

Table S6. Nucleotide Substitutions in *HTN1* 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
70,915,835	Upstream regions	G	G (100%)	A (6%)*	A (12%)	A (3%)*	
70,915,946	Upstream regions	A	A (100%)	A (100%)	G (14%)	A (100%)	
70,915,954	Upstream regions	G	G (100%)	G (100%)	A (12%)	C (4%)*	
70,915,975	Upstream regions	C	C (100%)	T (12%)	T (8%)*	C (100%)	
70,915,993	Upstream regions	C	C (100%)	C (100%)	T (16%)	C (100%)	
70,916,067	Upstream regions	A	G (12%)	G (4%)*	G (10%)*	A (100%)	
70,916,151	5'UTR	G	G (98%)	G (100%)	A (11%)	G (100%)	
70,916,157	5'UTR	C	T (4%)*	C (100%)	T (12%)	C (100%)	
70,916,201	5'UTR	C	C (100%)	T (14%)	C (100%)	C (100%)	
70,916,229	Intron 1	C	C (99%)	C (100%)	T (12%)	C (100%)	
70,916,433	Intron 1	T	G (93%)	G (86%) ** A (14%) **	G (67%) ** A (33%) **	T (100%)	
70,916,454	Intron 1	C	C (100%)	C (100%)	T (15%)	C (100%)	
70,916,507	Intron 1	G	G (97%)	A (5%)*	A (15%)	G (100%)	
70,916,513	Intron 1	G	G (100%)	A (13%)	G (100%)	G (100%)	
70,916,527	Intron 1	G	A (100%)	A (100%)	A (100%)**	A (100%)**	
70,916,549	Intron 1	G	A (7%)*	A (7%)*	A (15%)	T (8%)*	
70,916,629	Intron 1	C	C (100%)	T (4%)*	T (11%)	C (100%)	
70,916,745	Intron 1	C	C (100%)	T (11%)	C (100%)	C (100%)	
70,916,801	Intron 1	G	G (100%)	G (100%)	A (13%)	G (100%)	
70,916,810	Intron 1	C	T (2%)*	C (100%)	T (10%)*	T (100%)	
70,916,855	Intron 1	C	G (100%)	G (100%)	G (100%)	G (100%)	
70,916,970	Intron 1	C	C (100%)	T (13%)	T (3%)*	C (100%)	
70,917,037	Intron 1	G	G (100%)	A (3%)*	A (13%)	G (100%)	
70,917,063	Intron 1	G	G (100%)	A (13%)	A (3%)*	G (100%)	
70,917,193	Intron 1	G	A (100%)	A (100%)	A (100%)	A (100%)	
70,917,298	Intron 1	C	C (100%)	C (94%)	T (14%)	C (100%)	
70,917,390	Intron 1	C	C (100%)	T (11%)	C (100%)	C (100%)	
70,917,413	Intron 1	T	A (98%)	A (100%)	A (100%)	A (100%)	
70,917,415	Intron 1	G	G (100%)	A (11%)	A (3%)*	G (100%)	
70,917,429	Intron 1	A	G (98%)	G (97%)	G (100%)	G (100%)	

70,917,485	Intron 1	G	A (2%)*	G (100%)	A (11%)	G (100%)
70,917,487	Intron 1	G	G (100%)	G (100%)	A (16%)	G (100%)
70,917,494	Intron 1	G	G (100%)	A (11%)	A (6%)*	G (100%)
70,917,498	Intron 1	G	A (2%)*	A (5%)*	A (19%)	G (100%)
70,917,669	Intron 1	A	C (100%)	C (100%)	C (100%)	A (100%)
70,917,801	Intron 1	C	C (100%)	C (100%)	T (13%)	C (100%)
70,917,152	Intron 1	C	C (100%)	T (3%)*	T (11%)	C (100%)
70,917,227	Intron 1	C	C (100%)	C (100%)	T (17%)	C (100%)
70,917,230	Intron 1	C	C (100%)	T (11%)	C (100%)	C (100%)
70,917,401	Intron 1	C	C (100%)	T (12%)	C (100%)	C (100%)
70,917,452	Intron 1	G	G (100%)	A (15%)	A (2%)*	G (100%)
70,917,509	Intron 1	C	A (2%)*	T (11%)	T (9%)*	T (3%)*
70,918,511	Intron 1	A	delA (15%)	delA (48%)	delA (32%)	delA (21%)
70,918,512	Intron 1	A	delA (5%)*	delA (14%)	delA (11%)	delA (3%)*
70,918,551	Intron 1	G	G (100%)	A (4%)*	A (15%)	G (100%)
70,918,555	Intron 1	C	C (100%)	T (10%)*	T (15%)	C (100%)
70,918,557	Intron 1	C	C (100%)	C (100%)	T (15%)	C (100%)
70,918,559	Intron 1	C	T (2%)*	C (100%)	T (17%)	C (100%)
70,918,620	Intron 1	G	A (2%)*	A (14%)	A (5%)*	G (100%)
70,918,780	Intron 1	G	G (100%)	A (3%)*	A (11%)	G (100%)
70,918,901	Intron 2	C	T (2%)*	T (6%)*	T (13%)	C (100%)
70,918,978	Intron 2	G	G (100%)	A (8%)*	A (12%)	G (100%)
70,918,980	Intron 2	G	G (100%)	A (8%)*	A (12%)	G (100%)
70,918,984	Intron 2	C	C (100%)	T (3%)*	T (13%)	C (97%)
70,919,320	Intron 2	A	C (100%)	C (100%)	C (100%)	A (100%)
70,919,342	Intron 2	A	A (100%)	A (100%)	A (100%)	G (100%)
70,919,353	Intron 2	G	A (2%)*	A (14%)	A (8%)*	G (100%)
70,919,355	Intron 2	C	C (100%)	T (3%)*	T (13%)	C (100%)
70,919,413	Intron 2	C	C (100%)	T (4%)*	T (11%)	C (100%)
70,919,418	Intron 2	T	C (100%)	C (100%)	C (100%)	T (100%)
70,919,448	Intron 2	C	C (100%)	T (11%)	C (100%)	C (100%)
70,919,615	Intron 2	G	G (100%)	A (11%)	G (100%)	G (100%)
70,919,739	Intron 2	A	G (95%)	G (100%)	G (97%)	A (100%)
70,919,775	Intron 2	C	C (100%)	T (13%)	T (5%)*	C (100%)
70,919,777	Intron 2	G	G (100%)	A (4%)*	A (14%)	G (100%)

70,919,796	Intron 2	T	G (95%)	G (100%)	G (92%)	T (100%)	
70,919,872	Intron 2	C	C (100%)	C (100%)	T (11%)	C (100%)	
70,920,060	Intron 3	G	G (98%)	A (11%)	A (7%)*	G (100%)	
70,920,149	Exon 4	GGG	GG <u>A</u> (2%)*	GG <u>A</u> (5%)*	GG <u>A</u> (17%)	GGG (100%)	GGG→G <sub>9</sub> GGA→G <sub>9</sub>
70,920,408	Intron 4	A	A (100%)	A (97%)	T (60%)	A (100%)	
70,920,473	Intron 4	G	G (100%)	A (10%)*	A (14%)	G (100%)	
70,920,499	Intron 4	G	A (5%)*	A (3%)*	A (12%)	G (100%)	
70,920,608	Intron 4	A	G (95%)	G (100%)	G (100%)	A (100%)	
70,920,715	Intron 4	G	G (100%)	A (17%)	G (100%)	G (100%)	
70,920,879	Intron 4	C	C (100%)	C (100%)	T (11%)	C (100%)	
70,920,880	Intron 4	C	C (100%)	C (100%)	T (11%)	C (100%)	
70,920,916	Intron 4	G	G (100%)	G (100%)	A (11%)	G (100%)	
70,920,989	Intron 4	G	A (2%)*	G (96%)	A (13%)	G (100%)	
70,921,018	Intron 4	C	C (100%)	C (100%)	T (12%)	C (100%)	
70,921,122	Intron 4	G	A (98%)	A (100%)	A (100%)	G (100%)	
70,921,164	Intron 4	T	C (99%)	C (90%)	C (97%)	C (100%)	
70,921,174	Intron 4	C	C (99%)	T (8%)*	T (15%)	C (100%)	
70,921,295	3'UTR	G	G (100%)	A (14%)	A (5%)*	G (100%)	
70,921,306	3'UTR	G	G (100%)	G (96%)	A (11%)	G (100%)	
70,921,319	3'UTR	G	G (100%)	G (100%)	A (11%)	G (100%)	
70,921,329	Intron 5	C	C (100%)	C (100%)	T (11%)	C (100%)	
70,921,331	Intron 5	C	C (100%)	C (100%)	T (11%)	C (100%)	
70,921,337	Intron 5	G	A (98%)	A (100%)	A (100%)	A (100%)	
70,921,357	Intron 5	C	C (100%)	T (11%)	T (10%)*	C (100%)	
70,921,393	Intron 5	C	T (2%)*	T (16%)	C (100%)	T (4%)*	
70,921,419	Intron 5	C	C (98%)	C (100%)	C (96%)	A (100%)	
70,921,450	Intron 5	G	A (98%)	A (100%)	A (100%)	A (100%)	
70,921,525	Intron 5		InsT (92%)	InsC (45%)	InsC (44%)	InsT (100%)	
70,921,537	Intron 5	G	G (100%)	G (100%)	A (20%)	G (100%)	
70,921,564	Intron 5	C	C (100%)	T (20%)	T (15%)	C (100%)	
70,921,603	Intron 5	C	C (100%)	T (5%)*	T (11%)	C (96%)	
70,921,704	Intron 5	A	G (93%)	G (96%)	G (67%)	A (100%)	
70,921,706	Intron 5	G	G (98%)	A (4%)*	A (14%)	G (100%)	
70,921,707	Intron 5	G	A (2%)*	A (4%)*	A (13%)	G (100%)	

70,921,710	Intron 5	A	G (89%)	G (89%)	G (72%)	A (100%)
70,921,724	Intron 5	G	G (100%)	G (96%)	A (11%)	G (100%)
70,921,748	Intron 5	A	G (93%)	G (81%)	G (59%)	A (100%)
70,921,788	Intron 5	A	T (92%)	T (81%)	T (72%)	A (100%)
70,921,911	Intron 5	T	C (52%)	C (49%)	C (26%)	T (100%)
70,921,969	Intron 5	G	A (3%)*	G (100%)	A (13%)	G (100%)
70,922,002	Intron 5	A	G (100%)	G (97%)	G (93%)	A (100%)
70,922,084	Intron 5	C	C (100%)	T (14%)	C (100%)	C (97%)
70,922,163	Intron 5	C	C (100%)	C (100%)	T (17%)	C (100%)
70,922,167	Intron 5	G	G (100%)	G (100%)	A (14%)	G (100%)
70,922,174	Intron 5	C	C (100%)	T (11%)	T (27%)	T (4%)*
70,922,175	Intron 5	C	T (2%)*	T (5%)*	T (27%)	C (100%)
70,922,194	Intron 5	C	C (100%)	C (100%)	T (13%)	C (100%)
70,922,298	Intron 5	A	A (100%)	A (100%)	A (100%)	G (43%)
70,922,383	Intron 5	C	T (2%)*	T (3%)*	T (11%)	C (100%)
70,922,567	Intron 5	C	C (100%)	C (100%)	T (12%)	C (100%)
70,922,656	Intron 5	G	G (100%)	A (11%)	A (7%)*	A (3%)*
70,922,713	Intron 5	G	G (100%)	A (12%)	G (100%)	G (100%)
70,922,717	Intron 5	C	T (2%)*	C (100%)	T (14%)	C (100%)
70,922,892	Intron 5	G	A (2%)*	A (13%)	G (100%)	G (100%)
70,923,008	Intron 5	A	T (5%)*	A (100%)	T (12%)	A (100%)
70,923,009	Intron 5	A	C (5%)*	A (100%)	C (12%)	A (100%)
70,923,015	Intron 5	C	T (4%)*	C (100%)	T (12%)	C (100%)
70,923,066	Intron 5	G	G (100%)	A (4%)*	A (11%)	G (100%)
70,923,077	Intron 5	G	G (100%)	A (5%)*	A (13%)	G (100%)
70,923,088	Intron 5	C	C (100%)	C (100%)	T (14%)	C (100%)
70,923,101	Intron 5	G	G (100%)	A (13%)	G (100%)	G (100%)
70,923,105	Intron 5	G	G (100%)	A (5%)*	A (15%)	G (100%)
70,923,108	Intron 5	G	G (97%)	A (4%)*	A (17%)	G (100%)
70,923,123	Intron 5		InsTT (100%)	InsTT (46%)	InsTT (50%)	-
70,923,154	Intron 5	C	T (93%)	T (90%)	T (50%)	C (100%)
70,923,180	Intron 5	G	G (100%)	G (100%)	A (11%)	G (100%)
70,923,254	Intron 5	C	C (100%)	T (5%)*	T (13%)	C (100%)
70,923,276	Intron 5	C	C (100%)	C (100%)	T (12%)	C (100%)
70,923,278	Intron 5	C	C (100%)	C (96%)	T (12%)	C (100%)

70,923,315	Intron 5	G	A (2%)*	A (10%)*	A (12%)	G (100%)
70,923,371	Intron 5	C	T (4%)*	C (100%)	T (13%)	T (6%)*
70,923,380	Intron 5	C	T (100%)	T (100%)	T (100%)	C (100%)
70,923,451	Intron 5	G	A (2%)*	G (100%)	A (11%)	G (100%)
70,923,457	Intron 5	G	G (100%)	A (5%)*	A (15%)	G (100%)
70,923,461	Intron 5	G	G (100%)	G (100%)	A (11%)	G (100%)
70,923,493	Intron 5	C	C (100%)	C (100%)	T (12%)	C (100%)
70,923,498	Intron 5	A	T (97%)	T (100%)	T (100%)	A (100%)
70,923,630	Intron 5	G	G (100%)	G (100%)	A (15%)	G (100%)
70,923,635	Intron 5	G	G (98%)	A (3%)*	A (11%)	G (100%)
70,923,826	Intron 5	C	C (99%)	T (2%)*	T (11%)	C (100%)
70,923,913	Intron 5	C	C (97%)	T (7%)*	T (12%)	C (100%)
70,923,922	Intron 5	C	C (100%)	C (100%)	T (11%)	C (100%)
70,923,931	Intron 5	T	G (98%)	G (100%)	G (100%)	G (100%)
70,923,932	Intron 5	A	C (98%)	C (100%)	C (94%)	A (100%)
70,923,940	Intron 5	G	G (100%)	A (44%)	A (13%)	G (100%)
70,923,943	Intron 5	G	G (100%)	G (100%)	A (17%)	G (100%)
70,924,022	Intron 5	G	G (100%)	G (100%)	A (16%)	G (100%)
70,924,034	Intron 5	G	G (100%)	A (4%)*	A (11%)	G (100%)
70,924,036	Intron 5	G	G (98%)	G (100%)	A (14%)	G (100%)
70,924,091	Intron 5	G	G (99%)	A (11%)	G (100%)	G (100%)
70,924,131	Intron 5	C	C (100%)	T (11%)	C (100%)	C (100%)
70,924,200	Intron 5	C	C (100%)	T (12%)	C (100%)	C (100%)
70,924,204	Intron 5	C	C (100%)	T (8%)*	T (15%)	C (100%)
70,924,250	Intron 5	G	A (2%)*	A (11%)	G (100%)	G (100%)
70,924,345	3'UTR	C	C (100%)	T (3%)*	T (11%)	C (100%)
70,924,346	3'UTR	C	C (100%)	T (3%)*	T (26%)	T (3%)*
70,924,358	3'UTR	C	C (100%)	T (14%)	C (100%)	C (100%)
70,924,359	3'UTR	C	C (100%)	T (15%)	C (100%)	A (3%)*
70,924,419	3'UTR	T	T (100%)	T (100%)	T (100%)	C (98%)
70,924,467	3'UTR	G	G (100%)	A (14%)	A (6%)*	G (100%)
70,924,568	Downstream regions	T	C (98%)	C (100%)	C (89%)	T (100%)
70,924,569	Downstream regions	G	A (5%)*	A (10%)*	A (12%)	G (100%)
70,924,574	Downstream regions	C	C (100%)	T (11%)	T (13%)	C (100%)

70,924,576	Downstream regions	C	C (100%)	C (100%)	T (12%)	C (100%)
70,924,584	Downstream regions	C	C (100%)	T (13%)	C (100%)	C (100%)
70,924,585-70,924,587	Downstream regions	TCT	delTCT (98%)	delTCT (74%)	delTCT (93%)	TCT (100%)
70,924,658	Downstream regions	G	G (100%)	A (13%)	A (3%)*	G (100%)
70,924,661	Downstream regions	C	C (100%)	C (100%)	T (12%)	C (100%)
70,924,669	Downstream regions	C	T (80%)	T (96%)	T (76%)	T (3%)*
70,924,671	Downstream regions	T	C (76%)	C (90%)	C (73%)	T (100%)
70,924,735	Downstream regions	G	G (100%)	G (100%)	A (16%)	G (100%)
70,924,736	Downstream regions	G	G (100%)	A (13%)	A (11%)	G (100%)
70,924,747	Downstream regions	C	C (100%)	C (100%)	T (11%)	C (100%)
70,924,758	Downstream regions	C	C (100%)	T (13%)	C (100%)	C (100%)
70,924,767	Downstream regions	C	C (100%)	C (100%)	T (12%)	C (100%)
70,924,794	Downstream regions	G	G (96%)	A (11%)	A (7%)*	G (100%)
70,924,833	Downstream regions	C	delC (97%)	delC (79%)	delC (95%)	C (100%)
70,924,852	Downstream regions	G	G (100%)	A (11%)	A (2%)*	G (100%)
70,924,873	Downstream regions	C	C (100%)	T (6%)*	T (18%)	C (96%)
70,924,894	Downstream regions	G	G (100%)	G (100%)	A (12%)	G (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.