

Analysis Summary: Tonsillectomy

Phenotype Description

Data on "tonsillectomy" comes from two sources:

- "Your Medical History" survey:
 - "Have you ever had any of the following surgeries? (Tonsillectomy)" (Yes, No, I don't know)
- Research snippet:
 - "Have you had your tonsils removed?" (Yes, No, I'm not sure)

Cases gave a positive response to either question, and controls gave a negative response to either question. Individuals with discordant responses were excluded.

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]
tonsillectomy	case	60098	29487	30611	3212	8715	17103	31068
	control	113323	60724	52599	19679	40698	29860	23086

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

Phenotype	Group	Total	v1/v2	v3	v4
tonsillectomy	case	60098	5971	45975	8152
	control	113323	9917	89122	14284

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	0.0536	0.000373	143.8	0.0	24418.6	0.0
sexF	0.1229	0.010907	11.3	1.9×10^{-29}	127.0	1.9×10^{-29}
pc.0	-0.0282	0.005345	-5.3	1.4×10^{-7}	27.7	1.4×10^{-7}
pc.1	-0.0444	0.005708	-7.8	7.9×10^{-15}	61.1	5.6×10^{-15}
pc.2	0.0270	0.005450	5.0	7.2×10^{-7}	24.5	7.4×10^{-7}
pc.3	0.0231	0.005478	4.2	2.4×10^{-5}	17.8	2.4×10^{-5}
pc.4	0.0242	0.005681	4.3	2.1×10^{-5}	18.4	1.8×10^{-5}

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	-6960	13772107
failed to converge	-14677	13757430

Genetic Association Tests

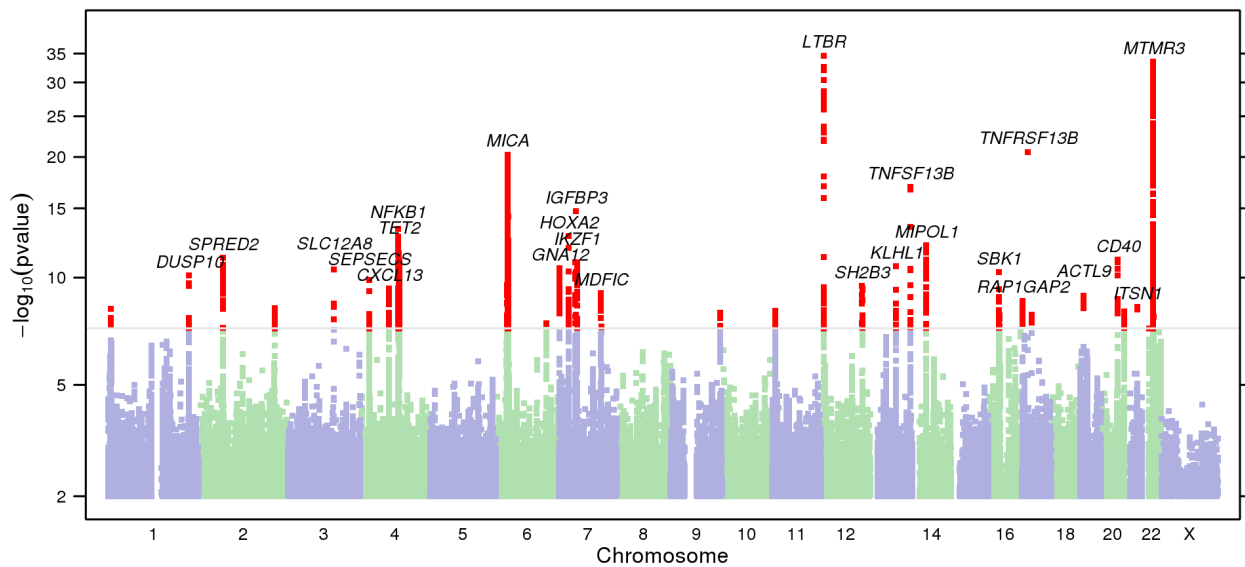
We performed logistic regression assuming an additive model for allelic effects, using the model:

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

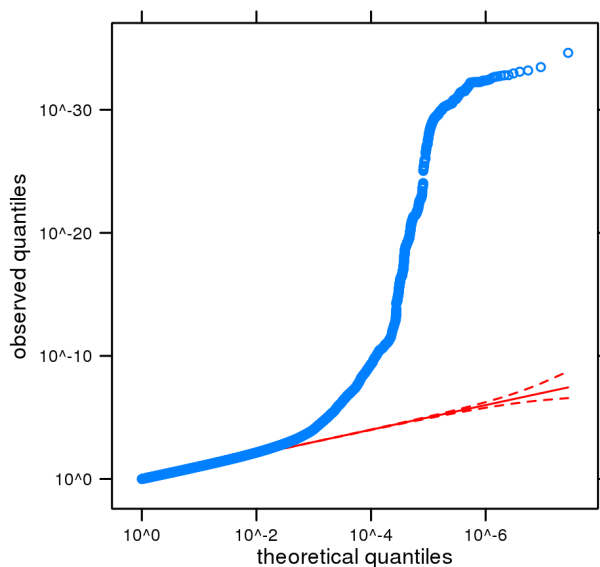
This genome-wide association analysis includes data from 60098 cases and 113323 controls of European ancestry, filtered to remove close relatives.

The results in this report have been adjusted for a genomic control inflation factor $\lambda=1.103$. The equivalent inflation factor for 1000 cases and 1000 controls $\lambda_{1000}=1.001$, and for 10000, $\lambda_{10000}=1.013$.

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
12p13.31	rs10849448	chr12	6493351	A/G	I	2.3×10 ⁻³⁵	0.887	[0.871,0.904]	[LTBR]
22q12.2	rs201112509	chr22	30324654	D/I	I	3.4×10 ⁻³⁴	1.120	[1.100,1.141]	[MTMR3]
17p11.2	rs34557412	chr17	16852187	A/G	I	2.7×10 ⁻²¹	1.595	[1.449,1.755]	[TNFRSF13B]
6p21.33	rs41543314	chr6	31322690	A/G	I	5.4×10 ⁻²¹	1.232	[1.180,1.286]	HLA-C--[]--MICA
13q33.3	rs200748895	chr13	108960380	D/I	I	1.2×10 ⁻¹⁷	0.794	[0.753,0.837]	[TNFSF13B]
7p12.3	rs80077929	chr7	46094089	C/T	I	1.7×10 ⁻¹⁵	1.111	[1.083,1.140]	IGFBP3---[]
4q24	rs230523	chr4	103462038	C/T	I	4.5×10 ⁻¹⁴	1.067	[1.050,1.086]	[NFKB1]
7p15.2	rs6668	chr7	27138183	C/T	I	1.7×10 ⁻¹³	1.065	[1.047,1.083]	HOXA1-[]-HOXA2
14q21.1	rs148131694	chr14	38025098	C/T	I	7.8×10 ⁻¹³	1.066	[1.047,1.084]	MIPOL1-[]--FOXA1
4q24	rs1391439	chr4	106151642	A/G	I	2.8×10 ⁻¹²	1.060	[1.043,1.077]	[TET2]
2p14	rs201473667	chr2	65562862	D/I	I	5.4×10 ⁻¹²	0.945	[0.930,0.960]	[SPRED2]
20q13.12	rs6032664	chr20	44739419	A/T	I	7.3×10 ⁻¹²	0.939	[0.922,0.956]	NCOA5--[]-CD40
7p12.2	rs876037	chr7	50308692	A/T	I	1.1×10 ⁻¹¹	1.061	[1.043,1.079]	C7orf72---[]--IKZF1
13q21.33	rs9542155	chr13	70579527	C/T	I	1.9×10 ⁻¹¹	1.060	[1.042,1.078]	[KLHL1]
7p22.2	rs2644312	chr7	2841164	A/G	I	2.7×10 ⁻¹¹	1.061	[1.043,1.080]	[GNA12]
3q21.2	rs1980080	chr3	124911841	C/T	I	3.2×10 ⁻¹¹	0.944	[0.929,0.960]	[SLC12A8]
16p11.2	rs141876325	chr16	28330968	D/I	I	4.7×10 ⁻¹¹	1.063	[1.044,1.083]	[SBK1]
1q41	rs12126292	chr1	222165909	G/T	I	7.2×10 ⁻¹¹	0.920	[0.897,0.943]	DUSP10---[]--HHIPL2
4p15.2	rs10939037	chr4	25114601	A/G	I	1.4×10 ⁻¹⁰	0.948	[0.933,0.964]	LG12--[]-SEPS2
12q24.12	rs3184504	chr12	111884608	C/T	I	3.1×10 ⁻¹⁰	1.052	[1.036,1.069]	[SH2B3]
4q21.1	rs7685785	chr4	78440425	C/T	I	4.5×10 ⁻¹⁰	0.919	[0.895,0.944]	[CXCL13]
7q31.2	rs2023703	chr7	114944919	A/C	I	7.8×10 ⁻¹⁰	1.055	[1.037,1.073]	MDFIC---[]--TFEC
19p13.2	rs2918308	chr19	8789912	A/C	I	1.1×10 ⁻⁹	0.934	[0.914,0.955]	ADAMTS10---[]--ACTL9
17p13.3	rs67968065	chr17	2729247	D/I	I	2.1×10 ⁻⁹	0.951	[0.936,0.967]	[RAP1GAP2]
21q22.11	rs200746495	chr21	35052995	D/I	I	4.4×10 ⁻⁹	0.930	[0.908,0.953]	[ITSN1]
2q33.3	rs1448903	chr2	207308961	A/G	I	5.1×10 ⁻⁹	1.085	[1.056,1.115]	[ADAM23]
1p36.23	rs12068123	chr1	8894346	A/G	I	5.6×10 ⁻⁹	1.049	[1.032,1.066]	RERE--[]--ENO1
11p15.4	rs11042055	chr11	8756856	A/G	I	7.0×10 ⁻⁹	0.954	[0.939,0.969]	[ST5]
20q13.33	rs41278232	chr20	62606617	A/G	I	7.5×10 ⁻⁹	1.078	[1.051,1.107]	[SAMD10]
9q34.2	rs635634	chr9	136155000	C/T	I	8.5×10 ⁻⁹	1.060	[1.039,1.081]	ABO-[]--SURF6
17q11.2	rs62066768	chr17	26858395	A/G	I	1.1×10 ⁻⁸	0.918	[0.892,0.946]	[FOXN1]
6q23.3	rs11757201	chr6	138003822	C/G	I	3.0×10 ⁻⁸	0.946	[0.928,0.965]	OLIG3---[]--TNFAIP3
6p21.31	rs201220830	chr6	34195176	D/I	I	3.6×10 ⁻⁸	0.898	[0.864,0.933]	GRM4--[]-HMGA1
16p11.2	rs12931792	chr16	30156398	A/G	I	4.1×10 ⁻⁸	1.057	[1.036,1.078]	MAPK3--[]--CORO1A
7q31.2	rs200608253	chr7	116917244	D/I	I	4.1×10 ⁻⁸	1.046	[1.029,1.063]	[WNT2]
22q11.21	rs41298830	chr22	19752943	A/C	I	5.0×10 ⁻⁸	0.947	[0.929,0.966]	[TBX1]

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
rs10849448	FALSE	FALSE	FALSE					0.9282	0.8265	0.30	0.7459	v2v3v4
rs201112509	FALSE	FALSE	FALSE					0.8998	0.8838	0.54	0.6935	v2v3v4
rs34557412	TRUE	FALSE	FALSE	0.9989	0.068	0.99	0.0048	0.7503	0.6731	9.3×10 ⁻¹⁹	0.0084	v2v3v4
rs41543314	FALSE	FALSE	TRUE	0.9921	0.0	1.1×10 ⁻⁹	0.0424	0.9403	0.8812	0.00035	0.0348	v2v3v4
rs200748895	FALSE	FALSE	FALSE					0.6785	0.5925	0.41	0.9659	v2v3v4
rs80077929	FALSE	FALSE	FALSE					0.8963	0.8625	0.33	0.1138	v2v3v4
rs230523	FALSE	FALSE	FALSE					0.9814	0.9796	0.17	0.6662	v2v3v4
rs6668	FALSE	TRUE	FALSE	0.9973	0.41	0.0093	0.3746	0.9586	0.8507	1.3×10 ⁻⁶	0.3719	v2v3v4
rs148131694	FALSE	FALSE	FALSE					0.8357	0.8296	0.048	0.4427	v2v3v4
rs1391439	FALSE	FALSE	FALSE					0.9991	0.9983	5.1×10 ⁻⁷	0.3980	v2v3v4
rs201473667	FALSE	FALSE	FALSE					0.9770	0.9667	0.91	0.5318	v2v3v4
rs6032664	FALSE	FALSE	FALSE					0.9996	0.9992	0.64	0.7400	v2v3v4
rs876037	FALSE	FALSE	FALSE					0.9974	0.9930	0.98	0.6789	v2v3v4
rs9542155	FALSE	FALSE	FALSE					0.9991	0.9941	0.42	0.3187	v2v3v4
rs2644312	FALSE	FALSE	FALSE					0.9949	0.9921	0.014	0.7107	v2v3v4
rs1980080	TRUE	TRUE	TRUE	0.9998	0.70	0.24	0.6715	0.9998	0.9996	0.57	0.6703	v2v3v4
rs141876325	FALSE	FALSE	FALSE					0.9325	0.7660	1.9×10 ⁻³³	0.2824	v2v3v4
rs12126292	FALSE	FALSE	FALSE					0.9064	0.8847	0.94	0.8767	v2v3v4
rs10939037	FALSE	FALSE	FALSE					0.9664	0.8961	2.8×10 ⁻⁶	0.4377	v2v3v4
rs3184504	TRUE	TRUE	TRUE	0.9998	2.4×10 ⁻⁵	0.00017	0.4996	0.9993	0.9985	0.30	0.4984	v2v3v4
rs7685785	FALSE	FALSE	FALSE					0.9704	0.8725	2.0×10 ⁻¹⁰	0.1051	v2v3v4
rs2023703	FALSE	FALSE	FALSE					0.9884	0.9779	0.0030	0.3254	v2v3v4
rs2918308	FALSE	FALSE	FALSE					0.9994	0.9936	0.83	0.1576	v2v3v4
rs67968065	FALSE	FALSE	FALSE					0.9500	0.9371	0.94	0.4760	v2v3v4
rs200746495	FALSE	FALSE	FALSE					0.9530	0.9479	0.33	0.1322	v2v3v4
rs1448903	TRUE	TRUE	TRUE	0.9992	0.15	0.067	0.0904	0.9993	0.9965	0.25	0.0911	v2v3v4
rs12068123	FALSE	FALSE	FALSE					0.9780	0.9695	3.8×10 ⁻⁹	0.4544	v2v3v4
rs11042055	TRUE	TRUE	TRUE	0.9983	1.6×10 ⁻⁷	0.53	0.5682	0.9999	0.9997	0.00059	0.5716	v2v3v4
rs41278232	FALSE	FALSE	FALSE					0.9714	0.8892	0.67	0.8888	v2v3v4
rs635634	FALSE	FALSE	FALSE					0.9997	0.9990	0.73	0.1976	v2v3v4
rs62066768	FALSE	FALSE	FALSE					0.9621	0.9290	0.13	0.9178	v2v3v4
rs11757201	FALSE	FALSE	FALSE					0.9991	0.9980	0.011	0.7962	v2v3v4
rs201220830	FALSE	FALSE	FALSE					0.9026	0.7062	0.30	0.9522	v2v3v4
rs12931792	FALSE	FALSE	FALSE					0.8560	0.8504	0.00010	0.5423	v3
rs200608253	FALSE	FALSE	FALSE					0.9823	0.9279	0.31	0.5247	v2v3v4
rs41298830	FALSE	FALSE	FALSE					0.9368	0.9052	0.031	0.2309	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
rs10849448				113323	0.7547				60098	0.7347
rs201112509				113323	0.6909				60098	0.7083
rs34557412	11833	93	0	113323	0.0076	7255	92	3	60098	0.0103
rs41543314	13296	627	240	113323	0.0332	7459	418	193	60098	0.0397
rs200748895				113323	0.9682				60098	0.9634
rs80077929				113323	0.1117				60098	0.1214
rs230523				113323	0.6608				60098	0.6720
rs6668	35564	41202	12100	113323	0.3662	17509	21558	6779	60098	0.3797
rs148131694				113323	0.4393				60098	0.4511
rs1391439				113323	0.3884				60098	0.3997
rs201473667				113323	0.5337				60098	0.5215
rs6032664				113323	0.7457				60098	0.7356
rs876037				113323	0.6766				60098	0.6884
rs9542155				113323	0.3143				60098	0.3235
rs2644312				113323	0.7043				60098	0.7155
rs1980080	11842	49717	51749	113323	0.6761	6677	26722	26691	60098	0.6665
rs141876325				113323	0.2800				60098	0.2889
rs12126292				113323	0.8793				60098	0.8731
rs10939037				113323	0.4455				60098	0.4337
rs3184504	29079	56319	27910	113323	0.4951	14663	29853	15565	60098	0.5078
rs7685785				113323	0.1065				60098	0.1001
rs2023703				113323	0.3204				60098	0.3306
rs2918308				113323	0.1604				60098	0.1527
rs67968065				113323	0.4798				60098	0.4704
rs200746495				113323	0.1346				60098	0.1284
rs1448903	93967	18371	888	113323	0.0892	49100	10361	564	60098	0.0960
rs12068123				113323	0.4445				60098	0.4570
rs11042055	21368	55011	36764	113323	0.5679	11828	29369	18803	60098	0.5580
rs41278232				113323	0.8861				60098	0.8929
rs635634				113323	0.1957				60098	0.2036
rs62066768				113323	0.9193				60098	0.9139
rs11757201				113323	0.7968				60098	0.7887
rs201220830				113323	0.9533				60098	0.9491
rs12931792				88447	0.5410				45608	0.5503
rs200608253				113323	0.5210				60098	0.5296
rs41298830				113323	0.2315				60098	0.2238

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
22q12.2	30324654	rs201112509	3.4×10^{-34}	12932	0.785	rs36600	6.0×10^{-13}	21725308	Lung cancer	MTMR3
22q12.2	30324654	rs201112509	3.4×10^{-34}	98806	0.589	rs12537	1.0×10^{-11}	22197929	IgA nephropathy	MTMR3
6p21.33	31322690	rs41543314	5.4×10^{-21}	109090	0.948	rs2395029	1.0×10^{-25}	21051598	HIV-1 control	HLA-B
6p21.33	31322690	rs41543314	5.4×10^{-21}	109090	0.948	rs2395029	5.0×10^{-35}	20041166	HIV-1 control	HCP5, HLA-B
6p21.33	31322690	rs41543314	5.4×10^{-21}	109090	0.948	rs2395029	9.0×10^{-33}	19483685	Drug-induced liver injury (flucloxacillin)	HCP5, HLA-B
6p21.33	31322690	rs41543314	5.4×10^{-21}	109090	0.948	rs2395029	3.0×10^{-19}	19115949	AIDS progression	HCP5, MICB, ...
6p21.33	31322690	rs41543314	5.4×10^{-21}	109090	0.948	rs2395029	2.0×10^{-26}	18369459	Psoriasis	HLA-C
4q24	103462038	rs230523	4.5×10^{-14}	-4620	0.740	rs230529	2.0×10^{-7}	22479419	Schizophrenia (treatment refractory)	NFKB1
4q24	103462038	rs230523	4.5×10^{-14}	49076	0.912	rs3774959	4.0×10^{-12}	23128233	Ulcerative colitis	NFKB1,MANBA
4q24	106151642	rs1391439	2.8×10^{-12}	-90108	0.854	rs7679673	3.0×10^{-14}	19767753	Prostate cancer	TET2
2p14	65562862	rs201473667	5.4×10^{-12}	32724	0.903	rs934734	5.0×10^{-10}	20453842	Rheumatoid arthritis	SPRED2
2p14	65562862	rs201473667	5.4×10^{-12}	46047	0.738	rs1876518	2.0×10^{-8}	21383967	Celiac disease and Rheumatoid arthritis	SPRED2
20q13.12	44739419	rs6032664	7.3×10^{-12}	-37299	0.679	rs2425752	5.0×10^{-10}	21833088	Multiple sclerosis	NCOA5, CD40
20q13.12	44739419	rs6032664	7.3×10^{-12}	777	0.973	rs6074022	5.0×10^{-6}	22190364	Multiple sclerosis	CD40
20q13.12	44739419	rs6032664	7.3×10^{-12}	777	0.973	rs6074022	1.0×10^{-7}	19525955	Multiple sclerosis	CD40
20q13.12	44739419	rs6032664	7.3×10^{-12}	2645	1.000	rs1569723	1.0×10^{-13}	23128233	Inflammatory bowel disease	CD40,MMP9,PLTP
20q13.12	44739419	rs6032664	7.3×10^{-12}	2645	1.000	rs1569723	6.0×10^{-9}	22446961	Kawasaki disease	CD40
20q13.12	44739419	rs6032664	7.3×10^{-12}	8528	0.993	rs4810485	3.0×10^{-9}	20453842	Rheumatoid arthritis	CD40
20q13.12	44739419	rs6032664	7.3×10^{-12}	8528	0.993	rs4810485	8.0×10^{-9}	18794853	Rheumatoid arthritis	CD40
7p12.2	50308692	rs876037	1.1×10^{-11}	-2829	0.994	rs4917014	3.0×10^{-23}	19838193	Systemic lupus erythematosus	IKZF1
7p22.2	2841164	rs2644312	2.7×10^{-11}	-81369	0.832	rs798554	4.0×10^{-23}	23563607	Height	AMZ1
7p22.2	2841164	rs2644312	2.7×10^{-11}	-78062	0.825	rs798544	7.0×10^{-15}	18391951	Height	GNA12
7p22.2	2841164	rs2644312	2.7×10^{-11}	-51284	0.921	rs798502	6.0×10^{-17}	23128233	Ulcerative colitis	CARD11,GNA12,TTYH3
7p22.2	2841164	rs2644312	2.7×10^{-11}	-51284	0.921	rs798502	3.0×10^{-15}	21297633	Ulcerative colitis	GNA12
7p22.2	2841164	rs2644312	2.7×10^{-11}	-45207	0.951	rs798497	7.0×10^{-7}	20189936	Height	GNA12
7p22.2	2841164	rs2644312	2.7×10^{-11}	-39361	0.880	rs798489	2.0×10^{-33}	20881960	Height	GNA12
7p22.2	2841164	rs2644312	2.7×10^{-11}	28821	0.975	rs1182188	3.0×10^{-9}	19343178	Height	GNA12
12q24.12	111884608	rs3184504	3.1×10^{-10}	-176150	0.575	rs1265564	1.0×10^{-16}	22293688	Type 1 diabetes	CUX2
12q24.12	111884608	rs3184504	3.1×10^{-10}	0	1.000	rs3184504	5.0×10^{-11}	24026423	Platelet counts	SH2B3, ATXN2
12q24.12	111884608	rs3184504	3.1×10^{-10}	0	1.000	rs3184504	3.0×10^{-8}	23417110	Beta-2 microglubulin plasma levels	SH2B3, ATXN2
12q24.12	111884608	rs3184504	3.1×10^{-10}	0	1.000	rs3184504	4.0×10^{-19}	23222517	Red blood cell traits	SH2B3, ATXN2
12q24.12	111884608	rs3184504	3.1×10^{-10}	0	1.000	rs3184504	3.0×10^{-12}	22493691	Hypothyroidism	SH2B3, ATXN2, ...
12q24.12	111884608	rs3184504	3.1×10^{-10}	0	1.000	rs3184504	1.0×10^{-26}	22139419	Platelet counts	SH2B3
12q24.12	111884608	rs3184504	3.1×10^{-10}	0	1.000	rs3184504	4.0×10^{-25}	21909115	Diastolic blood pressure	SH2B3
12q24.12	111884608	rs3184504	3.1×10^{-10}	0	1.000	rs3184504	2.0×10^{-38}	21829393	Type 1 diabetes autoantibodies	SH2B3
12q24.12	111884608	rs3184504	3.1×10^{-10}	0	1.000	rs3184504	6.0×10^{-6}	21378990	Coronary heart disease	SH2B3

12q24.12	111884608	rs3184504	3.1×10 ⁻¹⁰	0	1.000	rs3184504	6.0×10 ⁻⁶	20453842	Rheumatoid arthritis	SH2B3
12q24.12	111884608	rs3184504	3.1×10 ⁻¹⁰	0	1.000	rs3184504	3.0×10 ⁻²⁷	19430480	Type 1 diabetes	SH2B3
12q24.12	111884608	rs3184504	3.1×10 ⁻¹⁰	0	1.000	rs3184504	5.0×10 ⁻⁹	19430479	Systolic blood pressure	SH2B3
12q24.12	111884608	rs3184504	3.1×10 ⁻¹⁰	0	1.000	rs3184504	3.0×10 ⁻¹⁴	19430479	Diastolic blood pressure	SH2B3
12q24.12	111884608	rs3184504	3.1×10 ⁻¹⁰	0	1.000	rs3184504	7.0×10 ⁻¹⁹	19198610	Eosinophil counts	SH2B3
12q24.12	111884608	rs3184504	3.1×10 ⁻¹⁰	19763	0.924	rs4766578	4.0×10 ⁻¹⁸	22561518	Vitiligo	SH2B3
12q24.12	111884608	rs3184504	3.1×10 ⁻¹⁰	25611	0.934	rs10774625	2.0×10 ⁻¹³	21060863	Retinal vascular caliber	ATXN2,PTPN11,SH2B3
12q24.12	111884608	rs3184504	3.1×10 ⁻¹⁰	123148	0.979	rs653178	7.0×10 ⁻¹²	23263486	Urate levels	ATXN2, PTPN11
12q24.12	111884608	rs3184504	3.1×10 ⁻¹⁰	123148	0.979	rs653178	7.0×10 ⁻²⁰	21909110	Blood pressure	ATXN2
12q24.12	111884608	rs3184504	3.1×10 ⁻¹⁰	123148	0.979	rs653178	3.0×10 ⁻¹⁹	21383967	Celiac disease and Rheumatoid arthritis	SH2B3
12q24.12	111884608	rs3184504	3.1×10 ⁻¹⁰	123148	0.979	rs653178	4.0×10 ⁻¹¹	20383146	Chronic kidney disease	ATXN2
12q24.12	111884608	rs3184504	3.1×10 ⁻¹⁰	123148	0.979	rs653178	7.0×10 ⁻²¹	20190752	Celiac disease	SH2B3
12q24.12	111884608	rs3184504	3.1×10 ⁻¹⁰	123148	0.979	rs653178	3.0×10 ⁻¹⁸	19430483	Diastolic blood pressure	ATXN2, SH2B3
12q24.12	111884608	rs3184504	3.1×10 ⁻¹⁰	123148	0.979	rs653178	8.0×10 ⁻⁸	18311140	Celiac disease	SH2B3, ATXN2
12q24.12	111884608	rs3184504	3.1×10 ⁻¹⁰	187816	0.808	rs11065987	8.0×10 ⁻¹¹	23297363	Tetralogy of Fallot	PTPN11
12q24.12	111884608	rs3184504	3.1×10 ⁻¹⁰	187816	0.808	rs11065987	2.0×10 ⁻⁹	20686565	LDL cholesterol	BRAP
12q24.12	111884608	rs3184504	3.1×10 ⁻¹⁰	187816	0.808	rs11065987	7.0×10 ⁻¹²	20686565	Cholesterol, total	BRAP
12q24.12	111884608	rs3184504	3.1×10 ⁻¹⁰	187816	0.808	rs11065987	1.0×10 ⁻¹¹	19862010	Hemoglobin	TRAFD1
12q24.12	111884608	rs3184504	3.1×10 ⁻¹⁰	187816	0.808	rs11065987	1.0×10 ⁻¹²	19862010	Hematocrit	SH2B3, ATXN2
19p13.2	8789912	rs2918308	1.1×10 ⁻⁹	-531	1.000	rs2164983	7.0×10 ⁻⁹	22197932	Atopic dermatitis	ACTL9
1p36.23	8894346	rs12068123	5.6×10 ⁻⁹	-368204	0.559	rs4908760	7.0×10 ⁻¹⁵	20410501	Vitiligo	RERE
9q34.2	136155000	rs635634	8.5×10 ⁻⁹	-5601	0.991	rs507666	3.0×10 ⁻⁹¹	21533024	Soluble ICAM-1	ABO
9q34.2	136155000	rs635634	8.5×10 ⁻⁹	-5601	0.991	rs507666	5.0×10 ⁻²⁹	18604267	Soluble ICAM-1	ABO
9q34.2	136155000	rs635634	8.5×10 ⁻⁹	-1125	0.841	rs651007	2.0×10 ⁻²⁵	23381943	End-stage coagulation	ABO
9q34.2	136155000	rs635634	8.5×10 ⁻⁹	-1125	0.841	rs651007	6.0×10 ⁻⁹	21909109	Metabolite levels	ABO
9q34.2	136155000	rs635634	8.5×10 ⁻⁹	-1125	0.841	rs651007	2.0×10 ⁻⁸²	20147318	E-selectin levels	ABO
9q34.2	136155000	rs635634	8.5×10 ⁻⁹	-832	0.841	rs579459	9.0×10 ⁻¹⁸	23222517	Red blood cell traits	ABO
9q34.2	136155000	rs635634	8.5×10 ⁻⁹	-832	0.841	rs579459	3.0×10 ⁻¹²³	22001757	Liver enzyme levels (alkaline phosphatase)	ABO
9q34.2	136155000	rs635634	8.5×10 ⁻⁹	-832	0.841	rs579459	4.0×10 ⁻¹⁴	21378990	Coronary heart disease	ABO
9q34.2	136155000	rs635634	8.5×10 ⁻⁹	-832	0.841	rs579459	2.0×10 ⁻⁴¹	20167578	Soluble levels of adhesion molecules	ABO
9q34.2	136155000	rs635634	8.5×10 ⁻⁹	-832	0.841	rs579459	1.0×10 ⁻²⁹	19729612	Soluble E-selectin levels	ABO
9q34.2	136155000	rs635634	8.5×10 ⁻⁹	-133	0.841	rs495828	3.0×10 ⁻¹⁶	22672568	Venous thromboembolism	ABO
9q34.2	136155000	rs635634	8.5×10 ⁻⁹	-133	0.841	rs495828	3.0×10 ⁻¹²	20139978	Red blood cell count	ABO
9q34.2	136155000	rs635634	8.5×10 ⁻⁹	-133	0.841	rs495828	6.0×10 ⁻¹⁰	20139978	Hematological and biochemical traits	ABO
9q34.2	136155000	rs635634	8.5×10 ⁻⁹	-133	0.841	rs495828	3.0×10 ⁻⁸	20066004	Angiotensin-converting enzyme activity	ABO
9q34.2	136155000	rs635634	8.5×10 ⁻⁹	0	1.000	rs635634	8.0×10 ⁻²²	20686565	LDL cholesterol	ABO
9q34.2	136155000	rs635634	8.5×10 ⁻⁹	0	1.000	rs635634	9.0×10 ⁻²¹	20686565	Cholesterol, total	ABO
6q23.3	138003822	rs11757201	3.0×10 ⁻⁸	-30754	0.963	rs2327832	4.0×10 ⁻¹⁹	20190752	Celiac disease	TNFAIP3
6q23.3	138003822	rs11757201	3.0×10 ⁻⁸	2682	1.000	rs6920220	1.0×10 ⁻²¹	23128233	Inflammatory bowel disease	TNFAIP3
6q23.3	138003822	rs11757201	3.0×10 ⁻⁸	2682	1.000	rs6920220	8.0×10 ⁻¹⁷	21297633	Ulcerative colitis	Intergenic
6q23.3	138003822	rs11757201	3.0×10 ⁻⁸	2682	1.000	rs6920220	9.0×10 ⁻¹³	20453842	Rheumatoid arthritis	TNFAIP3
6q23.3	138003822	rs11757201	3.0×10 ⁻⁸	2682	1.000	rs6920220	2.0×10 ⁻⁹	18794853	Rheumatoid arthritis	OLIG3, TNFIP3
6q23.3	138003822	rs11757201	3.0×10 ⁻⁸	2682	1.000	rs6920220	1.0×10 ⁻⁷	17982456	Rheumatoid arthritis	TNFAIP3, OLIG3
7q31.2	116917244	rs200608253	4.1×10 ⁻⁸	-24398	0.503	rs38904	1.0×10 ⁻⁸	23128233	Inflammatory bowel disease	Intergenic
7q31.2	116917244	rs200608253	4.1×10 ⁻⁸	-126	0.774	rs4730775	3.0×10 ⁻⁸	21732829	Dupuytren's disease	WNT2

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
17p11.2	16852187	rs34557412	2.7×10 ⁻²¹	0	1.000	rs34557412	TNFRSF13B	C104R
6p21.33	31322690	rs41543314	5.4×10 ⁻²¹	56174	0.974	rs41558312	MICA	Q114R
6p21.33	31322690	rs41543314	5.4×10 ⁻²¹	57312	0.923	rs199503730	MICA	V298C
6p21.33	31322690	rs41543314	5.4×10 ⁻²¹	152130	0.784	rs41293883	MICB	T212I
14q21.1	38025098	rs148131694	7.8×10 ⁻¹³	36644	0.649	rs7144658	FOXA1	A83T
12q24.12	111884608	rs3184504	3.1×10 ⁻¹⁰	0	1.000	rs3184504	SH2B3	W262R
11p15.4	8756856	rs11042055	7.0×10 ⁻⁹	-4967	0.963	rs3794153	ST5	K316N
17q11.2	26858395	rs62066768	1.1×10 ⁻⁸	-6793	0.972	rs2071587	FOXP1	R69C
22q11.21	19752943	rs41298830	5.0×10 ⁻⁸	1148	0.915	rs72646967	TBX1	N397H

Nearby Expression QTLs

region	position	our.name	our.pval	dist	rsqr	assay.name	eqtl.dist	eqtl.gene	eqtl.pval	eqtl.rsqr	tissue	pubmed.id
12p13.31	6493351	rs10849448	2.3×10 ⁻³⁵	-122985	0.792	rs12354	140619	NCAPD2	9.9×10 ⁻⁵		Lymphoblastoid	20220756
12p13.31	6493351	rs10849448	2.3×10 ⁻³⁵	-119304	0.854	rs9669611	-3344	LTBR	2.2×10 ⁻¹³	0.228	Lymphoblastoid	17873874
12p13.31	6493351	rs10849448	2.3×10 ⁻³⁵	-702	0.784	rs11064157	7826	LTBR	1.4×10 ⁻⁷	0.095	Monocyte	22446964
12p13.31	6493351	rs10849448	2.3×10 ⁻³⁵	0	1.000	rs10849448	9140	LTBR	3.5×10 ⁻³⁴	0.331	Lymphoblastoid	24037378

12p13.31	6493351	rs10849448	2.3×10 ⁻³⁵	1924	0.891	rs2364480	1941	LTBR	2.7×10 ⁻⁵	0.215	Lymphoblastoid	19644074
12p13.31	6493351	rs10849448	2.3×10 ⁻³⁵	1924	0.891	rs2364480	1941	LTBR	1.0×10 ⁻⁵	0.236	Fibroblast	19644074
12p13.31	6493351	rs10849448	2.3×10 ⁻³⁵	1924	0.891	rs2364480	384634	PTMS	0.00045	0.043	Monocyte	22446964
22q12.2	30324654	rs201112509	3.4×10 ⁻³⁴	-155218	0.600	rs131291	978891	XBP1	0.00044	0.157	T-cell	19644074
22q12.2	30324654	rs201112509	3.4×10 ⁻³⁴	179553	0.743	rs718772	-338257	UCRC	0.00038	0.044	Monocyte	22446964
22q12.2	30324654	rs201112509	3.4×10 ⁻³⁴	230562	0.736	rs9625933	103605	OSM	4.5×10 ⁻⁵	0.205	T-cell	19644074
6p21.33	31322690	rs41543314	5.4×10 ⁻²¹	188660	0.531	rs9267487	32449	MICB	1.9×10 ⁻²⁸	0.079	Monocyte	20502693
6p21.33	31322690	rs41543314	5.4×10 ⁻²¹	188660	0.531	rs9267487	284338	HSPA1B	1.2×10 ⁻⁶	0.016	Monocyte	20502693
6p21.33	31322690	rs41543314	5.4×10 ⁻²¹	221068	0.739	rs3093661	-64948	MICB	2.8×10 ⁻¹⁴	0.187	Monocyte	22446964
6p21.33	31322690	rs41543314	5.4×10 ⁻²¹	221068	0.739	rs3093661	-64948	MICB	5.9×10 ⁻³³	0.402	B-Cell	22446964
6p21.33	31322690	rs41543314	5.4×10 ⁻²¹	221068	0.739	rs3093661	-438256	PSORS1C2	2.1×10 ⁻⁵	0.063	Monocyte	22446964
4q24	103462038	rs230523	4.5×10 ⁻¹⁴	-18469	0.728	rs1599961	94537	NFKB1	0.00013	0.051	Monocyte	22446964
4q24	106151642	rs1391439	2.8×10 ⁻¹²	219449	1.000	rs1391439	740728	NPNT	7.2×10 ⁻¹⁴	0.234	Lymphoblastoid	17873874
20q13.12	44739419	rs6032664	7.3×10 ⁻¹²	777	0.973	rs6074022	-253602	ZSWIM3	0.0019	0.034	Monocyte	22446964
7p22.2	2841164	rs2644312	2.7×10 ⁻¹¹	-26529	0.506	rs7807978	0	GNA12	1.1×10 ⁻¹⁰	0.031	Monocyte	20502693
7p22.2	2841164	rs2644312	2.7×10 ⁻¹¹	-15577	0.607	rs2644296	-585133	FTSJ2	9.3×10 ⁻⁶		Lymphoblastoid	20220756
7p22.2	2841164	rs2644312	2.7×10 ⁻¹¹	-1391	0.957	rs1182181	-38685	GNA12	1.1×10 ⁻⁵		Lymphoblastoid	20220756
7p22.2	2841164	rs2644312	2.7×10 ⁻¹¹	28821	0.975	rs1182188	-102078	GNA12	2.8×10 ⁻²³	0.299	B-Cell	22446964
7p22.2	2841164	rs2644312	2.7×10 ⁻¹¹	31972	0.957	rs1182183	110514	AC006028.9	3.6×10 ⁻¹⁴	0.144	Lymphoblastoid	24037378
4p15.2	25114601	rs10939037	1.4×10 ⁻¹⁰	53816	0.647	rs13139513	251516	ANAPC4	1.1×10 ⁻⁵	0.067	B-Cell	22446964
21q22.11	35052995	rs200746495	4.4×10 ⁻⁹	-105139	0.588	rs9978525	-138221	IFNGR2	3.4×10 ⁻⁹	0.118	Monocyte	22446964
21q22.11	35052995	rs200746495	4.4×10 ⁻⁹	-105139	0.588	rs9978525	-136280	TMEM50B	9.6×10 ⁻⁸	0.097	Monocyte	22446964
1p36.23	8894346	rs12068123	5.6×10 ⁻⁹	-282242	0.573	rs6577499	0	RERE	1.5×10 ⁻¹⁰	0.030	Monocyte	20502693
1p36.23	8894346	rs12068123	5.6×10 ⁻⁹	-235634	0.593	rs1318218	-245857	RERE	8.0×10 ⁻⁶	0.069	Monocyte	22446964
11p15.4	8756856	rs11042055	7.0×10 ⁻⁹	58597	0.897	rs11042067	117045	ST5	1.2×10 ⁻⁷	0.073	Lymphoblastoid	24037378
11p15.4	8756856	rs11042055	7.0×10 ⁻⁹	119996	0.528	rs2742552	64296	C11ORF17	3.6×10 ⁻¹⁵	0.199	B-Cell	22446964
11p15.4	8756856	rs11042055	7.0×10 ⁻⁹	256783	0.574	rs7930026	-72126	C11ORF17	6.7×10 ⁻⁷	0.085	Monocyte	22446964
20q13.33	62606617	rs41278232	7.5×10 ⁻⁹	1003	0.593	rs817344	3741	SAMD10	4.6×10 ⁻¹⁵	0.153	Lymphoblastoid	24037378
20q13.33	62606617	rs41278232	7.5×10 ⁻⁹	24953	0.938	rs112790215	34127	LINC00176	1.1×10 ⁻¹⁰	0.106	Lymphoblastoid	24037378
16p11.2	30156398	rs12931792	4.1×10 ⁻⁸	-101632	0.571	rs7201780	218660	CD2BP2	0.00045		Lymphoblastoid	20220756
16p11.2	30156398	rs12931792	4.1×10 ⁻⁸	9327	0.693	rs4787495	40548	GDPD3	1.1×10 ⁻¹⁰	0.030	Monocyte	20502693
16p11.2	30156398	rs12931792	4.1×10 ⁻⁸	41753	0.705	rs1132812	-78602	GDPD3	6.3×10 ⁻⁵	0.056	B-Cell	22446964
16p11.2	30156398	rs12931792	4.1×10 ⁻⁸	163909	0.565	rs4471699	185677	MAPK3	7.9×10 ⁻¹⁹	0.055	Monocyte	20502693
16p11.2	30156398	rs12931792	4.1×10 ⁻⁸	183355	0.521	rs77948921	873781	RP11-345J4.5	4.7×10 ⁻¹²	0.121	Lymphoblastoid	24037378
16p11.2	30156398	rs12931792	4.1×10 ⁻⁸	184472	0.573	rs7404371	925520	RP11-231C14.2	5.2×10 ⁻⁷	0.066	Lymphoblastoid	24037378

Nearby Clinical Variants

source	region	our.name	our.pval	dist	rsqr	assay.name	gene	phenotype	accession
clinvar	17p11.2	rs34557412	2.7×10 ⁻²¹	0	1.000	rs34557412	TNFRSF13B	Immunoglobulin A deficiency 2	Office of Rare Diseases10198
clinvar	17p11.2	rs34557412	2.7×10 ⁻²¹	0	1.000	rs34557412	TNFRSF13B	Common variable immunodeficiency 2	NCBI curation

Regional Association Plots

