Analysis Summary: Hypermobility Beighton Score

Phenotype Description

The “beighton_hypermobility_qtl” phenotype comes from:

- “morphology” survey:
  - “Can you bring your right thumb to reach your forearm (see above image)?(right_thumb_flexibility)” (Yes, No, I’m not sure)
  - “Can you bring your left thumb to reach your forearm? (left_thumb_flexibility)” (Yes, No, I’m not sure)
  - “Can you extend your right forearm backwards to form a greater than 180 degree angle at your elbow (see above image)? (right_elbow_hyper)” (Yes, No, I’m not sure)
  - “Can you extend your left forearm backwards to form a greater than 180 degree angle at your elbow? (left_elbow_hyper)” (Yes, No, I’m not sure)
  - “Can you extend your lower left leg forward to form a greater than 180 degree angle at your knee (see above image)? (left_knee_hyper)” (Yes, No, I’m not sure)
  - “Can you extend your lower right leg forward to form a greater than 180 degree angle at your knee? (right_knee_hyper)” (Yes, No, I’m not sure)
  - “Can you bend down and place your entire palms on the floor while keeping your legs straight (see above image)? (palms_on_floor)” (Yes, No, I’m not sure)
  - “When you place your right hand flat on a table, can you lift your right little finger straight up to a vertical position or even further? (right_finger_flex)” (Yes, No, I’m not sure)
  - “When you place your left hand flat on a table, can you lift your left little finger straight up to a vertical position or even further? (left_finger_flex)” (Yes, No, I’m not sure)

Each question has three choices (yes, no, not sure). The “beighton_hypermobility_qtl” is the total counts of the positive answers to the 9 questions regarding joint mobility (beighton score). It was scored 0-9, higher scores signal greater hypermobility. We set NAs when people who didn’t answer.

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]
beighton_hypermobility_qtl	[0,1]	35942	22345	13597	3715	10312	9867	12048
	(1,2]	10202	5809	4393	1710	3639	2606	2247
	(2,3]	7029	2830	4199	1153	2479	1909	1488
	(3,5]	7676	3235	4441	1684	3129	1701	1162
	(5,9]	3294	973	2321	782	1308	728	476

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

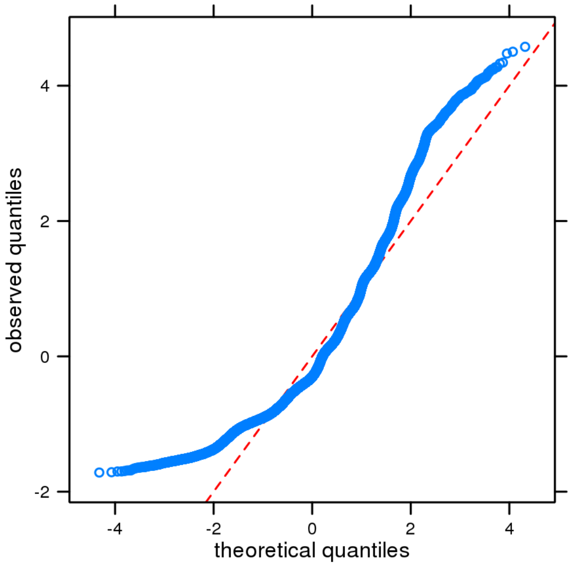
Phenotype	Group	Total	v1/v2	v3	v4
beighton_hypermobility_qtl	[0,1]	35942	4493	30033	1416
	(1,2]	10202	1168	8607	427
	(2,3]	7029	816	5862	351
	(3,5]	7676	826	6466	384
	(5,9]	3294	375	2734	185

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	−0.026858	0.000471	−57.1	0.0
sexF	0.931974	0.014818	62.9	0.0
pc.0	−0.000683	0.007378	−0.1	0.93
pc.1	0.010637	0.007409	1.4	0.15
pc.2	0.043504	0.007402	5.9	4.2×10^{-9}
pc.3	−0.003695	0.007384	−0.5	0.62
pc.4	−0.024697	0.007380	−3.3	0.00082

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	−17425	13761642
MAF < 0.1%	−1722150	12039492

Genetic Association Tests

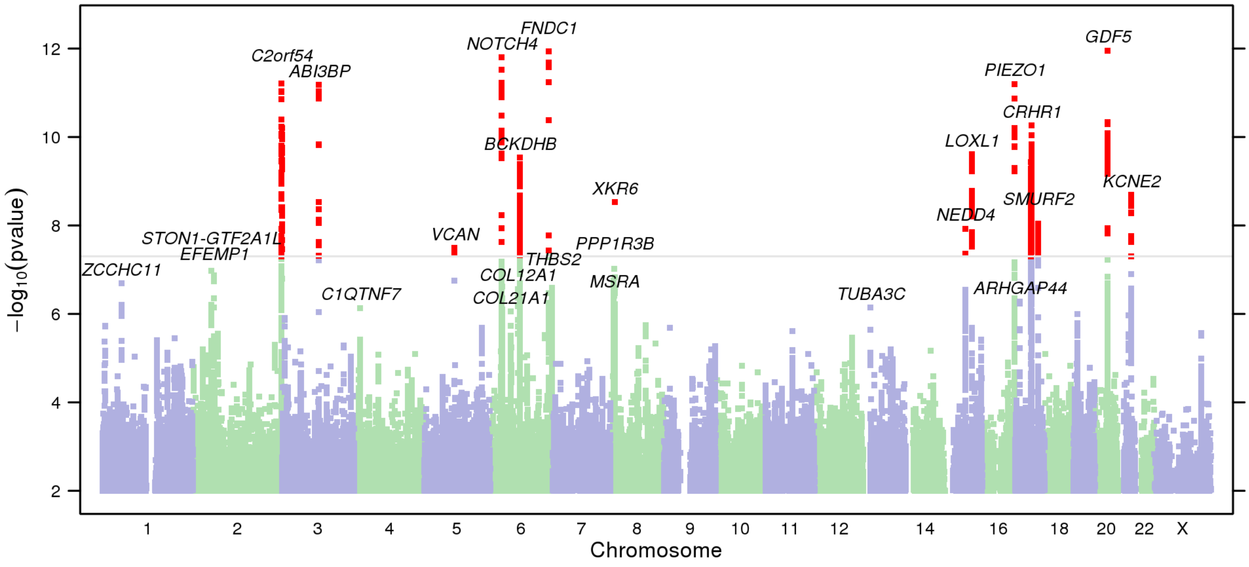
We performed linear regression assuming an additive model for allelic effects, using the model:

beighton_hypermobility_qtl ~ age + sex + pc.0 + pc.1 + pc.2 + pc.3 + pc.4 + genotype

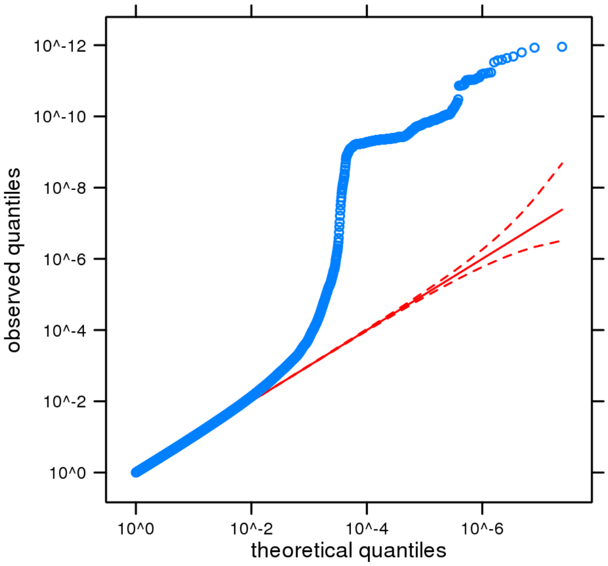
This genome-wide association analysis includes data from 64143 individuals of European ancestry, filtered to remove close relatives.

The results in this report have been adjusted for a genomic control inflation factor λ=1.090. The equivalent inflation factor rescaled for a sample size of 2000 would be λ_{2000}= 1.003 , and for 20000, λ_{20000}= 1.028 .

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
20q11.22	rs143384	chr20	34025756	A/G	I	1.1×10^{-12}	−0.079	[-0.100,-0.057]	[GDF5]
6q25.3	rs62432291	chr6	159621087	C/T	I	1.2×10^{-12}	−0.177	[-0.226,-0.128]	[FNDC1]
6p21.32	rs431722	chr6	32187722	C/T	I	1.6×10^{-12}	−0.080	[-0.102,-0.058]	[NOTCH4]
2q37.3	rs4417704	chr2	241846573	A/G	I	6.2×10^{-12}	0.087	[0.062,0.111]	C2orf54--[]--SNED1
16q24.3	rs78579285	chr16	88813060	C/T	I	6.3×10^{-12}	−0.104	[-0.133,-0.074]	[PIEZO1]
3q12.2	rs9828061	chr3	100559959	C/T	I	6.7×10^{-12}	0.078	[0.056,0.101]	[ABI3BP]
17q21.31	rs113518470	chr17	43850932	C/T	I	5.5×10^{-11}	−0.090	[-0.116,-0.063]	LRRC37A3---[]--CRHR1
15q24.1	rs150025731	chr15	74223500	D/I	I	2.5×10^{-10}	−0.075	[-0.098,-0.052]	[LOXL1]
6q14.1	rs4706831	chr6	81007699	C/T	I	2.9×10^{-10}	−0.069	[-0.090,-0.047]	[BCKDHB]
21q22.11	rs1467847	chr21	35714544	C/G	I	2.0×10^{-9}	0.066	[0.044,0.088]	MRPS6---[]--KCNE2
8p23.1	rs10109025	chr8	10815571	C/G	I	3.0×10^{-9}	−0.068	[-0.090,-0.045]	[XKR6]
17q24.1	rs6504249	chr17	62676149	A/G	I	9.2×10^{-9}	−0.072	[-0.097,-0.048]	SMURF2--[]---PLEKHM1P
15q21.3	rs13380185	chr15	56116563	G/T	I	1.2×10^{-8}	0.070	[0.046,0.094]	PRTG--[]-NEDD4
5q14.3	rs34580448	chr5	82810884	C/T	I	3.2×10^{-8}	0.156	[0.101,0.211]	[VCAN]
8p23.1	rs453301	chr8	9030387	G/T	I	9.6×10^{-8}	−0.059	[-0.080,-0.037]	PPP1R3B--[]---TNKS
2p16.3	rs77637988	chr2	48775583	C/G	I	1.0×10^{-7}	−0.071	[-0.096,-0.045]	PPP1R21--[]--STON1-GTF2A1L
2p16.1	rs3791679	chr2	56096892	A/G	I	1.4×10^{-7}	−0.067	[-0.092,-0.042]	[EFEMP1]
1p32.3	rs11581903	chr1	53030823	A/G	I	2.1×10^{-7}	0.078	[0.049,0.108]	ZCCHC11--[]--GPX7
6q27	rs1040	chr6	169616347	C/T	I	2.5×10^{-7}	−0.057	[-0.078,-0.035]	[THBS2]
6q14.1	rs41269313	chr6	75904844	C/G	I	2.7×10^{-7}	−0.108	[-0.149,-0.067]	[COL12A1]
8p23.1	rs187276360	chr8	9931621	C/G	I	3.8×10^{-7}	5.025	[3.087,6.963]	[MSRA]
17p12	rs8078472	chr17	12873387	A/G	I	5.3×10^{-7}	−0.089	[-0.123,-0.054]	[ARHGAP44]

13q11	rs12584385	chr13	19303482	A/C	I	7.1×10^{-7}	0.077	[0.046,0.107]	[]---TUBA3C
4p15.32	rs62289301	chr4	15449024	G/T	I	7.3×10^{-7}	−0.070	[-0.097,-0.042]	C1QTNF7-[]--CC2D2A
6p12.1	rs62413470	chr6	55999537	A/G	I	8.7×10^{-7}	0.074	[0.045,0.104]	[COL21A1]

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
rs143384	FALSE	TRUE	TRUE	0.9995	0.011	0.018	0.4278	0.9938	0.9381	0.025	0.4291	v2v3v4
rs62432291	FALSE	FALSE	FALSE					0.9144	0.8579	0.0067	0.0535	v2v3v4
rs431722	FALSE	FALSE	FALSE					0.9801	0.9750	0.0068	0.6106	v2v3v4
rs4417704	FALSE	FALSE	FALSE					0.9586	0.9461	0.035	0.7334	v2v3v4
rs78579285	FALSE	FALSE	FALSE					0.8695	0.7477	0.039	0.1734	v2v3v4
rs9828061	FALSE	FALSE	FALSE					0.9810	0.9717	0.18	0.6333	v2v3v4
rs113518470	FALSE	FALSE	FALSE					0.9395	0.9343	0.40	0.2167	v2v3v4
rs150025731	FALSE	FALSE	FALSE					0.9749	0.9729	0.96	0.6914	v2v3v4
rs4706831	FALSE	FALSE	FALSE					0.9935	0.9920	0.47	0.5508	v2v3v4
rs1467847	FALSE	FALSE	FALSE					0.9899	0.9855	0.14	0.4362	v2v3v4
rs10109025	FALSE	FALSE	FALSE					0.9313	0.9001	0.41	0.4121	v2v3v4
rs6504249	FALSE	TRUE	FALSE	0.9995	0.077	0.84	0.7377	0.9663	0.7375	2.6×10^{-33}	0.7379	v2v3v4
rs13380185	FALSE	TRUE	FALSE	0.9995	0.18	0.38	0.7275	0.9830	0.9220	0.00010	0.7260	v2v3v4
rs34580448	FALSE	FALSE	FALSE					0.8823	0.8642	0.019	0.9563	v2v3v4
rs453301	FALSE	FALSE	FALSE					0.9728	0.9590	0.55	0.5099	v2v3v4
rs77637988	FALSE	FALSE	FALSE					0.9563	0.9206	0.031	0.7729	v2v3v4
rs3791679	TRUE	TRUE	TRUE	0.9990	0.13	0.81	0.2376	0.9996	0.9993	0.12	0.2390	v2v3v4
rs11581903	FALSE	TRUE	FALSE	0.9501	0.0014	3.1×10^{-12}	0.8486	0.9949	0.9811	0.66	0.8511	v2v3v4
rs1040	FALSE	FALSE	FALSE					0.9955	0.9891	0.89	0.4279	v2v3v4
rs41269313	FALSE	FALSE	FALSE					0.9156	0.8991	0.014	0.9211	v2v3v4
rs187276360	FALSE	FALSE	FALSE					0.5008	0.4719	0.41	0.0024	v4
rs8078472	TRUE	TRUE	TRUE	0.9995	0.0060	0.39	0.1064	0.9997	0.9990	0.21	0.1063	v2v3v4
rs12584385	TRUE	TRUE	TRUE	0.9991	0.74	0.55	0.8549	0.9976	0.9888	0.16	0.8543	v2v3v4
rs62289301	FALSE	FALSE	FALSE					0.9323	0.9225	0.52	0.8041	v2v3v4
rs62413470	FALSE	FALSE	FALSE					0.9801	0.9777	2.4×10^{-5}	0.8418	v2v3v4

SNP Statistics in the GWAS Sample

assay.name	AA.0	AB.0	BB.0	im.num.0	dose.b.0
rs143384	18503	27527	10419	64143	0.4280
rs62432291				64143	0.0543
rs431722				64143	0.6048
rs4417704				64143	0.7362
rs78579285				64143	0.1756
rs9828061				64143	0.6349
rs113518470				64143	0.2168
rs150025731				64143	0.6882
rs4706831				64143	0.5524
rs1467847				64143	0.4330
rs10109025				64143	0.4105
rs6504249	3703	20897	29368	64143	0.7361
rs13380185	4065	21417	28207	64143	0.7233
rs34580448				64143	0.9564
rs453301				64143	0.5095
rs77637988				64143	0.7707
rs3791679	37406	23037	3604	64143	0.2361
rs11581903	1273	13577	37143	64143	0.8482
rs1040				64143	0.4269
rs41269313				64143	0.9214
rs187276360				2763	0.0017
rs8078472	51169	12232	704	64143	0.1064
rs12584385	1333	15977	46803	64143	0.8545
rs62289301				64143	0.8025
rs62413470				64143	0.8435

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
20q11.22	34025756	rs143384	1.1×10^{-12}	−118595	0.778	rs6060369	1.0×10^{-16}	18391950	Height	GDF5,UQCC
20q11.22	34025756	rs143384	1.1×10^{-12}	−118595	0.778	rs6060369	2.0×10^{-16}	18193045	Height	BFZB
20q11.22	34025756	rs143384	1.1×10^{-12}	−115972	0.532	rs6088792	8.0×10^{-7}	18391951	Height	UQCC, GDF5, …
20q11.22	34025756	rs143384	1.1×10^{-12}	−111548	0.789	rs6060373	2.0×10^{-7}	23207799	Spine bone size	UQCC
20q11.22	34025756	rs143384	1.1×10^{-12}	−111548	0.789	rs6060373	2.0×10^{-17}	18391952	Height	GDF5
20q11.22	34025756	rs143384	1.1×10^{-12}	−50575	0.831	rs6088813	1.0×10^{-13}	19343178	Height	UQCC
20q11.22	34025756	rs143384	1.1×10^{-12}	−1794	0.836	rs224333	8.0×10^{-37}	23563607	Height	GDF5
20q11.22	34025756	rs143384	1.1×10^{-12}	0	1.000	rs143384	1.0×10^{-58}	20881960	Height	GDF5
17q21.31	43850932	rs113518470	5.5×10^{-11}	−337491	0.668	rs11012	6.0×10^{-8}	20070850	Parkinson’s disease	PLEKHM1, MAPT, IMP5
17q21.31	43850932	rs113518470	5.5×10^{-11}	−334530	0.668	rs17631303	1.0×10^{-8}	23544013	Ovarian cancer in BRCA1 mutation carriers	
17q21.31	43850932	rs113518470	5.5×10^{-11}	−136082	0.934	rs2942168	1.0×10^{-28}	21292315	Parkinson’s disease	MAPT
17q21.31	43850932	rs113518470	5.5×10^{-11}	−131789	0.927	rs393152	2.0×10^{-16}	19915575	Parkinson’s disease	MAPT, C17orf69, …
17q21.31	43850932	rs113518470	5.5×10^{-11}	72751	0.934	rs12185268	3.0×10^{-14}	21738487	Parkinson’s disease	MAPT
17q21.31	43850932	rs113518470	5.5×10^{-11}	73287	0.926	rs12373124	5.0×10^{-10}	22693459	Male-pattern baldness	Intergenic
17q21.31	43850932	rs113518470	5.5×10^{-11}	126895	0.934	rs1864325	5.0×10^{-11}	22504420	Bone mineral density	MAPT

17q21.31	43850932	rs113518470	5.5×10^{-11}	205835	0.934	rs1981997	9.0×10^{-14}	23583980	Interstitial lung disease	MAPT
17q21.31	43850932	rs113518470	5.5×10^{-11}	230132	0.926	rs8070723	2.0×10^{-118}	21685912	Progressive supranuclear palsy	MAPT
17q21.31	43850932	rs113518470	5.5×10^{-11}	230132	0.926	rs8070723	7.0×10^{-12}	21044948	Parkinson's disease	MAPT
17q21.31	43850932	rs113518470	5.5×10^{-11}	336325	0.906	rs9303525	8.0×10^{-15}	22504418	Intracranial volume	MAPT, GRN, ...
15q24.1	74223500	rs150025731	2.5×10^{-10}	5565	0.897	rs893817	3.0×10^{-6}	19584346	Aortic root size	LOXL1
6q14.1	81007699	rs4706831	2.9×10^{-10}	−161710	0.696	rs2322633	3.0×10^{-9}	23563607	Height	BCKDHB
21q22.11	35714544	rs1467847	2.0×10^{-9}	−23758	0.619	rs2834442	5.0×10^{-12}	20881960	Height	KCNE2
21q22.11	35714544	rs1467847	2.0×10^{-9}	−16154	0.900	rs1013063	5.0×10^{-6}	23192594	Body mass index (interaction)	LOC100130240, KCNE2
15q21.3	56116563	rs13380185	1.2×10^{-8}	78314	0.645	rs8032158	6.0×10^{-13}	20711176	Keloid	NEDD4
8p23.1	9030387	rs453301	9.6×10^{-8}	−140289	0.527	rs1045529	3.0×10^{-6}	23049088	Myopia (pathological)	Intergenic
2p16.3	48775583	rs77637988	1.0×10^{-7}	−129184	0.536	rs10454142	1.0×10^{-7}	22829776	Sex hormone-binding globulin levels	LHCGR
2p16.1	56096892	rs3791679	1.4×10^{-7}	0	1.000	rs3791679	8.0×10^{-16}	23563607	Height	EFEMP1
2p16.1	56096892	rs3791679	1.4×10^{-7}	0	1.000	rs3791679	6.0×10^{-9}	20189936	Height	EFEMP1
2p16.1	56096892	rs3791679	1.4×10^{-7}	0	1.000	rs3791679	6.0×10^{-11}	18391951	Height	EFEMP1, PNPT1
2p16.1	56096892	rs3791679	1.4×10^{-7}	14417	0.993	rs3791675	3.0×10^{-35}	20881960	Height	EFEMP1
2p16.1	56096892	rs3791679	1.4×10^{-7}	14417	0.993	rs3791675	3.0×10^{-7}	20397748	Height	EFEMP1
2p16.1	56096892	rs3791679	1.4×10^{-7}	14417	0.993	rs3791675	4.0×10^{-6}	19893584	Height	EFEMP1
2p16.1	56096892	rs3791679	1.4×10^{-7}	14417	0.993	rs3791675	2.0×10^{-9}	19396169	Height	EFEMP1
2p16.1	56096892	rs3791679	1.4×10^{-7}	14417	0.993	rs3791675	2.0×10^{-12}	18391952	Height	EFEMP1
4p15.32	15449024	rs62289301	7.3×10^{-7}	96386	0.503	rs9918079	6.0×10^{-6}	23251661	Obesity-related traits	CC2D2A

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, $r^2 > 0.8$).

no relevant GWAS catalog terms were available

Nearby Nonsynonymous SNPs

region	position	our.name	our.pval	dist	rsqr	assay.name	gene	aa.chg
20q11.22	34025756	rs143384	1.1×10^{-12}	−53842	0.831	rs4911494	UQCC	R51Q
20q11.22	34025756	rs143384	1.1×10^{-12}	−3369	0.785	rs224331	GDF5	S276A
6q25.3	159621087	rs62432291	1.2×10^{-12}	0	1.000	rs62432291	FNDC1	P122S
6q25.3	159621087	rs62432291	1.2×10^{-12}	31844	0.513	rs420137	FNDC1	E463Q
6q25.3	159621087	rs62432291	1.2×10^{-12}	33464	0.504	rs370434	FNDC1	Q1003E
6q25.3	159621087	rs62432291	1.2×10^{-12}	33997	0.504	rs420054	FNDC1	D1180E
6q25.3	159621087	rs62432291	1.2×10^{-12}	34239	0.522	rs3003174	FNDC1	L1261P
6q25.3	159621087	rs62432291	1.2×10^{-12}	34296	0.522	rs2501176	FNDC1	Q1280R
6q25.3	159621087	rs62432291	1.2×10^{-12}	39792	0.522	rs386360	FNDC1	T1504K
6p21.32	32187722	rs431722	1.6×10^{-12}	661	0.562	rs422951	NOTCH4	T320A
16q24.3	88813060	rs78579285	6.3×10^{-12}	−32885	0.539	rs4782321	CTU2	V332I
16q24.3	88813060	rs78579285	6.3×10^{-12}	−31987	0.532	rs11549835	CTU2	R427Q
16q24.3	88813060	rs78579285	6.3×10^{-12}	−12662	0.651	rs144777557	PIEZO1	?749?
16q24.3	88813060	rs78579285	6.3×10^{-12}	−12275	0.651	rs35265318	PIEZO1	R720H
17q21.31	43850932	rs113518470	5.5×10^{-11}	−343635	0.573	rs12949256	ARHGAP27	A117T
17q21.31	43850932	rs113518470	5.5×10^{-11}	72010	0.934	rs62621252	SPPL2C	S224P
17q21.31	43850932	rs113518470	5.5×10^{-11}	72334	0.934	rs62054815	SPPL2C	A332T
17q21.31	43850932	rs113518470	5.5×10^{-11}	72722	0.934	rs12185233	SPPL2C	R461P
17q21.31	43850932	rs113518470	5.5×10^{-11}	72751	0.934	rs12185268	SPPL2C	I471V
17q21.31	43850932	rs113518470	5.5×10^{-11}	73141	0.934	rs12373123	SPPL2C	S601P
17q21.31	43850932	rs113518470	5.5×10^{-11}	73198	0.934	rs12373139	SPPL2C	G620R
17q21.31	43850932	rs113518470	5.5×10^{-11}	73268	0.926	rs12373142	SPPL2C	P643R
17q21.31	43850932	rs113518470	5.5×10^{-11}	73287	0.926	rs12373124	SPPL2C	H649H
17q21.31	43850932	rs113518470	5.5×10^{-11}	73299	0.926	rs12373140	SPPL2C	Q653Q
17q21.31	43850932	rs113518470	5.5×10^{-11}	209843	0.934	rs63750417	MAPT	P202L
17q21.31	43850932	rs113518470	5.5×10^{-11}	210091	0.934	rs62063786	MAPT	D285N
17q21.31	43850932	rs113518470	5.5×10^{-11}	210104	0.934	rs62063787	MAPT	V289A
17q21.31	43850932	rs113518470	5.5×10^{-11}	210346	0.934	rs17651549	MAPT	R370W
17q21.31	43850932	rs113518470	5.5×10^{-11}	216468	0.934	rs10445337	MAPT	S447P
17q21.31	43850932	rs113518470	5.5×10^{-11}	253479	0.910	rs199706121	MAPT	?1735?
17q21.31	43850932	rs113518470	5.5×10^{-11}	257554	0.717	rs200384907	KANSL1	?1225?
17q21.31	43850932	rs113518470	5.5×10^{-11}	257974	0.934	rs34579536	KANSL1	I1085T
17q21.31	43850932	rs113518470	5.5×10^{-11}	259600	0.934	rs36076725	KANSL1	F917F
17q21.31	43850932	rs113518470	5.5×10^{-11}	259609	0.934	rs35833914	KANSL1	D914D
17q21.31	43850932	rs113518470	5.5×10^{-11}	266187	0.934	rs34043286	KANSL1	S718P
17q21.31	43850932	rs113518470	5.5×10^{-11}	397905	0.765	rs35643216	KANSL1	N225D
17q21.31	43850932	rs113518470	5.5×10^{-11}	398267	0.765	rs17585974	KANSL1	K104T
15q24.1	74223500	rs150025731	2.5×10^{-10}	−3954	0.952	rs1048661	LOXL1	R141L
5q14.3	82810884	rs34580448	3.2×10^{-8}	4286	1.000	rs61749613	VCAN	K349E

Nearby Expression QTLs

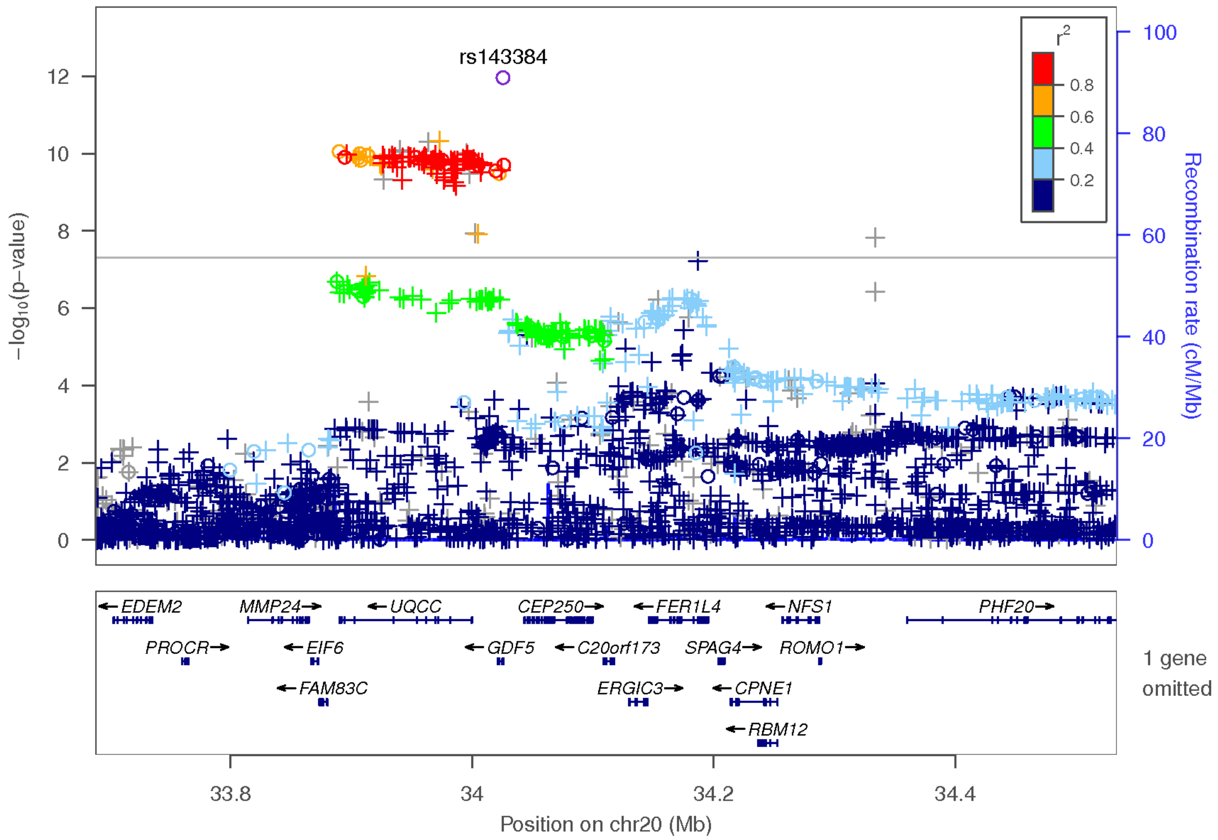
region	position	our.name	our.pval	dist	rsqr	assay.name	eqtl.dist	eqtl.gene	eqtl.pval	eqtl.rsqr	tissue	pubmed.id

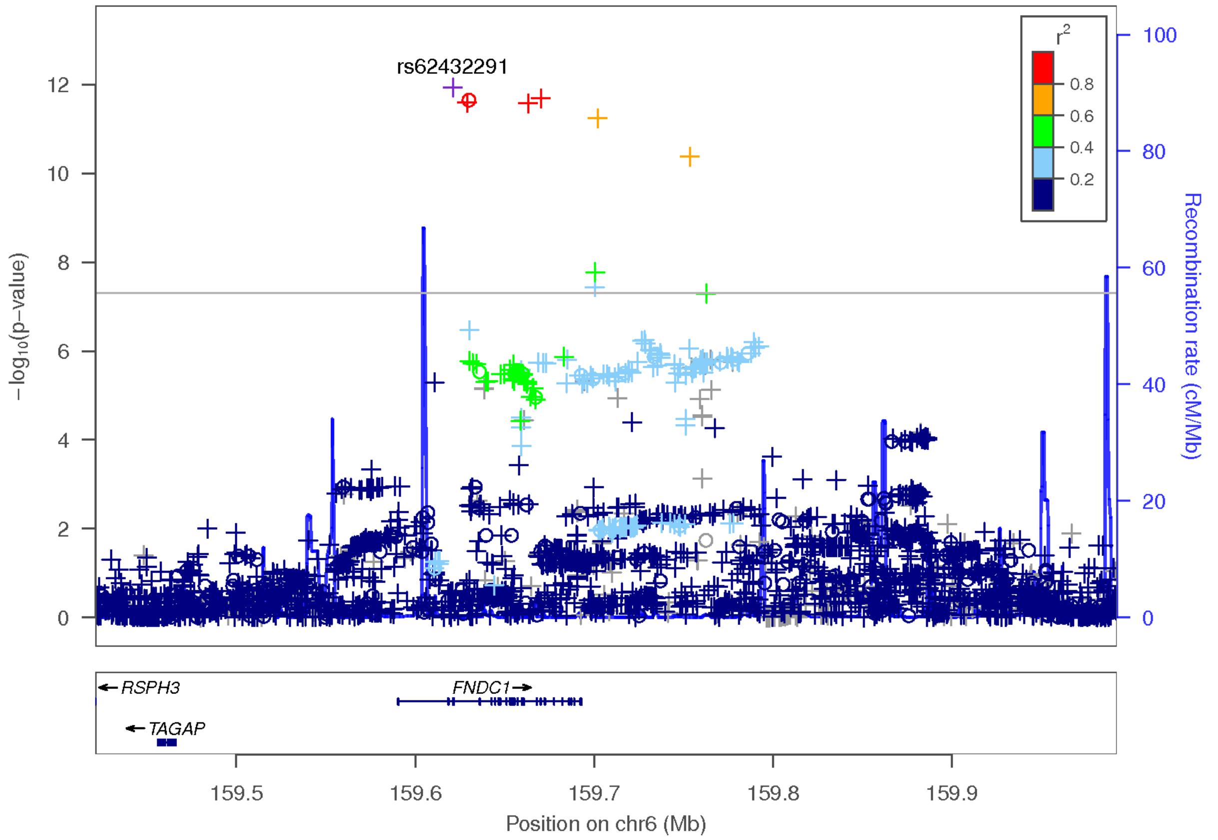
20q11.22	34025756	rs143384	1.1×10^{-12}	−137773	0.524	rs13038012	326158	CPNE1	0.00012	0.052	B-Cell	22446964
20q11.22	34025756	rs143384	1.1×10^{-12}	−115972	0.532	rs6088792	304357	CPNE1	6.0×10^{-6}	0.071	Monocyte	22446964
20q11.22	34025756	rs143384	1.1×10^{-12}	−73136	0.832	rs4911178	−61666	UQCC	6.0×10^{-8}	0.100	Monocyte	22446964
20q11.22	34025756	rs143384	1.1×10^{-12}	−67503	0.832	rs1570004	0	UQCC	2.5×10^{-47}	0.134	Monocyte	20502693
6p21.32	32187722	rs431722	1.6×10^{-12}	−3017	0.541	rs3134798	−580164	BAT2	0.00019	0.048	Monocyte	22446964
2q37.3	241846573	rs4417704	6.2×10^{-12}	−336989	0.568	rs10207380	394926	SEPT2	9.8×10^{-5}		Lymphoblastoid	20220756
16q24.3	88813060	rs78579285	6.3×10^{-12}	−31210	0.532	rs1061238	0	FAM38A	2.8×10^{-21}	0.062	Monocyte	20502693
17q21.31	43850932	rs113518470	5.5×10^{-11}	−337491	0.668	rs11012	201708	MGC57346	6.7×10^{-23}	0.297	B-Cell	22446964
17q21.31	43850932	rs113518470	5.5×10^{-11}	−301406	0.682	rs17631676	42490	LRRC37A4	6.0×10^{-208}	0.475	Monocyte	20502693
17q21.31	43850932	rs113518470	5.5×10^{-11}	−301406	0.682	rs17631676	165623	MGC57346	1.8×10^{-59}	0.613	Monocyte	22446964
17q21.31	43850932	rs113518470	5.5×10^{-11}	−301406	0.682	rs17631676	378322	HS.554608	0.00057	0.042	B-Cell	22446964
17q21.31	43850932	rs113518470	5.5×10^{-11}	−301406	0.682	rs17631676	170002	C17ORF69	7.7×10^{-5}	0.055	B-Cell	22446964
17q21.31	43850932	rs113518470	5.5×10^{-11}	−131789	0.927	rs393152	580720	NMT1	0.00045	0.156	Lymphoblastoid	19644074
17q21.31	43850932	rs113518470	5.5×10^{-11}	5407	0.550	rs117631329	446394	KANSL1	5.3×10^{-8}	0.077	Lymphoblastoid	24037378
17q21.31	43850932	rs113518470	5.5×10^{-11}	462337	0.587	rs140833087	354766	NSF	3.5×10^{-8}	0.079	Lymphoblastoid	24037378
17q21.31	43850932	rs113518470	5.5×10^{-11}	483356	0.875	rs142039218	666195	GOSR2	7.4×10^{-13}	0.130	Lymphoblastoid	24037378
17q21.31	43850932	rs113518470	5.5×10^{-11}	495738	0.730	rs139480590	647403	CRHR1	2.4×10^{-11}	0.113	Lymphoblastoid	24037378
6q14.1	81007699	rs4706831	2.9×10^{-10}	−115272	0.531	rs2490245	697719	LCA5	0.00043	0.158	Fibroblast	19644074
21q22.11	35714544	rs1467847	2.0×10^{-9}	−24045	0.651	rs2834440	−1558214	C21ORF66	0.0012	0.037	B-Cell	22446964
8p23.1	10815571	rs10109025	3.0×10^{-9}	3086	0.567	rs11250099	841532	FDFT1	3.0×10^{-9}	0.026	Monocyte	20502693
8p23.1	10815571	rs10109025	3.0×10^{-9}	3086	0.567	rs11250099	532863	BLK	1.6×10^{-6}	0.018	Monocyte	20502693
8p23.1	10815571	rs10109025	3.0×10^{-9}	4283	0.576	rs4841465	459280	C8ORF13	6.1×10^{-9}	0.114	Monocyte	22446964
8p23.1	10815571	rs10109025	3.0×10^{-9}	4283	0.576	rs4841465	−2072223	MFHAS1	2.1×10^{-6}	0.077	B-Cell	22446964
8p23.1	10815571	rs10109025	3.0×10^{-9}	16297	0.524	rs10096421	447113	C8orf13	0.00033	0.163	Lymphoblastoid	19644074
8p23.1	9030387	rs453301	9.6×10^{-8}	−200551	0.504	rs6983150	203154	PPP1R3B	2.6×10^{-15}	0.260	Lymphoblastoid	17873874
8p23.1	9030387	rs453301	9.6×10^{-8}	−119079	0.504	rs3989373	−349821	CLDN23	3.8×10^{-6}	0.074	Monocyte	22446964
8p23.1	9030387	rs453301	9.6×10^{-8}	−974	0.953	rs435581	−2285766	DEFA4	0.00052	0.042	B-Cell	22446964
8p23.1	9030387	rs453301	9.6×10^{-8}	−974	0.953	rs435581	−281782	MFHAS1	4.6×10^{-7}	0.087	B-Cell	22446964
8p23.1	9030387	rs453301	9.6×10^{-8}	54908	0.664	rs2929451	334164	MFHAS1	2.2×10^{-10}	0.029	Monocyte	20502693
1p32.3	53030823	rs11581903	2.1×10^{-7}	−415965	0.613	rs1983833	−968463	EPS15	0.00041		Lymphoblastoid	20220756
1p32.3	53030823	rs11581903	2.1×10^{-7}	−275952	0.691	rs1288706	−256807	KTI12	0.0013	0.036	B-Cell	22446964
1p32.3	53030823	rs11581903	2.1×10^{-7}	−234573	0.624	rs2762821	278078	GPX7	3.0×10^{-10}	0.133	Monocyte	22446964
17p12	12873387	rs8078472	5.3×10^{-7}	−4808	0.578	rs11655654	27443	ELAC2	0.00010	0.053	B-Cell	22446964

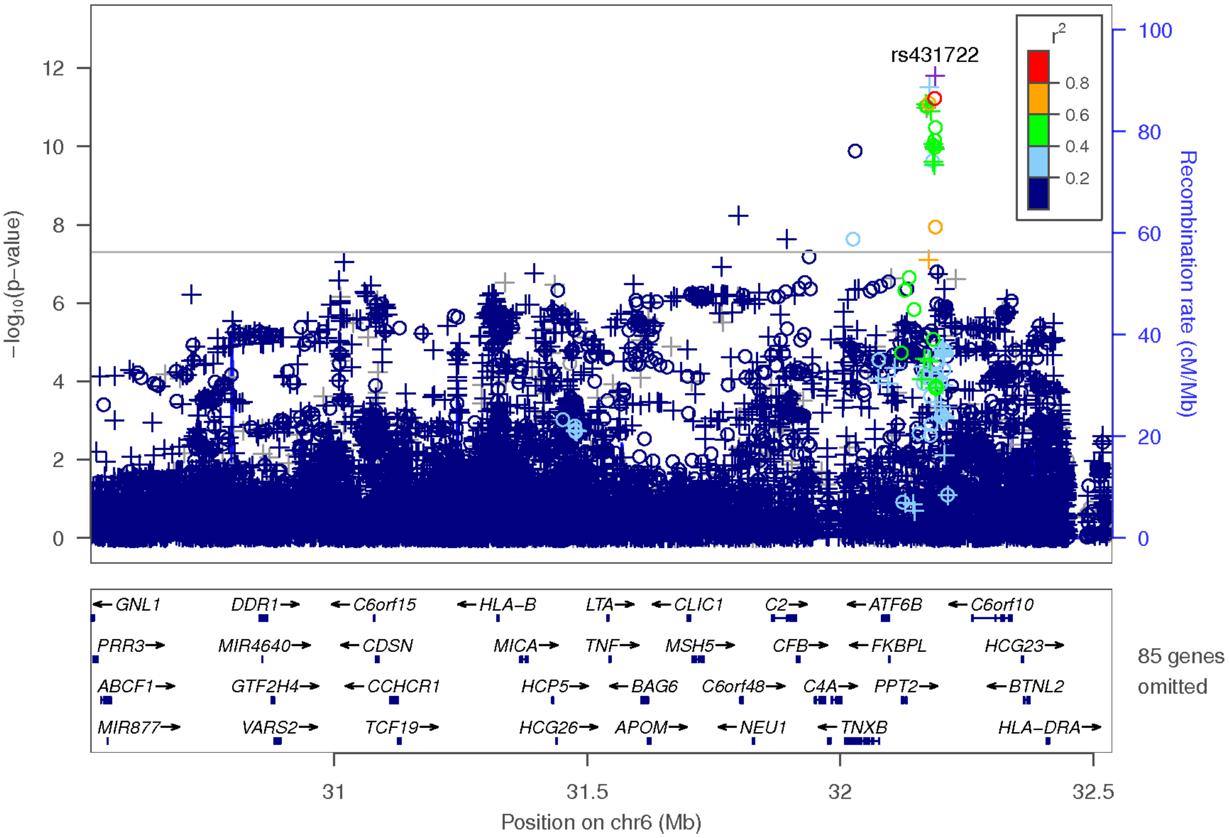
Nearby Clinical Variants

source	region	our.name	our.pval	dist	rsqr	assay.name	gene	phenotype	accession
clinvar	15q24.1	rs150025731	2.5×10^{-10}	−3954	0.952	rs1048661	LOXL1	Pseudoexfoliation glaucoma	SNOMED CT111514006
clinvar	6q14.1	rs4706831	2.9×10^{-10}	−170460	0.860	rs9448893	BCKDHB	Maple syrup urine disease	Office of Rare Diseases3228
clinvar	6q14.1	rs4706831	2.9×10^{-10}	−170460	0.860	rs9448893	BCKDHB	Maple syrup urine disease	SNOMED CT27718001
clinvar	6q14.1	rs4706831	2.9×10^{-10}	−170460	0.860	rs9448893	BCKDHB	Maple syrup urine disease type 1B	Office of Rare Diseases8597
clinvar	6q14.1	rs4706831	2.9×10^{-10}	−94953	0.527	rs3828753	BCKDHB	Maple syrup urine disease	Office of Rare Diseases3228
clinvar	6q14.1	rs4706831	2.9×10^{-10}	−94953	0.527	rs3828753	BCKDHB	Maple syrup urine disease	SNOMED CT27718001
clinvar	6q14.1	rs4706831	2.9×10^{-10}	−94953	0.527	rs3828753	BCKDHB	Maple syrup urine disease type 1B	Office of Rare Diseases8597

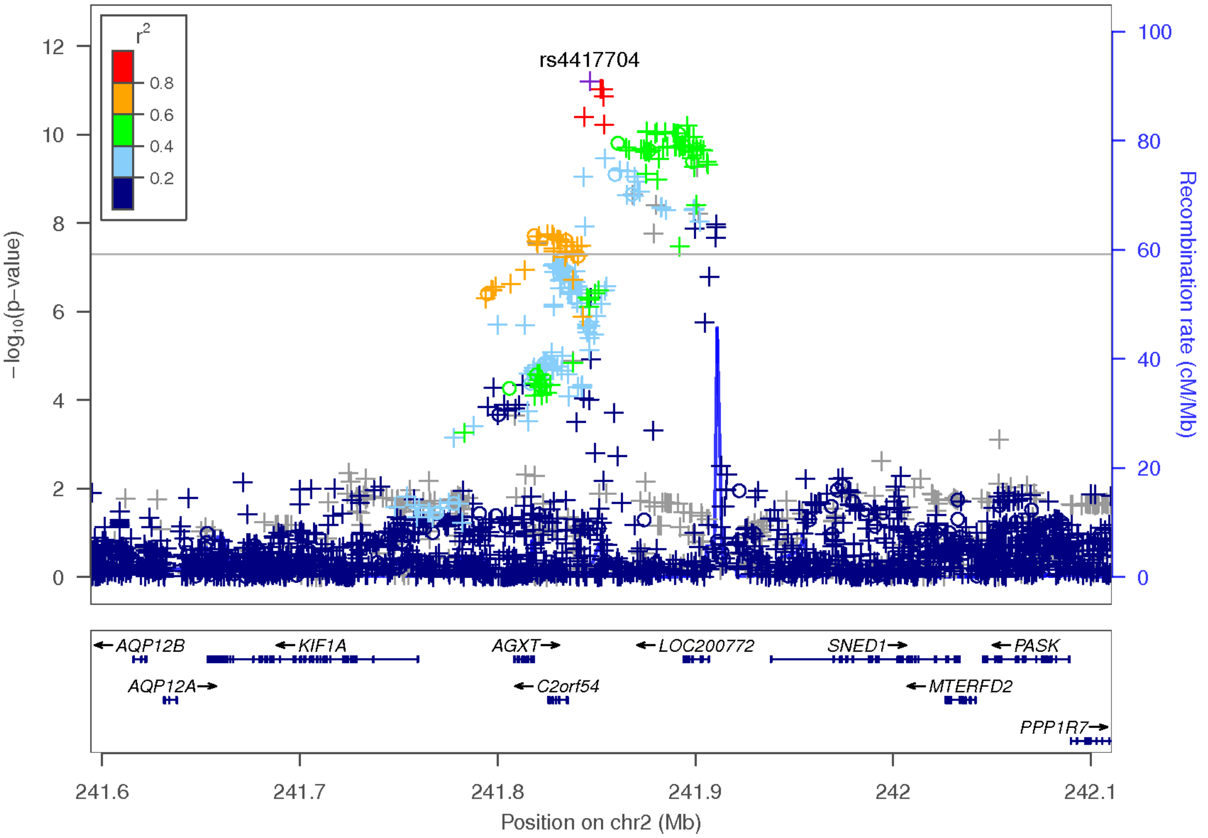
Regional Association Plots


rs143384
r²
0.8
0.6
0.4
0.2
−log₁₀(p−value)
0
2
4
6
8
10
12
Recombination rate (cM/Mb)
0
20
40
60
80
100
EDEM2
MMP24
UQCC
CEP250
FER1L4
NFS1
PHF20
PROCR
EIF6
GDF5
C20orf173
SPAG4
ROMO1
FAM83C
ERGIC3
CPNE1
RBM12
1 gene omitted
33.8
34
34.2
34.4
Position on chr20 (Mb)

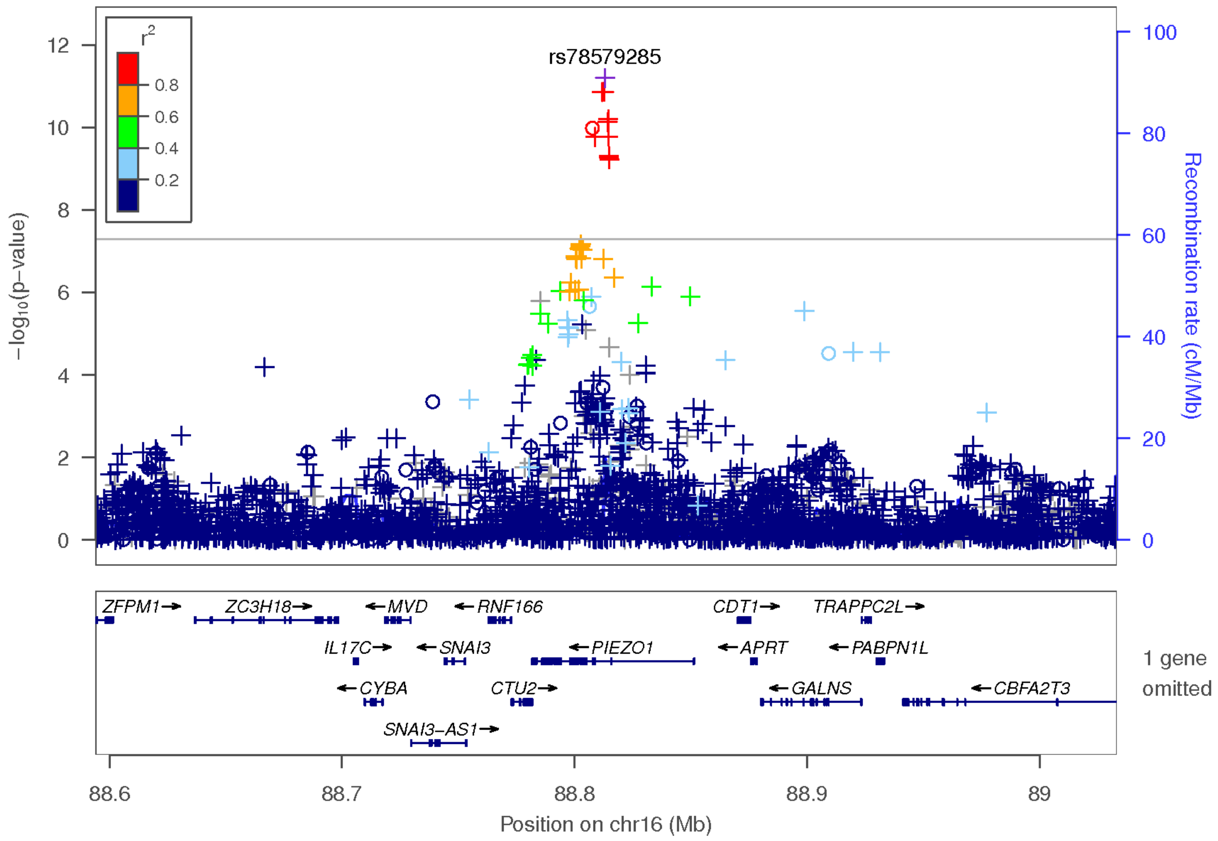

rs62432291
r²
0.8
0.6
0.4
0.2
−log₁₀(p−value)
0
2
4
6
8
10
12
Recombination rate (cM/Mb)
0
20
40
60
80
100
RSPH3
FNDC1
TAGAP
159.5
159.6
159.7
159.8
159.9
Position on chr6 (Mb)

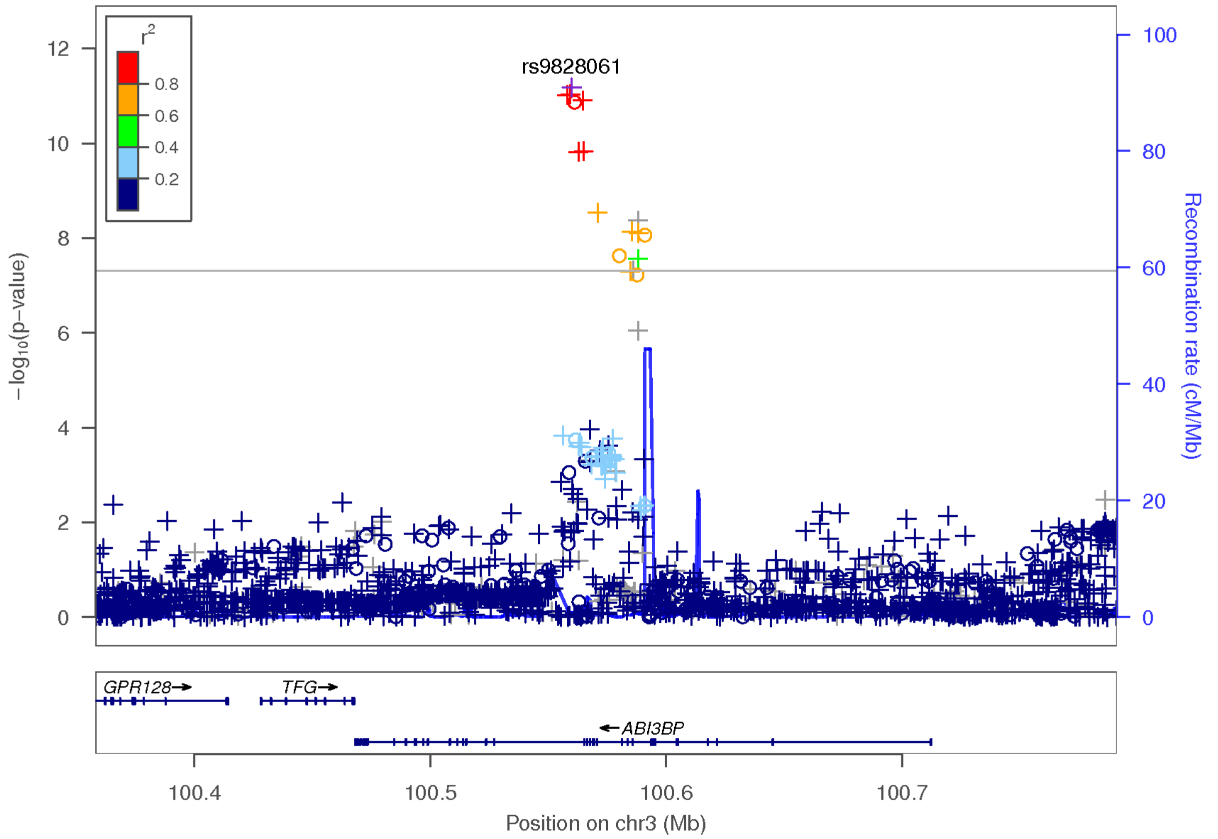


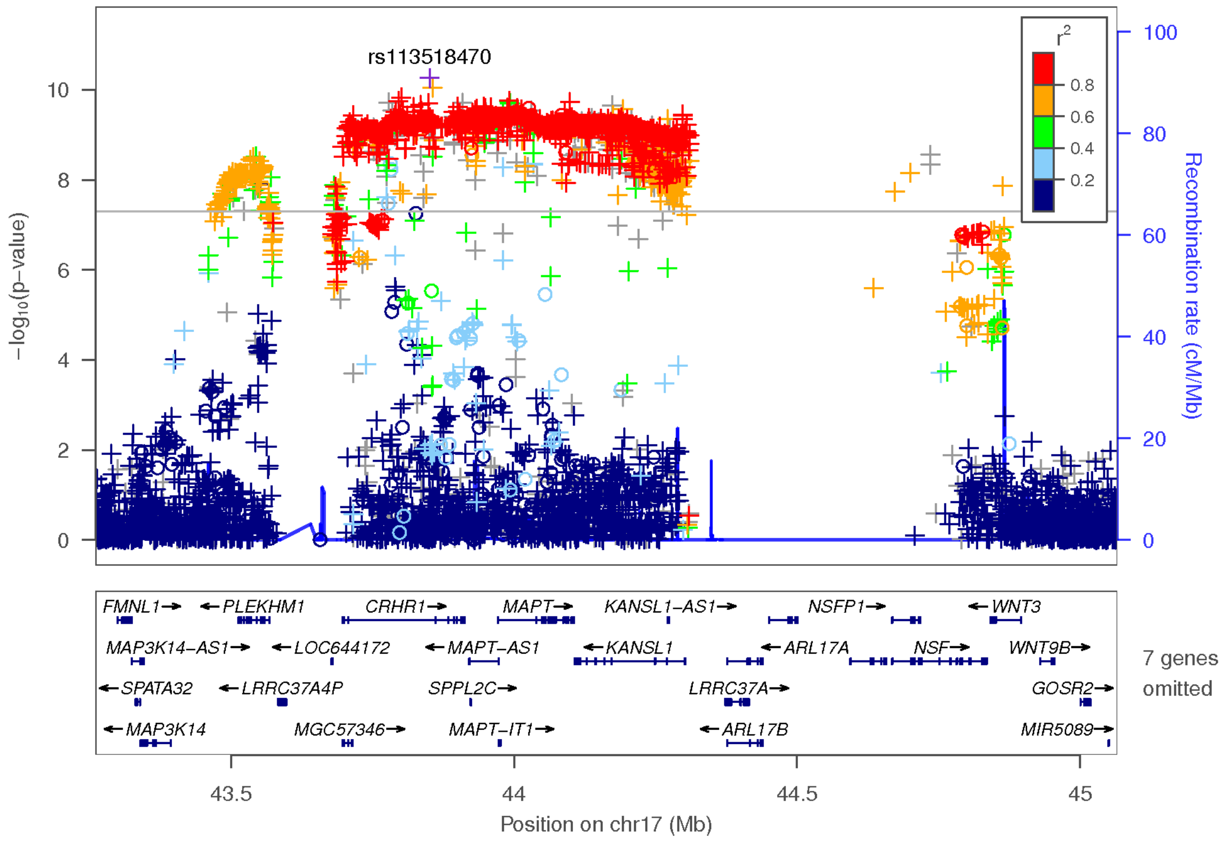
rs431722
r²
0.8
0.6
0.4
0.2
−log₁₀(p−value)
0
2
4
6
8
10
12
Recombination rate (cM/Mb)
0
20
40
60
80
100
GNL1
DDR1
C6orf15
HLA−B
LTA
CLIC1
C2
ATF6B
C6orf10
PRR3
MIR4640
CDSN
MICA
TNF
MSH5
CFB
FKBPL
HCG23
ABCF1
GTF2H4
CCHCR1
HCP5
BAG6
C6orf48
C4A
PPT2
BTNL2
MIR877
VARS2
TCF19
HCG26
APOM
NEU1
TNXB
HLA−DRA
85 genes omitted
31
31.5
32
32.5
Position on chr6 (Mb)

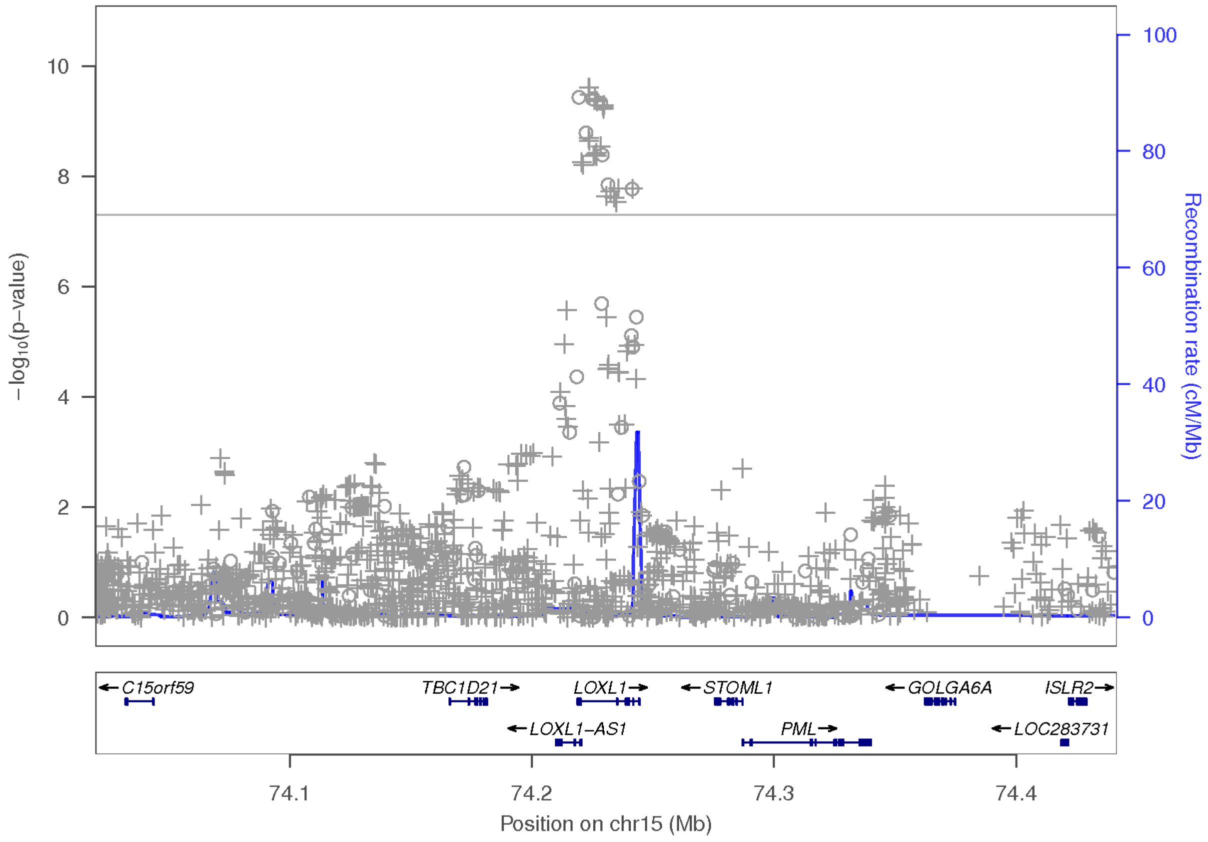


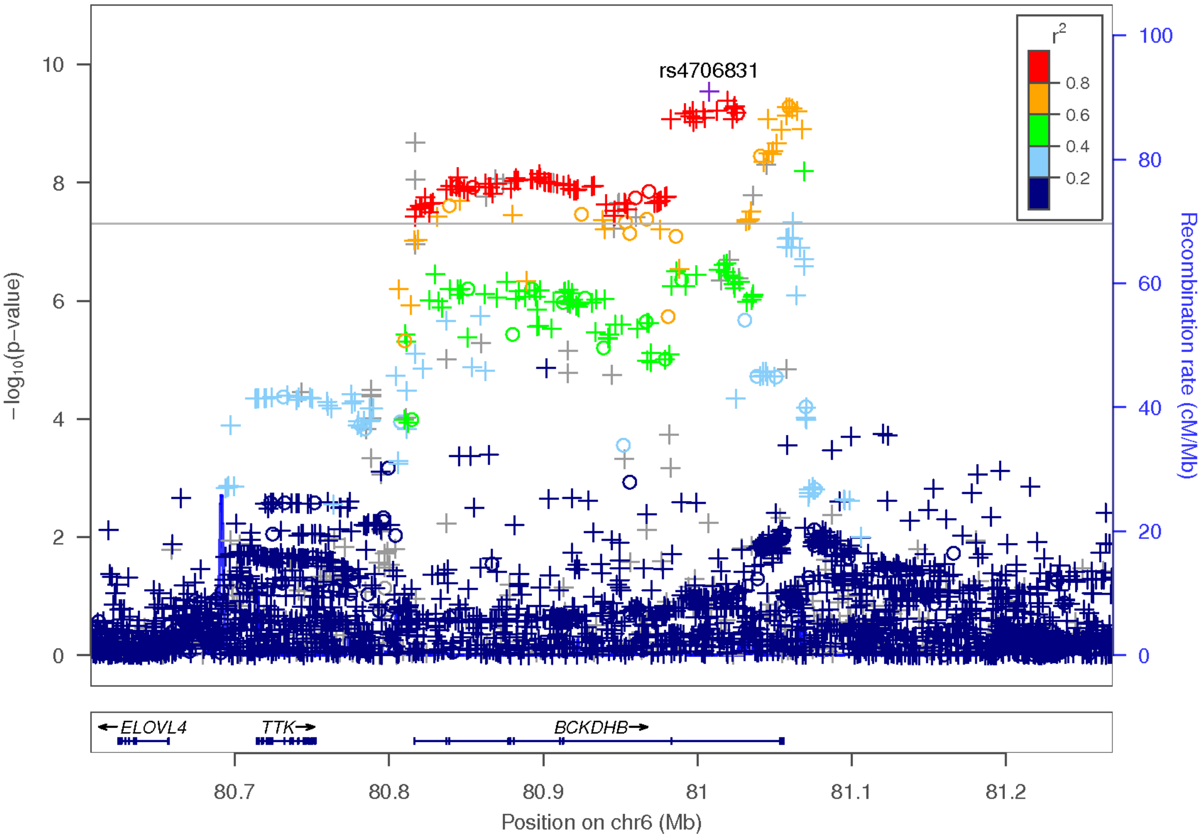
rs4417704
r²
0.8
0.6
0.4
0.2
−log₁₀(p−value)
0
2
4
6
8
10
12
Recombination rate (cM/Mb)
0
20
40
60
80
100
AQP12B
KIF1A
AGXT
LOC200772
SNED1
PASK
AQP12A
C2orf54
MTERFD2
PPP1R7
241.6
241.7
241.8
241.9
242
242.1
Position on chr2 (Mb)

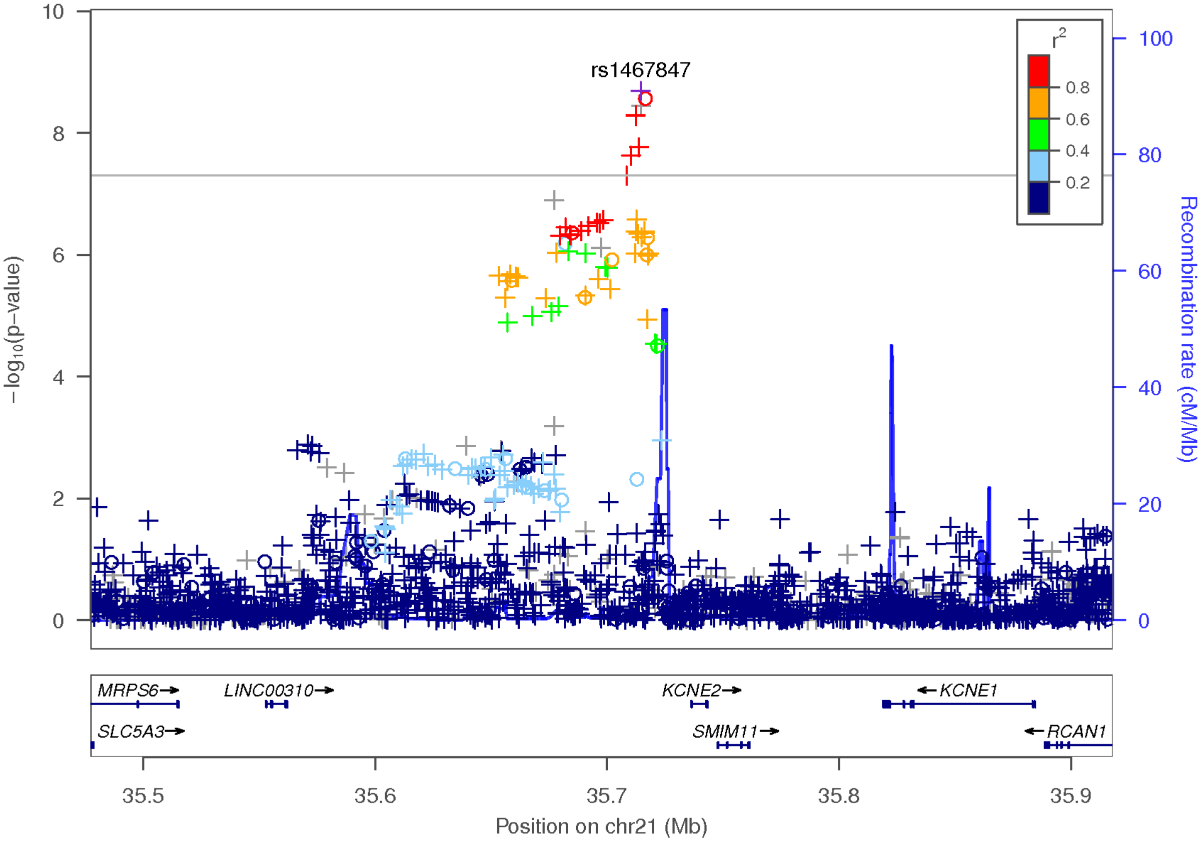

r^2
0.8
0.6
0.4
0.2
rs78579285
12
10
8
6
4
2
0
−log₁₀(p−value)
100
80
60
40
20
0
Recombination rate (cM/Mb)
ZFPM1→
ZC3H18→
←MVD
←RNF166
CDT1→
TRAPPC2L→
IL17C→
←SNAI3
←PIEZO1
←APRT
←PABPN1L
←CYBA
CTU2→
←GALNS
←CBFA2T3
SNAI3−AS1→
1 gene omitted
88.6
88.7
88.8
88.9
89
Position on chr16 (Mb)

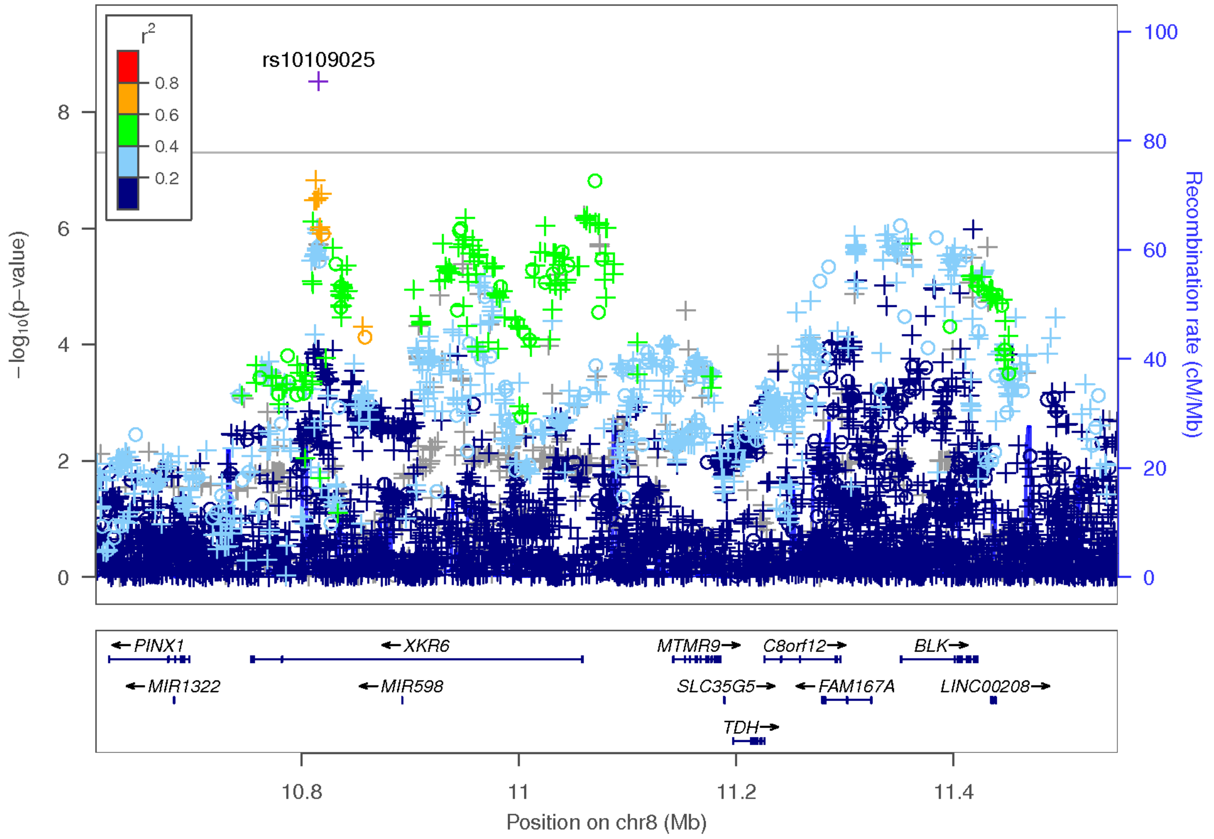

r^2
0.8
0.6
0.4
0.2
rs9828061
12
10
8
6
4
2
0
−log₁₀(p−value)
100
80
60
40
20
0
Recombination rate (cM/Mb)
GPR128→
TFG→
←ABI3BP
100.4
100.5
100.6
100.7
Position on chr3 (Mb)


rs113518470
r²
0.8
0.6
0.4
0.2
−log10(p−value)
Recombination rate (cM/Mb)
0
2
4
6
8
10
0
20
40
60
80
100
FMNL1→
←PLEKHM1
CRHR1→
MAPT→
KANSL1−AS1→
NSFP1→
←WNT3
MAP3K14−AS1→
←LOC644172
←MAPT−AS1
←KANSL1
←ARL17A
NSF→
WNT9B→
←SPATA32
←LRRC37A4P
SPPL2C→
LRRC37A→
GOSR2→
←MAP3K14
MGC57346→
MAPT−IT1→
←ARL17B
MIR5089→
7 genes omitted
43.5
44
44.5
45
Position on chr17 (Mb)

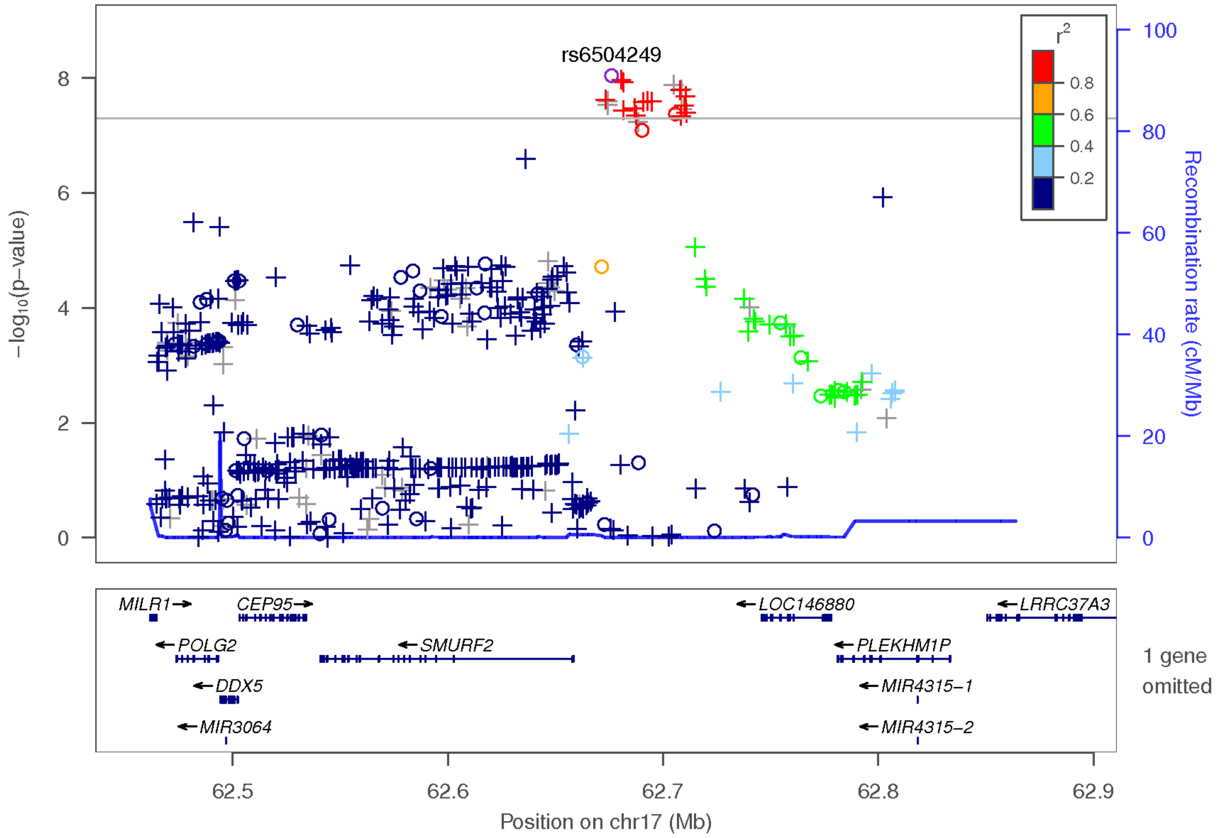

−log10(p−value)
Recombination rate (cM/Mb)
0
2
4
6
8
10
0
20
40
60
80
100
←C15orf59
TBC1D21→
LOXL1→
←STOML1
←GOLGA6A
ISLR2→
←LOXL1−AS1
PML→
←LOC283731
74.1
74.2
74.3
74.4
Position on chr15 (Mb)


rs4706831
r²
0.8
0.6
0.4
0.2
−log₁₀(p−value)
10
8
6
4
2
0
Recombination rate (cM/Mb)
100
80
60
40
20
0
ELOVL4
TTK
BCKDHB
80.7
80.8
80.9
81
81.1
81.2
Position on chr6 (Mb)

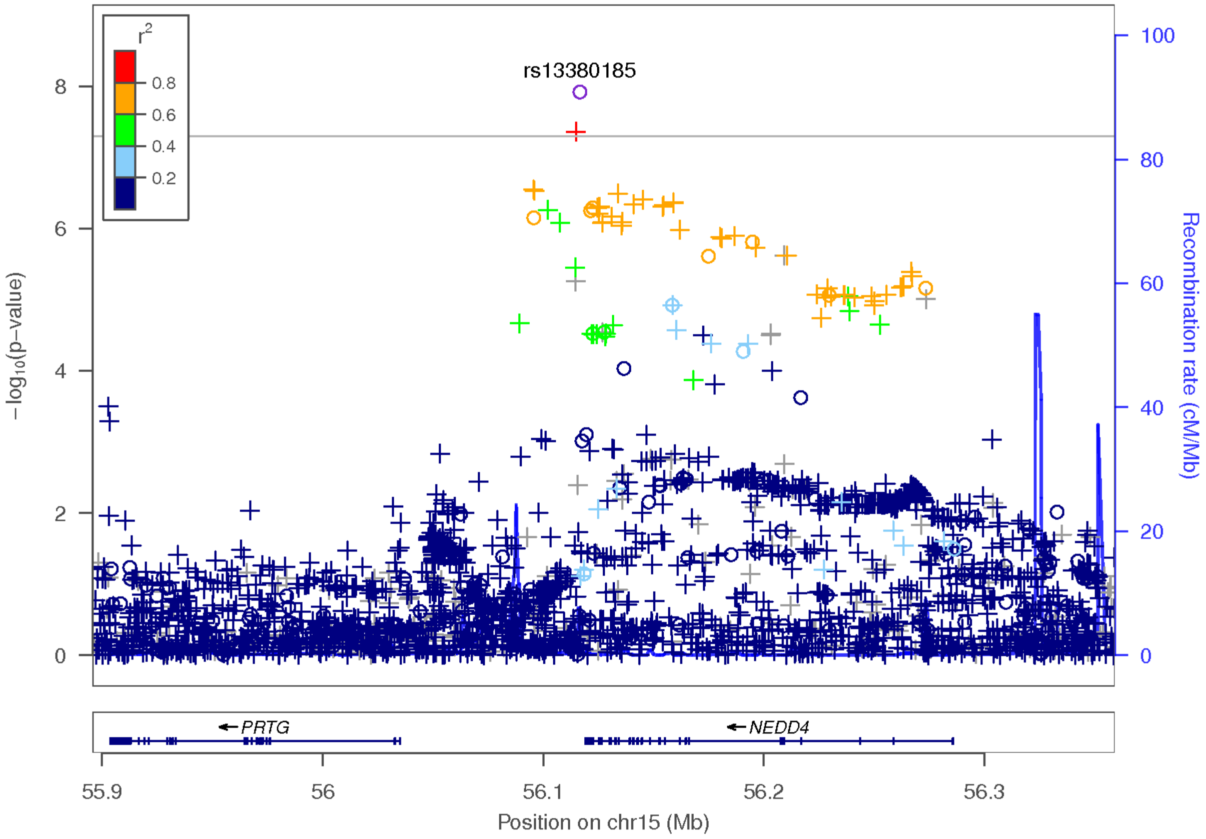

rs1467847
r²
0.8
0.6
0.4
0.2
−log₁₀(p−value)
10
8
6
4
2
0
Recombination rate (cM/Mb)
100
80
60
40
20
0
MRPS6
LINC00310
KCNE2
KCNE1
SLC5A3
SMIM11
RCAN1
35.5
35.6
35.7
35.8
35.9
Position on chr21 (Mb)

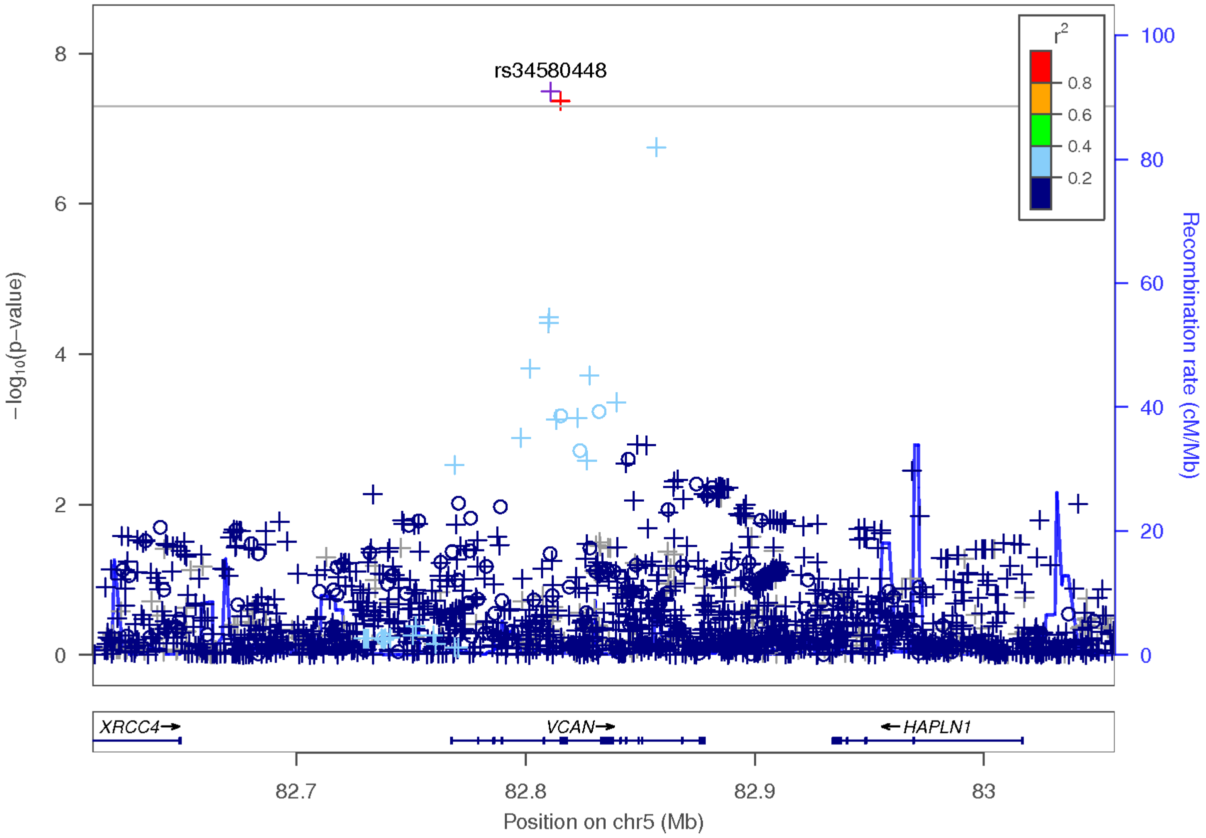


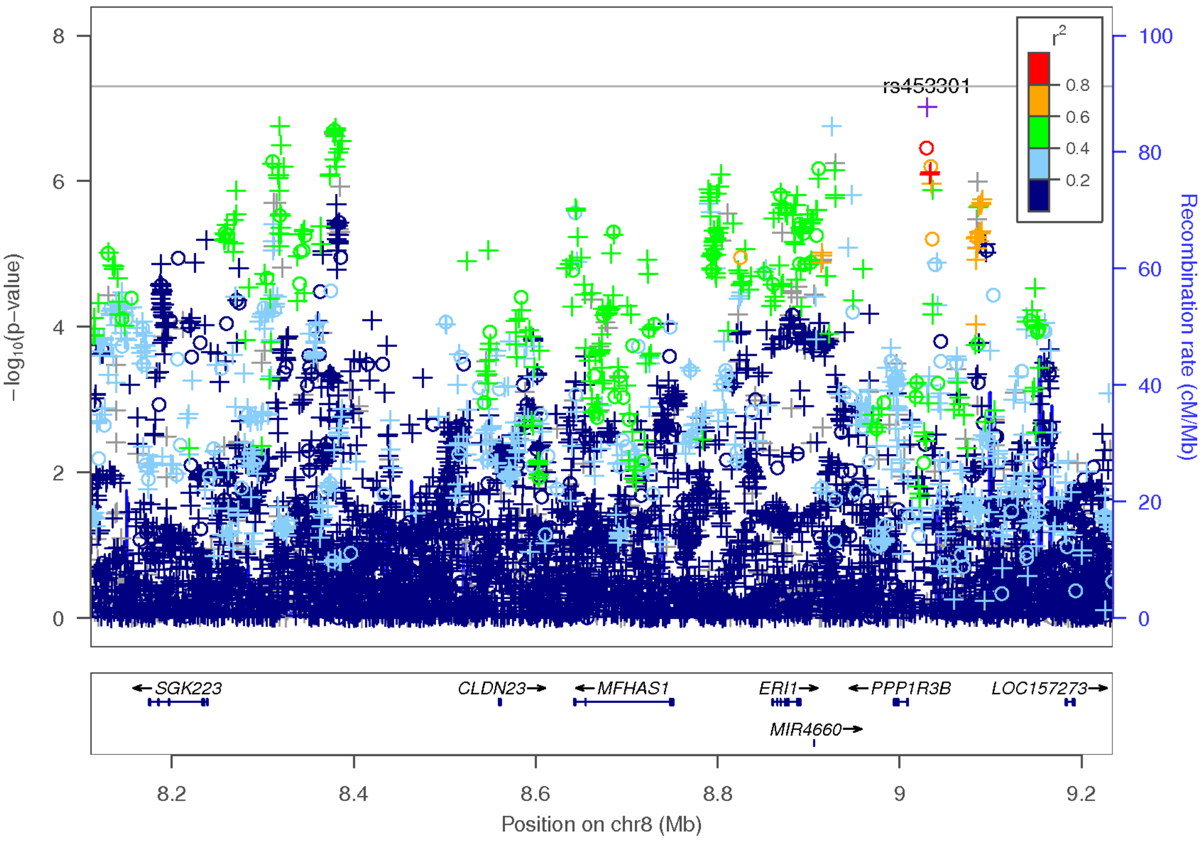
r²
0.8
0.6
0.4
0.2
rs10109025
−log₁₀(p−value)
0
2
4
6
8
Recombination rate (cM/Mb)
0
20
40
60
80
100
PINX1
XKR6
MTMR9
C8orf12
BLK
MIR1322
MIR598
SLC35G5
FAM167A
LINC00208
TDH
10.8
11
11.2
11.4
Position on chr8 (Mb)

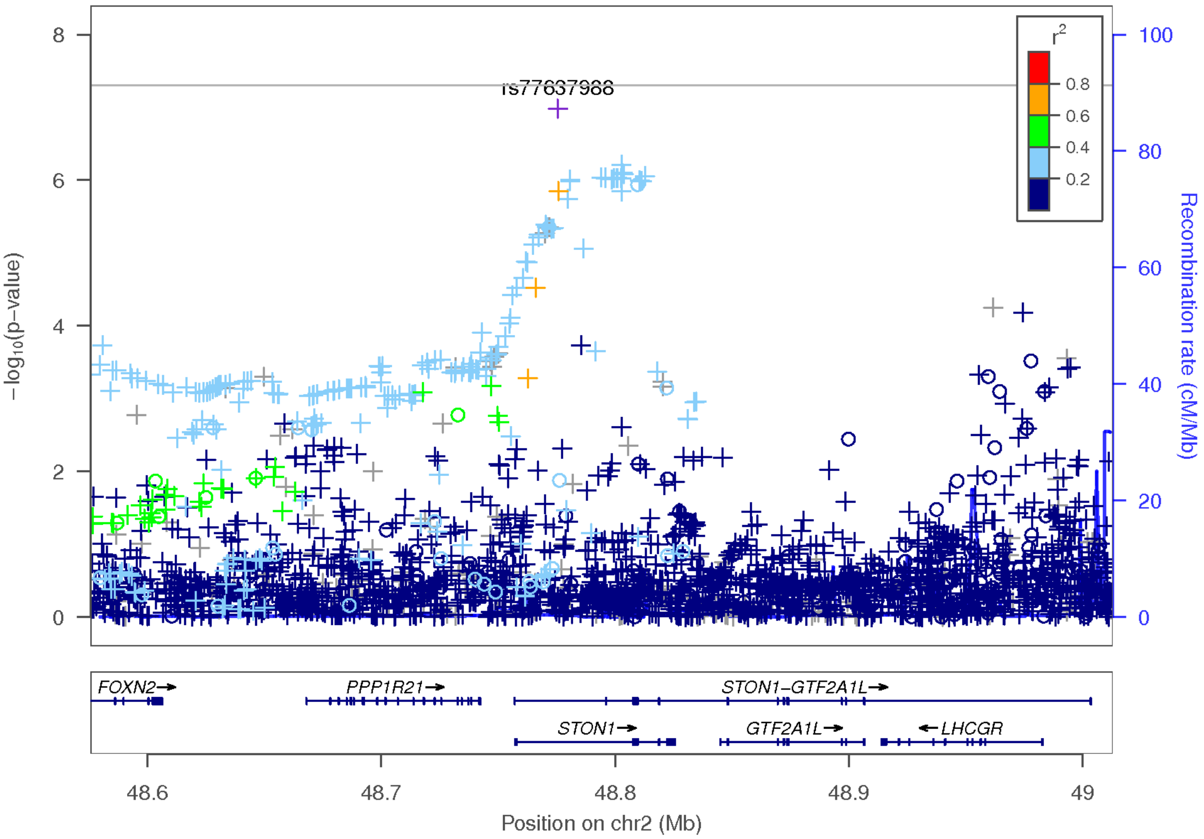


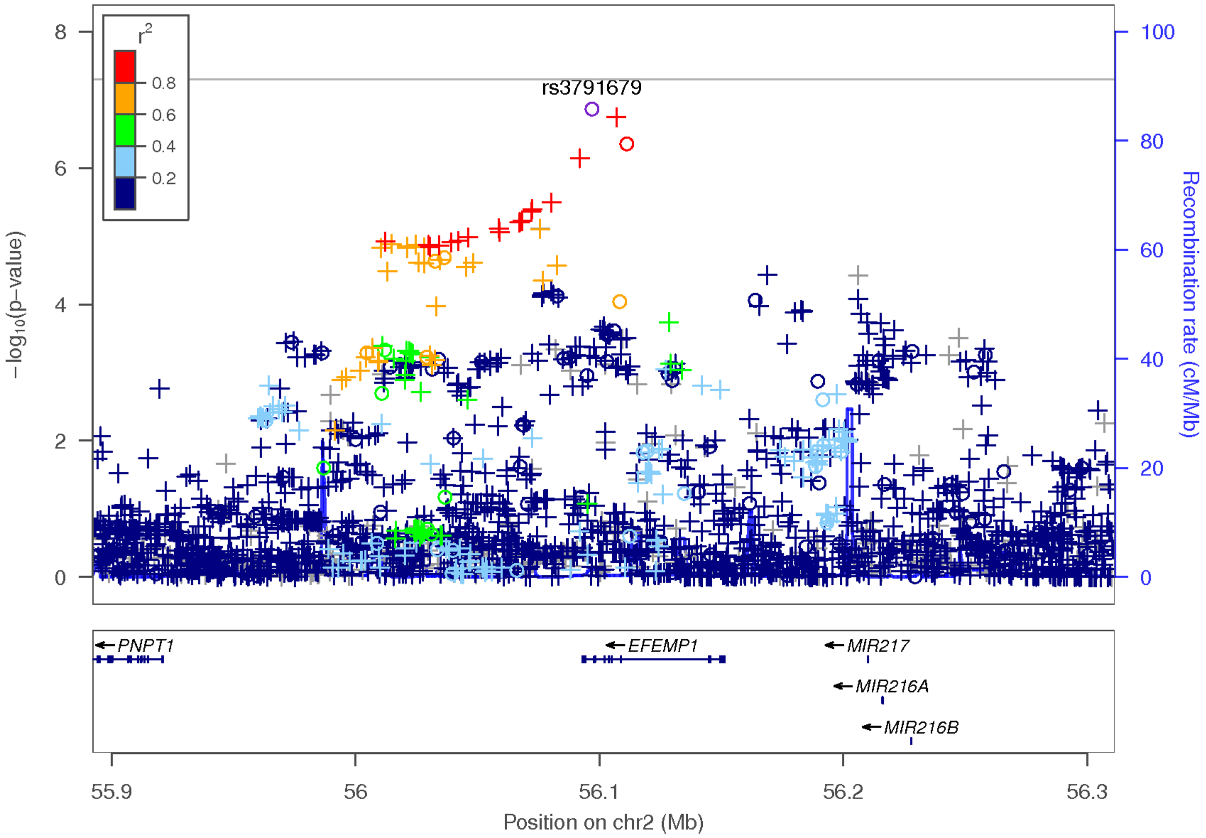
r²
0.8
0.6
0.4
0.2
rs6504249
−log₁₀(p−value)
0
2
4
6
8
Recombination rate (cM/Mb)
0
20
40
60
80
100
MILR1
CEP95
LOC146880
LRRC37A3
POLG2
SMURF2
PLEKHM1P
DDX5
MIR4315−1
MIR3064
MIR4315−2
1 gene
omitted
62.5
62.6
62.7
62.8
62.9
Position on chr17 (Mb)

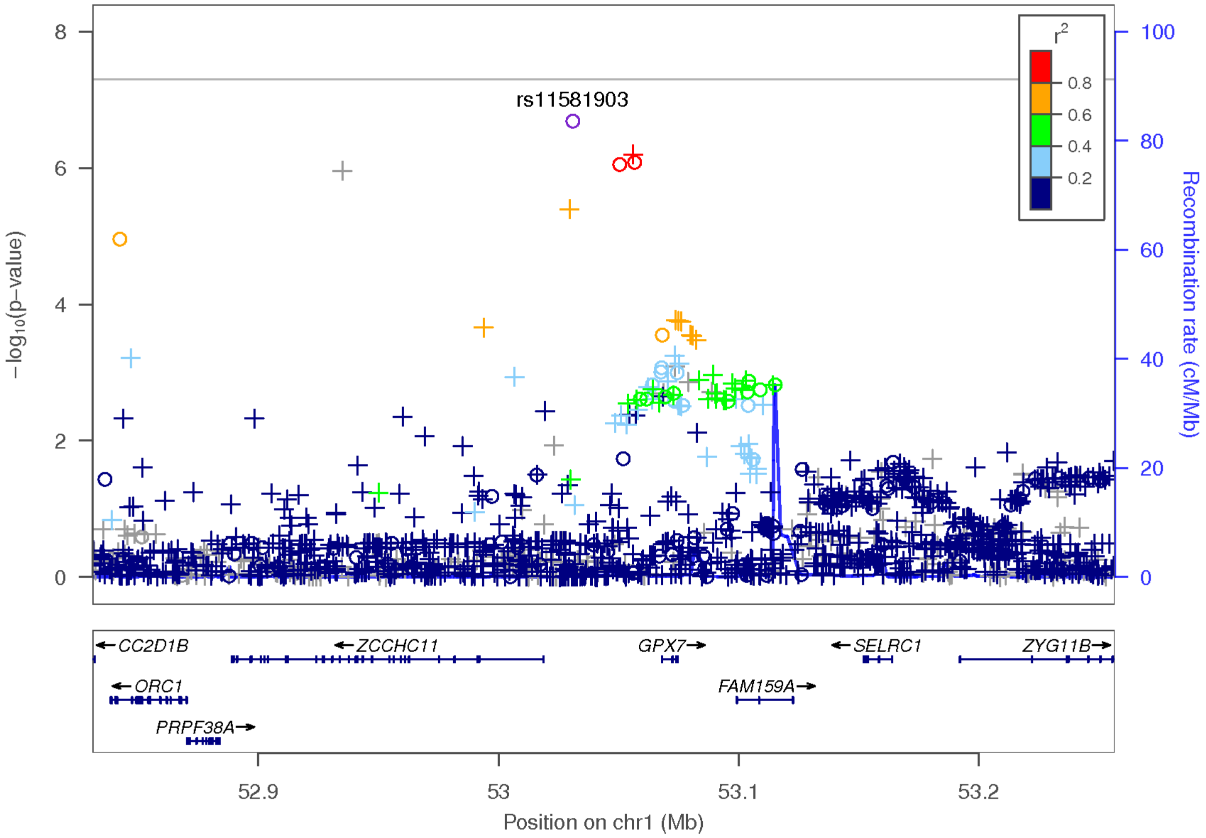

r^2
0.8
0.6
0.4
0.2
rs13380185
−log10(p−value)
0
2
4
6
8
Recombination rate (cM/Mb)
0
20
40
60
80
100
PRTG
NEDD4
55.9
56
56.1
56.2
56.3
Position on chr15 (Mb)

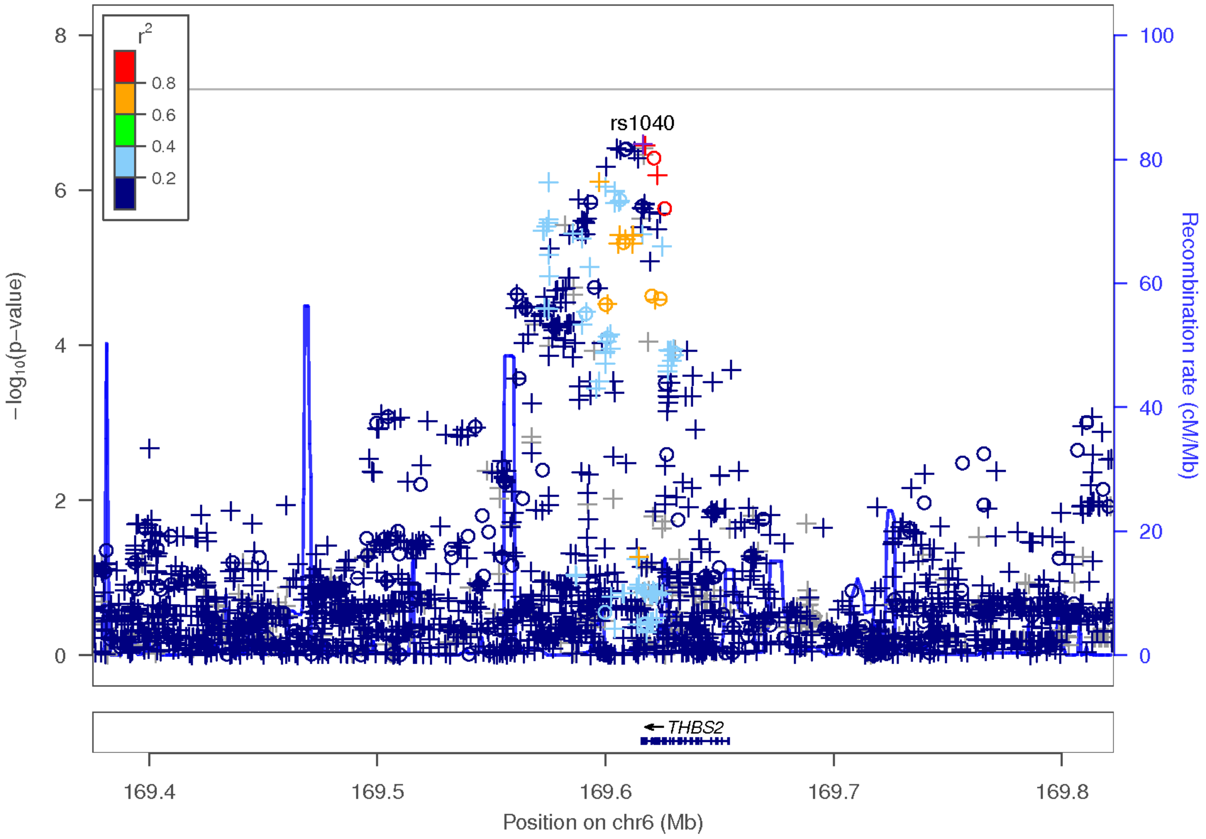

r^2
0.8
0.6
0.4
0.2
rs34580448
−log10(p−value)
0
2
4
6
8
Recombination rate (cM/Mb)
0
20
40
60
80
100
XRCC4
VCAN
HAPLN1
82.7
82.8
82.9
83
Position on chr5 (Mb)

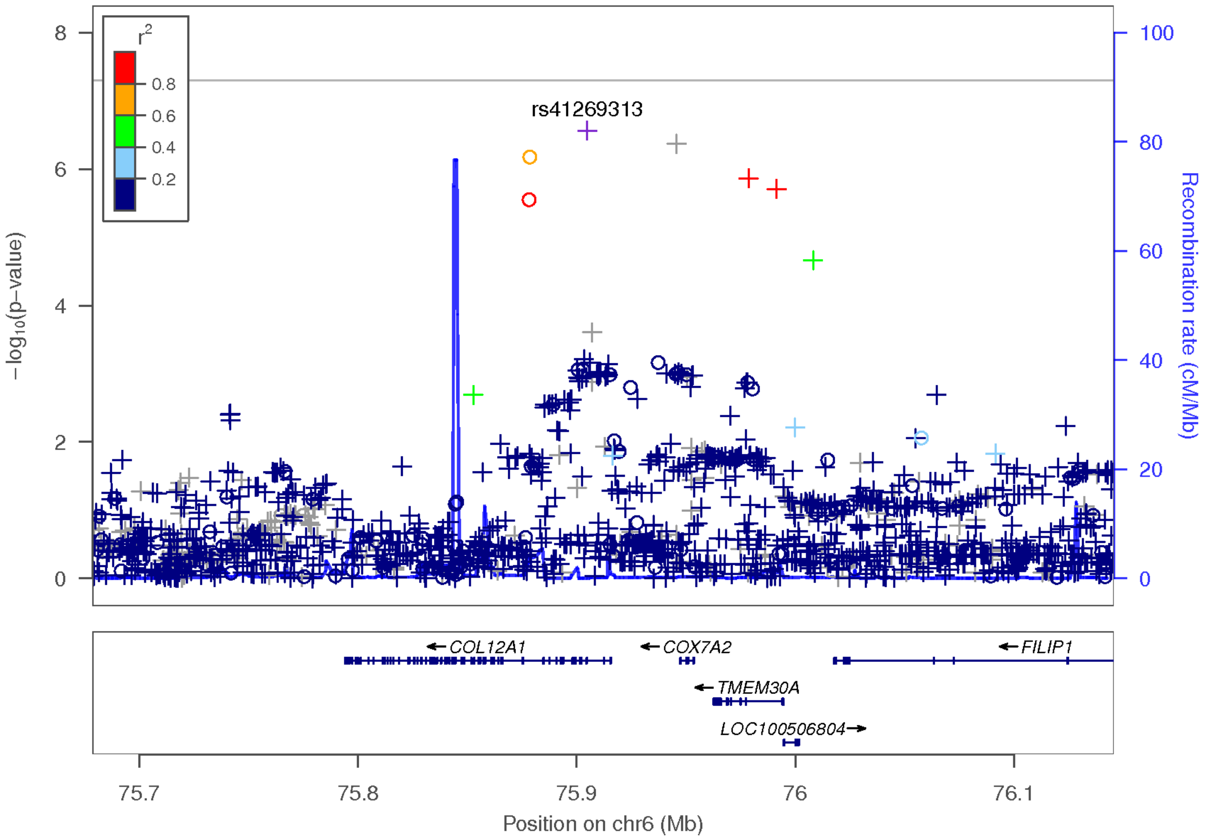

rs453301
r²
0.8
0.6
0.4
0.2
8
6
4
2
0
−log₁₀(p−value)
100
80
60
40
20
0
Recombination rate (cM/Mb)
←SGK223
CLDN23→
←MFHAS1
ERI1→
←PPP1R3B
LOC157273→
MIR4660→
8.2
8.4
8.6
8.8
9
9.2
Position on chr8 (Mb)

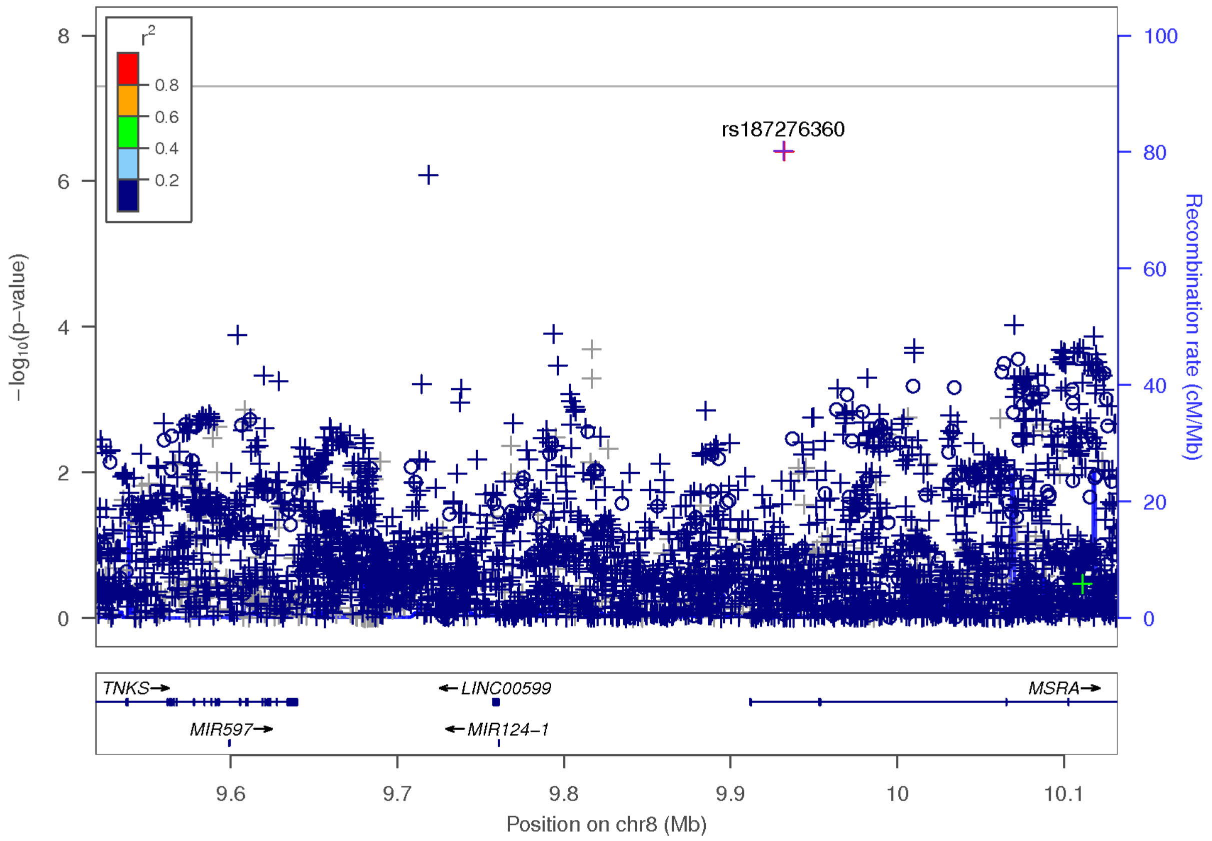

rs77637988
r²
0.8
0.6
0.4
0.2
8
6
4
2
0
−log₁₀(p−value)
100
80
60
40
20
0
Recombination rate (cM/Mb)
FOXN2→
PPP1R21→
STON1−GTF2A1L→
STON1→
GTF2A1L→
←LHCGR
48.6
48.7
48.8
48.9
49
Position on chr2 (Mb)

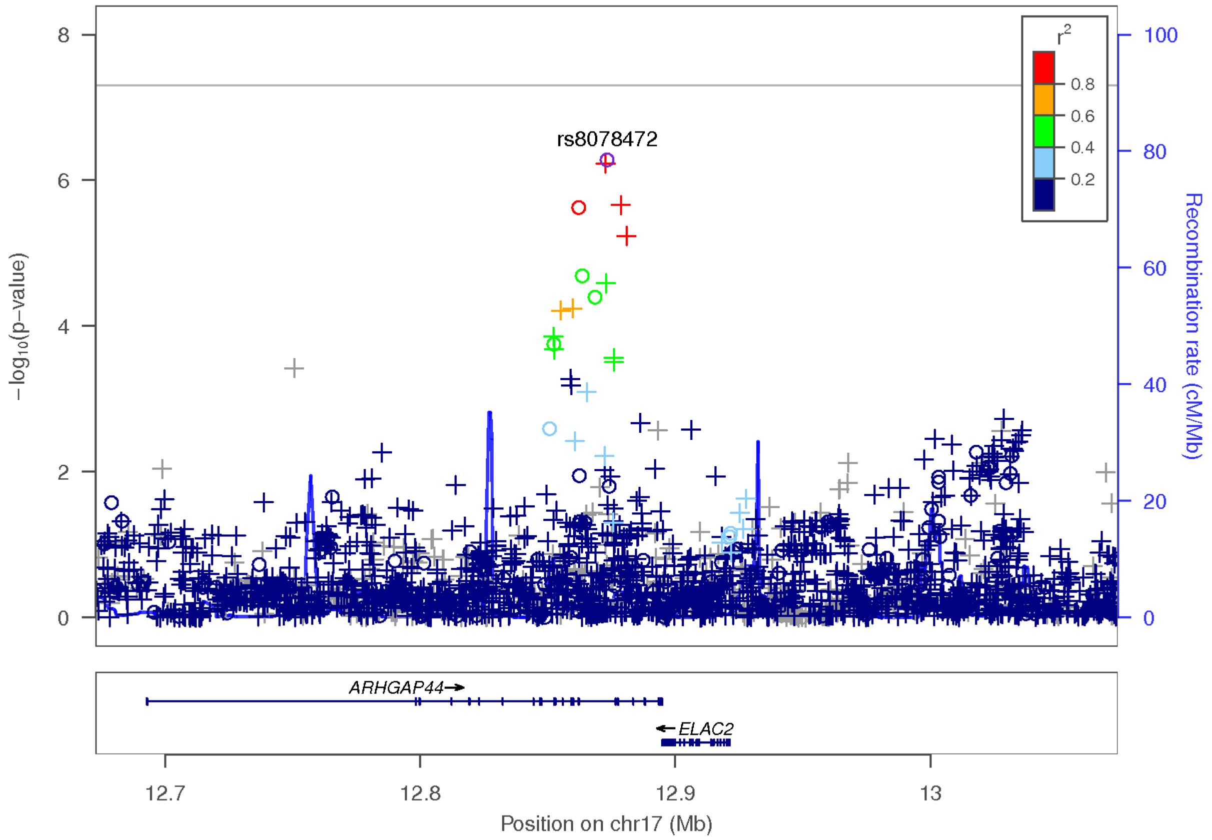

r²
0.8
0.6
0.4
0.2
rs3791679
−log₁₀(p−value)
0
2
4
6
8
Recombination rate (cM/Mb)
0
20
40
60
80
100
PNPT1
EFEMP1
MIR217
MIR216A
MIR216B
55.9
56
56.1
56.2
56.3
Position on chr2 (Mb)

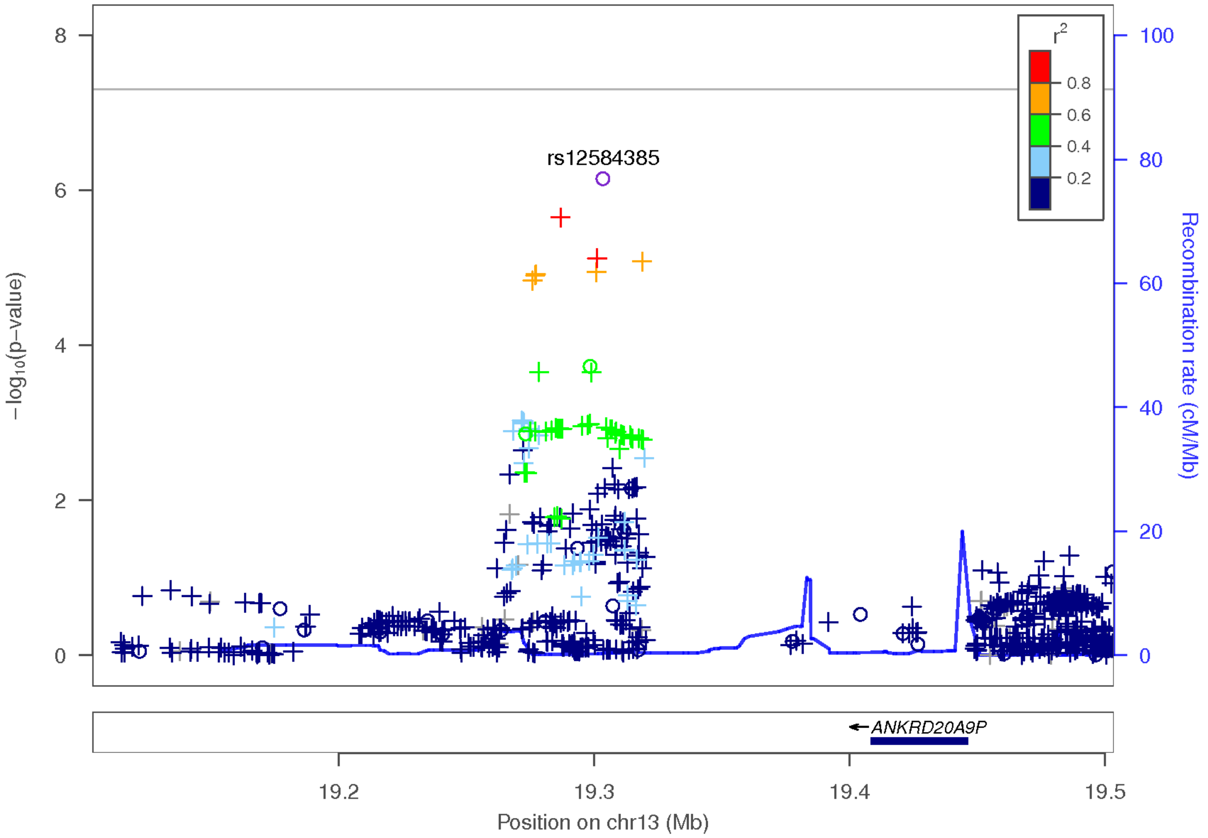

r²
0.8
0.6
0.4
0.2
rs11581903
−log₁₀(p−value)
0
2
4
6
8
Recombination rate (cM/Mb)
0
20
40
60
80
100
CC2D1B
ZCCHC11
GPX7
SELRC1
ZYG11B
ORC1
FAM159A
PRPF38A
52.9
53
53.1
53.2
Position on chr1 (Mb)

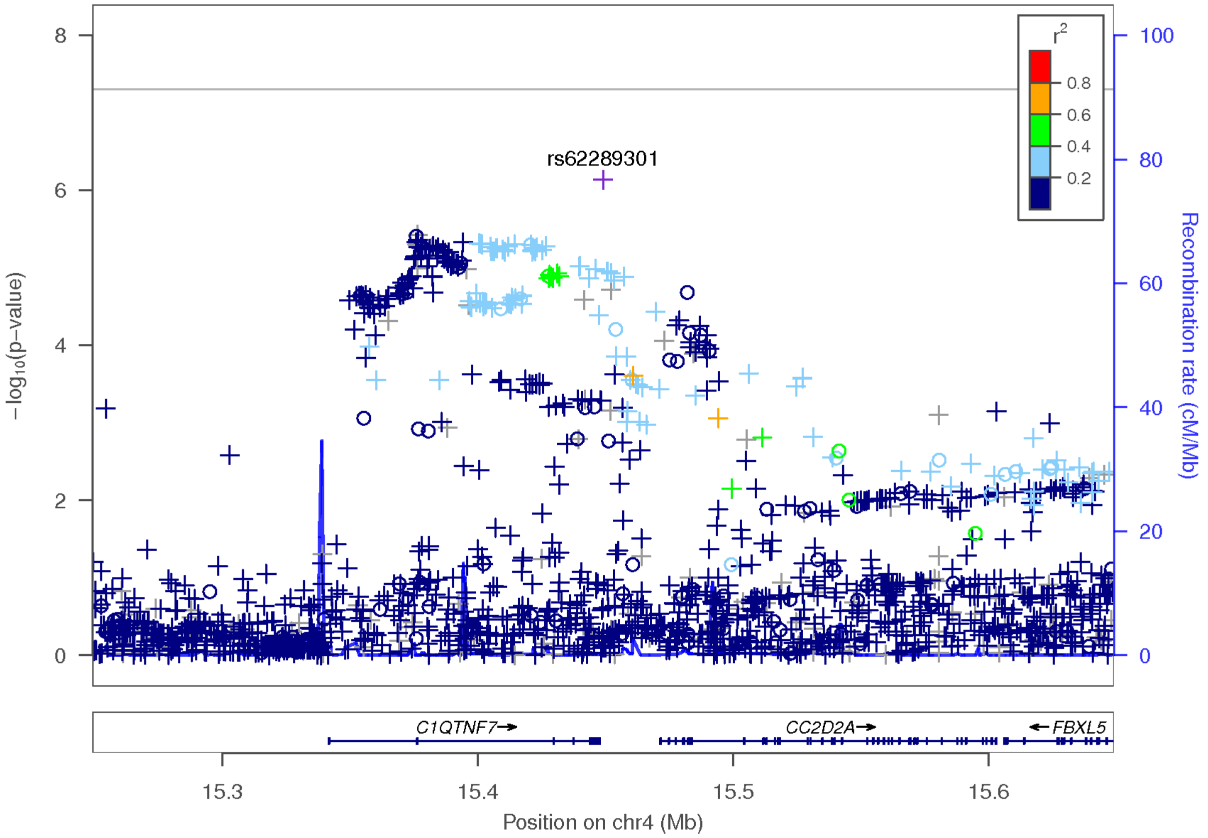

r^2
0.8
0.6
0.4
0.2
rs1040
8
6
4
2
0
$-\log_{10}$(p−value)
100
80
60
40
20
0
Recombination rate (cM/Mb)
THBS2
169.4
169.5
169.6
169.7
169.8
Position on chr6 (Mb)

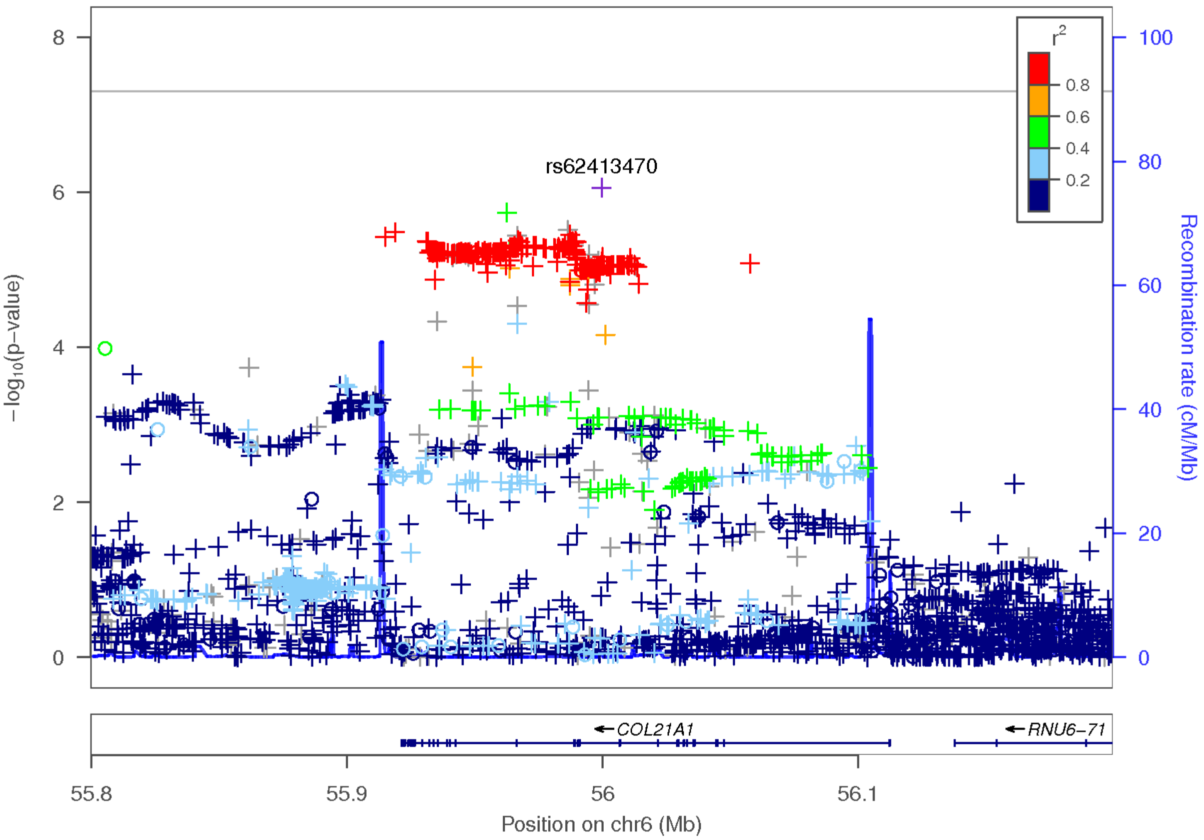

r^2
0.8
0.6
0.4
0.2
rs41269313
8
6
4
2
0
$-\log_{10}$(p−value)
100
80
60
40
20
0
Recombination rate (cM/Mb)
COL12A1
COX7A2
FILIP1
TMEM30A
LOC100506804
75.7
75.8
75.9
76
76.1
Position on chr6 (Mb)


r2
0.8
0.6
0.4
0.2
rs187276360
−log10(p−value)
8
6
4
2
0
Recombination rate (cM/Mb)
100
80
60
40
20
0
TNKS
MIR597
LINC00599
MIR124−1
MSRA
9.6
9.7
9.8
9.9
10
10.1
Position on chr8 (Mb)


r2
0.8
0.6
0.4
0.2
rs8078472
−log10(p−value)
8
6
4
2
0
Recombination rate (cM/Mb)
100
80
60
40
20
0
ARHGAP44
ELAC2
12.7
12.8
12.9
13
Position on chr17 (Mb)


rs12584385
r²
0.8
0.6
0.4
0.2
−log₁₀(p−value)
0
2
4
6
8
Recombination rate (cM/Mb)
0
20
40
60
80
100
ANKRD20A9P
19.2
19.3
19.4
19.5
Position on chr13 (Mb)


rs62289301
r²
0.8
0.6
0.4
0.2
−log₁₀(p−value)
0
2
4
6
8
Recombination rate (cM/Mb)
0
20
40
60
80
100
C1QTNF7
CC2D2A
FBXL5
15.3
15.4
15.5
15.6
Position on chr4 (Mb)


rs62413470
r²
0.8
0.6
0.4
0.2
8
6
4
2
0
−log₁₀(p−value)
100
80
60
40
20
0
Recombination rate (cM/Mb)
COL21A1
RNU6−71
55.8
55.9
56
56.1
Position on chr6 (Mb)