Analysis Summary: Breast Size

Phenotype Description

Quantitative trait looking at breast size (using bra cup size as a proxy) as defined by the following question:

- Physical Features ("What is your bra size? (Please report your bra size prior to any surgeries that affected that size of your breasts and prior to any pregnancies.)"

Respondents with a bra band size greater than 70 inches 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]
cup_size	[0,4]	12995	0	12995	2137	3884	3439	3535
	(4,5]	9408	0	9408	1208	2558	2660	2982
	(5,6]	5591	0	5591	753	1442	1590	1806
	(6,9]	5796	0	5796	765	1857	1635	1539

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

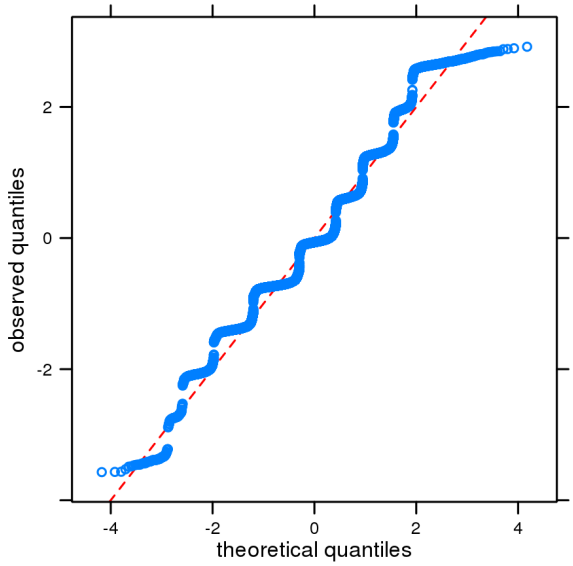
Phenotype	Group	Total	v1/v2	v3	v4
cup_size	[0,4]	12995	1600	10778	617
	(4,5]	9408	1113	7848	447
	(5,6]	5591	652	4635	304
	(6,9]	5796	618	4809	369

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	0.00121	0.000517	2.3	0.020
pc.0	−0.03893	0.008141	−4.8	1.8×10^{-6}
pc.1	−0.06898	0.008176	−8.4	3.4×10^{-17}
pc.2	−0.05233	0.008173	−6.4	1.5×10^{-10}
pc.3	−0.01672	0.008140	−2.1	0.040
pc.4	0.01108	0.008143	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	−23179	13755888
MAF < 0.1%	−1716401	12039487

Genetic Association Tests

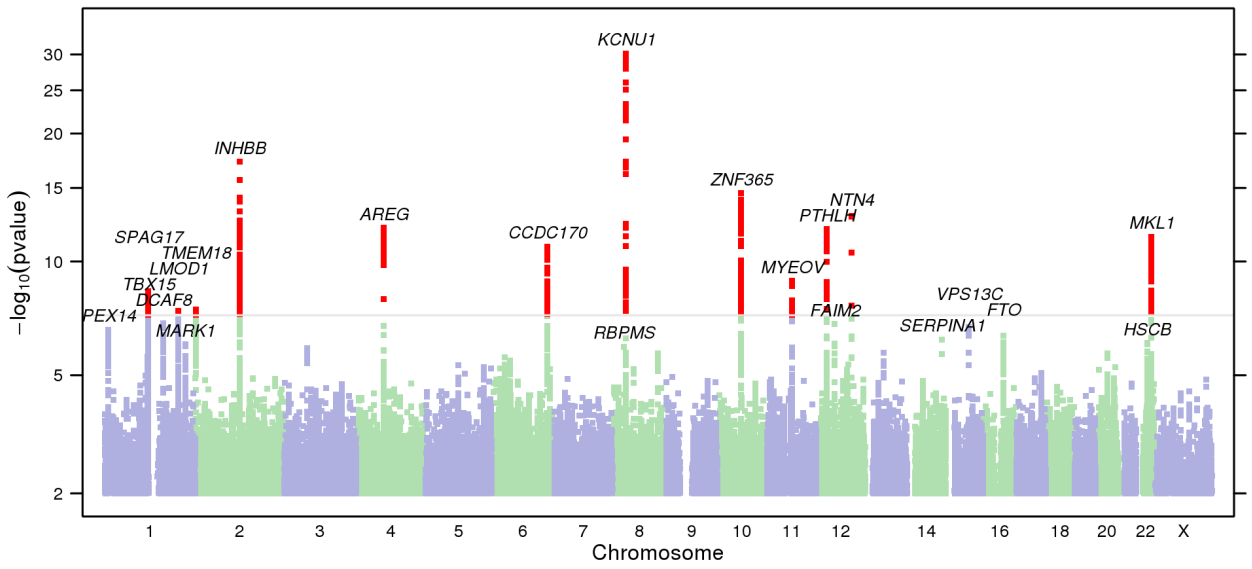
We performed linear regression assuming an additive model for allelic effects, using the model:

cup_size ~ age + pc.0 + pc.1 + pc.2 + pc.3 + pc.4 + genotype

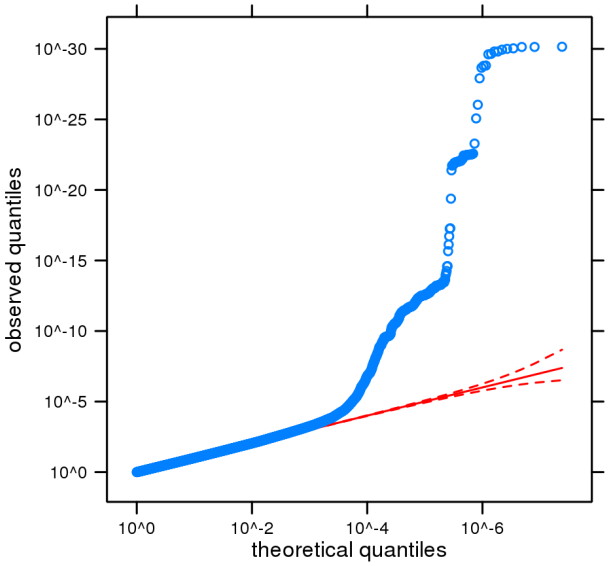
This genome-wide association analysis includes data from 33790 individuals of European ancestry, filtered to remove close relatives.

The results in this report have been adjusted for a genomic control inflation factor λ=1.085. The equivalent inflation factor rescaled for a sample size of 2000 would be λ_{2000}= 1.005 , and for 20000, λ_{20000}= 1.051 .

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
8p11.23	rs10110651	chr8	36847115	C/T	I	7.1×10^{-31}	0.177	[0.147,0.207]	KCNU1--[]---ZNF703
2q14.2	rs17625845	chr2	121089731	C/T	I	5.6×10^{-18}	−0.140	[-0.171,-0.108]	RALB--[]--INHBB
10q21.2	rs3081227	chr10	64178657	D/I	I	2.5×10^{-15}	−0.103	[-0.129,-0.078]	[ZNF365]
12q22	rs17356907	chr12	96027759	A/G	I	1.3×10^{-13}	0.097	[0.071,0.123]	USP44--[]--NTN4
4q13.3	rs7659874	chr4	75547013	A/G	I	8.6×10^{-13}	−0.105	[-0.134,-0.076]	AREG--[]---BTC
12p11.22	rs1838564	chr12	28154895	A/G	I	1.0×10^{-12}	0.136	[0.099,0.174]	PTHLH--[]---CCDC91
22q13.1	rs5995875	chr22	40960692	C/T	I	3.2×10^{-12}	−0.137	[-0.176,-0.099]	[MKL1]
6q25.1	rs9397437	chr6	151952332	A/G	I	1.3×10^{-11}	−0.158	[-0.204,-0.112]	CCDC170--[]---ESR1
11q13.3	rs7102705	chr11	69143284	A/G	I	1.1×10^{-9}	−0.089	[-0.118,-0.061]	MYEOV--[]---CCND1
1p12	rs2359714	chr1	118770271	C/T	I	3.8×10^{-9}	0.071	[0.047,0.095]	SPAG17--[]---TBX15
2p25.3	rs62105303	chr2	632922	A/G	I	2.8×10^{-8}	−0.086	[-0.117,-0.056]	FAM150B---[]--TMEM18
1q32.1	rs34091558	chr1	201886769	D/I	I	3.1×10^{-8}	−0.072	[-0.098,-0.047]	[LMOD1]
1p12	rs2645288	chr1	119500483	C/G	I	1.1×10^{-7}	−0.077	[-0.105,-0.049]	[TBX15]
1q23.2	rs6427508	chr1	160238857	C/T	I	1.1×10^{-7}	−0.067	[-0.092,-0.042]	DCAF8-[]--COPA
12q13.12	rs3205718	chr12	50261809	C/T	I	1.2×10^{-7}	0.067	[0.042,0.092]	[FAIM2]
15q22.2	rs148713408	chr15	62220772	C/T	I	1.6×10^{-7}	0.339	[0.212,0.466]	[VPS13C]
1p36.22	rs616402	chr1	10566272	C/T	I	2.0×10^{-7}	−0.067	[-0.093,-0.042]	[PEX14]
16q12.2	rs62033406	chr16	53824226	A/G	I	3.7×10^{-7}	0.062	[0.038,0.087]	[FTO]
14q32.13	rs28929474	chr14	94844947	C/T	I	5.1×10^{-7}	−0.215	[-0.299,-0.131]	[SERPINA1]
22q12.1	rs5752793	chr22	29160314	A/G	I	6.9×10^{-7}	0.065	[0.039,0.090]	HSCB-[]-CCDC117

1q41	rs200550907	chr1	220701165	D/I	I	7.9×10^{-7}	−0.077	[-0.107,-0.046]	RAB3GAP2---[]MARK1
8p12	rs113198678	chr8	30241618	C/T	I	9.7×10^{-7}	−0.120	[-0.169,-0.072]	DCTN6---[]RBPMS

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
rs10110651	FALSE	FALSE	FALSE					0.9989	0.9962	0.055	0.8062	v2v3v4
rs17625845	FALSE	TRUE	TRUE	0.9981	0.071	0.21	0.7998	0.9723	0.7026	1.6×10^{-50}	0.8018	v3v4
rs3081227	FALSE	FALSE	FALSE					0.9143	0.9033	0.047	0.6335	v2v3v4
rs17356907	TRUE	TRUE	TRUE	0.9999	0.65	0.39	0.2962	0.9977	0.9959	0.77	0.2959	v2v3v4
rs7659874	TRUE	TRUE	TRUE	0.9996	0.047	0.46	0.2111	0.9989	0.9973	0.56	0.2108	v2v3v4
rs1838564	FALSE	FALSE	FALSE					0.9946	0.9746	0.038	0.8912	v2v3v4
rs5995875	FALSE	FALSE	FALSE					0.9977	0.9951	0.72	0.1047	v2v3v4
rs9397437	FALSE	FALSE	FALSE					0.9985	0.9865	0.018	0.9276	v2v3v4
rs7102705	TRUE	TRUE	TRUE	0.9956	0.010	0.31	0.7921	0.9998	0.9989	0.010	0.7918	v2v3v4
rs2359714	FALSE	FALSE	FALSE					0.9902	0.9768	0.19	0.5057	v2v3v4
rs62105303	FALSE	FALSE	FALSE					0.9829	0.9721	0.21	0.1845	v2v3v4
rs34091558	FALSE	FALSE	FALSE					0.9811	0.9695	2.9×10^{-5}	0.6952	v2v3v4
rs2645288	FALSE	FALSE	FALSE					0.9161	0.9025	0.0078	0.2510	v2v3v4
rs6427508	FALSE	FALSE	FALSE					0.9106	0.9006	0.11	0.4723	v2v3v4
rs3205718	FALSE	FALSE	FALSE					0.9544	0.9272	4.4×10^{-5}	0.3818	v2v3v4
rs148713408	FALSE	FALSE	FALSE					0.6675	0.6126	0.012	0.0125	v2v3v4
rs616402	FALSE	FALSE	FALSE					0.9992	0.9982	0.19	0.3061	v2v3v4
rs62033406	FALSE	FALSE	FALSE					0.9910	0.9897	0.68	0.4166	v2v3v4
rs28929474	TRUE	TRUE	TRUE	0.9999	0.26	0.0027	0.0177	0.9960	0.9933	0.12	0.0176	v2v3v4
rs5752793	FALSE	TRUE	FALSE	0.9989	2.2×10^{-149}	0.023	0.3008	0.9838	0.9681	1.1×10^{-7}	0.3048	v2v3v4
rs200550907	FALSE	FALSE	FALSE					0.9236	0.9169	0.058	0.2076	v2v3v4
rs113198678	FALSE	FALSE	FALSE					0.7897	0.7525	0.0026	0.0797	v2v3v4

SNP Statistics in the GWAS Sample

assay.name	AA.0	AB.0	BB.0	im.num.0	dose.b.0
rs10110651				33790	0.8111
rs17625845	1179	9451	19141	29530	0.8015
rs3081227				33790	0.6352
rs17356907	16678	14098	3010	33790	0.2981
rs7659874	20998	11283	1499	33790	0.2113
rs1838564				33790	0.8894
rs5995875				33790	0.1036
rs9397437				33790	0.9284
rs7102705	1502	11095	20971	33790	0.7896
rs2359714				33790	0.5133
rs62105303				33790	0.1850
rs34091558				33790	0.6874
rs2645288				33790	0.2493
rs6427508				33790	0.4727
rs3205718				33790	0.3832
rs148713408				33790	0.0126
rs616402				33790	0.3069
rs62033406				33790	0.4054
rs28929474	32457	1316	15	33790	0.0199
rs5752793	14093	11141	2809	33790	0.2988
rs200550907				33790	0.2041
rs113198678				33790	0.0808

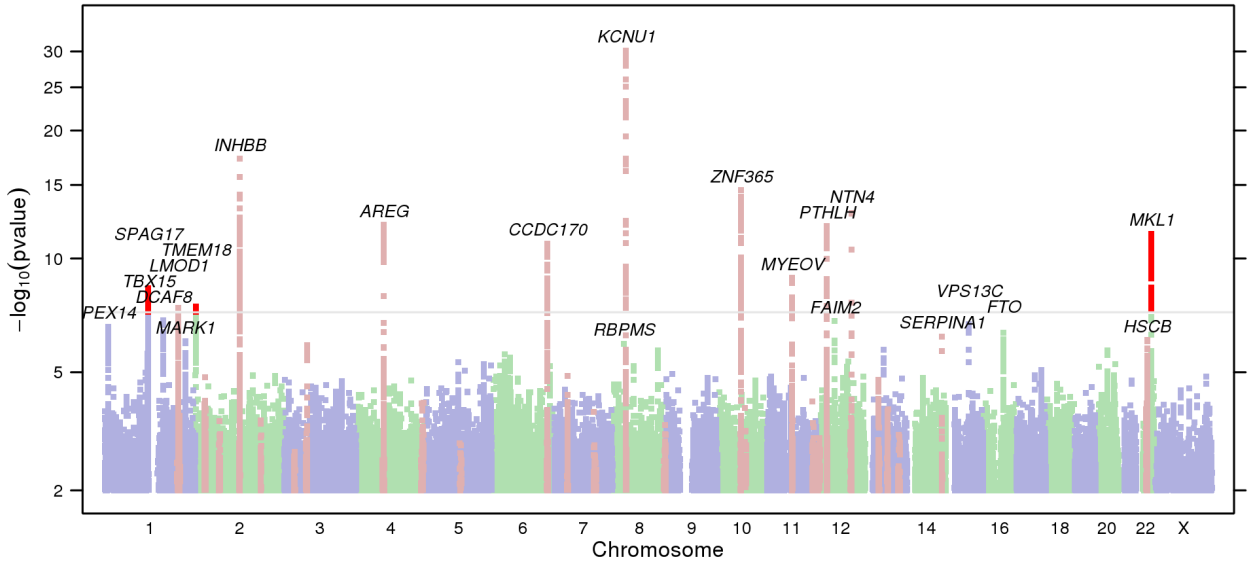
Annotations from NHGRI GWAS Catalog

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
8p11.23	36847115	rs10110651	7.1×10^{-31}	−1006	1.000	rs7816345	2.0×10^{-14}	22747683	Breast size	ZNF703,MRPS7P1,KCNU1
2q14.2	121089731	rs17625845	5.6×10^{-18}	0	1.000	rs17625845	5.0×10^{-10}	22747683	Breast size	INHBB
10q21.2	64178657	rs3081227	2.5×10^{-15}	8907	0.706	rs7089814	3.0×10^{-9}	22747683	Breast size	ZNF365, ALDH7A1P4, ADO
12q22	96027759	rs17356907	1.3×10^{-13}	0	1.000	rs17356907	9.0×10^{-6}	23535733	Breast cancer	NTN4
12q22	96027759	rs17356907	1.3×10^{-13}	0	1.000	rs17356907	2.0×10^{-22}	23535729	Breast cancer	NTN4
12q22	96027759	rs17356907	1.3×10^{-13}	0	1.000	rs17356907	1.0×10^{-6}	22747683	Breast size	NEDD1, MIR135A2
4q13.3	75547013	rs7659874	8.6×10^{-13}	−44526	0.630	rs62314947	5.0×10^{-8}	22747683	Breast size	AREG, EREG, …
12p11.22	28154895	rs1838564	1.0×10^{-12}	185	0.970	rs10771399	8.0×10^{-6}	23544013	Breast Cancer in BRCA1 mutation carriers	Intergenic
12p11.22	28154895	rs1838564	1.0×10^{-12}	185	0.970	rs10771399	2.0×10^{-12}	23535733	Breast cancer	PTHLH
12p11.22	28154895	rs1838564	1.0×10^{-12}	185	0.970	rs10771399	8.0×10^{-31}	23535729	Breast cancer	PTHLH
12p11.22	28154895	rs1838564	1.0×10^{-12}	1186	0.777	rs12371778	1.0×10^{-8}	22747683	Breast size	PTHLH, LOC100129646
22q13.1	40960692	rs5995875	3.2×10^{-12}	−84458	0.818	rs6001930	2.0×10^{-6}	23535733	Breast cancer	MKL1
22q13.1	40960692	rs5995875	3.2×10^{-12}	−84458	0.818	rs6001930	9.0×10^{-19}	23535729	Breast cancer	MKL1
22q13.1	40960692	rs5995875	3.2×10^{-12}	35675	0.895	rs17002034	4.0×10^{-6}	20889312	Bipolar disorder and schizophrenia	
6q25.1	151952332	rs9397437	1.3×10^{-11}	−38219	0.649	rs3757318	2.0×10^{-21}	23535729	Breast cancer	ESR1
6q25.1	151952332	rs9397437	1.3×10^{-11}	−38219	0.649	rs3757318	3.0×10^{-6}	20453838	Breast cancer	ESR1,C6orf97
6q25.1	151952332	rs9397437	1.3×10^{-11}	−12982	0.847	rs3734805	1.0×10^{-7}	21263130	Breast cancer	ESR1

6q25.1	151952332	rs9397437	1.3×10^{-11}	5382	0.728	rs12173570	6.0×10^{-11}	22747683	Breast size	ESR1, C6orf97
6q25.1	151952332	rs9397437	1.3×10^{-11}	35025	0.793	rs9383938	2.0×10^{-10}	22976474	Breast cancer	
11q13.3	69143284	rs7102705	1.1×10^{-9}	0	1.000	rs7102705	5.0×10^{-6}	22747683	Breast size	IFITM9P, CCND1
2p25.3	632922	rs62105303	2.8×10^{-8}	−18754	0.872	rs2947411	2.0×10^{-8}	21102462	Menarche (age at onset)	TMEM18
2p25.3	632922	rs62105303	2.8×10^{-8}	−10095	0.914	rs2867125	3.0×10^{-49}	20935630	Body mass index	TMEM18
2p25.3	632922	rs62105303	2.8×10^{-8}	−8888	0.914	rs6711012	6.0×10^{-35}	23563607	Obesity	TMEM18
2p25.3	632922	rs62105303	2.8×10^{-8}	−8244	0.914	rs2903492	6.0×10^{-15}	23563607	Body mass index	TMEM18
2p25.3	632922	rs62105303	2.8×10^{-8}	−3678	0.957	rs12463617	3.0×10^{-17}	23669352	Body mass index	TMEM18
2p25.3	632922	rs62105303	2.8×10^{-8}	−3678	0.957	rs12463617	2.0×10^{-13}	23563609	Obesity (early onset extreme)	TMEM18
2p25.3	632922	rs62105303	2.8×10^{-8}	1983	0.957	rs6548238	1.0×10^{-18}	19079261	Body mass index	TMEM18
2p25.3	632922	rs62105303	2.8×10^{-8}	12031	0.940	rs7561317	2.0×10^{-18}	19079260	Weight	TMEM18
2p25.3	632922	rs62105303	2.8×10^{-8}	12031	0.940	rs7561317	4.0×10^{-17}	19079260	Body mass index	TMEM18
1q32.1	201886769	rs34091558	3.1×10^{-8}	−1817	0.936	rs2819348	3.0×10^{-6}	22747683	Breast size	ATP2B4
12q13.12	50261809	rs3205718	1.2×10^{-7}	−14341	0.841	rs7138803	1.0×10^{-20}	23563607	Obesity	LOC144233
12q13.12	50261809	rs3205718	1.2×10^{-7}	−14341	0.841	rs7138803	2.0×10^{-9}	23563607	Body mass index	LOC144233
12q13.12	50261809	rs3205718	1.2×10^{-7}	−14341	0.841	rs7138803	2.0×10^{-17}	20935630	Body mass index	FAIM2
12q13.12	50261809	rs3205718	1.2×10^{-7}	−14341	0.841	rs7138803	8.0×10^{-7}	19557197	Waist circumference	FAIM2, BCDIN3D
12q13.12	50261809	rs3205718	1.2×10^{-7}	−14341	0.841	rs7138803	2.0×10^{-7}	19079260	Weight	BCDIN3D, FAIM2
12q13.12	50261809	rs3205718	1.2×10^{-7}	−14341	0.841	rs7138803	1.0×10^{-7}	19079260	Body mass index	BCDIN3D, FAIM2
1p36.22	10566272	rs616402	2.0×10^{-7}	−57	0.807	rs616488	1.0×10^{-8}	23535733	Breast cancer	PEX14
1p36.22	10566272	rs616402	2.0×10^{-7}	−57	0.807	rs616488	2.0×10^{-10}	23535729	Breast cancer	PEX14
1p36.22	10566272	rs616402	2.0×10^{-7}	15386	0.987	rs2056417	4.0×10^{-7}	23817569	Self-reported allergy	PEX14
16q12.2	53824226	rs62033406	3.7×10^{-7}	−23472	0.808	rs9940128	4.0×10^{-23}	23669352	Body mass index	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−23472	0.808	rs9940128	2.0×10^{-9}	22399527	Metabolic syndrome	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−23272	0.844	rs1421085	1.0×10^{-9}	23636237	Dietary macronutrient intake	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−23272	0.844	rs1421085	3.0×10^{-28}	23563609	Obesity (early onset extreme)	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−23272	0.844	rs1421085	1.0×10^{-28}	19151714	Obesity	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−20652	0.844	rs1558902	1.0×10^{-7}	21544081	Obesity	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−20652	0.844	rs1558902	5.0×10^{-120}	20935630	Body mass index	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−20652	0.844	rs1558902	7.0×10^{-13}	20421936	Obesity (extreme)	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−20652	0.844	rs1558902	5.0×10^{-19}	19557197	Waist circumference	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−14979	0.808	rs1121980	4.0×10^{-8}	18454148	Body mass index	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−14979	0.808	rs1121980	1.0×10^{-7}	18159244	Obesity (early onset extreme)	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−10859	0.892	rs17817449	6.0×10^{-7}	23535733	Breast cancer	MIR1972-2, FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−10859	0.892	rs17817449	6.0×10^{-14}	23535729	Breast cancer	MIR1972-2, FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−10859	0.892	rs17817449	2.0×10^{-12}	21552555	Obesity	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−10776	0.887	rs8043757	5.0×10^{-110}	23563607	Obesity	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−7951	0.887	rs8050136	6.0×10^{-6}	23209189	Type 2 diabetes	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−7951	0.887	rs8050136	3.0×10^{-26}	21706003	Adiposity	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−7951	0.887	rs8050136	4.0×10^{-8}	21037115	Body mass in chronic obstructive pulmonary disease	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−7951	0.887	rs8050136	5.0×10^{-36}	19079260	Weight	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−7951	0.887	rs8050136	1.0×10^{-47}	19079260	Body mass index	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−7951	0.887	rs8050136	2.0×10^{-17}	19056611	Type 2 diabetes	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−7951	0.887	rs8050136	7.0×10^{-6}	18372903	Type 2 diabetes	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−7951	0.887	rs8050136	7.0×10^{-14}	17463249	Type 2 diabetes	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−7951	0.887	rs8050136	1.0×10^{-12}	17463248	Type 2 diabetes	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−4333	0.897	rs11075990	2.0×10^{-51}	23563607	Body mass index	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−3699	0.897	rs9939609	1.0×10^{-20}	22693455	Type 2 diabetes	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−3699	0.897	rs9939609	3.0×10^{-8}	21102462	Menarche (age at onset)	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−3699	0.897	rs9939609	2.0×10^{-7}	19396169	Biomedical quantitative traits	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−3699	0.897	rs9939609	4.0×10^{-51}	19079261	Body mass index	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−3699	0.897	rs9939609	2.0×10^{-7}	17554300	Type 2 diabetes	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−3699	0.897	rs9939609	2.0×10^{-20}	17434869	Body mass index	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	−2611	0.903	rs7202116	2.0×10^{-10}	22982992	Body mass index	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	1262	0.865	rs9941349	6.0×10^{-12}	19553259	Obesity (extreme)	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	3840	0.898	rs17817964	1.0×10^{-10}	23583978	Body mass index	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	6239	0.818	rs9930506	9.0×10^{-7}	17658951	Obesity-related traits	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	7545	0.823	rs9922619	6.0×10^{-8}	22589738	Subcutaneous adipose tissue	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	14909	0.683	rs8044769	4.0×10^{-6}	22763110	Osteoarthritis	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	18682	0.829	rs12149832	5.0×10^{-22}	22344221	Body mass index	FTO
16q12.2	53824226	rs62033406	3.7×10^{-7}	21261	0.645	rs11642841	3.0×10^{-8}	20581827	Type 2 diabetes	FTO

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
1q32.1	201884952	rs2819348	8.9×10^{-8}	0	1.000	rs2819348	3.0×10^{-6}	22747683	Breast size	ATP2B4
2p23.3	27598097	rs4665972	0.00015	0	1.000	rs4665972	1.0×10^{-6}	22747683	Breast size	GCKR, SLC4A1AP
2p23.3	27742603	rs780093	0.00025	0	1.000	rs780093	5.0×10^{-6}	22747683	Breast size	SLC4A1AP
2p14	64495627	rs1529102	0.0020	0	1.000	rs1529102	6.0×10^{-6}	22747683	Breast size	RPL23AP37, LOC100128607
2q14.2	120513133	rs13401620	3.0×10^{-5}	0	1.000	rs13401620	2.0×10^{-6}	22747683	Breast size	EPB41L5
2q14.2	121089731	rs17625845	5.6×10^{-18}	0	1.000	rs17625845	5.0×10^{-10}	22747683	Breast size	INHBB
2q14.2	121245122	rs4849887	3.0×10^{-13}	0	1.000	rs4849887	3.0×10^{-11}	22747683	Breast size	INHBB, LOC84931, GLI2
2q31.2	179104298	rs34479159	0.0059	0	1.000	rs34479159	6.0×10^{-6}	22747683	Breast size	TTN
3p24.1	29209151	rs6549915	0.042	0	1.000	rs6549915	9.0×10^{-6}	22747683	Breast size	MESTP4, RBMS3
3p14.2	60763256	rs11919041	0.00092	0	1.000	rs11919041	2.0×10^{-6}	22747683	Breast size	NPCDR1,PTPRG
3p14.2	62733366	rs580384	1.8×10^{-6}	0	1.000	rs580384	4.0×10^{-6}	22747683	Breast size	CADPS
4q13.3	75502487	rs62314947	3.9×10^{-11}	0	1.000	rs62314947	5.0×10^{-8}	22747683	Breast size	AREG, EREG, EPGN, AREGB
4q34.3	180383312	rs2716816	0.00027	0	1.000	rs2716816	7.0×10^{-6}	22747683	Breast size	RPL19P8
6q25.1	151957714	rs12173570	2.6×10^{-9}	0	1.000	rs12173570	6.0×10^{-11}	22747683	Breast size	ESR1, C6orf97
7p14.2	36952021	rs10488023	0.00020	0	1.000	rs10488023	8.0×10^{-6}	22747683	Breast size	ELMO1
7q31.1	111642646	rs61159171	0.57	0	1.000	rs61159171	6.0×10^{-6}	22747683	Breast size	ZNF277
8p11.23	36846109	rs7816345	1.0×10^{-30}	0	1.000	rs7816345	2.0×10^{-14}	22747683	Breast size	ZNF703,MRPS7P1,KCNU1
8q24.3	142540925	rs7837045	0.36	0	1.000	rs7837045	5.0×10^{-6}	22747683	Breast size	FLJ43860
10q21.2	64187564	rs7089814	3.2×10^{-13}	0	1.000	rs7089814	3.0×10^{-9}	22747683	Breast size	ZNF365, ALDH7A1P4, ADO
10q22.2	76703006	rs10466033	0.39	0	1.000	rs10466033	6.0×10^{-6}	22747683	Breast size	COMTD1,SPA17P1
11q13.3	69143284	rs7102705	1.1×10^{-9}	0	1.000	rs7102705	5.0×10^{-6}	22747683	Breast size	IFITM9P, CCND1
11q24.2	125873102	rs7104745	0.039	0	1.000	rs7104745	6.0×10^{-6}	22747683	Breast size	KIRREL3
12p13.31	9380859	rs10771431	0.0020	0	1.000	rs10771431	2.0×10^{-6}	22747683	Breast size	LOC642846, LOC728715
12p11.22	28156081	rs12371778	1.0×10^{-10}	0	1.000	rs12371778	1.0×10^{-8}	22747683	Breast size	PTHLH, LOC100129646
12q22	96027759	rs17356907	1.3×10^{-13}	0	1.000	rs17356907	1.0×10^{-6}	22747683	Breast size	NEDD1, MIR135A2
13q13.3	36531548	rs34065801	0.0011	0	1.000	rs34065801	6.0×10^{-6}	22747683	Breast size	FAM48A
13q21.2	59953466	rs12585963	0.0048	0	1.000	rs12585963	2.0×10^{-6}	22747683	Breast size	TDRD3
13q31.3	93581791	rs9523848	0.042	0	1.000	rs9523848	3.0×10^{-6}	22747683	Breast size	GPC6
14q32.13	94796184	rs61280460	0.00032	0	1.000	rs61280460	8.0×10^{-7}	22747683	Breast size	SERPINA6, CLMN
22q12.1	29161007	rs4820792	2.8×10^{-6}	0	1.000	rs4820792	4.0×10^{-7}	22747683	Breast size	CHEK2, MTFP1, LOC646513, SEC14L2, SDC4P

Nearby Nonsynonymous SNPs

region	position	our.name	our.pval	dist	rsqr	assay.name	gene	aa.chg
8p11.23	36847115	rs10110651	7.1×10^{-31}	−58636	0.862	rs16885577	KCNU1	N916S
10q21.2	64178657	rs3081227	2.5×10^{-15}	−44461	0.609	rs3830349	ZNF365	?-10?
1p12	118770271	rs2359714	3.8×10^{-9}	−204318	0.510	rs10923472	SPAG17	P1348L
1q32.1	201886769	rs34091558	3.1×10^{-8}	−26143	0.904	rs2250377	SHISA4	I159M
1q32.1	201886769	rs34091558	3.1×10^{-8}	−17512	0.898	rs2820312	LMOD1	T295M
15q22.2	62220772	rs148713408	1.6×10^{-7}	−1435	1.000	rs75341202	VPS13C	L2157F
14q32.13	94844947	rs28929474	5.1×10^{-7}	0	1.000	rs28929474	SERPINA1	E366K

Nearby Expression QTLs

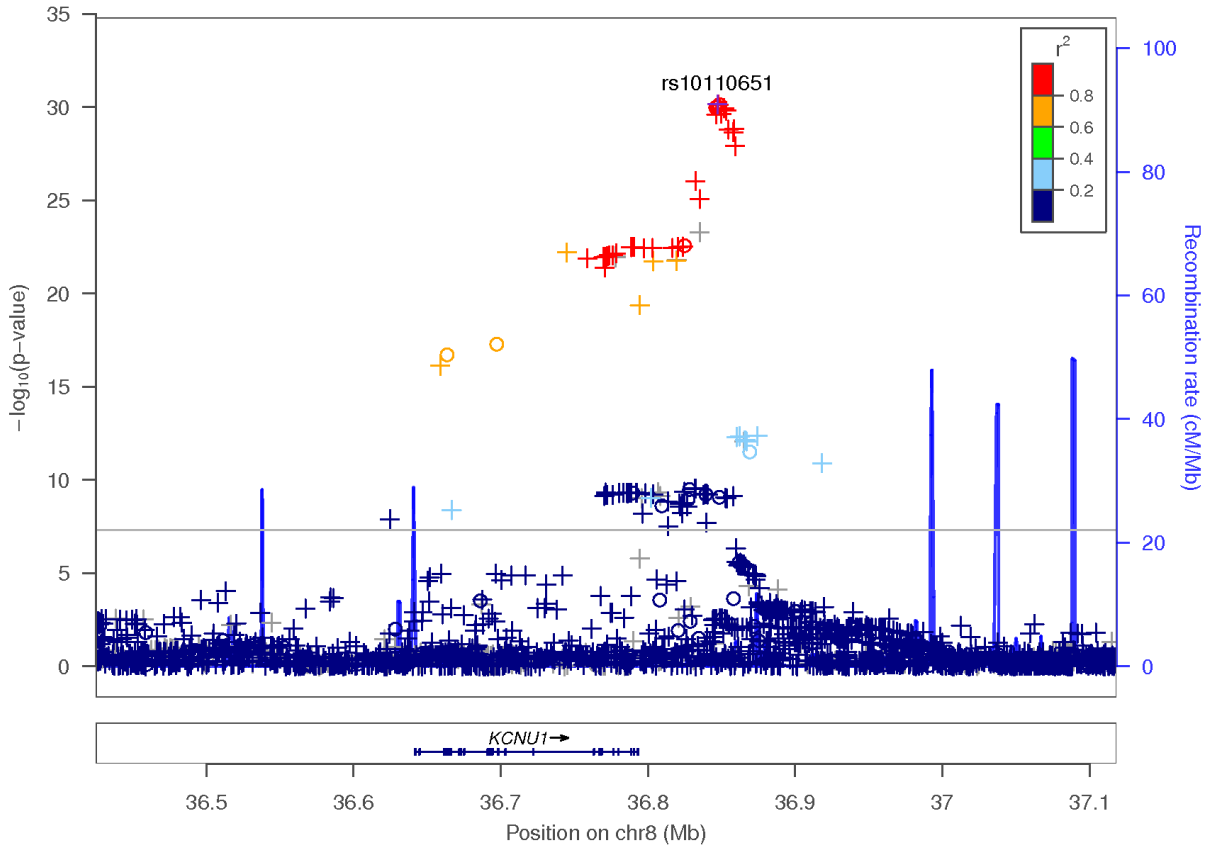
region	position	our.name	our.pval	dist	rsqr	assay.name	eqtl.dist	eqtl.gene	eqtl.pval	eqtl.rsqr	tissue	pubmed.id

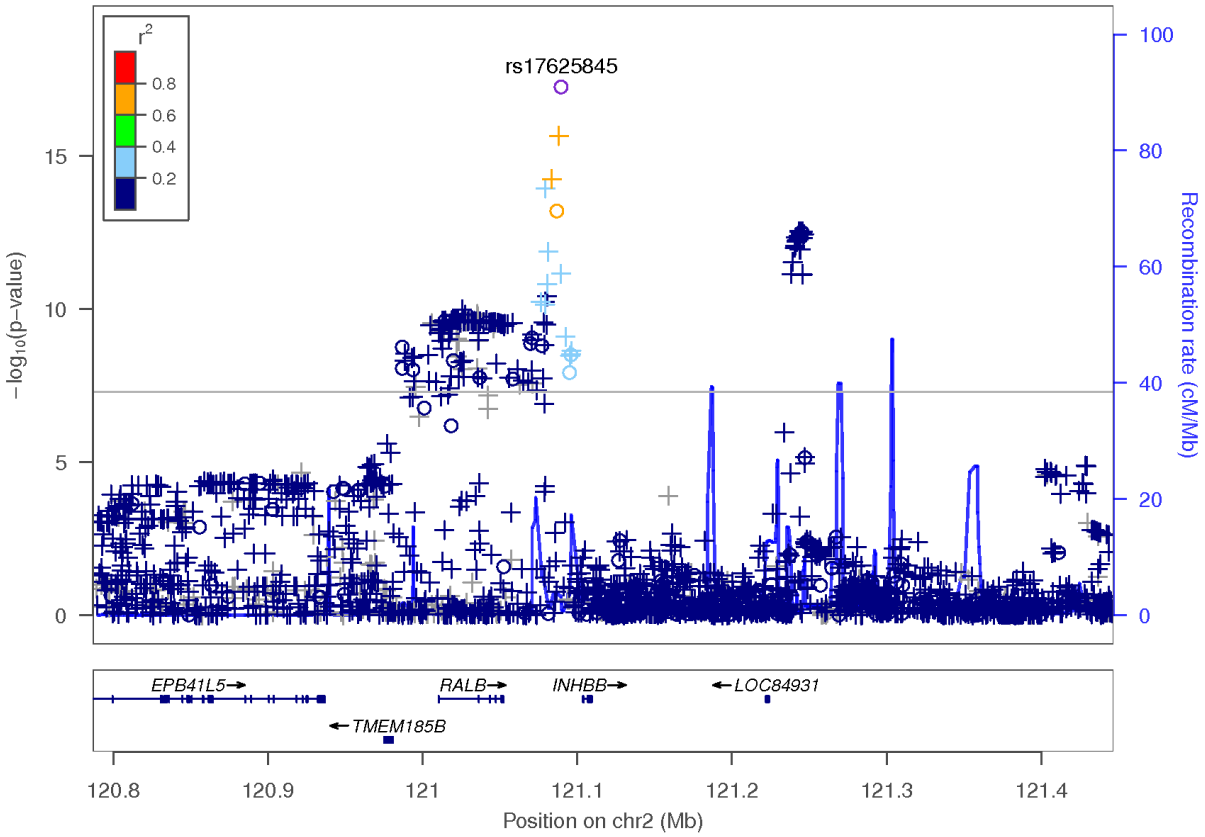
22q13.1	40960692	rs5995875	3.2×10^{-12}	35675	0.895	rs17002034	190074	SGSM3	5.1×10^{-6}	0.014	Monocyte	20502693
1p12	119500483	rs2645288	1.1×10^{-7}	37950	0.544	rs1409159	35406	WARS2	0.0	0.475	Lymphoblastoid	19644074
1p12	119500483	rs2645288	1.1×10^{-7}	37950	0.544	rs1409159	35406	WARS2	0.0	0.433	T-cell	19644074
1q23.2	160238857	rs6427508	1.1×10^{-7}	17147	0.889	rs2820421	2373	COPA	1.3×10^{-16}	0.048	Monocyte	20502693
1p36.22	10566272	rs616402	2.0×10^{-7}	−26729	0.875	rs607941	−750269	CLSTN1	0.00018	0.049	B-Cell	22446964
1p36.22	10566272	rs616402	2.0×10^{-7}	−25761	0.606	rs12402967	−153706	PGD	0.00010		Lymphoblastoid	20220756
1p36.22	10566272	rs616402	2.0×10^{-7}	−21725	0.875	rs668805	0	PEX14	1.2×10^{-19}	0.057	Monocyte	20502693
1p36.22	10566272	rs616402	2.0×10^{-7}	−2780	0.856	rs2506901	−72202	APITD1	6.9×10^{-9}	0.113	Monocyte	22446964
1q41	220701165	rs200550907	7.9×10^{-7}	−30130	0.581	rs3860322	201257	C1ORF115	4.8×10^{-12}	0.158	B-Cell	22446964

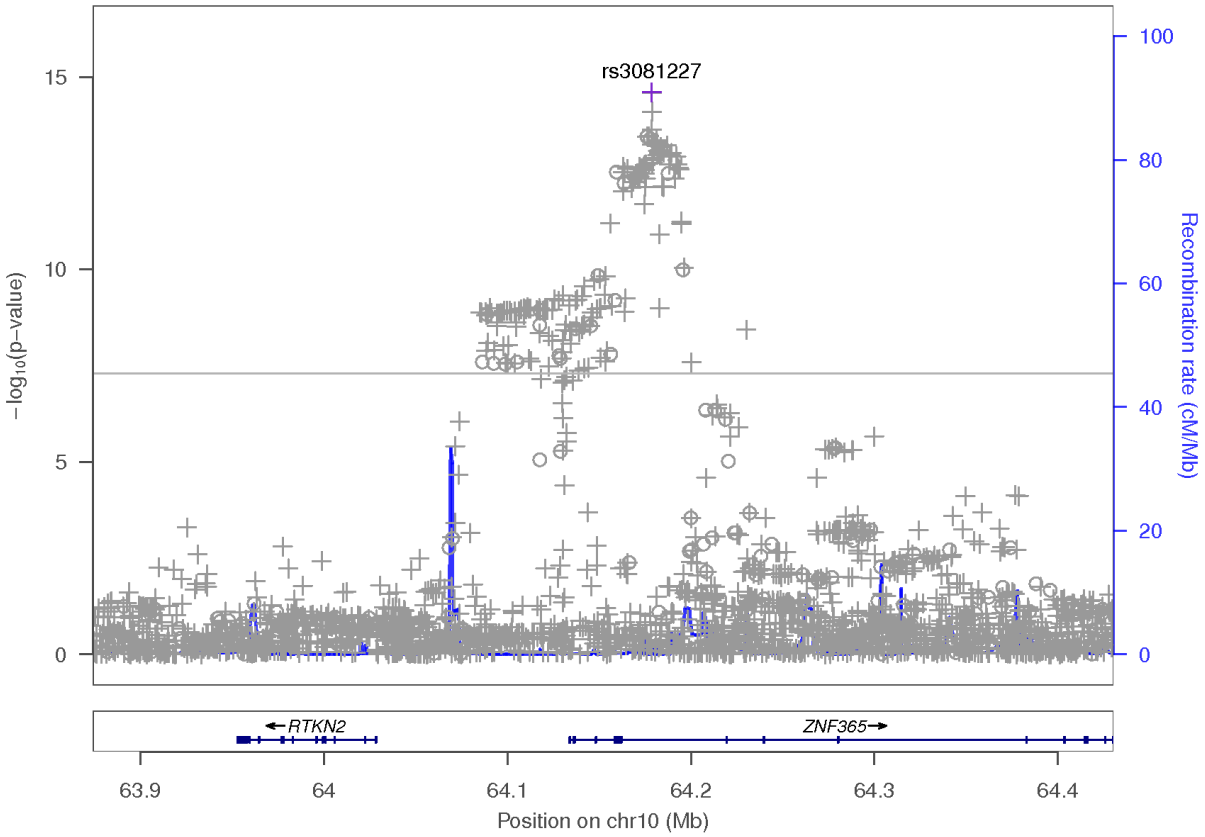
Nearby Clinical Variants

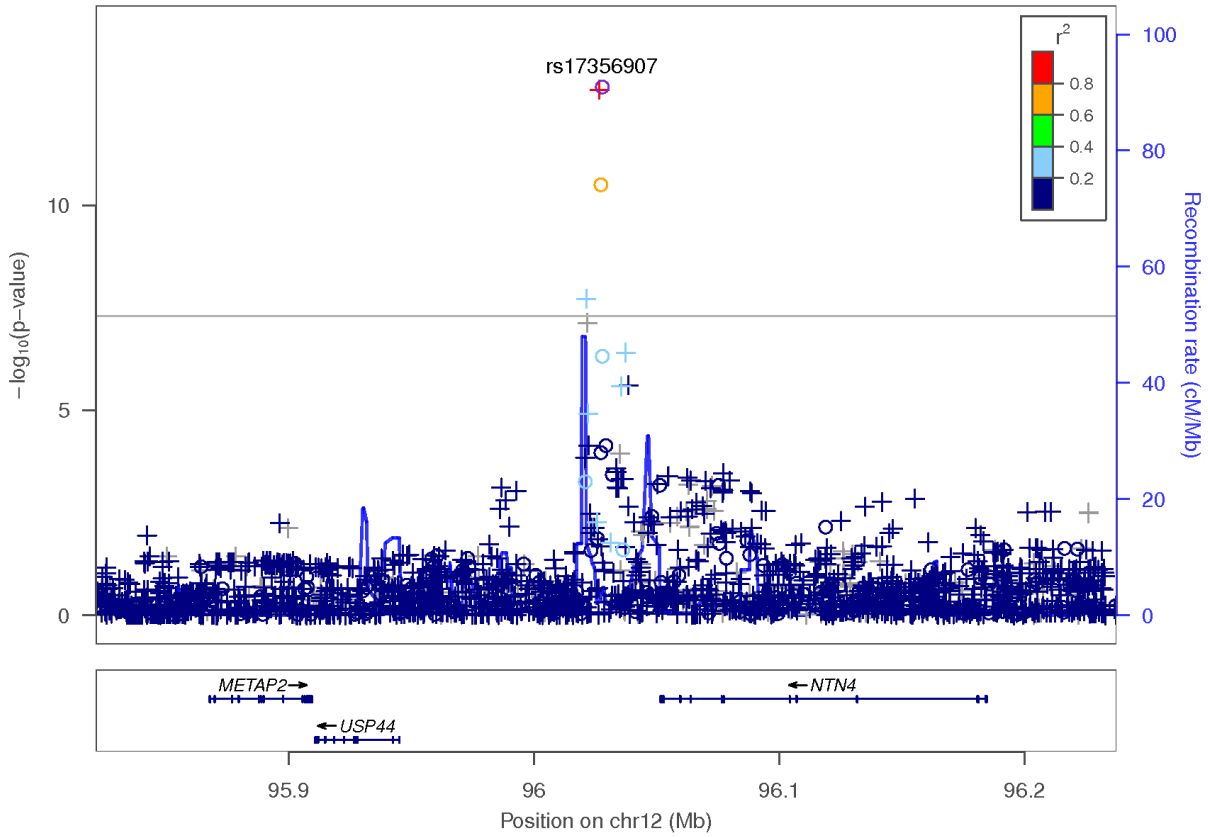
source	region	our.name	our.pval	dist	rsqr	assay.name	gene	phenotype	accession
clinvar	14q32.13	rs28929474	5.1×10^{-7}	0	1.000	rs28929474	SERPINA1	Chronic obstructive pulmonary disease	NCBI curation
clinvar	14q32.13	rs28929474	5.1×10^{-7}	0	1.000	rs28929474	SERPINA1	Alpha-1-antitrypsin deficiency	SNOMED CT30188007

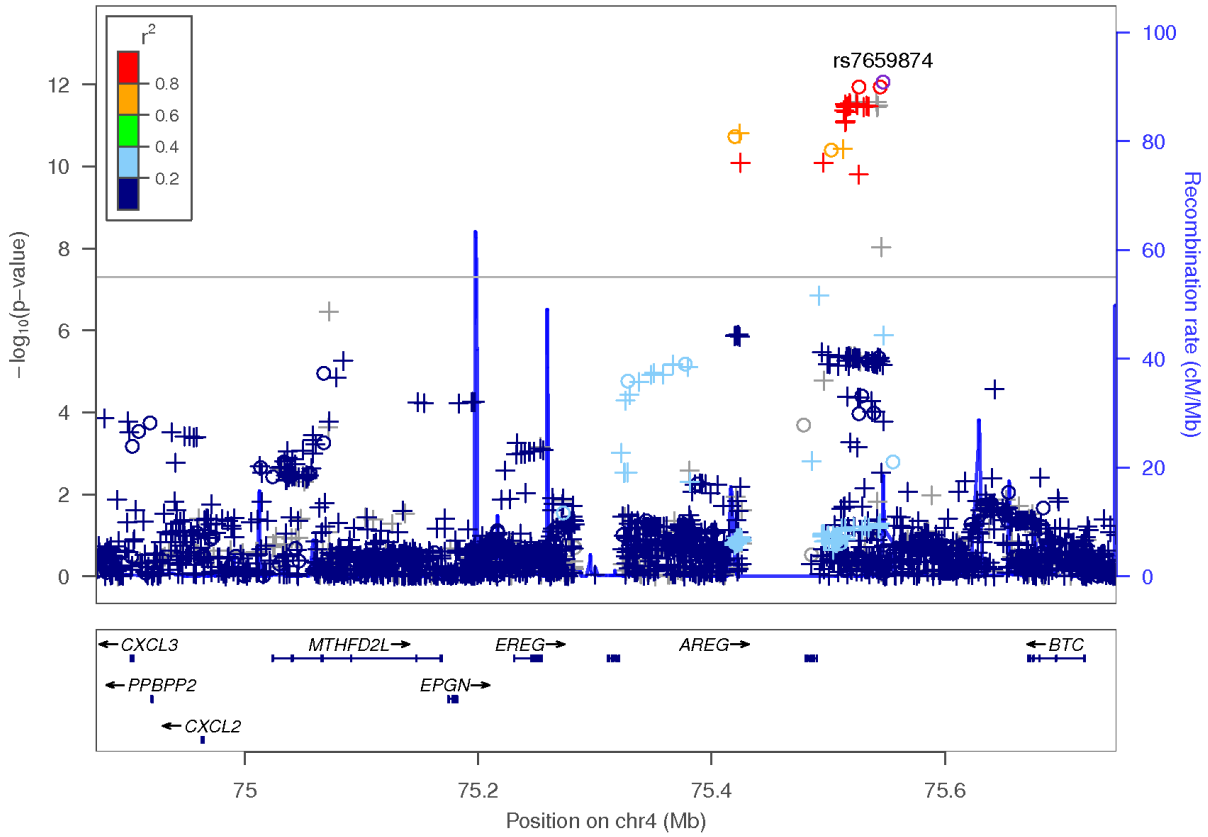
Regional Association Plots

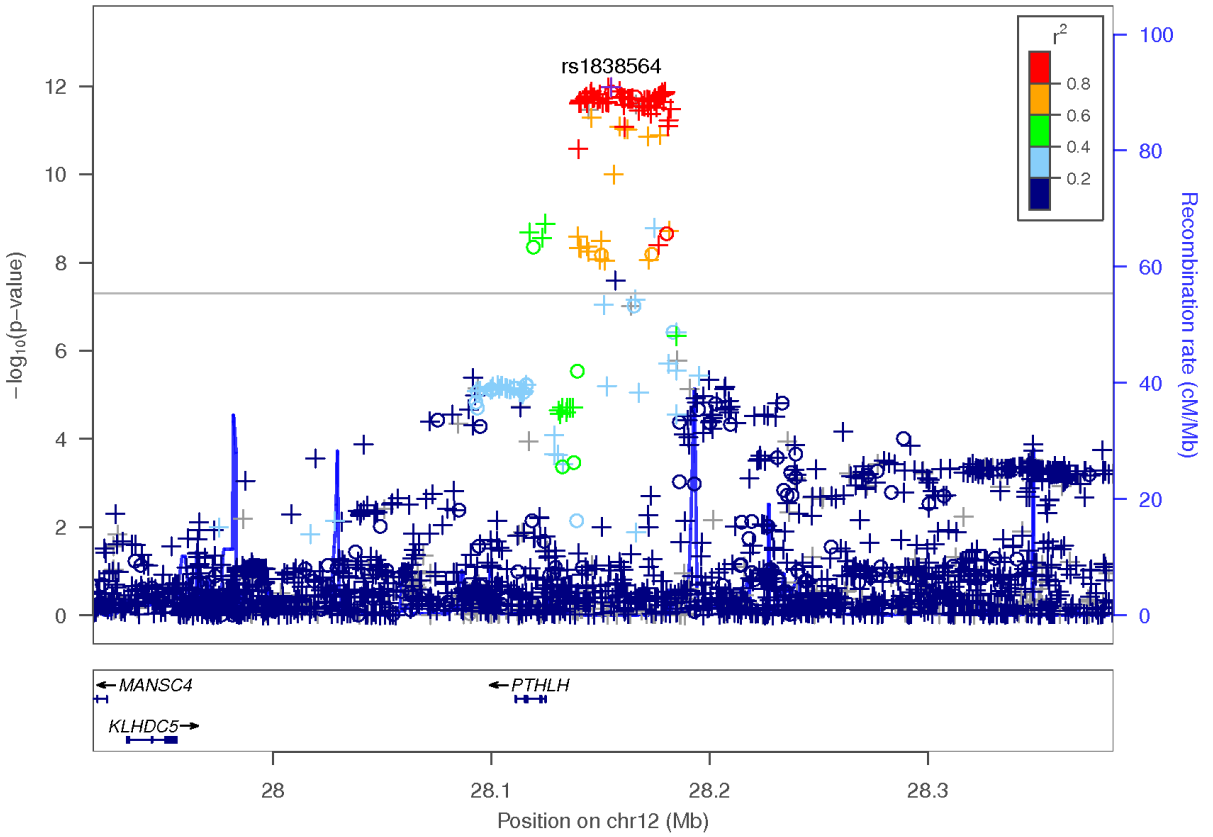


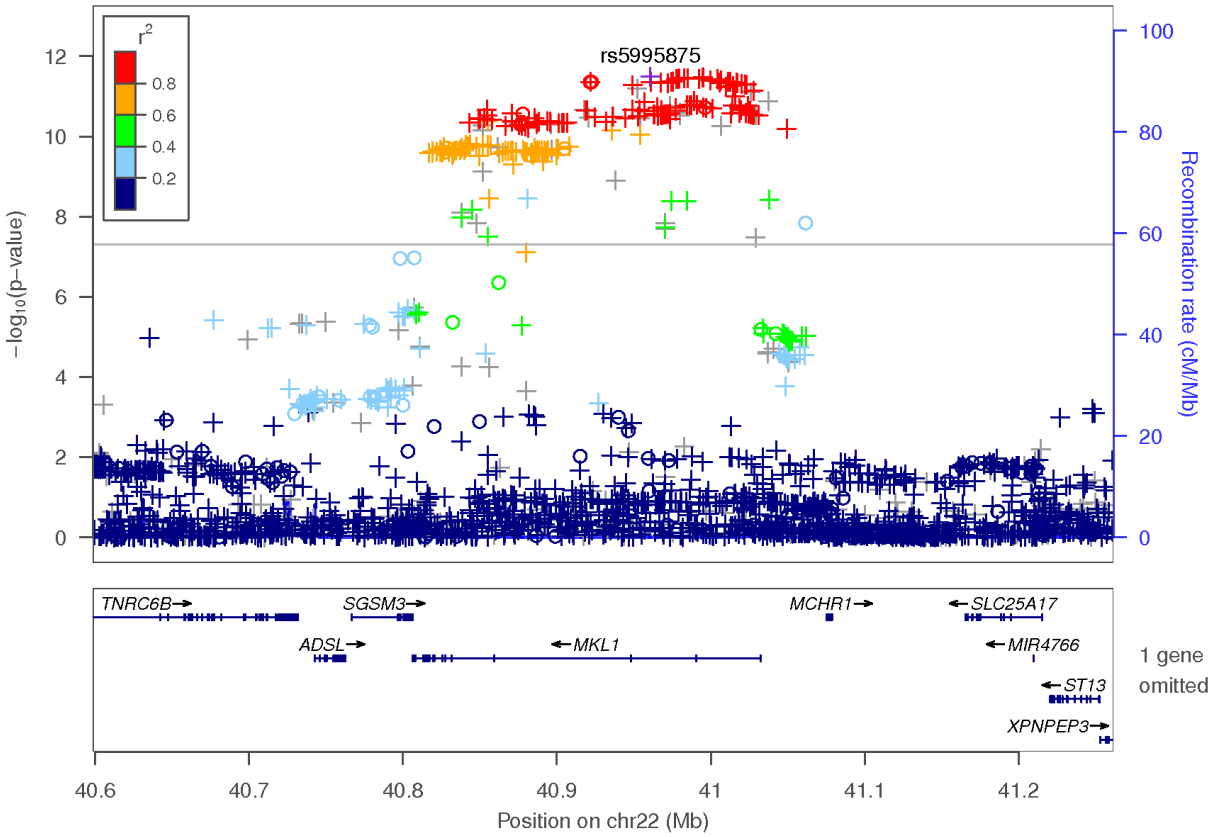

r²
0.8
0.6
0.4
0.2
rs17625845
−log₁₀(p−value)
Recombination rate (cM/Mb)
EPB41L5
RALB
INHBB
LOC84931
TMEM185B
120.8
120.9
121
121.1
121.2
121.3
121.4
Position on chr2 (Mb)

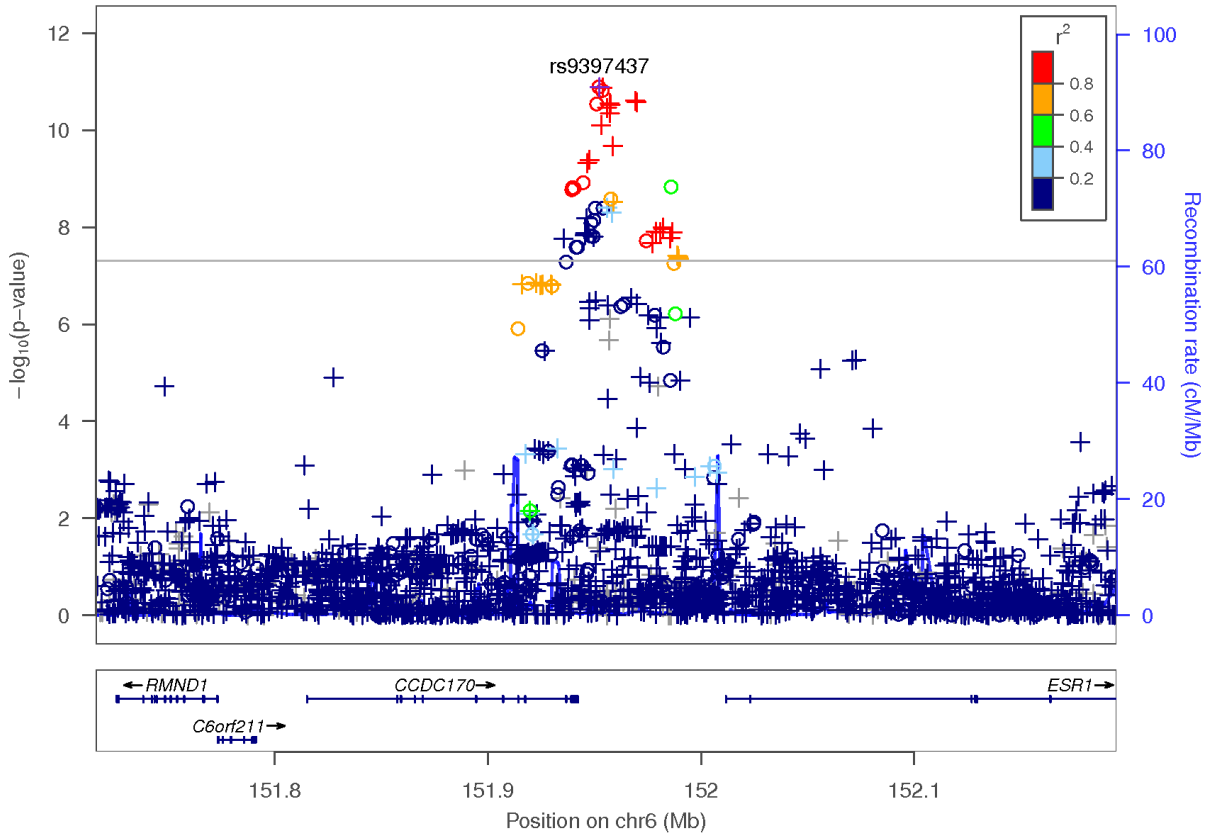

rs3081227
−log₁₀(p−value)
Recombination rate (cM/Mb)
RTKN2
ZNF365
63.9
64
64.1
64.2
64.3
64.4
Position on chr10 (Mb)

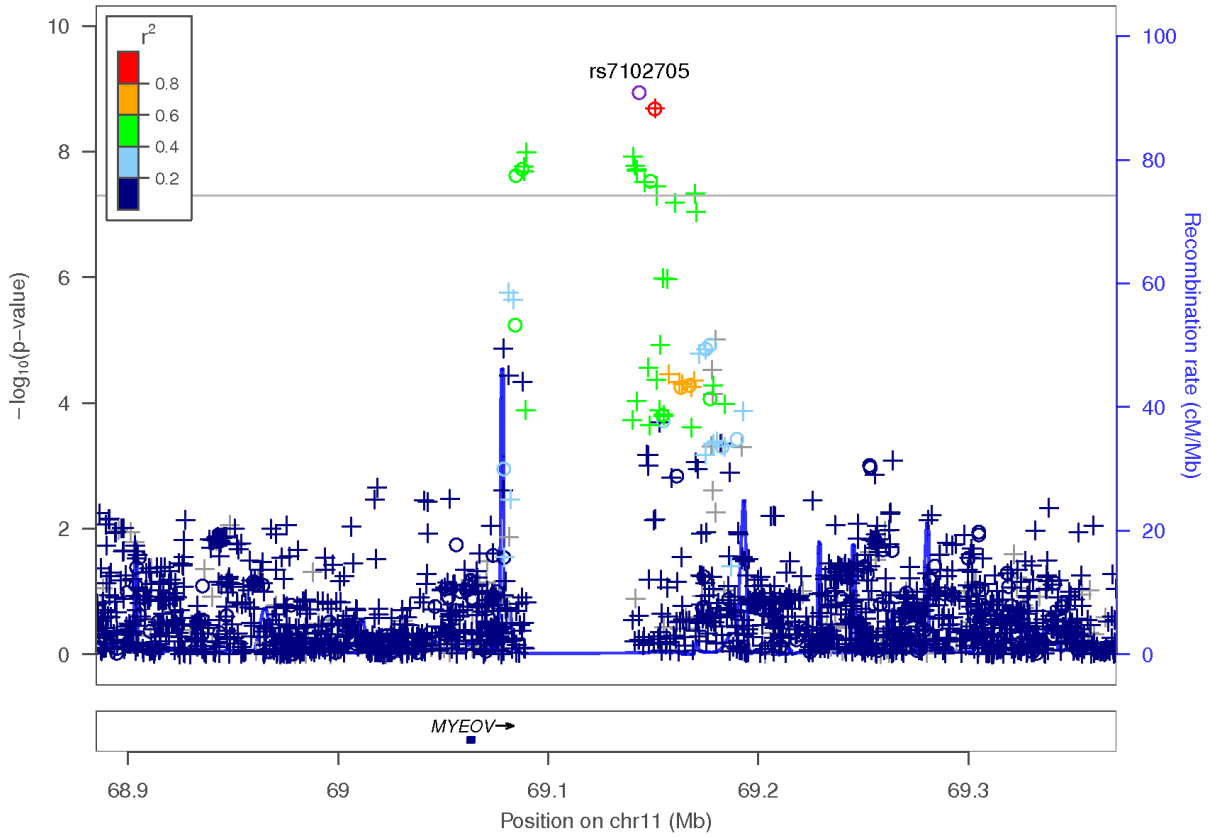

rs17356907
r²
0.8
0.6
0.4
0.2
−log₁₀(p−value)
Recombination rate (cM/Mb)
METAP2
USP44
NTN4
95.9
96
96.1
96.2
Position on chr12 (Mb)

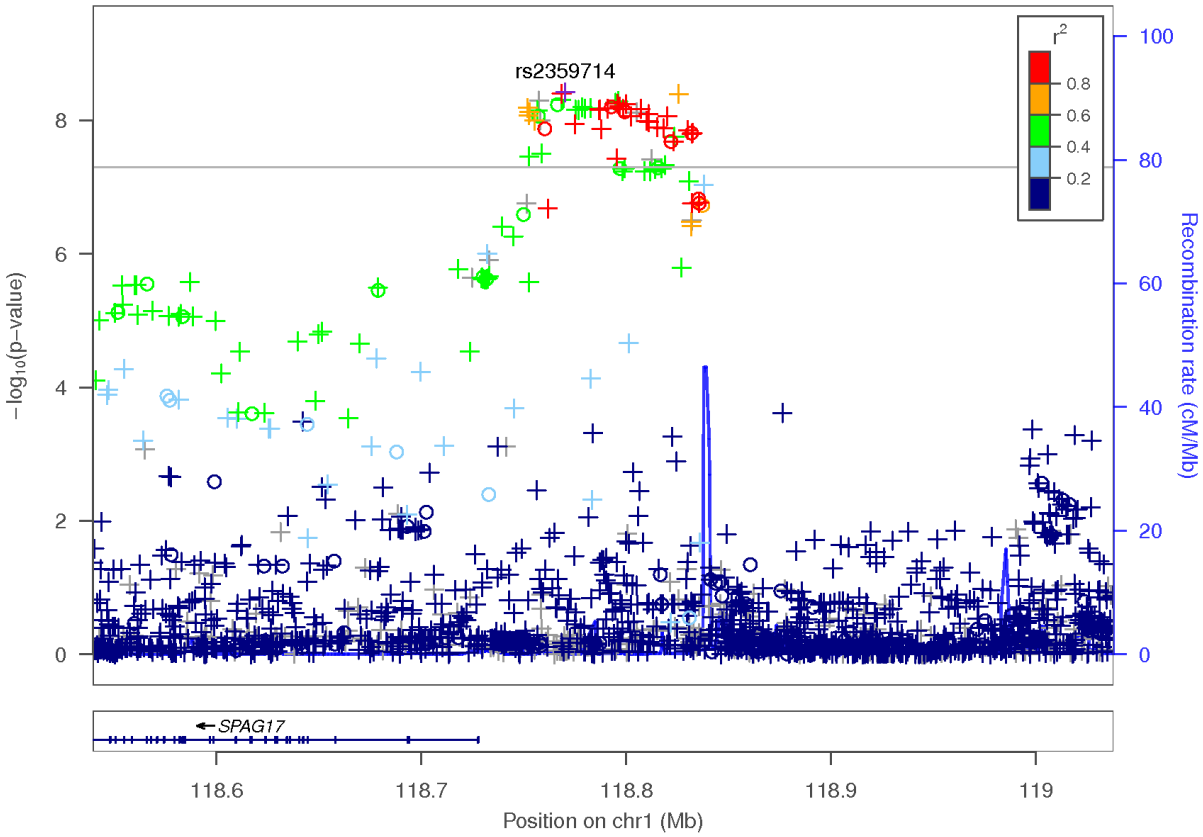

rs7659874
r²
0.8
0.6
0.4
0.2
−log₁₀(p−value)
Recombination rate (cM/Mb)
CXCL3
PPBPP2
CXCL2
MTHFD2L
EPGN
EREG
AREG
BTC
75
75.2
75.4
75.6
Position on chr4 (Mb)

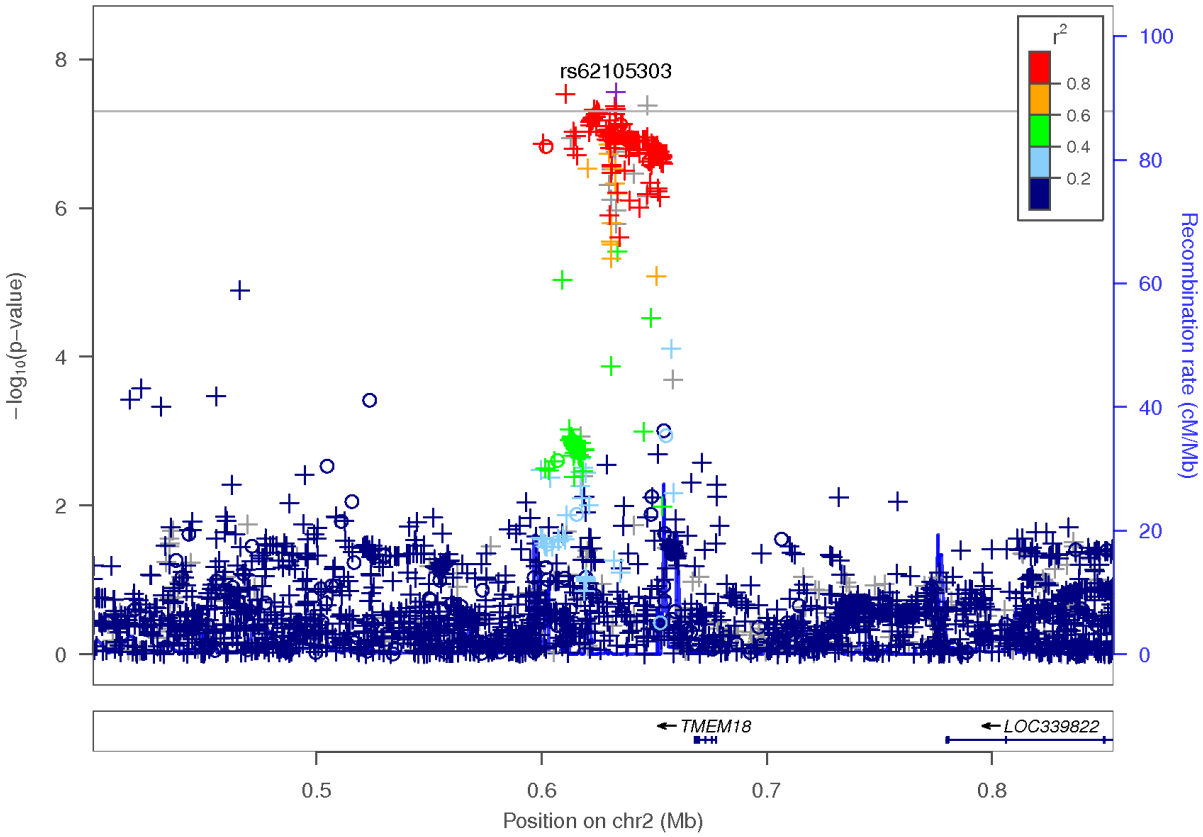

rs1838564
r²
0.8
0.6
0.4
0.2
−log₁₀(p−value)
Recombination rate (cM/Mb)
MANSC4
PTHLH
KLHDC5
28
28.1
28.2
28.3
Position on chr12 (Mb)

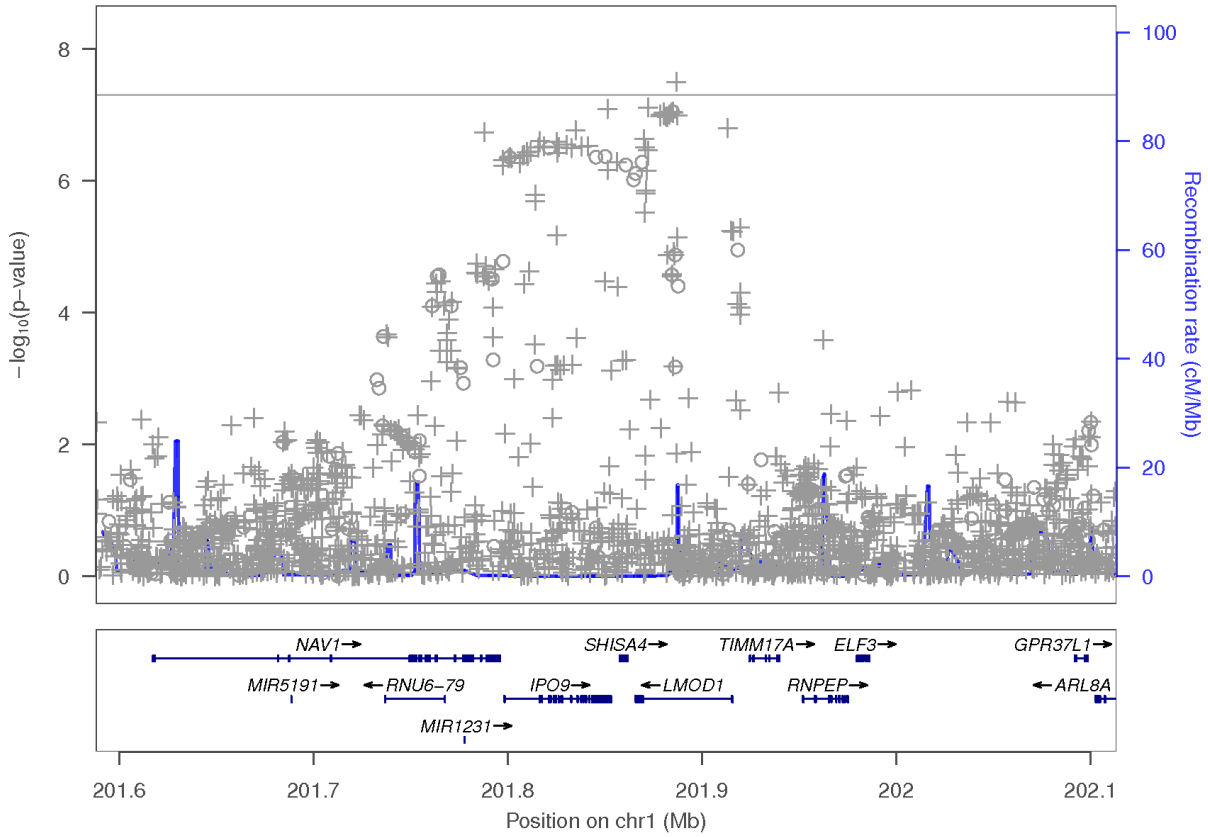

rs5995875
r²
0.8
0.6
0.4
0.2
−log₁₀(p−value)
Recombination rate (cM/Mb)
TNRC6B
SGSM3
MCHR1
SLC25A17
ADSL
MKL1
MIR4766
ST13
XPNPEP3
1 gene
omitted
40.6
40.7
40.8
40.9
41
41.1
41.2
Position on chr22 (Mb)

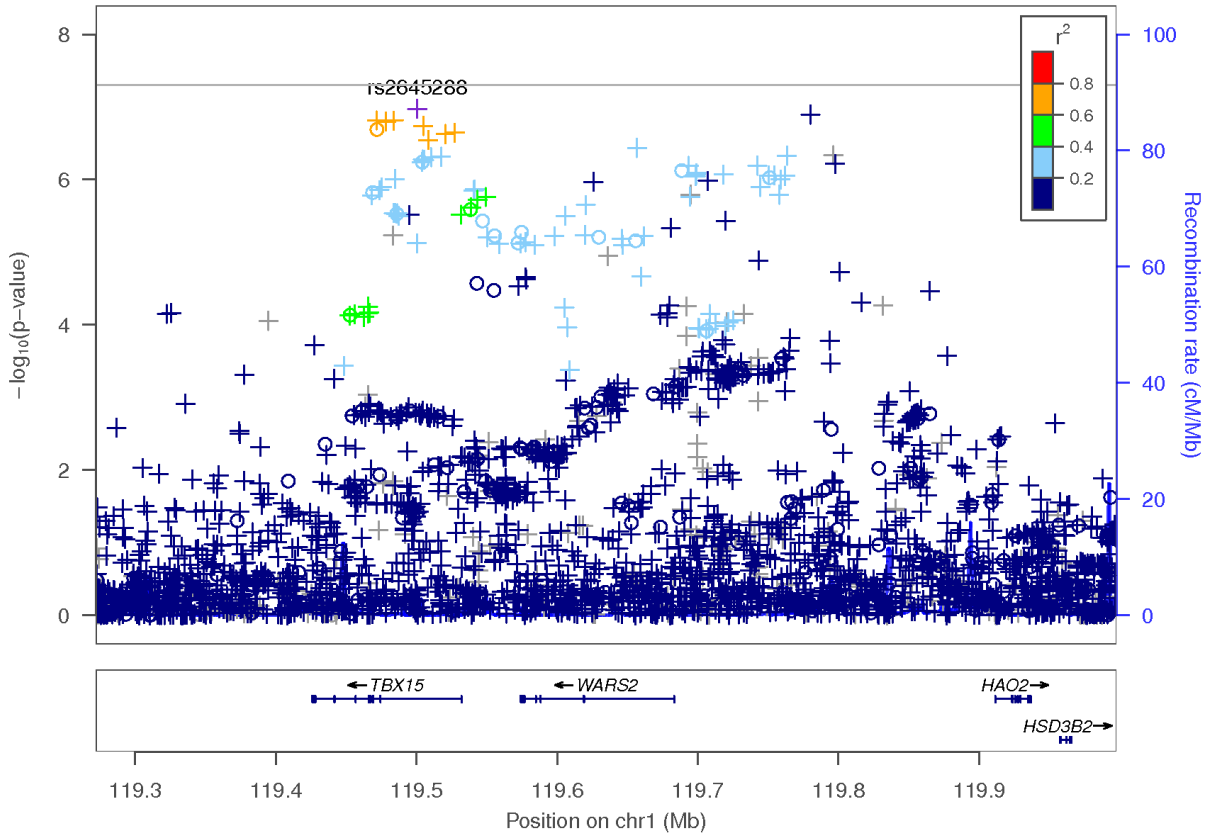

rs9397437
r2
0.8
0.6
0.4
0.2
−log10(p−value)
Recombination rate (cM/Mb)
RMND1
CCDC170
ESR1
C6orf211
151.8
151.9
152
152.1
Position on chr6 (Mb)

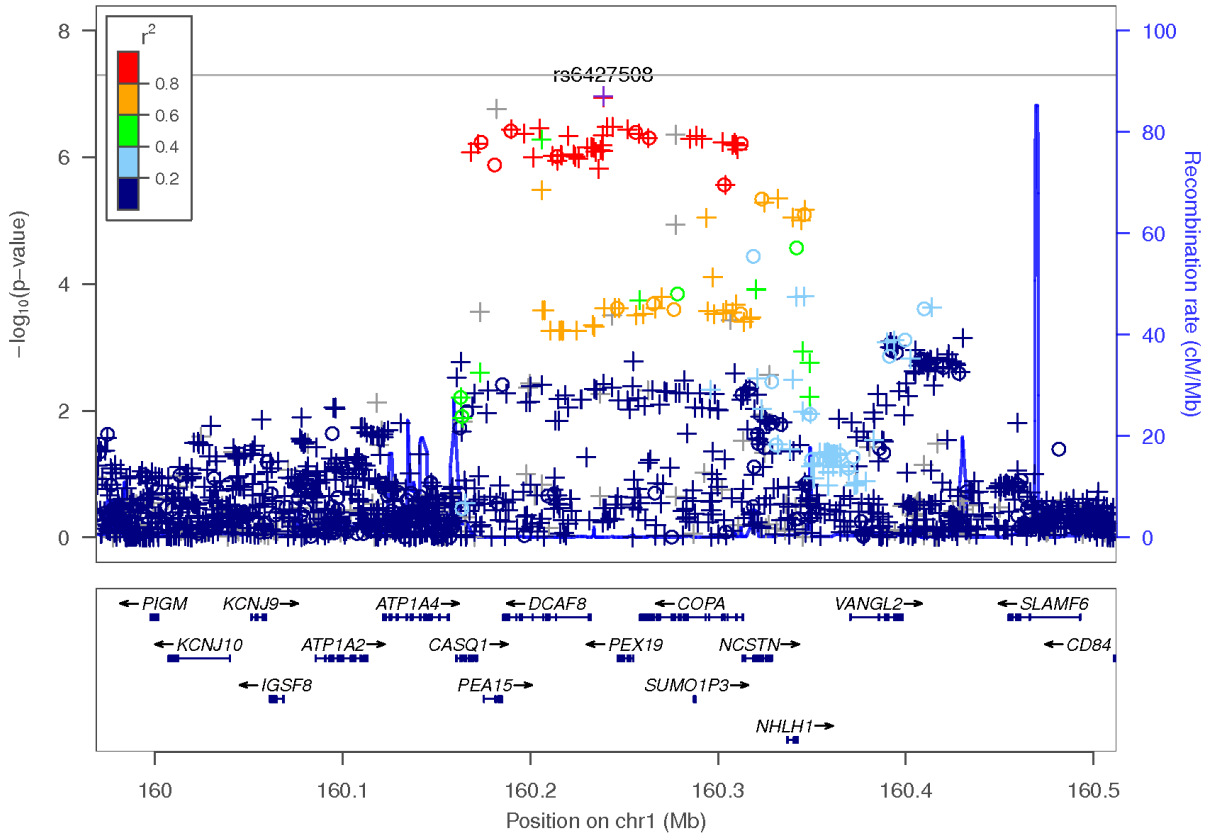

rs7102705
r2
0.8
0.6
0.4
0.2
−log10(p−value)
Recombination rate (cM/Mb)
MYEOV
68.9
69
69.1
69.2
69.3
Position on chr11 (Mb)

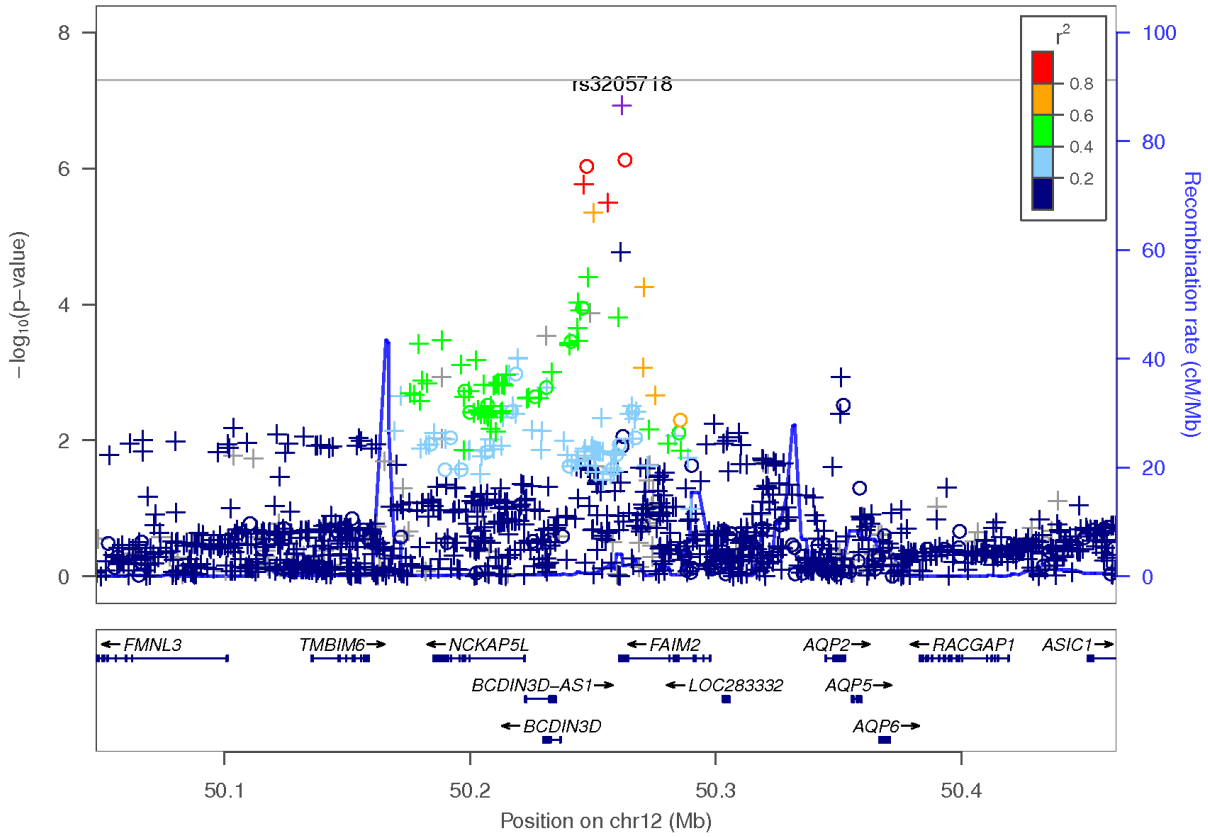

rs2359714
r²
0.8
0.6
0.4
0.2
−log₁₀(p−value)
Recombination rate (cM/Mb)
SPAG17
118.6
118.7
118.8
118.9
119
Position on chr1 (Mb)

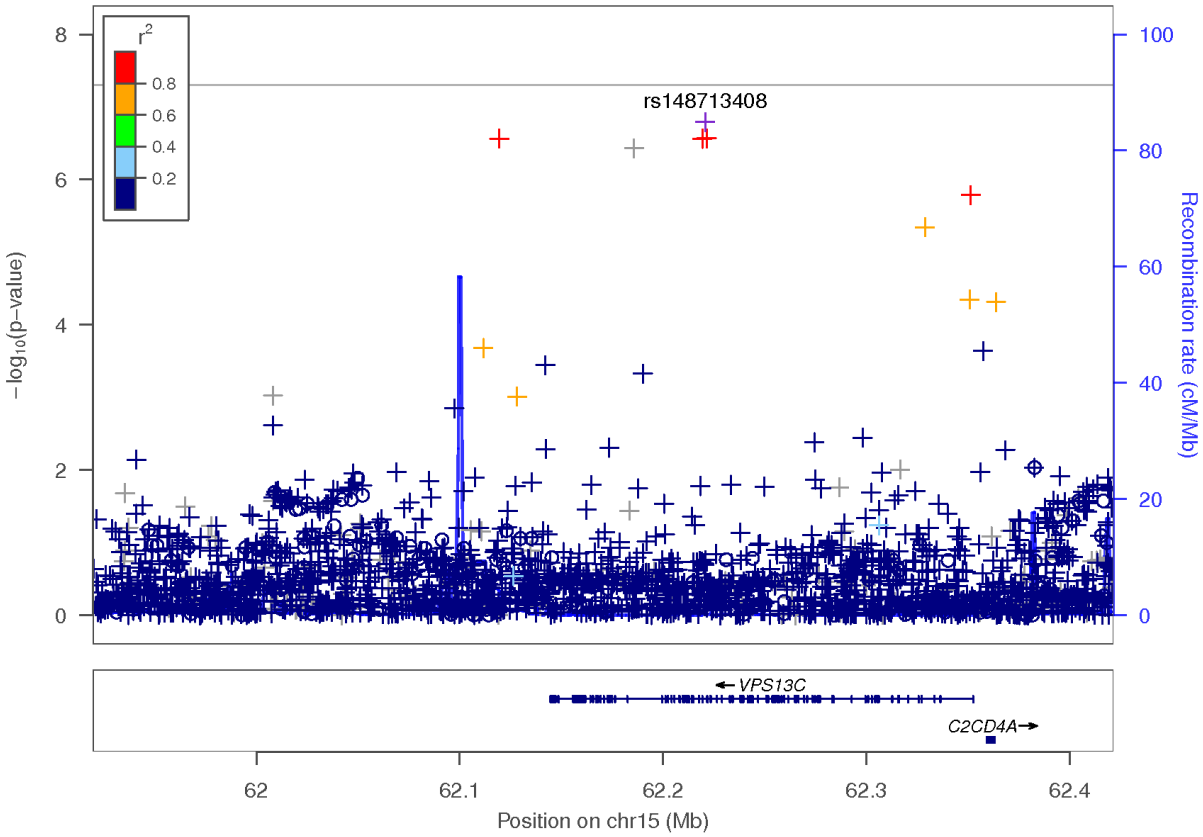

rs62105303
r²
0.8
0.6
0.4
0.2
−log₁₀(p−value)
Recombination rate (cM/Mb)
TMEM18
LOC339822
0.5
0.6
0.7
0.8
Position on chr2 (Mb)

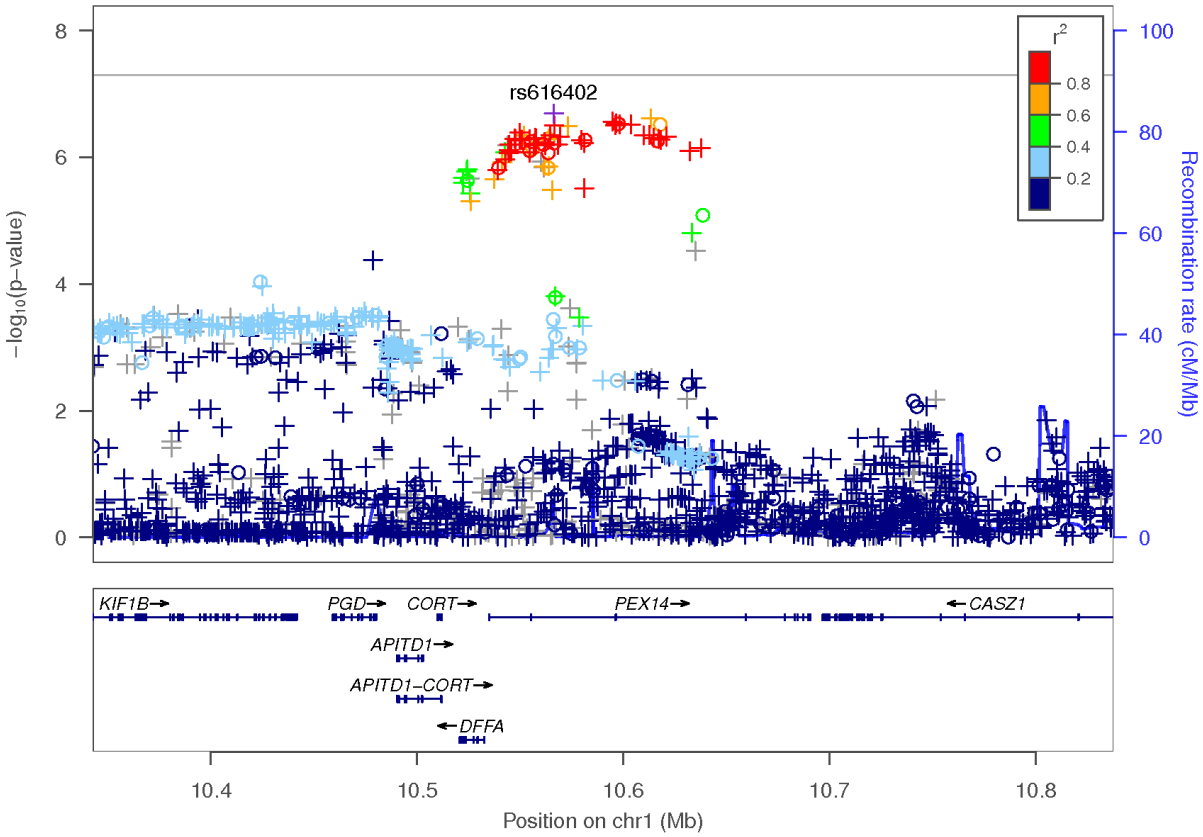

8
6
4
2
0
−log10(p−value)
100
80
60
40
20
0
Recombination rate (cM/Mb)
NAV1
SHISA4
TIMM17A
ELF3
GPR37L1
MIR5191
RNU6−79
IPO9
LMOD1
RNPEP
ARL8A
MIR1231
201.6
201.7
201.8
201.9
202
202.1
Position on chr1 (Mb)

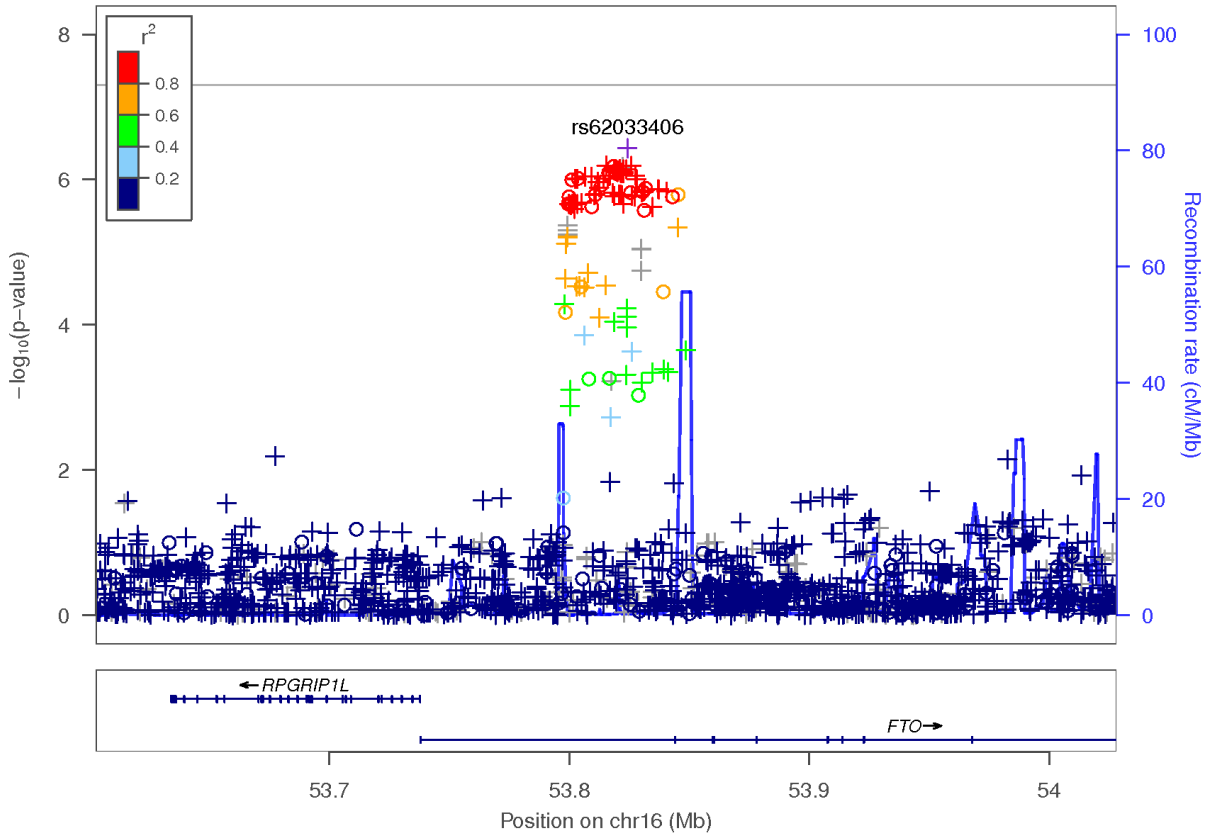

8
6
4
2
0
−log10(p−value)
rs2645288
r2
0.8
0.6
0.4
0.2
100
80
60
40
20
0
Recombination rate (cM/Mb)
TBX15
WARS2
HAO2
HSD3B2
119.3
119.4
119.5
119.6
119.7
119.8
119.9
Position on chr1 (Mb)

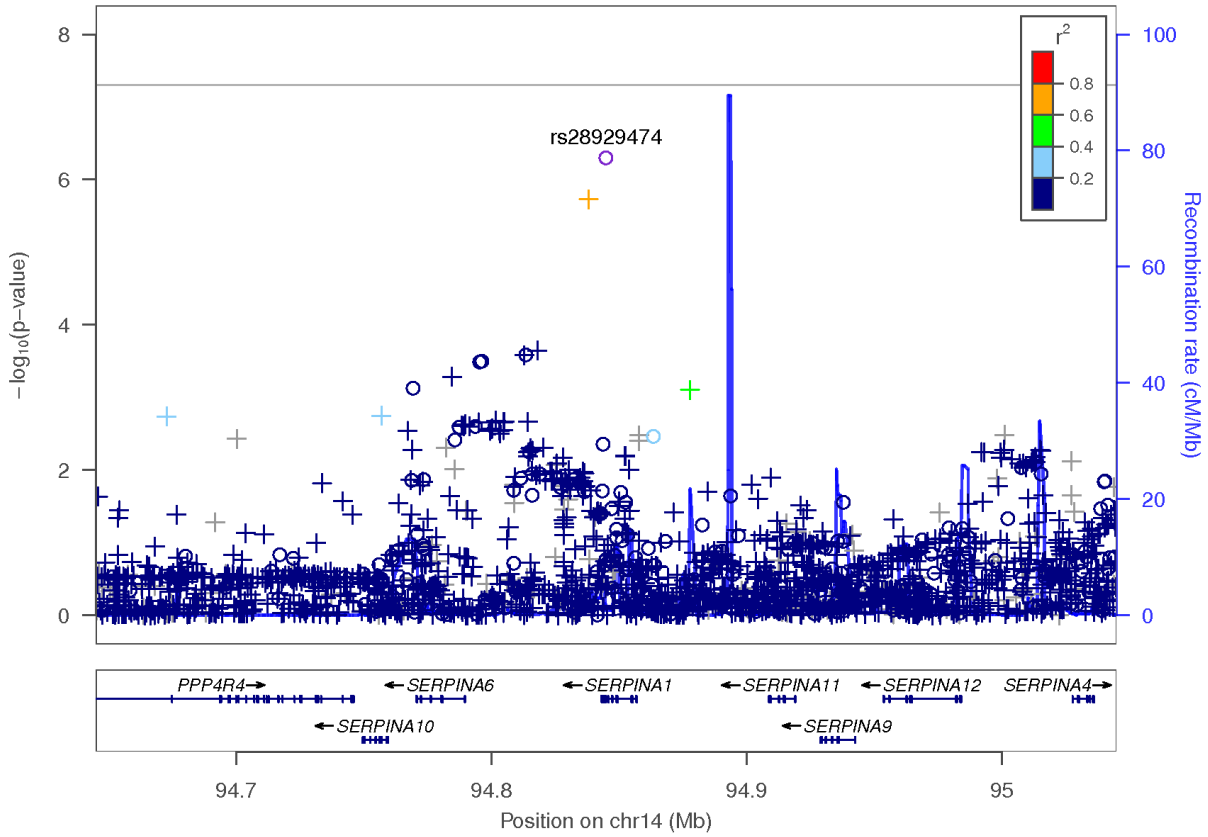

r²
0.8
0.6
0.4
0.2
rs6427508
−log₁₀(p−value)
Recombination rate (cM/Mb)
PIGM
KCNJ9
ATP1A4
DCAF8
COPA
VANGL2
SLAMF6
KCNJ10
ATP1A2
CASQ1
PEX19
NCSTN
CD84
IGSF8
PEA15
SUMO1P3
NHLH1
160
160.1
160.2
160.3
160.4
160.5
Position on chr1 (Mb)

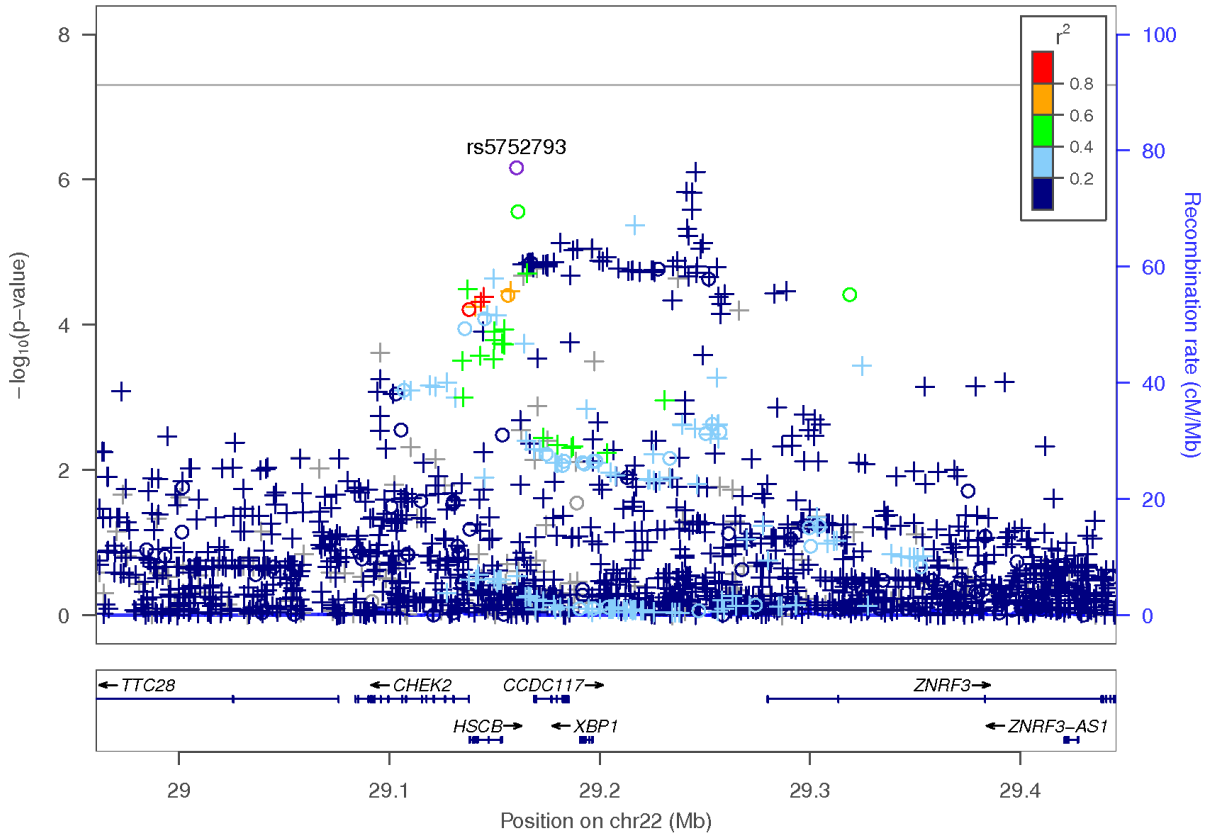

r²
0.8
0.6
0.4
0.2
rs3205718
−log₁₀(p−value)
Recombination rate (cM/Mb)
FMNL3
TMBIM6
NCKAP5L
FAIM2
AQP2
RACGAP1
ASIC1
BCDIN3D−AS1
LOC283332
AQP5
BCDIN3D
AQP6
50.1
50.2
50.3
50.4
Position on chr12 (Mb)

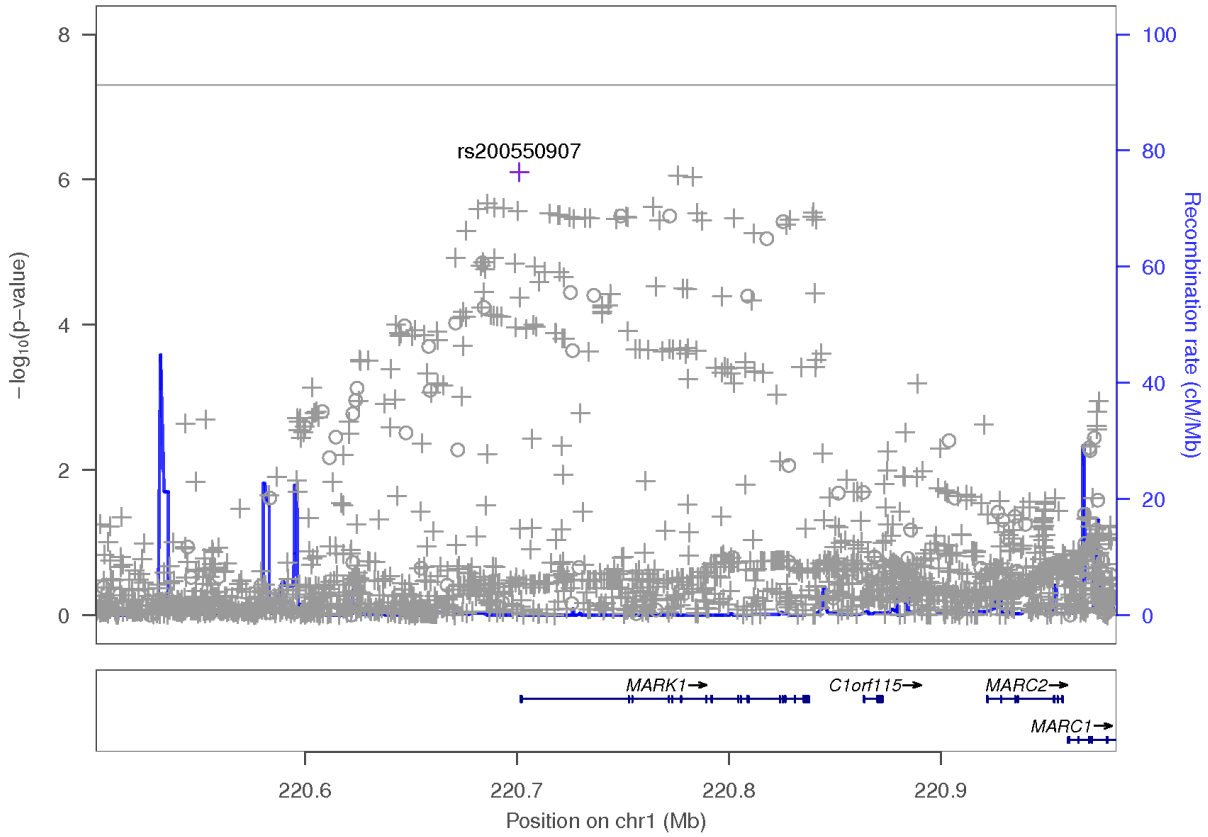

r²
0.8
0.6
0.4
0.2
rs148713408
−log₁₀(p−value)
Recombination rate (cM/Mb)
VPS13C
C2CD4A
62
62.1
62.2
62.3
62.4
Position on chr15 (Mb)

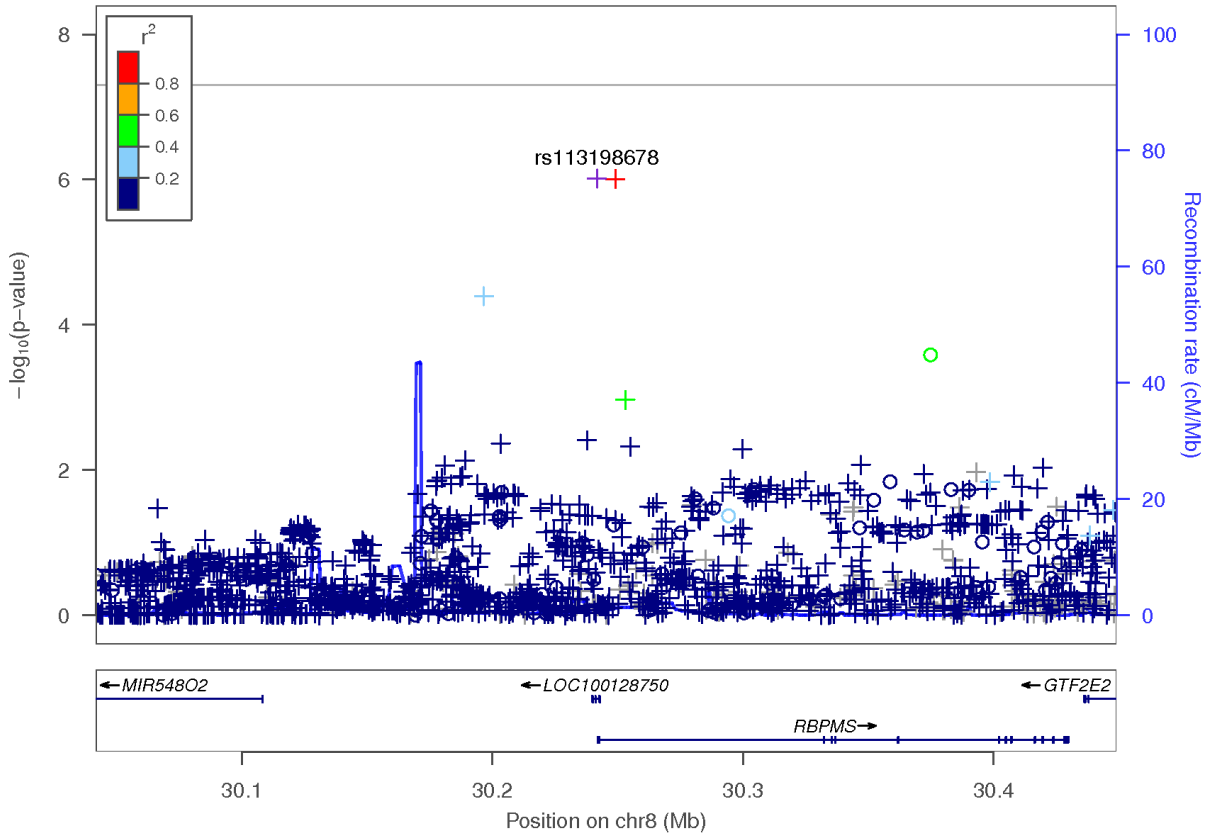

r²
0.8
0.6
0.4
0.2
rs616402
−log₁₀(p−value)
Recombination rate (cM/Mb)
KIF1B
PGD
CORT
PEX14
CASZ1
APITD1
APITD1−CORT
DFFA
10.4
10.5
10.6
10.7
10.8
Position on chr1 (Mb)


r²
0.8
0.6
0.4
0.2
rs62033406
−log10(p−value)
Recombination rate (cM/Mb)
RPGRIP1L
FTO
53.7
53.8
53.9
54
Position on chr16 (Mb)


r²
0.8
0.6
0.4
0.2
rs28929474
−log10(p−value)
Recombination rate (cM/Mb)
PPP4R4
SERPINA6
SERPINA1
SERPINA11
SERPINA12
SERPINA4
SERPINA10
SERPINA9
94.7
94.8
94.9
95
Position on chr14 (Mb)


rs5752793
r²
0.8
0.6
0.4
0.2
−log₁₀(p−value)
Recombination rate (cM/Mb)
TTC28
CHEK2
CCDC117
ZNRF3
HSCB
XBP1
ZNRF3−AS1
29
29.1
29.2
29.3
29.4
Position on chr22 (Mb)


rs200550907
−log₁₀(p−value)
Recombination rate (cM/Mb)
MARK1
C1orf115
MARC2
MARC1
220.6
220.7
220.8
220.9
Position on chr1 (Mb)


r²
0.8
0.6
0.4
0.2
rs113198678
−log₁₀(p−value)
Recombination rate (cM/Mb)
MIR548O2
LOC100128750
GTF2E2
RBPMS
30.1
30.2
30.3
30.4
Position on chr8 (Mb)