Supplementary Information SI6

Conservation of Ancient Hominin *AHR* 5'-UTR 185G Variant within Primates

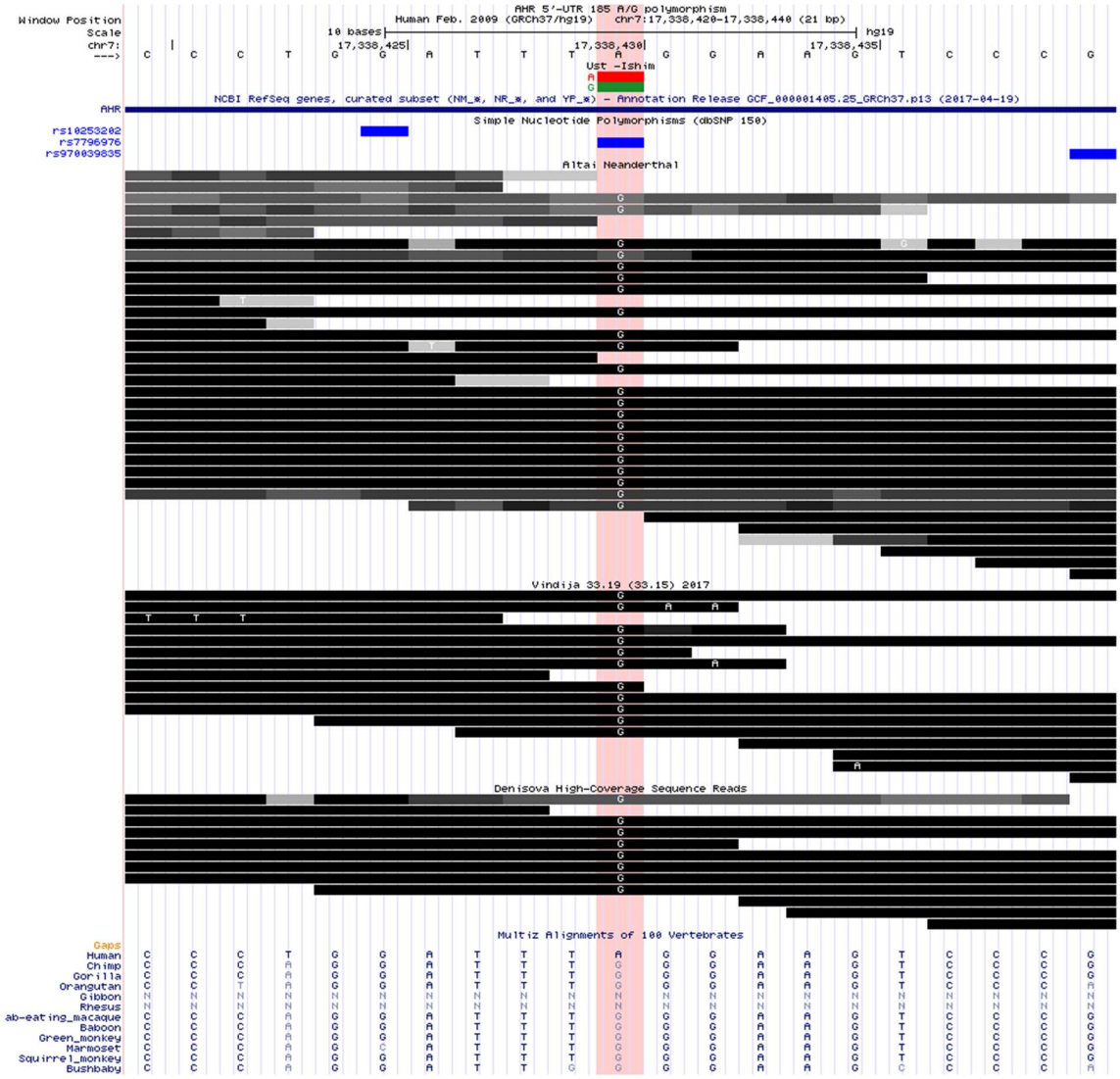

Fig. SI6.1. Using the UCSC Genome Browser the *AHR* mRNA exon 1 sequence around the 185A/G variant site (pink highlighting) from the modern human reference genome (assembly GRCh37/hg19; upper track) was aligned with the variants (VCF-format) observed in the genome of an ancient anatomically modern human (45 kya) found at Ust'-Ishim in Siberia (Fu et al. 2014) (second track), documenting that this individual is heterozygous (A/G) at position 185 (numbering of human AHR reference transcript; Ensembl ID: ENST00000242057.8). The third track demonstrates that the sequence shown is part of the RefSeq human *AHR* mRNA sequence NM_001621.4. The fourth track shows that the 185A/G variation is known in present-day humans as reference single nucleotide polymorphism (SNP) rs7796976 in dbSNP. The three tracks below show the alignment of the sequence reads obtained from the Altai-Neanderthal (Prüfer et al. 2014), Vindija-Neanderthal (Prüfer et al. 2017) and Denisovan (Meyer et al. 2012) genome, respectively, comprising only 185G reads. The bottom track displays the primate section of the "Multiz Alignments of 100 Vertebrates" available in the UCSC Genome Browser, showing that the 185G variant is highly conserved within primates, and the 185A variant is a newly derived allele, unique for the modern human lineage. The 185A allele frequency in the present-day human population is 21% (dbSNP, build 153, rs7796976, TopMed data set).

References

Fu Q, Li H, Moorjani P, Jay F, Slepchenko SM, Bondarev AA, Johnson PLF, Aximu-Petri A, Prufer K, de Filippo C et al. 2014. Genome sequence of a 45,000-year-old modern human from western Siberia. *Nature* 514(7523):445-449.

Meyer M, Kircher M, Gansauge MT, Li H, Racimo F, Mallick S, Schraiber JG, Jay F, Prufer K, de Filippo C et al. 2012. A high-coverage genome sequence from an archaic Denisovan individual. *Science* 338(6104):222-226.

Prüfer K, de Filippo C, Grote S, Mafessoni F, Korlević P, Hajdinjak M, Vernot B, Skov L, Hsieh P, Peyrégne S et al. 2017. A high-coverage Neandertal genome from Vindija Cave in Croatia. *Science*.

Prüfer K, Racimo F, Patterson N, Jay F, Sankararaman S, Sawyer S, Heinze A, Renaud G, Sudmant PH, de Filippo C et al. 2014. The complete genome sequence of a Neanderthal from the Altai Mountains. *Nature* 505(7481):43-49.