

Analysis Summary: Any Asthma

Phenotype Description

The "any asthma" phenotype combines reports of asthma diagnoses from several sources:

- "Your Health Profile"
 - "Have you ever been diagnosed or treated for any of the following conditions?" (Asthma: Yes, No, Don't Know)
- "Your Medical History"
 - "Have you ever been diagnosed by a doctor with any of the following types of *allergies*?" (Asthma: Yes, No, I don't know)
- "Allergies and Asthma" survey
 - "Have you ever had an asthma attack?" (Yes, No, I'm not sure)
- "Asthma Symptoms" survey
 - "Have you ever been diagnosed by a doctor with asthma or bronchial asthma?" (Yes, No, I'm not sure)
- The Roots into the Future intake form
 - "Have you ever been diagnosed or treated for any of the following conditions?" (Asthma: Yes, No, Don't Know)

We merge the yes/no responses from these questions, with inconsistent responses scored as missing: cases have at least one positive response and no negative responses, and controls have at least one negative response and no positive responses.

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]
any_asthma	case	28399	13045	15354	4549	8791	7307	7752
	control	128843	71655	57188	17452	37451	34020	39920

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

Phenotype	Group	Total	v1/v2	v3	v4
any_asthma	case	28399	2308	20625	5466
	control	128843	12342	91597	24904

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	-0.00869	0.000406	-21.4	6.3×10^{-102}	463.3	9.3×10^{-103}
sexF	0.40229	0.013243	30.4	1.1×10^{-202}	927.0	1.3×10^{-203}
pc.0	-0.01059	0.006554	-1.6	0.11	2.6	0.11
pc.1	-0.15937	0.007466	-21.3	4.1×10^{-101}	498.3	2.2×10^{-110}
pc.2	-0.08912	0.006928	-12.9	7.2×10^{-38}	170.6	5.5×10^{-39}
pc.3	-0.02524	0.006722	-3.8	0.00017	14.1	0.00017
pc.4	0.00249	0.007164	0.3	0.73	0.1	0.73

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	-6301	13772766
failed to converge	-24953	13747813

Genetic Association Tests

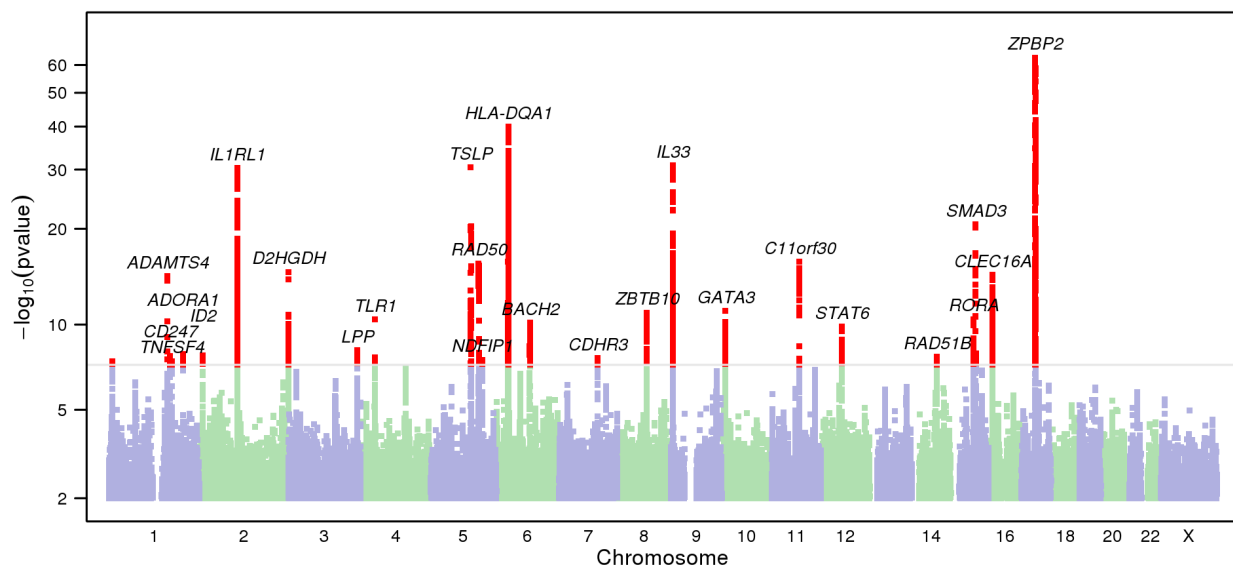
We performed logistic regression assuming an additive model for allelic effects, using the model:

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

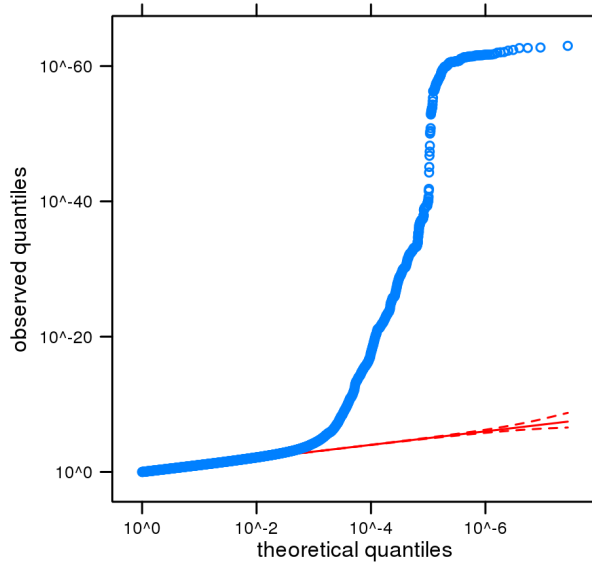
This genome-wide association analysis includes data from 28399 cases and 128843 controls of European ancestry, filtered to remove close relatives.

The results in this report have been adjusted for a genomic control inflation factor $\lambda=1.096$. The equivalent inflation factor for 1000 cases and 1000 controls $\lambda_{1000}=1.002$, and for 10000, $\lambda_{10000}=1.021$.

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
17q12	rs11655198	chr17	38026169	C/T	I	1.0×10^{-63}	0.847	[0.831,0.863]	[ZPBP2]
6p21.32	rs3104367	chr6	32603487	C/T	I	1.0×10^{-40}	0.872	[0.855,0.890]	HLA-DRB5---[]-HLA-DQA1
9p24.1	rs144829310	chr9	6208030	G/T	I	1.3×10^{-31}	1.165	[1.136,1.195]	RANBP6---[]-IL33
5q22.1	rs1837253	chr5	110401872	C/T	I	3.3×10^{-31}	0.876	[0.856,0.896]	SLC25A46---[]-TSLP
2q12.1	rs202011557	chr2	102913642	D/I	I	5.1×10^{-31}	0.842	[0.818,0.868]	IL1RL2--[]--IL1RL1
15q22.33	rs56375023	chr15	67448363	A/G	I	2.4×10^{-21}	0.897	[0.877,0.917]	[SMAD3]
11q13.5	rs7936323	chr11	76293758	A/G	I	1.4×10^{-16}	0.922	[0.905,0.940]	C11orf30--[]--LRRC32
6p21.33	rs2428494	chr6	31322197	A/T	I	1.4×10^{-16}	0.920	[0.902,0.939]	HLA-C-[]--MICA
5q31.1	rs2244012	chr5	131901225	A/G	I	2.1×10^{-16}	1.102	[1.077,1.128]	[RAD50]
2q37.3	rs34290285	chr2	242698640	A/G	I	1.8×10^{-15}	1.107	[1.079,1.135]	[D2HGDH]
16p13.13	rs7203459	chr16	11230703	C/T	I	3.5×10^{-15}	1.092	[1.068,1.117]	[CLEC16A]
1q23.3	rs4233366	chr1	161159147	C/T	I	4.8×10^{-15}	1.090	[1.067,1.114]	B4GALT3--[]ADAMTS4
10p14	rs12413578	chr10	9049253	C/T	I	8.1×10^{-12}	0.891	[0.862,0.921]	GATA3---[]
8q21.13	rs10957978	chr8	81285139	G/T	I	1.1×10^{-11}	0.934	[0.915,0.952]	TPD52---[]---ZBTB10
15q22.2	rs10519068	chr15	61068704	A/G	I	3.8×10^{-11}	1.100	[1.069,1.132]	RORA---[]
4p14	rs5743618	chr4	38798648	A/C	I	3.9×10^{-11}	1.082	[1.057,1.107]	[TLR1]
6q15	rs58521088	chr6	90985198	A/T	I	7.1×10^{-11}	0.934	[0.915,0.954]	[BACH2]
12q13.3	rs3001426	chr12	57509055	C/T	I	1.4×10^{-10}	0.938	[0.919,0.956]	STAT6-[]--LRP1
3q28	rs73196739	chr3	188402471	C/T	I	6.5×10^{-9}	0.922	[0.897,0.948]	[LPP]
1q32.1	rs6683383	chr1	203100504	A/T	I	1.1×10^{-8}	1.062	[1.040,1.084]	[ADORA1]
2p25.1	rs13412757	chr2	8458080	A/G	I	1.3×10^{-8}	1.062	[1.040,1.084]	[]---ID2
1q24.2	rs1723018	chr1	167433420	A/G	I	1.4×10^{-8}	0.945	[0.926,0.963]	[CD247]
14q24.1	rs3784099	chr14	68749927	A/G	I	1.6×10^{-8}	0.942	[0.922,0.961]	[RAD51B]
7q22.3	rs6959584	chr7	105676505	C/T	I	2.0×10^{-8}	1.086	[1.055,1.117]	[CDHR3]
5q31.3	rs200634877	chr5	141529761	D/I	I	2.5×10^{-8}	0.940	[0.919,0.961]	[NDFIP1]
1q25.1	rs6691738	chr1	173152036	G/T	I	2.9×10^{-8}	0.943	[0.923,0.963]	TNFSF18---[]TNFSF4
1p36.22	rs662064	chr1	10557251	C/T	I	3.2×10^{-8}	0.942	[0.922,0.962]	[PEX14]

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
rs11655198	FALSE	FALSE	FALSE					0.9882	0.9858	0.10	0.4874	v2v3v4
rs3104367	FALSE	FALSE	FALSE					0.9432	0.9214	0.023	0.4113	v2v3v4
rs144829310	FALSE	FALSE	FALSE					0.9945	0.9919	0.46	0.1610	v2v3v4
rs1837253	TRUE	TRUE	TRUE	0.9991	0.63	0.51	0.2628	0.9707	0.9576	0.27	0.2615	v2v3v4
rs202011557	FALSE	FALSE	FALSE					0.9517	0.9473	0.040	0.1419	v2v3v4
rs56375023	FALSE	FALSE	FALSE					0.9885	0.9784	0.42	0.7652	v2v3v4
rs7936323	FALSE	FALSE	FALSE					0.9923	0.9866	1.0	0.5171	v2v3v4
rs2428494	FALSE	FALSE	FALSE					0.9727	0.9451	0.0063	0.5793	v2v3v4
rs2244012	TRUE	TRUE	TRUE	0.9998	0.33	0.065	0.2144	0.9998	0.9995	0.091	0.2147	v2v3v4
rs34290285	FALSE	FALSE	FALSE					0.7736	0.5233	2.4×10^{-5}	0.7371	v2v3v4
rs7203459	FALSE	TRUE	FALSE	0.9836	0.36	1.5×10^{-6}	0.7316	0.9966	0.9848	0.60	0.7354	v2v3v4
rs4233366	TRUE	TRUE	TRUE	0.9996	0.83	0.51	0.2594	0.9998	0.9995	0.39	0.2604	v2v3v4
rs12413578	FALSE	FALSE	FALSE					0.8977	0.8389	0.021	0.1096	v2v3v4
rs10957978	FALSE	FALSE	FALSE					0.9981	0.9968	0.11	0.6373	v2v3v4
rs10519068	TRUE	TRUE	FALSE	0.4145	4.7×10^{-28}	1.8×10^{-8}	0.8846	0.9960	0.9918	0.13	0.8618	v2v3v4
rs5743618	FALSE	FALSE	TRUE	0.9963	0.0	0.81	0.4544	0.9073	0.8990	5.4×10^{-31}	0.7200	v2v3v4
rs58521088	FALSE	FALSE	FALSE					0.9760	0.9708	0.33	0.3480	v2v3v4
rs3001426	FALSE	FALSE	FALSE					0.9727	0.9631	0.0010	0.5780	v2v3v4

rs73196739	FALSE	FALSE	FALSE						0.9638	0.9481	0.18	0.1537	v2v3v4
rs6683383	FALSE	FALSE	FALSE						0.9968	0.9816	0.044	0.6762	v2v3v4
rs13412757	FALSE	FALSE	FALSE						0.9914	0.9700	0.0061	0.6737	v2v3v4
rs1723018	FALSE	FALSE	FALSE						0.9931	0.9816	0.12	0.3964	v2v3v4
rs3784099	FALSE	TRUE	TRUE	0.9998	0.00045	0.98	0.7029		0.9984	0.9872	0.044	0.7008	v2v3v4
rs6959584	FALSE	FALSE	FALSE						0.9514	0.8785	1.1×10 ⁻²²	0.1332	v2v3v4
rs200634877	FALSE	FALSE	FALSE						0.8496	0.8303	0.53	0.3501	v2v3v4
rs6691738	FALSE	FALSE	FALSE						0.9813	0.9440	7.6×10 ⁻⁶	0.6860	v2v3v4
rs662064	FALSE	FALSE	FALSE						0.9771	0.9743	0.31	0.2926	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
rs11655198				128843	0.4958				28399	0.4562
rs3104367				128843	0.4186				28399	0.3866
rs144829310				128843	0.1562				28399	0.1782
rs1837253	68978	50450	9265	128843	0.2664	16278	10411	1683	28399	0.2418
rs202011557				128843	0.1454				28399	0.1252
rs56375023				128843	0.7679				28399	0.7486
rs7936323				128843	0.5216				28399	0.5004
rs2428494				128843	0.5797				28399	0.5577
rs2244012	79996	42887	5922	128843	0.2124	17014	9970	1409	28399	0.2252
rs34290285				128843	0.7346				28399	0.7508
rs7203459	6639	35838	48086	128843	0.7324	1357	7706	11288	28399	0.7489
rs4233366	71268	49016	8517	128843	0.2566	14946	11295	2151	28399	0.2749
rs12413578				128843	0.1097				28399	0.1007
rs10957978				128843	0.6408				28399	0.6245
rs10519068	1201	13646	53463	128843	0.8606	191	2649	11210	28399	0.8727
rs743618	2208	22587	0	128843	0.7277	452	4994	0	28399	0.7431
rs58521088				128843	0.3523				28399	0.3386
rs3001426				128843	0.5772				28399	0.5614
rs73196739				128843	0.1563				28399	0.1456
rs6683383				128843	0.6737				28399	0.6866
rs13412757				128843	0.6681				28399	0.6816
rs1723018				128843	0.3974				28399	0.3856
rs3784099	10101	48233	58138	128843	0.7051	2531	10904	12649	28399	0.6926
rs6959584				128843	0.1305				28399	0.1386
rs200634877				128843	0.3531				28399	0.3422
rs6691738				128843	0.6907				28399	0.6798
rs662064				128843	0.2952				28399	0.2844

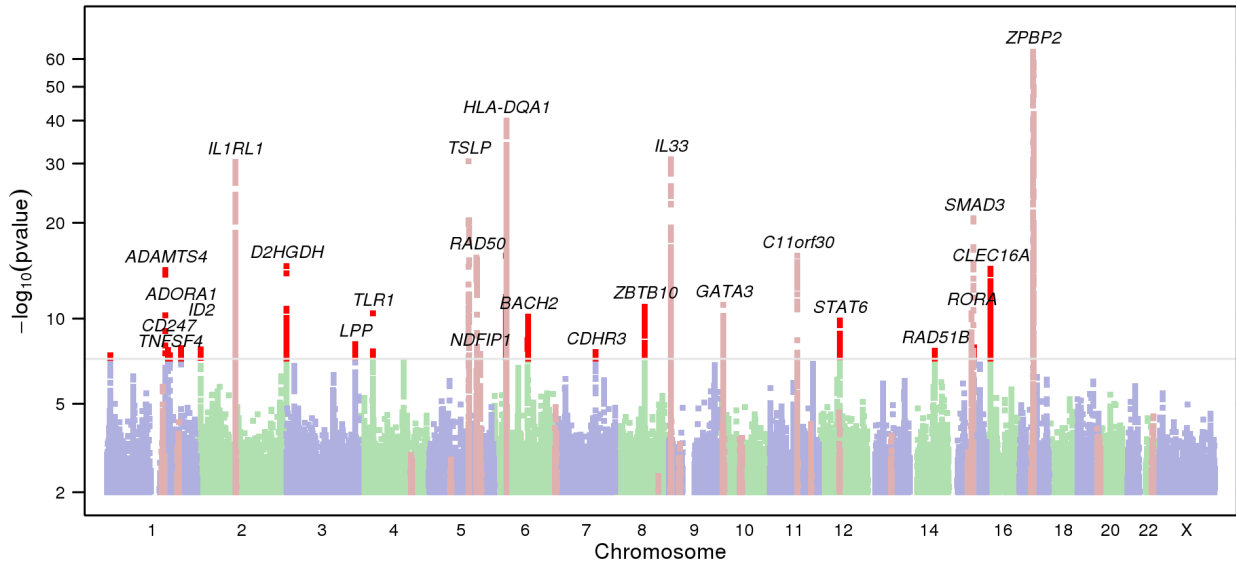
Annotations from NHGRI GWAS Catalog

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
17q12	38026169	rs11655198	1.0×10 ⁻⁶³	-113792	0.740	rs12946510	4.0×10 ⁻³⁸	23128233	Inflammatory bowel disease	IKZF3,ZPBP2,GSDMB,ORMDL3,GSDMA
17q12	38026169	rs11655198	1.0×10 ⁻⁶³	-103910	0.797	rs907092	8.0×10 ⁻⁶	19458352	Primary biliary cirrhosis	IKZF3
17q12	38026169	rs11655198	1.0×10 ⁻⁶³	-49700	0.922	rs9303277	7.0×10 ⁻⁶	23740937	Systemic lupus erythematosus and Systemic sclerosis	IKZF3
17q12	38026169	rs11655198	1.0×10 ⁻⁶³	-49700	0.922	rs9303277	4.0×10 ⁻⁹	23000144	Primary biliary cirrhosis	IKZF3, ZPBP2, ...
17q12	38026169	rs11655198	1.0×10 ⁻⁶³	-49700	0.922	rs9303277	2.0×10 ⁻⁹	20639880	Primary biliary cirrhosis	IKZF3,ZPBP2,GSDMB,ORMDL3
17q12	38026169	rs11655198	1.0×10 ⁻⁶³	14594	0.842	rs2872507	2.0×10 ⁻⁶	21829393	Type 1 diabetes autoantibodies	ORMDL3
17q12	38026169	rs11655198	1.0×10 ⁻⁶³	14594	0.842	rs2872507	5.0×10 ⁻¹¹	21297633	Ulcerative colitis	IKZF3, ORMDL3, ...
17q12	38026169	rs11655198	1.0×10 ⁻⁶³	14594	0.842	rs2872507	2.0×10 ⁻⁹	21102463	Crohn's disease	GSDML,ZPBP2,ORMDL3,IKZF3
17q12	38026169	rs11655198	1.0×10 ⁻⁶³	14594	0.842	rs2872507	9.0×10 ⁻⁷	20453842	Rheumatoid arthritis	IKZF3
17q12	38026169	rs11655198	1.0×10 ⁻⁶³	14594	0.842	rs2872507	5.0×10 ⁻⁹	18587394	Crohn's disease	ORMDL3
17q12	38026169	rs11655198	1.0×10 ⁻⁶³	25179	0.964	rs8067378	9.0×10 ⁻¹⁰	23817570	Cervical cancer	GSDMB
17q12	38026169	rs11655198	1.0×10 ⁻⁶³	36027	0.815	rs2305480	1.0×10 ⁻⁷	20860503	Asthma	GSDMB
17q12	38026169	rs11655198	1.0×10 ⁻⁶³	36027	0.815	rs2305480	3.0×10 ⁻⁸	20228799	Ulcerative colitis	ORMDL3,ZPBP2M,GSDML
17q12	38026169	rs11655198	1.0×10 ⁻⁶³	38236	0.811	rs11078927	2.0×10 ⁻¹⁶	21804549	Asthma	GSDMB
17q12	38026169	rs11655198	1.0×10 ⁻⁶³	40071	0.896	rs2290400	6.0×10 ⁻¹³	19430480	Type 1 diabetes	ORMDL3
17q12	38026169	rs11655198	1.0×10 ⁻⁶³	43780	0.885	rs7216389	9.0×10 ⁻¹¹	17611496	Asthma	ORMDL3
17q12	38026169	rs11655198	1.0×10 ⁻⁶³	47862	0.840	rs9303280	9.0×10 ⁻⁹	23817569	Self-reported allergy	GSDMB, IKZF3
17q12	38026169	rs11655198	1.0×10 ⁻⁶³	63175	0.677	rs4794820	1.0×10 ⁻⁸	22561531	Asthma	ORMDL3
6p21.32	32603487	rs3104367	1.0×10 ⁻⁴⁰	885	0.796	rs9272346	2.0×10 ⁻⁸	23181788	Asthma	HLA-DQA1
6p21.32	32603487	rs3104367	1.0×10 ⁻⁴⁰	885	0.796	rs9272346	6.0×10 ⁻¹²⁹	18978792	Type 1 diabetes	HLA
6p21.32	32603487	rs3104367	1.0×10 ⁻⁴⁰	885	0.796	rs9272346	5.0×10 ⁻¹³⁴	17554300	Type 1 diabetes	MHC
9p24.1	6208030	rs144829310	1.3×10 ⁻³¹	-35650	0.940	rs7032572	2.0×10 ⁻⁹	23817569	Self-reported allergy	RANBP6, IL33
9p24.1	6208030	rs144829310	1.3×10 ⁻³¹	-17954	0.959	rs1342326	9.0×10 ⁻¹⁰	20860503	Asthma	IL33
9p24.1	6208030	rs144829310	1.3×10 ⁻³¹	-14575	0.534	rs2381416	2.0×10 ⁻¹²	21804549	Asthma	IL33
5q22.1	110401872	rs1837253	3.3×10 ⁻³¹	0	1.000	rs1837253	1.0×10 ⁻¹⁴	21804549	Asthma	TSLP
5q22.1	110401872	rs1837253	3.3×10 ⁻³¹	0	1.000	rs1837253	1.0×10 ⁻¹⁶	21804548	Asthma	TSLP
2q12.1	102913642	rs202011557	5.1×10 ⁻³¹	-34178	0.819	rs10189629	2.0×10 ⁻¹⁶	23817569	Self-reported allergy	IL1RL2, IL1RL1
2q12.1	102913642	rs202011557	5.1×10 ⁻³¹	39975	0.915	rs3771180	2.0×10 ⁻¹⁵	21804549	Asthma	IL1RL1
2q12.1	102913642	rs202011557	5.1×10 ⁻³¹	41440	0.915	rs13408661	1.0×10 ⁻⁹	23028483	Asthma	IL1RL1, IL18R1

2q12.1	102913642	rs202011557	5.1×10^{-31}	46568	0.896	rs3771175	5.0×10^{-11}	23817571	Allergic sensitization	IL1RL1, IL18R1, IL18RAP
15q22.33	67448363	rs56375023	2.4×10^{-21}	-5767	0.984	rs17293632	6.0×10^{-16}	23128233	Inflammatory bowel disease	SMAD3
15q22.33	67448363	rs56375023	2.4×10^{-21}	-5767	0.984	rs17293632	3.0×10^{-19}	21102463	Crohn's disease	SMAD3
15q22.33	67448363	rs56375023	2.4×10^{-21}	1942	0.992	rs17228058	1.0×10^{-8}	23817569	Self-reported allergy	SMAD3
11q13.5	76293758	rs7936323	1.4×10^{-16}	-23075	0.570	rs7130588	4.0×10^{-13}	23886662	Atopic dermatitis	C11orf30,LRRC32
11q13.5	76293758	rs7936323	1.4×10^{-16}	-23075	0.570	rs7130588	2.0×10^{-8}	21907864	Asthma	LRRC32
11q13.5	76293758	rs7936323	1.4×10^{-16}	5436	0.898	rs2155219	1.0×10^{-18}	23817571	Allergic sensitization	C11orf30, LRRC32
11q13.5	76293758	rs7936323	1.4×10^{-16}	5436	0.898	rs2155219	2.0×10^{-19}	23817569	Self-reported allergy	C11orf30, LRRC32
11q13.5	76293758	rs7936323	1.4×10^{-16}	5436	0.898	rs2155219	4.0×10^{-36}	23128233	Inflammatory bowel disease	Intergenic
11q13.5	76293758	rs7936323	1.4×10^{-16}	5436	0.898	rs2155219	1.0×10^{-8}	22036096	IgE grass sensitization	C11orf30, LRRC32
11q13.5	76293758	rs7936323	1.4×10^{-16}	5436	0.898	rs2155219	4.0×10^{-8}	22036096	Allergic rhinitis	C11orf30, LRRC32
11q13.5	76293758	rs7936323	1.4×10^{-16}	5436	0.898	rs2155219	5.0×10^{-16}	21297633	Ulcerative colitis	Intergenic
11q13.5	76293758	rs7936323	1.4×10^{-16}	7558	0.642	rs7927894	8.0×10^{-10}	19349984	Atopic dermatitis	C11orf30
11q13.5	76293758	rs7936323	1.4×10^{-16}	7558	0.642	rs7927894	1.0×10^{-9}	18587394	Crohn's disease	C11orf30
11q13.5	76293758	rs7936323	1.4×10^{-16}	7617	0.642	rs7927997	6.0×10^{-13}	21102463	Crohn's disease	C11orf30
5q31.1	131901225	rs2244012	2.1×10^{-16}	0	1.000	rs2244012	3.0×10^{-7}	20159242	Asthma	RAD50
5q31.1	131901225	rs2244012	2.1×10^{-16}	71952	1.000	rs2040704	4.0×10^{-8}	18846228	IgE levels	RAD50
16p13.13	11230703	rs7203459	3.5×10^{-15}	-50830	0.508	rs12708716	5.0×10^{-14}	21829393	Type 1 diabetes autoantibodies	CLEC16A
16p13.13	11230703	rs7203459	3.5×10^{-15}	-50830	0.508	rs12708716	2.0×10^{-16}	19430480	Type 1 diabetes	CLEC16A
16p13.13	11230703	rs7203459	3.5×10^{-15}	-50830	0.508	rs12708716	7.0×10^{-13}	18978792	Type 1 diabetes	CLEC16A
16p13.13	11230703	rs7203459	3.5×10^{-15}	-50830	0.508	rs12708716	5.0×10^{-7}	17554300	Type 1 diabetes	KIAA0350
16p13.13	11230703	rs7203459	3.5×10^{-15}	-50830	0.508	rs12708716	3.0×10^{-18}	17554260	Type 1 diabetes	KIAA0350
16p13.13	11230703	rs7203459	3.5×10^{-15}	-20288	0.500	rs9923856	6.0×10^{-6}	23042114	Atopic dermatitis	CLEC16A
16p13.13	11230703	rs7203459	3.5×10^{-15}	0	1.000	rs7203459	2.0×10^{-7}	23817569	Self-reported allergy	CLEC16A
16p13.13	11230703	rs7203459	3.5×10^{-15}	8080	0.617	rs2903692	7.0×10^{-11}	17632545	Type 1 diabetes	KIAA0350
8q21.13	81285139	rs10957978	1.1×10^{-11}	-16984	0.853	rs6473223	8.0×10^{-8}	23817569	Self-reported allergy	TPD52, ZBTB10
8q21.13	81285139	rs10957978	1.1×10^{-11}	23011	0.561	rs7000782	1.0×10^{-6}	22197932	Atopic dermatitis	ZBTB10
15q22.2	61068704	rs10519068	3.8×10^{-11}	1284	0.955	rs11071559	1.0×10^{-7}	20860503	Asthma	RORA
4p14	38798648	rs5743618	3.9×10^{-11}	-13924	0.640	rs10004195	1.0×10^{-18}	23652523	Helicobacter pylori serologic status	TLR10,TLR1,FAM114A1,TLR6,KLF3
4p14	38798648	rs5743618	3.9×10^{-11}	-6124	0.619	rs4543123	9.0×10^{-6}	23743675	Alcohol consumption	Intergenic
4p14	38798648	rs5743618	3.9×10^{-11}	12903	0.594	rs2101521	5.0×10^{-21}	23817569	Self-reported allergy	TLR1,TLR6,TLR10
4p14	38798648	rs5743618	3.9×10^{-11}	14228	0.677	rs17616434	5.0×10^{-11}	23817571	Allergic sensitization	TLR1,TLR6, TLR10, ...
6q15	90985198	rs58521088	7.1×10^{-11}	-58586	0.713	rs10806425	4.0×10^{-10}	20190752	Celiac disease	BACH2, MAP3K7
6q15	90985198	rs58521088	7.1×10^{-11}	-27735	0.576	rs3757247	3.0×10^{-8}	22561518	Vitiligo	BACH2
6q15	90985198	rs58521088	7.1×10^{-11}	-27735	0.576	rs3757247	1.0×10^{-6}	18840781	Type 1 diabetes	BACH2
6q15	90985198	rs58521088	7.1×10^{-11}	-26967	0.616	rs11755527	3.0×10^{-8}	21829393	Type 1 diabetes autoantibodies	BACH2
6q15	90985198	rs58521088	7.1×10^{-11}	-26967	0.616	rs11755527	5.0×10^{-8}	19430480	Type 1 diabetes	BACH2
6q15	90985198	rs58521088	7.1×10^{-11}	-26967	0.616	rs11755527	5.0×10^{-12}	18978792	Type 1 diabetes	BACH2
6q15	90985198	rs58521088	7.1×10^{-11}	-12039	0.966	rs1847472	2.0×10^{-10}	23128233	Inflammatory bowel disease	Intergenic
6q15	90985198	rs58521088	7.1×10^{-11}	-12039	0.966	rs1847472	5.0×10^{-9}	21102463	Crohn's disease	BACH2
6q15	90985198	rs58521088	7.1×10^{-11}	11571	0.677	rs12212193	4.0×10^{-8}	21833088	Multiple sclerosis	BACH2
12q13.3	57509055	rs3001426	1.4×10^{-10}	-5280	0.674	rs167769	2.0×10^{-6}	20208534	Eosinophilic esophagitis (pediatric)	STAT6
12q13.3	57509055	rs3001426	1.4×10^{-10}	18228	0.602	rs11172113	1.0×10^{-10}	23793025	Migraine without aura	LRP1
12q13.3	57509055	rs3001426	1.4×10^{-10}	18228	0.602	rs11172113	1.0×10^{-6}	23793025	Migraine - clinic-based	LRP1
12q13.3	57509055	rs3001426	1.4×10^{-10}	18228	0.602	rs11172113	4.0×10^{-19}	23793025	Migraine	LRP1
12q13.3	57509055	rs3001426	1.4×10^{-10}	18228	0.602	rs11172113	8.0×10^{-6}	23284291	Pulmonary function (interaction)	LRP1
12q13.3	57509055	rs3001426	1.4×10^{-10}	18228	0.602	rs11172113	3.0×10^{-8}	22683712	Migraine	LRP1
12q13.3	57509055	rs3001426	1.4×10^{-10}	18228	0.602	rs11172113	1.0×10^{-8}	21946350	Pulmonary function	LRP1
12q13.3	57509055	rs3001426	1.4×10^{-10}	18228	0.602	rs11172113	4.0×10^{-9}	21666692	Migraine	LRP1
2p25.1	8458080	rs13412757	1.3×10^{-8}	-15832	0.761	rs10174949	1.0×10^{-7}	23817569	Self-reported allergy	ID2
1q24.2	167433420	rs1723018	1.4×10^{-8}	-12995	0.908	rs2056626	3.0×10^{-6}	21779181	Systemic sclerosis	CD247
1q24.2	167433420	rs1723018	1.4×10^{-8}	-12995	0.908	rs2056626	1.0×10^{-6}	21750679	Systemic sclerosis	CD247
1q24.2	167433420	rs1723018	1.4×10^{-8}	-12995	0.908	rs2056626	3.0×10^{-9}	20383147	Systemic sclerosis	CD247
14q24.1	68749927	rs3784099	1.6×10^{-8}	0	1.000	rs3784099	3.0×10^{-7}	22232737	Breast cancer (survival)	RAD51L1
14q24.1	68749927	rs3784099	1.6×10^{-8}	3666	0.982	rs911263	2.0×10^{-11}	21399635	Primary biliary cirrhosis	RAD51L1
14q24.1	68749927	rs3784099	1.6×10^{-8}	35150	0.718	rs8017304	9.0×10^{-11}	23455636	Age-related macular degeneration	RAD51B
14q24.1	68749927	rs3784099	1.6×10^{-8}	38997	0.718	rs1956529	3.0×10^{-8}	20195514	Primary tooth development (number of teeth)	RAD51L1
14q24.1	68749927	rs3784099	1.6×10^{-8}	63188	0.535	rs1570106	8.0×10^{-9}	20881960	Height	RAD51L1
5q31.3	141529761	rs200634877	2.5×10^{-8}	-16557	0.764	rs6863411	4.0×10^{-14}	23128233	Inflammatory bowel disease	SPRY4,NDFIP1
5q31.3	141529761	rs200634877	2.5×10^{-8}	-6761	0.764	rs1062158	2.0×10^{-6}	21833088	Multiple sclerosis	NDFIP1
1p36.22	10557251	rs662064	3.2×10^{-8}	8964	0.703	rs616488	1.0×10^{-8}	23535733	Breast cancer	PEX14
1p36.22	10557251	rs662064	3.2×10^{-8}	8964	0.703	rs616488	2.0×10^{-10}	23535729	Breast cancer	PEX14
1p36.22	10557251	rs662064	3.2×10^{-8}	24407	0.856	rs2056417	4.0×10^{-7}	23817569	Self-reported allergy	PEX14

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
1q21.3	152492559	rs4845783	0.097	0	1.000	rs4845783	6.0×10^{-6}	21804549	Asthma	CRCT1
1q21.3	154426264	rs4129267	0.0097	0	1.000	rs4129267	2.0×10^{-8}	21907864	Asthma	IL6R
1q23.1	158932555	rs1101999	0.51	0	1.000	rs1101999	4.0×10^{-9}	21804549	Asthma	PYHIN1
1q31.3	197325908	rs2786098	0.48	0	1.000	rs2786098	2.0×10^{-13}	20032318	Asthma	DENND1B, CRB1
2q12.1	102953617	rs3771180	9.0×10^{-30}	0	1.000	rs3771180	2.0×10^{-15}	21804549	Asthma	IL1RL1
2q12.1	102955082	rs13408661	1.1×10^{-29}	0	1.000	rs13408661	1.0×10^{-9}	23028483	Asthma	IL1RL1, IL18R1
2q12.1	102971200	rs9807989	2.5×10^{-24}	0	1.000	rs9807989	6.0×10^{-8}	22561531	Asthma	IL18R1, IL1R1
2q12.1	102986222	rs3771166	3.6×10^{-22}	0	1.000	rs3771166	3.0×10^{-9}	20860503	Asthma	IL18R1
4q31.21	144003159	rs7686660	0.37	0	1.000	rs7686660	2.0×10^{-12}	21804548	Asthma	LOC729675
4q31.21	144357737	rs3805236	0.62	0	1.000	rs3805236	7.0×10^{-8}	21804548	Asthma	GAB1
5q12.1	59369794	rs1588265	0.19	0	1.000	rs1588265	3.0×10^{-8}	19426955	Asthma	PDE4D
5q22.1	110401872	rs1837253	3.3×10^{-31}	0	1.000	rs1837253	1.0×10^{-14}	21804549	Asthma	TSLP
5q22.1	110401872	rs1837253	3.3×10^{-31}	0	1.000	rs1837253	1.0×10^{-16}	21804548	Asthma	TSLP
5q31.1	131723288	rs2073643	0.00042	0	1.000	rs2073643	2.0×10^{-7}	20860503	Asthma	SLC22A5
5q31.1	131796922	rs11745587	1.1×10^{-6}	0	1.000	rs11745587	2.0×10^{-6}	22561531	Asthma	C5orf56
5q31.1	131901225	rs2244012	2.1×10^{-16}	0	1.000	rs2244012	3.0×10^{-7}	20159242	Asthma	RAD50
5q31.1	131995843	rs1295686	2.5×10^{-13}	0	1.000	rs1295686	1.0×10^{-7}	20860503	Asthma	IL13
5q31.3	141445980	rs6867913	0.0070	0	1.000	rs6867913	4.0×10^{-6}	22561531	Asthma	NDFIP1
6p21.32	32155581	rs204993	1.0×10^{-6}	0	1.000	rs204993	2.0×10^{-15}	21804548	Asthma	PBX2
6p21.32	32184345	rs404860	0.11	0	1.000	rs404860	4.0×10^{-23}	21804548	Asthma	NOTCH4
6p21.32	32338695	rs3129943	0.58	0	1.000	rs3129943	3.0×10^{-15}	21804548	Asthma	C6orf10
6p21.32	32358513	rs3117098	4.2×10^{-6}	0	1.000	rs3117098	5.0×10^{-12}	21804548	Asthma	BTNL2
6p21.32	32379489	rs9268516	1.3×10^{-21}	0	1.000	rs9268516	1.0×10^{-8}	23028483	Asthma	BTNL2, HLA-DRA
6p21.32	32414273	rs3129890	0.13	0	1.000	rs3129890	5.0×10^{-13}	21804548	Asthma	HLA-DRA
6p21.32	32604372	rs9272346	6.8×10^{-34}	0	1.000	rs9272346	2.0×10^{-8}	23181788	Asthma	HLA-DQA1
6p21.32	32625869	rs9273349	9.2×10^{-33}	0	1.000	rs9273349	7.0×10^{-14}	20860503	Asthma	HLA-DQ
6p21.32	32658079	rs7775228	3.9×10^{-5}	0	1.000	rs7775228	5.0×10^{-15}	21804548	Asthma	HLA-DQB1
6p21.32	32687973	rs9275698	0.33	0	1.000	rs9275698	5.0×10^{-12}	21804548	Asthma	HLA-DQA2
6p21.32	32961361	rs9500927	2.6×10^{-5}	0	1.000	rs9500927	4.0×10^{-9}	21804548	Asthma	HLA-DOA
6p21.32	33042880	rs987870	2.8×10^{-6}	0	1.000	rs987870	2.0×10^{-10}	21814517	Asthma	HLA, DPB1
6q27	166534742	rs6456042	0.46	0	1.000	rs6456042	6.0×10^{-6}	22538805	Asthma	T
8q24.11	118025645	rs3019885	0.13	0	1.000	rs3019885	5.0×10^{-13}	21814517	Asthma	SLC30A8
9p24.1	6190076	rs1342326	1.7×10^{-31}	0	1.000	rs1342326	9.0×10^{-10}	20860503	Asthma	IL33
9p24.1	6193455	rs2381416	3.7×10^{-25}	0	1.000	rs2381416	2.0×10^{-12}	21804549	Asthma	IL33
9p21.1	32433526	rs10970976	0.61	0	1.000	rs10970976	4.0×10^{-6}	22561531	Asthma	ACO1
10p14	8972018	rs10508372	2.0×10^{-5}	0	1.000	rs10508372	2.0×10^{-15}	21804548	Asthma	LOC338591
10q21.1	53493473	rs7922491	0.23	0	1.000	rs7922491	5.0×10^{-7}	21907864	Asthma	PRKG1
11q13.5	76270683	rs7130588	1.4×10^{-11}	0	1.000	rs7130588	2.0×10^{-8}	21907864	Asthma	LRRC32
11q23.2	114231255	rs11214966	0.70	0	1.000	rs11214966	6.0×10^{-7}	21804549	Asthma	C11orf71
12q13.2	56364321	rs2069408	0.017	0	1.000	rs2069408	1.0×10^{-10}	21804548	Asthma	CDK2
12q13.2	56412487	rs1701704	0.0034	0	1.000	rs1701704	2.0×10^{-13}	21804548	Asthma	IKZF4
13q21.31	63638329	rs3119939	0.88	0	1.000	rs3119939	8.0×10^{-6}	21907864	Asthma	PCDH20
15q21.2	51969668	rs17525472	0.88	0	1.000	rs17525472	2.0×10^{-6}	20159242	Asthma	SCG3
15q22.2	61069988	rs11071559	6.8×10^{-11}	0	1.000	rs11071559	1.0×10^{-7}	20860503	Asthma	RORA
15q22.33	67446785	rs744910	2.2×10^{-11}	0	1.000	rs744910	4.0×10^{-9}	20860503	Asthma	SMAD3
17q12	38062196	rs2305480	2.6×10^{-62}	0	1.000	rs2305480	1.0×10^{-7}	20860503	Asthma	GSDMB
17q12	38064405	rs11078927	3.7×10^{-62}	0	1.000	rs11078927	2.0×10^{-16}	21804549	Asthma	GSDMB
17q12	38069949	rs7216389	5.0×10^{-62}	0	1.000	rs7216389	9.0×10^{-11}	17611496	Asthma	ORMDL3

17q12	38089344	rs4794820	4.5×10 ⁻⁴⁸	0	1.000	rs4794820	1.0×10 ⁻⁸	22561531	Asthma	ORMDL3
17q12	38095174	rs6503525	3.6×10 ⁻³⁸	0	1.000	rs6503525	5.0×10 ⁻⁷	21150878	Asthma	ORMDL3
17q21.1	38121993	rs3894194	1.5×10 ⁻³⁴	0	1.000	rs3894194	5.0×10 ⁻⁹	20860503	Asthma	GSDMA
19q13.42	53682042	rs16984547	0.74	0	1.000	rs16984547	4.0×10 ⁻⁶	22561531	Asthma	ZNF665
20p13	3827309	rs4815617	0.11	0	1.000	rs4815617	8.0×10 ⁻⁶	20159242	Asthma	KIAA1271
22q12.3	37534034	rs2284033	4.2×10 ⁻⁵	0	1.000	rs2284033	1.0×10 ⁻⁸	20860503	Asthma	IL2RB

Nearby Nonsynonymous SNPs

region	position	our.name	our.pval	dist	rsqr	assay.name	gene	aa.chg
17q12	38026169	rs11655198	1.0×10 ⁻⁶³	2465	0.974	rs11557467	ZBPB2	S173I
17q12	38026169	rs11655198	1.0×10 ⁻⁶³	36027	0.815	rs2305480	GSDMB	P311S
17q12	38026169	rs11655198	1.0×10 ⁻⁶³	36048	0.943	rs2305479	GSDMB	G304R
6p21.32	32603487	rs3104367	1.0×10 ⁻⁴⁰	1770	0.712	rs1047989	HLA-DQA1	L8M
6p21.32	32603487	rs3104367	1.0×10 ⁻⁴⁰	5618	0.700	rs1129740	HLA-DQA1	C34Y
6p21.32	32603487	rs3104367	1.0×10 ⁻⁴⁰	5639	0.868	rs1071630	HLA-DQA1	F41S
6p21.32	32603487	rs3104367	1.0×10 ⁻⁴⁰	5720	0.570	rs1142324	HLA-DQA1	A68V
6p21.32	32603487	rs3104367	1.0×10 ⁻⁴⁰	5825	0.581	rs1129808	HLA-DQA1	Y103S
6p21.32	32603487	rs3104367	1.0×10 ⁻⁴⁰	6522	0.583	rs2308891	HLA-DQA1	Q198?
2q12.1	102913642	rs202011557	5.1×10 ⁻³¹	41826	0.516	rs1041973	IL1RL1	A78E
2q37.3	242698640	rs34290285	1.8×10 ⁻¹⁵	-7965	0.814	rs1106639	D2HGDH	V338I
4p14	38798648	rs5743618	3.9×10 ⁻¹¹	0	1.000	rs5743618	TLR1	S602I
4p14	38798648	rs5743618	3.9×10 ⁻¹¹	1062	0.676	rs4833095	TLR1	N248S
7q22.3	105676505	rs6959584	2.0×10 ⁻⁸	-18054	0.693	rs6967330	CDHR3	C529Y

Nearby Expression QTLs

region	position	our.name	our.pval	dist	rsqr	assay.name	eqtl.dist	eqtl.gene	eqtl.pval	eqtl.rsqr	tissue	pubmed.id
1q23.3	161159147	rs4233366	4.8×10 ⁻¹⁵	0	1.000	rs4233366	29543	FCER1G	1.4×10 ⁻¹¹	0.151	Monocyte	22446964
1q23.3	161159147	rs4233366	4.8×10 ⁻¹⁵	19537	0.573	rs3924264	162917	USF1	7.6×10 ⁻¹⁵	0.043	Monocyte	20502693
1q23.3	161159147	rs4233366	4.8×10 ⁻¹⁵	30566	0.535	rs4489574	0	FCER1G	2.7×10 ⁻⁹	0.027	Monocyte	20502693
1q32.1	203100504	rs6683383	1.1×10 ⁻⁸	-9528	0.983	rs7555556	46049	MYBPH	4.1×10 ⁻⁸	0.102	Monocyte	22446964
1q32.1	203100504	rs6683383	1.1×10 ⁻⁸	-1686	1.000	rs10920570	38121	MYBPH	5.2×10 ⁻¹⁸	0.052	Monocyte	20502693
1q32.1	203100504	rs6683383	1.1×10 ⁻⁸	-1686	1.000	rs10920570	37655	ADORA1	2.0×10 ⁻³⁶	0.434	Monocyte	22446964
1q32.1	203100504	rs6683383	1.1×10 ⁻⁸	0	1.000	rs6683383	0	ADORA1	6.0×10 ⁻⁹⁶	0.255	Monocyte	20502693
1q32.1	203100504	rs6683383	1.1×10 ⁻⁸	8004	0.779	rs10920573	60644	PPFIA4	9.3×10 ⁻⁶	0.015	Monocyte	20502693
5q31.3	141529761	rs200634877	2.5×10 ⁻⁸	-36373	0.764	rs6863411	-24880	NDFIP1	8.1×10 ⁻⁶		Liver	18462017
5q31.3	141529761	rs200634877	2.5×10 ⁻⁸	-36373	0.764	rs6863411	-983365	PCDHB6	0.00012		Liver	18462017
5q31.3	141529761	rs200634877	2.5×10 ⁻⁸	-36373	0.764	rs6863411	637088	ARHGAP26	0.00017		Liver	18462017
5q31.3	141529761	rs200634877	2.5×10 ⁻⁸	-36373	0.764	rs6863411	-657635	PCDHGC3	0.00024		Liver	18462017
5q31.3	141529761	rs200634877	2.5×10 ⁻⁸	-36373	0.764	rs6863411	-802952	PCDHGA1	0.00035		Liver	18462017
5q31.3	141529761	rs200634877	2.5×10 ⁻⁸	-36373	0.764	rs6863411	-888057	PCDHB15	0.00051		Liver	18462017
5q31.3	141529761	rs200634877	2.5×10 ⁻⁸	-36373	0.764	rs6863411	459918	FGF1	0.0035		Liver	18462017
1p36.22	10557251	rs662064	3.2×10 ⁻⁸	-17708	0.980	rs607941	-750269	CLSTN1	0.00018	0.049	B-Cell	22446964
1p36.22	10557251	rs662064	3.2×10 ⁻⁸	-16740	0.533	rs12402967	-153706	PGD	0.00010		Lymphoblastoid	20220756
1p36.22	10557251	rs662064	3.2×10 ⁻⁸	-12704	0.980	rs668805	0	PEX14	1.2×10 ⁻¹⁹	0.057	Monocyte	20502693
1p36.22	10557251	rs662064	3.2×10 ⁻⁸	6241	0.949	rs2506901	-72202	APITD1	6.9×10 ⁻⁹	0.113	Monocyte	22446964

Nearby Clinical Variants

source	region	our.name	our.pval	dist	rsqr	assay.name	gene	phenotype	accession
clinvar	4p14	rs5743618	3.9×10 ⁻¹¹	0	1.000	rs5743618	TLR1	Leprosy 5	NCBI curation
clinvar	4p14	rs5743618	3.9×10 ⁻¹¹	1062	0.676	rs4833095	TLR1	Leprosy 5	NCBI curation

Regional Association Plots

