HABC Gentoype Data Submission Form

(assay-specific information for each of the 35 SNPs is on the attached table)

Submitting Investigator	
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Date Submitted	June 10, 2005
Polymorphism Information	
Name	See attached sheet for 35 different SNPs
RS# or Unique Identifier	See attached sheet
Gene	See attached sheet (gene context given if available)
Chromosome position	See attached sheet (position given in hg17 May 2004 assembly)
Alleles	See attached sheet for reference & variant alleles for 41 SNPs
Assay Information	
Genotyping Method	Sequenom MassArray
Genotypes in HWE (Y/N) (attach HWE Form)	Not possible to assess using ancestry informative SNPs in African Americans, because the markers are by their nature (highly differentiated in frequency across populations) out of Hardy-Weinberg equilibrium in an admixed populations
Amount of DNA used	5 ng per reaction
PCR primers	See attached sheet for each of the 35 SNPs.
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PCR Components (Taq, buffer, $MgCl_2$, primers, DMSO, other reagents)	Multiplex PCR reactions were performed using standard conditions in 384 well PCR plates (Eppendorf). The 6 uL PCR reactions contain 0.2 U of Taq polymerase (HotStar, Qiagen); 5 ng genomic DNA; 100 nM (0.6 pmol) of each PCR primer; and 200 uM (1.2 nmol) of each dNTP.
PCR Cycling conditions (time, temperature, # cycles)	Cycling conditions were initial denaturation at 95oC 15 minutes followed by 45 cycles of 95oC for 20 s, 56oC for 30s, 72oC for 30 s.
Detection Oligo(s) (if applicable)	See attached sheet for each of the 35 SNPs.
Other Reaction Conditions (detection reaction components, incubation conditions, gel %, etc.)	**Dephosphorylation protocol:** To remove residual unincorporated dNTPs from the PCR reaction, 2 uL mix of 0.3 U of Shrimp Alkaline Phosphatase (Amersham) and 5X Thermo Sequenase buffer (250 mM Tris, 10 mM MgCl2, pH 9.5) was added to the 6uL PCR reaction, followed by a 25 min incubation at 37oC and heat-inactivation of enzyme at 85oC for 10 min. **Primer Extension Protocol:** During the MassEXTEND reaction, a probe is extended by a specific number of nucleotides depending on the allele and the design of the assay. In the reaction mixture, all four nucleotides A, T, C, and G are present as either dNTPs or ddNTPs. Three nucleotides are present as ddNTPS and one as a dNTP. The incorporation of a ddNTP terminates the extension of the primer. For each type of SNP an optimal termination mix is used (C/T SNP uses A, C, G ddNTPs; C/G, A/G, and G/T SNPs use A, C, T ddNTPs; A/C use C, G, T ddNPTs; A/T uses C, G, T ddNTPs). Using a DNA polymerase that incorporates both dNTPs and ddNTPs, the reaction produces al;ele-specific extension products of different masses depending on the sequence analyzed. The reaction consists of 52 uM (5.2 nmol) of termination mix, 650 nM (~ 6 pmol) of each extension probe and 0.64 U of ThermoSequenase polymerase (Amersham Phamacia). 2 uL of the above reaction mixture was added to the dephosphorylated PCR products bringing the final reaction volume to 10 uL. Cycling conditions were 94 oC for 2 minutes, followed by 54 cycles of 94oC for 5 s, 50oC for 5 s, 72oC for 5 s.
Other Assay Info	MALDI-TOF MS analysis (automated genotype detection step): Approximately 7nl of the purified primer extension reaction was loaded onto a matrix pad (3-hydroxypicoloinicacid) of a SpectroCHIP (Sequenom, San Diego, CA). SpectroCHIPs were analyzed using a Bruker Biflex III MALDI-TOF (matrix-assisted laser desorption/ionization time-of-flight) mass spectrometer (SpectroREADER, Sequenom, San Diego, CA). The resulting mass spectra were processed by proprietary software (SpectroTYPER) to determine genotypes based on peaks intensities corresponding to the expected extension products.

rs#	Chromosome	hg17 position (May 2004 assembly)	Ref	Var	Primer 1	Primer 2
rs1011024	7	98842052	A	G	ACGTTGGATGGGACCAGAATACTGCATCAC	ACGTTGGATGTCACTGAATTGAACTGCCGG
rs10791800	11	105628804	G	T	ACGTTGGATGTGCTCTTGACAAAATCTGGC	ACGTTGGATGAAATCAGCTGGATACACCCC
rs11878536	19	47115315	C	G	ACGTTGGATGTATGTGTGGCTGTTGGATGG	ACGTTGGATGCCAGAAACAGCTTCACACAC
rs1374092	15	70595482	A	C	ACGTTGGATGGGTGGTGGAAACTAGGAATC	ACGTTGGATGTTGTTTCTAGCCACAAGTTC
rs1426654	15	46213776	A	G	ACGTTGGATGTTCAGCCCTTGGATTGTCTC	ACGTTGGATGAATTCAGGAGCTGAACTGCC
rs1454436	6	156941024	C	T	ACGTTGGATGAATGAAGCCTCTCTAGTCCC	ACGTTGGATGAGAGTGAAGGAGACCAACAG
rs1525760	4	117492984	C	T	ACGTTGGATGATAGAACAGACTGACCTCCC	ACGTTGGATGGGGAAGAGTTGCAGTTCAAG
rs1530044	4	166785016	A	T	ACGTTGGATGTGCAATCGGACACTTACTGG	ACGTTGGATGGCAATTGCAACACAGATATG
rs1861141	7	153498093	A	G	ACGTTGGATGCCAGAAGTGCTATGAGTTAC	ACGTTGGATGACATAAAGCCCTACCCAACC
rs1941141	18	7620982	A	G	ACGTTGGATGGTGCCATGTGAAAGTGAGAG	ACGTTGGATGTCCCCGGGATCTCTGAATTT
rs1956485	14	25933230	C	T	ACGTTGGATGGTAGACTGTAGCTATAGCCC	ACGTTGGATGCCTAACTGAACAACCCACTC
rs2060447	2	163426060	C	T	ACGTTGGATGGGTCATCAGCATTTTCAGGG	ACGTTGGATGTTACAGACACCAAGGGAGAG
rs2301550	9	20344796	A	G	ACGTTGGATGAGAAACGGAAGGCTTGTCAG	ACGTTGGATGGAATGGTGAATGTGACAAGG
rs2339475	2	29911441	A	G	ACGTTGGATGGTCAGAACCCTTGTTTCTGG	ACGTTGGATGTCCTTCAGGATACCACCTTG
rs2394931	10	74336071	A	G	ACGTTGGATGTAGCAACTTCTCAGGAACTC	ACGTTGGATGTCCTTTTGAGTCGTGGGTAC
rs2814778	1	155987756	C	T	ACGTTGGATGCATGGCACCGTTTGGTTCAG	ACGTTGGATGATGGCCCTCATTAGTCCTTG
rs2965404	7	21522742	C	T	ACGTTGGATGGATACTGTCACTTTTGGCAC	ACGTTGGATGTAGCATTGTGAGCAAAACTG
rs3024354	6	6237429	G	T	ACGTTGGATGAAATGGGAGGCATACAGCAC	ACGTTGGATGGTGGAAAATGCTAATGGAGG
rs3825663	14	89499502	C	T	ACGTTGGATGAAAGGCAGAAAAGCGGTTCC	ACGTTGGATGACTTTCTCAGCCTGCTTCTG
rs4242762	10	3797541	C	G	ACGTTGGATGAGCGGGCGGATGTTTTCATG	ACGTTGGATGTCTCTTCCATCAAGTTACCC
rs4436849	18	28809444	A	C	ACGTTGGATGTAATTCTGTATGGCGTGCCT	ACGTTGGATGCCAAAAGAAAACAGTGGACG
rs4759816	12	129980322	A	G	ACGTTGGATGGCGCCTGAAAATGGGTTCTG	ACGTTGGATGTACAGCTGAACTCCTTCTCC
rs4821667	22	36154581	A	C	ACGTTGGATGAGTTCCGGACCCAGGCAGCA	ACGTTGGATGGCAGCCTGCATTATTAAACC
rs6034866	20	17551728	A	G	ACGTTGGATGGCCTCTAGTTGTGAGTCAAG	ACGTTGGATGGATTGTGGTCACACATCCAG
rs6662385	1	72885596	C	T	ACGTTGGATGCATAGAACAATTACAGTCAGC	ACGTTGGATGTAGGTCGAACATGTATGTGG
rs680273	11	64296211	C	G	ACGTTGGATGGACTTCTTCACCTCCAACAG	ACGTTGGATGCAAACCTTCTTTGCTTCCTG
rs7201030	16	75081293	A	G	ACGTTGGATGAGTCACGGCTGGTCAATAAG	ACGTTGGATGGGAGCTAGTTGAGTAAGTGC
rs734908	1	53656058	G	T	ACGTTGGATGTGTACTGGTCCCTGGCTAAG	ACGTTGGATGCTGCTGAAGCTTCTGCTGCG
rs7865808	9	2903620	A	G	ACGTTGGATGGAAGCAGTGTGATTCTGTGG	ACGTTGGATGGACAGTTTTCATGCTCTCGG
rs803733	9	122912433	C	T	ACGTTGGATGACCAGTTTTCAACATTGCCG	ACGTTGGATGTAACACCTATAGCTGGAAAC
rs913607	13	105708805	C	G	ACGTTGGATGGCCCAAATCCCTGCTTTATC	ACGTTGGATGTTCTTCCAGAATGAGAGCAC
rs930072	5	36701828	C	T	ACGTTGGATGCTTCTGGCCCCTTTATAGAC	ACGTTGGATGTTCGGTTCCAGAACCCTTAG
rs9418990	10	135226847	A	G	ACGTTGGATGCACAAAAGTGGGTGAAGTGG	ACGTTGGATGTGGAAGCTGCCTTTGTAGGG
rs959071	17	19082819	C	T	ACGTTGGATGTCTAGAGCTACAAGAGGCAC	ACGTTGGATGCCAAGCTTCCTCTCATTATG
rs959354	11	129514791	A	G	ACGTTGGATGGCATTCCACCTATAGGAAGC	ACGTTGGATGGAGGAGAGTTGCTGTTCTTG

rs#	Detection_Probe	refs	hets	vars
rs1011024	CATCACTGCCAGGACAG	72	389	780
rs10791800	GTAGTATCTGAATTATCACTTAGC	54	389	851
rs11878536	TTGGATGGCGCATGTGT	615	509	110
rs1374092	GTTACTTTTGATAAACCAATAACA	131	487	601
rs1426654	GTCTCAGGATGTTGCAGGC	87	439	720
rs1454436	TCCTGTGCCCTCCTGCC	875	301	57
rs1525760	CTGACCTCCCCTGAGAAA	714	469	90
rs1530044	CCATAACGTTTTCCTTTATAC	666	498	124
rs1861141	TTATTCCAGTTTGTCCCCA	517	571	138
rs1941141	AAAGTGAGAGCCTGTGCCC	280	11	694
rs1956485	TGTAGCTATAGCCCAGTTAAGG	59	424	752
rs2060447	TTCAGGGTATGGCGAATCC	299	130	826
rs2301550	GAGCCCTCCTAGTAACAGACT	156	503	596
rs2339475	TAGGTCACAAGTAGTCCTG	59	359	861
rs2394931	AGTGAAATTCACCAATACAAATA	413	323	110
rs2814778	GCGCCTGTGCTTCCAAG	803	407	76
rs2965404	TCACAGAAGAAACATTCTAGATA	816	361	57
rs3024354	AGCACCTCTGCACAGCA	798	413	79
rs3825663	AGTGATGATGAGCTGCA	612	536	132
rs4242762	GATGTTTTCATGGCCAAAG	752	426	57
rs4436849	ATGGCGTGCCTACGGTAACTG	167	528	465
rs4759816	GGTTCTGCTAAAATCCC	146	531	540
rs4821667	CCCCCAGCATCTCCCTGGA	832	328	63
rs6034866	GAGTCAAGGCAAGCTGG	683	452	85
rs6662385	CAATTACAGTCAGCAAAGGT	837	351	46
rs680273	CCTCCAACAGGCATCGGCTA	77	446	706
rs7201030	GTCAATAAGCAAGGTAAGTAAACC	716	330	62
rs734908	AGGGCCCTGCCTCACAG	846	328	52
rs7865808	GGCAACCTAGAAGGAGAAAA	299	190	57
rs803733	ATTGCCGTTTATTCTCATCAT	61	369	812
rs913607	CTGCTTTATCAAATGCTATTTTAA	82	467	721
rs930072	GGCCCCTTTATAGACTAGCTT	734	375	82
rs9418990	GGTGAAGTGGAAACTGC	68	442	729
rs959071	CGTTTTCTGCCAAATCTTC	70	391	752
rs959354	AAGGCACCTTTCTCTAAAC	134	554	583

IL6R SNPs

rs	Chromosome	hg17 position (May 2004 assembly)	Ref	Var	Primer 1	Primer 2
rs11582424	1	151177058	A	C	ACGTTGGATGAATCATCTTGCCTCTGCCTC	ACGTTGGATGAAGGAAGTGCAGTGTGATGG
rs952146	1	151182001	A	G	ACGTTGGATGTGCTCTCCCCTTATATTCTC	ACGTTGGATGGGAAGAAGGAGTTAAGGATG
rs4845615	1	151188350	A	G	ACGTTGGATGTGAGCCAGAACACTTCTCAG	ACGTTGGATGCAATGTCTCCCAGCATTCAG
rs1552481	1	151189426	C	T	ACGTTGGATGAGAAAGATCCAGATGCACGG	ACGTTGGATGAATGGAACTCCCAAGTGGAC
rs4845617	1	151190971	A	G	ACGTTGGATGTCATGTGCGAGTGGGAAGTC	ACGTTGGATGTCTACACACACTGCGAGTCC
rs7549250	1	151217409	C	T	ACGTTGGATGCTGTTCTGAGAAGTGTGACG	ACGTTGGATGCGCAGGAATGCTGAACAAAG
rs4845625	1	151235140	C	T	ACGTTGGATGAGCTGTTGTCCTTACAGGTC	ACGTTGGATGTAGCTCGTAAGTGGTGGAAC
rs11265618	1	151243165	C	T	ACGTTGGATGCATCTTCGCTCCATTTGGTC	ACGTTGGATGTGAAGGCACTATTGGCTGAC
rs4329505	1	151245493	C	T	ACGTTGGATGGAATGTAGTGTATCATTCCGG	ACGTTGGATGGAACACCATGAAAGTCCCTC
rs7526293	1	151257282	C	T	ACGTTGGATGTGTGTGTGTGTAAGCATTC	ACGTTGGATGGGACATTGGGCAGCTTTATT
rs4845632	1	151272005	A	G	ACGTTGGATGAAAAACCCCATCCAGGCAAG	ACGTTGGATGCTTACAGCTTTGTATCAGGG

rs	Detection_Probe	refs	hets	vars
rs11582424	TACGTGTTGCTGGCATG	668	484	122
rs952146	ACATGAGCAAATGGTGGG	474	570	230
rs4845615	TCAGAGGAGATTGTGTATGT	16	195	1056
rs1552481	CCCAAGTGGACAGGGCCA	47	372	853
rs4845617	GGCTCCTCTCCCTCTGG	144	556	566
rs7549250	AGTGATGATTCATGTCCTGGG	223	640	408
rs4845625	GAACCAGCATTCCAGTCA	625	540	107
rs11265618	TTGGCTGACTTTTCTGCA	588	565	125
rs4329505	CAGCTGTGGAGGTAGCA	183	569	522
rs7526293	CTTAATTTAATAATGCATACACCT	476	599	208
rs4845632	TTGTATCAGGGAGCACA	91	513	661