

ANCIENT DNA



AARHUS
UNIVERSITY
DEPARTMENT OF MOLECULAR BIOLOGY AND GENETICS

EVOLUTIONARY THINKING 2023
WEEK 46

CALIN PANTEA
PHD STUDENT

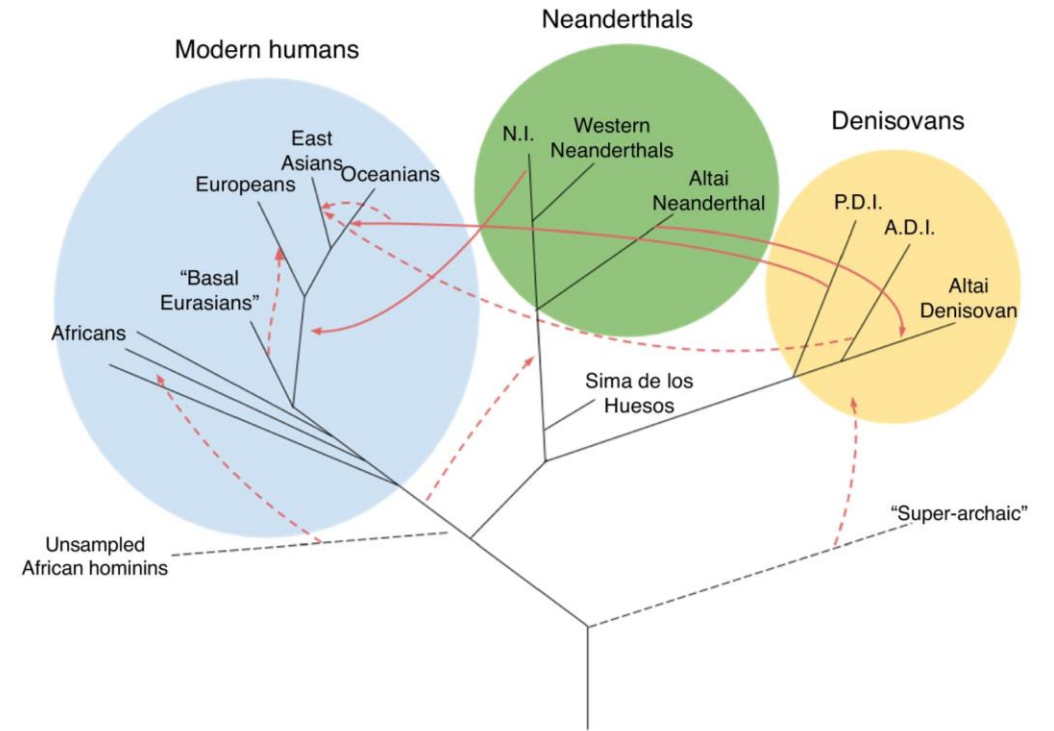


OVERVIEW

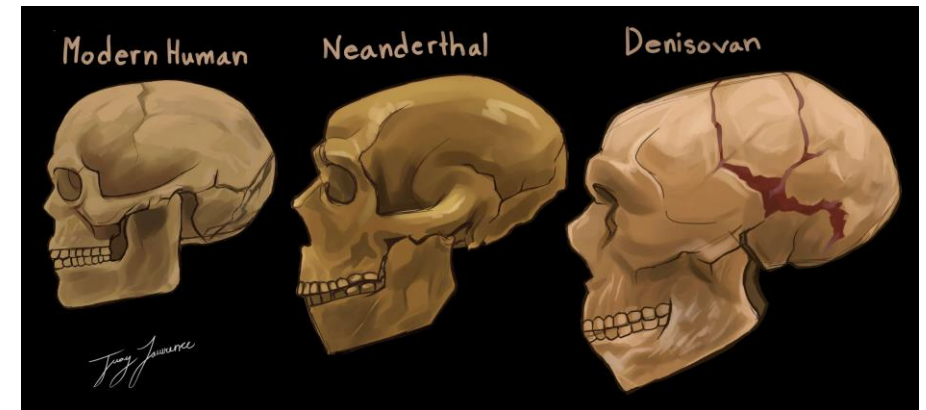
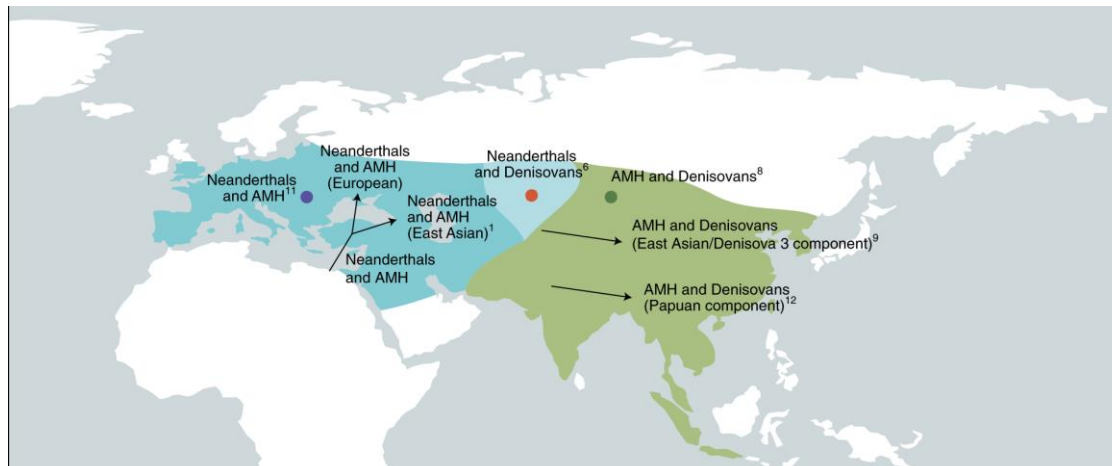
- some interesting **developments** in the field (instead of the lecture recap)
(12:15 – 12:30-ish)
- paper questions + discussion – **genome-wide** archaic introgression
(12:30 – 13:00 and 13:15 – 13:45)
- Menti quiz
(last 10 minutes or so)

ANCIENT DNA

- Current state of affairs
 - multiple introgression events
 - at least two species, likely 3+
- well-characterised archaeological evidence for two: Neanderthals and Denisovans



<https://doi.org/10.1016/j.gde.2018.05.009>

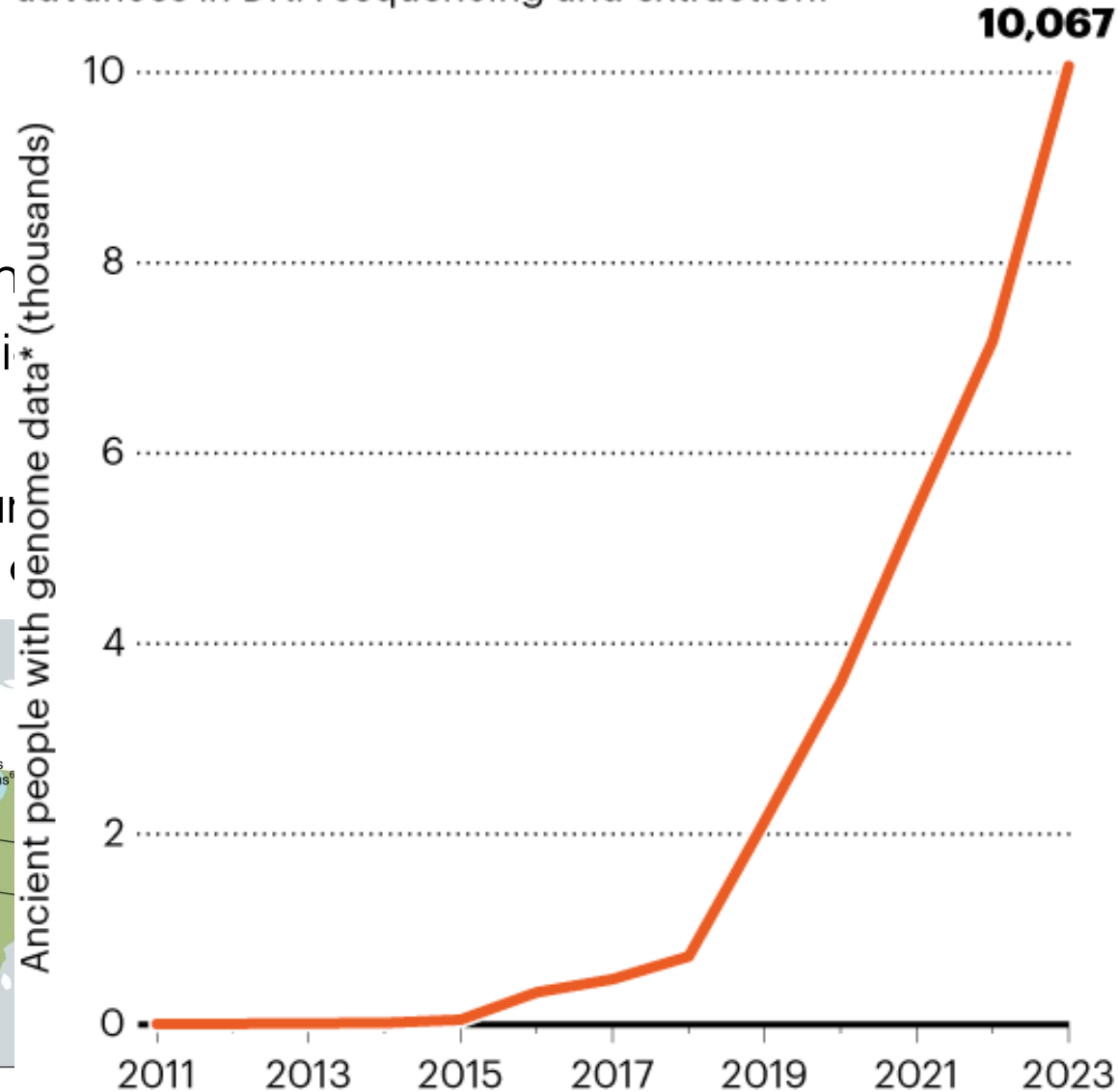
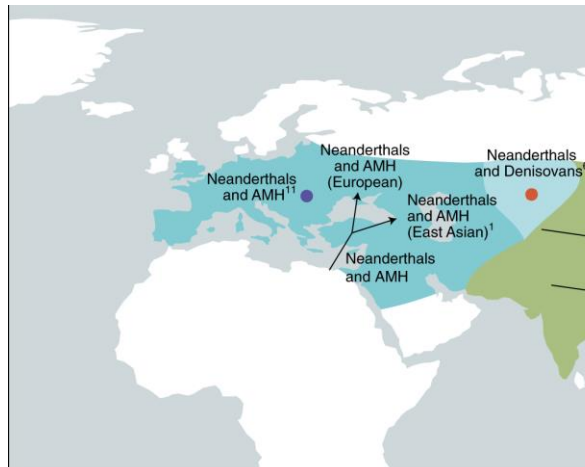


ANCIENT DNA

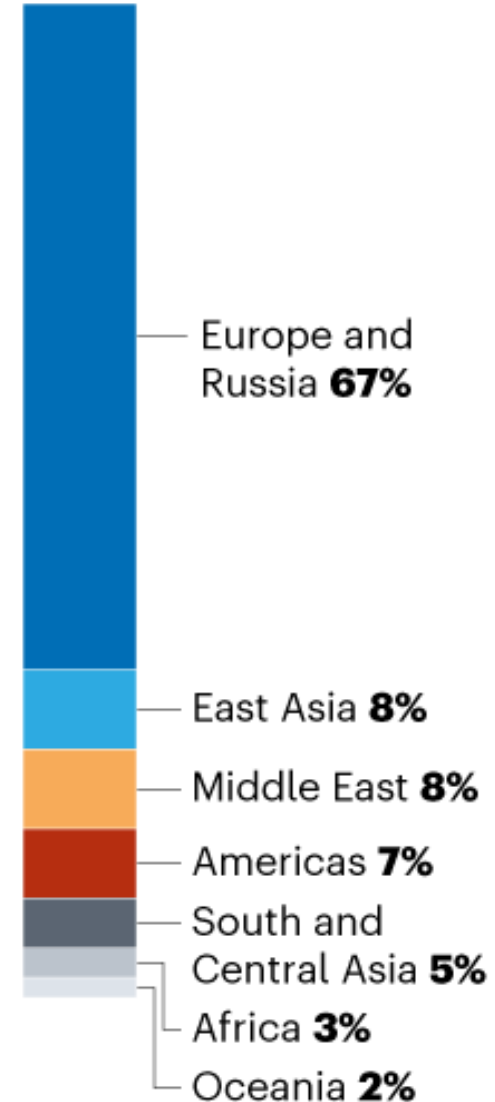
ANCIENT-DNA GOLDRUSH

The quantity of genome data from ancient-human remains has grown rapidly since 2018, owing to advances in DNA sequencing and extraction.

- Current state of affairs
 - multiple introgression
 - at least two species
 - well-characterised and for two: Neanderthals and AMH

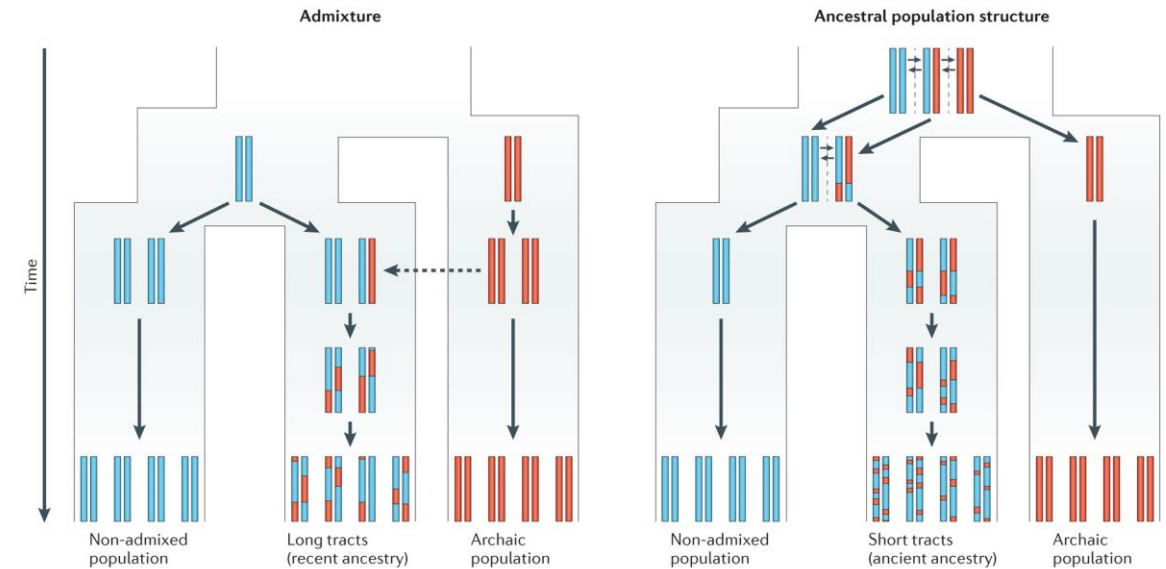


Geographical distribution



ANCIENT DNA

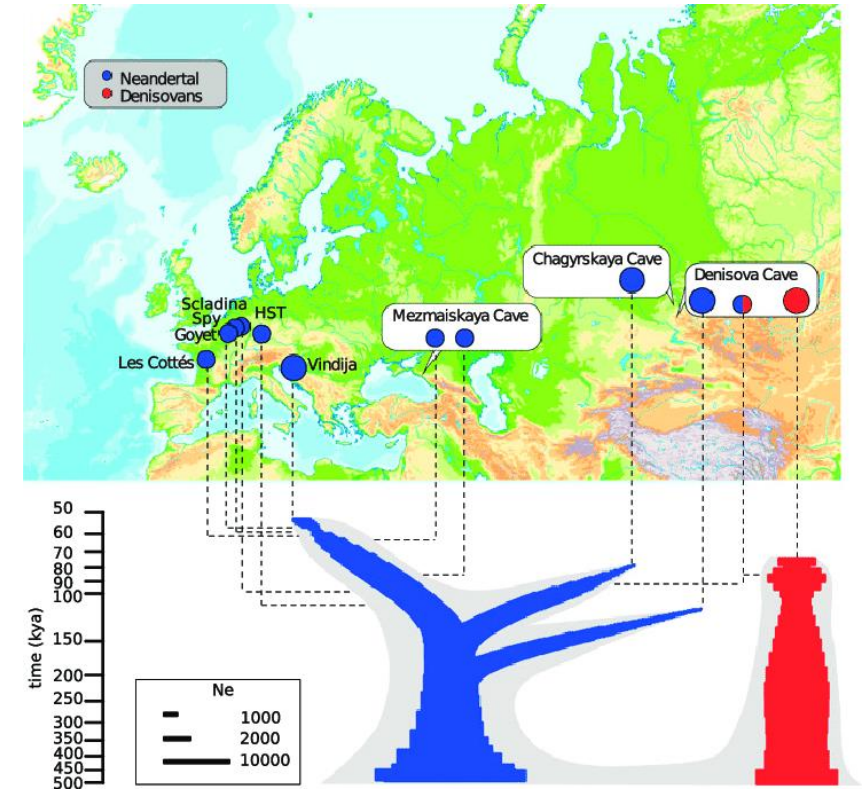
- tract length to infer introgression
- they argue that adaptive introgression should be seen as a separate phenomenon from selection on ancestral haplotypes
- reality – most likely something in-between



<https://doi.org/10.1038/nrg3936>

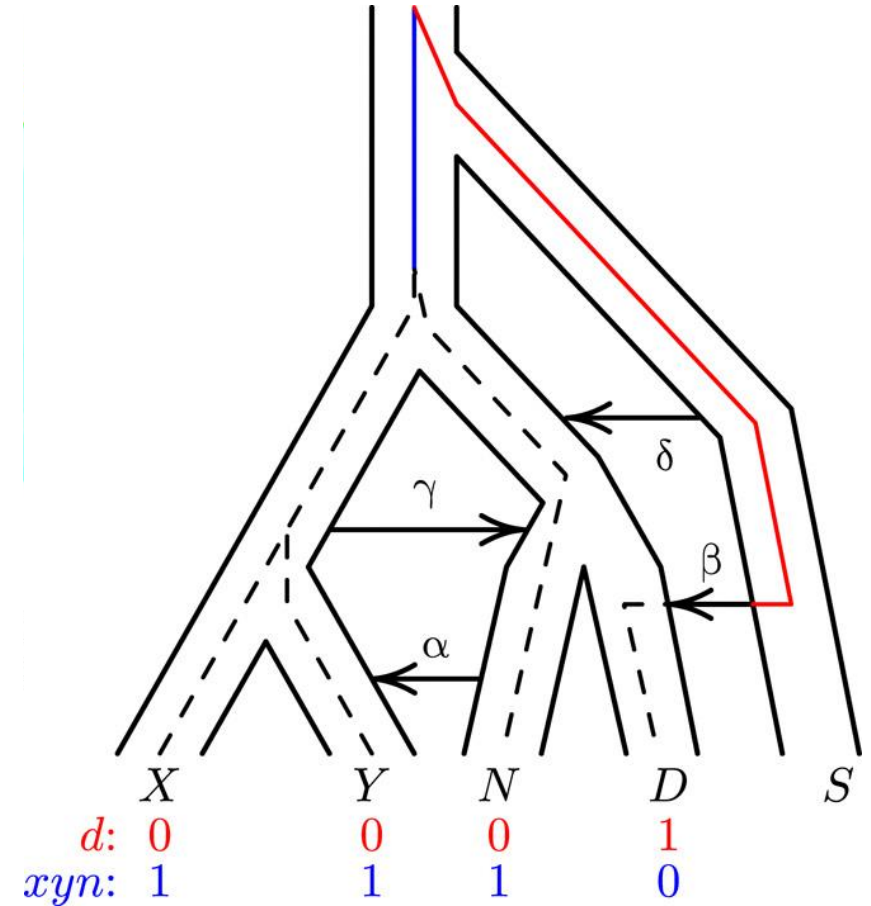
ANCIENT DNA

- Neandertal and Denisovan divergence
 - first attempts – mtDNA
 - as archaic genome resolution became better – variation in nuclear genome
 - complicated by a potential complex phylogeny with deep divergence and plenty of admixture



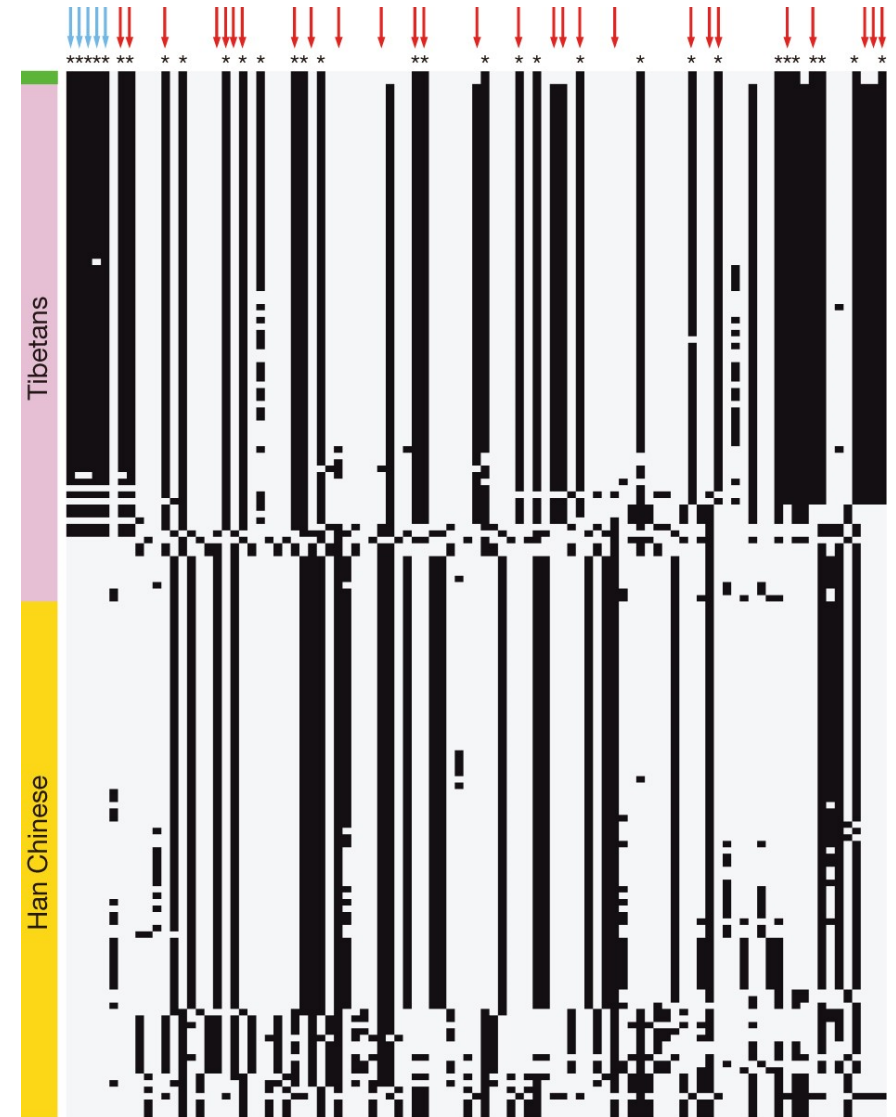
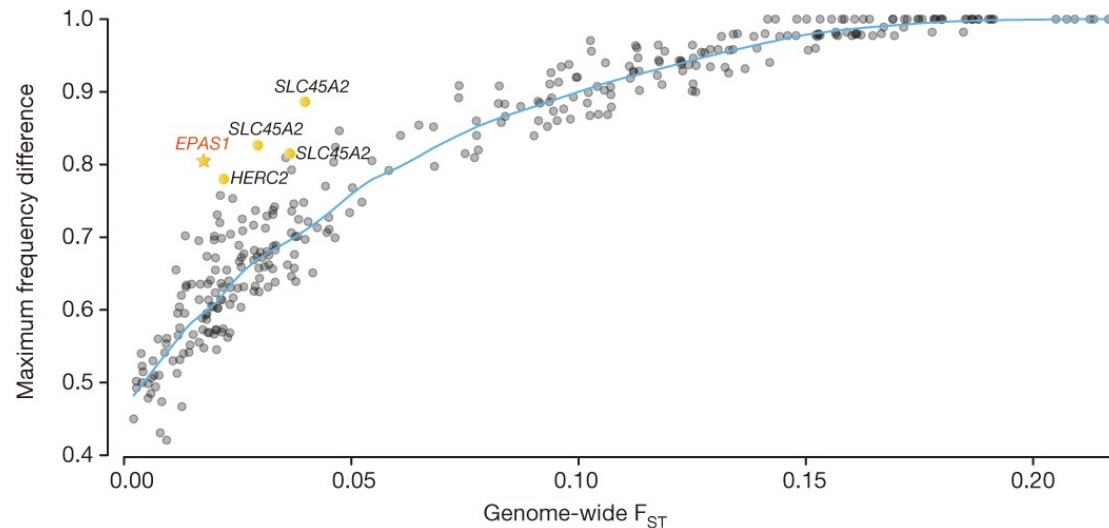
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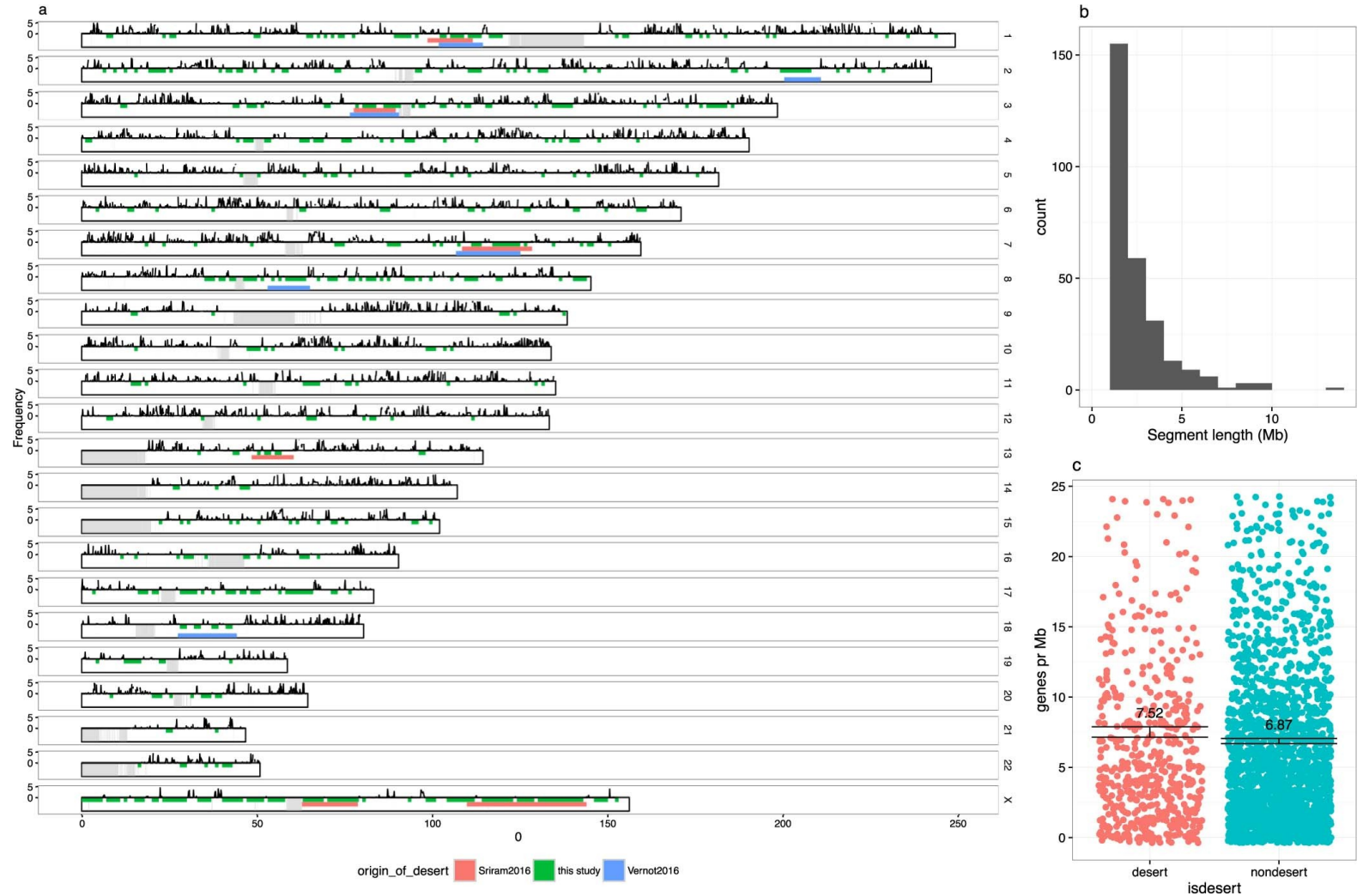
ANCIENT DNA

- Denisovan introgression in Tibetans contributed to altitude adaptation
 - Tibetan vs Han Chinese – closely related; F_{ST} differences reflecting selective pressure with higher confidence
 - Denisovan haplotypes present at high freq in Tibetans



ANCIENT DNA

- introgression does not always go as planned
- archaic introgression deserts (regions with no introgression)
- extensive on the X chromosome
- any guess why?



<https://doi.org/10.1038/s41586-020-2225-9>

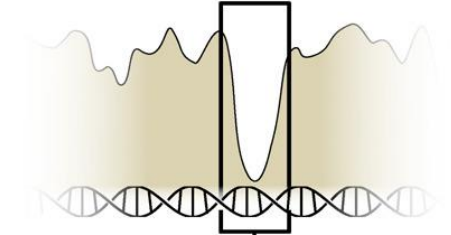
ANCIENT DNA

- non-African modern humans show long common haplotypes subject to strong selective sweeps
- post-introgression deserts
- swept haplotypes even present in an ancient Siberian modern human
- position of these human sweeps overlap with previously described sweep hotspots across great apes

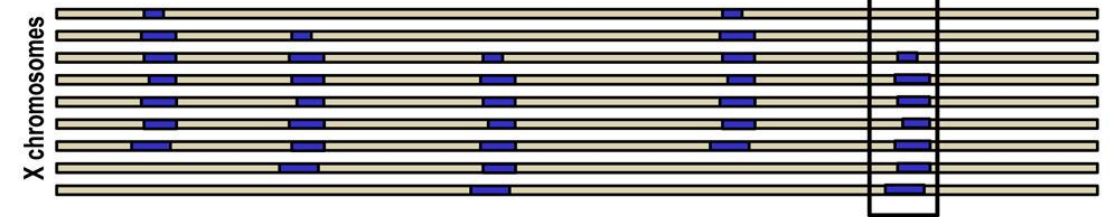
Global X chromosome sampling from SGDP



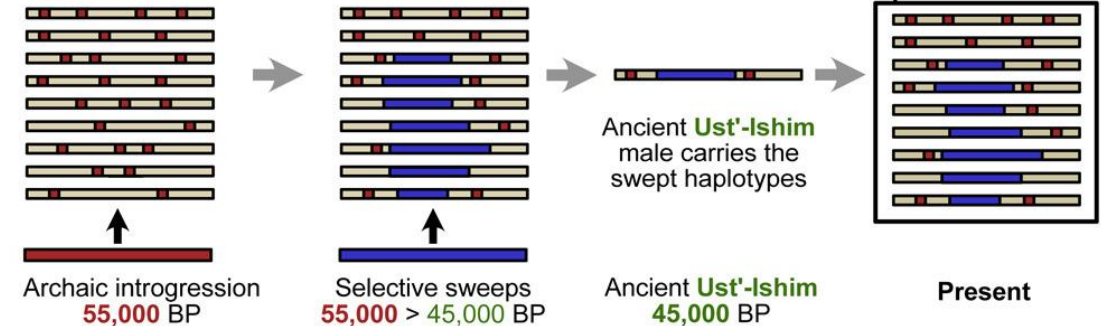
Strong reductions in non-African X chromosome diversity:



Strong diversity reduction are caused by long common haplotypes:



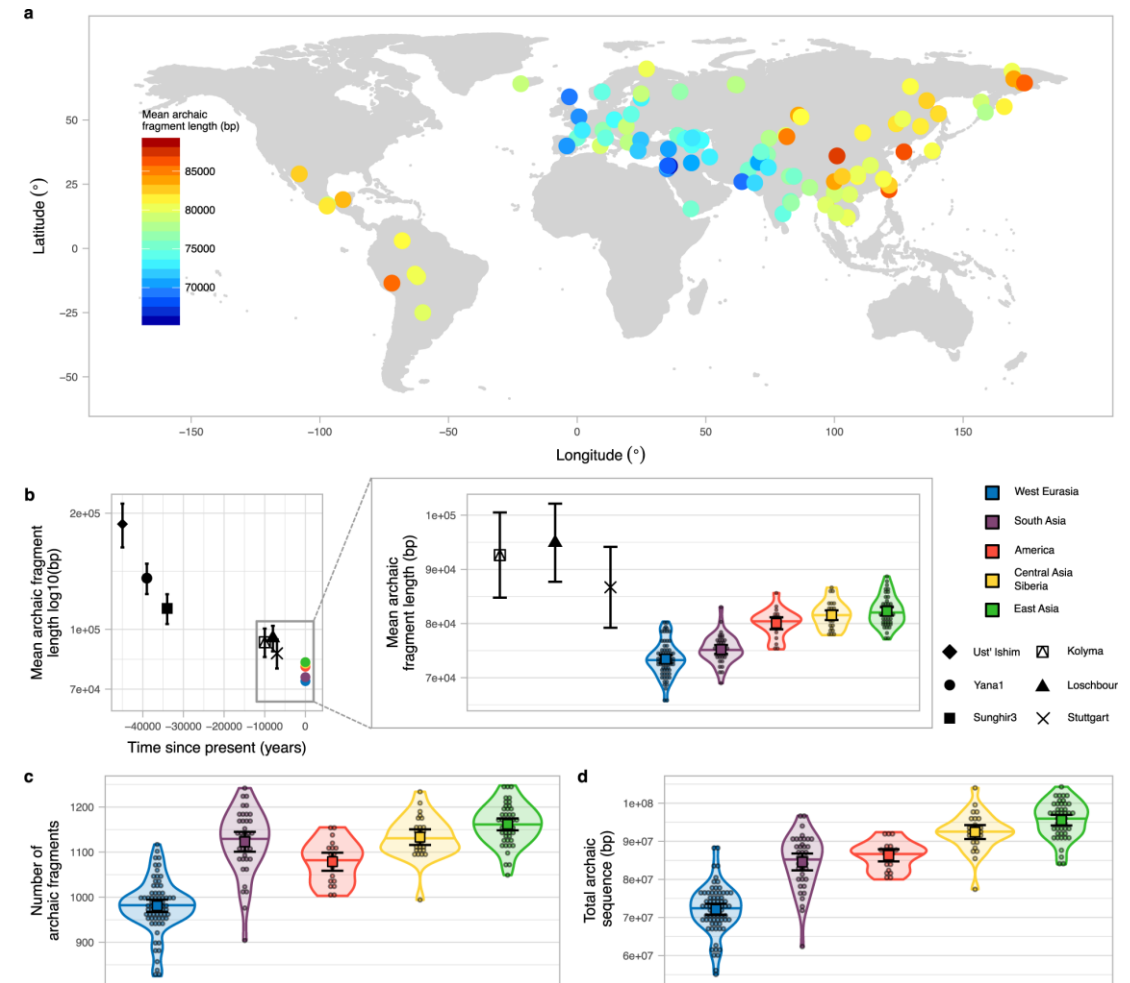
Selective sweeps displaced admixture 55,000 - 45,000 years BP:



<https://doi.org/10.1016/j.xgen.2023.100274>

ANCIENT DNA

- archaic fragment length varies between modern populations
- so do total numbers of archaic fragments per genome
- West Eurasian fragments subject to more recombination and increased mutation rates
- why?



<https://doi.org/10.1038/s41467-021-25524-4>

All this because we have been able to sequence more archaic genomes

But everything began here:

A Draft Sequence of the Neandertal Genome

RICHARD E. GREEN, JOHANNES KRAUSE, ADRIAN W. BRIGGS, TOMISLAV MARICIC, UDO STENZEL, MARTIN KIRCHER, NICK PATTERSON, HENG LI, WEIWEI ZHAI, [...], AND

SVANTE PÄÄBO

+46 authors

[Authors Info & Affiliations](#)

PAPER DISCUSSION

A Draft Sequence of the Neandertal Genome

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BREAK UNTIL 13:15



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