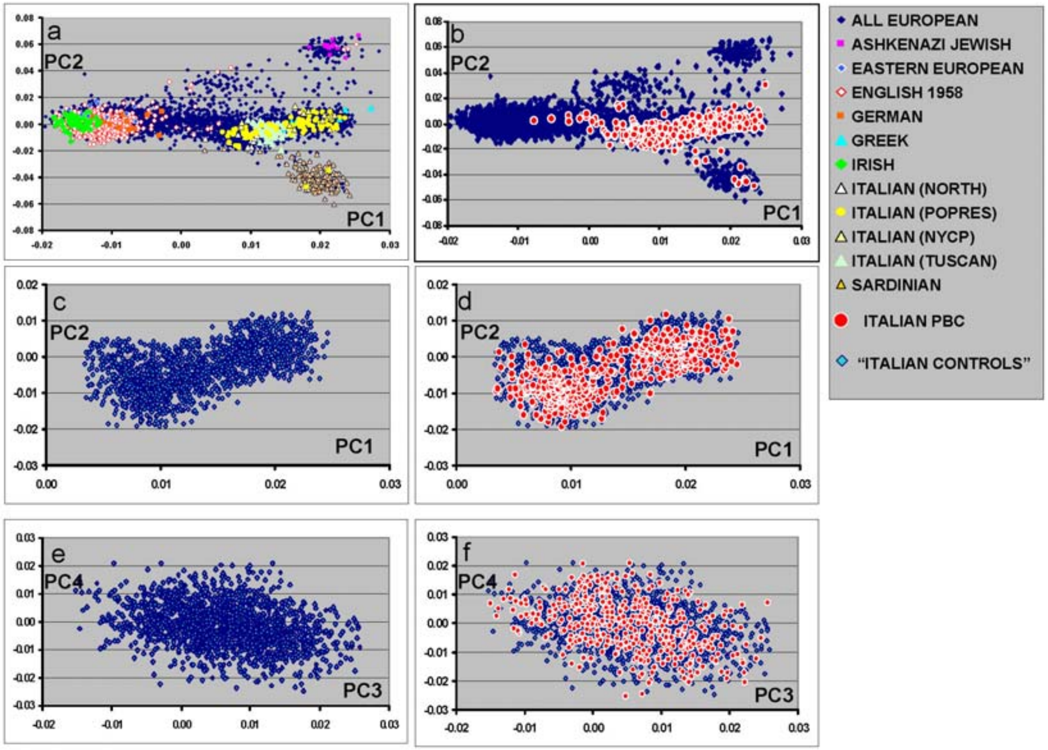
Genome-wide meta-analyses identifies three loci associated with primary biliary cirrhosis

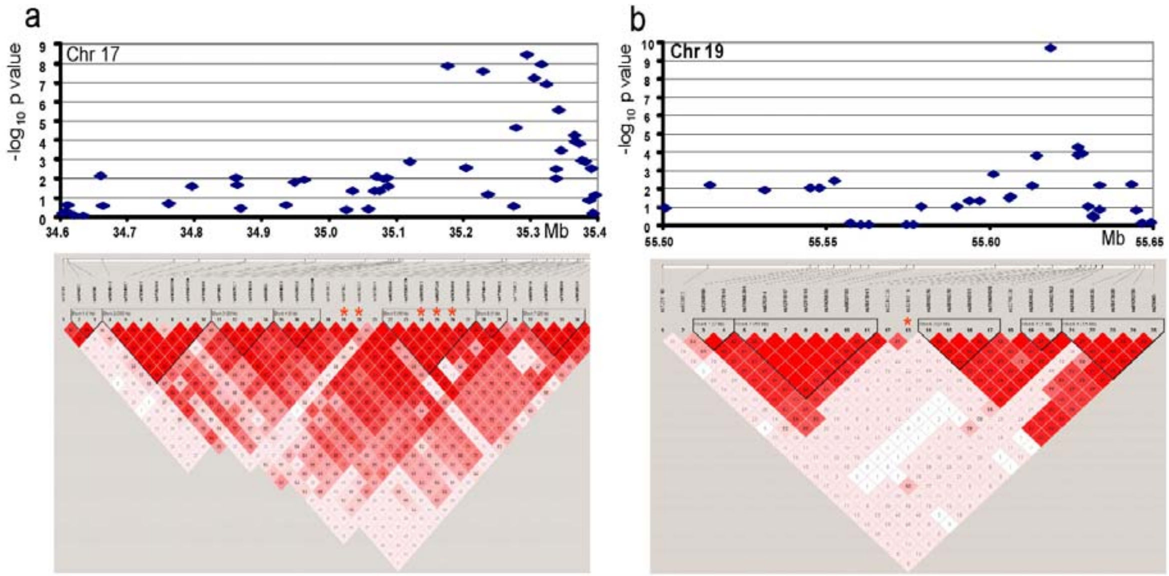
Xiangdong Liu, Pietro Invernizzi, Yue Lu, Roman Kosoy, Yan Lu, Ilaria Bianchi, Mauro Podda, Chun Xu, Gang Xie, Fabio Macciardi, Carlo Selmi, Sara Lupoli, Russell Shigeta, Michael Ransom, Ana Lleo, Annette T. Lee, Andrew L. Mason, Robert P. Myers, Kevork M. Peltekian, Cameron N. Ghent, Francesca Bernuzzi, Massimo Zuin, Floriano Rosina, Elisabetta Borghesio, Annarosa Floreani, Roberta Lazzari, Grazia Niro, Angelo Andriulli, Luigi Muratori, Paolo Muratori, Piero L. Almasio, Pietro Andreone, Marzia Margotti, Maurizia Brunetto, Barbara Coco, Domenico Alvaro, Maria C. Bragazzi, Fabio Marra, Alessandro Pisano, Cristina Rigamonti, Massimo Colombo, Marco Marzioni, Antonio Benedetti, Luca Fabris, Mario Strazzabosco, Piero Portincasa, Vincenzo O. Palmieri, Claudio Tiribelli, Lory Croce, Savino Bruno, Sonia Rossi, Maria Vinci, Cleofe Prisco, Alberto Mattalia, Pierluigi Toniutto, Antonio Picciotto, Andrea Galli, Carlo Ferrari, Silvia Colombo, Giovanni Casella, Lorenzo Morini, Nicola Caporaso, Agostino Colli, Giancarlo Spinzi, Renzo Montanari, Peter K. Gregersen, E. Jenny Heathcote, Gideon M. Hirschfield, Katherine A. Siminovitch, Christopher I. Amos, M. Eric Gershwin, and Michael F. Seldin.

Supplementary Information

1. **Supplementary Figure 1**. Principal components analysis (PCA) showing selection of homogeneous Italian cases and controls.
2. **Supplementary Figure 2**. Haplotype analysis of PBC susceptibility regions on chromosomes 17 and 19.
3. **Supplementary Table 1**. Summary of Subject Genotyping Information
4. **Supplementary Table 2**. Summary of Association Tests for Italian PBC and combined Italian PBC and Canadian PBC study
5. **Supplementary Table 3**. Analysis of *IL12A/SCHIP1* SNPs
6. **Supplementary Table 4**. Comparison of Meta-Analyses containing all PBC Cases and AMA only Cases
7. **Supplementary Table 5**. Comparison of PBC and Celiac Association for SCHIP1/IL12A Region
8. **Supplementary Methods**.



Supplementary Figure 1. (**a** - **f**) Principal components analysis (PCA) showing selection of homogeneous Italian cases and controls. PCA was performed using a large set of diverse European individuals (see Methods) and 34,000 SNPs distributed throughout each autosome excluding those with strong linkage disequilibrium. (**a**) Shows a plot of principal component (PC) 2 against PC1 with selected color coded European population groups. (**b**) Highlights the PBC cases collected in Italy (492 cases) in PC2/PC1 plot. (**c** and **d**) Shows the PC2/PC1 plot of homogeneous Italian controls and cases selected as described in Methods. (**e** and **f**) Shows the PC4/PC3 plot of homogeneous Italian controls and cases selected as described in Methods.



C Comparison of PBC and Asthma Association with SNPs on Chromosome 17

rs	bp	Allele	PBC -Log10 p value	PBC OR	ASTHMA -Log10 p value	ASTHMA OR
rs7216389	35323475	T	7.15	0.75	10	1.45
rs4795405	35341943	T	5.54	1.28	8.7	0.66
rs8079416	35346239	G	3.81	0.82	8.2	1.3
rs3894194	35375519	A	3.45	0.83	7.7	1.22
rs3859192	35382174	A	2.73	0.85	7.3	1.11

Supplementary Figure 2. Haplotype analysis of PBC susceptibility regions on chromosomes 17 and 19. In the top half of panels **a** and **b,** the association results using SNPTEST analysis is shown. The results include imputed SNPs (see **Supplementary Methods** and **Supplementary Table 6**. SNPTEST results were adjusted for population substructure using the PCA results from the first three principle components. The SNPTEST algorithm also adjusts for genotype uncertainty (**Supplementary Methods**). SNPs with –log10 p values > 7 are indicated with a red star above the haplotype heat plot and include rs907092, rs9303277, rs2872507, rs8067378 and rs2305480 for chromosome 17 and rs3745516 for chromosome 19. In panel **c**, the results for PBC are compared with those for pediatric asthma [derived from Moffatt, M.F. et al. Nature 448, 470-3 (2007)]. The odds ratios are presented for the minor allele on the forward strand and show an opposite direction for the odds ratio for these two diseases.

Supplementary Table 1. Summary of Subject Genotyping Information

			Excluded From Analyses				Analyzed Homogeneous
	Platform	Genotyped	% complete[a]	relationship[b]	nonEuropean[c]	on-Italian European	Italian[d]
Italian PBC Cases	Illumina 610K	524	15	13	6	37	453
Italian Controls	Illumina 1Mb	1027	29	16	11	971	637
British Blood Bank Controls	Illumina 550K	1357	19	3	18	1317	6
New York Cancer Project	Illumina 550K	1425	10	10	115	1280	159
Childrens Hospital of Philadelphia	Illumina 550K	1711	23	47	169	1472	129
NIH Laboratory of Neurogenetics	Illumina 550K	271	271	0	16	255	14
Total Controls Included in Analyses							945
							Canadian/European
Original Canadian PBC Cases[e]	Illumina 370K	536	6	30	19	NA	481
M.D. Anderson historical controls[e]	Illumina 300K	1137	0	4	15	NA	1118
Canadian subjects[e]	Illumina 370K	399	1	4	16	NA	378
Cancer Genetics Markers of Susceptibility	Illumina 550K	2236	0	12	14	NA	2210
Total Controls Included in Analyses							3706

a. Less than 95% complete genotypes and sex discrepency

b. PI^ >0.1 assessed using PLINK IBD

c. >10% nonEuropean admixture (see Supplemental Methods)

d. Based on PCA analyses (see Supplemental Methods and Supplemental Figure 1).

e. Genotypes used in original Canadian study.

Supplementary Table 2. Summary of Association Tests for Italian PBC and combined Italian PBC and Canadian PBC study[a]

SNP	Chr.	Position	Allele 1	Allele 2	Gene(s)	Italian Control Allele Frequency	Italian Case Allele Frequency	Italian p value	Italian Odds Ratio	Italian OR 95% Upper CI	Italian OR 95% Lower CI	Canadian Control Allele Frequency	Canadian Case Allele Frequency	Canadian p value	Canadian Odds Ratio	Canadian OR 95% Upper CI	Canadian OR 95% Lower CI	Meta-Analysis p value	Meta-Analysis Odds Ratio	Cochran's Q Statistic p value
rs3748816	1	2516606	G	A	*MMEL1*	0.305	0.305	9.14E-01	1.01	0.85	1.21	0.337	0.400	7.13E-05	1.32	1.15	1.51	1.90E-03	1.19	0.022
rs11588497	1	32803048	G	A	*ZBTB8A*	0.193	0.175	2.99E-01	0.89	0.72	1.11	0.190	0.140	4.68E-05	0.67	0.55	0.81	4.29E-04	0.77	0.069
rs3790565	1	67583944	C	T	*IL12RB2*	0.227	0.272	1.74E-02	1.26	1.04	1.51	0.174	0.256	6.44E-09	1.60	1.37	1.88	4.46E-09	1.44	0.065
rs3790567	1	67594965	A	G	*IL12RB2*	0.311	0.376	1.46E-03	1.32	1.11	1.57	0.232	0.331	4.40E-10	1.60	1.38	1.86	7.53E-12	1.48	0.093
rs4297265	1	67624923	G	A	*IL12RB2*	0.486	0.521	9.57E-02	1.15	0.98	1.35	0.410	0.479	1.37E-04	1.31	1.14	1.50	5.42E-05	1.24	0.232
rs2270614	1	67628609	A	G	*IL12RB2*	0.486	0.521	9.38E-02	1.15	0.98	1.35	0.410	0.481	8.10E-05	1.32	1.15	1.51	3.44E-05	1.25	0.206
rs1418426	1	111299634	C	T	*C1orf103*	0.405	0.389	2.92E-01	0.91	0.77	1.08	0.384	0.317	1.09E-05	0.72	0.62	0.83	1.01E-04	0.80	0.047
rs4636400	1	144155913	G	T	*NBPF3*	0.351	0.295	8.18E-03	0.79	0.66	0.94	0.397	0.347	4.23E-03	0.82	0.71	0.94	8.08E-05	0.80	0.837
rs1539414	1	196010129	A	G	*DENND1B*	0.251	0.300	4.16E-03	1.30	1.09	1.56	0.219	0.255	7.91E-03	1.23	1.06	1.44	1.82E-04	1.25	0.581
rs3024505	1	205006527	A	G	*MAPKAPK2*	0.141	0.150	4.74E-01	1.09	0.86	1.37	0.151	0.202	3.85E-05	1.43	1.21	1.70	1.54E-04	1.30	0.055
rs7527314	1	215886180	T	C	*SPATA17*	0.211	0.278	5.09E-05	1.48	1.22	1.78	0.225	0.245	1.45E-01	1.12	0.96	1.31	2.21E-04	1.26	0.029
rs3795366	1	230717373	A	G	*SIPA1L2*	0.042	0.077	5.21E-05	2.03	1.43	2.87	0.069	0.066	6.91E-01	0.95	0.72	1.24	3.46E-02	1.26	0.001
rs789633	1	230739777	C	T	*SIPA1L2*	0.040	0.072	9.81E-05	2.01	1.41	2.86	0.069	0.067	8.64E-01	0.98	0.75	1.28	2.98E-02	1.27	0.002
rs789645	1	230748744	G	A	*SIPA1L2*	0.038	0.073	4.85E-05	2.05	1.44	2.93	0.059	0.064	5.30E-01	1.09	0.83	1.43	2.86E-03	1.40	0.007
rs12049318	1	241660283	A	C	*SDCCAG8*	0.152	0.143	4.63E-01	0.92	0.73	1.16	0.170	0.117	5.94E-05	0.65	0.53	0.80	4.66E-04	0.76	0.031
rs7576701	2	3696102	C	T	*ALLC*	0.208	0.275	9.93E-05	1.45	1.20	1.75	0.219	0.234	2.98E-01	1.09	0.93	1.28	1.14E-03	1.23	0.021
rs1406230	2	29436825	T	C	*ALK*	0.327	0.409	8.67E-06	1.46	1.24	1.73	0.376	0.363	5.00E-01	0.95	0.83	1.10	1.55E-02	1.14	0.000
rs7569076	2	88554147	T	C	*none*	0.261	0.301	3.53E-02	1.22	1.01	1.46	0.214	0.274	2.11E-04	1.34	1.15	1.56	1.41E-05	1.30	0.355
rs2890845	2	146810855	C	A	*none*	0.493	0.491	8.96E-01	0.99	0.84	1.16	0.453	0.384	5.93E-05	0.75	0.66	0.86	1.66E-03	0.84	0.012
rs799770	2	154999825	G	A	*GALNT13*	0.093	0.152	1.08E-05	1.73	1.35	2.21	0.122	0.118	5.92E-01	0.94	0.76	1.17	1.12E-02	1.23	0.000
rs697651	2	155031524	C	A	*GALNT13*	0.360	0.279	2.93E-05	0.68	0.57	0.82	0.350	0.353	8.17E-01	1.02	0.88	1.17	1.21E-02	0.87	0.001
rs3769811	2	182745334	T	G	*PDE1A*	0.181	0.155	5.61E-02	0.81	0.65	1.01	0.188	0.138	3.52E-04	0.70	0.58	0.85	8.75E-05	0.75	0.365
rs7601754	2	191648696	G	A	*STAT4*	0.238	0.190	3.92E-03	0.74	0.61	0.91	0.174	0.134	7.91E-04	0.71	0.58	0.87	1.34E-05	0.73	0.841
rs6752770	2	191681808	G	A	*STAT4*	0.269	0.325	2.15E-03	1.32	1.11	1.58	0.294	0.334	6.97E-03	1.22	1.06	1.40	6.15E-05	1.26	0.483
rs10515940	2	201266944	A	C	*CASP10*	0.097	0.143	8.85E-05	1.63	1.27	2.08	0.146	0.136	5.79E-01	0.95	0.78	1.15	4.68E-02	1.17	0.001
rs1530239	2	213651879	T	C	*IKZF2*	0.110	0.058	3.14E-05	0.51	0.37	0.71	0.088	0.089	8.01E-01	1.03	0.81	1.31	2.38E-02	0.80	0.001
rs6437268	2	240649535	C	T	*none*	0.365	0.296	4.15E-04	0.73	0.61	0.87	0.339	0.299	1.06E-02	0.83	0.71	0.96	2.09E-05	0.78	0.298
rs2887401	2	240685626	A	G	*none*	0.265	0.211	3.02E-03	0.74	0.61	0.91	0.258	0.220	1.23E-02	0.81	0.69	0.96	9.63E-05	0.78	0.542
rs4685976	3	5716702	C	T	*none*	0.390	0.376	7.10E-01	0.97	0.82	1.15	0.442	0.490	2.17E-05	1.34	1.17	1.54	2.56E-03	1.18	0.004
rs1869241	3	5729732	T	C	*none*	0.269	0.259	7.21E-01	0.97	0.80	1.16	0.311	0.381	7.09E-06	1.38	1.20	1.58	1.01E-03	1.21	0.003
rs9311642	3	5748145	C	T	*none*	0.372	0.382	4.63E-01	1.07	0.90	1.26	0.396	0.459	8.79E-05	1.31	1.15	1.50	5.04E-04	1.21	0.062
rs4686174	3	7894746	T	C	*none*	0.254	0.303	6.33E-03	1.29	1.07	1.54	0.267	0.307	6.47E-03	1.23	1.06	1.42	9.96E-05	1.25	0.724
rs6797331	3	7915671	T	C	*none*	0.258	0.307	4.97E-03	1.30	1.08	1.55	0.270	0.313	3.71E-03	1.24	1.07	1.44	4.62E-05	1.27	0.761
rs1348109	3	16924518	G	A	*PLCL2*	0.419	0.436	3.88E-01	1.08	0.91	1.27	0.436	0.494	4.18E-05	1.33	1.16	1.52	2.08E-04	1.22	0.052
rs1372072	3	16930263	A	G	*PLCL2*	0.336	0.357	2.92E-01	1.10	0.92	1.30	0.350	0.428	2.70E-06	1.39	1.21	1.60	1.52E-05	1.27	0.033
rs9876137	3	16936269	G	A	*PLCL2*	0.337	0.358	2.90E-01	1.10	0.92	1.30	0.350	0.428	2.64E-06	1.39	1.21	1.60	1.42E-05	1.27	0.033
rs1025818	3	16940025	C	T	*PLCL2*	0.334	0.359	3.00E-01	1.09	0.92	1.30	0.330	0.409	1.28E-06	1.41	1.23	1.62	9.49E-06	1.28	0.023
rs4618210	3	17099388	G	A	*PLCL2*	0.467	0.490	3.34E-02	1.20	1.01	1.41	0.468	0.473	8.60E-04	1.26	1.10	1.45	3.78E-05	1.25	0.513
rs1158265	3	24311166	C	T	*THRB*	0.274	0.275	7.04E-01	1.04	0.86	1.24	0.306	0.244	9.54E-05	0.73	0.63	0.86	8.57E-03	0.85	0.006
rs1394761	3	24390831	A	C	*THRB*	0.293	0.236	4.76E-03	0.76	0.63	0.92	0.363	0.312	4.30E-03	0.81	0.70	0.94	7.56E-05	0.79	0.626
rs4683017	3	45009415	C	T	*EXOSC7*	0.453	0.452	9.26E-01	0.99	0.84	1.17	0.470	0.401	4.42E-05	0.75	0.65	0.86	1.95E-03	0.85	0.013
rs11130040	3	45024190	A	G	*EXOSC7*	0.465	0.461	7.90E-01	0.98	0.83	1.15	0.487	0.419	6.89E-05	0.76	0.66	0.87	1.67E-03	0.84	0.022
rs1404100	3	100273457	A	G	*Hs.581579*	0.218	0.219	8.83E-01	1.02	0.83	1.24	0.199	0.259	2.96E-05	1.40	1.19	1.63	6.95E-04	1.24	0.012
rs13078855	3	120589967	T	C	*CDGAP*	0.200	0.150	3.15E-03	0.72	0.58	0.90	0.180	0.138	1.63E-03	0.73	0.60	0.89	9.50E-06	0.72	0.989
rs12494314[b]	3	120605510	C	T	*CDGAP*	0.211	0.138	1.86E-05	0.62	0.49	0.77	0.176	0.135	2.94E-03	0.75	0.61	0.90	1.59E-07	0.67	0.112
rs1132200	3	120633526	T	C	*TMEM39A*	0.197	0.125	1.27E-05	0.60	0.47	0.76	0.159	0.122	5.03E-03	0.75	0.61	0.92	3.87E-07	0.67	0.192
rs7650774[b]	3	120687740	C	T	*KTELC1*	0.211	0.138	1.90E-05	0.62	0.49	0.77	0.175	0.134	2.64E-03	0.74	0.61	0.90	1.32E-07	0.67	0.116
rs2293370	3	120702624	A	G	*C3orf1*	0.216	0.145	3.30E-05	0.63	0.51	0.78	0.182	0.140	2.28E-03	0.74	0.61	0.90	3.03E-07	0.68	0.320
rs6804441	3	120743634	G	A	*CD80*	0.194	0.139	9.37E-04	0.69	0.55	0.86	0.170	0.138	1.55E-02	0.79	0.65	0.96	4.48E-05	0.74	0.413
rs979903	3	136776944	C	T	*none*	0.244	0.317	9.21E-05	1.43	1.20	1.71	0.308	0.312	7.19E-01	1.03	0.89	1.19	1.09E-02	1.16	0.003
rs11917011	3	155711698	T	C	*none*	0.083	0.072	1.98E-01	0.82	0.60	1.12	0.065	0.031	6.21E-05	0.46	0.31	0.67	3.69E-04	0.64	0.018
rs4680514	3	160944150	A	G	*SCHIP1*	0.187	0.161	8.21E-02	0.82	0.66	1.03	0.236	0.180	1.54E-04	0.71	0.60	0.85	7.94E-05	0.76	0.370
rs1461100	3	160945597	A	G	*SCHIP1*	0.178	0.150	4.87E-02	0.80	0.63	1.00	0.227	0.175	3.29E-04	0.72	0.61	0.86	7.28E-05	0.76	0.562

rs924247	3	160963022	C	T	*SCHIP1*	0.469	0.399	4.08E-03	0.78	0.66	0.93	0.547	0.517	8.08E-04	0.79	0.91	0.69	1.17E-05	0.79	0.912
rs3863075	3	160985457	C	T	*SCHIP1*	0.281	0.224	5.34E-03	0.76	0.63	0.92	0.353	0.288	2.08E-04	0.76	0.66	0.88	3.05E-06	0.76	0.916
rs7636819	3	160988297	G	T	*SCHIP1*	0.280	0.222	5.35E-03	0.76	0.63	0.92	0.353	0.290	2.98E-04	0.76	0.66	0.88	4.31E-06	0.76	0.965
rs6441276	3	161034716	T	C	*SCHIP1*	0.190	0.156	7.01E-02	0.82	0.66	1.02	0.250	0.185	3.71E-05	0.70	0.59	0.83	1.16E-05	0.74	0.256
rs26298	3	161093985	G	A	*SCHIP1*	0.345	0.299	1.84E-02	0.81	0.68	0.96	0.436	0.359	3.43E-05	0.75	0.65	0.86	1.65E-06	0.76	0.413
rs17811014	3	161170723	T	C	*none*	0.173	0.199	1.20E-01	1.18	0.96	1.46	0.158	0.213	5.44E-05	1.41	1.20	1.67	2.32E-05	1.33	0.161
rs2243123	3	161192345	C	T	*IL12A*	0.239	0.181	9.44E-05	0.66	0.54	0.82	0.285	0.230	7.31E-04	0.76	0.65	0.89	4.57E-07	0.72	0.305
rs583911	3	161193084	G	A	*IL12A*	0.471	0.534	9.29E-04	1.32	1.12	1.56	0.441	0.488	2.93E-05	1.34	1.17	1.53	1.41E-07	1.32	0.961
rs6441284	3	161200962	G	A	*IL12A*	0.369	0.294	1.97E-05	0.68	0.57	0.81	0.393	0.339	2.08E-03	0.80	0.69	0.92	2.91E-07	0.75	0.185
rs485497	3	161201826	G	A	*IL12A*	0.439	0.392	9.07E-03	0.80	0.68	0.95	0.498	0.431	7.54E-05	0.76	0.66	0.87	3.15E-06	0.78	0.657
rs6441286	3	161211572	G	T	*IL12A*	0.444	0.487	1.25E-04	1.38	1.17	1.63	0.394	0.495	1.35E-08	1.48	1.30	1.70	7.63E-12	1.44	0.492
rs574808	3	161215677	C	T	*IL12A*	0.340	0.277	2.85E-04	0.72	0.60	0.86	0.421	0.328	2.19E-07	0.68	0.59	0.79	3.30E-10	0.70	0.706
rs10513549	3	161237201	A	G	*IL12A*	0.228	0.184	2.36E-03	0.73	0.59	0.89	0.277	0.230	4.70E-03	0.79	0.68	0.93	5.93E-05	0.77	0.468
rs1075498	3	161239145	A	G	*IL12A*	0.277	0.203	7.21E-06	0.64	0.52	0.78	0.290	0.235	6.43E-04	0.76	0.65	0.89	6.54E-08	0.71	0.158
rs6762377	3	161323322	A	G	*none*	0.078	0.069	4.25E-01	0.88	0.64	1.21	0.131	0.081	3.89E-05	0.60	0.47	0.77	2.01E-04	0.69	0.060
rs4679904	3	161823590	T	C	*ARF7*	0.219	0.210	7.31E-01	0.97	0.79	1.18	0.289	0.204	2.67E-07	0.65	0.55	0.76	5.31E-05	0.77	0.003
rs4434248	4	18514674	G	A	*none*	0.423	0.345	4.83E-05	0.70	0.59	0.83	0.370	0.375	8.78E-01	1.01	0.88	1.16	1.24E-02	0.87	0.001
rs1452554	4	18529262	T	C	*none*	0.442	0.363	2.33E-05	0.70	0.59	0.82	0.395	0.401	8.58E-01	1.01	0.88	1.16	9.61E-03	0.87	0.001
rs7678820	4	22901080	C	T	*none*	0.167	0.142	1.34E-01	0.84	0.67	1.06	0.181	0.121	8.06E-06	0.63	0.51	0.77	1.33E-05	0.71	0.057
rs1912031	4	22938137	A	G	*none*	0.175	0.140	2.67E-02	0.77	0.62	0.97	0.184	0.141	1.48E-03	0.74	0.61	0.89	8.31E-05	0.74	0.657
rs4692196	4	26949002	C	T	*none*	0.070	0.063	5.13E-01	0.90	0.65	1.24	0.071	0.105	9.79E-05	1.57	1.25	1.96	7.73E-03	1.29	0.008
rs2903896	4	83892809	T	C	*SCD5*	0.198	0.268	8.68E-05	1.47	1.21	1.77	0.177	0.173	6.41E-01	0.96	0.80	1.15	1.36E-02	1.18	0.002
rs2869442	4	87640302	A	G	*MAPK10*	0.058	0.060	8.10E-01	1.04	0.74	1.47	0.051	0.083	5.22E-05	1.69	1.31	2.19	5.36E-04	1.43	0.023
rs10516775	4	87840497	C	T	*PTPN13*	0.056	0.054	9.09E-01	0.98	0.69	1.40	0.048	0.078	9.90E-05	1.68	1.30	2.19	1.67E-03	1.40	0.015
rs7665090	4	103770651	A	G	*MANBA*	0.491	0.448	1.45E-03	0.77	0.65	0.90	0.496	0.444	3.75E-03	0.82	0.71	0.94	3.47E-05	0.80	0.488
rs2866413	4	103776125	G	A	*MANBA*	0.490	0.450	1.78E-03	0.77	0.65	0.91	0.496	0.444	3.83E-03	0.82	0.71	0.94	4.11E-05	0.80	0.518
rs228611	4	103780757	A	G	*MANBA*	0.489	0.434	4.18E-03	0.79	0.67	0.93	0.482	0.428	2.52E-03	0.81	0.71	0.93	5.44E-05	0.81	0.709
rs228614	4	103797685	A	G	*MANBA*	0.496	0.438	2.31E-03	0.77	0.66	0.91	0.487	0.430	1.57E-03	0.80	0.70	0.92	2.19E-05	0.80	0.667
rs223420	4	103947120	A	C	*UBE2D3P*	0.465	0.529	7.94E-04	1.32	1.12	1.56	0.474	0.480	1.40E-02	1.19	1.04	1.36	7.66E-05	1.23	0.278
rs4698874	4	104149960	C	T	*NHEDC1*	0.458	0.480	1.05E-03	1.32	1.12	1.55	0.475	0.478	1.02E-02	1.19	1.04	1.37	5.83E-05	1.24	0.353
rs10516496	4	104161614	T	C	*NHEDC2*	0.341	0.391	3.27E-03	1.29	1.09	1.53	0.332	0.381	6.59E-03	1.21	1.06	1.39	8.98E-05	1.24	0.572
rs11946020	4	104164191	C	T	*NHEDC2*	0.343	0.394	2.96E-03	1.29	1.09	1.53	0.333	0.381	7.21E-03	1.21	1.05	1.39	8.47E-05	1.24	0.553
rs1358229	4	123116184	T	C	*TRPC3*	0.482	0.483	8.23E-02	1.16	0.98	1.36	0.445	0.477	1.17E-05	1.36	1.18	1.55	6.24E-06	1.27	0.140
rs6838639	4	123118615	A	G	*TRPC3*	0.250	0.224	1.13E-01	0.85	0.70	1.04	0.259	0.190	3.65E-06	0.67	0.56	0.79	6.63E-06	0.74	0.068
rs13144383	4	139310254	G	A	*SLC7A11*	0.148	0.190	5.92E-03	1.35	1.09	1.68	0.160	0.196	2.04E-03	1.32	1.11	1.57	6.21E-05	1.32	0.748
rs2291713	4	141764847	C	A	*TBC1D9*	0.139	0.134	4.77E-01	0.92	0.72	1.16	0.135	0.189	1.74E-05	1.48	1.24	1.76	2.66E-03	1.24	0.002
rs931283	5	2440550	T	C	*none*	0.447	0.459	2.68E-01	1.10	0.93	1.29	0.485	0.409	5.23E-05	0.75	0.65	0.86	1.44E-02	0.88	0.000
rs2923764	5	18216677	G	A	*ARSJ*	0.496	0.424	7.14E-05	0.72	0.61	0.84	0.436	0.425	4.10E-01	0.94	0.82	1.08	2.73E-03	0.85	0.008
rs4576132	5	158916817	A	G	*none*	0.403	0.398	7.70E-01	0.98	0.83	1.15	0.341	0.406	9.68E-05	1.32	1.15	1.51	5.65E-03	1.16	0.007
rs12206548	6	451231	G	A	*EXOSC2*	0.402	0.401	9.32E-01	1.01	0.85	1.19	0.413	0.347	7.40E-05	0.75	0.65	0.87	3.97E-03	0.85	0.010
rs9392056	6	463078	G	A	*EXOSC2*	0.395	0.397	8.14E-01	1.02	0.86	1.21	0.389	0.318	2.19E-05	0.73	0.63	0.85	2.96E-03	0.85	0.004
rs6918152	6	487159	A	G	*EXOSC2*	0.395	0.393	9.75E-01	1.00	0.84	1.18	0.388	0.317	1.58E-05	0.73	0.63	0.84	1.34E-03	0.84	0.006
rs9885921	6	14240590	G	A	*CD83*	0.427	0.410	3.88E-01	0.93	0.79	1.10	0.510	0.577	4.22E-05	1.33	1.52	1.16	1.29E-02	1.14	0.002
rs2244839	6	31546347	A	G	*Hs.615398*	0.369	0.366	5.07E-01	0.94	0.80	1.12	0.255	0.328	5.34E-05	1.35	1.17	1.57	5.42E-03	1.17	0.001
rs1055569	6	31548061	T	C	*Hs.615398*	0.457	0.474	6.34E-01	1.04	0.88	1.23	0.327	0.408	1.68E-05	1.36	1.18	1.57	1.89E-04	1.22	0.011
rs2516440	6	31548476	A	G	*Hs.615398*	0.460	0.477	6.45E-01	1.04	0.88	1.22	0.332	0.410	3.87E-05	1.34	1.17	1.54	3.42E-04	1.21	0.015
rs2395488	6	31553888	G	A	*none*	0.351	0.369	5.33E-01	1.06	0.89	1.25	0.342	0.412	1.95E-05	1.35	1.18	1.54	9.35E-05	1.24	0.018
rs2523647	6	31557757	G	A	*Hs.692412*	0.276	0.304	2.26E-01	1.12	0.93	1.34	0.218	0.280	4.86E-05	1.37	1.18	1.60	3.72E-05	1.28	0.054
rs2269475	6	31691910	T	C	*AIF1*	0.072	0.107	3.92E-04	1.65	1.25	2.19	0.122	0.160	1.58E-04	1.43	1.19	1.73	5.56E-07	1.49	0.382
rs1046089	6	31710946	A	G	*BAT2*	0.271	0.339	8.52E-05	1.42	1.19	1.70	0.338	0.382	1.82E-03	1.25	1.09	1.43	3.13E-06	1.30	0.197
rs805303	6	31724345	A	G	*BAT3*	0.318	0.379	5.99E-04	1.35	1.14	1.60	0.364	0.407	7.73E-03	1.21	1.05	1.38	4.81E-05	1.25	0.267
rs3130617	6	31735502	C	T	*C6orf47*	0.182	0.138	4.11E-03	0.72	0.57	0.90	0.260	0.208	2.14E-03	0.77	0.65	0.91	2.76E-05	0.75	0.624
rs2280800	6	31754377	A	C	*LY6G5C*	0.076	0.114	2.26E-04	1.66	1.27	2.18	0.123	0.163	1.17E-04	1.44	1.20	1.74	2.52E-07	1.50	0.373
rs805274	6	31773173	C	T	*BAT5*	0.272	0.340	9.67E-05	1.42	1.19	1.69	0.258	0.305	3.67E-03	1.24	1.07	1.44	4.03E-06	1.31	0.237
rs2242653	6	31783744	A	G	*C6orf21*	0.098	0.150	1.71E-05	1.69	1.33	2.16	0.141	0.181	2.19E-04	1.40	1.17	1.68	6.41E-08	1.49	0.197
rs480092	6	31872878	C	T	*HSP70*	0.145	0.200	4.95E-05	1.54	1.25	1.91	0.162	0.178	1.15E-01	1.16	0.97	1.38	2.13E-04	1.29	0.032
rs2227956	6	31886251	G	A	*HSPA1L*	0.094	0.072	6.40E-02	0.75	0.56	1.02	0.182	0.126	8.52E-05	0.67	0.54	0.82	2.47E-05	0.70	0.544
rs2763979	6	31902571	T	C	*HSPA1B*	0.325	0.391	2.68E-04	1.37	1.16	1.63	0.345	0.391	3.31E-03	1.23	1.07	1.41	6.86E-06	1.28	0.313
rs9267649	6	31932807	A	G	*NEU1*	0.101	0.072	2.73E-02	0.72	0.53	0.97	0.181	0.125	7.57E-05	0.66	0.54	0.81	9.33E-06	0.69	0.711
rs3134954	6	32179871	C	T	*TNXB*	0.074	0.052	4.44E-02	0.70	0.50	0.99	0.144	0.095	1.35E-04	0.64	0.51	0.81	1.06E-05	0.65	0.616

rs2269425	6	32231617	A	G	*PPT2*	0.085	0.119	4.60E-03	1.47	1.12	1.91	0.136	0.179	6.91E-05	1.43	1.20	1.71	4.06E-07	1.47	0.977
rs3134603	6	32233980	A	G	*PPT2*	0.089	0.065	5.59E-02	0.74	0.54	1.01	0.140	0.088	2.39E-05	0.60	0.48	0.76	3.30E-06	0.64	0.268
rs3134943	6	32255739	T	C	*RNF5*	0.077	0.050	1.84E-02	0.66	0.46	0.93	0.143	0.084	4.64E-06	0.57	0.45	0.73	1.70E-07	0.59	0.472
rs3132946	6	32298006	A	G	*NOTCH4*	0.061	0.042	7.57E-02	0.71	0.48	1.04	0.139	0.082	8.05E-06	0.58	0.45	0.73	1.33E-06	0.60	0.342
rs3130304[b]	6	32315159	A	G	*none*	0.349	0.273	6.67E-05	0.69	0.58	0.83	0.241	0.221	1.03E-01	0.87	0.74	1.03	1.29E-04	0.79	0.082
rs411326	6	32319295	T	C	*LOC401252*	0.234	0.271	2.56E-02	1.24	1.03	1.49	0.257	0.310	1.59E-04	1.33	1.15	1.55	6.96E-06	1.30	0.484
rs9267992	6	32328375	G	A	*LOC401252*	0.066	0.051	1.59E-01	0.77	0.54	1.10	0.142	0.091	4.98E-05	0.62	0.49	0.78	2.36E-05	0.66	0.286
rs570963[b]	6	32397572	G	A	*C6orf10*	0.158	0.246	8.41E-08	1.72	1.41	2.11	0.098	0.142	5.05E-05	1.51	1.24	1.85	2.18E-11	1.62	0.321
rs2076537	6	32425613	A	G	*C6orf10*	0.321	0.400	6.35E-05	1.41	1.19	1.67	0.355	0.389	1.52E-02	1.19	1.03	1.37	6.37E-06	1.28	0.148
rs2395148	6	32429532	T	G	*C6orf10*	0.021	0.056	8.42E-07	2.90	1.87	4.49	0.023	0.056	1.77E-09	2.67	1.94	3.68	9.95E-15	2.75	0.776
rs1265761[b]	6	32429575	G	A	*C6orf10*	0.087	0.143	3.72E-05	1.69	1.31	2.17	0.051	0.060	3.11E-01	1.16	0.87	1.54	1.01E-04	1.45	0.036
rs1265759[b]	6	32430371	C	T	*C6orf10*	0.387	0.468	9.28E-05	1.39	1.18	1.64	0.421	0.456	2.90E-02	1.17	1.02	1.34	1.88E-05	1.26	0.093
rs1265758[b]	6	32431507	A	G	*C6orf10*	0.382	0.471	1.55E-05	1.44	1.22	1.70	0.414	0.450	2.74E-02	1.17	1.02	1.34	6.03E-06	1.28	0.050
rs6907322	6	32432923	A	G	*C6orf10*	0.211	0.309	2.67E-08	1.68	1.40	2.02	0.178	0.237	6.70E-06	1.46	1.24	1.72	1.27E-12	1.55	0.253
rs2395150	6	32434023	G	A	*C6orf10*	0.381	0.467	3.50E-05	1.42	1.20	1.67	0.414	0.450	2.68E-02	1.17	1.02	1.34	1.23E-05	1.26	0.081
rs2073048	6	32443411	A	G	*C6orf10*	0.124	0.166	1.05E-03	1.47	1.17	1.85	0.127	0.176	1.05E-05	1.50	1.25	1.80	1.74E-08	1.50	0.781
rs3129943	6	32446673	G	A	*C6orf10*	0.213	0.286	9.31E-05	1.45	1.20	1.75	0.246	0.264	1.26E-01	1.13	0.97	1.32	2.28E-04	1.25	0.043
rs2395157	6	32456123	G	A	*none*	0.231	0.226	5.90E-01	0.95	0.78	1.15	0.272	0.344	3.37E-06	1.41	1.22	1.62	2.96E-04	1.24	0.001
rs4424066	6	32462406	G	A	*none*	0.345	0.357	6.18E-01	1.04	0.88	1.24	0.415	0.498	7.18E-07	1.40	1.23	1.60	1.71E-05	1.26	0.005
rs3117098	6	32466491	G	A	*BTNL2*	0.408	0.380	1.96E-01	0.90	0.76	1.06	0.317	0.224	2.09E-09	0.61	0.52	0.72	8.88E-08	0.73	0.001
rs2076530	6	32471794	C	T	*BTNL2*	0.371	0.368	8.98E-01	0.99	0.84	1.17	0.423	0.497	1.77E-06	1.38	1.21	1.58	1.56E-04	1.23	0.001
rs4248166	6	32474399	C	T	*BTNL2*	0.134	0.185	1.83E-04	1.52	1.22	1.89	0.160	0.216	3.11E-06	1.48	1.26	1.75	3.47E-09	1.49	0.868
rs3763305	6	32477466	A	G	*BTNL2*	0.024	0.035	1.37E-01	1.42	0.89	2.26	0.046	0.076	2.24E-05	1.78	1.36	2.33	9.11E-06	1.68	0.404
rs10947262[b]	6	32481290	T	C	*BTNL2*	0.088	0.139	5.99E-05	1.67	1.30	2.16	0.075	0.109	1.79E-04	1.53	1.23	1.91	3.20E-08	1.60	0.532
rs3806156	6	32481676	T	G	*BTNL2*	0.320	0.365	3.89E-02	1.20	1.01	1.42	0.349	0.453	3.45E-10	1.54	1.35	1.77	1.74E-10	1.42	0.012
rs3763313	6	32484449	C	A	*BTNL2*	0.167	0.225	1.01E-04	1.49	1.22	1.83	0.188	0.247	1.82E-05	1.41	1.20	1.65	4.24E-09	1.46	0.767
rs2395163	6	32495787	C	T	*BTNL2*	0.147	0.176	5.56E-02	1.24	0.99	1.54	0.201	0.258	2.13E-05	1.41	1.20	1.66	2.99E-06	1.36	0.325
rs3135363	6	32497626	G	A	*BTNL2*	0.363	0.260	4.80E-08	0.60	0.50	0.72	0.292	0.218	9.10E-07	0.67	0.57	0.78	2.00E-13	0.64	0.467
rs9501626	6	32508322	A	C	*HLA-DRA*	0.104	0.135	2.52E-02	1.33	1.03	1.71	0.109	0.174	3.15E-08	1.69	1.40	2.03	4.72E-09	1.56	0.119
rs2395175	6	32513004	A	G	*HLA-DRA*	0.057	0.074	6.83E-02	1.35	0.98	1.87	0.154	0.194	1.80E-04	1.40	1.18	1.68	2.11E-05	1.40	0.826
rs3129882	6	32517508	A	G	*HLA-DRA*	0.442	0.432	6.42E-01	0.96	0.82	1.13	0.567	0.634	2.58E-05	1.35	1.56	1.18	4.01E-03	1.17	0.002
rs6903608	6	32536263	C	T	*HLA-DRA*	0.462	0.428	9.76E-02	0.87	0.74	1.03	0.321	0.239	1.02E-07	0.65	0.56	0.76	4.96E-07	0.75	0.012
rs2395185	6	32541145	T	G	*HLA-DRA*	0.237	0.251	5.42E-01	1.06	0.88	1.28	0.321	0.388	5.33E-05	1.34	1.16	1.54	1.47E-04	1.24	0.041
rs660895	6	32685358	G	A	*HLA-DRB1*	0.102	0.177	2.41E-08	1.93	1.53	2.44	0.190	0.271	1.48E-09	1.62	1.39	1.90	1.14E-16	1.73	0.266
rs17533090[b]	6	32698700	T	G	*HLA-DQA1*	0.161	0.233	2.10E-05	1.55	1.27	1.90	0.159	0.190	2.47E-02	1.22	1.03	1.45	5.14E-06	1.36	0.067
rs3129763	6	32698903	A	G	*HLA-DQA1*	0.347	0.270	3.80E-05	0.69	0.57	0.82	0.240	0.188	9.62E-05	0.71	0.60	0.84	2.16E-08	0.70	0.732
rs9275184	6	32762692	C	T	*HLA-DQA1*	0.053	0.073	5.27E-02	1.38	0.99	1.92	0.100	0.145	3.24E-05	1.52	1.25	1.84	1.40E-06	1.52	0.524
rs7774434[b]	6	32765556	C	T	*HLA-DQB1*	0.346	0.475	2.33E-10	1.71	1.45	2.02	0.371	0.477	3.50E-18	1.82	1.59	2.09	2.38E-26	1.75	0.377
rs7775228	6	32766057	C	T	*HLA-DQB1*	0.149	0.186	1.16E-02	1.32	1.06	1.64	0.126	0.202	1.32E-10	1.77	1.48	2.10	9.74E-11	1.56	0.047
rs2647012	6	32772436	T	C	*HLA-DQB1*	0.303	0.314	5.09E-01	1.06	0.89	1.27	0.396	0.317	1.80E-05	0.73	0.63	0.84	2.37E-03	0.84	0.001
rs9357152	6	32772938	G	A	*HLA-DQB1*	0.396	0.309	5.10E-06	0.67	0.56	0.79	0.258	0.191	1.25E-06	0.65	0.55	0.78	4.24E-11	0.66	0.934
rs9275312	6	32773706	G	A	*HLA-DQB1*	0.084	0.148	1.42E-07	1.96	1.52	2.52	0.130	0.219	2.77E-13	1.88	1.59	2.22	2.14E-20	1.94	0.942
rs9275374[b]	6	32776504	T	C	*HLA-DQB1*	0.180	0.259	4.11E-07	1.65	1.36	2.01	0.246	0.364	4.45E-14	1.73	1.50	1.99	3.70E-19	1.69	0.581
rs9275388[b]	6	32777062	C	T	*HLA-DQB1*	0.179	0.261	1.98E-07	1.67	1.38	2.03	0.245	0.364	3.07E-14	1.73	1.50	1.99	1.50E-19	1.69	0.610
rs9275390	6	32777134	C	T	*HLA-DQB1*	0.180	0.262	1.90E-07	1.67	1.38	2.03	0.246	0.364	4.33E-14	1.73	1.50	1.99	3.31E-21	1.74	0.609
rs9275393[b]	6	32777417	A	G	*HLA-DQB1*	0.180	0.261	2.16E-07	1.67	1.37	2.03	0.246	0.364	4.47E-14	1.73	1.50	1.99	2.38E-19	1.69	0.619
rs2647050	6	32777745	C	T	*HLA-DQB1*	0.524	0.427	4.86E-07	0.66	0.56	0.77	0.363	0.321	3.31E-03	0.80	0.70	0.93	2.58E-08	0.73	0.074
rs9275406[b]	6	32777933	T	C	*HLA-DQB1*	0.180	0.261	2.16E-07	1.67	1.37	2.03	0.246	0.364	4.45E-14	1.73	1.50	1.99	2.38E-19	1.69	0.619
rs9275407[b]	6	32778015	T	G	*HLA-DQB1*	0.179	0.261	2.12E-07	1.67	1.38	2.03	0.246	0.364	5.85E-14	1.72	1.49	1.98	2.94E-19	1.69	0.636
rs9275418[b]	6	32778222	G	A	*HLA-DQB1*	0.179	0.261	1.96E-07	1.67	1.38	2.03	0.246	0.364	6.93E-14	1.72	1.49	1.98	3.17E-19	1.69	0.657
rs2856718	6	32778233	T	C	*HLA-DQB1*	0.524	0.426	3.63E-07	0.65	0.55	0.77	0.362	0.320	2.77E-03	0.80	0.69	0.93	1.53E-08	0.73	0.076
rs9275424[b]	6	32778554	G	A	*DQB1/DQA2 in*	0.179	0.264	9.21E-08	1.70	1.40	2.06	0.246	0.364	6.93E-14	1.72	1.49	1.98	1.55E-19	1.70	0.742
rs9275425[b]	6	32778852	A	C	*DQB1/DQA2 in*	0.180	0.257	6.45E-07	1.64	1.35	1.99	0.246	0.362	9.42E-14	1.71	1.49	1.97	1.21E-18	1.67	0.575
rs9275427[b]	6	32778893	T	C	*DQB1/DQA2 in*	0.180	0.261	2.16E-07	1.67	1.37	2.03	0.246	0.362	9.42E-14	1.71	1.49	1.97	4.75E-19	1.68	0.660
rs9275428[b]	6	32778956	G	A	*DQB1/DQA2 in*	0.180	0.261	2.16E-07	1.67	1.37	2.03	0.245	0.362	8.97E-14	1.71	1.49	1.97	4.75E-19	1.68	0.660
rs13192471[b]	6	32779081	C	T	*DQB1/DQA2 in*	0.121	0.184	2.09E-06	1.71	1.37	2.14	0.141	0.218	5.62E-10	1.70	1.44	2.01	2.46E-14	1.68	0.824
rs9275439[b]	6	32779499	C	T	*DQB1/DQA2 in*	0.180	0.261	2.16E-07	1.67	1.37	2.03	0.245	0.362	8.04E-14	1.72	1.49	1.98	4.08E-19	1.68	0.649
rs16898264[b]	6	32785130	A	G	*DQB1/DQA2 in*	0.524	0.425	4.05E-07	0.65	0.55	0.77	0.353	0.323	4.90E-03	0.81	0.70	0.94	8.92E-08	0.74	0.065

rs7745656	6	32788948	T	G	*HLA-DQA2*	0.486	0.400	6.65E-06	0.68	0.58	0.81	0.282	0.269	1.83E-01	0.90	0.77	1.05	4.01E-05	0.79	0.019
rs9276644	6	32853021	C	T	*HLA-DQA2*	0.427	0.387	2.24E-02	0.82	0.70	0.97	0.341	0.289	7.99E-04	0.78	0.67	0.90	6.52E-05	0.80	0.618
rs719654	6	32860117	A	G	*none*	0.213	0.225	4.16E-01	1.08	0.89	1.32	0.224	0.284	4.84E-05	1.37	1.18	1.59	2.98E-04	1.25	0.074
rs2219893	6	32877641	C	T	*none*	0.325	0.380	1.66E-03	1.31	1.11	1.56	0.321	0.378	6.99E-04	1.27	1.11	1.47	7.41E-06	1.28	0.714
rs3129299	6	33008765	T	C	*HLA-DMB*	0.201	0.243	1.59E-02	1.27	1.05	1.54	0.163	0.207	1.80E-03	1.31	1.11	1.55	6.56E-05	1.30	0.773
rs2395296	6	33019792	A	G	*HLA-DMB*	0.285	0.266	1.93E-01	0.89	0.74	1.06	0.314	0.252	6.77E-05	0.73	0.62	0.85	9.78E-05	0.79	0.111
rs194682	6	33022591	T	C	*HLA-DMB*	0.342	0.383	1.85E-02	1.22	1.03	1.45	0.333	0.388	9.18E-04	1.27	1.10	1.46	6.11E-05	1.25	0.784
rs209473	6	33030886	A	C	*HLA-DMA*	0.478	0.438	2.75E-02	0.83	0.71	0.98	0.451	0.391	2.64E-04	0.77	0.67	0.89	3.77E-05	0.80	0.548
rs209474	6	33032562	G	A	*HLA-DMA*	0.393	0.432	2.71E-02	1.21	1.02	1.42	0.404	0.465	4.35E-04	1.28	1.12	1.47	4.58E-05	1.25	0.611
rs9277535	6	33162839	G	A	*HLA-DPB1*	0.242	0.306	2.00E-04	1.41	1.17	1.68	0.242	0.327	2.58E-09	1.57	1.35	1.82	1.95E-12	1.50	0.344
rs9277554	6	33163516	T	C	*HLA-DPB1*	0.281	0.341	4.98E-04	1.36	1.14	1.62	0.291	0.358	3.69E-06	1.41	1.22	1.63	6.10E-09	1.39	0.787
rs9277565	6	33164875	T	C	*HLA-DPB1*	0.202	0.256	5.95E-04	1.40	1.15	1.69	0.203	0.280	1.03E-08	1.57	1.35	1.83	2.62E-11	1.50	0.341
rs2281389	6	33167774	G	A	*HLA-DPB1*	0.175	0.234	1.30E-04	1.48	1.21	1.80	0.171	0.244	9.21E-09	1.61	1.37	1.90	6.44E-12	1.55	0.525
rs3128917	6	33167974	G	T	*HLA-DPB1*	0.241	0.294	1.05E-03	1.36	1.13	1.63	0.252	0.313	1.41E-05	1.39	1.20	1.62	5.28E-08	1.38	0.846
rs3117222	6	33168927	T	C	*HLA-DPB1*	0.241	0.294	1.05E-03	1.36	1.13	1.63	0.253	0.313	1.52E-05	1.39	1.20	1.61	5.73E-08	1.37	0.855
rs2064478	6	33180244	T	C	*HLA-DPB2*	0.220	0.278	2.77E-04	1.41	1.17	1.70	0.221	0.276	3.75E-05	1.39	1.19	1.62	5.08E-08	1.39	0.829
rs3117230	6	33183613	G	A	*HLA-DPB2*	0.219	0.278	2.69E-04	1.41	1.17	1.71	0.221	0.275	5.07E-05	1.38	1.18	1.61	6.52E-08	1.39	0.793
rs1883414	6	33194426	A	G	*HLA-DPB2*	0.343	0.354	4.57E-01	1.07	0.90	1.27	0.309	0.376	1.39E-05	1.36	1.19	1.57	1.35E-04	1.24	0.029
rs3117016	6	33203494	A	G	*HLA-DPB2*	0.408	0.445	8.51E-02	1.16	0.98	1.36	0.394	0.464	1.06E-04	1.31	1.14	1.51	1.64E-05	1.26	0.185
rs6901221	6	33206254	C	A	*none*	0.121	0.172	3.14E-04	1.52	1.21	1.91	0.158	0.215	3.22E-06	1.50	1.26	1.77	2.15E-09	1.51	0.967
rs2855430	6	33249258	A	G	*COL11A2*	0.098	0.132	8.27E-03	1.40	1.09	1.81	0.133	0.191	2.96E-07	1.59	1.33	1.91	9.65E-09	1.53	0.433
rs2855459	6	33262634	A	G	*COL11A2*	0.096	0.128	1.46E-02	1.38	1.07	1.78	0.133	0.192	1.54E-07	1.61	1.35	1.93	1.02E-08	1.53	0.335
rs439205	6	33281820	A	G	*HSD17B8*	0.240	0.264	1.96E-01	1.13	0.94	1.36	0.236	0.314	3.90E-08	1.51	1.31	1.76	3.10E-07	1.35	0.016
rs421446	6	33282761	G	A	*HSD17B8*	0.287	0.323	6.79E-02	1.18	0.99	1.41	0.274	0.356	1.14E-07	1.47	1.28	1.70	9.06E-08	1.35	0.045
rs4706258	6	70409857	G	A	*none*	0.221	0.155	2.16E-05	0.63	0.50	0.78	0.285	0.279	9.73E-01	1.00	0.86	1.16	1.64E-02	0.86	0.001
rs13216266	6	140059450	T	G	*Hs.544400*	0.329	0.294	4.01E-02	0.83	0.70	0.99	0.328	0.397	1.99E-05	1.35	1.18	1.56	3.73E-02	1.12	0.000
rs6929952	6	158571383	T	C	*Hs.237825*	0.398	0.395	6.88E-01	1.04	0.88	1.22	0.417	0.346	5.52E-05	0.75	0.65	0.86	5.79E-03	0.86	0.004
rs2197100	6	165551781	A	G	*none*	0.232	0.161	8.09E-05	0.65	0.53	0.81	0.260	0.259	9.82E-01	1.00	0.86	1.17	2.29E-02	0.86	0.001
rs10250643	7	24502175	G	T	*none*	0.229	0.203	1.06E-01	0.85	0.69	1.04	0.278	0.208	8.83E-06	0.69	0.58	0.81	1.25E-05	0.75	0.139
rs2255365	7	24528186	C	A	*none*	0.436	0.411	1.43E-01	0.88	0.75	1.04	0.476	0.408	6.07E-05	0.76	0.66	0.87	9.23E-05	0.81	0.180
rs1476520	7	24762754	C	T	*DFNA5*	0.416	0.426	7.73E-01	1.03	0.87	1.21	0.474	0.402	5.06E-05	0.75	0.66	0.86	6.18E-03	0.86	0.008
rs1012775	7	24815354	A	G	*OSBPL3*	0.413	0.398	4.27E-01	0.93	0.79	1.10	0.345	0.416	2.67E-05	1.35	1.17	1.55	8.31E-03	1.15	0.001
rs7785415	7	25877852	G	A	*none*	0.149	0.133	2.44E-01	0.87	0.68	1.10	0.092	0.138	9.06E-05	1.49	1.22	1.82	1.75E-02	1.21	0.000
rs4728142	7	128361203	A	G	*IRF5/TNPO3*	0.424	0.479	9.48E-03	1.24	1.05	1.46	0.445	0.498	1.43E-03	1.25	1.09	1.43	5.97E-05	1.24	0.976
rs10488631	7	128381419	C	T	*IRF5/TNPO3*	0.083	0.121	4.24E-03	1.48	1.13	1.93	0.107	0.169	2.17E-08	1.71	1.42	2.06	2.78E-10	1.63	0.365
rs11784810	8	24482929	G	T	*none*	0.373	0.413	3.05E-02	1.20	1.02	1.42	0.373	0.432	8.54E-04	1.26	1.10	1.45	7.16E-05	1.24	0.626
rs12678383	8	24498107	T	G	*none*	0.255	0.289	4.75E-02	1.20	1.00	1.44	0.247	0.300	4.56E-04	1.30	1.12	1.51	7.50E-05	1.26	0.493
rs7821268	8	29520356	C	T	*none*	0.227	0.218	4.52E-01	0.93	0.76	1.13	0.213	0.281	8.49E-07	1.48	1.26	1.73	7.78E-04	1.23	0.000
rs4732980	8	29529350	A	G	*none*	0.333	0.315	1.86E-01	0.89	0.74	1.06	0.306	0.380	4.62E-06	1.39	1.21	1.59	6.25E-03	1.17	0.000
rs10866875	8	29532541	G	T	*none*	0.201	0.193	5.53E-01	0.94	0.76	1.15	0.185	0.247	4.27E-06	1.47	1.25	1.72	1.35E-03	1.23	0.001
rs12334531	8	74426184	T	C	*RDH10*	0.445	0.361	9.86E-06	0.68	0.58	0.81	0.423	0.400	2.29E-01	0.92	0.80	1.05	1.13E-04	0.81	0.011
rs279479	8	94180024	C	T	*none*	0.521	0.453	1.25E-03	0.76	0.65	0.90	0.489	0.447	1.11E-02	0.84	0.73	0.96	6.36E-05	0.81	0.390
rs7813101	8	97733748	A	G	*PGCP*	0.260	0.329	3.70E-04	1.38	1.16	1.65	0.288	0.328	1.46E-02	1.20	1.04	1.39	2.43E-05	1.27	0.252
rs7834444	8	99455892	T	C	*Hs.386298*	0.315	0.348	1.36E-01	1.14	0.96	1.35	0.318	0.249	2.50E-05	0.72	0.61	0.84	3.24E-02	0.88	0.000
rs7838130	8	99456091	T	C	*Hs.386298*	0.316	0.348	1.43E-01	1.14	0.96	1.35	0.318	0.249	2.47E-05	0.72	0.61	0.84	3.10E-02	0.88	0.000
rs4259391	8	99460461	T	C	*Hs.386298*	0.300	0.331	9.77E-02	1.16	0.97	1.38	0.310	0.242	3.05E-05	0.72	0.61	0.84	4.34E-02	0.89	0.000
rs6468721	8	101589274	C	T	*none*	0.232	0.259	1.14E-01	1.16	0.96	1.41	0.259	0.198	2.90E-05	0.70	0.59	0.83	3.35E-02	0.87	0.000
rs2935548	8	101749531	G	A	*none*	0.148	0.166	1.92E-01	1.16	0.93	1.45	0.095	0.138	8.21E-05	1.49	1.22	1.82	3.33E-04	1.31	0.141
rs1529335	8	129085529	T	C	*PVT1*	0.108	0.071	1.97E-03	0.63	0.46	0.84	0.172	0.133	3.79E-03	0.75	0.61	0.91	2.12E-05	0.70	0.376
rs895236	9	4028228	T	C	*GLIS3*	0.115	0.077	1.28E-03	0.62	0.46	0.83	0.132	0.102	1.20E-02	0.75	0.60	0.94	5.83E-05	0.69	0.323
rs2381904	9	8835177	A	C	*PTPRD*	0.311	0.291	2.64E-01	0.90	0.76	1.08	0.256	0.203	4.13E-05	0.70	0.59	0.83	4.65E-04	0.80	0.078
rs1972797	9	8837353	C	A	*PTPRD*	0.487	0.465	2.18E-01	0.90	0.77	1.06	0.428	0.365	9.13E-05	0.75	0.65	0.87	3.62E-04	0.82	0.137
rs10511828	9	28618500	C	T	*LINGO2*	0.064	0.028	1.41E-05	0.38	0.24	0.60	0.068	0.073	5.61E-01	1.08	0.83	1.40	1.05E-01	0.83	0.000
rs1529952	9	85909367	T	C	*none*	0.316	0.367	4.80E-03	1.28	1.08	1.52	0.336	0.386	1.31E-03	1.26	1.09	1.44	1.64E-05	1.27	0.911
rs3124607	9	138534660	A	G	*NOTCH1*	0.294	0.281	3.34E-01	0.91	0.76	1.10	0.328	0.257	1.20E-05	0.71	0.61	0.83	7.93E-05	0.79	0.037
rs7477689	10	30055806	G	A	*SVIL*	0.127	0.129	9.51E-01	0.99	0.78	1.27	0.092	0.135	3.82E-05	1.54	1.25	1.89	1.82E-03	1.28	0.008
rs1927461	10	30084320	C	T	*none*	0.185	0.173	2.73E-01	0.89	0.72	1.10	0.144	0.198	2.87E-05	1.45	1.22	1.73	8.40E-03	1.20	0.000
rs10993994	10	51219502	T	C	*MSMB*	0.428	0.477	3.54E-02	1.19	1.01	1.41	0.389	0.455	4.27E-04	1.28	1.12	1.46	2.92E-05	1.25	0.470
rs2345201	10	82662797	G	A	*none*	0.187	0.161	1.07E-01	0.84	0.67	1.04	0.168	0.220	5.47E-05	1.40	1.19	1.66	1.67E-02	1.17	0.000
rs12781461	10	82915023	T	C	*none*	0.336	0.405	2.69E-04	1.37	1.15	1.62	0.317	0.361	7.52E-03	1.21	1.05	1.40	9.41E-06	1.28	0.314

rs10886214	10	85117719	T	C	*none*	0.183	0.259	7.86E-06	1.56	1.28	1.89	0.217	0.227	4.77E-01	1.06	0.90	1.25	1.41E-03	1.22	0.002
rs10886221	10	85123936	T	C	*none*	0.240	0.318	1.30E-05	1.49	1.25	1.79	0.277	0.285	5.53E-01	1.05	0.90	1.21	1.93E-03	1.20	0.002
rs7068676	10	85173709	C	T	*none*	0.140	0.202	3.12E-05	1.58	1.27	1.95	0.161	0.155	6.27E-01	0.96	0.80	1.15	2.50E-02	1.17	0.000
rs11198735	10	85193717	A	C	*none*	0.099	0.156	3.25E-05	1.67	1.31	2.12	0.107	0.104	8.04E-01	0.97	0.78	1.21	1.19E-02	1.23	0.001
rs11190842	10	102885447	A	C	*TLX1*	0.125	0.090	1.18E-02	0.70	0.53	0.92	0.124	0.082	4.71E-04	0.65	0.51	0.83	2.67E-05	0.68	0.725
rs9420843	10	103254177	T	C	*BTRC*	0.490	0.498	8.48E-01	1.02	0.86	1.20	0.520	0.413	6.15E-05	0.76	0.66	0.87	1.91E-03	1.18	0.018
rs4910793	11	5419637	C	T	*OR51I1*	0.047	0.091	2.27E-06	2.12	1.54	2.92	0.061	0.048	8.46E-02	0.76	0.56	1.04	3.77E-02	1.27	0.000
rs541207	11	63881718	G	T	*none*	0.404	0.359	1.74E-02	0.81	0.69	0.96	0.454	0.382	5.29E-05	0.75	0.65	0.86	5.40E-06	0.78	0.521
rs538147	11	63886298	A	G	*RPS6KA4*	0.356	0.291	1.01E-03	0.74	0.62	0.89	0.383	0.328	1.74E-03	0.79	0.69	0.92	7.72E-06	0.78	0.553
rs645078	11	63891874	C	A	*RPS6KA4*	0.354	0.291	1.26E-03	0.75	0.63	0.89	0.384	0.328	1.50E-03	0.79	0.68	0.91	7.70E-06	0.78	0.601
rs670345	11	75123917	A	G	*none*	0.059	0.074	1.31E-01	1.28	0.93	1.77	0.064	0.100	4.77E-05	1.62	1.28	2.05	1.91E-05	1.51	0.228
rs3781730	11	75585731	T	C	*WNT11*	0.156	0.219	2.07E-05	1.56	1.27	1.92	0.151	0.166	3.22E-01	1.10	0.91	1.32	2.89E-04	1.29	0.014
rs4944640	11	86305653	C	T	*PRSS23*	0.156	0.216	9.87E-05	1.52	1.23	1.87	0.186	0.166	1.64E-01	0.88	0.74	1.05	1.32E-01	1.11	0.000
rs3858359	11	91148975	G	T	*none*	0.109	0.120	3.37E-01	1.13	0.88	1.46	0.122	0.071	1.09E-05	0.56	0.43	0.72	1.91E-02	0.80	0.000
rs11226664	11	104593463	G	A	*CARD17*	0.361	0.358	9.58E-01	1.00	0.84	1.18	0.378	0.309	5.46E-05	0.74	0.64	0.86	1.70E-03	0.84	0.009
rs1945975	11	104804934	T	C	*CARD17*	0.362	0.369	6.27E-01	1.04	0.88	1.24	0.380	0.313	5.70E-05	0.74	0.64	0.86	6.26E-03	0.86	0.003
rs598373	11	118169587	T	C	*DDX6*	0.341	0.299	2.11E-02	0.81	0.68	0.97	0.344	0.283	2.42E-04	0.76	0.65	0.88	2.44E-05	0.78	0.582
rs715412	11	118189820	A	G	*SET*	0.190	0.158	4.86E-02	0.80	0.65	1.00	0.209	0.158	3.87E-04	0.72	0.60	0.86	8.04E-05	0.75	0.461
rs10842549	12	25442292	G	A	*Hs.44647*	0.384	0.475	3.45E-05	1.43	1.21	1.69	0.405	0.408	8.15E-01	1.02	0.89	1.17	6.69E-03	1.16	0.002
rs2286025	12	46646251	T	C	*TMEM106C*	0.084	0.094	2.93E-01	1.17	0.88	1.55	0.100	0.141	1.43E-04	1.47	1.20	1.79	8.62E-05	1.39	0.147
rs3850022	12	102986504	C	A	*HCFC2*	0.110	0.157	8.58E-05	1.61	1.27	2.04	0.160	0.149	5.09E-01	0.94	0.78	1.13	5.07E-02	1.16	0.001
rs652157	13	101562517	T	C	*FGF14*	0.482	0.481	6.34E-02	1.17	0.99	1.37	0.564	0.626	2.82E-04	1.29	1.49	1.13	6.38E-05	1.24	0.326
rs2211312	13	107203928	C	A	*FAM155A*	0.104	0.108	8.77E-01	1.02	0.78	1.33	0.124	0.071	8.49E-06	0.55	0.42	0.72	1.59E-03	0.74	0.001
rs6573960	14	69926775	C	T	*SYNJ2BP*	0.157	0.126	4.21E-02	0.78	0.61	0.99	0.134	0.087	6.01E-05	0.61	0.48	0.78	2.02E-05	0.69	0.150
rs8017161	14	102632948	A	G	*C14orf73/TNFAIP2*	0.378	0.464	1.53E-05	1.44	1.22	1.69	0.399	0.446	4.23E-03	1.22	1.07	1.40	4.86E-07	1.31	0.152
rs8024749	15	36173868	A	G	*none*	0.137	0.117	1.24E-01	0.82	0.64	1.06	0.107	0.152	4.40E-05	1.50	1.23	1.82	1.94E-02	1.20	0.000
rs7199670	16	22782739	A	G	*HSRST2*	0.150	0.143	7.65E-01	0.97	0.77	1.22	0.112	0.071	6.62E-05	0.59	0.45	0.76	4.69E-03	0.78	0.005
rs6564470	16	76356809	G	A	*none*	0.443	0.383	2.79E-03	0.78	0.66	0.92	0.490	0.435	4.82E-03	0.82	0.72	0.94	3.36E-05	0.80	0.643
rs8048451	16	76368220	A	G	*none*	0.359	0.290	5.09E-04	0.73	0.61	0.87	0.412	0.366	1.15E-02	0.84	0.73	0.96	3.06E-05	0.79	0.264
rs4888683	16	76373529	C	T	*none*	0.346	0.291	5.51E-03	0.78	0.65	0.93	0.402	0.348	2.94E-03	0.81	0.70	0.93	4.48E-05	0.79	0.780
rs11645428	16	79816397	A	G	*BCMO1/PKD1L2*	0.482	0.386	2.69E-06	0.67	0.57	0.80	0.353	0.342	3.44E-01	0.93	0.81	1.08	1.42E-04	0.81	0.004
rs13290	17	7270356	T	G	*C17orf74*	0.359	0.277	3.37E-05	0.69	0.57	0.82	0.356	0.362	7.87E-01	1.02	0.88	1.18	2.06E-02	0.88	0.001
rs2302764	17	7300834	C	T	*CHRNB1*	0.196	0.130	7.26E-05	0.63	0.50	0.79	0.161	0.163	9.98E-01	1.00	0.83	1.20	1.58E-02	0.84	0.002
rs6503186	17	9032986	C	T	*NTN1*	0.149	0.160	4.12E-01	1.10	0.88	1.38	0.117	0.167	4.76E-05	1.47	1.22	1.77	2.81E-04	1.30	0.052
rs907092	17	35175785	A	G	*IKZF3*	0.407	0.484	5.06E-05	1.40	1.19	1.65	0.459	0.469	1.70E-05	1.35	1.18	1.55	2.70E-09	1.37	0.744
rs9303277	17	35229995	T	C	*IKZF3*	0.451	0.532	4.01E-05	1.41	1.20	1.66	0.506	0.578	2.59E-05	1.34	1.54	1.17	1.69E-09	1.38	0.771
rs11557467	17	35282160	T	G	*ZPBP2*	0.439	0.477	2.99E-05	1.42	1.20	1.67	0.501	0.566	7.83E-05	1.32	1.51	1.15	5.84E-09	1.37	0.579
rs2872507[b]	17	35294289	A	G	*ZPBP2*	0.415	0.498	2.35E-05	1.42	1.21	1.68	0.454	0.474	1.89E-05	1.34	1.17	1.54	3.12E-09	1.37	0.627
rs8067378	17	35304874	G	A	*GSDML*	0.453	0.532	8.06E-05	1.39	1.18	1.63	0.505	0.571	7.45E-05	1.32	1.51	1.15	1.12E-08	1.36	0.728
rs2305480	17	35315722	A	G	*ORMDL3*	0.412	0.493	3.62E-05	1.41	1.20	1.66	0.449	0.482	3.55E-05	1.33	1.16	1.53	4.49E-09	1.37	0.625
rs2290400	17	35319766	C	T	*ORMDL3*	0.452	0.528	1.42E-04	1.37	1.17	1.62	0.500	0.565	1.13E-04	1.31	1.50	1.14	3.31E-08	1.34	0.748
rs7216389	17	35323475	C	T	*ORMDL3*	0.443	0.521	8.48E-05	1.39	1.18	1.63	0.504	0.563	2.85E-04	1.29	1.47	1.12	7.08E-08	1.33	0.548
rs4795405	17	35341943	T	C	*ORMDL3*	0.402	0.463	1.75E-03	1.30	1.10	1.53	0.435	0.492	5.47E-04	1.27	1.11	1.45	2.88E-06	1.28	0.847
rs1007654	17	35364880	A	G	*GSDM1*	0.395	0.431	4.85E-02	1.18	1.00	1.39	0.363	0.425	1.99E-04	1.30	1.13	1.49	3.84E-05	1.25	0.384
rs3902025	17	35372780	G	T	*GSDM1*	0.465	0.488	1.50E-02	1.22	1.04	1.44	0.426	0.481	1.78E-03	1.24	1.09	1.43	6.08E-05	1.24	0.852
rs1617406	17	41140560	G	A	*LRRC37A*	0.183	0.247	4.78E-05	1.49	1.23	1.82	0.180	0.154	4.79E-02	0.83	0.69	1.00	1.99E-01	1.09	0.000
rs4792891	17	41329294	G	T	*MAPT*	0.414	0.342	9.44E-05	0.71	0.60	0.85	0.339	0.331	5.26E-01	0.95	0.83	1.10	3.17E-03	0.85	0.010
rs7220127	17	61976384	C	T	*PRKCA*	0.484	0.401	1.66E-05	0.69	0.59	0.82	0.423	0.414	4.54E-01	0.95	0.83	1.09	7.15E-04	0.83	0.005
rs9303778	18	12978783	C	T	*SEH1L*	0.092	0.073	1.14E-01	0.78	0.58	1.06	0.068	0.103	8.09E-05	1.59	1.26	2.00	3.17E-02	1.22	0.000
rs4043679	18	19907836	T	G	*TTC39C*	0.376	0.433	1.08E-02	1.24	1.05	1.47	0.404	0.452	3.97E-03	1.22	1.07	1.40	8.20E-05	1.24	0.947
rs8085148	18	19910288	A	C	*TTC39C*	0.382	0.432	2.95E-02	1.20	1.02	1.42	0.401	0.454	1.94E-03	1.24	1.08	1.42	9.42E-05	1.23	0.694
rs7230264	18	23181432	A	G	*FLJ45994*	0.075	0.110	2.11E-03	1.54	1.17	2.03	0.108	0.139	3.78E-03	1.34	1.10	1.64	4.34E-05	1.40	0.408
rs1474133	18	23234548	G	A	*FLJ45994*	0.374	0.468	5.05E-06	1.47	1.25	1.74	0.445	0.439	9.97E-01	1.00	0.87	1.15	3.74E-03	1.17	0.000
rs4274498	18	23249914	A	G	*Hs.201828*	0.426	0.482	1.55E-05	1.44	1.22	1.69	0.491	0.482	8.18E-01	0.98	0.86	1.13	9.85E-03	1.15	0.001
rs2879447	18	23256776	C	T	*Hs.201828*	0.434	0.474	1.46E-05	1.44	1.22	1.69	0.497	0.500	6.62E-01	1.03	0.90	1.18	1.87E-03	1.18	0.002
rs4447498	18	27037686	T	C	*DSC1*	0.074	0.125	2.40E-05	1.77	1.35	2.32	0.076	0.079	7.75E-01	1.04	0.81	1.34	4.15E-03	1.31	0.003
rs2063295	18	31264940	T	C	*none*	0.422	0.362	1.22E-03	0.76	0.64	0.90	0.363	0.323	9.86E-03	0.83	0.72	0.96	6.36E-05	0.80	0.404
rs4940235	18	48807971	C	A	*DCC*	0.468	0.542	8.55E-05	1.39	1.18	1.64	0.464	0.481	3.75E-01	1.06	0.93	1.21	1.32E-03	1.19	0.015

rs4077090	18	52992869	C	T	*none*	0.231	0.240	5.35E-01	1.06	0.88	1.29	0.268	0.332	1.81E-05	1.37	1.19	1.59	1.19E-04	1.26	0.034
rs1009081	18	53073827	T	C	*none*	0.196	0.192	8.42E-01	0.98	0.80	1.20	0.227	0.289	1.74E-05	1.39	1.20	1.62	7.13E-04	1.23	0.007
rs4808419	19	15961918	G	A	*none*	0.474	0.475	9.45E-01	0.99	0.84	1.17	0.511	0.435	2.96E-05	1.34	1.17	1.54	1.24E-03	1.19	0.004
rs2279144	19	43893621	T	C	*ACTN4*	0.414	0.327	2.30E-05	0.69	0.58	0.82	0.467	0.476	4.54E-01	1.05	0.92	1.21	5.64E-02	0.90	0.000
rs8107416	19	43929790	A	C	*ACTN4*	0.417	0.328	2.31E-05	0.69	0.58	0.82	0.477	0.478	7.24E-01	1.03	0.89	1.18	2.77E-02	0.89	0.000
rs3745516	19	55618554	A	G	*SPIB*	0.272	0.357	1.41E-05	1.47	1.24	1.75	0.231	0.307	8.88E-07	1.45	1.25	1.68	7.97E-11	1.46	0.875
rs3810276	19	55626751	A	G	*SPIB*	0.463	0.480	4.01E-03	1.27	1.08	1.50	0.414	0.471	2.06E-03	1.24	1.08	1.42	1.86E-05	1.26	0.866
rs6140113	20	691770	T	C	*C20orf54*	0.174	0.167	6.74E-01	0.95	0.77	1.19	0.164	0.108	2.10E-05	0.62	0.50	0.77	7.20E-04	0.76	0.004
rs2180368	20	987828	C	T	*none*	0.303	0.379	6.53E-05	1.41	1.19	1.68	0.280	0.291	7.31E-01	1.03	0.88	1.19	5.62E-03	1.17	0.004
rs5999665	22	33715028	C	T	*Hs.589934*	0.480	0.401	2.01E-05	0.70	0.59	0.82	0.466	0.473	7.20E-01	1.03	0.90	1.17	1.54E-02	0.88	0.001
rs739363	22	38090829	A	C	*SYNGR1*	0.263	0.324	2.56E-04	1.39	1.17	1.66	0.296	0.329	2.14E-02	1.19	1.03	1.37	5.25E-05	1.26	0.156
rs2879246	22	47188783	C	T	*none*	0.143	0.201	8.61E-05	1.53	1.24	1.89	0.188	0.192	6.11E-01	1.05	0.88	1.24	4.72E-03	1.21	0.006

a. SNPs with modest (p <1E-04) or stronger evidence of association in the Italian PBC, and/or Canadian PBC and/or the meta-analysis are presented. Allele 1 is the minor allele for the positive strand in the Italian population.

b. For these SNPs the combined data that included Canadian PBC and control data were based partially on imputation.

Supplementary Table 3 Analysis of *IL12A/SCHIP1* SNPs

Stepwise Logistic Regression[a]

SNP	BP	p value
rs6441284	161200962	5.0E-10
rs17811014	161170723	1.5E-05
rs583911	161193084	1.2E-05
rs1075498	161239145	5.9E-03
rs924247	160963022	2.9E-02

Haplotype Association Tests[b]

Haplotypes	OR	p value
GGAAA	0.58	3.5E-07
AGGCG	1.34	9.8E-05
AAACG	1.88	4.5E-08
GGAAG	0.58	8.4E-07
GGACA	0.311	1.7E-02
AGAAA	0.67	2.2E-02
GGGCG	1.19	4.8E-02
AGGAG	1.35	1.3E-02
AGAAG	0.72	1.1E-02

a. Results of stepwise logistic regression using combined PBC dataset. Conditionally independent results were observed with each of the listed SNPs. These analyses were performed using the combined Italian and Canadian datasets. For these SNPs there was no heterogeneity observed in the separate analyses [Cochran's Q test p values >0.1 (**Supplementary Table 2**)]

b. Haplotypes showing nominally significant results are shown for the SNPs identified in the setwise logistic regression. The first 4 haplotypes were strongly associated with PBC risk. The order of the SNPs is rs924247, rs17811014, rs583911, rs6441286, and rs107548.

Supplementary Table 4. Comparison of Meta-Analyses containing all PBC Cases and AMA only Cases[a]

SNP	CHR	BP	Minor Allele	META p	META OR	AMA p	AMA OR
rs4908830	1	9220458	G	7.61E-04	0.8063	8.71E-05	0.7492
rs3790565	1	67583944	C	4.46E-09	1.4358	1.00E-08	1.4796
rs3790567	1	67594965	A	7.53E-12	1.4753	2.96E-10	1.4921
rs4297265	1	67624923	G	5.42E-05	1.2404	8.52E-04	1.2218
rs2270614	1	67628609	A	3.44E-05	1.2471	5.68E-04	1.2298
rs7546194	1	81643352	A	4.31E-03	0.8579	5.55E-05	0.7868
rs4636400	1	144155913	G	8.08E-05	0.8007	1.01E-04	0.7842
rs1105749	2	4272264	G	4.09E-04	1.2553	9.74E-05	1.3234
rs6741449	2	70010093	G	1.34E-03	1.189	7.04E-05	1.2722
rs7569076	2	88554147	T	1.41E-05	1.2965	4.72E-03	1.209
rs3769811	2	182745334	T	8.75E-05	0.7469	3.25E-04	0.7379
rs7601754	2	191648696	G	1.34E-05	0.731	7.11E-05	0.7271
rs6752770	2	191681808	G	6.15E-05	1.2572	1.50E-04	1.2697
rs6437268	2	240649535	C	2.09E-05	0.7828	1.44E-05	0.7545
rs2887401	2	240685626	A	9.63E-05	0.78	1.04E-03	0.7923
rs4686174	3	7894746	T	9.96E-05	1.2548	8.36E-05	1.2882
rs6797331	3	7915671	T	4.62E-05	1.2671	4.46E-05	1.2988
rs1372072	3	16930263	A	1.52E-05	1.2671	1.19E-02	1.1664
rs9876137	3	16936269	G	1.42E-05	1.2681	1.14E-02	1.1675
rs1025818	3	16940025	C	9.49E-06	1.2753	1.63E-03	1.2142
rs4618210	3	17099388	G	3.78E-05	1.2463	1.15E-02	1.1615
rs1394761	3	24390831	A	7.56E-05	0.7927	4.23E-05	0.7584
rs1304138	3	39979370	A	1.38E-03	1.1885	5.67E-05	1.2768
rs1585287	3	39989394	A	3.58E-03	1.1683	4.64E-05	1.2767
rs13078855	3	120589967	T	9.50E-06	0.7201	2.20E-04	0.7349
rs1132200	3	120633526	T	3.87E-07	0.6723	4.36E-06	0.6663
rs2293370	3	120702624	A	3.03E-07	0.6847	6.82E-06	0.6878
rs6804441	3	120743634	G	4.48E-05	0.7365	3.91E-04	0.7418
rs4680514	3	160944150	A	7.94E-05	0.7599	8.53E-04	0.7725
rs1461100	3	160945597	A	7.28E-05	0.755	1.24E-03	0.7756
rs3863075	3	160985457	C	3.05E-06	0.7562	3.06E-07	0.7069
rs7636819	3	160988297	G	4.31E-06	0.7596	4.54E-07	0.711
rs6441276	3	161034716	T	1.16E-05	0.7386	6.72E-06	0.7027
rs33055	3	161086908	C	1.18E-04	1.2394	6.72E-05	1.2802
rs26298	3	161093985	G	1.65E-06	0.764	3.59E-07	0.7242
rs17811014	3	161170723	T	2.32E-05	1.3272	9.59E-04	1.2804
rs2243123	3	161192345	C	4.57E-07	0.7219	1.51E-06	0.7013
rs583911	3	161193084	G	1.41E-07	1.3242	1.67E-07	1.3719
rs6441284	3	161200962	G	2.91E-07	0.7482	1.17E-05	0.7555

rs485497	3	161201826	G	3.15E-06	0.7778	6.87E-06	0.7602
rs6441286	3	161211572	G	7.63E-12	1.4415	2.10E-08	1.395
rs574808	3	161215677	C	3.30E-10	0.698	9.10E-09	0.6877
rs10513549	3	161237201	A	5.93E-05	0.772	4.56E-04	0.774
rs1075498	3	161239145	A	6.54E-08	0.7113	2.78E-06	0.7157
rs4679904	3	161823590	T	5.31E-05	0.7688	2.88E-03	0.8071
rs7678820	4	22901080	C	1.33E-05	0.7138	6.55E-05	0.7068
rs1912031	4	22938137	A	8.31E-05	0.7449	2.27E-03	0.7786
rs7665090	4	103770651	A	3.47E-05	0.8012	3.19E-05	0.7793
rs2866413	4	103776125	G	4.11E-05	0.8027	3.95E-05	0.7816
rs228611	4	103780757	A	5.44E-05	0.805	7.69E-05	0.788
rs228614	4	103797685	A	2.19E-05	0.7965	4.20E-05	0.7806
rs223420	4	103947120	A	7.66E-05	1.2346	2.25E-03	1.2007
rs4698874	4	104149960	C	5.83E-05	1.2387	1.14E-03	1.2145
rs10516496	4	104161614	T	8.98E-05	1.2405	2.92E-03	1.1987
rs11946020	4	104164191	C	8.47E-05	1.2411	2.22E-03	1.2042
rs1358229	4	123116184	T	6.24E-06	1.2724	1.25E-05	1.2959
rs6838639	4	123118615	A	6.63E-06	0.7443	4.45E-04	0.7742
rs13144383	4	139310254	G	6.21E-05	1.317	1.20E-06	1.4519
rs954017	5	40066770	A	6.85E-04	1.3605	4.77E-05	1.4956
rs2395488	6	31553888	G	9.35E-05	1.2391	1.97E-03	1.2065
rs2523647	6	31557757	G	3.72E-05	1.2815	4.83E-03	1.2072
rs2269475	6	31691910	T	5.56E-07	1.4898	1.70E-05	1.4808
rs1046089	6	31710946	A	3.13E-06	1.2989	4.34E-06	1.3339
rs805303	6	31724345	A	4.81E-05	1.2505	3.81E-05	1.2862
rs3130617	6	31735502	C	2.76E-05	0.7508	1.06E-03	0.7767
rs2280800	6	31754377	A	2.52E-07	1.5002	1.25E-05	1.4857
rs805274	6	31773173	C	4.03E-06	1.3066	3.37E-05	1.303
rs2242653	6	31783744	A	6.41E-08	1.4873	1.67E-06	1.4966
rs805294	6	31796196	G	1.37E-04	1.2326	3.95E-05	1.2838
rs2227956	6	31886251	G	2.47E-05	0.6981	4.21E-05	0.6636
rs2763979	6	31902571	T	6.86E-06	1.2812	2.36E-05	1.296
rs9267649	6	31932807	A	9.33E-06	0.6862	1.21E-05	0.645
rs3134954	6	32179871	C	1.06E-05	0.6525	1.25E-04	0.6491
rs2269425	6	32231617	A	4.06E-07	1.4708	1.54E-05	1.4448
rs3134603	6	32233980	A	3.30E-06	0.6396	4.49E-05	0.6352
rs3134943	6	32255739	T	1.70E-07	0.5912	1.52E-06	0.5642
rs3132946	6	32298006	A	1.33E-06	0.6045	1.33E-05	0.5842
rs411326	6	32319295	T	6.96E-06	1.3046	2.66E-04	1.2747
rs9267992	6	32328375	G	2.36E-05	0.6588	1.89E-04	0.6491
rs2076537	6	32425613	A	6.37E-06	1.2817	2.53E-05	1.2982
rs2395148	6	32429532	T	9.95E-15	2.7547	1.54E-11	2.7113
rs6907322	6	32432923	A	1.27E-12	1.5523	1.01E-09	1.541
rs2395150	6	32434023	G	1.23E-05	1.264	6.10E-05	1.2775

rs2073048	6	32443411	A	1.74E-08	1.5049	3.59E-06	1.4531
rs4424066	6	32462406	G	1.71E-05	1.2616	1.11E-04	1.2615
rs3117098	6	32466491	G	8.88E-08	0.7287	2.51E-08	0.6897
rs4248166	6	32474399	C	3.47E-09	1.4937	1.31E-07	1.4951
rs3763305	6	32477466	A	9.11E-06	1.6844	1.12E-05	1.7923
rs3806156	6	32481676	T	1.74E-10	1.4153	8.29E-09	1.4215
rs3763313	6	32484449	C	4.24E-09	1.4555	4.75E-07	1.4322
rs2395163	6	32495787	C	2.99E-06	1.3556	2.15E-05	1.3712
rs3135363	6	32497626	G	2.00E-13	0.6351	1.34E-12	0.6103
rs9501626	6	32508322	A	4.72E-09	1.5582	2.73E-08	1.6001
rs2395175	6	32513004	A	2.11E-05	1.395	9.26E-04	1.3559
rs6903608	6	32536263	C	4.96E-07	0.7467	7.22E-08	0.7045
rs660895	6	32685358	G	1.14E-16	1.7273	1.13E-13	1.7497
rs3129763	6	32698903	A	2.16E-08	0.7014	1.17E-08	0.6671
rs9275184	6	32762692	C	1.40E-06	1.5164	4.93E-05	1.4815
rs7775228	6	32766057	C	9.74E-11	1.564	8.31E-10	1.5857
rs9357152	6	32772938	G	4.24E-11	0.6625	1.78E-12	0.6074
rs9275312	6	32773706	G	2.14E-20	1.9407	1.17E-16	1.9482
rs9275390	6	32777134	C	3.31E-21	1.744	2.49E-17	1.7463
rs2647050	6	32777745	C	2.58E-08	0.7333	3.32E-09	0.6896
rs2856718	6	32778233	T	1.53E-08	0.7291	1.83E-09	0.6844
rs7745656	6	32788948	T	4.01E-05	0.7893	1.37E-06	0.7308
rs9276644	6	32853021	C	6.52E-05	0.7976	2.01E-04	0.7908
rs2219893	6	32877641	C	7.41E-06	1.2813	1.33E-04	1.2696
rs3129299	6	33008765	T	6.56E-05	1.2966	3.31E-05	1.3504
rs2395296	6	33019792	A	9.78E-05	0.7905	2.03E-03	0.8134
rs194682	6	33022591	T	6.11E-05	1.2454	5.53E-03	1.1878
rs209473	6	33030886	A	3.77E-05	0.8005	4.41E-04	0.8074
rs209474	6	33032562	G	4.58E-05	1.2454	1.16E-03	1.2177
rs423639	6	33095752	T	1.70E-04	1.3843	2.39E-05	1.4851
rs9277535	6	33162839	G	1.95E-12	1.5046	4.90E-12	1.5773
rs9277554	6	33163516	T	6.10E-09	1.3877	2.71E-09	1.4604
rs9277565	6	33164875	T	2.62E-11	1.5025	6.17E-11	1.5725
rs2281389	6	33167774	G	6.44E-12	1.5514	7.93E-11	1.5988
rs3128917	6	33167974	G	5.28E-08	1.3753	2.37E-08	1.4454
rs3117222	6	33168927	T	5.73E-08	1.3741	2.54E-08	1.4443
rs2064478	6	33180244	T	5.08E-08	1.3898	5.44E-08	1.446
rs3130215	6	33182941	A	1.00E-04	0.8027	8.96E-05	0.7788
rs3117230	6	33183613	G	6.52E-08	1.387	7.01E-08	1.4419
rs3117016	6	33203494	A	1.64E-05	1.2598	2.16E-04	1.2475
rs6901221	6	33206254	C	2.15E-09	1.5112	2.33E-07	1.5027
rs2855430	6	33249258	A	9.65E-09	1.5253	4.51E-07	1.5321
rs2855459	6	33262634	A	1.02E-08	1.5268	4.53E-07	1.5344
rs439205	6	33281820	A	3.10E-07	1.353	5.28E-06	1.3554

rs421446	6	33282761	G	9.06E-08	1.3543	4.43E-06	1.3393
rs9371462	6	150910792	G	1.44E-04	1.2517	7.41E-05	1.2946
rs10250643	7	24502175	G	1.25E-05	0.7523	6.14E-05	0.7451
rs2255365	7	24528186	C	9.23E-05	0.8098	1.56E-03	0.8273
rs917017	7	43331539	G	1.65E-04	1.2228	9.79E-05	1.2615
rs4728142	7	128361203	A	5.97E-05	1.2386	2.03E-03	1.2015
rs10488631	7	128381419	C	2.78E-10	1.634	3.51E-07	1.5606
rs11784810	8	24482929	G	7.16E-05	1.2387	9.05E-03	1.1701
rs12678383	8	24498107	T	7.50E-05	1.2625	1.51E-03	1.2317
rs7813101	8	97733748	A	2.43E-05	1.2744	2.38E-04	1.2706
rs1529335	8	129085529	T	2.12E-05	0.7004	1.06E-03	0.7333
rs895236	9	4028228	T	5.83E-05	0.6932	4.44E-03	0.7547
rs1529952	9	85909367	T	1.64E-05	1.2684	3.60E-05	1.2915
rs3124607	9	138534660	A	7.93E-05	0.7901	1.02E-03	0.8009
rs10993994	10	51219502	T	2.92E-05	1.2513	9.50E-04	1.22
rs11596067	10	62688994	T	1.38E-03	1.268	4.15E-05	1.3974
rs7084300	10	62703010	C	1.48E-03	1.2658	4.20E-05	1.3949
rs12781461	10	82915023	T	9.41E-06	1.2778	2.20E-04	1.2598
rs11190842	10	102885447	A	2.67E-05	0.6769	1.05E-03	0.7134
rs7911360	10	122795537	A	1.08E-04	0.8034	7.66E-05	0.7796
rs1562902	11	14874792	C	9.72E-03	0.868	6.69E-05	0.7797
rs541207	11	63881718	G	5.40E-06	0.7783	3.18E-04	0.7993
rs538147	11	63886298	A	7.72E-06	0.7753	2.41E-04	0.7902
rs645078	11	63891874	C	7.70E-06	0.775	2.58E-04	0.7907
rs670345	11	75123917	A	1.91E-05	1.5088	2.92E-03	1.3824
rs598373	11	118169587	T	2.44E-05	0.7828	2.25E-04	0.7887
rs715412	11	118189820	A	8.04E-05	0.7541	8.78E-04	0.7663
rs2286025	12	46646251	T	8.62E-05	1.3881	1.98E-03	1.3347
rs3184504	12	110368991	C	1.53E-04	0.816	1.15E-05	0.7695
rs653178	12	110492139	T	1.68E-04	0.8175	9.46E-06	0.7677
rs11066320	12	111390798	A	1.19E-03	1.1889	8.83E-05	1.2625
rs3783316	14	34406943	A	1.46E-04	1.2314	7.80E-05	1.2717
rs6573960	14	69926775	C	2.02E-05	0.6896	6.13E-04	0.7258
rs8017161	14	102632948	A	4.86E-07	1.3101	3.97E-06	1.3225
rs1097863	16	10234830	G	2.65E-03	1.2306	2.90E-05	1.3673
rs1035089	16	10955851	A	5.13E-03	1.1612	8.13E-05	1.2632
rs8059260	16	10967652	G	5.07E-03	0.8086	3.12E-05	0.6907
rs2302558	16	10970512	T	8.34E-04	0.7591	9.71E-06	0.6528
rs8055893	16	10979296	T	7.92E-04	0.7553	2.26E-05	0.6601
rs9940096	16	11047095	G	5.20E-04	0.7462	2.87E-05	0.6624
rs741176	16	11053311	G	3.64E-04	0.7401	2.77E-05	0.6632
rs1345879	16	11312263	T	1.08E-03	1.2055	2.82E-05	1.3042
rs7186179	16	56265333	A	1.52E-03	1.2636	7.92E-05	1.3776
rs6564470	16	76356809	G	3.36E-05	0.7995	1.38E-03	0.8264

rs8048451	16	76368220	A	3.06E-05	0.7917	7.13E-04	0.8106
rs4888683	16	76373529	C	4.48E-05	0.7944	1.35E-03	0.8186
rs11645428	16	79816397	A	1.42E-04	0.8105	9.10E-05	0.7821
rs907092	17	35175785	A	2.70E-09	1.3737	4.08E-08	1.391
rs2305480	17	35315722	A	4.49E-09	1.3673	1.02E-07	1.3771
rs4795405	17	35341943	T	2.88E-06	1.2839	1.40E-05	1.3004
rs1007654	17	35364880	A	3.84E-05	1.2483	1.75E-04	1.255
rs3902025	17	35372780	G	6.08E-05	1.2377	6.48E-04	1.2272
rs10515129	17	51932026	G	9.17E-04	1.3239	2.65E-05	1.4786
rs4043679	18	19907836	T	8.20E-05	1.2356	2.00E-05	1.2921
rs8085148	18	19910288	A	9.42E-05	1.2334	3.56E-05	1.2752
rs1487435	18	19911764	T	1.07E-04	1.2302	6.82E-05	1.2684
rs7230264	18	23181432	A	4.34E-05	1.3997	6.67E-04	1.377
rs2063295	18	31264940	T	6.36E-05	0.7995	1.82E-03	0.8231
rs4077090	18	52992869	C	1.19E-04	1.2551	6.76E-05	1.3015
rs3745516	19	55618554	A	7.97E-11	1.4562	2.87E-08	1.4311
rs3810276	19	55626751	A	1.86E-05	1.2564	2.79E-04	1.2397
rs739363	22	38090829	A	5.25E-05	1.2605	1.33E-03	1.2253

a Results from the combined meta-analysis of ITN and Canadian datasets for all (959) or only AMA positive (701) PBC cases. All p values (<10-4) for either analyses are shown.

Supplementary Table 5. Comparison of PBC and Celiac Association for *SCHIP1/IL12A* Region

			PBC[a]		**Celiac UKGWAS[b]**		**Celiac CMH Overall**		**Celiac Italian Study[c]**	
		minor allele	P value	OR[d]	P value	OR	P value	OR	P value	OR
rs10513546	161034345	A	1.04E-03	1.20	1.20E-03	0.80	7.49E-02	0.93	NA	NA
rs6441276	161034716	T	1.16E-05	0.74	NA	NA	NA	NA	NA	NA
rs13088856	161037652	G	1.18E-03	1.20	1.13E-03	0.79	7.27E-02	0.93	NA	NA
rs17810546	161147744	G	2.76E-02	0.90	3.39E-04	1.38	1.07E-09	1.35	4.00E-04	1.71
rs9811792	161179692	C	3.30E-02	0.89	2.78E-03	1.21	5.24E-08	1.21	3.13E-02	1.21
rs583911	161193084	G	1.41E-07	1.32	NA	NA	NA	NA	NA	NA
rs6441286	161211572	G	7.63E-12	1.44	NA	NA	NA	NA	NA	NA

a. Results from analyses in the current study.

b. Results from Hunt, K.A. et al. Nat Genet 40, 395-402 (2008).

c. Results from Romanos, J. et al. J Med Genet 46, 60-3 (2009).

d. OR provided for minor allele in forward direction for all studies.

Supplementary Methods for Meta-analyses of Italian and Canadian Genome-Wide Association Screens Identify Multiple Loci Associated with Primary Biliary Cirrhosis

Subjects.

All PBC cases included in these studies met internationally accepted and American Association for the Study of Liver Diseases criteria for PBC[1], including a cholestatic pattern of serum biochemistry for >6 months and liver histology compatible with or diagnostic of PBC. Most individuals (see below) were positive for serum anti-mitochondrial antibodies (AMA) (titers of $\geq$1:40 by indirect immunofluorescence), but AMA positivity was not used as a criterion for case inclusion in view of previous data suggesting no effect of AMA status on the profile of disease-associated loci[2].

All cases and controls were obtained using IRB approved protocols at each institution or were obtained from genotype databases with appropriate approval. PBC Italian cases were collected from multiple centers in Italy including Milano, Bologna, Padova, Torino, Napoli, Pisa, Udine, Cuneo, Roma, Firenze, Genova, Bari, and Palermo. Of a total of 524 cases, 453 cases were included in the association testing after quality control (95% complete genotyping data), cryptic relationship analyses (PI^ < 0.1), and ancestry analysis (**Supplementary Table 1**). The cases were genotyped using the Illumina 610K platform at the Feinstein Institute. The Italian PBC cases were 90.3% female, 85.4% AMA positive, mean age of 55.2 years, 39.6 % were ANA positive and 31.7% had liver cirrhosis. The Canadian PBC cases were as previously described[2] and included 506 cases of European ancestry (see Ancestry).

The control genotypes were derived from multiple sources after quality control and selection for by ancestry criteria as detailed in **Supplementary Table 1**. For the Italian (phase 1 of this study) these include: 1) 637 normal controls collected in Italy genotyped using Illumina 1Mb platform at the University of Milan; 2) 6 samples from the British Blood Bank 1958 cohort genotyped at the Sanger Insitute using Illumina 550K platform; 3) 159 subjects from the New York Cancer Project (NYCP)[3] samples genotyped at the Feinstein Institute using the Illumina 550K platform; 4) 129 subjects genotyped using the Illumina 550K platform at Children's Hospital of Philadelphia (CHOP) and deposited in I-ControlDB (www.illumina.com/iControlDB, Illumina, San Diego, CA); and 5) 14 subjects from the NIH Laboratory of Neurogenetics (NIHLN) (http://neurogenetics.nia.nih.gov/paperdata/public/). For the Canadian (phase 2 of this study) these include: 1) 1118 M.D. Anderson historical controls genotyped using the 300K Illumina platform at the Center for Inherited Disease Research (CIDR, Johns Hopkins University); 2) 2210 subjects from the Cancer Genetics Markers of Susceptibility (CGEMS) genotyped using the 550K Illumina platform (http://cgems.cancer.gov/resource_room/dbgap.asp); and 3) 378 Canadian subjects genotyped using the 370K Illumina platform at the University of Toronto.

Data Filtering.

We used stringent quality control criteria to ensure that high-quality data were included in the final analysis. Specifically, we excluded individuals who had >5% missing data and all individuals with cryptic relatedness and duplicate samples based on identity-by-state status (PI_Hat > 0.10) using PLINK[4]. The SNP data was carefully reviewed and exclusion criteria applied to minimize potential batch effects as described below. The application of these criteria was particularly important given the derivation of genotypes from multiple platforms typed in different laboratories.

We included only SNPs with <5% missing data, Hardy-Weinberg (H-W) equilibrium p values > 1 times 10^{-4} in controls and >1 times 10^{-5} in combined cases and controls, minor allele frequency > 0.05%. These procedures were applied in a stepwise approach separately for the Italian and Canadian data sets. Thus, for each of the separately derived control genotyping sets (**Supplementary Table 1**), SNPs were excluded if they failed the above criteria within the individual control set (platform and laboratory) or in combination with any of the other control groups, or in the complete data set. The stringent H-W criteria were applied after exclusion of non-European individuals (see Ancestry). As another quality check, ten of the Italian controls genotyped on 1Mb platform at the University of Milan were also genotyped on the Illumina 610K platform at the Feinstein Institute together with the Italian PBC cases. The concordance of the genotypes from two different Illumina platforms genotyped at two different institutions was > 0.9998 and SNPs not fully concordant were excluded. Finally, after selection of the Italian control data set (see Ancestry), SNPs were excluded if allele frequency differences between different control subject groups was > 10%. For the Italian and Canadian study sets, 484,351 and 295,355 SNPs, respectively, passed these quality control steps. A total of 276,459 SNPs were common to both study groups and additional non-overlapping SNPs were imputed as described below (see **Imputation**).

Ancestry.

European ancestry was determined using a panel of 187 ancestry informative markers and analyzed using the STRUCTURE v2.1[5,6] program and subjects of known European, Amerindian, East Asian and West African origin as previously described[7]. We used STRUCTURE to exclude non-European and admixed study participants since the application of this Bayesian clustering method allows exclusion/inclusion criteria to be set using reference populations. Unlike principal components analysis (PCA), the clustering algorithm can be run under conditions that are not affected by the inclusion of the unknown samples. Briefly, analyses were performed using > 100,000 resamplings and > 50,000 burn-in cycles under the admixture model. For all analyses reported, we used the “infer α” option with a separate α is estimated for each population (where α is the Dirichlet parameter for degree of admixture). Runs were performed under the $\lambda = 1$ option where λ estimates the prior probability of the allele frequency and is based on the Dirichlet distribution of allele frequencies. Subjects with >15% non-European ancestry were excluded from further analysis.

Homogeneous Italian ancestry was defined by applying principal components analyses (PCA) performed using the EIGENSTRAT statistical package[8]. The analyses were performed using genotypes from a diverse set of subjects of European origin after the STRUCTURE analysis described above. These included 492 European PBC cases collected in Italy, the 857 European normal controls collected by the Milan University group, the 1280 European NYCP subjects, the 1472 subjects from the CHOP study, 255 subjects from the (NIHLN), the1317 subjects from the British Blood Bank 1958 cohort, 141 Sardinian subjects genotyped at University of Milan, 28 Sardinians and 13 Northern Italian and 7 Tuscan subjects from the Human Genome Diversity Panel obtained from I-ControlDB, 77 Tuscan subjects from HapMap phase III (http://hapmap.ncbi.nlm.nih.gov/) and 91 participants in the POPRES[9] study with 4 grandparent data indicated Italian origin were obtained from dbGaP (http://www.ncbi.nlm.nih.gov/dbgap/). We utilized a set of 34,000 genotypes common to multiple Illumina and Affymetrix platforms (since POPRES samples were genotyped using Affymetrix platform) that were selected to exclude regions of high linkage disequilibrium and exclude any paired SNPs with r^2 values > 0.2. We defined homogenous Italian subgroup using the definition that each subject was within 2 standard deviations of the mean of each of the first four principal components of a reference Italian subject group. We utilized the POPRES and HGDP Italian subjects as the reference group since these subjects were not included in our association testing. The results of the PCAs show the relationship of the homogeneous Italian population group among the diverse European subjects (**Supplementary Fig 1**).

Analytic Strategy and Association Testing Statistics.

The data for this study was analyzed separately in the two different population groups: 1) the homogeneous Italian study set and 2) the expanded Canadian study set (**Supplementary Table 1**). The Canadian study set was used as an in silico replication set for putative SNPs identified in the Italian discovery group. In addition, the results from the two independently analyzed study sets were combined together in a meta-analysis applying Cochran–Mantel–Haenszel (CMH) adjustments using PLINK[4,10] which allows allele and disease frequencies to vary among study populations. For the discovery set we pre-set an α criteria of $p<10^{-5}$ (corrected for population substructure and residual genome-wide inflation) as suggestive for association. For the replication (Canadian set) a Bonferroni correction was applied to ascertain the nominal p value corresponding to a false positive rate of $p < 0.05$. Since a total of 57 SNPs met the discovery criterion, the p value of 8×10^{-4} (0.05/57) was used as the replication criterion. In addition, for the meta-analysis we applied the genome-wide criterion of $p < 5 \times 10^{-8}$ for indicating significant association. We also checked for significant evidence of variation in the odds ratio associating disease alleles with PBC risk by applying Cochran's Q test of homogeneity[10,11].

For both data sets association methods controlling for population substructure were applied. PCA was performed using EIGENSTRAT software[8] after exclusion of chromosomal regions with high linkage disequilibrium as previously discussed[12]. The population substructure was defined using the first three PCs with no further reduction of the residual genome-wide inflation factor. The homogeneous Italian subset showed a small genome-wide χ^2 inflation both before (λ_{gc} = 1.035) and after eigenvalue correction

($\lambda_{gc} = 1.033$). For the Canadian data, the initial χ^2 inflation ($\lambda_{gc} = 1.85$) decreased to 1.050 after eigenvalue correction. The PCA results were used as covariates for logistic regression and the results were corrected for the small residual genome-wide χ^2 inflations. The analysis was performed using the PLINK software that also facilitated the combining results for the meta-analysis. The meta-analysis was performed using CMH adjustments for potential differences between the two study sets.

Imputation.

Before imputation, we applied stringent quality control checks on each of the data sets. The imputations were performed separately for those cases and controls genotyped on platforms with 550K or more genotypes and those genotyped with 370K or less genotypes. We next used IMPUTE version 2[13] to infer genotypes using HapMap Phase III CEU and TSI samples as a reference. [Prior testing of multiple genomic regions indicated that combining CEU with TSI haplotypes enabled more accurate imputation than either set alone (MFS, unpublished data)]. For inclusion of data we utilized only imputed genotypes with mean uncertainty scores of < 0.05 where the uncertainty is defined as the mean probability of assignment of each genotype (where if individual genotype probability < 0.5 = uncertainty and if probability > 0.5, uncertainty = 1-probability). Using this parameter our testing indicated that the maximum error rate for genotype assignment was < 0.05 and the mean error rate was < 0.01.

SNPTEST was used as the primary association test for the imputed genotypes for SNPs. The SNPTEST software uses the genotype probabilities for the imputed SNPs. For SNPTEST, the first three PC eigenvalue scores were used as continuous variables in the association test.

URLs.

CGEMS, http://cgems.cancer.gov/;

PLINK, http://pngu.mgh.harvard.edu/~purcell/plink/;

http://genepath.med.harvard.edu/~reich/EIGENSTRAT.htm;

SNPTEST, http://www.stats.ox.ac.uk/~marchini/software/gwas/snptest.html;

IMPUTE, https://mathgen.stats.ox.ac.uk/impute/impute.html;

HAPMAP, http://hapmap.ncbi.nlm.nih.gov/;

dbGaP, http://www.ncbi.nlm.nih.gov/sites/entrez?Db=gap

References

1. Lindor, K.D. et al. Primary biliary cirrhosis. *Hepatology* **50**, 291-308 (2009).
2. Hirschfield, G.M. et al. Primary biliary cirrhosis associated with HLA, IL12A, and IL12RB2 variants. *N Engl J Med* **360**, 2544-55 (2009).
3. Mitchell, M.K., Gregersen, P.K., Johnson, S., Parsons, R. & Vlahov, D. The New York Cancer project: rationale, organization, design, and baseline characteristics. *J Urban Health.* **81**, 301-310 (2004).
4. Purcell, S. et al. PLINK: a tool set for whole-genome association and population-based linkage analyses. *Am J Hum Genet* **81**, 559-75 (2007).
5. Pritchard, J.K., Stephens, M. & Donnelly, P. Inference of population structure using multilocus genotype data. *Genetics* **155**, 945-59 (2000).
6. Falush, D., Stephens, M. & Pritchard, J.K. Inference of population structure using multilocus genotype data: linked loci and correlated allele frequencies. *Genetics* **164**, 1567-87 (2003).
7. Kosoy, R. et al. Ancestry informative marker sets for determining continental origin and admixture proportions in common populations in America. *Hum Mutat* **30**, 69-78 (2009).
8. Price, A.L. et al. Principal components analysis corrects for stratification in genome-wide association studies. *Nat Genet* **38**, 904-9 (2006).
9. Nelson, M.R. et al. The Population Reference Sample, POPRES: a resource for population, disease, and pharmacological genetics research. *Am J Hum Genet* **83**, 347-58 (2008).
10. Saxena, R. et al. Genome-wide association analysis identifies loci for type 2 diabetes and triglyceride levels. *Science* **316**, 1331-6 (2007).
11. Cochran, W.G. The comparison of percentages in matched samples. *Biometrika* **37**, 256-66 (1950).

12. Tian, C., Gregersen, P.K. & Seldin, M.F. Accounting for ancestry: population substructure and genome-wide association studies. *Hum Mol Genet* **17**, R143-50 (2008).
13. Howie, B.N., Donnelly, P. & Marchini, J. A flexible and accurate genotype imputation method for the next generation of genome-wide association studies. *PLoS Genet* **5**, e1000529 (2009).