

**Table S1.** *SLC44A2* amplification primers

Amplicon	Specificity	Name	Location*	Position†	Nucleotide Sequence (5'- 3')	Orientation	PCR conditions		Amplicon Size
							Thermocycling conditions‡	MgCl <sub>2</sub> & primer concentration	
1	Exon 1	QEX1AMF	5'UTR	-242..-221	AGCCTCAATATCTCCTTGACGA	sense	Denaturation: 95°C for 30 sec Annealing: 58°C for 30 sec Extension: 72°C for 3 min Number of cycles: 35	MgCl <sub>2</sub> : 1.5 mM Forward and reverse primers: 0.1 µM each	825 bp
		QEX1AMR	Intron 1	564.. 545	TTCGGTCTGCCAGTCTTGC	antisense			
2	Exon 2	QEX2AMF1	Intron 1	23,417..23,436	CTTACTCTCCCCTACCCGAA	sense	Denaturation: 95°C for 30 sec Annealing: 57°C for 30 sec Extension: 72°C for 3 min Number of cycles: 35	MgCl <sub>2</sub> : 1.5 mM Forward and reverse primers: 0.1 µM each	1001 bp
		QEX2AMR1	Intron 2	24,398.. 24,379	CAGAAGACGTGCTTGAACCA	antisense			
3	Exon 3-4	CL2-A3bF	Intron 2	24,440..24,457	GACTACTCAGAGGCTACT	sense	Denaturation: 95°C for 30 sec Annealing: 58°C for 30 sec Extension: 72°C for 3 min Number of cycles: 35	MgCl <sub>2</sub> : 2 mM Forward and reverse primers: 0.15 µM each	2017 bp
		CL2-A3bR	Intron 4	26,456.. 26,439	GGCACATCTAACCATCAA	antisense			
4	Exon 5-10	QEX5-10AMF	Intron 4	28,513..28,533	ATCACACCATTGCACTCCATC	sense	Denaturation: 95°C for 30 sec Annealing: 59°C for 30 sec Extension: 72°C for 3 min Number of cycles: 35	MgCl <sub>2</sub> : 1.5 mM Forward and reverse primers: 0.1 µM each	1630 bp
		QEX5-10AMR	Intron 10	30,122.. 30,102	TGCATCCTTAACCTACGGTGCT	antisense			
5	Exon 11-14	QEX11-14AMF	Intron 10	31,930..31,948	GAATCGCTTGAACCCGTGA	sense	Denaturation: 95°C for 30 sec Annealing: 60°C for 30 sec Extension: 72°C for 3 min Number of cycles: 35	MgCl <sub>2</sub> : 1.5 mM Forward and reverse primers: 0.1 µM each	1267 bp
		QEX11-14AMR	Intron 14	33,176.. 33,156	CACCTAGCACAGGACATGCAA	antisense			
6	Exon 15-16	QEX15-16AMF	Intron 14	33,650..33,669	TTTCTGTCTCCATGCCCTC	sense	Denaturation: 95°C for 30 sec Annealing: 60°C for 30 sec Extension: 72°C for 3 min Number of cycles: 35	MgCl <sub>2</sub> : 1.5 mM Forward and reverse primers: 0.1 µM each	825 bp
		QEX15-16AMR1	Intron 16	34,455.. 34,436	GACTCAATCACGCCAACTCC	antisense			
7	Exon 17-20	CL2-A6bF	Intron 16	34,961..34,980	GGGATTGATGGTACTTTGTT	sense	Denaturation: 95°C for 30 sec Annealing: 58°C for 30 sec Extension: 72°C for 3 min Number of cycles: 35	MgCl <sub>2</sub> : 2 mM Forward and reverse primers: 0.15 µM each	1012 bp
		CL2-A6bR	Intron 20	35,972..35,955	CCACCTCTGACCTCTATC	antisense			
8	Exon 21-22	CL2-A7bF	Intron 20	39,803..39,820	TAATCCAAAGAGGGTTTC	sense	Denaturation: 95°C for 30 sec Annealing: 54°C for 30 sec Extension: 72°C for 3 min Number of cycles: 35	MgCl <sub>2</sub> : 2 mM Forward and reverse primers: 0.15 µM each	2401 bp
		CL2-A7bR	3'UTR	42,203.. 42,186	CTAGTTCAGAGACTCCTA	antisense			

\* 5'UTR: 5' untranslated region; 3'UTR: 3' untranslated region

† relative to NCBI Reference Sequence NC\_000019.9

‡ Initial denaturation was at 95°C for 2 min and final extension at 72 °C for 7 min. The final reaction volume was 25 ul containing 100 ng of genomic DNA; 0.2 mM of each dNTP and 0.125 µl Taq polymerase (FastStart High Fidelity PCR System; Roche Applied Science, Indianapolis, IN)

**Table S2.** *SLC44A2* sequencing primers

Name	Location *	Position †	Sequence	Orientation	Region sequenced
CL2-S1F1	5'UTR	-175..-154	ACCATGCACACCTCAAGAGGG	sense	5'UTR, Exon 1
CL2-S2F2	Intron 1	23,636..23,655	GGGGGCTTGGGTGTGGTCAC	sense	Exon 2
QS3F2	Intron 2	25,146..25166	TGGTACGGCATTCAAACGACT	sense	Exon 3
CL2-S2R1	Intron 4	25,712..25,692	TGCCTCGGCCTCCCAAAGTGT	antisense	Exon 4
CL2-S3R1	Intron 7	29,171..29,150	CAGAGGATGGCACCAGTCAGGC	antisense	Exon 5,6,7
CL2-S3F3	Intron 7	29,101..29,121	GGGATCCCAGGAGGCTCAGGT	sense	Exon 8, 9, 10
CL2-S4F1	Intron 10	32,222..32,242	GGGCGGTGGGAAGTCAAG	sense	Exon 11, 12
CL2-S4F2	Exon12/Intron 12	32,619..32,637	GAAGCCAGCAGGTGGGGGG	sense	Exon 13, 14
CL2-S4F3	Intron 14	33,781..33,804	CTACCTCATGGTTTTCTGGCCCC	sense	Exon 15, 16
CL2-S5F1	Intron 16	35,082..35,103	TGACAGCGTGGACTCAGGAGAC	sense	Exon 17, 18
CL2-S5F2	Exon 18	35,429..35,450	CAATTTCTGCACCTCGGCCAGG	sense	Exon 19, 20
CL2-S6F1	Intron 20	39,811..39,832	AGAGGGTTTCTCAGCAGGGGAAG	sense	Exon 21
CL2-S6F2	Intron 21	40,694..40,715	AGTCTGCCCCTCTCTGGTCCCA	sense	Exon 22
CL2-S6F3	3'UTR	41,051..41,070	TGCCCCTCCCCATGAGCCAG	sense	3'UTR
CL2-S6F4	3'UTR	41,390..41,410	GCTCAAGGGCCACCAGAAGCA	sense	3'UTR
CL2-S6F5	3'UTR	41,708..41,731	CCCGACTTCCTCTGCCAGTTATTG	sense	3'UTR

\* UTR- untranslated region

† relative to NCBI Reference Sequence NC\_000019.9 (range 10713121..10755235)

**Table S3.** Genetic variations identified in the *SLC44A2* exons

Exon	Nucleotide Change *	dbSNP reference no.	Protein residue change †	Donor observations									NHLBI ESP VAF		
				Caucasians (N=96)		VAF	95% CI	HWE <i>p</i> value‡	African Americans (N=200)		VAF	95% CI	HWE <i>p</i> value‡	Caucasian	African American
				Homozygous	Heterozygous				Homozygous	Heterozygous					
1 (5'UTR)	c.-77T>A ‡	Novel	NA	0	0	0%	0-1.7%	0	0	5	1%	0.5-2.8%	0.86	NA	NA
1	c.6G>A ‡	rs115559926	Glu2Glu ‡	0	0	0%	0-1.7%	0	0	15	4%	2.0-5.9%	0.58	NA	3.4% ¶
4	c.204T>C	rs3087969	Asp68Asp	6	26	20%	13.7-26.7%	0.15	2	48	13%	9.5-16.7%	0.39	21.4%	15.8%
7	c.457C>T	rs147820753	Leu153Phe	0	2	1%	0.2-2.9%	0.92	0	2	0.5%	0.09-1.4%	0.94	0.6%	0.1%
7	c.461G>A	rs2288904	Arg154Gln	6	25	19%	13.7-26.3%	0.11	0	15	4%	2.0-5.9%	0.58	21.3%	6.6%
10	c.732G>A	rs146127348	Ala244Ala	0	0	0%	0-1.7%	0	0	2	0.5%	0.09-1.4%	0.94	0%	0.5%
10	c.822C>T	rs141667578	Tyr274Tyr	0	0	0%	0-1.7%	0	0	1	0.3%	0.01-1.3%	0.97	0.01%	0.02%
12	c.1035A>G	rs35362308	Ala345Ala	0	0	0%	0-1.7%	0	1	26	7%	4.8-9.9%	0.98	0.02%	4%
13	c.1140C>T	rs7255431	Ser380Ser	0	0	0%	0-1.7%	0	1	28	8%	5.1-10.4%	0.89	0.02%	4%
22	c.2043G>A	rs34625417	Ser681Ser	0	1	0.5%	0.03-2.8%	0.96	2	30	8%	5.9-11.8%	0.61	0.06%	7.5%
22	c.2046C>G	rs151043533	Ala682Ala	0	0	0%	0-1.7%	0	0	1	0.3%	0.01-1.3%	0.97	0%	0.5%
22 (3'UTR)	c.2363T>C	rs116639546	NA	0	0	0%	0-1.7%	0	0	8	2%	0.8-3.7%	0.77	NA	3.4% ¶
22 (3'UTR)	2460G>A	rs1053007	NA	14	46	38%	30.6-47.8%	0.91	14	93	30%	24.8-36%	0.15	38.9% ¶	30% ¶
22 (3'UTR)	c.2874G>A	rs74795234	NA	1	5	4%	1.7-7.2%	0.01	0	11	3%	1.3-4.8%	0.69	2.2% €	
22 (3'UTR)	c.2965T>G	rs10948	NA	11	41	33%	24.8-41.6%	0.76	3	56	16%	11.9-19.7%	0.33	36.7% ¶	11.5% ¶

\* relative to NCBI Reference Sequence NM\_020428.3

† relative to NCBI Reference Sequence NP\_065161.3

‡ relative to NCBI Reference Sequence NM\_001145056.1 and NP\_001138528.1

¶ dbSNP VAF

€ global VAF from 1000Genome phase 1 genotype data from 1094 worldwide individuals

¥ HWE *p*-value with 1 degree of freedomNHLBI ESP – NHLBI Exome Sequencing Project (<http://evs.gs.washington.edu/EVS>). Total number of samples in Caucasians = 4300; and total number of samples in African Americans = 2203

MAF-minor allele frequency

NA- Not available

**Table S4.** Genetic variations detected in the *SLC44A2* introns

Location	Nucleotide Change *	dbSNP reference no.	Donors observed (n)			VAF observed						NHLBI ESP VAF	
			Zygoty	Caucasian (N=96)	African American (N=200)	Caucasian			African American			Caucasian	African American
						VAF	95% CI	HWE <i>p</i> value	VAF	95% CI	HWE <i>p</i> value		
Intron 1	c.31+12C>T †	rs189105123	Homozygous	0	0	1.5%	0.4-4.2%	0.87	0.2%	0.01-1.3%	0.97	NA	NA
			Heterozygous	3	1								
	c.31+85G>A †	rs74436647	Homozygous	1	0	6%	3.5-10.6%	0.28	2.5%	1.3-4.4%	0.72	NA	NA
			Heterozygous	10	10								
	c.31+185C>T †	rs113379022	Homozygous	6	0	30%	22.6-37.8%	0.23	4%	2.4-6.4%	0.56	NA	NA
			Heterozygous	45	16								
	c.32-62C>T †	rs114095820	Homozygous	0	0	0.5%	0.03-2.8%	0.96	2%	0.8-3.7%	0.77	0.03%	2.5%
			Heterozygous	1	8								
Intron 5	c.331-44G>A	rs12972963	Homozygous	7	2	20%	14.4-27.2%	0.06	12%	8.7-15.5%	0.60	21.4%	15.9%
			Heterozygous	25	43								
Intron 7	c.503-15C>T	rs1560711	Homozygous	6	2	20%	13.7-26.7%	0.15	13%	9.9-17.0%	0.35	21.4%	15.3%
			Heterozygous	26	49								
Intron 8	c.626+18T>C	rs1560710	Homozygous	6	1	20%	13.7-26.7%	0.15	13%	9.5-16.7%	0.14	21.4%	15.3%
			Heterozygous	26	50								
	c.626+38T>G	rs1560709	Homozygous	6	1	20%	13.7-26.7%	0.15	13%	9.5-16.7%	0.14	21.4%	15.3%
			Heterozygous	26	50								
Intron 9	c.711-41C>T	rs114989609	Homozygous	0	0	0%	0-1.7%	0	1%	0.3-2.4%	0.89	0%	1.8%
			Heterozygous	0	4								
Intron 10	c.823+164C>T	rs9797854	Homozygous	6	1	19%	13.7-26.3%	0.11	10%	6.6-12.8%	0.51	NA	NA
			Heterozygous	25	36								
	c.823+294C>T	rs9797861	Homozygous	6	1	19%	13.7-26.3%	0.11	13%	9.9-17.0%	0.79	NA	NA
			Heterozygous	25	51								
	c.823+304T>C	rs138838423	Homozygous	0	1	0%	0-1.7%	0	6%	4.2-9.0%	0.79	NA	NA
			Heterozygous	0	23								
Intron 12	c.1055+15C>T	rs2163832	Homozygous	12	0	33%	25.9-41.8%	0.54	6%	4.2-9.0%	0.35	32.5%	10%
			Heterozygous	40	25								
Intron 13	c.1148+11C>A	rs7255439	Homozygous	0	1	0%	0-1.7%	0	8%	5.1-10.4%	0.89	0.02%	4%
			Heterozygous	0	28								
	c.1148+41C>A	rs115004332	Homozygous	0	0	0.5%	0.03-2.8%	0.96	8%	5.7-11.3%	0.20	0.03%	9%
			Heterozygous	1	33								

Intron 14	c.1233+14G>A	rs115585815	Homozygous	0	0	0.5%	0.03-2.8%	0.96	8%	5.7-11.3%	0.20	0.03%	8.6%
			Heterozygous	1	33								
Intron 16	c.1592-25C>T	rs79521494	Homozygous	0	0	3%	2.8-9.9%	0.55	2%	1.1-4.2%	0.74	3.4%	2.6%
			Heterozygous	11	9								
Intron 18	c.1769+34C>T	rs139577429	Homozygous	0	0	0%	0-1.7%	0	1%	0.5-2.8%	0.86	0%	0.3%
			Heterozygous	0	5								
Intron 19	c.1840+51C>T	rs3859514	Homozygous	15	7	38%	29.7-47.0%	0.83	22%	17.8-27.4%	0.21	35.4%	28%
			Heterozygous	43	76								
	c.1841-53C>G	rs76374006	Homozygous	0	0	5%	2.8-9.2%	0.59	2%	0.8-3.4%	0.80	6.8%	1.2%
			Heterozygous	10	7								
Intron 21	c.2014+144C>T	rs149845008	Homozygous	0	0	0%	0-1.7%	0	2%	0.6-3.2%	0.83	NA	NA
			Heterozygous	0	6								
	c.2014+322T>C	rs79965874	Homozygous	0	0	0%	0-1.7%	0	2%	0.6-3.2%	0.83	NA	NA
			Heterozygous	0	6								
	c.2014+369G>A	rs114916976	Homozygous	0	0	0.5%	0.03-2.8%	0.96	2%	0.8-3.4%	0.80	NA	NA
			Heterozygous	1	7								

\* relative to NCBI Reference Sequence NM\_020428.3

† relative to NCBI Reference Sequence NM\_001145056.1

¶ dbSNP MAF

€ global MAF from 1000Genome phase 1 genotype data from 1094 worldwide individuals

NHLBI ESP – NHLBI Exome Sequencing Project (<http://evs.gs.washington.edu/EVS>). Total number of samples in Caucasians = 4300; and total number of samples in African Americans = 2203

MAF minor allele frequency

NA- Not available

**Table S5. SLC44A2 potential alleles and their frequencies in African Americans and Caucasians**

Allele number	Haplotype*	Haplotype frequency					
		African American			Caucasian		
		Observed (n)	Mean†	95% CI‡	Observed (n)	Mean†	95% CI‡
KM024996	TGCGCCTGCG <b>T</b> TTTCGCTTTACCCCGCCCCCTGGCTGGT	219	54.8%	47.4-62.3	76	39.6%	30.6-49.1
1	-----CA- <b>A</b> CCG-A-CC--T-----T-----A-G	2	0.5%	0.09-1.7	0	0%	0-1.7
2	-----CA- <b>A</b> CCG-----CC-----	0	0%	0-0.8	1	0.5%	0.03-2.8
3	-----CA--CCG---CC-----AA-----	1	0.25%	0.01-1.3	0	0%	0-1.7
4	-----CA--CCG---C-----AA--T-----A--	1	0.25%	0.01-1.3	0	0%	0-1.7
5	-----CA--CCG---C-----AA--T-T-----A--	1	0.25%	0.01-1.3	0	0%	0-1.7
6	-----CA--CCG-----AA--T-----A-G	1	0.25%	0.01-1.3	0	0%	0-1.7
7	-----CA--CCG---C-----AA--T-----CA-G	2	0.5%	0.09-1.7	0	0%	0-1.7
8	-----CA--CCG---CC-----AA--T-----CA-G	2	0.5%	0.09-1.7	0	0%	0-1.7
9	-----CA--CCG---C-----AA--T-----G-A-G	1	0.25%	0.01-1.3	0	0%	0-1.7
10	---A---A--CCG---CC-----AA--T-----A-G	1	0.25%	0.01-1.3	0	0%	0-1.7
11	-----A--CCG---C-----AA--T-----A-G	1	0.25%	0.01-1.3	0	0%	0-1.7
12	-----C---CCG---CC-----T-----A-G	2	0.5%	0.09-1.7	0	0%	0-1.7
13	A-----CC--T-----T-----A-G	1	0.25%	0.01-1.3	0	0%	0-1.7
14	-----AA--T-----A-G	1	0.25%	0.01-1.3	0	0%	0-1.7
15	-----AA--T-----A--	1	0.25%	0.01-1.3	0	0%	0-1.7
16	----T-----T-----A-G	0	0%	0-0.8	2	1.0%	0.18-2.9
17	-----A-G	1	0.25%	0.01-1.3	0	0%	0-1.7
18	-----T---C--TA-----A-A--	1	0.25%	0.01-1.3	0	0%	0-1.7
19	-----T---G-TA-----A--A--	2	0.5%	0.09-1.7	0	0%	0-1.7
20	-----G-TA-----	1	0.25%	0.01-1.3	0	0%	0-1.7
21	-----TA-----A--	1	0.25%	0.01-1.3	0	0%	0-1.7
22	---A-----T-T-----A--	1	0.25%	0.01-1.3	0	0%	0-1.7
23	----T-----T-T-----	0	0%	0-0.8	1	0.5%	0.03-2.8
24	-----CA-----T-T-----	0	0%	0-0.8	1	0.5%	0.03-2.8
25	-----T-T-----	1	0.25%	0.01-1.3	0	0%	0-1.7
26	---A--CA-----T-T-----A--	1	0.25%	0.01-1.3	0	0%	0-1.7
27	A-----C-----G-----	1	0.25%	0.01-1.3	0	0%	0-1.7
28	-A-----T-----	1	0.25%	0.01-1.3	0	0%	0-1.7
29	-A-----A-	1	0.25%	0.01-1.3	0	0%	0-1.7
30	---T-----C-----	1	0.25%	0.01-1.3	0	0%	0-1.7
31	---T-----T-----	1	0.25%	0.01-1.3	0	0%	0-1.7
32	-----A--A--	0	0%	0-0.8	1	0.5%	0.03-2.8
33	-----C-----	1	0.25%	0.01-1.3	0	0%	0-1.7
34	-----C-----	1	0.25%	0.01-1.3	0	0%	0-1.7
35	-----T-----	1	0.25%	0.01-1.3	0	0%	0-1.7
36	-----T-----	1	0.25%	0.01-1.3	0	0%	0-1.7
Confirmed haplotypes	See Table 1	145			110		
Total		400			192		

\* The nucleotides at the 61 SNP positions with variations are shown in 5' to 3' orientation (Table S3 and S4). The remaining 8131 nucleotide positions that we determined had no variation relative to the reference sequence KM024996. The SNP G/A (bold) determines the HNA-3a/b antigen polymorphism.

† Number of observed alleles (n)/Total number of alleles (African American=400 and Caucasian=192).

‡ 95% confidence interval (CI), Poisson distribution, two sided.<sup>32</sup>



