MITOCHONDRIAL PSEUDOGENES, GENE FLOW AMONG AFRICAN HOMININS AND HOMO HEIDELBERGENSIS

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Mitochondrial pseudogenes or nuclear DNA sequences of mitochondrial origin (numts) represent the fossils of vanished mtDNA molecules. Recent numt sequences have been inserted into the human nuclear genome after the reproductive separation of the hominin and chimpanzee lineages. In 2010 I characterized a 5841-bp numt on the human chromosome 1p36.33, called mtAncestor-1, and made a conclusion that it is likely to be a molecular relic of the mtDNA of Homo heidelbergensis. MtAncestor-1 was transferred into the nuclear genome of a common ancestor of Neanderthals and modern humans approximately 620,000 (440,000-820,000) years ago. In 2010 and 2014 the mtDNA sequences were published from the hominin remains found in Denisova Cave (Siberia, Russia) and Sima de los Huesos (SH) Cave (northern Spain). Phylogenetic analysis placed both sequences in the same group as mtAncestor-1 with a strong statistical support. This group diverged 840,000 (610,000–1,100,000) years ago from another cluster composing of the modern human and Neanderthal mtDNA sequences. The subsequent divergence of the human and Neanderthal mtDNAs starting 450,000 (320,000-600,000) years ago as well as the evolutionary radiation of the heidelbergensis-like (mtAncestor-1, Denisova and SH) mtDNAs approximately 570,000 (390,000–790,000) years ago demonstrated a deep division of mtDNA lineages that existed among African hominins in the Lower and Middle Pleistocene. The transposition of the heidelbergensis-like mtDNA to the modern human and Neanderthal nuclear genomes provided for the first time the genetic evidence of gene flow from one African hominin group to another. This mtDNA illustrates the evolutionary connections between the African H. heidelbergensis, the Denisova and SH hominins of Eurasia, and the ancestral African population of modern humans and Neanderthals. Assuming that the SH hominins belonged to the Neanderthal lineage, it is plausible to suggest the broad distribution of the heidelbergensis-like mtDNA outside Africa among the Eurasian hominins, including the daughter lineages of H. heidelbergensis and the primitive forms of Neanderthals. Hence, this type of mtDNA cannot serve as an evolutionary marker identifying new hominins in Eurasia in the absence of their remains. Instead, the ancient genome data should be considered in conjunction with paleoanthropological record.

Key words: mitochondrial pseudogenes, mtAncestor-1, Homo heidelbergensis, mtDNA, gene flow

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