

Table S8. Nucleotide Substitutions in *AMY1A* gene in introns and regulatory regions

Genomic Coordinates (hg19)	Gene Region	Modern human	Altai Neanderthal (Variant Frequency)	Chagyrskaya Neanderthal (Variant Frequency)	Vindija Neanderthal (Variant Frequency)	Denisovan (Variant Frequency)	Codon → Amino acid
104,239,478	Upstream region	G	G (100%)	G (100%)	A (17%)	G (100%)	
104,239,460	Upstream region	C	C (100%)	C (100%)	T (14%)	C (100%)	
104,239,450	Upstream region	G	G (100%)	G (100%)	A (13%)	G (100%)	
104,239,175	Upstream region	C	C (100%)	C (100%)	T (14%)	C (100%)	
104,239,005	5'UTR	G	G (100%)	G (100%)	A (27%)	G (100%)	
104,238,932	5'UTR	C	C (100%)	T (18%)	C (100%)	C (100%)	
104,238,918	5'UTR	G	G (100%)	G (100%)	A (11%)	G (100%)	
104,238,905	5'UTR	C	C (100%)	C (100%)	T (13%)	C (100%)	
104,238,850	5'UTR	G	G (100%)	G (100%)	A (15%)	G (100%)	
104,238,820	5'UTR	C	C (100%)	C (100%)	T (18%)	C (100%)	
104,238,815	5'UTR	C	C (100%)	C (100%)	T (17%)	C (100%)	
104,238,794	5'UTR	G	A (100%)	A (100%)	A (90%)	A (100%)**	
104,238,676	5'UTR	G	G (100%)	G (100%)	A (20%)	G (100%)	
104,238,649	Intron 1	G	G (98%)	A (8%)*	A (18%)	G (100%)	
104,238,648	Intron 1	G	A (3%)*	G (100%)	A (20%)	G (100%)	
104,238,610	Intron 1	C	T (3%)*	C (100%)	C (100%)	T (27%)	
104,238,487	Intron 1	T	delT (23%)	T (100%)	delT (30%)	delT (29%)	
104,238,486	Intron 1	C	T (14%)	C (100%)	T (17%)	T (21%)	
104,238,481	Intron 1	C	T (7%)*	C (100%)	C (100%)	T (14%)	
104,238,366	Intron 1	C	C (100%)	T (14%)	C (100%)	C (100%)	
104,238,362	Intron 1	C	C (100%)	C (100%)	T (11%)	C (100%)	
104,238,354	Intron 1	C	C (100%)	C (100%)	T (11%)	C (100%)	
104,238,317	Intron 1	G	G (100%)	G (91%)	A (11%)	G (100%)	
104,238,094	Exon 2	GTC	GTC (100%)	GTC (100%)	GTT (17%)	GTC (100%)	CAG→Q <sub>41</sub> CAA→Q <sub>41</sub>
104,238,054	Intron 2	C	G (100%)	G (100%)	G (100%)	G (100%)**	
104,238,050	Intron 2	C	C (100%)	T (6%)*	T (13%)	C (100%)	
104,238,047	Intron 2	C	C (100%)	T (12%)	T (15%)	C (100%)	
104,238,025	Intron 2	T	T (100%)	T (100%)	C (94%)	T (100%)	
104,238,017	Intron 2	G	G (100%)	A (8%)*	A (12%)	G (100%)	
104,237,948	Intron 2	C	C (100%)	C (100%)	T (18%)	C (100%)	

104,237,938	Intron 2	G	G (100%)	G (100%)	A (13%)	G (100%)	
104,237,897	Intron 2	C	C (100%)	T (8%)*	T (13%)	C (100%)	
104,237,850	Intron 2	G	G (100%)	G (100%)	A (15%)	G (100%)	
104,237,816	Intron 2	T	C (87%)	C (89%)	C (100%)	C (100%)	
104,237,595	Intron 3	C	C (100%)	C (100%)	T (14%)	C (100%)	
104,237,546	Intron 3	G	G (100%)	A (11%)	A (8%)*	G (100%)	
104,237,512	Intron 3	G	G (100%)	A (18%)	G (100%)	G (100%)	
104,237,472	Intron 3	T	C (100%)	C (100%)	T (100%)	T (100%)	
104,237,461	Intron 3	G	G (100%)	A (25%)	G (100%)	G (100%)	
104,237,384	Intron 3	T	G (100%)	G (100%)	G (100%)	G (100%)	
104,237,332	Intron 3	C	C (100%)	C (100%)	T (11%)	C (100%)	
104,237,331	Intron 3	C	C (100%)	C (100%)	T (11%)	C (100%)	
104,237,328	Intron 3	C	C (100%)	C (100%)	T (11%)	C (100%)	
104,237,319	Intron 3	C	C (100%)	T (12%)	C (100%)	C (100%)	
104,237,314	Intron 3	C	C (100%)	C (100%)	T (12%)	C (100%)	
104,237,254	Intron 3	G	G (100%)	G (100%)	G (100%)	A (93%)	
104,237,248	Intron 3	G	G (100%)	A (5%)*	G (100%)	A (13%)	
104,237,245	Intron 3	C	C (100%)	C (100%)	C (100%)	A (13%)	
104,237,229	Intron 3	C	C (100%)	T (12%)	C (100%)	C (100%)	
104,237,220	Intron 3	C	C (100%)	T (11%)	C (100%)	C (100%)	
104,237,214	Intron 3	G	G (100%)	A (14%)	A (7%)*	G (100%)	
104,237,205	Intron 3	C	C (100%)	T (8%)*	T (13%)	C (100%)	
104,237,078	Intron 3	C	A (100%)	A (100%)	A (100%)	A (100%)**	
104,236,994	Intron 3	C	T (100%)	T (100%)	T (100%)	T (100%)**	
104,236,974	Intron 3	C	C (100%)	T (7%)*	T (20%)	C (100%)	
104,236,963	Intron 3	C	C (100%)	C (100%)	T (20%)	C (100%)	
104,236,949	Intron 3	G	G (100%)	A (14%)	G (100%)	G (100%)	
104,236,905	Intron 3	C	C (100%)	C (100%)	C (100%)	T (79%)	
104,236,901	Intron 3	C	C (100%)	T (8%)*	T (11%)	C (100%)	
104,236,715	Exon 4	AT <u>G</u>	AT <u>G</u> (100%)	AT <u>G</u> (100%)	AT <u>A</u> (14%)	AT <u>G</u> (100%)	UAC→Y <sub>118</sub> UAU→Y <sub>118</sub>
104,236,425	Intron 4	A	A (100%)	A (100%)	A (100%)	C (100%)	
104,236,399	Intron 4	C	C (100%)	C (100%)	T (13%)	C (100%)	
104,236,396	Intron 4	G	G (100%)	A (13%)	A (6%)*	G (100%)	
104,236,381	Intron 4	G	G (100%)	G (100%)	A (11%)	G (100%)	

104,236,379	Intron 4	G	G (100%)	G (100%)	A (11%)	G (100%)	
104,236,340	Intron 4	G	G (100%)	A (14%)	A (10%)*	G (100%)	
104,236,262	Intron 4	C	C (100%)	C (100%)	T (13%)	C (100%)	
104,236,261	Intron 4	A	A (100%)	T (100%)	A (100%)	A (100%)	
104,236,192	Intron 4	C	T (3%)*	C (100%)	T (14%)	C (100%)	
104,236,153	Intron 4	C	C (100%)	C (100%)	T (13%)	C (100%)	
104,236,135	Exon 5	GAC	GAC (100%)	GAC (100%)	GAT (11%)	GAC (100%)	CUG→L <sub>162</sub> CUA→L <sub>162</sub>
104,236,108	Exon 5	CCC	CCC (100%)	CCT (11%)**	CCT (13%)	CCC (100%)	GGG→G <sub>171</sub> GGA→G <sub>171</sub>
104,235,967	Exon 5	TTG	TTG (100%)	TTA (14%)	TTG (100%)	TTG (100%)	AAC→N <sub>218</sub> AAU→N <sub>218</sub>
104,235,656	Intron 5	T	delT (97%)	delT (78%)	delT (73%)	delT (91%)	
104,235,449	Intron 5	C	C (100%)	C (100%)	T (19%)	T (11%)**	
104,235,370	Intron 5	G	G (100%)	G (100%)	A (13%)	G (100%)	
104,235,337	Intron 5	C	C (100%)	C (100%)	T (15%)	C (100%)	
104,235,186	Intron 5	G	G (100%)	G (100%)	A (14%)	G (100%)	
104,235,171	Exon 6	GAC	GAC (100%)	GAT (8%)*	GAT (11%)	GAC (100%)	CUG→L <sub>237</sub> CUA→L <sub>237</sub>
104,235,144	Exon 6	CTG	CTG (100%)	CTG (100%)	CTA (15%)	CTG (100%)	GAC→D <sub>246</sub> GAU→D <sub>246</sub>
104,235,096	Exon 6	GAG	GAA (3%)*	GAA (19%)	GAA (52%)	GAG (100%)	CUC→L <sub>262</sub> CUU→L <sub>262</sub>
104,234,993	Intron 6	G	G (100%)	A (20%)	A (10%)*	G (100%)	
104,234,937	Intron 6	C	C (100%)	C (100%)	T (27%)	C (100%)	
104,234,877	Intron 6	C	C (100%)	C (100%)	T (20%)	C (100%)	
104,234,553	Intron 6	G	G (100%)	G (100%)	A (11%)	A (11%)**	
104,234,436	Intron 6	C	C (100%)	T (20%)	C (100%)	C (100%)	
104,234,328	Intron 6	G	G (100%)	G (100%)	A (14%)	G (100%)	
104,234,256	Intron 6	C	T (28%)	T (33%)	T (10%)*	C (100%)**	
104,234,233	Intron 6	C	T (3%)*	C (100%)	T (15%)	C (100%)	
104,234,009	Exon 8	ATG	ATA (7%)*	ATA (14%)	ATA (14%)	ATA (13%)	UAC→Y <sub>321</sub> UAU→Y <sub>321</sub>
104,233,908	Intron 8	A	A (100%)	A (100%)	A (100%)	C (17%)	
104,233,903	Intron 8	C	C (100%)	C (100%)	T (18%)	C (100%)	
104,233,860	Intron 8	G	G (100%)	A (14%)	A (8%)*	G (100%)	
104,233,857	Intron 8	G	G (100%)	G (100%)	A (15%)	G (100%)	
104,233,840	Intron 8	G	G (100%)	G (100%)	A (13%)	G (100%)	
104,233,833	Intron 8	C	C (100%)	T (11%)	T (6%)*	C (100%)	
104,233,831	Intron 8	C	C (100%)	C (100%)	T (17%)	C (100%)	
104,233,787	Intron 8	C	C (100%)	T (9%)*	T (11%)	C (100%)	

104,233,775	Intron 8	T	T (100%)	T (100%)	T (100%)	C (10%)*
104,233,766	Intron 8	C	C (100%)	T (13%)	T (20%)	C (100%)
104,233,765	Intron 8	C	C (100%)	T (13%)	C (92%)	C (100%)
104,233,718	Intron 8	G	G (100%)	A (15%)	G (100%)	G (100%)
104,233,648	Intron 8	G	G (100%)	A (18%)	G (100%)	G (100%)
104,233,645	Intron 8	G	G (100%)	A (18%)	G (100%)	G (100%)
104,233,598	Intron 8	G	G (100%)	G (100%)	A (20%)	G (100%)
104,233,330	Intron 8	G	G (100%)	G (100%)	A (18%)	G (100%)
104,233,260	Intron 8	C	C (100%)	T (17%)	C (100%)	C (100%)
104,233,193	Intron 8	G	G (100%)	G (100%)	A (12%)	G (100%)
104,233,189	Intron 8	G	G (100%)	G (100%)	A (13%)	G (100%)
104,233,186	Intron 8	G	G (100%)	G (100%)	A (12%)	G (100%)
104,233,127	Intron 8	G	T (97%)	G (100%)	G (95%)	G (100%)**
104,233,126	Intron 8	G	G (100%)	G (100%)	A (11%)	G (100%)
104,232,949	Intron 8	G	G (100%)	A (11%)	G (100%)	G (100%)
104,232,911	Intron 8	G	G (100%)	A (11%)	G (100%)	G (100%)
104,232,902	Intron 8	C	C (100%)	C (100%)	T (11%)	C (100%)
104,232,892	Intron 8	C	C (100%)	T (20%)	T (12%)	C (100%)
104,232,891	Intron 8	C	C (100%)	T (20%)	T (6%)*	C (100%)
104,232,886	Intron 8	G	G (100%)	A (9%)*	A (11%)	G (100%)
104,232,884	Intron 8	G	G (100%)	G (100%)	A (12%)	G (100%)
104,232,882	Intron 8	C	C (100%)	C (100%)	T (11%)	C (100%)
104,232,646	Intron 8	A	G (97%)	G (86%)	G (100%)	G (100%)
104,232,616	Intron 8	C	C (100%)	C (100%)	C (100%)	T (10%)*
104,232,532	Intron 8	G	G (100%)	G (100%)	A (11%)	G (100%)
104,232,489	Intron 8	C	C (100%)	C (100%)	T (13%)	C (100%)
104,232,186	Intron 8	G	G (100%)	A (13%)	A (10%)*	G (100%)
104,232,184	Intron 8	C	C (100%)	T (13%)	T (20%)	C (100%)
104,232,161	Intron 8	C	C (100%)	C (100%)	T (17%)	C (100%)
104,232,130	Intron 8	C	C (100%)	T (25%)	C (100%)	C (100%)
104,232,117	Intron 8	G	G (100%)	G (100%)	G (100%)	T (10%)*
104,232,025	Intron 8	G	G (100%)	A (18%)	G (100%)	G (100%)
104,231,993	Intron 8	C	C (100%)	C (100%)	T (14%)	C (100%)
104,231,798	Intron 9	C	C (100%)	T (11%)	C (100%)	C (100%)
104,231,749	Intron 9	C	C (100%)	C (100%)	T (13%)	C (100%)

104,231,731	Intron 9	G	G (100%)	A (15%)	G (100%)	G (100%)	
104,231,642	Exon 10	CCC	CCT(4%)*	CCC(100%)	CCT(17%)	CCT(5%)*	GGG→G <sub>413</sub> GGA→G <sub>413</sub>
104,231,621	Exon 10	CCC	CCC(100%)	CCC(100%)	CCT(13%)	CCC(100%)	GGG→G <sub>420</sub> GGA→G <sub>420</sub>
104,231,542	Intron 10	C	C (100%)	C (100%)	T (13%)	C (100%)	
104,231,514	Intron 10	A	A (100%)	A (100%)	InsA (14%)	DelA (14%)**	
104,231,438	Intron 10	C	C (100%)	T (7%)*	T (14%)	C (100%)	
104,231,411	Intron 10	G	G (100%)	A (19%)	G (100%)	G (100%)	
104,231,404	Intron 10	C	T (4%)*	C (100%)	T (12%)	T (13%)**	
104,231,298	Intron 10	G	G (100%)	G (100%)	A (15%)	G (100%)	
104,231,278	Intron 10	C	C (97%)	C (100%)	T (12%)	C (100%)	
104,231,242	Intron 10	G	G (100%)	A (27%)	G (100%)	G (100%)	
104,231,201	Intron 10	C	C (100%)	C (94%)	T (14%)	C (100%)	
104,231,200	Intron 10	G	G (100%)	A (6%)*	A (14%)	G (100%)	
104,231,175	Intron 10	G	G (100%)	A (18%)	G (100%)	G (100%)	
104,231,134	Intron 10	G	G (100%)	A (17%)	A (17%)	G (100%)	
104,231,089	Intron 10	C	C (100%)	C (100%)	T (13%)	C (100%)	
104,231,088	Intron 10	G	G (100%)	G (100%)	A (14%)	G (100%)	
104,231,043	Intron 10	C	C (100%)	T (16%)	C (100%)	C (100%)	
104,231,040	Intron 10	C	C (100%)	C (100%)	T (15%)	C (100%)	
104,231,033	Intron 10	C	C (100%)	T (6%)*	T (15%)	C (100%)	
104,231,005	Intron 10	C	C (100%)	T (14%)	C (100%)	C (100%)	
104,230,805	Intron 10	G	G (100%)	G (100%)	A (13%)	G (100%)	
104,230,786	Intron 10	G	G (100%)	G (100%)	A (15%)	G (100%)	
104,230,664	Intron 10	G	G (100%)	A (20%)	G (100%)	G (100%)	
104,230,649	Intron 10	C	C (100%)	T (17%)	C (100%)	C (100%)	
104,230,635	Intron 10	A	T (90%)	T (100%)	T (100%)	T (100%)**	
104,230,594	Intron 10	C	C (100%)	T (18%)	C (100%)	C (100%)	
104,230,577	Intron 10	G	A (3%)*	G (100%)	A (14%)	G (100%)	
104,230,548	Intron 10	C	C (95%)	C (100%)	T (17%)	C (100%)	
104,230,496	Intron 10	G	G (100%)	A (14%)	G (100%)	G (100%)	
104,230,422	Intron 10	G	G (100%)	A (18%)	G (100%)	A (8%)*	
104,230,397	Intron 10	C	C (100%)	T (20%)	C (100%)	C (100%)	
104,230,384	Intron 10	C	C (100%)	C (100%)	T (17%)	C (100%)	
104,230,352	Intron 10	C	C (100%)	C (100%)	T (12%)	C (100%)	

104,230,319	Intron 10	C	C (100%)	T (12%)	C (100%)	C (100%)
104,230,014	Downstream region	G	G (100%)	G (100%)	A (30%)	G (100%)
104,229,887	Downstream region	C	C (100%)	T (14%)	C (100%)	C (100%)
104,229,778	Downstream region	G	G (100%)	A (8%)*	A (15%)	G (100%)
104,229,768	Downstream region	C	C (100%)	T (12%)	T (7%)*	C (100%)
104,229,767	Downstream region	C	C (100%)	C (100%)	T (13%)	C (100%)
104,229,761	Downstream region	G	G (100%)	G (100%)	A (18%)	G (100%)
104,229,742	Downstream region	G	G (100%)	A (13%)	G (100%)	G (100%)
104,229,617	Downstream region	G	G (100%)	A (17%)	G (100%)	G (100%)
104,229,602	Downstream region	C	C (100%)	T (25%)	C (100%)	C (100%)
104,229,591	Downstream region	G	G (100%)	A (13%)	G (100%)	G (100%)
104,229,568	Downstream region	C	C (100%)	T (5%)*	T (18%)	C (100%)
104,229,563	Downstream region	G	G (100%)	A (12%)	G (100%)	G (100%)
104,229,544	Downstream region	C	C (100%)	T (11%)	T (10%)*	C (100%)
104,229,543	Downstream region	C	C (100%)	T (11%)	C (100%)	C (100%)
104,229,502	Downstream region	C	C (100%)	T (17%)	T (8%)*	C (100%)

n.a.: not available; the variant frequency is referred to the percentage of the highlighted base in the sequenced ancient hominine genome, with\* frequency≤10% and \*\* counts<10. In light orange are underlined the variants fixed at 100% in modern human compared to ancient hominines.