

AH1 Variant Allele Frequencies
8/28/2008

Notes: These summary statistics were compiled from clinical tests.
Only variants that are within the coding regions and about 50 base pairs of flanking sequence on each side are reported.
At this time, these summary statistics do NOT include insertion/deletion variants.

Exon	Left Flanking	Sequence Variation	Right Flanking	Coding Change	SNP Reference	Allele Frequencies
4	-----TTGTGTGT	c.11-45 T>C	ATGATAGAGTCATTT	Intronic		T-99.5% C-0.5%
4	GCTTAAGACCCACAG	c.72 T>C	GATCTAATGCGTGAA	p.Ser24Ser		T-99.5% C-0.5%
6	TGACTTACTTTCCCT	c.662 C>G	AGATACTTTATTCCA	p.Ser221Stop		C-99.1% G-0.9%
6	AAGAAAAAGGAAGTT	c.724 C>T	CAGTCTTCTCTAAAG	p.Pro242Ser		C-99.1% T-0.9%
7	AAAGAAAGAATCTTC	c.804 A>C	GTTAGATCAGTTTCT	p.Ser268Ser	rs35528530	A-99.6% C-0.4%
8	TGGGAGTTTACATTC	c.1049 A>G	CCGAACTGATAGACT	p.His350Arg		A-99% G-1%
9	CCAGAGTGGGAAGAA	c.1267 C>T	AAATTGTATTTAATG	p.Gln423Stop		C-99.6% T-0.4%
10	AATTTTAGGTAGCTT	c.1345-24 A>T	GTCCTTCTGTGAATTT	Intronic		A-99.5% T-0.5%
10	GGGCATTTCTTAAGG	c.1440+2 T>C	ATGTTTTTCATCCAT	Intronic		T-99.5% C-0.5%
11	GAAATCATTACCCAT	c.1583 C>A	AACACTGTACGTAAC	p.Ser528Stop		C-99.6% A-0.4%
12	TAAAGCCATCTTACC	c.1643 G>A	CTCTATGATGGCTCT	p.Arg548His	rs35433555	G-99% A-1%
12	GTAATAAAGTGGAAA	c.1765 C>T	GACTCCCTGGGCAGG	p.Arg589Stop		C-99% T-1%
13	TTTATATTTTTTAAA	c.1780-14 C>T	GTATGTTTTTTTAGGC	Intronic	rs2757645	T-78.6% C-21.4%
13	AAAAACTAGCCTTCG	c.1780-47 C>T	GTTTTAATGTTTGCT	Intronic	rs17053651	C-96.4% T-3.6%
13	TTAGGCTTGCCGTAT	c.1791 C>T	CCAAACAAACACCTC	p.Ile597Ile		C-99.1% T-0.9%
14	CCTGGTCAAAAGATG	c.1997 A>T	TCACTACATCCTTAC	p.Asp666Val		A-99.6% T-0.4%
15	GGTCCGACAGTTTGA	c.2223 T>C	GTTCACAAAAGTTTT	p.Asp741Asp	rs2273761	T-98.2% C-1.8%
15	TGTTTTGATACTGAA	c.2266 G>T	GTATGTCAGAGACTA	p.Gly756Cys		G-99.1% T-0.9%
17	TCAGTAGTAGTATTT	c.2374-16 T>C	TTTTAATCATTTTAG	Intronic		T-99.5% C-0.5%
17	ATTTTAGGAAATTA	c.2382 A>G	GAAACTGAGTTTAAG	p.Lys794Lys		A-99% G-1%
17	AGAATTATGGATCTC	c.2488 C>T	GGATGTAAGTATATT	p.Arg830Trp		C-97.6% T-2.4%
19	TGAATCTCTCTTTTT	c.2624-6 A>G	TTTAGGAGAACAAGT	Intronic	rs41288015	A-97.1% G-2.9%
21	ATACTTCCTTATTTT	c.2962-16 A>T	TTGTATTTCTTTTAG	Intronic		A-99.5% T-0.5%
21	TTTAGGAGGTGATAC	c.2972 G>A	TTCCTGTGCTGCAA	p.Arg991His		G-99.5% A-0.5%

22	TCTCTCATTACTTC	c.3015 A>G	CCACCAGCAGTTTCC	p.Ser1005Ser	rs41287054	A-97.1% G-2.9%
23	AAAAGTGTTAAGAAA	c.3165+42 G>C	TACAAAATTATACTG	Intronic	rs7772864	G-97.6% C-2.4%
25	AAGGTAAATGTTTT	c.3426+13 G>A	AGCATACGTATTACA	Intronic	rs6914831	A-57.2% G-42.8%
25	AGATAAAGGAGCGAT	c.3368 C>T	CCCTCCTTTAAGCCC	p.Ser1123Phe		C-99.5% T-0.5%
26	TTTGCAGTGATAAAT	c.3485+38 G>C	GTTGGGGTTTTCTT-	Intronic	rs4896141	G-86.1% C-13.9%
26	AGTGATAAATCGTTG	c.3485+43 G>C	GGTTTTCTTTT----	Intronic		G-99.5% C-0.5%
27	AGGCAGAAAAGTCAC	c.3579 T>C	CTAATAGAGGTAAGG	p.Thr1193Thr		T-99.5% C-0.5%
28	AAGTTAAGAGCTGCC	c.*28 G>C	AAATGCACAGAGGTG	Post coding	rs9494209	G-88.9% C-11.1%