

Fossils within fossils: Using HPC in the classroom to test for ancient hybridization in Neanderthal genomes

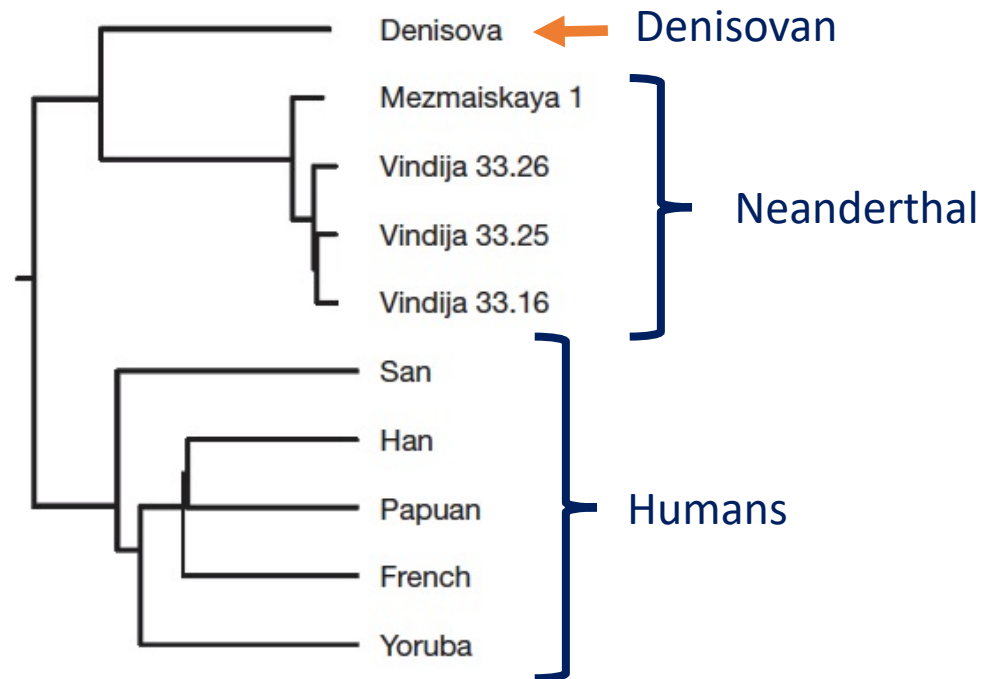


BIOL 435/535: Bioinformatics

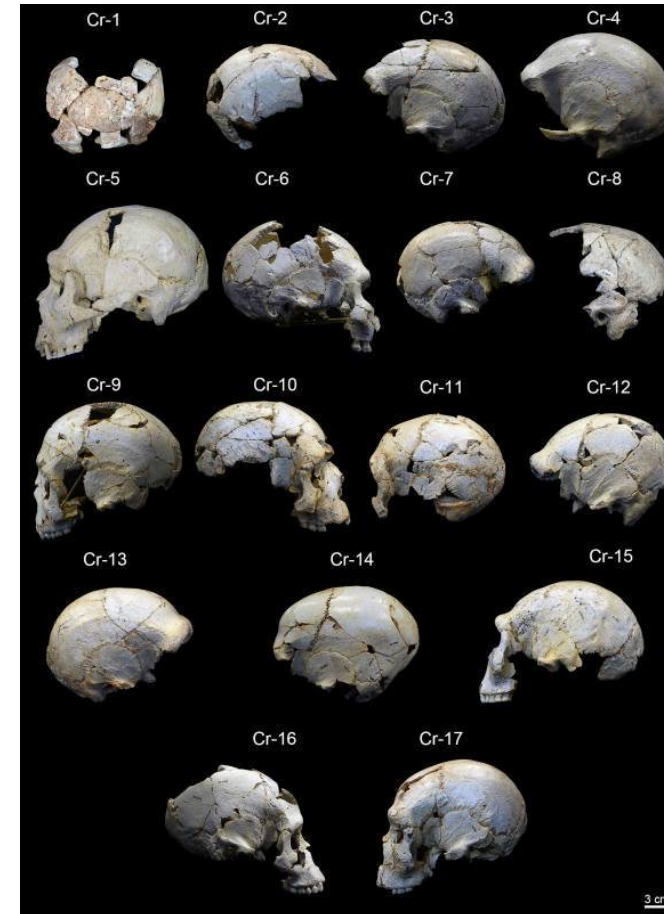
March 1st, 2022

Genetic history of an archaic hominin group from Denisova Cave in Siberia

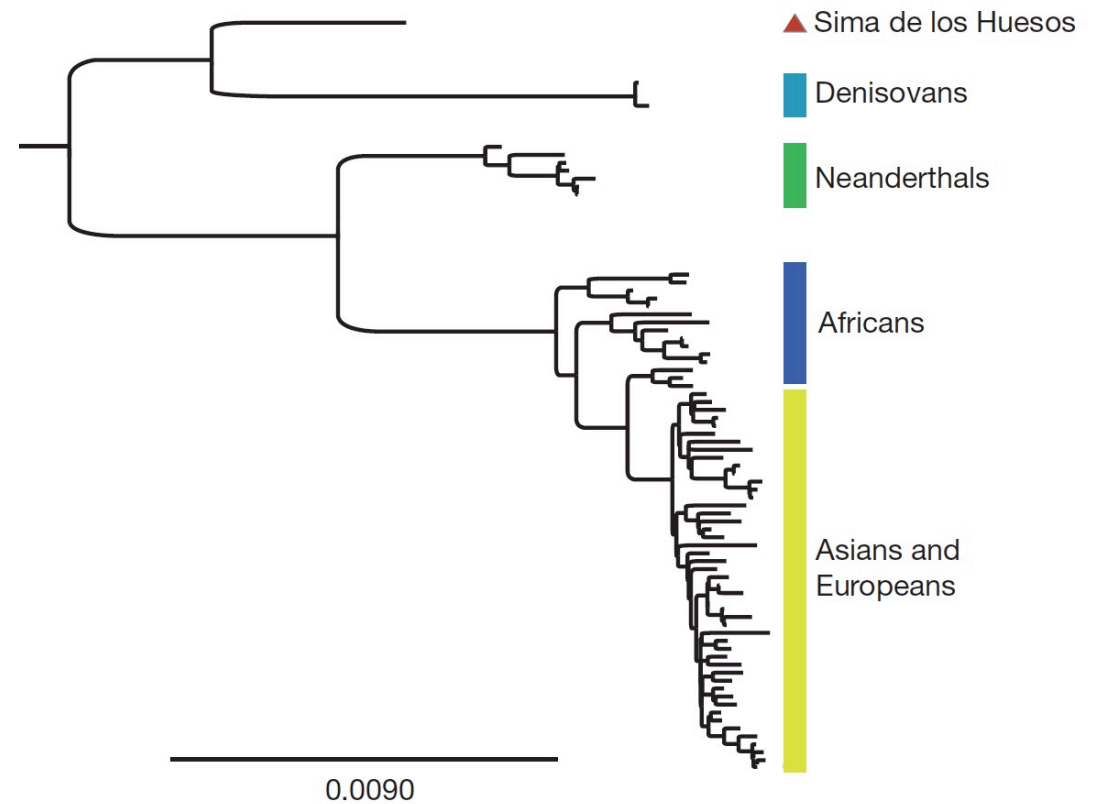
David Reich^{1,2*}, Richard E. Green^{3,4*}, Martin Kircher^{2*}, Johannes Krause^{2,5*}, Nick Patterson^{2*}, Eric V. Durand^{6*}, Bence Viola^{2,7*}, Adrian W. Briggs^{1,3}, Udo Stenzel³, Philip L. F. Johnson⁸, Tomislav Maricic³, Jeffrey M. Good⁹, Tomas Marques-Bonet^{10,11}, Can Alkan¹⁰, Qiaomei Hu^{2,12}, Swapan Mallick^{1,2}, Heng Li², Matthias Meyer³, Evan E. Eichler¹⁰, Mark Stoneking³, Michael Richards^{7,13}, Sahra Talamo⁷, Michael V. Shunkov^{1,4}, Anatoli P. Derevianko^{1,4}, Jean-Jacques Hublin⁷, Janet Kelso³, Montgomery Slatkin⁶ & Svante Pääbo³



Sima de los Huesos (Pit of Bones)

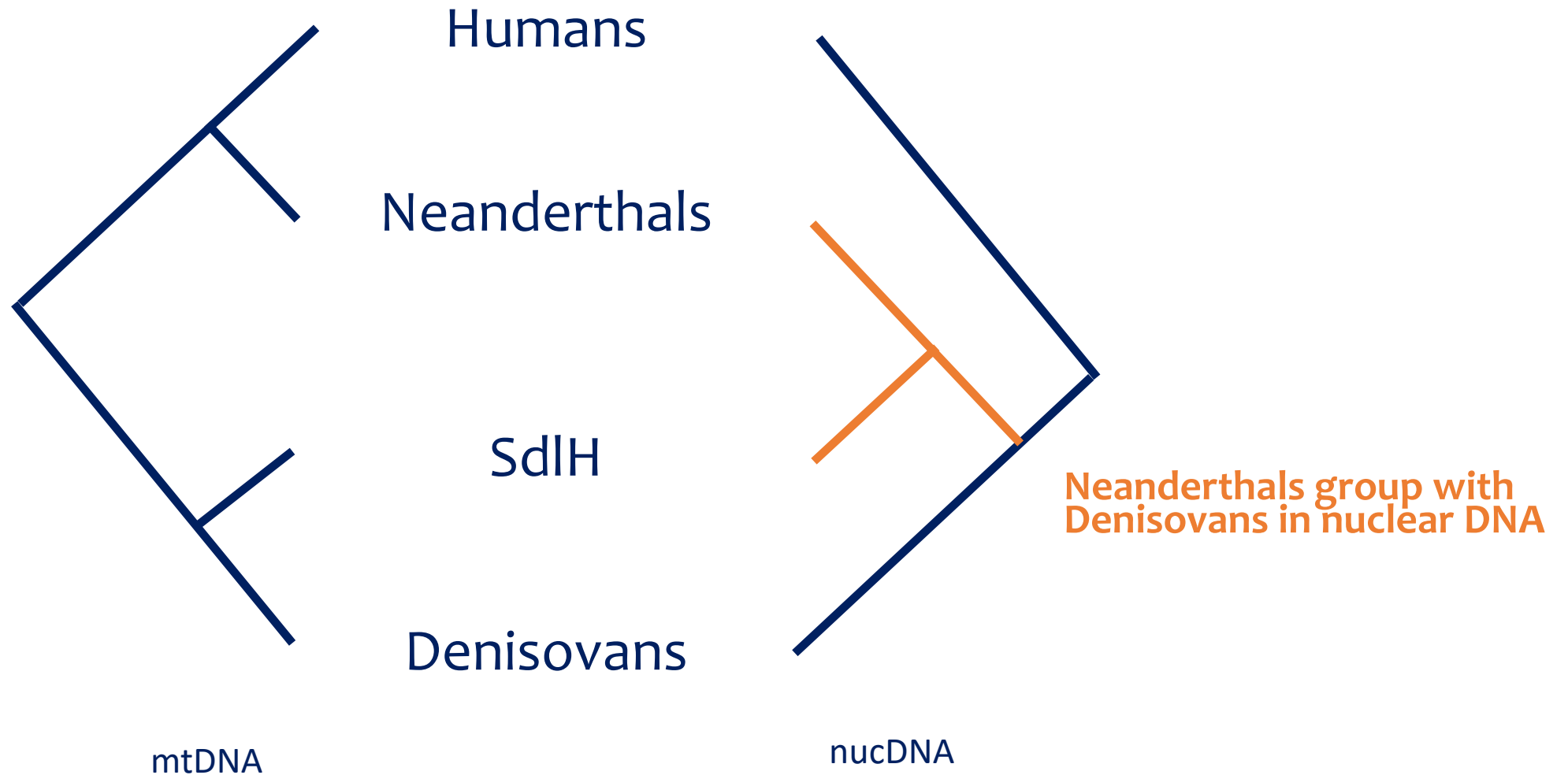


Sima de los Huesos (Pit of Bones)

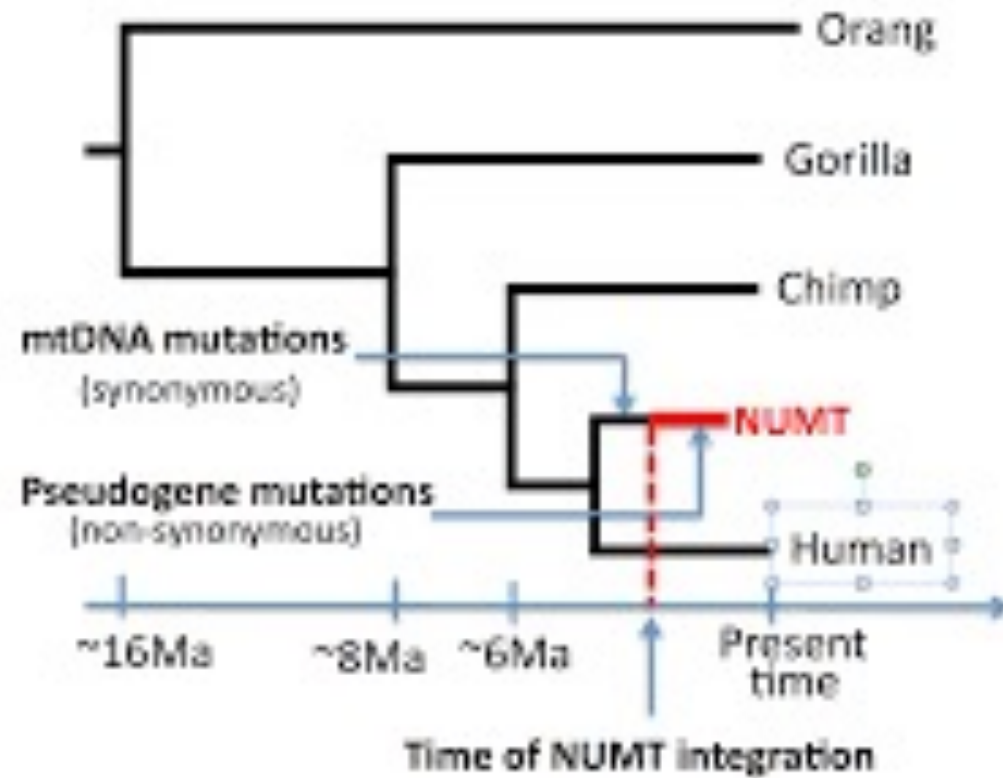
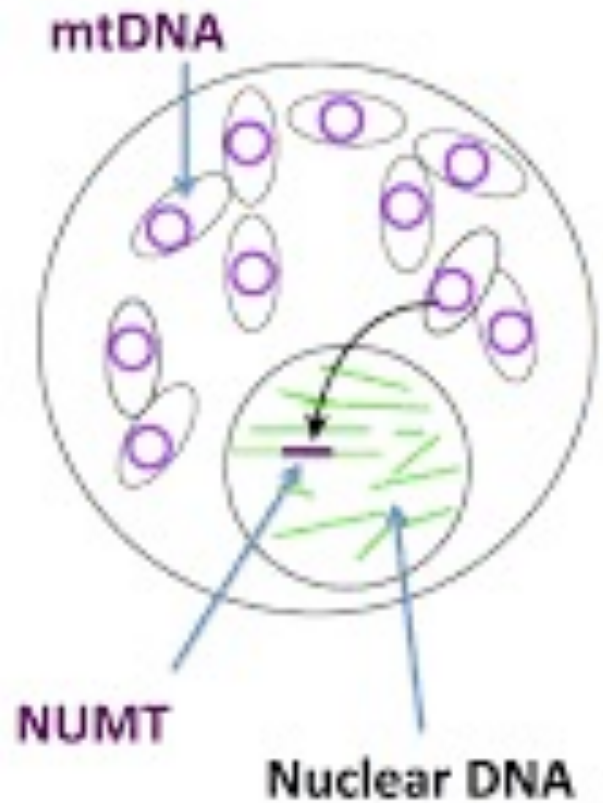


mtDNA

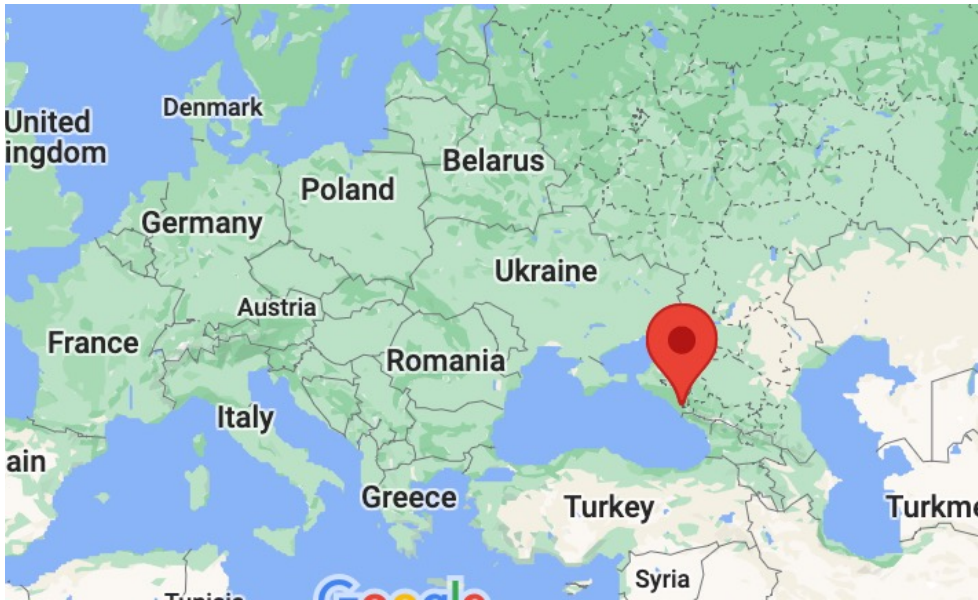
Mitochondrial replacement by ancient hominin?



Numts are “genomic fossils” recording the mtDNA across time



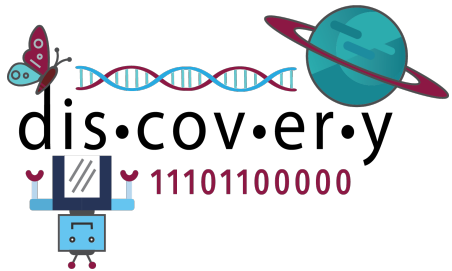
Numts are “genomic fossils” recording the mtDNA across time



Neanderthal Pos.	Mez-1 Neanderthal Allele	Mez-1 Denisovan Allele	No. Supporting Reads
3,478	A	G	66
8,463	T	C	16
9,525	T	C	7
9,711	T	C	13
11,618	T	C	15

mtDNA

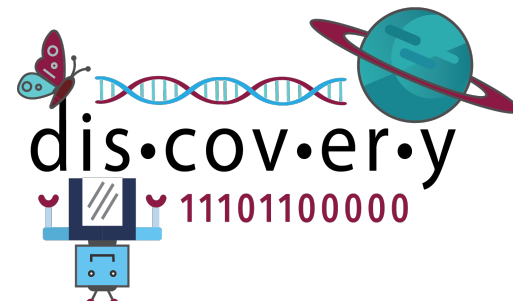
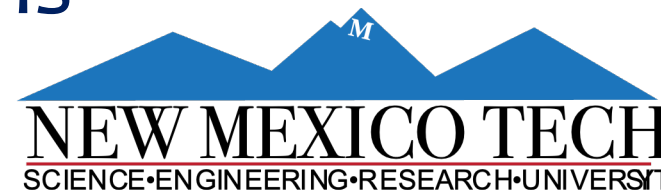
numt



Neanderthal **numts** resemble Denisovan mitochondrial DNA

Thus, ancient, human-like mitochondrial DNA invaded (**via hybridization**) and eventually **replaced** ancestral Neanderthal mitochondrial genomes

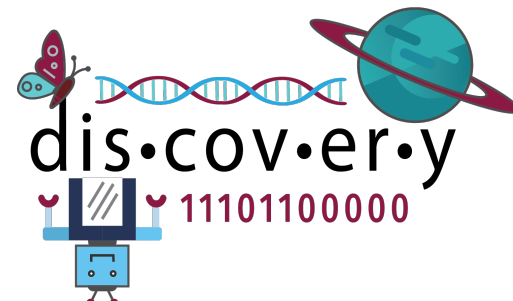
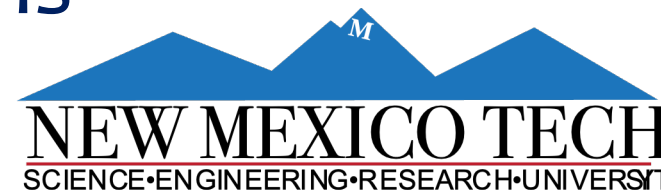
Implies a novel Out-of-Africa event,
prior to the evolution of humans



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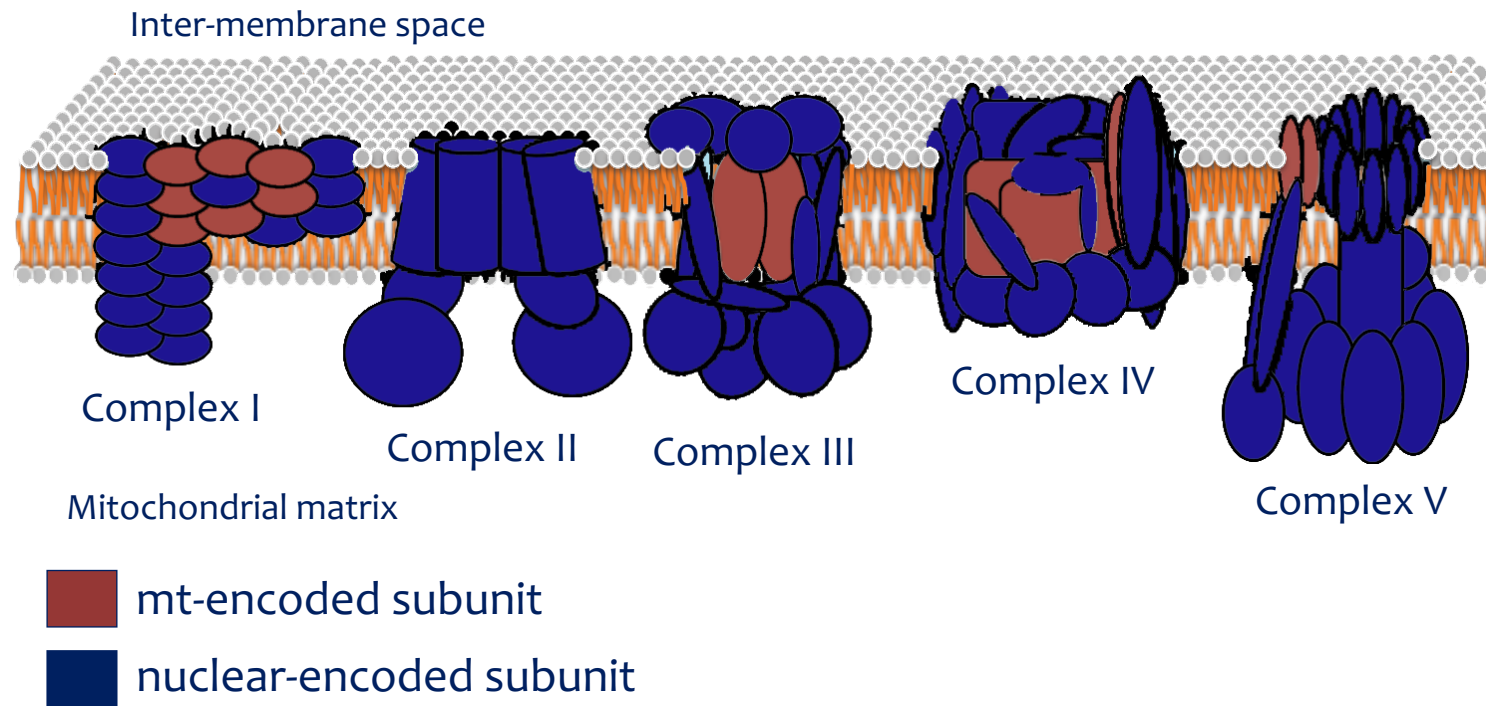
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Did nuclear-encoded mitochondria-targeted genes move along with them?

Oxidative Phosphorylation – “OXPHOS”



Rand et al 2004. *TREE*

