

Supplementary Information for
“Genomewide Scan Reveals Association of Psoriasis with IL-23 and NF-κB Pathways”

The Collaborative Association Study of Psoriasis (CASP)

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SUPPLEMENTARY TABLES

Supplementary Table 1 – Summary of Diagnostic Criteria and Key References for Each Sample.

The table summarizes case and control definitions for each sample, and provides relevant references for previously published sample sets. Note that, although case definitions are broadly comparable, control definitions varied slightly by site: some sites used controls known to be free of psoriasis and other autoimmune disorders, others focused on psoriasis free controls only and yet others relied on population controls. Since psoriasis typically affects <2% of the population, we expect these differences in control definition to play a very minor role in our results.

Supplementary Table 1. Summary of Diagnostic Criteria and Key References for Each Sample

Sample Set and References	Case Definition	N
Discovery Samples		
Collection of <i>J.T. Elder</i> ¹	Psoriasis diagnosed by dermatologist*	480
Collection of <i>G. Krueger</i> ^{2,3}	Psoriasis diagnosed by dermatologist, >18 years of age	476
Collection of <i>A. Bowcock</i> ^{4 **}	Psoriasis diagnosed by dermatologist, > 18 years of age, pustular psoriasis excluded	453
Follow-up Samples		
Collection of <i>J.T. Elder</i> ¹	Psoriasis diagnosed by dermatologist*	1642
Collection of <i>M. Weichenthal</i> ⁵	Psoriasis diagnosed by dermatologist	718
Celera Follow-up Set 1, <i>A. Begovich</i> ^{2,3}	Psoriasis diagnosed by dermatologist, PASI score	498
Celera Follow-up Set 2, <i>A. Begovich</i> ^{2,3}	Psoriasis diagnosed by dermatologist	483
Collection of <i>D. Gladman</i> ***	Psoriasis diagnosed by dermatologist	691
Collection of <i>J. Fisher</i> ⁶⁻⁸	Psoriasis diagnosed by dermatologist, isolated palmoplantar psoriasis excluded	346
Collection of <i>A. Bowcock</i> ^{4 **}	Psoriasis diagnosed by dermatologist, > 18 years of age, pustular psoriasis excluded	302
Collection of <i>P. Rahman</i> ⁹	Psoriasis documented by rheumatologist; most cases confirmed by dermatologist	368
Sample Set and References	Control Definition	N
Discovery Samples		
Collection of <i>J.T. Elder</i> ¹	No history of psoriasis, no family history of psoriasis, >18 years of age.	702
Collection of <i>G. Krueger</i> ^{2,3}	No history of psoriasis or auto-immune disorders in controls or first-degree relatives	473
Collection of <i>A. Bowcock</i> ^{4 **}	No history of psoriasis or auto-immune disorders	261
Follow-up Samples		
Collection of <i>J.T. Elder</i> ¹	No history of psoriasis, no family history of psoriasis, >18 years of age.	1101
Collection of <i>M. Weichenthal</i> ⁵	"Healthy" population controls with "no active disorders"	1464
Celera Follow-up Set 1, <i>A. Begovich</i> ^{2,3}	No history of psoriasis or other auto-immune disorders	498
Celera Follow-up Set 2, <i>A. Begovich</i> ^{2,3}	No history of psoriasis	427
Collection of <i>D. Gladman</i>	No evidence of psoriasis, rheumatoid arthritis or other auto-immune disorders	217
Collection of <i>J. Fisher</i> ⁶⁻⁸	No history of psoriasis	486
Collection of <i>A. Bowcock</i> ^{4 **}	Population controls; unknown psoriasis status	500
Collection of <i>P. Rahman</i> ⁹	No history of psoriasis or other auto-immune disorder	358

* Additional selection criteria for Michigan cases included a requirement of at least two psoriatic plaques or a single plaque occupying >1% of total body surface area outside scalp. Individuals that presented only palmoplantar psoriasis, inverse psoriasis or sebopsoriasis were excluded.

** The discovery sample used here overlaps partially with the replication sample of Liu et al⁴. The replication sample used here is the discovery sample of Liu et al⁴. The genotype data used for our analyses was generated independently and is distinct from the genotype data in Liu et al.⁴

*** Previously unpublished sample collected at University of Toronto affiliated hospitals.

References for Supplementary Table 1

1. Nair, R.P. et al. Polymorphisms of the IL12B and IL23R Genes Are Associated with Psoriasis. *J Invest Dermatol* (2008).
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4. Liu, Y. et al. A genome-wide association study of psoriasis and psoriatic arthritis identifies new disease Loci. *PLoS Genet* **4**, e1000041 (2008).
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6. Mahe, E. et al. Physicians' response to a letter to confirm diagnosis in a genetic study of psoriasis. *Eur J Dermatol* **12**, 66-9 (2002).
7. Lesueur, F. et al. Confirmation of psoriasis susceptibility loci on chromosome 6p21 and 20p13 in French families. *J Invest Dermatol* **127**, 1403-9 (2007).
8. Lesueur, F. et al. ADAM33, a new candidate for psoriasis susceptibility. *PLoS ONE* **2**, e906 (2007).
9. Rahman, P. et al. Association between the interleukin-1 family gene cluster and psoriatic arthritis. *Arthritis Rheum* **54**, 2321-5 (2006).

Additional Notes on Supplementary Table 1

Note that nearly all cases were collected in dermatology clinics and had their diagnosis confirmed by a dermatologist (except for a subset of the samples contributed by Dr. Rahman, which were diagnosed by a rheumatologist). A subset of controls for the follow-up sample (contributed by Drs. Bowcock and Weichenthal) were “population controls” and may include a small number of psoriasis cases. Since psoriasis is rare, we expect the impact on power to be negligible.

All cases and controls included in the study correspond to individuals who provided informed consent and reported their ancestry as “Caucasian” or “White, not Hispanic origin”.

Supplementary Table 2 – Detailed association test results and genotype counts.

The table provides detailed association test results including p-values and genotype counts in cases and controls for all successfully genotyped SNPs in each follow-up sample. A small number of SNPs failed genotyping or assay design in some follow-up samples (on average <2 SNPs failed per follow-up sample set) and, thus, their corresponding entries are marked as *NA* (not available) in the Table.

Supplementary Table 2

Locus	SNP	Chr	Pos	Alleles risk/non-risk	Cases			Controls					pvalue	OR
					1/1 ^a	1/2	2/2	Freq ^b	1/1	1/2	2/2	Freq		
GAIN samples (1359 cases, 1400 controls)														
HLA-C	rs12191877	6	31,360,904	T/C	105	642	612	0.313	34	326	1040	0.141	3.9E-53	2.788
IL12B	rs2082412	5	158,650,367	G/A	973	344	20	0.856	862	461	56	0.792	5.5E-10	1.564
TNIP1	rs17728338	5	150,458,511	A/G	10	232	1117	0.093	3	151	1246	0.056	2.0E-07	1.720
IL13	rs20541	5	132,023,863	G/A	929	389	33	0.832	848	491	57	0.783	5.8E-06	1.366
IL13	rs848 ^c	5	132,024,398	C/A	933	393	33	0.831	841	501	58	0.780	1.4E-06	1.391
TNFAIP3	rs610604	6	138,241,110	G/T	181	652	524	0.374	132	625	641	0.318	1.4E-05	1.279
IL23A,STA	rs2066808 ^c	12	55,024,239	A/G	1246	111	2	0.958	1215	177	8	0.931	1.7E-05	1.676
IL23A,STA	rs2066807	12	55,026,949	C/G	1229	111	2	0.957	1194	177	8	0.930	1.5E-05	1.681
IL23R	rs2201841	1	67,466,790	G/A	158	635	566	0.350	117	565	717	0.286	2.9E-07	1.347
TSC1	rs1076160	9	134,765,855	T/C	378	654	323	0.520	301	693	404	0.463	2.2E-05	1.257
IL1RN	rs397211	2	113,608,612	T/C	694	557	104	0.718	634	625	139	0.677	1.0E-03	1.213
SMARCA4	rs12983316	19	10,975,352	G/A	39	421	880	0.186	24	348	1007	0.144	2.3E-05	1.365
MAML2	rs10765787	11	95,455,226	T/C	34	376	944	0.164	19	310	1070	0.124	2.9E-05	1.381
CNTN5	rs12807920	11	98,996,747	C/T	403	682	273	0.548	345	698	355	0.496	1.3E-04	1.229
DYTN	rs13425179	2	207,251,268	G/A	717	513	118	0.722	600	634	149	0.663	2.2E-06	1.321
SESN1	rs2273668	6	109,430,212	G/T	1100	247	11	0.901	1045	328	26	0.864	2.3E-05	1.430
PRKRP1	rs10238421 ^c	7	101,831,694	A/G	792	499	68	0.766	731	561	108	0.723	1.9E-04	1.260
RGS6	rs2239247	14	71,871,989	A/G	65	433	837	0.211	38	373	968	0.163	5.5E-06	1.374
STK32A	rs4705036	5	146,665,485	T/C	992	336	31	0.854	908	438	53	0.806	2.2E-06	1.407
COL22A1	rs7845922 ^c	8	140,467,714	C/A	138	593	628	0.320	87	558	755	0.261	1.8E-06	1.328
PRKRP1	rs3988090	7	101,836,025	A/G	859	448	49	0.799	774	529	93	0.744	1.3E-06	1.366
University of Michigan follow-up samples (1642 cases, 1101 controls)														
HLA-C	rs12191877	6	31,360,904	T/C	101	758	766	0.295	22	251	816	0.135	1.0E-42	2.676
IL12B	rs2082412	5	158,650,367	G/A	1177	403	39	0.851	684	355	58	0.785	3.2E-10	1.567
TNIP1	rs17728338	5	150,458,511	A/G	11	262	1361	0.087	4	108	983	0.053	2.4E-06	1.702
IL13	rs20541	5	132,023,863	G/A	1115	465	44	0.830	653	382	54	0.775	5.3E-07	1.415
IL13	rs848	5	132,024,398	C/A	1105	472	45	0.827	656	387	57	0.772	6.5E-07	1.407
TNFAIP3	rs610604	6	138,241,110	G/T	205	757	659	0.360	108	481	512	0.317	0.00092	1.214
IL23A,STA	rs2066808	12	55,024,239	A/G	1472	151	5	0.951	947	139	7	0.930	0.0014	1.447
IL23A,STA	rs2066807	12	55,026,949	C/G	1480	145	4	0.953	965	125	8	0.936	0.0057	1.392
IL23R	rs2201841	1	67,466,790	G/A	167	724	735	0.325	93	454	551	0.291	0.0081	1.172
TSC1	rs1076160	9	134,765,855	T/C	384	817	427	0.487	237	532	316	0.464	0.094	1.098
IL1RN	rs397211	2	113,608,612	T/C	819	661	152	0.704	534	462	102	0.697	0.546	1.037
SMARCA4	rs12983316	19	10,975,352	G/A	41	428	1164	0.156	22	271	808	0.143	0.184	1.109
MAML2	rs10765787	11	95,455,226	T/C	33	388	1207	0.139	25	280	789	0.151	0.241	0.912
CNTN5	rs12807920	11	98,996,747	C/T	443	804	377	0.520	313	521	252	0.528	0.575	0.969
DYTN	rs13425179	2	207,251,268	G/A	761	674	173	0.683	555	439	100	0.708	0.049	0.888
SESN1	rs2273668	6	109,430,212	G/T	1260	337	30	0.878	836	247	18	0.871	0.475	1.061
PRKRP1	rs10238421	7	101,831,694	A/G	931	577	93	0.762	555	350	38	0.774	0.312	0.933
RGS6	rs2239247	14	71,871,989	A/G	50	509	1075	0.186	40	314	743	0.180	0.526	1.046
STK32A	rs4705036	5	146,665,485	T/C	1122	454	49	0.830	764	295	28	0.839	0.416	0.941
COL22A1	rs7845922	8	140,467,714	C/A	125	613	891	0.265	70	463	557	0.277	0.340	0.942
PRKRP1	rs3988090	7	101,836,025	A/G	949	587	94	0.762	638	403	45	0.773	0.359	0.942

Supplementary Table 2

Locus	SNP	Chr	Pos	Alleles risk/non-risk	Cases			Controls					pvalue	OR
					1/1 ^a	1/2	2/2	Freq ^b	1/1	1/2	2/2	Freq		
Newfoundland follow-up samples (368 cases, 358 controls)														
HLA-C	rs12191877	6	31,360,904	T/C	8	122	236	0.189	4	74	276	0.116	1.3E-04	1.774
IL12B	rs2082412	5	158,650,367	G/A	264	89	13	0.843	235	107	13	0.813	0.128	1.237
TNIP1	rs17728338	5	150,458,511	A/G	3	67	297	0.099	4	40	311	0.068	0.029	1.523
IL13	rs20541	5	132,023,863	G/A	251	105	11	0.827	227	115	13	0.801	0.212	1.184
IL13	rs848	5	132,024,398	C/A	236	103	11	0.821	216	115	13	0.795	0.212	1.186
TNFAIP3	rs610604	6	138,241,110	G/T	52	149	166	0.345	39	155	162	0.327	0.483	1.081
IL23A,STA	rs2066808	12	55,024,239	A/G	328	38	1	0.946	304	50	0	0.929	0.206	1.318
IL23A,STA	rs2066807	12	55,026,949	C/G	328	38	1	0.946	307	48	0	0.932	0.298	1.258
IL23R	rs2201841	1	67,466,790	G/A	33	154	163	0.314	44	138	162	0.328	0.571	0.937
TSC1	rs1076160	9	134,765,855	T/C	85	187	94	0.488	92	169	95	0.496	0.759	0.968
IL1RN	rs397211	2	113,608,612	T/C	180	155	32	0.702	158	166	31	0.679	0.350	1.112
SMARCA4	rs12983316	19	10,975,352	G/A	16	91	242	0.176	10	79	256	0.143	0.096	1.277
MAML2	rs10765787	11	95,455,226	T/C	11	95	261	0.159	7	96	252	0.155	0.815	1.034
CNTN5	rs12807920	11	98,996,747	C/T	86	196	83	0.504	98	173	84	0.520	0.554	0.939
DYTN	rs13425179	2	207,251,268	G/A	178	134	42	0.692	163	153	38	0.677	0.529	1.075
SESN1	rs2273668	6	109,430,212	G/T	274	87	5	0.867	281	68	6	0.887	0.251	0.831
PRKRP1	rs10238421	7	101,831,694	A/G	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
RGS6	rs2239247	14	71,871,989	A/G	12	100	255	0.169	8	97	249	0.160	0.633	1.070
STK32A	rs4705036	5	146,665,485	T/C	259	87	12	0.845	253	90	10	0.844	0.968	1.006
COL22A1	rs7845922	8	140,467,714	C/A	30	159	178	0.298	32	136	187	0.282	0.485	1.084
PRKRP1	rs3988090	7	101,836,025	A/G	227	111	11	0.809	209	127	7	0.794	0.484	1.099
Toronto follow-up samples (691 cases, 217 controls)														
HLA-C	rs12191877	6	31,360,904	T/C	52	250	381	0.259	4	55	158	0.145	9.4E-07	2.060
IL12B	rs2082412	5	158,650,367	G/A	491	176	17	0.846	148	53	16	0.804	0.038	1.343
TNIP1	rs17728338	5	150,458,511	A/G	8	126	551	0.104	0	32	185	0.074	0.066	1.453
IL13	rs20541	5	132,023,863	G/A	463	200	22	0.822	129	74	14	0.765	0.0087	1.418
IL13	rs848	5	132,024,398	C/A	453	199	24	0.817	127	71	15	0.763	0.014	1.390
TNFAIP3	rs610604	6	138,241,110	G/T	100	318	268	0.378	23	106	88	0.350	0.304	1.125
IL23A,STA	rs2066808	12	55,024,239	A/G	620	65	1	0.951	190	25	2	0.933	0.145	1.395
IL23A,STA	rs2066807	12	55,026,949	C/G	628	60	1	0.955	194	21	2	0.942	0.284	1.297
IL23R	rs2201841	1	67,466,790	G/A	83	289	308	0.335	23	94	98	0.326	0.730	1.041
TSC1	rs1076160	9	134,765,855	T/C	188	336	162	0.519	52	102	62	0.477	0.127	1.184
IL1RN	rs397211	2	113,608,612	T/C	357	269	60	0.716	102	87	28	0.671	0.067	1.242
SMARCA4	rs12983316	19	10,975,352	G/A	20	184	478	0.164	5	59	151	0.160	0.854	1.028
MAML2	rs10765787	11	95,455,226	T/C	14	198	472	0.165	5	49	163	0.136	0.146	1.258
CNTN5	rs12807920	11	98,996,747	C/T	193	329	163	0.522	56	109	52	0.509	0.645	1.052
DYTN	rs13425179	2	207,251,268	G/A	326	267	61	0.703	113	73	26	0.705	0.919	0.988
SESN1	rs2273668	6	109,430,212	G/T	551	129	5	0.899	169	45	3	0.882	0.342	1.179
PRKRP1	rs10238421	7	101,831,694	A/G	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
RGS6	rs2239247	14	71,871,989	A/G	22	198	467	0.176	8	72	137	0.203	0.210	0.841
STK32A	rs4705036	5	146,665,485	T/C	494	160	18	0.854	148	59	9	0.822	0.104	1.270
COL22A1	rs7845922	8	140,467,714	C/A	52	273	361	0.275	13	90	113	0.269	0.799	1.032
PRKRP1	rs3988090	7	101,836,025	A/G	407	233	39	0.771	120	81	13	0.750	0.371	1.122

Supplementary Table 2

Locus	SNP	Chr	Pos	Alleles risk/non-risk	Cases			Controls			pvalue	OR		
					1/1 ^a	1/2	2/2	Freq ^b	1/1	1/2	2/2			
Kiel follow-up samples (718 cases, 1464 controls)														
HLA-C	rs12191877	6	31,360,904	T/C	69	356	286	0.347	30	353	1057	0.143	1.0E-53	3.180
IL12B	rs2082412	5	158,650,367	G/A	519	176	16	0.854	945	441	53	0.810	3.8E-04	1.370
TNIP1	rs17728338	5	150,458,511	A/G	4	90	619	0.069	3	130	1312	0.047	0.0031	1.494
IL13	rs20541	5	132,023,863	G/A	462	220	25	0.809	866	502	63	0.781	0.031	1.191
IL13	rs848	5	132,024,398	C/A	464	223	24	0.809	875	498	67	0.781	0.029	1.194
TNFAIP3	rs610604	6	138,241,110	G/T	85	333	286	0.357	157	613	668	0.322	0.023	1.169
IL23A,STA	rs2066808	12	55,024,239	A/G	635	72	1	0.948	1256	185	12	0.928	0.014	1.405
IL23A,STA	rs2066807	12	55,026,949	C/G	638	69	1	0.950	1238	181	10	0.930	0.011	1.433
IL23R	rs2201841	1	67,466,790	G/A	71	301	337	0.312	122	559	772	0.276	0.014	1.190
TSC1	rs1076160	9	134,765,855	T/C	163	349	193	0.479	306	714	412	0.463	0.332	1.065
IL1RN	rs397211	2	113,608,612	T/C	362	282	57	0.718	689	619	133	0.693	0.099	1.126
SMARCA4	rs12983316	19	10,975,352	G/A	14	188	501	0.154	46	373	1020	0.162	0.504	0.942
MAML2	rs10765787	11	95,455,226	T/C	14	166	528	0.137	37	357	1057	0.149	0.313	0.910
CNTN5	rs12807920	11	98,996,747	C/T	186	362	164	0.515	442	700	296	0.551	0.029	0.868
DYTN	rs13425179	2	207,251,268	G/A	325	309	77	0.674	673	601	175	0.672	0.866	1.012
SESN1	rs2273668	6	109,430,212	G/T	566	137	6	0.895	1094	321	32	0.867	0.0088	1.307
PRKRP1	rs10238421	7	101,831,694	A/G	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
RGS6	rs2239247	14	71,871,989	A/G	20	227	461	0.189	37	436	955	0.179	0.426	1.069
STK32A	rs4705036	5	146,665,485	T/C	461	231	16	0.814	939	447	59	0.804	0.445	1.065
COL22A1	rs7845922	8	140,467,714	C/A	51	262	396	0.257	81	559	797	0.251	0.679	1.031
PRKRP1	rs3988090	7	101,836,025	A/G	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
Washington University follow-up samples (302 cases, 500 controls)														
HLA-C	rs12191877	6	31,360,904	T/C	32	126	140	0.319	13	104	376	0.132	3.0E-19	3.081
IL12B	rs2082412	5	158,650,367	G/A	220	72	8	0.853	325	150	18	0.811	0.032	1.353
TNIP1	rs17728338	5	150,458,511	A/G	1	53	247	0.091	1	42	453	0.044	1.6E-04	2.166
IL13	rs20541	5	132,023,863	G/A	204	85	12	0.819	323	160	12	0.814	0.811	1.033
IL13	rs848	5	132,024,398	C/A	199	81	12	0.820	316	147	14	0.817	0.857	1.025
TNFAIP3	rs610604	6	138,241,110	G/T	44	126	130	0.357	46	205	244	0.300	0.019	1.294
IL23A,STA	rs2066808	12	55,024,239	A/G	276	24	0	0.960	425	68	2	0.927	0.0079	1.882
IL23A,STA	rs2066807	12	55,026,949	C/G	280	21	0	0.965	430	67	1	0.931	0.0039	2.059
IL23R	rs2201841	1	67,466,790	G/A	31	137	132	0.332	49	205	239	0.307	0.312	1.119
TSC1	rs1076160	9	134,765,855	T/C	83	149	68	0.525	131	259	107	0.524	0.974	1.003
IL1RN	rs397211	2	113,608,612	T/C	146	151	2	0.741	231	251	5	0.732	0.702	1.046
SMARCA4	rs12983316	19	10,975,352	G/A	6	90	204	0.170	11	113	373	0.136	0.063	1.303
MAML2	rs10765787	11	95,455,226	T/C	6	70	222	0.138	16	144	335	0.178	0.036	0.738
CNTN5	rs12807920	11	98,996,747	C/T	89	147	61	0.547	142	236	111	0.532	0.552	1.064
DYTN	rs13425179	2	207,251,268	G/A	141	129	29	0.687	244	198	48	0.700	0.595	0.942
SESN1	rs2273668	6	109,430,212	G/T	229	64	5	0.876	379	108	6	0.878	0.885	0.977
PRKRP1	rs10238421	7	101,831,694	A/G	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
RGS6	rs2239247	14	71,871,989	A/G	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
STK32A	rs4705036	5	146,665,485	T/C	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
COL22A1	rs7845922	8	140,467,714	C/A	29	101	170	0.265	35	196	261	0.270	0.817	0.973
PRKRP1	rs3988090	7	101,836,025	A/G	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA

Supplementary Table 2

Locus	SNP	Chr	Pos	Alleles risk/non-risk	Cases			Controls				pvalue	OR	
					1/1 ^a	1/2	2/2	Freq ^b	1/1	1/2	2/2	Freq		
^dFrance follow-up samples (346 cases, 486 controls)														
HLA-C	rs12191877	6	31,360,904	T/C	38	199	105	0.402	21	175	283	0.227	5.3E-13	2.296
IL12B	rs2082412	5	158,650,367	G/A	234	99	9	0.829	308	156	20	0.798	0.294	1.230
TNIP1	rs17728338	5	150,458,511	A/G	2	51	279	0.083	1	48	413	0.054	0.035	1.578
IL13	rs20541	5	132,023,863	G/A	244	75	18	0.835	304	156	16	0.803	0.124	1.248
IL13	rs848	5	132,024,398	C/A	242	77	19	0.830	302	158	16	0.800	0.121	1.217
TNFAIP3	rs610604	6	138,241,110	G/T	51	155	136	0.376	56	212	214	0.336	0.186	1.189
IL23A,STA	rs2066808	12	55,024,239	A/G	313	29	0	0.958	447	35	0	0.964	0.877	0.851
IL23A,STA	rs2066807	12	55,026,949	C/G	322	21	0	0.969	452	31	0	0.968	0.383	1.051
IL23R	rs2201841	1	67,466,790	G/A	35	144	163	0.313	38	187	257	0.273	0.521	1.214
TSC1	rs1076160	9	134,765,855	T/C	78	180	84	0.491	121	231	127	0.494	0.943	0.990
IL1RN	rs397211	2	113,608,612	T/C	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
SMARCA4	rs12983316	19	10,975,352	G/A	8	73	264	0.129	9	95	373	0.118	0.448	1.103
MAML2	rs10765787	11	95,455,226	T/C	6	65	268	0.114	8	123	341	0.147	0.137	0.742
CNTN5	rs12807920	11	98,996,747	C/T	115	155	73	0.561	164	226	93	0.573	0.864	0.951
DYTN	rs13425179	2	207,251,268	G/A	160	145	34	0.686	229	201	50	0.686	0.804	0.997
SESN1	rs2273668	6	109,430,212	G/T	262	76	4	0.877	358	109	10	0.865	0.288	1.117
PRK RIP1	rs10238421	7	101,831,694	A/G	190	132	15	0.760	285	176	17	0.780	0.426	0.890
RGS6	rs2239247	14	71,871,989	A/G	17	97	227	0.192	14	152	319	0.186	0.775	1.043
STK32A	rs4705036	5	146,665,485	T/C	224	104	8	0.821	316	144	20	0.808	0.924	1.091
COL22A1	rs7845922	8	140,467,714	C/A	26	137	178	0.277	34	201	247	0.279	0.883	0.991
PRK RIP1	rs3988090	7	101,836,025	A/G	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
Celera Set 1 follow-up samples (498 cases, 498 controls)														
HLA-C	rs12191877	6	31,360,904	T/C	32	219	239	0.289	10	120	360	0.143	4.1E-15	2.436
IL12B	rs2082412	5	158,650,367	G/A	364	116	12	0.858	294	173	22	0.778	4.9E-06	1.719
TNIP1	rs17728338	5	150,458,511	A/G	1	75	417	0.078	2	44	445	0.049	0.0079	1.648
IL13	rs20541	5	132,023,863	G/A	339	142	12	0.832	312	171	12	0.803	0.100	1.212
IL13	rs848	5	132,024,398	C/A	339	142	12	0.832	308	171	13	0.800	0.068	1.237
TNFAIP3	rs610604	6	138,241,110	G/T	51	230	211	0.337	62	203	226	0.333	0.836	1.020
IL23A,STA	rs2066808	12	55,024,239	A/G	420	71	2	0.924	420	68	3	0.925	0.953	0.990
IL23A,STA	rs2066807	12	55,026,949	C/G	424	67	2	0.928	421	65	3	0.927	0.960	1.009
IL23R	rs2201841	1	67,466,790	G/A	63	194	235	0.325	49	207	236	0.310	0.468	1.073
TSC1	rs1076160	9	134,765,855	T/C	118	258	117	0.501	98	255	138	0.459	0.064	1.182
IL1RN	rs397211	2	113,608,612	T/C	235	202	56	0.682	242	198	51	0.695	0.535	0.941
SMARCA4	rs12983316	19	10,975,352	G/A	14	123	355	0.153	13	111	367	0.140	0.382	1.118
MAML2	rs10765787	11	95,455,226	T/C	7	127	359	0.143	10	146	335	0.169	0.111	0.820
CNTN5	rs12807920	11	98,996,747	C/T	144	218	131	0.513	140	248	102	0.539	0.256	0.902
DYTN	rs13425179	2	207,251,268	G/A	229	214	49	0.683	249	206	34	0.720	0.074	0.838
SESN1	rs2273668	6	109,430,212	G/T	386	98	8	0.884	380	105	4	0.884	0.983	0.997
PRK RIP1	rs10238421	7	101,831,694	A/G	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
RGS6	rs2239247	14	71,871,989	A/G	23	145	325	0.194	13	156	321	0.186	0.651	1.053
STK32A	rs4705036	5	146,665,485	T/C	329	149	14	0.820	340	137	14	0.832	0.488	0.921
COL22A1	rs7845922	8	140,467,714	C/A	29	213	251	0.275	39	188	263	0.271	0.865	1.017
PRK RIP1	rs3988090	7	101,836,025	A/G	285	182	26	0.763	295	161	31	0.771	0.661	0.954

Supplementary Table 2

Locus	SNP	Chr	Pos	Alleles risk/non-risk	Cases			Controls					pvalue	OR
					1/1 ^a	1/2	2/2	Freq ^b	1/1	1/2	2/2	Freq		
Celera Set 2 follow-up samples (483 cases, 427 controls)														
HLA-C	rs12191877	6	31,360,904	T/C	47	220	214	0.326	12	100	313	0.146	3.3E-19	2.837
IL12B	rs2082412	5	158,650,367	G/A	336	130	15	0.834	258	153	15	0.785	0.0085	1.371
TNIP1	rs17728338	5	150,458,511	A/G	3	80	397	0.090	2	59	365	0.074	0.226	1.232
IL13	rs20541	5	132,023,863	G/A	343	131	8	0.848	273	142	11	0.808	0.024	1.325
IL13	rs848	5	132,024,398	C/A	336	138	8	0.840	269	144	13	0.800	0.027	1.311
TNFAIP3	rs610604	6	138,241,110	G/T	67	218	196	0.366	42	164	220	0.291	0.001	1.405
IL23A,STA	rs2066808	12	55,024,239	A/G	424	57	0	0.941	367	52	2	0.933	0.526	1.131
IL23A,STA	rs2066807	12	55,026,949	C/G	429	52	0	0.946	376	50	0	0.941	0.669	1.091
IL23R	rs2201841	1	67,466,790	G/A	61	209	212	0.343	44	184	198	0.319	0.276	1.115
TSC1	rs1076160	9	134,765,855	T/C	124	243	114	0.510	100	199	126	0.469	0.082	1.178
IL1RN	rs397211	2	113,608,612	T/C	239	210	33	0.714	197	190	37	0.689	0.245	1.127
SMARCA4	rs12983316	19	10,975,352	G/A	14	139	329	0.173	12	107	307	0.154	0.263	1.153
MAML2	rs10765787	11	95,455,226	T/C	10	126	345	0.152	6	124	296	0.160	0.645	0.942
CNTN5	rs12807920	11	98,996,747	C/T	120	245	117	0.503	109	223	94	0.518	0.538	0.944
DYTN	rs13425179	2	207,251,268	G/A	239	200	42	0.705	214	178	33	0.713	0.703	0.961
SESN1	rs2273668	6	109,430,212	G/T	363	112	6	0.871	330	93	3	0.884	0.411	0.889
PRKRP1	rs10238421	7	101,831,694	A/G	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
RGS6	rs2239247	14	71,871,989	A/G	15	135	330	0.172	20	107	298	0.173	0.952	0.993
STK32A	rs4705036	5	146,665,485	T/C	333	130	15	0.833	290	123	12	0.827	0.753	1.040
COL22A1	rs7845922	8	140,467,714	C/A	39	203	239	0.292	29	169	228	0.266	0.224	1.136
PRKRP1	rs3988090	7	101,836,025	A/G	291	160	28	0.775	247	159	20	0.766	0.682	1.047

Notes:

a. "1" stands for the risk allele and "2" stands for the non-risk allele.

b. the frequency of the risk allele.

c. SNPs were not originally genotyped in the GAIN study, but instead were imputed using MACH. The most likely genotypes were used to calculate the genotype counts for these imputed SNPs.

d. France follow-up samples are family-based data. The genotype counts were calculated ignoring the family structure, while the association tests were performed taking the family structure into account.

Supplementary Table 3. Results of model building using forward selection with imputed and genotyped SNPs in the vicinity of replicated hits. For each locus, the table lists the selected SNP for each round of model fitting, the evidence for association with the selected SNP (in a logistic regression based analysis that included SNPs selected in previous rounds as covariates) and the odds ratio for association. The r^2 between each SNP and the SNP with confirmed association for each locus is also listed. Note that we preferentially selected genotyped SNPs for follow up, but the forward selection procedure often points to an imputed SNP that exhibits somewhat stronger association. We analyzed imputed SNPs, rather than just genotyped SNPs, because a single (untyped) associated SNP could result in strong association signals for multiple SNPs.

SNP	Position	p-value after adjustment for higher ranked SNPs		SNPs Tested	Odds Ratio	r^2 (w/replicated SNP)
		w/o Sidak correction	w/ Sidak correction			
HLA-C (models fitted including all SNPs within 4 Mb of replicated association at rs12191877)						
Round 1	rs12204500	31,184,375	8×10^{-57}	12,150	3.37	0.64
Round 2	rs13191343	31,349,087	2×10^{-10}	12,149	1.78	0.85
Round 3	rs2022544	32,428,981	1×10^{-7}	12,148	1.58	<0.01
Stop	rs2516514	31,555,114	2×10^{-5}	12,147	1.34	0.12
IL12B (models fitted including all SNPs within 200 Kb of replicated association at rs2082412)						
Round 1	rs3212220	158,686,773	1×10^{-10}	378	1.59	1.00
Round 2	rs10059288	158,791,476	6×10^{-7}	377	1.39	0.01
Stop	rs17056751	158,784,778	1×10^{-3}	376	5.92	<0.01
TNIP1 (models fitted including all SNPs within 200 Kb of replicated association at rs17728338)						
Round 1	rs17728338	150,458,511	1×10^{-7}	566	1.74	1.00
Stop	rs884520	150,464,980	9×10^{-5}	565	2.03	<0.01

(continued on next page ...)

Supplementary Table 3 (continued). Results of model building using forward selection with genotyped and imputed SNPs in the vicinity of replicated hits.

SNP	Position	p-value after adjustment for higher ranked SNPs		SNPs Tested	Odds Ratio	r^2 (w/replicated SNP)	
		w/o Sidak correction	w/ Sidak correction				
IL13 (models fitted including all SNPs within 200 Kb of replicated association at rs20541)							
Round 1	rs848	132,024,399	9×10^{-7}	2x10 ⁻⁴	272	1.41	0.96
Stop	rs2057656	131,837,204	4×10^{-3}	0.64	271	1.20	< 0.01
TNFAIP3 (models fitted including all SNPs within 200 Kb of replicated association at rs610604)							
Round 1	rs582757	138,239,517	7×10^{-6}	0.002	333	1.31	0.78
Stop	rs9321627	138,064,741	1×10^{-3}	0.38	332	1.24	< 0.01
IL23A (models fitted including all SNPs within 200 Kb of replicated association at rs2066808)							
Round 1	rs2066807	55,026,949	9×10^{-6}	0.001	140	1.69	1
Stop	rs872103	54,954,441	3×10^{-2}	0.98	139	1.34	< 0.01
IL23R (models fitted including all SNPs within 200 Kb of replicated association at rs2201841)							
Round 1	rs12119179	67,520,003	2×10^{-7}	6×10^{-5}	267	1.35	0.58
Round 2	rs11465804	67,475,114	5×10^{-5}	0.01	266	1.62	0.03
Stop	rs10789229	67,478,162	8×10^{-3}	0.88	265	1.18	0.26

Supplementary Table 4. Evidence for Heterogeneity at Replicated Hits.

SNP	Chr	Position (Mb)	Cochran's Q	Cochran's Q p-value	I^2 [%]	95% Confidence Interval for I^2	Notable Nearby Genes
rs12191877	6	31.36	17.59	0.0073	65.9	[23.6%; 84.8%]	<i>HLA-C</i>
rs2082412	5	158.65	5.65	0.46	0.0	[0.0%; 69.0%]	<i>IL12B</i>
rs17728338	5	150.46	5.19	0.52	0.0	[0.0%; 66.2%]	<i>TNIP1</i>
rs20541	5	132.02	6.65	0.35	9.8	[0.0%; 73.7%]	<i>IL13</i>
rs610604	6	138.24	7.06	0.32	15.0	[0.0%; 58.7%]	<i>TNFAIP3</i>
rs2066808 ^b	12	55.02	6.48	0.37	7.4	[0.0%; 73.0%]	<i>IL23A</i>
rs2201841	1	67.47	4.30	0.64	0.0	[0.0%; 59.3%]	<i>IL23R</i>

Evidence for heterogeneity was evaluated in the seven confirmed loci using Cochran's Q statistic and the I^2 statistic. Slightly stronger (but still non-significant) evidence for heterogeneity was observed at all loci when discovery samples were included in the analysis due to the winner's curse effect.

Supplementary Table 5. Evidence for Association with Psoriatic Arthritis at the Ten Loci Listed in Table 2.

SNP	Chr	Pos (Mb)	Alleles risk/ non-risk	Cutaneous Psoriasis Only (3523 cases, 5942 controls)			Psoriatic Arthritis Only (1755 cases, 5942 controls)			Psoriatic Arthritis vs. Cutaneous Cases (p-value)	Notable Genes	
				Frequency ^a		OR (meta) ^b	p-value (meta) ^c	Frequency ^a		OR (meta) ^b	p-value (meta) ^c	
				Case	Control			Case	Control			
rs12191877	6	31.36	T/C	.316	.139	2.87	3×10^{-178}	.260	.139	2.34	6×10^{-62}	.006 <i>HLA-C</i>
rs2082412	5	158.65	G/A	.846	.797	1.42	5×10^{-18}	.861	.797	1.63	4×10^{-16}	.01 <i>IL12B</i>
rs17728338	5	150.46	A/G	.083	.054	1.56	1×10^{-13}	.100	.054	1.78	2×10^{-14}	.07 <i>TNIP1</i>
rs20541	5	132.02	G/A	.821	.787	1.24	4×10^{-8}	.833	.787	1.34	1×10^{-7}	.11 <i>IL13</i>
rs610604	6	138.24	G/T	.364	.318	1.23	3×10^{-10}	.362	.318	1.18	3×10^{-4}	.67 <i>TNFAIP3</i>
rs2066807 ^d	12	55.03	C/G	.949	.932	1.33	2×10^{-5}	.955	.932	1.55	9×10^{-6}	.23 <i>IL23A</i>
rs2201841	1	67.47	G/A	.340	.295	1.23	2×10^{-10}	.318	.295	1.07	.142	.02 <i>IL23R</i>
rs1076160	9	134.77	T/C	.499	.471	1.13	6×10^{-5}	.511	.471	1.16	7×10^{-4}	.52 <i>TSC1</i>
rs12983316	19	10.98	G/A	.166	.148	1.15	7×10^{-4}	.174	.148	1.24	2×10^{-4}	.38 <i>SMARCA4</i>
rs397211	2	113.61	T/C	.714	.691	1.12	8×10^{-4}	.712	.691	1.11	.024	.79 <i>IL1RN</i>

^a Frequency of the risk allele.

^b Mantel-Haenszel estimator of the common odds ratio.

^c Two-tailed significance of allelic association across collection centers using the Cochran-Mantel-Haenszel test.

^d rs2066807 is a genotyped marker in strong LD with rs2066808 ($r^2 = 1.0$ in HapMap) where we observed the strongest overall evidence for association, but which was imputed in GWAS samples.

Supplementary Table 6. Differences in Expression between Psoriatic and Normal Skin for Genes Highlighted by Genomewide Scan and Follow-up Analyses.

Transcript	Raw Mean Expression (RMA, log scale)			Uninvolved vs. Control		Involved vs. Uninvolved	
	Control	Uninvolved	Involved	fold change	p-value	fold change	p-value
Genes in Loci With Confirmed Association ($p_{\text{combined}} < 5 \times 10^{-8}$)							
HLA-C	12.32	12.31	12.49	1.00	0.93	1.14	1.7×10^{-10}
IL12B	4.24	4.26	4.53	1.01	0.54	1.20	1.1×10^{-14}
TNIP1	9.62	9.45	10.01	0.90	0.00024	1.47	3.8×10^{-36}
IL13	5.76	5.86	5.93	1.07	0.013	1.05	0.033
TNFAIP3	8.54	8.60	8.59	1.03	0.34	0.99	0.79
IL23A	5.38	5.56	5.83	1.12	4.9×10^{-5}	1.20	4.0×10^{-12}
IL23R	4.29	4.30	4.36	1.01	0.77	1.04	0.051
Genes in Loci With Suggestive Association ($p_{\text{follow-up}} < 0.05$)							
TSC1	6.94	6.88	6.79	0.96	0.055	0.94	0.00090
SMARCA4	7.09	7.11	7.20	1.01	0.55	1.06	0.0053
IL1RN	7.65	7.76	8.48	1.08	0.018	1.64	8.2×10^{-34}

The table summarizes differences in expression levels for genes in regions of replicated association. The first column indicates the gene name for each of the transcripts investigated. When multiple transcripts for a gene were present on Affymetrix U133 2.0A arrays, results were averaged across all transcripts for the same gene. The next three columns summarize average of RMA expression levels for each tissue type. The last four columns summarize differences in expression levels between uninvolved and normal skin and between involved and uninvolved skin.

In comparisons of involved and uninvolved skin, approximately 25% of all genes show differences in expression similar to those observed for *HLA-C*, and thus we estimate the probability that among seven randomly selected genes four or more would show differences such as the highlighted genes is ~0.066.

Supplementary Table 7. Comparison with the top of Hits of Previously Published Scans.

SNP	Chr	Pos (Mb)	Alleles risk/ non-risk	Capon et al. ²⁶ Total Sample (2679 cases, 2215 controls)			This Study, Discovery Sample (1359 cases, 1400 controls)			Notable Nearby Genes		
				Frequency		OR	p-value	Frequency				
				Case	Control			Case	Control			
rs495337 ^a	20	47.9	C/T	.61	.55	1.25	1×10^{-8}	.63	.58	1.23	2×10^{-4}	ZNF313/RNF114
<hr/>												
SNP	Chr	Pos (Mb)	Alleles risk/ non-risk	Liu et al. ²⁷ Total Sample (960 cases, 1256 controls)			This Study, Discovery Sample ^b (924 cases, 1156 controls)			Notable Nearby Genes		
				Frequency		OR	p-value	Frequency				
				Case	Control			Case	Control			
rs6701216	1	151.0	T/C	.17	.13	1.45	5×10^{-5}	.15	.14	1.09	.305	EDC cluster
rs2164807	2	85.8	G/A	.47	.39	1.35	2×10^{-5}	.43	.43	1.00	.926	GNLY-ATOH8
rs11126740	2	79.7	C/T	.69	.62	1.28	1×10^{-4}	.66	.64	1.08	.170	CTNNA2
rs6804331	3	105.2	C/A	.45	.38	1.30	3×10^{-4}	.41	.41	0.97	.651	(gene desert)
rs3812888	13	39.1	C/T	.43	.35	.1.38	1×10^{-5}	.41	.39	1.07	.287	COG6
rs7993214	13	39.2	C/T	.72	.65	1.41	2×10^{-6}	.33	.35	1.08	.244	COG6
rs2282276	14	94.7	G/A	.10	.07	1.40	3×10^{-3}	.10	.08	1.29	.031	CLMN
rs4775912	15	49.1	G/A	.19	.15	1.41	6×10^{-5}	.17	.17	0.99	.869	USP8-TNFAIP8L3
rs3803369	15	49.2	A/G	.20	.15	1.43	3×10^{-5}	.17	.17	0.99	.878	USP8-TNFAIP8L3

^a The marker rs495337 was not examined in our sample. Instead, we report results for rs2235617, a nearby SNP with $r^2 = 1.0$.

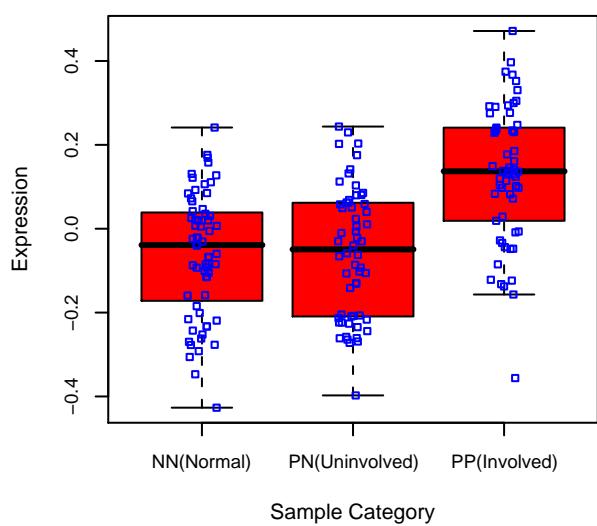
^b Results for this study exclude samples collected by Dr. Bowcock's group (row 3 in Table 1), since those overlap in part with the Liu et al. samples.

SUPPLEMENTARY FIGURE

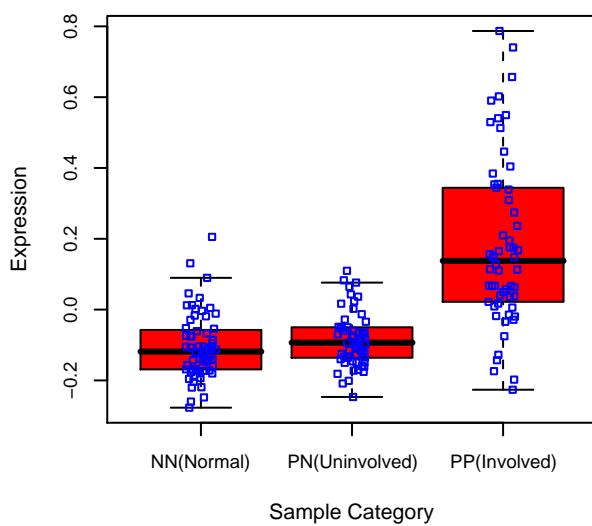
Supplementary Figure 1. Gene Expression Data for the Ten Loci Listed in Table 2.

For each of the transcripts examined, robust multi-chip average (RMA) expression levels were plotted after adjustment for batch and sex effects (Irizarry, R.A. et al. Summaries of Affymetrix GeneChip probe level data. Nucleic Acids Res 31, e15, 2003). We calculated the average impact of sex and batch on gene expression and then subtracted these from the measured RMA levels before plotting). The plots include both individual measurements and box-plots summarizing trait distribution for each of the transcripts.

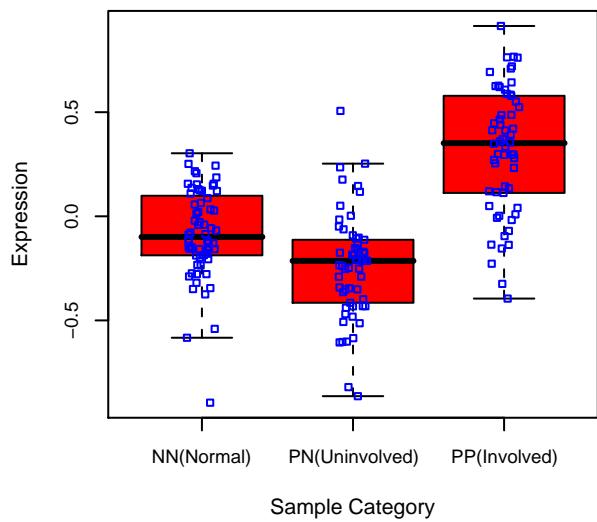
HLA-C
(PN vs. NN p = 0.93; PP vs. PN p = 1.7e-10)



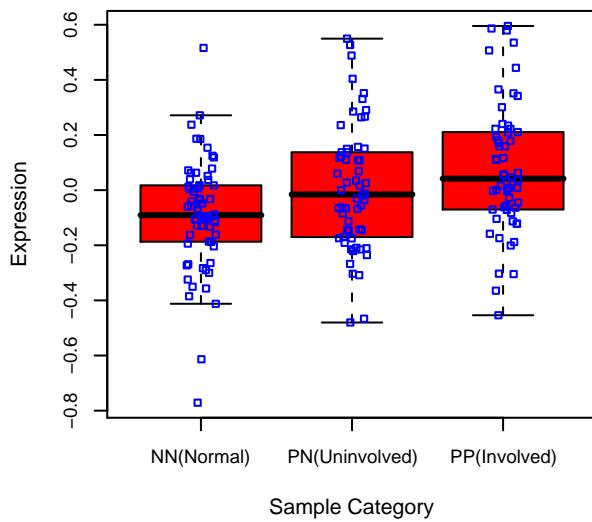
IL12B
(PN vs. NN p = 0.54; PP vs. PN p = 1.1e-14)



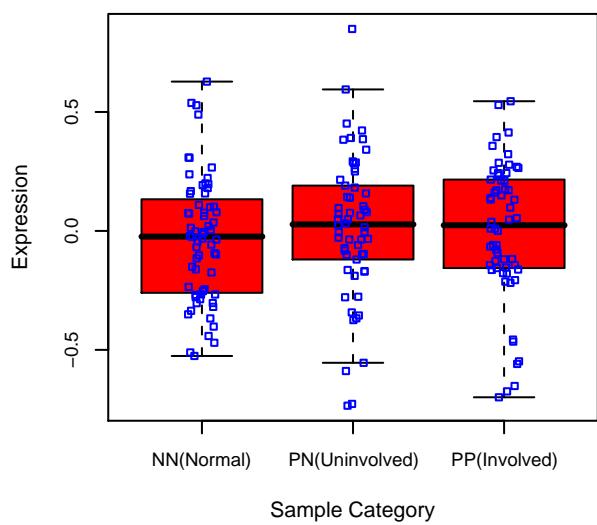
TNIP1
(PN vs. NN p = 0.00024; PP vs. PN p = 3.8e-36)



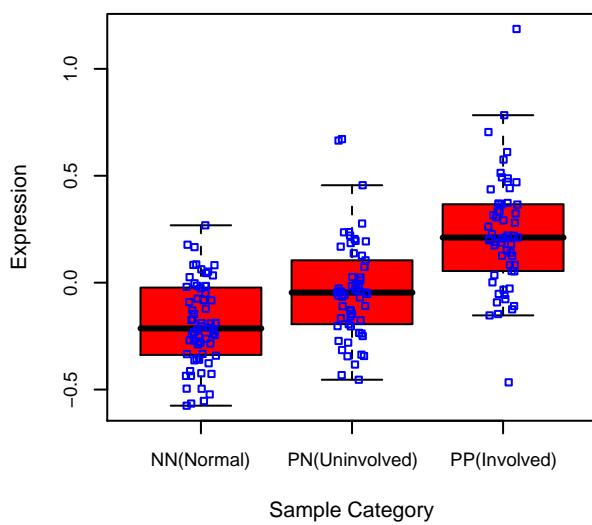
IL13
(PN vs. NN p = 0.013; PP vs. PN p = 0.033)



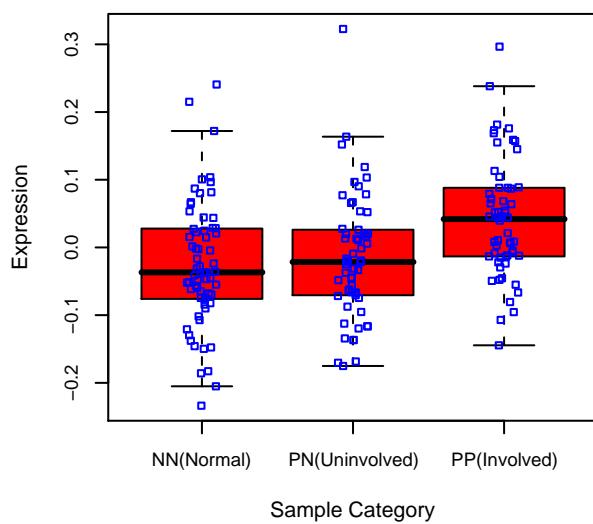
TNFAIP3
(PN vs. NN p = 0.34; PP vs. PN p = 0.79)



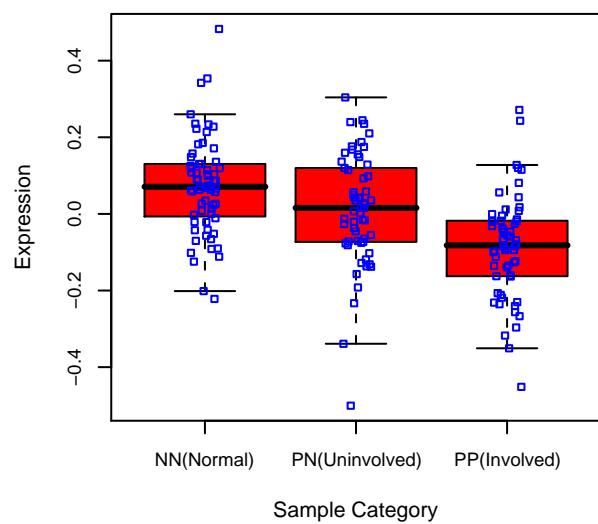
IL23A
(PN vs. NN p = 4.9e-05; PP vs. PN p = 4e-12)



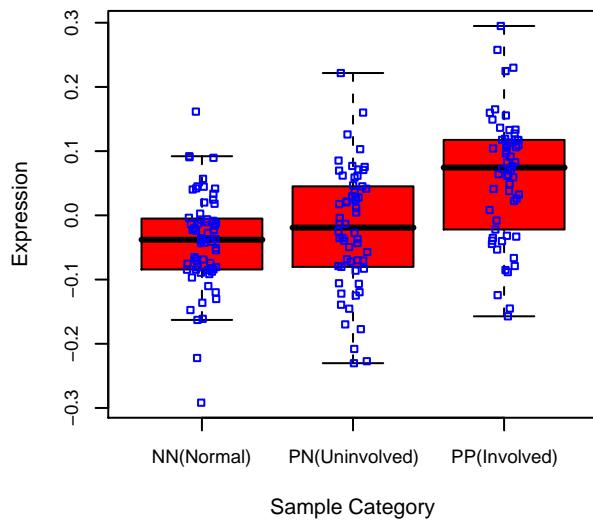
IL23R
(PN vs. NN p = 0.77; PP vs. PN p = 0.051)



TSC1
(PN vs. NN p = 0.055; PP vs. PN p = 9e-04)



SMARCA4
(PN vs. NN p = 0.55; PP vs. PN p = 0.0053)



IL1RN
(PN vs. NN p = 0.018; PP vs. PN p = 8.2e-34)

