

Supporting online material

A genome-wide association study for corneal curvature identifies the *PDGFRA* gene as quantitative trait locus for eye size in white Europeans

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Table S1.
Meta-analysis of corneal curvature GWAS results from the ALSPAC sample and the 4 samples analysed by Han et al. [10]

MARKER	POS	SP2 (N=2008)			SiMES (N=2281)			SINDI (N=2142)			SCORM (N=929)			ALSPAC (N=2023)			Meta-analysis			
		A1	beta	SE	P-value	beta	SE	P-value	beta	SE	P-value	beta	SE	P-value	beta	SE	P-value	beta	SE	P-value
rs7676985	54759330	A	-0.060	0.035	8.85E-02	-0.143	0.034	2.70E-05	-0.081	0.033	1.50E-02	-0.140	0.051	6.42E-03	-0.151	0.036	2.71E-05	-0.151	0.016	9.70E-12
rs7682912	54780377	G	-0.071	0.037	5.43E-02	-0.157	0.034	5.00E-06	-0.083	0.034	1.30E-02	-0.140	0.052	7.30E-03	-0.168	0.037	6.20E-06	-0.168	0.017	3.53E-13
rs17084051	54782338	A	-0.095	0.038	1.18E-02	-0.153	0.034	1.00E-05	-0.086	0.033	1.06E-02	-0.142	0.053	7.55E-03	-0.168	0.037	6.20E-06	-0.126	0.017	4.49E-14
rs2114039	54787383	C	-0.060	0.034	7.92E-02	-0.129	0.033	7.50E-05	-0.096	0.033	3.65E-03	-0.103	0.048	8.67E-02	-0.143	0.034	3.50E-05	0.107	0.016	1.48E-11
rs7677751	54819217	T	-0.094	0.039	1.53E-02	-0.125	0.035	3.65E-04	-0.078	0.034	2.18E-02	-0.143	0.054	7.75E-03	-0.145	0.045	1.37E-03	-0.111	0.018	4.22E-10
rs2307049	54824911	A	-0.104	0.040	8.62E-03	-0.133	0.034	1.16E-04	-0.080	0.034	1.78E-02	-0.125	0.055	2.36E-02	-0.130	0.045	4.21E-03	-0.112	0.018	3.14E-10
rs7660560	54829151	A	-0.116	0.039	3.40E-03	-0.127	0.035	2.51E-04	-0.081	0.034	1.61E-02	-0.125	0.055	2.35E-02	-0.129	0.045	4.55E-03	-0.112	0.018	2.75E-10

POS = human genomic position on chromosome 4 in reference assembly genome build 36.3; A1 = risk allele; beta = standardised beta coefficient; SE = standard error of beta; Rsq = Squared correlation between imputed and true genotypes computed by MACH; I^2 = Percentage of variation across studies due to heterogeneity.

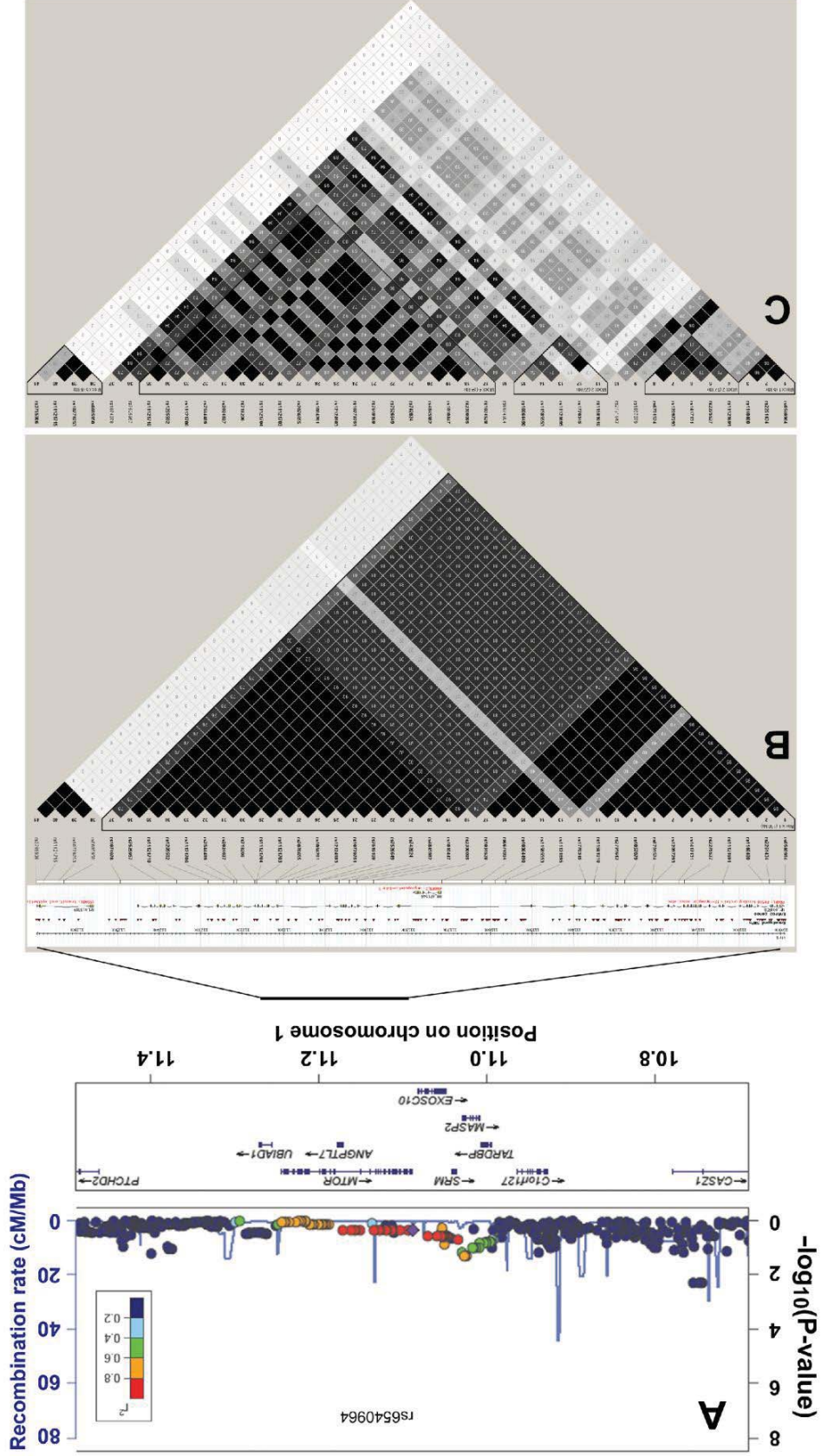


Figure S1. Results of ALSPAC GWAS for corneal curvature in the FRAP1 (MTOR) region, and LD profile in Europeans and Asians. GWAS results for corneal curvature in the FRAP1 region showed no evidence of association (Panel A). The pattern of LD differs between HapMap individuals of European ancestry (CEU; Panel B) and of Asian ancestry (CHB+JPT; Panel C). The greyscale and values shown in Panels B & C represent r^2 pairwise LD (0 = white, 1 = black).

Figure S2. Genomic region plot from ALSPAC GWAS for corneal curvature.

Results are shown from an analysis with the inclusion of the genotype at SNP rs6554163 as a covariate in the analysis (compare with Figure 2).

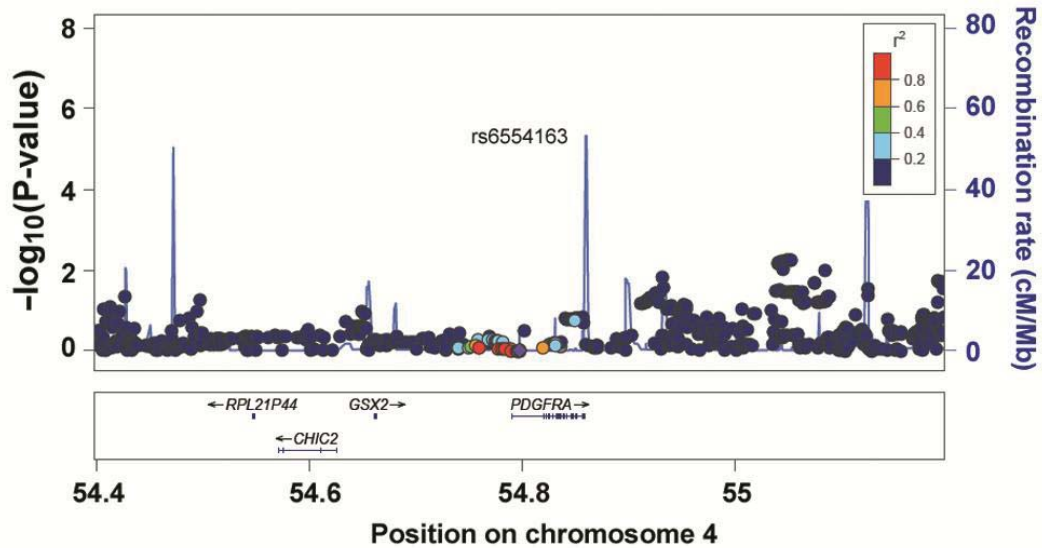


Figure S3. eQTL GWAS for probe ilmn_1681949. Panel A shows a Manhattan plot for probe ilmn_1681949 gene expression level in lymphocytes. Panel B shows a higher-resolution view of the most strongly linked region, on chromosome 6.

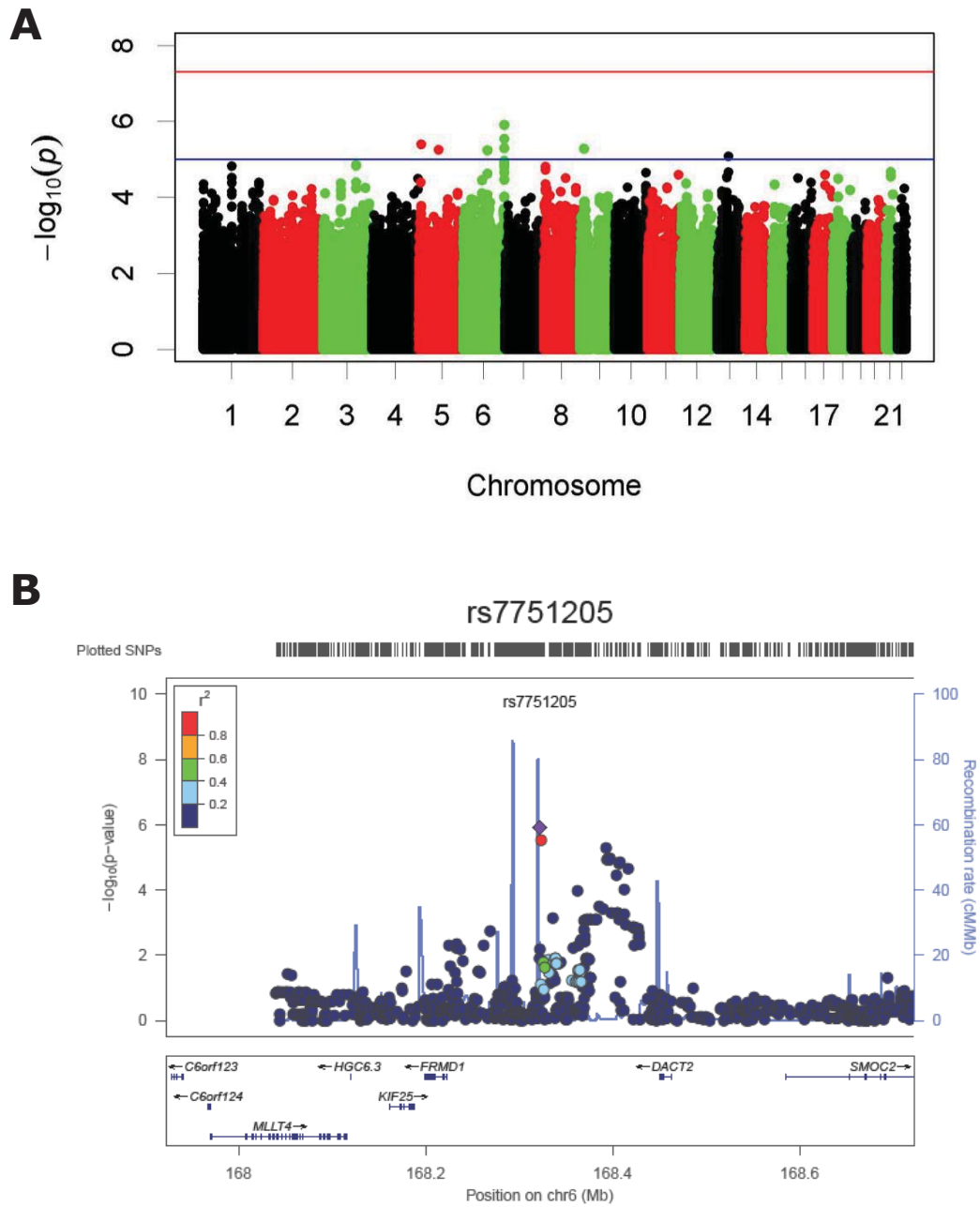


Figure S4. eQTL GWAS for probe ilmn_2086470. Panel A shows a Manhattan plot for probe ilmn_2086470 gene expression level in lymphocytes. Panel B shows a higher-resolution view of the most strongly linked region, on chromosome 7.

