

# Analysis Summary: Nearsightedness

## Phenotype Description

Nearsightedness cases are defined as having said "Yes" or checking nearsightedness to one of the following questions.

- Your Medical History ("Have you ever been diagnosed by a doctor with nearsightedness (near objects are clear, far objects are blurry)?" Yes)
- Research Snippets ("Are you nearsighted (near objects are clear, far objects are blurry)?" Yes)
- Refractive Error ("What vision problems do you have? Please check all that apply." Nearsightedness (near objects are clear, far objects are blurry))
- Refractive Error ("Prior to your LASIK eye surgery, what vision problems did you have? Please check all that apply." Nearsightedness (near objects are clear, far objects are blurry))

Controls are defined as having said "No" or not checking nearsightedness to at least one of the questions above. Discordant answers are removed.

## 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]
nearsightedness	case	106086	53935	52151	15103	33066	29503	28414
	control	85757	47488	38269	12935	24222	21975	26625

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

Phenotype	Group	Total	v1/v2	v3	v4
nearsightedness	case	106086	8140	82499	15447
	control	85757	6072	66218	13467

## 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	z value	Pr(> z )	LRT	Pr(>Chi)
age	-0.00489	0.000287	-17.1	$3.1 \times 10^{-65}$	291.4	$2.5 \times 10^{-65}$
sexF	0.19536	0.009255	21.1	$6.5 \times 10^{-99}$	446.4	$4.3 \times 10^{-99}$
pc.0	-0.03665	0.004638	-7.9	$2.7 \times 10^{-15}$	62.8	$2.3 \times 10^{-15}$
pc.1	0.03886	0.004648	8.4	$6.2 \times 10^{-17}$	70.2	$5.3 \times 10^{-17}$
pc.2	-0.01167	0.004623	-2.5	0.012	6.4	0.012
pc.3	0.02378	0.004604	5.2	$2.4 \times 10^{-7}$	26.7	$2.4 \times 10^{-7}$
pc.4	0.03085	0.004601	6.7	$2.0 \times 10^{-11}$	44.7	$2.2 \times 10^{-11}$

## 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	-5807	13773260
failed to converge	-10335	13762925

## Genetic Association Tests

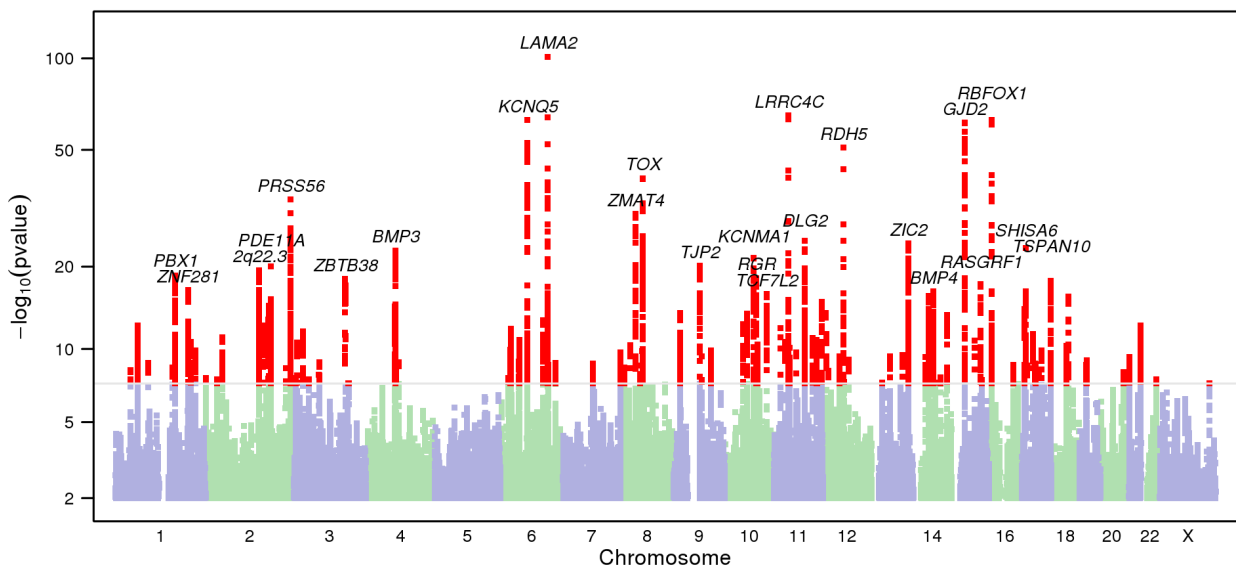
We performed logistic regression assuming an additive model for allelic effects, using the model:

$$\text{nearsightedness} \sim \text{age} + \text{sex} + \text{pc.0} + \text{pc.1} + \text{pc.2} + \text{pc.3} + \text{pc.4} + \text{genotype}$$

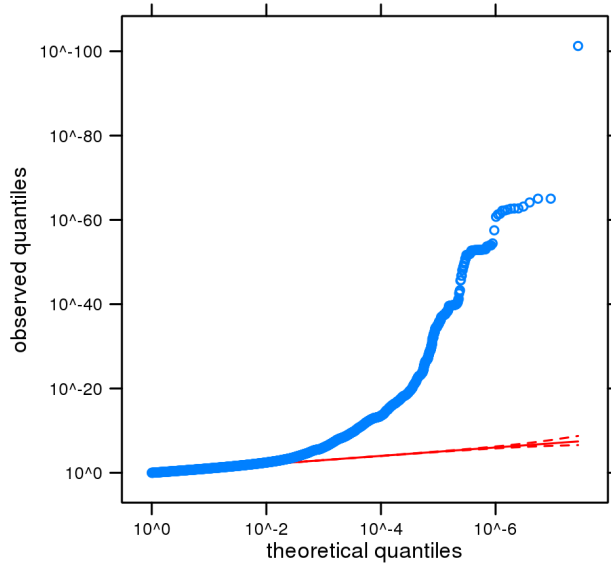
This genome-wide association analysis includes data from 106086 cases and 85757 controls of European ancestry, filtered to remove close relatives.

The results in this report have been adjusted for a genomic control inflation factor  $\lambda=1.230$ . The equivalent inflation factor for 1000 cases and 1000 controls  $\lambda_{1000}=1.002$ , and for 10000,  $\lambda_{10000}=1.024$ .

## Manhattan Plot



## Q-Q Plot of GWAS Results



## Index SNPs for Strongest Associations

cytoband	assay.name	scaffold	position	alleles	src	pvalue	OR	95% CI	gene.context
6q22.33	rs12193446	chr6	129820038	A/G	I	$5.4 \times 10^{-102}$	0.769	[0.751,0.788]	[LAMA2]
11p12	rs11606250	chr11	40149300	A/G	I	$9.2 \times 10^{-66}$	0.845	[0.829,0.862]	[LRRC4C]
6q13	rs7744813	chr6	73643289	A/C	I	$1.9 \times 10^{-63}$	0.883	[0.871,0.896]	[KCNQ5]
16p13.3	rs10500355	chr16	7459347	A/T	I	$2.0 \times 10^{-63}$	0.881	[0.868,0.894]	[RFX1]
15q14	rs524952	chr15	35005886	A/T	I	$3.5 \times 10^{-62}$	0.886	[0.874,0.899]	GOLGA8B---[]--GJD2
12q13.2	rs3138141	chr12	56115778	A/C	I	$1.5 \times 10^{-51}$	1.146	[1.126,1.167]	[RDH5]
8q12.1	rs10089517	chr8	60178721	A/C	I	$6.0 \times 10^{-41}$	1.107	[1.091,1.124]	TOX---[]---CA8
2q37.1	rs1550094	chr2	233385396	A/G	I	$9.2 \times 10^{-35}$	1.102	[1.085,1.119]	[PRSS56]
8p11.21	rs869422	chr8	40723970	A/G	I	$4.0 \times 10^{-31}$	0.903	[0.887,0.918]	[ZMAT4]
11q14.1	rs2155413	chr11	84634790	A/C	I	$2.3 \times 10^{-25}$	0.927	[0.914,0.941]	[DLG2]
13q32.3	rs9585327	chr13	100689354	A/G	I	$8.9 \times 10^{-25}$	0.928	[0.915,0.941]	ZIC2--[]--PCCA
17p12	rs2908972	chr17	11407259	A/T	I	$5.3 \times 10^{-24}$	0.927	[0.914,0.941]	[SHISA6]
4q21.21	rs5022942	chr4	81959966	A/G	I	$1.6 \times 10^{-23}$	0.917	[0.901,0.932]	[BMP3]
10q22.3	rs10824518	chr10	79063542	A/T	I	$3.3 \times 10^{-22}$	0.929	[0.916,0.943]	[KCNMA1]
9q21.11	rs11145488	chr9	71770939	A/G	I	$6.7 \times 10^{-21}$	0.920	[0.904,0.936]	FXN--[]--TJP2
2q31.2	rs17400325	chr2	178565913	C/T	I	$9.6 \times 10^{-21}$	0.844	[0.814,0.875]	[PDE11A]
2q22.3	rs61049169	chr2	146888708	A/G	I	$3.4 \times 10^{-20}$	1.070	[1.054,1.085]	[]
1q23.3	rs1556867	chr1	164213686	C/T	I	$2.7 \times 10^{-19}$	1.080	[1.062,1.098]	NUF2---[]---PBX1
10q23.1	rs10887265	chr10	86015573	C/G	I	$5.6 \times 10^{-19}$	0.931	[0.917,0.946]	[RGR]
3q23	rs9866391	chr3	141076084	C/T	I	$7.7 \times 10^{-19}$	1.069	[1.053,1.084]	[ZBTB38]
17q25.3	rs9747347	chr17	79606820	C/T	I	$1.6 \times 10^{-18}$	1.074	[1.057,1.092]	NPLOC4-[]-TSPAN10
15q25.1	rs13380104	chr15	79378821	C/T	I	$4.3 \times 10^{-18}$	1.066	[1.051,1.082]	[RASGRF1]
1q32.1	rs2808510	chr1	200336075	C/T	I	$3.1 \times 10^{-17}$	1.065	[1.049,1.081]	NR5A2---[]--ZNF281
14q22.2	rs2181346	chr14	54552428	A/T	I	$4.7 \times 10^{-17}$	1.079	[1.060,1.098]	BMP4---[]---CDKN3
10q25.2	rs56299331	chr10	114788436	C/T	I	$1.1 \times 10^{-16}$	1.078	[1.059,1.098]	[TCF7L2]
14q21.1	rs34217772	chr14	42273570	C/G	I	$2.0 \times 10^{-16}$	1.081	[1.061,1.102]	[LRFN5]
18q21.1	rs12965607	chr18	47391025	G/T	I	$2.4 \times 10^{-16}$	0.918	[0.899,0.937]	[MYO5B]
11q25	rs1793639	chr11	131931531	A/G	I	$9.7 \times 10^{-16}$	1.061	[1.046,1.076]	[NTM]
4q21.21	rs13129838	chr4	80508788	C/T	I	$2.6 \times 10^{-15}$	1.068	[1.051,1.086]	GK2---[]---ANTXR2
2q31.1	rs17428076	chr2	172851936	C/G	I	$3.2 \times 10^{-15}$	0.935	[0.919,0.951]	HAT1-[]--METAP1D
17p13.1	rs11658305	chr17	7429321	A/C	I	$7.9 \times 10^{-15}$	1.062	[1.046,1.078]	POLR2A--[]--TNFSF12-TNFSF13
9p22.2	rs10511652	chr9	18362865	A/G	I	$2.3 \times 10^{-14}$	1.058	[1.043,1.073]	SH3GL2---[]---ADAMTSL1
12p13.31	rs5442	chr12	6954864	A/G	I	$2.3 \times 10^{-14}$	0.895	[0.870,0.921]	[GNB3]
10q21.1	rs4948523	chr10	60339098	A/C	I	$2.8 \times 10^{-14}$	0.946	[0.933,0.960]	[BICC1]
14q32.12	rs34016308	chr14	92615741	D/I	I	$4.1 \times 10^{-14}$	0.929	[0.911,0.947]	[CPSF2]
6q22.1	rs1064583	chr6	116446576	A/G	I	$1.6 \times 10^{-13}$	1.056	[1.041,1.072]	[NT5DC1,COL10A1]
10q11.22	rs201140091	chr10	49408240	D/I	I	$3.7 \times 10^{-13}$	1.068	[1.049,1.087]	[FRMPD2]
21q22.3	rs73157695	chr21	47371947	A/G	I	$5.1 \times 10^{-13}$	0.944	[0.929,0.959]	PCBP3-[]--COL6A1
1p31.3	rs479445	chr1	61341632	A/T	I	$5.7 \times 10^{-13}$	0.946	[0.932,0.960]	C1orf87---[]---NFIA
2q24.1	rs297589	chr2	157358750	A/T	I	$5.8 \times 10^{-13}$	1.061	[1.044,1.078]	[GPD2]
11p15.1	rs1550870	chr11	18751041	C/T	I	$9.9 \times 10^{-13}$	0.950	[0.936,0.963]	[PTPN5]
6p22.1	rs2799081	chr6	28270584	C/T	I	$1.2 \times 10^{-12}$	0.943	[0.928,0.959]	PGBD1[]--ZSCAN31
3p24.2	rs826220	chr3	24268677	C/T	I	$2.3 \times 10^{-12}$	0.949	[0.936,0.963]	[THR3]
17q11.2	rs10512441	chr17	31239645	C/T	I	$3.8 \times 10^{-12}$	1.065	[1.046,1.084]	MYO1D--[]--TMEM98
12p13.31	rs7968679	chr12	9313304	A/G	I	$3.8 \times 10^{-12}$	1.057	[1.040,1.073]	[PZP]
2p21	rs2342406	chr2	45152748	C/T	I	$7.8 \times 10^{-12}$	1.065	[1.046,1.085]	CAMKMT---[]--SIX3
11q22.3	rs71041628	chr11	105665158	D/I	I	$9.7 \times 10^{-12}$	1.055	[1.039,1.072]	[GRIA4]
11q23.3	rs10892173	chr11	117672561	C/T	I	$1.4 \times 10^{-11}$	1.051	[1.036,1.066]	DSCAML1-[]--FXYD6-FXYD2
6p12.3	rs2207136	chr6	50809720	C/T	I	$1.5 \times 10^{-11}$	1.051	[1.036,1.066]	[TFAP2B]

## 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
rs12193446	TRUE	TRUE	TRUE	1.0000	0.026	0.64	0.0954	0.9946	0.9882	0.76	0.0947	v2v3v4
rs11606250	FALSE	FALSE	FALSE					0.9716	0.9313	0.088	0.8297	v2v3v4
rs7744813	FALSE	TRUE	TRUE	0.9991	0.89	0.40	0.4079	0.9913	0.9309	0.27	0.4071	v2v3v4
rs10500355	FALSE	FALSE	FALSE					0.9860	0.9681	0.095	0.6333	v2v3v4
rs524952	FALSE	FALSE	TRUE	1.0000	0.013	0.25	0.5239	0.9958	0.9643	0.060	0.5262	v2v3v4
rs3138141	FALSE	FALSE	FALSE					0.9861	0.9721	7.8×10 <sup>-5</sup>	0.7712	v3v4
rs10089517	FALSE	FALSE	TRUE	0.9936	0.059	0.063	0.6494	0.9874	0.9728	0.025	0.6452	v2v3v4
rs1550094	FALSE	TRUE	TRUE	0.9652	6.8×10 <sup>-10</sup>	1.9×10 <sup>-5</sup>	0.3067	0.9926	0.9326	0.22	0.3052	v2v3v4
rs869422	TRUE	TRUE	FALSE	0.9999	0.41	0.74	0.2157	0.9961	0.9777	0.85	0.2153	v2v3v4
rs2155413	TRUE	TRUE	TRUE	0.9998	0.34	0.064	0.5361	0.9960	0.9939	0.30	0.5319	v2v3v4
rs9585327	FALSE	FALSE	FALSE					0.9929	0.9864	0.22	0.5556	v2v3v4
rs2908972	FALSE	FALSE	TRUE	1.0000	0.26	0.59	0.6010	0.9815	0.9718	0.28	0.5981	v2v3v4
rs5022942	FALSE	FALSE	TRUE	1.0000	2.3×10 <sup>-5</sup>	0.41	0.7771	0.9983	0.9930	1.2×10 <sup>-5</sup>	0.7721	v2v3v4
rs10824518	FALSE	FALSE	FALSE					0.9916	0.9903	0.59	0.6461	v2v3v4
rs11145488	FALSE	FALSE	FALSE					0.9966	0.9930	0.24	0.7902	v2v3v4
rs17400325	FALSE	TRUE	TRUE	0.9998	0.24	0.067	0.9583	0.9834	0.9249	1.7×10 <sup>-10</sup>	0.9565	v2v3v4
rs61049169	FALSE	FALSE	FALSE					0.9902	0.9787	0.63	0.5593	v2v3v4
rs1556867	TRUE	TRUE	TRUE	0.9997	0.58	0.48	0.2330	0.9996	0.9989	0.43	0.2328	v2v3v4
rs10887265	FALSE	FALSE	FALSE					0.9277	0.9238	0.78	0.6763	v2v3v4
rs9866391	FALSE	FALSE	FALSE					0.9811	0.9383	0.020	0.6175	v2v3v4
rs9747347	FALSE	FALSE	FALSE					0.8344	0.7649	0.070	0.3738	v2v3v4
rs13380104	FALSE	FALSE	FALSE					0.9953	0.9835	0.063	0.4036	v2v3v4
rs2808510	FALSE	FALSE	FALSE					0.9926	0.9871	0.73	0.3822	v2v3v4
rs2181346	FALSE	FALSE	FALSE					0.7408	0.6697	0.39	0.3119	v2v3v4
rs56299331	FALSE	FALSE	FALSE					0.9875	0.9785	0.27	0.2025	v2v3v4
rs34217772	FALSE	FALSE	FALSE					0.9984	0.9953	0.014	0.1745	v2v3v4
rs12965607	FALSE	FALSE	FALSE					0.9496	0.9022	0.19	0.8539	v2v3v4
rs1793639	FALSE	FALSE	FALSE					0.9830	0.9621	0.27	0.5773	v2v3v4
rs13129838	FALSE	FALSE	FALSE					0.9229	0.9074	0.088	0.2826	v2v3v4
rs17428076	FALSE	FALSE	TRUE	1.0000	0.84	0.16	0.2363	0.9942	0.9910	0.48	0.2348	v2v3v4
rs11658305	FALSE	FALSE	FALSE					0.9165	0.8846	0.025	0.6085	v2v3v4
rs10511652	TRUE	TRUE	FALSE	0.9713	5.8×10 <sup>-13</sup>	9.8×10 <sup>-50</sup>	0.5994	0.9971	0.9825	0.17	0.5880	v2v3v4
rs5442	TRUE	TRUE	TRUE	0.9991	0.56	0.59	0.9336	0.9952	0.9929	0.51	0.9335	v2v3v4
rs4948523	FALSE	FALSE	FALSE					0.9943	0.9930	0.0071	0.5275	v2v3v4
rs34016308	FALSE	FALSE	FALSE					0.9841	0.9783	0.40	0.1659	v2v3v4
rs1064583	TRUE	TRUE	TRUE	0.9987	0.94	0.76	0.3889	1.0000	0.9998	0.95	0.3882	v2v3v4
rs201140091	FALSE	FALSE	FALSE					0.8150	0.8005	0.033	0.2682	v2v3v4
rs73157695	FALSE	FALSE	FALSE					0.9809	0.9711	0.23	0.7020	v2v3v4
rs479445	FALSE	FALSE	FALSE					0.9975	0.9875	0.056	0.6745	v2v3v4
rs297589	FALSE	FALSE	FALSE					0.8998	0.8746	0.15	0.3093	v2v3v4
rs1550870	TRUE	TRUE	TRUE	0.9983	0.022	0.87	0.4609	0.9989	0.9951	0.35	0.4587	v2v3v4
rs2799081	FALSE	FALSE	FALSE					0.9982	0.9974	0.031	0.7354	v2v3v4
rs826220	FALSE	FALSE	FALSE					0.9975	0.9950	0.17	0.5968	v2v3v4
rs10512441	FALSE	TRUE	TRUE	0.9999	0.017	0.46	0.2012	0.9855	0.8263	0.075	0.2023	v2v3v4
rs7968679	FALSE	FALSE	FALSE					0.9814	0.9709	0.0021	0.3038	v2v3v4
rs2342406	FALSE	TRUE	FALSE	0.9995	0.91	0.27	0.8083	0.9851	0.9171	0.63	0.8091	v2v3v4
rs71041628	FALSE	FALSE	FALSE					0.8855	0.8746	0.0013	0.3810	v2v3v4
rs10892173	FALSE	FALSE	FALSE					0.9946	0.9857	0.32	0.4030	v2v3v4
rs2207136	FALSE	FALSE	FALSE					0.9824	0.9767	0.0054	0.5511	v2v3v4
rs7162310	FALSE	FALSE	FALSE					0.9990	0.9953	0.19	0.2212	v2v3v4

## SNP Statistics in the GWAS Sample

assay.name	AA.0	AB.0	BB.0	im.num.0	dose.b.0	AA.1	AB.1	BB.1	im.num.1	dose.b.1
rs12193446	68256	16488	1009	85757	0.1076	88839	16416	829	106086	0.0850
rs11606250				85757	0.8425				106086	0.8201
rs7744813	26276	39453	14100	85757	0.4231	36189	46636	15314	106086	0.3936
rs10500355				85757	0.6508				106086	0.6217
rs524952	2824	6710	3933	85757	0.5427	3760	7651	4036	106086	0.5136
rs3138141				79302	0.7588				97352	0.7825
rs10089517	1755	6231	5397	85757	0.6330	1757	6913	6679	106086	0.6555
rs1550094	37631	32437	6499	85757	0.2939	43744	41278	9228	106086	0.3140
rs869422	43456	25388	3444	85757	0.2226	57188	29475	3971	106086	0.2060
rs2155413	17704	42366	25665	85757	0.5430	23526	52969	29567	106086	0.5251
rs9585327				85757	0.5657				106086	0.5477
rs2908972	2029	6355	5083	85757	0.6097	2608	7428	5411	106086	0.5920
rs5022942	652	4533	8282	85757	0.7848	828	5347	9272	106086	0.7690
rs10824518				85757	0.6519				106086	0.6356
rs11145488				85757	0.7963				106086	0.7831
rs17400325	118	5802	73751	85757	0.9613	204	8276	89450	106086	0.9545
rs61049169				85757	0.5470				106086	0.5643
rs1556867	51303	30044	4390	85757	0.2263	61329	38716	6011	106086	0.2391
rs10887265				85757	0.6849				106086	0.6703
rs9866391				85757	0.6065				106086	0.6220
rs9747347				85757	0.3684				106086	0.3820
rs13380104				85757	0.3983				106086	0.4130
rs2808510				85757	0.3770				106086	0.3912
rs2181346				85757	0.3044				106086	0.3171
rs56299331				85757	0.1946				106086	0.2061
rs34217772				85757	0.1718				106086	0.1825
rs12965607				85757	0.8591				106086	0.8491
rs1793639				85757	0.5703				106086	0.5849
rs13129838				85757	0.2758				106086	0.2878
rs17428076	7809	4888	770	85757	0.2409	9148	5504	795	106086	0.2289
rs11658305				85757	0.6020				106086	0.6151

rs10511652	11281	34437	24438	85757	0.5820	13365	42754	32020	106086	0.5951
rs5442	365	10123	75187	85757	0.9363	502	13843	91640	106086	0.9295
rs4948523				85757	0.5364				106086	0.5221
rs34016308				85757	0.1702				106086	0.1607
rs1064583	32689	40395	12565	85757	0.3827	38774	50622	16569	106086	0.3953
rs201140091				85757	0.2638				106086	0.2738
rs73157695				85757	0.7082				106086	0.6970
rs479445				85757	0.6833				106086	0.6709
rs297589				85757	0.3034				106086	0.3145
rs1550870	24181	42482	18935	85757	0.4694	31415	52323	22166	106086	0.4563
rs2799081				85757	0.7429				106086	0.7316
rs826220				85757	0.6051				106086	0.5920
rs10512441	51846	24707	3124	85757	0.1945	61877	31944	4108	106086	0.2051
rs7968679				85757	0.2995				106086	0.3102
rs2342406	2633	20862	42691	85757	0.8037	2869	25185	54405	106086	0.8135
rs71041628				85757	0.3757				106086	0.3867
rs10892173				85757	0.3990				106086	0.4103
rs2207136				85757	0.5407				106086	0.5534
rs7162310				85757	0.2240				106086	0.2143

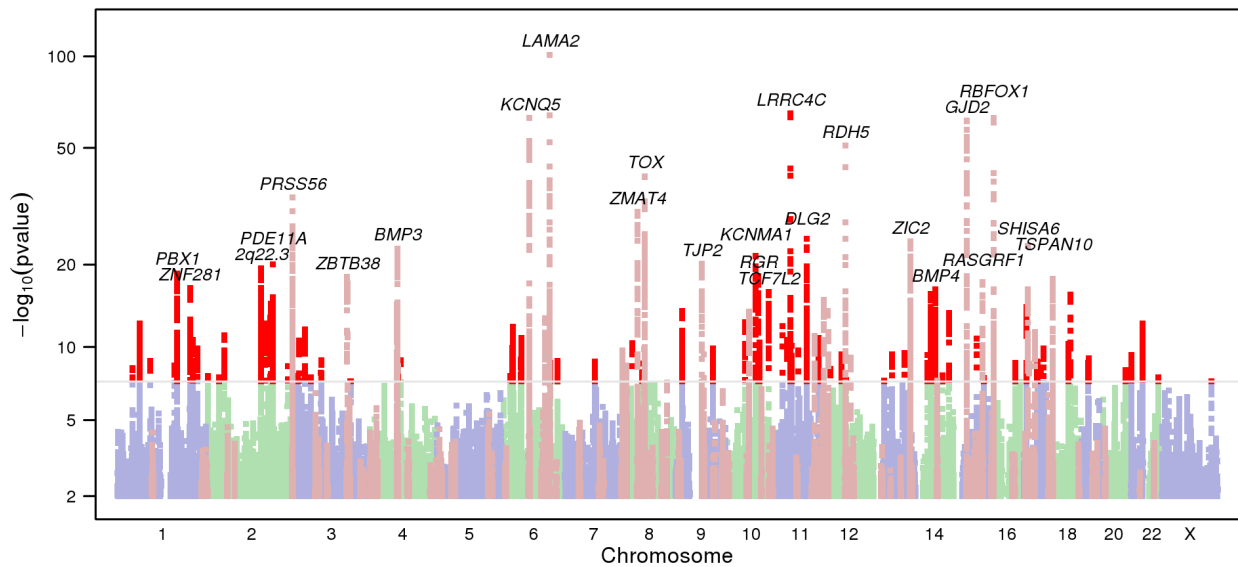
## Annotations from NHGRI GWAS Catalog

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
6q22.33	129820038	rs12193446	$5.4 \times 10^{-102}$	0	1.000	rs12193446	$1.0 \times 10^{-8}$	24144296	Axial length	LAMA2, ARHGAP18
6q22.33	129820038	rs12193446	$5.4 \times 10^{-102}$	14591	0.551	rs12205363	$2.0 \times 10^{-12}$	23396134	Refractive error	LAMA2
6q13	73643289	rs7744813	$1.9 \times 10^{-63}$	0	1.000	rs7744813	$4.0 \times 10^{-9}$	23396134	Refractive error	KCNQ5
16p13.3	7459347	rs10500355	$2.0 \times 10^{-63}$	0	1.000	rs10500355	$4.0 \times 10^{-9}$	23474815	Refractive error	RBFOX1
16p13.3	7459347	rs10500355	$2.0 \times 10^{-63}$	336	1.000	rs17648524	$6.0 \times 10^{-10}$	23396134	Refractive error	RBFOX1
15q14	35005886	rs524952	$3.5 \times 10^{-62}$	-16260	0.591	rs11073058	$4.0 \times 10^{-11}$	24144296	Axial length	GJD2, ACTC1
15q14	35005886	rs524952	$3.5 \times 10^{-62}$	0	1.000	rs524952	$1.0 \times 10^{-15}$	23396134	Refractive error	GJD2
15q14	35005886	rs524952	$3.5 \times 10^{-62}$	187	1.000	rs634990	$2.0 \times 10^{-14}$	20835239	Refractive error	GJD2,ACTC1,GOLGA8B
8q12.1	60178721	rs10089517	$6.0 \times 10^{-41}$	0	1.000	rs10089517	$2.0 \times 10^{-6}$	22589738	Visceral adipose tissue/subcutaneous adipose tissue ratio	TOX
8q12.1	60178721	rs10089517	$6.0 \times 10^{-41}$	365	0.576	rs7837791	$4.0 \times 10^{-12}$	23396134	Refractive error	CHD7, TOX
8p11.21	40723970	rs869422	$4.0 \times 10^{-31}$	2424	1.000	rs7829127	$4.0 \times 10^{-10}$	23396134	Refractive error	ZMAT4
17p12	11407259	rs2908972	$5.3 \times 10^{-24}$	642	0.621	rs2969180	$7.0 \times 10^{-11}$	23396134	Refractive error	SHISA6
9q21.11	71770939	rs11145488	$6.7 \times 10^{-21}$	-4346	1.000	rs11145465	$7.0 \times 10^{-9}$	23396134	Refractive error	TJP2
3q23	141076084	rs9866391	$7.7 \times 10^{-19}$	2104	0.685	rs9857275	$2.0 \times 10^{-6}$	22210626	Prion diseases	ZBTB38, RASA2
3q23	141076084	rs9866391	$7.7 \times 10^{-19}$	18125	0.547	rs6440003	$2.0 \times 10^{-10}$	21998595	Height	ZBTB38
3q23	141076084	rs9866391	$7.7 \times 10^{-19}$	18125	0.547	rs6440003	$2.0 \times 10^{-24}$	18391952	Height	ZBTB38
3q23	141076084	rs9866391	$7.7 \times 10^{-19}$	26749	0.542	rs6763931	$2.0 \times 10^{-8}$	21743467	Prostate cancer	ZBTB38
3q23	141076084	rs9866391	$7.7 \times 10^{-19}$	26749	0.542	rs6763931	$3.0 \times 10^{-12}$	19343178	Height	ZBTB38
3q23	141076084	rs9866391	$7.7 \times 10^{-19}$	26749	0.542	rs6763931	$1.0 \times 10^{-27}$	18391951	Height	ZBTB38
3q23	141076084	rs9866391	$7.7 \times 10^{-19}$	29486	0.542	rs724016	$3.0 \times 10^{-86}$	20881960	Height	ZBTB38
3q23	141076084	rs9866391	$7.7 \times 10^{-19}$	29486	0.542	rs724016	$8.0 \times 10^{-22}$	18391950	Height	ZBTB38
3q23	141076084	rs9866391	$7.7 \times 10^{-19}$	29486	0.542	rs724016	$1.0 \times 10^{-6}$	18193045	Height	ZBTB38
3q23	141076084	rs9866391	$7.7 \times 10^{-19}$	57366	0.509	rs1991431	$4.0 \times 10^{-47}$	23563607	Height	ZBTB38
15q25.1	79378821	rs13380104	$4.3 \times 10^{-18}$	-5946	0.899	rs4778879	$4.0 \times 10^{-11}$	23396134	Refractive error	RASGRF1
15q25.1	79378821	rs13380104	$4.3 \times 10^{-18}$	85026	0.660	rs1474256	$6.0 \times 10^{-6}$	23897914	Bronchopulmonary dysplasia	RASGRF1, LOC729911
10q25.2	114788436	rs56299331	$1.1 \times 10^{-16}$	-34348	0.543	rs7901695	$1.0 \times 10^{-6}$	21347282	Coronary heart disease	TCF7L2
10q25.2	114788436	rs56299331	$1.1 \times 10^{-16}$	-34348	0.543	rs7901695	$1.0 \times 10^{-48}$	17463249	Type 2 diabetes	TCF7L2
10q25.2	114788436	rs56299331	$1.1 \times 10^{-16}$	-32395	0.545	rs4506565	$1.0 \times 10^{-8}$	20081858	Fasting glucose-related traits	TCF7L2
10q25.2	114788436	rs56299331	$1.1 \times 10^{-16}$	-32395	0.545	rs4506565	$5.0 \times 10^{-12}$	17554300	Type 2 diabetes	TCF7L2
10q25.2	114788436	rs56299331	$1.1 \times 10^{-16}$	-30087	0.593	rs7903146	$2.0 \times 10^{-15}$	23945395	Type 2 diabetes	TCF7L2
10q25.2	114788436	rs56299331	$1.1 \times 10^{-16}$	-30087	0.593	rs7903146	$9.0 \times 10^{-75}$	23300278	Type 2 diabetes	TCF7L2
10q25.2	114788436	rs56299331	$1.1 \times 10^{-16}$	-30087	0.593	rs7903146	$1.0 \times 10^{-35}$	23209189	Type 2 diabetes	TCF7L2
10q25.2	114788436	rs56299331	$1.1 \times 10^{-16}$	-30087	0.593	rs7903146	$4.0 \times 10^{-21}$	22693455	Type 2 diabetes	TCF7L2
10q25.2	114788436	rs56299331	$1.1 \times 10^{-16}$	-30087	0.593	rs7903146	$3.0 \times 10^{-6}$	22581228	Fasting insulin-related traits (interaction with BMI)	TCF7L2
10q25.2	114788436	rs56299331	$1.1 \times 10^{-16}$	-30087	0.593	rs7903146	$2.0 \times 10^{-14}$	22581228	Fasting glucose-related traits (interaction with BMI)	TCF7L2
10q25.2	114788436	rs56299331	$1.1 \times 10^{-16}$	-30087	0.593	rs7903146	$2.0 \times 10^{-15}$	22101970	Type 2 diabetes	TCF7L2
10q25.2	114788436	rs56299331	$1.1 \times 10^{-16}$	-30087	0.593	rs7903146	$2.0 \times 10^{-20}$	21873549	Proinsulin levels	TCF7L2
10q25.2	114788436	rs56299331	$1.1 \times 10^{-16}$	-30087	0.593	rs7903146	$1.0 \times 10^{-7}$	20849430	Glycated hemoglobin levels	TCF7L2
10q25.2	114788436	rs56299331	$1.1 \times 10^{-16}$	-30087	0.593	rs7903146	$7.0 \times 10^{-7}$	20694148	Metabolic syndrome	TCF7L2
10q25.2	114788436	rs56299331	$1.1 \times 10^{-16}$	-30087	0.593	rs7903146	$2.0 \times 10^{-51}$	20581827	Type 2 diabetes	TCF7L2
10q25.2	114788436	rs56299331	$1.1 \times 10^{-16}$	-30087	0.593	rs7903146	$1.0 \times 10^{-30}$	19734900	Type 2 diabetes and other traits	TCF7L2
10q25.2	114788436	rs56299331	$1.1 \times 10^{-16}$	-30087	0.593	rs7903146	$8.0 \times 10^{-12}$	19401414	Type 2 diabetes	TCF7L2
10q25.2	114788436	rs56299331	$1.1 \times 10^{-16}$	-30087	0.593	rs7903146	$9.0 \times 10^{-30}$	19056611	Type 2 diabetes	TCF7L2
10q25.2	114788436	rs56299331	$1.1 \times 10^{-16}$	-30087	0.593	rs7903146	$3.0 \times 10^{-23}$	18372903	Type 2 diabetes	TCF7L2
10q25.2	114788436	rs56299331	$1.1 \times 10^{-16}$	-30087	0.593	rs7903146	$5.0 \times 10^{-8}$	17668382	Type 2 diabetes	TCF7L2
10q25.2	114788436	rs56299331	$1.1 \times 10^{-16}$	-30087	0.593	rs7903146	$1.0 \times 10^{-8}$	17463248	Type 2 diabetes	TCF7L2
10q25.2	114788436	rs56299331	$1.1 \times 10^{-16}$	-30087	0.593	rs7903146	$2.0 \times 10^{-31}$	17463246	Type 2 diabetes	TCF7L2
10q25.2	114788436	rs56299331	$1.1 \times 10^{-16}$	-30087	0.593	rs7903146	$2.0 \times 10^{-10}$	17460697	Type 2 diabetes	TCF7L2
10q25.2	114788436	rs56299331	$1.1 \times 10^{-16}$	-30087	0.593	rs7903146	$2.0 \times 10^{-34}$	17293876	Type 2 diabetes	TCF7L2
10q25.2	114788436	rs56299331	$1.1 \times 10^{-16}$	379	0.687	rs12243326	$1.0 \times 10^{-7}$	20081857	Two-hour glucose challenge	TCF7L2
4q21.21	80508788	rs13129838	$2.6 \times 10^{-15}$	21883	0.618	rs9307551	$1.0 \times 10^{-8}$	23396134	Refractive error	LOC100506035
17p13.1	7429321	rs11658305	$7.9 \times 10^{-15}$	-11658	0.860	rs6761	$3.0 \times 10^{-7}$	18464913	Protein quantitative trait loci	SHBG

10q21.1	60339098	rs4948523	$2.8 \times 10^{-14}$	-73694	0.927	rs7084402	$2.0 \times 10^{-13}$	23396134	Refractive error	BICC1
10q21.1	60339098	rs4948523	$2.8 \times 10^{-14}$	-20201	0.948	rs1658442	$3.0 \times 10^{-6}$	23322567	Corneal astigmatism	
6q22.1	116446576	rs1064583	$1.6 \times 10^{-13}$	-2841	0.989	rs3812111	$2.0 \times 10^{-8}$	23455636	Age-related macular degeneration	COL10A1
6p22.1	28270584	rs2799081	$1.2 \times 10^{-12}$	51712	0.791	rs6903823	$2.0 \times 10^{-10}$	21946350	Pulmonary function	ZKSCAN3, ZNF323
17q11.2	31239645	rs10512441	$3.8 \times 10^{-12}$	-161373	0.924	rs17183295	$1.0 \times 10^{-10}$	23396134	Refractive error	MYO1D

## 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,  $r^2 > 0.8$ ).

region	position	our.name	our.pval	dist	rsqr	assay.name	pvalue	pubmed.id	trait	genes
1p21.3	99524047	rs10747502	0.37	0	1.000	rs10747502	$1.0 \times 10^{-9}$	23049088	Myopia (pathological)	LOC100129620
1q42.2	230922951	rs12038826	0.75	0	1.000	rs12038826	$7.0 \times 10^{-6}$	23049088	Myopia (pathological)	CAPN9
1q44	249168436	rs12032643	0.35	0	1.000	rs12032643	$1.0 \times 10^{-9}$	23049088	Myopia (pathological)	Intergenic
2p16.2	54718181	rs4557020	0.070	0	1.000	rs4557020	$3.0 \times 10^{-7}$	22685421	Myopia (pathological)	SPTBN1
2p13.1	74939176	rs1137	0.029	0	1.000	rs1137	$4.0 \times 10^{-6}$	22685421	Myopia (pathological)	SEMA4F
3p14.2	59928851	rs931317	0.96	0	1.000	rs931317	$3.0 \times 10^{-6}$	23049088	Myopia (pathological)	FHIT
3p12.1	86255672	rs7428796	0.018	0	1.000	rs7428796	$2.0 \times 10^{-18}$	23049088	Myopia (pathological)	Intergenic
3q23	140247177	rs4683505	0.60	0	1.000	rs4683505	$4.0 \times 10^{-7}$	23049088	Myopia (pathological)	CLSTN2
3q24	143902027	rs4839680	0.68	0	1.000	rs4839680	$1.0 \times 10^{-15}$	23049088	Myopia (pathological)	Intergenic
3q26.32	177766464	rs7634528	0.65	0	1.000	rs7634528	$1.0 \times 10^{-6}$	23049088	Myopia (pathological)	Intergenic
3q27.3	186466252	rs1656966	0.99	0	1.000	rs1656966	$4.0 \times 10^{-21}$	23049088	Myopia (pathological)	Intergenic
3q27.3	187687890	rs16862782	0.64	0	1.000	rs16862782	$5.0 \times 10^{-7}$	23049088	Myopia (pathological)	Intergenic
4p16.1	10726853	rs16872571	0.92	0	1.000	rs16872571	$2.0 \times 10^{-10}$	23049088	Myopia (pathological)	Intergenic
4p15.2	24578097	rs6841898	0.19	0	1.000	rs6841898	$3.0 \times 10^{-26}$	23049088	Myopia (pathological)	DHX15
4q25	112611750	rs10034228	0.24	0	1.000	rs10034228	$8.0 \times 10^{-13}$	21505071	Myopia (pathological)	MYP11
4q25	112702635	rs1585471	0.22	0	1.000	rs1585471	$2.0 \times 10^{-6}$	21505071	Myopia (pathological)	MYP11
4q33	170880883	rs11723530	0.40	0	1.000	rs11723530	$4.0 \times 10^{-6}$	23049088	Myopia (pathological)	Intergenic
4q35.2	189120911	rs6857559	0.48	0	1.000	rs6857559	$3.0 \times 10^{-21}$	23049088	Myopia (pathological)	Intergenic
5p15.31	7368845	rs13172324	0.87	0	1.000	rs13172324	$3.0 \times 10^{-10}$	23049088	Myopia (pathological)	Intergenic
5p15.2	11169945	rs6885224	0.41	0	1.000	rs6885224	$8.0 \times 10^{-6}$	21095009	Myopia (pathological)	CTNND2
5p13.1	39979172	rs10053502	0.096	0	1.000	rs10053502	$1.0 \times 10^{-16}$	23049088	Myopia (pathological)	Intergenic
5q11.1	50026465	rs282544	0.0083	0	1.000	rs282544	$4.0 \times 10^{-6}$	22685421	Myopia (pathological)	PARP8
5q31.3	141189168	rs248471	0.62	0	1.000	rs248471	$2.0 \times 10^{-6}$	23406873	Myopia (pathological)	PCDH1
5q33.1	150473674	rs999556	0.32	0	1.000	rs999556	$1.0 \times 10^{-15}$	23049088	Myopia (pathological)	Intergenic
6p24.3	7743663	rs9505270	0.69	0	1.000	rs9505270	$4.0 \times 10^{-7}$	23049088	Myopia (pathological)	BMP6
6q21	108017671	rs12525668	0.52	0	1.000	rs12525668	$8.0 \times 10^{-11}$	23049088	Myopia (pathological)	Intergenic
6q22.1	117605991	rs13201929	0.23	0	1.000	rs13201929	$3.0 \times 10^{-6}$	23049088	Myopia (pathological)	Intergenic
6q24.3	147971574	rs1302019	0.78	0	1.000	rs1302019	$2.0 \times 10^{-23}$	23049088	Myopia (pathological)	Intergenic
7p14.1	38004406	rs1668357	0.46	0	1.000	rs1668357	$4.0 \times 10^{-8}$	23049088	Myopia (pathological)	Intergenic
7q21.3	97422926	rs1229542	0.39	0	1.000	rs1229542	$2.0 \times 10^{-6}$	23049088	Myopia (pathological)	Intergenic
7q22.3	104758250	rs6968355	0.047	0	1.000	rs6968355	$2.0 \times 10^{-7}$	23049088	Myopia (pathological)	SRPK2
7q36.3	157387441	rs10274279	0.53	0	1.000	rs10274279	$4.0 \times 10^{-11}$	23049088	Myopia (pathological)	PTPRN2
7q36.3	158846929	rs2730260	0.0030	0	1.000	rs2730260	$9.0 \times 10^{-14}$	23406873	Myopia (pathological)	VIPR2
8p23.1	8890098	rs1045529	$6.8 \times 10^{-6}$	0	1.000	rs1045529	$3.0 \times 10^{-6}$	23049088	Myopia (pathological)	Intergenic
8p23.1	8990577	rs189798	0.089	0	1.000	rs189798	$6.0 \times 10^{-7}$	23049088	Myopia (pathological)	PPP1R3B, MYP10, MIR4660, MIR124-1, MSRA
8p23.1	9814411	rs656319	0.0043	0	1.000	rs656319	$3.0 \times 10^{-6}$	23049088	Myopia (pathological)	Intergenic
8p23.1	11612698	rs804280	0.045	0	1.000	rs804280	$4.0 \times 10^{-6}$	23049088	Myopia (pathological)	GATA4
8q12.1	56599548	rs4737395	0.80	0	1.000	rs4737395	$5.0 \times 10^{-9}$	23049088	Myopia (pathological)	Intergenic
8q12.1	60961821	rs569688	0.083	0	1.000	rs569688	$4.0 \times 10^{-7}$	23049088	Myopia (pathological)	Intergenic
8q13.1	66822030	rs6472235	0.017	0	1.000	rs6472235	$1.0 \times 10^{-7}$	23049088	Myopia (pathological)	Intergenic
8q21.13	82151747	rs12216812	0.76	0	1.000	rs12216812	$3.0 \times 10^{-9}$	23049088	Myopia (pathological)	Intergenic



6p22.1	28270584	rs2799081	$1.2 \times 10^{-12}$	23966	0.521	rs853684	ZSCAN31	K205R
6p22.1	28270584	rs2799081	$1.2 \times 10^{-12}$	87736	0.604	rs1361385	ZSCAN12	C583R
6p22.1	28270584	rs2799081	$1.2 \times 10^{-12}$	87751	0.604	rs1416918	ZSCAN12	R578G
12p13.31	9313304	rs7968679	$3.8 \times 10^{-12}$	-81036	0.692	rs669	A2M	I1000V
12p13.31	9313304	rs7968679	$3.8 \times 10^{-12}$	-10008	0.981	rs10842971	PZP	I1443N

## Nearby Expression QTLs

region	position	our.name	our.pval	dist	rsqr	assay.name	eqtl.dist	eqtl.gene	eqtl.pval	eqtl.rsqr	tissue	pubmed.id
12q13.2	56115778	rs3138141	$1.5 \times 10^{-51}$	-193	0.975	rs3138142	728519	MIP	$7.2 \times 10^{-5}$	0.055	B-Cell	22446964
12q13.2	56115778	rs3138141	$1.5 \times 10^{-51}$	-193	0.975	rs3138142	2823	RDH5	$2.7 \times 10^{-42}$	0.485	Monocyte	22446964
12q13.2	56115778	rs3138141	$1.5 \times 10^{-51}$	-193	0.975	rs3138142	2823	RDH5	$5.2 \times 10^{-26}$	0.329	B-Cell	22446964
2q37.1	233385396	rs1550094	$9.2 \times 10^{-35}$	-9612	0.705	rs2117770	359210	ILMN_18124	0.00034	0.162	Lymphoblastoid	19644074
13q32.3	100689354	rs9585327	$8.9 \times 10^{-25}$	2013	0.652	rs8000973	49970	PCCA	$2.0 \times 10^{-8}$	0.024	Monocyte	20502693
3q23	141076084	rs9866391	$7.7 \times 10^{-19}$	17201	0.612	rs7624084	368312	RNF7	0.0011	0.038	B-Cell	22446964
17p13.1	7429321	rs11658305	$7.9 \times 10^{-15}$	-66233	0.624	rs9217	-2403	CHRNA1	$2.4 \times 10^{-22}$	0.287	Monocyte	22446964
17p13.1	7429321	rs11658305	$7.9 \times 10^{-15}$	-23187	0.662	rs11658168	45202	CHRNA1	$3.1 \times 10^{-66}$	0.184	Monocyte	20502693
10q21.1	60339098	rs4948523	$2.8 \times 10^{-14}$	-67274	0.588	rs7069916	318910	HS.100261	$4.4 \times 10^{-5}$	0.058	Monocyte	22446964
6q22.1	116446576	rs1064583	$1.6 \times 10^{-13}$	-23277	0.611	rs509002	143014	NT5DC1	0.0011	0.037	Monocyte	22446964
6q22.1	116446576	rs1064583	$1.6 \times 10^{-13}$	-7356	0.577	rs515745	17221	NT5DC1	$7.4 \times 10^{-15}$	0.151	Lymphoblastoid	24037378
6q22.1	116446576	rs1064583	$1.6 \times 10^{-13}$	100348	0.540	rs1204804	235609	FAM26F	$1.3 \times 10^{-7}$	0.021	Monocyte	20502693
6p22.1	28270584	rs2799081	$1.2 \times 10^{-12}$	-83483	0.510	rs1736891	213941	ZSCAN23	0.00085	0.039	B-Cell	22446964
6p22.1	28270584	rs2799081	$1.2 \times 10^{-12}$	29752	0.623	rs17312661	-460639	HIST1H3I	0.00018	0.049	B-Cell	22446964
6p22.1	28270584	rs2799081	$1.2 \times 10^{-12}$	54724	0.723	rs6922111	0	ZKSCAN3	$2.1 \times 10^{-8}$	0.024	Monocyte	20502693
6p22.1	28270584	rs2799081	$1.2 \times 10^{-12}$	119646	0.590	rs16894095	590925	HIST1H4K	$5.8 \times 10^{-6}$	0.016	Monocyte	20502693
6p22.1	28270584	rs2799081	$1.2 \times 10^{-12}$	256071	0.558	rs414745	-429050	ZSCAN16	0.00030	0.046	Monocyte	22446964
6p22.1	28270584	rs2799081	$1.2 \times 10^{-12}$	263362	0.558	rs418092	-728240	HIST1H2AK	0.00090	0.039	B-Cell	22446964
6p22.1	28270584	rs2799081	$1.2 \times 10^{-12}$	272680	0.643	rs17336532	-273277	PGBD1	$4.4 \times 10^{-5}$	0.058	B-Cell	22446964
6p22.1	28270584	rs2799081	$1.2 \times 10^{-12}$	276368	0.722	rs16901848	-2261548	HIST1H4H	0.00031	0.045	Monocyte	22446964
17q11.2	31239645	rs10512441	$3.8 \times 10^{-12}$	-203152	0.915	rs9912761	216865	MYO1D	$1.5 \times 10^{-6}$	0.274	T-cell	19644074
17q11.2	31239645	rs10512441	$3.8 \times 10^{-12}$	-52429	0.932	rs17781142	0	MYO1D	$4.2 \times 10^{-74}$	0.204	Monocyte	20502693
12p13.31	9313304	rs7968679	$3.8 \times 10^{-12}$	-199751	0.685	rs1805664	-14101	C12orf33	$3.4 \times 10^{-5}$		Liver	18462017
12p13.31	9313304	rs7968679	$3.8 \times 10^{-12}$	-94698	0.686	rs10771309	124548	PHC1	$4.9 \times 10^{-11}$	0.031	Monocyte	20502693

## Nearby Clinical Variants

source	region	our.name	our.pval	dist	rsqr	assay.name	gene	phenotype	accession
clinvar	10q25.2	rs56299331	$1.1 \times 10^{-16}$	-30087	0.593	rs7903146	TCF7L2	Diabetes mellitus type 2	SNOMED CT44054006
clinvar	10q25.2	rs56299331	$1.1 \times 10^{-16}$	20466	0.665	rs12255372	TCF7L2	Diabetes mellitus type 2	SNOMED CT44054006
clinvar	12p13.31	rs7968679	$3.8 \times 10^{-12}$	-81036	0.692	rs669	A2M	Alzheimer's disease	SNOMED CT26929004
clinvar	12p13.31	rs7968679	$3.8 \times 10^{-12}$	-81036	0.692	rs669	A2M	Alpha-2-macroglobulin deficiency	NCBI curation

## Regional Association Plots



