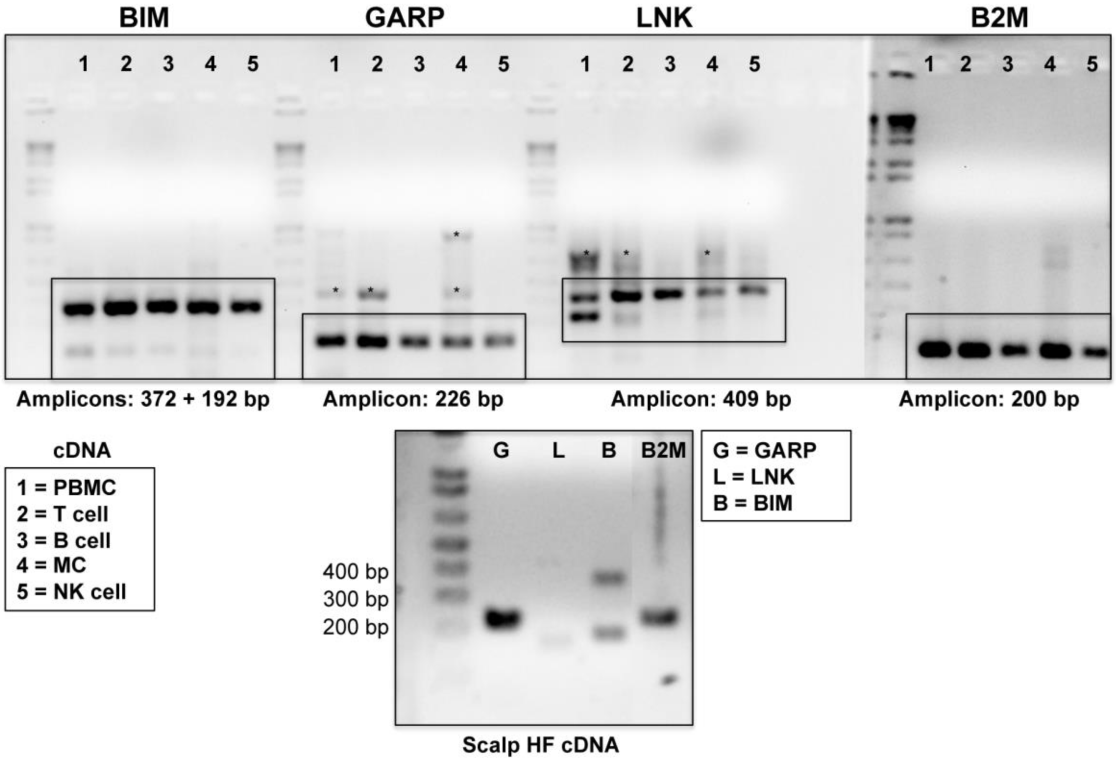
Supplementary Figures



Supplementary Figure 1. Full blot results of rt-PCR. The original gel images that were used to create Figure 4A are presented.

Supplementary Tables

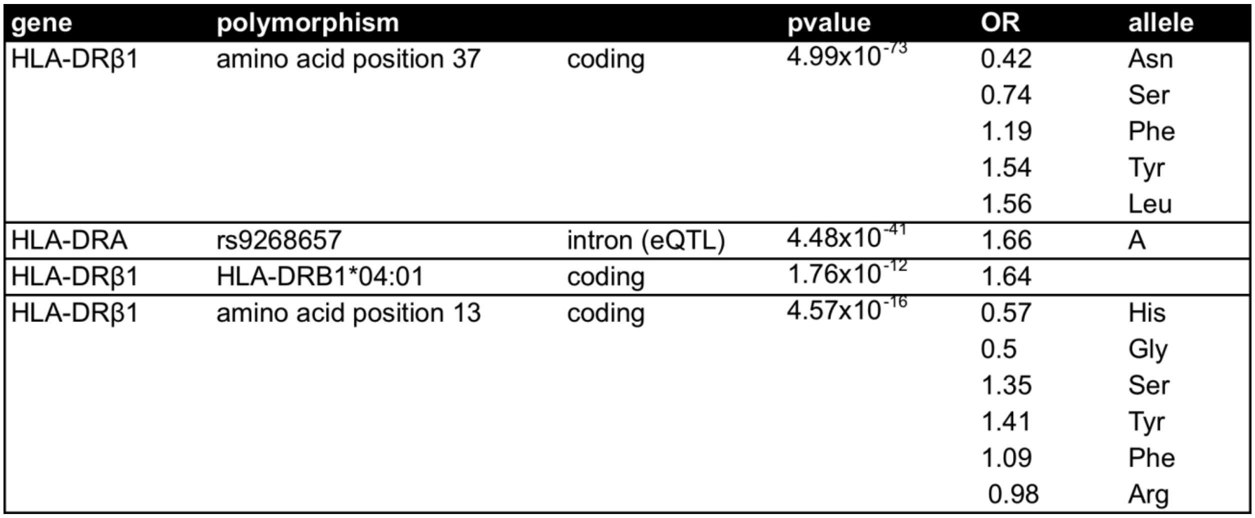
Supplementary Table 1. Study samples. Study samples for the meta-analysis and replication studies were ascertained in the United States (US) and central Europe. All US cases were ascertained through the National Alopecia Areata Registry (NAAR). The US cases included in the meta-analysis are the same cohort previously reported in our initial Genome-wide Association Study (GWAS).[1] The US controls used in the meta-analysis are the same cohorts used in our initial GWAS,[1] and were ascertained through the New York Canter Project[2] or the Cancer Genetic Markers of Susceptibility (CGEMS) breast[3] and prostate[4] cancer studies. The US cases used in the replication study are an independent sample of NAAR participants who self-report European ancestry. US controls for the replication study were either ascertained through NAAR (Sequenom genotyping) or were obtained from the NIDDK inflammatory bowel disease (IBD) genetics consortium (http://medicine.yale.edu/intmed/ibdgc/index.aspx) (ImmunoChip genotyping). The cohorts ascertained in central Europe had not been used in any previously published GWAS. Cases and controls were ascertained from outpatient clinics, private dermatology practices and *via* AA self-support groups in Belgium and Germany. Inclusion criteria followed published guidelines,[5] and additionally included diagnosis by a trained and experienced clinician. European GWAS controls included population-based controls established within the National Genome Research Network for use as universal controls (PopGen[6], KORA[7]), and the Heinz Nixdorf Recall (HNR) Study[8] and an additional population-based cohort from the HNR[8] study.

Analysis	Disease Status	Cohort (Ascertainment Source)	Genotyping Platform	Publication	Sample Count	Results	
meta analysis and HLA imputation	case	US1 (NAAR)	Illumina 610	PMID: 20596022	244	Table S2	Table S5
		US2 (NAAR)	Illumina 610	PMID: 20596022	808		
		Central Europe1	Illumina 660	current publication	629		
		Central Europe 2	Illumina Omni	current publication	651		
					2,332		
	control	US1 (NYCP)	Illumina 550	PMID: 20596022	1,034		
		US2 (CGEMS)	Illumina 550	PMID: 20596022	2,221		
		Central Europe1 (PopGen, KORA, HNR)	Illumina Omni	current publication	1,282		
		Central Europe2 (PopGen, KORA, HNR)	Illumina 550	current publication	696		
					5,233		
replication	case	US3 (NAAR)	Sequenom Immunochip	current publication	318		
	case	Central Europe 3	Sequenom	current publication	446		
					764		
	control	US3a (NAAR)	Sequenom	current publication	154		
	control	US3b (NIDDK IBDGC)	Immunochip	current publication	1,688		
	control	Central Europe 3	Sequenom	current publication	414		
					2,256		

Total Case Count 3,096

Total Control Count 7,489

Supplementary Table 2. Summary of the four independent effects found to underlie the HLA association with AA. Data is extracted from Supplementary Data 2. Odds ratios (OR) estimate the magnitude of effect in cases relative to controls.



gene	polymorphism		pvalue	OR	allele
HLA-DRβ1	amino acid position 37	coding	$4.99x10^{-73}$	0.42	Asn
				0.74	Ser
				1.19	Phe
				1.54	Tyr
				1.56	Leu
HLA-DRA	rs9268657	intron (eQTL)	$4.48x10^{-41}$	1.66	A
HLA-DRβ1	HLA-DRB1*04:01	coding	$1.76x10^{-12}$	1.64	
HLA-DRβ1	amino acid position 13	coding	$4.57x10^{-16}$	0.57	His
				0.5	Gly
				1.35	Ser
				1.41	Tyr
				1.09	Phe
				0.98	Arg

Supplementary Table 3. Summary of bona fide alopecia areata risk loci. For each of the loci in Table 1, p-values and effect estimates obtained from each of the analyses are presented. Data is extracted from Supplementary Data 1 and Supplementary Data 2.

Locus	Genes of Interest	SNP	CHR	BP	A1A2	Cohort	P	OR
6p21.32	HLA-DQA1-2,HLADQB2	rs9275524	6	32,783,087	TC	combined	**1.8E-60**	0.52
						US1	9.7E-14	0.38
						US2	9.9E-28	0.47
						CE1	4.5E-08	0.67
						CE2	1.4E-14	0.50
						replication	8.4E-07	0.54
10p15.1	IL15RA,IL2RA	rs3118470	10	6,141,719	TC	combined	**7.7E-21**	0.71
						US1	3.6E-04	0.66
						US2	1.3E-09	0.68
						CE1	1.2E-02	0.83
						CE2	2.2E-06	0.67
						replication	1.2E-04	0.71
2q33.2	CD28,CTLA4,ICOS	rs231775	2	204,440,959	AG	combined	**2.2E-20**	0.72
						US1	3.3E-06	0.60
						US2	1.3E-10	0.67
						CE1	5.2E-02	0.87
						CE2	4.8E-04	0.75
						replication	3.4E-05	0.69
6q25.1	RAET1L,ULBP3	rs12183587	6	150,396,301	TG	combined	**5.9E-24**	1.48
						US1	1.0E-09	2.04
						US2	4.2E-12	1.54
						CE1	1.7E-03	1.25
						CE2	6.6E-06	1.45
						replication	N/A	N/A
11q13	PRDX5	rs574087	11	63,859,524	AG	combined	**8.7E-14**	1.32
						US1	1.4E-01	1.19
						US2	5.1E-07	1.38
						CE1	2.9E-03	1.25
						CE2	4.9E-05	1.41
						replication	1.8E-02	1.26
12q13	IKZF4 **(Eos)**,ERBB3	rs2292239	12	54,768,447	TG	combined	**4.4E-09**	1.25
						US1	2.2E-01	1.15
						US2	1.2E-08	1.45
						CE1	5.9E-02	1.16
						CE2	4.1E-02	1.20
						replication	2.8E-01	1.11
4q27	IL21,**IL2**	rs7682481	4	123,743,476	CG	combined	**4.8E-09**	1.23
						US1	6.4E-04	1.45
						US2	2.5E-05	1.29
						CE1	2.1E-01	1.10
						CE2	6.1E-03	1.26
						replication	1.6E-01	1.14

5q31.1	**IL13**,1L4	rs848	5	132,024,399	AC	combined	**4.8E-09**	1.27
						US1	1.6E-05	1.71
						US2	1.3E-02	1.19
						CE1	9.2E-04	1.33
						CE2	1.3E-01	1.15
						replication	4.4E-02	1.24
2q13	ACOXL,BCL2L11(**BIM**)	rs3789129	2	111,414,511	AC	combined	**1.5E-08**	1.31
						US1	9.1E-02	1.29
						US2	2.9E-03	1.28
						CE1	1.7E-03	1.37
						CE2	2.5E-02	1.29
						replication	1.5E-02	1.31
11q13.5	GARP(LRRC32)	rs2155219	11	75,976,842	TG	combined	**4.1E-08**	1.21
						US1	1.9E-01	1.16
						US2	9.8E-03	1.17
						CE1	3.0E-01	1.08
						CE2	6.1E-04	1.32
						replication	2.8E-05	0.68
1p13.2	PTPN22	rs2476601	1	114,179,091	AG	combined	8.9E-08	1.34
						US1	1.3E-03	1.67
						US2	2.3E-02	1.24
						CE1	4.0E-02	1.26
						CE2	5.2E-05	1.76
						replication	4.3E-01	1.12
12q24.12	SH2B3(**LNK**),ATXN2	rs653178	12	110,492,139	TC	combined	1.6E-07	0.84
						US1	2.3E-02	0.79
						US2	2.8E-02	0.88
						CE1	1.1E-01	0.89
						CE2	1.6E-02	0.83
						replication	4.8E-04	0.79
16p13.13	CIITA,CLEC16A,SOCS1	rs3862469	16	11101581	TC	combined	1.7E-07	0.82
						US1	3.0E-02	0.77
						US2	8.8E-03	0.84
						CE1	8.0E-03	0.82
						CE2	1.4E-04	0.71
						replication	6.8E-01	0.96
9q31.1	STX17	rs10124366	9	101,727,524	AG	combined	1.1E-05	0.83
						US1	2.0E-01	0.86
						US2	1.3E-07	0.70
						CE1	8.9E-01	0.99
						CE2	2.1E-01	0.89
						replication	N/A	N/A