

Supplementary Materials: rs657075 (CSF2) Associated with the Disease Phenotype (BAS-G) of Ankylosing Spondylitis

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Table S1. The basic characteristics of the SNPs.

Gene	Pos (hg38)	Variant	Ref	Alt	AFR Freq	AMR Freq	ASN Freq	EUR Freq	JPT Freq	TWB Freq	HWE
<i>B3GNT2</i>	chr2:62225526	rs11900673	C	T	0.12	0.23	0.24	0.13	0.28	0.18	0.53
<i>CSF2</i>	chr5:132094425	rs657075	G	A	0.02	0.07	0.31	0.13	0.36	0.26	0.52
<i>CD83</i>	chr6:14096427	rs12529514	T	C	0.01	0.03	0.19	0.06	0.14	0.23	0.47
<i>NFKBIE</i>	chr6:44265183	rs2233434	A	G	0.05	0.02	0.16	0.04	0.21	0.16	0.85
<i>ARID5B</i>	chr10:62025330	rs10821944	G	T	0.57	0.64	0.70	0.69	0.64	0.74	0.92
<i>PDE2A-ARAP1</i>	chr11:72662452	rs3781913	T	G	0.59	0.48	0.28	0.54	0.31	0.28	0.45
<i>PLD4</i>	chr14:104924668	rs2841277	C	T	0.22	0.45	0.66	0.47	0.69	0.58	0.73
<i>PTPN2</i>	chr18:12797695	rs2847297	A	G	0.57	0.27	0.34	0.32	0.33	0.30	0.06

Freq frequency shows as alt allele. TWB frequency based on Taiwan Biobank. HWE *p* value for Hardy-Weinberg equilibrium.

Table S2. Genotype and allelic frequencies in a Taiwanese biobank of controls and patients with HLA-B27 (+) ankylosing spondylitis.

Single-Nucleotide Polymorphism	Genotype	Cases (%) (<i>n</i> = 431)	Control Subjects (%) (<i>n</i> = 11,301)	Allele	Cases (%) (<i>n</i> = 431)	Control Subjects (%) (<i>n</i> = 11,301)	Dominant <i>p</i> Value	Recessive <i>p</i> Value	Allelic <i>p</i> Value
<i>B3GNT2</i> rs11900673	TT	4 (1.2)	346 (3.0)	T	107 (15.4)	4008 (17.7)	0.2655	0.0394 *	0.1057
	CT	99 (28.4)	3316 (29.4)	C	589 (84.6)	18,574 (82.3)			
	CC	245 (70.4)	7629 (67.6)						
<i>CSF2</i> rs657075	AA	22 (6.3)	711 (6.3)	A	165 (23.6)	5715 (25.7)	0.2085	0.9996	0.3094
	GA	121 (34.7)	4293 (38.1)	G	533 (76.4)	16,481 (74.3)			
	GG	206 (59.0)	6274 (55.6)						
<i>CD83</i> rs12529514	CC	25 (6.9)	613 (5.4)	C	179 (24.7)	5319 (23.6)	0.7497	0.2250	0.4705
	TC	129 (35.6)	4093 (36.3)	T	545 (75.3)	17,251 (76.5)			
	TT	208 (57.5)	6579 (58.3)						
<i>NFKBIE</i> rs2233434	CC	10 (2.5)	12 (2.6)	C	127 (15.6)	151 (16.6)	0.5646	0.8674	0.5778
	TC	107 (26.3)	127 (28.0)	T	685 (84.4)	757 (83.4)			
	TT	289 (71.2)	315 (69.4)						
<i>ARID5B</i> rs10821944	GG	28 (7.9)	792 (7.0)	G	189 (25.5)	5988 (26.5)	0.3163	0.6940	0.5228
	TG	133 (35.6)	4404 (39.0)	T	553 (74.5)	16,588 (73.3)			
	TT	210 (56.5)	6092 (54.0)						
<i>PDE2A-ARAP1</i> rs3781913	CC	41 (10.3)	44 (8.9)	C	244 (30.7)	284 (28.6)	0.2362	0.3280	0.1731
	AC	162 (40.8)	196 (39.4)	A	550 (69.3)	710 (74.3)			
	AA	194 (48.9)	257 (51.7)						
<i>PLD4</i> rs2841277	CC	76 (18.6)	1954 (17.3)	C	344 (42.2)	9370 (41.5)	0.9902	0.4917	0.7148
	TC	192 (47.1)	5462 (48.4)	T	472 (57.8)	13,200 (58.5)			
	TT	140 (34.3)	3869 (34.3)						
<i>PTPN2</i> rs2847297	GG	38 (9.7)	1053 (9.2)	G	257 (32.9)	6728 (29.8)	0.0316 *	0.7836	0.0748
	AG	181 (46.3)	4658 (41.2)	A	525 (67.1)	15,862 (70.2)			
	AA	172 (44.0)	5602 (49.6)						

* Significant ($p < 0.05$) values are in bold.

Table S3. Differences in scores of the Bath Ankylosing Spondylosis (AS) Disease Activity Index (BASDAI), Bath AS Functional Index (BASFI), and Bath AS Global (BAS-G) among HLA-B27 (+) AS patients stratified by different genotypes.

Single-Nucleotide Polymorphism	Genotype	BASDAI	BASFI	BAS-G
<i>B3GNT2</i> rs11900673	TT	3.3 ± 0.9 ^a	0.7 ± 0.9	3.0 ± 3.1
	CT	4.3 ± 2.2	1.9 ± 2.2	4.4 ± 2.6
	CC	4.3 ± 2.1	2.1 ± 2.3	4.3 ± 2.7
<i>p</i> value [†]		0.539	0.269	0.354
<i>q</i> value		0.933	0.858	0.641
<i>CSF2</i> rs657075	AA	5.0 ± 2.2	2.4 ± 2.2	6.2 ± 2.8
	GA	4.3 ± 2.3	2.1 ± 2.3	4.5 ± 2.7
	GG	4.2 ± 2.2	2.0 ± 2.2	4.2 ± 2.7
<i>p</i> value [†]		0.574	0.821	0.021
<i>q</i> value		0.933	0.858	0.168
<i>CD83</i> rs12529514	CC	5.0 ± 2.2	2.3 ± 2.1	5.3 ± 3.0
	TC	4.2 ± 2.2	2.0 ± 2.3	4.5 ± 2.7
	TT	4.4 ± 2.1	2.1 ± 2.2	4.3 ± 2.7
<i>p</i> value [†]		0.583	0.858	0.401
<i>q</i> value		0.933	0.858	0.641
<i>NFKBIE</i> rs2233434	CC	4.7 ± 1.9	1.6 ± 1.7	3.6 ± 2.2
	TC	4.3 ± 2.2	2.0 ± 2.1	4.7 ± 2.7
	TT	4.3 ± 2.1	2.1 ± 2.3	4.3 ± 2.8
<i>p</i> value [†]		0.760	0.669	0.262
<i>q</i> value		0.987	0.858	0.641

Table S3. Cont.

Single-Nucleotide Polymorphism	Genotype	BASDAI	BASFI	BAS-G
<i>ARID5B</i>	GG	4.3 ± 2.2 ^a	2.3 ± 2.0	4.7 ± 3.1
rs10821944	TG	4.4 ± 2.2	2.1 ± 2.3	4.5 ± 2.7
	TT	4.3 ± 2.2	2.0 ± 2.3	4.3 ± 2.7
<i>p</i> value [†]		0.906	0.695	0.481
<i>q</i> value		0.987	0.858	0.641
<i>PDE2A-ARAP1</i>	CC	3.9 ± 2.3	1.7 ± 2.0	4.2 ± 2.8
rs3781913	AC	4.3 ± 2.2	2.0 ± 2.3	4.4 ± 2.7
	AA	4.4 ± 2.2	2.2 ± 2.2	4.5 ± 2.8
<i>p</i> value [†]		0.576	0.500	0.908
<i>q</i> value		0.933	0.858	0.917
<i>PLD4</i>	CC	4.2 ± 2.2	1.7 ± 1.7	4.2 ± 2.5
rs2841277	TC	4.3 ± 2.1	2.2 ± 2.3	4.4 ± 2.7
	TT	4.3 ± 2.3	2.0 ± 2.3	4.5 ± 3.0
<i>p</i> value [†]		0.987	0.192	0.917
<i>q</i> value		0.987	0.858	0.917
<i>PTPN2</i>	GG	4.6 ± 2.5	2.2 ± 2.5	4.6 ± 3.1
rs2847297	AG	4.2 ± 2.2	2.0 ± 2.2	4.2 ± 2.7
	AA	4.4 ± 2.1	2.1 ± 2.3	4.6 ± 2.7
<i>p</i> value [†]		0.534	0.565	0.378
<i>q</i> value		0.933	0.858	0.641

^a Data are presented as the mean ± SD. [†] Adjusted for the effects of age, sex, and disease duration.

Table S4. Differences in the values of immunoglobulin A (IgA), the erythrocyte sedimentation rate (ESR), and C-reactive protein (CRP) among HLA-B27 (+) ankylosing spondylosis (AS) patients stratified by different genotypes.

Single-Nucleotide Polymorphism	Genotype	IgA (mg/dL)	ESR (mm/h)	CRP (mg/dL)
<i>B3GNT2</i>	TT	261.50 ± 62.00 ^a	6.50 ± 2.65	0.45 ± 0.30
<i>rs11900673</i>	CT	312.89 ± 108.81	21.89 ± 17.93	0.94 ± 1.71
	CC	318.83 ± 121.71	26.48 ± 21.21	1.27 ± 1.97
<i>p</i> value [†]		0.684	0.029	0.178
<i>q</i> value		0.768	0.232	0.784
<i>CSF2</i>	AA	335.14 ± 137.68	22.18 ± 16.65	1.26 ± 1.72
<i>rs657075</i>	GA	307.86 ± 122.59	26.32 ± 23.15	1.23 ± 1.98
	GG	328.20 ± 120.08	23.62 ± 18.12	1.03 ± 1.63
<i>p</i> value [†]		0.486	0.846	0.473
<i>q</i> value		0.648	0.846	0.915
<i>CD83</i>	CC	335.14 ± 137.68	22.18 ± 16.65	1.46 ± 2.05
<i>rs12529514</i>	TC	307.86 ± 122.59	26.32 ± 23.15	1.13 ± 1.68
	TT	328.20 ± 120.08	23.62 ± 18.12	1.21 ± 1.99
<i>p</i> value [†]		0.037	0.553	0.801
<i>q</i> value		0.296	0.726	0.915
<i>NFKBIE</i>	CC	304.00 ± 122.81	33.00 ± 25.03	1.64 ± 2.25
<i>rs2233434</i>	TC	303.59 ± 113.37	24.20 ± 22.33	1.16 ± 1.69
	TT	332.09 ± 130.30	26.00 ± 20.96	1.20 ± 1.95
<i>p</i> value [†]		0.200	0.525	0.645
<i>q</i> value		0.376	0.726	0.915

Table S4. Cont.

Single-Nucleotide Polymorphism	Genotype	IgA (mg/dL)	ESR (mm/h)	CRP (mg/dL)
<i>ARID5B</i> rs10821944	GG	359.88 ± 171.70	29.07 ± 26.34	1.39 ± 1.87
	TG	331.17 ± 129.61	25.88 ± 20.91	1.26 ± 2.07
	TT	312.76 ± 112.04	25.01 ± 20.89	1.09 ± 1.74
<i>p</i> value [†]		0.235	0.635	0.694
<i>q</i> value		0.376	0.726	0.915
<i>PDE2A-ARAP1</i> rs3781913	CC	304.56 ± 135.02	22.00 ± 23.25	1.21 ± 2.23
	AC	308.21 ± 102.06	24.25 ± 18.72	1.17 ± 1.86
	AA	335.52 ± 132.66	26.25 ± 22.00	1.19 ± 1.81
<i>p</i> value [†]		0.141	0.552	0.983
<i>q</i> value		0.376	0.726	0.983
<i>PLD4</i> rs2841277	CC	327.62 ± 142.01	22.76 ± 18.40	1.13 ± 1.98
	TC	313.44 ± 126.69	27.81 ± 23.83	1.25 ± 1.92
	TT	336.22 ± 118.80	22.45 ± 18.09	1.07 ± 1.71
<i>p</i> value [†]		0.199	0.078	0.640
<i>q</i> value		0.376	0.312	0.915
<i>PTPN2</i> rs2847297	GG	325.47 ± 142.57	28.08 ± 21.87	1.70 ± 2.61
	AG	319.24 ± 128.04	25.86 ± 22.97	1.20 ± 1.72
	AA	324.53 ± 116.80	24.14 ± 18.88	1.07 ± 1.91
<i>p</i> value [†]		0.768	0.544	0.196
<i>q</i> value		0.768	0.726	0.784

^a Data are presented as the mean ± SD. [†] Adjusted for the effects of age, sex, and disease duration.