

Appendix

Some of the large tables can be found in spreadsheet format online, as supplementary material to the accompanying published manuscripts. Refer to the relevant chapters for citations.

Appendix Table 1. List of 91 lead genome-wide significant independent SNPs for disc size from UK Biobank

SNP	CHR	BP	Nearest Gene	Novel	EA	NEA	FREQ	BETA (UKBB)	P (UKBB)	Z score (IGGC + EPIC)	P (IGGC + EPIC)
rs12024620	1	3049362	<i>PRDM16</i>	0	C	T	0.94	-0.03	2.7E-16	4.71	2.4E-06
rs787540	1	68051137	<i>GADD45A</i>	0	A	G	0.66	-0.01	1.9E-11	-5.56	2.7E-08
rs12064002	1	81345620	<i>LPHN2</i>	1	C	T	0.75	-0.01	6.7E-09	2.38	0.02
rs1192415	1	92077097	<i>CDC7-TGFBR3</i>	0	G	A	0.19	0.06	1.8E-107	-17.11	1.3E-65
rs3754442	1	110611886	<i>ALX3</i>	1	C	T	0.41	-0.01	2.5E-10	1.90	0.06
rs12136690	1	116208944	<i>VANGL1</i>	1	C	T	0.76	-0.02	3.1E-24	5.40	6.7E-08
rs11264319	1	155083942	<i>EFNA1</i>	1	T	C	0.49	-0.01	3.2E-09	-3.09	2.0E-03
rs9332665	1	169486767	<i>F5</i>	0	T	G	0.67	0.01	4.4E-08	3.35	8.0E-04
rs742127	1	169564167	<i>SELP</i>	0	G	C	0.69	-0.01	4.9E-09	6.67	2.5E-11
rs56412756	1	201605477	<i>NAV1</i>	1	C	T	0.92	0.02	2.0E-08	-3.96	7.6E-05
rs114537871	1	227560794	<i>CDC42BPA</i>	0	G	A	0.92	0.05	1.1E-35	-7.45	9.4E-14
rs1100941	2	19410997	<i>MIR4757</i>	1	T	C	0.01	-0.06	4.1E-11	-1.81	0.07
rs116072427	2	33357096	<i>LTBP1</i>	1	G	C	0.93	0.02	1.1E-08	-3.34	8.3E-04
rs9967780	2	56234942	<i>MIR216B</i>	1	G	T	0.78	-0.01	3.0E-09	4.73	2.3E-06
rs6546560	2	70173690	<i>MXD1</i>	1	A	G	0.57	-0.01	1.5E-08	-0.91	0.36
rs4832012	2	86000500	<i>ATOH8</i>	1	G	C	0.49	-0.01	1.4E-11	3.69	2.3E-04
rs3748916	2	113984033	<i>PAX8</i>	1	G	A	0.53	0.01	5.5E-09	-2.50	0.01
rs1365902	2	145470699	<i>TEX41</i>	1	T	C	0.33	-0.01	8.7E-12	-4.91	9.1E-07
rs3914468	2	170157400	<i>LRP2</i>	1	A	G	0.70	-0.01	2.0E-10	-3.58	3.4E-04

rs7572800	2	180198241	SESTD 1	1	C	G	0.43	0.01	1.4E-08	2.18	0.03
rs1550094	2	233385396	PRSS56	1	G	A	0.31	-0.02	7.3E-13	1.98	0.05
rs4417830	3	25034135	RARB	0	A	G	0.70	-0.01	4.1E-11	-4.88	1.0E-06
rs6764521	3	25184389	RARB	0	A	G	0.34	-0.02	3.1E-12	-2.52	0.01
rs6787363	3	25388569	RARB	0	G	A	0.62	-0.02	3.0E-26	4.80	1.6E-06
rs13070614	3	25414588	RARB	0	C	G	0.78	0.02	1.5E-20	3.28	1.0E-03
rs114263587	3	57081641	ARHGE F3	1	T	C	0.97	0.03	3.5E-08	-1.39	0.17
rs71296770	3	62371837	CADPS	1	A	G	0.90	-0.02	5.4E-11	-1.76	0.08
rs77877421	3	71182447	FOXP1	1	A	T	0.94	-0.03	2.6E-09	-3.47	5.1E-04
rs793472	3	99546110	CMSS1; FILIP1L	1	A	G	0.22	-0.01	1.9E-08	-2.36	0.02
rs17398137	3	100625703	ABI3BP	0	G	A	0.82	0.02	6.4E-19	-6.02	1.7E-09
rs11133307	4	55018996	GSX2	1	C	T	0.65	-0.01	1.5E-09	2.57	0.01
rs74764079	4	81952637	BMP3	1	T	A	0.97	0.06	1.0E-19	-1.22	0.22
rs2654743	4	112361474	C4orf32	1	G	A	0.64	0.02	5.1E-14	-1.81	0.07
rs72759609	5	31952051	PDZD2	1	T	C	0.90	0.03	3.1E-17	4.02	5.9E-05
rs536376209	5	82740256	VCAN	1	A	G	0.60	0.01	1.2E-09	NA	NA
rs58531939	5	87823968	LINC00 461	1	T	C	0.91	-0.03	6.3E-16	-4.55	5.3E-06
rs11242236	5	134586980	C5orf66	1	G	A	0.55	-0.01	5.0E-09	0.11	0.91
rs2092524	6	39529692	KIF6	1	G	A	0.66	-0.01	8.5E-11	4.18	3.0E-05
rs7744813	6	73643289	KCNQ5	1	C	A	0.41	0.02	2.3E-19	-1.18	0.24
rs12661045	6	122682795	HSF2	1	C	T	0.70	0.02	6.1E-14	-3.46	5.5E-04
rs2152876	6	126761228	CENPW	1	G	A	0.54	-0.02	1.5E-18	5.36	8.2E-08
rs9401928	6	127298394	RSPO3	1	G	A	0.55	-0.02	2.7E-14	4.44	9.0E-06
rs80218807	8	1884839	ARHGE F10	1	C	T	0.89	-0.02	6.0E-09	0.47	0.64
rs1905014	8	13407297	C8orf48	1	T	C	0.57	-0.02	5.3E-13	-2.72	6.5E-03
rs3857971	8	61969579	LOC100 130298	1	G	A	0.70	0.02	2.2E-14	-2.75	5.9E-03

rs17734073	8	72598450	MSC	1	A	G	0.87	-0.02	5.6E-10	-2.75	6.0E-03
rs6999835	8	78948855	PKIA	1	T	C	0.63	0.01	5.1E-09	3.74	1.8E-04
rs7844015	8	88763923	DCAF4L2	0	A	G	0.48	0.01	2.4E-08	5.60	2.1E-08
rs10512176	9	89252706	ZCCHC6	1	T	C	0.72	-0.01	3.5E-11	-4.07	4.8E-05
rs73654567	9	98309513	PTCH1	1	A	G	0.89	-0.02	5.0E-09	-0.90	0.37
rs13283933	9	134584308	RAPGEF1	1	C	T	0.71	0.02	1.0E-11	-2.82	4.7E-03
rs10764494	10	25058144	ARHGA21	1	C	A	0.32	-0.01	4.7E-10	3.84	1.2E-04
rs750307744	10	69944990	MYPN	0	G	A	0.96	0.06	1.4E-19	NA	NA
rs7916697	10	69991853	ATOH7	0	A	G	0.24	-0.07	4.0E-169	-22.19	4.0E-109
rs565754424	10	70041770	ATOH7	0	T	A	0.97	0.10	6.1E-37	NA	NA
rs3835100	10	70051810	PBLD	0	A	G	0.06	-0.08	3.5E-51	-4.99	6.0E-07
rs10999459	10	72422431	ADAMTS14	1	G	A	0.84	0.01	4.0E-08	0.98	0.33
rs2149108	10	94866290	CYP26A1	1	C	T	0.60	0.01	1.5E-10	-1.08	0.28
rs4408249	10	104374653	SUFU	1	T	A	0.57	0.01	3.0E-11	-1.62	0.10
rs2447670	11	31330689	DCDC1	1	T	G	0.40	-0.01	2.2E-11	-3.25	1.2E-03
rs12421026	11	31809322	PAX6	0	C	T	0.50	-0.02	1.3E-17	4.94	7.8E-07
rs73048943	12	3383989	TSPAN9	1	C	A	0.94	-0.03	1.6E-12	3.15	1.6E-03
rs76567987	12	31037655	TSPAN11	1	A	G	0.84	0.02	8.4E-16	4.01	6.1E-05
rs711111	12	77995058	NAV3	1	A	G	0.66	-0.01	3.1E-08	-2.66	7.9E-03
rs61952220	12	83959234	TMTC2	0	C	A	0.45	-0.02	4.7E-20	NA	NA
rs9534439	13	47192049	LRCH1	1	T	C	0.19	0.02	5.1E-14	4.36	1.3E-05
rs2761882	14	54420309	BMP4	1	C	T	0.50	0.01	1.7E-09	-1.69	0.09
rs61985972	14	59550263	DAAM1	1	A	G	0.94	0.03	5.2E-11	5.02	5.2E-07
rs12147505	14	60660734	DHRS7	1	T	C	0.99	0.10	9.5E-26	2.20	0.03
rs112115087	14	60807865	PPM1A	0	T	G	0.61	0.02	1.3E-17	NA	NA
rs76320564	14	65115953	PLEKHG3	1	T	C	0.83	-0.02	3.1E-13	-2.24	0.02

rs12436074	14	85727904	SNORD3P3	1	G	A	0.44	0.02	6.9E-20	-2.89	3.8E-03
rs11627567	14	103851775	MARK3	1	C	T	0.79	-0.01	9.5E-09	1.41	0.16
rs1800407	15	28230318	OCA2	1	C	T	0.91	0.02	2.6E-08	-2.23	0.03
rs678510	15	35003309	GJD2	1	T	C	0.67	0.01	3.9E-09	-0.06	0.95
rs59484405	15	84476534	ADAMTSL3	1	A	T	0.74	-0.01	2.3E-08	-1.79	0.07
rs8034595	15	96719229	NR2F2-AS1	0	A	C	0.31	-0.02	1.9E-12	-3.88	1.1E-04
rs17648524	16	7459683	RBFOX1	1	G	C	0.63	0.01	3.3E-10	-3.28	1.0E-03
rs7196852	16	51462823	SALL1	0	G	A	0.30	0.02	1.9E-21	-7.38	1.5E-13
rs28514893	16	51573119	SALL1	0	C	T	0.79	-0.02	3.6E-14	4.79	1.6E-06
rs11655511	17	59287269	BCAS3	1	C	T	0.23	0.03	2.8E-30	-2.65	8.0E-03
rs72839495	17	61044550	TANC2	1	A	T	0.82	0.02	1.2E-08	2.95	3.2E-03
rs7405453	17	79615572	TSPAN10	1	A	G	0.36	-0.02	3.9E-23	-3.02	2.5E-03
rs11564401	18	25666690	CDH2	1	G	A	0.72	0.02	5.0E-12	-3.31	9.4E-04
rs10164395	19	32035891	THEG5	1	C	T	0.67	0.01	1.9E-08	-2.90	3.7E-03
rs743184	22	29102967	CHEK2	0	G	A	0.36	-0.02	4.8E-25	6.61	4.0E-11
rs7286335	22	37934531	CARD10	0	C	T	0.74	-0.02	2.5E-17	5.98	2.2E-09
rs9330797	22	46300967	WNT7B	1	A	G	0.58	-0.02	3.1E-14	-1.61	0.11
rs9330813	22	46364161	WNT7B	1	G	A	0.69	0.01	1.5E-09	-1.30	0.19
rs554433	23	68058027	EFNB1	1	C	T	0.40	-0.02	2.2E-31	NA	NA
rs113485657	23	136508373	ZIC3	1	C	A	0.94	-0.02	8.0E-09	NA	NA

Appendix Table 2. List of 115 lead genome-wide significant independent SNPs for disc size from UK Biobank and IGGC meta-analysis

SNP	CHR	BP	Nearest Gene	Novel	EA	NEA	FREQ	BETA (META)	P (META)	Direction	BETA (MTAG)	P (MTAG)
rs12024620	1	3049362	PRDM16	0	T	C	0.06	0.04	7.40E-20	++	0.04	5.70E-20
rs599892	1	39922884	MACF1	1	C	G	0.7	0.01	1.10E-08	++	0.01	1.50E-08

rs787540	1	68051137	GADD45A	0	A	G	0.66	-0.02	7.70E-16	--	-0.02	1.40E-15
rs6673575	1	81349395	LPHN2	1	A	G	0.32	0.01	8.80E-11	++	0.01	1.50E-10
rs4658101	1	92077409	CDC7 - TGFB3	0	A	G	0.19	0.06	2.80E-160	++	0.07	2.10E-156
rs3754442	1	110611886	ALX3	1	T	C	0.59	0.01	1.20E-09	++	0.01	1.50E-09
rs4839470	1	116209290	VANGL1	1	T	C	0.24	-0.02	3.50E-23	--	-0.02	8.40E-23
rs12137699	1	116209762	VANGL1	1	T	C	0.24	0.02	4.30E-27	++	0.03	9.40E-27
rs57742792	1	155071588	EFNA3	1	A	T	0.51	0.01	3.50E-11	++	0.01	4.80E-11
rs6030	1	169498975	F5	0	T	C	0.67	0.01	1.80E-10	++	0.01	3.00E-10
rs742127	1	169564167	SELP	0	C	G	0.31	0.02	3.60E-15	++	0.02	8.30E-15
rs11584075	1	201598915	NAV1	1	A	C	0.92	0.02	1.60E-10	++	0.02	6.30E-11
rs11808696	1	227563680	CDC42BPA	0	T	C	0.08	-0.05	5.80E-48	--	-0.05	3.40E-47
rs340762	2	16484005	FAM49A	1	T	G	0.45	0.01	9.40E-09	++	0.01	1.30E-08
rs72784719	2	19344098	MIR4757	1	A	C	0.99	0.06	1.00E-11	++	0.06	1.40E-11
rs13417287	2	33219147	LTBP1	1	T	C	0.77	-0.01	5.70E-10	--	-0.01	1.10E-09
rs116072427	2	33357096	LTBP1	1	C	G	0.07	-0.02	4.50E-11	--	-0.03	4.30E-11
rs163524	2	45157553	SIX3	1	A	C	0.18	-0.01	1.00E-08	--	-0.01	1.30E-08
rs9967780	2	56234942	MIR216B	1	T	G	0.22	0.02	1.80E-13	++	0.02	2.90E-13
rs4832012	2	86000500	ATOH8	1	C	G	0.51	0.01	8.90E-13	++	0.01	1.30E-12
rs11684168	2	88694167	FOXI3	1	T	C	0.83	0.01	2.90E-08	++	0.01	3.90E-08
rs12619508	2	113982040	PAX8	1	A	C	0.47	-0.01	7.70E-10	--	-0.01	1.10E-09
rs1901440	2	134437959	NCKAP5	1	A	C	0.66	-0.01	3.40E-08	--	-0.01	4.20E-08
rs12471698	2	145475412	TEX41	1	T	C	0.33	-0.01	5.60E-15	--	-0.02	1.00E-14
rs9287914	2	170155247	LRP2	1	A	T	0.31	0.01	8.40E-13	++	0.02	1.00E-12
rs13022913	2	180175927	SESTD1	1	T	C	0.57	-0.01	5.50E-09	--	-0.01	6.90E-09
rs10084346	2	218306736	DIRC3	0	T	C	0.16	0.01	2.20E-08	++	0.02	3.10E-08
rs934237	2	218467037	DIRC3	0	A	C	0.79	-0.02	1.70E-11	--	-0.02	3.20E-11
rs1550094	2	233385396	PRSS56	1	A	G	0.69	0.01	5.20E-13	++	0.02	1.10E-12
rs7620608	3	20095378	KAT2B	1	T	C	0.61	-0.01	1.40E-08	--	-0.01	1.80E-08
rs7615960	3	21730246	ZNF385D	1	A	T	0.93	-0.02	1.50E-08	--	-0.02	2.10E-08
rs4858678	3	25047446	RARB	0	A	G	0.72	-0.02	1.10E-15	--	-0.02	3.00E-15
rs6787363	3	25388569	RARB	0	A	G	0.38	0.02	2.30E-31	++	0.02	7.40E-31
rs13070614	3	25414588	RARB	0	C	G	0.78	0.02	2.00E-22	++	0.02	3.90E-22
rs71296770	3	62371837	CADPS	1	A	G	0.9	-0.02	8.20E-11	--	-0.02	6.10E-10
rs77877421	3	71182447	FOXP1	1	A	T	0.94	-0.03	1.30E-11	--	-0.03	1.40E-11
rs7610486	3	99732564	FILIP1L	1	A	G	0.77	0.01	1.60E-08	++	0.01	1.90E-08
rs17398137	3	100625703	ABI3BP	0	A	G	0.18	-0.03	4.00E-26	--	-0.03	1.40E-25
rs2220051	3	156764263	LEKR1	1	T	C	0.34	-0.01	2.00E-08	--	-0.01	2.90E-08
rs9852867	3	189969372	CLDN1	1	T	G	0.41	-0.01	3.20E-08	--	-0.01	4.00E-08
rs28603236	4	41561962	LIMCH1	1	A	G	0.85	0.01	4.30E-08	++	0.02	5.90E-08
rs2033054	4	55018617	GSX2	1	T	C	0.65	-0.01	1.30E-10	--	-0.01	1.80E-10
rs74764079	4	81952637	BMP3	1	A	T	0.03	-0.05	1.70E-19	--	-0.05	3.70E-18
rs906568	4	112402558	C4orf32	1	T	C	0.36	-0.01	2.00E-13	--	-0.02	3.00E-13
rs72759609	5	31952051	PDZD2	1	T	C	0.9	0.03	7.50E-19	++	0.03	2.00E-18
rs251526	5	52586585	FST	1	A	G	0.92	-0.02	3.90E-10	--	-0.02	5.70E-10
rs30371	5	55742832	ANKRD55	1	T	C	0.63	-0.01	3.40E-08	--	-0.01	4.30E-08
rs7717697	5	82744604	VCAN	1	T	C	0.59	0.01	2.40E-12	++	0.01	3.90E-12
rs73173591	5	87830536	LINC00461	1	A	T	0.09	0.03	2.60E-19	++	0.03	5.30E-19

rs6860726	5	126259178	MARCH3	1	C	G	0.49	-0.01	1.30E-08	--	-0.01	1.80E-08
rs62391624	6	1923520	GMDS	1	T	C	0.14	-0.02	8.70E-10	--	-0.02	1.30E-09
rs2092524	6	39529692	KIF6	1	A	G	0.34	0.01	1.30E-13	++	0.02	2.40E-13
rs7744813	6	73643289	KCNQ5	1	A	C	0.59	-0.02	1.00E-15	++	-0.02	2.10E-15
rs34607745	6	122675335	HSF2	1	T	G	0.3	-0.02	3.40E-16	--	-0.02	5.80E-16
rs112166936	6	126733140	CENPW	1	A	C	0.55	-0.02	1.40E-22	--	-0.02	2.80E-22
rs1930941	6	127316843	RSPO3	1	A	C	0.45	0.02	1.10E-15	++	0.02	1.90E-15
rs80218807	8	1884839	ARHGEF10	1	T	C	0.11	0.02	3.10E-08	++	0.02	9.40E-08
rs1905014	8	13407297	C8orf48	1	T	C	0.57	-0.01	4.40E-13	--	-0.01	6.50E-13
rs10957177	8	61911070	LOC100130298	1	A	G	0.75	0.01	1.30E-10	++	0.01	2.50E-10
rs3857971	8	61969579	LOC100130298	1	A	G	0.3	-0.02	1.90E-16	--	-0.02	3.40E-16
rs17734073	8	72598450	MSC	1	A	G	0.87	-0.02	2.70E-10	--	-0.02	3.30E-10
rs6999835	8	78948855	PKIA	1	T	C	0.63	0.01	2.40E-10	++	0.01	3.20E-10
rs12547416	8	88761223	DCAF4L2	0	T	C	0.51	-0.01	5.20E-14	--	-0.01	1.20E-13
rs13264644	8	143753546	PSCA	1	A	G	0.57	-0.01	4.50E-08	--	-0.01	5.60E-08
rs10512176	9	89252706	ZCCHC6	1	T	C	0.72	-0.02	1.70E-14	--	-0.02	2.60E-14
rs16909978	9	98307990	PTCH1	1	A	G	0.11	0.02	9.10E-10	++	0.02	1.40E-09
rs13283933	9	134584308	RAPGEF1	1	T	C	0.29	-0.02	2.20E-13	--	-0.02	3.40E-13
rs10764494	10	25058144	ARHGAP21	1	A	C	0.68	0.01	8.50E-13	++	0.02	1.50E-12
rs7916410	10	69995667	ATOH7	0	T	C	0.24	-0.07	3.30E-246	--	-0.08	2.10E-240
rs9988687	10	70024528	PBLD	0	A	G	0.77	0.07	3.40E-209	++	0.07	6.10E-206
rs3835100	10	70051810	PBLD	0	A	G	0.06	-0.08	1.20E-56	--	-0.07	5.00E-55
rs7096311	10	71610049	COL13A1	1	A	C	0.19	-0.01	2.70E-09	--	-0.02	3.80E-09
rs10823610	10	72506158	ADAMTS14	1	A	C	0.56	0.01	9.60E-09	++	0.01	1.20E-08
rs2149108	10	94866290	CYP26A1	1	T	C	0.4	-0.01	4.00E-10	--	-0.01	3.50E-10
rs4285804	10	104386309	SUFU	1	A	T	0.56	0.01	1.20E-11	++	0.01	1.70E-11
rs11602582	11	31285284	DCDC1	1	A	G	0.05	-0.04	4.10E-15	--	-0.04	2.50E-15
rs7482884	11	31683485	ELP4	0	A	T	0.23	0.02	4.20E-25	++	0.02	1.60E-24
rs78977588	11	68968149	MYEOV	1	A	C	0.12	0.02	6.30E-09	++	0.02	7.40E-09
rs1126809	11	89017961	TYR	1	A	G	0.3	-0.01	8.00E-09	--	-0.01	1.10E-08
rs74891390	12	3373823	TSPAN9	1	C	G	0.06	0.03	4.70E-15	++	0.03	7.20E-15
rs76567987	12	31037655	TSPAN11	1	A	G	0.84	0.02	1.70E-17	++	0.02	3.60E-17
rs698153	12	78023777	NAV3	1	A	G	0.95	-0.03	4.70E-11	--	-0.03	7.50E-11
rs10862708	12	84045567	TMTC2	0	A	T	0.56	0.02	1.80E-29	++	0.02	1.10E-28
rs1852148	12	85435434	LRR1Q1	1	A	G	0.47	-0.01	1.10E-08	--	-0.01	1.30E-08
rs11114001	12	108979235	TMEM119	1	C	G	0.19	0.01	7.30E-10	++	0.02	1.20E-09
rs9534439	13	47192049	LRCH1	1	T	C	0.19	0.02	9.90E-17	++	0.02	1.80E-16
rs2761882	14	54420309	BMP4	1	T	C	0.5	-0.01	6.40E-10	--	-0.01	9.00E-10
rs61975075	14	59505487	DAAM1	1	A	G	0.94	0.03	1.80E-14	++	0.03	2.40E-14
rs35285683	14	59519758	DAAM1	1	A	G	0.85	-0.02	2.30E-12	--	-0.02	3.30E-12
rs77587000	14	60624161	DHRS7	1	T	C	0.01	-0.09	2.90E-27	--	-0.09	3.40E-27
rs10483727	14	61072875	SIX1	1	T	C	0.39	-0.02	2.60E-21	--	-0.02	6.00E-21
rs74056359	14	65116231	PLEKHG3	1	A	G	0.83	-0.02	2.50E-16	--	-0.02	4.80E-16
rs12436074	14	85727904	SNORD3P3	1	A	G	0.56	-0.02	2.30E-19	--	-0.02	3.20E-19
rs2430356	14	92356289	FBLN5	1	A	T	0.77	-0.01	2.70E-08	--	-0.01	3.60E-08

rs11627052	14	103863476	MARK3	1	A	G	0.22	0.01	4.90E-09	++	0.01	5.50E-09
rs10910	15	74472199	STRA6	1	T	C	0.69	0.01	1.10E-08	++	0.01	1.30E-08
rs59199978	15	84484384	ADAMTSL3	1	A	G	0.82	-0.01	3.90E-09	--	-0.02	5.30E-09
rs8034595	15	96719229	NR2F2	0	A	C	0.31	-0.02	1.70E-15	--	-0.02	3.20E-15
rs7188859	16	7460426	RBFOX1	1	T	C	0.63	0.01	8.10E-12	++	0.01	1.20E-11
rs11639803	16	51396727	SALL1	0	A	G	0.77	0.02	4.80E-14	++	0.02	8.40E-14
rs7196852	16	51462823	SALL1	0	A	G	0.7	-0.02	2.30E-31	--	-0.02	1.30E-30
rs28514893	16	51573119	SALL1	0	T	C	0.21	0.02	3.90E-18	++	0.02	7.90E-18
rs11079419	17	59247327	BCAS3	1	T	C	0.2	0.03	2.30E-31	++	0.03	5.70E-31
rs34536594	17	61188396	TANC2	1	A	G	0.18	-0.02	5.50E-11	--	-0.02	1.00E-10
rs8076249	17	67573204	MAP2K6	1	T	C	0.6	-0.01	2.90E-08	--	-0.01	4.10E-08
rs9905786	17	79602063	NPLOC4	1	T	G	0.64	0.02	5.30E-25	++	0.02	2.50E-24
rs11564398	18	25668568	CDH2	1	T	C	0.72	0.01	4.30E-12	++	0.02	6.90E-12
rs10164395	19	32035891	THEG5	1	T	C	0.33	-0.01	1.70E-10	--	-0.01	2.60E-10
rs28840750	19	33519927	RHPN2	1	T	G	0.96	-0.03	2.40E-08	--	-0.03	2.70E-08
rs911526	20	31165018	LOC149950	1	A	G	0.32	-0.01	1.10E-08	--	-0.01	1.90E-08
rs2266963	22	21947467	UBE2L3	1	C	G	0.81	-0.01	1.90E-08	--	-0.01	2.30E-08
rs5762752	22	29100977	CHEK2	0	C	G	0.36	-0.02	7.90E-33	--	-0.02	2.70E-32
rs56385951	22	37909539	CARD10	0	A	G	0.11	0.03	2.90E-24	++	0.03	6.40E-24
rs9330799	22	46301368	WNT7B	1	A	G	0.53	-0.02	3.70E-14	--	-0.01	1.10E-12
rs9330813	22	46364161	WNT7B	1	A	G	0.31	-0.01	1.10E-09	--	-0.01	8.50E-09

Appendix Table 3. Top ten genetic correlated traits with disc size from LD Hub

Trait	PMID	Category	rg	se	z	p
Reason for glasses/contact lenses: For short-sightedness_ i.e. only or mainly for distance viewing such as driving_ cinema etc (called myopia)	0	ukbb	-0.2363	4.36E-02	-5.4205	5.94E-08
Reason for glasses/contact lenses: For long-sightedness_ i.e. for distance and near_ but particularly for near tasks like reading (called hypermetropia)	0	ukbb	0.2422	0.0619	3.9112	9.18E-05
Non-cancer illness code_ self-reported: retinal detachment	0	ukbb	-0.3521	0.11	-3.2015	0.0014
Job involves mainly walking or standing	0	ukbb	0.0976	0.0314	3.1126	0.0019
Years of schooling (proxy cognitive performance)	2520198	education	-0.1121	0.0371	-3.0226	0.0025
Years of schooling 2013	2372242	education	-0.1122	0.0373	-3.0055	0.0027
Years of schooling 2016	2722512	education	-0.0772	0.0265	-2.9126	0.0036
Overweight	2356360	anthropometric	0.078	0.0281	2.7779	0.0055
Forced expiratory volume in 1 second (FEV1)/Forced Vital capacity(FVC)	2194635	lung_function	0.1059	0.0396	2.6752	0.0075

Qualifications: NVQ or HND or HNC or equivalent	0 ukbb	0.1313	0.0494	2.6607	0.0078
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Appendix Table 4. Pleiotropy effects of lead disc size loci in GWAS Catalog

SNP	CHR	BP	Nearest Gene	PUBMED ID	trait	reported genes in GWAS Catalog	proxy SNP in GWAS Catalog	proxy SNP (R square)
rs12024620	1	3049362	PRDM16					
rs599892	1	39922884	MACF1					
rs787540	1	68051137	GADD45A	28073927	Optic disc area	AL590559.1 - RN7SL392P	rs787541	0.99
				28073927	Optic disc area	AL590559.1 - RN7SL392P	rs787541	0.99
rs6673575	1	81349395	LPHN2					
rs4658101	1	92077409	CDC7 - TGFB3	28073927	Vertical cup-disc ratio	AL714022.1 - HSP90B3P	rs1192414	0.88
				28073927	Vertical cup-disc ratio	AL714022.1 - HSP90B3P	rs1192414	0.88
				30595370	Lung function (FEV1/FVC)	AL714022.1 - HSP90B3P	rs1192415	0.97
				30595370	Lung function (FVC)	AL714022.1 - HSP90B3P	rs1192415	0.97
				21307088	Optic disc area	AL714022.1 - HSP90B3P	rs1192415	0.97
				20548946	Optic disc parameters	AL714022.1 - HSP90B3P	rs1192415	0.97
				20395239	Optic nerve measurement (disc area)	AL714022.1 - HSP90B3P	rs1192415	0.97
				28061514	Optic nerve measurement (cup-to-disc ratio)	AL714022.1 - HSP90B3P	rs1192419	1
				25631615	Optic disc area	AL714022.1 - HSP90B3P	rs1192419	1
				25631615	Optic disc area	AL714022.1 - HSP90B3P	rs1192419	1
				28073927	Optic disc area	AL714022.1 - HSP90B3P	rs4658101	1
				28073927	Optic disc area	AL714022.1 - HSP90B3P	rs4658101	1
				25241763	Vertical cup-disc ratio	AL714022.1 - HSP90B3P	rs4658101	1
rs3754442	1	110611886	ALX3	24816252	Blood metabolite levels	STRIP1 - LINC01397	rs1466788	0.87
				30038396	Cognitive performance (MTAG)	STRIP1	rs3768469	0.88
rs4839470	1	116209290	VANGL1					
rs12137699	1	116209762	VANGL1	27694959	Gut microbiota (bacterial taxa)	VANGL1	rs12137699	1

rs57742792	1	155071588	EFNA3					
rs6030	1	169498975	F5	27989323	Stem cell factor levels	F5	rs1557570	0.98
				27989323	Stem cell growth factor beta levels	AL021068.1 - F5	rs4656185	0.95
				23703922	Uric acid levels	F5	rs6030	1
rs742127	1	169564167	SELP	25631615	Optic disc area	F5	rs12406092	0.9
				25631615	Optic disc area	F5	rs12406092	0.9
				22703881	Activated partial thromboplastin time	F5	rs6028	0.87
rs11584075	1	201598915	NAV1					
rs11808696	1	227563680A	CDC42BP	30535121	Macular thickness	CDC42BPA - LINC01641	rs112123925	1
				28073927	Optic disc area	LINC01641	rs11811982	0.98
				28073927	Optic disc area	LINC01641	rs11811982	0.98
rs340762	2	16484005	FAM49A					
rs72784719	2	19344098	MIR4757					
rs13417287	2	33219147	LTBP1					
rs116072427	2	33357096	LTBP1	30593698	Fat-free mass	LTBP1	rs115179432	0.98
				30593698	Fat-free mass	LTBP1	rs115179432	0.98
				29235454	Intraocular pressure	LTBP1	rs115179432	0.98
				29760442	Central corneal thickness	LTBP1	rs115781177	1
				29617998	Intraocular pressure	LTBP1	rs116713089	0.97
rs163524	2	45157553	SIX3	30054594	Intraocular pressure	LINC01833	rs163524	1
				29785010	Intraocular pressure	LINC01833	rs163524	1
				29617998	Intraocular pressure	LINC01833	rs163524	1
rs9967780	2	56234942	MIR216B					
rs4832012	2	86000500	ATOH8					
rs11684168	2	88694167	FOXI3					
rs12619508	2	113982040	PAX8	27863252	Lymphocyte counts	PAX8, PAX8-AS1	rs2019137	0.87
rs1901440	2	134437959	NCKAP5	22293537	Response to gemcitabine in pancreatic cancer	RN7SKP93 - MGAT5	rs1901440	1
rs12471698	2	145475412	TEX41	29785010	Intraocular pressure	TEX41	rs10928224	0.84
				30595370	White blood cell count	TEX41	rs3856364	0.91
rs9287914	2	170155247	LRP2					

rs13022913	2	180175927	SESTD1					
rs10084346	2	218306736	DIRC3					
rs934237	2	218467037	DIRC3	28073927	Optic disc area	DIRC3	rs1367187	1
				28073927	Optic disc area	DIRC3	rs1367187	1
				25631615	Optic disc area	DIRC3	rs1549733	0.99
				25631615	Optic disc area	DIRC3	rs1549733	0.99
rs1550094	2	233385396	PRSS56	29808027	Myopia (age of diagnosis)	PRSS56	rs1550094	1
				29808027	Spherical equivalent or myopia (age of diagnosis)	PRSS56	rs1550094	1
				27182965	Myopia	PRSS56	rs1550094	1
rs7620608	3	20095378	KAT2B					
rs7615960	3	21730246	ZNF385D					
rs4858678	3	25047446	RARB	28073927	Optic disc area	AC092422.1	rs11129176	1
				28073927	Optic disc area	AC092422.1	rs11129176	1
				25631615	Optic disc area	AC092422.1	rs11129176	1
				25631615	Optic disc area	AC092422.1	rs11129176	1
				30535121	Macular thickness	AC092422.1	rs12487626	0.96
rs6787363	3	25388569	RARB					
rs13070614	3	25414588	RARB					
rs71296770	3	62371837	CADPS					
rs77877421	3	71182447	FOXP1					
rs7610486	3	99732564	FILIP1L	28232668	Nonsyndromic cleft lip with cleft palate	FILIP1L, CMSS1	rs1688766	0.82
				28054174	Cleft lip with or without cleft palate	FILIP1L, CMSS1	rs7616988	0.83
				29059683	Breast cancer	FILIP1L, CMSS1	rs9833888	1
				29059683	Breast cancer	FILIP1L, CMSS1	rs9837602	0.89
rs17398137	3	100625703	ABI3BP	28073927	Optic disc area	ABI3BP	rs9843102	0.98
				28073927	Optic disc area	ABI3BP	rs9843102	0.98
				25631615	Optic disc area	ABI3BP	rs9860250	0.95
				25631615	Optic disc area	ABI3BP	rs9860250	0.95
rs2220051	3	156764263	LEKR1					
rs9852867	3	189969372	CLDN1					
rs28603236	4	41561962	LIMCH1					

rs2033054	4	55018617	GSX2					
rs74764079	4	81952637	BMP3					
rs906568	4	112402558	C4orf32					
rs72759609	5	31952051	PDZD2	28073927	Vertical cup-disc ratio	PDZD2	rs72759609	1
				28073927	Vertical cup-disc ratio	PDZD2	rs72759609	1
rs251526	5	52586585	FST					
rs30371	5	55742832	ANKRD55	30535121	Macular thickness	HMG1P17 - LINC01948	rs30373	0.89
rs7717697	5	82744604	VCAN	28073927	Vertical cup-disc ratio	FTH1P9 - VCAN	rs7717697	1
				28073927	Vertical cup-disc ratio	FTH1P9 - VCAN	rs7717697	1
rs73173591	5	878305361	LINC00461	23776548	Retinal arteriolar caliber	AC091826.2 - LINC00461	rs2194025	0.81
				28250457	Macular telangiectasia type 2	LINC00461	rs73173548	0.87
rs6860726	5	126259178	MARCH3					
rs62391624	6	1923520	GMDS	25173105	Glaucoma (primary open-angle)	GMDS	rs11969985	0.9
rs2092524	6	39529692	KIF6					
rs7744813	6	73643289	KCNQ5	29808027	Myopia (age of diagnosis)	KCNQ5	rs7744813	1
				29808027	Spherical equivalent or myopia (age of diagnosis)	KCNQ5	rs7744813	1
				27182965	Myopia	KCNQ5	rs7744813	1
				27020472	Spherical equivalent (joint analysis main effects and education interaction)	KCNQ5	rs7744813	1
				27020472	Spherical equivalent (joint analysis main effects and education interaction)	KCNQ5	rs7744813	1
				23396134	Refractive error	KCNQ5	rs7744813	1
rs34607745	6	122675335	HSF2					
rs112166936	6	126733140	CENPW	29500382	Feeling nervous	CENPW - MIR588	rs1159619	0.83
				30573740	Male-pattern baldness	CENPW - MIR588	rs1361108	0.89
				29617998	Intraocular pressure	CENPW - MIR588	rs1361108	0.89
				21998595	Height	CENPW - MIR588	rs1361108	0.89
				21102462	Menarche (age at onset)	CENPW - MIR588	rs1361108	0.89
				18391951	Height	MIR588 - RNU6-200P	rs1490388	0.82

			25673412	Hip circumference adjusted for BMI	CENPW - MIR588	rs1538170	0.89
			25673412	Hip circumference adjusted for BMI	CENPW - MIR588	rs1538170	0.89
			25751624	Type 1 diabetes	CENPW - MIR588	rs1538171	0.89
			28270201	Height	CENPW - MIR588	rs1578060	0.89
			29844566	General cognitive ability	MIR588 - RNU6-200P	rs1602278	0.82
			30595370	Menarche (age at onset)	CENPW - MIR588	rs1844594	0.83
			30595370	Balding type 1	MIR588 - RNU6-200P	rs1907067	0.82
			27863252	Hematocrit	MIR588 - RNU6-200P	rs2326451	0.82
			24945404	Bone mineral density (paediatric, skull)	MIR588 - RNU6-200P	rs4418209	0.83
			25231870	Menarche (age at onset)	CENPW - MIR588	rs4895808	0.89
			28869590	Type 2 diabetes	CENPW - MIR588	rs4897182	0.89
			28869590	Type 2 diabetes	CENPW - MIR588	rs4897182	0.89
			29500382	Feeling nervous	PPP1R14BP5 - CENPW	rs6919397	0.85
			29500382	Neuroticism	PPP1R14BP5 - CENPW	rs6919397	0.85
			25281659	Infant length	CENPW - MIR588	rs9385399	0.89
			29844566	General cognitive ability	CENPW - MIR588	rs9388487	0.8
			19430480	Type 1 diabetes	CENPW - MIR588	rs9388489	0.88
			30593698	Fat-free mass	CENPW - MIR588	rs9388490	0.83
			30593698	Fat-free mass	CENPW - MIR588	rs9388490	0.83
			30593698	Fat-free mass	CENPW - MIR588	rs9388490	0.83
			30038396	Educational attainment (MTAG)	CENPW - MIR588	rs9388490	0.83
			30038396	Highest math class taken	CENPW - MIR588	rs9388490	0.83
			30038396	Educational attainment (years of education)	CENPW - MIR588	rs9388490	0.83
			30038396	Highest math class taken (MTAG)	CENPW - MIR588	rs9388490	0.83
			30038396	Cognitive performance (MTAG)	CENPW - MIR588	rs9388490	0.83
			29326435	Intelligence (MTAG)	CENPW - MIR588	rs9388490	0.83

				28196072	Male-pattern baldness	CENPW - MIR588	rs9388490	0.83
				29146897	Male-pattern baldness	CENPW - MIR588	rs9398803	0.8
				30595370	Lung function (FEV1/FVC)	CENPW - MIR588	rs9398804	0.85
rs1930941	6	127316843	RSPO3					
rs80218807	8	1884839	ARHGEF10					
rs1905014	8	13407297	C8orf48					
rs10957177	8	61911070	LOC100130298					
rs3857971	8	61969579	LOC100130298					
rs17734073	8	72598450	MSC					
rs6999835	8	78948855	PKIA	29617998	Intraocular pressure	AC062004.1 - AC084706.1	rs2219968	0.93
				30054594	Intraocular pressure	AC062004.1 - AC084706.1	rs35174414	0.91
				29785010	Intraocular pressure	AC062004.1 - AC084706.1	rs62508223	0.91
rs12547416	8	88761223	DCAF4L2					
rs13264644	8	143753546	PSCA					
rs10512176	9	89252706	ZCCHC6					
rs16909978	9	98307990	PTCH1					
rs13283933	9	1345843081	RAPGEF81					
rs10764494	10	25058144	ARHGAP21					
rs7916410	10	69995667	ATOH7	25631615	Optic disc area	AC016395.1	rs1900004	1
				25631615	Optic disc area	AC016395.1	rs1900004	1
				20548946	Optic disc parameters	AC016395.1	rs1900004	1
				20548946	Vertical cup-disc ratio	AC016395.1	rs1900004	1
				28073927	Vertical cup-disc ratio	AC016395.1	rs1900005	1
				28073927	Vertical cup-disc ratio	AC016395.1	rs1900005	1
				25241763	Vertical cup-disc ratio	AC016395.1	rs1900005	1
				28061514	Optic nerve measurement (cup-to-disc ratio)	AC016395.1	rs56238729	1
				28073927	Optic disc area	AC016395.1	rs7916410	1
				28073927	Optic disc area	AC016395.1	rs7916410	1
				21307088	Optic disc area	ATOH7	rs7916697	0.93
rs9988687	10	70024528	PBLD	25631615	Optic cup area	AC016395.1 - KRT19P4	rs3858145	0.86

				25631615	Optic cup area	AC016395.1 - KRT19P4	rs3858145	0.86
				20395239	Optic nerve measurement (disc area)	AC016395.1 - KRT19P4	rs3858145	0.86
				20395239	Optic nerve measurement (cup area)	AC016395.1 - KRT19P4	rs3858145	0.86
rs383510	10	70051810	PBLD					
rs709631	10	71610049	COL13A1					
rs10823610	10	72506158	ADAMTS14					
rs2149108	10	94866290	CYP26A1					
rs4285804	10	104386309	SUFU	28443625	Waist circumference adjusted for BMI (joint analysis main effects and smoking interaction)	SUFU	rs3862030	0.8
				28443625	Waist circumference adjusted for BMI (joint analysis main effects and smoking interaction)	SUFU	rs3862030	0.8
				28443625	Waist circumference adjusted for BMI (adjusted for smoking behaviour)	SUFU	rs3862030	0.8
				28443625	Waist circumference adjusted for BMI (adjusted for smoking behaviour)	SUFU	rs3862030	0.8
				25673412	Waist circumference adjusted for body mass index	SUFU	rs3862030	0.8
				25673412	Waist circumference adjusted for body mass index	SUFU	rs3862030	0.8
				30595370	Eczema	SUFU	rs7899004	0.81
rs11602582	11	31285284	DCDC1					
rs7482884	11	31683485	ELP4	25631615	Optic disc area	AC131571.1, ELP4	rs11031436	0.94
				25631615	Optic disc area	AC131571.1, ELP4	rs11031436	0.94
				28073927	Optic disc area	IMMP1L	rs61101201	0.84
				28073927	Optic disc area	IMMP1L	rs61101201	0.84
rs78977588	11	68968149	MYEOV					
rs1126809	11	89017961	TYR	30595370	Sunburns	TYR	rs1126809	1
				29739929	Low tan response	TYR	rs1126809	1
				27723757	Vitiligo	TYR	rs1126809	1
				27539887	Basal cell carcinoma	TYR	rs1126809	1
				27424798	Cutaneous squamous cell carcinoma	TYR	rs1126809	1

				26829030	Squamous cell carcinoma	TYR	rs1126809	1
				23548203	Tanning	TYR	rs1126809	1
				23548203	Sunburns	TYR	rs1126809	1
rs74891390	12	3373823	TSPAN9	28452372	Glomerular filtration rate (creatinine)	TSPAN9	rs67551338	0.89
rs76567987	12	31037655	TSPAN11					
rs698153	12	78023777	NAV3					
rs10862708	12	84045567	TMTC2	29891935	Glaucoma (primary open-angle)	AC090680.1 - AC093025.1	rs12309467	0.82
				25631615	Optic disc area	AC093025.1 - AC090679.2	rs1511589	0.99
				25631615	Optic disc area	AC093025.1 - AC090679.2	rs1511589	0.99
				25631615	Optic cup area	AC093025.1 - AC090679.2	rs1511589	0.99
				28073927	Vertical cup-disc ratio	AC090680.1 - AC093025.1	rs324780	0.89
				28073927	Vertical cup-disc ratio	AC090680.1 - AC093025.1	rs324780	0.89
rs1852148	12	85435434	LRR1Q1					
rs11114001	12	108979235	TMEM119					
rs9534439	13	47192049	LRCH1					
rs2761882	14	54420309	BMP4	29808027	Spherical equivalent or myopia (age of diagnosis)	BMP4	rs2855530	0.95
rs61975075	14	59505487	DAAM1					
rs35285683	14	59519758	DAAM1					
rs77587000	14	60624161	DHRS7					
rs10483727	14	61072875	SIX1	26752265	Glaucoma (high intraocular pressure)	AL049874.1 - SALR1	rs10483727	1
				25631615	Optic cup area	AL049874.1 - SALR1	rs10483727	1
				25631615	Optic cup area	AL049874.1 - SALR1	rs10483727	1
				25631615	Optic disc area	AL049874.1 - SALR1	rs10483727	1
				25173105	Glaucoma (primary open-angle)	AL049874.1 - SALR1	rs10483727	1
				22570617	Glaucoma (primary open-angle)	AL049874.1 - SALR1	rs10483727	1
				22419738	Glaucoma (primary open-angle)	AL049874.1 - SALR1	rs10483727	1
				20548946	Vertical cup-disc ratio	AL049874.1 - SALR1	rs10483727	1
				20395239	Optic nerve measurement (rim area)	AL049874.1 - SALR1	rs10483727	1

				29891935	Glaucoma (primary open-angle)	AL049874.1 - SALR1	rs34935520	0.97
				29891935	Glaucoma (primary open-angle)	AL049874.1 - SALR1	rs34935520	0.97
				28073927	Optic disc area	AL049874.1 - SALR1	rs34935520	0.97
				29891935	Glaucoma (primary open-angle)	AL049874.1 - SALR1	rs35155027	0.95
				30593698	Fat-free mass	SALR1	rs35320790	0.81
				30593698	Fat-free mass	SALR1	rs35320790	0.81
				30593698	Fat-free mass	SALR1	rs35320790	0.81
rs74056359	14	65116231	PLEKHG3					
rs12436074	14	85727904	SNORD3P3	23534349	QRS duration	AL357172.1 - AL049775.2	rs1867082	1
				28928442	Pneumonia	AL357172.1 - AL049775.2	rs8012486	0.85
rs2430356	14	92356289	FBLN5					
rs11627052	14	103863476	MARK36					
rs10910	15	74472199	STRA6					
rs59199978	15	84484384	ADAMTS L3					
rs8034595	15	96719229	NR2F2	25631615	Optic disc area	NR2F2-AS1, AC012409.2	rs8034595	1
rs7188859	16	7460426	RBFOX1	29808027	Spherical equivalent or myopia (age of diagnosis)	RBFOX1	rs10500355	0.99
				27182965	Myopia	RBFOX1	rs10500355	0.99
				23474815	Refractive error	RBFOX1	rs10500355	0.99
				29808027	Myopia (age of diagnosis)	RBFOX1	rs17648524	0.99
				25233373	Hyperopia	RBFOX1	rs17648524	0.99
				23396134	Refractive error	RBFOX1	rs17648524	0.99
				29808027	Spherical equivalent	RBFOX1	rs4332760	0.88
				29808027	Spherical equivalent	RBFOX1	rs4332760	0.88
rs11639803	16	51396727	SALL1					
rs7196852	16	51462823	SALL1	28073927	Optic disc area	AC007344.1	rs1345467	0.85
				28073927	Optic disc area	AC007344.1	rs1345467	0.85
				25241763	Vertical cup-disc ratio	AC007344.1	rs1345467	0.85
				25631615	Optic disc area	AC087564.1, AC007344.1	rs1362756	0.98
				25631615	Optic disc area	AC087564.1, AC007344.1	rs1362756	0.98
				20548946	Optic disc parameters	AC087564.1, AC007344.1	rs1362756	0.98
rs28514893	16	51573119	SALL1					
rs11079419	17	59247327	BCAS3	25631615	Optic cup area	BCAS3	rs11651885	0.85

				25631615	Optic cup area	BCAS3	rs11651885	0.85
				28073927	Optic cup area	BCAS3	rs11867840	0.86
				28073927	Optic cup area	BCAS3	rs11867840	0.86
				30595370	Lung function (FEV1/FVC)	BCAS3	rs2204928	0.94
				30535121	Macular thickness	BCAS3	rs723835	0.86
				28714975	Coronary artery disease (myocardial infarction, percutaneous transluminal coronary angioplasty, coronary artery bypass grafting, angina or chronic ischemic heart disease)	BCAS3	rs8068952	0.8
				20548946	Vertical cup-disc ratio	BCAS3	rs8068952	0.8
rs34536594	17	61188396	TANC2	29326435	Intelligence (MTAG)	TANC2	rs72843166	0.81
rs8076249	17	67573204	MAP2K6					
rs9905786	17	79602063	NPLOC4	30306274	Refractive astigmatism	NPLOC4	rs34635363	0.91
				30531825	Brown vs. black hair color	TSPAN10, NPLOC4	rs62075722	0.96
				30306274	Corneal astigmatism	TSPAN10, NPLOC4	rs62075722	0.96
				30306274	Corneal astigmatism	TSPAN10, NPLOC4	rs62075722	0.96
				29808027	Myopia (age of diagnosis)	NPLOC4, TSPAN10	rs62075723	0.92
				30595370	Lung function (FEV1/FVC)	NPLOC4, TSPAN10	rs6420484	0.96
				29808027	Spherical equivalent or myopia (age of diagnosis)	NPLOC4, TSPAN10	rs6420484	0.96
				29617998	Intraocular pressure	NPLOC4	rs67050149	0.92
				30535121	Macular thickness	TSPAN10	rs7405453	0.89
				29617998	Intraocular pressure	TSPAN10	rs7405453	0.89
				30595370	Hair color	NPLOC4, TSPAN10	rs9747347	0.91
				30531825	Red vs. brown/black hair color	NPLOC4, TSPAN10	rs9747347	0.91
				27182965	Myopia	NPLOC4, TSPAN10	rs9747347	0.91
rs11564398	18	25668568	CDH2	30038396	Educational attainment (years of education)	CDH2	rs1220027	0.83
rs10164395	19	32035891	THEG5					
rs28840750	19	33519927	RHPN2	30598549	Heel bone mineral density	RHPN2	rs28626308	0.98
				30048462	Heel bone mineral density	RHPN2	rs28626308	0.98
rs911526	20	31165018	LOC149950	30595370	Red blood cell count	NOL4L	rs159058	0.81
				27863252	Mean corpuscular hemoglobin	NOL4L	rs159058	0.81

				27863252	Immature fraction of reticulocytes	NOL4L	rs159058	0.81
				27863252	Mean corpuscular volume	NOL4L	rs159058	0.81
				30038396	Educational attainment (MTAG)	NOL4L, NOL4L-DT	rs6119267	0.92
				30038396	Highest math class taken (MTAG)	NOL4L, NOL4L-DT	rs6119267	0.92
				27863252	Red cell distribution width	NOL4L, NOL4L-DT	rs6119267	0.92
				27863252	White blood cell count (basophil)	NOL4L-DT, AL133343.1	rs67696533	0.88
				27863252	Basophil percentage of white cells	NOL4L-DT, AL133343.1	rs67696533	0.88
				27863252	Basophil percentage of granulocytes	NOL4L-DT, AL133343.1	rs67696533	0.88
rs226963	22	21947467	UBE2L3	30595370	Red cell distribution width	UBE2L3	rs131656	0.84
				27863252	High light scatter reticulocyte count	UBE2L3	rs131656	0.84
				27863252	High light scatter reticulocyte percentage of red cells	UBE2L3	rs131656	0.84
				28714469	Systemic lupus erythematosus	UBE2L3	rs131658	0.86
				27863252	Red cell distribution width	UBE2L3	rs138665726	0.93
				27863252	Mean corpuscular volume	UBE2L3	rs138665726	0.93
				30595370	Eczema	UBE2L3 - YDJC	rs1811069	0.91
				30595370	Eosinophil counts	UBE2L3 - YDJC	rs1811069	0.91
				30595370	Mean corpuscular hemoglobin	UBE2L3 - YDJC	rs1811069	0.91
				29507422	High density lipoprotein cholesterol levels	UBE2L3 - YDJC	rs1811069	0.91
				29507422	High density lipoprotein cholesterol levels	UBE2L3 - YDJC	rs1811069	0.91
				30019117	Adolescent idiopathic scoliosis	UBE2L3	rs181359	0.87
				26626624	Psoriasis vulgaris	UBE2L3	rs181359	0.87
				21102463	Crohn's disease	UBE2L3	rs181359	0.87
				28334899	HDL cholesterol levels	UBE2L3	rs181360	0.87
				29617998	Intraocular pressure	UBE2L3	rs181362	0.89
				29507422	High density lipoprotein cholesterol levels	UBE2L3	rs181362	0.89
				29507422	High density lipoprotein cholesterol levels	UBE2L3	rs181362	0.89
				24097068	HDL cholesterol	UBE2L3	rs181362	0.89
				20686565	HDL cholesterol	UBE2L3	rs181362	0.89
				26192919	Crohn's disease	UBE2L3	rs2256609	0.93
				28067908	Crohn's disease	UBE2L3	rs2266959	0.95
				28067908	Inflammatory bowel disease	UBE2L3	rs2266959	0.95
				28067908	Ulcerative colitis	UBE2L3	rs2266959	0.95

			23603761	Juvenile idiopathic arthritis (oligoarticular or rheumatoid factor-negative polyarticular)	UBE2L3	rs2266959	0.95
			23128233	Inflammatory bowel disease	UBE2L3	rs2266959	0.95
			26974007	Chronic inflammatory diseases (ankylosing spondylitis, Crohn's disease, psoriasis, primary sclerosing cholangitis, ulcerative colitis) (pleiotropy)	UBE2L3	rs2266961	0.95
			26192919	Inflammatory bowel disease	UBE2L3	rs2266961	0.95
			27193031	Systemic lupus erythematosus or rheumatoid arthritis	UBE2L3	rs2283790	0.9
			23749187	Ankylosing spondylitis	UBE2L3	rs2283790	0.9
			23749187	Ankylosing spondylitis	UBE2L3	rs2283790	0.9
			30048462	Heel bone mineral density	YDJC	rs2298428	0.95
			21383967	Celiac disease or Rheumatoid arthritis	YDJC	rs2298428	0.95
			20190752	Celiac disease	YDJC	rs2298428	0.95
			28017375	Mean corpuscular volume	UBE2L3	rs4820091	0.88
			20139978	Mean corpuscular volume	UBE2L3	rs4821112	0.9
			28334899	HDL cholesterol levels	UBE2L3	rs4821116	0.98
			28334899	HDL cholesterol levels	UBE2L3	rs4821116	0.98
			24162738	Hepatitis B	UBE2L3	rs4821116	0.98
			23143594	Psoriasis	UBE2L3 - YDJC	rs4821124	0.97
			22057235	Celiac disease	UBE2L3 - YDJC	rs4821124	0.97
			30595370	Autoimmune traits	UBE2L3	rs5754100	0.89
			27863252	Lymphocyte counts	UBE2L3	rs5754100	0.89
			30275531	Total cholesterol levels	UBE2L3	rs5754102	0.83
			29617998	Intraocular pressure	UBE2L3	rs5754102	0.83
			28017375	Mean corpuscular volume	UBE2L3	rs5754217	0.92
			23273568	Systemic lupus erythematosus	UBE2L3	rs5754217	0.92
			23222517	Red blood cell traits	UBE2L3	rs5754217	0.92
			21408207	Systemic lupus erythematosus	UBE2L3	rs5754217	0.92
			19838195	Systemic lupus erythematosus	UBE2L3	rs5754217	0.92
			25961943	HDL cholesterol	UBE2L3	rs5754344	0.97
			30598549	Heel bone mineral density	UBE2L3	rs5754387	0.89
			30595370	Heel bone mineral density	UBE2L3	rs5754387	0.89
			27863252	Neutrophil percentage of white cells	UBE2L3	rs5998509	0.91
			27863252	Lymphocyte percentage of white cells	UBE2L3	rs5998509	0.91
			30275531	HDL cholesterol	UBE2L3	rs7444	0.91
			28714469	Systemic lupus erythematosus	UBE2L3	rs7444	0.91

				27399966	Systemic lupus erythematosus	UBE2L3	rs7444	0.91
				27399966	Systemic lupus erythematosus	UBE2L3	rs7444	0.91
				26502338	Systemic lupus erythematosus	UBE2L3	rs7444	0.91
				29403010	High density lipoprotein cholesterol levels	UBE2L3	rs7445	0.89
				29403010	Mean corpuscular volume	UBE2L3	rs8137950	0.86
				30595370	White blood cell count	UBE2L3 - YDJC	rs878825	0.9
				28957414	Red cell distribution width	UBE2L3 - YDJC	rs878825	0.9
rs5762752	22	29100977	CHEK2	25241763	Vertical cup-disc ratio	CHEK2	rs1547014	0.83
				20548946	Vertical cup-disc ratio	CHEK2	rs1547014	0.83
				28073927	Vertical cup-disc ratio	CHEK2	rs5752773	0.8
				28073927	Vertical cup-disc ratio	CHEK2	rs5752773	0.8
				29891935	Glaucoma (primary open-angle)	CHEK2	rs5752774	0.8
				28073927	Optic disc area	CHEK2	rs5762752	1
				28073927	Optic disc area	CHEK2	rs5762752	1
rs56385951	22	37909539	CARD10	28073927	Optic disc area	CARD10	rs56385951	1
				28073927	Optic disc area	CARD10	rs56385951	1
rs9330799	22	46301368	WNT7B					
rs9330813	22	46364161	WNT7B	27863252	Hemoglobin concentration	WNT7B - BX537318.2	rs62228060	0.96
				30535121	Macular thickness	WNT7B - BX537318.2	rs75159625	0.88
				30595370	Red blood cell count	WNT7B	rs9330813	1
				30573740	Male-pattern baldness	WNT7B	rs9330813	1
				29847655	Central corneal thickness	WNT7B	rs9330813	1
				29847655	Central corneal thickness	WNT7B	rs9330813	1
				27863252	Red blood cell count	WNT7B	rs9330813	1
				27863252	Hematocrit	WNT7B	rs9330813	1

Appendix Table 5. 76 statistically independent SNPs from UKBB GWAS of VCDR (vertical DD adjusted)

SNP	Chr	Position	EA	NEA	Freq	BETA	SE	P	Nearest gene
rs12024620	1	3049362	C	T	0.94	-0.01	1.83E-03	1.2E-10	<i>PRDM16</i>
rs6690264	1	12613422	A	G	0.42	-6.08E-03	9.33E-04	7.2E-11	<i>DHRS3</i>

rs34151819	1	68773910	C	T	0.98	0.02	3.46E-03	5.4E-10	WLS
rs3125918	1	68846246	A	G	0.6	-7.08E-03	9.41E-04	5.2E-14	RPE65
rs4658101	1	92077409	A	G	0.19	0.01	1.12E-03	8.9E-35	CDC7-TGFBR3
rs1417488	1	218523730	C	T	0.75	-6.25E-03	1.05E-03	2.8E-09	TGFB2
rs77271542	1	227677723	A	T	0.94	-0.01	1.87E-03	6.5E-14	ZNF678
rs376096585	2	56094578	C	CT	0.78	9.33E-03	1.08E-03	3.9E-18	EFEMP1
rs2880192	2	111680818	A	G	0.65	6.39E-03	9.36E-04	9.0E-12	ACOXL
rs1579050	2	153364527	A	G	0.42	-5.39E-03	9.17E-04	4.1E-09	FMNL2
rs4858682	3	25066225	C	G	0.47	-5.26E-03	8.92E-04	3.6E-09	RARB
rs34010125	3	32878682	T	C	0.68	5.99E-03	9.53E-04	3.3E-10	TRIM71
3:88379094_AT_A	3	88379094	AT	A	0.45	5.53E-03	8.99E-04	7.8E-10	C3orf38
rs4928176	3	98972835	G	A	0.41	-7.20E-03	9.19E-04	4.5E-15	MIR548G
rs6804624	3	99159147	T	C	0.69	-7.35E-03	9.85E-04	8.5E-14	MIR548G
rs9827694	3	100648298	G	A	0.82	7.09E-03	1.16E-03	8.6E-10	ABI3BP
rs143351962	3	134089758	C	T	0.99	-0.03	4.58E-03	4.8E-09	AMOTL2
rs2162137	4	55005856	C	T	0.34	7.10E-03	9.63E-04	1.8E-13	GSX2
rs565335773	4	55095682	G	GA	0.79	7.81E-03	1.12E-03	3.7E-12	PDGFRA
rs72759609	5	31952051	T	C	0.9	0.01	1.47E-03	1.3E-14	PDZD2
rs158653	5	55578661	G	A	0.48	5.33E-03	8.74E-04	1.1E-09	ANKRD55
rs30372	5	55744230	T	C	0.24	-6.49E-03	1.07E-03	1.2E-09	LOC102467147
rs11749004	5	128918456	T	C	0.25	5.71E-03	1.04E-03	3.7E-08	ADAMTS19
5:133393380_GA_G	5	133393380	GA	G	0.84	8.98E-03	1.31E-03	8.1E-12	VDAC1
rs34471628	5	172196752	A	G	0.96	0.01	2.28E-03	3.4E-08	DUSP1
rs2761235	6	1951135	C	T	0.87	8.72E-03	1.36E-03	1.6E-10	GMDS
rs4960297	6	7212084	C	T	0.55	-6.54E-03	8.82E-04	1.2E-13	RREB1
rs12211825	6	36588140	C	T	0.78	-7.48E-03	1.08E-03	4.0E-12	SRSF3
rs2684249	6	122392511	T	C	0.59	5.59E-03	9.20E-04	1.2E-09	HSF2
rs10260511	7	14237240	C	A	0.84	-7.04E-03	1.25E-03	1.6E-08	DGKB
rs4518562	7	19627952	A	T	0.66	-5.79E-03	9.43E-04	8.0E-10	TWISTNB
rs7805378	7	28393403	A	C	0.56	5.48E-03	9.26E-04	3.3E-09	CREB5
rs2976932	8	8255319	T	C	0.11	-9.18E-03	1.43E-03	1.2E-10	SGK223
rs12543430	8	72278010	T	C	0.39	-5.48E-03	9.14E-04	2.0E-09	EYA1
rs78542921	9	18089832	T	A	0.96	-0.01	2.38E-03	8.1E-10	SH3GL2
rs7039467	9	22056213	A	G	0.69	-0.01	1.13E-03	2.0E-37	CDKN2B-AS1

rs7866783	9	22056359	A	G	0.43	-0.02	9.07E-04	5.4E-64	<i>CDKN2B-AS1</i>
10:21462896_GGC_G	10	21462896	GGC	G	0.99	-0.03	4.06E-03	2.4E-10	<i>NEBL</i>
rs7916697	10	69991853	A	G	0.24	-0.01	1.02E-03	6.8E-35	<i>ATOH7</i>
rs17108260	10	94950713	A	G	0.57	-6.05E-03	9.07E-04	2.6E-11	<i>CYP26A1</i>
rs11197820	10	118546046	G	A	0.58	-5.22E-03	8.75E-04	2.4E-09	<i>HSPA12A</i>
rs10835721	11	30934691	G	A	0.73	-5.48E-03	9.90E-04	3.1E-08	<i>DCDC5</i>
rs2753411	11	33405001	A	T	0.49	-4.74E-03	8.57E-04	3.1E-08	<i>HIPK3</i>
rs1346	11	65337251	A	T	0.8	8.26E-03	1.11E-03	1.2E-13	<i>SSSCA1-AS1</i>
rs2445575	11	86748437	T	C	0.81	6.34E-03	1.11E-03	1.1E-08	<i>TMEM135</i>
rs11021221	11	95308854	T	A	0.83	6.33E-03	1.15E-03	3.3E-08	<i>FAM76B</i>
rs4936099	11	130280725	C	A	0.41	-8.52E-03	8.92E-04	1.3E-21	<i>ADAMTS8</i>
rs6582298	12	76114872	G	A	0.6	5.14E-03	9.28E-04	3.0E-08	<i>KRR1</i>
rs61952219	12	83959231	G	A	0.79	-0.02	1.53E-03	8.0E-25	<i>TMTC2</i>
12:83973565_TTCTC_T	12	83973565	TTCTC	T	0.45	-0.01	8.80E-04	3.9E-58	<i>TMTC2</i>
rs9651957	12	107170128	T	C	0.64	6.11E-03	8.94E-04	8.6E-12	<i>RIC8B</i>
rs4765353	12	124667691	G	C	0.44	-5.57E-03	9.25E-04	1.8E-09	<i>ZNF664-FAM101A</i>
rs9546383	13	36683268	T	C	0.75	-8.77E-03	1.02E-03	9.2E-18	<i>DCLK1</i>
rs10162202	13	109267985	T	C	0.72	7.11E-03	9.95E-04	9.1E-13	<i>MYO16</i>
13:110778747_CCTTTT_C	13	110778747	CCTTTT	C	0.64	-7.29E-03	9.69E-04	5.3E-14	<i>COL4A1</i>
rs4982708	14	23391086	G	A	0.43	6.44E-03	8.69E-04	1.2E-13	<i>PRMT5</i>
rs2251171	14	53989447	G	A	0.6	5.71E-03	8.77E-04	7.9E-11	<i>DDHD1</i>
rs10162287	14	60808553	C	G	0.7	-9.99E-03	9.65E-04	4.1E-25	<i>PPM1A</i>
rs984586	14	86009142	G	A	0.76	-5.90E-03	1.03E-03	1.2E-08	<i>FLRT2</i>
rs893817	15	74229065	G	A	0.37	5.36E-03	9.01E-04	2.7E-09	<i>LOXL1</i>
rs148139847	15	101202269	C	CA	0.87	-9.78E-03	1.33E-03	1.6E-13	<i>ASB7</i>
rs373836950	16	51447492	C	CAA	0.8	0.01	1.14E-03	3.7E-21	<i>SALL1</i>
rs8053277	16	51469726	T	C	0.3	9.91E-03	9.63E-04	7.3E-25	<i>SALL1</i>
rs2720429	16	51571885	G	A	0.77	-0.01	1.12E-03	2.2E-19	<i>SALL1</i>
rs35526343	16	86465334	C	T	0.96	-0.02	2.23E-03	8.8E-13	<i>FENDRR</i>
rs847688	17	48228981	T	C	0.52	5.03E-03	9.00E-04	2.3E-08	<i>PPP1R9B</i>
rs2204928	17	59262040	C	T	0.21	-6.47E-03	1.08E-03	2.3E-09	<i>BCAS3</i>
rs146055611	19	816586	C	CATCT	0.66	6.39E-03	9.82E-04	7.3E-11	<i>LPPR3</i>
rs8102936	19	32027330	G	A	0.66	6.46E-03	1.03E-03	4.0E-10	<i>THEG5</i>
rs6140010	20	6473123	A	G	0.63	0.01	9.07E-04	9.4E-43	<i>CASC20</i>

rs3831804	20	31413015	T	TA	0.78	6.44E-03	1.07E-03	1.9E-09	<i>MAPRE1</i>
rs7287609	22	29096693	C	T	0.99	0.03	4.07E-03	8.2E-15	<i>CHEK2</i>
rs6005840	22	29101357	A	G	0.32	-0.01	9.54E-04	1.9E-38	<i>CHEK2</i>
rs713875	22	30592487	C	G	0.45	6.66E-03	8.93E-04	8.9E-14	<i>HORMAD2</i>
rs113605227	22	37908462	A	AC	0.83	-9.93E-03	1.26E-03	3.2E-15	<i>CARD10</i>
rs71324877	22	38184641	G	GAACA	0.36	7.19E-03	9.54E-04	4.7E-14	<i>TRIOBP</i>

Abbreviations: Chr, Chromosome; EA, effect allele; Freq, allele frequency of effect allele; NEA, non-effect allele; SE, Standard error; SNP, single nucleotide polymorphism; P, P value for VCDR (adjusted for VDD); VCDR, vertical cup-disc ratio; VDD, vertical disc diameter.

Appendix Table 6. 107 lead genome-wide significant independent SNPs for VCDR (adjusted for disc diameter) from UK Biobank and IGGC MTAG-analysis

SNP	Chr	Position	Nearest Gene	EA	NE	Freq	BETA (MTAG)	P (MTAG)	BETA (UKBB)	P (UKBB)	BETA(IGGC)	P (IGGC)
rs12021948	1	3049381	<i>PRDM16</i>	G	C	0.93	-0.01	1.4E-10	-0.01	4.4E-10	-6.4E-03	0.03
rs302714	1	8486131	<i>RERE</i>	A	C	0.66	-5.2E-03	8.6E-09	-3.9E-03	2.0E-05	-6.0E-03	1.9E-05
rs6690264	1	12613422	<i>VPS13D - DHRS3</i>	A	G	0.42	-6.8E-03	4.1E-15	-6.1E-03	7.2E-11	-6.5E-03	5.9E-06
rs34151819	1	68773910	<i>WLS - RPE65</i>	C	T	0.98	0.02	1.3E-10	0.02	5.4E-10	0.01	0.02
rs1925953	1	68848681	<i>WLS - RPE65</i>	A	T	0.60	-8.1E-03	4.4E-20	-7.1E-03	5.2E-14	-6.7E-03	3.1E-08
rs786914	1	89295765	<i>PKN2</i>	C	A	0.38	-5.1E-03	6.3E-09	-4.6E-03	1.6E-07	-3.8E-03	5.7E-03
rs4658101	1	92077409	<i>CDC7 - TGFB3</i>	A	G	0.19	0.02	7.2E-51	0.01	8.9E-35	0.01	1.5E-19
rs2187955	1	16953665	<i>F5</i>	G	A	0.52	-5.6E-03	1.0E-10	-3.5E-03	1.7E-05	-6.8E-03	1.0E-08
rs1417488	1	21852373	<i>TGFB2</i>	C	T	0.75	-6.0E-03	1.7E-09	-6.3E-03	2.8E-09	-3.1E-03	0.05
rs77271542	1	22767772	<i>CDC42BPA - ZNF678</i>	A	T	0.94	-0.01	3.1E-16	-0.01	6.5E-14	-0.01	2.7E-04
rs4666465	2	19338691	<i>NT5C1B - MIR4757</i>	C	T	0.64	5.5E-03	1.1E-09	4.3E-03	2.5E-06	5.9E-03	2.8E-05
rs851321	2	19420060	<i>NT5C1B - MIR4757</i>	C	T	0.01	-0.02	7.8E-09	-0.02	2.8E-06	-0.02	3.1E-04
rs2009262	2	56012214	<i>PNPT1 - EFEMP1</i>	T	C	0.79	9.7E-03	1.0E-19	9.4E-03	8.9E-18	5.7E-03	2.8E-04
rs2880192	2	11168081	<i>ACOXL</i>	A	G	0.65	7.1E-03	3.6E-15	6.4E-03	9.0E-12	5.8E-03	2.6E-05
rs74409360	2	23927627	<i>TRAF3IP1</i>	C	T	0.92	8.8E-03	1.6E-08	7.1E-03	1.8E-06	9.6E-03	1.3E-03
rs4858682	3	25066225	<i>MIR4792 - RARB</i>	C	G	0.47	-5.3E-03	7.2E-10	-5.3E-03	3.6E-09	-3.2E-03	0.02
rs56131903	3	32879823	<i>TRIM71</i>	A	T	0.68	6.9E-03	5.9E-14	6.0E-03	3.4E-10	6.1E-03	1.8E-05
rs6764184	3	58006266	<i>FLNB</i>	G	T	0.76	-6.7E-03	2.3E-11	-5.3E-03	2.3E-07	-6.9E-03	4.9E-06

rs9847377	3	66849734	LRIG1 - KBTBD8	T	C	0.52	4.9E-03	1.7E-08	3.8E-03	2.7E-05	5.0E-03	2.4E-05
rs9879264	3	88387796	C3orf38 - EPHA3	A	G	0.44	6.4E-03	1.2E-13	5.5E-03	1.2E-09	5.4E-03	9.2E-06
rs13076500	3	98943479	DCBLD2 - MIR548G	C	T	0.46	-7.4E-03	1.5E-17	-6.6E-03	4.4E-13	-5.7E-03	4.1E-06
rs2880364	3	99156869	DCBLD2 - MIR548G	G	A	0.64	-8.6E-03	7.9E-22	-7.3E-03	1.0E-14	-8.5E-03	1.8E-09
rs17398137	3	10062570 3	ABI3BP	G	A	0.82	7.6E-03	8.6E-12	7.0E-03	1.5E-09	5.4E-03	7.7E-04
rs2162137	4	55005856	GSX2 - PDGFRA	C	T	0.34	7.3E-03	1.1E-15	7.1E-03	1.8E-13	4.9E-03	6.1E-04
rs4864858	4	55089953	GSX2 - PDGFRA	T	A	0.78	8.1E-03	1.2E-14	7.5E-03	6.4E-12	6.0E-03	1.7E-04
rs1857901	4	12639627 0	FAT4	G	A	0.64	5.6E-03	2.7E-10	4.8E-03	1.9E-07	5.3E-03	2.5E-04
rs2611206	4	16657964 7	CPE - TLL1	G	A	0.85	-7.2E-03	2.9E-09	-6.3E-03	3.1E-07	-5.4E-03	1.7E-03
rs1445866	5	3621544	IRX1 - LOC1019291 53	G	A	0.67	-5.7E-03	6.7E-10	-5.1E-03	1.7E-07	-5.2E-03	5.1E-04
rs72759609	5	31952051	PDZD2	T	C	0.90	0.01	2.2E-21	0.01	1.3E-14	0.01	4.6E-09
rs10072364	5	52064349	ISL1 - PELO	T	C	0.47	-5.1E-03	2.4E-09	-3.9E-03	3.6E-06	-4.9E-03	5.0E-05
rs158653	5	55578661	ANKRD55 - LOC1024671 47	G	A	0.48	6.3E-03	2.3E-13	5.3E-03	1.1E-09	5.6E-03	2.9E-05
rs30372	5	55744230	ANKRD55 - LOC1024671 47	T	C	0.24	-7.2E-03	1.2E-12	-6.5E-03	1.2E-09	-6.2E-03	9.8E-05
rs16892523	5	60867273	ZSWIM6 - C5orf64	C	G	0.89	-7.4E-03	3.5E-08	-6.7E-03	3.0E-06	-6.2E-03	2.3E-03
rs12188947	5	82742118	XRCC4 - VCAN	A	C	0.60	5.3E-03	1.7E-09	3.6E-03	9.6E-05	7.1E-03	1.6E-07
rs17421410	5	87836307	LOC1025462 26 - LINC00461	A	G	0.93	-9.4E-03	1.0E-08	-6.4E-03	7.7E-05	-0.01	1.6E-06
rs13164684	5	12910255 0	KIAA1024L - CHSY3	A	G	0.08	1.0E-02	8.3E-10	7.4E-03	1.7E-05	0.01	6.9E-07
rs247463	5	13338433 7	VDAC1 - TCF7	T	G	0.22	7.1E-03	7.4E-12	6.5E-03	5.9E-09	6.1E-03	1.2E-04
rs60111777	5	13340291 1	VDAC1 - TCF7	C	T	0.90	0.01	4.6E-14	9.6E-03	3.2E-11	8.0E-03	1.7E-04
rs114503346	5	17219235 0	LOC1019280 93 - DUSP1	C	T	0.95	0.02	3.9E-14	0.01	4.9E-08	0.02	5.1E-09
rs17756712	6	625071	EXOC2	A	G	0.82	-7.3E-03	5.7E-11	-5.5E-03	3.5E-06	-8.4E-03	2.0E-07
rs56031192	6	1980208	GMDS	T	A	0.87	9.1E-03	6.4E-13	8.7E-03	2.5E-10	6.9E-03	4.0E-04
rs4960295	6	7205796	RREB1	G	A	0.54	-7.8E-03	2.0E-19	-6.3E-03	4.6E-13	-6.9E-03	1.7E-08
rs72852338	6	36554240	STK38 - SRSF3	A	C	0.78	-7.5E-03	8.0E-13	-7.4E-03	8.0E-12	-4.3E-03	8.8E-03
rs9367021	6	39566203	KIF6	G	A	0.59	-5.4E-03	7.5E-10	-4.0E-03	2.0E-06	-5.1E-03	2.8E-05
rs2684249	6	12239251 1	GJA1 - HSF2	T	C	0.59	6.5E-03	7.2E-14	5.6E-03	1.2E-09	6.5E-03	2.9E-06

rs10260511	7	14237240	DGKB	C	A	0.84	-9.0E-03	2.7E-14	-7.0E-03	1.6E-08	-9.6E-03	4.2E-08
rs12534570	7	19550247	FERD3L - TWISTNB	G	A	0.34	5.5E-03	2.1E-09	5.6E-03	2.7E-09	2.6E-03	0.07
rs7805378	7	28393403	CREB5	A	C	0.56	5.8E-03	2.2E-11	5.5E-03	3.3E-09	4.0E-03	1.1E-03
rs201530	7	10177738 2	CUX1	A	G	0.47	5.3E-03	6.9E-10	3.8E-03	1.5E-05	6.0E-03	7.5E-07
rs2976932	8	8255319	SGK223 - CLDN23	T	C	0.11	-0.01	8.1E-14	-9.2E-03	1.2E-10	-8.4E-03	7.1E-05
rs12543430	8	72278010	EYA1 - MSC	T	C	0.39	-5.5E-03	6.0E-10	-5.5E-03	2.0E-09	-3.7E-03	0.04
rs67120362	8	72604509	EYA1 - MSC	T	G	0.81	-6.7E-03	6.9E-10	-5.0E-03	8.8E-06	-7.7E-03	2.2E-06
rs78542921	9	18089832	SH3GL2 - ADAMTSL1	T	A	0.96	-0.01	2.0E-11	-0.01	8.1E-10	-0.01	3.0E-03
rs944801	9	22051670	CDKN2B-AS1	G	C	0.43	-0.02	4.7E-91	-0.02	7.6E-64	-0.01	3.8E-32
rs7039467	9	22056213	CDKN2B-AS1	A	G	0.69	-0.01	2.6E-54	-0.01	2.0E-37	-0.01	1.5E-21
rs10512176	9	89252706	ZCCHC6 - GAS1	T	C	0.72	-5.9E-03	8.9E-10	-5.0E-03	7.6E-07	-5.8E-03	1.0E-04
rs8176662	9	13614445 4	ABO	A	G	0.94	-0.01	4.2E-11	-0.01	6.1E-08	-0.01	6.3E-05
rs190927291	10	21437861	NEBL	C	G	0.99	-0.03	8.9E-12	-0.02	1.6E-09	-0.02	7.2E-04
rs7916697	10	69991853	ATOH7	A	G	0.24	-0.02	2.8E-67	-0.01	6.8E-35	-0.02	8.8E-43
rs72815193	10	94963391	CYP26A1 - MYOF	A	G	0.57	-7.0E-03	8.5E-16	-6.0E-03	3.3E-11	-5.8E-03	2.2E-06
rs2274224	10	96039597	PLCE1; PLCE1-AS1	G	C	0.57	-5.4E-03	4.8E-10	-3.6E-03	1.6E-04	-7.3E-03	5.3E-09
rs11197820	10	11854604 6	HSPA12A - ENO4	G	A	0.58	-6.4E-03	3.0E-13	-5.2E-03	2.4E-09	-5.8E-03	1.6E-05
rs7952509	11	31599910	ELP4	C	T	0.77	-6.4E-03	3.3E-10	-5.5E-03	6.0E-07	-7.8E-03	4.2E-05
rs2753411	11	33405001	HIPK3 - KIAA1549L	A	T	0.49	-5.2E-03	2.1E-09	-4.7E-03	3.1E-08	-3.6E-03	0.01
rs12789028	11	65326154	LTBP3 - SSSCA1-AS1	G	A	0.80	0.01	1.0E-25	8.3E-03	1.5E-13	0.01	1.1E-16
rs4944662	11	86740573	LOC1005063 68 - TMEM135	C	T	0.18	-7.6E-03	1.3E-11	-6.1E-03	5.6E-08	-6.9E-03	1.8E-05
rs7949779	11	95282879	LOC1001292 03 - FAM76B	G	C	0.61	5.6E-03	1.9E-10	4.7E-03	1.7E-07	5.4E-03	9.5E-05
rs4936099	11	13028072 5	ADAMTS8	C	A	0.41	-9.7E-03	1.6E-28	-8.5E-03	1.3E-21	-8.3E-03	2.8E-09
rs16930371	12	26392080	SSPN - ITPR2	A	G	0.82	6.2E-03	2.0E-08	5.0E-03	7.3E-06	5.8E-03	3.3E-04
rs12821310	12	47963700	PCED1B - RPAP3	C	T	0.82	7.1E-03	1.9E-10	5.0E-03	1.6E-05	8.7E-03	7.5E-08
rs324780	12	84003866	TMTC2 - SLC6A15	G	A	0.46	-0.02	1.6E-75	-0.01	5.4E-58	-0.01	5.7E-21
rs2125948	12	84120798	TMTC2 - SLC6A15	G	C	0.57	0.01	3.5E-36	0.01	1.9E-49	-6.0E-04	0.91
rs73153655	12	84133370	TMTC2 - SLC6A15	A	G	0.81	0.01	1.9E-32	0.01	3.0E-26	9.7E-03	9.2E-09
rs66716938	12	10719682 5	RIC8B	T	C	0.63	6.9E-03	7.7E-15	6.1E-03	1.1E-11	5.6E-03	7.6E-05
rs4765353	12	12466769 1	ZNF664- FAM101A	G	C	0.44	-6.7E-03	1.5E-14	-5.6E-03	1.8E-09	-6.2E-03	2.3E-07

rs9546383	13	36683268	DCLK1	T	C	0.75	-9.9E-03	2.3E-23	-8.8E-03	9.2E-18	-8.0E-03	9.4E-08
rs75764673	13	10926746 6	MYO16	A	G	0.73	6.8E-03	2.1E-12	6.9E-03	9.8E-12	4.2E-03	0.02
rs9521601	13	11076661 4	IRS2 - COL4A1	G	A	0.47	-5.6E-03	8.4E-11	-5.4E-03	7.2E-10	-3.0E-03	0.01
rs12437151	14	23372913	RBM23	G	A	0.43	7.1E-03	2.0E-16	6.4E-03	2.4E-13	4.8E-03	1.0E-04
rs2251069	14	53988050	LOC1019276 20 - MIR5580	T	C	0.57	6.7E-03	1.6E-14	5.2E-03	1.9E-09	6.2E-03	2.4E-07
rs2738265	14	54422399	BMP4	C	G	0.46	-4.8E-03	2.9E-08	-3.9E-03	9.4E-06	-4.3E-03	4.2E-04
rs7493429	14	60806759	PPM1A - C14orf39	A	C	0.70	-0.01	9.8E-36	-9.9E-03	8.3E-25	-0.01	4.0E-13
rs28761289	14	65113768	PPP1R36 - PLEKHG3	A	G	0.83	-7.5E-03	4.7E-11	-5.6E-03	1.5E-07	-6.8E-03	3.4E-05
rs984586	14	86009142	FLRT2	G	A	0.76	-7.0E-03	2.5E-12	-5.9E-03	1.2E-08	-6.5E-03	1.6E-05
rs35194812	15	71840327	THSD4	T	C	0.84	-7.5E-03	1.1E-10	-5.5E-03	4.5E-07	-7.4E-03	1.6E-05
rs893817	15	74229065	LOXL1	G	A	0.37	6.1E-03	6.3E-12	5.4E-03	2.7E-09	5.1E-03	1.9E-04
rs28612945	15	99458902	IGF1R	C	T	0.79	6.2E-03	5.7E-09	5.3E-03	5.5E-07	6.6E-03	1.5E-03
rs34222435	15	10120087 3	ASB7 - ALDH1A3	C	T	0.87	-0.01	2.4E-20	-9.7E-03	2.3E-13	-0.01	3.0E-09
rs1015438	16	51177517	SALL1	G	A	0.81	6.7E-03	1.4E-09	6.1E-03	1.8E-07	5.2E-03	1.4E-03
rs62038917	16	51425823	SALL1 - LOC1019273 64	G	T	0.78	0.01	1.2E-27	9.9E-03	3.9E-20	9.9E-03	9.3E-10
rs8053277	16	51469726	SALL1 - LOC1019273 64	T	C	0.30	0.01	6.1E-38	9.9E-03	7.3E-25	0.01	6.1E-16
rs2720429	16	51571885	SALL1 - LOC1019273 64	G	A	0.77	-9.9E-03	4.5E-22	-0.01	2.2E-19	-6.0E-03	7.3E-05
rs1728367	16	86386888	LINC00917 - FENDRR	C	T	0.09	0.01	9.7E-16	0.01	6.9E-12	9.7E-03	1.2E-05
rs7187191	16	86511858	FENDRR	T	G	0.18	8.2E-03	1.7E-13	7.0E-03	3.0E-09	7.5E-03	2.8E-06
rs847687	17	48229441	PPP1R9B - SGCA	T	G	0.52	6.0E-03	4.2E-12	5.0E-03	3.0E-08	5.8E-03	7.1E-06
rs2204928	17	59262040	BCAS3	C	T	0.21	-7.8E-03	2.2E-13	-6.5E-03	2.3E-09	-6.8E-03	1.0E-05
rs4790993	17	65035215	CACNG4 - CACNG1	T	C	0.70	-5.5E-03	6.9E-09	-5.3E-03	1.4E-07	-3.7E-03	8.5E-03
rs123698	19	807442	PTBP1	G	C	0.40	6.5E-03	9.9E-14	5.6E-03	3.8E-10	8.5E-03	2.3E-05
rs8102936	19	32027330	TSHZ3 - THEG5	G	A	0.66	6.1E-03	1.9E-11	6.5E-03	4.0E-10	3.9E-03	5.7E-03
rs6140009	20	6473054	CASC20	C	T	0.62	0.01	3.2E-48	0.01	1.5E-42	8.3E-03	1.3E-09
rs1819154	20	6528627	CASC20 - BMP2	C	T	0.02	-0.02	3.1E-10	-0.02	7.1E-08	-0.02	6.1E-04
rs6120005	20	31448849	MAPRE1 - SUN5	G	A	0.84	8.5E-03	3.8E-13	7.0E-03	2.1E-09	7.4E-03	1.8E-05
rs143032062	22	28709058	TTC28	T	C	0.98	0.03	1.3E-17	0.03	2.1E-14	0.04	4.2E-05
rs5752776	22	29108229	CHEK2	A	G	0.33	-0.01	2.3E-54	-0.01	3.9E-38	-0.01	5.7E-21
rs1003342	22	30570022	HORMAD2	A	G	0.45	7.7E-03	7.8E-19	6.4E-03	5.9E-13	6.6E-03	4.3E-08
rs6000759	22	37911190	CARD10	T	G	0.78	-9.6E-03	2.8E-20	-8.6E-03	1.0E-14	-8.5E-03	5.2E-08
rs5750494	22	38176979	TRIOBP - H1FO	T	G	0.37	7.3E-03	2.0E-16	6.8E-03	4.4E-13	5.8E-03	4.4E-05

Abbreviations: Chr, Chromosome; EA, effect allele; Freq, allele frequency of effect allele; NEA, non-effect allele; SE, Standard error; P, p value; SNP, single nucleotide polymorphism.

UKBB, UK biobank data; IGGC, International Glaucoma Genetic Consortium; MTAG, meta-analysis results of UKBB and IGGC datasets.

Chromosomal position is based on the NCBI RefSeq hg19 human genome reference assembly.

Appendix Table 7. 114 statistically independent SNPs for glaucoma risk from MTAG

GWAS

SNP	Chr	Position	Nearest Gene	EA	NEA	Freq	Glaucoma MTAG		Glaucoma ANZRAG		Glaucoma NEIGHBORHO OD		Glaucoma meta (ANZRAG + NEIGHBORHO OD)		Glaucoma meta (ANZRAG + NEIGHBORHO OD + UKBB)		Annotation*
							OR	P value	OR	P value	OR	P value	OR	P value	OR	P value	
rs274760	1	3657357	COL8A2	C	A	0.94	1.13	2.0E-08	1.29	9.8E-04	1.03	0.70	1.16	7.8E-03	1.12	1.5E-04	Known
rs10796912	1	3809159	RSPO1	G	A	0.56	0.94	1.1E-08	0.97	0.37	0.86	6.1E-06	0.92	1.8E-04	0.94	1.9E-05	Bonferroni
rs12068500	1	8661415	COL24A1	G	T	0.91	0.88	7.3E-13	0.93	0.47	0.93	0.26	0.93	0.18	0.93	4.8E-03	-
rs12139208	1	8821301	LOC100505768	T	C	0.38	1.08	2.2E-12	1.05	0.16	1.12	1.4E-03	1.08	1.3E-03	1.09	2.8E-09	Known
rs4658101	1	9207740	HSP90B3P	A	G	0.19	1.10	1.2E-13	1.01	0.78	1.09	0.04	1.05	0.11	1.05	7.2E-03	-
rs12045227	1	1131282	ST7L	G	A	0.80	0.92	5.3E-10	0.90	7.9E-03	0.98	0.65	0.94	0.02	0.96	8.3E-03	Nominal
rs10918274	1	1657144	TMCO1	T	C	0.12	1.37	5.6E-92	1.40	1.4E-13	1.39	3.3E-12	1.40	1.4E-24	1.38	1.4E-65	Known
rs73111535	1	2192228	LOC643723	C	A	0.93	1.17	3.4E-13	1.00	0.96	1.31	3.5E-04	1.12	0.02	1.15	6.2E-07	Known
rs6741499	2	2836943	BRE	C	T	0.74	0.92	3.8E-12	0.90	2.3E-03	0.99	0.82	0.94	0.02	0.94	5.8E-05	Known
rs113542380	2	4346481	THADA	G	A	0.92	0.87	9.7E-12	0.85	8.8E-03	0.91	0.12	0.88	2.9E-03	0.89	6.5E-06	Known
rs163524	2	4515755	SIX3	C	A	0.82	0.92	5.7E-09	0.94	0.13	1.00	0.95	0.96	0.24	0.94	1.7E-04	Known
rs74259971	2	5465920	SPTBN1	T	C	0.83	1.09	4.0E-10	0.96	0.38	1.09	0.05	1.02	0.49	1.08	6.8E-05	-
rs1084524	2	5593239	PNPT1	G	A	0.15	1.09	2.1E-09	1.06	0.32	0.98	0.56	1.00	0.95	1.07	1.9E-04	Known
rs13009933	2	6657439	MIR4778	C	T	0.64	0.93	1.3E-10	0.89	0.07	0.95	0.15	0.93	0.03	0.93	1.1E-05	Nominal
rs6732795	2	6941151	ANTXR1	A	C	0.61	0.93	7.2E-12	0.92	7.3E-03	0.96	0.25	0.94	5.3E-03	0.93	5.6E-07	Known
rs56405342	2	1016081	NPAS2	C	T	0.86	1.09	7.2E-09	1.06	0.25	1.03	0.54	1.05	0.20	1.05	0.01	-

rs1579050	2	153364527	FMNL2	A	G	3	0.89	7.5E-26	0.90	1.1E-03	0.96	0.19	0.92	9.0E-04	0.93	7.4E-08	Known
rs62172701	2	205998191	PARD3B	A	C	8	1.07	2.7E-08	1.05	0.20	0.96	0.35	1.01	0.75	1.04	0.02	Known
rs62188040	2	213723345	MIR4776-1	C	A	9	1.11	2.1E-09	1.08	0.16	1.03	0.64	1.05	0.18	1.12	1.6E-07	Known
rs1286771	3	25581512	RARB	T	C	7	1.11	4.9E-11	1.05	0.31	1.12	0.03	1.08	0.03	1.11	1.7E-06	Nominal
rs2526385	3	50181135	SEMA3F	T	G	0	1.08	1.1E-09	1.05	0.20	1.08	0.07	1.07	0.03	1.10	1.6E-08	Nominal
rs1867409	3	66860178	KBTBD8	G	A	0	1.10	1.1E-16	1.11	3.7E-03	1.06	0.12	1.09	1.3E-03	1.07	2.6E-06	Known
rs66500121	3	85137683	CADM2	A	G	1	1.08	2.6E-13	1.10	5.0E-03	1.09	0.01	1.09	1.6E-04	1.11	2.3E-14	Known
rs893830	3	124433550	KALRN	G	A	4	0.92	7.8E-09	0.94	0.17	0.97	0.59	0.96	0.16	0.93	2.1E-04	-
rs11710845	3	150065280	TSC2D2	C	T	4	1.07	2.7E-09	1.08	0.04	1.08	0.07	1.08	6.2E-03	1.10	4.6E-09	Known
rs9816799	3	169222771	MECOM	T	C	6	0.93	4.5E-11	0.94	0.08	0.91	6.8E-03	0.93	1.7E-03	0.93	1.8E-07	Bonferoni
rs9853115	3	186131600	LOC253573	T	A	0	1.15	1.1E-39	1.11	2.9E-03	1.17	1.4E-05	1.13	2.4E-07	1.12	1.2E-15	Known
rs4420855	3	188057207	LPP	A	G	0	1.08	3.8E-10	1.05	0.21	1.03	0.37	1.04	0.12	1.08	7.6E-07	Known
rs28795989	4	78915457891545	AFAP1	A	G	0	1.16	5.8E-42	1.16	2.8E-06	1.15	8.3E-05	1.16	7.7E-10	1.15	2.8E-25	Known
rs6838291	4	79178067917806	AFAP1	C	T	2	0.84	2.2E-36	0.87	1.0E-03	0.95	0.24	0.91	1.1E-03	0.87	3.2E-16	Known
rs447324	4	177430644	VEGFC	A	G	2	1.07	3.4E-09	1.03	0.43	1.01	0.71	1.02	0.40	1.05	7.1E-04	-
rs76325372	5	14837332	ANKH	A	C	3	1.11	4.3E-18	1.12	1.8E-03	1.04	0.31	1.08	2.7E-03	1.10	2.7E-10	Known
rs4703855	5	716938959	PTCD2	C	T	0	1.08	6.1E-11	0.99	0.79	1.00	0.98	0.99	0.83	1.05	3.9E-04	-
rs73220190	5	108053907	FER	T	C	3	1.13	1.9E-09	1.02	0.77	1.06	0.39	1.04	0.42	1.10	4.8E-04	-
rs11567976	5	137654218	CDC25C	C	T	3	0.94	1.7E-08	0.95	0.13	0.98	0.60	0.97	0.14	0.96	2.1E-03	-
rs1347141	5	147068784	JAKMIP2	A	C	7	1.06	2.9E-08	0.97	0.40	1.09	0.02	1.03	0.29	1.05	6.7E-04	-
rs57111852	6	606263606263	EXOC2	G	A	6	0.87	6.2E-22	0.93	0.12	0.83	1.0E-04	0.88	1.5E-04	0.87	8.5E-13	Known
rs2745572	6	15483691548369	FOXC1	A	G	7	1.14	1.2E-30	1.16	2.4E-05	1.25	2.4E-09	1.20	6.4E-13	1.15	6.4E-21	Known
rs7225856	6	17758631775863	GMDS	G	A	2	1.08	1.1E-11	1.04	0.27	1.04	0.31	1.04	0.13	1.05	6.3E-04	Known
rs6914444	6	19834401983440	GMDS	T	C	6	1.16	4.1E-21	1.23	4.2E-05	1.20	4.6E-04	1.21	6.3E-08	1.14	1.0E-10	Known
rs2145826	6	44763781	SUPT3H	G	A	3	1.07	1.3E-11	0.98	0.44	1.06	0.11	1.01	0.61	1.06	4.4E-05	-
rs2439042	6	51446615144661	PKHD1	T	G	0	1.15	7.6E-16	NA	NA	NA	NA	NA	NA	1.14	1.0E-05	Known

rs9494457	6	136474794	PDE7B	T	A	2	0.6	1.10	6.9E-17	1.13	4.2E-04	1.10	6.3E-03	1.12	7.6E-06	1.09	7.4E-10	Known
rs4709210	6	158971956	TMEM181	T	A	2	0.7	1.07	1.3E-09	1.06	0.31	1.06	0.11	1.06	0.06	1.08	1.4E-06	Nominal
rs2935072	6	170460912	LOC154449	C	T	8	0.8	0.89	2.4E-14	0.90	0.03	0.97	0.51	0.93	0.04	0.89	1.1E-09	Known
rs2526101	7	11677452	THSD7A	A	G	5	0.5	0.93	1.3E-10	0.89	5.2E-04	0.93	0.03	0.91	4.4E-05	0.90	3.7E-13	Known
rs1362227	7	33511631	BBS9	A	C	3	0.4	1.07	3.2E-09	1.02	0.52	1.05	0.17	1.04	0.16	1.06	8.0E-05	-
rs10435033	7	39054837	POU6F2	G	A	6	0.6	1.07	8.7E-09	1.12	7.8E-04	1.14	9.5E-04	1.13	2.2E-06	1.07	1.6E-06	Bonferoni
rs3277127	7	80836057	SEMA3C	C	G	7	0.5	1.09	1.7E-14	1.01	0.70	1.05	0.17	1.03	0.23	1.08	1.7E-07	Known
rs7799028	7	103346226	RELN	G	T	3	0.7	0.94	1.4E-08	1.01	0.76	0.96	0.27	0.99	0.59	0.95	1.4E-03	-
rs2896175	7	115811436	TES	A	C	2	0.6	1.10	1.7E-18	1.03	0.32	1.09	0.02	1.06	0.02	1.08	7.4E-08	Known
rs59454355	7	116156023	CAV2	C	G	3	0.7	0.84	4.4E-49	0.90	0.07	0.86	4.1E-05	0.87	8.9E-06	0.89	6.5E-14	Known
rs2188836	7	117635382	CTTNBP2	C	T	8	0.5	0.92	5.8E-15	0.93	0.03	0.91	4.1E-03	0.92	3.6E-04	0.92	5.1E-10	Known
rs4128399	7	151498679	PRKAG2	T	C	3	0.8	0.92	1.5E-09	0.95	0.35	0.99	0.89	0.97	0.44	0.91	2.1E-07	Known
rs4412362	8	78930579	PKIA	C	T	9	0.3	0.94	9.5E-10	1.00	0.89	0.99	0.87	1.00	0.83	0.96	6.1E-03	-
rs2881425	8	107836447	ABRA	A	G	8	0.6	1.07	1.6E-09	1.01	0.85	0.97	0.40	0.98	0.55	1.01	0.38	-
rs2022945	8	108251139	ANGPT1	A	G	3	0.1	0.83	1.1E-29	0.85	1.4E-03	0.86	4.9E-03	0.85	1.8E-05	0.85	1.5E-14	Known
rs1283696	8	108441531	ANGPT1	T	A	1	0.9	1.14	2.7E-11	1.04	0.46	1.01	0.85	1.03	0.50	1.10	1.1E-04	Known
rs17339357	8	124600906	FBXO32	T	A	3	0.9	0.86	9.2E-14	0.83	2.6E-03	0.87	0.04	0.85	3.0E-04	0.87	6.2E-08	Bonferoni
rs9448019	9	22051670	CDKN2B-AS1	G	C	3	0.4	0.87	2.0E-38	0.72	3.8E-22	0.70	6.7E-24	0.71	9.9E-45	0.80	6.8E-56	Known
rs10869665	9	78518999	PCSK5	C	T	0	0.7	1.08	2.7E-11	1.05	0.19	1.06	0.11	1.05	0.04	1.07	4.8E-06	Known
rs2472493	9	107695848	ABCA1	G	A	5	0.4	1.19	7.8E-59	1.32	2.9E-16	1.20	1.2E-07	1.26	6.1E-22	1.19	8.5E-38	Known
rs2275241	9	129370576	LMX1B	G	A	3	0.6	1.16	3.2E-40	1.10	4.0E-03	1.17	2.6E-05	1.13	5.9E-07	1.14	5.7E-19	Known
rs62580791	9	129757851	RALGPS1	A	C	4	0.5	0.93	1.1E-12	NA	NA	NA	NA	NA	NA	0.94	3.2E-04	Known
rs12216891	9	136127366	ABO	C	T	4	0.9	0.85	6.7E-14	1.00	1.00	0.94	0.32	0.97	0.49	0.92	4.2E-03	-
rs11008626	1	320372300	ARHGAP12	T	C	1	0.7	0.94	6.6E-09	1.00	0.93	0.94	0.08	0.97	0.25	0.94	7.3E-05	-
rs7089636	1	60283309	BICC1	T	G	4	0.5	0.92	9.8E-14	0.90	0.07	0.85	2.6E-06	0.87	6.8E-07	0.91	1.2E-11	Bonferoni
rs1616405	1	78581153	KCNMA1	A	G	9	0.4	1.06	4.0E-08	1.02	0.68	1.01	0.77	1.01	0.64	1.05	8.9E-04	-

rs12778014	10	94950273	CYP26A1	G	A	6	0.6	1.07	1.0E-08	1.11	2.5E-03	1.03	0.40	1.07	4.8E-03	1.07	3.0E-06	Nominal
rs2274224	10	96039597	PLCE1	G	C	7	0.5	0.93	2.7E-13	0.90	6.8E-04	0.98	0.54	0.93	3.3E-03	0.93	8.2E-08	Known
rs4141194	11	17011176	PLEKH A7	C	A	1	0.7	0.91	5.2E-15	0.98	0.48	0.95	0.22	0.97	0.18	0.94	1.2E-05	Known
rs12280392	11	28403473	METTL15	T	G	7	0.7	0.93	6.5E-09	0.94	0.12	0.96	0.33	0.95	0.07	0.95	7.7E-04	Nominal
rs56319620	11	47998922	PTPRJ	C	A	0	0.8	0.89	4.5E-21	0.95	0.25	0.92	0.04	0.94	0.03	0.93	6.3E-06	Known
rs4434990	11	51517073	OR4C46	G	A	7	0.8	0.91	2.8E-10	0.90	0.03	1.05	0.40	0.95	0.22	0.93	4.4E-04	Known
rs11229165	11	55112643	OR4A16	T	C	7	0.8	0.91	4.1E-10	0.90	0.03	1.01	0.80	0.95	0.13	0.93	1.9E-04	Known
rs4102217	11	65263895	MALAT1	G	C	0	0.8	1.11	7.8E-16	1.08	0.05	1.08	0.08	1.08	9.1E-03	1.10	1.3E-07	Nominal
rs11234741	11	86401678	ME3	A	G	0	0.8	0.90	2.7E-16	0.91	0.02	0.88	3.1E-03	0.90	1.6E-04	0.91	1.3E-08	Known
rs1126809	11	89017961	TYR	G	A	9	0.6	1.07	1.1E-08	1.10	6.0E-03	0.94	0.09	1.02	0.38	1.06	1.8E-04	-
rs58073046	11	120248493	ARHGE F12	A	G	8	0.8	0.86	1.1E-21	0.76	4.4E-08	0.87	6.2E-03	0.81	3.3E-09	0.85	1.3E-14	Known
rs7924522	11	128380742	ETS1	C	A	4	0.3	0.91	6.9E-17	0.89	7.3E-04	0.89	7.4E-04	0.89	1.6E-06	0.91	4.7E-11	Known
rs4936100	11	130287469	ADAMTS8	A	G	1	0.2	0.92	1.5E-09	0.88	2.9E-03	0.95	0.26	0.92	3.2E-03	0.94	1.6E-04	Nominal
rs3247622	12	83986665	TMTC2	A	G	6	0.4	0.93	2.9E-12	0.91	6.0E-03	0.92	0.02	0.92	2.6E-04	0.94	3.1E-05	Known
rs3184504	12	111884608	SH2B3	T	C	8	0.4	0.94	5.6E-09	0.92	0.01	0.86	6.2E-06	0.89	1.0E-06	0.94	2.9E-05	Known
rs9530143	13	73639371	KLF5	G	A	8	0.6	1.07	1.4E-08	1.09	9.0E-03	1.07	0.05	1.08	1.1E-03	1.08	1.2E-07	Bonferoni
rs9544024	13	76258720	LMO7	A	G	0	0.5	0.93	1.2E-12	0.87	2.5E-05	0.89	4.4E-04	0.88	3.4E-08	0.93	9.7E-08	Known
rs56152426	13	110893841	COL4A1	A	G	2	0.8	1.09	5.7E-10	1.04	0.42	1.18	3.1E-03	1.09	0.01	1.09	3.0E-06	Known
rs72669675	14	34715774	SPTSSA	A	T	3	0.6	0.93	1.0E-11	0.90	0.06	0.96	0.28	0.94	0.05	0.94	6.5E-06	Nominal
rs61755579	14	50655307	SOS2	C	T	7	0.9	1.18	4.5E-08	1.31	0.03	NA	NA	1.31	0.03	1.11	0.02	Nominal
rs8009633	14	53386834	FERMT2	G	C	7	0.7	1.10	1.5E-13	1.11	0.01	1.06	0.17	1.09	5.0E-03	1.06	2.1E-04	Nominal
rs35155027	14	61095174	SIX1	G	C	9	0.3	1.09	6.2E-17	1.29	3.1E-15	1.30	2.7E-14	1.30	2.5E-28	1.18	6.9E-33	Known
rs73294447	14	74929721	NPC2	T	C	9	0.9	1.36	1.9E-10	1.18	0.25	NA	NA	1.18	0.24	1.33	7.7E-05	Known
rs12913832	15	28365618	HERC2	A	G	1	0.2	1.08	7.1E-09	1.07	0.12	1.06	0.14	1.07	0.03	1.07	1.0E-04	Nominal
rs4601984	15	57060530	ZNF280D	G	T	4	0.6	0.93	2.4E-10	1.00	0.89	0.93	0.05	0.97	0.16	0.96	1.1E-03	Known
rs2249195	15	61958029	VPS13C	A	C	7	0.5	1.08	1.6E-13	1.06	0.09	1.11	2.0E-03	1.08	7.7E-04	1.06	1.6E-05	Bonferoni

rs11859314	16	51455794	SALL1	G	A	1	0.93	2.2E-10	0.94	0.09	0.97	0.51	0.96	0.08	0.94	7.3E-05	Nominal
rs74984957	16	65073799	CDH11	G	A	4	1.07	6.2E-11	1.12	0.04	1.15	9.1E-05	1.14	9.9E-06	1.09	1.3E-08	Known
rs75265191	16	77577844	ADAMTS18	G	A	3	0.86	1.4E-14	0.89	0.05	0.95	0.41	0.91	0.04	0.89	5.8E-06	Known
rs3743861	16	89818340	FANCA	G	C	8	1.07	4.0E-10	1.04	0.29	1.03	0.36	1.03	0.16	1.04	5.3E-03	-
rs1563966	17	2095954	SMG6	G	A	6	1.06	2.1E-08	1.06	0.10	1.08	0.04	1.07	8.7E-03	1.06	3.4E-05	Nominal
rs8064739	17	9842264	GAS7	A	C	9	0.92	1.2E-15	0.99	0.66	0.95	0.10	0.97	0.15	0.93	4.6E-07	Known
rs9913911	17	10031183	GAS7	A	G	2	1.19	1.1E-58	1.18	1.6E-06	1.24	5.0E-09	1.20	5.6E-14	1.18	6.8E-30	Known
rs36006455	17	47320418	FLJ40194	T	C	2	1.07	5.3E-09	1.02	0.69	0.95	0.32	0.99	0.74	1.04	0.02	-
rs3785856	17	59391379	BCAS3	A	G	0	0.91	3.7E-13	0.98	0.64	0.89	6.6E-03	0.94	0.03	0.93	5.1E-06	Known
rs3519739	17	795457	PTBP1	A	G	6	1.07	4.3E-09	1.07	0.11	0.96	0.41	1.03	0.43	1.03	0.06	-
rs4406779	17	11285390	KANK2	G	A	8	0.93	2.8E-10	0.95	0.11	0.97	0.37	0.96	0.07	0.94	3.5E-05	Nominal
rs6140009	20	64730540	CASC2	C	T	2	1.09	2.4E-14	1.07	0.05	1.17	5.9E-06	1.12	6.1E-06	1.09	1.7E-09	Bonferroni
rs34952318	20	11177055	LOC339593	G	A	5	1.18	1.4E-11	1.30	0.04	NA	NA	1.30	0.04	1.19	1.5E-05	Known
rs6065171	20	38538760	LOC339568	T	C	9	1.07	9.8E-10	1.07	0.05	1.03	0.42	1.05	0.04	1.06	6.8E-05	Known
rs7273775	20	49061320	PTPN1	C	T	1	1.07	9.4E-10	1.10	0.10	1.12	2.2E-03	1.11	5.4E-04	1.07	1.8E-05	Bonferroni
rs73148965	22	19872935	TXNRD2	G	A	5	1.11	9.4E-12	1.17	1.3E-03	1.27	4.1E-06	1.21	3.5E-08	1.15	9.5E-13	Known
rs7387222	22	29130012	CHEK2	T	C	0	0.91	2.3E-15	0.93	0.03	0.92	0.03	0.92	2.3E-03	0.91	3.3E-09	Nominal
rs9608740	22	29620325	EMID1	A	C	1	0.90	3.0E-14	0.92	0.06	NA	NA	0.92	0.06	0.93	1.9E-04	Known
rs7564812	22	33105227	SYN3	A	G	5	1.16	3.6E-09	1.04	0.61	NA	NA	1.04	0.60	1.10	0.01	-
rs4821712	22	38183847	TRIOBP	C	T	7	1.11	1.3E-20	1.05	0.38	1.02	0.57	1.03	0.34	1.06	1.6E-04	Known

Abbreviations: Chr, Chromosome; EA, effect allele; Freq, allele frequency of effect allele; MTAG, multiple trait analysis of GWAS; NEA, non-effect allele; SNP, single nucleotide polymorphism; OR, odds ratio; P, P value for VCDR (adjusted for VDD); VCDR, vertical cup-disc ratio; VDD, vertical disc diameter.

UKBB, UK biobank data; ANZRAG, the Australian & New Zealand Registry of Advanced Glaucoma study; NEIGHBORHOOD, the National Eye Institute Glaucoma Human Genetics Collaboration Heritable Overall Operational Database.

* Results are highlighted: those which are associated with glaucoma alone or previous reported loci are in bold italics (we also set IOP SNPs as "known" glaucoma loci if they were replicated in glaucoma

GWAS after correction for multiple comparisons in our previous study.(MacGregor et al. 2018)), those replicated in ANZRAG and NEIGHBORHOOD after Bonferroni correction ($P < 0.05/49 = 0.001$, one-sided test) are in bold, those replicated at a nominal significance level ($P < 0.05$) are in italics, with the remainder in normal font. “-”, remain to be confirmed.

Chromosomal position is based on the NCBI RefSeq hg19 human genome reference assembly.

Appendix Table 8. Normalised RNA-Seq expression of the identified genes in human ocular tissue in counts per million

Gene	Corneal epithelium	Corneal stroma	Descemet's membrane	Ciliary body	Trabecular meshwork	Iris	Optic nerve head	Optic nerve	Retina	Sclera
ADAMTS14	1631.2	349.8	1.3	155.3	226.5	754.6	279.2	874.8	11.7	12.6
ADAMTS L3	280.7	199.4	32.9	979.3	3004.3	1021.8	916.3	361.2	282.8	2883.9
ALX3	1.7	5.6	10.9	227.8	736.3	94.8	151.8	29.8	0.7	178.4
ARHGAP21	5026.4	4574.6	3598.4	998.5	2601.3	1551.8	9335.3	23183.3	11946.5	967.0
ARHGEF10	186.4	80.8	52.9	67.5	95.8	40.0	194.0	350.7	191.7	44.7
ATOH8	14.3	1690.0	1447.9	2475.5	2403.5	681.6	5757.7	10519.2	1550.2	834.7
BCAS3	2099.0	1282.6	595.4	2019.5	1670.8	750.8	2322.0	2086.0	2915.5	372.4
BMP3	135.6	424.4	1370.9	75.3	1578.2	6.8	381.7	18.3	216.8	2417.1
BMP4	115.7	1315.0	28.4	4654.7	2662.2	689.2	576.2	216.7	132.2	1215.4
C4orf32	547.7	471.8	259.7	709.3	988.5	423.2	996.8	920.3	1797.8	369.9
C8orf48	86.4	54.8	0.6	13.0	36.0	1.8	52.8	15.7	104.7	8.4
CADPS	288.1	256.4	11.3	313.8	134.5	534.6	4195.8	410.0	15298.3	56.4
CDH2	41.3	138.6	6803.6	2230.3	1578.5	701.2	5466.0	3416.2	5501.2	365.1
CENPW	80.7	59.6	11.1	61.8	56.7	35.4	27.0	36.7	20.7	20.4
CLDN1	51658.6	6858.0	17.3	224.2	81.2	222.8	2114.5	211.7	137.8	46.3
COL13A1	6.9	75.4	0.0	90.7	300.0	90.8	283.2	174.3	8.8	60.9
CYP26A1	3933.7	336.6	2.0	92.7	102.3	15.8	176.7	14.8	185.7	40.9
DAAM1	8586.9	1137.4	74.9	333.7	651.2	388.6	3088.2	2577.8	4489.5	83.6
DCDC1	14964.7	1276.2	23.4	147.7	50.8	1710.2	406.3	104.7	546.8	23.6
DHRS7	1149.1	2260.2	522.0	2467.7	1498.7	755.2	2725.2	2010.7	1964.0	655.0
EFNA3	189.2	214.4	198.7	115.7	46.3	15.2	142.3	65.3	322.5	8.6
FAM49A	725.2	123.6	0.4	642.8	891.3	344.6	1418.7	1235.7	1126.2	203.1
FBLN5	107.4	19286.4	542.1	4368.5	12943.0	467.8	3964.3	2278.0	97.2	6736.7
FILIP1L	494.1	71.4	593.3	137.3	1804.5	873.4	876.5	474.2	741.8	258.4
FOXI3	16.1	2.0	5.9	1865.8	11.3	59.6	55.3	17.7	203.3	3.0
FOXP1	4743.6	2756.6	1515.7	2067.7	3803.8	1953.4	2153.0	2046.5	1102.2	2421.3
FST	816.4	115.0	6.1	2831.7	179.8	199.6	501.2	157.0	39.8	265.6
GMDS	1447.4	725.0	1233.4	894.2	1159.7	199.8	624.7	394.5	127.3	739.7

GSX2	0.4	0.2	0.1	246.8	9.0	201.2	3.5	2.8	3.0	0.6
HSF2	1627.1	893.8	484.3	563.3	916.7	404.2	921.2	663.2	2073.0	209.0
KAT2B	11547.3	1555.6	1589.9	2606.3	4370.3	1018.6	3575.5	4500.2	3956.7	745.0
KCNQ5	39.9	71.8	92.3	382.8	1280.3	545.0	71.5	11.3	42.5	55.9
KIF6	18.1	48.6	72.1	89.2	53.5	32.6	338.5	1003.5	295.8	46.6
LEKR1	70.2	35.4	29.7	28.7	27.5	13.6	57.2	27.8	101.7	5.9
LIMCH1	136.0	1047.2	917.3	3129.3	3799.5	879.6	2986.5	10286.8	5522.2	2439.6
LINC004 61	5.0	40.2	17.9	16.5	90.7	3.8	1012.0	140.0	3100.3	2.0
LPHN2	1599.8	3107.8	1805.9	1511.3	3274.7	538.8	1792.2	333.2	4610.8	573.7
LRCH1	1643.4	1730.6	602.3	606.2	1298.3	514.6	1065.2	389.2	1372.0	321.9
LRP2	13.6	114.0	3.4	5974.5	59.2	2805.6	6722.3	6938.7	8366.3	1.9
LRR1Q1	19.7	70.4	32.0	15.8	69.5	35.6	332.3	702.0	686.2	55.9
LTBP1	5625.8	2287.0	21.0	3773.7	1929.5	2356.2	2063.5	2326.0	92.2	792.3
MACF1	31262.1	12607.8	9587.6	8226.7	14565.8	5396.6	17735.2	17826.5	28506.8	3577.9
MAP2K6	237.9	1753.4	460.1	73.2	210.7	63.2	803.3	212.3	196.7	52.7
43527	163.6	651.2	2600.4	1010.0	1132.7	187.6	726.0	156.8	1124.2	231.9
MARK3	10109.8	3999.6	1758.4	2264.8	3988.8	1407.2	3963.0	3820.8	5653.7	759.1
MSC	2.8	25.0	1.0	316.7	183.8	17.8	182.3	34.3	2.0	135.4
MYEOV	2568.2	197.2	0.4	6.3	4.7	4.2	16.3	2.3	13.8	2.4
NAV1	2536.0	7058.8	9029.0	2763.0	4438.3	2790.6	3100.3	5293.3	3227.0	1169.3
NAV3	205.8	84.2	14.3	224.5	260.8	360.4	435.7	838.7	1026.7	346.6
NCKAP5	1429.8	208.0	14.0	184.3	141.3	52.6	439.2	770.0	1099.7	34.4
NPLOC4	6327.6	5351.4	2877.1	8973.2	7902.8	3082.4	5885.8	4969.3	5065.8	1298.0
PAX8	164.9	81.6	53.0	360.7	105.3	63.6	11645.5	8306.7	335.2	26.9
PDZD2	2176.0	1335.0	2458.7	2649.0	2443.3	381.0	1782.0	1346.8	6258.7	866.7
PKIA	18.0	74.4	1.3	690.2	717.0	373.0	1833.0	1596.8	3270.0	10.3
PLEKHG 3	11476.2	2855.6	3897.4	7311.5	6548.3	4430.2	3060.8	8734.5	2499.3	628.3
PRSS56	0.0	24.6	6.1	0.8	2.5	2.6	206.5	1.0	2795.3	0.0
PSCA	13248.4	887.6	43.3	12.3	12.3	17.8	12.7	17.2	2.2	16.6
PTCH1	2279.1	1379.0	731.1	3562.3	1187.5	367.2	2434.3	1020.0	4071.5	452.1
RAPGEF 1	4456.2	2762.2	5146.9	8126.5	13220.3	3687.2	6465.0	2887.0	5820.5	1075.7
RBFOX1	410.9	183.2	371.4	384.3	130.0	283.6	383.5	67.8	1192.0	2.0
RHPN2	1351.7	138.8	40.1	792.0	139.2	93.8	2161.5	715.8	3073.8	17.0
RSPO3	1.4	25.2	2.4	22.0	164.2	126.2	199.5	17.3	23.5	267.4
SESTD1	208.3	1477.8	326.0	1273.5	2813.2	1200.8	2759.0	4869.5	2580.7	1896.4
SIX1	6.3	0.2	1.0	128.0	275.8	4.0	435.8	170.5	11.8	64.1
SIX3	22.2	74.6	18.9	6416.5	229.7	638.2	7151.8	4841.7	5238.5	40.9
STRA6	1702.3	3437.0	15.7	13481.5	237.2	1905.0	1199.7	379.0	40.7	37.7
SUFU	2662.6	1237.6	1021.6	2421.3	3171.8	758.4	1418.7	848.3	1103.0	420.4
TANC2	2694.3	1240.6	116.3	952.2	925.5	533.0	1238.8	1829.0	2276.7	459.1
TEX41	6.9	21.2	3.1	82.0	85.2	119.8	81.7	6.5	12.7	34.3
TMEM11 9	36.6	177.8	13.0	1378.8	3826.7	209.4	917.8	554.0	37.3	2915.7
TSPAN11	32.9	85.8	27.4	1028.3	1920.8	28.2	708.0	237.0	304.3	555.4
TSPAN9	3608.4	3507.0	1396.0	6301.5	4548.0	2837.0	3674.2	389.5	5132.0	564.1
TYR	44.0	67.4	8.3	12873.7	1094.8	6201.6	684.5	2.0	3.2	58.1

UBE2L3	3795.9	3020.0	3325.0	5628.8	3678.7	1255.2	4792.0	5573.0	3097.8	1117.6
VANGL1	980.1	396.8	8.3	774.7	310.5	342.4	532.3	512.3	334.0	127.0
VCAN	30.2	210.4	40.6	231.8	1010.8	179.8	1309.8	1757.7	69.7	761.9
WNT7B	7785.8	1072.0	6.1	5.3	14.8	9.4	380.2	67.0	20.7	7.4
ZCCHC6	3205.6	799.2	201.0	231.0	701.3	348.8	692.8	623.8	795.3	311.7
ZNF385D	118.9	238.6	398.3	1186.2	4177.8	401.6	737.8	206.3	2005.2	233.6

Appendix Table 9. Differential expression of the identified disc diameter genes in human ocular tissue.

SNP	Gene	GWAS beta	GWAS P-value	logFC vs all ocular tissue	P-value*
rs74891390	TSPAN9	0.03	4.70E-15	16.86479	3.91E-20
rs34607745	HSF2	-0.02	3.40E-16	9.964448	1.27E-14
rs7717697	VCAN	0.01	2.40E-12	19.35751	9.49E-11
rs13417287	LTBP1	-0.01	5.70E-10	25.20096	7.97E-26
rs116072427	LTBP1	-0.02	4.50E-11	25.20096	7.97E-26
rs28603236	LIMCH1	0.01	4.30E-08	18.47239	1.47E-19
rs698153	NAV3	-0.03	4.70E-11	15.71945	5.25E-15
rs4658101	TGFBR3	0.06	2.80E-160	13.2145	2.27E-17
rs11627052	MARK3	0.01	4.90E-09	8.042409	1.30E-18
rs4858678	RARB	-0.02	1.10E-15	18.5828	4.03E-25
rs6787363	RARB	0.02	2.30E-31	18.5828	4.03E-25
rs13070614	RARB	0.02	2.00E-22	18.5828	4.03E-25
rs1126809	TYR	-0.01	8.00E-09	53.30753	1.07E-22
rs7188859	RBFOX1	0.01	8.10E-12	21.46278	1.56E-17
rs9287914	LRP2	0.01	8.40E-13	51.80898	3.77E-23
rs10512176	ZCCHC6	-0.02	1.70E-14	4.92301	3.53E-05
rs2149108	CYP26A1	-0.01	4.00E-10	15.93865	1.99E-04
rs4658101	CDC7	0.06	2.80E-160	13.92986	2.93E-10
rs56385951	CARD10	0.03	2.90E-24	18.07626	9.99E-09
rs61975075	DAAM1	0.03	1.80E-14	8.123753	4.54E-06
rs35285683	DAAM1	-0.02	2.30E-12	8.123753	4.54E-06
rs77587000	DHRS7	-0.09	2.90E-27	18.15791	2.10E-32
rs11639803	SALL1	0.02	4.80E-14	42.75288	4.41E-19

rs7196852	SALL1	-0.02	2.30E-31	42.75288	4.41E-19
rs28514893	SALL1	0.02	3.90E-18	42.75288	4.41E-19
rs80218807	ARHGEF10	0.02	3.10E-08	14.12378	3.73E-10
rs13283933	RAPGEF1	-0.02	2.20E-13	14.5708	1.48E-24
rs10764494	ARHGAP21	0.01	8.50E-13	2.259635	8.11E-01
rs4285804	SUFU	0.01	1.20E-11	16.12064	1.52E-29
rs9988687	PBLD	0.07	3.40E-209	19.06877	5.09E-18
rs3835100	PBLD	-0.08	1.20E-56	19.06877	5.09E-18
rs8076249	MAP2K6	-0.01	2.90E-08	6.919825	7.48E-04
rs7482884	ELP4	0.02	4.20E-25	15.67269	1.78E-28
rs76567987	TSPAN11	0.02	1.70E-17	31.33882	1.75E-15
rs62391624	GMDS	-0.02	8.70E-10	14.60883	3.71E-23
rs7620608	KAT2B	-0.01	1.40E-08	11.0985	1.23E-12
rs77877421	FOXP1	-0.03	1.30E-11	8.741321	4.01E-13
rs787540	GADD45A	-0.02	7.70E-16	18.43828	1.23E-13
rs6673575	LPHN2	0.01	8.80E-11	9.46743	1.84E-10
rs2761882	BMP4	-0.01	6.40E-10	31.22687	4.25E-23
rs12619508	PAX8	-0.01	7.70E-10	31.39058	3.27E-22
rs10483727	SIX1	-0.02	2.60E-21	43.60995	1.05E-23
rs74056359	PLEKHG3	-0.02	2.50E-16	14.20603	2.41E-13
rs599892	MACF1	0.01	1.10E-08	5.515089	2.58E-07
rs28840750	RHPN2	-0.03	2.40E-08	28.00236	1.79E-24
rs72759609	PDZD2	0.03	7.50E-19	13.78282	1.67E-15
rs1852148	LRRIQ1	-0.01	1.10E-08	6.842777	0.225244
rs251526	FST	-0.02	3.90E-10	39.57679	4.08E-17
rs11584075	NAV1	0.02	1.60E-10	7.214548	1.18E-08
rs9534439	LRCH1	0.02	9.90E-17	8.678528	3.29E-10
rs10910	STRA6	0.01	1.10E-08	43.31332	4.73E-22
rs163524	SIX3	-0.01	1.00E-08	46.82838	1.75E-31
rs10823610	ADAMTS14	0.01	9.60E-09	21.59473	7.45E-10
rs2430356	FBLN5	-0.01	2.70E-08	21.46157	2.65E-14
rs11079419	BCAS3	0.03	2.30E-31	16.18692	4.83E-35
rs12024620	PRDM16	0.04	7.40E-20	39.70996	2.53E-24
rs57742792	EFNA3	0.01	3.50E-11	18.74617	2.60E-15

rs11808696	CDC42BPA	-0.05	5.80E-48	10.33683	2.28E-21
rs1930941	RSPO3	0.02	1.10E-15	17.18547	4.67E-05
rs7615960	ZNF385D	-0.02	1.50E-08	18.46011	2.44E-12
rs74764079	BMP3	-0.05	1.70E-19	1.680176	1.00E+00
rs17398137	ABI3BP	-0.03	4.00E-26	17.55565	7.87E-17
rs3754442	ALX3	0.01	1.20E-09	37.27406	2.23E-19
rs59199978	ADAMTSL3	-0.01	3.90E-09	18.12646	1.02E-13
rs9852867	CLDN1	-0.01	3.20E-08	11.79914	1.82E-04
rs71296770	CADPS	-0.02	8.20E-11	19.03748	5.26E-08
rs2092524	KIF6	0.01	1.30E-13	21.34942	9.37E-17
rs1905014	C8orf48	-0.01	4.40E-13	17.97731	1.22E-06
rs13264644	PSCA	-0.01	4.50E-08	1.106335	1.00E+00
rs7610486	FILIP1L	0.01	1.60E-08	3.997873	1.00E+00
rs4832012	ATOH8	0.01	8.90E-13	23.73023	2.35E-17
rs11564398	CDH2	0.01	4.30E-12	22.7985	3.66E-15
rs34536594	TANC2	-0.02	5.50E-11	13.3008	2.33E-11
rs11602582	DCDC1	-0.04	4.10E-15	6.705269	2.49E-01
rs6999835	PKIA	0.01	2.40E-10	34.49301	4.76E-18
rs78977588	MYEOV	0.02	6.30E-09	10.66949	1.00E+00
rs4839470	VANGL1	-0.02	3.50E-23	24.34075	3.09E-24
rs12137699	VANGL1	0.02	4.30E-27	24.34075	3.09E-24
rs6860726	MARCH3	-0.01	1.30E-08	16.68074	8.78E-18
rs742127	SELP	0.02	3.60E-15	55.34919	6.48E-21
rs906568	C4orf32	-0.01	2.00E-13	14.0661	6.36E-19
rs1901440	NCKAP5	-0.01	3.40E-08	16.93198	7.85E-09
rs17734073	MSC	-0.02	2.70E-10	42.9147	1.94E-17
rs10862708	TMTC2	0.02	1.80E-29	13.14176	6.43E-13
rs2033054	GSX2	-0.01	1.30E-10	55.51402	3.75E-21
rs9905786	NPLOC4	0.02	5.30E-25	15.42598	8.23E-39
rs11114001	TMEM119	0.01	7.30E-10	30.30984	2.74E-16
rs5762752	CHEK2	-0.02	7.90E-33	9.733569	2.17E-07
rs8034595	NR2F2	-0.02	1.70E-15	23.32289	1.79E-23
rs2266963	UBE2L3	-0.01	1.90E-08	16.5191	2.18E-19
rs7744813	KCNQ5	-0.02	1.00E-15	21.68797	6.48E-12

rs16909978	PTCH1	0.02	9.10E-10	20.4355	8.33E-30
rs13022913	SESTD1	-0.01	5.50E-09	13.41922	1.47E-18
rs9330799	WNT7B	-0.02	3.70E-14	1.750368	1
rs9330813	WNT7B	-0.01	1.10E-09	1.750368	1
rs7096311	COL13A1	-0.01	2.70E-09	32.70084	4.45E-17
rs340762	FAM49A	0.01	9.40E-09	26.35461	4.56E-26
rs2220051	LEKR1	-0.01	2.00E-08	17.17502	2.24E-07
rs6030	F5	0.01	1.80E-10	46.5512	3.92E-20
rs112166936	CENPW	-0.02	1.40E-22	19.40042	5.23E-10
rs11684168	FOXI3	0.01	2.90E-08	54.54089	1.36E-26
rs12471698	TEX41	-0.01	5.60E-15	27.67978	2.41E-17
rs10084346	DIRC3	0.01	2.20E-08	9.800193	0.114439
rs934237	DIRC3	-0.02	1.70E-11	9.800193	0.114439
rs1550094	PRSS56	0.01	5.20E-13	14.18323	1
rs73173591	LINC00461	0.03	2.60E-19	18.0087	1.13E-05

Appendix Table 10. Gene-based association analysis of UK Biobank and IGGC meta-analysis of disc diameter

SYMBOL	GENE	CH R	START	STOP	NSNP S	NPA RAM	N	ZSTAT	P
PRDM16	ENSG00000142611	1	2985732	3355185	987	158	89544	6.8618	3.40E-12
MACF1	ENSG00000127603	1	39546988	39952849	680	35	89544	6.0333	8.00E-10
KIAA0754	ENSG00000255103	1	39876151	39882154	8	3	89544	5.1266	1.50E-07
PABPC4	ENSG00000090621	1	40026488	40042462	29	8	89544	5.0984	1.70E-07
CDC7	ENSG00000097046	1	91966408	91991321	42	8	89544	6.4634	5.10E-11
TGFBR3	ENSG00000069702	1	92145902	92371892	638	66	89544	7.1469	4.40E-13
AHCYL1	ENSG00000168710	1	110527308	11056635 7	17	6	89544	4.7273	1.10E-06
STRIP1	ENSG00000143093	1	110574199	11061726 3	52	6	89544	5.9703	1.20E-09
ALX3	ENSG00000156150	1	110602616	11061332 2	16	3	89544	5.797	3.40E-09
VANGL1	ENSG00000173218	1	116184574	11624084 5	161	28	89544	7.5435	2.30E-14
CASQ2	ENSG00000118729	1	116242628	11631140 2	290	22	89544	4.9883	3.00E-07
ADAM15	ENSG00000143537	1	155023042	15503525 2	21	8	89544	5.9127	1.70E-09

EFNA3	ENSG00000143590	1	155036224	15506001 4	34	8	89544	5.4919	2.00E-08
EFNA1	ENSG00000169242	1	155099936	15510733 3	13	4	89544	5.7406	4.70E-09
KRTCAP2	ENSG00000163463	1	155141884	15514595 1	11	3	89544	5.6509	8.00E-09
RP11-201K10.3	ENSG00000273088	1	155141885	15515974 8	29	8	89544	5.5333	1.60E-08
ASH1L	ENSG00000116539	1	155305059	15553259 8	158	22	89544	4.7239	1.20E-06
MSTO1	ENSG00000125459	1	155579979	15571815 3	113	18	89544	4.9366	4.00E-07
YY1AP1	ENSG00000163374	1	155629237	15565879 1	24	8	89544	4.6122	2.00E-06
F5	ENSG00000198734	1	169483404	16955582 6	285	27	89544	8.0391	4.50E-16
SELP	ENSG00000174175	1	169558087	16959943 1	113	21	89544	5.5134	1.80E-08
PSEN2	ENSG00000143801	1	227057885	22708380 6	73	8	89544	5.1891	1.10E-07
ADCK3	ENSG00000163050	1	227085237	22717524 6	193	37	89544	5.7772	3.80E-09
CDC42BPA	ENSG00000143776	1	227177566	22750617 5	1545	29	89544	8.7056	1.60E-18
FAM150B	ENSG00000189292	2	279558	288851	16	4	89544	4.5947	2.20E-06
LTBP1	ENSG00000049323	2	33172039	33624576	1664	101	89544	6.7587	7.00E-12
ATOH8	ENSG00000168874	2	85978467	86015189	59	9	89544	6.0471	7.40E-10
MGAT4A	ENSG00000071073	2	99235569	99347589	200	24	89544	4.9065	4.60E-07
BCL2L11	ENSG00000153094	2	111876955	11192602 4	83	14	89544	5.1985	1.00E-07
PSD4	ENSG00000125637	2	113914902	11396697 3	138	28	89544	4.8044	7.80E-07
PAX8	ENSG00000125618	2	113973574	11403652 7	191	21	89544	4.5658	2.50E-06
NCKAP1	ENSG00000061676	2	183773843	18390358 6	281	26	89544	4.5766	2.40E-06
DIRC3	ENSG00000231672	2	218148742	21862131 6	1249	72	89544	7.7062	6.50E-15
PRSS56	ENSG00000237412	2	233385173	23339042 2	15	6	89544	6.5192	3.50E-11
KAT2B	ENSG00000114166	3	20081515	20195896	406	39	89544	5.4813	2.10E-08
ZNF385D	ENSG00000151789	3	21459915	22414812	3745	280	89544	5.0937	1.80E-07
RARB	ENSG00000077092	3	25215823	25639423	1441	125	89544	12.535	2.40E-36
FEZF2	ENSG00000153266	3	62355356	62359999	3	2	89544	4.6242	1.90E-06
FOXP1	ENSG00000114861	3	71003844	71633140	1116	141	89544	4.5718	2.40E-06
ZNF654	ENSG00000175105	3	88188254	88193815	8	2	89544	4.5751	2.40E-06
C3orf38	ENSG00000179021	3	88198893	88217879	32	4	89544	4.5564	2.60E-06

CMSS1	ENSG00000184220	3	99536678	99897447	402	30	89544	5.5457	1.50E-08
FILIP1L	ENSG00000168386	3	99548985	99833357	326	25	89544	5.6475	8.10E-09
ABI3BP	ENSG00000154175	3	100468000	100712359	601	50	89544	6.7008	1.00E-11
LEKR1	ENSG00000197980	3	156543270	156763918	577	29	89544	5.0417	2.30E-07
CRMP1	ENSG00000072832	4	5749811	5894785	507	86	89544	4.9451	3.80E-07
LIMCH1	ENSG00000064042	4	41361624	41702061	724	71	89544	7.0376	9.80E-13
FIP1L1	ENSG00000145216	4	54243810	55161439	2152	171	89544	5.1361	1.40E-07
GSX2	ENSG00000180613	4	54965690	54968672	7	2	89544	5.646	8.20E-09
BMP3	ENSG00000152785	4	81952119	81978685	46	7	89544	6.8424	3.90E-12
RASGEF1B	ENSG00000138670	4	82347547	82965397	1798	124	89544	4.9132	4.50E-07
FAT4	ENSG00000196159	4	126237554	126414087	434	49	89544	4.654	1.60E-06
PLEKHG4B	ENSG00000153404	5	140373	190085	160	14	89544	5.267	6.90E-08
PDCD6	ENSG00000249915	5	271736	353971	201	17	89544	4.7849	8.60E-07
AHRR	ENSG00000063438	5	304291	438406	387	42	89544	5.1151	1.60E-07
TMEM161B	ENSG00000164180	5	87485450	87565293	112	17	89544	5.8048	3.20E-09
MEF2C	ENSG00000081189	5	88013975	88199922	310	35	89544	4.7926	8.20E-07
DUSP1	ENSG00000120129	5	172195093	172198198	7	2	89544	4.9379	3.90E-07
GMDS	ENSG00000112699	6	1624041	2245926	1256	125	89544	6.1883	3.00E-10
KIF6	ENSG00000164627	6	39297766	39693181	956	71	89544	6.4714	4.90E-11
KCNQ5	ENSG00000185760	6	73331520	73908574	1228	117	89544	5.2014	9.90E-08
CENPW	ENSG00000203760	6	126661320	126670021	9	4	89544	8.209	1.10E-16
TNS3	ENSG00000136205	7	47314752	47622156	712	83	89544	4.7145	1.20E-06
ARHGEF10	ENSG00000104728	8	1772142	1906807	571	100	89544	4.6482	1.70E-06
NCOA2	ENSG00000140396	8	71021997	71316040	425	28	89544	4.5791	2.30E-06
RP11-382J12.1	ENSG00000246366	8	71520812	71575514	139	15	89544	4.9599	3.50E-07
LACTB2	ENSG00000147592	8	71547553	71581409	89	12	89544	4.9298	4.10E-07
CNBD1	ENSG00000176571	8	87878670	88627447	1805	86	89544	4.639	1.80E-06
PSCA	ENSG00000167653	8	143751726	143764142	62	6	89544	5.2481	7.70E-08
LY6K	ENSG00000160886	8	143781529	143786545	16	2	89544	4.6929	1.30E-06
THEM6	ENSG00000130193	8	143808621	143818345	26	7	89544	5.4459	2.60E-08
PTCH1	ENSG00000185920	9	98205262	98279339	139	15	89544	6.0632	6.70E-10
IKBKAP	ENSG00000070061	9	111629797	111696396	269	31	89544	4.5894	2.20E-06
FAM206A	ENSG00000119328	9	111696461	111713024	39	8	89544	5.4109	3.10E-08
CTNNAL1	ENSG00000119326	9	111704851	111775809	191	21	89544	5.6725	7.00E-09

DENND1A	ENSG00000119522	9	126141933	12669243 1	1202	65	89544	5.4291	2.80E-08
RAPGEF1	ENSG00000107263	9	134452157	13461546 1	372	34	89544	6.8592	3.50E-12
MSRB2	ENSG00000148450	10	23384435	23410942	53	9	89544	4.7302	1.10E-06
ANK3	ENSG00000151150	10	61786056	62493248	1922	102	89544	5.1367	1.40E-07
DNAJC12	ENSG00000108176	10	69556427	69597924	61	11	89544	5.2548	7.40E-08
SIRT1	ENSG00000096717	10	69644427	69678147	75	10	89544	5.5414	1.50E-08
HERC4	ENSG00000148634	10	69681665	69835105	254	24	89544	5.8496	2.50E-09
MYPN	ENSG00000138347	10	69865912	69971774	387	34	89544	17.787	4.50E-71
ATOH7	ENSG00000179774	10	69990386	69991871	1	1	89544	14.933	1.00E-50
PBLD	ENSG00000108187	10	70042417	70092806	156	11	89544	15.564	6.40E-55
HNRNPH3	ENSG00000096746	10	70090931	70102948	21	5	89544	15.961	1.20E-57
RUFY2	ENSG00000204130	10	70100864	70167051	121	9	89544	15.104	7.60E-52
DNA2	ENSG00000138346	10	70173821	70231879	175	16	89544	17.485	9.30E-69
SLC25A16	ENSG00000122912	10	70237756	70287231	177	15	89544	14.348	5.50E-47
TET1	ENSG00000138336	10	70320413	70454239	405	27	89544	7.2745	1.70E-13
CCAR1	ENSG00000060339	10	70480769	70552134	128	15	89544	5.7013	5.90E-09
STOX1	ENSG00000165730	10	70587298	70655188	180	27	89544	8.9892	1.20E-19
DDX50	ENSG00000107625	10	70661034	70706603	116	17	89544	7.4738	3.90E-14
DDX21	ENSG00000165732	10	70715884	70744829	46	12	89544	8.4679	1.20E-17
KIAA1279	ENSG00000198954	10	70748487	70776738	76	12	89544	7.5463	2.20E-14
HKDC1	ENSG00000156510	10	70980059	71027315	156	30	89544	4.6778	1.50E-06
ADAMTS14	ENSG00000138316	10	72432559	72522197	398	60	89544	6.1179	4.70E-10
RNLS	ENSG00000184719	10	90033621	90344287	642	42	89544	5.5793	1.20E-08
TMEM180	ENSG00000138111	10	104221149	10423680 2	50	6	89544	6.6061	2.00E-11
ACTR1A	ENSG00000138107	10	104238986	10426248 2	59	3	89544	5.7052	5.80E-09
SUFU	ENSG00000107882	10	104263744	10439329 2	254	15	89544	6.683	1.20E-11
TRIM8	ENSG00000171206	10	104404253	10441816 4	20	7	89544	5.8848	2.00E-09
MXI1	ENSG00000119950	10	111967363	11204712 3	189	20	89544	5.1651	1.20E-07
SMNDC1	ENSG00000119953	10	112050488	11206470 9	29	7	89544	4.791	8.30E-07
DCDC1	ENSG00000170959	11	30851916	31391357	636	27	89544	7.46	4.30E-14
DNAJC24	ENSG00000170946	11	31391387	31453396	80	9	89544	6.8183	4.60E-12
IMMP1L	ENSG00000148950	11	31453948	31531192	73	8	89544	8.945	1.90E-19
ELP4	ENSG00000109911	11	31531297	31805546	280	16	89544	9.147	2.90E-20
PAX6	ENSG00000007372	11	31806340	31839509	39	5	89544	7.5986	1.50E-14
FAT3	ENSG00000165323	11	92085262	92629618	1039	57	89544	4.7772	8.90E-07
TSPAN9	ENSG0000011105	12	3186521	3395730	786	99	89544	5.6631	7.40E-09
CAPRIN2	ENSG00000110888	12	30862486	30907885	119	13	89544	4.6922	1.40E-06

TSPAN19	ENSG00000231738	12	85408094	85430055	21	6	89544	5.5022	1.90E-08	
LRRIQ1	ENSG00000133640	12	85430092	85657002	245	18	89544	5.4126	3.10E-08	
TMEM119	ENSG00000183160	12	108983622	10899209	6	23	8	89544	5.3773	3.80E-08
LRCH1	ENSG00000136141	13	47127303	47327175	464	41	89544	7.4496	4.70E-14	
COL4A1	ENSG00000187498	13	110801318	11095949	6	591	74	89544	4.8923	5.00E-07
COL4A2	ENSG00000134871	13	110958159	11116537	4	840	107	89544	4.9261	4.20E-07
FRMD6	ENSG00000139926	14	51955818	52197445	701	96	89544	5.8839	2.00E-09	
BMP4	ENSG00000125378	14	54416454	54425479	15	4	89544	5.556	1.40E-08	
PPM1A	ENSG00000100614	14	60712470	60765805	80	8	89544	5.156	1.30E-07	
C14orf39	ENSG00000179008	14	60863187	60982261	218	15	89544	7.3108	1.30E-13	
SIX6	ENSG00000184302	14	60975669	60979568	3	2	89544	5.79	3.50E-09	
SIX1	ENSG00000126778	14	61110133	61124977	12	3	89544	7.2401	2.20E-13	
SIX4	ENSG00000100625	14	61176246	61191066	13	5	89544	5.7179	5.40E-09	
MNAT1	ENSG00000020426	14	61201460	61436671	216	37	89544	5.3193	5.20E-08	
ZBTB25	ENSG00000089775	14	64915824	64971931	103	11	89544	4.5661	2.50E-06	
ZBTB1	ENSG00000126804	14	64970430	65000408	76	8	89544	4.5643	2.50E-06	
PPP1R36	ENSG00000165807	14	65016620	65056098	98	14	89544	5.371	3.90E-08	
WDR72	ENSG00000166415	15	53805938	54055075	886	50	89544	4.7998	7.90E-07	
THSD4	ENSG00000187720	15	71389291	72075722	1869	185	89544	5.455	2.40E-08	
ISLR	ENSG00000129009	15	74466012	74469213	9	2	89544	4.7124	1.20E-06	
STRA6	ENSG00000137868	15	74471807	74504608	67	10	89544	5.5036	1.90E-08	
ADAMTSL3	ENSG00000156218	15	84322838	84708594	890	57	89544	5.9866	1.10E-09	
PDE8A	ENSG00000073417	15	85523671	85682376	475	34	89544	4.6224	1.90E-06	
BCAS3	ENSG00000141376	17	58754814	59470199	819	68	89544	13.032	4.00E-39	
TANC2	ENSG00000170921	17	61086917	61505060	551	27	89544	4.9573	3.60E-07	
NPLOC4	ENSG00000182446	17	79523913	79615495	354	25	89544	7.4394	5.10E-14	
PDE6G	ENSG00000185527	17	79617489	79630142	66	10	89544	5.7256	5.20E-09	
COLEC12	ENSG00000158270	18	319361	500722	719	77	89544	6.8445	3.80E-12	
CDH2	ENSG00000170558	18	25530930	25757410	414	46	89544	5.7914	3.50E-09	
C20orf112	ENSG00000197183	20	31030862	31172876	192	20	89544	5.3748	3.80E-08	
EFCAB8	ENSG00000215529	20	31446729	31549006	269	32	89544	4.6227	1.90E-06	
EYA2	ENSG00000064655	20	45523263	45817492	780	93	89544	5.6007	1.10E-08	
ERG	ENSG00000157554	21	39751949	40033704	788	98	89544	4.9303	4.10E-07	
UBE2L3	ENSG00000185651	22	21903736	21978323	82	3	89544	5.1071	1.60E-07	
YDJC	ENSG00000161179	22	21982378	21984353	2	1	89544	4.7512	1.00E-06	
TTC28	ENSG00000100154	22	28374004	29075853	1044	45	89544	9.1576	2.70E-20	
CHEK2	ENSG00000183765	22	29083731	29138410	122	20	89544	11.627	1.50E-31	
HSCB	ENSG00000100209	22	29138019	29153503	28	8	89544	6.006	9.50E-10	
CCDC117	ENSG00000159873	22	29168662	29185283	45	10	89544	6.539	3.10E-11	
XBP1	ENSG00000100219	22	29190543	29196585	7	3	89544	6.4161	7.00E-11	
ZNRF3	ENSG00000183579	22	29279580	29453475	313	34	89544	7.3969	7.00E-14	

CARD10	ENSG00000100065	22	37886400	37915549	84	15	89544	7.6576	9.50E-15
CDC42EP1	ENSG00000128283	22	37956454	37965412	17	4	89544	5.4447	2.60E-08
NOL12	ENSG00000100101	22	38077680	38170137	197	23	89544	4.5828	2.30E-06
TRIOBP	ENSG00000100106	22	38093011	38172563	170	20	89544	4.7419	1.10E-06
FBLN1	ENSG00000077942	22	45898118	45997015	306	50	89544	4.657	1.60E-06
WNT7B	ENSG00000188064	22	46316242	46373009	203	34	89544	6.0834	5.90E-10

Appendix Table 11. Gene-based analysis of glaucoma MTAG GWAS (list of 196 significant genes after Bonferroni correction)

GENE	CHR	START	STOP	NSNPS	NPARAM	ZSTAT	P	SYMBOL
ENSG00000143183	1	165696032	165796992	136	25	16.106	1.15E-58	<i>TMCO1</i>
ENSG00000007237	17	9813926	10101868	884	154	13.039	3.69E-39	<i>GAS7</i>
ENSG00000196526	4	7760441	7941653	836	53	12.522	2.81E-36	<i>AFAP1</i>
ENSG00000154188	8	108261721	108510283	737	66	10.626	1.12E-26	<i>ANGPT1</i>
ENSG00000136944	9	129376722	129463311	216	36	10.092	3.00E-24	<i>LMX1B</i>
ENSG00000112699	6	1624041	2245926	1258	119	9.9988	7.71E-24	<i>GMDS</i>
ENSG00000105971	7	115927434	116148595	494	49	9.701	1.49E-22	<i>CAV2</i>
ENSG00000157827	2	153191751	153506348	901	72	8.7735	8.66E-19	<i>FMNL2</i>
ENSG00000085276	3	168801287	169381406	1531	132	8.3933	2.36E-17	<i>MECOM</i>
ENSG00000105974	7	116164839	116201233	97	14	8.2351	8.97E-17	<i>CAV1</i>
ENSG00000122870	10	60272900	60591195	564	45	7.7995	3.11E-15	<i>BICC1</i>
ENSG00000134571	11	47352957	47374253	40	11	7.7552	4.41E-15	<i>MYBPC3</i>
ENSG00000135269	7	115850547	115898837	145	12	7.7277	5.48E-15	<i>TES</i>
ENSG00000149187	11	47487496	47587121	104	22	7.6975	6.94E-15	<i>CELF1</i>
ENSG00000154122	5	14704910	14871887	361	50	7.5972	1.51E-14	<i>ANKH</i>
ENSG00000196914	11	120207787	120360645	233	15	7.523	2.68E-14	<i>ARHGEF12</i>
ENSG00000100106	22	38093011	38172563	171	20	7.5211	2.72E-14	<i>TRIOBP</i>
ENSG00000165029	9	107543283	107690518	515	79	7.5088	2.98E-14	<i>ABCA1</i>
ENSG00000213619	11	47586888	47606114	15	6	7.4794	3.73E-14	<i>NDUFS3</i>
ENSG00000100101	22	38077680	38170137	198	23	7.4206	5.83E-14	<i>NOL12</i>
ENSG00000165916	11	47440320	47447993	13	4	7.3928	7.19E-14	<i>PSMC3</i>
ENSG00000183765	22	29083731	29138410	123	20	7.3464	1.02E-13	<i>CHEK2</i>
ENSG00000100116	22	38203912	38213183	19	3	7.3336	1.12E-13	<i>GCAT</i>
ENSG00000110514	11	47290712	47351582	123	14	7.2451	2.16E-13	<i>MADD</i>
ENSG00000181264	11	120195838	120204391	12	2	7.2262	2.48E-13	<i>TMEM136</i>

ENSG00000134574	11	47236493	47260767	51	7	7.2223	2.56E-13	<i>DDB2</i>
ENSG00000134954	11	128328656	128457453	269	50	7.1932	3.16E-13	<i>ETS1</i>
ENSG00000134917	11	130274820	130298888	61	16	7.0756	7.44E-13	<i>ADAMTS8</i>
ENSG00000109919	11	47638867	47664175	22	6	7.0407	9.56E-13	<i>MTCH2</i>
ENSG00000025434	11	47269851	47290396	42	7	7.0056	1.23E-12	<i>NR1H3</i>
ENSG00000196814	9	129089128	129269320	425	41	6.9592	1.71E-12	<i>MVB12B</i>
ENSG00000165917	11	47459308	47470730	31	6	6.914	2.36E-12	<i>RAPSN</i>
ENSG00000112685	6	485133	693117	565	47	6.8764	3.07E-12	<i>EXOC2</i>
ENSG00000168056	11	65306276	65326401	14	4	6.8757	3.08E-12	<i>LTBP3</i>
ENSG00000134575	11	47260853	47270457	24	4	6.8568	3.52E-12	<i>ACP2</i>
ENSG00000175161	3	85008132	86123579	2520	72	6.8394	3.98E-12	<i>CADM2</i>
ENSG00000100124	22	38226862	38245334	18	7	6.8283	4.30E-12	<i>ANKRD54</i>
ENSG00000140937	16	64977656	65160015	437	45	6.8072	4.98E-12	<i>CDH11</i>
ENSG00000110536	11	47586982	47595013	7	3	6.7968	5.35E-12	<i>PTPMT1</i>
ENSG00000100139	22	38301664	38338829	45	8	6.757	7.04E-12	<i>MICALL1</i>
ENSG00000149177	11	48002113	48189670	310	26	6.7299	8.49E-12	<i>PTPRJ</i>
ENSG00000136828	9	129677053	129985445	484	29	6.6688	1.29E-11	<i>RALGPS1</i>
ENSG00000100129	22	38244875	38285414	45	8	6.6577	1.39E-11	<i>EIF3L</i>
ENSG00000176540	11	48387037	48388017	1	1	6.5827	2.31E-11	<i>OR4C5</i>
ENSG00000075420	3	171757418	172119455	751	83	6.5649	2.60E-11	<i>FNDC3B</i>
ENSG00000066336	11	47376411	47400127	58	7	6.5285	3.32E-11	<i>SPI1</i>
ENSG00000142186	11	65292548	65306175	14	5	6.483	4.50E-11	<i>SCYL1</i>
ENSG00000171502	1	86194916	86622626	1297	76	6.4776	4.66E-11	<i>COL24A1</i>
ENSG00000136859	9	129849611	129885162	53	10	6.4641	5.10E-11	<i>ANGPTL2</i>
ENSG00000165915	11	47428683	47438047	20	4	6.4427	5.87E-11	<i>SLC39A13</i>
ENSG00000147883	9	22002902	22009362	6	3	6.4406	5.95E-11	<i>CDKN2B</i>
ENSG00000151376	11	86152150	86383678	815	65	6.4253	6.58E-11	<i>ME3</i>
ENSG00000173442	11	65343509	65360121	24	7	6.377	9.03E-11	<i>EHBP1L1</i>
ENSG00000030066	11	47799639	47870107	105	12	6.364	9.83E-11	<i>NUP160</i>
ENSG00000070366	17	1963133	2207065	542	27	6.3553	1.04E-10	<i>SMG6</i>
ENSG00000136153	13	76194570	76434004	561	53	6.313	1.37E-10	<i>LMO7</i>
ENSG00000140262	15	57210821	57591479	831	29	6.2902	1.59E-10	<i>TCF12</i>
ENSG00000128346	22	38339528	38349676	10	5	6.2606	1.92E-10	<i>C22orf23</i>
ENSG00000124813	6	45295894	45632086	821	80	6.2473	2.09E-10	<i>RUNX2</i>
ENSG00000148737	10	114710009	114927437	394	66	6.2188	2.50E-10	<i>TCF7L2</i>

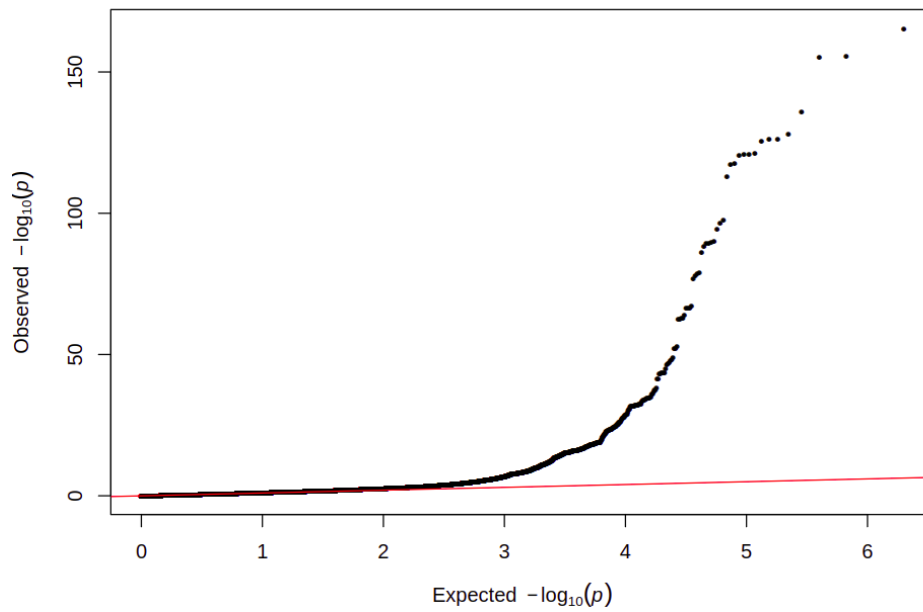
ENSG00000158019	2	28112808	28561768	815	57	6.1527	3.81E-10	<i>BRE</i>
ENSG00000155363	1	113215763	113243368	54	10	6.1485	3.91E-10	<i>MOV10</i>
ENSG00000178175	5	71738479	71803554	149	18	6.1474	3.94E-10	<i>ZNF366</i>
ENSG00000141376	17	58754814	59470199	825	68	6.1133	4.88E-10	<i>BCAS3</i>
ENSG00000165923	11	47681143	47736941	86	11	6.1109	4.95E-10	<i>AGBL2</i>
ENSG00000105967	7	115575202	115799950	494	45	6.0643	6.63E-10	<i>TFEC</i>
ENSG00000149179	11	46958240	47185936	280	25	6.014	9.05E-10	<i>C11orf49</i>
ENSG00000158955	17	44910567	44964096	95	12	6.0122	9.15E-10	<i>WNT9B</i>
ENSG00000196284	6	44777054	45345690	1544	37	6.0029	9.69E-10	<i>SUPT3H</i>
ENSG00000196159	4	126237554	126414087	433	48	5.9566	1.29E-09	<i>FAT4</i>
ENSG00000086205	11	49168187	49230222	167	16	5.897	1.85E-09	<i>FOLH1</i>
ENSG00000129521	14	34393437	34931980	1553	192	5.8103	3.12E-09	<i>EGLN3</i>
ENSG00000175619	11	48238344	48239314	2	1	5.7915	3.49E-09	<i>OR4B1</i>
ENSG00000160145	3	123798870	124445172	1779	114	5.7898	3.52E-09	<i>KALRN</i>
ENSG00000108379	17	44839872	44910520	86	16	5.7848	3.63E-09	<i>WNT3</i>
ENSG00000049192	5	64444563	64777747	667	46	5.7844	3.64E-09	<i>ADAMTS6</i>
ENSG00000100142	22	38348614	38437922	78	9	5.7716	3.93E-09	<i>POLR2F</i>
ENSG00000137871	15	56922379	57210769	496	35	5.7676	4.02E-09	<i>ZNF280D</i>
ENSG00000126391	11	65154070	65180996	31	9	5.7674	4.02E-09	<i>FRMD8</i>
ENSG00000169604	2	69240310	69476459	550	67	5.7471	4.54E-09	<i>ANTXR1</i>
ENSG00000007341	1	113066140	113163447	189	16	5.7206	5.31E-09	<i>ST7L</i>
ENSG00000138193	10	95753746	96092580	586	55	5.669	7.18E-09	<i>PLCE1</i>
ENSG00000109920	11	47738072	47788995	81	10	5.6557	7.76E-09	<i>FNBP4</i>
ENSG00000134569	11	46878419	46940193	95	12	5.648	8.12E-09	<i>LRP4</i>
ENSG00000116489	1	113161795	113214241	64	11	5.6394	8.53E-09	<i>CAPZA1</i>
ENSG00000176567	11	48285413	48286330	3	2	5.6392	8.54E-09	<i>OR4X1</i>
ENSG00000100154	22	28374004	29075853	1037	43	5.623	9.38E-09	<i>TTC28</i>
ENSG00000173327	11	65365226	65382853	29	6	5.6223	9.42E-09	<i>MAP3K11</i>
ENSG00000138035	2	55861400	55921045	189	24	5.6206	9.52E-09	<i>PNPT1</i>
ENSG00000179008	14	60863187	60982261	216	15	5.6124	9.98E-09	<i>C14orf39</i>
ENSG00000189056	7	103112231	103629963	1814	128	5.5802	1.20E-08	<i>RELN</i>
ENSG00000115970	2	43393800	43823185	936	47	5.5787	1.21E-08	<i>THADA</i>
ENSG00000171408	6	136172834	136516712	482	60	5.5619	1.33E-08	<i>PDE7B</i>
ENSG00000171812	1	36560837	36590821	47	7	5.499	1.91E-08	<i>COL8A2</i>
ENSG00000187498	13	110801318	110959496	591	75	5.476	2.18E-08	<i>COL4A1</i>

ENSG00000156804	8	124510129	124553446	114	27	5.4735	2.21E-08	<i>FBXO32</i>
ENSG00000011304	19	797075	812327	44	9	5.4614	2.36E-08	<i>PTBP1</i>
ENSG00000172733	8	30853321	30891231	46	10	5.4491	2.53E-08	<i>PURG</i>
ENSG00000183762	22	29469066	29564321	247	26	5.4466	2.57E-08	<i>KREMEN1</i>
ENSG00000215012	22	19833661	19842419	29	6	5.4253	2.89E-08	<i>C22orf29</i>
ENSG00000175216	11	46764598	46867847	133	18	5.4176	3.02E-08	<i>CKAP5</i>
ENSG00000143149	1	165631453	165668100	133	14	5.3996	3.34E-08	<i>ALDH9A1</i>
ENSG00000255073	11	58346645	58392112	69	13	5.3878	3.57E-08	<i>ZFP91-CNTF</i>
ENSG00000116117	2	205410516	206484886	2378	168	5.3551	4.28E-08	<i>PARD3B</i>
ENSG00000149182	11	47185848	47198676	21	7	5.3271	4.99E-08	<i>ARFGAP2</i>
ENSG00000174957	11	55944094	55945032	1	1	5.3054	5.62E-08	<i>OR5J2</i>
ENSG00000214891	11	49075266	49080664	15	4	5.2951	5.95E-08	<i>TRIM64C</i>
ENSG00000186660	11	58346584	58388515	67	13	5.2884	6.17E-08	<i>ZFP91</i>
ENSG00000163995	4	7967039	8160559	600	98	5.2871	6.21E-08	<i>ABLIM2</i>
ENSG00000115380	2	56093102	56151274	159	23	5.2786	6.51E-08	<i>EFEMP1</i>
ENSG00000068028	3	50367219	50378411	10	3	5.2726	6.73E-08	<i>RASSF1</i>
ENSG00000116863	1	36554476	36559533	1	1	5.2607	7.18E-08	<i>ADPRHL2</i>
ENSG00000185666	22	32908539	33454358	1371	142	5.25	7.60E-08	<i>SYN3</i>
ENSG00000204634	2	101624079	101869328	513	42	5.2411	7.98E-08	<i>TBC1D8</i>
ENSG00000169218	1	38076951	38100595	60	12	5.2191	8.99E-08	<i>RSPO1</i>
ENSG00000100146	22	38366693	38383429	8	2	5.1909	1.05E-07	<i>SOX10</i>
ENSG00000140873	16	77281710	77469011	903	64	5.189	1.06E-07	<i>ADAMTS18</i>
ENSG00000158805	16	89786808	89807311	94	14	5.1365	1.40E-07	<i>ZNF276</i>
ENSG00000231880	11	47599277	47599823	1	1	5.1199	1.53E-07	<i>KBTD4</i>
ENSG00000128250	22	29834572	29838444	9	2	5.1175	1.55E-07	<i>RFPL1</i>
ENSG00000143353	1	219347186	219386207	56	11	5.114	1.58E-07	<i>LYPLAL1</i>
ENSG00000198561	11	57520715	57587018	71	11	5.107	1.64E-07	<i>CTNND1</i>
ENSG00000173465	11	65337901	65341413	6	2	5.1008	1.69E-07	<i>SSSCA1</i>
ENSG00000001617	3	50192478	50226508	58	9	5.0735	1.95E-07	<i>SEMA3F</i>
ENSG00000198740	17	47366568	47439835	137	14	5.0731	1.96E-07	<i>ZNF652</i>
ENSG00000134697	1	38032417	38061536	58	11	5.0629	2.07E-07	<i>GNL2</i>
ENSG00000064655	20	45523263	45817492	780	92	5.0559	2.14E-07	<i>EYA2</i>
ENSG00000157368	16	70613798	70694585	211	18	5.054	2.16E-07	<i>IL34</i>
ENSG00000070081	11	17229700	17371521	263	22	5.0424	2.30E-07	<i>NUCB2</i>
ENSG00000138041	2	55774428	55846015	186	23	5.0422	2.30E-07	<i>SMEK2</i>

ENSG00000075399	16	89773542	89787394	31	10	5.032	2.43E-07	VPS9D1
ENSG00000189060	22	38201114	38203442	3	1	5.0315	2.43E-07	H1F0
ENSG00000122545	7	35840542	35944917	164	11	5.03	2.45E-07	SEPT7
ENSG00000164463	5	172483355	172566291	173	17	5.0093	2.73E-07	CREBRF
ENSG00000237388	11	48510269	48511332	4	2	5.0045	2.80E-07	OR4A47
ENSG00000129038	15	74218330	74244478	106	16	4.9967	2.92E-07	LOXL1
ENSG00000071575	2	12857015	12882860	49	10	4.9959	2.93E-07	TRIB2
ENSG00000179564	3	50316458	50325545	4	1	4.9869	3.07E-07	LSMEM2
ENSG00000073969	17	44668035	44834830	56	6	4.9754	3.26E-07	NSF
ENSG00000120733	5	137688285	137772717	179	22	4.9715	3.32E-07	KDM3B
ENSG00000197265	8	30435835	30515768	146	17	4.9525	3.66E-07	GTF2E2
ENSG00000187741	16	89803957	89883065	399	21	4.9418	3.87E-07	FANCA
ENSG00000184470	22	19863040	19929341	240	34	4.9377	3.95E-07	TXNRD2
ENSG00000011405	11	17099277	17229530	191	20	4.9241	4.24E-07	PIK3C2A
ENSG00000137834	15	66994566	67074338	197	26	4.9115	4.52E-07	SMAD6
ENSG00000113734	5	172571445	172591390	44	9	4.9083	4.59E-07	BNIP1
ENSG00000254462	11	57480077	57559058	84	11	4.903	4.72E-07	TMX2- CTNND1
ENSG00000158402	5	137620954	137674044	113	14	4.9006	4.78E-07	CDC25C
ENSG00000122507	7	33168856	33645680	1207	66	4.8941	4.94E-07	BBS9
ENSG00000188211	11	17373273	17398888	57	11	4.8896	5.05E-07	NCR3LG1
ENSG00000073712	14	53323986	53419153	172	20	4.8525	6.10E-07	FERMT2
ENSG00000054116	1	36602173	36615098	14	2	4.8271	6.93E-07	TRAPPC3
ENSG00000165912	11	47199076	47207994	18	5	4.8259	6.97E-07	PACSIN3
ENSG00000166689	11	16799842	17035990	689	55	4.8229	7.07E-07	PLEKHA7
ENSG00000178804	3	129262057	129270310	17	4	4.8163	7.31E-07	H1FOO
ENSG00000186998	22	29601840	29655586	140	25	4.8144	7.38E-07	EMID1
ENSG00000160293	9	136627016	136857726	893	102	4.7978	8.02E-07	VAV2
ENSG00000120709	5	137667624	137685416	40	10	4.7956	8.11E-07	FAM53C
ENSG00000106536	7	39017598	39532694	1340	80	4.792	8.26E-07	POU6F2
ENSG00000084636	1	32117848	32169920	83	16	4.7874	8.45E-07	COL16A1
ENSG00000112584	6	170599791	170716153	278	19	4.7803	8.75E-07	FAM120B
ENSG00000102554	13	73629114	73651676	42	10	4.7791	8.80E-07	KLF5
ENSG00000184178	4	53739149	54232242	913	77	4.778	8.85E-07	SCFD2
ENSG00000155366	1	113243728	113250056	15	5	4.7728	9.08E-07	RHOC

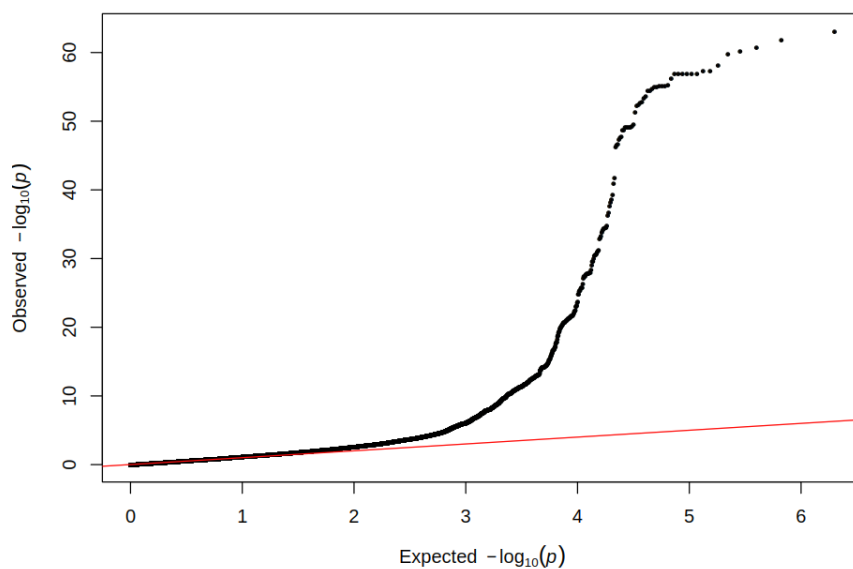
ENSG00000214706	3	50325163	50330349	4	2	4.751	1.01E-06	<i>IFRD2</i>
ENSG00000182185	14	68286496	69196935	1929	137	4.7495	1.02E-06	<i>RAD51B</i>
ENSG00000020426	14	61201460	61436671	210	36	4.7477	1.03E-06	<i>MNAT1</i>
ENSG00000142611	1	2985732	3355185	994	157	4.7464	1.04E-06	<i>PRDM16</i>
ENSG00000175806	8	9911778	10286401	1379	88	4.7301	1.12E-06	<i>MSRA</i>
ENSG00000164045	3	48198636	48229892	30	9	4.7297	1.12E-06	<i>CDC25A</i>
ENSG00000005108	7	11409984	11871824	1582	131	4.723	1.16E-06	<i>THSD7A</i>
ENSG00000198877	11	55540914	55541858	2	1	4.7213	1.17E-06	<i>OR5D13</i>
ENSG00000171132	2	45878484	46415129	1716	226	4.7176	1.19E-06	<i>PRKCE</i>
ENSG00000157110	8	30241944	30429778	286	24	4.7101	1.24E-06	<i>RBPMS</i>
ENSG00000163877	1	38000050	38019903	37	9	4.6983	1.31E-06	<i>SNIP1</i>
ENSG00000139624	12	50523575	50561288	49	5	4.6647	1.55E-06	<i>CERS5</i>
ENSG00000167822	11	55904247	55905194	5	2	4.6638	1.55E-06	<i>OR8J3</i>
ENSG00000198252	14	53196898	53241716	73	9	4.6611	1.57E-06	<i>STYX</i>
ENSG00000166979	21	33784314	33887707	257	24	4.6594	1.59E-06	<i>EVA1C</i>
ENSG00000214063	11	842808	867116	42	11	4.6569	1.61E-06	<i>TSPAN4</i>
ENSG00000008197	6	50681541	50740701	82	9	4.6558	1.61E-06	<i>TFAP2D</i>
ENSG00000177830	11	867357	915058	88	13	4.6555	1.62E-06	<i>CHID1</i>
ENSG00000123444	11	47593749	47600567	3	2	4.6521	1.64E-06	<i>KBTD4</i>
ENSG00000137692	11	102932805	102962944	44	8	4.6427	1.72E-06	<i>DCUN1D5</i>
ENSG00000114302	3	48782030	48885279	71	12	4.6369	1.77E-06	<i>PRKAR2A</i>
ENSG00000156599	11	57435219	57468659	46	9	4.6348	1.79E-06	<i>ZDHHC5</i>
ENSG00000137709	11	120107349	120190653	173	26	4.6344	1.79E-06	<i>POU2F3</i>
ENSG00000163875	1	37958176	37980375	69	12	4.6222	1.90E-06	<i>MEAF6</i>
ENSG00000162804	2	241938255	242034983	246	37	4.6123	1.99E-06	<i>SNED1</i>
ENSG00000079308	2	218664512	218867718	483	84	4.6016	2.10E-06	<i>TNS1</i>
ENSG00000163879	1	38022520	38032458	28	7	4.5882	2.24E-06	<i>DNALI1</i>
ENSG00000177479	3	48956254	49023815	61	11	4.5845	2.28E-06	<i>ARIH2</i>
ENSG00000175592	11	65659520	65668044	20	6	4.5798	2.33E-06	<i>FOSL1</i>
ENSG00000114395	3	50388126	50395891	7	3	4.5797	2.33E-06	<i>CYB561D2</i>
ENSG00000171103	2	29072687	29093167	28	5	4.5509	2.67E-06	<i>TRMT61B</i>
ENSG00000130158	19	11309971	11373157	119	16	4.5488	2.70E-06	<i>DOCK6</i>

Appendix Figure 1. Quantile-quantile plot for UK Biobank disc diameter GWAS



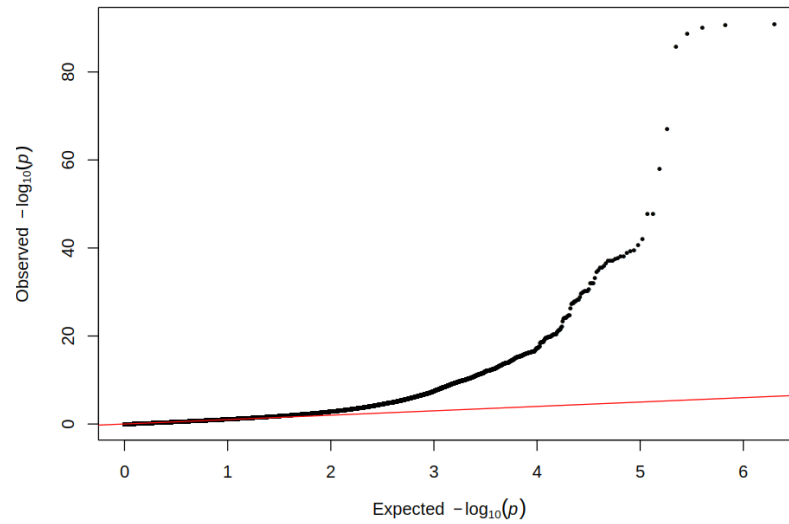
Linkage disequilibrium (LD) score regression intercept is used to assess the genomic inflation, and the intercept is 1.05 (standard error, SE=0.01) with lambda 1.20 and attenuation ratio $((\text{Intercept} - 1) / (\chi^2 - 1))$ 0.12 (SE=0.02). The quantile-quantile plot is based on 1×10^6 randomly selected SNPs.

Appendix Figure 2. Quantile-quantile plot for UKBB VCDR GWAS (vertical DD adjusted)



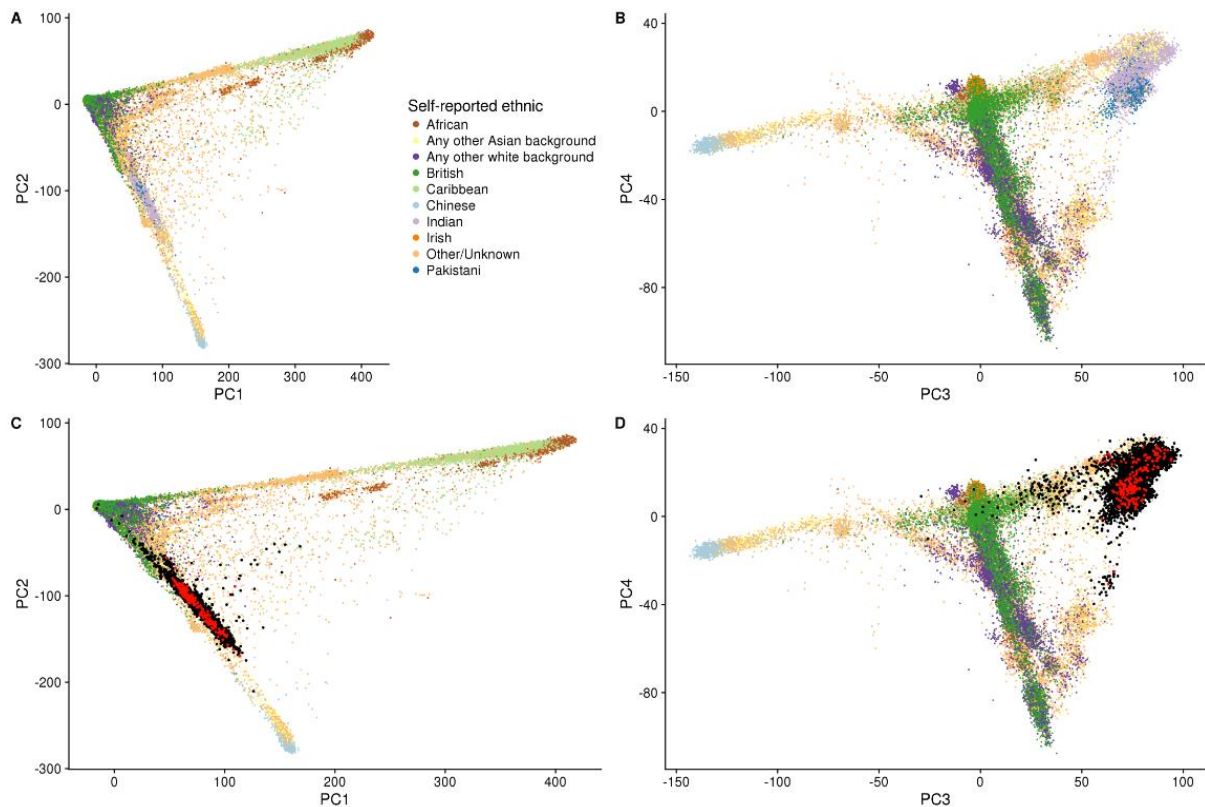
Linkage disequilibrium (LD) score regression intercept is used to assess the genomic inflation, and the intercept is 1.04 (standard error, SE=0.01) with lambda 1.20 and attenuation ratio $((\text{Intercept} - 1) / (\chi^2 - 1))$ 0.12 (SE=0.03). The quantile-quantile plot is based on 1×10^6 randomly selected SNPs.

Appendix Figure 3. Quantile-quantile plot for MTAG glaucoma GWAS result



In MTAG GWAS for glaucoma, LD score regression intercept is 0.94 (SE=0.01), and the lambda is 1.19. The quantile-quantile plot is based on 1×10^6 randomly selected SNPs.

Appendix Figure 4. Self-report ethnic background in the UK Biobank study.



The figures display different principal components for UK Biobank samples. Each point is colored based on their self-report ethnic background from UK Biobank Field 21000 (groups with less than 1,000 participants are shown as “Other/Unknown”). Panel A and Panel C are PC 1 vs PC 2, Panel B and Panel D are PC 3 vs PC 4. In Panel C and Panel D, the case-control status of South Asian ancestry (Indian or Pakistani ancestry) is superimposed with red color for glaucoma cases and black color for controls. South Asian samples have genetic ancestry which is clearly distinct from UK Biobank participants of European ancestry; the glaucoma cases of South Asian ancestry are mixed with non-glaucoma cases.