

Supplementary Figure S1: Chromosome 7 HLODs scores for gene-based two-point linkage analysis.

(A) the gene-based HLOD scores using only the rare variants (MAF >= 0.05) and (B) the gene-based HLOD scores using all variants. The lines at 3.3 and 1.9 represent the respective significant and suggestive thresholds as suggested by Lander and Kruglyak

Supplementary Figure S2: Heat map of mean ocular expression values for significant and suggestive genes from variant-based two-point linkage analysis.

CHR	POS	НГОД	GENE	Choroid. PLIER	Ciliary_body.PLIER	Cornea. PLIER	Iris. PLIER	Lens. PLIER	Optic_nerve. PLIER	Optic_nerve_head.PLIER	Retina. PLIER	Sclera. PLIER	Trabecular_meshwork.PLIER
7	25997536	4.34	NFE2L3	23.0	4.8	7.9	31.0	17.0	12.1	18.4	19.8	10.4	7.4
7	28754095	3.01	CREB5	51.6	46.0	32.7	36.9	67.9	96.4	113.0	44.6	49.9	47.4
7	27562660	2.82	HIBADH	53.5	55.9	67.2	51.1	91.0	70.7	74.7	59.8	50.6	108.2
7	27562660		EVX1	27.3	44.5	33.6	28.4	34.9	34.0	42.4	27.1	38.0	36.4
20	23424613		CSTL1	66.8	45.2	77.5	31.0	68.8	85.4	76.8	71.4	96.6	73.3
2	41792845		SLC8A1	20.6	16.7	10.8	21.9	19.7	28.8	33.3	54.9	30.8	22.2
7	33959239		BMPER	41.3	35.9	47.6	27.2	27.6	26.8	34.6	29.4	41.9	49.9
9	117118379		AKNA	34.5	35.8	35.0	30.8	40.0	40.3	40.3	41.8	43.4	34.3
7	31877261		PDE1C	57.7	21.7	13.0	39.5	32.3	76.8	71.9	30.8	71.8	46.9
7	36438709		ANLN	8.6	11.9	34.3	7.4	8.1	374.1	377.0	8.7	7.5	9.8
7	32611392		AVL9	25.4	17.4	45.8	25.3	39.3	38.4	44.3	43.2	23.2	42.3
6	152708310		SYNE1	39.4	42.4	52.4	27.2	31.0	46.4	73.2	73.0	54.1	70.5
1	26607726		SH3BGRL3	59.4	66.7	133.1	56.6	28.2	57.2	50.8	33.5	49.4	34.5
1 19	26612460		UBXN11	35.9	41.1	39.0	26.1	28.2	45.9	34.1	33.5	49.4	34.5
10	53086032 5920244		ZNF701 ANKRD16	15.9 35.2	7.7 27.4	39.0 32.5	3.3 28.3	22.1 25.2	12.5 28.9	6.8 28.6	17.3 22.8	12.6 33.9	18.1 33.7
18	3458997		TGIF1	42.4	38.3	46.0	28.7	40.7	37.2	30.9	26.5	42.6	43.5
	160610124		SLC22A2	22.3	33.7	22.5	23.6	22.4	30.9	35.8	28.5	22.0	24.0
6	160610124		SLC22A2	44.1	44.0	46.9	31.1	37.0	45.8	46.0	43.7	43.1	41.7
7	29647662		PRR15	66.2	97.1	76.2	46.7	51.5	66.4	71.5	70.5	65.6	64.9
7	24571485		MPP6	29.8	22.8	128.6	19.1	281.5	83.7	105.8	42.2	63.3	50.4
7	24571485	2.15		56.7	112.8	87.9	73.9	86.9	111.9	109.3	89.6	118.1	112.2
7	31178749		NEUROD6	11.3	19.4	9.4	21.7	9.3	14.6	10.4	14.3	16.6	8.2
7	31178749		ADCYAP1R1	37.5	31.2	42.2	33.3	30.8	65.0	70.0	42.9	40.8	38.6
1	1223385	2.13	SCNN1D	34.5	38.4	37.0	32.5	38.5	42.2	42.3	36.8	42.5	36.8
6	167438292	2.12	FGFR1OP	30.7	28.7	37.4	34.2	29.2	38.6	45.8	51.9	26.9	36.0
7	9266388	2.11	NXPH1	26.1	34.4	28.0	26.8	29.8	32.9	33.0	35.8	39.6	30.0
7	35423720	2.11	HERPUD2	59.7	60.6	62.2	44.5	120.8	59.5	81.8	61.6	55.3	59.5
2	73868446	2.11	NAT8	16.5	24.1	22.7	11.1	22.1	18.0	21.8	18.7	14.7	19.6
3	11400019	2.10	ATG7	34.3	27.0	37.8	24.1	30.2	34.0	46.1	35.2	40.5	49.3
7	26695215	2.08	SKAP2	64.6	35.9	29.6	28.1	69.3	43.1	64.1	36.9	75.7	40.9
9	37638744	2.05	томм5	245.6	306.6	107.3	272.8	300.6	86.8	94.6	80.4	112.8	210.6
9	37638744		FRMPD1	33.4	22.3	32.8	24.5	42.0	28.1	26.7	143.8	26.8	26.5
5	13769974		DNAH5	13.9	16.1	13.1	11.2	13.2	15.9	20.0	14.0	13.8	12.9
7	24311069		STK31	16.9	17.1	22.0	18.7	13.4	19.1	16.9	16.1	15.5	16.5
7	22929061		FAM126A	74.1	30.6	71.4	34.5	19.0	35.6	69.8	43.2	88.0	124.0
10	129903016		MKI67	17.2	15.4	34.4	12.1	17.6	18.0	16.3	15.0	24.4	16.6
1	36764770		THRAP3	116.1	106.7	173.0	69.5	162.7	162.6	195.9	131.7	112.2	173.5
20	36956026	1.92		23.1	22.1	24.2	21.6	23.7	26.9	26.9	23.0	25.3	22.2
7	33571828	1.91	BBS9	50.8	26.3	37.7	28.3	50.4	56.8	76.2	172.9	39.3	37.8

Genes are from the variant-based linkage analysis and were ranked by HLOD scores. The columns represent the chromosome, position, LOD and HLOD scores, gene symbol and ten eye tissues. Scale and color intensity represent PLIER values as reported in the ocular tissue database.

Supplementary Figure S3: Heat map of mean ocular expression values for significant and suggestive genes from gene-based two-point linkage analysis.

CHR	SOd	НГОД	GENE	Choroid. PLIER	Gliary_body.PLIER	Cornea.PLIER	Iris. PLIER	Lens. PLIER	Optic_nerve.PLIER	Optic_nerve_head. PLIER	Retina. PLIER	Sciera. PLIER	Trabecular_meshwork.PLIER
7	24571485		MPP6	29.8	22.8	128.6	19.1	281.5	83.7	105.8	42.2	63.3	50.4
7	24571485	2.15		56.7	112.8	87.9	73.9	86.9	111.9	109.3	89.6	118.1	112.2
7	31178749		NEUROD6	11.3	19.4	9.4	21.7	9.3	14.6	10.4	14.3	16.6	8.2
7	31178749		ADCYAP1R1	37.5	31.2	42.2	33.3	30.8	65.0	70.0	42.9	40.8	38.6
1	1223385		SCNN1D	34.5	38.4	37.0	32.5	38.5	42.2	42.3	36.8	42.5	36.8
6	167438292		FGFR1OP	30.7	28.7	37.4	34.2	29.2	38.6	45.8	51.9	26.9	36.0
7	9266388		NXPH1	26.1	34.4	28.0	26.8	29.8	32.9	33.0	35.8	39.6	30.0
7	35423720		HERPUD2	59.7	60.6	62.2	44.5	120.8	59.5	81.8	61.6	55.3	59.5
2	73868446		NAT8	16.5	24.1	22.7	11.1	22.1	18.0	21.8	18.7	14.7	19.6
3	11400019 26695215		ATG7 SKAP2	34.3 64.6	27.0 35.9	37.8 29.6	24.1	30.2	34.0	46.1 64.1	35.2 36.9	40.5	49.3 40.9
-	37638744		TOMM5	245.6	35.9	29.6 107.3	28.1 272.8	69.3 300.6	43.1 86.8	94.1		75.7 112.8	
9	37638744		FRMPD1	33.4	22.3	32.8	272.8	42.0	28.1	26.7	80.4 143.8	26.8	210.6 26.5
5	13769974		DNAH5	13.9	16.1	13.1	11.2	13.2	15.9	20.7	143.8	13.8	12.9
7	24311069		STK31	16.9	10.1	22.0	11.2	13.2	13.9	16.9	14.0	15.8	12.9
7			FAM126A	74.1	30.6	71.4	34.5	19.0	35.6	69.8	43.2	88.0	124.0
10			MKI67	17.2	15.4	34.4	12.1	19.0	18.0	16.3	45.2	24.4	16.6
10	36764770		THRAP3	116.1	106.7	173.0	69.5	162.7	162.6	195.9	131.7	112.2	173.5
20	36956026	1.92		23.1	22.1	24.2	21.6	23.7	26.9	26.9	23.0	25.3	22.2
7			BBS9	50.8	26.3	37.7	28.3	50.4	56.8	76.2		39.3	37.8
	33371628	1.91	9039	50.8	20.3	57.7	20.3	50.4	50.8	70.2	172.9	59.5	57.8

Genes are from the gene-based linkage analysis and were ranked by HLOD scores. The columns represent the chromosome, position, LOD and HLOD scores, gene symbol and ten eye tissues. Scale and color intensity represent PLIER values as reported in the ocular tissue database.