

The Genetic Links Between Archaic and Modern Humans

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Abstract. Our modern physiology is the mixture of many archaic humans that once roamed our planet. The evidence of these archaic humans is still present in our DNA. This poster reviews how our understanding of ancient human genetics has drastically changed due to advances in molecular genetics. Neanderthal and Denisovan remains have been sequenced for nuclear and mitochondrial DNA. Neanderthal and Denisovan genetic ancestry have been identified by genomic studies in modern human populations across Eurasia and Pacific Island regions. Studies have shown a gene flow of $4 \pm 1\%$ from Neanderthals to present-day Eurasians. Whereas Papuan and Melanesian individuals share $4 \pm 0.7\%$ more Denisovan alleles than other individuals studied. Neanderthal and Denisovan alleles have been linked to innate immunity. Neanderthal alleles have been linked to speech and environmental adaptations in modern humans. Neanderthal and Denisovan genes present in modern day humans continue to shape our biology. The study of our ancestors provides new insights about their influence in human genomic diversity.



Figure 1 Recreation of (a) a Denisovan girl from Siberian cave in Russia. Recreation of (b) a Neanderthal woman from Saint Césaire site in France. (Pictures adapted from Science Magazine and Smithsonian Magazine.)

References

- ¹Abi-Rached L., et al. (2011) The Shaping of Modern Human Immune System by Multiregional Admixture with Archaic Humans. *Science Express* 334 doi:10.1126/science.1209202
- ²Browning et al. (2018) Analysis of human sequence data reveals two pulses of archaic Denisovan admixture Cell 173, 53–61 doi:10.1016/j.cell.2018.02.031
- ³Sankararaman S., et al. (2014) The genomic landscape of Neanderthal ancestry in present-day humans. *Nature* 507 doi:10.1038/nature12961
- ⁴Reich et al., (2010) Genetic history of an archaic hominin group from Denisova Cave in Siberia. *Nature* 468 doi:10.1038/nature09710
- ⁵Vernot B., Akey M. J. (2014) Resurrecting surviving neanderthal lineages from modern human genomes. *Science* doi:10.1126/science.1245938

•DNA analysis suggests that Denisovan admixture happened in two different events involving two different Denisovan populations with modern human populations (Browning et. al., 2018). In contrast, evidence suggest only one Neanderthal admixture event with modern human or close related Neanderthal populations mated with modern humans.

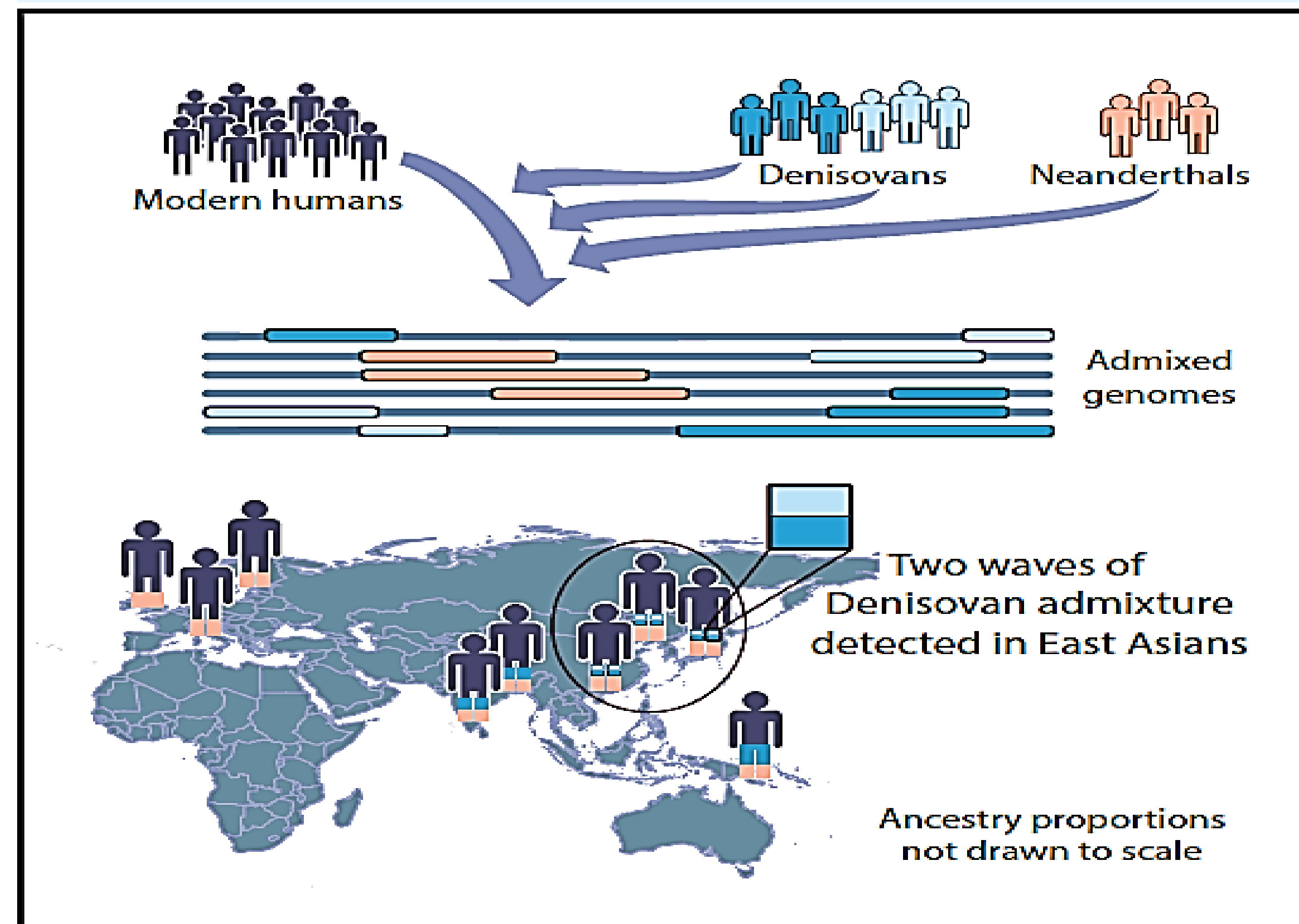


Figure 2 Schematic demonstrating the admixture between modern humans, Denisovans and Neanderthals and their geographical location. (Figure from Browning et. al.,2018)²

- The human leukocyte antigen (HLA) complex helps the immune system distinguish the body's own proteins from proteins made by foreign invaders such as viruses and bacteria.
- Denisovan and Neanderthal haplotypes of this HLA complex are present in modern humans (Abi-Rached et. al., 2011).
- The existence of Denisovan HLA-A-C haplotypes in modern humans suggest introgressive hybridization, transfer of genetic material from one species to another, from Denisovans. Denisovan admixture influenced HLA systems in Asians and Amerindians (Abi-Rached et. al., 2011).
- Neanderthal HLA-A-C haplotypes are present in high frequencies in Eurasia. Neanderthal admixture in Eurasia caused the introgression of these haplotypes in modern humans (Abi-Rached et. al., 2011).

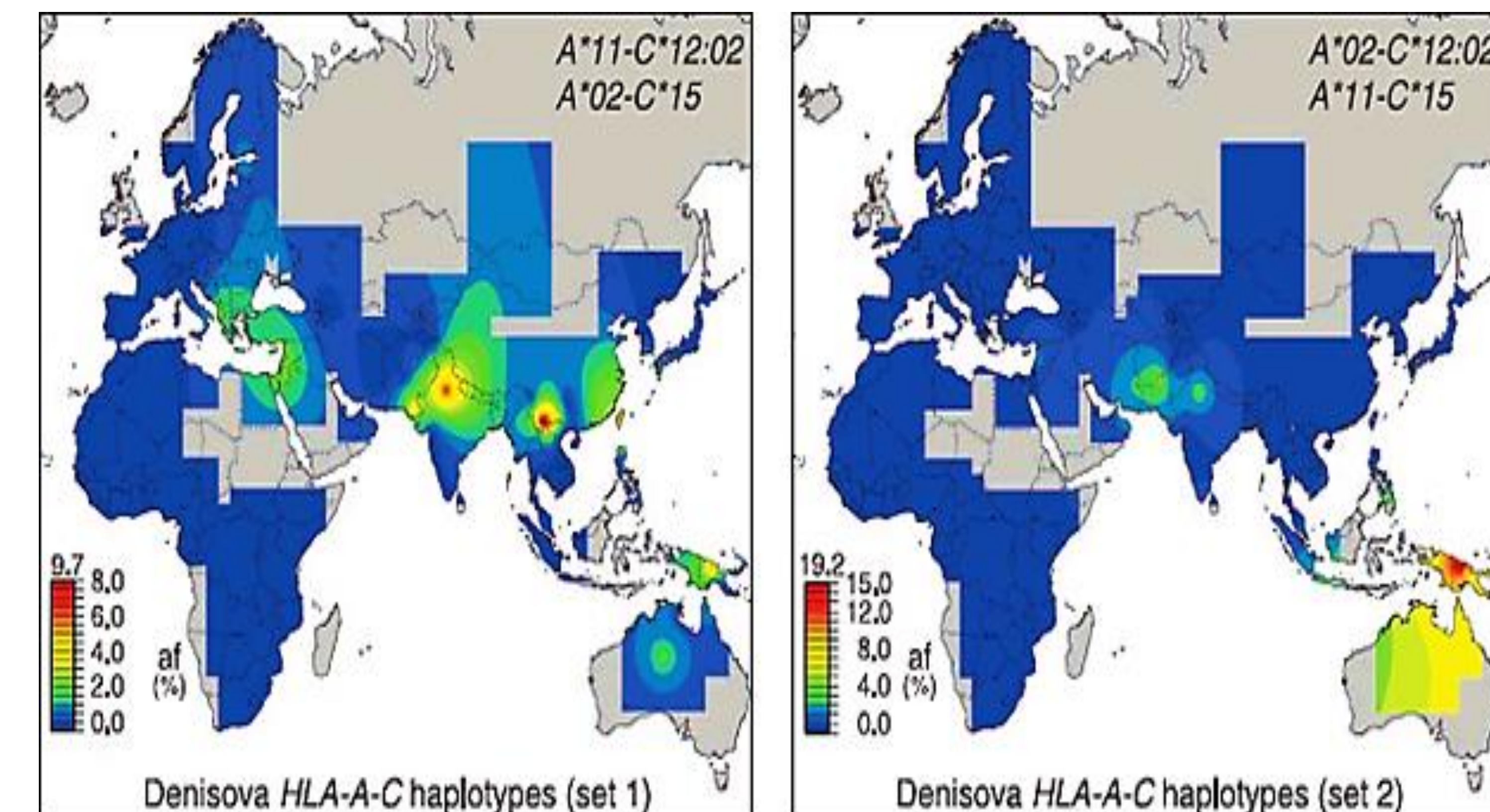


Figure 3 Worldwide distributions of the two possible Denisovan HLA-A-C haplotype combinations. Both are present in modern Asians and Oceanians but absent from sub-Saharan Africans. (Figure from Abi-Rached et. al., 2011)¹

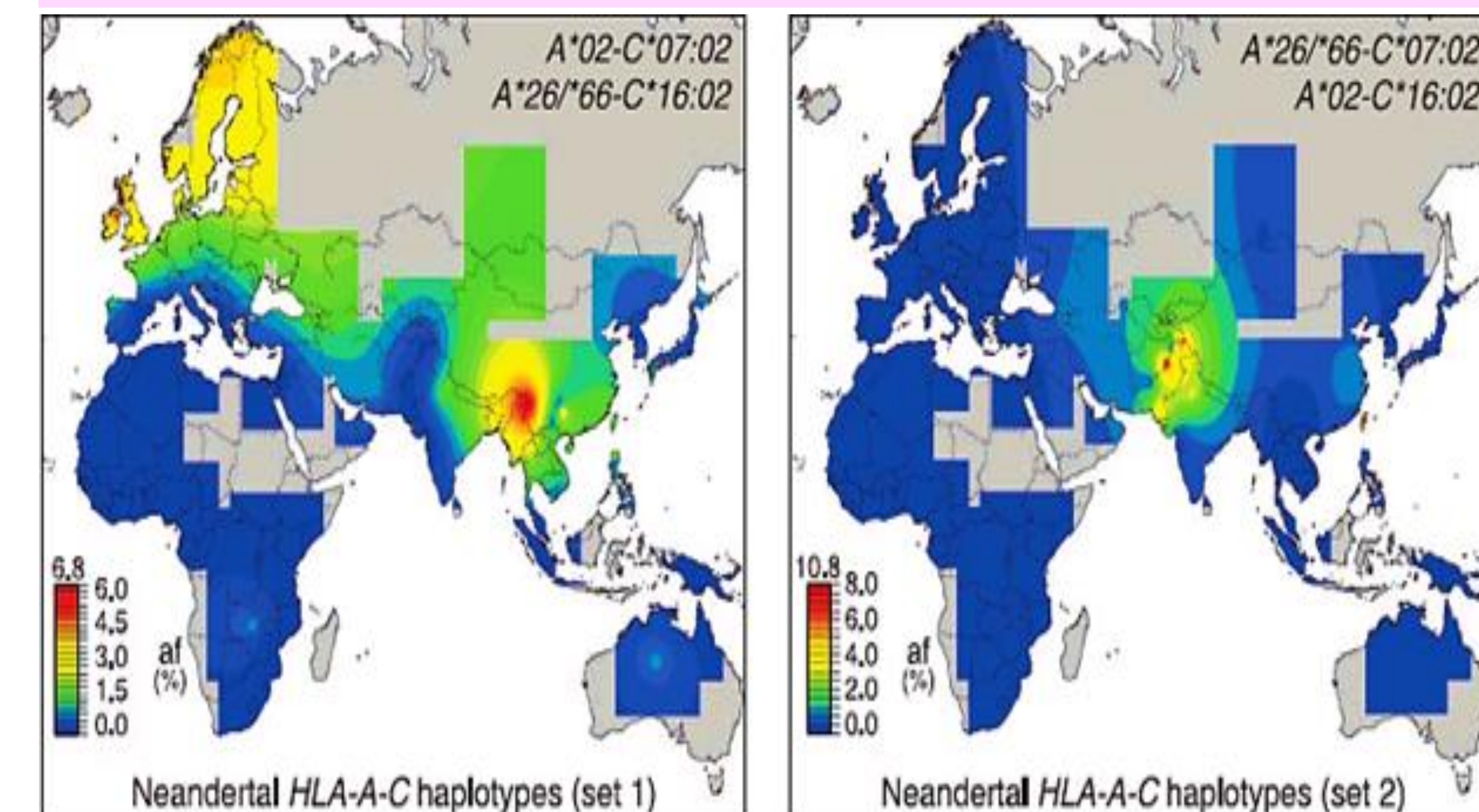


Figure 4 Worldwide distributions of the two possible Neanderthal HLA-A-C haplotype combinations. Both are present in modern Eurasians but absent from sub-Saharan Africans. (Figure from Abi-Rached et. al., 2011)¹

- Mating events between modern humans and archaic humans increased the fitness of migrant modern humans in the presence of new unknown pathogens.
- Adaptive introgression of HLA complex alleles toughen modern humans' lymphocytes, which are essential for immunity and reproduction (Abi-Rached et. al., 2011).
- As DNA technologies advance, the study of ancient humans may reveal new hypotheses about human evolutionary patterns. Genome comparisons can elucidate new aspects of our adaptive legacy. This can broaden our understanding of modern human biology.

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