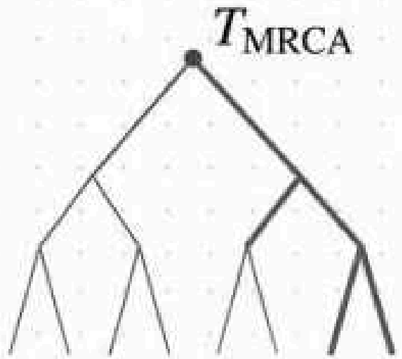
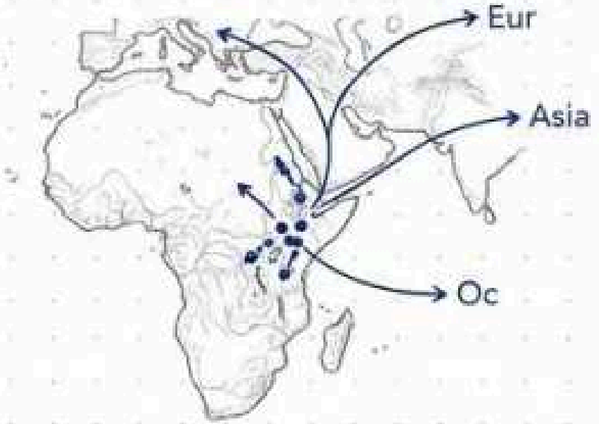
Populations, Ancestry, and Ancient DNA

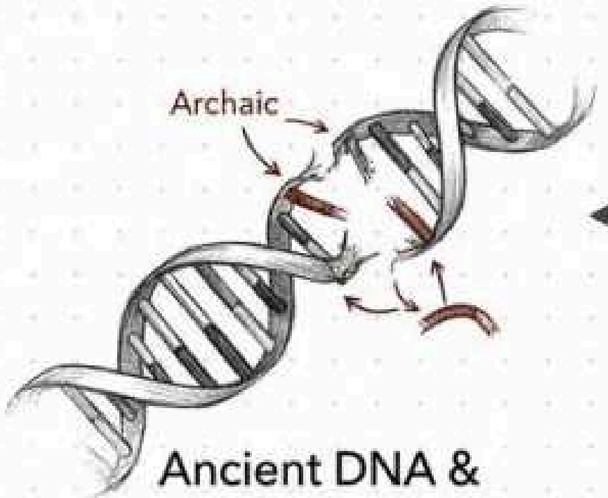
Reading human history from the genome



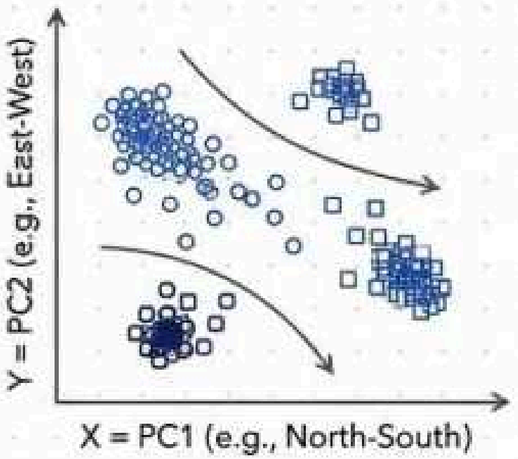
The Coalescent & N_e



Out of Africa



Ancient DNA & Archaic Admixture

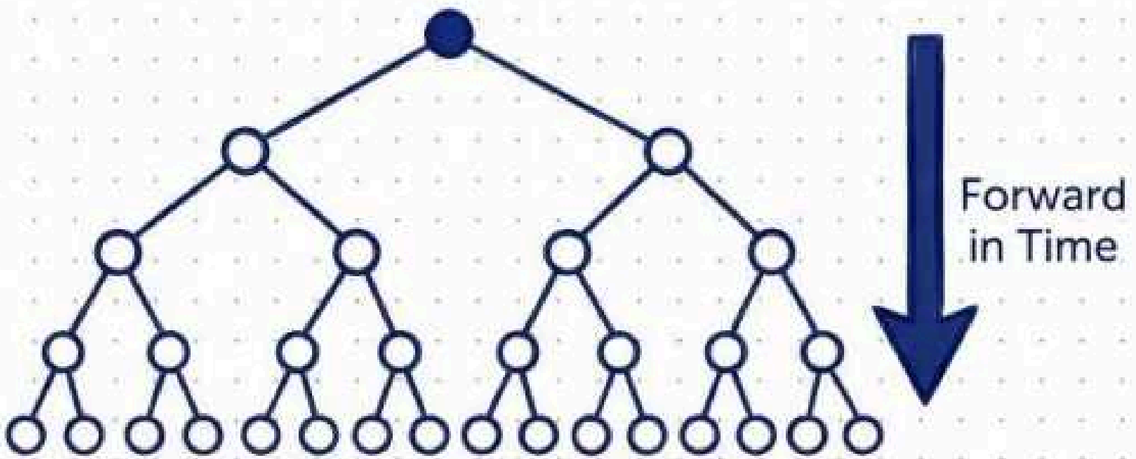


Genomic Geography

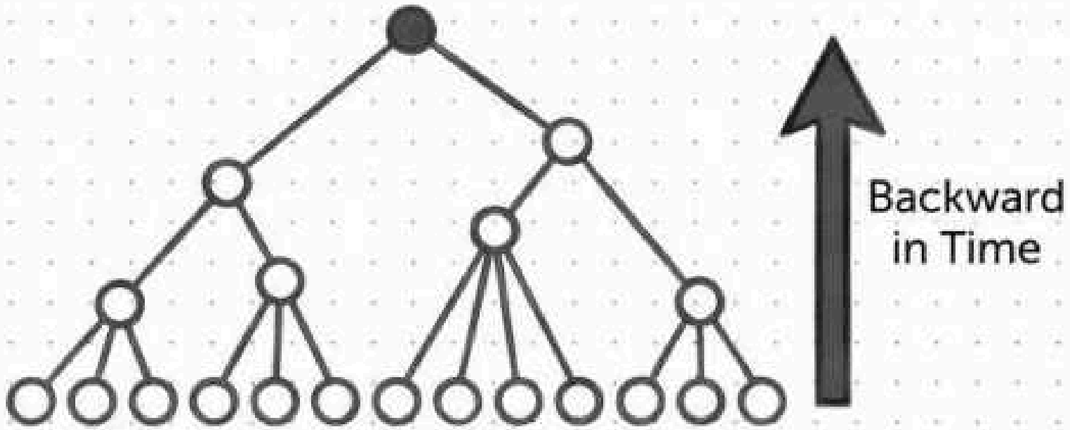
A shift from species to populations.
How living variation, historical bottlenecks, and ancient bones transform our view of the human past.

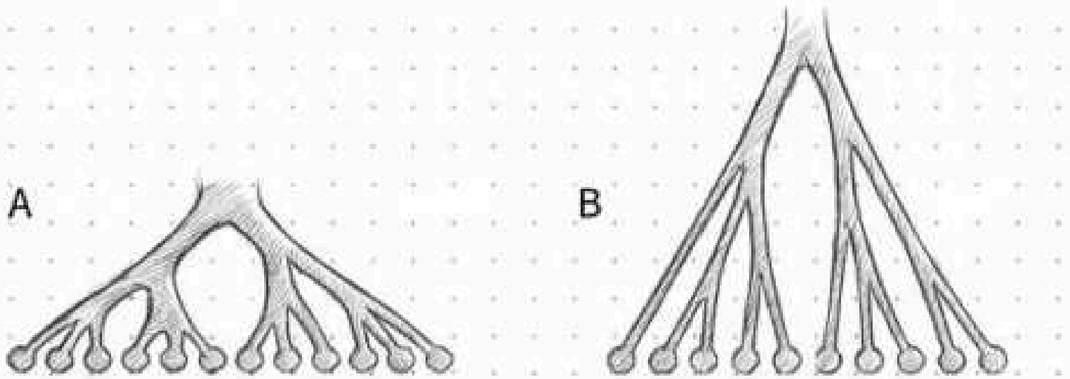
The Coalescent: Tracing Lineages Backward in Time

Standard Genealogy



The Coalescent (Kingman, 1982)



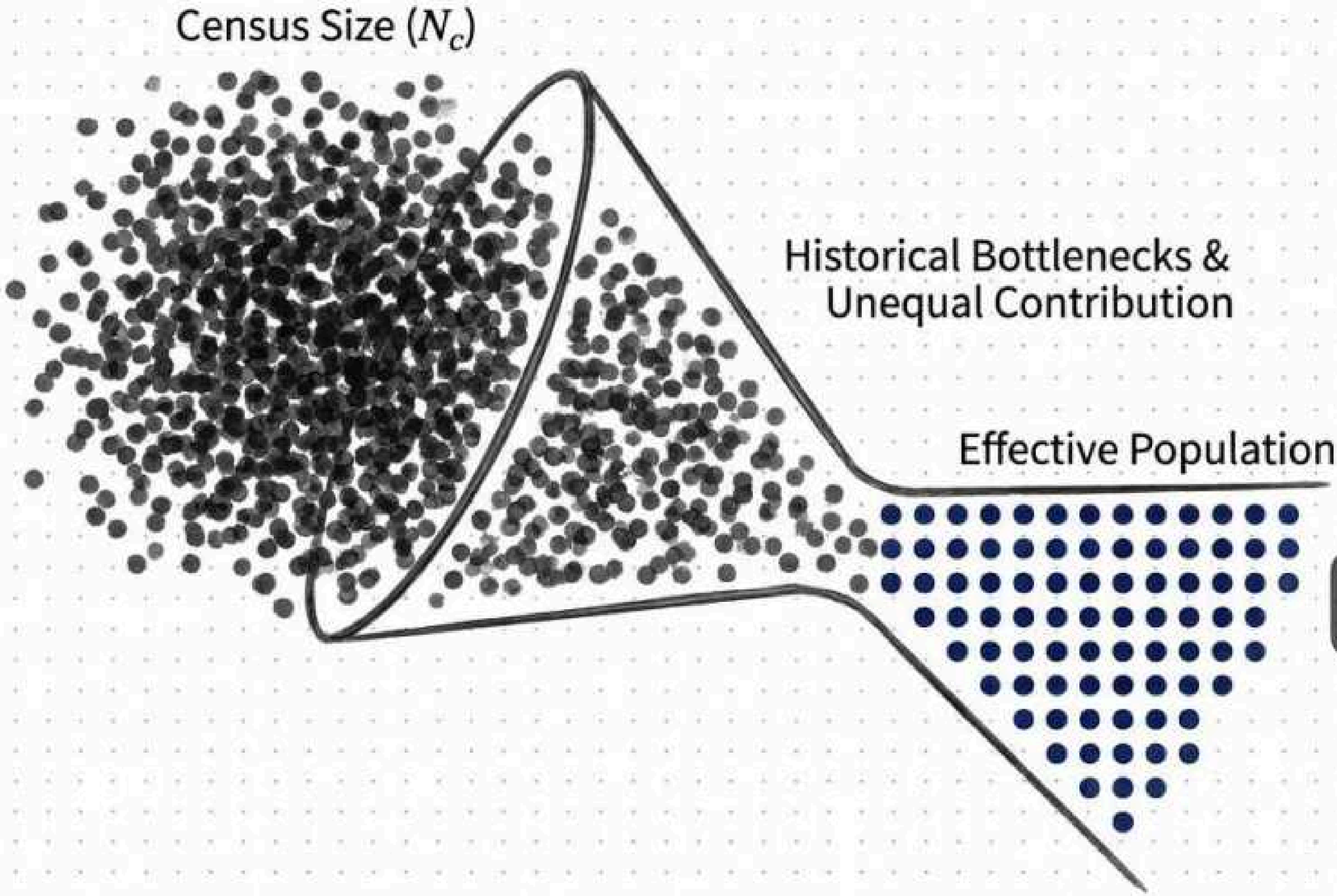


Small Population = Fast Coalescence

Large Population = Slow Coalescence

The shape of a gene tree directly records the history of population size.

Census Size (N_c) vs. Effective Population Size (N_e)



$$N_e \approx 10{,}000 - 20{,}000$$

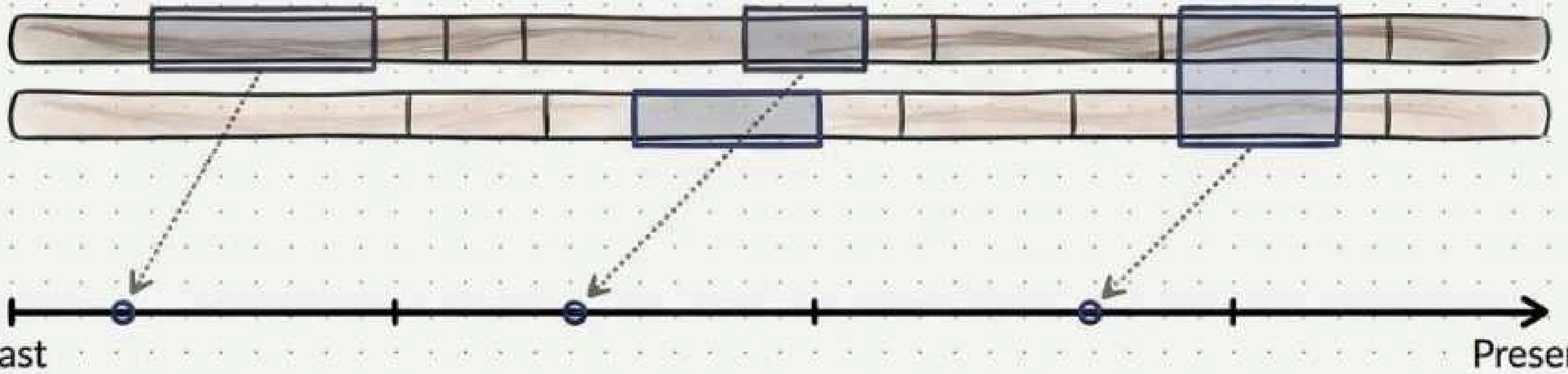
The size of an idealized population exhibiting the same amount of genetic drift.

Reading Time from a Single Genome (PSMC)

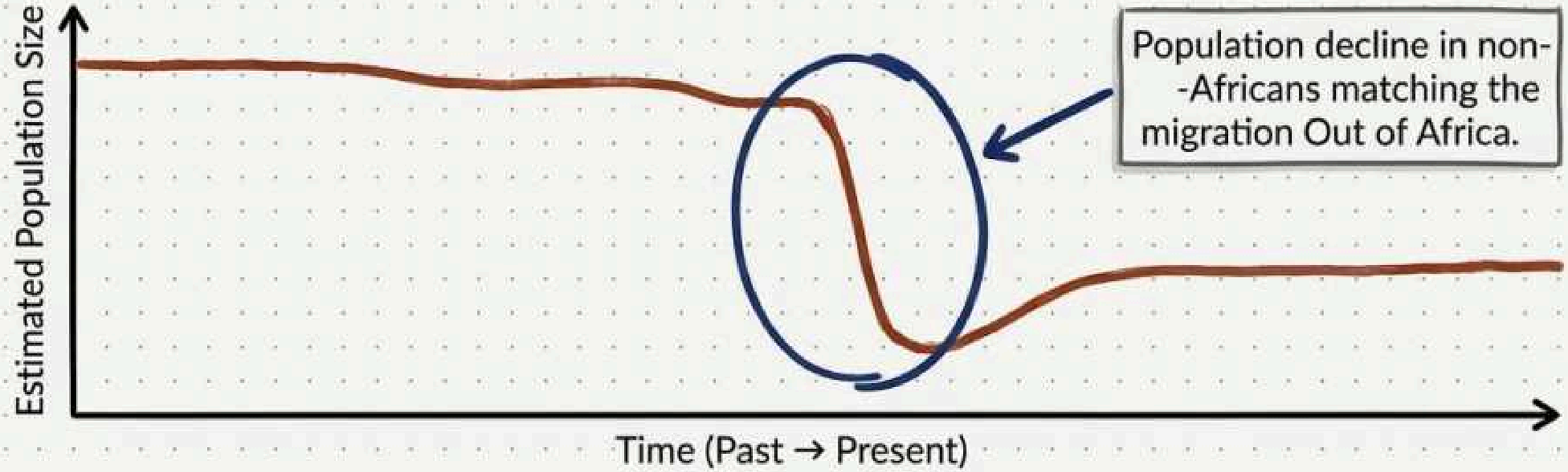
¶ 5.4

Pairwise Sequentially Markovian Coalescent (Li & Durbin, 2011)

Step 1

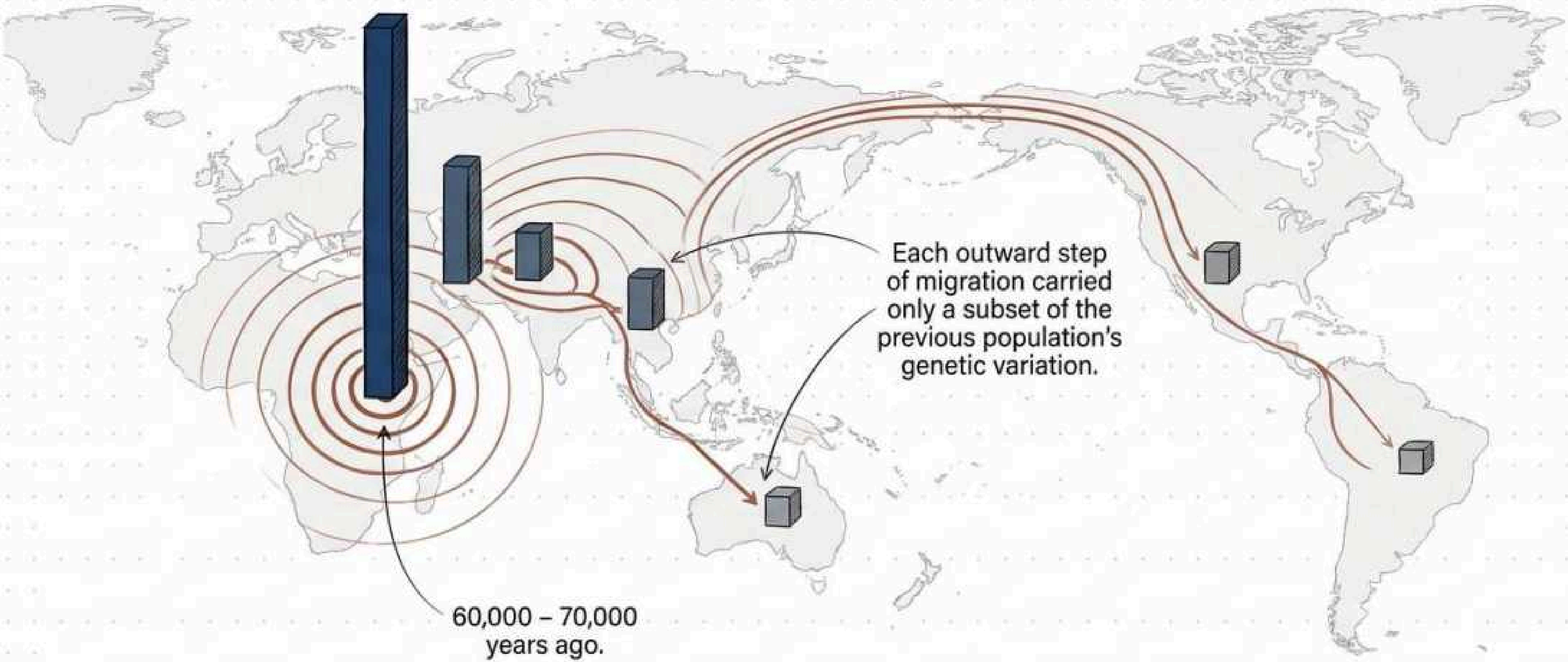


Step 2



The Serial Founder Effect and Out of Africa

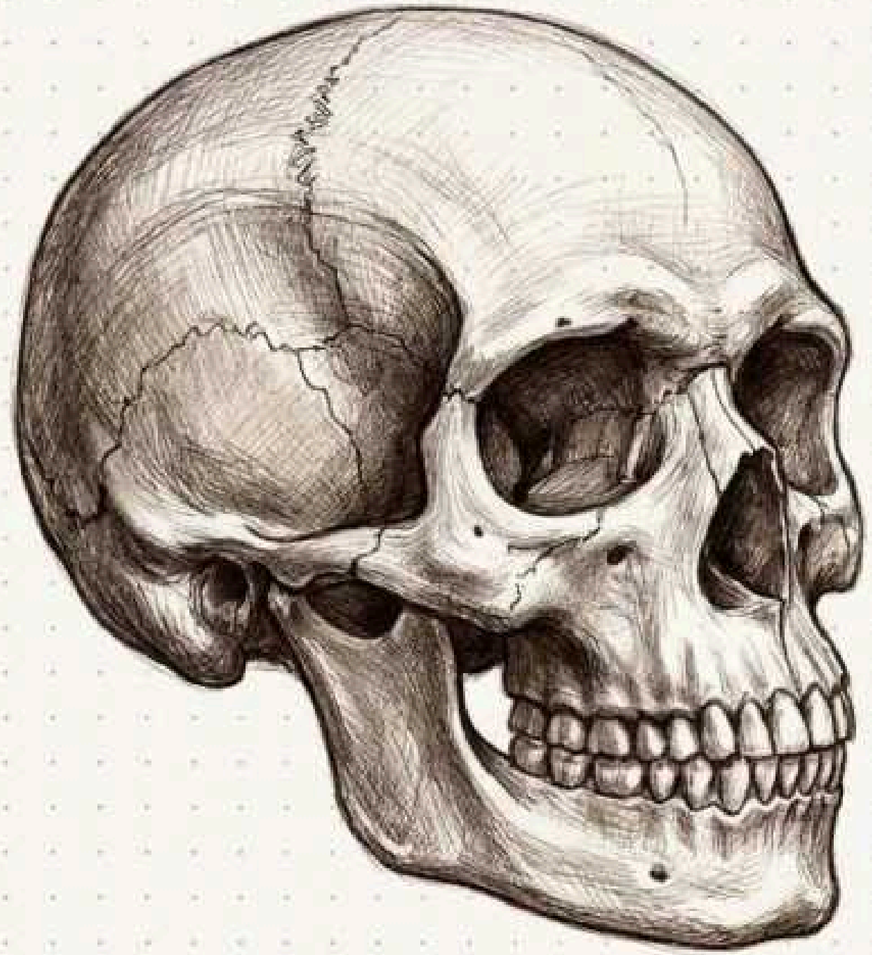
¶ 5.5

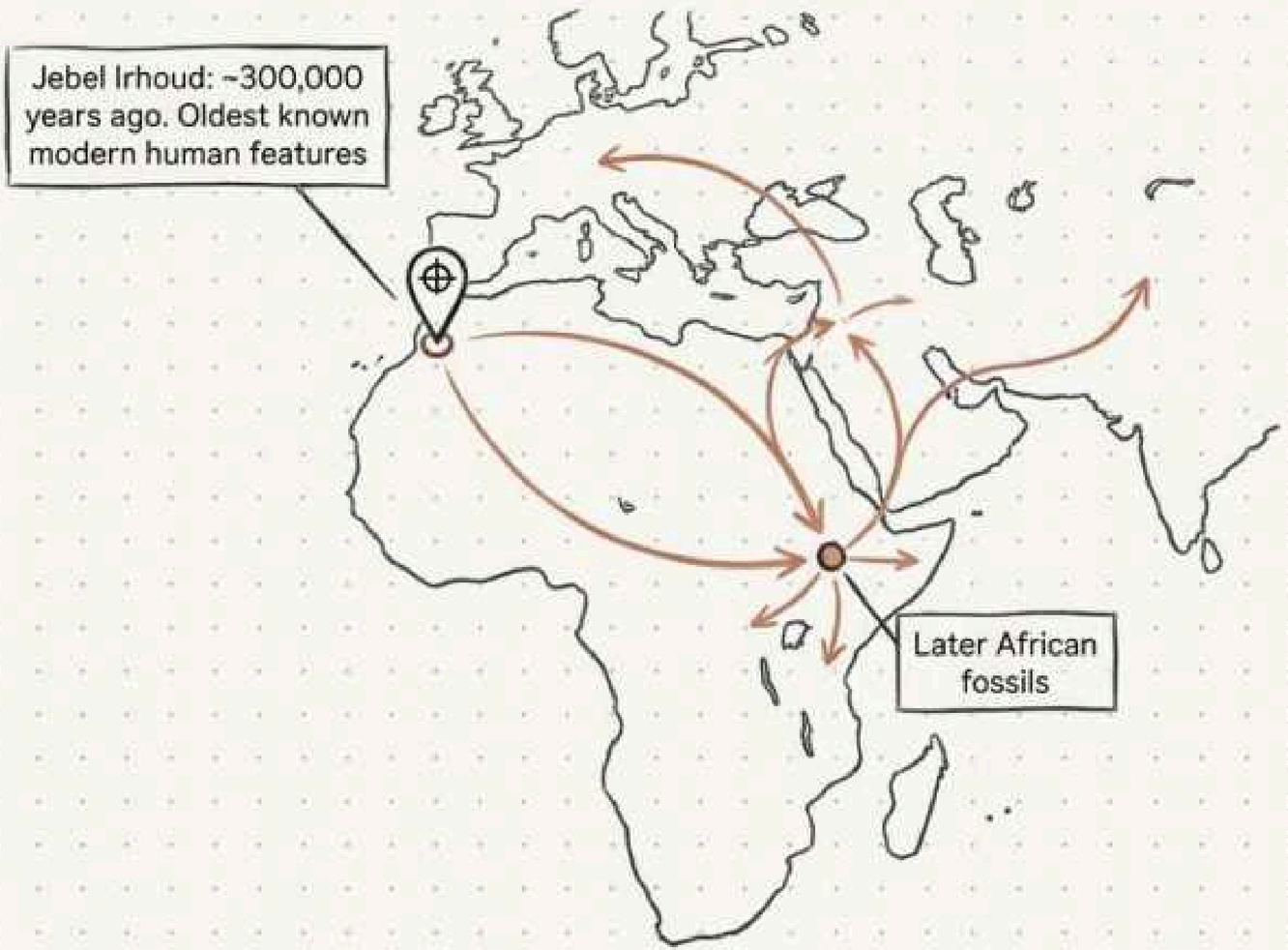


Conclusion: Genetic diversity is highest in Africa and strictly declines with geographic distance.

The Fossil Record Confirms the Homeland

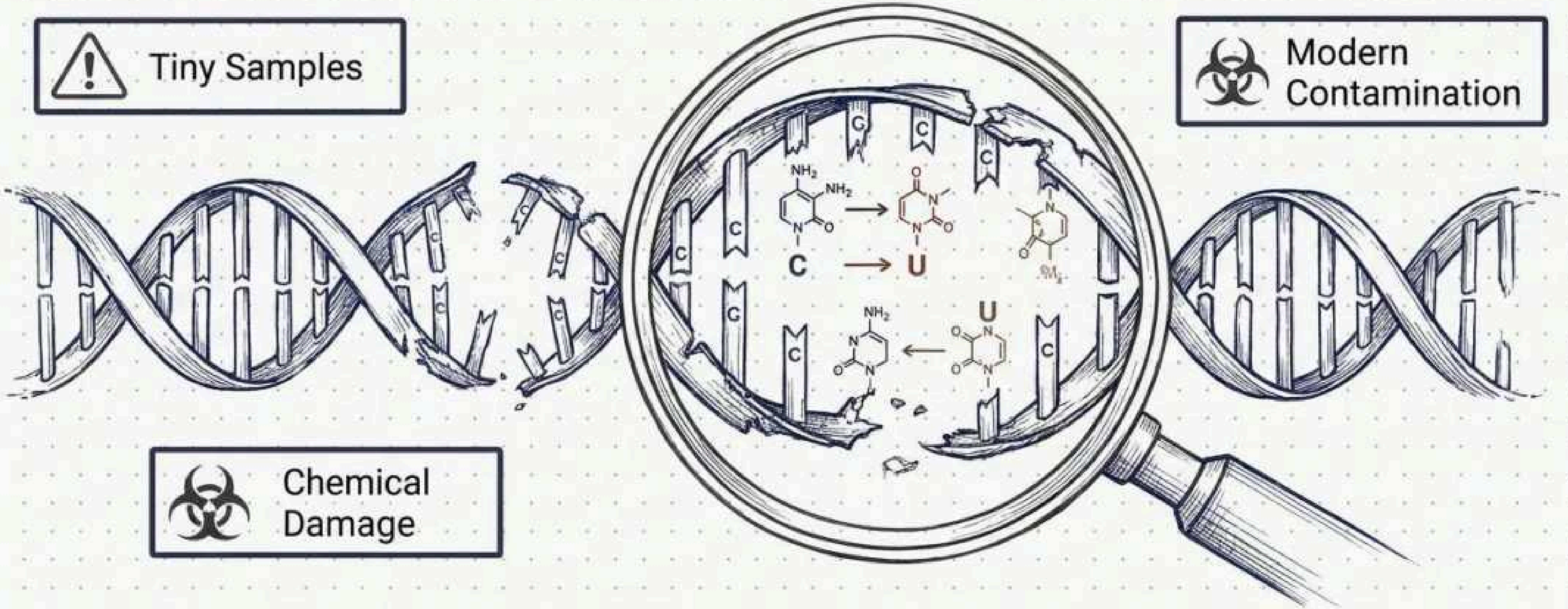
¶ 5.6





Genetics, fossils, and archaeology converge:
Africa is the clear homeland of our species.

The Ancient DNA Revolution



Nobel Prize in Physiology or Medicine (2022)

Svante **Pääbo**.

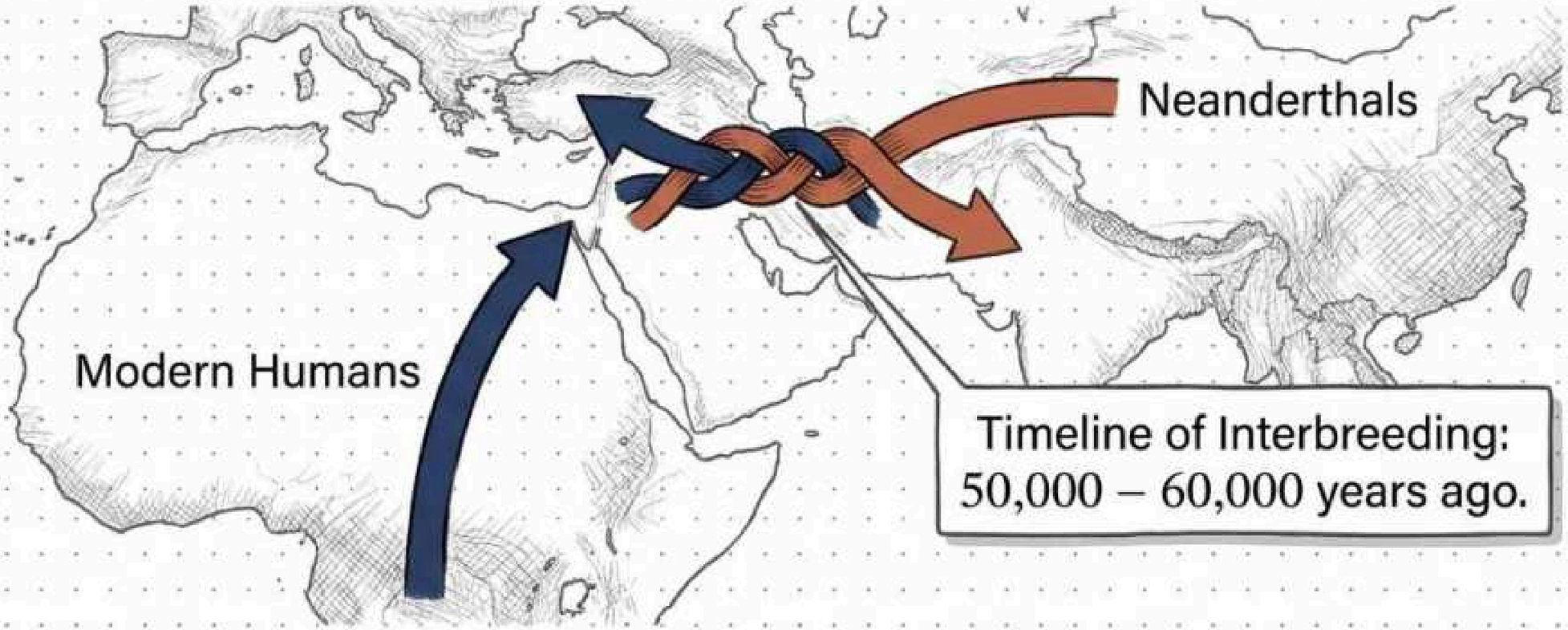
Methodology:

Decades of work utilizing clean rooms and recognizing typical damage patterns to sequence the un-sequenceable.

The Neanderthal Draft Genome (2010)

¶ 5.8

Green, Pääbo, and colleagues.



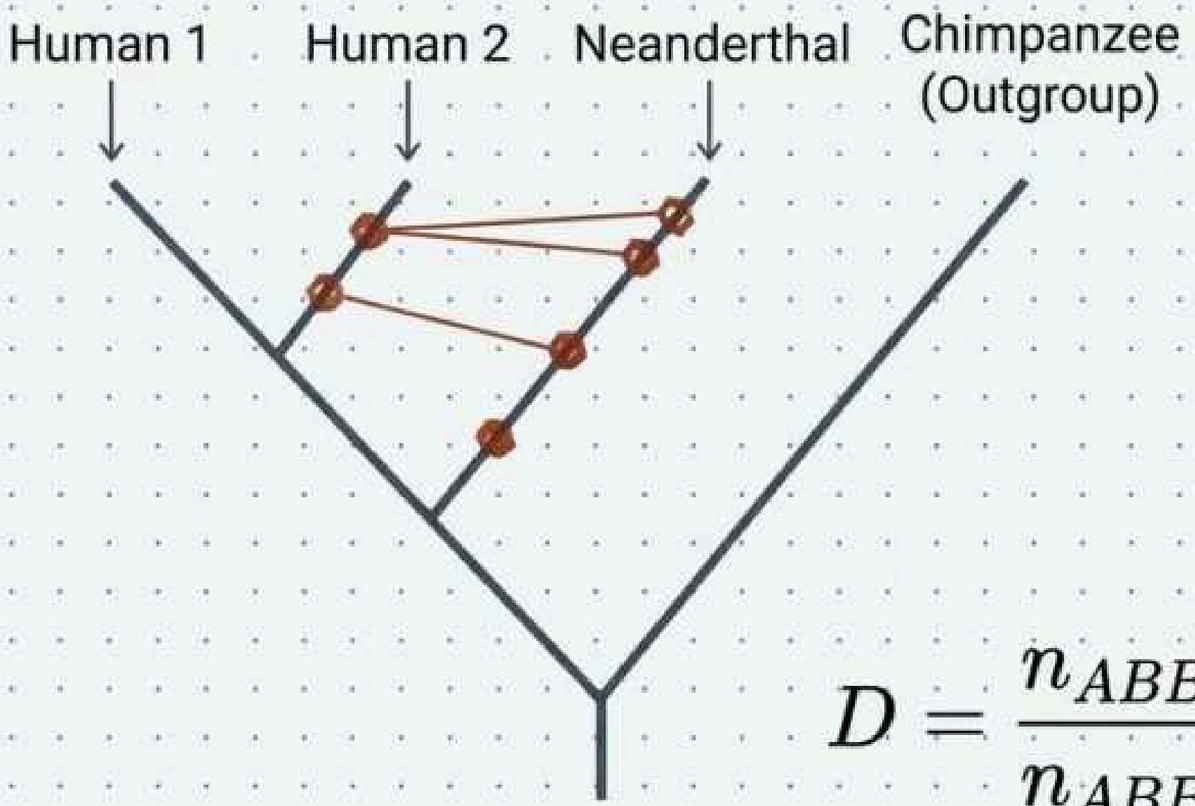


Impact: Non-Africans carry roughly 1% to 2% Neanderthal ancestry.

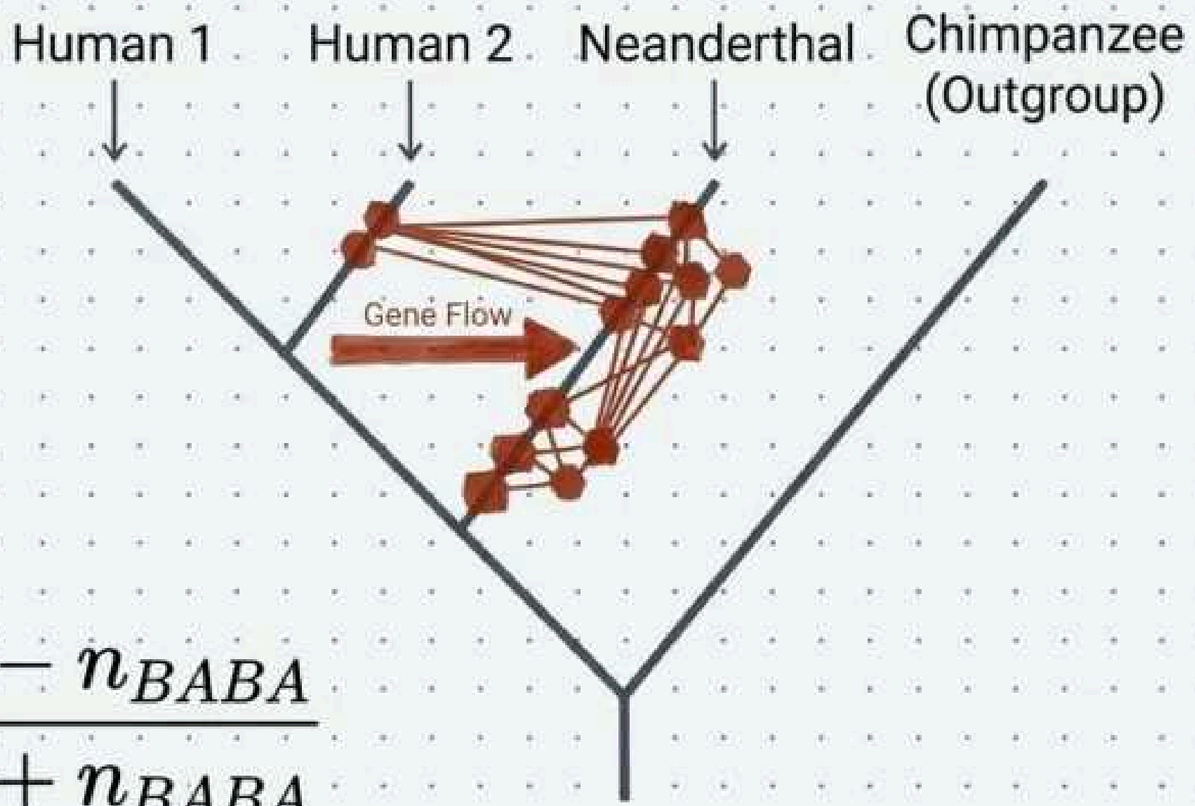
Detecting Admixture: The ABBA-BABA Test

¶ 5.9

Incomplete Lineage Sorting



Introgression

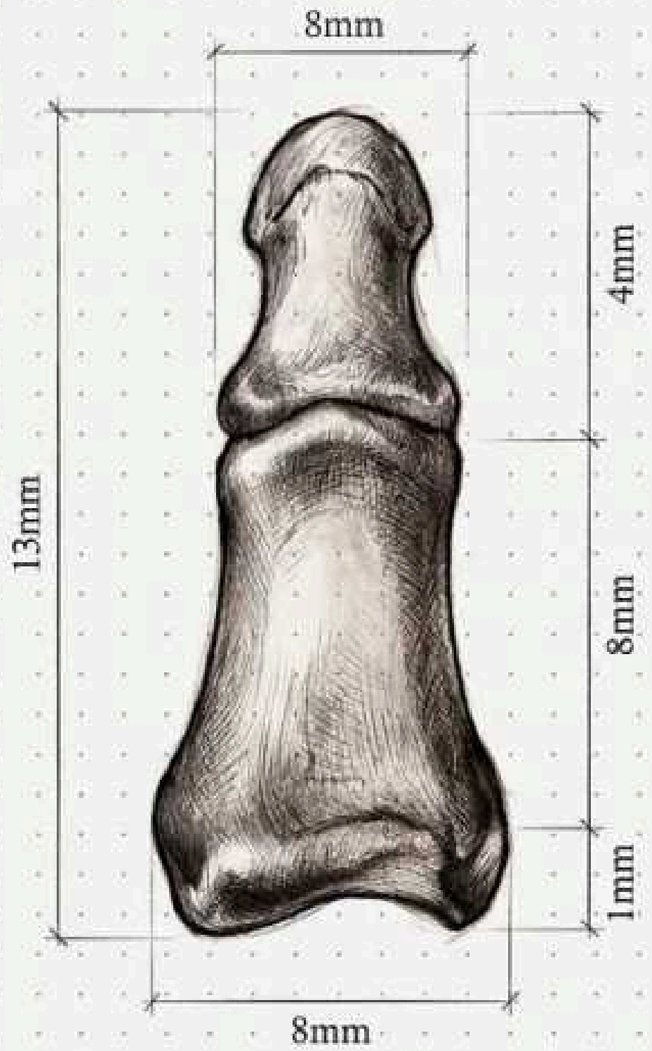


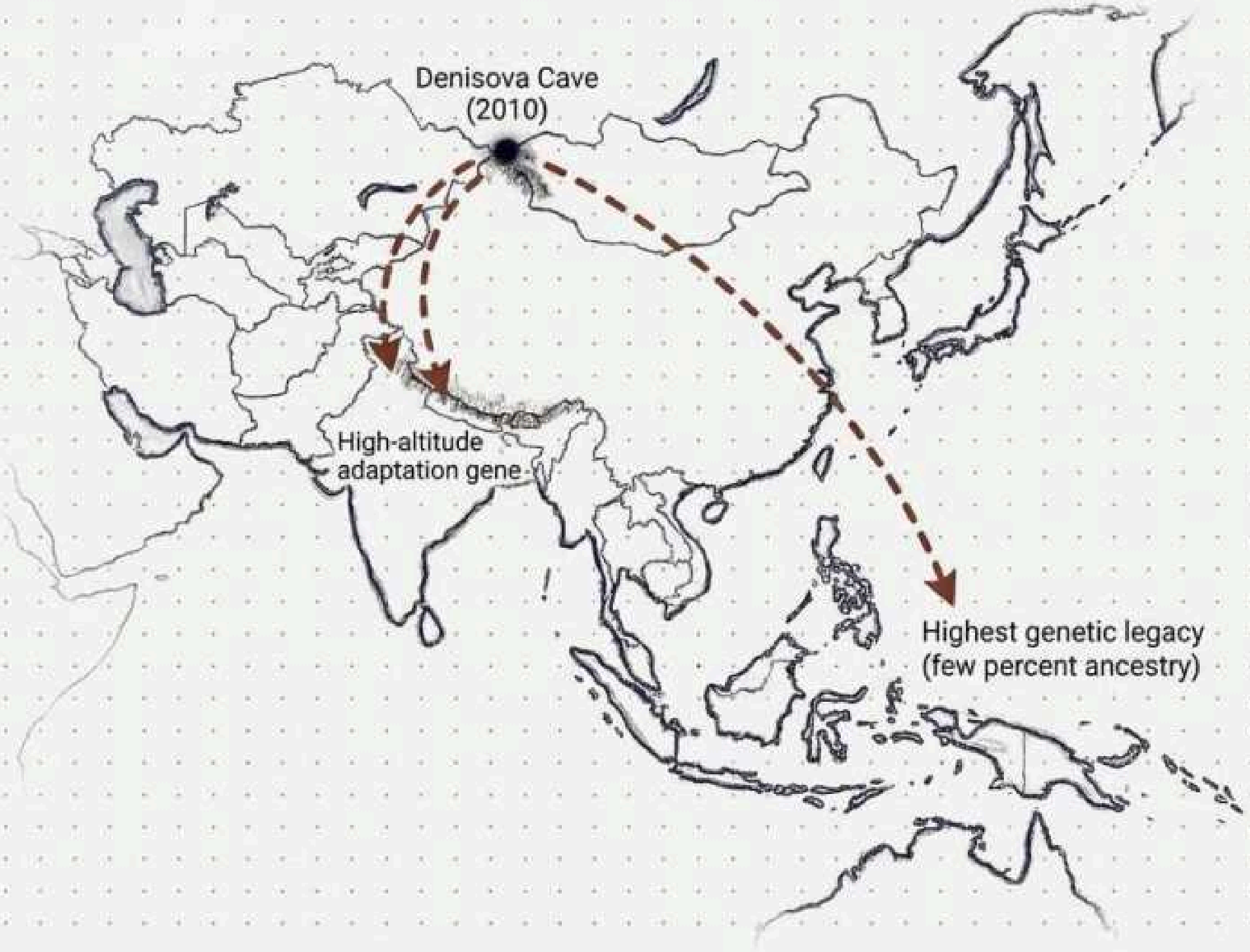
$$D = \frac{n_{ABBA} - n_{BABA}}{n_{ABBA} + n_{BABA}}$$

If no mixing occurred, sharing is equal.

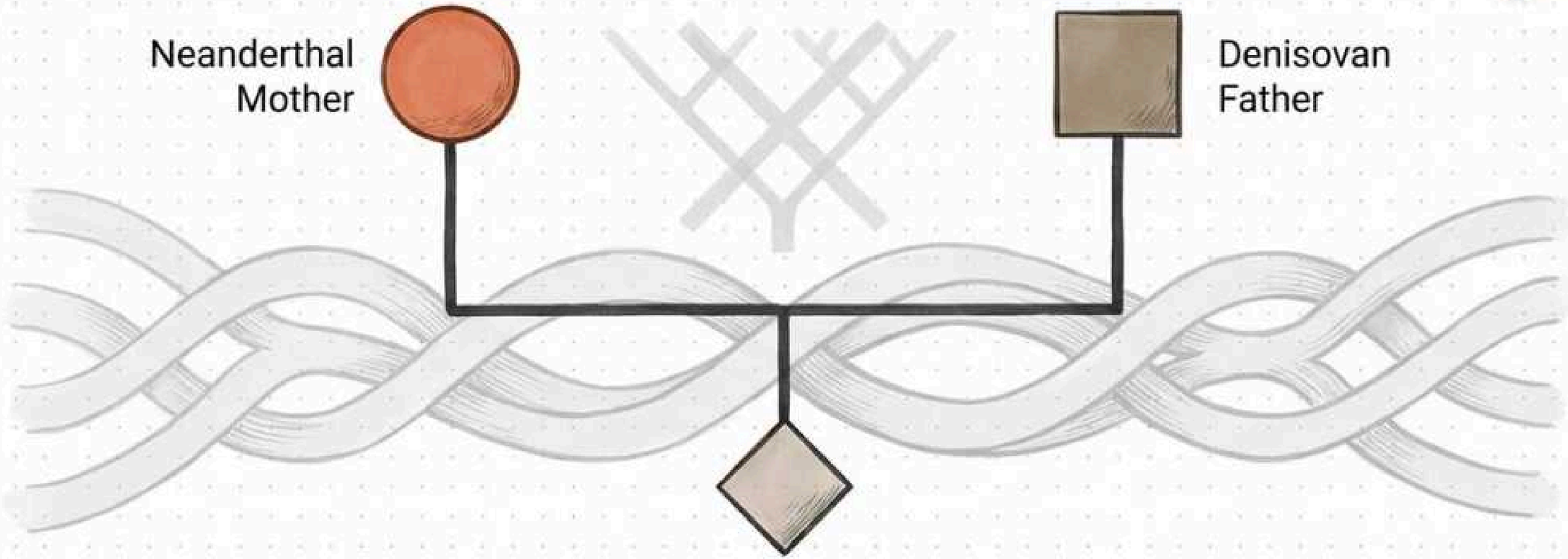
An excess of sharing with one modern human proves gene flow.

The Denisovans: A Ghost Population Revealed





“Denny”: Direct Evidence of Archaic Mixing

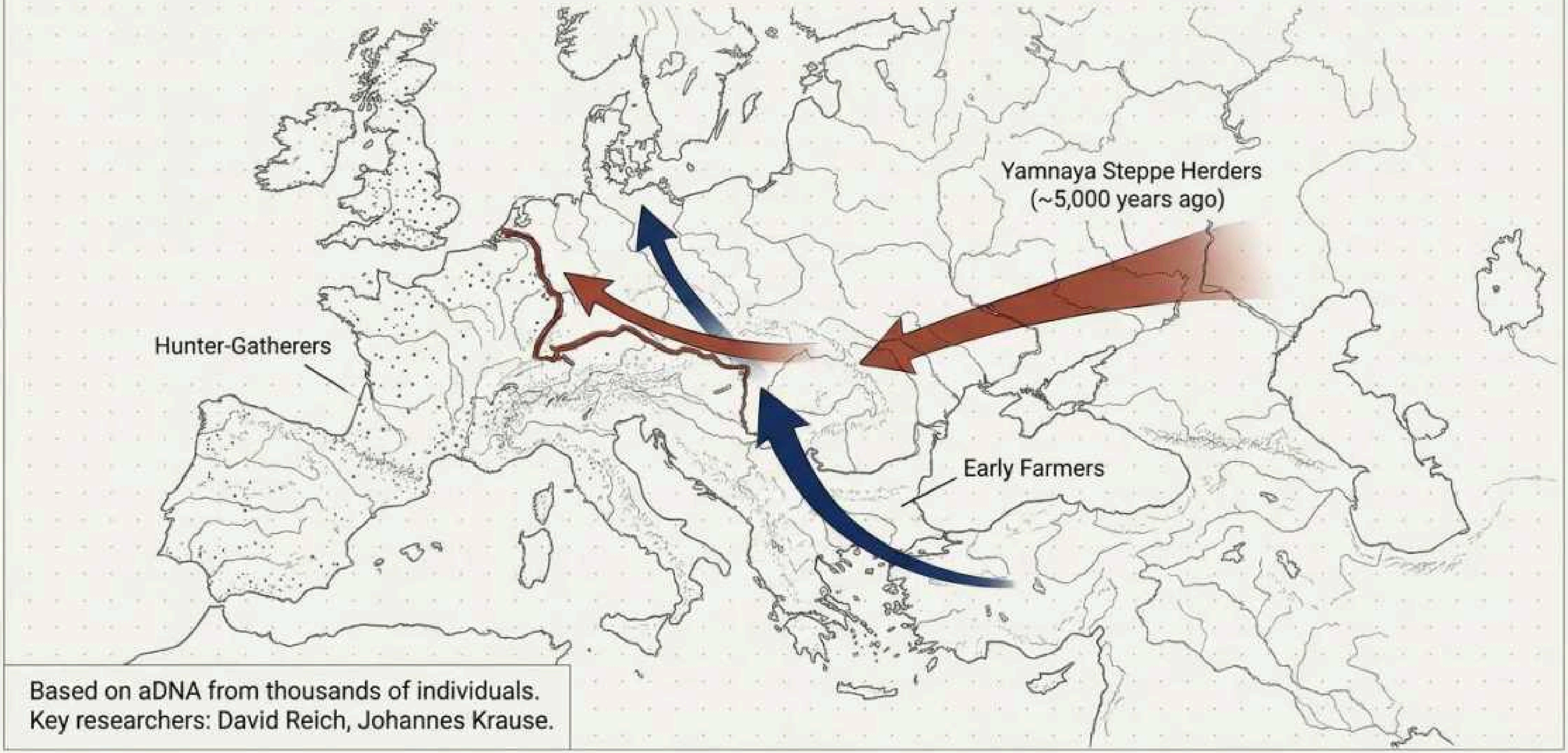


Published in 2018. Discovered in Denisova Cave.

The human past was not a simple tree of separate populations, but a branching network with frequent mixing.

¶ 5.12

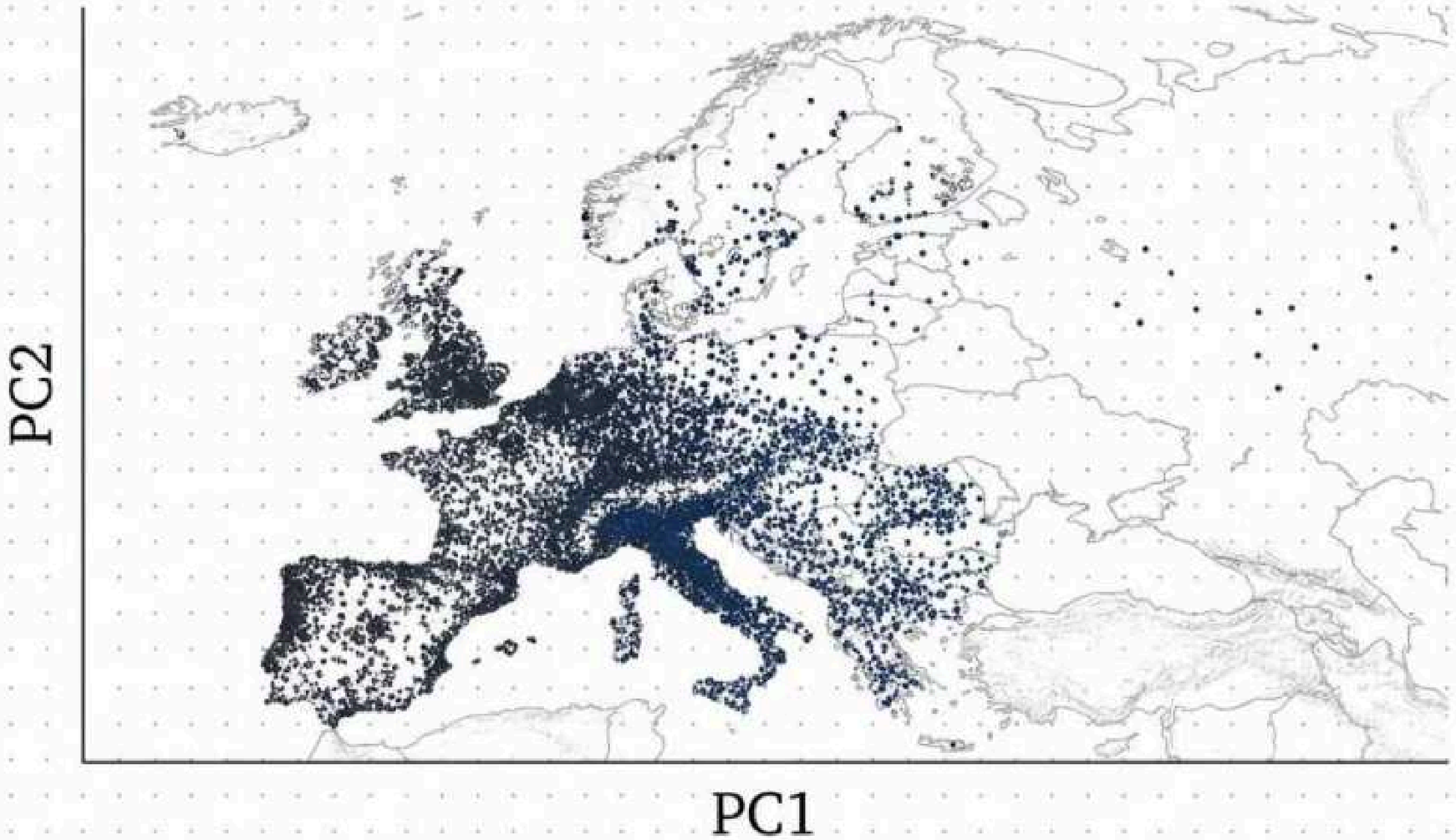
The Making of Modern Europe (<10,000 years ago)



Based on aDNA from thousands of individuals.
Key researchers: David Reich, Johannes Krause.

Principal Components: Genes Mirror Geography

Novembre et al., 2008.



PCA summarizes thousands of sites into a few dimensions. The pattern emerges because people historically had children with partners living nearby.

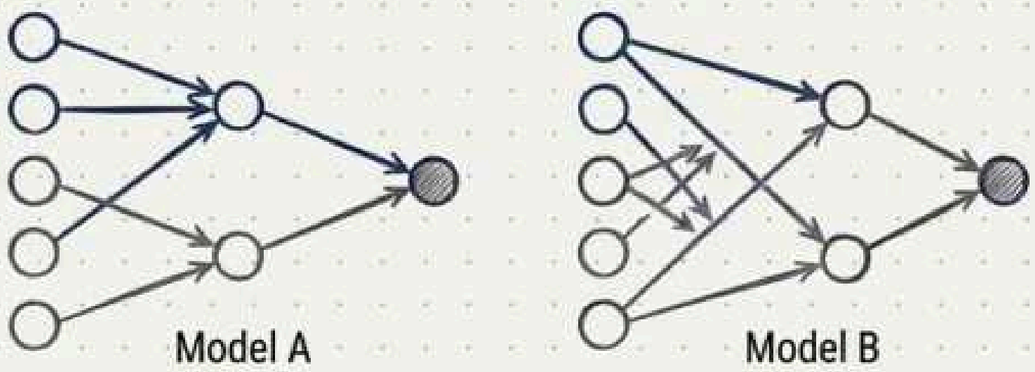
Scientific Guardrails and Caveats

Uneven Sampling



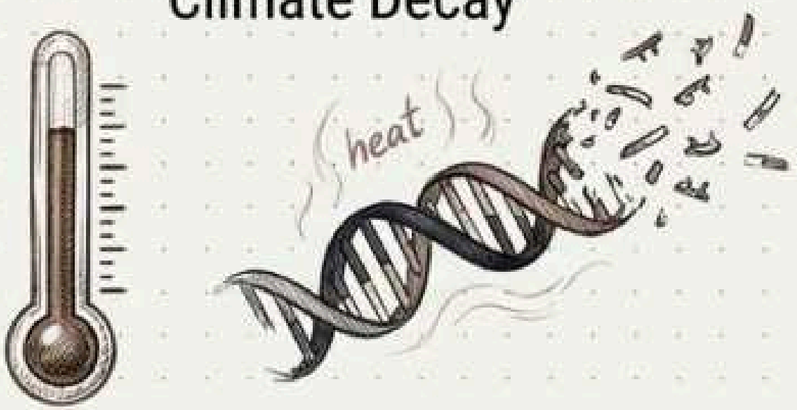
PCA can be heavily distorted by who is sequenced.

Model Dependency



Admixture estimates depend entirely on the chosen mathematical model.

Climate Decay



aDNA is exceptionally scarce in tropical climates where DNA decays rapidly.

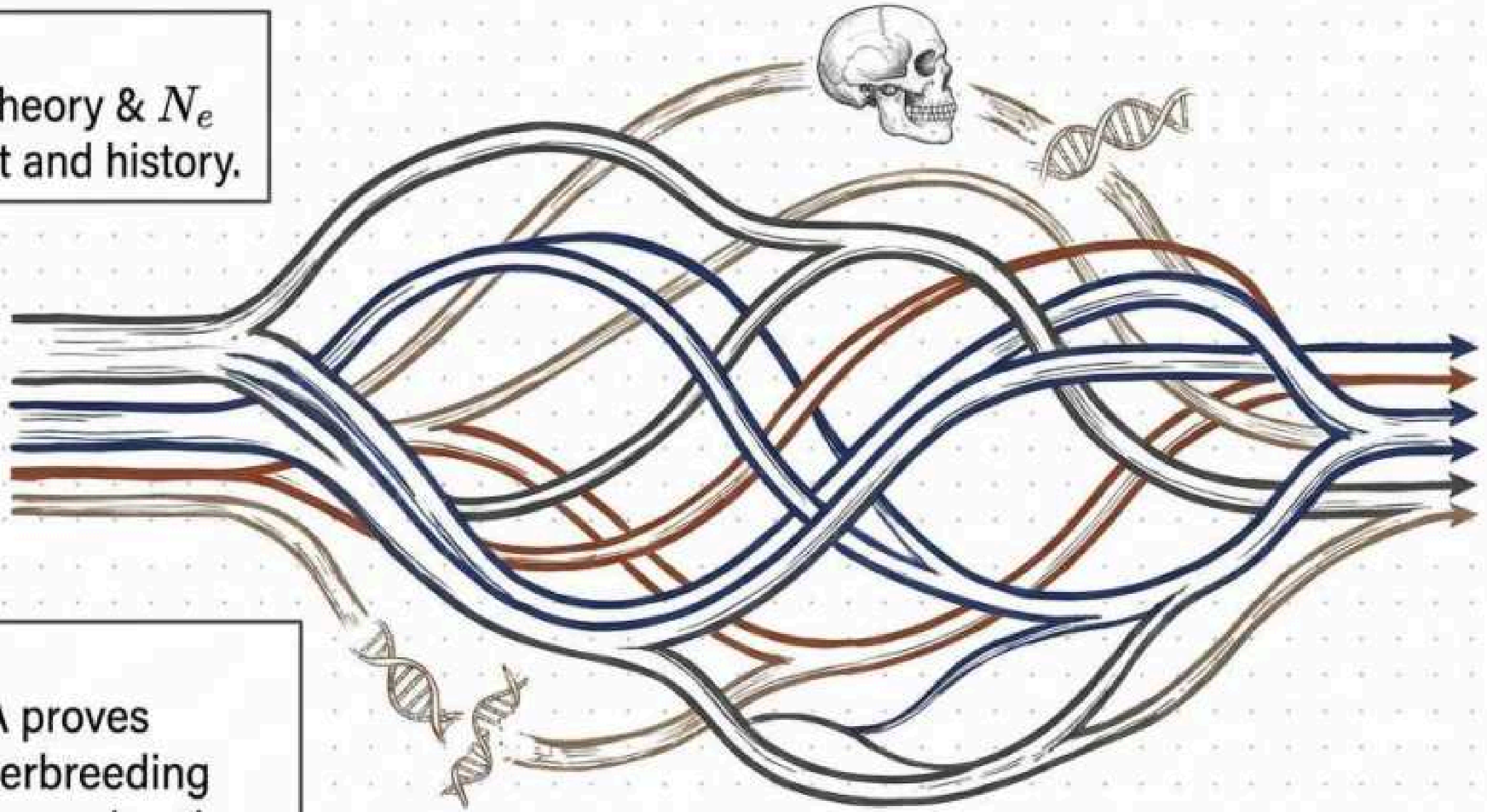
Ethical Misuse



Results must be communicated carefully to combat false claims of purity or superiority that the data explicitly contradict.

A Braided Stream: Synthesis of Population History

The Tool
Coalescent theory & N_e measure drift and history.



The Origin
Diversity and fossils lock our roots in Africa.

The Reality
Ancient DNA proves profound interbreeding and recent mass migrations.

Human history is a network of populations, not a set of separate branches.
Next: Testing Adaptation.