

Analysis Summary: Unibrow

Phenotype Description

Data comes from the "Physical Features" survey:

- "Which image best matches the extent of your natural brow line (without hair removal)?"

Subjects match their eyebrow to one of four pictures. The resulting "unibrow" phenotype is scored from 0–3, with 0=no hair between the eyebrows and 3=lots of hair in between.

Phenotype Statistics

The following table shows demographics of unrelated, European individuals included in the GWAS.

Phenotype	Group	Total	M	F	(0,30]	(30,45]	(45,60]	(60,Inf]
unibrow	none	37652	18032	19620	4447	10555	10292	12358
	little	26134	15256	10878	4746	9892	6235	5261
	some	5066	3497	1569	796	2020	1259	991
	lots	432	338	94	61	150	107	114

The following table shows the phenotypic distribution across 23andMe genotyping platforms for individuals included in the GWAS.

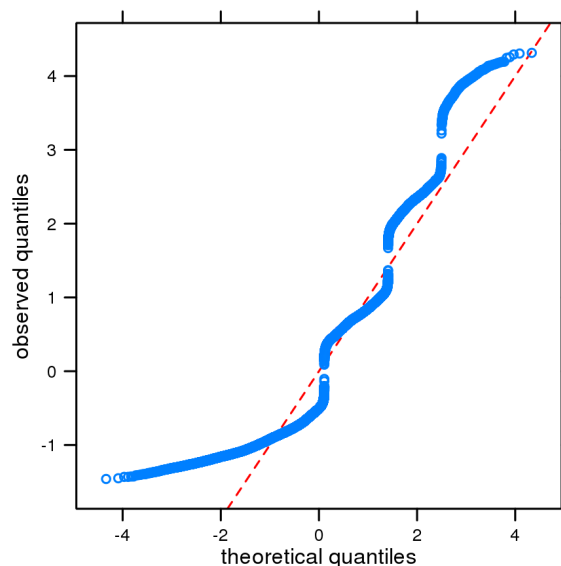
Phenotype	Group	Total	v1/v2	v3	v4
unibrow	none	37652	4505	31484	1663
	little	26134	3063	21959	1112
	some	5066	666	4206	194
	lots	432	55	354	23

Null Model with Covariates

The following table shows results of fitting a model for the trait based on just the covariates. Principal coordinates have been standardized, so these effect sizes are in units of standard deviations.

	Estimate	Std. Error	t value	Pr(> t)
age	-0.00594	0.000156	-38.1	0.0
sexF	-0.16928	0.004911	-34.5	3.5×10^{-258}
pc.0	-0.03161	0.002449	-12.9	4.6×10^{-38}
pc.1	-0.01742	0.002460	-7.1	1.5×10^{-12}
pc.2	0.01843	0.002458	7.5	6.5×10^{-14}
pc.3	-0.00345	0.002452	-1.4	0.16
pc.4	-0.00339	0.002452	-1.4	0.17

Q-Q plot of scaled residuals



SNP-level QC information

The following table shows results for QC filters on the genotyped data:

	failed	passed
no filters	0	1030430
not V1-only, chrM, chrY	4790	1025640
parent-offspring test	2129	1023511
MAF > 0%	3203	1020494
HWE > 1e-20	48225	972832
gt.rate > 90%	30775	952826
batch effects	28267	945446

The following table shows results for QC filters on the imputed dosage data:

	failed	passed
no filters	0	13733809
MAF > 0%	0	13733809
imputation quality	0	13733809
batch effects	2168	13731641

The following table shows results for QC filters on the merged association test results:

	passed	total
imputed only	12833621	12833621
both passed	898002	13731623
genotyped only	47444	13779067
no test result	-16606	13762461
MAF < 0.1%	-1722970	12039491

Genetic Association Tests

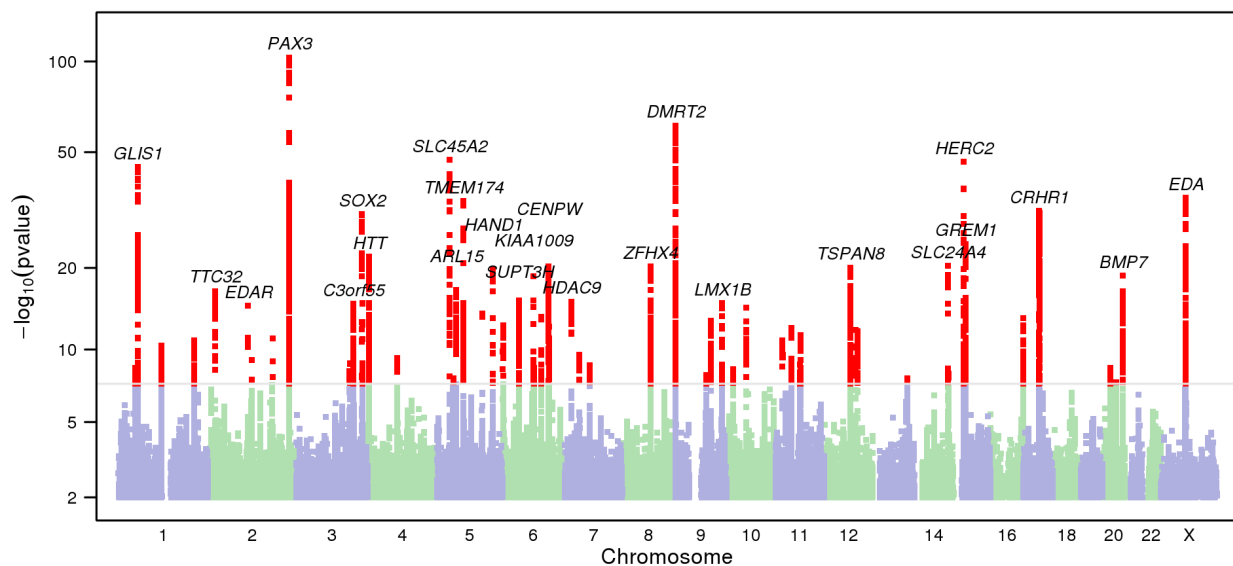
We performed linear regression assuming an additive model for allelic effects, using the model:

$$unibrow \sim age + sex + pc.0 + pc.1 + pc.2 + pc.3 + pc.4 + genotype$$

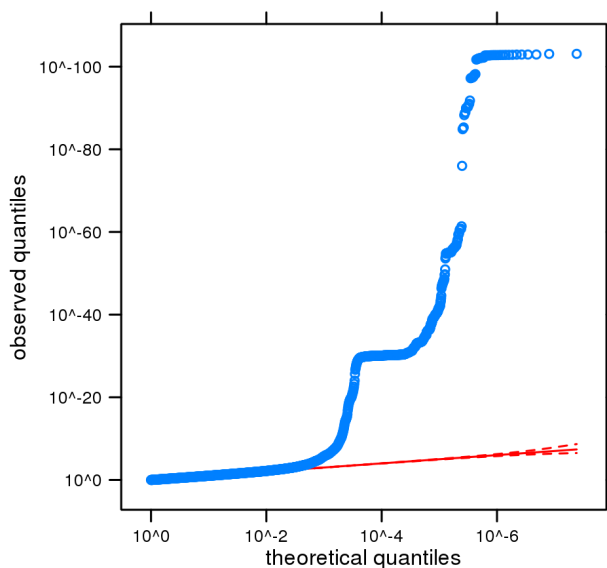
This genome-wide association analysis includes data from 69284 individuals of European ancestry, filtered to remove close relatives.

The results in this report have been adjusted for a genomic control inflation factor $\lambda=1.106$. The equivalent inflation factor rescaled for a sample size of 2000 would be $\lambda_{2000}=1.003$, and for 20000, $\lambda_{20000}=1.031$.

Manhattan Plot



Q-Q Plot of GWAS Results



Index SNPs for Strongest Associations

cytoband	assay.name	scaffold	position	alleles	src	pvalue	effect	95% CI	gene.context
2q36.1	rs2053316	chr2	223094489	C/T	I	7.9×10 ⁻¹⁰⁴	0.149	[0.135,0.162]	[PAX3]
9p24.3	rs7861975	chr9	1121353	C/T	I	4.2×10 ⁻⁶²	0.064	[0.056,0.071]	DMRT2--[--SMARCA2
5p13.2	rs16891982	chr5	33951693	C/G	I	6.8×10 ⁻⁴⁸	-0.148	[-0.168,-0.128]	[SLC45A2]
15q13.1	rs12916300	chr15	28410491	C/T	I	4.6×10 ⁻⁴⁷	-0.067	[-0.077,-0.058]	[HERC2]
1p32.3	rs702490	chr1	54195018	A/G	I	2.4×10 ⁻⁴⁵	-0.055	[-0.063,-0.047]	[GLIS1]
Xq13.1	rs2804362	chrX	69001854	A/G	I	8.9×10 ⁻³⁶	-0.037	[-0.043,-0.031]	[EDA]
5q13.2	rs6881396	chr5	72559339	G/T	I	6.7×10 ⁻³⁵	-0.046	[-0.054,-0.039]	TMEM174--[--FOXD1
17q21.31	17:43849572:AT_A	chr17	43849572	D/I	I	2.8×10 ⁻³²	-0.052	[-0.061,-0.044]	LRRC37A3---[--CRHR1
3q26.33	rs1345417	chr3	181511951	C/G	I	1.7×10 ⁻³¹	0.047	[0.039,0.055]	SOX2--[--ATP11B
15q13.3	rs3743103	chr15	33025627	C/T	I	5.3×10 ⁻²⁵	-0.038	[-0.045,-0.031]	[GREM1]
4p16.3	rs6828882	chr4	3166196	C/G	I	1.3×10 ⁻²²	-0.059	[-0.071,-0.048]	[HTT]
14q32.12	rs4144266	chr14	92789205	A/G	I	4.2×10 ⁻²¹	-0.036	[-0.043,-0.028]	CPSF2---[--SLC24A4
8q21.11	rs7837649	chr8	77658645	A/G	I	6.0×10 ⁻²¹	-0.046	[-0.056,-0.036]	[ZFHX4]
6q22.32	rs72422730	chr6	126825885	D/I	I	6.4×10 ⁻²¹	-0.035	[-0.042,-0.027]	CENPW---[--RSPO3
12q15	rs4760786	chr12	71446789	C/T	I	8.7×10 ⁻²¹	0.037	[0.029,0.044]	PTPRR---[--TSPAN8
5q33.2	rs199955185	chr5	153938270	D/I	I	1.8×10 ⁻²⁰	-0.036	[-0.043,-0.028]	HAND1--[--LARP1
20q13.31	rs230203	chr20	55791615	C/T	I	1.7×10 ⁻¹⁹	-0.034	[-0.041,-0.027]	[BMP7]
6q14.3	rs9449896	chr6	85118988	A/C	I	1.9×10 ⁻¹⁹	-0.044	[-0.053,-0.034]	KIAA1009---[--TBX18
5q11.2	rs1553477	chr5	53116966	A/C	I	2.3×10 ⁻¹⁷	-0.031	[-0.038,-0.024]	NDUFS4---[--ARL15
2p24.1	rs12467812	chr2	19995470	A/G	I	3.3×10 ⁻¹⁷	0.033	[0.025,0.040]	OSR1---[--TTC32
6p21.1	rs7227833	chr6	44681840	C/G	I	5.6×10 ⁻¹⁶	0.033	[0.025,0.042]	CDC5L---[--SUPT3H
7p21.1	rs1178101	chr7	18737197	A/C	I	7.3×10 ⁻¹⁶	0.039	[0.029,0.048]	[HDAC9]
9q33.3	rs3829849	chr9	129390800	C/T	I	1.2×10 ⁻¹⁵	0.030	[0.023,0.038]	[LMX1B]
3q25.32	rs67922471	chr3	157510696	D/I	I	1.4×10 ⁻¹⁵	-0.029	[-0.036,-0.022]	C3orf55---[--SHOX2
2q12.3	rs3827760	chr2	109513601	A/G	I	2.4×10 ⁻¹⁵	0.165	[0.124,0.206]	[EDAR]
10q21.1	rs7905367	chr10	54334653	C/G	I	4.4×10 ⁻¹⁵	-0.038	[-0.047,-0.028]	DKK1---[--MBL2
5q23.2	rs4476718	chr5	124075470	C/T	I	2.2×10 ⁻¹⁴	-0.037	[-0.047,-0.028]	[ZNF608]
6q21	rs61123157	chr6	106613530	C/T	I	4.6×10 ⁻¹⁴	0.028	[0.021,0.035]	PRDM1--[--ATG5
16q24.3	rs78498439	chr16	89967514	A/G	I	6.9×10 ⁻¹⁴	0.079	[0.059,0.100]	[TCF25]
9q22.32	rs199883506	chr9	98277366	D/I	I	1.3×10 ⁻¹³	-0.035	[-0.044,-0.026]	PTCH1-[---ERCC6L2
6p25.2	rs7763441	chr6	2452697	G/T	I	4.3×10 ⁻¹³	0.027	[0.020,0.034]	GMDS---[--C6orf195
11p11.2	rs66716358	chr11	44330610	C/T	I	7.6×10 ⁻¹³	-0.027	[-0.034,-0.019]	[ALX4]
12q21.31	rs11609649	chr12	85576492	A/G	I	1.3×10 ⁻¹²	0.033	[0.024,0.043]	[LRR1Q1]
12q21.33	rs11105799	chr12	91153721	C/T	I	1.8×10 ⁻¹²	0.028	[0.021,0.036]	[---CCER1
11q13.3	rs10626466	chr11	69683380	D/I	I	4.6×10 ⁻¹²	0.026	[0.019,0.033]	FGF3--[--ANO1
2q31.1	rs970797	chr2	177111819	G/T	I	9.0×10 ⁻¹²	-0.025	[-0.032,-0.018]	HOXD1--[--MTX2
1q32.2	rs7542091	chr1	210031948	C/G	I	1.5×10 ⁻¹¹	-0.026	[-0.033,-0.018]	DIEXF-[---SYT14
11p15.1	rs10500859	chr11	19740731	C/G	I	1.5×10 ⁻¹¹	-0.041	[-0.053,-0.029]	[NAV2]
9q22.32	rs111677654	chr9	98877903	C/T	I	3.8×10 ⁻¹¹	-0.032	[-0.042,-0.023]	[LOC158434]
1p12	rs6676180	chr1	119783223	A/T	I	4.3×10 ⁻¹¹	0.025	[0.018,0.033]	WARS2--[--HAO2
6p25.3	rs12203592	chr6	396321	C/T	I	1.4×10 ⁻¹⁰	0.031	[0.022,0.041]	[IRF4]
7p14.1	rs72328411	chr7	40319239	D/I	I	2.9×10 ⁻¹⁰	-0.075	[-0.099,-0.052]	[C7orf10]
4q21.21	rs3796606	chr4	81194090	A/G	I	5.5×10 ⁻¹⁰	0.023	[0.016,0.030]	[FGF5]
2q14.2	rs11694441	chr2	119541381	A/C	I	8.4×10 ⁻¹⁰	0.037	[0.025,0.049]	INSIG2---[--EN1
3q24	rs76036841	chr3	147804236	A/G	I	1.5×10 ⁻⁹	0.057	[0.039,0.076]	ZIC1---[--AGTR1
11q13.2	rs138341189	chr11	68224411	D/I	I	1.9×10 ⁻⁹	0.035	[0.023,0.046]	LRP5-[---PPP6R3

7q11.22	rs3094901	chr7	69937950	C/T	I	2.2×10 ⁻⁹	-0.022	[-0.029,-0.015]	[AUTS2]
20p11.22	rs6082544	chr20	21868713	A/G	I	3.5×10 ⁻⁹	-0.023	[-0.030,-0.015]	PAX1---[---FOXA2
1p33	rs1267303	chr1	46990457	A/G	I	3.6×10 ⁻⁹	0.025	[0.016,0.033]	DMBX1--[---KNCN
10p12.33	rs113334738	chr10	18271858	A/G	I	4.3×10 ⁻⁹	0.030	[0.020,0.040]	[SLC39A12]

Quality Statistics for Index SNPs

assay.name	is.v2	is.v3	is.v4	gt.rate	hw.p.value	p.date	freq.b	avg.rsqr	min.rsqr	p.batch	dose.b	qc.mask
rs2053316	FALSE	FALSE	FALSE					0.9648	0.9335	0.22	0.0770	v2v3v4
rs7861975	FALSE	TRUE	FALSE	0.9381	1.2×10 ⁻⁷⁴	1.9×10 ⁻⁵⁶	0.6223	0.9935	0.9919	0.014	0.6484	v2v3v4
rs16891982	TRUE	TRUE	TRUE	0.9997	4.3×10 ⁻²⁰	0.11	0.9573	0.9954	0.9915	1.8×10 ⁻⁴²	0.9494	v3
rs12916300	FALSE	FALSE	FALSE					0.7854	0.7531	2.5×10 ⁻³⁷	0.7077	v2v3v4
rs702490	FALSE	FALSE	FALSE					0.9710	0.8626	0.077	0.6567	v2v3v4
rs2804362	FALSE	FALSE	FALSE					0.9925	0.9836	0.0034	0.5398	v2v3v4
rs6881396	FALSE	FALSE	FALSE					0.9627	0.9510	0.0072	0.5980	v2v3v4
17:43849572:AT_A	FALSE	FALSE	FALSE					0.9774	0.9747	0.26	0.7749	v2v3v4
rs1345417	FALSE	FALSE	FALSE					0.8573	0.4988	0.56	0.5895	v2v3v4
rs3743103	FALSE	TRUE	FALSE	0.9998	0.88	0.018	0.5786	0.9926	0.9729	0.22	0.5797	v2v3v4
rs6828882	FALSE	FALSE	FALSE					0.9781	0.9742	0.017	0.1042	v2v3v4
rs4144266	FALSE	FALSE	FALSE					0.9458	0.8976	8.2×10 ⁻⁵	0.4434	v2v3v4
rs7837649	FALSE	FALSE	FALSE					0.9842	0.9784	0.73	0.8324	v2v3v4
rs72422730	FALSE	FALSE	FALSE					0.9785	0.9762	0.00014	0.5362	v2v3v4
rs4760786	FALSE	FALSE	FALSE					0.9946	0.9931	0.00020	0.6834	v2v3v4
rs199955185	FALSE	FALSE	FALSE					0.9065	0.8736	0.0017	0.4142	v2v3v4
rs230203	FALSE	FALSE	FALSE					0.9322	0.9211	0.045	0.4475	v2v3v4
rs9449896	FALSE	FALSE	FALSE					0.9872	0.9706	0.0044	0.1671	v2v3v4
rs1553477	FALSE	FALSE	FALSE					0.9894	0.9520	0.093	0.5395	v2v3v4
rs12467812	TRUE	FALSE	FALSE	0.9997	0.38	0.63	0.3330	0.9806	0.9553	0.017	0.3300	v2v3v4
rs227833	FALSE	FALSE	FALSE					0.9995	0.9965	0.46	0.7368	v2v3v4
rs1178101	FALSE	FALSE	FALSE					0.9936	0.9892	0.056	0.8233	v2v3v4
rs3829849	TRUE	TRUE	TRUE	0.9998	0.054	0.048	0.3584	0.9993	0.9988	0.41	0.3598	v2v3v4
rs67922471	FALSE	FALSE	FALSE					0.9781	0.9667	0.19	0.4837	v2v3v4
rs3827760	TRUE	TRUE	TRUE	1.0000	0.13	0.0089	0.0077	0.9883	0.9711	0.013	0.0081	v2v3v4
rs7905367	FALSE	FALSE	FALSE					0.8414	0.8082	0.0015	0.2193	v2v3v4
rs4476718	FALSE	FALSE	FALSE					0.9678	0.8677	1.5×10 ⁻¹⁵	0.8316	v2v3v4
rs61123157	FALSE	FALSE	FALSE					0.9879	0.9613	0.62	0.4053	v2v3v4
rs78498439	FALSE	FALSE	FALSE					0.6209	0.6043	4.7×10 ⁻⁹	0.9498	v2v3v4
rs199883506	FALSE	FALSE	FALSE					0.8928	0.8076	0.86	0.7940	v2v3v4
rs7763441	FALSE	FALSE	FALSE					0.9946	0.9857	0.11	0.5874	v2v3v4
rs66716358	FALSE	FALSE	FALSE					0.9619	0.8614	0.065	0.4935	v2v3v4
rs11609649	TRUE	TRUE	TRUE	0.9998	0.72	0.30	0.8153	0.9998	0.9996	0.64	0.8154	v2v3v4
rs11105799	FALSE	FALSE	FALSE					0.9912	0.9510	0.11	0.2854	v2v3v4
rs10626466	FALSE	FALSE	FALSE					0.9538	0.9397	0.34	0.4181	v2v3v4
rs970797	TRUE	TRUE	TRUE	0.9997	0.45	0.0039	0.4326	0.9997	0.9993	0.093	0.4334	v2v3v4
rs7542091	FALSE	FALSE	FALSE					0.9685	0.9549	0.91	0.6254	v2v3v4
rs10500859	FALSE	FALSE	FALSE					0.9393	0.9267	0.35	0.1058	v2v3v4
rs111677654	FALSE	FALSE	FALSE					0.9843	0.9723	0.28	0.1674	v2v3v4
rs6676180	FALSE	FALSE	FALSE					0.9712	0.9066	0.0043	0.6325	v2v3v4
rs12203592	TRUE	TRUE	TRUE	1.0000	9.7×10 ⁻³⁷	0.014	0.1728	0.9923	0.9819	4.2×10 ⁻⁶	0.1705	v2v3v4
rs72328411	FALSE	FALSE	FALSE					0.9681	0.9134	0.56	0.9757	v2v3v4
rs3796606	TRUE	TRUE	TRUE	0.9994	0.051	0.031	0.4191	0.9996	0.9993	0.54	0.4196	v2v3v4
rs11694441	FALSE	FALSE	FALSE					0.8667	0.8546	0.0048	0.8841	v2v3v4
rs76036841	FALSE	FALSE	FALSE					0.6946	0.6507	0.0081	0.0544	v2v3v4
rs138341189	FALSE	FALSE	FALSE					0.8640	0.8551	0.97	0.8691	v2v3v4
rs3094901	FALSE	FALSE	FALSE					0.9978	0.9953	0.83	0.4956	v2v3v4
rs6082544	FALSE	FALSE	FALSE					0.9232	0.9194	0.014	0.5640	v2v3v4
rs1267303	FALSE	TRUE	FALSE	0.9989	0.16	0.13	0.2558	0.9882	0.9561	0.0024	0.2585	v2v3v4
rs113334738	FALSE	FALSE	FALSE					0.9894	0.9786	0.15	0.8487	v2v3v4

SNP Statistics in the GWAS Sample

assay.name	AA.0	AB.0	BB.0	im.num.0	dose.b.0
rs2053316				69284	0.0781
rs7861975	7294	26600	19082	69284	0.6437
rs16891982	154	5312	63817	57281	0.9598
rs12916300				69284	0.7177
rs702490				69284	0.6588
rs2804362				69284	0.5353
rs6881396				69284	0.5963
17:43849572:AT_A				69284	0.7759
rs1345417				69284	0.5890
rs3743103	10247	28347	19398	69284	0.5796
rs6828882				69284	0.1015
rs4144266				69284	0.4517
rs7837649				69284	0.8334
rs72422730				69284	0.5394
rs4760786				69284	0.6788
rs199955185				69284	0.4116
rs230203				69284	0.4477
rs9449896				69284	0.1712
rs1553477				69284	0.5408
rs12467812	4810	4778	1165	69284	0.3278
rs227833				69284	0.7379
rs1178101				69284	0.8246
rs3829849	28491	31865	8912	69284	0.3591
rs67922471				69284	0.4833
rs3827760	68056	1052	7	69284	0.0078
rs7905367				69284	0.2180
rs4476718				69284	0.8290

rs61123157				69284	0.4051
rs78498439				69284	0.9497
rs199883506				69284	0.7957
rs7763441				69284	0.5892
rs66716358				69284	0.4932
rs11609649	2333	20802	46141	69284	0.8165
rs11105799				69284	0.2829
rs10626466				69284	0.4213
rs970797	22352	34078	12833	69284	0.4313
rs7542091				69284	0.6233
rs10500859				69284	0.1054
rs111677654				69284	0.1679
rs6676180				69284	0.6312
rs12203592	47467	19486	2327	69284	0.1747
rs72328411				69284	0.9757
rs3796606	23402	33779	12076	69284	0.4182
rs11694441				69284	0.8815
rs76036841				69284	0.0556
rs138341189				69284	0.8679
rs3094901				69284	0.4917
rs6082544				69284	0.5636
rs1267303	32122	21981	3826	69284	0.2574
rs113334738				69284	0.8498

Annotations from NHGRI GWAS Catalog

The following table shows, for each index SNP, all entries in the NHGRI GWAS Catalog that are within 500kb and in at least moderate linkage disequilibrium ($r^2 > 0.5$).

region	position	our.name	our.pval	dist	rsqr	assay.name	pvalue	pubmed.id	trait	genes
5p13.2	33951693	rs16891982	6.8×10^{-48}	0	1.000	rs16891982	4.0×10^{-20}	20585627	Hair color	SLC45A2
5p13.2	33951693	rs16891982	6.8×10^{-48}	0	1.000	rs16891982	1.0×10^{-12}	20585627	Eye color	SLC45A2
5p13.2	33951693	rs16891982	6.8×10^{-48}	0	1.000	rs16891982	3.0×10^{-11}	17999355	Skin pigmentation	SLC45A2
5p13.2	33951693	rs16891982	6.8×10^{-48}	3633	0.514	rs35390	2.0×10^{-7}	21983787	Melanoma	SLC45A2
5p13.2	33951693	rs16891982	6.8×10^{-48}	3980	0.514	rs35391	3.0×10^{-10}	19340012	Tanning	MATP
5p13.2	33951693	rs16891982	6.8×10^{-48}	7266	0.733	rs28777	9.0×10^{-14}	18483556	Black vs. red hair color	MATP
5p13.2	33951693	rs16891982	6.8×10^{-48}	7266	0.733	rs28777	1.0×10^{-17}	18483556	Black vs. blond hair color	MATP
15q13.1	28410491	rs12916300	4.6×10^{-47}	-53632	0.868	rs1129038	4.0×10^{-8}	22561518	Vitiligo	OCA2, HERC2
15q13.1	28410491	rs12916300	4.6×10^{-47}	-44873	0.880	rs12913832	1.0×10^{-22}	23548203	Tanning	HERC2
15q13.1	28410491	rs12916300	4.6×10^{-47}	-44873	0.880	rs12913832	1.0×10^{-167}	23548203	Hair color	HERC2
15q13.1	28410491	rs12916300	4.6×10^{-47}	-44873	0.880	rs12913832	1.0×10^{-158}	23548203	Eye color	HERC2
15q13.1	28410491	rs12916300	4.6×10^{-47}	-44873	0.880	rs12913832	9.0×10^{-88}	20585627	Hair color	OCA2
15q13.1	28410491	rs12916300	4.6×10^{-47}	-44873	0.880	rs12913832	3.0×10^{-52}	20585627	Eye color	OCA2
15q13.1	28410491	rs12916300	4.6×10^{-47}	-44873	0.880	rs12913832	1.0×10^{-300}	20463881	Eye color traits	HERC2, OCA
15q13.1	28410491	rs12916300	4.6×10^{-47}	-44873	0.880	rs12913832	1.0×10^{-77}	18483556	Black vs. red hair color	HERC2
15q13.1	28410491	rs12916300	4.6×10^{-47}	-44873	0.880	rs12913832	4.0×10^{-103}	18483556	Black vs. blond hair color	HERC2
15q13.1	28410491	rs12916300	4.6×10^{-47}	102873	0.591	rs916977	1.0×10^{-43}	18252221	Iris color	HERC2
15q13.1	28410491	rs12916300	4.6×10^{-47}	119691	0.617	rs1667394	2.0×10^{-20}	23118974	Eye color	HERC2, OCA2
15q13.1	28410491	rs12916300	4.6×10^{-47}	119691	0.617	rs1667394	2.0×10^{-53}	17952075	Blue vs. green eyes	OCA2
15q13.1	28410491	rs12916300	4.6×10^{-47}	119691	0.617	rs1667394	1.0×10^{-241}	17952075	Blue vs. brown eyes	OCA2
15q13.1	28410491	rs12916300	4.6×10^{-47}	119691	0.617	rs1667394	6.0×10^{-35}	17952075	Blond vs. brown hair color	OCA2
Xq13.1	69001854	rs2804362	8.9×10^{-36}	-196536	0.644	rs4844096	5.0×10^{-11}	20195514	Primary tooth development (number of teeth)	EDA
Xq13.1	69001854	rs2804362	8.9×10^{-36}	-108938	0.572	rs5936487	6.0×10^{-11}	20195514	Primary tooth development (time to first tooth eruption)	EDA
5q13.2	72559339	rs6881396	6.7×10^{-35}	-8205	0.920	rs7702331	6.0×10^{-10}	23128233	Crohn's disease	Intergenic
5q13.2	72559339	rs6881396	6.7×10^{-35}	-8205	0.920	rs7702331	6.0×10^{-12}	21102463	Crohn's disease	intergenic
14q32.12	92789205	rs4144266	4.2×10^{-21}	-25814	0.926	rs4900109	8.0×10^{-21}	21835309	Iris characteristics	SLC324A4
14q32.12	92789205	rs4144266	4.2×10^{-21}	-15542	0.926	rs12896399	2.0×10^{-36}	23548203	Hair color	SLC24A4
14q32.12	92789205	rs4144266	4.2×10^{-21}	-15542	0.926	rs12896399	4.0×10^{-11}	23548203	Eye color	SLC24A4
14q32.12	92789205	rs4144266	4.2×10^{-21}	-15542	0.926	rs12896399	5.0×10^{-13}	20585627	Hair color	SLC24A4
14q32.12	92789205	rs4144266	4.2×10^{-21}	-15542	0.926	rs12896399	1.0×10^{-16}	20585627	Eye color	SLC24A4
14q32.12	92789205	rs4144266	4.2×10^{-21}	-15542	0.926	rs12896399	8.0×10^{-21}	18483556	Black vs. blond hair color	SLC24A4
14q32.12	92789205	rs4144266	4.2×10^{-21}	-15542	0.926	rs12896399	4.0×10^{-38}	17952075	Blue vs. green eyes	SLC24A4
14q32.12	92789205	rs4144266	4.2×10^{-21}	-15542	0.926	rs12896399	1.0×10^{-48}	17952075	Blond vs. brown hair color	SLC24A4
8q21.11	77658645	rs7837649	6.0×10^{-21}	-47020	0.934	rs4735738	5.0×10^{-6}	23667675	Menarche (age at onset)	ZFXH4
6q22.32	126825885	rs72422730	6.4×10^{-21}	-127166	0.937	rs9388489	4.0×10^{-13}	19430480	Type 1 diabetes	C6orf173
6q22.32	126825885	rs72422730	6.4×10^{-21}	-58285	0.953	rs1361108	9.0×10^{-6}	21998595	Height	Intergenic
6q22.32	126825885	rs72422730	6.4×10^{-21}	-58285	0.953	rs1361108	2.0×10^{-8}	21102462	Menarche (age at onset)	C6orf173, TRMT11
6q22.32	126825885	rs72422730	6.4×10^{-21}	9770	0.958	rs1490388	6.0×10^{-7}	18391951	Height	C6orf173
6q22.32	126825885	rs72422730	6.4×10^{-21}	25275	0.822	rs1490384	1.0×10^{-16}	23563607	Height	C6orf173
6q22.32	126825885	rs72422730	6.4×10^{-21}	25275	0.822	rs1490384	4.0×10^{-21}	20881960	Height	C6orf173
6q22.32	126825885	rs72422730	6.4×10^{-21}	140423	0.748	rs4549631	5.0×10^{-13}	18391952	Height	LOC387103
6p25.3	396321	rs12203592	1.4×10^{-10}	0	1.000	rs12203592	3.0×10^{-23}	23548203	Tanning	IRF4
6p25.3	396321	rs12203592	1.4×10^{-10}	0	1.000	rs12203592	2.0×10^{-6}	23548203	Sunburns	IRF4
6p25.3	396321	rs12203592	1.4×10^{-10}	0	1.000	rs12203592	7.0×10^{-14}	23548203	Non-melanoma skin cancer	IRF4
6p25.3	396321	rs12203592	1.4×10^{-10}	0	1.000	rs12203592	1.0×10^{-28}	23548203	Hair color	IRF4
6p25.3	396321	rs12203592	1.4×10^{-10}	0	1.000	rs12203592	6.0×10^{-15}	21685912	Progressive supranuclear palsy	IRF4
6p25.3	396321	rs12203592	1.4×10^{-10}	0	1.000	rs12203592	4.0×10^{-7}	20585627	Hair color	IRF4
6p25.3	396321	rs12203592	1.4×10^{-10}	0	1.000	rs12203592	2.0×10^{-91}	20585627	Freckling	IRF4
6p25.3	396321	rs12203592	1.4×10^{-10}	0	1.000	rs12203592	2.0×10^{-15}	20585627	Eye color	IRF4

6p25.3	396321	rs12203592	1.4×10 ⁻¹⁰	0	1.000	rs12203592	9.0×10 ⁻²⁸	18483556	Black vs. red hair color	IRF4
6p25.3	396321	rs12203592	1.4×10 ⁻¹⁰	0	1.000	rs12203592	7.0×10 ⁻¹²⁷	18483556	Black vs. blond hair color	IRF4
11q13.2	68224411	rs138341189	1.9×10 ⁻⁹	-23116	0.612	rs3736228	2.0×10 ⁻²⁶	22504420	Bone mineral density	LRP5
11q13.2	68224411	rs138341189	1.9×10 ⁻⁹	-23116	0.612	rs3736228	6.0×10 ⁻¹²	18455228	Bone mineral density	LRP5
20p11.22	21868713	rs6082544	3.5×10 ⁻⁹	-15613	0.852	rs2180439	4.0×10 ⁻¹⁷	22032556	Male-pattern baldness	Intergenic PAX1, BQ013595, BE789145
20p11.22	21868713	rs6082544	3.5×10 ⁻⁹	-15613	0.852	rs2180439	3.0×10 ⁻¹⁵	18849994	Male-pattern baldness	PAX1, FOXA2
20p11.22	21868713	rs6082544	3.5×10 ⁻⁹	168862	0.500	rs6047844	2.0×10 ⁻³⁹	22693459	Male-pattern baldness	PAX1, FOXA2

Replication of GWAS Catalog Results

The following table shows, for each GWAS Catalog result for similar traits, our association test result for our best available proxy (distance < 100kb, r² > 0.8).

no relevant GWAS catalog terms were available

Nearby Nonsynonymous SNPs

region	position	our.name	our.pval	dist	rsqr	assay.name	gene	aa.chg
5p13.2	33951693	rs16891982	6.8×10 ⁻⁴⁸	0	1.000	rs16891982	SLC45A2	F374L
4p16.3	3166196	rs6828882	1.3×10 ⁻²²	23351	0.920	rs363125	HTT	T1720N
2q12.3	109513601	rs3827760	2.4×10 ⁻¹⁵	0	1.000	rs3827760	EDAR	V370A
16q24.3	89967514	rs78498439	6.9×10 ⁻¹⁴	159479	0.512	rs150196149	PRDM7	L330P
11p11.2	44330610	rs66716358	7.6×10 ⁻¹³	699	0.817	rs12421995	ALX4	P102S
11p11.2	44330610	rs66716358	7.6×10 ⁻¹³	899	0.989	rs3824915	ALX4	R35T
12q21.31	85576492	rs11609649	1.3×10 ⁻¹²	-137993	0.504	rs3765044	LRRIQ1	C83Y
12q21.31	85576492	rs11609649	1.3×10 ⁻¹²	-109769	0.504	rs17012533	LRRIQ1	A912T
11q13.2	68224411	rs138341189	1.9×10 ⁻⁹	-23116	0.612	rs3736228	LRP5	A1330V

Nearby Expression QTLs

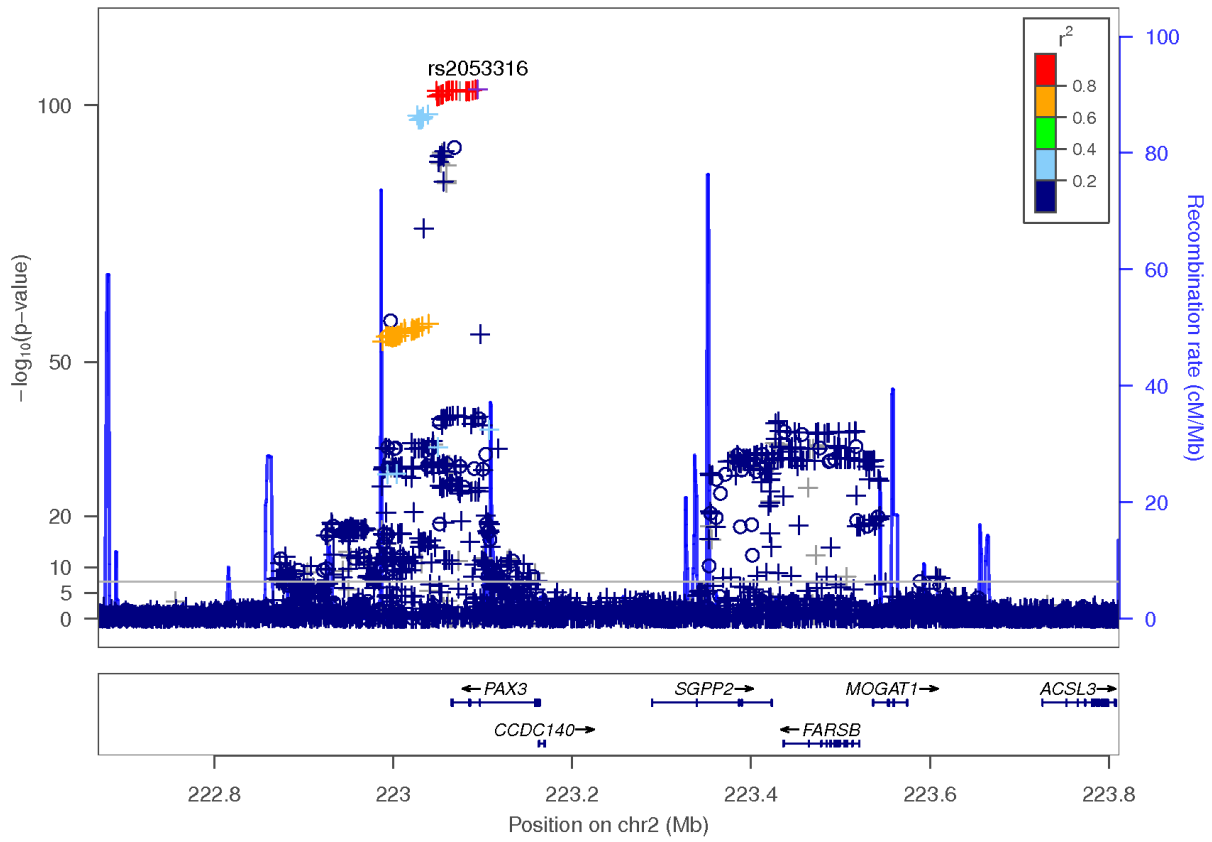
region	position	our.name	our.pval	dist	rsqr	assay.name	eqtl.dist	eqtl.gene	eqtl.pval	eqtl.rsqr	tissue	pubmed.id
5q33.2	153938270	rs199955185	1.8×10 ⁻²⁰	-18461	0.652	rs11749542	278243	C5orf4	0.00021	0.172	Fibroblast	19644074
20q13.31	55791615	rs230203	1.7×10 ⁻¹⁹	14725	0.623	rs186659	-183952	HS.132453	0.00020	0.048	Monocyte	22446964
5q11.2	53116966	rs1553477	2.3×10 ⁻¹⁷	37997	0.714	rs271251	-200566	NDUFS4	2.4×10 ⁻⁵	0.062	Monocyte	22446964
6p21.1	44681840	rs227833	5.6×10 ⁻¹⁶	118175	0.658	rs542444	545675	SUPT3H	2.2×10 ⁻¹²	0.125	Lymphoblastoid	24037378
6p21.1	44681840	rs227833	5.6×10 ⁻¹⁶	136028	0.821	rs4714813	211208	SUPT3H	6.9×10 ⁻¹⁰	0.165	Lymphoblastoid	17873874
16q24.3	89967514	rs78498439	6.9×10 ⁻¹⁴	18630	0.823	rs1805008	238999	CDK10	2.6×10 ⁻²⁴	0.244	Lymphoblastoid	24037378
16q24.3	89967514	rs78498439	6.9×10 ⁻¹⁴	18630	0.823	rs1805008	-223556	CDK10	4.9×10 ⁻⁶	0.072	Monocyte	22446964
16q24.3	89967514	rs78498439	6.9×10 ⁻¹⁴	18630	0.823	rs1805008	-223556	CDK10	1.8×10 ⁻⁷	0.093	B-Cell	22446964
16q24.3	89967514	rs78498439	6.9×10 ⁻¹⁴	18630	0.823	rs1805008	-212494	C16ORF7	1.4×10 ⁻⁸	0.109	B-Cell	22446964
11p11.2	44330610	rs66716358	7.6×10 ⁻¹³	-15220	0.595	rs1828656	325581	CD82	0.00038	0.044	Monocyte	22446964
2q31.1	177111819	rs970797	9.0×10 ⁻¹²	-291754	1.000	rs970797	-154287	HOXD13	2.8×10 ⁻⁵		Liver	18462017
2q31.1	177111819	rs970797	9.0×10 ⁻¹²	-291754	1.000	rs970797	983217	NFE2L2	0.0023		Liver	18462017
4q21.21	81194090	rs3796606	5.5×10 ⁻¹⁰	6555	0.961	rs6827834	773916	BMP3	0.0014	0.036	B-Cell	22446964
11q13.2	68224411	rs138341189	1.9×10 ⁻⁹	-23116	0.612	rs3736228	320795	CPT1A	3.9×10 ⁻¹⁷	0.047	Monocyte	20502693
11q13.2	68224411	rs138341189	1.9×10 ⁻⁹	-23116	0.612	rs3736228	320955	CPT1A	4.9×10 ⁻⁵	0.057	Monocyte	22446964
1p33	46990457	rs1267303	3.6×10 ⁻⁹	-230152	0.669	rs1267308	-831072	TMEM69	0.00018		Lymphoblastoid	20220756
1p33	46990457	rs1267303	3.6×10 ⁻⁹	-2739	0.669	rs1267308	-1867807	TMEM53	0.00020	0.048	Monocyte	22446964

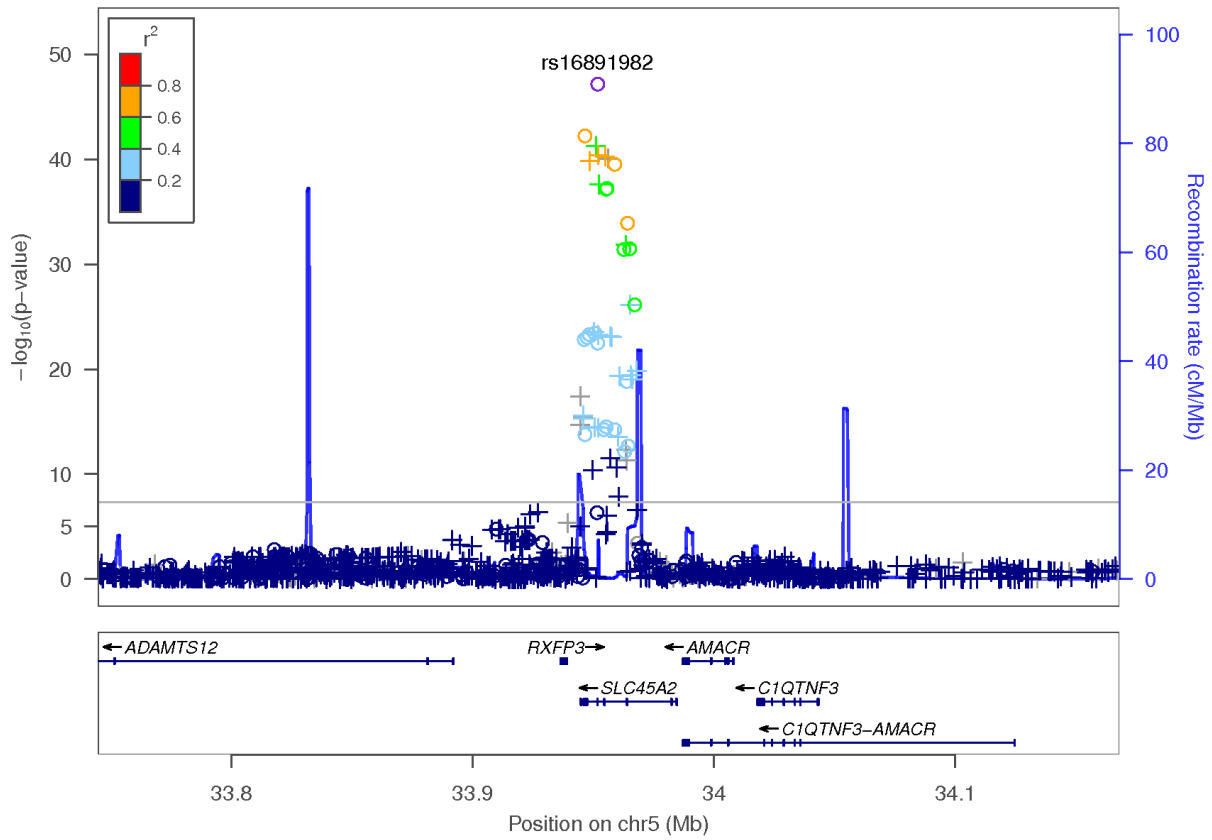
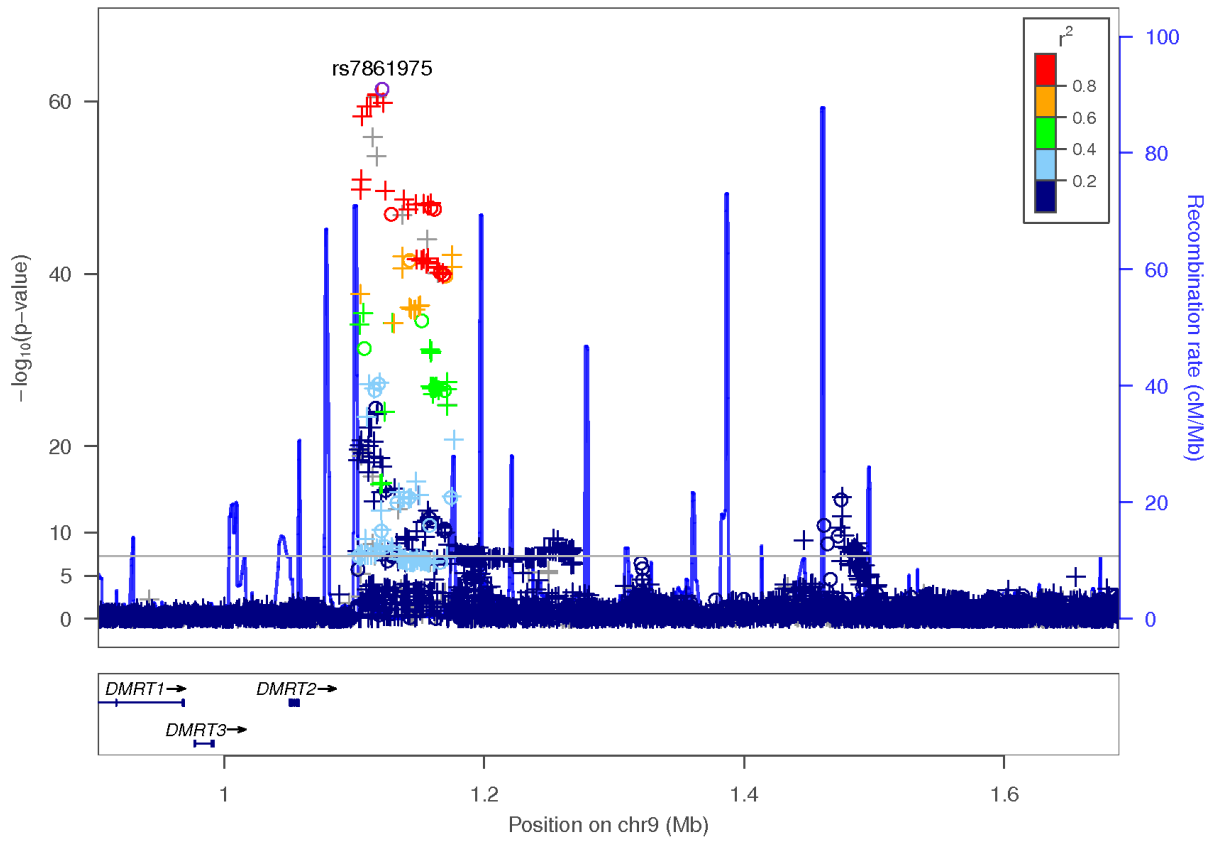
Nearby Clinical Variants

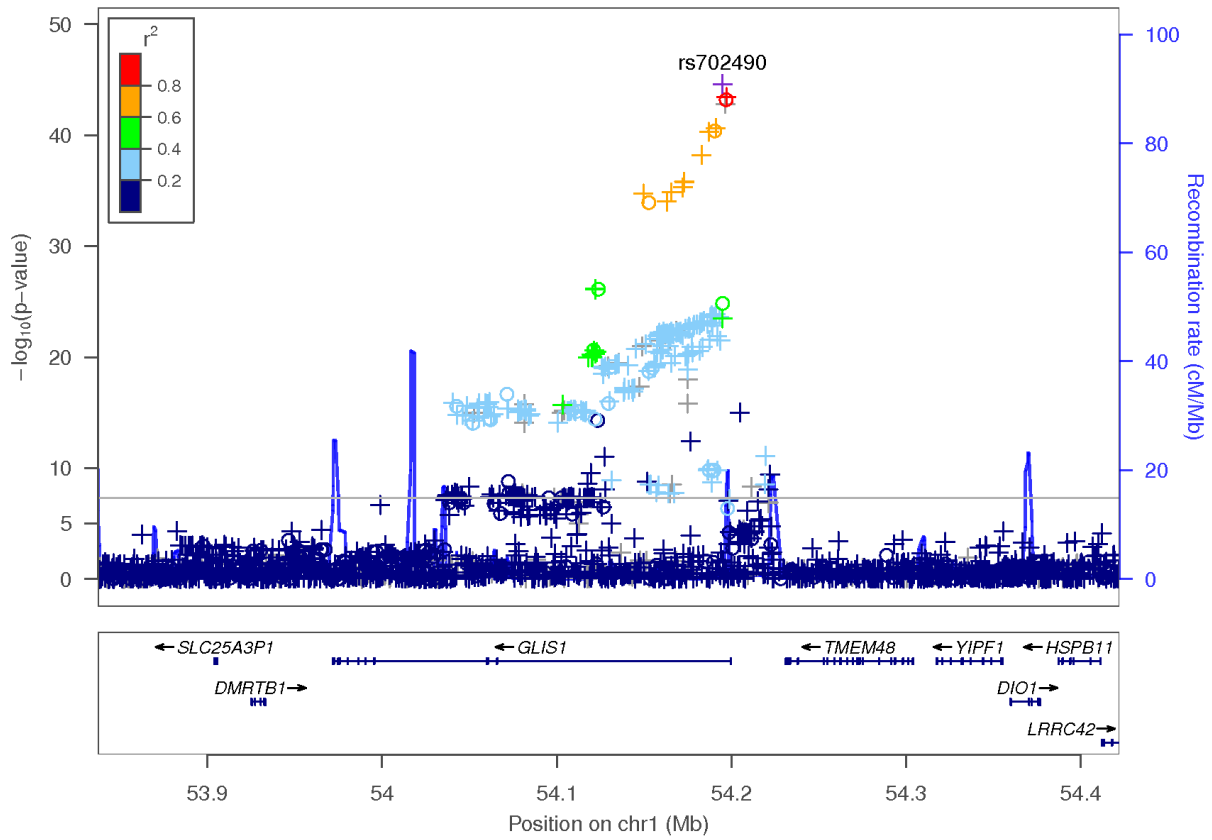
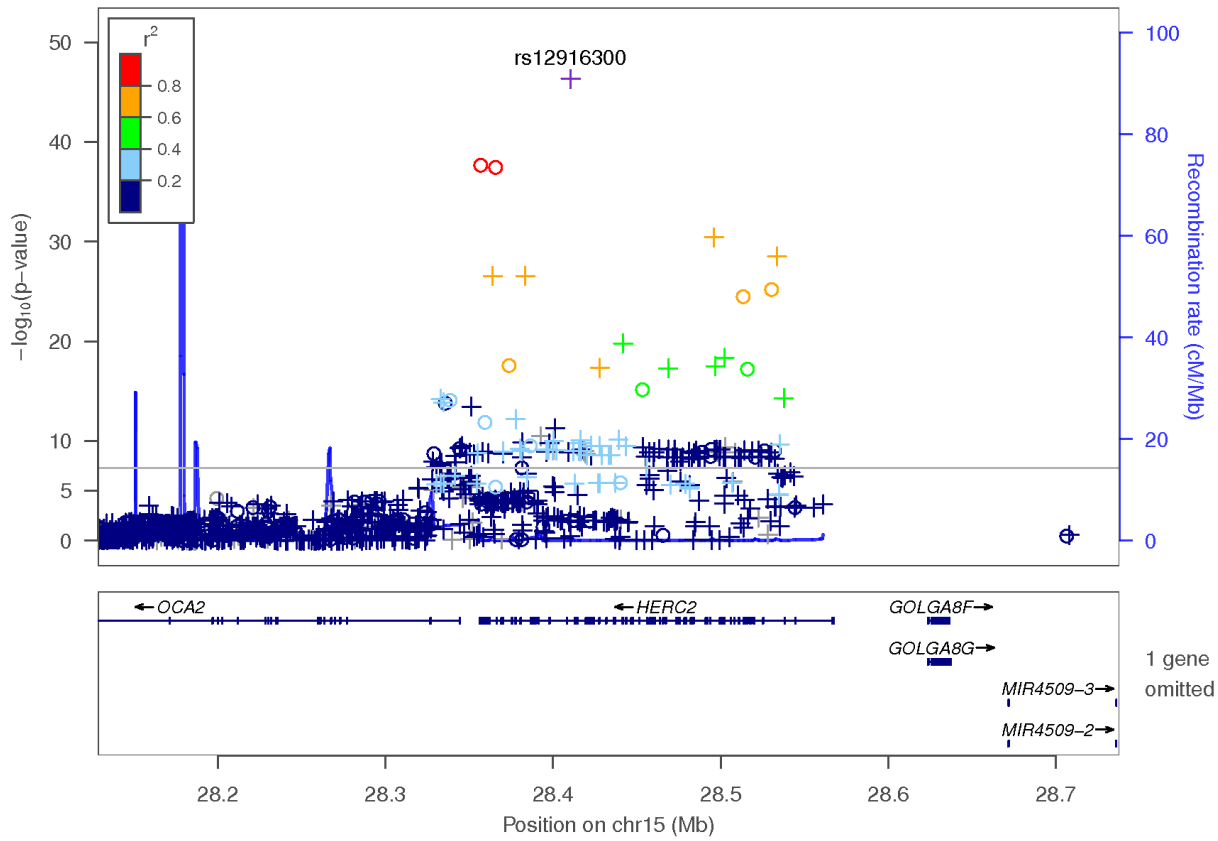
source	region	our.name	our.pval	dist	rsqr	assay.name	gene	phenotype	accession
clinvar	5p13.2	rs16891982	6.8×10 ⁻⁴⁸	0	1.000	rs16891982	SLC45A2	Skin/hair/eye pigmentation, variation in, 5	NCBI curation
clinvar	5p13.2	rs16891982	6.8×10 ⁻⁴⁸	0	1.000	rs16891982	SLC45A2	Oculocutaneous albinism type 4	NCBI curation
clinvar	15q13.1	rs12916300	4.6×10 ⁻⁴⁷	-44873	0.880	rs12913832	HERC2	Skin/hair/eye pigmentation, variation in, 1	NCBI curation Genetics Home Referenceprader-willi- syndrome
clinvar	15q13.1	rs12916300	4.6×10 ⁻⁴⁷	-44873	0.880	rs12913832	HERC2	Prader-Willi syndrome	SNOMED CT89392001
clinvar	15q13.1	rs12916300	4.6×10 ⁻⁴⁷	-44873	0.880	rs12913832	HERC2	Prader-Willi syndrome Mental retardation, autosomal recessive 38	NCBI curation
clinvar	14q32.12	rs4144266	4.2×10 ⁻²¹	-15542	0.926	rs12896399	SLC24A4	Skin/hair/eye pigmentation, variation in, 6	NCBI curation
clinvar	14q32.12	rs4144266	4.2×10 ⁻²¹	-15542	0.926	rs12896399	SLC24A4	AMELOGENESIS IMPERFECTA, HYPOMATURATION TYPE, IIA5	OMIM615887
clinvar	2q12.3	rs3827760	2.4×10 ⁻¹⁵	0	1.000	rs3827760	EDAR	Autosomal recessive hypohidrotic ectodermal dysplasia syndrome	SNOMED CT27025001
clinvar	2q12.3	rs3827760	2.4×10 ⁻¹⁵	0	1.000	rs3827760	EDAR	Autosomal dominant hypohidrotic ectodermal dysplasia	NCBI curation
clinvar	2q12.3	rs3827760	2.4×10 ⁻¹⁵	0	1.000	rs3827760	EDAR	Hair morphology 1	NCBI curation
clinvar	16q24.3	rs78498439	6.9×10 ⁻¹⁴	18630	0.823	rs1805008	MC1R	Cutaneous malignant melanoma 5	NCBI curation
clinvar	16q24.3	rs78498439	6.9×10 ⁻¹⁴	18630	0.823	rs1805008	MC1R	Skin/hair/eye pigmentation, variation in, 2	NCBI curation

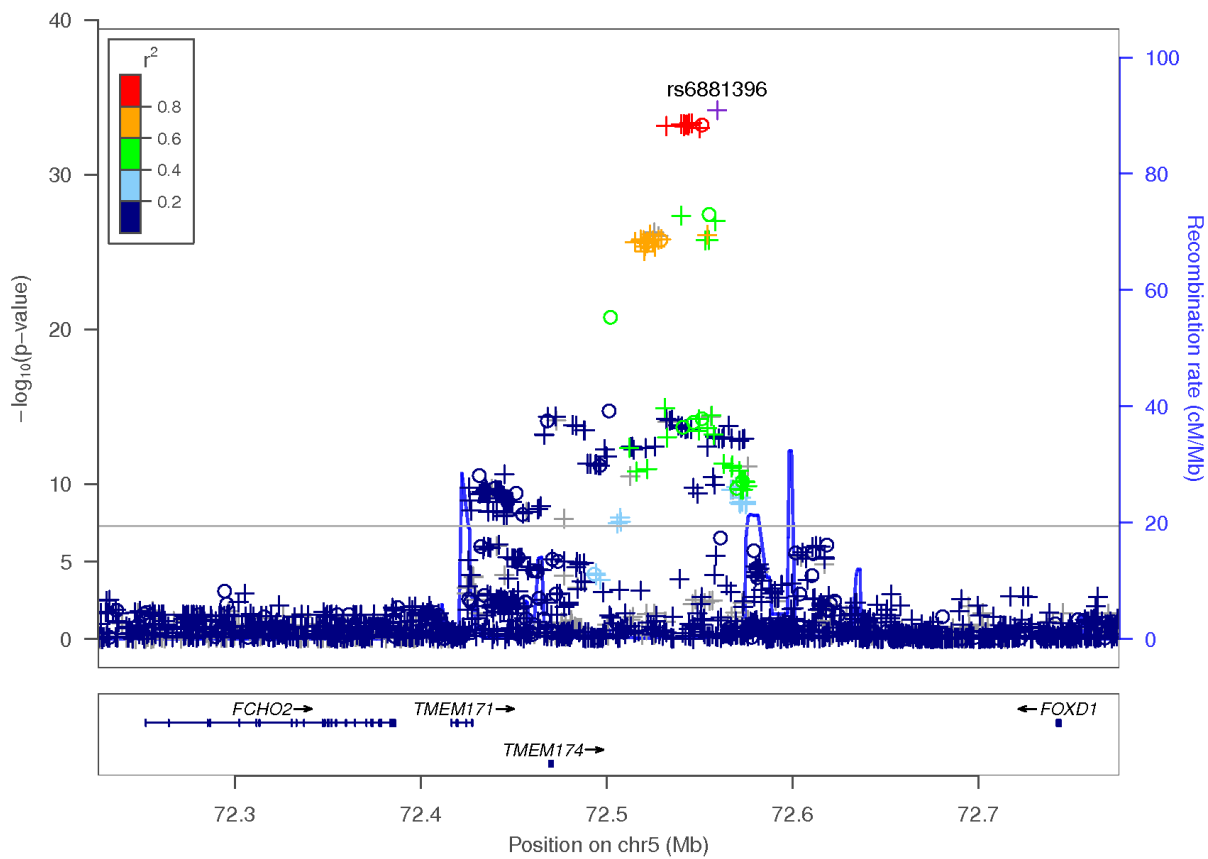
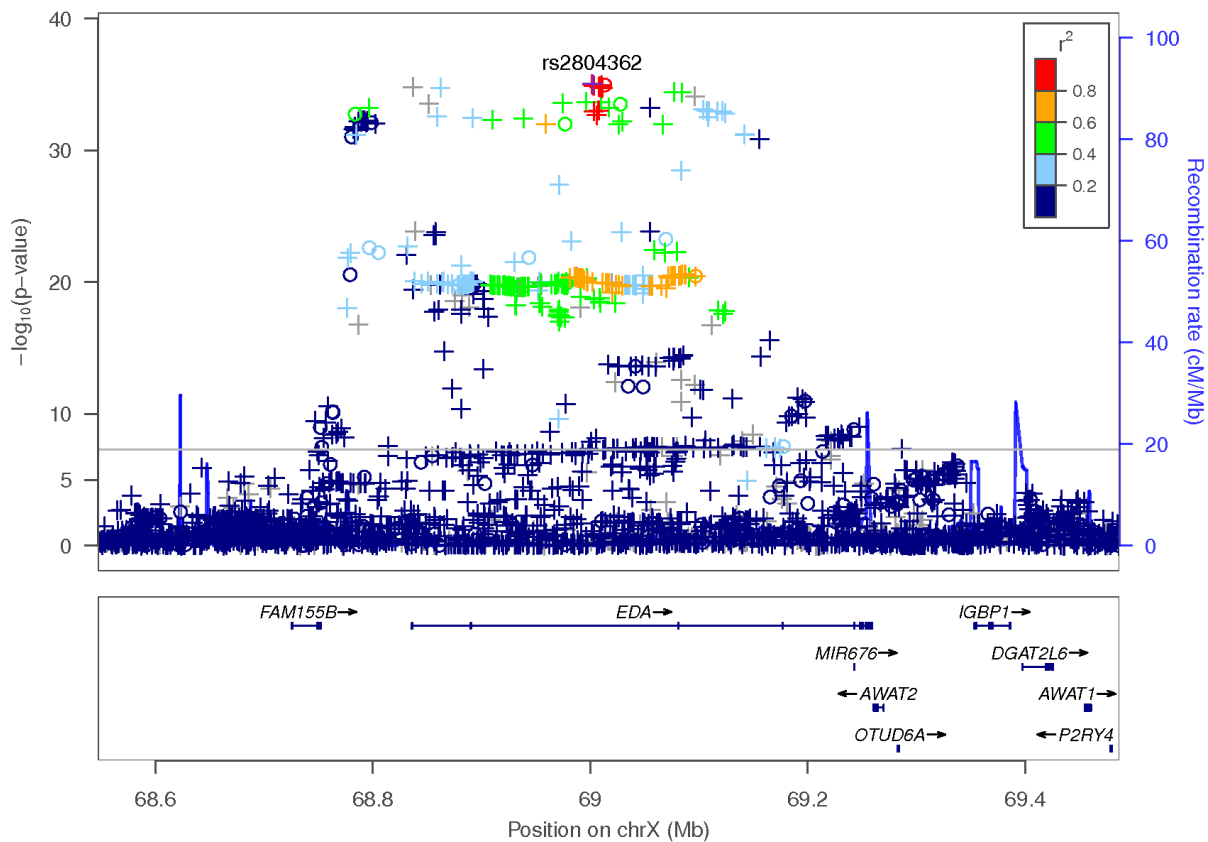
clinvar	16q24.3	rs78498439	6.9×10^{-14}	18630	0.823	rs1805008	MC1R	Increased analgesia from kappa-opioid receptor agonist, female-specific	NCBI curation
clinvar	16q24.3	rs78498439	6.9×10^{-14}	18630	0.823	rs1805008	MC1R	Tyrosinase-positive oculocutaneous albinism	SNOMED CT26336006
clinvar	16q24.3	rs78498439	6.9×10^{-14}	18630	0.823	rs1805008	MC1R	Tyrosinase-positive oculocutaneous albinism	Office of Rare Diseases4038

Regional Association Plots









locuszoom plot for 17:43849572:AT_A failed

