The American Journal of Human Genetics, Volume *89*

Supplemental Data

GWAS Findings for Human Iris Patterns: Associations with Variants in Genes that Influence Normal Neuronal Pattern Development

Mats Larsson, David L. Duffy, Gu Zhu, Jimmy Z. Liu, Stuart Macgregor, Allan F. McRae, Margaret J. Wright, Richard A. Sturm, David A. Mackey, Grant W. Montgomery, Nicholas G. Martin, and Sarah E. Medland

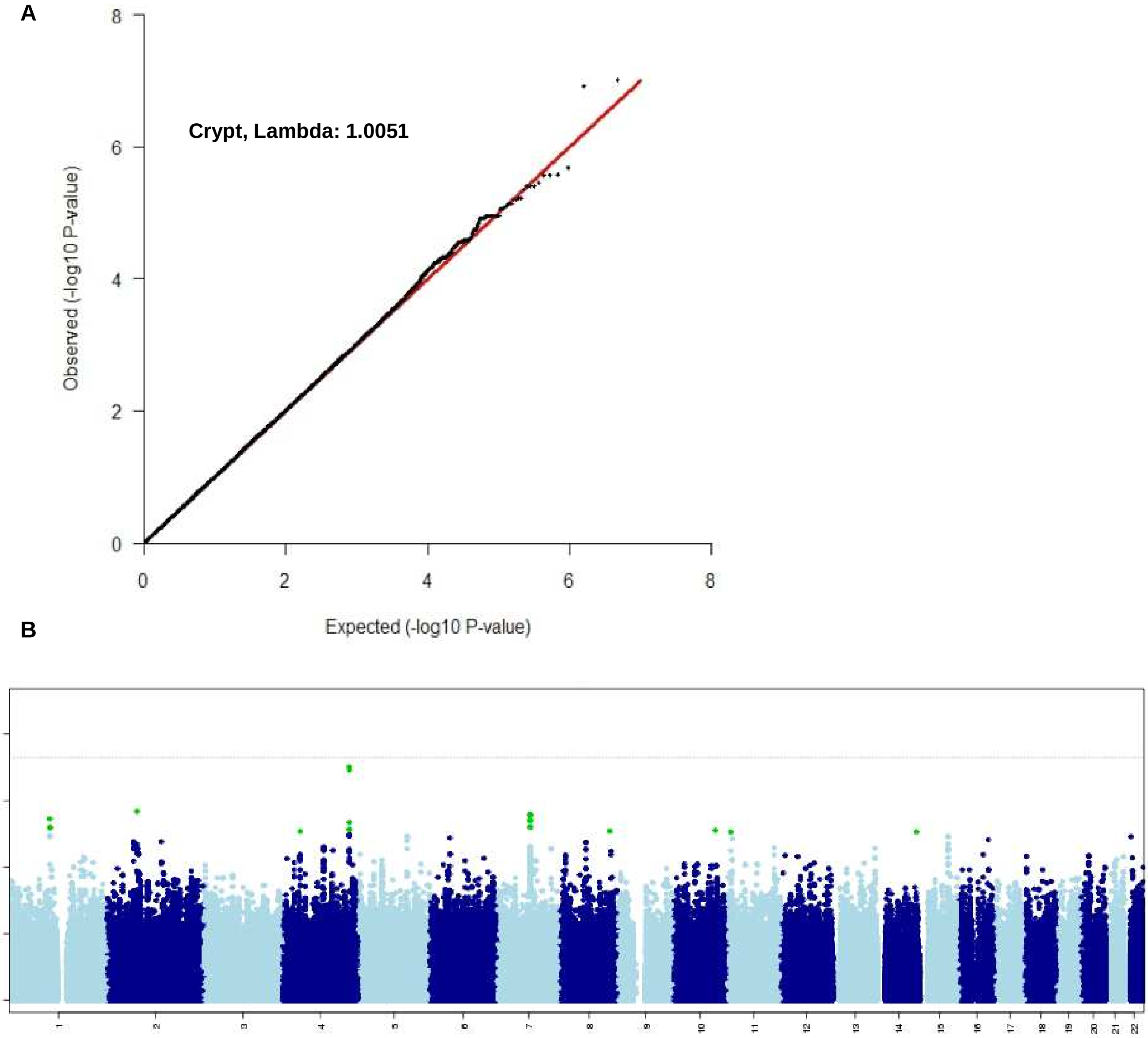

Figure S1. Q-Q and Manhattan plots for GWAS results for crypts frequency in the discovery sample, N=2121 Areas of suggestive association (1.0e-5) shown in green: 1p21.1 (rs10874518, p=3.5e-6); 2p13.1 (rs205611, p=2.1e-06); 4q32.3 (rs13140875, p=9.7e-08), 4p12 (rs6836590, p= 8.3e-06); 7q21.11 (rs10235789, p=2.7e-06); 8q24.13 (rs13262017, p=8.2e-06); 10q25.1 (rs4564255, p=7.8e-06); 11p15.4 (rs12272194, p=8.7e-06) and 14q32.2 (rs4905757, p=8.6e-06).

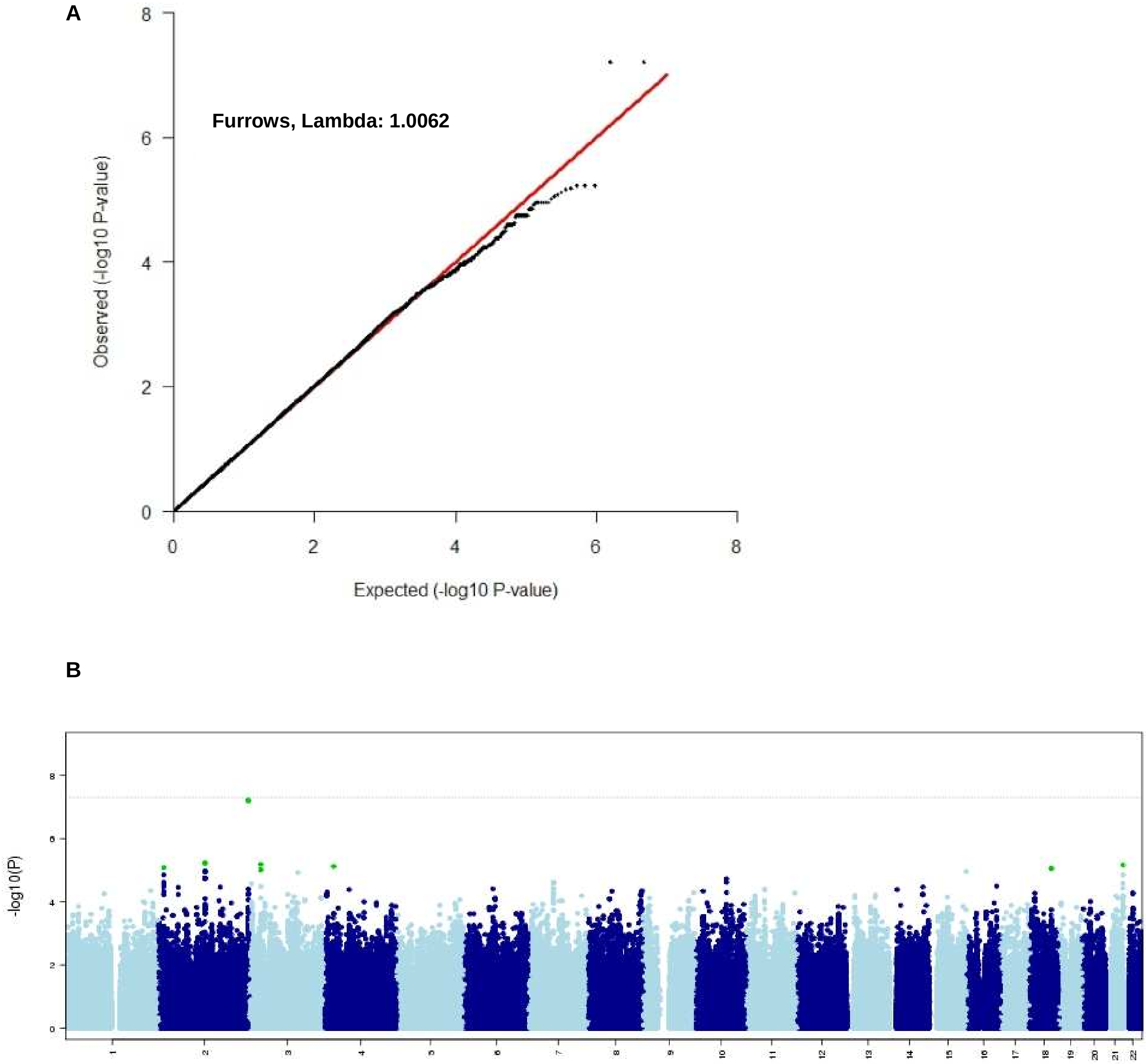

Figure S2. Q-Q and Manhattan plots for GWAS results for furrows in the discovery sample, N=2121

Areas of suggestive association (1.0e-5) shown in green: 2q37.3 (rs3739070, p=6.2e-08), 2q14.3 (rs17009210, p=6.0e-06), 2p24.3 (rs7568997, p=8.3e-06); 3p24.1 (rs2033702, p=6.6e-06); 4p15.2 (rs4312745, p=7.6e-06), 18q21.33 (rs10469238, p=8.9e-06) and 21q22.3 (rs13047753, p= 6.9e-06).

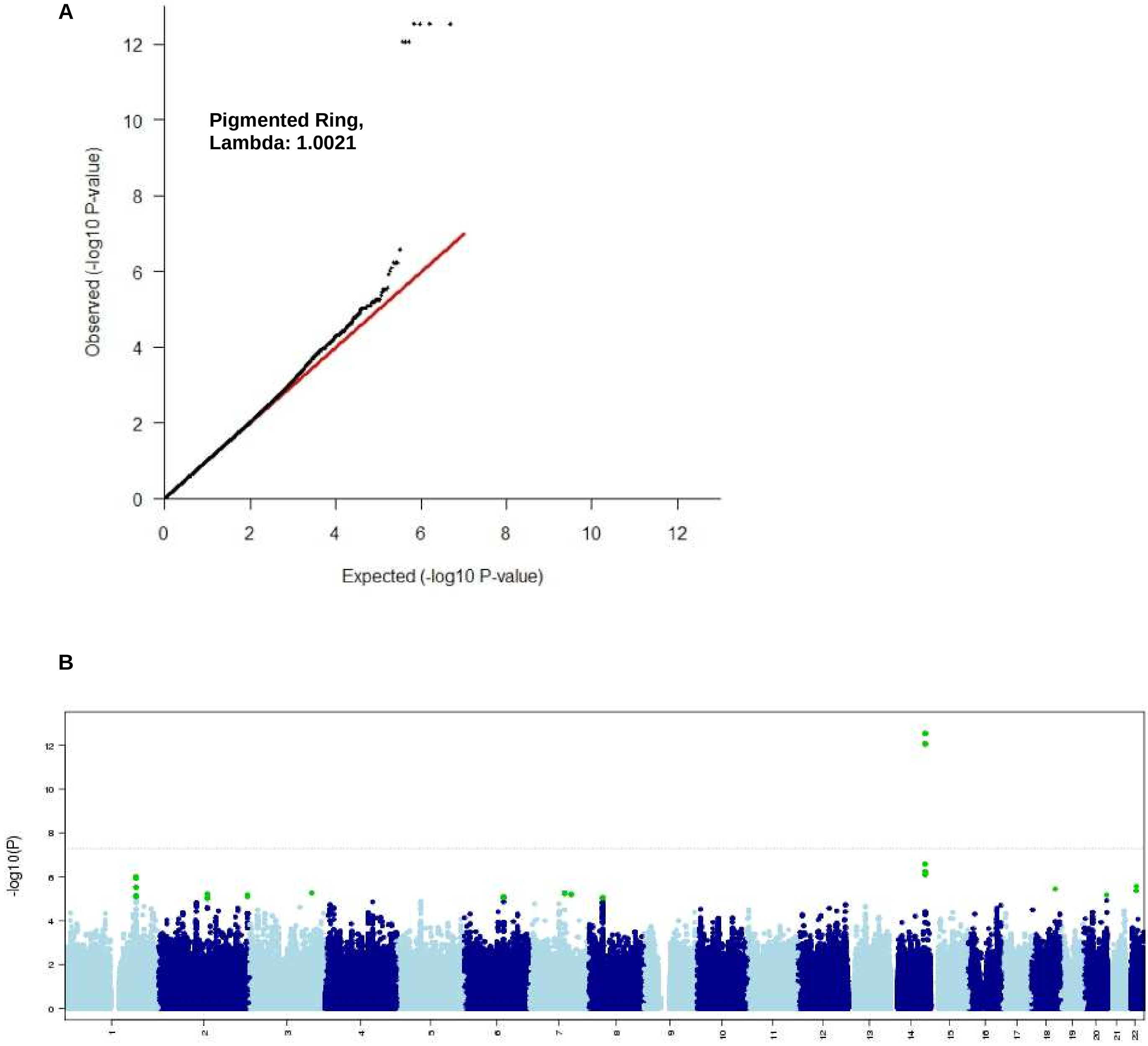

Figure S3. Q-Q and Manhattan plots for GWAS results for pigmented ring in the discovery sample, N=2121

Areas of suggestive association (1.0e-5) shown in green: 1q31.1 (rs2419670, p=1.2e-06); 2q21.1 (rs13400337, p=6.1e-06), 2q37.3 (rs4542797, p=6.7e-06); 3q26.1 (rs13081597, p=5.5e-06); 6q16.3 (rs9322729, p=8.2e-06); 7q21.3 (rs10808093, p=5.6e-06), 7q31.1 (rs6957003, p=6.4e-06); 8p11.23 (rs431626, p=8.8e-06); 14q32.12 (rs4900109, p=2.9e-13); 18q22.1 (rs9958293, p=3.6e-06); 20q13.33 (rs6061885, p=6.7e-06) and 22q12.2 (rs12159546, p=2.7e-06).

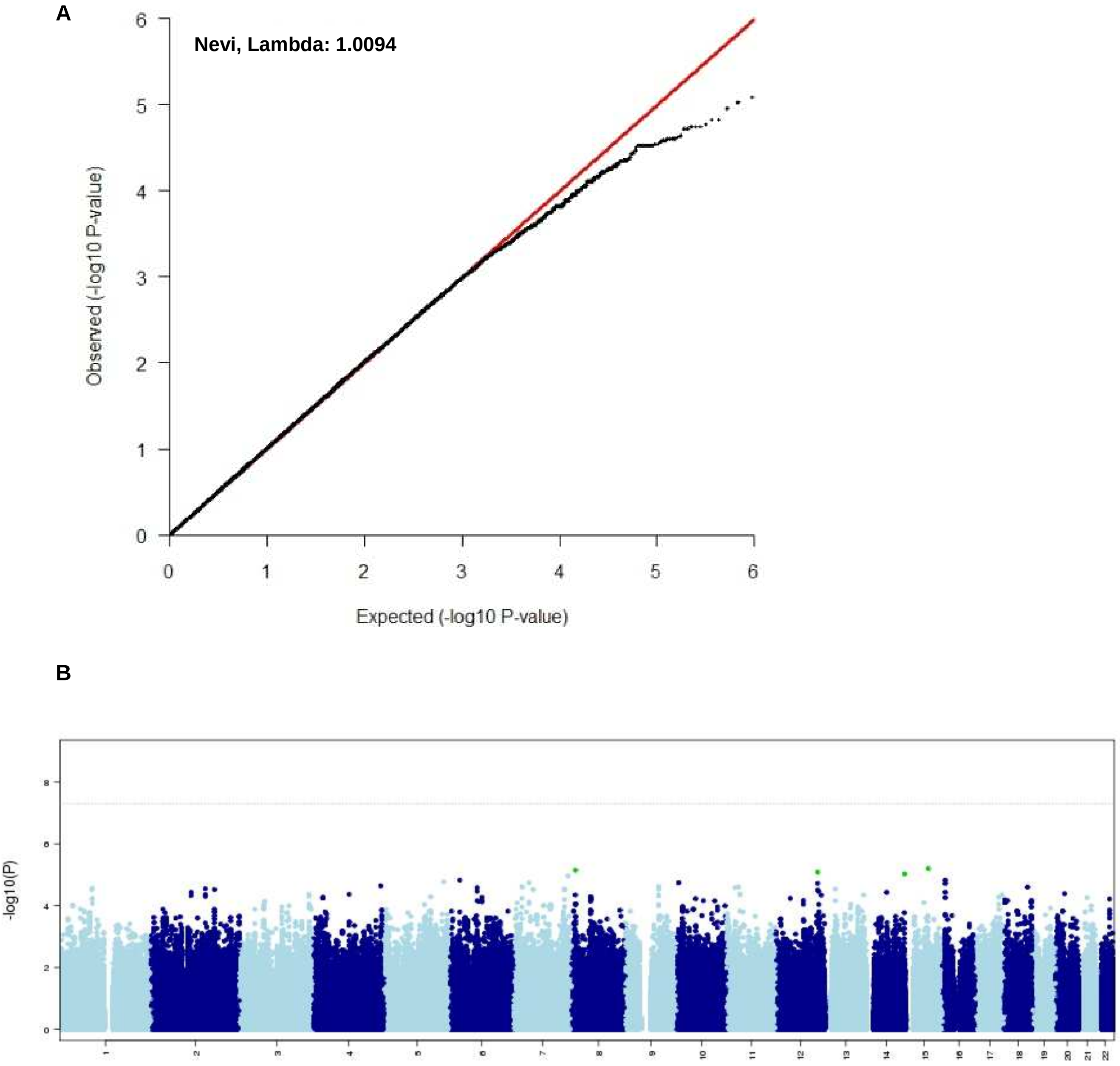

Figure S4. Q-Q and Manhattan plots for GWAS results for frequency of iris nevi in the discovery sample, N=2121

Areas of suggestive association (1.0e-5) shown in green: 8p23.1 (rs7822804, p=7.1e-06); 12q24.21 (rs11067162, p=8.2e-06); 14q32.33 (rs3212134, p=9.4e-06) and 15q22.31 (rs11630290, p=6.3e-06).

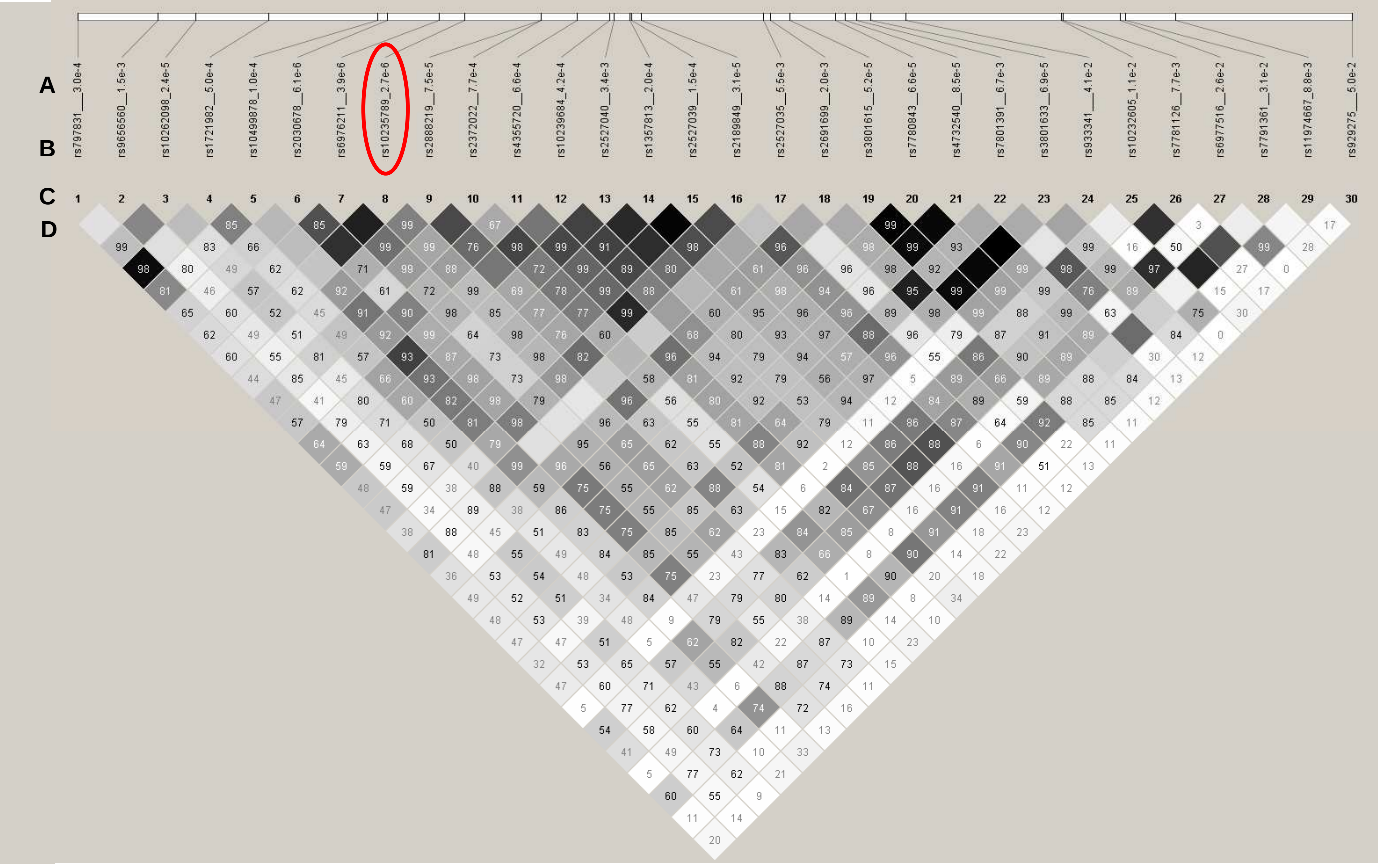

Figure S5. LD plot for SNPs nominally associated with crypts in *SEMA3A* surrounding rs10235789

The gene based test (VEGAS) included 379 SNPs in *SEMA3A*, 187 of these were nominally associated with crypts ($p < .05$). (A) P-value for the SNPs. (B) rs-number for the SNPs. (C) SNP order in the LD plot, the most strongly associated SNP (rs10235789, p=2.7e-6) is circled in red; (D) LD plot showing r^2 between 30 SNPs in *SEMA3A* within the discovery sample (N = 920).

Table S1. The test-retest and inter-rater reliability of iris characteristics

Scale	Number of Categories	Number of raters / Observations	Polychoric Reliability	Polychoric Correlation Between Right-Left Eye
Crypts	5	1/400	0.91	0.91
Furrows	5	1/400	0.94	0.94
Pigment Ring	3	2/982	0.94	0.95
Iris Nevi	4	1/400	0.96	0.94

Table S2. Distribution of Iris phenotypes in the discovery and replication samples

Sample	Mean Age (Std. Dev.)	Age Range	No. of Females	No. of Males	Total	Percentage of Subjects % in Each Category																
						Crypts					Furrows					Pigment Ring			Iris Nevi			
						1	2	3	4	5	1	2	3	4	5	1	2	3	1	2	3	4
GWAS	14.09 (5.36)	5 - 50	1126	995	2121	10.0	53.7	21.7	12.6	2.0	46.7	28.4	15.7	7.1	2.1	36.4	36.6	27.0	58.7	32.4	5.1	3.8
Rep 1	13.50 (1.96)	9 - 20	254	245	499	14.0	55.2	17.0	10.2	3.6	42.2	30.3	17.3	8.0	2.2	32.3	33.7	34.0	65.3	25.1	7.2	2.4
Rep 2	10.76 (4.14)	5 - 19	30	43	73	9.6	48.0	31.5	6.8	4.1	45.2	23.3	24.7	4.1	2.7	37.0	32.9	30.1	58.9	32.9	4.1	4.1

Table S3. Correlations between iris characteristics in the discovery sample for males (N = 995) and females (N = 1126)

	Males Below Diagonal / Females Above Diagonal					
Scale	**Eye Color**	**Crypt**	**Furrows**	**Pigment R.**	**Iris Nevi**	**Age**
Eye color		-0.14**	0.47**	0.55**	0.21**	0.04
Crypts	-0.13**		-0.23**	-0.13**	0.01	0.00
Furrows	0.48**	-0.29**		0.40**	0.13**	-0.04
Pigment Ring	0.60**	-0.17**	0.43**		0.28**	0.03
Iris Nevi	0.26**	0.04	0.16**	0.27**		0.24**
Age	0.07*	0.01	0.07*	0.04	0.15**	

* p < .05. ** p < .01. The correlation between sex (females coded 1, males coded 2) and crypt and furrows, are .17**, -.06** respectively. The correlation between sex and the other iris characteristics where not significant, p > .05.

Table S4. Estimated heritability of iris characteristics for the best-fitting ADE model with 95% lower and upper confidence intervals (thresholds are corrected for age and sex).

Characteristic	**No. of Categories**	**Additive Genetic Variance A (95% CI)**	**Non-additive Genetic Variance D (95% CI)**	**Non-shared Environmental Variance E (95% CI)**	**MZ Correlation 419 pairs**	**DZ Correlation 700 pairs**
Crypts	5	36% (11-60%)	48% (23-73%)	16% (13-20%)	0.76	0.24
Furrows	5	78% (52-88%)	9% (0-27%)	13% (10-17%)	0.81	0.35
Pigment Ring	3	46% (17-72%)	47% (20-75%)	7% (5-10%)	0.85	0.31
Iris Nevi	4	51% (17-72%)	20% (0-52%)	29% (22-36%)	0.57	0.25

Table S5. The 50 genes most strongly associated with crypts: VEGAS results for the discovery sample GWAS, N =2121

	Test statistics for the genes most strongly associated with Crypts					Test Statistics for the SNP most strongly associated within each gene					
#	**Gene**	**Position**	**#Simulations**	**#SNPs**	**P-value**	**SNP**	**P-value**	**Allele**	**beta**	**SE**	**r^2**
1	*SEMA3A*	7q21.11	1.00e+06	379	3.00e-06	rs10235789	2.70e-06	C	0.098	0.021	1.27%
2	*STRA6*	15q24.1	1.00e+06	87	0.00034	rs11858661	0.00075	C	0.080	0.024	0.65%
3	*ICA1L*	2q33.2	1.00e+06	59	0.00039	rs12053074	0.00043	G	-0.078	0.022	0.72%
4	*HSPA8*	11q24.1	1.00e+06	90	0.00046	rs10892958	0.00044	C	0.091	0.026	0.67%
5	*CCNL1*	3q25.31	1.00e+06	69	0.00050	rs2133952	0.00096	C	0.069	0.021	0.58%
6	*C1orf173*	1p31.1	1.00e+06	196	0.00052	rs7541513	0.00023	G	-0.077	0.021	0.77%
7	*VEPH1*	3q25.31	1.00e+06	364	0.00059	rs1587534	0.00096	A	0.069	0.021	0.58%
8	*IL1R2*	2q11.2	1.00e+06	198	0.00065	rs4851531	0.00037	C	0.076	0.021	0.73%
9	*DISP2*	15q15.1	1.00e+06	78	0.00070	rs875767	0.00023	C	0.113	0.031	0.76%
10	*TMEM160*	19q13.32	1.00e+06	26	0.00072	rs8101091	0.00042	G	-0.082	0.023	0.69%
11	*EPHA4*	2q36.1	1.00e+06	269	0.00074	rs4674596	0.00026	C	-0.076	0.021	0.74%
12	*TBC1D21*	15q24.1	1.00e+06	90	0.00077	rs1478558	1.20e-05	G	0.090	0.021	1.08%
13	*TRIM29*	11q23.3	1.00e+06	115	0.00079	rs470411	0.00075	C	0.080	0.024	0.64%
14	*CACNA1E*	1q25.3	1.00e+06	392	0.00083	rs3753737	7.00e-05	A	-0.091	0.023	0.93%
15	*CYP2D6*	22q13.2	1.00e+06	54	0.00084	rs742086	0.00034	T	0.083	0.023	0.73%
16	*ALS2CR13*	2q33.2	1.00e+06	62	0.00085	rs7577225	0.00071	C	-0.072	0.021	0.68%
17	*BPTF*	17q24.2	1.00e+06	59	0.00094	rs6504550	0.00074	A	-0.082	0.024	0.69%
18	*NPAS1*	19q13.32	1.00e+06	33	0.00098	rs2303107	0.00101	A	-0.074	0.023	0.59%
19	*C17orf58*	17q24.2	1.00e+06	14	0.00102	rs9891146	0.00102	C	-0.080	0.024	0.66%
20	*HPX*	11p15.4	1.00e+06	60	0.00103	rs2723647	0.0022	A	0.077	0.025	0.54%
21	*WNT7B*	22q13.31	1.00e+06	1	0.00111	rs6519955	0.0011	G	0.070	0.022	0.62%
22	*ISLR*	15q24.1	1.00e+06	65	0.00115	rs11639369	0.00117	G	-0.085	0.026	0.63%
23	*WEE1*	11p15.4	1.00e+06	66	0.00155	rs12272194	8.70e-06	T	-0.115	0.026	1.18%
24	*GPER*	7p22.3	1.00e+06	50	0.00178	rs12112166	0.00094	C	-0.069	0.021	0.63%
25	*C21orf2*	21q22.3	1.00e+06	67	0.00195	rs2236669	4.80e-05	T	-0.087	0.021	0.97%
26	*DNMT3A*	2p23.3	1.00e+06	84	0.00225	rs13420827	0.00011	C	0.104	0.027	0.86%
27	*CDC25A*	3p21.31	1.00e+06	34	0.00242	rs10446426	0.00021	G	0.079	0.021	0.75%
28	*NAGA*	22q13.2	1.00e+06	43	0.00243	rs2854827	0.00049	G	0.089	0.026	0.72%
29	*CD248*	11q13.2	1.00e+06	34	0.00248	rs479018	0.0023	G	-0.066	0.022	0.53%
30	*BCCIP*	10q26.2	1.00e+06	68	0.00337	rs4385801	0.00109	G	0.069	0.021	0.63%
31	*TMCO2*	1p34.2	1.00e+06	40	0.00349	rs12144950	0.00048	A	-0.093	0.027	0.71%
32	*COX6B1*	19q13.12	1.00e+06	50	0.00404	rs7254601	0.00024	A	-0.089	0.024	0.78%
33	*C7orf50*	7p22.3	1.00e+05	121	0.00136	rs2030958	0.00049	G	0.219	0.063	0.67%
34	*LYSMD4*	15q26.3	1.00e+05	92	0.00155	rs8041078	0.0022	G	0.097	0.032	0.52%
35	*MAP4K4*	2q11.2	1.00e+05	159	0.00177	rs3771903	0.00028	A	-0.076	0.021	0.76%
36	*OR10Z1*	1q23.1	1.00e+05	88	0.00178	rs12601	0.00031	A	0.075	0.021	0.73%
37	*KCNS2*	8q22.2	1.00e+05	66	0.00182	rs17370425	0.00029	C	0.113	0.031	0.76%
38	*ARMC4*	10p12.1	1.00e+05	216	0.00186	rs12218330	1.00e-04	A	-0.121	0.031	0.82%
39	*TMEM16H*	19p13.11	1.00e+05	51	0.00188	rs971694	0.00027	C	0.116	0.032	0.76%
40	*KIAA0947*	5p15.32	1.00e+05	183	0.00191	rs4702271	0.00016	T	-0.322	0.085	0.80%
41	*C8orf34*	8q13.2	1.00e+05	518	0.00200	rs11989456	0.00057	G	-0.083	0.024	0.66%
42	*CDK6*	7q21.2	1.00e+05	151	0.00215	rs11974095	0.00016	T	0.154	0.041	0.80%
43	*COX19*	7p22.3	1.00e+05	37	0.00237	rs10272099	0.00104	T	0.076	0.023	0.66%
44	*C15orf52*	15q15.1	1.00e+05	66	0.00248	rs6492944	0.0009	T	0.099	0.030	0.62%
45	*MEP1A*	6p12.3	1.00e+05	156	0.00272	rs1059276	0.00013	A	-0.083	0.022	0.85%
46	*SCN7A*	2q24.3	1.00e+05	176	0.00274	rs10930218	0.00024	T	-0.08	0.022	0.77%
47	*TPRX1*	19q13.33	1.00e+05	102	0.00322	rs11083901	0.00119	G	0.08	0.025	0.61%
48	*WNT11*	11q13.5	1.00e+05	129	0.00333	rs660981	8.10e-05	A	-0.082	0.021	0.87%
49	*PTX3*	3q25.32	1.00e+05	116	0.00344	rs1456110	0.0032	A	-0.06	0.020	0.48%
50	*CA11*	19q13.33	1.00e+05	41	0.00347	rs433852	0.0017	C	-0.081	0.026	0.54%

Shaded SNPs are also among the 50 best Krasanova[55] genes for crypts.

Table S6. The 50 genes most strongly associated with furrows: VEGAS results for the discovery sample GWAS, N =2121

	Test statistics for the genes most strongly associated with Furrows					Test Statistics for the SNP most strongly associated within each gene					
#	**Gene**	**Position**	**#Simulations**	**#SNPs**	**P-value**	**SNP**	**P-value**	**Allele**	**beta**	**SE**	**r^2**
1	*FANCF*	11p14.3	1.00e+06	66	9.10e-05	rs10833798	0.00006	A	0.127	0.032	0.92%
2	*HEPHL1*	11q21	1.00e+06	146	0.000149	rs4753115	0.00041	G	-0.086	0.024	0.71%
3	*MAPKAPK5*	12q24.12	1.00e+06	47	0.000402	rs7296651	0.00038	C	-0.115	0.032	0.72%
4	*TRIM62*	1p35.1	1.00e+06	64	0.000508	rs3912145	0.00014	T	-0.106	0.028	0.83%
5	*ALDH2*	12q24.12	1.00e+06	33	0.000512	rs440	0.00014	T	-0.118	0.031	0.86%
6	*USO1*	4q21.1	1.00e+06	121	0.000586	rs10028884	0.00037	G	-0.129	0.036	0.71%
7	*BRAP*	12q24.12	1.00e+06	31	0.000732	rs1544396	0.00046	T	-0.110	0.031	0.69%
8	*HIGD1B*	17q21.31	1.00e+06	69	0.000912	rs12947309	0.00230	C	0.082	0.027	0.54%
9	*PDE3B*	11p15.2	1.00e+06	136	0.001007	rs6486202	0.00023	C	0.091	0.025	0.80%
10	*TMEM116*	12q24.13	1.00e+06	67	0.001161	rs2339941	0.00028	G	-0.115	0.032	0.77%
11	*DIRC1*	2q32.2	1.00e+06	164	0.001228	rs1395864	0.00048	G	0.093	0.027	0.74%
12	*LAIR2*	19q13.42	1.00e+06	65	0.001281	rs8109847	0.00028	T	0.088	0.024	0.77%
13	*GFAP*	17q21.31	1.00e+06	104	0.001298	rs7225162	0.00035	T	-0.087	0.024	0.74%
14	*G3BP2*	4q21.1	1.00e+05	74	0.00135	rs1464071	0.00086	C	-0.123	0.037	0.63%
15	*SYT6*	1p13.2	1.00e+05	214	0.00154	rs2938334	0.00026	C	-0.128	0.035	0.75%
16	*OR4A47*	11p11.2	1.00e+05	62	0.00166	rs10838924	0.00004	T	0.183	0.045	1.13%
17	*TMCO7*	16q22.1	1.00e+06	139	0.001689	rs251100	0.00021	C	-0.094	0.025	0.85%
18	*ADAMTS5*	21q21.3	1.00e+05	173	0.00182	rs233896	0.00007	T	-0.097	0.024	0.91%
19	*CYP2R1*	11p15.2	1.00e+06	59	0.002114	rs1496167	0.00150	A	-0.084	0.027	0.56%
20	*COL6A1*	21q22.3	1.00e+06	94	0.002115	rs13047753	0.00001	G	0.164	0.036	1.27%
21	*FAM109A*	12q24.12	1.00e+05	33	0.00214	rs10774623	0.00095	A	-0.095	0.029	0.62%
22	*TPRX1*	19q13.33	1.00e+06	102	0.002153	rs11665715	0.00073	C	-0.097	0.029	0.66%
23	*ALDH3A2*	17p11.2	1.00e+05	66	0.00232	rs2386142	0.00064	G	-0.085	0.025	0.70%
24	*OR10J5*	1q23.2	1.00e+05	92	0.00236	rs4656837	0.00123	T	0.087	0.027	0.60%
25	*NUCB2*	11p15.1	1.00e+05	84	0.00269	rs7108315	0.00074	C	0.086	0.026	0.71%
26	*GJC1*	17q21.31	1.00e+05	75	0.00272	rs7223598	0.00230	T	0.082	0.027	0.54%
27	*MOBKL1B*	2p13.1	1.00e+05	77	0.00273	rs1667601	0.00023	G	0.090	0.025	0.79%
28	*ZDHHC21*	9p22.3	1.00e+06	275	0.002742	rs2382492	0.00014	G	0.096	0.025	0.88%
29	*LOC100131897*	5q35.1	1.00e+05	304	0.00287	rs30079	0.00008	C	-0.093	0.024	0.86%
30	*C2orf29*	2q11.2	1.00e+05	69	0.00302	rs13015194	0.00015	T	-0.109	0.029	0.82%
31	*RPS13*	11p15.1	1.00e+05	31	0.00312	rs17560341	0.00050	G	0.091	0.026	0.71%
32	*MEGF9*	9q33.2	1.00e+05	93	0.00314	rs735110	0.00133	A	-0.085	0.026	0.59%
33	*ADAD1*	4q27	1.00e+05	62	0.00323	rs10027161	0.00230	A	-0.135	0.044	0.53%
34	*PRMT6*	1p13.3	1.00e+05	85	0.00327	rs6675948	0.00038	C	-0.110	0.031	0.73%
35	*PDX1*	13q12.2	1.00e+05	70	0.00354	rs4769581	0.00033	C	-0.088	0.025	0.74%
36	*CEP192*	18p11.21	1.00e+05	142	0.00355	rs17604778	0.00066	C	0.093	0.027	0.70%
37	*HAMP*	19q13.12	1.00e+05	71	0.00356	rs928937	0.00160	A	0.080	0.025	0.59%
38	*ZNF704*	8q21.13	1.00e+05	168	0.00359	rs3907424	0.00026	C	0.133	0.036	0.83%
39	*SEH1L*	18p11.21	1.00e+05	88	0.00361	rs12605081	0.00096	G	-0.079	0.024	0.62%
40	*HAS3*	16q22.1	1.00e+06	34	0.003629	rs12325454	0.00095	T	0.109	0.033	0.63%
41	*CD200R2*	3q13.2	1.00e+05	132	0.0038	rs6775807	0.00010	C	-0.191	0.049	0.88%
42	*ZBTB10*	8q21.13	1.00e+05	96	0.00396	rs474402	0.00095	G	0.142	0.043	0.62%
43	*TMEM45A*	3q12.2	1.00e+05	135	0.00397	rs3924054	0.00006	G	0.198	0.049	0.98%
44	*MTERFD1*	8q22.1	1.00e+05	74	0.00431	rs2643343	0.00150	C	0.079	0.025	0.60%
45	*SASH1*	6q24.3	1.00e+05	327	0.00434	rs9399650	0.00030	T	0.093	0.026	0.74%
46	*PSMA1*	11p15.2	1.00e+05	107	0.00436	rs11023238	0.00025	T	-0.101	0.028	0.77%
47	*KCTD7*	7q11.21	1.00e+06	65	0.004417	rs10258739	0.00101	T	-0.080	0.024	0.64%
48	*FAM108C1*	15q25.1	1.00e+05	153	0.00442	rs2046389	0.00300	T	-0.105	0.035	0.52%
49	*SPAG9*	17q21.33	1.00e+05	98	0.00464	rs1860443	0.00021	C	0.099	0.027	0.81%
50	*SEMA3A*	7q21.11	1.00e+05	379	0.00465	rs10235789	0.00009	C	-0.095	0.024	0.86%

Shaded SNPs are also among the 50 best Krasanova[55] genes for furrows.

Table S7. The 50 genes most strongly associated with pigmented ring: VEGAS results for the discovery sample GWAS, N =2121

	Test statistics for the genes most strongly associated with Pigmented Ring					**Test Statistics for the SNP most strongly associated within each gene**					
#	**Gene**	**Position**	**#Simulations**	**#SNPs**	**P-value**	**SNP**	**P-value**	**Allele**	**beta**	**SE**	**r^2**
1	*GPR148*	2q21.1	1.00e+06	15	2.1e-05	rs13400337	6.10e-06	C	-0.473	0.104	1.29%
2	*SLC24A4*	14q32.12	1.00e+06	274	0.000143	rs4900109	2.90e-13	G	0.167	0.023	3.23%
3	*IL1R2*	2q11.2	1.00e+06	198	0.000146	rs1019031	0.00002	T	-0.101	0.023	1.08%
4	*ZNF30*	19q13.11	1.00e+06	69	0.000287	rs10422961	0.00030	C	0.087	0.024	0.79%
5	*GRSF1*	4q13.3	1.00e+06	56	0.000329	rs1020929	0.00043	C	-0.184	0.052	0.72%
6	*RUFY3*	4q13.3	1.00e+06	87	0.000428	rs7678779	0.00014	C	-0.203	0.053	0.84%
7	*GNG4*	1q42.3	1.00e+06	135	0.000449	rs10802790	0.00005	G	0.129	0.032	0.99%
8	*SLC25A40*	7q21.12	1.00e+06	51	0.000643	rs10225955	0.00091	C	-0.248	0.075	0.78%
9	*DUSP18*	22q12.2	1.00e+06	91	0.000679	rs12159546	0.00000	G	0.168	0.036	1.28%
10	*GUCA1C*	3q13.13	1.00e+06	205	0.000754	rs751298	0.00024	T	0.087	0.024	0.77%
11	*GLTPD1*	1p36.33	1.00e+06	10	0.000803	rs2296474	0.00320	G	0.266	0.090	0.45%
12	*ERAL1*	17q11.2	1.00e+06	34	0.000835	rs2242345	0.00040	T	0.099	0.028	0.72%
13	*OR10P1*	12q13.2	1.00e+06	80	0.000869	rs7978340	0.00030	G	0.081	0.022	0.75%
14	*FLJ20309*	2q33.3	1.00e+06	102	0.000921	rs6718656	0.00063	T	0.075	0.022	0.66%
15	*COL6A1*	21q22.3	1.00e+06	94	0.001214	rs13046217	0.00009	T	0.088	0.023	0.89%
16	*RUNDC3B*	7q21.12	1.00e+06	123	0.001278	rs10216040	0.00099	A	-0.244	0.074	0.76%
17	*MAP4K4*	2q11.2	1.00e+06	159	0.001335	rs7568226	0.00011	A	-0.096	0.025	0.84%
18	*C4orf37*	4q23	1.00e+06	494	0.001396	rs12233741	0.00060	C	-0.078	0.023	0.69%
19	*LIPT1*	2q11.2	1.00e+06	74	0.001666	rs13009147	0.00077	G	-0.077	0.023	0.63%
20	*NDST4*	4q26	1.00e+06	297	0.001702	rs7673947	0.00004	A	-0.150	0.037	0.94%
21	*ART5*	11p15.4	1.00e+06	117	0.001914	rs10834648	0.00003	A	0.098	0.023	0.95%
22	*CBR4*	4q32.3	1.00e+06	78	0.001925	rs11730894	0.00290	T	0.077	0.026	0.52%
23	*MUC5AC*	11p15.5	1.00e+06	70	0.002205	rs10902095	0.00030	C	-0.102	0.028	0.77%
24	*RPL23A*	17q11.2	1.00e+06	27	0.003348	rs4795456	0.00085	G	0.093	0.028	0.64%
25	*ZNF750*	17q25.3	1.00e+06	92	0.003757	rs3744159	0.00051	G	-0.086	0.025	0.68%
26	*LIN7B*	19q13.33	1.00e+06	60	0.005006	rs1009213	0.00086	A	0.078	0.023	0.62%
27	*GRAMD1A*	19q13.11	1.00e+06	64	0.006701	rs2651080	0.00032	G	0.089	0.025	0.77%
28	*ROCK1*	18q11.1	1.00e+05	40	0.00129	rs8085504	0.00057	G	0.183	0.053	0.64%
29	*SYCP3*	12q23.2	1.00e+05	85	0.00152	rs10492085	0.00015	G	-0.085	0.023	0.81%
30	*PES1*	22q12.2	1.00e+05	90	0.00171	rs2301955	0.00031	C	0.080	0.022	0.73%
31	*ANPEP*	15q26.1	1.00e+05	95	0.00223	rs16974181	0.00062	C	0.093	0.027	0.68%
32	*ANXA4*	2p13.3	1.00e+05	127	0.00254	rs7567400	0.00125	G	0.072	0.022	0.61%
33	*SLC6A12*	12p13.33	1.00e+05	128	0.00275	rs17692962	0.00113	A	0.075	0.023	0.61%
34	*PARL*	3q27.1	1.00e+05	125	0.00276	rs2056332	0.00028	T	-0.081	0.022	0.76%
35	*IGJ*	4q13.3	1.00e+05	33	0.00294	rs6813178	0.00069	G	-0.168	0.050	0.67%
36	*RAB39*	11q22.3	1.00e+05	66	0.003	rs12576453	0.00160	A	0.071	0.022	0.56%
37	*TRAF4*	17q11.2	1.00e+05	30	0.00313	rs12453504	0.00069	C	0.094	0.028	0.66%
38	*ZNF420*	19q13.12	1.00e+05	75	0.00316	rs11666786	0.00240	T	0.068	0.022	0.54%
39	*MED15*	22q11.21	1.00e+05	164	0.00319	rs165591	0.00021	G	-0.087	0.023	0.78%
40	*MED4*	13q14.2	1.00e+05	114	0.00327	rs2104480	0.00017	G	0.092	0.024	0.80%
41	*NPTX2*	7q22.1	1.00e+05	91	0.00334	rs817771	0.00017	A	0.097	0.026	0.90%
42	*ANGEL2*	1q32.3	1.00e+05	54	0.00349	rs3002284	0.00087	T	0.080	0.024	0.65%
43	*RNASEL*	1q25.3	1.00e+05	95	0.00352	rs11072	0.00005	A	-0.099	0.025	0.96%
44	*HRH3*	20q13.33	1.00e+05	84	0.00366	rs6062130	0.00001	C	0.116	0.027	1.16%
45	*RLN3*	19p13.12	1.00e+05	12	0.00384	rs7249702	0.00103	G	0.092	0.028	0.67%
46	*MYL3*	3p21.31	1.00e+05	42	0.00393	rs1077216	0.00020	C	-0.159	0.043	0.76%
47	*KLHL3*	5q31.2	1.00e+05	134	0.00397	rs757157	0.00013	C	-0.086	0.022	0.85%
48	*PON2*	7q21.3	1.00e+05	124	0.00437	rs10225097	0.00001	C	0.100	0.022	1.16%
49	*GABRG3*	15q12	1.00e+05	579	0.00444	rs3097489	0.00008	C	0.091	0.023	0.95%
50	*RAB32*	6q24.3	1.00e+05	56	0.00457	rs11155475	0.00230	G	-0.075	0.025	0.54%

Shaded SNPs are also among the 50 best Krasanova[55] genes for pigmented ring.

Table S8. The 50 genes most strongly associated with iris nevi: VEGAS results for the discovery sample, GWAS N =2121

	Test statistics for the genes most strongly associated with Nevi					Test Statistics for the SNP most strongly associated within each gene					
#	Gene	Position	#Simulations	#SNPs	P-value	SNP	P-value	Allele	beta	SE	r^2
1	*WIPI1*	17q24.2	1.00e+06	96	0.00025	rs1108683	5.3e-05	A	0.064	0.016	0.97%
2	*JHDM1D*	7q34	1.00e+06	107	0.00048	rs11972958	3.0e-05	G	0.065	0.016	0.99%
3	*TH*	11p15.5	1.00e+06	112	0.00091	rs7126629	0.00018	A	0.073	0.019	0.76%
4	*PC*	11q13.2	1.00e+06	50	0.00112	rs11227624	0.00108	C	0.051	0.016	0.61%
5	*TMEM151A*	11q13.2	1.00e+06	34	0.00115	rs479018	0.00210	G	0.050	0.016	0.53%
6	*HADH*	4q25	1.00e+06	143	0.00137	rs4956145	0.00113	C	-0.121	0.037	0.61%
7	*CYP2U1*	4q25	1.00e+06	94	0.00178	rs10025485	0.00170	C	-0.088	0.028	0.56%
8	*GRHPR*	9p13.2	1.00e+06	105	0.00209	rs765566	0.00149	A	0.049	0.015	0.55%
9	*NUDCD1*	8q23.1	1.00e+06	124	0.00230	rs2926220	0.00160	G	0.050	0.016	0.56%
10	*TIAL1*	10q26.11	1.00e+06	80	0.00268	rs10886513	0.00091	T	-0.052	0.016	0.58%
11	*KRTAP5-2*	11p15.5	1.00e+06	99	0.00270	rs7117114	0.00037	T	-0.059	0.016	0.70%
12	*LRFN4*	11q13.2	1.00e+06	24	0.00284	rs746018	0.00160	C	0.049	0.016	0.57%
13	*SLC25A40*	7q21.12	1.00e+06	51	0.00288	rs10263074	0.00340	C	-0.150	0.051	0.58%
14	*CNN2*	19p13.3	1.00e+06	72	0.00336	rs10408933	0.00064	G	0.091	0.027	0.71%
15	*DLX2*	2q31.1	1.00e+06	66	0.00684	rs4972632	0.00170	C	0.071	0.023	0.57%
16	*ACOT11*	1p32.3	1.00e+05	165	0.07374	rs12759156	0.00270	G	0.052	0.017	0.49%
17	*AKR7A2*	1p36.13	1.00e+05	88	0.02120	rs17477654	0.00390	A	-0.051	0.018	0.46%
18	*ALDH9A1*	1q24.1	1.00e+05	176	0.03136	rs9919190	0.00730	C	0.046	0.017	0.41%
19	*ALG6*	1p31.3	1.00e+05	100	0.03907	rs3938083	0.00190	C	0.071	0.023	0.53%
20	*ALPL*	1p36.12	1.00e+05	176	0.03578	rs2275363	0.00026	C	0.056	0.015	0.74%
21	*AMPD2*	1p13.3	1.00e+05	68	0.07654	rs12131828	0.00750	G	-0.146	0.054	0.46%
22	*AMY2A*	1p21.1	1.00e+05	9	0.07340	rs12136516	0.00450	T	0.060	0.021	0.49%
23	*ANGEL2*	1q32.3	1.00e+05	54	0.08161	rs11809007	0.00320	G	-0.155	0.053	0.53%
24	*ANGPTL1*	1q25.2	1.00e+05	52	0.09426	rs1413941	0.00970	G	0.065	0.025	0.35%
25	*APCS*	1q23.2	1.00e+05	118	0.04649	rs12029531	0.00820	C	-0.041	0.015	0.38%
26	*ATPAF1*	1p33	1.00e+05	94	0.06859	rs1150064	0.05900	T	0.033	0.017	0.20%
27	*B3GALNT2*	1q42.3	1.00e+05	123	0.05458	rs923942	0.00105	G	0.070	0.021	0.61%
28	*BARHL2*	1p22.2	1.00e+05	87	0.10807	rs6428601	0.00260	C	0.046	0.015	0.51%
29	*BPNT1*	1q41	1.00e+05	62	0.02687	rs2647458	0.00540	A	0.051	0.018	0.44%
30	*C1orf103*	1p13.3	1.00e+05	82	0.06300	rs10745314	0.00640	A	0.082	0.030	0.41%
31	*C1orf116*	1q32.2	1.00e+05	85	0.09035	rs760418	0.00190	C	0.047	0.015	0.51%
32	*C1orf157*	1q32.1	1.00e+05	91	0.09008	rs7525510	0.00480	G	-0.043	0.015	0.44%
33	*C1orf165*	1p33	1.00e+05	121	0.08205	rs7533353	0.01050	C	-0.044	0.017	0.38%
34	*C1orf201*	1p36.11	1.00e+05	128	0.01246	rs10903087	0.00053	G	-0.056	0.016	0.69%
35	*C1orf26*	1q25.3	1.00e+05	76	0.11315	rs12030554	0.03500	A	-0.034	0.016	0.25%
36	*CAMSAP1L1*	1q32.1	1.00e+05	161	0.07502	rs12042992	0.00980	C	-0.041	0.016	0.37%
37	*CAPZB*	1p36.13	1.00e+05	253	0.02883	rs17394154	0.00370	A	-0.050	0.017	0.45%
38	*CD2*	1p13.1	1.00e+05	78	0.07464	rs798048	0.02900	G	0.088	0.040	0.26%
39	*CD48*	1q23.3	1.00e+05	67	0.02616	rs11265461	0.00125	T	0.051	0.016	0.58%
40	*CD5L*	1q23.1	1.00e+05	109	0.09563	rs16839239	0.00360	C	-0.066	0.023	0.49%
41	*CDC42*	1p36.12	1.00e+05	106	0.09992	rs3820285	0.00073	G	-0.171	0.051	0.56%
42	*CDC7*	1p22.1	1.00e+05	140	0.08497	rs936340	0.00230	T	-0.048	0.016	0.54%
43	*CFHR3*	1q31.3	1.00e+05	43	0.04356	rs377298	0.01120	A	0.050	0.020	0.38%
44	*CGN*	1q21.3	1.00e+05	70	0.07205	rs12749	0.00430	G	-0.055	0.019	0.43%
45	*CHML*	1q43	1.00e+05	61	0.04338	rs3753216	0.00620	A	0.045	0.016	0.43%
46	*CLCA4*	1p22.3	1.00e+05	72	0.06977	rs1321681	0.01900	A	-0.049	0.021	0.31%
47	*CR1*	1q32.2	1.00e+05	169	0.06600	rs12022909	0.00580	T	0.043	0.016	0.43%
48	*CRP*	1q23.2	1.00e+05	97	0.00796	rs10437340	0.00042	G	0.058	0.016	0.72%
49	*DIRAS3*	1p31.3	1.00e+05	63	0.01148	rs269340	0.00059	C	-0.060	0.017	0.63%
50	*DMRTB1*	1p32.3	1.00e+05	74	0.04307	rs6588479	0.01020	G	0.040	0.015	0.36%

Shaded SNPs are also among the 50 best Krasanova[60] genes for iris nevi

Table S9. Human orthologs of the genes identified by Krasanova (2007)

	Gene Symbols		**Synonyms**	**Genes used in the polygenic score and candidate screens**
GeneBank#	**Mus musculus**	**Homo sapiens**		
BG087256	Abce1	*ABCE1*		*ABCE1*
BG084205	Acsl5	*ACSL5*		*ACSL5*
BG064835	Adamts10	*ADAMTS10*		*ADAMTS10*
BG063079	Adm	*ADM*		*ADM*
BQ551270	Aebp1	*AEBP1*		*AEBP1*
BC047281	Agpat4	*AGPAT4*		*AGPAT4*
BG077736	Aldh9a1	*ALDH9A1*		*ALDH9A1*
BG069982	Alox12	*ALOX12*		*ALOX12*
BQ551751	Ank3	*ANK3*		*ANK3*
BQ552281	Anxa6	*ANXA6*		*ANXA6*
BG078400	Ap1m2	*AP1M2*		*AP1M2*
BQ552159	Asb6	*ASB6*		*ASB6*
BG076762	No items found	No items found	ATPase, Na+/K+ transporting beta 1	*ATP1B1*
AU042390	Atp2a2	*ATP2A2*		*ATP2A2*
BG078689	Atp5a1	*ATP5A1*		*ATP5A1*
BG073437	Atp5b	*ATP5B*		*ATP5B*
AW537398	No items found	No items found	ATPase, H+ transporting, V1C1	*ATP6V1C1*
BG085675	Atp6v1g1	*ATP6V1G1*		*ATP6V1G1*
BQ552336	Atp7a	*ATP7A*		
BG075068	B4galt1	*B4GALT1*		*B4GALT1*
BG088471	Bcap29	*BCAP29*		*BCAP29*
BG063556	Bcor	*BCOR*		
BG071195	Best1	*BEST1*		*BEST1*
BQ551034	Bmp8b	*BMP8B*		*BMP8B*
BG077690	Bzw2	*BZW2*		*BZW2*
BG072229	Calb1	*CALB1*		*CALB1*
BC054805	Calm1	*CALM1*		*CALM1*
BQ550528	Ccne2	*CCNE2*		*CCNE2*
BG083664	Ccnl1	*CCNL1*		*CCNL1*
BG064770	Cct5	*CCT5*		*CCT5*
BQ550999	Cct6a	*CCT6A*		*CCT6A*
BG075757	Cd81	*CD81*		*CD81*
BG072071	Cdc42	*CDC42*		*CDC42*
BQ551224	Cdc7	*CDC7*		*CDC7*
BG064857	Cebpz	*CEBPZ*		*CEBPZ*
BG069861	Cfdp1	*CFDP1*		*CFDP1*
BG066368	Chek1	*CHEK1*		*CHEK1*
AB030378	Chst11	*CHST11*		*CHST11*
BG067882	Cog8	*COG8*		*COG8*
BQ552186	Col5a1	*COL5A1*		*COL5A1*
BG075779	Col6a1	*COL6A1*		*COL6A1*
AW554667	Cops5	*COPS5*		*COPS5*
BG085306	Cox6c	*COX6C*		*COX6C*
BG076988	Cox8a	*COX8A*		*COX8A*
BQ552082	No items found	*No items found*	Carboxypeptidase X 1	*CPXM1*
BG064928	Dab2	*DAB2*		*DAB2*
BG087134	Ddt	*DDT*		*DDT*
BG064782	Ddx5	*DDX5*		*DDX5*
BG085381	Degs1	*DEGS1*		*DEGS1*
BG088755	Dnaja1	*DNAJA1*		*DNAJA1*
BG065483	Dnajb1	*DNAJB1*		*DNAJB1*
BG077676	Dnajc7	*DNAJC7*		*DNAJC7*
BG086010	Dync1i2	*DYNC1I2*		*DYNC1I2*
AW538347	E2f4	*E2F4*		*E2F4*
BG064751	E4f1	*E4F1*		*E4F1*
BG084444	Eef1e1	*EEF1E1*		*EEF1E1*
BG077018	Eif1ax	*EIF1AX*		
BG077750	Eif3a	*EIF3A*		*EIF3A*
BG064923	Eif3i	*EIF3I*		*EIF3I*
BG078361	Eif4g2	*EIF4G2*		*EIF4G2*
BG082125	Elavl2	*ELAVL2*		*ELAVL2*
BG069686	Elovl6	*ELOVL6*		*ELOVL6*
BG076147	Emilin3	*EMILIN3*		*EMILIN3*
BG078410	Eno1	*ENO1*		*ENO1*
BG088166	Entpd4	*ENTPD4*		*ENTPD4*
BC034517	Ercc2	*ERCC2*		*ERCC2*
BG077757	Ero1l	*ERO1L*		*ERO1L*
BG087498	Etfa	*ETFA*		*ETFA*
BG078364	Ets1	*ETS1*		*ETS1*
BG064201	No items found	No items found	Ewing sarcoma homolog	*EWSR1*
BG069346	Eya2	*EYA2*		*EYA2*
C86521	Fbxo32	*FBXO32*		*FBXO32*
BG064795	No items found	No items found	Ferritin light chain 1	*FTL*
BG065384	Gabarapl2	*GABARAPL2*		*GABARAPL2*
BG079290	Gata1	*GATA1*		
BC064681	Gm5069	No items found	Glyceraldehyde-3-phosphate dehydrogenase	*GAPDH*
BQ51115	Ggps1	*GGPS1*		*GGPS1*
BG088919	Glg1	*GLG1*		*GLG1*

Table S9. Human orthologs of the genes identified by Krasanova (2007) [continued 2]

	Gene Symbols		**Synonyms**	**Genes used in the polygenic score and candidate screens**
GeneBank#	**Mus musculus**	**Homo sapiens**		
BG088057	Gmfb	*GMFB*		*GMFB*
AW556794	Gmpr2	*GMPR2*		*GMPR2*
BG078399	Gnb2l1	*GNB2L1*		*GNB2L1*
BG064771	Gnpda1	*GNPDA1*		*GNPDA1*
BG065213	No items found	No items found	Growth factor receptor bound protein 10	*GRB10*
BG086478	Gspt1	*GSPT1*		*GSPT1*
BC030444	Gstm4	*GSTM4*		*GSTM4*
AU015927	Gtf3c1	*GTF3C1*		*GTF3C1*
BG064826	Hax1	*HAX1*		*HAX1*
BG085818	Hba-a1	No items found	Hemoglobin alpha adult chain 1	*HBA1*
BG077101	Heyl	*HEYL*		*HEYL*
BG080836	Hipk3	*HIPK3*		*HIPK3*
AU020524	Hmgcs1	*HMGCS1*		*HMGCS1*
BG072533	Hnrnpa1	*HNRNPA1*		*HNRNPA1*
BG078653	Hnrnpd	*HNRNPD*		*HNRNPD*
BQ551016	Ddx49	*DDX49*	Homer, neuronal immediate early gene	*HOMER3*
BG074109	Hsp90aa1	*HSP90AA1*		*HSP90AA1*
BG087426	Hspa8	*HSPA8*		*HSPA8*
BG086000	Igf2bp1	*IGF2BP1*		*IGF2BP1*
BG063118	Incenp	*INCENP*		*INCENP*
BG066944	Itih1	*ITIH1*		*ITIH1*
BG071424	Itm2c	*ITM2C*		*ITM2C*
BG081243	Itpr1	*ITPR1*		*ITPR1*
BG080846	Jun	*JUN*		*JUN*
BG069106	Kcnh1	*KCNH1*		*KCNH1*
BG087137	Kit	*KIT*		*KIT*
BG063426	Krt7	*KRT7*		*KRT7*
BQ550072	Lcor	*LCOR*		*LCOR*
BG082595	Ldb1	*LDB1*		*LDB1*
BQ551553	Loxl3	*LOXL3*		*LOXL3*
BG064451	Lrp10	*LRP10*		*LRP10*
BG085146	ltf	*LTF*		*LTF*
BG071774	Lyar	*LYAR*		*LYAR*
AW558740	Man1a2	*MAN1A2*		*MAN1A2*
BG064865	Mcm5	*MCM5*		*MCM5*
BQ551346	Mcoln3	*MCOLN3*		*MCOLN3*
BG064680	Me1	*ME1*		*ME1*
BG080755	Mef2d	*MEF2D*		*MEF2D*
BG065235	Metap2	*METAP2*		*METAP2*
BG073306	Mrpl1	*MRPL1*		*MRPL1*
BQ552525	Mrpl45	*MRPL45*		*MRPL45*
BG084781	Mta3	*MTA3*		*MTA3*
BG077936	Mtf1	*MTF1*		*MTF1*
BG076333	Mthfd2	*MTHFD2*		*MTHFD2*
BG072457	Mtpn	*MTPN*		*MTPN*
BG072192	Naglu	*NAGLU*		*NAGLU*
BG075327	Ncor1	*NCOR1*		*NCOR1*
BG084093	Ndrg2	*NDRG2*		*NDRG2*
BQ550897	Ndufa10	*NDUFA10*		*NDUFA10*
BC024673	Ndufa8	*NDUFA8*		*NDUFA8*
BG063606	Ndufaf1	*NDUFAF1*		*NDUFAF1*
BQ550532	Nfkb2	*NFKB2*		*NFKB2*
BG088461	Ngfrap1	*NGFRAP1*		
BG073805	Nisch	*NISCH*		*NISCH*
BG063736	Nlk	*NLK*		*NLK*
BG087125	Nr0b2	*NR0B2*		*NR0B2*
BG063771	Pabpc1	*PABPC1*		*PABPC1*
BQ550908	Papola	*PAPOLA*		*PAPOLA*
BQ551164	Pask	*PASK*		*PASK*
BG085359	Pex19	*PEX19*		*PEX19*
BG077408	Pmm1	*PMM1*		*PMM1*
BG071480	Pole2	*POLE2*		*POLE2*
BG074714	Pon3	*PON3*		*PON3*
BG080374	Ppap2b	*PPAP2B*		*PPAP2B*
BG071790	Ppp1cc	*PPP1CC*		*PPP1CC*
C75970 Pp	Ppp2cb	*PPP2CB*		*PPP2CB*
BQ551079	Ppp2r2a	*PPP2R2A*		*PPP2R2A*
BG064735	Prdx6	*PRDX6*		*PRDX6*
BG069165	Preb	*PREB*		*PREB*
BG084015	Psat1	*PSAT1*		*PSAT1*
BC001982	Psma4	*PSMA4*		*PSMA4*
AU043387	Psmd12	*PSMD12*		*PSMD12*
BG077865	Psmg2	*PSMG2*		*PSMG2*
BG068173	Ptp4a1	*PTP4A1*		*PTP4A1*
BG071651	Rab11a	*RAB11A*		*RAB11A*
BG080166	Rabl3	*RABL3*		*RABL3*
BG071892	Rad21	*RAD21*		*RAD21*
BQ552743	Rbpms	*RBPMS*		*RBPMS*

Table S9. Human orthologs of the genes identified by Krasanova (2007) [continued 3]

	Gene Symbols		**Synonyms**	**Genes used in the polygenic score and candidate screens**
GeneBank#	**Mus musculus**	**Homo sapiens**		
BG064938	Rdbp	*RDBP*		*RDBP*
BQ552179	Rev3l	*REV3L*		*REV3L*
AU041357	Rhod	*RHOD*		*RHOD*
BG086349	Rnf11	*RNF11*		*RNF11*
BG074107	Rpl3	*RPL3*		*RPL3*
BC054388	Rpl37	*RPL37*	Ribosomal protein L37a	*RPL37A*
BG084847	Rpl39	*RPL39*		
BG065196	Rpl5	*RPL5*		*RPL5*
BG063883	Rpl7	*RPL7*		*RPL7*
BG077480	No items found	No items found	Ribosomal protein L8	*RPL8*
BG072802	Rps16	*RPS16*		*RPS16*
AW556256	Rps6	*RPS6*		*RPS6*
BG063083	Rps6ka2	*RPS6KA2*		*RPS6KA2*
BG080845	Rras2	*RRAS2*		*RRAS2*
BG085334	Sar1a	*SAR1A*		*SAR1A*
BG077146	Sars	*SARS*		*SARS*
BQ551360	Sema3a	*SEMA3A*		*SEMA3A*
BG070610	Senp3	*SENP3*		*SENP3*
BG077813	Senp6	*SENP6*		*SENP6*
BG077478	Serpinb9	*SERPINB9*		*SERPINB9*
BG088179	No items found	*SETDB1*	SET domain, bifurcated 1	*SETDB1*
BG083967	Sfxn1	*SFXN1*		*SFXN1*
BG087217	Sfxn3	*SFXN3*		*SFXN3*
BQ551075	Sh3bp1	*SH3BP1*		*SH3BP1*
BG064853	Slc16a3	*SLC16A3*		*SLC16A3*
BG085151	Slc27a4	*SLC27A4*		*SLC27A4*
BG075243	Slc30a3	*SLC30A3*		*SLC30A3*
BG064714	Slc3a2	*SLC3A2*		*SLC3A2*
BQ552559	Slc6a6	*SLC6A6*		*SLC6A6*
BG072114	Slco2a1	*SLCO2A1*		*SLCO2A1*
BG076183	Smc1b	*SMC1B*		*SMC1B*
BG085432	Smc3	*SMC3*		*SMC3*
BG077269	Smc4	*SMC4*		*SMC4*
BG070360	Socs4	*SOCS4*		*SOCS4*
BG084323	No items found	No items found	Solute carrier family 34	*SLC34A2*
BG079034	Sphk2	*SPHK2*		*SPHK2*
BQ550910	Srsf10	*SRSF10*	Neural-salient serine/arginine-rich	
BQ551716	St6galnac2	*ST6GALNAC2*	Sialyltransferase 7B	*ST6GALNAC2*
BG077776	Stk10	*STK10*		*STK10*
BG088027	Strn3	*STRN3*		*STRN3*
BQ551727	Unc84a / Sun1	*UNC84A / SUN1*		*UNC84A*
BG076926	Sypl1	*SYPL1*		*SYPL1*
BG065267	T	*T*		*T*
BG075593	Taf12	*TAF12*		*TAF12*
BG085163	Taf1a	*TAF1A*		*TAF1A*
BG084568	Tbca	*TBCA*		*TBCA*
BG088173	Tdrd1	*TDRD1*		*TDRD1*
BG075056	Tep1	*TEP1*		*TEP1*
BG072137	Tfb2m	*TFB2M*		*TFB2M*
AU040433	Tgfa	*TGFA*		*TGFA*
BG086002	Tia1	*TIA1*		*TIA1*
BG069221	Timd2	No items found	T-cell immunoglobulin and mucin domain 2	
BG075312	Tkt	*TKT*		*TKT*
BG075073	Tmsb4x	*TMSB4X*		
BG077775	Tnfrsf23	*TNFRSF23*	TNF receptor superfamily, member 23	
BG088746	Trim27	*TRIM27*		*TRIM27*
BG070343	Trim6	*TRIM6*		*TRIM6*
BG080898	Trpm7	*TRPM7*		*TRPM7*
BG076653	Tspo / BZRAP1	*TSPO / BZRAP1*	Benzodiazepine receptor, peripheral	*BZRAP1*
BG087420	Tubb4	*TUBB4*		*TUBB4*
BQ551468	Tubd1	*TUBD1*		*TUBD1*
BG077718	Twf1	*TWF1*		*TWF1*
BG065003	Usp21	*USP21*		*USP21*
BG069818	Usp3	*USP3*		*USP3*
BG076775	Usp39	*USP39*		*USP39*
BQ550549	Vasp	*VASP*		*VASP*
BG063694	Vcl	*VCL*		*VCL*
BC052677	Wisp1	*WISP1*		*WISP1*
L33417	Vldlr	*VLDLR*		*VLDLR*
BQ552129	Wnt5b	*WNT5B*		*WNT5B*
BG088199	Xrn2	*XRN2*		*XRN2*
AW539280	Yif1a	*YIF1A*		*YIF1A*
BQ551106	Ywhab	*YWHAB*		*YWHAB*

For each of the 229 GeneBank transcripts reported by Krasanova et al[55] the gene symbols are reported for *Mus musculus* and *Homo sapiens* according to NCIB Unigene and Entrez Gene data bases. For transcripts without records in these databases the gene name, as given by Krasanova was matched to a human gene with the same name using the database GeneCards. Ten genes identified by Krasanova et al[55] were not included in the analyses. Seven of these are situated on the X chromosome (Atp7a, Bcor, Eif1a, Gata1, Ngfrap1, Rpl39, Tmsb4x). Three others (BQ550910/Srsf13a, BG069221/Timd2, BG077775/Tnfrsf23) are currently regarded as being unique to mice.

Table S10. The 50 genes in the Krasanova gene set most strongly associated with crypts: VEGAS results for the discovery sample GWAS, N=2121

	Test statistics for the genes most strongly associated with Crypts					**Test Statistics for the SNP most strongly associated within each gene**					
#	**Gene**	**Position**	**#Simulations**	**#SNPs**	**P-value**	**SNP**	**P-value**	**Allele**	**beta**	**SE**	**r^2**
1	*SEMA3A*	7q21.11	1.00e+06	379	3.00e-06	rs10235789	2.70e-06	C	0.098	0.021	1.27%
2	*HSPA8*	11q24.1	1.00e+06	90	0.00046	rs10892958	0.00044	C	0.091	0.026	0.67%
3	*CCNL1*	3q25.31	1.00e+06	69	0.00050	rs2133952	0.00096	C	0.069	0.021	0.58%
4	*USP39*	2p11.2	1.00E+05	104	0.00700	rs1254901	0.00036	G	0.082	0.023	0.77%
5	*SMC1B*	22q13.31	1.00E+05	160	0.01113	rs6007032	0.00250	C	0.074	0.024	0.53%
6	*MRPL45*	17q12	1.00e+05	20	0.01192	rs8066255	0.00230	A	-0.092	0.030	0.55%
7	*MCOLN3*	1p22.3	1.00e+05	127	0.01316	rs1975365	0.00500	G	-0.061	0.022	0.45%
8	*TDRD1*	10q25.3	1.00e+05	119	0.01483	rs12262953	0.00146	G	0.121	0.038	0.58%
9	*YIF1A*	11q13.2	1.00E+05	33	0.01602	rs479018	0.00230	G	-0.066	0.022	0.53%
10	*SPHK2*	19q13.33	1.00e+05	70	0.01743	rs433852	0.00170	C	-0.081	0.026	0.54%
11	*EIF3A*	10q26.11	1.00E+05	76	0.03007	rs10787899	0.00650	A	0.057	0.021	0.43%
12	*PASK*	2q37.3	1.00e+05	134	0.03155	rs12987697	0.00037	A	0.076	0.021	0.72%
13	*WISP1*	8q24.22	1.00E+05	206	0.03178	rs753722	0.00190	G	-0.070	0.023	0.54%
14	*VLDLR*	9p24.2	1.00e+05	217	0.04526	rs1567712	0.00112	A	0.081	0.025	0.62%
15	*HNRNPA1*	12q13.13	1.00E+05	24	0.04917	rs10506328	0.00230	C	-0.067	0.022	0.54%
16	*GRB10*	7p12.1	1.00e+05	406	0.05621	rs2715119	0.00480	T	0.076	0.027	0.44%
17	*COL6A1*	21q22.3	1.00e+05	94	0.05761	rs13052792	0.00094	G	-0.131	0.040	0.64%
18	*METAP2*	12q22	1.00e+05	118	0.05769	rs10431429	0.00600	G	-0.059	0.021	0.45%
19	*COX6C*	8q22.2	1.00e+05	46	0.05990	rs12544943	0.02600	A	-0.071	0.032	0.30%
20	*MEF2D*	1q22	1.00e+05	83	0.06942	rs7548323	0.00200	G	-0.088	0.028	0.55%
21	*RBPMS*	8p12	1.00e+05	193	0.07294	rs6468319	0.01500	C	0.058	0.024	0.34%
22	*EYA2*	20q13.12	1.00E+05	475	0.07382	rs2868860	0.00059	T	0.107	0.031	0.74%
23	*CPXM1*	20p13	1.00E+05	72	0.08154	rs6037407	0.00880	A	-0.133	0.051	0.41%
24	*MTPN*	7q33	1.00E+05	122	0.09171	rs894793	0.00350	T	0.060	0.021	0.47%
25	*SERPINB9*	6p25.2	1.00E+05	118	0.09251	rs380779	0.00760	T	-0.060	0.022	0.40%
26	*DNAJB1*	19p13.12	1.00E+05	49	0.10006	rs10409467	0.00900	C	0.295	0.113	0.43%
27	*RPS6*	9p22.1	1000	75	0.10300	rs2277151	0.01300	A	0.072	0.029	0.36%
28	*TRIM27*	6p22.1	1000	112	0.11400	rs2038887	0.01110	T	-0.117	0.046	0.37%
29	*ANK3*	10q21.2	1000	547	0.11800	rs10821762	0.00135	G	-0.067	0.021	0.57%
30	*SENP3*	17p13.1	1000	47	0.12200	rs11078697	0.00810	C	0.273	0.103	0.46%
31	*GAPDH*	12p13.31	1000	74	0.12500	rs4764602	0.01900	A	-0.049	0.021	0.32%
32	*LCOR*	10q24.1	1000	80	0.12700	rs10399993	0.02600	T	-0.058	0.026	0.29%
33	*RPS6KA2*	6q27	1000	761	0.13300	rs10946173	0.00280	C	0.064	0.021	0.51%
34	*EMILIN3*	20q12	1000	75	0.14400	rs7275102	0.00880	A	0.060	0.023	0.38%
35	*GMPR2*	14q12	1000	53	0.14500	rs2256989	0.02200	T	-0.051	0.022	0.31%
36	*ST6GALNAC2*	17q25.1	1000	74	0.14700	rs895157	0.00850	A	0.068	0.026	0.40%
37	*GNPDA1*	5q31.3	1000	87	0.15200	rs164502	0.01700	G	0.052	0.022	0.34%
38	*UNC84A*	7p22.3	1000	76	0.15300	rs4275123	0.00460	A	-0.072	0.025	0.47%
39	*MTF1*	1p34.3	1000	28	0.15700	rs473279	0.03200	C	-0.047	0.022	0.25%
40	*GGPS1*	1q42.3	1000	64	0.15900	rs6666434	0.01800	C	0.068	0.029	0.32%
41	*TIA1*	2p13.3	1000	62	0.15900	rs7575953	0.04200	C	0.042	0.021	0.23%
42	*SLC6A6*	3p25.1	1000	214	0.17100	rs12632161	0.01700	C	0.119	0.050	0.33%
43	*GABARAPL2*	16q23.1	1000	74	0.17300	rs16940467	0.02200	C	-0.096	0.042	0.29%
44	*SFXN1*	5q35.2	1000	191	0.17600	rs10037961	0.00880	A	0.073	0.028	0.39%
45	*MTA3*	2p21	1000	173	0.17900	rs13424316	0.01800	C	0.050	0.021	0.33%
46	*CCT5*	5p15.2	1000	184	0.18000	rs1045369	0.00250	G	0.086	0.028	0.51%
47	*T*	6q27	1000	194	0.18000	rs9295321	0.00840	A	-0.059	0.022	0.40%
48	*NFKB2*	10q24.32	1000	28	0.18300	rs7086205	0.02000	T	0.066	0.028	0.32%
49	*STRN3*	14q12	1000	164	0.18400	rs2073204	0.00590	T	0.089	0.032	0.42%
50	*HMGCS1*	5p12	1000	84	0.18600	rs17244293	0.04600	G	-0.105	0.053	0.23%

Shaded SNPs are also among the 50 best genes for crypts, as computed by VEGAS

Table S11. The 50 genes in the Krasanova gene set most strongly associated with furrows: VEGAS results for the discovery sample GWAS, N=2121

Test statistics for the genes most strongly associated with Furrows						Test Statistics for the SNP most strongly associated within each gene					
#	**Gene**	**Position**	**#Simulations**	**#SNPs**	**P-value**	**SNP**	**P-value**	**Allele**	**beta**	**SE**	**r^2**
1	*COL6A1*	21q22.3	1.00e+06	94	0.00212	rs13047753	0.00001	G	0.164	0.036	1.27%
2	*MTHFD2*	2p13.	1.00e+06	65	0.00284	rs1667601	0.00023	G	0.090	0.025	0.79%
3	*SEMA3A*	7q21.11	1.00e+06	379	0.00465	rs10235789	0.00009	C	-0.095	0.024	0.86%
4	*SENP6*	6q14.1	1.00e+06	155	0.00818	rs17414086	0.00750	C	-0.067	0.025	0.42%
5	*DNAJC7*	17q21.2	1.00e+06	40	0.00845	rs11652600	0.00440	T	0.096	0.034	0.49%
6	*ERO1L*	14q22.1	1.00e+06	85	0.01736	rs17681013	0.00270	A	-0.072	0.024	0.51%
7	*PPP2CB*	8p12	1,00E+05	74	0.01786	rs1116003	0.00760	G	-0.091	0.034	0.39%
8	*MTF1*	1p34.3	1.00e+06	28	0.02026	rs3795494	0.00250	A	0.072	0.024	0.51%
9	*TRIM27*	6p22.1	1.00e+06	112	0.02688	rs16894407	0.00600	G	-0.184	0.067	0.43%
10	*GRB10*	7p12.1	1.00e+06	406	0.03018	rs2237481	0.00420	C	-0.100	0.035	0.46%
11	*CDC42*	1p36.12	1.00e+06	106	0.03652	rs760922	0.00250	C	0.140	0.046	0.49%
12	*RPL8*	8q24.3	1.00e+06	68	0.03848	rs1618523	0.01500	C	0.062	0.026	0.35%
13	*TDRD1*	10q25.3	1.00e+06	119	0.04856	rs682836	0.00019	A	-0.093	0.025	0.85%
14	*SAR1A*	10q22.1	1,00E+05	117	0.05357	rs10823490	0.00160	T	-0.078	0.025	0.57%
15	*BZW2*	7p21.1	1.00e+06	199	0.05572	rs1608698	0.00490	C	0.069	0.025	0.46%
16	*ITPR1*	3p26.1	1.00e+06	611	0.05709	rs6766684	0.00003	G	0.104	0.025	1.00%
17	*HEYL*	1p34.3	1.00e+06	80	0.05934	rs12021965	0.00240	C	-0.234	0.077	0.64%
18	*CALM1*	14q32.11	1.00e+06	86	0.06171	rs12147703	0.00860	A	-0.103	0.039	0.41%
19	*MTPN*	7q33	1.00e+06	122	0.06674	rs10244931	0.01180	A	0.063	0.025	0.37%
20	*LRP10*	14q11.2	1.00e+06	68	0.06964	rs4982695	0.00063	T	0.089	0.026	0.68%
21	*SLC27A4*	9q34.11	1.00e+06	33	0.07153	rs6478827	0.02400	C	0.134	0.059	0.31%
22	*ATP6V1G1*	9q32	1,00E+05	118	0.07939	rs2418315	0.00490	A	0.097	0.035	0.49%
23	*PPP2R2A*	8p21.2	1.00e+06	130	0.07959	rs3808582	0.00036	C	-0.227	0.064	0.77%
24	*T*	6q27	1.00e+06	194	0.08337	rs9457140	0.00190	T	0.093	0.030	0.58%
25	*TAF12*	1p35.3	1.00e+06	30	0.10044	rs12139243	0.04300	T	0.053	0.026	0.23%
26	*DNAJB1*	19p13.12	1.00e+06	49	0.10164	rs3815715	0.00160	C	-0.091	0.029	0.62%
27	*PPAP2B*	1p32.2	1.00e+06	202	0.10199	rs6691109	0.00740	C	-0.107	0.040	0.41%
28	*METAP2*	12q22	1000	118	0.10400	rs12823525	0.01180	G	0.063	0.025	0.39%
29	*PTP4A1*	6q12	1000	41	0.10800	rs1197892	0.04100	A	0.093	0.046	0.23%
30	*LYAR*	4p16.3	1000	112	0.10900	rs10033534	0.01800	A	0.064	0.027	0.32%
31	*SMC3*	10q25.2	1000	122	0.10900	rs11815960	0.00062	A	-0.201	0.059	0.65%
32	*ITM2C*	2q37.1	1000	74	0.11400	rs6759606	0.00370	A	0.086	0.030	0.49%
33	*RNF11*	1p32.3	1000	34	0.11600	rs11205826	0.03200	G	-0.111	0.052	0.27%
34	*BZRAP1*	17q22	1000	95	0.11700	rs2680688	0.00500	C	-0.069	0.024	0.45%
35	*TKT*	3p21.1	1000	109	0.12300	rs12635973	0.00250	C	-0.147	0.048	0.51%
36	*NDRG2*	14q11.2	1000	114	0.12600	rs10162349	0.00350	C	-0.086	0.030	0.50%
37	*TRPM7*	15q21.2	1000	164	0.12700	rs3098198	0.01700	A	-0.058	0.024	0.33%
38	*PSMG2*	18p11.21	1000	77	0.13800	rs2155734	0.01300	G	-0.083	0.033	0.33%
39	*INCENP*	11q12.3	1000	126	0.14000	rs1792949	0.01900	G	0.062	0.026	0.33%
40	*TFB2M*	1q44	1000	87	0.14500	rs7517207	0.00038	A	-0.117	0.033	0.76%
41	*YIF1A*	11q13.2	1000	33	0.14600	rs1151540	0.04500	A	-0.049	0.024	0.23%
42	*ME1*	6q14.2	1000	234	0.14700	rs7757427	0.04200	T	0.053	0.026	0.24%
43	*ADAMTS10*	19p13.2	1000	32	0.17600	rs3111576	0.01800	C	0.089	0.038	0.32%
44	*GNPDA1*	5q31.3	1000	87	0.18300	rs2232196	0.00670	G	-0.087	0.032	0.44%
45	*AGPAT4*	6q26	1000	266	0.18600	rs940520	0.00440	T	-0.137	0.048	0.49%
46	*AP1M2*	19p13.2	1000	48	0.18900	rs1465701	0.03700	C	0.059	0.029	0.25%
47	*RPL37A*	2q35	1000	97	0.19000	rs10932662	0.00470	T	-0.070	0.025	0.46%
48	*GSTM4*	1p13.3	1000	60	0.19500	rs1799875	0.01240	C	0.061	0.025	0.37%
49	*VCL*	10q22.2	1000	122	0.20000	rs11813221	0.03399	G	0.180	0.085	0.26%
50	*TUBD1*	17q23.1	1000	43	0.20700	rs8071475	0.04000	T	-0.058	0.028	0.25%

Shaded SNPs are also among the 50 best genes for furrow, as computed by VEGAS

Table S12. The 50 genes in the Krasanova gene set most strongly associated with pigmented ring: VEGAS results for the discovery sample GWAS, N=2121

	Test statistics for the genes most strongly associated with Pigmented RIng					Test Statistics for the SNP most strongly associated within each gene					
#	**Gene**	**Position**	**#Simulations**	**#SNPs**	**P-value**	**SNP**	**P-value**	**Allele**	**beta**	**SE**	**r^2**
1	*COL6A1*	21q22.3	1.00e+06	94	0.001214	rs13046217	0.00009	T	0.088	0.023	0.89%
2	*COX6C*	8q22.2	1.00e+05	46	0.00593	rs4434590	0.00101	C	-0.098	0.030	0.65%
3	*E2F4*	16q22.1	1.00e+05	31	0.01243	rs12051249	0.00480	A	-0.126	0.045	0.46%
4	*NCOR1*	17p11.2	1.00e+05	111	0.01653	rs178830	0.00300	T	-0.067	0.023	0.53%
5	*AP1M2*	19p13.2	1.00e+05	48	0.01789	rs17677316	0.00800	G	-0.069	0.026	0.42%
6	*TRIM6*	11p15.4	1.00e+05	282	0.02169	rs11038295	0.00380	G	0.110	0.038	0.51%
7	*TUBD1*	17q23.1	1.00e+05	43	0.02456	rs1295927	0.00640	A	-0.061	0.023	0.44%
8	*GMFB*	14q22.2	1.00e+05	44	0.03030	rs4901518	0.00111	T	0.106	0.033	0.60%
9	*CD81*	11p15.5	1.00e+05	54	0.03496	rs800345	0.01600	C	0.127	0.052	0.37%
10	*CDC42*	1p36.12	1.00e+05	106	0.03669	rs7526484	0.00061	C	-0.091	0.026	0.70%
11	*PON3*	7q21.3	1.00e+05	118	0.04334	rs10487133	0.00003	T	-0.141	0.034	1.02%
12	*RPL8*	8q24.3	1.00e+05	68	0.04568	rs2955195	0.02600	T	0.051	0.023	0.30%
13	*ADAMTS10*	19p13.2	1.00e+05	32	0.05013	rs2967715	0.00380	T	0.151	0.052	0.52%
14	*TFB2M*	1q44	1.00e+05	87	0.05115	rs3120693	0.01270	A	0.170	0.068	0.38%
15	*RABL3*	3q13.33	1.00e+05	52	0.05238	rs2292719	0.01800	G	-0.059	0.025	0.32%
16	*ALOX12*	17p13.1	1.00e+05	60	0.05416	rs188774	0.01700	G	-0.068	0.029	0.33%
17	*BCAP29*	7q22.3	1.00e+05	74	0.06144	rs11761775	0.00500	A	-0.063	0.022	0.45%
18	*PREB*	2p23.3	1.00e+05	43	0.06777	rs4665955	0.00080	C	-0.076	0.023	0.65%
19	*TIA1*	2p13.3	1,00E+05	62	0.08327	rs12713689	0.02700	A	0.050	0.022	0.28%
20	*KCNH1*	1q32.2	1.00e+05	516	0.08465	rs6540636	0.00090	G	0.116	0.035	0.63%
21	*SARS*	1p13.3	1,00E+05	118	0.08926	rs12239854	0.00350	A	0.090	0.031	0.51%
22	*CCNL1*	3q25.31	1000	69	0.10500	rs6441111	0.01090	C	0.055	0.022	0.36%
23	*SAR1A*	10q22.1	1000	117	0.10700	rs12262702	0.00620	C	-0.121	0.044	0.48%
24	*SLC3A2*	11q12.3	1000	68	0.10900	rs474209	0.03600	C	-0.049	0.024	0.26%
25	*SETDB1*	1q21.3	1000	56	0.11300	rs267734	0.01600	T	-0.065	0.027	0.34%
26	*ATP6V1G1*	9q32	1000	118	0.11500	rs10114562	0.00950	C	0.058	0.023	0.40%
27	*MCM5*	22q12.3	1000	91	0.11600	rs133420	0.00360	C	0.117	0.040	0.48%
28	*STRN3*	14q12	1000	164	0.11800	rs179658	0.00117	T	0.079	0.024	0.63%
29	*PPP1CC*	12q24.11	1000	50	0.11900	rs16940992	0.02800	A	-0.071	0.032	0.28%
30	*PSMG2*	18p11.21	1000	77	0.12400	rs888270	0.00380	G	0.087	0.030	0.51%
31	*COL5A1*	9q34.3	1000	276	0.12700	rs13301426	0.00095	C	0.134	0.041	0.62%
32	*PPP2CB*	8p12	1000	74	0.15600	rs12543324	0.04300	C	0.069	0.034	0.25%
33	*GNPDA1*	5q31.3	1000	87	0.16000	rs163918	0.06400	C	-0.072	0.039	0.20%
34	*ERCC2*	19q13.32	1000	65	0.17000	rs1799787	0.02200	G	0.055	0.024	0.31%
35	*FBXO32*	8q24.13	1000	133	0.17700	rs16898505	0.00054	A	-0.165	0.048	0.65%
36	*GGPS1*	1q42.3	1000	64	0.18700	rs4659821	0.00064	T	0.178	0.052	0.69%
37	*EMILIN3*	20q12	1000	75	0.19600	rs6102363	0.03000	A	0.138	0.064	0.29%
38	*DDX5*	17q23.3	1000	6	0.20100	rs7212377	0.07400	T	0.089	0.050	0.20%
39	*ELAVL2*	9p21.3	1000	295	0.20300	rs10966006	0.00170	G	0.134	0.043	0.57%
40	*COX8A*	11q13.1	1000	48	0.20800	rs578001	0.03000	G	0.048	0.022	0.27%
41	*CHST11*	12q23.3	1000	513	0.20800	rs3924373	0.00004	G	0.145	0.035	1.02%
42	*POLE2*	14q21.3	1000	64	0.21000	rs12890086	0.06500	G	-0.045	0.024	0.20%
43	*PPAP2B*	1p32.2	1000	202	0.21300	rs11206816	0.01120	A	0.064	0.025	0.37%
44	*RRAS2*	11p15.2	1000	98	0.21400	rs10766178	0.01500	A	0.063	0.026	0.33%
45	*ATP2A2*	12q24.11	1000	29	0.21800	rs11065630	0.03700	A	0.164	0.079	0.22%
46	*PABPC1*	8q22.3	1000	109	0.21900	rs2935548	0.00320	A	-0.104	0.035	0.48%
47	*LOXL3*	2p13.1	1000	32	0.22000	rs10454145	0.08500	C	-0.061	0.035	0.17%
48	*HOMER3*	19p13.11	1000	37	0.22400	rs4808167	0.03200	A	-0.052	0.024	0.30%
49	*SLC30A3*	2p23.3	1000	62	0.23000	rs13404327	0.11000	G	-0.041	0.026	0.15%
50	*SENP6*	6q14.1	1000	155	0.23100	rs13205073	0.01290	T	-0.076	0.031	0.34%

Shaded SNPs are also among the 50 best genes for the pigmented ring, as computed by VEGAS

Table S13. The 50 genes in the Krasanova gene set most strongly associated with iris nevi: VEGAS results for the discovery sample GWAS, N=2121

	Test statistics for the genes most strongly associated with Nevi					Test Statistics for the SNP most strongly associated within each gene					
#	**Gene**	**Position**	**#Simulations**	**#SNPs**	**P-value**	**SNP**	**P-value**	**Allele**	**beta**	**SE**	**h^2**
1	*YIF1A*	11q13.2	1,00E+05	33	0.00209	rs479018	0.00210	G	0.050	0.016	0.53%
2	*METAP2*	12q22	1.00e+05	118	0.00409	rs2197973	0.00053	T	-0.054	0.016	0.69%
3	*COX8A*	11q13.1	1.00e+05	48	0.00544	rs4980499	0.00270	G	0.047	0.016	0.50%
4	*RPS6*	9p22.1	1.00e+05	75	0.00578	rs10757054	0.00078	G	0.074	0.022	0.60%
5	*CCT6A*	7p11.2	1.00e+05	40	0.008	rs10043	0.00240	C	0.058	0.019	0.53%
6	*ELOVL6*	4q25	1.00e+05	219	0.01364	rs6533491	0.00044	C	-0.054	0.015	0.68%
7	*PON3*	7q21.3	1.00e+05	118	0.0142	rs13223537	0.00180	T	0.056	0.018	0.57%
8	*TRIM6*	11p15.4	1.00e+05	282	0.02632	rs7120668	0.00250	G	-0.049	0.016	0.53%
9	*RPL37A*	2q35	1.00e+05	97	0.03118	rs922987	0.00024	T	0.057	0.015	0.76%
10	*ALDH9A1*	1q24.1	1.00e+05	176	0.03136	rs9919190	0.00730	C	0.046	0.017	0.41%
11	*LRP10*	14q11.2	1.00e+05	68	0.03346	rs8006914	0.01500	C	0.041	0.017	0.35%
12	*RHOD*	11q13.2	1.00e+05	29	0.03808	rs7112925	0.02000	C	0.038	0.016	0.32%
13	*HMGCS1*	5p12	1,00E+05	84	0.04219	rs7380703	0.01800	T	-0.043	0.018	0.32%
14	*COPS5*	8q13.1	1.00e+05	3	0.04629	rs11785782	0.04500	C	-0.032	0.016	0.22%
15	*GMFB*	14q22.2	1.00e+05	44	0.05875	rs10162293	0.01500	A	-0.048	0.020	0.33%
16	*GSTM4*	1p13.3	1.00e+05	60	0.05978	rs12131828	0.00750	G	-0.146	0.054	0.46%
17	*AGPAT4*	6q26	1.00e+05	266	0.06367	rs1111040	0.00450	T	0.046	0.016	0.44%
18	*DNAJC7*	17q21.2	1.00e+05	40	0.06839	rs8079958	0.02400	G	-0.060	0.026	0.30%
19	*SENP6*	6q14.1	1.00e+05	155	0.07744	rs9447522	0.01260	A	0.039	0.016	0.34%
20	*PSAT1*	9q21.2	1.00e+05	166	0.07765	rs11137607	0.01600	A	0.038	0.016	0.34%
21	*DDT*	22q11.23	1.00e+05	31	0.07902	rs7291786	0.00460	C	0.064	0.023	0.47%
22	*CDC7*	1p22.1	1,00E+05	140	0.08497	rs936340	0.00230	T	-0.048	0.016	0.54%
23	*YWHAB*	20q13.12	1.00e+05	92	0.0863	rs1079900	0.01900	A	0.038	0.016	0.32%
24	*TRPM7*	15q21.2	1.00e+05	164	0.0889	rs8042172	0.00840	T	-0.050	0.019	0.39%
25	*AP1M2*	19p13.2	1.00e+05	48	0.09038	rs3745249	0.01700	C	-0.038	0.016	0.33%
26	*ITM2C*	2q37.1	1.00e+05	74	0.09093	rs6724157	0.00170	C	0.060	0.019	0.54%
27	*NDUFA8*	9q33.2	1.00e+05	125	0.09886	rs7030059	0.00770	T	0.058	0.022	0.41%
28	*CDC42*	1p36.12	1.00e+05	106	0.09992	rs3820285	0.00073	G	-0.171	0.051	0.56%
29	*ATP5B*	12q13.3	1000	36	0.105	rs17444799	0.04000	C	0.063	0.031	0.22%
30	*CFDP1*	16q23.1	1,00E+05	152	0.10618	rs4887828	0.00910	A	0.049	0.019	0.36%
31	*RBPMS*	8p12	1000	193	0.107	rs6985051	0.00260	G	0.055	0.018	0.51%
32	*PABPC1*	8q22.3	1.00e+05	109	0.10934	rs1039153	0.02400	T	-0.037	0.016	0.28%
33	*EMILIN3*	20q12	1000	75	0.114	rs6029636	0.01240	C	0.044	0.017	0.36%
34	*MCM5*	22q12.3	1000	91	0.115	rs2071737	0.01120	G	0.040	0.016	0.36%
35	*GGPS1*	1q42.3	1000	64	0.118	rs6666434	0.01800	C	0.050	0.021	0.32%
36	*CD81*	11p15.5	1000	54	0.118	rs808964	0.03500	A	0.038	0.018	0.25%
37	*REV3L*	6q21	1000	211	0.12	rs3851228	0.00067	A	0.110	0.032	0.65%
38	*PMM1*	22q13.2	1000	39	0.121	rs5996039	0.07500	A	0.036	0.020	0.19%
39	*ATP6V1C1*	8q22.3	1000	207	0.122	rs2515173	0.00057	T	0.053	0.015	0.65%
40	*ALOX12*	17p13.1	1000	60	0.123	rs11078663	0.00160	T	-0.052	0.016	0.59%
41	*CEBPZ*	2p22.2	1000	108	0.128	rs11124570	0.00590	A	0.081	0.029	0.42%
42	*LCOR*	10q24.1	1000	80	0.132	rs11188929	0.06200	G	0.050	0.027	0.21%
43	*SMC3*	10q25.2	1000	122	0.135	rs11591748	0.01050	G	0.068	0.026	0.39%
44	*WISP1*	8q24.22	1000	206	0.136	rs2929984	0.01290	G	0.049	0.020	0.37%
45	*ANK3*	10q21.2	1000	547	0.136	rs10761482	0.00690	C	0.051	0.019	0.42%
46	*USP3*	15q22.31	1000	127	0.136	rs11630290	0.00001	C	0.094	0.021	1.13%
47	*SLC16A3*	17q25.3	1000	44	0.137	rs4789670	0.06300	T	-0.029	0.016	0.19%
48	*TUBB4*	19p13.3	1000	32	0.137	rs3760749	0.00530	A	-0.071	0.025	0.44%
49	*TRIM27*	6p22.1	1000	112	0.151	rs9257319	0.01020	T	0.054	0.021	0.35%
50	*ADM*	11p15.4	1000	93	0.152	rs1442739	0.01900	C	0.045	0.019	0.29%

Shaded SNPs are also among the 50 best genes for iris nevi, as computed by VEGAS

Table S14. Results of the bootstrap analyses examining evidence for signal enhancement in SNPs located within the genes identified as influencing neural patterns by Krasanova[60]. Using sampling with replacement we examined evidence for signal enhancement in 1000 bootstrapped sets of 219 genes of similar size, containing similar numbers of SNPs (Mean SNPs per gene; Krasanova: 113.3, sd 102.2; random sets 123.8, sd 10.15). For crypts we found 542 of the 24812 SNPs from the Krasanova set attained p<=.01, whereas only 19 sets of the 1000 randomly sampled gene sets yielded 542 or more SNPs *P*<=.01; yielding an empirical p-value of .019. Likewise, we found 399 of the 24812 SNPs from the Krasanova set attained p<=.01, whereas only 34 sets of the 1000 randomly sampled gene sets yielded 399 or more SNPs *P*<=.01; yielding an empirical p-value of .034 for furrows. The empirical p-values for the two pigmentation characteristics, Pigment Ring and Iris Nevi, act as negative controls in these analyses, yielding no evidence of signal enhancement.

Iris Characteristic	N SNPs p<.01 from Krasanova's gene set	N random gene sets yielding the an equal or greater number of SNPs p<.01 as observed in Krasanova's gene set[a]	Empirical Probability
Crypt	542	19	19/1001 = 0.019
Furrow	399	34	34/1001 = 0.034
Pigment Ring	289	339	339/1001 = 0.339
Iris Nevi	375	91	91/1001 = 0.091

Table S15. Correlations between iris characteristics and allelic profile scores

(i) the 50 most significant genes derived from the VEGAS analyses of the discovery sample (Tables S5-8);

(ii) the 50 most significant genes from the VEGAS analyses of the discovery sample that appear in the Krasanova gene set (Tables S10-13);

(iii) the union of (*i)* and (*ii)*; and finally,

(iv) to examine the possibility that these effects were being driven by major gene effects, the three most significant genes from (*ii)*.

In calculating the profile scores, we selected the SNP that showed the strongest association signal for each gene in the GWAS analyses of the discovery sample and weighting the number of risk alleles at each locus by the allelic effect (beta) from discovery sample. To examine the extent to which these scores could explain variation across phenotypes within the discovery sample and predict variation in the replication sample we compared the profile scores to the iris characteristics in unrelated individuals in the discovery sample and replication sample 2. The within sample and across sample correlations are given below.

	Number of SNPs in each gene set		**Iris ratings for Crypts**		**Iris ratings for Furrows**		**Iris ratings for Pigmented Ring**		**Iris ratings for Nevi**	
Allelic profile scores	**GWAS**	**Rep 2**	**GWAS**	**Rep 2**	**GWAS**	**Rep 2**	**GWAS**	**Rep 2**	**GWAS**	**Rep 2**
i Crypts	50	50	.41**	.13	-.11**	-.14	-.07*	-.23	.03	-.03
ii Crypts	50	50	.41**	.20	-.04	.07	.01	-.06	.06	.06
iii Crypt	95	95	.50**	.18	-.10**	-.05	-.05	-.18	.04	.01
iv Crypts	3	3	.14**	.23	-.05	-.26	.03	-.23	.06	-.21
i Furrow	50	50	-.08*	.07	.33**	.20	.05	-.08	.01	.07
ii Furrow	50	50	-.12**	-.07	.37**	-.18	.07*	-.07	.01	-.13
iii Furrow	97	97	-.11**	.03	.44**	.07	.07*	-.15	.02	.00
iv Furrow	3	3	-.11**	-.01	.12**	.13	.01	.09	.02	-.05
i Pigmented Ring	50	50	-.04	-.02	.08*	.21	.38**	.27	.03	.42**
ii Pigmented Ring	50	50	-.03	.10	.11**	-.37*	.28**	-.30	.08*	-.06
iii Pigmented Ring	99	99	-.05	.03	.12**	.07	.43**	.12	.06	.38*
iv Pigmented Ring	3	3	.05	-.20	.04	-.28	.08*	-.16	.01	-.03
i Nevi	50	50	-.02	-.07	.05	.06	.08*	.27*	.27**	.03
ii Nevi	50	50	-.03	-.07	.02	.06	.10**	.00	.33**	-.13
iii Nevi	95	95	-.02	-.06	.03	.06	.09**	.16	.39**	-.08
iv Nevi	3	3	-.01	-.01	-.01	-.17	.01	-.07	.10**	-.26

* $p < .05$. ** $p < .0$