

Genetic comparison of Human and Neanderthal mtDNA

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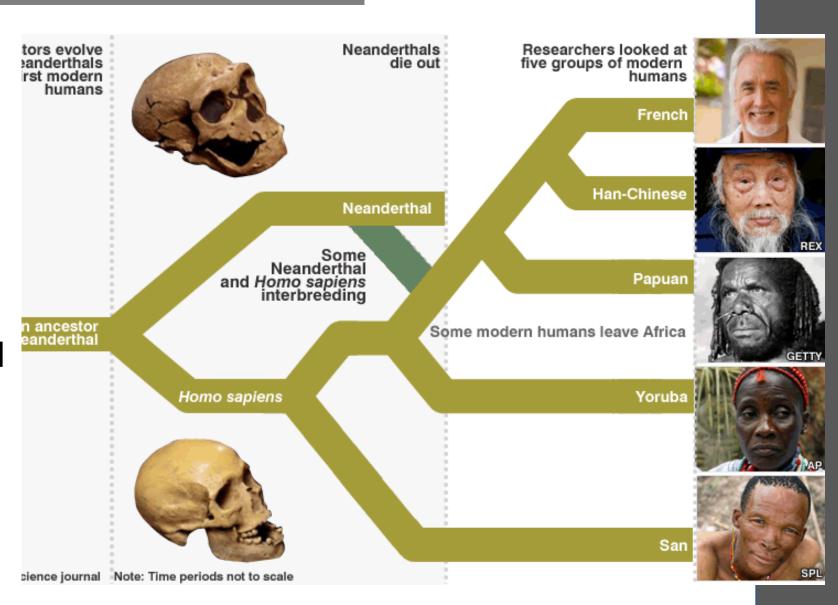
## Neanderthals

- Lived side by side with *H. sapiens* (that's us)
- Latest common ancestor: Homo erectus
- Evolved in Eurasia about 600.000 years ago
- Extinction: 30.000 years ago
- Related to Humans?



# Neanderthals and Humans

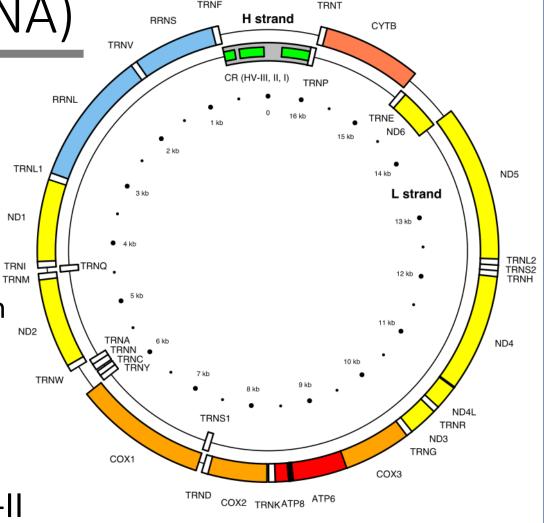
- About 202 differences in mtDNA compared to humans
- Geneflow from Neanderthals to Humans
- Gene mixing estimated at 1.15% to 1.3% in autosomes



#### Genetic Analysis

Mitochondrial DNA (mtDNA)

- 16,569 base pairs form 37 genes
- Maternally inherited
  - → No recombination
  - → mutations only through evolution
- Coding Region and Control Region
- Region of interest
  - → Control Region HV-I and HV-II



# Genealogical DNA-Testing

Alignment:

Compare Sequences and find Nucleotide Substitutions

Compute
Phylogenetic
Tree (UPGMA)







Use differences to create Distance-Matrix (Jukes-Cantor)

Deduce relatedness and geneflow

#### Experimental procedure

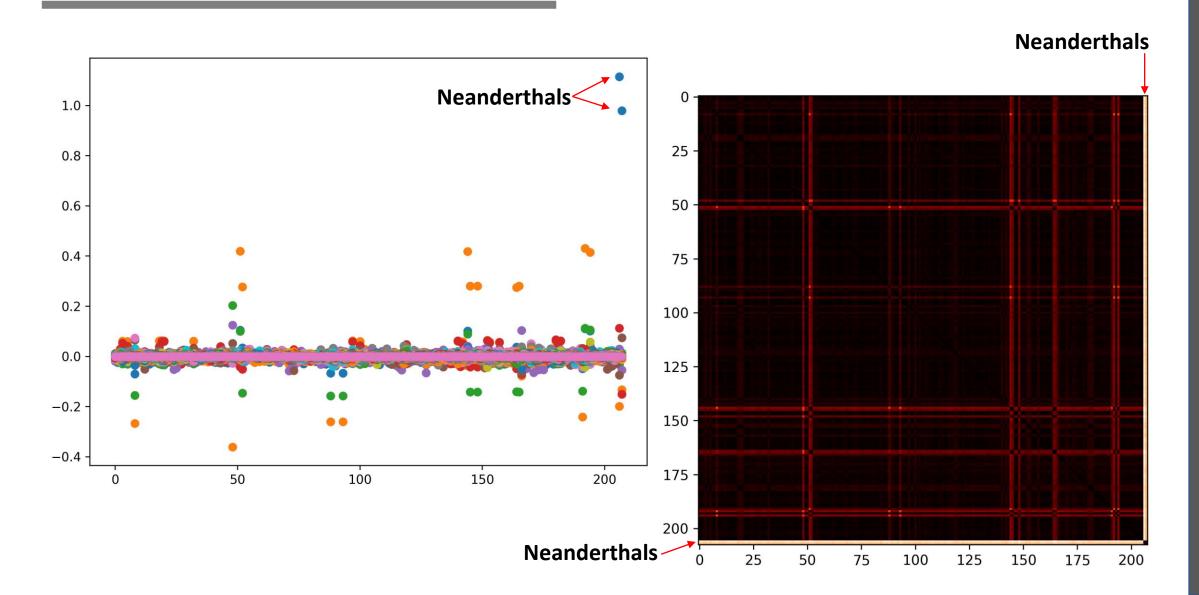
## Genetic Distances

Calculated mean distances...

- among any two H. sapiens: **0.0890**
- between Neanderthal and any modern human: 1.0739

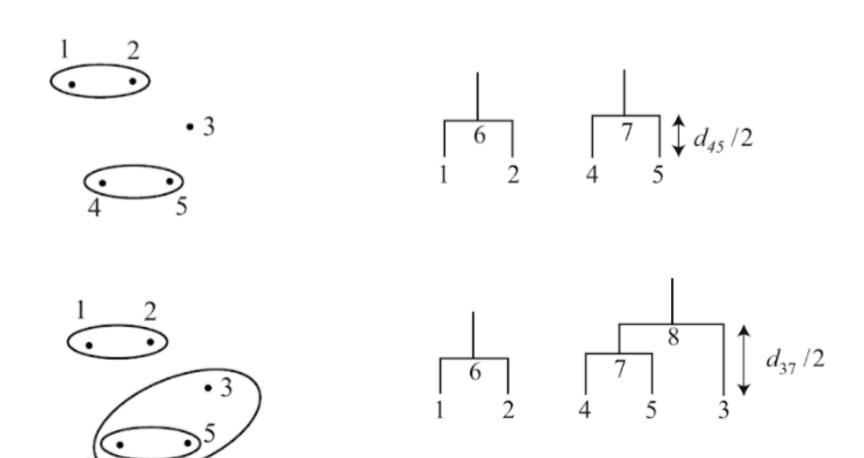
```
[[0. 0.02601417 0.06961516 ... 0.04374418 1.15120154 0.99861976]
[0.02601417 0. 0.07266085 ... 0.03478884 1.15120154 1.01045433]
[0.06961516 0.07266085 0. 0.07266085 1.12598908 0.99861976]
...
[0.04374418 0.03478884 0.07266085 ... 0. 1.12598908 0.99861976]
[1.15120154 1.15120154 1.12598908 ... 1.12598908 0. 0.13260893]
[0.99861976 1.01045433 0.99861976 ... 0.99861976 0.13260893 0. ]]
```

# Genetic Distances

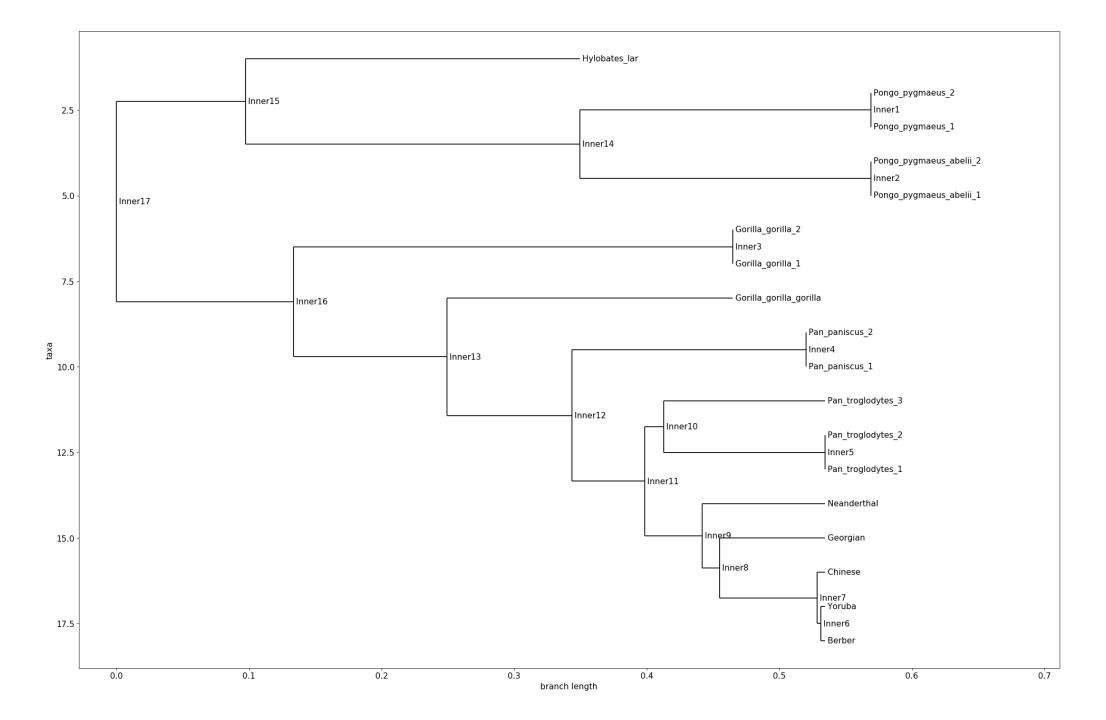


#### Experimental procedure

# **UPGMA**







### Conclusion

- Neanderthal closely related to modern humans, when compared to other primates
- The distance between Neanderthals and Humans show that they were two different species
- Our results don't show weather there was a geneflow between the two species
- The split-off of Georgians from the other human groups, indicates that there might be inaccurate measurements. This could potentially be fixed by using different distance matrices.

### References

- Sriram Sankararaman et al.: The genomic landscape of Neanderthal ancestry in present-day humans. In: Nature. Band 507, 2014, S. 354–357, doi:10.1038/nature12961
- https://www.ncbi.nlm.nih.gov/pubmed/20448178
- <a href="https://www.eurekalert.org/pub">https://www.eurekalert.org/pub</a> releases/2016-02/m-egf021616.php
- Richard E. Green et al.: A Complete Neandertal Mitochondrial Genome Sequence Determined by High-Throughput Sequencing
- https://www.theverge.com/2017/10/9/16448412/neanderthal-stone-age-human-genes-dna-schizophrenia-cholesterol-hair-skin-loneliness

#### Picture Courtesies

- title picture (skulls): hairymuseummat <a href="https://www.theverge.com/2017/10/9/16448412/neanderthal-stone-age-human-genes-dna-schizophrenia-cholesterol-hair-skin-loneliness">https://www.theverge.com/2017/10/9/16448412/neanderthal-stone-age-human-genes-dna-schizophrenia-cholesterol-hair-skin-loneliness</a>
- Neanderthal skeleton:
   <u>Claire Houck</u> from New York City, USA <u>Neanderthal Skeleton</u>
- geneflow picture: <u>http://news.bbc.co.uk/nol/shared/spl/hi/sci\_nat/10/neanderthal/img/neanderthals\_786.gif</u>
- mtDNA Picture: <u>Emmanuel Douzery</u>