

Supplementary Data 1. General characteristics of the AS genetic risk score (AS-GRS) construction and validation sets.

Parameter	AS-GRS construction set		Independent validation set	
	AS patients (n=285)	Controls (n=363)	AS patients (n=576)	Controls (n=680)
Age*				
Average	23.4±8.6	32.4±10.3	28.7±11.5	24.3±14.7
Median	22 (5-62)	29 (20-74)	26 (8-82)	23 (10-80)
Gender				
Male	254 (89.1%)	286 (78.8%)	494 (85.8%)	578 (85.0%)
Female	31 (10.9%)	77 (21.2%)	82 (14.2%)	102 (15.0%)
HLA-B27				
Positive	273 (95.8%)	24 (6.6%)	535 (92.9%)	56 (8.2%)
Negative	12 (4.2%)	339 (93.4%)	41 (7.1%)	624 (91.8%)

* The age at diagnosis was used for cases and the age at recruitment was used for controls.

Supplementary Data 2. Validation of TaqMan CNV assays.

1q32.2 Accuracy = 97.3%		Deletion-typing PCR	
		Deletion	≥ 2N
TaqMan CNV assay	Deletion	227	7
	≥ 2N	7	282

13q13.1 Accuracy = 99.8%		Deletion-typing PCR	
		Deletion	≥ 2N
TaqMan CNV assay	Deletion	217	0
	≥ 2N	1	304

Supplementary Data 3. Frequency distributions of genetic markers.

Genetic marker (risk event)	AS-GRS construction set				Independent validation set			
	AS patients (n=285)		Controls (n=363)		AS patients (n=576)		Controls (n=680)	
	Risk	No risk	Risk	No risk	Risk	No risk	Risk	No risk
1q32.2 (deletion)	179	106	107	256	338	238	308	372
2q31.2 (deletion)	185	100	138	225	-	-	-	-
13q13.1 (deletion)	161	124	88	275	303	273	236	444
16p13.3 (deletion)	38	247	17	346	51	525	32	648
rs10865331 (A)	222	63	182	181	402	174	388	292
rs27044 (G)	204	81	246	117	-	-	-	-
rs27434 (A)	208	77	258	105	-	-	-	-
rs27037 (T)	149	136	236	127	-	-	-	-
rs30187 (T)	208	77	259	104	-	-	-	-
rs17482078 (C)	277	8	356	7	-	-	-	-
rs10050860 (C)	285	0	362	1	-	-	-	-

Supplementary Data 4. SNP genotypes for AS-GRS construction set.

Sample_ID	Status	rs10865331	rs27044	rs27434	rs27037	rs30187	rs17482078	rs10050860
AS001	Case	GG	CC	GG	GG	CC	CC	CC
AS003	Case	AG	CG	AG	GG	CT	CC	CC
AS004	Case	AG	CG	AG	GT	CT	CC	CC
AS005	Case	AA	CC	GG	GG	CC	CC	CC
AS006	Case	AA	GG	AA	GT	TT	CC	CC
AS009	Case	AA	GG	AA	GT	TT	CC	CC
AS010	Case	GG	CG	AG	GT	CT	CC	CC
AS011	Case	GG	CG	AG	GT	CT	CC	CC
AS012	Case	AA	CG	AG	GT	CT	CC	CC
AS013	Case	AG	CC	AG	GG	CT	CC	CC
AS014	Case	GG	CC	GG	GG	CC	CC	CC
AS015	Case	AG	CG	AG	GT	CT	CC	CC
AS023	Case	AA	CC	AG	GG	CT	CC	CC
AS025	Case	GG	CG	AG	GT	CT	CC	CC
AS026	Case	GG	CG	AG	GG	CT	CC	CC
AS027	Case	AG	CG	AG	GG	CT	CC	CC
AS028	Case	AA	CG	AA	GT	TT	CC	CC
AS030	Case	GG	CG	AG	GG	CT	CT	CT
AS032	Case	AG	CG	AG	GT	CT	CC	CC
AS034	Case	AA	CG	AG	GT	CT	CC	CC
AS036	Case	AA	CG	AG	GG	CT	CC	CC
AS038	Case	AG	CG	AG	GT	CT	CC	CC
AS040	Case	AG	GG	AA	GG	TT	CC	CC
AS042	Case	GG	CG	AG	GG	CT	CT	CT
AS043	Case	AA	CG	AG	GG	CT	CC	CC
AS045	Case	AG	CG	AG	GT	CT	CC	CC
AS046	Case	AG	CC	GG	GG	CC	CC	CC
AS048	Case	GG	CC	GG	GG	CC	CC	CC
AS049	Case	AG	CG	AG	GG	CT	CC	CC
AS050	Case	GG	CG	AG	GT	CT	CT	CT
AS051	Case	GG	CG	AG	GG	CT	CT	CT
AS052	Case	AG	CG	AG	GG	CT	CC	CC
AS053	Case	AG	GG	AA	TT	TT	CC	CC
AS054	Case	AA	CG	AG	GT	CT	CC	CC
AS058	Case	AG	GG	AA	GT	TT	CC	CC
AS064	Case	AA	GG	AA	TT	TT	CC	CC
AS068	Case	AA	GG	AA	GT	TT	CC	CC
AS070	Case	AG	CG	AG	GG	CT	CC	CC
AS072	Case	AG	CC	GG	GG	CC	CC	CC
AS073	Case	AG	CC	GG	GG	CC	CC	CC
AS075	Case	AA	GG	AA	GT	TT	CC	CC
AS076	Case	AG	CG	AG	GG	CT	CC	CC
AS078	Case	AA	GG	AA	TT	TT	CC	CC
AS080	Case	AG	GG	AA	GT	TT	CC	CC
AS082	Case	AG	CC	GG	GG	CC	CC	CC
AS084	Case	AG	CG	AG	GT	CT	CC	CC
AS086	Case	AG	CG	AG	GT	CT	CC	CC
AS087	Case	AG	CG	AG	GT	CT	CC	CC
AS089	Case	AA	CG	AG	GT	CT	CC	CC
AS090	Case	AG	CC	GG	GG	CC	CC	CC
AS091	Case	GG	CG	AA	GG	TT	CC	CC
AS094	Case	AG	CC	GG	GG	CC	CC	CC
AS096	Case	GG	CG	AG	GT	CT	CC	CC
AS100	Case	AG	CG	AG	GG	CT	CC	CC
AS101	Case	AG	CG	AG	GG	CT	CC	CC
AS102	Case	GG	CC	GG	GG	CC	CC	CC
AS103	Case	AG	CC	GG	GG	CC	CT	CT
AS104	Case	AG	CC	GG	GG	CC	CC	CC
AS108	Case	AG	CG	AG	GT	CT	CC	CC

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AS110	Case	AG	CC	GG	GG	CC	CT	CT
AS112	Case	AA	CG	AG	GT	CT	CC	CC
AS116	Case	AG	GG	AA	TT	TT	CC	CC
AS121	Case	GG	CC	GG	GT	CC	CC	CC
AS125	Case	GG	CC	GG	GG	CC	CC	CC
AS126	Case	AG	GG	AA	TT	TT	CC	CC
AS127	Case	AG	CC	GG	GG	CC	CC	CC
AS128	Case	AG	GG	AA	GT	TT	CC	CC
AS130	Case	AG	CG	AG	GT	CT	CC	CC
AS131	Case	AG	CG	AG	GG	CT	CC	CC
AS132	Case	AG	GG	AA	TT	TT	CC	CC
AS133	Case	AG	GG	AA	TT	TT	CC	CC
AS135	Case	GG	CG	AG	GT	CT	CC	CC
AS137	Case	AG	CC	GG	GT	CC	CC	CC
AS138	Case	AG	GG	AA	GT	TT	CC	CC
AS140	Case	GG	CC	GG	GG	CC	CC	CC
AS143	Case	AG	GG	AA	GT	TT	CC	CC
AS144	Case	AG	CG	AG	GT	CT	CC	CC
AS145	Case	GG	CC	GG	GG	CC	CT	CT
AS146	Case	AG	CG	AG	GG	CT	CC	CC
AS148	Case	AG	CC	GG	GG	CC	CC	CC
AS149	Case	AG	GG	AA	TT	TT	CC	CC
AS150	Case	AA	CG	AG	GG	CT	CC	CC
AS151	Case	AG	CG	GG	GG	CC	CC	CC
AS152	Case	AG	CC	GG	GG	CC	CC	CC
AS153	Case	GG	CG	AG	GG	CT	CC	CC
AS155	Case	AA	CG	AG	GG	CT	CC	CC
AS156	Case	AG	CG	AG	GG	CT	CC	CC
AS157	Case	GG	GG	AA	GT	TT	CC	CC
AS158	Case	AG	CG	AG	GG	CT	CC	CC
AS159	Case	AA	GG	AA	TT	TT	CC	CC
AS162	Case	AA	CG	AG	GT	CT	CC	CC
AS163	Case	AG	CC	GG	GG	CC	CC	CC
AS164	Case	AG	CC	GG	GG	CC	CC	CC
AS166	Case	AA	CG	AG	GT	CT	CC	CC
AS169	Case	AG	GG	AA	GT	TT	CC	CC
AS170	Case	GG	CC	GG	GG	CC	CC	CC
AS171	Case	AG	CC	GG	GG	CC	CC	CC
AS174	Case	AG	CG	AG	GG	CT	CC	CC
AS176	Case	GG	CG	AG	GT	CT	CC	CC
AS179	Case	AG	CC	GG	GG	CC	CT	CT
AS182	Case	AG	CC	GG	GG	CC	CC	CC
AS183	Case	AG	CG	AG	GT	CT	CC	CC
AS184	Case	AA	CG	AG	GT	CT	CC	CC
AS185	Case	AG	GG	AA	TT	TT	CC	CC
AS186	Case	AG	GG	AA	GG	TT	CC	CC
AS188	Case	AA	CG	AG	GG	CT	CC	CC
AS189	Case	AG	CG	AG	GT	CT	CC	CC
AS191	Case	AG	CG	AG	GT	CT	CC	CC
AS192	Case	GG	CC	GG	GG	CC	CC	CC
AS198	Case	AG	CC	GG	GG	CC	CC	CC
AS199	Case	AG	CC	GG	GG	CC	CC	CC
AS200	Case	AG	CC	GG	GG	CC	CC	CC
AS201	Case	AA	CG	AG	GT	CT	CC	CC
AS204	Case	AA	CG	AA	GT	TT	CC	CC
AS206	Case	GG	GG	AA	GG	TT	CC	CC
AS207	Case	AA	CC	GG	GG	CC	CC	CC
AS209	Case	GG	GG	AA	GT	TT	CC	CC
AS211	Case	AA	GG	AA	TT	TT	CC	CC
AS213	Case	GG	CG	AG	GG	CT	CC	CC
AS215	Case	AA	CC	GG	GG	CC	CC	CC
AS216	Case	AA	GG	AA	GT	TT	CC	CC

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AS218	Case	AG	CG	AG	GG	CT	CC	CC
AS220	Case	AG	GG	AA	TT	TT	CC	CC
AS221	Case	GG	CC	GG	GG	CC	CC	CC
AS222	Case	AG	CC	GG	GG	CC	CC	CC
AS226	Case	AG	CG	AG	GT	CT	CC	CC
AS227	Case	GG	CG	AG	TT	CT	CC	CC
AS237	Case	AG	CG	AG	GG	CT	CC	CC
AS239	Case	AA	CG	AG	GT	CT	CC	CC
AS242	Case	AG	GG	AA	TT	TT	CC	CC
AS245	Case	GG	CG	AG	GT	CT	CC	CC
AS247	Case	AA	CG	AG	GT	CT	CC	CC
AS248	Case	AA	GG	AA	GT	TT	CC	CC
AS249	Case	AA	CG	AG	GT	CT	CC	CC
AS250	Case	GG	CC	GG	GG	CC	CC	CC
AS251	Case	AA	CC	GG	GG	CC	CC	CC
AS252	Case	AG	CG	AG	GT	CT	CC	CC
AS253	Case	AG	CG	AG	GG	CT	CT	CT
AS254	Case	AG	GG	AA	TT	TT	CC	CC
AS256	Case	AG	CG	AG	GT	CT	CC	CC
AS257	Case	AG	CG	AG	GT	CT	CC	CC
AS258	Case	AG	GG	AA	GT	TT	CC	CC
AS259	Case	AA	CG	AG	GT	CT	CC	CC
AS261	Case	AG	CC	GG	GG	CC	CC	CC
AS266	Case	GG	CC	GG	GG	CC	CC	CC
AS267	Case	AG	CG	AG	GT	CT	CC	CC
AS268	Case	AA	CG	AG	GG	CT	CC	CC
AS269	Case	AA	CG	AG	GT	CT	CC	CC
AS272	Case	AA	CG	AG	GT	CT	CC	CC
AS273	Case	AG	CC	GG	GG	CC	CC	CC
AS275	Case	GG	CG	AG	GT	CT	CC	CC
AS278	Case	AG	CG	AG	GT	CT	CC	CC
AS279	Case	AG	CG	AG	GG	CT	CC	CC
AS280	Case	AG	CC	GG	GG	CC	CC	CC
AS283	Case	AA	GG	AA	GT	TT	CC	CC
AS291	Case	AA	CG	AG	GT	CT	CC	CC
AS292	Case	AA	CG	AG	GT	CT	CC	CC
AS293	Case	AG	CG	AG	GG	CT	CC	CC
AS296	Case	AA	CC	GG	GG	CC	CC	CC
AS297	Case	AG	CG	AG	GG	CT	CC	CC
AS300	Case	AG	CG	AG	GG	CT	CC	CC
AS308	Case	AG	GG	AA	TT	TT	CC	CC
AS309	Case	AA	CG	AG	GG	CT	CT	CT
AS310	Case	AG	CG	AG	GT	CT	CC	CC
AS312	Case	GG	CG	AG	GG	CT	CC	CC
AS313	Case	AG	CG	AG	GT	CT	CC	CC
AS314	Case	AG	GG	AA	GT	TT	CC	CC
AS315	Case	AG	CG	AG	GG	CT	CC	CC
AS316	Case	AG	CG	AG	GG	CT	CC	CC
AS318	Case	GG	CG	AG	GT	CT	CC	CC
AS321	Case	AG	CG	AG	GT	CT	CC	CC
AS326	Case	AA	CC	GG	GG	CC	CC	CC
AS327	Case	GG	CG	AG	GT	CT	CC	CC
AS329	Case	AG	CG	AG	GG	CT	CT	CT
AS342	Case	AG	CG	AG	GG	CT	CC	CC
AS343	Case	AG	CG	AG	GT	CT	CC	CC
AS346	Case	AG	CC	GG	GG	CC	CC	CC
AS348	Case	GG	CC	GG	GG	CC	CT	CT
AS349	Case	AG	CC	GG	GG	CC	CC	CC
AS351	Case	AG	CG	AG	GT	CT	CC	CC
AS352	Case	AG	CG	AG	GG	CT	CC	CC
AS353	Case	AG	CG	AG	GG	CT	CT	CT
AS354	Case	GG	CG	AG	GT	CT	CC	CC

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AS357	Case	AG	GG	AA	GT	TT	CC	CC
AS359	Case	AG	GG	AA	TT	TT	CC	CC
AS360	Case	AG	CG	AG	GT	CT	CC	CC
AS361	Case	AG	GG	AA	GT	TT	CC	CC
AS363	Case	AA	CG	AG	GT	CT	CC	CC
AS364	Case	GG	CG	AG	GT	CT	CC	CC
AS367	Case	GG	CG	AG	GT	CT	CC	CC
AS368	Case	AG	CC	GG	GG	CC	CC	CC
AS370	Case	GG	CC	AG	GG	CT	CC	CC
AS371	Case	AG	CG	AG	GT	CT	CC	CC
AS372	Case	AG	CG	AG	GT	CT	CC	CC
AS373	Case	AG	CG	AG	GG	CT	CT	CT
AS374	Case	GG	CG	AG	GG	CT	CC	CC
AS375	Case	AG	CG	AG	GG	CT	CC	CC
AS376	Case	GG	CG	AG	GG	CT	CC	CC
AS377	Case	AG	CC	GG	GG	CC	CC	CC
AS384	Case	AG	CC	GG	GG	CC	CC	CC
AS385	Case	AA	CG	AG	GG	CT	CC	CC
AS387	Case	AG	GG	AA	GT	TT	CC	CC
AS388	Case	AG	CG	AA	GT	TT	CC	CC
AS392	Case	GG	CC	GG	GG	CC	CC	CC
AS394	Case	AA	CG	AG	GT	CT	CC	CC
AS395	Case	AA	CC	GG	GG	CC	CC	CC
AS398	Case	GG	CC	GG	GG	CC	CC	CC
AS399	Case	GG	CG	AG	GT	CT	CC	CC
AS400	Case	AG	CG	AG	GT	CT	CC	CC
ASR001	Case	AG	CC	GG	GG	CC	CC	CC
ASR002	Case	AG	CG	AG	GG	CT	CC	CC
ASR003	Case	GG	CC	GG	GG	CC	TT	CT
ASR004	Case	GG	CG	AG	GT	CT	CC	CC
ASR005	Case	GG	CG	AG	GT	CT	CC	CC
ASR008	Case	AA	GG	AA	GT	TT	CC	CC
ASR009	Case	AG	GG	AA	GG	TT	CC	CC
ASR010	Case	AG	CC	AG	GG	CT	CC	CC
ASR012	Case	AA	CG	AG	GG	CT	CC	CC
ASR013	Case	AG	GG	AA	TT	TT	CC	CC
ASR014	Case	GG	CC	GG	GG	CC	CC	CC
ASR015	Case	GG	CC	GG	GG	CC	CC	CC
ASR017	Case	AA	CG	AG	GT	CT	CC	CC
ASR018	Case	AG	GG	AA	TT	TT	CC	CC
ASR019	Case	AG	CC	GG	GG	CC	CC	CC
ASR025	Case	AG	GG	AA	TT	TT	CC	CC
ASR027	Case	AA	GG	AA	TT	TT	CC	CC
ASR029	Case	AG	GG	AA	TT	TT	CC	CC
ASR031	Case	AG	CC	GG	GG	CC	CC	CC
ASR032	Case	AG	GG	AA	TT	TT	CC	CC
ASR034	Case	AG	CC	AG	GG	CT	CC	CC
ASR041	Case	AG	CG	AG	GG	CT	CC	CC
ASR042	Case	AG	CG	AG	GT	CT	CC	CC
ASR044	Case	AA	GG	AA	GT	TT	CC	CC
ASR046	Case	AA	GG	AA	GT	TT	CC	CC
ASR047	Case	GG	CC	GG	GG	CC	CC	CC
ASR048	Case	AA	CG	AG	GT	CT	CC	CC
ASR054	Case	GG	CC	GG	GG	CC	CC	CC
ASR058	Case	AG	CC	GG	GG	CC	TT	CT
ASR059	Case	AG	CG	AG	GT	CT	CC	CC
ASR063	Case	GG	CG	AG	GT	CT	TT	CT
ASR067	Case	AG	CG	AG	GT	CT	CC	CC
ASR069	Case	AA	CG	AG	GT	CT	TT	CT
ASR071	Case	GG	CC	GG	GG	CC	CC	CC
ASR072	Case	AG	CC	GG	GG	CC	CC	CC
ASR077	Case	GG	CC	GG	GG	CC	CC	CC

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ASR078	Case	AG	CC	GG	GG	CC	TT	CT
ASR079	Case	AG	CG	AG	GT	CT	TT	CT
ASR080	Case	AG	CG	AG	GG	CT	TT	CT
ASR081	Case	AG	GG	AA	TT	TT	CC	CC
ASR082	Case	AG	CG	AG	GT	CT	CC	CC
ASR083	Case	AG	CC	GG	GG	CC	CC	CC
ASR084	Case	AA	CC	GG	GG	CC	CC	CC
ASR085	Case	AG	CG	AG	GT	CT	CC	CC
ASR086	Case	AG	CG	AG	GT	CT	CC	CC
ASR088	Case	AG	CG	AG	GT	CT	CC	CC
ASR090	Case	AG	CG	AG	GT	CT	CC	CC
ASR091	Case	AA	CG	AG	GT	CT	CC	CC
ASR092	Case	AG	GG	AA	TT	TT	CC	CC
ASR096	Case	AG	GG	AA	GT	TT	CC	CC
ASR099	Case	GG	CG	AG	GG	CT	TT	CT
ASR101	Case	AG	CG	AG	GT	CT	CC	CC
ASR102	Case	AA	CG	AG	GT	CT	CC	CC
ASR105	Case	GG	CC	GG	GG	CC	CC	CC
ASR106	Case	AG	CG	AA	GT	TT	CC	CC
ASR107	Case	AG	CC	GG	GG	CC	CC	CC
ASR109	Case	AG	CG	AG	GG	CT	CC	CC
ASR111	Case	GG	CG	AA	GT	TT	CC	CC
ASR112	Case	AG	GG	AA	GT	TT	CC	CC
ASR113	Case	GG	CC	GG	GG	CC	CC	CC
ASR118	Case	GG	CG	AG	GT	CT	CC	CC
ASR120	Case	AG	CC	GG	GG	CC	CC	CC
ASR122	Case	AG	GG	AA	GT	TT	CC	CC
ASR123	Case	AG	GG	AA	TT	TT	CC	CC
ASR126	Case	GG	CG	AA	GT	TT	CC	CC
ASR131	Case	AG	GG	AA	GT	TT	CC	CC
ASR134	Case	AG	GG	AA	TT	TT	CC	CC
ASR136	Case	AG	GG	AA	TT	TT	CC	CC
ASR137	Case	GG	CC	GG	GT	CC	CC	CC
ASR145	Case	AG	CC	GG	GG	CC	CC	CC
ASR147	Case	AG	CC	GG	GG	CC	CC	CC
ASR156	Case	AG	CC	GG	GG	CC	CC	CC
ASR159	Case	AG	GG	AA	TT	TT	CC	CC
ASR162	Case	AG	GG	AA	GG	TT	CC	CC
ASR165	Case	AG	GG	AA	GT	TT	CC	CC
ASR166	Case	AG	CG	AG	GT	CT	CC	CC
ASC002	Control	AG	CG	AG	GT	CT	CC	CC
ASC022	Control	GG	CG	AA	GT	TT	CC	CC
ASC035	Control	AG	CG	AG	GT	CT	CC	CC
ASC041	Control	AG	CC	GG	GG	CC	CC	CC
ASC047	Control	AG	GG	AA	GT	TT	CC	CC
ASC055	Control	GG	CG	AG	GT	CT	CC	CC
ASC060	Control	GG	CG	AG	GT	CT	CC	CC
ASC063	Control	AG	CG	AG	GT	CT	CC	CC
ASC064	Control	AG	CC	GG	GG	CC	CC	CC
ASC065	Control	AG	CG	AG	GT	CT	CC	CC
ASC066	Control	GG	GG	AA	TT	TT	CC	CC
ASC071	Control	GG	CG	AG	GT	CT	CC	CC
ASC072	Control	GG	GG	AA	GT	TT	CC	CC
ASC074	Control	GG	CG	AG	GT	CT	CC	CC
ASC081	Control	GG	CG	AG	GT	CT	CC	CC
ASC087	Control	GG	CC	AG	GG	CT	CC	CC
ASC089	Control	GG	CG	AG	GT	CT	CC	CC
ASC096	Control	AG	CG	AG	GT	CT	CC	CC
ASC097	Control	GG	CG	AG	GT	CT	CC	CC
ASC099	Control	GG	CC	GG	GG	CC	CC	CC
ASC1001	Control	AG	GG	AA	TT	TT	CC	CC
ASC1005	Control	AG	CG	AG	GG	CT	CC	CC

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ASC101	Control	AG	CG	AG	GT	CT	CC	CC
ASC1011	Control	AG	CG	AG	GT	CT	CC	CC
ASC1012	Control	AA	CC	GG	GT	CC	CC	CC
ASC1015	Control	AG	GG	AA	GT	TT	CC	CC
ASC1019	Control	AG	CG	AG	GT	CT	CC	CC
ASC104	Control	GG	CG	AG	GT	CT	CT	CT
ASC105	Control	AA	CC	GG	GG	CC	CC	CC
ASC107	Control	AG	CG	AG	GT	CT	CC	CC
ASC110	Control	AG	CG	AG	GT	CT	CC	CC
ASC114	Control	AG	CG	AG	GT	CT	CC	CC
ASC122	Control	AG	CC	GG	GG	CC	CC	CC
ASC129	Control	GG	GG	AA	TT	TT	CC	CC
ASC131	Control	GG	CC	GG	GT	CC	CC	CC
ASC133	Control	AG	CC	GG	GG	CC	CC	CC
ASC135	Control	GG	CC	GG	GG	CC	CC	CC
ASC136	Control	GG	GG	AA	GT	TT	CC	CC
ASC140	Control	AG	CC	GG	GG	CC	CC	CC
ASC143	Control	GG	CG	AG	GG	CT	CC	CC
ASC145	Control	AA	CC	GG	GG	CC	CC	CC
ASC146	Control	AG	CG	AG	GT	CT	CC	CC
ASC150	Control	GG	CC	GG	GG	CC	CC	CC
ASC154	Control	AG	CG	AG	GT	CT	CT	CT
ASC155	Control	GG	CC	GG	GG	CC	CC	CC
ASC157	Control	AG	GG	AA	TT	TT	CC	CC
ASC158	Control	GG	CG	AG	GT	CT	CT	CT
ASC160	Control	AA	GG	AA	GT	TT	CC	CC
ASC162	Control	GG	CC	GG	GG	CC	CC	CC
ASC163	Control	GG	CG	AG	GT	CT	CC	CC
ASC165	Control	AG	CG	AG	GT	CT	CC	CC
ASC176	Control	GG	CG	AG	GT	CT	CC	CC
ASC182	Control	AG	CG	AG	GT	CT	CT	CT
ASC184	Control	AG	CG	AG	GT	CT	CC	CC
ASC185	Control	GG	GG	AA	TT	TT	CC	CC
ASC186	Control	GG	CC	GG	GG	CC	CC	CC
ASC191	Control	GG	CC	GG	GG	CC	CC	CC
ASC201	Control	GG	CG	AG	GT	CT	CC	CC
ASC209	Control	AG	GG	AA	GT	TT	CC	CC
ASC214	Control	GG	CG	AG	GT	CT	CC	CC
ASC217	Control	AG	CC	GG	GG	CC	CC	CC
ASC239	Control	AG	CG	AG	GT	CT	CC	CC
ASC249	Control	GG	GG	AA	GT	TT	CC	CC
ASC259	Control	GG	CC	AG	GG	CT	CC	CC
ASC265	Control	GG	CC	GG	GG	CC	CC	CC
ASC270	Control	AG	CC	GG	GG	CC	CC	CC
ASC273	Control	GG	CG	AG	GT	CT	CC	CC
ASC276	Control	AG	GG	AA	TT	TT	CC	CC
ASC277	Control	GG	CC	GG	GG	CC	CC	CC
ASC281	Control	GG	CC	AG	GG	CT	CC	CC
ASC286	Control	GG	CC	GG	GG	CC	CC	CC
ASC292	Control	GG	CG	AG	GG	CT	CC	CC
ASC295	Control	GG	CC	GG	GG	CC	CC	CC
ASC307	Control	GG	GG	AA	GT	TT	CC	CC
ASC309	Control	AG	GG	AA	TT	TT	CC	CC
ASC310	Control	AA	GG	AA	GT	TT	CC	CC
ASC312	Control	AG	CG	AG	GT	CT	CC	CC
ASC313	Control	AG	CG	AG	GT	CT	CC	CC
ASC321	Control	AG	GG	AA	GT	TT	CC	CC
ASC330	Control	AA	CC	GG	GG	CC	CT	CT
ASC333	Control	GG	CC	GG	GG	CC	CC	CC
ASC334	Control	AG	CG	AG	GT	CT	CC	CC
ASC335	Control	AG	CC	GG	GG	CC	CC	CC
ASC338	Control	AG	CC	AG	GG	CT	CC	CC

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ASC349	Control	GG	CC	GG	GG	CC	CC	CC
ASC354	Control	AG	GG	AA	TT	TT	CC	CC
ASC361	Control	GG	GG	AA	TT	TT	CC	CC
ASC378	Control	GG	CC	GG	GG	CC	CC	CC
ASC381	Control	GG	CC	GG	GG	CC	CC	CC
ASC384	Control	AA	CG	AG	GT	CT	CT	CT
ASC387	Control	AG	CC	GG	GG	CC	CC	CC
ASC391	Control	GG	CC	GG	GG	CC	CC	CC
ASC393	Control	AG	CG	AG	GT	CT	CC	CC
ASC400	Control	GG	GG	AA	TT	TT	CC	CC
ASC404	Control	GG	CG	AG	GT	CT	CC	CC
ASC409	Control	AG	GG	AA	GT	TT	CC	CC
ASC415	Control	AG	GG	AA	GT	TT	CC	CC
ASC417	Control	AG	CG	AA	TT	TT	CC	CC
ASC422	Control	AG	CG	AG	GT	CT	CC	CC
ASC424	Control	AG	CC	GG	GG	CC	CC	CC
ASC433	Control	GG	CC	GG	GG	CC	CC	CC
ASC439	Control	AG	GG	AA	TT	TT	CC	CC
ASC444	Control	GG	CC	GG	GG	CC	CC	CC
ASC448	Control	GG	CG	AG	GT	CT	CT	CT
ASC450	Control	AG	CG	AG	GT	CT	CC	CC
ASC452	Control	AG	CG	AG	GT	CT	CC	CC
ASC453	Control	AG	CG	AG	GT	CT	CC	CC
ASC456	Control	GG	CC	AG	GG	CT	CC	CC
ASC457	Control	GG	GG	AA	TT	TT	CC	CC
ASC458	Control	GG	CC	GG	GG	CC	CC	CC
ASC460	Control	GG	GG	AA	GT	TT	CC	CC
ASC464	Control	AG	CG	AG	GT	CT	CC	CC
ASC466	Control	GG	CG	AG	GT	CT	CC	CC
ASC467	Control	GG	GG	AA	TT	TT	CC	CC
ASC468	Control	AG	CG	AG	GG	CT	CC	CC
ASC471	Control	AG	CC	GG	GG	CC	CC	CC
ASC479	Control	AG	CC	GG	GG	CC	CC	CC
ASC480	Control	GG	CG	AG	GT	CT	CC	CC
ASC481	Control	GG	CG	AG	GT	CT	CC	CC
ASC482	Control	AG	CC	GG	GG	CC	CC	CC
ASC485	Control	GG	CG	AG	GT	CT	CC	CC
ASC486	Control	GG	CC	GG	GG	CC	CC	CC
ASC489	Control	AG	CG	AG	GT	CT	CC	CC
ASC494	Control	GG	CG	AA	GT	TT	CC	CC
ASC495	Control	GG	CG	AG	TT	CT	CC	CC
ASC496	Control	GG	CG	AG	GT	CT	CC	CC
ASC497	Control	GG	CC	GG	GG	CC	CC	CC
ASC499	Control	GG	CC	GG	GG	CC	CC	CC
ASC507	Control	AA	GG	AA	TT	TT	CC	CC
ASC512	Control	AG	CG	AG	GT	CT	CC	CC
ASC515	Control	GG	GG	AA	TT	TT	CC	CC
ASC516	Control	AG	CC	GG	GG	CC	CC	CC
ASC519	Control	AG	CG	AG	GT	CT	CC	CC
ASC521	Control	GG	CC	GG	GG	CC	CT	CT
ASC524	Control	GG	CG	AG	GT	CT	CT	CT
ASC535	Control	GG	CG	AG	GT	CT	CC	CC
ASC536	Control	AG	CG	AG	GT	CT	CC	CC
ASC543	Control	AG	GG	AA	TT	TT	CC	CC
ASC545	Control	GG	CC	GG	GG	CC	CC	CC
ASC553	Control	GG	CG	AG	GT	CT	CC	CC
ASC555	Control	AG	CC	GG	GG	CC	CC	CC
ASC558	Control	AG	CG	AG	GT	CT	CC	CC
ASC560	Control	AG	CG	AG	GT	CT	CT	CT
ASC563	Control	GG	GG	AA	TT	TT	CC	CC
ASC570	Control	AG	CC	GG	GG	CC	CC	CC
ASC571	Control	AA	CG	AG	GT	CT	CC	CC

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ASC572	Control	AG	CC	GG	GG	CC	CC	CC
ASC575	Control	AG	GG	AA	TT	TT	CC	CC
ASC581	Control	AA	GG	AA	GT	TT	CC	CC
ASC587	Control	AG	CG	AG	GT	CT	CC	CC
ASC594	Control	GG	GG	AA	TT	TT	CC	CC
ASC595	Control	GG	CG	AA	GT	TT	CC	CC
ASC597	Control	GG	CG	AG	GG	CT	CC	CC
ASC598	Control	AG	GG	AA	GT	TT	CC	CC
ASC599	Control	AG	GG	AA	TT	TT	CC	CC
ASC600	Control	GG	CG	AG	GT	CT	CC	CC
ASC603	Control	GG	CG	AG	GT	CT	CC	CC
ASC604	Control	GG	CG	AG	GT	CT	CC	CC
ASC620	Control	AA	GG	AA	GG	TT	CC	CC
ASC628	Control	AA	CG	AG	GT	CT	CC	CC
ASC631	Control	GG	CC	GG	GG	CC	CC	CC
ASC632	Control	GG	CG	AG	GT	CT	CT	CT
ASC638	Control	AG	CC	GG	GG	CC	CT	CT
ASC640	Control	GG	GG	AA	GT	TT	CC	CC
ASC643	Control	GG	CG	AG	GT	CT	CC	CC
ASC646	Control	GG	CC	GG	GG	CC	CC	CC
ASC647	Control	AG	CG	AG	GG	CT	CC	CC
ASC649	Control	AG	CC	GG	GG	CC	CC	CC
ASC651	Control	AG	CG	AG	GT	CT	CC	CC
ASC655	Control	AG	CC	GG	GG	CC	CC	CC
ASC656	Control	GG	GG	AA	TT	TT	CC	CC
ASC658	Control	AG	CG	AG	GT	CT	CC	CC
ASC661	Control	GG	CC	GG	GG	CC	CC	CC
ASC664	Control	AA	CG	AG	GT	CT	CC	CC
ASC665	Control	AA	CG	AG	GT	CT	CC	CC
ASC668	Control	GG	CG	AG	GT	CT	CC	CC
ASC672	Control	AG	CG	AG	GG	CT	CC	CC
ASC673	Control	AG	CG	AG	GT	CT	CC	CC
ASC674	Control	AG	GG	AA	GT	TT	CC	CC
ASC675	Control	AG	GG	AA	GT	TT	CC	CC
ASC676	Control	GG	CC	GG	GG	CC	CT	CT
ASC678	Control	GG	CG	AG	GT	CT	CT	CT
ASC679	Control	GG	CG	AG	GT	CT	CC	CC
ASC687	Control	GG	CG	AG	GT	CT	CC	CC
ASC689	Control	GG	CG	AG	GT	CT	CC	CC
ASC703	Control	AA	GG	AA	GT	TT	CC	CC
ASC711	Control	AG	CC	AG	GG	CC	CC	CC
ASC712	Control	GG	CC	GG	GG	TT	TT	TT
ASC717	Control	GG	CC	GG	GG	CC	CC	CC
ASC718	Control	AA	GG	AA	GT	TT	CC	CC
ASC722	Control	GG	CG	AA	GT	CC	CC	CC
ASC725	Control	GG	GG	AA	GT	TT	CC	CC
ASC728	Control	GG	CC	GG	GG	CC	CC	CC
ASC730	Control	AG	CC	GG	GG	CT	CC	CC
ASC735	Control	AG	CC	AG	GG	CT	CC	CC
ASC744	Control	AG	CC	GG	GG	CT	CC	CC
ASC746	Control	GG	GG	AA	TT	CT	CC	CC
ASC757	Control	AG	CC	GG	GG	CC	CC	CC
ASC759	Control	AA	GG	AA	TT	TT	CC	CC
ASC769	Control	AG	GG	AA	TT	TT	CC	CC
ASC772	Control	GG	CG	AG	GT	CT	CC	CC
ASC782	Control	GG	CG	AG	GT	CT	CC	CC
ASC789	Control	GG	GG	AA	GT	TT	CC	CC
ASC790	Control	AG	GG	AA	TT	TT	CC	CC
ASC791	Control	AG	CG	AG	GT	CT	CC	CC
ASC792	Control	GG	CG	AG	TT	CT	CC	CC
ASC793	Control	AG	CG	AG	GT	CT	CC	CC
ASC795	Control	AG	CC	GG	GG	CC	CC	CC

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ASC796	Control	GG	CG	AG	GT	CT	CC	CC
ASC798	Control	GG	CC	GG	GG	CC	CC	CC
ASC803	Control	AG	CG	AG	GT	CT	CC	CC
ASC807	Control	AG	CG	AA	GT	TT	CC	CC
ASC808	Control	GG	CG	AG	GT	CT	CC	CC
ASC811	Control	AG	GG	AA	GT	TT	CC	CC
ASC812	Control	GG	GG	AA	TT	TT	CC	CC
ASC813	Control	GG	GG	AA	TT	TT	CC	CC
ASC815	Control	AG	CG	AG	GT	CT	CC	CC
ASC816	Control	GG	CG	AG	GT	CT	CC	CC
ASC828	Control	AG	CG	AG	GT	CT	CC	CC
ASC830	Control	GG	GG	AA	GT	TT	CC	CC
ASC833	Control	AA	GG	AA	GT	TT	CC	CC
ASC835	Control	AG	GG	AA	TT	TT	CC	CC
ASC836	Control	AG	CC	GG	GG	CC	CC	CC
ASC838	Control	GG	GG	AA	TT	TT	CC	CC
ASC841	Control	AA	CG	AG	GT	CT	CC	CC
ASC844	Control	AG	CC	GG	GG	CC	CC	CC
ASC846	Control	AG	CG	AG	GT	CT	CC	CC
ASC848	Control	GG	CC	GG	GG	CC	CC	CC
ASC852	Control	AA	GG	AA	GT	TT	CC	CC
ASC853	Control	GG	CG	AA	GT	TT	CC	CC
ASC858	Control	GG	CC	GG	GG	CC	CC	CC
ASC864	Control	AG	CC	GG	GG	CC	TT	CT
ASC869	Control	AG	CC	GG	GG	CC	CC	CC
ASC873	Control	GG	CC	GG	GG	CC	CC	CC
ASC890	Control	GG	CC	GG	GG	CC	CC	CC
ASC892	Control	AA	CG	AG	GT	CT	CC	CC
ASC894	Control	AG	CG	AG	GT	CT	CC	CC
ASC896	Control	AG	CC	GG	GG	CC	CC	CC
ASC899	Control	AG	CG	AG	GT	CT	TT	CT
ASC903	Control	AG	GG	AA	GT	TT	CC	CC
ASC904	Control	GG	CC	GG	GG	CC	CC	CC
ASC909	Control	AG	CG	AG	GT	CT	CC	CC
ASC915	Control	AG	CC	AG	GG	CT	CC	CC
ASC918	Control	GG	CG	AG	GG	CT	CC	CC
ASC919	Control	GG	CG	AG	GT	CT	CC	CC
ASC924	Control	GG	GG	AA	GT	TT	CC	CC
ASC930	Control	AA	CC	GG	GG	CC	CC	CC
ASC938	Control	GG	CG	AG	GT	CT	TT	CT
ASC941	Control	AG	CG	AG	GT	CT	CC	CC
ASC943	Control	AG	CG	AG	GT	CT	CC	CC
ASC944	Control	AG	CG	AG	GT	CT	CC	CC
ASC945	Control	AG	GG	AA	GT	TT	CC	CC
ASC950	Control	GG	CG	AG	GT	CT	CC	CC
ASC959	Control	AG	GG	AA	TT	TT	CC	CC
ASC962	Control	AG	CG	AG	GT	CT	CC	CC
ASC966	Control	GG	CG	AG	GT	CT	CC	CC
ASC969	Control	AG	CG	AG	GT	CT	CC	CC
ASC970	Control	GG	CC	GG	GG	CC	CC	CC
ASC972	Control	AG	CC	GG	GG	CC	CC	CC
ASC984	Control	GG	GG	AA	TT	TT	CC	CC
ASC988	Control	GG	GG	AA	GT	TT	CC	CC
ASC989	Control	GG	CC	GG	GG	CC	CC	CC
ASC992	Control	GG	CC	GG	GG	CC	CC	CC
ASC995	Control	GG	CC	GG	GG	CC	TT	CT
ASC996	Control	GG	CC	AG	GG	CT	CC	CC
PA008	Control	GG	CG	AA	GT	TT	CC	CC
PA009	Control	GG	CG	AG	GT	CT	CC	CC
PA010	Control	AG	GG	AA	GT	TT	CC	CC
PA015	Control	GG	CC	GG	GG	CC	CC	CC
PA016	Control	GG	CG	AG	GT	CT	CC	CC

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PA017	Control	GG	CG	AG	GT	CT	CC	CC
PA019	Control	AG	GG	AA	TT	TT	CC	CC
PA022	Control	AG	GG	AA	TT	TT	CC	CC
PA025	Control	GG	CG	AG	GT	CT	CC	CC
PA026	Control	GG	CC	GG	GG	CC	CC	CC
PA027	Control	AG	CG	AG	GT	CT	CC	CC
PA028	Control	GG	CC	GG	GG	CC	CC	CC
PA031	Control	AG	CG	AA	GG	TT	CC	CC
PA033	Control	GG	GG	AA	GT	TT	CC	CC
PA034	Control	AG	CG	AG	GT	CT	CC	CC
PA035	Control	GG	CC	GG	GG	CC	CC	CC
PA037	Control	GG	CG	AG	GT	CT	CC	CC
PA040	Control	AG	CC	GG	GG	CC	CC	CC
PA041	Control	GG	CC	GG	GG	CC	CC	CC
PA043	Control	AG	CG	AG	GT	CT	CC	CC
PA045	Control	GG	GG	AA	GT	TT	CC	CC
PA081	Control	AG	CG	AA	GT	TT	CC	CC
PA082	Control	AA	GG	AA	TT	TT	CC	CC
PA084	Control	GG	CG	AG	GT	CT	CC	CC
PA086	Control	GG	CG	AG	GT	CT	TT	CT
PA087	Control	GG	GG	AA	TT	TT	CC	CC
PA089	Control	AA	GG	AA	TT	TT	CC	CC
PA090	Control	AG	CG	AG	GT	CT	CC	CC
PA091	Control	GG	CG	AG	GT	CT	CC	CC
PA094	Control	GG	CG	AG	GT	CT	CC	CC
PA095	Control	GG	CC	GG	GG	CC	CC	CC
PA096	Control	GG	CC	GG	GG	CC	TT	CT
PA097	Control	AA	CC	GG	GG	CC	CC	CC
PA100	Control	AG	CG	AG	GT	CT	CC	CC
PA101	Control	AA	CG	AG	GT	CT	CC	CC
PA104	Control	AG	CC	GG	GG	CC	CC	CC
PA108	Control	GG	GG	AA	GT	TT	CC	CC
PA109	Control	GG	CG	AG	GT	CT	CC	CC
PA111	Control	GG	GG	AA	TT	TT	CC	CC
PA113	Control	AG	CG	AG	TT	CT	CC	CC
PA114	Control	GG	CG	AG	GT	CT	CC	CC
PA115	Control	AG	CG	AG	GT	CT	CC	CC
PA116	Control	GG	CC	GG	GG	CC	CC	CC
PA118	Control	GG	CG	AG	GT	CT	CC	CC
PA119	Control	AA	CC	GG	GG	CC	CC	CC
PA125	Control	GG	GG	AA	TT	TT	CC	CC
PA128	Control	AA	CG	AG	GT	CT	CC	CC
PA142	Control	GG	CG	AG	GG	CT	CC	CC
PA472	Control	GG	GG	AA	GT	TT	CC	CC
PA480	Control	AG	CG	AG	GT	CT	CC	CC
PA482	Control	AA	CG	AG	GT	CT	CC	CC
PA484	Control	AG	CG	AG	GT	CT	CC	CC
PA485	Control	GG	GG	AA	GT	TT	CC	CC
PA486	Control	GG	GG	AA	GT	TT	CC	CC
PA490	Control	AG	CG	AG	GT	CT	CC	CC
PA492	Control	AA	CG	AG	GT	CT	CC	CC
PA494	Control	AG	CG	AG	GT	CT	CC	CC
PA495	Control	GG	GG	AA	TT	TT	CC	CC
PA497	Control	GG	CC	AG	GG	CT	CC	CC
PA498	Control	AG	CC	GG	GG	CC	CC	CC
RC121	Control	GG	GG	AA	TT	TT	CC	CC
RC126	Control	GG	CG	AG	GT	CT	CC	CC
RC130	Control	AG	GG	AA	GT	TT	CC	CC
RC134	Control	AG	CC	GG	GG	CC	CC	CC
RC160	Control	AG	CC	GG	GG	CC	CC	CC
RC167	Control	AG	CC	GG	GG	CC	CC	CC
RC181	Control	AG	GG	AA	GT	TT	CC	CC

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RC186	Control	AG	GG	AA	GT	TT	CC	CC
RC200	Control	GG	CG	AG	GT	CT	CC	CC
RC208	Control	AG	CC	AG	GG	CT	CC	CC
RC258	Control	GG	CG	AG	GT	CT	CC	CC
RC259	Control	AG	CG	AG	GT	CT	CT	CT
RC262	Control	AG	GG	AA	GT	TT	CC	CC
RC265	Control	GG	CG	AG	GT	CT	CC	CC
RC273	Control	AG	CG	AG	GT	CT	CC	CC
RC298	Control	AG	CC	GG	GG	CC	CC	CC
RC300	Control	GG	CC	GG	GG	CC	CC	CC
RC317	Control	GG	CC	GG	GG	CC	CC	CC
RC336	Control	GG	CC	GG	GG	CC	CC	CC
RC337	Control	GG	CG	AG	GT	CT	CC	CC
RC338	Control	AG	CG	AG	GT	CT	CC	CC
RC373	Control	GG	GG	AA	GT	TT	CC	CC
RC385	Control	GG	CC	GG	GG	CC	CC	CC
RC412	Control	GG	CC	GG	GG	CC	CT	CT
RC450	Control	GG	CG	AG	GT	CT	CC	CC
RC466	Control	AG	CC	GG	GG	CC	CC	CC
RC529	Control	GG	CG	AG	GG	CT	CC	CC
RC534	Control	GG	CG	AG	GT	CT	CC	CC
RC567	Control	AG	CG	AG	GT	CT	CC	CC
RC588	Control	GG	CC	AG	GG	CT	CC	CC
RC606	Control	GG	CC	GG	GG	CC	CC	CC
RC611	Control	GG	GG	AA	GT	TT	CC	CC
RC699	Control	AG	CC	GG	GG	CC	CC	CC
SLE234	Control	GG	GG	AA	TT	TT	CC	CC
SLE235	Control	GG	CC	GG	GG	CC	CC	CC
SLE240	Control	AG	CG	AG	GT	CT	CC	CC
SLE241	Control	AG	CC	GG	GG	CC	CC	CC
SLE243	Control	AA	CC	GG	GG	CC	CT	CT

Supplementary Data 5. Associations of seven SNPs in the AS-GRS construction set and independent validation set.

SNP	Minor allele	Major allele	Model*	AS-GRS construction set			Independent validation set		
				Genotypes or alleles in cases	Genotypes or alleles in controls	P-value	Genotypes or alleles in cases	Genotypes or alleles in controls	P-value
rs10865331	A	G	Dominant	222/63	182/181	4.1x10 ⁻¹³	402/173	388/290	3.5x10 ⁻⁶
			Recessive	59/226	31/332	1.1x10 ⁻⁵	79/496	90/588	0.868
			Genotypic	59/163/63	31/151/181	1.3x10 ⁻¹³	79/323/173	90/298/290	9.4x10 ⁻⁶
			Allelic	281/289	213/513	3.1x10 ⁻¹³	481/669	478/878	0.001
rs27044	G	C	Dominant	204/81	246/117	0.304	-	-	-
			Recessive	61/224	85/278	0.571	-	-	-
			Genotypic	61/143/81	85/161/117	0.334	-	-	-
			Allelic	265/305	331/395	0.779	-	-	-
rs27434	A	G	Dominant	208/77	258/105	0.599	-	-	-
			Recessive	68/217	95/268	0.524	-	-	-
			Genotypic	68/140/77	95/163/105	0.560	-	-	-
			Allelic	276/294	353/373	0.955	-	-	-
rs27037	T	G	Dominant	149/136	236/127	0.001	-	-	-
			Recessive	29/256	45/318	0.387	-	-	-
			Genotypic	29/120/136	45/191/127	0.005	-	-	-
			Allelic	178/392	281/445	0.006	-	-	-
rs30187	T	C	Dominant	208/77	259/104	0.660	-	-	-
			Recessive	68/217	94/269	0.584	-	-	-
			Genotypic	68/140/77	94/165/104	0.659	-	-	-
			Allelic	276/294	353/373	0.955	-	-	-
rs17482078	T	C	Dominant	22/263	24/339	0.645	-	-	-
			Recessive	8/277	7/356	0.600	-	-	-

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			Genotypic	8/14/263	7/17/339	0.784	-	-	-
			Allelic	30/540	31/695	0.429	-	-	-
rs10050860	T	C	Dominant	22/263	24/339	0.645	-	-	-
			Recessive	0/285	1/362	1.000	-	-	-
			Genotypic	0/22/263	1/23/339	0.740	-	-	-
			Allelic	22/548	25/701	0.765	-	-	-

* Dominant: (DD, Dd) vs. dd, Recessive: DD vs. (Dd, dd), Genotypic: DD vs. Dd vs. dd, Allelic: D vs. d; D is the minor allele and d is the major allele

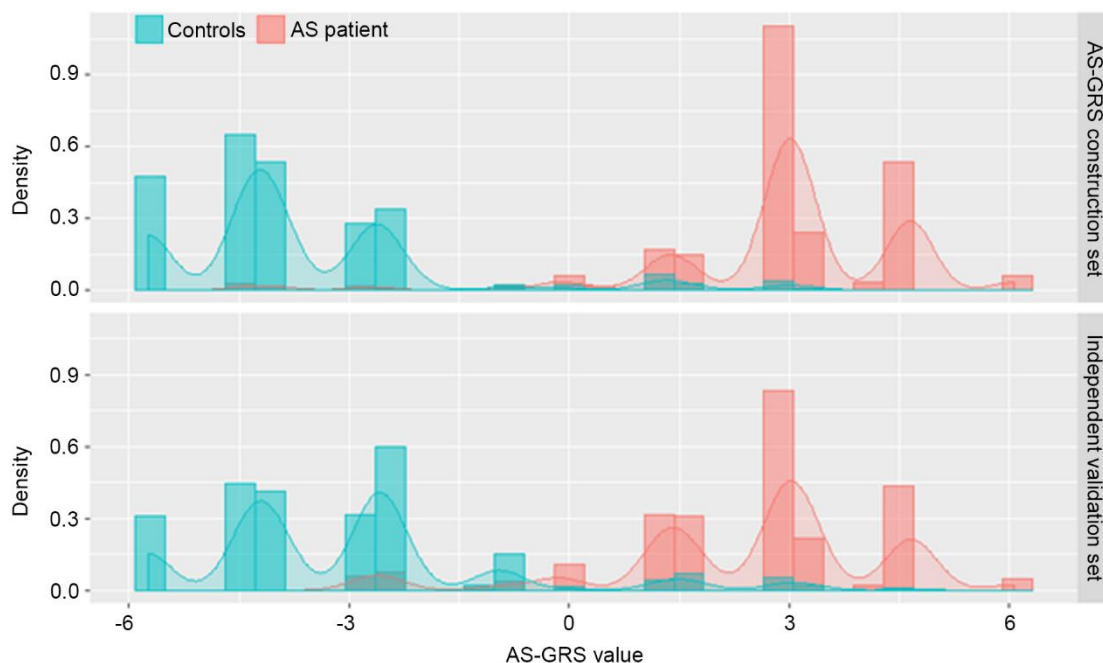
Supplementary Data 6. Associations of genetic factors in the AS-GRS model.

Genetic marker	Gene	AS-GRS construction set		Independent validation set	
		OR (95% CI)	<i>P</i>	OR (95% CI)	<i>P</i>
HLA-B27 positive	HLA-B27	273.07 (121.38-614.35)	6.91x10 ⁻⁴²	164.81 (101.46-267.70)	1.64x10 ⁻⁹⁴
1q32.2 deletion	<i>HHAT</i>	6.01 (2.76-13.05)	6.00x10 ⁻⁶	2.02 (1.30-3.14)	0.002
13q13.1 deletion	<i>EEF1DP3</i>	5.08 (2.34-11.02)	3.92x10 ⁻⁵	2.73 (1.74-4.30)	1.43x10 ⁻⁵
16p13.3 deletion	-	4.04 (1.15-14.14)	0.029	4.60 (1.92-11.03)	0.001
rs10865331 (A)	-	4.04 (1.86-8.77)	4.16x10 ⁻⁴	3.45 (2.15-5.56)	3.38x10 ⁻⁷

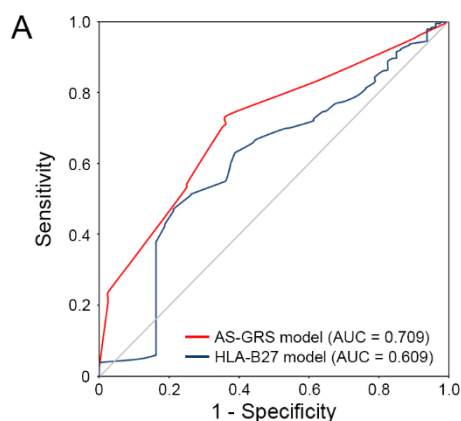
Logistic regression analysis for CNVs, SNP, and HLA-B27 factor.

95% CI = 95% confidence interval

Supplementary Data 7. Histograms by AS-GRS value in the AS-GRS construction set and independent validation set.



Supplementary Data 8. ROC curves for the AS-GRS and HLA-B27 models in HLA-B27 positive individuals. A, red and blue lines represent the AS-GRS and HLA-B27 based prediction models, respectively. B, sensitivity, specificity and accuracy of the AS-GRS model at specified thresholds.



B

Sensitivity (%)	Specificity (%)	Accuracy (%)	Threshold
100.0	0	91.0	-1.14
96.0	8.8	88.2	0.56
84.4	35.0	80.0	1.37
80.1	46.3	77.0	1.57
73.4	63.8	72.5	2.16
54.2	75.0	56.1	3.00
33.4	90.0	38.5	3.17
20.8	97.5	27.7	4.68
0	100.0	9.0	7.07

Online supplement to: Developing a Risk-scoring model for Ankylosing Spondylitis Based on a Combination of HLA-B27, Single-nucleotide Polymorphism, and Copy Number Variant Markers. *The Journal of Rheumatology*. doi:10.3899/jrheum.160347

Supplementary Data 9. Winner's curse effects.

Marker	MAF	A1	P-value	Unadjusted odds ratio	Bias-corrected odds ratio (95% CI)
1q32.2	0.171	Deletion	1.3×10^{-15}	2.87	2.90 (2.10-4.62)
13q13.1	0.131	Deletion	1.3×10^{-16}	3.13	2.78 (1.94-4.86)
16p13.3	0.025	Deletion	2.2×10^{-4}	2.77	2.72 (1.19-29.28)
rs10865331	0.293	A	9.7×10^{-14}	2.34	2.55 (1.83-4.27)

Winner's curse effects were estimated using 'BR-squared' software (Hum Genet 2011; 129:545-552). 95% CI = 95% confidence interval