

Table S15. Nucleotide Substitutions in *CST5* gene in introns and regulatory regions

Genomic Coordinates (hg19)	Gene Region	Modern human	Altai Neanderthal (Variant Frequency)	Chagyrskaya	Vindija	Denisovan (Variant Frequency)	Codon→ amino acid
				Neanderthal (Variant Frequency)	Neanderthal (Variant Frequency)		
23,860,839	Upstream regions	C	C (100%)	C (100%)	T (14%)	C (100%)	
23,860,785	Upstream regions	G	G (100%)	A (4%)*	A (11%)	G (100%)	
23,860,782	Upstream regions	G	G (100%)	A (4%)*	A (18%)	G (100%)	
23,860,761	Upstream regions	G	A (2%)*	A (4%)*	A (19%)	G (100%)	
23,860,736	Upstream regions	G	G (100%)	A (4%)*	A (11%)	G (100%)	
23,860,710	Upstream regions	G	G (100%)	A (12%)	A (7%)*	G (100%)	
23,860,681	Upstream regions	C	C (98%)	C (100%)	T (13%)	C (100%)	
23,860,680	Upstream regions	C	C (100%)	T (4%)*	T (13%)	C (100%)	
23,860,677	Upstream regions	C	C (96%)	T (4%)*	T (18%)	C (100%)	
23,860,626	Upstream regions	C	C (100%)	C (100%)	T (11%)	C (100%)	
23,860,613	Upstream regions	C	C (100%)	T (12%)	T (8%)*	C (100%)	
23,860,587	Upstream regions	C	T (3%)*	C (100%)	T (13%)	C (100%)	
23,860,567	Upstream regions	C	C (97%)	T (15%)	T (5%)*	C (100%)	
23,860,550	Upstream regions	C	C (97%)	T (9%)*	T (15%)	C (100%)	
23,860,548	Upstream regions	C	C (100%)	C (100%)	T (11%)	C (100%)	
23,860,523	Upstream regions	C	C (100%)	T (3%)*	T (19%)	C (100%)	
23,860,513	Upstream regions	C	C (100%)	T (9%)*	T (13%)	C (100%)	
23,860,498	Upstream regions	C	G (97%)	G (100%)	G (92%)	C (100%)	
23,860,482	Upstream regions	C	C (100%)	T (11%)**	T (13%)	C (100%)	
23,860,481	Upstream regions	C	C (100%)	C (100%)	T (14%)	C (100%)	
23,860,468	Upstream regions	C	C (100%)	C (100%)	T (14%)	C (100%)	
23,860,466	Upstream regions	C	C (100%)	C (100%)	T (14%)	C (100%)	
23,860,435	Upstream regions	C	C (100%)	T (7%)*	T (13%)	C (100%)	
23,860,434	Upstream regions	C	C (100%)	T (8%)*	T (13%)	C (100%)	
23,860,289	Exon 1 (Signal)	<u>G</u> AC	<u>G</u> AC (100%)	<u>A</u> AC (4%)*	<u>A</u> AC (17%)	<u>G</u> AC (100%)	CUG→L ^{9(sp)} UUG→L ^{9(sp)}
23,860,227	Exon 1	TG <u>G</u>	TG <u>G</u> (100%)	TG <u>A</u> (17%)	TG <u>A</u> (6%)*	TG <u>G</u> (100%)	ACC→T ₉ ACA→T ₉
23,860,212	Exon 1	TA <u>G</u>	TA <u>G</u> (100%)	TA <u>G</u> (92%)	TAA (12%)	TA <u>G</u> (100%)	AUC→I ₁₄ AUU→I ₁₄
23,860,158	Exon 1	TA <u>G</u>	TA <u>G</u> (100%)	TA <u>G</u> (100%)	TAA (13%)	TA <u>G</u> (100%)	AUC→I ₃₂ AUU→I ₃₂

23,860,152	Exon 1	<u>CTC</u>	<u>CTC</u> (100%)	<u>CTT</u> (7%)*	<u>CTT</u> (14%)	<u>CTC</u> (100%)	GAG→E ₃₄ GAA→E ₃₄
23,860,113	Exon 1	<u>GCG</u>	<u>GCG</u> (100%)	<u>GCA</u> (17%)	<u>GCA</u> (9%)*	<u>GCG</u> (100%)	CGC→R ₄₇ CGU→R ₄₇
23,860,083	Exon 1	<u>GTC</u>	<u>GTC</u> (100%)	<u>GTT</u> (7%)*	<u>GTT</u> (12%)	<u>GTC</u> (100%)	CAG→Q ₅₇ CAA→Q ₅₇
23,860,082	Intron 1	C	C (100%)	C (100%)	T (13%)	C (100%)	
23,860,079	Intron 1	G	G (100%)	A (6%)*	A (13%)	G (100%)	
23,860,046	Intron 1	T	T (100%)	T (100%)	T (100%)	G (100%)	
23,860,033	Intron 1	C	C (100%)	T (8%)*	T (18%)	C (100%)	
23,860,027	Intron 1	G	G (100%)	G (100%)	A (14%)	G (100%)	
23,859,994	Intron 1	G	G (100%)	A (8%)*	A (13%)	G (100%)	
23,859,956	Intron 1	G	G (100%)	A (13%)	A (3%)*	G (100%)	
23,859,950	Intron 1	C	C (100%)	T (14%)	T (6%)*	T (3%)*	
23,859,906	Intron 1	G	A (94%)	A (88%)	A (82%)	A (94%)	
23,859,861	Intron 1	G	G (100%)	G (100%)	A (14%)	G (100%)	
23,859,816	Intron 1	G	G (100%)	G (100%)	A (17%)	G (100%)	
23,859,798	Intron 1	C	C (100%)	C (100%)	T (12%)	C (100%)	
23,859,753	Intron 1	C	T (100%)	T (100%)	T (100%)	T (100%)	
23,859,750	Intron 1	C	C (100%)	T (3%)*	T (11%)	C (100%)	
23,859,700	Intron 1	G	G (100%)	A (4%)*	A (18%)	G (100%)	
23,859,653	Intron 1	C	C (100%)	T (11%)	C (100%)	C (100%)	
23,859,644	Intron 1	C	C (100%)	C (100%)	T (11%)	C (100%)	
23,859,604	Intron 1	G	G (100%)	A (23%)	G (100%)	G (100%)	
23,859,603	Intron 1	G	G (100%)	A (15%)	G (100%)	G (100%)	
23,859,586	Intron 1	C	C (100%)	C (83%)	T (14%)	C (100%)	
23,859,570	Intron 1	C	C (100%)	T (15%)	T (10%)*	C (100%)	
23,859,460	Intron 1	C	C (100%)	C (100%)	T (11%)	C (100%)	
23,859,440	Intron 1	G	G (100%)	G (100%)	A (11%)	G (100%)	
23,859,396	Intron 1	G	G (100%)	G (100%)	A (18%)	G (100%)	
23,859,394	Intron 1	C	C (100%)	C (100%)	T (12%)	C (100%)	
23,859,314	Intron 1	G	G (100%)	A (11%)	G (100%)	G (100%)	
23,859,282	Intron 1	G	G (100%)	G (100%)	A (11%)	G (100%)	
23,859,246	Intron 1	C	C (100%)	C (100%)	T (11%)	C (100%)	
23,859,213	Intron 1	C	C (100%)	C (100%)	T (14%)	C (100%)	
23,859,210	Intron 1	C	C (100%)	T (6%)*	T (15%)	C (100%)	
23,859,131	Intron 1	C	C (100%)	T (5%)*	T (13%)	C (100%)	

23,859,086	Intron 1	G	G (100%)	G (100%)	A (11%)	G (100%)	
23,859,050	Intron 1	G	A (27%)	A (36%)	A (47%)	G (97%)	
23,859,019	Intron 1	A	G (48%)	G (50%)	G (33%)	G (45%)	
23,859,017	Intron 1	A	C (47%)	C (43%)	C (65%)	C (43%)	
23,859,015	Intron 1	C	C (100%)	C (100%)	T (11%)	C (100%)	
23,858,984	Intron 1	G	T (48%)	T (43%)	T (41%)	T (46%)	
23,858,950	Intron 1	C	C (100%)	T (21%)	T (7%)*	C (100%)	
23,858,924	Intron 1	A	A (100%)	A (100%)	A (100%)	C (25%)	
23,858,901	Intron 1	C	C (100%)	T (3%)*	T (13%)	T (2%)*	
23,858,897	Intron 1	A	G (62%)	G (67%)	A (100%)	A (97%)	
23,858,775	Intron 1	A	G (95%)	G (93%)	G (87%)	A (100%)	
23,858,746	Intron 1	G	G (100%)	A (12%)	A (4%)*	G (100%)	
23,858,715	Intron 1	T	T (100%)	T (100%)	T (100%)	A (100%)	
23,858,705	Intron 1	G	G (100%)	A (9%)*	A (20%)	G (100%)	
23,858,701	Intron 1	G	G (100%)	A (12%)	G (100%)	G (100%)	
23,858,698	Intron 1	G	G (100%)	A (11%)	G (100%)	G (100%)	
23,858,689	Intron 1	C	C (100%)	C (100%)	T (13%)	C (100%)	
23,858,675	Intron 1	C	C (100%)	C (100%)	T (14%)	C (100%)	
23,858,614	Intron 1	C	G (100%)	G (95%)	G (91%)	C (96%)	
23,858,542	Intron 1	C	C (100%)	C (100%)	T (13%)	C (100%)	
23,858,526	Intron 1	G	G (100%)	G (100%)	A (18%)	G (100%)	
23,858,525	Intron 1	G	G (100%)	G (100%)	A (12%)	G (100%)	
23,858,521	Intron 1	G	G (100%)	A (6%)*	A (11%)	G (100%)	
23,858,517	Intron 1	G	G (100%)	G (100%)	A (11%)	G (100%)	
23,858,514	Intron 1	G	G (100%)	G (100%)	A (22%)	G (100%)	
23,858,509	Intron 1	G	G (98%)	A (11%)	G (100%)	G (100%)	
23,858,506	Intron 1	G	G (100%)	A (11%)	G (100%)	G (100%)	
23,858,428	Intron 1	A	G (100%)	G (100%)	G (100%)	G (100%)	
23,858,419	Intron 1	G	G (100%)	G (100%)	A (15%)	G (100%)	
23,858,324	Intron 1	C	C (100%)	T (12%)	T (5%)*	C (100%)	
23,858,263	Intron 1	C	C (100%)	C (100%)	T (25%)	C (100%)	
23,858,235	Exon 2	AT <u>G</u>	AT <u>G</u> (100%)	AT <u>A</u> (12%)	AT <u>G</u> (98%)	AT <u>G</u> (100%)	UAC→Y ₆₄ UAU →Y ₆₄
23,858,119	Intron 2	C	C (100%)	T (11%)	C (100%)	C (100%)	
23,858,110	Intron 2	C	C (100%)	T (4%)*	T (11%)	C (100%)	
23,858,107	Intron 2	G	G (100%)	G (100%)	A (11%)	G (100%)	

23,858,097	Intron 2	C	C (100%)	T (13%)	C (100%)	C (100%)
23,858,085	Intron 2	C	C (100%)	T (4%)*	T (14%)	C (100%)
23,858,067	Intron 2	C	C (100%)	C (100%)	T (17%)	C (97%)
23,858,062	Intron 2	G	A (2%)*	G (100%)	A (11%)	G (100%)
23,858,002	Intron 2	C	C (100%)	T (11%)	C (100%)	C (100%)
23,857,908	Intron 2	C	C (100%)	T (12%)	T (3%)*	C (100%)
23,857,868	Intron 2	C	C (100%)	T (2%)*	T (12%)	C (100%)
23,857,813	Intron 2	G	G (100%)	G (100%)	A (12%)	G (100%)
23,857,779	Intron 2	C	C (100%)	T (7%)*	T (12%)	C (100%)
23,857,772	Intron 2	C	C (100%)	C (100%)	T (12%)	C (100%)
23,857,767	Intron 2	C	C (100%)	T (3%)*	T (17%)	C (100%)
23,857,762	Intron 2	C	C (100%)	C (100%)	T (19%)	C (100%)
23,857,753	Intron 2	C	C (100%)	C (100%)	T (23%)	C (100%)
23,857,704	Intron 2	C	C (100%)	T (11%)	C (100%)	C (100%)
23,857,680	Intron 2	C	C (100%)	C (100%)	T (14%)	C (100%)
23,857,675	Intron 2	C	T (2%)*	T (19%)	T (5%)*	C (100%)
23,857,633	Intron 2	G	G (100%)	A (4%)*	A (20%)	G (100%)
23,857,624	Intron 2	C	C (100%)	C (100%)	T (11%)	C (100%)
23,857,615	Intron 2	C	C (100%)	C (95%)	T (12%)	C (100%)
23,857,603	Intron 2	C	T (3%)*	C (100%)	T (11%)	C (100%)
23,857,589	Intron 2	C	C (100%)	T (5%)*	T (14%)	C (100%)
23,857,530	Intron 2	G	G (100%)	A (5%)*	A (11%)	G (100%)
23,857,436	Intron 2	C	T (100%)	T (100%)	T (100%)	C (100%)
23,857,434	Intron 2	G	G (100%)	G (100%)	A (14%)	G (100%)
23,857,408	Intron 2	G	G (100%)	A (3%)*	A (14%)	G (100%)
23,857,366	Intron 2	C	C (98%)	T (8%)*	T (11%)	C (100%)
23,857,361	Intron 2	C	C (100%)	C (100%)	T (15%)	C (100%)
23,857,329	Intron 2	G	A (35%)	G (100%)	A (4%)*	G (100%)
23,857,261	Intron 2	C	C (100%)	T (11%)	T (10%)*	C (100%)
23,857,232	Intron 2	C	T (2%)*	C (100%)	T (18%)	C (100%)
23,857,230	Intron 2	C	C (98%)	C (100%)	T (15%)	C (100%)
23,857,228	Intron 2	C	C (98%)	T (4%)*	T (16%)	C (100%)
23,857,224	Intron 2	G	G (100%)	A (5%)*	A (11%)	G (100%)
23,857,208	Intron 2	G	G (100%)	A (5%)*	A (11%)	G (100%)
23,857,207	Intron 2	G	G (100%)	A (15%)	A (5%)*	G (100%)

23,857,206	Intron 2	G	G (100%)	A (10%)*	A (15%)	G (100%)
23,857,195	Intron 2	G	G (100%)	A (4%)*	A (15%)	G (100%)
23,857,183	Intron 2	G	G (100%)	A (14%)	A (8%)*	G (100%)
23,857,081	Intron 2	G	G (100%)	A (4%)*	A (13%)	G (95%)
23,857,075	Intron 2	G	G (100%)	A (4%)*	A (14%)	G (95%)
23,857,068	Intron 2	C	C (98%)	T (4%)*	T (27%)	C (100%)
23,857,062	Intron 2	G	G (100%)	G (100%)	A (13%)	G (95%)
23,857,058	Intron 2	C	C (100%)	C (100%)	T (13%)	C (100%)
23,857,044	Intron 2	C	C (100%)	C (100%)	T (13%)	T (5%)*
23,857,043	Intron 2	C	C (100%)	C (100%)	T (12%)	C (100%)
23,857,034	Intron 2	C	C (100%)	T (7%)*	T (14%)	C (100%)
23,857,027	Intron 2	C	C (100%)	T (12%)	T (6%)*	C (100%)
23,857,021	Intron 2	C	C (100%)	C (100%)	T (13%)	C (100%)
23,857,020	Intron 2	C	C (100%)	C (100%)	T (11%)	C (100%)
23,857,012	Intron 2	G	G (100%)	A (11%)	G (100%)	G (100%)
23,857,010	Intron 2	G	G (100%)	A (12%)	G (100%)	G (100%)
23,856,990	Intron 2	G	G (100%)	A (13%)	G (100%)	G (100%)
23,856,956	Intron 2	G	G (100%)	A (13%)	A (3%)*	G (100%)
23,856,822	3' UTR	C	C (100%)	C (100%)	T (11%)	C (100%)
23,856,812	3' UTR	C	C (100%)	T (6%)*	T (14%)	C (100%)
23,856,781	3' UTR	G	G (100%)	G (100%)	A (18%)	G (100%)
23,856,763	3' UTR	G	G (100%)	A (9%)*	A (15%)	G (100%)
23,856,748	3' UTR	G	G (100%)	G (100%)	A (11%)	G (100%)
23,856,745	3' UTR	G	G (100%)	A (15%)	A (5%)*	G (100%)
23,856,712	3' UTR	G	G (100%)	G (100%)	A (11%)	G (100%)
23,856,711	3' UTR	G	G (100%)	A (9%)*	A (11%)	G (100%)
23,856,709	3' UTR	C	C (100%)	C (95%)	T (11%)	C (100%)
23,856,663	3' UTR	T	C (97%)	C (91%)	C (100%)	C (100%)
23,856,661	3' UTR	C	C (100%)	T (11%)	C (100%)	C (100%)
23,856,657	3' UTR	A	A (100%)	G (11%)	A (100%)	A (100%)
23,856,655	3' UTR	G	G (100%)	A (11%)	A (8%)*	G (100%)
23,856,654	3' UTR	G	G (100%)	A (6%)*	A (14%)	G (100%)
23,856,639	3' UTR	G	G (100%)	A (5%)*	A (13%)	G (100%)
23,856,624	3' UTR	G	G (100%)	A (12%)	A (12%)	G (100%)
23,856,623	3' UTR	G	G (100%)	G (100%)	A (12%)	G (100%)

23,856,601	3' UTR	G	G (100%)	A (12%)	G (100%)	G (100%)
23,856,531	Downstream region	C	C (98%)	T (11%)	T (4%)*	C (100%)
23,856,478	Downstream region	C	C (100%)	T (4%)*	T (13%)	C (100%)
23,856,475	Downstream region	C	C (100%)	C (100%)	T (14%)	C (95%)
23,856,461	Downstream region	C	C (100%)	T (4%)*	T (14%)	C (95%)
23,856,444	Downstream region	C	C (100%)	C (100%)	T (17%)	C (100%)
23,856,432	Downstream region	C	C (100%)	T (6%)*	T (12%)	T (4%)*
23,856,407	Downstream region	C	C (100%)	T (15%)	T (4%)*	C (100%)
23,856,338	Downstream region	C	C (100%)	T (11%)	T (2%)*	C (100%)
23,856,305	Downstream region	G	G (100%)	A (11%)	A (17%)	G (100%)
23,856,263	Downstream region	G	G (100%)	A (13%)	G (100%)	G (100%)
23,856,223	Downstream region	C	C (100%)	T (11%)	T (5%)*	C (100%)
23,856,186	Downstream region	G	G (100%)	A (15%)	A (2%)*	G (100%)
23,856,165	Downstream region	G	G (100%)	A (14%)	G (100%)	G (97%)
23,856,108	Downstream region	C	C (100%)	C (100%)	T (13%)	C (100%)
23,856,092	Downstream region	G	G (100%)	A (11%)	G (100%)	G (100%)
23,856,083	Downstream region	G	G (100%)	A (13%)	G (100%)	G (100%)
23,856,029	Downstream region	C	C (100%)	T (11%)	T (5%)*	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.