Supplementary Table 1

Microsatellites and the primers

Locus	in hg19		H^b	Primer sequence		
	Physical position	Repeat unit[a]		Forward	Reverse	(°C)[c]
D6S2840	30407655-30407851	$(CT)_{14}$	0.890	AGATGGCATTTGGAGAGTGCAG	TCCTTACAGCAGAGATATGTGG	57
D6S2799	30475190-30475416	$(AAAG)_7$	0.495	TTCCGAGCAGTCTTCATTGG	TGAGGCAGGAGAATCGCTTG	59
D6S2827	30781051-30781275	$(GT)_9$	0.703	GAGCCACGGAGAGTCTCCCTTTATC	TCCAGGAACTGTGAGTAGTAAGAAC	57
D6S2825	30859182-30859405	$(GT)_{14}$	0.765	CCATCCCTCTTCTCAGCTCTTCCATC	TGTGGGGCAGACAATGTATTTATCTG	57
D6S2938	31061596-31062014	$(ATTT)_7$	0.572	AACCCAGGGAATCCTAGAAG	CAGTAGCTTTAGTCTTTCCC	57
D6S2931	31204137-31204564	$(TTTC)_{19}$	0.859	CAGCTGCTAGGTGTATCTGAATAC	CAATAAGAAATTTTGCTATAAGGTAAG	57
D6S2930	31209069-31209484	$(CTTC)_8$	0.860	TAGAAAACGCAATCTCGGCC	CTGGATTAACCTGGAGACTC	57
D6S2811	31259102-31259301	$(GT)_{23}$	0.903	CAGTAGTAAGCCAGAAGCTATTAC	AAGTCAAGCATATCTGCCATTTGG	57
MICA	31380107-31380335	$(GCT)_2G(GCT)_3$	0.777	GTGCTGGTGCTTCAGAGTCATT	CCTTGTCACCAACATGCCTA	60
D6S2793	31471706-31471915	$(TG)_{19}$	0.791	ATGGGCAAGACTTCAATGGC	CTACCTCCTTGCCAAACTTG	59
D6S2780	31709084-31709237	$(AC)_{23}$	0.819	GCTGCAGTGAGCCAAGATTG	TCCTCTGCTCTCTGGGATTG	62
D6S2924	31836518-31836670	$(CAAA)_8$	0.721	GGACATTGCTCTGACTTGAG	AGCTGAGATTGCACTGCTG	59
D6S2973	31906110-31906336	$(TA)_8$	0.163	ATGAGGATACCCACTCTACC	AGTTCGCACCACTGTAGTCC	59
D6S2920	32045736-32045948	$(GAAA)_{15}$	0.868	CCTGACCACAAAGCTTTCTC	AAAGGTTGCAGTGAGCCAAG	56
D6S2740	32219824-32220146	$(TG)_{10}$	0.703	ACCTGGTACATTCCTAAAGCC	ATGTGAGGATAGATCTCTGGG	53
D6S2892	32269688-32269904	$(ATA)_9$	0.812	ACTCAACCCTGCTGTTGTAG	TGCATGTCCTGTGAGGTAAG	53
D6S2885	32341989-32342152	$(TG)_{22}$	0.789	AGGTGACCTGGACCTTACTG	ACACTATGCTAGTCTGTGCC	57
D6S2883	32403351-32403488	$(AC)_7$	0.795	ACATTATGTTCTGTTGCATG	TACTTTCCTAATTCTCCTCC	57
D6S2876	32670036-32670255	$(GT)_{14}$	0.688	GACAGCTCTTCTTAACCTGC	GGTAAAATTCCTGACTGGCC	57
D6S2818	32737264-32737397	$(TTTG)_8$	0.701	ATGAAGTGAGCTGTGATCGC	AGAGCTGCAGTGCTGTATTG	59
D6S2820	32817465-32817660	$(CAAA)_4$	0.213	AGATCCTGGCTTGATGATGC	TTGCAGTGAGCCCAGATC	62
D6S2822	32853912-32854116	$(TATC)_{12}$	0.673	TCACTCATGGTTGCTTTTCC	GAATGATAGGAGTCCATTGTGG	57

[a]Repeat unit sequence are referred for the UCSC Genome Browser assembly GRCh37/hg19. [b]Heterozygosity of each microsatellite in the control subjects. [c]Annealing Temperature in each PCR reaction.

Supplementary Table 2

Overview of sequencing output by NGS

Subjects		Haplotype1			Haplotype2			Sequencing status							
		D6S2930 - *HLA-C* - *D6S2811*			*D6S2930* - *HLA-C* - *D6S2811*			Yield (mega bases)	Reads	Average bases	Mapped reads	Paired mapped reads	Mean coverage	% target bases at >=10X	% target bases at >=20X
AA083	Case	***441***	***C*04:01***	***208***	*437*	*C*15:02*	*194*	2320	22841942	101.6	13757754	13610648	280.5	94.5	93.5
AA086	Case	***441***	***C*04:01***	***208***	*437*	*C*15:02*	*194*	2300	22648454	101.5	12369001	12232126	251.8	95.2	94.1
AA211	Case	***441***	***C*04:01***	***208***	*429*	*C*12:02*	*204*	2039	20070420	101.6	11998428	11833266	244.5	96.0	95.1
AA215	Case	***441***	***C*04:01***	***208***	*441*	*C*03:04*	*194*	2070	20344272	101.8	13126105	12854246	267.7	94.8	93.7
AA267	Case	***441***	***C*04:01***	***208***	*433*	*C*07:02*	*192*	2372	23179050	102.3	8518411	8425394	174.3	94.5	92.9
AA214	Case	*437*	*C*03:03*	*196*	*441*	*C*03:03*	*196*	1970	19313488	102.0	11133559	11000613	227.7	94.9	93.8
AA249	Case	*437*	*C*01:02*	*216*	*441*	*C*01:02*	*216*	2363	23107648	102.3	13823924	13678332	283.5	95.1	94.2
AA289	Case	*437*	*C*01:02*	*212*	*433*	*C*03:03*	*196*	2020	19820778	101.9	11607431	11448500	237.2	96.0	95.1
AA292	Case	*409*	*C*07:02*	*196*	*445*	*C*01:02*	*212*	2040	20035423	101.8	12452906	12280422	254.4	95.5	94.5
AA298	Case	*433*	*C*04:01*	*210*	*469*	*C*14:03*	*216*	2210	21667416	102.0	13204169	13036608	270.0	95.3	94.3
T416	Control	*437*	*C*01:02*	*212*	*469*	*C*14:03*	*216*	2035	19952372	102.0	12467409	12320619	255.1	95.5	94.6
T438	Control	*409*	*C*07:02*	*196*	*461*	*C*14:02*	*198*	2165	21199733	102.1	12264395	12065036	250.8	95.5	94.6

Risk haplotypes associated with AA are shown in bold.

Supplementary Table 3

Primer sets for PCR direct sequencing in all exons of *CCHCR1* gene

Target exon	Amplicon size (bp)	Physical position	Amplification and sequencing primer		(°C)[a]	Additional sequencing primer
			Forward	Reverse		
01 - 03	1,299	31124423-31125721	GGGCAGGCTATTGAGAGATGG	GGCCGAAATAGGGTAAGGAGTT	61	GGATTATGACTTTGTTATTGTAGG
04	464	31122226-31122689	TCCCCACACGGAAGCAGAGG	CCCAGCTCTCCGTTATGAATTTGA	65	AGGGGTCCTCTTTTCCTTCT
05 - 08	1,248	31117761-31119008	GCTGATTGGTTGGGTCATTCTCTA	AACACTGGTTGAATGGATGCCAC	63	AGGAGCGTGTAGAGCACAGC AGCTCATGTTGCCCAGGCAG
9 - 10	576	31116034-31116609	GCATGGCATTCTTACAGAGATCTC	GGAACATTGATAGAGCTAAATCTGC	59	
11 - 15	1,333	31112369-31113701	TGCGTTCTGTACTTGTATAATGGGAC	TGAAGCTTTGAACACACTTTGAGG	61	GCTGGGACCCCCAAAACCAT
16 - 18	1,148	31110146-31111293	AGCCACAGAGTTGGTGACCCAG	AATCTCCAAGAGAGATGGCTGCA	64	TCTCCCATTCCTCATTTCC CAGCGTTTGTTCCTGTCTTC

[a]Annealing temperature used for each PCR reaction.

Supplementary Table 4
Allelic association analysis of 23 loci spanning 2.45 Mb in the MHC region

	For locus			For an allele in each locus						
		HWE[b]			Allele frequency		Allelic association[d]			
Locus	No of allele[a]	Case	Control	Allele[c]	Case	Control	OR	(95% CI)		*P*
D6S2840	19	0.549	0.295	*217*	0.073	0.049	1.51	0.93	- 2.46	1.02×10^{-1}
D6S2799	3	0.107	0.335	*222*	0.693	0.649	1.22	0.93	- 1.60	1.52×10^{-1}
D6S2827	8	0.212	0.917	*218*	0.105	0.054	1.99	1.30	- 3.06	2.03×10^{-3}
D6S2825	14	0.138	0.445	*235*	0.298	0.370	0.73	0.57	- 0.95	1.66×10^{-2}
D6S2938	6	0.057	0.781	*429*	0.158	0.104	1.60	1.13	- 2.27	9.41×10^{-3}
D6S2931	17	0.211	0.380	*371*	0.234	0.170	1.48	1.10	- 1.97	9.89×10^{-3}
D6S2930	15	0.187	0.265	*441*	0.228	0.144	1.81	1.32	- 2.48	2.54×10^{-4}
HLA-C	17	0.043	0.424	*C*04:01*	0.082	0.036	2.47	1.48	- 4.11	5.33×10^{-4}
D6S2811[e]	21	0.368	0.014	*208*	0.076	0.025	3.41	1.94	- 5.99	3.39×10^{-5}
MICA	5	0.933	0.056	*A5*	0.371	0.293	1.43	1.10	- 1.84	6.77×10^{-3}
D6S2793	16	0.807	0.549	*224*	0.020	0.004	4.74	1.48	- 15.1	8.92×10^{-3}
D6S2780	19	0.914	0.325	*161*	0.012	0.003	4.45	0.99	- 20.1	5.43×10^{-2}
D6S2924	8	0.113	0.873	*158*	0.181	0.146	1.31	0.94	- 1.82	1.10×10^{-1}
D6S2973	4	0.828	0.721	*232*	0.012	0.004	3.33	0.82	- 13.5	1.00×10^{-1}
D6S2920	15	0.631	0.396	*208*	0.135	0.095	1.50	1.03	- 2.18	3.80×10^{-2}
D6S2740	5	0.204	0.740	*322*	0.155	0.235	0.59	0.43	- 0.82	1.16×10^{-3}
D6S2892	11	0.271	0.441	*206*	0.079	0.102	0.76	0.49	- 1.17	2.02×10^{-1}
D6S2885	17	0.097	0.306	*162*	0.070	0.038	1.88	1.12	- 3.14	1.34×10^{-2}
D6S2883	9	0.248	0.248	*138*	0.184	0.246	0.70	0.51	- 0.94	1.71×10^{-2}
D6S2876	8	0.264	0.346	*215*	0.152	0.121	1.30	0.92	- 1.85	1.42×10^{-1}
D6S2818	5	0.846	0.450	*124*	0.120	0.188	0.59	0.41	- 0.85	2.63×10^{-3}
D6S2820	2	0.475	0.321	*194*	0.120	0.121	0.99	0.68	- 1.43	9.39×10^{-1}
D6S2822	8	0.959	0.167	*196*	0.257	0.220	1.24	0.93	- 1.66	1.40×10^{-1}

Allelic association analysis of 23 loci spanning 2.45 Mb in the MHC region. [a]Number of alleles observed in each locus. [b]*P*-values for exact test of Hardy-Weinberg proportion. [c]Allele showing smallest *P*-value and greater than 0.01 allele frequency in case subjects among all alleles. [d]D6S2811 was shown to be a statistically significant locus. The *P*-value for significance after Bonferroni correction was below 1.98×10^{-4} in all statistical tests (252 tests).

Supplementary Table 5

Allelic association analysis of *HLA-C* gene

	Allele frequency		Allelic association		
Allele	Case	Cont	OR	(95% CI)	*P*
*C*01:02*	0.1579	0.1821	0.84	0.60 - 1.17	2.92×10^{-1}
*C*01:03*	0.0000	0.0054	0.00	- -	3.45×10^{-1}
*C*03:02*	0.0146	0.0063	2.09	0.71 - 6.16	2.63×10^{-1}
*C*03:03*	0.1491	0.1295	1.18	0.83 - 1.68	3.49×10^{-1}
*C*03:04*	0.0848	0.1268	0.62	0.41 - 0.96	2.51×10^{-2}
*C*04:01*	0.0819	0.0357	2.47	1.48 - 4.11	5.33×10^{-4}
*C*05:01*	0.0029	0.0036	0.82	0.09 - 7.36	8.55×10^{-1}
*C*06:02*	0.0000	0.0054	0.00	- -	3.45×10^{-1}
*C*07:02*	0.1754	0.1232	1.48	1.07 - 2.04	1.96×10^{-2}
*C*07:04*	0.0117	0.0161	0.72	0.24 - 2.16	5.47×10^{-1}
*C*08:01*	0.0731	0.0857	0.84	0.52 - 1.33	4.45×10^{-1}
*C*08:03*	0.0117	0.0107	1.09	0.35 - 3.44	8.79×10^{-1}
*C*12:02*	0.0819	0.1143	0.70	0.46 - 1.06	8.42×10^{-2}
*C*12:03*	0.0000	0.0009	0.00	- -	8.42×10^{-1}
*C*14:02*	0.0468	0.0670	0.67	0.38 - 1.18	4.10×10^{-1}
*C*14:03*	0.0526	0.0652	0.80	0.47 - 1.35	7.11×10^{-1}
*C*15:02*	0.0556	0.0223	2.45	1.34 - 4.46	4.32×10^{-3}

Supplementary Table 6

Filtering of variants identified by MHC region sequencing

No of variant		Filtering parameters
77,040	←	Total variants obserbed in 12 individuals
↓	→	Excluding variants in segmental duplication regions
60,255		
↓	→	Excluding variants in control individuals
32,256		
↓	→	Excluding variants in individuals with non-risk haplotype
9,290		
↓	←	Selecting heterozygous variants in individuals with risk haplotype
3,895		

Supplementary Table 7

Overview of 16 variants identical in 5 individuals with risk haplotype

Physical position						Annotation by	1000 Genomes[d]			Sequencing reads in resequencing by NGS										Mean of
										AA083		AA086		AA211		AA215		AA267		
Start	End	Ref[a]	Alt[b]	SNV ID	Function (gene)	RepeatMasker[c]	CEU	CHB	JPT	Ref[e]	Alt[f]	Ref[e]	Alt[f]	Ref[e]	Alt[f]	Ref[e]	Alt[f]	Ref[e]	Alt[f]	total reads
31112701	31112701	G	A	rs142986308	exonic (*CCHCR1*)	Non	A=0.0000	A=0.0309	A=0.0169	86	117	114	126	56	97	123	115	44	83	192
31162402	31162402	C	T	rs140726956	intergenic	L2b	T=0.0000	T=0.0309	T=0.0112	425	329	387	400	431	476	378	438	230	243	747
31253444	31253444	C	T	rs9461684	intergenic	Non	T=0.1000	T=0.0722	T=0.0169	189	161	144	138	171	176	136	178	83	108	297
31253866	31253866	G	A	rs9468919	intergenic	Non	A=0.1000	A=0.0722	A=0.0169	250	208	156	157	186	194	210	156	113	100	346
31255762	31255762	A	C	rs28577989	intergenic	Non	C=0.0765	C=0.0670	C=0.0169	269	268	332	281	198	239	247	203	161	174	474
31255809	31255809	A	G	rs28506073	intergenic	Non	G=0.1000	G=0.0722	G=0.0169	289	263	306	291	225	218	252	233	164	187	486
31260888	31260888	G	A	rs60582327	intergenic	L1MB8	A=0.1000	A=0.0722	A=0.0169	103	125	104	117	100	106	99	101	48	70	195
31272553	31272553	G	A	rs9264904	intergenic	MLT1F2	A=0.2765	A=0.1907	A=0.1011	423	422	405	368	380	311	299	301	270	287	693
31274582	31274582	G	A	rs9468944	intergenic	Non	A=0.1353	A=0.0825	A=0.0169	111	114	85	100	97	91	93	76	87	47	180
31435267	31435267	C	T	rs147602969	intergenic	ERV3-16A3_I-int	T=0.0000	T=0.0206	T=0.0169	244	239	233	241	171	227	259	288	175	193	454
31482200	31482200	G	T	rs138518912	intergenic	L1M5	T=0.0000	T=0.0258	T=0.0281	74	96	94	72	91	100	88	70	71	44	160
31510328	31510343	TGTCCGAGGTAGCTGA	Deletion	rs201744045	intergenic	Non	Del=0.0000	Del=0.0258	Del=0.0169	125	62	127	47	70	45	146	50	63	41	155
31530306	31530306	T	C	rs142636125	intergenic	Non	C=0.0000	C=0.0258	C=0.0169	117	109	115	100	78	87	162	139	110	83	220
31537331	31537331	A	G	rs185544867	intergenic	AluSx	G=0.0000	G=0.0258	G=0.0337	41	58	41	40	45	71	77	66	23	41	101
31637474	31637474	C	T	rs140257968	intronic (*CSNK2B*)	Non	T=0.0000	T=0.0155	T=0.0169	228	180	266	186	118	90	257	253	110	109	359
31764424	31764424	C	T	rs150974840	intergenic	AluSx	T=0.0000	T=0.0155	T=0.0169	18	15	21	15	11	16	14	21	18	8	31

[a]Reference sequence in the UCSC Genome Browser. [b]Sequence observed in 5 individual with risk haplotype. [c]Name of repetitive sequence annotated by RepeatMasker (http://genome.ucsc.edu/). [d]Allele frequencies in each population in 1000 genomes Browser (http://www.ncbi.nlm.nih.gov/variation/tools/1000genomes/). [e]Sequence read number of reference sequence. [f]Sequence read number of observed variant.

Supplementary Table 8

SNV discovery and allelic association with *CCHCR1*

SNV	Physical position	Exon	Substitution[a]		Allele	Predicted Coiled-coil[c]	Functional predictions			Frequency	Allele frequency[e]		Allelic association		
			cDNA[b]	Amino acid			PolyPhen2	SIFT	PhyloP	in EA[d]	Case	Control	OR	(95% CI)	*P*
rs72856718	31125257	01	c.G121T	p.Glu41X	T	Non	NA	NA	Non	0.0854	0.0877	0.0375	2.47	1.51 - 4.05	4.25×10^{-4}
rs3130453	31124849	02	c.G234A	p.Trp78X	A	Non	NA	NA	Non	0.4817	0.3684	0.3545	1.06	0.83 - 1.36	6.45×10^{-1}
rs130075	31122502	04	c.G305A	p.Arg102Gln	A	Coiled-coil	Probably damaging	Tolerated	Conserved	0.0501	0.0906	0.1304	0.67	0.45 - 1.01	4.54×10^{-2}
rs130065	31122500	04	c.C307T	p.Arg103Trp	T	Coiled-coil	Benign	Tolerated	Conserved	0.2050	0.0000	0.0054	-	- - -	3.45×10^{-1}
rs130076	31122482	04	c.C325T	p.Arg109Trp	T	Coiled-coil	Benign	Tolerated	Conserved	0.2100	0.0000	0.0054	-	- - -	3.45×10^{-1}
rs130066	31122315	04	c.C492G	p.Ser164Arg	G	Coiled-coil	Benign	Tolerated	Non	0.4288	0.3626	0.3500	1.05	0.82 - 1.35	6.78×10^{-1}
rs11540822	31118898	05	c.T536A	p.Leu179Gln	A	Coiled-coil	Possibly damaging	Damaging	Conserved	0.0855	0.0819	0.0375	2.41	1.45 - 4.03	1.08×10^{-3}
rs130067	31118511	06	c.A825C	p.Glu275Asp	C	Coiled-coil	Benign	Tolerated	Non	0.1973	0.3596	0.3446	1.06	0.83 - 1.36	6.19×10^{-1}
rs2027937	31116502	09	c.G1099A	p.Ala367Thr	A	Coiled-coil	Benign	Tolerated	Conserved	0.0725	0.0439	0.0223	2.06	1.06 - 4.00	3.95×10^{-2}
rs130068	31116246	10	c.C1249T	p.Arg417Trp	T	Coiled-coil	Benign	Tolerated	Non	0.4444	0.2895	0.2571	1.19	0.90 - 1.57	2.24×10^{-1}
rs130069	31116245	10	c.G1250A	p.Arg417Gln	A	Coiled-coil	Benign	Tolerated	Conserved	0.0484	0.0029	0.0045	0.65	0.08 - 5.63	6.85×10^{-1}
rs147733073	31113239	12	c.G1458C	p.Gln486His	C	Non	Probably damaging	Damaging	Conserved	0.0002	0.0497	0.0679	0.74	0.44 - 1.24	3.95×10^{-1}
rs2073720	31112823	14	c.A1637G	p.Lys546Arg	G	Coiled-coil	Benign	Tolerated	Non	0.0026	0.1023	0.1250	0.79	0.53 - 1.18	2.44×10^{-1}
rs750668764	31112755	14	c.C1705A	p.Leu569Met	A	Coiled-coil	Possibly damaging	Damaging	Non	Non	0.0117	0.0027	4.45	0.99 - 20.1	5.43×10^{-2}
rs130079	31112737	14	c.G1723T	p.Gly575Cys	T	Coiled-coil	Benign	Tolerated	Non	0.2362	0.0000	0.0054	-	- - -	1.00
rs142986308	31112701	14	c.C1759T	p.Arg587Trp	T	Coiled-coil	Probably damaging	Damaging	Conserved	0.0001	0.0760	0.0250	3.41	1.94 - 5.99	3.39×10^{-5}
rs202129359	31112532	15	c.G1832A	p.Arg611Gln	A	Coiled-coil	Probably damaging	Damaging	Non	Non	0.0000	0.0027	-	- - -	1.00
rs130072	31112484	15	c.G1880A	p.Arg627Gln	A	Coiled-coil	Probably damaging	Damaging	Conserved	0.0867	0.0877	0.0375	2.47	1.51 - 4.05	4.25×10^{-4}
rs130074	31111174	16	c.G1917T	p.Gln639His	T	Coiled-coil	Probably damaging	Damaging	Non	0.0233	0.1667	0.1920	0.84	0.60 - 1.16	2.81×10^{-1}
rs116969494	31111148	16	c.A1943T	p.Lys648Met	T	Coiled-coil	Probably damaging	Damaging	Conserved	0.0002	0.0789	0.0955	0.81	0.52 - 1.26	3.48×10^{-1}
rs73397100	31111125	16	c.C1966T	p.Arg656Cys	T	Coiled-coil	Probably damaging	Damaging	Conserved	0.0004	0.0789	0.0955	0.81	0.52 - 1.26	3.48×10^{-1}
rs1576	31110391	18	c.C2327G	p.Ser776Cys	G	Non	Benign	Tolerated	Non	0.3228	0.0819	0.0429	2.09	1.26 - 3.45	5.20×10^{-3}

Sequencing analysis focused on variants resulting in amino acid substitution, though synonymous variants were also observed. [a]The positions of rs72856718 and rs3130453 were determined as based on transcript variant 2 (NM_001105563), while the positions of other variants were based on transcript variant 3 (NM_019052) (Supplementary Fig. 7.). [b]Sequences of cDNA for *CCHCR1*. [c]Coiled-coil structure prediction in the reference sequence (GRCh37/hg19) using COILS v2.2. [d]Allele frequencies in European Americans reported in NHLBI Exome Sequencing Project. [e]Allele frequencies from this study.

Supplementary Table 9
Haplotype analysis of 22 SNVs with amino acid substitution in *CCHCR1*

	SNV[a]																						Frequency			Haplotype association		
Haplotype	rs72856718	rs3130453	rs130075	rs130065	rs130076	rs130066	rs11540822	rs130067	rs2027937	rs130068	rs130069	rs147733073	rs2073720	rs750668764	rs130079	rs142986308	rs202129359	rs130072	rs130074	rs116969494	rs73397100	rs1576	Case	Control	OR	(95% CI)		*P*
Hap01	G	G	G	C	C	C	T	A	G	C	G	G	A	C	G	C	G	G	G	A	C	C	0.1667	0.1223	1.43	1.02	2.00	3.97×10^{-2}
Hap02	G	G	A	C	C	C	T	A	G	C	G	G	A	C	G	C	G	G	G	A	C	C	0.0906	0.1295	0.68	0.45	1.01	5.02×10^{-2}
Hap03	G	A	G	C	C	G	T	A	G	C	G	G	A	C	G	C	G	G	G	A	C	C	0.0029	0.0000	-	-	-	2.34×10^{-1}
Hap04	G	A	G	C	C	G	T	C	G	C	G	G	A	C	G	C	G	G	G	A	C	C	0.2865	0.3170	0.87	0.66	1.13	2.86×10^{-1}
Hap05	T	A	G	C	C	G	T	C	G	C	G	G	A	C	G	C	G	G	G	A	C	C	0.0058	0.0000	-	-	-	5.45×10^{-2}
Hap06	G	A	G	C	C	G	T	C	A	T	G	G	A	C	G	C	G	G	G	A	C	C	0.0439	0.0223	2.06	1.06	4.00	3.95×10^{-2}
Hap07	G	A	G	C	C	C	T	A	G	C	A	G	A	C	G	C	G	G	G	A	C	C	0.0029	0.0045	0.65	0.08	5.63	6.85×10^{-1}
Hap08	G	A	G	C	C	G	T	C	G	C	G	G	A	C	G	C	G	G	G	A	C	C	0.0029	0.0009	3.29	0.20	52.9	4.14×10^{-1}
Hap09	G	G	G	C	C	C	T	A	G	C	G	C	A	C	G	C	G	G	G	A	C	C	0.0468	0.0652	0.71	0.41	1.23	5.41×10^{-1}
Hap10	G	G	A	C	C	C	T	A	G	C	G	C	A	C	G	C	G	G	G	A	C	C	0.0000	0.0009	-	-	-	-	1.00
Hap11	G	A	G	C	C	G	T	C	G	C	G	G	A	A	G	C	G	G	G	A	C	C	0.0117	0.0027	4.45	0.99	20.1	5.43×10^{-2}
Hap12	G	A	G	C	C	G	T	C	G	C	G	G	A	C	G	C	G	G	G	A	C	C	0.0088	0.0018	4.98	0.83	30.1	7.94×10^{-2}
Hap13	G	G	G	C	C	C	T	A	G	C	G	G	A	C	G	C	A	G	G	A	C	C	0.0000	0.0027	-	-	-	-	1.00
Hap14	G	G	G	C	C	C	T	A	G	T	G	G	G	C	G	C	G	A	G	A	C	C	0.0029	0.0000	-	-	-	2.34×10^{-1}
Hap15	G	G	G	C	C	C	T	A	G	C	G	G	A	C	G	C	G	G	T	A	C	C	0.0029	0.0000	-	-	-	2.34×10^{-1}
Hap16	G	G	G	C	C	C	T	A	G	T	G	G	A	C	G	C	G	G	T	A	C	C	0.0585	0.0670	0.86	0.51	1.45	7.67×10^{-1}
Hap17	G	A	G	C	C	C	T	A	G	T	G	G	A	C	G	C	G	G	T	A	C	C	0.0029	0.0000	-	-	-	2.34×10^{-1}
Hap18	G	G	G	C	C	C	T	A	G	C	G	C	A	C	G	C	G	G	T	A	C	C	0.0029	0.0000	-	-	-	2.34 x 10-1
Hap19	G	G	G	C	C	C	T	A	G	T	G	G	G	C	G	C	G	G	T	A	C	C	0.0965	0.1241	0.75	0.5	1.12	1.51 x 10-1
Hap20	G	G	G	C	C	C	T	A	G	T	G	C	G	C	G	C	G	G	T	A	C	C	0.0000	0.0009	-	-	-	-	1.00
Hap21	G	G	G	C	C	C	T	A	G	T	G	G	G	C	G	C	G	A	T	A	C	C	0.0029	0.0000	-	-	-	2.34×10^{-1}
Hap22	G	G	G	C	C	C	T	A	G	C	G	G	A	C	G	C	G	G	G	T	T	C	0.0789	0.0946	0.82	0.53	1.27	3.74×10^{-1}
Hap23	G	G	G	C	C	C	T	A	G	C	G	C	A	C	G	C	G	G	G	T	T	C	0.0000	0.0009	-	-	-	-	1.00
Hap24	G	A	G	T	T	G	T	A	G	T	G	G	A	C	T	C	G	G	G	A	C	G	0.0000	0.0054	-	-	-	-	3.45×10^{-1}
Hap25	T	G	G	C	C	C	A	A	G	T	G	G	A	C	G	C	G	A	G	A	C	G	0.0058	0.0125	0.46	0.10	2.05	2.66×10^{-1}
Hap26	T	G	G	C	C	C	A	A	G	T	G	G	A	C	G	T	G	A	G	A	C	G	0.0760	0.0250	3.41	1.94	5.99	3.39×10^{-5}

[a]All SNVs with amino acid substitution used in analysis of haplotypes. Gray area indicates relationship between rs142986308 and Hap26.

Supplementary Table 10

Allele frequency of rs142986308 in gnomAD v2.1.1 (https://gnomad.broadinstitute.org/)

Population	Allele Count	Allele Nunmber	Allele Frequency
Eest Asian	553	19946	0.02772
South Asian	11	30616	0.00028
African	1	24934	0.00004
European (non-Finnish)	4	129060	0.00003
Latino	0	35432	0.00000
Ashkenazi Jewish	0	10350	0.00000
European (Finnish)	0	24126	0.00000

Supplementary Table 11

Homology of amino acid sequence to Hap26 in various species

	Hap25	Pan troglodytes	Macaca mulatta	Canis lupus familiaris	Bos taurus	Mus musculus	Xenopus tropicalis
		NP_001009009	NP_001108422	XP_532064	NP_001019707	NP_666360	NP_001116918
Identity (%)	99.87	98.59	95.50	83.61	63.50	72.96	32.51

Supplementary Table 12
List of 265 probes showing greater than 2-fold up- or down-regulation in Cchcr1 mice as compared to wild type.

Rank[a]	ProbeName	GeneSymbol	Regulation	AA mouse 01	AA mouse 02	Chr.	GeneName (type of keratin)	GenbankAccession
				Fold change	Fold change			
1	A_51_P221823	*Krtap21-1*	up	3,298	11,957	16	keratin associated protein 21-1	NM_028621
2	A_51_P380991	*Krt34*	up	3,019	9,125	11	keratin 34 (Hair keratin)	NM_027563
3	A_55_P2095054	*Krtap6-1*	up	2,522	6,552	16	keratin associated protein 6-1	NM_010672
4	A_52_P321831	*Krtap3-1*	up	2,402	6,325	11	keratin associated protein 3-1	NM_023511
5	A_55_P2151225	*Krtap16-1*	up	1,938	6,126	16	keratin associated protein 16-1	NM_130870
6	A_55_P1985648	*Krtap6-2*	up	1,861	5,741	16	keratin associated protein 6-2	NM_010673
7	A_51_P100624	*Krtap9-3*	up	1,966	4,668	11	keratin associated protein 9-3	NM_029351
8	A_55_P2154994	*Krtap28-13*	up	1,297	5,534	1	keratin associated protein 28-13	XM_006543615
9	A_55_P2088755	*Krtap4-1*	up	1,755	3,959	11	keratin associated protein 4-1	NM_001048196
10	A_51_P104681	*Krtap6-5*	up	1,709	3,845	16	keratin associated protein 6-5	NM_130856
11	A_52_P463962	*Krtap16-3*	up	1,319	4,169	16	keratin associated protein 16-3	NM_183296
12	A_55_P2032930	*Krtap3-3*	up	1,281	3,848	11	keratin associated protein 3-3	NM_025524
13	A_55_P2092475	*Krtap20-2*	up	1,293	3,687	16	keratin associated protein 20-2	NM_001163615
14	A_51_P172424	*Krtap4-16*	up	1,255	2,507	11	keratin associated protein 4-16	NM_001013823
15	A_51_P132400	*Krtap14*	up	881	2,568	16	keratin associated protein 14	NM_013707
16	A_51_P189899	*Olfr1134*	up	784	2,816	2	olfactory receptor 1134	NM_147030
17	A_55_P2140022	*Krt35*	up	693	2,093	11	keratin 35 (Hair keratin)	NM_016880
18	A_55_P2086334	*Krt85*	up	729	1,906	15	keratin 85 (Hair keratin)	NM_016879
19	A_55_P1966450	*Krt83*	up	570	1,926	15	keratin 83 (Hair keratin)	NM_001003668
20	A_55_P2006812	*Krtap11-1*	up	596	1,697	16	keratin associated protein 11-1	NM_001113406
21	A_55_P2154987	*Krtap5-2*	up	431	2,278	7	keratin associated protein 5-2	NM_027844
22	A_51_P269721	*Krtap28-13*	up	442	1,647	1	keratin associated protein 28-13	XM_896507
23	A_55_P2042161	*Krtap26-1*	up	426	1,593	16	keratin associated protein 26-1	NM_027105
24	A_55_P2009001	*Gprc5d*	up	441	1,510	6	G protein-coupled receptor, family C, group 5, member D	NM_001205396
25	A_55_P2005525	*Krtap12-1*	up	239	1,414	10	keratin associated protein 12-1	NM_010670
26	A_51_P475816	*Krtap2-4*	up	373	819	11	keratin associated protein 2-4	NM_027800
27	A_55_P2027213	*Krtap20-2*	up	315	876	16	keratin associated protein 20-2	NM_001163615
28	A_51_P450549	*Padi3*	up	307	842	4	peptidyl arginine deiminase, type III	NM_011060
29	A_55_P2017347	*Krtap11-1*	up	232	560	16	keratin associated protein 11-1	NM_001113406
30	A_55_P2070940	*Krtap20-2*	up	205	583	16	keratin associated protein 20-2	NM_001163615
31	A_51_P468456	*S100a3*	up	181	624	3	S100 calcium binding protein A3	NM_011310
32	A_51_P501873	*Krt26*	up	219	499	11	keratin 26 (Inner root sheath in hair follicle)	NM_001033397
33	A_55_P1985298	*Krtap5-2*	up	181	508	7	keratin associated protein 5-2	NM_027844
34	A_52_P468068	*Tchh*	up	221	405	3	trichohyalin	NM_001163098
35	A_55_P2158067	*Krtap4-16*	up	191	438	11	keratin associated protein 4-16	NM_001013823
36	A_55_P2110698	*Krtap4-9*	up	203	343	11	keratin associated protein 4-9	NM_001085548
37	A_66_P138462	*Sprr2h*	up	192	286	3	small proline-rich protein 2H	NM_011474
38	A_55_P2091831	*Krtap1-4*	up	146	280	11	keratin associated protein 1-4	NM_001039502
39	A_55_P1976972	*Krt28*	up	143	276	11	keratin 28 (Inner root sheath in hair follicle)	NM_027574
40	A_55_P2145626	*Krt82*	up	95	413	15	keratin 82 (Hair keratin)	NM_053249
41	A_55_P2178698	*Gprc5d*	up	115	333	6	G protein-coupled receptor, family C, group 5, member D	NM_053118
42	A_51_P136680	*Krtap31-1*	up	80	475	11	keratin associated protein 31-1	NM_027568
43	A_66_P135018	*Krtap5-1*	up	112	323	7	keratin associated protein 5-1	NM_015808
44	A_55_P2006311	*Krtap9-1*	up	141	226	11	keratin associated protein 9-1	NM_015741
45	A_52_P90805	*Lrrc15*	up	117	265	16	leucine rich repeat containing 15	NM_028973
46	A_66_P114558	*Krtap1-3*	up	127	238	11	keratin associated protein 1-3	NM_001085526
47	A_55_P2139703	*Krtap5-4*	up	77	315	7	keratin associated protein 5-4	NM_015809
48	A_55_P2009732	*Prr9*	up	105	220	3	proline rich 9	NM_175424
49	A_55_P2140042	*Krt31*	up	88	247	11	keratin 31 (Hair keratin)	NM_010659
50	A_55_P2060011	*Krtap5-5*	up	77	271	7	keratin associated protein 5-5	NM_001037822
51	A_55_P2111172	*Mecom*	up	57	335	3	MDS1 and EVI1 complex locus	NM_021442
52	A_51_P493437	*Krtap28-10*	up	77	234	1	keratin associated protein 28-10	AK160243
53	A_55_P1973970	*Krt73*	up	82	195	15	keratin 73 (Inner root sheath in hair follicle)	NM_212485
54	A_51_P474701	*Fbp1*	up	74	212	13	fructose bisphosphatase 1	NM_019395
55	A_51_P259694	*Pinlyp*	up	82	172	7	phospholipase A2 inhibitor and LY6/PLAUR domain containing	NM_001037143
56	A_55_P2095859	*Rdh18-ps*	up	39	290	10	retinol dehydrogenase 18, pseudogene	NR_037604
57	A_55_P1985304	*Krtap5-3*	up	49	230	7	keratin associated protein 5-3	NM_023860
58	A_55_P2043639	*Rpl6*	up	38	250	5	ribosomal protein L6	NM_011290
59	A_55_P2403159	*Fam26d*	up	66	138	10	family with sequence similarity 26, member D	NM_001081165
60	A_55_P2052145	*Ly6g6d*	up	46	197	17	lymphocyte antigen 6 complex, locus G6D	NM_033478
61	A_55_P1964672	*Krt28*	up	57	136	11	keratin 28 (Inner root sheath in hair follicle)	NM_027574
62	A_55_P1966445	*Krt81*	up	47	160	15	keratin 81 (Hair keratin)	NM_001166157
63	A_55_P1997126	*Ctse*	up	62	110	1	cathepsin E	NM_007799
64	A_55_P2055727	*Vmn1r52*	up	48	140	6	vomeronasal 1 receptor 52	NM_053222
65	A_55_P2071716	*Klre1*	up	25	201	6	killer cell lectin-like receptor family E member 1	NM_153590
66	A_55_P2030269	*Dnah6*	up	26	158	6	dynein, axonemal, heavy chain 6	NM_001164669
67	A_55_P2047778	*Krtap4-8*	up	43	93	11	keratin associated protein 4-8	NM_001085547
68	A_55_P2006436	*Olfr282*	up	27	150	15	olfactory receptor 282	NM_146457
69	A_51_P264695	*Crym*	up	42	91	7	crystallin, mu	NM_016669
70	A_55_P2114308	*Krt40*	up	29	113	11	keratin 40 (Hair keratin)	NM_001039666
71	A_55_P1988714	*Psors1c2*	up	31	95	17	psoriasis susceptibility 1 candidate 2 (human)	NM_020576
72	A_51_P338031	*Trpm1*	up	40	70	7	transient receptor potential cation channel, subfamily M, member 1	NM_018752
73	A_51_P143712	*Tdh*	up	43	62	14	L-threonine dehydrogenase	NM_021480
74	A_55_P1961477	*Kcnj11*	up	20	128	7	potassium inwardly rectifying channel, subfamily J, member 11	NM_010602
75	A_52_P534583	*Ahsp*	up	44	56	0	alpha hemoglobin stabilizing protein	NM_133245
76	A_55_P2165439	*Krt36*	up	30	79	11	keratin 36 (Hair keratin)	NM_001174099
77	A_55_P2173768	*Pkhd1*	up	21	107	1	polycystic kidney and hepatic disease 1	AK143992
78	A_55_P2114492	*Icmt*	up	18	115	4	isoprenylcysteine carboxyl methyltransferase	NM_133788
79	A_52_P163660	*Krtap3-2*	up	27	76	11	keratin associated protein 3-2	NM_025720
80	A_55_P2065159	*Krtap9-5*	up	23	89	11	keratin associated protein 9-5	NM_001085527
81	A_52_P383089	*Cd4*	up	15	105	6	CD4 antigen	NM_013488
82	A_51_P480202	*Dlx2*	up	24	50	2	distal-less homeobox 2	NM_010054
83	A_55_P2140571	*Krtap24-1*	up	22	51	16	keratin associated protein 24-1	NM_001163141
84	A_55_P2140057	*Krt32*	up	18	59	11	keratin 32 (Hair keratin)	NM_001159374
85	A_55_P2134938	*Krtap10-4*	up	15	70	10	keratin associated protein 10-4	NM_001135991
86	A_51_P183561	*Csdc2*	up	17	51	15	cold shock domain containing C2, RNA binding	NM_145473
87	A_51_P484158	*Steap1*	up	20	38	5	six transmembrane epithelial antigen of the prostate 1	NM_027399
88	A_55_P2109407	*Rnase12*	up	20	35	14	ribonuclease, RNase A family, 12 (non-active)	NM_001011875
89	A_52_P482251	*Gjb6*	up	20	34	14	gap junction protein, beta 6	NM_001010937
90	A_55_P1958919	*Olfr1347*	up	10	67	7	olfactory receptor 1347	NM_146385
91	A_55_P2002477	*Zfp707*	up	11	58	15	zinc finger protein 707	NM_001081065
92	A_55_P2181634	*Spef2*	up	7.9	78	15	sperm flagellar 2	XM_006520087
93	A_55_P2043777	*Trpm1*	up	13	47	7	transient receptor potential cation channel, subfamily M, member 1	BC082560
94	A_51_P324814	*Krt18*	up	15	38	15	keratin 18 (Epithelial keratins)	NM_010664
95	A_55_P2078960	*Tdh*	up	19	29	14	L-threonine dehydrogenase	NM_021480
96	A_55_P1998456	*Birc7*	down	39	14	2	baculoviral IAP repeat-containing 7 (livin)	NM_001163247
97	A_55_P2038752	*Olfr18*	up	11	48	9	olfactory receptor 18	NM_146563
98	A_55_P2081283	*Dlx4*	up	17	32	11	distal-less homeobox 4	NM_007867
99	A_51_P468073	*Ggt1*	up	16	31	10	gamma-glutamyltransferase 1	NM_008116
100	A_55_P2092671	*Nppc*	up	15	31	1	natriuretic peptide type C	NM_010933
101	A_55_P2000034	*Cryba4*	up	13	36	5	crystallin, beta A4	NM_021351

102	A_52_P498193	*Aldh1l2*	down	28	15	10	aldehyde dehydrogenase 1 family, member L2	NM_153543
103	A_51_P108659	*Pon1*	down	17	24	6	paraoxonase 1	NM_011134
104	A_51_P205385	*Uox*	up	12	33	3	urate oxidase	NM_009474
105	A_55_P2162379	*Actbl2*	up	12	29	13	actin, beta-like 2	NM_175497
106	A_55_P2062469	*Col12a1*	up	15	20	9	collagen, type XII, alpha 1	NM_001290308
107	A_52_P355169	*Tnc*	up	16	19	4	tenascin C	NM_011607
108	A_51_P304397	*Cpm*	up	12	25	10	carboxypeptidase M	NM_027468
109	A_55_P1985288	*Bhlha9*	up	7.2	40	11	basic helix-loop-helix family, member a9	NM_177182
110	A_51_P362066	*Chil1*	up	16	18	1	chitinase-like 1	NM_007695
111	A_52_P69020	*Slc24a5*	up	10	27	2	solute carrier family 24, member 5	NM_175034
112	A_66_P132496	*Asic3*	up	12	21	5	acid-sensing (proton-gated) ion channel 3	AY261387
113	A_51_P112734	*Slc7a8*	up	10	23	14	solute carrier family 7 (cationic amino acid transporter, y+ system), member 8	NM_016972
114	A_65_P08727	*Adamts18*	up	12	17	8	a disintegrin-like and metallopeptidase (reprolysin type) with thrombospondin type 1 motif, 18	AK081122
115	A_52_P62121	*Gpr37*	up	13	16	6	G protein-coupled receptor 37	NM_010338
116	A_55_P2172096	*Mc1r*	up	7.9	25	8	melanocortin 1 receptor	NM_008559
117	A_55_P2116435	*Gpr143*	up	10	19	X	G protein-coupled receptor 143	NM_010951
118	A_55_P2110076	*C1qtnf6*	down	11	16	15	C1q and tumor necrosis factor related protein 6	NM_001204153
119	A_55_P2041828	*Tubb3*	up	8.7	19	8	tubulin, beta 3 class III	NM_023279
120	A_51_P486810	*Gpx2*	up	10	17	12	glutathione peroxidase 2	NM_030677
121	A_55_P2027831	*Cryba4*	up	6.9	22	5	crystallin, beta A4	XM_006534748
122	A_51_P142767	*Glt8d2*	down	15	10	10	glycosyltransferase 8 domain containing 2	NM_029102
123	A_52_P399934	*Dusp2*	up	7.9	19	2	dual specificity phosphatase 2	NM_010090
124	A_55_P2414619	*Cdh6*	up	11	13	15	cadherin 6	XM_006520017
125	A_55_P2084965	*Klk13*	up	11	13	7	kallikrein related-peptidase 13	NM_001039042
126	A_55_P1953573	*Tyrp1*	up	11	14	4	tyrosinase-related protein 1	NM_031202
127	A_55_P1989519	*Fndc1*	down	6.9	21	17	fibronectin type III domain containing 1	NM_001081416
128	A_55_P2100375	*Col11a1*	up	9.1	15	3	collagen, type XI, alpha 1	NM_007729
129	A_55_P2010197	*Serpina10*	up	4.9	28	12	serine (or cysteine) peptidase inhibitor, clade A (alpha-1 antiproteinase, antitrypsin), member 10	NM_144834
130	A_52_P184042	*Adamts2*	down	12	12	11	a disintegrin-like and metallopeptidase (reprolysin type) with thrombospondin type 1 motif, 2	NM_175643
131	A_52_P151393	*Lrrc75b*	up	8.4	16	10	leucine rich repeat containing 75B	NM_198860
132	A_51_P425071	*C1qtnf3*	down	10	13	15	C1q and tumor necrosis factor related protein 3	NM_030888
133	A_51_P419637	*Dclk3*	up	7.4	17	9	doublecortin-like kinase 3	NM_172928
134	A_51_P282760	*Per2*	down	4.9	24	1	period circadian clock 2	NM_011066
135	A_55_P1973945	*Krt75*	up	7.1	16	15	keratin 75 (Hair follicle)	NM_133357
136	A_51_P455866	*Elf5*	up	8.3	13	2	E74-like factor 5	NM_010125
137	A_55_P2023118	*Prss28*	up	4.2	26	17	protease, serine 28	NM_053259
138	A_55_P1999301	*Cyp2e1*	down	11	10	7	cytochrome P450, family 2, subfamily e, polypeptide 1	NM_021282
139	A_51_P283968	*Adamts18*	up	6.4	17	8	a disintegrin-like and metallopeptidase (reprolysin type) with thrombospondin type 1 motif, 18	NM_172466
140	A_55_P1988368	*Upp1*	up	6.5	15	11	uridine phosphorylase 1	NM_009477
141	A_55_P1955778	*Adamts4*	up	8.2	12	1	a disintegrin-like and metallopeptidase (reprolysin type) with thrombospondin type 1 motif, 4	NM_172845
142	A_55_P2062573	*C1qtnf3*	down	11	8.8	15	C1q and tumor necrosis factor related protein 3	NM_001204134
143	A_51_P481398	*Kif11*	up	8.1	12	19	kinesin family member 11	NM_010615
144	A_51_P245414	*Klk1*	down	10	9.4	7	kallikrein 1	NM_010639
145	A_55_P2104219	*Hoxc13*	up	6.5	15	15	homeobox C13	NM_010464
146	A_55_P2043486	*Msx2*	up	5.8	16	13	msh homeobox 2	NM_013601
147	A_51_P123405	*Bub1*	up	8.0	11	2	budding uninhibited by benzimidazoles 1 homolog (S. cerevisiae)	NM_009772
148	A_55_P2086329	*Krt85*	up	5.4	16	15	keratin 85 (Hair keratin)	NM_016879
149	A_55_P2020532	*BC089491*	up	3.8	23	7	cDNA sequence BC089491	NM_175033
150	A_55_P1966690	*Cyp2e1*	down	9.2	9.4	7	cytochrome P450, family 2, subfamily e, polypeptide 1	NM_021282
151	A_66_P139618	*Stfa2*	up	7.4	12	16	stefin A2	NM_001082545
152	A_52_P627269	*Ces2b*	down	6.6	13	8	carboxyesterase 2B	NM_198171
153	A_52_P198898	*Samd5*	up	6.4	12	10	sterile alpha motif domain containing 5	NM_177271
154	A_52_P514061	*Padi4*	up	7.8	10	4	peptidyl arginine deiminase, type IV	NM_011061
155	A_55_P2135276	*Mcoln3*	up	4.7	16	3	mucolipin 3	NM_134160
156	A_55_P2002933	*Klk1b5*	down	16	4.6	7	kallikrein 1-related peptidase b5	NM_008456
157	A_52_P227391	*Kif15*	up	6.6	11	9	kinesin family member 15	NM_010620
158	A_55_P2279977	*D2Bwg0886e*	up	3.4	22	2	DNA segment, Chr 2, Brigham & Women's Genetics 0886 expressed	BE573844
159	A_55_P2023717	*Kcnq5*	down	12	5.6	1	potassium voltage-gated channel, subfamily Q, member 5	NM_023872
160	A_51_P486681	*Ap3b2*	up	7.4	8.9	7	adaptor-related protein complex 3, beta 2 subunit	NM_021492
161	A_55_P2090025	*Mest*	up	15	4.3	6	mesoderm specific transcript	NM_001252292
162	A_55_P2092219	*Serpina9*	up	5.0	12	12	serine (or cysteine) peptidase inhibitor, clade A (alpha-1 antiproteinase, antitrypsin), member 9	NM_027997
163	A_52_P520466	*Kif18b*	up	6.2	10	11	kinesin family member 18B	NM_197959
164	A_51_P162162	*Inmt*	down	9.0	6.4	6	indolethylamine N-methyltransferase	NM_009349
165	A_55_P2005956	*Egfbp2*	down	14	4.0	7	epidermal growth factor binding protein type B	NM_010115
166	A_51_P270949	*Hist1h1b*	up	5.4	10	13	histone cluster 1, H1b	NM_020034
167	A_51_P389888	*Slc40a1*	up	5.7	10	1	solute carrier family 40 (iron-regulated transporter), member 1	NM_016917
168	A_55_P1983769	*Birc5*	up	5.8	9.3	11	baculoviral IAP repeat-containing 5	NM_001012273
169	A_51_P154379	*Tmem150c*	down	8.8	6.0	5	transmembrane protein 150C	NM_182841
170	A_51_P367866	*Egr1*	up	5.1	10	18	early growth response 1	NM_007913
171	A_55_P1969276	*Hhip*	up	4.6	11	8	Hedgehog-interacting protein	NM_020259
172	A_55_P2025006	*Klk1b26*	down	11	4.5	7	kallikrein 1-related petidase b26	NM_010644
173	A_55_P1997141	*Mybl2*	up	4.7	10	2	myeloblastosis oncogene-like 2	NM_008652
174	A_52_P68028	*Ace*	down	10	4.6	11	angiotensin I converting enzyme (peptidyl-dipeptidase A) 1	NM_009598
175	A_55_P2069221	*Prr11*	up	6.2	7.6	11	proline rich 11	NM_175563
176	A_51_P292008	*Gpx3*	down	6.5	7.2	11	glutathione peroxidase 3	NM_008161
177	A_55_P1955656	*Ctla2a*	down	13	3.5	13	cytotoxic T lymphocyte-associated protein 2 alpha	NM_007796
178	A_51_P207892	*Pla2g5*	down	6.3	7.0	4	phospholipase A2, group V	NM_011110
179	A_55_P2066309	*Slc39a8*	up	5.0	8.6	3	solute carrier family 39 (metal ion transporter), member 8	NM_001135150
180	A_55_P2016014	*Cpsf4l*	up	5.0	8.5	11	cleavage and polyadenylation specific factor 4-like	NM_001164532
181	A_55_P2158011	*Knstrn*	up	5.5	7.6	2	kinetochore-localized astrin/SPAG5 binding	NM_026412
182	A_51_P451151	*Ube2c*	up	5.3	8.0	2	ubiquitin-conjugating enzyme E2C	NM_026785
183	A_66_P130035	*Klk1b24*	down	8.9	4.6	7	kallikrein 1-related peptidase b24	NM_010643
184	A_55_P1996941	*Ube2c*	up	5.6	7.2	2	ubiquitin-conjugating enzyme E2C	NM_026785
185	A_51_P413147	*Klk1b3*	down	7.3	5.3	7	kallikrein 1-related peptidase b3	NM_008693
186	A_55_P2039704	*S100a2*	up	4.3	8.4	3	S100 calcium binding protein A2	NM_001195760
187	A_51_P164014	*Cenpe*	up	4.7	7.6	3	centromere protein E	NM_173762
188	A_52_P447284	*Clic6*	up	3.6	10	16	chloride intracellular channel 6	NM_172469
189	A_55_P1996963	*Mtcl1*	up	4.4	7.9	17	microtubule crosslinking factor 1	NM_001114098
190	A_51_P419439	*Gnmt*	up	3.1	11	17	glycine N-methyltransferase	NM_010321
191	A_55_P2127934	*Palm3*	up	4.3	8.0	8	paralemmin 3	NM_028877
192	A_51_P489522	*Ctla2b*	down	10	3.3	13	cytotoxic T lymphocyte-associated protein 2 beta	NM_007797
193	A_52_P197402	*Tbc1d30*	up	4.3	7.8	10	TBC1 domain family, member 30	NM_029057
194	A_51_P329949	*Fam13a*	down	3.5	9.5	6	family with sequence similarity 13, member A	NM_153574
195	A_51_P415395	*C2cd4b*	up	3.9	8.5	9	C2 calcium-dependent domain containing 4B	NM_001081314
196	A_51_P486239	*Clec3b*	down	5.6	5.8	9	C-type lectin domain family 3, member b	NM_011606
197	A_52_P482897	*Areg*	up	3.3	9.6	5	amphiregulin	NM_009704
198	A_55_P2027386	*Gpr156*	up	5.5	5.8	16	G protein-coupled receptor 156	NM_153394
199	A_55_P2117614	*Tnfrsf13c*	up	4.4	7.2	15	tumor necrosis factor receptor superfamily, member 13c	NM_028075
200	A_52_P622850	*Hes5*	up	2.1	15	4	hairy and enhancer of split 5 (Drosophila)	NM_010419
201	A_51_P259603	*Adcyap1r1*	down	4.4	6.9	6	adenylate cyclase activating polypeptide 1 receptor 1	NM_007407
202	A_66_P115004	*Gabrp*	up	3.9	7.6	11	gamma-aminobutyric acid (GABA) A receptor, pi	NM_146017
203	A_51_P253803	*Mki67*	up	4.4	6.7	7	antigen identified by monoclonal antibody Ki 67	NM_001081117
204	A_52_P114905	*Ifitm10*	up	4.3	6.9	7	interferon induced transmembrane protein 10	BC049666
205	A_51_P179461	*Eid3*	up	5.2	5.5	10	EP300 interacting inhibitor of differentiation 3	NM_025499
206	A_55_P2072453	*Marc1*	up	8.2	3.5	1	mitochondrial amidoxime reducing component 1	NM_001290273

207	A_55_P2158510	*Dcpp2*	down	7.7	3.6	17	demilune cell and parotid protein 2	NM_001039238
208	A_52_P374897	*Arg2*	up	4.9	5.6	12	arginase type II	NM_009705
209	A_55_P2141093	*Eya2*	up	4.6	6.0	2	eyes absent 2 homolog (Drosophila)	NM_010165
210	A_51_P490924	*Hapln4*	down	4.5	5.9	8	hyaluronan and proteoglycan link protein 4	NM_177900
211	A_55_P2030368	*Tm7sf3*	down	4.3	6.0	6	transmembrane 7 superfamily member 3	NM_026281
212	A_55_P1983768	*Birc5*	up	4.4	5.8	11	baculoviral IAP repeat-containing 5	NM_009689
213	A_55_P1994781	*Vamp1*	down	6.6	3.7	6	vesicle-associated membrane protein 1	AK020615
214	A_51_P320304	*Plod1*	down	4.5	5.0	4	procollagen-lysine, 2-oxoglutarate 5-dioxygenase 1	NM_011122
215	A_52_P676819	*Bnc1*	up	4.4	5.1	7	basonuclin 1	NM_007562
216	A_51_P150653	*Ptprv*	up	3.8	5.9	1	protein tyrosine phosphatase, receptor type, V	NM_007955
217	A_52_P341886	*Lzts1*	up	2.8	8.0	8	leucine zipper, putative tumor suppressor 1	NM_199364
218	A_55_P2013019	*Lpar1*	down	4.1	5.4	4	lysophosphatidic acid receptor 1	NM_172989
219	A_55_P2035320	*Nfil3*	up	4.0	5.4	13	nuclear factor, interleukin 3, regulated	NM_017373
220	A_52_P639043	*Glt8d2*	down	5.8	3.7	10	glycosyltransferase 8 domain containing 2	NM_029102
221	A_55_P2060238	*Itgbl1*	down	6.2	3.3	14	integrin, beta-like 1	NM_145467
222	A_51_P474169	*Proser2*	up	2.9	7.2	2	proline and serine rich 2	NM_144883
223	A_66_P131110	*Zfp473*	up	4.7	4.4	7	zinc finger protein 473	NM_001289836
224	A_55_P2034625	*Sema6a*	up	5.0	4.0	18	sema domain, transmembrane domain (TM), and cytoplasmic domain, (semaphorin) 6A	AK082711
225	A_52_P263658	*Hes2*	up	3.0	6.6	4	hairy and enhancer of split 2 (Drosophila)	NM_008236
226	A_52_P58283	*Kcna5*	down	6.4	3.0	6	potassium voltage-gated channel, shaker-related subfamily, member 5	NM_145983
227	A_55_P2070373	*Ccdc155*	up	2.9	6.6	7	coiled-coil domain containing 155	NM_201374
228	A_51_P255699	*Mmp3*	down	6.9	2.7	9	matrix metallopeptidase 3	NM_010809
229	A_66_P121480	*Sardh*	up	3.1	5.4	2	sarcosine dehydrogenase	NM_138665
230	A_52_P145415	*Ptch2*	up	3.1	5.1	4	patched homolog 2	BC058397
231	A_55_P2178539	*Nacc2*	down	3.5	4.4	2	nucleus accumbens associated 2, BEN and BTB (POZ) domain containing	NM_001037098
232	A_66_P130911	*Proser2*	up	3.2	4.7	2	proline and serine rich 2	AK132370
233	A_55_P2219243	*Usp43*	up	3.9	3.8	11	ubiquitin specific peptidase 43	NM_173754
234	A_55_P1953030	*Tnfsf12*	down	2.2	6.8	11	tumor necrosis factor (ligand) superfamily, member 12	NM_011614
235	A_51_P482121	*Edn2*	up	3.1	4.5	4	endothelin 2	NM_007902
236	A_55_P2024439	*Gaa*	down	2.7	5.1	11	glucosidase, alpha, acid	NM_008064
237	A_55_P2167530	*Scube3*	up	2.7	5.1	17	signal peptide, CUB domain, EGF-like 3	XM_006524323
238	A_52_P401504	*Thbs4*	up	4.3	3.2	13	thrombospondin 4	NM_011582
239	A_55_P2105152	*Trim59*	up	3.1	4.3	3	tripartite motif-containing 59	NM_025863
240	A_66_P111021	*Nlrp6*	down	3.7	3.5	7	NLR family, pyrin domain containing 6	NM_133946
241	A_52_P35048	*Serpinf1*	down	3.0	4.4	11	serine (or cysteine) peptidase inhibitor, clade F, member 1	NM_011340
242	A_51_P295034	*Klk1b4*	down	3.7	3.0	7	kallikrein 1-related pepidase b4	NM_010915
243	A_55_P1974923	*Olfr568*	down	3.3	3.3	7	olfactory receptor 568	NM_147091
244	A_55_P1961499	*Ly6c1*	down	2.7	4.0	15	lymphocyte antigen 6 complex, locus C1	NM_001252058
245	A_51_P116651	*Dpt*	down	2.5	4.1	1	dermatopontin	NM_019759
246	A_55_P2106074	*Wfikkn1*	up	2.4	4.3	17	WAP, FS, Ig, KU, and NTR-containing protein 1	NM_001100454
247	A_51_P247184	*Npr3*	up	4.7	2.1	15	natriuretic peptide receptor 3	NM_008728
248	A_52_P281145	*Kank4*	up	3.9	2.6	4	KN motif and ankyrin repeat domains 4	NM_172872
249	A_51_P183894	*Fbxo15*	down	4.5	2.1	18	F-box protein 15	NM_015798
250	A_55_P2062437	*Begain*	up	2.4	3.9	12	brain-enriched guanylate kinase-associated	NM_001163175
251	A_55_P2085412	*Ankle1*	up	2.6	3.6	8	ankyrin repeat and LEM domain containing 1	NM_172756
252	A_55_P2035315	*Rasgef1b*	up	2.6	3.5	5	RasGEF domain family, member 1B	NM_145839
253	A_51_P200494	*Klk1b1*	down	2.4	3.6	7	kallikrein 1-related peptidase b1	NM_010645
254	A_55_P2042247	*Nlrp2*	down	4.2	2.0	7	NLR family, pyrin domain containing 2	NM_177690
255	A_55_P2061991	*Pnldc1*	up	2.6	3.3	17	poly(A)-specific ribonuclease (PARN)-like domain containing 1	NM_001034866
256	A_55_P2144075	*Pofut2*	down	2.6	3.3	10	protein O-fucosyltransferase 2	NM_030262
257	A_55_P1974527	*Ntsr2*	down	3.0	2.7	12	neurotensin receptor 2	NM_008747
258	A_55_P2181538	*Sult1d1*	down	3.7	2.1	5	sulfotransferase family 1D, member 1	NM_016771
259	A_55_P2185397	*Copz2*	down	2.2	3.4	11	coatomer protein complex, subunit zeta 2	NM_019877
260	A_51_P164939	*Tmem150a*	down	2.8	2.6	6	transmembrane protein 150A	NM_144916
261	A_52_P602091	*Csf1r*	down	2.3	3.2	18	colony stimulating factor 1 receptor	NM_001037859
262	A_55_P2162160	*Anp32a*	down	2.9	2.3	9	acidic (leucine-rich) nuclear phosphoprotein 32 family, member A	NM_009672
263	A_55_P2116647	*Myo1c*	down	2.6	2.5	11	myosin IC	NM_001080774
264	A_55_P2014755	*Pclo*	up	3.2	2.0	5	piccolo (presynaptic cytomatrix protein)	NM_001110796
265	A_66_P114614	*Ndst1*	down	2.5	2.5	18	N-deacetylase/N-sulfotransferase (heparan glucosaminyl) 1	NM_008306

[a] Fold change values, calculated using mean normalized expression values for both AA mice, are listed in descending order.

Supplementary Table 13
45 genes showing over 2-fold up- or down-regulation in AA patients with the T allele of rs142986308 as compared to an AA patient without the allele

Genesymbol	Fold change value		Regulation
	AA patient 1 (rs142986308:C/T)	AA patient 2	
ANGPTL7	9.00	8.23	up
KRT25	7.11	8.44	up
KRT74	6.23	8.66	up
KRT28	6.54	7.71	up
KRT71	4.53	4.47	up
KRT73	4.03	4.03	up
TCHH	4.03	3.76	up
KRT27	3.19	3.49	up
FABP9	2.74	3.52	up
AMTN	2.56	3.34	up
SLC7A5	2.69	3.00	up
PADI3	2.75	2.87	up
GJB6	2.78	2.39	up
ISOC1	2.31	2.74	up
SLC7A1	2.26	2.51	up
HIST2H2BA	2.25	2.39	up
KRTAP5-5	2.39	21.6	down
KRTAP5-6	2.39	10.8	down
KRTAP5-10	3.67	8.73	down
KRTAP10-11	3.26	8.79	down
KRTAP5-8	2.23	9.61	down
KRTAP10-4	2.79	8.22	down
KRTAP19-6	5.59	3.58	down
KRTAP13-2	2.53	6.52	down
KRTAP6-1	3.15	5.25	down
KRTAP10-3	2.04	5.27	down
KRTAP9-7	4.06	2.95	down
PLA2G2E	2.63	3.90	down
BMP2	2.37	3.58	down
PDK4	2.99	2.94	down
ZNF652	3.54	2.28	down
KRTAP20-1	2.59	3.04	down
KRTAP10-9	2.28	3.35	down
SNORD14A	2.90	2.32	down
KRTAP12-1	2.44	2.53	down
TNS4	2.68	2.22	down
KRTAP19-7	2.49	2.34	down
KRT83	2.76	2.03	down
OGFRL1	2.17	2.53	down
PLB1	2.01	2.60	down
KRTAP10-5	2.05	2.45	down
KRTAP5-2	2.10	2.35	down
LPCAT3	2.20	2.18	down
IGHD2-21	2.07	2.14	down
HIF1A	2.11	2.09	down