# Characterisation of X Chromosomal Short Tandem Repeat Markers for Forensic Use

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## Declaration Page

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# Abstract

The use of X chromosomal short tandem repeat (STR) markers has been greatly increasing in the forensic setting. The marker system offers the potential to provide information in addition to that which is obtained from autosomal STR systems currently employed at forensic and paternity laboratories and in the courtroom. In certain scenarios, markers on the X chromosome may be the only means of obtaining the required information. Any investigated relationship situation where at least one female is involved will benefit from the use of X STRs, which can be applied to cases of missing persons, criminal incest, immigration, deficiency paternity or other questioned relationships. In-depth characterisation of the marker system is the first step in maximizing the power of this additional tool in the forensic arsenal.

Using guidelines set forth within the 1991 report of the International Society for Forensic Genetics (ISFG) relating to the use of DNA polymorphisms, all aspects of the feasibility of routine X STR use were evaluated. Two mini-X chromosomal STR multiplexes capable of amplifying 15 total markers (DXS6795, DXS9902, DXS8378, DXS7132, DXS6803, DXS6789, DXS7424, DXS101, GATA172D05, DXS7130, GATA165B12, HPRTB, GATA31E08, DXS10147, and DXS7423) were developed and optimised for use in the investigation and characterisation of allele nomenclature, allele/genotype frequencies, mutation rates, and linkage between markers. To simplify the transition into routine use, the assays utilised techniques and instrumentation already present in most forensic laboratories as well as analyses familiar to forensic scientists. Several large sample sets from four U.S. populations (African Americans, U.S. Asians, U.S. Caucasians, and U.S. Hispanics) were studied to address the lack of relevant data available for the United States, where the use of X STRs remains limited. Much of the knowledge gained, however, is universally applicable.

Though commercial kits are available that probe a wide variety of genetic markers on both the Y chromosome and the autosomes, there is only one commercial kit currently manufactured assaying markers on the X chromosome, the Investigator Argus X-12 kit (QIAGEN, Hilden, Germany). Due to patent and intellectual property issues between the United States and European STR kit manufacturers, the

kit cannot be sold or marketed in the U.S. at this time. Nonetheless, four markers overlapping with those present in the Argus X-12 kit were included in the optimised multiplexes and were the subject of an extensive concordance study. With greater than 99% concordance across 975 alleles, comparability of the data was established for these four markers while the utility of the kit with U.S. populations was evaluated for the first time.

This extensive developmental validation study investigated each aspect of the X STR marker system that would require consideration before implementation by a forensic laboratory. The optimised assays were found to be robust and the markers discriminating, while the mutation rate was estimated with high accuracy and the extent of linkage between the 15 markers was thoroughly evaluated. The combination of the results obtained as part of this study form the foundation upon which the introduction and routine use of X STRs may be built.

## Acknowledgements

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# Abbreviations

| | |
|---|---|
| µL | Microliter |
| µM | Micromolar |
| A | Adenine |
| AFDIL | Armed Forces DNA Identification Laboratory |
| BLAST | Basic Local Alignment Search Tool |
| BLAT | BLAST-like Alignment Tool |
| bp | Base pair(s) |
| BSA | Bovine serum albumin |
| C | Cytosine |
| CEPH | Centre d'Etudes du Polymorphisme Humaine |
| CI | Confidence interval |
| cM | Centimorgan |
| cm | Centimeter(s) |
| CODIS | Combined DNA Index System |
| Ct | Cycle threshold |
| DAB | DNA Advisory Board |
| DF | Dilution factor |
| dGTP | Deoxyguanosine triphosphate |
| $dH_2O$ | Distilled water |
| DNA | Deoxyribonucleic acid |
| dNTP | Deoxyribonucleotide triphosphate |
| EDNAP | European DNA Profiling Group |
| EDTA | Ethylenediaminetetraacetic acid |
| EMPOP | EDNAP mtDNA Population Database |
| FamID | Family identification number |
| FID | Father's identification number |
| $F_{ST}$ | Fixation index |
| G | Guanine |
| H(exp) | Expected heterozygosity |
| H(obs) | Observed heterozygosity |
| INDEL | Insertion/deletion marker |
| IPC | Internal PCR control |
| IRBO | Institutional Review Board Office |
| ISFG | International Society for Forensic Genetics |
| ISFH | International Society for Forensic Hameogenetics |
| kb | Kilobase(s) |
| kV | Kilovolts |
| LD | Linkage disequilibrium |
| LG | Linkage group |
| LISA | Laboratory Information Systems Applications |
| LOD | Logarithm of the odds |
| LR | Likelihood ratio |
| Mb | Megabase(s) |
| MEC | Mean exclusion chance |
| MECI | Mean exclusion chance in trios involving daughters |
| MECII | Mean exclusion chance in father/daughter duos |
| $MgCl_2$ | Magnesium chloride |
| MID | Mother's identification number |
| MIXplex | Mixture multiplex |

| | |
|---|---|
| mM | Millimolar |
| MRMC | U.S. Army Medical Research and Materiel Command |
| mtDNA | Mitochondrial DNA |
| NCBI | National Center for Biotechnology Information |
| ng | Nanograms |
| NIGMS | National Institute of General Medical Sciences |
| NIST | National Institute of Standards and Technology |
| $^{o}$C | Degrees Celcius |
| p (HWE) | P value of the exact test for Hardy-Weinberg equilibrium |
| PCR | Polymerase chain reaction |
| PDf | Power of discrimination in females |
| PDm | Power of discrimination in males |
| pg | Picograms |
| PIC | Polymorphism information content |
| PID | Patient identification number |
| Ref. | Reference |
| RF or Θ | Recombination fraction (or recombination rate) |
| SNP | Single nucleotide polymorphism |
| SRY | Sex-determining region of the Y chromosome |
| STR | Short tandem repeat |
| STRBase | Short Tandem Repeat Database |
| SWGDAM | Scientific Working Group on DNA Analysis Methods |
| T | Thymine |
| TLE | Tris-low-EDTA buffer |
| $T_m$ | Melting temperature |
| U.S. | United States |
| UCSC | University of California Santa Cruz |
| YHRD | Y Chromosome Haplotype Reference Database |
| Z | LOD score |

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# Chapter 1. Introduction

*1.1. Introduction to forensic short tandem repeat testing*

Autosomal short tandem repeat (STR) testing has been the cornerstone of forensic identity testing since the mid-1990s, when the first fluorescently-labelled STR markers were described. STR polymorphisms are stretches of DNA in which short nucleotide sequences (typically 3-5 base pairs in length) are repeated directly adjacent to one another and variation results from different numbers of repeated sequences. DNA testing replaced serological testing in forensic science in the mid-1980s, and offered several advantages over that traditional method including wide application to all body tissues and fluids, greater discriminatory potential, greater sensitivity and chemical robustness, and potential application to mixed samples [1]. The polymerase chain reaction (PCR) further increased the sensitivity of DNA testing, allowing analysis from just a few cells. Today, profiles are technically simple to generate through the use of multiplex PCR and fluorescent detection platforms. Numerous publications have characterised thousands of polymorphic STR markers that can be used for these purposes (see [2-6], for example), and commercial kits are available that facilitate comparisons between laboratories. While forensic use of autosomal and Y chromosomal STR markers is well-documented and accepted by both the scientific community and courts of law, markers on the X chromosome are less commonly applied, especially in the United States, and much of the groundwork for use of these markers is still in progress.

Autosomal STR loci across the genome are typically applied in combination to generate a genetic profile; this profile is used to putatively identify an individual through comparison to a reference profile, such as a suspect in a criminal investigation, or establish a genetic relationship, such as in mass disaster victim identification or paternity cases. Additionally, combinations of these markers have been used for ancestry investigations [7], population genetic comparisons, and the detection of chimeric mixtures [8], to name just a few applications. Male-specific Y chromosomal markers are commonly used in deficiency paternity cases or sexual assault mixtures. For example, in motherless cases, it is possible that autosomal STRs will not provide sufficient discrimination in order to assign paternity [9]. Instead, a Y STR profile can be used to distinguish between a son's two possible fathers, as occurred in a particular scenario where the 16-marker autosomal profiles

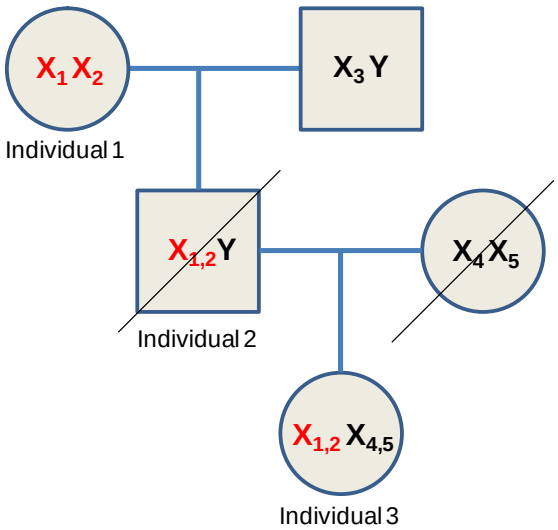
of two potential fathers resulted in similar paternity indices [10]. In another interesting case, four-marker Y STR haplotypes were used to exclude a male individual from the ancient Königsfelder paternal lineage despite archaeological evidence indicating a patrilineal association [11]. In missing persons cases, Y chromosomal STRs expand the pool of family reference samples that can be utilised to confirm identity [12]. Y chromosomal STRs have also proven useful in situations where the male portion of a mixture is of interest, often in the presence of large excesses of female DNA, as would be the case with a vaginal swab from a sexual assault [13-15]. Additionally, Y STRs have been used to investigate male lineages [16-19] and infer patterns of migration [20-23].

More recently, STR markers located on the X chromosome have emerged as additional tools in this forensic arsenal. X STRs can be used to supplement traditional STR typing due to their unique inheritance pattern and, correspondingly, the breadth of published literature on the subject has expanded greatly in recent years. STR markers on the X chromosome may be useful in several forensic contexts. To begin, missing persons cases usually require the analysis of relatives due to a lack of direct reference material. Often times, mitochondrial DNA (mtDNA) typing can be used to address the potential for degraded or low quantity specimens such as skeletal remains, particularly in closed populations and when a direct maternal reference is available, due to its relatively high copy number and protected location within the mitochondria of the cell [24]. However, mtDNA is maternally inherited; therefore where maternal references are unavailable or where the unidentified individual matches one of the most common mtDNA haplotypes, mtDNA testing alone may be inadequate. In such cases, markers on the X chromosome may provide additional information [25,26], offering the potential to both augment traditional STR testing and mtDNA sequencing for human remains identification as well as differentiate pedigrees that would be otherwise indistinguishable with unlinked autosomal STRs [27].

X chromosomal STRs can be particularly useful for any parent-child relationship that involves at least one female (e.g. father-daughter, mother-son, or mother-daughter) [28]. For example, Figure 1.1 shows the relationship pedigree of parents with one son who has a daughter. In this scenario, if the son and the biological mother of his

daughter are unavailable for testing, it may be necessary to use the grandparents' DNA profiles to re-associate their granddaughter. In this specific scenario, there is on average only one-quarter sharing of autosomal alleles between a grandparent and a grandchild, generally resulting in a low likelihood ratio (LR) of a relationship. X STRs, on the other hand, prove to be more useful since the X chromosome of the son is inherited entirely from his mother's genome with no contribution from his father (in the non-recombining region). This X chromosome is then passed in full to the daughter of the missing son. In this example, one would expect to see one allele from each X STR marker of the grandmother present in the granddaughter's X STR profile; therefore, the X STRs will most likely outperform autosomal STRs. Other maternally-related scenarios such as identifying cousins or aunt-niece relationships using X chromosomal STRs to augment or replace autosomal STR testing have been proposed [29].

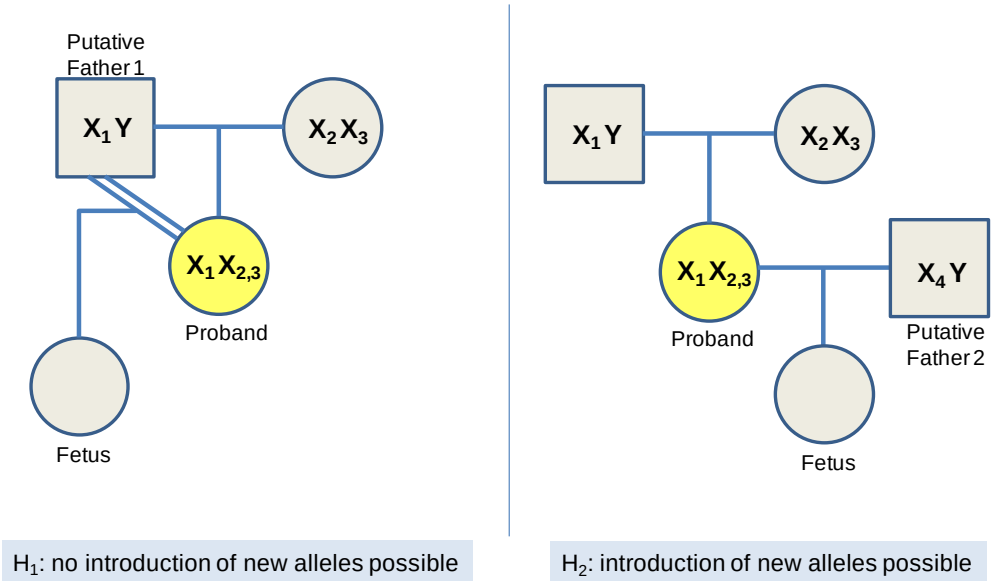
**Figure 1.1. Example of X chromosomal STR use for reassociation.** In this scenario, the matriarch (individual 1) passes a combination of her X chromosomes ($X_{1,2}$) to her son (individual 2). The son passes this combination in its entirety to his daughter (individual 3) without recombination. Without knowing the X STR profile of either parent, the granddaughter will share at least one allele at each X STR marker (50% allele sharing) with her paternal grandmother. Using autosomal STRs, the grandmother and granddaughter share only one quarter of their alleles by chance.




In alleged criminal incest investigations, the use of X STRs could be potentially more informative than any other marker system (Figure 1.2). In this published case example, the use of X STRs to exclude incest in a case of questioned paternity

without involving either potential father is illustrated [30]. A pregnant female (proband) presented with a question as to whether her father (putative father 1) or an unrelated man (putative father 2) is the true father of her unborn daughter. A combination of 21 autosomal STRs were uninformative as to the likely father since only the proband, her mother, and her fetus were available for testing. Additionally, Y chromosomal STRs and mtDNA analysis are both useless in this case. X chromosomal STRs, however, could provide confidence in an inclusion, since the fetus would not possess any alleles not present in the proband if that fetus indeed resulted from the incestuous relationship. That is, if the father of the proband was the true father of the proband's child, then the child would exhibit 100% allele sharing with the father's X chromosome ($X_1$) along with a combination of alleles from the X chromosomes of the proband ($X_1$ from the father and $X_{2,3}$ inherited from the grandmother). Alternatively, if the unrelated man was the true father, then the child would show additional alleles at some or all of the X STR markers tested due to the contribution from his X chromosome.

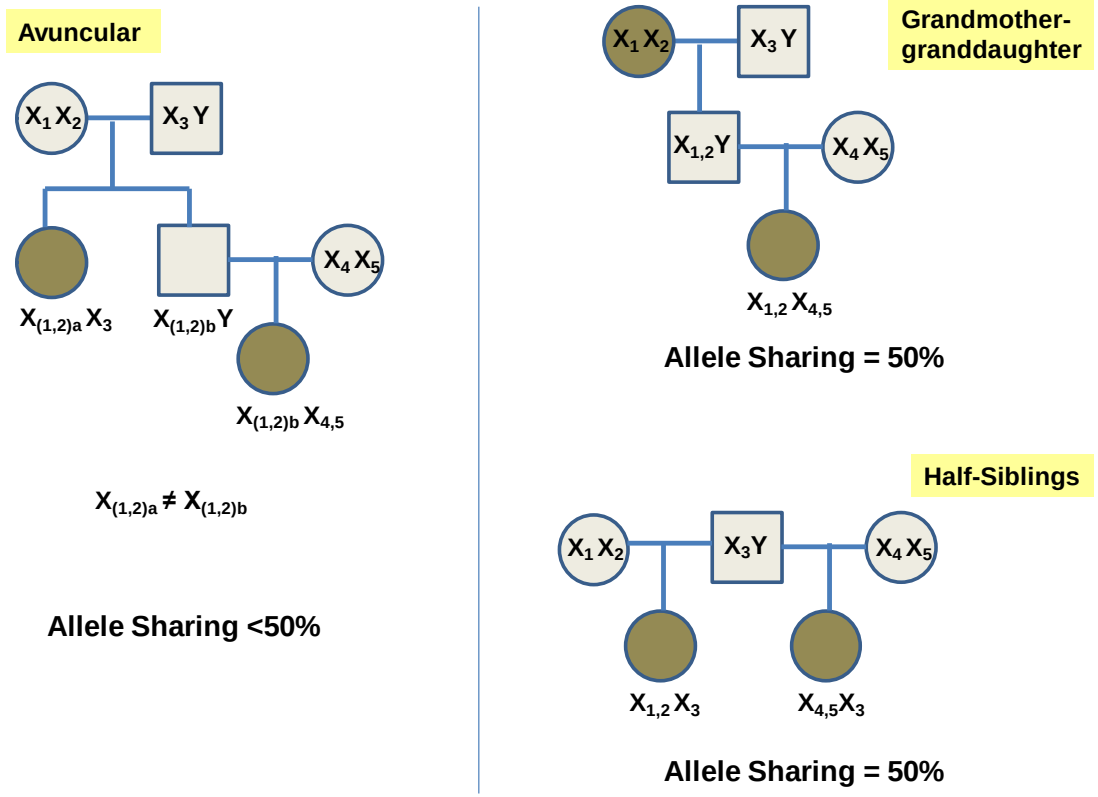
**Figure 1.2. Example of X chromosomal STR use in a paternity scenario involving incest.** In this scenario, there are two hypotheses being investigated. In hypothesis 1 ($H_1$), the father of the proband (yellow) is the putative father of her unborn child. Here, the female child (fetus) would inherit one half of her X STR alleles from the father ($X_1$), and a number of $X_1$ alleles would be transmitted to her from her mother (who also inherited the same $X_1$ chromosome from the father). A portion of the alleles transmitted to the child from her mother (proband) will contain the alleles from the proband's mother (present on chromosome $X_{2,3}$). Therefore, under $H_1$, it is expected that only alleles present in the mother's profile will be present in the fetus' profile and that there would be a high degree of homozygosity. Under the second hypothesis ($H_2$), the $X_4$ chromosome of the alternate putative father would be transmitted in its entirety to the fetus. Here, the fetus will have a number of alleles that are not present in her mother. The X chromosomal STRs can provide additional evidence to the investigator, while autosomal STRs provided little evidence given the limited number of individuals available for testing (see [30] for details).




It has been demonstrated that X STRs can differentiate pedigrees that are otherwise indistinguishable using only unlinked autosomal markers [27]. For example, using autosomal STRs, the avuncular, half-sibling, and grandparent-grandchild relationships cannot be differentiated [31]. Figure 1.3 depicts just one example of how X STR markers could distinguish the avuncular relationship from the other two in the case of a questioned relationship between two females. In the grandmother-granddaughter pedigree, similar to Figure 1.1, the grandmother passes on a combination of her X chromosomes ($X_1$, $X_2$) to her son, who passes along this X chromosome ($X_{1,2}$) in its entirety to the granddaughter. The grandmother and granddaughter, then, can be expected to share exactly one allele at every X STR

locus. Similarly, in the case of half-siblings, the common father passes his X chromosome ($X_3$) in its entirety without recombination to each of his daughters. The half-sisters therefore can be expected to share exactly one allele at every X locus. However, a closer look at the aunt-niece relationship depicted here reveals that the two women would not be expected to share an allele at each locus, and could potentially share none. Due to recombination of the unlinked X markers, the combination of $X_1$ and $X_2$ that each female receives will be different (barring unlikely recombination events generating two identical X chromosomes). Unlinked X chromosomal markers are similarly useful in the same pedigree scenarios involving one male and one female as well as two males [27].

**Figure 1.3. Example of X chromosomal STR use in questioned relationships.** In this scenario, the relationship between two women is questioned; no additional relatives are available for testing. Autosomal STRs cannot distinguish between the three relationship scenarios depicted here, and unlinked X chromosomal markers are utilised to further discriminate between the avuncular relationship and the grandparent-grandchild and half-sibling relationships. While exactly 50% of X STR alleles are expected to be shared by a grandmother and granddaughter or half-sisters, this sharing will be less than 50% between an aunt and her niece due to recombination.



Because the use of autosomal and Y chromosomal STRs is well-established in forensic genetics, these marker systems can be used as a model for the validation of the X chromosomal markers. Specifically, the development process of the Y chromosomal marker system can be used to highlight what steps have been completed, what needs to be accomplished, and the challenges still facing widespread X chromosomal marker use. As for the autosomes and Y chromosome, other non-STR markers have been investigated on the X chromosome, such as single-nucleotide polymorphisms (SNPs) [32-35] and insertion/deletion markers (INDELs) [36-38], and research has included additional species such as dogs [39]. However, the focus of this overview will centre on the forensic development of human-specific X STR markers in particular.

### *1.2. Validation of a marker system*

Guidelines used to validate a marker system such as autosomal or Y STRs for forensic use have been described previously [40,41]. These guidelines can, of course, be applied to any forensic marker system as technology changes and the scope of the questions asked by forensic scientists broadens. The aspects of the new marker system that must be addressed are as follows: 1.) selection of suitable markers with the intended purpose in mind; 2.) development of a multiplex assay that is robust and sensitive enough to accommodate targeted sample types; 3.) generation of high-quality population databases large enough to provide sufficient statistical vigour to the conclusions made based on the genetic information; 4.) assessment of the requirements for statistical interpretation of a match between two genetic profiles; and 5.) the potential for exchange/comparability of data between laboratories.

#### *1.2.1. Marker Selection*

A genetic locus (or marker) was defined early on as a segment of unique DNA sequence occupying a specific position on a chromosome [40]. Polymorphic loci, specifically STRs, have proven to be informative genetic loci for routine forensic use [2]. In general, STR markers are selected for forensic use based upon their potential discriminatory power (variability) and repeat size/structure (complexity). One definition of a useful microsatellite locus was set forth in a study describing those present on the Y chromosome [42]. Here, an acceptable locus has a repeat unit size

of greater than 3 base pairs, a repeat count greater than 8 repeats, and demonstrated polymorphism in 8 samples of different haplogroups (as defined by a set of bi-allelic markers). The large amount of data collected during this study was used to investigate the source of variation between individuals at these markers, and the most significant factor was found to be the average repeat count of the longest homogeneous array. Lack of variation, therefore, was found to be associated with a low repeat count, and complexity alone was not an indication of variability. This information allowed marker selection to favour simple repeats that could eliminate nomenclature conflicts (see Section 1.2.5) while maintaining polymorphism information content for use in forensic genetic investigations. For the Y chromosome specifically, simple but less variable markers could be chosen for comparing distant lineages [42]. After discovery and characterisation of a number of Y STR markers showing potential for forensic use, additional criteria were recommended by the ISFG, including consideration of a marker's potential for multiplexing, resolving mixtures (single-copy loci), and increasing overall haplotype discrimination (vs. discrimination value of the single STR) [43].

Marker selection for the community at large has been standardised for autosomal and Y STR markers. In the U.S., the Federal Bureau of Investigation has chosen a core set of 13 autosomal loci for inclusion into the national database, CODIS: CSF1PO, FGA, TH01, TPOX, VWA, D3S1358, D5S818, D7S820, D8S1179, D13S317, D16S539, D18S51, and D21S11. Recently, extending this set to include six additional autosomal markers (D2S1338, D19S433, D1S1656, D12S391, D2S441, and D10S1248) and one Y chromosomal marker (DYS391) was recommended [44,45]. The European Union, United Kingdom, and Germany also have defined sets of core loci. A core set of 9 markers has similarly been established for Y chromosomal STR comparisons: DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, and DYS385a/b [46,47]. These loci are collectively termed the "minimal haplotype" and, as for autosomal STRs, are required for database inclusion. An extended haplotype is also defined for Y markers and includes the loci DYS438, DYS439, DYS437, DYS448, DYS456, DYS458, DYS635, and YGATAH4 [48-50]. At the time of writing, there has been no move to standardise the set of X STR markers utilised in the community.

*1.2.2. Assay Development*

After appropriate markers have been identified as candidates for a particular forensic genetic application, further evaluation is then necessary to determine their suitability for inclusion in a multiplex PCR assay. Considerations include the ability to design acceptable primers within the regions flanking the repeats, the size range of the alleles for the particular marker in relation to other markers to be included in the multiplex, and the amplification efficiency of the individual primers in the context of a multiplex reaction. Because DNA templates encountered in the forensic setting are often degraded (for example, as the result of prolonged exposure to environmental extremes), amplicon size should be considered when designing primers. Amplicon size ranges for forensically-employed STR markers are typically less than 400 base pairs. When specifically targeting DNA that is known to be degraded, even shorter amplicon sizes are favoured with the goal of recovering the maximum number of alleles [51]. Genotyping using reduced-size amplicons for the 13 core autosomal loci used in the United States produced an increase in the ability to recover information from compromised samples while maintaining concordant profiles [52], and additional mini-STR loci were characterised to further increase the information that could be obtained from degraded samples [53]. In another study examining degraded samples, Asamura *et al*. demonstrated the success of two X STR quadruplex reactions consisting of amplicons ranging from 76-169 base pairs (bp) in length [54], and reduced size amplicons have since been designed for additional X STRs [55].

Primer design can most simply take place using a web-based program such as Primer3 [56], or on the NCBI website Primer-BLAST, which combines primer design using Primer3 and a BLAST search to ensure specificity of the primers for the intended template [57]. Primers are typically tested in singleplex reactions to determine the success and robustness of amplification. Selection of primers for a set of markers must take the annealing temperature of each individual primer pair into consideration, and aim for optimal amplification at one annealing temperature. However, adjustments can be made to the multiplex primer mix to account for any fluctuations in inter-locus balance due to a difference in amplification efficiency at any of the markers. Prior to selection of a particular primer pair, detection of potential primer interactions within the multiplex such as primer-dimer formation or

hairpin structures may be accomplished through a software program such as AutoDimer [58] or MultiPLX [59]. While such pre-screening may help avoid unwanted artefacts resulting from such interactions, evaluation of the chosen primer pairs in a multiplex reaction will reveal any additional artefacts or preferential amplification that could interfere with interpretation or prevent locus detection. Other considerations once the multiplex is assembled include avoiding allele size range overlap of adjacent markers and minimizing amplification artefacts such as stutter or incomplete adenylation. The physical separation of alleles belonging to markers adjacent to one another in the multiplex reaction may be accomplished by the additional of a short homopolymeric tail that increases the detected size of the amplicons. Primer tailing can also be an effective strategy for promoting the non-template adenylation of amplicons by *Taq* polymerase [60], thereby minimizing the doublet peak that can occur when both the adenylated and non-adenylated forms are present in appreciable amounts. Brownstein *et al*. [61] noted that placing a G base at the 5' end of the reverse primer favours adenylation and the addition of the sequence GTTTCTT to the 5' end resulted in nearly 100% adenylation of the PCR product. Ballard *et al*. [62] confirmed these results using both di- and tetranucleotide repeats and several different detection platforms and analysis methods.

Characterisation of stutter percentages and heterozygote peak balance also increases the ease of interpretation of multiplex data [63,64]. Stutter peaks occur at STR markers due to *Taq* slippage [65] and resultant amplification of a product that is typically one repeat unit smaller than the authentic allele [66,67]. Though both high stutter percentages and heterozygote peak imbalance can interfere with interpretation of potentially mixed samples, methodical characterisation of both the typical occurrence of these phenomena and the optimal input for the particular assay can mitigate their effects.

Ideally, multiple flanking region sequences would be examined in an alignment to look for variation that could affect primer binding prior to inclusion of the particular primer pair in a multiplex [68]. However, polymorphisms that result in reduced primer binding efficiency are often only recognised when a large sample set, such as that required for a population database, is processed with a multiplex. In one example, an excess of homozygosity at D8S1179 was observed in a population study

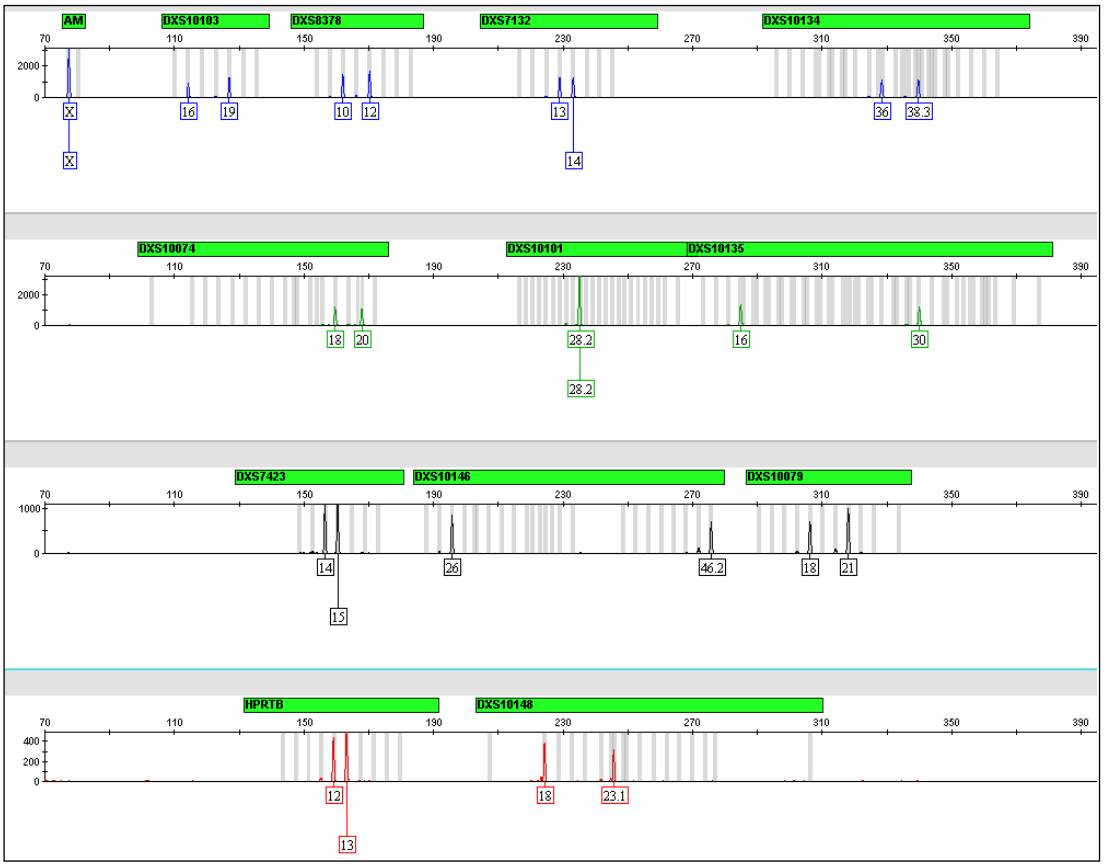
of Asian samples using the AmpF*l*STR® Profiler Plus® PCR amplification kit (Life Technologies), revealing a population-specific mutation under the reverse primer binding site [69]. In this case, a degenerate primer was designed and added to the primer mix and previously null alleles were recovered in the population samples. Similarly, null alleles have been detected for X STR loci, such as the null allele caused by a U.S. Hispanic-specific G to A substitution under the reverse primer binding site for GATA172D05 observed in two different multiplexes using the same primers [55,70]. This particular null allele was observed only twice in a total of 998 samples. In contrast to the D8S1179 strategy of adding an additional primer, no changes were made to either multiplex due to the rarity of the GATA172D05 mutation. This was also the case for additional null alleles observed at markers DXS7132, GATA165B12, and GATA31E08 [55]. It was recommended for the Y chromosome that all SNPs be reported in a central location, the Y Chromosome Haplotype Reference Database (YHRD; see Section 5), so that null alleles might be predicted before they were encountered in practice [43]. The assay design strategy used for a Y STR decaplex addressed some of these aspects in more detail and may be used as a general model [68].

Once a functioning multiplex has been designed, additional developmental validation studies should be performed to determine its suitability for use in human forensic genetics investigations; such studies have been outlined for use within the United States by the DNA Advisory Board (DAB) and Scientific Working Group on DNA Analysis Methods (SWGDAM) [71,72]. Given the potential for low quantity samples in this setting, the sensitivity of the assay should be evaluated and an optimal input range for reliable results determined. Additionally, the cross-reactivity of the primers used in the multiplex with other forensically relevant nonhuman species should be documented. Primers designed to be human-specific may also amplify primate DNA sequences but should not amplify bacterial or fungal contaminants, for example. Mixture studies might also be performed to determine the limitations of the new assay with regards to discriminating individual contributors.

For both the Y chromosome and the autosomes, commercial kits are available that probe a wide variety of genetic markers, and commonly the work completed during

development of these multiplexes is published; PowerPlex® 16 (Promega Corporation) [73], AmpF*l*STR® Minifiler™ (Life Technologies) [74], PowerPlex® Y (Promega Corporation) [75], AmpF*l*STR® Yfiler® (Life Technologies) [50], PowerPlex® ESX/ESI (Promega Corporation) [76,77], and AmpF*l*STR® Identifiler® (Life Technologies) [78] are all examples of commercial kits used by forensic laboratories with published validation studies. Wallin *et al*. [79] describes an approach to the process of developmental validation for a variety of early STR kits. For X chromosomal STRs, there have been several iterations of one commercial kit. The Mentype® Argus X-UL kit simultaneously amplified four X STR markers (DXS8378, DXS7423, HPRTB, and DXS7132) plus amelogenin in a single dye channel [80]. The Mentype® Argus X-8 kit expanded the number of markers to eight by adding four additional STRs (DXS10134, DXS10074, DXS10101, and DXS10135) [81]. The current iteration of this multiplex is produced by Qiagen® and is known as the Investigator Argus X-12 kit, which includes yet another four markers (DXS10103, DXS10079, DXS10146, and DXS10148) present in four dye channels [82] (Figure 1.4).

**Figure 1.4. Qiagen® Investigator Argus X-12 amplification kit example.** Profile for 500 pg of control DNA XX28 (included with the kit).




Due to patent and intellectual property issues between United States and European STR kit manufacturers, these kits cannot be sold or marketed in the United States at this time. It is believed that going forward, as more laboratories demand X STRs, additional X STR kits will be manufactured for the forensic community. For now, most X STR typing currently relies upon non-commercial multiplex assays, simultaneously amplifying 2 to 12 markers in a single reaction, that have been designed and published by individual laboratories interested in X STR typing (see [54,55,70,83-111] and Table 1.1).

**Table 1.1. Published multiplex PCR assays targeting X chromosomal STR markers.** Thirty-five published X STR multiplexes have been established in the forensic community and used for population studies, paternity investigations, and other purposes. Thirty-six of the most commonly used markers were included in the various multiplexes. $N_M$: number of marker in the multiplex.

| Ref. | $N_M$ | Population studied | DXS101 | HPRTB | DXS7423 | DXS6789 | DXS8377 | DXS7132 | DXS7424 | DXS8378 | DXS7133 | GATA172D05 | DXS6809 | DXS6807 | DXS9898 | GATA31E08 | DXS6800 | DXS981 | GATA165B12 | DXS10011 | DXS9902 | DXS6803 | HumARA | DXS6797 | DXS7130 | DXS6801 | DXS9895 | DXS10074 | DXS10079 | DXS6799 | DXS6854 | DXS6804 | DXS10075 | DXS6795 | DXS6808 | DXS10147 | DXS10078 | DXS6810 | Additional studies using multiplex & population studied |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| [94] | 5 | Cantabria | X | X | X | | X | | | | | | | | | | | | | | | | X | | | | | | | | | | | | | | | | Cantabria & Basque [112] |
| [87] | 4 | Germany | X | X | | | X | | | | | | | | | | | X | | | | | | | | | | | | | | | | | | | | | Tuscany [113]; case example [114] |
| [90] | 7 | Italians | X | X | X | X | | | | | | | | X | | | | | | X | | | X | | | | | | | | | | | | | | | | |
| [92] | 4 | Korean | | | | | | | X | | | | X | | X | | | | | X | | | | | | | | | | | | | | | | | | | |
| [93] | 4 | Korean | X | X | | | X | | | | | X | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| [91] | 10 | NE German | X | X | | | X | | X | X | X | | | X | X | | | | | X | | | X | | | | | | | | | | | | | | | | Latvia [115] |
| [89] | 5 | China, Japan, Thai, German, Philippines | X | X | | X | X | | | | | | | | | | | X | | | | | | | | | | | | | | | | | | | | | Kurd & German [116] |
| [54] | 8 | Japan | X | | X | X | | | X | X | X | | | | | X | | | X | | | | | | | | | | | | | | | | | | | | |
| [84] | 8 | Japan | | X | | | X | X | | | | X | | X | X | | | | | X | | | | X | | | | | | | | | | | | | | | |
| [105] | 6 | Italy | X | | X | X | X | | | | | | | X | | | | | | | | | X | | | | | | | | | | | | | | | | Italy [117] |
| [109] | 4 | N. Spain | X | X | X | | X | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| [86] | 12 | NW Italian | X | X | | X | X | X | X | X | | X | X | | | | X | | | X | | | | | | X | | | | | | | | | | | | | Algeria [118]; Ivory Coast [119]; case example [120] |
| [70] | 10 | United States, 3 groups | X | X | X | X | X | X | | X | | X | X | | X | | | | | | | | | | | | | | | | | | | | | | | | Spain [121]; Columbia [122]; Angola [123]; Mozambique [123]; Uganda [123]; N. Portugal [124] |
| [99] | 12 | N. Italy | X | X | X | X | | X | X | X | X | X | X | X | | X | | | | | | | | | | | | | | | | | | | | | | | India [125] |

Table continues on next page.

| Ref. | $N_M$ | Population studied | DXS101 | HPRTB | DXS7423 | DXS6789 | DXS8377 | DXS7132 | DXS7424 | DXS8378 | DXS7133 | GATA172D05 | DXS6809 | DXS6807 | DXS9898 | GATA31E08 | DXS6800 | DXS981 | GATA165B12 | DXS10011 | DXS9902 | DXS6803 | HumARA | DXS6797 | DXS7130 | DXS6801 | DXS9895 | DXS10074 | DXS10079 | DXS6799 | DXS6854 | DXS6804 | DXS10075 | DXS6795 | DXS6808 | DXS10147 | DXS10078 | DXS6810 | Additional studies using multiplex & population studied |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| [107] | 4 | Belarus, Slovakia | X | X | X | | X | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| [98] | 5 | Han, China | | | | X | | X | | | | | X | | | | | X | | | | X | | | | | | | | | | | | | | | | |
| [106] | 4 | Peru | | X | | | | | | | | X | | | | | X | | | | | | | X | | | | | | | | | | | | | | |
| [102] | 4 | Bauru, Brazil | | | | | | X | X | | | | | | | | | | | | | | | | | | | | | | X | | | | X | | | | Brazil [126] |
| [97] | 11 | Brazil Amazon | | X | X | X | | X | | | X | | | | X | X | X | | | X | | | | | X | | X | | | | | | | | | | | | Brazil [127,128] |
| [104] | 6 | Brazil | | | X | | X | | X | X | X | | | X | | | | | | | | | | | | | | | | | | | | | | | | | paternity testing [25,129] |
| [83] | 10 | Iberian, Latin American | | | X | X | | X | | X | X | X | X | | X | X | | | | | | X | | | | | | | | | | | | | | | | | Azores [130]; Uganda [131]; paternity testing [132,133]; Brazil [134,135]; Macau [136]; Portuguese Gypsies [137]; Argentina [138,139]; prostate cancer study [140]; Spain [141-144] |
| [96] | 13 | Taiwan | X | X | X | X | X | X | X | X | | X | X | X | X | | | | | | X | | | | | | | | | | | | | | | | | | Taiwan [145] |
| [101] | 4 | Beijing Han | | | | | | X | | | | | | | | | X | | | | X | | | | | | | | | X | | | | | | | | |
| [95] | 5 | Pakistan | | | | | | | | | X | | | | | | X | X | X | | | | | X | | | | | | | | | | | | | | |
| [146] | 11 | Ghana | X | X | X | | X | X | X | X | X | | | X | X | | X | | | | | | | | | | | | | | | | | | | | | | Morocco [147]; Madagascar [147] |
| [103] | 5 | Pakistan | X | | | X | | | X | | | | X | | | | | | | | | | | | | X | | | | | | | | | | | | | |
| [85] | 7 | Chinese Han | X | | | X | | | X | | X | | | | | X | | X | X | | | | | | | | | | | | | | | | | | | | |
| [100] | 10 | Chinese Daur | X | X | X | X | | X | | X | X | | | | | | | | | | | | | | X | | | | | X | | X | | | | | | | China [148] |
| [110] | 5 | Brazil | X | | | | | | X | | | | | | | | | | | | | | | | | | | X | X | | | | X | | | | | | |
| [108] | 16 | Japan | X | X | X | X | X | X | X | | X | X | X | X | | X | X | X | | | | X | | | | | X | | | | | | | | | | | | |
| [55] | 15 | United States, 4 groups | X | X | X | X | | X | X | X | | X | | | | X | | | X | | X | X | | | X | | | | | | | | | | X | | X | | | United States, 3 groups [149]; Bosnia & Herzegovina [150] |
| [151] | 11 | Tibet | | X | X | | X | X | | X | X | | | | X | | | | X | | | X | | | | X | | | | | | | | X | | | | | China [152] |
| [111] | 9 | China, 3 groups | | X | X | | X | | | X | | X | X | X | | | | | | | X | | | | | | | | | X | | | | | | | | | |
| [153] | 12 | Chinese Han | | | | X | | | | | | X | | | | X | | | X | | | X | | X | X | | X | X | | | | X | | | | | X | X | |
| [154] | 15 | China, 3 groups | X | | | X | | X | X | | X | | X | | X | X | X | X | X | | | | | | | X | | X | X | | | | X | | | | | | |

*1.2.3. Reference Population Database Generation & Mutation Rate Analysis*

Population databases must be generated as part of the validation of a specific marker system in order to ensure the ability to quantify the value of a "match" between two samples. These reference databases should contain at least 100-250 unrelated individuals from relevant populations [40,41] and are used to calculate population-specific allele or haplotype frequencies. Observed and expected genotype frequencies are used to confirm that the population is in Hardy-Weinberg equilibrium, and additional parameters such as expected heterozygosity (H(exp)), power of discrimination (PD), polymorphism information content (PIC), and mean exclusion chance (MEC) are also calculated to evaluate the usefulness of the selected set of markers. Software programs such as PowerMarker [155] and Arlequin [156] can be employed to aid in these calculations for large sample sets. The MEC and PD are calculated differently for X chromosomal markers than for autosomal markers and depending upon sex and/or kinship situation, i.e. trios involving a daughter versus father-daughter duos [29]. The Forensic ChrX Research website's "Evaluate & Calculate" section can be used to automatically calculate these (and other) forensic efficiency parameters specifically for X STR markers [157].

There are a number of population studies that have been performed with the various iterations of the commercial X STR kits [119,158-184] and these are summarised in Table 1.2. Also, a number of the non-commercially produced published multiplexes have been used by additional laboratories to increase the population data available for a particular marker set [25,113-118,121-127,129-141,145,150,181,185-187] and are noted in Table 1.1.

**Table 1.2. Summary of published population studies using commercial X STR multiplexes.** "X-UL" refers to the Mentype® Argus X-UL; "X-8" refers to the Mentype® Argus X-8 kit; and "X-12" refers to the Qiagen® Investigator Argus X-12 kit. "X-8+4" indicates that the same twelve markers in the Qiagen® Investigator Argus X-12 kit were typed but that the Mentype® Argus X-8 kit was used in combination with a non-commercial quadruplex. N: total number of individuals included in study. *The three Hungarian populations are identical, but were processed separately with all three kits.

| Reference | Kit | Population studied | N |
|---|---|---|---|
| [158] | X-UL | Poland | 240 |
| [159] | X-UL | Italy | 90 |
| [160] | X-UL | United Kingdom | 200 |
| [160] | X-UL | Ireland | 200 |
| [160] | X-UL | South Asia | 200 |
| [161] | X-UL | China | 500 |
| [162] | X-UL | Brazil | 184 |
| [163] | X-UL | Portugal | 200 |
| [164] | X-UL | Poland, 2 minority groups | 420 |
| [165] | X-UL | Hungary* | 384 |
| [166] | X-8 | Italy | 100 |
| [167] | X-8 | Germany | 259 |
| [167] | X-8 | Ghana | 59 |
| [167] | X-8 | Japan | 93 |
| [168] | X-8 | Italy | 131 |
| [169] | X-8 | Japan | 258 |
| [170] | X-8 | Ghana | 182 |
| [171] | X-8 | Hungary* | 384 |
| [172] | X-8 | Turkey | 100 |
| [173] | X-8 | Finland | 300 |
| [173] | X-8 | Somalia | 300 |
| [174] | X-8 | Korea | 300 |
| [175] | X-8 | NE Italy | 176 |
| [176] | X-8 | Japan | 492 |
| [177] | X-8 | Poland | 311 |
| [178] | X-8 | China | 303 |
| [179] | X-8 | Korea | 138 |
| [180] | X-8 | China | 198 |
| [181] | X-8+4 | Algeria | 210 |
| [119] | X-8+4 | Ivory Coast | 125 |
| [188] | X-12 | Morocco | 145 |
| [189] | X-12 | United States, 4 groups | 853 |
| [190] | X-12 | Germany | 1037 |
| [182] | X-12 | Hungary* | 407 |
| [184] | X-12 | NE Italy | 207 |
| [183] | X-12 | Chinese Han | 272 |
| [191] | X-12 | Kuala Lumpur | 283 |
| [192] | X-12 | Sweden | 652 |
| [193] | X-12 | Greenland | 198 |
| [193] | X-12 | Denmark | 210 |
| [193] | X-12 | Somalia | 441 |

According to the 1991 report of the International Society for Forensic Genetics (ISFG; formerly ISFH (Hameogenetics)) relating to the use of DNA polymorphisms in paternity testing, mutation rates must be known in order to adequately address possible mismatches attributable to mutational events [40]. A known mutation rate can be used to distinguish closely related individuals or define the time to most recent common ancestor (i.e. dating of Y chromosome lineage origins) using a large set (>200) of Y STR markers, as described by Kayser *et al*. [42]. Mutations typically occur as a result of strand slippage during DNA replication, and are the major mechanism of the high degree of polymorphism seen in human microsatellites [194]. Single-step mutations (insertions or deletions of one repeat unit) are most common, affecting longer alleles more frequently than shorter ones [195]. This trend is observed for markers on the Y chromosome [196], and no difference is expected on the X chromosome. Mutation rates can differ, however, between males and females, with one estimate of the ratio of paternal to maternal mutations at 17:3 [195]. Since twice the number of X chromosomes are present in females than in males, however, it is possible that this ratio will be shifted, causing the difference between the sexes to become less obvious. Mutation rates can also vary with population, as was demonstrated in several studies of Y chromosomal markers where African Americans had a slightly higher rate than other populations [196,197]. This variation is most likely related to the length of the most frequently observed alleles in each population.

Typical mutation rates for autosomal and Y chromosomal markers are in the range of ~1-5 x $10^{-3}$ [41]. Existing reports of X STR mutation rates fall within this range, though many are based upon a relatively small number of meiotic events (Table 1.3, reproduced here from supplementary material associated with manuscript in Chapter 5, Section 5.1). As evident from Table 1.3, there is a need to investigate mutation rates in populations outside of Europe in general and Germany specifically. Currently, there are no studies in the literature that investigate mutation rates among U.S. population groups, or from the continent of Australia, for example. Additionally, marker-specific mutation rates, along with the features of those markers that may affect the mutation rate, have previously been recognised as important for forensic and population genetic purposes [195,198,199]. For example, the two markers with the highest mutation rate in the literature are ARA and

DXS8377, both of which contain trinucleotide repeats [200]. In addition, both have very long repeat stretches (on the order of 10-50 repeat units) that are highly polymorphic (>20 alleles). The contribution of each of these characteristics, as well as the need for marker-specific rates, has not yet been investigated for X STR markers. A summary of pooled marker-specific mutation rates from the literature are noted in Table 1.4 (reproduced here from supplementary material associated with manuscript in Chapter 5, Section 5.1). Note that the ARA locus is no longer considered suitable for forensic use because it falls within the coding region of a gene in which a mutation would give rise to X-linked spinal and bulbar muscular atrophy [201].

In order to enhance the practical application and interpretation of X STR markers, the rate of mutation should be estimated through examination of a substantial number of meioses [41], and the dependence of mutation rates upon the origin, length, and structure of the allele should be investigated.

**Table 1.3. Published overall X STR mutation rates.** The overall mutation rates from 34 published studies were compiled and 95% confidence intervals (CI) calculated.

| Ref. | Population | Markers (N) | Mutations (N) | Meioses (N) | Mutation rate (x $10^{-3}$) | 95% CI (x $10^{-3}$) |
|---|---|---|---|---|---|---|
| [202] | Argentina | 7 | 1 | 1015 | 0.99 | 0.02-5.5 |
| [203] | Austria & Germany | 3 | 0 | 834 | 0.00 | 0-4.4 |
| [204] | China | 2 | 0 | 312 | 0.00 | 0-11.7 |
| [161] | China | 4 | 0 | 424 | 0.00 | 0-8.7 |
| [98] | China | 5 | 8 | 4295 | 1.86 | 0.8-3.7 |
| [154] | China | 15 | 13 | 11850 | 1.10 | 0.6-1.9 |
| [122] | Columbia | 10 | 4 | 1460 | 2.74 | 0.7-7.0 |
| [205] | Germany | 1 | 2 | 580 | 3.45 | 0.42-12.4 |
| [206] | Germany | 1 | 0 | 340 | 0.00 | 0-10.8 |
| [207] | Germany | 1 | 0 | 404 | 0.00 | 0-9.1 |
| [208] | Germany | 1 | 0 | 300 | 0.00 | 0-12.2 |
| [87] | Germany | 4 | 0 | 372 | 0.00 | 0-9.9 |
| [209] | Germany | 16 | 16 | 7658 | 2.09 | 1.2-3.4 |
| [91] | Germany | 10 | 0 | 500 | 0.00 | 0-7.3 |
| [167] | Germany | 8 | 1 | 2800 | 0.36 | 0.01-2.0 |
| [210] | Germany | 3 | 3 | 1029 | 2.92 | 0.6-8.5 |
| [211] | Germany | 8 | 8 | 1680 | 4.76 | 2.1-9.4 |
| [146] | Ghana | 11 | 0 | 198 | 0.00 | 0-18.5 |
| [193] | Greenland, Denmark, Somalia | 12 | 20 | 6156 | 3.25 | 2.0-5.0 |
| [165] | Hungary | 4 | 1 | 768 | 1.30 | 0.03-7.2 |
| [212] | Italy | 3 | 0 | 240 | 0.00 | 0-15.2 |
| [99] | Italy | 12 | 0 | 1080 | 0.00 | 0-3.4 |
| [213] | Japan | 12 | 0 | 648 | 0.00 | 0-5.7 |
| [93] | Korea | 5 | 1 | 180 | 5.56 | 0.14-30.1 |
| [214] | Pakistan | 13 | 0 | 1300 | 0.00 | 0-2.8 |
| [95] | Pakistan | 5 | 0 | 840 | 0.00 | 0-4.4 |
| [89] | Phillipines | 5 | 1 | 445 | 2.25 | 0.06-12.5 |
| [158] | Poland | 4 | 0 | 320 | 0.00 | 0-11.5 |
| [215] | Poland | 4 | 0 | 264 | 0.00 | 0-13.9 |
| [164] | Poland | 4 | 0 | 600 | 0.00 | 0-6.1 |
| [216] | Spain | 2 | 1 | 214 | 4.67 | 0.12-25.8 |
| [94] | Spain | 5 | 0 | 125 | 0.00 | 0-29.1 |
| [217] | Spain | 6 | 4 | 1164 | 3.44 | 0.9-8.8 |
| [218] | Mixed | 12 | 8 | 10290 | 0.78 | 0.34-1.5 |
| | | **Totals** | **92** | **60685** | **1.52** | **1.2-1.9** |

**Table 1.4. Marker-specific X STR mutation rates from published studies.** Mutations have been studied in 36 X STR markers and reported in at least 34 publications. Pooled marker-specific rates are shown here with 95% confidence intervals (CI). Markers also investigated as part of the present study are shown in bold.

| Marker | Mutations (N) | Meioses (N) | Pooled mutation rate ($\times 10^{-3}$) | 95% CI ($\times 10^{-3}$) | Reference |
|---|---|---|---|---|---|
| ARA | 4 | 673 | 5.94 | 1.6-15.1 | [91,93,94,209] |
| DXS8377 | 10 | 1702 | 5.88 | 2.8-10.8 | [87,89,91,93,94,122,146,203,209,214,216] |
| DXS10135 | 9 | 2318 | 3.88 | 1.8-7.4 | [167,193,210,211,218] |
| DXS10079 | 7 | 2293 | 3.05 | 1.2-6.3 | [154,193,217,218] |
| DXS10103 | 2 | 1310 | 1.53 | 0.18-5.5 | [193,218] |
| DXS10146 | 2 | 1258 | 1.59 | 0.19-5.7 | [193,218] |
| DXS10134 | 6 | 2027 | 2.96 | 1.1-6.4 | [167,193,211,213,218] |
| DXS10148 | 5 | 1598 | 3.13 | 1.0-7.3 | [193,210,218] |
| DXS10075 | 3 | 984 | 3.05 | 0.63-8.9 | [154,217] |
| **DXS7132** | **11** | **4965** | **2.22** | **1.1-4.0** | [98,99,122,146,154,158,161,164,165,167,193,209,211-215,218] |
| DXS10074 | 6 | 3014 | 1.99 | 0.73-4.3 | [154,167,193,211,213,217,218] |
| **DXS6803** | **2** | **1015** | **1.97** | **0.24-7.1** | [98,204] |
| DXS6809 | 4 | 2133 | 1.88 | 0.51-4.8 | [98,99,122,154,213,217] |
| **HPRTB** | **6** | **4530** | **1.32** | **0.49-2.9** | [87,89,91,93,94,99,122,146,158,161,164,165,167,193,202,205,209,211,214,215,218] |
| **DXS7424** | **2** | **1805** | **1.11** | **0.13-4.0** | [91,99,146,154,202,208,209] |
| **DXS8378** | **4** | **3882** | **1.03** | **0.28-2.6** | [91,99,122,146,158,161,164,165,167,193,202,209-211,213-215,218] |
| DXS10101 | 1 | 1973 | 0.51 | 0.01-2.8 | [167,193,211,218] |
| **GATA31E08** | **1** | **1127** | **0.89** | **0.02-4.9** | [99,154,202,213,214] |
| **DXS6789** | **3** | **3478** | **0.86** | **0.18-2.5** | [89,98,99,122,154,207,209,213,214,217] |
| **DXS7423** | **2** | **3515** | **0.57** | **0.07-2.1** | [94,99,122,146,158,161,164,165,167,193,202,209,211,213-216,218] |
| DXS9898 | 1 | 1936 | 0.52 | 0.01-2.9 | [91,122,146,154,202,209,213] |
| **DXS101** | **1** | **2534** | **0.39** | **0.01-2.2** | [87,89,91,93,94,99,122,146,154,202,203,206,209] |
| DXS10011 | 0 | 50 | 0.00 | 0-71.1 | [91] |
| **DXS10147** | **0** | **54** | **0.00** | **0-66.0** | [213] |
| DXS6810 | 0 | 100 | 0.00 | 0-36.2 | [214] |
| DXS6793 | 0 | 100 | 0.00 | 0-36.2 | [214] |
| DXS6797 | 0 | 168 | 0.00 | 0-21.7 | [95] |
| **DXS9902** | **0** | **458** | **0.00** | **0-8.0** | [209,213,214] |
| DXS6807 | 0 | 598 | 0.00 | 0-6.2 | [91,99,146,209] |
| **GATA172D05** | **0** | **876** | **0.00** | **0-4.2** | [93,99,122,209,212-214] |
| DXS9895 | 0 | 917 | 0.00 | 0-4.0 | [204,209] |
| **GATA165B12** | **0** | **958** | **0.00** | **0-3.8** | [95,154] |
| DXS6801 | 0 | 1084 | 0.00 | 0-3.4 | [154,214,217] |
| DXS7133 | 0 | 1459 | 0.00 | 0-2.5 | [91,95,99,146,154,209,212] |
| DXS6800 | 0 | 1694 | 0.00 | 0-2.2 | [95,146,154,203,209] |
| DXS981 | 0 | 2099 | 0.00 | 0-1.8 | [87,89,95,98,154,214] |

*1.2.4. Requirements for statistical interpretation of a match*

Standard 8.1.3.2 of the U.S. DAB's Quality Assurance Standards for Forensic DNA Testing Laboratories states that laboratories shall establish and document match criteria on the basis of empirical data [71]. Another requirement of the 1991 ISFG report relating to the use of DNA polymorphisms is that questions of independent assortment and linkage disequilibrium be addressed [40]. For autosomal STRs, this ensures that the product rule can be used to multiply individual marker frequencies together to determine the overall rarity of a profile. It does not preclude the use of linked markers, however. Y chromosomal STRs, for example, are linked to one another and are considered together as a group called a haplotype. Haplotype frequencies are measured directly from population data, and the counting method is just one approach used to determine the rarity of the profile [219]. It follows that X chromosomal STRs may be a combination of the two techniques: the organisation of several physically close markers into linkage groups, forming haplotypes, whose frequencies could then be multiplied together once independent assortment of the groups was established.

From the ISFG report, it is clear that both linkage and linkage disequilibrium must be studied. Linkage refers to the co-segregation of closely located markers within a pedigree and can be measured by calculating the recombination fraction (RF) from family samples. Families in which the gametic phase of the parents is known (requires grandparental profiles) and which have multiple offspring are used to calculate the RF using informative meioses. A meiosis is informative for linkage if it can be unambiguously determined that the gamete is recombinant. Homozygous markers, for example, will prevent a meiosis from being informative. A statistical test (logarithm of the odds, or LOD) is then applied to determine if the observed RF value is significantly different from that expected for independent assortment (0.5). Computer programs, such as LINKAGE [220,221] or Genetic Data Analysis (GDA) [222], are available that will perform these calculations for complicated pedigrees and/or large sets of autosomal markers.

A set of families satisfying the requirements of linkage study (multiple generations and offspring) have been established at the National Institute of General Medical

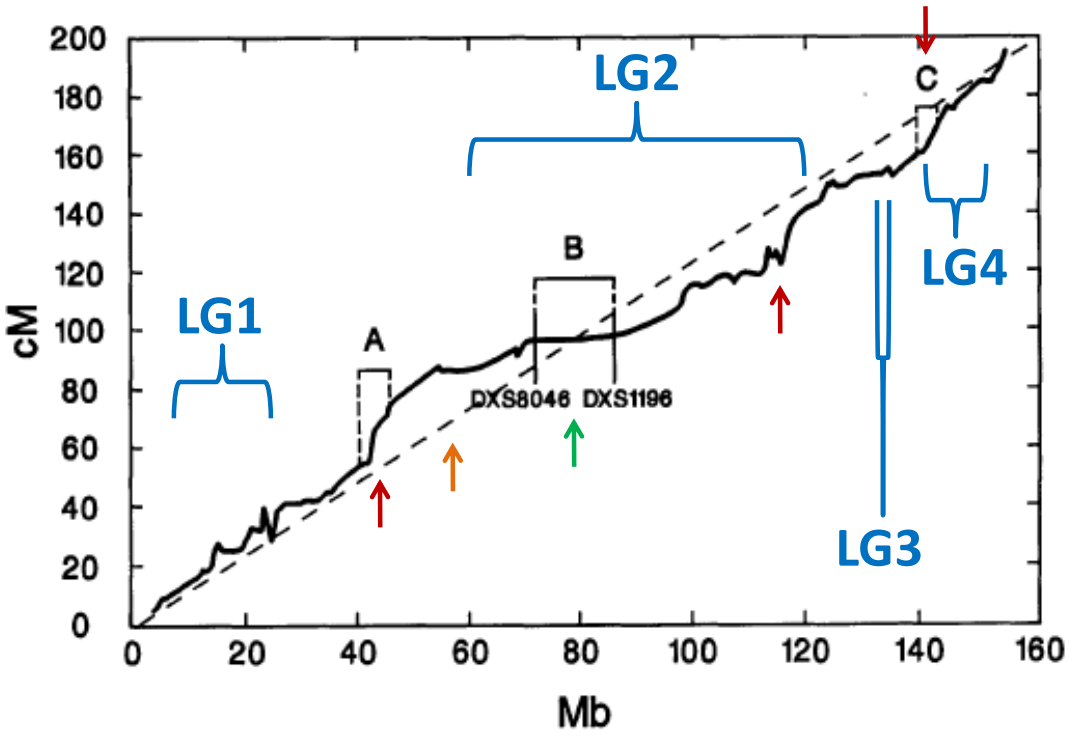
Sciences (NIGMS) repository from lymphoblastic cell lines donated by the Centre d'Etudes du Polymorphisme Humaine (CEPH). The collection includes families from Utah, France, Venezuela, and one Old Order Amish family from Pennsylvania, making this an important resource for the characterisation of DNA polymorphisms and the construction of the human genetic map. In addition, any research effort that requires access to a common dataset can find value in the use of the CEPH reference families, as evidenced by the large amount of data that has already been collected from them and shared in a database made available to contributing researchers. Several reliable linkage maps of the human genome have resulted from such collaborations [223-225]. In fact, most forensic publications refer to the location of markers on the X chromosome according to the Marshfield map [225], which is based upon analysis of recombination rates in a subset of 8 CEPH families. The CEPH families were also used to create the Rutgers combined linkage-physical map of the human genome, which is a denser map incorporating both sequence-based positional information as well as recombination-based data [226]. This Rutgers map has been used to generate a consolidated list of physical and genetic distances between 39 commonly-used forensic X chromosomal markers [227], but since the entire set of 39 markers had not been directly measured, some genetic distances were interpolated. Additionally, certain X chromosomal markers are missing from this analysis (DXS6795, for example). Therefore, while the physical map of the X chromosome may be well-established, further study of chosen markers to create a genetic map through family studies is required.

Linkage disequilibrium measures the non-random association of two or more alleles that are not necessarily closely located on a chromosome, and is estimated from allele and haplotype frequencies. Statistical tests designed to indicate linkage disequilibrium across the genome can potentially highlight co-segregating markers on the X chromosome that may be physically or genetically linked [228]. However, large sample sizes are required to obtain reliable estimates, and this measure alone cannot establish groups of markers that should be considered as haplotypes. While most population studies include a test for linkage disequilibrium, little scrutiny of linkage on the X chromosome has been performed. However, such information is necessary to the application of information obtained from such markers. Early linkage studies performed with 182 mother-multiple son constellations produced a

map of the X chromosome that divided 16 X chromosomal STRs into four linkage groups [209] (Chapter 5, Figure 5.1). The hypothesis was that alleles at linked markers combine to form haplotypes that could recombine during meiosis as "blocks." In the same study, linkage disequilibrium was estimated from a population of over 200 males and showed association between only two markers: DXS7424 and DXS101. No further confirmation of this proposed linkage situation was undertaken at this point. Since this initial work, a number of different sets of physically close markers have been studied, demonstrating that alleles do indeed co-segregate as stable haplotypes [210,229,230], especially markers located around the centromere where recombination rates are reduced [231,232]. Still, little research on the linkage situation has been completed with the growing number of available markers across the entire chromosome.

Support for this theory of four linkage groups can be found in a study published six years prior. Here, Nagaraja *et al*. [233] produced a high resolution map of the X chromosome allowing regional differences in recombination rate to be highlighted through comparison of genetic and physical differences. Figure 1.5, reproduced from [233], plots genetic versus physical distance across the chromosome. Regions with a steep slope (i.e. larger Mb/cM ratio) revealed a locally higher recombination rate, indicating potential recombination "hot spots." Interestingly, these potential "hot spots" coincide with the borders between the proposed linkage groups. Additionally, one region of locally lower recombination rate can be observed within linkage group 2, rather than at the centromere, as expected. This region may prove especially useful for targeting tightly-linked markers.

**Figure 1.5. Independent support for the four proposed linkage groups.** This figure depicts cumulative genetic vs. physical distances across the X chromosome. Distances in cM between successive markers that detect polymorphism are summed and plotted against the corresponding distances in Mb (abscissa) from Xpter to Xqter. The broken line is drawn with the average (overall) slope of 1.3 cM/Mb. The approximate locations of the four proposed linkage groups are superimposed on the plot in blue. Red arrows indicate possible hot spots for recombination while the green arrow indicates a region of low recombination rate. The centromere is highlighted with the orange arrow. Figure and portion of legend text reproduced from [233] with coloured labelling added for clarity and emphasis. LG: linkage group.




The X chromosomal STR commercial kits Argus X-UL, X-8, and X-12 were designed with the specialised linkage situation on the X chromosome in mind. Because the chosen markers reside on a single chromosome, the initial four markers were chosen because they were physically far apart and unlinked. Additional loci were chosen specifically based upon their reported linkage to the original markers, creating four linkage trios that could be viewed as haplotypes: DXS8378-DXS10135-DXS10148, DXS7132-DXS10079-DXS10074, DXS10103-HPRTB-DXS10101, and DXS10146-DXS10134-DXS7423. This first trio was proposed for inclusion into the commercial multiplex after confirmation of heterogeneity in a German population and a small recombination study (89 informative meioses) in which the stability of region containing the three proposed markers was assessed

[210]. DXS10074 and DXS10079 were validated for forensic use through a study of their allele structure and recombination rate in a German population [229,234]. Though no recombination was observed during this study, less than 92 informative meioses were examined and linkage disequilibrium was established. In comparison, a study of two Brazilian populations revealed ambiguous results for DXS10079 and DXS10074 and a third marker within a 280-kb region of Xq12, where significant linkage disequilibrium was confirmed in the absence of an indication of significant linkage [110]. Evaluation of DXS10103 and DXS10101 for acceptable heterozygosity and reliable amplification was performed as part of a study of the 133.14-133.45 Mb region of the X chromosome surrounding HPRTB [235]. Haplotype stability of the trio DXS10146-DXS10134-DXS7423 was assessed in a recombination study of less than 109 informative meioses [236]. Though linkage disequilibrium was not tested due to the small sample size, two crossing over events were observed between DXS10146 and DXS10135 out of 80 informative meioses. The authors still recommended these two markers for inclusion into the new commercial kit due to their high degree of polymorphism, pending a more accurate estimation of the genetic distance between them.

In a study of the Argus X-8 kit, Tillmar *et al.* [237] were able to use observed haplotype frequencies to reveal linkage disequilibrium between markers within the same linkage group, but not between markers located in different linkage groups. The study showed that the paternity index would be significantly influenced if this observed linkage disequilibrium was not taken into account. Additionally, 32 families were studied and the recombination fraction between linkage groups 3 and 4 was found to be approximately 25%, indicating non-random assortment. Though it was shown that both linkage and linkage disequilibrium should be taken into account when using X chromosomal STRs, the limited number of informative meioses in these studied families did not allow a detailed picture of the linkage situation.

Lastly, in another recent recombination report, three-generation pedigrees were analyzed at 39 X STR markers [238]. Previous studies were confirmed, including a loose linkage between groups 3 and 4, which could be potentially misleading in certain cases of kinship analysis. The need for larger, collaborative recombination studies was emphasised by these authors.

*1.2.5. Exchange & comparability of data*

Reproducibility both within and between laboratories is a requirement of any forensically useful scientific technique. An important step towards comparability of STR data between laboratories was the recommendation for inclusion of allelic ladders in the typing process [239,240]. Composed of the most common alleles at each locus, the allelic ladder allows consistent identification of alleles across detection platforms, between laboratories, and from run to run within a laboratory. Each allele in the ladder is assigned the appropriate numerical designation according to the nomenclature guidelines for STR markers proposed by the ISFG [43,239,241,242] (see below for further nomenclature discussion). An early assessment of the potential to achieve standardisation of STR testing across laboratories using allelic ladders revealed success typing markers containing simple repeat units across a variety of platforms [243]. Additionally, when employing standardised amplification and detection systems, more complex repeats were suitable for forensic use. Given that dye-labelled amplification primers, allelic ladders, and capillary electrophoresis instruments are almost universal in forensic laboratories today, discrepancies between STR typing results from different laboratories must arise from other sources, most glaringly the lack of standardised nomenclature.

As the number of new markers that are described in the literature increases, the potential for nomenclature differences also increases, making comparisons of published population data between laboratories complicated. Nomenclature discrepancies have been a particular problem on the Y chromosome because of its repetitive nature [43]. The same primer pair, then, can potentially amplify a STR marker present in duplicate at two physically separate locations on the chromosome, as is generally the case for DYS385, or two physically close STRs can fall within the same primer pair, as is the case for DYS389. Gusmão *et al*. [244] determined that for 10 Y STR markers, repetitive units in the flanking region (defined as greater than 7 base pairs from the variable stretch) do not add to the marker's variability and should not be considered in the nomenclature for that particular marker. Similarly, insertions or deletions within this flanking sequence should not be included as part of the repeat size, but rather distinguished with a particular notation as described in

[43]. Several publications have addressed nomenclature concerns specifically for Y STRs [43,241,245].

There are at least 36 markers that are employed by forensic laboratories studying X STRs (Table 1.1), and in some cases, differences in allele nomenclature make comparisons between the laboratories at best tedious and at worst impossible. Though several publications have explicitly addressed these nomenclature discrepancies for certain X STR markers such as DXS7423, DXS8377 [246], and HPRTB [247], other instances still exist. DXS6803, GATA172D05, and GATA31E08 are all examples of X STR markers for which nomenclature variations between publications affects the allele designation (see Chapter 3, section 3.2 for details). Additionally, DXS9902 and DXS7424 are examples of markers for which the nomenclature differences exist despite attempts at adhering to the ISFG guidelines. Publication of corresponding allele sequencing data and/or definition of the repeat structure used in a particular study can ease a portion of this confusion.

Peer-reviewed publication can also aid in further establishing the validity of underlying scientific principles as well as the reproducibility of a specific technique. SWGDAM's Revised Validation Guidelines, for example, require not only the internal validation of a technique in a particular laboratory, but the publication of the developmental validation in order to further address the standard of reproducibility necessary in a forensic laboratory [72]. Additional efforts to foster the exchange of data between laboratories can be seen in the advent of websites designed to compile and disseminate information, such as YHRD [248], the U.S. National Institute of Standards and Technology's (NIST) Short Tandem Repeat database (STRBase) [249], the European DNA Profiling Group's (EDNAP) mtDNA population database (EMPOP) [250] and Forensic ChrX Research [157]. Routine proficiency testing [251], concordance studies [252,253], and other inter-laboratory collaborative exercises [254,255] also further the cause.

*1.3. Study goals*

In order to maximize the potential for forensic, kinship, and missing persons applications, specifically in the United States but more generally across the forensic community, this study sought to address for X chromosomal STRs each aspect of the

developmental validation described here for autosomal and Y chromosomal STR markers. Given the lack of a commercially available X STR amplification kit within the United States, this process must begin with assay design. Target markers for inclusion into a multiplex will be chosen based upon a set of criteria defined by the goals of the analysis: high discriminatory power coupled with simple, efficient analyses. Next, thorough evaluation of the developed multiplexes will provide a robust method with which to genotype relevant population samples, establishing databases which can be relied upon for frequency data. The focus of this portion of the project is an increase in under-represented U.S. population data, but data will also be reported for a population from Bosnia & Herzegovina for the first time. Further assessment of the rate of both mutation and recombination will provide the basis for statistical interpretation of a match between profiles, while comparability of the data across laboratories utilizing X chromosomal markers will be assessed through sequencing of selected alleles and evaluation of a X STR kit available in Europe. Lastly, the potential for the use of X STR markers for additional forensic applications will be investigated through the creation of a mixture multiplex designed to use gonosomal markers to guide interpretation.

# Chapter 2. Materials & Methods

*2.1. Marker selection*

*2.1.1. X chromosomal STR multiplexes*

A review of X chromosomal STR literature resulted in a list of potential markers. Selection was predominantly limited to markers for which there were published population genetic studies in order to exploit the collective knowledge of these established markers and their relevant characteristics, simplifying the process. Forensic utility was assessed according to the following criteria: (a) potential for small amplicon size; (b) heterozygosity in published materials greater than 0.65; and (c) distribution between the four proposed linkage groups across the X chromosome. The list of candidate markers was further limited by selecting those markers that could be electrophoretically separated in a four-dye multiplex. In order to balance number of markers with size of amplicons, two multiplexes were designed.

*2.1.2. Mixture multiplex*

A similar strategy was employed in the generation of a multiplex created to examine the potential of X STRs in mixed samples, termed the MIXplex. Only published markers were considered for inclusion into the mixture multiplex; again, the goal was small amplicons (<200 bp in length) with the intention of maintaining the ability to target potentially degraded samples. A combination of markers located on both sex chromosomes was pursued, including X and Y STRs as well as STRs within the X-Y homologous regions, termed XY markers. A minimum of two markers of each type were targeted for inclusion into the final multiplex, along with SRY. Other requirements included single-copy markers, simple repeat regions, established use within the forensic community, and high heterozygosity (if known).

*2.2. Primer design*

Markers best matching the selection criteria were chosen for inclusion into two multiplexes and organised according to amplicon size. Maximal use of the electrophoretic space across four dye channels within a multiplex was achieved by choosing markers with known allele ranges. Approximately 200 bp flanking either side of the repeat regions for the chosen markers were downloaded from the University of California Santa Cruz (UCSC) Genome Browser [256] using their

BLAT In Silico PCR search [257] and the published primers. In many cases, published amplification primers were sufficient for incorporation into the multiplexes, but several markers required one or both primers be redesigned. When necessary, primers were designed using the web-based program Primer3 [56], and selected primers were screened for use in multiplex reactions using the web-based algorithm AutoDimer [58]. One primer for each marker was labelled at the 5' end with a fluorescent dye, either 6FAM™, VIC®, NED™, or PET™ (Applied Biosystems, Foster City, CA). A tail was added to the complementary primer in the set at the 5' end in order to promote the complete adenylation of PCR products [61] and, in some cases, provide adequate spacing between amplicons in the multiplex. This tail was either 5'-GTTTCTT-3', 5'-ATT-3', or a single G (Chapter 3, Tables 3.3 & 3.4).

*2.3. Sample selection & preparation*

A set of commercially available, highly concentrated control DNA samples with known profiles at each of the markers chosen for inclusion in the various multiplexes were characterised (Table 2.1), as per the recommendations of Szibor *et al.* [258]. One exception was an internally maintained female control DNA designated here as AFDIL-1. Results were compared to published profiles before allele designations were made. This panel of known samples was used for optimisation and quality control testing of all STR multiplexes and allele sequencing reactions, and as positive controls during amplification of unknowns. Additionally, since a large quantity of extract would be needed to optimise the mixture multiplex and later generate and test multiple different mixtures, these control DNAs were a practical and reliable solution for this study as well. All controls were given a sample ID that would allow rapid identification of sex with just a glance at the name (i.e. male 1, male 2, female 1, etc.) for simplicity during the development and testing process.

**Table 2.1. Panel of Control DNAs used for evaluation and quality control experiments.**

| Control DNA | Source | Part Number | Sex | Sample ID |
|---|---|---|---|---|
| Quantifiler® Human DNA Standard | Quantifiler® Human DNA Quantification Kit, Applied Biosystems | 4343895 | Male | Male 1 |
| 9948 | Promega Corporation | DD2061 | Male | Male 2 |
| 2800M | Promega Corporation | DD7101 | Male | Male 3 |
| 9947A | Promega Corporation | DD1001 | Female | Female 1 |
| AFDIL-1 | In-house | NA | Female | Female 2 |
| K562 | Promega Corporation | DD2011 | Female | Female 3 |

Extracts that were part of the large-scale studies using the optimised multiplexes (population database creation, mutation study, linkage study) were collected and processed in high-throughput, 96-well plate format with a witness present at the initial plate creation step.

*2.3.1. Population databases*

In order to study the genetic parameters of the chosen set of X STRs in relevant populations, unrelated, anonymous individuals from four major U.S. populations (African Americans, U.S. Asians, U.S. Caucasians, and U.S. Hispanics) were sought. More information on the sources of these sample sets can be found in Chapter 4 and Table 4.1.

Sample set A represented four U.S. populations: African Americans (174 females, 175 males), U.S. Asians (300 females, 201 males), U.S. Caucasians (146 females, 122 males), and U.S. Hispanics (122 females, 123 males). Bloodstain cards were extracted on the Biomek® 2000 robot (Beckman Coulter, Brea, CA) using the DNA IQ™ system (Promega Corporation, Madison, WI) or on the Qiagen 9604 robot using the Qiagen QIAmp DNA kit (Qiagen, Gaithersburg, MD). Approximate final concentrations of 1 ng/µL were obtained for most of the samples, and no individual normalisation was performed.

Sample set B donors were 701 individuals from four U.S. population groups, African American (260 males), U.S. Asian (3 males), U.S. Caucasian (260 males, 38 females), and U.S. Hispanic (140 males). Blood samples were collected and extracted according to [259] and extracts were provided for typing during this study at a final concentration of 500 pg/µL. Although this sample set had not been typed

with X STRs prior to this study, other marker systems including autosomal STRs [252,259,260], autosomal SNPs [261], Y chromosomal STRs [262], and mitochondrial DNA [263,264] have been completed.

Sample set C extracts were part of a larger cohort of family samples collected to study mutation rates and are described in more detail below (see Section 2.3.2.).

An additional set of samples from a non-U.S. population was also studied. Profiles from a total of 154 (68 female and 86 male) unrelated individuals were analyzed in a population from Bosnia and Herzegovina. After obtaining informed consent, either blood stains or buccal swabs were collected and extracted using the QIAmp DNA Micro Kit (Qiagen GmbH, Hilden, Germany) at the Institute for Genetic Engineering and Biotechnology in Bosnia & Herzegovina; aliquots of these extracts were then brought to the Armed Forces DNA Identification Laboratory (AFDIL) for typing. Test amplifications replaced the quantitation step, and 1 µL of extract was used to amplify all samples from this set.

All samples were collected with Institutional Review Board approval at the respective institutions, and use of these samples for this project was reviewed and approved by the U.S. Army Medical Research and Materiel Command (MRMC) Institutional Review Board Office (IRBO).

*2.3.2. Mutation rate study*

In order to determine the mutation rate of each of the 15 markers included in the two multiplexes utilised here, individuals from a variety of relevant family groups were investigated. X STR alleles that are identical by descent can be unambiguously identified in mother-son pairs, father-daughter pairs, and most family trios. Note that the inheritance pattern of the X chromosome makes trios involving male children equivalent to mother-son pairs for the purposes of mutation rate analyses since the father does not contribute any alleles in this situation. Conversely, it is possible that a family trio involving female children will from time to time result in offspring that are homozygous at a particular marker, making it impossible to tell from which parent the mutation arose. Since this study seeks to define both the rate and the origin of mutation, certain informative family groups will not be helpful in

determining both. Therefore, the final number of samples processed will depend upon the profiles encountered during analyses.

Anonymous extracts from non-probative paternity cases with probability of paternity >99% and at least one child were representative of the three major U.S. populations: African American, U.S. Caucasian, and U.S. Hispanic. Six additional U.S. Caucasian families previously typed at a subset of autosomal STR markers [265] were also included. In total, 958 families (parent-child duos and trios) were included in this study (Table 2.2).

**Table 2.2. Summary of samples examined to determine mutation rate.** The total number of samples analyzed during the mutation rate study is broken down by family type and population. In total, 958 families comprised of 2022 individuals were typed.

| | Population | | | |
|---|---|---|---|---|
| **Type** | **African American** | **U.S. Caucasian** | **U.S. Hispanic** | **U.S. Total** |
| Mother-Son Duos | 147 | 182 | 161 | 490 |
| Mother-Daughter Duos | 11 | 15 | 14 | 40 |
| Father-Daughter Duos | 2 | 2 | 7 | 11 |
| Mother-Father-Daughter Trios | 115 | 167 | 135 | 417 |
| Total Duos & Trios | 275 | 366 | 317 | 958 |
| Total Individuals | 584 | 755 | 683 | 2022 |

Unrelated individuals from this mutation rate study were used to generate a U.S. population database described separately. In total, 314 African American (108 males, 206 females), 434 U.S. Caucasian (165 males, 269 females), and 398 U.S. Hispanic (150 males, 248 females) individuals were used to generate U.S. allele and haplotype frequencies.

Extracts originated from samples processed at the Analytical Genetic Testing Center (Denver, CO) and were provided by Dr. Moses Schanfield of the George Washington University's Department of Forensic Sciences, where the extracts are currently archived. Collection was accomplished by manually aliquoting 5-25 µL extract from individual stock tubes into 96-well plates with a witness to verify sample placement. Samples from mothers and children were organised together into plates separate from samples from fathers; these plates were processed first in order

to determine the sex of the child(ren) involved. Only female children would necessitate processing of associated paternal samples, as a father does not pass an X chromosome on to his male progeny. Additionally, trio samples were organised into plates separate from families with >1 child. A naming convention of PopulationFamily#-Member was used; for example, the father from U.S. Caucasian family trio number 006 would be named CN006-F and the second child from African American family number 511 was named AA511-C2. Family numbers <500 correspond to family trios, while family numbers ≥500 correspond to families with more than one child and/or mother/father.

Sample extracts had concentrations that varied widely but were on the order of tens to hundreds of nanograms per microliter (personal communication with collaborator). Additionally, samples from different years were extracted by different methods, resulting in a difference in average concentration, though it was unknown which year corresponded to which concentration. Therefore a subset of samples from each case year were quantified by a casework analyst using the Quantifiler® Human DNA Quantification Kit (Applied Biosystems) and/or amplified as part of this study at several dilutions in order to determine an appropriate dilution factor to apply to the sample set as a whole. Quantitation values, as well as tested and selected dilution factors (adjusted throughout processing of the sample set as results dictated), are shown in Table 2.3.

**Table 2.3. Sample set C extract dilutions by collection year.** A subset of samples from each collection year was quantified and test amplifications were performed to determine a final dilution factor, which was then modified as additional amplifications indicated. Note that for italicised quantities, associated IPC Ct values were greater than 28 cycles, indicating the quantity may be underestimated. DF: dilution factor. *Space considerations in the dilution plate (Elution Microtubes CL, QIAGEN) allowed a maximum dilution of 1:600, though 1:1000 would have been chosen based upon the test data.

| Collection year | Sample | Quantity (ng/ul) | Test dilutions | Selected DF | Modified DF1 | Modified DF2 |
|---|---|---|---|---|---|---|
| 1995 | Sample 1 | *1781.44* | 1:100, 1:1000 | 1:600** | 1:500 | |
| | Sample 2 | *1599.4* | 1:100, 1:1000 | | | |
| | Sample 3 | *1857.09* | 1:100, 1:1000 | | | |
| | Sample 4 | *1443.3* | 1:100, 1:1000 | | | |
| | Sample 5 | *1041.45* | 1:100, 1:1000 | | | |
| | Sample 5 1:50 | 20.6 | | | | |
| | Sample 6 | *2206.32* | 1:100, 1:1000 | | | |
| | Sample 6 1:100 | 13.18 | | | | |
| | Sample 7 | | 1:100, 1:500, 1:1000 | | | |
| | Sample 8 | | 1:100, 1:500, 1:1000 | | | |
| | Sample 9 | | 1:100, 1:500, 1:1000 | | | |
| 1998 | Sample 1 | | 1:100, 1:500, 1:1000 | 1:600** | 1:500 | |
| | Sample 2 | | 1:100, 1:500, 1:1000 | | | |
| | Sample 3 | | 1:100, 1:500, 1:1000 | | | |
| 1999 | Sample 1 | | 1:100, 1:500, 1:1000 | 1:600** | 1:500 | |
| | Sample 2 | | 1:100, 1:500, 1:1000 | | | |
| | Sample 3 | | 1:100, 1:500, 1:1000 | | | |
| 2000 | Sample 1 | | 1:100, 1:500, 1:1000 | 1:600** | 1:350 | |
| | Sample 2 | | 1:100, 1:500, 1:1000 | | | |
| | Sample 3 | | 1:100, 1:500, 1:1000 | | | |
| 2001 | Sample 1 | *158.41* | 1:100, 1:1000 | 1:600** | 1:250 | 1:350 |
| | Sample 1 1:50 | 3.23 | | | | |
| | Sample 2 | *80.53* | 1:100, 1:1000 | | | |
| | Sample 2 1:100 | 0.866 | | | | |
| | Sample 3 | *171.08* | 1:100, 1:1000 | | | |
| | Sample 4 | *196.03* | 1:100, 1:1000 | | | |
| | Sample 5 | *131.46* | 1:100, 1:1000 | | | |
| | Sample 6 | *65.34* | 1:100, 1:1000 | | | |
| | Sample 7 | | 1:100, 1:500, 1:1000 | | | |
| | Sample 8 | | 1:100, 1:500, 1:1000 | | | |
| | Sample 9 | | 1:100, 1:500, 1:1000 | | | |
| 2002 | Sample 1 | | 1:100, 1:500, 1:1000 | 1:100 | 1:250 | |
| | Sample 2 | | 1:100, 1:500, 1:1000 | | | |
| | Sample 3 | | 1:100, 1:500, 1:1000 | | | |
| 2003 | Sample 1 | | 1:100, 1:500, 1:1000 | 1:100 | 1:250 | |
| | Sample 2 | | 1:100, 1:500, 1:1000 | | | |
| | Sample 3 | | 1:100, 1:500, 1:1000 | | | |

### *2.3.3. Recombination study*

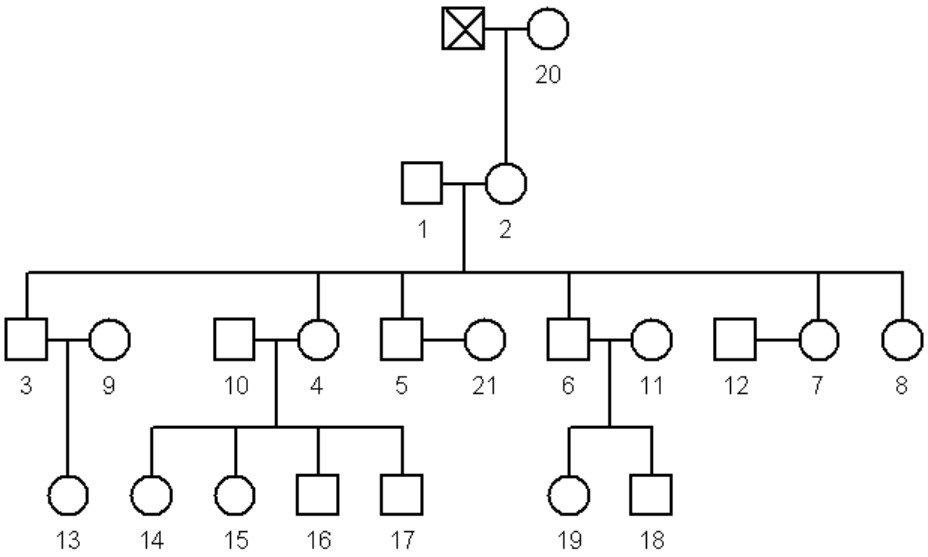
Before any test of linkage was initiated, the appropriate sample sets were identified for analyses. Such sets included the following family types:

Type I: three-generation families typically including a maternal grandfather, mother, and her child(ren); these families include, at a minimum
A maternal grandfather, a mother, and her son
A maternal grandfather, a mother, a father, and their daughter

Type II: two-generation families including a mother and two or more of her children; this type can include several different scenarios such as
A mother and multiple sons
A mother, a father, and two or more daughters
A mother, a father, one son, and one daughter

Complex pedigrees including several generations and multiple offspring could therefore include multiple different families as defined above; see Figure 2.1 for an example.

**Figure 2.1. Multiple family types present within one pedigree.** This example of a pedigree used for linkage analysis (FamID 6) contains distinct sub-families of both type I and type II, as described in the accompanying table. Complex pedigrees such as this one were divided into their respective sub-families for analysis.




| FamID | Family type | Individuals included | Description |
|---|---|---|---|
| 6A | II | 1, 2, 3, 4, 5, 6, 7, 8 | Mother and father with multiple children |
| 6B | II | 6, 11, 18, 19 | Mother and father with son and daughter |
| 600 | I | 1, 2, 4, 10, 14, 15, 16, 17 | Maternal grandparents, mother & father, multiple children |

A set of commercially-available family extracts satisfying these requirements was purchased from the NIGMS repository. This collection of DNA from lymphoblastic cell lines included CEPH families from Utah, France, Venezuela, and an Old Order

Amish family from Pennsylvania. Additionally, six U.S. Caucasian families previously typed at a subset of autosomal STR markers [265] and also used as part of the mutation rate study were included, as well as a subset of other families from the mutation rate study. In total, 158 families (54 Type I and 104 Type II) that were appropriate for linkage analysis according to the criteria defined above were identified within this dataset.

*2.4. PCR amplification & detection*

Experiments were performed to optimise a variety of important aspects of the amplification and detection process, resulting in a final set of robust parameters. For these initial experiments, amplification was performed in a 10 µL reaction that consisted of 1x PCR buffer II (Applied Biosystems), 1, 1.5, or 2 units Ampli*Taq* Gold™ DNA polymerase (Applied Biosystems), 0.25 mM dNTP Mix (Applied Biosystems), 0.15 mg/mL bovine serum albumin (BSA, Sigma-Aldrich, St. Louis, MO), 2 mM magnesium chloride solution (Applied Biosystems) and 1 or 2 µL of primer mix. Primer mix concentrations were adjusted empirically to balance peak heights within each multiplex. Each preparation of primer mix was tested on a panel of known control samples to ensure consistent quality and correct genotypes could be obtained before use on unknowns.

Annealing temperatures of 55, 56, 57, 58, 59, and 60 $^{o}$C were evaluated with extension times of 60 and 90 minutes on a GeneAmp® 9700 (Applied Biosystems). Final optimised thermal cycling parameters were as follows: initial incubation at 96 $^{o}$C for 10 minutes, amplification with 30 cycles of 94 $^{o}$C for 1 minute, 60 $^{o}$C for 1 minute, and 72 $^{o}$C for 1 minute, extension at 60 $^{o}$C for 45 minutes, and final soak at 4 $^{o}$C.

AmpF*l*STR® Identifiler® and Investigator Argus X-12 (QIAGEN) amplifications were performed according to manufacturers' recommended protocols [82,266]. Though the X-12 kit was not available for purchase within the United States at the time of this study, a collaboration with QIAGEN and the National Institute of Standards and Technology allowed access to a limited number of reactions for the purposes described here (see Chapter 6 for more details).

Samples were prepared for capillary electrophoresis by adding1 µL amplified product to 8.7 µL Hi-Di™ formamide (Applied Biosystems) and 0.3 µL LIZ-500 size standard (Applied Biosystems). Samples were injected at 3.0 kV for 5, 10, or 20 seconds and run using a 36 cm array and POP-6™ Polymer on a 3130xl Genetic Analyzer (Applied Biosystems).

Electrophoretic data were analyzed using GeneMapper® ID-X version 1.2 or 1.3 (Applied Biosystems) with custom bins and panels or those provided by the manufacturer (for the commercial kits). New alleles were inferred based upon electrophoretic mobility and, when possible, sequenced to confirm repeat structure and length according to the protocol described in [55]; sequencing was also performed on all samples that exhibited null alleles. Electronic transfer of allele calls from GeneMapper® export files to a master file combining alleles from both multiplexes by sample was accomplished through the use of a custom macro in order to reduce the possibility of transcription errors. A single marker (DXS9902) was included in both X STR multiplexes and compared during analysis to ensure concordance for each sample.

*2.5. DNA sequencing*

In order to fully characterise both the number of repeating units and flanking sequence, a set of sequencing primers located outside the target STR amplicon was designed as outlined above. Each set of primers was then tested on male control DNA samples to confirm high quality sequence data results. If sequence data were unacceptable, additional primers were designed and retested until high quality sequence data were obtained.

Template DNA was amplified in a 50 µL reaction consisting of 1x PCR buffer, 0.4 µM each forward and reverse primer (Applied Biosystems), 0.2 mM dNTPs, and 2.5 units Ampli*Taq* Gold™ DNA polymerase. Thermal cycling was performed on a GeneAmp® 9700 using the following parameters: initial incubation at 96 $^{o}$C for 10 minutes, amplification with 36 cycles of 94 $^{o}$C for 30 seconds, 56 $^{o}$C for 30 seconds, and 72 $^{o}$C for 1 minute, extension at 72 $^{o}$C for 7 minutes, and final soak at 4 $^{o}$C. Successful amplification was confirmed on a 1% agarose gel, and PCR products

were then treated with ExoSAP-IT® (USB Corporation, Cleveland, OH) to remove excess primers and dNTPs.

When necessary, heterozygous alleles in female samples were separated using a 4% agarose gel and 10 µL amplification product run at ~110 volts for 45 minutes to 2 hours. Separated bands were excised from the gel and purified using the QIAquick Gel Extraction Kit (Qiagen) using the manufacturer's protocol. Extracted DNA was then re-amplified according to the protocol described above, and the presence of a single amplification product was confirmed via agarose gel before treatment with ExoSAP-IT®.

Cycle sequencing was performed using the BigDye® Terminator v1.1 kit (Applied Biosystems). The sequencing reactions consisted of 8 µL $dH_2O$, 6 µL 2.5x sequencing buffer (400 mM Tris, 10 mM $MgCl_2$, pH 9.0), 1.5 µL BigDye® Terminator v1.1 Ready Reaction Mix, 0.5 µL dGTP mix, 2 µL 10 mM forward or reverse primer, and 2 µL amplified product. Thermal cycling was performed on a GeneAmp® 9700 using the following parameters: initial incubation at 96 $^{o}$C for 1 minute, amplification with 25 cycles of 96 $^{o}$C for 15 seconds, 50 $^{o}$C for 5 seconds, and 60 $^{o}$C for 2 minutes, and final soak at 4 $^{o}$C.

Sequencing products were purified using a Performa® DTR 96-well plate (Edge Biosystems, Gaithersburg, MD) and 1 µL filtrate was added to 9 µL Hi-Di™ formamide for injection onto the 3130xl or 3730 instruments using either POP-6™ polymer and a 36 cm array or POP-7™ polymer and a 50 cm array, respectively. Sequences were aligned to a GenBank reference sequence and analyzed using Sequencher® version 4.1, 4.7, or 4.8 (GeneCodes, Ann Arbor, MI).

*2.6. Sensitivity testing*

Sensitivity testing was performed to evaluate the lower limits of the various multiplexes. For the two X STR assays, the initial concentration of three female and two male samples was determined by a casework analyst using the Quantifiler® Human DNA Quantification Kit. Samples were then serially diluted with Tris-low-EDTA buffer (TLE; 10 mM Tris, 0.1 mM EDTA, pH 8.0) to form the following dilution series: 1000, 500, 200, 100, 50, and 25 pg/µL. One microliter of each

concentration was tested in duplicate to determine the minimum quantity of input DNA required to reliably obtain full profiles for both males and females with each multiplex.

Additionally, an evaluation of the performance of the two multiplexes was performed on samples that mimicked those typically received at the AFDIL. Five bone samples greater than 65 years old were tested to determine the multiplexes' utility with potentially degraded specimens. DNA was extracted from bones using a procedure that results in the total demineralisation of the bone matrix [267]. Previously, PowerPlex® 16 (Promega) testing with a modified protocol (increased polymerase concentration and increased PCR cycle number) was performed on these bone samples as part of a separate study, and dropout at many of the larger loci (>300 bp) was observed, indicating DNA degradation. For mini-X STR testing, the total volume of the amplification reaction was increased to allow greater extract input volume and an increased concentration of polymerase. All volumes were adjusted proportionally from the standard volume (10 µL) to 25 µL, with up to 8 units Ampli*Taq* Gold™ DNA polymerase per reaction.

Sensitivity testing was also performed to evaluate the lower limits of the mixture multiplex with single-source samples. The initial concentration of two female and one male sample was determined by a casework analyst using the Quantifiler® Human DNA Quantification Kit. Samples were then serially diluted with TLE to form the following dilution series: 1000, 500, 200, 100, 50, and 25 pg/µL. One microliter of each concentration was tested in triplicate to determine the minimum quantity of input DNA required to reliably obtain full profiles for both male and female samples.

*2.7. Population Genetic Analyses*

Allele frequencies were calculated by hand using a spreadsheet program. The software PowerMarker [155] was used to determine both the observed heterozygosity value (H(obs)) and the p value of the exact test for Hardy–Weinberg equilibrium (p(HWE)) using only the female genotype data. Since both parameters depend upon a diploid dataset, neither value was calculated for populations that were predominantly male. All other forensic efficiency statistics (H(exp), PIC, PDm, PDf,

MECI, and MECII) were calculated using the Forensic ChrX Research website version 2.0 [157]. The formulae required for these calculations are detailed in Table 2.4. Pairwise genetic distance calculations were performed either via the chi square test or in Arlequin v3.1 or v3.5 [156]. Specifically, Arlequin was used to compute pairwise $F_{ST}$ and corresponding p values using 10,000 permutations and a significance level of 0.05. Linkage disequilibrium between all pairs of loci was assessed either within PowerMarker or using Arlequin and the default settings.

**Table 2.4. Genetic formulae required for the calculation of forensic efficiency parameters.** *X STR formula applies only to trios involving daughters.

| Parameter | General Formula | X STR Formula (if different) |
|---|---|---|
| Mean exclusion chance in trios (MECI)* | $\sum_i fi^3(1-fi)^2 + \sum_i fi(1-fi)^3 + \sum_{i<j} fifj(fi+fj)(1-fi-fj)^2$ | 1. $\sum_i fi^3(1-fi) + \sum_i fi(1-fi)^2 + \sum_{i<j} fifj(fi+fj)(1-fi-fj)$ [268] |
| | | 2. $1-\sum_i f_i^2 + \sum_i f_i^4 - \left(\sum_{i<j} f_i^2\right)^2$ [269] |
| Mean exclusion chance in father/ daughter duos (MECII) | NA | $1-2\sum_i f_i^2 + \sum_i f_i^3$ |
| Power of discrimination in females (PDf) | $1-2\left(\sum_i f_i^2\right)^2 + \sum_i f_i^4$ | Same |
| Power of discrimination in males (PDm) | $1-2\left(\sum_i f_i^2\right)^2 + \sum_i f_i^4$ | $1-\sum_i f_i^2$ |
| Expected heterozygosity (H(exp)) | $1-\sum_i f_i^2$ | Same |
| Polymorphism information content (PIC) | $1-\sum_i f_i^2 - \left(\sum_i f_i^2\right)^2 + \sum_i f_i^4$ | Same |
| Power of exclusion (PE) | $H^2(1-(1-H)H^2)$, H = H(exp) | Same |
| Paternity index (PI) | $\frac{1}{2\sum_i f_i^2}$ | Same |

*2.8. Mutation Rate Analysis*

Mutations within families were identified by comparison to appropriate parent(s) using a spreadsheet program. Extracts from all families in which a potential mutation was discovered were sequenced at the affected marker in order to confirm

profiles and rule out the presence of null alleles. Additionally in families with mutations, autosomal STR results were obtained using the AmpF*l*STR® Identifiler® PCR Amplification Kit according to the manufacturer's recommended protocol in order to confirm relationships. Custom software known as Laboratory Information Systems Applications (LISA, Future Technologies, Inc., Fairfax, Virginia) was used to determine the parentage index in each case using a one parent-one child calculation.

X STR mutation rates were calculated as the number of mutations divided by the total number of meioses. Of note, the resultant mutation rate is an estimation of the true rate, as only observed mutations were considered during this study. Confidence intervals (CI) were estimated using the exact binomial distribution [270] via spreadsheet formulae provided at http://statpages.org/confint.html. For the purposes of this study, overall mutation rate refers to the general X STR mutation rate resulting from observed meioses at more than one marker, while the marker-specific rate describes the rate at only a single marker.

In order to confirm the findings of this study by comparison as well as compile a robust large-scale dataset for use by the forensic community, a literature review of published mutation rate studies available at the time of manuscript preparation was conducted. Data mining necessary to compile the included list of published and combined (this study plus published) mutation rates by marker was accomplished by recalculating the mutation rate within 34 independent studies [87,89,91,93-95,98,99,122,146,154,158,161,164,165,167,193,202-218] according to the described number of mutations and meioses broken down by marker. Individual studies (and, for the combined dataset, this study) were then combined for each marker to determine both the marker-specific and overall mutation rates, and confidence intervals were assigned to these pooled values. Studies in which there was any ambiguity as to the exact value of the parameters necessary for these calculations were excluded. A complete list of published mutation rate studies can be found in Table 1.3.

*2.9. Recombination rate assessment & linkage analysis*

Once appropriate sample sets were identified and typed, analyses were performed using two different methods. To begin, the direct method utilized a pairwise comparison between adjacent markers and phased source chromosome. This method was particularly suited to analysis of X markers since the recombination events could be observed directly. However, only Type I families could be analyzed this way owing to the inability to unambiguously determine the phase of each of the mother's X chromosomes without the maternal grandfather's profile. The general steps were as follows:

1. Use the maternal grandfather's profile to determine the paternal contribution to mother's profile and deduce maternal contribution.
2. Use father's profile to assign and remove the paternal contribution from analysis of daughters.
3. Compare maternal contribution (source chromosome) in each of the children to the mother's two chromosomes, looking specifically for instances where alleles of adjacent markers resulted from different chromosomes, therefore indicating a potential recombination event had occurred.

Each observed recombination event was noted and totalled for each marker pair. This total was then divided by the total number of unambiguous meioses for that marker pair, producing the observed recombination rate. An example detailing this manual analysis is presented in Appendix A. This analysis was then followed with a manual (analytical) calculation of the logarithm of the odds (LOD) scores (Z) using the following formula:

$$Z = \frac{\log_{10}((1-\theta)^{NR}\theta^{R})}{0.5^{NR+R}}$$

where $\theta$ is the recombination rate

NR represents the number of meiosis for which no recombination was observed (or were ambiguous)

R represents the number of meiosis for which recombination was observed.

The second analysis method was performed as described by Nothnagel *et al.* [218] with modifications on the front-end specific to this dataset. A PLINK format text file was created by assigning the following to each sample: family identification number (FamID), patient identification number (PID), father's identification number (FID), mother's identification number (MID), sex (1 for male and 2 for female), phenotype (not used; coded zero for all samples) followed by 2 alleles for each of the 15 X STR markers. The second allele in male samples and any markers with missing allelic information were coded with a zero as a placeholder.

The design of the scripts used to perform likelihood calculations in this strategy limited the family structure to only those with male children; that is, type I families with a maternal grandfather, mother, and one or more sons, or a type II family with a mother and >1 son. Since the dataset included many families that did not conform to this structure, it was necessary to further modify the data before analysis to maximise the overall number of meiosis that could be included. This process was similar to that described for the second step of the manual analysis above; fathers' profiles were used to manually remove the paternal contribution from the daughter's profiles, effectively turning these daughters into sons. In doing this, several families required separation into multiple distinct families to allow a female profile to serve as both a daughter-turned-son as well as a mother. This was the case, for example, with the family shown in Figure 2.1, where individual 4 served as a mother in family 600 and a daughter-turned-son in family 6A. Also, any paternal mutations discovered during this process were catalogued.

Since this process of converting the daughters' profiles to mimic sons required manual manipulation of the data, additional quality control measures were incorporated to ensure the resulting dataset was error-free. To begin, all meioses were screened for mutations using a spreadsheet program. In particular, for daughters-turned-sons and their mothers, a mutation could indicate either a mistake in the process of removing the paternal contribution or a true maternal mutation. Mistakes were corrected by referring to the original unedited profiles, and true mutations were categorised as maternal, paternal, or unknown origin. Mutations with unknown origin discovered in daughters-turned-sons were considered paternal in order to allow inclusion in the linkage analysis. Since the linkage analysis scripts

also had the limitation of being unable to tolerate mutations of >1 step or between maternal grandfathers and mothers, the markers in these rare cases were rendered uninformative for linkage by assigning either a 0,0 genotype in the affected individuals or a homo- or hemizygous genotype to all individuals at that particular marker. Other mutations that did not have an impact on linkage analysis (between a maternal grandmother and mother, for example) or could be determined to be maternal were maintained in the dataset.

Lastly, duplications and triplications were reduced to the appropriate allele number by examining parental profiles and using a zero designation at any ambiguous or undeterminable loci. This file was submitted for analysis to the authors of the referenced publication, who made several modifications to the original scripts to accommodate this larger set of markers. Additionally, since this method allowed for incorporation of the mutation rate during the calculation of recombination rate, the value obtained during the mutation rate study performed as part of this work was used (see Section 5.1).

*2.10. Mixture testing*

In order to efficiently evaluate the effectiveness of the mixture multiplex (MIXplex), control DNA samples with known profiles for the included markers were chosen. By doing so, allele sequencing steps necessary to confirm the allele call for each sample at each marker were avoided, and published allele calls could be relied upon instead. Predictions could also then be made concerning the outcome of mixture testing with each pair of control DNAs prior to amplification, allowing comparisons between expected and observed results.

Six control DNAs (Table 2.1) were quantified by a casework analyst using the Quantifiler® Human DNA Quantification Kit and normalised to 1 ng/µL using TLE. A subset of two male (Quantifiler® standard, 2800M) and two female (AFDIL-1, K562) samples were chosen to create 4 male-female mixtures, 1 male-male mixture, and 1 female-female mixture at the following ratios: 0:100, 10:90, 20:80, 30:70, 40:60, 50:50, 60:40, 70:30, 80:20, 90:10, 100:0. One microliter of each mixture at each ratio was amplified in duplicate or triplicate with the MIXplex and once with

the AmpF*l*STR® Identifiler® autosomal STR amplification kit according to the manufacturer's recommended protocol.

In additional to the creation of these artificial mixtures used to test the performance of the MIXplex assay, the profiles of all six controls were used to generate 63 theoretical mixture profiles for all possible combinations of male(s)-female(s), multiple males, and multiple females. These profiles were then coded with generic numeric identifiers and randomised in order to mask the sex and number of contributors to the profile, and used to test the theoretical ability of the MIXplex compared to the Identifiler® kit. Table 2.5 lists both the artificial and theoretical mixtures and their components.

**Table 2.5. Naming conventions and components of mixtures created for evaluation of MIXplex.** Samples/mixtures in bold were prepared and amplified while the others' profiles were theoretically determined based on known profiles of the individual contributors.

| Sample identifier | Profile identifier | Quantifiler® Standard (male) | 9948 (male) | 2800M (male) | 9947A (female) | AFDIL-1 (female) | K562 (female) |
|---|---|---|---|---|---|---|---|
| **female 1** | **123** | | | | **X** | | |
| **female 2** | **113** | | | | | **X** | |
| **female 3** | **152** | | | | | | **X** |
| **male 1** | **106** | **X** | | | | | |
| **male 2** | **133** | | **X** | | | | |
| **male 3** | **138** | | | **X** | | | |
| mixture 1-A1 | 136 | X | | | X | | |
| **mixture 1-A2** | **118** | **X** | | | | **X** | |
| **mixture 1-A3** | **135** | **X** | | | | | **X** |
| mixture 1-A4 | 149 | | X | | X | | |
| mixture 1-A5 | 119 | | X | | | X | |
| mixture 1-A6 | 137 | | X | | | | X |
| mixture 1-A7 | 109 | | | X | X | | |
| **mixture 1-A8** | **139** | | | **X** | | **X** | |
| **mixture 1-A9** | **148** | | | **X** | | | **X** |
| mixture 1-F1 | 160 | | | | X | X | |
| mixture 1-F2 | 163 | | | | X | | X |
| **mixture 1-F3** | **107** | | | | | **X** | **X** |
| mixture 1-M1 | 108 | X | X | | | | |
| **mixture 1-M2** | **140** | **X** | | **X** | | | |
| mixture 1-M3 | 121 | | X | X | | | |
| mixture 3-A1 | 141 | X | X | | X | | |

Table continues on next page

| Sample identifier | Profile identifier | Quantifiler® Standard (male) | 9948 (male) | 2800M (male) | 9947A (female) | AFDIL-1 (female) | K562 (female) |
|---|---|---|---|---|---|---|---|
| mixture 3-A2 | 120 | X | X | | | X | |
| mixture 3-A3 | 147 | X | X | | | | X |
| mixture 3-A4 | 146 | X | | X | X | | |
| mixture 3-A5 | 145 | X | | X | | X | |
| mixture 3-A6 | 142 | X | | X | | | X |
| mixture 3-A7 | 110 | | X | X | X | | |
| mixture 3-A8 | 122 | | X | X | | X | |
| mixture 3-A9 | 134 | | X | X | | | X |
| mixture 3-B1 | 143 | X | | | X | X | |
| mixture 3-B2 | 101 | | X | | X | X | |
| mixture 3-B3 | 132 | | | X | X | X | |
| mixture 3-B4 | 111 | X | | | X | | X |
| mixture 3-B5 | 131 | | X | | X | | X |
| mixture 3-B6 | 117 | | | X | X | | X |
| mixture 3-B7 | 124 | X | | | | X | X |
| mixture 3-B8 | 112 | | X | | | X | X |
| mixture 3-B9 | 103 | | | X | | X | X |
| mixture 3-F | 144 | | | | X | X | X |
| mixture 3-M | 125 | X | X | X | | | |
| mixture 4-A1 | 102 | X | X | X | X | | |
| mixture 4-A2 | 151 | X | X | X | | X | |
| mixture 4-A3 | 157 | X | X | X | | | X |
| mixture 4-B1 | 150 | X | | | X | X | X |
| mixture 4-B2 | 114 | | X | | X | X | X |
| mixture 4-B3 | 126 | | | X | X | X | X |
| mixture 4-C1 | 128 | X | X | | X | X | |
| mixture 4-C2 | 116 | X | X | | X | | X |
| mixture 4-C3 | 127 | X | X | | | X | X |
| mixture 4-C4 | 156 | X | | X | X | X | |
| mixture 4-C5 | 153 | X | | X | X | | X |
| mixture 4-C6 | 104 | X | | X | | X | X |
| mixture 4-C7 | 129 | | X | X | X | X | |
| mixture 4-C8 | 158 | | X | X | X | | X |
| mixture 4-C9 | 161 | | X | X | | X | X |
| mixture 5A | 105 | X | X | X | X | X | |
| mixture 5B | 130 | X | X | X | X | | X |
| mixture 5C | 155 | X | X | X | | X | X |
| mixture 5D | 159 | X | X | | X | X | X |
| mixture 5E | 162 | X | | X | X | X | X |
| mixture 5F | 154 | | X | X | X | X | X |
| mixture 6P | 115 | X | X | X | X | X | X |

# Chapter 3. X Chromosomal Short Tandem Repeat Multiplex Assay Development

See also Appendix C for the following publication:

C1. Toni M. Diegoli, Michael D. Coble, Development and characterization of two mini-X chromosomal short tandem repeat multiplexes, Forensic Sci. Int. Genet. 5 (2011) 415-421.

*3.1. Assay Design*

Organisation and selection of the initial set of markers for investigation were based upon forensic utility as described previously in the literature (small amplicon size, heterozygosity values, and distribution among the proposed linkage groups) as well as practical considerations (arrangement within two multiplexes, spacing between allele ranges for markers within the same dye channel, primer compatibility, successful amplification, etc.); nineteen candidate markers were identified (Table 3.1).

**Table 3.1. List of potential X STR markers for inclusion in two mini-STR multiplexes.** bp: base pairs. *According to Refs. [157,209,271].

| Marker name | Published amplicon size range (bp) | Heterozygosity (published) | Linkage group* |
|---|---|---|---|
| DXS7423 | 99-115 | 0.734 | 4 |
| GATA165B12 | 90-110 | 0.630 | 3 |
| DXS7133 | 76-100 | 0.658 | 2 |
| DXS8378 | 95-111 | 0.714 | 1 |
| DXS6803 | 105-128 | 0.812 | 2 |
| DXS9895 | 139-161 | 0.704 | 1 |
| DXS101 | 142-169 | 0.885 | 2 |
| DXS6789 | 122-162 | 0.746 | 2 |
| DXS8377 | 204-261 | 0.922 | 4 |
| DXS7130 | 89-124 | 0.788 | 1 |
| DXS7424 | 79-100 | 0.836 | 2 |
| DXS6801 | 113-137 | 0.626 | 2 |
| DXS7132 | 131-155 | 0.883 | 2 |
| GATA172D05 | 108-136 | 0.804 | 2 |
| GATA31E08 | 101-133 | 0.804 | 4 |
| HPRTB | 147-179 | 0.919 | 3 |
| DXS9902 | 160-176 | 0.743 | 1 |
| DXS9898 | 185-217 | 0.745 | 2 |
| DXS6800 | 194-218 | 0.694 | 2 |

Singleplex amplifications were performed first to evaluate primer selection for successful amplification, complete adenylation, and peak migration. At this stage,

primer sets were redesigned when necessary to address practical issues, and unlabelled sequencing primers were designed for additional troubleshooting and quality control purposes (Table 3.2). Various combinations of STR amplification primers were then re-tested in singleplex when necessary to determine the optimal set with which to proceed to multiplex testing. A list of all labelled STR amplification primers designed and tested during this process is detailed in Table 3.3.

**Table 3.2. Unlabelled primers used for amplification and sequencing of X STR alleles.** F: forward primer; R: reverse primer.

| Marker Name | Primer Sequence (5'-3') | |
|---|---|---|
| DXS6789 | F | TCAAGCTTGCAGACAGCCTA |
| | R | TCGAAAAGATAGCCAATCACTG |
| DXS7130 | F | GCCCATGGAGCTATCTTGAA |
| | R | ATCAGCCTGAAATGCTTTGG |
| GATA31E08 | F | AGCAAGGGGAGAAGGCTAGA |
| | R | TCAGCTGACAGAGCACAGAGA |
| DXS7424 | F | AGTTATGCCAGCCACTGGAC |
| | R | TCATCCAGGGTTCATAGTGTCA |
| GATA165B12 | F | TTGACAACAGATTTCTAAGCCAAG |
| | R | AATCATTTTCACTGTGTATGCTTT |
| DXS101 | F | CAGAAAAAGCCCTCAGCAAA |
| | R | GCAAGGGAAGGGATAGCATT |
| DXS7133 | F | CACTTCCAAAAGGGGAAAAA |
| | R | CTTGTACTTGGTGGGAGGAA |
| DXS6795 | F | TTCATGCTGTTGCTTTCCAG |
| | R | CCATCCCCTAAACCTCTCAT |
| GATA172D05 | F | GTGGTTACCAGGGACTGGAG |
| | R | AAACAGCATGGTGGTTCCTC |
| DXS10147 | F | TCCCTTCAACCTAGGAGGTG |
| | R | GCAGATAATGACCAGGACAGG |
| | F2 | AAAATTAGCCAGGCATGGTG |
| | R2 | AGCAAAACACAGAGGGCAGT |
| | F3 | TCACACCAGTAATCCCAGCA |
| DXS8378 | F | TTGCAGTCCTACGCTTTTCC |
| | R | GGGTTCTGGGCTGTAGCATA |
| DXS7132 | F | CAGATTTGAATTGGGCTAACCT |
| | R | TGGACAATCAGTGCTTTCTG |
| DXS6803 | F | TGCACACGTATCCTGGAATTT |
| | R | TGTTAAACAGGCAAATGAAAACT |
| HPRTB | F | CTCTCCAGAATAGTTAGATGTAGGT |
| | R | ATGCCACAGATAATACACATCCCC |
| DXS7423 | F | TGCGAGCCCACTCTTTCTAT |
| | R | TGGCCTTTGTCTCCAGTACC |
| DXS9902 | F | CGAAACGCGCTATCTAAAGG |
| | R | CACATCCTGCACATGTACCC |

**Table 3.3. STR amplification primer sequences tested for 19 candidate X STR markers and SRY.** Bases in bold are tails added to the primer sequence to promote complete adenylation of the amplicon. [a]Primer from [272] but modified to match GenBank sequence in this study through addition of bases in brackets.

| Marker | Primer name | Primer sequence (5'-3') | Reference |
|---|---|---|---|
| DXS7424 | DXS7424 F | 6FAM-AAAACAGGAAGACCCCATC | [54] |
| | DXS7424 Fc | 6FAM-AACACAGGAAGACCCCATC | This study |
| | DXS7424 Fc.0 | ATTAACACAGGAAGACCCCATC | This study |
| | DXS7424 F2 | NED-GGACTGCTTGAGTCCAGGAA | This study |
| | DXS7424 R | **ATT**GGCTAAGAAGAATCCCGCACA | [54] |
| | DXS7424 Rc | **ATT**GCTAAGAAGAATCCCGCACA | [54] |
| | DXS7424 Rc.G | **G**GCTAAGAAGAATCCCGCACA | [54] |
| | DXS7424 Rc.0 | 6FAM-GCTAAGAAGAATCCCGCACA | [54] |
| | DXS7424 R2 | **G**GGAACACGCACATTTGAGAA | This study |
| DXS6789 | DXS6789 F | 6FAM-CCTCGTGATCATGTAAGTTGG | [54] |
| | DXS6789 R | **ATT**GCAGAACCAATAGGAGATAGATGGT | [54] |
| | DXS6789 Rc | **ATT**CAGAACCAATAGGAGATAGATGGT | [54] |
| DXS7130 | DXS7130 F | VIC-AATATAGAGGAAGGGGAAATCATTA | This study |
| | DXS7130 R | ATCAGCCTGAAATGCTTTGG | This study |
| | DXS7130 R2 | **ATT**CAAAGAAATGAGAACAAAAATCAGG | This study |
| | DXS7130 R3 | **ATT**TCAAAGAAATGAGAACAAAAATCA | This study |
| | DXS7130 R4 | **ATT**AAAGAAATGAGAACAAAAATCAGG | This study |
| DXS7423 | DXS7423 F | NED-AGATTTCCTCCCCATCCATC | [54] |
| | DXS7423 Fv3 | PET-AGATTTCCTCCCCATCCATC | [54] |
| | DXS7423 R | **ATT**GTTGTCACACAAATAAATGAATGAGT | [54] |
| | DXS7423 Rc | **ATT**TTGTCACACAAATAAATGAATGAGT | [54] |
| GATA165B12 | GATA165B12 F | PET-TCATCAATCATCTATCCGTATATCA | [54] |
| | GATA165B12 R | **ATT**GAAGTTGACTGTGATTCCTGGTTT | [54] |
| | GATA165B12 Rc | **ATT**AAGTTGACTGTGATTCCTGGTTT | [54] |
| DXS101 | DXS101 F | PET-TCTCCCTTCAAAAACAAAGATAA | [54] |
| | DXS101 F2 | PET-TCAGTCCAAATATCTCCCTTCAA | This study |
| | DXS101 F.0 | **ATT**TCTCCCTTCAAAAACAAAGATAA | [54] |
| | DXS101 F2.0 | **ATT**TCAGTCCAAATATCTCCCTTCAA | This study |
| | DXS101 R | **ATT**GTGCATATTCTGCGCATGT | [54] |
| | DXS101 Rc | **ATT**TGCATATTCTGCGCATGT | [54] |
| | DXS101 R2 | **ATT**GCGCATGTATCCCAGAACTT | This study |
| | DXS101 Rc.0 | PET-TGCATATTCTGCGCATGT | [54] |
| DXS7133 | DXS7133 F | 6FAM-AGCTTCCTTAGATGGCATTCA | [54] |
| | DXS7133 R | **ATT**GTTTTTAACGGTGTTCATGCTT | [54] |
| | DXS7133 Rc | **ATT**TTTTTAACGGTGTTCATGCTT | [54] |
| GATA172D05 | GATA172D05 F | 6FAM-TAGTGGTGATGGTTGCACAG | [272] |
| | GATA172D05 Ftail | 6FAM-**GTTTCTT**TAGTGGTGATGGTTGCACAG | [272] |
| | GATA172D05 R | **GTTTCTT**ATAATTGAAAGCCCGGATTC | [272] |
| DXS8378 | DXS8378 F | VIC-GCTCCTGGCAGGTCACTATC | [54] |
| | DXS8378 R | **ATT**GCGACAAGAGCGAAACTCCA | [54] |
| | DXS8378 Rx | **ATT**GCGACAAGACGCAAACTCCA | This study |
| DXS7132 | DXS7132 F | VIC-GAGCCCATTTT[cat]AATAAATCC | [272]/this study[a] |
| | DXS7132 F2 | VIC-AATAGTGTGAGCCCATTTTCA | This study |
| | DXS7132 R | **GTTTCTT**GCCAAACTCTATTAGTCAAC | [272] |
| | DXS7132 R2 | **ATT**CAAAGAAATGAGAACAAAAATCAGG | This study |
| DXS6803 | DXS6803 F | NED-GAAATGTGCTTTGACAGGAA | [273] |
| | DXS6803 R | **G**CAAAAAGGGACATATGCTACTT | [273] |
| | DXS6803 Rc | **G**CAAAAAGGAACATATGCTACTT | This study |
| HPRTB | HPRTB F | NED-TCTCTATTTCCATCTCTGTCTCC | [157] |
| | HPRTB R | GTCACCCCTGTCTATGGTCTCG | [157] |

Table continues on next page.

| Marker | Primer name | Primer sequence (5'-3') | Reference |
|---|---|---|---|
| GATA31E08 | GATA31E08 F | PET-CAGAGCTGGTGATGATAGATGA | [54] |
| | GATA31E08 Fv3 | NED-CAGAGCTGGTGATGATAGATGA | [54] |
| | GATA31E08 R | **ATT**GCTCACTTTTATGTGTGTATGTATCTCC | [54] |
| | GATA31E08 Rc | **ATT**CTCACTTTTATGTGTGTATGTATCTCC | [54] |
| | GATA31E08 Rg | **G**CTCACTTTTATGTGTGTATGTATCTCC | [54] |
| DXS9902 | DXS9902 F1 | VIC-TGGAGTCTCTGGGTGAAGAG | [274] |
| | DXS9902 F2 | PET-TGGAGTCTCTGGGTGAAGAG | [274] |
| | DXS9902 R | **ATT**CAGGAGTATGGGATCACCAG | [274] |
| DXS6795 | DXS6795 F | 6FAM-TGACATGGCTTTCTTTACAATTAC | This study |
| | DXS6795 R | **G**CCATGTTACATAAACAAGGAGTTATG | This study |
| DXS10147 | DXS10147 F | NED-CTGGGCGACAGAGTGAGATT | This study |
| | DXS10147 F2 | NED-AGGAGGTGAAGGTTGTGGTG | This study |
| | DXS10147 F2v3 | 6FAM-AGGAGGTGAAGGTTGTGGTG | This study |
| | DXS10147 R | **ATT**CTAATGGCCTGGGACTCTTC | This study |
| | DXS10147 R3 | **ATT**TGGGACTCTTCCCTTAAATGC | This study |
| DXS10101 | DXS10101 F | 6FAM-TGTGTTTCAATCTTTCCATAATAAAA | This study |
| | DXS10101 R | **G**AGGCTTATCCTGATACCGTATTT | This study |
| DXS10103 | DXS10103 F | 6FAM-CCTTCATAATCACATATCACATGAGC | This study |
| | DXS10103 R | **G**AAACAGAACCAGGGGAATGAA | This study |
| DXS9895 | DXS9895 F | NED-TTGGGTGGGGACACAGAG | This study |
| | DXS9895 F.big | NED-CACGTGGGAATTATGGGATG | This study |
| | DXS9895 R | **ATT**CCTGGCTCAAGGAAATTCAA | This study |
| | DXS9895 R2 | **ATT**GAATGGCTCTGCTCAAGGAA | This study |
| SRY | SRY F | NED-TGGCGATTAAGTCAAATTCGC | [275] |
| | SRY F2 | NED-AAAAATTGGCGATTAAGTCAAA | This study |
| | SRY F3 | NED-AAATTGGCGATTAAGTCAAATTC | This study |
| | SRY R | **ATT**AGCAGGGCAAGTAGTCAACG | This study |
| | SRY R2 | **G**TTGACTACTTGCCCTGCTGA | This study |
| | SRY R2.att | **ATT**TTGACTACTTGCCCTGCTGA | This study |

The first version of the multiplexes was designed as follows: mini X-plex 1 consisted of DXS7424, DXS6789, DXS7130, DXS9902, DXS7423, GATA165B12, and DXS101 while mini X-plex 2 consisted of DXS7133, GATA172D05, DXS8378, DXS7132, DXS6803, HPRTB, GATA31E08, and DXS9902. The primer sequences chosen for both versions as well as the organisation within the multiplexes are shown in Table 3.4. Note that marker DXS9902 was included in both multiplexes for concordance and quality control between the two multiplexes. Additionally, confirmation of sex in the form of a marker located within the sex-determining region of the Y chromosome (SRY) [275,276] was also included in both multiplexes. SRY was chosen over the more widely-used amelogenin locus in order to avoid known cases of amelogenin allele dropout (see [277-282] for example).

**Table 3.4. Mini-X STR amplification primers & conditions.** Bases in bold are tails added to the primer sequence to promote complete adenylation. Final primer sequences for markers DXS7424, DXS6795, and DXS10147 were designed as part of this study. Final primer concentrations and amplicon size ranges are given only for the final set of primer sequences. Amplicon size ranges include tails. $MP_O$: original multiplex; $MP_F$: final multiplex; [primer]: primer concentration. *Basic primer sequence obtained from publication but underlined base modified in this study to match GenBank sequence.

| Marker name | Original primer sequence (5'-3') | Original reference | $MP_O$ | Final primer sequence (5'-3') | $MP_F$ | Final [primer] (uM) | Amplicon size range (bp) |
|---|---|---|---|---|---|---|---|
| DXS6789 | 6FAM-CCTCGTGATCATGTAAGTTGG | [54] | 1 | same as original | 1 | 0.8 | 124-168 |
| | **ATT**CAGAACCAATAGGAGATAGATGGT | [54] | | same as original | | 0.8 | |
| DXS7130 | VIC-AATATAGAGGAAGGGGAAATCATTA | This study | 1 | same as original | 1 | 1.6 | 93-136 |
| | **ATT**CAAAGAAATGAGAACAAAAATCAGG | This study | | same as original | | 1.6 | |
| GATA31E08 | PET-CAGAGCTGGTGATGATAGATGA | [54] | 2 | NED-CAGAGCTGGTGATGATAGATGA | 1 | 1.2 | 95-131 |
| | **ATT**CTCACTTTTATGTGTGTATGTATCTCC | [54] | | same as original | | 1.2 | |
| DXS7424 | 6FAM-AA<u>C</u>ACAGGAAGACCCCATC | [54]* | 1 | NED-GGACTGCTTGAGTCCAGGAA | 1 | 1.2 | 146-185 |
| | **ATT**GCTAAGAAGAATCCCGCACA | [54] | | **G**GGAACACGCACATTTGAGAA | | 1.2 | |
| GATA165B12 | PET-TCATCAATCATCTATCCGTATATCA | [54] | 1 | same as original | 1 | 3.2 | 92-112 |
| | **ATT**AAGTTGACTGTGATTCCTGGTTT | [54] | | same as original | | 3.2 | |
| DXS101 | PET-TCTCCCTTCAAAAACAAAGATAA | [54] | 1 | **ATT**TCTCCCTTCAAAAACAAAGATAA | 1 | 1.2 | 126-177 |
| | **ATT**TGCATATTCTGCGCATGT | [84] | | PET-TGCATATTCTGCGCATGT | | 1.2 | |
| DXS7133 | 6FAM-AGCTTCCTTAGATGGCATTCA | [84] | 2 | Not included in final multiplexes | n/a | n/a | n/a |
| | **ATT**TTTTTAACGGTGTTCATGCTT | [84] | | Not included in final multiplexes | | n/a | |
| DXS9902 | VIC-TGGAGTCTCTGGGTGAAGAG | [274] | 1 | same as original | 1 | 0.4 | 167-191 |
| | **ATT**CAGGAGTATGGGATCACCAG | [274] | | same as original | | 0.4 | |
| DXS6795 | *Not included in original multiplexes* | *n/a* | *n/a* | 6FAM-TGACATGGCTTTCTTTACAATTAC | 2 | 0.6 | 90-111 |
| | *Not included in original multiplexes* | *n/a* | | **G**CCATGTTACATAAACAAGGAGTTATG | | 0.6 | |

Table continues on next page.

| Marker name | Original primer sequence (5'-3') | Original reference | $MP_O$ | Final primer sequence (5'-3') | $MP_F$ | Final [primer] (uM) | Amplicon size range (bp) |
|---|---|---|---|---|---|---|---|
| GATA172D05 | 6FAM-TAGTGGTGATGGTTGCACAG | [272] | 2 | 6FAM-**GTTTCTTT**AGTGGTGATGGTTGCACAG | 2 | 2.8 | 122-150 |
| | **GTTTCTT**ATAATTGAAAGCCCGGATTC | [272] | | same as original | | 2.8 | |
| DXS10147 | *Not included in original multiplexes* | *n/a* | *n/a* | 6FAM-AGGAGGTGAAGGTTGTGGTG | 2 | 0.2 | 165-185 |
| | *Not included in original multiplexes* | *n/a* | | **ATT**TGGGACTCTTCCCTTAAATGC | | 0.2 | |
| DXS8378 | VIC-GCTCCTGGCAGGTCACTATC | [84] | 2 | same as original | 2 | 1.6 | 94-118 |
| | **ATT**GCGACAAGAGCGAAACTCCA | [84] | | same as original | | 1.6 | |
| DXS7132 | VIC-AATAGTGTGAGCCCATTTTCA | This study | 2 | same as original | 2 | 0.4 | 149-177 |
| | **GTTTCTT**GCCAAACTCTATTAGTCAAC | [272] | | same as original | | 0.4 | |
| DXS6803 | NED-GAAATGTGCTTTGACAGGAA | [273] | 2 | same as original | 2 | 0.4 | 102-130 |
| | **G**CAAAAAGGAACATATGCTACTT | [273]* | | same as original | | 0.4 | |
| HPRTB | NED-TCTCTATTTCCATCTCTGTCTCC | [157] | 2 | same as original | 2 | 0.4 | 148-176 |
| | **G**TCACCCCTGTCTATGGTCTCG | [157] | | same as original | | 0.4 | |
| DXS7423 | NED-AGATTTCCTCCCCATCCATC | [84] | 1 | PET-AGATTTCCTCCCCATCCATC | 2 | 1.2 | 85-125 |
| | **ATT**TTGTCACACAAATAAATGAATGAGT | [84] | | same as original | | 1.2 | |
| DXS9902 | PET-TGGAGTCTCTGGGTGAAGAG | [274] | 2 | same as original | 2 | 1.2 | 167-191 |
| | **ATT**CAGGAGTATGGGATCACCAG | [274] | | same as original | | 1.2 | |
| SRY | NED-AAAAATTGGCGATTAAGTCAAA | This study | Both | same as original | Both | 0.4 | 86 |
| | **G**TTGACTACTTGCCCTGCTGA | This study | | same as original | | 0.4 | |

After the continued testing and allele sequencing, complications with the current set of chosen markers became evident, suggesting that reorganisation might provide more optimal multiplexes. To begin, two of the three defined selection criteria were not met by the initial version of the multiplexes. First, not all of the chosen markers had heterozygosity values in U.S. populations that were ideal for forensic purposes (as high as possible). Second, heavy reliance on markers from the second linkage group reduced the overall effectiveness of the multiplexes by limiting the discriminatory power of the other three linkage groups. The best example of these two violations was marker DXS7133, a part of the over-represented linkage group 2. Though a relatively high heterozygosity value (greater than 0.65) had been published for this marker [91,115,200], initial testing revealed some of the lowest observed heterozygosity values of these 14 markers in U.S. populations (0.5400–0.6494). Additionally, allele sequencing results of control DNAs revealed a primer binding site mutation (G→A) located 7 base pairs from the 3' end of the forward STR amplification primer that resulted in reduced peak height of the affected allele (Figure 3.1). Though the frequency of this mutation in the general population is unknown, its discovery was sufficient to confirm the decision to remove this marker from the final set of chosen informative markers.

**Figure 3.1. DXS7133 primer binding site mutation.** **A.** Singleplex DXS7133 amplification results showed dramatically reduced peak height for male control DNA 1 compared to other controls. **B.** Sequencing results revealed that a single nucleotide polymorphism located seven bases from the 3' end of the primer binding site is the cause of this reduction in peak height.

**A. Singleplex DXS7133 amplification of three control DNAs.**

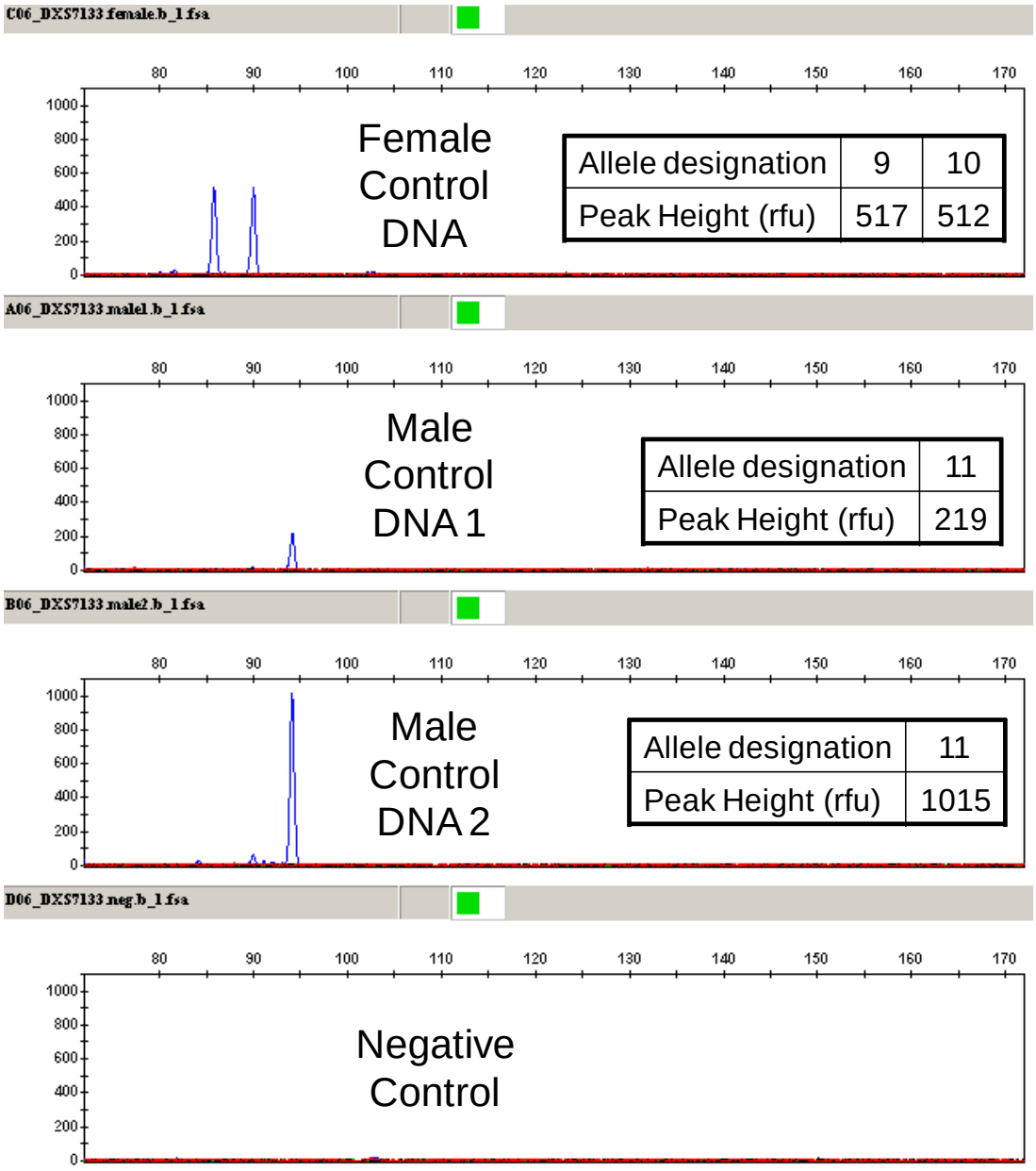
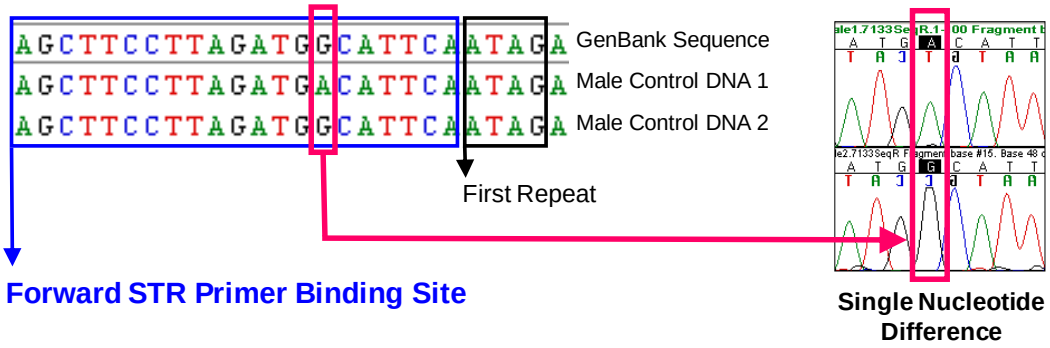



Figure continues on next page.

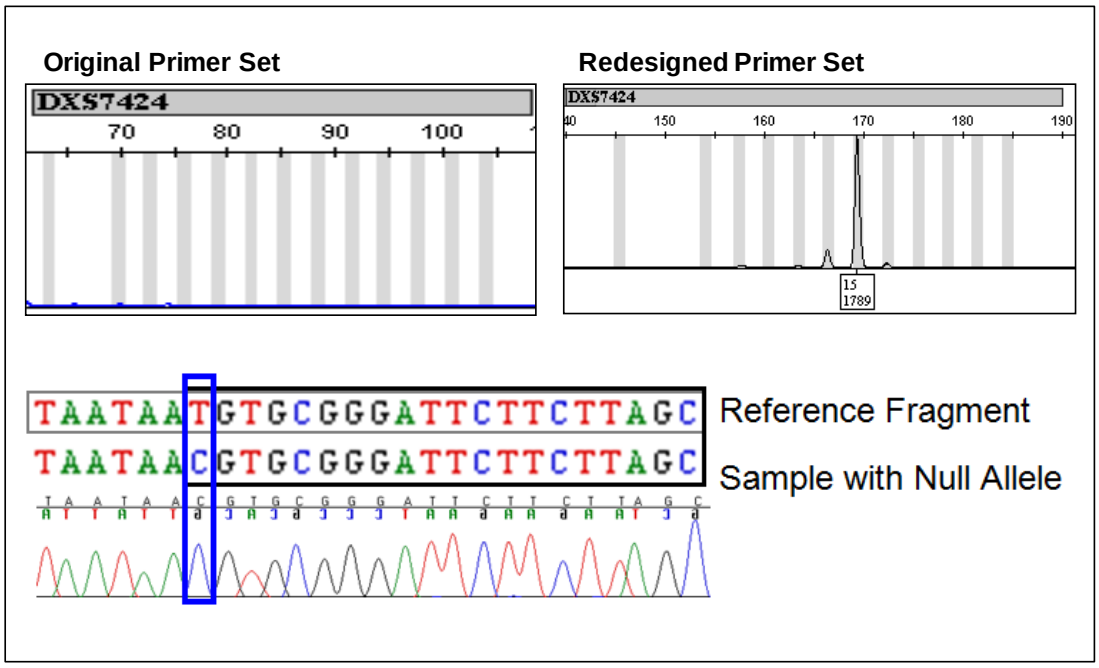
**B. Sequencing results for marker DXS7133.**




Other revisions to the initial multiplexes included (1) the addition of two markers meeting the defined selection criteria; (2) the redesign of primers affected by primer binding site mutations; and (3) the relocation of two markers to alternate dye channels. First, DXS10147 and DXS6795 were added to the multiplexes primarily for their contribution to the diversity of proposed linkage groups included; neither was part of linkage group 2. DXS10147 belongs to linkage group 4 while DXS6795 belongs to linkage group 1, increasing the number of markers representing each of the respective linkage groups from 2 to 3. Second, the STR amplification primers for marker DXS7424 were redesigned to avoid three individual primer binding site mutations that occurred frequently during initial experiments (Figure 3.2). The label on the forward primer for this marker was changed from 6FAM™ to NED™ in order to accommodate the slightly larger amplicon size resulting from the redesigned primer set.

**Figure 3.2. DXS7424 primer binding site mutations.** When amplified using the original published STR primer pair (left), affected alleles are either (A.) missing (null) or (B.) much lower than the rest of the profile (partially null). Sequencing results revealed variation (blue box) under the reverse primer binding site (black box). When re-amplified with a primer pair designed to avoid such sequence variation, detection of affected alleles is restored (right). C. Schematic of STR amplification primer placement for marker DXS7424. Strand orientation within figure is 5' → 3'.

**A.**




**B.**

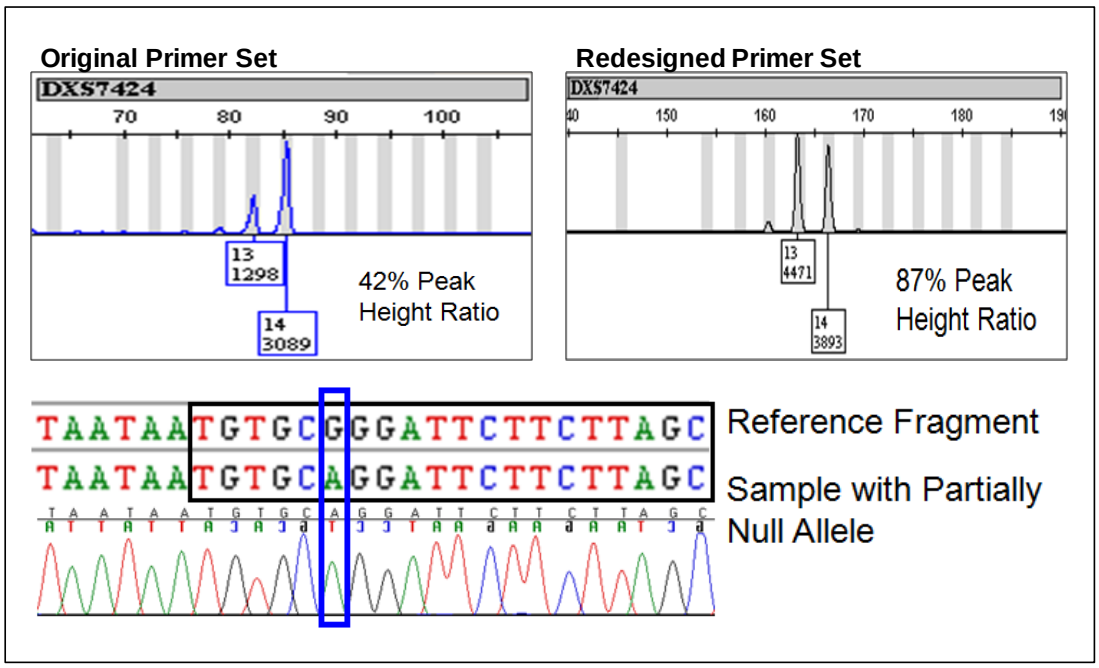
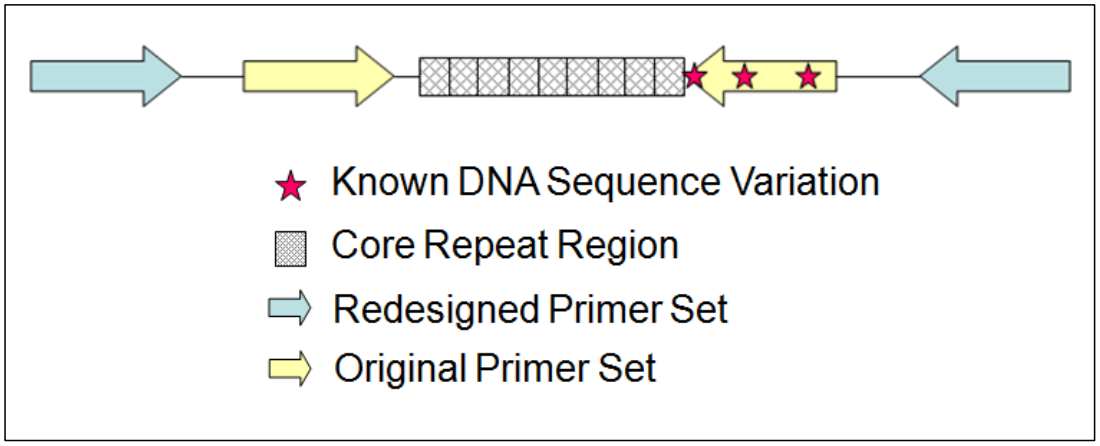



Figure continues on next page.

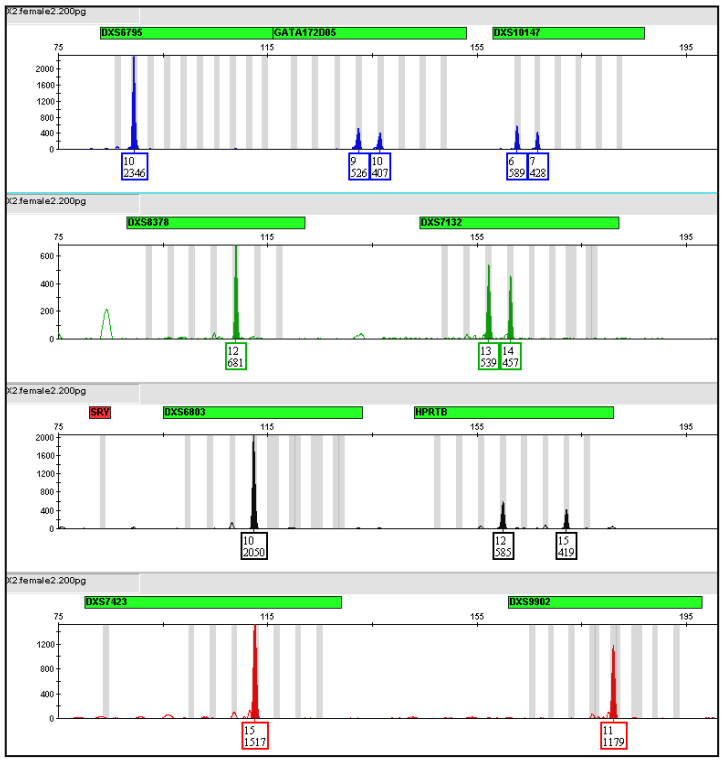
**C.**




Additionally, the forward primer for DXS101 was redesigned while the PET® dye label was moved to the reverse primer; both changes resulted in a more robust amplification of this locus. Third, dye labels and multiplex locations were switched for markers DXS7423 and GATA31E08, as detailed in Table 3.4. No changes were made to the primer sequences. This move was completed in order to situate the four markers also present in the commercially available kit (DXS8378, DXS7132, HPRTB, and DXS7423) into a single multiplex, allowing concordance to be performed with ease between laboratories using the multiplexes proposed here and those using the commercially available kit. The final multiplexes and primers are listed in Table 3.4. Example electropherograms for the two final multiplexes are shown in Figure 3.3.

**Figure 3.3. Example electropherograms for two X STR multiplexes.** (A) Multiplex 1 and (B) multiplex 2 both amplified using 200 pg input DNA from a female control sample in the sensitivity study demonstrating a full profile. (C) Multiplex 1 and (D) multiplex 2 both resulting from an old bone sample in the degraded samples study.

**A.**




**B.**

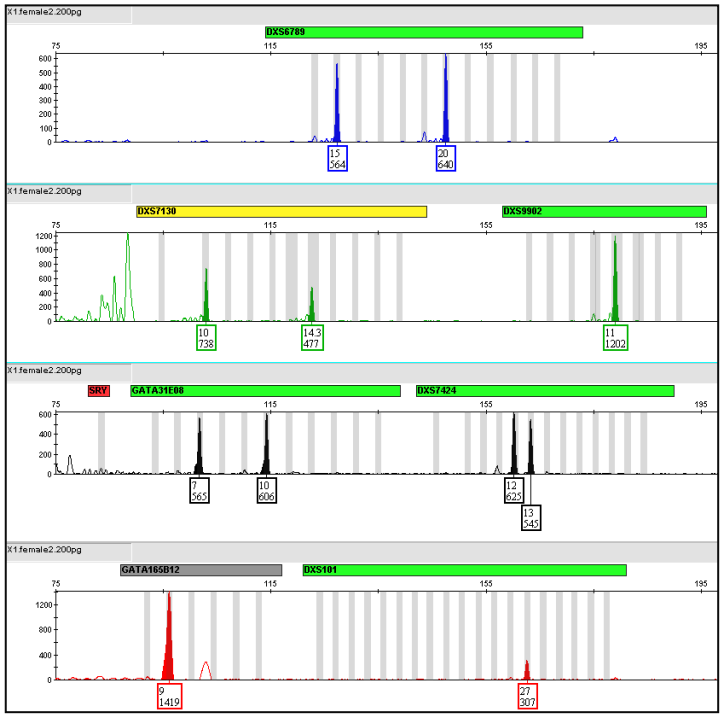
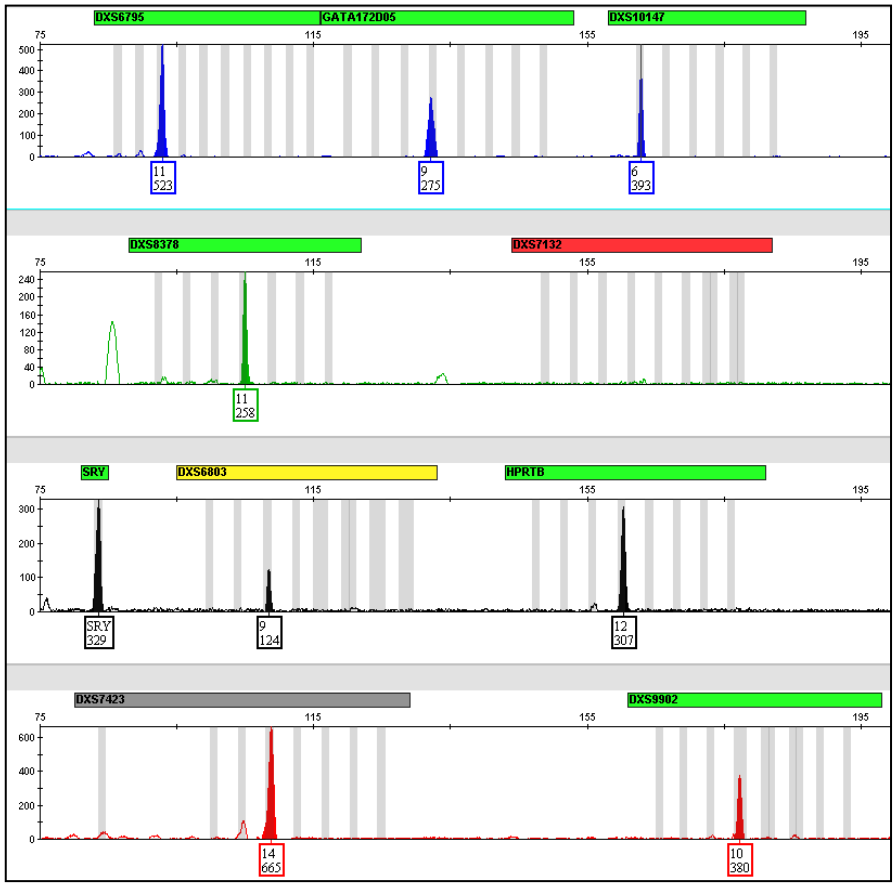
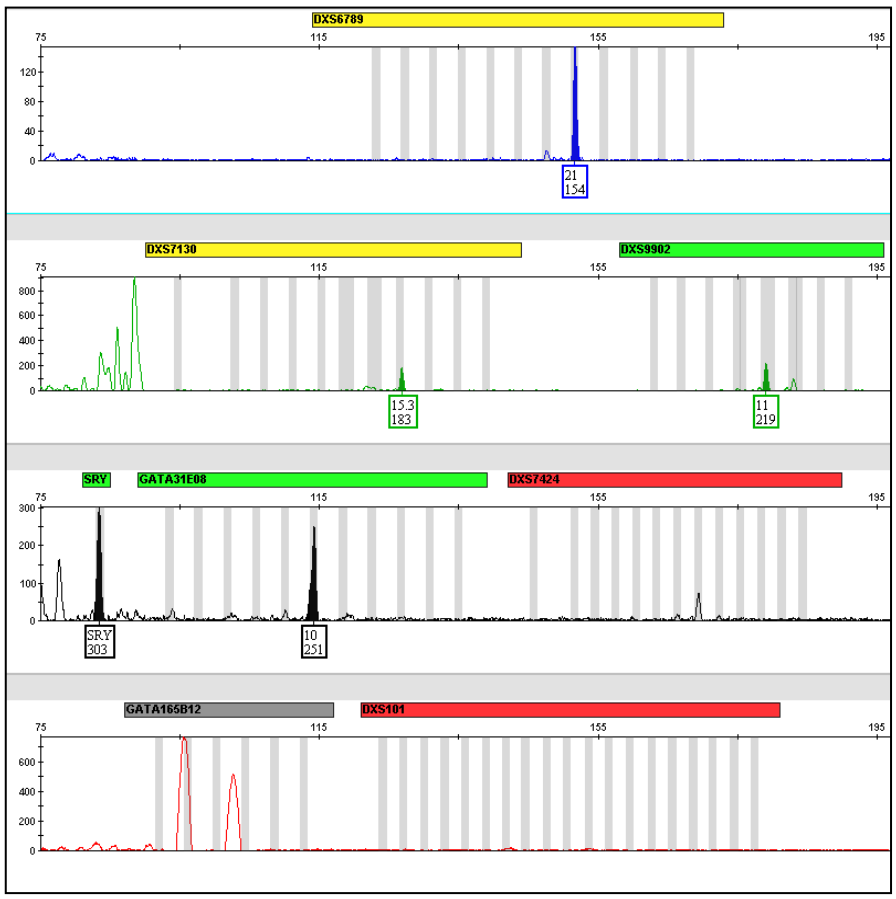



Figure continues on next page.

**C.**


DXS6795
GATA172D05
DXS10147
11
523
9
275
6
393
DXS8378
DXS7132
11
258
SRY
DXS6803
HPRTB
SRY
329
9
124
12
307
DXS7423
DXS9902
14
665
10
380

**D.**


DXS6789
21
154
DXS7130
DXS9902
15.3
183
11
219
SRY
GATA31E08
DXS7424
SRY
303
10
251
GATA165B12
DXS101

*3.2. Nomenclature*

Sample genotyping followed published allele nomenclature according to [200,207,216,236,247,258,272-274,283,284] and guidelines set forth by the DNA Commission of the ISFG [242]. Table 3.5 lists the repeat motif and profile of control DNA 9947A for all markers; other control DNAs matched the profiles published in [258] and on the Forensic ChrX Research website version 2.0 except for markers GATA31E08 and DXS6803. All control DNAs were sequenced to confirm their repeat structure. Sequencing results for marker GATA31E08 revealed additional variation resulting from AGGG repeats before the reported AGAT repeat unit. Most commonly, two AGGG repeats are present before the AGAT units, and variation results from additional AGAT repeats (also noted in [284]). Including the initial AGGG repeats increases the allele designation for this marker by two repeat units compared to that using solely the AGAT repeat unit. Additional details on the allele sequencing results can be found in Chapter 7. Further, though there is no conflict with the published repeat unit (TCTA) for marker DXS6803, the allele designation used in this study differs from the published profiles [258] of control DNAs 9947A, 9948, and K562 by +1 repeat unit. Additionally, the observed profile of 9947A was a heterozygote 11.3, 12 (Table 3.5), rather than a homozygote 11[258,273].

**Table 3.5. Characteristics of the 16 X STR markers examined in this study.** $N_8$: TCTGTCCT; x and y: variables representing the number of repeats; Ref.: nomenclature reference. *According to [157,209,271].

| Marker name | Observed allele range | Repeat motif | Ref. | Linkage group* | DNA profile 9947A |
|---|---|---|---|---|---|
| DXS6789 | 14-25 | $(TATC)_{(0-1)}$-$(TATG)_x$-$(TATC)_y$ | [207] | 2 | 21, 22 |
| DXS7130 | 9-14, 16, 13.3-18.3 | $(TATC)_5$-$ATC_{(0-1)}$-$(TATC)_x$ | [273] | 2 | 15.3, 15.3 |
| GATA31E08 | 7-16 | $(AGGG)_x$-$(AGAT)_y$ | [284] | 4 | 13, 13 |
| DXS7424 | 9-20 | $(TAA)_x$ | [200] | 2 | 14, 16 |
| GATA165B12 | 8-13 | $(AGAT)_x$ | [274] | 2 | 9, 11 |
| DXS101 | 14-31, 33 | $(CTT)_x$-$(ATT)_y$ | [283] | 2 | 24, 26 |
| DXS6795 | 6, 8-17 | ATT-$ATC_{(0-1)}$-$(ATT)_x$ | This study | 1 | 12, 13 |
| GATA172D05 | 6-13 | $(TAGA)_x$ | [272] | 2 | 10, 10 |
| DXS10147 | 5-11 | $(AAAC)_x$ | [236] | 4 | 8, 8 |
| DXS8378 | 8-15 | $(CTAT)_x$ | [200] | 1 | 10, 11 |
| DXS7132 | 10-18, 16.3 | $(TCTA)_x$-$(TCA)_{(0-1)}$-$(TCTA)_2$ | [272] | 2 | 12, 12 |
| DXS6803 | 7-14, 16, 10.3-14.3 | $(TCTA)_x$-$(TCA)_{(0-1)}$-TCTA | [273] | 2 | 11.3, 12 |
| HPRTB | 7-16 | $(ATCT)_x$ | [200,247] | 3 | 14, 14 |
| DXS7423 | 8, 12-17 | $(TCCA)_3$-$(N_8)_{(0-1)}$-$(TCCA)_x$ | [216,272] | 4 | 14, 15 |
| DXS9902 | 7-14, 10.1-12.1 | $(GATA)_x$ | [200] | 1 | 11, 11 |
| DXS7133 | 6-14 | $(ATAG)_x$ | [200] | 2 | 9, 10 |

Of note, GATA172D05 allele calls were determined based upon the TAGA repeat as suggested by Edelmann *et al*. [272], rather than the GATA repeat used in earlier studies [200]. This distinction results in one additional repeat unit, for a corrected allelic range of 6–13 repeats (rather than 5–12). The repeat motif for marker HPRTB has also been published with conflicting nomenclature. An overview of the discrepancies and a recommendation to use the AGAT repeat motif according to Edwards *et al.* [285] can be found in [247]. In the present study, the ATCT coding strand equivalent of this recommended repeat unit was used and is in agreement with Szibor *et al*. [258].

Though the ISFG guidelines suggest that the first 5' nucleotides that can define a repeat motif for an STR marker should be used, there were several situations where this recommendation was not followed. Instead, allele numbering was based upon published and widely-used repeat motifs in favour of maintaining consistency with an established historical nomenclature and avoiding unnecessary confusion, which is itself in accordance with the ISFG guidelines [242]. For example, at locus DXS9902, the first 5' nucleotides that define a repeat motif are TAGA [286]; however, the GATA repeat unit nomenclature first published in [200] was used instead, which results in a one repeat unit difference. Similarly, although the repeat motif AAT may be the most 5' motif for DXS7424 [287], the published and widely-used TAA repeat unit [200] (which also results in a one repeat unit difference) was employed for allele designation here.

As evidenced by these 16 markers, further study is needed in order to reach a consensus regarding both STR nomenclature and allele designation in the community of X chromosome researchers. In the future, it might be useful for publications to include the nomenclature used for assigning allele number in addition to the control DNA profiles already suggested by Szibor *et al*. [258] in order to facilitate comparisons between different groups.

*3.3. Sensitivity and degraded sample testing*

In this study, full STR profiles were reliably obtained with as little as 200 pg of input DNA (Figure 3.3A), with a loss of only 1–2 markers at 100 pg. Because there are multiple markers from each linkage group present in the multiplexes (except for

linkage group 3), the information obtained even with a loss of 1–2 markers per multiplex will still be adequate in most situations. Below 100 pg, typing was less reliable when using the standard protocol described here, and a low copy number approach employing some combination of replicate analyses, increased cycle number, or increased polymerase could be considered. There was no difference observed between the two multiplexes, or between male and female profiles in this study.

Typing attempts on bone samples that were decades old revealed partial to full profiles with both multiplexes (Figure 3.3B). With this particular set of samples, multiplex 2 performed better, resulting in a full profile for two of the five bone samples tested, whereas multiplex 1 did not produce any full profiles. Further testing would be necessary in order to assess the true utility of these multiplexes with degraded DNA.

# Chapter 4. Population Databases

The forensic utility of an STR profile rests in the ability to determine how rare that particular profile is in the population. The generation of adequate databases composed of unrelated individuals from the relevant population group is therefore a critical component in the validation of any new STR marker system. Though X STR databases have been established for a number of populations (see Table 1.1), the work described in this chapter aimed to fill in some of the gaps. Additionally, the generation of thousands of individual profiles with the two novel multiplexes provided the opportunity to observe and characterize important aspects of their performance, including the occurrence of null alleles and the accuracy of allele calls through comparisons to similar published populations.

## *4.1. United States populations*

Three discrete sample sets were obtained via various collaborations, and are detailed in Table 4.1. Both male and female samples were included with a minimum of 140 individuals per population (with the exception of the U.S. Asians part of sample set B, which were not considered a distinct population group). Sample set C was composed of unrelated individuals from the mutation rate study (see Chapter 5).

**Table 4.1. United States population databases by contributor.** Three sample sets representing the four major U.S. populations were typed for 15 X STR markers. M: males; F: females; I: individuals; A: alleles.

| Sample Set | Contributor | Population | $N_M$ | $N_F$ | $N_I$ | $N_A$ |
|---|---|---|---|---|---|---|
| A | Orange County Sheriff-Coroner's Department Crime Laboratory | African Americans | 175 | 174 | 349 | 523 |
| | | U.S. Asians | 201 | 300 | 501 | 801 |
| | | U.S. Caucasians | 122 | 146 | 268 | 414 |
| | | U.S. Hispanics | 123 | 122 | 245 | 367 |
| B | National Institute of Standards and Technology | African Americans | 259 | 0 | 259 | 260 |
| | | U.S. Asians | 3 | 0 | 3 | 3 |
| | | U.S. Caucasians | 260 | 38 | 298 | 336 |
| | | U.S. Hispanics | 138 | 0 | 138 | 138 |
| C | George Washington University | African Americans | 108 | 206 | 314 | 520 |
| | | U.S. Caucasians | 165 | 269 | 434 | 703 |
| | | U.S. Hispanics | 150 | 248 | 398 | 646 |
| **Combined** | **Sample Sets A, B, & C** | **African Americans** | **542** | **380** | **923** | **1303** |
| | | **U.S. Asians** | **204** | **300** | **504** | **804** |
| | | **U.S. Caucasians** | **547** | **453** | **1000** | **1453** |
| | | **U.S. Hispanics** | **411** | **370** | **781** | **1151** |

Similar analyses were completed for each of the three sample sets, and sample sets were compared both to each other as well as to other published populations for quality control purposes and prior to combining any of the individual U.S. populations.

*4.1.1. Sample set A*

Sample set A was processed as part of the original development and characterisation of the two X STR mini-plexes. Please also see Appendix C for the following publication:

**C1.** Toni M. Diegoli, Michael D. Coble, **Development and characterization of two mini-X chromosomal short tandem repeat multiplexes**, Forensic Sci. Int. Genet. 5 (2011) 415-421.

The distribution of allele frequencies in the male and female populations was examined using the chi-square test for independence (Table 4.2). The resultant p values showed no significant differences ($p < 0.05$) and frequency data was pooled at each marker. Allele frequencies obtained for the 15 X chromosomal STRs examined in four U.S. population groups are shown in Table 4.3. In total, 158 alleles were observed across 15 markers, with 6 to 19 alleles at each marker.

**Table 4.2. P values from chi-square test comparing distribution of male and female allele frequencies from sample set A by marker.**

| Marker | P value |
|---|---|
| DXS6795 | 0.2549 |
| DXS9902 | 0.6957 |
| DXS8378 | 0.9732 |
| GATA172D05 | 0.6950 |
| DXS7132 | 0.5678 |
| DXS6803 | 0.1655 |
| DXS6789 | 0.5345 |
| GATA165B12 | 0.3398 |
| DXS7130 | 0.1018 |
| DXS101 | 0.6837 |
| DXS7424 | 0.4061 |
| HPRTB | 0.3178 |
| DXS7423 | 0.3038 |
| DXS10147 | 0.8049 |
| GATA31E08 | 0.1702 |

**Table 4.3. Allele frequencies and summary statistics for 15 X chromosomal STR markers in 4 U.S. population groups from sample set A.** P values from the exact test indicating a deviation from Hardy-Weinberg equilibrium ($p < 0.05$) are in bold. AA: African American (N=349), AS: U.S. Asian (N=501), CN: U.S. Caucasian (N=268), Hisp: U.S. Hispanic (N=245), H(exp): expected heterozygosity, H(obs): observed heterozygosity, PIC: polymorphism information content, PDf: power of discrimination in females, PDm: power of discrimination in males, MECI: mean exclusion chance in trios involving daughter, MECII: mean exclusion chance in father/daughter duos, p (HWE): p value of the exact test for Hardy-Weinberg equilibrium.

| | AA | AS | CN | Hisp | | AA | AS | CN | Hisp |
|---|---|---|---|---|---|---|---|---|---|
| **DXS9902** | | | | | **DXS6789** | | | | |
| 7 | 0.0019 | 0.0037 | | | 14 | 0.0057 | 0.0050 | 0.0024 | 0.0054 |
| 8 | 0.0325 | | | 0.0027 | 15 | 0.2122 | 0.1561 | 0.0483 | 0.0599 |
| 9 | 0.0746 | 0.0175 | 0.0338 | 0.0272 | 16 | 0.1147 | 0.3633 | 0.0193 | 0.0627 |
| 10 | 0.2849 | 0.4220 | 0.3357 | 0.3733 | 17 | 0.0115 | 0.0312 | 0.0048 | 0.0054 |
| 10.1 | | | | 0.0054 | 18 | 0.0172 | | | 0.0027 |
| 11 | 0.3690 | 0.3346 | 0.3865 | 0.3515 | 19 | 0.0516 | 0.0250 | 0.0217 | 0.0354 |
| 11.1 | 0.0096 | | 0.0217 | 0.0327 | 20 | 0.2065 | 0.1935 | 0.3575 | 0.4005 |
| 12 | 0.2237 | 0.2122 | 0.2150 | 0.2016 | 21 | 0.2065 | 0.1386 | 0.3188 | 0.2561 |
| 12.1 | | | | 0.0054 | 22 | 0.1090 | 0.0712 | 0.1425 | 0.1199 |
| 13 | 0.0038 | 0.0087 | 0.0072 | | 23 | 0.0554 | 0.0137 | 0.0700 | 0.0409 |
| 14 | | 0.0012 | | | 24 | 0.0076 | 0.0025 | 0.0145 | 0.0082 |
| | | | | | 25 | 0.0019 | | | 0.0027 |
| H(exp) | 0.7259 | 0.6645 | 0.6900 | 0.6946 | H(exp) | 0.8384 | 0.7801 | 0.7419 | 0.7491 |
| H(obs) | 0.6954 | 0.6933 | 0.6438 | 0.7459 | H(obs) | 0.8736 | 0.8200 | 0.6849 | 0.7213 |
| PIC | 0.6784 | 0.5983 | 0.6311 | 0.6376 | PIC | 0.8183 | 0.7516 | 0.7024 | 0.7163 |
| PDf | 0.8774 | 0.8212 | 0.8450 | 0.8498 | PDf | 0.9538 | 0.9231 | 0.8939 | 0.9043 |
| PDm | 0.7259 | 0.6645 | 0.6900 | 0.6946 | PDm | 0.8384 | 0.7801 | 0.7419 | 0.7491 |
| MECI | 0.6784 | 0.5983 | 0.6311 | 0.6376 | MECI | 0.8183 | 0.7516 | 0.7024 | 0.7163 |
| MECII | 0.5368 | 0.4512 | 0.4856 | 0.4929 | MECII | 0.7071 | 0.6223 | 0.5653 | 0.5814 |
| p (HWE) | 0.5968 | 0.8160 | 0.2615 | 0.4521 | p (HWE) | 0.4370 | 0.3430 | 0.2227 | 0.9175 |
| | | | | | | | | | |
| **GATA165B12** | | | | | **DXS7423** | | | | |
| 8 | 0.0382 | 0.0037 | 0.0121 | 0.0136 | 8 | 0.0076 | | | |
| 9 | 0.1702 | 0.2547 | 0.3140 | 0.2452 | 12 | 0.0057 | | | |
| 10 | 0.3308 | 0.5643 | 0.3140 | 0.4496 | 13 | 0.0822 | 0.0050 | 0.0966 | 0.0300 |
| 11 | 0.3595 | 0.1511 | 0.3333 | 0.2561 | 14 | 0.4818 | 0.3833 | 0.3502 | 0.2997 |
| 12 | 0.0975 | 0.0262 | 0.0266 | 0.0354 | 15 | 0.3270 | 0.5581 | 0.3816 | 0.4796 |
| 13 | 0.0038 | | | | 16 | 0.0765 | 0.0524 | 0.1256 | 0.0954 |
| | | | | | 17 | 0.0191 | 0.0012 | 0.0459 | 0.0954 |
| H(exp) | 0.7214 | 0.5932 | 0.6909 | 0.6707 | H(exp) | 0.6479 | 0.5388 | 0.7045 | 0.6612 |
| H(obs) | 0.7184 | 0.5967 | 0.6712 | 0.6148 | H(obs) | 0.6897 | 0.5233 | 0.6712 | 0.6721 |
| PIC | 0.6734 | 0.5338 | 0.6271 | 0.6111 | PIC | 0.5893 | 0.4448 | 0.6538 | 0.6074 |
| PDf | 0.8744 | 0.7751 | 0.8407 | 0.8319 | PDf | 0.8174 | 0.6933 | 0.8620 | 0.8315 |
| PDm | 0.7214 | 0.5932 | 0.6909 | 0.6707 | PDm | 0.6479 | 0.5388 | 0.7045 | 0.6612 |
| MECI | 0.6734 | 0.5338 | 0.6271 | 0.6111 | MECI | 0.5893 | 0.4448 | 0.6538 | 0.6074 |
| MECII | 0.5313 | 0.3860 | 0.4807 | 0.4639 | MECII | 0.4436 | 0.3080 | 0.5106 | 0.4612 |
| p (HWE) | 0.7492 | 0.7070 | 0.9359 | 0.1552 | p (HWE) | 0.7291 | 0.4901 | 0.3688 | 0.7558 |

Table continues on next page.

| | AA | AS | CN | Hisp | | AA | AS | CN | Hisp |
|---|---|---|---|---|---|---|---|---|---|
| **HPRTB** | | | | | **DXS7132** | | | | |
| 7 | | | 0.0024 | | 10 | | 0.0012 | | |
| 8 | 0.0019 | | | | 11 | 0.0115 | 0.0037 | 0.0145 | 0.0054 |
| 9 | 0.0382 | | 0.0024 | 0.0027 | 12 | 0.1013 | 0.0799 | 0.0966 | 0.0817 |
| 10 | 0.0134 | | 0.0024 | 0.0109 | 13 | 0.2581 | 0.1723 | 0.3092 | 0.2752 |
| 11 | 0.0975 | 0.0886 | 0.1159 | 0.0736 | 14 | 0.3709 | 0.3695 | 0.3599 | 0.3215 |
| 12 | 0.2964 | 0.2584 | 0.3406 | 0.2561 | 15 | 0.2180 | 0.2734 | 0.1522 | 0.2507 |
| 13 | 0.2887 | 0.4095 | 0.3188 | 0.3951 | 16 | 0.0306 | 0.0824 | 0.0628 | 0.0545 |
| 14 | 0.1740 | 0.1860 | 0.1618 | 0.1989 | 16.3 | 0.0019 | | | |
| 15 | 0.0727 | 0.0524 | 0.0459 | 0.0490 | 17 | 0.0038 | 0.0175 | 0.0024 | 0.0109 |
| 16 | 0.0172 | 0.0050 | 0.0097 | 0.0136 | 18 | 0.0038 | | 0.0024 | |
| H(exp) | 0.7818 | 0.7203 | 0.7405 | 0.7306 | H(exp) | 0.7369 | 0.7455 | 0.7382 | 0.7483 |
| H(obs) | 0.7931 | 0.7267 | 0.8151 | 0.7869 | H(obs) | 0.7701 | 0.7533 | 0.7945 | 0.7295 |
| PIC | 0.7499 | 0.6759 | 0.6979 | 0.6883 | PIC | 0.6935 | 0.7060 | 0.6962 | 0.7053 |
| PDf | 0.9205 | 0.8774 | 0.8900 | 0.8851 | PDf | 0.8873 | 0.8957 | 0.8895 | 0.8937 |
| PDm | 0.7818 | 0.7203 | 0.7405 | 0.7306 | PDm | 0.7369 | 0.7455 | 0.7382 | 0.7483 |
| MECI | 0.7499 | 0.6759 | 0.6979 | 0.6883 | MECI | 0.6935 | 0.7060 | 0.6962 | 0.7053 |
| MECII | 0.6203 | 0.5338 | 0.5589 | 0.5481 | MECII | 0.5535 | 0.5682 | 0.5573 | 0.5671 |
| p (HWE) | 0.8825 | 0.3569 | 0.8683 | 0.9161 | p (HWE) | 0.3662 | 0.9305 | 0.7262 | 0.2625 |
| **DXS8378** | | | | | **GATA172D05** | | | | |
| 8 | 0.0096 | 0.0012 | 0.0024 | 0.0027 | 6 | 0.1855 | 0.0549 | 0.1787 | 0.1117 |
| 9 | 0.0115 | 0.0212 | 0.0072 | 0.0082 | 7 | 0.0382 | 0.0087 | 0.0024 | 0.0082 |
| 10 | 0.2811 | 0.5630 | 0.3551 | 0.3815 | 8 | 0.1778 | 0.1860 | 0.1618 | 0.1335 |
| 11 | 0.3595 | 0.2672 | 0.3454 | 0.3161 | 9 | 0.2772 | 0.0936 | 0.0507 | 0.0708 |
| 12 | 0.3059 | 0.1311 | 0.2585 | 0.2698 | 10 | 0.1606 | 0.3833 | 0.2971 | 0.3134 |
| 13 | 0.0268 | 0.0112 | 0.0266 | 0.0191 | 11 | 0.1090 | 0.2272 | 0.2029 | 0.2507 |
| 14 | 0.0057 | 0.0037 | 0.0048 | 0.0027 | 12 | 0.0516 | 0.0462 | 0.0990 | 0.1090 |
| 15 | | 0.0012 | | | 13 | | | 0.0072 | 0.0027 |
| H(exp) | 0.6972 | 0.5939 | 0.6870 | 0.6814 | H(exp) | 0.8153 | 0.7529 | 0.8000 | 0.7917 |
| H(obs) | 0.6322 | 0.5700 | 0.7329 | 0.7213 | H(obs) | 0.8563 | 0.7400 | 0.7534 | 0.8033 |
| PIC | 0.6372 | 0.5348 | 0.6236 | 0.6163 | PIC | 0.7901 | 0.7173 | 0.7713 | 0.7625 |
| PDf | 0.8483 | 0.7760 | 0.8387 | 0.8334 | PDf | 0.9407 | 0.9034 | 0.9313 | 0.9274 |
| PDm | 0.6972 | 0.5939 | 0.6870 | 0.6814 | PDm | 0.8153 | 0.7529 | 0.8000 | 0.7917 |
| MECI | 0.6372 | 0.5348 | 0.6236 | 0.6163 | MECI | 0.7901 | 0.7173 | 0.7713 | 0.7625 |
| MECII | 0.4917 | 0.3875 | 0.4773 | 0.4695 | MECII | 0.6696 | 0.5813 | 0.6457 | 0.6353 |
| p (HWE) | 0.4949 | 0.5703 | 0.6491 | 0.1022 | p (HWE) | 0.5409 | 0.5898 | 0.4575 | 0.9691 |
| **DXS6795** | | | | | **GATA31E08** | | | | |
| 6 | 0.0019 | | | | 7 | 0.0210 | | | |
| 8 | 0.0019 | | | | 8 | 0.0287 | 0.0012 | 0.0024 | 0.0027 |
| 9 | 0.1166 | 0.0412 | 0.3019 | 0.1826 | 9 | 0.1396 | 0.0986 | 0.1691 | 0.1771 |
| 10 | 0.2677 | 0.1710 | 0.0169 | 0.1090 | 10 | 0.1472 | 0.0275 | 0.0193 | 0.0872 |
| 11 | 0.1950 | 0.3121 | 0.4710 | 0.2234 | 11 | 0.0612 | 0.1935 | 0.2222 | 0.1798 |
| 12 | 0.0880 | 0.0325 | 0.0290 | 0.1471 | 12 | 0.2486 | 0.2397 | 0.2053 | 0.3188 |
| 13 | 0.0975 | 0.4195 | 0.1787 | 0.2997 | 13 | 0.2620 | 0.3208 | 0.2609 | 0.1853 |
| 14 | 0.0516 | 0.0212 | | 0.0191 | 14 | 0.0822 | 0.1049 | 0.1111 | 0.0327 |
| 15 | 0.1702 | 0.0025 | 0.0024 | 0.0191 | 15 | 0.0076 | 0.0125 | 0.0097 | 0.0163 |
| 16 | 0.0076 | | | | 16 | 0.0019 | 0.0012 | | |
| 17 | 0.0019 | | | | | | | | |
| H(exp) | 0.8278 | 0.6942 | 0.6539 | 0.7927 | H(exp) | 0.8166 | 0.7805 | 0.7990 | 0.7914 |
| H(obs) | 0.8448 | 0.6567 | 0.5685 | 0.8197 | H(obs) | 0.8448 | 0.7633 | 0.8288 | 0.8607 |
| PIC | 0.8059 | 0.6419 | 0.5927 | 0.7620 | PIC | 0.7924 | 0.7479 | 0.7684 | 0.7615 |
| PDf | 0.9484 | 0.8542 | 0.8190 | 0.9263 | PDf | 0.9421 | 0.9192 | 0.9290 | 0.9266 |
| PDm | 0.8278 | 0.6942 | 0.6539 | 0.7927 | PDm | 0.8166 | 0.7805 | 0.7990 | 0.7914 |
| MECI | 0.8059 | 0.6419 | 0.5927 | 0.7620 | MECI | 0.7924 | 0.7479 | 0.7684 | 0.7615 |
| MECII | 0.6904 | 0.4977 | 0.4456 | 0.6340 | MECII | 0.6732 | 0.6173 | 0.6416 | 0.6336 |
| p (HWE) | 0.9080 | 0.1521 | 0.2011 | 0.5093 | p (HWE) | 0.7598 | 0.6962 | 0.4349 | 0.6854 |

Table continues on next page.

| | AA | AS | CN | Hisp | | AA | AS | CN | Hisp |
|---|---|---|---|---|---|---|---|---|---|
| **DXS6803** | | | | | **DXS7130** | | | | |
| 7 | 0.0019 | | | 0.0054 | 9 | 0.0076 | | 0.0024 | |
| 8 | 0.0096 | | | | 10 | 0.0229 | 0.0075 | | 0.0027 |
| 9 | 0.0249 | | 0.0024 | | 11 | 0.0765 | 0.2122 | 0.0362 | 0.0572 |
| 10 | 0.1434 | 0.0112 | 0.0435 | 0.0245 | 12 | 0.1931 | 0.1810 | 0.1014 | 0.2125 |
| 10.3 | | | | 0.0027 | 13 | 0.1491 | 0.0674 | 0.0580 | 0.0790 |
| 11 | 0.3671 | 0.1948 | 0.2633 | 0.2561 | 13.3 | 0.0229 | 0.0050 | 0.0435 | 0.0191 |
| 11.3 | 0.0096 | 0.0687 | 0.0097 | 0.0572 | 14 | 0.0440 | 0.0050 | 0.0169 | 0.0163 |
| 12 | 0.2333 | 0.1423 | 0.2633 | 0.3488 | 14.3 | 0.1893 | 0.0799 | 0.2246 | 0.2234 |
| 12.3 | 0.0688 | 0.4419 | 0.1087 | 0.1308 | 15.3 | 0.2218 | 0.3371 | 0.3406 | 0.3324 |
| 13 | 0.0803 | 0.0400 | 0.1280 | 0.0627 | 16 | 0.0019 | | | |
| 13.3 | 0.0363 | 0.0936 | 0.1546 | 0.1117 | 16.3 | 0.0612 | 0.0961 | 0.1449 | 0.0409 |
| 14 | 0.0229 | 0.0025 | 0.0097 | | 17.3 | 0.0076 | 0.0087 | 0.0314 | 0.0163 |
| 14.3 | | 0.0050 | 0.0169 | | 18.3 | 0.0019 | | | |
| 16 | 0.0019 | | | | | | | | |
| H(exp) | 0.7764 | 0.7313 | 0.8069 | 0.7753 | H(exp) | 0.8427 | 0.7882 | 0.7944 | 0.7824 |
| H(obs) | 0.7701 | 0.7100 | 0.8151 | 0.7623 | H(obs) | 0.8161 | 0.7500 | 0.7671 | 0.7295 |
| PIC | 0.7480 | 0.6992 | 0.7802 | 0.7444 | PIC | 0.8237 | 0.7595 | 0.7687 | 0.7518 |
| PDf | 0.9216 | 0.8957 | 0.9360 | 0.9186 | PDf | 0.9562 | 0.9265 | 0.9320 | 0.9221 |
| PDm | 0.7764 | 0.7313 | 0.8069 | 0.7753 | PDm | 0.8427 | 0.7882 | 0.7944 | 0.7824 |
| MECI | 0.7480 | 0.6992 | 0.7802 | 0.7444 | MECI | 0.8237 | 0.7595 | 0.7687 | 0.7518 |
| MECII | 0.6189 | 0.5604 | 0.6575 | 0.6140 | MECII | 0.7145 | 0.6320 | 0.6441 | 0.6230 |
| p (HWE) | 0.7304 | 0.8852 | 0.7232 | 0.0502 | p (HWE) | 0.1264 | 0.3760 | 0.4379 | 0.5786 |
| **DXS101** | | | | | **DXS7424** | | | | |
| 14 | 0.0019 | | | | 9 | 0.0019 | | 0.0024 | |
| 15 | 0.0038 | | 0.0411 | 0.0082 | 10 | 0.0115 | 0.0012 | 0.0048 | 0.0082 |
| 16 | 0.0038 | | | | 11 | 0.0593 | 0.0100 | 0.0048 | 0.0027 |
| 17 | 0.0019 | | 0.0048 | | 12 | 0.0554 | 0.0112 | 0.0459 | 0.0191 |
| 18 | 0.0631 | | 0.0966 | 0.0572 | 13 | 0.2161 | 0.0662 | 0.0580 | 0.1253 |
| 19 | 0.0746 | 0.0087 | 0.0459 | 0.0272 | 14 | 0.2218 | 0.1336 | 0.2005 | 0.1798 |
| 20 | 0.0746 | 0.0050 | 0.0242 | 0.0245 | 15 | 0.1931 | 0.3221 | 0.2536 | 0.2343 |
| 21 | 0.1147 | 0.0087 | 0.0290 | 0.0327 | 16 | 0.1721 | 0.3695 | 0.2464 | 0.2779 |
| 22 | 0.0669 | 0.0549 | 0.0048 | 0.0381 | 17 | 0.0497 | 0.0624 | 0.1377 | 0.1226 |
| 23 | 0.0784 | 0.1186 | 0.0676 | 0.0599 | 18 | 0.0153 | 0.0212 | 0.0386 | 0.0245 |
| 24 | 0.1033 | 0.2784 | 0.1908 | 0.2507 | 19 | 0.0019 | 0.0025 | 0.0048 | 0.0054 |
| 25 | 0.0975 | 0.2235 | 0.1812 | 0.1580 | 20 | 0.0019 | | 0.0024 | |
| 26 | 0.0975 | 0.1735 | 0.1473 | 0.2125 | H(exp) | 0.8278 | 0.7329 | 0.8088 | 0.8037 |
| 27 | 0.1166 | 0.0749 | 0.0652 | 0.0736 | H(obs) | 0.8448 | 0.7400 | 0.8562 | 0.7951 |
| 28 | 0.0612 | 0.0387 | 0.0507 | 0.0300 | PIC | 0.8050 | 0.6913 | 0.7820 | 0.7757 |
| 29 | 0.0229 | 0.0100 | 0.0290 | 0.0136 | PDf | 0.9476 | 0.8871 | 0.9367 | 0.9335 |
| 30 | 0.0096 | 0.0050 | 0.0193 | 0.0109 | PDm | 0.8278 | 0.7329 | 0.8088 | 0.8037 |
| 31 | 0.0038 | | 0.0024 | | MECI | 0.8050 | 0.6913 | 0.7820 | 0.7757 |
| 33 | 0.0038 | | | 0.0027 | MECII | 0.6893 | 0.5526 | 0.6598 | 0.6515 |
| H(exp) | 0.9134 | 0.8180 | 0.8819 | 0.8496 | p (HWE) | 0.4000 | 0.7182 | 0.7307 | 0.7423 |
| H(obs) | 0.9023 | 0.8433 | 0.8493 | 0.8525 | | | | | |
| PIC | 0.9067 | 0.7945 | 0.8709 | 0.8337 | **DXS10147** | | | | |
| PDf | 0.985{{}}8 | 0.9434 | 0.9751 | 0.9614 | 5 | 0.0019 | 0.0012 | | |
| PDm | 0.9134 | 0.8180 | 0.8819 | 0.8496 | 6 | 0.1300 | 0.2772 | 0.2198 | 0.3406 |
| MECI | 0.9067 | 0.7945 | 0.8709 | 0.8337 | 7 | 0.2811 | 0.1985 | 0.0386 | 0.0599 |
| MECII | 0.8350 | 0.6762 | 0.7817 | 0.7295 | 8 | 0.3633 | 0.4757 | 0.2995 | 0.4469 |
| p (HWE) | 0.4268 | **0.0003** | 0.0956 | 0.5742 | 9 | 0.1740 | 0.0474 | 0.4130 | 0.1390 |
| | | | | | 10 | 0.0440 | | 0.0242 | 0.0136 |
| | | | | | 11 | 0.0057 | | 0.0048 | |
| | | | | | H(exp) | 0.7398 | 0.6552 | 0.6893 | 0.6612 |
| | | | | | H(obs) | 0.7069 | 0.6400 | 0.6370 | 0.6557 |
| | | | | | PIC | 0.6970 | 0.5950 | 0.6323 | 0.6001 |
| | | | | | PDf | 0.8895 | 0.8209 | 0.8464 | 0.8241 |
| | | | | | PDm | 0.7398 | 0.6552 | 0.6893 | 0.6612 |
| | | | | | MECI | 0.6970 | 0.5950 | 0.6323 | 0.6001 |
| | | | | | MECII | 0.5574 | 0.4473 | 0.4866 | 0.4540 |
| | | | | | p (HWE) | 0.5620 | 0.4224 | **0.0356** | 0.6517 |

Several alleles that had not previously been noted in the literature at the time of publication were observed in this dataset: 10.1 at DXS9902; 6, 8, 16 and 17 at DXS6795; 5 and 11 at DXS10147; 33 at DXS101; and 16 at DXS6803 (Table 4.4). These new alleles are rare, occurring primarily in populations for which little published genetic data are available. While new alleles 5 and 11 at DXS10147 and 33 at DXS101 were observed in multiple population groups, the 10.1 allele at DXS9902 was observed exclusively in the U.S. Hispanic population group. Similarly, new alleles at DXS6795 and DXS6803 were observed only in the African American population. The structure of the repeat regions of these novel alleles generally followed the standard motif except for marker DXS6803, which was shown to have an irregular TCCA repeat with a stretch of the regular TCTA repeat. Given that microvariant alleles containing an incomplete TCA repeat have also been observed at this marker, the discovery of this structure indicates that the variation present within this repeat region may be largely unexplored.

**Table 4.4. Novel alleles observed in U.S. populations of sample set A.** Nine alleles never previously noted in the literature were observed in this dataset. Eight of these alleles were sequenced and the repeat structure is presented below. N = number of times allele observed in population. *Repeat structure was unable to be determined because only female heterozygote samples were available for analysis and allele separation obtained was insufficient to distinguish with certainty the number of CTT repeats versus ATT repeats within this allele.

| Marker name | Allele | Repeat structure (if sequenced) | Population | N |
|---|---|---|---|---|
| DXS9902 | 10.1 | *Not sequenced* | U.S. Hispanic | 2 |
| DXS6795 | 6 | (ATT)6 | African American | 1 |
| | 8 | (ATT)8 | African American | 1 |
| | 16 | (ATT)16 | African American | 4 |
| | 17 | (ATT)17 | African American | 1 |
| DXS10147 | 5 | (AAAC)5 | African American | 1 |
| | | | U.S. Asian | 1 |
| | 11 | (AAAC)11 | African American | 3 |
| | | | U.S. Caucasian | 2 |
| DXS101 | 33 | *Unable to determine** | African American | 2 |
| | | | U.S. Hispanic | 1 |
| DXS6803 | 16 | (TCTA)12-TCCA-(TCTA)3 | African American | 1 |

The rare 8 allele at marker DXS7423 has thus far only been observed in populations from Africa or with African admixture, and an African specificity of this allele has been postulated [286]. The data presented here seem to be in agreement with this hypothesis, as the 8 allele was observed solely in the African American population (Table 4.3). Other rare alleles were noted in populations in which they had not been

previously observed. For example, alleles 7 and 10.3 at DXS6803 as well as 13 at GATA165B12 have previously been seen in Asian populations [54,85,98,288,289] but were noted in the African American and/or U.S. Hispanic population groups exclusively in this study. Similarly, microvariant alleles at marker DXS7132 (15.3, 16.3, 17.3, 18.3) have only been observed in South American individuals from Brazil, Argentina, and Columbia [185], suggesting a Native American origin of these alleles [286]. Here, the only microvariant observed (16.3) was found in the African American population group.

*4.1.1.1. Null alleles*

Null alleles were noted for four different markers: GATA172D05, GATA165B12, GATA31E08, and DXS7132 (Table 4.5). Loss of an allele at GATA172D05 was observed in one U.S. Hispanic sample. Sequencing of the sample revealed a nucleotide substitution (G→A) in the reverse primer binding site located 7 bases from the 3' end. The same mutation and resultant null allele has been observed once before in a U.S. Hispanic sample, where 377 chromosomes were studied [70]. Here, the frequency was even less, 1 in 621 male chromosomes, though it cannot be ruled out that an additional allele carrying this mutation went undetected amongst the females. Similarly, one male null was observed for each of the other three markers: African American samples at GATA165B12 and GATA31E08, and a U.S. Hispanic sample at DXS7132. Sequencing confirmed the location of a nucleotide substitution under a primer binding site and the correct allele call for all samples. Due to the rarity of these mutations, no measures were taken to change the primer sequences for these markers.

**Table 4.5. Null alleles observed in U.S. populations from sample set A.** Four null alleles observed in four U.S. populations are the result of primer binding site mutations. N = number of times null allele observed in population. *Position refers to the number of base pairs from the 3' end of the indicated primer.

| Marker name | Base change | Position* | Primer orientation | Null allele | Population | N |
|---|---|---|---|---|---|---|
| GATA172D05 | G→A | 7 | Reverse | 12 | U.S. Hispanic | 1 |
| GATA165B12 | C→T | 9 | Forward | 11 | African American | 1 |
| GATA31E08 | A→G | 1 | Forward | 11 | African American | 1 |
| DXS7132 | T→C | 3 | Forward | 15 | U.S. Hispanic | 1 |

*4.1.1.2. Forensic efficiency parameters*

Forensic efficiency parameters calculated for all four population groups are shown in Table 4.3. Marker DXS101 showed the highest observed heterozygosity values for the African American (0.9023) and U.S. Asian (0.8433) populations while the highest value for the U.S. Caucasian population was at DXS7424 (0.8562) and at GATA31E08 for the U.S. Hispanic population (0.8607). The lowest values varied by population, none of which exhibited the lowest value at the same marker. The marker with the lowest observed heterozygosity value was DXS8378 (0.6322) in the African American population, DXS7423 (0.5233) in the U.S. Asian population, DXS6795 (0.5685) in the U.S. Caucasian population, and GATA165B12 (0.6148) in the U.S. Hispanic population. All markers, however, possessed high forensic efficiency values in the majority of the studied populations, confirming their utility for forensic purposes. The high MEC values for the selected markers support the potential of the two multiplexes in a certain specific kinship situations involving female offspring, while the values for the power of discrimination in both males and females indicate their usefulness in forensic identity testing.

Departures from Hardy-Weinberg equilibrium (indicated by a p value for the exact test that is less than 0.05; shown in bold in Table 4.3) occurred in only 2 of the 60 marker-population combinations: the U.S. Caucasian population at DXS10147 ($p = 0.0356$) and the U.S. Asian population at DXS101 ($p = 0.0003$). These deviations observed could potentially be due to population sampling effects.

*4.1.1.3. Comparisons of pairs of population samples by marker*

Population pairwise $F_{ST}$ and corresponding p values were calculated at each marker for the four U.S. population groups using male and female allele frequency data (Table 4.6). Only 10 pairs of populations out of 90 comparisons were not found to be significantly different from one another (indicated by a p value greater than 0.05). Consequently, the four U.S. populations should not be pooled into a single database for use with this set of 15 X STR markers, but rather each should be treated independently. This result concerning the genetic structure of U.S. populations is consistent with similar findings in studies of both X and Y STR markers [70,290,291].

**Table 4.6. Pairwise $F_{ST}$ values (below diagonal) and corresponding p values with standard deviations (above diagonal) comparing individual populations from sample set A by marker.** P values indicative of populations that are not significantly different from one another ($p > 0.5$) are bolded. AA: African American; AS: U.S. Asian; CN: U.S. Caucasian, Hisp: U.S. Hispanic.

Table appears on next page.

| Marker | Population | AA | AS | CN | Hisp |
|---|---|---|---|---|---|
| DXS101 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | AS | 0.04367 | * | 0.00000±0.0000 | 0.00059±0.0002 |
| | CN | 0.01818 | 0.01533 | * | 0.00554±0.0008 |
| | Hisp | 0.02743 | 0.00705 | 0.00510 | * |
| DXS6789 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | AS | 0.04272 | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.04237 | 0.11292 | * | 0.04871±0.0021 |
| | Hisp | 0.03781 | 0.09242 | 0.00372 | * |
| DXS6795 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | AS | 0.09782 | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.123 | 0.11257 | * | 0.00000±0.0000 |
| | Hisp | 0.05502 | 0.03749 | 0.07062 | * |
| DXS6803 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | AS | 0.11858 | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.02293 | 0.08661 | * | 0.00257±0.0005 |
| | Hisp | 0.03087 | 0.08656 | 0.00807 | * |
| DXS7130 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 | 0.00010±0.0001 |
| | AS | 0.03067 | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.02339 | 0.03654 | * | 0.00000±0.0000 |
| | Hisp | 0.01027 | 0.02844 | 0.01304 | * |
| DXS7132 | AA | * | 0.00010±0.0001 | 0.04703±0.0023 | **0.22602±0.0043** |
| | AS | 0.00768 | * | 0.00000±0.0000 | 0.00228±0.0005 |
| | CN | 0.00333 | 0.02093 | * | 0.01822±0.0013 |
| | Hisp | 0.00095 | 0.00748 | 0.00597 | * |
| DXS7423 | AA | * | 0.00000±0.0000 | 0.00020±0.0001 | 0.00000±0.0000 |
| | AS | 0.0553 | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.0152 | 0.03696 | * | 0.00267±0.0005 |
| | Hisp | 0.04549 | 0.01881 | 0.01183 | * |
| DXS7424 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | AS | 0.05403 | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.02521 | 0.01862 | * | **0.10949±0.0030** |
| | Hisp | 0.01741 | 0.01468 | 0.00207 | * |
| DXS8378 | AA | * | 0.00000±0.0000 | **0.05712±0.0025** | 0.01059±0.0012 |
| | AS | 0.08428 | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.00358 | 0.0484 | * | **0.63192±0.0049** |
| | Hisp | 0.0073 | 0.04036 | -0.0013 | * |
| DXS9902 | AA | * | 0.00000±0.0000 | **0.12504±0.0030** | 0.01505±0.0012 |
| | AS | 0.01595 | * | 0.01119±0.0011 | **0.21146±0.0037** |
| | CN | 0.00194 | 0.00621 | * | **0.42451±0.0047** |
| | Hisp | 0.00628 | 0.00104 | -0.00034 | * |
| DXS10147 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | AS | 0.03961 | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.08016 | 0.12549 | * | 0.00000±0.0000 |
| | Hisp | 0.06533 | 0.02228 | 0.07412 | * |
| GATA31E08 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | AS | 0.02229 | * | 0.00673±0.0008 | 0.00000±0.0000 |
| | CN | 0.02613 | 0.00492 | * | 0.00000±0.0000 |
| | Hisp | 0.01811 | 0.02281 | 0.01674 | * |
| GATA165B12 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | AS | 0.07816 | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.01685 | 0.07185 | * | 0.00020±0.0001 |
| | Hisp | 0.02204 | 0.01748 | 0.01842 | * |
| GATA172D05 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | AS | 0.06754 | * | 0.00000±0.0000 | 0.00079±0.0003 |
| | CN | 0.04656 | 0.01649 | * | **0.07989±0.0032** |
| | Hisp | 0.05494 | 0.00831 | 0.00268 | * |
| HPRTB | AA | * | 0.00010±0.0001 | **0.14434±0.0033** | 0.00267±0.0005 |
| | AS | 0.01068 | * | 0.00040±0.0002 | **0.86496±0.0037** |
| | CN | 0.00145 | 0.00932 | * | 0.00545±0.0007 |
| | Hisp | 0.00809 | -0.00142 | 0.00835 | * |

At the time of writing, there were few publications that investigated the distributions of X STR alleles in U.S. population groups, and none that encompassed all 15 markers and 4 population groups from this study. However, seven markers (DXS8378, HPRTB, GATA172D05, DXS7423, DXS7132, DXS101, and DXS6789) and three population groups (African Americans, U.S. Asians, and U.S. Hispanics) from [70] overlap with those studied here. A comparison of the allele frequency distributions across these seven markers for the three population groups was conducted using $F_{ST}$ and corresponding p values (Table 4.7). In general, corresponding population groups were not significantly different from one another, as expected, except for the U.S. Asian population at marker DXS101 ($p = 0.02$) and the U.S. Hispanic population at markers DXS7423 ($p = 0.02$) and GATA172D05 ($p = 0.004$). However, 16 (7.8%) of the 204 comparisons performed revealed unexpected similarity to one another. The majority (75%) of these pairs involved an association between the published U.S. Hispanic population and the remaining three groups from sample set A. Given the heterogenous nature of Hispanic ancestry within the United States, these results are not surprising, but rather indicate a need to be cautious when dealing with such a population.

Similarly, there was generally no significant difference between the African American, U.S. Asian, and U.S. Caucasian populations presented here, and similar published populations at marker HPRTB [292]. However, the results did show a significant difference between the two U.S. Hispanic populations, as well as unexpected similarities between 7 of the 12 other population pairs (58%). Two additional overlapping populations were compared at marker DXS101; both the African American and U.S. Caucasian populations from this study and those published previously [283] were not found to be significantly different from one another, though the published U.S. Caucasian population also did not differ from the U.S. Hispanic population from sample set A. Overall, however, these results confirm the quality of the data presented here for overlapping markers and populations, as well as the population-specificity of the allelic distributions at these markers.

**Table 4.7. P values (with standard deviations) corresponding to pairwise $F_{ST}$ values (not shown) comparing sample set A populations (top) to published populations (left) at overlapping markers.** P values indicative of populations that are not significantly different from one another ($p > 0.5$) are bolded. Values at the intersection of equivalent populations are shaded in gray. AA: African American; AS: U.S. Asian; CN: U.S. Caucasian, Hisp: U.S. Hispanic.

| Marker | Ref. | Population | AA | AS | CN | Hisp |
|---|---|---|---|---|---|---|
| DXS101 | [70] | AA | **0.26512±0.0039** | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | | AS | 0.00000±0.0000 | 0.01752±0.0014 | 0.00000±0.0000 | 0.00495±0.0007 |
| | | Hisp | 0.00317±0.0006 | 0.00119±0.0003 | **0.22414±0.0040** | **0.11365±0.0032** |
| | [283] | AA | **0.07752±0.0025** | 0.00000±0.0000 | 0.00010±0.0001 | 0.00000±0.0000 |
| | | CN | 0.00010±0.0001 | 0.01158±0.0012 | **0.23988±0.0045** | **0.23027±0.0039** |
| DXS6789 | [70] | AA | **0.38402±0.0048** | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | | AS | 0.00000±0.0000 | **0.41620±0.0055** | 0.00000±0.0000 | 0.00000±0.0000 |
| | | Hisp | 0.00040±0.0002 | 0.00000±0.0000 | **0.11623±0.0036** | **0.36284±0.0045** |
| DXS7132 | [70] | AA | **0.52500±0.0050** | 0.03564±0.0018 | **0.68191±0.0044** | **0.46055±0.0053** |
| | | AS | **0.25473±0.0043** | **0.71013±0.0040** | 0.01950±0.0014 | **0.51886±0.0046** |
| | | Hisp | **0.10375±0.0032** | **0.24364±0.0041** | 0.02317±0.0015 | **0.06960±0.0028** |
| DXS7423 | [70] | AA | **0.16493±0.0036** | 0.00000±0.0000 | 0.00050±0.0002 | 0.00000±0.0000 |
| | | AS | 0.00119±0.0003 | **0.50094±0.0052** | 0.00069±0.0003 | 0.01188±0.0012 |
| | | Hisp | **0.10276±0.0030** | 0.00089±0.0003 | **0.45372±0.0049** | 0.02277±0.0015 |
| DXS8378 | [70] | AA | **0.25394±0.0038** | 0.00000±0.0000 | 0.00772±0.0009 | 0.00139±0.0003 |
| | | AS | 0.00010±0.0001 | **0.09385±0.0030** | 0.00525±0.0007 | 0.00762±0.0009 |
| | | Hisp | **0.19256±0.0039** | 0.00000±0.0000 | **0.52995±0.0050** | **0.77774±0.0045** |
| GATA172D05 | [70] | AA | **0.16355±0.0032** | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | | AS | 0.00000±0.0000 | **0.47738±0.0049** | 0.00119±0.0004 | 0.03425±0.0018 |
| | | Hisp | 0.00267±0.0005 | 0.00000±0.0000 | 0.01356±0.0012 | 0.00396±0.0006 |
| HPRTB | [70] | AA | **0.32927±0.0047** | 0.00030±0.0002 | 0.04079±0.0020 | 0.00386±0.0006 |
| | | AS | 0.00703±0.0009 | **0.76349±0.0037** | 0.02614±0.0017 | **0.61687±0.0054** |
| | | Hisp | **0.65102±0.0047** | **0.19206±0.0036** | **0.30452±0.0042** | **0.34700±0.0043** |
| | [292] | AA | **0.08969±0.0027** | 0.00356±0.0006 | 0.00614±0.0009 | **0.05099±0.0021** |
| | | AS | 0.00000±0.0000 | **0.07583±0.0025** | 0.00050±0.0002 | **0.05445±0.0022** |
| | | CN | **0.07772±0.0026** | **0.32274±0.0046** | **0.14157±0.0032** | **0.57846±0.0045** |
| | | Hisp | **0.12692±0.0032** | 0.00000±0.0000 | **0.88001±0.0031** | 0.00040±0.0002 |

#### *4.1.1.4. Linkage disequilibrium*

The likelihood ratio test for linkage disequilibrium was performed for all pairs of markers in the four population groups using only the female genotype data (Table 4.8). Thirty-one pairs of markers showed significant p values ($p < 0.05$) in at least one population, with two pairs that were significant in three populations. An indication of linkage disequilibrium for DXS101–DXS7424 has been observed previously in German [208] and Brazilian [110] populations, and a recombination study has demonstrated the association between DXS7423 and DXS10147 [236].

This latter pair alone resulted in a p value that remained significant in two populations after the Bonferroni correction ($p < 0.003$) was applied. Ten pairs of markers confirmed suspected associations based upon the classification of markers into the four proposed linkage groups (see Chapter 5 for more on linkage groups). The majority (68%) of the pairs with significant p values were not located within the same linkage group or in close proximity on the X chromosome, and do not remain significant after the Bonferroni correction. Additionally, the association was significant in only a subset of the four studied populations, rather than consistently regardless of population as would be expected. It is therefore possible that the p values for these pairings do not indicate a true linkage, but rather population sampling effects or substructure, and further investigation is necessary.

**Table 4.8. Marker pairs with significant p values at the p = 0.05 level indicating the presence of linkage disequilibrium by population for sample set A.** Only those marker pairs that have p values less than 0.05 are shown. P values remaining significant after Bonferroni correction ($p < 0.003$) are bolded. StDev: standard deviation.

| Marker Pair | Population | P value | StDev |
|---|---|---|---|
| DXS101-DXS6803 | AS | 0.04822 | 0.00212 |
| DXS101-DXS7423 | AS | 0.00683 | 0.00079 |
| DXS101-DXS7424 | AA | 0.00564 | 0.00076 |
| | AS | 0.01564 | 0.00127 |
| | Hisp | 0.01545 | 0.00116 |
| DXS101-DXS9902 | AS | 0.03713 | 0.00179 |
| DXS101-HPRTB | Hisp | 0.03832 | 0.00190 |
| DXS6789-DXS101 | Hisp | 0.01574 | 0.00110 |
| DXS6789-DXS7130 | CN | 0.00713 | 0.00088 |
| DXS6789-GATA31E08 | AA | 0.01396 | 0.00119 |
| | CN | 0.02574 | 0.00193 |
| DXS6795-DXS101 | AS | 0.01911 | 0.00152 |
| | Hisp | 0.02465 | 0.00143 |
| DXS6795-DXS7130 | CN | 0.02960 | 0.00162 |
| DXS6795-DXS7423 | AA | 0.03099 | 0.00170 |
| DXS7423-DXS6803 | CN | 0.03426 | 0.00165 |
| DXS7423-DXS7132 | AA | 0.04881 | 0.00219 |
| DXS7423-DXS10147 | AA | 0.01238 | 0.00115 |
| | AS | **0.00020** | **0.00014** |
| | CN | **0.00000** | **0.00000** |
| DXS7423-GATA31E08 | Hisp | 0.03416 | 0.00185 |
| DXS7424-DXS7130 | CN | 0.02663 | 0.00152 |
| DXS7424-DXS9902 | AA | 0.00386 | 0.00070 |
| | CN | 0.03455 | 0.00190 |
| DXS7424-DXS10147 | CN | 0.03248 | 0.00177 |
| DXS8378-DXS6803 | AA | 0.01475 | 0.00134 |
| DXS8378-DXS7130 | AS | 0.02267 | 0.00152 |
| DXS8378-GATA172D05 | AA | 0.03218 | 0.00170 |
| DXS8378-HPRTB | AA | 0.04455 | 0.00230 |
| | AS | 0.01020 | 0.00095 |
| DXS10147-DXS7130 | Hisp | 0.00891 | 0.00093 |
| DXS10147-DXS7132 | Hisp | 0.01921 | 0.00136 |
| DXS10147-GATA31E08 | AA | 0.02267 | 0.00150 |
| GATA165B12-DXS7130 | CN | 0.03634 | 0.00180 |
| GATA165B12-DXS9902 | AA | 0.01673 | 0.00150 |
| GATA172D05-DXS7423 | Hisp | 0.04525 | 0.00212 |
| GATA172D05-DXS7424 | AA | 0.00901 | 0.00103 |
| GATA172D05-DXS10147 | CN | 0.00515 | 0.00076 |
| GATA31E08-DXS7130 | Hisp | 0.04743 | 0.00208 |

For reference, haplotypes based upon the proposed linkage groups are presented in Appendix B, Tables B1-B3 (except for linkage group 3, which is represented by HPRTB alone in these multiplexes). The most common haplotype for proposed linkage group 1 in sample set A was 10-10-13 (alleles in order of markers on the chromosome: DXS8378-DXS9902-DXS6795), which was observed a total of 36 times, mostly in U.S. Asians. Linkage group 2 haplotypes were all unique in this

sample set, completely discriminating between the 621 males. The most common haplotype from proposed linkage group 4 was 13-8-15 (alleles in order of markers on the chromosome: GATA31E08-DXS10147-DXS7423), which was observed a total of 40 times, also mainly in U.S. Asians.

While these data may help to provide an initial understanding of the power of X chromosomal STR markers treated as both independent markers and linked haplotypes, the population size is too small to provide an accurate depiction of the frequency of each observed haplotype. Additionally, as evidenced by the inconclusive linkage disequilibrium results observed here and elsewhere, further studies should be performed to more thoroughly assess the association between markers and better define the proposed linkage groups.

*4.1.2. Sample Set B*

Allele frequencies obtained for the 15 X chromosomal STR markers in three U.S. population groups are shown in Table 4.9. In total, 144 alleles were observed across 15 markers, with 5 to 17 alleles observed within a particular population at each marker.

**Table 4.9. Allele frequencies and forensic efficiency parameters for 15 X STR markers in three U.S. populations from sample set B.** AA: African American; CN: U.S. Caucasian; Hisp: U.S. Hispanic; N: number of alleles; H(exp): expected heterozygosity; PIC: polymorphism information content; PDf: power of discrimination in females; PDm: power of discrimination in males; MECI: mean exclusion chance in trios involving daughter; MECII: mean exclusion chance in father/daughter duos.

| | DXS6789 | | | DXS7130 | | | DXS9902 | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | |
| N | 260 | 336 | 138 | 260 | 336 | 138 | 260 | 336 | 138 | N |
| 6 | | | | | | | | | | 6 |
| 7 | | | | | | | | 0.0030 | | 7 |
| 8 | | | | | | | 0.0538 | | | 8 |
| 9 | | | | 0.0038 | | | 0.0538 | 0.0387 | 0.0217 | 9 |
| 10 | | | | 0.0269 | 0.0149 | | 0.3154 | 0.3423 | 0.3986 | 10 |
| 10.1 | | | | | | | | | 0.0145 | 10.1 |
| 11 | | | | 0.0615 | 0.0387 | 0.0797 | 0.3500 | 0.3423 | 0.2971 | 11 |
| 11.1 | | | | | | | 0.0077 | 0.0417 | 0.0217 | 11.1 |
| 11.3 | | | | | | | | | | 11.3 |
| 12 | | | | 0.2538 | 0.0833 | 0.1667 | 0.2115 | 0.2173 | 0.2391 | 12 |
| 12.1 | | | | | | | | 0.0030 | | 12.1 |
| 12.3 | | | | | | | | | | 12.3 |
| 13 | | | | 0.1654 | 0.0238 | 0.0652 | 0.0077 | 0.0119 | 0.0072 | 13 |
| 13.3 | | | | 0.0115 | 0.0536 | 0.0217 | | | | 13.3 |
| 14 | | | | 0.0538 | 0.0089 | 0.0145 | | | | 14 |
| 14.3 | | | | 0.1500 | 0.2321 | 0.1957 | | | | 14.3 |
| 15 | 0.2500 | 0.0565 | 0.0290 | | | | | | | 15 |
| 15.3 | | | | 0.2000 | 0.3869 | 0.4058 | | | | 15.3 |
| 16 | 0.1077 | 0.0208 | 0.0362 | | | | | | | 16 |
| 16.3 | | | | 0.0731 | 0.1429 | 0.0435 | | | | 16.3 |
| 17 | 0.0038 | 0.0030 | | | | | | | | 17 |
| 17.3 | | | | | 0.0149 | 0.0072 | | | | 17.3 |
| 18 | 0.0115 | 0.0030 | 0.0072 | | | | | | | 18 |
| 19 | 0.0577 | 0.0179 | 0.0435 | | | | | | | 19 |
| 20 | 0.1731 | 0.3839 | 0.3768 | | | | | | | 20 |
| 21 | 0.2192 | 0.2470 | 0.2971 | | | | | | | 21 |
| 22 | 0.1385 | 0.1696 | 0.1667 | | | | | | | 22 |
| 23 | 0.0346 | 0.0625 | 0.0362 | | | | | | | 23 |
| 24 | 0.0038 | 0.0327 | 0.0072 | | | | | | | 24 |
| 25 | | 0.0030 | | | | | | | | 25 |
| 26 | | | | | | | | | | 26 |
| 27 | | | | | | | | | | 27 |
| 28 | | | | | | | | | | 28 |
| 29 | | | | | | | | | | 29 |
| 30 | | | | | | | | | | 30 |
| 31 | | | | | | | | | | 31 |
| 32 | | | | | | | | | | 32 |
| H(exp) | 0.8251 | 0.7539 | 0.7362 | 0.8335 | 0.7636 | 0.7563 | 0.7270 | 0.7151 | 0.6946 | H(exp) |
| PIC | 0.8020 | 0.7196 | 0.6954 | 0.8128 | 0.7335 | 0.7265 | 0.6795 | 0.6636 | 0.6371 | PIC |
| PDf | 0.9463 | 0.9052 | 0.8896 | 0.9515 | 0.9140 | 0.9108 | 0.8780 | 0.8674 | 0.8493 | PDf |
| PDm | 0.8251 | 0.7539 | 0.7362 | 0.8335 | 0.7636 | 0.7563 | 0.7270 | 0.7151 | 0.6946 | PDm |
| MECI | 0.8020 | 0.7196 | 0.6954 | 0.8128 | 0.7335 | 0.7265 | 0.6795 | 0.6636 | 0.6371 | MECI |
| MECII | 0.6853 | 0.5848 | 0.5570 | 0.6998 | 0.6014 | 0.5926 | 0.5383 | 0.5208 | 0.4919 | MECII |

Table continues on next page.

| | GATA31E08 | | | DXS7424 | | | GATA165B12 | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | |
| N | 260 | 336 | 138 | 261 | 336 | 138 | 260 | 336 | 138 | N |
| 6 | | | | | | | | | | 6 |
| 7 | 0.0192 | | | | | | | | | 7 |
| 8 | 0.0308 | | 0.0217 | | | | 0.0269 | 0.0119 | | 8 |
| 9 | 0.1577 | 0.2143 | 0.2174 | | | | 0.1154 | 0.3423 | 0.1957 | 9 |
| 10 | 0.1192 | 0.0149 | 0.0072 | 0.0153 | 0.0030 | | 0.3269 | 0.3214 | 0.4855 | 10 |
| 10.1 | | | | | | | | | | 10.1 |
| 11 | 0.0962 | 0.2351 | 0.1739 | 0.0613 | 0.0060 | 0.0145 | 0.4231 | 0.2917 | 0.2754 | 11 |
| 11.1 | | | | | | | | | | 11.1 |
| 11.3 | | | | | | | | | | 11.3 |
| 12 | 0.3346 | 0.1964 | 0.2899 | 0.0728 | 0.0387 | 0.0217 | 0.0923 | 0.0327 | 0.0217 | 12 |
| 12.1 | | | | | | | | | | 12.1 |
| 12.3 | | | | | | | | | | 12.3 |
| 13 | 0.1769 | 0.2083 | 0.2174 | 0.2184 | 0.0923 | 0.1159 | 0.0154 | | 0.0217 | 13 |
| 13.3 | | | | | | | | | | 13.3 |
| 14 | 0.0577 | 0.1250 | 0.0725 | 0.2452 | 0.1726 | 0.1667 | | | | 14 |
| 14.3 | | | | | | | | | | 14.3 |
| 15 | 0.0077 | 0.0060 | | 0.1724 | 0.2887 | 0.2174 | | | | 15 |
| 15.3 | | | | | | | | | | 15.3 |
| 16 | | | | 0.1686 | 0.2768 | 0.2971 | | | | 16 |
| 16.3 | | | | | | | | | | 16.3 |
| 17 | | | | 0.0421 | 0.0952 | 0.1377 | | | | 17 |
| 17.3 | | | | | | | | | | 17.3 |
| 18 | | | | 0.0038 | 0.0208 | 0.0290 | | | | 18 |
| 19 | | | | | 0.0060 | | | | | 19 |
| 20 | | | | | | | | | | 20 |
| 21 | | | | | | | | | | 21 |
| 22 | | | | | | | | | | 22 |
| 23 | | | | | | | | | | 23 |
| 24 | | | | | | | | | | 24 |
| 25 | | | | | | | | | | 25 |
| 26 | | | | | | | | | | 26 |
| 27 | | | | | | | | | | 27 |
| 28 | | | | | | | | | | 28 |
| 29 | | | | | | | | | | 29 |
| 30 | | | | | | | | | | 30 |
| 31 | | | | | | | | | | 31 |
| 32 | | | | | | | | | | 32 |
| H(exp) | 0.8045 | 0.8010 | 0.7874 | 0.8240 | 0.7907 | 0.8024 | 0.6911 | 0.6932 | 0.6512 | H(exp) |
| PIC | 0.7804 | 0.7701 | 0.7542 | 0.8005 | 0.7607 | 0.7749 | 0.6394 | 0.6308 | 0.5900 | PIC |
| PDf | 0.9378 | 0.9295 | 0.9217 | 0.9456 | 0.9262 | 0.9334 | 0.8528 | 0.8434 | 0.8172 | PDf |
| PDm | 0.8045 | 0.8010 | 0.7874 | 0.8240 | 0.7907 | 0.8024 | 0.6911 | 0.6932 | 0.6512 | PDm |
| MECI | 0.7804 | 0.7701 | 0.7542 | 0.8005 | 0.7607 | 0.7749 | 0.6394 | 0.6308 | 0.5900 | MECI |
| MECII | 0.6582 | 0.6433 | 0.6243 | 0.6834 | 0.6334 | 0.6505 | 0.4953 | 0.4846 | 0.4425 | MECII |

Table continues on next page.

| | DXS101 | | | DXS6795 | | | GATA172D05 | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | |
| N | 260 | 337 | 138 | 260 | 336 | 138 | 260 | 336 | 138 | N |
| 6 | | | | | | | 0.1885 | 0.1399 | 0.1232 | 6 |
| 7 | | | | | | | 0.0692 | | 0.0362 | 7 |
| 8 | | | | | | | 0.1692 | 0.1756 | 0.1667 | 8 |
| 9 | | | | 0.1231 | 0.3155 | 0.1884 | 0.2923 | 0.0625 | 0.0507 | 9 |
| 10 | | | | 0.3615 | 0.0208 | 0.0725 | 0.1269 | 0.3155 | 0.3116 | 10 |
| 10.1 | | | | | | | | | | 10.1 |
| 11 | | | | 0.1538 | 0.4315 | 0.3116 | 0.0923 | 0.1875 | 0.2101 | 11 |
| 11.1 | | | | | | | | | | 11.1 |
| 11.3 | | | | | | | | | | 11.3 |
| 12 | | | | 0.0808 | 0.0387 | 0.1377 | 0.0615 | 0.1161 | 0.1014 | 12 |
| 12.1 | | | | | | | | | | 12.1 |
| 12.3 | | | | | | | | | | 12.3 |
| 13 | | | 0.0072 | 0.0577 | 0.1875 | 0.2609 | | 0.0030 | | 13 |
| 13.3 | | | | | | | | | | 13.3 |
| 14 | | | 0.0072 | 0.0269 | | 0.0217 | | | | 14 |
| 14.3 | | | | | | | | | | 14.3 |
| 15 | | 0.0297 | 0.0072 | 0.1769 | 0.0060 | 0.0072 | | | | 15 |
| 15.3 | | | | | | | | | | 15.3 |
| 16 | 0.0038 | 0.0030 | | 0.0038 | | | | | | 16 |
| 16.3 | | | | | | | | | | 16.3 |
| 17 | | 0.0089 | | 0.0115 | | | | | | 17 |
| 17.3 | | | | | | | | | | 17.3 |
| 18 | 0.0462 | 0.0861 | 0.0580 | 0.0038 | | | | | | 18 |
| 19 | 0.1077 | 0.0445 | 0.0507 | | | | | | | 19 |
| 20 | 0.0423 | 0.0237 | 0.0362 | | | | | | | 20 |
| 21 | 0.1808 | 0.0326 | 0.0435 | | | | | | | 21 |
| 22 | 0.0769 | 0.0445 | 0.0072 | | | | | | | 22 |
| 23 | 0.0577 | 0.0772 | 0.0580 | | | | | | | 23 |
| 24 | 0.0808 | 0.1721 | 0.1812 | | | | | | | 24 |
| 25 | 0.0769 | 0.1602 | 0.1522 | | | | | | | 25 |
| 26 | 0.1154 | 0.1128 | 0.2246 | | | | | | | 26 |
| 27 | 0.1154 | 0.1009 | 0.0870 | | | | | | | 27 |
| 28 | 0.0462 | 0.0593 | 0.0580 | | | | | | | 28 |
| 29 | 0.0308 | 0.0237 | 0.0072 | | | | | | | 29 |
| 30 | 0.0154 | 0.0178 | 0.0145 | | | | | | | 30 |
| 31 | | 0.0030 | | | | | | | | 31 |
| 32 | 0.0038 | | | | | | | | | 32 |
| H(exp) | 0.9005 | 0.8975 | 0.8714 | 0.7888 | 0.6771 | 0.7740 | 0.8179 | 0.7975 | 0.8031 | H(exp) |
| PIC | 0.8922 | 0.8889 | 0.8590 | 0.7629 | 0.6187 | 0.7388 | 0.7943 | 0.7692 | 0.7763 | PIC |
| PDf | 0.9819 | 0.9809 | 0.9710 | 0.9295 | 0.8373 | 0.9137 | 0.9433 | 0.9307 | 0.9344 | PDf |
| PDm | 0.9005 | 0.8975 | 0.8714 | 0.7888 | 0.6771 | 0.7740 | 0.8179 | 0.7975 | 0.8031 | PDm |
| MECI | 0.8922 | 0.8889 | 0.8590 | 0.7629 | 0.6187 | 0.7388 | 0.7943 | 0.7692 | 0.7763 | MECI |
| MECII | 0.8130 | 0.8082 | 0.7645 | 0.6363 | 0.4727 | 0.6061 | 0.6753 | 0.6430 | 0.6524 | MECII |

Table continues on next page.

| | DXS10147 | | | DXS8378 | | | DXS7132 | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | |
| N | 258 | 336 | 138 | 260 | 336 | 138 | 260 | 336 | 138 | N |
| 6 | 0.1008 | 0.2381 | 0.2754 | | | | | | | 6 |
| 7 | 0.3333 | 0.0476 | 0.0725 | | | | | | | 7 |
| 8 | 0.3333 | 0.2679 | 0.3696 | | | | | | | 8 |
| 9 | 0.1938 | 0.4315 | 0.2536 | | 0.0149 | 0.0290 | | | | 9 |
| 10 | 0.0349 | 0.0149 | 0.0290 | 0.2769 | 0.3393 | 0.4130 | | | | 10 |
| 10.1 | | | | | | | | | | 10.1 |
| 11 | 0.0039 | | | 0.4000 | 0.3393 | 0.3043 | 0.0115 | 0.0089 | | 11 |
| 11.1 | | | | | | | | | | 11.1 |
| 11.3 | | | | | | | | | | 11.3 |
| 12 | | | | 0.2846 | 0.2649 | 0.2319 | 0.0885 | 0.0774 | 0.0725 | 12 |
| 12.1 | | | | | | | | | | 12.1 |
| 12.3 | | | | | | | | | | 12.3 |
| 13 | | | | 0.0385 | 0.0417 | 0.0145 | 0.2231 | 0.3304 | 0.2391 | 13 |
| 13.3 | | | | | | | | | | 13.3 |
| 14 | | | | | | 0.0072 | 0.3615 | 0.3542 | 0.3696 | 14 |
| 14.3 | | | | | | | | | | 14.3 |
| 15 | | | | | | | 0.2423 | 0.1875 | 0.2536 | 15 |
| 15.3 | | | | | | | | | 0.0072 | 15.3 |
| 16 | | | | | | | 0.0654 | 0.0417 | 0.0435 | 16 |
| 16.3 | | | | | | | | | 0.0072 | 16.3 |
| 17 | | | | | | | 0.0077 | | 0.0072 | 17 |
| 17.3 | | | | | | | | | | 17.3 |
| 18 | | | | | | | | | | 18 |
| 19 | | | | | | | | | | 19 |
| 20 | | | | | | | | | | 20 |
| 21 | | | | | | | | | | 21 |
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| 24 | | | | | | | | | | 24 |
| 25 | | | | | | | | | | 25 |
| 26 | | | | | | | | | | 26 |
| 27 | | | | | | | | | | 27 |
| 28 | | | | | | | | | | 28 |
| 29 | | | | | | | | | | 29 |
| 30 | | | | | | | | | | 30 |
| 31 | | | | | | | | | | 31 |
| 32 | | | | | | | | | | 32 |
| H(exp) | 0.7284 | 0.6829 | 0.7172 | 0.6829 | 0.6976 | 0.6807 | 0.7487 | 0.7225 | 0.7335 | H(exp) |
| PIC | 0.6809 | 0.6253 | 0.6655 | 0.6199 | 0.6376 | 0.6199 | 0.7085 | 0.6744 | 0.6889 | PIC |
| PDf | 0.8788 | 0.8419 | 0.8683 | 0.8364 | 0.8486 | 0.8372 | 0.8966 | 0.8749 | 0.8844 | PDf |
| PDm | 0.7284 | 0.6829 | 0.7172 | 0.6829 | 0.6976 | 0.6807 | 0.7487 | 0.7225 | 0.7335 | PDm |
| MECI | 0.6809 | 0.6253 | 0.6655 | 0.6199 | 0.6376 | 0.6199 | 0.7085 | 0.6744 | 0.6889 | MECI |
| MECII | 0.5395 | 0.4789 | 0.5222 | 0.4730 | 0.4920 | 0.4735 | 0.5707 | 0.5326 | 0.5486 | MECII |

Table continues on next page.

| | DXS6803 | | | HPRTB | | | DXS7423 | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | |
| N | 260 | 336 | 138 | 260 | 336 | 138 | 260 | 336 | 138 | N |
| 6 | | | | | | | | | | 6 |
| 7 | 0.0038 | | | | | | | | | 7 |
| 8 | 0.0115 | | | 0.0038 | | | 0.0077 | | | 8 |
| 9 | 0.0462 | | 0.0145 | 0.0346 | 0.0089 | | | | | 9 |
| 10 | 0.0923 | 0.0536 | 0.0580 | 0.0231 | 0.0149 | | | | | 10 |
| 10.1 | | | | | | | | | | 10.1 |
| 11 | 0.3500 | 0.2173 | 0.2319 | 0.0846 | 0.1161 | 0.1087 | | | | 11 |
| 11.1 | | | | | | | | | | 11.1 |
| 11.3 | 0.0154 | 0.0119 | 0.0217 | | | | | | | 11.3 |
| 12 | 0.2615 | 0.2589 | 0.2899 | 0.3154 | 0.2768 | 0.3188 | 0.0154 | | | 12 |
| 12.1 | | | | | | | | | | 12.1 |
| 12.3 | 0.0654 | 0.1607 | 0.2029 | | | | | | | 12.3 |
| 13 | 0.1154 | 0.1488 | 0.0652 | 0.2115 | 0.3631 | 0.3478 | 0.0808 | 0.1220 | 0.0725 | 13 |
| 13.3 | 0.0192 | 0.1399 | 0.0942 | | | | | | | 13.3 |
| 14 | 0.0192 | 0.0030 | 0.0217 | 0.1923 | 0.1667 | 0.1594 | 0.5000 | 0.3363 | 0.2391 | 14 |
| 14.3 | | 0.0060 | | | | | | | | 14.3 |
| 15 | | | | 0.1192 | 0.0327 | 0.0580 | 0.2885 | 0.3512 | 0.4928 | 15 |
| 15.3 | | | | | | | | | | 15.3 |
| 16 | | | | 0.0154 | 0.0208 | 0.0072 | 0.1038 | 0.1518 | 0.1304 | 16 |
| 16.3 | | | | | | | | | | 16.3 |
| 17 | | | | | | | 0.0038 | 0.0387 | 0.0652 | 17 |
| 17.3 | | | | | | | | | | 17.3 |
| 18 | | | | | | | | | | 18 |
| 19 | | | | | | | | | | 19 |
| 20 | | | | | | | | | | 20 |
| 21 | | | | | | | | | | 21 |
| 22 | | | | | | | | | | 22 |
| 23 | | | | | | | | | | 23 |
| 24 | | | | | | | | | | 24 |
| 25 | | | | | | | | | | 25 |
| 26 | | | | | | | | | | 26 |
| 27 | | | | | | | | | | 27 |
| 28 | | | | | | | | | | 28 |
| 29 | | | | | | | | | | 29 |
| 30 | | | | | | | | | | 30 |
| 31 | | | | | | | | | | 31 |
| 32 | | | | | | | | | | 32 |
| H(exp) | 0.7794 | 0.8152 | 0.8071 | 0.7954 | 0.7626 | 0.7342 | 0.6496 | 0.7241 | 0.6723 | H(exp) |
| PIC | 0.7507 | 0.7893 | 0.7810 | 0.7670 | 0.7299 | 0.6904 | 0.5958 | 0.6768 | 0.6278 | PIC |
| PDf | 0.9226 | 0.9399 | 0.9367 | 0.9297 | 0.9109 | 0.8855 | 0.8234 | 0.8766 | 0.8481 | PDf |
| PDm | 0.7794 | 0.8152 | 0.8071 | 0.7954 | 0.7626 | 0.7342 | 0.6496 | 0.7241 | 0.6723 | PDm |
| MECI | 0.7507 | 0.7893 | 0.7810 | 0.7670 | 0.7299 | 0.6904 | 0.5958 | 0.6768 | 0.6278 | MECI |
| MECII | 0.6224 | 0.6683 | 0.6590 | 0.6410 | 0.5976 | 0.5503 | 0.4491 | 0.5350 | 0.4815 | MECII |

Two alleles that had not previously been noted in the literature at the time of writing or in the other U.S. populations studied here (sample sets A or C) were observed in this dataset: 18 at DXS6795 and 13 at DXS101 (Table 4.10). Both were sequenced to confirm repeat structure, which conformed to the reported motif. Two additional alleles were observed for the first time in U.S. populations within this dataset: 15.3 at DXS7132 and 32 at DXS101. Both alleles, however, have been observed in several

published populations. The 15.3 allele at DXS7132 has been noted at low frequencies in populations in Central and South America [83,139,293], the Iberian Peninsula [83], and Asia [136]. These results correspond well with the observation of the only instance of this 15.3 allele in the U.S. Hispanic population. The distribution of the 32 allele at DXS101 is more widespread, having been observed in populations from Europe [87,89,115,200], India [125], Asia [85,154,294], and Northern Africa [118]. In this dataset, the only observation of the 32 allele was in a single African American sample.

**Table 4.10. Novel alleles observed in U.S. populations from sample set B.** Four alleles never previously observed in U.S. populations were noted in this dataset. Three of these alleles were sequenced and the repeat structure is presented below. N = number of times allele observed in population.

| Marker name | Allele | Repeat structure (if sequenced) | Population | N |
|---|---|---|---|---|
| DXS6795 | 18 | (ATT)18 | African American | 1 |
| DXS101 | 13 | (CTT)4-(ATT)9 | U.S. Hispanic | 1 |
| | 32 | *Not sequenced* | African American | 1 |
| DXS7132 | 15.3 | (TCTA)13-(TCA)-(TCTA)2 | U.S. Hispanic | 1 |

*4.1.2.1. Null alleles*

Several examples of reduced or complete suppression of amplification (null alleles) were observed within this dataset (Table 4.11). Ten different null alleles were determined through allele sequencing to be the result of single nucleotide changes occurring under one of the primer binding sites for that particular marker. Often times, the distance from the 3' end of the primer can be correlated to the degree of amplification suppression, with base changes closest to the 3' end inducing complete absence of binding (and therefore amplification).

**Table 4.11. Null alleles observed in U.S. populations from sample set B.** Ten null alleles observed in three U.S. populations are the result of primer binding site mutations. N = number of times null allele observed in population. *Position refers to the number of base pairs from the 3' end of the indicated primer.

| Marker name | Amplification suppression | Base change | Position* | Primer orientation | Null allele | Population | N |
|---|---|---|---|---|---|---|---|
| DXS7130 | Partial | G→A | 11 | Reverse | 11 | U.S. Hispanic | 1 |
| | | | | | 12 | African American | 2 |
| | | | | | 13 | African American | 2 |
| DXS8378 | Partial | C→T | 9 | Reverse | 11 | African American | 1 |
| HPRTB | Partial | AGdel | 14-15 | Reverse | 12 | African American | 1 |
| | | | | | | U.S. Caucasian | 1 |
| DXS6803 | Complete | G→A | 3 | Reverse | 10 | African American | 1 |
| DXS7132 | Complete | C→T | 8 | Forward | 14 | African American | 1 |
| GATA172D05 | Complete | G→A | 7 | Reverse | 9 | U.S. Caucasian | 1 |
| | | | | | 10 | U.S. Caucasian | 1 |
| | | | | | 11 | U.S. Caucasian | 1 |

Null alleles were observed at six different markers (DXS7130, DXS8378, HPRTB, DXS6803, DXS7132, and GATA172D05) and in all three studied populations. Most causal primer binding site mutations occurred under the reverse primer, but the base change at DXS7132 affected the forward primer. Three different DXS7130 null alleles (11, 12, and 13) were observed in 5 samples from two populations (African American and U.S. Hispanic). Sequencing revealed that a single G→A change 11 bases from the 3' end of the reverse primer binding site was responsible for the partially null alleles in all samples. The frequency of this base change is 0.0153 and 0.0071 in the African American and U.S. Hispanic populations respectively. One null 11 allele at DXS8378 in an African American sample was found to be due to a C→T transition located 9 base pairs from the 3' end of the reverse primer binding site. This polymorphism appears to be relatively rare, at a frequency of 0.0038 in the African American population alone. Null 12 alleles were also noted for one sample in each of two populations (African American and U.S. Caucasian) at HPRTB. Sequencing confirmed the presence of a previously published deletion of two bases (AG) [70,295] that happen to fall under the reverse primer binding site in this assay.

Complete suppression of amplification was observed for 5 alleles at three markers (DXS6803, DXS7132, and GATA172D05) in both African American and U.S. Caucasian samples. A null 10 allele at marker DXS6803 in one African American sample was found to be the result of a G→A transition located 3 bases from the 3' end of the reverse primer binding site. Sequencing revealed a C→T transition 8

bases from the 3' end of the forward primer binding site in another African American sample responsible for a null 14 allele at DXS7132. Three different null alleles (9, 10, and 11) were observed in three U.S. Caucasian samples at marker GATA172D05. Sequencing revealed the complete suppression of amplification was due to a previously reported G→A change located 7 bases from the 3' end of the reverse primer binding site [55,70]. The frequency of this polymorphism in the U.S. Caucasian population is 0.0089. See Chapter 6 for more details of the allele sequencing results performed as part of this study for these and other samples.

In addition to the above-mentioned null alleles, there were two African American samples with observed null alleles at marker DXS10147 for which no sequence data were able to be obtained (Figure 4.1). After the standard DXS10147 allele sequencing amplification primers (DXS10147 F and DXS10147 R; Table 3.2) failed to produce an amplicon, amplification primers targeting a larger region surrounding the repeat were designed: DXS10147 F2, DXS10147 R2, and DXS10147 F3 (Table 3.2). Figure 4.2 depicts the location of these primers relative to the STR amplification primers and repeat region. Since these larger targeted regions (433 bp and 546 bp) also did not produce amplicons, it was concluded that the null allele was likely the result of a larger-scale deletion in the region of the STR itself, or the region directly adjacent. No further sequencing was attempted for these samples, and a similar situation was not observed again within any of the populations studied here. Additionally, because of the rarity of this observed loss of amplification, no attempts were made to alter the STR amplification primers routinely used.

**Figure 4.1. STR profiles for two African American samples showing null alleles at DXS10147.**

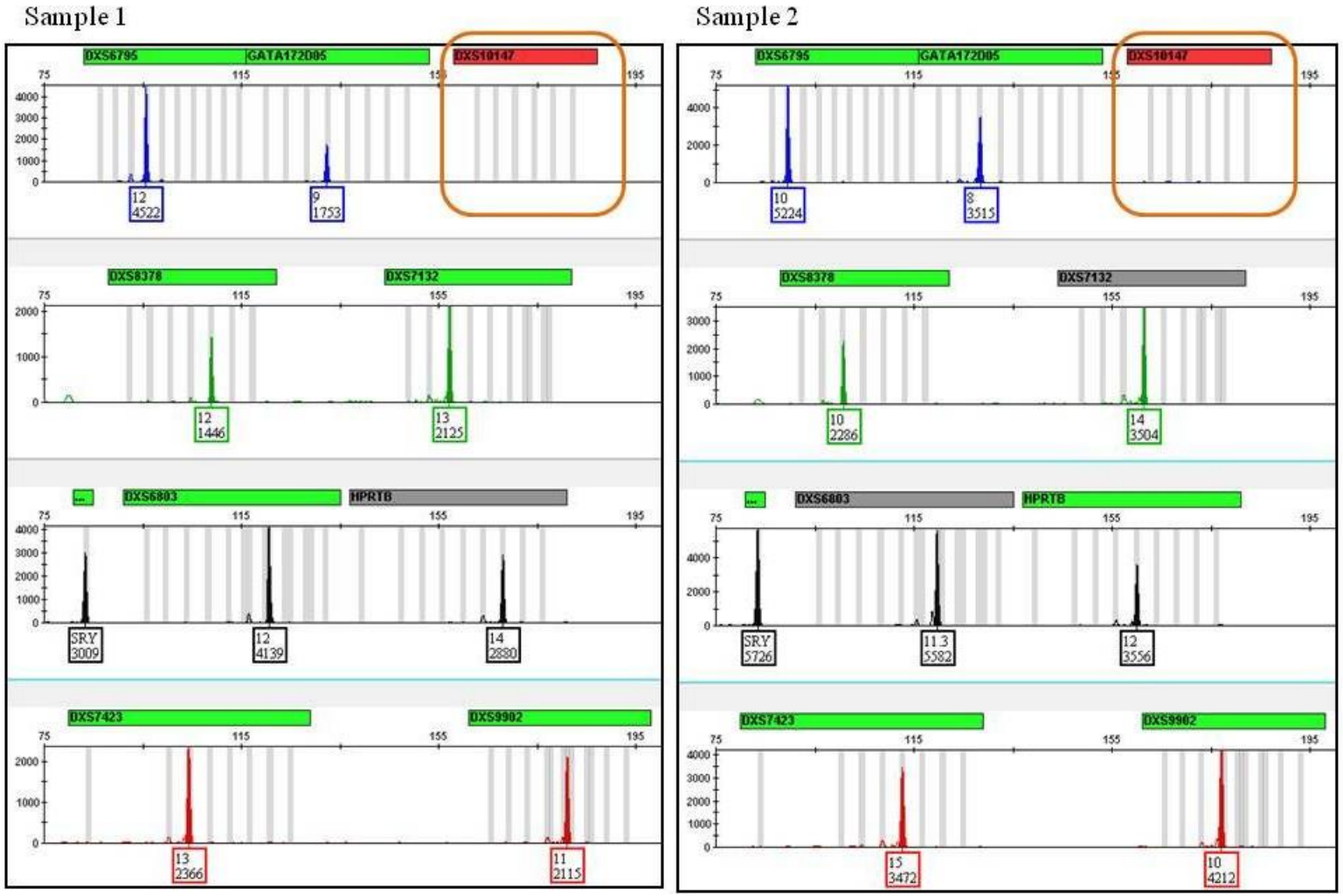


**Figure 4.2. Relative location and resulting amplificon size of various pairs of amplification primers designed for sequencing of marker DXS10147.** The position of the five allele sequencing amplification primers are shown labelled with corresponding sequence underlined. The STR amplification primer sequences are shown in blue. The AAAC repeat region is bolded.

CGGGTGTGGTGGCTCACACCAGTAATCCCAGCACTTTGGGAGACCAAGGTAGGCAGATCACCTGAGGT
DXS10147 F3

CAGGAGTTCAAGACCAGCCTGGCCAACATGGCGAAATCCTGTCTCTACTAAAAATACAAAAATTAGCC
DXS10147

AGGCATGGTGGCATGTGCCTGTAATCCTAGCTACTCAGGAGGCTGAGGCAGGAGAATCCCTTCAACCT
F2 DXS10147 F

AGGAGGTGAAGGTTGTGGTGAGCTGAGATTGTGCCACTACACTCCAGCCTGGGCGACAGAGTGAGATT

CCGTCTCA**AAACAAACAAACAAACAAACAAACAAACAAAC**AACAGCAACAGCAAAAAACCTTAGTGGA

AGACACAATCTAAGCATTTAAGGGAAGAGTCCCAGGCCATTAGTAGATGAAGGCTTTCAGAAGAAGAG

AGGTTGACCTGTCCTGGTCATTATCTGCCTTCAGCTTCCCGGGAATGTGTAGAGCCCACCACCATCA
DXS10147 R

GGACACTAGCAGAGTTGTGGACATGGTATTGGATATGCAGAAGAAATATGGCCAAAGAGATTGGACTG

CCCTCTGTGTTTTGCTTTCAAGCTCACCACAGCCTGCCTCCAGTACCTCCAGTTCCCAGGT
DXS10147 R2

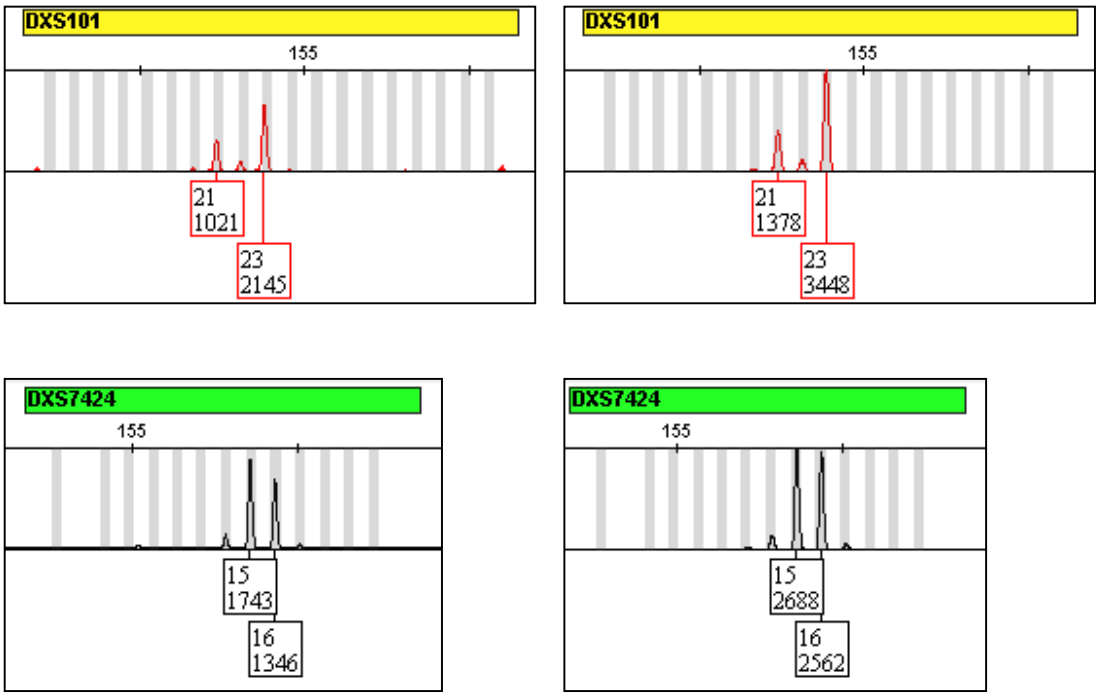
| Primer set | Forward primer | Reverse primer | Amplicon size (bp) |
|---|---|---|---|
| Standard | DXS10147 F | DXS10147 R | 244 |
| Redesigned | DXS10147 F2 | DXS10147 R2 | 433 |
| Redesigned | DXS10147 F3 | DXS10147 R2 | 546 |

*4.1.2.2. Atypical profiles*

Of note, there were several instances of irregular patterns of peaks observed within this dataset. In two instances, there were apparent duplications, which are deduced in male samples by the presence of two alleles (only one is expected) at a single marker (Figure 4.3). One instance occurred in an African American sample at marker DXS7424 while the other was found in a U.S. Caucasian sample at marker DXS101. Interestingly, these two markers are physically close to one another on the chromosome, with linkage between them having been previously documented [110,208]. Each sample was reamplified at least twice using a multiplex reaction and once using a singleplex reaction to confirm the reproducibility of the observed duplications. The presence of two alleles at defined peak height ratios was consistent across all amplifications. Additionally, autosomal (Identifiler®) and Y (Yfiler®) STR typing was performed and confirmed the presence of a single male in each case. Since the duplications appeared to be authentic according to these

criteria, no corrective action was taken to account for these two anomalies, and the additional alleles were included in the population database.

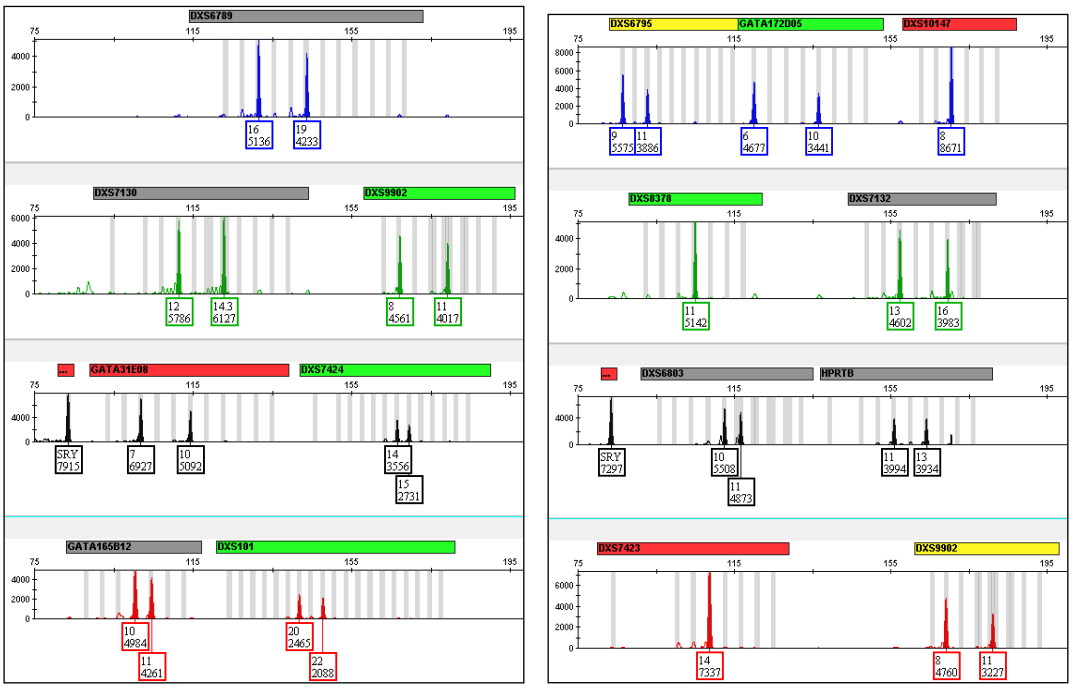
**Figure 4.3. Presumed duplications observed in U.S. populations from sample set B.** Multiplex amplification results are shown in the left panels, and singleplex amplification results are shown in the right panels.




Autosomal triallelic patterns are typically characterised by alleles of either unequal (Type I) or equal (Type II) signal intensities [65,296]. Using an analogous definition to describe X STR diallelic patterns in males, the observed duplication at DXS7424 would be categorised as Type II due to an observed average peak height ratio across three amplifications of 86%. Type II mutations are suspected to result from chromosomal rearrangement, either gross or local [65]. Since duplication of the X chromosome itself can be ruled out in this case due to the observation of the expected single allele at each of the other X STR markers as well as a balanced peak height ratio (92%) at amelogenin, this pattern is likely due to a more localised duplication of the region of the chromosome surrounding DXS7424. On the other hand, the duplication at DXS101 revealed an average peak height ratio of 45% across four amplifications, indicating it is a Type I pattern, which is thought to be the result of somatic mutation [65].

Another uncharacteristic profile was observed in an African American sample that was presumed to be male based upon previous autosomal STR testing [259] and the presence of a single SRY peak within the two X multiplex profiles. However, two alleles were observed at 12 of the 15 X STR markers typed (Figure 4.4). Reamplification of both the original extract as well as freshly-collected extract with one X STR multiplex confirmed that the profile was reproducible. To eliminate the possibility of a mixture of two male individuals, autosomal (Identifiler®) and Y (Yfiler®) STR typing was performed, revealing a single-source male profile at all markers. Of note, however, was a reduced peak height ratio of 41% at amelogenin, roughly corresponding to a 2:1 ratio of X chromosomal signal to Y chromosomal signal. Though this observation alone was not definitive evidence of an additional X chromosome present within this individual, the combination of X STR amplification results was compelling. Therefore, all alleles were included in the population database. Alleles that appeared only once were considered to be identical on both copies of the X chromosome (equivalent to a diploid homozygote) and were counted twice during the calculation of allele frequencies.

**Figure 4.4. Electropherograms depicting results of two multiplexes used to amplify 15 X STR markers in a presumed XXY individual from sample set B.**



An XXY karyotype is considered to be a genetic disorder known as Klinefelter's syndrome. Some form of this disorder is estimated to occur in 1 of 500 to 1 of 1000 live births [297]. Many individuals with this karyotype do not show symptoms, however, and the discovery of such information should not be revealed or utilised for forensic purposes. Other chromosomal disorders are also easily detected with a gonosomal assay such as those targeting X STRs, and care should be given to interpretation and reporting of results.

*4.1.2.3. Forensic efficiency parameters*

The PIC for these three populations was higher than 0.59 in all cases. The highest PIC values observed in all three populations were at marker DXS101 (0.8922 in African Americans, 0.8889 in U.S. Caucasians, and 0.8590 in U.S. Hispanics), which also exhibited the largest number of different alleles (20). The marker with the lowest PIC value varied by population: DXS7423 for African American (0.5958), DXS6795 for U.S. Caucasian (0.6187), and GATA165B12 in U.S. Hispanic (0.5900). In general, markers performed similarly across the three populations. Three notable exceptions are DXS7130, DXS6803, and DXS6795, for which PIC values vary by population. Marker DXS7130 has the second highest PIC value in the African American population (0.8128) while its value falls within the middle range of the 15 markers in both the U.S. Caucasian and U.S. Hispanic populations (0.7335 and 0.7265, respectively). Similarly, while PIC values for DXS6803 are the second highest in the U.S. Caucasian and U.S. Hispanic populations (0.7893 and 0.7810, respectively), its relative value within the African American population falls towards the middle (0.7507). Lastly, DXS6795 exhibited the lowest PIC value (0.6187) of the 15 markers in the U.S. Caucasian population but fell within the middle range in both the African American and U.S. Hispanic populations (0.7629 and 0.7388, respectively).

*4.1.2.4. Comparisons of pairs of population samples by marker*

Though the population substructure of the U.S. had been established both for sample set A (Section 4.1.1.3) and within other studies of STR markers [70,290,291], the pairwise $F_{ST}$ values and corresponding p values were calculated for each of the three pairs of sample set B populations at all 15 markers for confirmation (Table 4.12). The majority of pairs (69%) resulted in p values indicative of significant differences

between the populations ($p < 0.05$), but there were 14 comparisons that revealed similarities between pairs. At all but one marker (GATA165B12), the U.S. Caucasian and the U.S. Hispanic populations were comparable, and at an additional 2 markers (DXS7132 and DXS9902), the U.S. Asian and U.S. Hispanic populations showed a likeness. Despite these associations (and two others), the populations were deemed disparate enough to warrant consideration of each group separately for application of this set of markers.

**Table 4.12. Pairwise $F_{ST}$ values (below diagonal) and corresponding p values with standard deviations (above diagonal) comparing populations from sample set B by marker.** P values indicative of populations that are not significantly different from one another ($p > 0.5$) are bolded. AA: African American; CN: U.S. Caucasian, Hisp: U.S. Hispanic.

| Marker | Population | AA | CN | Hisp |
|---|---|---|---|---|
| DXS101 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.02148 | * | **0.06128±0.0022** |
| | Hisp | 0.02351 | 0.00395 | * |
| DXS6789 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.05336 | * | **0.58885±0.0049** |
| | Hisp | 0.05584 | -0.00162 | * |
| DXS6795 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.15696 | * | 0.00040±0.0002 |
| | Hisp | 0.10055 | 0.02672 | * |
| DXS6803 | AA | * | 0.00000±0.0000 | 0.00099±0.0003 |
| | CN | 0.02476 | * | **0.17582±0.0038** |
| | Hisp | 0.02097 | 0.00273 | * |
| DXS7130 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.05586 | * | 0.01901±0.0015 |
| | Hisp | 0.03376 | 0.01033 | * |
| DXS7132 | AA | * | 0.03247±0.0017 | **0.95099±0.0021** |
| | CN | 0.00643 | * | **0.16543±0.0035** |
| | Hisp | -0.00451 | 0.00325 | * |
| DXS7423 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.02159 | * | 0.00604±0.0007 |
| | Hisp | 0.07244 | 0.0175 | * |
| DXS7424 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.02883 | * | **0.32759±0.0047** |
| | Hisp | 0.02358 | 0.00065 | * |
| DXS8378 | AA | * | **0.17731±0.0036** | 0.01129±0.0010 |
| | CN | 0.00243 | * | **0.27235±0.0046** |
| | Hisp | 0.01765 | 0.00138 | * |
| DXS9902 | AA | * | **0.37264±0.0048** | **0.19305±0.0036** |
| | CN | 0.00003 | * | **0.52708±0.0049** |
| | Hisp | 0.00303 | -0.00149 | * |
| DXS10147 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.10114 | * | 0.00059±0.0002 |
| | Hisp | 0.06463 | 0.02674 | * |
| GATA31E08 | AA | * | 0.00000±0.0000 | 0.01653±0.0014 |
| | CN | 0.03162 | * | **0.11484±0.0031** |
| | Hisp | 0.01069 | 0.00424 | * |
| GATA165B12 | AA | * | 0.00000±0.0000 | 0.00040±0.0002 |
| | CN | 0.04654 | * | 0.00089±0.0003 |
| | Hisp | 0.03199 | 0.02901 | * |
| GATA172D05 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.05877 | * | **0.91506±0.0027** |
| | Hisp | 0.05789 | -0.00364 | * |
| HPRTB | AA | * | 0.00020±0.0001 | 0.01564±0.0011 |
| | CN | 0.01782 | * | **0.83309±0.0033** |
| | Hisp | 0.01213 | -0.00327 | * |

At the time of writing, there were a limited number of publications examining populations and markers similar to those studied here in sample set B. However, there were 3 studies in particular that presented allele frequencies for some

combination of the markers used here, and the p values for the pairwise $F_{ST}$ values resulting from comparison to sample set B populations are shown in Table 4.13. As expected, like populations generally showed no significant differences from one another, with the exception of the U.S. Hispanic populations from sample set B and Gomez, *et al.* [70] at markers DXS7423 and GATA172D05, and the African American populations from sample set B and Edwards, *et al.* [292] at marker HPRTB. Additionally, the U.S. Caucasian population from sample set B was similar to all published U.S. Hispanic populations at overlapping markers except one, DXS7132, where it instead showed similarity to the published African American population.

**Table 4.13. P values (with standard deviations) corresponding to pairwise $F_{ST}$ values (not shown) comparing sample set B populations (top) to published populations (left) at overlapping markers.** P values indicative of populations that are not significantly different from one another ($p > 0.5$) are bolded. Values at the intersection of equivalent populations are shaded in gray. AA: African American; CN: U.S. Caucasian, Hisp: U.S. Hispanic.

| Marker | Ref. | Population | AA | CN | Hisp |
|---|---|---|---|---|---|
| DXS101 | [70] | AA | **0.33452±0.0046** | 0.00000±0.0000 | 0.00010±0.0001 |
| | | Hisp | 0.00000±0.0000 | **0.72587±0.0047** | **0.63271±0.0047** |
| | [283] | AA | **0.39768±0.0042** | 0.00059±0.0002 | 0.00208±0.0004 |
| | | CN | 0.00000±0.0000 | **0.10593±0.0031** | **0.67310±0.0044** |
| DXS6789 | [70] | AA | **0.46461±0.0051** | 0.00020±0.0001 | 0.00020±0.0001 |
| | | Hisp | 0.00000±0.0000 | **0.15137±0.0035** | **0.31898±0.0045** |
| DXS7132 | [70] | AA | **0.57242±0.0044** | **0.52975±0.0050** | **0.57331±0.0049** |
| | | Hisp | **0.48748±0.0054** | 0.00842±0.0008 | **0.26641±0.0042** |
| DXS7423 | [70] | AA | **0.44114±0.0056** | 0.00059±0.0002 | 0.00000±0.0000 |
| | | Hisp | 0.04604±0.0023 | **0.13603±0.0035** | 0.01208±0.0011 |
| DXS8378 | [70] | AA | **0.41164±0.0046** | 0.01921±0.0013 | 0.00168±0.0004 |
| | | Hisp | **0.08564±0.0025** | **0.54737±0.0046** | **0.36462±0.0045** |
| GATA172D05 | [70] | AA | **0.32759±0.0054** | 0.00000±0.0000 | 0.00000±0.0000 |
| | | Hisp | 0.00059±0.0002 | 0.00772±0.0009 | 0.02574±0.0014 |
| HPRTB | [70] | AA | **0.89803±0.0030** | 0.00851±0.0009 | **0.06871±0.0023** |
| | | Hisp | **0.07930±0.0025** | **0.56489±0.0050** | **0.65380±0.0045** |
| | [292] | AA | 0.02574±0.0014 | 0.02267±0.0014 | **0.09702±0.0031** |
| | | CN | 0.00832±0.0009 | **0.05653±0.0023** | **0.50351±0.0051** |
| | | Hisp | 0.00287±0.0006 | **0.29027±0.0043** | **0.66479±0.0045** |

For reference, male haplotypes based upon the proposed linkage groups are presented in Appendix B, Tables B4-B6 (except for linkage group 3, which is represented by HPRTB alone in these multiplexes). The total number of haplotypes observed in this sample set for proposed linkage groups 1, 2, and 4 were 122, 656, and 128, respectively. Within linkage group 1, three individual haplotypes were found to be the most common, each observed 23 times: 10-11-11, 11-11-9, and 12-

10-11 (alleles in order of markers on the chromosome: DXS8378-DXS9902-DXS6795). Each of these haplotypes was observed most often in the U.S. Caucasian population within sample set B, but could also be found to a lesser extent within the other three groups. One linkage group 2 haplotype was found to be common to two U.S. Caucasian samples, but all other 655 haplotypes were unique. The most common haplotype for linkage group 4 was 12-8-14 (alleles in order of markers on the chromosome: GATA31E08-DXS10147-DXS7423), which was observed in a total of 30 samples.

*4.1.3. Sample Set C*

The following publication describes the population genetic analyses of a total of 1214 African American, U.S. Caucasian, and U.S. Hispanic individuals using the two X STR multiplexes described here. Additionally, the publication presents results of a combined U.S. dataset resulting from sample set A and C.

Toni M. Diegoli, Adrian Linacre, Michael D. Coble, **Population genetic data for 15 X chromosomal short tandem repeat markers in three U.S. populations**, Forensic Sci. Int. Genet. 8 (2014) 64-67, doi: 10.1016/j.fsigen.2013.07.008.

Supplementary tables and figure referred to within publication text can be found immediately following or in Appendix B, and are labelled as described in Table 4.14.

**Table 4.14. Table and figure labels corresponding to supplementary material referenced in publication describing sample set C.**

| Label | Publication reference | Title |
|---|---|---|
| Table 4.15 | Supplementary Table 1 | Allele frequencies and summary statistics for 15 X-chromosomal STR markers in 3 U.S. population groups from sample set C |
| Figure 4.5 | Supplementary Figure 1 | Observed triallelic pattern at DXS7132 |
| Table 4.16 | Supplementary Table 2 | Combined allele frequencies and summary statistics for 15 X-chromosomal STR markers in 3 U.S. population groups from sample sets A and C |
| Table 4.17 | Supplementary Table 3 | P values from multi-locus exact test for linkage disequilibrium results for original and combined populations |
| Appendix B, Table B7 | Supplementary Table 4 | Haplotypes observed within sample set C for proposed linkage group 1 |
| Appendix B, Table B8 | Supplementary Table 5 | Haplotypes observed within sample set C for proposed linkage group 2 |
| Appendix B, Table B9 | Supplementary Table 6 | Haplotypes observed within sample set C for proposed linkage group 4 |

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Short communication

# Population genetic data for 15 X chromosomal short tandem repeat markers in three U.S. populations

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ABSTRACT

314 African American, 434 U.S. Caucasian, and 398 U.S. Hispanic individuals were typed at X STR markers DXS6795, DXS9902, DXS8378, DXS7132, DXS6803, DXS6789, DXS7424, DXS101, GATA172D05, DXS7130, GATA165B12, HPRTB, GATA31E08, DXS10147, DXS7423. High forensic efficiency parameter values confirm the potential usefulness of these markers in certain specific kinship situations involving female offspring as well as identity testing. Alleles previously unobserved in U.S. populations were noted in this study at 8 different markers. Additionally, null alleles and a triallelic pattern were observed and described. Pairwise comparisons indicated consistency with similar published populations at overlapping markers. These data represent a substantial increase in the quantity of U.S. X chromosomal short tandem repeat data available to the community.



## 1. Population

Anonymous extracts from unrelated individuals in non-probative paternity cases representing three major U.S. populations (African American, U.S. Caucasian, and U.S. Hispanic) or from six U.S. Caucasian families previously typed for a subset of autosomal STR markers [1] were used to generate U.S. allele and haplotype frequencies. In total, 314 African American (108 males, 206 females), 434 U.S. Caucasian (165 males, 269 females), and 398 U.S. Hispanic (150 males, 248 females) individuals were included in this study. These data serve to greatly increase the amount of X STR information available for U.S. populations, for which only four previous studies exist [2–5]. This project was reviewed and approved by the U.S. Army Medical Research and Materiel Command Institutional Review Board Office.

## 2. PCR amplification and typing

A subset of representative extracts was quantified using Quantifiler® Human DNA Quantification Kit (Applied Biosystems, Foster City, CA) and all extracts were normalized to approximately 1 ng/μL. Profiles were generated for 15 X chromosomal short tandem repeat (X STR) markers (DXS6795, DXS9902, DXS8378, DXS7132, DXS6803, DXS6789, DXS7424, DXS101, GATA172D05, DXS7130, GATA165B12, HPRTB, GATA31E08, DXS10147, DXS7423) using two mini-X STR multiplexes according to the protocol described in [4].

## 3. Analysis of data

Electrophoretic data were analyzed using GeneMapper® ID-X version 1.2 or 1.3 (Applied Biosystems) with custom bins and panels. Allele frequencies and forensic efficiency parameters were generated as described in [4] using PowerMarker version 3.25 [6] and the Forensic ChrX Research website version 2.0 [7]. Pairwise linkage disequilibrium between the 15 markers was also tested using PowerMarker.

## 4. Quality control

Extracts were collected and processed in high-throughput, 96-well plate format with a witness present at the initial plate creation step. Electronic transfer of allele calls from GeneMapper® export files to a master file combining alleles from both multiplexes by sample was accomplished through the use of a custom macro in order to reduce the possibility of transcription errors. A single

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marker (DXS9902) was included in both multiplexes and compared during analysis to ensure concordance for each sample. New alleles were inferred based upon electrophoretic mobility and, when possible, sequenced to confirm repeat structure and length as described in [4]. Sequencing was also performed on all samples that exhibited null alleles. Comparisons at overlapping markers with similar published populations were performed using the chi square test.

## 5. Results and discussion

Allele frequencies and forensic efficiency parameters calculated for each of the 15 markers in three population groups are shown in Supplementary Table 1. In total, 160 alleles were observed across 15 markers, with 7–17 alleles at each marker. Marker DXS101 was the marker with the highest number of observed alleles (17) in the populations studied; DXS8378, GATA165B12 and DXS10147 were the markers with the lowest number (7). DXS101 also exhibited the highest observed heterozygosity values within all three populations (0.9466, 0.8513, and 0.8629 in African Americans, U.S. Caucasians, and U.S. Hispanics respectively) while the lowest heterozygosity values varied by population (0.6845 at DXS8378 in African Americans; 0.6691 at DXS6795 in U.S. Caucasians; 0.6532 at DXS9902 in U.S. Hispanics). The usefulness of certain markers was strongly dependent upon the population to which they were applied. For example, DXS6795 exhibited the second highest observed heterozygosity value (0.8105) in the U.S. Hispanic population, the lowest value (0.6691) in the U.S. Caucasian population, and a value (0.8107) at the midpoint of the 15 markers in the African American population. Overall, the forensic efficiency parameter values confirm the potential usefulness of these markers in certain specific kinship situations involving female offspring as well as identity testing.

Twelve alleles not previously observed in other U.S. populations were noted at 8 markers in this study (Table 1): 9.3, 15, and 15.3 at DXS6803; 17 and 20.2 at DXS7130; 17.3 at DXS7132; 11 and 18 at DXS7423; 10.3 at DXS9902; 13 at DXS10147; 7 at GATA165B12; and 17 at HPRTB. A subset of 6 of these alleles was sequenced to confirm repeat structure. At DXS6803, the sequenced alleles 9.3 and 15.3 conformed to the published repeat structure [4,8] and were observed only in the U.S. Caucasian population. Previously, the 9.3 allele has been observed in Han population of China [9,10] while the 15.3 allele was observed in populations from Croatia [11] and Japan [12]. The 15 allele at DXS6803 had not been previously reported at the time of publication in any world-wide population, and was observed here in two African American samples. Both the 17 and 20.2 alleles newly observed at DXS7130 in U.S. populations exhibited novel repeat structures. The typical repeat motif in non-microvariant DXS7130 alleles is TATC [4,8], but the 17 allele included both this standard TATC as well as one AATC. This allele was observed only once in the African American population, but had been previously observed in populations from the Brazilian Amazon [13], Japanese immigrants residing in Brazil [14], and northwestern China [15]. While microvariant alleles at DXS7130 typically exhibit a $(TATC)_5$-ATC-$(TATC)_x$ pattern [4,8], the 20.2 allele contained an additional partial repeat (ATC). The 20.2 allele was not previously reported at the time of publication, and was observed here in a single U.S. Caucasian sample. Both the 17.3 allele at DXS7132 and the 11 and 18 alleles at DXS7423 were observed previously in non-U.S. populations. Observed here in a single U.S. Hispanic sample, the 17.3 allele at DXS7132 had been observed one time each in populations from Northern Portugal [16], Galicia (Spain) [16], and Nicaragua [17]. The presence of both the 11 and 18 alleles at DXS7423 had been reported in many global populations including those from Europe [16,18,19], Asia [20,21], northern Africa [22], and South America [16,23,24], though each was observed in only one U.S. Caucasian sample in this study. The 10.3 allele at DXS9902 and 13 allele at DXS10147 had not previously been observed in any population at the time of publication. Both the 7 allele at GATA165B12 and 17 allele at HPRTB were consistent with published simple repeat structures [25–27] and were observed for the first time in U.S. populations in the U.S. Hispanic population. Previously, the 7 allele at GATA165B12 had been noted in several Chinese populations [21,28,29] while the 17 allele at HPRTB had been widely detected throughout the world.

A triallelic pattern was observed in one U.S. Caucasian sample at marker DXS7132 (Supplementary Figure 1). Because the three alleles were of even signal intensity, this example was characterized as a Type 2 pattern, which was found to be less frequent than Type 1 triallelic patterns that consist of alleles of unequal signal strength [30,31]. Ancillary testing of this individual's husband, daughter and son revealed transmission of the 13 and 14 allele as a unit from the mother to the son, while the 15 allele was transmitted alone from the mother to the daughter. This inheritance pattern supports the hypothesis that Type 2 triallelic patterns are due to localized duplication events affecting the individual's germ cells [30] rather than X chromosomal aneuploidy, which in this case would be accompanied by the observation of an additional allele at the other 14 markers.

In addition to new or rare alleles, several instances of primer binding site mutations resulting in null alleles were observed in this dataset at 7 markers: DXS101, DXS6795, DXS6803, DXS7130, DXS7132, GATA172D05, and HPRTB (Table 2). One null allele was observed at marker DXS101 in a U.S. Caucasian sample at a frequency of 0.0014. Sequencing revealed a C→T transition located 6 base pairs (bp) from the 3′ end of the reverse primer binding site which resulted in complete suppression of amplification. Two null

**Table 1**
Novel alleles observed in U.S. populations. Twelve alleles never previously observed in U.S. populations were noted in this study. Six of these alleles were sequenced and the repeat structure is presented below.

| Marker name | Allele | Repeat structure (if sequenced) | Population | *N* |
|---|---|---|---|---|
| DXS6803 | 9.3 | $(TCTA)_8$-TCA-TCTA | U.S. Caucasian | 1 |
| | 15 | Not sequenced | African American | 2 |
| | 15.3 | $(TCTA)_{14}$-TCA-(TCTA) | U.S. Caucasian | 2 |
| DXS7130 | 17 | $(TATC)_4$-AATC-$(TATC)_{12}$ | African American | 1 |
| | 20.2 | $(TATC)_5$-ATC-$(TATC)_4$-ATC-$(TATC)_{10}$ | U.S. Caucasian | 1 |
| DXS7132 | 17.3 | Not sequenced | U.S. Hispanic | 1 |
| DXS7423 | 11 | Not sequenced | U.S. Caucasian | 1 |
| | 18 | Not sequenced | U.S. Caucasian | 1 |
| DXS9902 | 10.3 | Not sequenced | U.S. Hispanic | 1 |
| DXS10147 | 13 | Not sequenced | African American | 1 |
| GATA165B12 | 7 | $(AGAT)_7$ | African American | 1 |
| HPRTB | 17 | $(ATCT)_{17}$ | African American | 1 |

*N*, number of times null allele observed in population.

**Table 2**
Null alleles observed in U.S. populations. Ten null alleles observed in three U.S. populations are the result of primer binding site mutations.

| Marker name | Suppression of amplification | Base change | Position[a] | Primer orientation | Null allele | Population | *N* |
|---|---|---|---|---|---|---|---|
| DXS101 | Complete | C→T | 6 | Reverse | 23 | U.S. Caucasian | 1 |
| DXS6795 | Partial | T→C | 15 | Forward | 10 | African American | 6 |
| | | | | | | U.S. Hispanic | 2 |
| | Complete | A→G | 2 | Forward | 11 | U.S. Hispanic | 1 |
| DXS6803 | Partial | C→T | 15 | Reverse | 11 | U.S. Hispanic | 2 |
| DXS7130 | Partial | G→A | 11 | Reverse | 12 | African American | 2 |
| | | | | | 13 | African American | 3 |
| | | | | | 14 | African American | 1 |
| DXS7132 | Partial | C→A | 20 | Reverse | 17 | U.S. Hispanic | 1 |
| GATA172D05 | Complete | G→A | 7 | Reverse | 12 | U.S. Caucasian | 1 |
| HPRTB | Partial | AGdel | 14–15 | Reverse | 12 | U.S. Hispanic | 2 |

*N*, number of times null allele observed in population.
[a] Position refers to the number of base pairs from the 3′ end of the indicated primer.

11 alleles at DXS6803 were found to be caused by a C→T transition under the reverse primer binding site located 15 bp from the 3′ end. In both cases, residual amplification product could be observed under the detection threshold, indicating partial binding of the primer could still occur. This particular null allele was present only in the U.S. Hispanic population (at a frequency of 0.0031), suggesting a possible population-specificity to this particular polymorphism associated with an 11 allele. At DXS6795, a T→C transition 15 bp from the 3′ end of the forward primer resulted in partially null alleles 6 times in the African American population (frequency of 0.0115) and 2 times in the U.S. Hispanic population (frequency of 0.0031). An additional null allele at DXS6795 was observed in one U.S. Hispanic sample due to an A→G transition 2 bp from the 3′ end of the forward primer binding site. Six instances of reduced amplification efficiency resulting in a partially null allele were observed at marker DXS7130 due to a G→A transition in the reverse primer binding site 11 bp from the 3′ end. This polymorphism was observed in combination with a 12, 13, or 14 allele with an overall frequency of 0.0115 in the African American population, making this base change more common than certain rare alleles at this locus (9, 15, 17, and 17.3). Again, the possible population-specificity of the polymorphism was highlighted by its presence within the African American population only. One U.S. Hispanic sample exhibited a partially null 17 allele at DXS7132 due to a C→A transversion at the 5′-most end (20 bp from the 3′ end) of the reverse primer binding site; this was the only observed transversion. At GATA172D05, a null allele was observed at one U.S. Caucasian sample resulting from a G→A transition 7 bp from the 3′ end of the reverse primer binding site. This mutation has been described previously in the U.S. Hispanic population [3,4], but this is the first instance of its observation in a U.S. Caucasian. Lastly, two partially null 12 alleles at HPRTB were observed in the U.S. Hispanic population. A previously noted AG deletion in the flanking region [3,32] fell under the reverse primer binding site used in this study 14 and 15 bp from the 3′ end. Since many of these primer sets are shared with other published X STR assays, knowledge of the frequency of observed null alleles and the populations in which they occur can aid in interpretation. The frequency and diversity of these polymorphisms suggest the additional discriminatory power that may be available should routine sequencing of STR repeats become feasible for forensic laboratories in the future.

Departures from Hardy–Weinberg equilibrium (indicated by a *p*-value for the exact test that is less than 0.05; shown in bold in Supplementary Table 1) occurred in 3 of the 45 marker-population combinations: HPRTB in the African American population ($p = 0.0084$) and U.S. Caucasian population ($p = 0.0420$), and DXS7130 in the U.S. Hispanic population ($p = 0.0229$). After application of the Bonferroni correction, however, none of these values remain significant ($p < 0.0033$).

Previous pairwise population comparisons with X STRs and U.S. populations revealed that the individual groups cannot be combined into one pooled database for forensic use and must instead be treated as three distinct databases [3,4]. This structure was maintained in this study and similarity to other published populations was determined by comparison using the chi square test. Though few studies addressed the same populations investigated here, and none covered all 15 markers, a subset of seven markers (DXS8378, HPRTB, GATA172D05, DXS7423, DXS7132, DXS101, and DXS6789) overlapped with this study for the African American and U.S. Hispanic populations [3]. Significant differences (indicated by a *p* value for the chi square test of less than 0.05) were found between the U.S. Hispanic populations at markers DXS8378 ($p = 0.0179$), DXS7132 ($p = 0.0119$), DXS101 ($p = 0.0300$), and GATA172D05 ($p = 0.0004$) while none were observed for the African American populations. These results may suggest an inherent heterogeneity in self-identified U.S. Hispanic sample populations such as these. Additionally, no significant differences were observed at marker HPRTB from the African American, U.S. Caucasian, and U.S. Hispanic populations described in [2].

The original publication describing the development of the multiplexes used here to amplify the 15 X STR markers included a population study of 349 African Americans, 268 U.S. Caucasians, and 245 U.S. Hispanics [4]. Of note, no significant differences between the three populations present in both studies were observed. Therefore, these databases could be combined to create a larger single database for each group (Supplementary Table 2). When combined, the U.S. populations exhibit similar forensic efficiency statistics with none of the marker-population combinations resulting in a significant deviation from Hardy–Weinberg equilibrium after the Bonferroni correction ($p < 0.0033$).

A test of multi-locus linkage disequilibrium in both the population described in this study as well as the combined population revealed three marker pairs with significant *p* values after the Bonferroni correction ($p < 0.0011$) in both populations (Supplementary Table 3). One of these pairs (DXS6789–GATA165B12) was a set of markers that is not adjacent on the X chromosome, and therefore the association may instead be due to sampling effects. However, the pairs of DXS7423–DXS10147 and DXS10147–GATA31E08 both had *p* values equal to zero in both populations and are adjacent on the chromosome within the originally proposed 4th linkage group [33,34]. These results confirmed a previous study that observed linkage disequilibrium between markers DXS10147 and DXS7423 [35] and indicated that these markers should likely be considered as a haplotype for statistical purposes. Nine additional significant associations (after Bonferroni correction) were noted in the combined population; two of these were between adjacent markers: DXS6803–DXS6789, which are within ~9 Mb of each other, and DXS101–DXS7424, for

which previously reported linkage disequilibrium exists [36,37]. For reference, haplotype frequencies for markers included in each of the originally proposed linkage groups 1, 2, and 4 are presented in Supplementary Tables 4–6 (linkage group 3 is represented by HPRTB alone in this set of markers).

This study substantially increases the volume of X STR data available for U.S. populations while simultaneously confirming the potential utility of the chosen markers for use in both kinship and identity testing. While further investigation of linkage between markers is necessary to fully realize this potential, initial linkage disequilibrium results indicate the need to consider certain pairs or groups of markers as haplotypes.

## 6. Other remarks

This paper followed both the recommendations of the International Society for Forensic Genetics [38] and the guidelines for publication of population data according to [39], and the authors understand and accept the requirements requested therein.

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## Appendix A. Supplementary data

Supplementary material related to this article can be found, in the online version, at doi:10.1016/j.fsigen.2013.07.008.

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**Table 4.15. (Supplementary Table 1). Allele frequencies and summary statistics for 15 X-chromosomal STR markers in 3 U.S. population groups from sample set C.** AA: African American, CN: U.S. Caucasian, Hisp: U.S. Hispanic, N: number of alleles, H(exp): expected heterozygosity, H(obs): observed heterozygosity, PIC: polymorphism information content, PDf: power of discrimination in females, PDm: power of discrimination in males, MECI: mean exclusion chance in trios involving daughter, MECII: mean exclusion chance in father/daughter duos, p (HWE): p value of the exact test for Hardy-Weinberg equilibrium. Significant p values ($p < 0.05$) are shown in bold.

Table begins on next page.

| | DXS6795 | | | DXS9902 | | | DXS8378 | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | |
| N | 520 | 703 | 646 | 520 | 703 | 646 | 520 | 703 | 646 | N |
| 6 | | | | | | | | | | 6 |
| 7 | | | | | | | | | | 7 |
| 8 | | | | 0.0615 | | 0.0046 | | 0.0085 | | 8 |
| 8.3 | | | | | | | | | | 8.3 |
| 9 | 0.1212 | 0.2859 | 0.1300 | 0.0692 | 0.0498 | 0.0108 | 0.0115 | 0.0185 | 0.0170 | 9 |
| 9.3 | | | | | | | | | | 9.3 |
| 10 | 0.3038 | 0.0327 | 0.1068 | 0.3135 | 0.3172 | 0.3622 | 0.2635 | 0.3286 | 0.4443 | 10 |
| 10.3 | | | | | | 0.0015 | | | | 10.3 |
| 11 | 0.1577 | 0.4651 | 0.2415 | 0.3231 | 0.3613 | 0.4241 | 0.3712 | 0.3556 | 0.3313 | 11 |
| 11.1 | | | | 0.0077 | 0.0327 | 0.0279 | | | | 11.1 |
| 11.3 | | | | | | | | | | 11.3 |
| 12 | 0.0827 | 0.0327 | 0.1548 | 0.2096 | 0.2205 | 0.1563 | 0.3115 | 0.2518 | 0.1811 | 12 |
| 12.1 | | | | | 0.0028 | 0.0031 | | | | 12.1 |
| 12.3 | | | | | | | | | | 12.3 |
| 13 | 0.0846 | 0.1792 | 0.3297 | 0.0135 | 0.0156 | 0.0062 | 0.0423 | 0.0356 | 0.0248 | 13 |
| 13.3 | | | | | | | | | | 13.3 |
| 14 | 0.0442 | 0.0014 | 0.0217 | 0.0019 | | 0.0031 | | 0.0014 | 0.0015 | 14 |
| 14.3 | | | | | | | | | | 14.3 |
| 15 | 0.1865 | 0.0028 | 0.0139 | | | | | | | 15 |
| 15.3 | | | | | | | | | | 15.3 |
| 16 | 0.0135 | | 0.0015 | | | | | | | 16 |
| 16.3 | | | | | | | | | | 16.3 |
| 17 | 0.0058 | | | | | | | | | 17 |
| 17.3 | | | | | | | | | | 17.3 |
| 18 | | | | | | | | | | 18 |
| 18.3 | | | | | | | | | | 18.3 |
| 19 | | | | | | | | | | 19 |
| 20 | | | | | | | | | | 20 |
| 20.3 | | | | | | | | | | 20.3 |
| 21 | | | | | | | | | | 21 |
| 22 | | | | | | | | | | 22 |
| 23 | | | | | | | | | | 23 |
| 24 | | | | | | | | | | 24 |
| 25 | | | | | | | | | | 25 |
| 26 | | | | | | | | | | 26 |
| 27 | | | | | | | | | | 27 |
| 28 | | | | | | | | | | 28 |
| 29 | | | | | | | | | | 29 |
| 30 | | | | | | | | | | 30 |
| 31 | | | | | | | | | | 31 |
| PIC | 0.7944 | 0.6118 | 0.7479 | 0.7019 | 0.6655 | 0.6005 | 0.6333 | 0.6424 | 0.5950 | PIC |
| H(exp) | 0.8172 | 0.6677 | 0.7800 | 0.7446 | 0.7164 | 0.6635 | 0.6938 | 0.7005 | 0.6591 | H(exp) |
| H(obs) | 0.8107 | 0.6691 | 0.8105 | 0.7621 | 0.7138 | 0.6532 | 0.6845 | 0.6952 | 0.6653 | H(obs) |
| PDf | 0.9438 | 0.8336 | 0.9194 | 0.8920 | 0.8687 | 0.8238 | 0.8457 | 0.8523 | 0.8197 | PDf |
| PDm | 0.8172 | 0.6677 | 0.7800 | 0.7446 | 0.7164 | 0.6635 | 0.6938 | 0.7005 | 0.6591 | PDm |
| MECI | 0.7944 | 0.6118 | 0.7479 | 0.7019 | 0.6655 | 0.6005 | 0.6333 | 0.6424 | 0.5950 | MECI |
| MECII | 0.6759 | 0.4652 | 0.6171 | 0.5635 | 0.5228 | 0.4547 | 0.4874 | 0.4974 | 0.4483 | MECII |
| p(HWE) | 0.7551 | 0.0707 | 0.3724 | 0.8238 | 0.0879 | 0.8353 | 0.9894 | 0.7500 | 0.9458 | p(HWE) |

Table continues on next page.

| | GATA172D05 | | | DXS7132 | | | DXS6803 | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | |
| N | 520 | 703 | 646 | 520 | 704 | 646 | 520 | 703 | 646 | N |
| 6 | 0.2038 | 0.1636 | 0.1192 | | | | | | | 6 |
| 7 | 0.0692 | | 0.0031 | | | | 0.0077 | | | 7 |
| 8 | 0.1750 | 0.1807 | 0.1455 | | | | 0.0173 | | | 8 |
| 8.3 | | | | | | | | | | 8.3 |
| 9 | 0.2923 | 0.0469 | 0.0588 | | | | 0.0327 | | 0.0031 | 9 |
| 9.3 | | | | | | | | 0.0014 | | 9.3 |
| 10 | 0.1269 | 0.2774 | 0.2817 | | | 0.0015 | 0.1058 | 0.0299 | 0.0170 | 10 |
| 10.3 | | | | | | | | | | 10.3 |
| 11 | 0.1019 | 0.2134 | 0.2988 | 0.0212 | 0.0099 | 0.0015 | 0.3673 | 0.2418 | 0.2771 | 11 |
| 11.1 | | | | | | | | | | 11.1 |
| 11.3 | | | | | | | 0.0058 | 0.0171 | 0.0588 | 11.3 |
| 12 | 0.0288 | 0.1181 | 0.0913 | 0.1038 | 0.0810 | 0.1022 | 0.2404 | 0.2646 | 0.3142 | 12 |
| 12.1 | | | | | | | | | | 12.1 |
| 12.3 | | | | | | | 0.0827 | 0.1323 | 0.1656 | 12.3 |
| 13 | 0.0019 | | 0.0015 | 0.2577 | 0.2827 | 0.2492 | 0.0788 | 0.1536 | 0.0666 | 13 |
| 13.3 | | | | | | | 0.0269 | 0.1366 | 0.0898 | 13.3 |
| 14 | | | | 0.3269 | 0.3608 | 0.3514 | 0.0269 | 0.0043 | 0.0062 | 14 |
| 14.3 | | | | | | | 0.0038 | 0.0128 | 0.0015 | 14.3 |
| 15 | | | | 0.2269 | 0.2045 | 0.2353 | 0.0038 | | | 15 |
| 15.3 | | | | | | | | 0.0057 | | 15.3 |
| 16 | | | | 0.0423 | 0.0497 | 0.0387 | | | | 16 |
| 16.3 | | | | 0.0058 | | 0.0015 | | | | 16.3 |
| 17 | | | | 0.0135 | 0.0099 | 0.0124 | | | | 17 |
| 17.3 | | | | | | 0.0015 | | | | 17.3 |
| 18 | | | | 0.0019 | 0.0014 | 0.0046 | | | | 18 |
| 18.3 | | | | | | | | | | 18.3 |
| 19 | | | | | | | | | | 19 |
| 20 | | | | | | | | | | 20 |
| 20.3 | | | | | | | | | | 20.3 |
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| 29 | | | | | | | | | | 29 |
| 30 | | | | | | | | | | 30 |
| 31 | | | | | | | | | | 31 |
| PIC | 0.7847 | 0.7728 | 0.7526 | 0.7240 | 0.6958 | 0.7052 | 0.7535 | 0.7839 | 0.7492 | PIC |
| H(exp) | 0.8103 | 0.8020 | 0.7842 | 0.7620 | 0.7389 | 0.7469 | 0.7801 | 0.8104 | 0.7808 | H(exp) |
| H(obs) | 0.8447 | 0.7955 | 0.7500 | 0.8301 | 0.7621 | 0.6976 | 0.8204 | 0.7770 | 0.7782 | H(obs) |
| PDf | 0.9384 | 0.9316 | 0.9218 | 0.9053 | 0.8887 | 0.8942 | 0.9251 | 0.9376 | 0.9204 | PDf |
| PDm | 0.8103 | 0.8020 | 0.7842 | 0.7620 | 0.7389 | 0.7469 | 0.7801 | 0.8104 | 0.7808 | PDm |
| MECI | 0.7847 | 0.7728 | 0.7526 | 0.7240 | 0.6958 | 0.7052 | 0.7535 | 0.7839 | 0.7492 | MECI |
| MECII | 0.6628 | 0.6471 | 0.6231 | 0.5890 | 0.5565 | 0.5669 | 0.6260 | 0.6619 | 0.6196 | MECII |
| p(HWE) | 0.2128 | 0.6158 | 0.5084 | 0.2895 | 0.6342 | 0.2715 | 0.9786 | 0.5050 | 0.7068 | p(HWE) |

Table continues on next page.

| | DXS6789 | | | GATA165B12 | | | DXS7130 | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | |
| N | 520 | 703 | 646 | 520 | 703 | 646 | 520 | 703 | 646 | N |
| 6 | | | | | | | | | | 6 |
| 7 | | | | 0.0038 | | | | | | 7 |
| 8 | | | | 0.0404 | 0.0043 | 0.0077 | | | | 8 |
| 8.3 | | | | | | | | | | 8.3 |
| 9 | | | | 0.1038 | 0.3030 | 0.2430 | 0.0038 | | | 9 |
| 9.3 | | | | | | | | | | 9.3 |
| 10 | | | | 0.3269 | 0.3428 | 0.4830 | 0.0269 | 0.0043 | 0.0046 | 10 |
| 10.3 | | | | | | | | | | 10.3 |
| 11 | | | | 0.4154 | 0.3172 | 0.2446 | 0.0673 | 0.0413 | 0.0588 | 11 |
| 11.1 | | | | | | | | | | 11.1 |
| 11.3 | | | | | | | | | | 11.3 |
| 12 | | | | 0.1000 | 0.0327 | 0.0217 | 0.2154 | 0.1067 | 0.1966 | 12 |
| 12.1 | | | | | | | | | | 12.1 |
| 12.3 | | | | | | | | | | 12.3 |
| 13 | | | | 0.0096 | | | 0.1673 | 0.0356 | 0.0542 | 13 |
| 13.3 | | | | | | | 0.0212 | 0.0640 | 0.0248 | 13.3 |
| 14 | 0.0038 | 0.0043 | 0.0093 | | | | 0.0404 | 0.0057 | 0.0201 | 14 |
| 14.3 | | | | | | | 0.1615 | 0.2048 | 0.1687 | 14.3 |
| 15 | 0.2423 | 0.0384 | 0.0341 | | | | 0.0038 | | 0.0031 | 15 |
| 15.3 | | | | | | | 0.2288 | 0.3642 | 0.3560 | 15.3 |
| 16 | 0.1154 | 0.0142 | 0.0325 | | | | | | | 16 |
| 16.3 | | | | | | | 0.0558 | 0.1494 | 0.0882 | 16.3 |
| 17 | 0.0038 | 0.0014 | 0.0046 | | | | 0.0019 | | | 17 |
| 17.3 | | | | | | | 0.0058 | 0.0228 | 0.0232 | 17.3 |
| 18 | 0.0135 | 0.0014 | 0.0031 | | | | | | | 18 |
| 18.3 | | | | | | | | | 0.0015 | 18.3 |
| 19 | 0.0692 | 0.0256 | 0.0341 | | | | | | | 19 |
| 20 | 0.1962 | 0.3912 | 0.3994 | | | | | | | 20 |
| 20.3 | | | | | | | | 0.0014 | | 20.3 |
| 21 | 0.2173 | 0.2717 | 0.3111 | | | | | | | 21 |
| 22 | 0.0981 | 0.1479 | 0.1099 | | | | | | | 22 |
| 23 | 0.0365 | 0.0868 | 0.0495 | | | | | | | 23 |
| 24 | 0.0038 | 0.0171 | 0.0124 | | | | | | | 24 |
| 25 | | | | | | | | | | 25 |
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| 31 | | | | | | | | | | 31 |
| PIC | 0.8036 | 0.7035 | 0.6851 | 0.6483 | 0.6246 | 0.5844 | 0.8164 | 0.7575 | 0.7649 | PIC |
| H(exp) | 0.8263 | 0.7411 | 0.7255 | 0.6981 | 0.6890 | 0.6473 | 0.8367 | 0.7841 | 0.7904 | H(exp) |
| H(obs) | 0.8155 | 0.7286 | 0.6855 | 0.7136 | 0.7175 | 0.7097 | 0.8155 | 0.7881 | 0.7702 | H(obs) |
| PDf | 0.9471 | 0.8953 | 0.8843 | 0.8591 | 0.8389 | 0.8127 | 0.9530 | 0.9268 | 0.9306 | PDf |
| PDm | 0.8263 | 0.7411 | 0.7255 | 0.6981 | 0.6890 | 0.6473 | 0.8367 | 0.7841 | 0.7904 | PDm |
| MECI | 0.8036 | 0.7035 | 0.6851 | 0.6483 | 0.6246 | 0.5844 | 0.8164 | 0.7575 | 0.7649 | MECI |
| MECII | 0.6875 | 0.5661 | 0.5464 | 0.5049 | 0.4780 | 0.4363 | 0.7048 | 0.6300 | 0.6394 | MECII |
| p(HWE) | 0.1127 | 0.3011 | 0.0790 | 0.4856 | 0.1170 | 0.2379 | 0.8648 | 0.4266 | **0.0229** | p(HWE) |

Table continues on next page.

| | DXS101 | | | DXS7424 | | | HPRTB | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | |
| N | 520 | 703 | 646 | 520 | 703 | 646 | 520 | 703 | 646 | N |
| 6 | | | | | | | | | | 6 |
| 7 | | | | | | | | | | 7 |
| 8 | | | | | | | | 0.0014 | | 8 |
| 8.3 | | | | | | | | | | 8.3 |
| 9 | | | | 0.0019 | | | 0.0385 | 0.0043 | | 9 |
| 9.3 | | | | | | | | | | 9.3 |
| 10 | | | | 0.0077 | 0.0043 | 0.0046 | 0.0154 | 0.0057 | 0.0015 | 10 |
| 10.3 | | | | | | | | | | 10.3 |
| 11 | | | | 0.0808 | 0.0057 | 0.0046 | 0.0904 | 0.1294 | 0.0851 | 11 |
| 11.1 | | | | | | | | | | 11.1 |
| 11.3 | | | | | | | | | | 11.3 |
| 12 | | | | 0.0731 | 0.0398 | 0.0464 | 0.2712 | 0.3272 | 0.2817 | 12 |
| 12.1 | | | | | | | | | | 12.1 |
| 12.3 | | | | | | | | | | 12.3 |
| 13 | | | | 0.2250 | 0.0626 | 0.1084 | 0.2577 | 0.3229 | 0.3762 | 13 |
| 13.3 | | | | | | | | | | 13.3 |
| 14 | | | | 0.2750 | 0.2119 | 0.2121 | 0.2212 | 0.1394 | 0.1842 | 14 |
| 14.3 | | | | | | | | | | 14.3 |
| 15 | | 0.0242 | 0.0170 | 0.1692 | 0.2603 | 0.1950 | 0.0923 | 0.0569 | 0.0526 | 15 |
| 15.3 | | | | | | | | | | 15.3 |
| 16 | 0.0019 | 0.0028 | 0.0015 | 0.1192 | 0.2703 | 0.2678 | 0.0135 | 0.0114 | 0.0186 | 16 |
| 16.3 | | | | | | | | | | 16.3 |
| 17 | 0.0058 | 0.0043 | | 0.0365 | 0.1238 | 0.1053 | | 0.0014 | | 17 |
| 17.3 | | | | | | | | | | 17.3 |
| 18 | 0.0327 | 0.0612 | 0.0697 | 0.0096 | 0.0156 | 0.0511 | | | | 18 |
| 18.3 | | | | | | | | | | 18.3 |
| 19 | 0.1000 | 0.0569 | 0.0449 | 0.0019 | 0.0057 | 0.0031 | | | | 19 |
| 20 | 0.0558 | 0.0171 | 0.0155 | | | 0.0015 | | | | 20 |
| 20.3 | | | | | | | | | | 20.3 |
| 21 | 0.1692 | 0.0270 | 0.0263 | | | | | | | 21 |
| 22 | 0.0635 | 0.0199 | 0.0124 | | | | | | | 22 |
| 23 | 0.0635 | 0.0669 | 0.0666 | | | | | | | 23 |
| 24 | 0.0923 | 0.2048 | 0.2399 | | | | | | | 24 |
| 25 | 0.0827 | 0.1821 | 0.1811 | | | | | | | 25 |
| 26 | 0.1038 | 0.1252 | 0.1904 | | | | | | | 26 |
| 27 | 0.1442 | 0.1010 | 0.0820 | | | | | | | 27 |
| 28 | 0.0423 | 0.0640 | 0.0402 | | | | | | | 28 |
| 29 | 0.0250 | 0.0256 | 0.0077 | | | | | | | 29 |
| 30 | 0.0135 | 0.0128 | 0.0015 | | | | | | | 30 |
| 31 | 0.0038 | 0.0043 | 0.0031 | | | | | | | 31 |
| PIC | 0.8911 | 0.8696 | 0.8363 | 0.7936 | 0.7625 | 0.7932 | 0.7619 | 0.7091 | 0.6921 | PIC |
| H(exp) | 0.8995 | 0.8806 | 0.8523 | 0.8176 | 0.7931 | 0.8176 | 0.7926 | 0.7491 | 0.7348 | H(exp) |
| H(obs) | 0.9466 | 0.8513 | 0.8629 | 0.8544 | 0.8253 | 0.8024 | 0.7767 | 0.7881 | 0.6895 | H(obs) |
| PDf | 0.9815 | 0.9748 | 0.9622 | 0.9428 | 0.9266 | 0.9424 | 0.9263 | 0.8971 | 0.8869 | PDf |
| PDm | 0.8995 | 0.8806 | 0.8523 | 0.8176 | 0.7931 | 0.8176 | 0.7926 | 0.7491 | 0.7348 | PDm |
| MECI | 0.8911 | 0.8696 | 0.8363 | 0.7936 | 0.7625 | 0.7932 | 0.7619 | 0.7091 | 0.6921 | MECI |
| MECII | 0.8112 | 0.7799 | 0.7326 | 0.6748 | 0.6354 | 0.6741 | 0.6346 | 0.5719 | 0.5523 | MECII |
| p(HWE) | 0.6232 | 0.3965 | 0.9997 | 0.8320 | 0.3977 | 0.1601 | **0.0083** | **0.0393** | 0.7030 | p(HWE) |

Table continues on next page.

| | DXS7423 | | | DXS10147 | | | GATA31E08 | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | |
| N | 520 | 703 | 646 | 520 | 703 | 646 | 520 | 703 | 646 | N |
| 6 | | | | 0.1385 | 0.2376 | 0.3452 | 0.0019 | | | 6 |
| 7 | | | | 0.2788 | 0.0413 | 0.0604 | 0.0212 | | 0.0077 | 7 |
| 8 | 0.0077 | | | 0.3615 | 0.2959 | 0.3947 | 0.0250 | 0.0014 | 0.0062 | 8 |
| 8.3 | | | | | | | | | | 8.3 |
| 9 | | | | 0.1808 | 0.4139 | 0.1889 | 0.1635 | 0.1878 | 0.1130 | 9 |
| 9.3 | | | | | | | | | | 9.3 |
| 10 | | | | 0.0365 | 0.0114 | 0.0077 | 0.1596 | 0.0284 | 0.0325 | 10 |
| 10.3 | | | | | | | | | | 10.3 |
| 11 | | 0.0014 | | 0.0019 | | 0.0031 | 0.0635 | 0.2020 | 0.1811 | 11 |
| 11.1 | | | | | | | | | | 11.1 |
| 11.3 | | | | | | | | | | 11.3 |
| 12 | 0.0038 | | 0.0015 | | | | 0.2635 | 0.2119 | 0.3808 | 12 |
| 12.1 | | | | | | | | | | 12.1 |
| 12.3 | | | | | | | | | | 12.3 |
| 13 | 0.1115 | 0.1024 | 0.0325 | 0.0019 | | | 0.2135 | 0.2518 | 0.2121 | 13 |
| 13.3 | | | | | | | | | | 13.3 |
| 14 | 0.4500 | 0.3001 | 0.3003 | | | | 0.0692 | 0.0982 | 0.0557 | 14 |
| 14.3 | | | | | | | | | | 14.3 |
| 15 | 0.3212 | 0.3997 | 0.4830 | | | | 0.0192 | 0.0156 | 0.0108 | 15 |
| 15.3 | | | | | | | | | | 15.3 |
| 16 | 0.0981 | 0.1664 | 0.0991 | | | | | 0.0014 | | 16 |
| 16.3 | | | | | | | | | | 16.3 |
| 17 | 0.0077 | 0.0284 | 0.0836 | | | | | 0.0014 | | 17 |
| 17.3 | | | | | | | | | | 17.3 |
| 18 | | 0.0014 | | | | | | | | 18 |
| 18.3 | | | | | | | | | | 18.3 |
| 19 | | | | | | | | | | 19 |
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| 20.3 | | | | | | | | | | 20.3 |
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| 31 | | | | | | | | | | 31 |
| PIC | 0.6165 | 0.6623 | 0.6049 | 0.6945 | 0.6225 | 0.6266 | 0.7994 | 0.7759 | 0.7268 | PIC |
| H(exp) | 0.6721 | 0.7112 | 0.6587 | 0.7384 | 0.6829 | 0.6856 | 0.8226 | 0.8049 | 0.7601 | H(exp) |
| H(obs) | 0.7184 | 0.6952 | 0.6976 | 0.7816 | 0.6691 | 0.7097 | 0.8107 | 0.8216 | 0.7258 | H(obs) |
| PDf | 0.8369 | 0.8677 | 0.8297 | 0.8877 | 0.8390 | 0.8421 | 0.9453 | 0.9329 | 0.9092 | PDf |
| PDm | 0.6721 | 0.7112 | 0.6587 | 0.7384 | 0.6829 | 0.6856 | 0.8226 | 0.8049 | 0.7601 | PDm |
| MECI | 0.6165 | 0.6623 | 0.6049 | 0.6945 | 0.6225 | 0.6266 | 0.7994 | 0.7759 | 0.7268 | MECI |
| MECII | 0.4709 | 0.5190 | 0.4587 | 0.5543 | 0.4760 | 0.4809 | 0.6822 | 0.6512 | 0.5925 | MECII |
| p(HWE) | 0.9471 | 0.1218 | 0.7420 | 0.0637 | 0.8893 | 0.5185 | 0.2097 | 0.6188 | 0.3914 | p(HWE) |

**Figure 4.5. (Supplementary Figure 1). Observed triallelic pattern at DXS7132.** A single U.S. Caucasian sample (mother) in the studied populations revealed a Type 2 triallelic pattern. Ancillary testing of additional family members (son, daughter, husband) revealed an inheritance pattern consistent with localized duplication of the region containing the STR repeat. The 13 and 14 alleles were passed as a unit from the mother to her son, whose profile would typically exhibit only one X STR allele per marker. The 15 allele was passed separately to the daughter, whose 16 allele was inherited from her father (husband).

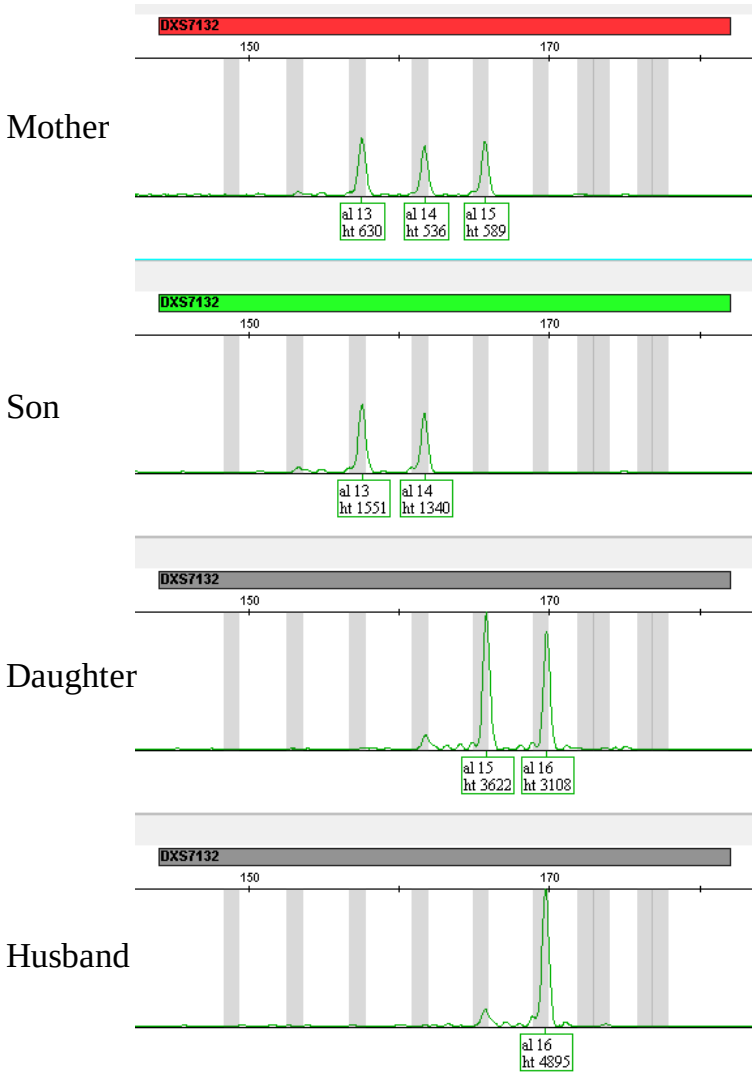


**Table 4.16. (Supplementary Table 2). Combined allele frequencies and summary statistics for 15 X-chromosomal STR markers in 3 U.S. population groups from sample sets A and C.** AA: African American, CN: U.S. Caucasian, Hisp: U.S. Hispanic, N: number of alleles, H(exp): expected heterozygosity, H(obs): observed heterozygosity, PIC: polymorphism information content, PDf: power of discrimination in females, PDm: power of discrimination in males, MECI: mean exclusion chance in trios involving daughter, MECII: mean exclusion chance in father/daughter duos, p (HWE): p value of the exact test for Hardy-Weinberg equilibrium. Significant p values ($p < 0.05$) are shown in bold.

Table begins on next page.

| | DXS6795 | | | DXS9902 | | | DXS8378 | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | |
| N | 1043 | 1117 | 1013 | 1043 | 1117 | 1013 | 1043 | 1117 | 1013 | N |
| 5 | | | | | | | | | | 5 |
| 6 | 0.0010 | | | | | | | | | 6 |
| 7 | | | | 0.0010 | | | | | | 7 |
| 8 | 0.0010 | | | 0.0470 | | 0.0039 | 0.0048 | 0.0063 | 0.0010 | 8 |
| 8.3 | | | | | | | | | | 8.3 |
| 9 | 0.1189 | 0.2919 | 0.1491 | 0.0719 | 0.0439 | 0.0168 | 0.0115 | 0.0143 | 0.0138 | 9 |
| 9.3 | | | | | | | | | | 9.3 |
| 10 | 0.2857 | 0.0269 | 0.1076 | 0.2991 | 0.3241 | 0.3662 | 0.2723 | 0.3384 | 0.4215 | 10 |
| 10.1 | | | | | | 0.0020 | | | | 10.1 |
| 10.3 | | | | | | 0.0010 | | | | 10.3 |
| 11 | 0.1764 | 0.4673 | 0.2349 | 0.3461 | 0.3706 | 0.3978 | 0.3653 | 0.3518 | 0.3258 | 11 |
| 11.1 | | | | 0.0086 | 0.0286 | 0.0296 | | | | 11.1 |
| 11.3 | | | | | | | | | | 11.3 |
| 12 | 0.0853 | 0.0313 | 0.1520 | 0.2167 | 0.2184 | 0.1728 | 0.3087 | 0.2543 | 0.2132 | 12 |
| 12.1 | | | | | 0.0018 | 0.0039 | | | | 12.1 |
| 12.3 | | | | | | | | | | 12.3 |
| 13 | 0.0911 | 0.1791 | 0.3189 | 0.0086 | 0.0125 | 0.0039 | 0.0345 | 0.0322 | 0.0227 | 13 |
| 13.3 | | | | | | | | | | 13.3 |
| 14 | 0.0479 | 0.0009 | 0.0207 | 0.0010 | | 0.0020 | 0.0029 | 0.0027 | 0.0020 | 14 |
| 14.3 | | | | | | | | | | 14.3 |
| 15 | 0.1783 | 0.0027 | 0.0158 | | | | | | | 15 |
| 15.3 | | | | | | | | | | 15.3 |
| 16 | 0.0105 | | 0.0010 | | | | | | | 16 |
| 16.3 | | | | | | | | | | 16.3 |
| 17 | 0.0038 | | | | | | | | | 17 |
| 17.3 | | | | | | | | | | 17.3 |
| 18 | | | | | | | | | | 18 |
| 18.3 | | | | | | | | | | 18.3 |
| 19 | | | | | | | | | | 19 |
| 20 | | | | | | | | | | 20 |
| 20.3 | | | | | | | | | | 20.3 |
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| 31 | | | | | | | | | | 31 |
| 33 | | | | | | | | | | 33 |
| PIC | 0.8011 | 0.6048 | 0.7541 | 0.6913 | 0.6533 | 0.6159 | 0.6356 | 0.6358 | 0.6060 | PIC |
| H(exp) | 0.8233 | 0.6627 | 0.7855 | 0.7363 | 0.7070 | 0.6766 | 0.6958 | 0.6958 | 0.6700 | H(exp) |
| H(obs) | 0.8263 | 0.6337 | 0.8135 | 0.7316 | 0.6892 | 0.6838 | 0.6605 | 0.7084 | 0.6838 | H(obs) |
| PDf | 0.9465 | 0.8284 | 0.9226 | 0.8855 | 0.8605 | 0.8347 | 0.8473 | 0.8475 | 0.8271 | PDf |
| PDm | 0.8233 | 0.6627 | 0.7855 | 0.7363 | 0.7070 | 0.6766 | 0.6958 | 0.6958 | 0.6700 | PDm |
| MECI | 0.8011 | 0.6048 | 0.7541 | 0.6913 | 0.6533 | 0.6159 | 0.6356 | 0.6358 | 0.6060 | MECI |
| MECII | 0.6843 | 0.4580 | 0.6245 | 0.5514 | 0.5095 | 0.4704 | 0.4899 | 0.4903 | 0.4592 | MECII |
| p(HWE) | 0.8655 | 0.0984 | 0.2196 | 0.6772 | **0.0315** | 0.5959 | 0.7691 | 0.7213 | 0.7444 | p(HWE) |

Table continues on next page.

| | GATA172D05 | | | DXS7132 | | | DXS6803 | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | |
| N | 1043 | 1117 | 1013 | 1043 | 1118 | 1013 | 1043 | 1117 | 1013 | N |
| 5 | | | | | | | | | | 5 |
| 6 | 0.1946 | 0.1692 | 0.1165 | | | | | | | 6 |
| 7 | 0.0537 | 0.0009 | 0.0049 | | | | 0.0048 | | 0.0020 | 7 |
| 8 | 0.1764 | 0.1737 | 0.1412 | | | | 0.0134 | | | 8 |
| 8.3 | | | | | | | | | | 8.3 |
| 9 | 0.2848 | 0.0483 | 0.0632 | | | | 0.0288 | 0.0009 | 0.0020 | 9 |
| 9.3 | | | | | | | | 0.0009 | | 9.3 |
| 10 | 0.1438 | 0.2847 | 0.2932 | | | 0.0010 | 0.1246 | 0.0349 | 0.0197 | 10 |
| 10.1 | | | | | | | | | | 10.1 |
| 10.3 | | | | | | | | | 0.0010 | 10.3 |
| 11 | 0.1055 | 0.2095 | 0.2813 | 0.0163 | 0.0116 | 0.0030 | 0.3672 | 0.2498 | 0.2695 | 11 |
| 11.1 | | | | | | | | | | 11.1 |
| 11.3 | | | | | | | 0.0077 | 0.0143 | 0.0582 | 11.3 |
| 12 | 0.0403 | 0.1110 | 0.0977 | 0.1026 | 0.0868 | 0.0948 | 0.2368 | 0.2641 | 0.3268 | 12 |
| 12.1 | | | | | | | | | | 12.1 |
| 12.3 | | | | | | | 0.0757 | 0.1235 | 0.1530 | 12.3 |
| 13 | 0.0010 | 0.0027 | 0.0020 | 0.2579 | 0.2925 | 0.2586 | 0.0796 | 0.1441 | 0.0652 | 13 |
| 13.3 | | | | | | | 0.0316 | 0.1432 | 0.0977 | 13.3 |
| 14 | | | | 0.3490 | 0.3605 | 0.3406 | 0.0249 | 0.0063 | 0.0039 | 14 |
| 14.3 | | | | | | | 0.0019 | 0.0143 | 0.0010 | 14.3 |
| 15 | | | | 0.2224 | 0.1852 | 0.2409 | 0.0019 | | | 15 |
| 15.3 | | | | | | | | 0.0036 | | 15.3 |
| 16 | | | | 0.0364 | 0.0546 | 0.0444 | 0.0010 | | | 16 |
| 16.3 | | | | 0.0038 | | 0.0010 | | | | 16.3 |
| 17 | | | | 0.0086 | 0.0072 | 0.0118 | | | | 17 |
| 17.3 | | | | | | 0.0010 | | | | 17.3 |
| 18 | | | | 0.0029 | 0.0018 | 0.0030 | | | | 18 |
| 18.3 | | | | | | | | | | 18.3 |
| 19 | | | | | | | | | | 19 |
| 20 | | | | | | | | | | 20 |
| 20.3 | | | | | | | | | | 20.3 |
| 21 | | | | | | | | | | 21 |
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| 31 | | | | | | | | | | 31 |
| 33 | | | | | | | | | | 33 |
| PIC | 0.7884 | 0.7726 | 0.7572 | 0.7093 | 0.6971 | 0.7058 | 0.7597 | 0.7833 | 0.7483 | PIC |
| H(exp) | 0.8136 | 0.8016 | 0.7878 | 0.7500 | 0.7395 | 0.7480 | 0.7845 | 0.8096 | 0.7796 | H(exp) |
| H(obs) | 0.8500 | 0.7807 | 0.7676 | 0.8026 | 0.7735 | 0.7081 | 0.7974 | 0.7904 | 0.773 | H(obs) |
| PDf | 0.9401 | 0.9317 | 0.9243 | 0.8968 | 0.8898 | 0.8943 | 0.9287 | 0.9374 | 0.9202 | PDf |
| PDm | 0.8136 | 0.8016 | 0.7878 | 0.7500 | 0.7395 | 0.7480 | 0.7845 | 0.8096 | 0.7796 | PDm |
| MECI | 0.7884 | 0.7726 | 0.7572 | 0.7093 | 0.6971 | 0.7058 | 0.7597 | 0.7833 | 0.7483 | MECI |
| MECII | 0.6675 | 0.6470 | 0.6287 | 0.5718 | 0.5581 | 0.5677 | 0.6343 | 0.6612 | 0.6186 | MECII |
| p(HWE) | 0.2151 | 0.1429 | 0.8982 | 0.3650 | 0.6347 | 0.2813 | 0.9855 | 0.6405 | 0.3303 | p(HWE) |

Table continues on next page.

| | DXS6789 | | | GATA165B12 | | | DXS7130 | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | |
| N | 1043 | 1117 | 1013 | 1043 | 1117 | 1013 | 1043 | 1117 | 1013 | N |
| 5 | | | | | | | | | | 5 |
| 6 | | | | | | | | | | 6 |
| 7 | | | | 0.0019 | | | | | | 7 |
| 8 | | | | 0.0393 | 0.0072 | 0.0099 | | | | 8 |
| 8.3 | | | | | | | | | | 8.3 |
| 9 | | | | 0.1371 | 0.3071 | 0.2438 | 0.0058 | 0.0009 | | 9 |
| 9.3 | | | | | | | | | | 9.3 |
| 10 | | | | 0.3289 | 0.3321 | 0.4709 | 0.0249 | 0.0027 | 0.0039 | 10 |
| 10.1 | | | | | | | | | | 10.1 |
| 10.3 | | | | | | | | | | 10.3 |
| 11 | | | | 0.3873 | 0.3232 | 0.2488 | 0.0719 | 0.0394 | 0.0582 | 11 |
| 11.1 | | | | | | | | | | 11.1 |
| 11.3 | | | | | | | | | | 11.3 |
| 12 | | | | 0.0988 | 0.0304 | 0.0267 | 0.2042 | 0.1047 | 0.2024 | 12 |
| 12.1 | | | | | | | | | | 12.1 |
| 12.3 | | | | | | | | | | 12.3 |
| 13 | | | | 0.0067 | | | 0.1582 | 0.0439 | 0.0632 | 13 |
| 13.3 | | | | | | | 0.0221 | 0.0564 | 0.0227 | 13.3 |
| 14 | 0.0048 | 0.0036 | 0.0079 | | | | 0.0422 | 0.0098 | 0.0188 | 14 |
| 14.3 | | | | | | | 0.1755 | 0.2122 | 0.1885 | 14.3 |
| 15 | 0.2272 | 0.0421 | 0.0434 | | | | 0.0019 | | 0.0020 | 15 |
| 15.3 | | | | | | | 0.2253 | 0.3554 | 0.3475 | 15.3 |
| 16 | 0.1151 | 0.0161 | 0.0434 | | | | 0.0010 | | | 16 |
| 16.3 | | | | | | | 0.0585 | 0.1477 | 0.0711 | 16.3 |
| 17 | 0.0077 | 0.0027 | 0.0049 | | | | 0.0010 | | | 17 |
| 17.3 | | | | | | | 0.0067 | 0.0260 | 0.0207 | 17.3 |
| 18 | 0.0153 | 0.0009 | 0.0030 | | | | | | | 18 |
| 18.3 | | | | | | | 0.0010 | | 0.0010 | 18.3 |
| 19 | 0.0604 | 0.0242 | 0.0346 | | | | | | | 19 |
| 20 | 0.2013 | 0.3787 | 0.3998 | | | | | | | 20 |
| 20.3 | | | | | | | | 0.0009 | | 20.3 |
| 21 | 0.2119 | 0.2892 | 0.2912 | | | | | | | 21 |
| 22 | 0.1035 | 0.1459 | 0.1135 | | | | | | | 22 |
| 23 | 0.0460 | 0.0806 | 0.0464 | | | | | | | 23 |
| 24 | 0.0058 | 0.0161 | 0.0109 | | | | | | | 24 |
| 25 | 0.0010 | | 0.0010 | | | | | | | 25 |
| 26 | | | | | | | | | | 26 |
| 27 | | | | | | | | | | 27 |
| 28 | | | | | | | | | | 28 |
| 29 | | | | | | | | | | 29 |
| 30 | | | | | | | | | | 30 |
| 31 | | | | | | | | | | 31 |
| 33 | | | | | | | | | | 33 |
| PIC | 0.8116 | 0.7040 | 0.6979 | 0.6632 | 0.6258 | 0.5944 | 0.8206 | 0.7623 | 0.7620 | PIC |
| H(exp) | 0.8329 | 0.7423 | 0.7351 | 0.7117 | 0.6900 | 0.6561 | 0.8402 | 0.7884 | 0.7890 | H(exp) |
| H(obs) | 0.8421 | 0.7133 | 0.6973 | 0.7158 | 0.7012 | 0.6784 | 0.8158 | 0.7807 | 0.7568 | H(obs) |
| PDf | 0.9508 | 0.8953 | 0.8926 | 0.8684 | 0.8397 | 0.8200 | 0.9549 | 0.9291 | 0.9285 | PDf |
| PDm | 0.8329 | 0.7423 | 0.7351 | 0.7117 | 0.6900 | 0.6561 | 0.8402 | 0.7884 | 0.7890 | PDm |
| MECI | 0.8116 | 0.7040 | 0.6979 | 0.6632 | 0.6258 | 0.5944 | 0.8206 | 0.7623 | 0.7620 | MECI |
| MECII | 0.6981 | 0.5669 | 0.5607 | 0.5206 | 0.4793 | 0.4466 | 0.7104 | 0.6360 | 0.6358 | MECII |
| p(HWE) | 0.1885 | 0.0993 | 0.4868 | 0.8493 | 0.3855 | 0.9952 | 0.5784 | 0.3077 | **0.0463** | p(HWE) |

Table continues on next page.

| | DXS101 | | | DXS7424 | | | HPRTB | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | |
| N | 1043 | 1117 | 1013 | 1043 | 1117 | 1013 | 1043 | 1117 | 1013 | N |
| 5 | | | | | | | | | | 5 |
| 6 | | | | | | | | | | 6 |
| 7 | | | | | | | | 0.0009 | | 7 |
| 8 | | | | | | | 0.0010 | 0.0009 | | 8 |
| 8.3 | | | | | | | | | | 8.3 |
| 9 | | | | 0.0019 | 0.0009 | | 0.0384 | 0.0036 | 0.0010 | 9 |
| 9.3 | | | | | | | | | | 9.3 |
| 10 | | | | 0.0096 | 0.0045 | 0.0059 | 0.0144 | 0.0045 | 0.0049 | 10 |
| 10.1 | | | | | | | | | | 10.1 |
| 10.3 | | | | | | | | | | 10.3 |
| 11 | | | | 0.0700 | 0.0054 | 0.0039 | 0.0940 | 0.1244 | 0.0809 | 11 |
| 11.1 | | | | | | | | | | 11.1 |
| 11.3 | | | | | | | | | | 11.3 |
| 12 | | | | 0.0642 | 0.0421 | 0.0365 | 0.2838 | 0.3321 | 0.2725 | 12 |
| 12.1 | | | | | | | | | | 12.1 |
| 12.3 | | | | | | | | | | 12.3 |
| 13 | | | | 0.2205 | 0.0609 | 0.1145 | 0.2733 | 0.3214 | 0.3830 | 13 |
| 13.3 | | | | | | | | | | 13.3 |
| 14 | 0.0010 | | | 0.2483 | 0.2077 | 0.2004 | 0.1975 | 0.1477 | 0.1895 | 14 |
| 14.3 | | | | | | | | | | 14.3 |
| 15 | 0.0019 | 0.0304 | 0.0138 | 0.1812 | 0.2578 | 0.2093 | 0.0825 | 0.0528 | 0.0513 | 15 |
| 15.3 | | | | | | | | | | 15.3 |
| 16 | 0.0029 | 0.0018 | 0.0010 | 0.1457 | 0.2614 | 0.2715 | 0.0153 | 0.0107 | 0.0168 | 16 |
| 16.3 | | | | | | | | | | 16.3 |
| 17 | 0.0038 | 0.0045 | | 0.0431 | 0.1289 | 0.1115 | | 0.0009 | | 17 |
| 17.3 | | | | | | | | | | 17.3 |
| 18 | 0.0479 | 0.0743 | 0.0652 | 0.0125 | 0.0242 | 0.0415 | | | | 18 |
| 18.3 | | | | | | | | | | 18.3 |
| 19 | 0.0872 | 0.0528 | 0.0385 | 0.0019 | 0.0054 | 0.0039 | | | | 19 |
| 20 | 0.0652 | 0.0197 | 0.0188 | 0.0010 | 0.0009 | 0.0010 | | | | 20 |
| 20.3 | | | | | | | | | | 20.3 |
| 21 | 0.1419 | 0.0278 | 0.0286 | | | | | | | 21 |
| 22 | 0.0652 | 0.0143 | 0.0217 | | | | | | | 22 |
| 23 | 0.0709 | 0.0671 | 0.0642 | | | | | | | 23 |
| 24 | 0.0978 | 0.1996 | 0.2438 | | | | | | | 24 |
| 25 | 0.0901 | 0.1817 | 0.1728 | | | | | | | 25 |
| 26 | 0.1007 | 0.1334 | 0.1984 | | | | | | | 26 |
| 27 | 0.1304 | 0.0877 | 0.0790 | | | | | | | 27 |
| 28 | 0.0518 | 0.0591 | 0.0365 | | | | | | | 28 |
| 29 | 0.0240 | 0.0269 | 0.0099 | | | | | | | 29 |
| 30 | 0.0115 | 0.0152 | 0.0049 | | | | | | | 30 |
| 31 | 0.0038 | 0.0036 | 0.0020 | | | | | | | 31 |
| 33 | 0.0019 | | 0.0010 | | | | | | | 33 |
| PIC | 0.9008 | 0.8713 | 0.8361 | 0.8015 | 0.7703 | 0.7882 | 0.7571 | 0.7053 | 0.6911 | PIC |
| H(exp) | 0.9082 | 0.8821 | 0.8520 | 0.8245 | 0.7993 | 0.8137 | 0.7882 | 0.7462 | 0.7337 | H(exp) |
| H(obs) | 0.9263 | 0.8506 | 0.8595 | 0.8500 | 0.8361 | 0.8000 | 0.7842 | 0.7976 | 0.7216 | H(obs) |
| PDf | 0.9842 | 0.9753 | 0.9622 | 0.9462 | 0.9307 | 0.9399 | 0.9240 | 0.8947 | 0.8865 | PDf |
| PDm | 0.9082 | 0.8821 | 0.8520 | 0.8245 | 0.7993 | 0.8137 | 0.7882 | 0.7462 | 0.7337 | PDm |
| MECI | 0.9008 | 0.8713 | 0.8361 | 0.8015 | 0.7703 | 0.7882 | 0.7571 | 0.7053 | 0.6911 | MECI |
| MECII | 0.8259 | 0.7823 | 0.7326 | 0.6848 | 0.6450 | 0.6676 | 0.6288 | 0.5675 | 0.5512 | MECII |
| p(HWE) | 0.3703 | **0.0498** | 0.9489 | 0.6025 | 0.1476 | **0.0377** | 0.6834 | **0.0403** | 0.7875 | p(HWE) |

Table continues on next page.

| | DXS7423 | | | DXS10147 | | | GATA31E08 | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** | |
| N | 1043 | 1117 | 1013 | 1043 | 1117 | 1013 | 1043 | 1117 | 1013 | N |
| 5 | | | | 0.0010 | | | | | | 5 |
| 6 | | | | 0.1342 | 0.2310 | 0.344 | 0.0010 | | | 6 |
| 7 | | | | 0.2800 | 0.0403 | 0.060 | 0.0211 | | 0.0049 | 7 |
| 8 | 0.0077 | | | 0.3624 | 0.2972 | 0.414 | 0.0268 | 0.0018 | 0.0049 | 8 |
| 8.3 | | | | | | | | | | 8.3 |
| 9 | | | | 0.1774 | 0.4136 | 0.171 | 0.1515 | 0.1808 | 0.1362 | 9 |
| 9.3 | | | | | | | | | | 9.3 |
| 10 | | | | 0.0403 | 0.0161 | 0.010 | 0.1534 | 0.0251 | 0.0523 | 10 |
| 10.1 | | | | | | | | | | 10.1 |
| 10.3 | | | | | | | | | | 10.3 |
| 11 | | 0.0009 | | 0.0038 | 0.0018 | 0.002 | 0.0623 | 0.2095 | 0.1807 | 11 |
| 11.1 | | | | | | | | | | 11.1 |
| 11.3 | | | | | | | | | | 11.3 |
| 12 | 0.0048 | | 0.0010 | | | | 0.2560 | 0.2095 | 0.3583 | 12 |
| 12.1 | | | | | | | | | | 12.1 |
| 12.3 | | | | | | | | | | 12.3 |
| 13 | 0.0968 | 0.1003 | 0.0316 | 0.0010 | | | 0.2378 | 0.2551 | 0.2024 | 13 |
| 13.3 | | | | | | | | | | 13.3 |
| 14 | 0.4660 | 0.3187 | 0.3001 | | | | 0.0757 | 0.1030 | 0.0474 | 14 |
| 14.3 | | | | | | | | | | 14.3 |
| 15 | 0.3241 | 0.3930 | 0.4817 | | | | 0.0134 | 0.0134 | 0.0128 | 15 |
| 15.3 | | | | | | | | | | 15.3 |
| 16 | 0.0872 | 0.1513 | 0.0977 | | | | 0.0010 | 0.0009 | | 16 |
| 16.3 | | | | | | | | | | 16.3 |
| 17 | 0.0134 | 0.0349 | 0.0879 | | | | | 0.0009 | | 17 |
| 17.3 | | | | | | | | | | 17.3 |
| 18 | | 0.0009 | | | | | | | | 18 |
| 18.3 | | | | | | | | | | 18.3 |
| 19 | | | | | | | | | | 19 |
| 20 | | | | | | | | | | 20 |
| 20.3 | | | | | | | | | | 20.3 |
| 21 | | | | | | | | | | 21 |
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| 31 | | | | | | | | | | 31 |
| 33 | | | | | | | | | | 33 |
| PIC | 0.6037 | 0.6604 | 0.6059 | 0.6958 | 0.6263 | 0.6184 | 0.7969 | 0.7735 | 0.7429 | PIC |
| H(exp) | 0.6606 | 0.7098 | 0.6596 | 0.7391 | 0.6854 | 0.6780 | 0.8205 | 0.8030 | 0.7743 | H(exp) |
| H(obs) | 0.7053 | 0.6867 | 0.6892 | 0.7474 | 0.6578 | 0.6919 | 0.8263 | 0.8241 | 0.7703 | H(obs) |
| PDf | 0.8279 | 0.8664 | 0.8304 | 0.8886 | 0.8419 | 0.8367 | 0.9442 | 0.9317 | 0.9177 | PDf |
| PDm | 0.6606 | 0.7098 | 0.6596 | 0.7391 | 0.6854 | 0.6780 | 0.8205 | 0.8030 | 0.7743 | PDm |
| MECI | 0.6037 | 0.6604 | 0.6059 | 0.6958 | 0.6263 | 0.6184 | 0.7969 | 0.7735 | 0.7429 | MECI |
| MECII | 0.4579 | 0.5172 | 0.4597 | 0.5559 | 0.4801 | 0.4726 | 0.6790 | 0.6481 | 0.6115 | MECII |
| p(HWE) | 0.9073 | 0.0936 | 0.8123 | 0.1265 | 0.3951 | 0.3083 | 0.3373 | 0.5504 | 0.8781 | p(HWE) |

**Table 4.17. (Supplementary Table 3). P values from multi-locus exact test for linkage disequilibrium results for original and combined populations.** P values for marker pairs with significant p values ($p < 0.05$) in at least one of the populations are shown. Bold values are those that remain significant after the Bonferroni correction ($p < 0.0011$). The combined population represents sample sets A and C together.

| Locus combination | Sample set C | Combined population |
|---|---|---|
| DXS6795-DXS9902 | 0.0510 | 0.0030 |
| DXS6795-DXS8378 | 0.0510 | 0.0160 |
| DXS9902-GATA172D05 | 0.0030 | **0.0000** |
| DXS8378-GATA172D05 | 0.0510 | 0.0140 |
| GATA172D05-DXS6803 | 0.0510 | 0.0070 |
| DXS7132-DXS6789 | 0.0330 | 0.0510 |
| DXS6803-DXS6789 | 0.0510 | **0.0000** |
| DXS6803-GATA165B12 | 0.0510 | **0.0000** |
| DXS6789-GATA165B12 | **0.0000** | **0.0000** |
| DXS6789-DXS7130 | 0.0060 | **0.0000** |
| GATA165B12-DXS7130 | 0.0510 | 0.0120 |
| GATA165B12-DXS101 | 0.0060 | **0.0000** |
| DXS7130-DXS101 | 0.0050 | **0.0000** |
| DXS7130-DXS7424 | 0.0510 | 0.0420 |
| DXS101-DXS7424 | 0.0020 | **0.0000** |
| DXS7424-HPRTB | 0.0510 | **0.0010** |
| DXS7424-DXS7423 | 0.0510 | **0.0010** |
| HPRTB-DXS7423 | 0.0130 | 0.0090 |
| HPRTB-DXS10147 | **0.0010** | 0.0070 |
| DXS7423-DXS10147 | **0.0000** | **0.0000** |
| DXS10147-GATA31E08 | **0.0000** | **0.0000** |

Pairwise $F_{ST}$ value comparisons for populations from sample set C revealed that all populations were significantly different from one another (indicated by a corresponding p value < 0.05) at 13 of the 15 markers (Table 4.18). The U.S. Caucasian and U.S. Hispanic populations did not differ significantly at marker DXS6789, however, and all three populations were similar to one another at marker DXS7132. Given these results, the three populations were determined to be disctinct with respect to the multiplexes applied here, and were maintained separately for all analyses mentioned within the publication.

**Table 4.18. Pairwise $F_{ST}$ values (below diagonal) and corresponding p values with standard deviations (above diagonal) comparing populations from sample set C by marker.** P values indicative of populations that are not significantly different from one another ($p > 0.5$) are bolded. AA: African American; CN: U.S. Caucasian, Hisp: U.S. Hispanic.

| Marker | Population | AA | CN | Hisp |
|---|---|---|---|---|
| DXS101 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.02692 | * | 0.00822±0.0009 |
| | Hisp | 0.03828 | 0.00266 | * |
| DXS6789 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.05889 | * | **0.06405±0.0024** |
| | Hisp | 0.06051 | 0.00187 | * |
| DXS6795 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.14077 | * | 0.00000±0.0000 |
| | Hisp | 0.08007 | 0.07417 | * |
| DXS6803 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.02543 | * | 0.00000±0.0000 |
| | Hisp | 0.02143 | 0.00889 | * |
| DXS7130 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.03579 | * | 0.00010±0.0001 |
| | Hisp | 0.0175 | 0.00834 | * |
| DXS7132 | AA | * | **0.28740±0.0053** | **0.76349±0.0043** |
| | CN | 0.00034 | * | **0.20325±0.0042** |
| | Hisp | -0.00095 | 0.00071 | * |
| DXS7423 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.02218 | * | 0.00000±0.0000 |
| | Hisp | 0.04198 | 0.01248 | * |
| DXS7424 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.04349 | * | 0.00713±0.0009 |
| | Hisp | 0.02987 | 0.00347 | * |
| DXS8378 | AA | * | 0.02198±0.0016 | 0.00000±0.0000 |
| | CN | 0.00419 | * | 0.00010±0.0001 |
| | Hisp | 0.03518 | 0.01241 | * |
| DXS9902 | AA | * | 0.04762±0.0022 | 0.00000±0.0000 |
| | CN | 0.00271 | * | 0.00218±0.0005 |
| | Hisp | 0.01415 | 0.00696 | * |
| DXS10147 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.0801 | * | 0.00000±0.0000 |
| | Hisp | 0.05958 | 0.04885 | * |
| GATA31E08 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.02413 | * | 0.00000±0.0000 |
| | Hisp | 0.02734 | 0.02225 | * |
| GATA165B12 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.03689 | * | 0.00000±0.0000 |
| | Hisp | 0.05484 | 0.0195 | * |
| GATA172D05 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.06217 | * | 0.00149±0.0004 |
| | Hisp | 0.07581 | 0.00565 | * |
| HPRTB | AA | * | 0.00020±0.0001 | 0.00000±0.0000 |
| | CN | 0.01001 | * | 0.00703±0.0009 |
| | Hisp | 0.0105 | 0.00453 | * |

In addition to using the chi square test to compare the populations in sample set C to similar published populations, pairwise $F_{ST}$ and corresponding p values were calculated (Table 4.19). Using the $F_{ST}$ value may in fact be a better way in which to

analyze and compare populations using allele frequency distributions that often include expected allele counts less than 1 and/or greater than 20% of these counts are less than 5, which tends to make the chi square test less reliable [298].

In general, equivalent populations were found to be similar to one another at overlapping markers with the exception of the U.S. Hispanic populations at markers DXS7423, DXS8378, and GATA172D05. For two of these markers (DXS7423 and DXS8378), the U.S. Hispanic population from Gomez, *et al.* [70] was more similar to the African American or U.S. Caucasian populations from sample set C. Other unexpected associations involving the U.S. Hispanic populations were observed, underscoring the variability of self-described U.S. Hispanic populations.

**Table 4.19. P values (with standard deviations) corresponding to pairwise $F_{ST}$ values (not shown) comparing sample set C populations (top) to published populations (left) at overlapping markers.** P values indicative of populations that are not significantly different from one another ($p > 0.5$) are bolded. Values at the intersection of equivalent populations are shaded in gray. AA: African American; CN: U.S. Caucasian, Hisp: U.S. Hispanic.

| Marker | Ref. | Population | AA | CN | Hisp |
|---|---|---|---|---|---|
| DXS101 | [70] | AA | **0.13523±0.0036** | 0.00000±0.0000 | 0.00000±0.0000 |
| | | Hisp | 0.00000±0.0000 | **0.56509±0.0043** | **0.18701±0.0033** |
| | [283] | AA | **0.12048±0.0033** | 0.00000±0.0000 | 0.00000±0.0000 |
| | | CN | 0.00000±0.0000 | **0.09197±0.0027** | **0.25047±0.0042** |
| DXS6789 | [70] | AA | **0.38303±0.0049** | 0.00000±0.0000 | 0.00000±0.0000 |
| | | Hisp | 0.00000±0.0000 | **0.06118±0.0024** | **0.05702±0.0022** |
| DXS7132 | [70] | AA | **0.57321±0.0050** | **0.87556±0.0036** | **0.44451±0.0053** |
| | | Hisp | **0.13276±0.0031** | **0.06178±0.0025** | **0.16276±0.0038** |
| DXS7423 | [70] | AA | **0.12979±0.0032** | 0.00000±0.0000 | 0.00000±0.0000 |
| | | Hisp | **0.11019±0.0031** | 0.03435±0.0020 | 0.01406±0.0010 |
| DXS8378 | [70] | AA | **0.45728±0.0039** | 0.01921±0.0012 | 0.00000±0.0000 |
| | | Hisp | **0.07940±0.0026** | **0.27265±0.0048** | 0.01406±0.0011 |
| GATA172D05 | [70] | AA | **0.40263±0.0043** | 0.00000±0.0000 | 0.00000±0.0000 |
| | | Hisp | 0.00010±0.0001 | 0.00673±0.0008 | 0.00050±0.0002 |
| HPRTB | [70] | AA | **0.73240±0.0044** | 0.01436±0.0011 | 0.00653±0.0008 |
| | | Hisp | **0.45322±0.0042** | **0.24275±0.0046** | **0.49203±0.0048** |
| | [292] | AA | **0.43550±0.0051** | 0.00050±0.0002 | 0.02812±0.0018 |
| | | CN | 0.00139±0.0004 | **0.68498±0.0048** | 0.00347±0.0006 |
| | | Hisp | 0.00990±0.0011 | 0.04594±0.0019 | **0.75171±0.0040** |

*4.1.4. Additional markers*

As part of a larger concordance study (see Chapter 6), 853 U.S. population samples were typed using the Investigator Argus X-12 commercial kit. The population was a combination of sample sets A (152 U.S. Asian) and B (260 African American, 3 U.S. Asian, 298 U.S. Caucasian, 140 U.S. Hispanic). U.S. allele frequencies and population genetic data were gained for 8 X STR markers (DXS10103, DXS10134, DXS10074, DXS10101, DXS10135, DXS10146, DXS10079, DXS10148) in addition to the 15 markers previously typed using the two multiplexes developed during this study and described in Sections 4.1.1 and 4.1.2.

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# Allele frequency distribution of twelve X-chromosomal short tandem repeat markers in four U.S. population groups

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ABSTRACT

A total of 853 samples from the four major U.S. population groups (African American, Asian, Caucasian, and Hispanic) were typed using the Qiagen® Investigator Argus X-12 kit. Allele frequency distributions are reported here for each of the 12 X-STR markers (DXS10103, DXS8378, DXS7132, DXS10134, DXS10074, DXS10101, DXS10135, DXS7423, DXS10146, DXS10079, DXS10148, and HPRTB).

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## 1. Introduction

Though a large number of multiplex assays using X-chromosomal short tandem repeat (X-STR) markers have been described in the literature, there is currently only one commercial kit available, the Qiagen® Investigator Argus X-12 kit, which simultaneously detects 12 X-STR markers plus Amelogenin. Several recent publications have described its performance on high quality samples and presented allele and haplotype frequency data from populations in Germany [1], Morocco [2], China [3], Hungary [4], and Sweden [5]. Further study of these 12 markers is required before the potential of the kit can be fully realized for use in the forensic setting. To this end, 853 samples from the four major U.S. population groups (African American, Asian, Caucasian, and Hispanic) were typed using this commercial kit.

## 2. Materials and methods

Donors were 701 individuals from four self-identified U.S. population groups, African American (260 males), U.S. Asian (3 males), U.S. Caucasian (260 males, 38 females), and U.S. Hispanic (140 males) whose blood samples were collected and extracted as described in [6]. A supplementary U.S. Asian population (69 males, 83 females), described in [7], was also included in the study. Amplification and data analysis were performed according to the manufacturer's instructions [8] using a GeneAmp® thermal cycler, 3130*xl* Genetic Analyzer, and Genemapper ID-X version 1.1 (all Applied Biosystems, Foster City, CA).

## 3. Results and discussion

Allele frequencies for the four U.S. population groups are shown in Table 1. A comparison of the allele frequency distributions across overlapping markers and population groups using the chi-square test revealed no significant differences ($p < 0.05$; data not shown) from published studies [7,9,10]. This study represents a substantial increase in the number of U.S. population samples with X-STR allele frequencies currently available as well as the first report of Investigator Argus X-12 allele frequencies in U.S. populations.

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No external funding was received to conduct this study.

## Conflict of interest

No conflict of interest exists.

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**Table 1**
X-12 marker allele frequencies for four U.S. populations.

| | AF | AS | CN | Hisp |
|---|---|---|---|---|
| **DXS10135** | | | | |
| **N** | 261 | 238 | 337 | 140 |
| **15** | 0.011 | | 0.003 | 0.021 |
| **16** | 0.023 | | 0.009 | 0.007 |
| **17** | 0.031 | 0.017 | 0.030 | 0.029 |
| **17.1** | 0.019 | | | 0.007 |
| **18** | 0.065 | 0.025 | 0.039 | 0.014 |
| **18.1** | 0.019 | | 0.009 | 0.007 |
| **19** | 0.096 | 0.088 | 0.062 | 0.079 |
| **19.1** | 0.073 | | 0.012 | 0.021 |
| **20** | 0.050 | 0.097 | 0.050 | 0.071 |
| **20.1** | 0.034 | | 0.012 | |
| **21** | 0.069 | 0.122 | 0.059 | 0.086 |
| **21.1** | 0.057 | | 0.006 | 0.021 |
| **22** | 0.073 | 0.134 | 0.068 | 0.107 |
| **22.1** | 0.011 | | 0.021 | |
| **23** | 0.046 | 0.092 | 0.089 | 0.114 |
| **23.1** | 0.011 | | | |
| **24** | 0.054 | 0.109 | 0.098 | 0.071 |
| **24.1** | 0.008 | | 0.003 | 0.014 |
| **25** | 0.038 | 0.050 | 0.107 | 0.071 |
| **25.1** | 0.004 | | 0.003 | |
| **26** | 0.042 | 0.021 | 0.086 | 0.071 |
| **27** | 0.031 | 0.067 | 0.059 | 0.100 |
| **27.1** | 0.008 | | 0.003 | |
| **28** | 0.015 | 0.055 | 0.047 | 0.043 |
| **28.1** | 0.004 | | | |
| **29** | 0.027 | 0.042 | 0.036 | 0.021 |
| **30** | 0.015 | 0.025 | 0.042 | 0.014 |
| **31** | 0.019 | 0.021 | 0.024 | |
| **32** | | 0.021 | 0.009 | |
| **33** | | 0.008 | 0.009 | |
| **34** | | | 0.003 | 0.007 |
| **34.1** | 0.004 | | | |
| **35** | 0.004 | 0.004 | | |
| **35.1** | 0.004 | | | |
| **35.2** | 0.011 | | | |
| **36.1** | 0.004 | | | |
| **38** | | | 0.003 | |
| **38.2** | 0.008 | | | |
| **39.2** | 0.011 | | | |

| | AF | AS | CN | Hisp |
|---|---|---|---|---|
| **DXS10101** | | | | |
| **N** | 261 | 238 | 336 | 141 |
| **23** | 0.004 | | | |
| **24.2** | 0.038 | | | 0.035 |
| **25** | 0.004 | | | |
| **25.2** | | | 0.009 | |
| **26** | 0.015 | | | |
| **26.2** | 0.019 | | 0.015 | |
| **27** | 0.008 | 0.004 | 0.003 | 0.007 |
| **27.2** | 0.027 | 0.008 | 0.051 | 0.050 |
| **28** | 0.073 | 0.034 | 0.018 | 0.028 |
| **28.2** | 0.031 | 0.017 | 0.095 | 0.014 |
| **29** | 0.096 | 0.034 | 0.030 | 0.028 |
| **29.2** | 0.038 | 0.084 | 0.119 | 0.057 |
| **30** | 0.092 | 0.122 | 0.071 | 0.057 |
| **30.2** | 0.073 | 0.084 | 0.158 | 0.156 |
| **31** | 0.134 | 0.164 | 0.137 | 0.113 |
| **31.2** | 0.038 | 0.092 | 0.122 | 0.121 |
| **32** | 0.138 | 0.130 | 0.074 | 0.156 |
| **32.2** | 0.015 | 0.097 | 0.024 | 0.050 |
| **33** | 0.130 | 0.084 | 0.045 | 0.071 |
| **33.2** | 0.004 | 0.017 | 0.009 | 0.007 |
| **34** | 0.023 | 0.025 | 0.018 | 0.043 |
| **35** | | 0.004 | | 0.007 |
| **37** | | | 0.003 | |

| | AF | AS | CN | Hisp |
|---|---|---|---|---|
| **DXS10148** | | | | |
| **N** | 261 | 238 | 336 | 140 |
| **13.3** | 0.103 | | | |
| **14** | | | 0.003 | |
| **16** | 0.004 | | | |
| **17** | | 0.004 | 0.003 | |
| **18** | 0.123 | 0.143 | 0.134 | 0.157 |
| **19** | 0.050 | 0.059 | 0.015 | 0.007 |
| **20** | 0.008 | 0.025 | | 0.014 |
| **20.1** | 0.011 | | | |
| **21** | | 0.008 | | |
| **21.1** | | 0.008 | 0.006 | 0.007 |
| **22** | 0.011 | | | 0.014 |
| **22.1** | 0.008 | 0.042 | 0.015 | 0.007 |
| **23** | 0.011 | | 0.057 | 0.050 |
| **23.1** | 0.038 | 0.092 | 0.071 | 0.036 |
| **24** | 0.034 | | 0.012 | 0.014 |
| **24.1** | 0.034 | 0.155 | 0.158 | 0.064 |
| **25** | 0.019 | | | |
| **25.1** | 0.054 | 0.134 | 0.161 | 0.229 |
| **26** | 0.015 | | | |
| **26.1** | 0.034 | 0.097 | 0.185 | 0.200 |
| **26.3** | 0.004 | | | |
| **27** | 0.008 | | | |
| **27.1** | 0.031 | 0.105 | 0.098 | 0.093 |
| **27.2** | 0.008 | 0.004 | | |
| **28** | 0.023 | | | |
| **28.1** | 0.023 | 0.050 | 0.045 | 0.036 |
| **29** | 0.019 | | | |
| **29.1** | 0.027 | 0.038 | 0.033 | 0.036 |
| **29.2** | | 0.004 | | |
| **30** | 0.023 | | | |
| **30.1** | 0.011 | 0.017 | 0.006 | 0.014 |
| **31** | 0.019 | | | |
| **31.1** | | 0.008 | | 0.007 |
| **32** | 0.008 | | | |
| **32.2** | | 0.004 | | |
| **35.1** | 0.004 | | | |
| **null** | 0.234 | | | 0.014 |

| | AF | AS | CN | Hisp |
|---|---|---|---|---|
| **DXS10074** | | | | |
| **N** | 261 | 238 | 336 | 140 |
| **7** | 0.115 | | 0.083 | 0.036 |
| **8** | 0.015 | | 0.146 | 0.057 |
| **9** | 0.004 | | 0.009 | 0.036 |
| **10** | 0.019 | | | |
| **11** | 0.034 | | | 0.021 |
| **11.3** | | 0.004 | | |
| **12** | 0.077 | | | 0.021 |
| **13** | 0.096 | 0.004 | 0.009 | 0.007 |
| **14** | 0.111 | 0.008 | 0.015 | 0.057 |
| **14.2** | | | | 0.007 |
| **14.3** | 0.008 | | | |
| **15** | 0.180 | 0.076 | 0.077 | 0.171 |
| **15.3** | 0.004 | 0.004 | | |
| **16** | 0.153 | 0.143 | 0.196 | 0.150 |
| **16.3** | 0.011 | | | |
| **17** | 0.111 | 0.336 | 0.199 | 0.243 |
| **18** | 0.034 | 0.244 | 0.167 | 0.107 |
| **19** | 0.023 | 0.160 | 0.086 | 0.071 |
| **19.2** | | | 0.003 | |
| **20** | | 0.017 | 0.006 | 0.014 |
| **21** | 0.004 | | 0.003 | |
| **22** | | 0.004 | | |

| | AF | AS | CN | Hisp |
|---|---|---|---|---|
| **DXS10146** | | | | |
| **N** | 261 | 238 | 336 | 140 |
| **23** | 0.004 | | | 0.014 |
| **24** | 0.004 | 0.034 | 0.006 | 0.007 |
| **25** | 0.019 | 0.055 | 0.060 | 0.036 |
| **26** | 0.065 | 0.092 | 0.134 | 0.079 |
| **27** | 0.080 | 0.218 | 0.128 | 0.164 |
| **28** | 0.080 | 0.218 | 0.164 | 0.121 |
| **29** | 0.069 | 0.160 | 0.140 | 0.250 |
| **29.2** | 0.008 | | | |
| **30** | 0.073 | 0.084 | 0.119 | 0.064 |
| **30.2** | 0.004 | | | |
| **31** | 0.069 | 0.063 | 0.039 | 0.050 |
| **31.2** | 0.004 | | | |
| **32** | 0.073 | 0.029 | 0.006 | 0.014 |
| **32.2** | 0.008 | | | |
| **33** | 0.069 | 0.013 | 0.003 | |
| **33.2** | 0.011 | | | |
| **34** | 0.011 | 0.008 | | |
| **34.2** | 0.054 | | | |
| **35** | 0.008 | | | |
| **35.2** | 0.042 | | | 0.014 |
| **36.2** | 0.011 | | | 0.007 |
| **38.2** | 0.038 | | 0.006 | 0.021 |
| **39.1** | 0.008 | | | |
| **39.2** | 0.011 | | 0.039 | 0.029 |
| **40.2** | 0.034 | 0.004 | 0.030 | 0.029 |
| **41** | | | 0.003 | |
| **41.2** | 0.011 | 0.004 | 0.012 | 0.014 |
| **42.2** | 0.027 | 0.013 | 0.012 | 0.007 |
| **42.3** | 0.004 | | | |
| **43.2** | 0.015 | | 0.048 | 0.007 |
| **44.2** | 0.008 | | 0.033 | 0.036 |
| **45.2** | 0.004 | | 0.009 | 0.007 |
| **46.2** | | 0.004 | 0.012 | |
| **47.2** | | | | 0.007 |
| **null** | 0.073 | | | 0.021 |

| | AF | AS | CN | Hisp |
|---|---|---|---|---|
| **DXS10079** | | | | |
| **N** | 262 | 238 | 337 | 140 |
| **13** | 0.015 | | 0.003 | 0.007 |
| **14** | 0.008 | | 0.006 | 0.064 |
| **15** | | 0.004 | 0.024 | 0.007 |
| **16** | 0.008 | 0.025 | 0.021 | 0.021 |
| **17** | 0.084 | 0.055 | 0.065 | 0.050 |
| **18** | 0.126 | 0.105 | 0.166 | 0.107 |
| **19** | 0.221 | 0.223 | 0.217 | 0.250 |
| **20** | 0.260 | 0.261 | 0.300 | 0.271 |
| **21** | 0.168 | 0.139 | 0.128 | 0.157 |
| **22** | 0.084 | 0.164 | 0.065 | 0.043 |
| **23** | 0.019 | 0.025 | 0.006 | 0.021 |
| **24** | 0.008 | | | |

| | AF | AS | CN | Hisp |
|---|---|---|---|---|
| **DXS7423** | | | | |
| **N** | 261 | 238 | 336 | 140 |
| **8** | 0.008 | | | |
| **12** | 0.015 | | | |
| **13** | 0.080 | | 0.122 | 0.071 |
| **14** | 0.498 | 0.357 | 0.336 | 0.243 |
| **15** | 0.291 | 0.597 | 0.351 | 0.493 |
| **16** | 0.103 | 0.046 | 0.152 | 0.129 |
| **17** | 0.004 | | 0.039 | 0.064 |

| | AF | AS | CN | Hisp |
|---|---|---|---|---|
| **DXS8378** | | | | |
| **N** | 261 | 238 | 336 | 140 |
| **9** | | 0.038 | 0.015 | 0.029 |
| **10** | 0.276 | 0.576 | 0.339 | 0.414 |
| **11** | 0.398 | 0.227 | 0.339 | 0.307 |
| **12** | 0.284 | 0.147 | 0.265 | 0.229 |
| **13** | 0.042 | 0.008 | 0.042 | 0.014 |
| **14** | | 0.004 | | 0.007 |

| | AF | AS | CN | Hisp |
|---|---|---|---|---|
| **DXS10134** | | | | |
| **N** | 261 | 238 | 337 | 140 |
| **28** | 0.004 | | | |
| **28.1** | 0.004 | | | |
| **29** | 0.011 | | | 0.007 |
| **30** | 0.073 | | | 0.014 |
| **30.1** | 0.004 | | | |
| **31** | 0.019 | 0.004 | 0.003 | 0.014 |
| **32** | 0.042 | 0.017 | 0.024 | 0.014 |
| **32.1** | 0.004 | | | |
| **33** | 0.084 | 0.034 | 0.047 | 0.050 |
| **33.2** | | 0.004 | | |
| **33.3** | 0.004 | | | |
| **34** | 0.084 | 0.071 | 0.125 | 0.050 |
| **35** | 0.142 | 0.181 | 0.151 | 0.150 |
| **35.1** | 0.004 | | 0.003 | 0.007 |
| **35.2** | 0.004 | | | |
| **36** | 0.199 | 0.223 | 0.211 | 0.250 |
| **36.1** | | | | 0.007 |
| **37** | 0.176 | 0.210 | 0.160 | 0.171 |
| **37.2** | | | 0.003 | |
| **37.3** | 0.004 | 0.008 | 0.006 | 0.014 |
| **38** | 0.103 | 0.134 | 0.080 | 0.121 |
| **38.3** | 0.004 | 0.025 | 0.021 | 0.007 |
| **39** | 0.015 | 0.063 | 0.024 | 0.021 |
| **39.2** | | | | 0.007 |
| **39.3** | 0.004 | | 0.024 | 0.029 |
| **40** | | 0.004 | 0.009 | 0.007 |
| **40.3** | | 0.004 | 0.033 | 0.021 |
| **41** | | 0.008 | | |
| **41.3** | 0.011 | 0.004 | 0.030 | 0.014 |
| **42.3** | | 0.004 | 0.039 | 0.007 |
| **43.3** | | | 0.003 | 0.014 |
| **44.3** | | | 0.006 | |

| | AF | AS | CN | Hisp |
|---|---|---|---|---|
| **HPRTB** | | | | |
| **N** | 261 | 238 | 336 | 140 |
| **8** | 0.004 | | | |
| **9** | 0.034 | | 0.009 | |
| **10** | 0.023 | | 0.015 | |
| **11** | 0.084 | 0.071 | 0.116 | 0.107 |
| **12** | 0.314 | 0.265 | 0.277 | 0.314 |
| **13** | 0.215 | 0.387 | 0.363 | 0.357 |
| **14** | 0.192 | 0.189 | 0.167 | 0.157 |
| **15** | 0.119 | 0.080 | 0.033 | 0.057 |
| **16** | 0.015 | 0.008 | 0.021 | 0.007 |

| | AF | AS | CN | Hisp |
|---|---|---|---|---|
| **DXS10103** | | | | |
| **N** | 261 | 238 | 336 | 140 |
| **15** | | 0.013 | 0.021 | 0.021 |
| **16** | 0.138 | 0.265 | 0.116 | 0.243 |
| **17** | 0.069 | 0.105 | 0.140 | 0.114 |
| **18** | 0.203 | 0.181 | 0.164 | 0.136 |
| **19** | 0.444 | 0.353 | 0.423 | 0.386 |
| **20** | 0.134 | 0.076 | 0.122 | 0.071 |
| **21** | 0.011 | 0.008 | 0.015 | 0.029 |

| | AF | AS | CN | Hisp |
|---|---|---|---|---|
| **DXS7132** | | | | |
| **N** | 261 | 238 | 336 | 140 |
| **11** | 0.011 | 0.008 | 0.009 | |
| **12** | 0.088 | 0.084 | 0.077 | 0.071 |
| **13** | 0.226 | 0.164 | 0.330 | 0.243 |
| **14** | 0.360 | 0.340 | 0.354 | 0.371 |
| **15** | 0.241 | 0.294 | 0.188 | 0.250 |
| **15.3** | | | | 0.007 |
| **16** | 0.065 | 0.092 | 0.042 | 0.043 |
| **16.3** | | | | 0.007 |
| **17** | 0.008 | 0.013 | | 0.007 |
| **null** | | 0.004 | | |

AF: African American; AS: U.S. Asian; CN: U.S. Caucasian; Hisp: U.S. Hispanic; N: Number of alleles

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Forensic efficiency parameters calculated for the additional 8 markers in each U.S. population group are shown in Table 4.20. Results for the 4 markers that overlap with the two multiplexes developed as part of this study are shown for ease of comparison. Note that the U.S. Asian population studied with the Investigator Argus X-12 kit was only a subset of that studied in sample set A and included the 3 samples from sample set B; therefore, these parameters have been calculated based on this specific population size. Because both observed heterozygosity and the test for Hardy-Weinberg equilibrium require diploid individuals, neither parameter can be calculated in populations that are predominantly male such as these.

**Table 4.20. Forensic efficiency parameters for 12 X STR markers and 4 U.S. populations.** The first 8 markers listed are exclusive to the Investigator Argus X-12 kit. Pop.: population; N(alleles): number of alleles; AA: African American; AS: U.S. Asian; CN: U.S. Caucasian; Hisp: U.S. Hispanic; H(exp): expected heterozygosity; PIC: polymorphism information content, PDf: power of discrimination in females; PDm: power of discrimination in males; MECI: mean exclusion chance in trios involving daughter; MECII: mean exclusion chance in father/daughter duos.

| Marker | Pop. | N(alleles) | H(exp) | PIC | PDf | PDm | MECI | MECII |
|---|---|---|---|---|---|---|---|---|
| DXS10135 | AA | 261 | 0.9504 | 0.9482 | 0.9953 | 0.9504 | 0.9482 | 0.9039 |
| | AS | 238 | 0.9147 | 0.9084 | 0.9864 | 0.9147 | 0.9084 | 0.8381 |
| | CN | 337 | 0.9363 | 0.9326 | 0.9923 | 0.9363 | 0.9326 | 0.8775 |
| | Hisp | 140 | 0.9262 | 0.9214 | 0.9897 | 0.9262 | 0.9214 | 0.8589 |
| DXS10148 | AA | 261 | 0.9038 | 0.8979 | 0.9848 | 0.9038 | 0.8979 | 0.8239 |
| | AS | 238 | 0.8980 | 0.8893 | 0.9808 | 0.8980 | 0.8893 | 0.8085 |
| | CN | 336 | 0.8757 | 0.8631 | 0.9720 | 0.8757 | 0.8631 | 0.7697 |
| | Hisp | 140 | 0.8627 | 0.8490 | 0.9674 | 0.8627 | 0.8490 | 0.7507 |
| DXS10146 | AA | 261 | 0.9430 | 0.9400 | 0.9938 | 0.9430 | 0.9400 | 0.8898 |
| | AS | 238 | 0.8540 | 0.8381 | 0.9627 | 0.8540 | 0.8381 | 0.7349 |
| | CN | 336 | 0.8938 | 0.8844 | 0.9793 | 0.8938 | 0.8844 | 0.8013 |
| | Hisp | 140 | 0.8767 | 0.8665 | 0.9745 | 0.8767 | 0.8665 | 0.7764 |
| DXS10134 | AA | 261 | 0.8762 | 0.8640 | 0.9725 | 0.8762 | 0.8640 | 0.7714 |
| | AS | 238 | 0.8442 | 0.8257 | 0.9573 | 0.8442 | 0.8257 | 0.7176 |
| | CN | 337 | 0.8771 | 0.8655 | 0.9732 | 0.8771 | 0.8655 | 0.7739 |
| | Hisp | 140 | 0.8626 | 0.8492 | 0.9677 | 0.8626 | 0.8492 | 0.7513 |
| DXS10101 | AA | 261 | 0.9103 | 0.9034 | 0.9851 | 0.9103 | 0.9034 | 0.8304 |
| | AS | 238 | 0.8987 | 0.8900 | 0.9810 | 0.8987 | 0.8900 | 0.8094 |
| | CN | 336 | 0.9006 | 0.8923 | 0.9818 | 0.9006 | 0.8923 | 0.8131 |
| | Hisp | 141 | 0.9025 | 0.8946 | 0.9826 | 0.9025 | 0.8946 | 0.8169 |
| DXS10103 | AA | 261 | 0.7194 | 0.6820 | 0.8839 | 0.7194 | 0.6820 | 0.5402 |
| | AS | 238 | 0.7558 | 0.7178 | 0.9023 | 0.7558 | 0.7178 | 0.5815 |
| | CN | 336 | 0.7460 | 0.7149 | 0.9044 | 0.7460 | 0.7149 | 0.5780 |
| | Hisp | 140 | 0.7544 | 0.7202 | 0.9055 | 0.7544 | 0.7202 | 0.5849 |
| DXS10074 | AA | 261 | 0.8874 | 0.8769 | 0.9768 | 0.8874 | 0.8769 | 0.7899 |
| | AS | 238 | 0.7756 | 0.7426 | 0.9167 | 0.7756 | 0.7426 | 0.6110 |
| | CN | 336 | 0.8518 | 0.8343 | 0.9605 | 0.8518 | 0.8343 | 0.7285 |
| | Hisp | 140 | 0.8623 | 0.8483 | 0.9671 | 0.8623 | 0.8483 | 0.7493 |
| DXS10079 | AA | 262 | 0.8247 | 0.8020 | 0.9466 | 0.8247 | 0.8020 | 0.6856 |
| | AS | 238 | 0.8212 | 0.7975 | 0.9443 | 0.8212 | 0.7975 | 0.6794 |
| | CN | 337 | 0.8098 | 0.7849 | 0.9390 | 0.8098 | 0.7849 | 0.6638 |
| | Hisp | 140 | 0.8182 | 0.7952 | 0.9440 | 0.8182 | 0.7952 | 0.6776 |
| DXS8378 | AA | 261 | 0.6829 | 0.6199 | 0.8364 | 0.6829 | 0.6199 | 0.4730 |
| | AS | 238 | 0.5940 | 0.5421 | 0.7833 | 0.5940 | 0.5421 | 0.3937 |
| | CN | 336 | 0.6976 | 0.6376 | 0.8486 | 0.6976 | 0.6376 | 0.4920 |
| | Hisp | 140 | 0.6807 | 0.6199 | 0.8372 | 0.6807 | 0.6199 | 0.4735 |
| DXS7423 | AA | 261 | 0.6496 | 0.5958 | 0.8234 | 0.6496 | 0.5958 | 0.4491 |
| | AS | 238 | 0.5144 | 0.4216 | 0.6714 | 0.5144 | 0.4216 | 0.2868 |
| | CN | 336 | 0.7241 | 0.6768 | 0.8766 | 0.7241 | 0.6768 | 0.5350 |
| | Hisp | 140 | 0.6723 | 0.6278 | 0.8481 | 0.6723 | 0.6278 | 0.4815 |
| DXS7132 | AA | 261 | 0.7487 | 0.7085 | 0.8966 | 0.7487 | 0.7085 | 0.5707 |
| | AS | 238 | 0.7550 | 0.7167 | 0.9017 | 0.7550 | 0.7167 | 0.5806 |
| | CN | 336 | 0.7225 | 0.6744 | 0.8749 | 0.7225 | 0.6744 | 0.5326 |
| | Hisp | 140 | 0.7335 | 0.6889 | 0.8844 | 0.7335 | 0.6889 | 0.5486 |
| HPRTB | AA | 261 | 0.7954 | 0.7670 | 0.9297 | 0.7954 | 0.7670 | 0.6410 |
| | AS | 238 | 0.7332 | 0.6906 | 0.8862 | 0.7332 | 0.6906 | 0.5503 |
| | CN | 336 | 0.7626 | 0.7299 | 0.9109 | 0.7626 | 0.7299 | 0.5976 |
| | Hisp | 140 | 0.7342 | 0.6904 | 0.8855 | 0.7342 | 0.6904 | 0.5503 |

Within this set of 12 markers, DXS10135 had the highest PIC in all four populations (>0.90 in all cases). DXS10101 was also a highly discriminating marker across all four population groups (PIC >0.89). The marker with the lowest PIC was DXS7423 for the African American, U.S. Asian, and U.S. Hispanic populations (0.5958, 0.4216, and 0.6278, respectively) and DXS8378 for the U.S. Caucasian population (0.6376). Of note, the four markers that overlap with the multiplexes developed as part of this study (DXS8378, DXS7423, DXS7132, HPRTB), along with DXS10103, had the 5 lowest PIC values in all four populations. For a detailed comparison between the two assays (Investigator Argus X-12 kit versus the two multiplexes developed here) using U.S. populations, see Chapter 6.

*4.1.5. Combined United States Population Database*

The final objective of the databasing study was to merge the individual populations from sample sets A, B, and C into one U.S. population dataset representing the four main population groups (African American, U.S. Asian, U.S. Caucasian, U.S. Hispanic) in order to provide a large and robust database for application in cases involving suspected or known U.S. X STR profiles. Before combining the sample sets, however, the genetic similarity of the populations was considered. Pairwise $F_{ST}$ values comparing populations within each sample set at each marker were calculated, and the corresponding p values are shown in Table 4.21. P values indicative of populations that are not significantly different from one another ($p > 0.05$) were observed for 127 (94%) of 135 comparisons between equivalent populations (gray shading in Table 4.21). The 8 significant differences were observed at 6 of the 15 markers with African American or U.S. Hispanic populations only. The most common occurrence of significant differences occurred 3 times between the U.S. Hispanic populations of sample sets B & C; all other differences were observed in only one or two comparisons or at one or two markers. Similarly, 293 (81%) of 360 comparisons between unmatched populations generated p values indicating significant differences from one another ($p < 0.05$), with a lack of similarity confirmed for every unmatched pair at 5 markers (DXS6795, DXS6803, DXS7130, DXS10147, and GATA165B12). Of the 67 comparisons exhibiting no significant differences, the majority (52%) occurred between some combination of U.S. Caucasian and U.S. Hispanic populations across one or more of the 10 affected

markers. Marker DXS7132 displayed the weakest ability to discriminate between these U.S. populations, failing to detect significant differences between 16 (67%) of 24 comparisons of unmatched populations.

**Table 4.21. P values (with standard deviations) corresponding to pairwise $F_{ST}$ values (not shown) comparing the individual U.S. populations from each sample set.** P values indicative of populations that are significantly different from one another ($p < 0.5$) are bolded. Values at the intersection of equivalent populations in different sample sets are shaded in gray. AA: African American; AS: U.S. Asian; CN: U.S. Caucasian; Hisp: U.S. Hispanic.

| | | | Sample Set B | | | Sample Set C | | |
|---|---|---|---|---|---|---|---|---|
| **Marker** | **Sample Set** | **Popul-ation** | **AA** | **CN** | **Hisp** | **AA** | **CN** | **Hisp** |
| **DXS101** | **A** | **AA** | 0.07465 ±0.0027 | **0.00000 ±0.0000** | **0.00000 ±0.0000** | 0.03643 ±0.0019 | **0.00000 ±0.0000** | **0.00000± 0.0000** |
| | | **AS** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00030 ±0.0002** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00000± 0.0000** |
| | | **CN** | **0.00000 ±0.0000** | 0.33442 ±0.0042 | 0.19998 ±0.0043 | **0.00000 ±0.0000** | 0.26760 ±0.0050 | 0.05089± 0.0022 |
| | | **Hisp** | **0.00000 ±0.0000** | **0.00020 ±0.0001** | 0.40283 ±0.0047 | **0.00000 ±0.0000** | **0.00050 ±0.0002** | 0.59509± 0.0052 |
| | **C** | **AA** | 0.96000 ±0.0020 | **0.00000 ±0.0000** | **0.00000 ±0.0000** | | | |
| | | **CN** | **0.00000 ±0.0000** | 0.54262 ±0.0056 | 0.08039 ±0.0028 | | | |
| | | **Hisp** | **0.00000 ±0.0000** | **0.00129 ±0.0004** | 0.38907 ±0.0044 | | | |
| **DXS6789** | **A** | **AA** | 0.52777 ±0.0046 | **0.00000 ±0.0000** | **0.00000 ±0.0000** | 0.69686 ±0.0044 | **0.00000 ±0.0000** | **0.00000± 0.0000** |
| | | **AS** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00000± 0.0000** |
| | | **CN** | **0.00000 ±0.0000** | 0.13246 ±0.0035 | 0.63627 ±0.0053 | **0.00000 ±0.0000** | 0.23582 ±0.0043 | 0.23453± 0.0043 |
| | | **Hisp** | **0.00000 ±0.0000** | 0.21196 ±0.0042 | 0.46679 ±0.0051 | **0.00000 ±0.0000** | 0.09158 ±0.0032 | 0.18107± 0.0038 |
| | **C** | **AA** | 0.78002 ±0.0041 | **0.00000 ±0.0000** | **0.00000 ±0.0000** | | | |
| | | **CN** | **0.00000 ±0.0000** | 0.54391 ±0.0052 | 0.52628 ±0.0055 | | | |
| | | **Hisp** | **0.00000 ±0.0000** | **0.03257 ±0.0017** | 0.50421 ±0.0052 | | | |
| **DXS6795** | **A** | **AA** | **0.02317 ±0.0015** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | 0.38075 ±0.0047 | **0.00000 ±0.0000** | **0.00000± 0.0000** |
| | | **AS** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00000± 0.0000** |
| | | **CN** | **0.00000 ±0.0000** | 0.61736 ±0.0045 | **0.00000 ±0.0000** | **0.00000 ±0.0000** | 0.87130 ±0.0031 | **0.00000± 0.0000** |
| | | **Hisp** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | 0.18038 ±0.0036 | **0.00000 ±0.0000** | **0.00000 ±0.0000** | 0.29126± 0.0039 |
| | **C** | **AA** | 0.43253 ±0.0051 | **0.00000 ±0.0000** | **0.00000 ±0.0000** | | | |
| | | **CN** | **0.00000 ±0.0000** | 0.47282 ±0.0052 | **0.00010 ±0.0001** | | | |
| | | **Hisp** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.04831 ±0.0020** | | | |

Table continues on next page.

| Marker | Sample Set | Popul-ation | Sample Set B AA | CN | Hisp | Sample Set C AA | CN | Hisp |
|---|---|---|---|---|---|---|---|---|
| **DXS6803** | **A** | **AA** | 0.22800 ±0.0042 | **0.00000 ±0.0000** | **0.00000 ±0.0000** | 0.63885 ±0.0050 | **0.00000 ±0.0000** | **0.00000± 0.0000** |
| | | **AS** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00000± 0.0000** |
| | | **CN** | **0.00000 ±0.0000** | 0.25443 ±0.0047 | **0.03287 ±0.0019** | **0.00000 ±0.0000** | 0.57123 ±0.0053 | **0.00000± 0.0000** |
| | | **Hisp** | **0.00000 ±0.0000** | **0.00129 ±0.0004** | 0.11306 ±0.0030 | **0.00000 ±0.0000** | **0.00040 ±0.0002** | 0.36185± 0.0046 |
| | **C** | **AA** | 0.58885 ±0.0047 | **0.00000 ±0.0000** | **0.00030 ±0.0002** | | | |
| | | **CN** | **0.00000 ±0.0000** | 0.68874 ±0.0048 | **0.03059 ±0.0016** | | | |
| | | **Hisp** | **0.00000 ±0.0000** | **0.00030 ±0.0002** | 0.24196 ±0.0042 | | | |
| **DXS7130** | **A** | **AA** | 0.20859 ±0.0043 | **0.00000 ±0.0000** | **0.00000 ±0.0000** | 0.76012 ±0.0040 | **0.00000 ±0.0000** | **0.00000± 0.0000** |
| | | **AS** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00030 ±0.0002** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00000± 0.0000** |
| | | **CN** | **0.00000 ±0.0000** | 0.36066 ±0.0049 | **0.01772 ±0.0014** | **0.00000 ±0.0000** | 0.59578 ±0.0046 | **0.00089± 0.0003** |
| | | **Hisp** | **0.00010 ±0.0001** | **0.00010 ±0.0001** | 0.28027 ±0.0038 | **0.00010 ±0.0001** | **0.00000 ±0.0000** | 0.07673± 0.0028 |
| | **C** | **AA** | 0.66875 ±0.0048 | **0.00000 ±0.0000** | **0.00000 ±0.0000** | | | |
| | | **CN** | **0.00000 ±0.0000** | 0.62439 ±0.0049 | **0.01346 ±0.0010** | | | |
| | | **Hisp** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | 0.35264 ±0.0045 | | | |
| **DXS7132** | **A** | **AA** | 0.54034 ±0.0052 | 0.10207 ±0.0034 | 0.79180 ±0.0041 | 0.48371 ±0.0052 | 0.57767 ±0.0055 | 0.59271± 0.0048 |
| | | **AS** | 0.29522 ±0.0048 | **0.00000 ±0.0000** | 0.29888 ±0.0044 | **0.00040 ±0.0002** | **0.00000 ±0.0000** | **0.00178± 0.0004** |
| | | **CN** | **0.02010 ±0.0015** | 0.58588 ±0.0045 | 0.07890 ±0.0026 | **0.02188 ±0.0015** | 0.23156 ±0.0041 | **0.00683± 0.0008** |
| | | **Hisp** | 0.46441 ±0.0048 | 0.10662 ±0.0027 | 0.67775 ±0.0054 | 0.70874 ±0.0049 | 0.28641 ±0.0044 | 0.44926± 0.0047 |
| | **C** | **AA** | 0.54371 ±0.0048 | 0.06138 ±0.0022 | 0.62073 ±0.0052 | | | |
| | | **CN** | 0.27878 ±0.0050 | 0.45025 ±0.0054 | 0.59271 ±0.0047 | | | |
| | | **Hisp** | 0.70557 ±0.0048 | **0.02416 ±0.0016** | 0.81992 ±0.0038 | | | |
| **DXS7423** | **A** | **AA** | 0.52094 ±0.0053 | **0.00000 ±0.0000** | **0.00000 ±0.0000** | 0.36086 ±0.0051 | **0.00000 ±0.0000** | **0.00000± 0.0000** |
| | | **AS** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00129 ±0.0004** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00000± 0.0000** |
| | | **CN** | **0.00030 ±0.0002** | 0.58648 ±0.0054 | **0.01663 ±0.0012** | **0.00337 ±0.0005** | 0.14593 ±0.0038 | **0.00079± 0.0003** |
| | | **Hisp** | **0.00000 ±0.0000** | **0.00010 ±0.0001** | 0.32393 ±0.0046 | **0.00000 ±0.0000** | **0.00020 ±0.0001** | 0.98931± 0.0010 |
| | **C** | **AA** | 0.30551 ±0.0050 | **0.00307 ±0.0006** | **0.00000 ±0.0000** | | | |
| | | **CN** | **0.00000 ±0.0000** | 0.23661 ±0.0040 | 0.05415 ±0.0022 | | | |
| | | **Hisp** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | 0.35036 ±0.0048 | | | |

Table continues on next page.

| Marker | Sample Set | Popul-ation | Sample Set B | | | Sample Set C | | |
|---|---|---|---|---|---|---|---|---|
| | | | AA | CN | Hisp | AA | CN | Hisp |
| **DXS7424** | **A** | **AA** | 0.92961 ±0.0025 | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.04316 ±0.0021** | **0.00000 ±0.0000** | **0.00000± 0.0000** |
| | | **AS** | **0.00000 ±0.0000** | **0.00366 ±0.0005** | **0.00535 ±0.0008** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00000± 0.0000** |
| | | **CN** | **0.00000 ±0.0000** | 0.19404 ±0.0044 | 0.23018 ±0.0048 | **0.00000 ±0.0000** | 0.75448 ±0.0041 | **0.04069± 0.0019** |
| | | **Hisp** | **0.00000 ±0.0000** | 0.32324 ±0.0045 | 0.97228 ±0.0016 | **0.00000 ±0.0000** | 0.10484 ±0.0031 | 0.23166± 0.0042 |
| | **C** | **AA** | 0.51579 ±0.0052 | **0.00000 ±0.0000** | **0.00000 ±0.0000** | | | |
| | | **CN** | **0.00000 ±0.0000** | 0.29868 ±0.0043 | 0.21879 ±0.0043 | | | |
| | | **Hisp** | **0.00010 ±0.0001** | **0.00901 ±0.0009** | 0.49797 ±0.0051 | | | |
| **DXS8378** | **A** | **AA** | 0.55074 ±0.0045 | 0.16078 ±0.0039 | **0.01129 ±0.0010** | 0.77497 ±0.0042 | 0.09286 ±0.0030 | **0.00000± 0.0000** |
| | | **AS** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00178 ±0.0005** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00010± 0.0001** |
| | | **CN** | 0.08861 ±0.0029 | 0.90110 ±0.0028 | 0.37610 ±0.0050 | **0.01287 ±0.0010** | 0.70755 ±0.0047 | **0.00446± 0.0007** |
| | | **Hisp** | **0.01238 ±0.0012** | 0.47777 ±0.0045 | 0.63736 ±0.0047 | **0.00208 ±0.0005** | 0.15830 ±0.0036 | **0.00911± 0.0011** |
| | **C** | **AA** | 0.66835 ±0.0049 | 0.06296 ±0.0025 | **0.00307 ±0.0005** | | | |
| | | **CN** | 0.17761 ±0.0037 | 0.87298 ±0.0033 | 0.14781 ±0.0032 | | | |
| | | **Hisp** | **0.00000 ±0.0000** | **0.00218 ±0.0004** | 0.50975 ±0.0048 | | | |
| **DXS9902** | **A** | **AA** | 0.59400 ±0.0052 | 0.09474 ±0.0030 | **0.03455 ±0.0018** | 0.20998 ±0.0039 | 0.21701 ±0.0042 | **0.00020± 0.0001** |
| | | **AS** | **0.00901 ±0.0009** | **0.04316 ±0.0021** | 0.61509 ±0.0048 | **0.00010 ±0.0001** | **0.00069 ±0.0003** | **0.00010± 0.0001** |
| | | **CN** | 0.30522 ±0.0044 | 0.55460 ±0.0051 | 0.19691 ±0.0039 | **0.02713 ±0.0017** | 0.64211 ±0.0049 | 0.08771± 0.0025 |
| | | **Hisp** | 0.19493 ±0.0043 | 0.72646 ±0.0052 | 0.60707 ±0.0048 | **0.02495 ±0.0014** | 0.20869 ±0.0043 | 0.05475± 0.0025 |
| | **C** | **AA** | 0.90545 ±0.0027 | 0.10425 ±0.0032 | 0.09672 ±0.0031 | | | |
| | | **CN** | 0.37076 ±0.0052 | 0.76418 ±0.0039 | 0.17978 ±0.0041 | | | |
| | | **Hisp** | **0.01020 ±0.0009** | **0.01634 ±0.0014** | **0.02129 ±0.0016** | | | |
| **DXS10147** | **A** | **AA** | 0.28562 ±0.0044 | **0.00000 ±0.0000** | **0.00000 ±0.0000** | 0.98832 ±0.0011 | **0.00000 ±0.0000** | **0.00000± 0.0000** |
| | | **AS** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00000± 0.0000** |
| | | **CN** | **0.00000 ±0.0000** | 0.64994 ±0.0048 | **0.00248 ±0.0005** | **0.00000 ±0.0000** | 0.88763 ±0.0032 | **0.00000± 0.0000** |
| | | **Hisp** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | 0.02663 ±0.0017 | **0.00000 ±0.0000** | **0.00000 ±0.0000** | 0.14672± 0.0037 |
| | **C** | **AA** | 0.24493 ±0.0041 | **0.00000 ±0.0000** | **0.00000 ±0.0000** | | | |
| | | **CN** | **0.00000 ±0.0000** | 0.74062 ±0.0045 | **0.00139 ±0.0004** | | | |
| | | **Hisp** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | 0.22216 ±0.0039 | | | |

Table continues on next page.

| Marker | Sample Set | Popul-ation | Sample Set B | | | Sample Set C | | |
|---|---|---|---|---|---|---|---|---|
| | | | AA | CN | Hisp | AA | CN | Hisp |
| **GATA31E08** | **A** | **AA** | **0.00287 ±0.0006** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | 0.32432 ±0.0044 | **0.00000 ±0.0000** | **0.00000± 0.0000** |
| | | **AS** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00238 ±0.0005** | **0.00000 ±0.0000** | **0.00020 ±0.0001** | **0.00000± 0.0000** |
| | | **CN** | **0.00000 ±0.0000** | 0.29878 ±0.0047 | 0.11187 ±0.0033 | **0.00000 ±0.0000** | 0.85001 ±0.0037 | **0.00000± 0.0000** |
| | | **Hisp** | 0.09524 ±0.0029 | **0.00000 ±0.0000** | 0.14405 ±0.0038 | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00574± 0.0008** |
| | **C** | **AA** | 0.07128 ±0.0026 | **0.00000 ±0.0000** | **0.00010 ±0.0001** | | | |
| | | **CN** | **0.00000 ±0.0000** | 0.23107 ±0.0044 | 0.23235 ±0.0043 | | | |
| | | **Hisp** | **0.00099 ±0.0003** | **0.00000 ±0.0000** | **0.01614 ±0.0012** | | | |
| **GATA165B12** | **A** | **AA** | 0.14771 ±0.0031 | **0.00000 ±0.0000** | **0.00188 ±0.0004** | **0.03970 ±0.0020** | **0.00000 ±0.0000** | **0.00000± 0.0000** |
| | | **AS** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00307 ±0.0005** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00020± 0.0002** |
| | | **CN** | **0.00000 ±0.0000** | 0.52054 ±0.0051 | **0.00089 ±0.0003** | **0.00000 ±0.0000** | 0.63687 ±0.0046 | **0.00000± 0.0000** |
| | | **Hisp** | **0.00000 ±0.0000** | **0.00079 ±0.0003** | 0.45075 ±0.0051 | **0.00000 ±0.0000** | **0.00079 ±0.0003** | 0.61499± 0.0052 |
| | **C** | **AA** | 0.97347 ±0.0016 | **0.00000 ±0.0000** | **0.00010 ±0.0001** | | | |
| | | **CN** | **0.00000 ±0.0000** | 0.41659 ±0.0049 | **0.00307 ±0.0005** | | | |
| | | **Hisp** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | 0.44095 ±0.0049 | | | |
| **GATA172D05** | **A** | **AA** | 0.72904 ±0.0040 | **0.00000 ±0.0000** | **0.00000 ±0.0000** | 0.39372 ±0.0053 | **0.00000 ±0.0000** | **0.00000± 0.0000** |
| | | **AS** | **0.00000 ±0.0000** | **0.00030 ±0.0002** | **0.02990 ±0.0014** | **0.00000 ±0.0000** | **0.00000 ±0.0000** | **0.00000± 0.0000** |
| | | **CN** | **0.00000 ±0.0000** | 0.64607 ±0.0054 | 0.75270 ±0.0049 | **0.00000 ±0.0000** | 0.75735 ±0.0040 | **0.00277± 0.0005** |
| | | **Hisp** | **0.00000 ±0.0000** | 0.19434 ±0.0042 | 0.66152 ±0.0044 | **0.00000 ±0.0000** | **0.03663 ±0.0017** | 0.30690± 0.0048 |
| | **C** | **AA** | 0.92169 ±0.0029 | **0.00000 ±0.0000** | **0.00000 ±0.0000** | | | |
| | | **CN** | **0.00000 ±0.0000** | 0.50054 ±0.0053 | 0.70775 ±0.0044 | | | |
| | | **Hisp** | **0.00000 ±0.0000** | **0.00267 ±0.0006** | 0.16424 ±0.0034 | | | |
| **HPRTB** | **A** | **AA** | 0.10435 ±0.0032 | 0.05960 ±0.0022 | 0.36769 ±0.0050 | 0.21295 ±0.0043 | 0.05881 ±0.0023 | **0.00614± 0.0008** |
| | | **AS** | **0.00000 ±0.0000** | 0.20988 ±0.0039 | 0.30888 ±0.0043 | **0.00000 ±0.0000** | **0.00000 ±0.0000** | 0.43025± 0.0051 |
| | | **CN** | **0.00455 ±0.0007** | 0.16810 ±0.0039 | 0.82913 ±0.0037 | **0.00149 ±0.0004** | 0.82655 ±0.0034 | **0.03257± 0.0017** |
| | | **Hisp** | **0.00020 ±0.0001** | 0.33234 ±0.0052 | 0.33858 ±0.0050 | **0.00010 ±0.0001** | **0.00069 ±0.0003** | 0.72626± 0.0041 |
| | **C** | **AA** | 0.30284 ±0.0046 | **0.00109 ±0.0003** | **0.02891 ±0.0017** | | | |
| | | **CN** | **0.00119 ±0.0003** | 0.13127 ±0.0034 | 0.83091 ±0.0040 | | | |
| | | **Hisp** | **0.00000 ±0.0000** | 0.60746 ±0.0046 | 0.75448 ±0.0039 | | | |

The described inconsistencies when comparing population groups from the various sample sets could be due to a number of factors, including sampling effects related to admixture and the use of self-described ancestry, especially within the African

American and U.S. Hispanic groups, as alluded to previously. A published study examining maternally, paternally, and bi-parentally inherited (and forensically-relevant) DNA markers using samples overlapping with sample set B demonstrated the existence of a difference in relative composition of continental ancestry for the three modes of inheritance in the African American and U.S. Hispanic population groups [299]. Further, this ancestral composition was shown to vary by region throughout the country, which could also contribute to the variation observed for X STRs in these sample sets. Overall, however, equivalent populations were sufficiently consistent and unmatched populations sufficiently different to warrant combination into a larger database for routine use. Allele frequencies and forensic genetic parameters for this combined dataset are presented in Table 4.22.

**Table 4.22. Allele frequencies and forensic efficiency parameters for 15 X STR markers in four U.S. populations.** AA: African American; AS: U.S. Asian; CN: U.S. Caucasian; Hisp: U.S. Hispanic; N: number of alleles; PIC: polymorphism information content; H(exp): expected heterozygosity; H(obs): observed heterozygosity; PDf: power of discrimination in females; PDm: power of discrimination in males; MECI: mean exclusion chance in trios involving daughter; MECII: mean exclusion chance in father/daughter duos; p(HWE): p value for the exact test for Hardy-Weinberg equilibrium.

Table begins on next page.

| | DXS6795 | | | | DXS9902 | | | | DXS8378 | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | **AA** | **AS** | **CN** | **Hisp** | **AA** | **AS** | **CN** | **Hisp** | **AA** | **AS** | **CN** | **Hisp** | |
| N | 1303 | 804 | 1453 | 1151 | 1303 | 804 | 1453 | 1151 | 1303 | 804 | 1453 | 1151 | N |
| 5 | | | | | | | | | | | | | 5 |
| 6 | 0.0008 | | | | | | | | | | | | 6 |
| 7 | | | | | 0.0008 | 0.0037 | 0.0007 | | | | | | 7 |
| 8 | 0.0008 | | | | 0.0483 | | | 0.0035 | 0.0038 | 0.0012 | 0.0048 | 0.0009 | 8 |
| 8.3 | | | | | | | | | | | | | 8.3 |
| 9 | 0.1197 | 0.0410 | 0.2973 | 0.1538 | 0.0683 | 0.0174 | 0.0427 | 0.0174 | 0.0092 | 0.0224 | 0.0145 | 0.0156 | 9 |
| 9.3 | | | | | | | | | | | | | 9.3 |
| 10 | 0.3008 | 0.1704 | 0.0255 | 0.1034 | 0.3024 | 0.4229 | 0.3283 | 0.3701 | 0.2732 | 0.5622 | 0.3386 | 0.4205 | 10 |
| 10.1 | | | | | | | | 0.0035 | | | | | 10.1 |
| 10.3 | | | | | | | | 0.0009 | | | | | 10.3 |
| 11 | 0.1719 | 0.3122 | 0.4591 | 0.2441 | 0.3469 | 0.3346 | 0.3641 | 0.3858 | 0.3722 | 0.2662 | 0.3489 | 0.3232 | 11 |
| 11.1 | | | | | 0.0084 | | 0.0317 | 0.0287 | | | | | 11.1 |
| 11.3 | | | | | | | | | | | | | 11.3 |
| 12 | 0.0844 | 0.0323 | 0.0330 | 0.1503 | 0.2157 | 0.2114 | 0.2182 | 0.1807 | 0.3039 | 0.1306 | 0.2567 | 0.2155 | 12 |
| 12.1 | | | | | | | 0.0021 | 0.0035 | | | | | 12.1 |
| 12.3 | | | | | | | | | | | | | 12.3 |
| 13 | 0.0844 | 0.4204 | 0.1810 | 0.3119 | 0.0084 | 0.0087 | 0.0124 | 0.0043 | 0.0353 | 0.0124 | 0.0344 | 0.0217 | 13 |
| 13.3 | | | | | | | | | | | | | 13.3 |
| 14 | 0.0437 | 0.0211 | 0.0007 | 0.0209 | 0.0008 | 0.0012 | | 0.0017 | 0.0023 | 0.0037 | 0.0021 | 0.0026 | 14 |
| 14.3 | | | | | | | | | | | | | 14.3 |
| 15 | 0.1781 | 0.0025 | 0.0034 | 0.0148 | | | | | | 0.0012 | | | 15 |
| 15.3 | | | | | | | | | | | | | 15.3 |
| 16 | 0.0092 | | | 0.0009 | | | | | | | | | 16 |
| 16.3 | | | | | | | | | | | | | 16.3 |
| 17 | 0.0054 | | | | | | | | | | | | 17 |
| 17.3 | | | | | | | | | | | | | 17.3 |
| 18 | 0.0008 | | | | | | | | | | | | 18 |
| 18.3 | | | | | | | | | | | | | 18.3 |
| 19 | | | | | | | | | | | | | 19 |
| 20 | | | | | | | | | | | | | 20 |
| 20.2 | | | | | | | | | | | | | 20.2 |
| 20.3 | | | | | | | | | | | | | 20.3 |
| 21 | | | | | | | | | | | | | 21 |
| 22 | | | | | | | | | | | | | 22 |
| 23 | | | | | | | | | | | | | 23 |
| 24 | | | | | | | | | | | | | 24 |
| 25 | | | | | | | | | | | | | 25 |
| 26 | | | | | | | | | | | | | 26 |
| 27 | | | | | | | | | | | | | 27 |
| 28 | | | | | | | | | | | | | 28 |
| 29 | | | | | | | | | | | | | 29 |
| 30 | | | | | | | | | | | | | 30 |
| 31 | | | | | | | | | | | | | 31 |
| 32 | | | | | | | | | | | | | 32 |
| 33 | | | | | | | | | | | | | 33 |
| PIC | 0.7948 | 0.6413 | 0.6083 | 0.7537 | 0.6891 | 0.5978 | 0.6560 | 0.6199 | 0.6328 | 0.5368 | 0.6364 | 0.6079 | PIC |
| H(exp) | 0.8177 | 0.6936 | 0.6663 | 0.7856 | 0.7345 | 0.6641 | 0.7092 | 0.6801 | 0.6935 | 0.5953 | 0.6963 | 0.6715 | H(exp) |
| H(obs) | 0.8263 | 0.6567 | 0.6358 | 0.8135 | 0.7316 | 0.6933 | 0.6954 | 0.6838 | 0.6605 | 0.5700 | 0.7130 | 0.6838 | H(obs) |
| PDf | 0.9439 | 0.8538 | 0.8306 | 0.9222 | 0.8841 | 0.8209 | 0.8623 | 0.8375 | 0.8453 | 0.7777 | 0.8478 | 0.8285 | PDf |
| PDm | 0.8177 | 0.6936 | 0.6663 | 0.7856 | 0.7345 | 0.6641 | 0.7092 | 0.6801 | 0.6935 | 0.5953 | 0.6963 | 0.6715 | PDm |
| MECI | 0.7948 | 0.6413 | 0.6083 | 0.7537 | 0.6891 | 0.5978 | 0.6560 | 0.6199 | 0.6328 | 0.5368 | 0.6364 | 0.6079 | MECI |
| MECII | 0.6763 | 0.4970 | 0.4617 | 0.6241 | 0.5489 | 0.4508 | 0.5125 | 0.4745 | 0.4869 | 0.3895 | 0.4909 | 0.4611 | MECII |
| p(HWE) | 0.8104 | 0.1521 | 0.0919 | 0.1968 | 0.7303 | 0.8160 | 0.0773 | 0.6524 | 0.8187 | 0.5703 | 0.5423 | 0.8691 | p(HWE) |

Table continues on next page.

| | GATA172D05 | | | | DXS7132 | | | | DXS6803 | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | **AA** | **AS** | **CN** | **Hisp** | **AA** | **AS** | **CN** | **Hisp** | **AA** | **AS** | **CN** | **Hisp** | |
| N | 1303 | 804 | 1453 | 1151 | 1303 | 804 | 1454 | 1151 | 1303 | 804 | 1453 | 1151 | N |
| 5 | | | | | | | | | | | | | 5 |
| 6 | 0.1934 | 0.0560 | 0.1624 | 0.1173 | | | | | | | | | 6 |
| 7 | 0.0568 | 0.0087 | 0.0007 | 0.0087 | | | | | 0.0046 | | | 0.0017 | 7 |
| 8 | 0.1750 | 0.1866 | 0.1741 | 0.1442 | | | | | 0.0130 | | | | 8 |
| 8.3 | | | | | | | | | | | | | 8.3 |
| 9 | 0.2863 | 0.0933 | 0.0516 | 0.0617 | | | | | 0.0322 | | 0.0007 | 0.0035 | 9 |
| 9.3 | | | | | | | | | | | 0.0007 | | 9.3 |
| 10 | 0.1404 | 0.3831 | 0.2918 | 0.2954 | | 0.0012 | | 0.0009 | 0.1182 | 0.0112 | 0.0392 | 0.0243 | 10 |
| 10.1 | | | | | | | | | | | | | 10.1 |
| 10.3 | | | | | | | | | | | | 0.0009 | 10.3 |
| 11 | 0.1028 | 0.2264 | 0.2044 | 0.2728 | 0.0153 | 0.0037 | 0.0110 | 0.0026 | 0.3638 | 0.1940 | 0.2423 | 0.2650 | 11 |
| 11.1 | | | | | | | | | | | | | 11.1 |
| 11.3 | | | | | | | | | 0.0092 | 0.0684 | 0.0138 | 0.0539 | 11.3 |
| 12 | 0.0445 | 0.0460 | 0.1122 | 0.0982 | 0.0998 | 0.0808 | 0.0846 | 0.0921 | 0.2417 | 0.1430 | 0.2629 | 0.3223 | 12 |
| 12.1 | | | | | | | | | | | | | 12.1 |
| 12.3 | | | | | | | | | 0.0737 | 0.4415 | 0.1321 | 0.1590 | 12.3 |
| 13 | 0.0008 | | 0.0028 | 0.0017 | 0.2510 | 0.1716 | 0.3012 | 0.2563 | 0.0867 | 0.0398 | 0.1452 | 0.0652 | 13 |
| 13.3 | | | | | | | | | 0.0292 | 0.0945 | 0.1425 | 0.0973 | 13.3 |
| 14 | | | | | 0.3515 | 0.3694 | 0.3590 | 0.3440 | 0.0238 | 0.0025 | 0.0055 | 0.0061 | 14 |
| 14.3 | | | | | | | | | 0.0015 | 0.0050 | 0.0124 | 0.0009 | 14.3 |
| 15 | | | | | 0.2264 | 0.2724 | 0.1857 | 0.2424 | 0.0015 | | | | 15 |
| 15.3 | | | | | | | | 0.0009 | | | 0.0028 | | 15.3 |
| 16 | | | | | 0.0422 | 0.0833 | 0.0516 | 0.0443 | 0.0008 | | | | 16 |
| 16.3 | | | | | 0.0031 | | | 0.0017 | | | | | 16.3 |
| 17 | | | | | 0.0084 | 0.0174 | 0.0055 | 0.0113 | | | | | 17 |
| 17.3 | | | | | | | | 0.0009 | | | | | 17.3 |
| 18 | | | | | 0.0023 | | 0.0014 | 0.0026 | | | | | 18 |
| 18.3 | | | | | | | | | | | | | 18.3 |
| 19 | | | | | | | | | | | | | 19 |
| 20 | | | | | | | | | | | | | 20 |
| 20.2 | | | | | | | | | | | | | 20.2 |
| 20.3 | | | | | | | | | | | | | 20.3 |
| 21 | | | | | | | | | | | | | 21 |
| 22 | | | | | | | | | | | | | 22 |
| 23 | | | | | | | | | | | | | 23 |
| 24 | | | | | | | | | | | | | 24 |
| 25 | | | | | | | | | | | | | 25 |
| 26 | | | | | | | | | | | | | 26 |
| 27 | | | | | | | | | | | | | 27 |
| 28 | | | | | | | | | | | | | 28 |
| 29 | | | | | | | | | | | | | 29 |
| 30 | | | | | | | | | | | | | 30 |
| 31 | | | | | | | | | | | | | 31 |
| 32 | | | | | | | | | | | | | 32 |
| 33 | | | | | | | | | | | | | 33 |
| PIC | 0.7899 | 0.7178 | 0.7724 | 0.7604 | 0.7097 | 0.7067 | 0.6923 | 0.7041 | 0.7521 | 0.6996 | 0.7853 | 0.7537 | PIC |
| H(exp) | 0.8147 | 0.7532 | 0.8011 | 0.7905 | 0.7502 | 0.7461 | 0.7359 | 0.7465 | 0.7795 | 0.7316 | 0.8114 | 0.7840 | H(exp) |
| H(obs) | 0.8500 | 0.7400 | 0.7881 | 0.7676 | 0.8026 | 0.7533 | 0.7660 | 0.7081 | 0.7974 | 0.7100 | 0.7947 | 0.7730 | H(obs) |
| PDf | 0.9409 | 0.9037 | 0.9317 | 0.926 | 0.8971 | 0.8962 | 0.8866 | 0.8933 | 0.9240 | 0.8959 | 0.9383 | 0.9231 | PDf |
| PDm | 0.8147 | 0.7532 | 0.8011 | 0.7905 | 0.7502 | 0.7461 | 0.7359 | 0.7465 | 0.7795 | 0.7316 | 0.8114 | 0.7840 | PDm |
| MECI | 0.7899 | 0.7178 | 0.7724 | 0.7604 | 0.7097 | 0.7067 | 0.6923 | 0.7041 | 0.7521 | 0.6996 | 0.7853 | 0.7537 | MECI |
| MECII | 0.6695 | 0.5818 | 0.6468 | 0.6327 | 0.5723 | 0.5690 | 0.5526 | 0.5657 | 0.6241 | 0.5608 | 0.6636 | 0.6252 | MECII |
| p(HWE) | 0.2259 | 0.5898 | 0.1658 | 0.8860 | 0.4097 | 0.9305 | 0.5191 | 0.2614 | 0.9913 | 0.8852 | 0.8488 | 0.2787 | p(HWE) |

Table continues on next page.

| | DXS6789 | | | | GATA165B12 | | | | DXS7130 | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | **AA** | **AS** | **CN** | **Hisp** | **AA** | **AS** | **CN** | **Hisp** | **AA** | **AS** | **CN** | **Hisp** | |
| N | 1303 | 804 | 1453 | 1151 | 1303 | 804 | 1453 | 1151 | 1303 | 804 | 1453 | 1151 | N |
| 5 | | | | | | | | | | | | | 5 |
| 6 | | | | | | | | | | | | | 6 |
| 7 | | | | | 0.0015 | | | | | | | | 7 |
| 8 | | | | | 0.0368 | 0.0037 | 0.0083 | 0.0087 | | | | | 8 |
| 8.3 | | | | | | | | | | | | | 8.3 |
| 9 | | | | | 0.1328 | 0.2575 | 0.3152 | 0.2381 | 0.0054 | | 0.0007 | | 9 |
| 9.3 | | | | | | | | | | | | | 9.3 |
| 10 | | | | | 0.3285 | 0.5622 | 0.3297 | 0.4726 | 0.0253 | 0.0075 | 0.0055 | 0.0035 | 10 |
| 10.1 | | | | | | | | | | | | | 10.1 |
| 10.3 | | | | | | | | | | | | | 10.3 |
| 11 | | | | | 0.3945 | 0.1505 | 0.3159 | 0.2520 | 0.0698 | 0.2127 | 0.0392 | 0.0608 | 11 |
| 11.1 | | | | | | | | | | | | | 11.1 |
| 11.3 | | | | | | | | | | | | | 11.3 |
| 12 | | | | | 0.0975 | 0.0261 | 0.0310 | 0.0261 | 0.2141 | 0.1816 | 0.0998 | 0.1981 | 12 |
| 12.1 | | | | | | | | | | | | | 12.1 |
| 12.3 | | | | | | | | | | | | | 12.3 |
| 13 | | | | | 0.0084 | | | 0.0026 | 0.1596 | 0.0672 | 0.0392 | 0.0634 | 13 |
| 13.3 | | | | | | | | | 0.0200 | 0.0050 | 0.0557 | 0.0226 | 13.3 |
| 14 | 0.0038 | 0.0050 | 0.0028 | 0.0070 | | | | | 0.0445 | 0.0050 | 0.0096 | 0.0182 | 14 |
| 14.3 | | | | | | | | | 0.1704 | 0.0796 | 0.2168 | 0.1894 | 14.3 |
| 15 | 0.2318 | 0.1567 | 0.0454 | 0.0417 | | | | | 0.0015 | | | 0.0017 | 15 |
| 15.3 | | | | | | | | | 0.2203 | 0.3371 | 0.3627 | 0.3545 | 15.3 |
| 16 | 0.1136 | 0.3644 | 0.0172 | 0.0426 | | | | | 0.0008 | | | | 16 |
| 16.3 | | | | | | | | | 0.0614 | 0.0958 | 0.1466 | 0.0678 | 16.3 |
| 17 | 0.0069 | 0.0311 | 0.0028 | 0.0043 | | | | | 0.0008 | | | | 17 |
| 17.3 | | | | | | | | | 0.0054 | 0.0087 | 0.0234 | 0.0191 | 17.3 |
| 18 | 0.0146 | | 0.0014 | 0.0035 | | | | | | | | | 18 |
| 18.3 | | | | | | | | | 0.0008 | | | 0.0009 | 18.3 |
| 19 | 0.0599 | 0.0249 | 0.0227 | 0.0356 | | | | | | | | | 19 |
| 20 | 0.1957 | 0.1928 | 0.3799 | 0.3970 | | | | | | | | | 20 |
| 20.2 | | | | | | | | | | | 0.0007 | | 20.2 |
| 20.3 | | | | | | | | | | | | | 20.3 |
| 21 | 0.2134 | 0.1381 | 0.2794 | 0.2919 | | | | | | | | | 21 |
| 22 | 0.1105 | 0.0709 | 0.1514 | 0.1199 | | | | | | | | | 22 |
| 23 | 0.0437 | 0.0137 | 0.0764 | 0.0452 | | | | | | | | | 23 |
| 24 | 0.0054 | 0.0025 | 0.0200 | 0.0104 | | | | | | | | | 24 |
| 25 | 0.0008 | | 0.0007 | 0.0009 | | | | | | | | | 25 |
| 26 | | | | | | | | | | | | | 26 |
| 27 | | | | | | | | | | | | | 27 |
| 28 | | | | | | | | | | | | | 28 |
| 29 | | | | | | | | | | | | | 29 |
| 30 | | | | | | | | | | | | | 30 |
| 31 | | | | | | | | | | | | | 31 |
| 32 | | | | | | | | | | | | | 32 |
| 33 | | | | | | | | | | | | | 33 |
| PIC | 0.8102 | 0.7510 | 0.7083 | 0.6982 | 0.6588 | 0.5345 | 0.6275 | 0.5946 | 0.8198 | 0.7593 | 0.7562 | 0.7585 | PIC |
| H(exp) | 0.8318 | 0.7796 | 0.7456 | 0.7357 | 0.7078 | 0.5943 | 0.6912 | 0.656 | 0.8396 | 0.7880 | 0.7831 | 0.7858 | H(exp) |
| H(obs) | 0.8421 | 0.8200 | 0.7152 | 0.6973 | 0.7158 | 0.5967 | 0.6887 | 0.6784 | 0.8158 | 0.7500 | 0.7815 | 0.7568 | H(obs) |
| PDf | 0.9501 | 0.9229 | 0.8980 | 0.8927 | 0.8656 | 0.7756 | 0.8409 | 0.8202 | 0.9545 | 0.9263 | 0.9260 | 0.9269 | PDf |
| PDm | 0.8318 | 0.7796 | 0.7456 | 0.7357 | 0.7078 | 0.5943 | 0.6912 | 0.6560 | 0.8396 | 0.7880 | 0.7831 | 0.7858 | PDm |
| MECI | 0.8102 | 0.7510 | 0.7083 | 0.6982 | 0.6588 | 0.5345 | 0.6275 | 0.5946 | 0.8198 | 0.7593 | 0.7562 | 0.7585 | MECI |
| MECII | 0.6962 | 0.6217 | 0.5718 | 0.5609 | 0.5159 | 0.3868 | 0.4811 | 0.4468 | 0.7093 | 0.6316 | 0.6287 | 0.6314 | MECII |
| p(HWE) | 0.2180 | 0.3430 | 0.0612 | 0.4908 | 0.7937 | 0.7070 | 0.4085 | 0.9928 | 0.5319 | 0.3760 | 0.4767 | 0.0508 | p(HWE) |

Table continues on next page.

| | DXS101 | | | | DXS7424 | | | | HPRTB | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | **AA** | **AS** | **CN** | **Hisp** | **AA** | **AS** | **CN** | **Hisp** | **AA** | **AS** | **CN** | **Hisp** | |
| N | 1303 | 804 | 1453 | 1151 | 1303 | 804 | 1453 | 1151 | 1303 | 804 | 1453 | 1151 | N |
| 5 | | | | | | | | | | | | | 5 |
| 6 | | | | | | | | | | | | | 6 |
| 7 | | | | | | | | | | | 0.0007 | | 7 |
| 8 | | | | | | | | | 0.0015 | | 0.0007 | | 8 |
| 8.3 | | | | | | | | | | | | | 8.3 |
| 9 | | | | | 0.0015 | | 0.0007 | | 0.0376 | | 0.0048 | 0.0009 | 9 |
| 9.3 | | | | | | | | | | | | | 9.3 |
| 10 | | | | | 0.0107 | 0.0012 | 0.0041 | 0.0052 | 0.0161 | | 0.0069 | 0.0043 | 10 |
| 10.1 | | | | | | | | | | | | | 10.1 |
| 10.3 | | | | | | | | | | | | | 10.3 |
| 11 | | | | | 0.0683 | 0.0100 | 0.0055 | 0.0052 | 0.0921 | 0.0883 | 0.1225 | 0.0843 | 11 |
| 11.1 | | | | | | | | | | | | | 11.1 |
| 11.3 | | | | | | | | | | | | | 11.3 |
| 12 | | | | | 0.0660 | 0.0112 | 0.0413 | 0.0348 | 0.2901 | 0.2575 | 0.3193 | 0.2780 | 12 |
| 12.1 | | | | | | | | | | | | | 12.1 |
| 12.3 | | | | | | | | | | | | | 12.3 |
| 13 | | | | 0.0009 | 0.2201 | 0.0659 | 0.0681 | 0.1147 | 0.2609 | 0.4117 | 0.3310 | 0.3788 | 13 |
| 13.3 | | | | | | | | | | | | | 13.3 |
| 14 | 0.0008 | | | 0.0009 | 0.2477 | 0.1331 | 0.1996 | 0.1964 | 0.1965 | 0.1853 | 0.1521 | 0.1859 | 14 |
| 14.3 | | | | | | | | | | | | | 14.3 |
| 15 | 0.0015 | | 0.0303 | 0.0130 | 0.1794 | 0.3209 | 0.2650 | 0.2103 | 0.0898 | 0.0522 | 0.0482 | 0.0521 | 15 |
| 15.3 | | | | | | | | | | | | | 15.3 |
| 16 | 0.0031 | | 0.0021 | 0.0009 | 0.1503 | 0.3706 | 0.2650 | 0.2745 | 0.0153 | 0.0050 | 0.0131 | 0.0156 | 16 |
| 16.3 | | | | | | | | | | | | | 16.3 |
| 17 | 0.0031 | | 0.0055 | | 0.0429 | 0.0634 | 0.1211 | 0.1147 | | | 0.0007 | | 17 |
| 17.3 | | | | | | | | | | | | | 17.3 |
| 18 | 0.0476 | | 0.0770 | 0.0643 | 0.0107 | 0.0211 | 0.0234 | 0.0400 | | | | | 18 |
| 18.3 | | | | | | | | | | | | | 18.3 |
| 19 | 0.0913 | 0.0100 | 0.0509 | 0.0400 | 0.0015 | 0.0025 | 0.0055 | 0.0035 | | | | | 19 |
| 20 | 0.0606 | 0.0050 | 0.0206 | 0.0209 | 0.0008 | | 0.0007 | 0.0009 | | | | | 20 |
| 20.2 | | | | | | | | | | | | | 20.2 |
| 20.3 | | | | | | | | | | | | | 20.3 |
| 21 | 0.1497 | 0.0087 | 0.0289 | 0.0304 | | | | | | | | | 21 |
| 22 | 0.0675 | 0.0547 | 0.0213 | 0.0200 | | | | | | | | | 22 |
| 23 | 0.0683 | 0.1182 | 0.0695 | 0.0634 | | | | | | | | | 23 |
| 24 | 0.0944 | 0.2774 | 0.1933 | 0.2363 | | | | | | | | | 24 |
| 25 | 0.0875 | 0.2251 | 0.1768 | 0.1703 | | | | | | | | | 25 |
| 26 | 0.1036 | 0.1729 | 0.1286 | 0.2016 | | | | | | | | | 26 |
| 27 | 0.1274 | 0.0746 | 0.0908 | 0.0799 | | | | | | | | | 27 |
| 28 | 0.0507 | 0.0386 | 0.0591 | 0.0391 | | | | | | | | | 28 |
| 29 | 0.0253 | 0.0100 | 0.0261 | 0.0096 | | | | | | | | | 29 |
| 30 | 0.0123 | 0.0050 | 0.0158 | 0.0061 | | | | | | | | | 30 |
| 31 | 0.0031 | | 0.0034 | 0.0017 | | | | | | | | | 31 |
| 32 | 0.0008 | | | | | | | | | | | | 32 |
| 33 | 0.0015 | | | 0.0009 | | | | | | | | | 33 |
| PIC | 0.8998 | 0.7947 | 0.8760 | 0.8398 | 0.8014 | 0.6914 | 0.7690 | 0.7871 | 0.7601 | 0.6749 | 0.7073 | 0.6915 | PIC |
| H(exp) | 0.9072 | 0.8181 | 0.8861 | 0.8551 | 0.8245 | 0.7329 | 0.7981 | 0.8127 | 0.7906 | 0.7193 | 0.7478 | 0.7342 | H(exp) |
| H(obs) | 0.9263 | 0.8433 | 0.8631 | 0.8595 | 0.8500 | 0.7400 | 0.8411 | 0.8000 | 0.7842 | 0.7267 | 0.7991 | 0.7216 | H(obs) |
| PDf | 0.9839 | 0.9435 | 0.9769 | 0.9637 | 0.9461 | 0.8872 | 0.9301 | 0.9393 | 0.9257 | 0.8768 | 0.8959 | 0.8867 | PDf |
| PDm | 0.9072 | 0.8181 | 0.8861 | 0.8551 | 0.8245 | 0.7329 | 0.7981 | 0.8127 | 0.7906 | 0.7193 | 0.7478 | 0.7342 | PDm |
| MECI | 0.8998 | 0.7947 | 0.8760 | 0.8398 | 0.8014 | 0.6914 | 0.7690 | 0.7871 | 0.7601 | 0.6749 | 0.7073 | 0.6915 | MECI |
| MECII | 0.8244 | 0.6765 | 0.7891 | 0.7376 | 0.6847 | 0.5527 | 0.6435 | 0.6661 | 0.6326 | 0.5327 | 0.5698 | 0.5517 | MECII |
| p(HWE) | 0.3468 | **0.0003** | 0.0713 | 0.9938 | 0.5497 | 0.7182 | 0.1054 | 0.0325 | 0.7132 | 0.3569 | **0.0334** | 0.7700 | p(HWE) |

Table continues on next page.

| | DXS7423 | | | | DXS10147 | | | | GATA31E08 | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | **AA** | **AS** | **CN** | **Hisp** | **AA** | **AS** | **CN** | **Hisp** | **AA** | **AS** | **CN** | **Hisp** | |
| N | 1303 | 804 | 1453 | 1151 | 1301 | 804 | 1453 | 1151 | 1303 | 804 | 1453 | 1151 | N |
| 5 | | | | | 0.0008 | 0.0012 | | | | | | | 5 |
| 6 | | | | | 0.1276 | 0.2774 | 0.2326 | 0.3354 | 0.0008 | | | | 6 |
| 7 | | | | | 0.2905 | 0.1990 | 0.0420 | 0.0617 | 0.0207 | | | 0.0043 | 7 |
| 8 | 0.0077 | | | | 0.3566 | 0.4751 | 0.2904 | 0.4083 | 0.0276 | 0.0012 | 0.0014 | 0.0070 | 8 |
| 8.3 | | | | | | | | | | | | | 8.3 |
| 9 | | | | | 0.1806 | 0.0473 | 0.4178 | 0.1807 | 0.1527 | 0.0983 | 0.1886 | 0.1460 | 9 |
| 9.3 | | | | | | | | | | | | | 9.3 |
| 10 | | | | | 0.0392 | | 0.0158 | 0.0122 | 0.1466 | 0.0274 | 0.0227 | 0.0469 | 10 |
| 10.1 | | | | | | | | | | | | | 10.1 |
| 10.3 | | | | | | | | | | | | | 10.3 |
| 11 | | | 0.0007 | | 0.0038 | | 0.0014 | 0.0017 | 0.0691 | 0.1928 | 0.2154 | 0.1798 | 11 |
| 11.1 | | | | | | | | | | | | | 11.1 |
| 11.3 | | | | | | | | | | | | | 11.3 |
| 12 | 0.0069 | | | 0.0009 | | | | | 0.2717 | 0.2413 | 0.2065 | 0.3501 | 12 |
| 12.1 | | | | | | | | | | | | | 12.1 |
| 12.3 | | | | | | | | | | | | | 12.3 |
| 13 | 0.0936 | 0.0050 | 0.1053 | 0.0365 | 0.0008 | | | | 0.2256 | 0.3209 | 0.2443 | 0.2042 | 13 |
| 13.3 | | | | | | | | | | | | | 13.3 |
| 14 | 0.4728 | 0.3818 | 0.3228 | 0.2928 | | | | | 0.0721 | 0.1045 | 0.1081 | 0.0504 | 14 |
| 14.3 | | | | | | | | | | | | | 14.3 |
| 15 | 0.3170 | 0.5597 | 0.3833 | 0.4831 | | | | | 0.0123 | 0.0124 | 0.0117 | 0.0113 | 15 |
| 15.3 | | | | | | | | | | | | | 15.3 |
| 16 | 0.0906 | 0.0522 | 0.1514 | 0.1017 | | | | | 0.0008 | 0.0012 | 0.0007 | | 16 |
| 16.3 | | | | | | | | | | | | | 16.3 |
| 17 | 0.0115 | 0.0012 | 0.0358 | 0.0851 | | | | | | | 0.0007 | | 17 |
| 17.3 | | | | | | | | | | | | | 17.3 |
| 18 | | | 0.0007 | | | | | | | | | | 18 |
| 18.3 | | | | | | | | | | | | | 18.3 |
| 19 | | | | | | | | | | | | | 19 |
| 20 | | | | | | | | | | | | | 20 |
| 20.2 | | | | | | | | | | | | | 20.2 |
| 20.3 | | | | | | | | | | | | | 20.3 |
| 21 | | | | | | | | | | | | | 21 |
| 22 | | | | | | | | | | | | | 22 |
| 23 | | | | | | | | | | | | | 23 |
| 24 | | | | | | | | | | | | | 24 |
| 25 | | | | | | | | | | | | | 25 |
| 26 | | | | | | | | | | | | | 26 |
| 27 | | | | | | | | | | | | | 27 |
| 28 | | | | | | | | | | | | | 28 |
| 29 | | | | | | | | | | | | | 29 |
| 30 | | | | | | | | | | | | | 30 |
| 31 | | | | | | | | | | | | | 31 |
| 32 | | | | | | | | | | | | | 32 |
| 33 | | | | | | | | | | | | | 33 |
| PIC | 0.6026 | 0.4444 | 0.6647 | 0.6095 | 0.6939 | 0.5952 | 0.6263 | 0.6259 | 0.7959 | 0.7474 | 0.7737 | 0.7462 | PIC |
| H(exp) | 0.6589 | 0.5382 | 0.7136 | 0.6619 | 0.7379 | 0.6555 | 0.6850 | 0.6841 | 0.8194 | 0.7801 | 0.8034 | 0.7775 | H(exp) |
| H(obs) | 0.7053 | 0.5233 | 0.6799 | 0.6892 | 0.7474 | 0.6400 | 0.6578 | 0.6919 | 0.8263 | 0.7633 | 0.8190 | 0.7703 | H(obs) |
| PDf | 0.8274 | 0.6929 | 0.8690 | 0.8333 | 0.8873 | 0.8211 | 0.8421 | 0.8420 | 0.9438 | 0.9189 | 0.9316 | 0.9192 | PDf |
| PDm | 0.6589 | 0.5382 | 0.7136 | 0.6619 | 0.7379 | 0.6555 | 0.6850 | 0.6841 | 0.8194 | 0.7801 | 0.8034 | 0.7775 | PDm |
| MECI | 0.6026 | 0.4444 | 0.6647 | 0.6095 | 0.6939 | 0.5952 | 0.6263 | 0.6259 | 0.7959 | 0.7474 | 0.7737 | 0.7462 | MECI |
| MECII | 0.4567 | 0.3076 | 0.5218 | 0.4633 | 0.5538 | 0.4476 | 0.4801 | 0.4802 | 0.6777 | 0.6166 | 0.6481 | 0.6154 | MECII |
| p(HWE) | 0.9261 | 0.4901 | **0.0310** | 0.8342 | 0.1358 | 0.4224 | 0.3669 | 0.2966 | 0.3421 | 0.6962 | 0.5607 | 0.8702 | p(HWE) |

Overall, 177 unique alleles were observed across 15 markers in all four population groups, with a mean value of 12 alleles per marker. The lowest number of unique alleles was observed at marker GATA165B12 while the largest occurred at DXS101, which contains the only compound repeat region composed of two variable

trinucleotide stretches in this set of markers (Table 3.5). Since the major mechanism responsible for microsatellite variation is thought to be strand slippage [194] which, in the form of stutter, is more often observed for trinucleotide repeats than tetranucleotide repeats (see, for example, [300]), it follows that the high degree of polymorphism observed at DXS101 might be related to its repeat structure. See Chapter 5 for additional discussion of strand slippage and STR mutation rate.

The forensic efficiency statistics indicate the relative usefulness of the chosen set of markers in the U.S. populations studied. The observed heterozygosity values ranged from 0.5233 for DXS7423 in the U.S. Asian population to 0.9263 at DXS101 in the African American population. Despite occasional values lower than originally targeted (> 0.65) at certain markers within individual populations, all markers exhibited observed heterozygosity values higher than 0.70 in at least one population group, and 11 or more markers in each population group showed values greater than 0.65. The studied markers, then, are likely to be especially informative in these U.S. populations when applied in combination. Indeed, a comparison of PIC values confirms this same trend. While the highest PIC values in all studied populations occurred for marker DXS101 as expected, the relative value of the other 14 markers varied by population. The lowest PIC values were observed for DXS7423 in the African American (0.6026) and U.S. Asian (0.4444) populations while DXS6795 and GATA165B12 exhibited the lowest values within the U.S. Caucasian (0.6083) and U.S. Hispanic (0.5946) populations respectively. The mean PIC value for all populations was greater then 0.65. Additionally, there is at least one marker within each of the four proposed linkage groups (see Chapter 5 for more on linkage groups) with a PIC value greater than 0.64 in all four populations. When used in combination, these 15 markers provide a highly discriminating system for use with U.S. populations.

All but three marker-population combinations were found to be in Hardy-Weinberg equilibrium in the combined dataset: the U.S. Asian population at DXS101 ($p = 0.003$), and the U.S. Caucasian population at both HPRTB ($p = 0.0334$) and DXS7423 ($p = 0.0310$). As mentioned previously, Hardy-Weinberg equilibrium describes an ideal state against which sampled populations can be compared, and incorporates assumptions about the population itself. One such assumption is that

there is no migration, mutation or selection, which is rarely accurate when describing real populations. Given that the vast majority of marker-population combinations exhibited no significant deviations from Hardy-Weinberg equilibrium, it is likely that these populations represent a robust, unbiased sampling suitable for forensic use.

Though it has been established both within the analyses of the individual sample sets examined here as well as in published studies [55,70] that the four U.S. population groups differ significantly from each other with respect to X STR profiles, pairwise $F_{ST}$ and corresponding p values were calculated at every marker for confirmation (Table 4.23). In only two cases (out of 90) were the p values indicative of populations that are not significantly different from one another: for the African American and U.S. Hispanic populations at marker DXS7132 ($p = 0.7356$) and for the U.S. Asian and U.S. Hispanic populations at marker HPRTB ($p = 0.3650$). All other pairs of populations at every marker were found to be significantly different from one another, supporting the separation of the U.S. population into these four individual groups for forensic genetic use.

**Table 4.23. Pairwise $F_{ST}$ values (below diagonal) and corresponding p values with standard deviations (above diagonal) comparing populations within the combined U.S. population dataset by marker.** P values indicative of populations that are not significantly different from one another ($p > 0.5$) are bolded. AA: African American; AS: U.S. Asian; CN: U.S. Caucasian, Hisp: U.S. Hispanic.

Table begins on next page.

| Marker | Population | AA | AS | CN | Hisp |
|---|---|---|---|---|---|
| DXS101 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | AS | 0.05058 | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.02189 | 0.01343 | * | 0.00000±0.0000 |
| | Hisp | 0.03204 | 0.00825 | 0.00418 | * |
| DXS6789 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | AS | 0.04531 | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.05161 | 0.11548 | * | 0.02891±0.0015 |
| | Hisp | 0.05264 | 0.10919 | 0.00145 | * |
| DXS6795 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | AS | 0.10872 | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.1361 | 0.10848 | * | 0.00000±0.0000 |
| | Hisp | 0.07334 | 0.02996 | 0.06652 | * |
| DXS6803 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | AS | 0.11371 | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.0254 | 0.07559 | * | 0.00000±0.0000 |
| | Hisp | 0.02429 | 0.07086 | 0.00857 | * |
| DXS7130 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | AS | 0.03158 | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.03577 | 0.03733 | * | 0.00000±0.0000 |
| | Hisp | 0.01689 | 0.02198 | 0.01117 | * |
| DXS7132 | AA | * | 0.00000±0.0000 | 0.00515±0.0008 | **0.73557±0.0043** |
| | AS | 0.00638 | * | 0.00000±0.0000 | 0.00040±0.0002 |
| | CN | 0.00231 | 0.01609 | * | 0.00198±0.0004 |
| | Hisp | -0.00044 | 0.00593 | 0.00298 | * |
| DXS7423 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | AS | 0.05772 | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.02152 | 0.04008 | * | 0.00000±0.0000 |
| | Hisp | 0.04849 | 0.01835 | 0.01385 | * |
| DXS7424 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | AS | 0.06553 | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.03303 | 0.01367 | * | 0.00139±0.0003 |
| | Hisp | 0.02358 | 0.01872 | 0.00278 | * |
| DXS8378 | AA | * | 0.00000±0.0000 | 0.00059±0.0002 | 0.00000±0.0000 |
| | AS | 0.08639 | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.00434 | 0.05168 | * | 0.00000±0.0000 |
| | Hisp | 0.02226 | 0.02231 | 0.00596 | * |
| DXS9902 | AA | * | 0.00000±0.0000 | 0.00842±0.0009 | 0.00000±0.0000 |
| | AS | 0.01264 | * | 0.00000±0.0000 | 0.00505±0.0007 |
| | CN | 0.00239 | 0.00729 | * | 0.01247±0.0012 |
| | Hisp | 0.00781 | 0.00444 | 0.00236 | * |
| DXS10147 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | AS | 0.0426 | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.08574 | 0.12732 | * | 0.00000±0.0000 |
| | Hisp | 0.06463 | 0.03123 | 0.05529 | * |
| GATA31E08 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | AS | 0.02592 | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.02618 | 0.00887 | * | 0.00000±0.0000 |
| | Hisp | 0.01742 | 0.01844 | 0.01723 | * |
| GATA165B12 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | AS | 0.09166 | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.03031 | 0.05975 | * | 0.00000±0.0000 |
| | Hisp | 0.03937 | 0.01392 | 0.02109 | * |
| GATA172D05 | AA | * | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | AS | 0.07632 | * | 0.00000±0.0000 | 0.00000±0.0000 |
| | CN | 0.05584 | 0.01566 | * | 0.00000±0.0000 |
| | Hisp | 0.06577 | 0.01118 | 0.00411 | * |
| HPRTB | AA | * | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | AS | 0.01634 | * | 0.00000±0.0000 | **0.36501±0.0045** |
| | CN | 0.0067 | 0.00762 | * | 0.00030±0.0002 |
| | Hisp | 0.01035 | 0.00002 | 0.00377 | * |

Lastly, as an additional quality control measure, this combined U.S. dataset was compared to similar published populations at overlapping markers in the same manner as was performed for each individual sample set. P values corresponding to pairwise $F_{ST}$ values for each comparison are shown in Table 4.24. Of the 27 comparisons between similar populations, only 3 (11%) had p values indicating the published populations were actually significantly different from this combined population: the U.S. Asians at marker DXS101 and the U.S. Hispanics at markers DXS7423 and GATA172D05, both populations from Ref. [70]. Similar populations from two additional publications were found to be similar to the combined dataset at two overlapping markers. Comparisons between unmatched populations, however, revealed 19 (23%) of 81 pairs that were not significantly different from one another. As encountered through analyses of the individual sample sets, most of the population pairs show similarities at marker DXS7132. Additionally, the African American and U.S. Hispanic populations are often found to lack significant differences from other population groups. These results are consistent with previous findings, and taken together, verify the suitability of these populations.

**Table 4.24. P values (with standard deviations) corresponding to pairwise $F_{ST}$ values (not shown) comparing the combined U.S. populations (top) to similar published populations (left) at overlapping markers.** P values indicative of populations that are not significantly different from one another ($p > 0.5$) are bolded. P values for comparisons between equivalent populations are in gray shaded boxes. AA: African American; AS: U.S. Asian; CN: U.S. Caucasian, Hisp: U.S. Hispanic.

| Marker | Ref. | Population | AA | AS | CN | Hisp |
|---|---|---|---|---|---|---|
| DXS101 | [70] | AA | **0.22671±0.0040** | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | | AS | 0.00000±0.0000 | 0.01554±0.0012 | 0.00010±0.0001 | 0.00119±0.0004 |
| | | Hisp | 0.00000±0.0000 | 0.00109±0.0003 | **0.49035±0.0054** | **0.15751±0.0033** |
| | [283] | AA | **0.12524±0.0031** | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | | CN | 0.00000±0.0000 | 0.01079±0.0011 | **0.11316±0.0031** | **0.27542±0.0042** |
| DXS6789 | [70] | AA | **0.36383±0.0050** | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | | AS | 0.00000±0.0000 | **0.42600±0.0049** | 0.00000±0.0000 | 0.00000±0.0000 |
| | | Hisp | 0.00000±0.0000 | 0.00000±0.0000 | **0.06752±0.0027** | **0.10652±0.0027** |
| DXS7132 | [70] | AA | **0.56173±0.0053** | 0.03356±0.0018 | **0.79596±0.0040** | **0.42511±0.0049** |
| | | AS | **0.34729±0.0053** | **0.68320±0.0045** | **0.05940±0.0018** | **0.50728±0.0047** |
| | | Hisp | **0.12910±0.0038** | **0.25898±0.0043** | 0.01792±0.0013 | **0.08613±0.0031** |
| DXS7423 | [70] | AA | **0.17177±0.0036** | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | | AS | 0.00020±0.0001 | **0.47956±0.0044** | 0.00000±0.0000 | 0.00317±0.0007 |
| | | Hisp | **0.06772±0.0025** | 0.00109±0.0003 | **0.08672±0.0028** | 0.00762±0.0007 |
| DXS8378 | [70] | AA | **0.31086±0.0042** | 0.00000±0.0000 | 0.00792±0.0008 | 0.00000±0.0000 |
| | | AS | 0.00000±0.0000 | **0.08841±0.0026** | 0.00158±0.0004 | **0.08484±0.0030** |
| | | Hisp | **0.07979±0.0029** | 0.00000±0.0000 | **0.35105±0.0048** | **0.08623±0.0031** |
| GATA172D05 | [70] | AA | **0.23750±0.0045** | 0.00000±0.0000 | 0.00000±0.0000 | 0.00000±0.0000 |
| | | AS | 0.00000±0.0000 | **0.47273±0.0042** | 0.00050±0.0002 | 0.01188±0.0009 |
| | | Hisp | 0.00020±0.0001 | 0.00000±0.0000 | 0.00505±0.0006 | 0.00158±0.0004 |
| HPRTB | [70] | AA | **0.70874±0.0046** | 0.00040±0.0002 | 0.01089±0.0012 | 0.00366±0.0006 |
| | | AS | 0.00050±0.0002 | **0.78062±0.0043** | 0.03168±0.0015 | **0.40026±0.0050** |
| | | Hisp | **0.41877±0.0047** | **0.17681±0.0038** | **0.32126±0.0046** | **0.43313±0.0045** |
| | [292] | AA | **0.10969±0.0031** | 0.00317±0.0006 | 0.00129±0.0004 | 0.01386±0.0012 |
| | | AS | 0.00000±0.0000 | **0.08276±0.0025** | 0.00020±0.0001 | 0.01535±0.0011 |
| | | CN | 0.00277±0.0005 | 0.00000±0.0000 | **0.45283±0.0049** | 0.00020±0.0001 |
| | | Hisp | 0.00812±0.0009 | **0.29750±0.0045** | **0.07742±0.0025** | **0.68993±0.0041** |

Because the markers reside on a single chromosome, the exact test for multi-locus linkage disequilibrium was performed using female profiles and resultant p values indicating a significant association between markers are given in Table 4.25. In general, the pairs associated statistically correspond to pairs that are physically close together on the chromosome and belong to the same proposed linkage group. Four exceptions were noted, however (italicized pairs in Table 4.25). The markers in each of these four pairs are neither physically adjacent to one another on the chromosome nor hypothesized to belong to the same linkage group, and may not represent true

associations but rather population sampling effects or substructure. Either way additional study to understand the extent of both linkage and linkage disequilibrium between this set of markers is necessary.

**Table 4.25. P values indicating significance ($p < 0.05$) for the exact test for multi-locus linkage disequilibrium within the combined dataset.** Marker pairs that do not belong to the same proposed linkage group and are not physically close to one another are shown in italics.

| Locus combination | P Value |
|---|---|
| DXS6795-DXS9902 | 0.0120 |
| *DXS9902-GATA172D05* | *0.0000* |
| DXS6803-DXS6789 | 0.0140 |
| DXS6803-GATA165B12 | 0.0250 |
| DXS6789-GATA165B12 | 0.0000 |
| DXS6789-DXS7130 | 0.0000 |
| GATA165B12-DXS7130 | 0.0090 |
| GATA165B12-DXS101 | 0.0220 |
| DXS7130-DXS101 | 0.0030 |
| DXS101-DXS7424 | 0.0020 |
| *DXS7424-DXS7423* | *0.0330* |
| *HPRTB-DXS7423* | *0.0460* |
| *HPRTB-DXS10147* | *0.0040* |
| DXS7423-DXS10147 | 0.0000 |
| DXS10147-GATA31E08 | 0.0000 |

For reference, male haplotypes for each of the four proposed linkage groups are provided in Appendix B, Tables B10-B12 (excluding linkage group 3, which is represented by HPRTB alone in this set of markers). The number of individual haplotypes observed for proposed linkage groups 1 and 4 are generally similar to each other (173 and 162 respectively) and to the individual samples sets A, B, and C (102 to 128). These data suggest that this combined population is capturing the vast majority of common haplotypes for these marker combinations; typing of additional samples will have diminishing returns. The diversity of haplotypes from proposed linkage group 2, however, increased proportionally with the population size. There were 1698 individual haplotypes observed in 1701 male samples, with only three of these haplotypes observed more than once. Additional profiles will be necessary before a true picture of the haplotype diversity possible with this set of 8 markers can be assessed. Equally important, however, is understanding whether the 8 markers are actually physically and/or genetically linked to one another and unlinked to the

remaining markers in this set, a question that cannot be answered with additional population sampling.

*4.2. Bosnian & Herzegovinian Population Database*

A population from Bosnia and Herzegovina was studied to determine allele frequencies and appropriate forensic efficiency parameters for 14 commonly-used X STR markers using the original version of the multiplexes developed here (see Chapter 3 for details). Profiles from a total of 154 (68 female and 86 male) unrelated individuals living in Bosnia and Herzegovina were analyzed as part of this study. The chi-square test for independence was used to examine the distribution of allele frequencies in the male and female donors (Table 4.26). The resultant p values revealed only one significant differences ($p = 0.0299$) at marker DXS7423. Given the small population size as well as previous results observed for U.S. populations, this outlier was disregarded and frequency data was pooled at each locus.

**Table 4.26. P values from chi-square test comparing distribution of male and female allele frequencies from a Bosnian & Herzegovinian population by marker.** P values indicative of a significant difference (< 0.05) are bolded.

| Marker | P value |
|---|---|
| DXS8378 | 0.9729 |
| DXS9902 | 0.1974 |
| DXS7424 | 0.3056 |
| DXS6789 | 0.1751 |
| DXS7130 | 0.1390 |
| GATA165B12 | 0.4707 |
| DXS101 | 0.3634 |
| GATA172D05 | 0.4055 |
| DXS7132 | 0.6609 |
| DXS6803 | 0.9425 |
| DXS7133 | 0.4495 |
| HPRTB | 0.4938 |
| GATA31E08 | 0.2404 |
| DXS7423 | **0.0299** |

Allele frequencies and forensic efficiency parameters calculated for the 14 X STR markers are presented in Table 4.27.

**Table 4.27. Allele frequencies and summary statistics for 14 X STR markers in a population from Bosnia and Herzegovina.** H(exp): expected heterozygosity, H(obs): observed heterozygosity, PIC: polymorphism information content, PDf: power of discrimination in females, PDm: power of discrimination in males, MECI: mean exclusion chance in trios involving daughters, MECII: mean exclusion chance in father/daughter duos, p (HWE): p value of the exact test for Hardy-Weinberg equilibrium. Bold values are p values for the exact test that are less than 0.05, indicating departure from Hardy-Weinberg equilibrium.

| Allele | DXS8378 | DXS9902 | DXS7424 | DXS6789 | DXS7130 | GATA165B12 | DXS101 | Allele |
|---|---|---|---|---|---|---|---|---|
| **6** | | | | | | | | **6** |
| **7** | | | | | | | | **7** |
| **8** | | | | | | | | **8** |
| **9** | 0.0541 | 0.0450 | | | | 0.2973 | | **9** |
| **10** | 0.3288 | 0.3784 | 0.0090 | | | 0.3018 | | **10** |
| **10.1** | | 0.0045 | | | | | | **10.1** |
| **11** | 0.3333 | 0.3153 | | | 0.0495 | 0.3604 | | **11** |
| **11.1** | | 0.0405 | | | | | | **11.1** |
| **11.3** | | | | | | | | **11.3** |
| **12** | 0.2568 | 0.2027 | 0.0090 | | 0.0946 | 0.0405 | | **12** |
| **12.1** | | 0.0090 | | | | | | **12.1** |
| **12.3** | | | | | | | | **12.3** |
| **13** | 0.0270 | 0.0045 | 0.0901 | | 0.0270 | | | **13** |
| **13.3** | | | | | 0.0450 | | | **13.3** |
| **14** | | | 0.2297 | | 0.0270 | | | **14** |
| **14.3** | | | | | 0.1757 | | | **14.3** |
| **15** | | | 0.2568 | 0.0676 | | | 0.0450 | **15** |
| **15.3** | | | | | 0.3964 | | | **15.3** |
| **16** | | | 0.2703 | | | | 0.0135 | **16** |
| **16.3** | | | | | 0.1712 | | | **16.3** |
| **17** | | | 0.1171 | | | | | **17** |
| **17.3** | | | | | 0.0135 | | | **17.3** |
| **18** | | | 0.0135 | | | | 0.0631 | **18** |
| **19** | | | 0.0045 | 0.0135 | | | 0.0450 | **19** |
| **20** | | | | 0.4144 | | | 0.0135 | **20** |
| **21** | | | | 0.2477 | | | 0.0270 | **21** |
| **22** | | | | 0.2342 | | | 0.0045 | **22** |
| **23** | | | | 0.0180 | | | 0.0631 | **23** |
| **24** | | | | 0.0045 | | | 0.2568 | **24** |
| **25** | | | | | | | 0.1577 | **25** |
| **26** | | | | | | | 0.1216 | **26** |
| **27** | | | | | | | 0.0586 | **27** |
| **28** | | | | | | | 0.1126 | **28** |
| **29** | | | | | | | 0.0045 | **29** |
| **30** | | | | | | | 0.0090 | **30** |
| **31** | | | | | | | 0.0045 | **31** |
| **H(exp)** | 0.7112 | 0.7125 | 0.7860 | 0.7070 | 0.7676 | 0.6890 | 0.8650 | **H(exp)** |
| **H(obs)** | 0.7353 | 0.7647 | 0.8088 | 0.5882 | 0.7206 | 0.6765 | 0.8676 | **H(obs)** |
| **PIC** | 0.6562 | 0.6620 | 0.7530 | 0.6574 | 0.7402 | 0.6253 | 0.8522 | **PIC** |
| **PDf** | 0.8616 | 0.8668 | 0.9212 | 0.8646 | 0.9186 | 0.8395 | 0.9690 | **PDf** |
| **PDm** | 0.7112 | 0.7125 | 0.7860 | 0.7070 | 0.7676 | 0.6890 | 0.8650 | **PDm** |
| **MECI** | 0.6562 | 0.6620 | 0.7530 | 0.6574 | 0.7402 | 0.6253 | 0.8522 | **MECI** |
| **MECII** | 0.5121 | 0.5191 | 0.6232 | 0.5135 | 0.6091 | 0.4786 | 0.7551 | **MECII** |
| **p (HWE)** | 0.7793 | 0.7268 | **0.0291** | **0.0248** | 0.2664 | 0.6202 | 0.4761 | **p (HWE)** |

Table continues on next page.

| Allele | GATA172D05 | DXS7132 | DXS6803 | DXS7133 | HPRTB | GATA31E08 | DXS7423 | Allele |
|---|---|---|---|---|---|---|---|---|
| **6** | 0.1532 | | | 0.0045 | | | | **6** |
| **7** | | | | 0.0090 | 0.0090 | | | **7** |
| **8** | 0.1667 | | | | 0.0090 | | | **8** |
| **9** | 0.0450 | | 0.0045 | 0.3964 | 0.0270 | 0.2117 | | **9** |
| **10** | 0.3288 | | 0.0405 | 0.1712 | 0.0045 | 0.0405 | | **10** |
| **10.1** | | | | | | | | **10.1** |
| **11** | 0.2207 | 0.0090 | 0.2523 | 0.3333 | 0.1396 | 0.1892 | | **11** |
| **11.1** | | | | | | | | **11.1** |
| **11.3** | | | 0.0135 | | | | | **11.3** |
| **12** | 0.0856 | 0.0811 | 0.2297 | 0.0586 | 0.2973 | 0.1667 | | **12** |
| **12.1** | | | | | | | | **12.1** |
| **12.3** | | | 0.1441 | | | | | **12.3** |
| **13** | | 0.3153 | 0.1486 | 0.0270 | 0.3514 | 0.2838 | 0.0856 | **13** |
| **13.3** | | | 0.1441 | | | | | **13.3** |
| **14** | | 0.4054 | 0.0090 | | 0.1171 | 0.0991 | 0.3243 | **14** |
| **14.3** | | | 0.0135 | | | | | **14.3** |
| **15** | | 0.1486 | | | 0.0405 | 0.0090 | 0.3964 | **15** |
| **15.3** | | | | | | | | **15.3** |
| **16** | | 0.0180 | | | 0.0045 | | 0.1802 | **16** |
| **16.3** | | | | | | | | **16.3** |
| **17** | | 0.0225 | | | | | 0.0135 | **17** |
| **17.3** | | | | | | | | **17.3** |
| **18** | | | | | | | | **18** |
| **19** | | | | | | | | **19** |
| **20** | | | | | | | | **20** |
| **21** | | | | | | | | **21** |
| **22** | | | | | | | | **22** |
| **23** | | | | | | | | **23** |
| **24** | | | | | | | | **24** |
| **25** | | | | | | | | **25** |
| **26** | | | | | | | | **26** |
| **27** | | | | | | | | **27** |
| **28** | | | | | | | | **28** |
| **29** | | | | | | | | **29** |
| **30** | | | | | | | | **30** |
| **31** | | | | | | | | **31** |
| **H(exp)** | 0.7826 | 0.7067 | 0.8179 | 0.6982 | 0.7524 | 0.7995 | 0.6977 | **H(exp)** |
| **H(obs)** | 0.7794 | 0.7647 | 0.8088 | 0.7206 | 0.7794 | 0.8235 | 0.7059 | **H(obs)** |
| **PIC** | 0.7507 | 0.6580 | 0.7929 | 0.6450 | 0.7147 | 0.7700 | 0.6432 | **PIC** |
| **PDf** | 0.9209 | 0.8653 | 0.9418 | 0.8557 | 0.9010 | 0.9303 | 0.8541 | **PDf** |
| **PDm** | 0.7826 | 0.7067 | 0.8179 | 0.6982 | 0.7524 | 0.7995 | 0.6977 | **PDm** |
| **MECI** | 0.7507 | 0.6580 | 0.7929 | 0.6450 | 0.7147 | 0.7700 | 0.6432 | **MECI** |
| **MECII** | 0.6204 | 0.5151 | 0.6732 | 0.5010 | 0.5788 | 0.6438 | 0.4983 | **MECII** |
| **p (HWE)** | 0.4936 | 0.4618 | 0.3779 | 0.9174 | 0.2490 | 0.1055 | 0.9458 | **p (HWE)** |

In total, 110 different alleles were observed across 14 markers, with 4 to16 alleles per marker. The highest observed heterozygosity value was noted at marker DXS101 (0.8676), which also exhibited the most alleles (16), while the lowest was observed at DXS6789 (0.5882). All markers possessed high forensic efficiency values with the studied population sample, supporting the utility of the multiplexes for forensic purposes.

Two markers (DXS7424 and DXS6789) showed a departure from Hardy-Weinberg equilibrium, indicated by a p value for the exact test that is less than 0.05 (bold values in Table 4.27). The observed deviations could be due to the small population size used to evaluate Hardy-Weinberg equilibrium (68 females). After applying the Bonferroni correction, however, none of the values remained significant.

Analysis of pairwise linkage disequilibrium using only the female profiles revealed marginally significant results ($p < 0.05$) in the studied population for seven pairs of markers: GATA172D05-GATA31E08, DXS7132-DXS7423, DXS9902-HPRTB, DXS7130-DXS6803, DXS6789-GATA172D05, DXS8378-GATA172D05, and DXS7424-DXS7130. Only one pair showed a significant p value less than 0.01 (DXS8378-GATA165B12). While it is possible that the p values for these pairings indicate true linkage, the population tested was relatively small (68 females) and analyses may be skewed by non-random sampling or substructure. For reference, male haplotype data according to the proposed linkage groups [209,271] are presented in Appendix B, Tables B13-B15. A count of the number of observed haplotypes was performed for groups 1, 2, and 4 (group 3 is represented by HPRTB alone in these multiplexes). There were 20 unique haplotypes for linkage group 1 (DXS8378 and DXS9902) and 24 for linkage group 4 (GATA31E08 and DXS7423) while every haplotype was unique across linkage group 2. The most common haplotype (DXS8378-10, DXS9902-10) was observed 14 times.

The data from this population were compared with six other populations representing various regions of Europe: Hungary [165] and Latvia [115] (eastern Europe); Italy [86,99] and Germany [200,246,273] (central Europe); and Portugal [83,124] and Spain [83,94] (western Europe). The allele frequency distributions of overlapping

markers were assessed using the chi-square test and significant differences ($p < 0.05$) were observed for several markers in several populations: DXS9902 and DXS7130 in Germans, HPRTB and DXS7133 in Latvians, and DXS8378 in Northern Portuguese. Because of the relatively small sample size studied here, however, further investigation would be necessary to determine if these populations truly differ at these markers. In general, the allelic distribution of this set of X STR markers in the population from Bosnia and Herzogovina is similar to that observed in other populations across Europe.

This study represents the first study of a population from Bosnia and Herzegovina using 14 X STR markers.

# Chapter 5. Mutation Rate & Recombination Rate

A combination of two- and three-generation families was used to determine both the mutation rate and recombination rate for this set of 15 X STR markers. Both parameters are required to maximize their utility and accurately assign weight to comparisons between resulting profiles.

## *5.1. Mutation rate study*

The results presented here are part of a manuscript accepted for publication in the *International Journal of Legal Medicine*, and has been formatted accordingly. Table, figure, and section numbers have been modified for inclusion in this body of work.

### *5.1.1. Overall mutation rates*

A total of 20625 meioses in confirmed family trios or duos were analyzed at 15 X STR markers (DXS6789, DXS7130, GATA31E08, GATA165B12, GATA172D05, DXS10147, DXS8378, DXS7132, DXS6803, HPRTB, DXS7423, and DXS9902, DXS7424, DXS101, and DXS6795) and eighteen mutations were observed across 7 of the 15 markers and in all three U.S. population groups, resulting in an overall mutation rate of $8.73 \times 10^{-4}$ (95% CI: $0.52\text{-}1.4 \times 10^{-3}$; Table 5.1). The probability of paternity or maternity for the families showing mutation(s) were all $\geq 99.99\%$ and allele sequencing results confirmed profiles (data not shown). The overall mutation rate for X STR markers observed in this study was similar to that reported for autosomal STRs [301-304] and Y STRs [196-199,305-307]. Since the mechanism of mutation is likely the same for each system, this result was expected.

**Table 5.1.A-C. X STR mutation rates from this study (A & B), a literature summary (C), and the combined datasets (C).**

**A.**

| | African American | | | | U.S. Caucasian | | | |
|---|---|---|---|---|---|---|---|---|
| **Marker** | **Mutations** | **Meioses** | **Mutation rate ($x10^{-3}$)** | **95% CI ($x10^{-3}$)** | **Mutations** | **Meioses** | **Mutation rate ($x10^{-3}$)** | **95% CI ($x10^{-3}$)** |
| DXS8378 | 0 | 390 | 0.00 | 0-9.4 | 3 | 533 | 5.63 | 1.2-16.4 |
| DXS9902 | 0 | 390 | 0.00 | 0-9.4 | 1 | 533 | 1.88 | 0.05-10.4 |
| DXS6795 | 0 | 390 | 0.00 | 0-9.4 | 0 | 533 | 0.00 | 0-6.9 |
| DXS7132 | 1 | 390 | 2.56 | 0.06-14.2 | 3 | 533 | 5.63 | 1.2-16.4 |
| DXS6803 | 0 | 390 | 0.00 | 0-9.4 | 0 | 533 | 0.00 | 0-6.9 |
| DXS6789 | 0 | 390 | 0.00 | 0-9.4 | 0 | 533 | 0.00 | 0-6.9 |
| DXS7424 | 0 | 390 | 0.00 | 0-9.4 | 0 | 533 | 0.00 | 0-6.9 |
| DXS101 | 0 | 390 | 0.00 | 0-9.4 | 0 | 533 | 0.00 | 0-6.9 |
| GATA172D05 | 0 | 390 | 0.00 | 0-9.4 | 1 | 533 | 1.88 | 0.05-10.4 |
| DXS7130 | 0 | 390 | 0.00 | 0-9.4 | 0 | 533 | 0.00 | 0-6.9 |
| GATA165B12 | 0 | 390 | 0.00 | 0-9.4 | 0 | 533 | 0.00 | 0-6.9 |
| HPRTB | 0 | 390 | 0.00 | 0-9.4 | 1 | 533 | 1.88 | 0.05-10.4 |
| GATA31E08 | 0 | 390 | 0.00 | 0-9.4 | 0 | 533 | 0.00 | 0-6.9 |
| DXS10147 | 0 | 390 | 0.00 | 0-9.4 | 0 | 533 | 0.00 | 0-6.9 |
| DXS7423 | 0 | 390 | 0.00 | 0-9.4 | 0 | 533 | 0.00 | 0-6.9 |
| **Overall** | **1** | **5850** | **0.17** | **0.004-0.95** | **9** | **7995** | **1.13** | **0.51-2.1** |

**B.**

| | U.S. Hispanic | | | | This study (U.S. total) | | | |
|---|---|---|---|---|---|---|---|---|
| **Marker** | **Mutations** | **Meioses** | **Mutation rate ($x10^{-3}$)** | **95% CI ($x10^{-3}$)** | **Mutations** | **Meioses** | **Mutation rate ($x10^{-3}$)** | **95% CI ($x10^{-3}$)** |
| DXS8378 | 1 | 452 | 2.21 | 0.06-12.3 | 4 | 1375 | 2.91 | 0.79-7.4 |
| DXS9902 | 3 | 452 | 6.64 | 1.4-19.3 | 4 | 1375 | 2.91 | 0.79-7.4 |
| DXS6795 | 0 | 452 | 0.00 | 0-8.1 | 0 | 1375 | 0.00 | 0-2.7 |
| DXS7132 | 0 | 452 | 0.00 | 0-8.1 | 4 | 1375 | 2.91 | 0.79-7.4 |
| DXS6803 | 1 | 452 | 2.21 | 0.06-12.3 | 1 | 1375 | 0.73 | 0.02-4.1 |
| DXS6789 | 2 | 452 | 4.42 | 0.54-15.9 | 2 | 1375 | 1.45 | 0.18-5.2 |
| DXS7424 | 0 | 452 | 0.00 | 0-8.1 | 0 | 1375 | 0.00 | 0-2.7 |
| DXS101 | 0 | 452 | 0.00 | 0-8.1 | 0 | 1375 | 0.00 | 0-2.7 |
| GATA172D05 | 0 | 452 | 0.00 | 0-8.1 | 1 | 1375 | 0.73 | 0.02-4.1 |
| DXS7130 | 0 | 452 | 0.00 | 0-8.1 | 0 | 1375 | 0.00 | 0-2.7 |
| GATA165B12 | 0 | 452 | 0.00 | 0-8.1 | 0 | 1375 | 0.00 | 0-2.7 |
| HPRTB | 1 | 452 | 2.21 | 0.06-12.3 | 2 | 1375 | 1.45 | 0.18-5.2 |
| GATA31E08 | 0 | 452 | 0.00 | 0-8.1 | 0 | 1375 | 0.00 | 0-2.7 |
| DXS10147 | 0 | 452 | 0.00 | 0-8.1 | 0 | 1375 | 0.00 | 0-2.7 |
| DXS7423 | 0 | 452 | 0.00 | 0-8.1 | 0 | 1375 | 0.00 | 0-2.7 |
| Other | | | | | | | | |
| **Overall** | **8** | **6780** | **1.18** | **0.51-2.3** | **18** | **20625** | **0.87** | **0.52-1.4** |

Table continues on next page.

**C.**

| | Literature summary | | | | Combined | | | |
|---|---|---|---|---|---|---|---|---|
| **Marker** | **Mutations** | **Meioses** | **Mutation rate ($x10^{-3}$)** | **95% CI ($x10^{-3}$)** | **Mutations** | **Meioses** | **Mutation rate ($x10^{-3}$)** | **95% CI ($x10^{-3}$)** |
| DXS8378 | 4 | 3882 | 1.03 | 0.28-2.6 | 8 | 5257 | 1.52 | 0.66-3.0 |
| DXS9902 | 0 | 458 | 0.00 | 0-8.0 | 4 | 1833 | 2.18 | 0.59-5.6 |
| DXS6795 | *na* | *na* | *na* | *na* | 0 | 1375 | 0.00 | 0-2.7 |
| DXS7132 | 11 | 4965 | 2.22 | 1.1-4.0 | 15 | 6340 | 2.37 | 1.3-3.9 |
| DXS6803 | 2 | 1015 | 1.97 | 0.24-7.1 | 3 | 2390 | 1.26 | 0.26-3.7 |
| DXS6789 | 3 | 3478 | 0.86 | 0.18-2.5 | 5 | 4853 | 1.03 | 0.33-2.4 |
| DXS7424 | 2 | 1805 | 1.11 | 0.13-4.0 | 2 | 3180 | 0.63 | 0.08-2.3 |
| DXS101 | 1 | 2534 | 0.39 | 0.01-2.2 | 1 | 3909 | 0.26 | 0.01-1.42 |
| GATA172D05 | 0 | 876 | 0.00 | 0-4.2 | 1 | 2251 | 0.44 | 0.01-2.5 |
| DXS7130 | *na* | *na* | *na* | *Na* | 0 | 1375 | 0.00 | 0-2.7 |
| GATA165B12 | 0 | 958 | 0.00 | 0-3.8 | 0 | 2333 | 0.00 | 0-1.6 |
| HPRTB | 6 | 4530 | 1.32 | 0.49-2.9 | 8 | 5905 | 1.35 | 0.59-2.7 |
| GATA31E08 | 1 | 1127 | 0.89 | 0.02-4.9 | 1 | 2502 | 0.40 | 0.01-2.2 |
| DXS10147 | 0 | 54 | 0.00 | 0-66.0 | 0 | 1429 | 0.00 | 0-2.6 |
| DXS7423 | 2 | 3515 | 0.57 | 0.07-2.1 | 2 | 4890 | 0.41 | 0.05-1.5 |
| Other | 60 | 31488 | 1.91 | 1.5-2.5 | 60 | 31488 | 1.91 | 1.5-2.5 |
| **Overall** | **92** | **60685** | **1.52** | **1.2-1.9** | **110** | **81310** | **1.35** | **1.1-1.6** |

The overall mutation rates from published X STR studies also demonstrated general similarity with previously reported Y and autosomal rates (see Table 1.3). Compared with the overall rate in the present study, 14 published X STR studies reported higher mutation rates, but only three approached significance based upon confidence interval bounds: $4.76 \times 10^{-3}$ (95% CI: $2.1\text{-}9.4 \times 10^{-3}$) [211], $2.09 \times 10^{-3}$ (95% CI: $1.2\text{-}3.4 \times 10^{-3}$) [209], both observed in German populations, and $3.25 \times 10^{-3}$ (95% CI: $2.0\text{-}5.0 \times 10^{-3}$) [193]. None of the studies with smaller overall mutation rates were significantly different from this study's overall rate. Many studies that exhibited low overall mutation rate values also relied upon a smaller number of meioses (hundreds rather than thousands), and the uncertainty of the resultant rate was reflected in larger confidence intervals. One notable exception was the study relying upon only 180 meioses in a Korean population that observed the highest overall mutation rate [93]. The need for larger sample sizes was underscored by these data, and therefore mutation rates taking into account published data as well as data collected as part of this study were calculated.

The combined overall rate of $1.35 \times 10^{-3}$ (95% CI: $1.1\text{-}1.6 \times 10^{-3}$; Table 5.1), like the overall rate from the literature summary, was higher than that observed for the total U.S. dataset in the present study. This difference can be explained by considering that additional markers were included in the calculation of the higher two rates that were not included in the total U.S. dataset. When the three markers with the highest marker-specific mutation rates in the literature summary dataset and the combined

dataset (ARA, DXS8377, and DXS10135) were removed, the overall mutation rates became 1.23 x $10^{-3}$ (95% CI: 0.96-1.6 x $10^{-3}$) and 1.14 x $10^{-3}$ (95% CI: 0.91-1.4 x $10^{-3}$) respectively. These values further decrease to 1.10 x $10^{-3}$ (95% CI: 0.75-1.6 x $10^{-3}$) for the literature summary dataset and 1.00 x $10^{-3}$ (95% CI: 0.74-1.3 x $10^{-3}$) for the combined dataset if only the 15 markers used in this study are considered. These values are more similar to the overall rate observed in the present study, and illustrate the variation of mutation rate with STR marker.

*5.1.2. Population-specific mutation rates*

The overall rate for the African American population was the lowest overall rate, and was significantly smaller than both the overall rate from the literature summary as well as the combined overall rate. The total number of observed meioses for the African American population was at least 13% less than that of either the U.S. Caucasian and U.S. Hispanic populations, indicating that further study would be necessary to determine if this difference was authentic. This trend is in contrast to that observed for Y chromosomal STRs, where a higher mutation rate in African Americans compared with U.S. Caucasians or U.S. Hispanics was detected [196,197].

Considering the significance of the difference between overall rates for the African American population compared to the literature summary and combined study, populations from the literature summary with more than one published mutation rate study (Table 1.3) were combined to obtain one overall mutation rate per population. The overall rates for populations from China, Germany, Italy, Pakistan, Poland, and Spain showed no significant differences based upon confidence interval bounds from each other, the literature summary, the combined study, or the U.S. Caucasian and U.S. Hispanic populations. However, the overall rates for the published German and Spanish populations were significantly larger than that of the African American population studied here.

Mutations at certain markers appeared to be population-specific in this study. For example, mutations at DXS7132 were only observed in the African American and U.S. Caucasian populations while mutations at DXS6789 occurred only in U.S. Hispanics. Confidence intervals for each marker-specific rate in all three

populations demonstrated substantial overlap, however. For the individual U.S. population groups, more meioses will need to be observed before the true marker-specific and/or population-specific mutation rates can be assessed.

*5.1.3. Marker-specific mutation rates*

In general, the observed marker-specific mutation rates within this study were similar across populations and markers as well as consistent with the overall rate (Table 5.1). There were three instances, however, where the difference between the marker-specific rate and the overall rate approached significance based upon overlap of the confidence intervals: markers DXS8378 and DXS7132 in the U.S. Caucasian population and marker DXS9902 in the U.S. Hispanic population, which exhibited mutation rates higher than the overall rate observed in this study. However, these rates were not significantly different from the population-specific overall rates, and in the collective U.S. population, the marker-specific rates for DXS8378, DXS7132, and DXS9902 agreed with the overall rate.

Two markers exhibited a mutation rate of zero in both this study and published studies: GATA165B12 and DXS10147. Both markers have a relatively small allele range; in U.S. populations, seven alleles were observed at GATA165B12 and eight at DXS10147 (Table 4.16). Markers for which no mutation rate studies have been published (DXS7130 and DXS6795) exhibited no mutations in this study. Mutations were observed for the first time at DXS9902 and GATA172D05 in this study, likely owing to the larger number of total meioses examined here.

Within the combined dataset representing the pooled rates from this study and the literature summary, there were differences between the mutation rates of different markers (Table 5.1). The difference between the mutation rate for DXS7132, which exhibited the largest marker-specific rate, and DXS101, GATA165B12, and DXS7423 approached significance based on the confidence interval bounds. Additionally, the combined overall mutation rate, though generally higher than many marker-specific rates in the combined dataset, yielded a confidence interval completely contained within those of the marker-specific rates for all but DXS101 and DXS7423. Given the relatively large (>1300) number of meioses for all markers and the small confidence interval ranges, it is likely this difference is genuine for at

least these two markers, and their true mutation rates are indeed lower than for other markers or for X STRs in general. Further study, however, could help confirm this hypothesis before marker-specific rates were incorporated into practice.

### *5.1.4. Characterization of observed mutations*

Characteristics of the 18 mutations observed in this study are summarised in Table 5.2. Through sequencing of regions flanking an STR mutational event and analysis of haplotypes in families with multiple children, it has been demonstrated that one-step mutations are far more frequent than mutations involving a change of more than one repeat unit or a partial repeat unit [195,301,308]. Therefore, when assigning "Origin" and "Result" for each observed inconsistency, one-step mutations and single mutational events were favoured, as was consistent with the approach of other mutation rate studies [195,302,309]. As a consequence, multi-step mutations that have been artificially ignored may actually exist in this study, but the overall mutation rate should remain unchanged. For example, the maternal mutational event at DXS9902 in which the obligate paternal allele contained 10 repeats was characterised as resulting from the mutation of the mother's 11 allele into a 10 allele (a loss of one repeat unit; Table 5.2). However, it is also possible that the maternal 12 allele could be the progenitor of the daughter's 10 allele, requiring a two-step mutational event. Of note, the assignment of "Origin" and "Result" in this case would not be changed, and the scenario nevertheless contributes one mutation to the overall rate. In total and including this example, five mutations (the aforementioned example, the mutation of unknown origin at DXS9902, and the three mutations involving mother-son duos) observed in this study could be alternatively explained by a two-step mutation without requiring an additional mutational event. In all cases, the designation for "Origin" and "Result" as well as the overall number of observed mutations would remain unchanged if a two-step mutation were considered.

**Table 5.2. Characteristics of the 18 observed mutations.** One-step mutations and single mutational events were assumed for assigning "Origin" and "Result," as other types of mutations are less common. Paternal genotypes are not listed in the table for families with sons.

| Marker | Population | Origin | Maternal Genotype | Paternal Genotype | Progenal Genotype | Result |
|---|---|---|---|---|---|---|
| DXS8378 | U.S. Caucasian | Maternal | 11,12 | -- | 10 | Loss |
| DXS8378 | U.S. Caucasian | Unknown | 10,12 | 10 | 10,**11** | Unknown |
| DXS8378 | U.S. Caucasian* | Paternal | 10,10 | 12 | 10,**11** | Loss |
| DXS8378 | U.S. Hispanic | Maternal | 11,12 | -- | 13 | Gain |
| DXS9902 | U.S. Caucasian** | Paternal | 9,10 | 10 | 9,**9** | Loss |
| DXS9902 | U.S. Hispanic | Paternal | 10,10 | 11 | 10,**12** | Gain |
| DXS9902 | U.S. Hispanic | Unknown | 10,11 | 11 | 11,**12** | Gain |
| DXS9902 | U.S. Hispanic | Maternal | 11,12 | 10 | **10**,10 | Loss |
| DXS7132 | African American | Paternal | 14,16 | 14 | **15**,16 | Gain |
| DXS7132 | U.S. Caucasian* | Maternal | 13,14 | -- | **15**,16 | Gain |
| DXS7132 | U.S. Caucasian | Paternal | 13,14 | 15 | 14,**16** | Gain |
| DXS7132 | U.S. Caucasian** | Paternal | 15,15 | 14 | **13**,15 | Loss |
| DXS6803 | U.S. Hispanic | Maternal | 12,12.3 | 13.3 | **13.3**,13.3 | Gain |
| DXS6789 | U.S. Hispanic | Paternal | 20,23 | 22 | 20,**23** | Gain |
| DXS6789 | U.S. Hispanic | Paternal | 20,23 | 23 | 20,**22** | Loss |
| GATA172D05 | U.S. Caucasian | Unknown | 6,10 | 10 | 10,**11** | Gain |
| HPRTB | U.S. Caucasian | Maternal | 11,12 | -- | 13 | Gain |
| HPRTB | U.S. Hispanic | Paternal | 14,14 | 12 | **13**,14 | Gain |

*Two mutations observed within a single family.
**Two mutations observed within a second single family.

Examination of the progenal and parental genotypes revealed that all observed mutations could be explained by a change of one repeat unit (Table 5.2), which is consistent with the model of strand slippage during replication as the mechanism of microsatellite mutation [194]. Both gains and losses of repeat units were noted in the observed mutations, with gains outnumbering losses by approximately 2:1. This bias towards microsatellite expansion has been noted in other mutation rate studies [198,199,307,309], though both an excess of losses [195,301] as well as equal rates [98,196,197,302,305] have been noted by others. For approximately half of the mutations, the progenitor allele was the most frequent for that particular marker and population, reflecting the greater opportunity for observing mutation. Additionally, since in most cases the intermediate alleles tended to be the most frequent, mutations involving the largest or the smallest alleles were not observed in this dataset. Because the opportunity to observe mutations in these extremes was so much lower than for the intermediate-sized alleles, no attempt to correlate mutation rate with progenitor allele size was made in this study.

Two U.S. Caucasian families exhibited two mutations each; one family displayed paternal mutations resulting in a loss of a repeat unit at both DXS9902 & DXS7132 while the other family showed a maternal mutation resulting in a gain of a repeat unit at DXS7132 and a paternal mutation resulting in a loss of a repeat unit at DXS8378. Individually, these three markers had the highest mutation rates in this study and within the combined dataset. Though multiple mutational events within one family are rare, they should not be entirely unexpected. Based upon the upper confidence interval bounds of the two markers with the highest mutation rates (DXS9902 and DXS7132), two simultaneous mutations could be expected to occur approximately once in every 41,500 meioses.

*5.1.5. Mutation rate and repeat structure*

Studies of Y STR mutation rates have previously indicated a bias towards a higher mutation rate for longer repeat units [198,306]. To examine whether the mutation rates for the 12 tetranucleotide and 3 trinucleotide X STR markers used in this study might follow this same trend, tetranucleotide and trinucleotide mutation rates were calculated (Table 5.3). Though the population-specific rates appeared similar for both types of repeats, the rates for the total U.S. and combined datasets showed values approaching significance (based on confidence interval bounds) that were higher for tetranucleotide repeats compared with trinucleotide. The relatively small number of markers with trinucleotide repeats compared to those with tertranucleotide repeats may begin to explain this difference, and further study of trinucleotide markers would be necessary to confirm this trend for X STRs.

**Table 5.3. X STR mutation rate by repeat length and type.** The 15 markers used to determine the overall mutation rate in this study were grouped either according to repeat length (tetra- or trinucleotide) or repeat type (simple or compound/complex), and corresponding mutation rates and 95% confidence intervals (CI) were calculated. Compound/complex repeat type included markers with microvariants. Markers with tetranucleotide repeat motifs included DXS6789, DXS7130, GATA31E08, GATA165B12, GATA172D05, DXS10147, DXS8378, DXS7132, DXS6803, HPRTB, DXS7423, and DXS9902. Markers with trinucleotide repeat motifs included DXS7424, DXS6795, and DXS101. Markers with simple repeat motifs were DXS7424, GATA165B12, GATA172D05, DXS10147, DXS8378, HPRTB, and DXS9902. Markers with compound/complex repeat motifs (including microvariants) were DXS6789, DXS7130, GATA31E08, DXS101, DXS6795, DXS7132, DXS6803, and DXS7423.

| Repeat | Markers (N) | Mutations | Meioses | Mutation rate ($x10^{-3}$) | 95% CI ($x10^{-3}$) | Mutations | Meioses | Mutation rate ($x10^{-3}$) | 95% CI ($x10^{-3}$) | Mutations | Meioses | Mutation rate ($x10^{-3}$) | 95% CI ($x10^{-3}$) |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | **African American** | | | | **U.S. Caucasian** | | | | **U.S. Hispanic** | | | |
| Tetranucleotide | 12 | 1 | 4680 | 0.21 | 0.01-1.2 | 9 | 6396 | 1.41 | 0.64-2.7 | 8 | 5424 | 1.47 | 0.64-2.9 |
| Trinucleotide | 3 | 0 | 1170 | 0.00 | 0-3.1 | 0 | 1599 | 0.00 | 0-2.3 | 0 | 1356 | 0.00 | 0-2.7 |
| Simple | 7 | 0 | 2730 | 0.00 | 0-1.3 | 6 | 3731 | 1.61 | 0.6-3.5 | 5 | 3164 | 1.58 | 0.51-3.7 |
| Compound/ Complex | 8 | 1 | 3120 | 0.32 | 0.01-1.8 | 3 | 4264 | 0.70 | 0.15-2.0 | 3 | 3616 | 0.83 | 0.17-2.4 |
| | | **This study** | | | | **Combined** | | | | | | | |
| Tetranucleotide | 12 | 18 | 16500 | 1.09 | 0.65-1.7 | 47 | 41358 | 1.14 | 0.84-1.5 | | | | |
| Trinucleotide | 3 | 0 | 4125 | 0.00 | 0-0.89 | 3 | 8464 | 0.35 | 0.07-1.0 | | | | |
| Simple | 7 | 11 | 9625 | 1.14 | 0.57-2.0 | 23 | 22188 | 1.04 | 0.66-1.6 | | | | |
| Compound/ Complex | 8 | 7 | 11000 | 0.64 | 0.26-1.3 | 26 | 27634 | 0.98 | 0.64-1.4 | | | | |

Though it has been previously noted for autosomal and Y STRs that mutations observed at microvariant and/or compound repeats appeared more common than at simple repeats [199,301,306], this study did not yield the same results. Separating the 15 markers into two groups based upon repeat structure ("simple" and "compound/complex" including microvariant), two sets of approximately equal numbers of markers and meioses were formed (Table 5.3). Mutation rates appeared slightly higher for simple repeats than for compound/complex repeats in the U.S. Caucasian, U.S. Hispanic, and total U.S. populations. However, the addition of data from published studies, considerably increasing the size of the dataset, revealed almost identical rates for both types of repeats.

*5.1.6. Maternal versus paternal mutation rate*

Paternal mutations outnumbered maternal mutations, resulting in different mutation rates depending upon allele origin (Table 5.4). Despite examination of more than 2.2 times as many maternal as paternal meioses, the maternal mutation rate remained comparable to the overall observed rate and almost an order of magnitude smaller than the paternal rate. This trend was consistent across populations and overall, with the paternal rate reaching almost three times the maternal rate in the U.S. Caucasian and U.S. Hispanic populations. Though the confidence intervals for the overall mutation rates for maternal and paternal transfers overlap by a small margin, both rates fell outside of the 95% CI for the overall mutation rate for all meioses. Previous studies have also found that paternal mutations are more frequent for both autosomal STRs [195,301,302,304,308-310] as well as X STRs [193,211]. This difference has been attributed to the variation in the number of cell divisions that occur between the sexes; the formation of oocytes is generally complete by birth, while sperm cells are renewed throughout the life of the male [195,309,311,312]. Additionally, the higher paternal X STR mutation rate corroborates the idea that the mechanism of microsatellite mutation may be independent of recombination [199,313] (which is absent within a paternally-inherited X chromosome) and explains the similarity in overall mutation rate for both the gonosomes and autosomes.

**Table 5.4. X STR mutation rate by origin of mutation.** Maternal and paternal mutation rates for each of the studied populations are shown.

| Population | Origin | Mutations | Meioses | Mutation Rate (x $10^{-3}$) | 95% CI (x $10^{-3}$) |
|---|---|---|---|---|---|
| African American | Maternal | 0 | 4095 | 0.00 | 0-0.9 |
| | Paternal | 1 | 1755 | 0.57 | 0.01-3.2 |
| U.S. Caucasian | Maternal | 3 | 5458 | 0.55 | 0.1-1.6 |
| | Paternal | 4 | 2533 | 1.58 | 0.4-4.0 |
| U.S. Hispanic | Maternal | 3 | 4649 | 0.65 | 0.1-1.9 |
| | Paternal | 4 | 2129 | 1.88 | 0.5-4.8 |
| **This study (U.S. total)** | **Maternal** | **6** | **14202** | **0.42** | **0.2-0.9** |
| | **Paternal** | **9** | **6417** | **1.40** | **0.6-2.7** |

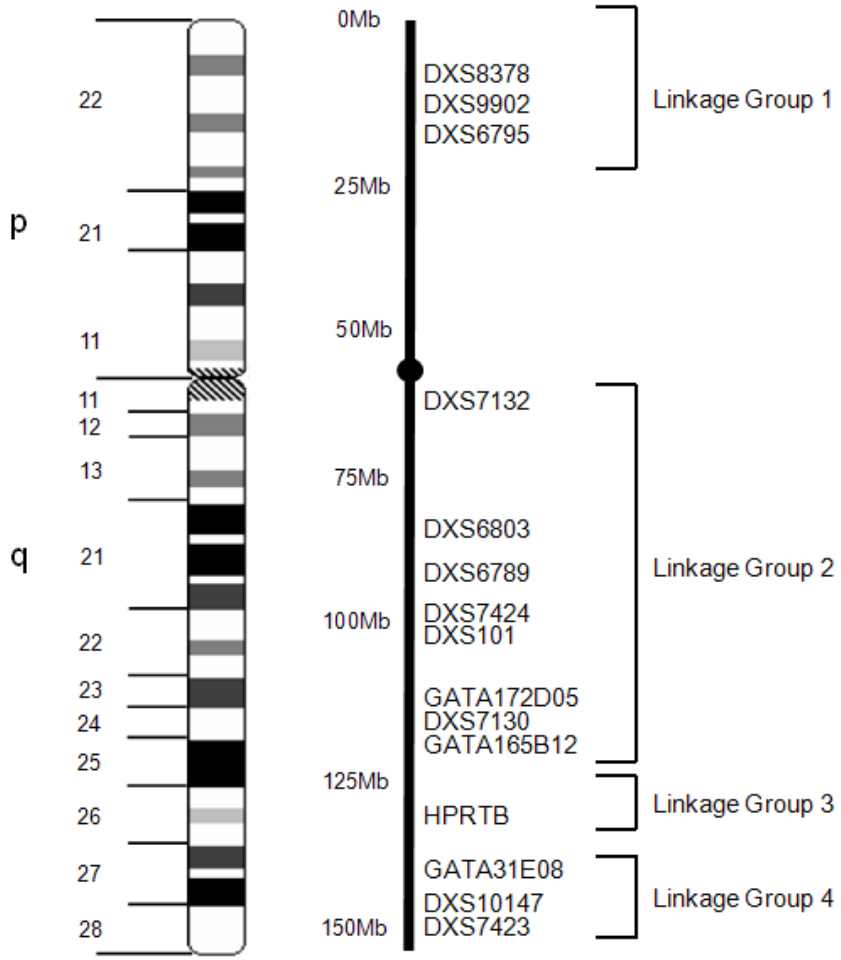
*5.2. Recombination Study*

In Chapter 4, linkage disequilibrium within U.S. population groups was evaluated for the described 15 markers and haplotypes based upon prosed linkage groups were presented for reference. Here, the linkage between the 15 markers and four linkage groups will be assessed using classical analyses of recombination rates in multi-generational family pedigrees.

*5.2.1. Physical location of markers*

Marker locations were determined based upon In Silico PCR BLAT searches [257] and organised along the chromosome in Figure 5.1. Of the 15 markers studies here, the four original proposed linkage groups described by Szibor *et al.* [209] contained the following markers: DXS8378 and DXS9902 in linkage group 1; DXS7132, DXS6789, DXS101, DXS7424, and GATA172D05 in linkage group 2; HPRTB in linkage group 3; and DXS7423 in linkage group 4. Additional markers (DXS6795, DXS6803, DXS7130, GATA165B12, GATA31E08, and DXS10147) included within each linkage group were hypothesised based upon location, the Forensic ChrX Research website [157], and linkage disequilibrium analysis (see Chapter 4).

**Figure 5.1. Physical location of 15 X STR markers and four proposed linkage groups on the chromosome.**




*5.2.2. Recombination rate assessment*

Of the Type I families identified within the dataset as appropriate for linkage analysis, 50 families were analyzed using the manual method. Homozygous genotypes and mutations that rendered a marker uninformative for recombination were excluded, and recombination was defined as a change in source chromosome between two adjacent markers. The recombination rate (Θ) and LOD scores (Z) based upon observed instances of recombination is shown in Table 5.5. Logarithm of the odds (LOD) scores above 3 are generally considered strongly indicative of linkage; however, an LOD of 2 was used initially to identify the four linkage groups [209].

**Table 5.5. Observed recombination rate between 14 pairs of X STR markers.**
LG: linkage group, LOD: logarithm of the odds.

| Location | X STR marker pair | Total number of informative meioses | Instances of observed recombination | Observed recombination rate | LOD scores (Z) |
|---|---|---|---|---|---|
| Linkage Group 1 | DXS8378-DXS9902 | 187 | 21 | 0.11 | 27.76 |
| | DXS9902-DXS6795 | 194 | 26 | 0.13 | 25.21 |
| Border LG1-LG2 | DXS6795-DXS7132 | 158 | 63 | 0.40 | 1.42 |
| Linkage Group 2 | DXS7132-DXS6803 | 186 | 23 | 0.12 | 25.77 |
| | DXS6803-DXS6789 | 182 | 15 | 0.08 | 32.29 |
| | DXS6789-DXS7424 | 172 | 6 | 0.03 | 40.47 |
| | DXS7424-DXS101 | 179 | 0 | 0 | 53.88 |
| | DXS101-GATA172D05 | 194 | 15 | 0.08 | 35.47 |
| | GATA172D05-DXS7130 | 149 | 14 | 0.09 | 24.69 |
| | DXS7130-GATA165B12 | 91 | 4 | 0.04 | 20.27 |
| Border LG2-LG3 | GATA165B12-HPRTB | 141 | 16 | 0.11 | 20.79 |
| Border LG3-LG4 | HPRTB-GATA31E08 | 200 | 19 | 0.10 | 32.94 |
| Linkage Group 4 | GATA31E08-DXS10147 | 177 | 37 | 0.21 | 13.87 |
| | DXS10147-DXS7423 | 134 | 0 | 0 | 40.34 |

The observed recombination rate varied from zero (marker pairs DXS7424-DXS101 and DXS10147-DXS743) to 0.21 (marker pair GATA31E08-DXS10147) within linkage groups and from 0.10 (border of linkage groups 3 and 4) to 0.40 (border between linkage groups 1 and 2) between linkage groups. The highest recombination rate was observed for the pair of markers defining the boundary between linkage groups 1 and 2; however, free recombination ($\Theta = 0.5$) was never observed. All marker pairs, including those within linkage groups, exhibited a non-zero recombination rate except two: DXS7424-DXS101 in linkage group 2 and DXS10147-DXS7423 in linkage group 4. LOD scores, however, strongly indicated linkage between all pairs except those at the boundary of linkage groups 1 and 2.

The computer-based analyses undertaken with this dataset required the modification of scripts previously designed to accommodate only 12 X STR markers [218]. Unforeseen complications in the ability to perform the necessary computations with available computer systems prevented the analyses of the Type II families at the time of writing. Instead a slightly larger number (58) of Type I families were analyzed using three different starting values: distance-interpolated recombination rates, all recombination rates equal to 0.25, and four groups of unlinked markers. Despite these different starting points, the optimisation converged at the same location in

each case, indicating a robust optimisation result; the values for the recombination rates as well as their 95% support intervals (see [218] for a complete definition) are given in Table 5.6.

**Table 5.6. Maximum likelihood estimates of recombination rate between 14 pairs of X STR markers.** The computer-based analysis described by Nothnagel *et al.* [218] was performed on 58 Type I families using three different starting values: distance-interpolated recombination rates, all recombination rates equal to 0.25, and four groups of unlinked markers. LG: linkage group.

| Location | X STR marker pair | Recombination rate | 95% support interval |
|---|---|---|---|
| Linkage Group 1 | DXS8378-DXS9902 | 0.1114 | (0.0714-0.1514) |
| | DXS9902-DXS6795 | 0.1223 | (0.0795-0.1650) |
| Border LG1-LG2 | DXS6795-DXS7132 | 0.4462 | (0.3865-0.5060) |
| Linkage Group 2 | DXS7132-DXS6803 | 0.0964 | (0.0587-0.1341) |
| | DXS6803-DXS6789 | 0.0935 | (0.0578-0.1292) |
| | DXS6789-DXS7424 | 0.0663 | (0.0350-0.0977) |
| | DXS7424-DXS101 | 0.0000 | (NaN-NaN) |
| | DXS101-GATA172D05 | 0.0924 | (0.0592-0.1256) |
| | GATA172D05-DXS7130 | 0.0736 | (0.0411-0.1062) |
| | DXS7130-GATA165B12 | 0.0484 | (0.0149-0.0819) |
| Border LG2-LG3 | GATA165B12-HPRTB | 0.0971 | (0.0589-0.1353) |
| Border LG3-LG4 | HPRTB-GATA31E08 | 0.1092 | (0.0696-0.1488) |
| Linkage Group 4 | GATA31E08-DXS10147 | 0.2045 | (0.1569-0.2521) |
| | DXS10147-DXS7423 | 0.0000 | (NaN-NaN) |

The recombination rates obtained with the computer-based method generally agreed with the values calculated manually, further indicating a robust computation. These values ranged from zero (marker pairs DXS7424-DXS101 and DXS10147-DXS743) to 0.2045 (marker pair GATA31E08-DXS10147) within linkage groups. Between linkage groups, the lowest rate occurred at the border of linkage groups 2 and 3 (0.0971) rather than between linkage groups 3 and 4 as for the classical analyses. The border between linkage groups 1 and 2 revealed the highest overall recombination rate for both the manual (0.40) and the computer-based analyses (0.4462), including a 95% support interval that supports free recombination ($\Theta$ = 0.5). As noted in the classical analyses, all marker pairs, including those within linkage groups, exhibited a non-zero recombination rate using the computer-based method except two: DXS7424-DXS101 in linkage group 2 and DXS10147-DXS7423 in linkage group 4.

Because mutations were ignored in the manual analyses, it was thought that the true recombination rate had likely been overestimated. In the family described in Figure 2.1 (Chapter 2), regarding the 13 allele at DXS7132 in individual 5 as a 14→13 mutational event rather than recombination between DXS7132 and the markers above and below would eliminate 2 of 18 observed events, for example. However, when comparing the classical analyses to computer-based estimates performed using a maximum likelihood approach taking mutation rates into account, the values were found to be very similar. In all cases, the manually-calculated observed recombination rate fell within the 95% support intervals of the computer-based values except for marker pair DXS6789-DXS7424 where the rate of 0.03 fell just outside the lower limit of the 95% support interval (0.0350). Taken together, the results of this combination of methods indicate a robust estimate of the recombination rate between these 14 marker pairs has been achieved.

*5.2.3. Genetic Distance Calculations*

The genetic distance values calculated as part of the computer-based analyses varied only marginally by starting value, and are given for each marker pair in Table 5.7. The relative rate of recombination was generally positively correlated with physical distance between markers in this study: as the distance between the markers increased, the recombination rate increased. There was one notable exception to this trend. Marker pair GATA31E08-DXS10147 exhibited a much higher mutation rate (0.21 and 0.2045) than marker pairs with similar genetic distances separating them, as reflected by the genetic distance estimates. Further study is necessary to determine whether this result may indicate a true recombination "hot spot" or may be influenced by factors such as linkage disequilibrium, sampling bias, and/or population substructure. However, the results obtained with this set of markers also agreed with five published recombination studies [237,238,314-316] with regards to recombination between linkage groups 3 & 4 even though their borders were defined by different markers in each. Using a mapping function to compare observed to expected recombination rates for this pair based on physical distance, the expected rates in the published studies were lower than the observed rates. In this study, this phenomenon is reflected in a genetic distance calculation that is almost 1.7 times greater than that of the physical distance. These concordant results may indicate the

presence of a region of locally enhanced recombination that may also include marker pair GATA31E08-DXS10147.

**Table 5.7. Genetic distances between 14 X STR marker pairs given for three different starting value schemes.** LG: linkage group; Mb: megabases; cM: centimorgans; theta: recombination rate.

| Location | X STR marker pair | Physical distance (Mb) | Starting value scheme (cM) | | |
|---|---|---|---|---|---|
| | | | Distance-interpolated | Theta equals 0.25 | 4 unlinked groups |
| Linkage Group 1 | DXS8378-DXS9902 | 5.90 | 11.3310 | 11.3310 | 11.3314 |
| | DXS9902-DXS6795 | 7.92 | 12.4783 | 12.4782 | 12.4781 |
| Border LG1-LG2 | DXS6795-DXS7132 | 41.42 | 71.6852 | 71.6863 | 71.6929 |
| Linkage Group 2 | DXS7132-DXS6803 | 21.75 | 9.7662 | 9.7664 | 9.7663 |
| | DXS6803-DXS6789 | 9.02 | 9.4599 | 9.4602 | 9.4599 |
| | DXS6789-DXS7424 | 5.17 | 6.6694 | 6.6696 | 6.6698 |
| | DXS7424-DXS101 | 0.79 | 0.0000 | 0.0000 | 0.0000 |
| | DXS101-GATA172D05 | 11.76 | 9.3463 | 9.3462 | 9.3481 |
| | GATA172D05-DXS7130 | 5.02 | 7.4173 | 7.4175 | 7.4159 |
| | DXS7130-GATA165B12 | 2.62 | 4.8550 | 4.8552 | 4.8555 |
| Border LG2-LG3 | GATA165B12-HPRTB | 12.74 | 9.8320 | 9.8321 | 9.8313 |
| Border LG3-LG4 | HPRTB-GATA31E08 | 6.62 | 11.1001 | 11.1000 | 11.0994 |
| Linkage Group 4 | GATA31E08-DXS10147 | 9.35 | 21.7218 | 21.7221 | 21.7211 |
| | DXS10147-DXS7423 | 0.05 | 0.0000 | 0.0000 | 0.0000 |

*5.2.4. Comparisons to Published Studies*

A relatively limited number of X STR recombination rate studies have been performed to date, none of which cover the same set of 15 markers studied here. However, several studies included markers also studied here, and comparisons to those results were made.

In a study of the Argus X-8 kit, 32 two-generation families were studied to estimate recombination rate [237]. Of note, the mothers used in this study did not have confirmed haplotypes; that is, the gametic phase was unknown since no grandparents were included in the study. Therefore, two possibilities had to be considered for each child and accounted for in their analysis of linkage. Their results indicated that while a high degree of (but not complete) linkage existed between pairs of markers within the linkage groups, the recombination rate between linkage groups did not consistently indicate free recombination ($\Theta = 0.5$). Even the border between linkage groups 2 and 3 (defined by DXS10074 & HPRTB in this set of markers), which

exhibited a recombination rate of 0.5, produced a large confidence interval (0.34-0.50) suggesting caution should be taken with the assumption of independence between the linkage groups until further study with a larger sample set could be undertaken. Interestingly, the recombination rate between linkage groups 1 and 2 in the published study (0.45) agreed well with the results obtained here with the maximum likelihood estimation (0.4465) despite the border of linkage group 1 being defined by two different markers (DXS8378 for the published study and DXS6795 here).

A study of 39 X STR markers in 90 three-generation pedigrees analyzed linkage between four groups of two physically-close markers [238]. Strong evidence for independent assortment between linkage groups 1 & 2 and linkage groups 2 & 3 was found, which was generally larger than found in this study. The markers defining the linkage groups, however, differed and may have contributed to the discordant measurements. This same publication also provided LOD scores from a previous study analyzing two-generation families [209]. Four marker pairs overlapped with those studied here: DXS8378-DXS9902, DXS6789-DXS7424, DXS7424-DXS101, and DXS101-GATA172D05. In both studies, the LOD scores indicated nonrandom association between the two markers ($Z > 2$), with higher scores obtained in this study for all pairs.

Additional support for independent assortment between linkage groups 1 & 2 was provided in a study of 20 X STR markers in 80 two-generation families [314]; a recombination rate of 0.49980 was observed between border markers DXS8378 and DXS7132. Also similar to the findings of this study, free recombination could not be assumed between linkage groups 2 & 3 and linkage groups 3 & 4 in the published study, though different markers defined the boundaries of these linkage groups as well. Intragroup trends reported in the published study were also confirmed by results presented here for two of the three overlapping marker pairs. Strong evidence for linkage was observed in both studies for marker pairs DXS7424-DXS101 and DXS101-GATA172D05; however the observed recombination rates for marker pair DXS6789-DXS7424 differ significantly. While this study obtained an observed recombination rate (calculated by maximum likelihood estimate) of 0.0633, the

published study reported a rate of 0.14267 which falls outside the 95% support interval associated with this study's value (0.0350-0.0977).

The computer-based analyses employed here for this set of 15 markers was originally described in a study of 12 markers on two- and three-generation pedigrees [218]. Results of the published study generally agree with those of this study in regards to intergroup recombination rate for linkage groups 1 & 2 (free recombination) and linkage groups 3 & 4 (less than free recombination). Although the published study also found a recombination rate less than 0.5 between linkage groups 2 & 3 (0.4252), the reduction was not as dramatic as observed in this study (0.11/0.0971) and falls well outside the 95% support interval calculated in association with the computer-based method (0.0589-0.1353). This discrepancy is likely due to the difference in the two markers defining the linkage group boundaries in each study (DXS10101 and DXS10146 in the published study and GATA165B12 and HPRTB here). None of the intragroup marker pairs overlapped between the two studies.

Lastly, in a recent study examining the recombination rate of 25 marker pairs in two- and three-generation pedigrees, three markers pairs overlapping with those in this study were investigated: DXS8378-DXS9902, DXS7424-DXS101, and HPRTB-GATA31E08 [316]. Of the two intragroup marker pairs, the recombination rates for DXS8378-DXS9902 agreed for both studies, with overlapping 95% support intervals (0.135 (95% CI: 0.0729-0.2366) for the published study and 0.1114 (95% SI: 0.0714-0.1514) here). The rate for marker pair DXS7424-DXS101 was slightly higher in the published study (0.031) since no recombination events were observed in this study, but LOD scores were high (35.627 and 53.88) for both pairs. Marker pair HPRTB-GATA31E08 defines the boundary between linkage groups 3 & 4 in both studies, and the observed recombination fractions agree with overlapping support intervals (0.133 (95% CI: 0.0844-0.2068) for the published study and 0.1092 (95% SI: 0.0696-0.1488) here).

The common themes resonating through the published studies were both the importance of linkage in the interpretation of X STR data and the need for larger, collaborative recombination studies investigating a comprehensive set of markers.

The present study contributes to these requirements with a large-scale evaluation in which a portion of the pedigrees are widely available to the scientific community for future study.

# Chapter 6. Exchange & Comparability of Data

In order to ensure that results obtained in different laboratories can be compared to one another in a meaningful way, the repeat number assigned to an allele must correspond to exactly the same allele in another laboratory. The easiest way in which to ensure this accuracy is to sequence a number of representative alleles at each marker and clearly define the repeat structure used to determine the allele call; this process is especially important when using a custom multiplex designed by an individual laboratory. As increasing routine use of X STRs motivates the production and wide-scale availability of commercial kits, allele calls will be standardized and the nomenclature concerns will decline. Comparison of the custom multiplex described here to the limited-availability commercial kit, QIAGEN® Investigator Argus X-12, can serve to verify nomenclature currently employed for overlapping markers. In conjunction, this commercial kit was evaluated for use with U.S. populations for the first time.

*6.1. Allele Sequencing*

In addition to the routine allele sequencing necessary during the development of the multiplex assays, sequencing of a subset of samples at each marker was accomplished in an attempt to better define the variation present at each marker as well as to verify concordance of the repeat structure with that of published data obtained using larger primer sets. Any anomalous profile characteristics that might be clarified with sequence data were also analyzed, such as novel microvariants. Though the ultimate goal might be to obtain sequence data from every observed allele at each marker, this study was limited to a subset of representative alleles for which male or homozygous female profiles were available. During this initial characterisation, the sequencing process revealed other pertinent information such as primer binding site mutations, for example. For nine markers in particular – DXS9902, DXS7130, DXS7132, DXS8378, DXS101, DXS6795, DXS10147, HPRTB, and DXS6803 – new alleles and/or microvariants were confirmed.

A minimum of 2 and a maximum of 73 alleles from each marker were sequenced as part of the ongoing quality control process to confirm and/or clarify repeat structures.

Table 6.1 lists each allele that was sequenced for each marker and the structure observed.

**Table 6.1. Allele sequencing results for a sampling of alleles from 16 X STR markers studied here.** Alleles that were unpublished at the time they were observed and sequenced are bolded. $N_8$: TCTGTCCT. *Microvariant alleles result from variation in the number of bases present in a poly-A stretch 6 base pairs prior to the start of the repeat.

| Marker | Allele | Observed repeat structure | N |
|---|---|---|---|
| DXS9902 | 9 | $(GATA)_9$ | 1 |
| | 10 | $(GATA)_{10}$ | 2 |
| | **10.1** | **$(GATA)_{10}$*** | **1** |
| | **10.3** | **$(GATA)_{11}$*** | **1** |
| | **11.1** | **$(GATA)_{11}$*** | **1** |
| | 12 | $(GATA)_{12}$ | 1 |
| | **12.1** | **$(GATA)_{12}$*** | **1** |
| DXS7130 | **9** | **$(TATC)_9$** | **1** |
| | 11 | $(TATC)_{11}$ | 1 |
| | 12 | $(TATC)_{12}$ | 5 |
| | 13 | $(TATC)_{13}$ | 8 |
| | **14** | **$(TATC)_{14}$** | **2** |
| | 14.3 | $(TATC)_5$-ATC-$(TATC)_9$ | 1 |
| | 16 | $(TATC)_4$-AATC-$(TATC)_{11}$ | 1 |
| | 16.3 | $(TATC)_5$-ATC-$(TATC)_{11}$ | 1 |
| | 17 | $(TATC)_4$-AATC-$(TATC)_{12}$ | 1 |
| | **20.2** | **$(TATC)_5$-ATC-$(TATC)_4$-ATC-$(TATC)_{10}$** | **1** |
| GATA31E08 | 8 | $(AGGG)_2$-$(AGAT)_6$ | 4 |
| | 9 | $(AGGG)_2$-$(AGAT)_7$ | 10 |
| | 10 | $(AGGG)_2$-$(AGAT)_8$ | 10 |
| | 11 | $(AGGG)_2$-$(AGAT)_9$ | 8 |
| | 12 | $(AGGG)_2$-$(AGAT)_{10}$ | 8 |
| | | $(AGGG)_3$-$(AGAT)_9$ | 2 |
| | 13 | $(AGGG)_2$-$(AGAT)_{11}$ | 7 |
| | | $(AGGG)_3$-$(AGAT)_{10}$ | 4 |
| | 14 | $(AGGG)_2$-$(AGAT)_{12}$ | 9 |
| | | $(AGGG)_3$-$(AGAT)_{11}$ | 1 |
| | 15 | $(AGGG)_3$-$(AGAT)_{12}$ | 2 |
| | | $(AGGG)_2$-$(AGAT)_{13}$ | 6 |
| | 16 | $(AGGG)_2$-$(AGAT)_{14}$ | 2 |
| DXS7132 | 10 | $(TCTA)_{10}$ | 1 |
| | 13 | $(TCTA)_{13}$ | 1 |
| | 14 | $(TCTA)_{14}$ | 4 |
| | 15 | $(TCTA)_{15}$ | 3 |
| | **15.3** | **$(TCTA)_{13}$-TCA-$(TCTA)_2$** | **1** |
| | **16.3** | **$(TCTA)_{14}$-TCA-$(TCTA)_2$** | **1** |
| | 17 | $(TCTA)_{17}$ | 2 |
| DXS7133 | 11 | $(ATAG)_{11}$ | 2 |
| DXS8378 | 11 | $(CTAT)_{11}$ | 2 |
| | 12 | $(CTAT)_{12}$ | 1 |
| | **15** | **$(CTAT)_{15}$** | **1** |
| DXS7423 | 8 | $(TCCA)_{10}$ | 2 |
| | 11 | $(TCCA)_3$-$N_8$-$(TCCA)_8$ | 2 |
| | 12 | $(TCCA)_3$-$N_8$-$(TCCA)_9$ | 2 |
| | 14 | $(TCCA)_3$-$N_8$-$(TCCA)_{11}$ | 2 |

Table continues on next page.

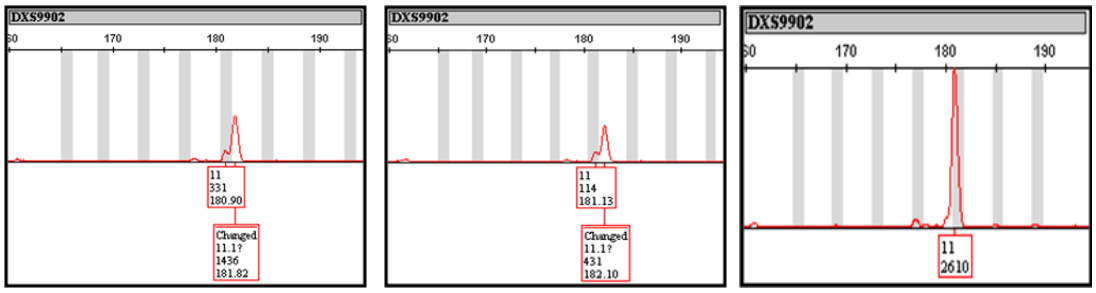
| Marker | Allele | Observed repeat structure | N |
|---|---|---|---|
| DXS6789 | 14 | $(TATG)_{10}$-$(TATC)_4$ | 1 |
| | 15 | $(TATG)_9$-$(TATC)_6$ | 1 |
| | 16 | $(TATC)_{10}$-$(TATC)_6$ | 1 |
| | 17 | $(TATG)_{10}$-$(TATC)_7$ | 1 |
| | 18 | TATC-$(TATG)_9$-$(TATC)_8$ | 1 |
| | 19 | TATC-$(TATG)_9$-$(TATC)_9$ | 1 |
| | 20 | TATC-$(TATG)_9$-$(TATC)_{10}$ | 1 |
| | | TATC-$(TATG)_{10}$-$(TATC)_9$ | 1 |
| | 21 | TATC-$(TATG)_{10}$-$(TATC)_{10}$ | 1 |
| | 22 | TATC-$(TATG)_{10}$-$(TATC)_{11}$ | 1 |
| | 23 | TATC-$(TATG)_{11}$-$(TATC)_{11}$ | 1 |
| | 24 | TATC-$(TATG)_{10}$-$(TATC)_{13}$ | 1 |
| DXS101 | **13** | **$(CTT)_4$-$(ATT)_9$** | **1** |
| | 21 | $(CTT)_{16}$-$(ATT)_5$ | 1 |
| | 23 | $(CTT)_{19}$-$(ATT)_4$ | 2 |
| | 24 | $(CTT)_{15}$-$(ATT)_9$ | 1 |
| | 25 | $(CTT)_{14}$-$(ATT)_{11}$ | 1 |
| DXS7424 | 13 | $(TAA)_{13}$ | 1 |
| | 16 | $(TAA)_{16}$ | 1 |
| DXS6795 | **6** | **$(ATT)_6$** | **1** |
| | **8** | **$(ATT)_8$** | **1** |
| | 9 | ATT-ATC-$(ATT)_7$ | 4 |
| | 10 | $(ATT)_{10}$ | 3 |
| | | ATT-ATC-$(ATT)_8$ | 2 |
| | 11 | $(ATT)_{11}$ | 3 |
| | 12 | $(ATT)_{12}$ | 4 |
| | 13 | $(ATT)_{13}$ | 2 |
| | 14 | $(ATT)_{14}$ | 2 |
| | 15 | $(ATT)_{15}$ | 2 |
| | **17** | **$(ATT)_{17}$** | **3** |
| | **18** | **$(ATT)_{18}$** | **1** |
| DXS10147 | **5** | **$(AAAC)_5$** | **2** |
| | 8 | $(AAAC)_8$ | 2 |
| | 9 | $(AAAC)_9$ | 1 |
| | 10 | $(AAAC)_{10}$ | 1 |
| | **11** | **$(AAAC)_{11}$** | **1** |
| GATA165B12 | 7 | $(AGAT)_7$ | 1 |
| | 10 | $(AGAT)_{10}$ | 2 |
| | 11 | $(AGAT)_{11}$ | 1 |
| GATA172D05 | 6 | $(TAGA)_6$ | 2 |
| | 9 | $(TAGA)_9$ | 2 |
| | 10 | $(TAGA)_{10}$ | 1 |
| | 11 | $(TAGA)_{11}$ | 1 |
| | 12 | $(TAGA)_{12}$ | 3 |
| HPRTB | **7** | **$(ATCT)_7$** | **1** |
| | **8** | **$(ATCT)_8$** | **1** |
| | 12 | $(ATCT)_{12}$ | 12 |
| | 13 | $(ATCT)_{13}$ | 1 |
| | 14 | $(ATCT)_{14}$ | 3 |
| | 15 | $(ATCT)_{15}$ | 2 |
| | **17** | **$(ATCT)_{17}$** | **2** |
| DXS6803 | 7 | $(TCTA)_7$ | 1 |
| | 9.3 | $(TCTA)_8$-TCA-TCTA | 1 |
| | **10.3** | **$(TCTA)_9$-TCA-TCTA** | **1** |
| | 11 | $(TCTA)_{11}$ | 3 |
| | 12.3 | $(TCTA)_{11}$-TCA-(TCTA) | 1 |
| | 13 | $(TCTA)_{13}$ | 1 |
| | **15.3** | **$(TCTA)_{14}$-TCA-TCTA** | **1** |
| | **16** | **$(TCTA)_{12}$-TCCA-$(TCTA)_3$** | **1** |

The allele sequencing process was particularly useful in determining the origin of a peculiar but reproducible microvariant peak observed at DXS9902 for certain samples (Figure 6.1). At these potential "X.1 microvariants," an additional peak was consistently observed that was not present with non-microvariant alleles. Increasing the final extension step to as long as 4 hours did not resolve this anomaly, thus excluding incomplete adenylation as the source. Sequencing results revealed a poly-adenine stretch within the STR amplicon prior to the core repeat region which can be prone to polymerase slippage (Figure 6.2). The result is a mixture of two amplicons that have an identical number of expected GATA repeats whenever an X.1 microvariant allele is present: one with the standard 9 adenine residues prior to the core repeat region (standard allele), and the other with and additional $10^{th}$ adenine (microvariant allele). The decreased peak height ratio observed for heterozygous females with profiles comprised of both the standard allele and its X.1 microvariant is then explained by the summation of signal from the 9-adenine form of the X.1 allele with that of the standard allele (Figure 6.1B). Confirmation of both the structure and cause of the abnormal peak morphology allowed routine calling of X.1 microvariants, even in heterozygous females. Similary, the 10.3 microvariant allele was found to be the result of an allele with 11 GATA repeats but only 8 adenine residues (data not shown), highlighting the potential for other as-yet unobserved microvariants. Given that the frequency of occurrence of these microvariants is not negligible (see Table 4.22), clarification of this result was integral to determining the true discriminatory power of this marker.

In a study of this marker comparing human sequences to those of chimpanzees, the same sequence structure of X.1 microvariants was observed [286]. Due to the presence of nonconsensus alleles in only Europeans in this study, a recent emergence of the version with an additional adenine was postulated. However, in this study, microvariants can be noted in African American, U.S. Caucasian, and U.S. Hispanic populations, seemingly contradicting this hypothesis. Further study would be necessary to reveal further variation and confirm true population specificity.

**Figure 6.1. Abnormal peak morphology observed for potential X.1 microvariants at DXS9902.** A. Example profiles from male samples with presumed 11.1 (first two panels) and standard 11 (right panel) alleles. The extension time was varied for the sample with the presumed 11.1 allele, and is shown below the electropherograms for each example. B. Analogous example profiles for female samples with presumed 11.1 allele (left panel) and 11, 11.1 genotype (right panel). Female samples were amplified using the standard 45 minute extension time only.

**A.**




45 minutes (standard) 240 minutes 45 minutes (standard)

**B.**

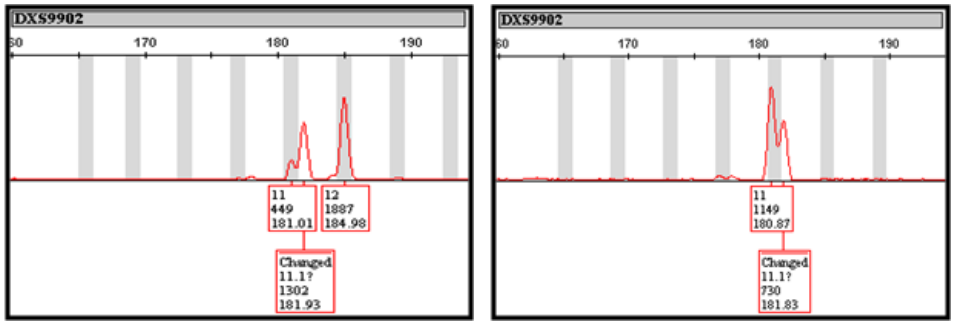
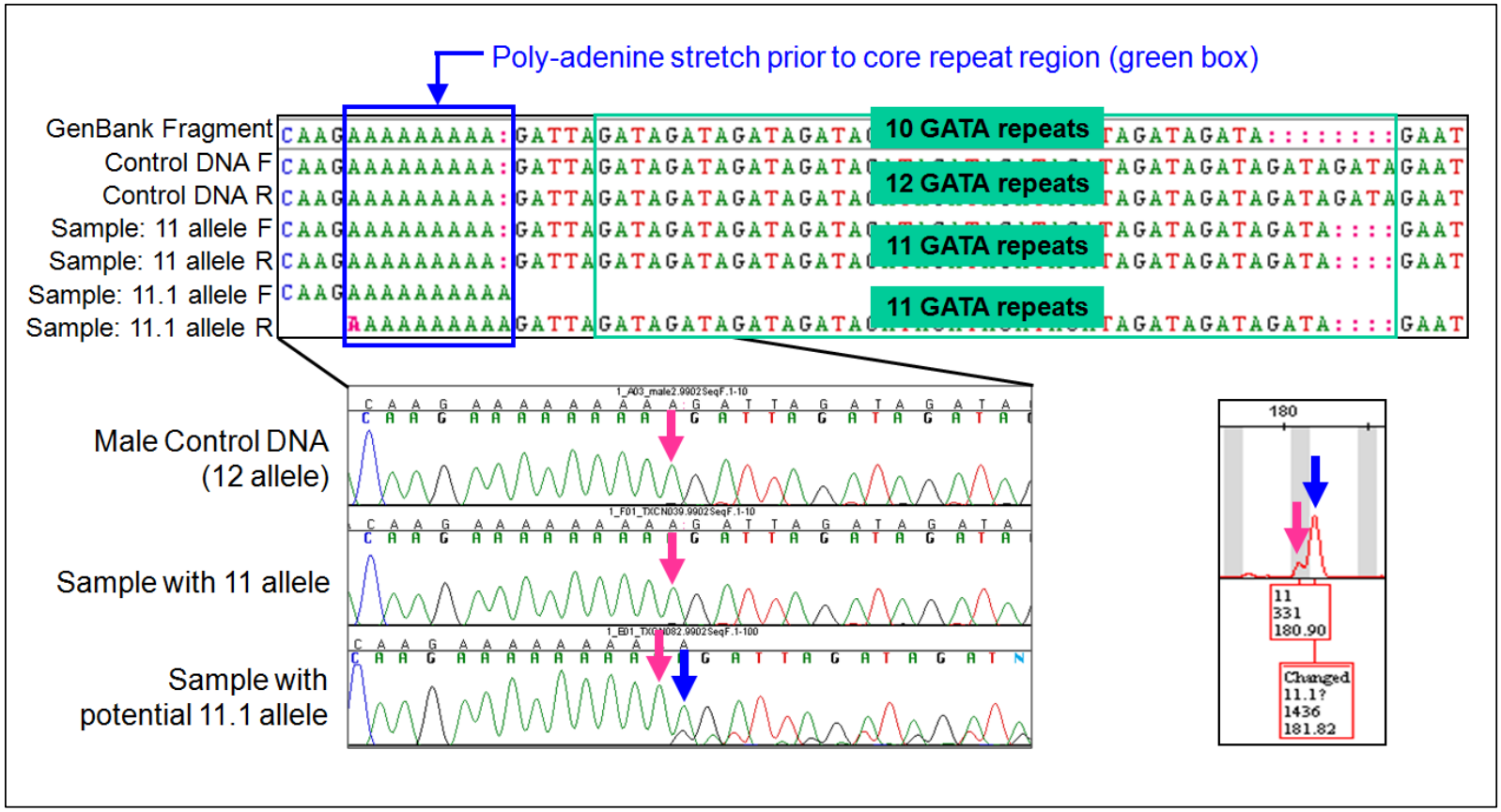


**Figure 6.2. Results of sequencing presumed microvariant alleles at DXS9902.** Sequencing results and corresponding electropherograms (forward sequences only) for three male samples: a control sample with a 12 allele, a sample with an 11 allele, and a sample with a presumed 11.1 allele and abnormal peak morphology.



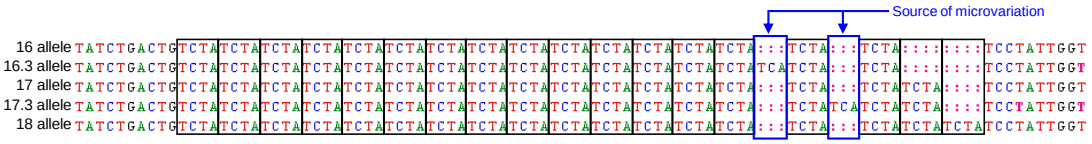
Sequencing of marker DXS7130 revealed a structure for both standard alleles and microvariants that conformed to that previously published by Edelman and Szibor [273]. However, additional sequence variation that has been previously unobserved was present in several alleles in this study. The typical TATC repeat was augmented by an ATTC repeat in both the 16 and 17 alleles sequenced here. Additionally, the novel microvariant 20.2 allele was composed of two incomplete ATC units following either four or five standard TATC units, consistent with other sequenced alleles for which the nonstandard AATC repeat or the ATC incomplete repeat followed four or five standard TATC repeats. This observation may suggest that the loss of an A from the nonstandard AATC repeat results in the incomplete ATC units rather than the loss of the first T of the standard TATC repeat. Further study would be necessary to confirm such a correlation.

Seventy-three alleles were sequenced for marker GATA31E08 in order to understand the variation possible within this compound repeat. Though initial studies referred only to a variable number of AGAT repeats, additional variation was later found in the number of preceding AGGG units [284]. Similar to the published study, either 2 or 3 AGGG repeats were observed before the AGAT repeats, which exhibit larger variation in their numbers. The variation in the number of AGGG units seems to increase with allele size, revealing identical allele calls based upon repeat number for alleles with different sequence structure.

Because microvariation was previously unreported at DXS7132 at the time of observation, both standard and the new microvariant alleles were sequenced to determine the pattern of variation. As shown in Figure 6.3, the source of microvariation was determined to be an incomplete TCA repeat. Therefore, the generalised repeat motif could be redefined from $(TCTA)_x$ to $(TCTA)_x$-$(TCA)_{(0-1)}$-$(TCTA)_2$. In a study comparing the sequences of human and chimpanzee alleles at this marker, a recommendation to call this repeat CTAT (rather than TCTA) was made based upon comparison between the two species. In accordance with ISFG guidelines [242] and an early study describing marker DXS7132 [272], the TCTA repeat designation was used in the present study. Regardless of the unit's structure, the repeat number does not change, and the incomplete repeat units observed within

microvariant alleles agree. Based upon the presence of these microvariants limited to populations originating in South America, it was postulated that the origin of this variation may be Native American. The present study supports that idea, with the majority of microvariant alleles observed in U.S. Hispanic populations (Table 4.22).

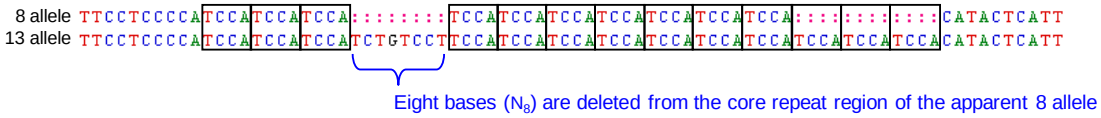
**Figure 6.3. DXS7132 previously unpublished microvariants.** Sequence data obtained for 5 different DXS7132 alleles, including two new microvariants. The source of microvariation was determined to be an incomplete TCA repeat (shown in blue boxes). Standard repeat units (black boxes) confirmed allele calls. Sequence direction is 5' → 3'.




Only male control DNAs were sequenced for marker DXS7133, which was removed from the final set of markers studied, to confirm the reported ATAG nomenclature. Similarly, only a small subset of alleles was sequenced for marker DXS8378, all of which were found to follow the straightforward CTAT structure.

Initial nomenclature discrepancies between studies of DXS7423 were resolved by revising the structure from $(TCCA)_x$ to $(TCCA)_3$-$N_8$-$(TCCA)_x$ [246]. However, allele calls were based upon the number of TCCA repeats only, ignoring the stretch of 8 nucleotides interspersed between them. During sequencing of a newly observed 8 allele in this study, however, it was discovered that this nonstandard stretch could vary as well (Figure 6.4). Therefore, an allele with 10 TCCA repeats missing this 8 nucleotide stretch would be called an 8 allele. Interestingly, no intermediate 9 or 10 alleles were noted in this study, and were only rarely observed in the literature [83,124,317].

**Figure 6.4. Previously unpublished 8 allele at marker DXS7423 shows uncharacteristic repeat pattern.** Sequence data obtained for a standard 13 allele and an apparent 8 allele at DXS7423, revealing unexpected variation. Sequence direction is 5' → 3'.




Sequencing results from a variety of alleles at marker DXS6789 confirmed a compound repeat structure composed of both TATC and TATG units as previously reported [284]. This previous study described the invariant TATC at the start of the region as present in alleles 17 or larger. However, the 17 allele studied here lacked this first TATC unit, proving that additional unreported variation exists at this locus. Also, similar to the results of the previous study, greater variation in the number of TATC units (4-13) than TATG units (9 or 10) was noted here.

Only a small amount of the seemingly unlimited variation potential at marker DXS101 was probed here, which is composed of a variable number of CTT and ATT repeats. Though both 23 alleles sequenced in this study exhibited the same structure ($(CTT)_{19}$-$(ATT)_4$), an alternate 23 allele structure ($(CTT)_{15}$-$(ATT)_7$) was reported by Edelmann, *et al.* [206], and 5 different repeat structures were observed after sequencing just 8 total alleles designated as 25 alleles by fragment analysis methods in both that study and this one. Both the CTT and ATT components of this marker were present in fluctuating number, and the large observed allele range (13-33, Table 4.22) reflects the result of the large number of possible combinations. Another trinucleotide repeat, marker DXS7424, was found to exhibit a simple TAA structure.

Since marker DXS6795 was relatively newly described [318] and unstudied when adopted for inclusion into the described set of markers, there were no allele sequencing studies available. Twenty-eight alleles were sequenced as part of this study, and the variation was found to primarily result from a variable number of ATT repeat units. However, in some alleles, the second ATT unit was replaced by an ATC repeat, and variation resulted from the subsequent ATT repeats. Both

structures could result in the same allele call, as evidenced by the 10 alleles sequenced. This expanded repeat structure was reported for the first time through this work, and a subsequent study confirmed the observations [151].

Similar to DXS6795, marker DXS10147 was relatively novel when this work was started; limited sequencing results were available, however. Edelmann, *et al.* [236] had described a simple AAAC repeat structure that varied in number from 6 to 10. Here, new alleles 5 and 11 were also sequenced, and found to follow this same structure. Markers GATA165B12 and GATA172D05 were also composed of simple repeats (AGAT and TAGA, respectively), but had been more widely studied prior to incorporation into these multiplexes. Though initially described as a simple AGAT repeat at GATA165B12 [274], sequencing performed in a later publication included an additional AGAT unit present at the end of the repeat after 4 invariable bases [319]. The structure suggested in this publication was $(AGAT)_x$-$N_4$-$(AGAT)_y$, where x + y determined the allele call. However, since this terminal unit was not variable, and ISFG guidelines dictate the use of the first described structure, this additional unit was dropped, and the simple AGAT variable unit was used in this study and others since [54,153]. No variability in the simple TAGA repeat unit of GATA172D05 was observed in this study or others [272,284]. Several newly observed alleles at marker HPRTB were sequenced as part of this study and also confirmed to follow the described simple ATCT repeat structure.

Lastly, additional DXS6803 alleles observed within the studied populations were sequenced and generally found to conform to the described structure for both standard and microvariants. One exception was the newly described 16 allele that included a TCCA unit in addition to the standard TCTA repeats. This additional repeat unit had not been previously described in the initial study of the marker [273] and is the first time this additional variation was reported. Similar to the case of marker DXS7130, it appears that the discovery of this additional nonstandard repeat unit might indicate that the incomplete repeat present in DXS6803 microvariant alleles results from a loss of the repeated cytosine within the TCCA rather than the loss of a thymine from the standard TCTA unit.

In summary, the allele sequencing results presented here allow the structure of each marker to be clearly defined and communicated to other laboratories or scientists, increasing the utility of the chosen set. Moreover, an initial understanding of the additional variation present within the repeat structure itself was gained. Harnessing this additional variation in the future through techniques that allow rapid, cost-effective allele sequence interrogation such as next generation sequencing technologies could allow even further discrimination with this same set of markers than traditional fragment analysis.

*6.2. Evaluation of Investigator Argus X-12 Kit Using U.S. Populations*

Though a large number of multiplex assays using X STR markers have been described in the literature (see Table 1.1), there is currently only one commercial kit available, the QIAGEN® Investigator Argus X-12 kit. This kit simultaneously detects 12 X STR markers plus amelogenin. Several recent publications have described its performance on high quality samples and presented allele and haplotype frequency data from populations in Germany [190], Morocco [188], China [183], Hungary [182], and Sweden [192] (see Table 1.2 for more). Further study of these 12 markers is required before the potential of the kit can be fully realised for use in the forensic setting. To this end, 853 samples from the four major U.S. population groups (African American, U.S. Asian, U.S. Caucasian, and U.S. Hispanic) were typed using this commercial kit. Resultant profiles were examined for kit performance measures such as stutter and incidence of off-ladder alleles. Alleles that were off-ladder, unpublished, or designated as null were noted and many were sequenced to confirm repeat structure and number. Additionally, a concordance study between the X-12 kit and the four overlapping markers also present in a published mini X-STR assay [55] was completed. Nearly all of the samples typed were concordant, with only 1 sample (0.1%) exhibiting discordance due to a null allele using the commercial kit. The forensic utility of markers present in both the X-12 kit and the mini-X assays (23 markers total) in these U.S. populations was also compared, demonstrating that the highest mean exclusion chance values corresponded to those X-12 kit markers composed of the most complex repeat structures.

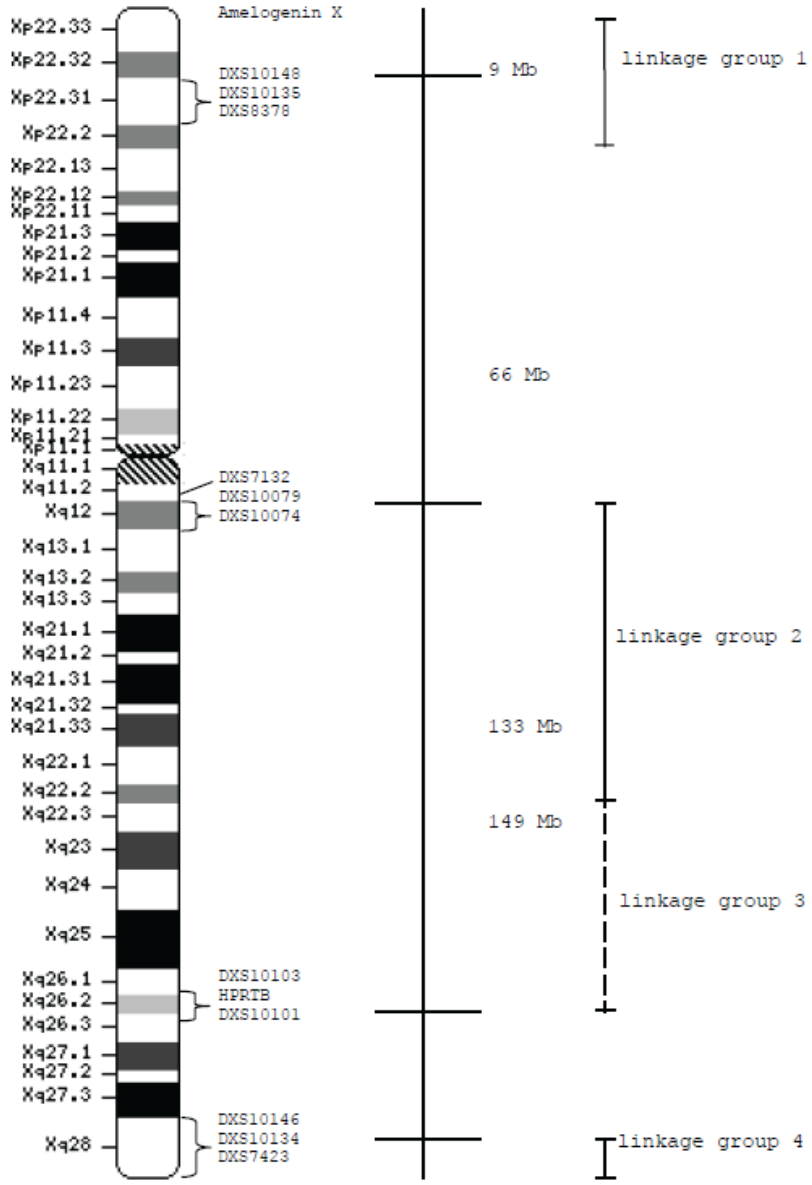
Samples used in this study consisted of a combination of a subset of Sample Set A and Sample Set B used for population databasing (see Chapter 4). Utilizing a predominantly male sample set allowed straightforward assessment of parameters such as stutter and incidence of null alleles. A summary of this sample set is shown in Table 6.2. Because the U.S. Asian individuals typed as part of this study do not necessarily overlap with those typed as part of the population database described earlier, some comparisons will occur between three populations only.

**Table 6.2. Summary of population samples used for evaluation of Investigator Argus X-12 kit.**

| Sample Set | Population | N(males) | N(females) | N(individuals) | N(alleles) |
|---|---|---|---|---|---|
| A | U.S. Asians | 69 | 83 | 152 | 235 |
| B | African Americans | 260 | 0 | 260 | 261 |
| | U.S. Asians | 3 | 0 | 3 | 3 |
| | U.S. Caucasians | 260 | 38 | 298 | 336 |
| | U.S. Hispanics | 140 | 0 | 140 | 140 |
| | **Total** | **732** | **121** | **853** | **975** |

The markers included in the X-12 kit represent the four proposed linkage groups with three markers each: DXS10148, DXS10135, & DXS8378 in linkage group 1; DXS7132, DXS10079, & DXS10074 in linkage group 2; DXS10103, HPRTB, & DXS10101 in linkage group 3; and DXS10146, DXS10134, & DXS7423 in linkage group 4. Physical locations across the chromosome are shown in Figure 6.5 [82].

**Figure 6.5. Physical location of 12 markers included in Investigator Argus X-12 Kit.** Three closely-located markers represent each of the four proposed linkage groups. Figure reproduced from [82].




These 12 markers are simple, compound, and complex repeat motifs, and exhibit allele ranges that include a variety of microvariants; their characteristics are detailed in Table 6.3. The observed allele range for each marker highlights the number and type of microvariants observed in U.S. populations.

**Table 6.3. Characteristics of the 12 X STR markers of the Investigator Argus X-12 kit.** The observed allele range was based upon study of the four U.S. populations. The variables "x" and "y" indicate repeat units that are present in different numbers in different alleles, i.e. the source of length variation. Additional lower case characters represent bases that fall within the repeat region, but do not contribute to the total repeat count defining the allele call.

| Marker | Observed Allele Range | Repeat Motif |
|---|---|---|
| DXS8378 | 9-14 | CTAT |
| HPRTB | 8-16 | AGAT |
| DXS7132 | 11-17; 15.3, 16.3 | $(TCTA)_x$-$(TCA)_{(0-1)}$-$(TCTA)_2$ |
| DXS7423 | 8, 12-17 | $(TCCA)_3$-$(TCTGTCCT)_{(0-1)}$-$(TCCA)_x$ |
| DXS10103 | 15-21 | $(TAGA)_{(1-2)}$-CTGACAGA-$(TAGA)_x$-$(CAGA)_4$-TAGA |
| DXS10079 | 13-24 | $(AGAA)_x$-AGAG-$(AGAA)_y$ |
| DXS10074 | 7-22; 14.2, 19.2; 11.3, 15.3, 16.3 | $(AA)_{(0-1)}$-$(AAGA)_x$-$(AAGG)_{(0-1)}$-$(AAGA)_{(0-2)}$ |
| DXS10101 | 23, 25-35, 37; 24.2-33.2 | $(AAAG)_x$-GAAAGAAG-$(GAAA)_3$-A-$(GAAA)_4$-AAGA-$(AAAG)_5$-[aaaaagaa-$(AAAG)_4$]$_y$-$(AAAG)_z$-$(AA)_{(0-1)}$ |
| DXS10135 | 15-35, 38; 17.1-25.1, 27.1, 28.1, 34.1-36.1; 35.2, 38.2, 39.2 | $(AAGA)_3$-GAAAG-$(GAAA)_x$ |
| DXS10148 | 14, 16-32; 20.1-32.1, 35.1; 27.2, 29.2, 32.2; 13.3, 26.3 | $(GGAA)_x$-$(AAGA)_y$-$(AAAG)_4$-AAGGAAAG-$(AAGG)_2$-AAAGAAGG |
| DXS10146 | 23-35, 41; 39.1; 29.2-36.2, 38.2-47.2; 42.3 | $(TTCC)_x$-T-$(TTCC)_4$-TCCCTTCC-$(TCCC)_2$-TTCTTCTTTC-$(TTCC)_2$-TTTCTT-$(TT)_{(0-1)}$-$(CTTT)_y$-T-$(CTTT)_2$ |
| DXS10134 | 28-41; 28.1, 30.1, 32.1, 35.1, 36.1; 33.2, 35.2, 37.2, 39.2; 33.3, 37.3-44.3 | $(GAAA)_3$-GAGA-$(GAAA)_4$-AAGAAAGAGA-$(GAAA)_4$-GAGA-$(GACAGA)_{(2-3)}$-GAAAGTAA-$(GAAA)_3$-AAA-$(GAAA)_4$-AAA-$(GAAA)_x$ |

*6.2.1. Off-ladder alleles in U.S. populations*

The complex repeat patterns of a majority of the markers lend themselves to a higher frequency of microvariation, and a robust, validated capillary electrophoresis protocol must be maintained to detect the frequent 1 bp differences between alleles. A generally comprehensive allelic ladder is used for ease of interpretation, and an example of this ladder is shown in Figure 6.6. Notable deficiencies exist in the allelic ladder between alleles 35.2 & 39.2 for DXS10146 and between 13.3 & 18 and 31 & 38.1 for DXS10148, where 3.3% (32) of the alleles observed in this study fell. A total of 5.8% (15) of the African American samples were affected by the gap at DXS10146 alone. Tables 6.4 to 6.6 summarize the off-ladder alleles (OLAs) observed in the U.S. dataset, where 80 unique OLAs were observed 334 times in total. Since the majority of the validation testing performed using the commercial kit and responsible for the current composition of the allelic ladder employed European populations, the frequency of OLAs was population-dependent: 55% of all OLAs

were observed in African American samples. Fourty-two (52.5%) OLAs were observed more than once, with the most common (DXS10148-24) observed 44 times in 3 populations (African American, U.S. Caucasian, and U.S. Hispanic). More details on the frequency of individual OLAs can be found in the allele frequency tables for these makers in Chapter 4, Section 4.1.4.

**Figure 6.6. Example of Investigator Argus X-12 allelic**

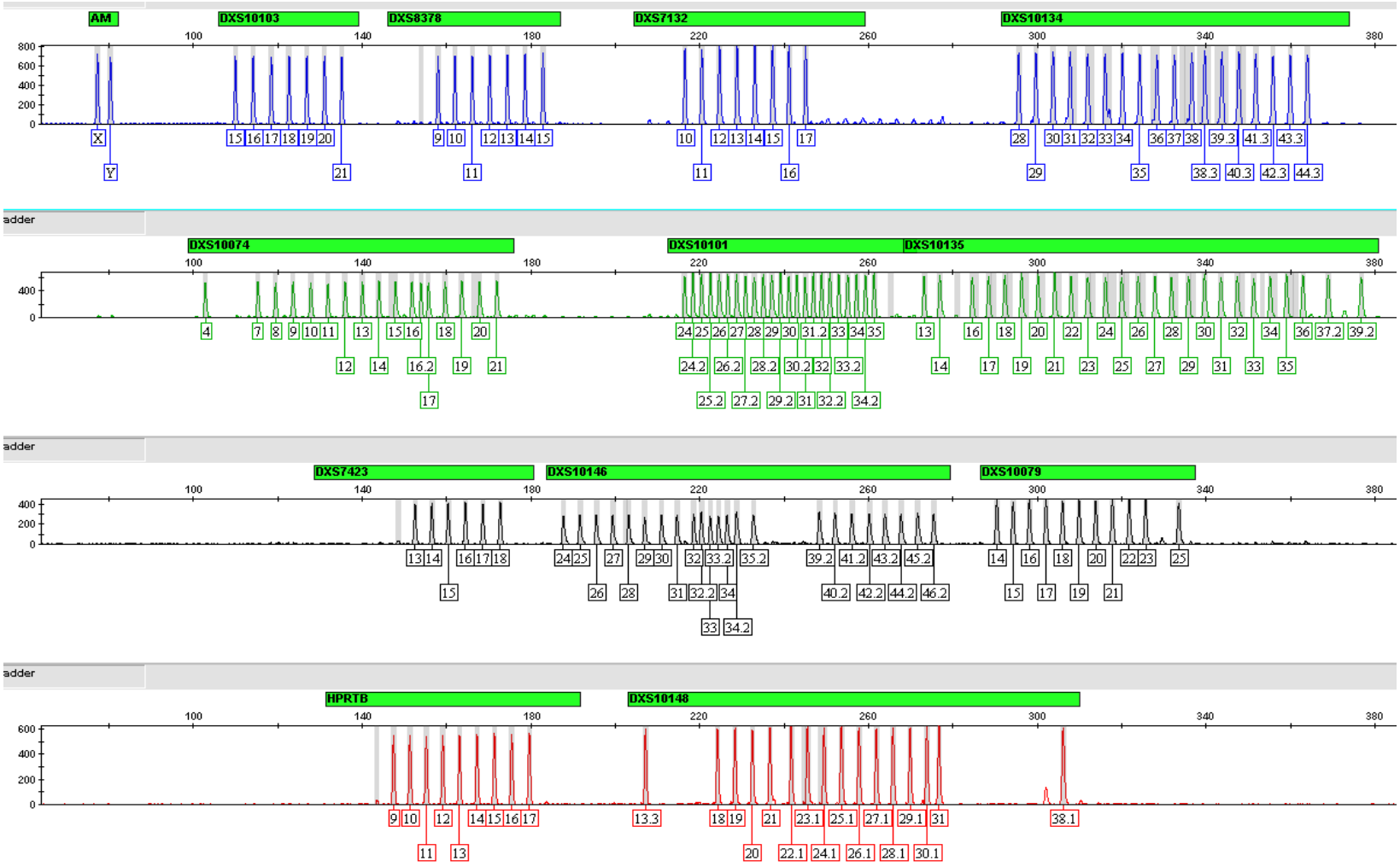


**Table 6.4. Off-ladder alleles observed using Investigator Argus X-12 kit and U.S. populations.**

| Marker | Off-ladder alleles |
|---|---|
| DXS10101 | 23, 37 |
| DXS10079 | 13, 24 |
| DXS7132 | 15.3, 16.3 |
| DXS7423 | 8, 12 |
| HPRTB | 8 |
| DXS10146 | 23, 29.2-31.2, 35, 36.2, 38.2, 39.1, 41, 42.3, 47.2 |
| DXS10148 | 14, 16, 17, 20.1, 21.1, 22-26, 26.3, 27, 27.2, 28, 29, 29.2, 30, 31.1, 32, 32.2, 35.1 |
| DXS10134 | 28.1, 30.1, 32.1, 33.2, 33.3, 35.1, 35.2, 36.1, 37.2, 37.3, 39, 39.2, 40, 41 |
| DXS10135 | 15, 17.1-25.1, 27.1, 28.1, 34.1, 35.1, 35.2, 36.1, 38, 38.2 |
| DXS10074 | 11.3, 14.2, 14.3, 15.3, 16.3, 19.2, 22 |

**Table 6.5. Number of unique off-ladder alleles observed by marker and population.** AA: African American; AS: U.S. Asian; CN: U.S. Caucasian; Hisp: U.S. Hispanic.

| Marker | AA | AS | CN | Hisp | Overall |
|---|---|---|---|---|---|
| DXS10101 | 1 | 0 | 1 | 0 | 2 |
| DXS10079 | 2 | 0 | 1 | 1 | 2 |
| DXS7132 | 0 | 0 | 0 | 2 | 2 |
| DXS7423 | 2 | 0 | 0 | 0 | 2 |
| HPRTB | 1 | 0 | 0 | 0 | 1 |
| DXS10146 | 9 | 0 | 2 | 4 | 11 |
| DXS10148 | 15 | 6 | 5 | 5 | 21 |
| DXS10134 | 8 | 5 | 5 | 6 | 14 |
| DXS10135 | 17 | 0 | 10 | 6 | 18 |
| DXS10074 | 3 | 3 | 1 | 1 | 7 |
| Total | 58 | 14 | 25 | 25 | 80 |

**Table 6.6. Total number of off-ladder alleles observed by marker and population.** AA: African American; AS: U.S. Asian; CN: U.S. Caucasian; Hisp: U.S. Hispanic.

| Marker | AA | AS | CN | Hisp | Overall |
|---|---|---|---|---|---|
| DXS10101 | 1 | 0 | 1 | 0 | 2 |
| DXS10079 | 6 | 0 | 1 | 1 | 8 |
| DXS7132 | 0 | 0 | 0 | 2 | 2 |
| DXS7423 | 6 | 0 | 0 | 0 | 6 |
| HPRTB | 1 | 0 | 0 | 0 | 1 |
| DXS10146 | 23 | 0 | 3 | 7 | 33 |
| DXS10148 | 53 | 8 | 27 | 13 | 101 |
| DXS10134 | 11 | 21 | 15 | 9 | 56 |
| DXS10135 | 76 | 0 | 25 | 13 | 114 |
| DXS10074 | 6 | 3 | 1 | 1 | 11 |
| Total | 183 | 32 | 73 | 46 | 334 |

A number of the observed OLAs were sequenced as part of this study, and the details can be found in the following section.

*6.2.2. Allele Sequencing*

A subset of alleles for most of the twelve markers included in this commercial kit was sequenced in order to confirm repeat structure. In the case of microvariant or null alleles, sequencing was often necessary to verify the true allele call. Like the number of microvariants present in this marker set, the number of null alleles was also large in the studied populations (Table 6.7). Particularly in the African American population, null alleles presented a real problem for 29% of the total number of alleles analyzed. In addition to the null alleles noted in Table 6.7, eight samples exhibited alleles with reduced peak height due to a primer binding site mutation at DXS7130, HPRTB, or DXS8378. A small number of additional null alleles may be possible also, appearing as homozygous females. However, only 2.6% of the total number of alleles typed have the potential to remain undetected in this manner. Additional information on the frequency of null alleles in these populations can be found in Table 4.22.

**Table 6.7. Null alleles observed using Investigator Argus X-12 kit with U.S. populations.** AA: African American; AS: U.S. Asian; CN: U.S. Caucasian; Hisp: U.S. Hispanic.

| Marker | AA | AS | CN | Hisp | Total |
|---|---|---|---|---|---|
| DXS10146 | 19 (7%) | 0 | 0 | 3 (2%) | 22 (3%) |
| DXS10148 | 61 (23%) | 0 | 0 | 2 (1%) | 63 (7%) |
| DXS7132 | 0 | 1 (1%) | 0 | 0 | 1 (0.1%) |
| Total | 76 (29%) | 1 (1%) | 0 | 5 (4%) | 82 (10%) |

In particular, marker DXS10148 exhibited a large proportion of null alleles in the African American population. Therefore, fifty-six samples with DXS10148 nulls were sequenced, representing 13 unique alleles, and the results are shown in Table 6.8. Of note, there is a wide variation in the composition of each individual allele; 40 unique sequence structures were noted for only 13 total allele calls by fragment analysis. Additionally, individual bases (typically an adenine) are interspersed

within the repeat units, resulting in microvariants. One of the sequenced alleles was also an off-ladder allele, 35.1; its repeat structure appears to follow the same patterns as the other X.1 microvariant alleles. In general, much of the variation in these sequenced alleles resulted from the internal AAGA and AAAG repeats found at the middle of the repeat structure. Given that the individual repeat units are composed of a combination of adenine and guanine residues, the potential for *in vivo* polymerase slippage during replication seems a likely cause of the large amount of variation and number of allele calls present for this marker.

**Table 6.8. Sequence structure of fifty-six DXS10148 null alleles observed within U.S. populations.** The italicized 22 allele is the Qiagen reference allele defined in the kit manual. Allele 35.1 (shaded) is also an off-ladder allele. N: number of alleles sequenced. $N_8$: AAGGAAAG, not included in repeat count for allele designation.

| Allele | Repeat Structure | N |
|---|---|---|
| *22* | *[GGAA]$_4$-[AAGA]$_{12}$-[AAAG]$_4$-N$_8$-[AAGG]$_2$* | |
| 23 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{15}$-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 1 |
| 24 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{16}$-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 1 |
| 25 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{17}$-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 1 |
| 32.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{15}$-[AAAG]$_9$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 1 |
| 32.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{15}$-[AAAG]$_{13}$-A-[AAAG]$_4$-N$_8$ | 1 |
| 33 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{16}$-[AAAG]$_{19}$-N$_8$-[AAGG]$_2$ | 1 |
| 35.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{12}$-[AAAG]$_{10}$-ATAG-[AAAG]$_4$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 1 |
| 35.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{14}$-[AAAG]$_{13}$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 1 |
| 35.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{15}$-[AAAG]$_{12}$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 2 |
| 36.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{13}$-[AAAG]$_{10}$-ATAG-[AAAG]$_4$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 1 |
| 36.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{13}$-[AAAG]$_{15}$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 1 |
| 36.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{14}$-[AAAG]$_{14}$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 2 |
| 36.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{15}$-[AAAG]$_{13}$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 2 |
| 36.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{16}$-[AAAG]$_{12}$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 2 |
| 37.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{15}$-[AAAG]$_{14}$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 3 |
| 37.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{15}$-[AAAG]$_9$-ATAG-[AAAG]$_4$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 2 |
| 37.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{16}$-[AAAG]$_{13}$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 2 |
| 38.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{15}$-[AAAG]$_{10}$-ATAG-[AAAG]$_4$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 1 |
| 38.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{15}$-[AAAG]$_{15}$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 2 |
| 38.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{16}$-[AAAG]$_{14}$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 4 |
| 38.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{16}$-[AAAG]$_9$-ATAG-[AAAG]$_4$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 1 |
| 38.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{17}$-[AAAG]$_{13}$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 2 |
| 38.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{18}$-[AAAG]$_{12}$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 1 |
| 39.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{15}$-[AAAG]$_{11}$-ATAG-[AAAG]$_4$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 1 |
| 39.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{16}$-[AAAG]$_{13}$-AGAG-AAAG-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 1 |
| 39.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{16}$-[AAAG]$_{15}$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 1 |
| 39.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{17}$-[AAAG]$_{14}$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 2 |
| 39.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{17}$-[AAAG]$_9$-ATAG-[AAAG]$_4$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 3 |
| 39.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{18}$-[AAAG]$_{13}$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 1 |
| 39.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{18}$-[AAAG]$_8$-ATAG-[AAAG]$_4$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 1 |
| 40.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{16}$-[AAAG]$_{16}$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 2 |
| 40.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{17}$-[AAAG]$_{10}$-ATAG-[AAAG]$_4$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 1 |
| 40.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{17}$-[AAAG]$_{15}$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 2 |
| 40.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{18}$-[AAAG]$_{14}$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 2 |
| 40.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{18}$-[AAAG]$_9$-ATAG-[AAAG]$_4$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 1 |
| 40.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_5$-AATA-[AAGA]$_{11}$-[AAAG]$_{15}$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 1 |
| 41.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{17}$-[AAAG]$_{16}$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 1 |
| 41.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{18}$-[AAAG]$_{15}$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 2 |
| 41.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{19}$-[AAAG]$_{14}$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 1 |
| 41.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{19}$-[AAAG]$_9$-ATAG-[AAAG]$_4$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 1 |
| 43.1 | [GGAA]$_2$-AGAAGGAA-[AAGA]$_{17}$-[AAAG]$_{18}$-A-[AAAG]$_4$-N$_8$-[AAGG]$_2$ | 1 |

Forty-nine alleles were sequenced at marker DXS10146, 22 of which were null alleles when amplified with the kit (Table 6.9). Like DXS10148, the sequence structure was found to be highly complex, with individual bases responsible for microvariation scattered throughout. Fifteen alleles were represented by this sequencing according to the allele call from fragment-based analysis, but 22 unique allele structures were observed. Four of these unique sequence structures were confined to null alleles (those in bold). However, certain structures were present in both amplified and null alleles. The 31 allele structure that was sequenced 4 times was found in 3 null alleles and one that amplified normally. Similarly, both the 32 allele structure sequenced 2 times and the 34 allele were observed in one null allele each.

**Table 6.9. Sequence structure of forty-nine DXS10146 alleles observed within U.S. populations.** Shading indicates off-ladder alleles. N: number of alleles sequenced. The bolded structures were only found in alleles that were also null with the kit. $N_4$: TTCC; $N_6$: TTTCTT; $N_{10}$: TTCTTCTTTC, none of which are included in repeat count for allele designation.

| Allele | Repeat Structure | N |
|---|---|---|
| 23 | $[TTCC]_3$--T-$[TTCC]_3$-TTC-ctccc-$N_4$-$[TCCC]_2$-$N_{10}$-$[TTCC]_2$-$N_6$-$[CTTT]_{10}$-t-$[CTTT]_2$ | 2 |
| 24 | $[TTCC]_3$-T-$[TTCC]_3$-TTC-ctccc-$N_4$-$[TCCC]_2$-$N_{10}$-$[TTCC]_2$-$N_6$-$[CTTT]_{11}$-t-$[CTTT]_2$ | 2 |
| **24** | **$[TTCC]_6$-TTTC-cttcc-$N_4$-[TTCC]-[TCCC]-[TTCC]-$[TCCC]_2$-$N_{10}$-$[TTCC]_2$-$N_6$-$[CTTT]_8$-t-$[CTTT]_2$** | **11** |
| 25 | $[TTCC]_3$-T-$[TTCC]_3$-TTC-ctccc-$N_4$-$[TCCC]_2$-$N_{10}$-$[TTCC]_2$-$N_6$-$[CTTT]_{12}$-t-$[CTTT]_2$ | 2 |
| **25** | **$[TTCC]_6$-TTTC-cttcc-$N_4$-[TTCC]-[TCCC]-[TTCC]-$[TCCC]_2$-$N_{10}$-$[TTCC]_2$-$N_6$-$[CTTT]_9$-t-$[CTTT]_2$** | 4 |
| **25** | **$[TTCC]_6$-TTTC-cttcc-$N_4$-[TTCC]-[TCCC]-[TTCC]-$[TCCC]_2$-$N_{10}$-$[TTCC]_2$-$N_6$-$[CTTT]_5$-CTTA-$[CTTT]_3$-t-$[CTTT]_2$** | 1 |
| 26 | $[TTCC]_3$-T-$[TTCC]_3$-TTC-ctccc-$N_4$-$[TCCC]_2$-$N_{10}$-$[TTCC]_2$-$N_6$-$[CTTT]_{13}$-t-$[CTTT]_2$ | 2 |
| 27 | $[TTCC]_3$-T-$[TTCC]_3$-TTC-ctccc-$N_4$-$[TCCC]_2$-$N_{10}$-$[TTCC]_2$-$N_6$-$[CTTT]_{14}$-t-$[CTTT]_2$ | 2 |
| **27** | **$[TTCC]_6$-TTTC-cttcc-$N_4$-[TTCC]-[TCCC]-[TTCC]-$[TCCC]_2$-$N_{10}$-$[TTCC]_2$-$N_6$-$[CTTT]_{11}$-t-$[CTTT]_2$** | **1** |
| 28 | $[TTCC]_6$-TTTC-cttcc-$N_4$-[TTCC]-[TCCC]-[TTCC]-$[TCCC]_2$-$N_{10}$-$[TTCC]_2$-$N_6$-$[CTTT]_{12}$-t-$[CTTT]_2$ | 1 |
| 28 | $[TTCC]_3$-T-$[TTCC]_3$-TTC-ctccc-$N_4$-$[TCCC]_2$-$N_{10}$-$[TTCC]_2$-$N_6$-$[CTTT]_{15}$-t-$[CTTT]_2$ | 1 |
| 29 | $[TTCC]_3$-T-$[TTCC]_3$-TTC-ctccc-$N_4$-$[TCCC]_2$-$N_{10}$-$[TTCC]_2$-$N_6$-$[CTTT]_{16}$-t-$[CTTT]_2$ | 2 |
| 29.2 | $[TTCC]_3$-T-$[TTCC]_3$-TTC-ctccc-$N_4$-$[TCCC]_2$-$N_{10}$-$[TTCC]_2$-$N_6$-TT-$[CTTT]_{16}$-t-$[CTTT]_2$ | 3 |
| 30 | $[TTCC]_3$-T-$[TTCC]_3$-TTC-ctccc-$N_4$-$[TCCC]_2$-$N_{10}$-$[TTCC]_2$-$N_6$-$[CTTT]_{17}$-t-$[CTTT]_2$ | 1 |
| 30.2 | $[TTCC]_3$-T-$[TTCC]_3$-TTC-ctccc-$N_4$-$[TCCC]_2$-$N_{10}$-$[TTCC]_2$-$N_6$-TT-$[CTTT]_{17}$-t-$[CTTT]_2$ | 1 |
| 31 | $[TTCC]_3$-T-$[TTCC]_3$-TTC-ctccc-$N_4$-$[TCCC]_2$-$N_{10}$-$[TTCC]_2$-$N_6$-$[CTTT]_{18}$-t-$[CTTT]_2$ | 1 |
| 31 | $[TTCC]_6$-TTTC-cttcc-$N_4$-[TTCC]-[TCCC]-[TTCC]-$[TCCC]_2$-$N_{10}$-$[TTCC]_2$-$N_6$-$[CTTT]_{15}$-t-$[CTTT]_2$ | 4 |
| 31.2 | $[TTCC]_3$-T-$[TTCC]_3$-TTC-ctccc-$N_4$-$[TCCC]_2$-$N_{10}$-$[TTCC]_2$-$N_6$-TT-$[CTTT]_{18}$-t-$[CTTT]_2$ | 1 |
| 32 | $[TTCC]_3$-T-$[TTCC]_3$-TTC-ctccc-$N_4$-$[TCCC]_2$-$N_{10}$-$[TTCC]_2$-$N_6$-$[CTTT]_{19}$-t-$[CTTT]_2$ | 1 |
| 32 | $[TTCC]_6$-TTTC-cttcc-$N_4$-[TTCC]-[TCCC]-[TTCC]-$[TCCC]_2$-$N_{10}$-$[TTCC]_2$-$N_6$-$[CTTT]_{16}$-t-$[CTTT]_2$ | 2 |
| 33 | $[TTCC]_6$-TTTC-cttcc-$N_4$-[TTCC]-[TCCC]-[TTCC]-$[TCCC]_2$-$N_{10}$-$[TTCC]_2$-$N_6$-$[CTTT]_{17}$-t-$[CTTT]_2$ | 1 |
| 34 | $[TTCC]_6$-TTTC-cttcc-$N_4$-[TTCC]-[TCCC]-[TTCC]-$[TCCC]_2$-$N_{10}$-$[TTCC]_2$-$N_6$-$[CTTT]_{18}$-t-$[CTTT]_2$ | 3 |

Though the primers used within the Investigator Argus X-12 kit are not published, a limited number of primer pairs have been validated for use, one of which corresponds to the DXS10146 amplicon lengths obtained using the kit [236]. Indeed, sequencing results revealed the presence of a 4 bp deletion in the middle of this putative reverse primer binding site specific to samples exhibiting null alleles for this marker (Figure 6.7).

**Figure 6.7. Allele sequencing results comparing the putative reverse primer binding sites of null and non-null alleles observed in U.S. populations.** A 4-bp CTTT deletion present 10 bp downstream of the repeat motif was present in all null samples that were sequenced (N = 22) but was lacking in all other (non-null) samples (N = 30). Boxed sequence indicates published DXS10146 reverse amplification primer [236].


Besides markers DXS10146 and DXS10148, one null allele was observed in a U.S. Asian sample for marker DXS7132 using the commercial kit. Sequencing results for this allele are presented in Section 6.2.3 as part of a discussion of concordance.

In addition to confirmation of allele calls for null alleles, routine sequencing of standard alleles was undertaken for markers DXS10079 (Table 6.10) and DXS10101 (Table 6.11). Eleven alleles sequenced for DXS10079 revealed a compound repeat structure composed of AGAA and AGAG units. The variation, however, could be wholly attributed to the leading AGGA repeat units, which varied in number while the AGAG and trailing AGAA repeats did not. Further study would be necessary to confirm the stability of the last 4 repeats.

**Table 6.10. Sequence structure of eleven standard DXS10079 alleles observed within U.S. populations.** The italicized 21 allele is the Qiagen reference allele defined in the kit manual. N: number of alleles sequenced.

| Allele | Repeat structure | N |
|---|---|---|
| *21* | *$[AGAA]_{17}$-AGAG-$[AGAA]_3$* | |
| 13 | $[AGAA]_9$-AGAG-$[AGAA]_3$ | 2 |
| 14 | $[AGAA]_{10}$-AGAG-$[AGAA]_3$ | 1 |
| 17 | $[AGAA]_{13}$-AGAG-$[AGAA]_3$ | 1 |
| 18 | $[AGAA]_{14}$-AGAG-$[AGAA]_3$ | 1 |
| 19 | $[AGAA]_{15}$-AGAG-$[AGAA]_3$ | 1 |
| 20 | $[AGAA]_{16}$-AGAG-$[AGAA]_3$ | 1 |
| 23 | $[AGAA]_{19}$-AGAG-$[AGAA]_3$ | 1 |
| 24 | $[AGAA]_{20}$-AGAG-$[AGAA]_3$ | 2 |
| 22 | $[AGAA]_{17}$-AGAG-$[AGAA]_3$ | 1 |

The structure observed for marker DXS10101 revealed a complex repeat unit with nonvariable regions present throughout. Only six alleles were sequenced as part of this study, mainly to confirm off-ladder allele structure, and examples from an additional study [171] were added for comparison. The composition of the repeat structures found in the present study, including the off-ladder alleles, follows that of the published study.

**Table 6.11. Sequence structure of ten DXS10101 alleles.** Both structures observed in this study in U.S. populations and those noted in the literature were compiled in this table. The italicized 28.2 allele is the Qiagen reference allele defined in the kit manual. Shading indicates off-ladder alleles. N: number of alleles sequenced (this study only). $N_4$: AAGA; $N_8$: GAAAGAAG, none of which are included in the repeat count for allele designation.

| Ref. | Allele | Repeat Structure | N |
|---|---|---|---|
| This study | 23 | $[AAAG]_3$-$N_8$-$[GAAA]_3$-a-$[GAAA]_4$-$N_4$-$[AAAG]_5$-aaaaagaa-$[AAAG]_4$-$[AAAG]_4$ | 1 |
| [171] | 26 | $[AAAG]_3$-$N_8$-$[GAAA]_3$-a-$[GAAA]_4$-$N_4$-$[AAAG]_5$-aaaaagaa-[AAAG]-AA-$[AAAG]_9$-AA | |
| [82] | *28.2* | *$[AAAG]_3$-$N_8$-$[GAAA]_3$-a-$[GAAA]_4$-$N_4$-$[AAAG]_5$-aaaaagaa-$[AAAG]_4$-$[AAAG]_9$-AA* | |
| This study | 29 | $[AAAG]_3$-$N_8$-$[GAAA]_3$-a-$[GAAA]_4$-$N_4$-$[AAAG]_5$-aaaaagaa-$[AAAG]_4$-$[AAAG]_{10}$ | 1 |
| This study | 31 | $[AAAG]_2$-$N_8$-$[GAAA]_3$-a-$[GAAA]_4$-$N_4$-$[AAAG]_5$-aaaaagaa-$[AAAG]_4$-$[AAAG]_{13}$ | 1 |
| This study | 32 | $[AAAG]_3$-$N_8$-$[GAAA]_3$-a-$[GAAA]_4$-$N_4$-$[AAAG]_5$-aaaaagaa-$[AAAG]_4$-$[AAAG]_{12}$ | 1 |
| This study | 34 | $[AAAG]_3$-$N_8$-$[GAAA]_3$-a-$[GAAA]_4$-$N_4$-$[AAAG]_5$-aaaaagaa-$[AAAG]_4$-$[AAAG]_{15}$ | 1 |
| [171] | 34.2 | $[AAAG]_3$-$N_8$-$[GAAA]_3$-a-$[GAAA]_4$-$N_4$-$[AAAG]_5$-aaaaagaa-$[AAAG]_4$-$[AAAG]_{15}$-AA | |
| [171] | 35 | $[AAAG]_3$-$N_8$-$[GAAA]_3$-a-$[GAAA]_4$-$N_4$-$[AAAG]_5$-aaaaagaa-$[AAAG]_4$-$[AAAG]_{16}$ | |
| This study | 37 | $[AAAG]_3$-$N_8$-$[GAAA]_3$-a-$[GAAA]_4$-$N_4$-$[AAAG]_5$-(aaaaagaa-$[AAAG]_4)_2$-$[AAAG]_{12}$ | 1 |

*6.2.3. Concordance study*

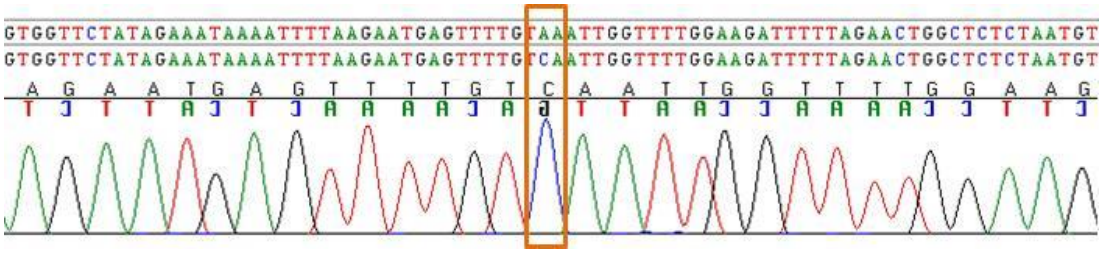
A total of 3900 alleles were examined for concordance across the 4 markers overlapping with the set studied here: DXS7132, DXS7423, DXS8378, and HPRTB. Three discordant alleles were noted (Table 6.12), resulting in 99.92% concordance. Only one discordant allele call resulted from a null allele in a profile amplified with the commercial kit: the null 17 allele at marker DXS7132 mentioned previously.

**Table 6.12. Summary of discordant allele calls.**

| Population | Marker | X-12 profile | Mini-STR profile |
|---|---|---|---|
| U.S. Asian | DXS7132 | Null | 17 |
| African American | DXS7132 | 14 | Null |
| U.S. Caucasian | HPRTB | 12,14 | 14,14 |

All instances of discordance were sequenced to determine the probable cause. To begin, the sequencing results for the U.S. Asian sample exhibiting discordance are shown in Figure 6.8. Sequencing results revealed a standard 17 allele with an A→C mutation 95 bp downstream of the repeat motif. Again, though the commercial kit primers are not published, it could be postulated that the observed A→C mutation may fall at the 3' end of the reverse primer binding site, causing the resultant null allele. Fortunately, this mutation appears to be relatively rare, occurring only once in 975 alleles typed as part of this study.

**Figure 6.8. DXS7132 sequencing results for the discordant U.S. Asian sample.** An A→C mutation under the putative reverse primer binding site is surrounded with an orange box.



Mutations resulting in the two additional null alleles could be localized to the appropriate primer binding sites since primer sequences for this multiplex was known. A C→T transition resulted in complete loss of amplification at marker DXS7132 in an African American sample (Figure 6.9A). This mutation has been observed only once in a total of 2216 (0.045%) U.S. population samples processed using this mini-STR primer set. Additionally, this mutation has not been reported to occur in published populations. The AG deletion falling under the reverse primer binding site of marker HPRTB (Figure 6.9B), however, was originally defined in 1999 by Mertens, *et al.* [295]. With amplicons of a larger size, this deletion may fall within the flanking region rather than the primer binding site, resulting in an apparent X.2 microvariant instead of a null allele. Such was the case in one U.S. population study [70] as well as a variety of other published worldwide population samples [107,113,116,118,121,124,125,165,167,173,176,292].

**Figure 6.9. Sequencing results for primer binding sites involved in mini-STR null alleles.** (A) DXS7132 forward primer binding site mutation. (B) HPRTB reverse primer binding site mutation. Sequence direction is 5'→3'.

**A.**


**B.**


The Investigator Argus X12 Kit provides reliable amplification and separation of 12 X STR markers plus amelogenin in one multiplex reaction. This study represents the first time its use on U.S. samples has been examined, and the markers proved to be highly informative for U.S. populations (see Chapter 4 for more details). One consideration for the kit's use with U.S. populations is the high rate of null and off-ladder alleles observed, particularly for the African American population group. The potential for mistyping due to missing or incorrectly assigned allele calls increases

when female samples are considered. Further study of the null and off-ladder alleles observed in these populations as well as additional considerations such as the mutation rate and linkage situation will be necessary for routine forensic use of the Investigator Argus X-12 kit.

# Chapter 7. Further Applications: Mixture Multiplex

*7.1. Assay development*

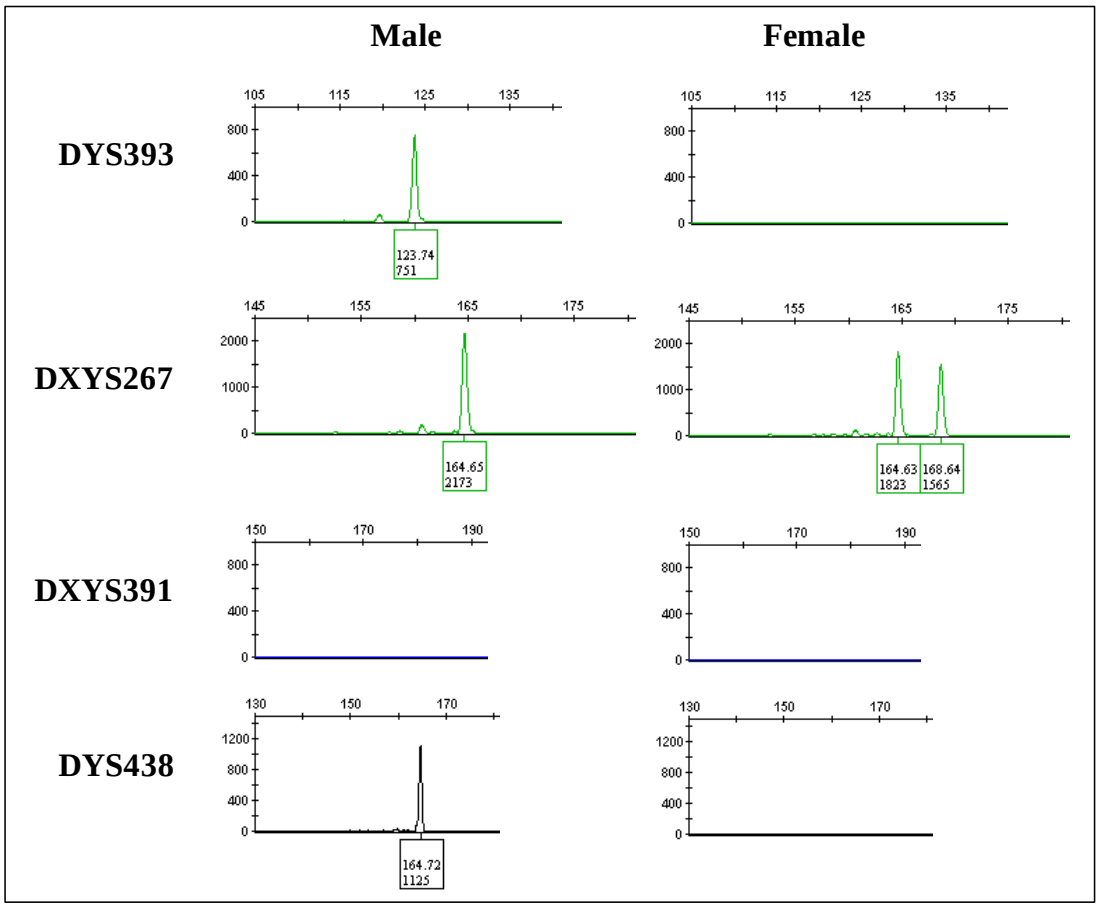
A list of a subset of the markers that were considered for inclusion in the MIXplex, along with details of those that were chosen, appears in Table 7.1. Other markers were considered but excluded due to some combination of undesirable factors including but not limited to: a complex repeat structure (DYS390, DYS635); published amplicons that were too large or too small (DYS19, DYS390, DYS439, DYS635, DYS392, Y-GATA-H4, DYS448); and/or unresolved allele nomenclature issues (DYS439, Y-GATA-H4).

**Table 7.1. List of potential STR markers for inclusion in a mixture multiplex.** In addition to the markers characterised for inclusion in the mini-X STR multiplexes, Y and XY markers were needed for the mixture multiplex. Below are markers that were considered, and their selection-relevant characteristics. F: forward; R: reverse; x: number of repeats; bp: base pairs; $T_m$: melting temperature; Ref: reference.

| Marker | Repeat motif | Amplicon size (bp) | Amplification Primer (5'→3') | $T_m$ (°C) | Ref. |
|---|---|---|---|---|---|
| **DYS393** | $AGAT_x$ | 107-139 | F GTGGTCTTCTACTTGTGTCAATAC | 54.7 | [46] |
| | | | R AACTCAAGTCCAAAAAATGAGG | 57.4 | |
| **DXYS267** | TATA-$GATA_x$-GACA-GATA | 147-179 | F GTGGTCTTCTACTTGTGTCAATAC | 54.7 | [320] |
| | | | R CTAAATAAAAGTCATATCAGCTGC | 53.1 | |
| **DXYS391** | $TCTA_x$ | 151-207 | F TTCATTCAATCATACACCCATATC | 57.8 | [321] |
| | | | R GGAATAAAATCTCCCTGGTTG | 57.5 | |
| **DYS438** | $TTTTC_x$ | 133-173 | F TGGGGAATAGTTGAACGGTAA | 59.3 | [249] |
| | | | R GGAGGTTGTGGTGAGTCGAG | 60.7 | |
| **DYS437** | $TCTA_x$-$(TCTG)_2$-$(TCTA)_4$ | 181-197 | F GACTATGGGCGTGAGTGCAT | 61.1 | [48] |
| | | | R AGACCCTGTCATTCACAGATGA | 59.6 | |
| **DYS458** | $GAAA_x$ | 132-160 | F GCAACAGGAATGAAACTCCAAT | 60.4 | [262] |
| | | | R GTTCTGGCATTACAAGCATGAG | 57.5 | |
| **DYS391** | $TCTA_x$ | 147-179 | F CTATTCATTCAATCATACACCCATAT | 57.5 | [46] |
| | | | R ACATAGCCAAATATCTCCTGGG | 59.4 | |
| **DYS456** | $AGAT_x$ | 137-161 | F GGACCTTGTGATAATGTAAGATA | 52.8 | [49] |
| | | | R CCCATCAACTCAGCCCAAAAC | 63.9 | |

Singleplex amplifications were performed first to evaluate primer selection for successful amplification, complete adenylation, and peak migration (Figure 7.1). The first version of the MIXplex targeted GATA172D05 and DXYS391 in the blue channel, DYS393 and DXYS267 in the green channel, and SRY, GATA31E08, and DYS438 in the yellow channel. All primer sets exhibited expected amplification for both males and females except DXYS391.

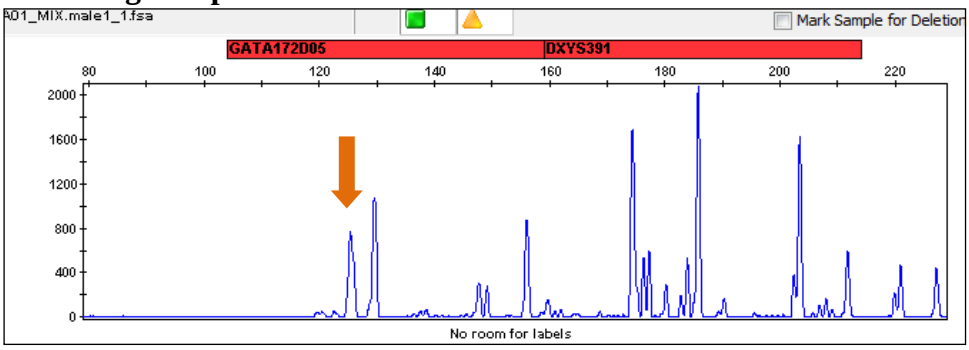
**Figure 7.1. Singleplex testing results.** Candidate primer sets that had not been previously evaluated during the development of the X STR multiplexes were tested in singleplex with two male and three female samples. Here, representative male and female profiles at each marker are shown.



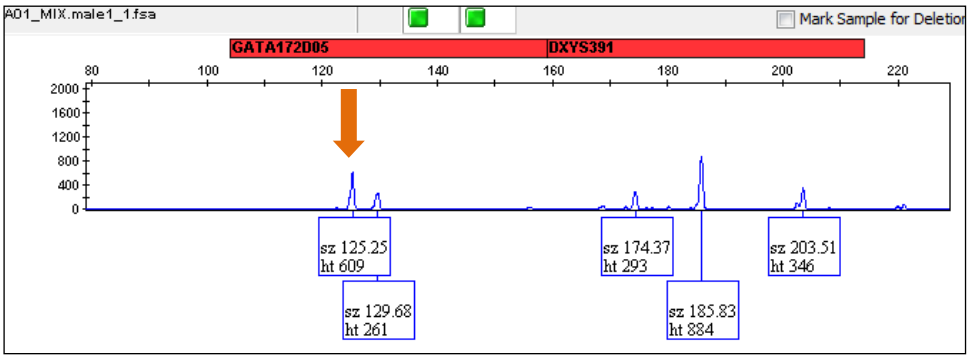

When the primer sets were combined for multiplex amplification, extraneous peaks appeared in the 6-FAM™ (blue) channel, obscuring the true allele peaks for the samples despite alteration of the annealing temperature (Figure 7.2).

**Figure 7.2. First version of a mixture multiplex amplified at both 55 °C (top) and 60 °C (bottom).** The 6-FAM™ (blue) channel exhibited multiple non-specific peaks at both annealing temperatures used. Authentic GATA172D05 allele peak in each electropherogram is indicated with an arrow. All other peaks (green and yellow channels; not shown) appeared as expected.

**Annealing Temperature: 55 °C**




**Annealing Temperature: 60 °C**

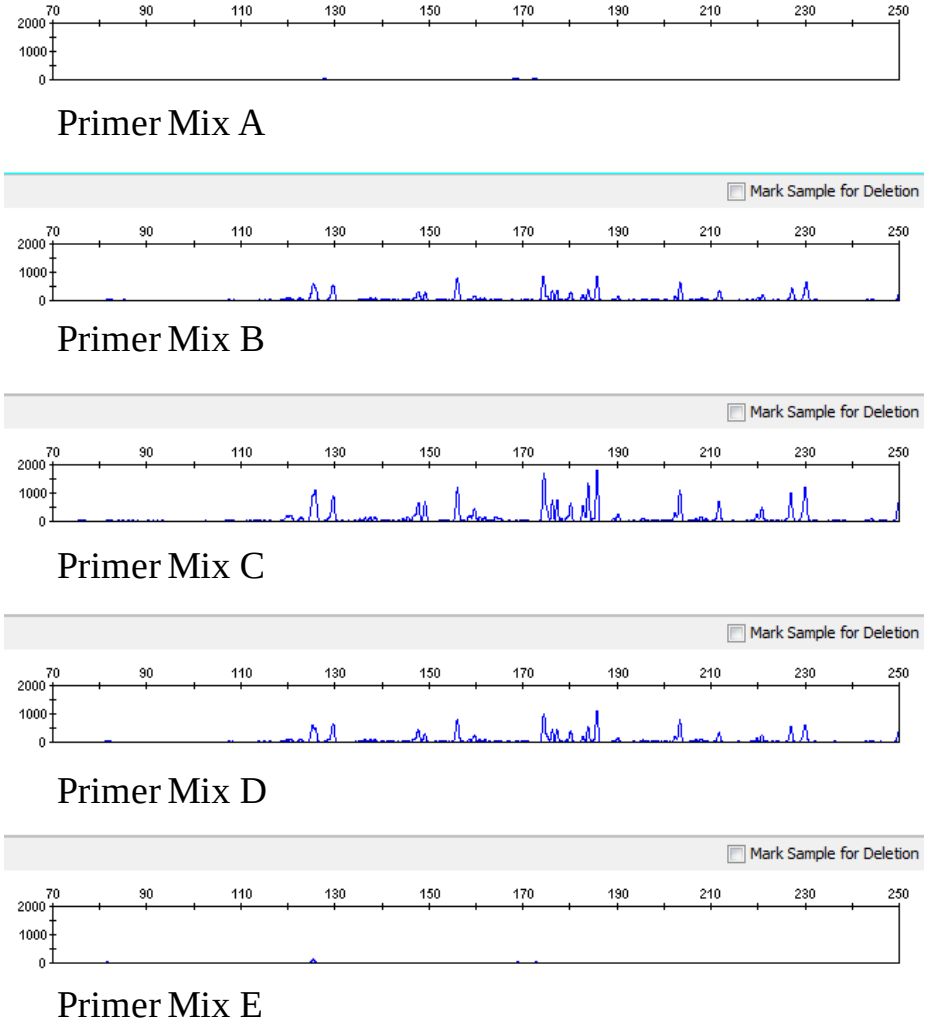



A series of multiplex primer mixes was created that omitted one primer set at a time in order to attempt to uncover which primer(s) might be responsible for the unspecific priming through process of elimination (Table 7.2). Amplification revealed that when the primer set for either GATA172D05 or DYS438 were removed from the multiplex primer mix (primer mix A and E respectively), the extraneous peaks disappeared (Figure 7.3). Because these extraneous peaks were seen only in the 6-FAM™ (blue) channel, the forward (labelled) GATA172D05 primer and the reverse (unlabelled) DYS438 primer were likely binding elsewhere in the genome to produce the additional amplicons. A BLAST search using these two primers revealed at least 10 additional amplicons between 70 and 250 bp.

**Table 7.2. Components of 5 primer mixes used for troubleshooting.** One primer set was omitted from each primer mix in order to observe affects on extraneous peaks occurring during multiplex amplification of an early version of the MIXplex. "X" indicates that the primer pair was included in the primer mix.

| | Primer mix | | | | |
|---|---|---|---|---|---|
| **Primer set** | **A** | **B** | **C** | **D** | **E** |
| GATA172D05 | | X | X | X | X |
| DYS393/DXYS267 | X | | X | X | X |
| SRY | X | X | | X | X |
| GATA31E08 | X | X | X | | X |
| DYS438 | X | X | X | X | |

**Figure 7.3. Results of amplification with primer mixes A-E.** Electropherograms showing 6-FAM™ (blue) channel results of amplification of male 1 with primer mixes A-E (Table 7.2). The data shown are between 70 and 250 base pairs in size.



As a result of the initial poor performance of the DXYS391 primer set and the artefacts generated as a result of the GATA172D05 forward primer, both markers were subsequently replaced with the X STR markers DXS6795 and DXS6789, which were known to be robust and polymorphic based upon previous X STR multiplex development work (see Chapter 3 & [55]). The organisation of the final MIXplex is show in Figure 7.4A. Further optimisation aimed at balancing the inter-locus peak heights was undertaken, and example electropherograms depicting the final multiplex are shown in Figure 7.4B. Though additional improvements, such as decreasing the rate of incomplete adenylation, would be necessary before routine use in a casework laboratory, the multiplex as shown was sufficient to test the principle of gonosomal markers to aid in mixture interpretation. Primer sequences and concentrations in the primer mix are detailed in Table 7.3.

**Figure 7.4. Multiplex organisation of the finalised MIXplex. A.** The electrophoretic position and known amplicon size range is shown. Markers with pink borders are X STRs, markers with blue borders are Y chromosomal markers, and the one with a black border is an XY marker. **B.** Example electropherograms of male, female, and 50:50 male-female mixed profiles generated using the final mixture multiplex.

**A.**

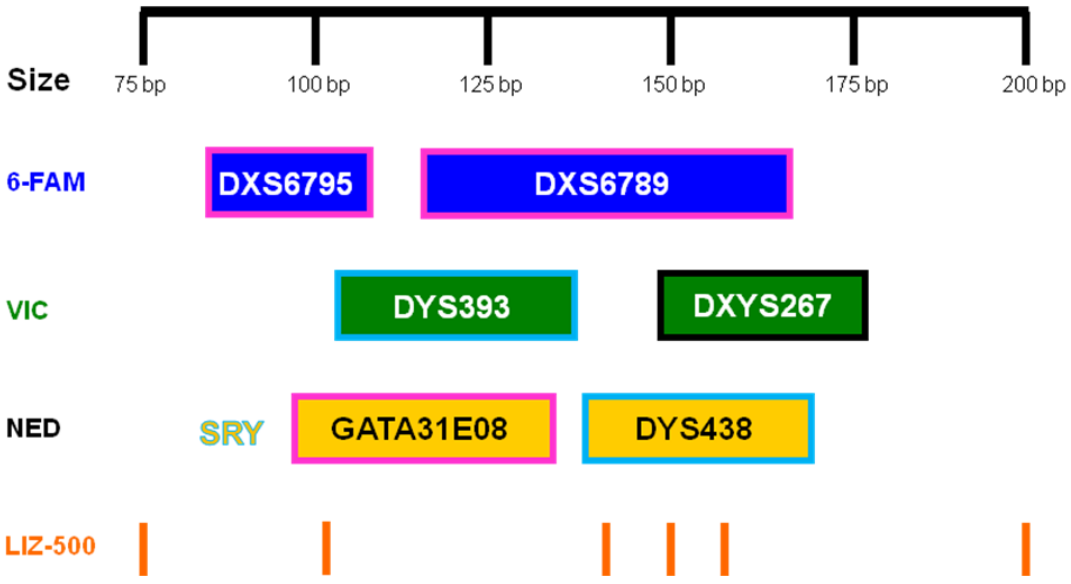



Figure continues on next page.

**B.**

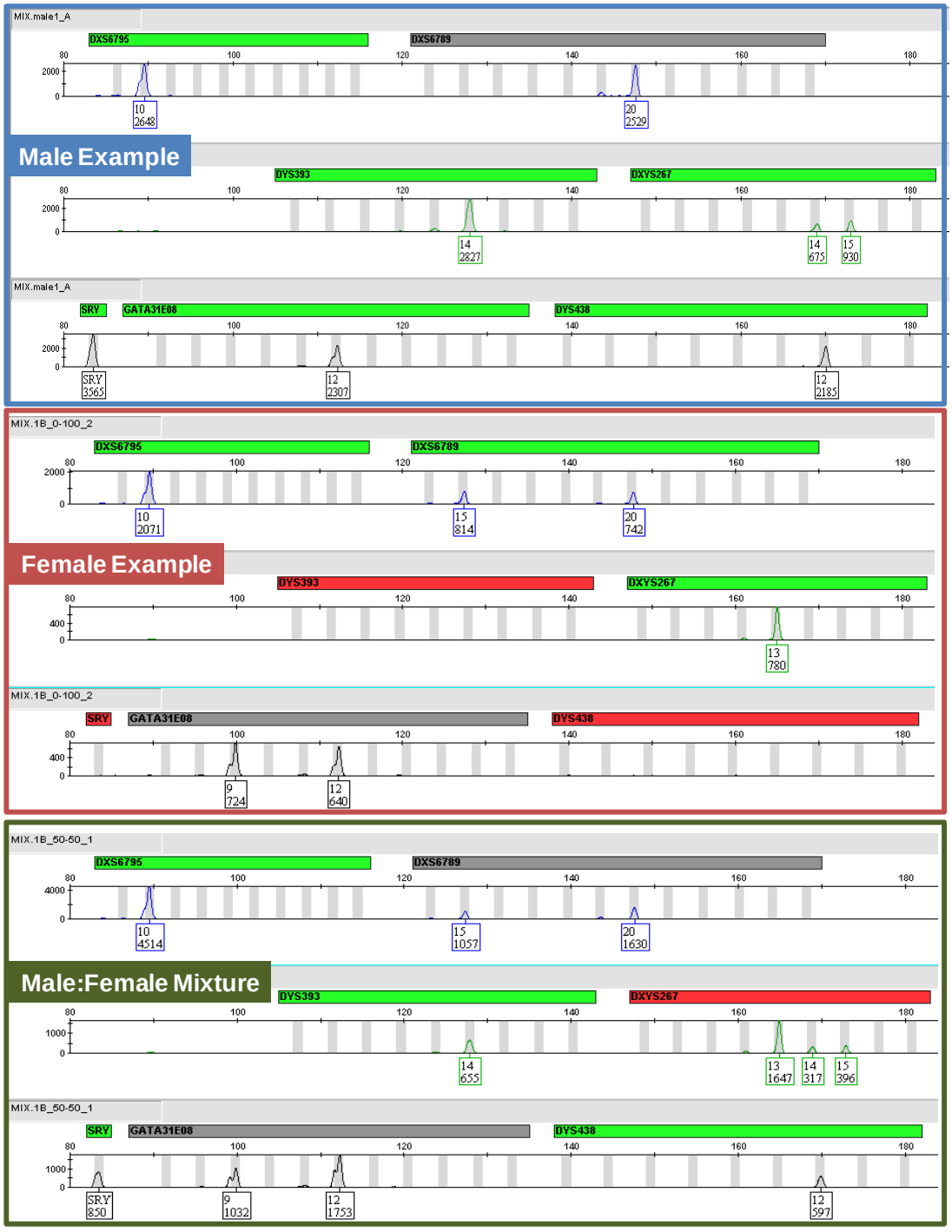

Male Example
MIX.male1_A
DXS6795
DXS6789
10 2648
20 2529
DYS393
DXYS267
14 2827
14 675
15 930
SRY
GATA31E08
DYS438
SRY 3565
12 2307
12 2185
Female Example
MIX.1B_0-100_2
10 2071
15 814
20 742
13 780
9 724
12 640
Male:Female Mixture
MIX.1B_50-50_1
10 4514
15 1057
20 1630
14 655
13 1647
14 317
15 396
SRY 850
9 1032
12 1753
12 597

**Table 7.3. Final primer sequences and concentrations used in MIXplex assay.** Bases in bold are tails added to the primer sequence to promote complete adenylation of the amplicon or to improve electrophoretic separation of adjacent amplicons. Amplicon size ranges include tails. Conc.: concentration; bp: base pairs. [a]Basic primer sequence obtained from publication with underlined base modified in this study to match GenBank sequence.

| Marker | | Primer Sequence (5'→3') | Ref. | Final Conc. (µM) | Amplicon Size (bp) |
|---|---|---|---|---|---|
| DXS6795 | F | 6FAM-TGACATGGCTTTCTTTACAATTAC | [55] | 0.8 | 81-114 |
| | R | **G**CCATGTTACATAAACAAGGAGTTATG | [55] | 0.8 | |
| DXS6789 | F | 6FAM-CCTCGTGATCATGTAAGTTGG | [54] | 1.2 | 124-168 |
| | R | **ATT**CAGAACCAATAGGAGATAGATGGT | [54] | 1.2 | |
| DYS393/ DXYS267 | F | VIC-GTGGTCTTCTACTTGTGTCAATAC | [320] | 1.6 | |
| DYS393 | R | **G**AACTCAAGTCCAAAAAATGAGG | [320] | 2.0 | 108-140 |
| DXYS267 | R | **G**CTAAATAAA<u>G</u>TCATATCAGCTGC | [320][a] | 0.1 | 148-180 |
| SRY | F | NED-AAAAATTGGCGATTAAGTCAAA | [55] | 0.8 | 86 |
| | R | **G**TTGACTACTTGCCCTGCTGA | [55] | 0.8 | |
| GATA31E08 | F | NED-CAGAGCTGGTGATGATAGATGA | [54] | 2.0 | 99-143 |
| | R | **ATT**CTCACTTTTATGTGTGTATGTATCTCC | [54] | 2.0 | |
| DYS438 | F | NED-TGGGGAATAGTTGAACGGTAA | [48] | 1.2 | 140-180 |
| | R | **GTTTCTT**GGAGGTTGTGGTGAGTCGAG | [249] | 1.2 | |

*7.2. Allele Sequencing*

In anticipation of a need to sequence certain alleles to confirm repeat number and structure, two new sequencing primer pairs were designed for markers DYS393 and DYS438. The primer sequences are shown in Table 7.4 along with previously designed sequencing primers (Chapter 3) for the three X markers.

**Table 7.4. Unlabelled primers used for sequencing purposes.**

| Marker | Forward Primer Sequence (5'→3') | Reverse Primer Sequence (5'→3') |
|---|---|---|
| DYS393 | TTGTAGTTATGTTTTATTTGTCATTCC | AAATGTTACAAAAAGAATGGCCTA |
| DYS438 | TGATGCAAGAAAGATTCACTGAT | AGGAGAATCGCTTGAACCTG |
| DXS6795 | TTCATGCTGTTGCTTTCCAG | CCATCCCCTAAACCTCTCAT |
| DXS6789 | TCAAGCTTGCAGACAGCCTA | TCGAAAAGATAGCCAATCACTG |
| GATA31E08 | AGCAAGGGGAGAAGGCTAGA | TCAGCTGACAGAGCACAGAGA |

*7.3. Sensitivity testing*

Full single-source MIXplex profiles were reliably obtained with as little as 200 pg of input DNA (Table 7.5). Complete loss of an allele despite triplicate amplification

was seen in two samples (one male and one female) at one marker (DXYS267 and GATA321E08 respectively) with 50 pg of input, and multiple alleles were lost from profiles amplified from only 25 pg of input. Sensitivity results did not vary by sex of the sample, as might be expected due to the chromosomal differences between the samples. Similarly, the entirely homozygous sample female 3 did not show increased sensitivity compared to the other two samples. Heterozygous peak height ratios remained above 60% for recovered alleles in all replicates (37) except two, both at DXYS267: one replicate of male 3 and one replicate of female 2 (both 52%). Marker DXYS267 appeared to be the least robust amplicon, as it was the first marker to exhibit loss of recovery in all three samples. In contrast, marker DXS6795 may be the most robust amplicon, with complete loss of amplification for only one allele (9) in one sample (male 3).

**Table 7.5. MIXplex sensitivity testing results.** Each single-source sample was amplified in triplicate at each of 6 different input quantities (54 amplifications total). Thresholds of 100 RFU for male or heterozygous alleles and 200 RFU for homozygous alleles were used to define the presence of a peak. Triplicate results were combined for the purposes of this table, with green boxes representing alleles that were above threshold in all three amplifications, yellow boxes representing alleles that were present in only 1 or 2 amplifications, and red boxes indicating that no alleles were above threshold. Allele calls are designated for each sample within the boxes.

| Quantity (pg) | Sample | DXS6795 | DXS6789 | DXS6789 | DYS393 | DXYS267 | DXYS267 | SRY | GATA31E08 | GATA31E08 | DYS438 |
|---|---|---|---|---|---|---|---|---|---|---|---|
| 1000 | Male 3 | 9 | 21 | | 13 | 12 | 13 | SRY | 14 | | 9 |
| | Female 2 | 10 | 15 | 20 | | 13 | | | 9 | 12 | |
| | Female 3 | 9 | 21 | | | 15 | | | 11 | | |
| 500 | Male 3 | 9 | 21 | | 13 | 12 | 13 | SRY | 14 | | 9 |
| | Female 2 | 10 | 15 | 20 | | 13 | | | 9 | 12 | |
| | Female 3 | 9 | 21 | | | 15 | | | 11 | | |
| 200 | Male 3 | 9 | 21 | | 13 | 12 | 13 | SRY | 14 | | 9 |
| | Female 2 | 10 | 15 | 20 | | 13 | | | 9 | 12 | |
| | Female 3 | 9 | 21 | | | 15 | | | 11 | | |
| 100 | Male 3 | 9 | 21 | | 13 | 12 | 13 | SRY | 14 | | 9 |
| | Female 2 | 10 | 15 | 20 | | 13 | | | 9 | 12 | |
| | Female 3 | 9 | 21 | | | 15 | | | 11 | | |
| 50 | Male 3 | 9 | 21 | | 13 | 12 | 13 | SRY | 14 | | 9 |
| | Female 2 | 10 | 15 | 20 | | 13 | | | 9 | 12 | |
| | Female 3 | 9 | 21 | | | 15 | | | 11 | | |
| 25 | Male 3 | 9 | 21 | | 13 | 12 | 13 | SRY | 14 | | 9 |
| | Female 2 | 10 | 15 | 20 | | 13 | | | 9 | 12 | |
| | Female 3 | 9 | 21 | | | 15 | | | 11 | | |

*7.4. Mixture testing*

*7.4.1. Artificial Mixtures*

The results of duplicate or triplicate amplification of six two-person mixtures (four female-male, one male-male, and one female-female) at varying mixture ratios is shown in Table 7.6. Complete profiles (defined as containing all expected alleles >100 RFU) were reliably obtained for mixtures where the minor component was 20% or greater in most cases. This value coincided well with the single-source sensitivity results for the MIXplex, which showed that reliable profiles were generated with just 200 pg input. Like the sensitivity results, the mixture testing also revealed that near-complete or complete profiles could also be obtained for most mixtures with the minor component at only 10%, or 100 pg input. Several notable

exceptions did exist, however. The minor component of mixture 1-A2 (Table 7.6A), 1-A3 (Table 7.6B), and 1-M2 (Table 7.6E) was completely undetected for the 10:90 (but not the 90:10) amplifications. In each case, the amplification reaction itself appeared to be suboptimal, as indicated by low peak heights and/or missing alleles for the major component (at 900 pg). It is likely that re-amplification may generate more robust profiles for both the major and minor components which more accurately reflect the sensitivity of the MIXplex.

When allele peak heights were divided into two categories, above or below 1000 RFU (shaded blue or not shaded, respectively, in Table 7.6 below), the pattern highlighted the minor component in more extreme mixture ratios (10:90, 20:80, 30:70, 70:30, 80:20, 90:10) and heterozygous markers in the relatively even mixture ratios (40:60, 50:50, 60:40), as expected. Little bias towards sex of the contributor or marker type was seen. Therefore, input DNA concentration was found to have the largest effect on allele recovery with the MIXplex, as would be a desired characteristic of an assay aimed at aiding mixture interpretation.

**Table 7.6 A-F. Mixture testing results.** Six two-person mixtures (four female-male, one male-male, and one female-female) were amplified in duplicate or triplicate both neat and at nine different mixture ratios. Expected profiles for the individual components as well as the mixtures are shown above each table, and RFU values for each peak greater than the 100 RFU reporting threshold were recorded within each box. Blue boxes highlight values >1000 RFU and uncoloured boxes contain values between 100 and 1000 RFU. Orange boxes highlight missing alleles (<100 RFU). Male 1 or M1: Quantifiler® Human DNA Standard; male 3 or M3: 2800M; female 2 or F2: AFDIL-1; female 3: K562.

## A. Mixture testing: female-male mixture 1-A2

| | DXS6795 | DXS6789 | DXS6789 | DYS393 | DXYS267 | DXYS267 | DXYS267 | SRY | GATA31E08 | GATA31E08 | DYS438 |
|---|---|---|---|---|---|---|---|---|---|---|---|
| **female 2** | **10** | **15** | **20** | | **13** | | | | **9** | **12** | |
| **male 1** | **10** | | **20** | **14** | | **14** | **15** | **SRY** | | **12** | **12** |
| **female 2:male 1** | **10** | **15** | **20** | **14** | **13** | **14** | **15** | **SRY** | **9** | **12** | **12** |
| 0:100 | 1320 | 320 | 334 | | 529 | | | | 372 | 387 | |
| | 2071 | 814 | 742 | | 780 | | | | 724 | 640 | |
| | 1880 | 513 | 586 | | 889 | | | | 597 | 581 | |
| 10:90 | 1015 | 230 | 257 | | 317 | | | | 279 | 279 | |
| | 384 | 115 | 132 | | 113 | | | | 117 | 113 | |
| | 186 | | | | | | | | | | |
| 20:80 | 8624 | 3145 | 4123 | 432 | 3798 | 190 | 212 | 554 | 2604 | 3085 | 383 |
| | 6835 | 2303 | 2604 | 511 | 2408 | 242 | 162 | 489 | 1504 | 1761 | 321 |
| | 3405 | 827 | 726 | 100 | 831 | | | 169 | 650 | 686 | 123 |
| 30:70 | 5067 | 1743 | 2066 | 557 | 2282 | 211 | 292 | 487 | 1708 | 1634 | 474 |
| | 5053 | 1320 | 1937 | 344 | 1652 | 169 | 175 | 358 | 1391 | 1602 | 331 |
| | 2443 | 792 | 916 | 203 | 842 | | | 206 | 691 | 649 | 158 |
| 40:60 | 5780 | 2113 | 2973 | 605 | 2257 | 251 | 312 | 604 | 1772 | 2175 | 634 |
| | 3758 | 1255 | 1586 | 259 | 1197 | 113 | 125 | 412 | 848 | 1254 | 296 |
| | 5271 | 1774 | 2081 | 509 | 1634 | 255 | 178 | 677 | 1600 | 1837 | 371 |
| 50:50 | 4514 | 1057 | 1630 | 655 | 1647 | 317 | 396 | 850 | 1032 | 1753 | 597 |
| | 4711 | 1354 | 1651 | 445 | 1421 | 210 | 322 | 606 | 839 | 1570 | 594 |
| | 2453 | 626 | 1036 | 394 | 587 | 180 | 166 | 412 | 577 | 673 | 390 |
| 60:40 | 3673 | 743 | 1563 | 647 | 871 | 283 | 399 | 888 | 867 | 1368 | 712 |
| | 3585 | 823 | 1747 | 916 | 1049 | 406 | 320 | 754 | 851 | 1335 | 618 |
| | 4971 | 1285 | 1818 | 928 | 1279 | 478 | 401 | 700 | 524 | 2002 | 838 |
| 70:30 | 2613 | 537 | 1199 | 702 | 650 | 324 | 387 | 846 | 489 | 1158 | 687 |
| | 2571 | 677 | 1249 | 758 | 575 | 334 | 378 | 746 | 533 | 1117 | 655 |
| | 2601 | 538 | 1119 | 615 | 573 | 278 | 267 | 625 | 408 | 845 | 544 |
| 80:20 | 2350 | 504 | 1458 | 1157 | 475 | 478 | 420 | 1288 | 456 | 1058 | 646 |
| | 1984 | 442 | 1213 | 688 | 398 | 297 | 313 | 1045 | 332 | 915 | 574 |
| | 3950 | 452 | 2128 | 1125 | 754 | 578 | 677 | 1454 | 549 | 1344 | 1074 |
| 90:10 | 1724 | | 922 | 829 | 185 | 331 | 480 | 874 | 135 | 1005 | 676 |
| | 2064 | 235 | 1143 | 1055 | 211 | 466 | 450 | 1021 | 144 | 834 | 594 |
| | 1836 | 120 | 985 | 686 | 206 | 331 | 417 | 1061 | 250 | 882 | 809 |
| 100:0 | 2466 | | 1477 | 1823 | | 768 | 867 | 2089 | | 1126 | 1859 |
| | 1874 | | 1392 | 1064 | | 478 | 478 | 1158 | | 902 | 1100 |
| | 2187 | | 1614 | 1263 | | 542 | 631 | 1571 | | 975 | 1399 |

## B. Mixture testing: female-male mixture 1-A3

| | DXS6795 | DXS6795 | DXS6789 | DXS6789 | DYS393 | DXYS267 | DXYS267 | SRY | GATA31E08 | GATA31E08 | DYS438 |
|---|---|---|---|---|---|---|---|---|---|---|---|
| **female 3** | **9** | | | **21** | | | **15** | | | **13** | |
| **male 1** | | **10** | **20** | | **14** | **14** | **15** | **SRY** | **12** | | **12** |
| **female 3:male 1** | **9** | **10** | **20** | **21** | **14** | **14** | **15** | **SRY** | **12** | **13** | **12** |
| 0:100 | 908 | | | 536 | | | 322 | | | 485 | |
| | 2647 | | | 1697 | | | 957 | | | 1608 | |
| | 1078 | | | 652 | | | 359 | | | 576 | |
| 10:90 | 143 | | | | | | | | | | |
| | 616 | | | 297 | | | 138 | | | 307 | |
| | 816 | | | 426 | | | 196 | | | 448 | |
| 20:80 | 5500 | 561 | 678 | 3368 | 282 | 341 | 2410 | 362 | 412 | 2686 | 425 |
| | 4173 | 231 | 476 | 2601 | 213 | 224 | 1497 | 254 | 249 | 1573 | 231 |
| | 3838 | 324 | 498 | 2026 | 165 | 185 | 1341 | 128 | 277 | 1629 | 235 |
| 30:70 | 5334 | 902 | 922 | 3123 | 596 | 493 | 2422 | 394 | 688 | 2680 | 725 |
| | 5573 | 874 | 940 | 3497 | 417 | 333 | 1820 | 797 | 801 | 2999 | 883 |
| | 3614 | 508 | 616 | 1690 | 318 | 200 | 915 | 344 | 314 | 1352 | 266 |
| 40:60 | 3782 | 831 | 804 | 2502 | 588 | 335 | 1467 | 797 | 431 | 1660 | 515 |
| | 4200 | 1050 | 1136 | 2326 | 609 | 351 | 1273 | 575 | 610 | 1652 | 554 |
| | 4006 | 1043 | 825 | 2044 | 563 | 295 | 1440 | 738 | 569 | 1935 | 482 |
| 50:50 | 2380 | 666 | 814 | 1132 | 442 | 273 | 1034 | 625 | 605 | 1159 | 615 |
| | 3262 | 866 | 874 | 1495 | 703 | 390 | 1191 | 840 | 770 | 1352 | 594 |
| | 2274 | 737 | 581 | 1355 | 382 | 210 | 828 | 517 | 467 | 962 | 410 |
| 60:40 | 4390 | 2548 | 1427 | 2162 | 1255 | 730 | 1816 | 1312 | 1391 | 1656 | 1271 |
| | 2311 | 1148 | 1303 | 1963 | 1096 | 547 | 1317 | 738 | 946 | 1443 | 850 |
| | 2661 | 1252 | 1105 | 1492 | 848 | 437 | 926 | 869 | 632 | 1332 | 806 |
| 70:30 | 1672 | 1051 | 907 | 988 | 528 | 282 | 736 | 733 | 584 | 548 | 494 |
| | 1568 | 1166 | 807 | 669 | 596 | 278 | 832 | 905 | 546 | 599 | 772 |
| | 1585 | 1424 | 1031 | 1107 | 731 | 318 | 628 | 876 | 776 | 788 | 609 |
| 80:20 | 1534 | 1616 | 1540 | 699 | 1284 | 615 | 1191 | 1389 | 939 | 951 | 1257 |
| | 1088 | 1571 | 904 | 732 | 1077 | 547 | 766 | 999 | 579 | 394 | 902 |
| | 1147 | 1510 | 930 | 547 | 1136 | 491 | 749 | 969 | 952 | 485 | 786 |
| 90:10 | 515 | 1228 | 1083 | 411 | 720 | 359 | 817 | 1073 | 714 | 249 | 925 |
| | 825 | 1719 | 951 | 472 | 1085 | 520 | 788 | 1051 | 990 | 231 | 949 |
| | 771 | 1427 | 1148 | 341 | 768 | 374 | 660 | 1631 | 1029 | 318 | 1146 |
| 100:0 | | 2247 | 1586 | | 1393 | 643 | 597 | 1614 | 1104 | | 1199 |
| | | 2008 | 1259 | | 1162 | 516 | 570 | 1505 | 980 | | 1130 |
| | | 1853 | 1518 | | 1371 | 582 | 576 | 1112 | 1154 | | 906 |

## C. Mixture testing: female-male mixture 1-A8

| | DXS6795 | DXS6795 | DXS6789 | DXS6789 | DXS6789 | DYS393 | DXYS267 | DXYS267 | SRY | GATA31E08 | GATA31E08 | GATA31E08 | DYS438 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| **F2** | | **10** | **15** | **20** | | | | **13** | | **9** | **12** | | |
| **M3** | **9** | | | | **21** | **13** | **12** | **13** | **SRY** | | | **14** | **9** |
| **F2:M3** | **9** | **10** | **15** | **20** | **21** | **13** | **12** | **13** | **SRY** | **9** | **12** | **14** | **9** |
| 0:100 | | 6612 | 2625 | 2429 | | | | 3025 | | 2051 | 1764 | | |
| | | 6301 | 2411 | 2206 | | | | 2642 | | 1472 | 1499 | | |
| | | 8140 | 2663 | 2398 | | | | 4191 | | 2387 | 2044 | | |
| 10:90 | 291 | 3787 | 1227 | 1463 | 187 | 216 | 285 | 1971 | 285 | 1229 | 1232 | 207 | 232 |
| | 400 | 4785 | 1752 | 1476 | 306 | 154 | 224 | 1931 | 112 | 1098 | 1211 | 134 | 103 |
| | 557 | 4563 | 2028 | 1730 | 172 | 279 | 314 | 2397 | 239 | 1352 | 1540 | 187 | 169 |
| 20:80 | 836 | 5079 | 2013 | 1583 | 697 | 339 | 369 | 2178 | 633 | 1101 | 1251 | 362 | 382 |
| | 703 | 4388 | 1569 | 1430 | 437 | 245 | 470 | 2060 | 426 | 1191 | 972 | 262 | 313 |
| | 1077 | 5941 | 1966 | 2093 | 464 | 527 | 414 | 2934 | 550 | 1392 | 1490 | 367 | 385 |
| 30:70 | 1091 | 3607 | 1201 | 1199 | 512 | 569 | 548 | 1978 | 804 | 1334 | 1991 | 584 | 552 |
| | 623 | 3499 | 1383 | 1472 | 553 | 444 | 442 | 1631 | 551 | 920 | 901 | 314 | 569 |
| | 775 | 3648 | 1264 | 1096 | 599 | 353 | 582 | 2231 | 691 | 1117 | 878 | 390 | 505 |
| 40:60 | 1833 | 4535 | 1987 | 2003 | 1140 | 937 | 878 | 2262 | 848 | 1250 | 1347 | 1003 | 735 |
| | 1049 | 3628 | 1312 | 1278 | 884 | 531 | 543 | 1460 | 693 | 809 | 1055 | 613 | 625 |
| | 1791 | 5108 | 1898 | 1997 | 1150 | 1031 | 896 | 3053 | 1371 | 1486 | 1381 | 716 | 737 |
| 50:50 | 1246 | 2746 | 839 | 1078 | 753 | 1041 | 709 | 1837 | 962 | 928 | 866 | 969 | 854 |
| | 1593 | 2594 | 1136 | 975 | 1018 | 773 | 545 | 1327 | 846 | 601 | 695 | 774 | 677 |
| | 1753 | 2917 | 1126 | 1113 | 895 | 1091 | 686 | 2254 | 1234 | 821 | 800 | 800 | 921 |
| 60:40 | 2452 | 2885 | 777 | 1000 | 1301 | 1277 | 822 | 1754 | 1071 | 516 | 639 | 1257 | 1075 |
| | 1898 | 2349 | 751 | 1036 | 907 | 1206 | 738 | 1488 | 1262 | 586 | 568 | 876 | 758 |
| | 2560 | 3070 | 1070 | 886 | 1585 | 1088 | 1295 | 1979 | 1465 | 937 | 602 | 1256 | 968 |
| 70:30 | 1686 | 1161 | 481 | 504 | 987 | 930 | 858 | 1117 | 1210 | 460 | 522 | 922 | 921 |
| | 1867 | 1729 | 608 | 727 | 1098 | 1001 | 648 | 1103 | 1163 | 379 | 385 | 887 | 779 |
| | 2310 | 1787 | 534 | 757 | 1425 | 1100 | 932 | 1468 | 1174 | 480 | 410 | 1236 | 922 |
| 80:20 | 3205 | 1733 | 552 | 608 | 1913 | 1577 | 1060 | 1413 | 1660 | 379 | 499 | 1471 | 1520 |
| | 2430 | 1651 | 354 | 577 | 1341 | 1142 | 950 | 994 | 1479 | 348 | 423 | 1133 | 1091 |
| | 2838 | 1612 | 550 | 752 | 2026 | 1452 | 1217 | 1482 | 1740 | 376 | 382 | 1427 | 1297 |
| 90:10 | 2935 | 460 | 188 | 452 | 1855 | 1538 | 1166 | 1066 | 2182 | | 244 | 1697 | 1712 |
| | 2551 | 748 | 109 | 379 | 1568 | 1168 | 905 | 810 | 1454 | 157 | 109 | 1177 | 1211 |
| | 2466 | 589 | 248 | 392 | 1629 | 1250 | 1180 | 871 | 1527 | | 244 | 1289 | 1423 |
| 100:0 | 3840 | | | | 2769 | 2291 | 1384 | 1035 | 2506 | | | 1863 | 1810 |
| | 3425 | | | | 2400 | 1486 | 1261 | 763 | 2021 | | | 1332 | 1573 |
| | 3758 | | | | 2578 | 2003 | 1458 | 939 | 2242 | | | 1679 | 1814 |

## D. Mixture testing: female-male mixture 1-A9

| | DXS6795 | DXS6789 | DYS393 | DXYS267 | DXYS267 | DXYS267 | SRY | GATA31E08 | GATA31E08 | DYS438 |
|---|---|---|---|---|---|---|---|---|---|---|
| **female 3** | **9** | **21** | | | | **15** | | **13** | | |
| **male 3** | **9** | **21** | **13** | **12** | **13** | | **SRY** | | **14** | **9** |
| **female 3:male 3** | **9** | **21** | **13** | **12** | **13** | **15** | **SRY** | **13** | **14** | **9** |
| 0:100 | 6397 | 3941 | | | | 2715 | | 2907 | | |
| | 7003 | 3415 | | | | 2043 | | 2689 | | |
| | 6412 | 4133 | | | | 2059 | | 3208 | | |
| 10:90 | 4315 | 2820 | 191 | 236 | 123 | 1618 | 207 | 1764 | 208 | 221 |
| | 5253 | 2948 | 266 | 191 | 127 | 1569 | 355 | 1953 | 233 | 272 |
| | 5934 | 2686 | 189 | 158 | 114 | 1533 | 312 | 2042 | 333 | 199 |
| 20:80 | 5288 | 3441 | 356 | 355 | 182 | 1655 | 519 | 2115 | 295 | 538 |
| | 5191 | 3298 | 433 | 386 | 231 | 1421 | 559 | 1932 | 342 | 288 |
| | 5157 | 3130 | 424 | 213 | 229 | 1488 | 306 | 1637 | 284 | 338 |
| 30:70 | 5070 | 2989 | 432 | 433 | 266 | 1519 | 778 | 1258 | 526 | 599 |
| | 5279 | 3142 | 566 | 480 | 345 | 1393 | 757 | 1521 | 591 | 513 |
| | 4810 | 3205 | 528 | 449 | 261 | 1119 | 852 | 1522 | 473 | 561 |
| 40:60 | 6371 | 4254 | 867 | 732 | 454 | 1308 | 1375 | 1660 | 841 | 778 |
| | 5036 | 3258 | 749 | 578 | 445 | 1226 | 1210 | 1757 | 801 | 735 |
| | 5528 | 2717 | 701 | 640 | 353 | 1333 | 1000 | 1823 | 882 | 779 |
| 50:50 | 4461 | 2630 | 922 | 782 | 470 | 840 | 1013 | 1301 | 834 | 961 |
| | 5351 | 2903 | 1150 | 799 | 605 | 1286 | 1178 | 1631 | 899 | 1101 |
| | 5031 | 2938 | 967 | 864 | 465 | 811 | 1026 | 1404 | 995 | 1197 |
| 60:40 | 5189 | 3242 | 1431 | 1156 | 879 | 1030 | 1824 | 1444 | 1296 | 1114 |
| | 4831 | 3101 | 1214 | 775 | 666 | 785 | 1614 | 1155 | 1310 | 1065 |
| | 4198 | 2993 | 1354 | 819 | 669 | 594 | 1239 | 1040 | 1269 | 1075 |
| 70:30 | 3954 | 2520 | 1043 | 885 | 623 | 538 | 1545 | 855 | 1228 | 1328 |
| | 4780 | 2732 | 1330 | 1078 | 812 | 525 | 1753 | 947 | 1228 | 1466 |
| | 4048 | 2331 | 1318 | 889 | 647 | 412 | 1986 | 984 | 1232 | 1428 |
| 80:20 | 4318 | 2708 | 1520 | 1192 | 807 | 465 | 2242 | 592 | 1517 | 1432 |
| | 4314 | 2706 | 1420 | 1129 | 788 | 368 | 2293 | 574 | 1198 | 1425 |
| | 2949 | 1961 | 949 | 993 | 694 | 340 | 1495 | 393 | 1123 | 992 |
| 90:10 | 3911 | 2376 | 1447 | 1359 | 726 | 221 | 1865 | 370 | 1641 | 1768 |
| | 3986 | 2316 | 1808 | 1507 | 981 | 348 | 2548 | 368 | 1700 | 2018 |
| | 4189 | 2295 | 1868 | 1106 | 948 | 227 | 1971 | 417 | 1623 | 1823 |
| 100:0 | 4168 | 2594 | 1925 | 1690 | 1022 | | 2953 | | 2208 | 2255 |
| | 3874 | 2159 | 1866 | 1235 | 988 | | 2614 | | 1953 | 1857 |
| | 3840 | 2225 | 2003 | 1299 | 900 | | 2387 | | 1701 | 1716 |

## E. Mixture testing: male-male mixture 1-M2

| | DXS6795 | DXS6795 | DXS6789 | DXS6789 | DYS393 | DYS393 | DXYS267 | DXYS267 | DXYS267 | DXYS267 | SRY | GATA31E08 | GATA31E08 | DYS438 | DYS438 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| **M1** | | **10** | **20** | | | **14** | | | **14** | **15** | | **12** | | | **12** |
| **M3** | **9** | | | **21** | **13** | | **12** | **13** | | | **SRY** | | **14** | **9** | |
| **M3:M1** | **9** | **10** | **20** | **21** | **13** | **14** | **12** | **13** | **14** | **15** | **SRY** | **12** | **14** | **9** | **12** |
| 0:100 | 694 | | | 450 | 424 | | 182 | 141 | | | 519 | | 418 | 422 | |
| | 581 | | | 371 | 418 | | 236 | 161 | | | 582 | | 378 | 384 | |
| 10:90 | 250 | | | 156 | 139 | | | | | | 209 | | 143 | 173 | |
| | 420 | | | 233 | 246 | | 130 | | | | 368 | | 262 | 258 | |
| 20:80 | 4651 | 937 | 866 | 3396 | 3058 | 367 | 2151 | 1200 | 168 | 102 | 3733 | 317 | 2659 | 2919 | 508 |
| | 3986 | 479 | 731 | 2469 | 2387 | 347 | 1418 | 875 | 147 | 164 | 3056 | 432 | 2390 | 2381 | 261 |
| 30:70 | 3757 | 812 | 1357 | 2645 | 2997 | 535 | 1542 | 1187 | 225 | 238 | 3355 | 532 | 2352 | 2895 | 827 |
| | 3547 | 882 | 1060 | 2758 | 2393 | 839 | 1527 | 869 | 362 | 339 | 2988 | 436 | 2047 | 2539 | 599 |
| 40:60 | 2416 | 1217 | 743 | 1683 | 1492 | 451 | 997 | 572 | 184 | 237 | 2429 | 400 | 1677 | 1470 | 650 |
| | 2157 | 660 | 954 | 1290 | 1744 | 518 | 834 | 669 | 209 | 232 | 1863 | 380 | 1401 | 1650 | 519 |
| 50:50 | 2179 | 1031 | 1275 | 1720 | 1266 | 650 | 699 | 524 | 324 | 483 | 1852 | 629 | 1138 | 989 | 765 |
| | 2347 | 992 | 863 | 1365 | 1601 | 662 | 784 | 607 | 325 | 352 | 2153 | 546 | 963 | 916 | 865 |
| 60:40 | 1471 | 1050 | 1222 | 1296 | 1124 | 1074 | 626 | 431 | 410 | 382 | 1812 | 642 | 990 | 1133 | 7965 |
| | 1597 | 1263 | 878 | 1013 | 1249 | 736 | 532 | 469 | 326 | 380 | 1821 | 597 | 948 | 961 | 852 |
| 70:30 | 1517 | 1594 | 1890 | 1192 | 1375 | 2247 | 333 | 264 | 425 | 337 | 2113 | 1178 | 851 | 769 | 1078 |
| | 844 | 878 | 730 | 578 | 448 | 534 | 302 | 179 | 229 | 255 | 1282 | 638 | 703 | 440 | 519 |
| 80:20 | 556 | 1983 | 1156 | 236 | 265 | 1011 | 121 | | 413 | 479 | 1364 | 1038 | 117 | 204 | 816 |
| | 735 | 846 | 846 | 367 | 487 | 837 | 217 | 157 | 329 | 237 | 1480 | 655 | 206 | 374 | 645 |
| 90:10 | | 1715 | 1335 | | | 1111 | | | 458 | 627 | 1447 | 877 | | | 1263 |
| | 307 | 1626 | 999 | 210 | 271 | 821 | | | 366 | 405 | 1427 | 982 | 160 | 194 | 898 |
| 100:0 | | 1998 | 887 | | | 1087 | | | 449 | 412 | 1008 | 1096 | | | 1014 |

**F. Mixture testing: female-female mixture 1-F3**

| | DXS6795 | DXS6795 | DXS6789 | DXS6789 | DXS6789 | DXYS267 | DXYS267 | GATA31E08 | GATA31E08 | GATA31E08 |
|---|---|---|---|---|---|---|---|---|---|---|
| **female 2** | | **10** | **15** | **20** | | **13** | | **9** | **12** | |
| **female 3** | **9** | | | | **21** | | **15** | | | **13** |
| **female 3:female 2** | **9** | **10** | **15** | **20** | **21** | **13** | **15** | **9** | **12** | **13** |
| 0:100 | 7011 | | | | 4161 | | 2596 | | | 3497 |
| | 5026 | | | | 2666 | | 1723 | | | 2737 |
| 10:90 | 4544 | 479 | 253 | 452 | 2470 | 364 | 1291 | 263 | 312 | 1888 |
| | 4055 | 710 | 181 | 569 | 2503 | 305 | 1568 | 195 | 376 | 1993 |
| 20:80 | 4742 | 1613 | 541 | 723 | 2319 | 595 | 1359 | 439 | 490 | 1793 |
| | 4335 | 1131 | 416 | 512 | 2244 | 577 | 1459 | 453 | 342 | 2138 |
| 30:70 | 3799 | 1849 | 623 | 862 | 1966 | 697 | 1076 | 521 | 505 | 1587 |
| | 3652 | 1944 | 815 | 702 | 2133 | 996 | 1364 | 482 | 590 | 1666 |
| 40:60 | 4465 | 2785 | 1209 | 1472 | 2264 | 1486 | 1398 | 835 | 1205 | 1320 |
| | 3005 | 2523 | 1186 | 1272 | 1788 | 1260 | 1041 | 976 | 911 | 1548 |
| 50:50 | 2990 | 3022 | 973 | 1439 | 1531 | 1005 | 956 | 644 | 822 | 1208 |
| | 3000 | 3283 | 1095 | 1454 | 1704 | 1253 | 846 | 813 | 967 | 1504 |
| 60:40 | 2402 | 4283 | 1457 | 1636 | 1502 | 1315 | 827 | 982 | 1122 | 1221 |
| | 1875 | 3417 | 1156 | 1083 | 1039 | 1228 | 672 | 780 | 889 | 1073 |
| 70:30 | 1794 | 4365 | 1533 | 1212 | 922 | 1678 | 573 | 1212 | 1381 | 764 |
| | 1599 | 3674 | 1171 | 1448 | 781 | 1560 | 761 | 989 | 1096 | 703 |
| 80:20 | 1462 | 5453 | 2162 | 2291 | 944 | 2215 | 382 | 1574 | 1521 | 643 |
| | 1117 | 4549 | 1449 | 1426 | 491 | 2074 | 268 | 1428 | 1204 | 384 |
| 90:10 | 561 | 4712 | 1826 | 1730 | 212 | 2079 | 237 | 1175 | 1329 | 191 |
| | 638 | 4845 | 1734 | 1343 | 257 | 1737 | 225 | 1304 | 1189 | 229 |
| 100:0 | | 7605 | 2844 | 2534 | | 3198 | | 1585 | 1828 | |
| | | 6197 | 2705 | 2500 | | 2695 | | 2206 | 2330 | |

The same mixtures and mixture ratios were amplified with the autosomal STR kit Identifiler® (Applied Biosystems) and analyzed for the ability to detect the presence of a mixture. Based upon the criteria of observing at least two markers with >2 alleles above 100 RFU, all mixtures with a minor component 20% or greater as well as most mixtures with a minor component of 10% or greater, were correctly identified as mixtures, as expected. Assignment of the sex of the contributors, however, was more complicated due to normal variation in peak height seen with the single sex-typing marker, amelogenin.

*7.4.2. Theoretical mixtures*

Sixty-three theoretical MIXplex profiles were analyzed to determine the minimum number and sex of contributors without knowledge of the profile source(s). During this process, a general method and sequence of analysis was found to be optimal and is summarised in Table 7.7. To begin, the presence or absence of the SRY peak was

noted. Its presence indicated that at least one male contributed to the profile. Its absence, however, did not necessarily preclude a male contribution. In order to account for the possibility of an SRY null allele, the presence or absence of alleles at the two Y chromosomal markers DYS393 and DYS438 was noted. If the SRY peak was missing, but at least one peak is present at a Y marker, then it can was assumed there was a male component to the mixture. Thus far, an SRY null allele has never been observed during the typing of over 3000 samples described in Chapters 4 & 5, or in the literature at the time of writing.

**Table 7.7. Interpretation method used in this study to determine the minimum number and sex of contributors for MIXplex profiles in the absence of peak height information.** *Must round up to nearest whole number. **Must see all DYS393 alleles captured in DXYS267 profile.

| Order | Marker type | Inference(s) made |
|---|---|---|
| 1 | SRY | Determine if there is a male component present |
| 2 | DYS | 1. Confirm presence or absence of male component<br>2. Maximum number of alleles at one marker = minimum number of males |
| 3 | DXS | (Maximum number of alleles at one marker – minimum number of males)/2 = minimum number of females* |
| 4 | DXYS | 1. Confirm conclusions regarding the minimum number of total contributors<br>2. Confirm minimum number of male contributors** |

Once the presence of a male component was established, the Y STR markers were used to determine the minimum number of male contributors to the mixture. The largest number of detectable alleles present at a Y STR marker in the multiplex represented the minimum number of males present in the mixture. Additional males may have contributed to the profile but were not represented due to allele sharing.

After establishing the minimum number of male contributors or the absence of a male component, the X STR markers were examined to determine the possible presence of a female component to the mixture. The previously-determined minimum number of males in the mixture was subtracted from the number of alleles at the X STR marker with the maximum number of alleles and divided in half. After rounding up to the nearest whole number, the result was the minimum number of

female contributors to the profile. Again, additional females may have been present but were masked by allele sharing.

Lastly, the XY homologous marker was used to confirm inferences made in the previous three steps concerning sex and minimum number of contributors. The number of alleles could be used to confirm the minimum number of contributors in the same way as with autosomal profiles; there had to be at least one allele present for each suspected contributor (male or female). Additionally, since the DXYS primer pair was simply amplifying a larger flanking region surrounding DYS393, all of the DYS393 alleles must also have been represented in the DXYS267 profile.

In contrast, the interpretation logic used for the theoretical Identifiler® mixtures in the absence of peak height information was relatively simplistic (Table 7.8). Since there were only two marker types present in this assay, inferences were limited to the presence or absence of a minimum of one male contributor, and a determination of the minimum number of contributors. No additional conclusions regarding the sex or the number of each sex of contributors were possible.

**Table 7.8. Interpretation method used in this study to determine the minimum number of contributors for Identifiler® profiles in the absence of peak height information.** *Must round up to nearest whole number.

| Order | Marker type | Inference(s) made |
|---|---|---|
| 1 | Amelogenin | Determine if there is a male component present |
| 2 | Remaining autosomal markers | Maximum number of alleles at one marker ÷ 2 = minimum number of contributors* |

Once established, these analysis methods were used on all 63 theoretical mixtures to define the sex and minimum number of suspected contributors. These results were subsequently compared to the true contributors, and the difference was noted (Table 7.9). For ease of comparison, Identifiler® differences were designated as female when both male(s) and female(s) contributors were not inferred, and a loss of sex information despite the correct number of contributors was captured as a difference of "1 sex."

**Table 7.9. Blind analysis of the MIXplex profiles of 63 theoretical mixtures.** The "actual combination" indicates the true sex and number of contributors used to create the theoretical profile. The "minimum combination" describes the number and sex of contributors determined according the analysis methods above (Tables 7.7 & 7.8) for both the MIXplex and Identifiler® kit. The difference between the minimum and actual combination is shown in the last column. Complete loss of information is highlighted in red text, while loss of either contributor number information or sex determination losses are noted in blue and green text, respectively. "Sex" in the "difference" column designates a loss of contributor sex determination though the number of contributors was captured correctly.

| Profile Identifier | Actual Combination | MIXplex Minimum Combination | MIXplex Difference | Identifiler® Minimum Combination | Identifiler® Difference |
|---|---|---|---|---|---|
| 101 | 1 male/2 females | 1 male/2 females | | 2 total, 1 male | 1 female + 1 sex |
| 102 | 3 males/1 female | 3 males/1 female | | 3 total, 1 male | 1 female + 2 sex |
| 103 | 1 male/2 females | 1 male/2 females | | 3 total, 1 male | 1 female + 2 sex |
| 104 | 2 males/2 females | 2 males/1 female | 1 female | 4 total, 1 male | 3 sex |
| 105 | 3 males/2 females | 3 males/1 female | 1 female | 4 total, 1 male | 1 female + 3 sex |
| 106 | 1 male | 1 male | | 1 male | |
| 107 | 2 females | 2 females | | 2 females | |
| 108 | 2 males | 2 males | | 2 total, 1 male | 1 male |
| 109 | 1 male/1 female | 1 male/1 female | | 2 total, 1 male | 1 female |
| 110 | 2 males/1 female | 2 males/1 female | | 2 total, 1 male | 1 female + 1 sex |
| 111 | 1 male/2 females | 1 male/2 females | | 2 total, 1 male | 1 female + 1 sex |
| 112 | 1 male/2 females | 1 male/1 female | 1 female | 2 total, 1 male | 1 female + 1 sex |
| 113 | 1 female | 1 female | | 1 female | |
| 114 | 1 male/3 females | 1 male/2 females | 1 female | 3 total, 1 male | 1 female + 2 sex |
| 115 | 3 males/3 females | 3 males/1 female | 2 females | 4 total, 1 male | 2 females + 3 sex |
| 116 | 2 males/2 females | 2 males/2 females | | 3 total, 1 male | 1 female + 2 sex |
| 117 | 1 male/2 females | 1 male/1 female | 1 female | 3 total, 1 male | 2 sex |
| 118 | 1 male/1 female | 1 male/1 female | | 2 total, 1 male | 1 sex |
| 119 | 1 male/1 female | 1 male/1 female | | 2 total, 1 male | 1 sex |
| 120 | 2 males/1 female | 2 males | 1 female | 3 total, 1 male | 2 sex |
| 121 | 2 males | 2 males | | 2 total, 1 male | 1 sex |
| 122 | 2 males/1 female | 2 males/1 female | | 3 total, 1 male | 2 sex |
| 123 | 1 female | 1 female | | 1 female | |
| 124 | 1 male/2 females | 1 male/1 female | 1 female | 3 total, 1 male | 2 sex |
| 125 | 3 males | 3 males | | 3 total, 1 male | 2 sex |
| 126 | 1 male/3 females | 1 male/2 females | 1 female | 3 total, 1 male | 1 female + 2 sex |
| 127 | 2 males/2 females | 2 males/1 female | 1 female | 3 total, 1 male | 1 female + 2 sex |
| 128 | 2 males/2 females | 2 males/1 female | 1 female | 3 total, 1 male | 1 female + 2 sex |
| 129 | 2 males/2 females | 2 males/2 females | | 3 total, 1 male | 1 female + 2 sex |
| 130 | 3 males/2 females | 3 males/1 female | 1 female | 4 total, 1 male | 1 female + 3 sex |
| 131 | 1 male/2 females | 1 male/2 females | | 2 total, 1 male | 1 female + 1 sex |
| 132 | 1 male/2 females | 1 male/2 females | | 3 total, 1 male | 2 sex |
| 133 | 1 male | 1 male | | 1 male | |
| 134 | 2 males/1 female | 2 males/1 female | | 3 total, 1 male | 2 sex |

Table continues on next page.

| Profile Identifier | Actual Combination | MIXplex Minimum Combination | MIXplex Difference | Identifiler® Minimum Combination | Identifiler® Difference |
|---|---|---|---|---|---|
| 135 | 1 male/1 female | 1 male/1 female | | 2 total, 1 male | 1 sex |
| 136 | 1 male/1 female | 1 male/1 female | | 2 total, 1 male | 1 sex |

| Profile Identifier | Actual Combination | MIXplex Minimum Combination | MIXplex Difference | Identifiler® Minimum Combination | Identifiler® Difference |
|---|---|---|---|---|---|
| 137 | 1 male/1 female | 1 male/1 female | | 2 total, 1 male | 1 sex |
| 138 | 1 male | 1 male | | 1 male | |
| 139 | 1 male/1 female | 1 male/1 female | | 2 total, 1 male | 1 sex |
| 140 | 2 males | 2 males | | 2 total, 1 male | 1 sex |
| 141 | 2 males/1 females | 2 males/1 females | | 3 total, 1 male | 2 sex |
| 142 | 2 males/1 females | 2 males/1 females | | 3 total, 1 male | 2 sex |
| 143 | 1 male/2 females | 1 male/2 females | | 3 total, 1 male | 2 sex |
| 144 | 3 females | 2 females | 1 female | 3 females | |
| 145 | 2 males/1 female | 2 males/1 female | | 3 total, 1 male | 2 sex |
| 146 | 2 males/1 female | 2 males/1 female | | 3 total, 1 male | 2 sex |
| 147 | 2 males/1 female | 2 males/1 female | | 3 total, 1 male | 2 sex |
| 148 | 1 male/1 female | 1 male/1 female | | 2 total, 1 male | 1 sex |
| 149 | 1 male/1 female | 1 male/1 female | | 2 total, 1 male | 1 sex |
| 150 | 1 male/3 females | 1 male/2 females | 1 female | 3 total, 1 male | 1 female + 2 sex |
| 151 | 3 males/1 female | 3 males | 1 female | 4 total, 1 male | 3 sex |
| 152 | 1 female | 1 female | | 1 female | |
| 153 | 2 males/2 females | 2 males/1 female | 1 female | 3 total, 1 male | 1 female + 2 sex |
| 154 | 2 males/3 females | 2 males/2 females | 1 female | 3 total, 1 male | 2 females + 2 sex |
| 155 | 3 males/2 females | 3 males/1 female | 1 female | 4 total, 1 male | 1 female + 3 sex |
| 156 | 2 males/2 females | 2 males/1 female | 1 female | 3 total, 1 male | 1 female + 2 sex |
| 157 | 3 males/1 female | 3 males | 1 female | 4 total, 1 male | 3 sex |
| 158 | 2 males/2 females | 2 males/1 female | 1 female | 3 total, 1 male | 1 female + 2 sex |
| 159 | 2 males/3 females | 2 males/2 females | 1 female | 4 total, 1 male | 1 female + 3 sex |
| 160 | 2 females | 2 females | | 2 females | |
| 161 | 2 males/2 females | 2 males/1 female | 1 female | 3 total, 1 male | 1 female + 2 sex |
| 162 | 2 males/3 females | 2 males/2 females | 1 female | 4 total, 1 male | 1 female + 3 sex |
| 163 | 2 females | 2 females | | 2 females | |

Using the MIXplex, in no instance was the determination of the minimum number or sex of contributors incorrect; these basic parameters were correctly inferred for all 63 theoretical mixtures of varying sex and number of contributors without reliance on peak heights. Additionally, for this particular set of theoretical mixtures, the absolute number of male contributors was estimated correctly in every case. Upon closer inspection, this outcome was likely due to the particular DYS alleles present in the male profiles used in this study, which lacked allele sharing at both Y STR markers at the same time. It is possible for male contributors to share the same Y STR alleles at both DYS393 and DYS438, decreasing the minimum number of male contributors inferred. Though Identifiler® analysis allowed correct inference of the minimum number of contributors also, the MIXplex analysis was able to determine this number with more accuracy. Additionally, the only sex information that could be inferred using Identifiler® was the presence or absence of a male.

The maximum difference between the minimum and the actual number and sex of contributors using the MIXplex was 2 females, which occurred with the only 6-person mixture. Identifiler® analysis also differed by these 2 females, and was additionally unable to determine the sex of three contributors to this mixture. For both assays, it is likely that as the total number of contributors increases, the difference between the minimum and the actual number of contributors will increase due to a finite number of possible alleles and the frequency of the most common alleles. A difference between the minimum and the actual contributor combination of 1 female was observed 23 times, or at a frequency of 0.365, using the MIXplex. Consequently, these results indicated that the correct sex and number of contributors was obtained from 62% of the MIXplex profiles from this set of samples. On the other hand, the correct determination of these parameters was only possible for 16% of the profiles generated with the Identifiler® kit. Of note, 6 of the correctly identified profiles (60% of the 16%) were single-source samples.

*7.5. Utility*

The design and potential of the MIXplex combined several key elements of mixture interpretation. Generally, reporting of mixed profiles centres on estimating the minimum number of contributors as well as attempting to assign a sex to the individual contributors in some two-person mixtures. The MIXplex allowed correct identification of the sex and minimum number of contributors in all cases of artificial and theoretical mixtures tested as part of this study, and allowed correct assignment of the actual number and sex of contributors 62% of the time. Currently, with autosomal STRs, sex can only be reliably assigned when both contributors are of the same type, or the male contributor is the minor component of a male-female mixture [322]. Additional testing, such as a Y STR assay, is usually necessary to correctly infer and confirm these characteristics of a mixture, and the MIXplex offers an additional alternative. Corroboration of the suspected number and/or sex of contributors using this assay could direct future analysis, potentially saving time and money. Pre-screening samples thought to contain multiple contributors with this relatively inexpensive assay to 1.) confirm a mixture is present, and 2.) decide which assay, if any, would be most appropriate could eliminate uninformative testing altogether. Additionally, the MIXplex can clarify situations where the male allele at the amelogenin locus is not amplified due to a deletion on the Y chromosome

[279,323] without complete Y STR typing. Moreover, profile subtraction, which is the elimination of alleles from a known contributor (usually female) to the mixture, is simplified in an assay where only four loci of seven markers are found on a female's chromosomes, while all markers are present within a male's chromosomes. Even when a male and a female contributor share alleles, there are an additional four markers at which the male alleles would be the only ones present.

While it is clear from this initial development and characterisation study that the MIXplex cannot solve all of the questions surrounding the interpretation of a mixed profile, there are benefits to its use in certain situations that justify continued study. Additional optimisation of assay parameters such as the reporting threshold and stutter ratios would be helpful to increase confidence in allele calls. Analysis of additional mixtures, both theoretical and actual, could illustrate both the strengths and the limitations of this combination of markers, as well as suggest additional configurations that might aid interpretation even further. Casework mixtures should eventually be evaluated with a final assay in order to assess its performance and value in real-world settings.

Two of the key elements of mixture analysis rest in the initial determination of the minimum number of contributors to the mixed profile as well as the sex of these contributors [322], which the MIXplex helps to address. However, the same authors recognise that a standardised mixture interpretation protocol that will be appropriate for every mixed profile an analyst encounters is not feasible. Where the MIXplex, or an improved version of it, fits into the overall forensic mixture interpretation scheme remains to be uncovered.

# Chapter 8. Conclusions & Impact

The use of X chromosomal short tandem repeat (STR) markers has been greatly increasing in the forensic setting over the past decade. The United States, however, has fallen behind other parts of the world with regards to their implementation. The marker system offers the potential to provide information in addition to what is obtained from autosomal STR systems currently used at crime laboratories and in the courtroom. In certain scenarios, markers on the X chromosome may be the only means of obtaining this information. In-depth characterisation of the marker system is the first step in maximizing the power of this additional tool in the forensic arsenal. Pieces of this puzzle have certainly been addressed, but not comprehensively for a single set of markers in populations relevant to the United States. The combination of results obtained as part of this study form the foundation upon which the routine use of X STRs with U.S. populations may be built, providing a path towards implementation with a finish line in sight.

Guidelines used to validate a marker system such as autosomal or Y STRs for forensic use had been described previously [40,41] and were applied here to the X STR marker system and U.S. populations. The aspects of interest that were addressed are as follows: 1.) selection of suitable markers with the intended purpose in mind (i.e. potential for short amplicons to aid in the analysis of degraded DNA); 2.) development of multiplex assays that are robust and sensitive enough to accommodate targeted sample types; 3.) generation of relevant high-quality population databases large enough to provide sufficient statistical vigour to the conclusions made based on available genetic information; 4.) assessment of the requirements for statistical interpretation of a match between two genetic profiles; and 5.) data compatibility between laboratories.

To begin, the list of potential markers resulting from a review of literature capitalized on the collective knowledge of these established markers and their relevant characteristics, simplifying the process. In the end, fifteen markers were identified for inclusion in two multiplexes: DXS6789, DXS7130, DXS9902, GATA31E08, DXS7424, DXS6795, GATA172D05, DXS10147, GATA165B12, DXS101, DXS8378, DXS7132, DXS6803, HPRTB, and DXS7423. In order to balance number of markers with size of amplicons, not one but two multiplexes were

designed; all amplicons were less than 200 bp to ensure increased utility with potentially degraded extracts [51]. The two proposed multiplexes are exceptional in the world of X STRs because they represent one of the first efforts at combining maximum discrimination with maximum utility for real-world forensic samples. When the project described in these chapters was initially undertaken, there existed but one other study describing the use of mini-X chromosomal STRs [54]. Indeed, seven of the nineteen mini-X STR primer sets investigated for inclusion in the mini-plexes described here were adopted from this work. However, the two multiplexes in the current study contain almost twice the number of markers as well as a marker from each linkage group, unlike the two published quadruplexes that lack a representative from linkage group 3. The two multiplexes described here were also carefully designed to be complementary to one another; markers representative of each proposed linkage group as well as markers overlapping with those present in the commercial kit available at the time (Argus X-UL) were combined in a 10-plex assay, while additional supplementary markers from three of the four proposed linkage groups make up an additional 8-plex. Though not put into practice in this study, one could imagine routine use of the 10-plex to generate X STR profiles for the majority of laboratory samples, adding the 8-plex profile only in cases where additional discriminatory power was required. This approach would be both cost-effective and time-saving for a forensic laboratory seeking to add X STR analysis on degraded samples to their bag of tricks.

Of note, the two assays described here were found to work well with moderately degraded DNA that might represent a typical case sample as well as with high quality DNA obtained from population or reference samples; this is not always the case with commercially available mini-STR kits designed specifically for compromised samples, such as the AmpFℓSTR® MiniFiler™ PCR amplification kit. Often times, profiles generated from high-quality DNA samples using the MiniFiler™ kit suffer from artifacts such as imbalance and pull-up presumably related to an enhanced buffer, smaller primer sets, and increased PCR cycle number targeting compromised samples. Processing high-quality references with a standard kit, such as AmpFℓSTR® Identifiler® PCR Amplification Kit or the PowerPlex® 16 System, avoids the increased time it takes to reprocess these samples or analyze these difficult profiles. The ability to analyze a variety of sample types means that a

separate multiplex with standard-sized amplicons designed solely to generate reference profiles can be eliminated, which is ideal for the use of X STRs in the United States where currently there is no commercial kit available and an in-house solution would need to be implemented.

In order to study the genetic parameters of the chosen set of X STRs in relevant populations, 3,208 unrelated, anonymous individuals from four major U.S. populations (African Americans, U.S. Asians, U.S. Caucasians, and U.S. Hispanics) were typed with the two multiplex assays. Until this study, published U.S. population databases of X STR profiles were limited in size as well as number of markers probed [70,283,292]. Moreover, profiles from a total of 154 unrelated individuals were analyzed as part of the first study of X STRs in a population from Bosnia and Herzegovina. In general for both populations, heterozygosity values for the selected markers were high and the populations were not statistically different from similar published populations, reinforcing the utility of the chosen set of markers.

Both the rate of mutation and the extent of linkage between the 15 markers were also investigated so that the biological processes of mutation and recombination could be taken into account during data interpretation. To begin, overall and marker-specific mutation rates were estimated for the chosen set of 15 markers using 958 U.S. families (African American, U.S. Caucasian, and U.S. Hispanic). In 20,625 meioses, 18 mutations were observed, resulting in an overall mutation rate of $8.73 \times 10^{-4}$ (95% CI: $0.52\text{-}1.4 \times 10^{-3}$). In order to continue to work towards routine use of X STRs for relationship testing in the forensic setting, datasets that combine results from multiple studies serve to maximise the information that can be concluded from individual profiles. Therefore, thirty-four published X STR mutation rate studies were collated here; combining the 20,625 meioses from this study with those from published studies yielded a robust dataset of 81,310 meioses for use by the forensic community. The mutation rate for this combined population was $1.35 \times 10^{-3}$ (95% CI: $1.1\text{-}1.6 \times 10^{-3}$). Both rates are consistent with other studies of X STRs as well as Y and autosomal STRs. As the largest X STR mutation rate study to date, and the only one to investigate U.S. populations, the total number of meioses available to the community for consideration has increased by over 33%.

Also influencing the statistical probability of a match are the phenomena of linkage and linkage disequilibrium. Tested by comparing observed and expected genotypes of population samples, linkage disequilibrium can occur between markers regardless of their physical proximity. Few reproducible instances of linkage disequilibrium could be found for these 15 markers; only 15 pairs (14%) out of a potential 105 pairs showed any indication of linkage disequilibrium in the final combined U.S. population. However linkage disequilibrium is subject to sampling effects and population substructure, which may explain a lack of consistency between this study and other published results. Equally as important, linked markers will have a similar effect on a likelihood ratio, but are measured through recombination events independent of population. To study linkage here, 158 families with several generations and/or multiple offspring were typed and the recombination rate was assessed used two different methods (direct calculation using observed instances of recombination and computer-based analyses as described in [218]). The values obtained using the different methods were generally similar, indicating a robust computation. The hypotheses of complete linkage within linkage groups and of free recombination between linkage groups were both contradicted by the results of this study. The ultimate goal, however, is to gain a better understanding of how potential linkage between this set of 15 X STR markers should direct likelihood calculations in kinship testing. These preliminary results may indicate a need to delve even deeper with more comprehensive analyses and even larger sample sets, as the effect of either linkage disequilibrium or linkage has been shown by others to be non-negligible [218]. Furthermore, strategies to accommodate these two genetic concepts during the calculation of likelihood ratios for both autosomal [265,324,325] and X chromosomal markers [28,326] have already been proposed.

The practical ability to apply a set of X STR markers to forensic samples is also impacted by the comparability of the results between laboratories. Compatible results allow data exchange, such as the generation of shared databases, and helps ensure efficient communication of findings. Important aspects of this comparability can be accessed through techniques such as allele sequencing, which can serve to standardize allele calls. In addition to the routine allele sequencing necessary in this study during the development of the multiplex assays, sequencing of a subset of

samples at each marker was accomplished to verify consistency of the repeat structure with that of published data obtained using larger primer sets. Results revealed variation from published studies in repeat number or structure for four markers (DXS6803, GATA31E08, GATA172D05, and HPRTB). A side effect of sequencing individual alleles was a glimpse into the abundance of variation found within alleles that are otherwise indistinguishable with fragment analysis. Variation was observed within and around the repeat region itself in the form of single-base changes as well as differences in the number of individual repeat units found within a compound repeat region, revealing the potential for increased discrimination possible with the identical set of markers if allelic sequence data were interrogated. Recent advances in sequencing technologies capable of analyzing hundreds of forensically relevant markers at once may allow access to this variation sooner rather than later. The baseline allele sequence data generated for this set of 15 markers could facilitate the incorporation of X STR markers into future workflows, especially for laboratories currently without X STR capabilities.

As a further assessment of the comparability of X STR data between laboratories, a concordance study was completed between the only currently available commercial X STR kit, the QIAGEN® Investigator Argus X-12, and the four overlapping markers also present in the set of 15 markers studied here. Nearly all of the samples typed were concordant, with only 3 instances of discordance due to a null allele (99.92% concordance). The kit provided robust and reliable amplification and proved highly informative for U.S. populations. One concern for routine use of the kit stems from the complexity of the repeat regions of certain markers. Without reconsidering the markers included in the kit, these interpretation concerns could be alleviated through additional sequence data aimed at clarifying the repeat structures, and additional alleles within the ladder better representing observed microvariation. Straightforward repeat structures that can be described and analyzed without ambiguity, such as those probed within the two assays described here, may be favourable to these highly complex regions that test the limits of fragment analysis. Additionally, certain markers within the commercial kit were plagued with null alleles specific to U.S. population groups. This issue can be overcome during the analysis of typical male samples since a lack of amplification is simple to spot (only one allele at each marker is expected); however, the potential for mistyping due to

missing or incorrectly assigned allele calls increases when female samples are considered.

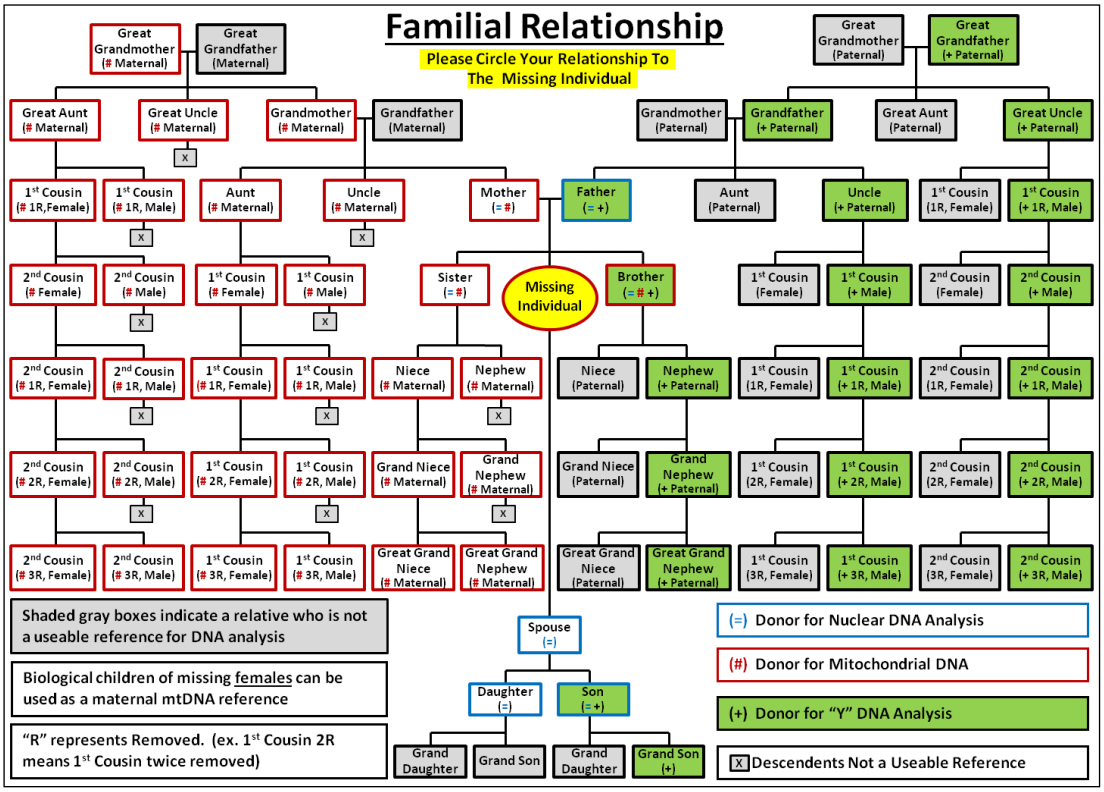
Lastly, the potential for the use of X STRs in other forensic applications was investigated through the creation of a gonosomal STR marker multiplex termed the MIXplex. Incorporating markers from each sex chromosome provided more information regarding the sex of contributors, and in some case the minimum number of contributors, than a multiplex of autosomal STRs and amelogenin with various experimental and theoretical mixed samples. The application is likely limited to cases or laboratories that require such information, but understanding the synergy between these different types of markers was accomplished here and contributes to the body of knowledge characterizing the X STR marker system. Beyond forensic laboratories, the application of these described multiplexes and the marker system itself extend into other scientific arenas such as human evolutionary history and population genetics.

With these goals addressed, X STRs may become more accessible to laboratories currently interested in their potential, but unsure of their practical utility. One such laboratory is the Armed Forces DNA Identification Laboratory (AFDIL), for which the addition of X STR typing could further expand the pool of family reference samples available for comparisons. The old and compromised skeletal remains received by the laboratory must typically be processed using sequencing of the maternally-inherited mitochondrial DNA, accessing the better preserved and abundant mitochondrial genome rather than autosomal markers. Recently, however, a modified Y STR typing procedure has been implemented, allowing the inclusion of certain paternal male relatives in the family reference database to which unknown profiles are compared [327]. Mini-X STRs could provide the link to even more relatives, especially since the assay design and amplification protocols could accommodate the sample type.

Figure 8.1 depicts the family tree used by AFDIL to aid family members in describing their relationship to a missing service member. The individual is asked to identify themselves on the tree, and the potential utility of their profile is assessed. If they are determined not to be a useable reference, no sample is collected; these are

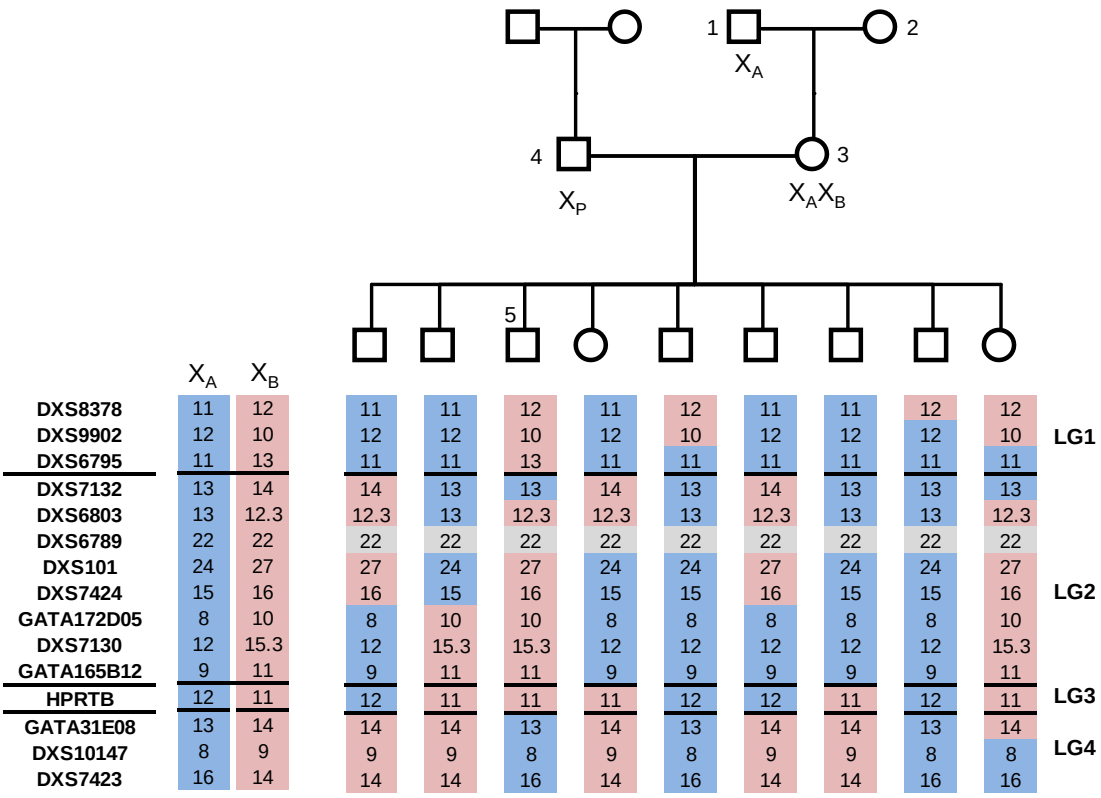
individuals in gray shaded boxes in Figure 8.1, and any descendents not depicted on the tree. In addition to being able to lend further support to results from the lineage markers (mtDNA and Y STRs), X STRs have the potential to turn a number of the "gray relatives" into useful ones, depending on the sex of the missing individual. It would therefore be recommended to obtain DNA samples from these additional relatives in the event that other modes of identification are not fruitful. It is unlikely X STRs will replace current modalities on an everyday basis for personnel accounting, which already obtains a greater than 80% match success rate with currently-employed markers; however if their application in certain specific kinship scenarios might help to bring even one missing service member back to his/her family, then their purpose has been fulfilled. Similarly, other laboratories that currently do not use X STRs may gain a benefit from their use in certain scenarios where only X STRs can provide the required information to catch a criminal or confirm a relationship.

**Figure 8.1. Pedigree depicting potential family references for missing individuals encountered by the Armed Forces DNA Identification Laboratory.**



# Appendix A. Example of Recombination Assessment using a Multigenerational Pedigree

In this example, a three-generation pedigree is used to determine the phase of each X chromosome in the second and third generations, and uncover potential recombination events between adjacent markers and linkage groups.




| | $X_A$ | $X_B$ | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| DXS8378 | 11 | 12 | 11 | 11 | 12 | 11 | 12 | 11 | 11 | 12 | 12 | |
| DXS9902 | 12 | 10 | 12 | 12 | 10 | 12 | 10 | 12 | 12 | 12 | 10 | LG1 |
| DXS6795 | 11 | 13 | 11 | 11 | 13 | 11 | 11 | 11 | 11 | 11 | 11 | |
| DXS7132 | 13 | 14 | 14 | 13 | 13 | 14 | 13 | 14 | 13 | 13 | 13 | |
| DXS6803 | 13 | 12.3 | 12.3 | 13 | 12.3 | 12.3 | 13 | 12.3 | 13 | 13 | 12.3 | |
| DXS6789 | 22 | 22 | 22 | 22 | 22 | 22 | 22 | 22 | 22 | 22 | 22 | |
| DXS101 | 24 | 27 | 27 | 24 | 27 | 24 | 24 | 27 | 24 | 24 | 27 | |
| DXS7424 | 15 | 16 | 16 | 15 | 16 | 15 | 15 | 16 | 15 | 15 | 16 | LG2 |
| GATA172D05 | 8 | 10 | 8 | 10 | 10 | 8 | 8 | 8 | 8 | 8 | 10 | |
| DXS7130 | 12 | 15.3 | 12 | 15.3 | 15.3 | 12 | 12 | 12 | 12 | 12 | 15.3 | |
| GATA165B12 | 9 | 11 | 9 | 11 | 11 | 9 | 9 | 9 | 9 | 9 | 11 | |
| HPRTB | 12 | 11 | 12 | 11 | 11 | 11 | 12 | 12 | 11 | 12 | 11 | LG3 |
| GATA31E08 | 13 | 14 | 14 | 14 | 13 | 14 | 13 | 14 | 14 | 13 | 14 | |
| DXS10147 | 8 | 9 | 9 | 9 | 8 | 9 | 8 | 9 | 9 | 8 | 8 | LG4 |
| DXS7423 | 16 | 14 | 14 | 14 | 16 | 14 | 16 | 14 | 14 | 16 | 16 | |

**Figure A-1.** Maternally-inherited X STR haplotypes for each of the nine children are shown in this figure beside the source haplotypes ($X_A$ and $X_B$) present in the mother (individual 3). Alleles originating from each of the source haplotypes are colour-coded and separated into the four linkage groups. Ambiguous source haplotypes (homozygous markers) are shown in gray. $X_A$: X STR haplotype passed from maternal grandfather (individual 1) in its entirety to mother (individual 3). $X_B$: X STR haplotype present in mother (individual 3) resulting from recombination of the two X STR haplotypes present in the maternal grandmother (individual 2). $X_P$: X STR haplotype present in father (individual 4).

Procedure: The profile of the maternal grandfather (individual) 1 is used to determine the source chromosome ($X_A$ and $X_B$) for each allele present in the mother (individual 3). The profile of the father (individual 4) is used to separate the maternally-inherited alleles present in daughters through process of elimination.

Maternally-inherited X STR haplotypes of children are compared to source haplotypes ($X_A$ and $X_B$) from the mother (individual 3) and then used to infer potential recombination events between markers.

There are several challenges encountered during this analysis that are representative of those encoutnered in a wide variety of the families included in the study. First, homozygous markers are uninformative. In this example, individual 3 possesses a homozygous 22 allele at marker DXS6789. It is therefore impossible to tell from which source haplotype ($X_A$ or $X_B$) an offspring's 22 allele originated. Without this knowledge, recombination between DXS6789 and the two bordering markers (DXS6803 & GATA165B12) cannot be assessed. Though only one marker is homozygous in this example, 290 marker pairs (41%) were rendered uninformative in this study due to homozygous genotypes. Second, mutations must be considered in addition to recombination. In this example, the 13 allele at DXS7132 in the profile of individual 3 was inherited from her father (individual 1). The same allele in individual 5, however, could have been inherited from $X_A$ through two recombination events (between DXS6795 & DXS7132 and between DXS7132 & DXS6803) OR from $X_B$ through a 14→13 mutation. Incorporating information on the rate of mutation could aid in an understanding which scenario is more likely. Laslty, there is a lack of software tools available. Given the unique inheritance pattern of the X chromosome, software tools designed to accommodate autosomal markers are not necessarily helpful for markers on the X chromosome. An ideal tool must be able to simultaneously accommodate both haploid (male) and diploid (female) genotypes.

# Appendix B. Observed Haplotype Frequencies by Linkage Group

**Table B1. Haplotypes observed within sample set A for proposed linkage group 1.** AA: African American, AS: U.S. Asian, CN: U.S. Caucasian, Hisp: U.S. Hispanic, N: number of samples.

| DXS8378 | DXS9902 | DXS6795 | AA N=175 | AS N=201 | CN N=122 | Hisp N=123 | Overall N=621 |
|---|---|---|---|---|---|---|---|
| 8 | 12 | 10 | 1 | | | | 1 |
| 8 | 12 | 15 | 1 | | | | 1 |
| 9 | 10 | 12 | | 1 | | | 1 |
| 9 | 10 | 13 | | 2 | | | 2 |
| 9 | 11 | 8 | 1 | | | | 1 |
| 9 | 11 | 10 | | 1 | | | 1 |
| 9 | 11 | 13 | | 2 | | | 2 |
| 9 | 11 | 14 | | 1 | | | 1 |
| 9 | 12 | 9 | 1 | | | | 1 |
| 10 | 7 | 9 | | 1 | | | 1 |
| 10 | 8 | 9 | | | | 1 | 1 |
| 10 | 8 | 10 | 1 | | | | 1 |
| 10 | 9 | 11 | | | 3 | | 3 |
| 10 | 9 | 12 | | | | 1 | 1 |
| 10 | 9 | 13 | | 1 | 2 | | 3 |
| 10 | 10 | 9 | 1 | 3 | 6 | 4 | 14 |
| 10 | 10 | 10 | 3 | 8 | | 3 | 14 |
| 10 | 10 | 11 | 4 | 11 | 5 | 5 | 25 |
| 10 | 10 | 12 | 1 | 2 | | 4 | 7 |
| 10 | 10 | 13 | 2 | 20 | 5 | 9 | 36 |
| 10 | 10 | 14 | 1 | | | | 1 |
| 10 | 10 | 15 | 1 | | | | 1 |
| 10 | 11 | 9 | 2 | 2 | 9 | 2 | 15 |
| 10 | 11 | 10 | 3 | 4 | | 2 | 9 |
| 10 | 11 | 11 | 1 | 11 | 7 | 4 | 23 |
| 10 | 11 | 12 | 2 | 2 | | 1 | 5 |
| 10 | 11 | 13 | 2 | 19 | 5 | 2 | 28 |
| 10 | 11 | 14 | 1 | | | 1 | 2 |
| 10 | 11 | 15 | 5 | | | | 5 |
| 10 | 11.1 | 11 | | | 1 | 1 | 2 |
| 10 | 12 | 9 | | 2 | 5 | | 7 |
| 10 | 12 | 10 | 2 | 6 | 1 | 1 | 10 |
| 10 | 12 | 11 | 2 | 9 | 3 | 2 | 16 |
| 10 | 12 | 12 | 2 | | | 2 | 4 |
| 10 | 12 | 13 | 3 | 11 | | 4 | 18 |
| 10 | 12 | 14 | | 1 | | | 1 |
| 10 | 12 | 15 | 2 | | | | 2 |
| 10 | 13 | 13 | | 1 | | | 1 |
| 11 | 8 | 13 | 1 | | | | 1 |
| 11 | 9 | 9 | 1 | | | 1 | 2 |
| 11 | 9 | 10 | 2 | | | | 2 |
| 11 | 9 | 11 | 1 | | | | 1 |
| 11 | 9 | 12 | 1 | | | 1 | 2 |
| 11 | 9 | 13 | | | | 1 | 1 |
| 11 | 10 | 9 | 2 | 1 | 2 | 1 | 6 |
| 11 | 10 | 10 | 3 | 2 | 1 | 1 | 7 |
| 11 | 10 | 11 | 1 | 11 | 7 | 4 | 23 |
| 11 | 10 | 12 | 3 | 1 | 1 | 2 | 7 |
| 11 | 10 | 13 | 1 | 10 | 2 | 2 | 15 |

| DXS8378 | DXS9902 | DXS6795 | AA N=175 | AS N=201 | CN N=122 | Hisp N=123 | Overall N=621 |
|---|---|---|---|---|---|---|---|
| 11 | 10 | 14 | 1 | 1 | | | 2 |
| 11 | 10 | 15 | 2 | | | | 2 |
| 11 | 10 | 16 | 1 | | | | 1 |
| 11 | 11 | 9 | 3 | 1 | 4 | 1 | 9 |
| 11 | 11 | 10 | 7 | 3 | 1 | 2 | 13 |
| 11 | 11 | 11 | 8 | 6 | 10 | 3 | 27 |
| 11 | 11 | 12 | 1 | 1 | | 2 | 4 |
| 11 | 11 | 13 | 3 | 7 | 2 | 4 | 16 |
| 11 | 11 | 14 | 1 | | | | 1 |
| 11 | 11 | 15 | 6 | | | | 6 |
| 11 | 11.1 | 9 | | | 1 | | 1 |
| 11 | 11.1 | 13 | | | | 1 | 1 |
| 11 | 12 | 6 | 1 | | | | 1 |
| 11 | 12 | 9 | | | 2 | 2 | 4 |
| 11 | 12 | 10 | 4 | 2 | | | 6 |
| 11 | 12 | 11 | 6 | 4 | 5 | 1 | 16 |
| 11 | 12 | 12 | 2 | 1 | 1 | | 4 |
| 11 | 12 | 13 | | 3 | | 7 | 10 |
| 11 | 12 | 14 | 1 | | | | 1 |
| 11 | 12 | 15 | 5 | | | | 5 |
| 11 | 13 | 12 | 1 | | | | 1 |
| 12 | 8 | 10 | 2 | | | | 2 |
| 12 | 8 | 12 | 1 | | | | 1 |
| 12 | 8 | 15 | 1 | | | | 1 |
| 12 | 9 | 9 | 1 | | 1 | 1 | 3 |
| 12 | 9 | 13 | | 1 | | 1 | 2 |
| 12 | 10 | 9 | 3 | | 5 | 2 | 10 |
| 12 | 10 | 10 | 6 | | | 1 | 7 |
| 12 | 10 | 11 | 1 | 2 | 2 | 1 | 6 |
| 12 | 10 | 12 | 1 | | | 3 | 4 |
| 12 | 10 | 13 | 2 | 4 | 4 | 4 | 14 |
| 12 | 10 | 14 | 1 | | | | 1 |
| 12 | 10 | 15 | 5 | | | | 5 |
| 12 | 11 | 9 | 2 | 3 | 1 | 1 | 7 |
| 12 | 11 | 10 | 6 | | | 2 | 8 |
| 12 | 11 | 11 | 6 | 3 | 5 | 4 | 18 |
| 12 | 11 | 12 | 1 | 1 | | 2 | 4 |
| 12 | 11 | 13 | | 3 | 1 | 5 | 9 |
| 12 | 11 | 15 | 2 | | | | 2 |
| 12 | 11 | 17 | 1 | | | | 1 |
| 12 | 11.1 | 9 | | | 1 | 1 | 2 |
| 12 | 11.1 | 11 | 1 | | | | 1 |
| 12 | 12 | 9 | 1 | | 3 | 2 | 6 |
| 12 | 12 | 10 | 6 | 3 | | 1 | 10 |
| 12 | 12 | 11 | 2 | | 2 | 1 | 5 |
| 12 | 12 | 12 | 1 | | | | 1 |
| 12 | 12 | 13 | 2 | 4 | | 5 | 11 |
| 12 | 12 | 14 | | 1 | | | 1 |
| 12 | 12 | 15 | | | | 1 | 1 |
| 12 | 13 | 11 | | | 1 | | 1 |
| 13 | 8 | 10 | 1 | | | | 1 |
| 13 | 10 | 11 | | | 1 | | 1 |
| 13 | 11 | 9 | 2 | | 1 | | 3 |
| 13 | 11 | 11 | | | 1 | | 1 |
| 13 | 11 | 13 | | 1 | | | 1 |
| 13 | 12 | 9 | | | 1 | | 1 |
| 13 | 12 | 11 | 1 | | | | 1 |
| 13 | 12 | 14 | 1 | | | | 1 |
| 14 | 9 | 14 | 1 | | | | 1 |
| 14 | 11 | 9 | | | 1 | | 1 |

| DXS8378 | DXS9902 | DXS6795 | AA N=175 | AS N=201 | CN N=122 | Hisp N=123 | Overall N=621 |
|---|---|---|---|---|---|---|---|
| Total number of haplotypes: | | | 79 | 49 | 41 | 53 | 109 |
| Total number of unique haplotypes: | | | 39 | 17 | 17 | 23 | 43 |
| Count of most common haplotype: | | | 8 | 20 | 10 | 9 | 36 |

**Table B2. Haplotypes observed within sample set A for proposed linkage group 2.** AA: African American, AS: U.S. Asian, CN: U.S. Caucasian, Hisp: U.S. Hispanic, N: number of samples.

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=175 | AS N=201 | CN N=122 | Hisp N=123 | Overall N=621 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 11 | 10 | 19 | 13 | 28 | 8 | 15.3 | 11 | 1 | | | | 1 |
| 11 | 11 | 21 | 15 | 26 | 10 | 17.3 | 10 | | | | 1 | 1 |
| 11 | 11 | 22 | 16 | 18 | 10 | 14.3 | 11 | 1 | | | | 1 |
| 12 | 8 | 21 | 13 | 28 | 8 | 14.3 | 12 | 1 | | | | 1 |
| 12 | 9 | 19 | 14 | 27 | 10 | 13 | 8 | 1 | | | | 1 |
| 12 | 10 | 15 | 13 | 19 | 6 | 12 | 12 | 1 | | | | 1 |
| 12 | 10 | 20 | 14 | 29 | 10 | 16.3 | 9 | | | 1 | | 1 |
| 12 | 10 | 23 | 15 | 24 | 6 | 14.3 | 11 | 1 | | | | 1 |
| 12 | 11 | 14 | 12 | 21 | 12 | 14.3 | 10 | 1 | | | | 1 |
| 12 | 11 | 15 | 16 | 25 | 9 | 16.3 | 11 | 1 | | | | 1 |
| 12 | 11 | 16 | 16 | 22 | 10 | 11 | 10 | | 1 | | | 1 |
| 12 | 11 | 16 | 16 | 25 | 9 | 12 | 9 | | 1 | | | 1 |
| 12 | 11 | 19 | 14 | 19 | 6 | 13 | 10 | 1 | | | | 1 |
| 12 | 11 | 20 | 13 | 24 | 9 | 13 | 11 | 1 | | | | 1 |
| 12 | 11 | 20 | 15 | 24 | 11 | 14.3 | 10 | | | 1 | | 1 |
| 12 | 11 | 20 | 15 | 27 | 10 | 15.3 | 12 | 1 | | | | 1 |
| 12 | 11 | 20 | 16 | 25 | 6 | 12 | 9 | | | 1 | | 1 |
| 12 | 11 | 21 | 13 | 26 | 10 | 14.3 | 10 | | | | 1 | 1 |
| 12 | 11 | 21 | 14 | 25 | 8 | 14.3 | 11 | 1 | | | | 1 |
| 12 | 11 | 21 | 14 | 27 | 10 | 13 | 11 | | | 1 | | 1 |
| 12 | 11 | 21 | 16 | 19 | 12 | 15.3 | 10 | 1 | | | | 1 |
| 12 | 11 | 21 | 16 | 21 | 10 | 15.3 | 10 | | | 1 | | 1 |
| 12 | 11 | 21 | 16 | 24 | 11 | 11 | 10 | | 1 | | | 1 |
| 12 | 11 | 22 | 13 | 25 | 6 | 14 | 11 | 1 | | | | 1 |
| 12 | 11.3 | 16 | 15 | 23 | 10 | 15.3 | 11 | | 1 | | | 1 |
| 12 | 11.3 | 19 | 16 | 25 | 10 | 15.3 | 11 | | | | 1 | 1 |
| 12 | 11.3 | 21 | 13 | 26 | 10 | 16.3 | 9 | | | 1 | | 1 |
| 12 | 11.3 | 22 | 13 | 25 | 11 | 14.3 | 9 | | | | 1 | 1 |
| 12 | 12 | 20 | 13 | 25 | 11 | 15.3 | 8 | 1 | | | | 1 |
| 12 | 12 | 20 | 15 | 23 | 10 | 12 | 9 | 1 | | | | 1 |
| 12 | 12 | 20 | 15 | 25 | 10 | 12 | 10 | | | | 1 | 1 |
| 12 | 12 | 20 | 15 | 26 | 8 | 15.3 | 10 | | | | 1 | 1 |
| 12 | 12 | 20 | 15 | 27 | 10 | 15.3 | 9 | | | 1 | | 1 |
| 12 | 12 | 20 | 16 | 18 | 9 | 16.3 | 9 | | | 1 | | 1 |
| 12 | 12 | 20 | 16 | 18 | 11 | 14.3 | 10 | | | | 1 | 1 |
| 12 | 12 | 21 | 13 | 22 | 9 | 13 | 10 | 1 | | | | 1 |
| 12 | 12 | 21 | 14 | 22 | 8 | 9 | 11 | 1 | | | | 1 |
| 12 | 12 | 21 | 16 | 25 | 6 | 15.3 | 10 | 1 | | | | 1 |
| 12 | 12 | 22 | 13 | 24 | 11 | 11 | 10 | | | 1 | | 1 |
| 12 | 12 | 22 | 16 | 23 | 6 | 13 | 12 | 1 | | | | 1 |
| 12 | 12 | 23 | 17 | 15 | 10 | 15.3 | 11 | 1 | | | | 1 |
| 12 | 12.3 | 15 | 16 | 25 | 10 | 15.3 | 11 | | 1 | | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=175 | AS N=201 | CN N=122 | Hisp N=123 | Overall N=621 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 12 | 12.3 | 16 | 16 | 24 | 10 | 11 | 11 | | 1 | | | 1 |
| 12 | 12.3 | 19 | 16 | 23 | 11 | 16.3 | 10 | | | 1 | | 1 |
| 12 | 12.3 | 20 | 15 | 25 | 11 | 14 | 9 | | 1 | | | 1 |
| 12 | 12.3 | 20 | 15 | 26 | 6 | 12 | 10 | | | | 1 | 1 |
| 12 | 12.3 | 20 | 16 | 22 | 12 | 13 | 10 | | | | 1 | 1 |
| 12 | 12.3 | 21 | 15 | 24 | 10 | 15.3 | 10 | | 1 | | | 1 |
| 12 | 12.3 | 21 | 16 | 24 | 10 | 12 | 9 | | | | 1 | 1 |
| 12 | 12.3 | 21 | 16 | 25 | 10 | 12 | 10 | | | | 1 | 1 |
| 12 | 12.3 | 21 | 16 | 26 | 8 | 11 | 10 | | 1 | | | 1 |
| 12 | 13 | 16 | 16 | 23 | 12 | 14.3 | 9 | | 1 | | | 1 |
| 12 | 13 | 20 | 12 | 25 | 10 | 14.3 | 9 | | | 1 | | 1 |
| 12 | 13 | 20 | 15 | 24 | 8 | 12 | 11 | | 1 | | | 1 |
| 12 | 13 | 20 | 15 | 26 | 11 | 17.3 | 11 | | 1 | | | 1 |
| 12 | 13 | 21 | 16 | 27 | 10 | 16.3 | 9 | | 1 | | | 1 |
| 12 | 13.3 | 20 | 15 | 25 | 10 | 14.3 | 10 | | 1 | | | 1 |
| 12 | 13.3 | 21 | 15 | 19 | 11 | 15.3 | 10 | | | | 1 | 1 |
| 12 | 13.3 | 21 | 17 | 23 | 11 | 16.3 | 11 | | | 1 | | 1 |
| 12 | 13.3 | 22 | 12 | 28 | 6 | 15.3 | 9 | | | | 1 | 1 |
| 12 | 13.3 | 22 | 15 | 25 | 6 | 14.3 | 11 | | | 1 | | 1 |
| 13 | 7 | 22 | 12 | 20 | 6 | 13 | 10 | | | | 1 | 1 |
| 13 | 8 | 22 | 16 | 26 | 6 | 12 | 11 | 1 | | | | 1 |
| 13 | 10 | 15 | 13 | 21 | 12 | 15.3 | 9 | 1 | | | | 1 |
| 13 | 10 | 15 | 13 | 24 | 10 | 14.3 | 10 | 1 | | | | 1 |
| 13 | 10 | 20 | 16 | 18 | 8 | 15.3 | 10 | | | 1 | | 1 |
| 13 | 10 | 21 | 17 | 18 | 6 | 12 | 9 | 1 | | | | 1 |
| 13 | 10 | 22 | 16 | 28 | 10 | 14.3 | 11 | 1 | | | | 1 |
| 13 | 11 | 15 | 14 | 21 | 9 | 13 | 11 | 1 | | | | 1 |
| 13 | 11 | 15 | 14 | 22 | 9 | 15.3 | 10 | 1 | | | | 1 |
| 13 | 11 | 15 | 16 | 24 | 9 | 12 | 11 | 1 | | | | 1 |
| 13 | 11 | 16 | 12 | 24 | 9 | 12 | 10 | 1 | | | | 1 |
| 13 | 11 | 16 | 13 | 25 | 8 | 15.3 | 10 | | 1 | | | 1 |
| 13 | 11 | 16 | 14 | 18 | 7 | 12 | 12 | 1 | | | | 1 |
| 13 | 11 | 16 | 14 | 25 | 9 | 13 | 11 | 1 | | | | 1 |
| 13 | 11 | 16 | 15 | 25 | 11 | 12 | 9 | | 1 | | | 1 |
| 13 | 11 | 16 | 16 | 27 | 8 | 12 | 10 | | 1 | | | 1 |
| 13 | 11 | 18 | 15 | 19 | 11 | 13 | 10 | | | | 1 | 1 |
| 13 | 11 | 19 | 17 | 25 | 10 | 14.3 | 10 | | | | 1 | 1 |
| 13 | 11 | 19 | 19 | 25 | 8 | 13 | 11 | 1 | | | | 1 |
| 13 | 11 | 20 | 11 | 22 | 6 | 15.3 | 9 | 1 | | | | 1 |
| 13 | 11 | 20 | 11 | 23 | 9 | 14.3 | 10 | 1 | | | | 1 |
| 13 | 11 | 20 | 13 | 24 | 12 | 12 | 10 | 1 | | | | 1 |
| 13 | 11 | 20 | 13 | 26 | 6 | 9 | 9 | | | 1 | | 1 |
| 13 | 11 | 20 | 13 | 27 | 9 | 13 | 10 | 1 | | | | 1 |
| 13 | 11 | 20 | 14 | 24 | 9 | 15.3 | 11 | | | | 1 | 1 |
| 13 | 11 | 20 | 14 | 24 | 12 | 12 | 11 | | | | 1 | 1 |
| 13 | 11 | 20 | 15 | 26 | 8 | 14.3 | 11 | | | 1 | | 1 |
| 13 | 11 | 20 | 16 | 24 | 11 | 14.3 | 10 | 1 | | | | 1 |
| 13 | 11 | 20 | 16 | 25 | 11 | 12 | 10 | | | | 1 | 1 |
| 13 | 11 | 20 | 16 | 27 | 6 | 11 | 11 | 1 | | | | 1 |
| 13 | 11 | 21 | 12 | 24 | 9 | 13 | 10 | 1 | | | | 1 |
| 13 | 11 | 21 | 13 | 21 | 10 | 12 | 11 | 1 | | | | 1 |
| 13 | 11 | 21 | 13 | 27 | 8 | 15.3 | 11 | 1 | | | | 1 |
| 13 | 11 | 21 | 14 | 24 | 10 | 16.3 | 11 | | 1 | | | 1 |
| 13 | 11 | 21 | 15 | 24 | 10 | 14.3 | 9 | | | 1 | | 1 |
| 13 | 11 | 21 | 15 | 24 | 11 | 12 | 10 | | 1 | | | 1 |
| 13 | 11 | 21 | 15 | 25 | 8 | 14.3 | 10 | | | 1 | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=175 | AS N=201 | CN N=122 | Hisp N=123 | Overall N=621 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 13 | 11 | 21 | 15 | 26 | 11 | 13 | 10 | | | | 1 | 1 |
| 13 | 11 | 21 | 15 | 28 | 6 | 15.3 | 9 | | | 1 | | 1 |
| 13 | 11 | 21 | 16 | 18 | 10 | 13 | 11 | | | 1 | | 1 |
| 13 | 11 | 21 | 16 | 26 | 6 | 12 | 9 | | | | 1 | 1 |
| 13 | 11 | 21 | 16 | 26 | 11 | 12 | 10 | | 1 | | | 1 |
| 13 | 11 | 21 | 17 | 27 | 6 | 16.3 | 10 | | | 1 | | 1 |
| 13 | 11 | 21 | 18 | 15 | 8 | 12 | 9 | | | 1 | | 1 |
| 13 | 11 | 22 | 13 | 25 | 6 | 14.3 | 10 | 1 | | | | 1 |
| 13 | 11 | 22 | 14 | 28 | 12 | 14.3 | 10 | | | 1 | | 1 |
| 13 | 11 | 22 | 15 | 26 | 8 | 14.3 | 11 | | | 1 | | 1 |
| 13 | 11 | 23 | 13 | 30 | 7 | 14.3 | 10 | 1 | | | | 1 |
| 13 | 11 | 23 | 15 | 27 | 6 | 13.3 | 9 | | 1 | | | 1 |
| 13 | 11 | 23 | 17 | 18 | 11 | 15.3 | 11 | | | | 1 | 1 |
| 13 | 11 | 23 | 17 | 24 | 12 | 12 | 10 | | | | 1 | 1 |
| 13 | 11 | 24 | 16 | 20 | 10 | 13 | 9 | | | 1 | | 1 |
| 13 | 11 | 25 | 14 | 24 | 9 | 12 | 11 | 1 | | | | 1 |
| 13 | 11.3 | 16 | 16 | 29 | 10 | 11 | 10 | | 1 | | | 1 |
| 13 | 11.3 | 21 | 14 | 24 | 11 | 12 | 10 | | | | 1 | 1 |
| 13 | 11.3 | 24 | 16 | 21 | 10 | 15.3 | 10 | | 1 | | | 1 |
| 13 | 12 | 15 | 16 | 24 | 10 | 15.3 | 10 | | 1 | | | 1 |
| 13 | 12 | 15 | 16 | 25 | 6 | 14.3 | 10 | | | | 1 | 1 |
| 13 | 12 | 16 | 13 | 24 | 11 | 16.3 | 11 | | 1 | | | 1 |
| 13 | 12 | 16 | 14 | 21 | 9 | 12 | 8 | 1 | | | | 1 |
| 13 | 12 | 16 | 15 | 23 | 10 | 14.3 | 11 | | 1 | | | 1 |
| 13 | 12 | 16 | 15 | 25 | 8 | 14.3 | 9 | | 1 | | | 1 |
| 13 | 12 | 16 | 17 | 25 | 12 | 15.3 | 9 | | 1 | | | 1 |
| 13 | 12 | 17 | 13 | 20 | 9 | 14.3 | 11 | 1 | | | | 1 |
| 13 | 12 | 17 | 14 | 24 | 12 | 17.3 | 11 | | | 1 | | 1 |
| 13 | 12 | 19 | 13 | 26 | 8 | 12 | 10 | | | | 1 | 1 |
| 13 | 12 | 20 | 12 | 26 | 11 | 15.3 | 12 | | | | 1 | 1 |
| 13 | 12 | 20 | 13 | 26 | 6 | 15.3 | 10 | 1 | | | | 1 |
| 13 | 12 | 20 | 13 | 26 | 10 | 12 | 9 | | 1 | | | 1 |
| 13 | 12 | 20 | 13 | 26 | 10 | 14.3 | 10 | | | | 1 | 1 |
| 13 | 12 | 20 | 14 | 26 | 12 | 12 | 11 | 1 | | | | 1 |
| 13 | 12 | 20 | 14 | 27 | 10 | 12 | 11 | | | | 1 | 1 |
| 13 | 12 | 20 | 14 | 28 | 10 | 15.3 | 10 | | 1 | | | 1 |
| 13 | 12 | 20 | 15 | 26 | 10 | 16.3 | 10 | | | | 1 | 1 |
| 13 | 12 | 20 | 16 | 23 | 11 | 15.3 | 10 | | | 1 | | 1 |
| 13 | 12 | 20 | 16 | 24 | 12 | 12 | 10 | | | 1 | | 1 |
| 13 | 12 | 20 | 16 | 27 | 11 | 15.3 | 10 | | | | 1 | 1 |
| 13 | 12 | 20 | 17 | 25 | 9 | 15.3 | 9 | | 1 | | | 1 |
| 13 | 12 | 20 | 17 | 30 | 10 | 15.3 | 9 | | | 1 | | 1 |
| 13 | 12 | 21 | 13 | 22 | 7 | 9 | 11 | 1 | | | | 1 |
| 13 | 12 | 21 | 13 | 26 | 12 | 15.3 | 9 | | | 1 | | 1 |
| 13 | 12 | 21 | 14 | 24 | 10 | 12 | 10 | | | 1 | | 1 |
| 13 | 12 | 21 | 15 | 23 | 8 | 15.3 | 11 | 1 | | | | 1 |
| 13 | 12 | 21 | 17 | 18 | 8 | 13.3 | 12 | 1 | | | | 1 |
| 13 | 12 | 21 | 17 | 19 | 11 | 16.3 | 9 | | | | 1 | 1 |
| 13 | 12 | 21 | 17 | 25 | 10 | 14.3 | 10 | | | 1 | | 1 |
| 13 | 12 | 22 | 14 | 24 | 9 | 12 | 10 | | | | 1 | 1 |
| 13 | 12 | 22 | 14 | 26 | 10 | 12 | 9 | | 1 | | | 1 |
| 13 | 12 | 22 | 16 | 24 | 10 | 15.3 | 10 | | | | 1 | 1 |
| 13 | 12 | 22 | 16 | 25 | 10 | 14 | 9 | | | 1 | | 1 |
| 13 | 12 | 22 | 16 | 25 | 10 | 15.3 | 11 | | | | 1 | 1 |
| 13 | 12 | 23 | 15 | 30 | 6 | 14.3 | 9 | | | 1 | | 1 |
| 13 | 12 | 23 | 15 | 30 | 10 | 13 | 9 | | | | 1 | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=175 | AS N=201 | CN N=122 | Hisp N=123 | Overall N=621 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 13 | 12 | 23 | 16 | 24 | 10 | 12 | 9 | | | | 1 | 1 |
| 13 | 12 | 23 | 17 | 18 | 11 | 16.3 | 9 | | | 1 | | 1 |
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| 13 | 12.3 | 20 | 16 | 27 | 10 | 12 | 9 | | | | 1 | 1 |
| 13 | 12.3 | 21 | 13 | 21 | 12 | 14.3 | 9 | | | | 1 | 1 |
| 13 | 12.3 | 21 | 14 | 24 | 11 | 11 | 10 | | 1 | | | 1 |
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| 13 | 12.3 | 22 | 14 | 26 | 11 | 14.3 | 11 | | | 1 | | 1 |
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| 13 | 13.3 | 21 | 16 | 26 | 10 | 15.3 | 11 | 1 | | | | 1 |
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| 13 | 14 | 20 | 16 | 25 | 9 | 15.3 | 10 | | | 1 | | 1 |
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| 13 | 14.3 | 15 | 16 | 24 | 10 | 15.3 | 11 | | | 1 | | 1 |
| 13 | 14.3 | 20 | 16 | 17 | 12 | 13.3 | 9 | | | 1 | | 1 |
| 13 | 16 | 22 | 16 | 25 | 8 | 14.3 | 11 | 1 | | | | 1 |
| 14 | 9 | 18 | 14 | 27 | 9 | 14.3 | 11 | 1 | | | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=175 | AS N=201 | CN N=122 | Hisp N=123 | Overall N=621 |
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| 14 | 10 | 20 | 18 | 18 | 10 | 14.3 | 11 | | | 1 | | 1 |
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| 14 | 10 | 22 | 14 | 23 | 10 | 16.3 | 10 | 1 | | | | 1 |
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| 14 | 11 | 16 | 15 | 18 | 11 | 16.3 | 11 | 1 | | | | 1 |
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| 14 | 11 | 16 | 15 | 23 | 10 | 12 | 10 | | 1 | | | 1 |
| 14 | 11 | 16 | 15 | 25 | 8 | 15.3 | 9 | | 1 | | | 1 |
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| 14 | 11 | 16 | 15 | 27 | 9 | 12 | 10 | | 1 | | | 1 |
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| 14 | 11 | 20 | 15 | 26 | 8 | 15.3 | 9 | | 1 | | | 1 |
| 14 | 11 | 20 | 15 | 26 | 8 | 15.3 | 12 | | 1 | | | 1 |
| 14 | 11 | 20 | 15 | 26 | 12 | 17.3 | 9 | | | 1 | | 1 |
| 14 | 11 | 21 | 13 | 27 | 10 | 15.3 | 11 | | | | 1 | 1 |
| 14 | 11 | 21 | 14 | 28 | 9 | 15.3 | 10 | 1 | | | | 1 |
| 14 | 11 | 21 | 14 | 31 | 8 | 13 | 10 | 1 | | | | 1 |
| 14 | 11 | 21 | 15 | 24 | 8 | 15.3 | 10 | | | | 1 | 1 |
| 14 | 11 | 21 | 15 | 25 | 8 | 16.3 | 11 | | | 1 | | 1 |
| 14 | 11 | 21 | 15 | 25 | 9 | 12 | 11 | | 1 | | | 1 |
| 14 | 11 | 21 | 15 | 25 | 11 | 16.3 | 10 | | | | 1 | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=175 | AS N=201 | CN N=122 | Hisp N=123 | Overall N=621 |
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| 14 | 11 | 21 | 15 | 26 | 9 | 14.3 | 10 | | | | 1 | 1 |
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| 14 | 11 | 21 | 16 | 24 | 11 | 12 | 10 | | 1 | | | 1 |
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| 14 | 11 | 23 | 16 | 17 | 11 | 14.3 | 11 | | | 1 | | 1 |
| 14 | 11 | 23 | 16 | 27 | 6 | 12 | 12 | 1 | | | | 1 |
| 14 | 11 | 24 | 15 | 24 | 6 | 14.3 | 11 | 1 | | | | 1 |
| 14 | 11.3 | 16 | 13 | 26 | 12 | 15.3 | 9 | | | | 1 | 1 |
| 14 | 11.3 | 16 | 15 | 28 | 6 | 15.3 | 11 | | 1 | | | 1 |
| 14 | 11.3 | 16 | 16 | 27 | 12 | 12 | 11 | | | | 1 | 1 |
| 14 | 11.3 | 20 | 13 | 24 | 6 | 15.3 | 11 | 1 | | | | 1 |
| 14 | 11.3 | 20 | 15 | 25 | 9 | 12 | 10 | | 1 | | | 1 |
| 14 | 11.3 | 20 | 15 | 27 | 11 | 14.3 | 11 | 1 | | | | 1 |
| 14 | 12 | 15 | 12 | 28 | 8 | 15.3 | 10 | 1 | | | | 1 |
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| 14 | 12 | 15 | 16 | 19 | 7 | 15.3 | 12 | 1 | | | | 1 |
| 14 | 12 | 15 | 16 | 22 | 10 | 15.3 | 10 | | 1 | | | 1 |
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| 14 | 12 | 16 | 14 | 26 | 9 | 11 | 11 | | 1 | | | 1 |
| 14 | 12 | 16 | 14 | 26 | 10 | 11 | 10 | | 1 | | | 1 |
| 14 | 12 | 16 | 15 | 23 | 11 | 14.3 | 9 | | 1 | | | 1 |
| 14 | 12 | 16 | 15 | 26 | 11 | 15.3 | 10 | | 1 | | | 1 |
| 14 | 12 | 16 | 15 | 27 | 11 | 16.3 | 9 | 1 | | | | 1 |
| 14 | 12 | 16 | 16 | 25 | 11 | 16.3 | 11 | | | 1 | | 1 |
| 14 | 12 | 16 | 16 | 29 | 10 | 16.3 | 10 | | 1 | | | 1 |
| 14 | 12 | 19 | 14 | 18 | 6 | 15.3 | 10 | | | | 1 | 1 |
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| 14 | 12 | 20 | 15 | 19 | 12 | 15.3 | 11 | | | 1 | | 1 |
| 14 | 12 | 20 | 15 | 24 | 11 | 12 | 11 | | | | 1 | 1 |
| 14 | 12 | 20 | 15 | 26 | 11 | 15.3 | 10 | | | | 1 | 1 |
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| 14 | 12 | 20 | 16 | 24 | 11 | 14.3 | 11 | | | 1 | | 1 |
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| 14 | 12 | 20 | 17 | 25 | 11 | 14.3 | 11 | | | | 1 | 1 |
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| 14 | 12 | 21 | 13 | 19 | 9 | 13 | 10 | | | | 1 | 1 |
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| 14 | 12 | 21 | 14 | 23 | 8 | 13 | 11 | | | 1 | | 1 |
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| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=175 | AS N=201 | CN N=122 | Hisp N=123 | Overall N=621 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 14 | 12 | 21 | 15 | 26 | 10 | 14.3 | 9 | 1 | | | | 1 |
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| 14 | 12.3 | 21 | 16 | 19 | 10 | 15.3 | 11 | | 1 | | | 1 |
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| 14 | 12.3 | 22 | 14 | 26 | 10 | 13 | 9 | | 1 | | | 1 |
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| 14 | 12.3 | 22 | 16 | 24 | 13 | 14.3 | 11 | | | 1 | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=175 | AS N=201 | CN N=122 | Hisp N=123 | Overall N=621 |
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| 14 | 13.3 | 16 | 15 | 15 | 11 | 14 | 9 | | | 1 | | 1 |
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| 14 | 13.3 | 17 | 15 | 27 | 11 | 11 | 10 | | 1 | | | 1 |
| 14 | 13.3 | 20 | 12 | 26 | 7 | 14.3 | 9 | | | 1 | | 1 |
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| 15 | 10 | 16 | 14 | 21 | 9 | 14 | 11 | 1 | | | | 1 |
| 15 | 10 | 19 | 13 | 28 | 10 | 13 | 10 | 1 | | | | 1 |
| 15 | 10 | 21 | 13 | 26 | 10 | 12 | 11 | 1 | | | | 1 |
| 15 | 10 | 21 | 16 | 20 | 9 | 13 | 9 | 1 | | | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=175 | AS N=201 | CN N=122 | Hisp N=123 | Overall N=621 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 15 | 10 | 21 | 16 | 25 | 10 | 15.3 | 9 | | 1 | | | 1 |
| 15 | 10 | 22 | 15 | 20 | 8 | 12 | 10 | 1 | | | | 1 |
| 15 | 10 | 23 | 14 | 28 | 10 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 11 | 14 | 14 | 27 | 10 | 15.3 | 11 | | 1 | | | 1 |
| 15 | 11 | 15 | 12 | 23 | 8 | 13.3 | 9 | 1 | | | | 1 |
| 15 | 11 | 15 | 13 | 28 | 9 | 16.3 | 10 | 1 | | | | 1 |
| 15 | 11 | 15 | 15 | 21 | 9 | 11 | 10 | 1 | | | | 1 |
| 15 | 11 | 15 | 15 | 25 | 9 | 14 | 9 | 1 | | | | 1 |
| 15 | 11 | 15 | 15 | 26 | 10 | 13 | 11 | 1 | | | | 1 |
| 15 | 11 | 15 | 15 | 27 | 9 | 13 | 9 | 1 | | | | 1 |
| 15 | 11 | 15 | 15 | 28 | 11 | 15.3 | 9 | | | | 1 | 1 |
| 15 | 11 | 16 | 11 | 19 | 6 | 15.3 | 10 | 1 | | | | 1 |
| 15 | 11 | 16 | 11 | 25 | 7 | 14.3 | 11 | 1 | | | | 1 |
| 15 | 11 | 16 | 13 | 26 | 8 | 11 | 9 | | | | 1 | 1 |
| 15 | 11 | 16 | 13 | 27 | 10 | 12 | 8 | | | | 1 | 1 |
| 15 | 11 | 16 | 15 | 24 | 8 | 15.3 | 10 | | 1 | | | 1 |
| 15 | 11 | 16 | 15 | 27 | 6 | 14.3 | 12 | 1 | | | | 1 |
| 15 | 11 | 16 | 16 | 24 | 8 | 11 | 9 | 1 | | | | 1 |
| 15 | 11 | 16 | 16 | 24 | 8 | 16.3 | 12 | | 1 | | | 1 |
| 15 | 11 | 16 | 16 | 24 | 10 | 12 | 12 | | 1 | | | 1 |
| 15 | 11 | 16 | 16 | 24 | 12 | 15.3 | 10 | | 1 | | | 1 |
| 15 | 11 | 16 | 16 | 27 | 10 | 13 | 10 | | 1 | | | 1 |
| 15 | 11 | 16 | 17 | 28 | 10 | 11 | 9 | | 1 | | | 1 |
| 15 | 11 | 16 | 18 | 24 | 10 | 12 | 9 | | | | 1 | 1 |
| 15 | 11 | 20 | 14 | 27 | 10 | 15.3 | 11 | 1 | | | | 1 |
| 15 | 11 | 20 | 16 | 19 | 8 | 13.3 | 11 | 1 | | | | 1 |
| 15 | 11 | 20 | 16 | 20 | 6 | 16.3 | 11 | 1 | | | | 1 |
| 15 | 11 | 20 | 16 | 24 | 11 | 11 | 10 | | | | 1 | 1 |
| 15 | 11 | 20 | 16 | 25 | 10 | 11 | 10 | | 1 | | | 1 |
| 15 | 11 | 20 | 16 | 26 | 10 | 12 | 10 | | 1 | | | 1 |
| 15 | 11 | 20 | 16 | 26 | 11 | 15.3 | 10 | | 1 | | | 1 |
| 15 | 11 | 20 | 17 | 24 | 11 | 13 | 9 | | 1 | | | 1 |
| 15 | 11 | 21 | 13 | 25 | 12 | 13.3 | 11 | | | 1 | | 1 |
| 15 | 11 | 21 | 14 | 21 | 6 | 16.3 | 9 | | | 1 | | 1 |
| 15 | 11 | 21 | 15 | 28 | 10 | 12 | 10 | | 1 | | | 1 |
| 15 | 11 | 21 | 16 | 26 | 10 | 14.3 | 11 | | | | 1 | 1 |
| 15 | 11 | 21 | 17 | 18 | 8 | 15.3 | 11 | 1 | | | | 1 |
| 15 | 11 | 21 | 18 | 15 | 12 | 14.3 | 9 | | | 1 | | 1 |
| 15 | 11 | 22 | 14 | 24 | 12 | 14.3 | 10 | | | | 1 | 1 |
| 15 | 11 | 22 | 18 | 24 | 10 | 12 | 11 | | | | 1 | 1 |
| 15 | 11 | 23 | 16 | 15 | 10 | 16.3 | 11 | | | 1 | | 1 |
| 15 | 11 | 23 | 16 | 18 | 8 | 14.3 | 10 | 1 | | | | 1 |
| 15 | 11 | 23 | 16 | 19 | 11 | 12 | 11 | | | | 1 | 1 |
| 15 | 11.3 | 15 | 16 | 26 | 8 | 15.3 | 10 | | 1 | | | 1 |
| 15 | 11.3 | 16 | 15 | 24 | 10 | 15.3 | 11 | | 1 | | | 1 |
| 15 | 11.3 | 20 | 14 | 28 | 11 | 11 | 10 | | | 1 | | 1 |
| 15 | 11.3 | 20 | 16 | 24 | 10 | 12 | 11 | | 1 | | | 1 |
| 15 | 11.3 | 20 | 17 | 25 | 10 | 15.3 | 10 | | 1 | | | 1 |
| 15 | 11.3 | 21 | 15 | 24 | 9 | 11 | 11 | 1 | | | | 1 |
| 15 | 11.3 | 22 | 14 | 24 | 8 | 11 | 10 | | 1 | | | 1 |
| 15 | 12 | 15 | 10 | 27 | 9 | 12 | 10 | 1 | | | | 1 |
| 15 | 12 | 15 | 13 | 22 | 11 | 14.3 | 9 | | | | 1 | 1 |
| 15 | 12 | 15 | 14 | 24 | 11 | 15.3 | 12 | | | 1 | | 1 |
| 15 | 12 | 15 | 15 | 23 | 11 | 12 | 10 | | 1 | | | 1 |
| 15 | 12 | 15 | 16 | 25 | 11 | 15.3 | 9 | | 1 | | | 1 |
| 15 | 12 | 15 | 16 | 26 | 8 | 14 | 10 | 1 | | | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=175 | AS N=201 | CN N=122 | Hisp N=123 | Overall N=621 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 15 | 12 | 16 | 14 | 20 | 9 | 12 | 11 | 1 | | | | 1 |
| 15 | 12 | 16 | 15 | 23 | 11 | 16.3 | 10 | | 1 | | | 1 |
| 15 | 12 | 16 | 15 | 25 | 11 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 12 | 16 | 16 | 25 | 9 | 12 | 9 | | 1 | | | 1 |
| 15 | 12 | 16 | 16 | 28 | 8 | 12 | 9 | | 1 | | | 1 |
| 15 | 12 | 18 | 16 | 27 | 7 | 13 | 10 | 1 | | | | 1 |
| 15 | 12 | 19 | 15 | 25 | 10 | 15.3 | 9 | | | | 1 | 1 |
| 15 | 12 | 20 | 13 | 26 | 6 | 11 | 9 | | | | 1 | 1 |
| 15 | 12 | 20 | 13 | 26 | 10 | 14.3 | 10 | | | | 1 | 1 |
| 15 | 12 | 20 | 14 | 24 | 10 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 12 | 20 | 14 | 27 | 11 | 14 | 8 | | | | 1 | 1 |
| 15 | 12 | 20 | 15 | 24 | 11 | 13.3 | 9 | 1 | | | | 1 |
| 15 | 12 | 20 | 15 | 24 | 11 | 15.3 | 10 | | 1 | | | 1 |
| 15 | 12 | 20 | 15 | 26 | 10 | 14.3 | 9 | | | | 1 | 1 |
| 15 | 12 | 20 | 15 | 27 | 10 | 13.3 | 9 | | | | 1 | 1 |
| 15 | 12 | 20 | 16 | 25 | 6 | 11 | 10 | 1 | | | | 1 |
| 15 | 12 | 20 | 17 | 26 | 8 | 15.3 | 13 | 1 | | | | 1 |
| 15 | 12 | 21 | 12 | 26 | 6 | 12 | 10 | 1 | | | | 1 |
| 15 | 12 | 21 | 13 | 26 | 10 | 14.3 | 10 | | | | 1 | 1 |
| 15 | 12 | 21 | 13 | 26 | 11 | 16.3 | 10 | 1 | | | | 1 |
| 15 | 12 | 21 | 14 | 27 | 6 | 15.3 | 11 | | | 1 | | 1 |
| 15 | 12 | 21 | 15 | 24 | 11 | 14 | 10 | | | | 1 | 1 |
| 15 | 12 | 21 | 16 | 23 | 9 | 12 | 9 | 1 | | | | 1 |
| 15 | 12 | 21 | 16 | 25 | 10 | 11 | 11 | | | 1 | | 1 |
| 15 | 12 | 21 | 16 | 25 | 11 | 14.3 | 10 | | | 1 | | 1 |
| 15 | 12 | 21 | 16 | 26 | 8 | 15.3 | 10 | | 1 | | | 1 |
| 15 | 12 | 22 | 16 | 24 | 10 | 15.3 | 10 | | 1 | | | 1 |
| 15 | 12 | 23 | 14 | 24 | 6 | 14.3 | 11 | | | 1 | | 1 |
| 15 | 12 | 23 | 17 | 18 | 6 | 15.3 | 11 | | | 1 | | 1 |
| 15 | 12.3 | 15 | 15 | 23 | 10 | 15.3 | 9 | | 1 | | | 1 |
| 15 | 12.3 | 15 | 15 | 24 | 6 | 15.3 | 11 | | | 1 | | 1 |
| 15 | 12.3 | 15 | 15 | 26 | 8 | 15.3 | 10 | | 1 | | | 1 |
| 15 | 12.3 | 15 | 16 | 23 | 9 | 14.3 | 10 | | 1 | | | 1 |
| 15 | 12.3 | 15 | 16 | 25 | 11 | 15.3 | 9 | | 1 | | | 1 |
| 15 | 12.3 | 16 | 11 | 25 | 10 | 11 | 10 | | 1 | | | 1 |
| 15 | 12.3 | 16 | 12 | 25 | 10 | 15.3 | 9 | | 1 | | | 1 |
| 15 | 12.3 | 16 | 14 | 23 | 8 | 15.3 | 9 | | | | 1 | 1 |
| 15 | 12.3 | 16 | 14 | 24 | 8 | 12 | 10 | | 1 | | | 1 |
| 15 | 12.3 | 16 | 15 | 23 | 8 | 10 | 10 | | 1 | | | 1 |
| 15 | 12.3 | 16 | 15 | 24 | 6 | 16.3 | 10 | | 1 | | | 1 |
| 15 | 12.3 | 16 | 15 | 25 | 10 | 12 | 9 | | 1 | | | 1 |
| 15 | 12.3 | 16 | 16 | 22 | 8 | 16.3 | 10 | | 1 | | | 1 |
| 15 | 12.3 | 16 | 16 | 24 | 10 | 14.3 | 9 | | | | 1 | 1 |
| 15 | 12.3 | 16 | 16 | 25 | 10 | 12 | 10 | | 1 | | | 1 |
| 15 | 12.3 | 16 | 16 | 28 | 9 | 11 | 10 | | 1 | | | 1 |
| 15 | 12.3 | 17 | 15 | 23 | 10 | 15.3 | 11 | | 1 | | | 1 |
| 15 | 12.3 | 17 | 15 | 26 | 8 | 12 | 9 | | 1 | | | 1 |
| 15 | 12.3 | 17 | 15 | 27 | 6 | 16.3 | 9 | | 1 | | | 1 |
| 15 | 12.3 | 20 | 15 | 23 | 12 | 14.3 | 9 | | 1 | | | 1 |
| 15 | 12.3 | 20 | 16 | 20 | 8 | 10 | 11 | 1 | | | | 1 |
| 15 | 12.3 | 20 | 16 | 24 | 10 | 11 | 10 | | 1 | | | 1 |
| 15 | 12.3 | 20 | 16 | 24 | 10 | 12 | 9 | | 1 | | | 1 |
| 15 | 12.3 | 21 | 14 | 25 | 11 | 15.3 | 11 | | 1 | | | 1 |
| 15 | 12.3 | 21 | 15 | 24 | 8 | 15.3 | 11 | | 1 | | | 1 |
| 15 | 12.3 | 21 | 15 | 24 | 9 | 15.3 | 10 | | 1 | | | 1 |
| 15 | 12.3 | 21 | 16 | 25 | 8 | 11 | 9 | | 1 | | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=175 | AS N=201 | CN N=122 | Hisp N=123 | Overall N=621 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 15 | 12.3 | 21 | 17 | 24 | 12 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 12.3 | 21 | 17 | 25 | 9 | 11 | 10 | | 1 | | | 1 |
| 15 | 12.3 | 22 | 15 | 23 | 10 | 15.3 | 10 | | 1 | | | 1 |
| 15 | 12.3 | 22 | 15 | 24 | 10 | 15.3 | 11 | | 1 | | | 1 |
| 15 | 12.3 | 22 | 15 | 27 | 6 | 15.3 | 11 | | 1 | | | 1 |
| 15 | 12.3 | 22 | 16 | 22 | 8 | 13.3 | 11 | | 1 | | | 1 |
| 15 | 12.3 | 22 | 16 | 25 | 10 | 15.3 | 10 | | 1 | | | 1 |
| 15 | 12.3 | 23 | 16 | 24 | 10 | 15.3 | 9 | | 1 | | | 1 |
| 15 | 13 | 15 | 15 | 23 | 11 | 12 | 10 | 1 | | | | 1 |
| 15 | 13 | 15 | 16 | 26 | 11 | 14.3 | 11 | | | | 1 | 1 |
| 15 | 13 | 16 | 14 | 24 | 10 | 12 | 11 | | 1 | | | 1 |
| 15 | 13 | 16 | 15 | 27 | 10 | 15.3 | 10 | | 1 | | | 1 |
| 15 | 13 | 16 | 17 | 25 | 10 | 13 | 10 | | 1 | | | 1 |
| 15 | 13 | 20 | 14 | 24 | 10 | 15.3 | 9 | | 1 | | | 1 |
| 15 | 13 | 20 | 16 | 24 | 8 | 14.3 | 11 | | | | 1 | 1 |
| 15 | 13 | 20 | 17 | 26 | 10 | 14.3 | 11 | | | | 1 | 1 |
| 15 | 13 | 21 | 16 | 24 | 11 | 14.3 | 10 | | | | 1 | 1 |
| 15 | 13 | 22 | 16 | 19 | 6 | 15.3 | 11 | | | 1 | | 1 |
| 15 | 13 | 24 | 17 | 18 | 10 | 14.3 | 9 | | | | 1 | 1 |
| 15 | 13.3 | 15 | 16 | 19 | 8 | 12 | 10 | | | | 1 | 1 |
| 15 | 13.3 | 16 | 14 | 25 | 8 | 15.3 | 12 | 1 | | | | 1 |
| 15 | 13.3 | 16 | 17 | 23 | 6 | 15.3 | 11 | | | 1 | | 1 |
| 15 | 13.3 | 16 | 17 | 24 | 8 | 13 | 9 | | 1 | | | 1 |
| 15 | 13.3 | 19 | 13 | 29 | 10 | 14.3 | 11 | 1 | | | | 1 |
| 15 | 13.3 | 20 | 13 | 23 | 11 | 12 | 9 | | | | 1 | 1 |
| 15 | 13.3 | 20 | 15 | 18 | 10 | 12 | 9 | | | | 1 | 1 |
| 15 | 13.3 | 20 | 15 | 23 | 10 | 15.3 | 9 | | 1 | | | 1 |
| 15 | 13.3 | 20 | 15 | 25 | 10 | 15.3 | 10 | | 1 | | | 1 |
| 15 | 13.3 | 21 | 14 | 27 | 10 | 13.3 | 9 | | | | 1 | 1 |
| 15 | 13.3 | 21 | 16 | 24 | 6 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 13.3 | 22 | 14 | 28 | 8 | 15.3 | 11 | | | 1 | | 1 |
| 15 | 13.3 | 22 | 16 | 24 | 11 | 16.3 | 9 | | 1 | | | 1 |
| 15 | 13.3 | 23 | 16 | 24 | 10 | 13.3 | 9 | | | | 1 | 1 |
| 15 | 14.3 | 15 | 15 | 25 | 12 | 12 | 11 | | | 1 | | 1 |
| 16 | 10 | 21 | 13 | 26 | 8 | 12 | 9 | 1 | | | | 1 |
| 16 | 11 | 15 | 16 | 21 | 8 | 13 | 11 | 1 | | | | 1 |
| 16 | 11 | 16 | 16 | 22 | 6 | 15.3 | 10 | | 1 | | | 1 |
| 16 | 11 | 20 | 14 | 26 | 12 | 14.3 | 10 | | | 1 | | 1 |
| 16 | 11 | 20 | 15 | 26 | 6 | 15.3 | 12 | | | | 1 | 1 |
| 16 | 11 | 21 | 14 | 23 | 12 | 13 | 10 | | 1 | | | 1 |
| 16 | 11 | 23 | 15 | 19 | 11 | 14 | 11 | | | 1 | | 1 |
| 16 | 12 | 15 | 14 | 24 | 11 | 15.3 | 10 | | 1 | | | 1 |
| 16 | 12 | 15 | 17 | 18 | 12 | 13 | 10 | | | 1 | | 1 |
| 16 | 12 | 16 | 16 | 23 | 10 | 14.3 | 10 | | 1 | | | 1 |
| 16 | 12 | 16 | 16 | 26 | 8 | 15.3 | 10 | | 1 | | | 1 |
| 16 | 12 | 16 | 17 | 25 | 10 | 15.3 | 10 | | 1 | | | 1 |
| 16 | 12 | 20 | 14 | 26 | 6 | 15.3 | 10 | | | 1 | | 1 |
| 16 | 12 | 20 | 16 | 22 | 8 | 14.3 | 10 | 1 | | | | 1 |
| 16 | 12 | 20 | 16 | 24 | 12 | 15.3 | 10 | 1 | | | | 1 |
| 16 | 12 | 21 | 16 | 24 | 9 | 15.3 | 11 | | | 1 | | 1 |
| 16 | 12 | 21 | 16 | 28 | 12 | 16.3 | 11 | | | 1 | | 1 |
| 16 | 12 | 21 | 17 | 24 | 8 | 12 | 10 | | 1 | | | 1 |
| 16 | 12 | 24 | 16 | 25 | 8 | 14.3 | 9 | | | 1 | | 1 |
| 16 | 12.3 | 15 | 15 | 23 | 10 | 15.3 | 10 | | 1 | | | 1 |
| 16 | 12.3 | 16 | 15 | 24 | 10 | 11 | 11 | | 1 | | | 1 |
| 16 | 12.3 | 16 | 15 | 26 | 8 | 14.3 | 11 | | 1 | | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=175 | AS N=201 | CN N=122 | Hisp N=123 | Overall N=621 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 16 | 12.3 | 16 | 16 | 25 | 10 | 12 | 9 | | 1 | | | 1 |
| 16 | 12.3 | 19 | 14 | 25 | 11 | 14.3 | 10 | | 1 | | | 1 |
| 16 | 12.3 | 20 | 14 | 25 | 8 | 12 | 10 | | 1 | | | 1 |
| 16 | 12.3 | 20 | 17 | 26 | 10 | 12 | 9 | | | | 1 | 1 |
| 16 | 12.3 | 21 | 15 | 24 | 10 | 15.3 | 12 | | 1 | | | 1 |
| 16 | 12.3 | 22 | 14 | 25 | 10 | 12 | 10 | 1 | | | | 1 |
| 16 | 12.3 | 22 | 17 | 25 | 11 | 11 | 11 | | 1 | | | 1 |
| 16 | 13 | 14 | 15 | 26 | 9 | 11 | 11 | | | | 1 | 1 |
| 16 | 13 | 20 | 14 | 23 | 6 | 15.3 | 9 | | | | 1 | 1 |
| 16 | 13 | 20 | 17 | 18 | 8 | 14.3 | 10 | | | | 1 | 1 |
| 16 | 13.3 | 21 | 15 | 22 | 10 | 15.3 | 10 | | 1 | | | 1 |
| 16 | 13.3 | 21 | 15 | 28 | 11 | 12 | 11 | | | 1 | | 1 |
| 17 | 11 | 20 | 14 | 28 | 6 | 13 | 11 | 1 | | | | 1 |
| 17 | 12 | 16 | 16 | 25 | 10 | 15.3 | 10 | | 1 | | | 1 |
| 17 | 12 | 19 | 17 | 27 | 6 | 12 | 10 | | 1 | | | 1 |
| 17 | 12 | 20 | 14 | 25 | 10 | 15.3 | 10 | | | | 1 | 1 |
| 17 | 12.3 | 19 | 14 | 24 | 9 | 12 | 10 | | 1 | | | 1 |
| 18 | 12 | 15 | 16 | 18 | 8 | 9 | 11 | 1 | | | | 1 |
| 18 | 12 | 21 | 12 | 24 | 11 | 14.3 | 9 | | | 1 | | 1 |
| | | | | | | | Total number of haplotypes: | 175 | 201 | 122 | 123 | 621 |
| | | | | | | | Total number of unique haplotypes: | 175 | 201 | 122 | 123 | 621 |
| | | | | | | | Count of most common haplotype: | 1 | 1 | 1 | 1 | 1 |

**Table B3. Haplotypes observed within sample set A for proposed linkage group 4.** AA: African American, AS: U.S. Asian, CN: U.S. Caucasian, Hisp: U.S. Hispanic, N: number of samples.

| GATA31E08 | DXS10147 | DXS7423 | AA N=175 | AS N=201 | CN N=122 | Hisp N=123 | Overall N=621 |
|---|---|---|---|---|---|---|---|
| 8 | 7 | 14 | 1 | | | | 1 |
| 8 | 8 | 13 | 1 | | | | 1 |
| 8 | 8 | 14 | 2 | | | | 2 |
| 9 | 6 | 13 | | | 1 | | 1 |
| 9 | 6 | 14 | 2 | 2 | 2 | 1 | 7 |
| 9 | 6 | 15 | 3 | 1 | 2 | 5 | 11 |
| 9 | 6 | 16 | | | 2 | 1 | 3 |
| 9 | 6 | 17 | 1 | | | | 1 |
| 9 | 7 | 12 | 1 | | | | 1 |
| 9 | 7 | 14 | 5 | 2 | | | 7 |
| 9 | 7 | 15 | 2 | 4 | 1 | 1 | 8 |
| 9 | 7 | 16 | | 1 | | | 1 |
| 9 | 8 | 14 | 6 | 2 | 3 | 3 | 14 |
| 9 | 8 | 15 | 4 | 10 | | 2 | 16 |
| 9 | 8 | 16 | | 1 | | 1 | 2 |
| 9 | 8 | 17 | 1 | | | 2 | 3 |
| 9 | 9 | 13 | | 1 | | | 1 |
| 9 | 9 | 14 | 1 | | 3 | 1 | 5 |
| 9 | 9 | 15 | 1 | | 3 | 1 | 5 |
| 9 | 9 | 16 | | | 1 | | 1 |
| 9 | 9 | 17 | | | 1 | | 1 |
| 9 | 11 | 13 | | | 1 | | 1 |
| 10 | 6 | 15 | | | 2 | 1 | 3 |

| GATA31E08 | DXS10147 | DXS7423 | AA N=175 | AS N=201 | CN N=122 | Hisp N=123 | Overall N=621 |
|---|---|---|---|---|---|---|---|
| 10 | 6 | 16 | 1 | | 1 | 1 | 3 |
| 10 | 7 | 14 | 3 | 1 | | 1 | 5 |
| 10 | 7 | 15 | 6 | 1 | | | 7 |
| 10 | 7 | 16 | 1 | 1 | | 1 | 3 |
| 10 | 8 | 12 | 1 | | | | 1 |
| 10 | 8 | 13 | 2 | | | | 2 |
| 10 | 8 | 14 | 3 | | | 2 | 5 |
| 10 | 8 | 15 | 4 | 2 | | 1 | 7 |
| 10 | 8 | 16 | 1 | | | | 1 |
| 10 | 8 | 17 | | | | 1 | 1 |
| 10 | 9 | 13 | 2 | | | 1 | 3 |
| 10 | 9 | 14 | 2 | | | | 2 |
| 10 | 9 | 15 | 1 | 1 | | | 2 |
| 10 | 9 | 16 | | | | 1 | 1 |
| 10 | 10 | 15 | 1 | | | | 1 |
| 11 | 6 | 14 | | 3 | | | 3 |
| 11 | 6 | 15 | | 5 | 6 | 9 | 20 |
| 11 | 6 | 16 | 1 | 1 | 1 | 1 | 4 |
| 11 | 6 | 17 | | | 3 | | 3 |
| 11 | 7 | 14 | | 6 | 1 | | 7 |
| 11 | 7 | 15 | | 2 | | | 2 |
| 11 | 7 | 16 | | | | 1 | 1 |
| 11 | 7 | 17 | | | | 1 | 1 |
| 11 | 8 | 13 | | 1 | | | 1 |
| 11 | 8 | 14 | 6 | 5 | 5 | 3 | 19 |
| 11 | 8 | 15 | 1 | 9 | 3 | 5 | 18 |
| 11 | 8 | 16 | | | 1 | 1 | 2 |
| 11 | 8 | 17 | | | | 1 | 1 |
| 11 | 9 | 13 | | | 2 | 1 | 3 |
| 11 | 9 | 14 | | 3 | 2 | | 5 |
| 11 | 9 | 15 | | 2 | 4 | 2 | 8 |
| 11 | 9 | 16 | | | 1 | | 1 |
| 11 | 10 | 15 | | | | 1 | 1 |
| 12 | 6 | 13 | | | 1 | | 1 |
| 12 | 6 | 14 | 3 | 6 | | | 9 |
| 12 | 6 | 15 | | 9 | 3 | 9 | 21 |
| 12 | 6 | 16 | 1 | | 1 | 1 | 3 |
| 12 | 6 | 17 | | | | 1 | 1 |
| 12 | 7 | 13 | | | | 1 | 1 |
| 12 | 7 | 14 | 6 | 8 | | | 14 |
| 12 | 7 | 15 | 5 | 1 | | 2 | 8 |
| 12 | 7 | 16 | 1 | | | 1 | 2 |
| 12 | 8 | 13 | | | 1 | | 1 |
| 12 | 8 | 14 | 6 | 7 | 3 | 7 | 23 |
| 12 | 8 | 15 | 4 | 19 | 1 | 8 | 32 |
| 12 | 8 | 16 | | 1 | | 1 | 2 |
| 12 | 8 | 17 | | | 1 | 3 | 4 |
| 12 | 9 | 13 | 2 | | 1 | | 3 |
| 12 | 9 | 14 | 5 | 1 | 3 | 3 | 12 |
| 12 | 9 | 15 | 2 | | 7 | 1 | 10 |
| 12 | 9 | 16 | 3 | 3 | | | 6 |
| 12 | 10 | 14 | 1 | | | | 1 |
| 12 | 10 | 16 | | | | 1 | 1 |
| 13 | 6 | 13 | | | | 1 | 1 |
| 13 | 6 | 14 | 5 | 7 | 3 | 1 | 16 |
| 13 | 6 | 15 | 1 | 12 | 3 | 4 | 20 |
| 13 | 6 | 16 | | | 1 | 1 | 2 |
| 13 | 6 | 17 | | | 1 | 1 | 2 |
| 13 | 7 | 14 | 11 | 4 | | | 15 |
| 13 | 7 | 15 | 2 | 5 | | | 7 |

| GATA31E08 | DXS10147 | DXS7423 | AA N=175 | AS N=201 | CN N=122 | Hisp N=123 | Overall N=621 |
|---|---|---|---|---|---|---|---|
| 13 | 7 | 16 | 1 | | 1 | | 2 |
| 13 | 7 | 17 | | | 1 | 1 | 2 |
| 13 | 8 | 13 | 2 | | | 1 | 3 |
| 13 | 8 | 14 | 12 | 7 | 3 | 1 | 23 |
| 13 | 8 | 15 | 7 | 18 | 5 | 10 | 40 |
| 13 | 8 | 16 | 1 | 2 | 2 | 1 | 6 |
| 13 | 9 | 13 | 2 | | 2 | | 4 |
| 13 | 9 | 14 | 3 | 3 | 4 | 2 | 12 |
| 13 | 9 | 15 | 2 | | 3 | 1 | 6 |
| 13 | 9 | 17 | | | | 1 | 1 |
| 13 | 10 | 13 | | | 1 | | 1 |
| 13 | 10 | 14 | 1 | | | | 1 |
| 13 | 10 | 15 | 2 | | | | 2 |
| 13 | 11 | 14 | 1 | | | | 1 |
| 13 | 11 | 15 | | | 1 | | 1 |
| 14 | 5 | 14 | 1 | | | | 1 |
| 14 | 6 | 13 | 1 | | | | 1 |
| 14 | 6 | 14 | 3 | | 1 | | 4 |
| 14 | 6 | 15 | | 4 | 1 | 1 | 6 |
| 14 | 7 | 14 | 1 | 1 | | | 2 |
| 14 | 7 | 15 | 3 | 2 | | | 5 |
| 14 | 7 | 16 | 1 | | | | 1 |
| 14 | 7 | 17 | | | 1 | | 1 |
| 14 | 8 | 14 | 1 | 1 | 1 | | 3 |
| 14 | 8 | 15 | | 7 | 3 | 1 | 11 |
| 14 | 9 | 13 | | | 4 | | 4 |
| 14 | 9 | 14 | | | 2 | 1 | 3 |
| 14 | 9 | 15 | 1 | | | | 1 |
| 14 | 9 | 16 | 1 | | 2 | | 3 |
| 14 | 10 | 14 | | | 1 | | 1 |
| 14 | 10 | 15 | 1 | | | | 1 |
| 15 | 6 | 15 | | 1 | | 1 | 2 |
| 15 | 8 | 14 | 1 | | | | 1 |
| 15 | 8 | 15 | | 2 | 1 | | 3 |
| 15 | 8 | 16 | | 1 | | | 1 |
| 15 | 9 | 15 | | 1 | | | 1 |
| 16 | 6 | 14 | | 1 | | | 1 |
| 16 | 8 | 16 | 1 | | | | 1 |
| | | Total number of haplotypes: | 69 | 51 | 58 | 60 | 121 |
| | | Total number of unique haplotypes: | 33 | 19 | 28 | 42 | 45 |
| | | Coutn of most common haplotype: | 12 | 19 | 7 | 10 | 40 |

**Table B4. Haplotypes observed within sample set B for proposed linkage group 1.** AA: African American, AS: U.S. Asian, CN: U.S. Caucasian, Hisp: U.S. Hispanic, N: number of samples.

| DXS8378 | DXS9902 | DXS6795 | AA N=258 | AS N=3 | CN N=260 | Hisp N=138 | Overall N=659 |
|---|---|---|---|---|---|---|---|
| 9 | 10 | 11 | | | 1 | 1 | 2 |
| 9 | 10 | 13 | | 1 | | | 1 |
| 9 | 11 | 13 | | | 1 | 1 | 2 |
| 9 | 12 | 11 | | | | 1 | 1 |
| 9 | 12 | 13 | | | | 1 | 1 |
| 10 | 8 | 10 | 1 | | | | 1 |
| 10 | 8 | 14 | 1 | | | | 1 |

| DXS8378 | DXS9902 | DXS6795 | AA N=258 | AS N=3 | CN N=260 | Hisp N=138 | Overall N=659 |
|---|---|---|---|---|---|---|---|
| 10 | 8 | 15 | 1 | | | | 1 |
| 10 | 9 | 10 | 3 | | | | 3 |
| 10 | 9 | 11 | | | 2 | | 2 |
| 10 | 9 | 12 | | | 1 | | 1 |
| 10 | 9 | 13 | 1 | | | | 1 |
| 10 | 10 | 9 | 3 | | 9 | 5 | 17 |
| 10 | 10 | 10 | 10 | | 1 | 3 | 14 |
| 10 | 10 | 11 | 3 | | 10 | 4 | 17 |
| 10 | 10 | 12 | 1 | | 3 | 4 | 8 |
| 10 | 10 | 13 | 4 | | 4 | 9 | 17 |
| 10 | 10 | 15 | 3 | | | | 3 |
| 10 | 10 | 17 | 2 | | | | 2 |
| 10 | 10.1 | 11 | | | | 2 | 2 |
| 10 | 11 | 9 | 2 | | 8 | 3 | 13 |
| 10 | 11 | 10 | 13 | | | 1 | 14 |
| 10 | 11 | 11 | 1 | 1 | 16 | 5 | 23 |
| 10 | 11 | 12 | 2 | | | 3 | 5 |
| 10 | 11 | 13 | | | 5 | 5 | 10 |
| 10 | 11 | 15 | 6 | | | | 6 |
| 10 | 11.1 | 11 | 1 | | 2 | | 3 |
| 10 | 12 | 9 | | | 10 | 2 | 12 |
| 10 | 12 | 10 | 6 | | 1 | 1 | 8 |
| 10 | 12 | 11 | 1 | | 9 | 5 | 15 |
| 10 | 12 | 12 | 1 | | 2 | 3 | 6 |
| 10 | 12 | 13 | 2 | | 3 | 1 | 6 |
| 10 | 12 | 14 | | | | 1 | 1 |
| 10 | 12 | 15 | 3 | | | | 3 |
| 10 | 12.1 | 9 | | | 1 | | 1 |
| 10 | 13 | 11 | 1 | | | | 1 |
| 10 | 13 | 12 | | | 1 | | 1 |
| 11 | 8 | 9 | 1 | | | | 1 |
| 11 | 8 | 11 | 1 | | | | 1 |
| 11 | 8 | 15 | 2 | | | | 2 |
| 11 | 9 | 9 | 2 | | 1 | 1 | 4 |
| 11 | 9 | 10 | 4 | | | | 4 |
| 11 | 9 | 11 | 1 | | 1 | 1 | 3 |
| 11 | 9 | 15 | 1 | | | | 1 |
| 11 | 10 | 9 | 3 | | 10 | 3 | 16 |
| 11 | 10 | 10 | 8 | | | 2 | 10 |
| 11 | 10 | 11 | 5 | | 10 | 5 | 20 |
| 11 | 10 | 12 | 3 | | 1 | 4 | 8 |
| 11 | 10 | 13 | 1 | | 6 | 6 | 13 |
| 11 | 10 | 14 | 2 | | | | 2 |
| 11 | 10 | 15 | 8 | | 1 | | 9 |
| 11 | 11 | 9 | 8 | | 12 | 3 | 23 |
| 11 | 11 | 10 | 11 | | | 1 | 12 |
| 11 | 11 | 11 | 3 | | 12 | 3 | 18 |
| 11 | 11 | 12 | 2 | | 1 | 2 | 5 |
| 11 | 11 | 13 | 3 | | 7 | 2 | 12 |
| 11 | 11 | 14 | 1 | | | | 1 |
| 11 | 11 | 15 | 10 | | | | 10 |
| 11 | 11 | 16 | 1 | | | | 1 |
| 11 | 11 | 18 | 1 | | | | 1 |
| 11 | 11.1 | 9 | | | 2 | | 2 |
| 11 | 11.1 | 10 | 1 | | | | 1 |

| DXS8378 | DXS9902 | DXS6795 | AA N=258 | AS N=3 | CN N=260 | Hisp N=138 | Overall N=659 |
|---|---|---|---|---|---|---|---|
| 11 | 11.1 | 12 | | | 1 | | 1 |
| 11 | 11.1 | 13 | | | 2 | 1 | 3 |
| 11 | 12 | 9 | 2 | | 6 | | 8 |
| 11 | 12 | 10 | 6 | | 1 | 1 | 8 |
| 11 | 12 | 11 | 4 | | 10 | 2 | 16 |
| 11 | 12 | 12 | 4 | | | | 4 |
| 11 | 12 | 13 | 1 | | 5 | 3 | 9 |
| 11 | 12 | 15 | 2 | | | 1 | 3 |
| 11 | 13 | 9 | | | | 1 | 1 |
| 11 | 13 | 11 | | | 1 | | 1 |
| 12 | 8 | 9 | 1 | | | | 1 |
| 12 | 8 | 10 | 2 | | | | 2 |
| 12 | 8 | 15 | 1 | | | | 1 |
| 12 | 8 | 17 | 1 | | | | 1 |
| 12 | 9 | 9 | | | 1 | | 1 |
| 12 | 9 | 10 | | | | 1 | 1 |
| 12 | 9 | 11 | 1 | | 1 | | 2 |
| 12 | 9 | 14 | 1 | | | | 1 |
| 12 | 10 | 9 | 4 | | 11 | | 15 |
| 12 | 10 | 10 | 10 | | | | 10 |
| 12 | 10 | 11 | 5 | | 15 | 3 | 23 |
| 12 | 10 | 12 | 2 | | | 1 | 3 |
| 12 | 10 | 13 | | | 4 | 2 | 6 |
| 12 | 10 | 14 | | | | 1 | 1 |
| 12 | 10 | 15 | 1 | | 1 | | 2 |
| 12 | 11 | 9 | 4 | | 6 | 3 | 13 |
| 12 | 11 | 10 | 6 | | | | 6 |
| 12 | 11 | 11 | 4 | | 8 | 6 | 18 |
| 12 | 11 | 12 | 3 | | 2 | | 5 |
| 12 | 11 | 13 | | | 6 | 3 | 9 |
| 12 | 11 | 14 | 1 | | | | 1 |
| 12 | 11 | 15 | 5 | | | | 5 |
| 12 | 11.1 | 11 | | | 1 | 1 | 2 |
| 12 | 11.1 | 12 | | | | 1 | 1 |
| 12 | 11.1 | 13 | | | 1 | | 1 |
| 12 | 12 | 9 | | | 3 | 4 | 7 |
| 12 | 12 | 10 | 7 | | 1 | | 8 |
| 12 | 12 | 11 | 8 | | 6 | 4 | 18 |
| 12 | 12 | 12 | 3 | | | | 3 |
| 12 | 12 | 13 | 2 | | 1 | 1 | 4 |
| 12 | 12 | 14 | 1 | | | 1 | 2 |
| 12 | 12 | 15 | 1 | | | | 1 |
| 13 | 7 | 11 | | | 1 | | 1 |
| 13 | 8 | 10 | 1 | | | | 1 |
| 13 | 9 | 11 | | | 1 | | 1 |
| 13 | 9 | 12 | | | 1 | | 1 |
| 13 | 9 | 13 | | | 1 | | 1 |
| 13 | 10 | 10 | 2 | | | | 2 |
| 13 | 10 | 11 | | | 2 | | 2 |
| 13 | 10 | 12 | | | | 1 | 1 |
| 13 | 10 | 13 | 1 | 1 | | 1 | 3 |
| 13 | 10 | 15 | 1 | | | | 1 |
| 13 | 11 | 9 | 1 | | 2 | | 3 |
| 13 | 11 | 10 | 2 | | | | 2 |
| 13 | 11 | 13 | | | 1 | | 1 |

| DXS8378 | DXS9902 | DXS6795 | AA N=258 | AS N=3 | CN N=260 | Hisp N=138 | Overall N=659 |
|---|---|---|---|---|---|---|---|
| 13 | 12 | 10 | 1 | | | | 1 |
| 13 | 12 | 11 | | | 2 | | 2 |
| 13 | 13 | 9 | | | 1 | | 1 |
| 13 | 13 | 15 | 1 | | | | 1 |
| 14 | 12 | 9 | | | | 1 | 1 |
| Total number of haplotypes: | | | 85 | 3 | 64 | 56 | 122 |
| Total number of unique haplotypes: | | | 35 | 3 | 28 | 25 | 45 |
| Count of most common haplotype: | | | 13 | 1 | 16 | 9 | 23 |

**Table B5. Haplotypes observed within sample set B for proposed linkage group 2.** AA: African American, AS: U.S. Asian, CN: U.S. Caucasian, Hisp: U.S. Hispanic, N: number of samples.

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=257 | AS N=3 | CN N=259 | Hisp N=138 | Overall N=657 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 11 | 11 | 15 | 15 | 27 | 8 | 14.3 | 11 | 1 | | | | 1 |
| 11 | 12 | 15 | 14 | 26 | 6 | 15.3 | 11 | 1 | | | | 1 |
| 11 | 12.3 | 15 | 13 | 19 | 6 | 15.3 | 11 | 1 | | | | 1 |
| 11 | 13 | 20 | 13 | 25 | 12 | 13 | 10 | | | 1 | | 1 |
| 11 | 14.3 | 20 | 17 | 18 | 12 | 15.3 | 9 | | | 1 | | 1 |
| 12 | 10 | 19 | 16 | 27 | 9 | 13 | 10 | 1 | | | | 1 |
| 12 | 10 | 20 | 12 | 24 | 10 | 12 | 11 | 1 | | | | 1 |
| 12 | 10 | 20 | 16 | 26 | 10 | 12 | 11 | 1 | | | | 1 |
| 12 | 10 | 20 | 17 | 18 | 11 | 13.3 | 11 | 1 | | | | 1 |
| 12 | 10 | 21 | 13 | 26 | 9 | 13 | 11 | 1 | | | | 1 |
| 12 | 10 | 21 | 15 | 29 | 9 | 15.3 | 9 | | | 1 | | 1 |
| 12 | 11 | 15 | 11 | 21 | 9 | 15.3 | 11 | 1 | | | | 1 |
| 12 | 11 | 15 | 16 | 24 | 12 | 14.3 | 11 | | | 1 | | 1 |
| 12 | 11 | 16 | 14 | 21 | 6 | 11 | 11 | 1 | | | | 1 |
| 12 | 11 | 16 | 15 | 27 | 8 | 14.3 | 11 | 1 | | | | 1 |
| 12 | 11 | 19 | 14 | 22 | 6 | 10 | 9 | 1 | | | | 1 |
| 12 | 11 | 19 | 15 | 26 | 10 | 13.3 | 11 | | | | 1 | 1 |
| 12 | 11 | 20 | 10 | 28 | 10 | 15.3 | 12 | 1 | | | | 1 |
| 12 | 11 | 20 | 14 | 29 | 8 | 16.3 | 12 | | | 1 | | 1 |
| 12 | 11 | 20 | 15 | 23 | 8 | 14.3 | 9 | | | 1 | | 1 |
| 12 | 11 | 20 | 15 | 26 | 10 | 15.3 | 9 | | | | 1 | 1 |
| 12 | 11 | 20 | 16 | 25 | 8 | 13 | 9 | 1 | | | | 1 |
| 12 | 11 | 21 | 13 | 29 | 8 | 13 | 9 | 1 | | | | 1 |
| 12 | 11 | 21 | 14 | 27 | 10 | 13.3 | 9 | | | 1 | | 1 |
| 12 | 11 | 21 | 16 | 24 | 6 | 14 | 11 | 1 | | | | 1 |
| 12 | 11 | 22 | 12 | 27 | 8 | 16.3 | 9 | | | 1 | | 1 |
| 12 | 11 | 22 | 13 | 26 | 9 | 15.3 | 11 | | | | 1 | 1 |
| 12 | 11 | 22 | 15 | 18 | 10 | 14.3 | 11 | | | 1 | | 1 |
| 12 | 12 | 15 | 13 | 26 | 8 | 15.3 | 10 | 1 | | | | 1 |
| 12 | 12 | 15 | 16 | 25 | 10 | 15.3 | 11 | | | 1 | | 1 |
| 12 | 12 | 16 | 14 | 19 | 7 | 14.3 | 10 | 1 | | | | 1 |
| 12 | 12 | 18 | 13 | 26 | 11 | 15.3 | 10 | | | | 1 | 1 |
| 12 | 12 | 19 | 16 | 27 | 9 | 14.3 | 11 | 1 | | | | 1 |
| 12 | 12 | 20 | 13 | 21 | 9 | 12 | 10 | 1 | | | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=257 | AS N=3 | CN N=259 | Hisp N=138 | Overall N=657 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 12 | 12 | 20 | 14 | 24 | 8 | 12 | 9 | | | 1 | | 1 |
| 12 | 12 | 20 | 14 | 26 | 8 | 14.3 | 9 | | | 1 | | 1 |
| 12 | 12 | 20 | 17 | 18 | 11 | 15.3 | 9 | | | 1 | | 1 |
| 12 | 12 | 21 | 13 | 27 | 10 | 16.3 | 9 | | | | 1 | 1 |
| 12 | 12 | 21 | 14 | 23 | 6 | 15.3 | 11 | | | | 1 | 1 |
| 12 | 12 | 21 | 14 | 24 | 10 | 14.3 | 9 | | | 1 | | 1 |
| 12 | 12 | 21 | 14 | 26 | 12 | 12 | 13 | | | | 1 | 1 |
| 12 | 12 | 21 | 16 | 24 | 10 | 15.3 | 11 | | | 1 | | 1 |
| 12 | 12 | 21 | 17 | 21 | 6 | 14.3 | 10 | 1 | | | | 1 |
| 12 | 12.3 | 21 | 14 | 24 | 6 | 15.3 | 11 | | | 1 | | 1 |
| 12 | 12.3 | 21 | 14 | 25 | 11 | 12 | 11 | | | 1 | | 1 |
| 12 | 12.3 | 21 | 15 | 27 | 10 | 15.3 | 11 | | | 1 | | 1 |
| 12 | 12.3 | 21 | 16 | 24 | 9 | 11 | 10 | | | | 1 | 1 |
| 12 | 13 | 15 | 14 | 26 | 8 | 12 | 10 | 1 | | | | 1 |
| 12 | 13 | 15 | 14 | 26 | 11 | 11 | 10 | 1 | | | | 1 |
| 12 | 13 | 20 | 10 | 21 | 8 | 15.3 | 11 | 1 | | | | 1 |
| 12 | 13 | 20 | 15 | 19 | 12 | 12 | 11 | | | 1 | | 1 |
| 12 | 13 | 20 | 16 | 24 | 12 | 16.3 | 10 | | | 1 | | 1 |
| 12 | 13 | 22 | 13 | 26 | 12 | 15.3 | 10 | | | 1 | | 1 |
| 12 | 13.3 | 15 | 17 | 25 | 10 | 11 | 9 | | 1 | | | 1 |
| 12 | 13.3 | 20 | 13 | 26 | 11 | 14.3 | 9 | | | 1 | | 1 |
| 12 | 13.3 | 20 | 15 | 26 | 10 | 15.3 | 9 | | | 1 | | 1 |
| 12 | 13.3 | 20 | 16 | 25 | 11 | 15.3 | 11 | | | 1 | | 1 |
| 12 | 13.3 | 20 | 16 | 30 | 10 | 11 | 9 | | | | 1 | 1 |
| 12 | 13.3 | 22 | 16 | 25 | 6 | 15.3 | 11 | 1 | | | | 1 |
| 12 | 13.3 | 22 | 17 | 18 | 6 | 15.3 | 9 | | | | 1 | 1 |
| 12 | 14 | 20 | 13 | 22 | 9 | 12 | 8 | 1 | | | | 1 |
| 13 | 8 | 20 | 13 | 19 | 7 | 13 | 10 | 1 | | | | 1 |
| 13 | 10 | 16 | 11 | 21 | 9 | 15.3 | 12 | 1 | | | | 1 |
| 13 | 10 | 20 | 11 | 26 | 11 | 15.3 | 9 | | | | 1 | 1 |
| 13 | 10 | 20 | 12 | 19 | 10 | 15.3 | 8 | 1 | | | | 1 |
| 13 | 10 | 20 | 14 | 27 | 7 | 12 | 11 | 1 | | | | 1 |
| 13 | 10 | 20 | 16 | 22 | 6 | 15.3 | 10 | | | 1 | | 1 |
| 13 | 10 | 20 | 19 | 15 | 10 | 15.3 | 9 | | | 1 | | 1 |
| 13 | 10 | 21 | 15 | 24 | 8 | 15.3 | 9 | | | 1 | | 1 |
| 13 | 10 | 22 | 13 | 24 | 10 | 17.3 | 9 | | | 1 | | 1 |
| 13 | 10 | 22 | 13 | 27 | 6 | 13 | 11 | | | | 1 | 1 |
| 13 | 10 | 23 | 16 | 29 | 10 | 12 | 12 | | | 1 | | 1 |
| 13 | 11 | 15 | 11 | 21 | 6 | 14.3 | 10 | 1 | | | | 1 |
| 13 | 11 | 15 | 12 | 25 | 12 | 15.3 | 11 | 1 | | | | 1 |
| 13 | 11 | 15 | 13 | 26 | 9 | 13 | 11 | 1 | | | | 1 |
| 13 | 11 | 15 | 14 | 19 | 6 | 15.3 | 11 | 1 | | | | 1 |
| 13 | 11 | 15 | 14 | 22 | 9 | 14.3 | 10 | 1 | | | | 1 |
| 13 | 11 | 15 | 15 | 26 | 11 | 14.3 | 10 | | | | 1 | 1 |
| 13 | 11 | 15 | 15 | 27 | 9 | 13 | 11 | 1 | | | | 1 |
| 13 | 11 | 15 | 16 | 20 | 8 | 12 | 9 | 1 | | | | 1 |
| 13 | 11 | 16 | 14 | 28 | 9 | 13 | 8 | 1 | | | | 1 |
| 13 | 11 | 16 | 16 | 23 | 7 | 13 | 10 | 1 | | | | 1 |
| 13 | 11 | 19 | 16 | 25 | 10 | 11 | 11 | | | 1 | | 1 |
| 13 | 11 | 20 | 12 | 25 | 11 | 12 | 11 | | | | 1 | 1 |
| 13 | 11 | 20 | 13 | 27 | 8 | 16.3 | 11 | 1 | | | | 1 |
| 13 | 11 | 20 | 14 | 25 | 11 | 14.3 | 9 | | | 1 | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=257 | AS N=3 | CN N=259 | Hisp N=138 | Overall N=657 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 13 | 11 | 20 | 15 | 25 | 6 | 16.3 | 10 | | | 1 | | 1 |
| 13 | 11 | 20 | 16 | 18 | 8 | 13.3 | 10 | | | 1 | | 1 |
| 13 | 11 | 20 | 16 | 19 | 6 | 15.3 | 10 | | | | 1 | 1 |
| 13 | 11 | 20 | 16 | 28 | 12 | 12 | 10 | | | | 1 | 1 |
| 13 | 11 | 21 | 11 | 18 | 7 | 14 | 12 | 1 | | | | 1 |
| 13 | 11 | 21 | 13 | 19 | 6 | 13 | 10 | 1 | | | | 1 |
| 13 | 11 | 21 | 14 | 19 | 9 | 13 | 9 | 1 | | | | 1 |
| 13 | 11 | 21 | 14 | 24 | 10 | 15.3 | 10 | | | 1 | | 1 |
| 13 | 11 | 21 | 15 | 25 | 6 | 15.3 | 10 | | | 1 | | 1 |
| 13 | 11 | 21 | 15 | 25 | 10 | 11 | 9 | | | 1 | | 1 |
| 13 | 11 | 21 | 16 | 24 | 11 | 14.3 | 10 | | | | 1 | 1 |
| 13 | 11 | 21 | 16 | 27 | 6 | 12 | 11 | 1 | | | | 1 |
| 13 | 11 | 21 | 16 | 28 | 6 | 12 | 9 | 1 | | | | 1 |
| 13 | 11 | 21 | 17 | 23 | 10 | 14.3 | 10 | | | | 1 | 1 |
| 13 | 11 | 22 | 12 | 21 | 11 | 16.3 | 10 | 1 | | | | 1 |
| 13 | 11 | 22 | 13 | 29 | 9 | 12 | 12 | 1 | | | | 1 |
| 13 | 11 | 22 | 14 | 29 | 11 | 16.3 | 11 | 1 | | | | 1 |
| 13 | 11 | 22 | 14 | 31 | 9 | 14 | 10 | | | 1 | | 1 |
| 13 | 11 | 22 | 15 | 19 | 7 | 14 | 10 | 1 | | | | 1 |
| 13 | 11 | 22 | 15 | 20 | 8 | 12 | 11 | 1 | | | | 1 |
| 13 | 11 | 22 | 15 | 27 | 8 | 14.3 | 9 | | | | 1 | 1 |
| 13 | 11 | 23 | 14 | 27 | 9 | 13 | 10 | 1 | | | | 1 |
| 13 | 11 | 23 | 15 | 25 | 12 | 15.3 | 9 | | | 1 | | 1 |
| 13 | 11 | 23 | 16 | 24 | 8 | 15.3 | 9 | | | 1 | | 1 |
| 13 | 11 | 24 | 12 | 27 | 12 | 14.3 | 11 | | | 1 | | 1 |
| 13 | 11.3 | 16 | 13 | 27 | 12 | 12 | 10 | | | | 1 | 1 |
| 13 | 11.3 | 20 | 14 | 26 | 8 | 13 | 10 | 1 | | | | 1 |
| 13 | 11.3 | 20 | 14 | 26 | 8 | 14.3 | 10 | | | | 1 | 1 |
| 13 | 11.3 | 20 | 16 | 27 | 10 | 14.3 | 10 | | | 1 | | 1 |
| 13 | 12 | 15 | 14 | 24 | 9 | 11 | 10 | | | 1 | | 1 |
| 13 | 12 | 15 | 16 | 21 | 11 | 14.3 | 10 | 1 | | | | 1 |
| 13 | 12 | 15 | 16 | 24 | 6 | 15.3 | 11 | | | 1 | | 1 |
| 13 | 12 | 15 | 16 | 25 | 8 | 14.3 | 8 | | | 1 | | 1 |
| 13 | 12 | 16 | 15 | 18 | 9 | 14 | 11 | 1 | | | | 1 |
| 13 | 12 | 16 | 15 | 28 | 10 | 16.3 | 9 | | | 1 | | 1 |
| 13 | 12 | 16 | 16 | 25 | 10 | 16.3 | 11 | | | 1 | | 1 |
| 13 | 12 | 19 | 14 | 27 | 11 | 12 | 10 | 1 | | | | 1 |
| 13 | 12 | 20 | 12 | 21 | 6 | 12 | 10 | 1 | | | | 1 |
| 13 | 12 | 20 | 13 | 20 | 6 | 12 | 11 | 1 | | | | 1 |
| 13 | 12 | 20 | 14 | 24 | 10 | 15.3 | 9 | | | 1 | | 1 |
| 13 | 12 | 20 | 14 | 27 | 10 | 15.3 | 11 | | | 1 | | 1 |
| 13 | 12 | 20 | 15 | 24 | 10 | 15.3 | 9 | | | 1 | | 1 |
| 13 | 12 | 20 | 16 | 15 | 8 | 15.3 | 9 | | | 1 | | 1 |
| 13 | 12 | 20 | 16 | 18 | 12 | 13.3 | 9 | | | 1 | | 1 |
| 13 | 12 | 20 | 16 | 19 | 9 | 11 | 9 | | | 1 | | 1 |
| 13 | 12 | 20 | 16 | 21 | 6 | 15.3 | 12 | | | 1 | | 1 |
| 13 | 12 | 20 | 16 | 21 | 8 | 14 | 11 | 1 | | | | 1 |
| 13 | 12 | 20 | 16 | 25 | 11 | 15.3 | 11 | | | 1 | | 1 |
| 13 | 12 | 20 | 16 | 27 | 10 | 14.3 | 11 | | | 1 | | 1 |
| 13 | 12 | 21 | 13 | 26 | 10 | 13 | 11 | | | 1 | | 1 |
| 13 | 12 | 21 | 13 | 26 | 12 | 11 | 9 | 1 | | | | 1 |
| 13 | 12 | 21 | 14 | 25 | 8 | 15.3 | 9 | 1 | | | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=257 | AS N=3 | CN N=259 | Hisp N=138 | Overall N=657 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 13 | 12 | 21 | 14 | 26 | 8 | 15.3 | 10 | | | | 1 | 1 |
| 13 | 12 | 21 | 14 | 27 | 10 | 16.3 | 10 | | | 1 | | 1 |
| 13 | 12 | 21 | 14 | 28 | 6 | 12 | 10 | 1 | | | | 1 |
| 13 | 12 | 21 | 15 | 24 | 6 | 14.3 | 10 | | | 2 | | 2 |
| 13 | 12 | 21 | 15 | 26 | 10 | 14.3 | 10 | 1 | | | | 1 |
| 13 | 12 | 21 | 15 | 26 | 11 | 15.3 | 11 | | | | 1 | 1 |
| 13 | 12 | 21 | 15 | 28 | 12 | 16.3 | 10 | 1 | | | | 1 |
| 13 | 12 | 21 | 16 | 19 | 10 | 14.3 | 11 | | | 1 | | 1 |
| 13 | 12 | 21 | 16 | 22 | 11 | 15.3 | 9 | | | 1 | | 1 |
| 13 | 12 | 21 | 17 | 18 | 10 | 15.3 | 10 | | | | 1 | 1 |
| 13 | 12 | 21 | 17 | 19 | 10 | 15.3 | 10 | | | 1 | | 1 |
| 13 | 12 | 21 | 17 | 24 | 11 | 15.3 | 9 | | | | 1 | 1 |
| 13 | 12 | 21 | 17 | 28 | 12 | 14.3 | 10 | | | 1 | | 1 |
| 13 | 12 | 21 | 18 | 15 | 10 | 16.3 | 11 | | | 1 | | 1 |
| 13 | 12 | 22 | 13 | 21 | 8 | 13 | 9 | 1 | | | | 1 |
| 13 | 12 | 22 | 13 | 26 | 6 | 16.3 | 11 | | | 1 | | 1 |
| 13 | 12 | 22 | 14 | 26 | 10 | 15.3 | 9 | 1 | | | | 1 |
| 13 | 12 | 22 | 14 | 28 | 6 | 13.3 | 11 | | | | 1 | 1 |
| 13 | 12 | 22 | 15 | 21 | 6 | 14.3 | 9 | | | | 1 | 1 |
| 13 | 12 | 22 | 15 | 22 | 12 | 14.3 | 11 | | | 1 | | 1 |
| 13 | 12 | 22 | 15 | 26 | 11 | 15.3 | 11 | | | 1 | | 1 |
| 13 | 12 | 22 | 16 | 25 | 8 | 12 | 10 | | | | 1 | 1 |
| 13 | 12 | 22 | 17 | 24 | 10 | 14.3 | 10 | | | 1 | | 1 |
| 13 | 12 | 22 | 17 | 25 | 11 | 14.3 | 10 | | | | 1 | 1 |
| 13 | 12 | 23 | 13 | 24 | 10 | 13.3 | 10 | | | 1 | | 1 |
| 13 | 12 | 23 | 17 | 23 | 11 | 16.3 | 10 | 1 | | | | 1 |
| 13 | 12 | 24 | 15 | 18 | 6 | 15.3 | 10 | | | 1 | | 1 |
| 13 | 12.3 | 15 | 13 | 23 | 11 | 16.3 | 12 | 1 | | | | 1 |
| 13 | 12.3 | 15 | 16 | 15 | 10 | 15.3 | 9 | | | 1 | | 1 |
| 13 | 12.3 | 16 | 15 | 24 | 11 | 14.3 | 9 | | | 1 | | 1 |
| 13 | 12.3 | 19 | 15 | 25 | 11 | 16.3 | 9 | | | | 1 | 1 |
| 13 | 12.3 | 20 | 13 | 28 | 8 | 11 | 10 | | | | 1 | 1 |
| 13 | 12.3 | 20 | 14 | 18 | 10 | 13.3 | 9 | | | 1 | | 1 |
| 13 | 12.3 | 20 | 14 | 27 | 8 | 12 | 9 | | | | 1 | 1 |
| 13 | 12.3 | 20 | 15 | 24 | 12 | 16.3 | 9 | | | | 1 | 1 |
| 13 | 12.3 | 20 | 16 | 18 | 11 | 16.3 | 11 | | | 1 | | 1 |
| 13 | 12.3 | 20 | 16 | 19 | 10 | 12 | 11 | | | | 1 | 1 |
| 13 | 12.3 | 20 | 16 | 28 | 11 | 16.3 | 9 | | | 1 | | 1 |
| 13 | 12.3 | 20 | 18 | 19 | 8 | 11 | 9 | | | 1 | | 1 |
| 13 | 12.3 | 20 | 18 | 26 | 6 | 14.3 | 10 | | | | 1 | 1 |
| 13 | 12.3 | 21 | 13 | 25 | 11 | 14.3 | 11 | | | 1 | | 1 |
| 13 | 12.3 | 21 | 14 | 24 | 12 | 15.3 | 10 | 1 | | | | 1 |
| 13 | 12.3 | 21 | 14 | 26 | 8 | 13 | 9 | | | | 1 | 1 |
| 13 | 12.3 | 21 | 16 | 28 | 11 | 12 | 10 | | | 1 | | 1 |
| 13 | 12.3 | 22 | 14 | 19 | 10 | 15.3 | 10 | | | 1 | | 1 |
| 13 | 12.3 | 22 | 15 | 26 | 8 | 16.3 | 10 | | | 1 | | 1 |
| 13 | 12.3 | 22 | 16 | 21 | 10 | 11 | 10 | 1 | | | | 1 |
| 13 | 12.3 | 22 | 16 | 25 | 9 | 14 | 11 | 1 | | | | 1 |
| 13 | 12.3 | 22 | 16 | 25 | 11 | 17.3 | 11 | | | 1 | | 1 |
| 13 | 12.3 | 23 | 14 | 23 | 11 | 12 | 10 | | | | 1 | 1 |
| 13 | 13 | 15 | 14 | 23 | 6 | 12 | 9 | 1 | | | | 1 |
| 13 | 13 | 15 | 14 | 27 | 6 | 15.3 | 10 | | | 1 | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=257 | AS N=3 | CN N=259 | Hisp N=138 | Overall N=657 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 13 | 13 | 15 | 16 | 28 | 8 | 11 | 11 | 1 | | | | 1 |
| 13 | 13 | 18 | 17 | 25 | 9 | 13 | 11 | 1 | | | | 1 |
| 13 | 13 | 19 | 15 | 18 | 10 | 15.3 | 11 | | | 1 | | 1 |
| 13 | 13 | 19 | 16 | 24 | 10 | 16.3 | 9 | | | 1 | | 1 |
| 13 | 13 | 20 | 14 | 26 | 10 | 14.3 | 9 | | | 1 | | 1 |
| 13 | 13 | 20 | 15 | 19 | 10 | 16.3 | 10 | | | 1 | | 1 |
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| 13 | 13 | 21 | 14 | 24 | 12 | 15.3 | 11 | 1 | | | | 1 |
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| 13 | 13 | 21 | 17 | 24 | 10 | 14.3 | 11 | | | 1 | | 1 |
| 13 | 13 | 21 | 17 | 26 | 6 | 12 | 9 | | | | 1 | 1 |
| 13 | 13 | 22 | 12 | 23 | 6 | 14.3 | 10 | 1 | | | | 1 |
| 13 | 13 | 22 | 16 | 20 | 10 | 16.3 | 11 | | | 1 | | 1 |
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| 13 | 13.3 | 21 | 14 | 28 | 10 | 15.3 | 9 | | | 1 | | 1 |
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| 13 | 13.3 | 21 | 15 | 27 | 11 | 15.3 | 9 | | | 1 | | 1 |
| 13 | 13.3 | 21 | 16 | 19 | 6 | 15.3 | 9 | | | 1 | | 1 |
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| 13 | 13.3 | 22 | 13 | 26 | 10 | 15.3 | 10 | | | | 1 | 1 |
| 13 | 13.3 | 22 | 14 | 24 | 11 | 15.3 | 11 | | | 1 | | 1 |
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| 13 | 13.3 | 23 | 17 | 25 | 12 | 10 | 9 | | | 1 | | 1 |
| 13 | 14 | 20 | 13 | 19 | 9 | 15.3 | 11 | 1 | | | | 1 |
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| 14 | 9 | 15 | 11 | 22 | 6 | 15.3 | 9 | 1 | | | | 1 |
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| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=257 | AS N=3 | CN N=259 | Hisp N=138 | Overall N=657 |
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| 14 | 9 | 22 | 13 | 24 | 6 | 12 | 9 | 1 | | | | 1 |
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| 14 | 10 | 16 | 17 | 27 | 6 | 11 | 11 | 1 | | | | 1 |
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| 14 | 10 | 24 | 15 | 18 | 7 | 10 | 9 | 1 | | | | 1 |
| 14 | 11 | 15 | 13 | 19 | 8 | 12 | 12 | 1 | | | | 1 |
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| 14 | 11 | 20 | 14 | 27 | 6 | 13 | 11 | 1 | | | | 1 |
| 14 | 11 | 20 | 14 | 29 | 10 | 13.3 | 9 | | | 1 | | 1 |
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| 14 | 11 | 20 | 15 | 21 | 9 | 14 | 11 | 1 | | | | 1 |
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| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=257 | AS N=3 | CN N=259 | Hisp N=138 | Overall N=657 |
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| 14 | 11 | 20 | 16 | 24 | 11 | 14.3 | 9 | | | 1 | | 1 |
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| 14 | 11 | 20 | 16 | 26 | 11 | 12 | 10 | | | | 1 | 1 |
| 14 | 11 | 20 | 17 | 18 | 8 | 15.3 | 9 | | | 1 | | 1 |
| 14 | 11 | 20 | 17 | 24 | 10 | 14.3 | 10 | | | | 1 | 1 |
| 14 | 11 | 20 | 17 | 24 | 11 | 12 | 10 | | | 1 | | 1 |
| 14 | 11 | 20 | 18 | 26 | 6 | 15.3 | 9 | | | 1 | | 1 |
| 14 | 11 | 21 | 10 | 27 | 6 | 15.3 | 11 | 1 | | | | 1 |
| 14 | 11 | 21 | 12 | 26 | 6 | 14.3 | 9 | | | | 1 | 1 |
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| 14 | 11 | 21 | 14 | 25 | 11 | 15.3 | 9 | | | | 1 | 1 |
| 14 | 11 | 21 | 14 | 27 | 9 | 14.3 | 11 | 1 | | | | 1 |
| 14 | 11 | 21 | 14 | 27 | 10 | 14.3 | 9 | | | 1 | | 1 |
| 14 | 11 | 21 | 14 | 30 | 7 | 11 | 11 | | | | 1 | 1 |
| 14 | 11 | 21 | 15 | 22 | 8 | 16.3 | 9 | | | 1 | | 1 |
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| 14 | 11 | 22 | 14 | 21 | 9 | 15.3 | 12 | 1 | | | | 1 |
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| 14 | 11 | 22 | 15 | 24 | 11 | 13 | 9 | | | | 1 | 1 |
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| 14 | 12 | 15 | 14 | 18 | 7 | 14.3 | 10 | 1 | | | | 1 |
| 14 | 12 | 15 | 16 | 22 | 9 | 13.3 | 11 | 1 | | | | 1 |
| 14 | 12 | 15 | 16 | 24 | 6 | 11 | 10 | 1 | | | | 1 |
| 14 | 12 | 15 | 16 | 26 | 9 | 13 | 9 | 1 | | | | 1 |
| 14 | 12 | 15 | 16 | 26 | 12 | 15.3 | 11 | | | | 1 | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=257 | AS N=3 | CN N=259 | Hisp N=138 | Overall N=657 |
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| 14 | 12 | 16 | 13 | 27 | 6 | 14.3 | 10 | 1 | | | | 1 |
| 14 | 12 | 16 | 15 | 21 | 6 | 13 | 11 | 1 | | | | 1 |
| 14 | 12 | 16 | 17 | 27 | 11 | 15.3 | 11 | | | 1 | | 1 |
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| 14 | 12 | 20 | 11 | 21 | 7 | 15.3 | 12 | 1 | | | | 1 |
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| 14 | 12 | 20 | 12 | 18 | 11 | 10 | 10 | 1 | | | | 1 |
| 14 | 12 | 20 | 12 | 30 | 11 | 11 | 9 | | | 1 | | 1 |
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| 14 | 12 | 20 | 14 | 25 | 9 | 12 | 10 | 1 | | | | 1 |
| 14 | 12 | 20 | 14 | 25 | 10 | 12 | 10 | | | | 1 | 1 |
| 14 | 12 | 20 | 14 | 27 | 12 | 15.3 | 10 | | | 1 | | 1 |
| 14 | 12 | 20 | 15 | 25 | 11 | 14.3 | 9 | | | 1 | | 1 |
| 14 | 12 | 20 | 15 | 28 | 10 | 14.3 | 12 | | | 1 | | 1 |
| 14 | 12 | 20 | 16 | 19 | 8 | 15.3 | 11 | | | 1 | | 1 |
| 14 | 12 | 20 | 16 | 21 | 8 | 14.3 | 11 | | | | 1 | 1 |
| 14 | 12 | 20 | 16 | 21 | 11 | 15.3 | 11 | | | 1 | | 1 |
| 14 | 12 | 20 | 16 | 25 | 9 | 14.3 | 10 | | | 1 | | 1 |
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| 14 | 12 | 21 | 15 | 15 | 6 | 15.3 | 11 | | | | 1 | 1 |
| 14 | 12 | 21 | 15 | 24 | 12 | 15.3 | 10 | | | 1 | | 1 |
| 14 | 12 | 21 | 16 | 13 | 10 | 12 | 11 | | | | 1 | 1 |
| 14 | 12 | 21 | 16 | 18 | 10 | 14.3 | 10 | 1 | | | | 1 |
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| 14 | 12 | 22 | 14 | 27 | 8 | 14.3 | 11 | | | 1 | | 1 |
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| 14 | 12 | 22 | 15 | 24 | 8 | 11 | 9 | | | | 1 | 1 |
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| 14 | 12 | 22 | 16 | 24 | 8 | 12 | 13 | 1 | | | | 1 |
| 14 | 12 | 22 | 17 | 25 | 8 | 10 | 10 | | | 1 | | 1 |
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| 14 | 12 | 23 | 16 | 24 | 8 | 15.3 | 9 | | | 1 | | 1 |
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| 14 | 12 | 24 | 13 | 25 | 9 | 16.3 | 11 | | | 1 | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=257 | AS N=3 | CN N=259 | Hisp N=138 | Overall N=657 |
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| 14 | 12.3 | 16 | 16 | 24 | 9 | 12 | 9 | 1 | | | | 1 |
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| 14 | 12.3 | 20 | 16 | 20 | 11 | 15.3 | 9 | | | | 1 | 1 |
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| 14 | 12.3 | 20 | 16 | 27 | 10 | 13 | 10 | | | | 1 | 1 |
| 14 | 12.3 | 21 | 11 | 23 | 6 | 15.3 | 9 | | | 1 | | 1 |
| 14 | 12.3 | 21 | 14 | 22 | 8 | 10 | 11 | 1 | | | | 1 |
| 14 | 12.3 | 21 | 15 | 22 | 11 | 15.3 | 11 | | | | 1 | 1 |
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| 14 | 12.3 | 22 | 14 | 25 | 8 | 14.3 | 10 | | | 1 | | 1 |
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| 14 | 12.3 | 22 | 16 | 18 | 10 | 15.3 | 11 | | | | 1 | 1 |
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| 14 | 12.3 | 24 | 18 | 18 | 10 | 11 | 10 | | | | 1 | 1 |
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| 14 | 13 | 20 | 13 | 27 | 8 | 15.3 | 9 | 1 | | | | 1 |
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| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=257 | AS N=3 | CN N=259 | Hisp N=138 | Overall N=657 |
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| 14 | 13.3 | 21 | 16 | 14 | 12 | 15.3 | 10 | | | | 1 | 1 |
| 14 | 13.3 | 21 | 16 | 19 | 8 | 12 | 9 | | | 1 | | 1 |
| 14 | 13.3 | 22 | 13 | 23 | 10 | 15.3 | 10 | | | 1 | | 1 |
| 14 | 13.3 | 22 | 16 | 26 | 11 | 15.3 | 10 | | | 1 | | 1 |
| 14 | 14 | 15 | 14 | 21 | 9 | 13 | 11 | 1 | | | | 1 |
| 14 | 14 | 16 | 15 | 25 | 11 | 13 | 11 | 1 | | | | 1 |
| 14 | 14 | 21 | 16 | 19 | 9 | 12 | 11 | | | | 1 | 1 |
| 15 | 8 | 20 | 15 | 26 | 11 | 14.3 | 9 | 1 | | | | 1 |
| 15 | 9 | 15 | 13 | 21 | 9 | 13 | 11 | 1 | | | | 1 |
| 15 | 9 | 15 | 14 | 22 | 8 | 14.3 | 11 | 1 | | | | 1 |
| 15 | 9 | 15 | 14 | 27 | 10 | 15.3 | 10 | 1 | | | | 1 |
| 15 | 9 | 15 | 14 | 29 | 8 | 13 | 10 | 1 | | | | 1 |
| 15 | 9 | 19 | 12 | 24 | 6 | 12 | 10 | 1 | | | | 1 |
| 15 | 10 | 15 | 14 | 30 | 8 | 12 | 9 | 1 | | | | 1 |
| 15 | 10 | 16 | 10 | 20 | 11 | 12 | 10 | 1 | | | | 1 |
| 15 | 10 | 21 | 13 | 19 | 9 | 13 | 12 | 1 | | | | 1 |
| 15 | 10 | 21 | 14 | 26 | 11 | 14.3 | 10 | | | | 1 | 1 |
| 15 | 10 | 21 | 15 | 27 | 12 | 15.3 | 9 | | | 1 | | 1 |
| 15 | 10 | 22 | 11 | 21 | 11 | 14.3 | 8 | 1 | | | | 1 |
| 15 | 10 | 22 | 14 | 23 | 10 | 14.3 | 11 | | | 1 | | 1 |
| 15 | 10 | 22 | 14 | 27 | 11 | 12 | 10 | | | 1 | | 1 |
| 15 | 10 | 22 | 15 | 22 | 9 | 15.3 | 9 | | | 1 | | 1 |
| 15 | 10 | 22 | 17 | 26 | 10 | 14.3 | 12 | | | | 1 | 1 |
| 15 | 10 | 23 | 15 | 27 | 12 | 11 | 10 | | | | 1 | 1 |
| 15 | 11 | 15 | 13 | 19 | 12 | 13 | 11 | 1 | | | | 1 |
| 15 | 11 | 15 | 14 | 22 | 11 | 12 | 9 | 1 | | | | 1 |
| 15 | 11 | 15 | 14 | 24 | 7 | 12 | 10 | 1 | | | | 1 |
| 15 | 11 | 15 | 14 | 28 | 6 | 14.3 | 10 | 1 | | | | 1 |
| 15 | 11 | 15 | 15 | 21 | 6 | 14.3 | 10 | 1 | | | | 1 |
| 15 | 11 | 15 | 16 | 20 | 9 | 15.3 | 10 | | | 1 | | 1 |
| 15 | 11 | 16 | 14 | 21 | 11 | 12 | 9 | 1 | | | | 1 |
| 15 | 11 | 16 | 14 | 23 | 8 | 12 | 11 | | | 1 | | 1 |
| 15 | 11 | 18 | 17 | 25 | 11 | 13 | 11 | | | 1 | | 1 |
| 15 | 11 | 19 | 13 | 21 | 8 | 9 | 10 | 1 | | | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=257 | AS N=3 | CN N=259 | Hisp N=138 | Overall N=657 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 15 | 11 | 19 | 14 | 19 | 9 | 14.3 | 10 | 1 | | | | 1 |
| 15 | 11 | 19 | 15 | 21 | 10 | 13 | 11 | 1 | | | | 1 |
| 15 | 11 | 20 | 13 | 29 | 9 | 13 | 11 | | | | 1 | 1 |
| 15 | 11 | 20 | 14 | 19 | 6 | 14.3 | 9 | 1 | | | | 1 |
| 15 | 11 | 20 | 15 | 23 | 11 | 13.3 | 11 | | | 1 | | 1 |
| 15 | 11 | 20 | 15 | 26 | 8 | 16.3 | 11 | | | 1 | | 1 |
| 15 | 11 | 20 | 16 | 26 | 11 | 16.3 | 11 | 1 | | | | 1 |
| 15 | 11 | 20 | 17 | 18 | 11 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 11 | 20 | 18 | 20 | 11 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 11 | 21 | 11 | 19 | 9 | 16.3 | 11 | 1 | | | | 1 |
| 15 | 11 | 21 | 11 | 21 | 7 | 12 | 10 | 1 | | | | 1 |
| 15 | 11 | 21 | 14 | 28 | 9 | 14 | 10 | 1 | | | | 1 |
| 15 | 11 | 21 | 15 | 21 | 12 | 13 | 10 | 1 | | | | 1 |
| 15 | 11 | 21 | 15 | 22 | 8 | 12 | 9 | | | 1 | | 1 |
| 15 | 11 | 21 | 15 | 23 | 8 | 16.3 | 11 | 1 | | | | 1 |
| 15 | 11 | 21 | 15 | 23 | 10 | 15.3 | 10 | | | 1 | | 1 |
| 15 | 11 | 21 | 15 | 27 | 9 | 12 | 11 | 1 | | | | 1 |
| 15 | 11 | 21 | 16 | 23 | 9 | 12 | 11 | 1 | | | | 1 |
| 15 | 11 | 21 | 16 | 25 | 6 | 13 | 10 | | | 1 | | 1 |
| 15 | 11 | 22 | 13 | 22 | 9 | 12 | 12 | 1 | | | | 1 |
| 15 | 11 | 22 | 14 | 28 | 10 | 15.3 | 12 | 1 | | | | 1 |
| 15 | 11 | 23 | 14 | 18 | 12 | 16.3 | 10 | | | 1 | | 1 |
| 15 | 11 | 23 | 15 | 20 | 9 | 15.3 | 10 | 1 | | | | 1 |
| 15 | 11 | 23 | 16 | 26 | 8 | 16.3 | 10 | | | | 1 | 1 |
| 15 | 11 | 24 | 13 | 26 | 6 | 13 | 9 | | | 1 | | 1 |
| 15 | 11.3 | 15 | 14 | 21 | 9 | 12 | 11 | 1 | | | | 1 |
| 15 | 11.3 | 16 | 14 | 30 | 8 | 13 | 11 | 1 | | | | 1 |
| 15 | 11.3 | 19 | 13 | 19 | 9 | 15.3 | 11 | 1 | | | | 1 |
| 15 | 12 | 15 | 12 | 25 | 12 | 14.3 | 11 | 1 | | | | 1 |
| 15 | 12 | 15 | 13 | 19 | 10 | 15.3 | 11 | 1 | | | | 1 |
| 15 | 12 | 15 | 16 | 20 | 10 | 14.3 | 10 | | | | 1 | 1 |
| 15 | 12 | 15 | 16 | 23 | 9 | 12 | 10 | 1 | | | | 1 |
| 15 | 12 | 16 | 14 | 30 | 10 | 12 | 11 | 1 | | | | 1 |
| 15 | 12 | 18 | 14 | 21 | 11 | 13 | 10 | 1 | | | | 1 |
| 15 | 12 | 19 | 13 | 20 | 9 | 15.3 | 10 | 1 | | | | 1 |
| 15 | 12 | 19 | 16 | 25 | 10 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 12 | 20 | 13 | 21 | 6 | 12 | 10 | | | | 1 | 1 |
| 15 | 12 | 20 | 13 | 24 | 7 | 14.3 | 12 | | | | 1 | 1 |
| 15 | 12 | 20 | 15 | 18 | 6 | 12 | 10 | 1 | | | | 1 |
| 15 | 12 | 20 | 15 | 22 | 10 | 14.3 | 10 | | | 1 | | 1 |
| 15 | 12 | 20 | 15 | 23 | 8 | 15.3 | 10 | | | 1 | | 1 |
| 15 | 12 | 20 | 15 | 23 | 10 | 15.3 | 9 | | | | 1 | 1 |
| 15 | 12 | 20 | 15 | 26 | 8 | 12 | 9 | | | | 1 | 1 |
| 15 | 12 | 20 | 17 | 25 | 10 | 14 | 9 | | | | 1 | 1 |
| 15 | 12 | 20 | 18 | 24 | 10 | 15.3 | 9 | | | 1 | | 1 |
| 15 | 12 | 21 | 12 | 27 | 8 | 14.3 | 10 | | | | 1 | 1 |
| 15 | 12 | 21 | 13 | 22 | 10 | 14 | 10 | 1 | | | | 1 |
| 15 | 12 | 21 | 14 | 26 | 10 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 12 | 21 | 15 | 26 | 10 | 15.3 | 10 | | | 1 | | 1 |
| 15 | 12 | 21 | 15 | 26 | 11 | 15.3 | 11 | | | | 1 | 1 |
| 15 | 12 | 21 | 15 | 26 | 11 | 15.3 | 12 | 1 | | | | 1 |
| 15 | 12 | 21 | 16 | 19 | 12 | 12 | 11 | 1 | | | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=257 | AS N=3 | CN N=259 | Hisp N=138 | Overall N=657 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 15 | 12 | 21 | 16 | 25 | 9 | 15.3 | 10 | 1 | | | | 1 |
| 15 | 12 | 21 | 16 | 26 | 12 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 12 | 21 | 16 | 27 | 10 | 15.3 | 11 | | | 1 | | 1 |
| 15 | 12 | 21 | 17 | 19 | 10 | 14.3 | 11 | 1 | | | | 1 |
| 15 | 12 | 22 | 12 | 24 | 9 | 13 | 11 | 1 | | | | 1 |
| 15 | 12 | 22 | 12 | 26 | 8 | 12 | 11 | 1 | | | | 1 |
| 15 | 12 | 22 | 12 | 28 | 11 | 15.3 | 10 | | | 1 | | 1 |
| 15 | 12 | 22 | 14 | 27 | 10 | 15.3 | 11 | | | | 1 | 1 |
| 15 | 12 | 22 | 15 | 23 | 10 | 12 | 10 | | | 1 | | 1 |
| 15 | 12 | 22 | 15 | 25 | 6 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 12 | 22 | 16 | 19 | 6 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 12 | 23 | 14 | 18 | 9 | 15.3 | 11 | 1 | | | | 1 |
| 15 | 12 | 23 | 15 | 18 | 10 | 10 | 12 | | | 1 | | 1 |
| 15 | 12 | 23 | 15 | 24 | 6 | 14.3 | 11 | 1 | | | | 1 |
| 15 | 12 | 23 | 15 | 24 | 6 | 15.3 | 11 | | | 1 | | 1 |
| 15 | 12 | 23 | 17 | 25 | 10 | 16.3 | 11 | | | | 1 | 1 |
| 15 | 12 | 24 | 12 | 23 | 6 | 17.3 | 12 | | | 1 | | 1 |
| 15 | 12.3 | 15 | 13 | 27 | 9 | 14.3 | 11 | 1 | | | | 1 |
| 15 | 12.3 | 16 | 15 | 24 | 6 | 15.3 | 11 | | | | 1 | 1 |
| 15 | 12.3 | 19 | 13 | 19 | 8 | 11 | 10 | 1 | | | | 1 |
| 15 | 12.3 | 20 | 14 | 24 | 6 | 15.3 | 10 | 1 | | | | 1 |
| 15 | 12.3 | 20 | 14 | 26 | 12 | 14.3 | 10 | | | | 1 | 1 |
| 15 | 12.3 | 20 | 15 | 18 | 9 | 15.3 | 10 | | | 1 | | 1 |
| 15 | 12.3 | 20 | 15 | 18 | 11 | 15.3 | 9 | | | 1 | | 1 |
| 15 | 12.3 | 20 | 15 | 26 | 10 | 14.3 | 12 | | | 1 | | 1 |
| 15 | 12.3 | 20 | 16 | 25 | 8 | 11 | 10 | | | | 1 | 1 |
| 15 | 12.3 | 20 | 17 | 24 | 8 | 14.3 | 9 | | | | 1 | 1 |
| 15 | 12.3 | 20 | 17 | 25 | 8 | 15.3 | 11 | 1 | | | | 1 |
| 15 | 12.3 | 21 | 15 | 24 | 10 | 15.3 | 9 | | | 1 | | 1 |
| 15 | 12.3 | 21 | 15 | 30 | 10 | 16.3 | 11 | | | 1 | | 1 |
| 15 | 12.3 | 21 | 16 | 24 | 12 | 15.3 | 11 | | | | 1 | 1 |
| 15 | 12.3 | 22 | 15 | 22 | 10 | 15.3 | 9 | | | 1 | | 1 |
| 15 | 12.3 | 22 | 15 | 25 | 10 | 16.3 | 11 | | | | 1 | 1 |
| 15 | 12.3 | 22 | 16 | 26 | 8 | 11 | 9 | | | 1 | | 1 |
| 15 | 13 | 15 | 13 | 16 | 11 | 16.3 | 10 | 1 | | | | 1 |
| 15 | 13 | 15 | 13 | 27 | 8 | 13 | 12 | 1 | | | | 1 |
| 15 | 13 | 16 | 11 | 21 | 6 | 15.3 | 10 | 1 | | | | 1 |
| 15 | 13 | 16 | 14 | 23 | 6 | 12 | 11 | 1 | | | | 1 |
| 15 | 13 | 16 | 14 | 24 | 11 | 15.3 | 10 | | | 1 | | 1 |
| 15 | 13 | 20 | 11 | 21 | 10 | 14 | 9 | | | 1 | | 1 |
| 15 | 13 | 20 | 15 | 21 | 8 | 15.3 | 11 | 1 | | | | 1 |
| 15 | 13 | 20 | 16 | 18 | 8 | 12 | 11 | | | 1 | | 1 |
| 15 | 13 | 20 | 16 | 23 | 8 | 14.3 | 11 | | | 1 | | 1 |
| 15 | 13 | 20 | 16 | 24 | 10 | 15.3 | 11 | | | | 1 | 1 |
| 15 | 13 | 20 | 16 | 28 | 12 | 15.3 | 9 | | | 1 | | 1 |
| 15 | 13 | 21 | 15 | 19 | 6 | 15.3 | 10 | | | 1 | | 1 |
| 15 | 13 | 21 | 15 | 23 | 10 | 17.3 | 10 | | | | 1 | 1 |
| 15 | 13 | 21 | 16 | 23 | 12 | 15.3 | 11 | | | 1 | | 1 |
| 15 | 13 | 21 | 17 | 18 | 6 | 14.3 | 9 | | | | 1 | 1 |
| 15 | 13 | 22 | 13 | 29 | 6 | 15.3 | 9 | | | 1 | | 1 |
| 15 | 13 | 22 | 14 | 19 | 9 | 11 | 10 | 1 | | | | 1 |
| 15 | 13 | 22 | 16 | 24 | 8 | 12 | 9 | | | | 1 | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=257 | AS N=3 | CN N=259 | Hisp N=138 | Overall N=657 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 15 | 13 | 22 | 16 | 24 | 11 | 15.3 | 11 | | | 1 | | 1 |
| 15 | 13 | 22 | 16 | 24 | 12 | 12 | 10 | 1 | | | | 1 |
| 15 | 13 | 22 | 17 | 18 | 12 | 16.3 | 10 | | | 1 | | 1 |
| 15 | 13 | 23 | 12 | 23 | 10 | 14.3 | 9 | | | 1 | | 1 |
| 15 | 13.3 | 19 | 15 | 29 | 8 | 16.3 | 11 | | | 1 | | 1 |
| 15 | 13.3 | 20 | 16 | 27 | 12 | 15.3 | 10 | | | 1 | | 1 |
| 15 | 13.3 | 20 | 17 | 18 | 10 | 15.3 | 12 | | | | 1 | 1 |
| 15 | 13.3 | 21 | 14 | 25 | 10 | 16.3 | 9 | 1 | | | | 1 |
| 15 | 13.3 | 21 | 16 | 24 | 11 | 13.3 | 11 | | | | 1 | 1 |
| 15 | 13.3 | 22 | 16 | 25 | 10 | 14.3 | 9 | | | 1 | | 1 |
| 15 | 13.3 | 22 | 18 | 27 | 8 | 14.3 | 11 | | | 1 | | 1 |
| 15 | 14 | 20 | 17 | 24 | 11 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 14 | 23 | 16 | 22 | 9 | 12 | 11 | 1 | | | | 1 |
| 15.3 | 12 | 16 | 16 | 25 | 8 | 11 | 10 | | | | 1 | 1 |
| 16 | 7 | 21 | 15 | 27 | 10 | 15.3 | 11 | 1 | | | | 1 |
| 16 | 9 | 15 | 15 | 24 | 9 | 14 | 11 | 1 | | | | 1 |
| 16 | 10 | 21 | 14 | 22 | 10 | 15.3 | 9 | 1 | | | | 1 |
| 16 | 11 | 16 | 16 | 26 | 9 | 11 | 11 | 1 | | | | 1 |
| 16 | 11 | 20 | 15 | 25 | 12 | 14.3 | 10 | | | | 1 | 1 |
| 16 | 11 | 20 | 16 | 28 | 6 | 12 | 10 | | | | 1 | 1 |
| 16 | 11 | 21 | 15 | 21 | 8 | 13 | 10 | 1 | | | | 1 |
| 16 | 11 | 21 | 16 | 25 | 10 | 13 | 10 | | | | 1 | 1 |
| 16 | 11 | 22 | 14 | 21 | 9 | 13 | 11 | 1 | | | | 1 |
| 16 | 11 | 23 | 11 | 21 | 9 | 14 | 11 | 1 | | | | 1 |
| 16 | 11 | 23 | 14 | 27 | 10 | 14.3 | 11 | | | 1 | | 1 |
| 16 | 12 | 16 | 13 | 23 | 8 | 16.3 | 8 | 1 | | | | 1 |
| 16 | 12 | 16 | 16 | 19 | 6 | 15.3 | 9 | | 1 | | | 1 |
| 16 | 12 | 16 | 16 | 27 | 9 | 15.3 | 12 | 1 | | | | 1 |
| 16 | 12 | 18 | 15 | 22 | 8 | 12 | 8 | 1 | | | | 1 |
| 16 | 12 | 20 | 12 | 18 | 8 | 14.3 | 11 | 1 | | | | 1 |
| 16 | 12 | 20 | 13 | 29 | 6 | 14.3 | 11 | | | 1 | | 1 |
| 16 | 12 | 20 | 14 | 21 | 11 | 14.3 | 9 | | | | 1 | 1 |
| 16 | 12 | 20 | 14 | 26 | 11 | 12 | 11 | 1 | | | | 1 |
| 16 | 12 | 22 | 15 | 21 | 9 | 13 | 12 | 1 | | | | 1 |
| 16 | 12 | 24 | 17 | 18 | 9 | 12 | 10 | | | 1 | | 1 |
| 16 | 12.3 | 15 | 12 | 22 | 9 | 12 | 11 | 1 | | | | 1 |
| 16 | 12.3 | 20 | 17 | 16 | 10 | 16.3 | 9 | | | 1 | | 1 |
| 16 | 12.3 | 21 | 13 | 24 | 10 | 16.3 | 12 | | | 1 | | 1 |
| 16 | 12.3 | 21 | 15 | 24 | 9 | 13 | 9 | 1 | | | | 1 |
| 16 | 13 | 15 | 15 | 21 | 10 | 15.3 | 10 | 1 | | | | 1 |
| 16 | 13 | 15 | 15 | 28 | 9 | 15.3 | 10 | | | 1 | | 1 |
| 16 | 13 | 21 | 15 | 27 | 6 | 16.3 | 10 | | | 1 | | 1 |
| 16 | 13 | 23 | 16 | 24 | 9 | 14.3 | 11 | | | 1 | | 1 |
| 16 | 13.3 | 20 | 15 | 24 | 8 | 14.3 | 11 | | | 1 | | 1 |
| 16 | 13.3 | 20 | 17 | 25 | 10 | 15.3 | 11 | | | | 1 | 1 |
| 16 | 14 | 23 | 15 | 23 | 10 | 15.3 | 13 | | | | 1 | 1 |
| 16.3 | 13.3 | 22 | 15 | 26 | 11 | 14.3 | 10 | | | | 1 | 1 |
| 17 | 9 | 22 | 13 | 19 | 6 | 12 | 13 | 1 | | | | 1 |
| 17 | 10 | 21 | 16 | 25 | 10 | 15.3 | 10 | 1 | | | | 1 |
| 17 | 12 | 21 | 16 | 26 | 11 | 15.3 | 11 | | | | 1 | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=257 | AS N=3 | CN N=259 | Hisp N=138 | Overall N=657 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | Total number of haplotypes: | 257 | 3 | 258 | 138 | 656 |
| | | | | | | | Total number of unique haplotypes: | 257 | 3 | 257 | 138 | 655 |
| | | | | | | | Count of most common haplotype: | 1 | 1 | 2 | 1 | 2 |

**Table B6. Haplotypes observed within sample set B for proposed linkage group 4.** AA: African American, AS: U.S. Asian, CN: U.S. Caucasian, Hisp: U.S. Hispanic, N: number of samples.

| GATA31E08 | DXS10147 | DXS7423 | AA N=256 | AS N=3 | CN N=260 | Hisp N=138 | Overall N=657 |
|---|---|---|---|---|---|---|---|
| 7 | 8 | 14 | 2 | | | | 2 |
| 7 | 9 | 14 | 1 | | | | 1 |
| 7 | 9 | 15 | 1 | | | | 1 |
| 7 | 9 | 16 | 1 | | | | 1 |
| 8 | 6 | 13 | 1 | | | | 1 |
| 8 | 6 | 14 | 1 | | | | 1 |
| 8 | 7 | 14 | 1 | | | | 1 |
| 8 | 7 | 15 | 3 | | | | 3 |
| 8 | 8 | 13 | | | | 1 | 1 |
| 8 | 8 | 14 | 1 | | | | 1 |
| 8 | 8 | 17 | | | | 1 | 1 |
| 8 | 9 | 13 | 1 | | | | 1 |
| 8 | 9 | 14 | | | | 1 | 1 |
| 9 | 6 | 13 | 1 | | 3 | | 4 |
| 9 | 6 | 14 | 2 | | 5 | 1 | 8 |
| 9 | 6 | 15 | | | 6 | 7 | 13 |
| 9 | 6 | 16 | 1 | | 2 | 1 | 4 |
| 9 | 6 | 17 | 1 | | 2 | 1 | 4 |
| 9 | 7 | 14 | 7 | | 1 | 1 | 9 |
| 9 | 7 | 15 | 2 | | | | 2 |
| 9 | 7 | 16 | 3 | | | | 3 |
| 9 | 8 | 13 | | | 2 | | 2 |
| 9 | 8 | 14 | 10 | | 5 | 3 | 18 |
| 9 | 8 | 15 | 5 | | 2 | 2 | 9 |
| 9 | 8 | 16 | | | 1 | | 1 |
| 9 | 8 | 17 | | | | 2 | 2 |
| 9 | 9 | 13 | 1 | | 5 | | 6 |
| 9 | 9 | 14 | 4 | | 5 | 1 | 10 |
| 9 | 9 | 15 | | | 11 | 8 | 19 |
| 9 | 9 | 16 | | | 4 | 2 | 6 |
| 9 | 10 | 15 | 3 | | 1 | 1 | 5 |
| 10 | 6 | 14 | 2 | | | | 2 |
| 10 | 6 | 15 | 1 | | | 1 | 2 |
| 10 | 7 | 14 | 4 | | | | 4 |
| 10 | 7 | 15 | 7 | | | | 7 |
| 10 | 7 | 16 | 1 | | | | 1 |
| 10 | 8 | 14 | 8 | | 1 | | 9 |
| 10 | 8 | 15 | 4 | | 1 | | 5 |
| 10 | 9 | 13 | | | 1 | | 1 |

| GATA31E08 | DXS10147 | DXS7423 | AA N=256 | AS N=3 | CN N=260 | Hisp N=138 | Overall N=657 |
|---|---|---|---|---|---|---|---|
| 10 | 9 | 14 | 3 | | | | 3 |
| 10 | 9 | 15 | | | 1 | | 1 |
| 10 | 10 | 15 | 1 | | | | 1 |
| 11 | 6 | 14 | 1 | | 5 | | 6 |
| 11 | 6 | 15 | 1 | | 7 | 5 | 13 |
| 11 | 6 | 16 | | | 3 | | 3 |
| 11 | 7 | 12 | 2 | | | | 2 |
| 11 | 7 | 14 | 5 | | | | 5 |
| 11 | 7 | 15 | 1 | | | 1 | 2 |
| 11 | 7 | 16 | 2 | | 1 | 1 | 4 |
| 11 | 7 | 17 | | | 3 | | 3 |
| 11 | 8 | 13 | | | 2 | | 2 |
| 11 | 8 | 14 | 4 | | 9 | 5 | 18 |
| 11 | 8 | 15 | 3 | | 8 | 4 | 15 |
| 11 | 8 | 16 | | | 2 | | 2 |
| 11 | 9 | 13 | 2 | | 5 | 3 | 10 |
| 11 | 9 | 14 | 2 | | 7 | | 9 |
| 11 | 9 | 15 | | | 4 | 3 | 7 |
| 11 | 9 | 16 | 2 | | 1 | 1 | 4 |
| 11 | 9 | 17 | | | 1 | | 1 |
| 11 | 10 | 14 | | | 1 | | 1 |
| 11 | 10 | 15 | | | | 1 | 1 |
| 12 | 6 | 13 | | | | 1 | 1 |
| 12 | 6 | 14 | 3 | | 2 | 2 | 7 |
| 12 | 6 | 15 | 4 | 1 | 6 | 6 | 17 |
| 12 | 6 | 16 | | | 2 | 1 | 3 |
| 12 | 6 | 17 | | | 1 | | 1 |
| 12 | 7 | 8 | 2 | | | | 2 |
| 12 | 7 | 12 | 2 | | | | 2 |
| 12 | 7 | 13 | 1 | | 1 | | 2 |
| 12 | 7 | 14 | 11 | | | 2 | 13 |
| 12 | 7 | 15 | 13 | 1 | | 1 | 15 |
| 12 | 7 | 16 | 1 | | | 2 | 3 |
| 12 | 8 | 13 | 2 | | 3 | 1 | 6 |
| 12 | 8 | 14 | 18 | | 6 | 6 | 30 |
| 12 | 8 | 15 | 2 | | 3 | 7 | 12 |
| 12 | 8 | 16 | 3 | | 3 | 1 | 7 |
| 12 | 8 | 17 | | | | 3 | 3 |
| 12 | 9 | 13 | 4 | | 4 | 1 | 9 |
| 12 | 9 | 14 | 4 | | 2 | 1 | 7 |
| 12 | 9 | 15 | 4 | | 7 | 3 | 14 |
| 12 | 9 | 16 | 6 | | 8 | 2 | 16 |
| 12 | 9 | 17 | | | 1 | | 1 |
| 12 | 10 | 13 | 1 | | | | 1 |
| 12 | 10 | 14 | 1 | | | | 1 |
| 12 | 10 | 15 | 1 | | 2 | | 3 |
| 12 | 11 | 14 | 1 | | | | 1 |
| 13 | 6 | 13 | | | | 1 | 1 |
| 13 | 6 | 14 | 3 | | 6 | 1 | 10 |
| 13 | 6 | 15 | 2 | | 5 | 4 | 11 |
| 13 | 6 | 16 | 2 | | 3 | 2 | 7 |
| 13 | 7 | 13 | 2 | | | | 2 |
| 13 | 7 | 14 | 8 | | | | 8 |
| 13 | 7 | 15 | 1 | | | | 1 |
| 13 | 7 | 16 | | | | 1 | 1 |
| 13 | 7 | 17 | | | 1 | | 1 |

| GATA31E08 | DXS10147 | DXS7423 | AA N=256 | AS N=3 | CN N=260 | Hisp N=138 | Overall N=657 |
|---|---|---|---|---|---|---|---|
| 13 | 8 | 13 | 1 | | | 1 | 2 |
| 13 | 8 | 14 | 10 | | 7 | 4 | 21 |
| 13 | 8 | 15 | 5 | 1 | 5 | 4 | 15 |
| 13 | 8 | 16 | 3 | | 1 | 1 | 5 |
| 13 | 8 | 17 | | | | 1 | 1 |
| 13 | 9 | 13 | 2 | | 1 | 1 | 4 |
| 13 | 9 | 14 | 3 | | 9 | 3 | 15 |
| 13 | 9 | 15 | 3 | | 11 | 3 | 17 |
| 13 | 9 | 16 | | | 6 | 1 | 7 |
| 13 | 9 | 17 | | | 2 | | 2 |
| 13 | 10 | 14 | 1 | | | | 1 |
| 13 | 10 | 15 | | | | 1 | 1 |
| 13 | 10 | 16 | | | | 1 | 1 |
| 14 | 6 | 13 | | | 1 | | 1 |
| 14 | 6 | 14 | | | 1 | 1 | 2 |
| 14 | 6 | 15 | | | 4 | 3 | 7 |
| 14 | 6 | 16 | | | 2 | | 2 |
| 14 | 7 | 14 | 3 | | 1 | | 4 |
| 14 | 7 | 15 | 1 | | 1 | | 2 |
| 14 | 7 | 16 | 2 | | 2 | 1 | 5 |
| 14 | 8 | 14 | | | 3 | | 3 |
| 14 | 8 | 15 | 2 | | 3 | 3 | 8 |
| 14 | 8 | 16 | | | 1 | | 1 |
| 14 | 8 | 17 | | | | 1 | 1 |
| 14 | 9 | 13 | 1 | | 2 | | 3 |
| 14 | 9 | 14 | 3 | | 5 | 1 | 9 |
| 14 | 9 | 15 | 2 | | 4 | | 6 |
| 14 | 9 | 16 | | | 2 | | 2 |
| 14 | 10 | 15 | 1 | | | | 1 |
| 15 | 7 | 15 | 1 | | | | 1 |
| 15 | 8 | 14 | 1 | | | | 1 |
| 15 | 8 | 15 | | | 1 | | 1 |
| 15 | 9 | 13 | | | 1 | | 1 |
| | Total number of haplotypes: | | 86 | 3 | 77 | 63 | 128 |
| | Total number of unique haplotypes: | | 32 | 3 | 24 | 35 | 41 |
| | Count of most common haplotype: | | 18 | 1 | 11 | 8 | 30 |

**Table B7. (Supplementary Table 4). Haplotypes observed within sample set C for proposed linkage group 1.** AA: African American, AS: U.S. Asian, CN: U.S. Caucasian, Hisp: U.S. Hispanic, N: number of samples.

| DXS8378 | DXS9902 | DXS6795 | AA N=108 | CN N=165 | Hisp N=150 | Overall N=423 |
|---|---|---|---|---|---|---|
| 8 | 10 | 11 | | 1 | | 1 |
| 9 | 9 | 11 | | 1 | | 1 |
| 9 | 10 | 9 | | 1 | | 1 |
| 9 | 10 | 11 | | 1 | | 1 |
| 9 | 11 | 9 | | 1 | | 1 |
| 9 | 11 | 13 | | | 1 | 1 |
| 9 | 11 | 15 | 1 | | | 1 |
| 9 | 11.1 | 12 | | | 1 | 1 |
| 10 | 8 | 10 | 1 | | | 1 |
| 10 | 8 | 11 | 1 | | | 1 |

| DXS8378 | DXS9902 | DXS6795 | AA N=108 | CN N=165 | Hisp N=150 | Overall N=423 |
|---|---|---|---|---|---|---|
| 10 | 8 | 13 | 1 | | | 1 |
| 10 | 9 | 9 | | 1 | | 1 |
| 10 | 9 | 11 | | 2 | | 2 |
| 10 | 9 | 12 | 1 | | | 1 |
| 10 | 9 | 15 | 1 | | | 1 |
| 10 | 10 | 9 | 1 | 4 | 7 | 12 |
| 10 | 10 | 10 | 2 | 1 | 4 | 7 |
| 10 | 10 | 11 | 2 | 12 | 6 | 20 |
| 10 | 10 | 12 | | | 4 | 4 |
| 10 | 10 | 13 | 2 | 5 | 9 | 16 |
| 10 | 10 | 14 | 1 | | | 1 |
| 10 | 10 | 15 | 2 | | | 2 |
| 10 | 11 | 9 | 2 | 7 | 2 | 11 |
| 10 | 11 | 10 | 2 | 1 | 4 | 7 |
| 10 | 11 | 11 | 2 | 9 | 8 | 19 |
| 10 | 11 | 12 | 1 | 1 | 6 | 8 |
| 10 | 11 | 13 | | 4 | 7 | 11 |
| 10 | 11 | 14 | | | 1 | 1 |
| 10 | 11 | 15 | 2 | | | 2 |
| 10 | 11.1 | 9 | 1 | | 1 | 2 |
| 10 | 11.1 | 11 | | 1 | | 1 |
| 10 | 11.1 | 13 | | 1 | | 1 |
| 10 | 12 | 9 | | 5 | 1 | 6 |
| 10 | 12 | 10 | | | 2 | 2 |
| 10 | 12 | 11 | 1 | 8 | 1 | 10 |
| 10 | 12 | 12 | | 1 | 2 | 3 |
| 10 | 12 | 13 | | 3 | 2 | 5 |
| 10 | 12 | 15 | 2 | | | 2 |
| 10 | 12 | 16 | 1 | | | 1 |
| 10 | 13 | 10 | 1 | | | 1 |
| 11 | 8 | 15 | 1 | | | 1 |
| 11 | 9 | 10 | 1 | | | 1 |
| 11 | 9 | 11 | | 1 | | 1 |
| 11 | 9 | 12 | 1 | | | 1 |
| 11 | 9 | 13 | | | 1 | 1 |
| 11 | 10 | 9 | 1 | 6 | 3 | 10 |
| 11 | 10 | 10 | 4 | | 1 | 5 |
| 11 | 10 | 11 | 2 | 8 | 3 | 13 |
| 11 | 10 | 12 | 2 | 1 | 1 | 4 |
| 11 | 10 | 13 | 1 | 3 | 10 | 14 |
| 11 | 10 | 14 | 1 | | 1 | 2 |
| 11 | 10 | 15 | 2 | 1 | 1 | 4 |
| 11 | 11 | 9 | 3 | 6 | 2 | 11 |
| 11 | 11 | 10 | 2 | 3 | 2 | 7 |
| 11 | 11 | 11 | | 12 | 7 | 19 |
| 11 | 11 | 12 | 1 | 1 | | 2 |
| 11 | 11 | 13 | 1 | 3 | 7 | 11 |
| 11 | 11 | 14 | | | 1 | 1 |
| 11 | 11 | 15 | 1 | | | 1 |
| 11 | 11.1 | 9 | | | 2 | 2 |
| 11 | 11.1 | 13 | 1 | 1 | 1 | 3 |
| 11 | 12 | 9 | | 1 | 1 | 2 |
| 11 | 12 | 10 | 7 | | 2 | 9 |
| 11 | 12 | 11 | 1 | 4 | 5 | 10 |
| 11 | 12 | 13 | | 1 | 1 | 2 |

| DXS8378 | DXS9902 | DXS6795 | AA N=108 | CN N=165 | Hisp N=150 | Overall N=423 |
|---|---|---|---|---|---|---|
| 11 | 12 | 15 | 2 | | | 2 |
| 11 | 13 | 11 | 2 | | | 2 |
| 12 | 8 | 9 | 1 | | | 1 |
| 12 | 8 | 10 | 1 | | | 1 |
| 12 | 9 | 10 | 1 | | | 1 |
| 12 | 9 | 13 | | 2 | | 2 |
| 12 | 10 | 9 | 1 | 7 | 2 | 10 |
| 12 | 10 | 10 | 1 | | 1 | 2 |
| 12 | 10 | 11 | 2 | 2 | 3 | 7 |
| 12 | 10 | 12 | 2 | | 4 | 6 |
| 12 | 10 | 13 | 1 | 2 | 4 | 7 |
| 12 | 10 | 15 | 4 | | | 4 |
| 12 | 11 | 9 | 1 | 6 | | 7 |
| 12 | 11 | 10 | 5 | | | 5 |
| 12 | 11 | 11 | 4 | 4 | 6 | 14 |
| 12 | 11 | 12 | 1 | | 1 | 2 |
| 12 | 11 | 13 | 1 | 1 | 2 | 4 |
| 12 | 11 | 14 | 1 | | | 1 |
| 12 | 11 | 16 | 1 | | | 1 |
| 12 | 11.1 | 9 | | 1 | | 1 |
| 12 | 12 | 9 | 2 | 3 | 1 | 6 |
| 12 | 12 | 10 | 1 | 1 | | 2 |
| 12 | 12 | 11 | 1 | 3 | 2 | 6 |
| 12 | 12 | 12 | | 1 | 1 | 2 |
| 12 | 12 | 15 | 3 | | | 3 |
| 13 | 8 | 14 | 1 | | | 1 |
| 13 | 10 | 9 | 1 | 1 | | 2 |
| 13 | 10 | 11 | 1 | 1 | | 2 |
| 13 | 10 | 13 | | 1 | | 1 |
| 13 | 11 | 10 | | | 1 | 1 |
| 13 | 11 | 11 | | 1 | | 1 |
| 13 | 11 | 13 | | 1 | | 1 |
| 13 | 11.1 | 9 | | | 1 | 1 |
| 13 | 11.1 | 11 | | 1 | | 1 |
| 13 | 12 | 9 | | 1 | | 1 |
| 13 | 12 | 13 | 1 | 1 | | 2 |
| 13 | 12 | 15 | 1 | | | 1 |
| 14 | 11.1 | 11 | | 1 | | 1 |
| | | Total number of haplotypes: | 67 | 58 | 51 | 103 |
| | | Total number of unique haplotypes: | 42 | 32 | 21 | 43 |
| | | Count of most common haplotype: | 7 | 12 | 10 | 20 |

**Table B8. (Supplementary Table 5). Haplotypes observed within sample set C for proposed linkage group 2.** AA: African American, AS: U.S. Asian, CN: U.S. Caucasian, Hisp: U.S. Hispanic, N: number of samples.

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=108 | CN N=165 | Hisp N=150 | Overall N=423 |
|---|---|---|---|---|---|---|---|---|---|---|---|
| 11 | 11 | 15 | 16 | 28 | 7 | 12 | 10 | 1 | | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=108 | CN N=165 | Hisp N=150 | Overall N=423 |
|---|---|---|---|---|---|---|---|---|---|---|---|
| 11 | 11 | 16 | 16 | 24 | 9 | 15.3 | 10 | 1 | | | 1 |
| 11 | 12.3 | 21 | 15 | 26 | 6 | 14.3 | 10 | 1 | | | 1 |
| 12 | 10 | 20 | 11 | 26 | 6 | 15.3 | 12 | 1 | | | 1 |
| 12 | 10 | 21 | 16 | 15 | 8 | 13.3 | 10 | | 1 | | 1 |
| 12 | 11 | 15 | 14 | 18 | 9 | 12 | 11 | 1 | | | 1 |
| 12 | 11 | 15 | 14 | 22 | 11 | 12 | 9 | 1 | | | 1 |
| 12 | 11 | 16 | 16 | 24 | 6 | 14.3 | 11 | 1 | | | 1 |
| 12 | 11 | 19 | 11 | 21 | 9 | 12 | 11 | 1 | | | 1 |
| 12 | 11 | 19 | 16 | 18 | 11 | 14.3 | 10 | | | 1 | 1 |
| 12 | 11 | 19 | 16 | 28 | 11 | 11 | 10 | | | 1 | 1 |
| 12 | 11 | 19 | 16 | 28 | 11 | 14.3 | 11 | | | 1 | 1 |
| 12 | 11 | 20 | 16 | 28 | 6 | 12 | 9 | | | 1 | 1 |
| 12 | 11 | 21 | 15 | 26 | 10 | 16.3 | 11 | 1 | | | 1 |
| 12 | 11 | 22 | 14 | 26 | 9 | 14.3 | 11 | 1 | | | 1 |
| 12 | 11.3 | 21 | 14 | 23 | 11 | 14 | 10 | | | 1 | 1 |
| 12 | 12 | 14 | 15 | 25 | 11 | 13.3 | 9 | | 1 | | 1 |
| 12 | 12 | 15 | 14 | 28 | 8 | 15.3 | 11 | 1 | | | 1 |
| 12 | 12 | 20 | 12 | 26 | 10 | 15.3 | 9 | | | 1 | 1 |
| 12 | 12 | 20 | 12 | 28 | 6 | 10 | 10 | 1 | | | 1 |
| 12 | 12 | 20 | 13 | 26 | 9 | 15.3 | 11 | 1 | | | 1 |
| 12 | 12 | 20 | 13 | 30 | 12 | 14 | 11 | 1 | | | 1 |
| 12 | 12 | 20 | 16 | 19 | 10 | 16.3 | 10 | | | 1 | 1 |
| 12 | 12 | 21 | 15 | 23 | 8 | 16.3 | 11 | | | 1 | 1 |
| 12 | 12 | 22 | 15 | 23 | 10 | 14.3 | 11 | | | 1 | 1 |
| 12 | 12 | 22 | 16 | 26 | 9 | 12 | 10 | 1 | | | 1 |
| 12 | 12 | 23 | 14 | 21 | 9 | 11 | 10 | 1 | | | 1 |
| 12 | 12.3 | 21 | 16 | 24 | 8 | 16.3 | 10 | 1 | | | 1 |
| 12 | 12.3 | 22 | 14 | 26 | 10 | 12 | 10 | | | 1 | 1 |
| 12 | 13 | 20 | 16 | 28 | 12 | 15.3 | 12 | | 1 | | 1 |
| 12 | 13 | 21 | 12 | 26 | 11 | 15.3 | 10 | | 1 | | 1 |
| 12 | 13 | 21 | 16 | 24 | 12 | 14.3 | 9 | | 1 | | 1 |
| 12 | 13.3 | 20 | 16 | 25 | 6 | 12 | 10 | | 1 | | 1 |
| 12 | 13.3 | 21 | 14 | 24 | 11 | 12 | 10 | | | 1 | 1 |
| 12 | 13.3 | 21 | 16 | 21 | 9 | 14.3 | 10 | | | 1 | 1 |
| 12 | 14 | 15 | 15 | 26 | 11 | 12 | 12 | 1 | | | 1 |
| 12 | 15.3 | 20 | 16 | 24 | 6 | 15.3 | 10 | | 1 | | 1 |
| 12 | 15.3 | 20 | 16 | 25 | 11 | 15.3 | 11 | | 1 | | 1 |
| 13 | 10 | 16 | 13 | 27 | 9 | 11 | 10 | 1 | | | 1 |
| 13 | 10 | 20 | 13 | 27 | 9 | 15.3 | 11 | 1 | | | 1 |
| 13 | 10 | 21 | 14 | 25 | 9 | 14.3 | 10 | 1 | | | 1 |
| 13 | 11 | 14 | 16 | 24 | 8 | 14.3 | 10 | | | 1 | 1 |
| 13 | 11 | 15 | 13 | 19 | 11 | 12 | 11 | 1 | | | 1 |
| 13 | 11 | 15 | 13 | 29 | 8 | 13.3 | 11 | 1 | | | 1 |
| 13 | 11 | 16 | 14 | 29 | 10 | 15.3 | 11 | 1 | | | 1 |
| 13 | 11 | 16 | 15 | 21 | 11 | 14.3 | 11 | 1 | | | 1 |
| 13 | 11 | 20 | 12 | 24 | 9 | 15.3 | 9 | | | 1 | 1 |
| 13 | 11 | 20 | 15 | 24 | 8 | 12 | 10 | | | 1 | 1 |
| 13 | 11 | 20 | 16 | 21 | 10 | 16.3 | 9 | | | 1 | 1 |
| 13 | 11 | 20 | 16 | 22 | 12 | 16.3 | 10 | | 1 | | 1 |
| 13 | 11 | 20 | 16 | 23 | 6 | 15.3 | 10 | | 1 | | 1 |
| 13 | 11 | 20 | 16 | 25 | 12 | 15.3 | 10 | | | 1 | 1 |
| 13 | 11 | 20 | 17 | 18 | 12 | 16.3 | 10 | | 1 | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=108 | CN N=165 | Hisp N=150 | Overall N=423 |
|---|---|---|---|---|---|---|---|---|---|---|---|
| 13 | 11 | 21 | 14 | 24 | 6 | 14.3 | 10 | | | 1 | 1 |
| 13 | 11 | 21 | 15 | 24 | 9 | 13 | 10 | | | 1 | 1 |
| 13 | 11 | 21 | 15 | 26 | 10 | 15.3 | 12 | | | 1 | 1 |
| 13 | 11 | 21 | 16 | 19 | 7 | 12 | 11 | 1 | | | 1 |
| 13 | 11 | 21 | 16 | 23 | 10 | 12 | 10 | | | 1 | 1 |
| 13 | 11 | 22 | 13 | 26 | 8 | 15.3 | 9 | 1 | | | 1 |
| 13 | 11 | 22 | 14 | 21 | 11 | 16.3 | 12 | 1 | | | 1 |
| 13 | 11 | 22 | 14 | 28 | 10 | 14.3 | 11 | | 1 | | 1 |
| 13 | 11 | 22 | 15 | 21 | 7 | 14.3 | 9 | 1 | | | 1 |
| 13 | 11 | 22 | 15 | 24 | 10 | 15.3 | 9 | | 1 | | 1 |
| 13 | 11 | 22 | 16 | 25 | 11 | 13 | 10 | | 1 | | 1 |
| 13 | 11 | 22 | 16 | 28 | 12 | 13 | 10 | | | 1 | 1 |
| 13 | 11 | 22 | 17 | 24 | 8 | 16.3 | 10 | | 1 | | 1 |
| 13 | 11.3 | 16 | 17 | 26 | 11 | 15.3 | 11 | | | 1 | 1 |
| 13 | 11.3 | 20 | 14 | 24 | 11 | 16.3 | 10 | | | 1 | 1 |
| 13 | 11.3 | 20 | 14 | 26 | 6 | 12 | 10 | | | 1 | 1 |
| 13 | 11.3 | 23 | 13 | 26 | 10 | 16.3 | 9 | | | 1 | 1 |
| 13 | 12 | 15 | 13 | 29 | 11 | 14.3 | 12 | 1 | | | 1 |
| 13 | 12 | 15 | 14 | 24 | 8 | 12 | 10 | | | 1 | 1 |
| 13 | 12 | 19 | 14 | 24 | 12 | 12 | 10 | | 1 | | 1 |
| 13 | 12 | 20 | 13 | 25 | 10 | 12 | 10 | | | 1 | 1 |
| 13 | 12 | 20 | 14 | 24 | 10 | 15.3 | 11 | | 1 | | 1 |
| 13 | 12 | 20 | 14 | 25 | 12 | 12 | 9 | | | 1 | 1 |
| 13 | 12 | 20 | 15 | 18 | 10 | 14.3 | 10 | | | 1 | 1 |
| 13 | 12 | 20 | 15 | 21 | 11 | 14.3 | 11 | | 1 | | 1 |
| 13 | 12 | 20 | 15 | 24 | 8 | 14.3 | 11 | | 1 | | 1 |
| 13 | 12 | 20 | 15 | 26 | 6 | 11 | 9 | | 1 | | 1 |
| 13 | 12 | 20 | 15 | 27 | 10 | 15.3 | 10 | | 1 | | 1 |
| 13 | 12 | 20 | 16 | 19 | 10 | 15.3 | 11 | | | 1 | 1 |
| 13 | 12 | 20 | 16 | 25 | 11 | 12 | 10 | | | 1 | 1 |
| 13 | 12 | 20 | 16 | 25 | 12 | 12 | 10 | | | 1 | 1 |
| 13 | 12 | 20 | 17 | 20 | 12 | 12 | 11 | | | 1 | 1 |
| 13 | 12 | 21 | 13 | 19 | 9 | 13 | 11 | 1 | | | 1 |
| 13 | 12 | 21 | 13 | 20 | 8 | 15.3 | 11 | 1 | | | 1 |
| 13 | 12 | 21 | 13 | 24 | 10 | 15.3 | 9 | | | 1 | 1 |
| 13 | 12 | 21 | 14 | 21 | 6 | 14.3 | 11 | 1 | | | 1 |
| 13 | 12 | 21 | 14 | 24 | 8 | 15.3 | 10 | | 1 | | 1 |
| 13 | 12 | 21 | 15 | 23 | 6 | 15.3 | 9 | | 1 | | 1 |
| 13 | 12 | 21 | 16 | 25 | 11 | 14.3 | 11 | | 1 | | 1 |
| 13 | 12 | 21 | 17 | 24 | 11 | 12 | 11 | | 1 | | 1 |
| 13 | 12 | 21 | 17 | 25 | 10 | 12 | 10 | | | 1 | 1 |
| 13 | 12 | 21 | 17 | 26 | 11 | 14.3 | 11 | | | 1 | 1 |
| 13 | 12 | 22 | 12 | 19 | 9 | 12 | 10 | 1 | | | 1 |
| 13 | 12 | 22 | 13 | 26 | 11 | 15.3 | 9 | | | 1 | 1 |
| 13 | 12 | 22 | 15 | 27 | 11 | 15.3 | 9 | | 1 | | 1 |
| 13 | 12 | 22 | 16 | 19 | 11 | 10 | 10 | | 1 | | 1 |
| 13 | 12 | 22 | 17 | 25 | 12 | 15.3 | 10 | | 1 | | 1 |
| 13 | 12 | 23 | 15 | 26 | 11 | 14.3 | 12 | | 1 | | 1 |
| 13 | 12 | 23 | 16 | 25 | 6 | 14.3 | 10 | | 1 | | 1 |
| 13 | 12.3 | 16 | 13 | 25 | 11 | 12 | 10 | | | 1 | 1 |
| 13 | 12.3 | 19 | 16 | 18 | 8 | 15.3 | 9 | | | 1 | 1 |
| 13 | 12.3 | 20 | 13 | 26 | 6 | 12 | 9 | | | 1 | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=108 | CN N=165 | Hisp N=150 | Overall N=423 |
|---|---|---|---|---|---|---|---|---|---|---|---|
| 13 | 12.3 | 20 | 14 | 22 | 8 | 15.3 | 11 | | | 1 | 1 |
| 13 | 12.3 | 20 | 14 | 28 | 10 | 16.3 | 9 | | 1 | | 1 |
| 13 | 12.3 | 20 | 16 | 24 | 8 | 16.3 | 11 | | 1 | | 1 |
| 13 | 12.3 | 20 | 18 | 24 | 6 | 16.3 | 10 | | | 1 | 1 |
| 13 | 12.3 | 21 | 15 | 27 | 11 | 12 | 10 | | 1 | | 1 |
| 13 | 12.3 | 21 | 17 | 25 | 6 | 16.3 | 11 | | | 1 | 1 |
| 13 | 12.3 | 22 | 14 | 23 | 8 | 16.3 | 11 | | | 1 | 1 |
| 13 | 12.3 | 23 | 15 | 24 | 10 | 14.3 | 9 | | 1 | | 1 |
| 13 | 12.3 | 23 | 16 | 20 | 8 | 12 | 11 | | 1 | | 1 |
| 13 | 13 | 20 | 16 | 26 | 8 | 15.3 | 10 | | 1 | | 1 |
| 13 | 13 | 20 | 17 | 25 | 12 | 13.3 | 10 | | 1 | | 1 |
| 13 | 13 | 21 | 12 | 28 | 10 | 15.3 | 9 | | 1 | | 1 |
| 13 | 13 | 21 | 14 | 18 | 9 | 15.3 | 11 | 1 | | | 1 |
| 13 | 13 | 21 | 14 | 20 | 10 | 12 | 10 | 1 | | | 1 |
| 13 | 13 | 21 | 15 | 24 | 10 | 15.3 | 9 | | 1 | | 1 |
| 13 | 13 | 21 | 16 | 24 | 6 | 13 | 9 | | 1 | | 1 |
| 13 | 13 | 21 | 16 | 26 | 9 | 15.3 | 10 | | 1 | | 1 |
| 13 | 13 | 22 | 17 | 24 | 9 | 13.3 | 12 | | 1 | | 1 |
| 13 | 13 | 23 | 16 | 26 | 6 | 15.3 | 9 | | 1 | | 1 |
| 13 | 13.3 | 15 | 17 | 18 | 12 | 15.3 | 10 | | 1 | | 1 |
| 13 | 13.3 | 20 | 15 | 25 | 11 | 16.3 | 11 | | 1 | | 1 |
| 13 | 13.3 | 20 | 16 | 19 | 11 | 14.3 | 9 | | | 1 | 1 |
| 13 | 13.3 | 21 | 13 | 26 | 10 | 12 | 10 | | | 1 | 1 |
| 13 | 13.3 | 21 | 15 | 23 | 6 | 15.3 | 11 | | 1 | | 1 |
| 13 | 13.3 | 21 | 15 | 26 | 10 | 15.3 | 10 | | | 1 | 1 |
| 13 | 13.3 | 21 | 16 | 15 | 11 | 15.3 | 9 | | 1 | | 1 |
| 13 | 13.3 | 21 | 18 | 24 | 10 | 13 | 11 | | | 1 | 1 |
| 13 | 13.3 | 22 | 14 | 26 | 10 | 15.3 | 11 | | 1 | | 1 |
| 13 | 13.3 | 22 | 15 | 25 | 9 | 16.3 | 9 | | 1 | | 1 |
| 13 | 13.3 | 23 | 12 | 24 | 10 | 14.3 | 10 | | | 1 | 1 |
| 13 | 13.3 | 23 | 16 | 24 | 10 | 14.3 | 11 | | 1 | | 1 |
| 13 | 14 | 20 | 13 | 23 | 10 | 9 | 12 | 1 | | | 1 |
| 13 | 14.3 | 22 | 17 | 24 | 8 | 15.3 | 10 | | 1 | | 1 |
| 14 | 8 | 19 | 11 | 21 | 9 | 13 | 10 | 1 | | | 1 |
| 14 | 9 | 20 | 14 | 25 | 10 | 15.3 | 9 | | | 1 | 1 |
| 14 | 9 | 21 | 16 | 26 | 10 | 15.3 | 11 | 1 | | | 1 |
| 14 | 10 | 15 | 11 | 21 | 9 | 13 | 10 | 1 | | | 1 |
| 14 | 10 | 18 | 11 | 21 | 6 | 15.3 | 10 | 1 | | | 1 |
| 14 | 10 | 20 | 16 | 24 | 10 | 12 | 8 | | 1 | | 1 |
| 14 | 10 | 21 | 14 | 24 | 10 | 13.3 | 9 | | 1 | | 1 |
| 14 | 10 | 21 | 15 | 18 | 6 | 14.3 | 9 | | 1 | | 1 |
| 14 | 10 | 21 | 17 | 21 | 7 | 14 | 11 | 1 | | | 1 |
| 14 | 10 | 23 | 14 | 27 | 11 | 15.3 | 10 | | | 1 | 1 |
| 14 | 11 | 15 | 12 | 15 | 10 | 15.3 | 11 | | 1 | | 1 |
| 14 | 11 | 15 | 14 | 25 | 8 | 13.3 | 10 | | 1 | | 1 |
| 14 | 11 | 15 | 14 | 25 | 9 | 12 | 9 | 1 | | | 1 |
| 14 | 11 | 15 | 15 | 21 | 10 | 14.3 | 10 | 1 | | | 1 |
| 14 | 11 | 15 | 15 | 23 | 9 | 15.3 | 10 | 1 | | | 1 |
| 14 | 11 | 15 | 16 | 22 | 9 | 12 | 11 | 1 | | | 1 |
| 14 | 11 | 15 | 17 | 18 | 12 | 13.3 | 11 | | 1 | | 1 |
| 14 | 11 | 16 | 13 | 25 | 9 | 12 | 10 | 1 | | | 1 |
| 14 | 11 | 16 | 13 | 27 | 6 | 15.3 | 12 | 1 | | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=108 | CN N=165 | Hisp N=150 | Overall N=423 |
|---|---|---|---|---|---|---|---|---|---|---|---|
| 14 | 11 | 16 | 15 | 27 | 9 | 13 | 11 | 1 | | | 1 |
| 14 | 11 | 16 | 17 | 26 | 9 | 15.3 | 8 | 1 | | | 1 |
| 14 | 11 | 19 | 13 | 26 | 11 | 16.3 | 10 | | | 1 | 1 |
| 14 | 11 | 19 | 14 | 25 | 9 | 12 | 11 | | 1 | | 1 |
| 14 | 11 | 20 | 13 | 26 | 8 | 12 | 10 | | | 1 | 1 |
| 14 | 11 | 20 | 14 | 22 | 11 | 14.3 | 11 | 1 | | | 1 |
| 14 | 11 | 20 | 14 | 24 | 8 | 14.3 | 10 | | | 1 | 1 |
| 14 | 11 | 20 | 14 | 26 | 8 | 13 | 10 | 1 | | | 1 |
| 14 | 11 | 20 | 14 | 27 | 6 | 11 | 11 | | 1 | | 1 |
| 14 | 11 | 20 | 16 | 19 | 11 | 12 | 11 | 1 | | | 1 |
| 14 | 11 | 20 | 16 | 25 | 10 | 10 | 10 | | 1 | | 1 |
| 14 | 11 | 20 | 16 | 25 | 11 | 13 | 9 | 1 | | | 1 |
| 14 | 11 | 20 | 16 | 26 | 6 | 15.3 | 10 | 1 | | | 1 |
| 14 | 11 | 20 | 17 | 19 | 10 | 12 | 11 | | 1 | | 1 |
| 14 | 11 | 21 | 13 | 19 | 6 | 13 | 11 | 1 | | | 1 |
| 14 | 11 | 21 | 13 | 23 | 10 | 14.3 | 10 | 1 | | | 1 |
| 14 | 11 | 21 | 14 | 18 | 11 | 14 | 10 | | | 1 | 1 |
| 14 | 11 | 21 | 14 | 22 | 7 | 14.3 | 10 | 1 | | | 1 |
| 14 | 11 | 21 | 14 | 24 | 11 | 12 | 9 | | | 1 | 1 |
| 14 | 11 | 21 | 14 | 25 | 9 | 15.3 | 10 | | | 1 | 1 |
| 14 | 11 | 21 | 14 | 27 | 12 | 13 | 10 | | 1 | | 1 |
| 14 | 11 | 21 | 15 | 21 | 8 | 13 | 11 | 1 | | | 1 |
| 14 | 11 | 21 | 15 | 25 | 10 | 16.3 | 10 | | 1 | | 1 |
| 14 | 11 | 21 | 16 | 24 | 12 | 16.3 | 9 | | 1 | | 1 |
| 14 | 11 | 21 | 16 | 27 | 10 | 16.3 | 8 | 1 | | | 1 |
| 14 | 11 | 21 | 18 | 24 | 11 | 11 | 10 | | | 1 | 1 |
| 14 | 11 | 21 | 19 | 25 | 11 | 15.3 | 11 | | | 1 | 1 |
| 14 | 11 | 22 | 16 | 18 | 11 | 15.3 | 10 | 1 | | | 1 |
| 14 | 11 | 22 | 16 | 26 | 8 | 16.3 | 9 | | 1 | | 1 |
| 14 | 11.3 | 20 | 14 | 23 | 8 | 13 | 10 | 1 | | | 1 |
| 14 | 11.3 | 20 | 14 | 26 | 12 | 15.3 | 10 | | | 1 | 1 |
| 14 | 11.3 | 20 | 16 | 25 | 12 | 15.3 | 10 | | | 1 | 1 |
| 14 | 11.3 | 20 | 16 | 27 | 10 | 15.3 | 11 | | 1 | | 1 |
| 14 | 11.3 | 20 | 17 | 19 | 8 | 16.3 | 10 | | | 1 | 1 |
| 14 | 11.3 | 20 | 17 | 24 | 11 | 13.3 | 10 | | 1 | | 1 |
| 14 | 11.3 | 21 | 14 | 25 | 12 | 12 | 9 | | 1 | | 1 |
| 14 | 11.3 | 21 | 15 | 27 | 12 | 17.3 | 11 | | 1 | | 1 |
| 14 | 11.3 | 21 | 16 | 25 | 6 | 14.3 | 10 | | 1 | | 1 |
| 14 | 11.3 | 21 | 18 | 24 | 10 | 14.3 | 11 | | 1 | | 1 |
| 14 | 11.3 | 21 | 18 | 24 | 11 | 13 | 10 | | | 1 | 1 |
| 14 | 11.3 | 22 | 17 | 24 | 11 | 15.3 | 11 | | | 1 | 1 |
| 14 | 12 | 15 | 11 | 23 | 8 | 14 | 11 | 1 | | | 1 |
| 14 | 12 | 15 | 13 | 27 | 10 | 11 | 11 | 1 | | | 1 |
| 14 | 12 | 15 | 15 | 25 | 6 | 16.3 | 9 | | 1 | | 1 |
| 14 | 12 | 15 | 16 | 19 | 6 | 14.3 | 10 | | 1 | | 1 |
| 14 | 12 | 20 | 13 | 19 | 7 | 10 | 10 | 1 | | | 1 |
| 14 | 12 | 20 | 14 | 23 | 10 | 15.3 | 9 | | | 1 | 1 |
| 14 | 12 | 20 | 14 | 24 | 11 | 12 | 9 | | | 1 | 1 |
| 14 | 12 | 20 | 15 | 24 | 10 | 15.3 | 11 | | 1 | | 1 |
| 14 | 12 | 20 | 15 | 25 | 11 | 13 | 11 | | | 1 | 1 |
| 14 | 12 | 20 | 15 | 26 | 8 | 16.3 | 10 | | 1 | | 1 |
| 14 | 12 | 20 | 16 | 25 | 10 | 13.3 | 9 | | 1 | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=108 | CN N=165 | Hisp N=150 | Overall N=423 |
|---|---|---|---|---|---|---|---|---|---|---|---|
| 14 | 12 | 20 | 16 | 26 | 10 | 12 | 10 | | | 1 | 1 |
| 14 | 12 | 21 | 13 | 27 | 11 | 16.3 | 11 | | 1 | | 1 |
| 14 | 12 | 21 | 14 | 24 | 11 | 14.3 | 11 | | | 1 | 1 |
| 14 | 12 | 21 | 14 | 26 | 11 | 15.3 | 9 | | 1 | | 1 |
| 14 | 12 | 21 | 14 | 31 | 10 | 12 | 10 | | 1 | | 1 |
| 14 | 12 | 21 | 15 | 18 | 11 | 14.3 | 12 | 1 | | | 1 |
| 14 | 12 | 21 | 15 | 26 | 8 | 15.3 | 9 | | 1 | | 1 |
| 14 | 12 | 21 | 15 | 27 | 12 | 15.3 | 9 | | | 1 | 1 |
| 14 | 12 | 21 | 16 | 24 | 11 | 14.3 | 10 | | | 1 | 1 |
| 14 | 12 | 21 | 16 | 24 | 11 | 16.3 | 11 | | | 1 | 1 |
| 14 | 12 | 21 | 16 | 25 | 10 | 16.3 | 10 | | | 1 | 1 |
| 14 | 12 | 22 | 11 | 21 | 9 | 12 | 12 | 1 | | | 1 |
| 14 | 12 | 22 | 14 | 27 | 10 | 14.3 | 11 | | 1 | | 1 |
| 14 | 12 | 22 | 15 | 28 | 10 | 15.3 | 11 | | | 1 | 1 |
| 14 | 12 | 22 | 15 | 29 | 11 | 14.3 | 10 | | | 1 | 1 |
| 14 | 12 | 22 | 16 | 18 | 6 | 15.3 | 11 | | 1 | | 1 |
| 14 | 12 | 22 | 16 | 24 | 6 | 13 | 10 | | | 1 | 1 |
| 14 | 12 | 22 | 16 | 24 | 11 | 13 | 10 | | | 1 | 1 |
| 14 | 12 | 22 | 16 | 25 | 10 | 13 | 11 | | 1 | | 1 |
| 14 | 12 | 22 | 17 | 20 | 10 | 15.3 | 10 | | 1 | | 1 |
| 14 | 12 | 22 | 17 | 24 | 11 | 14.3 | 11 | | 1 | | 1 |
| 14 | 12 | 22 | 18 | 26 | 10 | 15.3 | 11 | | | 1 | 1 |
| 14 | 12 | 23 | 14 | 26 | 6 | 12 | 11 | | | 1 | 1 |
| 14 | 12 | 23 | 14 | 26 | 12 | 14.3 | 9 | | 1 | | 1 |
| 14 | 12 | 24 | 14 | 24 | 10 | 15.3 | 10 | | | 1 | 1 |
| 14 | 12.3 | 16 | 13 | 27 | 10 | 12 | 11 | 1 | | | 1 |
| 14 | 12.3 | 20 | 14 | 26 | 8 | 14.3 | 9 | | | 1 | 1 |
| 14 | 12.3 | 20 | 15 | 26 | 12 | 14.3 | 11 | | | 1 | 1 |
| 14 | 12.3 | 20 | 16 | 18 | 10 | 14.3 | 11 | | | 1 | 1 |
| 14 | 12.3 | 20 | 16 | 24 | 6 | 12 | 10 | 1 | | | 1 |
| 14 | 12.3 | 20 | 16 | 24 | 12 | 15.3 | 10 | 1 | | | 1 |
| 14 | 12.3 | 20 | 16 | 26 | 6 | 12 | 10 | | 1 | | 1 |
| 14 | 12.3 | 20 | 16 | 26 | 8 | 15.3 | 10 | | | 1 | 1 |
| 14 | 12.3 | 20 | 17 | 18 | 8 | 15.3 | 9 | | | 1 | 1 |
| 14 | 12.3 | 21 | 14 | 27 | 6 | 13 | 10 | | 1 | | 1 |
| 14 | 12.3 | 21 | 14 | 28 | 11 | 12 | 10 | | 1 | | 1 |
| 14 | 12.3 | 21 | 15 | 24 | 6 | 14.3 | 10 | | 1 | | 1 |
| 14 | 12.3 | 21 | 15 | 25 | 12 | 16.3 | 11 | | | 1 | 1 |
| 14 | 12.3 | 21 | 16 | 20 | 6 | 15.3 | 10 | | | 1 | 1 |
| 14 | 12.3 | 22 | 12 | 24 | 10 | 15.3 | 10 | | | 1 | 1 |
| 14 | 12.3 | 22 | 16 | 25 | 8 | 15.3 | 11 | | 1 | | 1 |
| 14 | 12.3 | 22 | 17 | 18 | 10 | 15.3 | 10 | | | 1 | 1 |
| 14 | 12.3 | 22 | 17 | 23 | 8 | 15.3 | 11 | | 1 | | 1 |
| 14 | 12.3 | 23 | 16 | 24 | 10 | 14.3 | 9 | | | 1 | 1 |
| 14 | 12.3 | 24 | 15 | 24 | 6 | 12 | 9 | | 1 | | 1 |
| 14 | 13 | 15 | 13 | 25 | 7 | 13 | 12 | 1 | | | 1 |
| 14 | 13 | 16 | 11 | 27 | 6 | 13 | 11 | 1 | | | 1 |
| 14 | 13 | 16 | 17 | 18 | 10 | 14.3 | 10 | | | 1 | 1 |
| 14 | 13 | 20 | 13 | 25 | 6 | 15.3 | 11 | | 1 | | 1 |
| 14 | 13 | 20 | 15 | 24 | 11 | 12 | 9 | | 1 | | 1 |
| 14 | 13 | 20 | 15 | 25 | 6 | 15.3 | 11 | | 1 | | 1 |
| 14 | 13 | 20 | 15 | 25 | 10 | 14.3 | 11 | | | 1 | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=108 | CN N=165 | Hisp N=150 | Overall N=423 |
|---|---|---|---|---|---|---|---|---|---|---|---|
| 14 | 13 | 20 | 15 | 26 | 8 | 16.3 | 10 | | 1 | | 1 |
| 14 | 13 | 20 | 16 | 15 | 11 | 11 | 10 | | 1 | | 1 |
| 14 | 13 | 20 | 16 | 15 | 12 | 15.3 | 10 | | | 1 | 1 |
| 14 | 13 | 20 | 16 | 21 | 8 | 14.3 | 9 | | 1 | | 1 |
| 14 | 13 | 20 | 16 | 25 | 8 | 16.3 | 11 | | 1 | | 1 |
| 14 | 13 | 20 | 17 | 18 | 11 | 15.3 | 10 | | | 1 | 1 |
| 14 | 13 | 20 | 17 | 24 | 10 | 15.3 | 10 | | 1 | | 1 |
| 14 | 13 | 21 | 11 | 21 | 9 | 11 | 10 | 1 | | | 1 |
| 14 | 13 | 21 | 13 | 25 | 11 | 12 | 10 | 1 | | | 1 |
| 14 | 13 | 21 | 13 | 27 | 10 | 16.3 | 10 | | 1 | | 1 |
| 14 | 13 | 21 | 15 | 24 | 6 | 14.3 | 9 | | 1 | | 1 |
| 14 | 13 | 21 | 15 | 25 | 11 | 15.3 | 10 | | 1 | | 1 |
| 14 | 13 | 21 | 17 | 18 | 12 | 16.3 | 12 | | 1 | | 1 |
| 14 | 13 | 21 | 18 | 25 | 8 | 15.3 | 9 | | | 1 | 1 |
| 14 | 13 | 22 | 14 | 19 | 8 | 13 | 10 | | 1 | | 1 |
| 14 | 13 | 22 | 17 | 18 | 10 | 13.3 | 10 | | 1 | | 1 |
| 14 | 13 | 23 | 14 | 28 | 6 | 14.3 | 11 | | | 1 | 1 |
| 14 | 13 | 23 | 17 | 28 | 11 | 15.3 | 11 | | 1 | | 1 |
| 14 | 13.3 | 19 | 13 | 25 | 8 | 14 | 12 | | 1 | | 1 |
| 14 | 13.3 | 20 | 12 | 30 | 12 | 15.3 | 10 | | 1 | | 1 |
| 14 | 13.3 | 20 | 14 | 27 | 10 | 15.3 | 10 | | 1 | | 1 |
| 14 | 13.3 | 20 | 15 | 23 | 10 | 15.3 | 11 | | 1 | | 1 |
| 14 | 13.3 | 20 | 15 | 26 | 10 | 11 | 9 | | 1 | | 1 |
| 14 | 13.3 | 20 | 16 | 27 | 12 | 15.3 | 10 | | | 1 | 1 |
| 14 | 13.3 | 21 | 13 | 25 | 11 | 15.3 | 10 | | 1 | | 1 |
| 14 | 13.3 | 21 | 14 | 26 | 12 | 15.3 | 11 | | 1 | | 1 |
| 14 | 13.3 | 22 | 14 | 29 | 11 | 16.3 | 9 | | 1 | | 1 |
| 14 | 13.3 | 22 | 16 | 30 | 6 | 15.3 | 11 | | 1 | | 1 |
| 14 | 14.3 | 21 | 14 | 24 | 6 | 15.3 | 10 | | | 1 | 1 |
| 14 | 15.3 | 20 | 13 | 25 | 10 | 12 | 11 | | 1 | | 1 |
| 15 | 8 | 18 | 15 | 20 | 9 | 11 | 11 | 1 | | | 1 |
| 15 | 9 | 15 | 13 | 19 | 7 | 11 | 11 | 1 | | | 1 |
| 15 | 9 | 21 | 13 | 26 | 9 | 10 | 12 | 1 | | | 1 |
| 15 | 9.3 | 20 | 13 | 26 | 8 | 12 | 10 | | 1 | | 1 |
| 15 | 10 | 16 | 13 | 21 | 9 | 12 | 11 | 1 | | | 1 |
| 15 | 10 | 16 | 14 | 16 | 11 | 15.3 | 10 | 1 | | | 1 |
| 15 | 10 | 20 | 14 | 24 | 8 | 13.3 | 9 | | 1 | | 1 |
| 15 | 10 | 20 | 16 | 18 | 11 | 16.3 | 11 | | 1 | | 1 |
| 15 | 10 | 23 | 13 | 19 | 9 | 13 | 11 | 1 | | | 1 |
| 15 | 10 | 23 | 13 | 19 | 11 | 11 | 10 | | | 1 | 1 |
| 15 | 11 | 14 | 15 | 24 | 11 | 15.3 | 9 | | 1 | | 1 |
| 15 | 11 | 15 | 11 | 23 | 10 | 16.3 | 10 | | | 1 | 1 |
| 15 | 11 | 15 | 13 | 19 | 10 | 13 | 10 | | | 1 | 1 |
| 15 | 11 | 15 | 14 | 19 | 11 | 11 | 11 | 1 | | | 1 |
| 15 | 11 | 16 | 14 | 20 | 9 | 11 | 11 | 1 | | | 1 |
| 15 | 11 | 19 | 9 | 27 | 9 | 14 | 10 | 1 | | | 1 |
| 15 | 11 | 19 | 16 | 24 | 10 | 14.3 | 9 | | | 1 | 1 |
| 15 | 11 | 19 | 18 | 25 | 10 | 15.3 | 10 | | 1 | | 1 |
| 15 | 11 | 20 | 13 | 24 | 10 | 15.3 | 11 | | | 1 | 1 |
| 15 | 11 | 20 | 13 | 25 | 11 | 14.3 | 11 | | | 1 | 1 |
| 15 | 11 | 20 | 13 | 25 | 11 | 15.3 | 9 | | 1 | | 1 |
| 15 | 11 | 20 | 15 | 27 | 11 | 13.3 | 11 | | 1 | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=108 | CN N=165 | Hisp N=150 | Overall N=423 |
|---|---|---|---|---|---|---|---|---|---|---|---|
| 15 | 11 | 20 | 16 | 25 | 10 | 15.3 | 11 | 1 | | | 1 |
| 15 | 11 | 20 | 17 | 18 | 12 | 11 | 11 | | 1 | | 1 |
| 15 | 11 | 20 | 17 | 24 | 8 | 12 | 9 | | | 1 | 1 |
| 15 | 11 | 21 | 11 | 23 | 12 | 15.3 | 10 | | 1 | | 1 |
| 15 | 11 | 21 | 12 | 25 | 8 | 15.3 | 9 | 1 | | | 1 |
| 15 | 11 | 21 | 13 | 25 | 6 | 15.3 | 10 | | | 1 | 1 |
| 15 | 11 | 21 | 13 | 26 | 12 | 12 | 9 | | | 1 | 1 |
| 15 | 11 | 21 | 13 | 27 | 8 | 12 | 11 | 1 | | | 1 |
| 15 | 11 | 21 | 13 | 27 | 12 | 15.3 | 10 | 1 | | | 1 |
| 15 | 11 | 21 | 13 | 28 | 10 | 13 | 10 | 1 | | | 1 |
| 15 | 11 | 21 | 15 | 27 | 12 | 14.3 | 11 | | 1 | | 1 |
| 15 | 11 | 21 | 16 | 18 | 8 | 15.3 | 9 | | 1 | | 1 |
| 15 | 11 | 21 | 16 | 23 | 10 | 14.3 | 11 | | | 1 | 1 |
| 15 | 11 | 21 | 17 | 23 | 10 | 13.3 | 11 | | | 1 | 1 |
| 15 | 11 | 21 | 18 | 23 | 9 | 15.3 | 10 | | | 1 | 1 |
| 15 | 11 | 21 | 18 | 24 | 12 | 12 | 11 | | | 1 | 1 |
| 15 | 11 | 22 | 13 | 19 | 9 | 15.3 | 11 | 1 | | | 1 |
| 15 | 11 | 22 | 13 | 25 | 10 | 15.3 | 11 | | | 1 | 1 |
| 15 | 11 | 22 | 14 | 25 | 12 | 15.3 | 9 | 1 | | | 1 |
| 15 | 11 | 22 | 16 | 22 | 10 | 15.3 | 9 | | 1 | | 1 |
| 15 | 11 | 22 | 16 | 22 | 11 | 15.3 | 9 | | | 1 | 1 |
| 15 | 11 | 23 | 16 | 28 | 11 | 12 | 10 | | | 1 | 1 |
| 15 | 11 | 23 | 17 | 27 | 12 | 12 | 10 | | | 1 | 1 |
| 15 | 11 | 24 | 14 | 31 | 6 | 15.3 | 10 | | | 1 | 1 |
| 15 | 11.3 | 20 | 16 | 21 | 9 | 15.3 | 9 | | | 1 | 1 |
| 15 | 11.3 | 22 | 16 | 24 | 7 | 15 | 10 | | | 1 | 1 |
| 15 | 12 | 14 | 15 | 26 | 10 | 14.3 | 10 | | | 1 | 1 |
| 15 | 12 | 15 | 13 | 24 | 6 | 13 | 10 | 1 | | | 1 |
| 15 | 12 | 15 | 13 | 29 | 10 | 10 | 8 | 1 | | | 1 |
| 15 | 12 | 15 | 14 | 27 | 9 | 16.3 | 11 | 1 | | | 1 |
| 15 | 12 | 15 | 18 | 24 | 10 | 15.3 | 10 | | | 1 | 1 |
| 15 | 12 | 16 | 13 | 19 | 7 | 15.3 | 12 | 1 | | | 1 |
| 15 | 12 | 16 | 16 | 25 | 11 | 11 | 9 | | | 1 | 1 |
| 15 | 12 | 20 | 13 | 18 | 8 | 15.3 | 10 | | 1 | | 1 |
| 15 | 12 | 20 | 13 | 25 | 10 | 15.3 | 9 | | | 1 | 1 |
| 15 | 12 | 20 | 14 | 24 | 10 | 12 | 10 | | | 1 | 1 |
| 15 | 12 | 20 | 14 | 26 | 10 | 15.3 | 10 | | | 1 | 1 |
| 15 | 12 | 20 | 14 | 27 | 9 | 12 | 12 | 1 | | | 1 |
| 15 | 12 | 20 | 15 | 27 | 6 | 12 | 11 | | 1 | | 1 |
| 15 | 12 | 20 | 16 | 25 | 10 | 15.3 | 10 | | 1 | | 1 |
| 15 | 12 | 20 | 16 | 25 | 11 | 14.3 | 10 | | | 1 | 1 |
| 15 | 12 | 20 | 16 | 25 | 12 | 15.3 | 9 | | 1 | | 1 |
| 15 | 12 | 21 | 14 | 21 | 6 | 12 | 10 | 1 | | | 1 |
| 15 | 12 | 21 | 14 | 24 | 8 | 14.3 | 12 | | 1 | | 1 |
| 15 | 12 | 21 | 15 | 21 | 9 | 17 | 10 | 1 | | | 1 |
| 15 | 12 | 21 | 16 | 22 | 9 | 17.3 | 11 | 1 | | | 1 |
| 15 | 12 | 21 | 16 | 24 | 12 | 16.3 | 9 | | 1 | | 1 |
| 15 | 12 | 22 | 13 | 24 | 10 | 15.3 | 10 | | | 1 | 1 |
| 15 | 12 | 22 | 15 | 26 | 11 | 15.3 | 10 | | | 1 | 1 |
| 15 | 12 | 22 | 16 | 27 | 10 | 15.3 | 10 | | 1 | | 1 |
| 15 | 12 | 23 | 14 | 26 | 11 | 15.3 | 9 | | 1 | | 1 |
| 15 | 12 | 24 | 17 | 25 | 8 | 15.3 | 10 | | 1 | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=108 | CN N=165 | Hisp N=150 | Overall N=423 |
|---|---|---|---|---|---|---|---|---|---|---|---|
| 15 | 12.3 | 16 | 13 | 25 | 11 | 12 | 10 | | | 1 | 1 |
| 15 | 12.3 | 19 | 14 | 24 | 11 | 16.3 | 10 | | 1 | | 1 |
| 15 | 12.3 | 19 | 17 | 24 | 11 | 15.3 | 9 | | | 1 | 1 |
| 15 | 12.3 | 20 | 10 | 26 | 12 | 15.3 | 11 | | | 1 | 1 |
| 15 | 12.3 | 20 | 13 | 25 | 8 | 15.3 | 10 | | 1 | | 1 |
| 15 | 12.3 | 20 | 14 | 24 | 11 | 14.3 | 10 | 1 | | | 1 |
| 15 | 12.3 | 20 | 14 | 24 | 11 | 15.3 | 10 | | | 1 | 1 |
| 15 | 12.3 | 20 | 16 | 22 | 10 | 14.3 | 11 | | 1 | | 1 |
| 15 | 12.3 | 20 | 16 | 26 | 11 | 11 | 11 | | | 1 | 1 |
| 15 | 12.3 | 21 | 13 | 24 | 10 | 14.3 | 10 | | | 1 | 1 |
| 15 | 12.3 | 21 | 14 | 28 | 11 | 16.3 | 10 | | 1 | | 1 |
| 15 | 12.3 | 21 | 16 | 18 | 11 | 15.3 | 11 | | | 1 | 1 |
| 15 | 12.3 | 21 | 16 | 25 | 11 | 15.3 | 10 | | 1 | | 1 |
| 15 | 12.3 | 22 | 15 | 26 | 10 | 14.3 | 10 | | | 1 | 1 |
| 15 | 12.3 | 24 | 14 | 27 | 10 | 14.3 | 11 | | 1 | | 1 |
| 15 | 13 | 16 | 11 | 27 | 8 | 17.3 | 11 | 1 | | | 1 |
| 15 | 13 | 20 | 16 | 24 | 8 | 14.3 | 11 | | | 1 | 1 |
| 15 | 13 | 20 | 16 | 24 | 8 | 15.3 | 10 | | 1 | | 1 |
| 15 | 13 | 20 | 17 | 25 | 10 | 15.3 | 10 | | 1 | | 1 |
| 15 | 13 | 21 | 15 | 26 | 12 | 15.3 | 11 | | 1 | | 1 |
| 15 | 13 | 21 | 16 | 25 | 12 | 15.3 | 10 | | 1 | | 1 |
| 15 | 13 | 22 | 14 | 22 | 11 | 13.3 | 11 | | 1 | | 1 |
| 15 | 13 | 22 | 15 | 26 | 6 | 15.3 | 9 | | | 1 | 1 |
| 15 | 13.3 | 17 | 14 | 28 | 11 | 14.3 | 10 | | | 1 | 1 |
| 15 | 13.3 | 20 | 10 | 26 | 8 | 14 | 11 | | | 1 | 1 |
| 15 | 13.3 | 20 | 16 | 18 | 6 | 12 | 9 | | | 1 | 1 |
| 15 | 13.3 | 21 | 14 | 28 | 8 | 14.3 | 10 | | 1 | | 1 |
| 15 | 13.3 | 21 | 15 | 27 | 10 | 15.3 | 10 | | | 1 | 1 |
| 15 | 13.3 | 22 | 12 | 26 | 10 | 17.3 | 9 | | 1 | | 1 |
| 15 | 13.3 | 23 | 15 | 23 | 6 | 15.3 | 10 | | 1 | | 1 |
| 15 | 14 | 20 | 11 | 21 | 6 | 12 | 9 | 1 | | | 1 |
| 16 | 10 | 22 | 16 | 27 | 11 | 17.3 | 9 | | 1 | | 1 |
| 16 | 11 | 15 | 16 | 29 | 8 | 15.3 | 11 | 1 | | | 1 |
| 16 | 11 | 20 | 14 | 24 | 10 | 15.3 | 10 | | 1 | | 1 |
| 16 | 11 | 20 | 15 | 22 | 10 | 14 | 10 | | | 1 | 1 |
| 16 | 11 | 21 | 14 | 28 | 9 | 15.3 | 11 | | 1 | | 1 |
| 16 | 11 | 23 | 16 | 19 | 6 | 12 | 10 | 1 | | | 1 |
| 16 | 11.3 | 21 | 17 | 23 | 8 | 14.3 | 11 | | | 1 | 1 |
| 16 | 12 | 15 | 14 | 29 | 10 | 14.3 | 10 | | 1 | | 1 |
| 16 | 12 | 16 | 17 | 28 | 8 | 14.3 | 11 | | | 1 | 1 |
| 16 | 12 | 20 | 14 | 25 | 12 | 15.3 | 10 | | 1 | | 1 |
| 16 | 12 | 20 | 16 | 24 | 6 | 10 | 9 | | | 1 | 1 |
| 16 | 12 | 20 | 16 | 25 | 10 | 14.3 | 10 | | | 1 | 1 |
| 16 | 12.3 | 16 | 14 | 24 | 8 | 14.3 | 11 | 1 | | | 1 |
| 16 | 12.3 | 20 | 17 | 25 | 10 | 15.3 | 11 | | 1 | | 1 |
| 16 | 13 | 20 | 14 | 29 | 11 | 15.3 | 11 | | 1 | | 1 |
| 16 | 13 | 22 | 13 | 21 | 9 | 10 | 12 | 1 | | | 1 |
| 16 | 13 | 22 | 15 | 24 | 8 | 15.3 | 11 | | 1 | | 1 |
| 16 | 13.3 | 21 | 14 | 27 | 11 | 15.3 | 10 | | 1 | | 1 |
| 16 | 13.3 | 21 | 15 | 24 | 12 | 16.3 | 11 | 1 | | | 1 |
| 17 | 11 | 15 | 12 | 18 | 9 | 13 | 11 | 1 | | | 1 |
| 17 | 11 | 20 | 15 | 20 | 10 | 16.3 | 11 | | | 1 | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=108 | CN N=165 | Hisp N=150 | Overall N=423 |
|---|---|---|---|---|---|---|---|---|---|---|---|
| 17 | 11 | 23 | 12 | 25 | 10 | 12 | 11 | | 1 | | 1 |
| 17 | 12 | 20 | 14 | 25 | 11 | 16.3 | 9 | | | 1 | 1 |
| 17 | 12.3 | 20 | 13 | 25 | 10 | 15.3 | 10 | | | 1 | 1 |
| 17 | 13 | 21 | 11 | 23 | 8 | 12 | 10 | 1 | | | 1 |
| 17 | 13.3 | 21 | 15 | 19 | 8 | 16.3 | 11 | | 1 | | 1 |
| 18 | 12 | 15 | 14 | 24 | 10 | 16.3 | 10 | | 1 | | 1 |
| | | | | | | | Total number of unique haplotypes: | 108 | 165 | 150 | 423 |
| | | | | | | | Total number of singletons: | 108 | 165 | 150 | 423 |
| | | | | | | | Most common haplotype count: | 1 | 1 | 1 | 1 |

**Table B9. (Supplementary Table 6). Haplotypes observed within sample set C for proposed linkage group 4.** AA: African American, AS: U.S. Asian, CN: U.S. Caucasian, Hisp: U.S. Hispanic, N: number of samples.

| GATA31E08 | DXS10147 | DXS7423 | AA N=108 | CN N=165 | Hisp N=150 | Overall N=423 |
|---|---|---|---|---|---|---|
| 7 | 7 | 16 | 1 | | | 1 |
| 7 | 8 | 15 | 1 | | | 1 |
| 7 | 9 | 14 | 1 | | | 1 |
| 8 | 6 | 17 | | | 1 | 1 |
| 8 | 7 | 15 | 1 | | | 1 |
| 8 | 8 | 15 | 1 | | | 1 |
| 8 | 9 | 14 | 1 | | | 1 |
| 9 | 6 | 14 | | 2 | 2 | 4 |
| 9 | 6 | 15 | 1 | 9 | 6 | 16 |
| 9 | 6 | 16 | | 1 | | 1 |
| 9 | 7 | 14 | 4 | 1 | | 5 |
| 9 | 7 | 15 | 4 | | 1 | 5 |
| 9 | 7 | 17 | | 1 | | 1 |
| 9 | 8 | 13 | | | 1 | 1 |
| 9 | 8 | 14 | 4 | 2 | 5 | 11 |
| 9 | 8 | 15 | 1 | 6 | 4 | 11 |
| 9 | 8 | 16 | 2 | 1 | | 3 |
| 9 | 8 | 17 | | | 1 | 1 |
| 9 | 9 | 13 | | 3 | 1 | 4 |
| 9 | 9 | 14 | 1 | 3 | | 4 |
| 9 | 9 | 15 | 3 | 3 | 4 | 10 |
| 9 | 9 | 16 | 1 | 3 | 1 | 5 |
| 9 | 9 | 17 | | 1 | 1 | 2 |
| 10 | 6 | 15 | | | 3 | 3 |
| 10 | 7 | 14 | 1 | | | 1 |
| 10 | 7 | 15 | 1 | | | 1 |
| 10 | 7 | 16 | | 1 | | 1 |
| 10 | 8 | 14 | 7 | 1 | 1 | 9 |
| 10 | 8 | 15 | 4 | | | 4 |
| 10 | 8 | 17 | | | 2 | 2 |
| 10 | 9 | 13 | | | 1 | 1 |
| 10 | 9 | 14 | 1 | | | 1 |
| 10 | 9 | 15 | 1 | 1 | | 2 |

| GATA31E08 | DXS10147 | DXS7423 | AA N=108 | CN N=165 | Hisp N=150 | Overall N=423 |
|---|---|---|---|---|---|---|
| 10 | 9 | 16 | | | 1 | 1 |
| 10 | 10 | 16 | 1 | | | 1 |
| 11 | 6 | 14 | 2 | 2 | | 4 |
| 11 | 6 | 15 | 1 | 5 | 4 | 10 |
| 11 | 7 | 14 | | | 1 | 1 |
| 11 | 7 | 15 | 2 | | 1 | 3 |
| 11 | 7 | 16 | | 1 | | 1 |
| 11 | 7 | 17 | | 1 | | 1 |
| 11 | 8 | 14 | 2 | 5 | 6 | 13 |
| 11 | 8 | 15 | 1 | 3 | 2 | 6 |
| 11 | 8 | 16 | | 2 | 1 | 3 |
| 11 | 8 | 17 | | | 2 | 2 |
| 11 | 9 | 13 | | 2 | | 2 |
| 11 | 9 | 14 | 1 | 4 | 1 | 6 |
| 11 | 9 | 15 | | 5 | 2 | 7 |
| 11 | 9 | 16 | | 1 | 2 | 3 |
| 12 | 6 | 13 | 2 | | | 2 |
| 12 | 6 | 14 | 2 | 1 | 2 | 5 |
| 12 | 6 | 15 | 1 | 5 | 16 | 22 |
| 12 | 6 | 16 | | | 3 | 3 |
| 12 | 6 | 17 | | | 1 | 1 |
| 12 | 7 | 14 | 4 | | 2 | 6 |
| 12 | 7 | 15 | 1 | | 2 | 3 |
| 12 | 7 | 17 | | 2 | | 2 |
| 12 | 8 | 14 | 4 | 4 | 11 | 19 |
| 12 | 8 | 15 | 4 | 7 | 9 | 20 |
| 12 | 8 | 16 | | 1 | 1 | 2 |
| 12 | 8 | 17 | | | 1 | 1 |
| 12 | 9 | 13 | | 4 | 2 | 6 |
| 12 | 9 | 14 | 2 | 5 | 2 | 9 |
| 12 | 9 | 15 | | 4 | 1 | 5 |
| 12 | 9 | 16 | 1 | 3 | 1 | 5 |
| 12 | 9 | 17 | | | 1 | 1 |
| 12 | 10 | 15 | 1 | | | 1 |
| 12 | 10 | 16 | 1 | | 1 | 2 |
| 13 | 6 | 14 | 1 | 1 | 1 | 3 |
| 13 | 6 | 15 | 1 | 1 | 6 | 8 |
| 13 | 6 | 16 | | 1 | 1 | 2 |
| 13 | 6 | 17 | | 1 | | 1 |
| 13 | 7 | 14 | 5 | | | 5 |
| 13 | 7 | 15 | 3 | | 1 | 4 |
| 13 | 7 | 16 | | 1 | | 1 |
| 13 | 7 | 17 | | 1 | 1 | 2 |
| 13 | 8 | 13 | | | 1 | 1 |
| 13 | 8 | 14 | 6 | 5 | 3 | 14 |
| 13 | 8 | 15 | 3 | 4 | 7 | 14 |
| 13 | 8 | 16 | 1 | 4 | | 5 |
| 13 | 8 | 17 | | | 2 | 2 |
| 13 | 9 | 13 | | 5 | 1 | 6 |
| 13 | 9 | 14 | 3 | 6 | 2 | 11 |
| 13 | 9 | 15 | | 9 | 2 | 11 |
| 13 | 9 | 16 | | 2 | | 2 |
| 13 | 10 | 15 | 1 | 1 | | 2 |
| 13 | 10 | 16 | 1 | | | 1 |
| 14 | 6 | 15 | | 1 | 2 | 3 |

| GATA31E08 | DXS10147 | DXS7423 | AA N=108 | CN N=165 | Hisp N=150 | Overall N=423 |
|---|---|---|---|---|---|---|
| 14 | 7 | 14 | 2 | | | 2 |
| 14 | 8 | 13 | | 1 | | 1 |
| 14 | 8 | 14 | 2 | 3 | 2 | 7 |
| 14 | 8 | 15 | | 4 | 1 | 5 |
| 14 | 8 | 16 | | 1 | | 1 |
| 14 | 8 | 17 | | | 1 | 1 |
| 14 | 9 | 13 | 2 | 2 | | 4 |
| 14 | 9 | 14 | | | 1 | 1 |
| 14 | 9 | 15 | | 2 | 1 | 3 |
| 15 | 6 | 14 | | 1 | | 1 |
| 15 | 7 | 15 | 1 | | | 1 |
| 15 | 9 | 13 | | 1 | | 1 |
| 15 | 9 | 14 | | 1 | | 1 |
| 15 | 9 | 15 | 1 | | | 1 |
| | Total number of haplotypes: | | 54 | 61 | 61 | 102 |
| | Total number of unique haplotypes: | | 30 | 26 | 31 | 38 |
| | Count of most common haplotype: | | 7 | 9 | 16 | 22 |

**Table B10. Haplotypes observed within the combined U.S. dataset for proposed linkage group 1.** AA: African American, AS: U.S. Asian, CN: U.S. Caucasian, Hisp: U.S. Hispanic, N: number of samples.

| DXS8378 | DXS9902 | DXS6795 | AA N=541 | AS N=326 | CN N=425 | Hisp N=411 | Overall N=1703 |
|---|---|---|---|---|---|---|---|
| 8 | 10 | 11 | | | 1 | | 1 |
| 8 | 12 | 10 | 1 | | | | 1 |
| 8 | 12 | 15 | 1 | | | | 1 |
| 9 | 9 | 11 | | | 1 | | 1 |
| 9 | 10 | 9 | | | 1 | | 1 |
| 9 | 10 | 11 | | | 2 | 1 | 3 |
| 9 | 10 | 12 | | 1 | | | 1 |
| 9 | 10 | 13 | | 3 | | | 3 |
| 9 | 11 | 8 | 1 | | | | 1 |
| 9 | 11 | 9 | | | 1 | | 1 |
| 9 | 11 | 10 | | 1 | | | 1 |
| 9 | 11 | 13 | | 2 | 1 | 2 | 5 |
| 9 | 11 | 14 | | 1 | | | 1 |
| 9 | 11 | 15 | 1 | | | | 1 |
| 9 | 11.1 | 12 | | | | 1 | 1 |
| 9 | 12 | 9 | 1 | | | | 1 |
| 9 | 12 | 11 | | | | 1 | 1 |
| 9 | 12 | 13 | | | | 1 | 1 |
| 10 | 7 | 9 | | 1 | | | 1 |
| 10 | 8 | 9 | | | | 1 | 1 |
| 10 | 8 | 10 | 3 | | | | 3 |
| 10 | 8 | 11 | 1 | | | | 1 |
| 10 | 8 | 13 | 1 | | | | 1 |
| 10 | 8 | 14 | 1 | | | | 1 |
| 10 | 8 | 15 | 1 | | | | 1 |
| 10 | 9 | 9 | | | 1 | | 1 |
| 10 | 9 | 10 | 3 | | | | 3 |
| 10 | 9 | 11 | | 3 | 4 | | 7 |
| 10 | 9 | 12 | 1 | | 1 | 1 | 3 |

| DXS8378 | DXS9902 | DXS6795 | AA N=541 | AS N=326 | CN N=425 | Hisp N=411 | Overall N=1703 |
|---|---|---|---|---|---|---|---|
| 10 | 9 | 13 | 1 | 3 | | | 4 |
| 10 | 9 | 15 | 1 | | | | 1 |
| 10 | 10 | 9 | 5 | 9 | 13 | 16 | 43 |
| 10 | 10 | 10 | 15 | 8 | 2 | 10 | 35 |
| 10 | 10 | 11 | 9 | 16 | 22 | 15 | 62 |
| 10 | 10 | 12 | 2 | 2 | 3 | 12 | 19 |
| 10 | 10 | 13 | 8 | 25 | 9 | 27 | 69 |
| 10 | 10 | 14 | 2 | | | | 2 |
| 10 | 10 | 15 | 6 | | | | 6 |
| 10 | 10 | 17 | 2 | | | | 2 |
| 10 | 10.1 | 11 | | | | 2 | 2 |
| 10 | 11 | 9 | 6 | 11 | 15 | 7 | 39 |
| 10 | 11 | 10 | 18 | 4 | 1 | 7 | 30 |
| 10 | 11 | 11 | 4 | 19 | 25 | 17 | 65 |
| 10 | 11 | 12 | 5 | 2 | 1 | 10 | 18 |
| 10 | 11 | 13 | 2 | 24 | 9 | 14 | 49 |
| 10 | 11 | 14 | 1 | | | 2 | 3 |
| 10 | 11 | 15 | 13 | | | | 13 |
| 10 | 11.1 | 9 | 1 | | | 1 | 2 |
| 10 | 11.1 | 11 | 1 | 1 | 3 | 1 | 6 |
| 10 | 11.1 | 13 | | | 1 | | 1 |
| 10 | 12 | 9 | | 7 | 15 | 3 | 25 |
| 10 | 12 | 10 | 8 | 7 | 1 | 4 | 20 |
| 10 | 12 | 11 | 4 | 12 | 17 | 8 | 41 |
| 10 | 12 | 12 | 3 | | 3 | 7 | 13 |
| 10 | 12 | 13 | 5 | 11 | 6 | 7 | 29 |
| 10 | 12 | 14 | | 1 | | 1 | 2 |
| 10 | 12 | 15 | 7 | | | | 7 |
| 10 | 12 | 16 | 1 | | | | 1 |
| 10 | 12.1 | 9 | | | 1 | | 1 |
| 10 | 13 | 10 | 1 | | | | 1 |
| 10 | 13 | 11 | 1 | | | | 1 |
| 10 | 13 | 12 | | | 1 | | 1 |
| 10 | 13 | 13 | | 1 | | | 1 |
| 11 | 8 | 9 | 1 | | | | 1 |
| 11 | 8 | 11 | 1 | | | | 1 |
| 11 | 8 | 13 | 1 | | | | 1 |
| 11 | 8 | 15 | 3 | | | | 3 |
| 11 | 9 | 9 | 3 | | 1 | 2 | 6 |
| 11 | 9 | 10 | 7 | | | | 7 |
| 11 | 9 | 11 | 2 | | 2 | 1 | 5 |
| 11 | 9 | 12 | 2 | | | 1 | 3 |
| 11 | 9 | 13 | | | | 2 | 2 |
| 11 | 9 | 15 | 1 | | | | 1 |
| 11 | 10 | 9 | 6 | 3 | 16 | 7 | 32 |
| 11 | 10 | 10 | 15 | 3 | | 4 | 22 |
| 11 | 10 | 11 | 8 | 18 | 18 | 12 | 56 |
| 11 | 10 | 12 | 8 | 2 | 2 | 7 | 19 |
| 11 | 10 | 13 | 3 | 12 | 9 | 18 | 42 |
| 11 | 10 | 14 | 4 | 1 | | 1 | 6 |
| 11 | 10 | 15 | 12 | | 2 | 1 | 15 |
| 11 | 10 | 16 | 1 | | | | 1 |
| 11 | 11 | 9 | 14 | 5 | 18 | 6 | 43 |
| 11 | 11 | 10 | 20 | 4 | 3 | 5 | 32 |
| 11 | 11 | 11 | 11 | 16 | 24 | 13 | 64 |

| DXS8378 | DXS9902 | DXS6795 | AA N=541 | AS N=326 | CN N=425 | Hisp N=411 | Overall N=1703 |
|---|---|---|---|---|---|---|---|
| 11 | 11 | 12 | 4 | 1 | 2 | 4 | 11 |
| 11 | 11 | 13 | 7 | 9 | 10 | 13 | 39 |
| 11 | 11 | 14 | 2 | | | 1 | 3 |
| 11 | 11 | 15 | 17 | | | | 17 |
| 11 | 11 | 16 | 1 | | | | 1 |
| 11 | 11 | 18 | 1 | | | | 1 |
| 11 | 11.1 | 9 | | 1 | 2 | 2 | 5 |
| 11 | 11.1 | 10 | 1 | | | | 1 |
| 11 | 11.1 | 12 | | | 1 | | 1 |
| 11 | 11.1 | 13 | 1 | | 3 | 3 | 7 |
| 11 | 12 | 6 | 1 | | | | 1 |
| 11 | 12 | 9 | 2 | 2 | 7 | 3 | 14 |
| 11 | 12 | 10 | 17 | 2 | 1 | 3 | 23 |
| 11 | 12 | 11 | 11 | 9 | 14 | 8 | 42 |
| 11 | 12 | 12 | 6 | 2 | | | 8 |
| 11 | 12 | 13 | 1 | 3 | 6 | 11 | 21 |
| 11 | 12 | 14 | 1 | | | | 1 |
| 11 | 12 | 15 | 9 | | | 1 | 10 |
| 11 | 13 | 9 | | | | 1 | 1 |
| 11 | 13 | 11 | 2 | | 1 | | 3 |
| 11 | 13 | 12 | 1 | | | | 1 |
| 12 | 8 | 9 | 2 | | | | 2 |
| 12 | 8 | 10 | 5 | | | | 5 |
| 12 | 8 | 12 | 1 | | | | 1 |
| 12 | 8 | 15 | 2 | | | | 2 |
| 12 | 8 | 17 | 1 | | | | 1 |
| 12 | 9 | 9 | 1 | 1 | 1 | 1 | 4 |
| 12 | 9 | 10 | 1 | | | 1 | 2 |
| 12 | 9 | 11 | 1 | | 1 | | 2 |
| 12 | 9 | 13 | | 1 | 2 | 1 | 4 |
| 12 | 9 | 14 | 1 | | | | 1 |
| 12 | 10 | 9 | 8 | 5 | 18 | 4 | 35 |
| 12 | 10 | 10 | 17 | | | 2 | 19 |
| 12 | 10 | 11 | 8 | 4 | 17 | 7 | 36 |
| 12 | 10 | 12 | 5 | | | 8 | 13 |
| 12 | 10 | 13 | 3 | 8 | 6 | 10 | 27 |
| 12 | 10 | 14 | 1 | | | 1 | 2 |
| 12 | 10 | 15 | 10 | | 1 | | 11 |
| 12 | 11 | 9 | 7 | 4 | 12 | 4 | 27 |
| 12 | 11 | 10 | 17 | | | 2 | 19 |
| 12 | 11 | 11 | 14 | 8 | 12 | 16 | 50 |
| 12 | 11 | 12 | 5 | 1 | 2 | 3 | 11 |
| 12 | 11 | 13 | 1 | 4 | 7 | 10 | 22 |
| 12 | 11 | 14 | 2 | | | | 2 |
| 12 | 11 | 15 | 7 | | | | 7 |
| 12 | 11 | 16 | 1 | | | | 1 |
| 12 | 11 | 17 | 1 | | | | 1 |
| 12 | 11.1 | 9 | | 1 | 1 | 1 | 3 |
| 12 | 11.1 | 11 | 1 | | 1 | 1 | 3 |
| 12 | 11.1 | 12 | | | | 1 | 1 |
| 12 | 11.1 | 13 | | | 1 | | 1 |
| 12 | 12 | 9 | 3 | 3 | 6 | 7 | 19 |
| 12 | 12 | 10 | 14 | 3 | 2 | 1 | 20 |
| 12 | 12 | 11 | 11 | 2 | 7 | 7 | 27 |
| 12 | 12 | 12 | 4 | | 1 | 1 | 6 |

| DXS8378 | DXS9902 | DXS6795 | AA N=541 | AS N=326 | CN N=425 | Hisp N=411 | Overall N=1703 |
|---|---|---|---|---|---|---|---|
| 12 | 12 | 13 | 4 | 4 | 1 | 6 | 15 |
| 12 | 12 | 14 | 1 | 1 | | 1 | 3 |
| 12 | 12 | 15 | 4 | | | 1 | 5 |
| 12 | 13 | 11 | | 1 | 2 | | 3 |
| 13 | 7 | 11 | | | 1 | | 1 |
| 13 | 8 | 10 | 2 | | | | 2 |
| 13 | 8 | 14 | 1 | | | | 1 |
| 13 | 9 | 11 | | | 1 | | 1 |
| 13 | 9 | 12 | | | 1 | | 1 |
| 13 | 9 | 13 | | | 1 | | 1 |
| 13 | 10 | 9 | 1 | | 1 | | 2 |
| 13 | 10 | 10 | 2 | | | | 2 |
| 13 | 10 | 11 | 1 | 1 | 3 | | 5 |
| 13 | 10 | 12 | | | | 1 | 1 |
| 13 | 10 | 13 | 1 | 1 | 1 | 1 | 4 |
| 13 | 10 | 15 | 1 | | | | 1 |
| 13 | 11 | 9 | 3 | 1 | 2 | | 6 |
| 13 | 11 | 10 | 2 | | | 1 | 3 |
| 13 | 11 | 11 | | 1 | 1 | | 2 |
| 13 | 11 | 13 | | 1 | 2 | | 3 |
| 13 | 11.1 | 9 | | | | 1 | 1 |
| 13 | 11.1 | 11 | | | 1 | | 1 |
| 13 | 12 | 9 | | 1 | 1 | | 2 |
| 13 | 12 | 10 | 1 | | | | 1 |
| 13 | 12 | 11 | 1 | | 2 | | 3 |
| 13 | 12 | 13 | 1 | | 1 | | 2 |
| 13 | 12 | 14 | 1 | | | | 1 |
| 13 | 12 | 15 | 1 | | | | 1 |
| 13 | 13 | 9 | | | 1 | | 1 |
| 13 | 13 | 15 | 1 | | | | 1 |
| 14 | 9 | 14 | 1 | | | | 1 |
| 14 | 11 | 9 | | 1 | | | 1 |
| 14 | 11.1 | 11 | | | 1 | | 1 |
| 14 | 12 | 9 | | | | 1 | 1 |
| Total number of haplotypes: | | | 126 | 65 | 84 | 81 | 173 |
| Total number of unique haplotypes: | | | 55 | 23 | 37 | 32 | 68 |
| Count of most common haplotype: | | | 20 | 25 | 25 | 27 | 69 |

**Table B11. Haplotypes observed within the combined U.S. dataset for proposed linkage group 2.** AA: African American, AS: U.S. Asian, CN: U.S. Caucasian, Hisp: U.S. Hispanic, N: number of samples.

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 11 | 10 | 19 | 13 | 28 | 8 | 15.3 | 11 | 1 | | | | 1 |
| 11 | 11 | 15 | 15 | 27 | 8 | 14.3 | 11 | 1 | | | | 1 |
| 11 | 11 | 15 | 16 | 28 | 7 | 12 | 10 | 1 | | | | 1 |
| 11 | 11 | 16 | 16 | 24 | 9 | 15.3 | 10 | 1 | | | | 1 |
| 11 | 11 | 21 | 15 | 26 | 10 | 17.3 | 10 | | | | 1 | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 11 | 11 | 22 | 16 | 18 | 10 | 14.3 | 11 | 1 | | | | 1 |
| 11 | 12 | 15 | 14 | 26 | 6 | 15.3 | 11 | 1 | | | | 1 |
| 11 | 12.3 | 15 | 13 | 19 | 6 | 15.3 | 11 | 1 | | | | 1 |
| 11 | 12.3 | 21 | 15 | 26 | 6 | 14.3 | 10 | 1 | | | | 1 |
| 11 | 13 | 20 | 13 | 25 | 12 | 13 | 10 | | | 1 | | 1 |
| 11 | 14.3 | 20 | 17 | 18 | 12 | 15.3 | 9 | | | 1 | | 1 |
| 12 | 8 | 21 | 13 | 28 | 8 | 14.3 | 12 | 1 | | | | 1 |
| 12 | 9 | 19 | 14 | 27 | 10 | 13 | 8 | 1 | | | | 1 |
| 12 | 10 | 15 | 13 | 19 | 6 | 12 | 12 | 1 | | | | 1 |
| 12 | 10 | 19 | 16 | 27 | 9 | 13 | 10 | 1 | | | | 1 |
| 12 | 10 | 20 | 11 | 26 | 6 | 15.3 | 12 | 1 | | | | 1 |
| 12 | 10 | 20 | 12 | 24 | 10 | 12 | 11 | 1 | | | | 1 |
| 12 | 10 | 20 | 14 | 29 | 10 | 16.3 | 9 | | 1 | | | 1 |
| 12 | 10 | 20 | 16 | 26 | 10 | 12 | 11 | 1 | | | | 1 |
| 12 | 10 | 20 | 17 | 18 | 11 | 13.3 | 11 | 1 | | | | 1 |
| 12 | 10 | 21 | 13 | 26 | 9 | 13 | 11 | 1 | | | | 1 |
| 12 | 10 | 21 | 15 | 29 | 9 | 15.3 | 9 | | | 1 | | 1 |
| 12 | 10 | 21 | 16 | 15 | 8 | 13.3 | 10 | | | 1 | | 1 |
| 12 | 10 | 23 | 15 | 24 | 6 | 14.3 | 11 | 1 | | | | 1 |
| 12 | 11 | 14 | 12 | 21 | 12 | 14.3 | 10 | 1 | | | | 1 |
| 12 | 11 | 15 | 11 | 21 | 9 | 15.3 | 11 | 1 | | | | 1 |
| 12 | 11 | 15 | 14 | 18 | 9 | 12 | 11 | 1 | | | | 1 |
| 12 | 11 | 15 | 14 | 22 | 11 | 12 | 9 | 1 | | | | 1 |
| 12 | 11 | 15 | 16 | 24 | 12 | 14.3 | 11 | | | 1 | | 1 |
| 12 | 11 | 15 | 16 | 25 | 9 | 16.3 | 11 | 1 | | | | 1 |
| 12 | 11 | 16 | 14 | 21 | 6 | 11 | 11 | 1 | | | | 1 |
| 12 | 11 | 16 | 15 | 27 | 8 | 14.3 | 11 | 1 | | | | 1 |
| 12 | 11 | 16 | 16 | 22 | 10 | 11 | 10 | | 1 | | | 1 |
| 12 | 11 | 16 | 16 | 24 | 6 | 14.3 | 11 | 1 | | | | 1 |
| 12 | 11 | 16 | 16 | 25 | 9 | 12 | 9 | | 1 | | | 1 |
| 12 | 11 | 19 | 11 | 21 | 9 | 12 | 11 | 1 | | | | 1 |
| 12 | 11 | 19 | 14 | 19 | 6 | 13 | 10 | 1 | | | | 1 |
| 12 | 11 | 19 | 14 | 22 | 6 | 10 | 9 | 1 | | | | 1 |
| 12 | 11 | 19 | 15 | 26 | 10 | 13.3 | 11 | | | | 1 | 1 |
| 12 | 11 | 19 | 16 | 18 | 11 | 14.3 | 10 | | | | 1 | 1 |
| 12 | 11 | 19 | 16 | 28 | 11 | 11 | 10 | | | | 1 | 1 |
| 12 | 11 | 19 | 16 | 28 | 11 | 14.3 | 11 | | | | 1 | 1 |
| 12 | 11 | 20 | 10 | 28 | 10 | 15.3 | 12 | 1 | | | | 1 |
| 12 | 11 | 20 | 13 | 24 | 9 | 13 | 11 | 1 | | | | 1 |
| 12 | 11 | 20 | 14 | 29 | 8 | 16.3 | 12 | | | 1 | | 1 |
| 12 | 11 | 20 | 15 | 23 | 8 | 14.3 | 9 | | | 1 | | 1 |
| 12 | 11 | 20 | 15 | 24 | 11 | 14.3 | 10 | | 1 | | | 1 |
| 12 | 11 | 20 | 15 | 26 | 10 | 15.3 | 9 | | | | 1 | 1 |
| 12 | 11 | 20 | 15 | 27 | 10 | 15.3 | 12 | 1 | | | | 1 |
| 12 | 11 | 20 | 16 | 25 | 6 | 12 | 9 | | 1 | | | 1 |
| 12 | 11 | 20 | 16 | 25 | 8 | 13 | 9 | 1 | | | | 1 |
| 12 | 11 | 20 | 16 | 28 | 6 | 12 | 9 | | | | 1 | 1 |
| 12 | 11 | 21 | 13 | 26 | 10 | 14.3 | 10 | | | | 1 | 1 |
| 12 | 11 | 21 | 13 | 29 | 8 | 13 | 9 | 1 | | | | 1 |
| 12 | 11 | 21 | 14 | 25 | 8 | 14.3 | 11 | 1 | | | | 1 |
| 12 | 11 | 21 | 14 | 27 | 10 | 13 | 11 | | 1 | | | 1 |
| 12 | 11 | 21 | 14 | 27 | 10 | 13.3 | 9 | | | 1 | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 12 | 11 | 21 | 15 | 26 | 10 | 16.3 | 11 | 1 | | | | 1 |
| 12 | 11 | 21 | 16 | 19 | 12 | 15.3 | 10 | 1 | | | | 1 |
| 12 | 11 | 21 | 16 | 21 | 10 | 15.3 | 10 | | 1 | | | 1 |
| 12 | 11 | 21 | 16 | 24 | 6 | 14 | 11 | 1 | | | | 1 |
| 12 | 11 | 21 | 16 | 24 | 11 | 11 | 10 | | 1 | | | 1 |
| 12 | 11 | 22 | 12 | 27 | 8 | 16.3 | 9 | | | 1 | | 1 |
| 12 | 11 | 22 | 13 | 25 | 6 | 14 | 11 | 1 | | | | 1 |
| 12 | 11 | 22 | 13 | 26 | 9 | 15.3 | 11 | | | | 1 | 1 |
| 12 | 11 | 22 | 14 | 26 | 9 | 14.3 | 11 | 1 | | | | 1 |
| 12 | 11 | 22 | 15 | 18 | 10 | 14.3 | 11 | | | 1 | | 1 |
| 12 | 11.3 | 16 | 15 | 23 | 10 | 15.3 | 11 | | 1 | | | 1 |
| 12 | 11.3 | 19 | 16 | 25 | 10 | 15.3 | 11 | | | | 1 | 1 |
| 12 | 11.3 | 21 | 13 | 26 | 10 | 16.3 | 9 | | 1 | | | 1 |
| 12 | 11.3 | 21 | 14 | 23 | 11 | 14 | 10 | | | | 1 | 1 |
| 12 | 11.3 | 22 | 13 | 25 | 11 | 14.3 | 9 | | | | 1 | 1 |
| 12 | 12 | 14 | 15 | 25 | 11 | 13.3 | 9 | | | 1 | | 1 |
| 12 | 12 | 15 | 13 | 26 | 8 | 15.3 | 10 | 1 | | | | 1 |
| 12 | 12 | 15 | 14 | 28 | 8 | 15.3 | 11 | 1 | | | | 1 |
| 12 | 12 | 15 | 16 | 25 | 10 | 15.3 | 11 | | | 1 | | 1 |
| 12 | 12 | 16 | 14 | 19 | 7 | 14.3 | 10 | 1 | | | | 1 |
| 12 | 12 | 18 | 13 | 26 | 11 | 15.3 | 10 | | | | 1 | 1 |
| 12 | 12 | 19 | 16 | 27 | 9 | 14.3 | 11 | 1 | | | | 1 |
| 12 | 12 | 20 | 12 | 26 | 10 | 15.3 | 9 | | | | 1 | 1 |
| 12 | 12 | 20 | 12 | 28 | 6 | 10 | 10 | 1 | | | | 1 |
| 12 | 12 | 20 | 13 | 21 | 9 | 12 | 10 | 1 | | | | 1 |
| 12 | 12 | 20 | 13 | 25 | 11 | 15.3 | 8 | 1 | | | | 1 |
| 12 | 12 | 20 | 13 | 26 | 9 | 15.3 | 11 | 1 | | | | 1 |
| 12 | 12 | 20 | 13 | 30 | 12 | 14 | 11 | 1 | | | | 1 |
| 12 | 12 | 20 | 14 | 24 | 8 | 12 | 9 | | | 1 | | 1 |
| 12 | 12 | 20 | 14 | 26 | 8 | 14.3 | 9 | | | 1 | | 1 |
| 12 | 12 | 20 | 15 | 23 | 10 | 12 | 9 | 1 | | | | 1 |
| 12 | 12 | 20 | 15 | 25 | 10 | 12 | 10 | | | | 1 | 1 |
| 12 | 12 | 20 | 15 | 26 | 8 | 15.3 | 10 | | | | 1 | 1 |
| 12 | 12 | 20 | 15 | 27 | 10 | 15.3 | 9 | | 1 | | | 1 |
| 12 | 12 | 20 | 16 | 18 | 9 | 16.3 | 9 | | 1 | | | 1 |
| 12 | 12 | 20 | 16 | 18 | 11 | 14.3 | 10 | | | | 1 | 1 |
| 12 | 12 | 20 | 16 | 19 | 10 | 16.3 | 10 | | | | 1 | 1 |
| 12 | 12 | 20 | 17 | 18 | 11 | 15.3 | 9 | | | 1 | | 1 |
| 12 | 12 | 21 | 13 | 22 | 9 | 13 | 10 | 1 | | | | 1 |
| 12 | 12 | 21 | 13 | 27 | 10 | 16.3 | 9 | | | | 1 | 1 |
| 12 | 12 | 21 | 14 | 22 | 8 | 9 | 11 | 1 | | | | 1 |
| 12 | 12 | 21 | 14 | 23 | 6 | 15.3 | 11 | | | | 1 | 1 |
| 12 | 12 | 21 | 14 | 24 | 10 | 14.3 | 9 | | | 1 | | 1 |
| 12 | 12 | 21 | 14 | 26 | 12 | 12 | 13 | | | | 1 | 1 |
| 12 | 12 | 21 | 15 | 23 | 8 | 16.3 | 11 | | | | 1 | 1 |
| 12 | 12 | 21 | 16 | 24 | 10 | 15.3 | 11 | | | 1 | | 1 |
| 12 | 12 | 21 | 16 | 25 | 6 | 15.3 | 10 | 1 | | | | 1 |
| 12 | 12 | 21 | 17 | 21 | 6 | 14.3 | 10 | 1 | | | | 1 |
| 12 | 12 | 22 | 13 | 24 | 11 | 11 | 10 | | 1 | | | 1 |
| 12 | 12 | 22 | 15 | 23 | 10 | 14.3 | 11 | | | | 1 | 1 |
| 12 | 12 | 22 | 16 | 23 | 6 | 13 | 12 | 1 | | | | 1 |
| 12 | 12 | 22 | 16 | 26 | 9 | 12 | 10 | 1 | | | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 12 | 12 | 23 | 14 | 21 | 9 | 11 | 10 | 1 | | | | 1 |
| 12 | 12 | 23 | 17 | 15 | 10 | 15.3 | 11 | 1 | | | | 1 |
| 12 | 12.3 | 15 | 16 | 25 | 10 | 15.3 | 11 | | 1 | | | 1 |
| 12 | 12.3 | 16 | 16 | 24 | 10 | 11 | 11 | | 1 | | | 1 |
| 12 | 12.3 | 19 | 16 | 23 | 11 | 16.3 | 10 | | 1 | | | 1 |
| 12 | 12.3 | 20 | 15 | 25 | 11 | 14 | 9 | | 1 | | | 1 |
| 12 | 12.3 | 20 | 15 | 26 | 6 | 12 | 10 | | | | 1 | 1 |
| 12 | 12.3 | 20 | 16 | 22 | 12 | 13 | 10 | | | | 1 | 1 |
| 12 | 12.3 | 21 | 14 | 24 | 6 | 15.3 | 11 | | | 1 | | 1 |
| 12 | 12.3 | 21 | 14 | 25 | 11 | 12 | 11 | | | 1 | | 1 |
| 12 | 12.3 | 21 | 15 | 24 | 10 | 15.3 | 10 | | 1 | | | 1 |
| 12 | 12.3 | 21 | 15 | 27 | 10 | 15.3 | 11 | | | 1 | | 1 |
| 12 | 12.3 | 21 | 16 | 24 | 8 | 16.3 | 10 | 1 | | | | 1 |
| 12 | 12.3 | 21 | 16 | 24 | 9 | 11 | 10 | | | | 1 | 1 |
| 12 | 12.3 | 21 | 16 | 24 | 10 | 12 | 9 | | | | 1 | 1 |
| 12 | 12.3 | 21 | 16 | 25 | 10 | 12 | 10 | | | | 1 | 1 |
| 12 | 12.3 | 21 | 16 | 26 | 8 | 11 | 10 | | 1 | | | 1 |
| 12 | 12.3 | 22 | 14 | 26 | 10 | 12 | 10 | | | | 1 | 1 |
| 12 | 13 | 15 | 14 | 26 | 8 | 12 | 10 | 1 | | | | 1 |
| 12 | 13 | 15 | 14 | 26 | 11 | 11 | 10 | 1 | | | | 1 |
| 12 | 13 | 16 | 16 | 23 | 12 | 14.3 | 9 | | 1 | | | 1 |
| 12 | 13 | 20 | 10 | 21 | 8 | 15.3 | 11 | 1 | | | | 1 |
| 12 | 13 | 20 | 12 | 25 | 10 | 14.3 | 9 | | 1 | | | 1 |
| 12 | 13 | 20 | 15 | 19 | 12 | 12 | 11 | | | 1 | | 1 |
| 12 | 13 | 20 | 15 | 24 | 8 | 12 | 11 | | 1 | | | 1 |
| 12 | 13 | 20 | 15 | 26 | 11 | 17.3 | 11 | | 1 | | | 1 |
| 12 | 13 | 20 | 16 | 24 | 12 | 16.3 | 10 | | | 1 | | 1 |
| 12 | 13 | 20 | 16 | 28 | 12 | 15.3 | 12 | | | 1 | | 1 |
| 12 | 13 | 21 | 12 | 26 | 11 | 15.3 | 10 | | | 1 | | 1 |
| 12 | 13 | 21 | 16 | 24 | 12 | 14.3 | 9 | | | 1 | | 1 |
| 12 | 13 | 21 | 16 | 27 | 10 | 16.3 | 9 | | 1 | | | 1 |
| 12 | 13 | 22 | 13 | 26 | 12 | 15.3 | 10 | | | 1 | | 1 |
| 12 | 13.3 | 15 | 17 | 25 | 10 | 11 | 9 | | 1 | | | 1 |
| 12 | 13.3 | 20 | 13 | 26 | 11 | 14.3 | 9 | | | 1 | | 1 |
| 12 | 13.3 | 20 | 15 | 25 | 10 | 14.3 | 10 | | 1 | | | 1 |
| 12 | 13.3 | 20 | 15 | 26 | 10 | 15.3 | 9 | | | 1 | | 1 |
| 12 | 13.3 | 20 | 16 | 25 | 6 | 12 | 10 | | | 1 | | 1 |
| 12 | 13.3 | 20 | 16 | 25 | 11 | 15.3 | 11 | | | 1 | | 1 |
| 12 | 13.3 | 20 | 16 | 30 | 10 | 11 | 9 | | | | 1 | 1 |
| 12 | 13.3 | 21 | 14 | 24 | 11 | 12 | 10 | | | | 1 | 1 |
| 12 | 13.3 | 21 | 15 | 19 | 11 | 15.3 | 10 | | | | 1 | 1 |
| 12 | 13.3 | 21 | 16 | 21 | 9 | 14.3 | 10 | | | | 1 | 1 |
| 12 | 13.3 | 21 | 17 | 23 | 11 | 16.3 | 11 | | 1 | | | 1 |
| 12 | 13.3 | 22 | 12 | 28 | 6 | 15.3 | 9 | | | | 1 | 1 |
| 12 | 13.3 | 22 | 15 | 25 | 6 | 14.3 | 11 | | 1 | | | 1 |
| 12 | 13.3 | 22 | 16 | 25 | 6 | 15.3 | 11 | 1 | | | | 1 |
| 12 | 13.3 | 22 | 17 | 18 | 6 | 15.3 | 9 | | | | 1 | 1 |
| 12 | 14 | 15 | 15 | 26 | 11 | 12 | 12 | 1 | | | | 1 |
| 12 | 14 | 20 | 13 | 22 | 9 | 12 | 8 | 1 | | | | 1 |
| 12 | 15.3 | 20 | 16 | 24 | 6 | 15.3 | 10 | | | 1 | | 1 |
| 12 | 15.3 | 20 | 16 | 25 | 11 | 15.3 | 11 | | | 1 | | 1 |
| 13 | 7 | 22 | 12 | 20 | 6 | 13 | 10 | | | | 1 | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 13 | 8 | 20 | 13 | 19 | 7 | 13 | 10 | 1 | | | | 1 |
| 13 | 8 | 22 | 16 | 26 | 6 | 12 | 11 | 1 | | | | 1 |
| 13 | 10 | 15 | 13 | 21 | 12 | 15.3 | 9 | 1 | | | | 1 |
| 13 | 10 | 15 | 13 | 24 | 10 | 14.3 | 10 | 1 | | | | 1 |
| 13 | 10 | 16 | 11 | 21 | 9 | 15.3 | 12 | 1 | | | | 1 |
| 13 | 10 | 16 | 13 | 27 | 9 | 11 | 10 | 1 | | | | 1 |
| 13 | 10 | 20 | 11 | 26 | 11 | 15.3 | 9 | | | | 1 | 1 |
| 13 | 10 | 20 | 12 | 19 | 10 | 15.3 | 8 | 1 | | | | 1 |
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| 13 | 10 | 20 | 14 | 27 | 7 | 12 | 11 | 1 | | | | 1 |
| 13 | 10 | 20 | 16 | 18 | 8 | 15.3 | 10 | | 1 | | | 1 |
| 13 | 10 | 20 | 16 | 22 | 6 | 15.3 | 10 | | | 1 | | 1 |
| 13 | 10 | 20 | 19 | 15 | 10 | 15.3 | 9 | | | 1 | | 1 |
| 13 | 10 | 21 | 14 | 25 | 9 | 14.3 | 10 | 1 | | | | 1 |
| 13 | 10 | 21 | 15 | 24 | 8 | 15.3 | 9 | | | 1 | | 1 |
| 13 | 10 | 21 | 17 | 18 | 6 | 12 | 9 | 1 | | | | 1 |
| 13 | 10 | 22 | 13 | 24 | 10 | 17.3 | 9 | | | 1 | | 1 |
| 13 | 10 | 22 | 13 | 27 | 6 | 13 | 11 | | | | 1 | 1 |
| 13 | 10 | 22 | 16 | 28 | 10 | 14.3 | 11 | 1 | | | | 1 |
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| 13 | 11 | 14 | 16 | 24 | 8 | 14.3 | 10 | | | | 1 | 1 |
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| 13 | 11 | 15 | 13 | 29 | 8 | 13.3 | 11 | 1 | | | | 1 |
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| 13 | 11 | 15 | 14 | 22 | 9 | 14.3 | 10 | 1 | | | | 1 |
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| 13 | 11 | 15 | 15 | 27 | 9 | 13 | 11 | 1 | | | | 1 |
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| 13 | 11 | 16 | 13 | 25 | 8 | 15.3 | 10 | | 1 | | | 1 |
| 13 | 11 | 16 | 14 | 18 | 7 | 12 | 12 | 1 | | | | 1 |
| 13 | 11 | 16 | 14 | 25 | 9 | 13 | 11 | 1 | | | | 1 |
| 13 | 11 | 16 | 14 | 28 | 9 | 13 | 8 | 1 | | | | 1 |
| 13 | 11 | 16 | 14 | 29 | 10 | 15.3 | 11 | 1 | | | | 1 |
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| 13 | 11 | 16 | 15 | 25 | 11 | 12 | 9 | | 1 | | | 1 |
| 13 | 11 | 16 | 16 | 23 | 7 | 13 | 10 | 1 | | | | 1 |
| 13 | 11 | 16 | 16 | 27 | 8 | 12 | 10 | | 1 | | | 1 |
| 13 | 11 | 18 | 15 | 19 | 11 | 13 | 10 | | | | 1 | 1 |
| 13 | 11 | 19 | 16 | 25 | 10 | 11 | 11 | | | 1 | | 1 |
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| 13 | 11 | 20 | 12 | 25 | 11 | 12 | 11 | | | | 1 | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
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| 13 | 11 | 20 | 16 | 22 | 12 | 16.3 | 10 | | | 1 | | 1 |
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| 13 | 11 | 21 | 12 | 24 | 9 | 13 | 10 | 1 | | | | 1 |
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| 13 | 11 | 21 | 16 | 19 | 7 | 12 | 11 | 1 | | | | 1 |
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| 13 | 11 | 21 | 16 | 27 | 6 | 12 | 11 | 1 | | | | 1 |
| 13 | 11 | 21 | 16 | 28 | 6 | 12 | 9 | 1 | | | | 1 |
| 13 | 11 | 21 | 17 | 23 | 10 | 14.3 | 10 | | | | 1 | 1 |
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| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
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| 13 | 11 | 22 | 14 | 21 | 11 | 16.3 | 12 | 1 | | | | 1 |
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| 13 | 11 | 23 | 14 | 27 | 9 | 13 | 10 | 1 | | | | 1 |
| 13 | 11 | 23 | 15 | 25 | 12 | 15.3 | 9 | | | 1 | | 1 |
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| 13 | 11.3 | 16 | 13 | 27 | 12 | 12 | 10 | | | | 1 | 1 |
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| 13 | 11.3 | 20 | 14 | 26 | 6 | 12 | 10 | | | | 1 | 1 |
| 13 | 11.3 | 20 | 14 | 26 | 8 | 13 | 10 | 1 | | | | 1 |
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| 13 | 12 | 15 | 16 | 25 | 8 | 14.3 | 8 | | | 1 | | 1 |
| 13 | 12 | 16 | 13 | 24 | 11 | 16.3 | 11 | | 1 | | | 1 |
| 13 | 12 | 16 | 14 | 21 | 9 | 12 | 8 | 1 | | | | 1 |
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| 13 | 12 | 16 | 15 | 23 | 10 | 14.3 | 11 | | 1 | | | 1 |
| 13 | 12 | 16 | 15 | 25 | 8 | 14.3 | 9 | | 1 | | | 1 |
| 13 | 12 | 16 | 15 | 28 | 10 | 16.3 | 9 | | | 1 | | 1 |
| 13 | 12 | 16 | 16 | 25 | 10 | 16.3 | 11 | | | 1 | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
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| 13 | 12 | 17 | 13 | 20 | 9 | 14.3 | 11 | 1 | | | | 1 |
| 13 | 12 | 17 | 14 | 24 | 12 | 17.3 | 11 | | 1 | | | 1 |
| 13 | 12 | 19 | 13 | 26 | 8 | 12 | 10 | | | | 1 | 1 |
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| 13 | 12 | 19 | 14 | 27 | 11 | 12 | 10 | 1 | | | | 1 |
| 13 | 12 | 20 | 12 | 21 | 6 | 12 | 10 | 1 | | | | 1 |
| 13 | 12 | 20 | 12 | 26 | 11 | 15.3 | 12 | | | | 1 | 1 |
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| 13 | 12 | 20 | 13 | 26 | 6 | 15.3 | 10 | 1 | | | | 1 |
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| 13 | 12 | 20 | 14 | 24 | 10 | 15.3 | 9 | | | 1 | | 1 |
| 13 | 12 | 20 | 14 | 24 | 10 | 15.3 | 11 | | | 1 | | 1 |
| 13 | 12 | 20 | 14 | 25 | 12 | 12 | 9 | | | | 1 | 1 |
| 13 | 12 | 20 | 14 | 26 | 12 | 12 | 11 | 1 | | | | 1 |
| 13 | 12 | 20 | 14 | 27 | 10 | 12 | 11 | | | | 1 | 1 |
| 13 | 12 | 20 | 14 | 27 | 10 | 15.3 | 11 | | | 1 | | 1 |
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| 13 | 12 | 20 | 15 | 18 | 10 | 14.3 | 10 | | | | 1 | 1 |
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| 13 | 12 | 20 | 15 | 26 | 6 | 11 | 9 | | | 1 | | 1 |
| 13 | 12 | 20 | 15 | 26 | 10 | 16.3 | 10 | | | | 1 | 1 |
| 13 | 12 | 20 | 15 | 27 | 10 | 15.3 | 10 | | | 1 | | 1 |
| 13 | 12 | 20 | 16 | 15 | 8 | 15.3 | 9 | | | 1 | | 1 |
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| 13 | 12 | 20 | 16 | 19 | 9 | 11 | 9 | | | 1 | | 1 |
| 13 | 12 | 20 | 16 | 19 | 10 | 15.3 | 11 | | | | 1 | 1 |
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| 13 | 12 | 20 | 16 | 24 | 12 | 12 | 10 | | 1 | | | 1 |
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| 13 | 12 | 20 | 16 | 25 | 11 | 15.3 | 11 | | | 1 | | 1 |
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| 13 | 12 | 20 | 16 | 27 | 11 | 15.3 | 10 | | | | 1 | 1 |
| 13 | 12 | 20 | 17 | 20 | 12 | 12 | 11 | | | | 1 | 1 |
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| 13 | 12 | 20 | 17 | 30 | 10 | 15.3 | 9 | | 1 | | | 1 |
| 13 | 12 | 21 | 13 | 19 | 9 | 13 | 11 | 1 | | | | 1 |
| 13 | 12 | 21 | 13 | 20 | 8 | 15.3 | 11 | 1 | | | | 1 |
| 13 | 12 | 21 | 13 | 22 | 7 | 9 | 11 | 1 | | | | 1 |
| 13 | 12 | 21 | 13 | 24 | 10 | 15.3 | 9 | | | | 1 | 1 |
| 13 | 12 | 21 | 13 | 26 | 10 | 13 | 11 | | | 1 | | 1 |
| 13 | 12 | 21 | 13 | 26 | 12 | 11 | 9 | 1 | | | | 1 |
| 13 | 12 | 21 | 13 | 26 | 12 | 15.3 | 9 | | 1 | | | 1 |
| 13 | 12 | 21 | 14 | 21 | 6 | 14.3 | 11 | 1 | | | | 1 |
| 13 | 12 | 21 | 14 | 24 | 8 | 15.3 | 10 | | | 1 | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 13 | 12 | 21 | 14 | 24 | 10 | 12 | 10 | | 1 | | | 1 |
| 13 | 12 | 21 | 14 | 25 | 8 | 15.3 | 9 | 1 | | | | 1 |
| 13 | 12 | 21 | 14 | 26 | 8 | 15.3 | 10 | | | | 1 | 1 |
| 13 | 12 | 21 | 14 | 27 | 10 | 16.3 | 10 | | | 1 | | 1 |
| 13 | 12 | 21 | 14 | 28 | 6 | 12 | 10 | 1 | | | | 1 |
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| 13 | 12 | 21 | 15 | 23 | 8 | 15.3 | 11 | 1 | | | | 1 |
| 13 | 12 | 21 | 15 | 24 | 6 | 14.3 | 10 | | | 2 | | 2 |
| 13 | 12 | 21 | 15 | 26 | 10 | 14.3 | 10 | 1 | | | | 1 |
| 13 | 12 | 21 | 15 | 26 | 11 | 15.3 | 11 | | | | 1 | 1 |
| 13 | 12 | 21 | 15 | 28 | 12 | 16.3 | 10 | 1 | | | | 1 |
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| 13 | 12 | 21 | 17 | 19 | 11 | 16.3 | 9 | | | | 1 | 1 |
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| 13 | 12 | 22 | 14 | 24 | 9 | 12 | 10 | | | | 1 | 1 |
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| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
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| 13 | 12.3 | 15 | 16 | 15 | 10 | 15.3 | 9 | | | 1 | | 1 |
| 13 | 12.3 | 15 | 16 | 23 | 6 | 11 | 10 | | 1 | | | 1 |
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| 13 | 12.3 | 16 | 16 | 24 | 6 | 15.3 | 10 | | 1 | | | 1 |
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| 13 | 12.3 | 20 | 14 | 27 | 8 | 12 | 9 | | | | 1 | 1 |
| 13 | 12.3 | 20 | 14 | 27 | 12 | 14.3 | 9 | | 1 | | | 1 |
| 13 | 12.3 | 20 | 14 | 28 | 10 | 16.3 | 9 | | | 1 | | 1 |
| 13 | 12.3 | 20 | 15 | 23 | 10 | 11 | 9 | | 1 | | | 1 |
| 13 | 12.3 | 20 | 15 | 24 | 12 | 16.3 | 9 | | | | 1 | 1 |
| 13 | 12.3 | 20 | 16 | 18 | 11 | 16.3 | 11 | | | 1 | | 1 |
| 13 | 12.3 | 20 | 16 | 19 | 10 | 12 | 11 | | | | 1 | 1 |
| 13 | 12.3 | 20 | 16 | 24 | 8 | 16.3 | 11 | | | 1 | | 1 |
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| 13 | 12.3 | 20 | 16 | 26 | 11 | 16.3 | 10 | | 1 | | | 1 |
| 13 | 12.3 | 20 | 16 | 27 | 10 | 12 | 9 | | | | 1 | 1 |
| 13 | 12.3 | 20 | 16 | 28 | 11 | 16.3 | 9 | | | 1 | | 1 |
| 13 | 12.3 | 20 | 18 | 19 | 8 | 11 | 9 | | | 1 | | 1 |
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| 13 | 12.3 | 20 | 18 | 26 | 6 | 14.3 | 10 | | | | 1 | 1 |
| 13 | 12.3 | 21 | 13 | 21 | 12 | 14.3 | 9 | | | | 1 | 1 |
| 13 | 12.3 | 21 | 13 | 25 | 11 | 14.3 | 11 | | | 1 | | 1 |
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| 13 | 12.3 | 21 | 14 | 24 | 12 | 15.3 | 10 | 1 | | | | 1 |
| 13 | 12.3 | 21 | 14 | 26 | 8 | 13 | 9 | | | | 1 | 1 |
| 13 | 12.3 | 21 | 15 | 27 | 11 | 12 | 10 | | | 1 | | 1 |
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| 13 | 12.3 | 21 | 16 | 23 | 10 | 16.3 | 10 | | 1 | | | 1 |
| 13 | 12.3 | 21 | 16 | 28 | 11 | 12 | 10 | | | 1 | | 1 |
| 13 | 12.3 | 21 | 17 | 25 | 6 | 16.3 | 11 | | | | 1 | 1 |
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| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
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| 13 | 12.3 | 22 | 15 | 26 | 8 | 16.3 | 10 | | | 1 | | 1 |
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| 13 | 13 | 18 | 17 | 25 | 9 | 13 | 11 | 1 | | | | 1 |
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| 13 | 13 | 19 | 16 | 24 | 10 | 16.3 | 9 | | | 1 | | 1 |
| 13 | 13 | 20 | 14 | 26 | 10 | 14.3 | 9 | | | 1 | | 1 |
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| 13 | 13 | 21 | 15 | 24 | 9 | 15.3 | 11 | 1 | | | | 1 |
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| 13 | 13 | 21 | 17 | 24 | 10 | 14.3 | 11 | | | 1 | | 1 |
| 13 | 13 | 21 | 17 | 26 | 6 | 12 | 9 | | | | 1 | 1 |
| 13 | 13 | 22 | 12 | 23 | 6 | 14.3 | 10 | 1 | | | | 1 |
| 13 | 13 | 22 | 16 | 20 | 10 | 16.3 | 11 | | | 1 | | 1 |
| 13 | 13 | 22 | 17 | 24 | 9 | 13.3 | 12 | | | 1 | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
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| 13 | 13.3 | 20 | 16 | 22 | 10 | 12 | 10 | | 1 | | | 1 |
| 13 | 13.3 | 20 | 16 | 25 | 10 | 12 | 8 | | | | 1 | 1 |
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| 13 | 13.3 | 21 | 13 | 26 | 8 | 14.3 | 10 | | 1 | | | 1 |
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| 13 | 13.3 | 21 | 15 | 27 | 11 | 15.3 | 9 | | | 1 | | 1 |
| 13 | 13.3 | 21 | 16 | 15 | 11 | 15.3 | 9 | | | 1 | | 1 |
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| 13 | 13.3 | 22 | 14 | 24 | 11 | 15.3 | 11 | | | 1 | | 1 |
| 13 | 13.3 | 22 | 14 | 26 | 10 | 15.3 | 11 | | | 1 | | 1 |
| 13 | 13.3 | 22 | 15 | 25 | 9 | 16.3 | 9 | | | 1 | | 1 |
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| 13 | 13.3 | 22 | 18 | 26 | 6 | 15.3 | 10 | | | 1 | | 1 |
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| 13 | 13.3 | 23 | 12 | 28 | 12 | 12 | 9 | 1 | | | | 1 |
| 13 | 13.3 | 23 | 16 | 24 | 10 | 14.3 | 11 | | | 1 | | 1 |
| 13 | 13.3 | 23 | 17 | 18 | 10 | 16.3 | 9 | | | 1 | | 1 |
| 13 | 13.3 | 23 | 17 | 25 | 12 | 10 | 9 | | | 1 | | 1 |
| 13 | 14 | 20 | 13 | 19 | 9 | 15.3 | 11 | 1 | | | | 1 |
| 13 | 14 | 20 | 13 | 23 | 10 | 9 | 12 | 1 | | | | 1 |
| 13 | 14 | 20 | 14 | 24 | 10 | 11 | 10 | 1 | | | | 1 |
| 13 | 14 | 20 | 16 | 25 | 9 | 15.3 | 10 | | 1 | | | 1 |
| 13 | 14 | 22 | 13 | 23 | 6 | 14.3 | 10 | | | 1 | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 13 | 14 | 22 | 14 | 28 | 12 | 15.3 | 9 | 1 | | | | 1 |
| 13 | 14.3 | 15 | 16 | 24 | 10 | 15.3 | 11 | | 1 | | | 1 |
| 13 | 14.3 | 20 | 16 | 17 | 12 | 13.3 | 9 | | 1 | | | 1 |
| 13 | 14.3 | 22 | 17 | 24 | 8 | 15.3 | 10 | | | 1 | | 1 |
| 13 | 16 | 22 | 16 | 25 | 8 | 14.3 | 11 | 1 | | | | 1 |
| 14 | 8 | 16 | 14 | 23 | 9 | 14 | 12 | 1 | | | | 1 |
| 14 | 8 | 19 | 11 | 21 | 9 | 13 | 10 | 1 | | | | 1 |
| 14 | 9 | 15 | 11 | 22 | 6 | 15.3 | 9 | 1 | | | | 1 |
| 14 | 9 | 15 | 14 | 28 | 7 | 11 | 10 | | | | 1 | 1 |
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| 14 | 9 | 20 | 14 | 25 | 10 | 15.3 | 9 | | | | 1 | 1 |
| 14 | 9 | 21 | 14 | 21 | 12 | 14 | 10 | 1 | | | | 1 |
| 14 | 9 | 21 | 16 | 26 | 10 | 15.3 | 11 | 1 | | | | 1 |
| 14 | 9 | 22 | 13 | 24 | 6 | 12 | 9 | 1 | | | | 1 |
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| 14 | 10 | 18 | 11 | 21 | 6 | 15.3 | 10 | 1 | | | | 1 |
| 14 | 10 | 19 | 16 | 20 | 6 | 12 | 11 | 1 | | | | 1 |
| 14 | 10 | 19 | 17 | 18 | 6 | 10 | 10 | 1 | | | | 1 |
| 14 | 10 | 20 | 13 | 25 | 9 | 14 | 10 | 1 | | | | 1 |
| 14 | 10 | 20 | 14 | 27 | 10 | 14.3 | 11 | 1 | | | | 1 |
| 14 | 10 | 20 | 15 | 22 | 8 | 13.3 | 9 | | | 1 | | 1 |
| 14 | 10 | 20 | 15 | 24 | 8 | 12 | 12 | 1 | | | | 1 |
| 14 | 10 | 20 | 16 | 21 | 8 | 15.3 | 11 | | | | 1 | 1 |
| 14 | 10 | 20 | 16 | 24 | 8 | 14.3 | 10 | | | | 1 | 1 |
| 14 | 10 | 20 | 16 | 24 | 10 | 12 | 8 | | | 1 | | 1 |
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| 14 | 10 | 22 | 13 | 25 | 6 | 12 | 9 | 1 | | | | 1 |
| 14 | 10 | 22 | 14 | 23 | 10 | 16.3 | 10 | 1 | | | | 1 |
| 14 | 10 | 22 | 14 | 27 | 6 | 14.3 | 11 | 1 | | | | 1 |
| 14 | 10 | 23 | 14 | 27 | 11 | 15.3 | 10 | | | | 1 | 1 |
| 14 | 10 | 23 | 16 | 27 | 6 | 14.3 | 11 | 1 | | | | 1 |
| 14 | 10 | 24 | 15 | 18 | 7 | 10 | 9 | 1 | | | | 1 |
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| 14 | 11 | 15 | 13 | 21 | 8 | 15.3 | 10 | 1 | | | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 14 | 11 | 15 | 13 | 21 | 9 | 12 | 12 | 1 | | | | 1 |
| 14 | 11 | 15 | 13 | 22 | 8 | 14.3 | 11 | 1 | | | | 1 |
| 14 | 11 | 15 | 13 | 24 | 8 | 15.3 | 9 | | 1 | | | 1 |
| 14 | 11 | 15 | 13 | 25 | 9 | 15.3 | 9 | 1 | | | | 1 |
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| 14 | 11 | 15 | 16 | 24 | 10 | 14.3 | 9 | | 1 | | | 1 |
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| 14 | 11 | 15 | 16 | 28 | 11 | 13.3 | 11 | | | 1 | | 1 |
| 14 | 11 | 15 | 17 | 18 | 12 | 13.3 | 11 | | | 1 | | 1 |
| 14 | 11 | 15 | 19 | 15 | 10 | 15.3 | 12 | | | 1 | | 1 |
| 14 | 11 | 16 | 11 | 24 | 9 | 13 | 11 | 1 | | | | 1 |
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| 14 | 11 | 16 | 13 | 25 | 9 | 12 | 10 | 1 | | | | 1 |
| 14 | 11 | 16 | 13 | 27 | 6 | 15.3 | 12 | 1 | | | | 1 |
| 14 | 11 | 16 | 14 | 18 | 6 | 15.3 | 11 | | | 1 | | 1 |
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| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
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| 14 | 11 | 19 | 12 | 25 | 9 | 15.3 | 11 | 1 | | | | 1 |
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| 14 | 11 | 20 | 11 | 23 | 10 | 11 | 12 | | | | 1 | 1 |
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| 14 | 11 | 20 | 16 | 22 | 8 | 15.3 | 12 | 1 | | | | 1 |
| 14 | 11 | 20 | 16 | 24 | 11 | 14.3 | 9 | | | 1 | | 1 |
| 14 | 11 | 20 | 16 | 24 | 11 | 16.3 | 10 | | | 1 | | 1 |
| 14 | 11 | 20 | 16 | 25 | 10 | 10 | 10 | | | 1 | | 1 |
| 14 | 11 | 20 | 16 | 25 | 11 | 13 | 9 | 1 | | | | 1 |
| 14 | 11 | 20 | 16 | 26 | 6 | 15.3 | 10 | 1 | | | | 1 |
| 14 | 11 | 20 | 16 | 26 | 11 | 12 | 10 | | | | 1 | 1 |
| 14 | 11 | 20 | 17 | 18 | 8 | 15.3 | 9 | | | 1 | | 1 |
| 14 | 11 | 20 | 17 | 19 | 10 | 12 | 11 | | | 1 | | 1 |
| 14 | 11 | 20 | 17 | 24 | 10 | 14.3 | 10 | | | | 1 | 1 |
| 14 | 11 | 20 | 17 | 24 | 11 | 12 | 10 | | | 1 | | 1 |
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| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
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| 14 | 11 | 21 | 12 | 26 | 6 | 14.3 | 9 | | | | 1 | 1 |
| 14 | 11 | 21 | 13 | 19 | 6 | 13 | 11 | 1 | | | | 1 |
| 14 | 11 | 21 | 13 | 23 | 10 | 14.3 | 10 | 1 | | | | 1 |
| 14 | 11 | 21 | 13 | 24 | 8 | 15.3 | 9 | 1 | | | | 1 |
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| 14 | 11 | 21 | 14 | 25 | 9 | 15.3 | 10 | | | | 1 | 1 |
| 14 | 11 | 21 | 14 | 25 | 11 | 15.3 | 9 | | | | 1 | 1 |
| 14 | 11 | 21 | 14 | 27 | 9 | 14.3 | 11 | 1 | | | | 1 |
| 14 | 11 | 21 | 14 | 27 | 10 | 14.3 | 9 | | | 1 | | 1 |
| 14 | 11 | 21 | 14 | 27 | 12 | 13 | 10 | | | 1 | | 1 |
| 14 | 11 | 21 | 14 | 28 | 9 | 15.3 | 10 | 1 | | | | 1 |
| 14 | 11 | 21 | 14 | 30 | 7 | 11 | 11 | | | | 1 | 1 |
| 14 | 11 | 21 | 14 | 31 | 8 | 13 | 10 | 1 | | | | 1 |
| 14 | 11 | 21 | 15 | 21 | 8 | 13 | 11 | 1 | | | | 1 |
| 14 | 11 | 21 | 15 | 22 | 8 | 16.3 | 9 | | | 1 | | 1 |
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| 14 | 11 | 21 | 15 | 25 | 8 | 16.3 | 11 | | 1 | | | 1 |
| 14 | 11 | 21 | 15 | 25 | 9 | 12 | 11 | | 1 | | | 1 |
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| 14 | 11 | 21 | 16 | 18 | 10 | 14.3 | 9 | | | 1 | | 1 |
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| 14 | 11 | 21 | 17 | 26 | 10 | 13 | 11 | | 1 | | | 1 |
| 14 | 11 | 21 | 17 | 27 | 6 | 13 | 9 | | 1 | | | 1 |
| 14 | 11 | 21 | 18 | 15 | 10 | 14.3 | 11 | | 1 | | | 1 |
| 14 | 11 | 21 | 18 | 24 | 11 | 11 | 10 | | | | 1 | 1 |
| 14 | 11 | 21 | 19 | 25 | 11 | 15.3 | 11 | | | | 1 | 1 |
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| 14 | 11 | 22 | 13 | 25 | 6 | 15.3 | 9 | | | | 1 | 1 |
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| 14 | 11 | 22 | 13 | 27 | 9 | 12 | 11 | 1 | | | | 1 |
| 14 | 11 | 22 | 14 | 21 | 9 | 15.3 | 12 | 1 | | | | 1 |
| 14 | 11 | 22 | 14 | 22 | 6 | 15.3 | 12 | 1 | | | | 1 |
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| 14 | 11 | 22 | 14 | 28 | 9 | 13 | 11 | 1 | | | | 1 |
| 14 | 11 | 22 | 14 | 28 | 10 | 15.3 | 11 | | 1 | | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 14 | 11 | 22 | 14 | 29 | 8 | 15.3 | 11 | | 1 | | | 1 |
| 14 | 11 | 22 | 15 | 24 | 11 | 13 | 9 | | | | 1 | 1 |
| 14 | 11 | 22 | 15 | 24 | 11 | 15.3 | 10 | | | | 1 | 1 |
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| 14 | 11 | 23 | 16 | 27 | 6 | 12 | 12 | 1 | | | | 1 |
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| 14 | 11.3 | 16 | 13 | 26 | 12 | 15.3 | 9 | | | | 1 | 1 |
| 14 | 11.3 | 16 | 15 | 28 | 6 | 15.3 | 11 | | 1 | | | 1 |
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| 14 | 11.3 | 20 | 13 | 24 | 6 | 15.3 | 11 | 1 | | | | 1 |
| 14 | 11.3 | 20 | 13 | 26 | 11 | 12 | 10 | | | | 1 | 1 |
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| 14 | 11.3 | 20 | 16 | 23 | 8 | 14.3 | 10 | | | 1 | | 1 |
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| 14 | 11.3 | 20 | 16 | 27 | 10 | 15.3 | 11 | | | 1 | | 1 |
| 14 | 11.3 | 20 | 17 | 19 | 8 | 16.3 | 10 | | | | 1 | 1 |
| 14 | 11.3 | 20 | 17 | 24 | 11 | 13.3 | 10 | | | 1 | | 1 |
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| 14 | 11.3 | 21 | 18 | 24 | 11 | 13 | 10 | | | | 1 | 1 |
| 14 | 11.3 | 22 | 17 | 24 | 11 | 15.3 | 11 | | | | 1 | 1 |
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| 14 | 12 | 15 | 12 | 25 | 7 | 14.3 | 11 | 1 | | | | 1 |
| 14 | 12 | 15 | 12 | 28 | 8 | 15.3 | 10 | 1 | | | | 1 |
| 14 | 12 | 15 | 13 | 21 | 11 | 10 | 11 | 1 | | | | 1 |
| 14 | 12 | 15 | 13 | 25 | 11 | 15.3 | 9 | | 1 | | | 1 |
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| 14 | 12 | 15 | 13 | 27 | 10 | 11 | 11 | 1 | | | | 1 |
| 14 | 12 | 15 | 13 | 29 | 7 | 12 | 11 | 1 | | | | 1 |
| 14 | 12 | 15 | 14 | 18 | 7 | 14.3 | 10 | 1 | | | | 1 |
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| 14 | 12 | 15 | 15 | 25 | 6 | 16.3 | 9 | | | 1 | | 1 |
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| 14 | 12 | 15 | 16 | 22 | 9 | 13.3 | 11 | 1 | | | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
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| 14 | 12 | 15 | 16 | 24 | 6 | 11 | 10 | 1 | | | | 1 |
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| 14 | 12 | 16 | 15 | 21 | 6 | 13 | 11 | 1 | | | | 1 |
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| 14 | 12 | 16 | 15 | 27 | 11 | 16.3 | 9 | 1 | | | | 1 |
| 14 | 12 | 16 | 16 | 25 | 11 | 16.3 | 11 | | 1 | | | 1 |
| 14 | 12 | 16 | 16 | 29 | 10 | 16.3 | 10 | | 1 | | | 1 |
| 14 | 12 | 16 | 17 | 27 | 11 | 15.3 | 11 | | | 1 | | 1 |
| 14 | 12 | 19 | 13 | 26 | 12 | 12 | 11 | 1 | | | | 1 |
| 14 | 12 | 19 | 13 | 28 | 10 | 15.3 | 10 | | | | 1 | 1 |
| 14 | 12 | 19 | 14 | 18 | 6 | 15.3 | 10 | | | | 1 | 1 |
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| 14 | 12 | 20 | 13 | 28 | 6 | 12 | 9 | | | 1 | | 1 |
| 14 | 12 | 20 | 14 | 23 | 10 | 15.3 | 9 | | | | 1 | 1 |
| 14 | 12 | 20 | 14 | 24 | 11 | 12 | 9 | | | | 1 | 1 |
| 14 | 12 | 20 | 14 | 24 | 11 | 14.3 | 10 | | | 1 | | 1 |
| 14 | 12 | 20 | 14 | 24 | 11 | 15.3 | 10 | | 1 | | | 1 |
| 14 | 12 | 20 | 14 | 25 | 9 | 12 | 10 | 1 | | | | 1 |
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| 14 | 12 | 20 | 15 | 25 | 11 | 14.3 | 9 | | | 1 | | 1 |
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| 14 | 12 | 20 | 15 | 28 | 10 | 14.3 | 12 | | | 1 | | 1 |
| 14 | 12 | 20 | 16 | 19 | 8 | 15.3 | 11 | | | 1 | | 1 |
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| 14 | 12 | 20 | 16 | 21 | 11 | 15.3 | 11 | | | 1 | | 1 |
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| 14 | 12 | 20 | 16 | 25 | 9 | 14.3 | 10 | | | 1 | | 1 |
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| 14 | 12 | 20 | 16 | 26 | 11 | 14.3 | 10 | | 1 | | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 14 | 12 | 20 | 16 | 27 | 6 | 16.3 | 11 | 1 | | | | 1 |
| 14 | 12 | 20 | 16 | 27 | 11 | 12 | 10 | | | 1 | | 1 |
| 14 | 12 | 20 | 17 | 24 | 8 | 13 | 12 | | 1 | | | 1 |
| 14 | 12 | 20 | 17 | 25 | 11 | 14.3 | 11 | | | | 1 | 1 |
| 14 | 12 | 20 | 18 | 25 | 8 | 14 | 10 | | | | 1 | 1 |
| 14 | 12 | 20 | 19 | 15 | 10 | 14.3 | 10 | | 1 | | | 1 |
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| 14 | 12 | 21 | 13 | 25 | 10 | 15.3 | 11 | | | 1 | | 1 |
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| 14 | 12 | 21 | 14 | 24 | 10 | 14.3 | 11 | | | | 1 | 1 |
| 14 | 12 | 21 | 14 | 24 | 11 | 14.3 | 11 | | | | 1 | 1 |
| 14 | 12 | 21 | 14 | 25 | 6 | 13.3 | 11 | | | 1 | | 1 |
| 14 | 12 | 21 | 14 | 26 | 11 | 15.3 | 9 | | | 1 | | 1 |
| 14 | 12 | 21 | 14 | 27 | 6 | 12 | 9 | | | 1 | | 1 |
| 14 | 12 | 21 | 14 | 31 | 10 | 12 | 10 | | | 1 | | 1 |
| 14 | 12 | 21 | 15 | 15 | 6 | 15.3 | 11 | | | | 1 | 1 |
| 14 | 12 | 21 | 15 | 18 | 11 | 14.3 | 12 | 1 | | | | 1 |
| 14 | 12 | 21 | 15 | 22 | 9 | 12 | 11 | | | | 1 | 1 |
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| 14 | 12 | 21 | 15 | 24 | 12 | 15.3 | 10 | | | 1 | | 1 |
| 14 | 12 | 21 | 15 | 25 | 11 | 15.3 | 10 | | 1 | | | 1 |
| 14 | 12 | 21 | 15 | 26 | 8 | 15.3 | 9 | | | 1 | | 1 |
| 14 | 12 | 21 | 15 | 26 | 10 | 14.3 | 9 | 1 | | | | 1 |
| 14 | 12 | 21 | 15 | 26 | 12 | 16.3 | 9 | | 1 | | | 1 |
| 14 | 12 | 21 | 15 | 27 | 12 | 15.3 | 9 | | | | 1 | 1 |
| 14 | 12 | 21 | 16 | 13 | 10 | 12 | 11 | | | | 1 | 1 |
| 14 | 12 | 21 | 16 | 18 | 10 | 14.3 | 10 | 1 | | | | 1 |
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| 14 | 12 | 22 | 13 | 23 | 10 | 16.3 | 10 | | 1 | | | 1 |
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| 14 | 12 | 22 | 14 | 26 | 12 | 16.3 | 10 | | | 1 | | 1 |
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| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
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| 14 | 12 | 22 | 15 | 26 | 10 | 12 | 9 | 1 | | | | 1 |
| 14 | 12 | 22 | 15 | 28 | 10 | 15.3 | 11 | | | | 1 | 1 |
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| 14 | 12 | 22 | 17 | 20 | 10 | 15.3 | 10 | | | 1 | | 1 |
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| 14 | 12.3 | 15 | 12 | 26 | 12 | 12 | 9 | | | 1 | | 1 |
| 14 | 12.3 | 15 | 13 | 23 | 10 | 13 | 11 | | 1 | | | 1 |
| 14 | 12.3 | 15 | 13 | 27 | 11 | 12 | 11 | | | 1 | | 1 |
| 14 | 12.3 | 15 | 15 | 20 | 10 | 15.3 | 9 | | | 1 | | 1 |
| 14 | 12.3 | 15 | 15 | 23 | 10 | 16.3 | 10 | | | 1 | | 1 |
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| 14 | 12.3 | 16 | 15 | 27 | 11 | 11 | 10 | | 1 | | | 1 |
| 14 | 12.3 | 16 | 15 | 27 | 11 | 13 | 9 | | 1 | | | 1 |
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| 14 | 12.3 | 16 | 16 | 22 | 10 | 11 | 11 | | 1 | | | 1 |
| 14 | 12.3 | 16 | 16 | 24 | 9 | 12 | 9 | 1 | | | | 1 |
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| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
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| 14 | 12.3 | 17 | 13 | 26 | 9 | 12 | 11 | 1 | | | | 1 |
| 14 | 12.3 | 19 | 14 | 23 | 10 | 12 | 10 | | | | 1 | 1 |
| 14 | 12.3 | 19 | 17 | 25 | 11 | 14 | 9 | | | 1 | | 1 |
| 14 | 12.3 | 20 | 12 | 25 | 11 | 15.3 | 9 | | | 1 | | 1 |
| 14 | 12.3 | 20 | 13 | 24 | 8 | 15.3 | 11 | | 1 | | | 1 |
| 14 | 12.3 | 20 | 13 | 25 | 11 | 15.3 | 10 | | | | 1 | 1 |
| 14 | 12.3 | 20 | 14 | 26 | 8 | 14.3 | 9 | | | | 1 | 1 |
| 14 | 12.3 | 20 | 15 | 20 | 10 | 15.3 | 11 | | | | 1 | 1 |
| 14 | 12.3 | 20 | 15 | 24 | 10 | 12 | 9 | | 1 | | | 1 |
| 14 | 12.3 | 20 | 15 | 25 | 11 | 14.3 | 11 | | | 1 | | 1 |
| 14 | 12.3 | 20 | 15 | 26 | 11 | 12 | 10 | | | | 1 | 1 |
| 14 | 12.3 | 20 | 15 | 26 | 12 | 14.3 | 11 | | | | 1 | 1 |
| 14 | 12.3 | 20 | 16 | 18 | 10 | 14.3 | 11 | | | | 1 | 1 |
| 14 | 12.3 | 20 | 16 | 20 | 10 | 15.3 | 10 | | | | 1 | 1 |
| 14 | 12.3 | 20 | 16 | 20 | 11 | 15.3 | 9 | | | | 1 | 1 |
| 14 | 12.3 | 20 | 16 | 21 | 8 | 11 | 9 | | | | 1 | 1 |
| 14 | 12.3 | 20 | 16 | 23 | 10 | 13 | 10 | | 1 | | | 1 |
| 14 | 12.3 | 20 | 16 | 24 | 6 | 12 | 10 | 1 | | | | 1 |
| 14 | 12.3 | 20 | 16 | 24 | 6 | 16.3 | 11 | | | | 1 | 1 |
| 14 | 12.3 | 20 | 16 | 24 | 10 | 15.3 | 11 | 1 | | | | 1 |
| 14 | 12.3 | 20 | 16 | 24 | 12 | 15.3 | 10 | 1 | | | | 1 |
| 14 | 12.3 | 20 | 16 | 25 | 10 | 14.3 | 11 | | | 1 | | 1 |
| 14 | 12.3 | 20 | 16 | 25 | 10 | 15.3 | 11 | | 1 | | | 1 |
| 14 | 12.3 | 20 | 16 | 26 | 6 | 12 | 10 | | | 1 | | 1 |
| 14 | 12.3 | 20 | 16 | 26 | 8 | 15.3 | 10 | | | | 1 | 1 |
| 14 | 12.3 | 20 | 16 | 27 | 10 | 12 | 11 | | | | 1 | 1 |
| 14 | 12.3 | 20 | 16 | 27 | 10 | 13 | 10 | | | | 1 | 1 |
| 14 | 12.3 | 20 | 16 | 27 | 12 | 15.3 | 11 | 1 | | | | 1 |
| 14 | 12.3 | 20 | 17 | 18 | 8 | 15.3 | 9 | | | | 1 | 1 |
| 14 | 12.3 | 20 | 17 | 24 | 10 | 12 | 10 | | 1 | | | 1 |
| 14 | 12.3 | 20 | 20 | 18 | 10 | 14.3 | 9 | 1 | | | | 1 |
| 14 | 12.3 | 21 | 11 | 23 | 6 | 15.3 | 9 | | | 1 | | 1 |
| 14 | 12.3 | 21 | 14 | 20 | 10 | 15.3 | 9 | | 1 | | | 1 |
| 14 | 12.3 | 21 | 14 | 22 | 8 | 10 | 11 | 1 | | | | 1 |
| 14 | 12.3 | 21 | 14 | 27 | 6 | 13 | 10 | | | 1 | | 1 |
| 14 | 12.3 | 21 | 14 | 28 | 11 | 12 | 10 | | | 1 | | 1 |
| 14 | 12.3 | 21 | 15 | 20 | 8 | 10 | 11 | 1 | | | | 1 |
| 14 | 12.3 | 21 | 15 | 22 | 11 | 15.3 | 11 | | | | 1 | 1 |
| 14 | 12.3 | 21 | 15 | 23 | 8 | 15.3 | 9 | | | 1 | | 1 |
| 14 | 12.3 | 21 | 15 | 23 | 11 | 15.3 | 9 | | 1 | | | 1 |
| 14 | 12.3 | 21 | 15 | 24 | 6 | 14.3 | 10 | | | 1 | | 1 |
| 14 | 12.3 | 21 | 15 | 25 | 12 | 16.3 | 11 | | | | 1 | 1 |
| 14 | 12.3 | 21 | 16 | 15 | 8 | 15.3 | 10 | | 1 | | | 1 |
| 14 | 12.3 | 21 | 16 | 18 | 9 | 15.3 | 10 | | | | 1 | 1 |
| 14 | 12.3 | 21 | 16 | 18 | 10 | 12 | 10 | | | 1 | | 1 |
| 14 | 12.3 | 21 | 16 | 19 | 10 | 15.3 | 11 | | 1 | | | 1 |
| 14 | 12.3 | 21 | 16 | 20 | 6 | 15.3 | 10 | | | | 1 | 1 |
| 14 | 12.3 | 21 | 16 | 21 | 10 | 15.3 | 11 | | 1 | | | 1 |
| 14 | 12.3 | 21 | 16 | 21 | 12 | 15.3 | 11 | 1 | | | | 1 |
| 14 | 12.3 | 21 | 16 | 24 | 10 | 15.3 | 9 | | 1 | | | 1 |
| 14 | 12.3 | 21 | 16 | 24 | 12 | 15.3 | 10 | | | | 1 | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 14 | 12.3 | 21 | 17 | 18 | 11 | 15.3 | 10 | | | | 1 | 1 |
| 14 | 12.3 | 22 | 12 | 24 | 10 | 15.3 | 10 | | | | 1 | 1 |
| 14 | 12.3 | 22 | 13 | 24 | 8 | 11 | 11 | | 1 | | | 1 |
| 14 | 12.3 | 22 | 14 | 25 | 8 | 14.3 | 10 | | | 1 | | 1 |
| 14 | 12.3 | 22 | 14 | 26 | 10 | 13 | 9 | | 1 | | | 1 |
| 14 | 12.3 | 22 | 15 | 25 | 8 | 11 | 11 | | | | 1 | 1 |
| 14 | 12.3 | 22 | 15 | 26 | 13 | 14.3 | 11 | | | 1 | | 1 |
| 14 | 12.3 | 22 | 16 | 18 | 10 | 15.3 | 11 | | | | 1 | 1 |
| 14 | 12.3 | 22 | 16 | 24 | 11 | 14.3 | 10 | 1 | | | | 1 |
| 14 | 12.3 | 22 | 16 | 24 | 13 | 14.3 | 11 | | 1 | | | 1 |
| 14 | 12.3 | 22 | 16 | 25 | 8 | 15.3 | 11 | | | 1 | | 1 |
| 14 | 12.3 | 22 | 17 | 18 | 10 | 13 | 11 | 1 | | | | 1 |
| 14 | 12.3 | 22 | 17 | 18 | 10 | 15.3 | 10 | | | | 1 | 1 |
| 14 | 12.3 | 22 | 17 | 23 | 8 | 15.3 | 11 | | | 1 | | 1 |
| 14 | 12.3 | 23 | 15 | 26 | 12 | 14.3 | 11 | | | 1 | | 1 |
| 14 | 12.3 | 23 | 16 | 22 | 10 | 15.3 | 10 | | 1 | | | 1 |
| 14 | 12.3 | 23 | 16 | 24 | 10 | 14.3 | 9 | | | | 1 | 1 |
| 14 | 12.3 | 24 | 15 | 24 | 6 | 12 | 9 | | | 1 | | 1 |
| 14 | 12.3 | 24 | 15 | 24 | 11 | 15.3 | 10 | | | 1 | | 1 |
| 14 | 12.3 | 24 | 18 | 18 | 10 | 11 | 10 | | | | 1 | 1 |
| 14 | 13 | 14 | 16 | 25 | 6 | 14.3 | 11 | | 1 | | | 1 |
| 14 | 13 | 15 | 11 | 23 | 9 | 13 | 11 | 1 | | | | 1 |
| 14 | 13 | 15 | 13 | 25 | 7 | 13 | 12 | 1 | | | | 1 |
| 14 | 13 | 15 | 14 | 27 | 10 | 15.3 | 11 | 1 | | | | 1 |
| 14 | 13 | 15 | 15 | 24 | 7 | 13 | 11 | 1 | | | | 1 |
| 14 | 13 | 15 | 16 | 21 | 6 | 14.3 | 10 | | | | 1 | 1 |
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| 14 | 13 | 19 | 15 | 21 | 9 | 16.3 | 9 | 1 | | | | 1 |
| 14 | 13 | 20 | 13 | 20 | 9 | 14.3 | 10 | 1 | | | | 1 |
| 14 | 13 | 20 | 13 | 25 | 6 | 15.3 | 11 | | | 1 | | 1 |
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| 14 | 13 | 20 | 15 | 25 | 12 | 15.3 | 9 | | | 1 | | 1 |
| 14 | 13 | 20 | 15 | 26 | 8 | 16.3 | 10 | | | 1 | | 1 |
| 14 | 13 | 20 | 15 | 26 | 12 | 16.3 | 9 | | | 1 | | 1 |
| 14 | 13 | 20 | 15 | 27 | 11 | 16.3 | 11 | | 1 | | | 1 |
| 14 | 13 | 20 | 16 | 15 | 11 | 11 | 10 | | | 1 | | 1 |
| 14 | 13 | 20 | 16 | 15 | 12 | 15.3 | 10 | | | | 1 | 1 |
| 14 | 13 | 20 | 16 | 17 | 6 | 15.3 | 10 | | | 1 | | 1 |
| 14 | 13 | 20 | 16 | 21 | 8 | 14.3 | 9 | | | 1 | | 1 |
| 14 | 13 | 20 | 16 | 24 | 11 | 15.3 | 10 | | | | 1 | 1 |
| 14 | 13 | 20 | 16 | 25 | 8 | 16.3 | 11 | | | 1 | | 1 |
| 14 | 13 | 20 | 16 | 25 | 11 | 15.3 | 9 | | | 1 | | 1 |
| 14 | 13 | 20 | 17 | 18 | 11 | 15.3 | 10 | | | | 1 | 1 |
| 14 | 13 | 20 | 17 | 24 | 10 | 15.3 | 10 | | | 1 | | 1 |
| 14 | 13 | 21 | 11 | 21 | 9 | 11 | 10 | 1 | | | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 14 | 13 | 21 | 11 | 23 | 12 | 12 | 11 | 1 | | | | 1 |
| 14 | 13 | 21 | 13 | 19 | 6 | 15.3 | 9 | | 1 | | | 1 |
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| 14 | 13 | 21 | 13 | 25 | 11 | 12 | 10 | 1 | | | | 1 |
| 14 | 13 | 21 | 13 | 26 | 10 | 15.3 | 11 | 1 | | | | 1 |
| 14 | 13 | 21 | 13 | 27 | 10 | 16.3 | 10 | | | 1 | | 1 |
| 14 | 13 | 21 | 14 | 26 | 6 | 16.3 | 11 | | | 1 | | 1 |
| 14 | 13 | 21 | 14 | 29 | 10 | 15.3 | 10 | | | 1 | | 1 |
| 14 | 13 | 21 | 15 | 18 | 12 | 13.3 | 11 | 1 | | | | 1 |
| 14 | 13 | 21 | 15 | 24 | 6 | 14.3 | 9 | | | 1 | | 1 |
| 14 | 13 | 21 | 15 | 25 | 11 | 15.3 | 10 | | | 1 | | 1 |
| 14 | 13 | 21 | 15 | 26 | 6 | 16.3 | 8 | 1 | | | | 1 |
| 14 | 13 | 21 | 16 | 21 | 10 | 15.3 | 11 | | | | 1 | 1 |
| 14 | 13 | 21 | 16 | 22 | 10 | 16.3 | 10 | | | 1 | | 1 |
| 14 | 13 | 21 | 16 | 24 | 10 | 14.3 | 10 | | | | 1 | 1 |
| 14 | 13 | 21 | 16 | 27 | 10 | 14.3 | 12 | | | 1 | | 1 |
| 14 | 13 | 21 | 17 | 18 | 10 | 15.3 | 10 | 1 | | | | 1 |
| 14 | 13 | 21 | 17 | 18 | 12 | 14.3 | 10 | | | 1 | | 1 |
| 14 | 13 | 21 | 17 | 18 | 12 | 16.3 | 12 | | | 1 | | 1 |
| 14 | 13 | 21 | 17 | 26 | 9 | 14.3 | 10 | | 1 | | | 1 |
| 14 | 13 | 21 | 17 | 26 | 11 | 15.3 | 11 | 1 | | | | 1 |
| 14 | 13 | 21 | 18 | 25 | 8 | 15.3 | 9 | | | | 1 | 1 |
| 14 | 13 | 21 | 18 | 32 | 7 | 14.3 | 11 | 1 | | | | 1 |
| 14 | 13 | 22 | 14 | 19 | 8 | 13 | 10 | | | 1 | | 1 |
| 14 | 13 | 22 | 14 | 20 | 9 | 13 | 11 | 1 | | | | 1 |
| 14 | 13 | 22 | 14 | 24 | 8 | 15.3 | 10 | | | 1 | | 1 |
| 14 | 13 | 22 | 14 | 24 | 11 | 15.3 | 9 | 1 | | | | 1 |
| 14 | 13 | 22 | 14 | 27 | 9 | 12 | 11 | 1 | | | | 1 |
| 14 | 13 | 22 | 15 | 25 | 11 | 15.3 | 10 | | | 1 | | 1 |
| 14 | 13 | 22 | 15 | 26 | 8 | 16.3 | 11 | | 1 | | | 1 |
| 14 | 13 | 22 | 17 | 18 | 10 | 13.3 | 10 | | | 1 | | 1 |
| 14 | 13 | 22 | 17 | 21 | 12 | 14.3 | 10 | | | 1 | | 1 |
| 14 | 13 | 23 | 12 | 27 | 8 | 15.3 | 11 | 1 | | | | 1 |
| 14 | 13 | 23 | 14 | 28 | 6 | 14.3 | 11 | | | | 1 | 1 |
| 14 | 13 | 23 | 15 | 18 | 8 | 11 | 10 | 1 | | | | 1 |
| 14 | 13 | 23 | 17 | 28 | 11 | 15.3 | 11 | | | 1 | | 1 |
| 14 | 13.3 | 16 | 14 | 26 | 11 | 15.3 | 10 | | 1 | | | 1 |
| 14 | 13.3 | 16 | 14 | 27 | 9 | 15.3 | 9 | | 1 | | | 1 |
| 14 | 13.3 | 16 | 15 | 15 | 11 | 14 | 9 | | 1 | | | 1 |
| 14 | 13.3 | 16 | 15 | 23 | 8 | 12 | 11 | 1 | | | | 1 |
| 14 | 13.3 | 16 | 15 | 27 | 10 | 15.3 | 11 | | 1 | | | 1 |
| 14 | 13.3 | 17 | 15 | 27 | 11 | 11 | 10 | | 1 | | | 1 |
| 14 | 13.3 | 19 | 13 | 25 | 8 | 14 | 12 | | | 1 | | 1 |
| 14 | 13.3 | 20 | 12 | 26 | 7 | 14.3 | 9 | | 1 | | | 1 |
| 14 | 13.3 | 20 | 12 | 30 | 12 | 15.3 | 10 | | | 1 | | 1 |
| 14 | 13.3 | 20 | 13 | 19 | 11 | 14.3 | 9 | | | | 1 | 1 |
| 14 | 13.3 | 20 | 14 | 24 | 6 | 14.3 | 10 | | | | 1 | 1 |
| 14 | 13.3 | 20 | 14 | 24 | 6 | 15.3 | 11 | | | | 1 | 1 |
| 14 | 13.3 | 20 | 14 | 26 | 10 | 14.3 | 11 | | | 1 | | 1 |
| 14 | 13.3 | 20 | 14 | 27 | 8 | 11 | 10 | | | 1 | | 1 |
| 14 | 13.3 | 20 | 14 | 27 | 10 | 15.3 | 10 | | | 1 | | 1 |
| 14 | 13.3 | 20 | 15 | 23 | 10 | 15.3 | 11 | | | 1 | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 14 | 13.3 | 20 | 15 | 26 | 10 | 11 | 9 | | | 1 | | 1 |
| 14 | 13.3 | 20 | 15 | 28 | 6 | 15.3 | 10 | | | 1 | | 1 |
| 14 | 13.3 | 20 | 15 | 28 | 8 | 12 | 11 | | 1 | | | 1 |
| 14 | 13.3 | 20 | 15 | 28 | 9 | 16.3 | 10 | | | | 1 | 1 |
| 14 | 13.3 | 20 | 16 | 19 | 8 | 16.3 | 9 | | | 1 | | 1 |
| 14 | 13.3 | 20 | 16 | 20 | 11 | 14.3 | 9 | | | 1 | | 1 |
| 14 | 13.3 | 20 | 16 | 27 | 12 | 15.3 | 10 | | | | 1 | 1 |
| 14 | 13.3 | 20 | 17 | 24 | 11 | 15.3 | 13 | | | | 1 | 1 |
| 14 | 13.3 | 21 | 10 | 29 | 8 | 15.3 | 9 | | | | 1 | 1 |
| 14 | 13.3 | 21 | 13 | 24 | 8 | 16.3 | 9 | | | 1 | | 1 |
| 14 | 13.3 | 21 | 13 | 24 | 11 | 14.3 | 11 | | | 1 | | 1 |
| 14 | 13.3 | 21 | 13 | 25 | 11 | 15.3 | 10 | | | 1 | | 1 |
| 14 | 13.3 | 21 | 14 | 24 | 8 | 15.3 | 10 | | | | 1 | 1 |
| 14 | 13.3 | 21 | 14 | 25 | 10 | 14.3 | 11 | | | 1 | | 1 |
| 14 | 13.3 | 21 | 14 | 26 | 12 | 15.3 | 11 | | | 1 | | 1 |
| 14 | 13.3 | 21 | 14 | 27 | 9 | 13 | 11 | 1 | | | | 1 |
| 14 | 13.3 | 21 | 15 | 15 | 10 | 15.3 | 8 | | | 1 | | 1 |
| 14 | 13.3 | 21 | 16 | 14 | 12 | 15.3 | 10 | | | | 1 | 1 |
| 14 | 13.3 | 21 | 16 | 19 | 8 | 12 | 9 | | | 1 | | 1 |
| 14 | 13.3 | 21 | 17 | 25 | 10 | 13.3 | 9 | | 1 | | | 1 |
| 14 | 13.3 | 21 | 17 | 25 | 10 | 14.3 | 8 | | 1 | | | 1 |
| 14 | 13.3 | 22 | 13 | 23 | 10 | 15.3 | 10 | | | 1 | | 1 |
| 14 | 13.3 | 22 | 14 | 29 | 8 | 14.3 | 10 | | 1 | | | 1 |
| 14 | 13.3 | 22 | 14 | 29 | 11 | 16.3 | 9 | | | 1 | | 1 |
| 14 | 13.3 | 22 | 15 | 25 | 12 | 13 | 11 | | 1 | | | 1 |
| 14 | 13.3 | 22 | 16 | 26 | 11 | 15.3 | 10 | | | 1 | | 1 |
| 14 | 13.3 | 22 | 16 | 30 | 6 | 15.3 | 11 | | | 1 | | 1 |
| 14 | 13.3 | 23 | 15 | 26 | 10 | 16.3 | 9 | | 1 | | | 1 |
| 14 | 14 | 15 | 14 | 21 | 9 | 13 | 11 | 1 | | | | 1 |
| 14 | 14 | 15 | 15 | 25 | 6 | 12 | 10 | 1 | | | | 1 |
| 14 | 14 | 16 | 13 | 27 | 11 | 15.3 | 9 | | 1 | | | 1 |
| 14 | 14 | 16 | 15 | 25 | 11 | 13 | 11 | 1 | | | | 1 |
| 14 | 14 | 17 | 16 | 27 | 10 | 12 | 9 | 1 | | | | 1 |
| 14 | 14 | 21 | 14 | 24 | 10 | 11 | 9 | 1 | | | | 1 |
| 14 | 14 | 21 | 16 | 19 | 9 | 12 | 11 | | | | 1 | 1 |
| 14 | 14.3 | 21 | 14 | 24 | 6 | 15.3 | 10 | | | | 1 | 1 |
| 14 | 14.3 | 21 | 15 | 23 | 10 | 11 | 11 | | 1 | | | 1 |
| 14 | 15.3 | 20 | 13 | 25 | 10 | 12 | 11 | | | 1 | | 1 |
| 15 | 7 | 21 | 14 | 30 | 9 | 13 | 10 | | | | 1 | 1 |
| 15 | 8 | 18 | 15 | 20 | 9 | 11 | 11 | 1 | | | | 1 |
| 15 | 8 | 20 | 15 | 26 | 11 | 14.3 | 9 | 1 | | | | 1 |
| 15 | 9 | 15 | 13 | 19 | 7 | 11 | 11 | 1 | | | | 1 |
| 15 | 9 | 15 | 13 | 21 | 9 | 13 | 11 | 1 | | | | 1 |
| 15 | 9 | 15 | 14 | 22 | 8 | 14.3 | 11 | 1 | | | | 1 |
| 15 | 9 | 15 | 14 | 27 | 10 | 15.3 | 10 | 1 | | | | 1 |
| 15 | 9 | 15 | 14 | 29 | 8 | 13 | 10 | 1 | | | | 1 |
| 15 | 9 | 15 | 15 | 21 | 11 | 15.3 | 12 | 1 | | | | 1 |
| 15 | 9 | 19 | 12 | 24 | 6 | 12 | 10 | 1 | | | | 1 |
| 15 | 9 | 21 | 13 | 26 | 9 | 10 | 12 | 1 | | | | 1 |
| 15 | 9 | 21 | 18 | 27 | 8 | 15.3 | 10 | 1 | | | | 1 |
| 15 | 9.3 | 20 | 13 | 26 | 8 | 12 | 10 | | | 1 | | 1 |
| 15 | 10 | 15 | 13 | 22 | 6 | 14.3 | 12 | 1 | | | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 15 | 10 | 15 | 14 | 26 | 9 | 15.3 | 10 | 1 | | | | 1 |
| 15 | 10 | 15 | 14 | 30 | 8 | 12 | 9 | 1 | | | | 1 |
| 15 | 10 | 15 | 15 | 20 | 11 | 16.3 | 9 | | 1 | | | 1 |
| 15 | 10 | 15 | 16 | 25 | 10 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 10 | 16 | 10 | 20 | 11 | 12 | 10 | 1 | | | | 1 |
| 15 | 10 | 16 | 13 | 21 | 9 | 12 | 11 | 1 | | | | 1 |
| 15 | 10 | 16 | 14 | 16 | 11 | 15.3 | 10 | 1 | | | | 1 |
| 15 | 10 | 16 | 14 | 19 | 11 | 12 | 11 | 1 | | | | 1 |
| 15 | 10 | 16 | 14 | 21 | 9 | 14 | 11 | 1 | | | | 1 |
| 15 | 10 | 19 | 13 | 28 | 10 | 13 | 10 | 1 | | | | 1 |
| 15 | 10 | 20 | 14 | 24 | 8 | 13.3 | 9 | | | 1 | | 1 |
| 15 | 10 | 20 | 16 | 18 | 11 | 16.3 | 11 | | | 1 | | 1 |
| 15 | 10 | 21 | 13 | 19 | 9 | 13 | 12 | 1 | | | | 1 |
| 15 | 10 | 21 | 13 | 26 | 10 | 12 | 11 | 1 | | | | 1 |
| 15 | 10 | 21 | 14 | 26 | 11 | 14.3 | 10 | | | | 1 | 1 |
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| 15 | 10 | 21 | 16 | 20 | 9 | 13 | 9 | 1 | | | | 1 |
| 15 | 10 | 21 | 16 | 25 | 10 | 15.3 | 9 | | 1 | | | 1 |
| 15 | 10 | 22 | 11 | 21 | 11 | 14.3 | 8 | 1 | | | | 1 |
| 15 | 10 | 22 | 14 | 23 | 10 | 14.3 | 11 | | | 1 | | 1 |
| 15 | 10 | 22 | 14 | 27 | 11 | 12 | 10 | | | 1 | | 1 |
| 15 | 10 | 22 | 15 | 20 | 8 | 12 | 10 | 1 | | | | 1 |
| 15 | 10 | 22 | 15 | 22 | 9 | 15.3 | 9 | | | 1 | | 1 |
| 15 | 10 | 22 | 17 | 26 | 10 | 14.3 | 12 | | | | 1 | 1 |
| 15 | 10 | 23 | 13 | 19 | 9 | 13 | 11 | 1 | | | | 1 |
| 15 | 10 | 23 | 13 | 19 | 11 | 11 | 10 | | | | 1 | 1 |
| 15 | 10 | 23 | 14 | 28 | 10 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 10 | 23 | 15 | 27 | 12 | 11 | 10 | | | | 1 | 1 |
| 15 | 11 | 14 | 14 | 27 | 10 | 15.3 | 11 | | 1 | | | 1 |
| 15 | 11 | 14 | 15 | 24 | 11 | 15.3 | 9 | | | 1 | | 1 |
| 15 | 11 | 15 | 11 | 23 | 10 | 16.3 | 10 | | | | 1 | 1 |
| 15 | 11 | 15 | 12 | 23 | 8 | 13.3 | 9 | 1 | | | | 1 |
| 15 | 11 | 15 | 13 | 19 | 10 | 13 | 10 | | | | 1 | 1 |
| 15 | 11 | 15 | 13 | 19 | 12 | 13 | 11 | 1 | | | | 1 |
| 15 | 11 | 15 | 13 | 28 | 9 | 16.3 | 10 | 1 | | | | 1 |
| 15 | 11 | 15 | 14 | 19 | 11 | 11 | 11 | 1 | | | | 1 |
| 15 | 11 | 15 | 14 | 22 | 11 | 12 | 9 | 1 | | | | 1 |
| 15 | 11 | 15 | 14 | 24 | 7 | 12 | 10 | 1 | | | | 1 |
| 15 | 11 | 15 | 14 | 28 | 6 | 14.3 | 10 | 1 | | | | 1 |
| 15 | 11 | 15 | 15 | 21 | 6 | 14.3 | 10 | 1 | | | | 1 |
| 15 | 11 | 15 | 15 | 21 | 9 | 11 | 10 | 1 | | | | 1 |
| 15 | 11 | 15 | 15 | 25 | 9 | 14 | 9 | 1 | | | | 1 |
| 15 | 11 | 15 | 15 | 26 | 10 | 13 | 11 | 1 | | | | 1 |
| 15 | 11 | 15 | 15 | 27 | 9 | 13 | 9 | 1 | | | | 1 |
| 15 | 11 | 15 | 15 | 28 | 11 | 15.3 | 9 | | | | 1 | 1 |
| 15 | 11 | 15 | 16 | 20 | 9 | 15.3 | 10 | | | 1 | | 1 |
| 15 | 11 | 16 | 11 | 19 | 6 | 15.3 | 10 | 1 | | | | 1 |
| 15 | 11 | 16 | 11 | 25 | 7 | 14.3 | 11 | 1 | | | | 1 |
| 15 | 11 | 16 | 13 | 26 | 8 | 11 | 9 | | | | 1 | 1 |
| 15 | 11 | 16 | 13 | 27 | 10 | 12 | 8 | | | | 1 | 1 |
| 15 | 11 | 16 | 14 | 20 | 9 | 11 | 11 | 1 | | | | 1 |
| 15 | 11 | 16 | 14 | 21 | 11 | 12 | 9 | 1 | | | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
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| 15 | 11 | 16 | 15 | 24 | 8 | 15.3 | 10 | | 1 | | | 1 |
| 15 | 11 | 16 | 15 | 27 | 6 | 14.3 | 12 | 1 | | | | 1 |
| 15 | 11 | 16 | 16 | 24 | 8 | 11 | 9 | 1 | | | | 1 |
| 15 | 11 | 16 | 16 | 24 | 8 | 16.3 | 12 | | 1 | | | 1 |
| 15 | 11 | 16 | 16 | 24 | 10 | 12 | 12 | | 1 | | | 1 |
| 15 | 11 | 16 | 16 | 24 | 12 | 15.3 | 10 | | 1 | | | 1 |
| 15 | 11 | 16 | 16 | 27 | 10 | 13 | 10 | | 1 | | | 1 |
| 15 | 11 | 16 | 17 | 28 | 10 | 11 | 9 | | 1 | | | 1 |
| 15 | 11 | 16 | 18 | 24 | 10 | 12 | 9 | | | | 1 | 1 |
| 15 | 11 | 18 | 17 | 25 | 11 | 13 | 11 | | | 1 | | 1 |
| 15 | 11 | 19 | 9 | 27 | 9 | 14 | 10 | 1 | | | | 1 |
| 15 | 11 | 19 | 13 | 21 | 8 | 9 | 10 | 1 | | | | 1 |
| 15 | 11 | 19 | 14 | 19 | 9 | 14.3 | 10 | 1 | | | | 1 |
| 15 | 11 | 19 | 15 | 21 | 10 | 13 | 11 | 1 | | | | 1 |
| 15 | 11 | 19 | 16 | 24 | 10 | 14.3 | 9 | | | | 1 | 1 |
| 15 | 11 | 19 | 18 | 25 | 10 | 15.3 | 10 | | | 1 | | 1 |
| 15 | 11 | 20 | 13 | 24 | 10 | 15.3 | 11 | | | | 1 | 1 |
| 15 | 11 | 20 | 13 | 25 | 11 | 14.3 | 11 | | | | 1 | 1 |
| 15 | 11 | 20 | 13 | 25 | 11 | 15.3 | 9 | | | 1 | | 1 |
| 15 | 11 | 20 | 13 | 29 | 9 | 13 | 11 | | | | 1 | 1 |
| 15 | 11 | 20 | 14 | 19 | 6 | 14.3 | 9 | 1 | | | | 1 |
| 15 | 11 | 20 | 14 | 27 | 10 | 15.3 | 11 | 1 | | | | 1 |
| 15 | 11 | 20 | 15 | 23 | 11 | 13.3 | 11 | | | 1 | | 1 |
| 15 | 11 | 20 | 15 | 26 | 8 | 16.3 | 11 | | | 1 | | 1 |
| 15 | 11 | 20 | 15 | 27 | 11 | 13.3 | 11 | | | 1 | | 1 |
| 15 | 11 | 20 | 16 | 19 | 8 | 13.3 | 11 | 1 | | | | 1 |
| 15 | 11 | 20 | 16 | 20 | 6 | 16.3 | 11 | 1 | | | | 1 |
| 15 | 11 | 20 | 16 | 24 | 11 | 11 | 10 | | | | 1 | 1 |
| 15 | 11 | 20 | 16 | 25 | 10 | 11 | 10 | | 1 | | | 1 |
| 15 | 11 | 20 | 16 | 25 | 10 | 15.3 | 11 | 1 | | | | 1 |
| 15 | 11 | 20 | 16 | 26 | 10 | 12 | 10 | | 1 | | | 1 |
| 15 | 11 | 20 | 16 | 26 | 11 | 15.3 | 10 | | 1 | | | 1 |
| 15 | 11 | 20 | 16 | 26 | 11 | 16.3 | 11 | 1 | | | | 1 |
| 15 | 11 | 20 | 17 | 18 | 11 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 11 | 20 | 17 | 18 | 12 | 11 | 11 | | | 1 | | 1 |
| 15 | 11 | 20 | 17 | 24 | 8 | 12 | 9 | | | | 1 | 1 |
| 15 | 11 | 20 | 17 | 24 | 11 | 13 | 9 | | 1 | | | 1 |
| 15 | 11 | 20 | 18 | 20 | 11 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 11 | 21 | 11 | 19 | 9 | 16.3 | 11 | 1 | | | | 1 |
| 15 | 11 | 21 | 11 | 21 | 7 | 12 | 10 | 1 | | | | 1 |
| 15 | 11 | 21 | 11 | 23 | 12 | 15.3 | 10 | | | 1 | | 1 |
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| 15 | 11 | 21 | 13 | 25 | 6 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 11 | 21 | 13 | 25 | 12 | 13.3 | 11 | | 1 | | | 1 |
| 15 | 11 | 21 | 13 | 26 | 12 | 12 | 9 | | | | 1 | 1 |
| 15 | 11 | 21 | 13 | 27 | 8 | 12 | 11 | 1 | | | | 1 |
| 15 | 11 | 21 | 13 | 27 | 12 | 15.3 | 10 | 1 | | | | 1 |
| 15 | 11 | 21 | 13 | 28 | 10 | 13 | 10 | 1 | | | | 1 |
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| 15 | 11 | 21 | 14 | 28 | 9 | 14 | 10 | 1 | | | | 1 |
| 15 | 11 | 21 | 15 | 21 | 12 | 13 | 10 | 1 | | | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 15 | 11 | 21 | 15 | 22 | 8 | 12 | 9 | | | 1 | | 1 |
| 15 | 11 | 21 | 15 | 23 | 8 | 16.3 | 11 | 1 | | | | 1 |
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| 15 | 11 | 21 | 15 | 27 | 9 | 12 | 11 | 1 | | | | 1 |
| 15 | 11 | 21 | 15 | 27 | 12 | 14.3 | 11 | | | 1 | | 1 |
| 15 | 11 | 21 | 15 | 28 | 10 | 12 | 10 | | 1 | | | 1 |
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| 15 | 11 | 21 | 18 | 23 | 9 | 15.3 | 10 | | | | 1 | 1 |
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| 15 | 11 | 22 | 13 | 22 | 9 | 12 | 12 | 1 | | | | 1 |
| 15 | 11 | 22 | 13 | 25 | 10 | 15.3 | 11 | | | | 1 | 1 |
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| 15 | 11 | 22 | 16 | 22 | 10 | 15.3 | 9 | | | 1 | | 1 |
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| 15 | 11 | 22 | 18 | 24 | 10 | 12 | 11 | | | | 1 | 1 |
| 15 | 11 | 23 | 14 | 18 | 12 | 16.3 | 10 | | | 1 | | 1 |
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| 15 | 11 | 23 | 16 | 15 | 10 | 16.3 | 11 | | 1 | | | 1 |
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| 15 | 11.3 | 21 | 15 | 24 | 9 | 11 | 11 | 1 | | | | 1 |
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| 15 | 12 | 15 | 12 | 25 | 12 | 14.3 | 11 | 1 | | | | 1 |
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| 15 | 12 | 15 | 13 | 22 | 11 | 14.3 | 9 | | | | 1 | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
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| 15 | 12 | 15 | 14 | 24 | 11 | 15.3 | 12 | | 1 | | | 1 |
| 15 | 12 | 15 | 14 | 27 | 9 | 16.3 | 11 | 1 | | | | 1 |
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| 15 | 12 | 15 | 16 | 23 | 9 | 12 | 10 | 1 | | | | 1 |
| 15 | 12 | 15 | 16 | 25 | 11 | 15.3 | 9 | | 1 | | | 1 |
| 15 | 12 | 15 | 16 | 26 | 8 | 14 | 10 | 1 | | | | 1 |
| 15 | 12 | 15 | 18 | 24 | 10 | 15.3 | 10 | | | | 1 | 1 |
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| 15 | 12 | 16 | 16 | 25 | 9 | 12 | 9 | | 1 | | | 1 |
| 15 | 12 | 16 | 16 | 25 | 11 | 11 | 9 | | | | 1 | 1 |
| 15 | 12 | 16 | 16 | 28 | 8 | 12 | 9 | | 1 | | | 1 |
| 15 | 12 | 18 | 14 | 21 | 11 | 13 | 10 | 1 | | | | 1 |
| 15 | 12 | 18 | 16 | 27 | 7 | 13 | 10 | 1 | | | | 1 |
| 15 | 12 | 19 | 13 | 20 | 9 | 15.3 | 10 | 1 | | | | 1 |
| 15 | 12 | 19 | 15 | 25 | 10 | 15.3 | 9 | | | | 1 | 1 |
| 15 | 12 | 19 | 16 | 25 | 10 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 12 | 20 | 13 | 18 | 8 | 15.3 | 10 | | | 1 | | 1 |
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| 15 | 12 | 20 | 13 | 25 | 10 | 15.3 | 9 | | | | 1 | 1 |
| 15 | 12 | 20 | 13 | 26 | 6 | 11 | 9 | | | | 1 | 1 |
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| 15 | 12 | 20 | 14 | 24 | 10 | 12 | 10 | | | | 1 | 1 |
| 15 | 12 | 20 | 14 | 24 | 10 | 15.3 | 10 | | | | 1 | 1 |
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| 15 | 12 | 20 | 14 | 27 | 11 | 14 | 8 | | | | 1 | 1 |
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| 15 | 12 | 20 | 15 | 22 | 10 | 14.3 | 10 | | | 1 | | 1 |
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| 15 | 12 | 20 | 15 | 23 | 10 | 15.3 | 9 | | | | 1 | 1 |
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| 15 | 12 | 20 | 15 | 24 | 11 | 15.3 | 10 | | 1 | | | 1 |
| 15 | 12 | 20 | 15 | 26 | 8 | 12 | 9 | | | | 1 | 1 |
| 15 | 12 | 20 | 15 | 26 | 10 | 14.3 | 9 | | | | 1 | 1 |
| 15 | 12 | 20 | 15 | 27 | 6 | 12 | 11 | | | 1 | | 1 |
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| 15 | 12 | 20 | 16 | 25 | 6 | 11 | 10 | 1 | | | | 1 |
| 15 | 12 | 20 | 16 | 25 | 10 | 15.3 | 10 | | | 1 | | 1 |
| 15 | 12 | 20 | 16 | 25 | 11 | 14.3 | 10 | | | | 1 | 1 |
| 15 | 12 | 20 | 16 | 25 | 12 | 15.3 | 9 | | | 1 | | 1 |
| 15 | 12 | 20 | 17 | 25 | 10 | 14 | 9 | | | | 1 | 1 |
| 15 | 12 | 20 | 17 | 26 | 8 | 15.3 | 13 | 1 | | | | 1 |
| 15 | 12 | 20 | 18 | 24 | 10 | 15.3 | 9 | | | 1 | | 1 |
| 15 | 12 | 21 | 12 | 26 | 6 | 12 | 10 | 1 | | | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
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| 15 | 12 | 21 | 13 | 22 | 10 | 14 | 10 | 1 | | | | 1 |
| 15 | 12 | 21 | 13 | 26 | 10 | 14.3 | 10 | | | | 1 | 1 |
| 15 | 12 | 21 | 13 | 26 | 11 | 16.3 | 10 | 1 | | | | 1 |
| 15 | 12 | 21 | 14 | 21 | 6 | 12 | 10 | 1 | | | | 1 |
| 15 | 12 | 21 | 14 | 24 | 8 | 14.3 | 12 | | | 1 | | 1 |
| 15 | 12 | 21 | 14 | 26 | 10 | 15.3 | 10 | | | | 1 | 1 |
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| 15 | 12 | 21 | 15 | 21 | 9 | 17 | 10 | 1 | | | | 1 |
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| 15 | 12 | 21 | 15 | 26 | 10 | 15.3 | 10 | | | 1 | | 1 |
| 15 | 12 | 21 | 15 | 26 | 11 | 15.3 | 11 | | | | 1 | 1 |
| 15 | 12 | 21 | 15 | 26 | 11 | 15.3 | 12 | 1 | | | | 1 |
| 15 | 12 | 21 | 16 | 19 | 12 | 12 | 11 | 1 | | | | 1 |
| 15 | 12 | 21 | 16 | 22 | 9 | 17.3 | 11 | 1 | | | | 1 |
| 15 | 12 | 21 | 16 | 23 | 9 | 12 | 9 | 1 | | | | 1 |
| 15 | 12 | 21 | 16 | 24 | 12 | 16.3 | 9 | | | 1 | | 1 |
| 15 | 12 | 21 | 16 | 25 | 9 | 15.3 | 10 | 1 | | | | 1 |
| 15 | 12 | 21 | 16 | 25 | 10 | 11 | 11 | | 1 | | | 1 |
| 15 | 12 | 21 | 16 | 25 | 11 | 14.3 | 10 | | 1 | | | 1 |
| 15 | 12 | 21 | 16 | 26 | 8 | 15.3 | 10 | | 1 | | | 1 |
| 15 | 12 | 21 | 16 | 26 | 12 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 12 | 21 | 16 | 27 | 10 | 15.3 | 11 | | | 1 | | 1 |
| 15 | 12 | 21 | 17 | 19 | 10 | 14.3 | 11 | 1 | | | | 1 |
| 15 | 12 | 22 | 12 | 24 | 9 | 13 | 11 | 1 | | | | 1 |
| 15 | 12 | 22 | 12 | 26 | 8 | 12 | 11 | 1 | | | | 1 |
| 15 | 12 | 22 | 12 | 28 | 11 | 15.3 | 10 | | | 1 | | 1 |
| 15 | 12 | 22 | 13 | 24 | 10 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 12 | 22 | 14 | 27 | 10 | 15.3 | 11 | | | | 1 | 1 |
| 15 | 12 | 22 | 15 | 23 | 10 | 12 | 10 | | | 1 | | 1 |
| 15 | 12 | 22 | 15 | 25 | 6 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 12 | 22 | 15 | 26 | 11 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 12 | 22 | 16 | 19 | 6 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 12 | 22 | 16 | 24 | 10 | 15.3 | 10 | | 1 | | | 1 |
| 15 | 12 | 22 | 16 | 27 | 10 | 15.3 | 10 | | | 1 | | 1 |
| 15 | 12 | 23 | 14 | 18 | 9 | 15.3 | 11 | 1 | | | | 1 |
| 15 | 12 | 23 | 14 | 24 | 6 | 14.3 | 11 | | 1 | | | 1 |
| 15 | 12 | 23 | 14 | 26 | 11 | 15.3 | 9 | | | 1 | | 1 |
| 15 | 12 | 23 | 15 | 18 | 10 | 10 | 12 | | | 1 | | 1 |
| 15 | 12 | 23 | 15 | 24 | 6 | 14.3 | 11 | 1 | | | | 1 |
| 15 | 12 | 23 | 15 | 24 | 6 | 15.3 | 11 | | | 1 | | 1 |
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| 15 | 12 | 23 | 17 | 25 | 10 | 16.3 | 11 | | | | 1 | 1 |
| 15 | 12 | 24 | 12 | 23 | 6 | 17.3 | 12 | | | 1 | | 1 |
| 15 | 12 | 24 | 17 | 25 | 8 | 15.3 | 10 | | | 1 | | 1 |
| 15 | 12.3 | 15 | 13 | 27 | 9 | 14.3 | 11 | 1 | | | | 1 |
| 15 | 12.3 | 15 | 15 | 23 | 10 | 15.3 | 9 | | 1 | | | 1 |
| 15 | 12.3 | 15 | 15 | 24 | 6 | 15.3 | 11 | | 1 | | | 1 |
| 15 | 12.3 | 15 | 15 | 26 | 8 | 15.3 | 10 | | 1 | | | 1 |
| 15 | 12.3 | 15 | 16 | 23 | 9 | 14.3 | 10 | | 1 | | | 1 |
| 15 | 12.3 | 15 | 16 | 25 | 11 | 15.3 | 9 | | 1 | | | 1 |
| 15 | 12.3 | 16 | 11 | 25 | 10 | 11 | 10 | | 1 | | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 15 | 12.3 | 16 | 12 | 25 | 10 | 15.3 | 9 | | 1 | | | 1 |
| 15 | 12.3 | 16 | 13 | 25 | 11 | 12 | 10 | | | | 1 | 1 |
| 15 | 12.3 | 16 | 14 | 23 | 8 | 15.3 | 9 | | | | 1 | 1 |
| 15 | 12.3 | 16 | 14 | 24 | 8 | 12 | 10 | | 1 | | | 1 |
| 15 | 12.3 | 16 | 15 | 23 | 8 | 10 | 10 | | 1 | | | 1 |
| 15 | 12.3 | 16 | 15 | 24 | 6 | 15.3 | 11 | | | | 1 | 1 |
| 15 | 12.3 | 16 | 15 | 24 | 6 | 16.3 | 10 | | 1 | | | 1 |
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| 15 | 12.3 | 16 | 16 | 22 | 8 | 16.3 | 10 | | 1 | | | 1 |
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| 15 | 12.3 | 16 | 16 | 25 | 10 | 12 | 10 | | 1 | | | 1 |
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| 15 | 12.3 | 19 | 14 | 24 | 11 | 16.3 | 10 | | | 1 | | 1 |
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| 15 | 12.3 | 20 | 10 | 26 | 12 | 15.3 | 11 | | | | 1 | 1 |
| 15 | 12.3 | 20 | 13 | 25 | 8 | 15.3 | 10 | | | 1 | | 1 |
| 15 | 12.3 | 20 | 14 | 24 | 6 | 15.3 | 10 | 1 | | | | 1 |
| 15 | 12.3 | 20 | 14 | 24 | 11 | 14.3 | 10 | 1 | | | | 1 |
| 15 | 12.3 | 20 | 14 | 24 | 11 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 12.3 | 20 | 14 | 26 | 12 | 14.3 | 10 | | | | 1 | 1 |
| 15 | 12.3 | 20 | 15 | 18 | 9 | 15.3 | 10 | | | 1 | | 1 |
| 15 | 12.3 | 20 | 15 | 18 | 11 | 15.3 | 9 | | | 1 | | 1 |
| 15 | 12.3 | 20 | 15 | 23 | 12 | 14.3 | 9 | | 1 | | | 1 |
| 15 | 12.3 | 20 | 15 | 26 | 10 | 14.3 | 12 | | | 1 | | 1 |
| 15 | 12.3 | 20 | 16 | 20 | 8 | 10 | 11 | 1 | | | | 1 |
| 15 | 12.3 | 20 | 16 | 22 | 10 | 14.3 | 11 | | | 1 | | 1 |
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| 15 | 12.3 | 20 | 16 | 25 | 8 | 11 | 10 | | | | 1 | 1 |
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| 15 | 12.3 | 20 | 17 | 24 | 8 | 14.3 | 9 | | | | 1 | 1 |
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| 15 | 12.3 | 21 | 16 | 24 | 12 | 15.3 | 11 | | | | 1 | 1 |
| 15 | 12.3 | 21 | 16 | 25 | 8 | 11 | 9 | | 1 | | | 1 |
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| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
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| 15 | 12.3 | 22 | 15 | 26 | 10 | 14.3 | 10 | | | | 1 | 1 |
| 15 | 12.3 | 22 | 15 | 27 | 6 | 15.3 | 11 | | 1 | | | 1 |
| 15 | 12.3 | 22 | 16 | 22 | 8 | 13.3 | 11 | | 1 | | | 1 |
| 15 | 12.3 | 22 | 16 | 25 | 10 | 15.3 | 10 | | 1 | | | 1 |
| 15 | 12.3 | 22 | 16 | 26 | 8 | 11 | 9 | | | 1 | | 1 |
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| 15 | 13 | 20 | 16 | 24 | 8 | 14.3 | 11 | | | | 2 | 2 |
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| 15 | 13 | 21 | 15 | 26 | 12 | 15.3 | 11 | | | 1 | | 1 |
| 15 | 13 | 21 | 16 | 23 | 12 | 15.3 | 11 | | | 1 | | 1 |
| 15 | 13 | 21 | 16 | 24 | 11 | 14.3 | 10 | | | | 1 | 1 |
| 15 | 13 | 21 | 16 | 25 | 12 | 15.3 | 10 | | | 1 | | 1 |
| 15 | 13 | 21 | 17 | 18 | 6 | 14.3 | 9 | | | | 1 | 1 |
| 15 | 13 | 22 | 13 | 29 | 6 | 15.3 | 9 | | | 1 | | 1 |
| 15 | 13 | 22 | 14 | 19 | 9 | 11 | 10 | 1 | | | | 1 |
| 15 | 13 | 22 | 14 | 22 | 11 | 13.3 | 11 | | | 1 | | 1 |
| 15 | 13 | 22 | 15 | 26 | 6 | 15.3 | 9 | | | | 1 | 1 |
| 15 | 13 | 22 | 16 | 19 | 6 | 15.3 | 11 | | 1 | | | 1 |
| 15 | 13 | 22 | 16 | 24 | 8 | 12 | 9 | | | | 1 | 1 |
| 15 | 13 | 22 | 16 | 24 | 11 | 15.3 | 11 | | | 1 | | 1 |
| 15 | 13 | 22 | 16 | 24 | 12 | 12 | 10 | 1 | | | | 1 |
| 15 | 13 | 22 | 17 | 18 | 12 | 16.3 | 10 | | | 1 | | 1 |
| 15 | 13 | 23 | 12 | 23 | 10 | 14.3 | 9 | | | 1 | | 1 |
| 15 | 13 | 24 | 17 | 18 | 10 | 14.3 | 9 | | | | 1 | 1 |
| 15 | 13.3 | 15 | 16 | 19 | 8 | 12 | 10 | | | | 1 | 1 |
| 15 | 13.3 | 16 | 14 | 25 | 8 | 15.3 | 12 | 1 | | | | 1 |
| 15 | 13.3 | 16 | 17 | 23 | 6 | 15.3 | 11 | | 1 | | | 1 |
| 15 | 13.3 | 16 | 17 | 24 | 8 | 13 | 9 | | 1 | | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 15 | 13.3 | 17 | 14 | 28 | 11 | 14.3 | 10 | | | | 1 | 1 |
| 15 | 13.3 | 19 | 13 | 29 | 10 | 14.3 | 11 | 1 | | | | 1 |
| 15 | 13.3 | 19 | 15 | 29 | 8 | 16.3 | 11 | | | 1 | | 1 |
| 15 | 13.3 | 20 | 10 | 26 | 8 | 14 | 11 | | | | 1 | 1 |
| 15 | 13.3 | 20 | 13 | 23 | 11 | 12 | 9 | | | | 1 | 1 |
| 15 | 13.3 | 20 | 15 | 18 | 10 | 12 | 9 | | | | 1 | 1 |
| 15 | 13.3 | 20 | 15 | 23 | 10 | 15.3 | 9 | | 1 | | | 1 |
| 15 | 13.3 | 20 | 15 | 25 | 10 | 15.3 | 10 | | 1 | | | 1 |
| 15 | 13.3 | 20 | 16 | 18 | 6 | 12 | 9 | | | | 1 | 1 |
| 15 | 13.3 | 20 | 16 | 27 | 12 | 15.3 | 10 | | | 1 | | 1 |
| 15 | 13.3 | 20 | 17 | 18 | 10 | 15.3 | 12 | | | | 1 | 1 |
| 15 | 13.3 | 21 | 14 | 25 | 10 | 16.3 | 9 | 1 | | | | 1 |
| 15 | 13.3 | 21 | 14 | 27 | 10 | 13.3 | 9 | | | | 1 | 1 |
| 15 | 13.3 | 21 | 14 | 28 | 8 | 14.3 | 10 | | | 1 | | 1 |
| 15 | 13.3 | 21 | 15 | 27 | 10 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 13.3 | 21 | 16 | 24 | 6 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 13.3 | 21 | 16 | 24 | 11 | 13.3 | 11 | | | | 1 | 1 |
| 15 | 13.3 | 22 | 12 | 26 | 10 | 17.3 | 9 | | | 1 | | 1 |
| 15 | 13.3 | 22 | 14 | 28 | 8 | 15.3 | 11 | | 1 | | | 1 |
| 15 | 13.3 | 22 | 16 | 24 | 11 | 16.3 | 9 | | 1 | | | 1 |
| 15 | 13.3 | 22 | 16 | 25 | 10 | 14.3 | 9 | | | 1 | | 1 |
| 15 | 13.3 | 22 | 18 | 27 | 8 | 14.3 | 11 | | | 1 | | 1 |
| 15 | 13.3 | 23 | 15 | 23 | 6 | 15.3 | 10 | | | 1 | | 1 |
| 15 | 13.3 | 23 | 16 | 24 | 10 | 13.3 | 9 | | | | 1 | 1 |
| 15 | 14 | 20 | 11 | 21 | 6 | 12 | 9 | 1 | | | | 1 |
| 15 | 14 | 20 | 17 | 24 | 11 | 15.3 | 10 | | | | 1 | 1 |
| 15 | 14 | 23 | 16 | 22 | 9 | 12 | 11 | 1 | | | | 1 |
| 15 | 14.3 | 15 | 15 | 25 | 12 | 12 | 11 | | 1 | | | 1 |
| 15.3 | 12 | 16 | 16 | 25 | 8 | 11 | 10 | | | | 1 | 1 |
| 16 | 7 | 21 | 15 | 27 | 10 | 15.3 | 11 | 1 | | | | 1 |
| 16 | 9 | 15 | 15 | 24 | 9 | 14 | 11 | 1 | | | | 1 |
| 16 | 10 | 21 | 13 | 26 | 8 | 12 | 9 | 1 | | | | 1 |
| 16 | 10 | 21 | 14 | 22 | 10 | 15.3 | 9 | 1 | | | | 1 |
| 16 | 10 | 22 | 16 | 27 | 11 | 17.3 | 9 | | | 1 | | 1 |
| 16 | 11 | 15 | 16 | 21 | 8 | 13 | 11 | 1 | | | | 1 |
| 16 | 11 | 15 | 16 | 29 | 8 | 15.3 | 11 | 1 | | | | 1 |
| 16 | 11 | 16 | 16 | 22 | 6 | 15.3 | 10 | | 1 | | | 1 |
| 16 | 11 | 16 | 16 | 26 | 9 | 11 | 11 | 1 | | | | 1 |
| 16 | 11 | 20 | 14 | 24 | 10 | 15.3 | 10 | | | 1 | | 1 |
| 16 | 11 | 20 | 14 | 26 | 12 | 14.3 | 10 | | 1 | | | 1 |
| 16 | 11 | 20 | 15 | 22 | 10 | 14 | 10 | | | | 1 | 1 |
| 16 | 11 | 20 | 15 | 25 | 12 | 14.3 | 10 | | | | 1 | 1 |
| 16 | 11 | 20 | 15 | 26 | 6 | 15.3 | 12 | | | | 1 | 1 |
| 16 | 11 | 20 | 16 | 28 | 6 | 12 | 10 | | | | 1 | 1 |
| 16 | 11 | 21 | 14 | 23 | 12 | 13 | 10 | | 1 | | | 1 |
| 16 | 11 | 21 | 14 | 28 | 9 | 15.3 | 11 | | | 1 | | 1 |
| 16 | 11 | 21 | 15 | 21 | 8 | 13 | 10 | 1 | | | | 1 |
| 16 | 11 | 21 | 16 | 25 | 10 | 13 | 10 | | | | 1 | 1 |
| 16 | 11 | 22 | 14 | 21 | 9 | 13 | 11 | 1 | | | | 1 |
| 16 | 11 | 23 | 11 | 21 | 9 | 14 | 11 | 1 | | | | 1 |
| 16 | 11 | 23 | 14 | 27 | 10 | 14.3 | 11 | | | 1 | | 1 |
| 16 | 11 | 23 | 15 | 19 | 11 | 14 | 11 | | 1 | | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 16 | 11 | 23 | 16 | 19 | 6 | 12 | 10 | 1 | | | | 1 |
| 16 | 11.3 | 21 | 17 | 23 | 8 | 14.3 | 11 | | | | 1 | 1 |
| 16 | 12 | 15 | 14 | 24 | 11 | 15.3 | 10 | | 1 | | | 1 |
| 16 | 12 | 15 | 14 | 29 | 10 | 14.3 | 10 | | | 1 | | 1 |
| 16 | 12 | 15 | 17 | 18 | 12 | 13 | 10 | | 1 | | | 1 |
| 16 | 12 | 16 | 13 | 23 | 8 | 16.3 | 8 | 1 | | | | 1 |
| 16 | 12 | 16 | 16 | 19 | 6 | 15.3 | 9 | | 1 | | | 1 |
| 16 | 12 | 16 | 16 | 23 | 10 | 14.3 | 10 | | 1 | | | 1 |
| 16 | 12 | 16 | 16 | 26 | 8 | 15.3 | 10 | | 1 | | | 1 |
| 16 | 12 | 16 | 16 | 27 | 9 | 15.3 | 12 | 1 | | | | 1 |
| 16 | 12 | 16 | 17 | 25 | 10 | 15.3 | 10 | | 1 | | | 1 |
| 16 | 12 | 16 | 17 | 28 | 8 | 14.3 | 11 | | | | 1 | 1 |
| 16 | 12 | 18 | 15 | 22 | 8 | 12 | 8 | 1 | | | | 1 |
| 16 | 12 | 20 | 12 | 18 | 8 | 14.3 | 11 | 1 | | | | 1 |
| 16 | 12 | 20 | 13 | 29 | 6 | 14.3 | 11 | | | 1 | | 1 |
| 16 | 12 | 20 | 14 | 21 | 11 | 14.3 | 9 | | | | 1 | 1 |
| 16 | 12 | 20 | 14 | 25 | 12 | 15.3 | 10 | | | 1 | | 1 |
| 16 | 12 | 20 | 14 | 26 | 6 | 15.3 | 10 | | 1 | | | 1 |
| 16 | 12 | 20 | 14 | 26 | 11 | 12 | 11 | 1 | | | | 1 |
| 16 | 12 | 20 | 16 | 22 | 8 | 14.3 | 10 | 1 | | | | 1 |
| 16 | 12 | 20 | 16 | 24 | 6 | 10 | 9 | | | | 1 | 1 |
| 16 | 12 | 20 | 16 | 24 | 12 | 15.3 | 10 | 1 | | | | 1 |
| 16 | 12 | 20 | 16 | 25 | 10 | 14.3 | 10 | | | | 1 | 1 |
| 16 | 12 | 21 | 16 | 24 | 9 | 15.3 | 11 | | 1 | | | 1 |
| 16 | 12 | 21 | 16 | 28 | 12 | 16.3 | 11 | | 1 | | | 1 |
| 16 | 12 | 21 | 17 | 24 | 8 | 12 | 10 | | 1 | | | 1 |
| 16 | 12 | 22 | 15 | 21 | 9 | 13 | 12 | 1 | | | | 1 |
| 16 | 12 | 24 | 16 | 25 | 8 | 14.3 | 9 | | 1 | | | 1 |
| 16 | 12 | 24 | 17 | 18 | 9 | 12 | 10 | | | 1 | | 1 |
| 16 | 12.3 | 15 | 12 | 22 | 9 | 12 | 11 | 1 | | | | 1 |
| 16 | 12.3 | 15 | 15 | 23 | 10 | 15.3 | 10 | | 1 | | | 1 |
| 16 | 12.3 | 16 | 14 | 24 | 8 | 14.3 | 11 | 1 | | | | 1 |
| 16 | 12.3 | 16 | 15 | 24 | 10 | 11 | 11 | | 1 | | | 1 |
| 16 | 12.3 | 16 | 15 | 26 | 8 | 14.3 | 11 | | 1 | | | 1 |
| 16 | 12.3 | 16 | 16 | 25 | 10 | 12 | 9 | | 1 | | | 1 |
| 16 | 12.3 | 19 | 14 | 25 | 11 | 14.3 | 10 | | 1 | | | 1 |
| 16 | 12.3 | 20 | 14 | 25 | 8 | 12 | 10 | | 1 | | | 1 |
| 16 | 12.3 | 20 | 17 | 16 | 10 | 16.3 | 9 | | | 1 | | 1 |
| 16 | 12.3 | 20 | 17 | 25 | 10 | 15.3 | 11 | | | 1 | | 1 |
| 16 | 12.3 | 20 | 17 | 26 | 10 | 12 | 9 | | | | 1 | 1 |
| 16 | 12.3 | 21 | 13 | 24 | 10 | 16.3 | 12 | | | 1 | | 1 |
| 16 | 12.3 | 21 | 15 | 24 | 9 | 13 | 9 | 1 | | | | 1 |
| 16 | 12.3 | 21 | 15 | 24 | 10 | 15.3 | 12 | | 1 | | | 1 |
| 16 | 12.3 | 22 | 14 | 25 | 10 | 12 | 10 | 1 | | | | 1 |
| 16 | 12.3 | 22 | 17 | 25 | 11 | 11 | 11 | | 1 | | | 1 |
| 16 | 13 | 14 | 15 | 26 | 9 | 11 | 11 | | | | 1 | 1 |
| 16 | 13 | 15 | 15 | 21 | 10 | 15.3 | 10 | 1 | | | | 1 |
| 16 | 13 | 15 | 15 | 28 | 9 | 15.3 | 10 | | | 1 | | 1 |
| 16 | 13 | 20 | 14 | 23 | 6 | 15.3 | 9 | | | | 1 | 1 |
| 16 | 13 | 20 | 14 | 29 | 11 | 15.3 | 11 | | | 1 | | 1 |
| 16 | 13 | 20 | 17 | 18 | 8 | 14.3 | 10 | | | | 1 | 1 |
| 16 | 13 | 21 | 15 | 27 | 6 | 16.3 | 10 | | | 1 | | 1 |

| DXS7132 | DXS6803 | DXS6789 | DXS7424 | DXS101 | GATA172D05 | DXS7130 | GATA165B12 | AA N=540 | AS N=326 | CN N=424 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 16 | 13 | 22 | 13 | 21 | 9 | 10 | 12 | 1 | | | | 1 |
| 16 | 13 | 22 | 15 | 24 | 8 | 15.3 | 11 | | | 1 | | 1 |
| 16 | 13 | 23 | 16 | 24 | 9 | 14.3 | 11 | | | 1 | | 1 |
| 16 | 13.3 | 20 | 15 | 24 | 8 | 14.3 | 11 | | | 1 | | 1 |
| 16 | 13.3 | 20 | 17 | 25 | 10 | 15.3 | 11 | | | | 1 | 1 |
| 16 | 13.3 | 21 | 14 | 27 | 11 | 15.3 | 10 | | | 1 | | 1 |
| 16 | 13.3 | 21 | 15 | 22 | 10 | 15.3 | 10 | | 1 | | | 1 |
| 16 | 13.3 | 21 | 15 | 24 | 12 | 16.3 | 11 | 1 | | | | 1 |
| 16 | 13.3 | 21 | 15 | 28 | 11 | 12 | 11 | | 1 | | | 1 |
| 16 | 14 | 23 | 15 | 23 | 10 | 15.3 | 13 | | | | 1 | 1 |
| 16.3 | 13.3 | 22 | 15 | 26 | 11 | 14.3 | 10 | | | | 1 | 1 |
| 17 | 9 | 22 | 13 | 19 | 6 | 12 | 13 | 1 | | | | 1 |
| 17 | 10 | 21 | 16 | 25 | 10 | 15.3 | 10 | 1 | | | | 1 |
| 17 | 11 | 15 | 12 | 18 | 9 | 13 | 11 | 1 | | | | 1 |
| 17 | 11 | 20 | 14 | 28 | 6 | 13 | 11 | 1 | | | | 1 |
| 17 | 11 | 20 | 15 | 20 | 10 | 16.3 | 11 | | | | 1 | 1 |
| 17 | 11 | 23 | 12 | 25 | 10 | 12 | 11 | | | 1 | | 1 |
| 17 | 12 | 16 | 16 | 25 | 10 | 15.3 | 10 | | 1 | | | 1 |
| 17 | 12 | 19 | 17 | 27 | 6 | 12 | 10 | | 1 | | | 1 |
| 17 | 12 | 20 | 14 | 25 | 10 | 15.3 | 10 | | | | 1 | 1 |
| 17 | 12 | 20 | 14 | 25 | 11 | 16.3 | 9 | | | | 1 | 1 |
| 17 | 12 | 21 | 16 | 26 | 11 | 15.3 | 11 | | | | 1 | 1 |
| 17 | 12.3 | 19 | 14 | 24 | 9 | 12 | 10 | | 1 | | | 1 |
| 17 | 12.3 | 20 | 13 | 25 | 10 | 15.3 | 10 | | | | 1 | 1 |
| 17 | 13 | 21 | 11 | 23 | 8 | 12 | 10 | 1 | | | | 1 |
| 17 | 13.3 | 21 | 15 | 19 | 8 | 16.3 | 11 | | | 1 | | 1 |
| 18 | 12 | 15 | 14 | 24 | 10 | 16.3 | 10 | | | 1 | | 1 |
| 18 | 12 | 15 | 16 | 18 | 8 | 9 | 11 | 1 | | | | 1 |
| 18 | 12 | 21 | 12 | 24 | 11 | 14.3 | 9 | | 1 | | | 1 |
| | | | | | | Total number of haplotypes: | | 540 | 326 | 423 | 410 | 1698 |
| | | | | | | Total number of unique haplotypes: | | 540 | 326 | 422 | 409 | 1695 |
| | | | | | | Coun of most common haplotype: | | 1 | 1 | 2 | 2 | 2 |

**Table B12. Haplotypes observed within the combined U.S. dataset for proposed linkage group 4.** AA: African American, AS: U.S. Asian, CN: U.S. Caucasian, Hisp: U.S. Hispanic, N: number of samples.

| GATA31E08 | DXS10147 | DXS7423 | AA N=539 | AS N=326 | CN N=425 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|
| 7 | 7 | 16 | 1 | | | | 1 |
| 7 | 8 | 14 | 2 | | | | 2 |
| 7 | 8 | 15 | 1 | | | | 1 |
| 7 | 9 | 14 | 2 | | | | 2 |
| 7 | 9 | 15 | 1 | | | | 1 |
| 7 | 9 | 16 | 1 | | | | 1 |
| 8 | 6 | 13 | 1 | | | | 1 |
| 8 | 6 | 14 | 1 | | | | 1 |
| 8 | 6 | 17 | | | | 1 | 1 |
| 8 | 7 | 14 | 2 | | | | 2 |

| GATA31E08 | DXS10147 | DXS7423 | AA N=539 | AS N=326 | CN N=425 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|
| 8 | 7 | 15 | 4 | | | | 4 |
| 8 | 8 | 13 | 1 | | | 1 | 2 |
| 8 | 8 | 14 | 3 | | | | 3 |
| 8 | 8 | 15 | 1 | | | | 1 |
| 8 | 8 | 17 | | | | 1 | 1 |
| 8 | 9 | 13 | 1 | | | | 1 |
| 8 | 9 | 14 | 1 | | | 1 | 2 |
| 9 | 6 | 13 | 1 | 1 | 3 | | 5 |
| 9 | 6 | 14 | 4 | 4 | 7 | 4 | 19 |
| 9 | 6 | 15 | 4 | 3 | 15 | 18 | 40 |
| 9 | 6 | 16 | 1 | 2 | 3 | 2 | 8 |
| 9 | 6 | 17 | 2 | | 2 | 1 | 5 |
| 9 | 7 | 12 | 1 | | | | 1 |
| 9 | 7 | 14 | 16 | 2 | 2 | 1 | 21 |
| 9 | 7 | 15 | 8 | 5 | | 2 | 15 |
| 9 | 7 | 16 | 3 | 1 | | | 4 |
| 9 | 7 | 17 | | | 1 | | 1 |
| 9 | 8 | 13 | | | 2 | 1 | 3 |
| 9 | 8 | 14 | 20 | 5 | 7 | 11 | 43 |
| 9 | 8 | 15 | 10 | 10 | 8 | 8 | 36 |
| 9 | 8 | 16 | 2 | 1 | 2 | 1 | 6 |
| 9 | 8 | 17 | 1 | | | 5 | 6 |
| 9 | 9 | 13 | 1 | 1 | 8 | 1 | 11 |
| 9 | 9 | 14 | 6 | 3 | 8 | 2 | 19 |
| 9 | 9 | 15 | 4 | 3 | 14 | 13 | 34 |
| 9 | 9 | 16 | 1 | 1 | 7 | 3 | 12 |
| 9 | 9 | 17 | | 1 | 1 | 1 | 3 |
| 9 | 10 | 15 | 3 | | 1 | 1 | 5 |
| 9 | 11 | 13 | | 1 | | | 1 |
| 10 | 6 | 14 | 2 | | | | 2 |
| 10 | 6 | 15 | 1 | 2 | | 5 | 8 |
| 10 | 6 | 16 | 1 | 1 | | 1 | 3 |
| 10 | 7 | 14 | 8 | 1 | | 1 | 10 |
| 10 | 7 | 15 | 14 | 1 | | | 15 |
| 10 | 7 | 16 | 2 | 1 | 1 | 1 | 5 |
| 10 | 8 | 12 | 1 | | | | 1 |
| 10 | 8 | 13 | 2 | | | | 2 |
| 10 | 8 | 14 | 18 | | 2 | 3 | 23 |
| 10 | 8 | 15 | 12 | 2 | 1 | 1 | 16 |
| 10 | 8 | 16 | 1 | | | | 1 |
| 10 | 8 | 17 | | | | 3 | 3 |
| 10 | 9 | 13 | 2 | | 1 | 2 | 5 |
| 10 | 9 | 14 | 6 | | | | 6 |
| 10 | 9 | 15 | 2 | 1 | 2 | | 5 |
| 10 | 9 | 16 | | | | 2 | 2 |
| 10 | 10 | 15 | 2 | | | | 2 |
| 10 | 10 | 16 | 1 | | | | 1 |
| 11 | 6 | 14 | 3 | 3 | 7 | | 13 |
| 11 | 6 | 15 | 2 | 11 | 12 | 18 | 43 |
| 11 | 6 | 16 | 1 | 2 | 3 | 1 | 7 |
| 11 | 6 | 17 | | 3 | | | 3 |
| 11 | 7 | 12 | 2 | | | | 2 |
| 11 | 7 | 14 | 5 | 7 | | 1 | 13 |
| 11 | 7 | 15 | 3 | 2 | | 2 | 7 |
| 11 | 7 | 16 | 2 | | 2 | 2 | 6 |

| GATA31E08 | DXS10147 | DXS7423 | AA N=539 | AS N=326 | CN N=425 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|
| 11 | 7 | 17 | | | 4 | 1 | 5 |
| 11 | 8 | 13 | | 1 | 2 | | 3 |
| 11 | 8 | 14 | 12 | 10 | 14 | 14 | 50 |
| 11 | 8 | 15 | 5 | 12 | 11 | 11 | 39 |
| 11 | 8 | 16 | | 1 | 4 | 2 | 7 |
| 11 | 8 | 17 | | | | 3 | 3 |
| 11 | 9 | 13 | 2 | 2 | 7 | 4 | 15 |
| 11 | 9 | 14 | 3 | 5 | 11 | 1 | 20 |
| 11 | 9 | 15 | | 6 | 9 | 7 | 22 |
| 11 | 9 | 16 | 2 | 1 | 2 | 3 | 8 |
| 11 | 9 | 17 | | | 1 | | 1 |
| 11 | 10 | 14 | | | 1 | | 1 |
| 11 | 10 | 15 | | | | 2 | 2 |
| 12 | 6 | 13 | 2 | 1 | | 1 | 4 |
| 12 | 6 | 14 | 8 | 6 | 3 | 4 | 21 |
| 12 | 6 | 15 | 5 | 13 | 11 | 31 | 60 |
| 12 | 6 | 16 | 1 | 1 | 2 | 5 | 9 |
| 12 | 6 | 17 | | | 1 | 2 | 3 |
| 12 | 7 | 8 | 2 | | | | 2 |
| 12 | 7 | 12 | 2 | | | | 2 |
| 12 | 7 | 13 | 1 | | 1 | 1 | 3 |
| 12 | 7 | 14 | 21 | 8 | | 4 | 33 |
| 12 | 7 | 15 | 19 | 2 | | 5 | 26 |
| 12 | 7 | 16 | 2 | | | 3 | 5 |
| 12 | 7 | 17 | | | 2 | | 2 |
| 12 | 8 | 13 | 2 | 1 | 3 | 1 | 7 |
| 12 | 8 | 14 | 28 | 10 | 10 | 24 | 72 |
| 12 | 8 | 15 | 10 | 20 | 10 | 24 | 64 |
| 12 | 8 | 16 | 3 | 1 | 4 | 3 | 11 |
| 12 | 8 | 17 | | 1 | | 7 | 8 |
| 12 | 9 | 13 | 6 | 1 | 8 | 3 | 18 |
| 12 | 9 | 14 | 11 | 4 | 7 | 6 | 28 |
| 12 | 9 | 15 | 6 | 7 | 11 | 5 | 29 |
| 12 | 9 | 16 | 10 | 3 | 11 | 3 | 27 |
| 12 | 9 | 17 | | | 1 | 1 | 2 |
| 12 | 10 | 13 | 1 | | | | 1 |
| 12 | 10 | 14 | 2 | | | | 2 |
| 12 | 10 | 15 | 2 | | 2 | | 4 |
| 12 | 10 | 16 | 1 | | | 2 | 3 |
| 12 | 11 | 14 | 1 | | | | 1 |
| 13 | 6 | 13 | | | | 2 | 2 |
| 13 | 6 | 14 | 9 | 10 | 7 | 3 | 29 |
| 13 | 6 | 15 | 4 | 15 | 6 | 14 | 39 |
| 13 | 6 | 16 | 2 | 1 | 4 | 4 | 11 |
| 13 | 6 | 17 | | 1 | 1 | 1 | 3 |
| 13 | 7 | 13 | 2 | | | | 2 |
| 13 | 7 | 14 | 24 | 4 | | | 28 |
| 13 | 7 | 15 | 6 | 5 | | 1 | 12 |
| 13 | 7 | 16 | 1 | 1 | 1 | 1 | 4 |
| 13 | 7 | 17 | | 1 | 2 | 2 | 5 |
| 13 | 8 | 13 | 3 | | | 3 | 6 |
| 13 | 8 | 14 | 28 | 10 | 12 | 8 | 58 |
| 13 | 8 | 15 | 15 | 24 | 9 | 21 | 69 |
| 13 | 8 | 16 | 5 | 4 | 5 | 2 | 16 |
| 13 | 8 | 17 | | | | 3 | 3 |

| GATA31E08 | DXS10147 | DXS7423 | AA N=539 | AS N=326 | CN N=425 | Hisp N=411 | Overall N=1701 |
|---|---|---|---|---|---|---|---|
| 13 | 9 | 13 | 4 | 2 | 6 | 2 | 14 |
| 13 | 9 | 14 | 9 | 7 | 15 | 7 | 38 |
| 13 | 9 | 15 | 5 | 3 | 20 | 6 | 34 |
| 13 | 9 | 16 | | | 8 | 1 | 9 |
| 13 | 9 | 17 | | | 2 | 1 | 3 |
| 13 | 10 | 13 | | 1 | | | 1 |
| 13 | 10 | 14 | 2 | | | | 2 |
| 13 | 10 | 15 | 3 | | 1 | 1 | 5 |
| 13 | 10 | 16 | 1 | | | 1 | 2 |
| 13 | 11 | 14 | 1 | | | | 1 |
| 13 | 11 | 15 | | 1 | | | 1 |
| 14 | 5 | 14 | 1 | | | | 1 |
| 14 | 6 | 13 | 1 | | 1 | | 2 |
| 14 | 6 | 14 | 3 | 1 | 1 | 1 | 6 |
| 14 | 6 | 15 | | 5 | 5 | 6 | 16 |
| 14 | 6 | 16 | | | 2 | | 2 |
| 14 | 7 | 14 | 6 | 1 | 1 | | 8 |
| 14 | 7 | 15 | 4 | 2 | 1 | | 7 |
| 14 | 7 | 16 | 3 | | 2 | 1 | 6 |
| 14 | 7 | 17 | | 1 | | | 1 |
| 14 | 8 | 13 | | | 1 | | 1 |
| 14 | 8 | 14 | 3 | 2 | 6 | 2 | 13 |
| 14 | 8 | 15 | 2 | 10 | 7 | 5 | 24 |
| 14 | 8 | 16 | | | 2 | | 2 |
| 14 | 8 | 17 | | | | 2 | 2 |
| 14 | 9 | 13 | 3 | 4 | 4 | | 11 |
| 14 | 9 | 14 | 3 | 2 | 5 | 3 | 13 |
| 14 | 9 | 15 | 3 | | 6 | 1 | 10 |
| 14 | 9 | 16 | 1 | 2 | 2 | | 5 |
| 14 | 10 | 14 | | 1 | | | 1 |
| 14 | 10 | 15 | 2 | | | | 2 |
| 15 | 6 | 14 | | | 1 | | 1 |
| 15 | 6 | 15 | | 1 | | 1 | 2 |
| 15 | 7 | 15 | 2 | | | | 2 |
| 15 | 8 | 14 | 2 | | | | 2 |
| 15 | 8 | 15 | | 3 | 1 | | 4 |
| 15 | 8 | 16 | | 1 | | | 1 |
| 15 | 9 | 13 | | | 2 | | 2 |
| 15 | 9 | 14 | | | 1 | | 1 |
| 15 | 9 | 15 | 1 | 1 | | | 2 |
| 16 | 6 | 14 | | 1 | | | 1 |
| 16 | 8 | 16 | 1 | | | | 1 |
| Total number of haplotypes: | | | 119 | 84 | 87 | 94 | 162 |
| Total number of unique haplotypes: | | | 35 | 35 | 22 | 34 | 32 |
| Count of most common haplotype: | | | 28 | 24 | 20 | 31 | 72 |

**Table B13. Haplotypes observed for proposed linkage group 1 in a population from Bosnia & Herzegovina (N=86).**

| DXS8378 | DXS9902 | Haplotype Count |
|---|---|---|
| 9 | 9 | 1 |
| 9 | 10 | 1 |
| 9 | 11 | 1 |
| 9 | 12 | 2 |
| 10 | 10 | 14 |
| 10 | 11 | 8 |
| 10 | 12 | 7 |
| 11 | 9 | 4 |
| 11 | 10 | 7 |
| 11 | 11 | 8 |
| 11 | 12 | 8 |
| 11 | 12.1 | 1 |
| 12 | 9 | 2 |
| 12 | 10 | 8 |
| 12 | 10.1 | 1 |
| 12 | 11 | 7 |
| 12 | 11.1 | 1 |
| 12 | 12 | 2 |
| 13 | 10 | 2 |
| 13 | 11 | 1 |
| Total number of haplotypes: | | 20 |

**Table B14. Haplotypes observed for proposed linkage group 2 in a population from Bosnia & Herzegovina (N=86).**

| DXS7424 | DXS6789 | DXS7130 | GATA165B12 | DXS101 | GATA172D05 | DXS7132 | DXS6803 | DXS7133 |
|---|---|---|---|---|---|---|---|---|
| 10 | 20 | 14.3 | 10 | 25 | 8 | 14 | 12 | 12 |
| 10 | 20 | 15.3 | 10 | 18 | 6 | 13 | 12 | 9 |
| 12 | 20 | 14.3 | 10 | 24 | 10 | 13 | 12 | 9 |
| 13 | 15 | 16.3 | 10 | 19 | 8 | 14 | 13 | 9 |
| 13 | 20 | 14.3 | 9 | 25 | 10 | 13 | 11 | 11 |
| 13 | 20 | 14.3 | 12 | 25 | 10 | 14 | 12 | 9 |
| 13 | 20 | 16.3 | 10 | 25 | 6 | 14 | 11.3 | 10 |
| 13 | 21 | 16.3 | 9 | 25 | 11 | 14 | 13.3 | 9 |
| 13 | 22 | 14.3 | 10 | 26 | 9 | 12 | 13 | 9 |
| 13 | 22 | 16.3 | 11 | 15 | 11 | 13 | 12.3 | 9 |
| 14 | 15 | 15.3 | 11 | 27 | 9 | 13 | 13 | 9 |
| 14 | 15 | 16.3 | 11 | 24 | 6 | 13 | 13 | 9 |
| 14 | 19 | 14.3 | 9 | 23 | 8 | 13 | 11 | 9 |
| 14 | 20 | 14.3 | 11 | 24 | 11 | 13 | 11 | 9 |
| 14 | 20 | 15.3 | 10 | 27 | 10 | 13 | 12.3 | 9 |
| 14 | 20 | 16.3 | 10 | 23 | 10 | 15 | 13.3 | 9 |
| 14 | 20 | 16.3 | 11 | 25 | 10 | 15 | 13 | 10 |
| 14 | 21 | 14.3 | 9 | 24 | 8 | 13 | 12.3 | 9 |
| 14 | 21 | 14.3 | 10 | 25 | 8 | 16 | 11 | 10 |

| DXS7424 | DXS6789 | DXS7130 | GATA165B12 | DXS101 | GATA172D05 | DXS7132 | DXS6803 | DXS7133 |
|---|---|---|---|---|---|---|---|---|
| 14 | 21 | 17.3 | 9 | 25 | 8 | 15 | 13.3 | 13 |
| 14 | 22 | 12 | 11 | 28 | 6 | 14 | 10 | 9 |
| 14 | 22 | 14.3 | 9 | 24 | 10 | 14 | 12 | 11 |
| 14 | 22 | 14.3 | 10 | 25 | 6 | 12 | 11 | 10 |
| 14 | 22 | 16.3 | 10 | 28 | 10 | 14 | 13.3 | 10 |
| 14 | 22 | 16.3 | 11 | 24 | 6 | 13 | 13.3 | 11 |
| 15 | 15 | 15.3 | 11 | 20 | 11 | 14 | 13.3 | 9 |
| 15 | 19 | 15.3 | 10 | 24 | 11 | 12 | 13.3 | 10 |
| 15 | 20 | 11 | 11 | 25 | 10 | 13 | 11 | 9 |
| 15 | 20 | 12 | 10 | 24 | 11 | 13 | 11 | 11 |
| 15 | 20 | 14.3 | 11 | 28 | 11 | 13 | 13 | 11 |
| 15 | 20 | 15.3 | 9 | 26 | 10 | 14 | 12 | 11 |
| 15 | 20 | 15.3 | 10 | 24 | 6 | 14 | 11 | 11 |
| 15 | 20 | 15.3 | 10 | 24 | 8 | 14 | 14.3 | 9 |
| 15 | 20 | 15.3 | 10 | 24 | 12 | 13 | 12.3 | 9 |
| 15 | 20 | 15.3 | 10 | 26 | 12 | 14 | 13.3 | 11 |
| 15 | 20 | 15.3 | 11 | 23 | 10 | 15 | 13.3 | 9 |
| 15 | 20 | 16.3 | 9 | 24 | 9 | 17 | 13 | 11 |
| 15 | 21 | 14.3 | 11 | 24 | 6 | 14 | 13.3 | 10 |
| 15 | 21 | 14.3 | 11 | 25 | 6 | 13 | 11 | 9 |
| 15 | 21 | 15.3 | 9 | 24 | 8 | 14 | 10 | 11 |
| 15 | 21 | 15.3 | 9 | 26 | 8 | 13 | 10 | 10 |
| 15 | 21 | 15.3 | 10 | 15 | 6 | 14 | 12 | 9 |
| 15 | 21 | 15.3 | 11 | 19 | 11 | 14 | 12 | 11 |
| 15 | 21 | 16.3 | 9 | 19 | 11 | 14 | 12 | 9 |
| 15 | 21 | 16.3 | 10 | 24 | 6 | 14 | 12.3 | 9 |
| 15 | 21 | 17.3 | 9 | 24 | 6 | 12 | 11 | 10 |
| 15 | 21 | 17.3 | 11 | 19 | 11 | 14 | 13.3 | 9 |
| 15 | 22 | 14 | 10 | 27 | 9 | 14 | 12.3 | 9 |
| 15 | 22 | 15.3 | 9 | 26 | 6 | 13 | 13.3 | 11 |
| 15 | 22 | 15.3 | 11 | 18 | 6 | 13 | 13.3 | 10 |
| 15 | 22 | 15.3 | 11 | 28 | 8 | 14 | 13 | 12 |
| 16 | 15 | 13.3 | 11 | 15 | 12 | 13 | 11 | 13 |
| 16 | 20 | 12 | 10 | 20 | 11 | 13 | 13 | 9 |
| 16 | 20 | 13.3 | 9 | 16 | 11 | 14 | 13.3 | 11 |
| 16 | 20 | 13.3 | 10 | 28 | 8 | 15 | 14 | 11 |
| 16 | 20 | 13.3 | 11 | 16 | 11 | 14 | 12 | 11 |
| 16 | 20 | 14 | 9 | 18 | 6 | 15 | 11 | 9 |
| 16 | 20 | 14.3 | 10 | 26 | 10 | 13 | 12 | 9 |
| 16 | 20 | 15.3 | 10 | 15 | 8 | 15 | 11 | 9 |
| 16 | 20 | 15.3 | 10 | 27 | 10 | 14 | 12 | 11 |
| 16 | 20 | 15.3 | 11 | 23 | 6 | 13 | 11 | 9 |
| 16 | 20 | 15.3 | 11 | 27 | 10 | 13 | 12 | 9 |
| 16 | 20 | 15.3 | 12 | 19 | 11 | 14 | 13 | 11 |
| 16 | 20 | 15.3 | 12 | 25 | 6 | 13 | 11 | 11 |
| 16 | 20 | 16.3 | 11 | 23 | 11 | 14 | 13 | 11 |
| 16 | 20 | 16.3 | 11 | 28 | 8 | 14 | 12.3 | 11 |
| 16 | 20 | 16.3 | 11 | 28 | 10 | 13 | 12 | 10 |
| 16 | 21 | 15.3 | 11 | 24 | 10 | 14 | 12.3 | 11 |
| 16 | 22 | 12 | 9 | 28 | 11 | 15 | 11 | 11 |
| 16 | 22 | 13 | 11 | 26 | 8 | 16 | 12 | 10 |
| 16 | 22 | 15.3 | 10 | 25 | 10 | 15 | 12 | 9 |
| 16 | 22 | 16.3 | 11 | 25 | 10 | 13 | 11 | 9 |
| 17 | 20 | 12 | 9 | 25 | 10 | 14 | 11.3 | 11 |
| 17 | 20 | 13.3 | 9 | 22 | 10 | 13 | 13 | 13 |

| DXS7424 | DXS6789 | DXS7130 | GATA165B12 | DXS101 | GATA172D05 | DXS7132 | DXS6803 | DXS7133 |
|---|---|---|---|---|---|---|---|---|
| 17 | 20 | 14.3 | 9 | 25 | 11 | 14 | 13 | 11 |
| 17 | 20 | 14.3 | 11 | 26 | 10 | 14 | 12.3 | 9 |
| 17 | 20 | 15.3 | 9 | 24 | 8 | 14 | 11 | 9 |
| 17 | 20 | 15.3 | 11 | 25 | 12 | 12 | 12 | 11 |
| 17 | 21 | 16.3 | 11 | 24 | 10 | 13 | 12.3 | 11 |
| 17 | 22 | 12 | 11 | 25 | 12 | 14 | 12 | 10 |
| 17 | 22 | 14.3 | 9 | 24 | 8 | 15 | 13.3 | 11 |
| 17 | 22 | 15.3 | 11 | 25 | 10 | 13 | 12 | 11 |
| 17 | 22 | 16.3 | 10 | 18 | 6 | 14 | 11 | 10 |
| 18 | 20 | 11 | 10 | 24 | 10 | 13 | 12.3 | 9 |
| 18 | 22 | 11 | 10 | 28 | 10 | 15 | 11 | 11 |
| 19 | 20 | 16.3 | 10 | 15 | 10 | 15 | 13 | 11 |
| | | | | | | | Total number of haplotypes: | 86 |

**Table B15. Haplotypes observed for proposed linkage group 4 in a population from Bosnia & Herzegovina (N=86).**

| GATA31E08 | DXS7423 | Haplotype Count |
|---|---|---|
| 9 | 13 | 3 |
| 9 | 14 | 5 |
| 9 | 15 | 4 |
| 9 | 16 | 4 |
| 9 | 17 | 1 |
| 10 | 13 | 2 |
| 10 | 14 | 2 |
| 10 | 15 | 1 |
| 11 | 13 | 3 |
| 11 | 14 | 3 |
| 11 | 15 | 3 |
| 11 | 16 | 2 |
| 12 | 13 | 2 |
| 12 | 14 | 4 |
| 12 | 15 | 5 |
| 12 | 16 | 2 |
| 13 | 13 | 3 |
| 13 | 14 | 10 |
| 13 | 15 | 13 |
| 13 | 16 | 4 |
| 14 | 13 | 1 |
| 14 | 14 | 2 |
| 14 | 15 | 5 |
| 14 | 16 | 2 |
| | Total number of haplotypes: | 24 |

# Appendix C. Additional Publications

*Peer-reviewed Publications*

**C1.** Toni M. Diegoli, Michael D. Coble, Development and characterization of two mini-X chromosomal short tandem repeat multiplexes, Forensic Sci. Int. Genet. 5 (2011) 415-421.

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## Development and characterization of two mini-X chromosomal short tandem repeat multiplexes

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ABSTRACT

This study presents the development and characterization of two X chromosomal short tandem repeat (STR) multiplexes utilizing reduced-size amplicons (less than 200 base pairs) for identity and kinship testing with degraded DNA. Approximately 1360 samples across 4 U.S. population groups were typed for 15 X chromosomal STR markers: DXS6789, DXS7130, DXS9902, GATA31E08, DXS7424, GATA165B12, DXS101, DXS6795, GATA172D05, DXS10147, DXS8378, DXS7132, DXS6803, HPRTB, and DXS7423. A high degree of polymorphism was observed for each marker and both multiplexes were sensitive down to 200 pg of pristine DNA. The two proposed multiplexes are suitable for forensic use, and show potential for improved analysis of compromised bone samples.

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### 1. Introduction

The multiplex detection and analysis of short tandem repeat (STR) markers is a common tool used for genetic identity testing in the forensic setting. Numerous publications have characterized genetic markers located throughout the autosomes and male-specific Y chromosome that can be used for this purpose. More recently, markers located on the X chromosome have emerged as additional tools in this forensic arsenal. X chromosomal STRs can be used to supplement traditional kinship testing due to their unique inheritance pattern and, correspondingly, the breadth of published literature on the subject has expanded greatly in recent years.

For both the Y chromosome and the autosomes, commercial kits are available that probe a wide variety of genetic markers (see Refs. [1–5], for example). When it comes to the X chromosome, however, only one kit is currently being manufactured with limited availability, the Investigator Argus X12 kit (Qiagen, Hilden, Germany). This kit simultaneously amplifies and detects twelve X chromosomal STRs (DXS8378, HPRTB, DXS7423, DXS7132, DXS10134, DXS10074, DXS10101, DXS10103, DXS10148, DXS10146, DXS10079, and DXS10135) plus amelogenin in four fluorescent dye channels [6]. With amplicon sizes ranging from approximately 104 to 375 base pairs (bp), the kit's detection limit is 100 pg of input DNA [6]. Noncommercial multiplex assays have also been published, amplifying 2–12 loci in a single reaction (see Refs. [7–19], for example).

At the Armed Forces DNA Identification Laboratory (AFDIL), kinship testing is routinely used to support the identification of skeletal remains. Many times, mitochondrial DNA (mtDNA) typing will reveal the answer to the question of identity, particularly in closed populations and when a direct maternal reference is available. However, where maternal references are unavailable or where the unidentified individual matches one of the most common mtDNA haplotypes, mtDNA testing alone may be inadequate. Sufficient statistical power must then result from fewer, smaller-amplicon STR loci or low copy number analyses. In such cases, markers on the X chromosome may provide additional information. Consequently, the selection of candidate X chromosomal markers and the development of these markers into STR multiplexes with reduced amplicon sizes (or "mini-STRs") offers the potential to augment both traditional STR testing and mtDNA sequencing. Other applications of X chromosomal STRs include immigration or maternity cases as well as paternity cases with female children. X chromosomal markers may also prove useful as a complement to mtDNA and Y chromosomal markers in the study of human evolutionary history [20].

In general, STR markers are selected for forensic use based upon their observed heterozygosity, discriminatory power, and repeat size/structure. Another criterion for selecting candidate STR markers

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is pairwise linkage; in order to take advantage of the product rule in the calculation of random match statistics, markers should not be linked to one another. Marker selection restricted to a single chromosome introduces an additional challenge because some loci will necessarily be linked. Additionally, because DNA templates encountered in the forensic setting are often degraded (for example, as the result of prolonged exposure to environmental extremes), amplicon size should be considered in selecting potential markers. In such cases, shorter amplicon sizes are favored with the goal of recovering the maximum number of alleles [21]. In a study examining degraded samples, Asamura et al. demonstrated the success of two X chromosomal STR quadruplex reactions consisting of amplicons ranging from 76 to 169 bp in length [13].

Here, two complementary mini-X chromosomal STR multiplexes, one 8-plex and one 10-plex, were assembled with the above parameters in mind. Markers were selected to maintain high heterozygosity and represent all four X chromosomal linkage groups identified thus far [22,23]. Further, all amplicon sizes were kept small (<190 bp) to facilitate application to degraded samples. This study describes the development and optimization of these two multiplexes as well as their application to the analysis of four U.S. population groups: African Americans, U.S. Asians, U.S. Caucasians, and U.S. Hispanics.

## 2. Materials and methods

### 2.1. Selection of markers and primer design

A review of X chromosomal STR literature resulted in a list of potential markers. Forensic utility was assessed according to the following criteria: (a) potential for small amplicon size; (b) heterozygosity in published materials; and (c) distribution between the proposed linkage groups across the X chromosome. Markers best matching these criteria (Table 1) were chosen for inclusion into two multiplexes and organized according to amplicon size (Table 2).

Approximately 200 bp flanking either side of the repeat regions for the chosen markers were downloaded from the UCSC Genome Browser [24] using their BLAT In Silico PCR search [25] and the published primers. In many cases, published amplification primers were sufficient for incorporation into the multiplexes, but several markers required one or both primers be redesigned (see Section 3.1). When necessary, primers were designed using the web-based program Primer3 [26], and selected primers were screened for use in multiplex reactions using the web-based algorithm AutoDimer [27].

One primer for each marker was labeled at the 5′ end with a fluorescent dye, either 6FAM, VIC, NED, or PET (Applied Biosystems, Foster City, CA). A tail was added to the complementary primer in the set at the 5′ end in order to promote the complete adenylation of PCR products [28] and, in some cases, provide adequate spacing between amplicons in the multiplex. This tail was either GTTTCTT, ATT, or a single G (Table 2).

### 2.2. Source and extraction of DNA samples

Unrelated, anonymous bloodstains represented four U.S. populations: African Americans (174 females, 175 males), U.S. Asians (300 females, 201 males), U.S. Caucasians (146 females, 122 males), and U.S. Hispanics (122 females, 123 males). Bloodstain cards were extracted on the Biomek® 2000 robot (Beckman Coulter, Brea, CA) using the DNA IQ™ system (Promega Corporation, Madison, WI) or on the Qiagen 9604 robot using the Qiagen QIAmp DNA kit (Qiagen, Gaithersburg, MD). Three control DNA standards – 9948 (Applied Biosystems and Promega Corporation), 9947a (Applied Biosystems), K562 (Promega Corporation) – were also characterized, as per the recommendations of Szibor et al. [29]. Results were compared to published profiles before allele designations were made (see Section 3.2 and Table 1).

### 2.3. STR amplification, detection, and analysis

Amplification was performed in a 10 μL reaction that consisted of 1× PCR buffer II (Applied Biosystems), 2 units AmpliTaq Gold™ DNA polymerase (Applied Biosystems), 0.25 mM dNTP Mix (Applied Biosystems), 0.15 mg/mL bovine serum albumin (Sigma–Aldrich, St. Louis, MO), 2 mM magnesium chloride solution (Applied Biosystems) and 2 μL of primer mix. Primer mix concentrations were adjusted empirically to balance peak heights within each multiplex and individual concentrations are listed in Table 2. Thermal cycling was performed on a GeneAmp® 9700 (Applied Biosystems) using the following parameters: initial incubation at 96 °C for 10 min, amplification with 30 cycles of 94 °C for 1 min, 60 °C for 1 min, and 72 °C for 1 min, extension at 60 °C for 45 min, and final soak at 4 °C.

Samples were prepared for capillary electrophoresis by adding1 μL amplified product to 8.7 μL Hi-Di™ formamide (Applied Biosystems) and 0.3 μL LIZ-500 size standard (Applied Biosystems). Samples were injected at 3.0 kV for 10 s and run using a 36 cm array and POP6 on a 3130xl Genetic Analyzer (Applied Biosystems). Data were analyzed using Genemapper ID version 3.2 or ID-X version 1.1 (Applied Biosystems). As mentioned, allele designations were achieved through comparison with control DNAs 9947a, 9948, and K562. Final bins and panels were created based upon an average of all alleles observed in the profiles of population samples.

**Table 1**
Characteristics of the 15 X-STRs examined in this study.

| Marker name | Observed allele range | Repeat motif | Nomenclature reference | Linkage group[a] | DNA profile 9947a |
|---|---|---|---|---|---|
| DXS6789 | 14–25 | (TATC)(0-1)-(TATG)x-(TATC)y | [44] | 2 | 21, 22 |
| DXS7130 | 9–14, 16, 13.3–18.3 | (TATC)5-ATC(0-1)-(TATC)x | [47] | 2 | 15.3, 15.3 |
| GATA31E08 | 7–16 | (AGGG)x-(AGAT)y | [52] | 4 | 13, 13 |
| DXS7424 | 9–20 | TAA | [43] | 2 | 14, 16 |
| GATA165B12 | 8–13 | AGAT | [45] | 2 | 9, 11 |
| DXS101 | 14–31, 33 | (CTT)x-(ATT)y | [50] | 2 | 24, 26 |
| DXS6795 | 6, 8–17 | ATT-ATC(0-1)-(ATT)x | This study | 1 | 12, 13 |
| GATA172D05 | 6–13 | TAGA | [46] | 2 | 10, 10 |
| DXS10147 | 5–11 | AAAC | [49] | 4 | 8, 8 |
| DXS8378 | 8–15 | CTAT | [43] | 1 | 10, 11 |
| DXS7132 | 10–18, 16.3 | (TCTA)x-(TCA)(0-1)-(TCTA)2 | [46] | 2 | 12, 12 |
| DXS6803 | 7–14, 16, 10.3–14.3 | (TCTA)x-(TCA)(0-1)-TCTA | [47] | 2 | 11.3, 12 |
| HPRTB | 7–16 | ATCT | [43,48] | 3 | 14, 14 |
| DXS7423 | 8, 12–17 | (TCCA)3-(N8)(0-1)-(TCCA)x | [46,51] | 4 | 14, 15 |
| DXS9902 | 7–14, 10.1–12.1 | GATA | [43] | 1 | 11, 11 |

[a] According to Refs. [22,23,33]. N8 = TCTGTCCT.

**Table 2**
Mini-X STR primers sequences and concentrations used in this study.

| Marker name | Primer sequence (5′–3′) | Reference | Multiplex | Final primer concentration (μM) | Amplicon size range (bp) |
|---|---|---|---|---|---|
| DXS6789 | 6FAM-CCTCGTGATCATGTAAGTTGG | [13] | 1 | 0.8 | 124–168 |
| | **ATT**CAGAACCAATAGGAGATAGATGGT | [13] | | 0.8 | |
| DXS7130 | VIC-AATATAGAGGAAGGGGAAATCATTA | This study | 1 | 1.6 | 93–136 |
| | **ATT**CAAAGAAATGAGAACAAAAATCAGG | This study | | 1.6 | |
| GATA31E08 | NED-CAGAGCTGGTGATGATAGATGA | [13] | 1 | 1.2 | 95–131 |
| | **ATT**CTCACTTTTATGTGTGTATGTATCTCC | [13] | | 1.2 | |
| DXS7424 | NED-GGACTGCTTGAGTCCAGGAA | This study | 1 | 1.2 | 146–185 |
| | **G**GGAACACGCACATTTGAGAA | This study | | 1.2 | |
| GATA165B12 | PET-TCATCAATCATCTATCCGTATATCA | [13] | 1 | 3.2 | 92–112 |
| | **ATT**AAGTTGACTGTGATTCCTGGTTT | [13] | | 3.2 | |
| DXS101 | **ATT**TCTCCCTTCAAAAACAAAGATAA | [13] | 1 | 1.2 | 126–177 |
| | PET-TGCATATTCTGCGCATGT | [13] | | 1.2 | |
| DXS9902 | VIC-TGGA GTCTCTGGGTGAAGAG | [45] | 1 | 0.4 | 167–191 |
| | **ATT**CAGGAGTATGGGATCACCAG | [45] | | 0.4 | |
| DXS6795 | 6FAM-TGACATGGCTTTCTTTACAATTAC | This study | 2 | 0.6 | 90–111 |
| | **G**CCATGTTACATAAACAAGGAGTTATG | This study | | 0.6 | |
| GATA172D05 | 6FAM-**GTTTCTT**TAGTGGTGATGGTTGCACAG | [46] | 2 | 2.8 | 122–150 |
| | **GTTTCTT**ATAATTGAAAGCCCGGATTC | [46] | | 2.8 | |
| DXS10147 | 6FAM-AGGAGGTGAAGGTTGTGGTG | This study | 2 | 0.2 | 165–185 |
| | **ATT**TGGGACTCTTCCCTTAAATGC | This study | | 0.2 | |
| DXS8378 | VIC-GCTCCTGGCAGGTCACTATC | [13] | 2 | 1.6 | 94–118 |
| | **ATT**GCGACAAGACGCCAAACTCCA | [13][a] | | 1.6 | |
| DXS7132 | VIC-AATAGTGTGAGCCCATTTTCA | This study | 2 | 0.4 | 149–177 |
| | **GTTTCTT**GCCAAACTCTATTAGTCAAC | [46] | | 0.4 | |
| DXS6803 | NED-GAAATGTGCTTTGACAGGAA | [47] | 2 | 0.4 | 102–130 |
| | **G**CAAAAAGGAACATATGCTACTT | [47][b] | | 0.4 | |
| HPRTB | NED-TCTCTATTTCCATCTCTGTCTCC | [33] | 2 | 0.4 | 148–176 |
| | **GT**CACCCCTGTCTATGGTCTCG | [33] | | 0.4 | |
| DXS7423 | PET-AGATTTCCTCCCCATCCATC | [13] | 2 | 1.2 | 85–125 |
| | **ATT**TTGTCACACAAATAAATGAATGAGT | [13] | | 1.2 | |
| DXS9902 | PET-TGGAGTCTCTGGGTGAAGAG | [45] | 2 | 1.2 | 167–191 |
| | **ATT**CAGGAGTATGGGATCACCAG | [45] | | 1.2 | |
| SRY | NED-AAAAATTGGCGATTAAGTCAAA | This study | Both | 0.4 | 86 |
| | **GT**TGACTACTTGCCCTGCTGA | This study | | 0.4 | |

Bases in bold are tails added to the primer sequence to promote complete adenylation of the amplicon. Amplicon size ranges include tails.
[a] Basic primer sequence obtained from publication with underlined bases modified in this study to prevent artifact formation.
[b] Basic primer sequence obtained from publication with underlined base modified in this study to match GenBank sequence.

### 2.4. Sensitivity and degraded sample testing

The initial concentration of three female and two male samples was determined using the Quantifiler® Human DNA Quantification Kit (Applied Biosystems). Samples were then serially diluted with Tris–low-EDTA buffer (TLE; 10 mM Tris, 0.1 mM EDTA, pH 8.0) to form the following dilution series: 1000, 500, 200, 100, 50, and 25 pg/μL. One microliter of each concentration was tested in duplicate to determine the minimum quantity of input DNA required to reliably obtain full profiles for both males and females with each multiplex.

Five bone samples greater than 65 years old were tested to determine the multiplexes' utility on potentially degraded DNA. DNA was extracted from bones using a procedure that results in the total demineralization of the bone matrix [30]. Previously, PowerPlex® 16 (Promega, Madison, WI) testing with a modified protocol (increased Taq and increased PCR cycle number) was performed on these bone samples and dropout at many of the larger loci (>300 bp) was observed, indicating DNA degradation. For mini-X STR testing, the total volume of the amplification reaction was increased to allow greater extract input volume and an increased quantity of polymerase. All volumes were adjusted proportionally from the standard volume (10 μL) to 25 μL, with up to 8 units AmpliTaq Gold™ DNA polymerase used per reaction.

### 2.5. DNA sequencing

A minimum of five male samples for each X chromosomal STR marker were sequenced to confirm allele designation and repeat structure. In order to fully characterize both the number of repeating units and flanking sequence, a set of sequencing primers located outside the target STR amplicon was designed as outlined above (Supplementary Table 1). Template DNA was amplified in a 50 μL reaction consisting of 1× PCR buffer, 0.4 μM each forward and reverse primer (Applied Biosystems), 0.2 mM dNTPs, and 2.5 units AmpliTaq Gold™ DNA polymerase. Thermal cycling was performed on a GeneAmp® 9700 (Applied Biosystems) using the following parameters: initial incubation at 96 °C for 10 min, amplification with 36 cycles of 94 °C for 30 s, 56 °C for 30 s, and 72 °C for 1 min, extension at 72 °C for 7 min, and final soak at 4 °C. PCR products were then treated with ExoSAP-IT® (USB Corporation, Cleveland, OH) to remove excess primers and dNTPs.

Cycle sequencing was performed using the BigDye® Terminator v1.1 kit (Applied Biosystems). The sequencing reactions consisted of 8 μL $dH_2O$, 6 μL 2.5× Sequencing Buffer (400 mM Tris, 10 mM $MgCl_2$, pH 9.0), 1.5 μL BigDye® Terminator v1.1 Ready Reaction Mix, 0.5 μL dGTP mix, 2 μL 10 μM forward or reverse primer, and 2 μL amplified product. Thermal cycling was performed on a GeneAmp® 9700 (Applied Biosystems) using the following parameters: initial incubation at 96 °C for 1 min, amplification with 25 cycles of 96 °C for 15 s, 50 °C for 5 s, and 60 °C for 2 min, and final soak at 4 °C.

Sequencing products were purified using a Performa® DTR 96-well plate (Edge Biosystems, Gaithersburg, MD) and 1 μL filtrate was added to 9 μL Hi-Di™ formamide for injection onto the 3130xl or 3730 instruments using either POP6 polymer and a 36 cm array or POP7 polymer and a 50 cm array, respectively. Sequences were aligned to a GenBank reference sequence and analyzed using Sequencher® version 4.1, 4.7, or 4.8 (GeneCodes, Ann Arbor, MI).

### 2.6. Statistical analysis

Allele frequencies were calculated by hand using a spreadsheet program. The software PowerMarker [31] was used to determine both the observed heterozygosity value (H(obs)) and the *p* value of the exact test for Hardy–Weinberg equilibrium (*p*(HWE)) using only the female genotype data. All other forensic efficiency statistics (expected heterozygosity (H(exp)), polymorphism information content (PIC), power of discrimination in both males (PDm) and females (PDf), mean exclusion chance in trios involving a daughter (MECI) and in father/daughter duos (MECII) [32]) were calculated using the Forensic ChrX Research website version 2.0 [33]. Pairwise genetic distance and linkage disequilibrium calculations were performed in Arlequin v3.1 [34].

## 3. Results and discussion

### 3.1. Organization of the multiplexes

Organization and selection of 15 markers were based upon forensic utility as described above (small amplicon size, heterozygosity values, and distribution among the proposed linkage groups) as well as practical considerations (arrangement within two multiplexes, spacing between allele ranges for markers within the same dye channel, primer compatibility, successful amplification, etc.). Singleplex amplifications were performed first to evaluate primer selection for successful amplification, complete adenylation, and peak migration; multiplex testing then confirmed the success of the typing process. At this stage, primer sets were redesigned when necessary to address practical issues. For example, the STR amplification primers for marker DXS7424 were redesigned to avoid three individual primer binding site mutations that occurred commonly in the U.S. populations examined when using previously published primers (manuscript in preparation). The organization of the chosen markers resulted in two final multiplexes, and primers are listed in Table 2. Note that marker DXS9902 was included in both multiplexes for concordance. Gender confirmation in the form of a marker located within the sex-determining region of the Y chromosome (SRY) [35,36] was also included in both multiplexes. SRY was chosen over the more widely-used amelogenin locus in order to avoid known cases of amelogenin allele dropout (see Refs. [37–42], for example). Additionally, the four markers that are also present in the commercially available kit Argus X8 (DXS8378, DXS7132, HPRTB, and DXS7423) were included in a single multiplex (multiplex 2), allowing concordance to be performed with ease between laboratories using the multiplex(es) proposed here and those using the commercially available kit. Example electropherograms for the two final multiplexes are shown in Supplementary Fig. 1.

### 3.2. Nomenclature

Sample genotyping followed published allele nomenclature according to [29,43–52] and guidelines set forth by the DNA Commission of the International Society for Forensic Genetics (ISFG) [53]. Table 1 lists the repeat motif and profile of 9947a for all markers; other control DNAs matched the profiles published in [29] and on the Forensic ChrX Research website version 2.0 except for markers GATA31E08 and DXS6803. All control DNAs were sequenced to confirm the repeat structure with primers listed in Supplementary Table 1. Sequencing results for marker GATA31E08 revealed additional variation resulting from AGGG repeats before the reported AGAT repeat unit. Most commonly, two AGGG repeats are present before the AGAT units, and variation results from additional AGAT repeats (also noted in Ref. [52]). Including the initial AGGG repeats increases the allele designation for this marker by two repeat units compared to that using solely the AGAT repeat unit. Further, though there is no conflict with the published repeat unit (TCTA) for marker DXS6803, the allele designation used in this study differs from the published profiles [29] of control DNAs 9947a, 9948, and K562 by +1 repeat unit. Additionally, the observed profile of 9947a was a heterozygote 11.3, 12 (Table 1), rather than a homozygote 11.

Of note, GATA172D05 allele calls were determined based upon the TAGA repeat as suggested by Edelmann et al. [46], rather than the GATA repeat used in earlier studies [43]. This distinction results in one additional repeat unit, for a corrected allelic range of 6–13 repeats (rather than 5–12). The repeat motif for marker HPRTB has also been published with conflicting nomenclature. An overview of the discrepancies and a recommendation to use the AGAT repeat motif according to Ref. [55] can be found in Ref. [48]. In the present study, the ATCT coding strand equivalent of this recommended repeat unit was used and is in agreement with Ref. [29].

Though the ISFG guidelines suggest that the first 5′ nucleotides that can define a repeat motif for an STR marker should be used, there were several situations where this recommendation was not followed. Instead, allele numbering was based upon published and widely-used repeat motifs in favor of maintaining consistency with an established historical nomenclature and avoiding unnecessary confusion, which is itself in accordance with the ISFG guidelines [53]. For example, at marker DXS9902, the first 5′ nucleotides that define a repeat motif are TAGA [54]; however, the GATA repeat unit nomenclature first published in Ref. [43] was used instead, which results in a one repeat unit difference. Similarly, although the repeat motif AAT may be the most 5′ motif for DXS7424 [56], the published and widely-used TAA repeat unit [43] (which also results in a one repeat unit difference) was employed for allele designation here.

As evidenced by these 15 markers, further study is needed in order to reach a consensus regarding both STR nomenclature and allele designation in the community of X chromosome researchers. In the future, it might be useful for publications to include the nomenclature used for assigning allele number in addition to the control DNA profiles already suggested by Szibor et al. [29] in order to facilitate comparisons between different groups.

### 3.3. Sensitivity and degraded sample testing

In this study, full STR profiles were reliably obtained with as little as 200 pg of input DNA (Supplementary Fig. 2A), with a loss of only 1–2 markers at 100 pg. Because there are multiple markers from each linkage group present in the multiplexes (except for linkage group 3), the information obtained even with a loss of 1–2 markers per multiplex will still be adequate in most situations. Below 100 pg, typing was less reliable when using the standard protocol described here, and a low copy number approach employing some combination of replicate analyses, increased cycle number, or increased polymerase could be considered. There was no difference observed between the two multiplexes, or between male and female profiles in this study.

Typing attempts on bone samples that were decades old revealed partial to full profiles with both multiplexes (Supplementary Fig. 2B). With this particular set of samples, multiplex 2 performed better, resulting in a full profile for two of the five bone samples tested, whereas multiplex 1 did not produce any full profiles. Further testing will be necessary in order to assess the true utility of these multiplexes with degraded DNA.

### 3.4. Alleles

Allele frequencies obtained for the 15 X chromosomal STRs examined in four U.S. population groups are shown in Supplemen-

tary Table 2. The distribution of allele frequencies in the male and female subjects was examined using the chi-square test for independence. The resultant $p$ values showed no significant differences ($p > 0.05$) and frequency data was pooled at each marker. In total, 158 alleles were observed across 15 markers, with 6–19 alleles at each marker.

Several alleles that had not previously been noted in the literature at the time of publication were observed in this dataset: 10.1 at DXS9902; 6, 8, 16 and 17 at DXS6795; 5 and 11 at DXS10147; 33 at DXS101; and 16 at DXS6803. All new alleles were inferred based upon electrophoretic mobility and then sequenced to confirm the uniformity and number of repeats. These new alleles are rare, occurring in populations for which little published genetic data is available. The 10.1 allele at DXS9902 was observed exclusively in the U.S. Hispanic population group while new alleles at DXS10147 and 33 at DXS101 were observed in both the African American and U.S. Hispanic population groups. Similarly, new alleles at DXS6795 and 16 at DXS6803 were observed only in the African American population.

The rare 8 allele at marker DXS7423 has thus far only been observed in populations from Africa or with African admixture, and an African specificity of this allele has been postulated [54]. The data presented here seem to be in agreement with this hypothesis, as the 8 allele was observed solely in the African American population (Supplementary Table 2). Other rare alleles were noted in populations in which they had not been previously observed. For example, alleles 7 and 10.3 at DXS6803 as well as 13 at GATA165B12 have previously been seen in Asian populations [13,57–60] but were noted in the African American and/or U.S. Hispanic population groups exclusively in this study. Similarly, microvariant alleles at marker DXS7132 (15.3, 16.3, 17.3, 18.3) have only been observed in South American individuals from Brazil, Argentina, and Columbia [19], suggesting a Native American origin of these alleles [54]. Here, the only microvariant observed (16.3) was found in the African American population group.

### 3.5. Null alleles

Null alleles were noted for four different markers: GATA172D05, GATA165B12, GATA31E08, and DXS7132. Loss of an allele at GATA172D05 was observed in one U.S. Hispanic sample. Sequencing of the sample revealed a nucleotide substitution (G → A) in the reverse primer binding site located 7 bases from the 3′ end. The same mutation and resultant null allele has been observed once before in a U.S. Hispanic sample, where 377 chromosomes were studied [16]. Here, the frequency was even less, 1 in 621 male chromosomes, though it cannot be ruled out that an additional allele carrying this mutation went undetected amongst the females. Similarly, one male null was observed for each of the other three markers: African American samples at GATA165B12 and GATA31E08, and a U.S. Hispanic sample at DXS7132. Sequencing confirmed the location of a nucleotide substitution under a primer binding site and the correct allele call (data not shown). Due to the rarity of these mutations, no measures were taken to change the primer sequences for these markers.

### 3.6. Forensic efficiency parameters

Forensic efficiency parameters calculated for all four population groups are shown in Supplementary Table 2. Marker DXS101 showed the highest observed heterozygosity values for the African American (0.9023) and U.S. Asian (0.8433) populations while the highest value for the U.S. Caucasian population was at DXS7424 (0.8562) and at GATA31E08 for the U.S. Hispanic population (0.8607). The lowest values varied by population, none of which possessed the lowest value at the same marker. The marker with the lowest observed heterozygosity value was DXS8378 (0.6322) in the African American population, DXS7423 (0.5233) in the U.S. Asian population, DXS6795 (0.5685) in the U.S. Caucasian population, and GATA165B12 (0.6148) in the U.S. Hispanic population. All markers, however, possessed high forensic efficiency values in the majority of the studied populations, confirming their utility for forensic purposes. The high MEC values for the selected markers support the potential of the two multiplexes in a certain specific kinship situations involving female offspring, while the values for the power of discrimination in both males and females indicate their usefulness in forensic identity testing.

Departures from Hardy–Weinberg equilibrium (indicated by a $p$ value for the exact test that is less than 0.05; shown in bold in Supplementary Table 2) occurred in only 2 of the 60 marker–population combinations: the U.S. Caucasian population at DXS10147 ($p = 0.0356$) and the U.S. Asian population at DXS101 ($p = 0.0003$). These deviations observed could potentially be due to population sampling effects.

### 3.7. Comparisons of pairs of population samples by marker

Population pairwise Fst values were calculated at each marker for the four U.S. population groups using male and female allele frequency data ($p < 0.05$; data not shown). All markers investigated showed significant genetic distances between at least 3 pairs of populations (out of 6); most markers showed significance for 5 or all 6 pairs. Consequently, the four U.S. populations should not be pooled into a single database for use with these two multiplexes, but rather each treated independently for forensic statistical analysis purposes.

At the time of publication, there were few publications that investigated the distributions of X chromosomal STRs in U.S. population groups, and none that encompassed all 15 markers and 4 population groups from this study. However, seven markers (DXS8378, HPRTB, GATA172D05, DXS7423, DXS7132, DXS101, and DXS6789) and three population groups (African Americans, U.S. Asians, and U.S. Hispanics) from Ref. [16] overlap with those studied here. A comparison of the allele frequency distributions across these seven markers for the three population groups using the chi-square test revealed no significant differences ($p < 0.05$; data not shown) except for the U.S. Hispanic population at marker GATA172D05 ($p = 0.04$). Similarly, there was no difference in the allele frequency distribution at marker DXS101 for the African American, U.S. Asian, U.S. Hispanic, and U.S. Caucasian populations presented here, and those published for similar populations from Ref. [61].

### 3.8. Linkage disequilibrium

The likelihood ratio test for linkage disequilibrium was performed for all pairs of markers in the four population groups using only the female genotype data. Thirty-one pairs of markers showed significant $p$ values ($p < 0.05$) in at least one population (data not shown), with two pairs that were significant in three populations (DXS101–DXS7424 and DXS7423–DXS10147). An indication of linkage disequilibrium for DXS101–DXS7424 has been observed previously in German [62] and Brazilian [63] populations, and a recombination study has demonstrated the association between DXS7423 and DXS10147 [49]. This latter pair alone resulted in a $p$ value that remained significant in two populations after the Bonferroni correction ($p < 0.003$) was applied. Ten pairs of markers confirmed suspected associations based upon the classification of markers into the four linkage groups. The majority of the pairs with significant $p$ values were not located within the same linkage group or in close proximity on the X chromosome, and do not remain significant after the Bonferroni correction. It is therefore possible that the $p$ values for these pairings do not indicate a true

linkage, but rather population sampling effects or substructure, and further investigation is necessary.

For reference, haplotypes based upon the proposed linkage groups are presented in Supplementary Tables 3–5 (except for linkage group 3, which is represented by HPRTB alone in these multiplexes). While these data may help to provide an initial understanding of the power of X chromosomal STR markers treated as both independent markers and linked haplotypes, the population size is too small to provide an accurate depiction of the frequency of each observed haplotype. Additionally, as evidenced by the inconclusive linkage disequilibrium results observed here and elsewhere, further studies should be performed to more thoroughly assess the linkage between markers and better define the proposed linkage groups.

## 4. Conclusions

The two proposed multiplexes are exceptional in the world of X STRs because they represent one of the first efforts at combining maximum discrimination with maximum utility on real-world forensic samples. There exists but one other publication describing the use of mini-X chromosomal STRs [13]. Indeed, seven of the sixteen mini-X STR primer sets used in these mini-plexes were adopted from this work. However, the two multiplexes in the current study contain almost twice the number of markers as well as a marker from each linkage group, unlike the two quadruplexes that lack a representative from linkage group 3. Though the present study lacks an organized comparison of mini-STR results to those with larger-amplicon multiplexes, the two mini X-plexes show promise in this area. To this end, future work will include concordance studies between the reduced-amplicon multiplexes proposed here and the overlapping markers with larger amplicons in the Argus X12 kit. In conclusion, this study demonstrates the potential of these multiplexes for routine forensic casework and inquiries into human evolutionary history.

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## Appendix A. Supplementary data

Supplementary data associated with this article can be found, in the online version, at doi:10.1016/j.fsigen.2010.08.019.

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For supplementary materials, see Chapter 4.

**C2.** Toni M. Diegoli, Lejla Kovacevic, Naris Pojskic, Michael D. Coble, Damir Marjanovic, Population study of fourteen X chromosomal short tandem repeat loci in a population from Bosnia and Herzegovina, Forensic Sci. Int. Genet. 5 (2011) 350-1.

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Letter to the Editor

## Population study of fourteen X chromosomal short tandem repeat loci in a population from Bosnia and Herzegovina

*Dear Editor,*

X chromosomal short tandem repeat (STR) loci can be used in combination with autosomal STRs to solve cases of complex kinship. In order to properly apply STR results to forensic cases, population databases must be established to which comparisons can be made and the rarity of profiles determined. Here, a population from Bosnia and Herzegovina was studied to determine the appropriate forensic efficiency parameters with 14 widely used X chromosomal STR loci.

Profiles from a total of 154 (68 female and 86 male) unrelated individuals living in Bosnia and Herzegovina were analyzed as part of this study. After obtaining informed consent, either blood stains or buccal swabs were collected and extracted using the QIAmp DNA Micro Kit (Qiagen GmbH, Hilden, Germany). Multiplex PCR amplification and electrophoretic detection was performed using the original multiplexes as described in [1], and the recommendations of the International Society for Forensic Genetics (ISFG) concerning STR nomenclature were followed [2,3] (see [1] for detailed discussion). Per the recommendations of Szibor et al. [4], allele designations were achieved through comparison with three control DNAs: 9948 (Applied Biosystems, Foster City, CA, USA and Promega Corporation, Madison, WI, USA), 9947a (Applied Biosystems), and K562 (Promega Corporation). This paper follows the guidelines for publication of population data requested by the journal [5], and the authors understand and accept the conditions requested within.

Allele frequencies and forensic efficiency parameters calculated for the 14 X chromosomal STRs examined are presented in Supplementary Table 1. The chi-square test for independence was used to examine the distribution of allele frequencies in the male and female donors. The resultant $p$ values showed no significant differences ($p > 0.05$) and frequency data was pooled at each locus. Both the software PowerMarker [6] and the Forensic ChrX Research website version 2.0 [7] were used to calculate the appropriate forensic efficiency statistics, and analysis of linkage disequilibrium was performed in Arlequin v3.1 [8]. In total, 110 different alleles were observed across 14 loci, with 4–16 alleles per locus. The highest observed heterozygosity was noted at locus DXS101 (0.8676), which also had the most alleles (16), while the lowest was observed at DXS6789 (0.5882). All markers possessed high forensic efficiency values with the studied population sample, supporting the utility of the multiplexes for forensic purposes.

Two markers (DXS7424 and DXS6789) showed a departure from Hardy–Weinberg equilibrium, indicated by a $p$ value for the exact test that is less than 0.05 (bold values in Supplementary Table 1). The observed deviations could be due to the small population size used to evaluate Hardy–Weinberg equilibrium (68 females). After applying the Bonferroni correction, however, none of the values remained significant.

Analysis of pairwise linkage disequilibrium revealed marginally significant results ($p < 0.05$) in the studied population for seven pairs of markers: GATA172D05–GATA31E08, DXS7132–DXS7423, DXS9902–HPRTB, DXS7130–DXS6803, DXS6789–GATA172D05, DXS8378–GATA172D05, and DXS7424–DXS7130. Only one pair showed a significant p value less than 0.01 (DXS8378–GATA165B12). While it is possible that the p values for these pairing indicate a true linkage, the population tested was relatively small (68 females) and analyses may be skewed by non-random sampling or substructure. For reference, male haplotype data according to the proposed linkage groups [9,10] are presented in Supplementary Tables 2–4. A count of the number of observed haplotypes was performed for groups 1, 2, and 4 (group 3 is represented by HPRTB alone in these multiplexes). There were 20 unique haplotypes for linkage group 1 (DXS8378 and DXS9902) and 24 for linkage group 4 (GATA31E08 and DXS7423) while every haplotype was unique across linkage group 2. The most common haplotype (DXS8378–10, DXS9902–10) was observed 14 times.

The data from this population was compared with six other populations representing various regions of Europe: Hungary [11] and Latvia [12] (eastern Europe); Italy [13,14] and Germany [15–17] (central Europe); and Portugal [18,19] and Spain [19–21] (western Europe). The allele frequency distributions of overlapping loci were assessed using the chi-square test and significant differences ($p < 0.05$) were observed for several loci in several populations: DXS9902 and DXS7130 in Germans, HPRTB and DXS7133 in Latvians, and DXS8378 in Northern Portuguese (data not shown). Because of the relatively small sample size studied here, however, further investigation would be necessary to determine if these populations truly differ at these loci. In general, the allelic distribution of these particular X chromosomal STRs in the population from Bosnia and Herzogovina is similar to that in other populations across Europe.

This work represents the first study of a population from Bosnia and Herzegovina using 14 X chromosomal STRs.

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### Appendix A. Supplementary data

Supplementary data associated with this article can be found, in the online version, at doi:10.1016/j.fsigen.2010.01.007.

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For supplementary materials, see Chapter 4.

*Non-Peer-Reviewed Publications*

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# Developmental validation of 15 X chromosomal short tandem repeat markers

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ABSTRACT

The use of X chromosomal short tandem repeat (STR) markers has been greatly increasing in the forensic setting. Using guidelines set forth previously for the validation of autosomal and Y STRs, aspects of the feasibility of routine X chromosomal STR use were evaluated. Two mini-X chromosomal STR multiplexes capable of amplifying 15 total markers were developed and utilized to determine allele nomenclature, allele/genotype frequencies, mutation rates, and linkage between markers. Additionally, a concordance study between these multiplexes and a commercially available kit was performed. Here, the authors present an overview of this extensive developmental validation study.



## 1. Introduction

The use of X chromosomal short tandem repeat (STR) markers has been greatly increasing in the forensic setting. The marker system offers the potential to provide information in addition to what is obtained from autosomal STR systems currently used at crime laboratories and in the courtroom. In certain scenarios, markers on the X chromosome may be the only means of obtaining this information. In-depth characterization of the marker system is the first step in maximizing the power of this additional tool in the forensic arsenal.

Guidelines used to validate a marker system such as autosomal or Y STRs for forensic use have been described previously [1,2]. These guidelines can, of course, be applied to any forensic marker system as technology changes and the scope of the questions asked by forensic scientists broadens. The aspects that must be addressed are as follows: (1) selection of suitable markers with the intended purpose in mind; (2) development of a multiplex assay that is robust and sensitive enough to accommodate targeted sample types; (3) generation of high-quality population databases large enough to provide sufficient statistical vigor to the conclusions made based on the genetic information; (4) assessment of the requirements for statistical interpretation of a match between two genetic profiles; and (5) data compatibility between laboratories.

## 2. Methods, results and discussion

### 2.1. Marker selection

A review of literature resulted in a list of potential markers, capitalizing on the collective knowledge of these established markers and their relevant characteristics, simplifying the process. Utility was assessed according to the following criteria: (1) potential for small amplicon size; (2) heterozygosity in published materials; and (3) distribution between the four proposed linkage groups across the X chromosome [3]. The list of candidate markers was further limited by selecting those markers that could be electrophoretically separated in a four-dye multiplex.

Fifteen markers were identified for inclusion in two multiplexes: DXS6789, DXS7130, DXS9902, GATA165B12, DXS101, GATA31E08, DXS7424, DXS6795, GATA172D05, DXS10147, DXS8378, DXS7132, DXS6803, HPRTB, and DXS7423 [4].

### 2.2. Assay development

In order to balance number of markers with size of amplicons, two multiplexes were designed with all amplicons under 200 bp in

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order to increase utility with potentially degraded extracts [5]. This process is detailed in [4].

### 2.3. Population databases

In order to study the genetic parameters of the chosen set of X STRs in relevant populations, unrelated, anonymous individuals from four major U.S. populations (African Americans, U.S. Asians, U.S. Caucasians, and U.S. Hispanics) were investigated [4,6]. In addition, profiles from a total of 154 unrelated individuals were analyzed as part of the first study of X STRs in a population from Bosnia and Herzegovina [7].

In general for both populations, heterozygosity values for the selected markers were high and the populations were not statistically different from similar published populations, reinforcing the utility of the chosen set of markers.

### 2.4. Match criteria

Both the rate of mutation and the linkage between the 15 markers were investigated. The overall mutation rate was determined using 958 (275 African American, 366 U.S. Caucasian, and 317 U.S. Hispanic) families (2022 individuals). In 20,625 meioses, 18 mutations were observed, resulting in a mutation rate of $8.73 \times 10^{-4}$ (manuscript undergoing revisions).

To study linkage, 158 families with several generations and/or multiple offspring were typed and the recombination rate was assessed used two different methods (direct calculation using observed instances of recombination and computer-based analyses as described in [8]). The values obtained using the different methods were generally similar, indicating a robust computation.

### 2.5. Data concordance

In addition to the routine allele sequencing necessary during the development of the multiplex assays, sequencing of a subset of samples at each marker was accomplished to begin to define the variation present at each marker as well as to verify consistency of the repeat structure with that of published data obtained using larger primer sets. Results revealed variation from published studies in repeat number or structure for four markers (DXS6803, GATA31E08, GATA172D05, and HPRTB).

A concordance study between the only currently available commercial X STR kit, the QIAGEN® Investigator Argus X-12, and the four overlapping markers also present the studied set of 15 markers was completed. Nearly all of the samples typed were concordant, with only 3 instances of discordance due to a null allele (99.92% concordance).

## 3. Conclusions

This extensive developmental validation study investigated each aspect of the 15-marker system that would require consideration before implementation by a forensic laboratory. The selected markers were found to be discriminating and the optimized assay robust, while the mutation rate was determined with high accuracy and the extent of linkage between the 15 markers was evaluated. Comparability was established for four overlapping markers while the utility of the only commercially available X STR kit was evaluated with U.S. populations for the first time. The combination of the results obtained as part of this study form the foundation upon which the routine use of X STRs with U.S. populations may be built.

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## Conflict of interest

None.

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# A gonosomal marker multiplex to aid in mixture interpretation

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ABSTRACT

Mixture interpretation is an important part of the forensic scientist's role in evaluating evidence from a crime scene, where mixed specimens can be common. To this end, a mixture multiplex has been created that combines markers from both the X and the Y chromosomes in an attempt to aid in the interpretation of such mixtures, providing clues as to the sex and number of contributors. By maximizing the information gained, the direction of further testing could potentially be influenced and optimized.



## 1. Introduction

Mixture interpretation is an important part of the forensic scientist's role in evaluating evidence from a crime scene. However, routine analysis of mixtures continues to be a challenge, and varying solutions have been proposed and adopted [1–3]. One approach that has thus far only been partially explored is the use of markers on the sex chromosomes. Several studies reported the ability of Y chromosomal markers to detect the male component in male–female mixtures from sexual assault cases [4,5]. Similarly, it has been suggested that X chromosomal markers may help to expose a female profile in male background, such as vaginal cells on a penis or female cells from male fingernail scrapings [6]. Due to their unique inheritance patterns, gonosomal markers hold the potential to supplement traditional mixture testing in certain specific situations.

To this end, a mixture multiplex, termed "MIXplex," has been created that combines markers from both the X and Y chromosomes in an attempt to aid the interpretation of mixed samples, providing clues as to the sex and number of contributors. The benefits and limitations of using gonosomal STRs in the evaluation of mixed evidence are uncovered and discussed in the context of current and future efforts.

## 2. Materials and methods

A combination of short tandem repeat (STR) markers located on both sex chromosomes was targeted, including STRs within the XY homologous region, termed XY markers. A review of the literature resulted in a list of potential markers, thus exploiting the collective knowledge of these established markers and their relevant characteristics, simplifying the process. Potential utility was assessed according to the following criteria: (a) potential for small amplicon size; (b) large allele range with high degree of polymorphism; and (c) established use within the forensic community.

Extracts utilized in this study were six (3 male, 3 female) generally commercially available control DNAs for which the profiles at each of the chosen markers were known. PCR amplification, electrophoresis, and detection occurred in a manner similar to the protocol described in [7]. Using these same six controls, 63 total mixtures of various sex and number of contributors were created and a subset was amplified using the MIXplex. Blind analyses focused on determination of the minimum number and sex of contributors.

## 3. Results and discussion

After consideration of candidate gonosomal markers, several suboptimal primer pairs were excluded and the final multiplex consisted of three X STRs (DXS6795, DXS6789, and GATA31E08), two Y STRs (DYS393 and DYS438), one XY STR (DXYS267), and a portion of the sex determining region of the Y chromosome, SRY. Primer mix concentrations were adjusted empirically to balance peak heights within each multiplex, and tested on a panel of

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known control samples to ensure consistent quality and correct genotypes could be obtained. Using a calling threshold of 100 RFU, sensitivity testing with single-source samples revealed that full profiles could be obtained with as little as 200 pg of input DNA and heterozygous peak height ratios generally remained above 60%. With duplicate or triplicate amplification of six two-person mixtures (four female–male, one male–male, and one female–female) at varying mixture ratios, complete profiles were reliably obtained for mixtures where the minor component was 20% or greater in most cases, which coincided well with the single-source sensitivity results. Like the sensitivity results, the mixture testing also revealed that near-complete or complete profiles could be obtained for most mixtures with the minor component at only 10%, or 100 pg input.

The design and potential of the MIXplex combined several key elements of mixture interpretation. Generally, reporting of mixed profiles centers on estimating the minimum number of contributors as well as attempting (mainly in two-person mixtures) to assign a sex to each. The MIXplex correctly identified the sex and minimum number of contributors in all cases of artificial and theoretical mixtures tested in this study, and correctly assigned the actual number and sex of contributors 62% of the time. Currently, with autosomal STRs, sex can only be reliably assigned when both contributors are of the same gender, or the male contributor is the minor component of a male–female mixture [8]. The MIXplex offers an alternative to using additional Y STR testing to infer and confirm these characteristics. Corroboration of the suspected number and/or sex of contributors through this assay could direct future analyses, potentially saving time and money. Additionally, the MIXplex can clarify situations where the male allele at the amelogenin locus is not amplified due to a deletion on the Y chromosome [9,10]. Moreover, profile subtraction is simplified in an assay where only four of seven markers are found on a female's chromosomes. Even when a male and a female contributor share alleles, there are an additional three markers at which only male alleles would be present.

While it is clear from this initial study that the MIXplex cannot solve all of the questions surrounding the interpretation of mixed profiles, there are benefits to its use in certain situations that justify continued study. Additional characterization of assay parameters such as the reporting threshold and stutter ratios would be helpful to increase confidence in allele calls. Analysis of additional mixtures, both theoretical and actual, could further illustrate both the strengths and the limitations of this combination of markers as well as suggest additional configurations that might aid interpretation even further. Casework mixtures should eventually be evaluated with a final assay in order to assess its performance and value in real-world settings.

Two key elements of mixture analysis rest in the initial determination of the sex and minimum number of contributors [8], which the MIXplex helps to address. However, these same authors recognize that a standardized mixture interpretation protocol that will be appropriate for every mixed profile an analyst encounters is not feasible. Where the MIXplex, or an improved version of it, fits into the overall forensic mixture interpretation scheme remains to be uncovered.

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## Conflict of interest

None.

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