Report: Evolutionary Analysis of Human mtDNA and Related Lineages
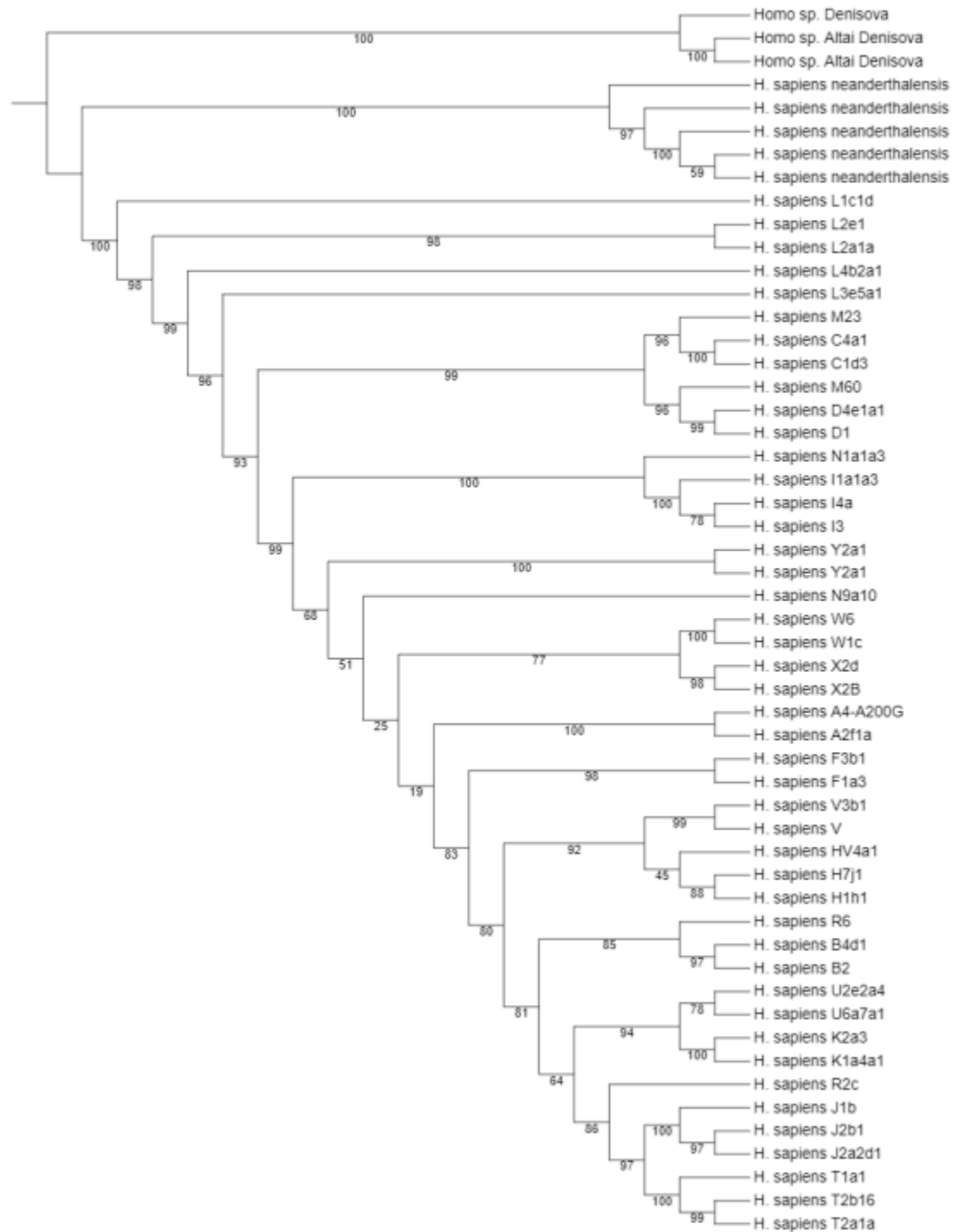
Steps Performed

The multiple sequence alignment: MAFFT algorithm.

A phylogenetic tree was constructed IQ-tree software and then refined using the iTOL platform.

The calculated the matrix of pairwise distances between all species: MEGA 11.

The mtDNA sequences of humans, Denisovans, and Neanderthals were combined into a single FASTA file.

1. **Mitochondrial Eve**

Homo sp. Denisova
Homo sp. Altai Denisova
Homo sp. Altai Denisova
H. sapiens neanderthalensis
H. sapiens neanderthalensis
H. sapiens neanderthalensis
H. sapiens neanderthalensis
H. sapiens neanderthalensis
H. sapiens L1c1d
H. sapiens L2e1
H. sapiens L2a1a
H. sapiens L4b2a1
H. sapiens L3e5a1
H. sapiens M23
H. sapiens C4a1
H. sapiens C1d3
H. sapiens M60
H. sapiens D4e1a1
H. sapiens D1
H. sapiens N1a1a3
H. sapiens I1a1a3
H. sapiens I4a
H. sapiens I3
H. sapiens Y2a1
H. sapiens Y2a1
H. sapiens N9a10
H. sapiens W6
H. sapiens W1c
H. sapiens X2d
H. sapiens X2B
H. sapiens A4-A200G
H. sapiens A2f1a
H. sapiens F3b1
H. sapiens F1a3
H. sapiens V3b1
H. sapiens V
H. sapiens HV4a1
H. sapiens H7j1
H. sapiens H1h1
H. sapiens R6
H. sapiens B4d1
H. sapiens B2
H. sapiens U2e2a4
H. sapiens U6a7a1
H. sapiens K2a3
H. sapiens K1a4a1
H. sapiens R2c
H. sapiens J1b
H. sapiens J2b1
H. sapiens J2a2d1
H. sapiens T1a1
H. sapiens T2b16
H. sapiens T2a1a

To estimate the age of a specific sequence, it is necessary to calculate the number of genetic variations between that sequence and others. Mitochondrial Eve (mtEve) is considered the common ancestor of all modern humans.

 Based on the literature, the L1 haplogroup is as the most ancient lineage. This haplogroup is also the first to branch from the root, which includes Denisovan sequences.

Using MEGA, the average number of nucleotide differences between the L1 haplogroup and other haplogroups was calculated to be 85,045. A human mtDNA mutation rate of 1.91 × 10⁻⁸ mutations per site per year (msy) was adopted based on recent studies. (https://www.nature.com/articles/s41598-021-84583-1)

Age of mtEve can be calculated as:

85.045/16569 /(1.91 × 10^{-8}) = 268731.8 years

This result is consistent with previous scientific estimates ranging from 200,000 to 300,000 years.

Separation time and branching order of the Neanderthal and Denisovan population:

Mean distance between Homo sapiens neanderthalensis and all modern
Homo sapiens sequences was amounted to 208.64
nucleotides. Hence the separation of Neanderthal population happened
208.64 / 16569 (1.91 × 10^{-8}) = 659277 years ago.
Mean distance between Denisovan population and modern humans after
calculations is equal to 379.98. Using the formula we obtain:
208.64 / 16569 (1.91 × 10^{-8}) = 1200690.5 years