

Archaic human introgression

Population Genetics 2026

Martin Petr

mp@bodkan.net

February 25, 2026

1856: Discovery of a ‘new human’



<https://www.donsmaps.com/neanderthaloriginal.html>



<https://twitter.com/Qafzeh/status/805339276334333953>

1856: Discovery of a ‘new human’



<https://www.donsmaps.com/neanderthaloriginal.html>



<https://twitter.com/Qafzeh/status/805339276334333953>

Neander's valley

1856: Discovery of a ‘new human’



<https://www.donsmaps.com/neanderthaloriginal.html>



<https://twitter.com/Qafzeh/status/805339276334333953>

Neander's valley

valley = Thal in German

1856: Discovery of a ‘new human’



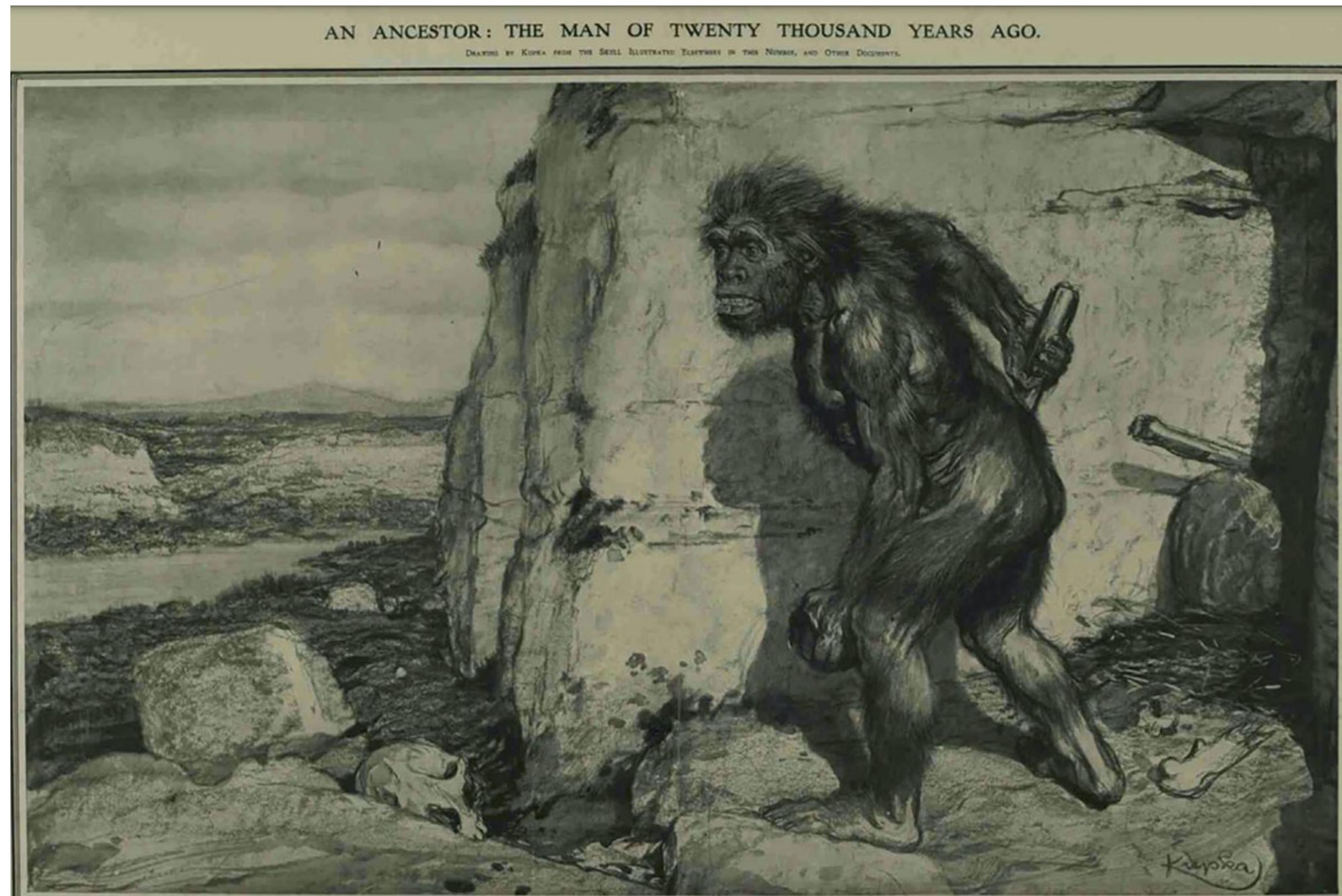
<https://www.donsmaps.com/neanderthaloriginal.html>

Neander's valley
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<https://twitter.com/Qafzeh/status/805339276334333953>

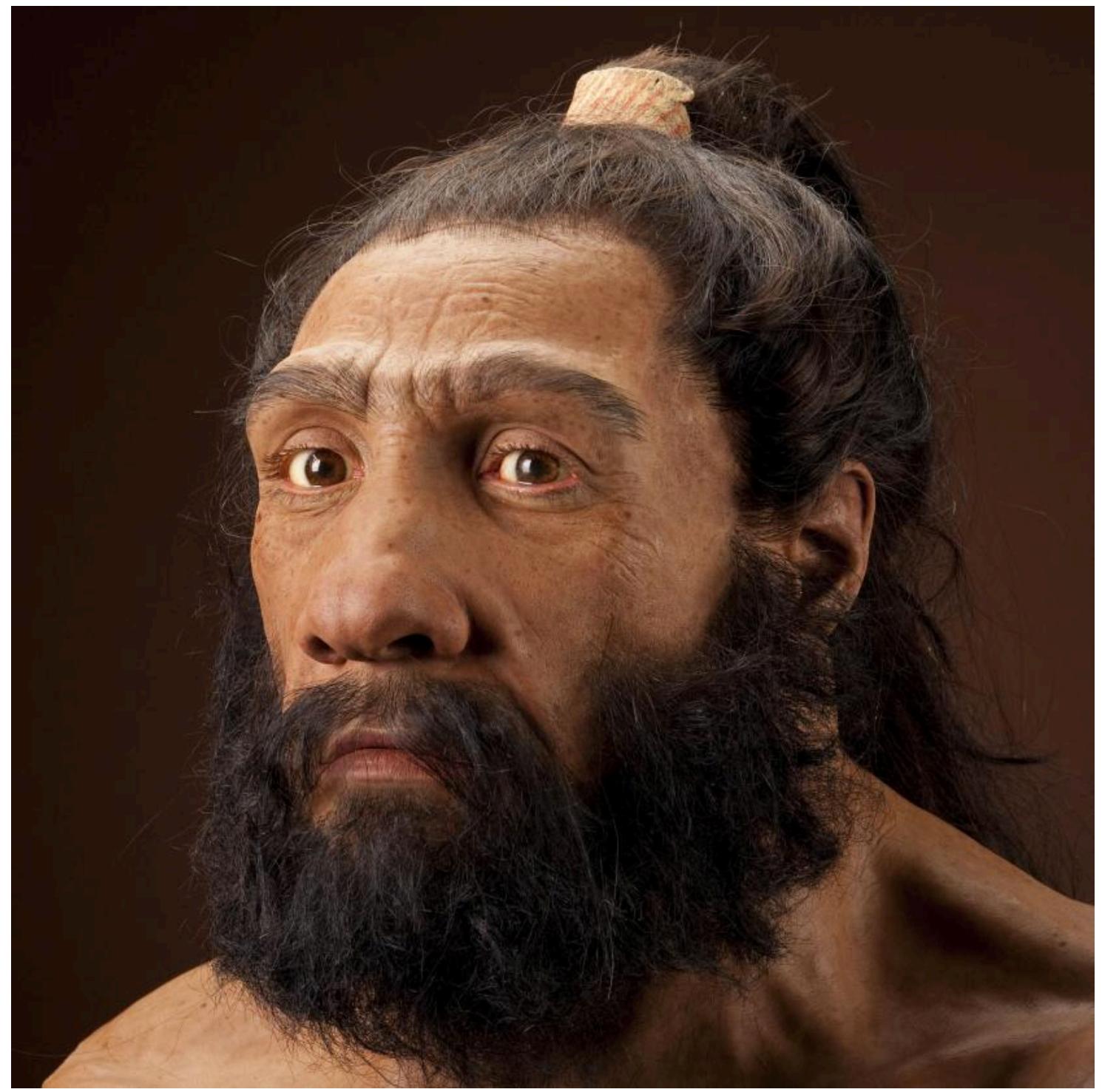
Neanderthal



The Illustrated London News, 1909



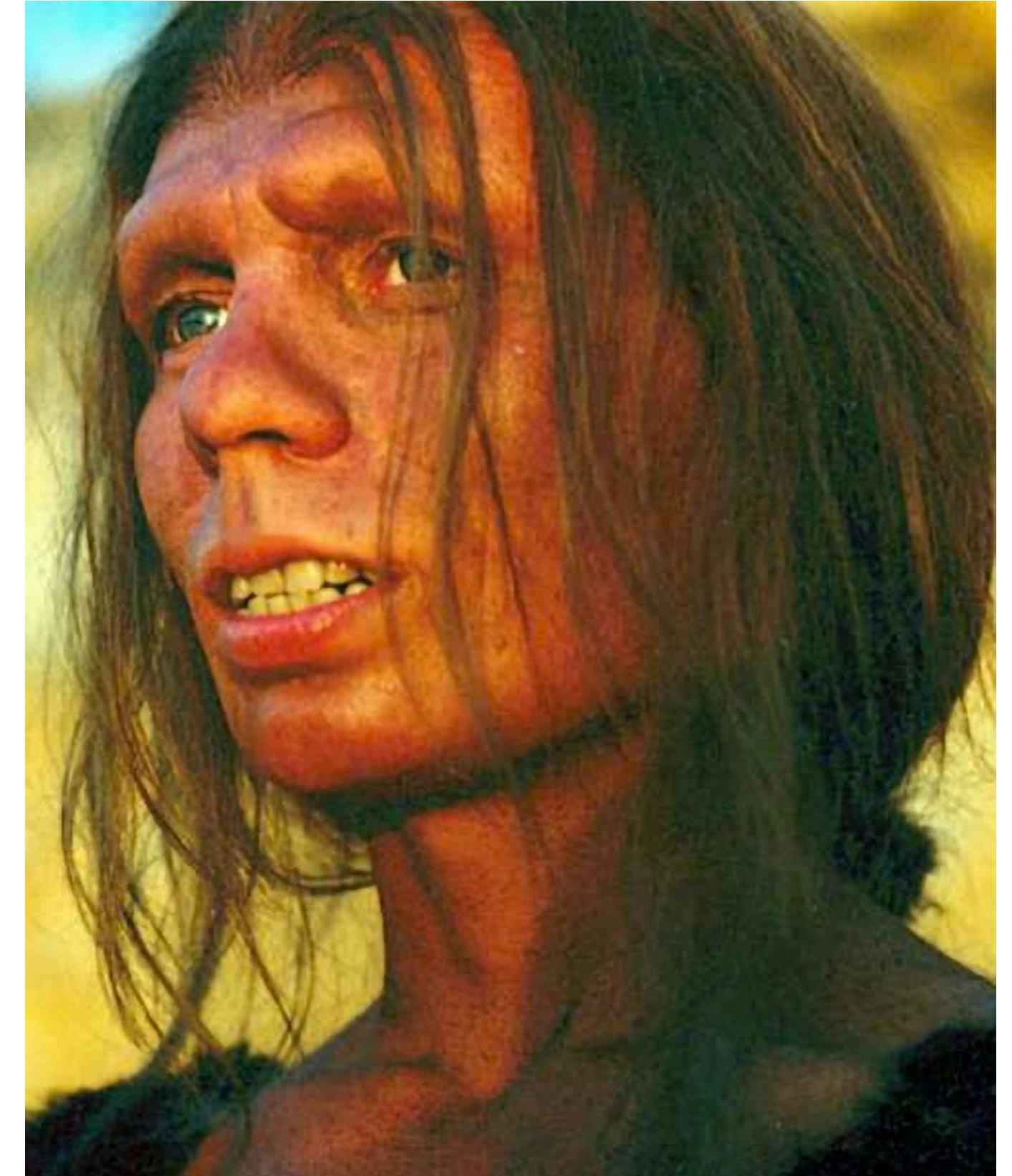
The Neanderthal Man movie poster, 1953



<http://humanorigins.si.edu/evidence/human-fossils/species/homo-neanderthalensis>



<https://www.neanderthal.de>



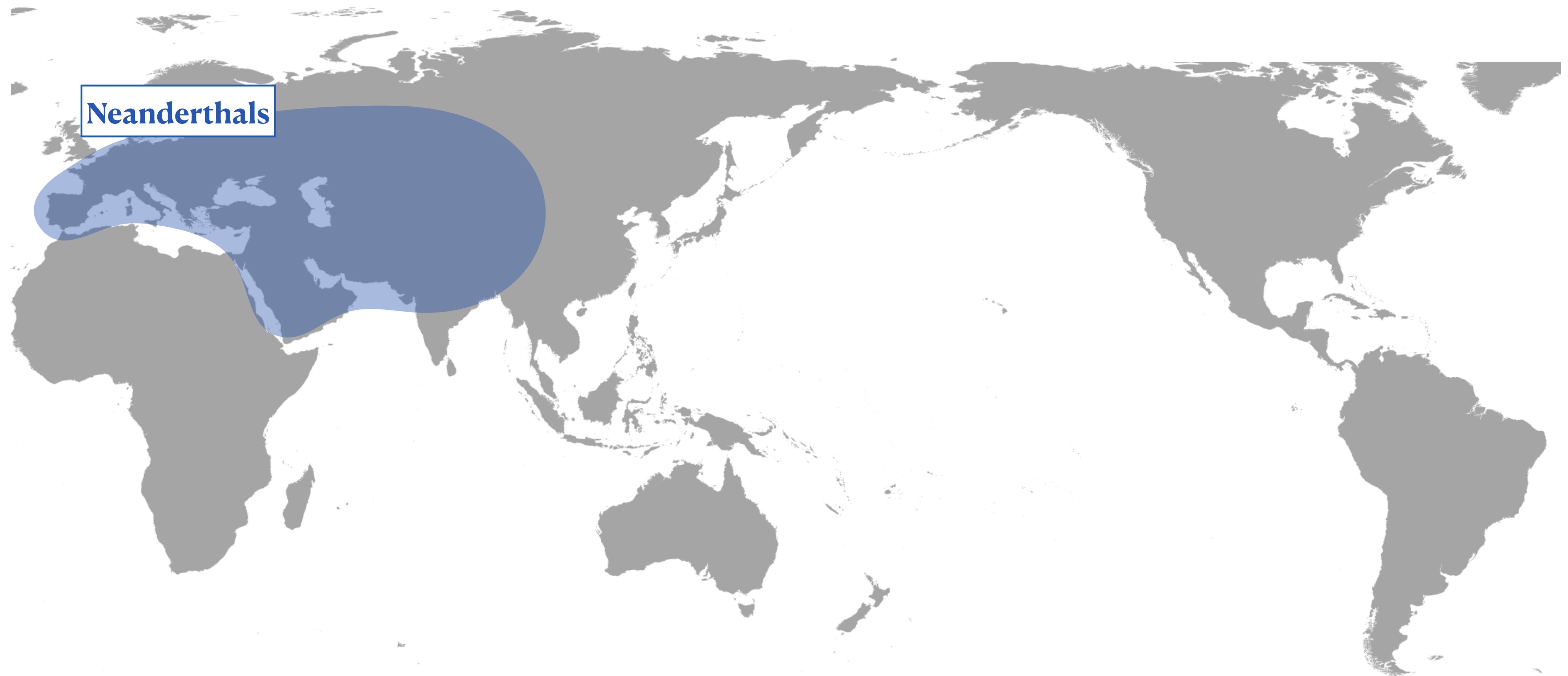
<https://journals.plos.org/plosbiology/article?id=10.1371/journal.pbio.0020449>



Neanderthal Museum, Mettmann, Germany

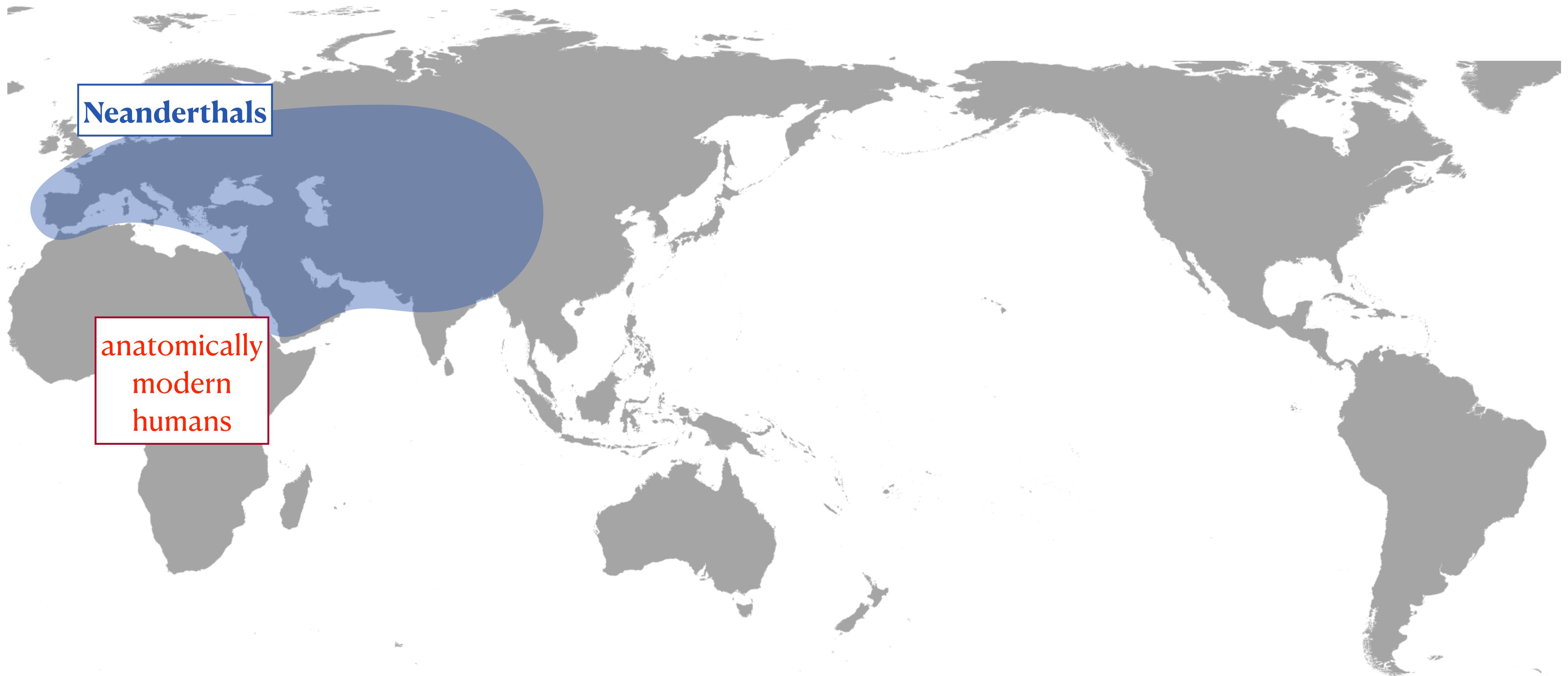


~600 kya B.P.: Neanderthals in Eurasia



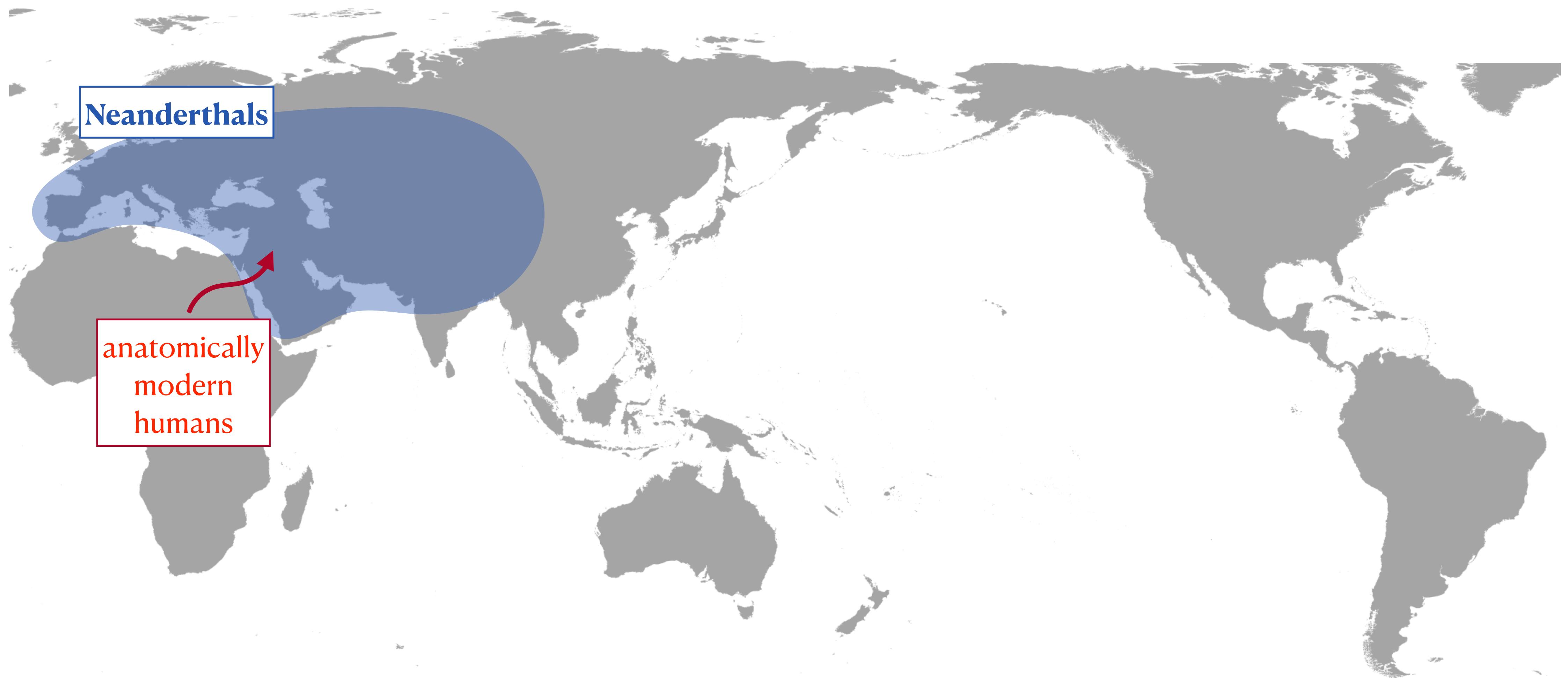
B.P. = "before present"

~300 kya B.P.: anatomically modern humans in Africa



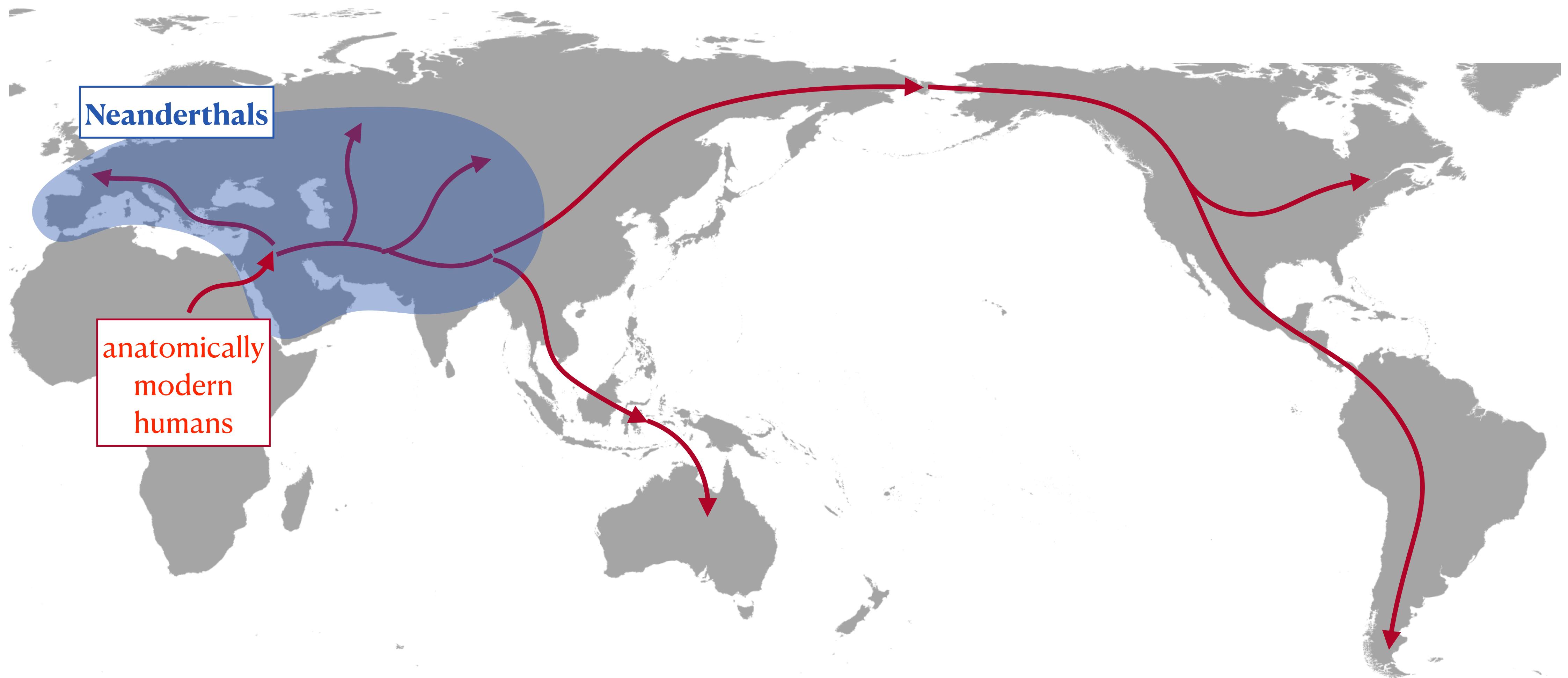
B.P. = "before present"

~70 kya B.P.: modern humans migrated out of Africa



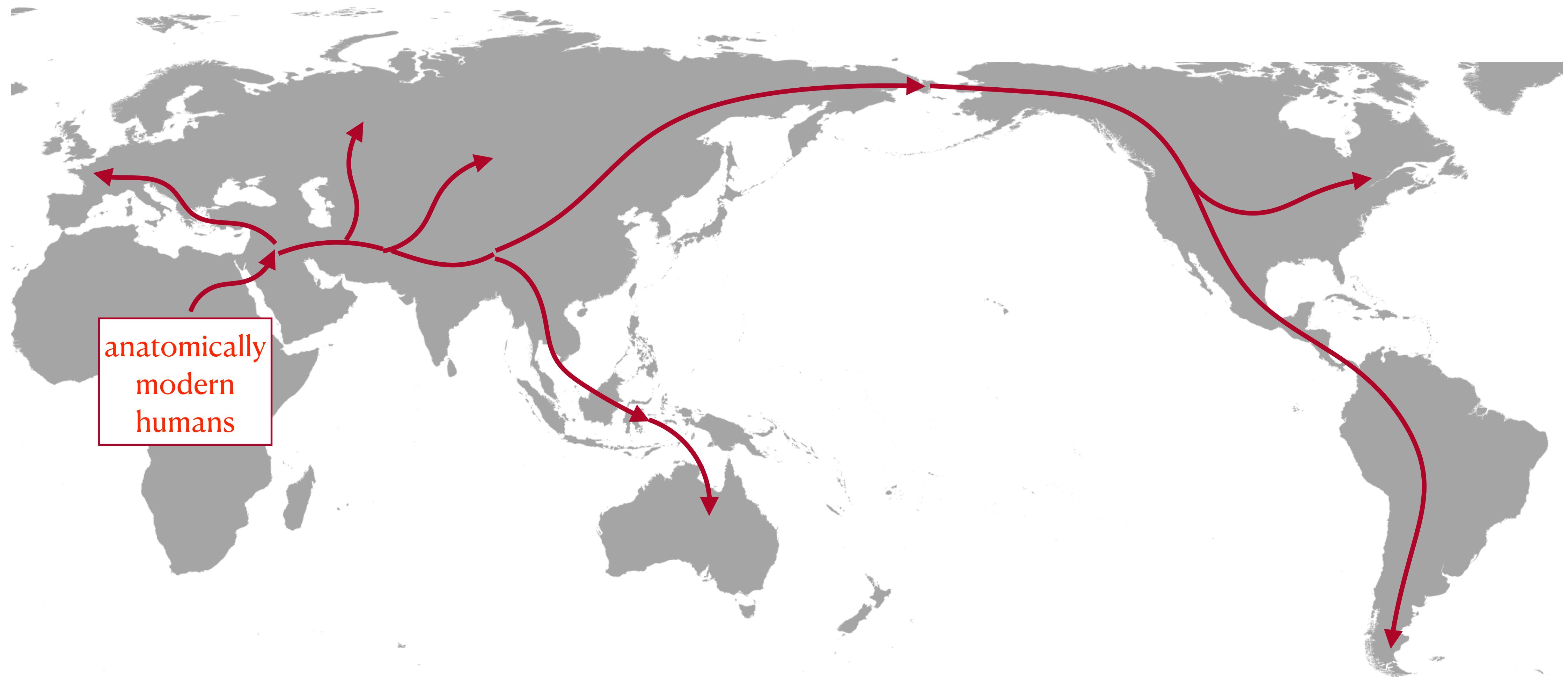
B.P. = "before present"

~70 kya B.P.: modern humans migrated out of Africa ...and dispersed around the world



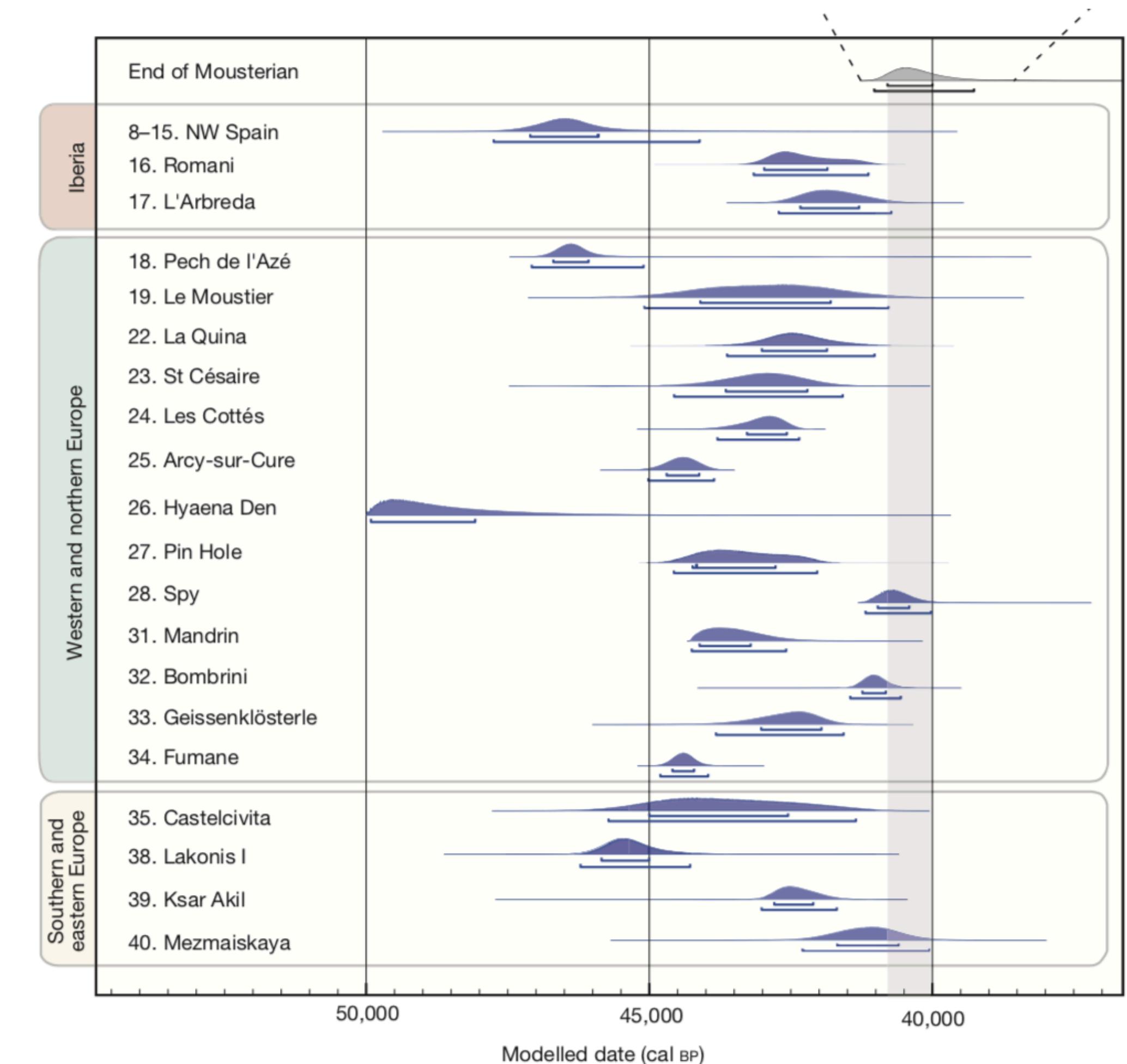
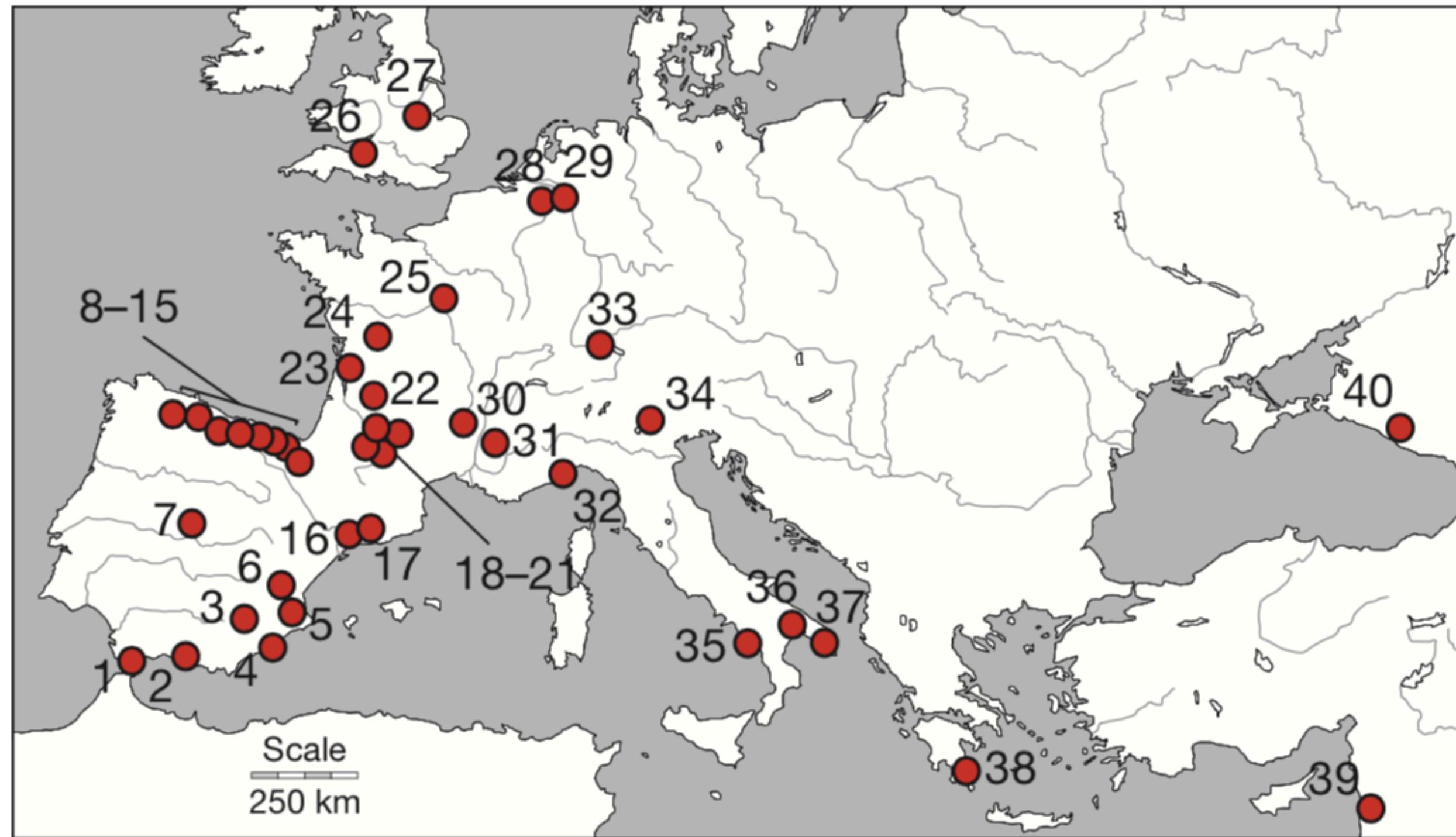
B.P. = "before present"

~40 kya: Neanderthals vanished

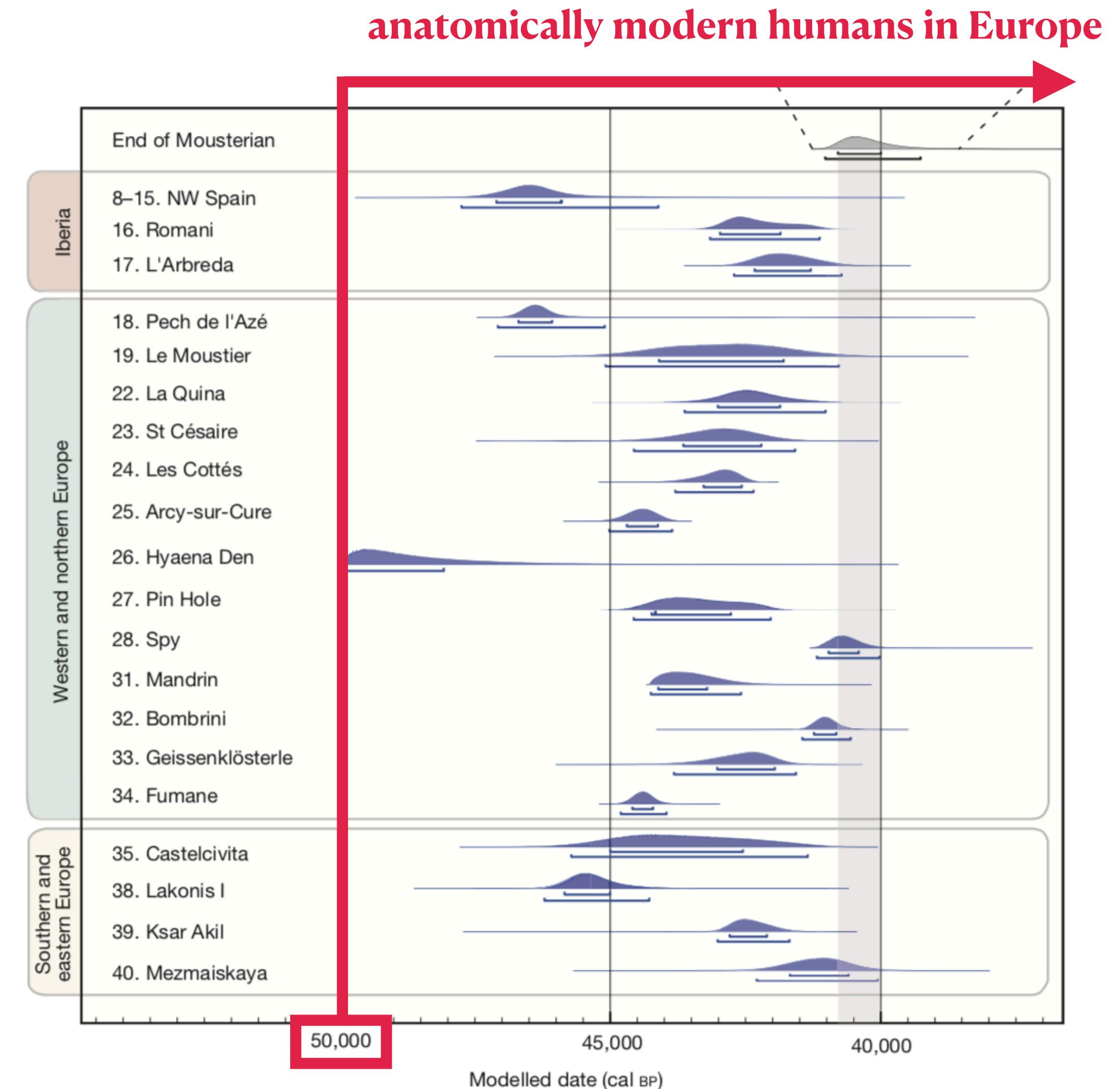
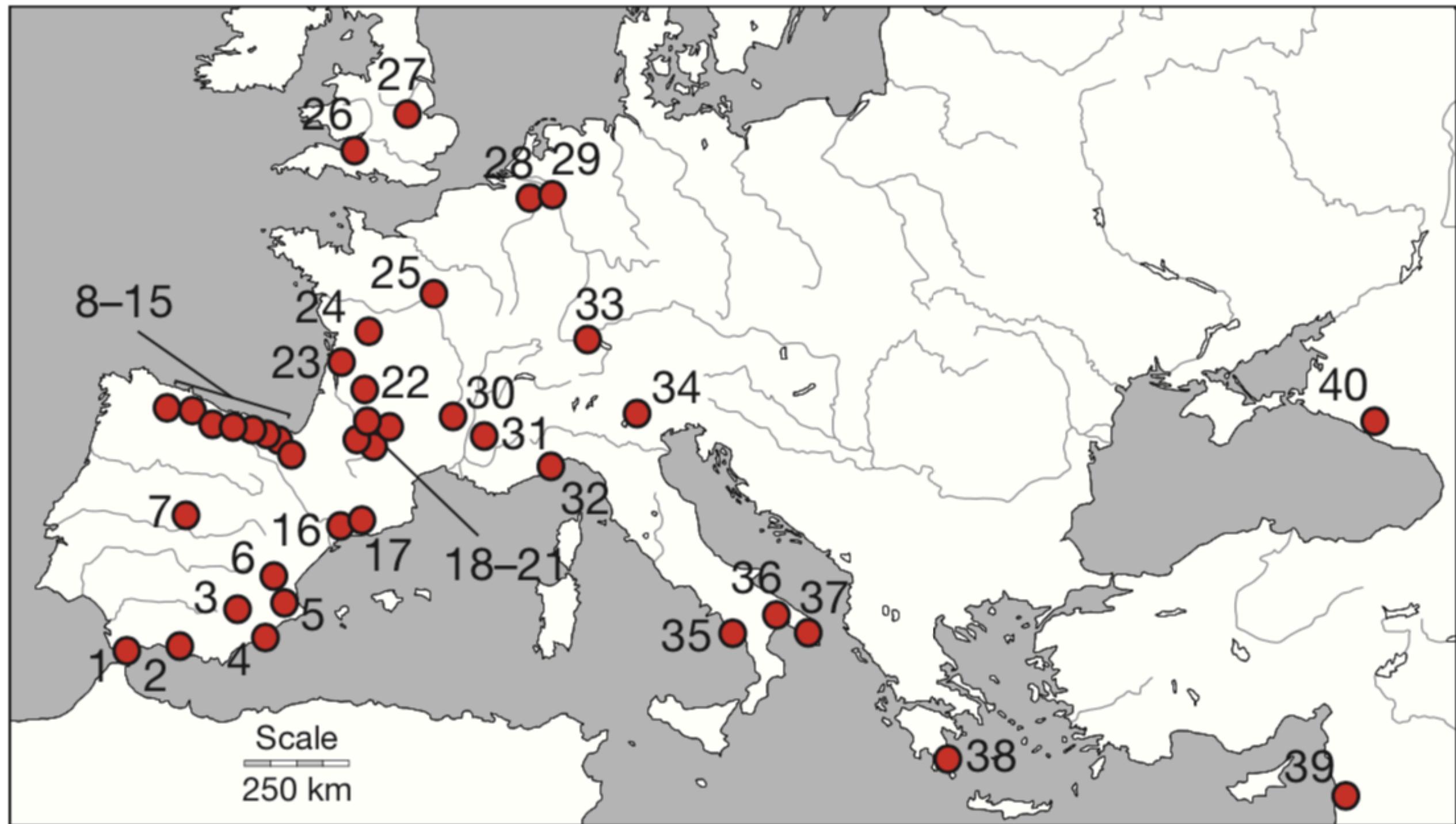


B.P. = "before present"

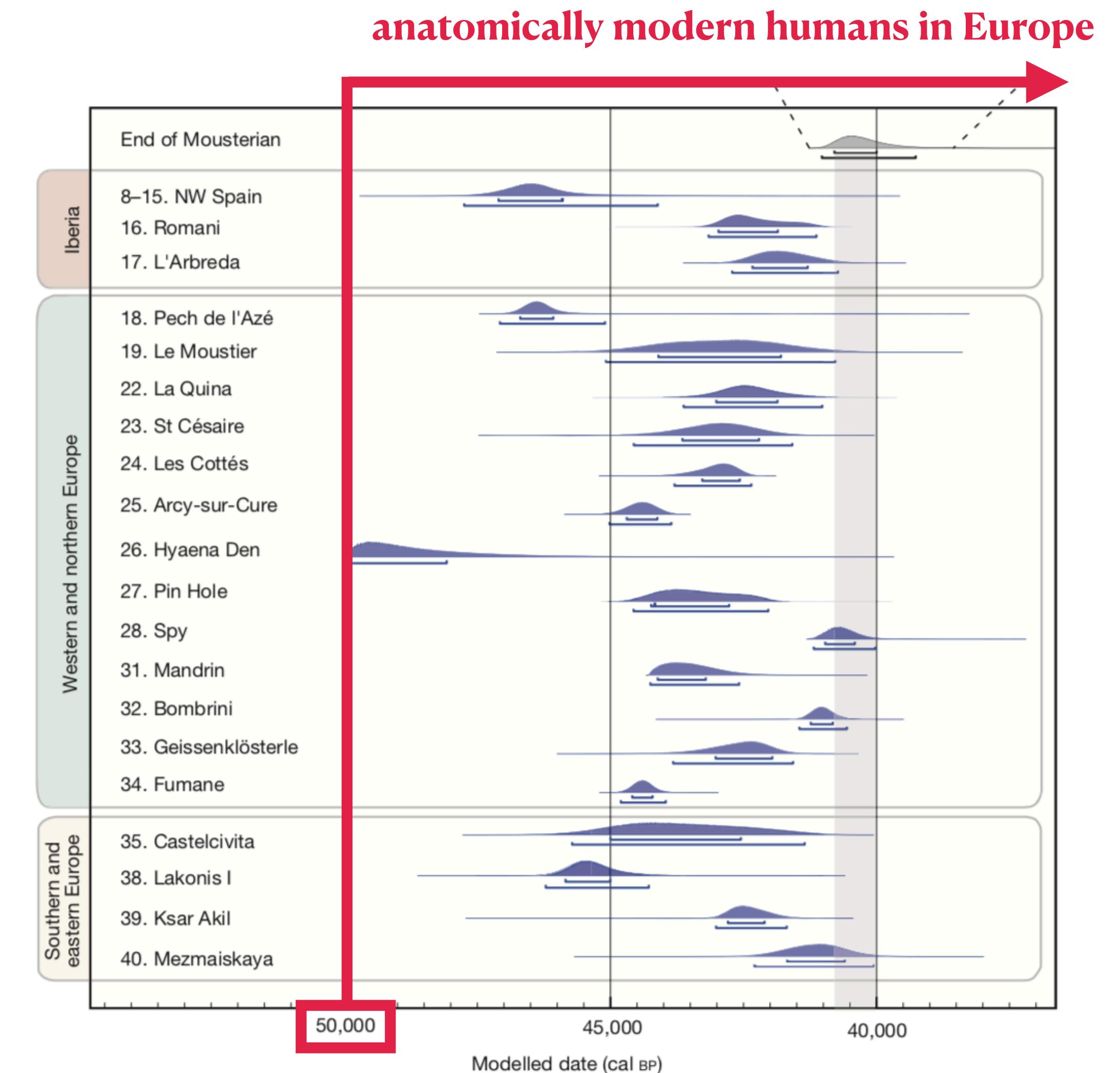
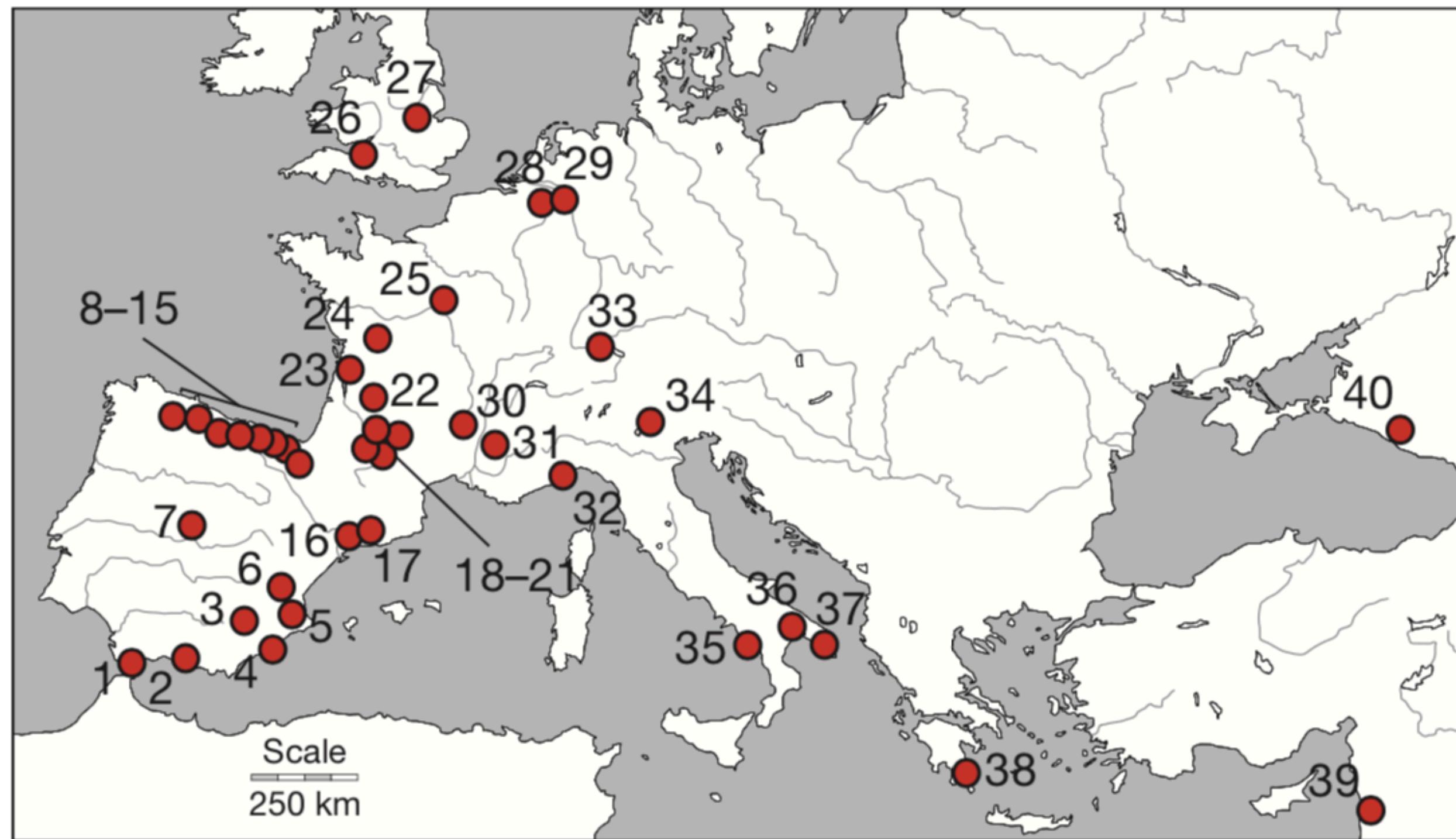
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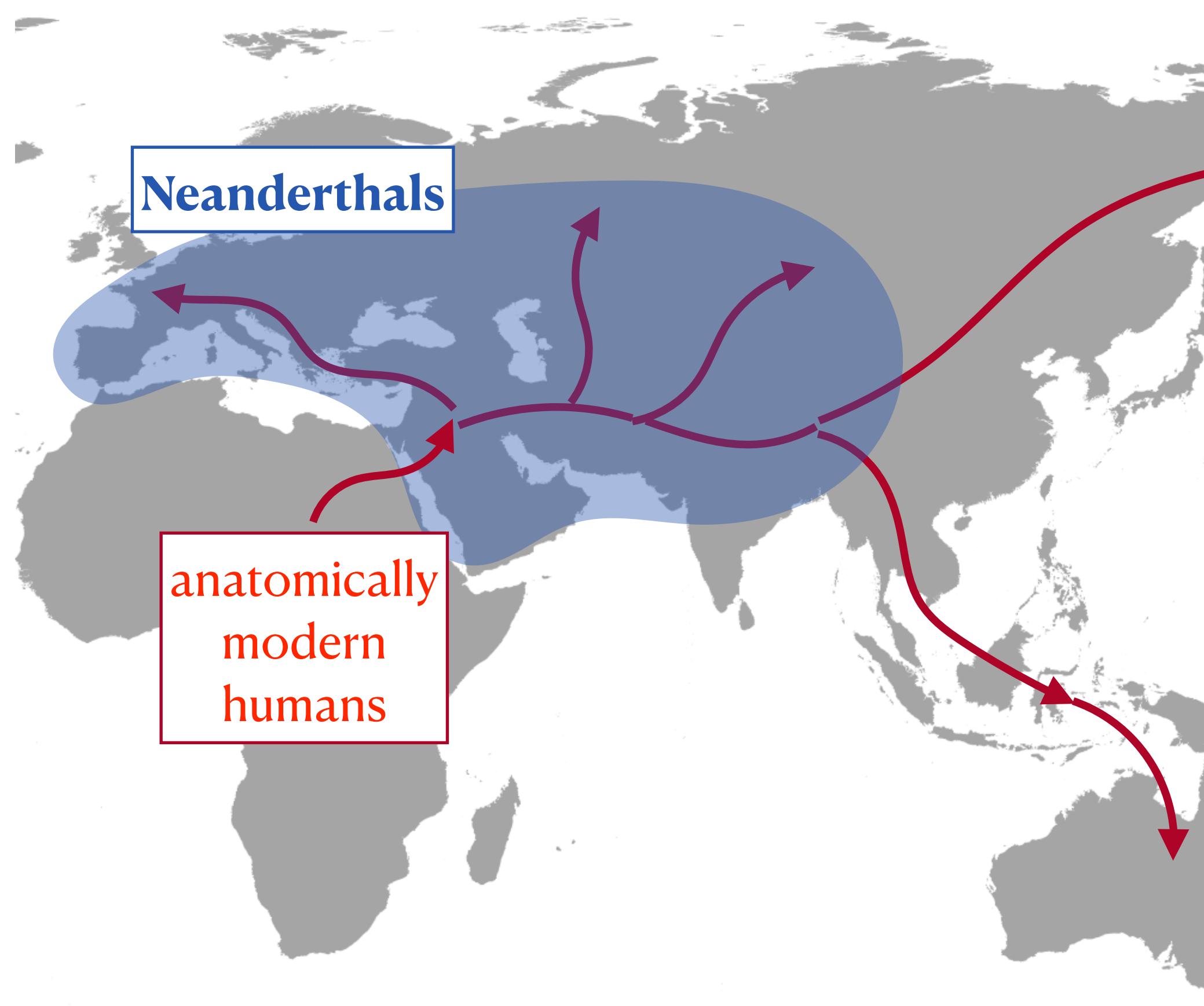
~40 kya: Neanderthals vanished



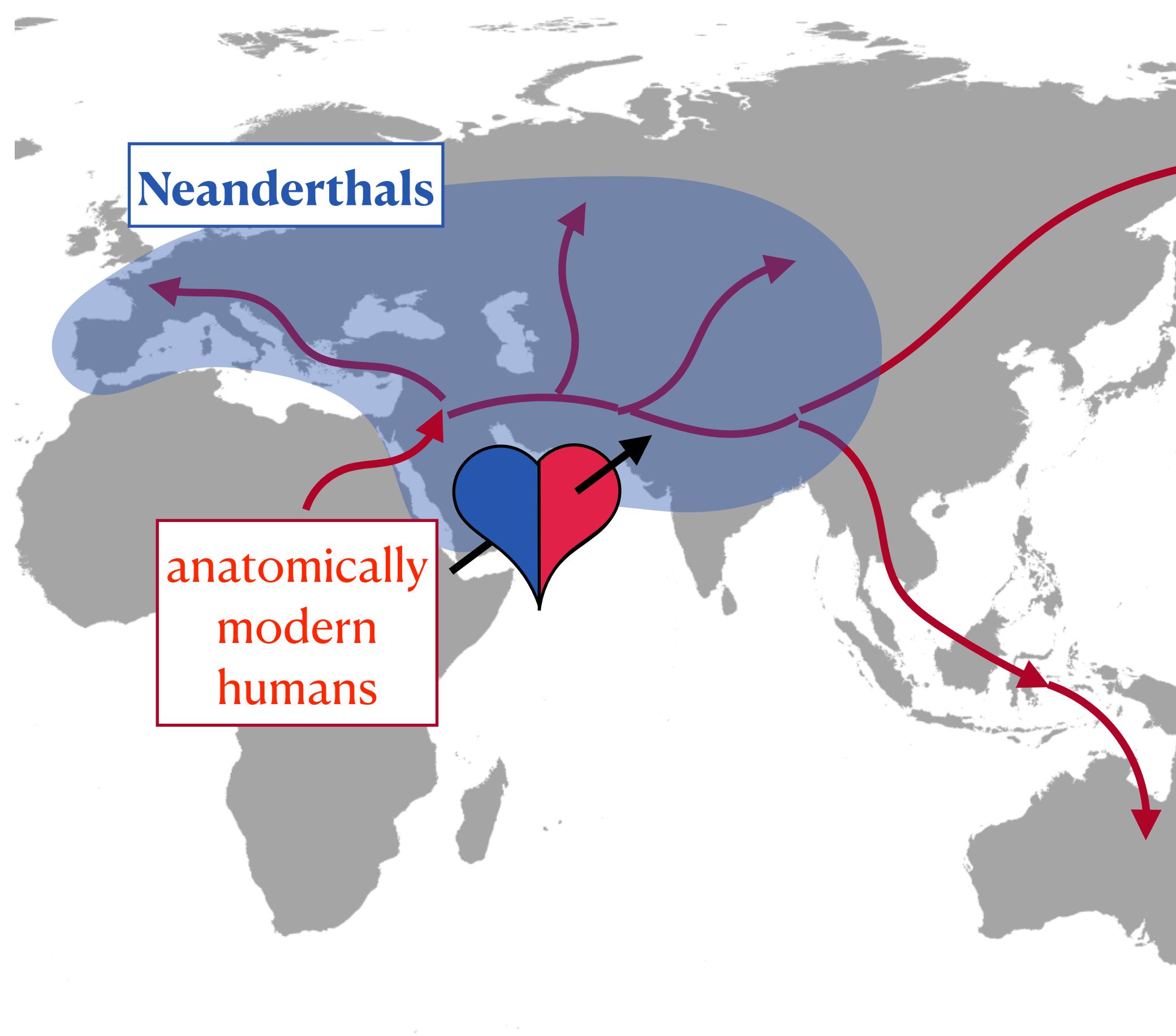
What happened in the few thousand years of overlap?



What happened in the few thousand years of overlap?



Introgression / gene flow / admixture?



<https://twitter.com/ijhublin/status/739866080764628993>

Morphological evidence?



**~40 thousand years old
remains of a modern human
Peștera cu Oase, Romania**

Neanderthal DNA? (ancient DNA, aDNA)



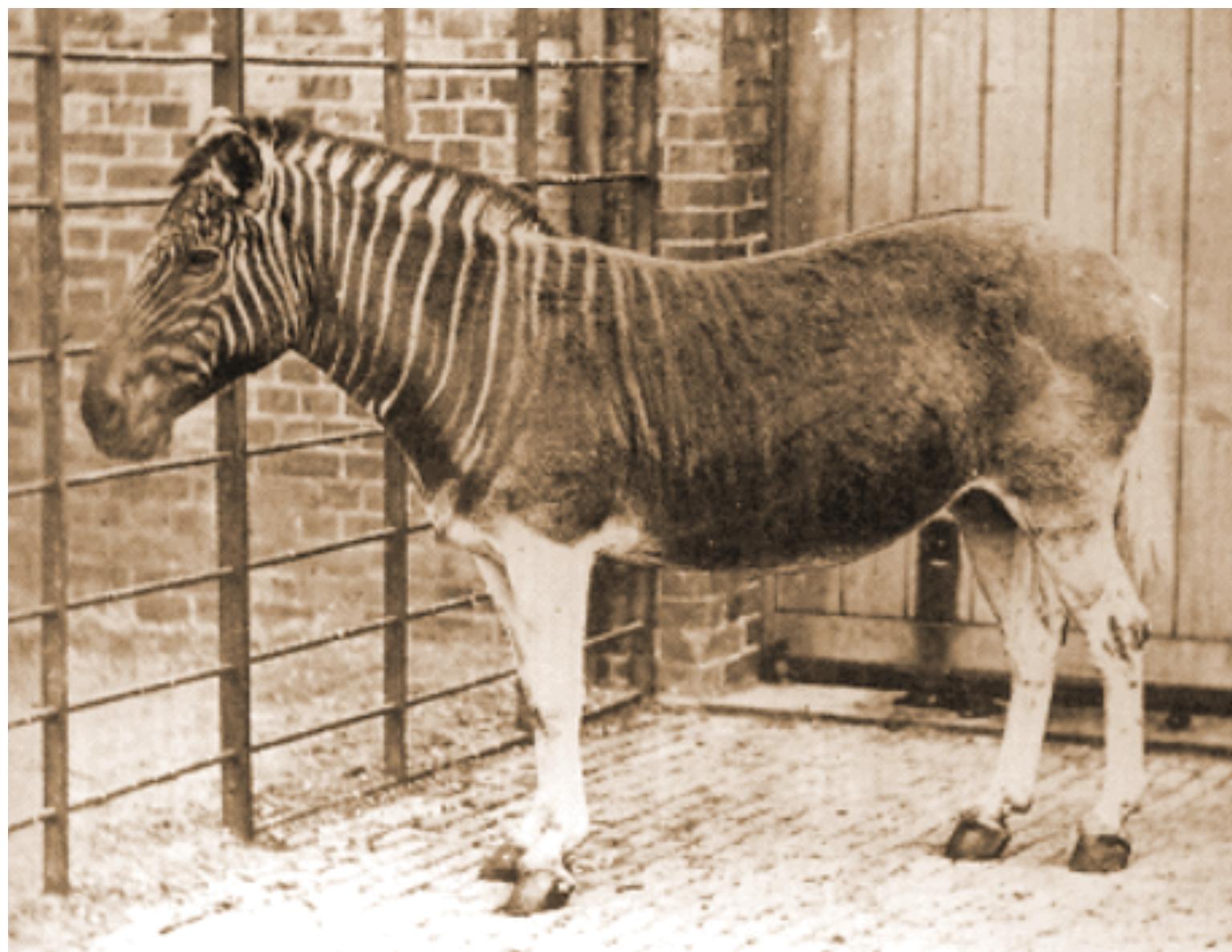
1984

DNA sequences from the quagga, an extinct member of the horse family

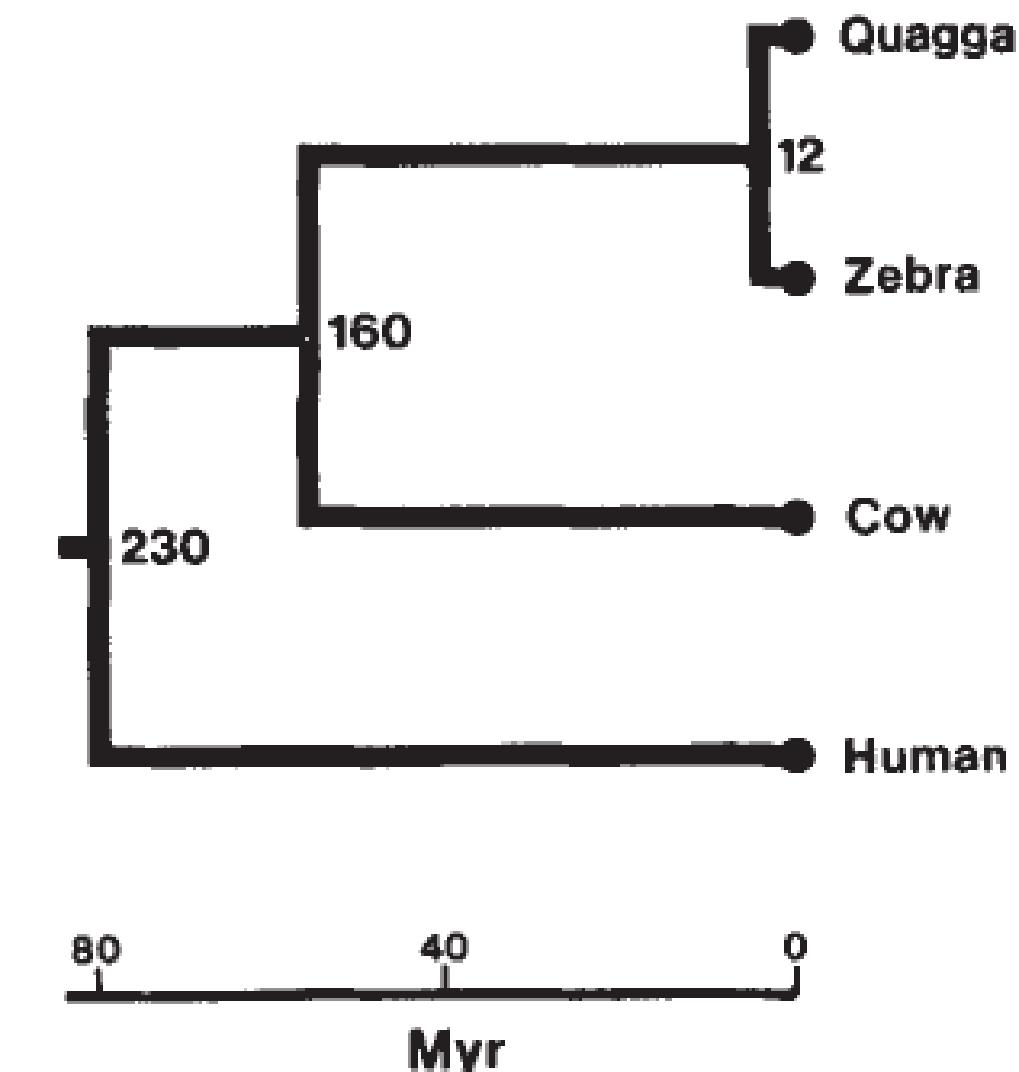
Russell Higuchi*, Barbara Bowman*, Mary Freiberger*,
Oliver A. Ryder† & Allan C. Wilson*

* Department of Biochemistry, University of California, Berkeley,
California 94720, USA

† Research Department, San Diego Zoo, San Diego,
California 92103, USA



- 150 years old tissue from a museum specimen
- 229 bp mitochondrial DNA



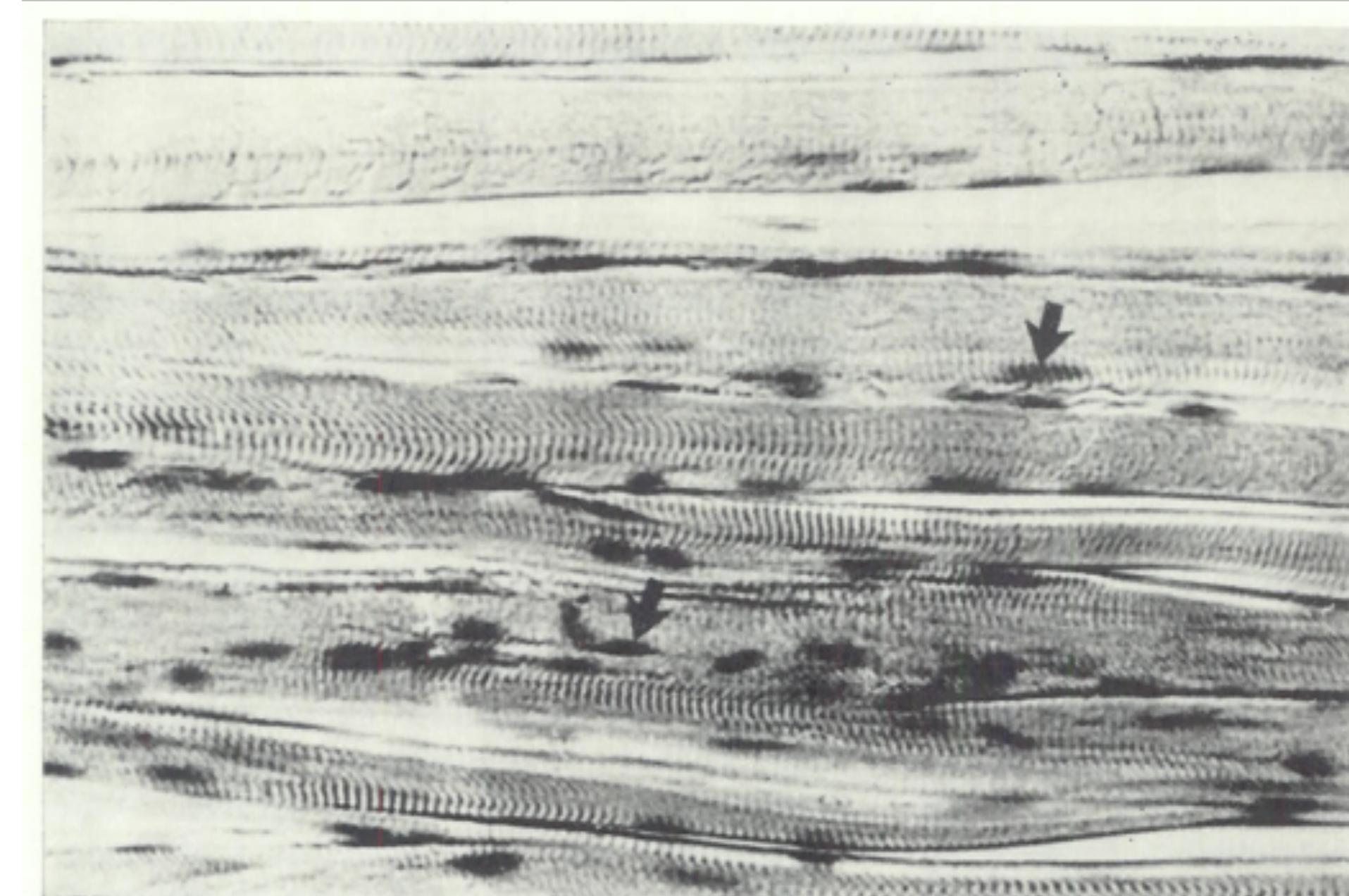
1985

Molecular cloning of Ancient Egyptian mummy DNA

Svante Pääbo

Department of Cell Research, The Wallenberg Laboratory,
University of Uppsala, Box 562, S-75122 Uppsala, Sweden and
Institute of Egyptology, Gustavianum, University of Uppsala,
S-75120 Uppsala, Sweden

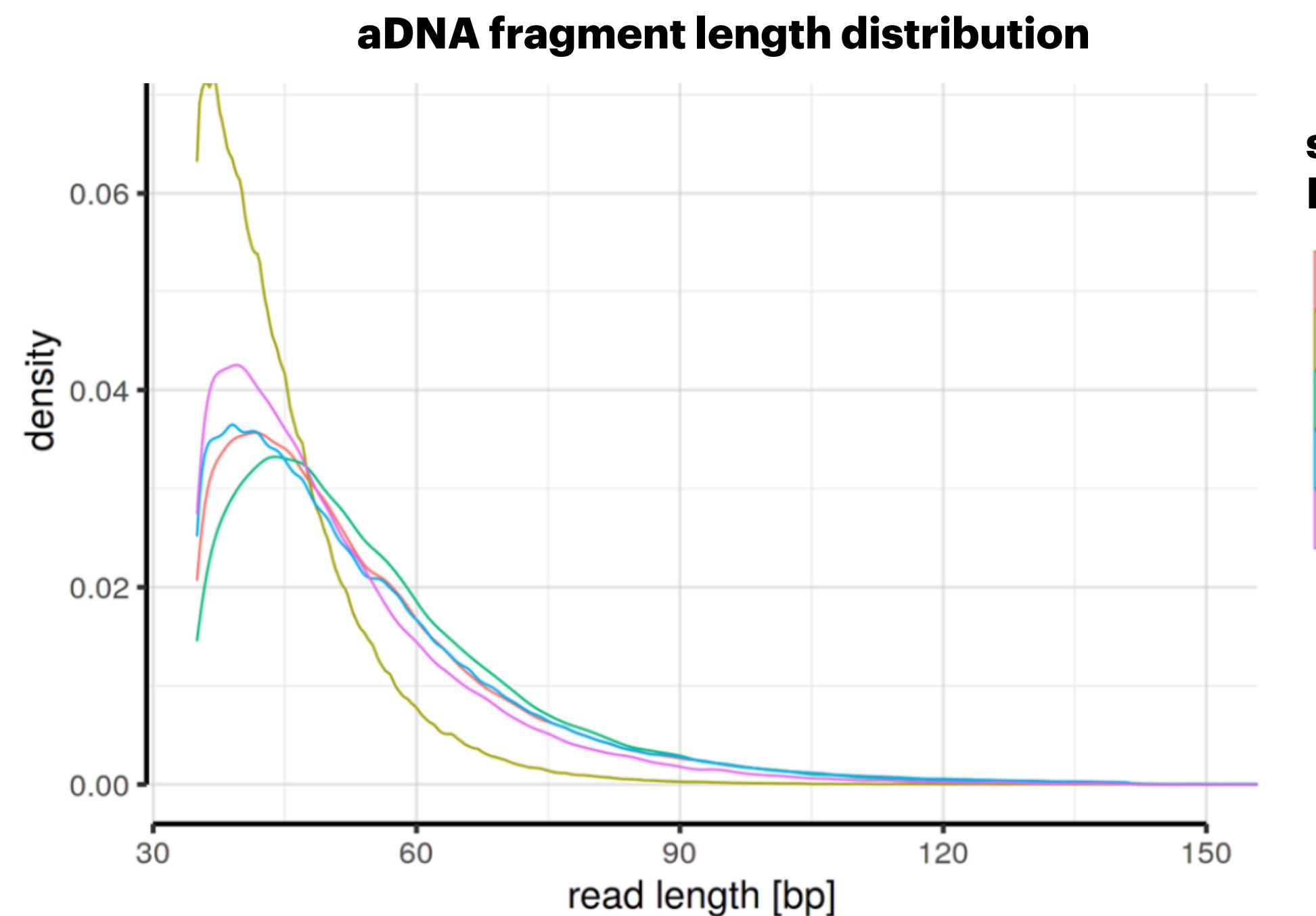
- ~2400 years old mummy
- 3400 bp nuclear DNA



Molecular characteristics of aDNA

Molecular characteristics of aDNA

- highly fragmented

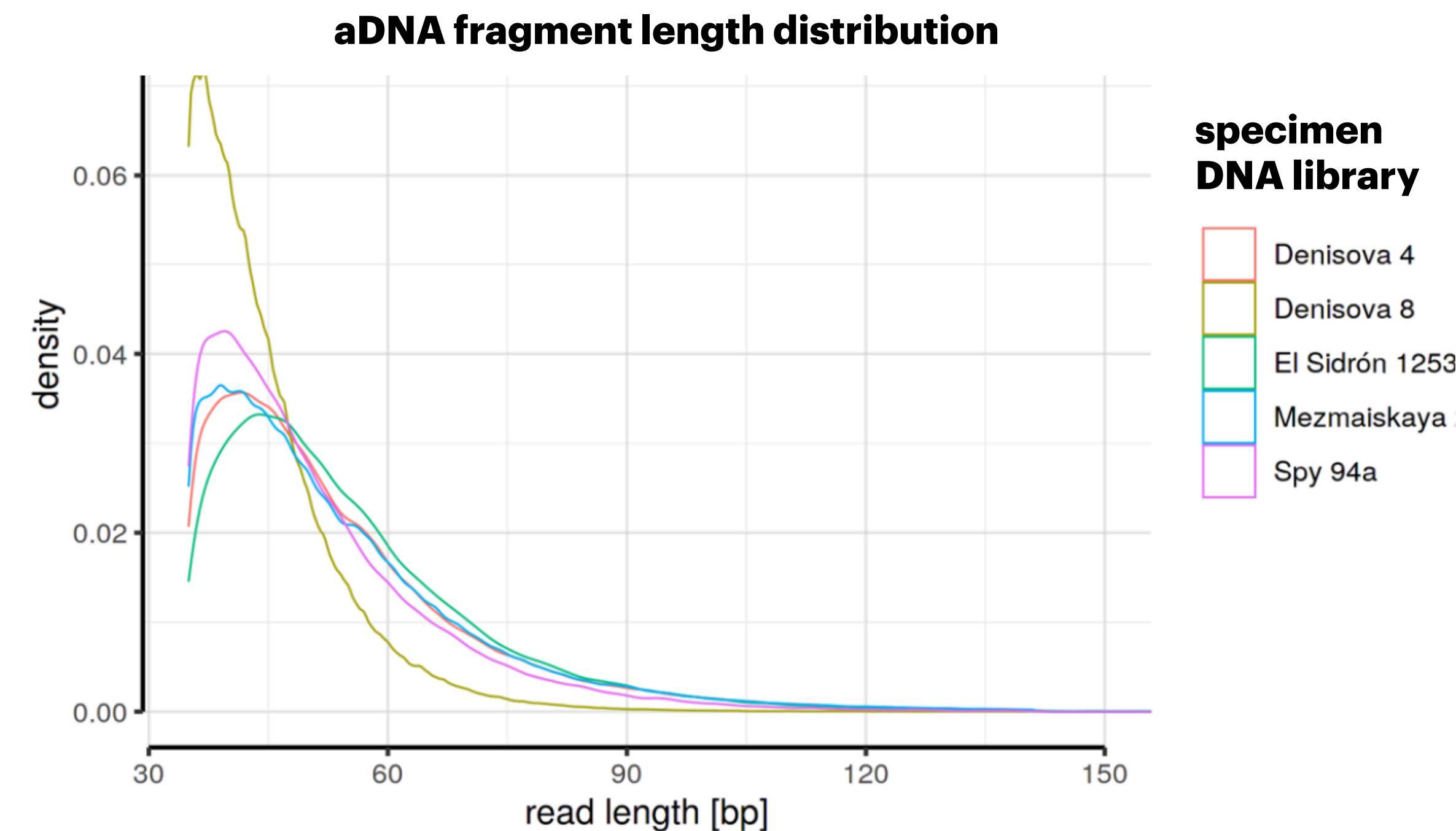
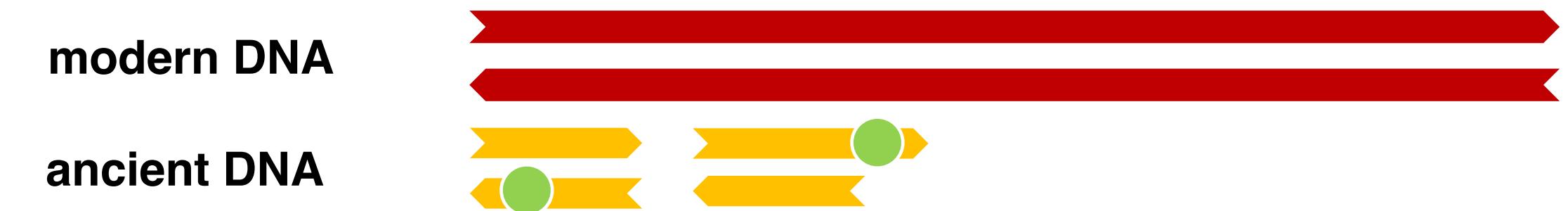
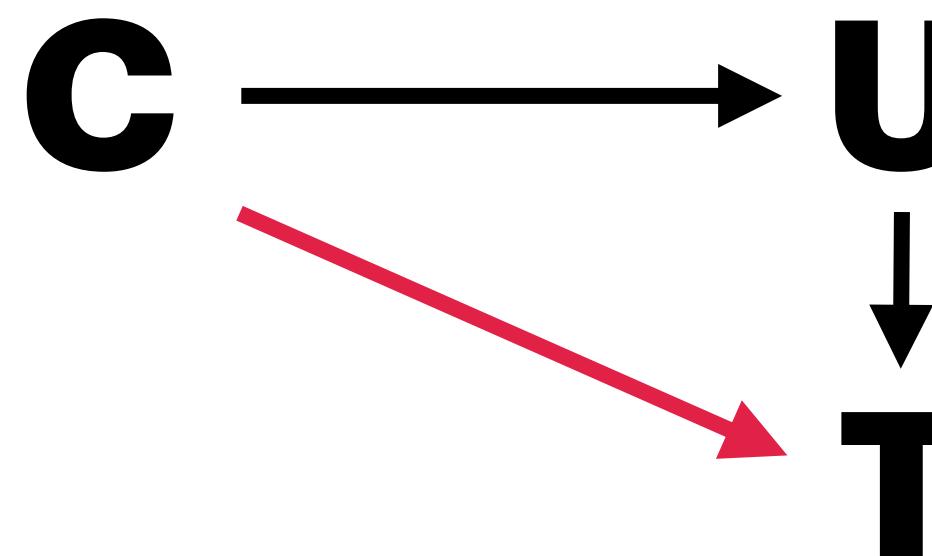
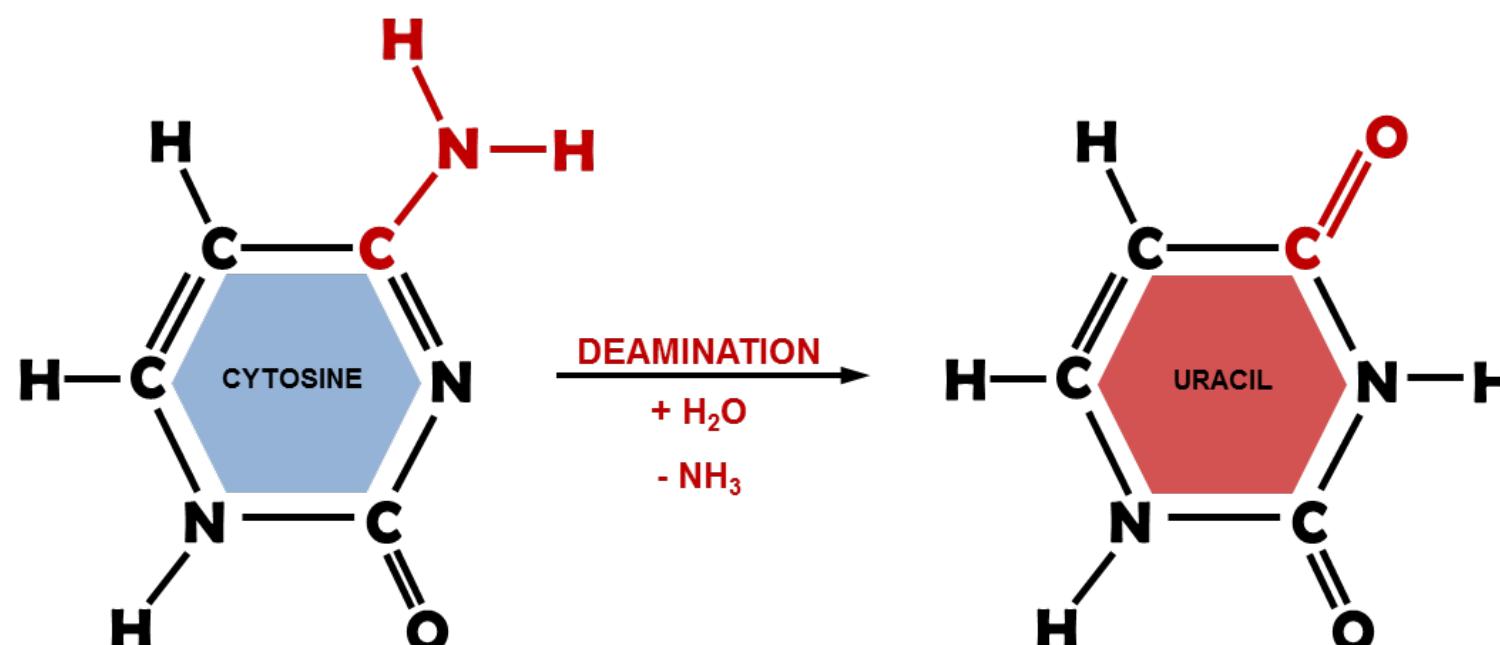


specimen DNA library

| |
|----------------|
| Denisova 4 |
| Denisova 8 |
| El Sidrón 1253 |
| Mezmaiskaya |
| Spy 94a |

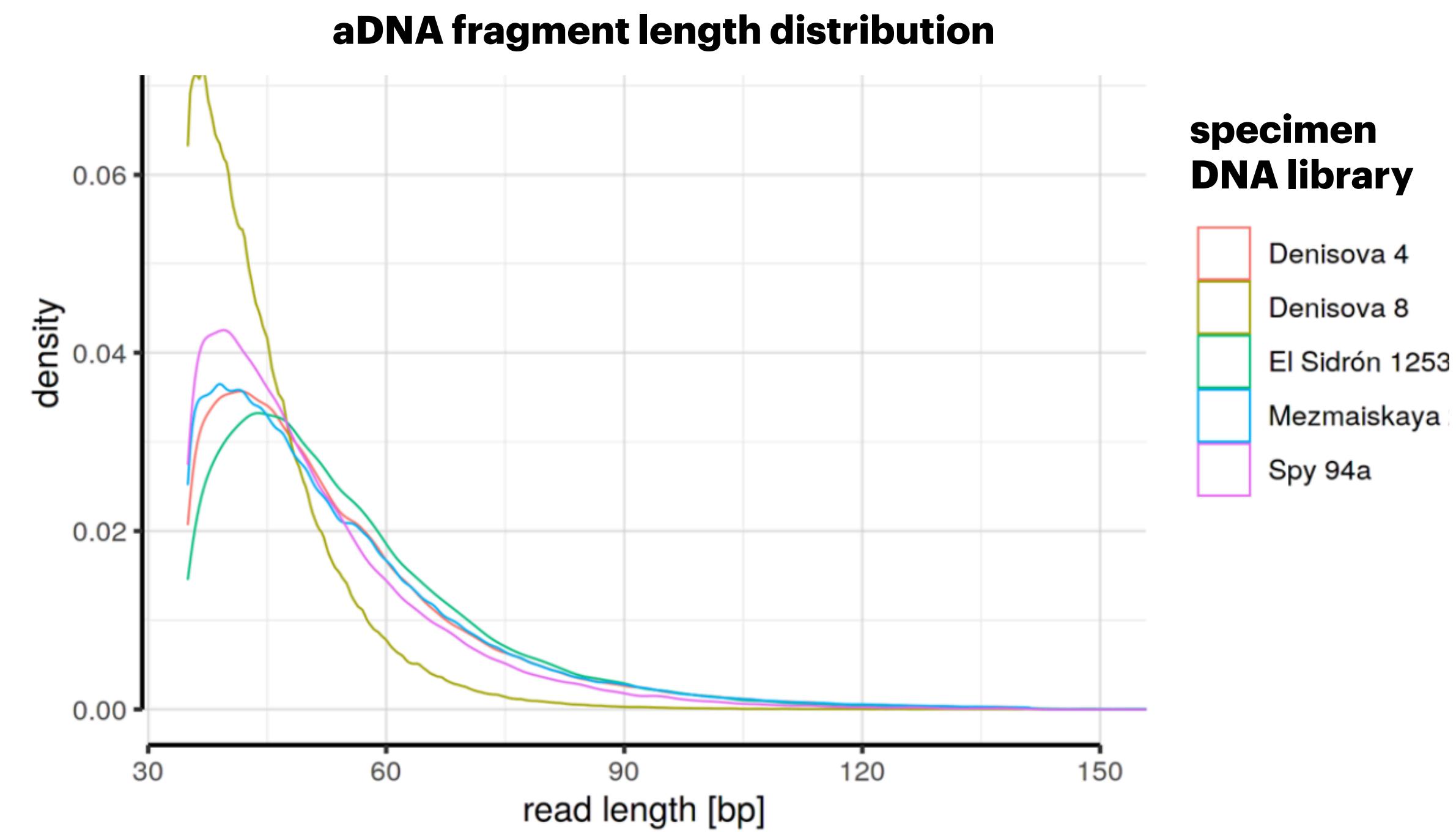
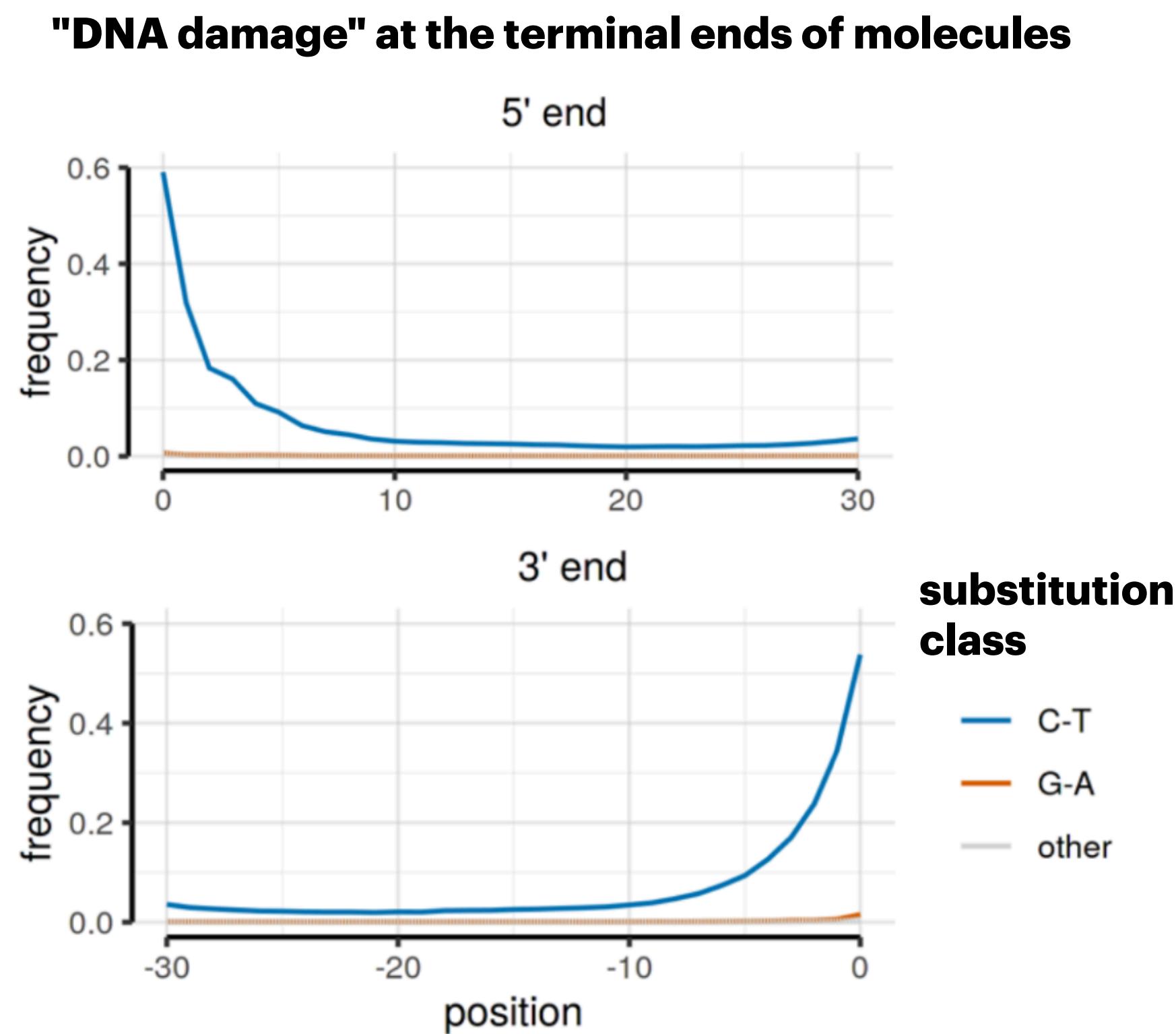
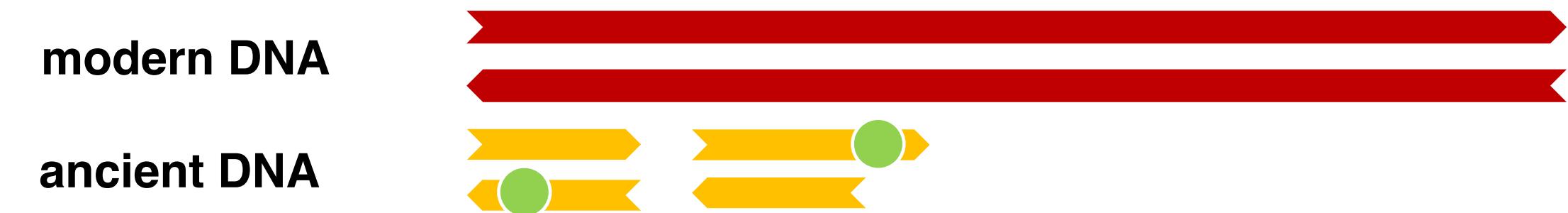
Molecular characteristics of aDNA

- highly fragmented
- post-mortem chemical modifications



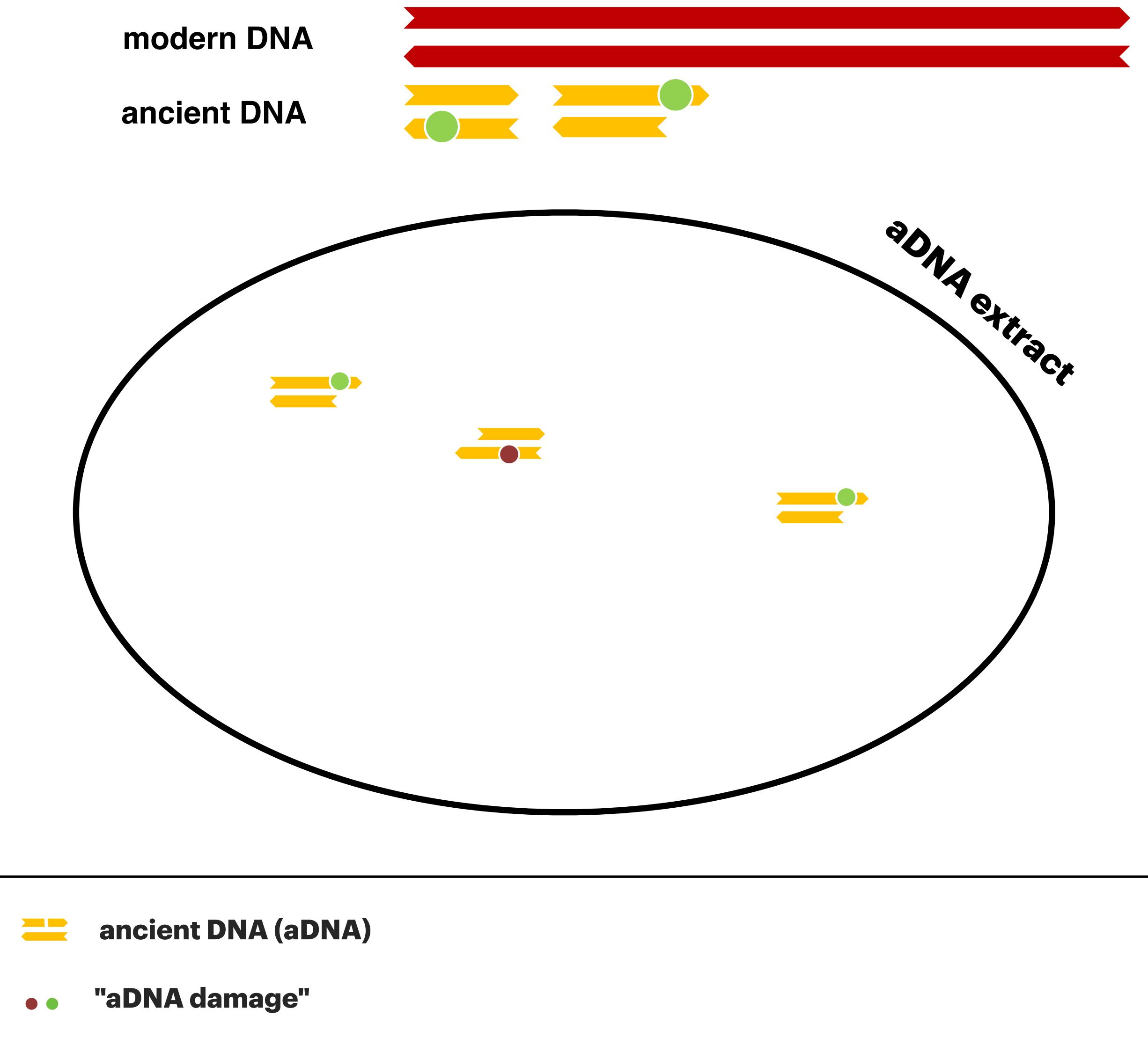
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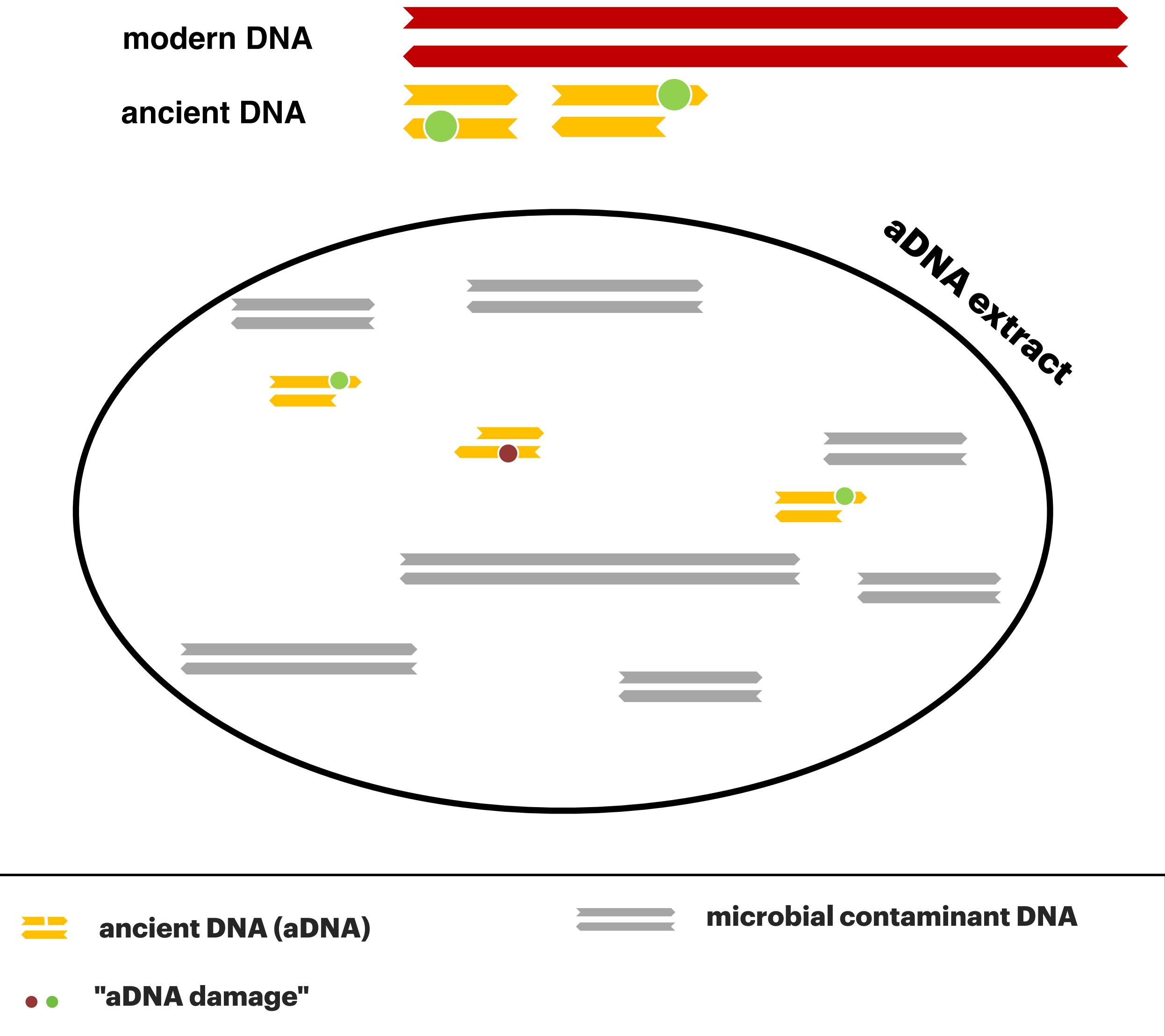
Molecular characteristics of aDNA

- highly fragmented
- post-mortem chemical modifications
- contamination:
 - microbial DNA
 - human DNA (excavation, museum, lab)



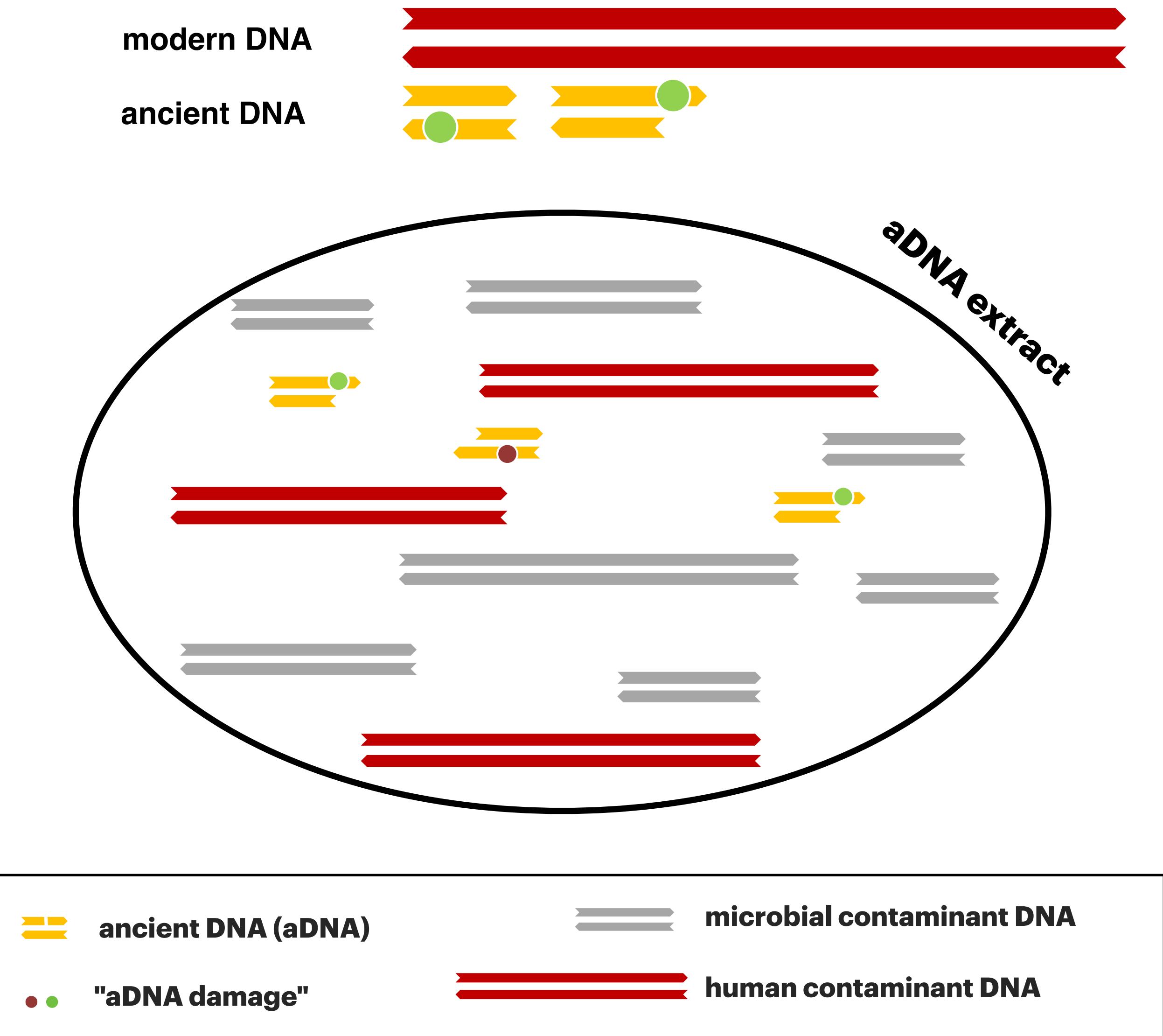
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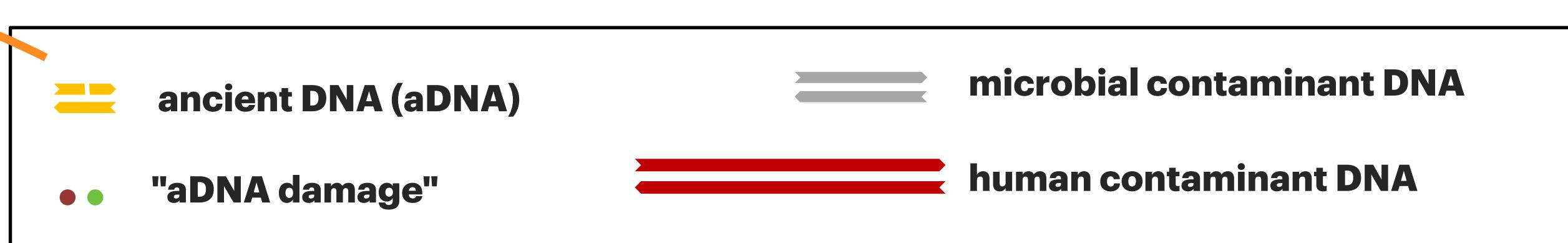
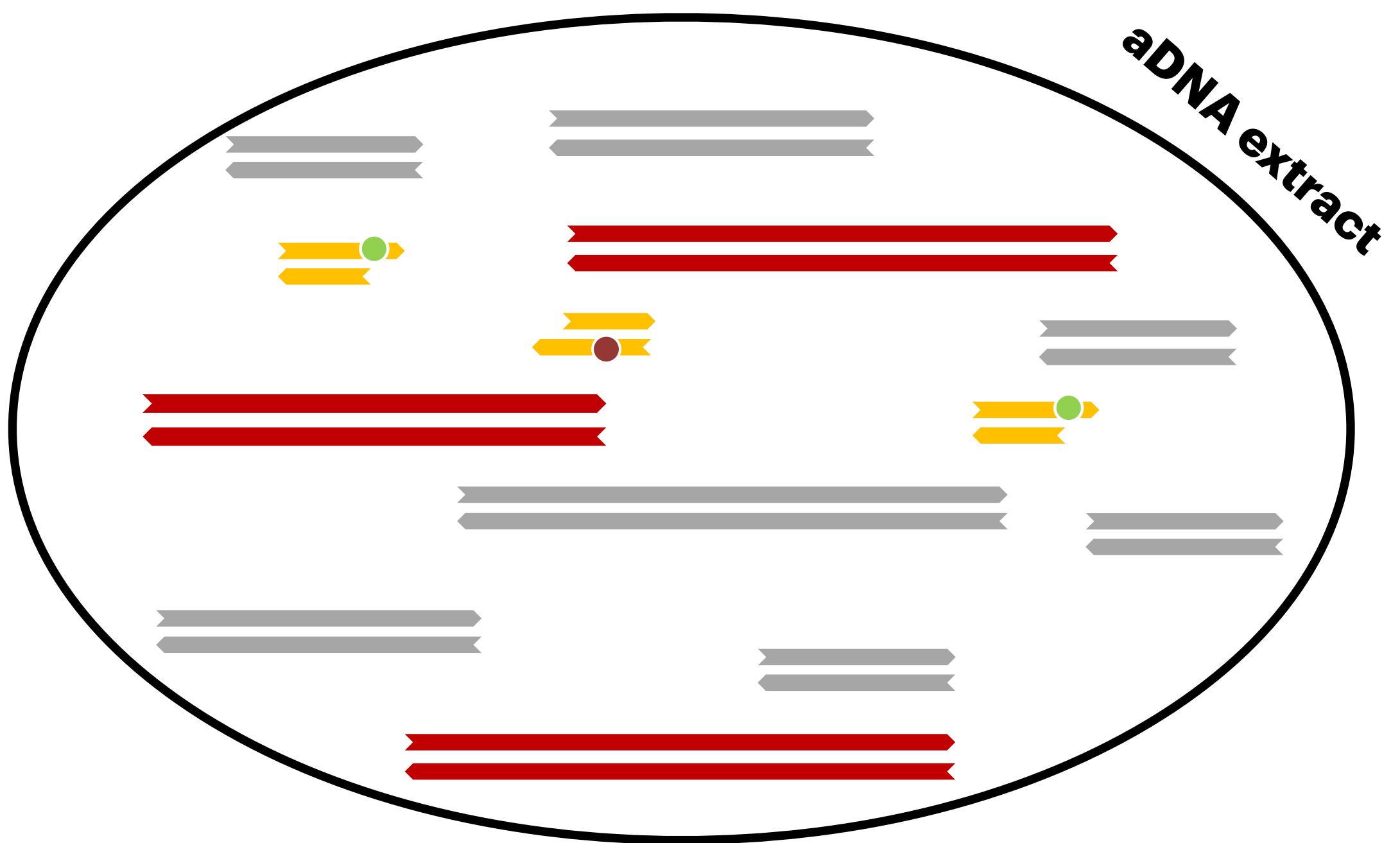
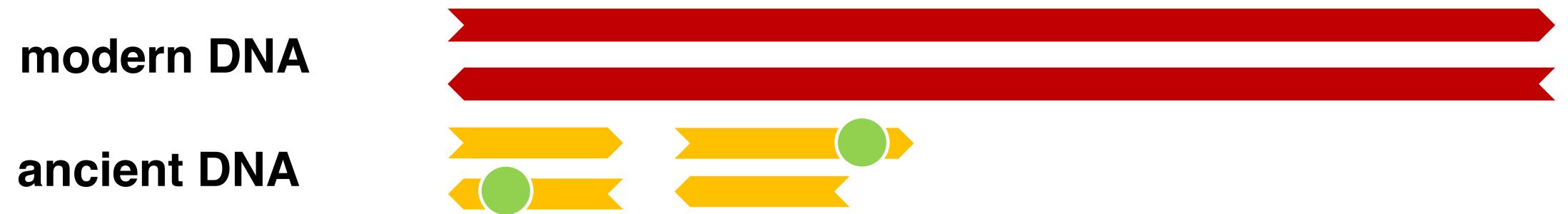


Molecular characteristics of aDNA

- highly fragmented
- post-mortem chemical modifications
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aDNA extract

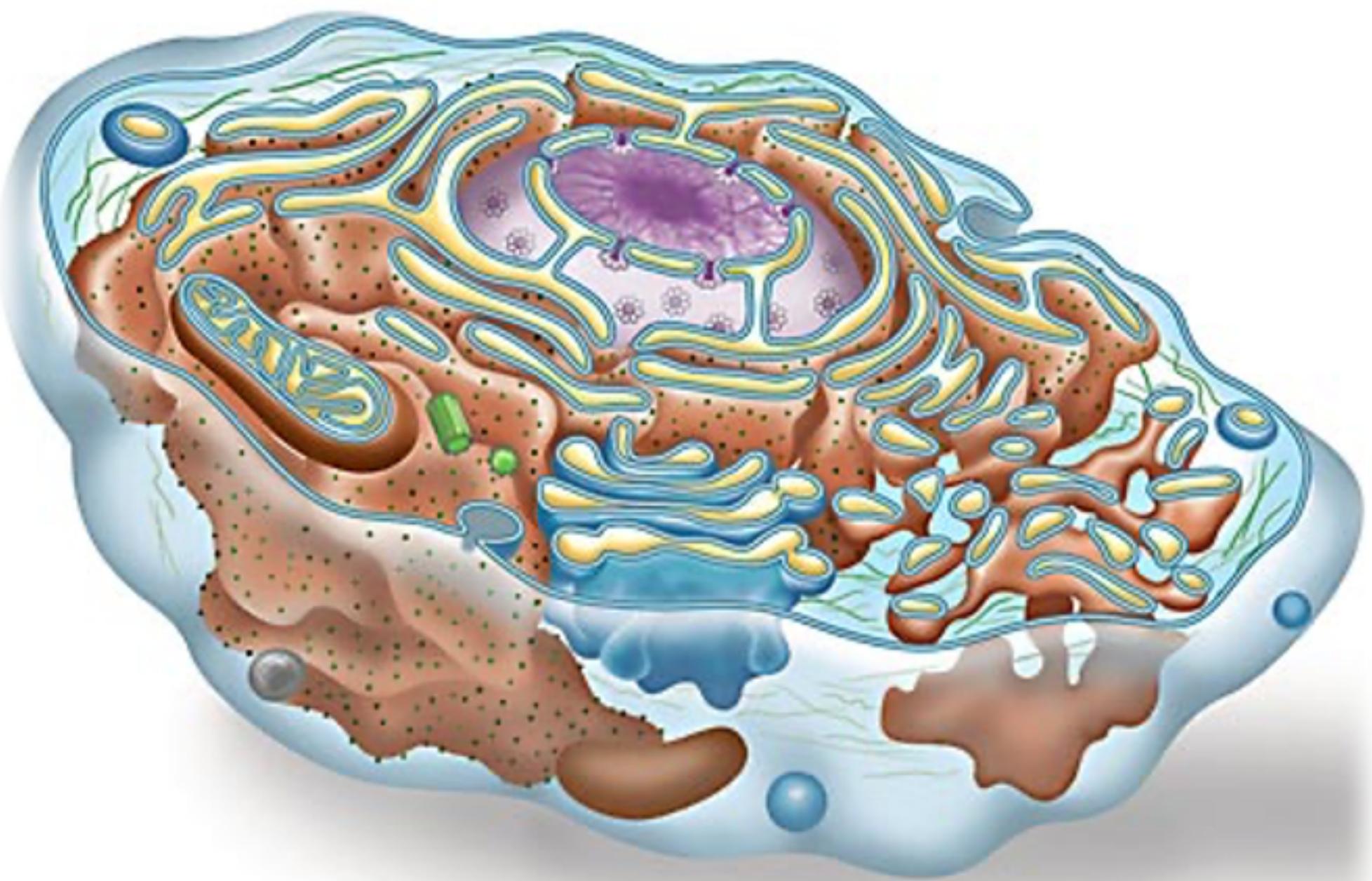
often < 1% endogenous DNA



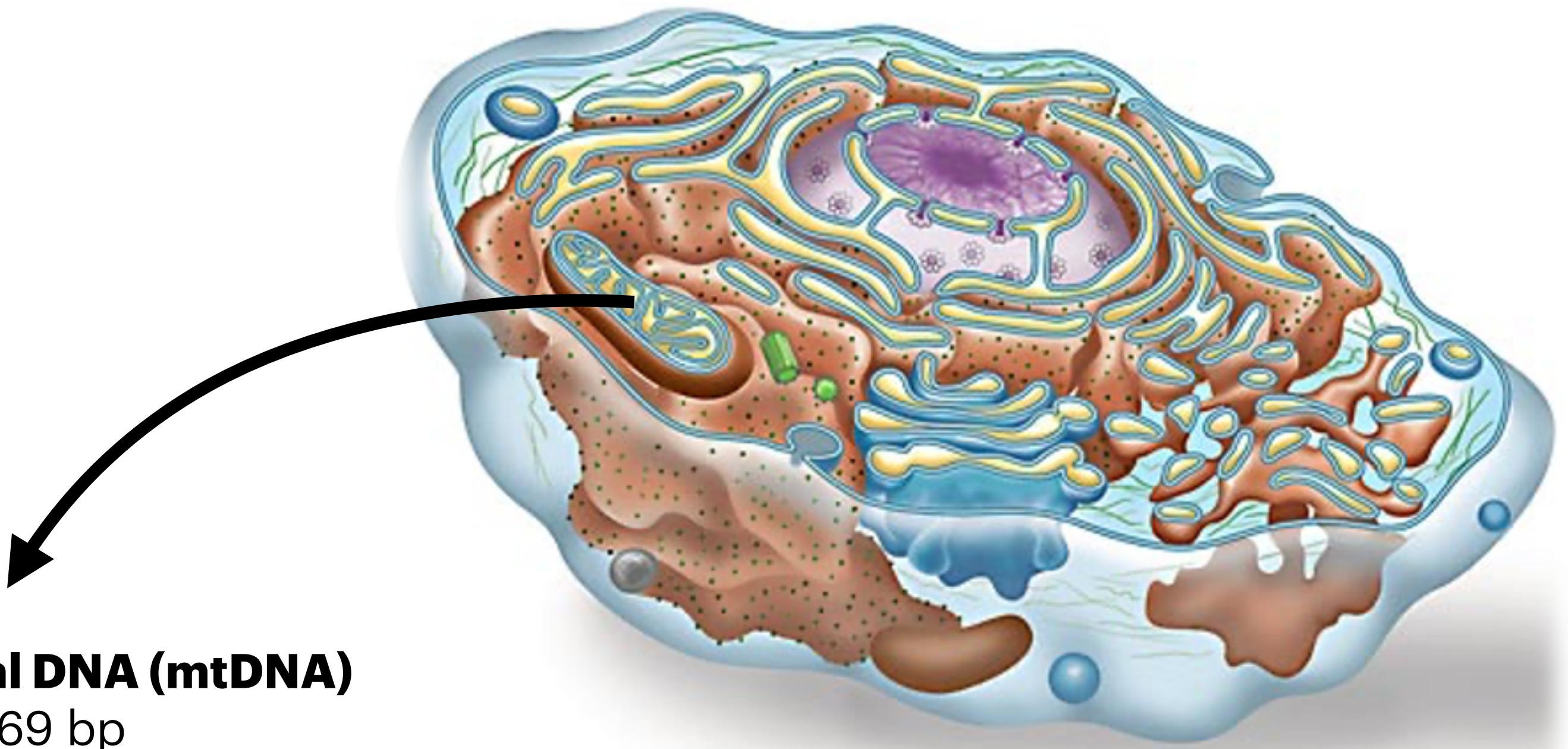
Neanderthal DNA?



Two sources of DNA



Two sources of DNA

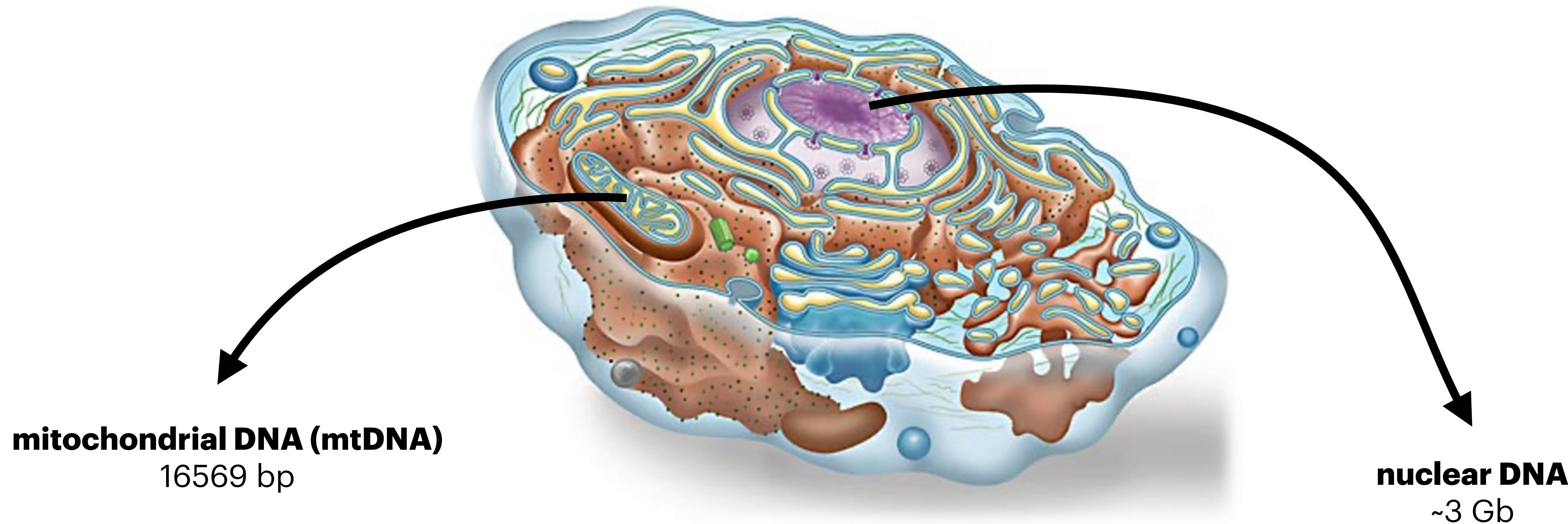


mitochondrial DNA (mtDNA)

16569 bp

- just one locus
- transmitted along maternal line
- fast mutation rate
- **thousands of copies in a cell**

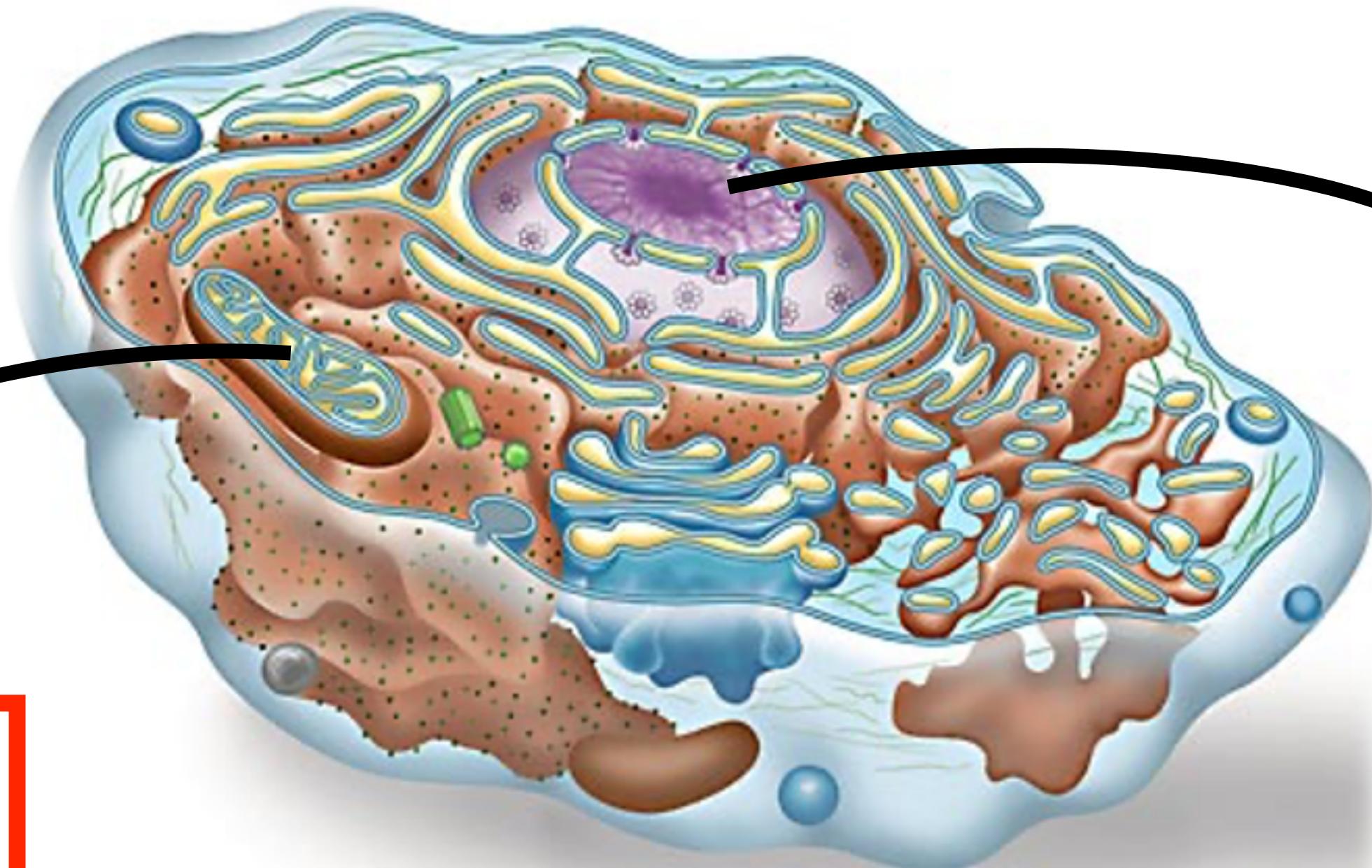
Two sources of DNA



- just one locus
- transmitted along maternal line
- fast mutation rate
- **thousands of copies in a cell**

- thousands of loci due to recombination
- complex mosaic of genetic history
- **two copies in a human cell**

Two sources of DNA



mitochondrial DNA (mtDNA)

16569 bp

- just one locus
- transmitted along maternal line
- fast mutation rate
- **thousands of copies in a cell**

nuclear DNA

~3 Gb

- thousands of loci due to recombination
- complex mosaic of genetic history
- **two copies in a human cell**

1997: Neanderthal mitochondrial DNA!

Cell, Vol. 90, 19–30, July 11, 1997, Copyright ©1997 by Cell Press

Neandertal DNA Sequences and the Origin of Modern Humans

Matthias Krings,* Anne Stone,† Ralf W. Schmitz,‡
Heike Krainitzki,§ Mark Stoneking,† and Svante Pääbo*



| | refseq | ACAGCAATCAGCCCTCAACTATCACACATCAACTGCAACTCCAAAGCCACCCCT-CACCCACTAGGGATACCAACAAACCTAACAGTACATAGTACATAAACCCATTACCGTACATAGCACATTACAGTC* |
|-------------------------------|--------------------------|---|
| A12.1 | L16,269 | T...G...T.....A.....A.G...T.A.....T.....T.G.....G.....C.....T..... |
| A12.2 | | T...G...T.....A.....A.G...T.A.....T.....T.G.....G.....C.....T..... |
| A12.3 | | T...G...T.....A.....A.G...T.A.....T.....T.G.....G.....CG.....T..... |
| A12.4 | | T...G...T.....A.....A.G...T.A.....T.....T.G.....G.....C.....T..... |
| A12.5 | | T...G...T.....A.....A.G...T.A.....T.....T.G.....G.....C.....T..... |
| A12.6 | | T...G...T.....A.....A.G...T.A.....T.....T.G.....G.....C.....T..... |
| A12.7 | | T...G...T.....A.....A.G...T.A.....T.....T.G.....G.....C.....T..... |
| A12.8 | | T...G...T.....A.....A.G...T.A.....T.....T.G.....G.....C.....T..... |
| A12.9 | | T...G...T.....A.....A.G...T.A.....T.....T.G.....G.....C.....T..... |
| A12.10 | | T...G...T.....A.....A.G...T.A.....T.....T.G.....G.....C.....T..... |
| A12.11 | | T...G...T.....A.....A.G...T.A.....T.....T.G.....G.....C.....T..... |
| A12.12 | | T...G...T.....A.....A.G...T.A.....T.....T.G.....G.....C.....T..... |
| A12.13 | | T...G...T.....A.....A.G...T.A.....T.....T.G.....G.....C.....T..... |
| B13.1 | CACACATCAACTGCAACTCCAA | A.G...T.A.....T.....G.....G.....C.....T..... |
| B13.2 | L16,254 | A.G...T.A.....T.....G.....G.....C.....T..... |
| B13.3 | | A.G...T.A.....T.....G.....G.....C.....T..... |
| B13.4 | | A.G...T.A.....T.....G.....G.....C.....T..... |
| B13.5 | | A.G...T.A.....T.....G.....G.....C.....T..... |
| B13.6 | | A.G...T.A.....T.....G.....G.....C.....T..... |
| B13.7 | | A.G...T.A.....T.....G.....G.....C.....T..... |
| B13.8 | | A.G...T.A.....T.....G.....G.....C.....T..... |
| B14.1 | ACTACAACTCCAAAGRCGCCCTTA |G.....C.....C.....T.....T..... |
| B14.2 | ML16,263/264 |T.....G.....C.....C.....T..... |
| B14.3 | |T.....G.....C.....C.....T..... |
| B14.4 | |T.....G.....C.....G.....T..... |
| B14.5 | |T.....G.....C.....C.....T..... |
| B14.6 | |T.....G.....C.....C.....T..... |
| B14.7 | |T.....G.....C.....C.....T..... |
| B14.8 | |T.....G.....C.....C.....T..... |
| B14.9 | |T.....G.....C.....C.....T..... |
| B14.10 | |T.....G.....C.....C.....TT..... |
| B14.11 | |T.....G.....C.....C.....T..... |
| B14.12 | |T.....G.....C.....C.....T..... |
| B14.13 | |T.....G.....C.....C.....T..... |
| A15.1 | | CGTACATAGCACATTACAGT |
| A15.2 | | L16,347 |
| A15.3 | |C.....C.....T.....T..... |
| A15.4 | |C.....C.....T.....T..... |
| A15.5 | |C.....C.....T.....T..... |
| A15.6 | |C.....C.....T.....T..... |
| A15.7 | |C.....C.....AA.....T..... |
| A15.8 | |C.....C.....AA.....T..... |
| A15.9 | |C.....C.....AA.....T..... |
| A15.10 | |C.....C.....AA.....T..... |
| Neandertal (16,210-16,400) | |T.....G...T.....A.....A.G...T.A.....T.....G.....C.....T..... |

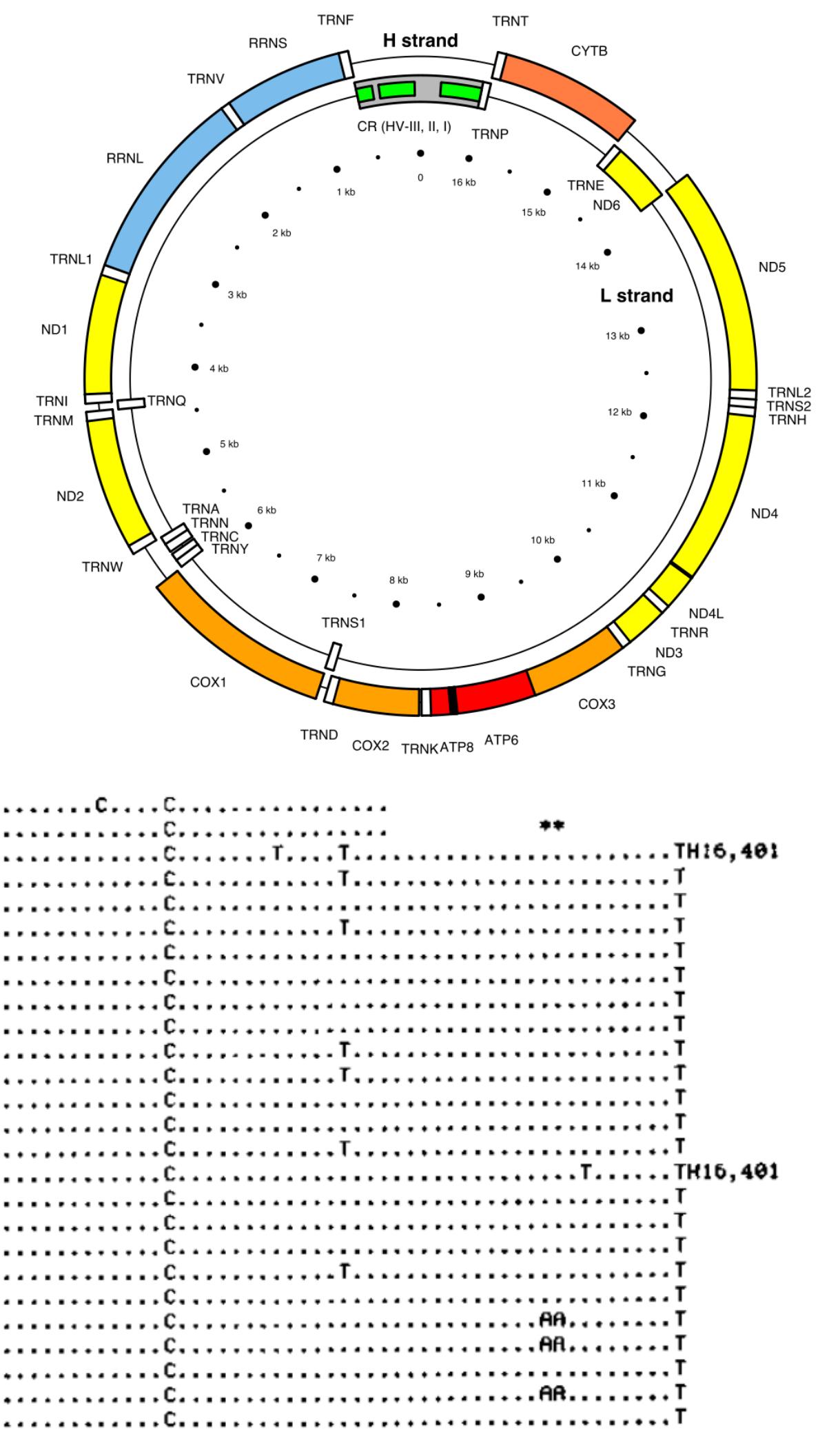
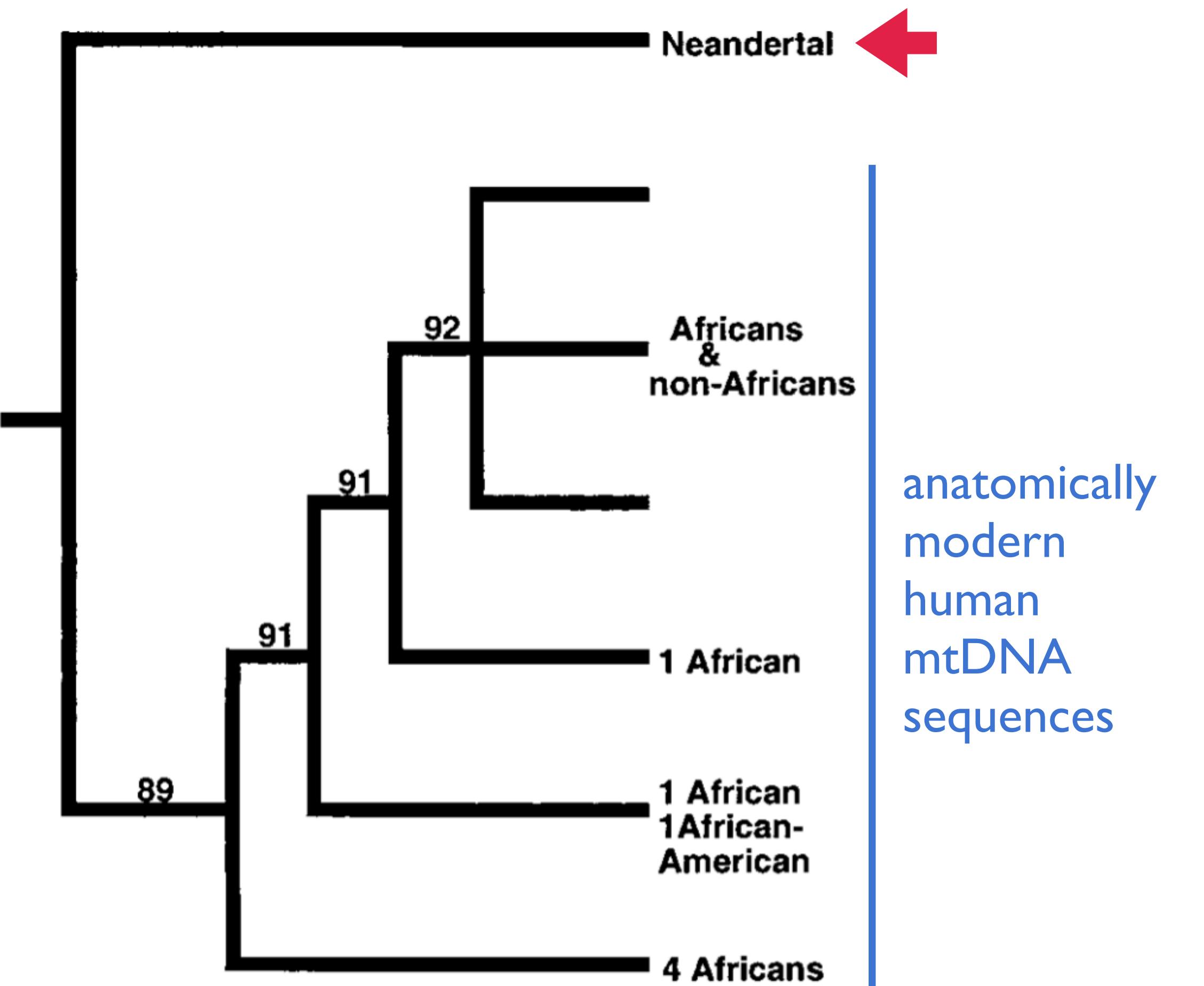
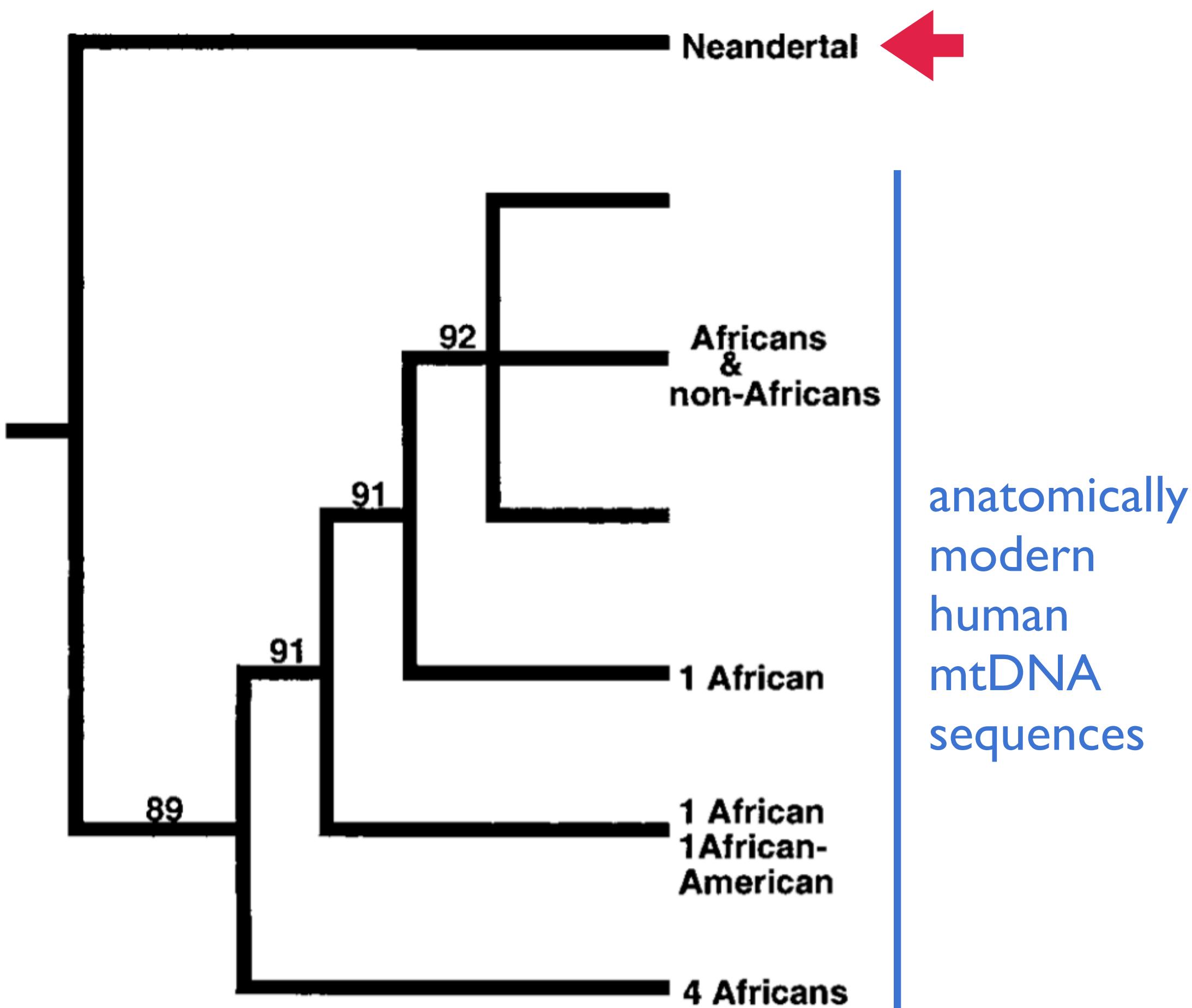


Figure 5. The DNA Sequences of Clones Used to Infer the Sequence of the Hypervariable Region I of the Neandertal Individual



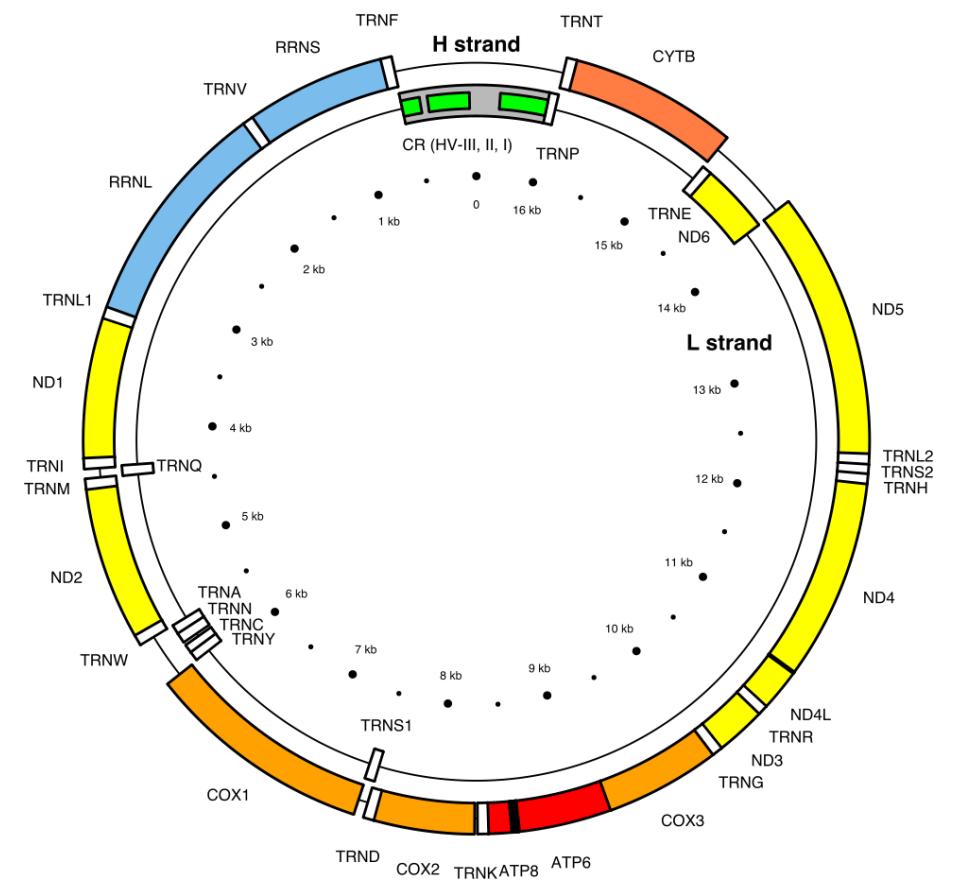
anatomically
modern
human
mtDNA
sequences

No introgression?

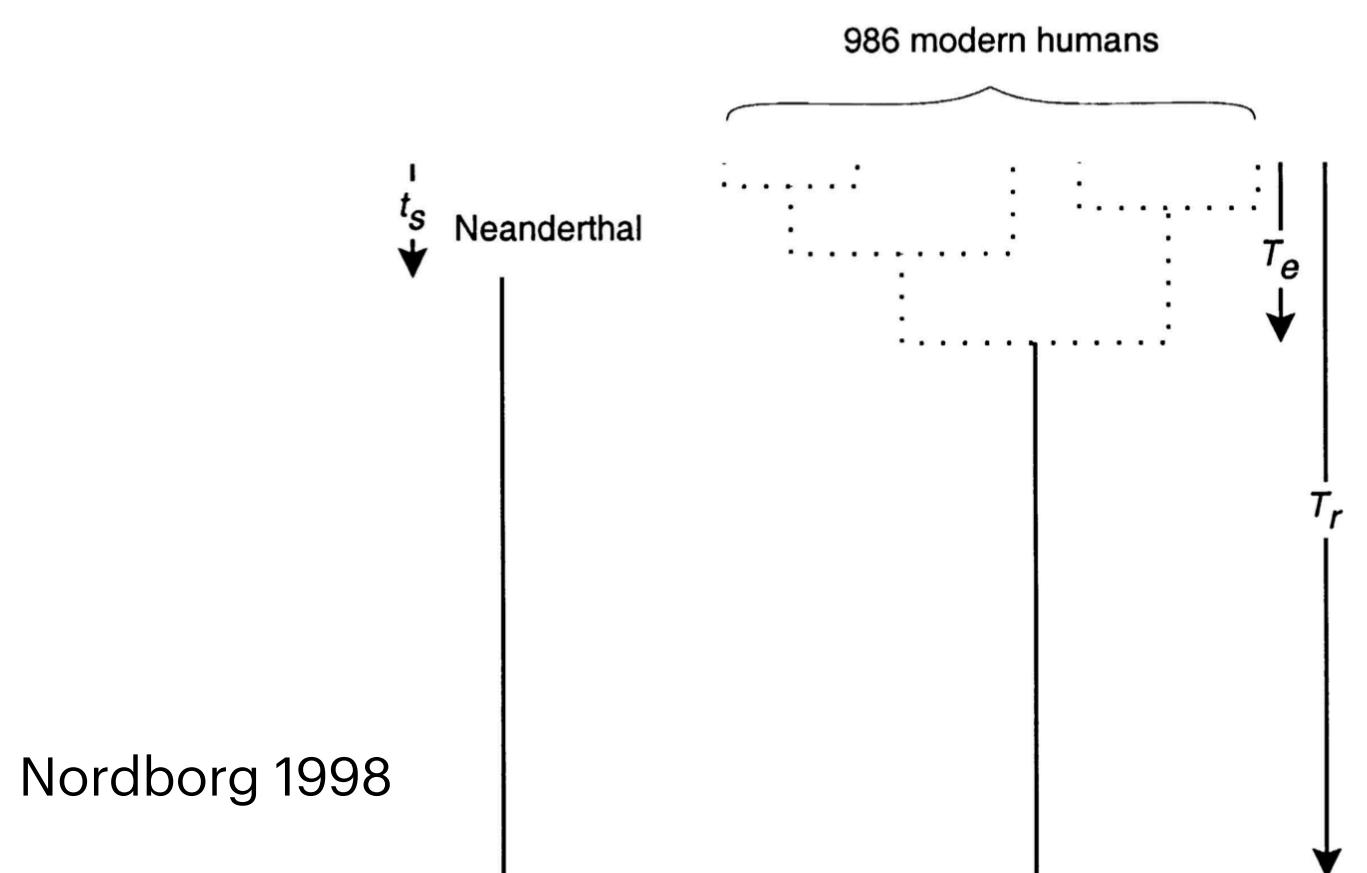


When the Neandertal mtDNA sequence is used to root a neighbor joining tree of modern human mtDNA sequences (Figure 7a), the first three branches consist exclusively of African sequences. The Neandertal mtDNA sequence thus supports a scenario in which modern humans arose recently in Africa as a distinct species and replaced Neandertals with little or no interbreeding.

mitochondrial DNA



16 kb

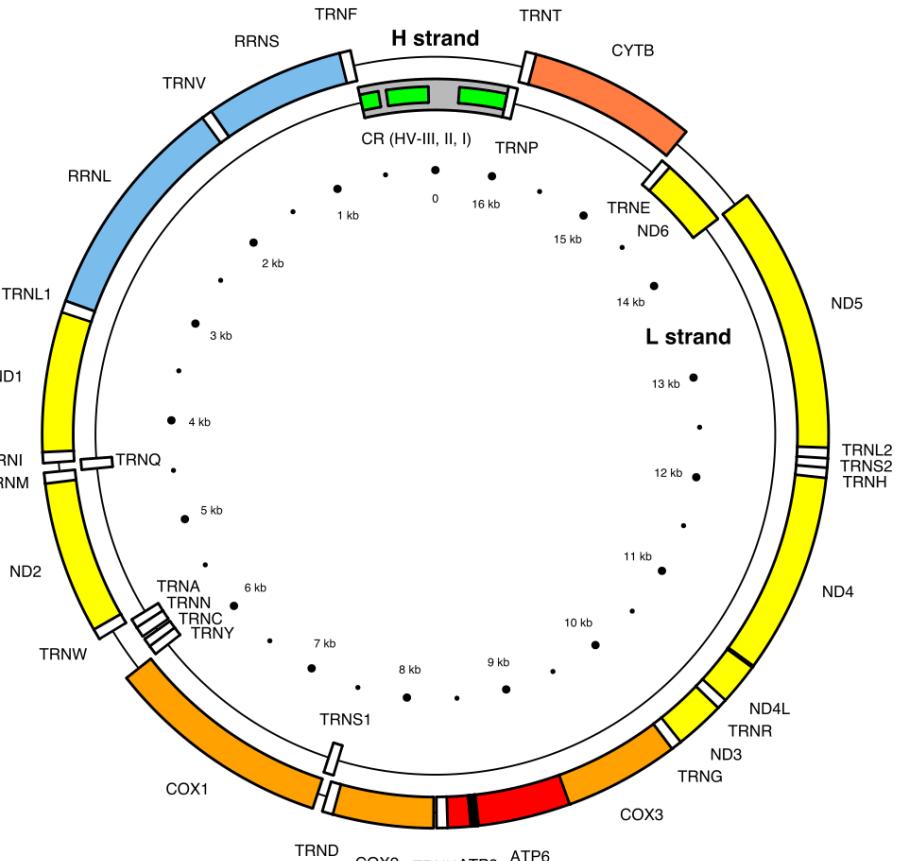


one mtDNA phylogenetic tree
(maternal history)

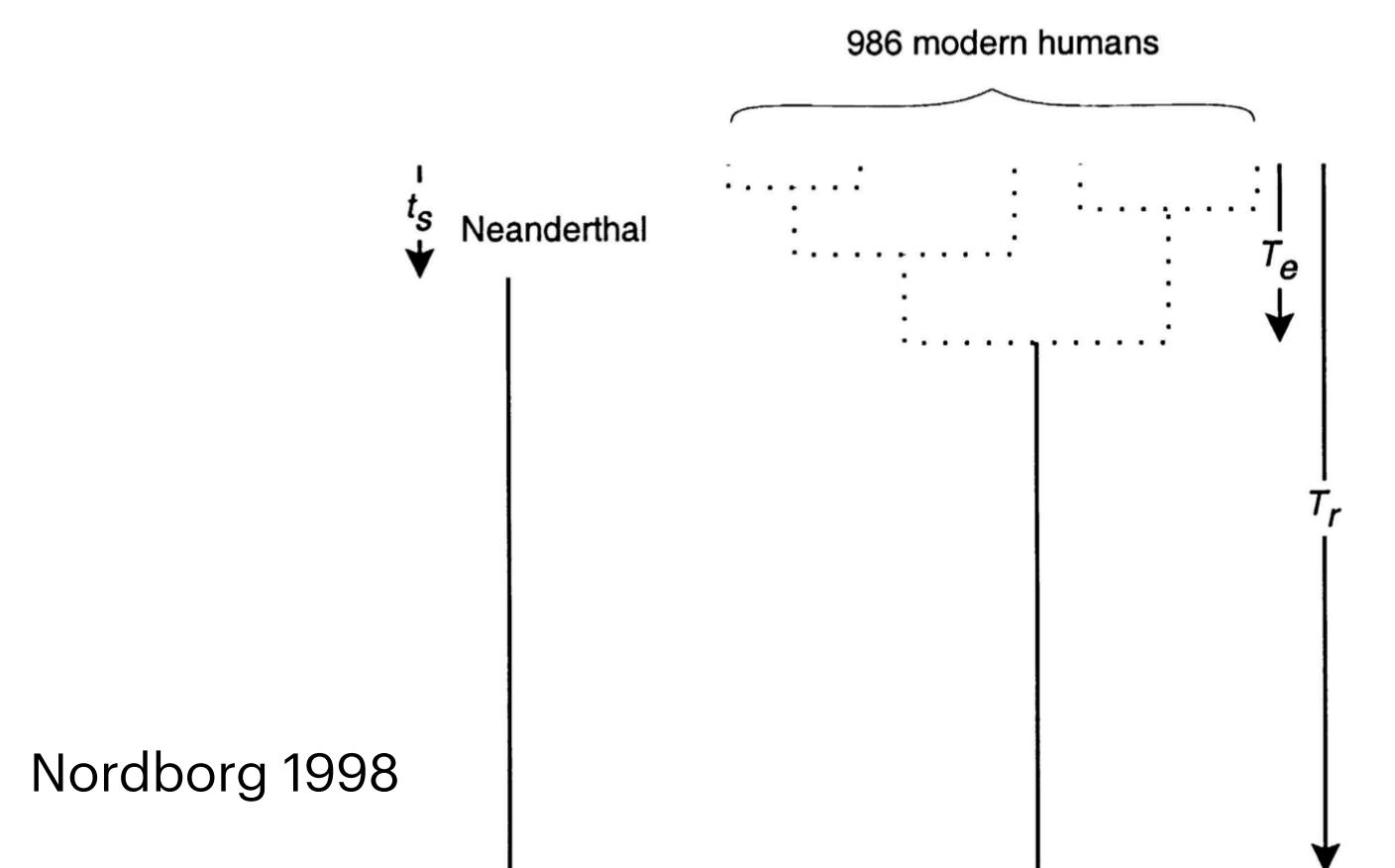
mitochondrial DNA

vs

nuclear DNA



3 Gb



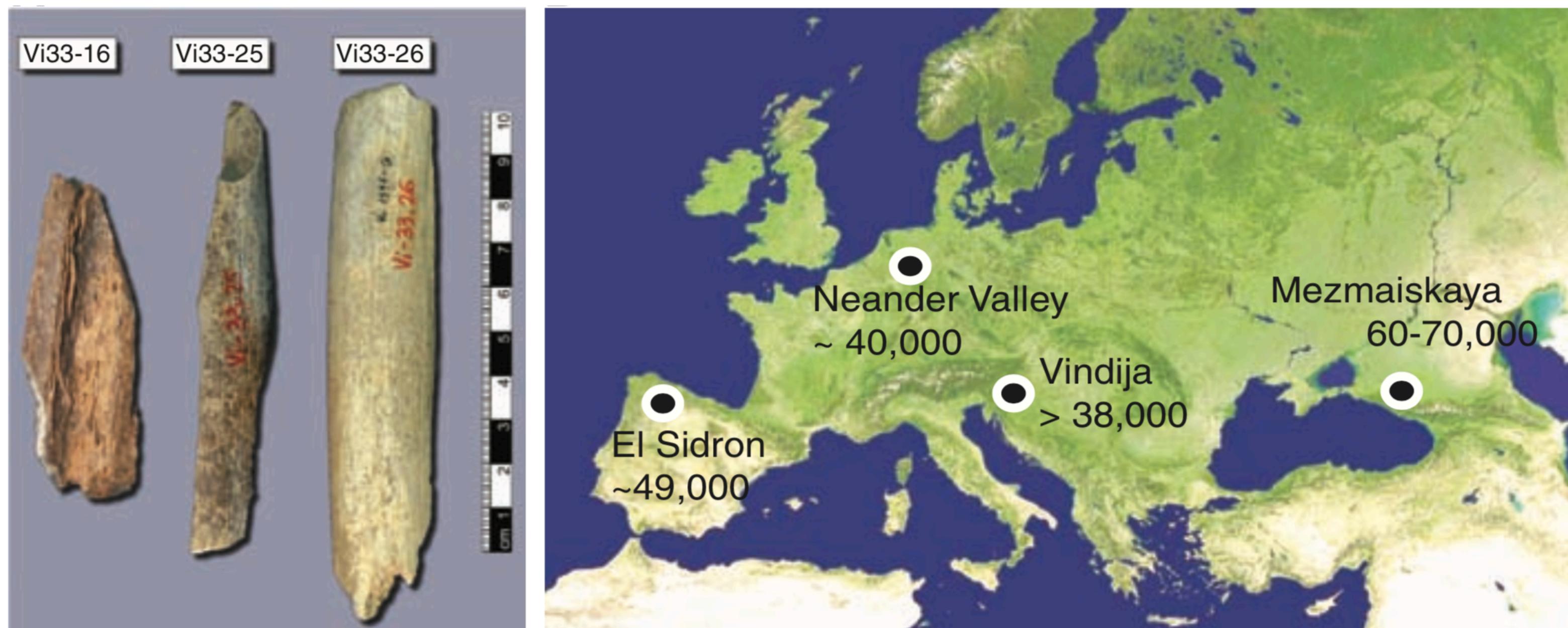
one mtDNA phylogenetic tree
(maternal history)

thousands of trees along the nuclear genome
(result of recombination)



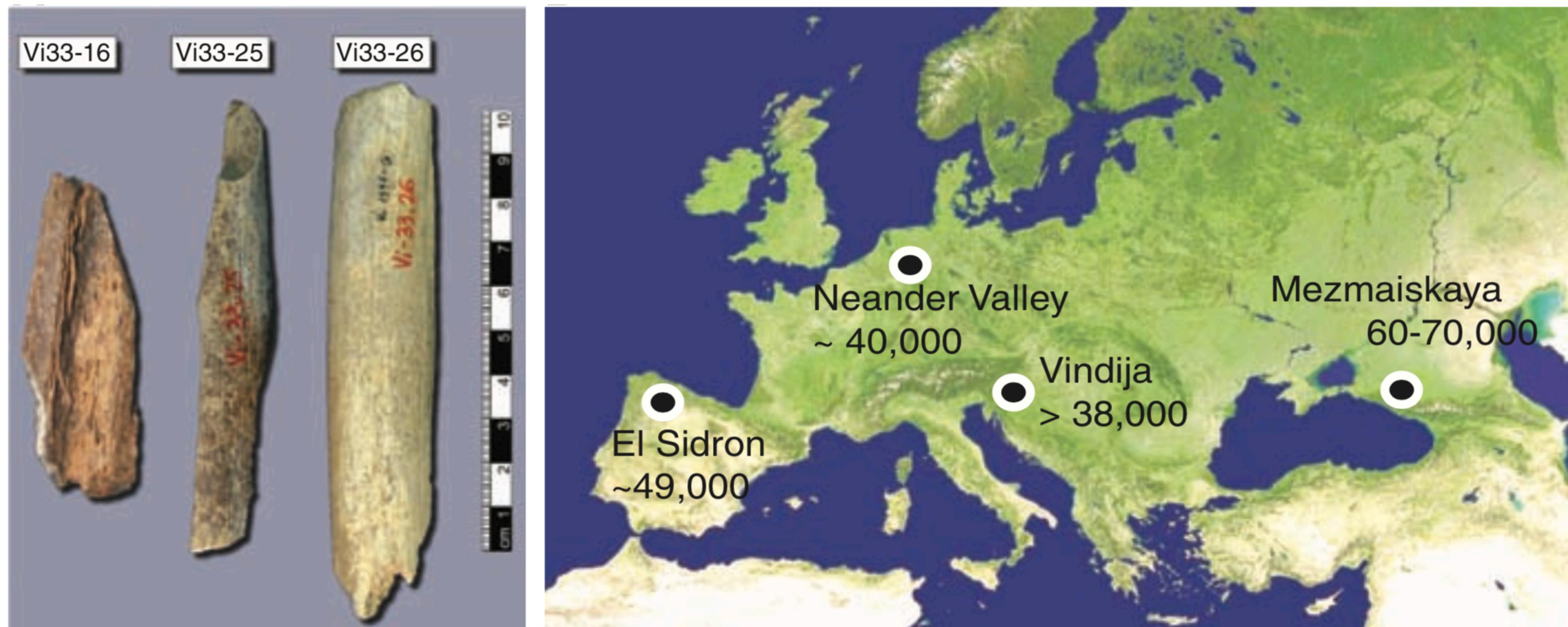
"Neanderthal genome consortium" – 2010, Leipzig, Germany

2010: "draft" Neanderthal genome



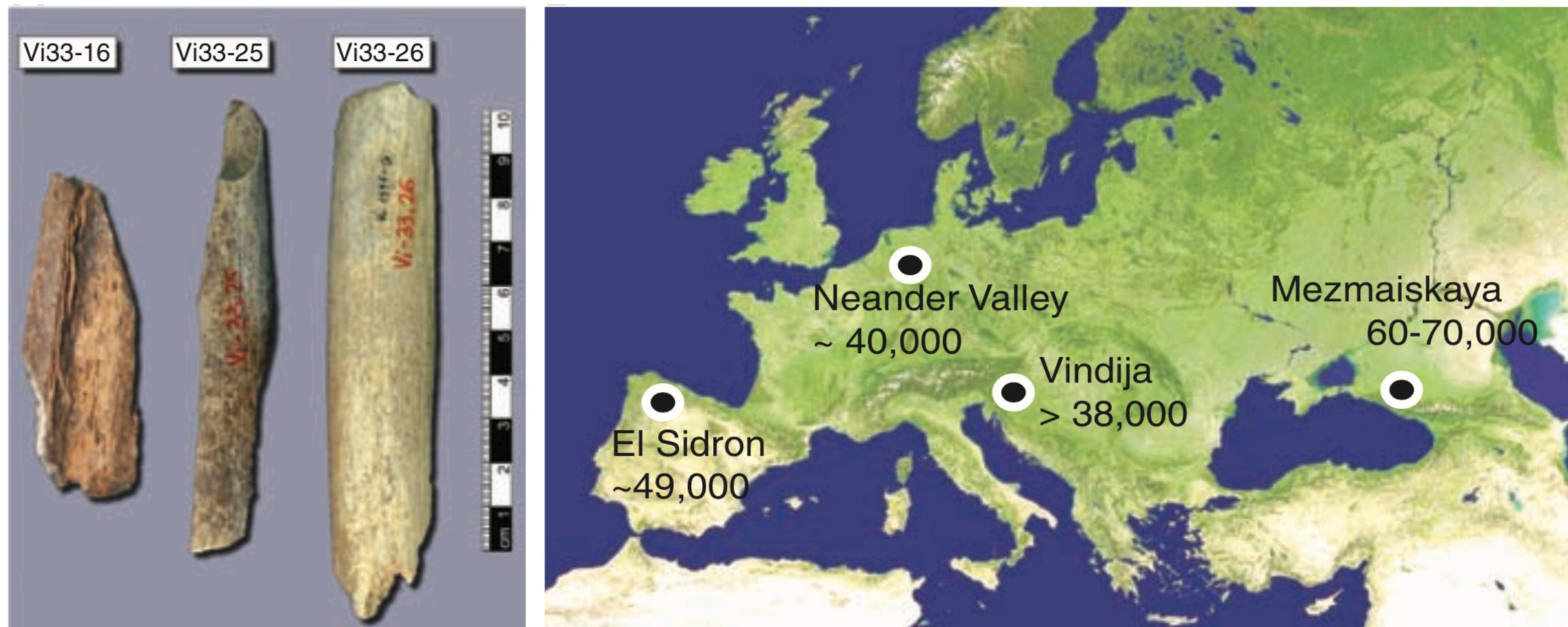
- DNA extracted from bone powder of three bones

2010: "draft" Neanderthal genome



- DNA extracted from bone powder of three bones
- ~1.3X coverage genome (on average, each position in the genome covered by 1.3 fragments)

2010: "draft" Neanderthal genome

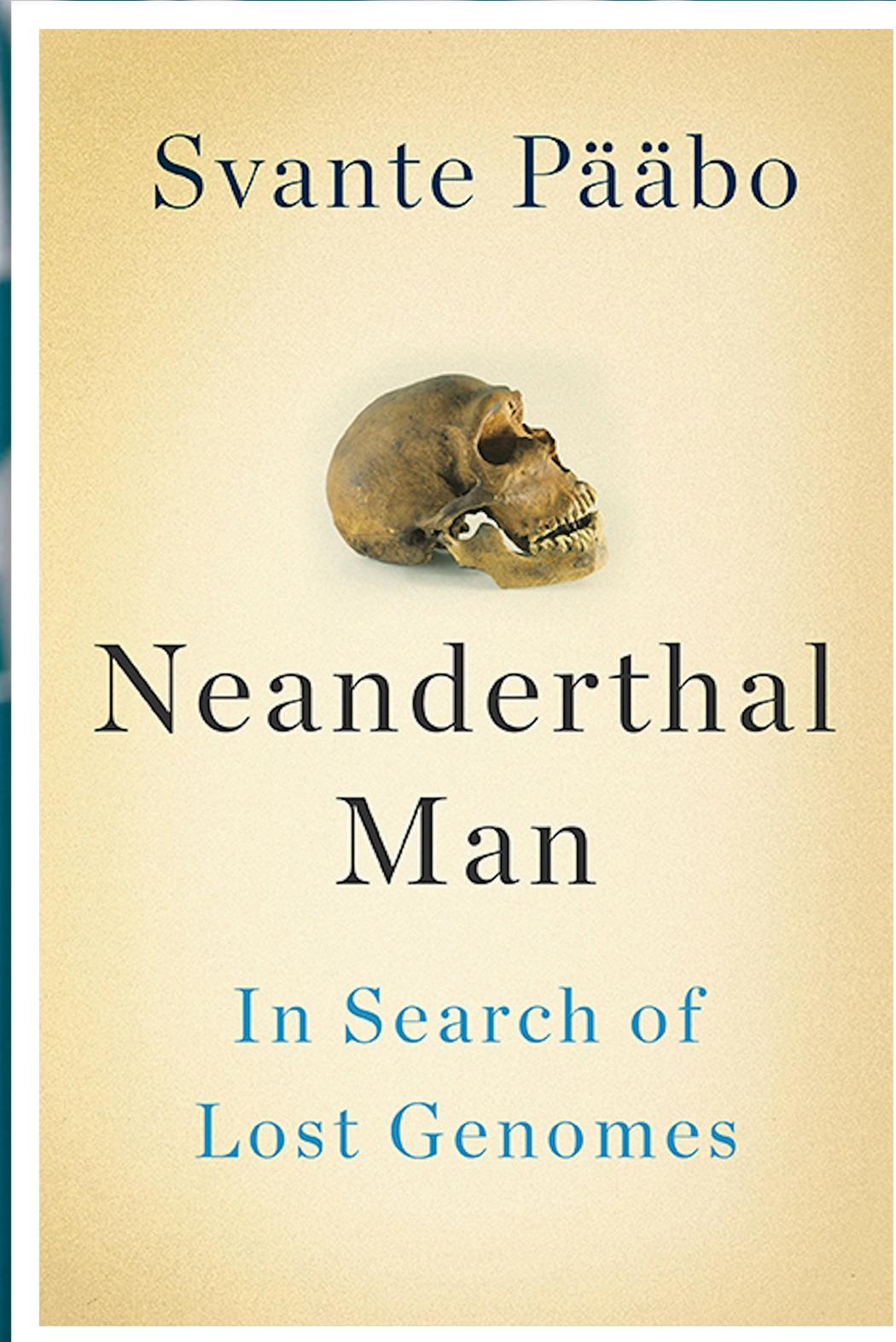


- DNA extracted from bone powder of three bones
- ~1.3X coverage genome (on average, each position in the genome covered by 1.3 fragments)
- to this date we have four more high-coverage archaic genomes up to ~40X

Svante Pääbo
Nobel Prize 2022



Svante Pääbo Nobel Prize 2022



Typical aDNA workflow



Typical aDNA workflow



**dentist
drill**

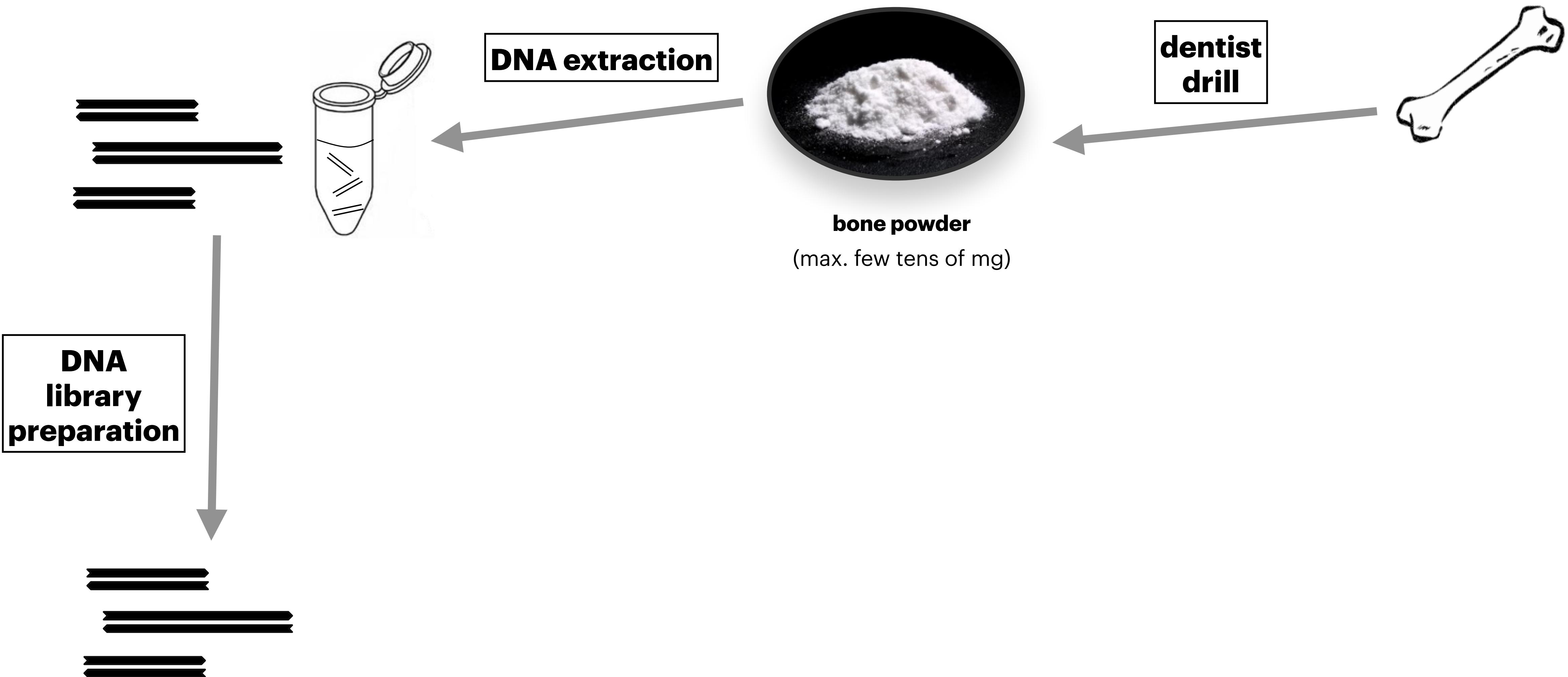


bone powder
(max. few tens of mg)

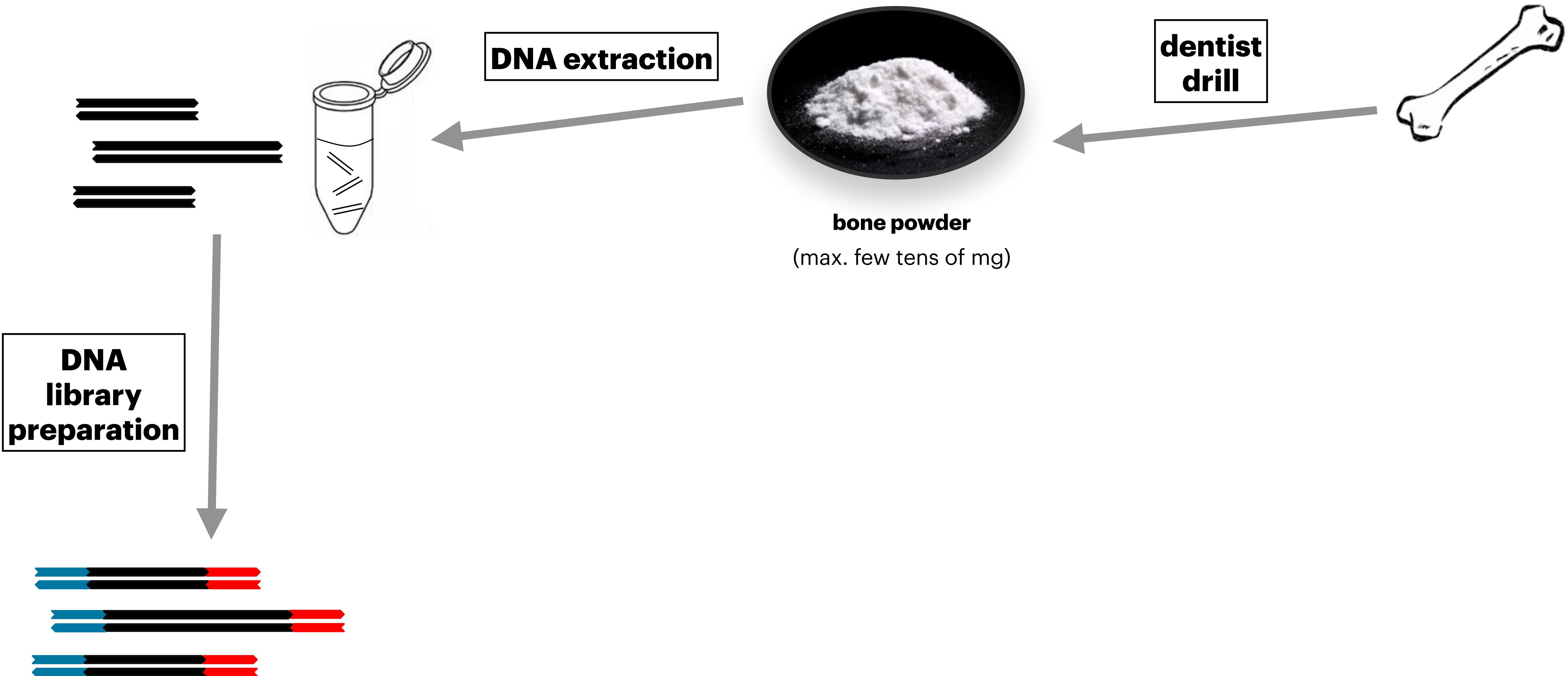
Typical aDNA workflow



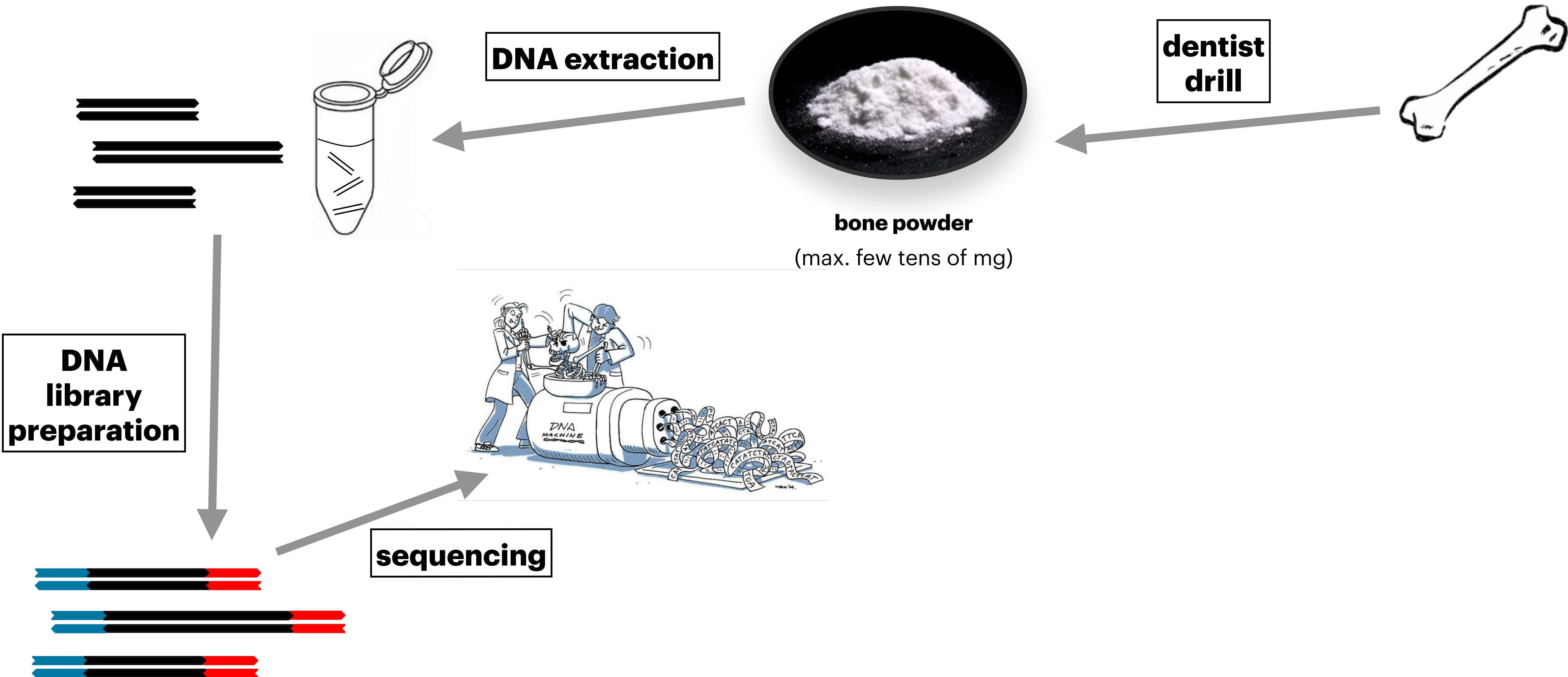
Typical aDNA workflow



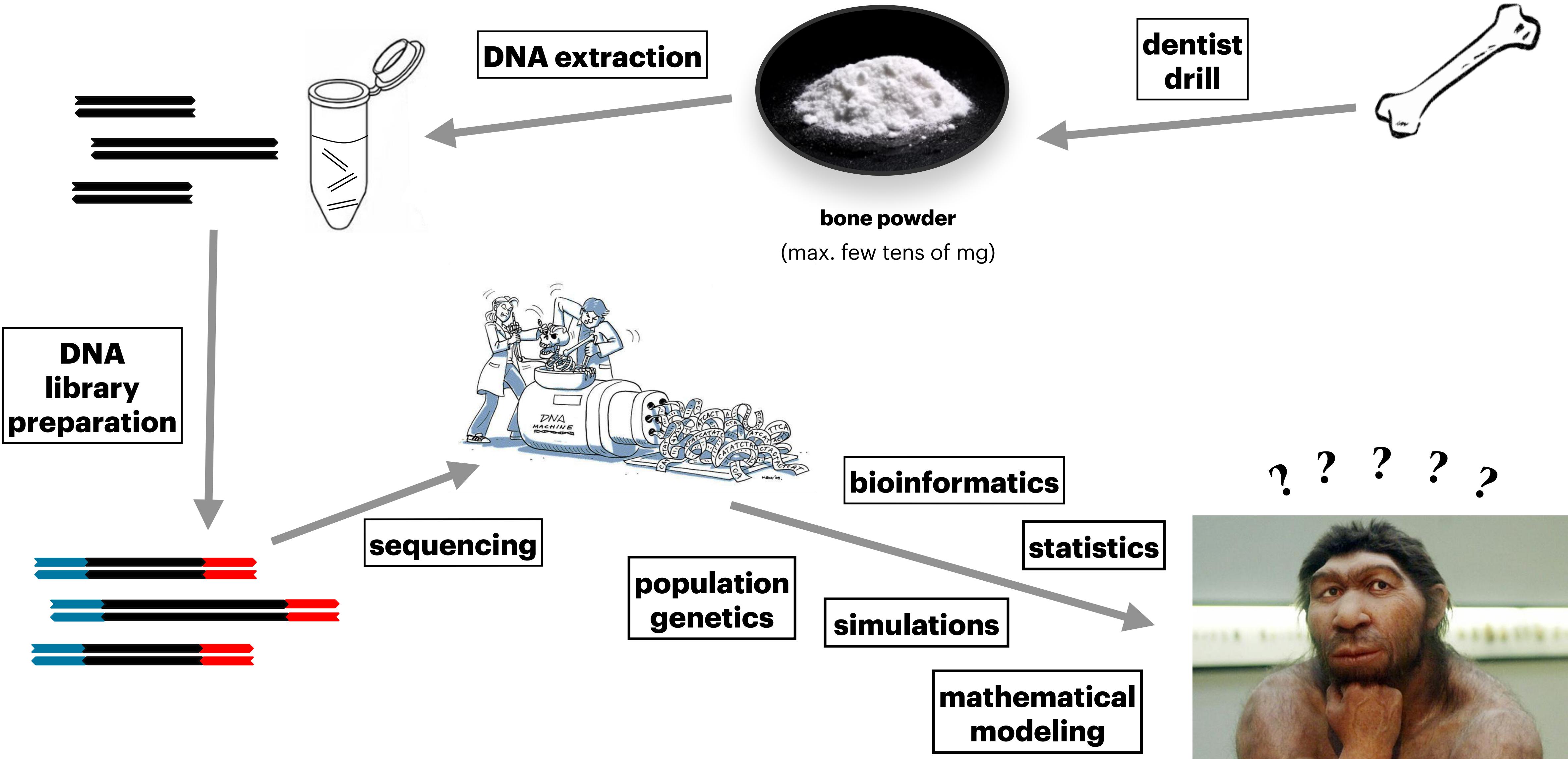
Typical aDNA workflow



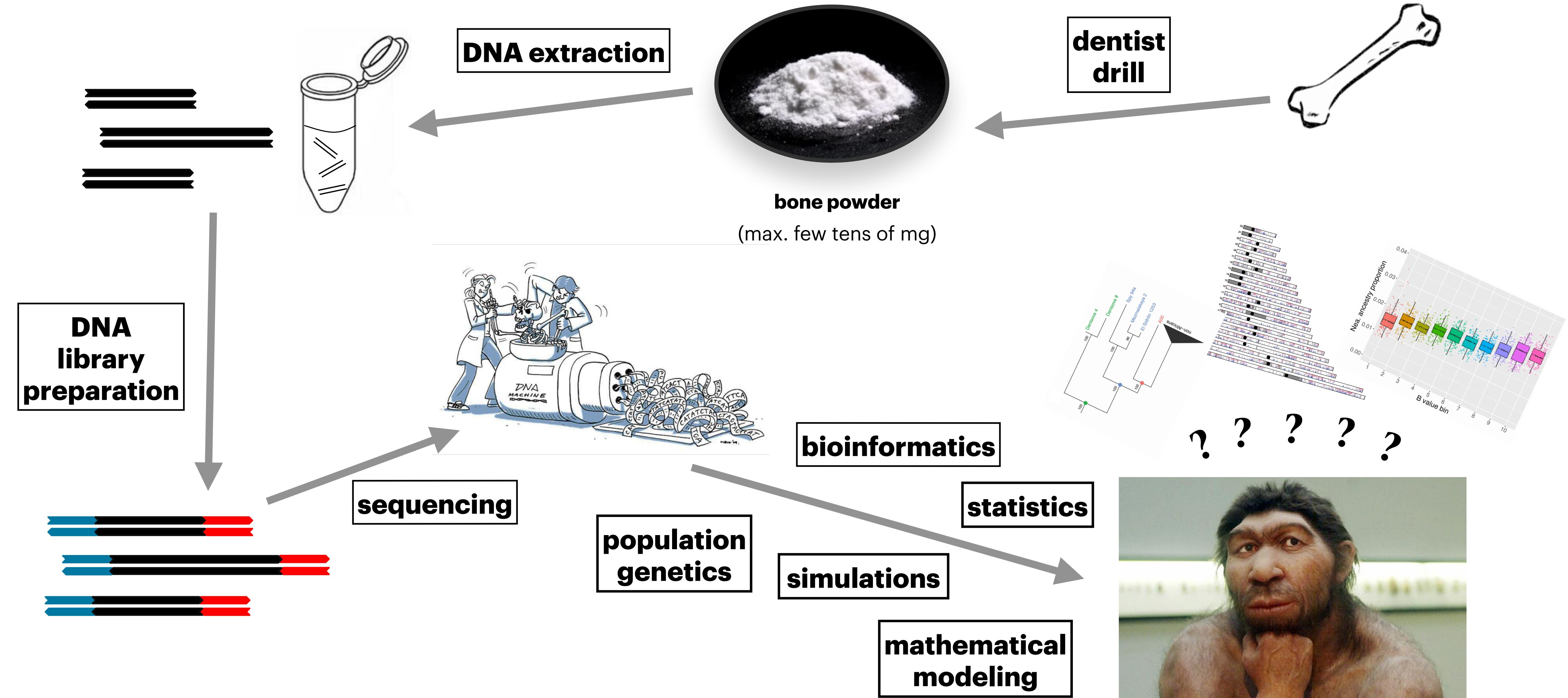
Typical aDNA workflow



Typical aDNA workflow



Typical aDNA workflow



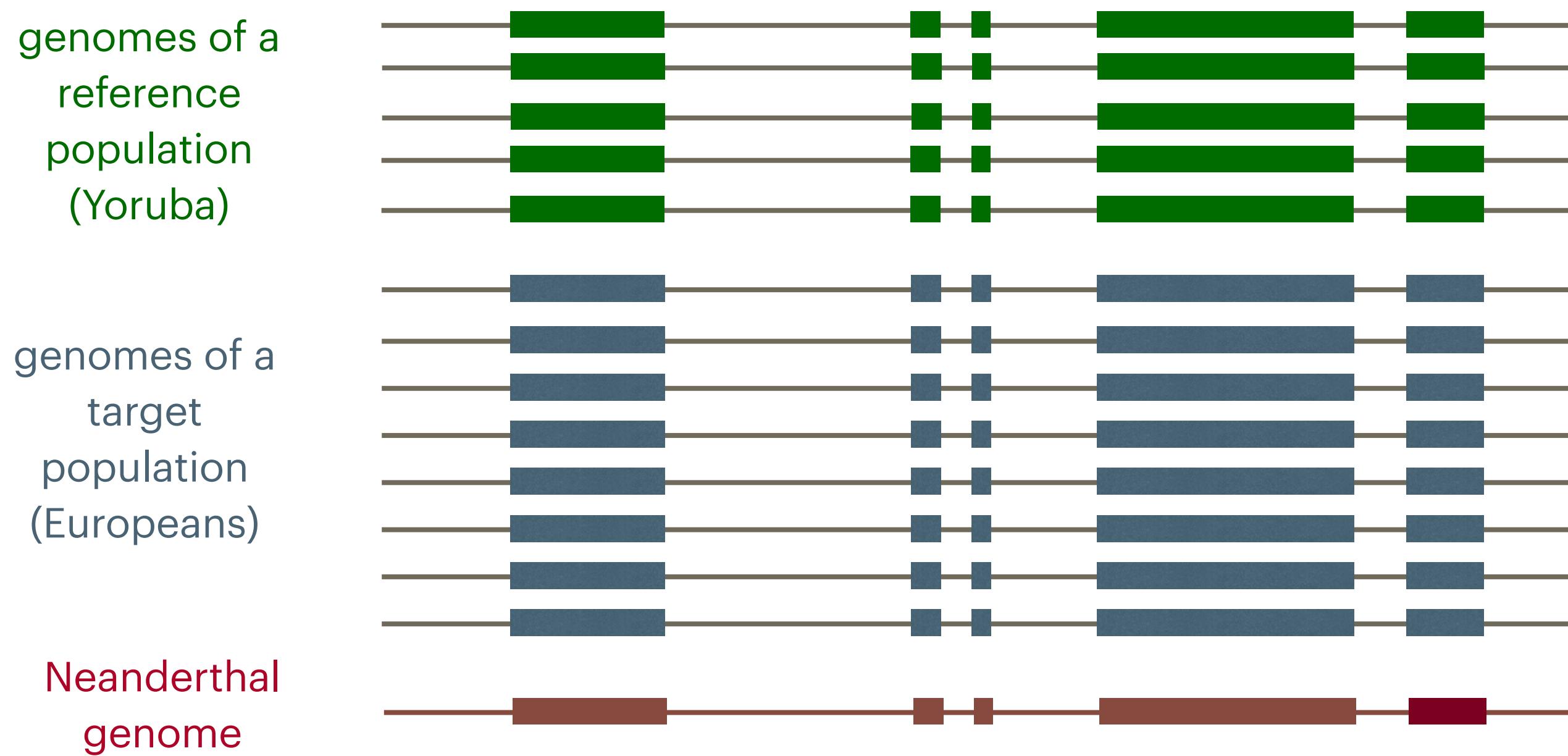
So we have a Neanderthal genome...



...how to test the gene-flow hypothesis?



How to test the gene-flow hypothesis?



David Reich

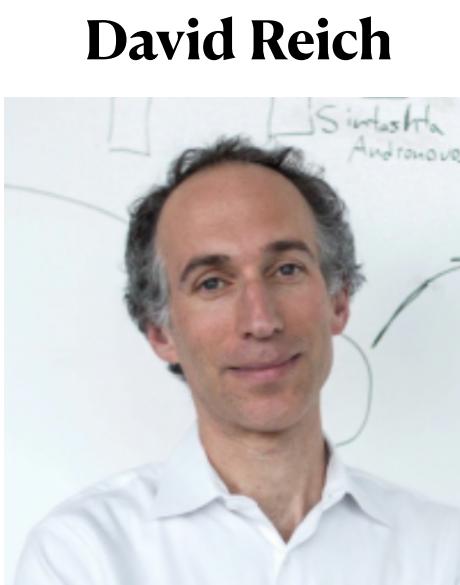


Nick Patterson



Green et al. (Science 2010)

How to test the gene-flow hypothesis?



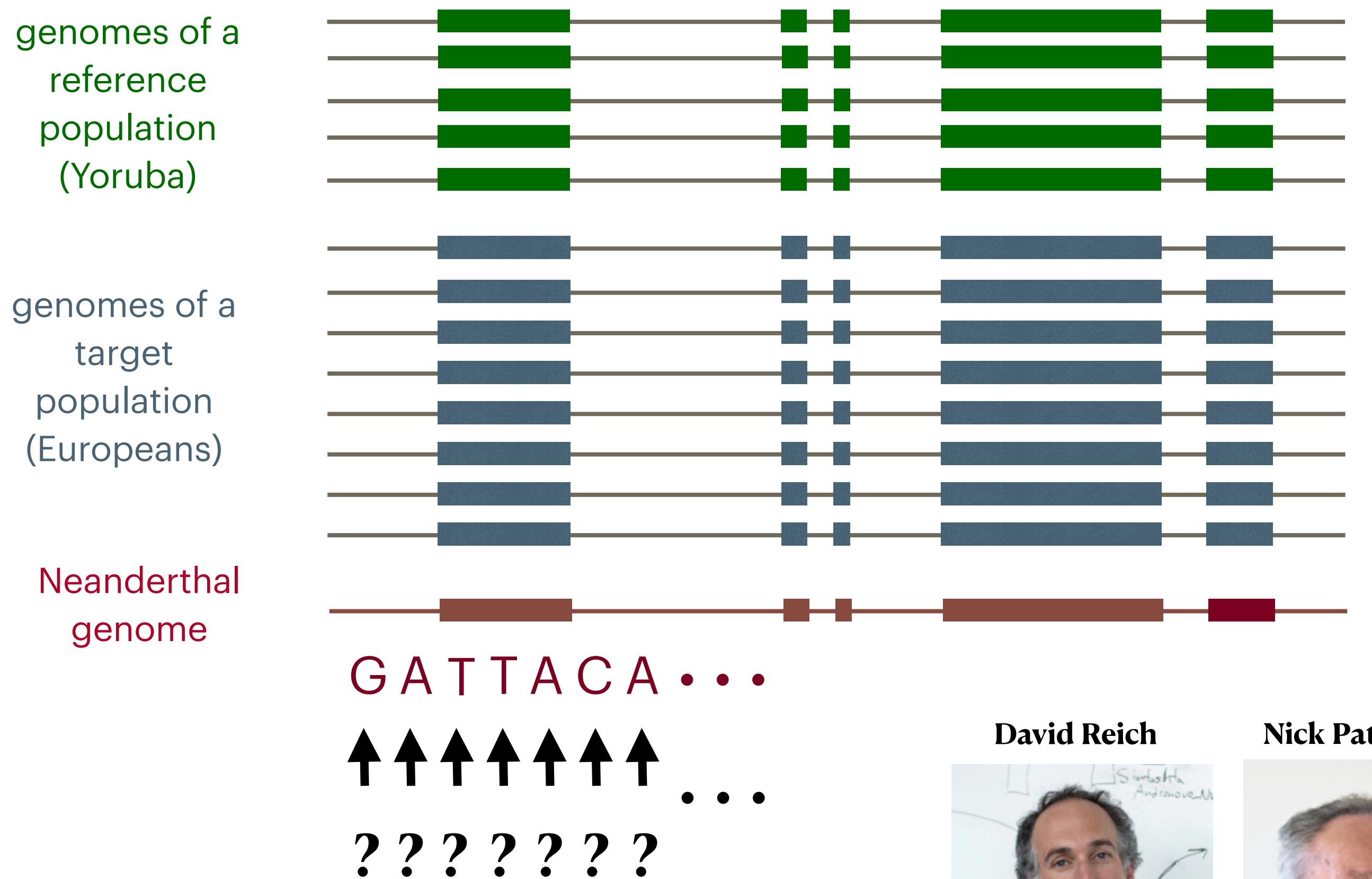
David Reich



Nick Patterson

Green et al. (Science 2010)

How to test the gene-flow hypothesis?



David Reich

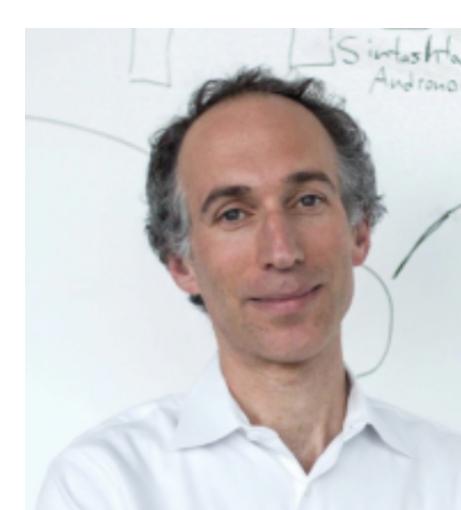
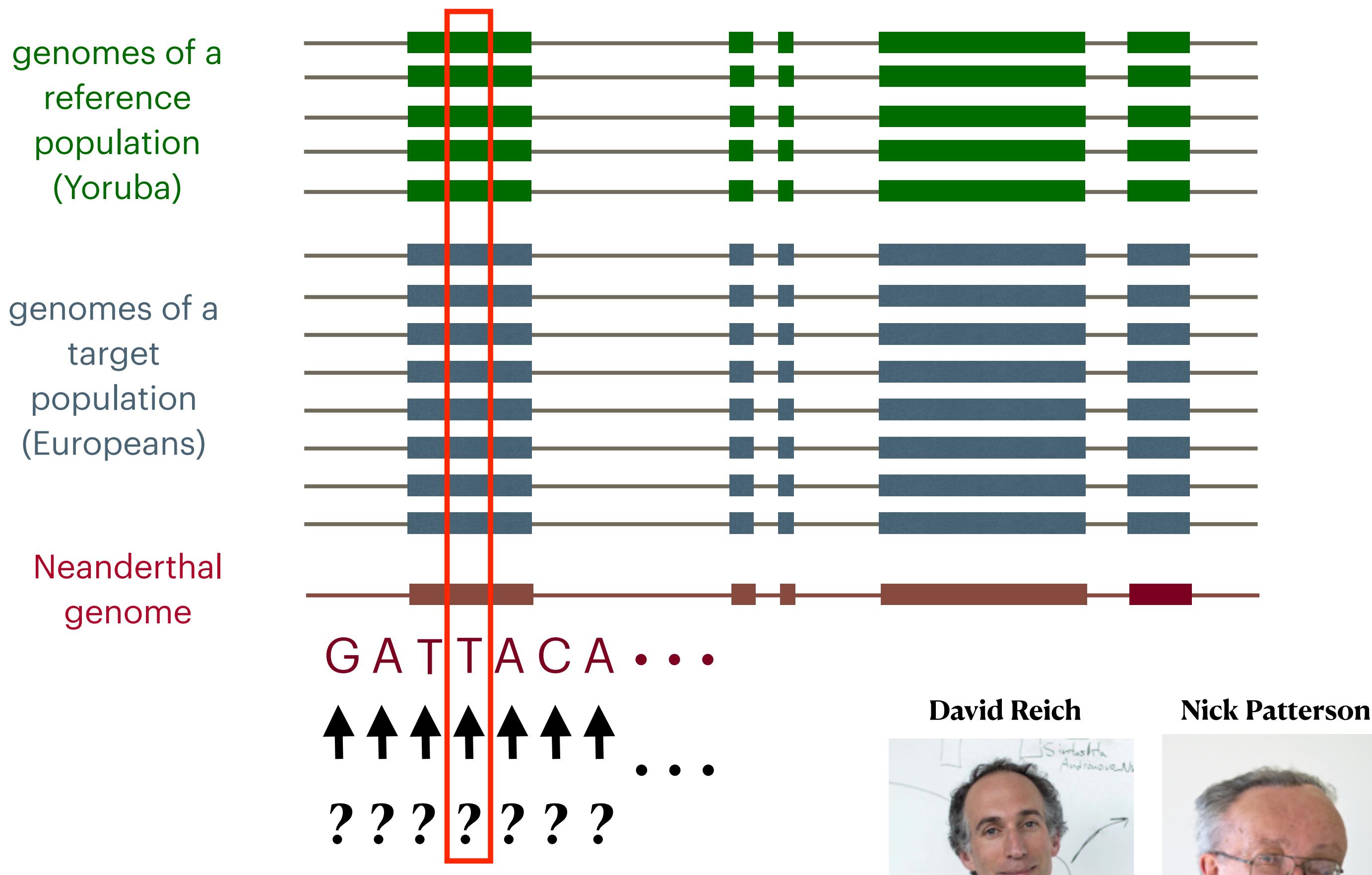


Nick Patterson

Green et al. (Science 2010)

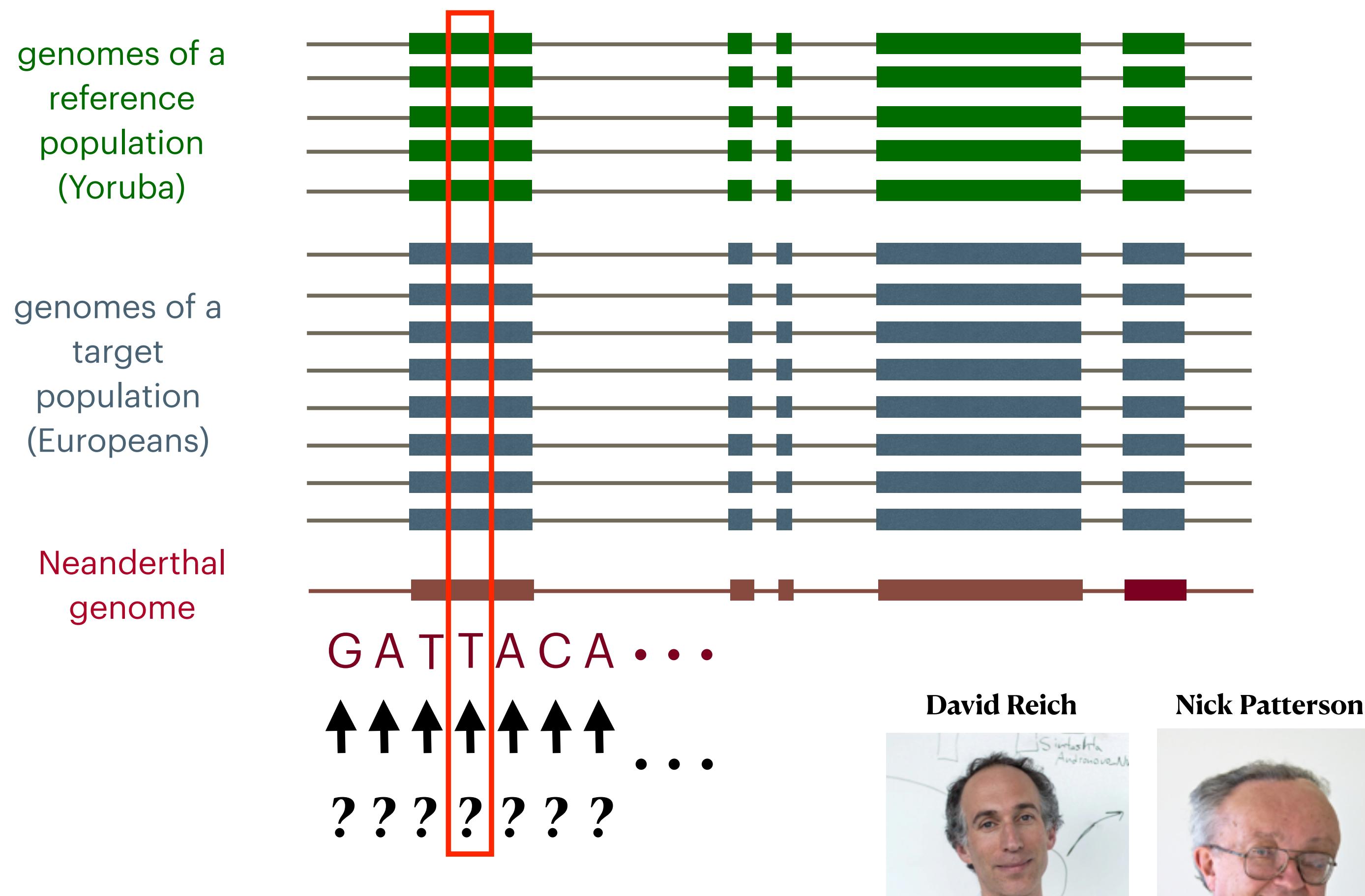
How to test the gene-flow hypothesis?

Situation at one locus...



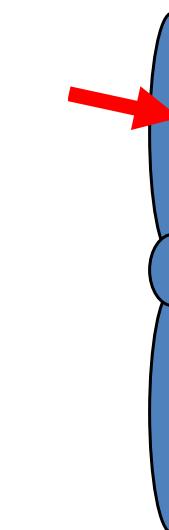
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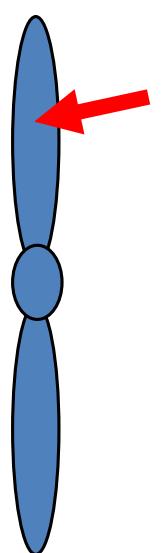


Situation at one locus...

person of an African ancestry



person of a non-african ancestry (i.e. European)

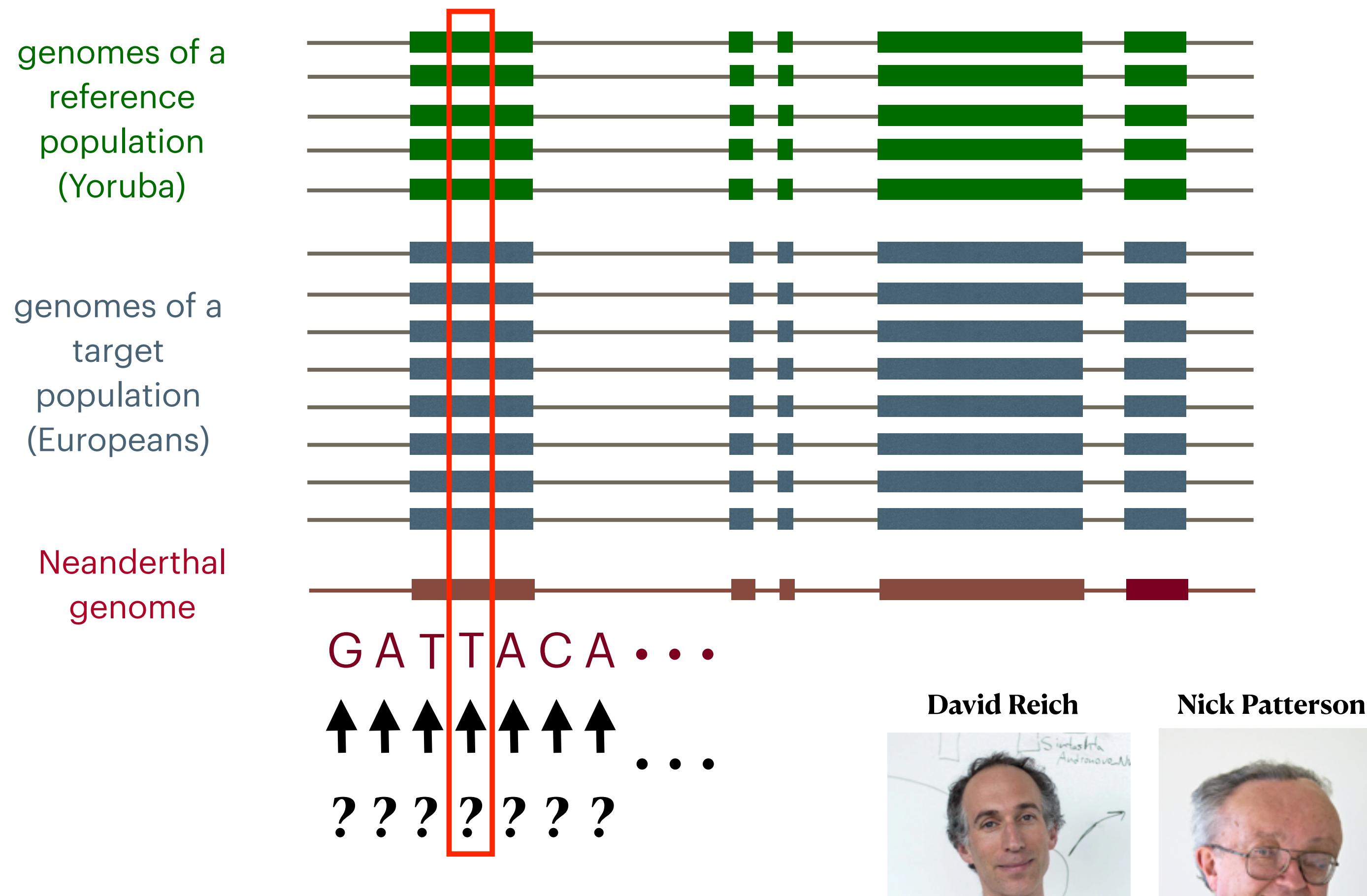


Neanderthal



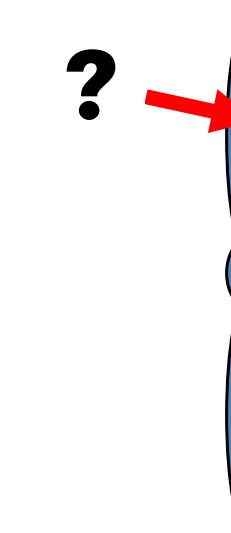
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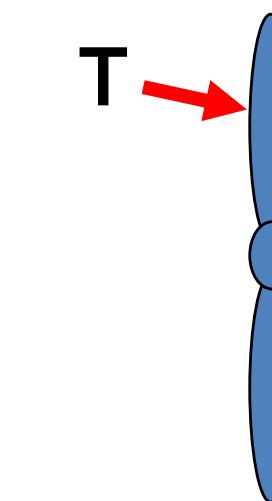
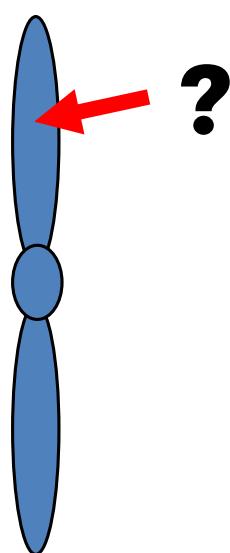


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Neanderthal



Green et al. (Science 2010)

Formal test of introgression

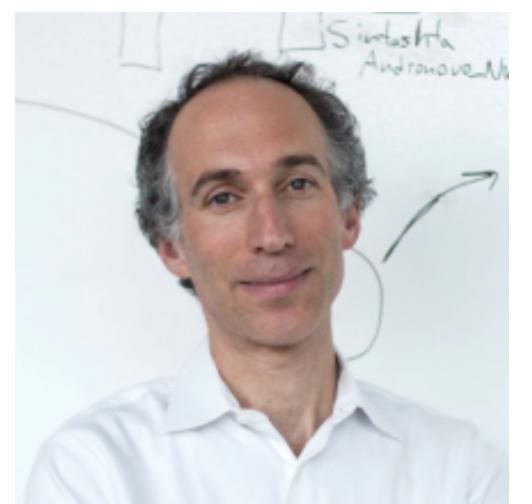
f_4 statistic

Given a set of 4 samples, f_4 compares counts of observed **BABA** vs **ABBA** site patterns.

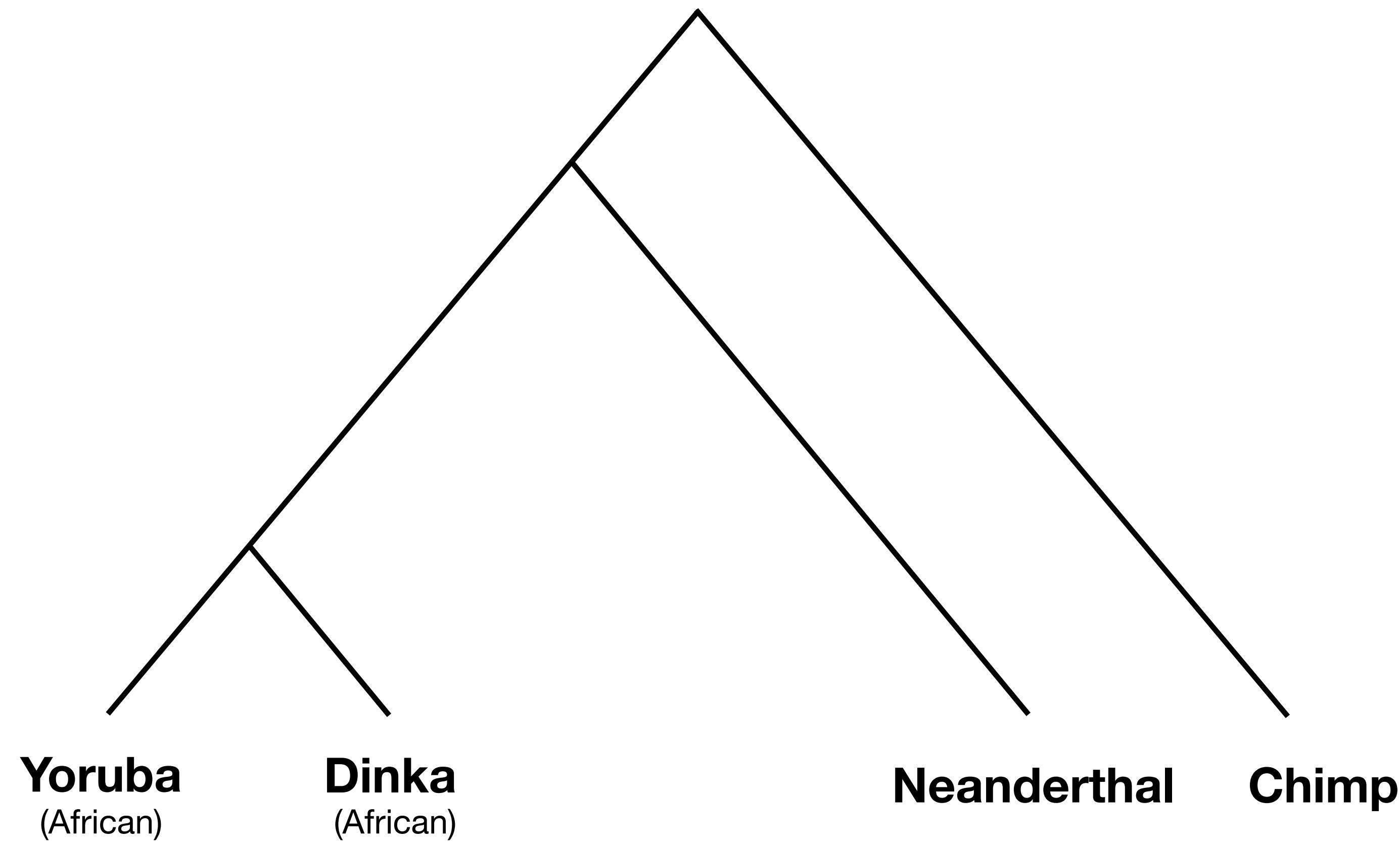
Nick Patterson



David Reich



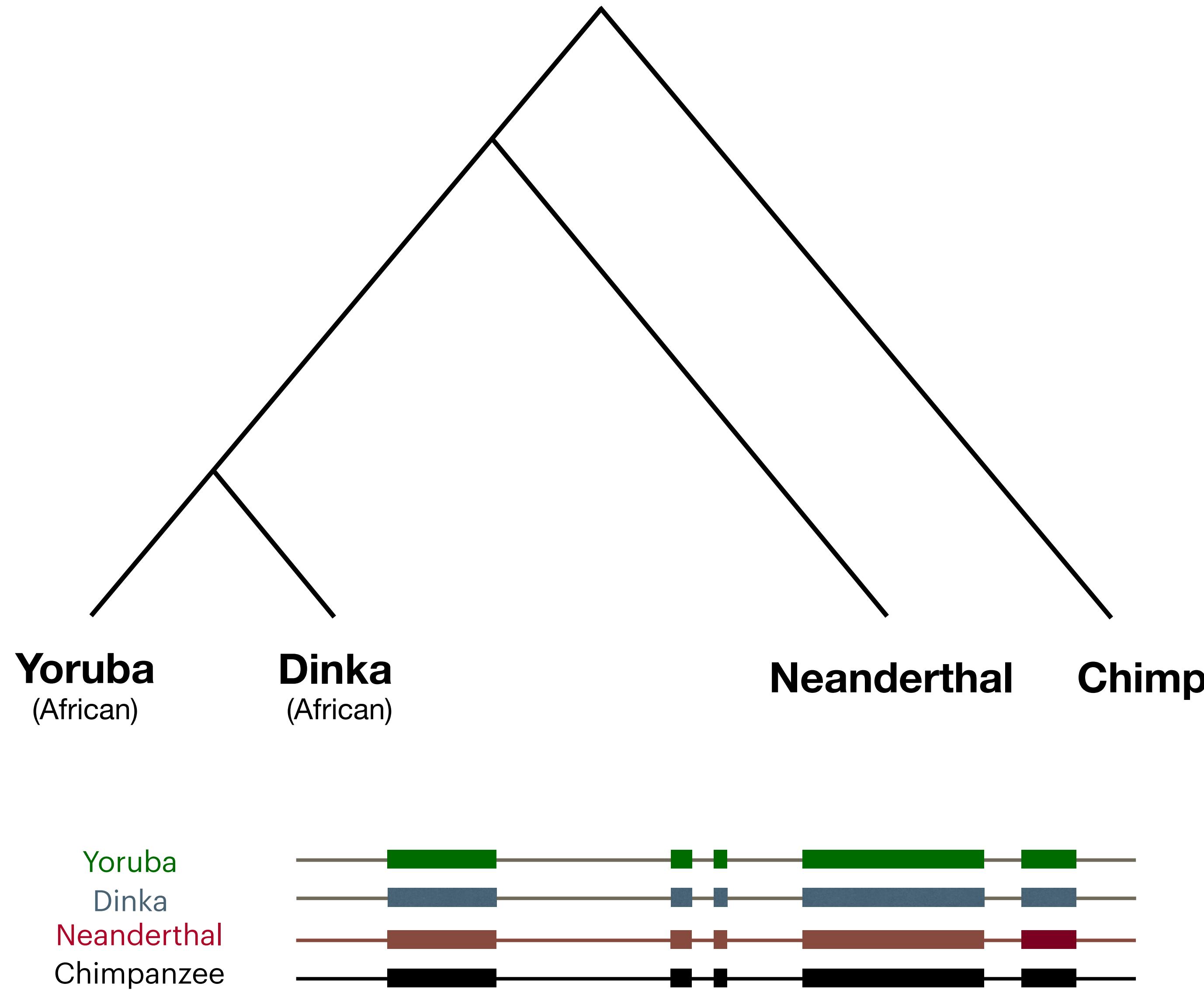
A variant of this statistic that is used equally often is known as **D statistic**.

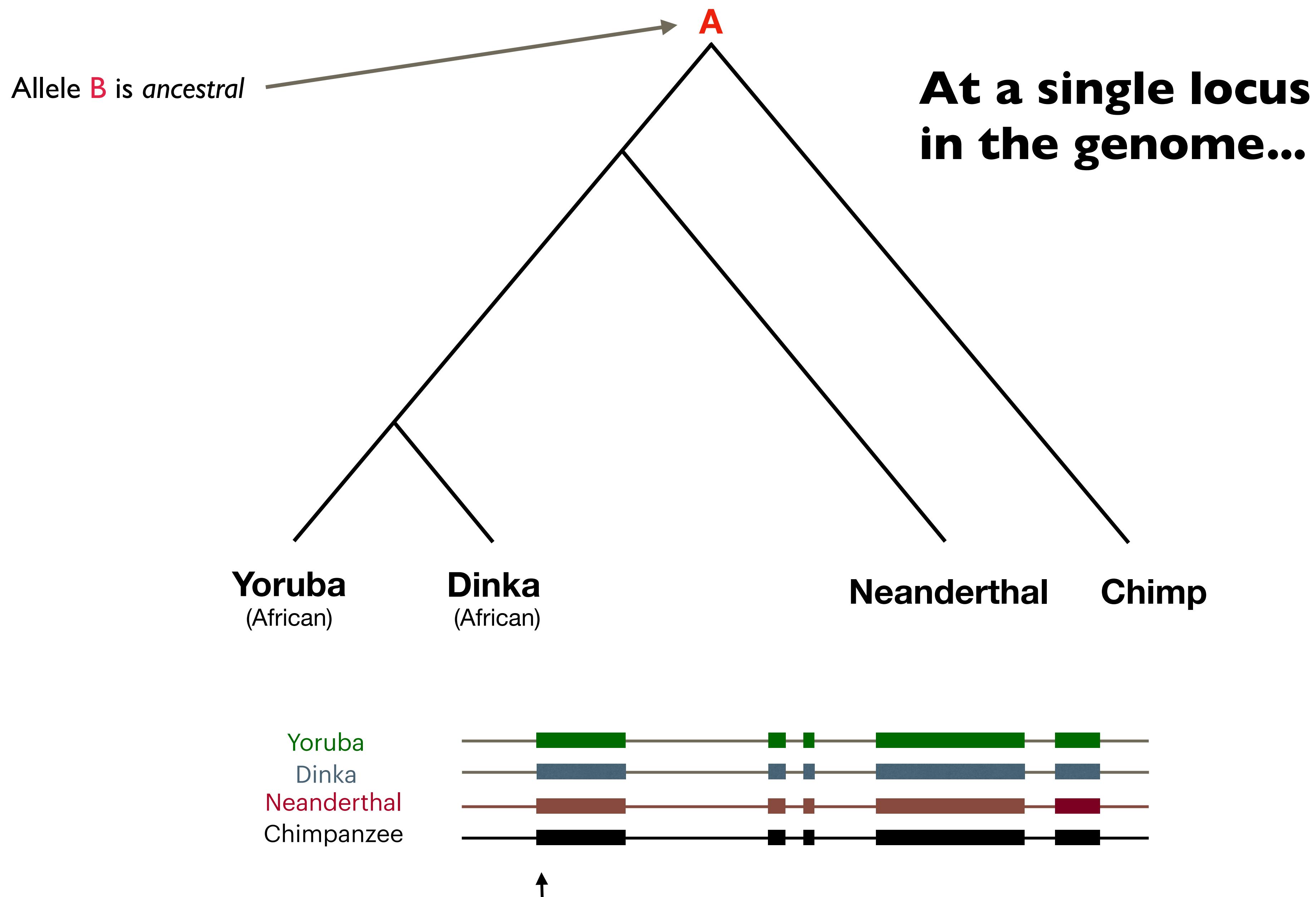


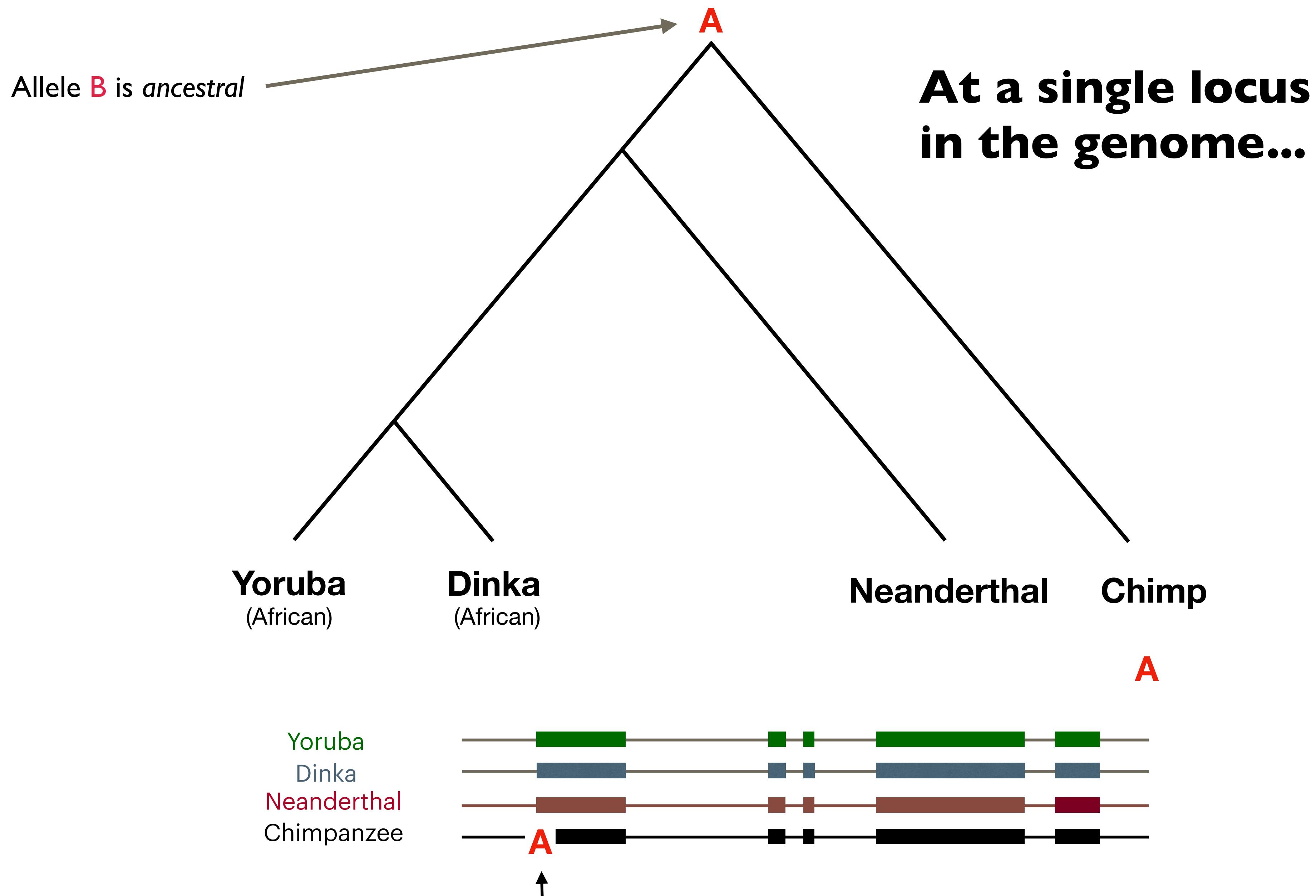
Yoruba
(African)

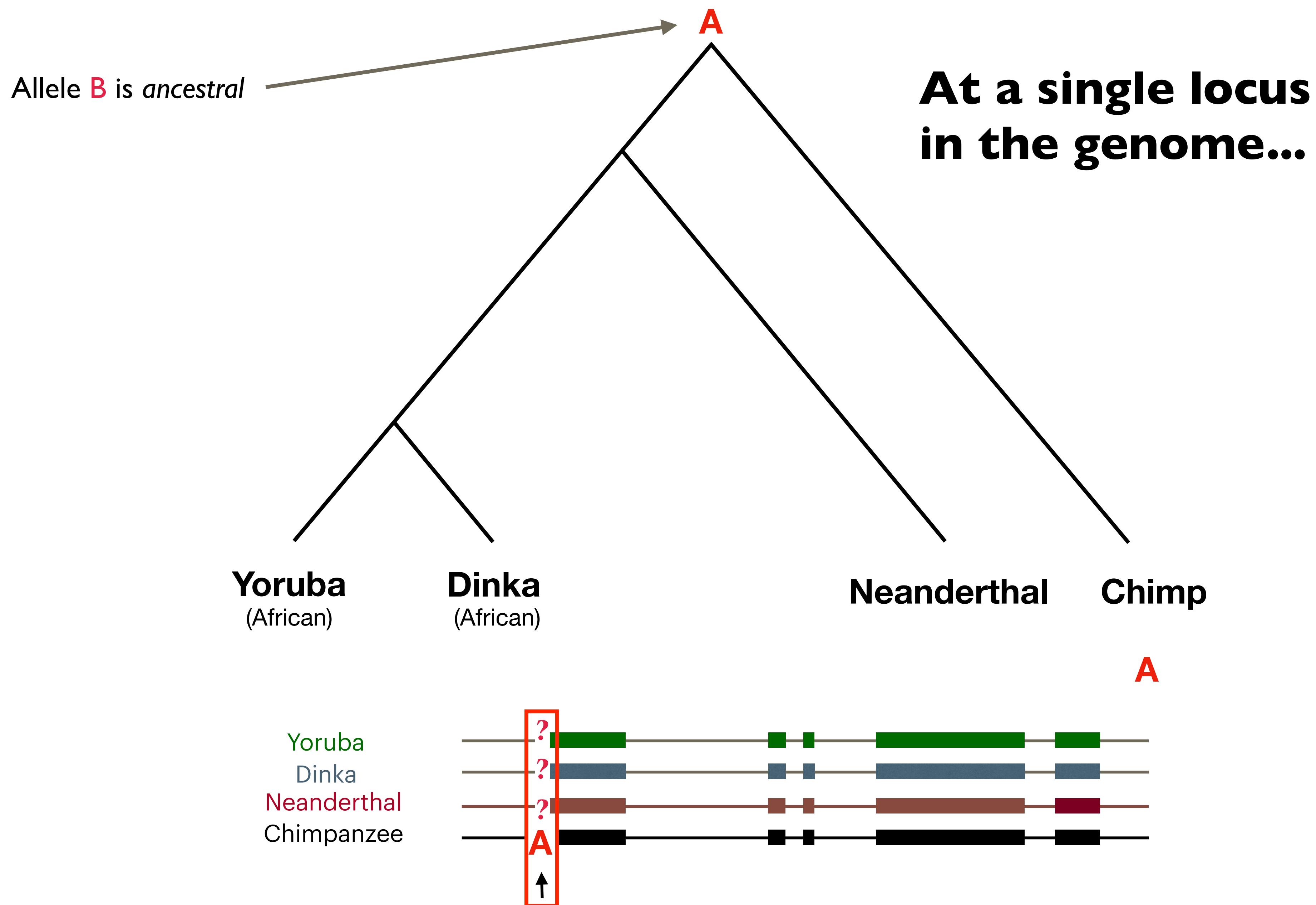
Dinka
(African)

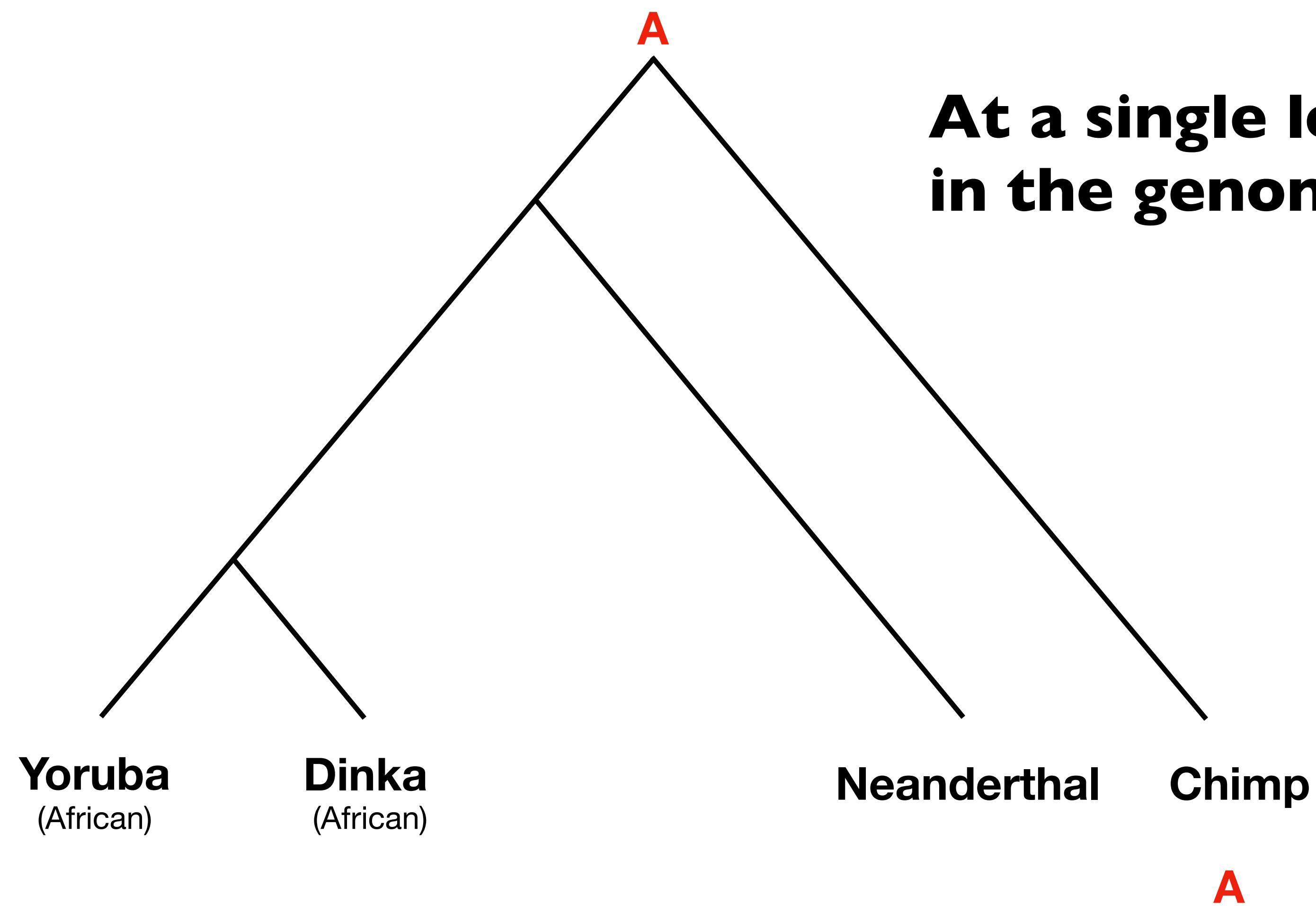
Neanderthal **Chimp**





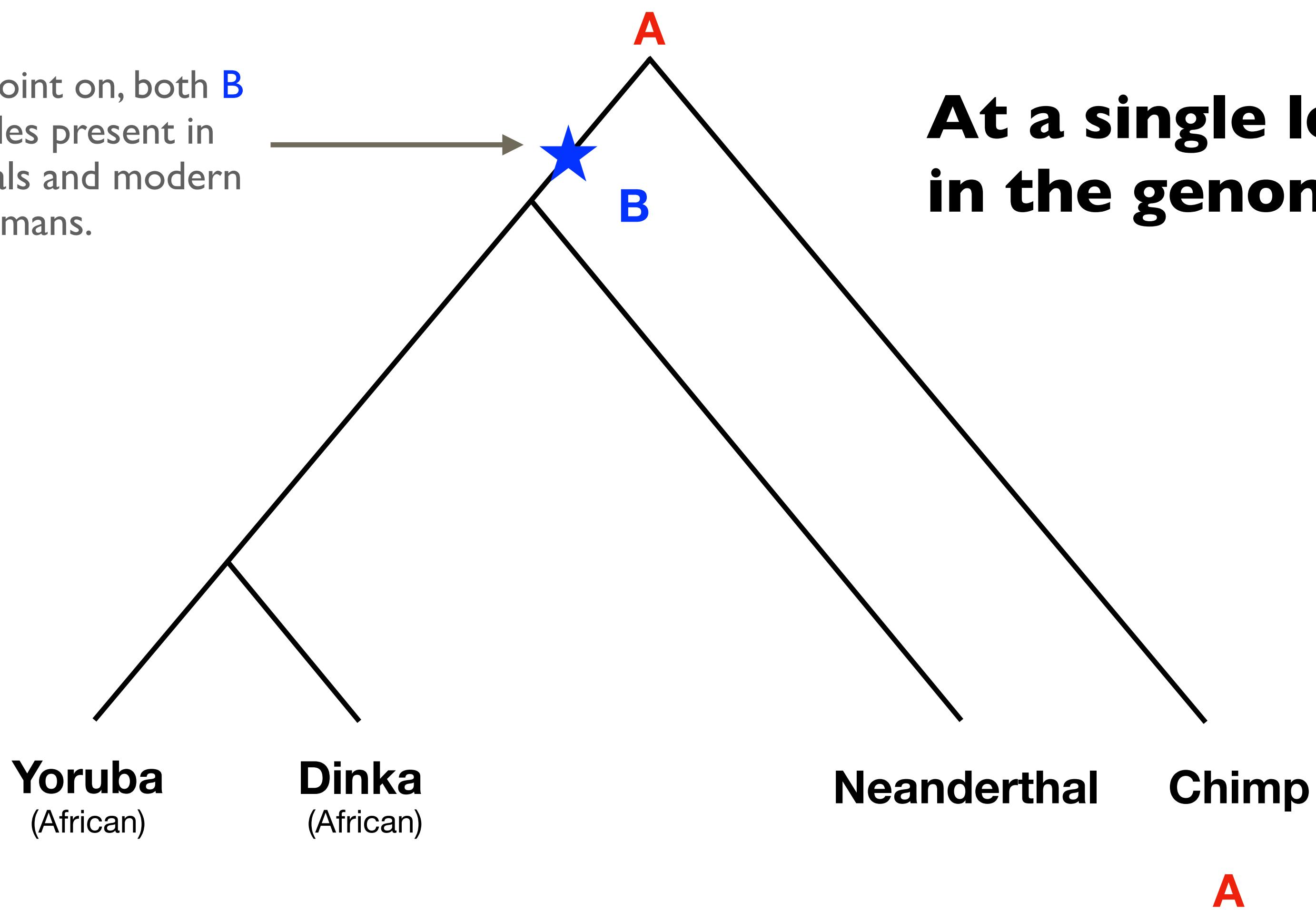






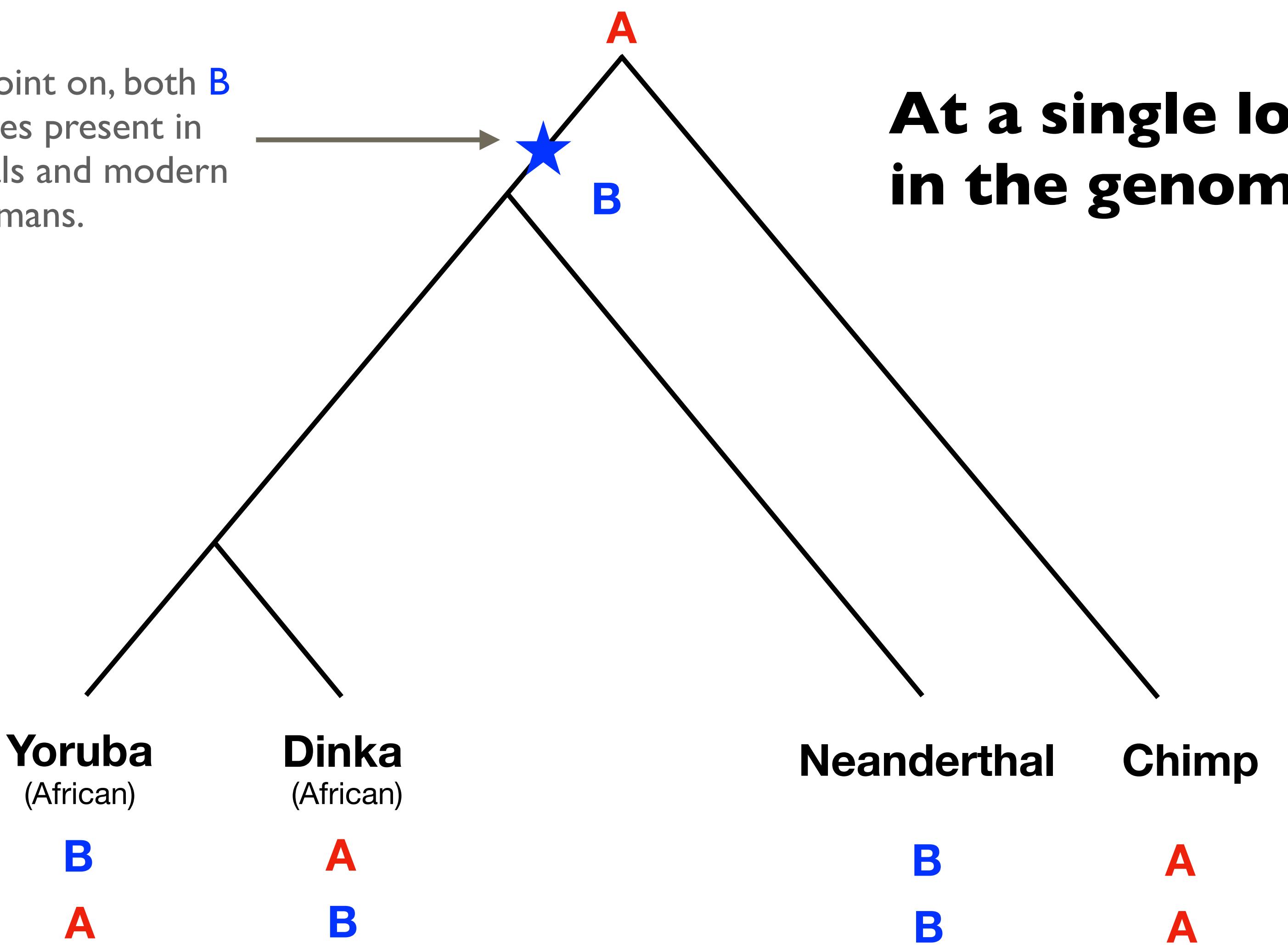
**At a single locus
in the genome...**

From this point on, both **B** and **A** alleles present in Neanderthals and modern humans.



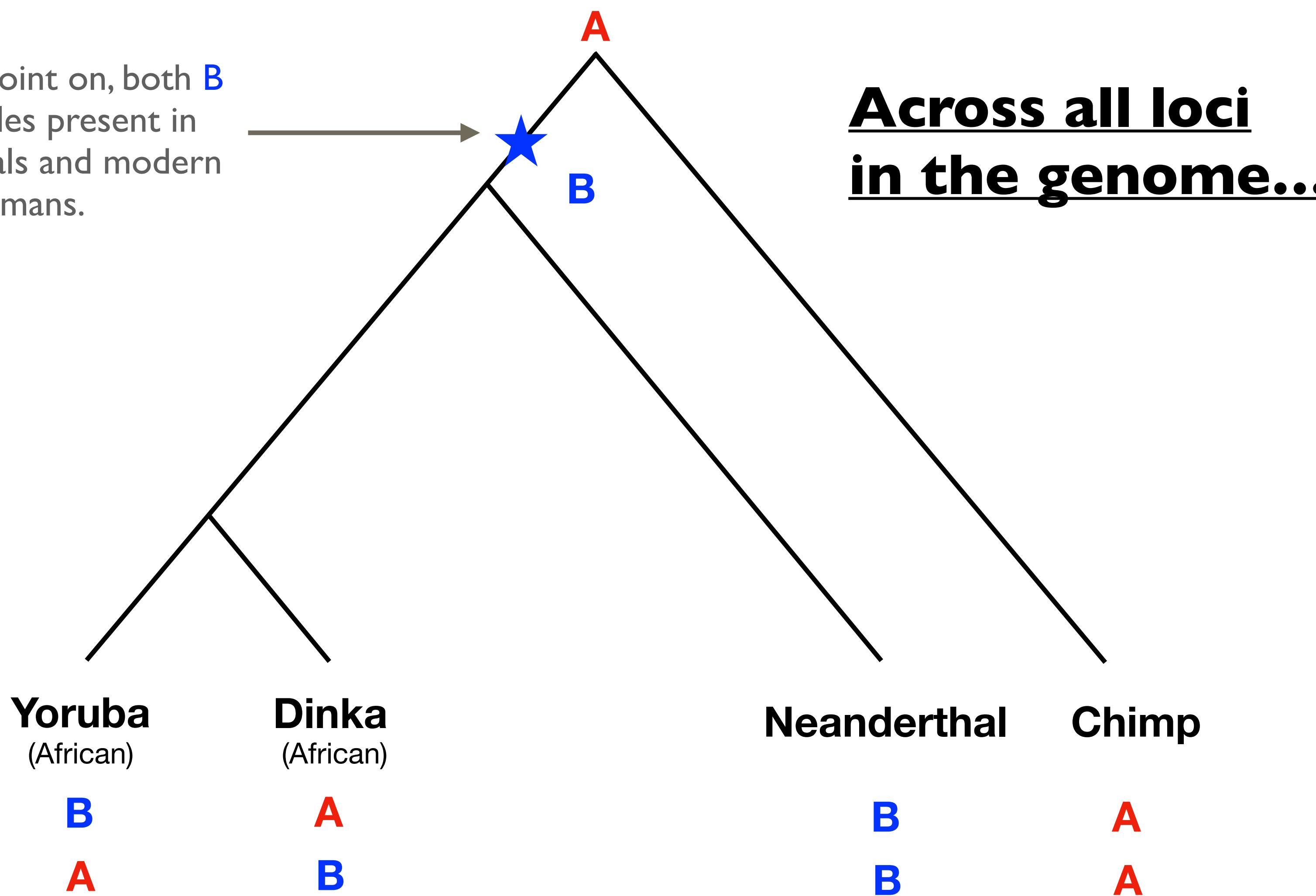
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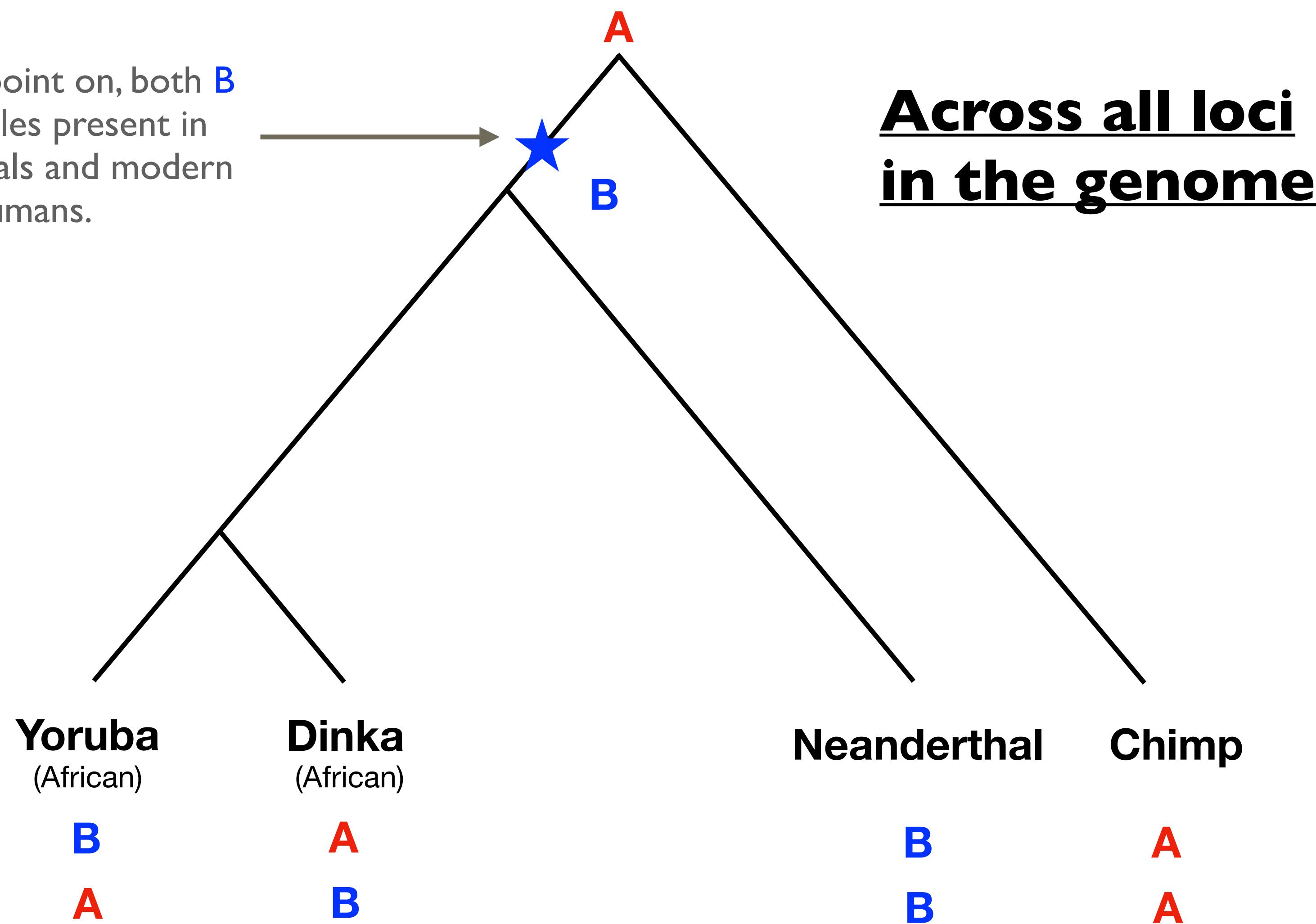


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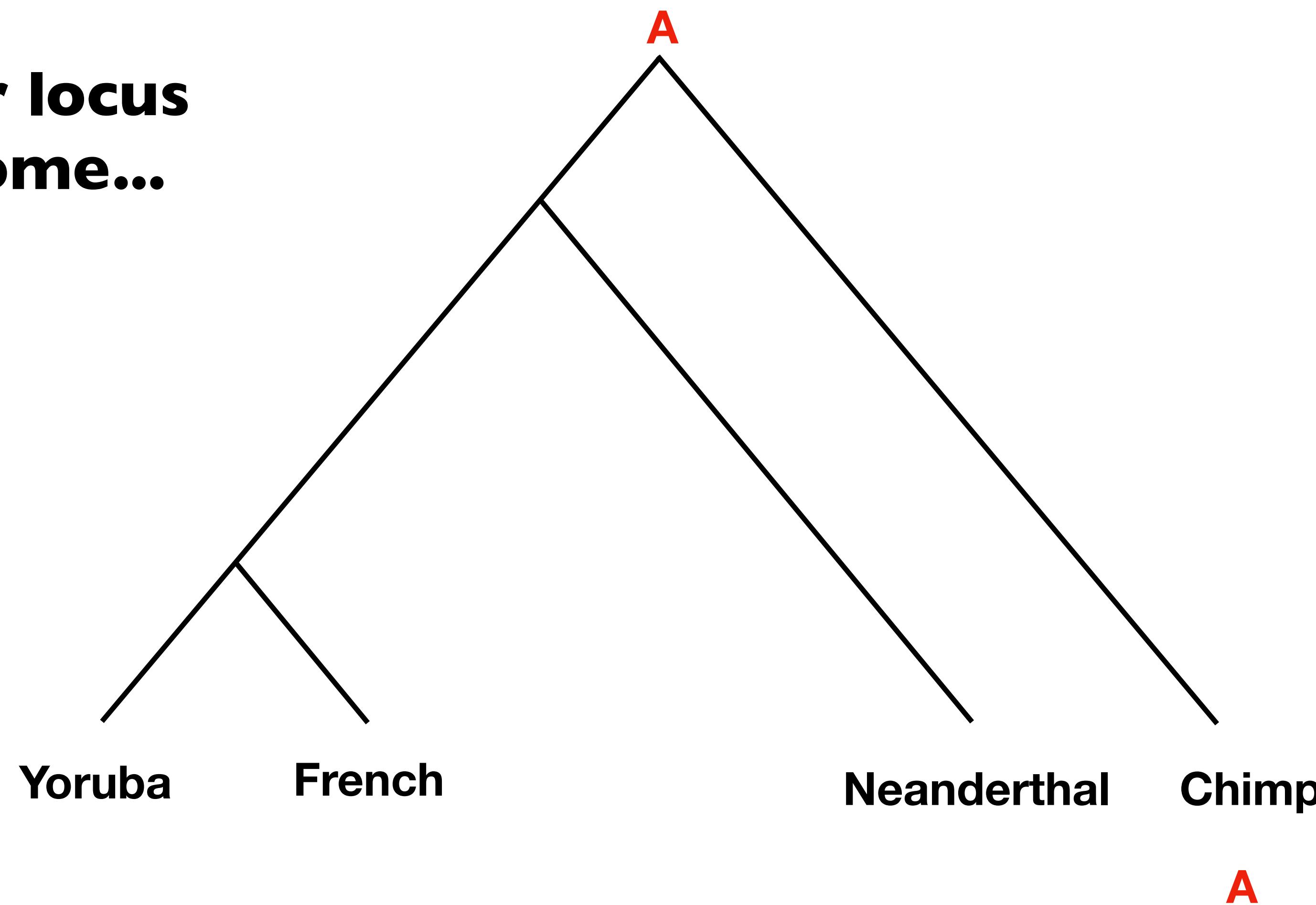
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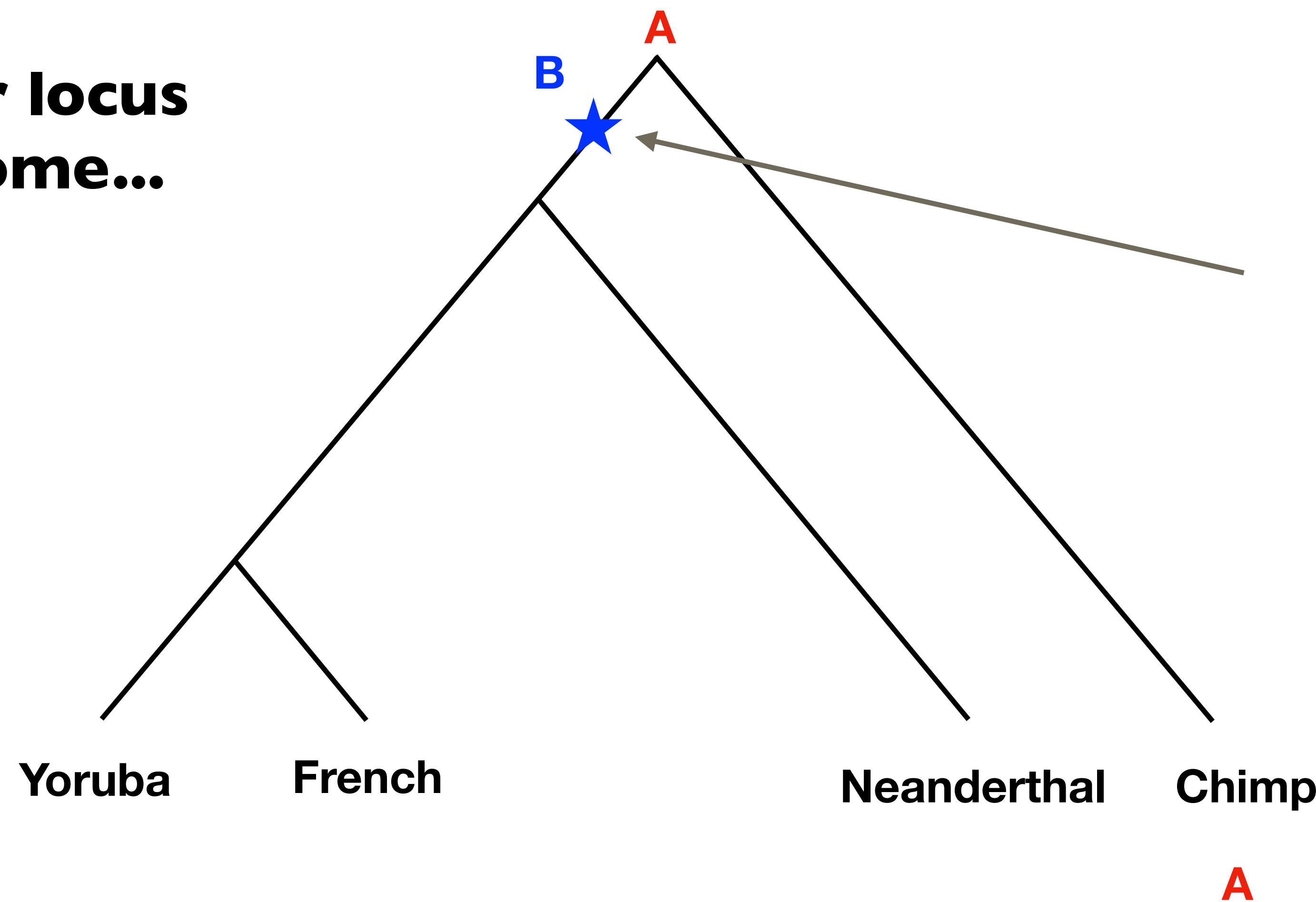
**Across all loci
in the genome...**

No introgression: *f4 statistic* = (# BABA - #ABBA) / # SNPs ~ 0

**At another locus
in the genome...**



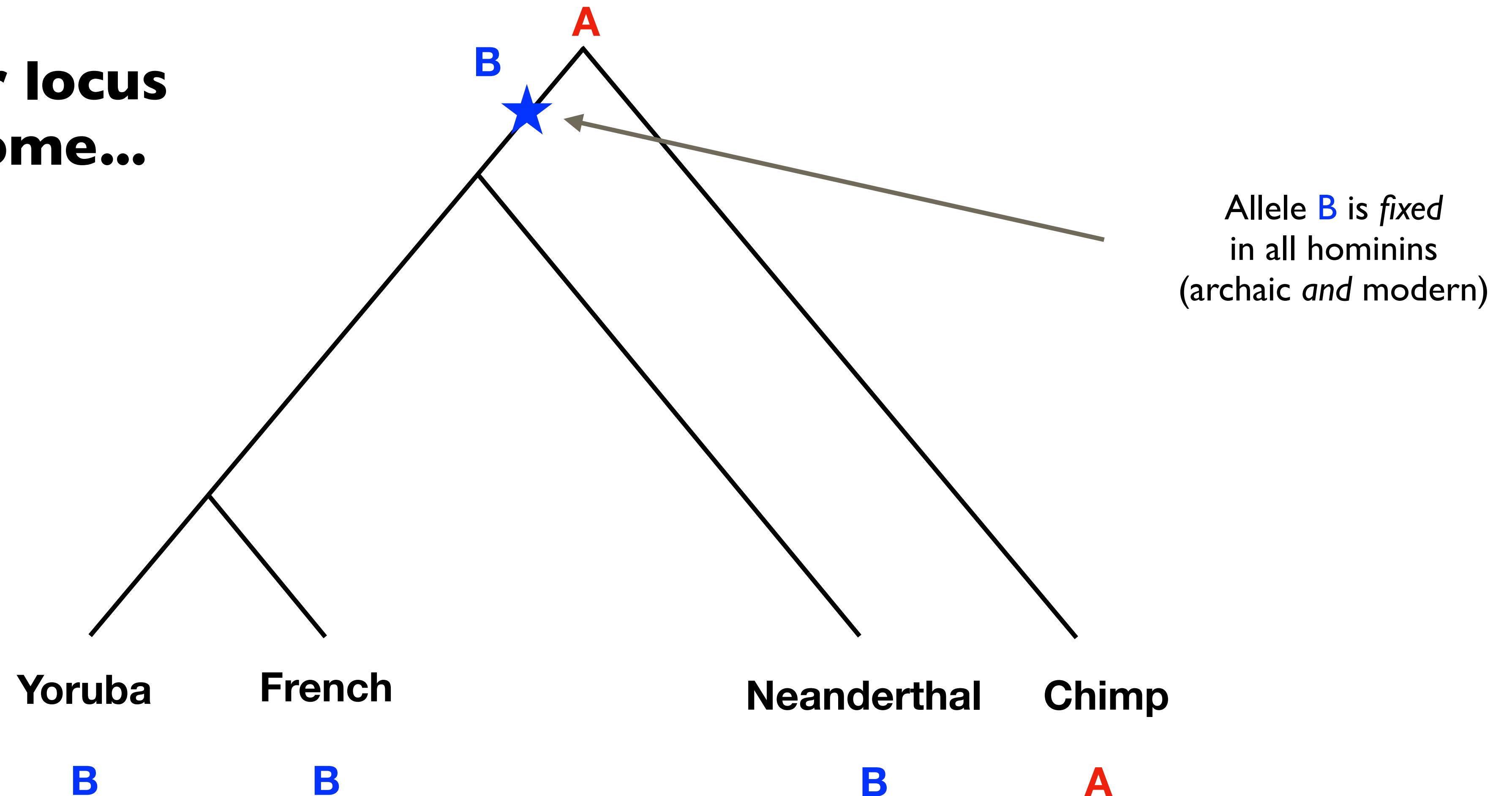
**At another locus
in the genome...**



Allele **B** is *fixed*
in all hominins
(archaic *and* modern)

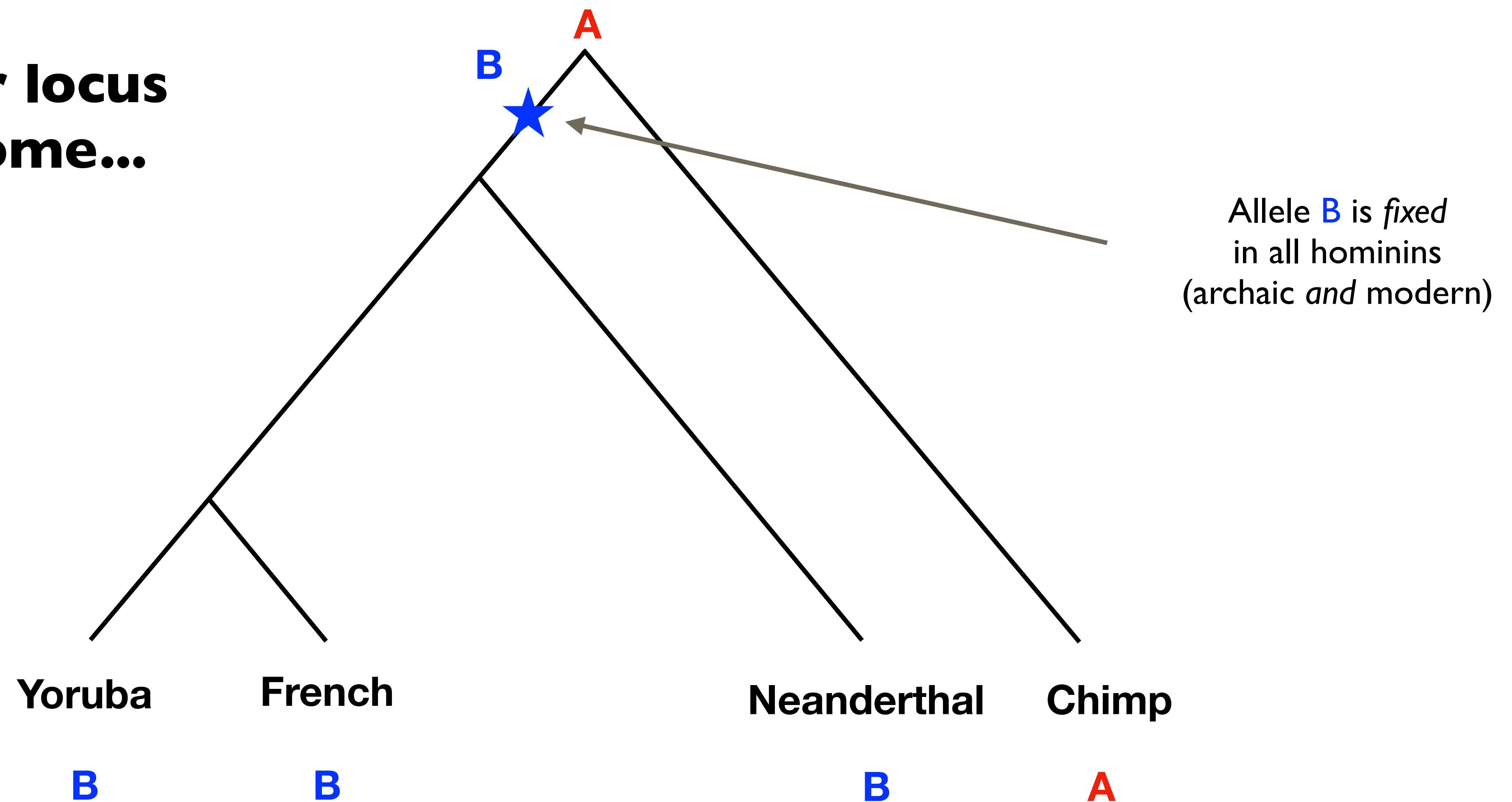
A

**At another locus
in the genome...**



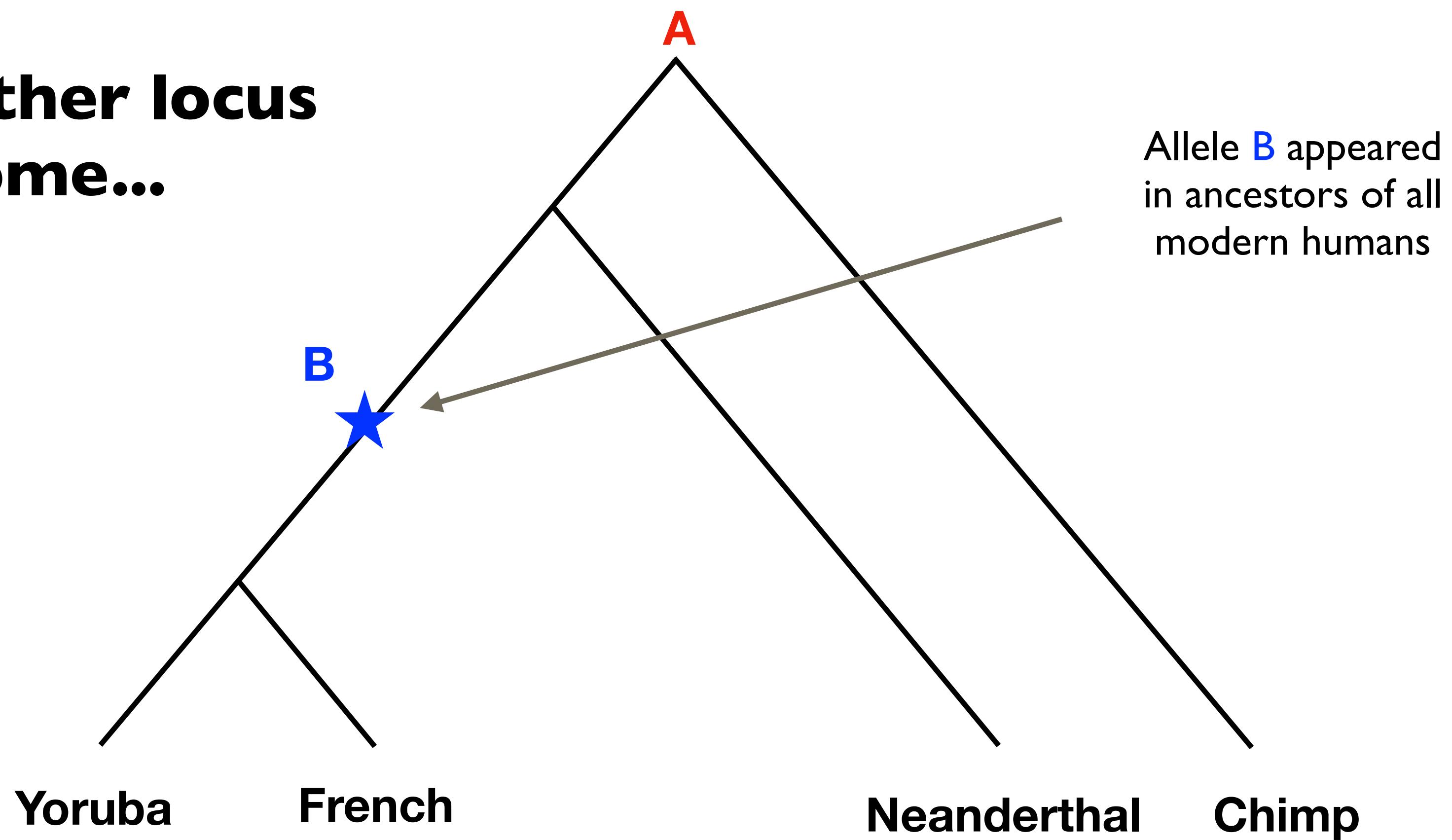
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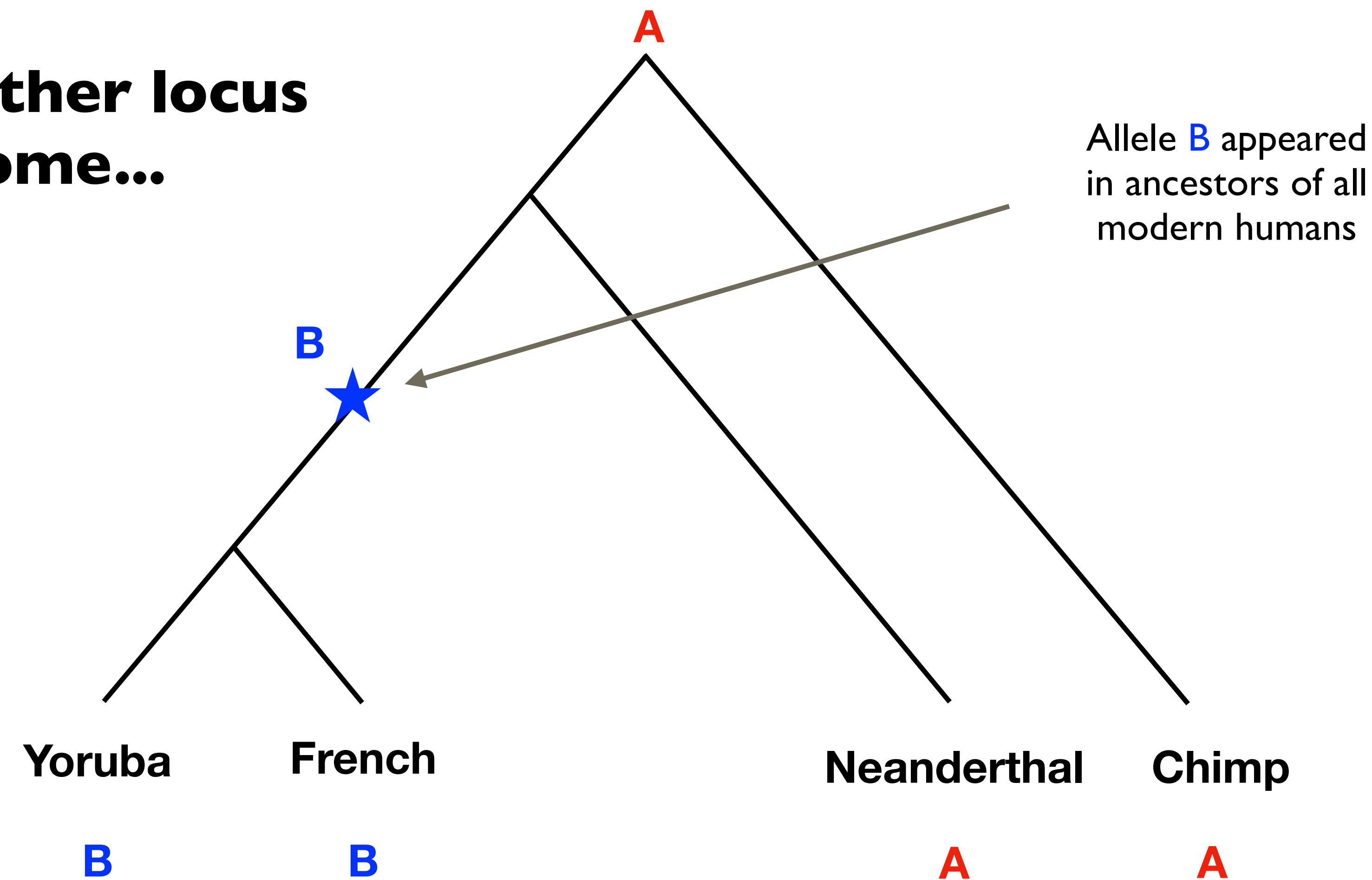


This “allele configuration” tells us nothing about introgression!

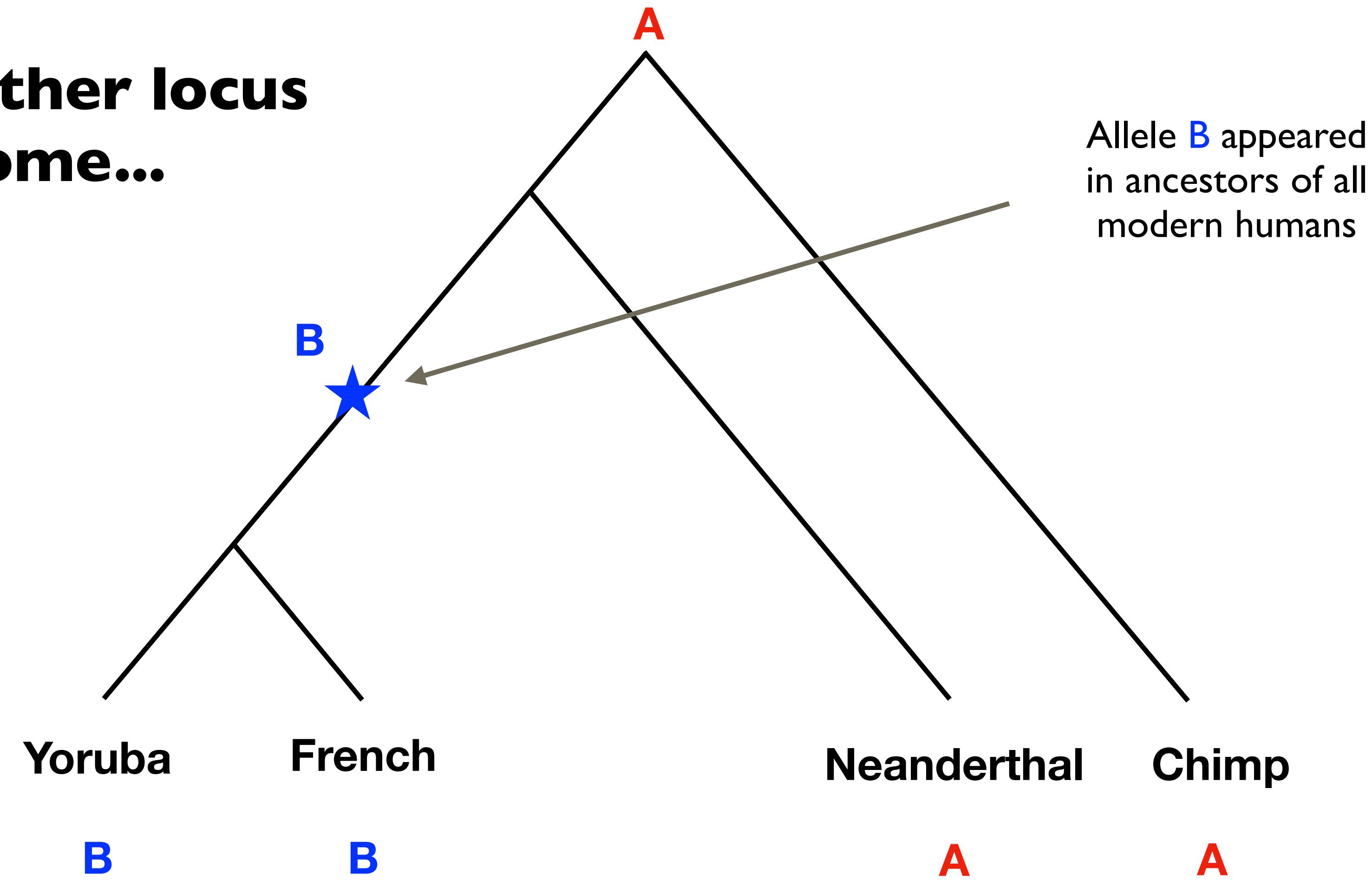
**At yet another locus
in the genome...**



**At yet another locus
in the genome...**

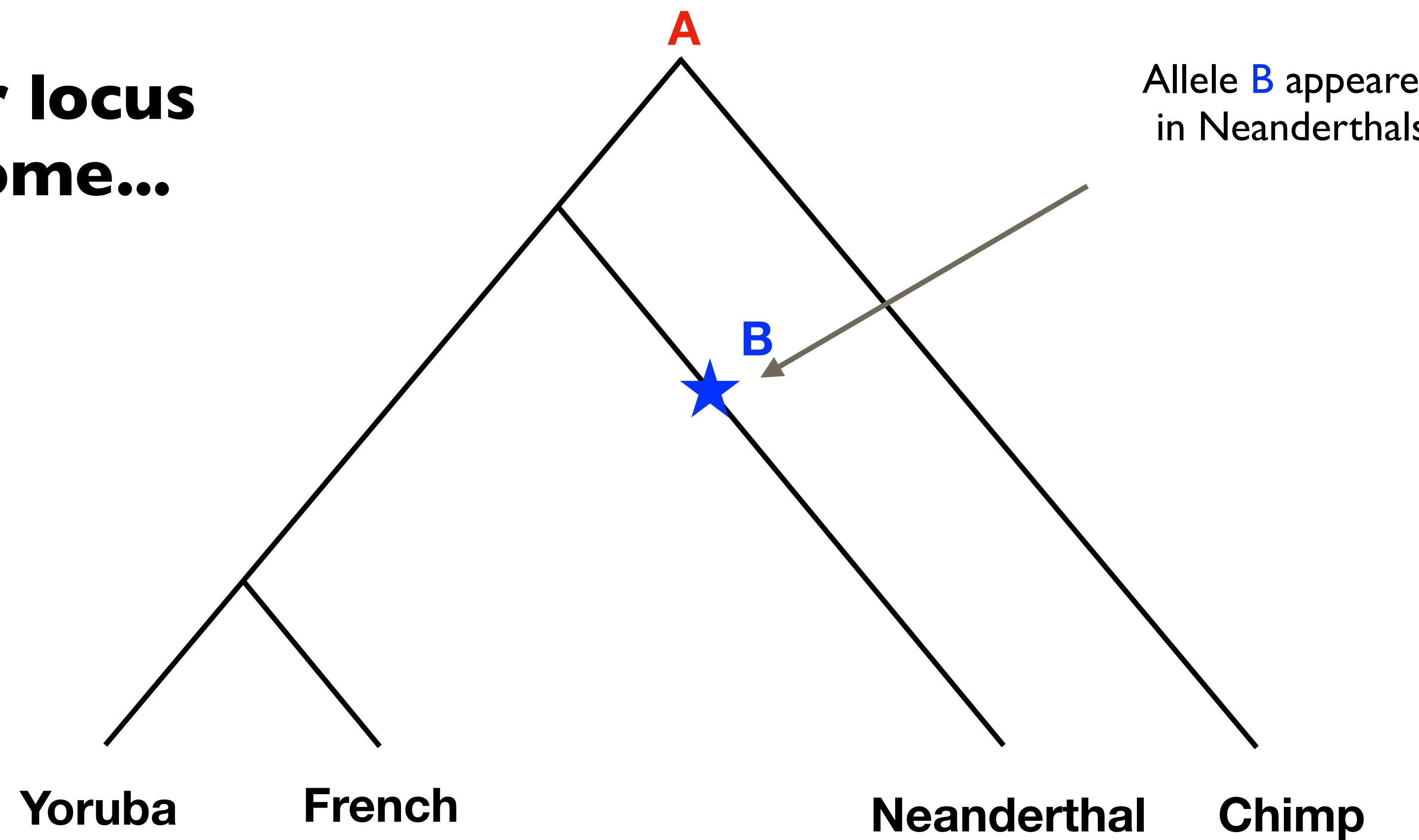


**At yet another locus
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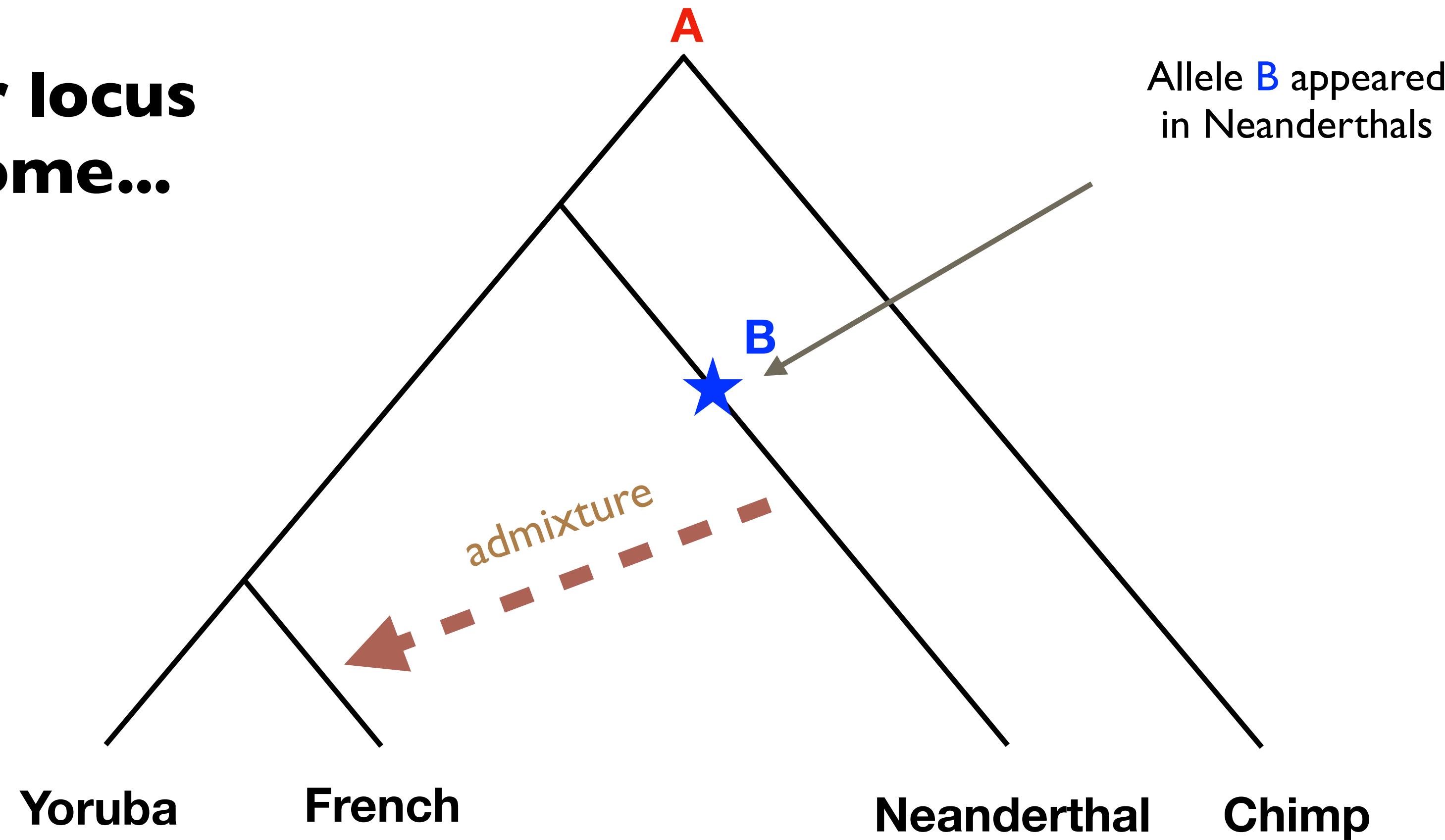


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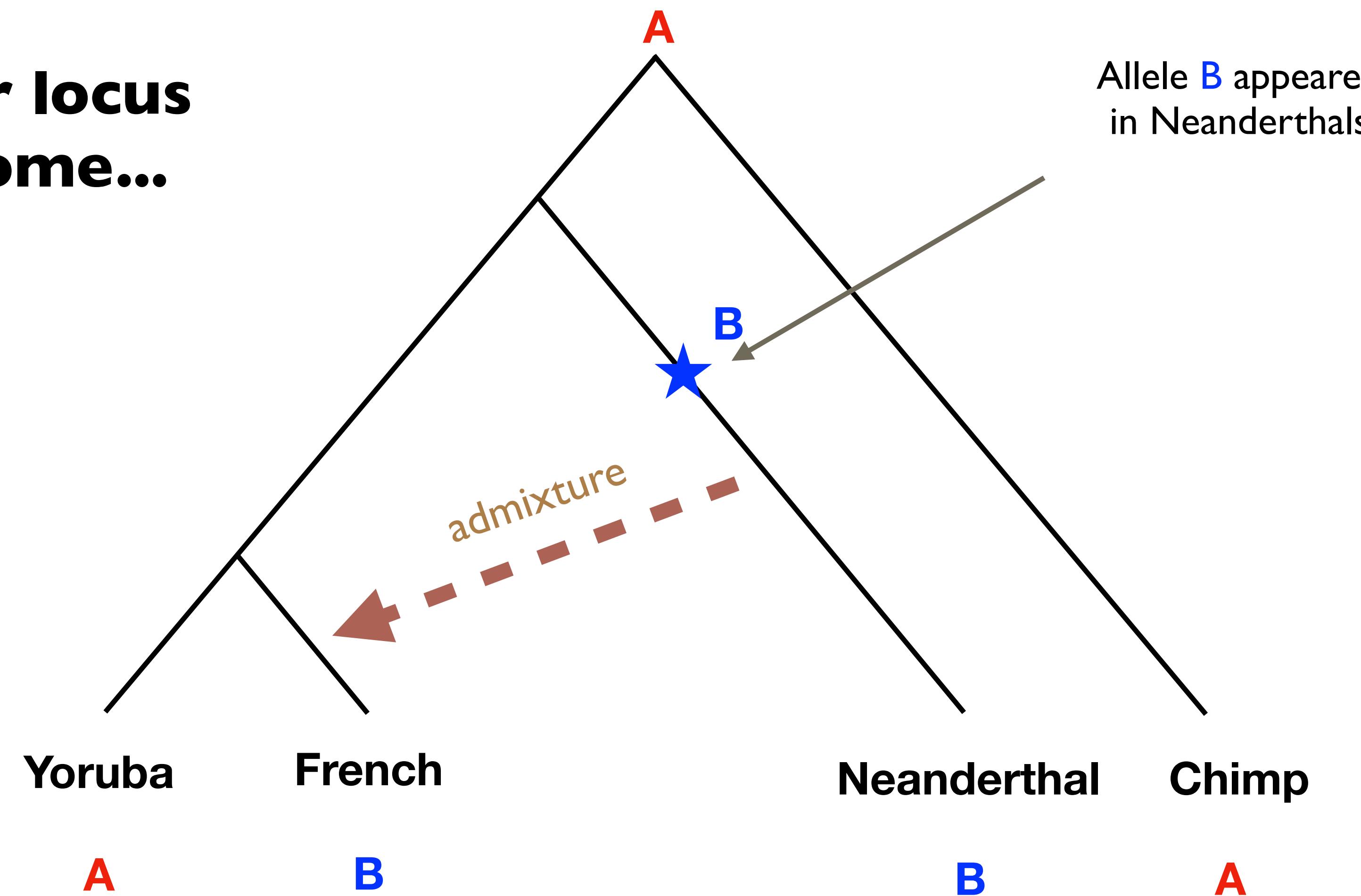
**At another locus
in the genome...**



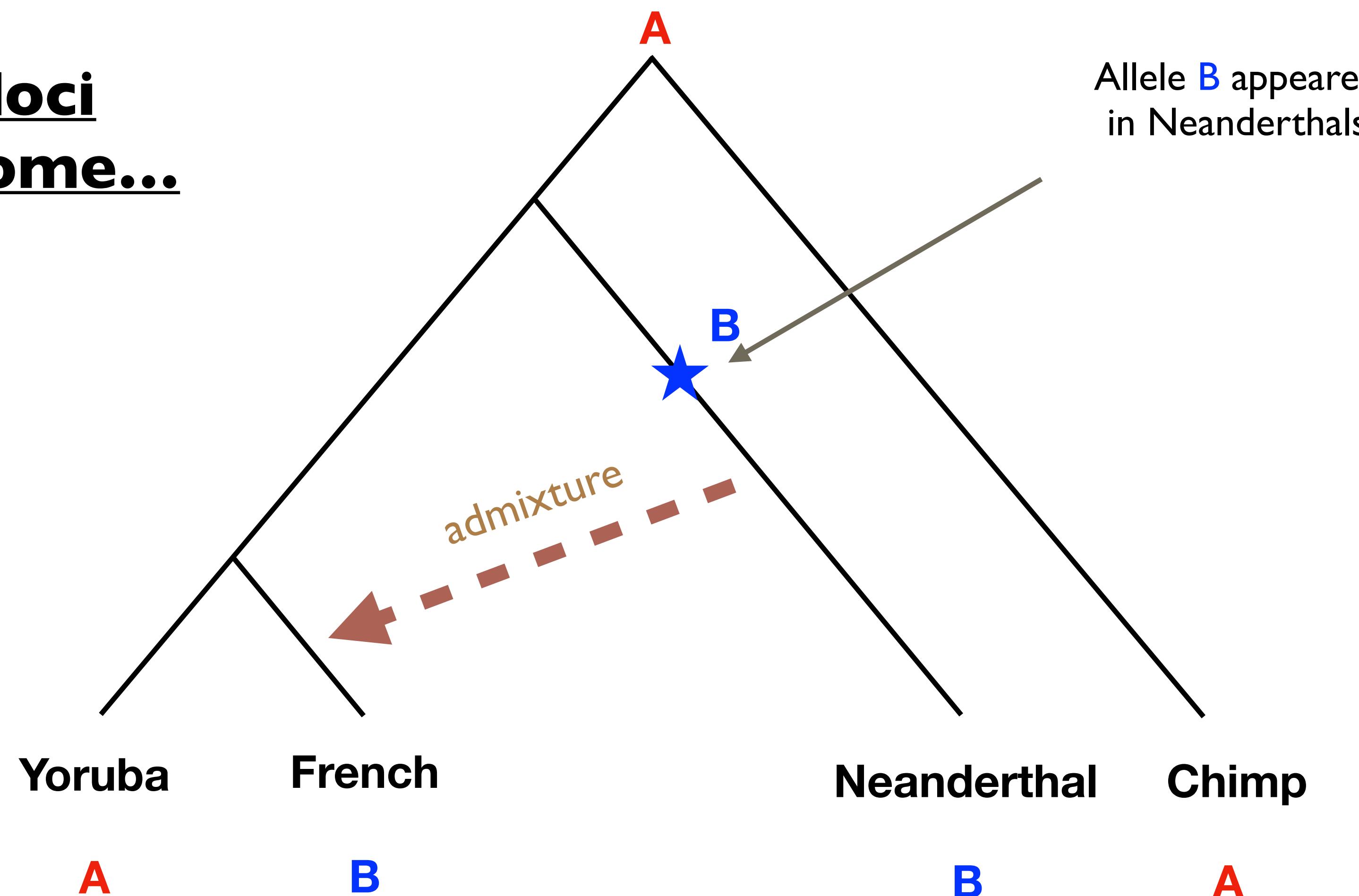
**At another locus
in the genome...**



**At another locus
in the genome...**



**Across all loci
in the genome...**



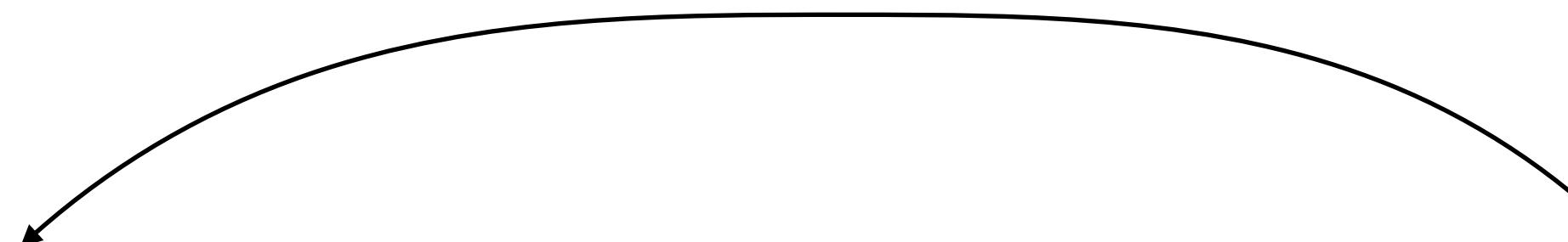
Introgression: *f4 statistic* = (# BABA - # ABBA) / # SNPs < 0

f_4 statistic quantifies allele configurations at only those sites which are “informative” of introgression

only **BABA** vs **ABBA** site patterns

(we ignore all others as “uninformative”)

Test of Neanderthal admixture in practice



$f_4(\text{Yoruba}, \mathbf{X}; \text{Neanderthal}, \text{Chimp}) \dots$ where $\mathbf{X} = \text{Dinka or French}$

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| X | #BABA | #ABBA | #total | f4 |
|---------------|-------|-------|---------|---------|
| French | 44409 | 46865 | 1436967 | -0.0017 |
| Dinka | 43025 | 43182 | 1436978 | -0.0001 |

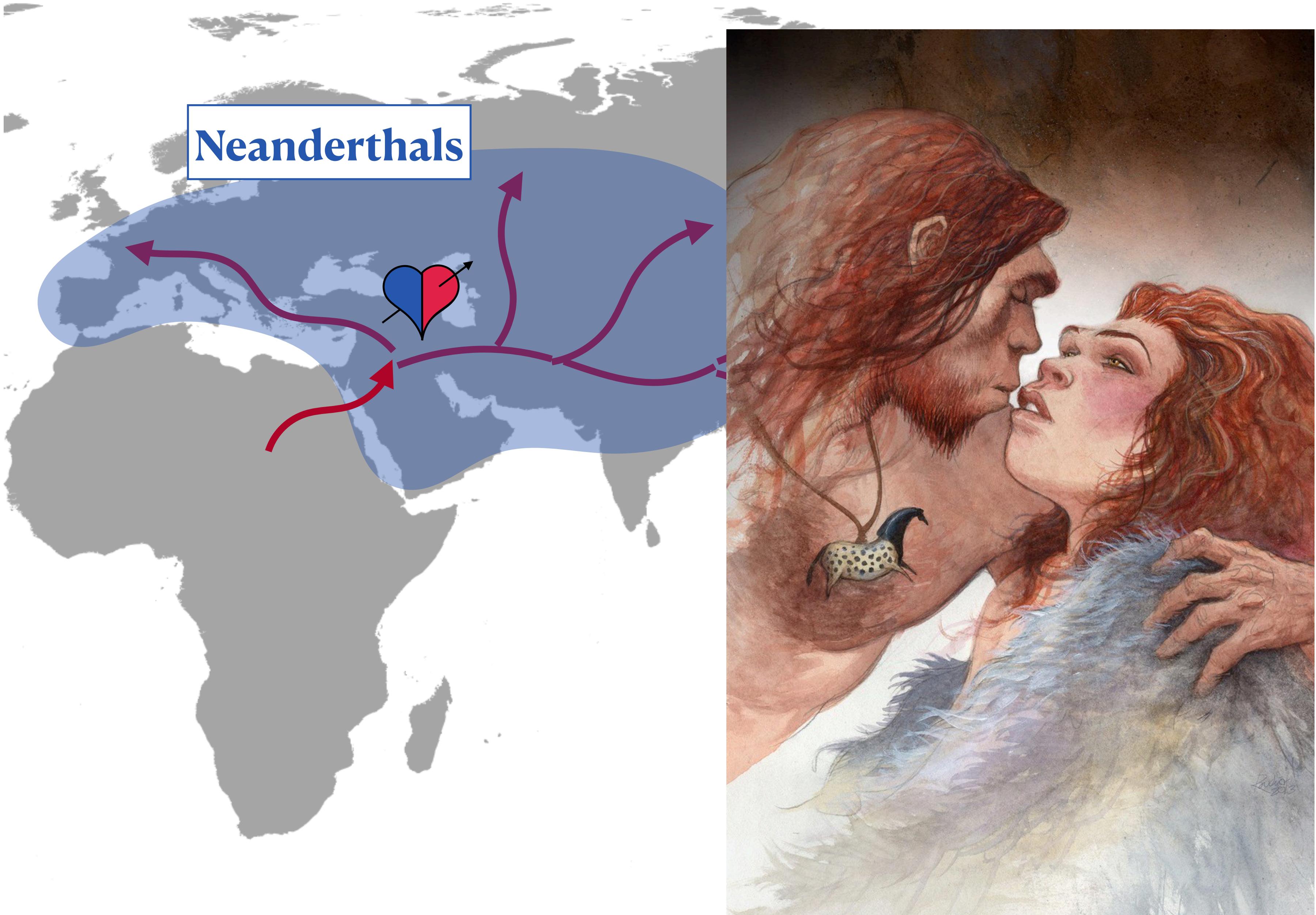
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| X | #BABA | #ABBA | #total | f4 | Z |
|---------------|-------|-------|---------|---------|-------|
| French | 44409 | 46865 | 1436967 | -0.0017 | -7.83 |
| Dinka | 43025 | 43182 | 1436978 | -0.0001 | -0.72 |

$|Z \text{ score}| > 3$ considered significant



What about the proportion of Neanderthal ancestry?

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We compare the rate of allele sharing of X and a Neanderthal, with the rate of sharing between two Neanderthals (Altai and Vindija).

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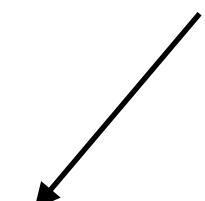


| X | #BABA | #ABBA | #sites | f4 |
|----------------|-------|--------|---------|----------------|
| French | 44409 | 46865 | 1436967 | -0.0017 |
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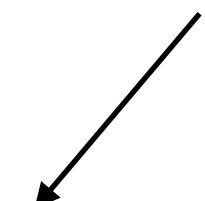


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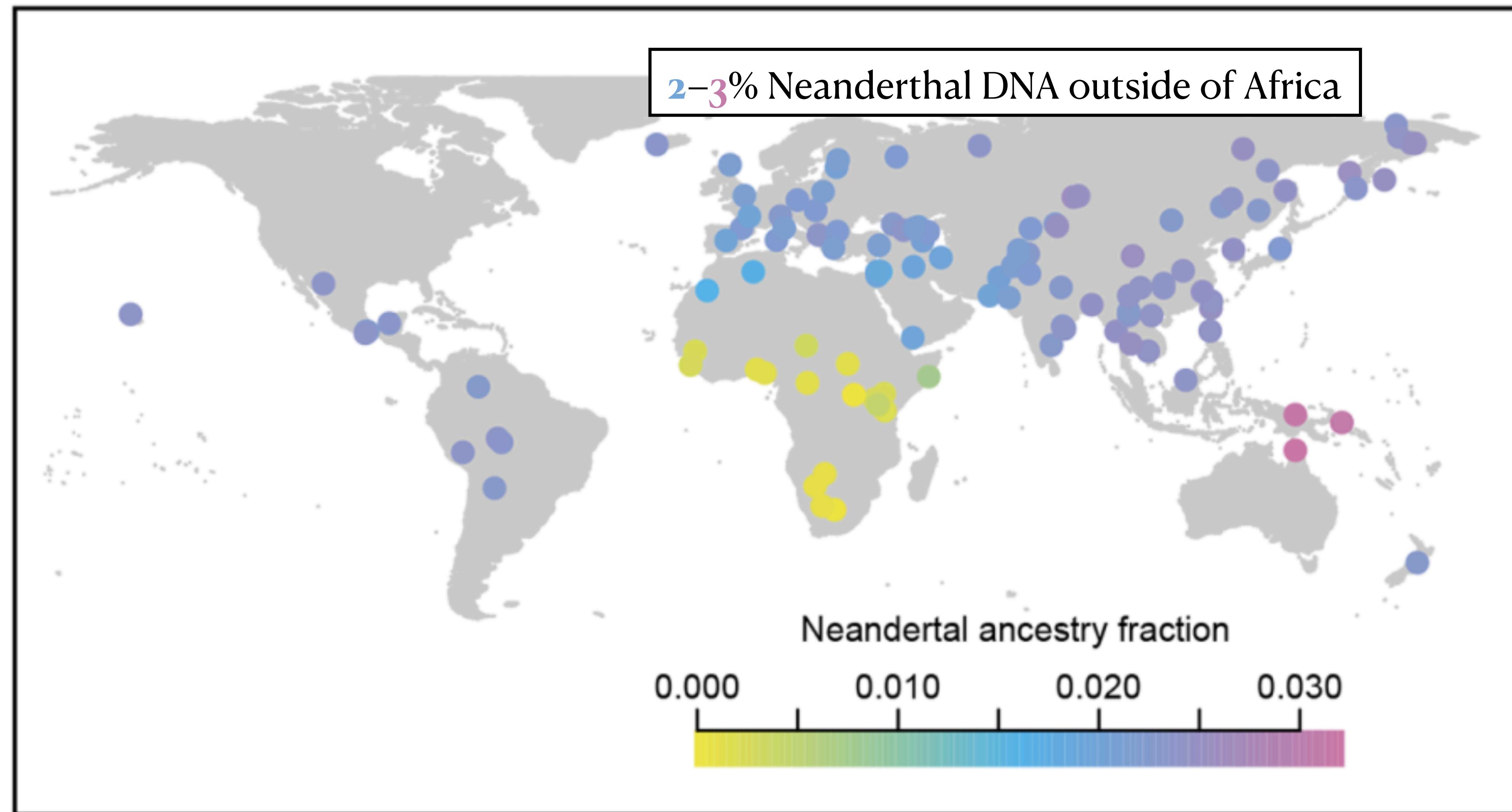
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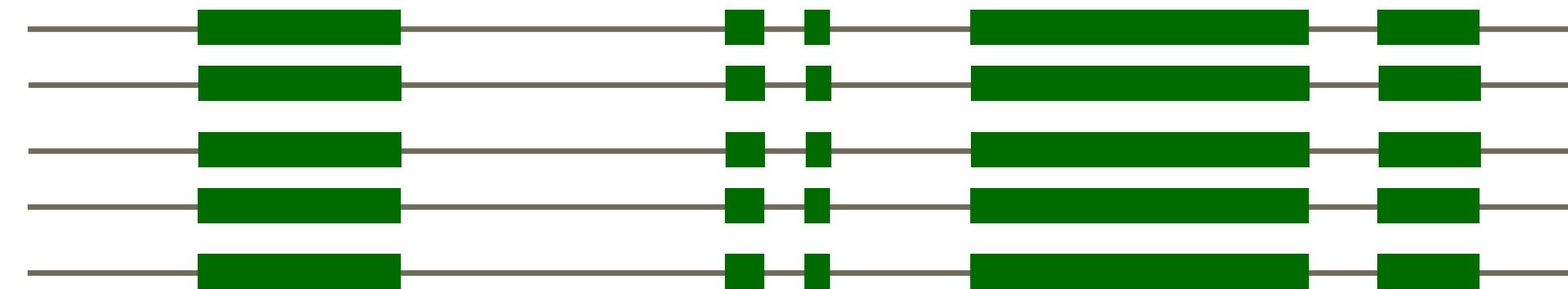


| X | #BABA | #ABBA | #sites | f4 | ratio |
|---------|-------|--------|---------|---------|-------|
| French | 44409 | 46865 | 1436967 | -0.0017 | 2.2% |
| Dinka | 43025 | 43182 | 1436978 | -0.0001 | 0.1% |
| Vindija | 7337 | 118956 | 1436703 | -0.0777 | 100% |

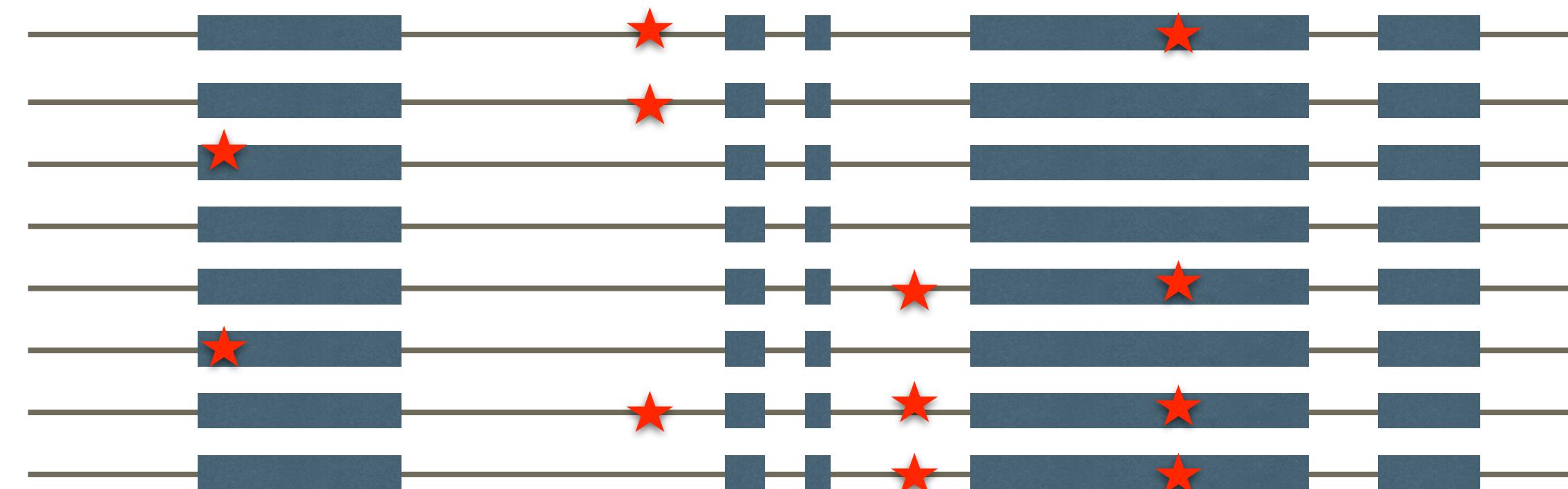
Proportion of Neanderthal DNA in humans today



We can go from detecting SNPs of Neanderthal origin...



genomes of a
reference population
(Yoruba)



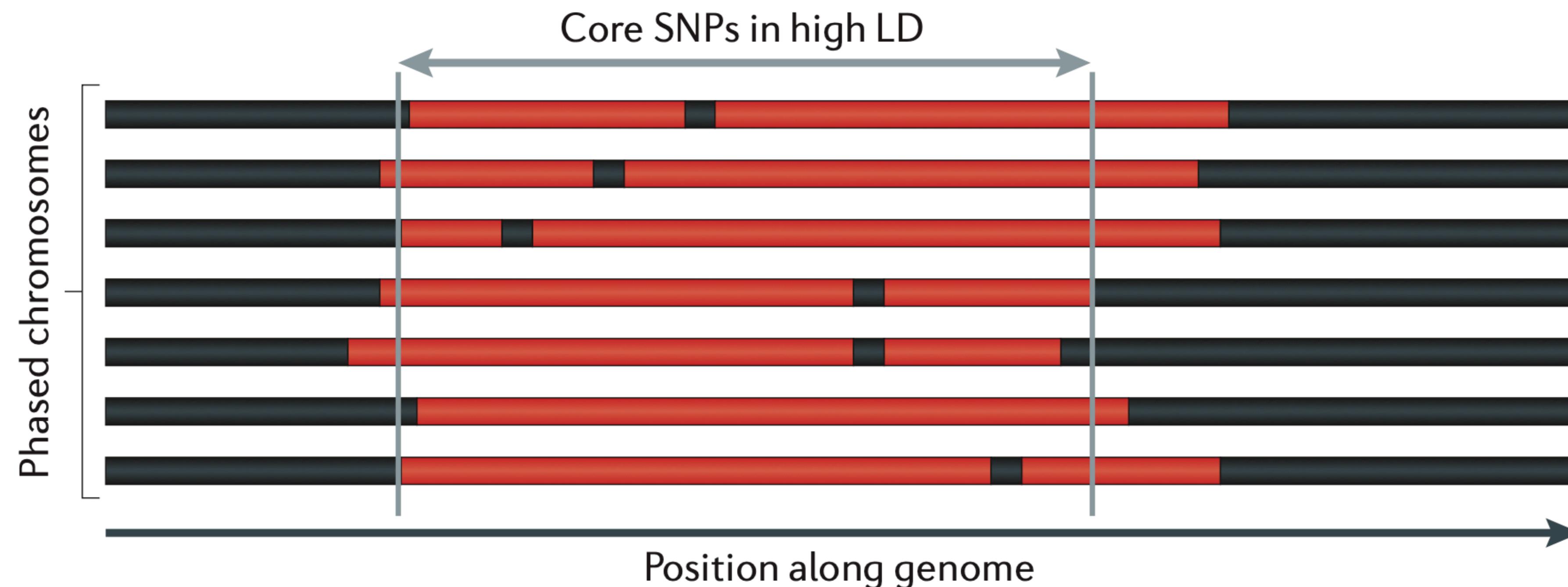
genomes of a
target population
(Europeans)



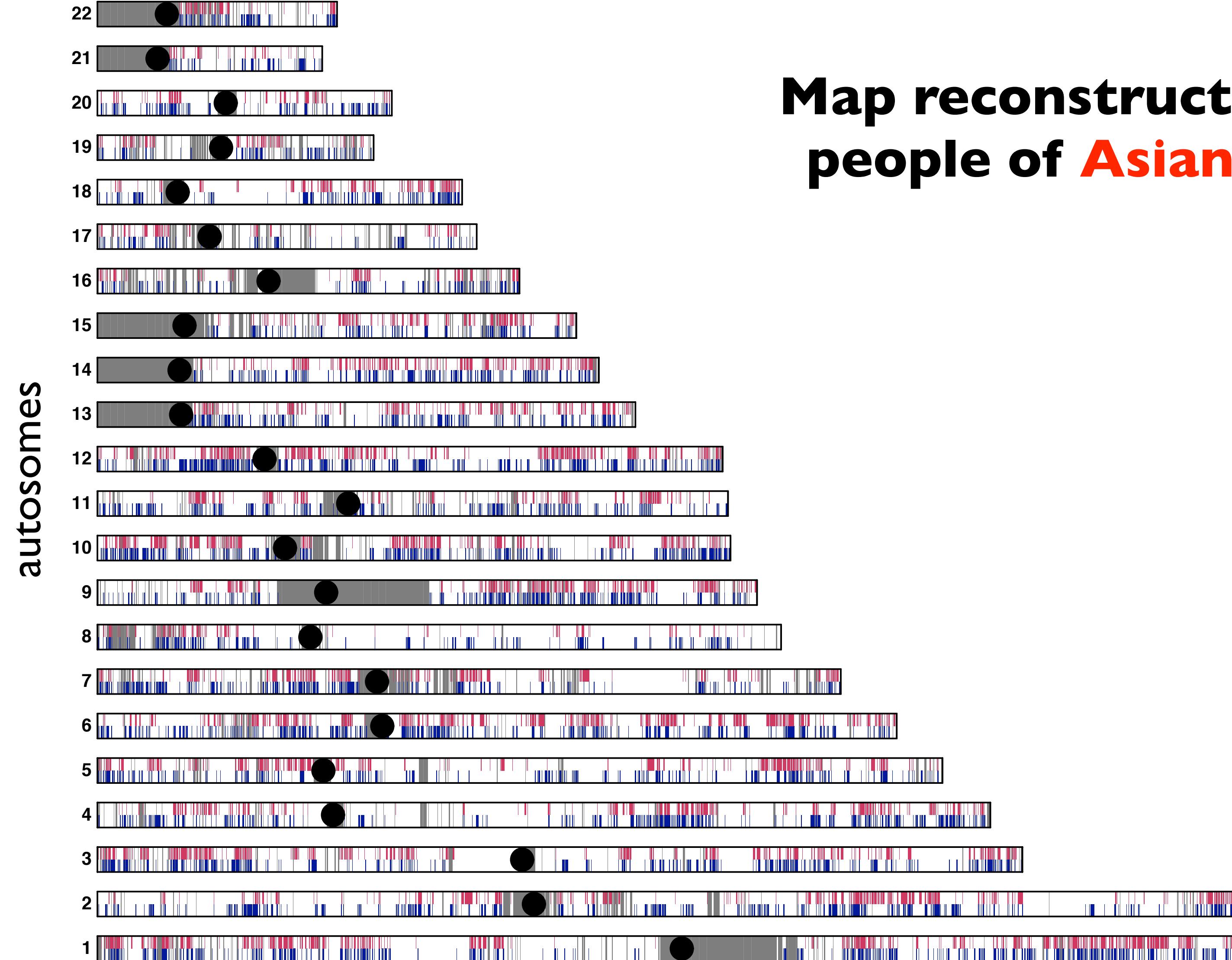
Neanderthal
genome

★ = Neanderthal-derived mutation

...to continuous segments of Neanderthal DNA

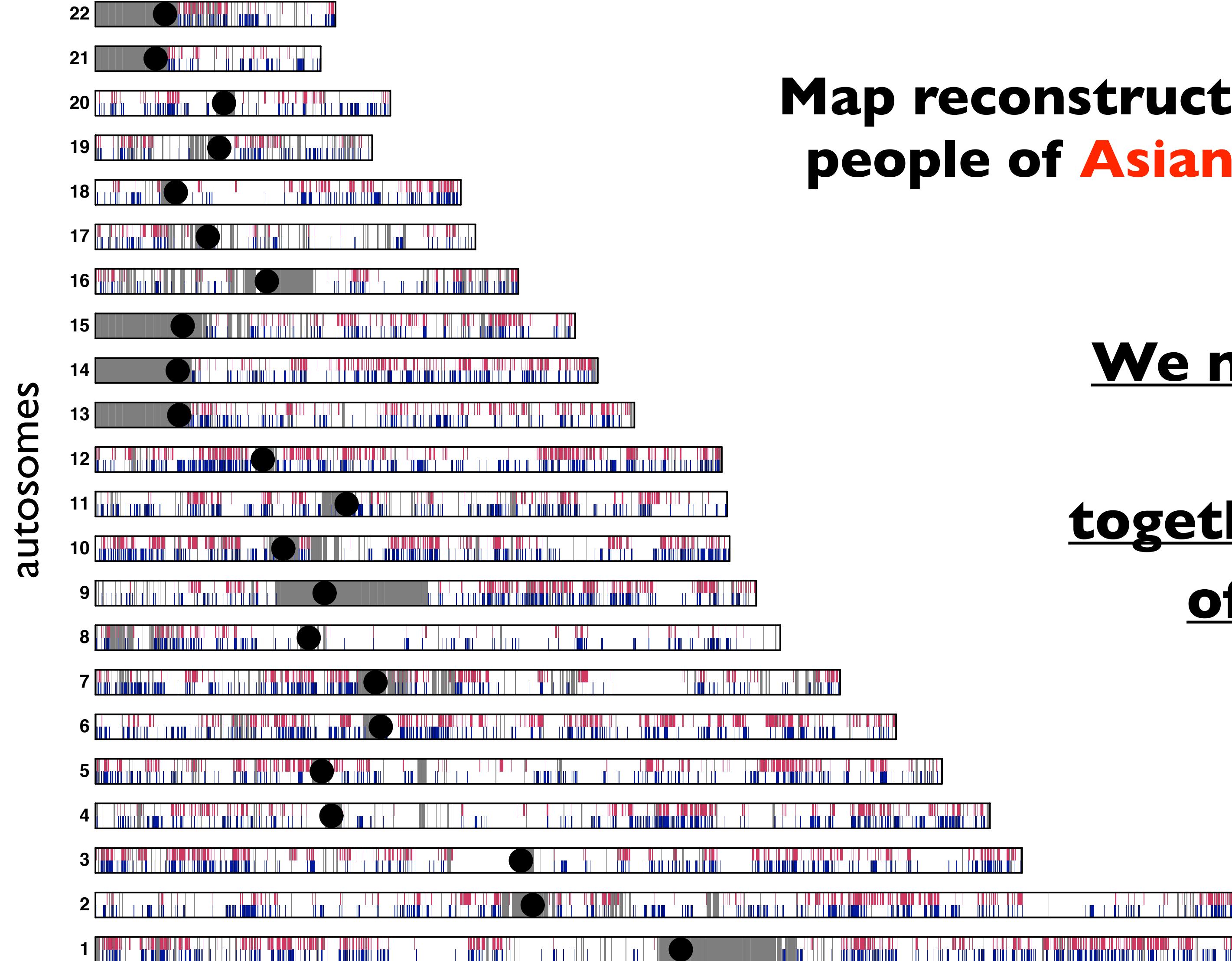


Genomic maps of Neanderthal DNA today



Map reconstructed from genomes of 700 people of Asian and European ancestry.

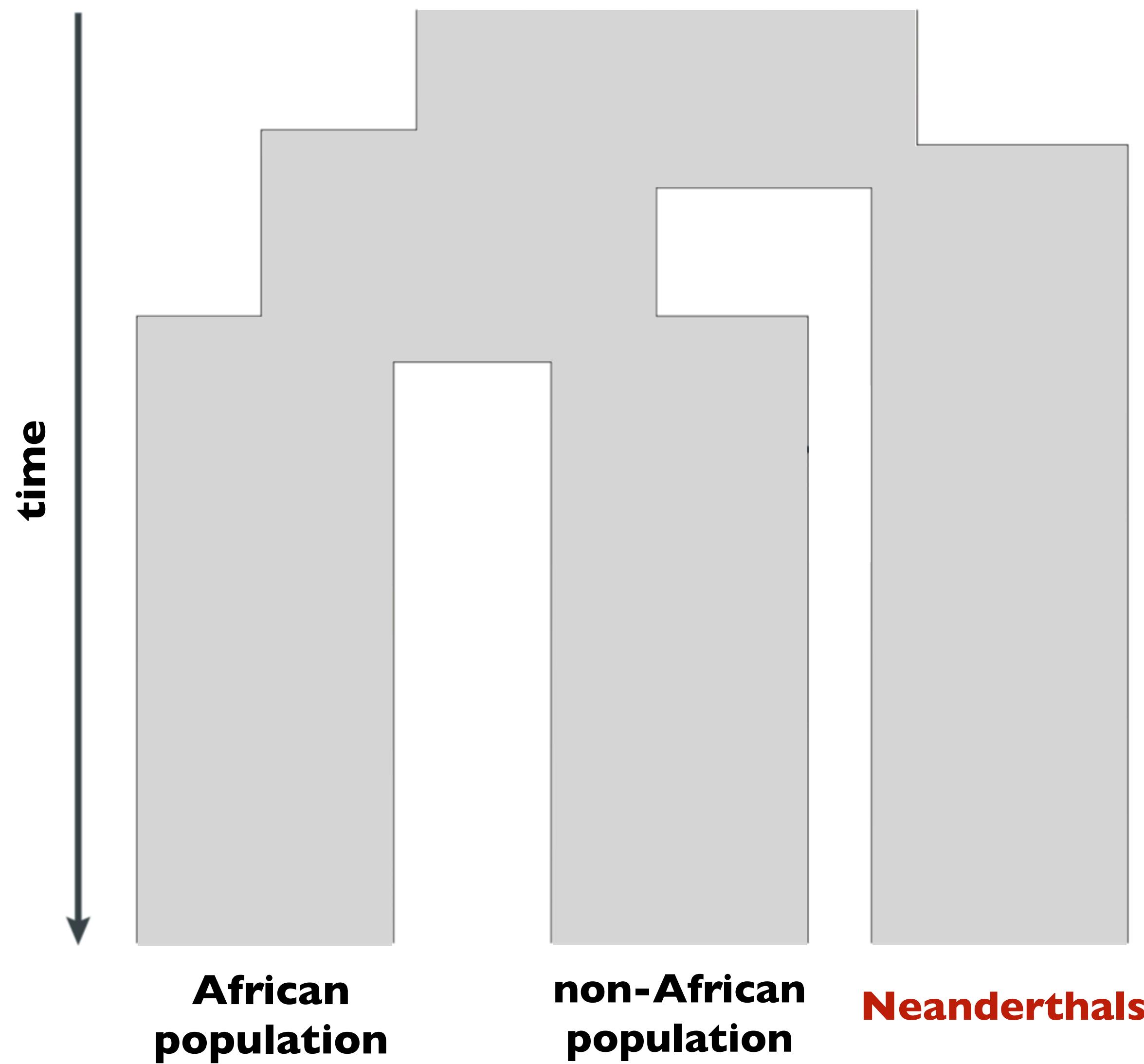
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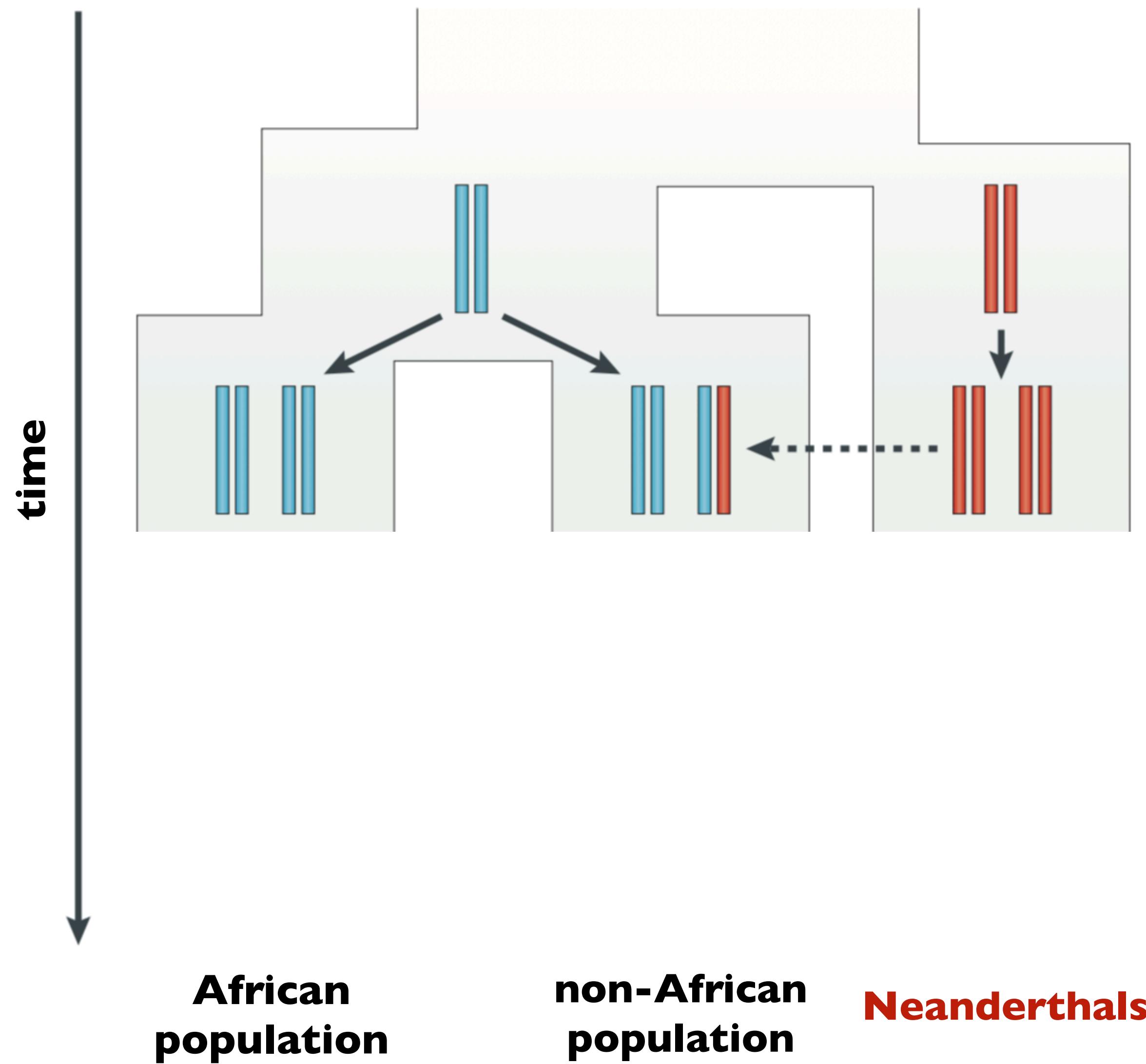
Map reconstructed from genomes of 700 people of **Asian and **European** ancestry.**

We may each have only 2% of Neanderthal DNA, but together we carry around 40% of a Neanderthal genome!

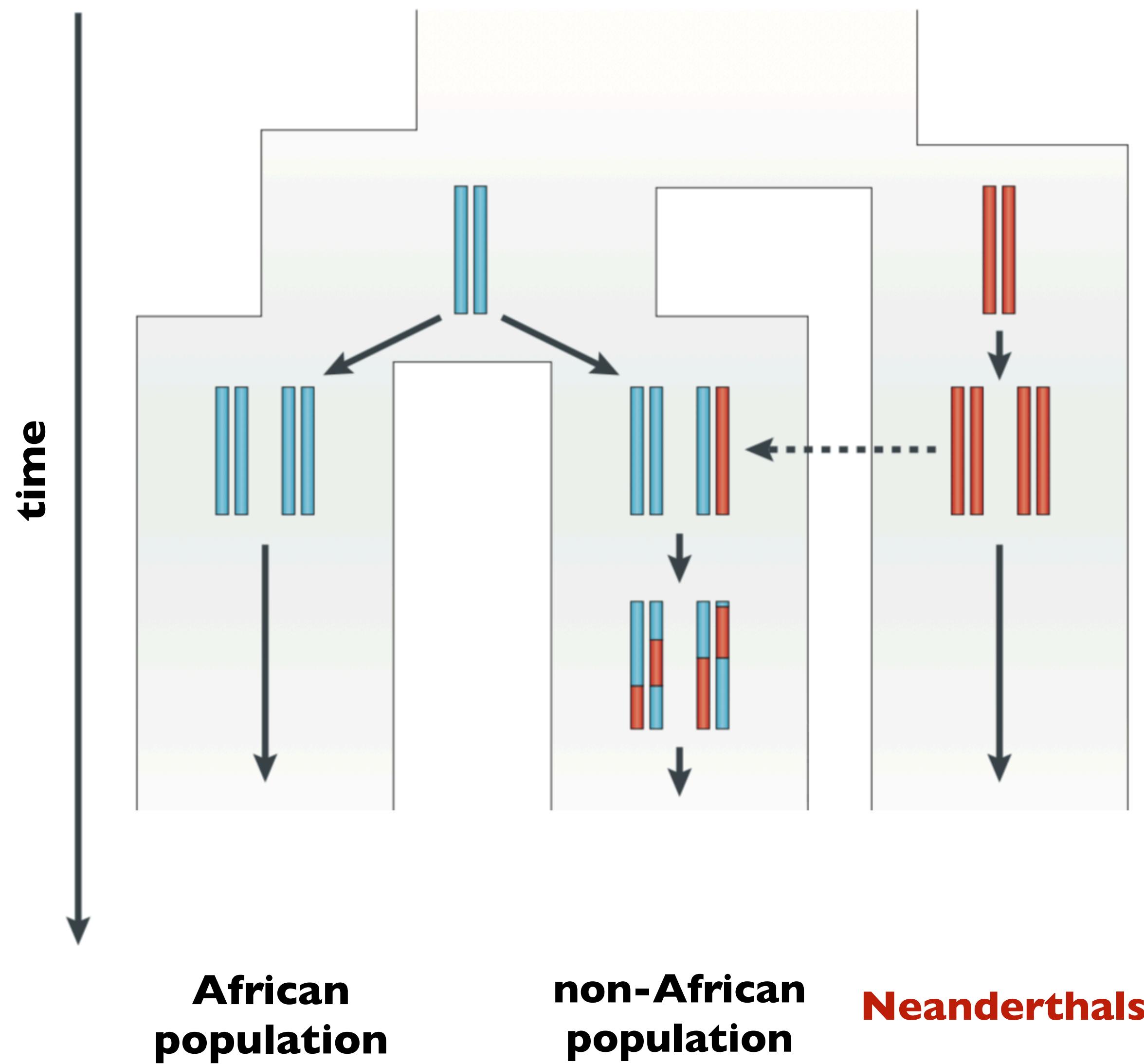
When did introgression happen?



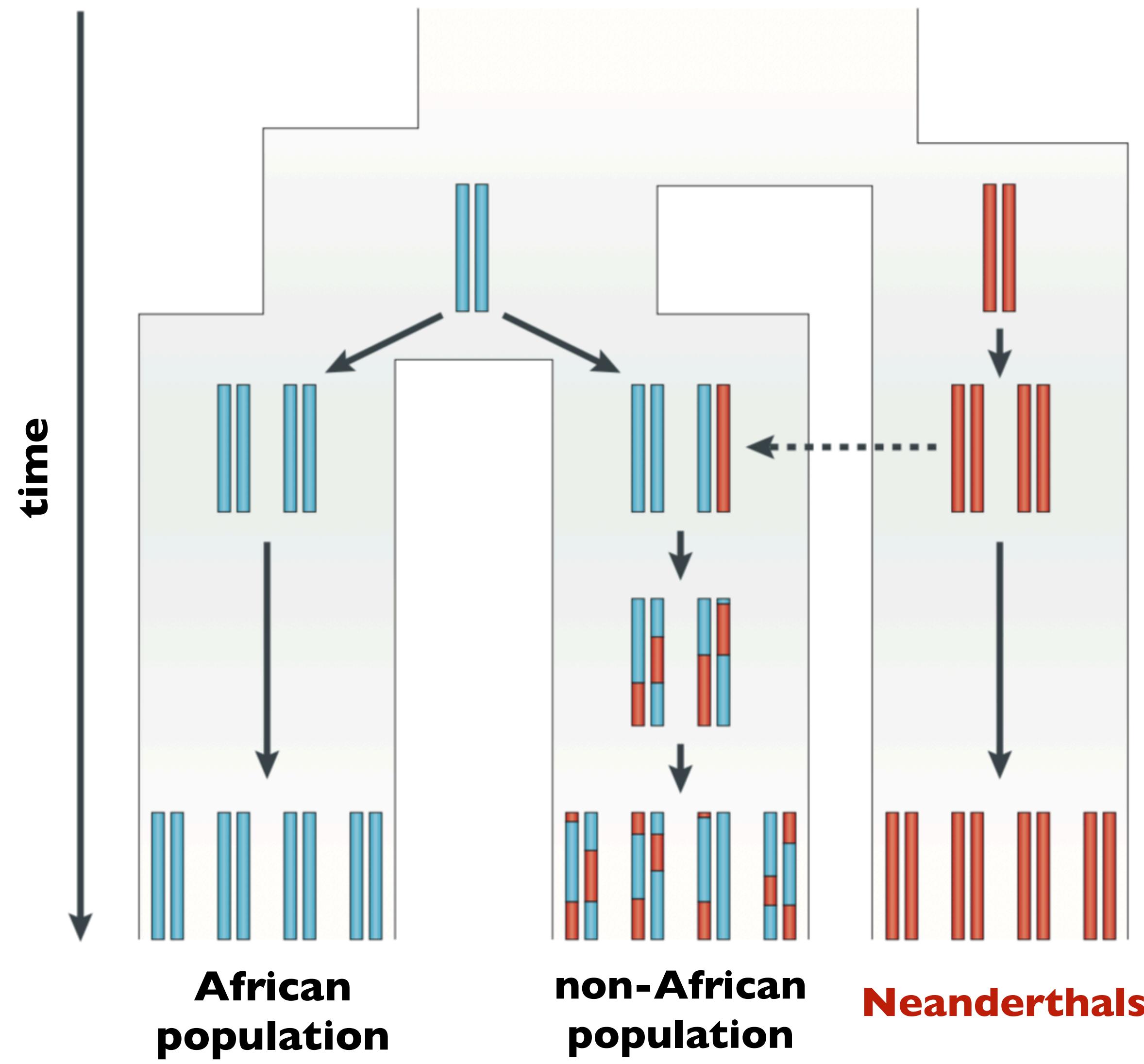
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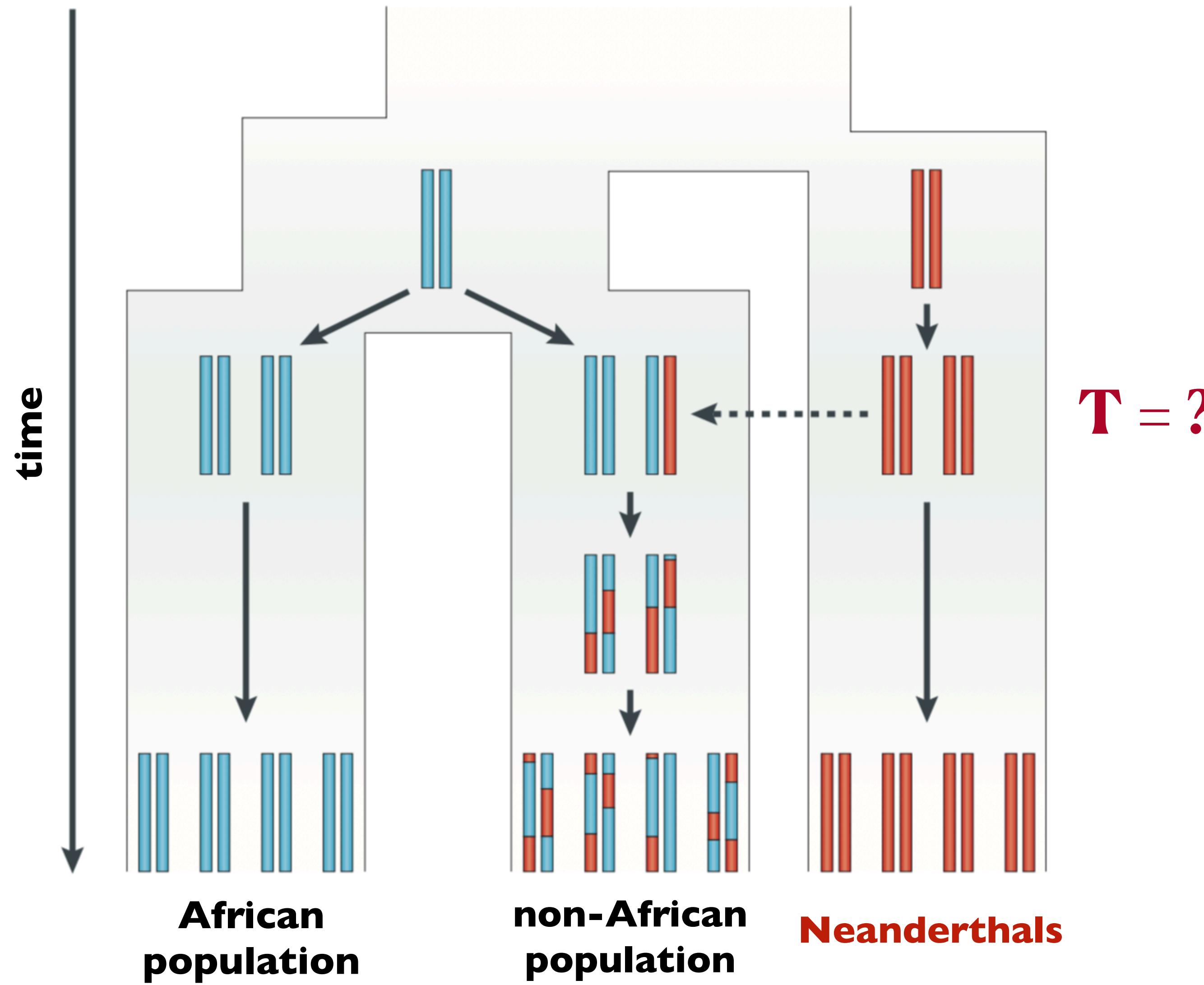
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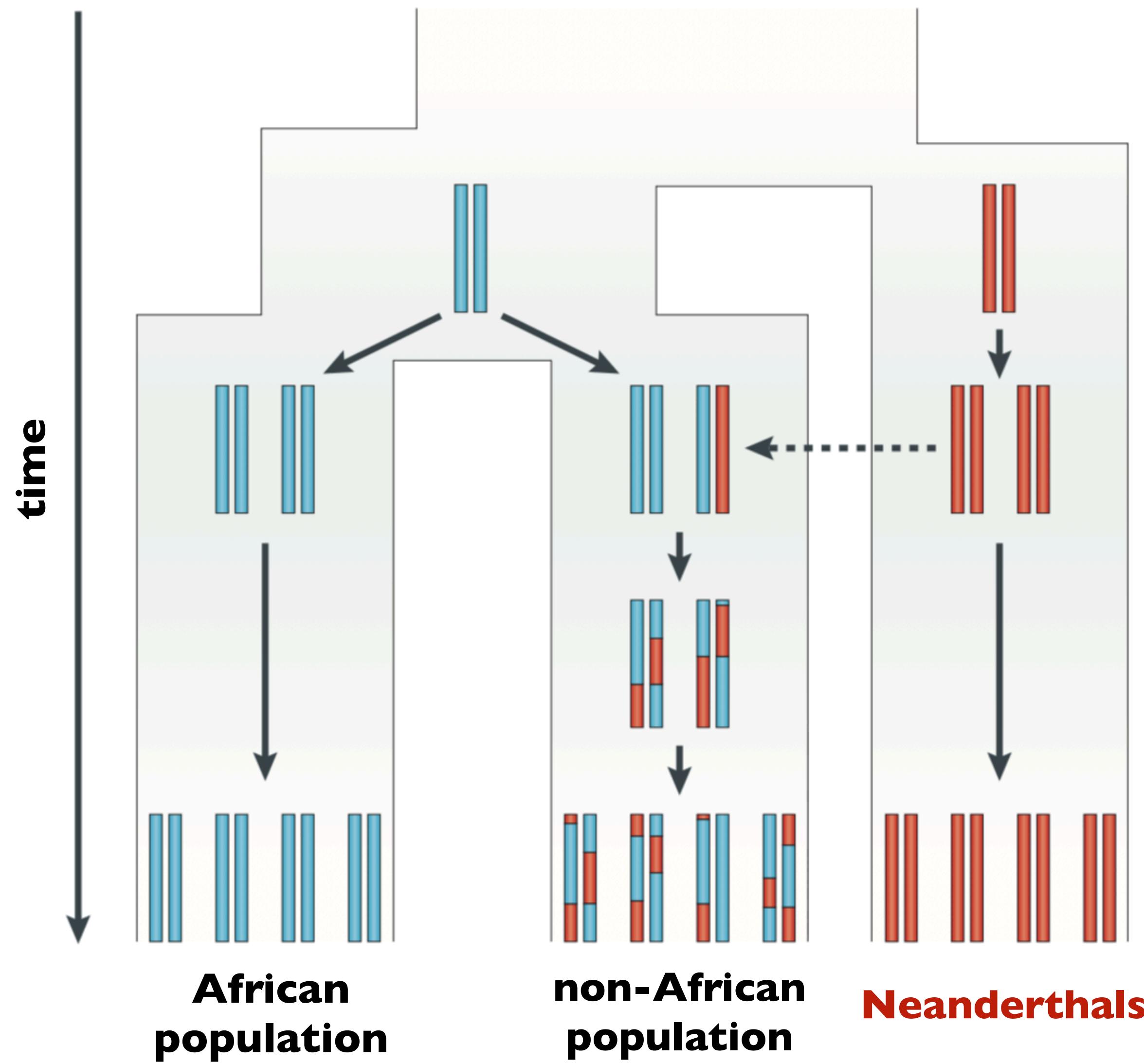
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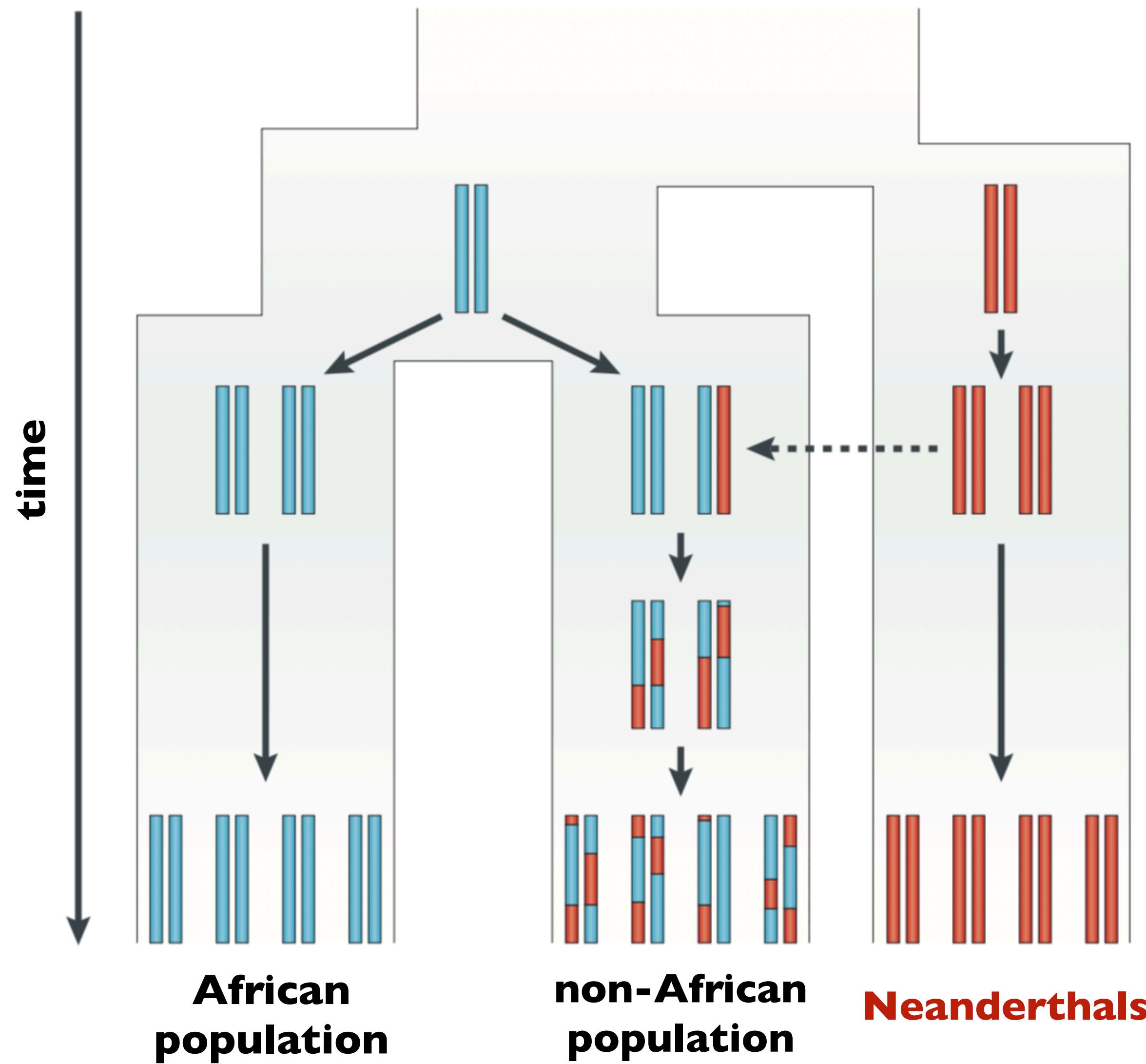


When did introgression happen?



Haplotypes carrying Neanderthal alleles get shorter over time due to recombination (statistically “regularly”).

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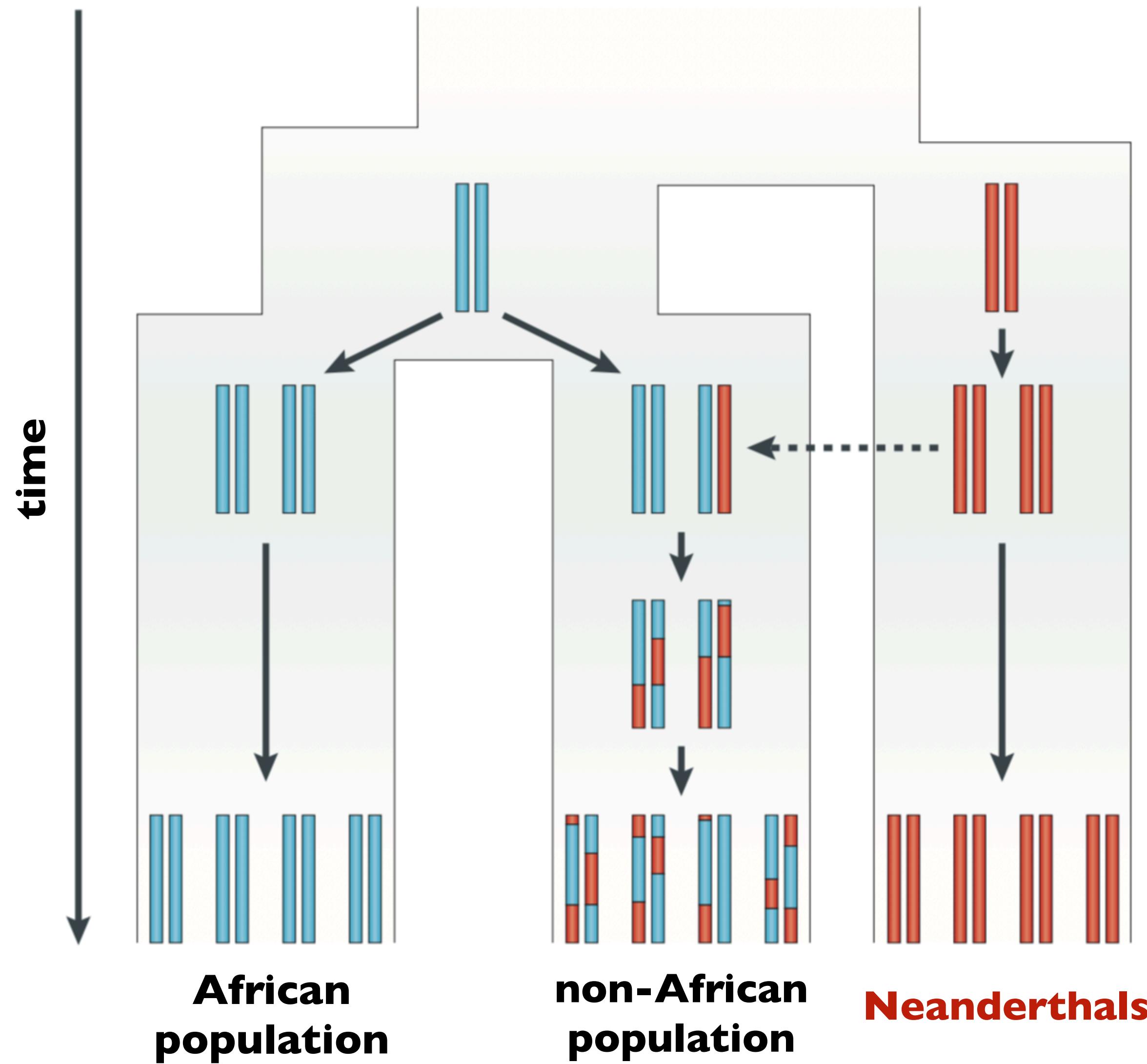


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$T = ?$

Expected length of these haplotypes follows the principle of exponential decay.

When did introgression happen?



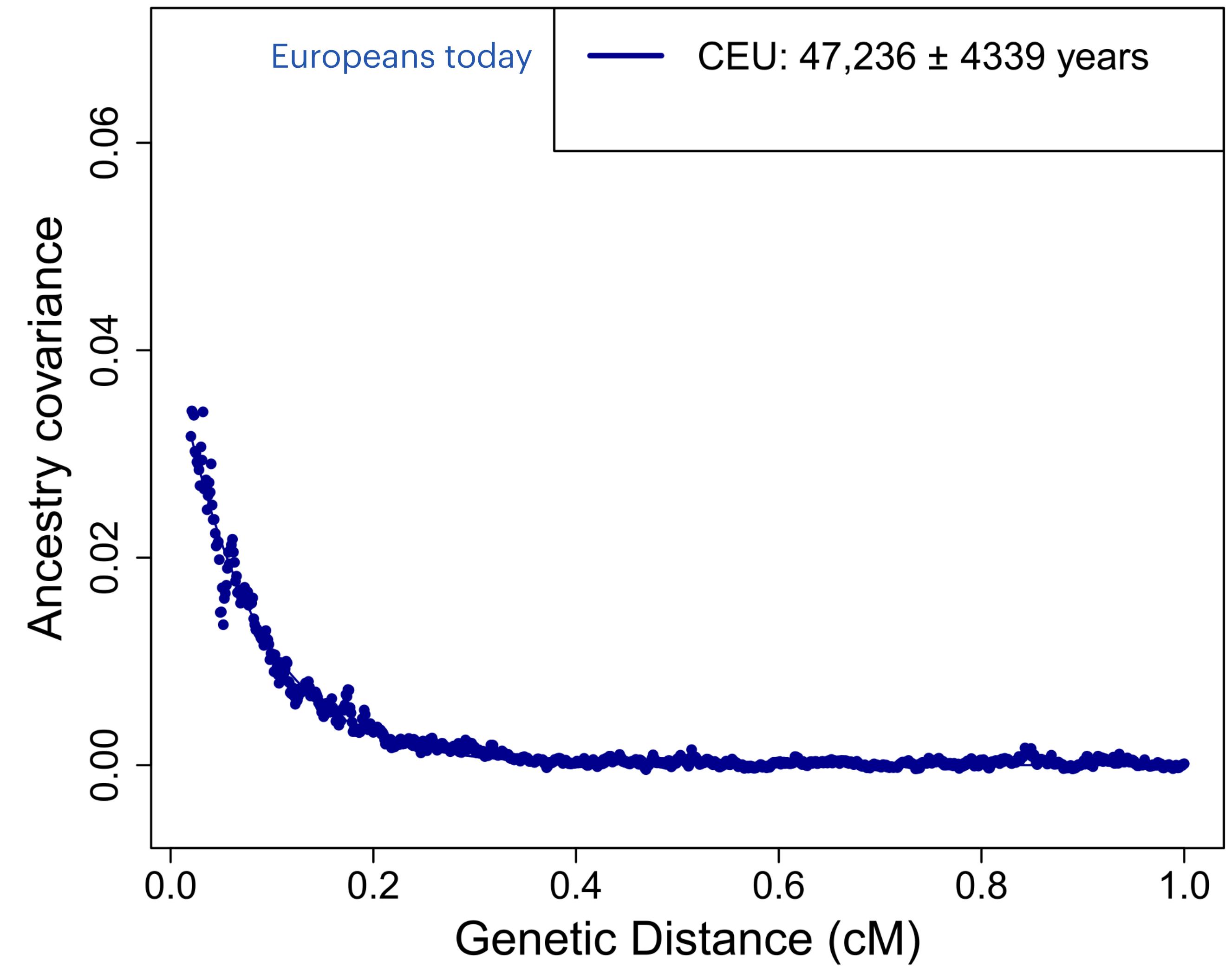
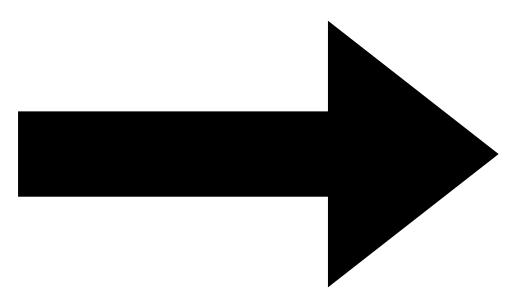
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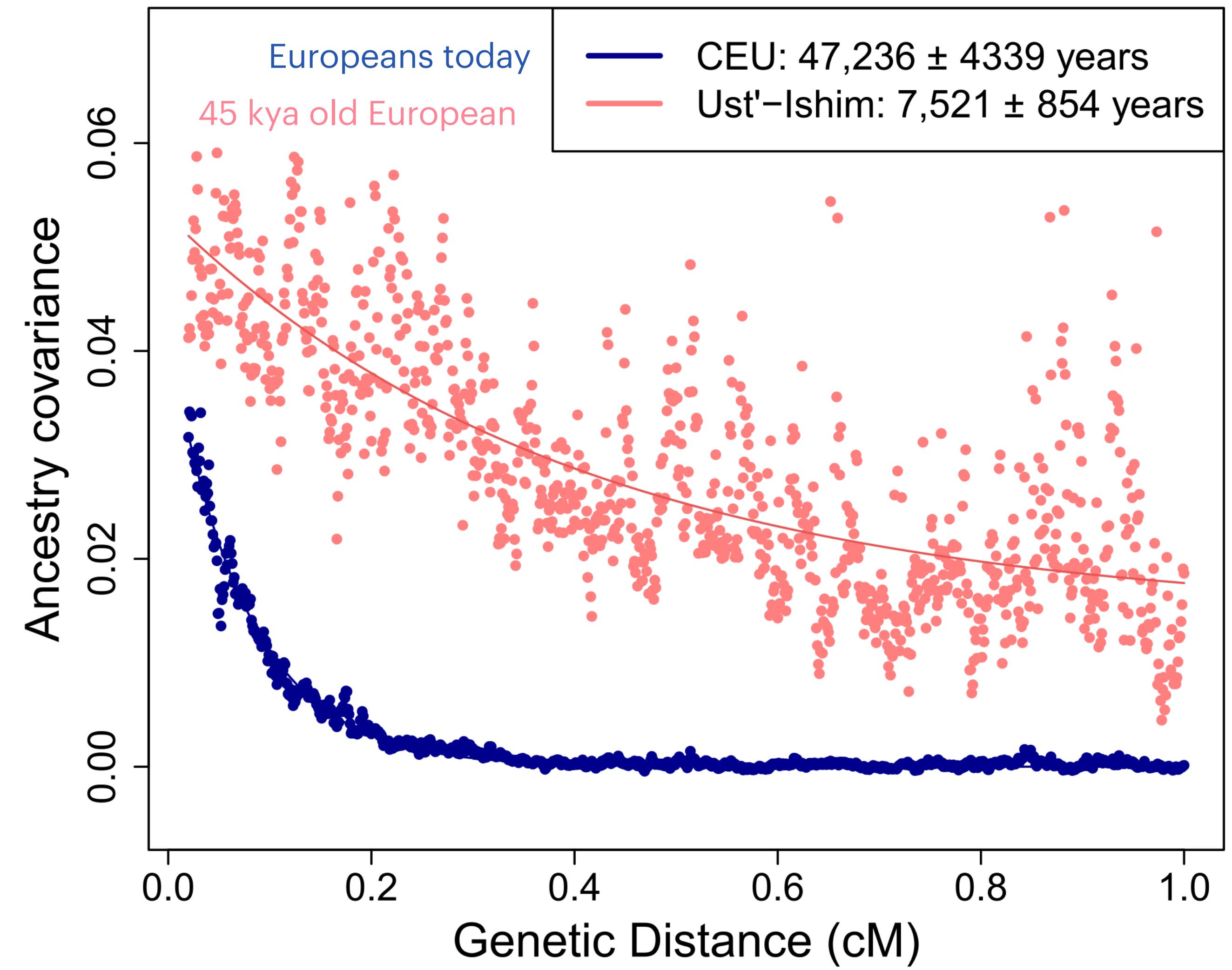
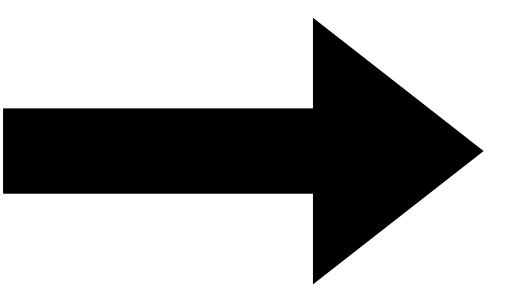
Expected length of these haplotypes follows the principle of exponential decay.

The rate of this decay gives us the time since introgression.

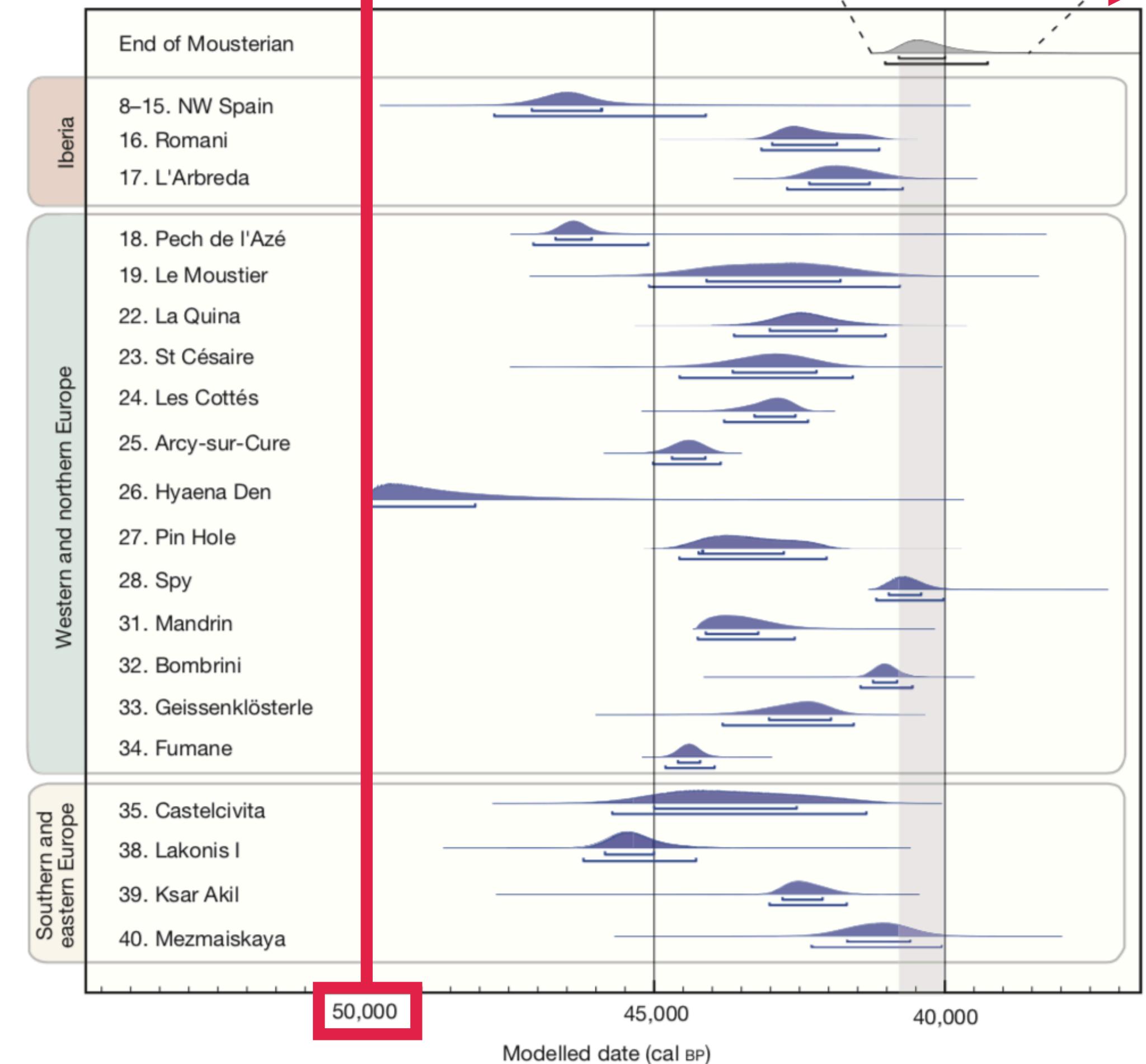
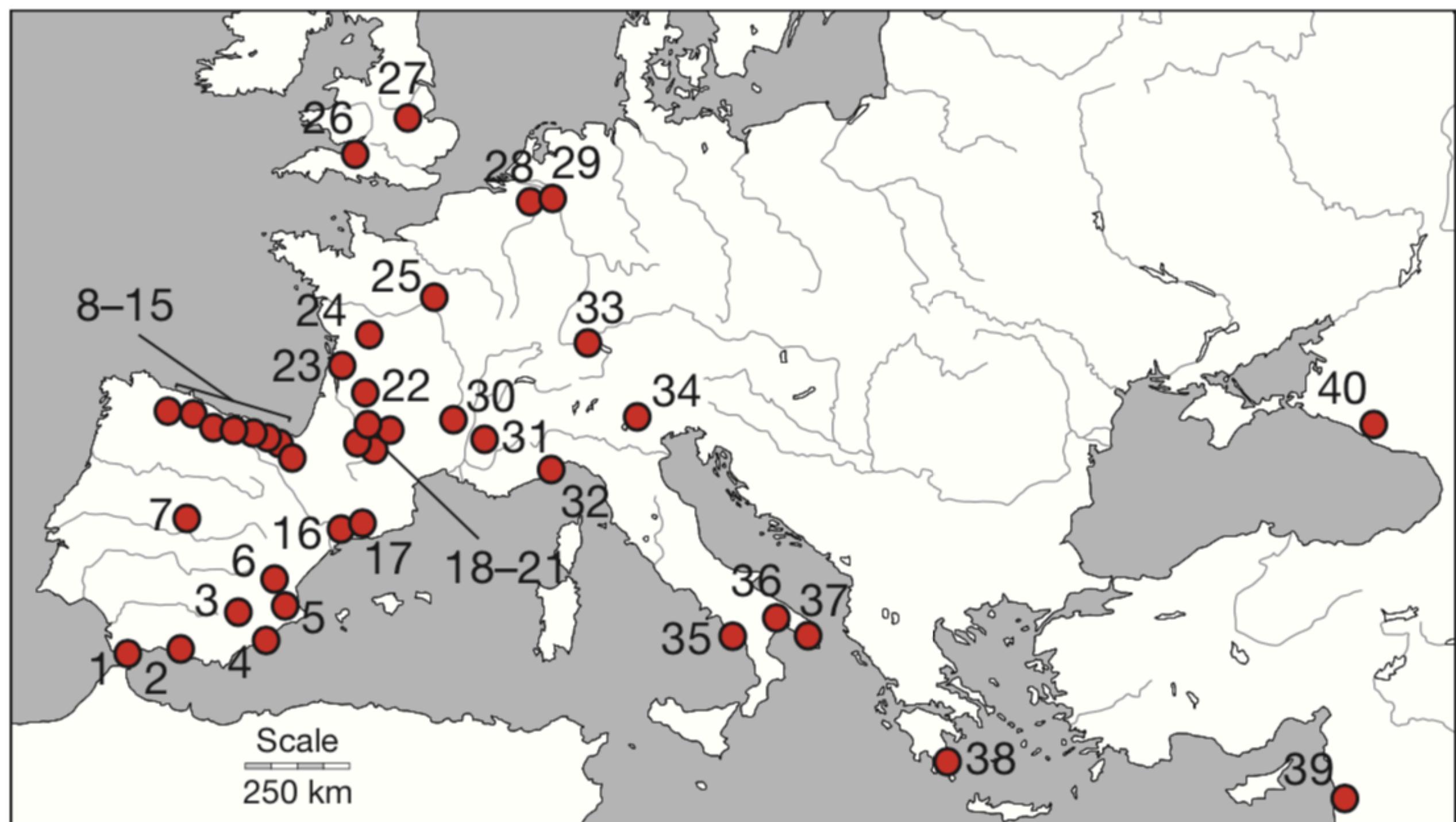
Introgression happened around 50 thousand years ago



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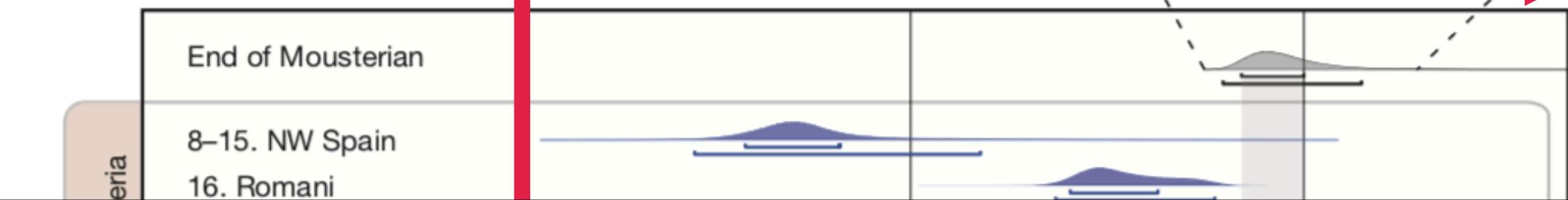


anatomically modern humans in Europe

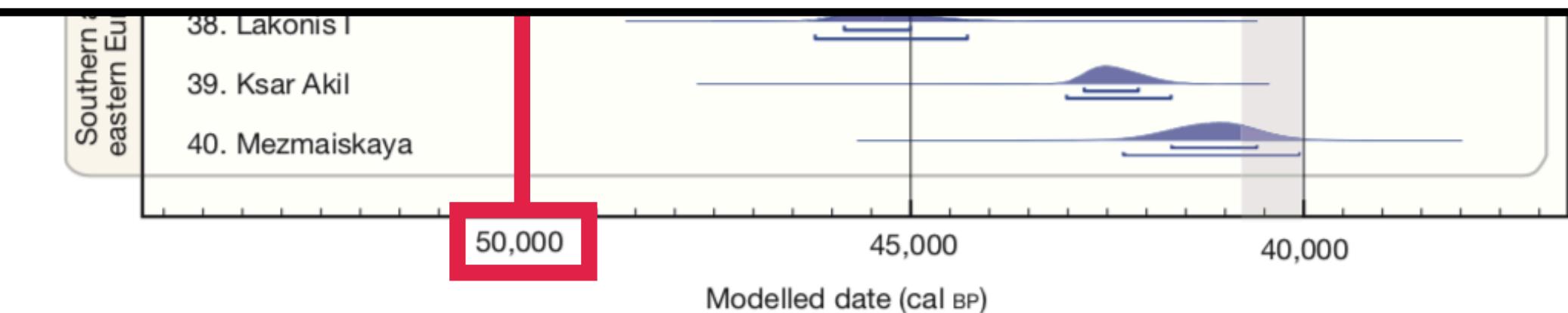


Higham et al. (Nature, 2014)

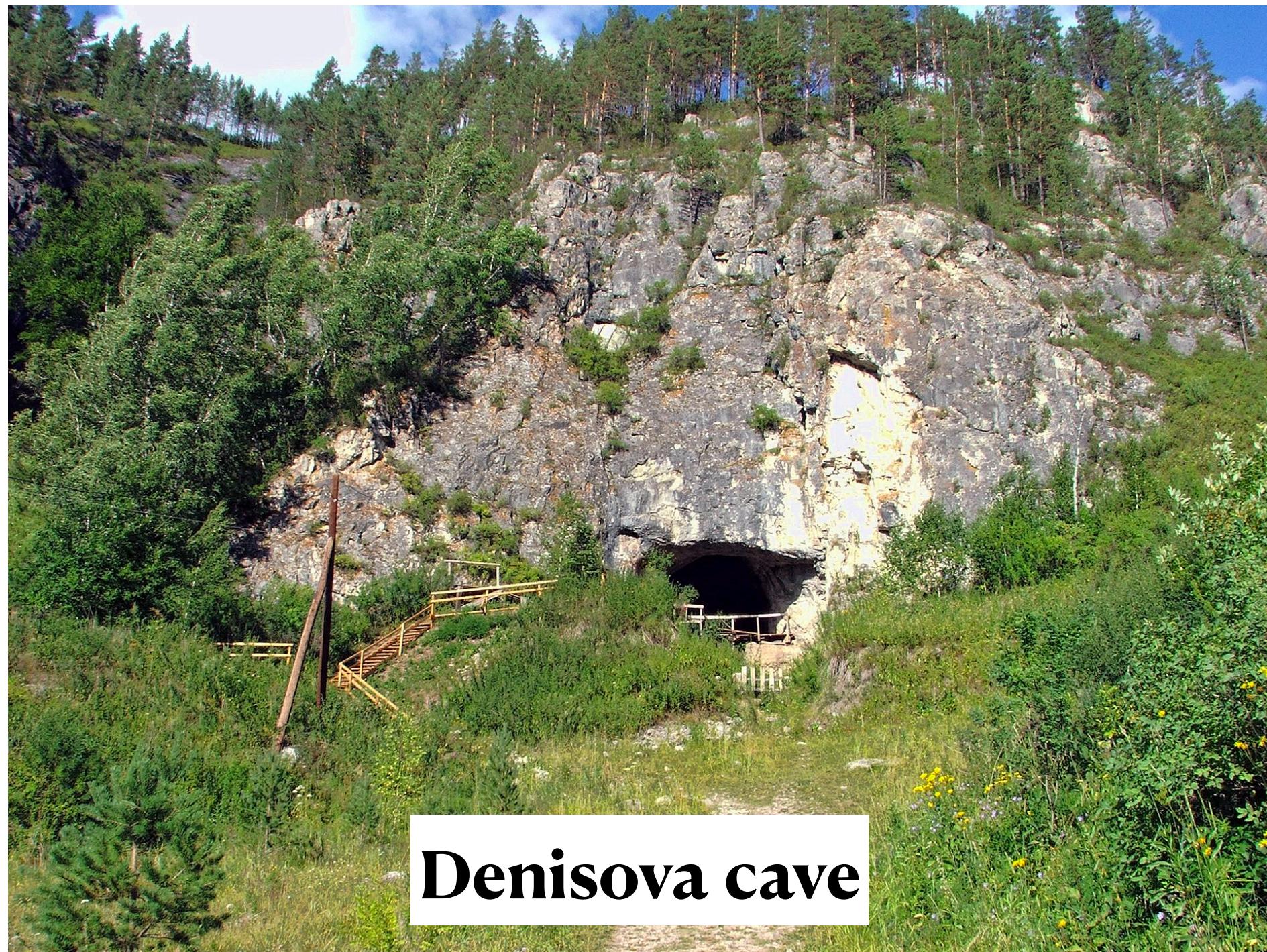
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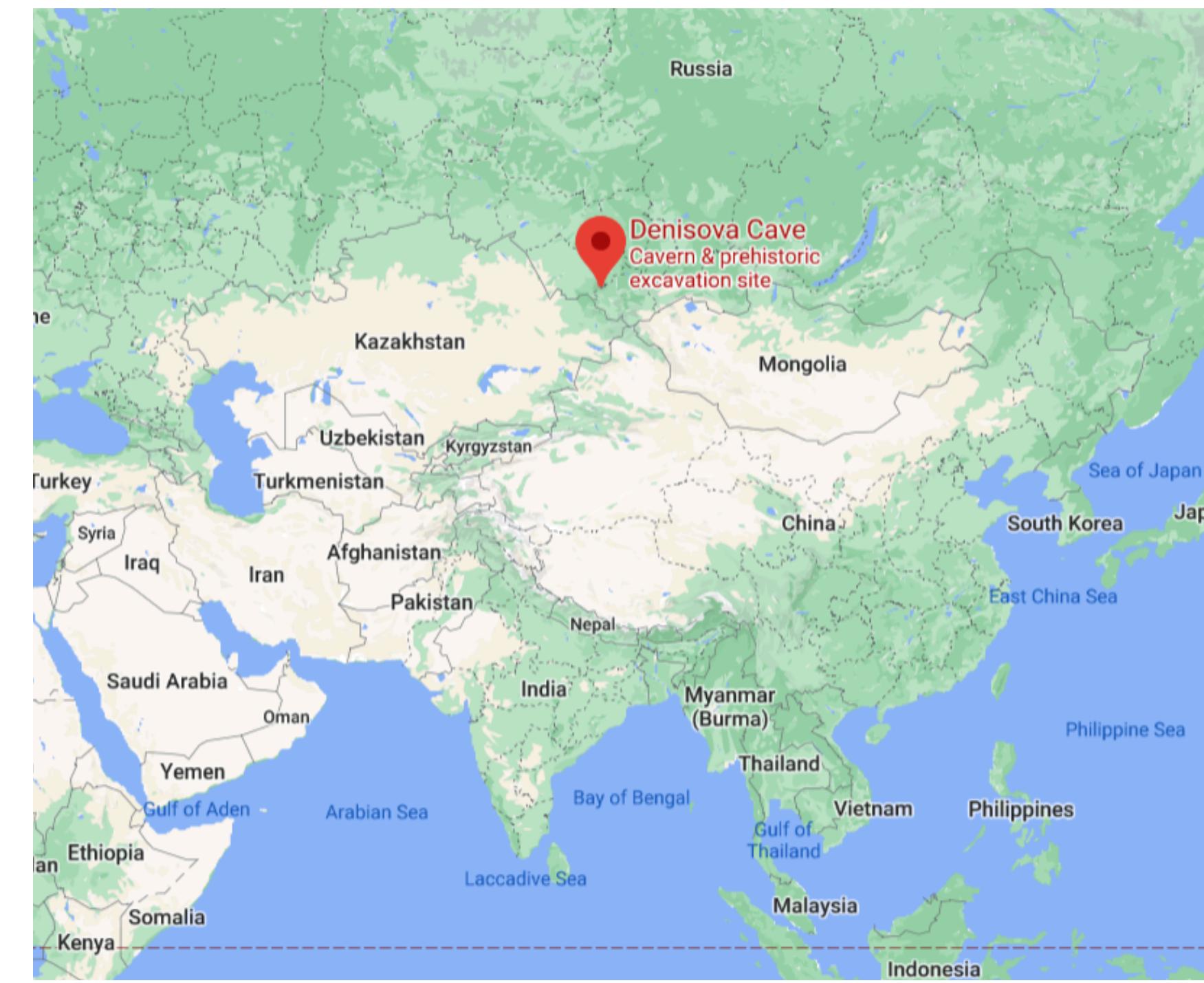
Neanderthals were not the only archaics around at the time

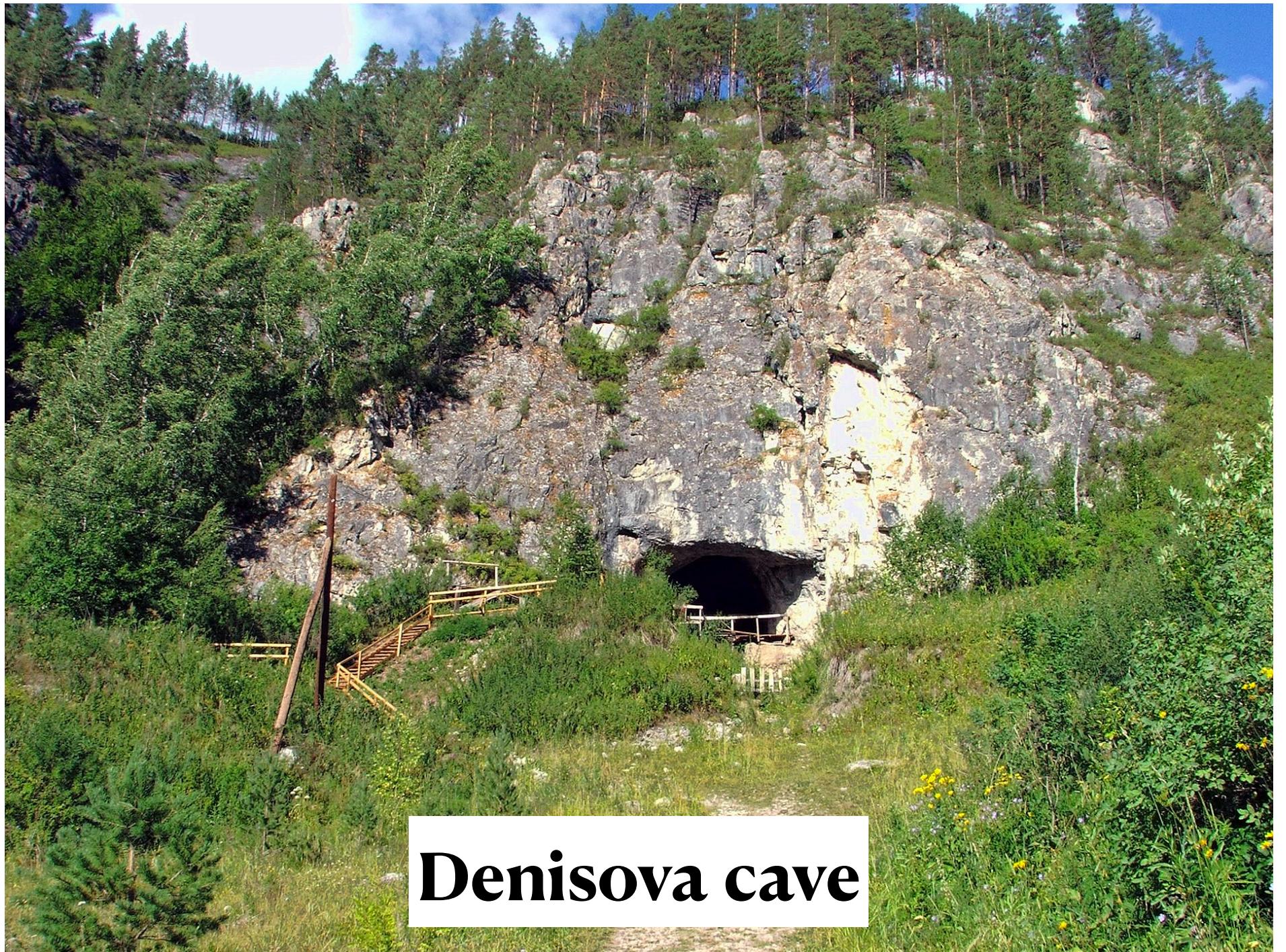


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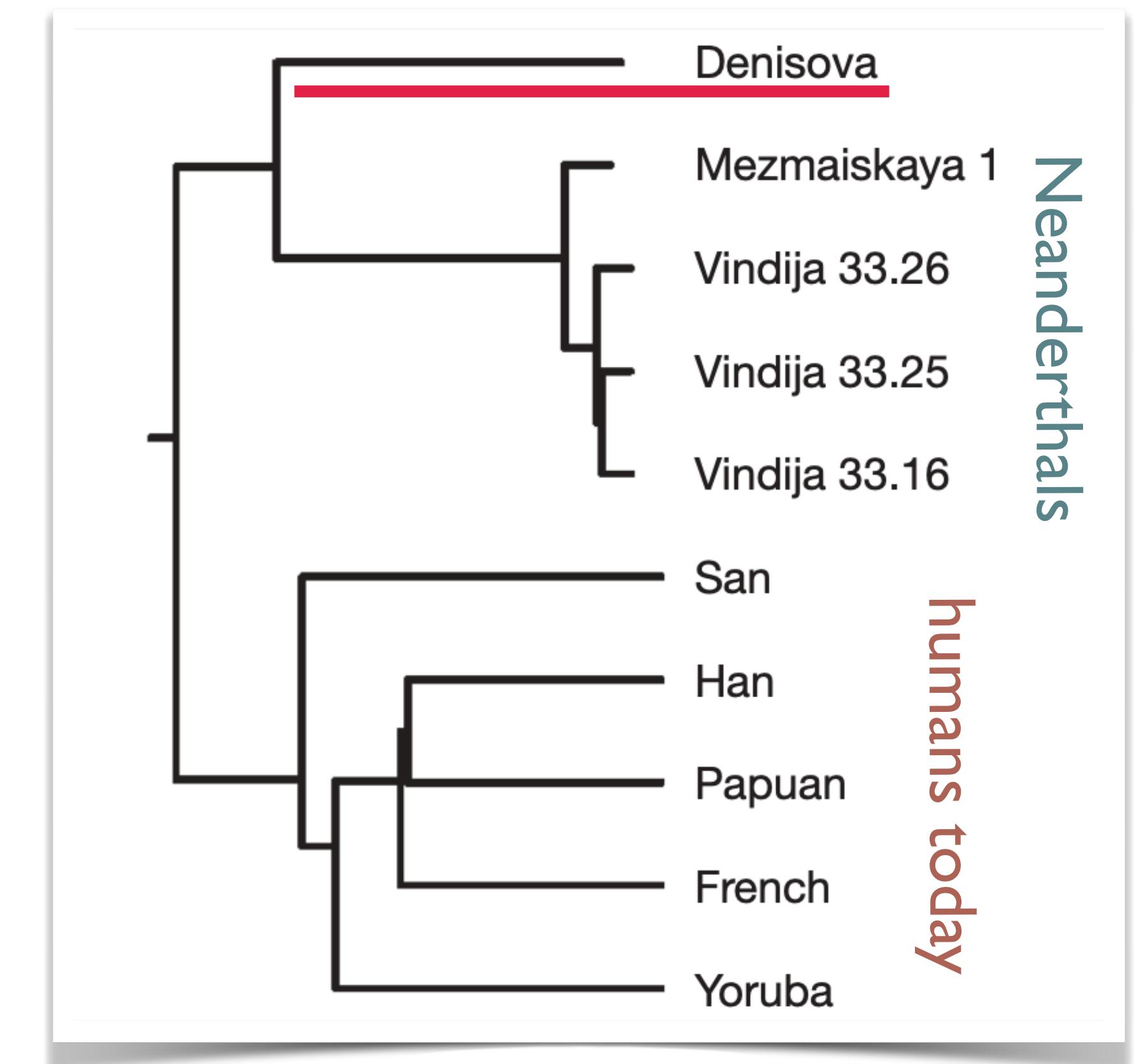


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nature

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

David Reich , Richard E. Green, Martin Kircher, Johannes Krause, Nick Patterson, Eric Y. Durand, Bence Viola , Adrian W. Briggs, Udo Stenzel, Philip L. F. Johnson, Tomislav Maricic, Jeffrey M. Good, Tomas Marques-Bonet, Can Alkan, Qiaomei Fu, Swapan Mallick, Heng Li, Matthias Meyer, Evan E. Eichler, Mark Stoneking, Michael Richards, Sahra Talamo, Michael V. Shunkov, Anatoli P. Derevianko, Jean-Jacques Hublin, Janet Kelso, Montgomery Slatkin & Svante Pääbo  — Show fewer authors

Nature 468, 1053–1060 (2010) | [Cite this article](#)

Traces of Denisovans in genomes of present-day humans

$f4(\text{human 1}, \text{human 2}; \text{Denisovan}, \text{Chimp})$

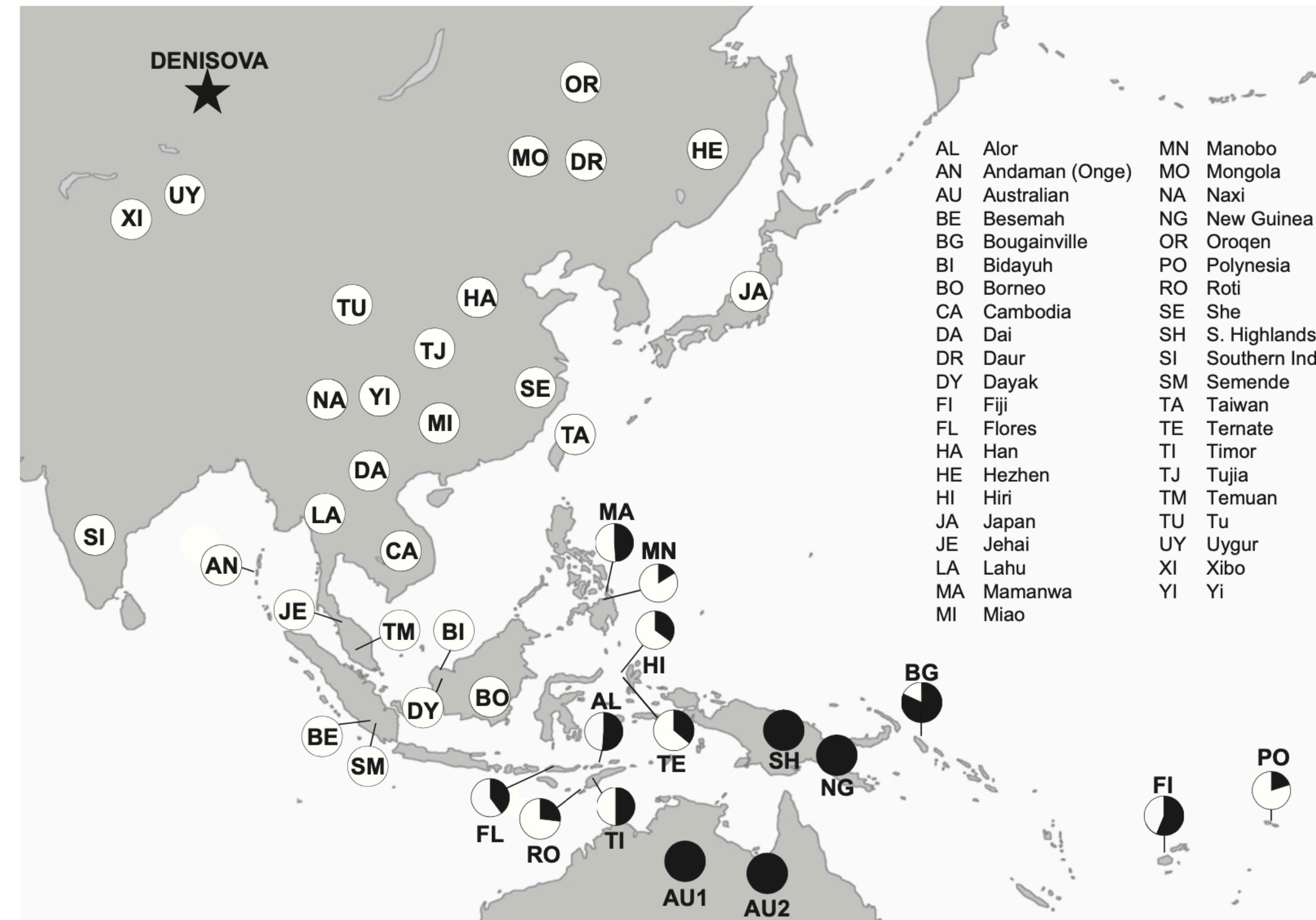


Figure 1. Denisovan Genetic Material as a Fraction of that in New Guineans

Reich et al., 2011

Traces of Denisovans in genomes of present-day humans

$f4(\text{human 1}, \text{human 2}; \text{Denisovan}, \text{Chimp})$

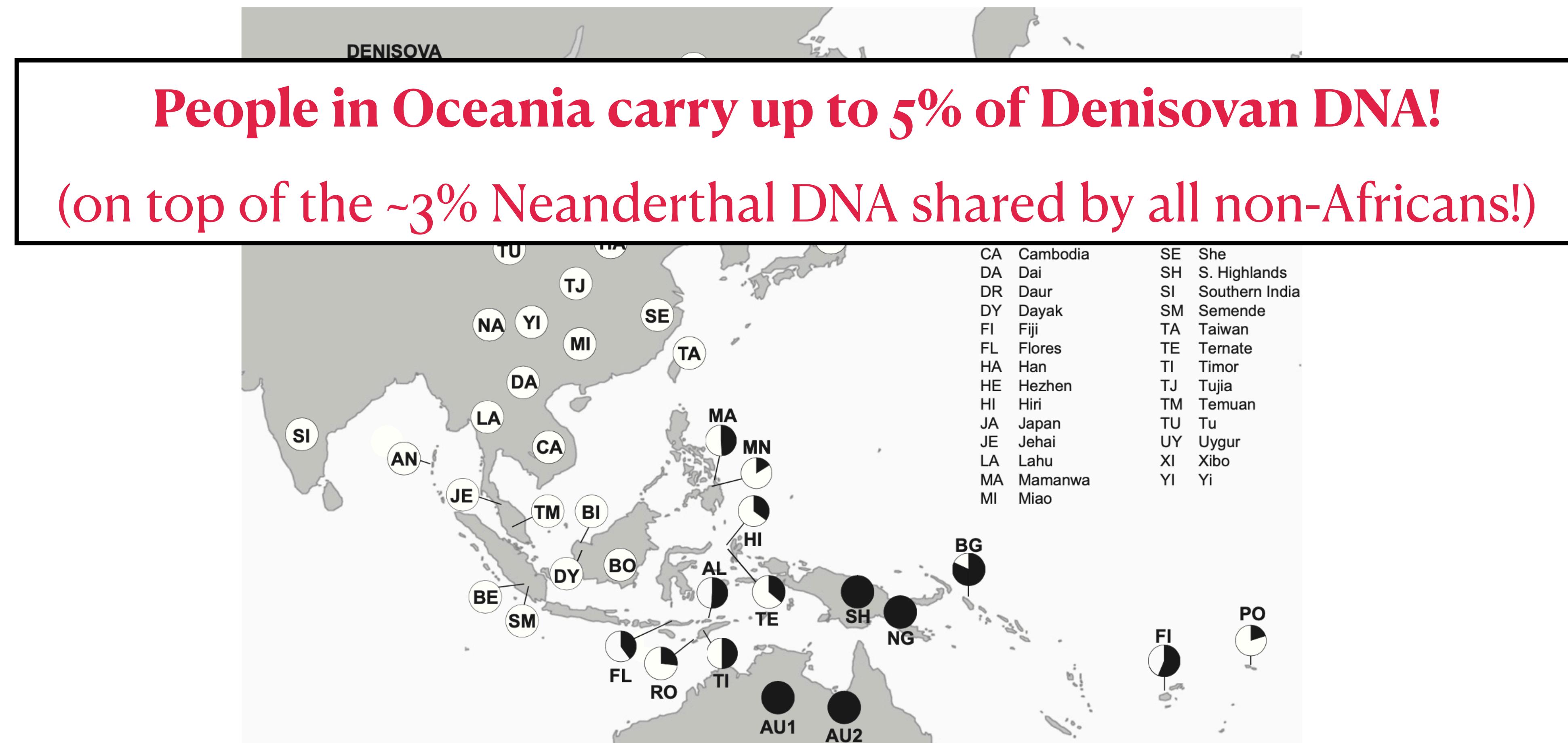
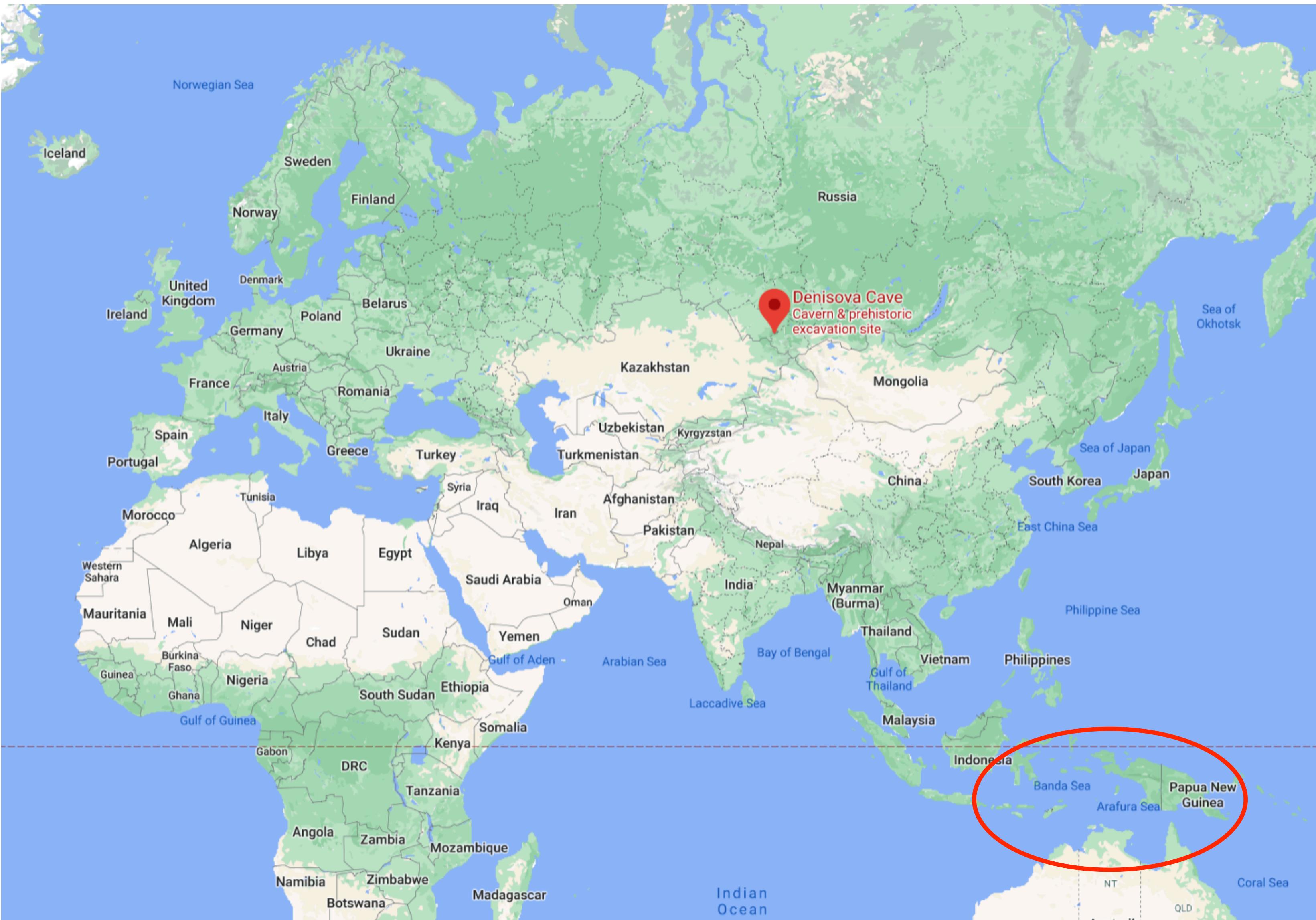


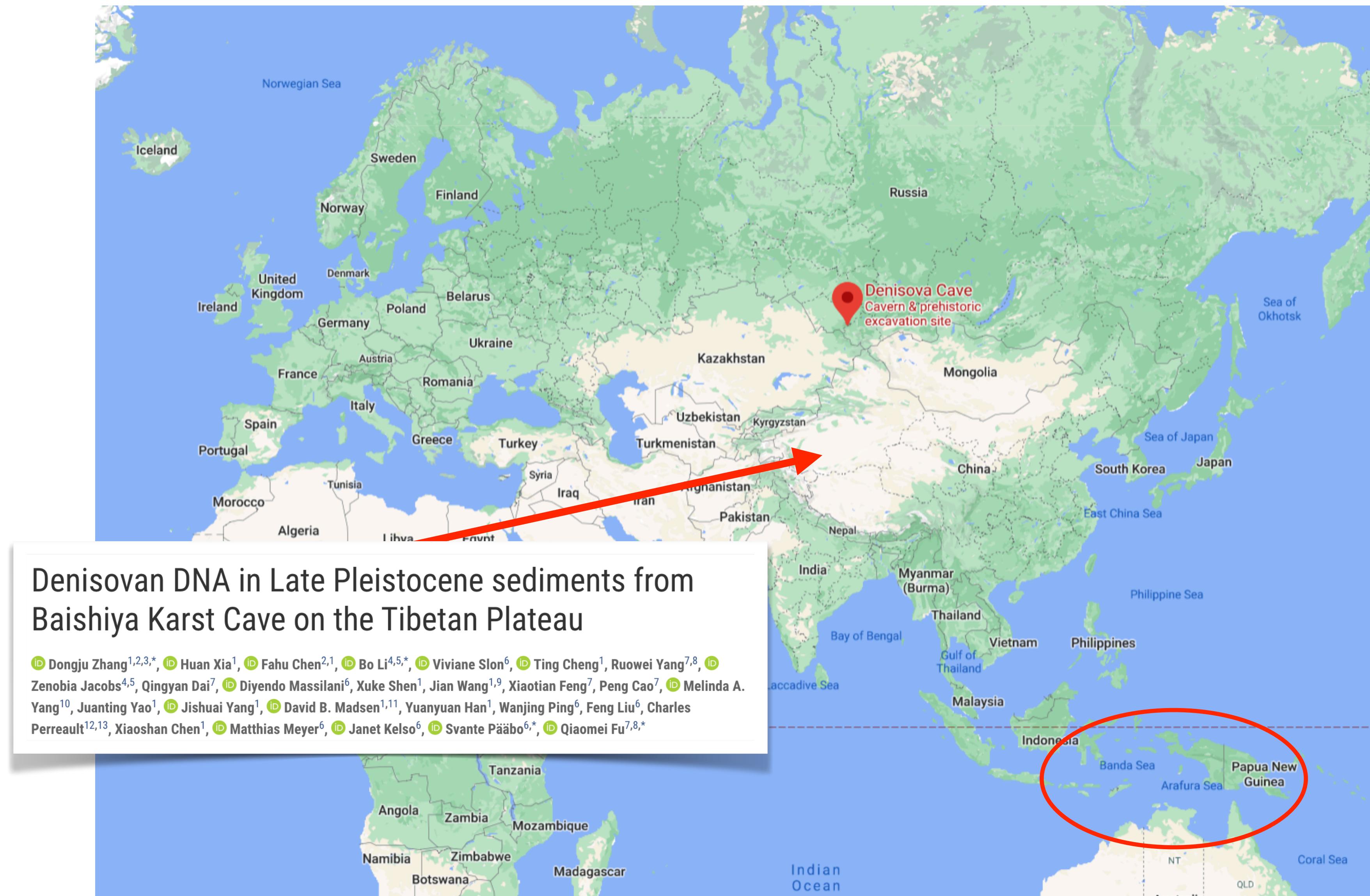
Figure 1. Denisovan Genetic Material as a Fraction of that in New Guineans

Reich et al., 2011

Geographic range of Denisovans?

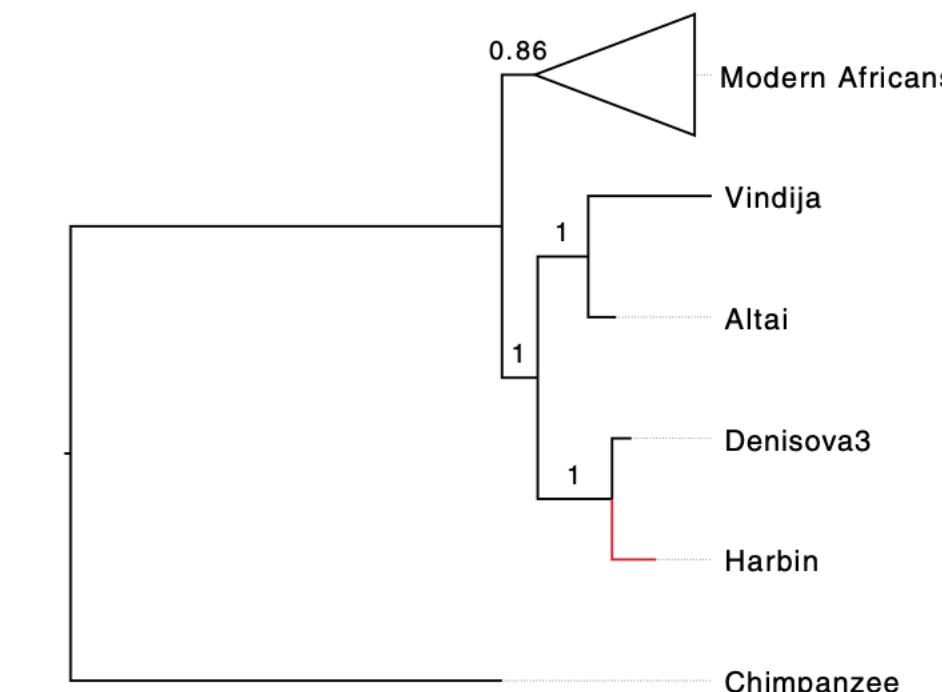
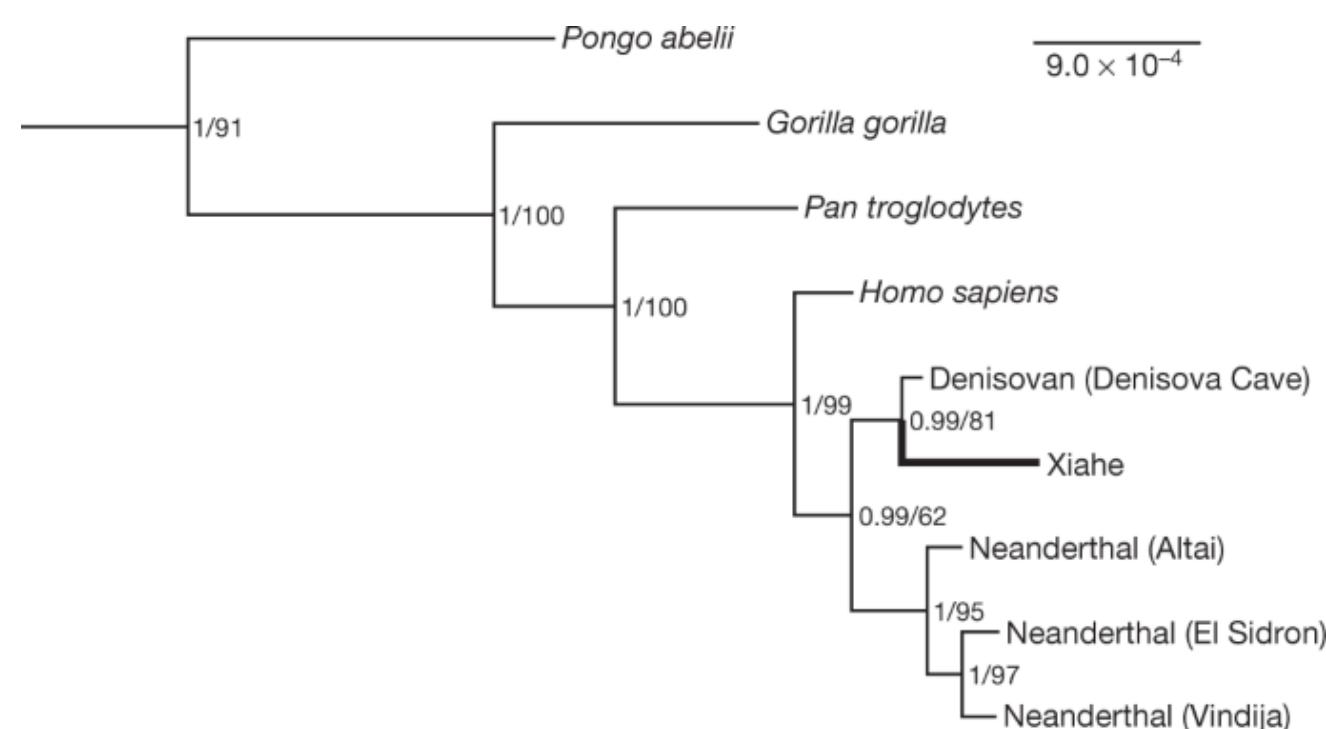
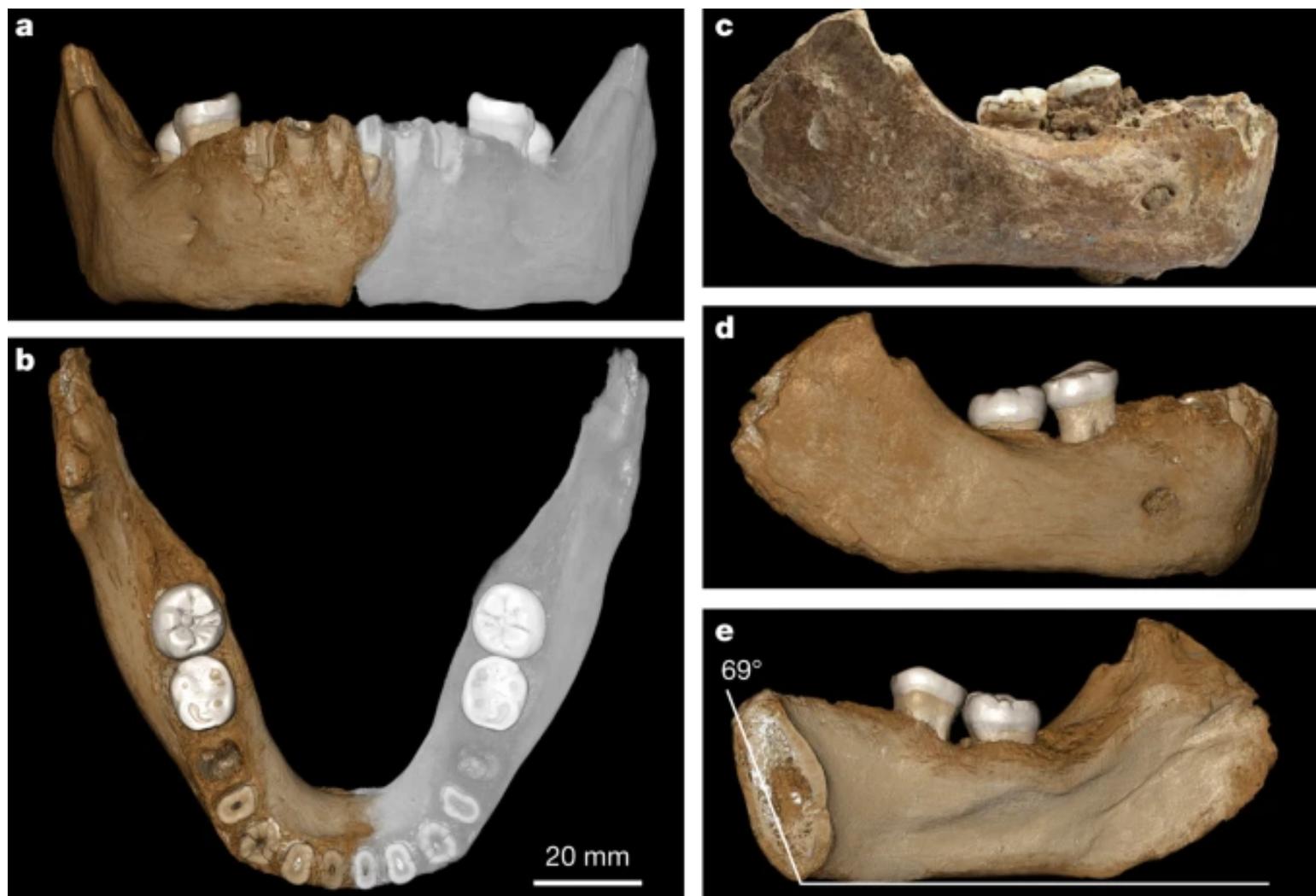


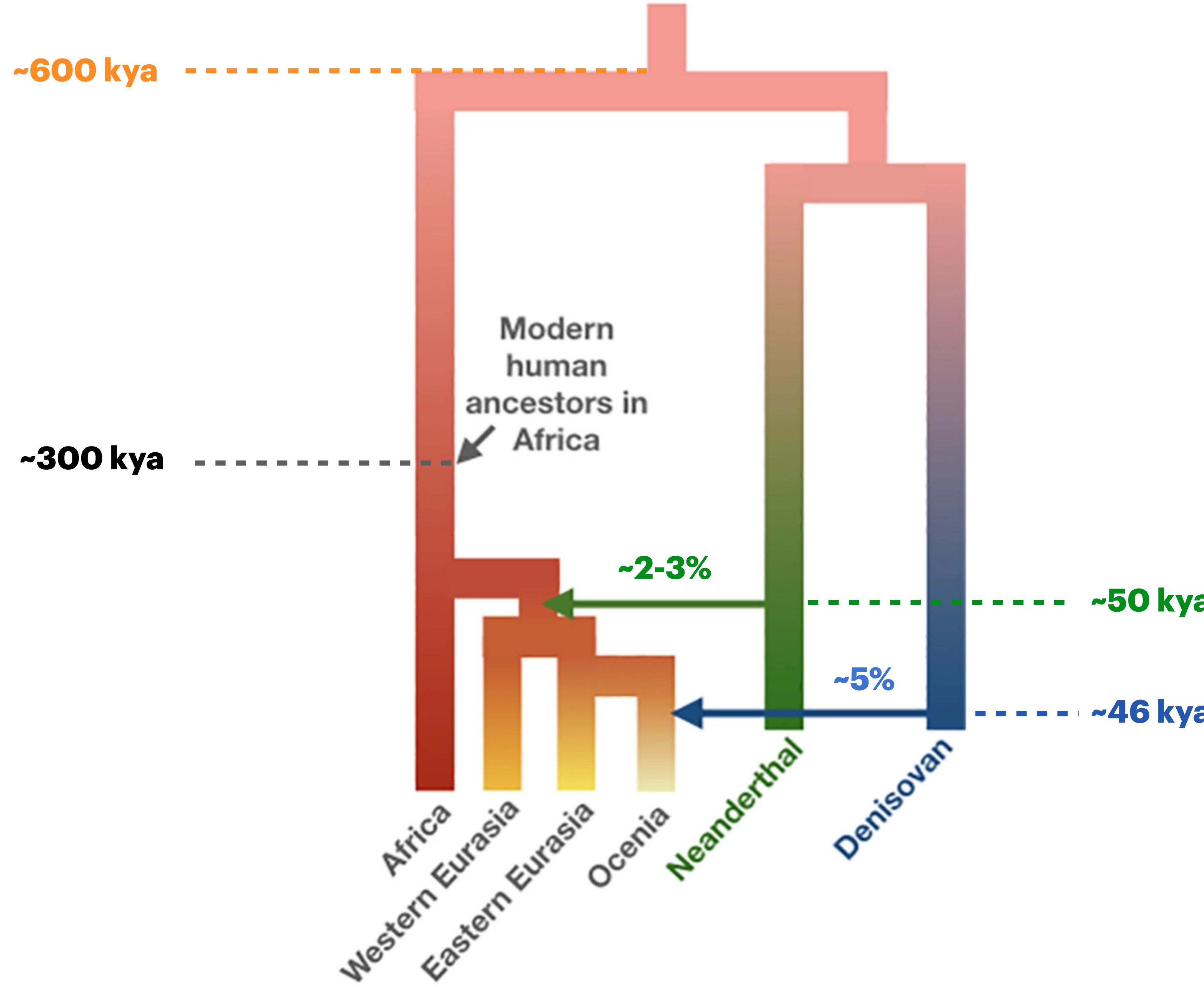
Geographic range of Denisovans?



Glimpses of a Denisovan anatomy

(thanks to ancient proteins!)





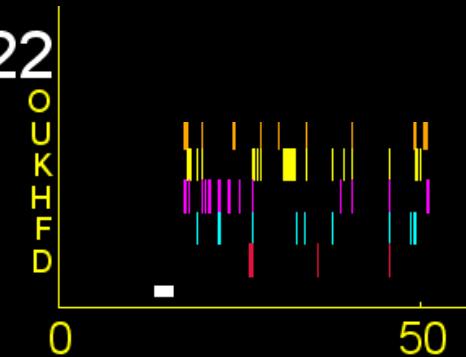
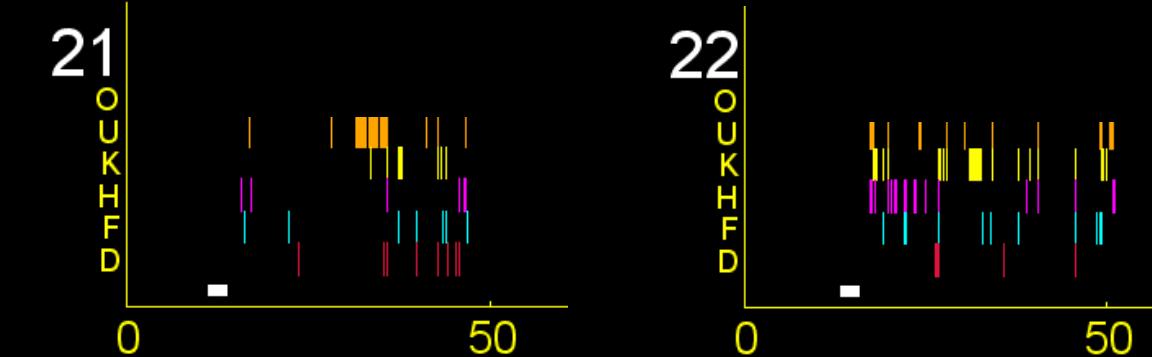
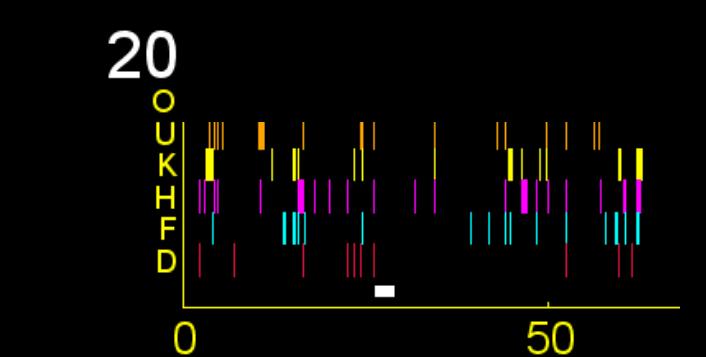
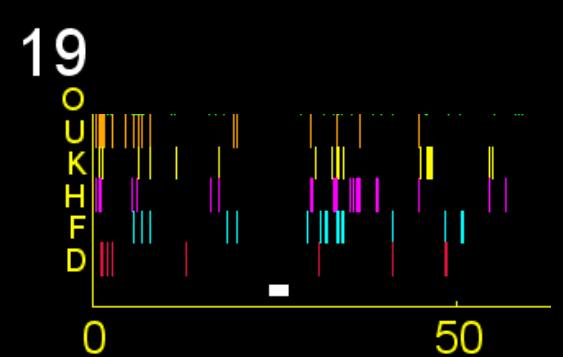
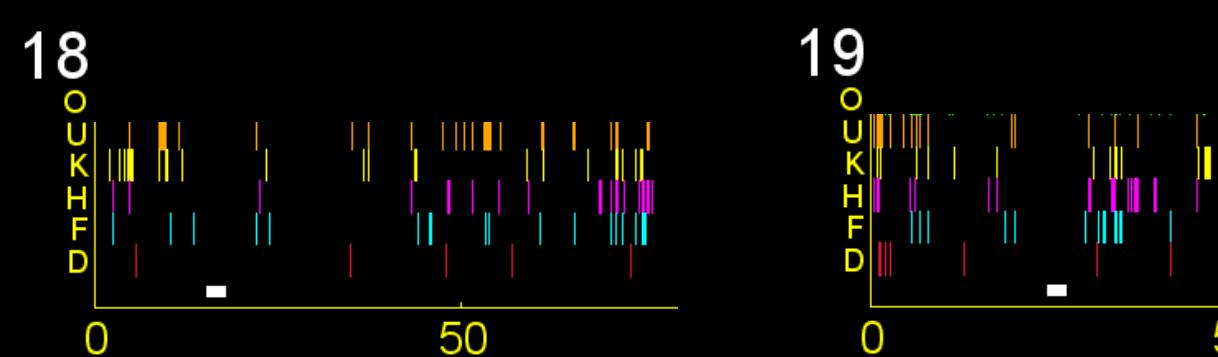
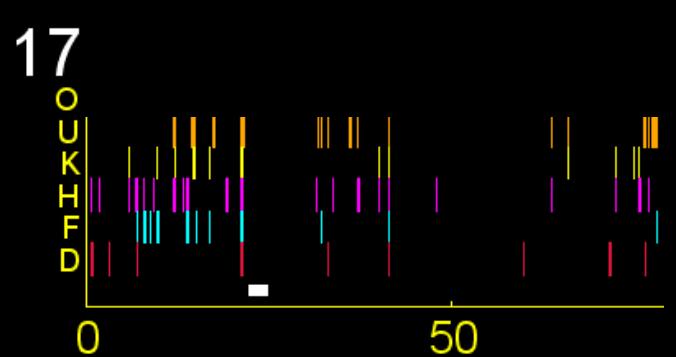
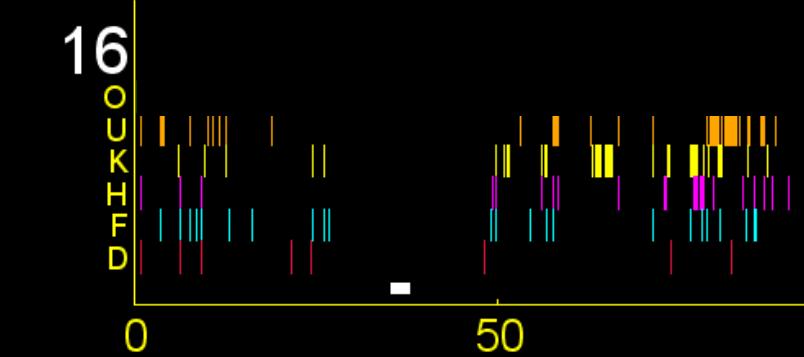
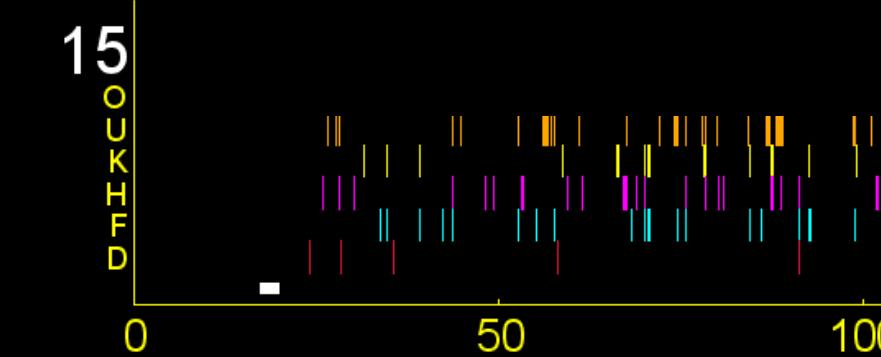
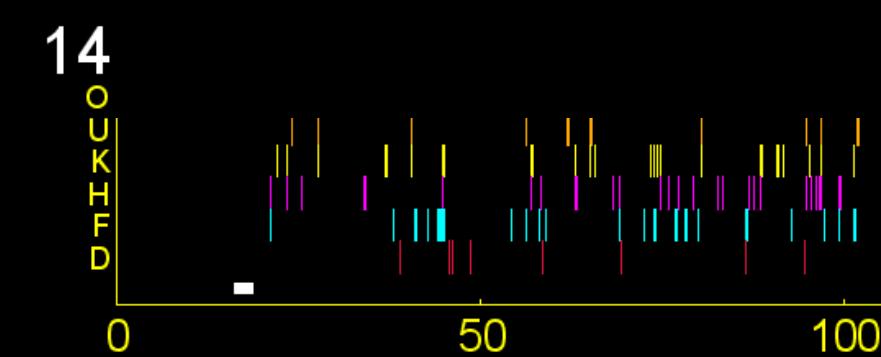
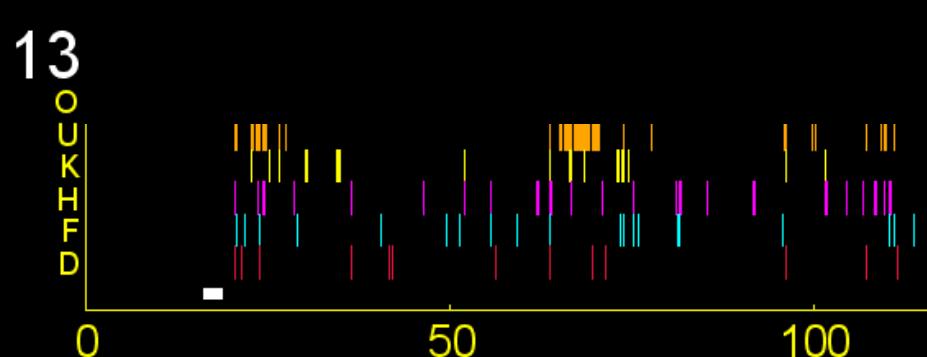
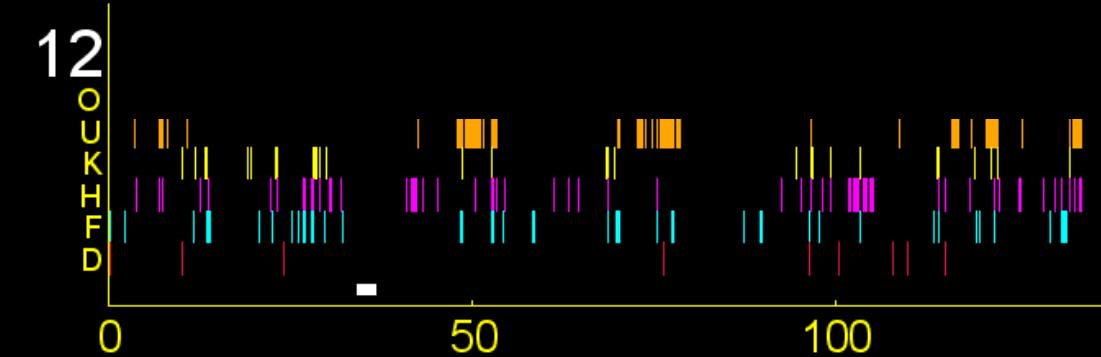
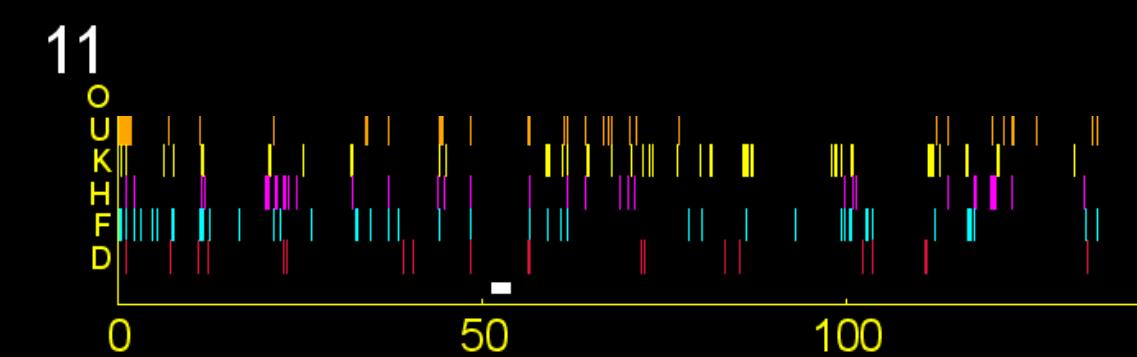
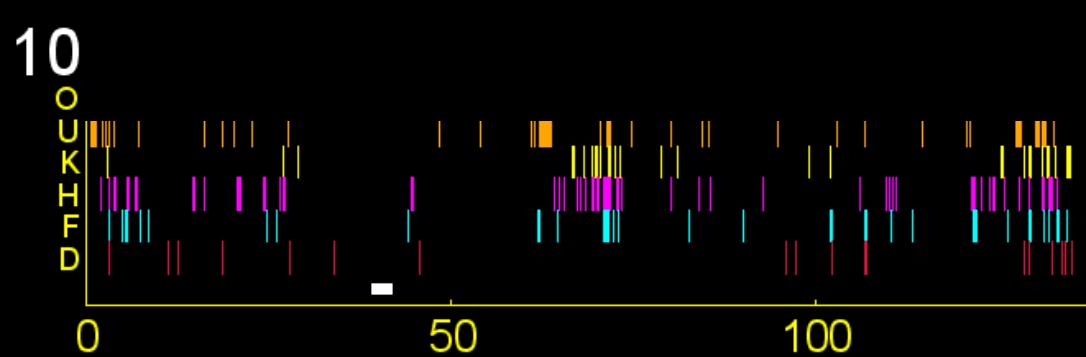
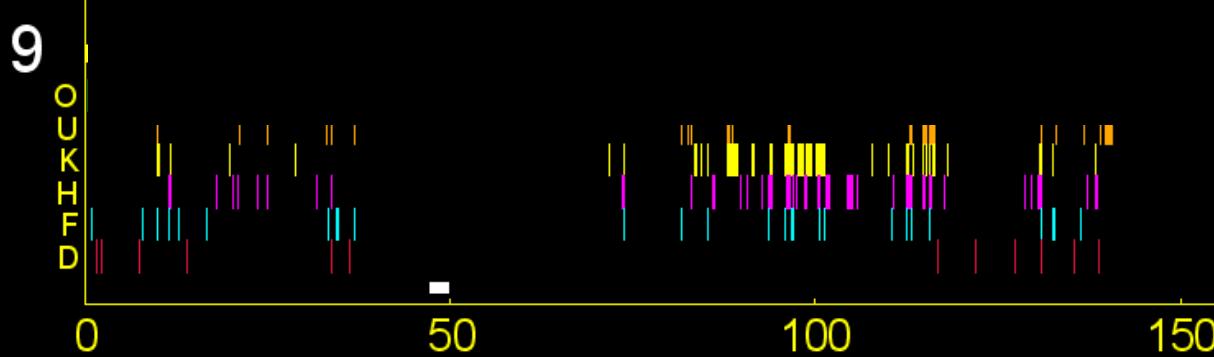
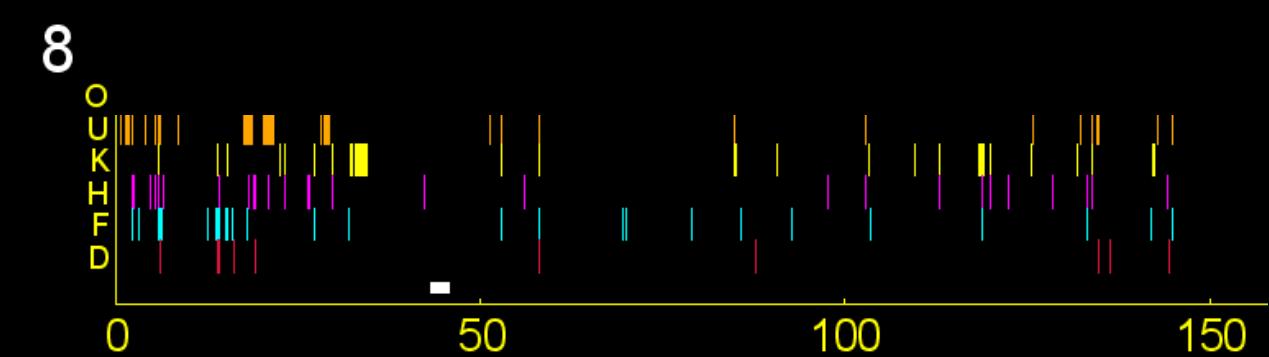
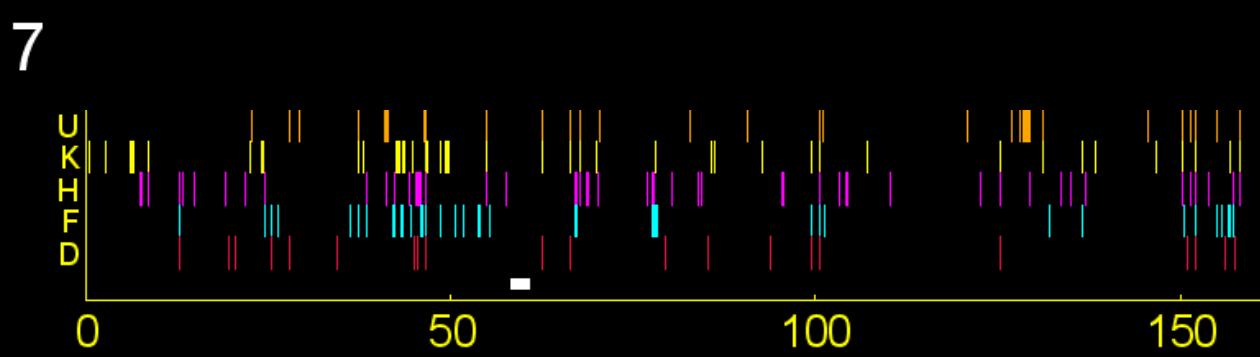
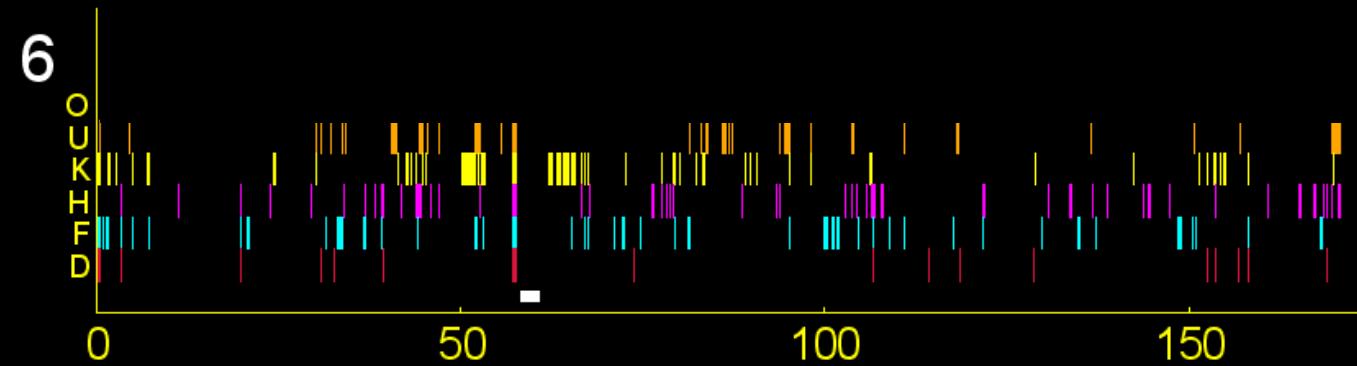
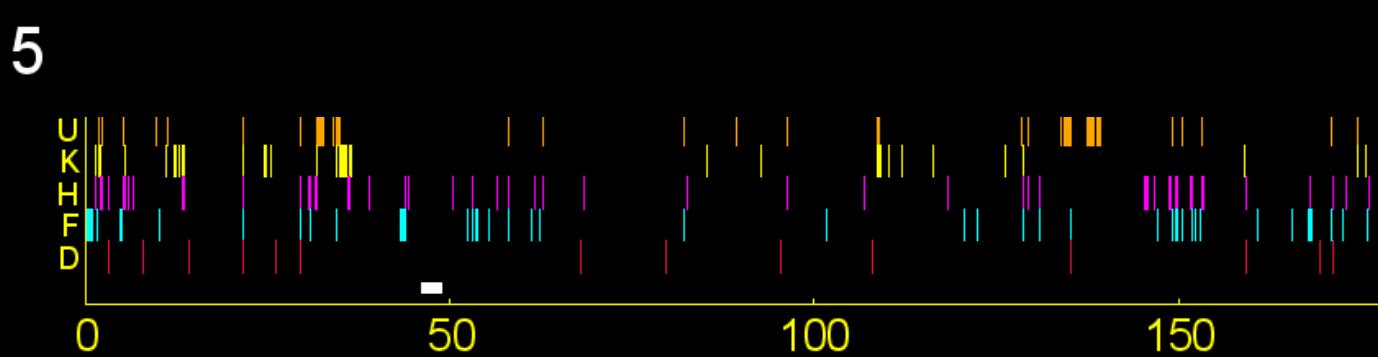
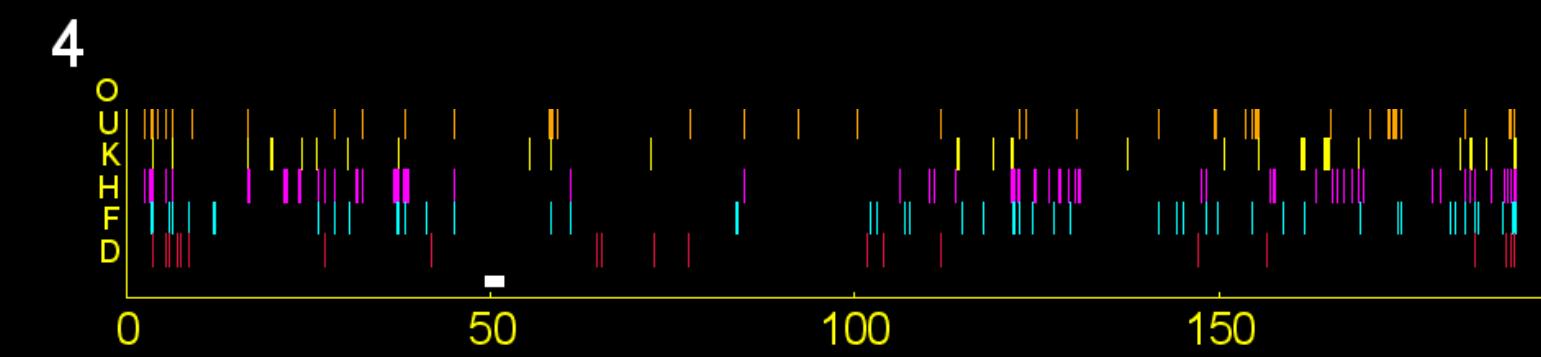
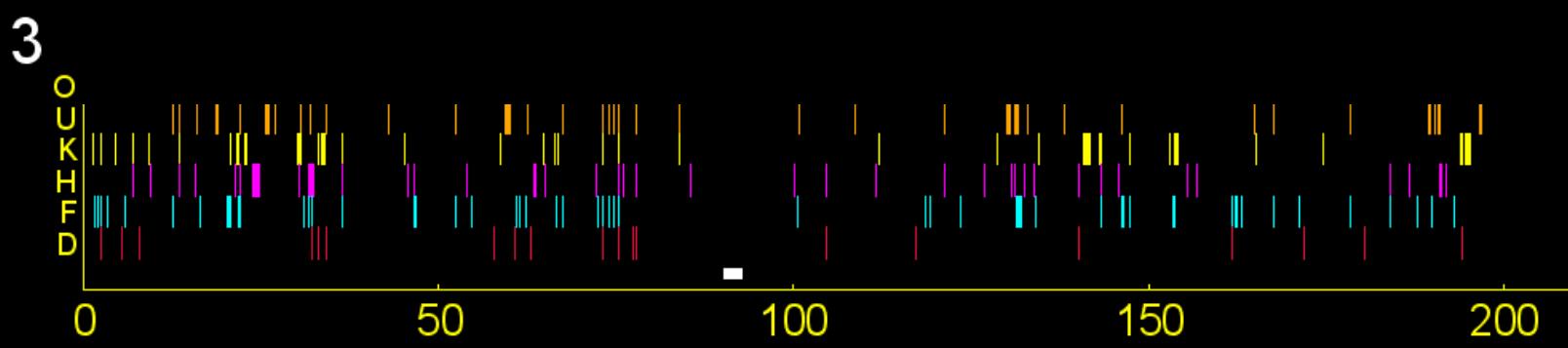
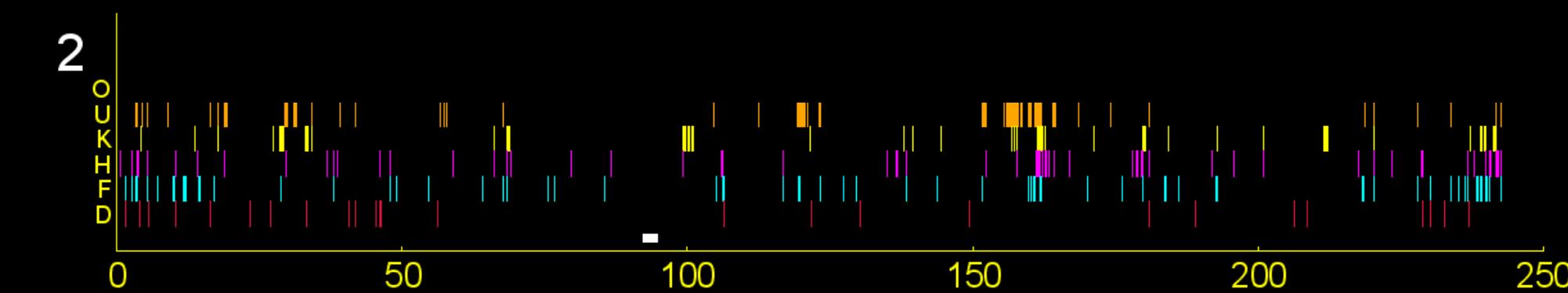
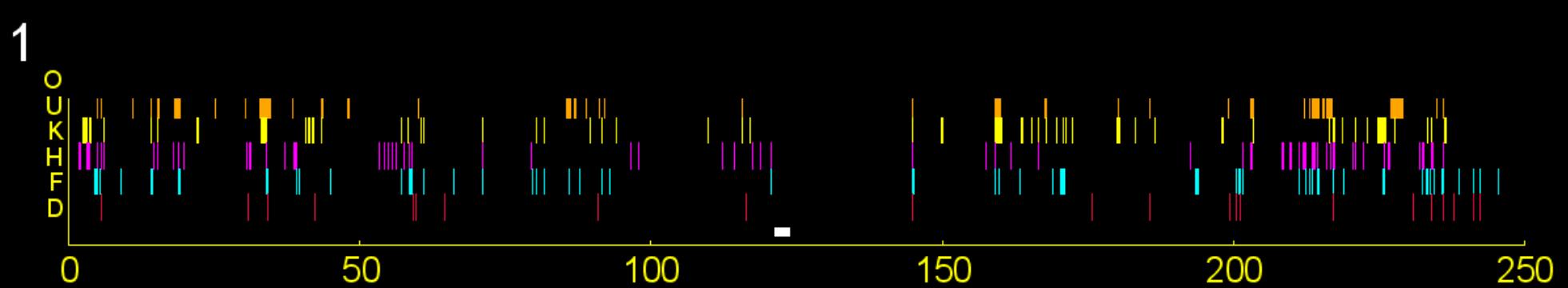
adapted from the Yearbook of Physical Anthropology (2019) by Gokcumen

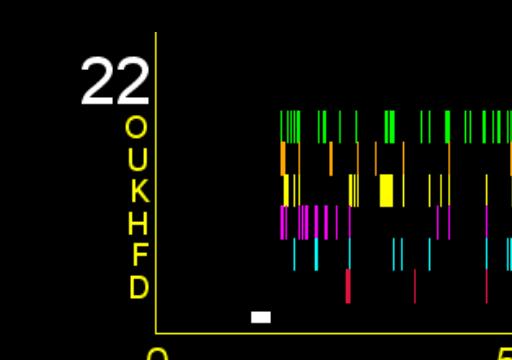
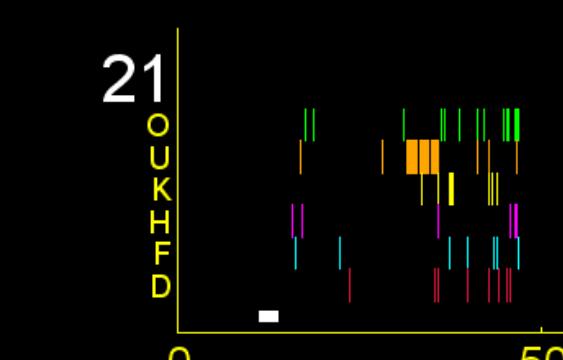
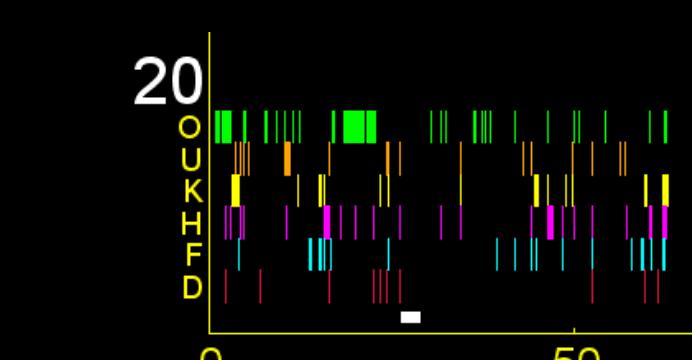
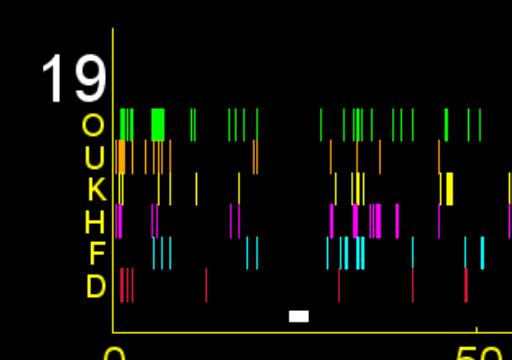
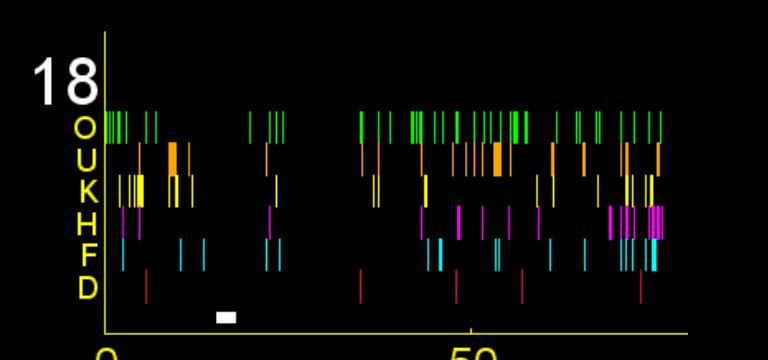
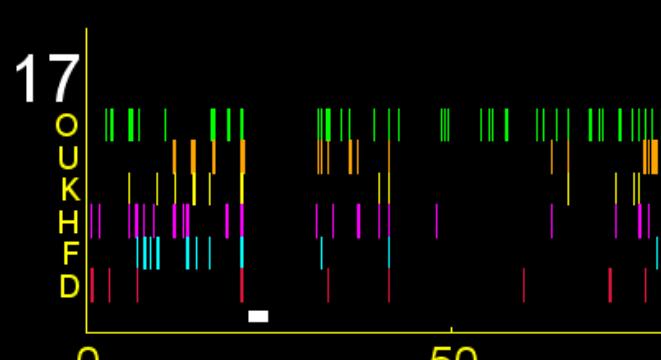
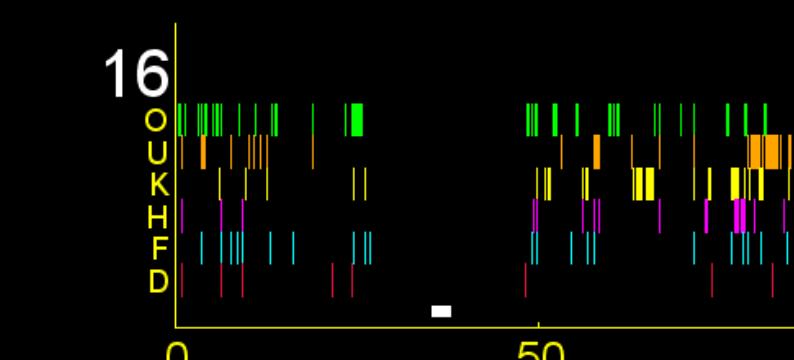
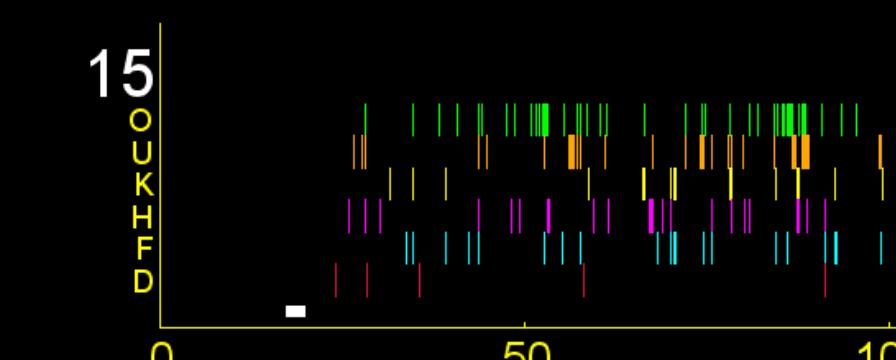
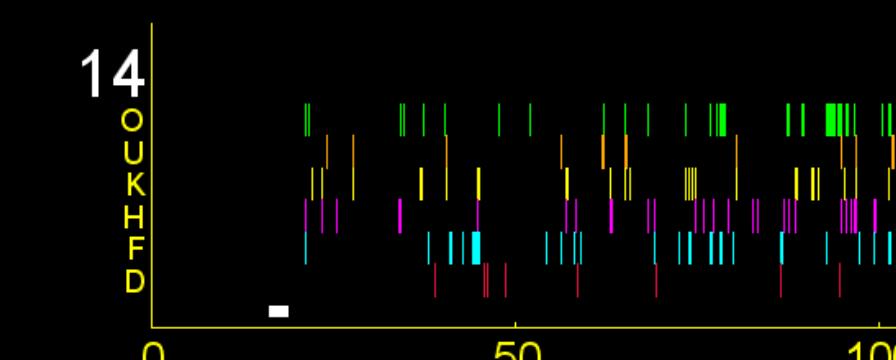
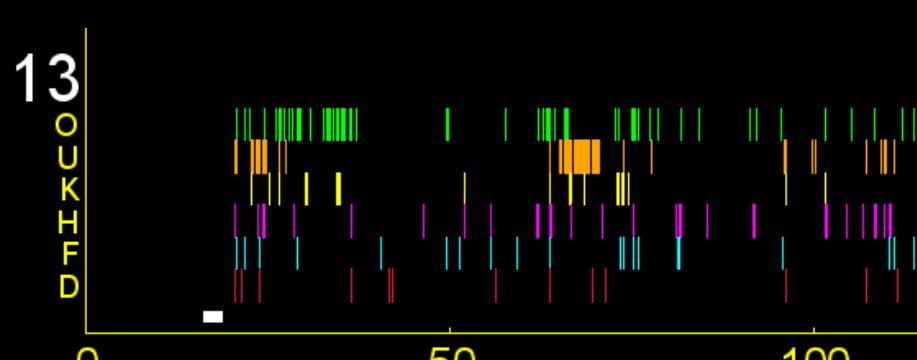
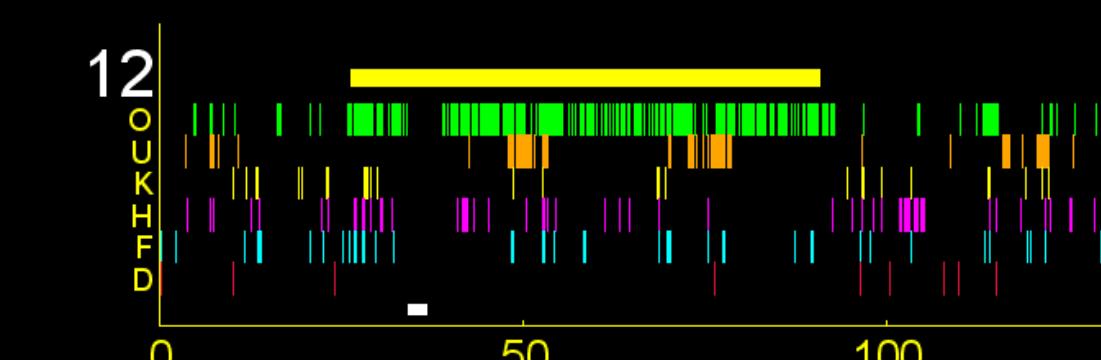
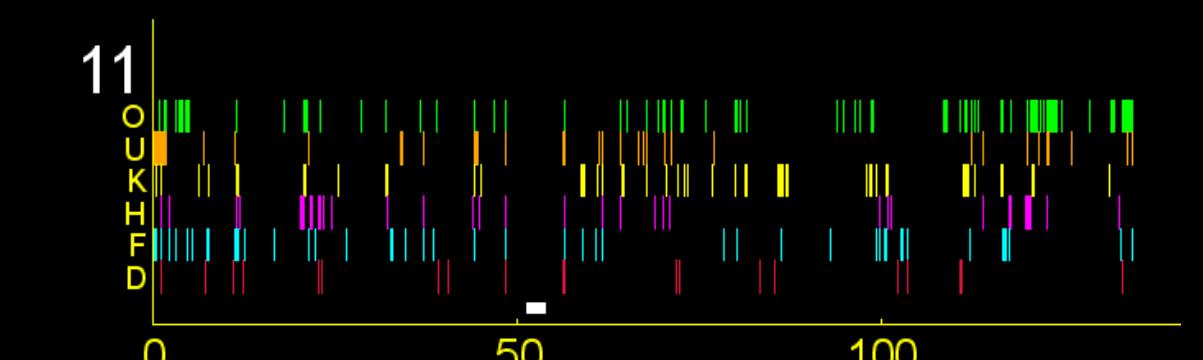
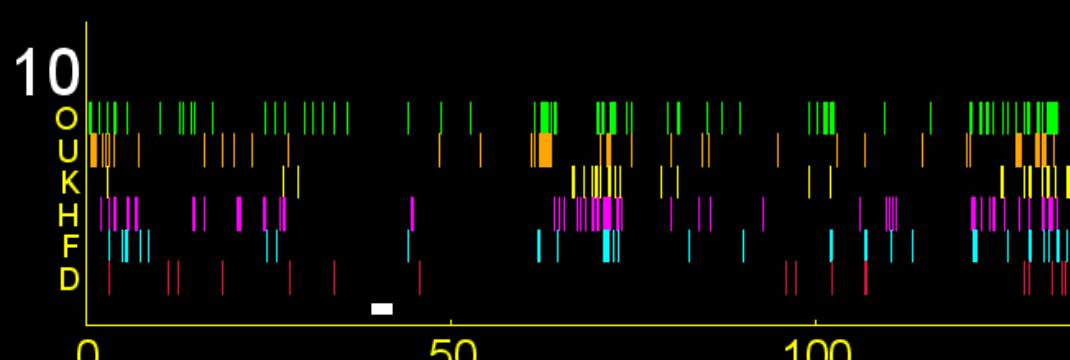
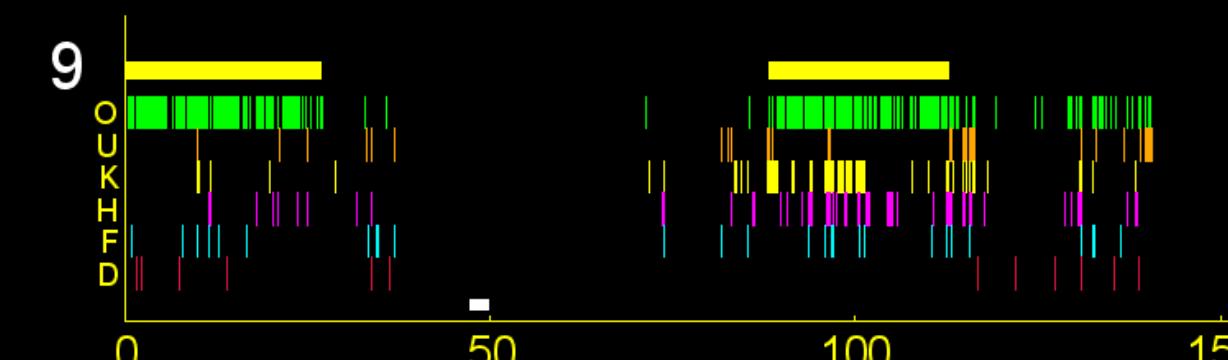
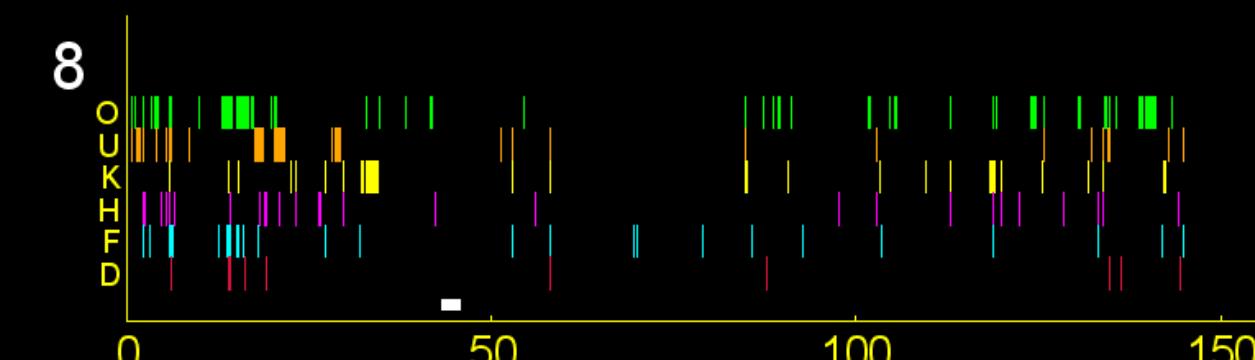
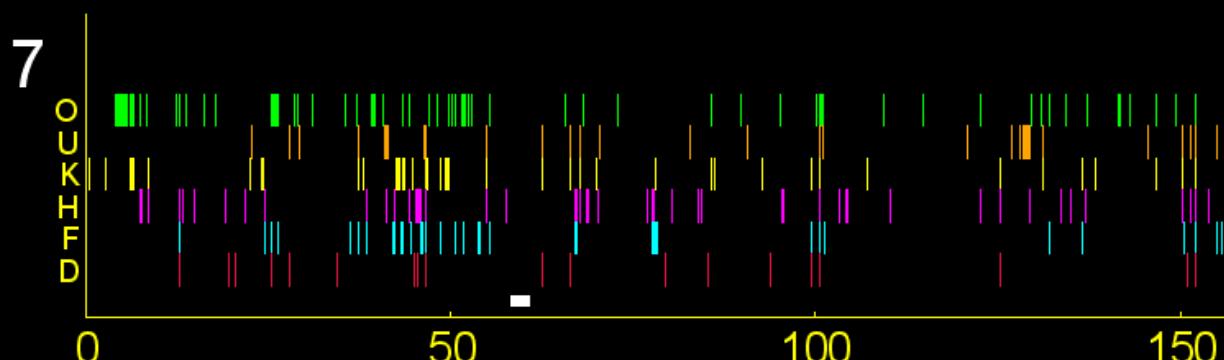
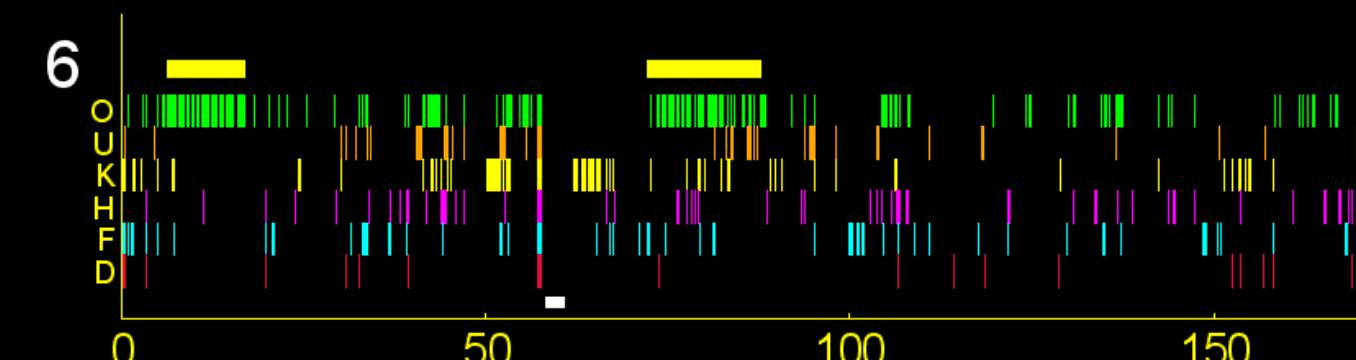
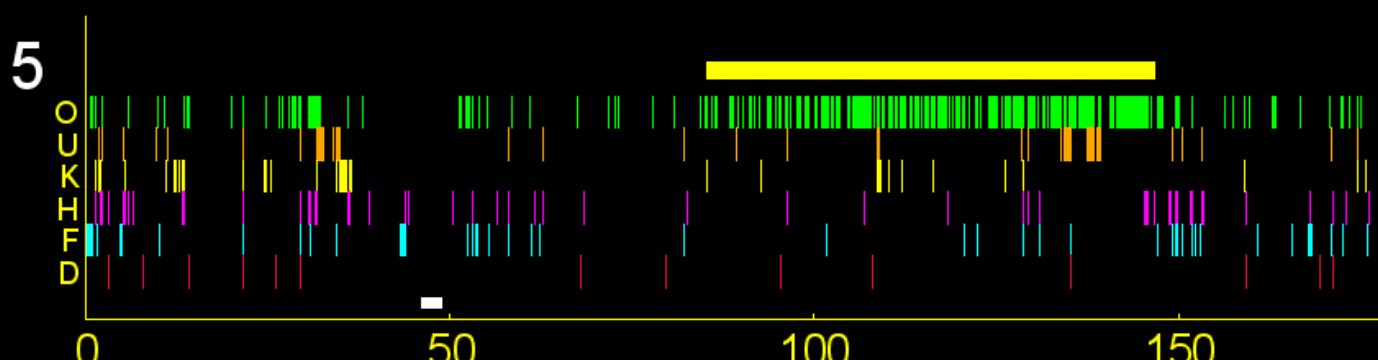
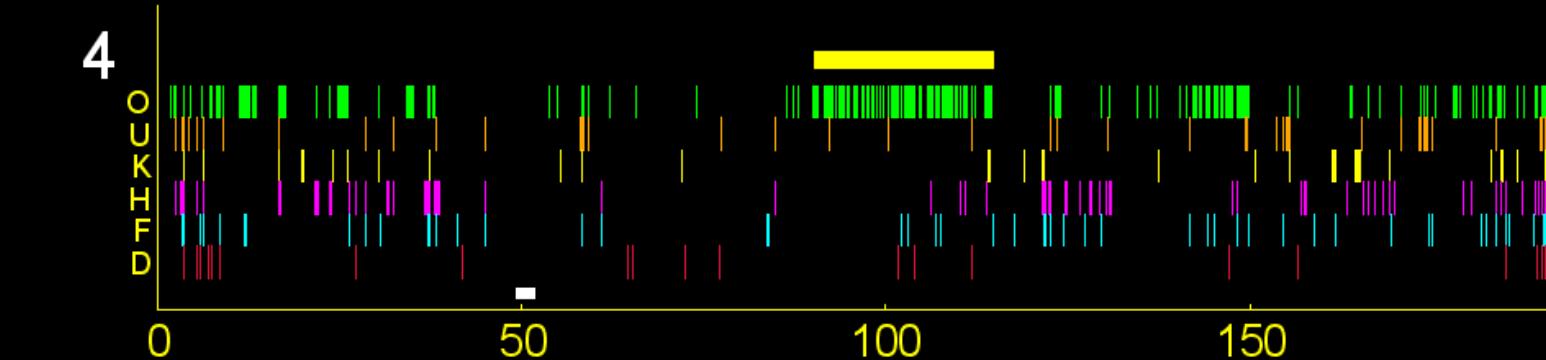
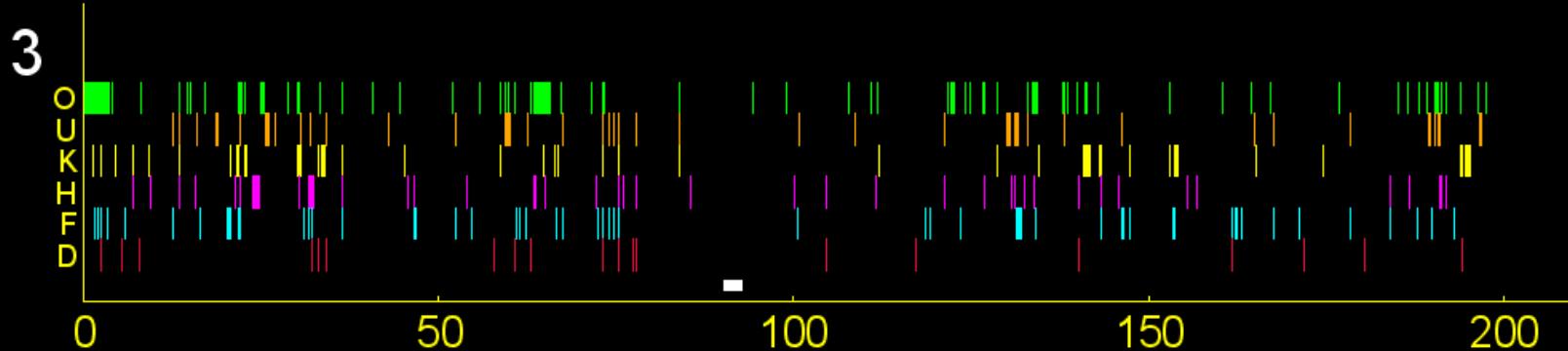
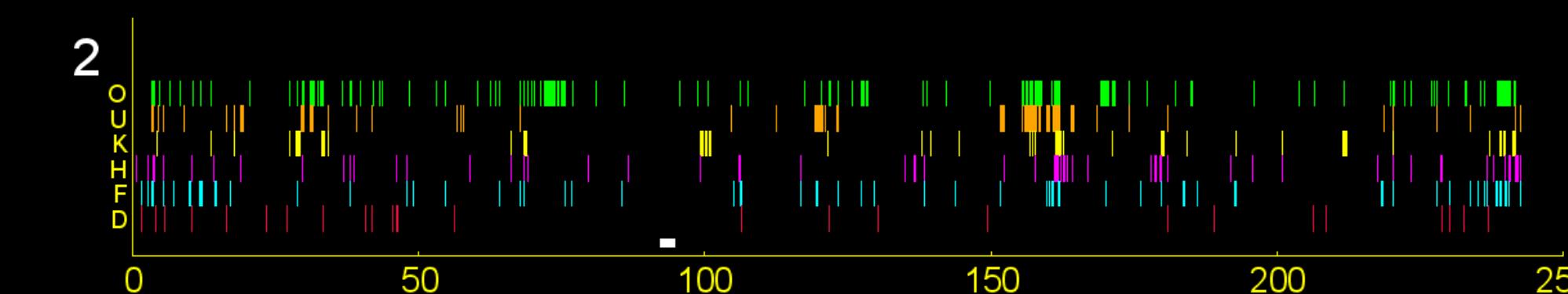
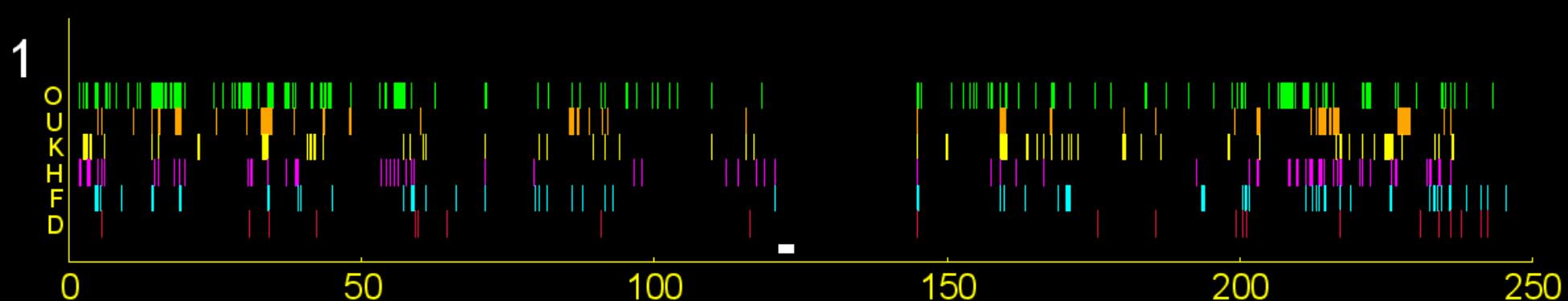
Morphological evidence?

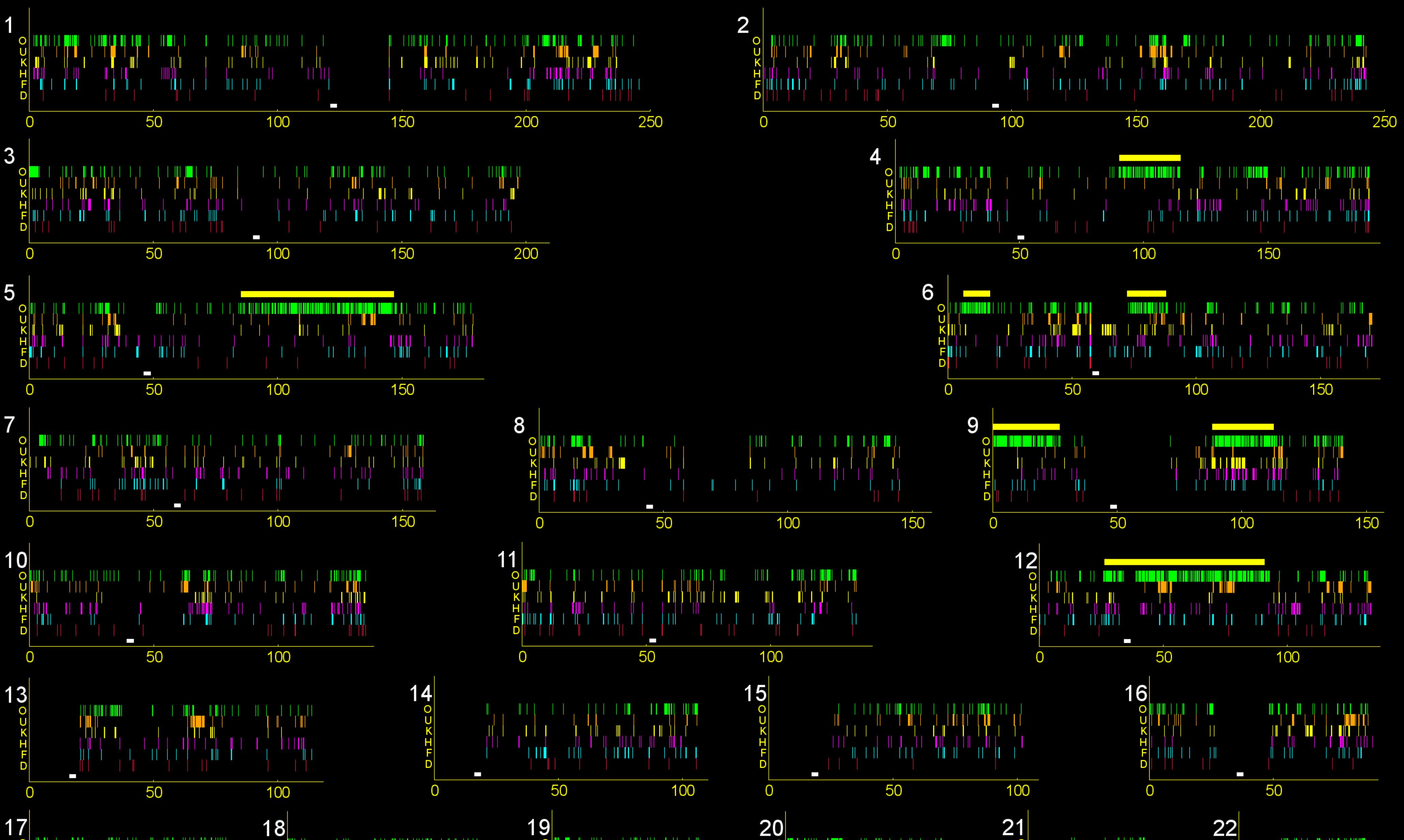


**~40 thousand years old
remains of a modern human
Peștera cu Oase, Romania**

Trinkaus *et al.* PNAS, 2003





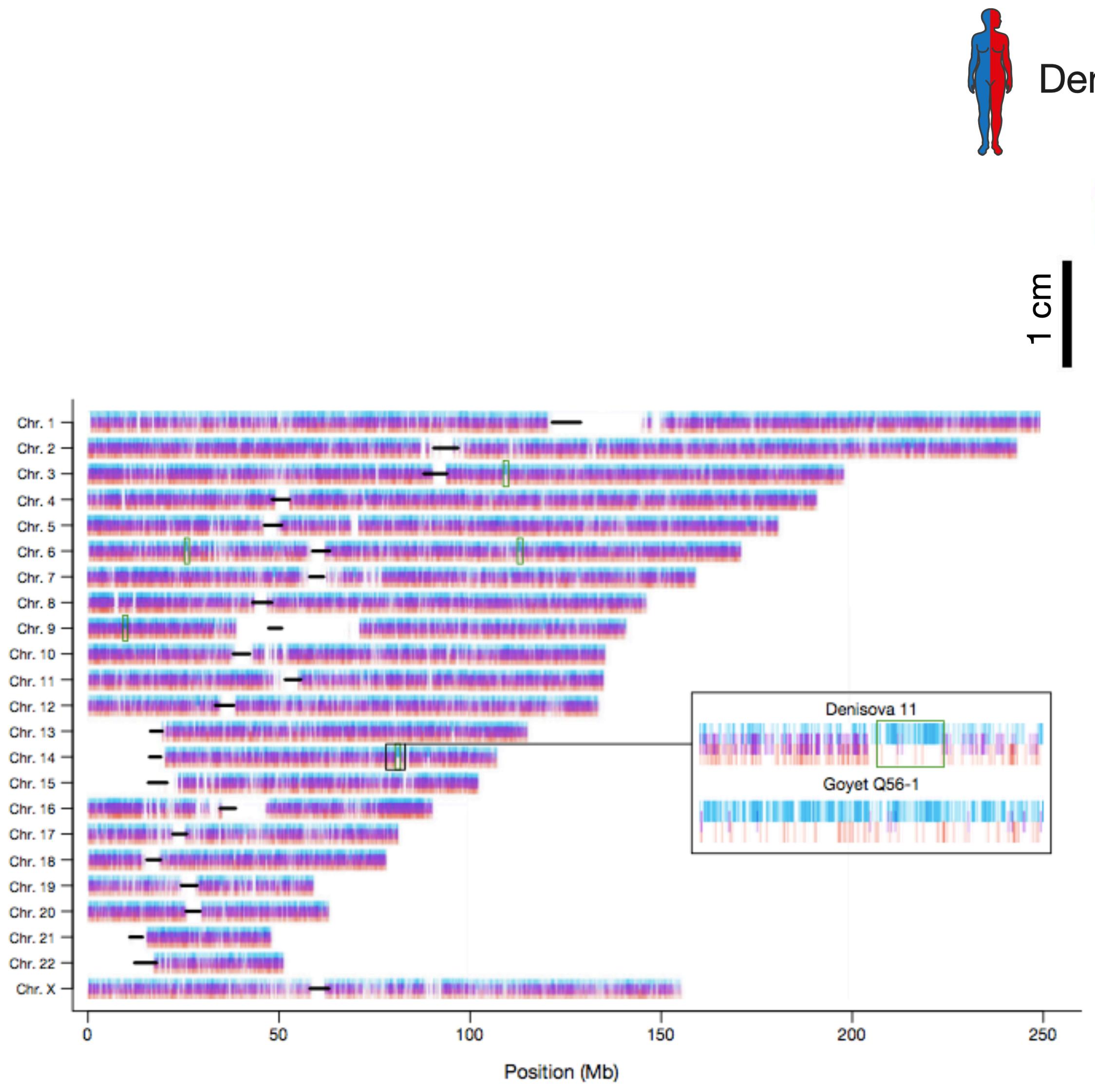


Neanderthal ancestor four to six generations back!

Denisova 11



By Демин Алексей Барнаул - Own work, CC BY-SA 4.0, <https://commons.wikimedia.org/w/index.php?curid=48890364>



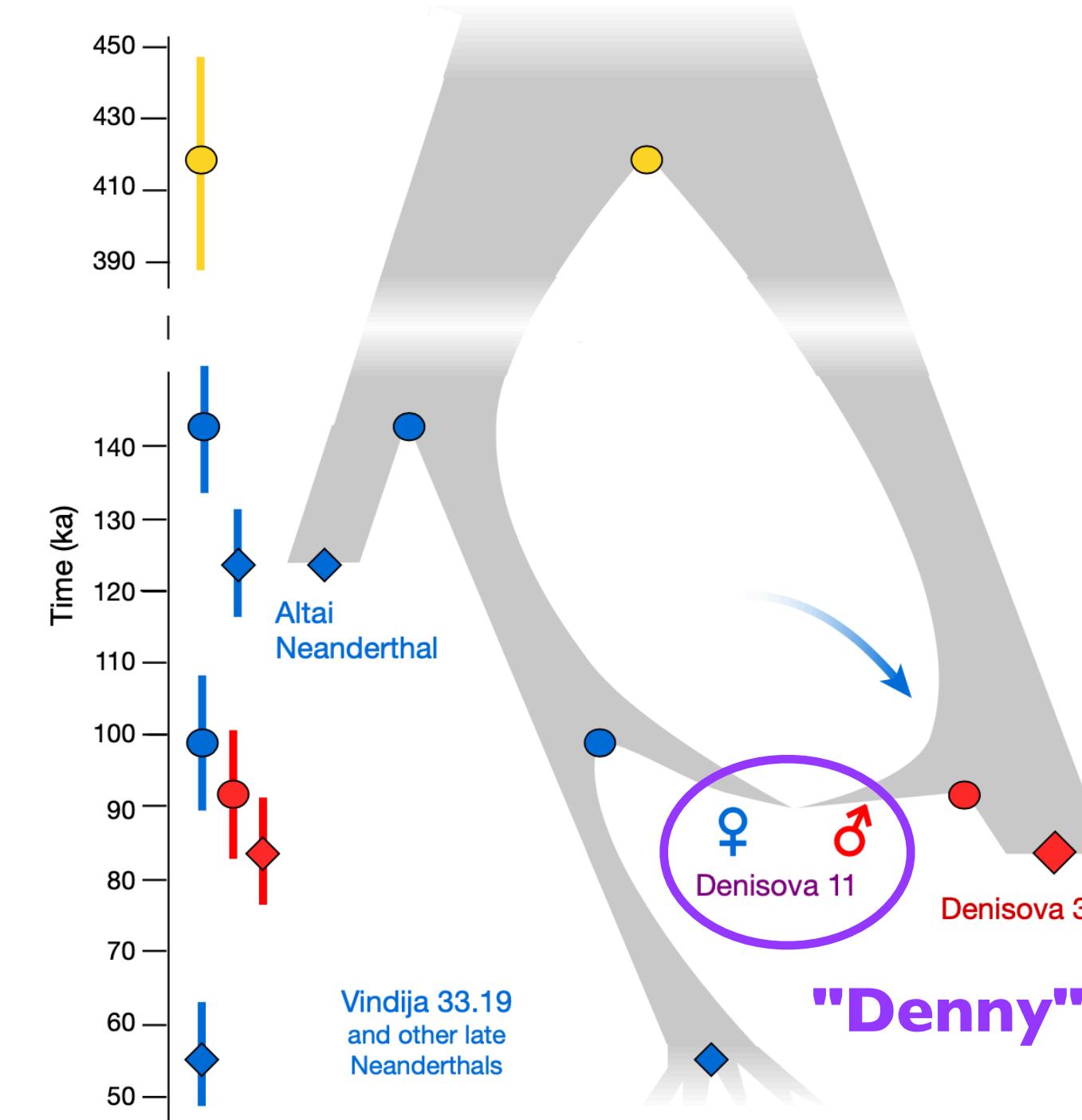
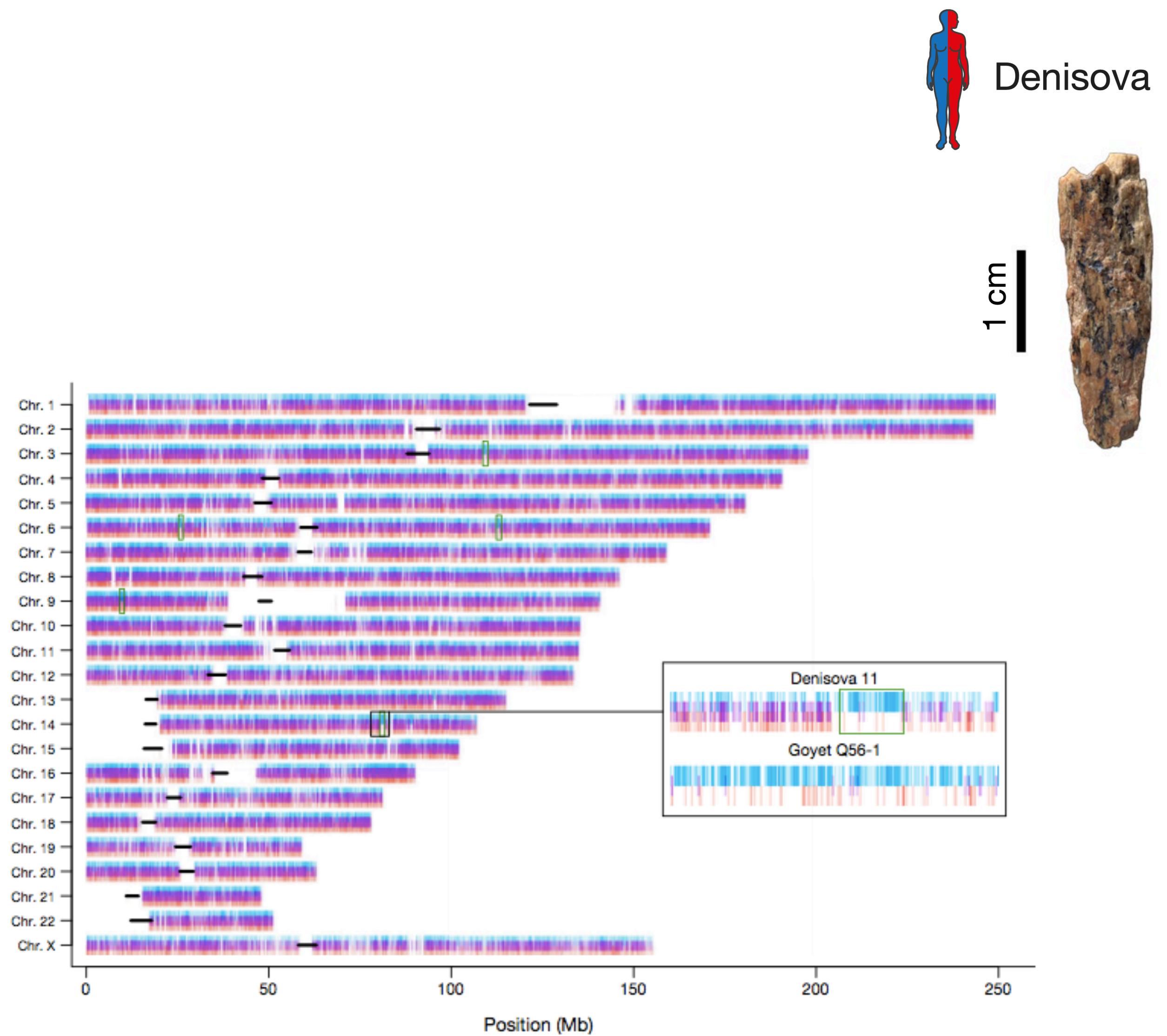
Denisova 11



1 cm

The genome of the offspring of a Neanderthal mother and a Denisovan father

Viviane Slon , Fabrizio Mafessoni, Benjamin Vernot, Cesare de Filippo, Steffi Grote, Bence Viola, Mateja Hajdinjak, Stéphane Peyrégne, Sarah Nagel, Samantha Brown, Katerina Douka, Tom Higham, Maxim B. Kozlikin, Michael V. Shunkov, Anatoly P. Derevianko, Janet Kelso, Matthias Meyer, Kay Prüfer & Svante Pääbo 





mp@bodkan.net



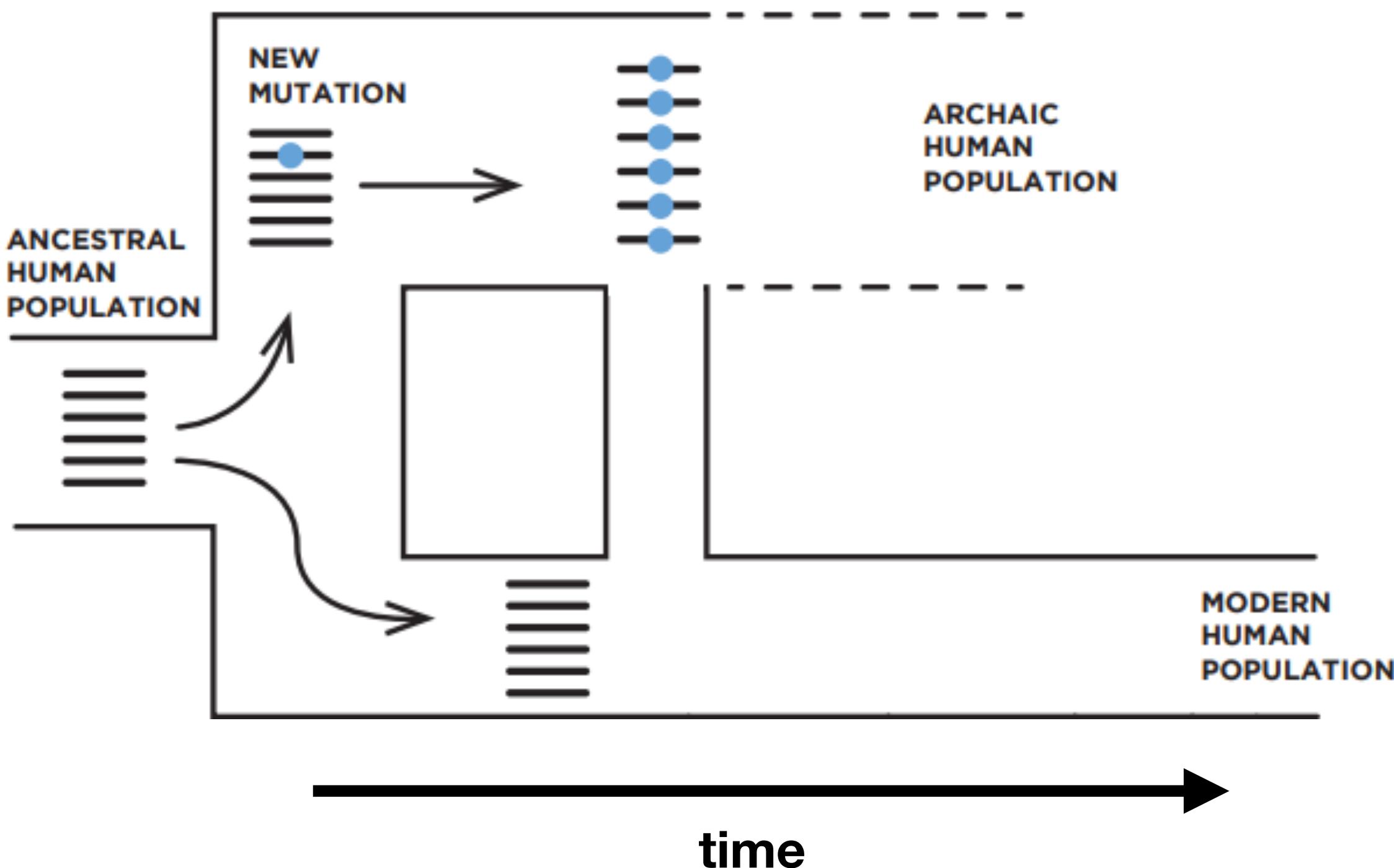
ABBA/BABA EXERCISES

github.com/bodkan/ku-introgression2026

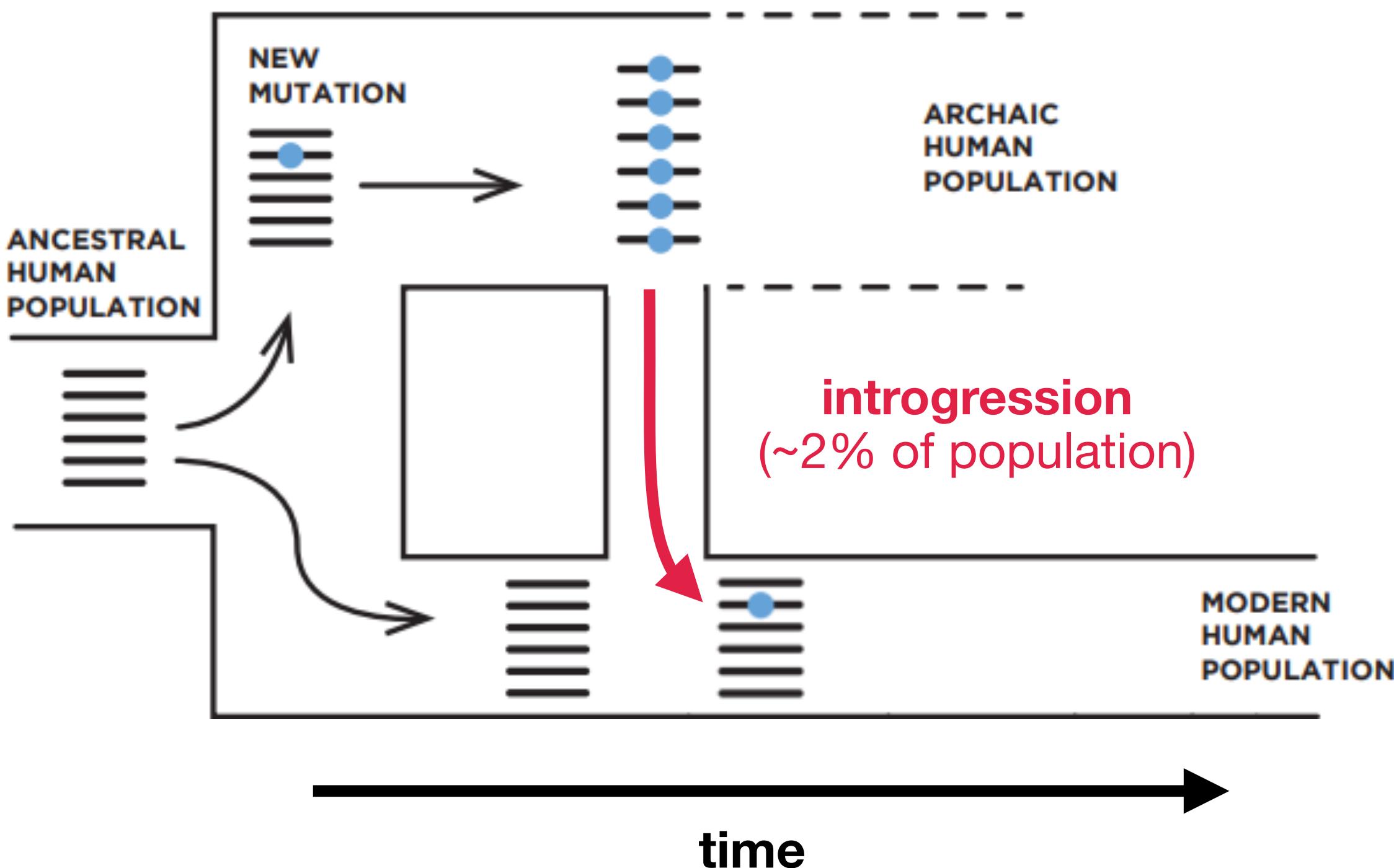
Bonus content

Consequences of introgression on human functional biology?

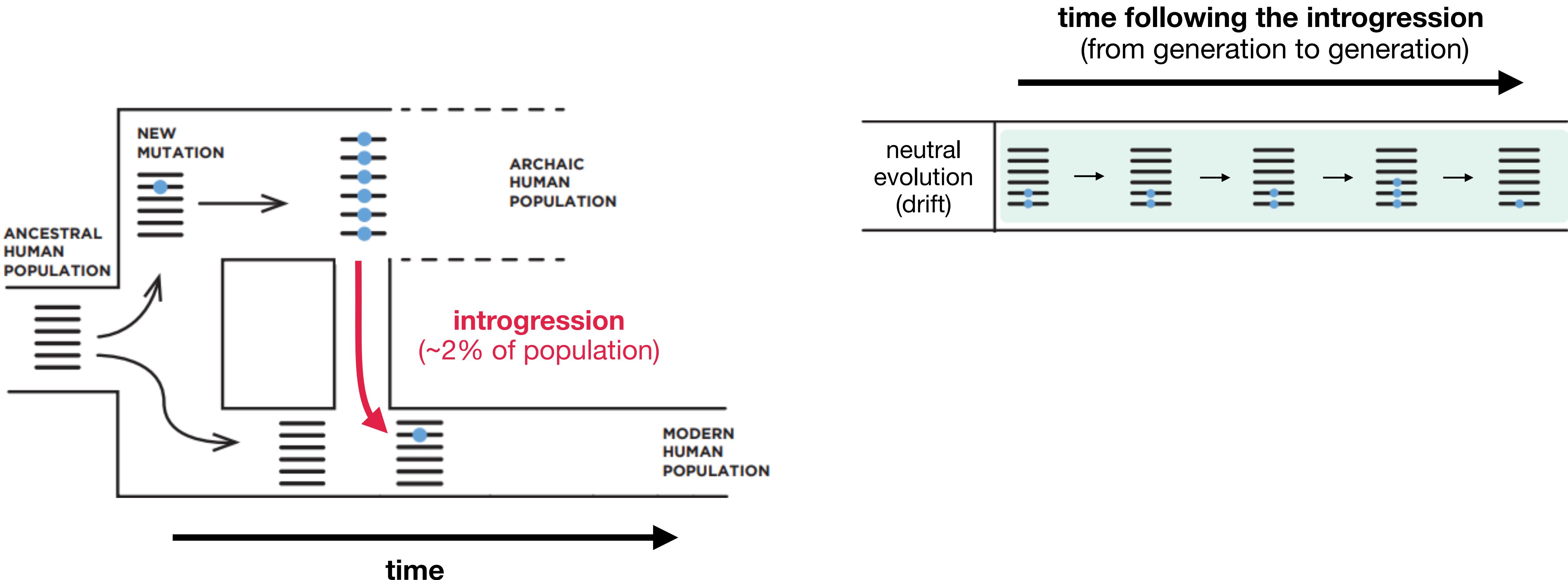
What are the possible fates of introgressed DNA?



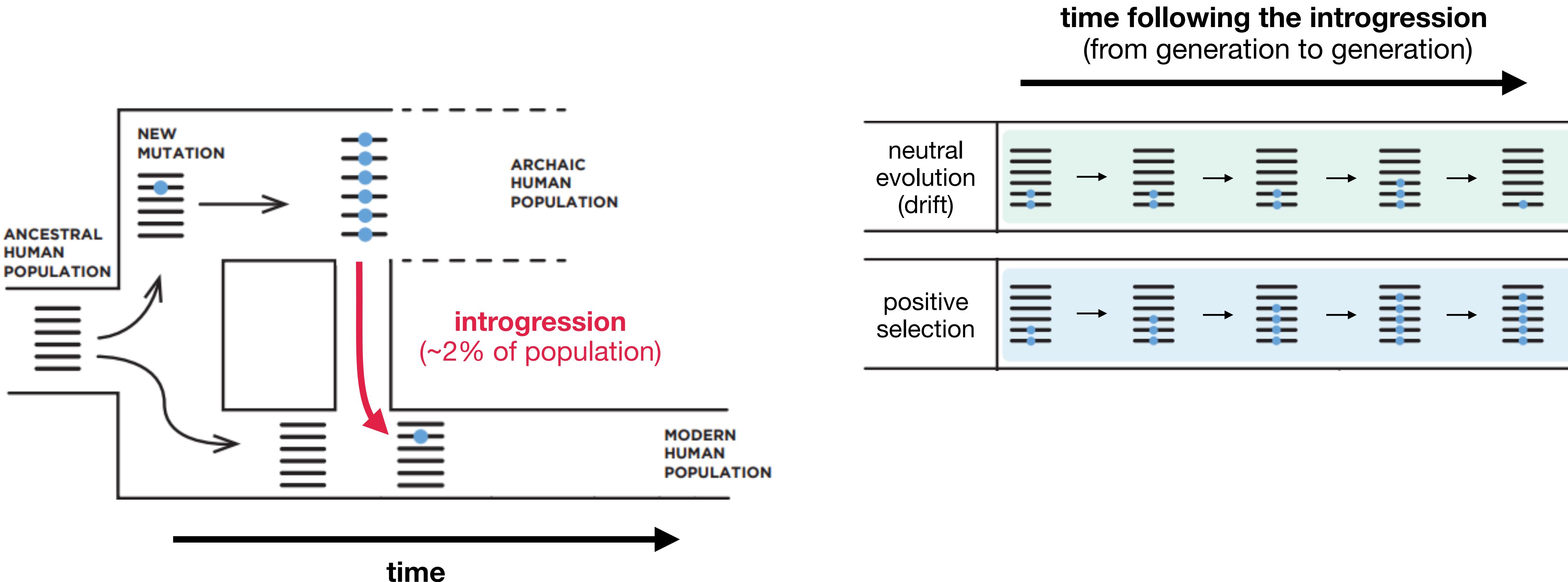
What are the possible fates of introgressed DNA?



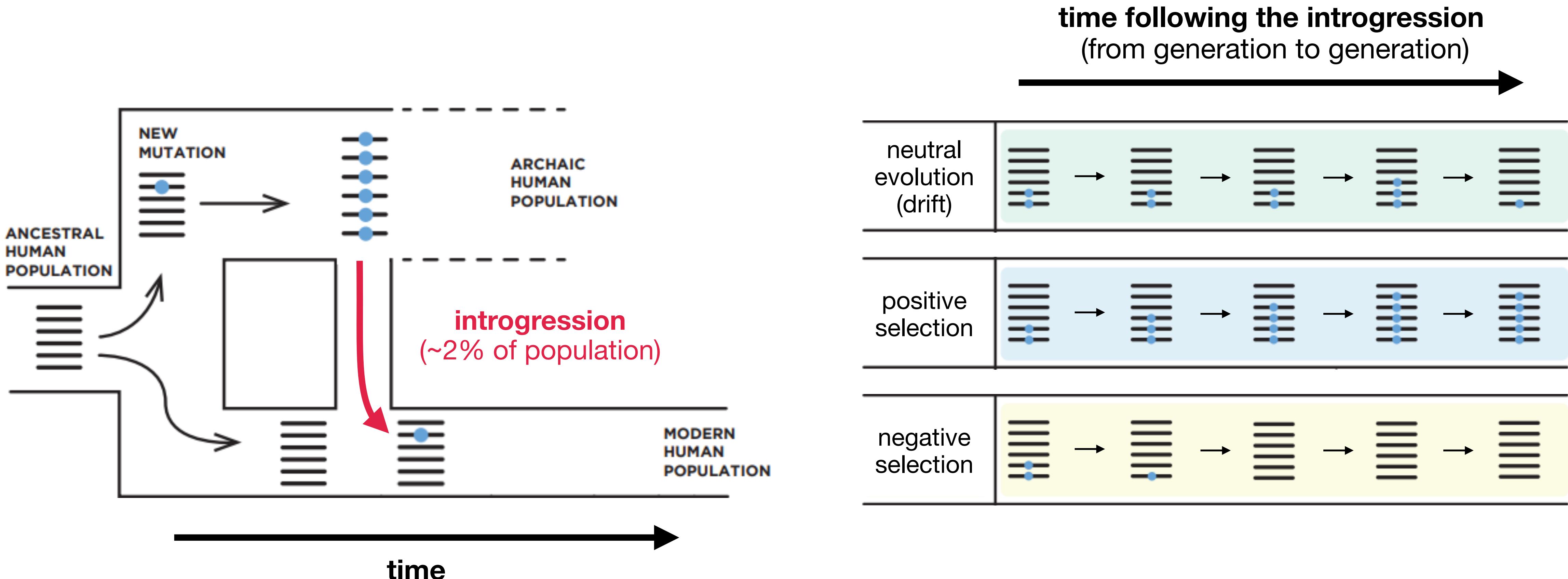
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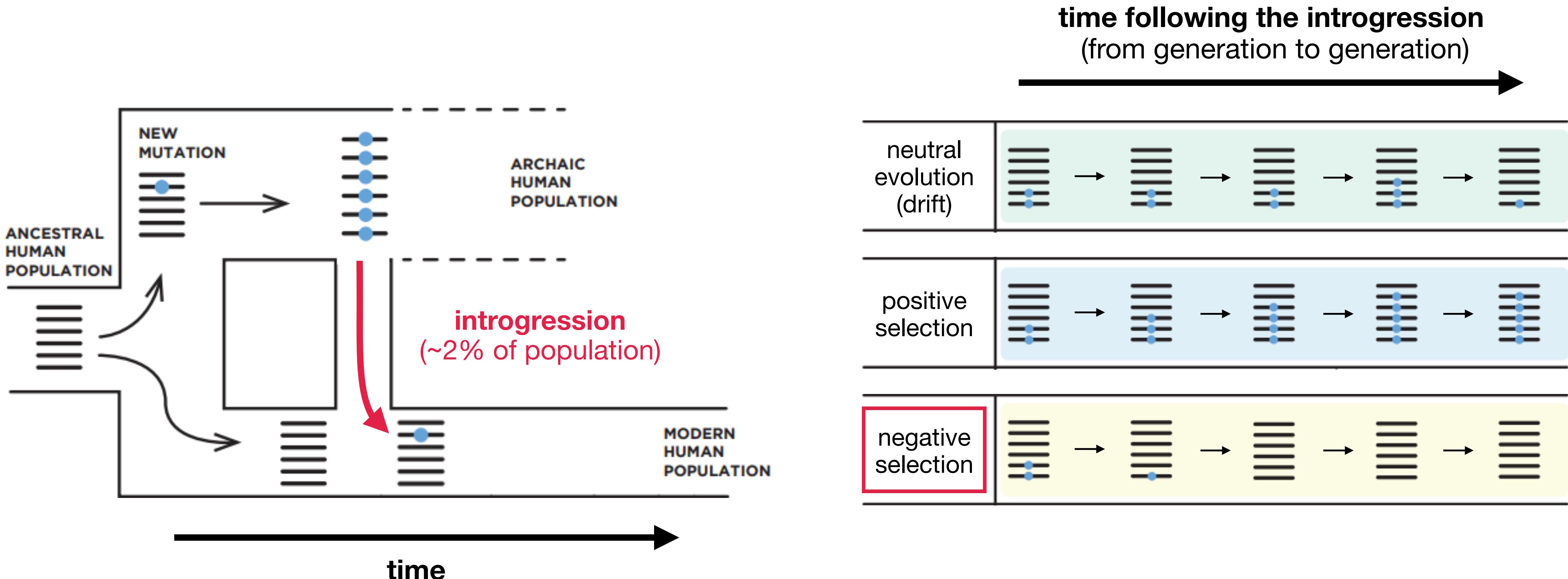
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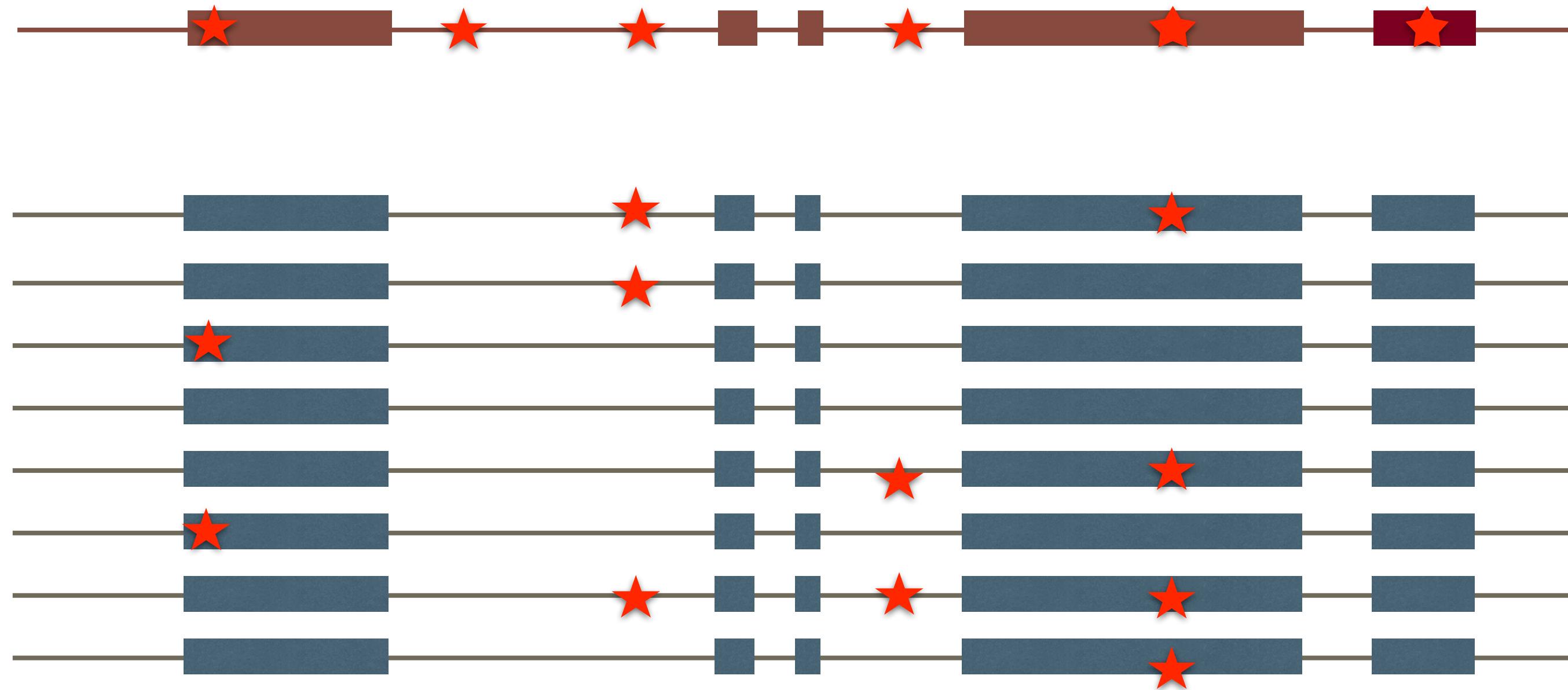


What are the possible fates of introgressed DNA?



What are the possible fates of introgressed DNA?





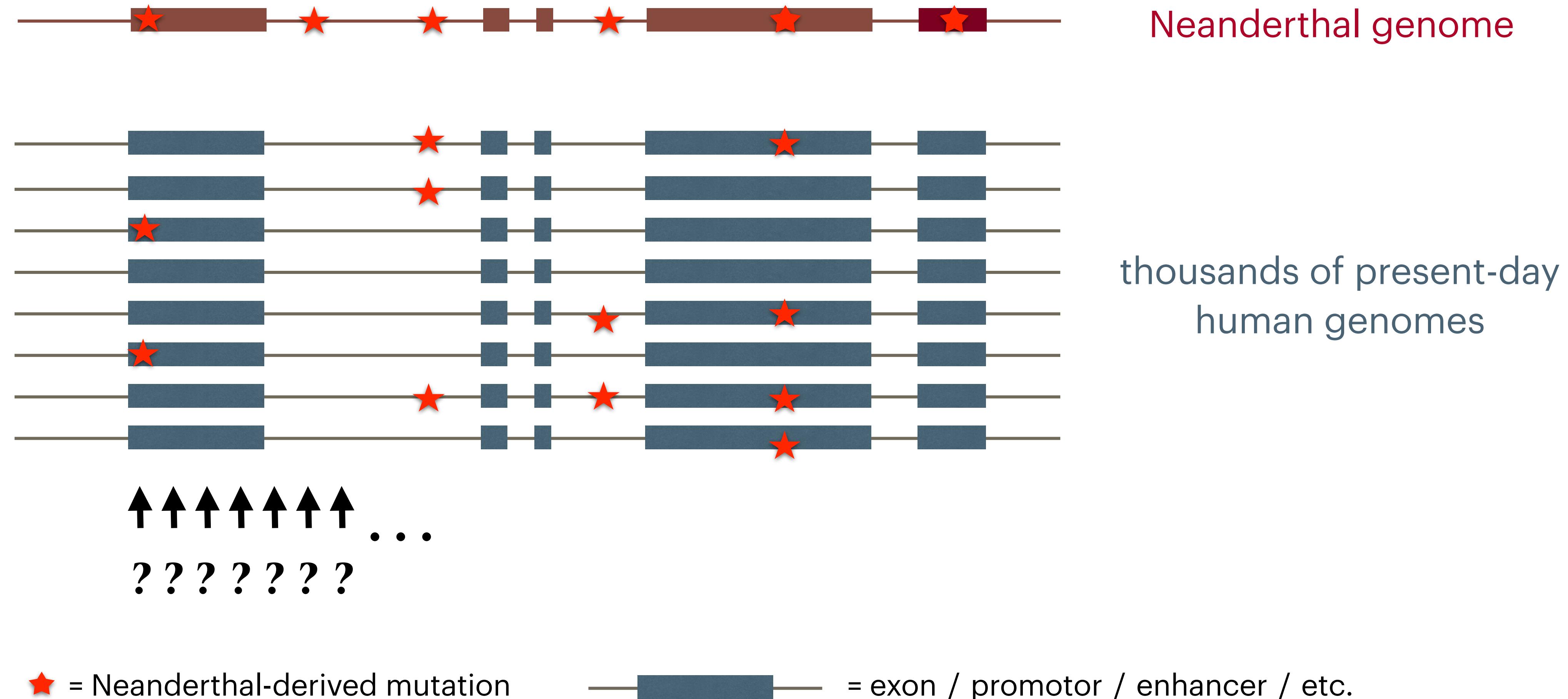
Neanderthal genome

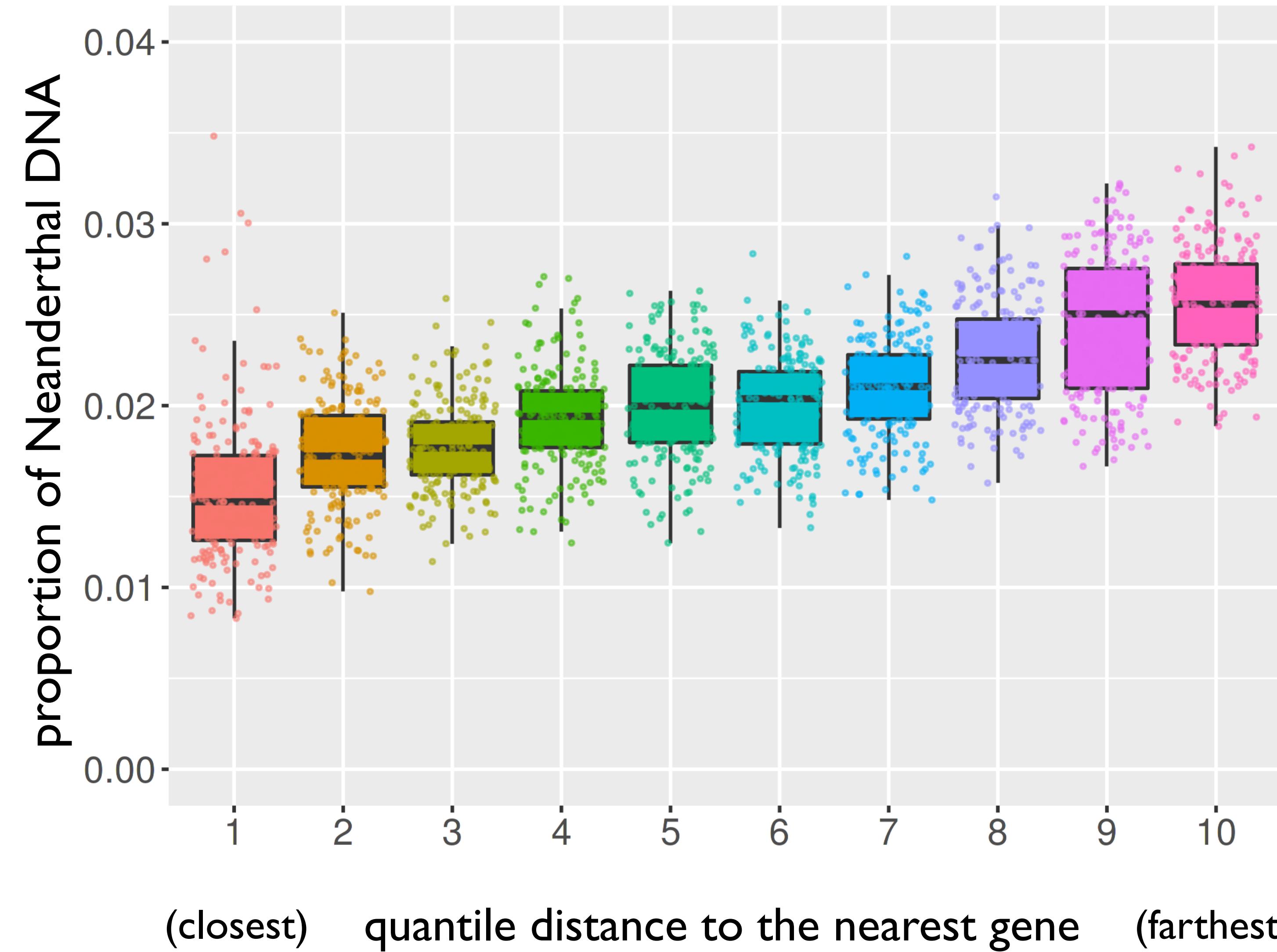
thousands of present-day
human genomes

★ = Neanderthal-derived mutation

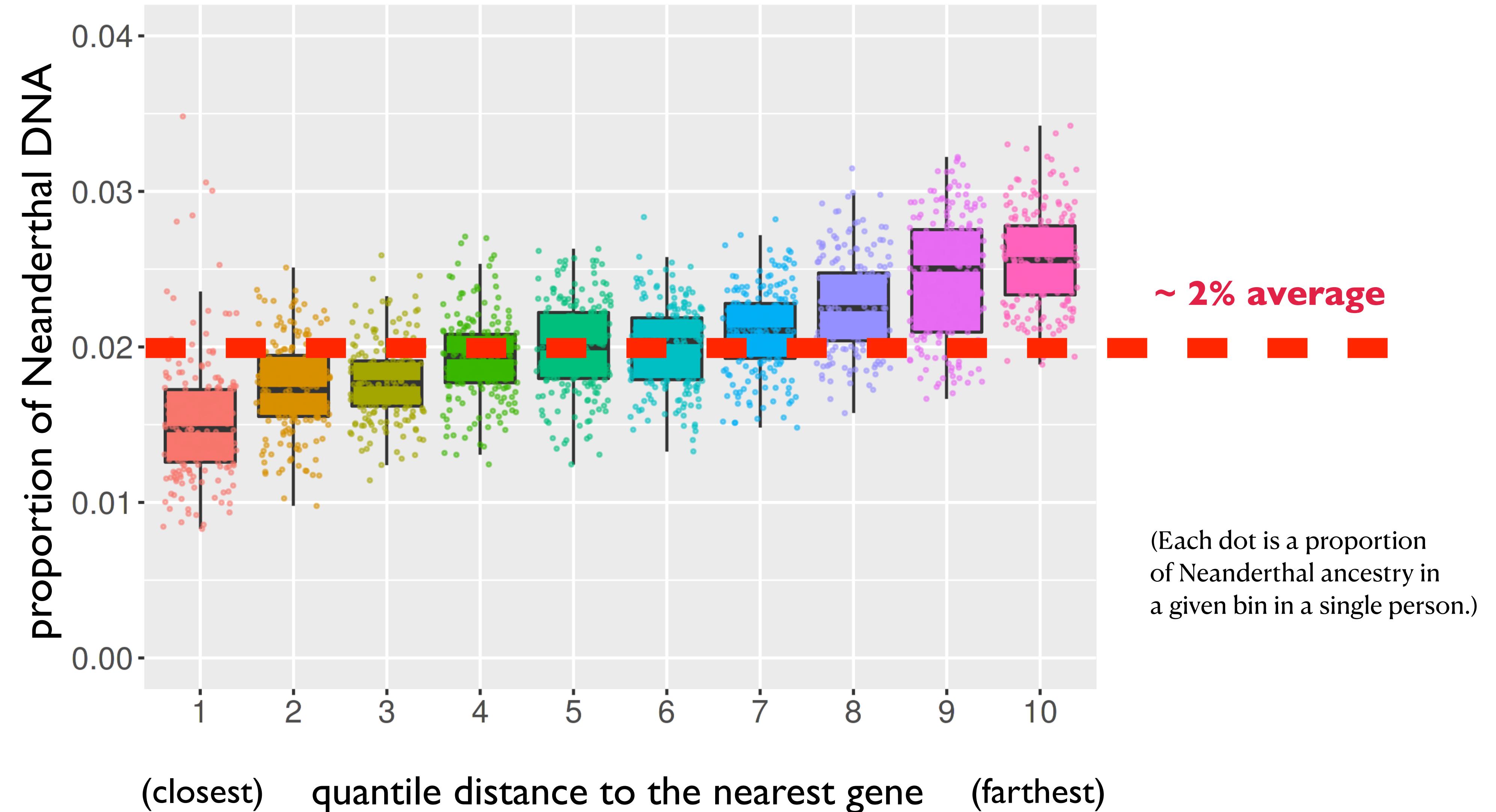
—■— = exon / promotor / enhancer / etc.

Is there a relationship between the presence of a Neanderthal allele and its distance to the nearest important gene?

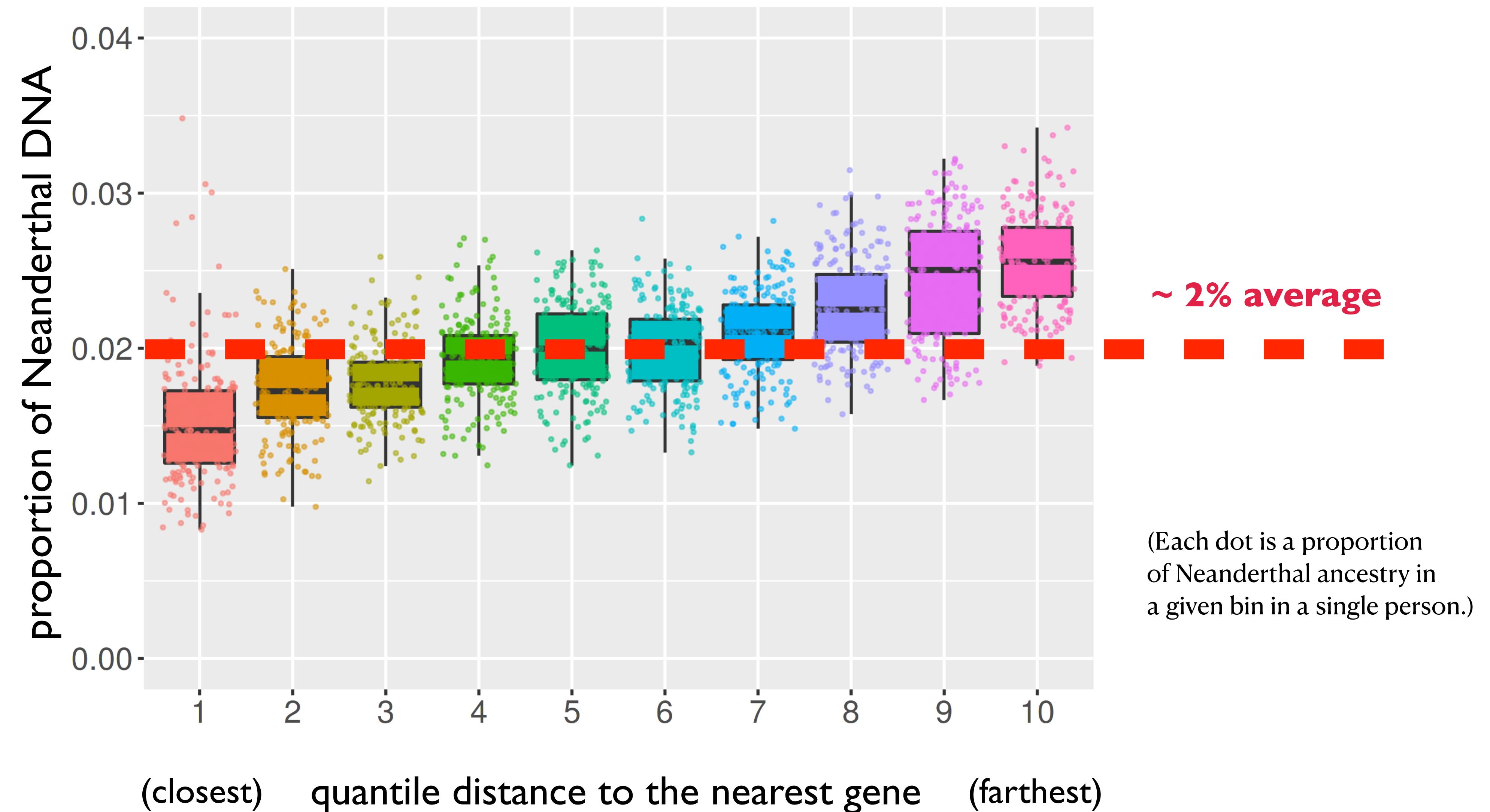




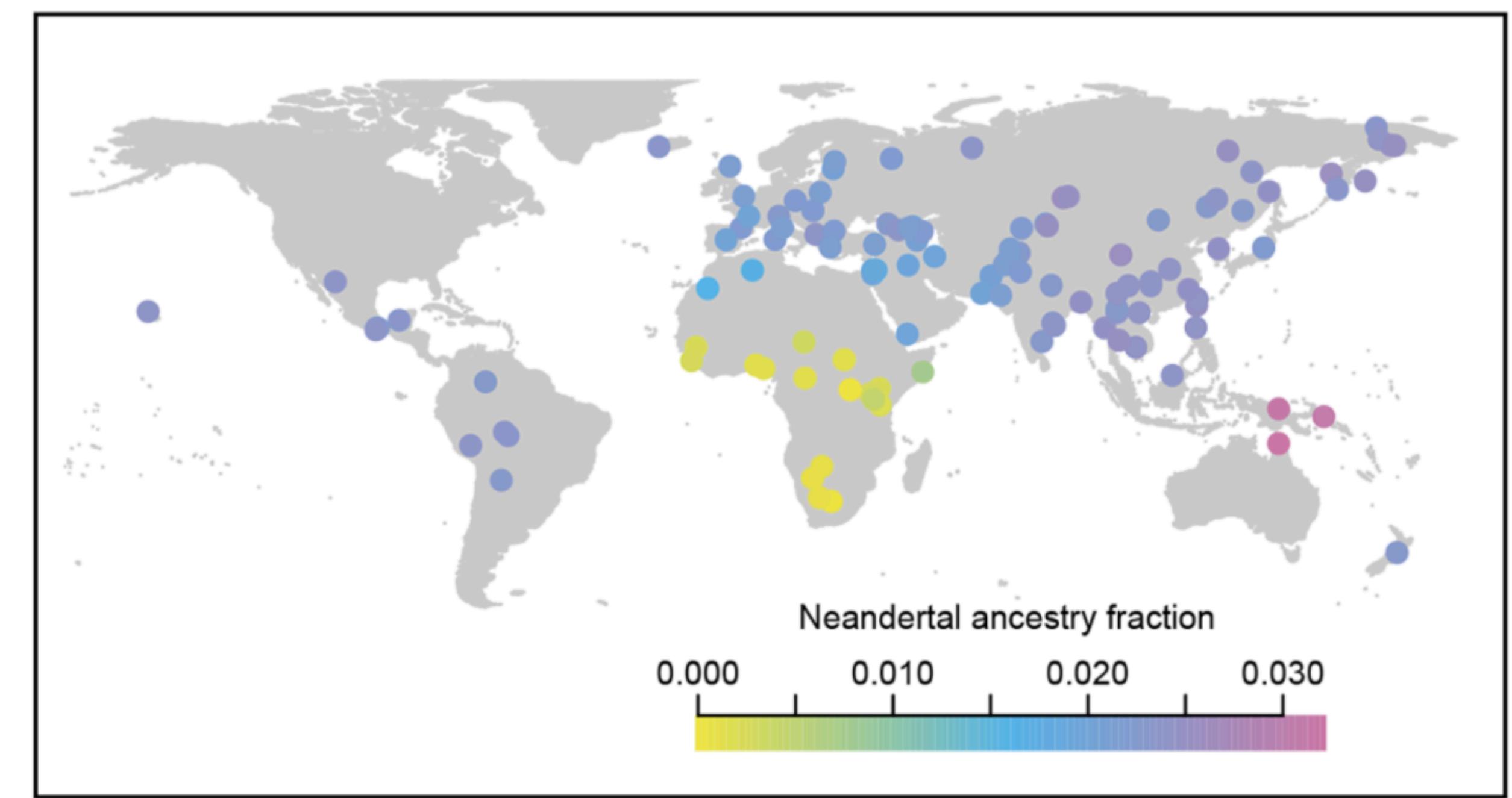
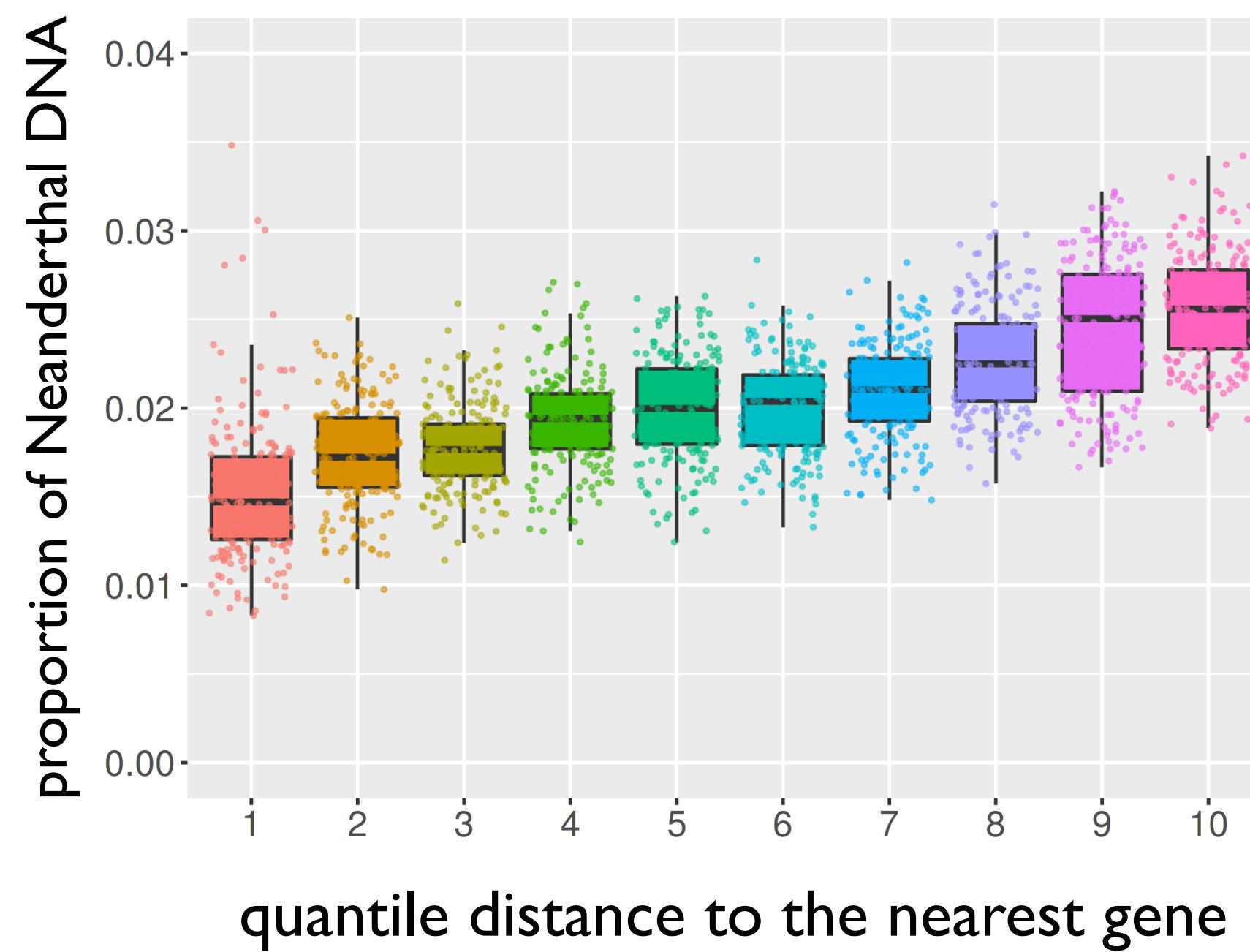
(Each dot is a proportion
of Neanderthal ancestry in
a given bin in a single person.)



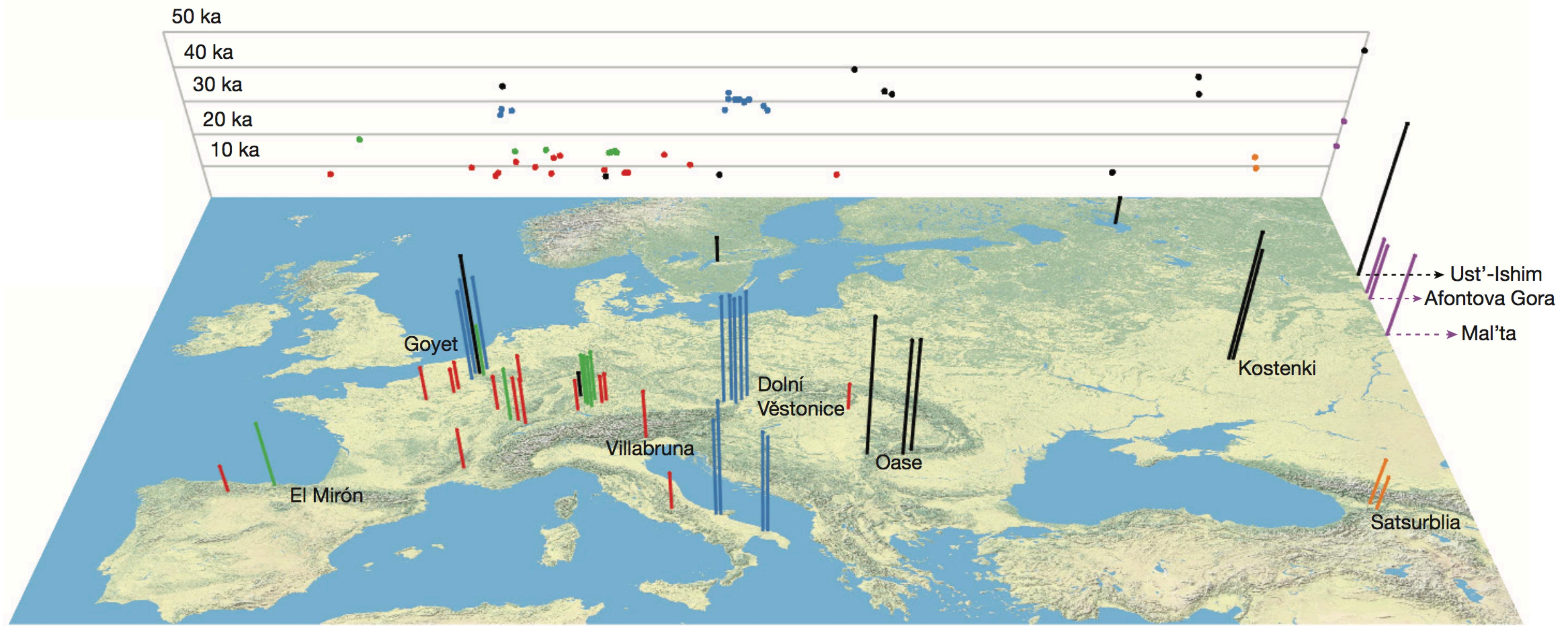
Genome-wide selection *against* Neanderthal DNA!



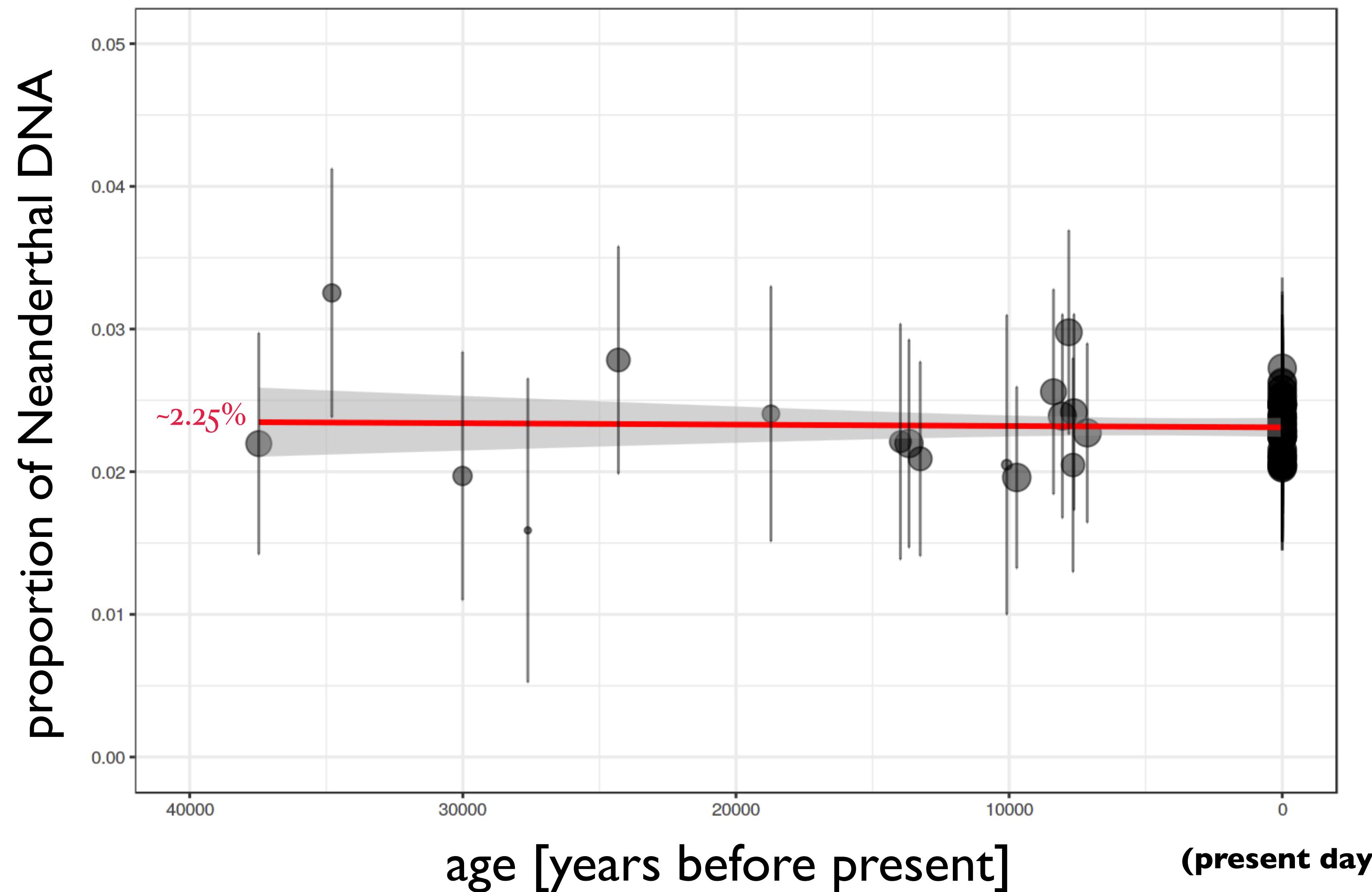
On what time scale did this happen?



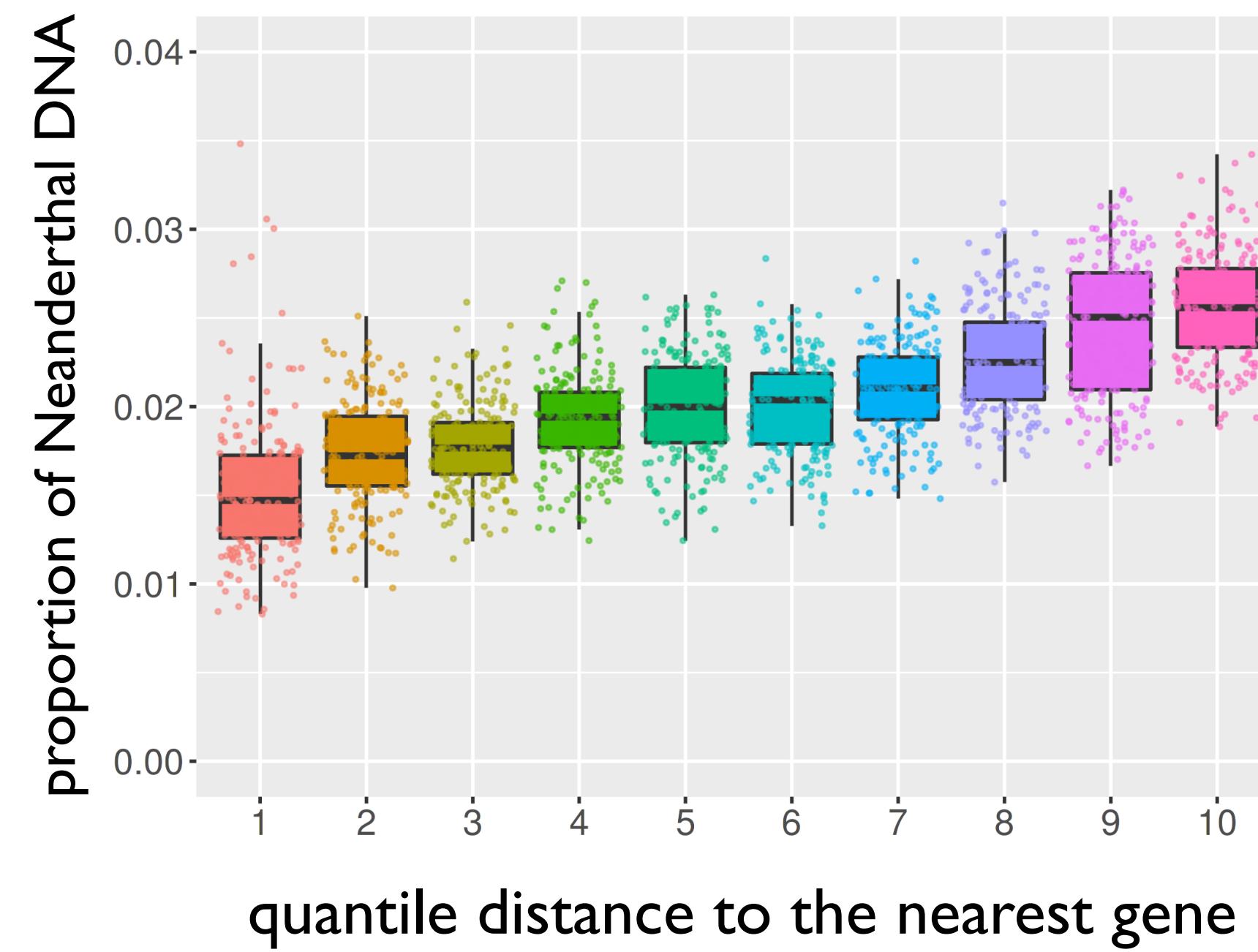
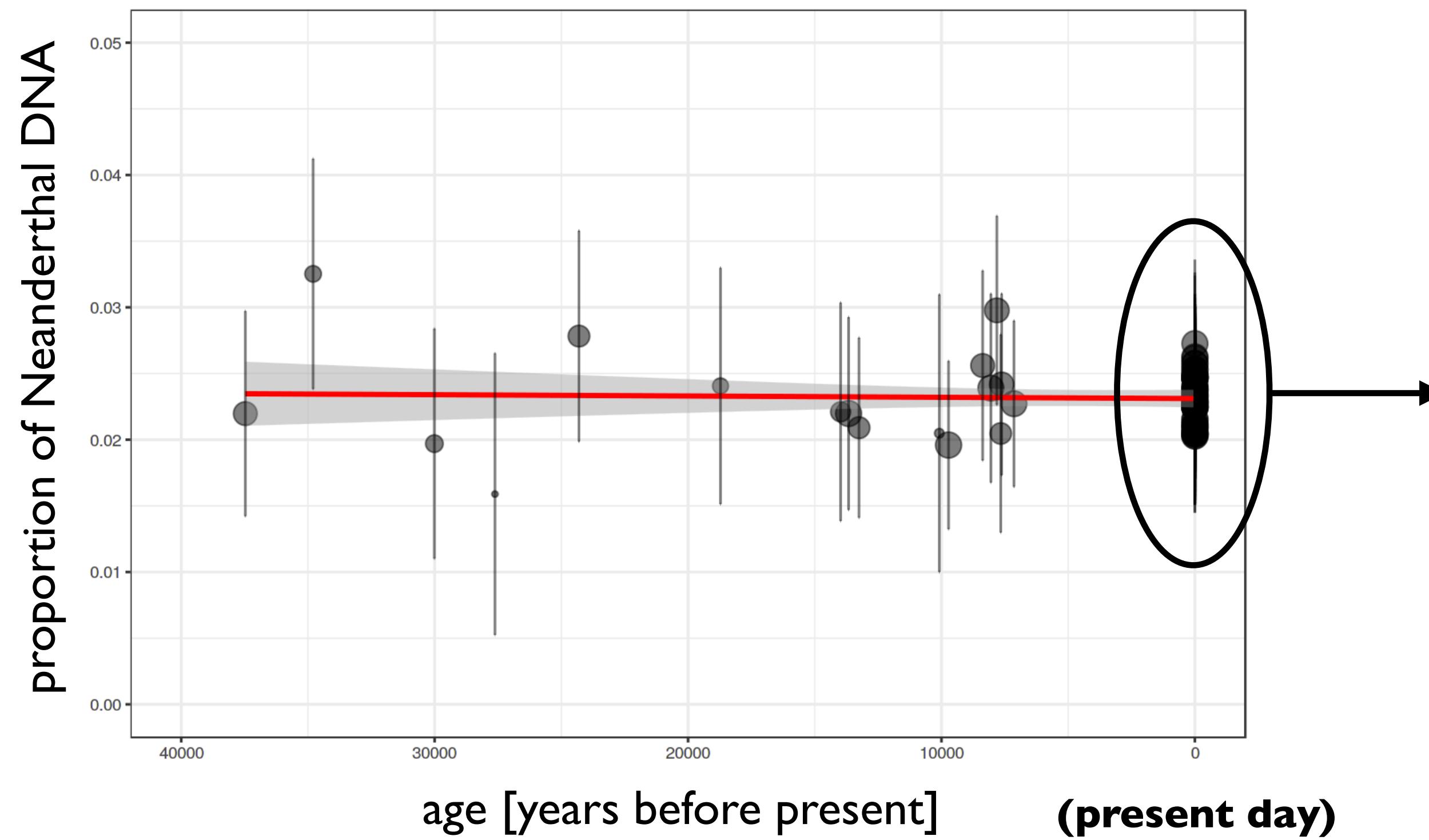
aDNA of “early-modern humans” from Europe (45-10 kya BP)



Proportion of Neanderthal ancestry in modern humans remained constant for tens of thousands of years

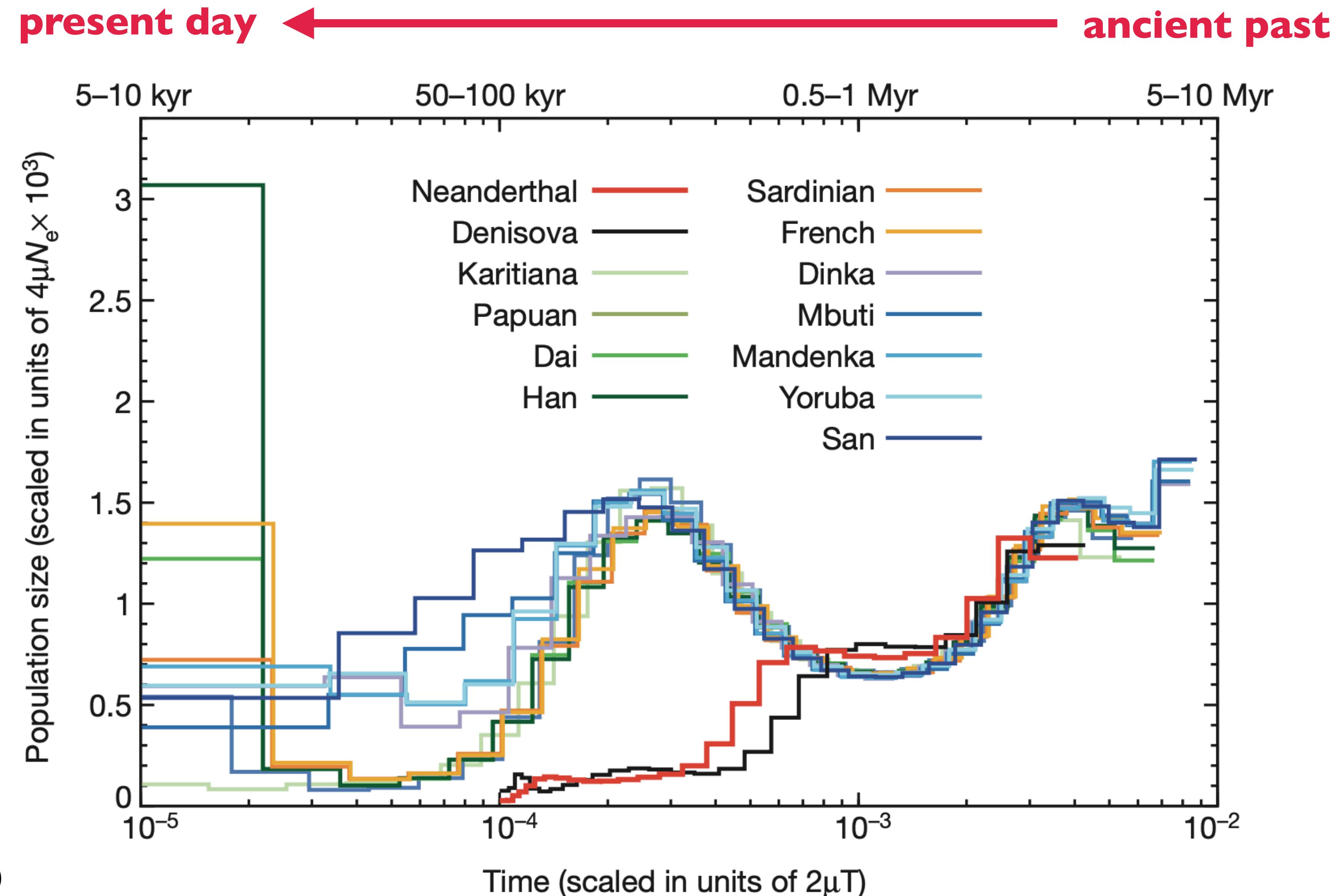


Empirical trajectory of Neanderthal ancestry contradicts the signal of negative selection?



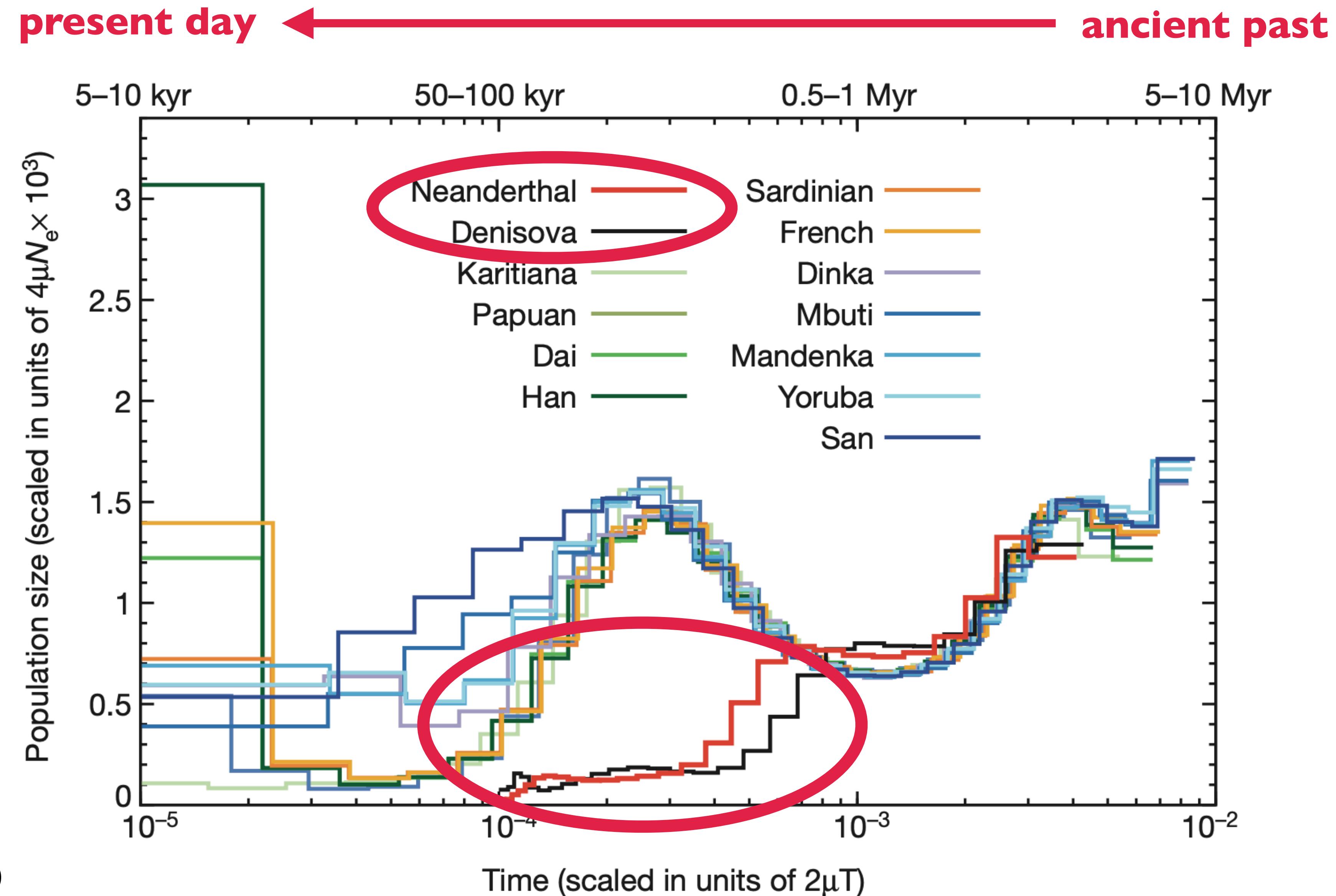
Demographic collapse of archaic human populations

Pairwise Sequentially
Markovian Coalescent
(PSMC) model



Demographic collapse of archaic human populations

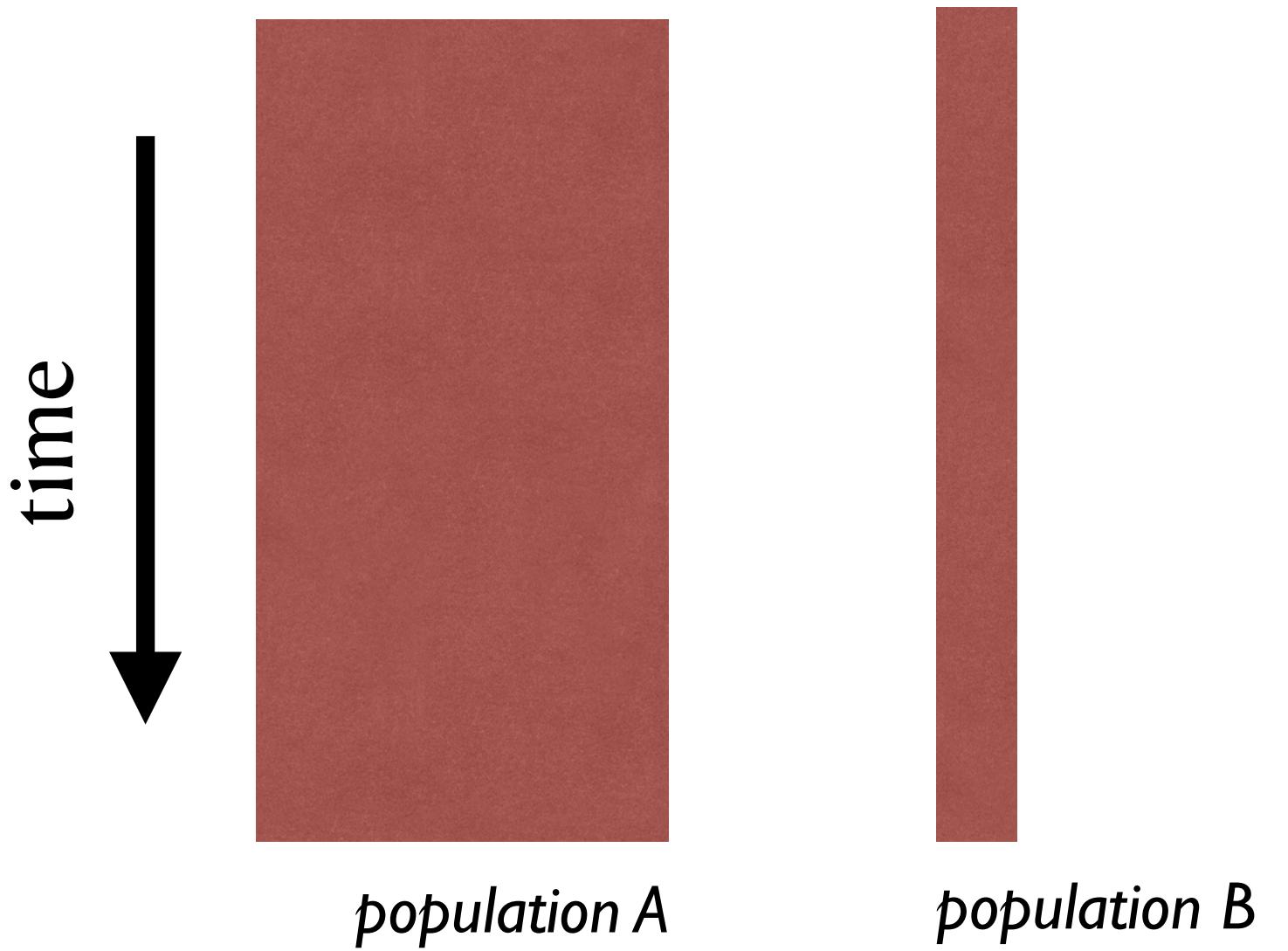
Pairwise Sequentially
Markovian Coalescent
(PSMC) model



**What was the impact of this on
accumulation of deleterious mutations?**

Nearly-neutral theory of molecular evolution

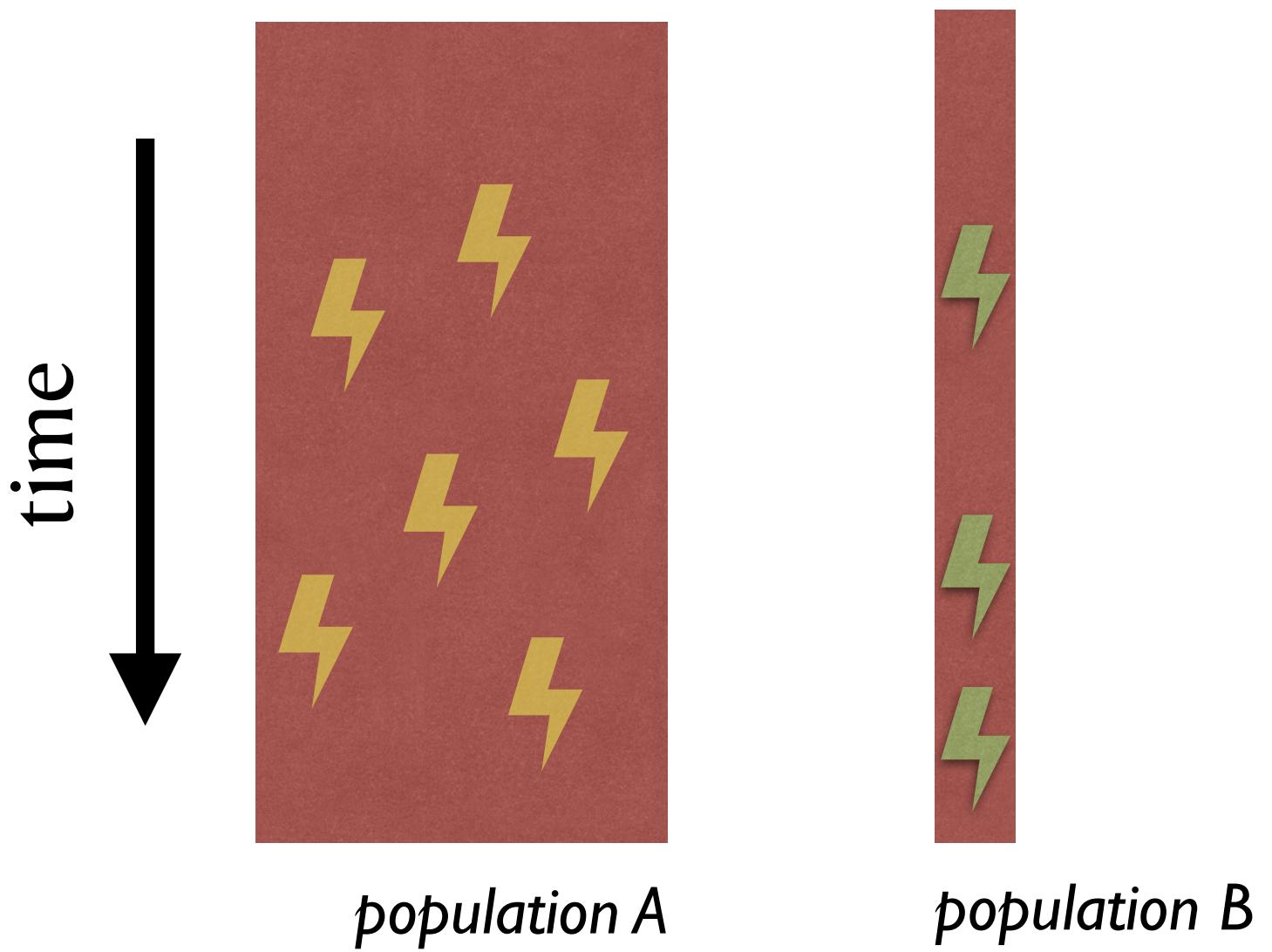
**Tomoko
Ohta**



<https://womentyoushouldknow.net/population-genetics-tomoko-ohta/>

Nearly-neutral theory of molecular evolution

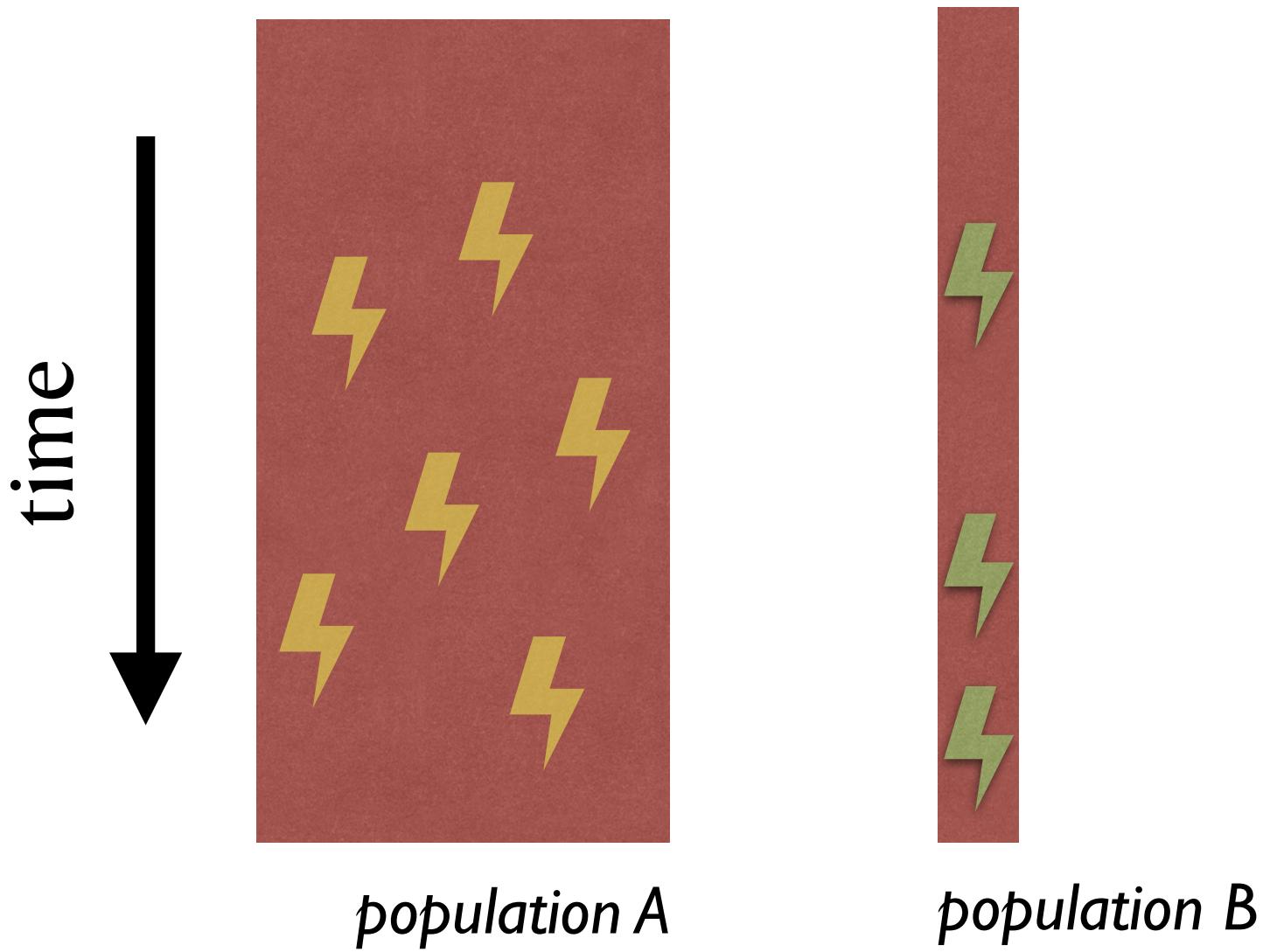
**Tomoko
Ohta**



We know that genomes accumulate mutations

Nearly-neutral theory of molecular evolution

**Tomoko
Ohta**



We know that genomes accumulate mutations

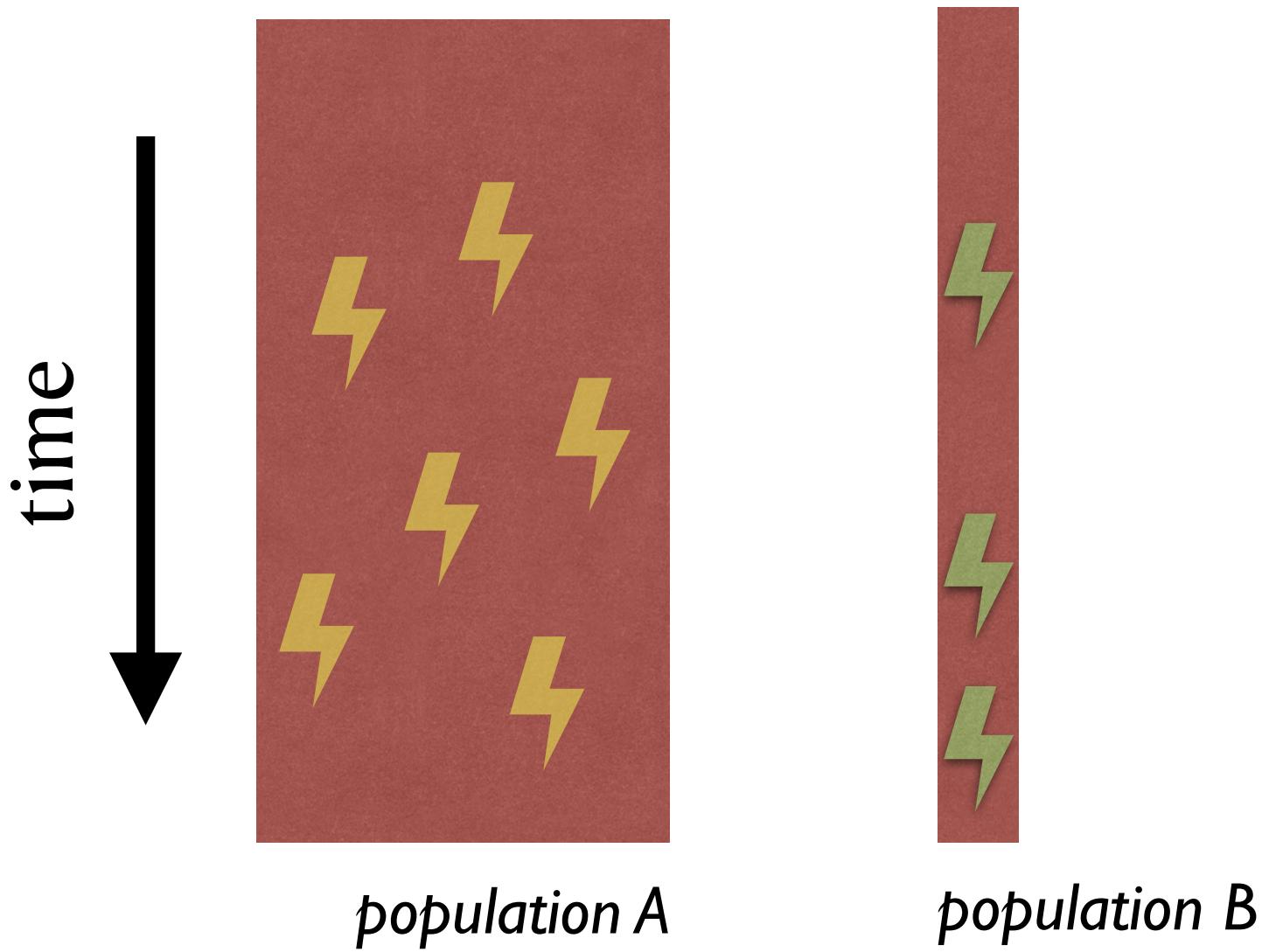
- a tiny number are extremely negative (**lethal**)
- a tiny number are positive (**adaptive**)
- a large number does nothing (**neutral**)
- many are “mildly deleterious” (**nearly neutral**)

Nearly-neutral theory of molecular evolution

**Tomoko
Ohta**



<https://womentyoushouldknow.net/population-genetics-tomoko-ohta/>



We know that genomes accumulate mutations

- a tiny number are extremely negative (**lethal**)
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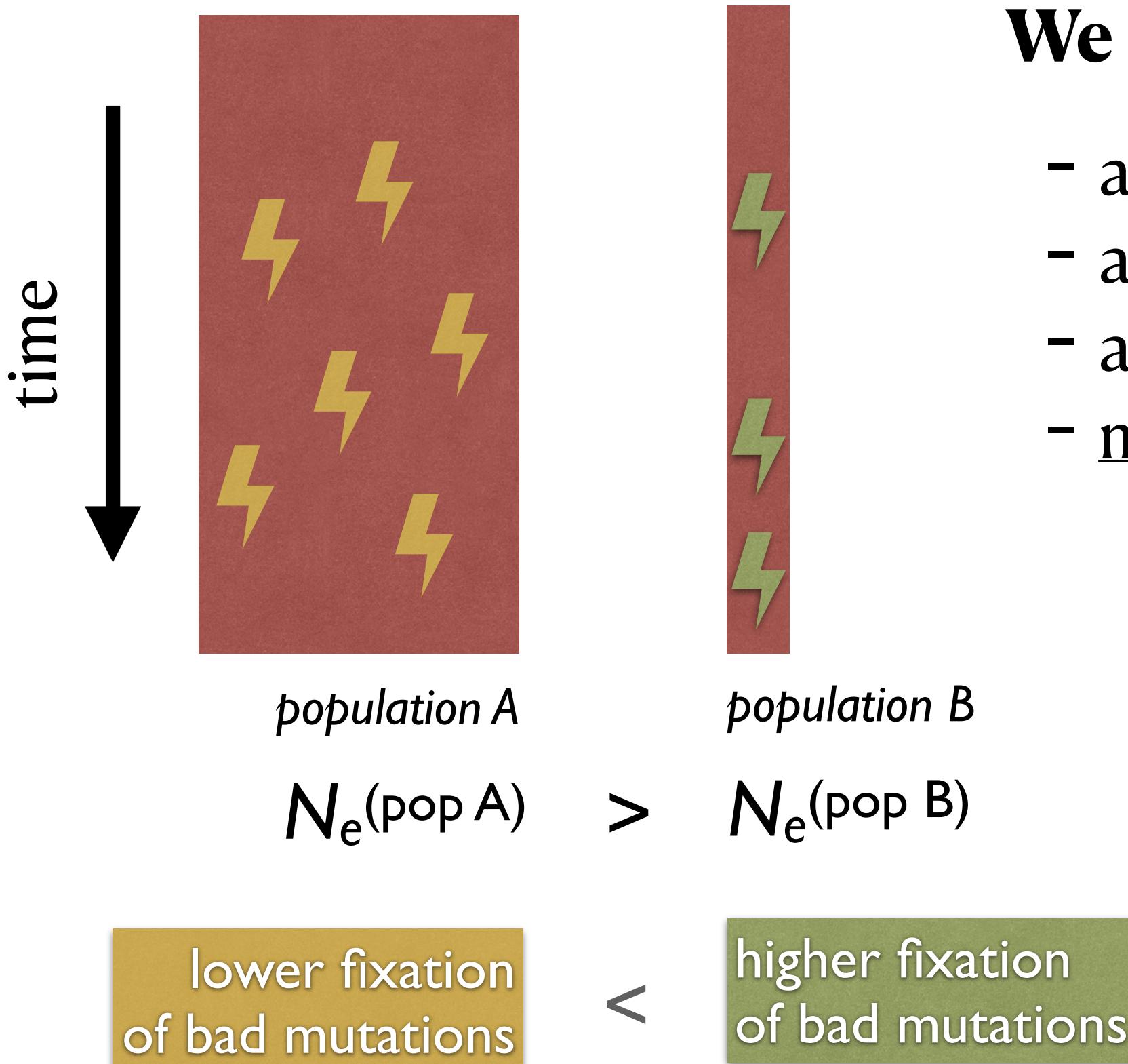
**Efficacy of negative selection
depends on the effective
population size (N_e).**

Nearly-neutral theory of molecular evolution

**Tomoko
Ohta**



<https://womentyoushouldknow.net/population-genetics-tomoko-ohta/>



We know that genomes accumulate mutations

- a tiny number are extremely negative (**lethal**)
- a tiny number are positive (**adaptive**)
- a large number does nothing (**neutral**)
- many are “mildly deleterious” (**nearly neutral**)

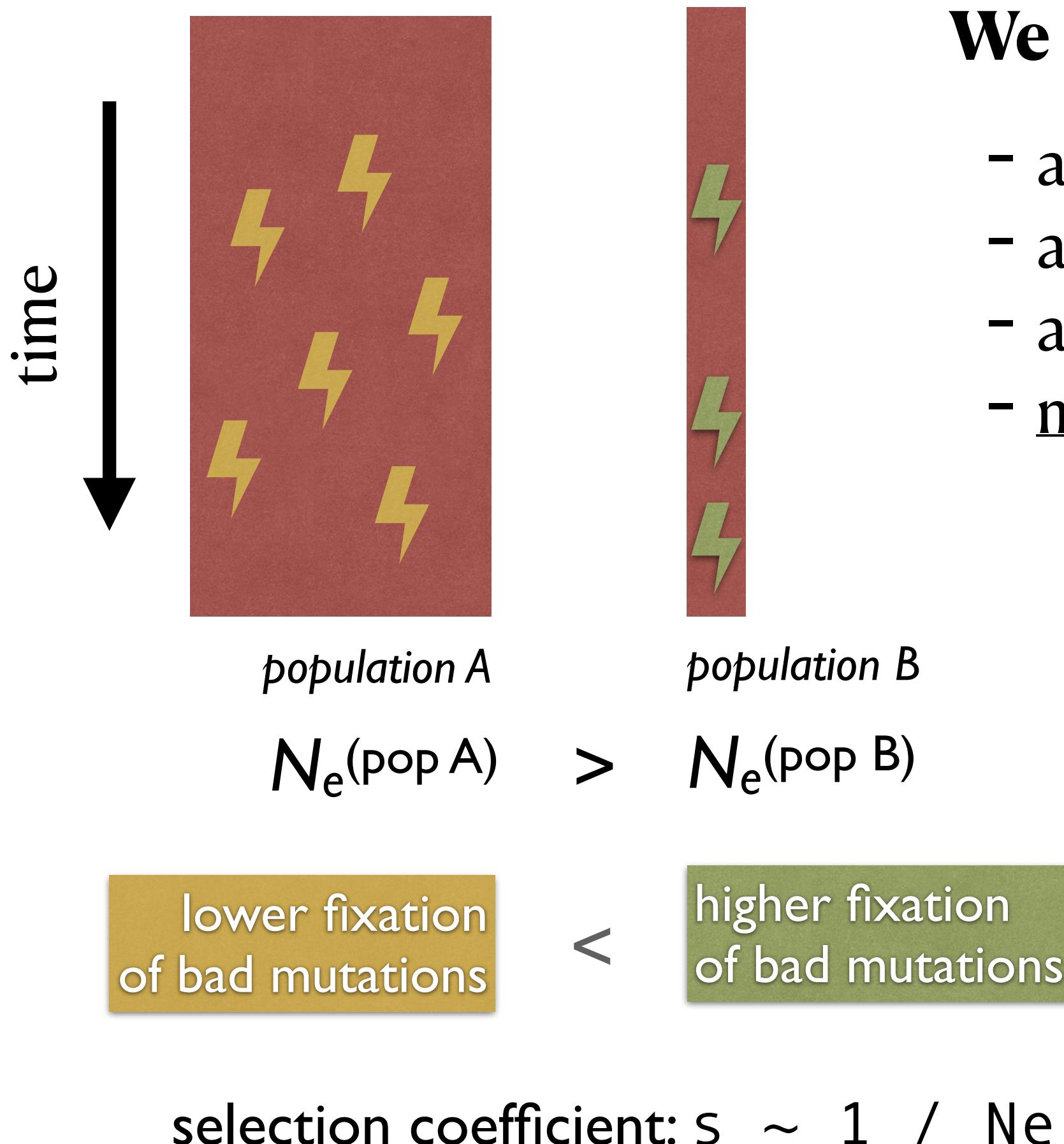
Efficacy of negative selection depends on the effective population size (N_e).

Nearly-neutral theory of molecular evolution

**Tomoko
Ohta**



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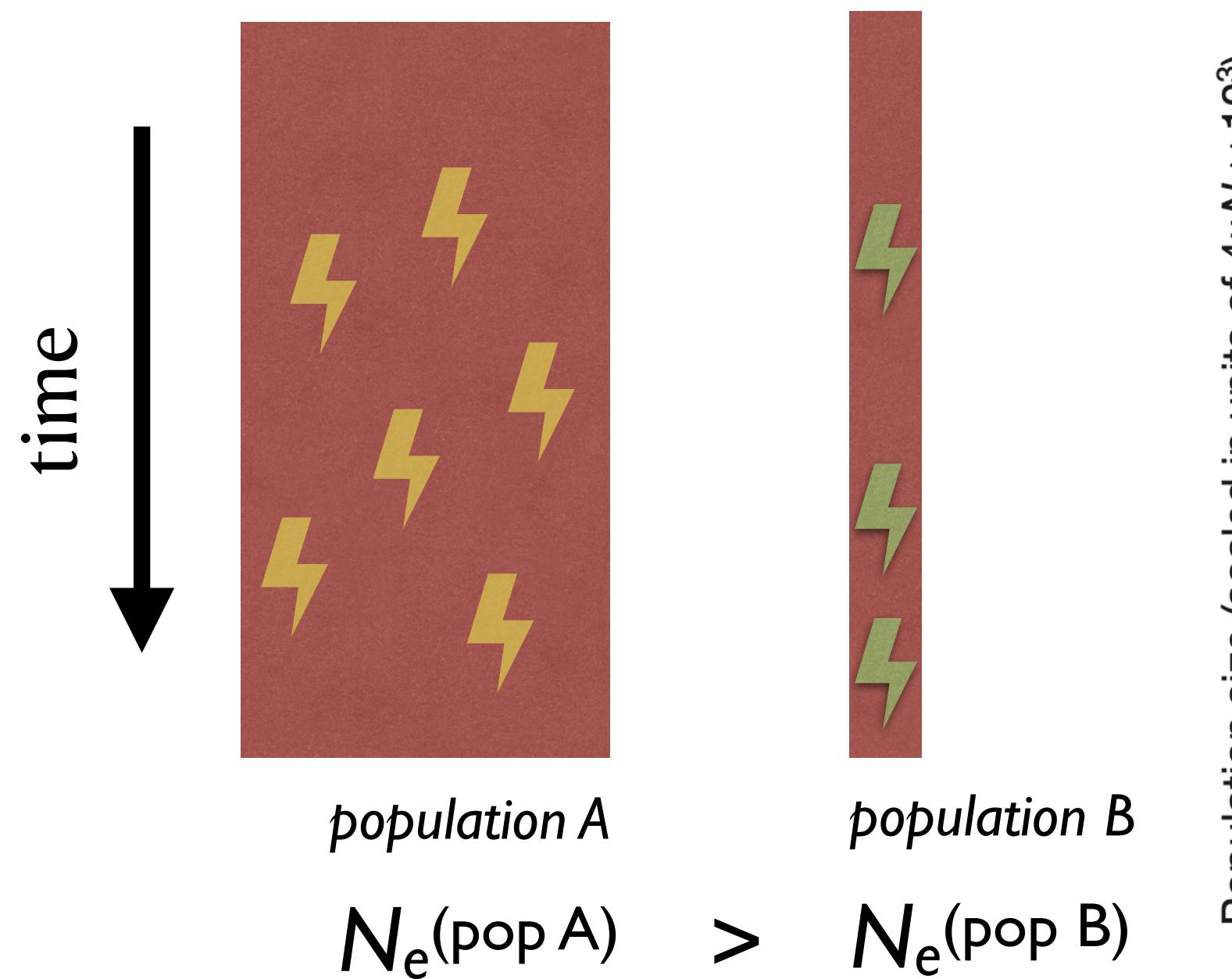
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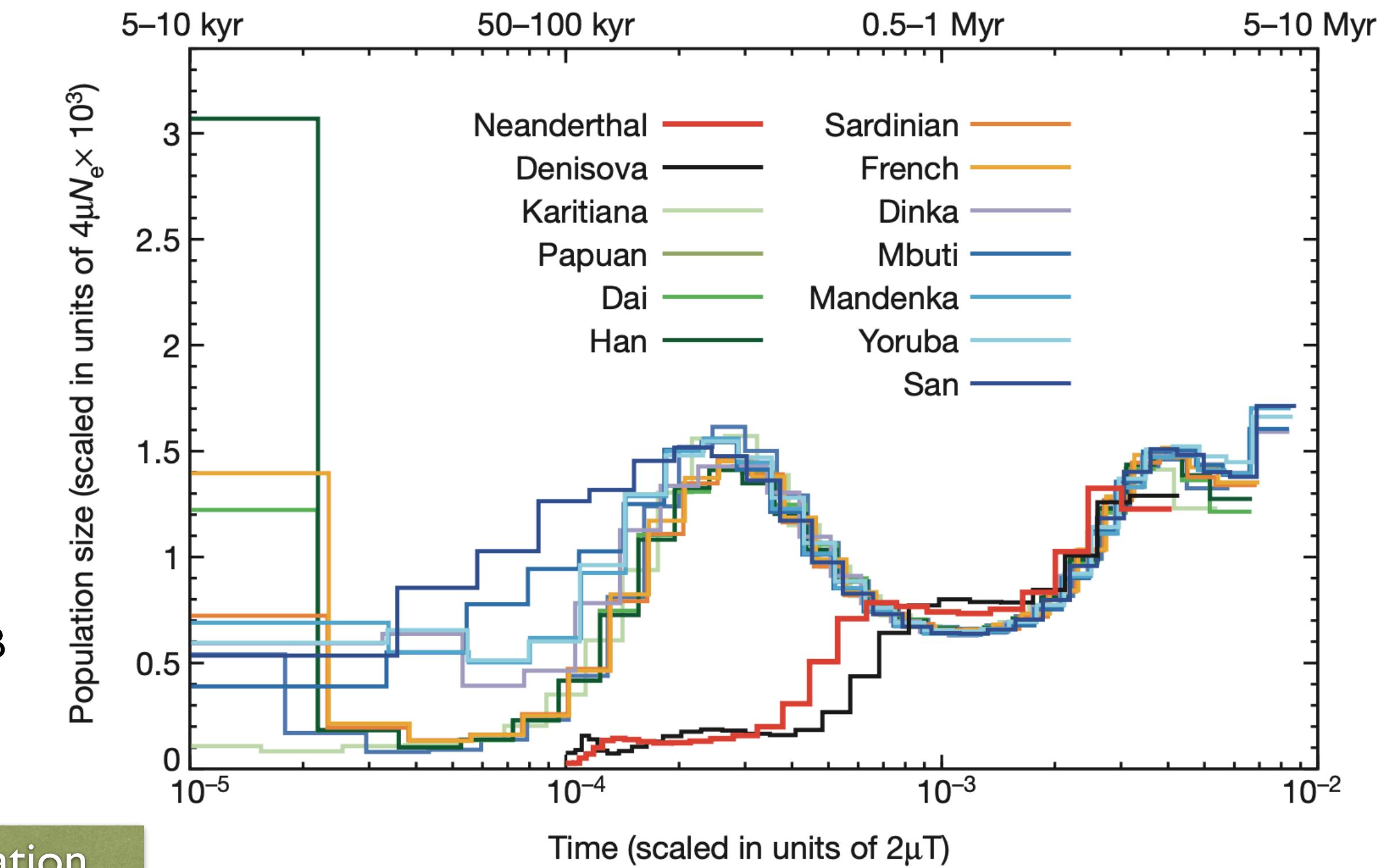


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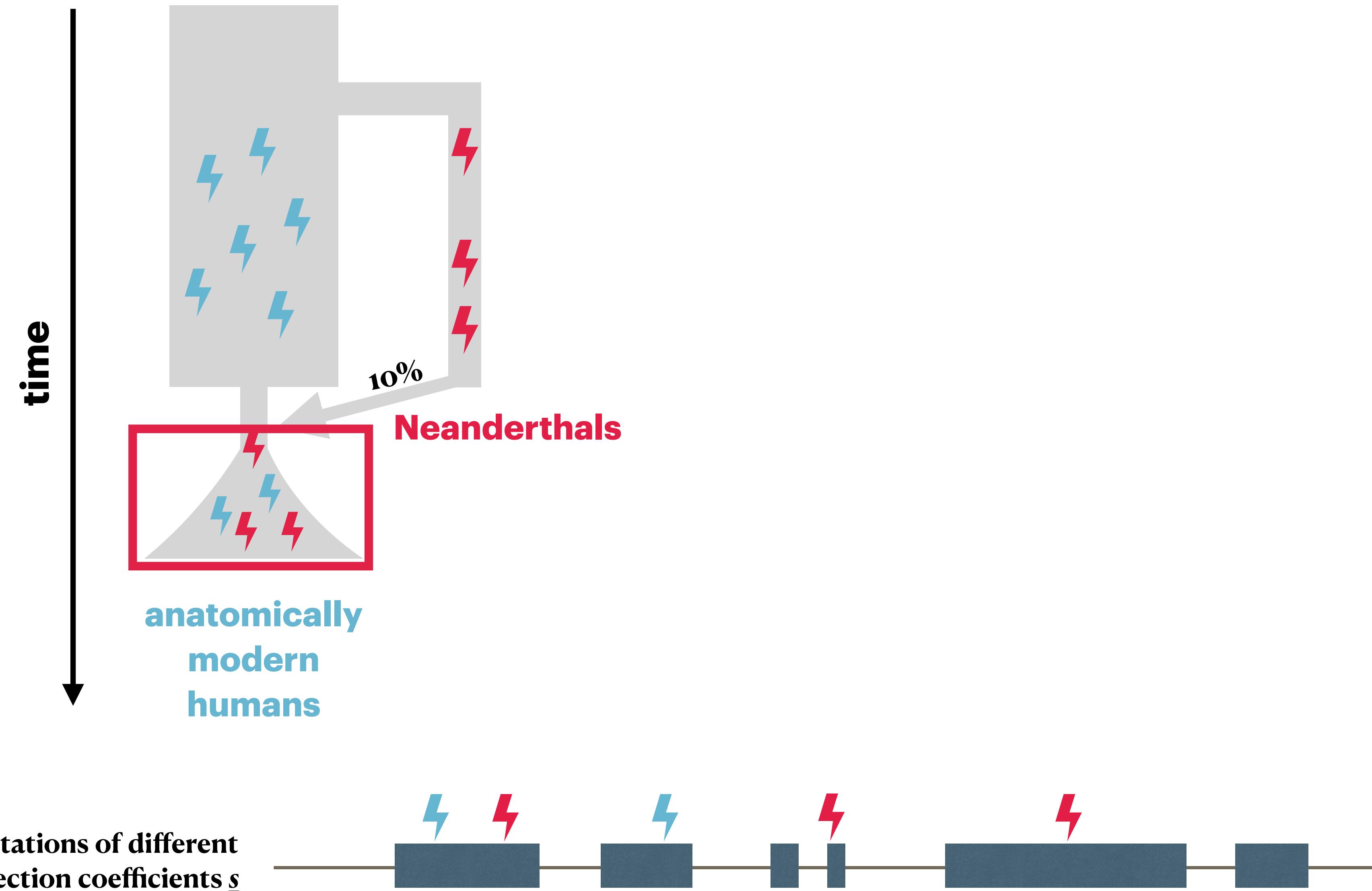
"anatomically
modern humans"

"Neanderthals"

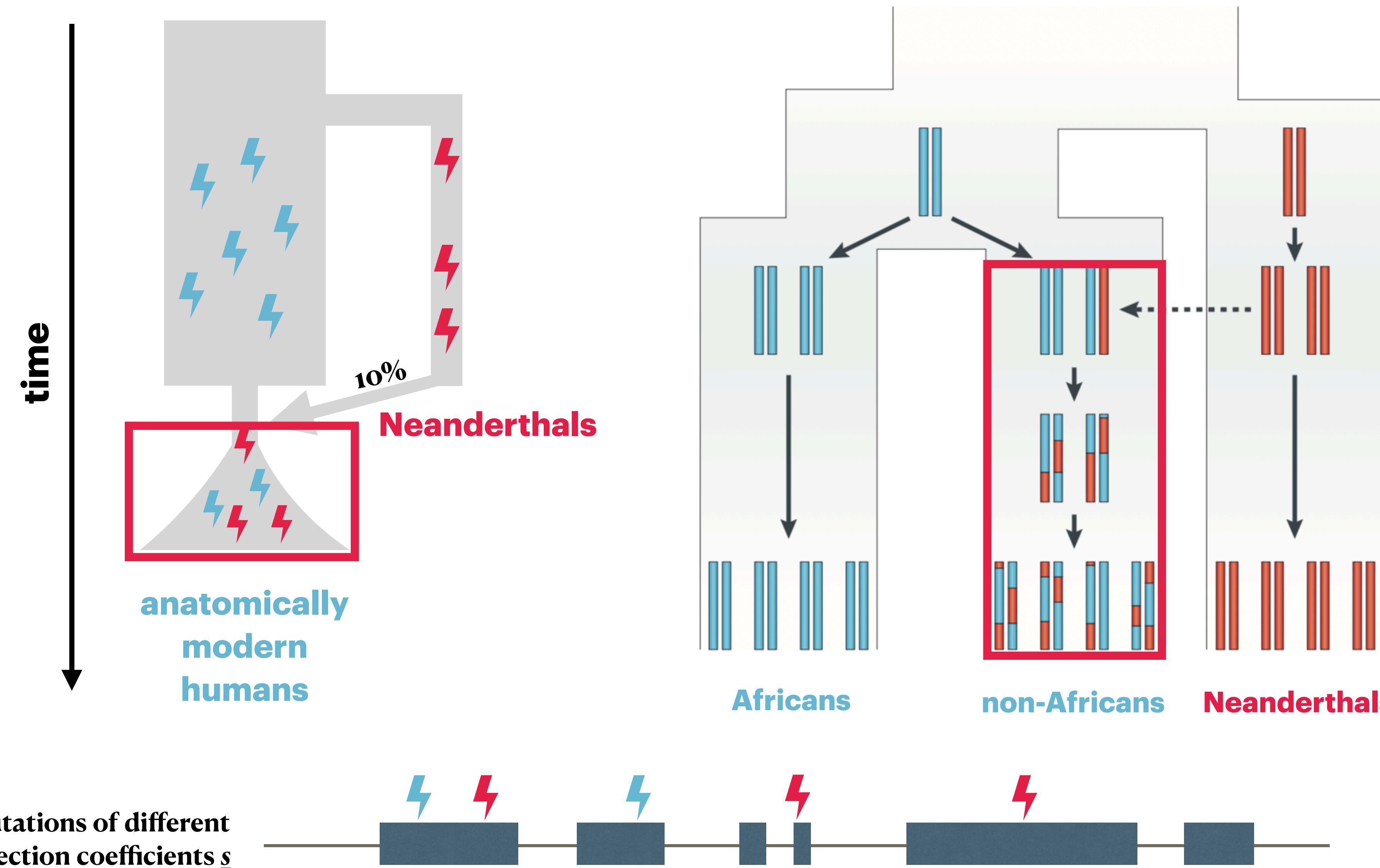


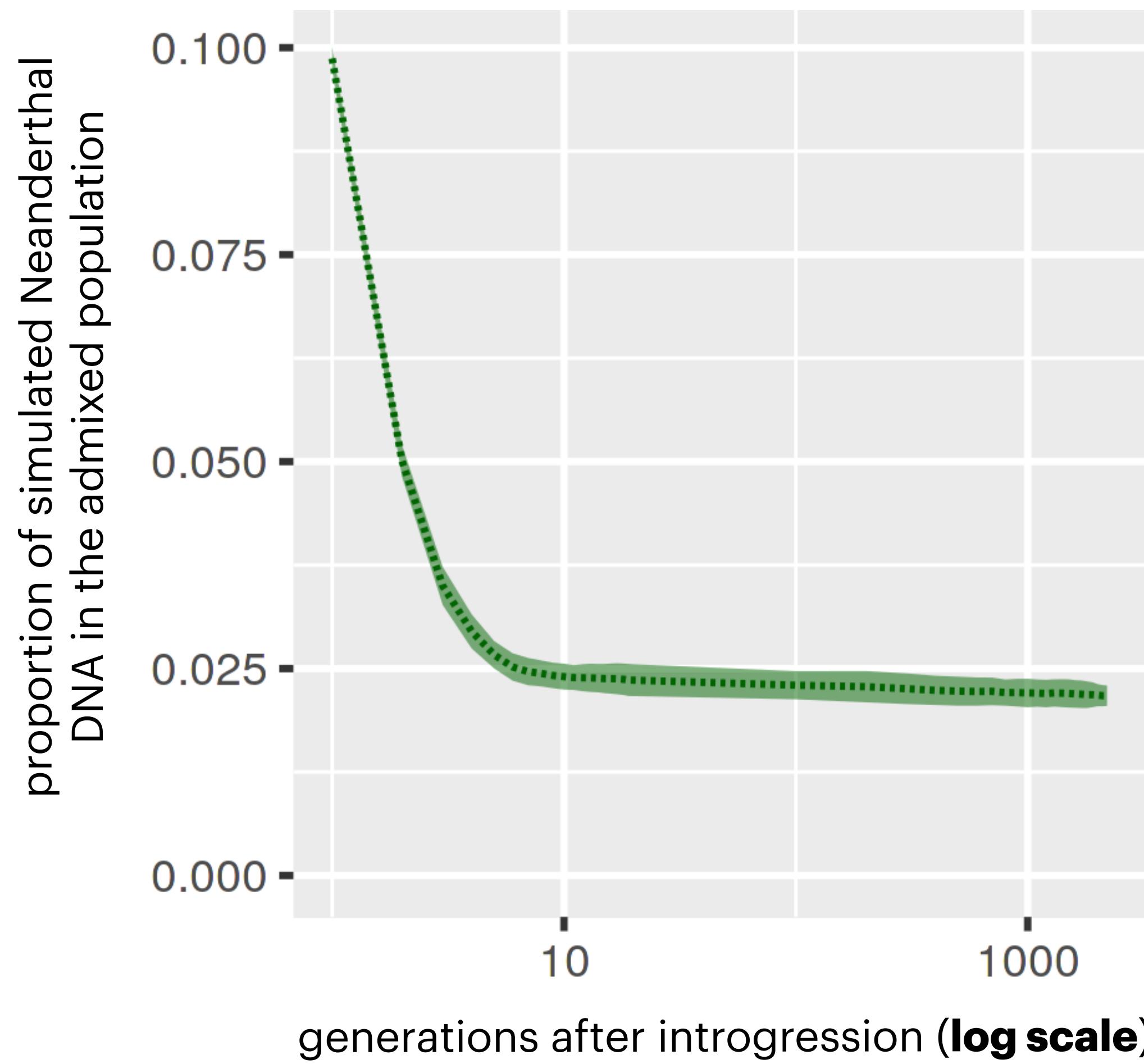
Ohta (Nature, 1973)

Simulations: tracing the trajectory of introgressed DNA over time

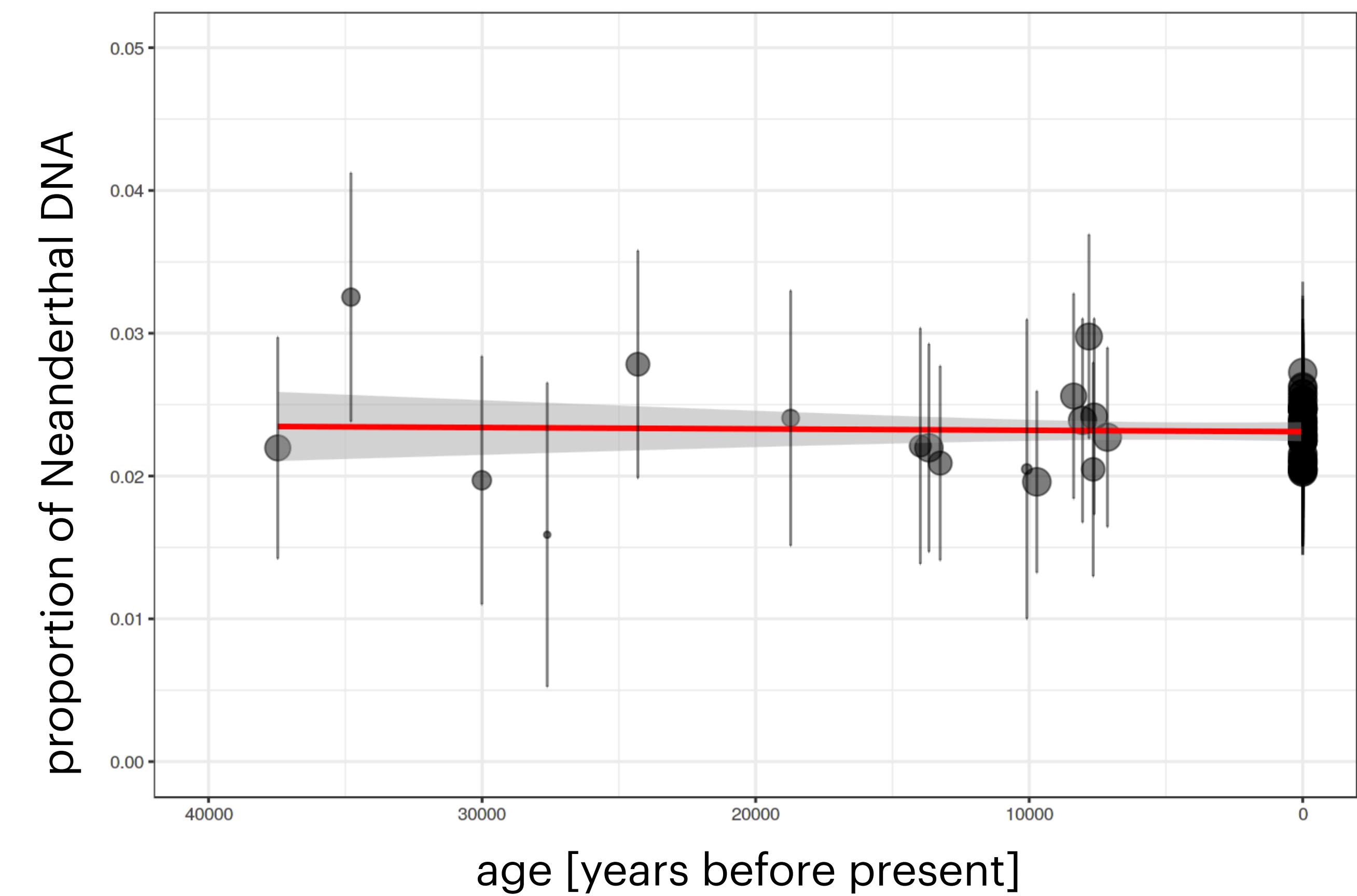
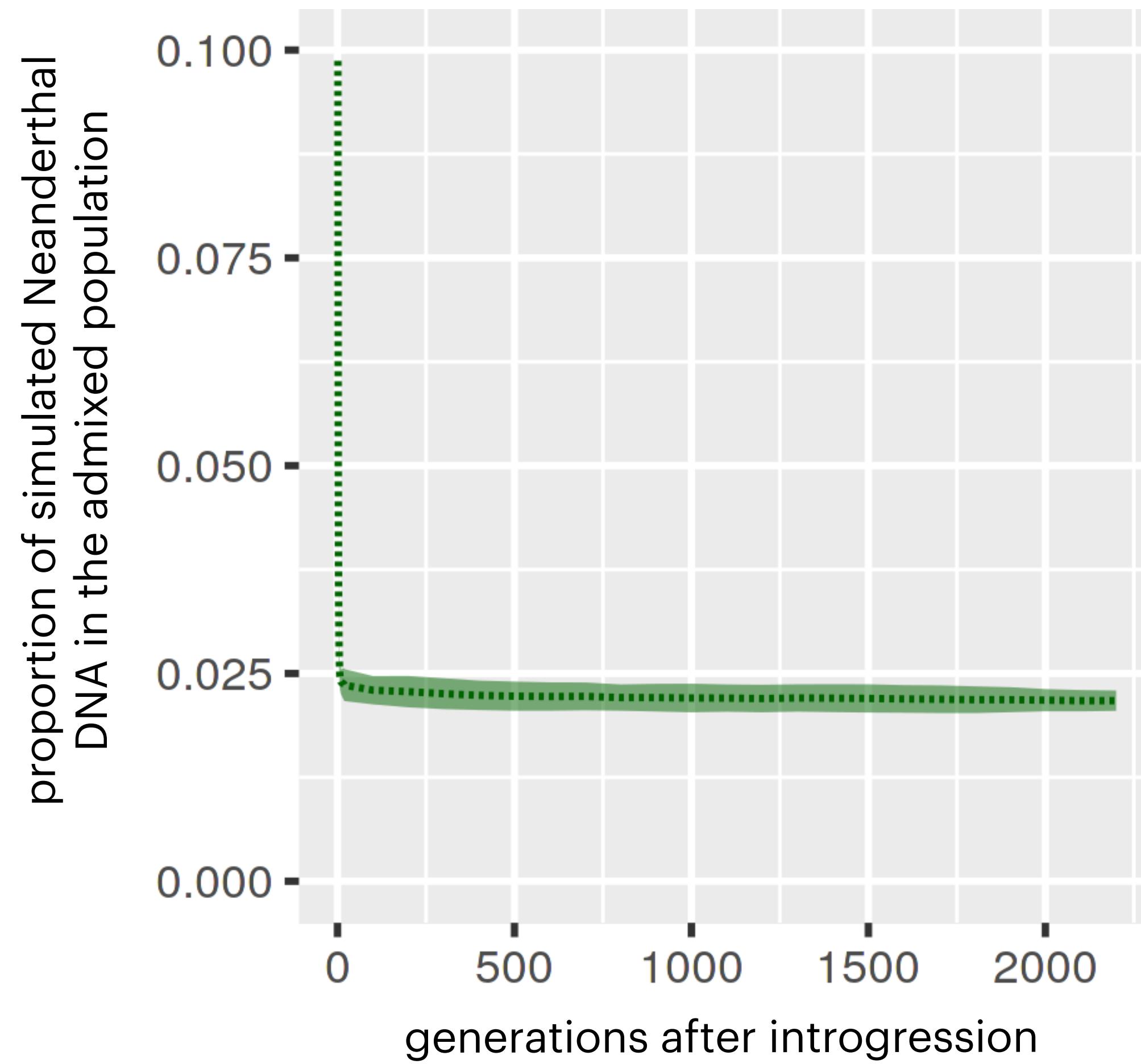


Simulations: tracing the trajectory of introgressed DNA over time



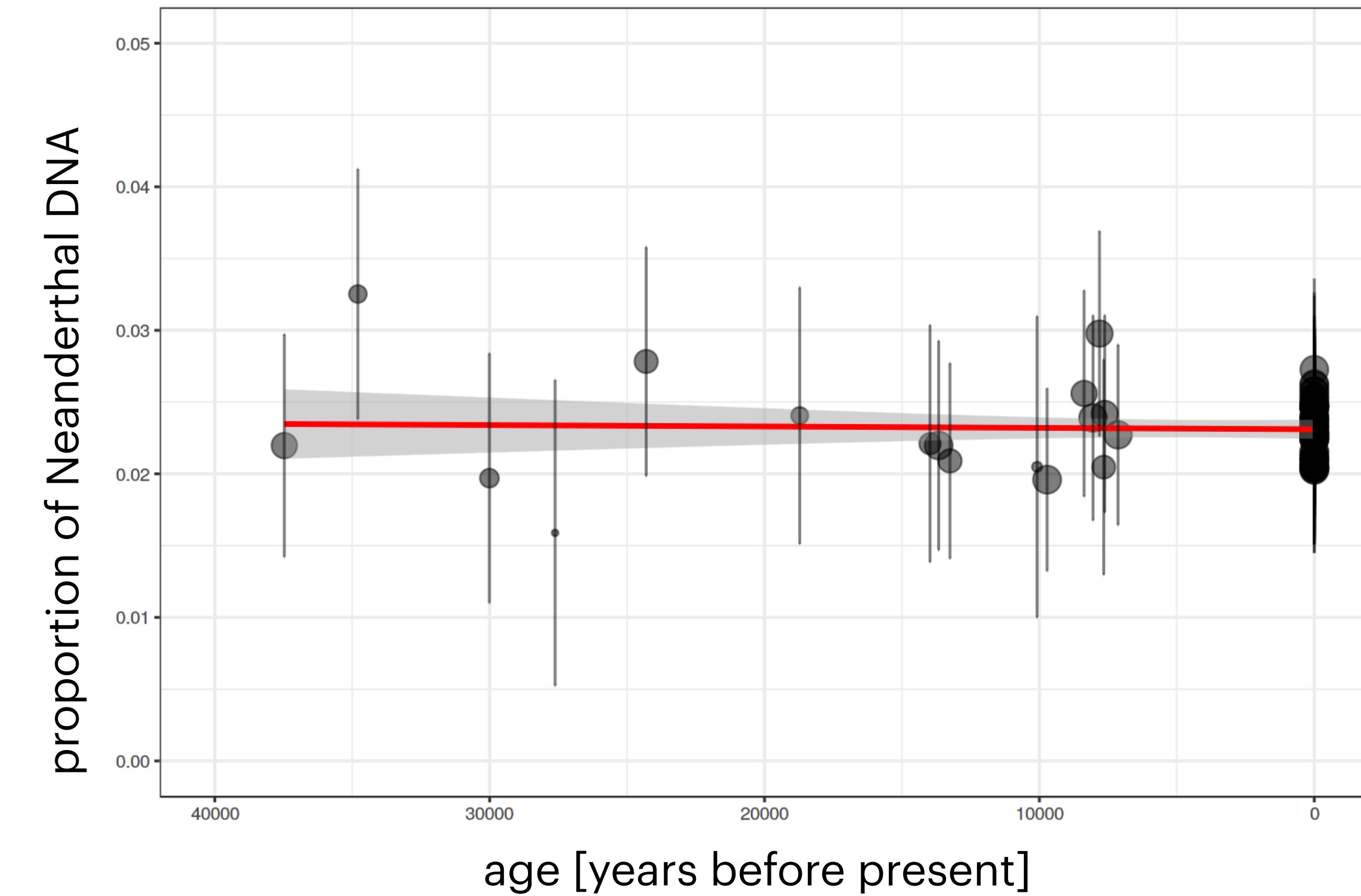
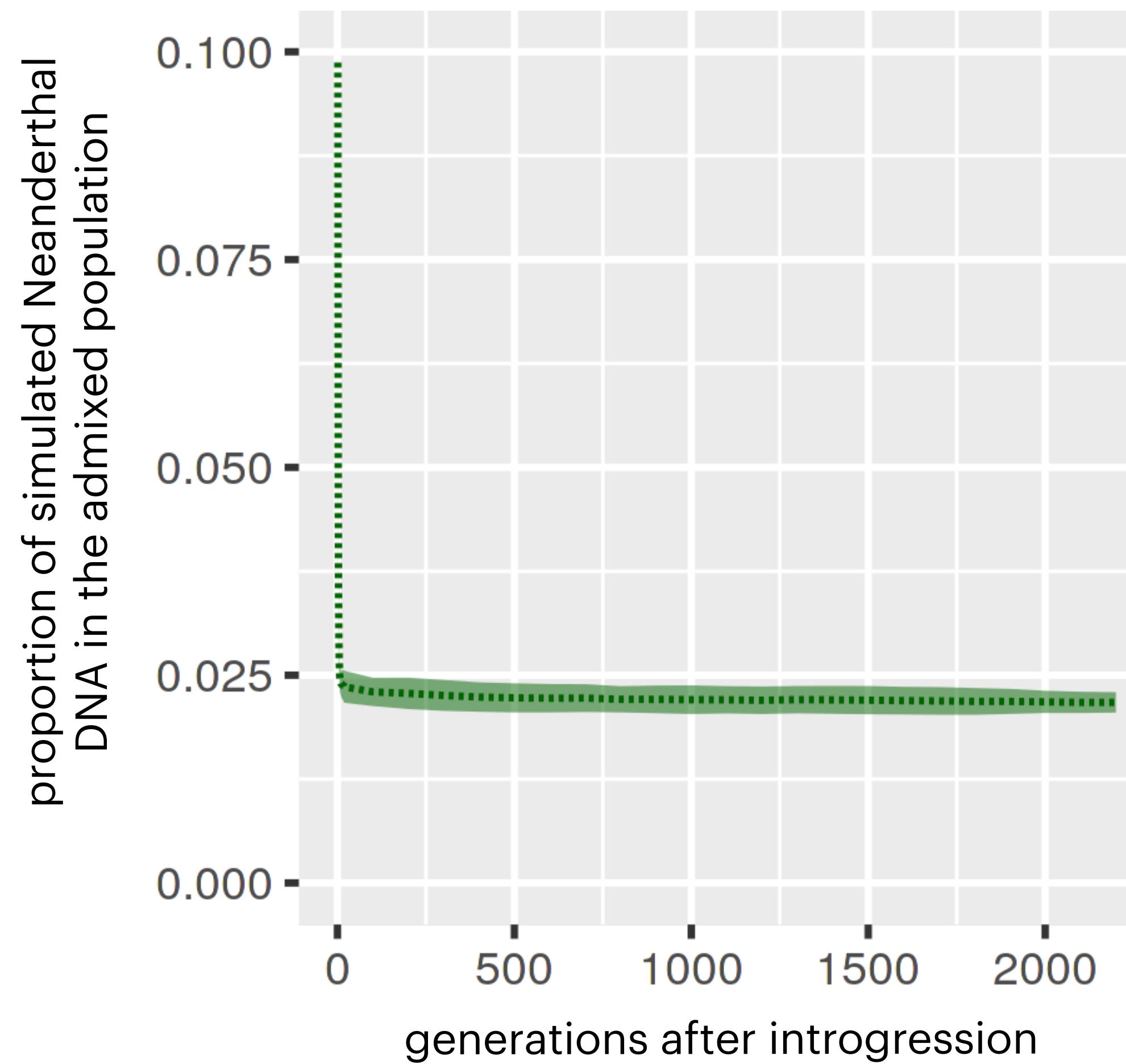


Harris et al. (2016); Juric et al. (2016); Petr et al. (2019)



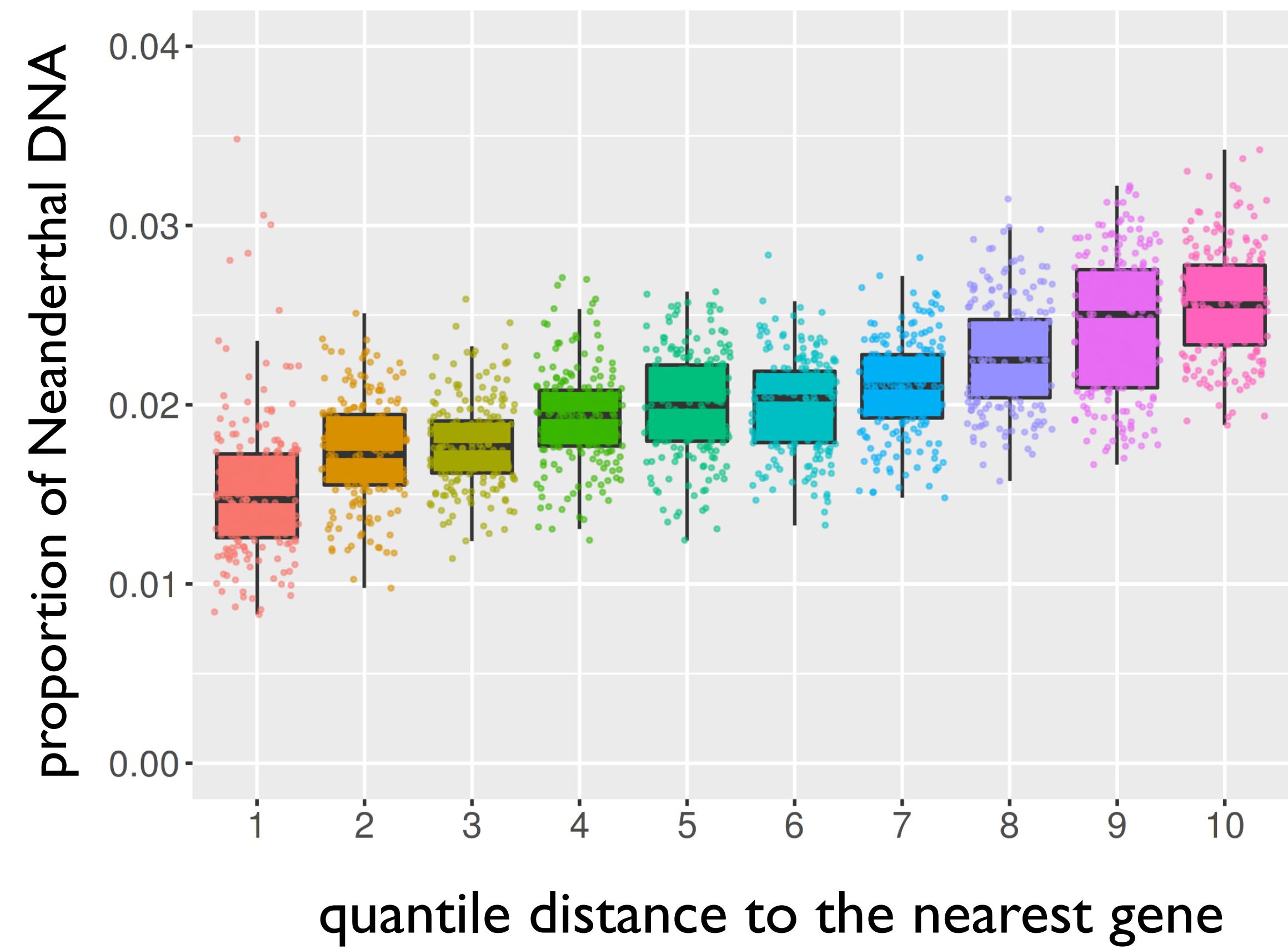
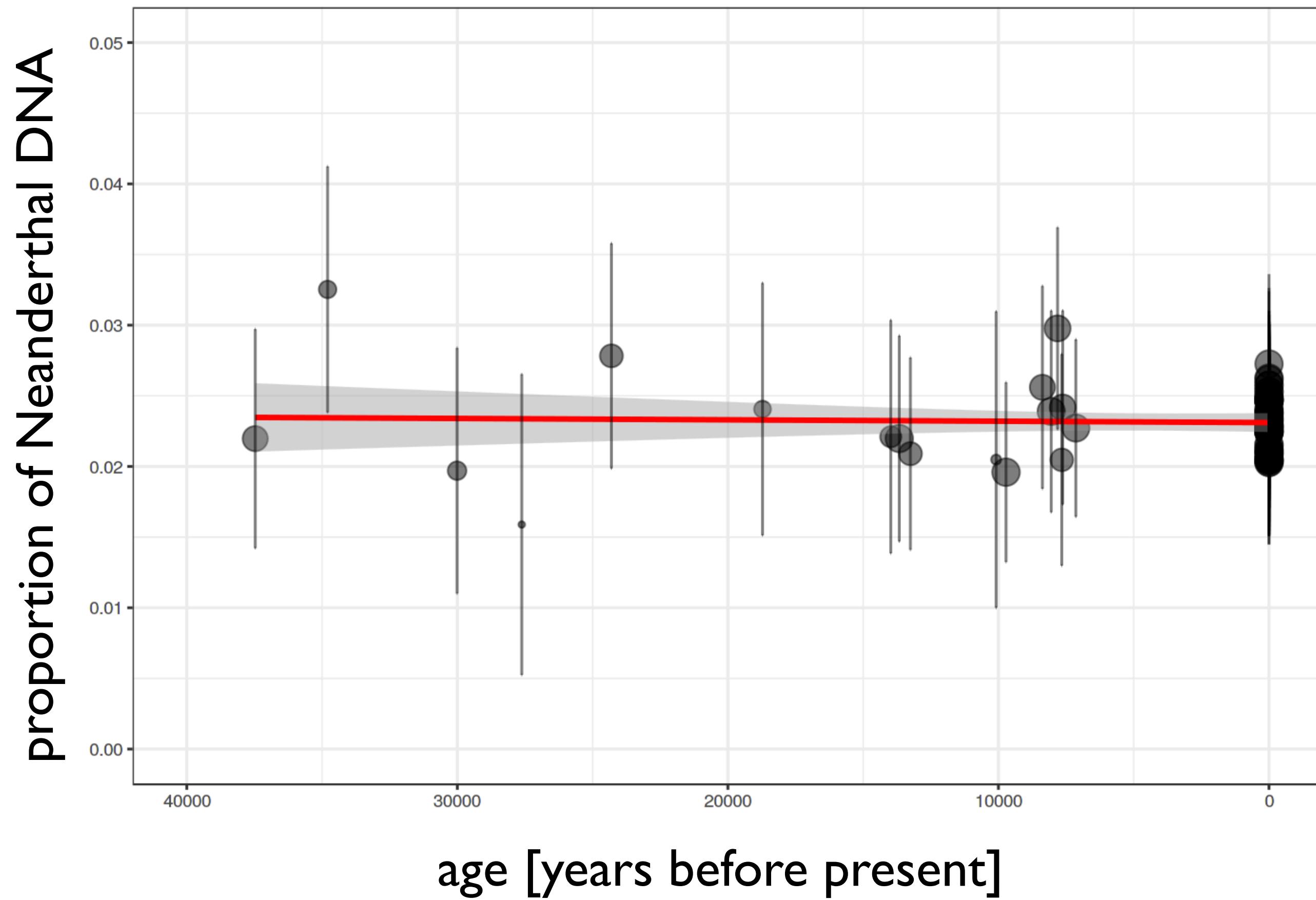
Harris et al. (2016); Juric et al. (2016); Petr et al. (2019)

Neanderthal ancestry trajectory matches simulations...

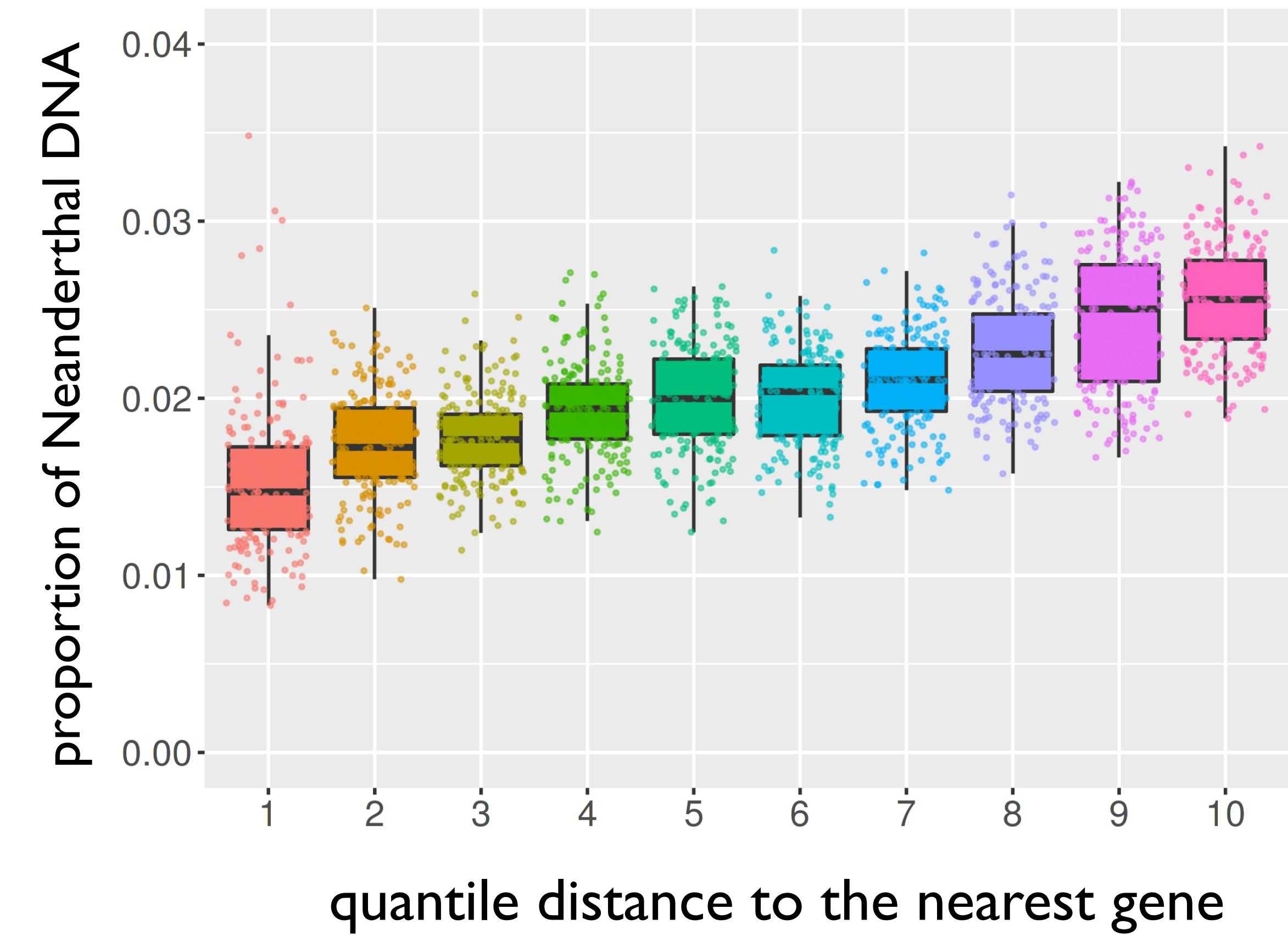
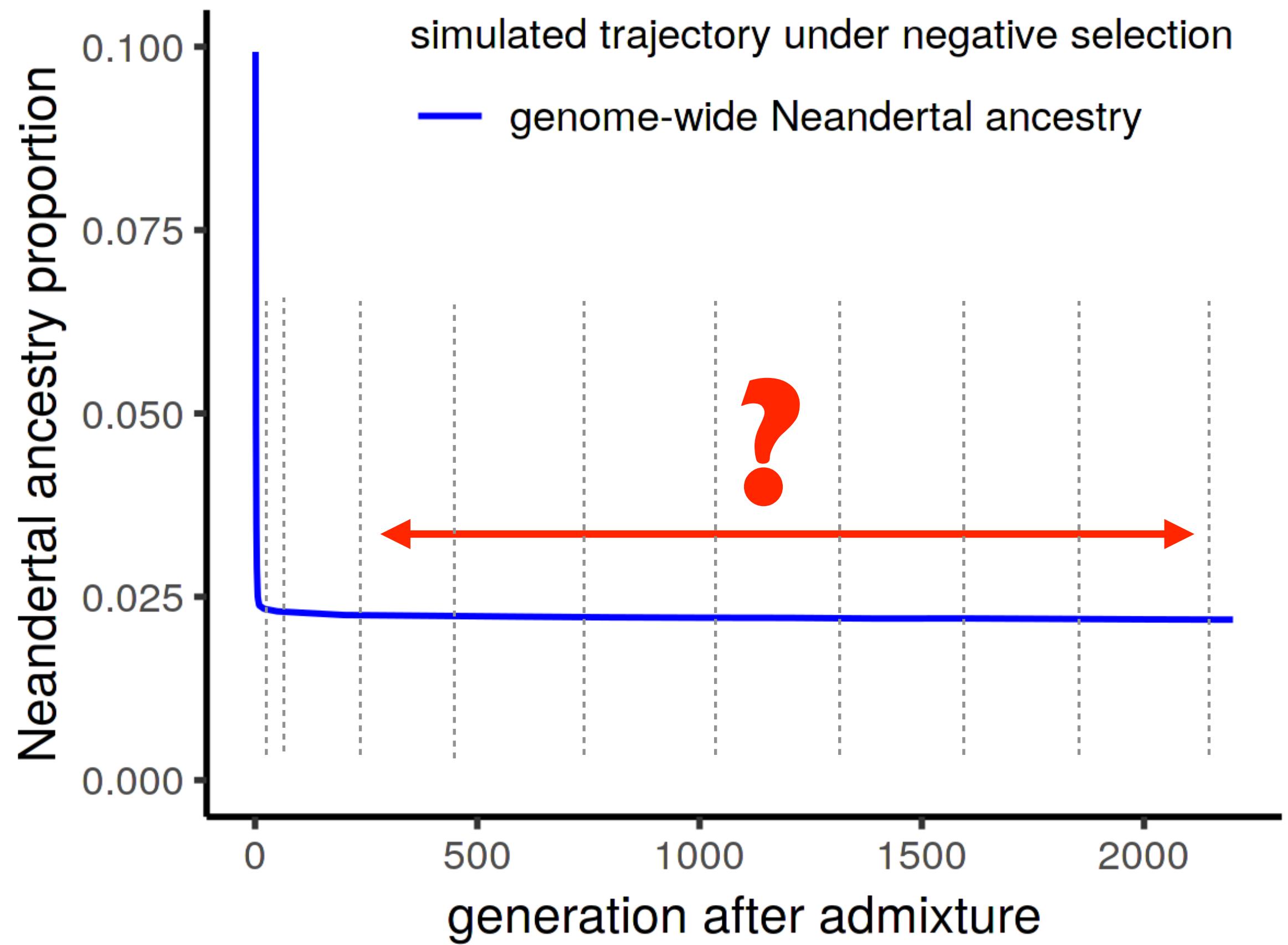


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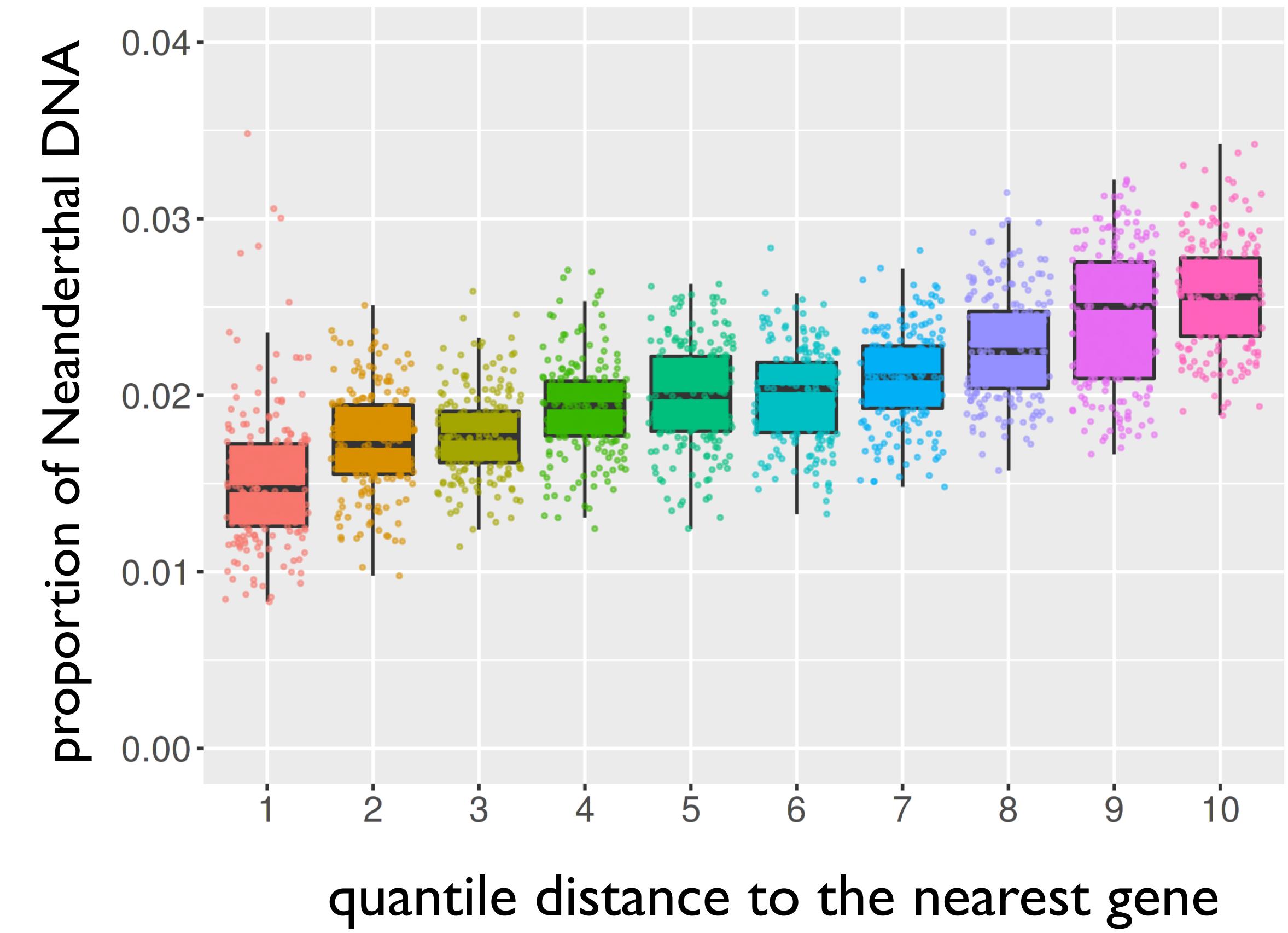
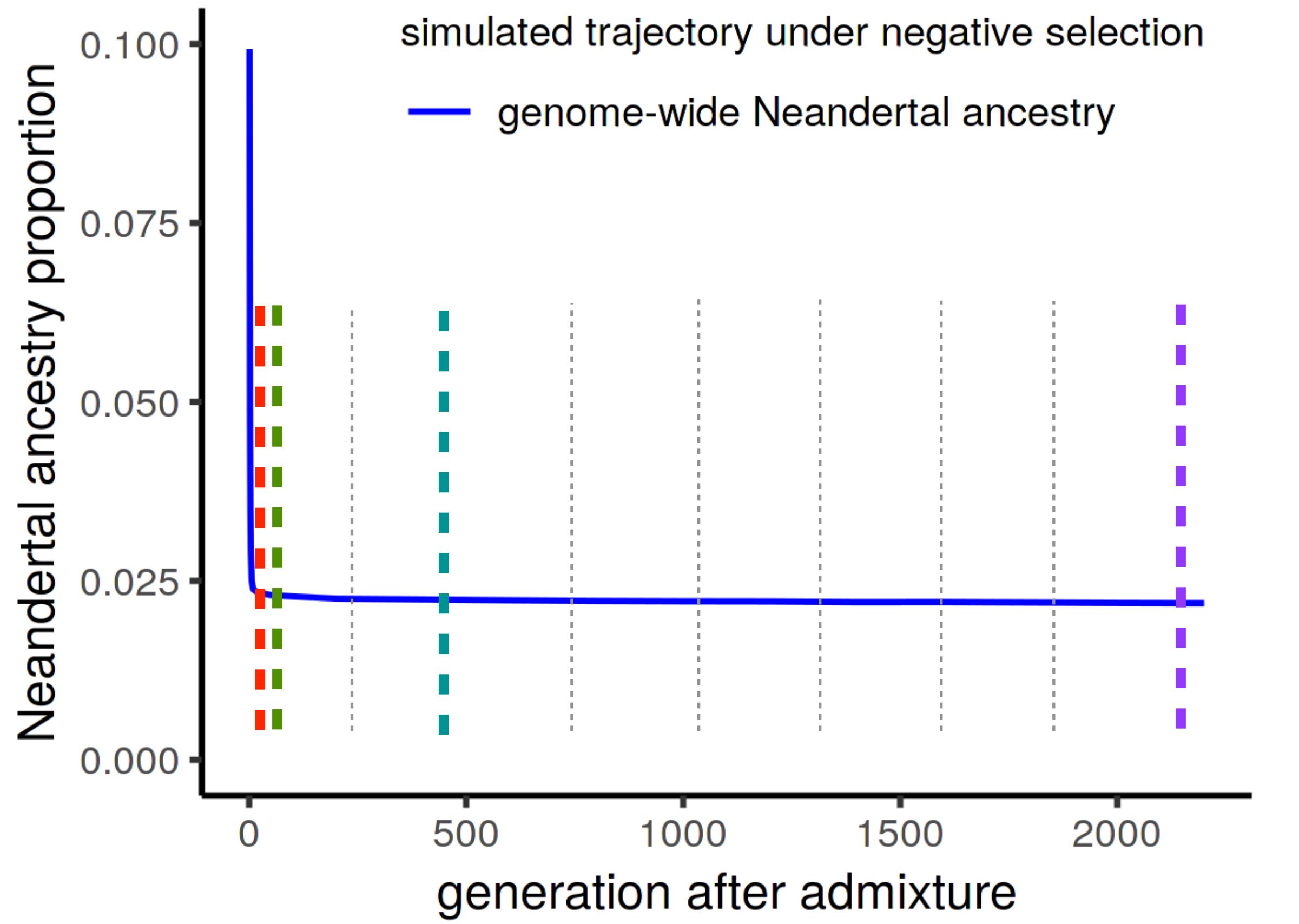
... what about the whole-genome negative selection signal?



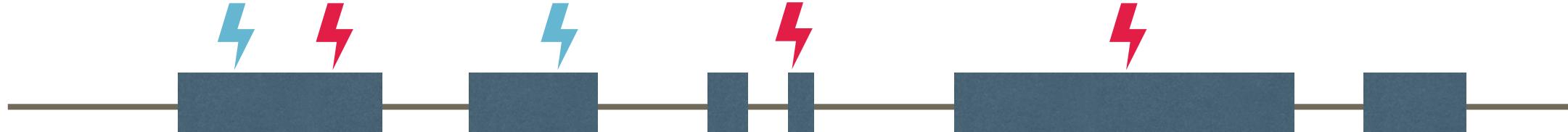
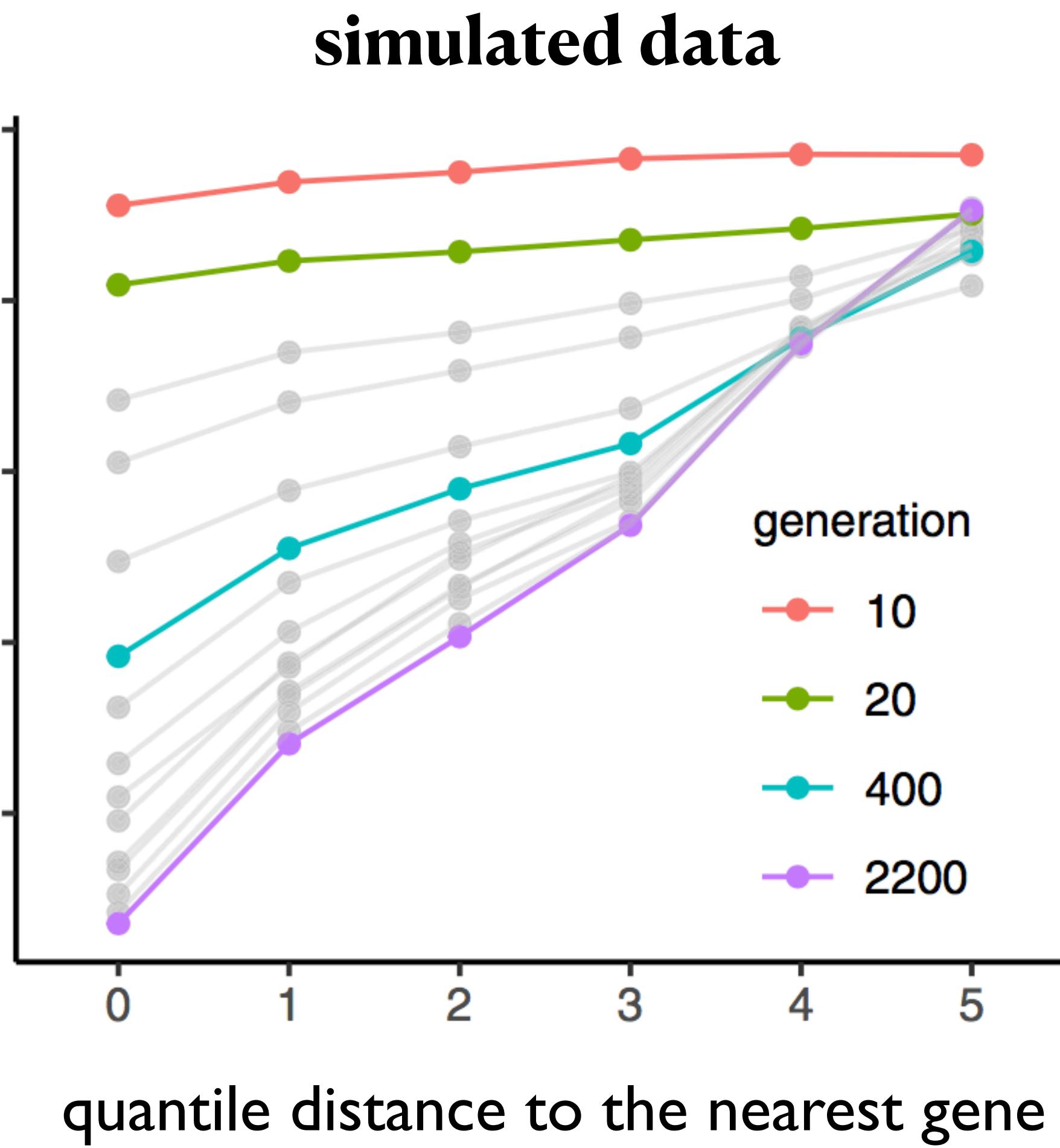
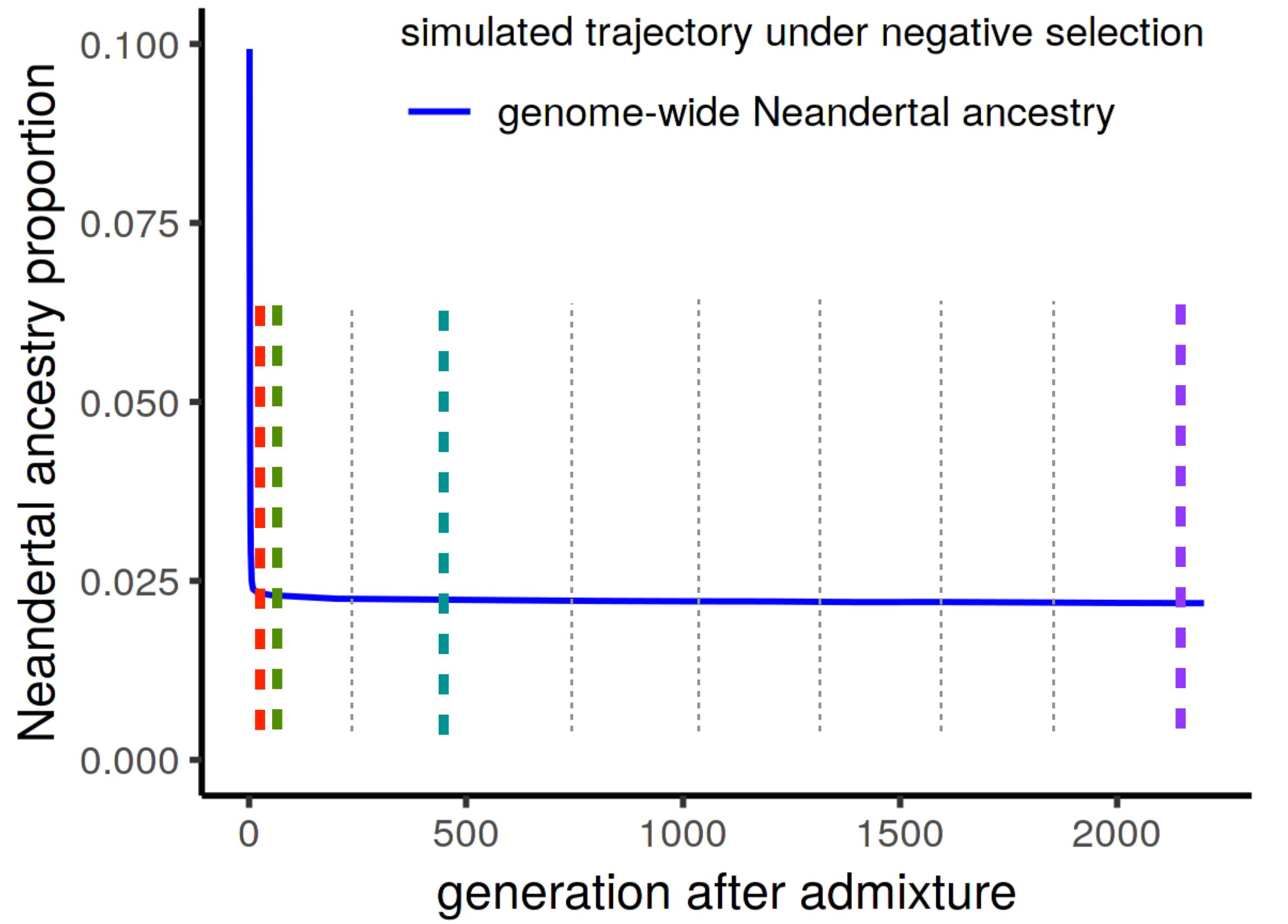
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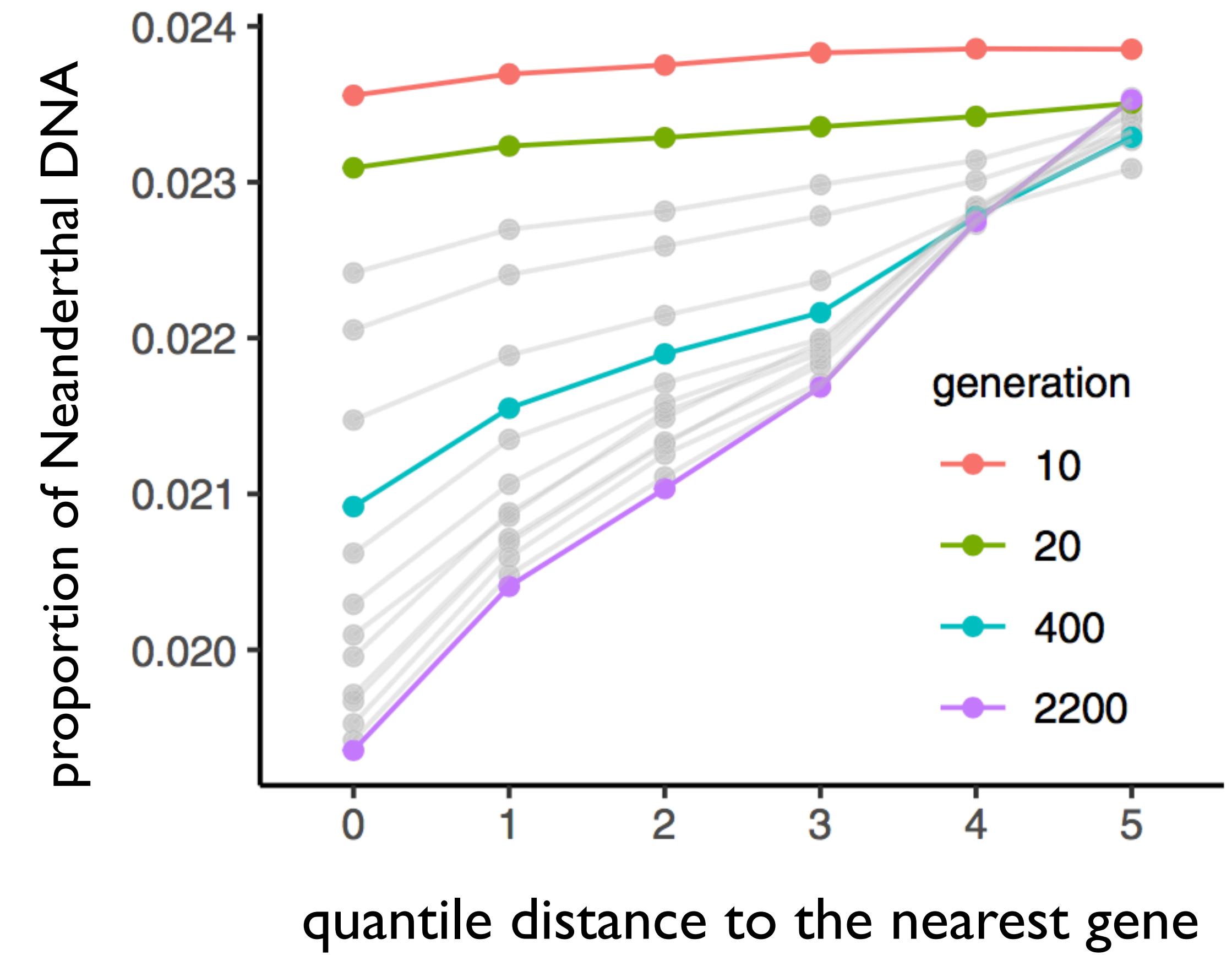
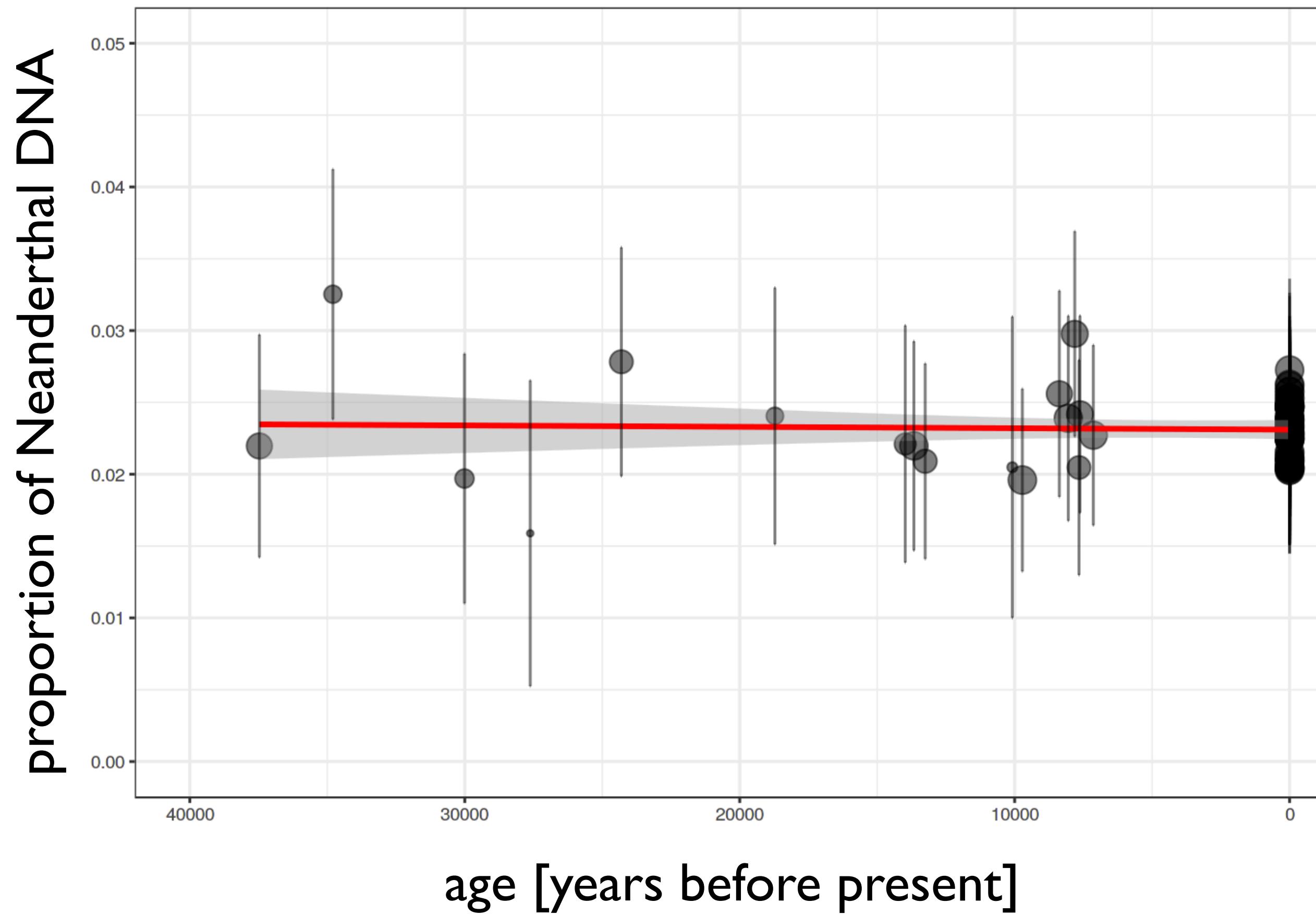
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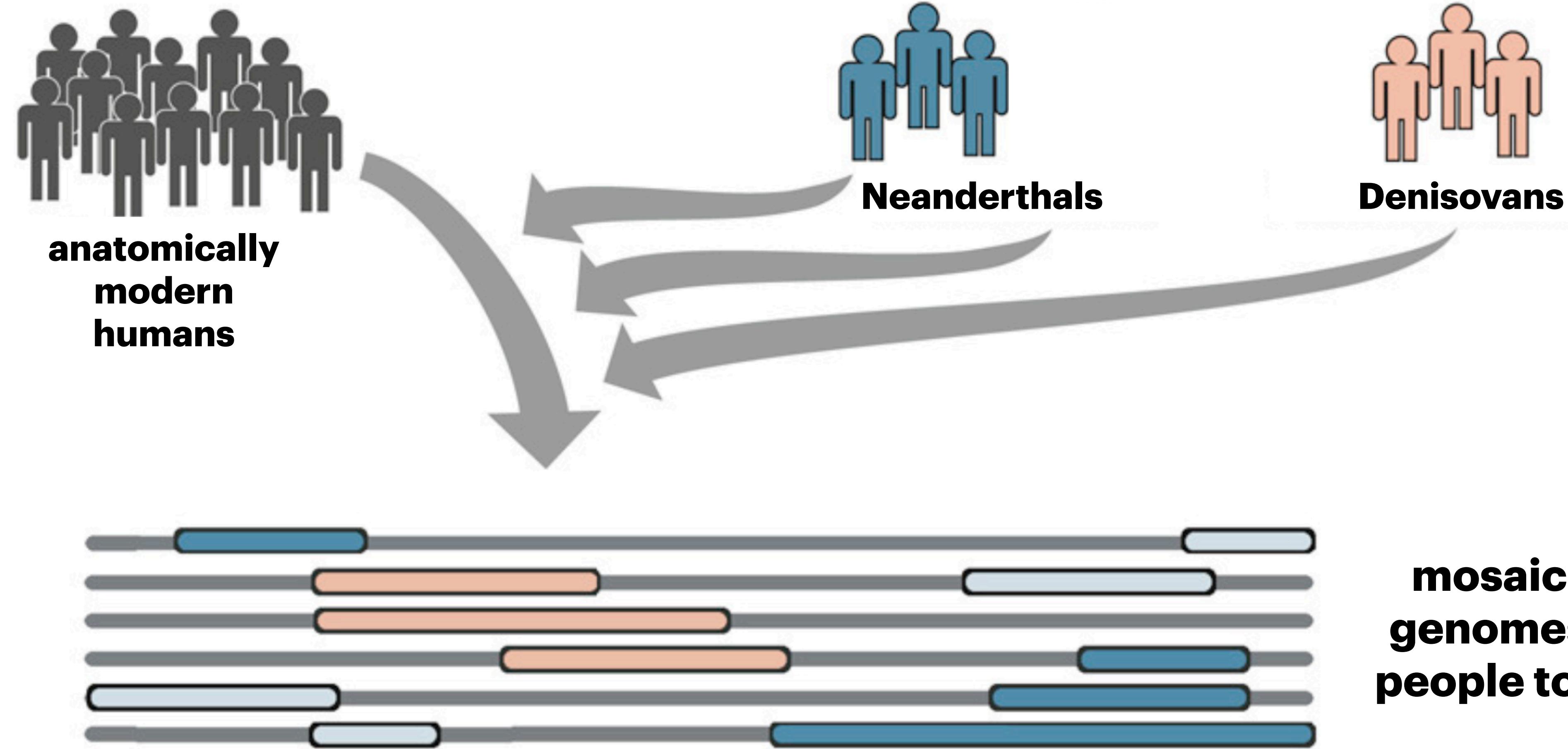
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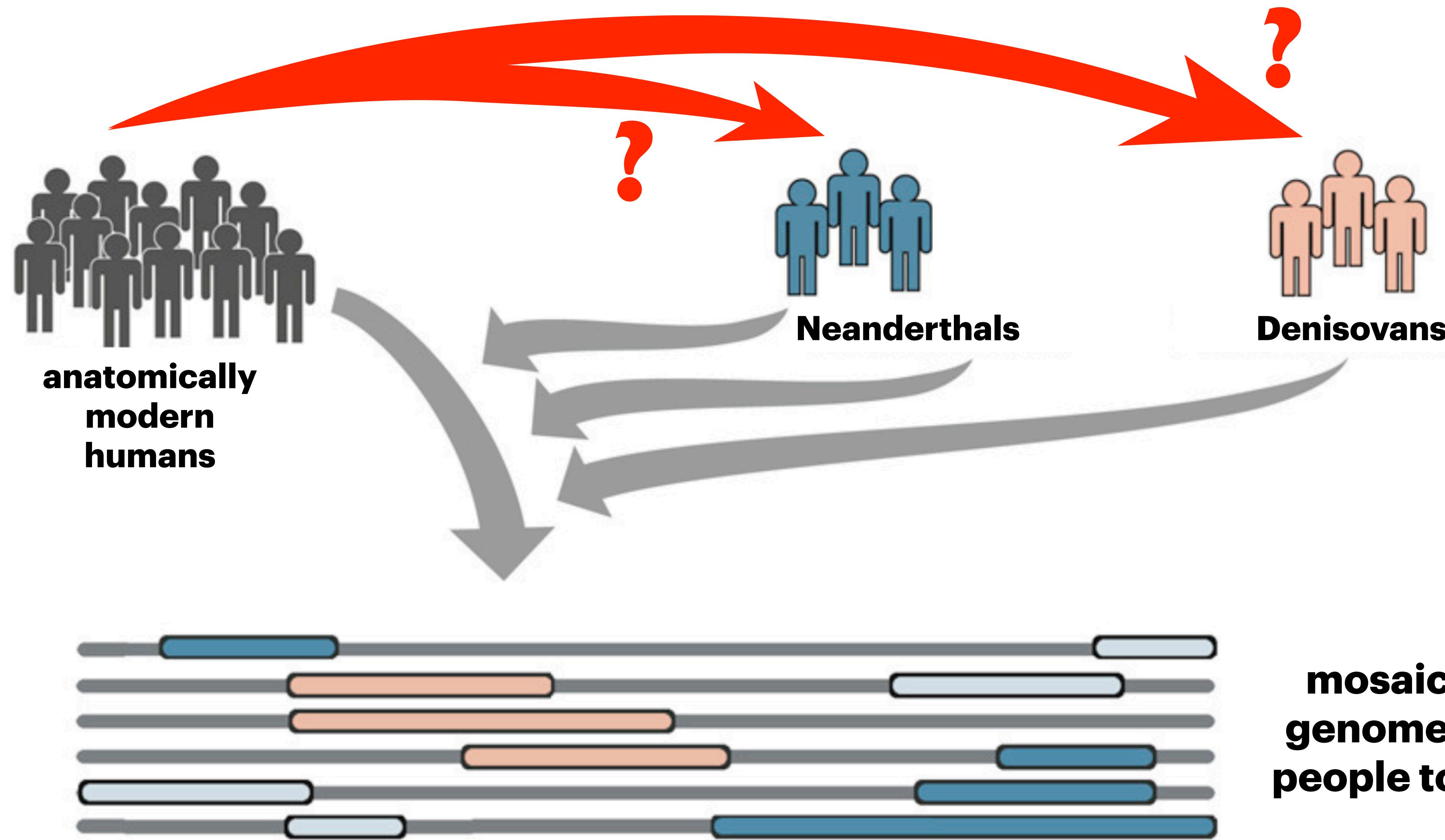
Both empirical results match population genetic theory!



This might have had an effect in
many other contexts too...

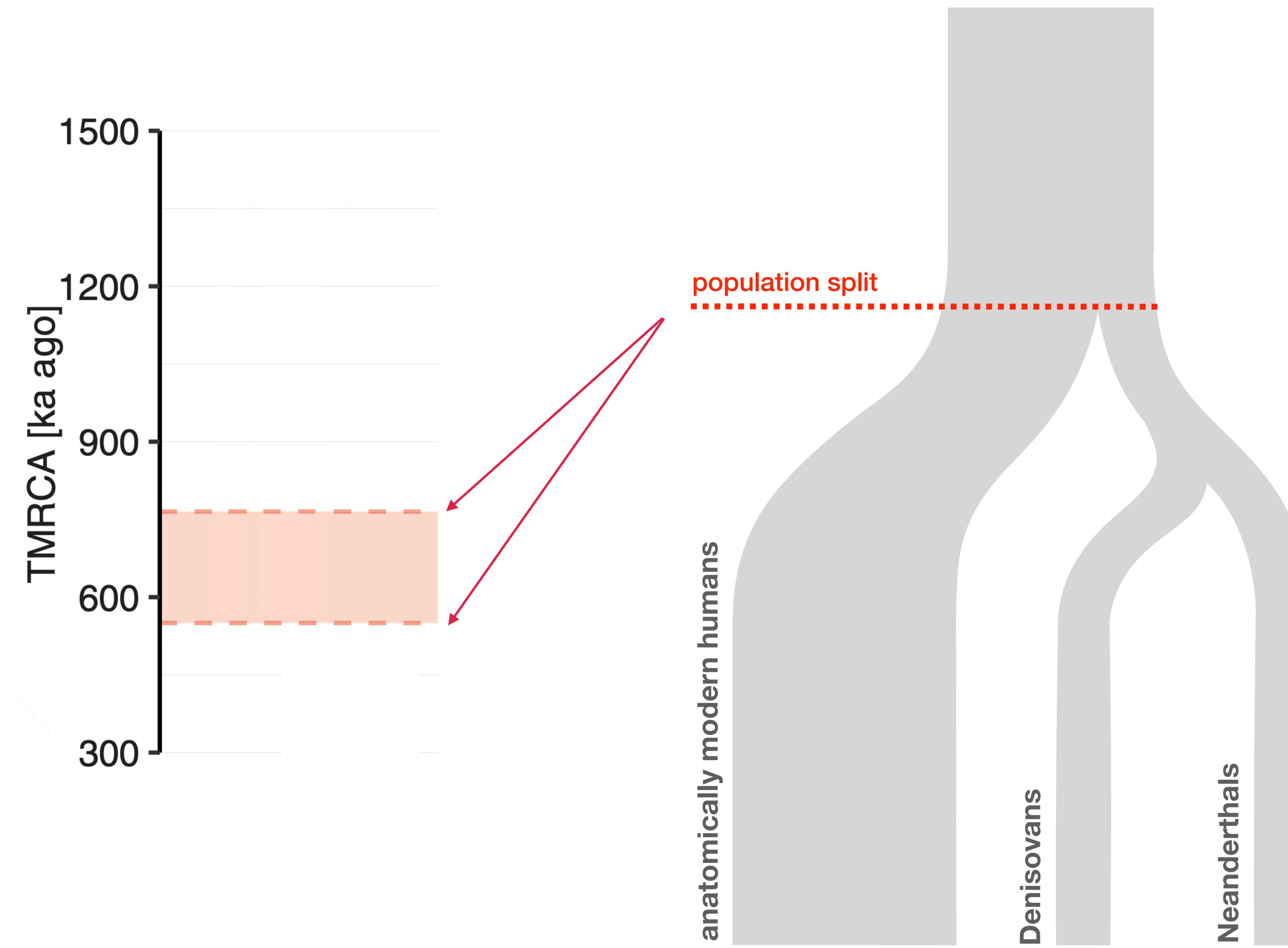


adapted from Browning et al., Cell, 2018

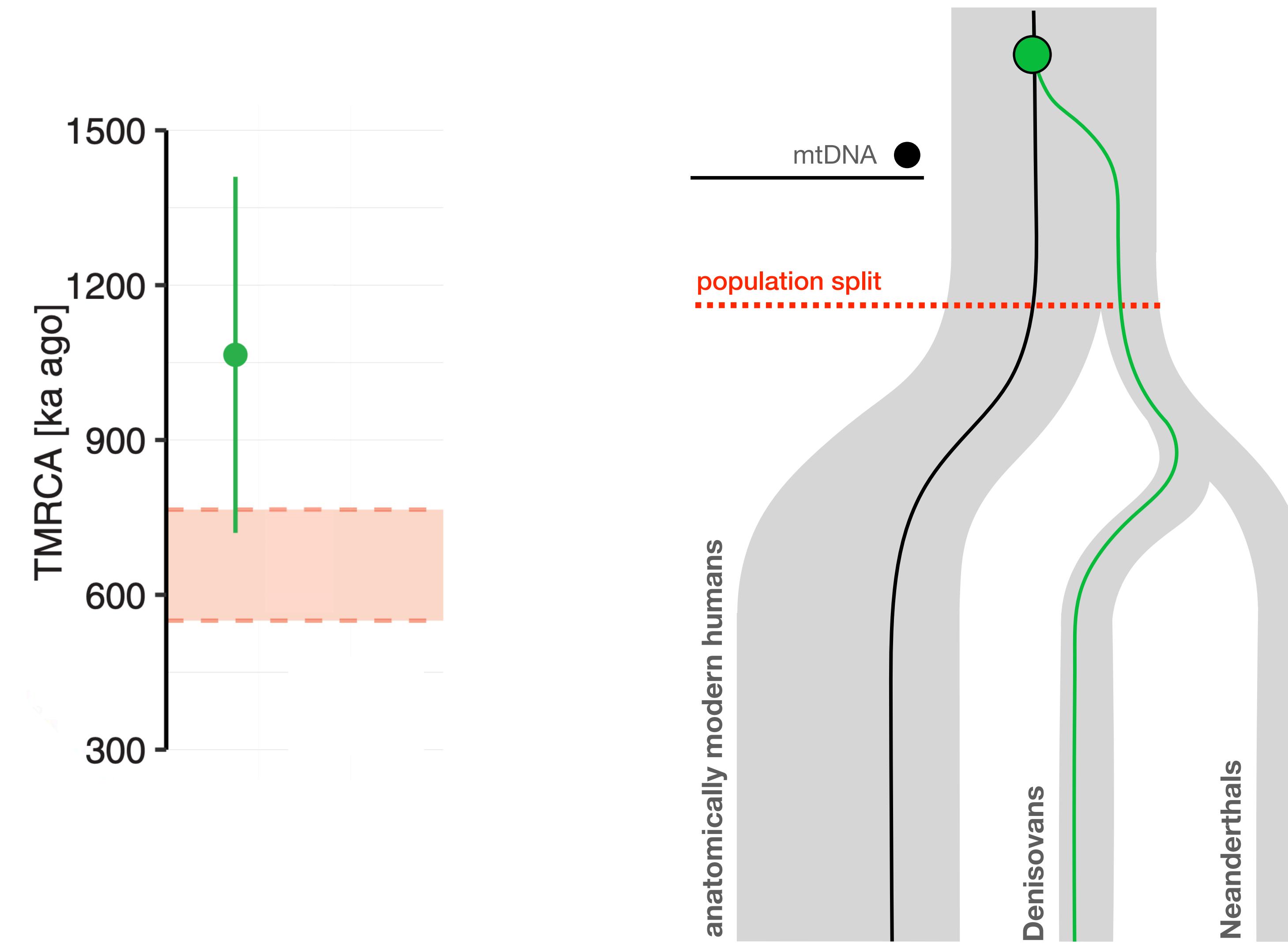


adapted from Browning et al., Cell, 2018

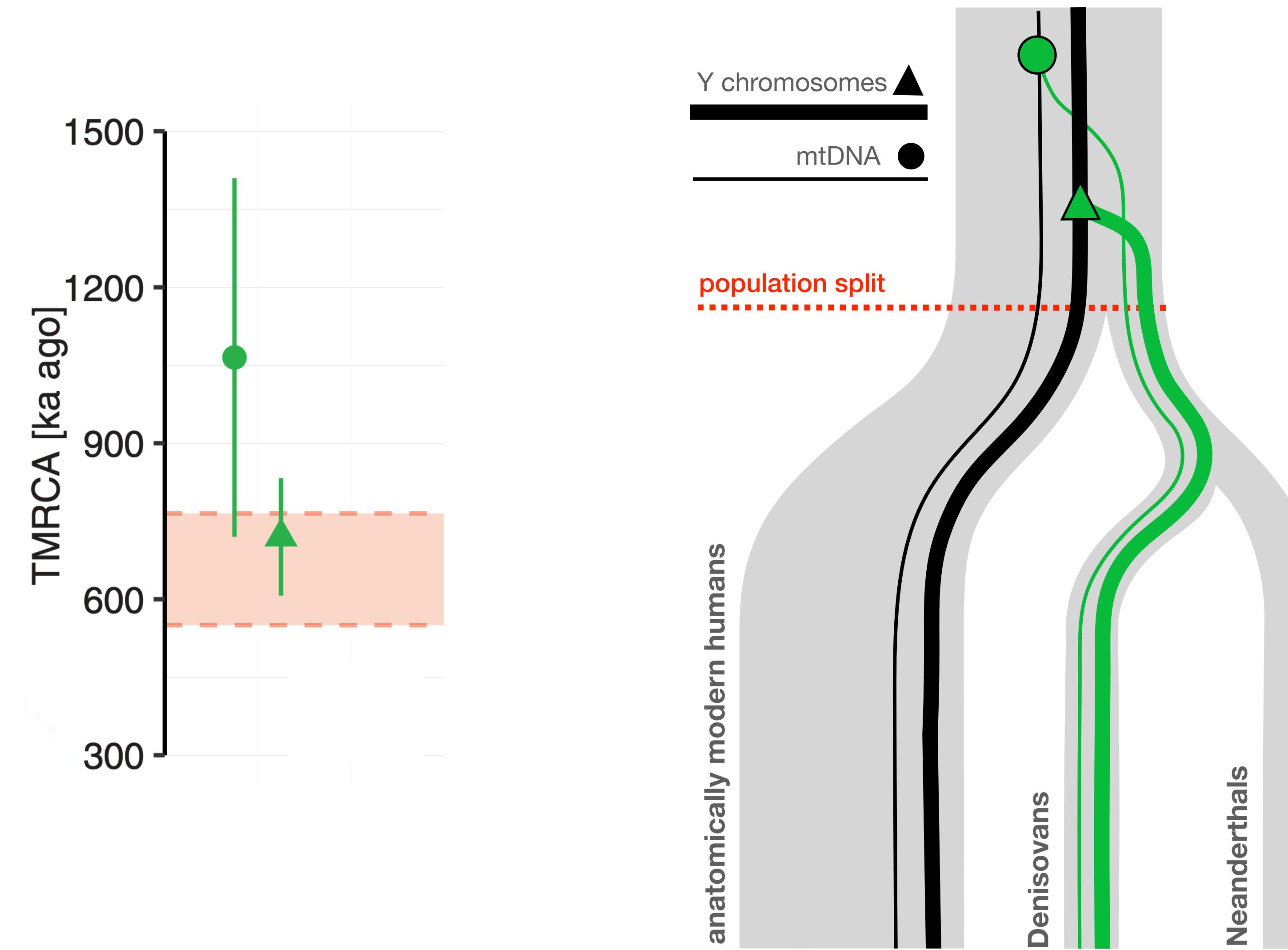
Population history on a nuclear DNA level



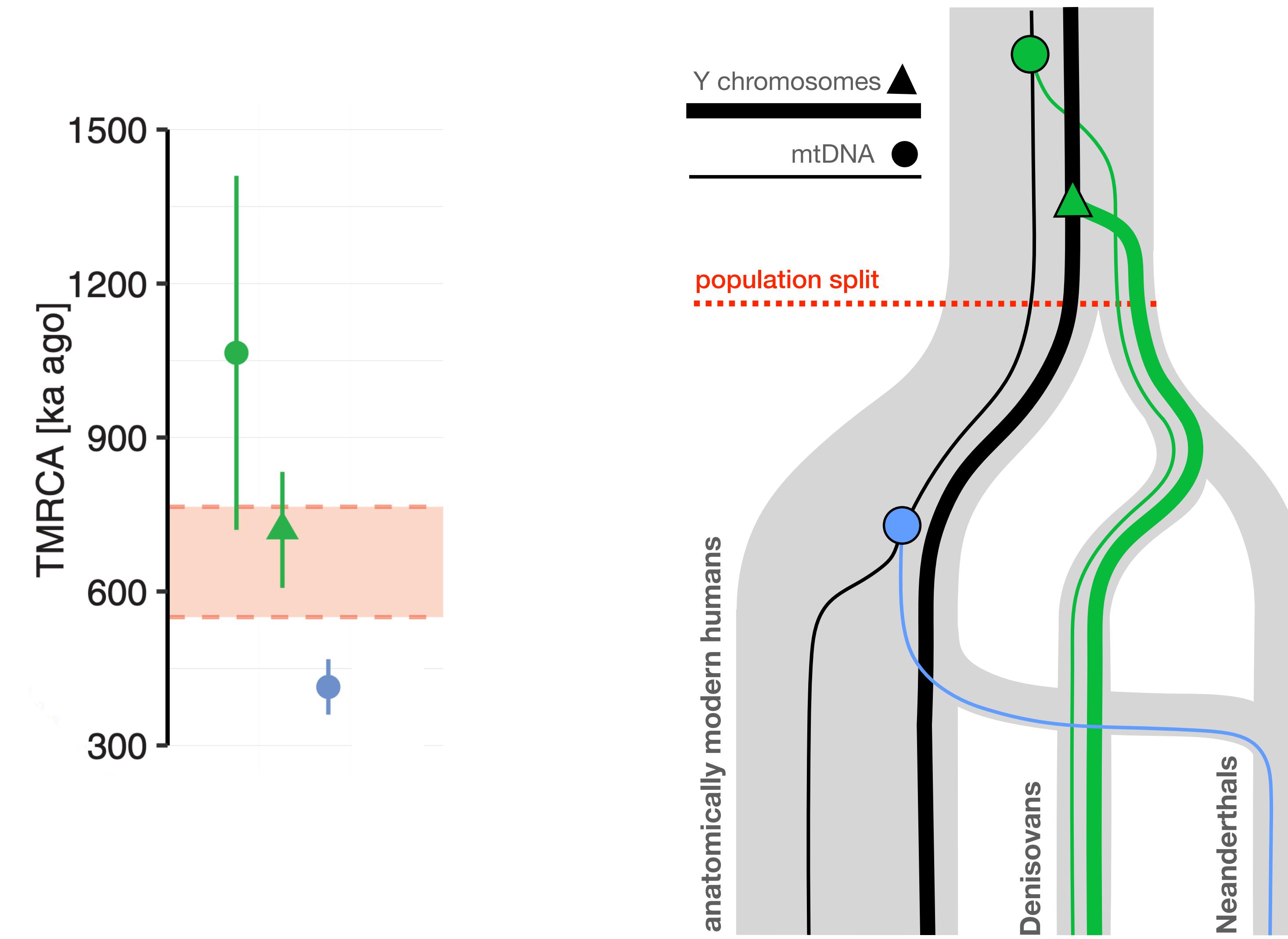
Denisovan mt and Y DNA follow known population history



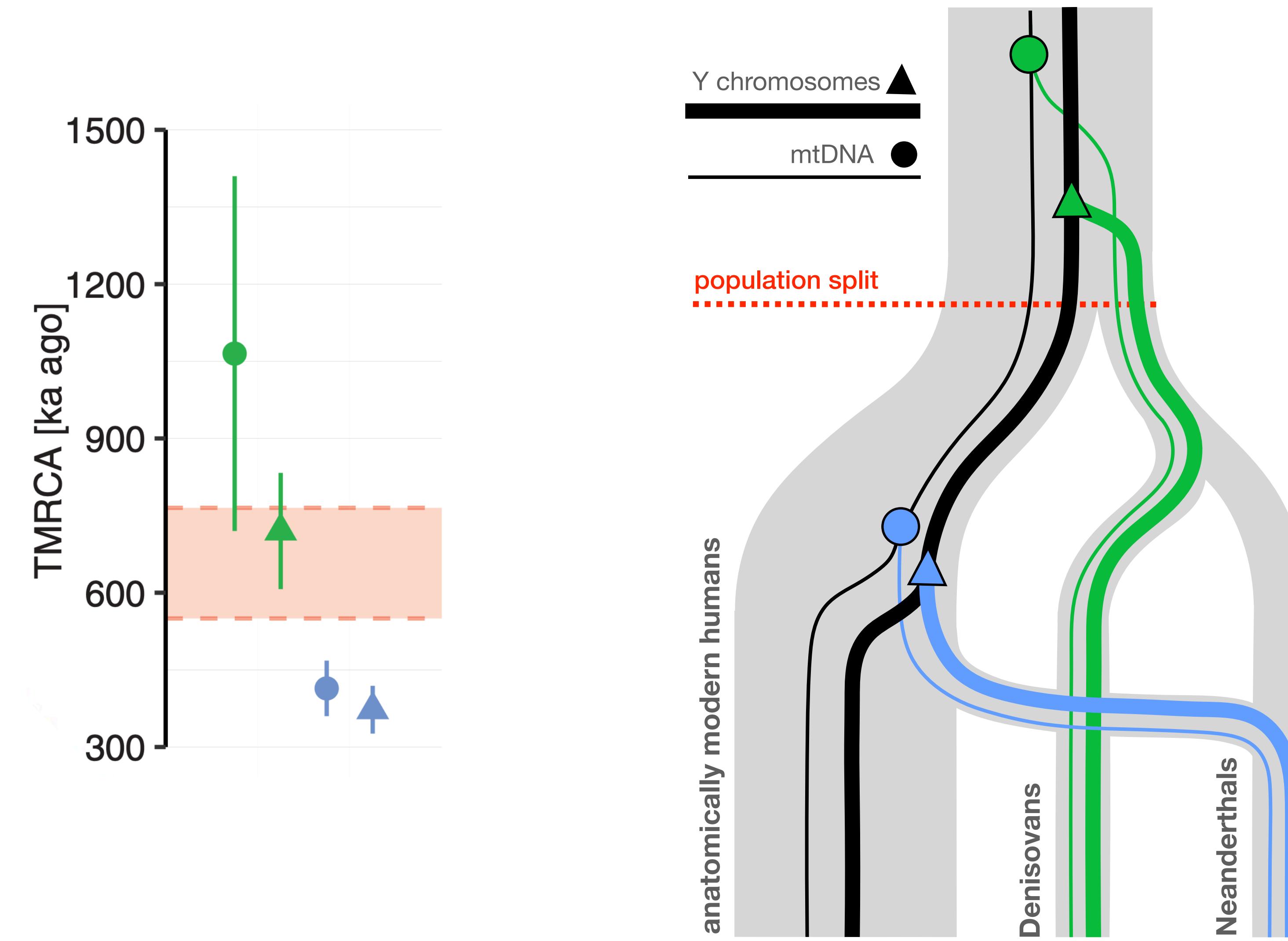
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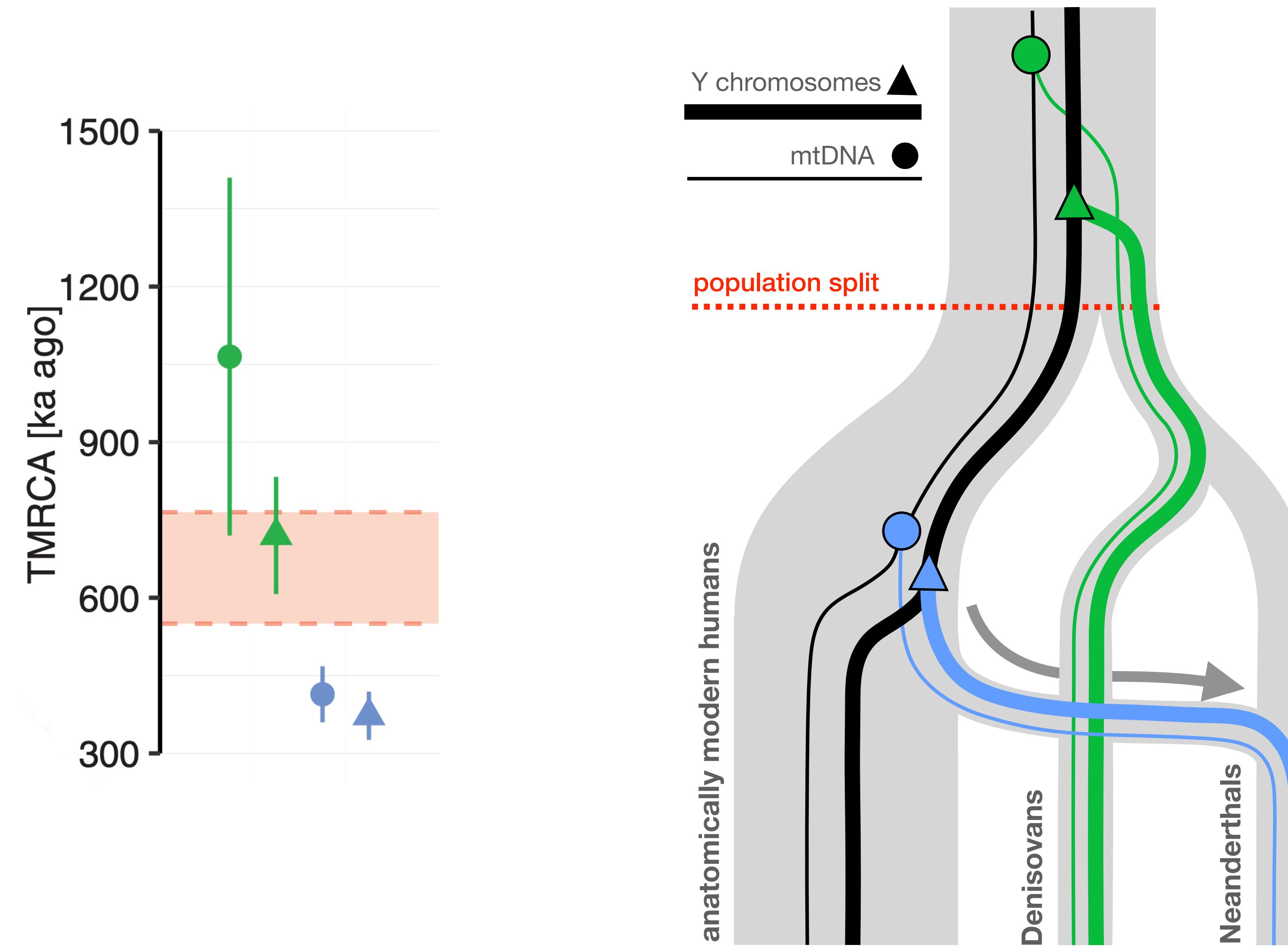
Neanderthal mitochondrial DNAs contradict this!



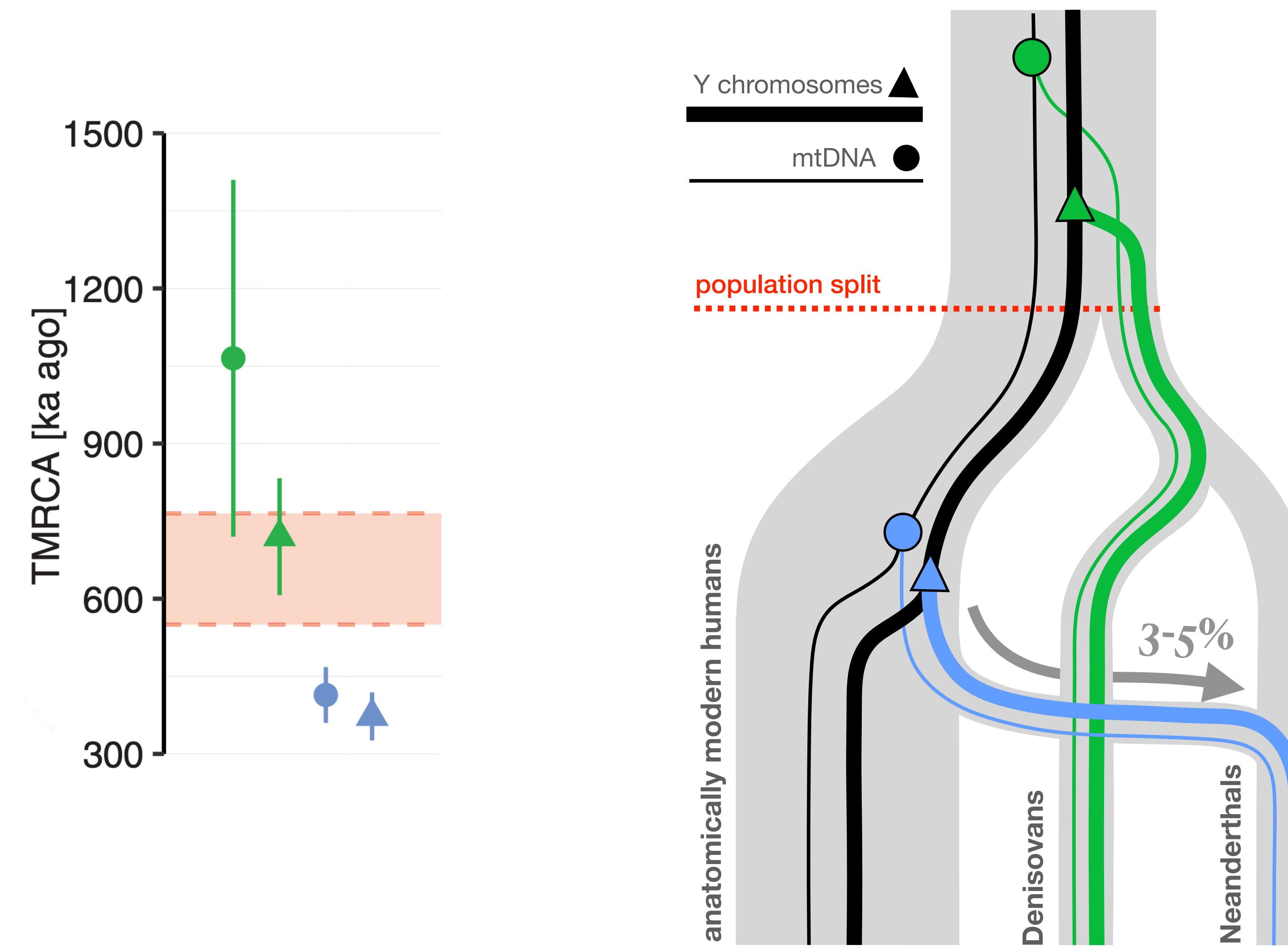
Neanderthal Y chromosomes also contradict this!



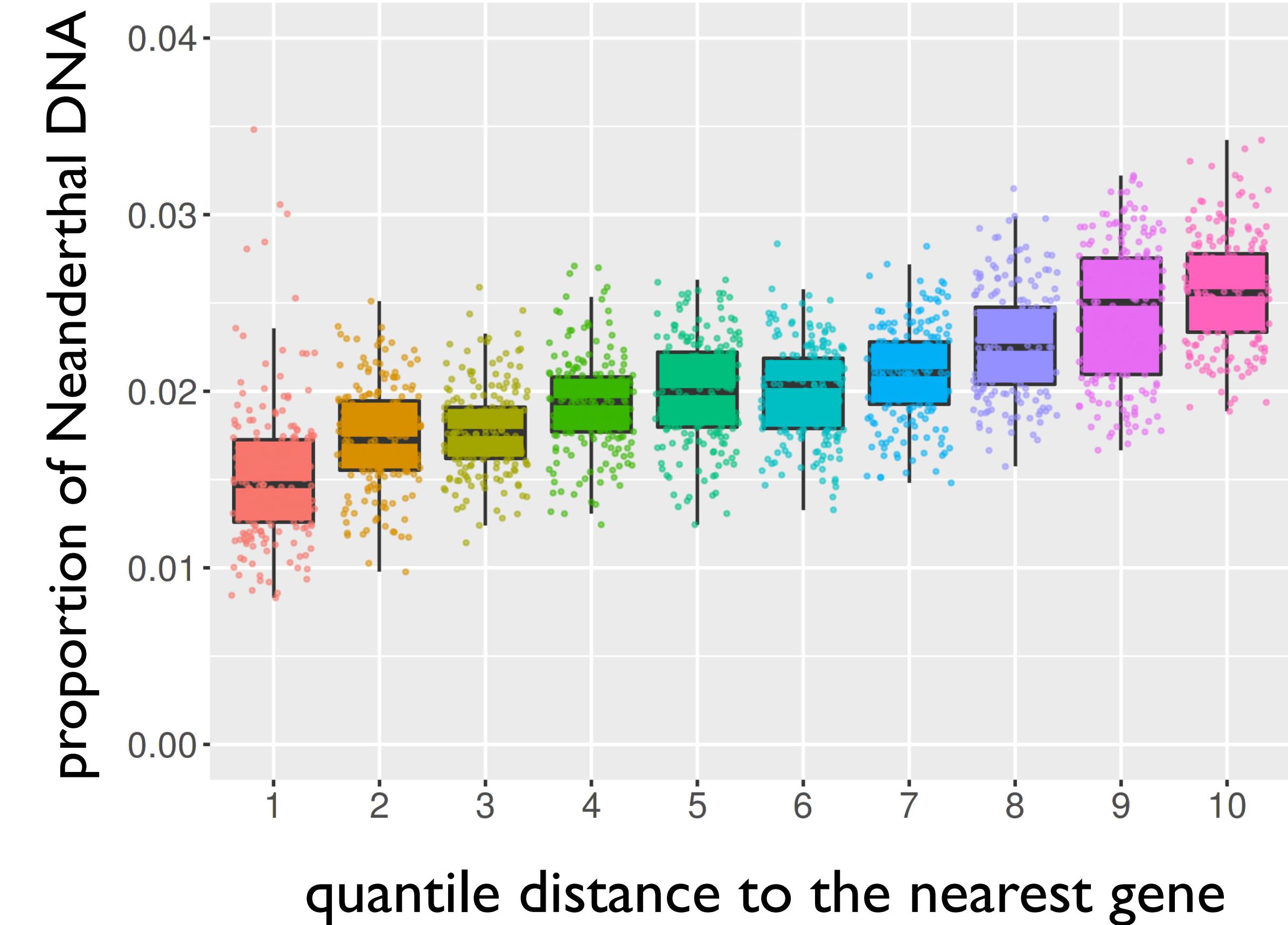
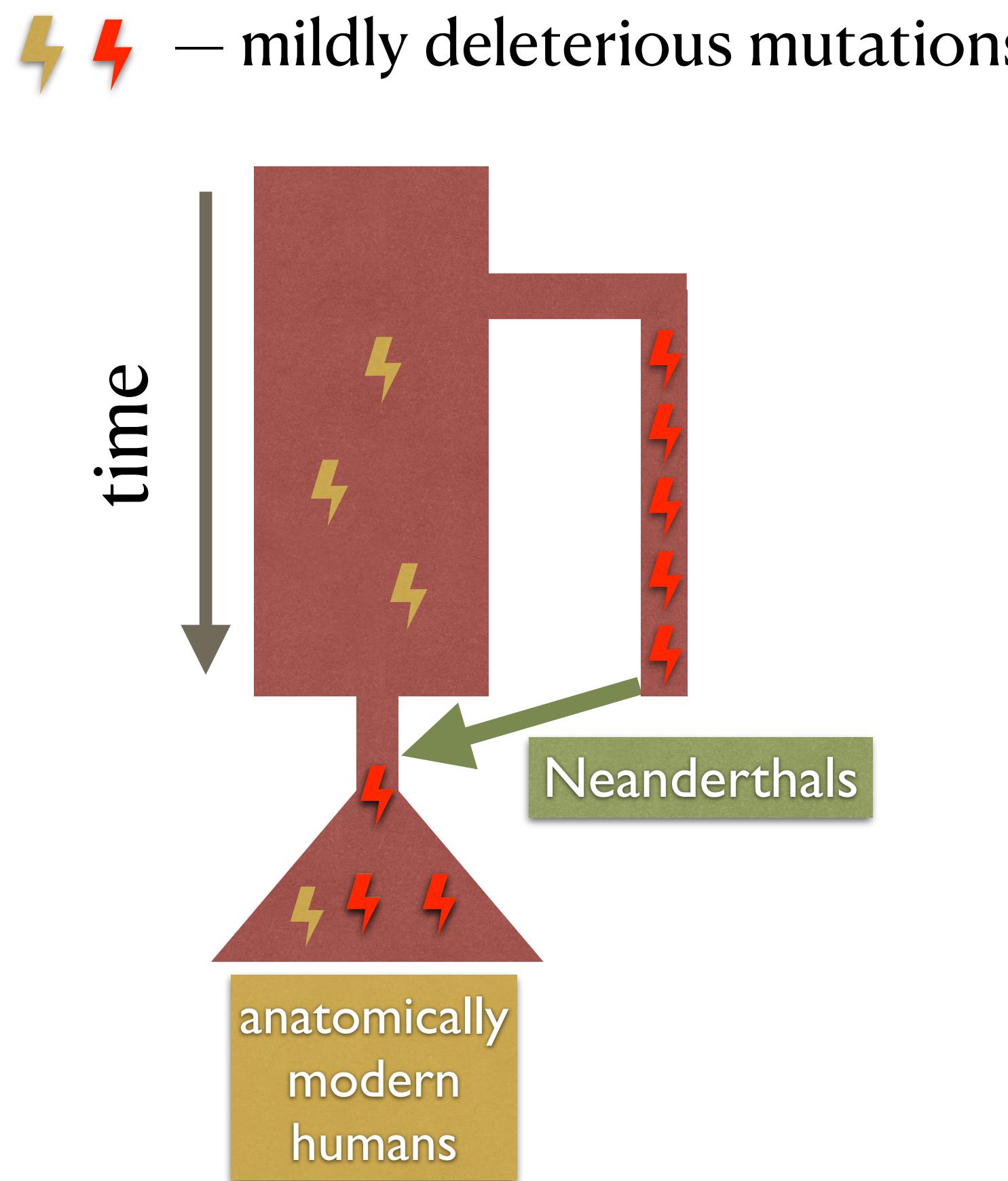
Original Neanderthal mtDNA/Y experienced replacement?



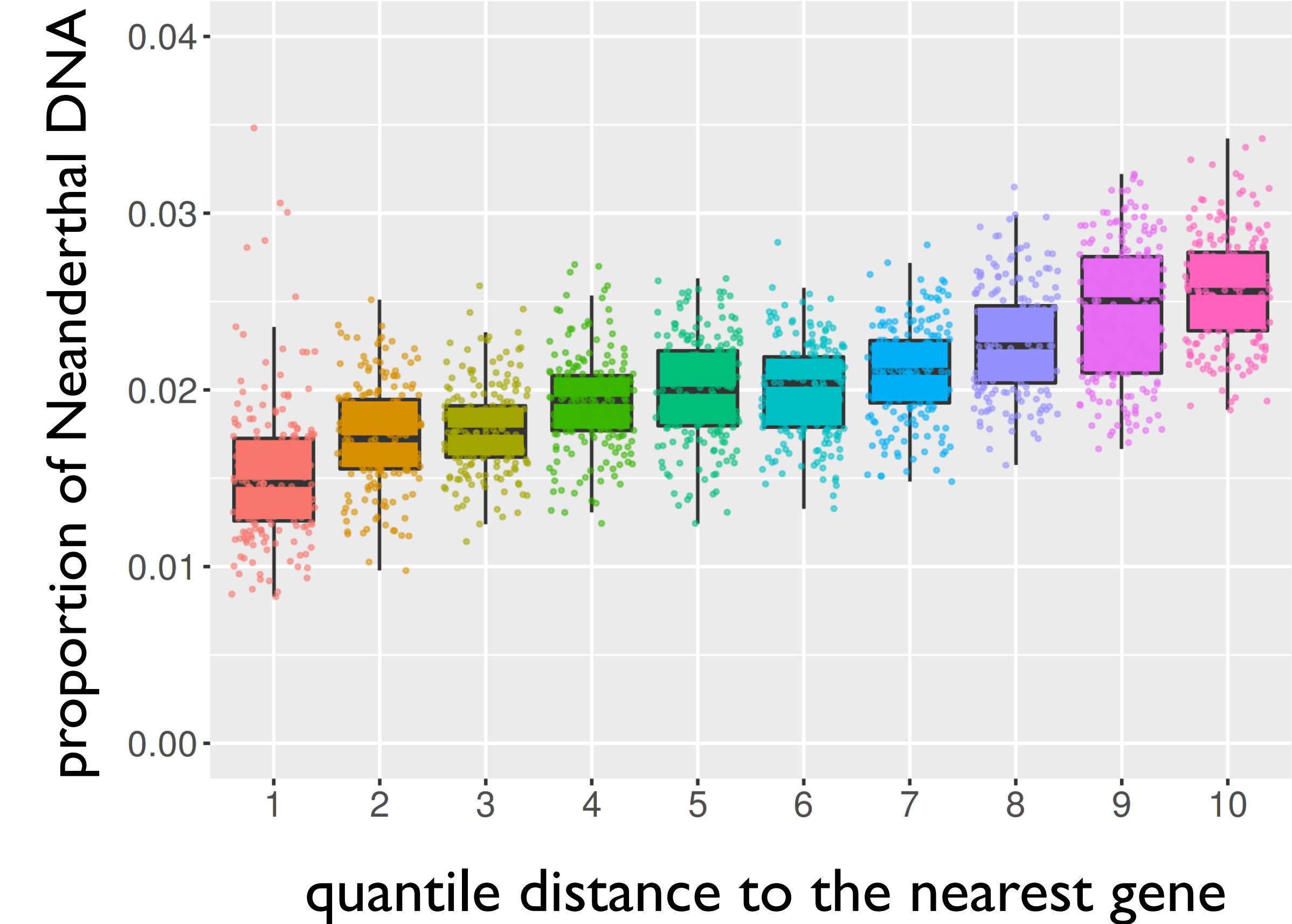
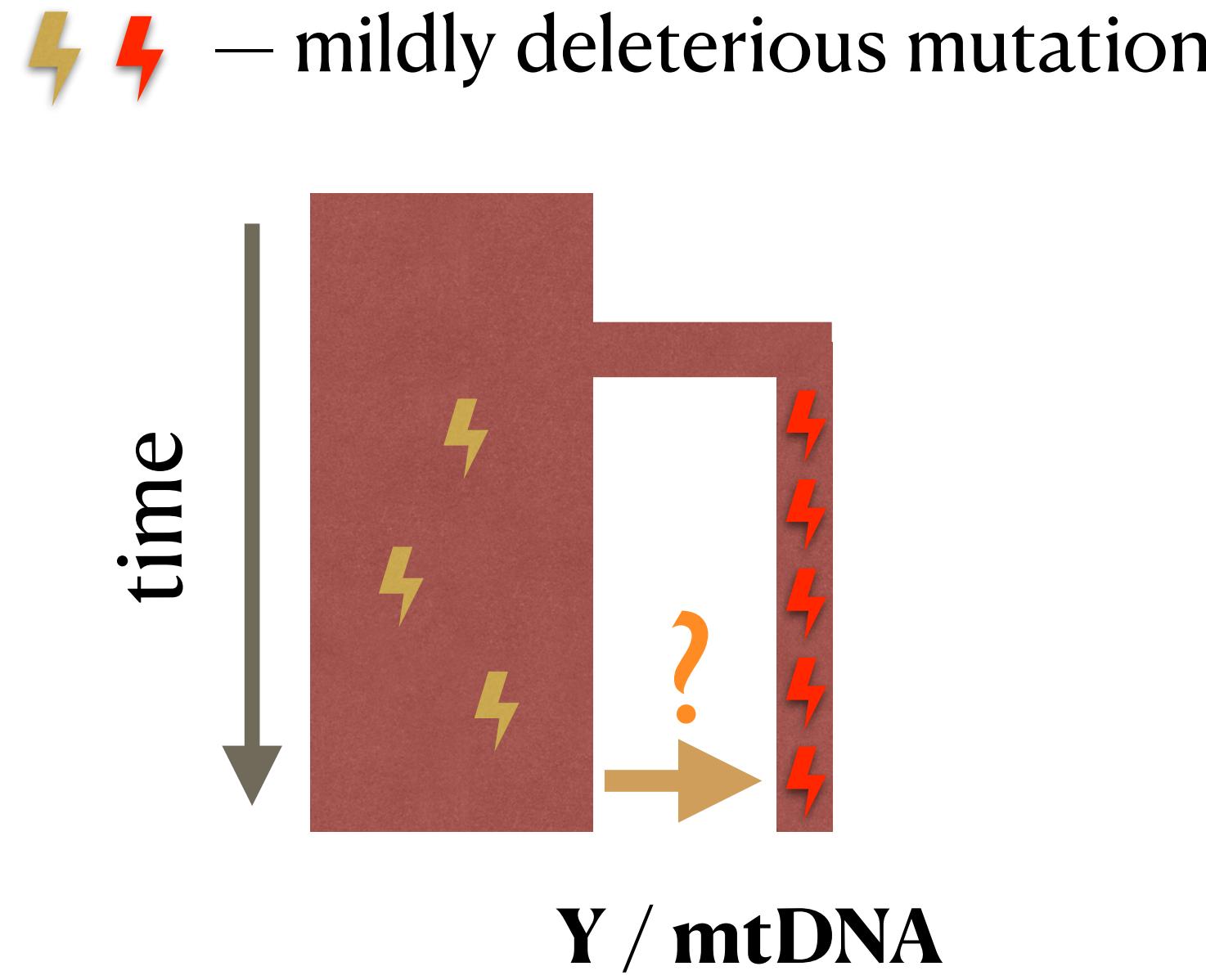
Original Neanderthal mtDNA/Y experienced replacement? (very unlikely under neutrality!)



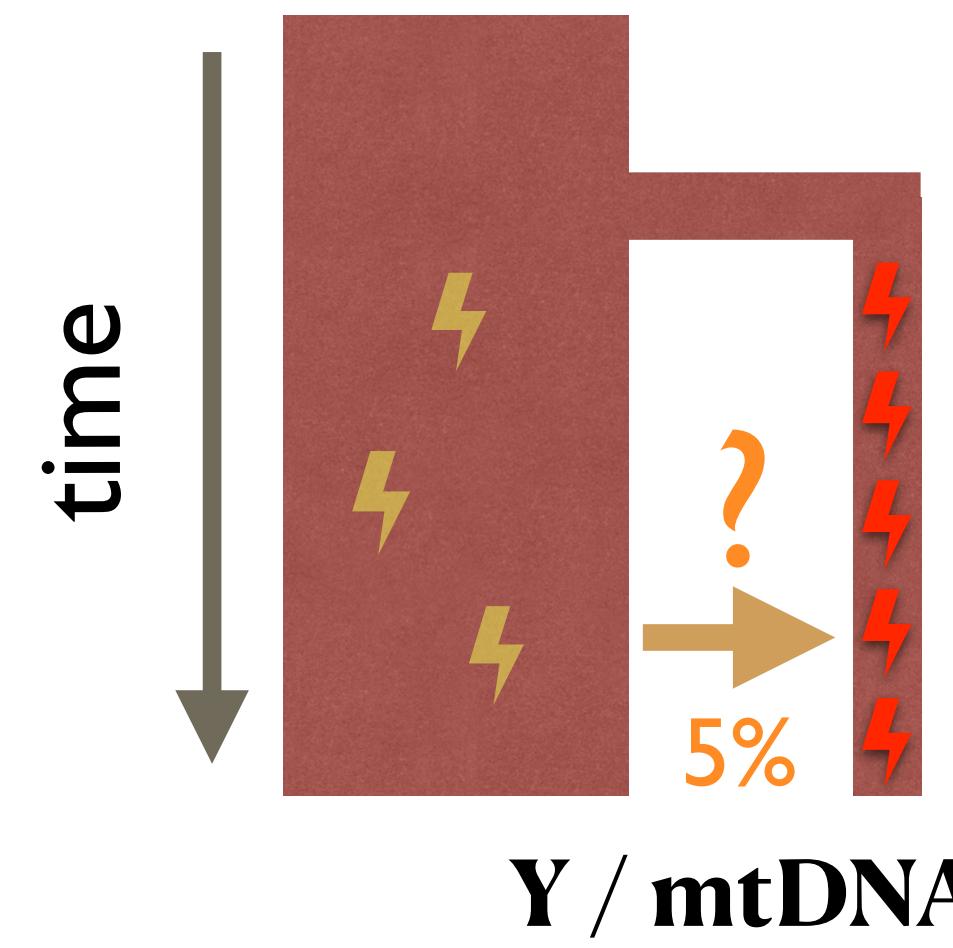
...but we know introgression was not neutral!



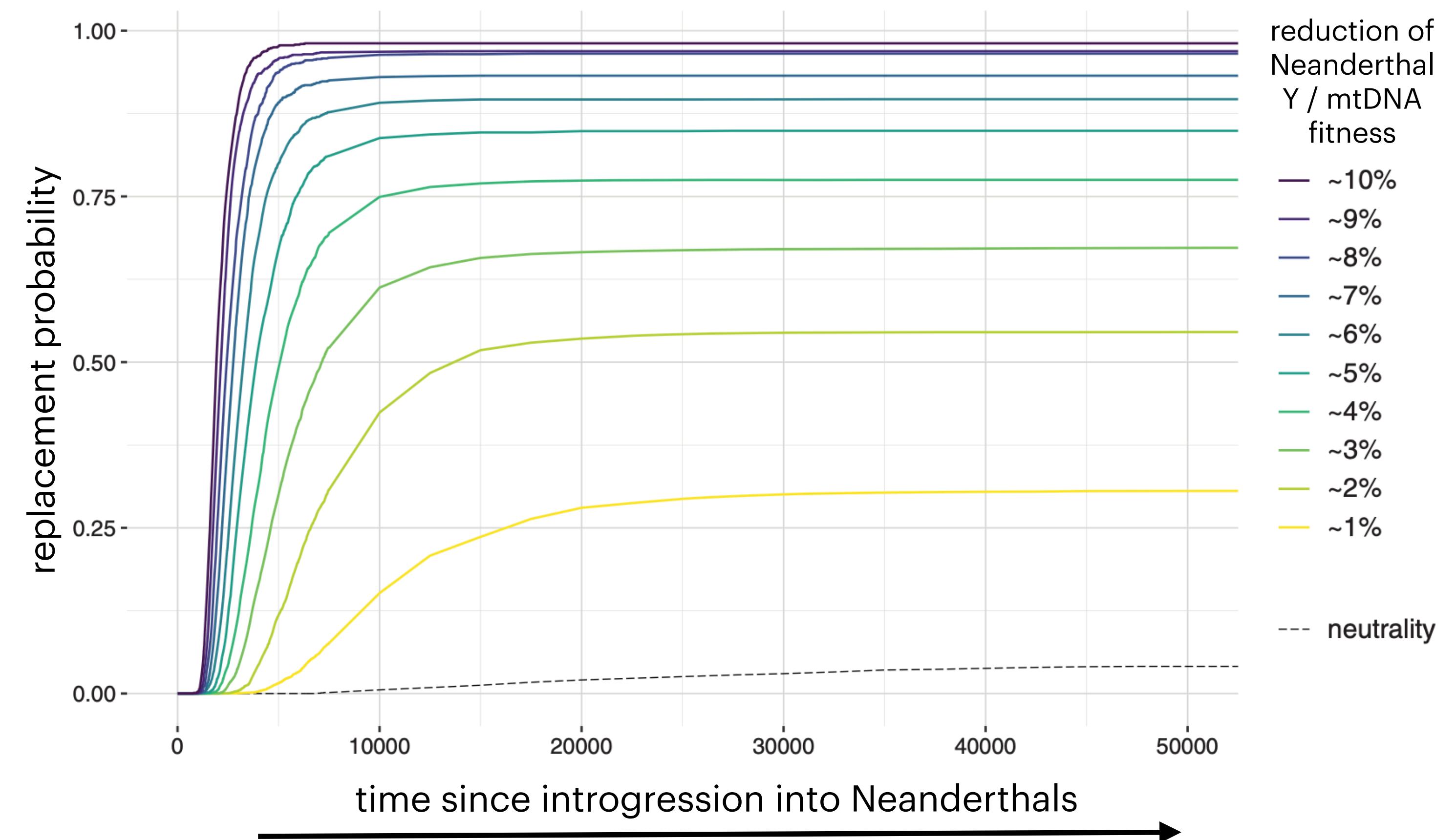
What is the expected scenario for Y / mtDNA introgression?



Natural selection in Neanderthals favors modern human Y / mtDNA



Simulation of 5% gene flow from modern humans



It wasn't all negative though

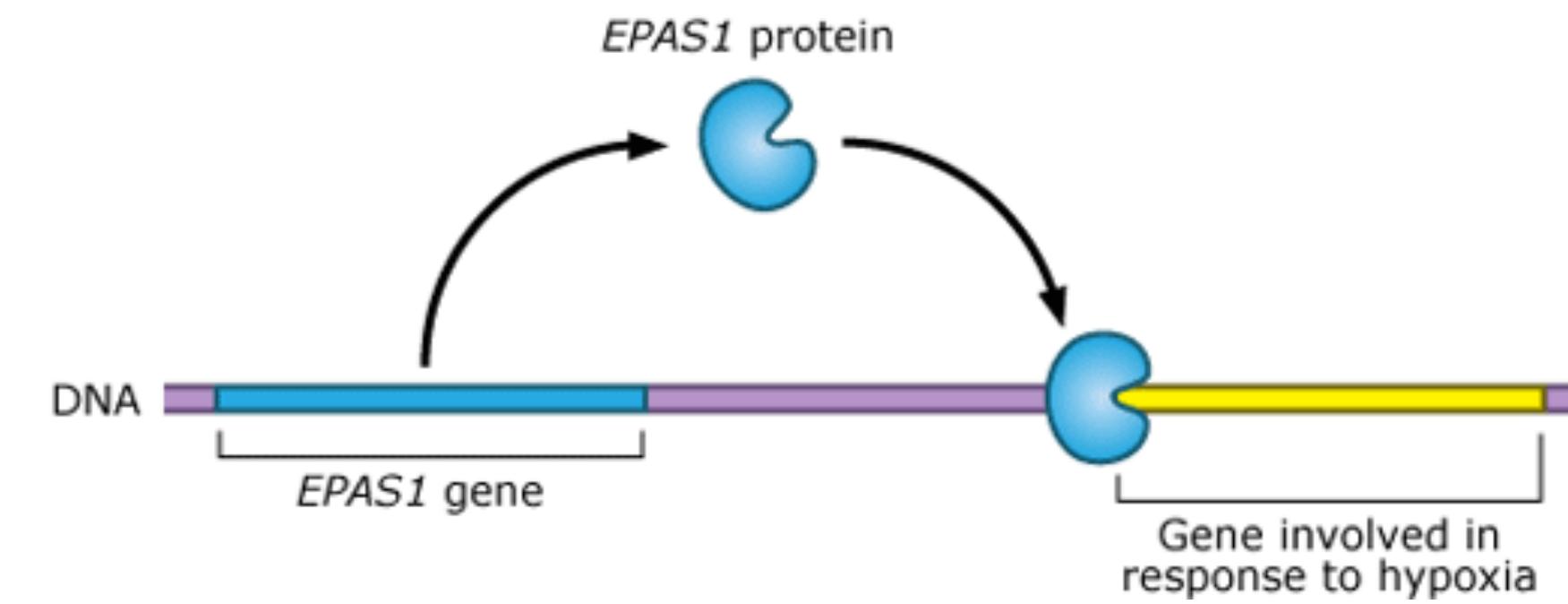
positive selection / adaptive introgression

High altitude adaptation in Tibetans

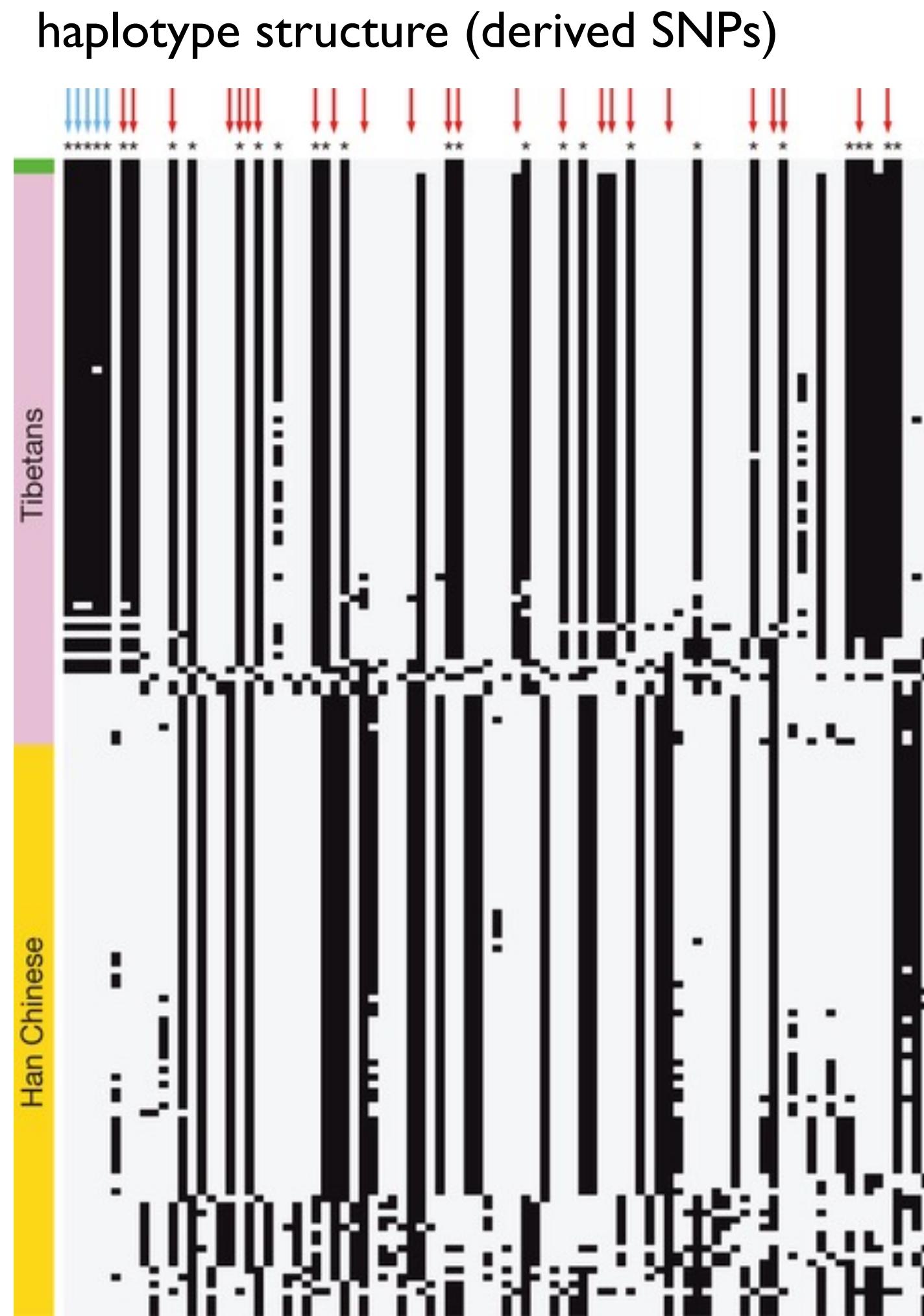
haplotype structure (derived SNPs)



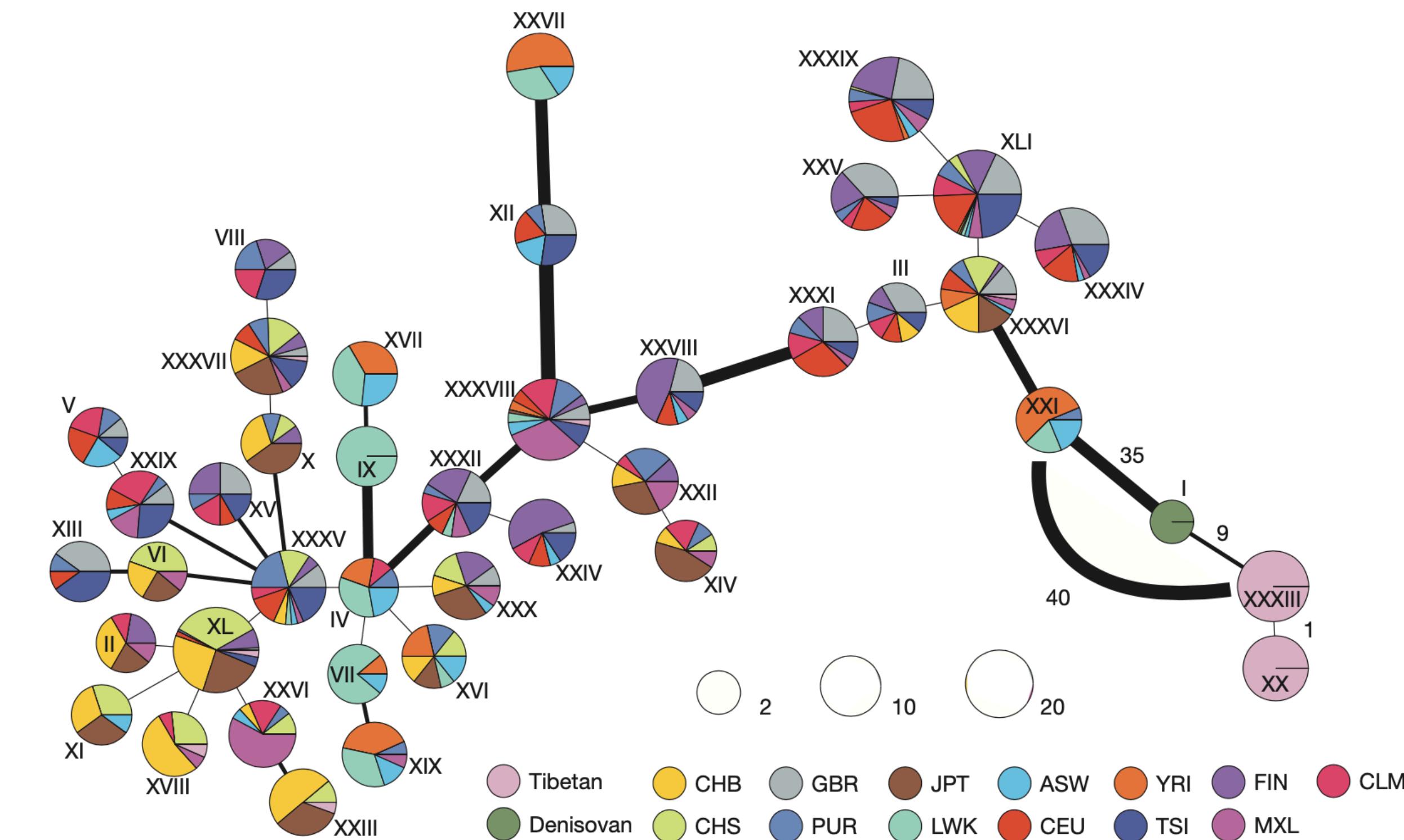
Schematic of *EPAS1* function



High altitude adaptation via Denisovan introgression!



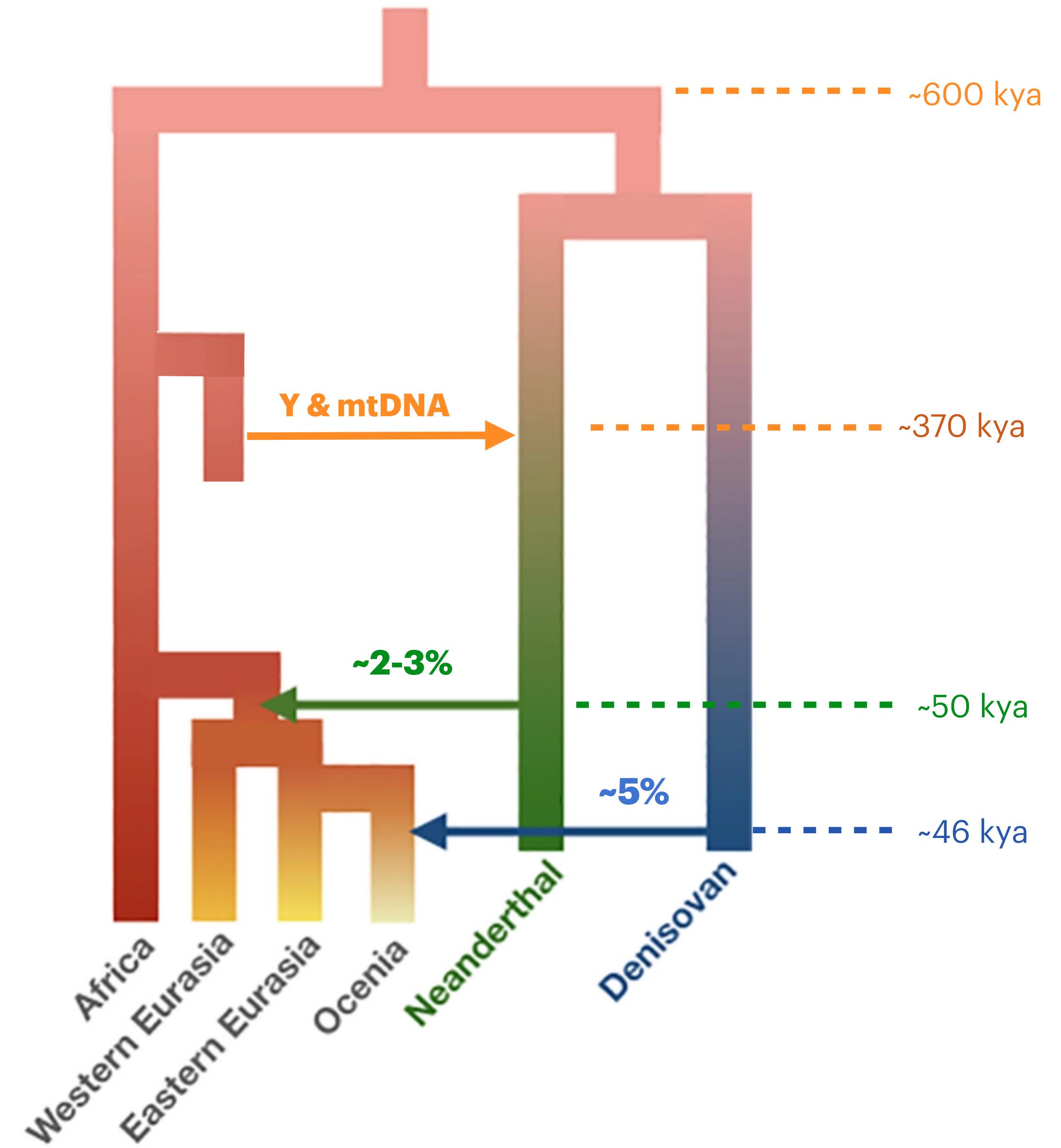
haplotype network based on pairwise-differences



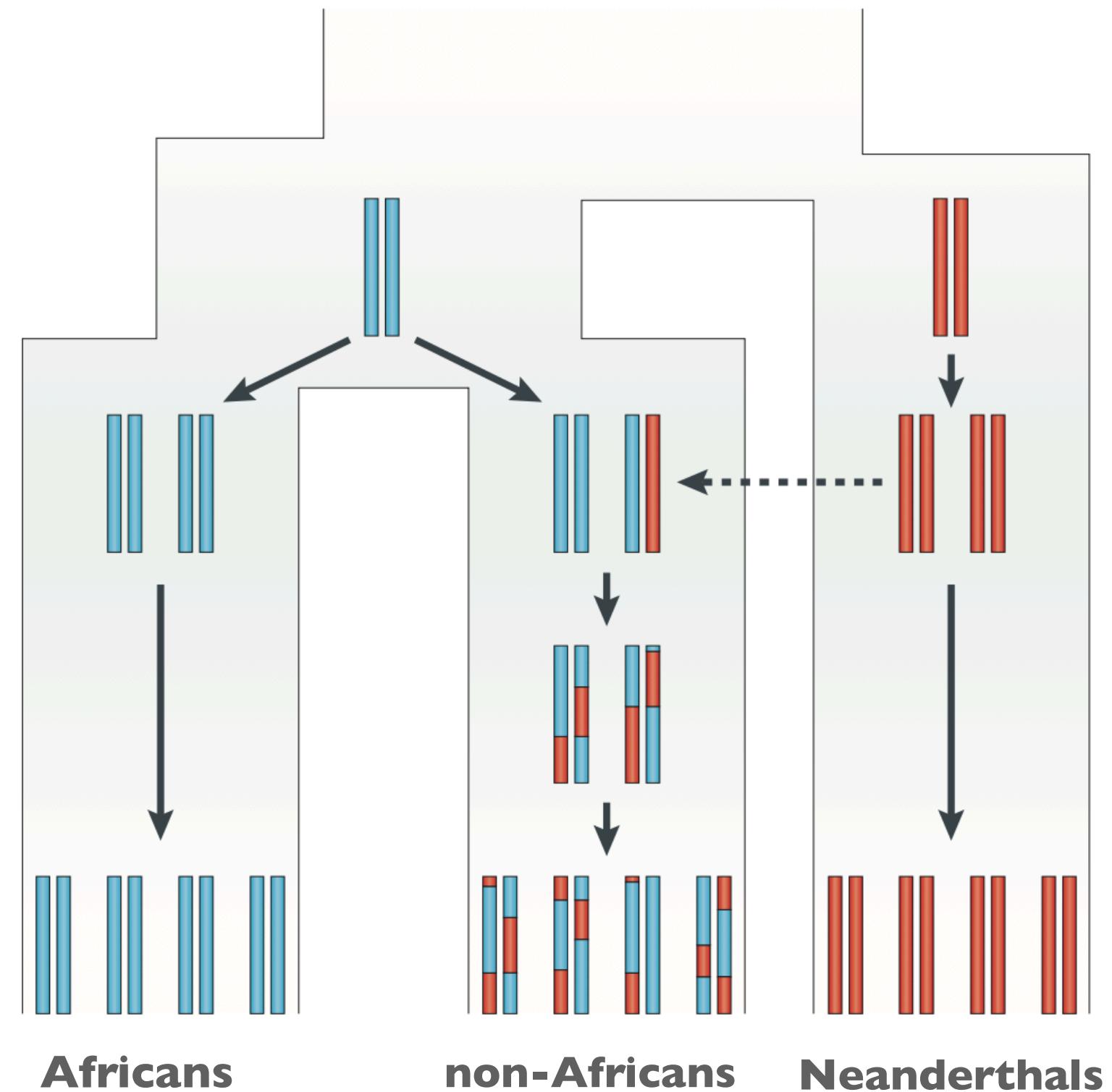
Introgression has been a very frequent occurrence throughout human history.

Introgressed DNA can have a strong impact on the biology of “recipient populations” (not just negative!).

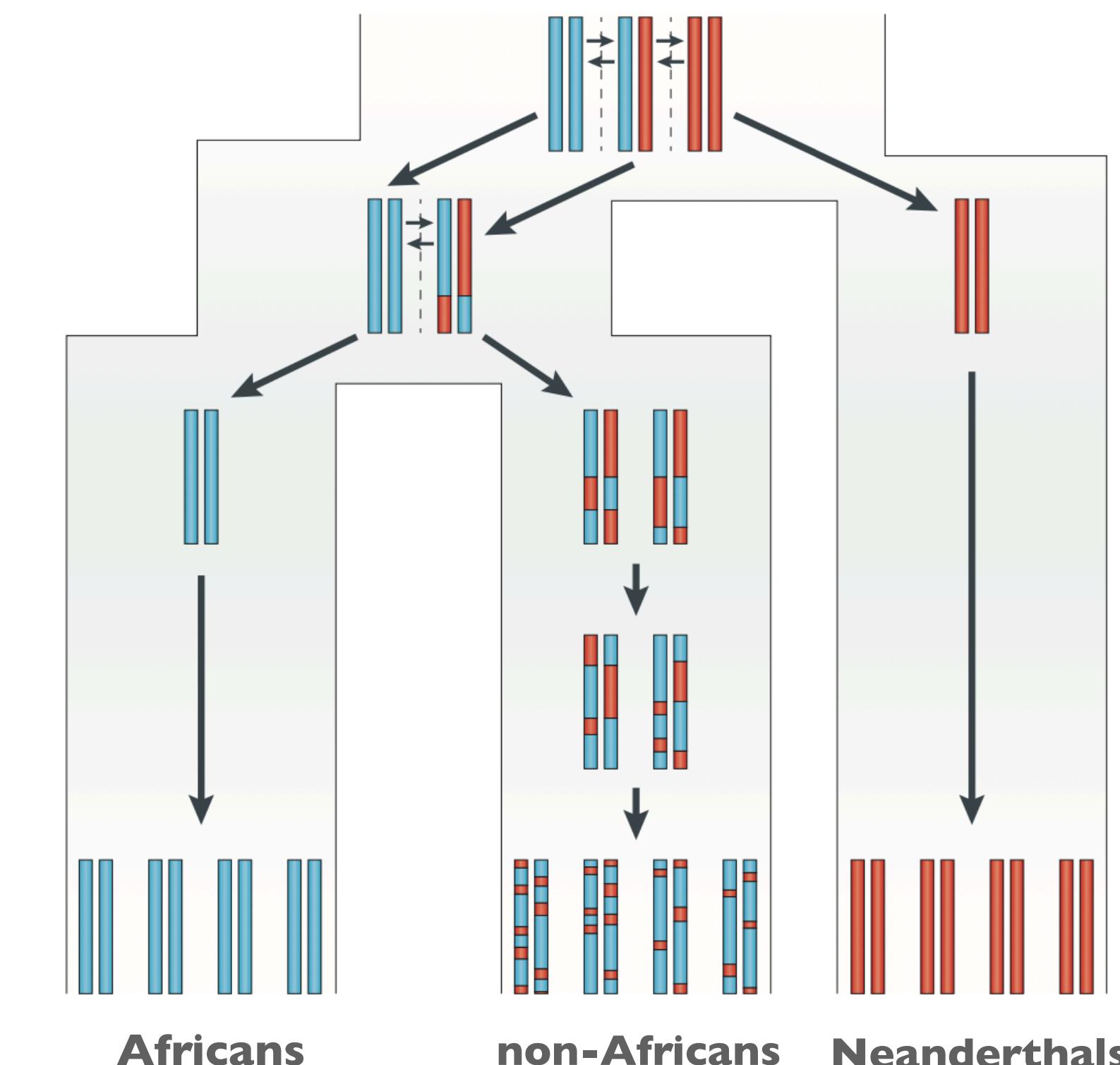
Population genetic simulations critical for testing evolutionary hypotheses.



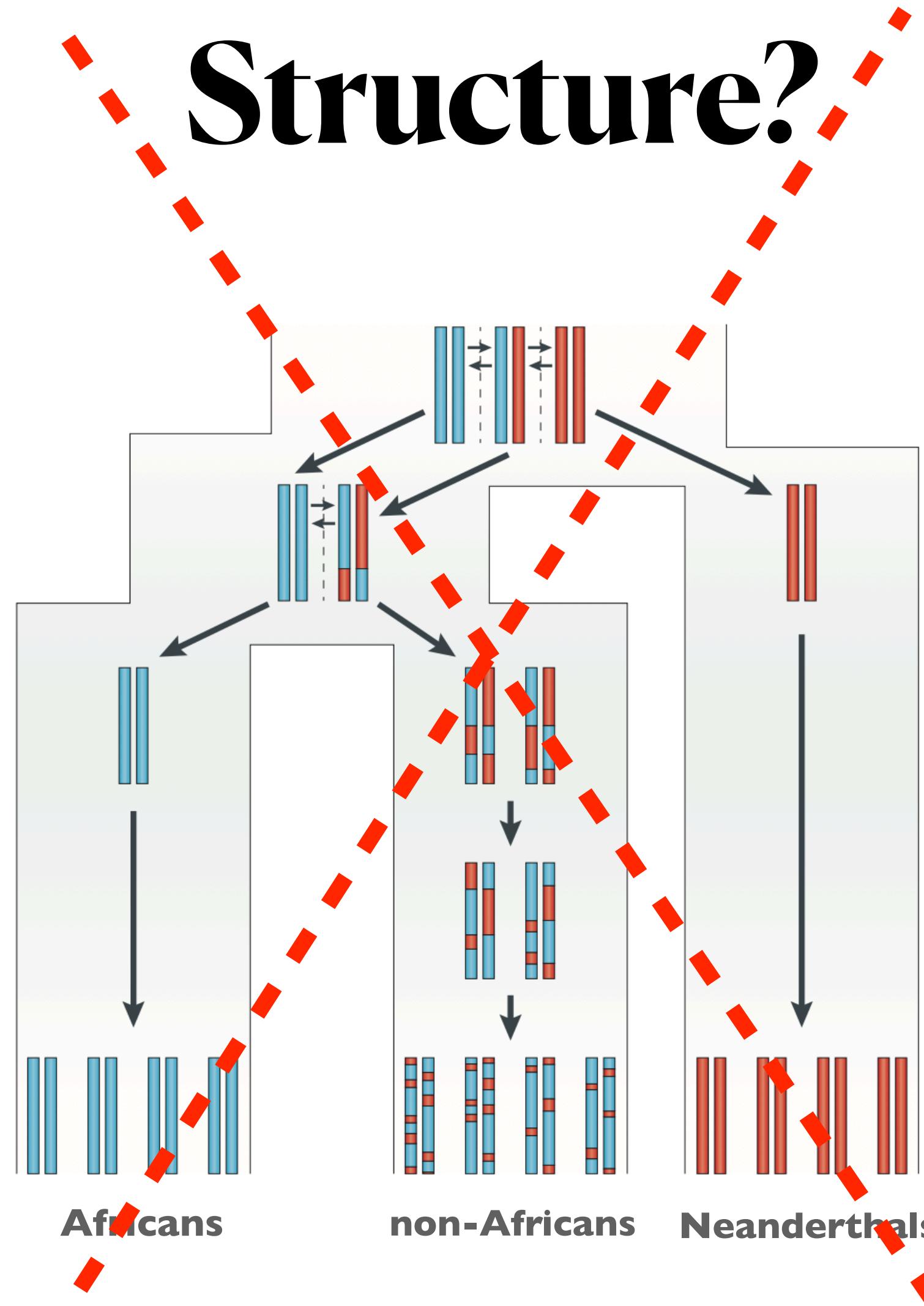
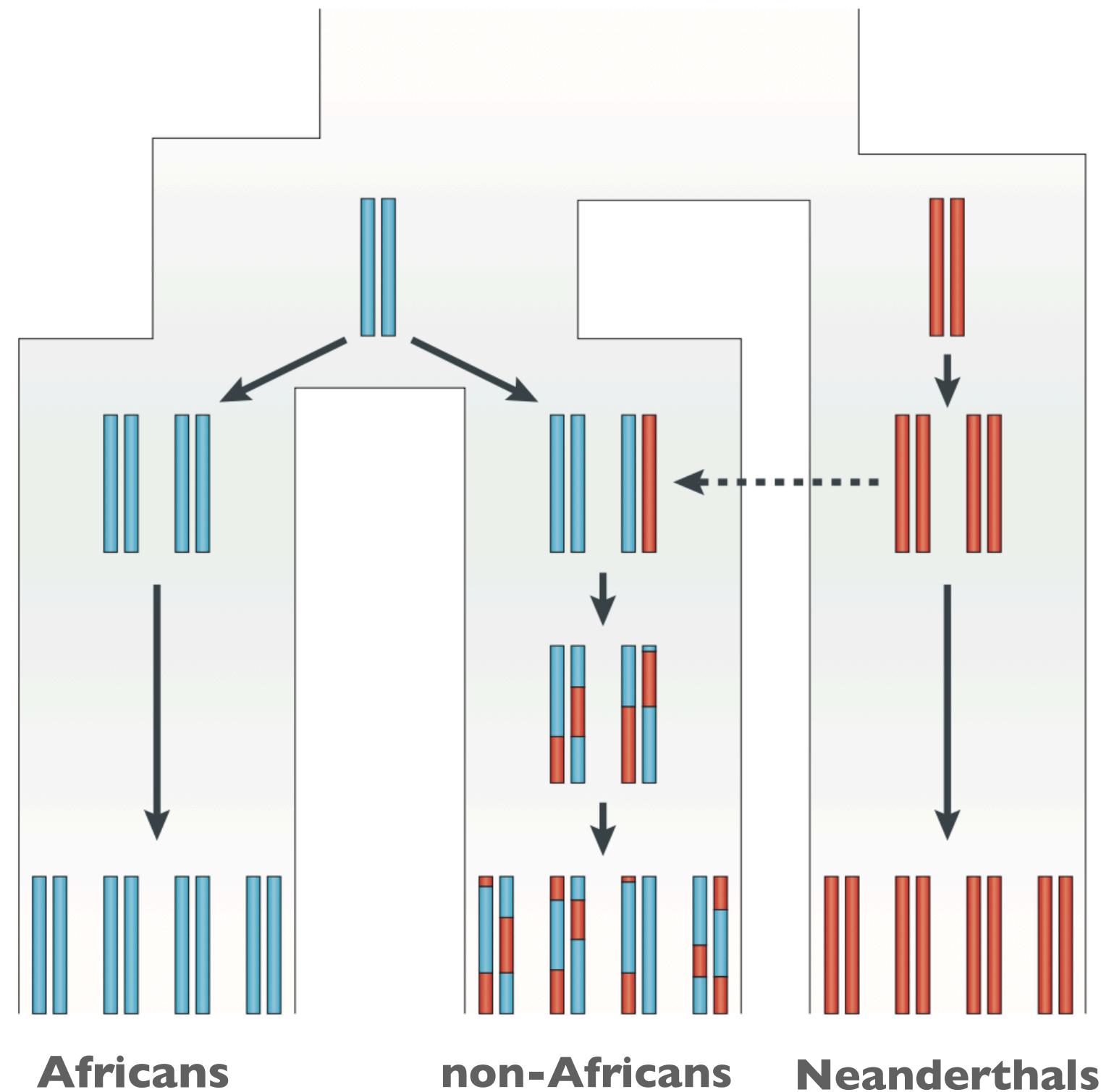
Introgression?



Structure?

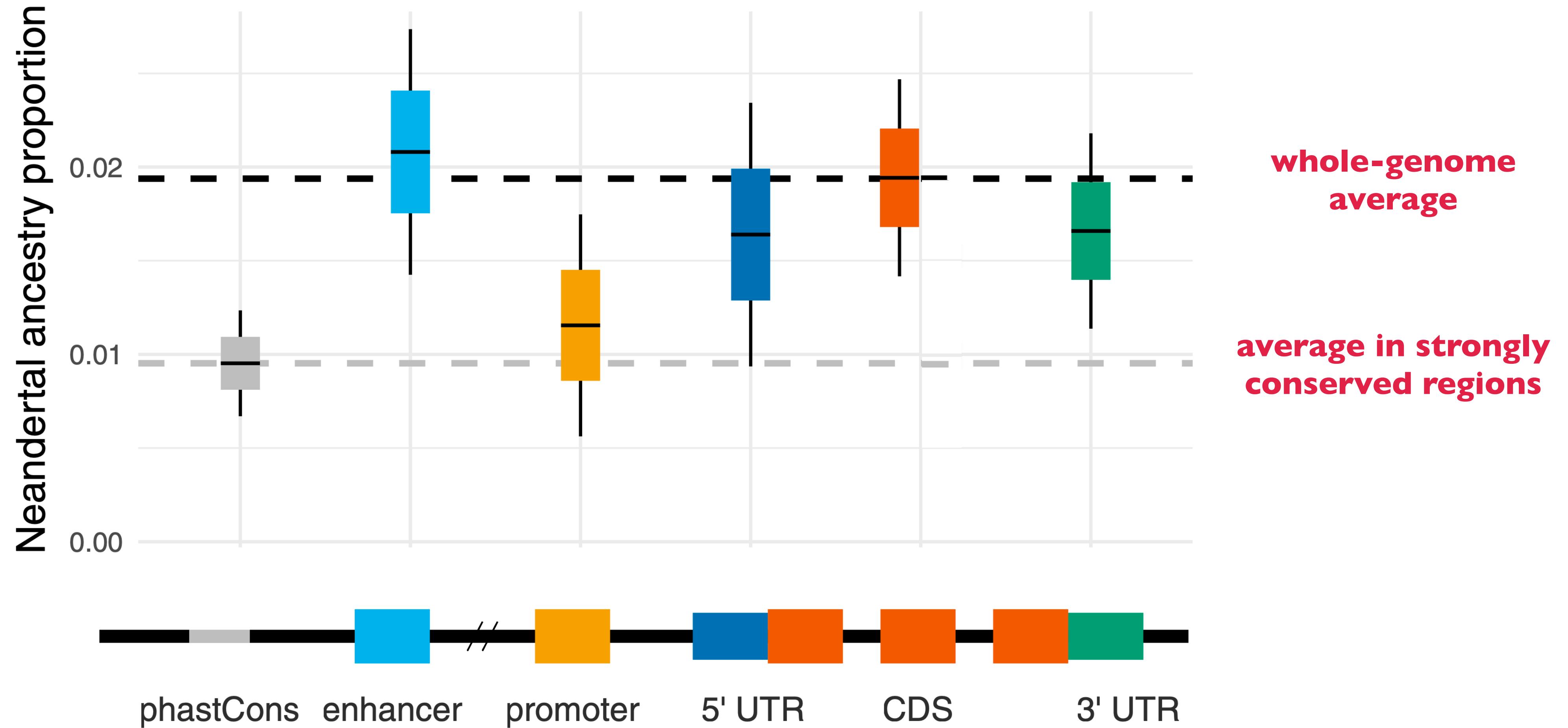


Introgression?

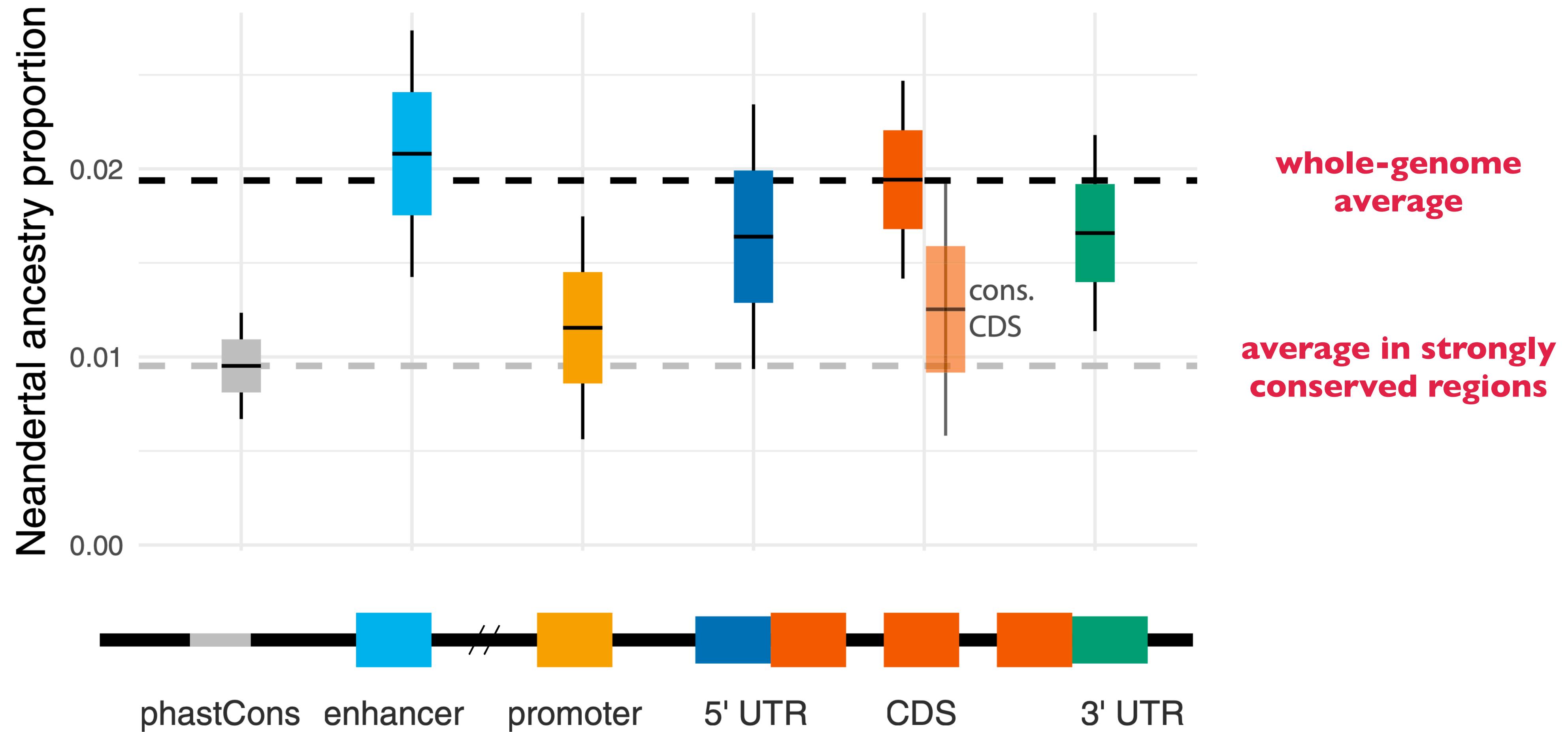


Under structure, “Neanderthal-looking”
haplotypes will be much shorter!

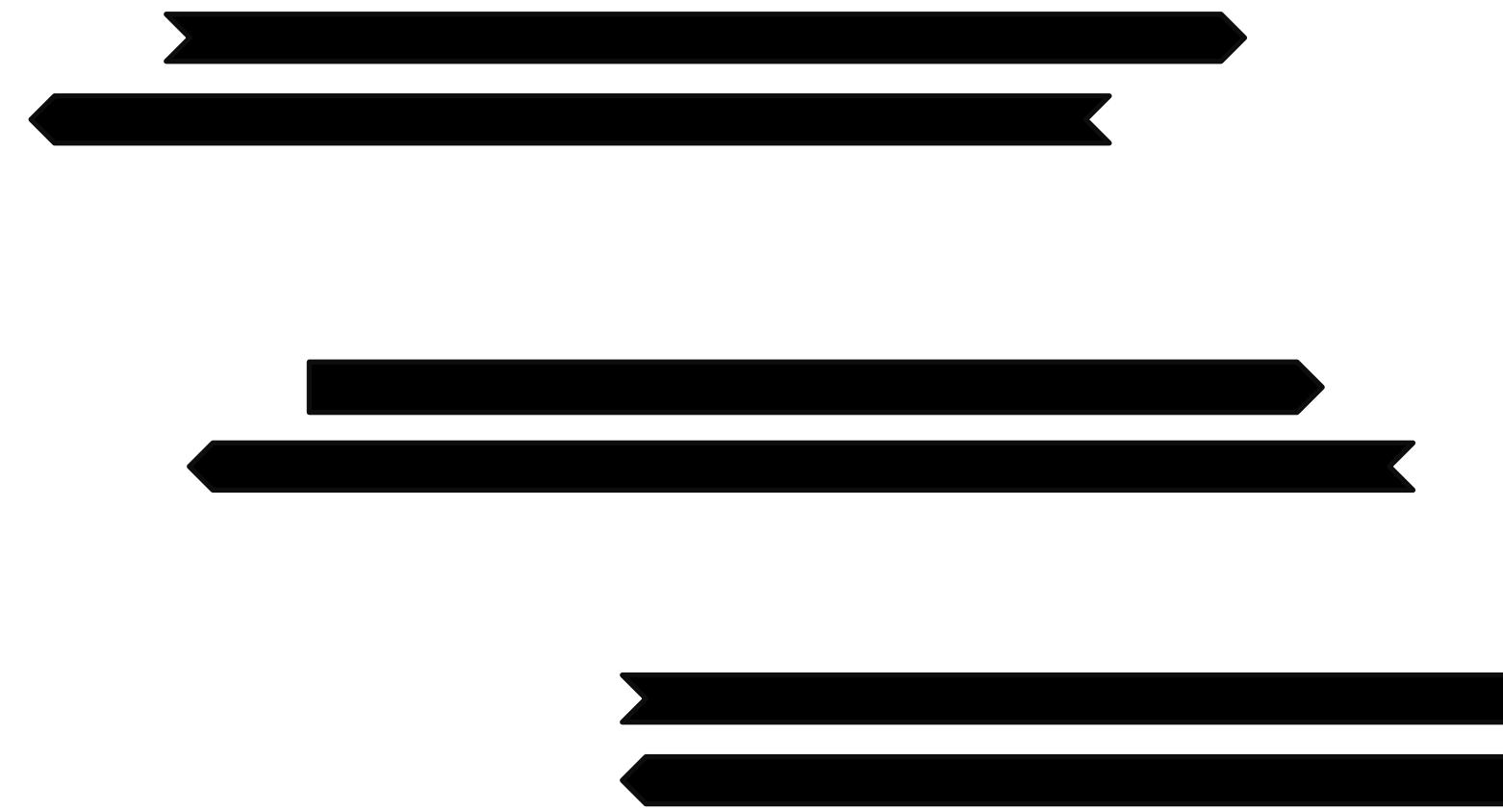
The story is much more complicated if we zoom in...



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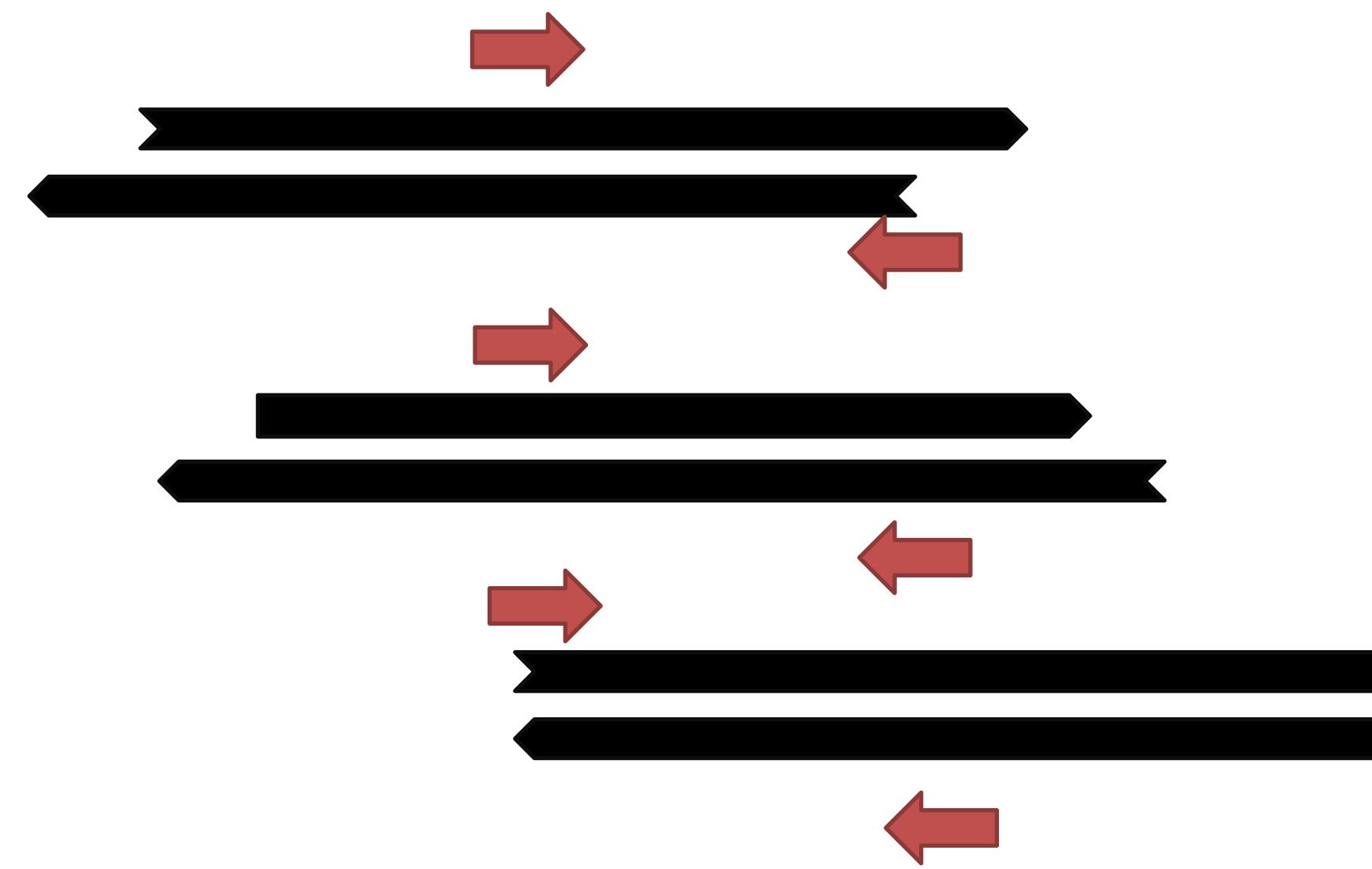
PCR



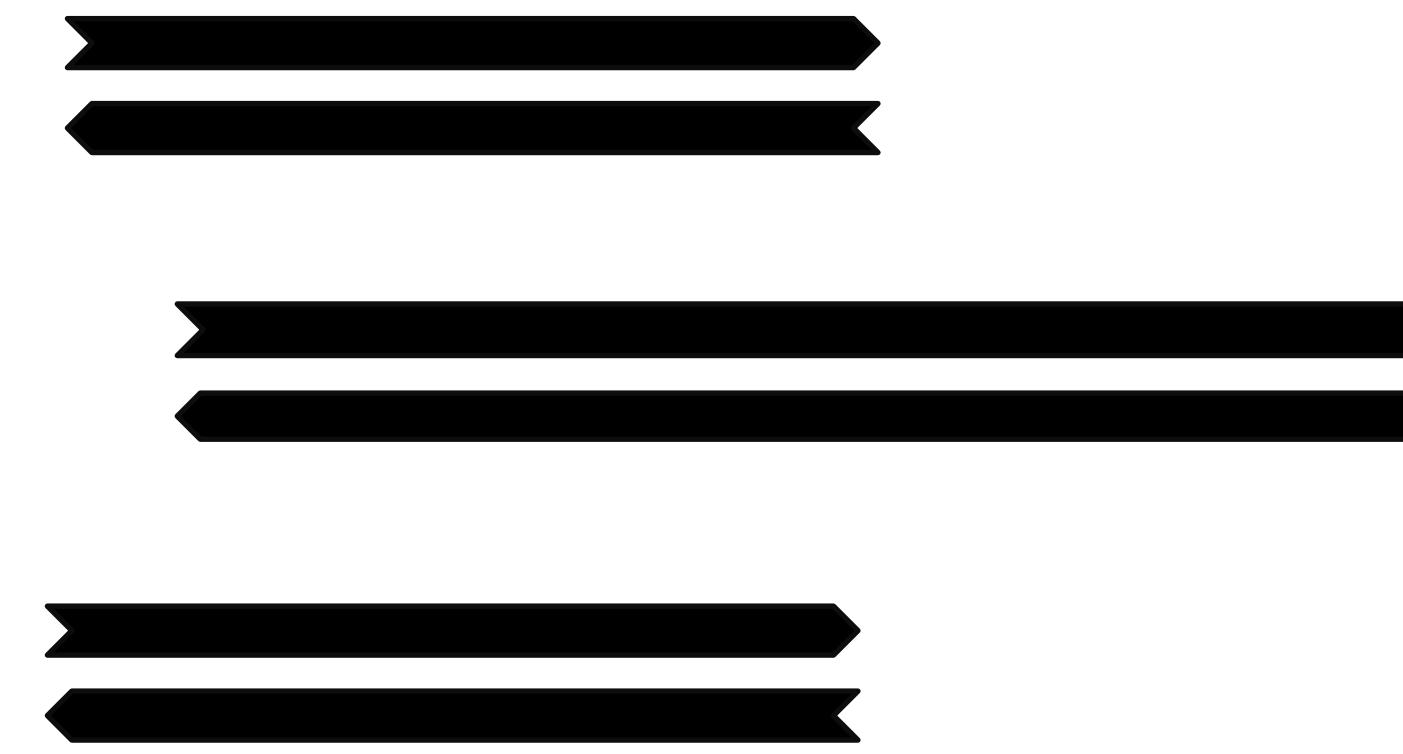
Library preparation



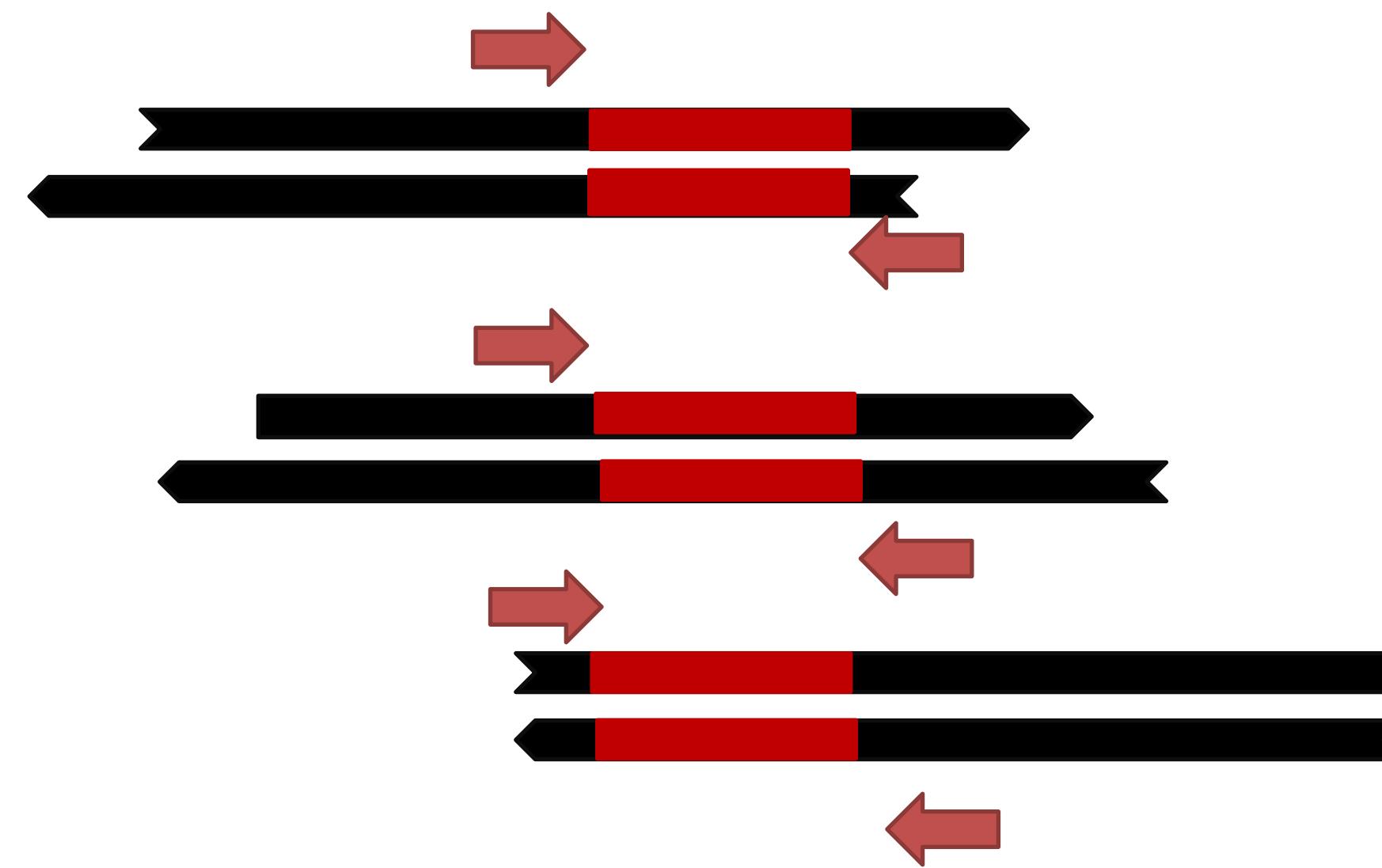
PCR



Library preparation



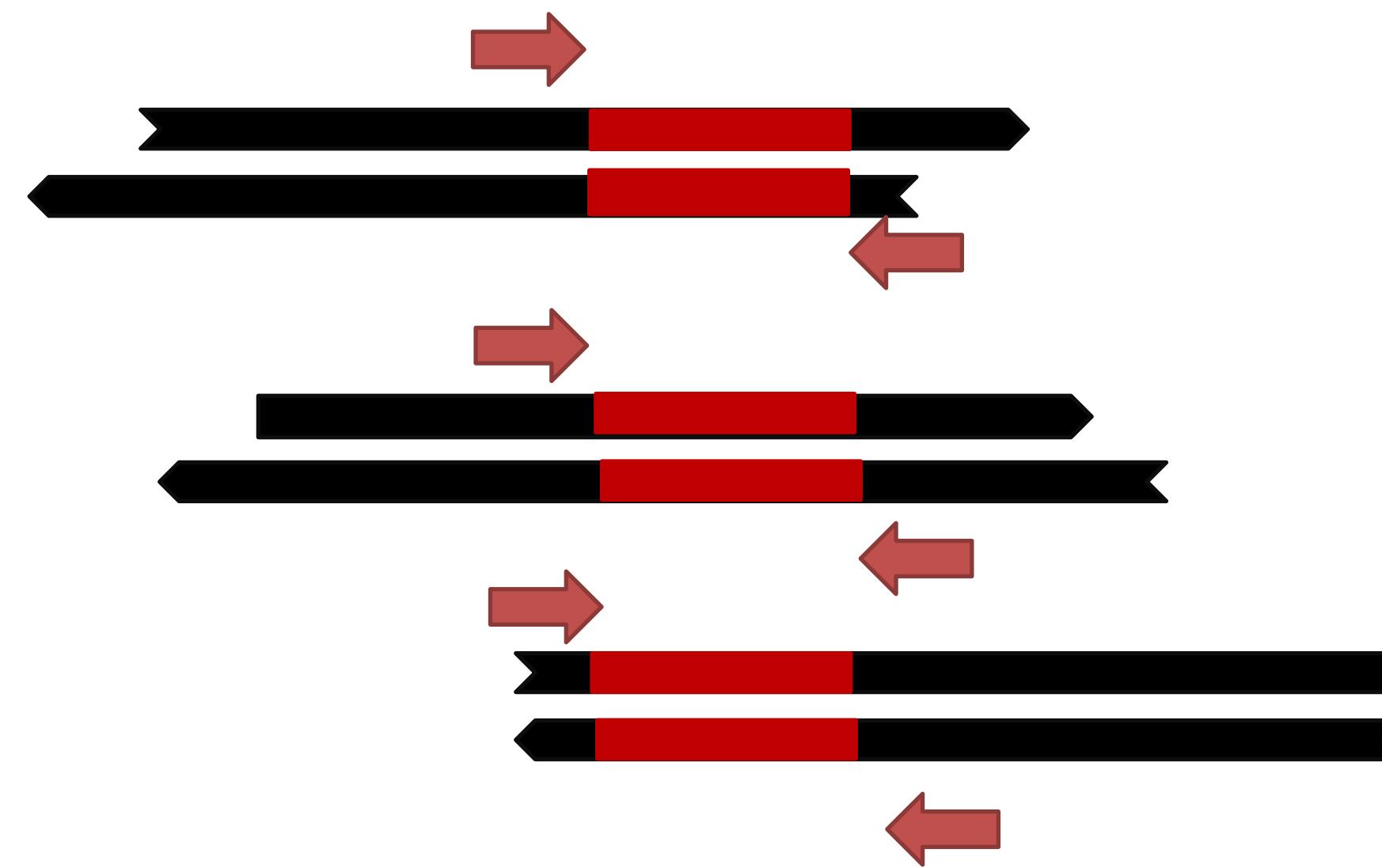
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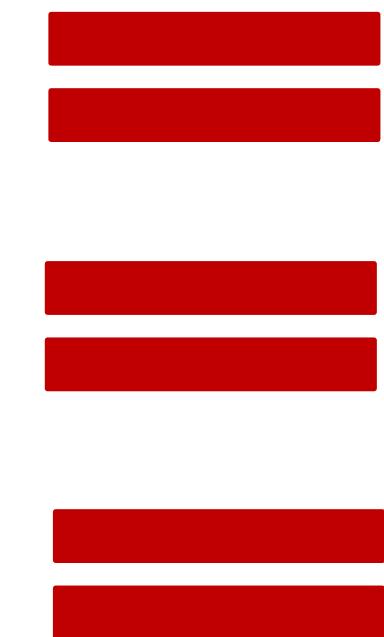
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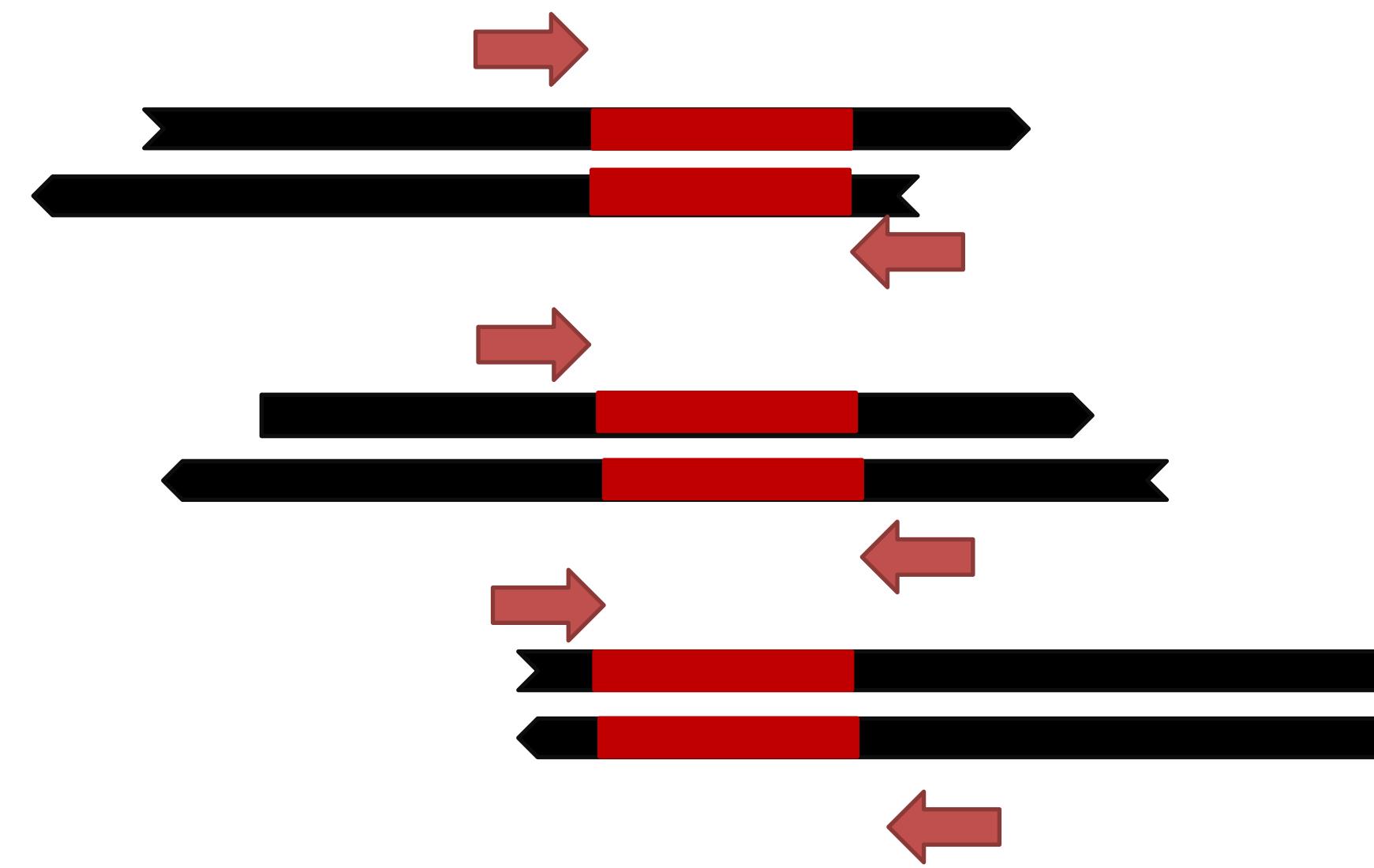
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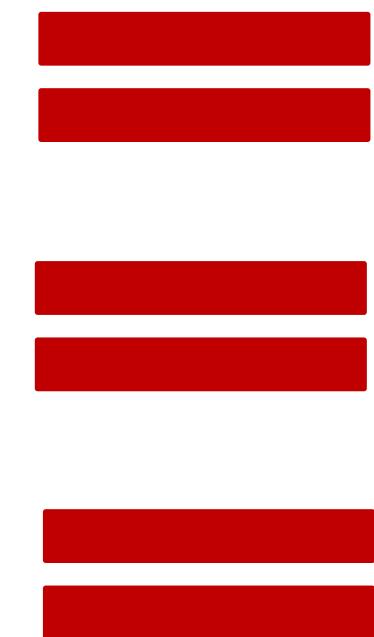
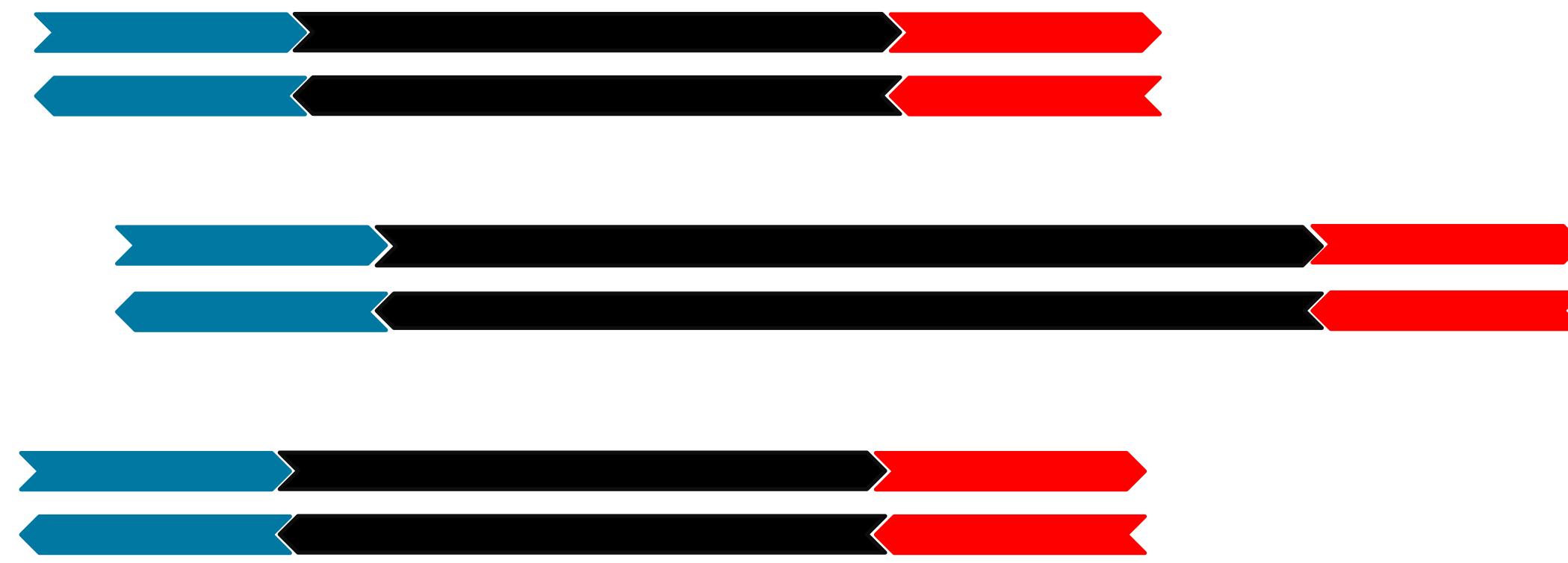
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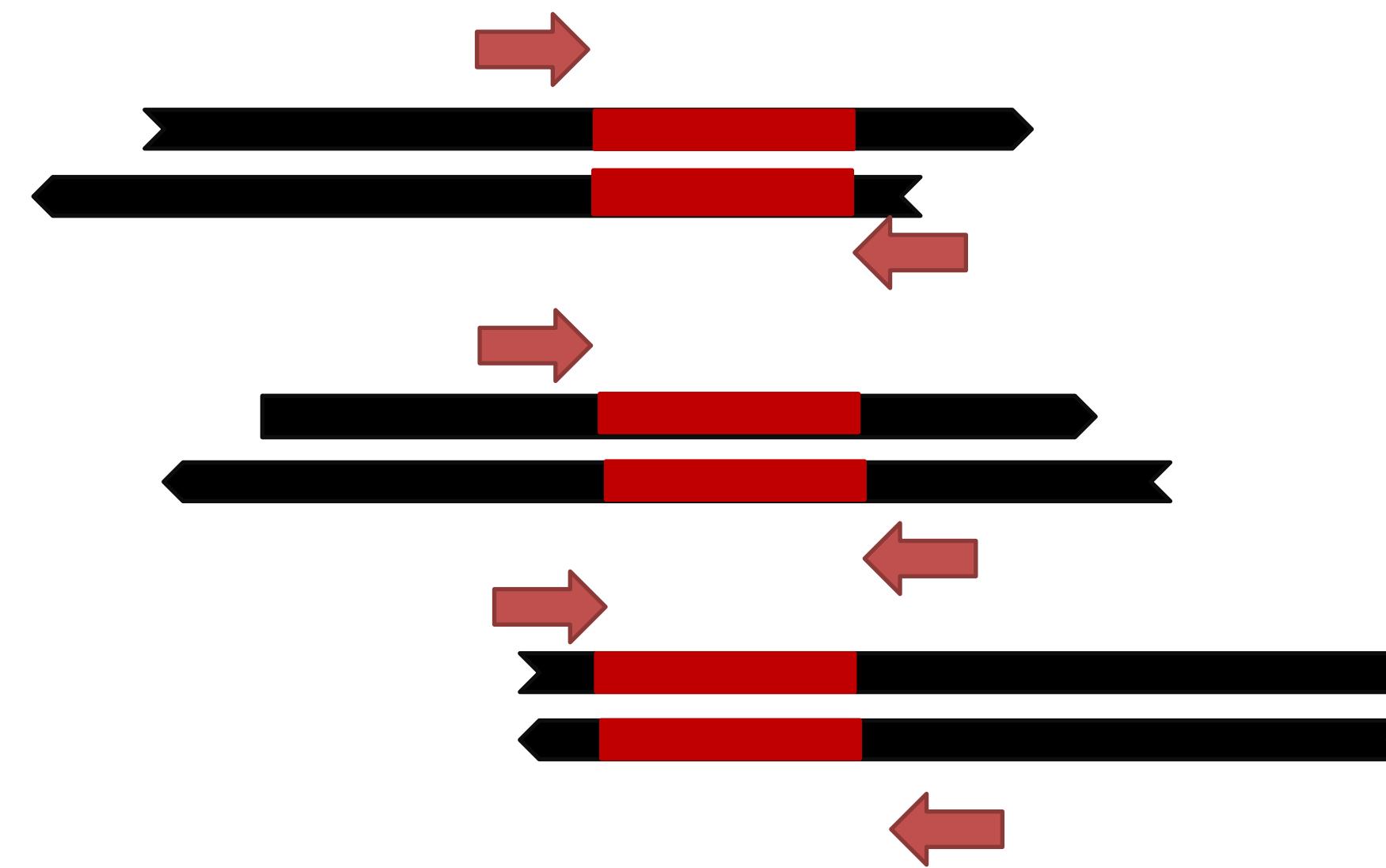
PCR



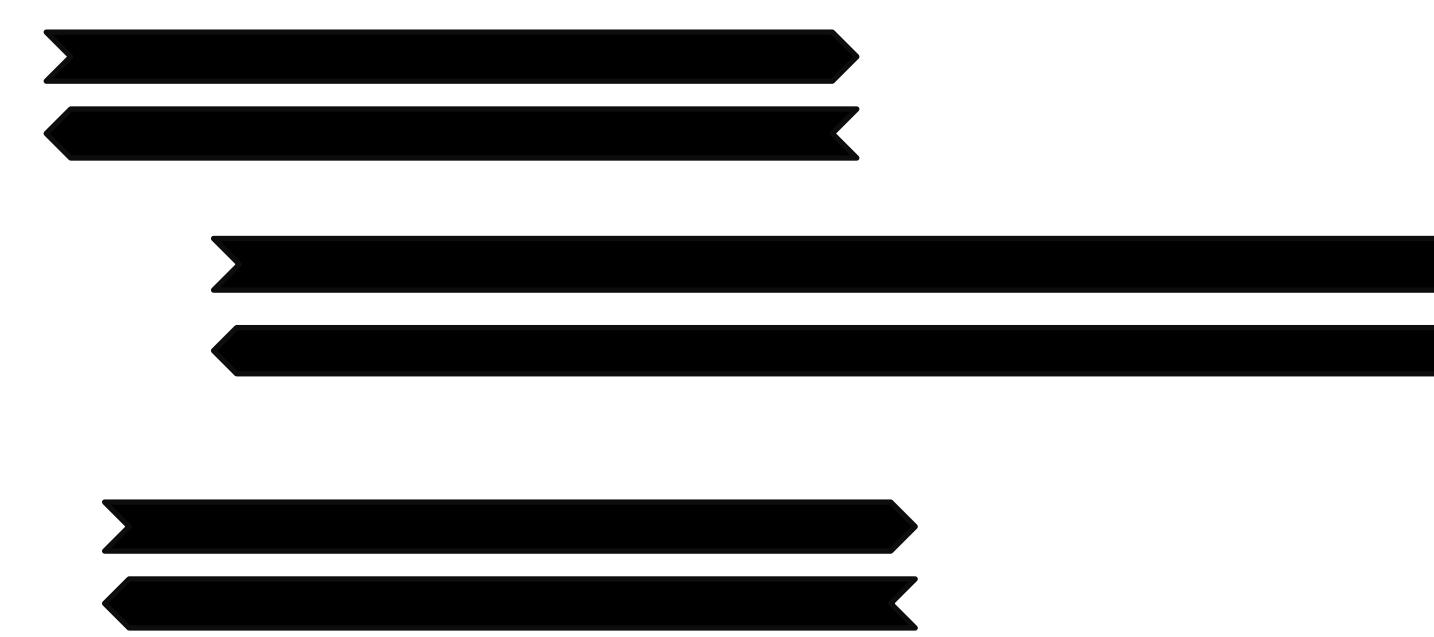
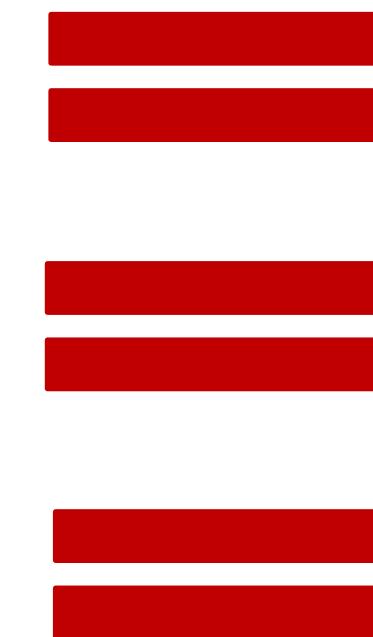
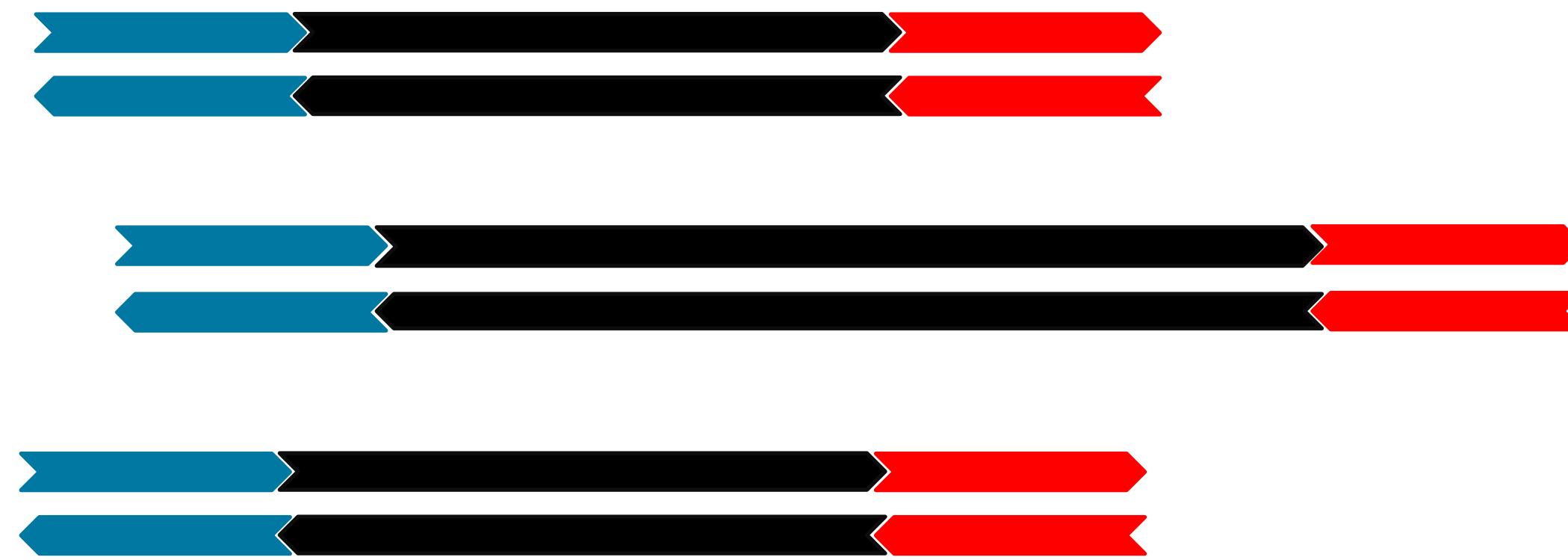
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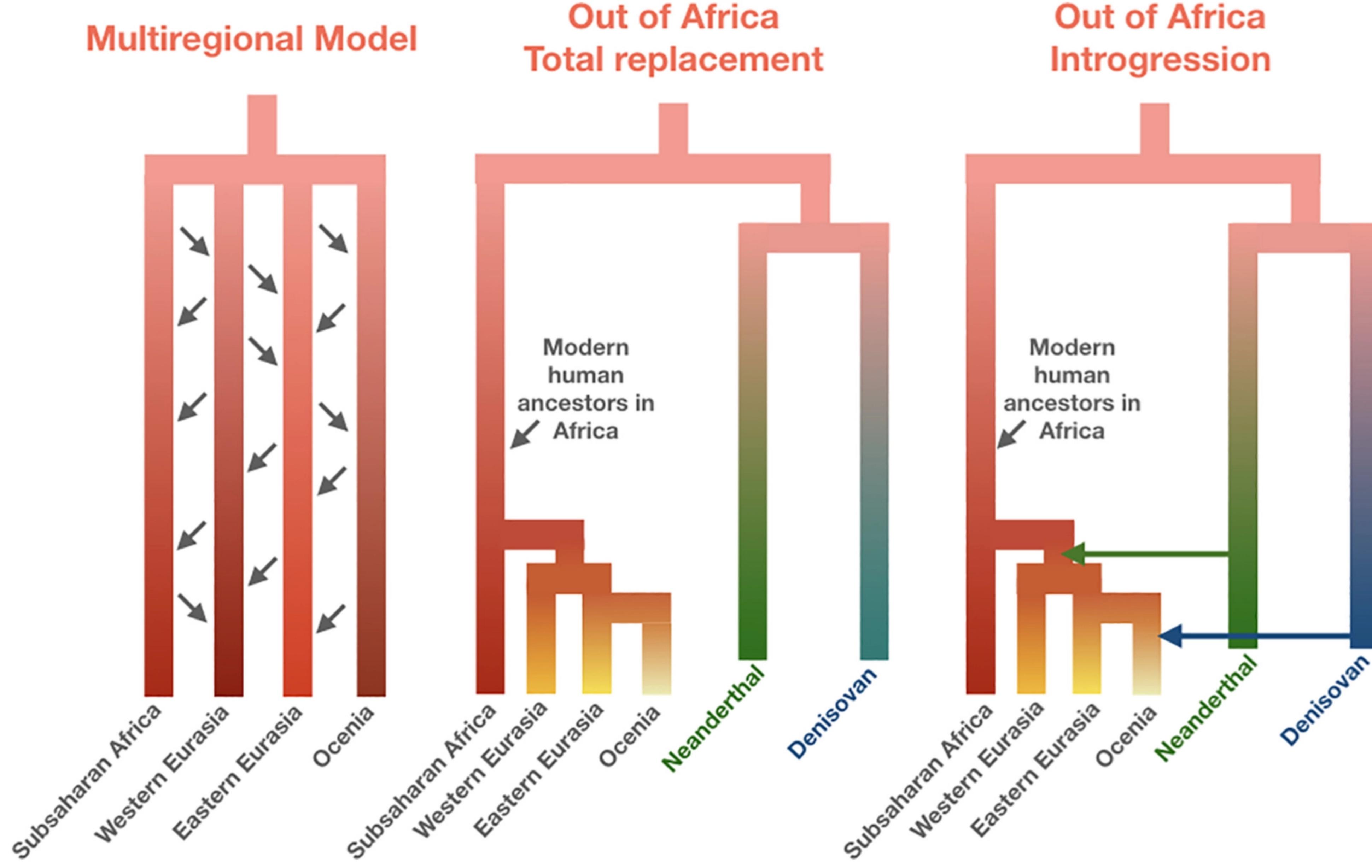


PCR



Library preparation





Genomics in practice...

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Genomics in practice...

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12 60474 . G A 100 PASS . GT:AP 0|0:0.000,0.000 0|1:0.015,0.705 0|1:0.020,0.775
12 60614 . C A 100 PASS . GT:AP 0|0:0.000,0.000 0|0:0.000,0.005 0|0:0.000,0.015
12 60628 . T C 100 PASS . GT:AP 0|0:0.000,0.000 0|0:0.000,0.070 0|0:0.000,0.000
12 60654 . G A 100 PASS . GT:AP 0|0:0.000,0.000 0|0:0.000,0.000 0|0:0.000,0.000
12 61021 . C T 100 PASS . GT:AP 0|0:0.000,0.000 0|0:0.000,0.000 0|0:0.000,0.000
12 61107 . G T 100 PASS . GT:AP 0|0:0.000,0.000 0|0:0.000,0.015 0|0:0.000,0.000
12 61172 . G A 100 PASS . GT:AP 0|0:0.000,0.000 0|0:0.000,0.000 0|0:0.000,0.000
12 61220 . G A 100 PASS . GT:AP 0|0:0.000,0.000 0|0:0.005,0.265 0|1:0.020,0.665
12 61258 . C T 100 PASS . GT:AP 0|0:0.000,0.000 0|0:0.005,0.465 0|1:0.020,0.895
12 61272 . T C 100 PASS . GT:AP 0|0:0.000,0.000 0|0:0.000,0.000 0|0:0.000,0.000
12 61329 . C T 100 PASS . GT:AP 0|0:0.000,0.000 0|0:0.000,0.000 0|0:0.000,0.000
12 61341 . G A 100 PASS . GT:AP 0|0:0.000,0.000 0|1:0.010,0.560 0|1:0.020,0.855
12 61368 . C T 100 PASS . GT:AP 0|0:0.000,0.000 0|1:0.020,0.630 0|1:0.020,0.955
12 61392 . T A 100 PASS . GT:AP 0|0:0.000,0.000 0|0:0.000,0.000 0|0:0.000,0.000
12 61405 . G C 100 PASS . GT:AP 0|0:0.000,0.000 0|0:0.000,0.000 0|0:0.000,0.000
12 61411 . C A 100 PASS . GT:AP 0|0:0.000,0.000 0|0:0.000,0.000 0|0:0.000,0.015
12 61416 . G A 100 PASS . GT:AP 0|0:0.000,0.025 0|0:0.000,0.010 0|0:0.015,0.075
12 61422 . C T 100 PASS . GT:AP 0|0:0.000,0.000 0|0:0.000,0.105 0|0:0.005,0.010
12 61476 . C G 100 PASS . GT:AP 0|0:0.000,0.000 0|0:0.000,0.000 0|0:0.005,0.000
12 61510 . G A 100 PASS . GT:AP 0|0:0.000,0.000 0|0:0.000,0.000 0|0:0.000,0.000
12 61516 . C T 100 PASS . GT:AP 0|0:0.000,0.000 0|0:0.000,0.005 0|0:0.000,0.000
12 61552 . C T 100 PASS . GT:AP 0|0:0.000,0.000 0|0:0.000,0.000 0|0:0.000,0.000
12 61604 . T G 100 PASS . GT:AP 0|0:0.000,0.000 0|0:0.000,0.000 0|0:0.000,0.000
12 61687 . G A 100 PASS . GT:AP 1|0:1.000,0.000 0|1:0.015,0.625 0|1:0.020,0.960
12 61700 . C T 100 PASS . GT:AP 0|0:0.005,0.000 0|0:0.000,0.035 0|0:0.025,0.060

```

chromosome position

Genomics in practice...

| #fileformat=VCFv4.0 ##source=BCM:SNPTools:hapfuse ##reference=1000Genomes-NCBI37 ##FORMAT=<ID=GT,Number=1>Type=String,Description="Genotype"> ##FORMAT=<ID=AP,Number=2>Type=Float,Description="Allelic Probability, P(Allele=1 Haplotype)"> | | | | | | | | | | | | | | |
|---|-------|----|-----|-----|------|--------|------|--------|-----------------|-----------------|-----------------|---------|---------|---------|
| #CHROM | POS | ID | REF | ALT | QUAL | FILTER | INFO | FORMAT | HG00096 | HG00097 | HG00099 | HG00100 | HG00101 | HG00102 |
| 12 | 60076 | . | A | C | 100 | PASS | . | GT:AP | 1 0:1.000,0.000 | 0 0:0.000,0.005 | 0 0:0.000,0.005 | | | |
| 12 | 60252 | . | A | G | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 0:0.000,0.300 | 0 1:0.010,0.590 | | | |
| 12 | 60317 | . | C | T | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 0:0.000,0.000 | 0 0:0.000,0.000 | | | |
| 12 | 60344 | . | C | A | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 0:0.020,0.025 | 0 0:0.005,0.000 | | | |
| 12 | 60383 | . | G | A | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 0:0.000,0.000 | 0 0:0.000,0.000 | | | |
| 12 | 60405 | . | T | C | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 0:0.000,0.000 | 0 0:0.000,0.000 | | | |
| 12 | 60474 | . | G | A | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 1:0.015,0.705 | 0 1:0.020,0.775 | | | |
| 12 | 60614 | . | C | A | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 0:0.000,0.005 | 0 0:0.000,0.015 | | | |
| 12 | 60628 | . | T | C | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 0:0.000,0.070 | 0 0:0.000,0.000 | | | |
| 12 | 60654 | . | G | A | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 0:0.000,0.000 | 0 0:0.000,0.000 | | | |
| 12 | 61021 | . | C | T | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 0:0.000,0.000 | 0 0:0.000,0.000 | | | |
| 12 | 61107 | . | G | T | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 0:0.000,0.015 | 0 0:0.000,0.000 | | | |
| 12 | 61172 | . | G | A | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 0:0.000,0.000 | 0 0:0.000,0.000 | | | |
| 12 | 61220 | . | G | A | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 0:0.005,0.265 | 0 1:0.020,0.665 | | | |
| 12 | 61258 | . | C | T | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 0:0.005,0.465 | 0 1:0.020,0.895 | | | |
| 12 | 61272 | . | T | C | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 0:0.000,0.000 | 0 0:0.000,0.000 | | | |
| 12 | 61329 | . | C | T | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 0:0.000,0.000 | 0 0:0.000,0.000 | | | |
| 12 | 61341 | . | G | A | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 1:0.010,0.560 | 0 1:0.020,0.855 | | | |
| 12 | 61368 | . | C | T | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 1:0.020,0.630 | 0 1:0.020,0.955 | | | |
| 12 | 61392 | . | T | A | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 0:0.000,0.000 | 0 0:0.000,0.000 | | | |
| 12 | 61405 | . | G | C | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 0:0.000,0.000 | 0 0:0.000,0.000 | | | |
| 12 | 61411 | . | C | A | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 0:0.000,0.000 | 0 0:0.000,0.015 | | | |
| 12 | 61416 | . | G | A | 100 | PASS | . | GT:AP | 0 0:0.000,0.025 | 0 0:0.000,0.010 | 0 0:0.015,0.075 | | | |
| 12 | 61422 | . | C | T | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 0:0.000,0.105 | 0 0:0.005,0.010 | | | |
| 12 | 61476 | . | C | G | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 0:0.000,0.000 | 0 0:0.005,0.000 | | | |
| 12 | 61510 | . | G | A | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 0:0.000,0.000 | 0 0:0.000,0.000 | | | |
| 12 | 61516 | . | C | T | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 0:0.000,0.005 | 0 0:0.000,0.000 | | | |
| 12 | 61552 | . | C | T | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 0:0.000,0.000 | 0 0:0.000,0.000 | | | |
| 12 | 61604 | . | T | G | 100 | PASS | . | GT:AP | 0 0:0.000,0.000 | 0 0:0.000,0.000 | 0 0:0.000,0.000 | | | |
| 12 | 61687 | . | G | A | 100 | PASS | . | GT:AP | 1 0:1.000,0.000 | 0 1:0.015,0.625 | 0 1:0.020,0.960 | | | |
| 12 | 61700 | . | C | T | 100 | PASS | . | GT:AP | 0 0:0.005,0.000 | 0 0:0.000,0.035 | 0 0:0.025,0.060 | | | |

chromosome position

SNP

Genomics in practice...

| VCF File Content | | | | | | | | | | | | | | | |
|------------------|-------|-----|----|-----|-----|------|--------|------|--------|---------|--------------|---------|--------------|---------|--------------|
| #CHROM | | POS | ID | REF | ALT | QUAL | FILTER | INFO | FORMAT | HG00096 | HG00097 | HG00099 | HG00100 | HG00101 | HG00102 |
| 12 | 60076 | . | | A | C | 100 | PASS | . | GT:AP | 1 0 | 1.000, 0.000 | 0 0 | 0.000, 0.005 | 0 0 | 0.000, 0.005 |
| 12 | 60252 | . | | A | G | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.300 | 0 1 | 0.010, 0.590 |
| 12 | 60317 | . | | C | T | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 |
| 12 | 60344 | . | | C | A | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 0 | 0.020, 0.025 | 0 0 | 0.005, 0.000 |
| 12 | 60383 | . | | G | A | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 |
| 12 | 60405 | . | | T | C | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 |
| 12 | 60474 | . | | G | A | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 1 | 0.015, 0.705 | 0 1 | 0.020, 0.775 |
| 12 | 60614 | . | | C | A | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.005 | 0 0 | 0.000, 0.015 |
| 12 | 60628 | . | | T | C | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.070 | 0 0 | 0.000, 0.000 |
| 12 | 60654 | . | | G | A | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 |
| 12 | 61021 | . | | C | T | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 |
| 12 | 61107 | . | | G | T | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.015 | 0 0 | 0.000, 0.000 |
| 12 | 61172 | . | | G | A | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 |
| 12 | 61220 | . | | G | A | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 0 | 0.005, 0.265 | 0 1 | 0.020, 0.665 |
| 12 | 61258 | . | | C | T | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 0 | 0.005, 0.465 | 0 1 | 0.020, 0.895 |
| 12 | 61272 | . | | T | C | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 |
| 12 | 61329 | . | | C | T | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 |
| 12 | 61341 | . | | G | A | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 1 | 0.010, 0.560 | 0 1 | 0.020, 0.855 |
| 12 | 61368 | . | | C | T | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 1 | 0.020, 0.630 | 0 1 | 0.020, 0.955 |
| 12 | 61392 | . | | T | A | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 |
| 12 | 61405 | . | | G | C | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 |
| 12 | 61411 | . | | C | A | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.015 |
| 12 | 61416 | . | | G | A | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.025 | 0 0 | 0.000, 0.010 | 0 0 | 0.015, 0.075 |
| 12 | 61422 | . | | C | T | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.105 | 0 0 | 0.005, 0.010 |
| 12 | 61476 | . | | C | G | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 | 0 0 | 0.005, 0.000 |
| 12 | 61510 | . | | G | A | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 |
| 12 | 61516 | . | | C | T | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.005 | 0 0 | 0.000, 0.000 |
| 12 | 61552 | . | | C | T | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 |
| 12 | 61604 | . | | T | G | 100 | PASS | . | GT:AP | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 | 0 0 | 0.000, 0.000 |
| 12 | 61687 | . | | G | A | 100 | PASS | . | GT:AP | 1 0 | 1.000, 0.000 | 0 1 | 0.015, 0.625 | 0 1 | 0.020, 0.960 |
| 12 | 61700 | . | | C | T | 100 | PASS | . | GT:AP | 0 0 | 0.005, 0.000 | 0 0 | 0.000, 0.035 | 0 0 | 0.025, 0.060 |

chromosome position

SNP

genotype ind. 1 genotype ind. 2 genotype ind. 3 ...