

Supplementary Information

Table S1. Allele and genotype frequencies in AAU

Gene	SNP	Allele and Genotype	AAU	Controls	P	Pc	OR(95%CI)
JAK1	rs310241	C	555(30.6%)	392(27.5%)	0.06	NS	1.16(1.00-1.35)
		CC	74(8.2%)	45(6.3%)	0.16	NS	1.32(0.90-1.93)
		CT	407(44.8%)	302(42.4%)	0.32	NS	1.11(0.91-1.35)
		TT	427(47.0%)	366(51.3%)	0.09	NS	0.84(0.70-1.02)
	rs2780815	G	1478(86.6%)	1250(87.5%)	0.46	NS	0.92(0.75-1.14)
		GG	642(75.3%)	548(76.7%)	0.49	NS	0.92(0.73-1.16)
		GT	194(22.7%)	154(21.6%)	0.58	NS	1.07(0.84-1.36)
JAK2	rs10758669	TT	17(2.0%)	12(1.7%)	0.65	NS	1.19(0.56-2.51)
		A	1242(67.3%)	980(69.0%)	0.30	NS	0.92(0.80-1.07)
		AA	405(43.9%)	346(48.7%)	0.05	NS	0.82(0.68-1.00)
		AC	432(46.8%)	288(40.6%)	0.01	NS	1.29(1.06-1.57)
	CC	86(9.3%)	76(10.7%)	0.35	NS	0.86(0.62-1.18)	
rs10975003	C	406(23.7%)	272(21.2%)	0.10	NS	1.16(0.97-1.38)	

		CC	42(4.9%)	28(4.4%)	0.62	NS	1.13(0.70-1.85)
		CT	322(37.6%)	216(33.6%)	0.11	NS	1.19(0.96-1.48)
		TT	492(57.5%)	399(62.1%)	0.07	NS	0.83(0.67-1.02)
STAT1	rs1547550	C	1601(87.6%)	1254(87.8%)	0.84	NS	0.98(0.80-1.21)
		CC	697(76.3%)	555(77.7%)	0.48	NS	0.92(0.73-1.16)
		CG	207(22.6%)	144(20.2%)	0.23	NS	1.16(0.91-1.48)
		GG	10(1.1%)	15(2.1%)	0.10	NS	0.52(0.23-1.15)
	rs2066802	C	346(19.7%)	288(20.7%)	0.47	NS	0.94(0.79-1.12)
		CC	29(3.3%)	27(3.9%)	0.87	NS	0.95(0.56-1.63)
		CT	288(32.8%)	234(33.7%)	0.75	NS	0.96(0.78-1.19)
		TT	562(63.9%)	434(62.4%)	0.54	NS	1.07(0.87-1.31)
	rs6718902	C	1017(56.2%)	778(54.5%)	0.32	NS	1.07(0.93-1.24)
		CC	289(32.0%)	209(29.3%)	0.24	NS	1.14(0.94-1.27)
		CT	439(48.5%)	360(50.4%)	0.46	NS	0.93(0.76-1.13)
		TT	176(19.5%)	145(20.3%)	0.67	NS	0.95(0.74-1.21)
	rs10199181	A	453(27.0%)	397(27.8%)	0.62	NS	0.96(0.82-1.13)
		AA	72(8.6%)	55(7.7%)	0.53	NS	1.13(0.78-1.62)

		AT	309(36.8%)	287(40.2%)	0.17	NS	0.87(0.71-1.07)
		TT	458(54.6%)	372(52.1%)	0.33	NS	1.11(0.91-1.35)
IRF1	rs2070721	A	590(34.9%)	487(34.1%)	0.65	NS	1.04(0.89-1.20)
		AA	98(11.6%)	74(10.4%)	0.44	NS	1.13(0.82-1.56)
		AC	394(46.6%)	339(47.4%)	0.72	NS	0.96(0.79-1.18)
		CC	354(41.8%)	301(42.2%)	0.90	NS	0.99(0.81-1.21)
NOS2	rs2297518	A	317(17.6%)	209(16.2%)	0.30	NS	1.11(0.91-1.34)
		AA	22(2.4%)	12(1.9%)	0.44	NS	1.32(0.65-2.69)
		AG	273(30.3%)	185(28.6%)	0.48	NS	1.08(0.87-1.35)
		GG	607(67.3%)	450(69.5%)	0.35	NS	0.90(0.73-1.12)
	rs4795067	A	1377(74.1%)	1008(76.1%)	0.19	NS	0.90(0.76-1.06)
		AA	514(55.3%)	382(57.7%)	0.35	NS	0.91(0.74-1.11)
		AG	349(37.6%)	244(36.9%)	0.77	NS	1.03(0.84-1.27)
		GG	66(7.1%)	36(5.4%)	0.18	NS	1.33(0.88-2.02)

OR = odds ratio; 95%CI = 95% confidence interval;

Pc = P value adjusted by Bonferroni correction.

Table S2. Allele and genotype frequencies in male AAU patients and male controls

Gene	SNP	Allele and Genotype	AAU⁺AS⁺ (Male)	AAU⁺AS⁻ (Male)	Controls (Male)	P (AS⁺)	Pc (AS⁺)	OR (95%CI)	P (AS⁻)	Pc (AS⁻)	OR (95%CI)
JAK1	rs310241	C	193(29.9%)	144(31.0%)	236(26.8%)	0.18	NS	1.17(0.93-1.46)	0.10	NS	1.23(0.96-1.58)
		CC	29(9.0%)	19(8.2%)	28(6.4%)	0.17	NS	1.46(0.85-2.50)	0.37	NS	1.32(0.72-2.41)
		CT	135(41.8%)	106(45.7%)	180(40.8%)	0.79	NS	1.04(0.78-1.39)	0.22	NS	1.22(0.89-1.68)
		TT	159(49.2%)	107(46.1%)	233(52.8%)	0.32	NS	0.87(0.65-1.15)	0.09	NS	0.76(0.56-1.05)
	rs2780815	G	524(87.0%)	395(89.4%)	755(89.0%)	0.25	NS	0.83(0.60-1.14)	0.86	NS	1.04(0.71-1.50)
		GG	228(75.7%)	176(79.6%)	336(79.2%)	0.26	NS	0.82(0.58-1.16)	0.91	NS	1.02(0.69-1.53)
		GT	68(22.6%)	43(19.5%)	83(19.6%)	0.32	NS	1.20(0.84-1.72)	0.97	NS	0.99(0.66-1.50)
		TT	5(1.7%)	2(0.9%)	5(1.2%)	0.58	NS	1.42(0.41-4.93)	0.75	NS	0.77(0.15-3.98)
JAK2	rs10758669	A	415(64.4%)	329(67.7%)	537(67.1%)	0.29	NS	0.89(0.71-1.10)	0.83	NS	1.21(0.81-1.31)
		AA	126(39.1%)	116(47.7%)	202(46.5%)	0.04	NS	0.74(0.55-0.99)	0.77	NS	1.05(0.77-1.44)
		AC	163(50.6%)	97(39.9%)	177(40.8%)	7.17×10 ⁻³	NS	1.49(1.11-1.99)	0.83	NS	0.97(0.70-1.33)
		CC	33(10.2%)	30(12.3%)	55(12.7%)	0.30	NS	0.79(0.50-1.24)	0.90	NS	0.97(0.60-1.56)
	rs10975003	C	155(26.4%)	90(19.7%)	179(23.4%)	0.21	NS	1.17(0.92-1.51)	0.14	NS	0.81(0.61-1.07)
		CC	20(6.8%)	9(3.9%)	20(5.2%)	0.39	NS	1.33(0.70-2.51)	0.47	NS	0.75(0.33-1.67)

		CT	115(39.1%)	72(31.6%)	139(36.3%)	0.45	NS	1.13(0.82-1.54)	0.24	NS	0.81(0.57-1.15)
		TT	159(54.1%)	147(64.5%)	224(58.5%)	0.25	NS	0.84(0.62-1.14)	0.14	NS	1.29(0.92-1.81)
STAT1	rs1547550	C	571(87.8%)	414(86.6%)	659(87.9%)	0.99	NS	1.00(0.72-1.38)	0.52	NS	0.89(0.63-1.26)
		CC	246(75.7%)	181(75.7%)	292(77.9%)	0.50	NS	0.89(0.62-1.26)	0.54	NS	0.88(0.61-1.30)
		CG	79(24.3%)	52(21.8%)	75(20.0%)	0.17	NS	1.29(0.90-1.84)	0.60	NS	1.11(0.75-1.66)
		GG	0(0.0%)	6(2.5%)	8(2.1%)	-	-	-	0.76	NS	1.18(0.41-3.45)
	rs2066802	C	127(21.0%)	92(20.0%)	175(20.4%)	0.81	NS	1.03(0.80-1.33)	0.85	NS	0.97(0.73-1.29)
		CC	14(4.6%)	8(3.5%)	19(4.4%)	0.91	NS	1.04(0.51-2.11)	0.55	NS	0.78(0.33-1.80)
		CT	99(32.7%)	76(33.0%)	137(32.0%)	0.85	NS	1.03(0.75-1.41)	0.79	NS	1.05(0.75-1.48)
		TT	190(62.7%)	146(63.5%)	272(63.6%)	0.82	NS	0.96(0.71-1.31)	0.99	NS	1.00(0.72-1.39)
	rs6718902	C	349(55.6%)	277(57.5%)	470(54.9%)	0.80	NS	1.03(0.84-1.26)	0.37	NS	1.11(0.89-1.39)
		CC	102(32.5%)	79(32.8%)	127(29.7%)	0.41	NS	1.14(0.83-1.56)	0.40	NS	1.16(0.82-1.62)
		CT	145(46.2%)	119(49.4%)	216(50.4%)	0.25	NS	0.84(0.63-1.13)	0.79	NS	0.96(0.70-1.31)
		TT	67(21.3%)	43(17.8%)	85(19.9%)	0.62	NS	1.10(0.76-1.57)	0.52	NS	0.88(0.58-1.32)
	rs10199181	A	165(27.9%)	120(27.8%)	249(29.6%)	0.47	NS	0.92(0.73-1.16)	0.49	NS	0.91(0.71-1.18)
		AA	30(10.1%)	16(7.4%)	36(8.6%)	0.48	NS	1.20(0.72-2.00)	0.61	NS	0.85(0.46-1.58)
		AT	105(35.5%)	88(40.7%)	177(42.1%)	0.07	NS	0.76(0.56-1.03)	0.73	NS	0.94(0.68-1.32)

		TT	161(54.4%)	112(51.9%)	207(49.3%)	0.18	NS	1.23(0.91-1.65)	0.54	NS	1.11(0.80-1.54)
IRF1	rs2070721	A	197(33.6%)	153(34.8%)	271(34.0%)	0.89	NS	0.99(0.79-1.23)	0.77	NS	1.04(0.81-1.32)
		AA	29(9.9%)	28(12.7%)	45(11.3%)	0.56	NS	0.86(0.53-1.42)	0.59	NS	1.15(0.69-1.90)
		AC	139(47.4%)	97(44.1%)	181(45.3%)	0.59	NS	1.09(0.80-1.47)	0.76	NS	0.95(0.68-1.32)
		CC	125(42.7%)	95(43.2%)	173(43.4%)	0.86	NS	0.97(0.72-1.32)	0.97	NS	0.99(0.71-1.38)
NOS2	rs2297518	A	114(18.1%)	82(17.6%)	133(16.3%)	0.36	NS	1.14(0.86-1.50)	0.54	NS	1.10(.81-1.49)
		AA	7(2.3%)	6(2.6%)	10(2.4%)	0.84	NS	0.91(0.34-2.41)	0.92	NS	1.06(0.38-2.94)
		AG	100(31.7%)	70(30.0%)	113(27.6%)	0.23	NS	1.22(0.88-1.68)	0.52	NS	1.13(0.79-1.60)
		GG	208(66.0%)	157(67.4%)	286(70.0%)	0.26	NS	0.84(0.61-1.15)	0.50	NS	0.89(0.63-1.26)
	rs4795067	A	476(73.0%)	359(73.9%)	569(74.9%)	0.43	NS	0.91(0.72-1.15)	0.69	NS	0.95(0.73-1.23)
		AA	171(52.5%)	136(56.0%)	213(56.1%)	0.34	NS	0.87(0.64-1.16)	0.98	NS	1.00(0.72-1.38)
		AG	134(41.1%)	87(35.8%)	143(37.6%)	0.35	NS	1.16(0.85-1.57)	0.64	NS	0.92(0.66-1.29)
		GG	21(6.4%)	20(8.2%)	24(6.3%)	0.95	NS	1.02(0.56-1.87)	0.36	NS	1.33(0.72-2.47)

OR = odds ratio; 95%CI = 95% confidence interval;

Pc = P value adjusted by Bonferroni correction.

Table S3. Allele and genotype frequencies in AAU stratified by AS status

Gene	SNP	Allele and Genotype				P	Pc	OR	P	Pc	OR
			AAU ⁺ AS ⁺	AAU ⁺ AS ⁻	Controls	(AAU ⁺ AS ⁺)	(AAU ⁺ AS ⁺)	(95%CI)	(AAU ⁺ AS ⁻)	(AAU ⁺ AS ⁻)	(95%CI)
JAK1	rs310241	C	273(30.8%)	282(30.3%)	392(27.5%)	0.09	NS	1.18(0.98-1.41)	0.14	NS	1.15(0.96-1.38)
		CC	41(9.3%)	33(7.0%)	45(6.3%)	0.06	NS	1.51(0.97-2.35)	0.73	NS	1.13(0.71-1.81)
		CT	191(43.1%)	216(46.5%)	302(42.4%)	0.80	NS	1.03(0.81-1.31)	0.17	NS	1.18(0.93-1.49)
		TT	211(47.6%)	216(46.5%)	366(51.3%)	0.22	NS	0.86(0.68-1.09)	0.10	NS	0.82(0.65-1.04)
	rs2780815	G	683(85.0%)	795(88.1%)	1250(87.5%)	0.08	NS	0.80(0.63-1.03)	0.66	NS	1.06(0.82-1.37)
		GG	291(72.4%)	351(77.8%)	548(76.7%)	0.11	NS	0.79(0.60-1.05)	0.67	NS	1.06(0.80-1.41)
		GT	101(25.1%)	93(20.6%)	154(21.6%)	0.17	NS	1.22(0.92-1.63)	0.70	NS	0.95(0.71-1.26)
		TT	10(2.5%)	7(1.6%)	12(1.7%)	0.35	NS	1.49(0.64-3.49)	0.87	NS	0.92(0.36-2.36)
JAK2	rs10758669	A	585(66.9%)	657(67.6%)	980(69.0%)	0.30	NS	0.91(0.76-1.09)	0.47	NS	0.94(0.79-1.12)
		AA	185(42.3%)	220(45.3%)	346(48.7%)	0.04	NS	0.77(0.61-0.98)	0.24	NS	0.87(0.69-1.10)
		AC	215(49.2%)	217(44.7%)	288(40.6%)	4.21×10 ⁻³	NS	1.42(1.12-1.80)	0.16	NS	1.18(0.94-1.49)
		CC	37(8.5%)	49(10.0%)	76(10.7%)	0.22	NS	0.77(0.51-1.17)	0.70	NS	0.93(0.64-1.36)
	rs10975003	C	199(24.8%)	207(22.7%)	272(21.2%)	0.05	NS	1.23(0.10-1.52)	0.37	NS	1.10(0.89-1.35)
		CC	22(5.5%)	20(4.4%)	28(4.4%)	0.41	NS	1.28(0.72-2.26)	0.97	NS	1.01(0.56-1.82)

		CT	155(38.7%)	167(36.7%)	216(33.5%)	0.10	NS	1.25(0.96-1.61)	0.28	NS	1.15(0.89-1.47)
		TT	224(55.8%)	268(58.9%)	399(62.1%)	0.05	NS	0.77(0.60-0.10)	0.29	NS	0.88(0.69-1.12)
STAT1	rs1547550	C	776(87.6%)	825(87.6%)	1254(87.8%)	0.87	NS	0.98(0.76-1.26)	0.86	NS	0.98(0.76-1.26)
		CC	334(75.4%)	363(77.1%)	555(77.7%)	0.36	NS	0.88(0.66-1.16)	0.79	NS	0.96(0.73-1.27)
		CG	108(24.4%)	99(21.0%)	144(20.2%)	0.09	NS	1.28(0.96-1.70)	0.72	NS	1.05(0.79-1.40)
		GG	1(0.2%)	9(1.9%)	15(2.1%)	0.01	NS	0.11(0.01-0.80)	0.82	NS	0.91(0.39-2.09)
	rs2066802	C	173(20.8%)	173(18.7%)	288(20.7%)	0.97	NS	1.00(0.81-1.24)	0.23	NS	0.88(0.71-1.08)
		CC	15(3.6%)	14(3.0%)	27(3.9%)	0.82	NS	0.93(0.49-1.76)	0.44	NS	0.77(0.40-1.49)
		CT	143(34.4%)	145(31.3%)	234(33.7%)	0.81	NS	1.03(0.80-1.33)	0.40	NS	0.90(0.70-1.16)
		TT	258(62.0%)	304(65.7%)	434(62.4%)	0.89	NS	0.98(0.76-1.26)	0.27	NS	1.15(0.90-1.47)
	rs6718902	C	481(56.7%)	536(55.8%)	778(54.5%)	0.30	NS	1.10(0.92-1.30)	0.52	NS	1.06(0.90-1.25)
		CC	138(32.5%)	151(31.5%)	209(29.3%)	0.25	NS	1.17(0.90-1.51)	0.42	NS	1.11(0.86-1.43)
		CT	205(48.3%)	234(48.7%)	360(50.4%)	0.50	NS	0.92(0.72-1.17)	0.57	NS	0.94(0.74-1.18)
		TT	81(19.2%)	95(19.8%)	145(20.3%)	0.62	NS	0.93(0.68-1.26)	0.83	NS	0.97(0.73-1.30)
	rs10199181	A	216(27.1%)	237(26.9%)	397(27.8%)	0.71	NS	0.96(0.80-1.17)	0.65	NS	0.96(0.79-1.16)
		AA	36(9.0%)	36(8.2%)	55(7.7%)	0.44	NS	1.19(0.77-1.84)	0.77	NS	1.07(0.69-1.66)
		AT	144(36.1%)	165(37.5%)	287(40.2%)	0.18	NS	0.84(0.65-1.08)	0.36	NS	0.89(0.70-1.14)

		TT	219(54.9%)	239(54.3%)	372(52.1%)	0.37	NS	1.12(0.88-1.43)	0.46	NS	1.09(0.86-1.39)
IRF1	rs2070721	A	283(35.5%)	307(34.3%)	487(34.1%)	0.52	NS	1.06(0.89-1.27)	0.91	NS	1.01(0.85-1.21)
		AA	44(11.0%)	54(12.1%)	74(10.4%)	0.73	NS	1.07(0.72-1.59)	0.36	NS	1.19(0.82-1.73)
		AC	195(48.9%)	199(44.5%)	339(47.4%)	0.66	NS	1.06(0.83-1.35)	0.33	NS	0.89(0.70-1.13)
		CC	160(40.1%)	194(43.4%)	301(42.2%)	0.50	NS	0.92(0.72-1.18)	0.68	NS	1.05(0.83-1.34)
NOS2	rs2297518	A	166(18.8%)	151(16.4%)	209(16.2%)	0.11	NS	1.20(0.96-1.50)	0.87	NS	1.02(0.81-1.28)
		AA	10(2.3%)	12(2.6%)	12(1.9%)	0.64	NS	1.22(0.53-2.86)	0.40	NS	1.42(0.63-3.19)
		AG	146(33.0%)	127(27.6%)	185(28.6%)	0.12	NS	1.23(0.95-1.60)	0.72	NS	0.95(0.73-1.24)
		GG	286(64.7%)	321(69.8%)	450(69.5%)	0.09	NS	0.80(0.62-1.04)	0.93	NS	1.01(0.78-1.31)
	rs4795067	A	649(73.3%)	728(74.9%)	1008(76.1%)	0.13	NS	0.86(0.71-1.04)	0.50	NS	0.94(0.77-1.13)
		AA	233(52.6%)	281(57.8%)	382(57.7%)	0.09	NS	0.81(0.64-1.04)	0.97	NS	1.01(0.79-1.27)
		AG	183(41.3%)	166(34.2%)	244(36.9%)	0.14	NS	1.21(0.94-1.54)	0.35	NS	0.89(0.70-1.14)
		GG	27(6.1%)	39(8.0%)	36(5.4%)	0.64	NS	1.13(0.68-1.89)	0.08	NS	1.52(0.95-2.43)

OR = odds ratio; 95%CI = 95% confidence interval;

Pc = P value adjusted by Bonferroni correction

Table S4. Multivariate logistic regression analysis of rs10758669 polymorphisms in patients with AAU and healthy controls stratified by AS and HLA-B27 status after adjustment for gender and age.

Model	Genotype	AAU					AAU+ AS+					HLA-B27(+)				
		control (n=710)	Case (n=923)	OR (95% CI)	P	Pc	Control (n=710)	Case (n=437)	OR (95% CI)	P	Pc	Control (n=710)	Case (n=535)	(95% CI)	P	Pc
Additive				1.09(0.93-1.27)	0.27	NS			1.07(0.88-1.28)	0.51	NS			1.15(0.97-1.37)	0.12	NS
Codominant	AA	346(48.7%)	405(43.9%)	Reference			346(48.7%)	185(42.3%)	Reference			346(48.7%)	220(41.1%)	Reference		
	CA	288(40.6%)	432(46.8%)	1.28(1.04-1.58)	0.02	NS	288(40.6%)	215(49.2%)	1.36(1.05-1.76)	0.02	NS	288(40.6%)	265(49.5%)	1.43(1.13-1.82)	3.50×10 ⁻³	NS
	CC	76(10.7%)	86(9.3%)	0.97(0.69-1.38)	0.88	NS	76(10.7%)	37(8.5%)	0.84(0.54-1.31)	0.44	NS	76(10.7%)	50(9.4%)	1.01(0.68-1.52)	0.92	NS
Dominant	AA	346(48.7%)	405(43.9%)	Reference			346(48.7%)	185(42.3%)	Reference			346(48.7%)	220(41.1%)	Reference		
	CA+CC	364(51.3%)	518(56.1%)	1.22(1.00-1.49)	0.05	NS	364(51.3%)	252(57.7%)	1.25(0.98-1.60)	0.07	NS	364(51.3%)	315(58.9%)	1.35(1.07-1.69)	0.01	NS
Recessive	AA+CA	634(89.3%)	837(90.7%)	Reference			634(89.3%)	400(91.5%)	Reference			634(89.3%)	485(90.6%)	Reference		
	CC	76(10.7%)	86(9.3%)	0.86(0.62-1.20)	0.38	NS	76(10.7%)	37(8.5%)	0.72(0.47-1.10)	0.12	NS	76(10.7%)	50(9.4%)	0.85(0.58-1.25)	0.41	NS

OR = odds ratio; 95%CI = 95% confidence interval.

Pc = P value adjusted by Bonferroni correction.

Table S5. Multivariate logistic regression analysis of rs10975003 polymorphisms in female AAU patients and female controls after adjustment for gender and age.

Model	Genotype	AAU(Female)		OR (95% CI)	P	Pc
		Control (Female) (n=643)	Case (Female) (n=334)			
Additive				1.46(1.08-1.97)	0.01	NS
Codominant	TT	399(62.0%)	186(55.7%)	Reference		
	CT	216(33.6%)	135(40.4%)	1.57(1.11-2.23)	0.01	NS
	CC	28(4.4%)	13(3.9%)	1.59(0.64-3.96)	0.32	NS
Dominant	TT	399(62.0%)	186(55.7%)	Reference		
	CT+CC	244(38.0%)	148(44.3%)	1.57(1.12-2.21)	9.10×10 ⁻³	NS
Recessive	TT+CT	615(95.6%)	321(96.1%)	Reference		
	CC	28(4.4%)	13(3.9%)	1.36(0.55-3.35)	0.51	NS

OR = odds ratio; 95%CI = 95% confidence interval.

Pc = P value adjusted by Bonferroni correction.