Supplementary Materials

Association of polymorphisms in the *Hepatocyte Growth Factor* (*HGF*) gene promoter with keratoconus.

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Supplementary Table 1: SNPs genotyped in individual DNA samples, identified through genome-wide association scan of pooled DNA. P-values in the pooled GWAS and in individual typing of the same samples (validation) are shown, as well as in the primary replication cohort for validated SNPs. Validated SNPs (defined as $p<9.0x10^{-3}$ in the individual typing) are indicated in **bold**. Replicated SNPs (defined as $p<0.05$ in replication cohort as well as being validated) are in ***bold italics.***

Nearest Gene	SNP	Chr	Position (bp)	P-value in pooled GWAS	P-value in validation	P-value in replication
C1orf94	rs771392	1	34521402	1.64E-08	0.1023	
ADORA3	rs1361133	1	111828556	4.39E-06	0.0229	
FLJ36116	rs4838968	1	113161960	1.38E-11	**0.0005**	0.4426
OSR1	rs1727177	2	19356060	5.94E-08	**0.0004**	0.6836
LRP1B	rs10496894	2	142222836	3.41E-05	0.0749	
ADAM23	rs13420297	2	207165510	7.63E-03	0.0370	
ADAM23	rs10490745	2	207174805	9.51E-05	0.0138	
ADAM23	rs13396148	2	207192617	3.08E-06	0.0370	
ATP8A1	rs3749530	4	42287997	7.30E-08	**0.0004**	0.6746
ATP8A1	rs7681922	4	42300164	9.34E-05	**0.0002**	0.8880
CHSY3	rs12188544	5	129768423	4.93E-05	0.0274	
CHSY3	rs6893341	5	129846454	5.65E-04	0.0274	
CHSY3	rs10520081	5	129862210	3.25E-05	0.0292	
KRT18P50	rs567149	6	96542399	3.28E-07	0.0128	
MTHFD1L	rs1771798	6	151323260	5.83E-07	**0.0007**	0.4095
ARID1B	rs17087267	6	156627585	1.19E-08	**0.0019**	0.2617
ARID1B	rs9480347	6	156910347	8.13E-05	0.0167	
ARID1B	rs6927999	6	156913513	8.67E-03	0.0135	
HGF	rs7799610	7	81128458	2.31E-06	0.0518	
HGF	rs1014091	7	81246485	4.99E-05	**0.0084**	***0.0083***
NUB1	rs6963048	7	150700229	6.55E-06	**4.22E-05**	0.1605
ZFHX4	rs4548159	8	77496519	9.22E-06	**0.0050**	0.0763
SDC2	rs1020344	8	97526709	6.41E-05	**0.0079**	0.2569
PTPRD	rs1331660	9	9029222	7.17E-08	0.0102	
COL27A1	rs10817573	9	115947269	2.91E-06	**0.0001**	0.8186
CDH23	rs10999876	10	72982127	7.93E-03	0.0185	
CTSC	rs520864	11	87623983	7.75E-03	0.1778	
SOX5	rs10842183	12	23624709	6.05E-08	0.0863	
CS	rs744051	12	54953565	3.24E-06	**0.0066**	0.2683
PAN2	rs2291361	12	55004072	1.07E-05	**0.0066**	0.2683
PAN2	rs2643626	12	55012785	9.91E-03	0.0196	
IL23A	rs11171806	12	55019798	7.18E-03	0.0059	
TMEM132C	rs7959567	12	127755803	8.96E-10	0.0188	
LOC432369	rs9512928	13	27411301	1.90E-08	0.0200	
ABCC4	rs17189540	13	94706508	1.58E-03	0.0336	
ABCC4	rs17268275	13	94710845	6.60E-05	0.0336	
ACTN1	rs1950712	14	68550317	9.68E-03	0.0259	
FBN1	rs16961065	15	46593831	6.71E-03	0.0550	
FBN1	rs2118181	15	46703176	3.46E-03	0.0820	
CHSY1	rs12148369	15	99584813	1.06E-03	0.0096	
CDH11	rs16967999	16	63486525	1.04E-05	**$2.30x10^{-5}$**	0.5215
CDH11	rs185394	16	63527844	2.10E-04	**$1.83x10^{-5}$**	0.6040
CDH11	rs257341	16	63554514	8.75E-03	**$4.18\ x10^{-5}$**	0.7943

CDH11	rs35195	16	63583219	6.44E-04	**2.00×10^{-5}**	0.7170
CDH11	rs257490	16	63609142	3.45E-03	**3.33×10^{-5}**	0.7170
CDH1	rs3785076	16	67408284	3.87E-06	0.0172	
KC6	rs2848758	18	37355417	5.59E-03	0.1026	
NETO1	rs9963971	18	69047309	4.99E-06	0.0164	
MACROD2	rs200744	20	15542367	7.41E-07	**0.0001**	0.7974
ZHX3	rs6016541	20	39369419	3.14E-06	0.0315	
NCAM2	rs2826767	21	21582667	1.90E-05	0.0158	
TIMP3	rs11703156	22	31559804	9.48E-04	0.0355	

Supplementary Figure 1: Flowchart depicting genotyping strategies and cohorts utilised in the two independent genetic studies.