

Archaic human introgression

Population Genetics 2025

Martin Petr

mp@bodkan.net

February 26, 2025

1856: Discovery of a ‘new man’



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



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

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



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

Neander's valley

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

Neander's valley
valley = Thal in German

1856: Discovery of a ‘new man’



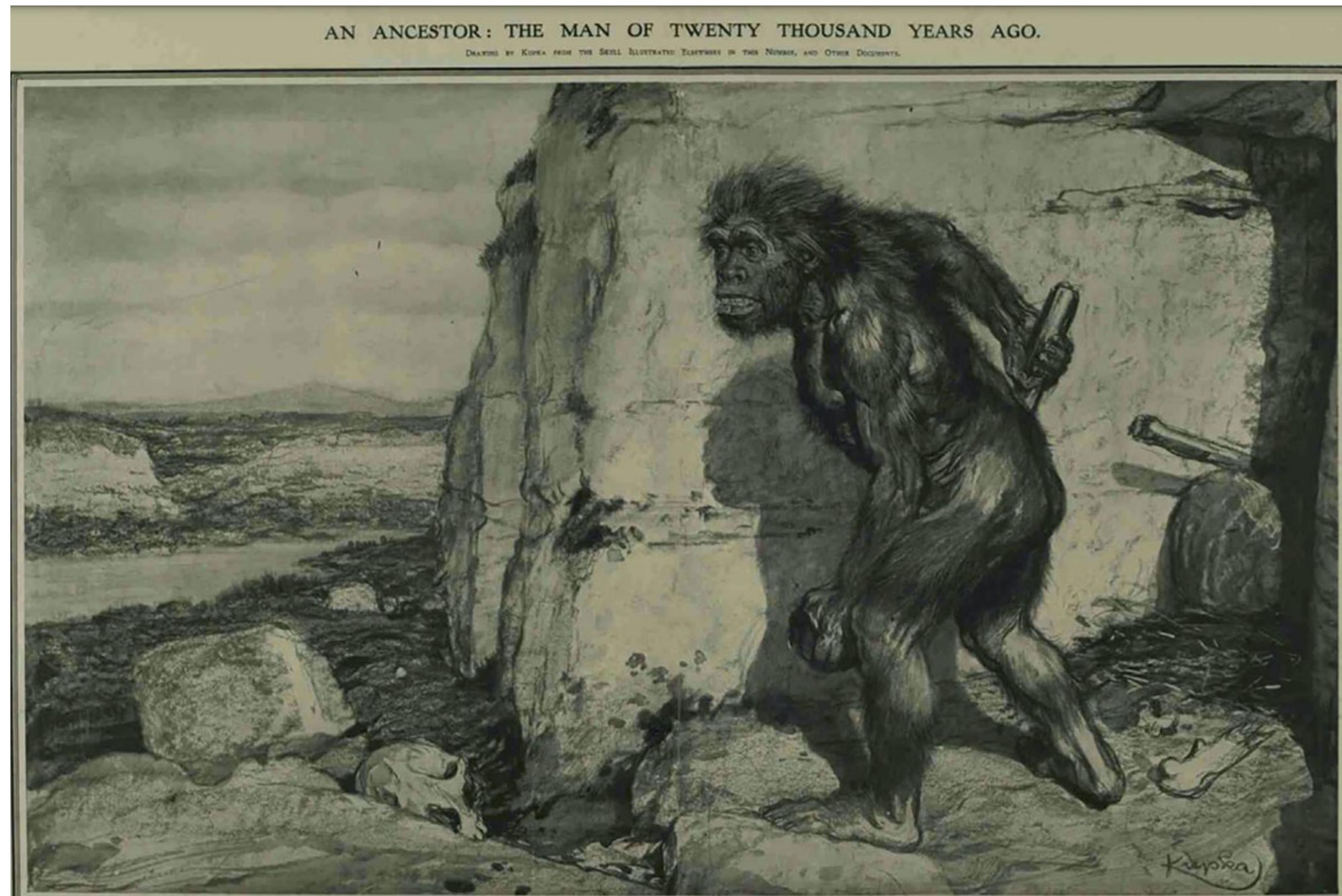
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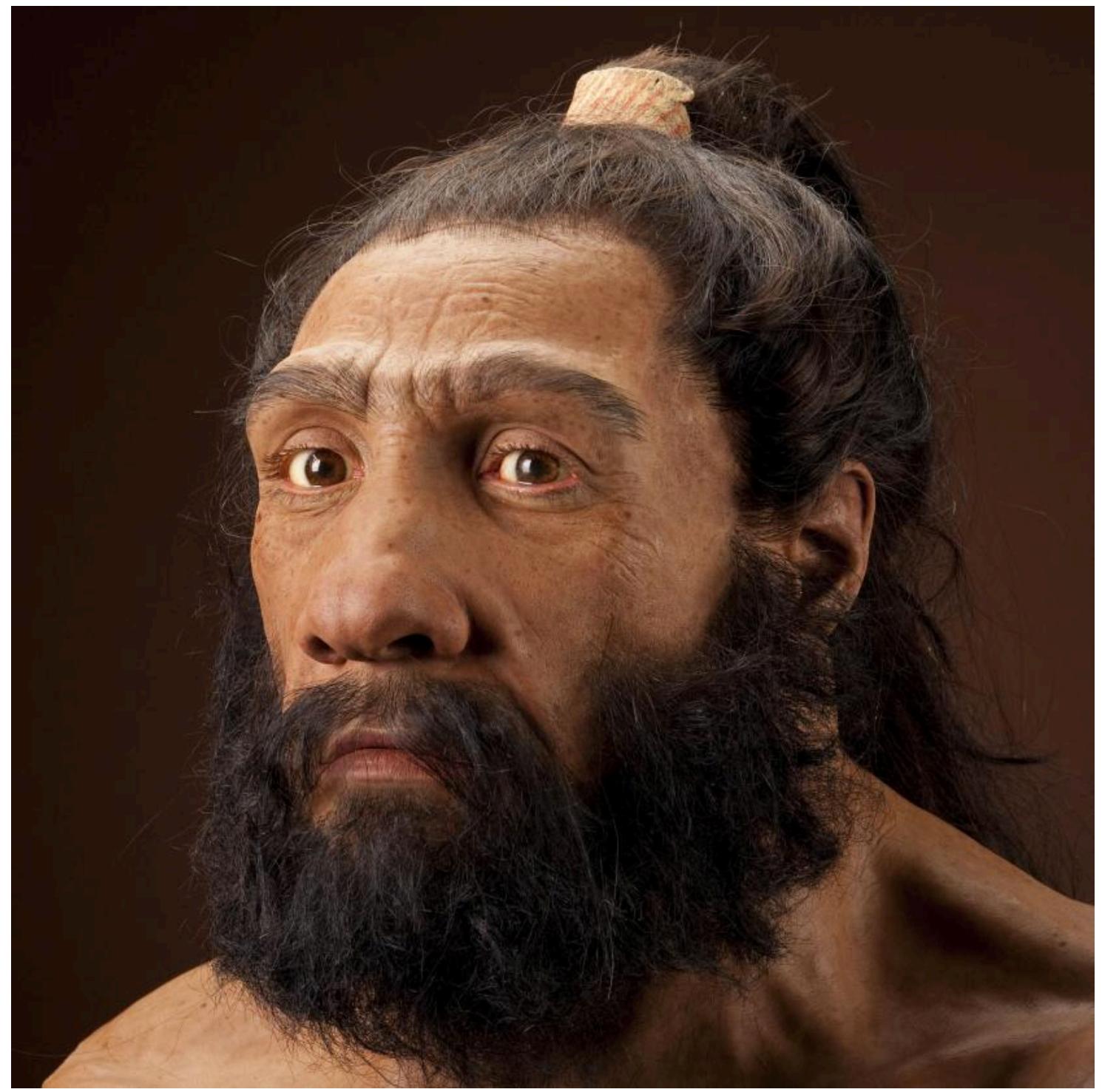
Neanderthal



The Illustrated London News, 1909



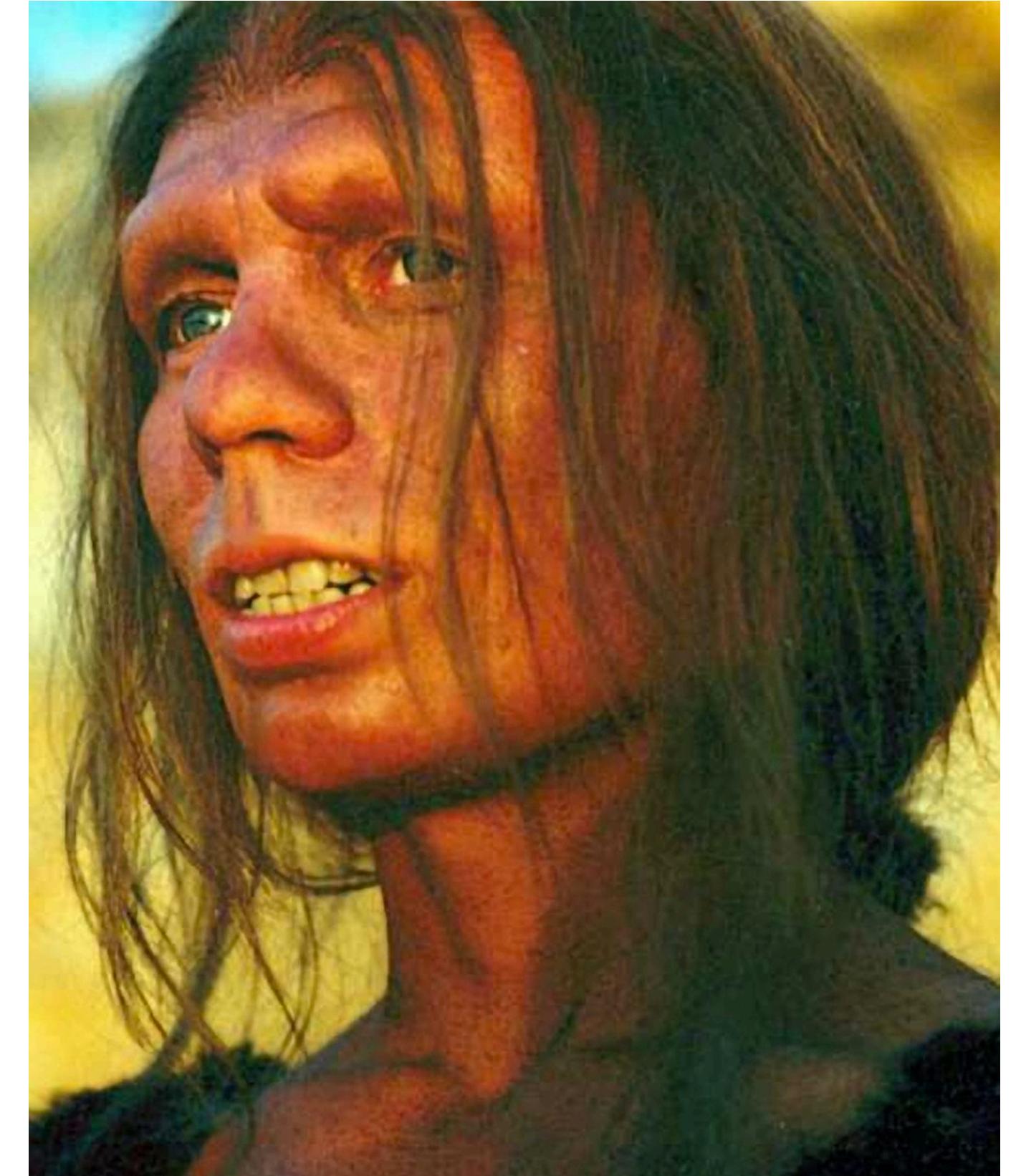
The Neanderthal Man movie poster, 1953



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



<https://www.neanderthal.de>



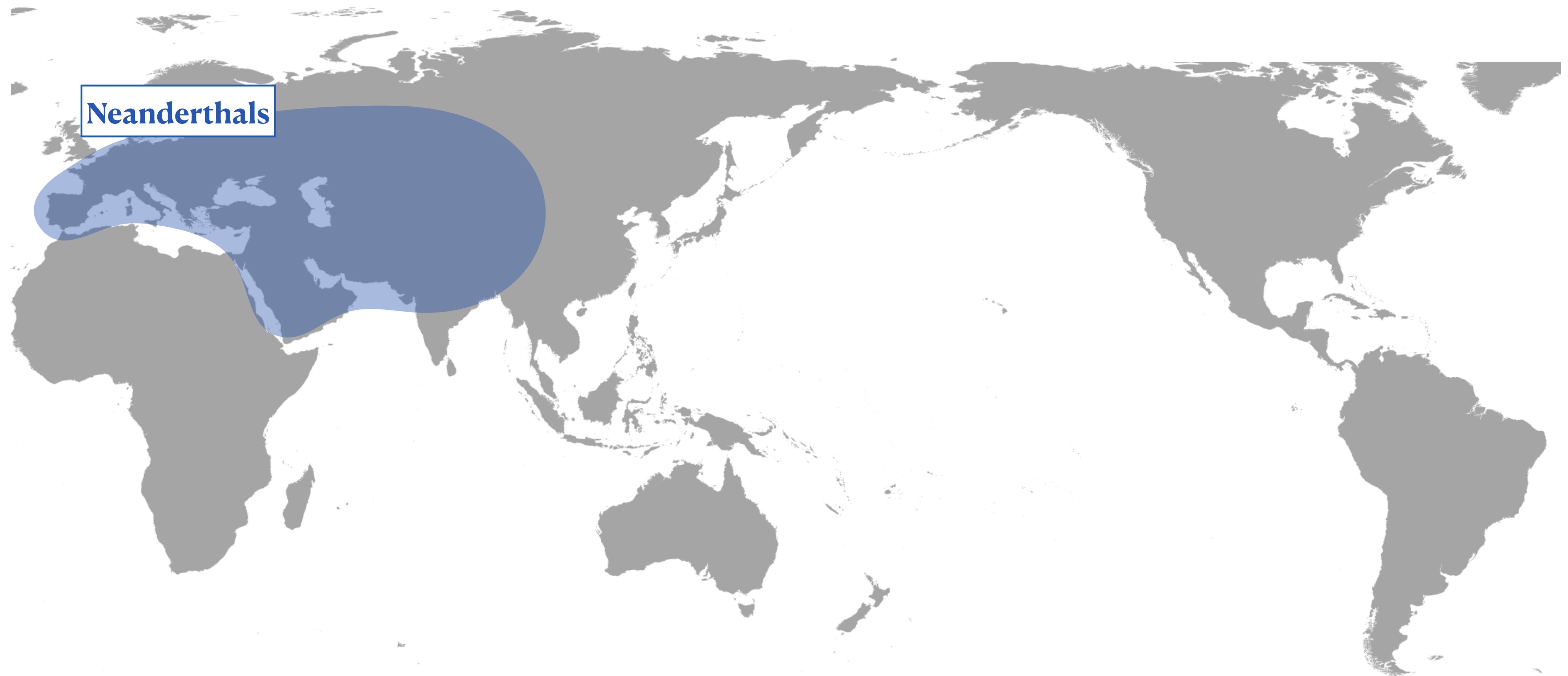
<https://journals.plos.org/plosbiology/article?id=10.1371/journal.pbio.0020449>



Neanderthal Museum, Mettmann, Germany

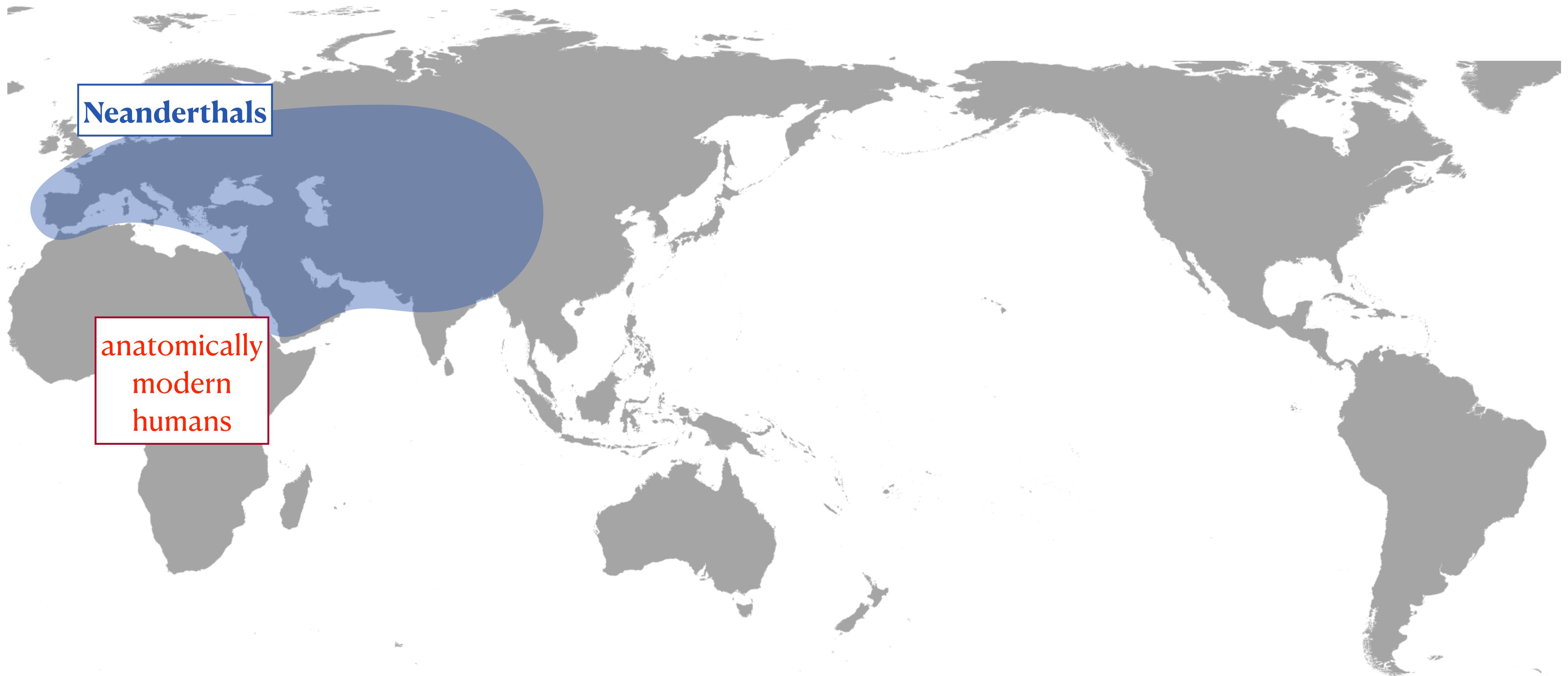


~600 kya B.P.: Neanderthals in Eurasia



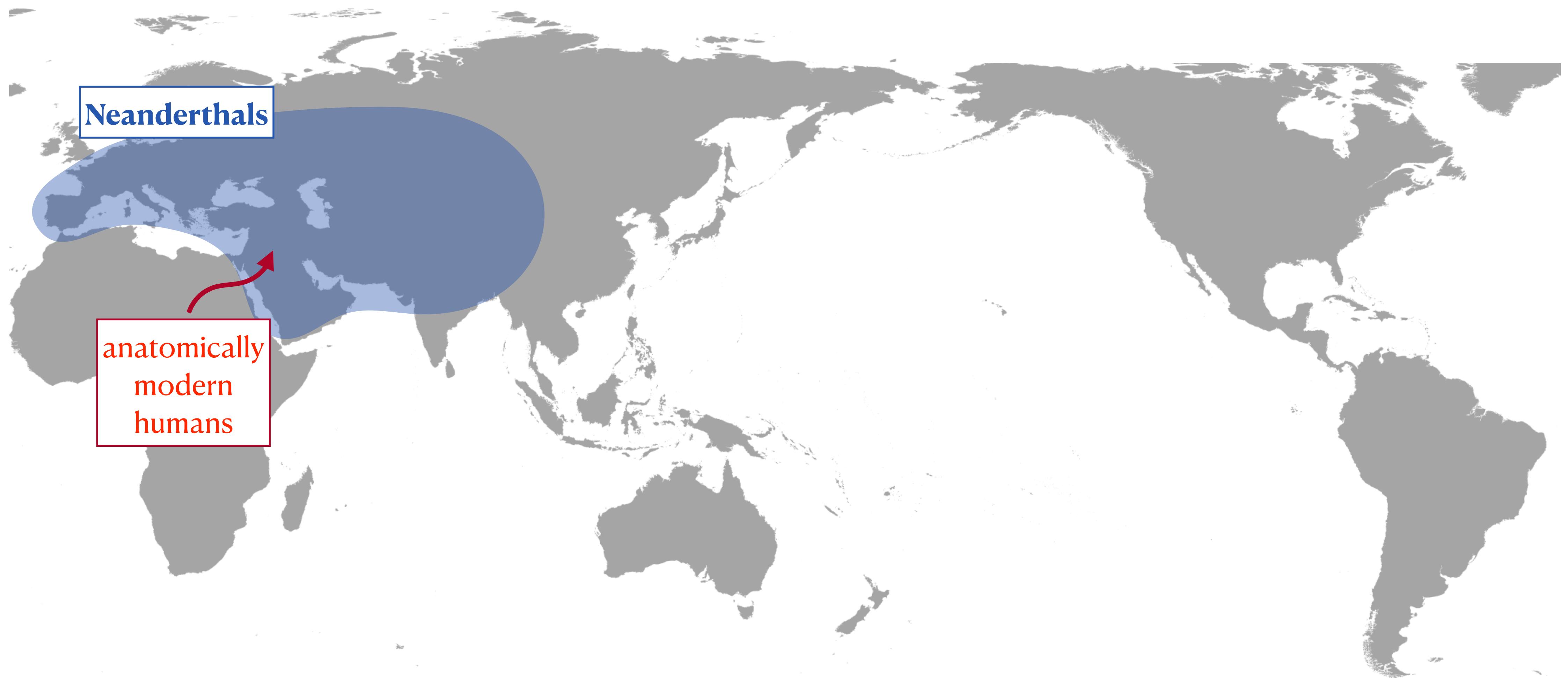
B.P. = "before present"

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



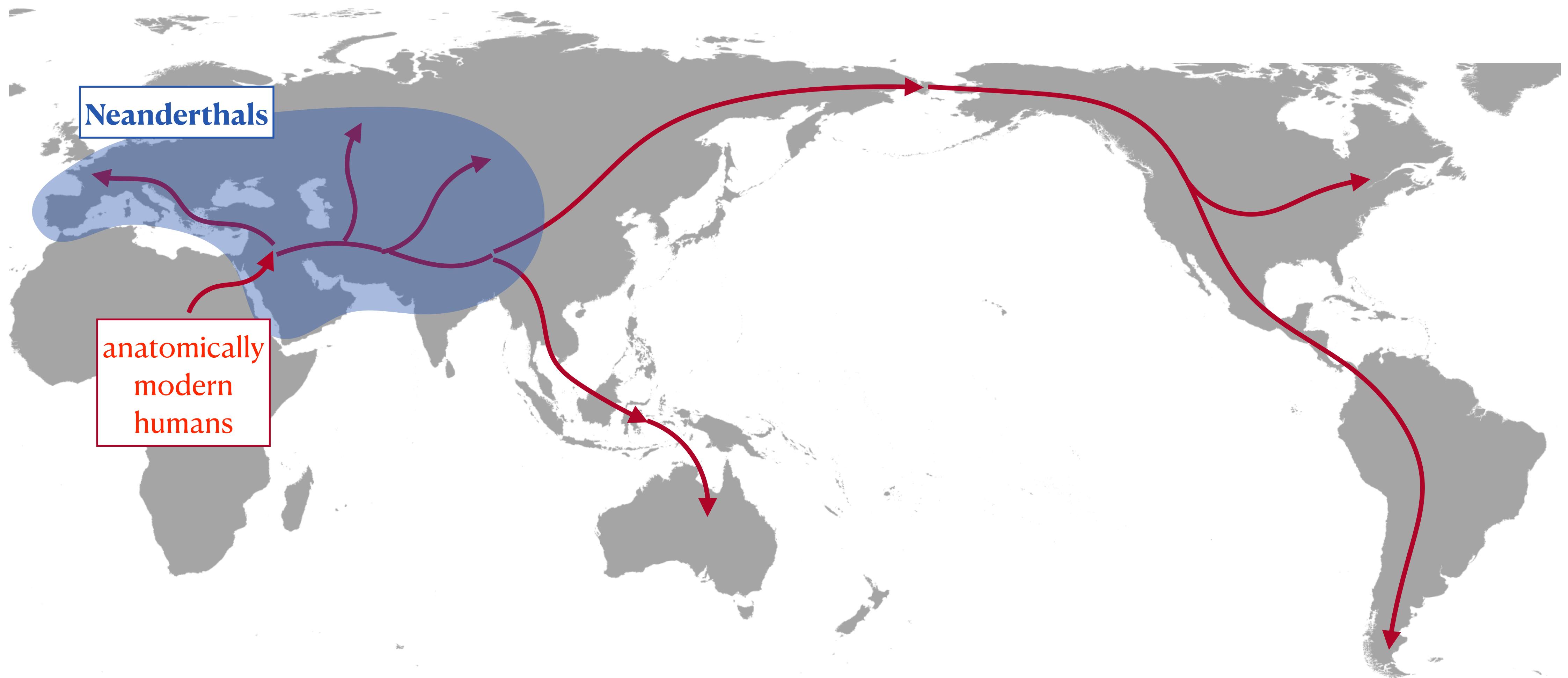
B.P. = "before present"

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



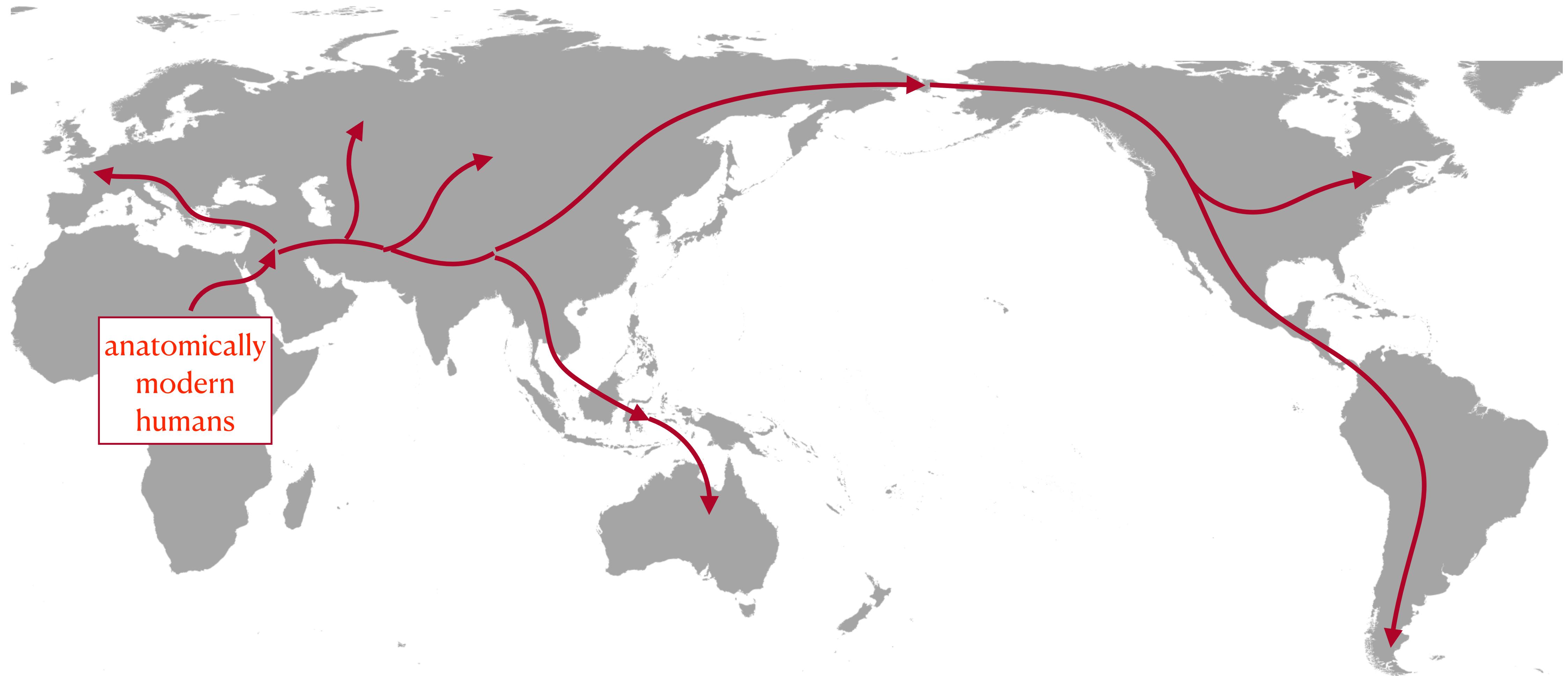
B.P. = "before present"

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



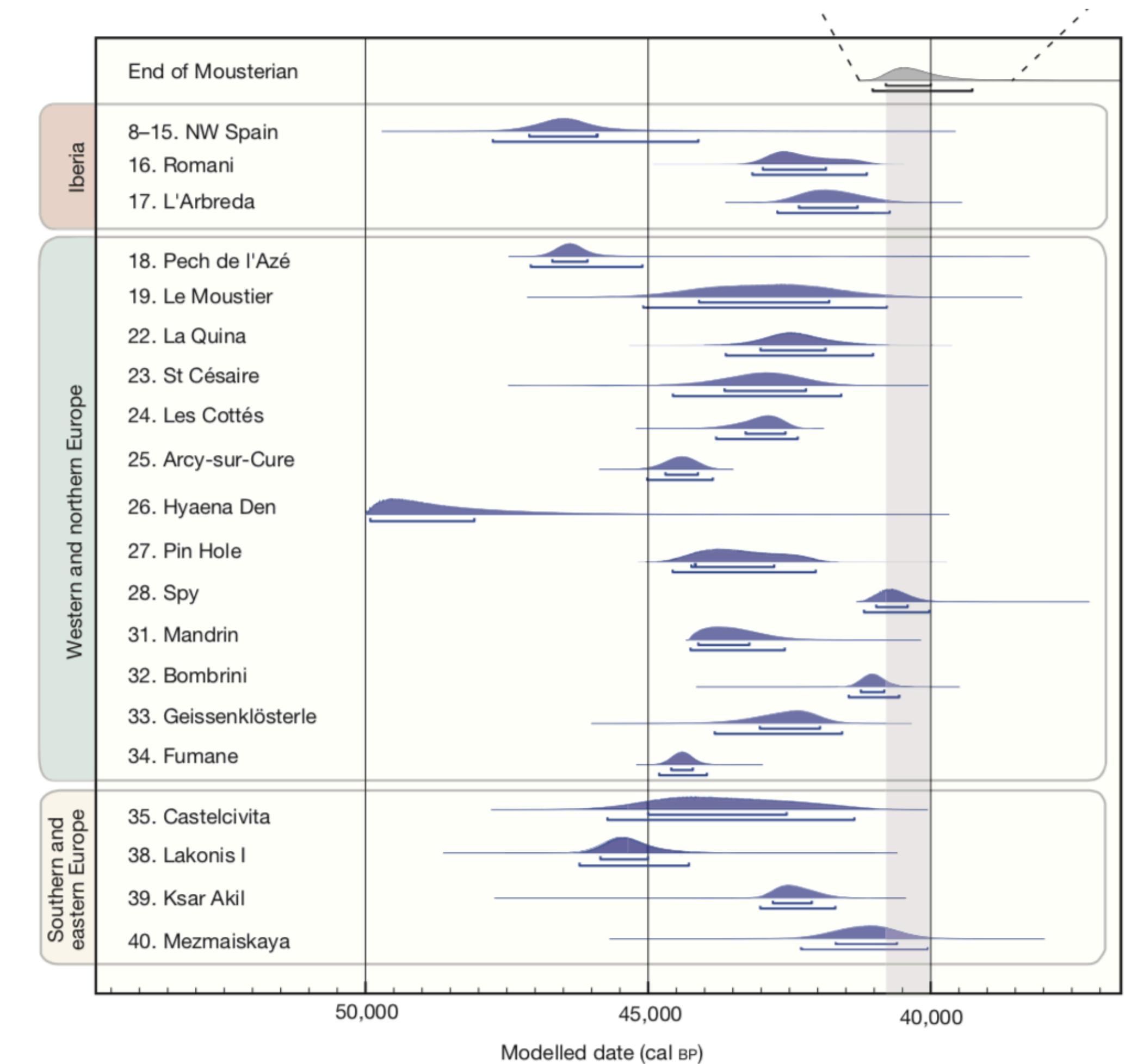
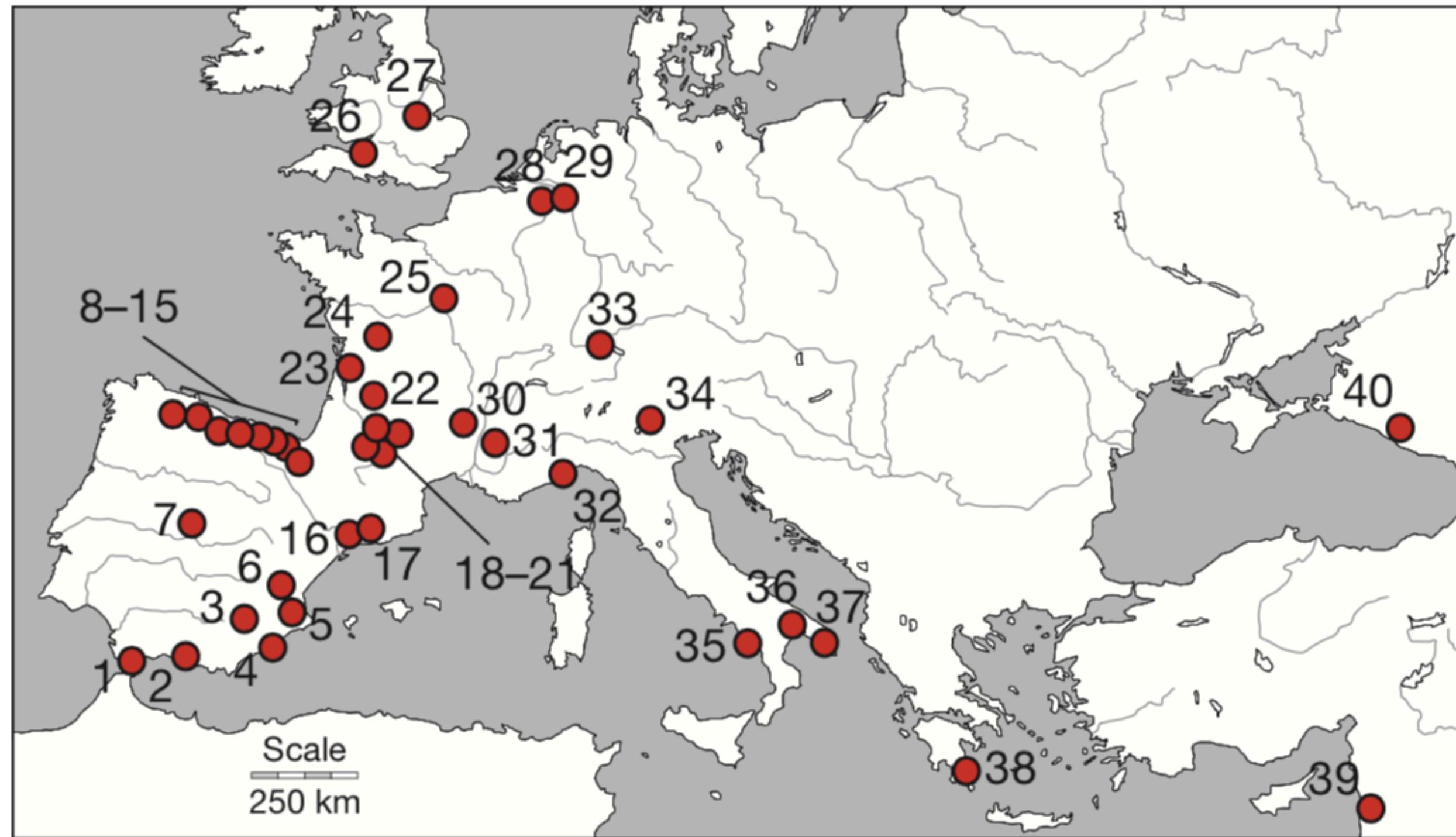
B.P. = "before present"

\sim 40 kya: Neanderthals vanished

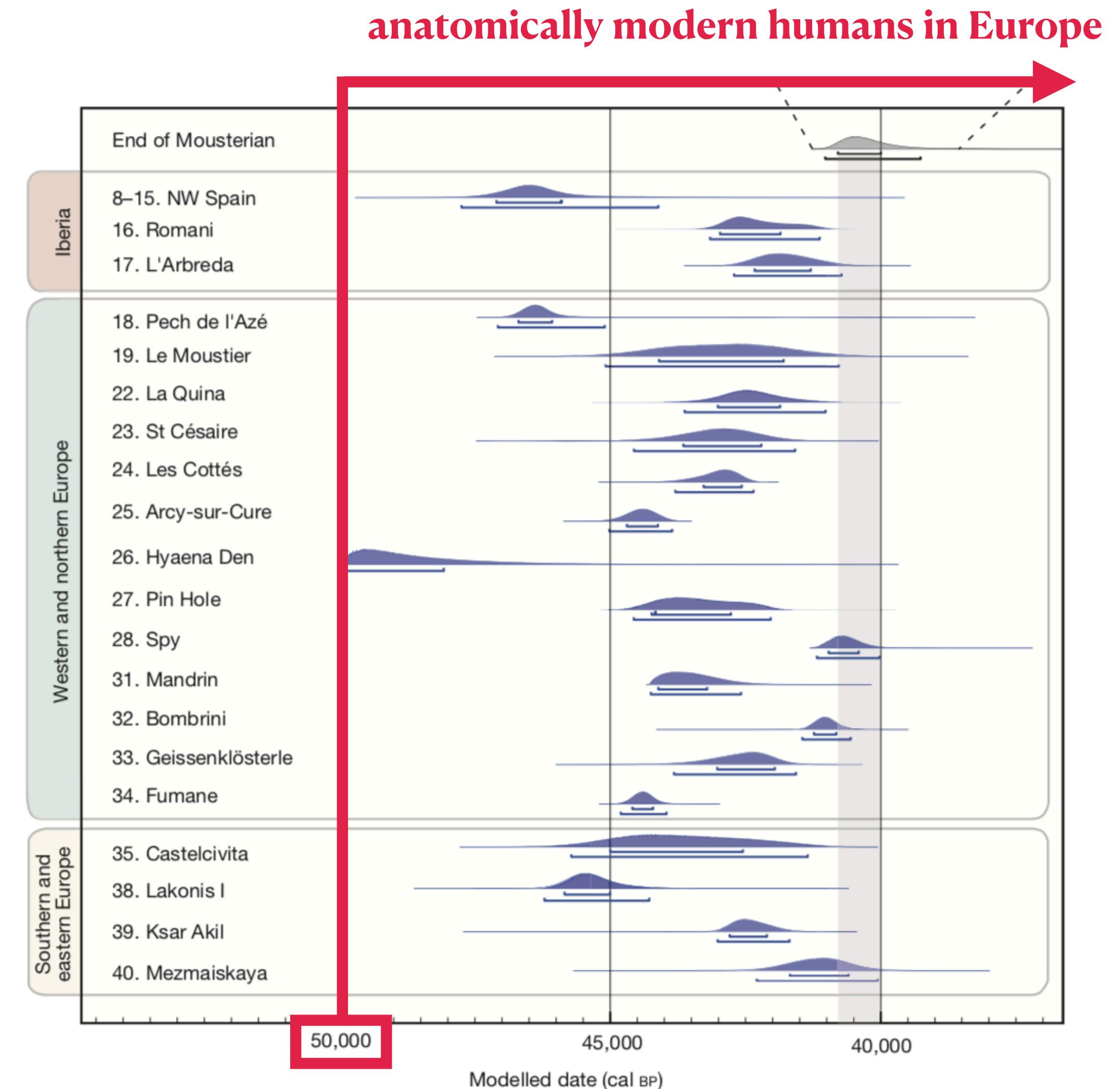
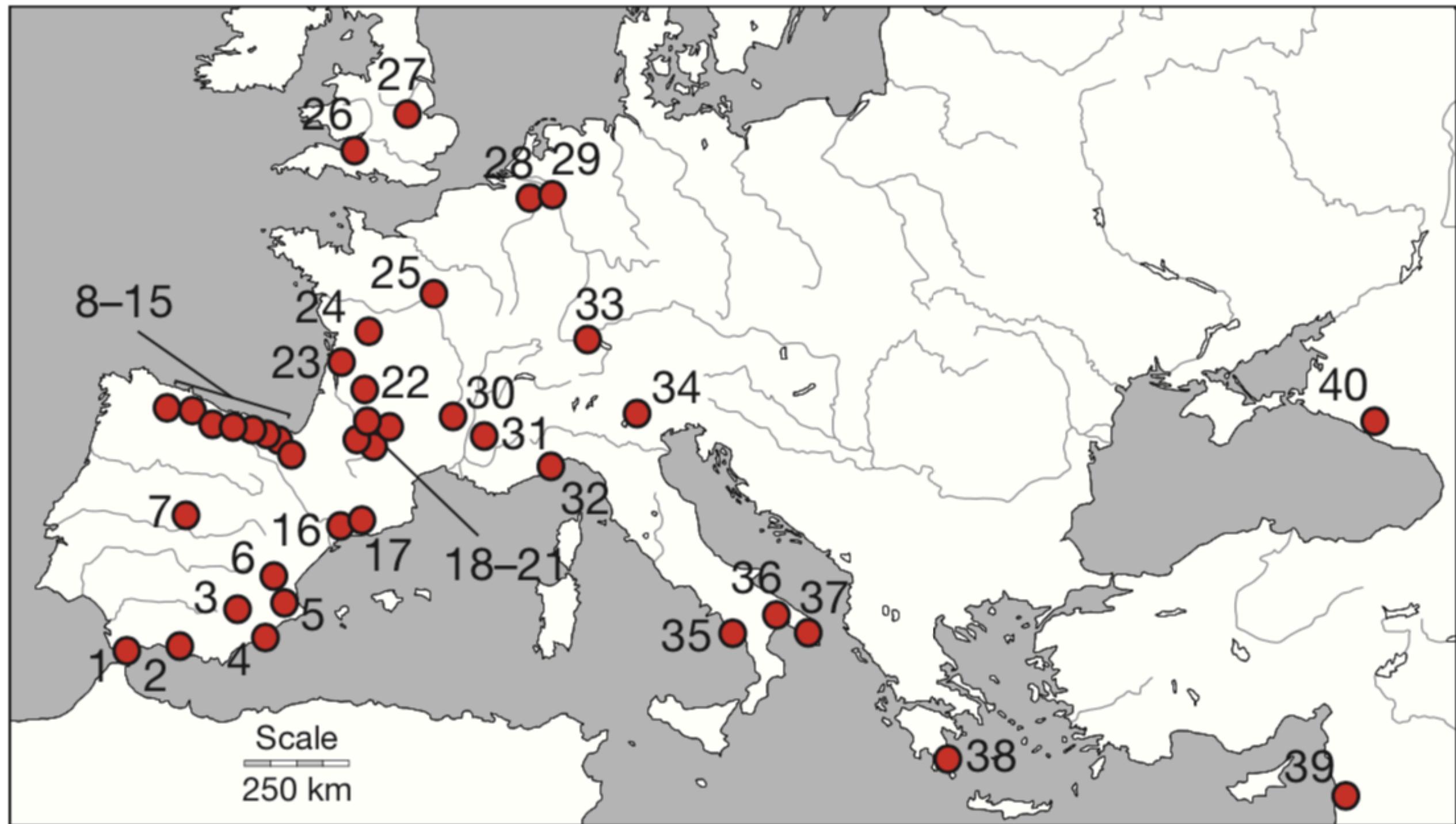


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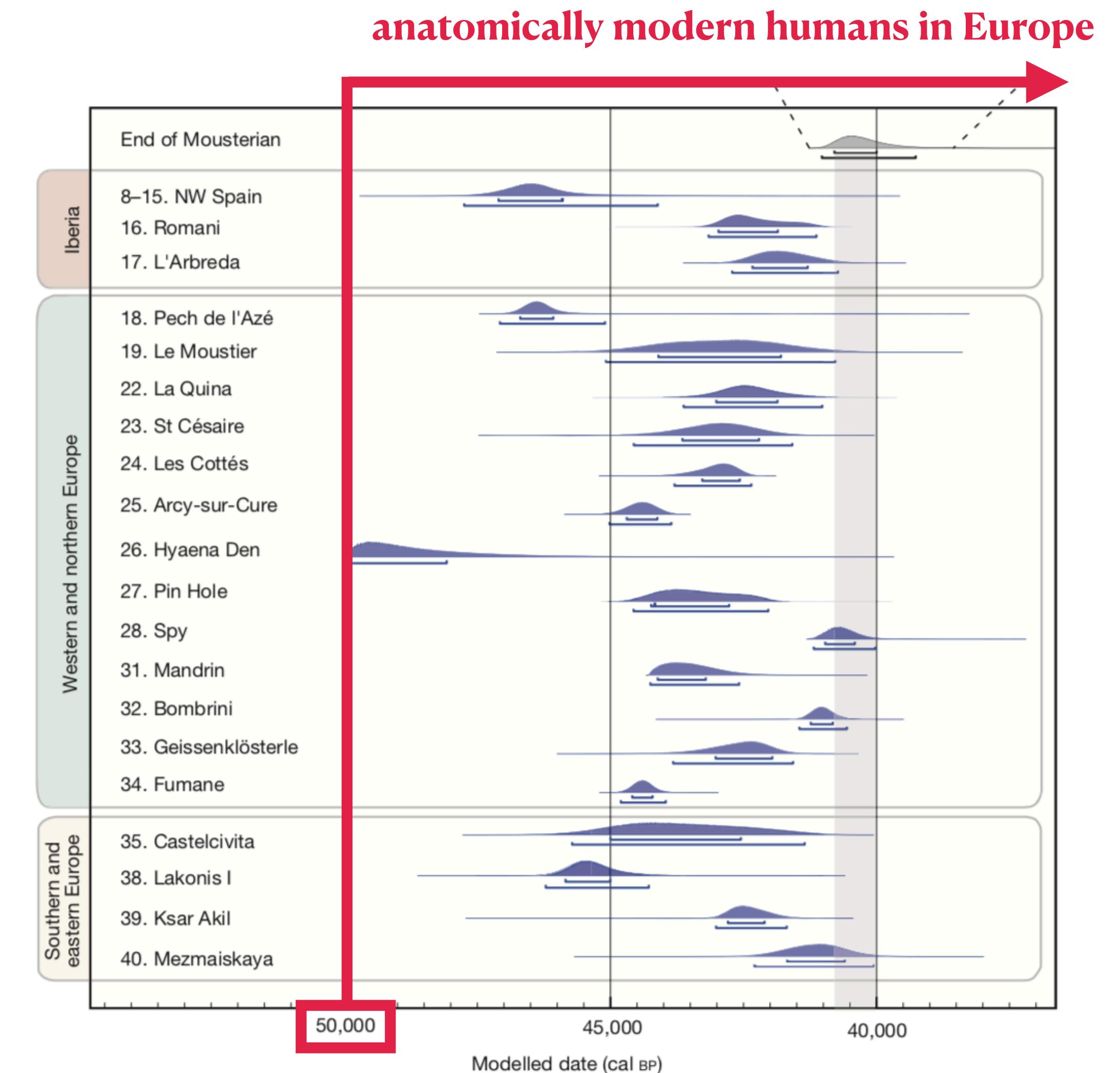
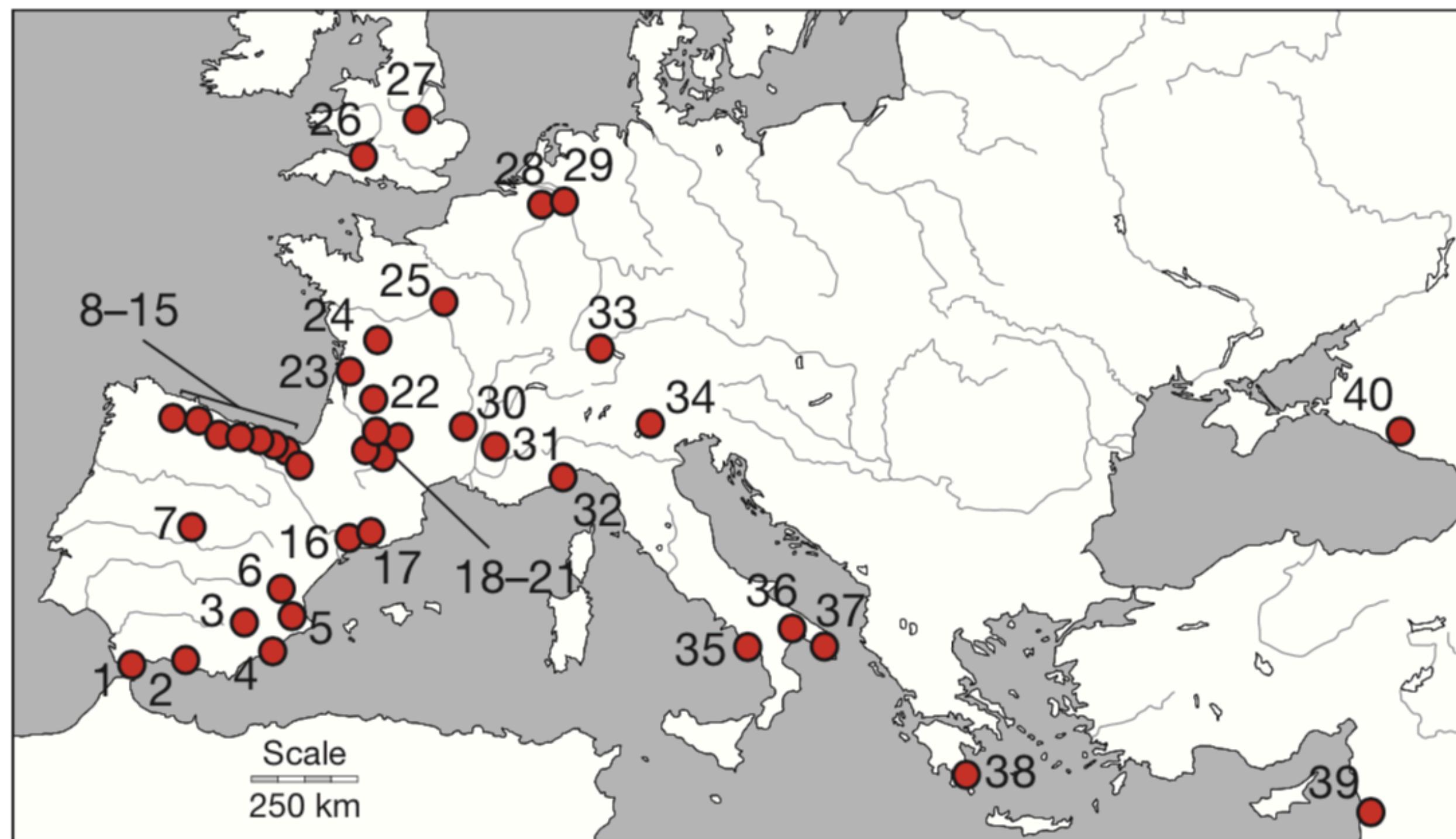
~40 kya: Neanderthals vanished



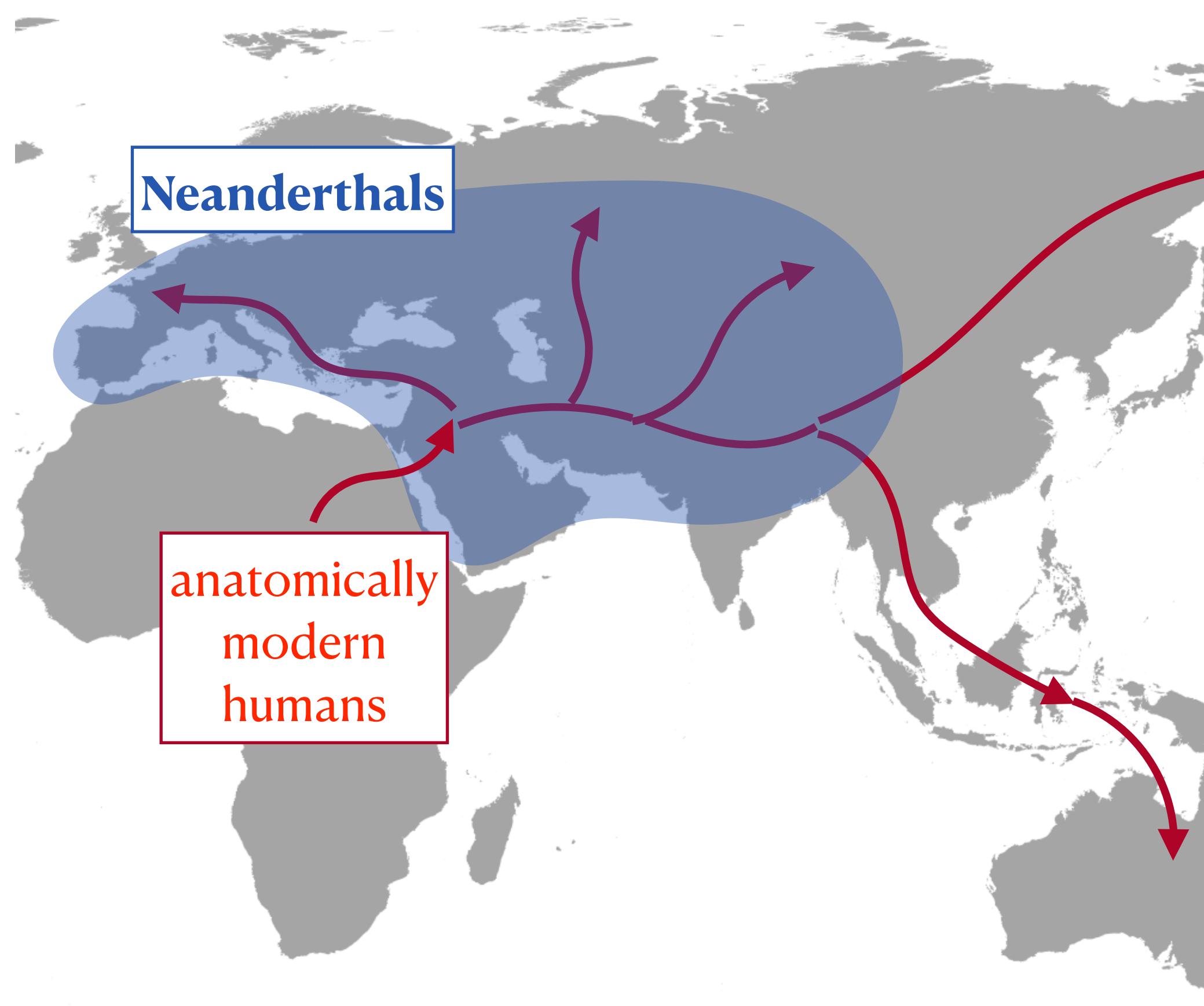
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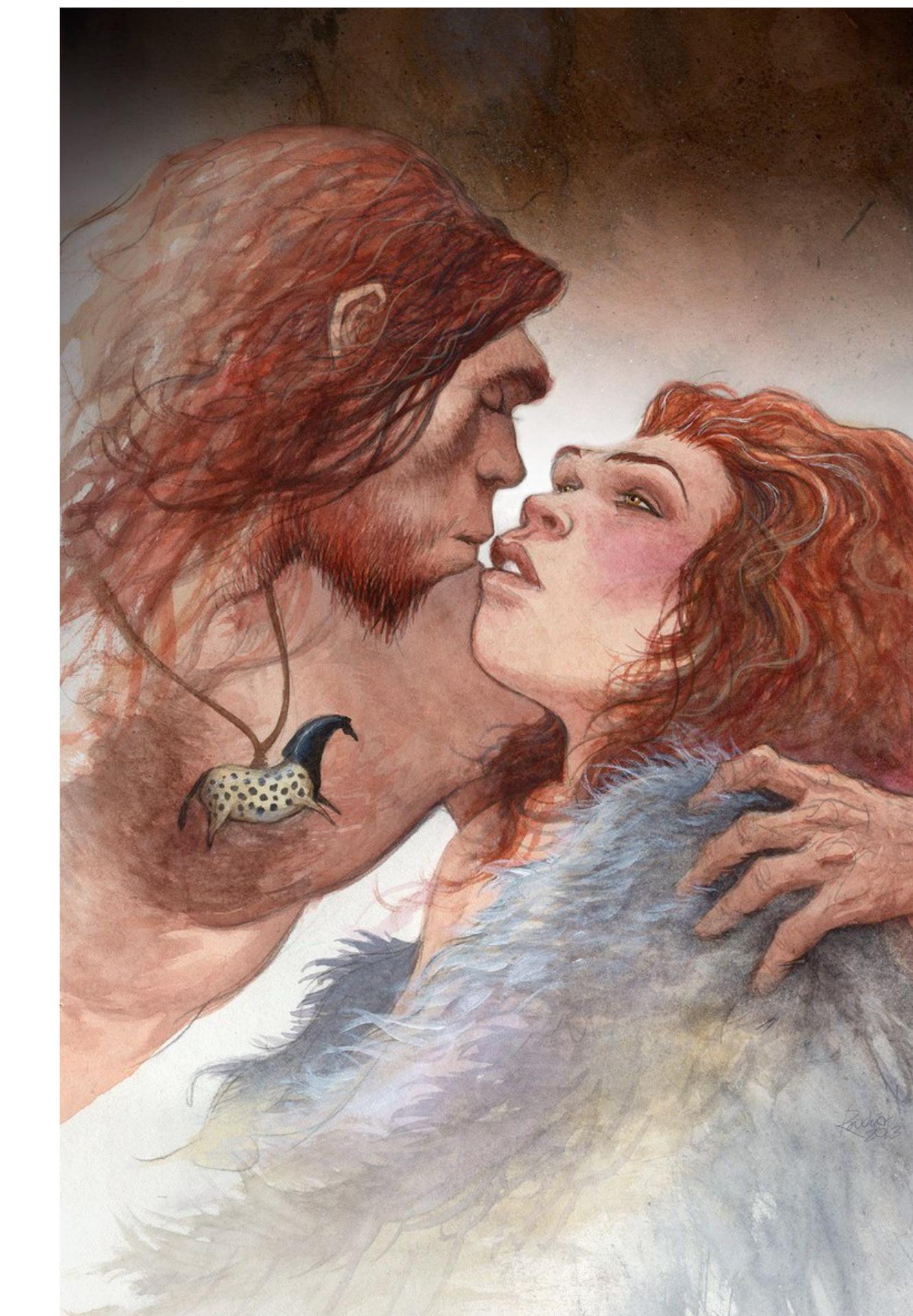
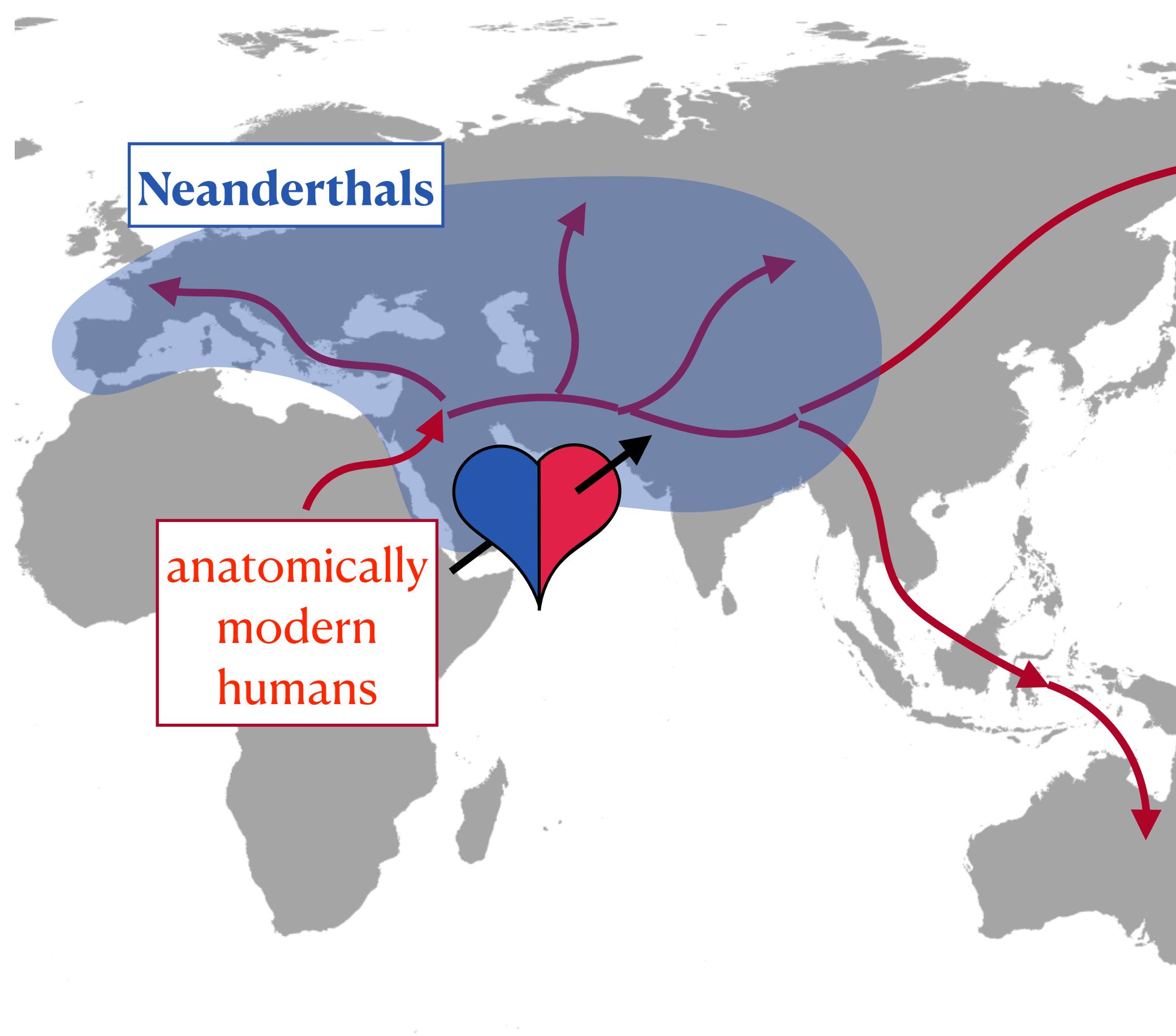
What happened in the few thousand years of overlap?



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)



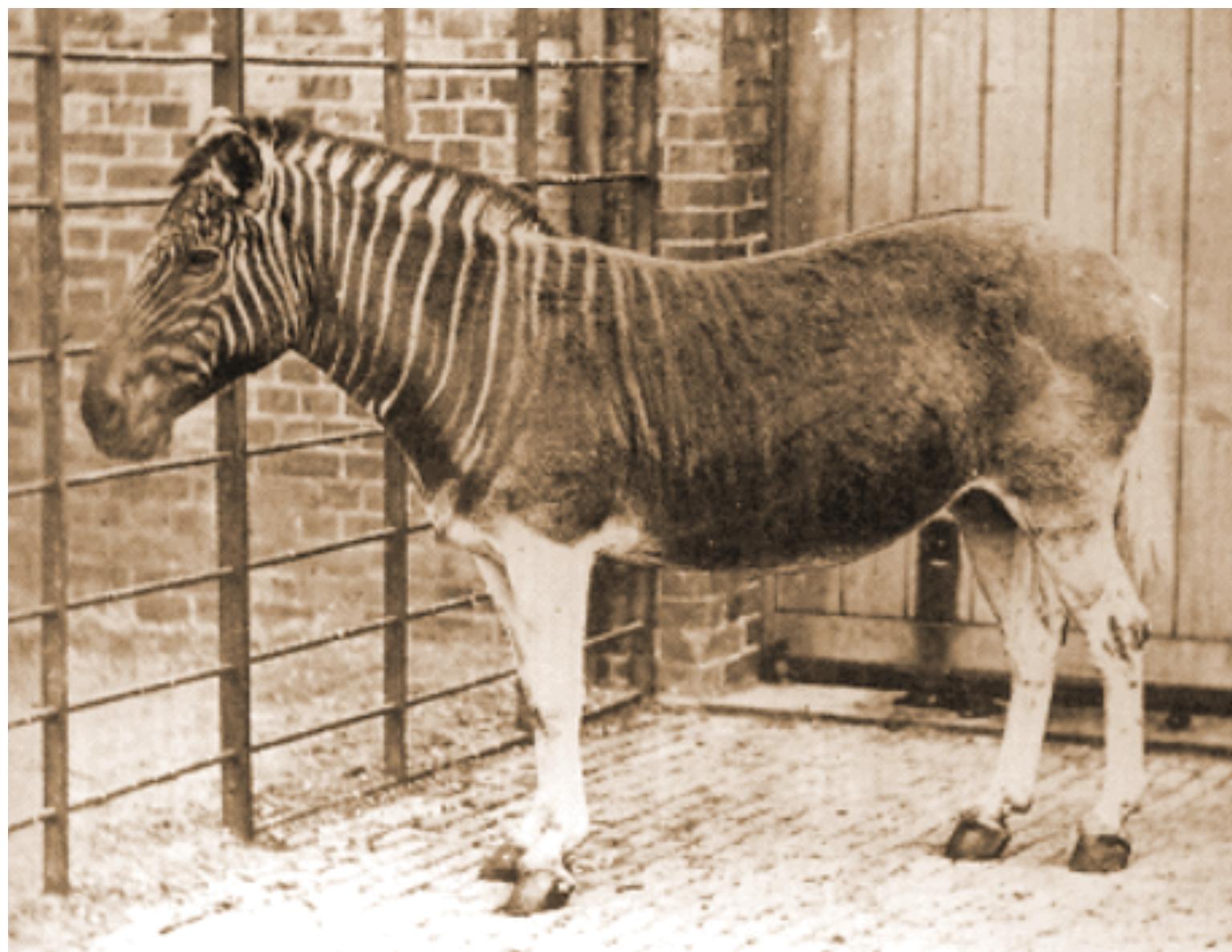
1984

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

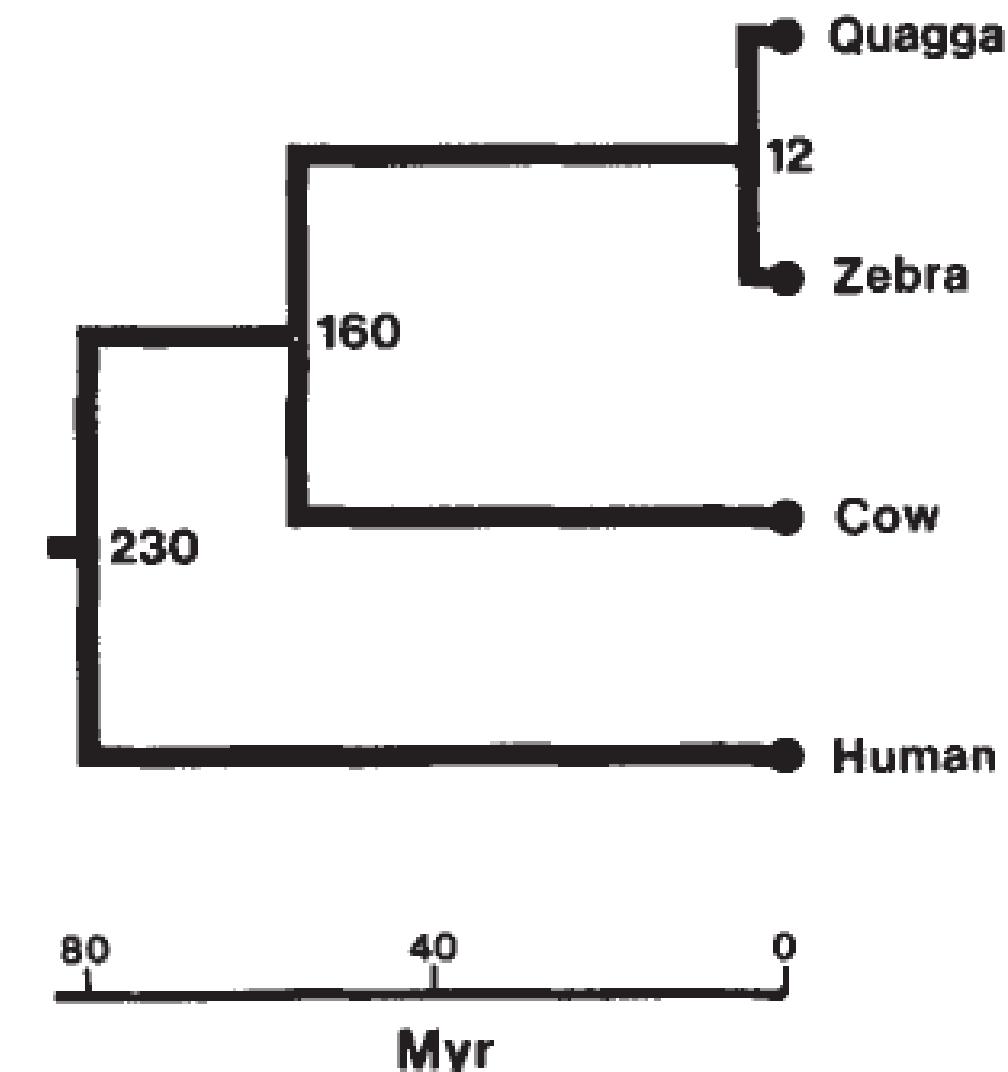
Russell Higuchi*, Barbara Bowman*, Mary Freiberger*,
Oliver A. Ryder† & Allan C. Wilson*

* 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



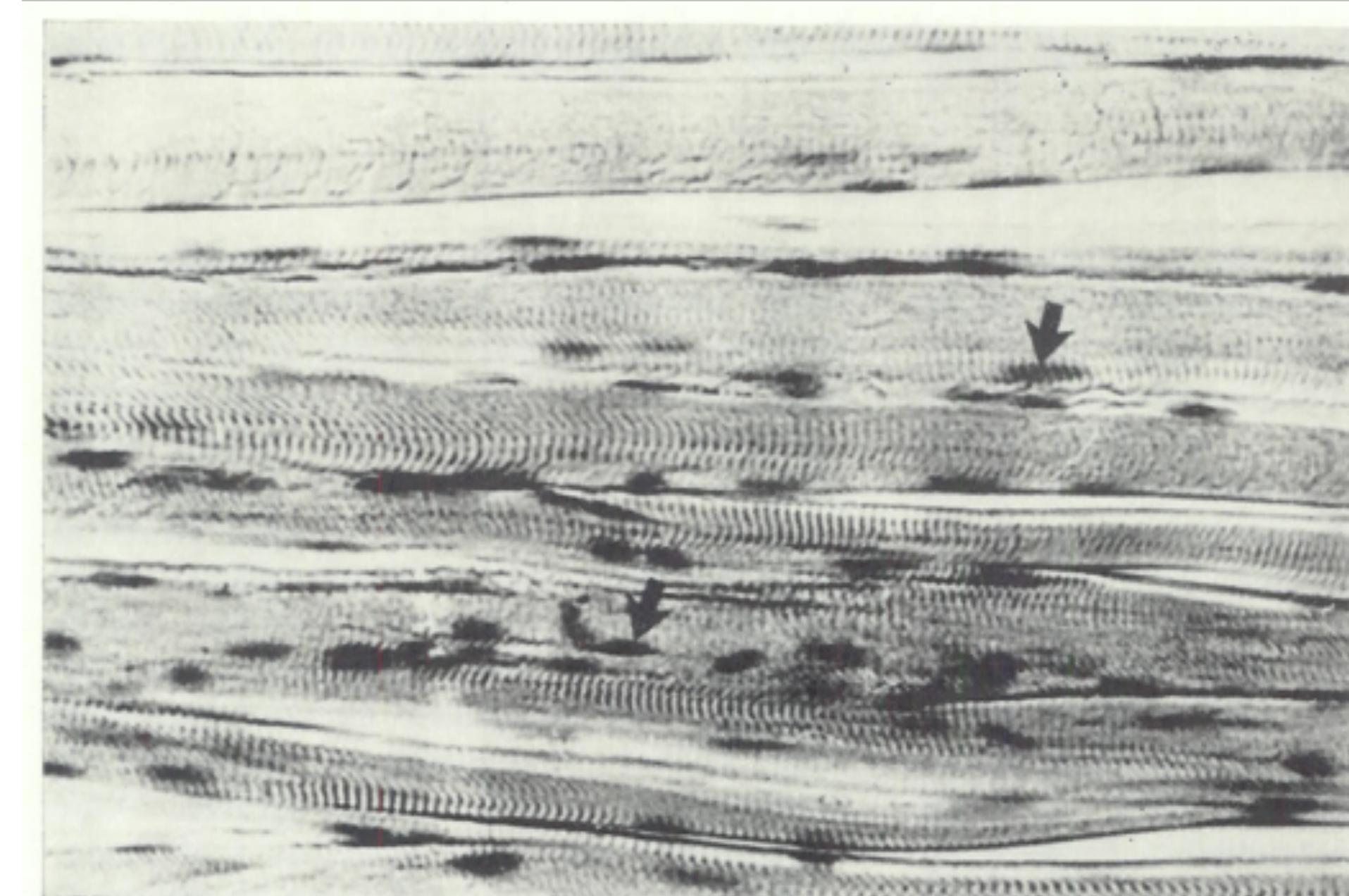
1985

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

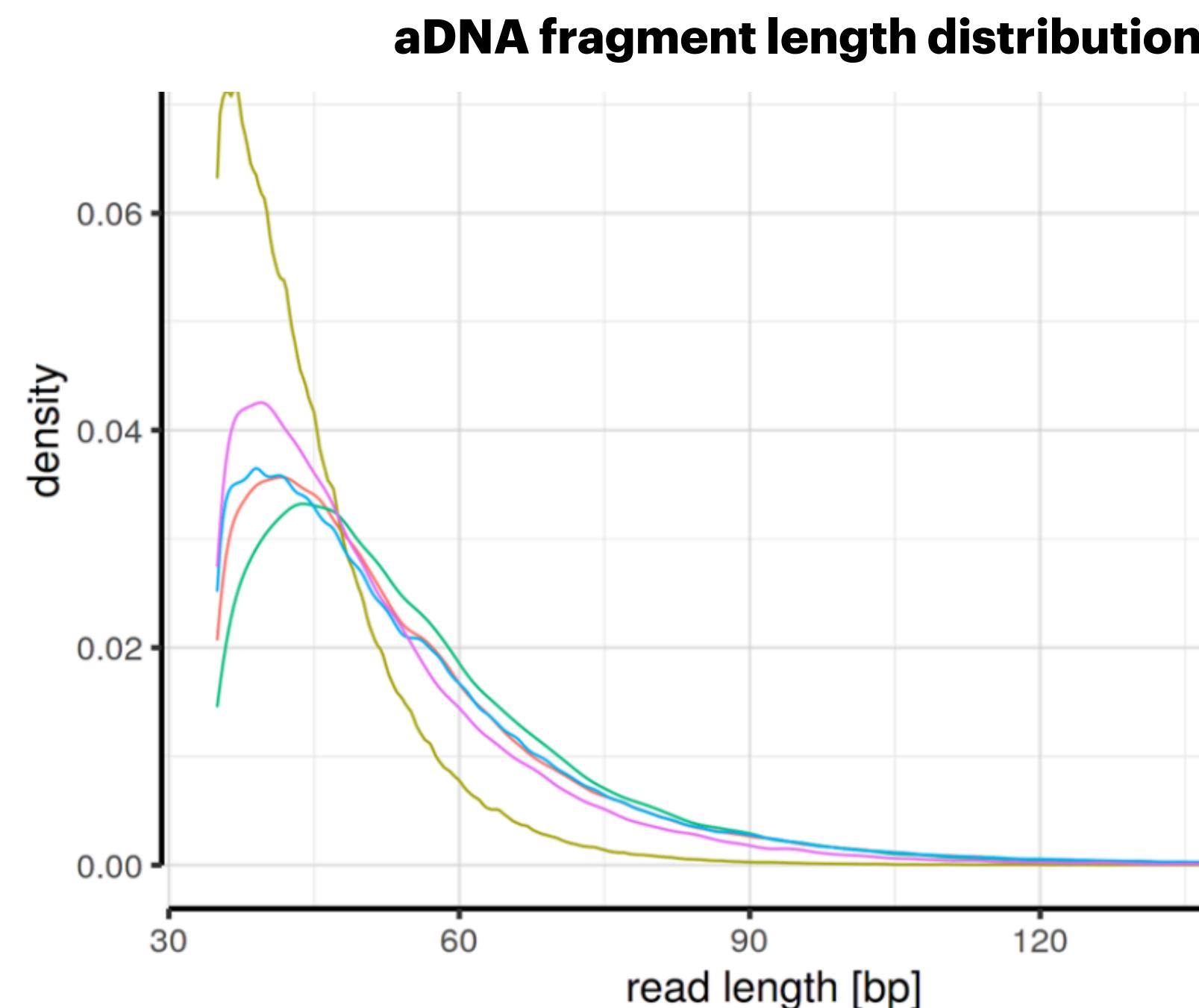
- ~2400 years old mummy
- 3400 bp nuclear DNA



Molecular characteristics of aDNA

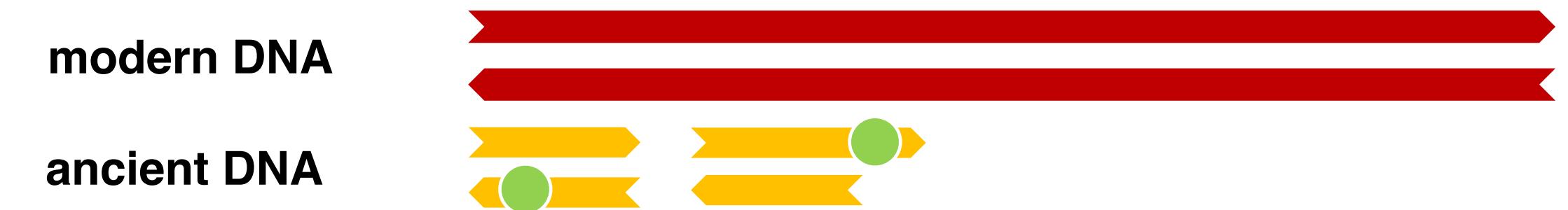
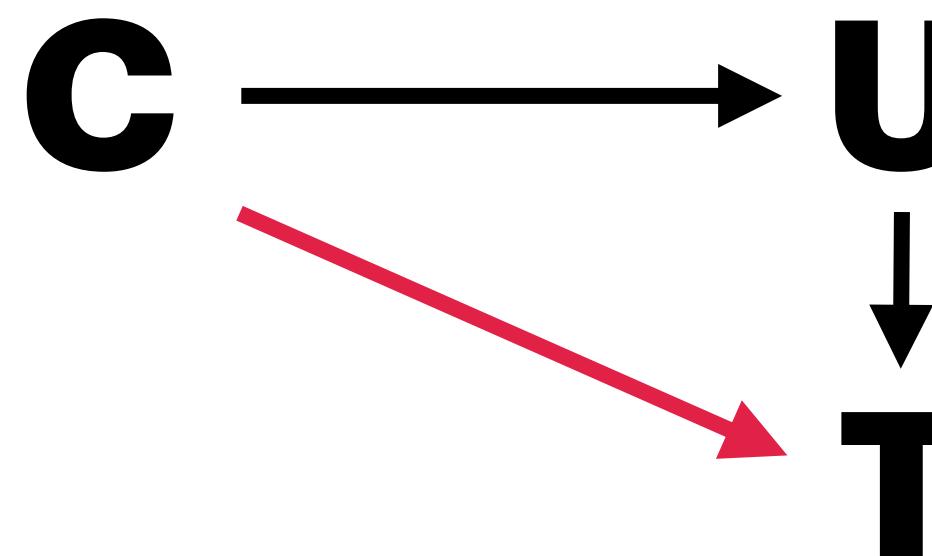
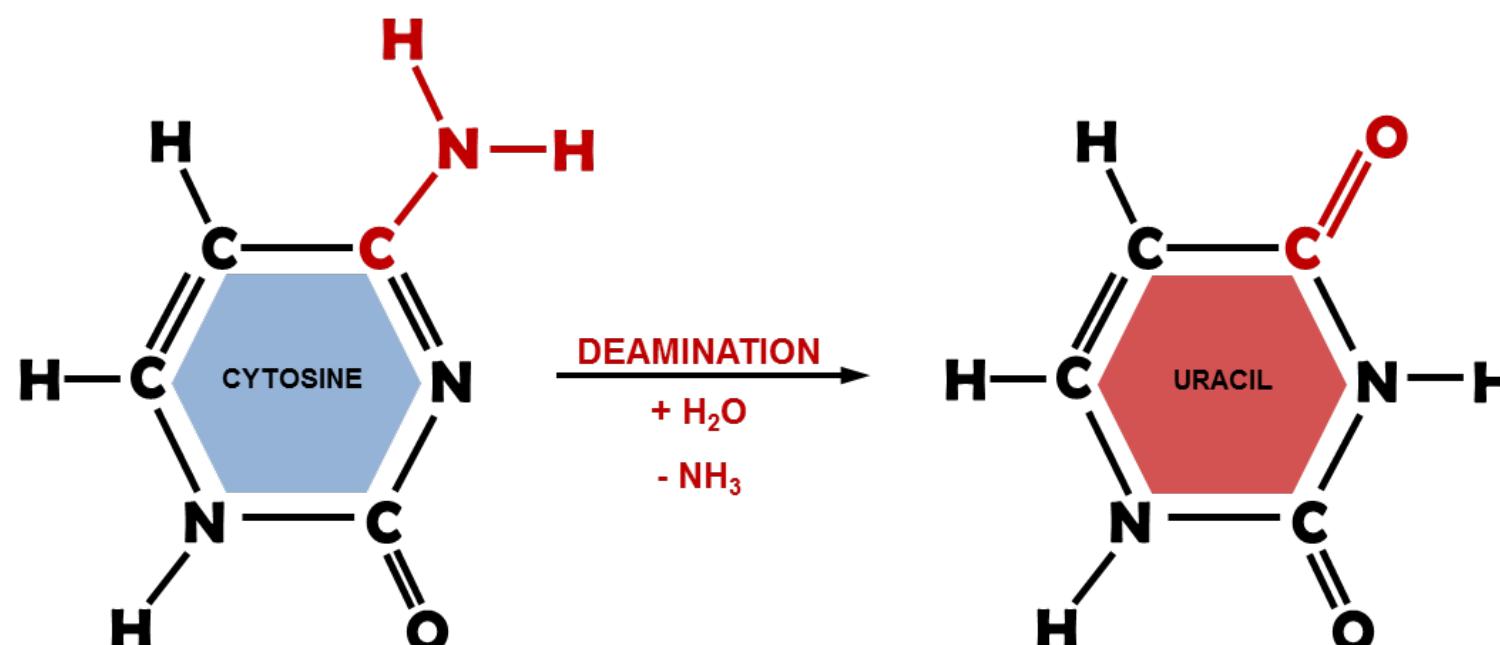
Molecular characteristics of aDNA

- highly fragmented

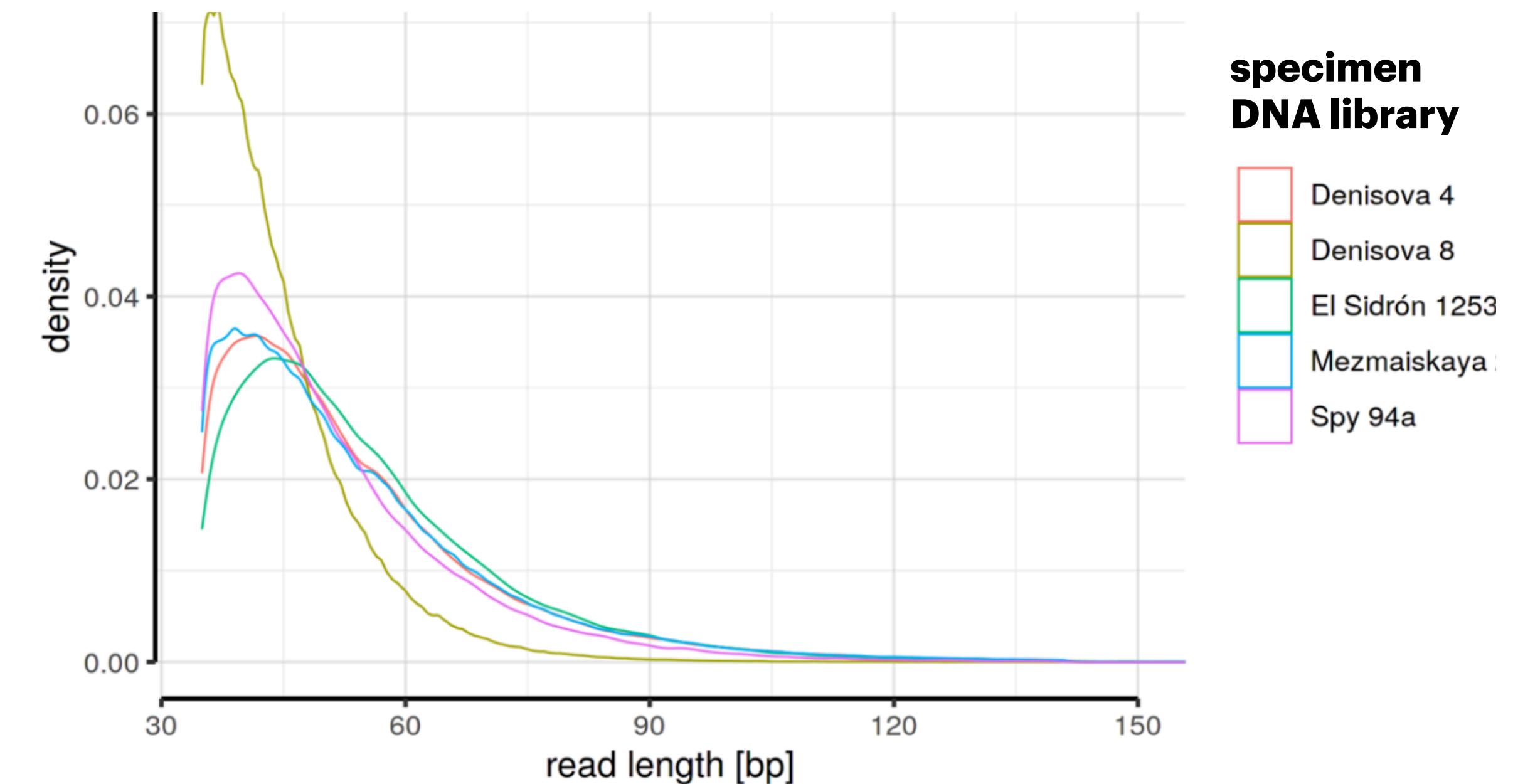


Molecular characteristics of aDNA

- highly fragmented
- post-mortem chemical modifications

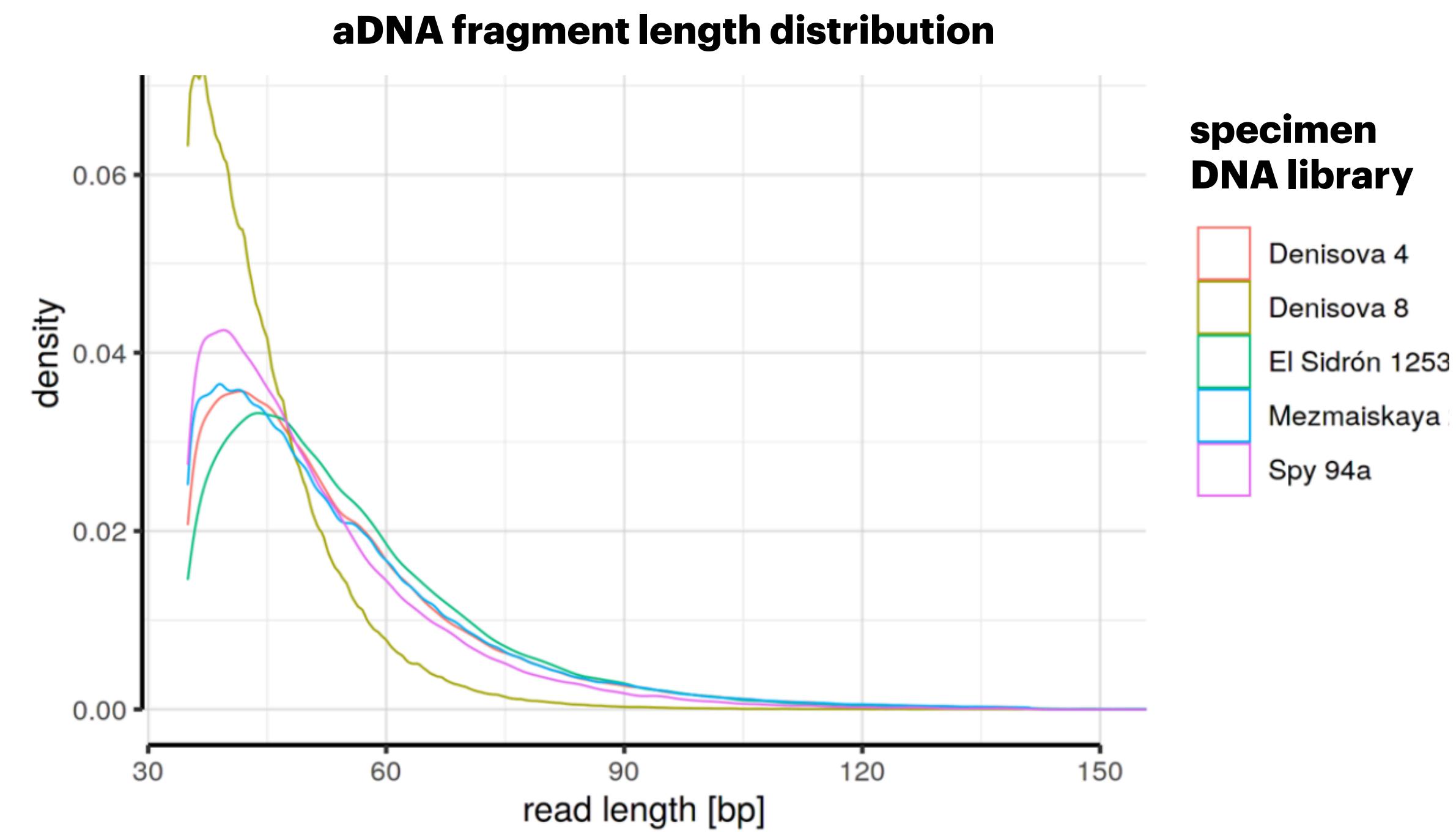
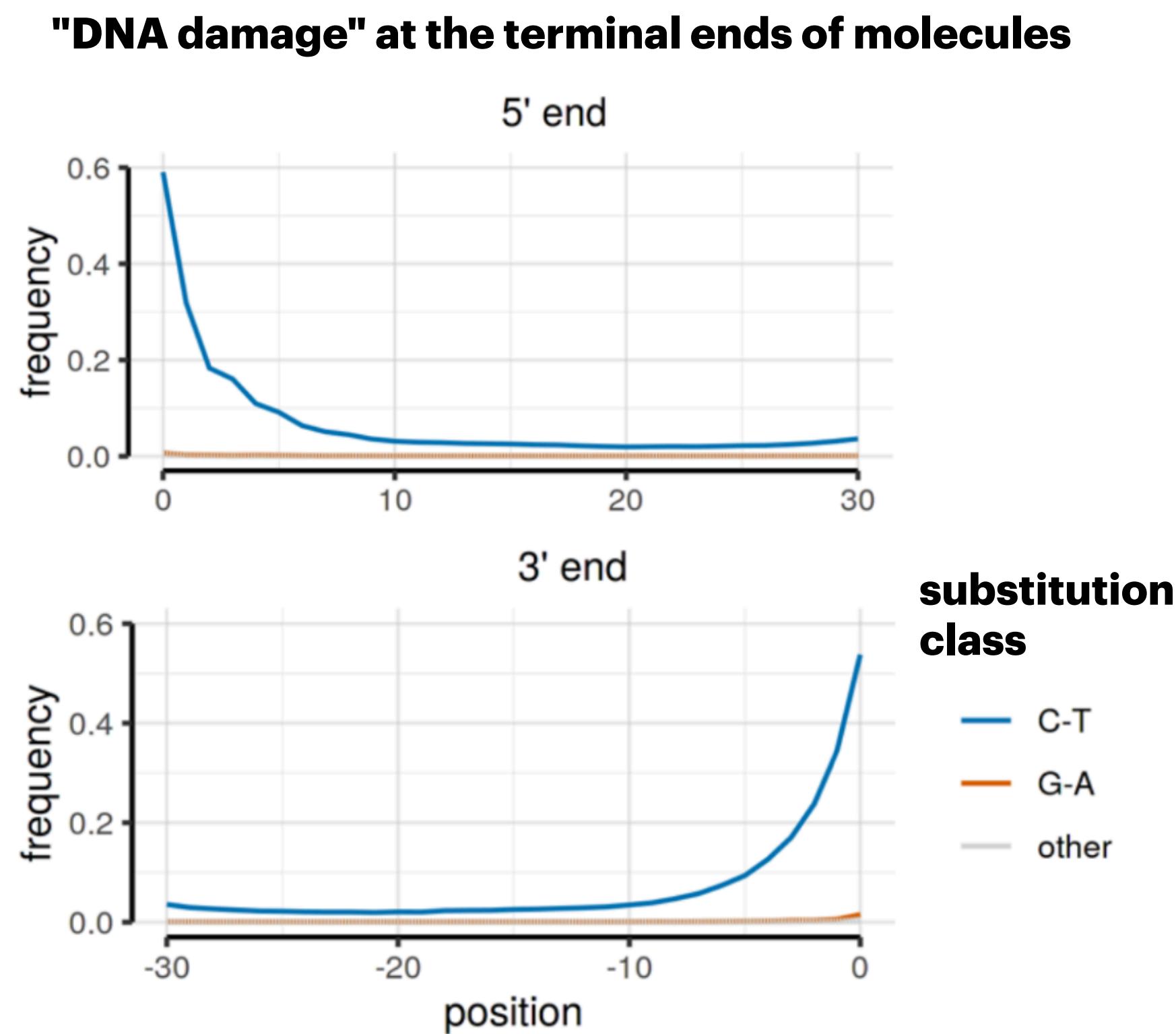
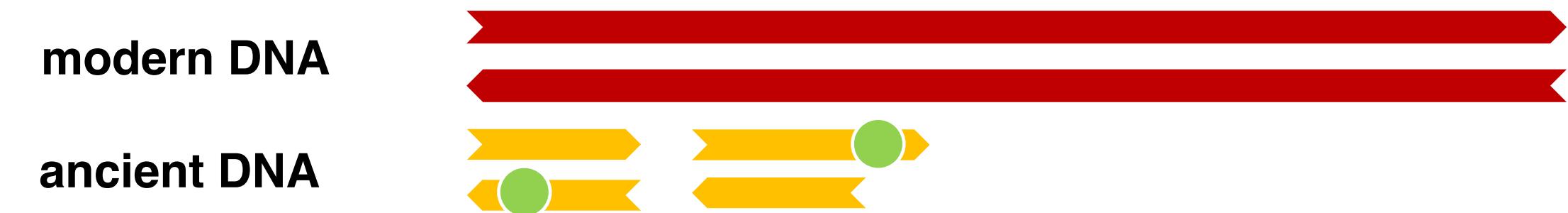


aDNA fragment length distribution



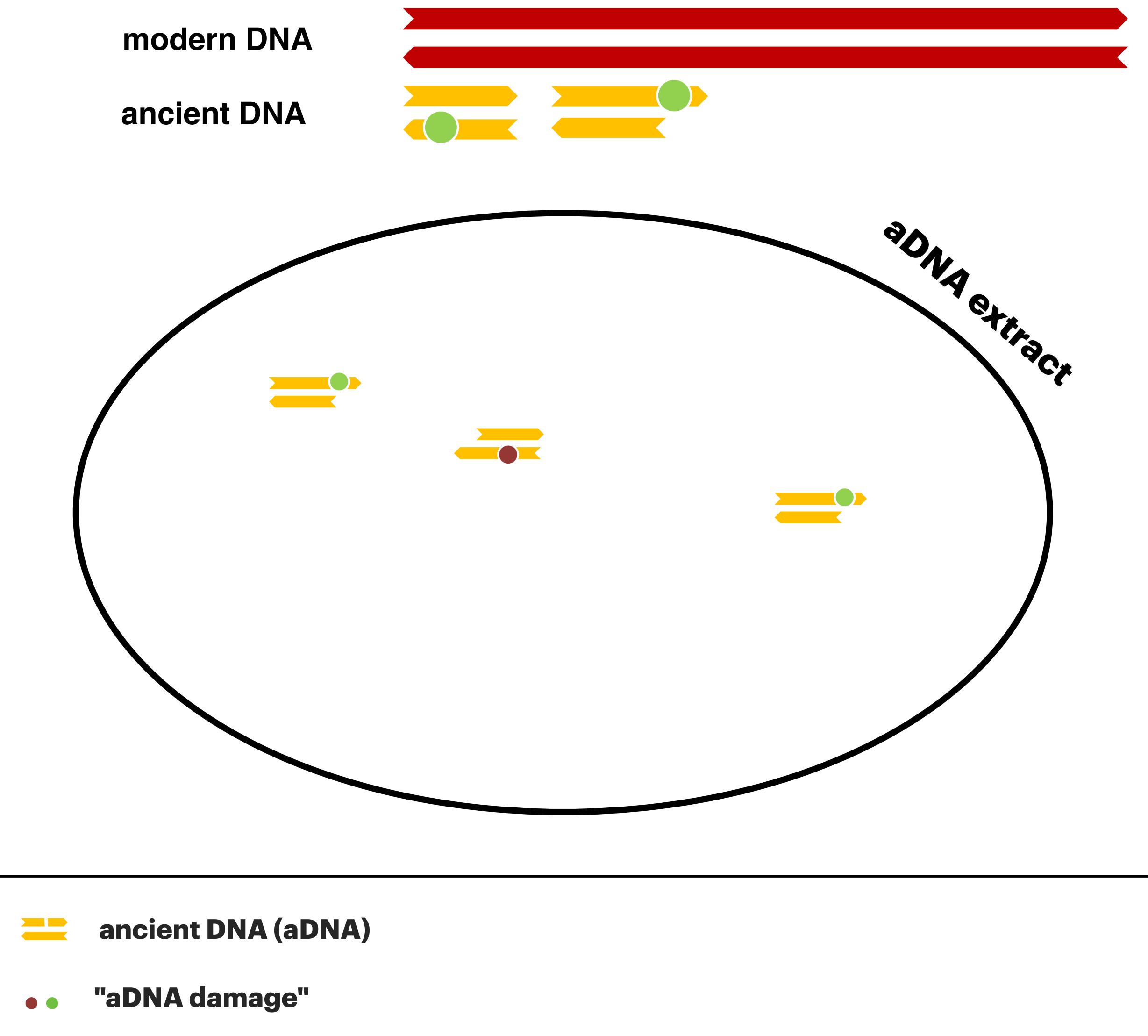
Molecular characteristics of aDNA

- highly fragmented
- post-mortem chemical modifications



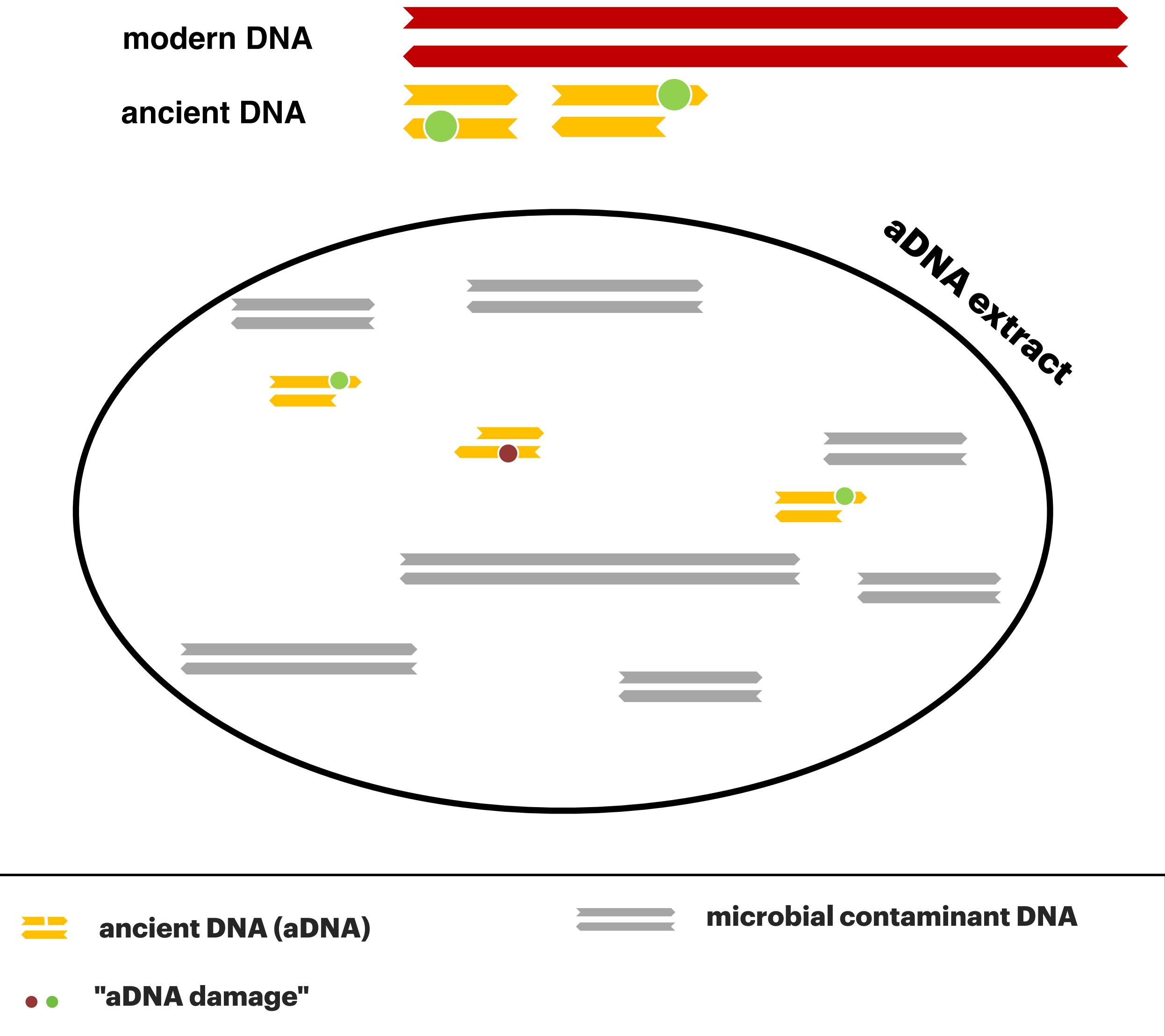
Molecular characteristics of aDNA

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



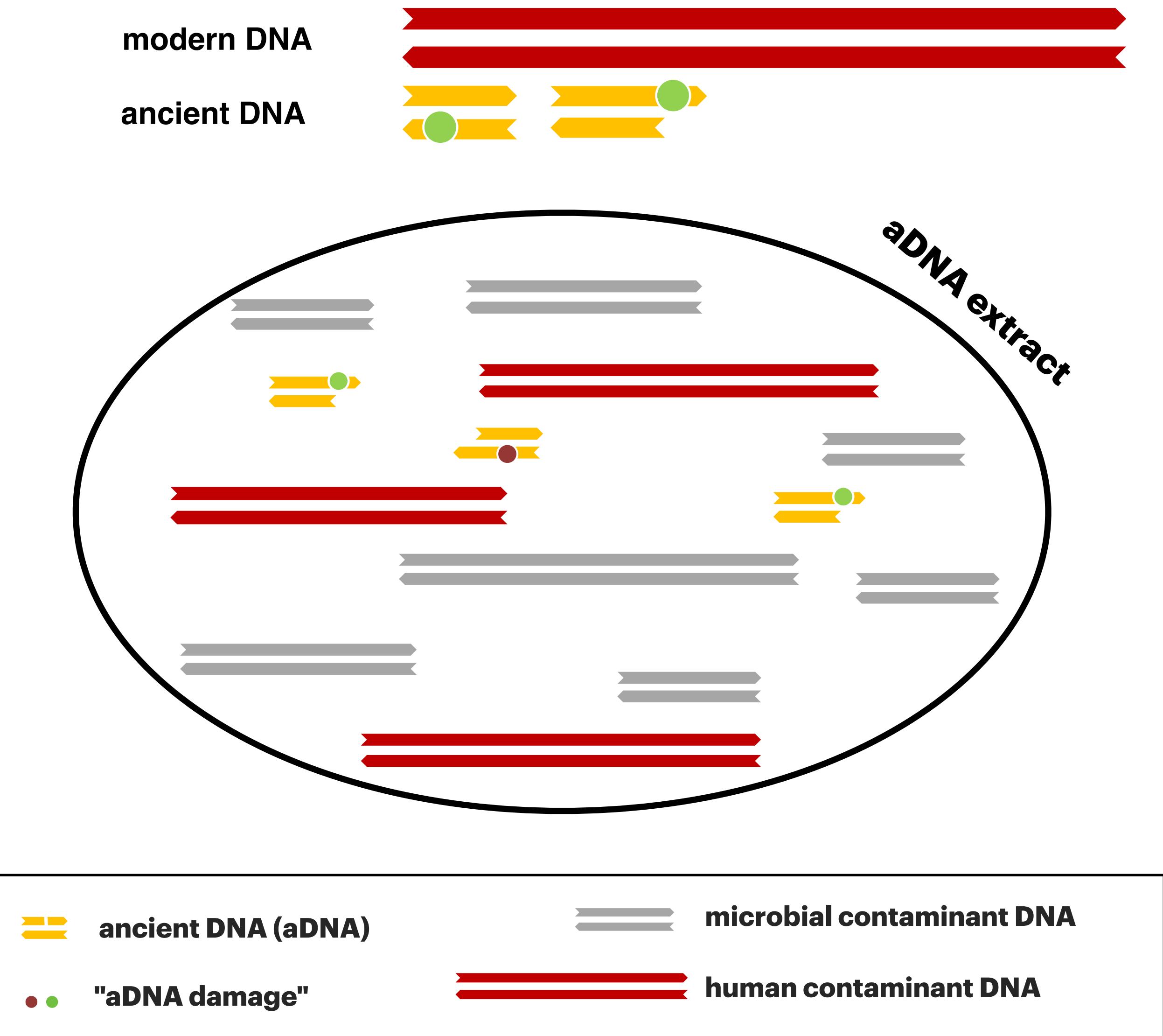
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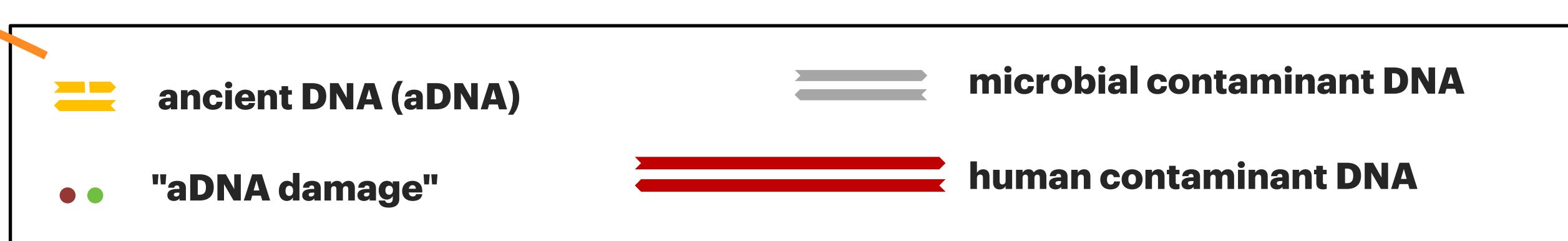
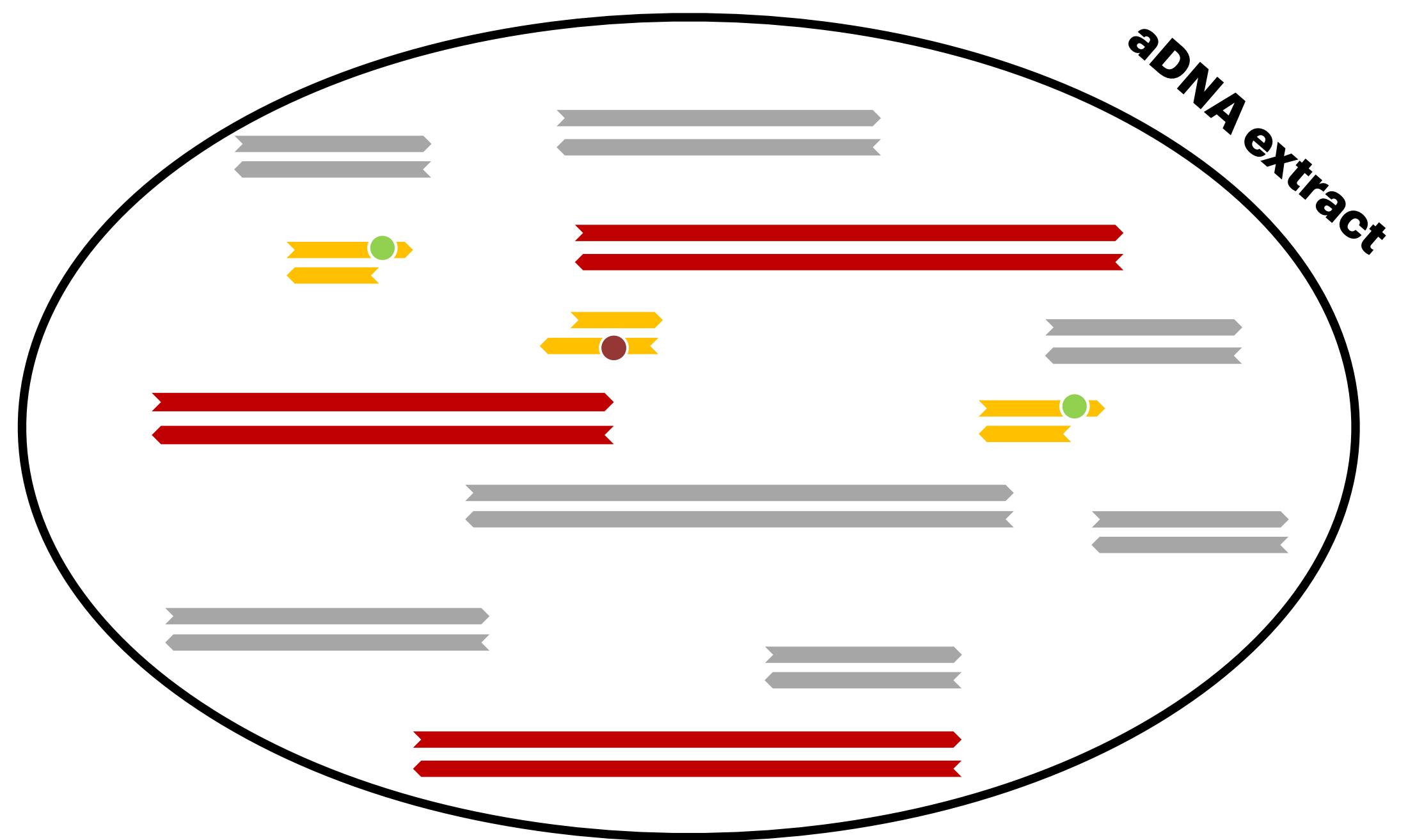


Molecular characteristics of aDNA

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aDNA extract

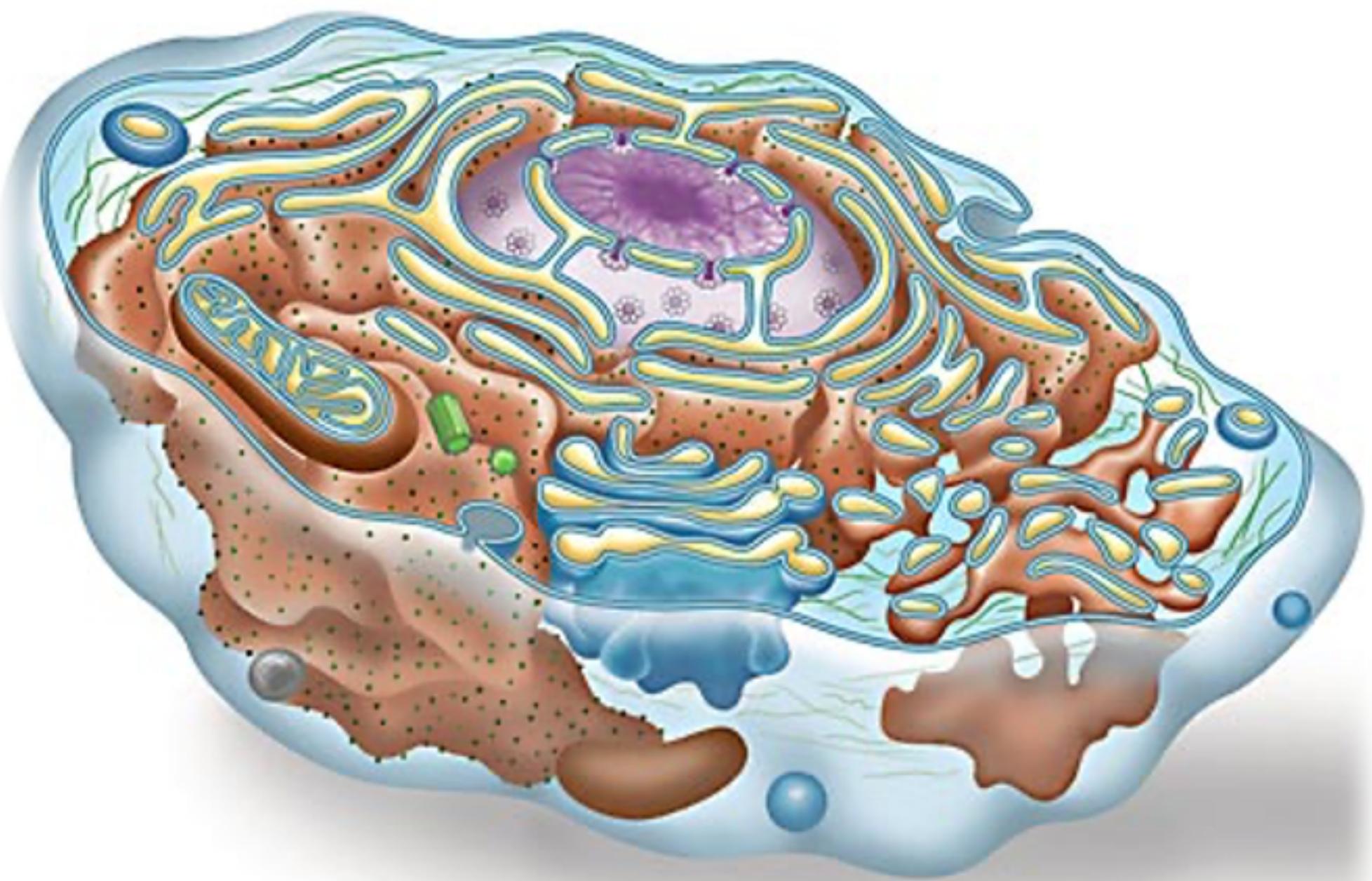
often < 1% endogenous DNA



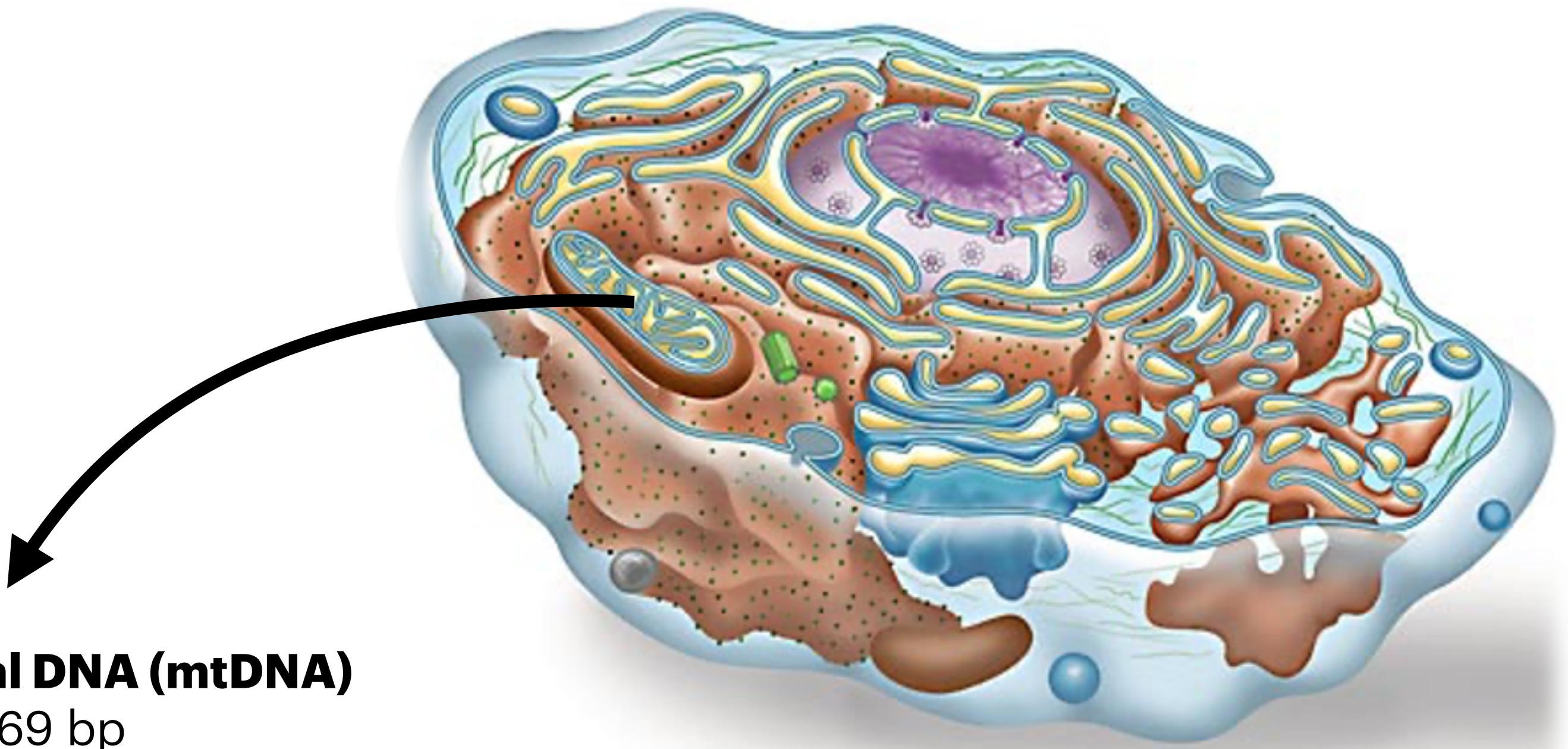
Neanderthal DNA?



Two sources of DNA



Two sources of DNA

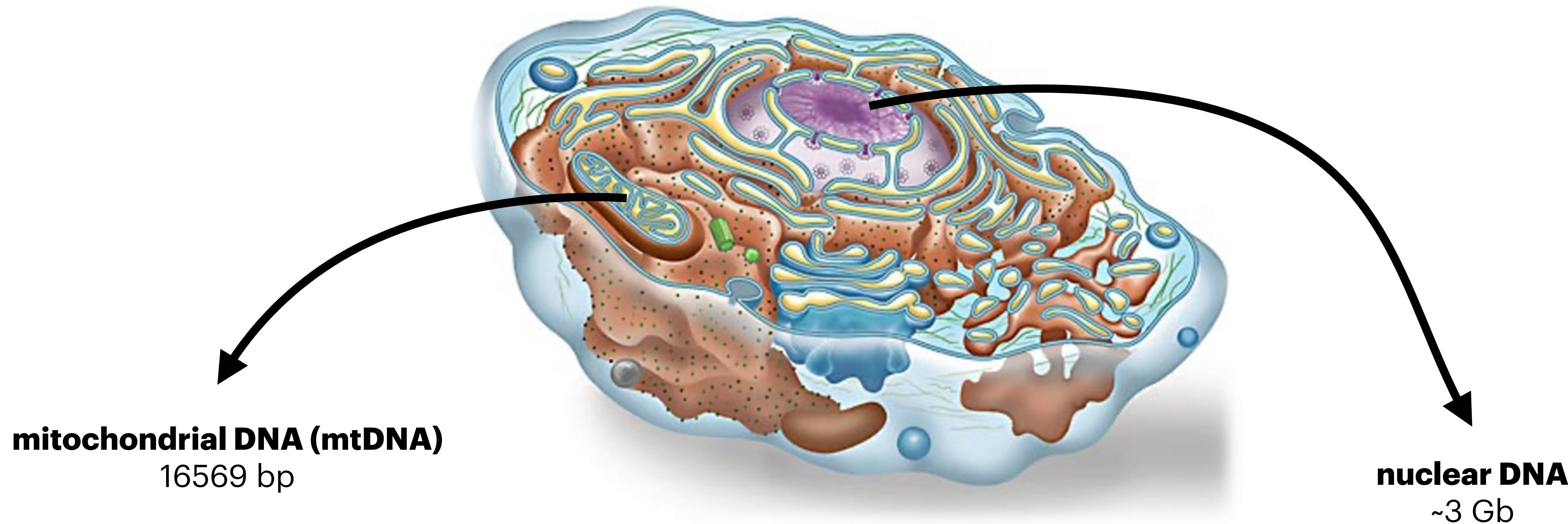


mitochondrial DNA (mtDNA)

16569 bp

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

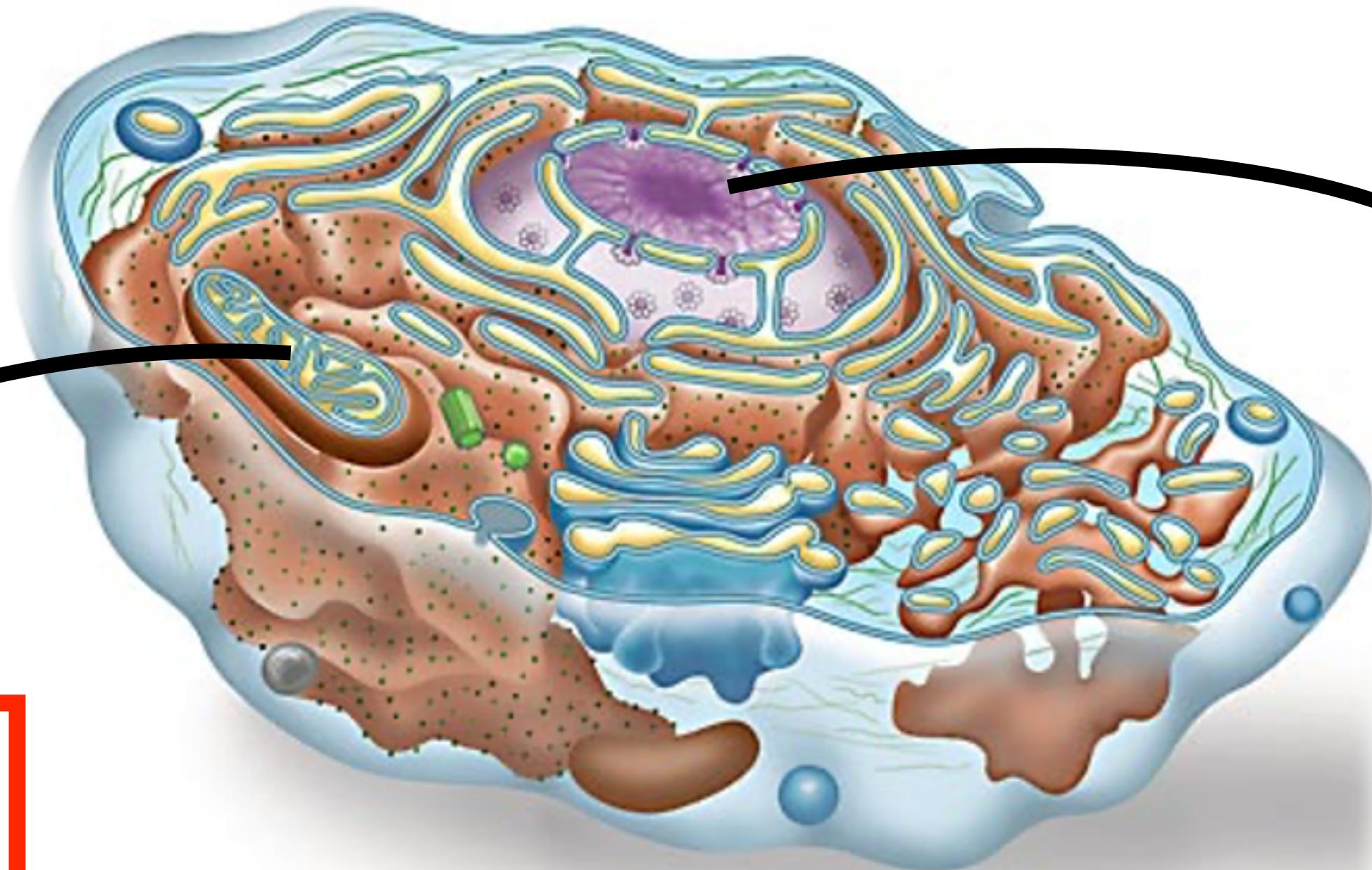
Two sources of DNA



- just one locus
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- **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*



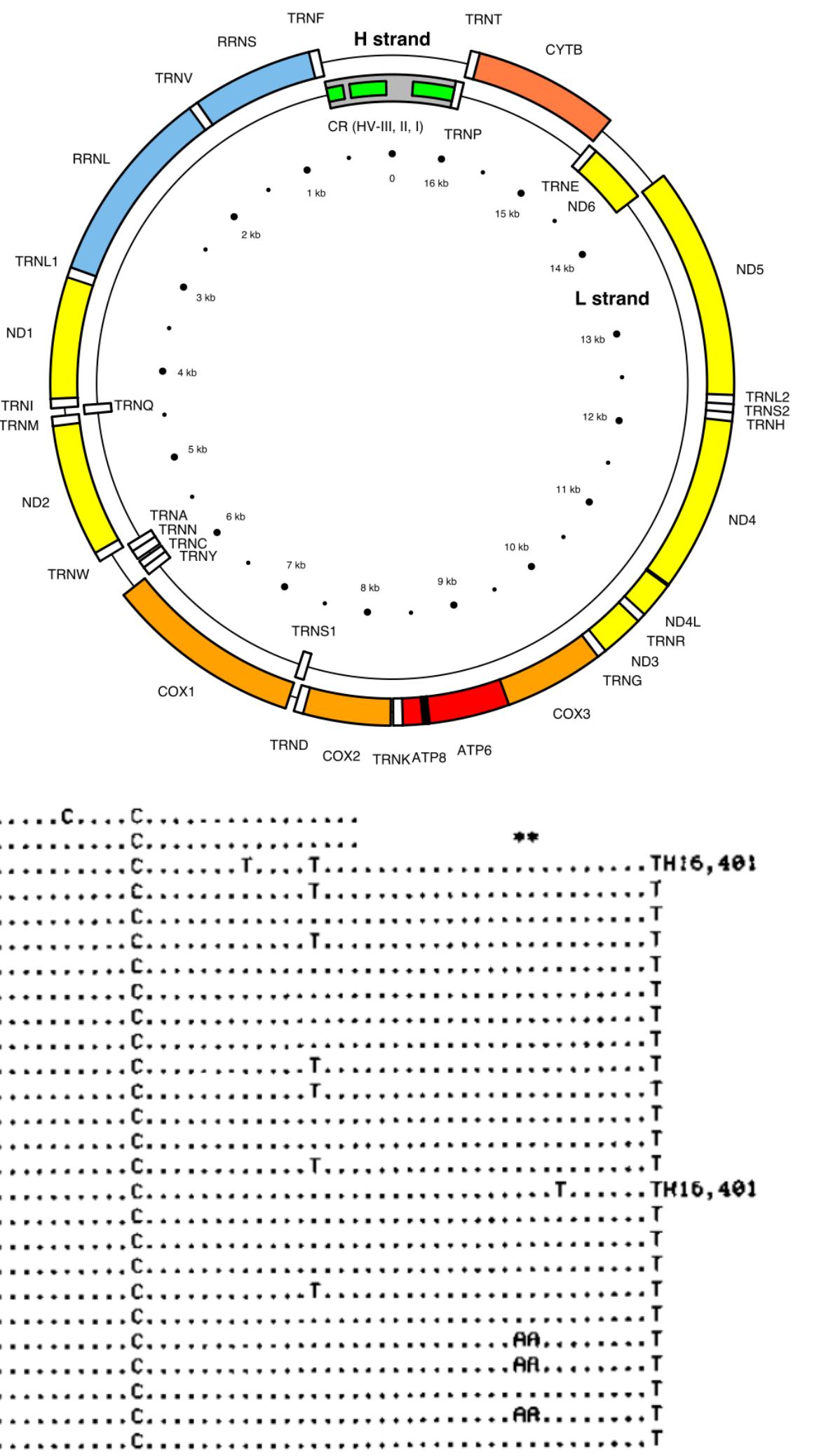
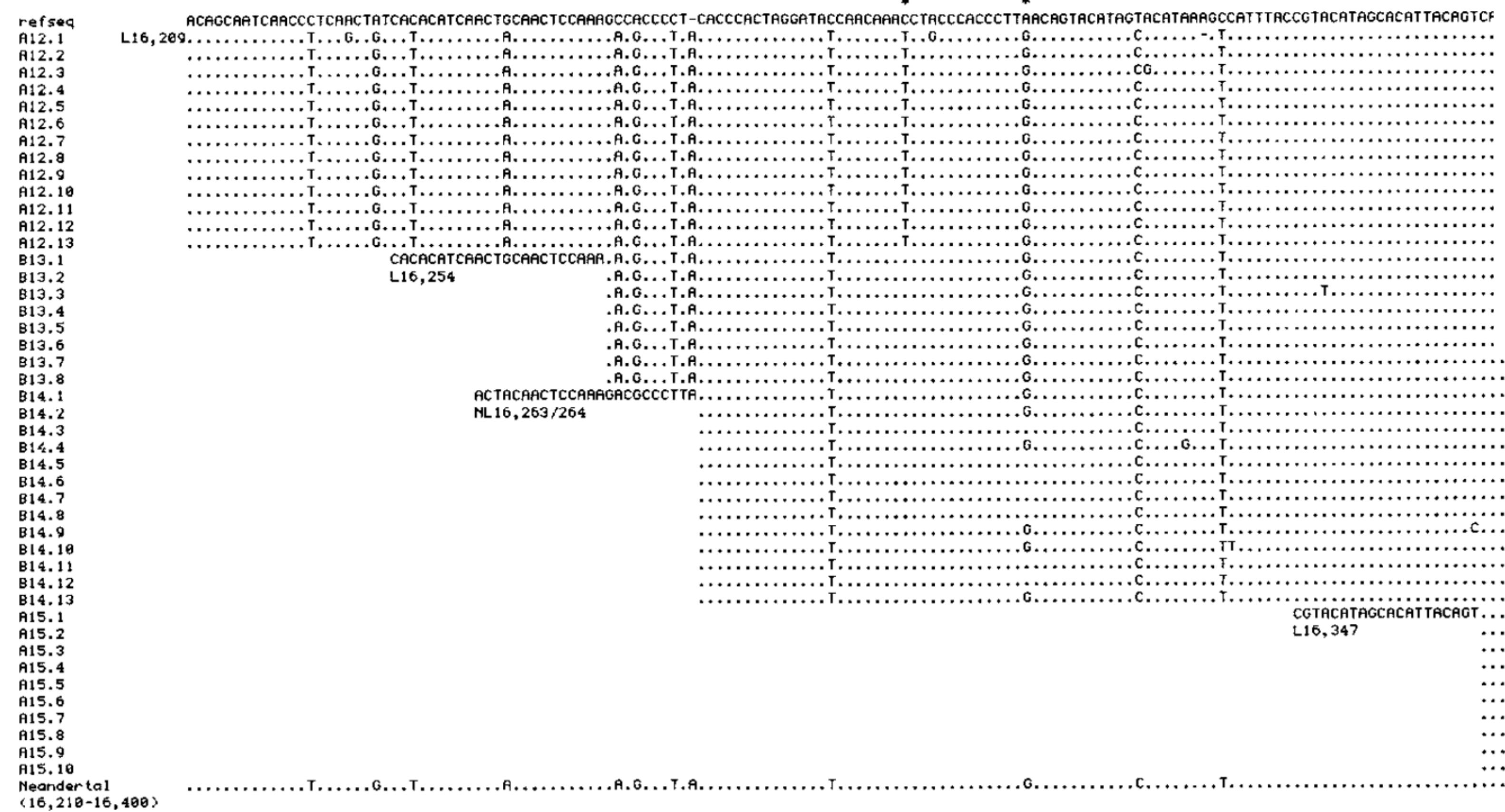
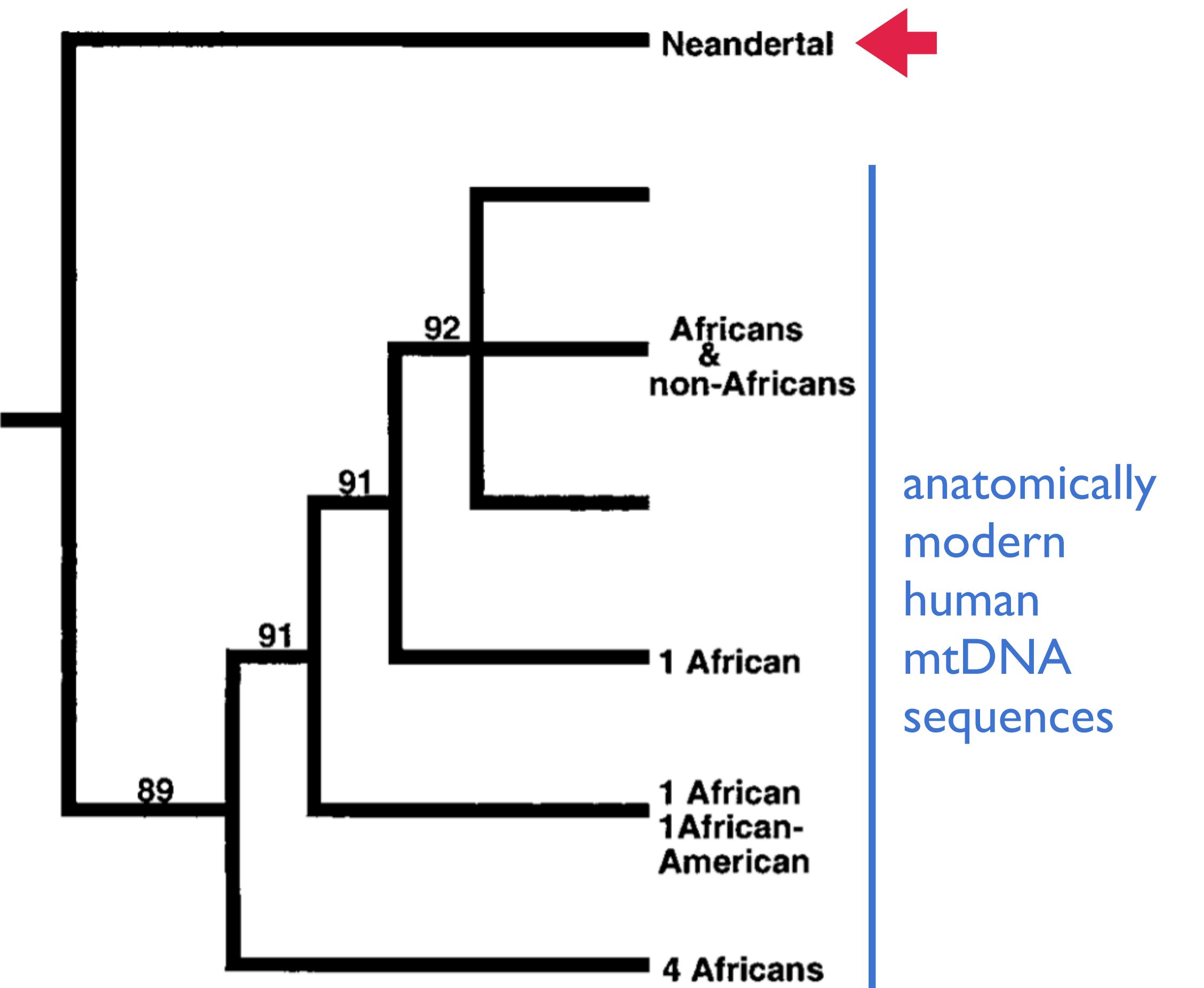
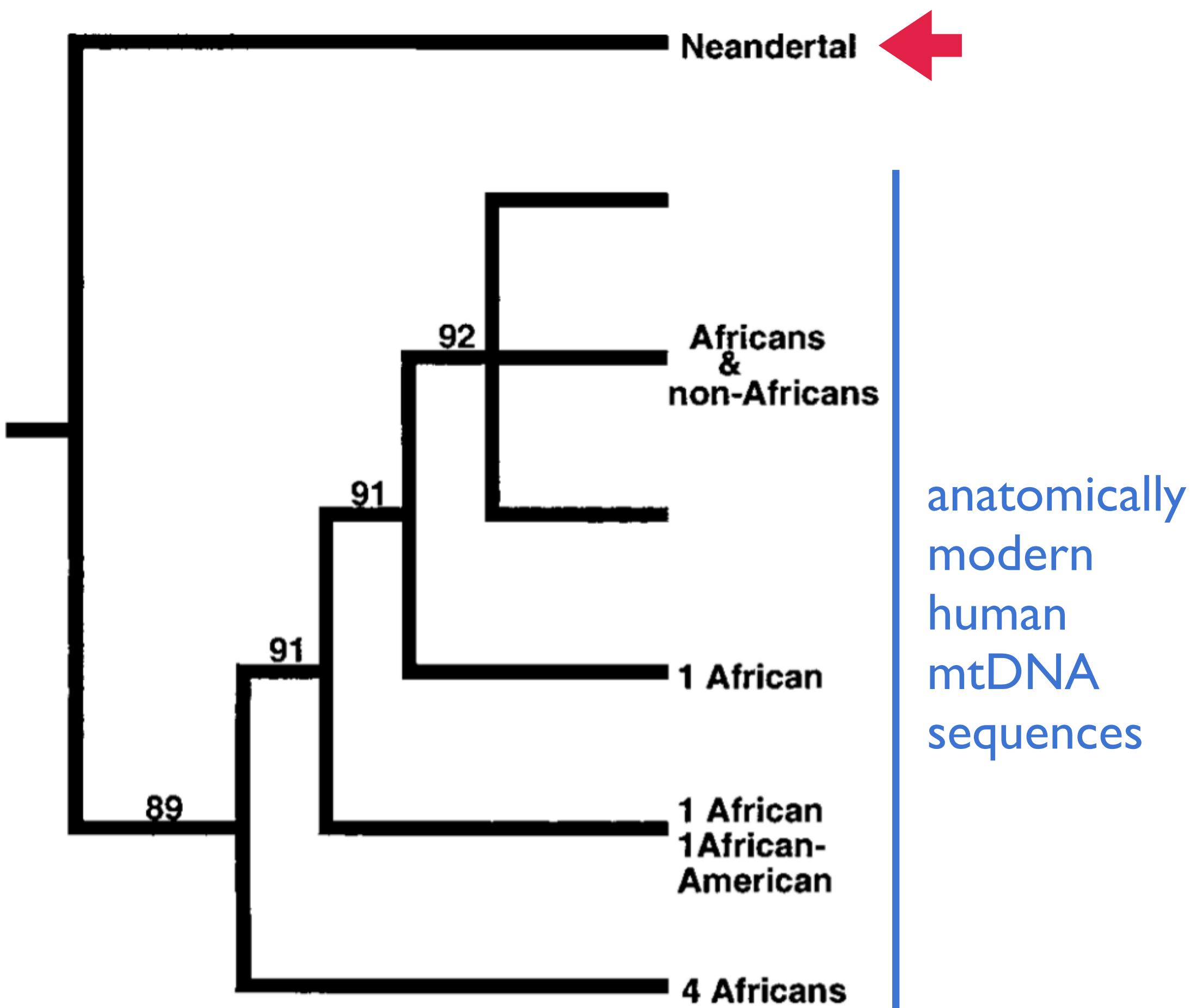


Figure 5. The DNA Sequences of Clones Used to Infer the Sequence of the Hypervariable Region I of the Neandertal Individual

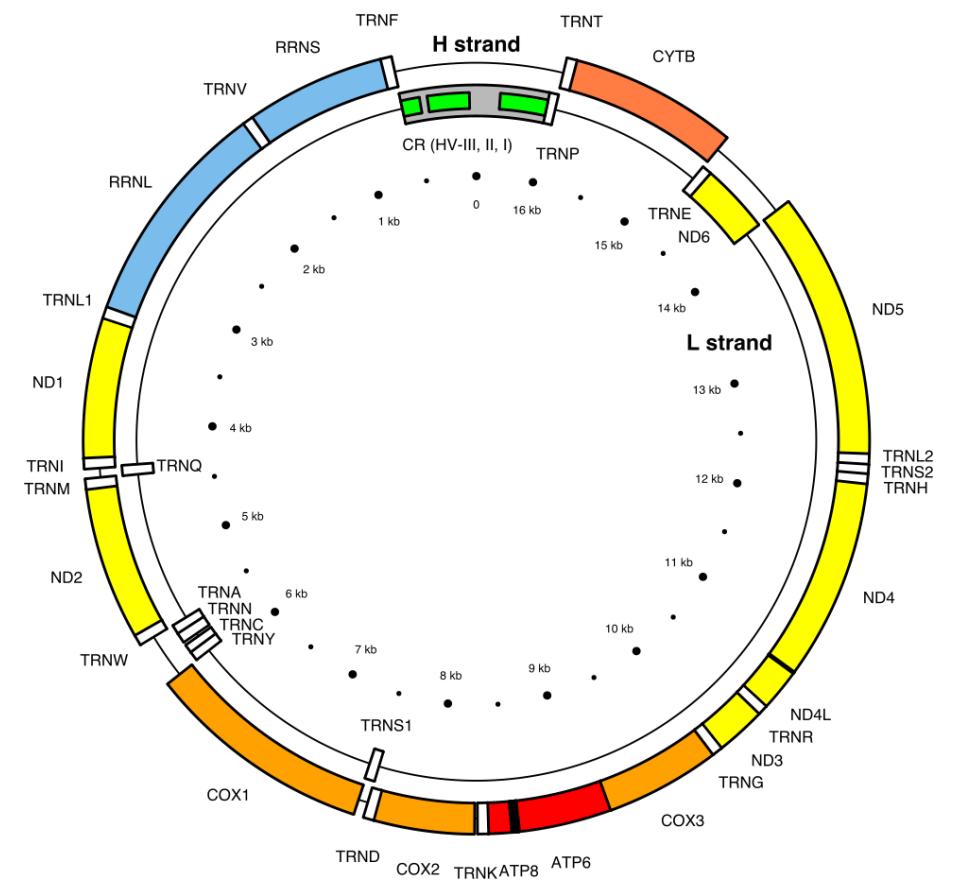


No introgression?

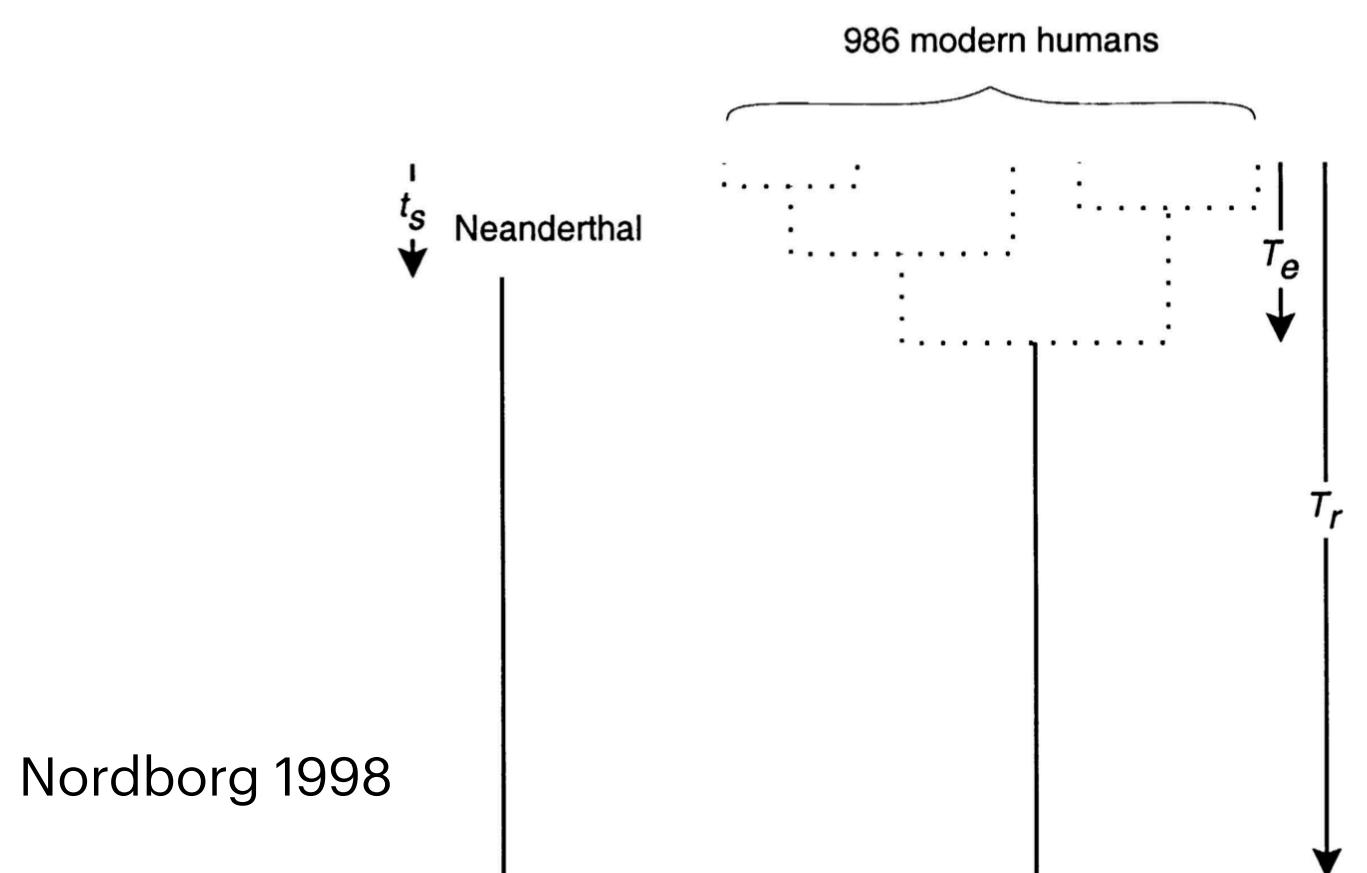


When the Neandertal mtDNA sequence is used to root a neighbor joining tree of modern human mtDNA sequences (Figure 7a), the first three branches consist exclusively of African sequences. The Neandertal mtDNA sequence thus supports a scenario in which modern humans arose recently in Africa as a distinct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

mitochondrial DNA



16 kb

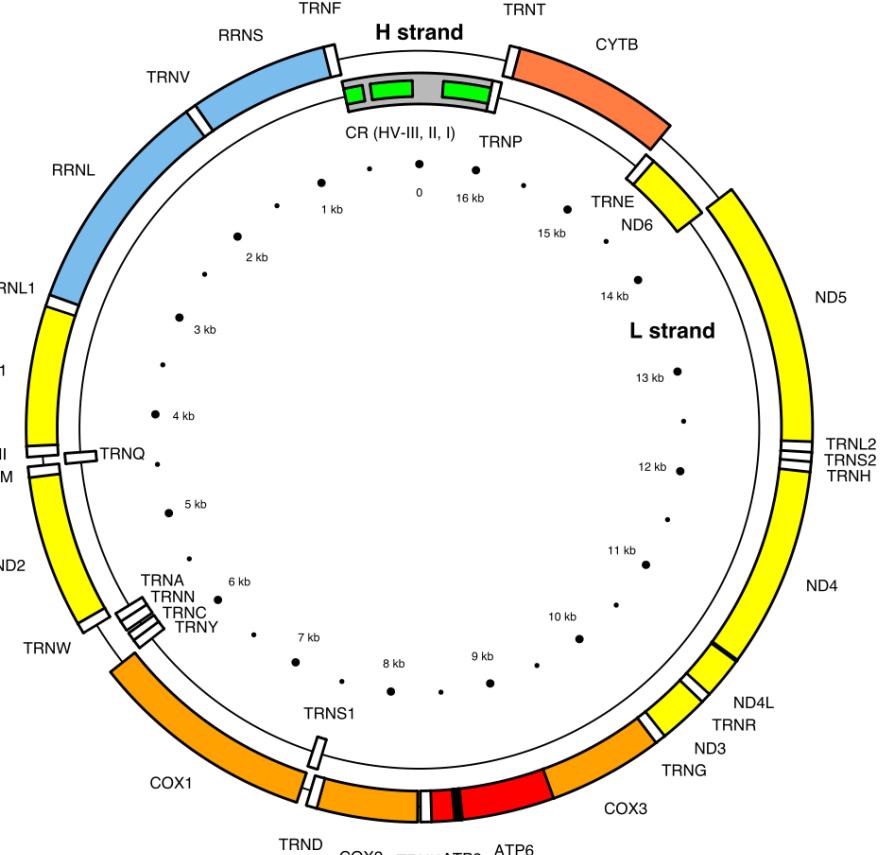


one mtDNA phylogenetic tree
(maternal history)

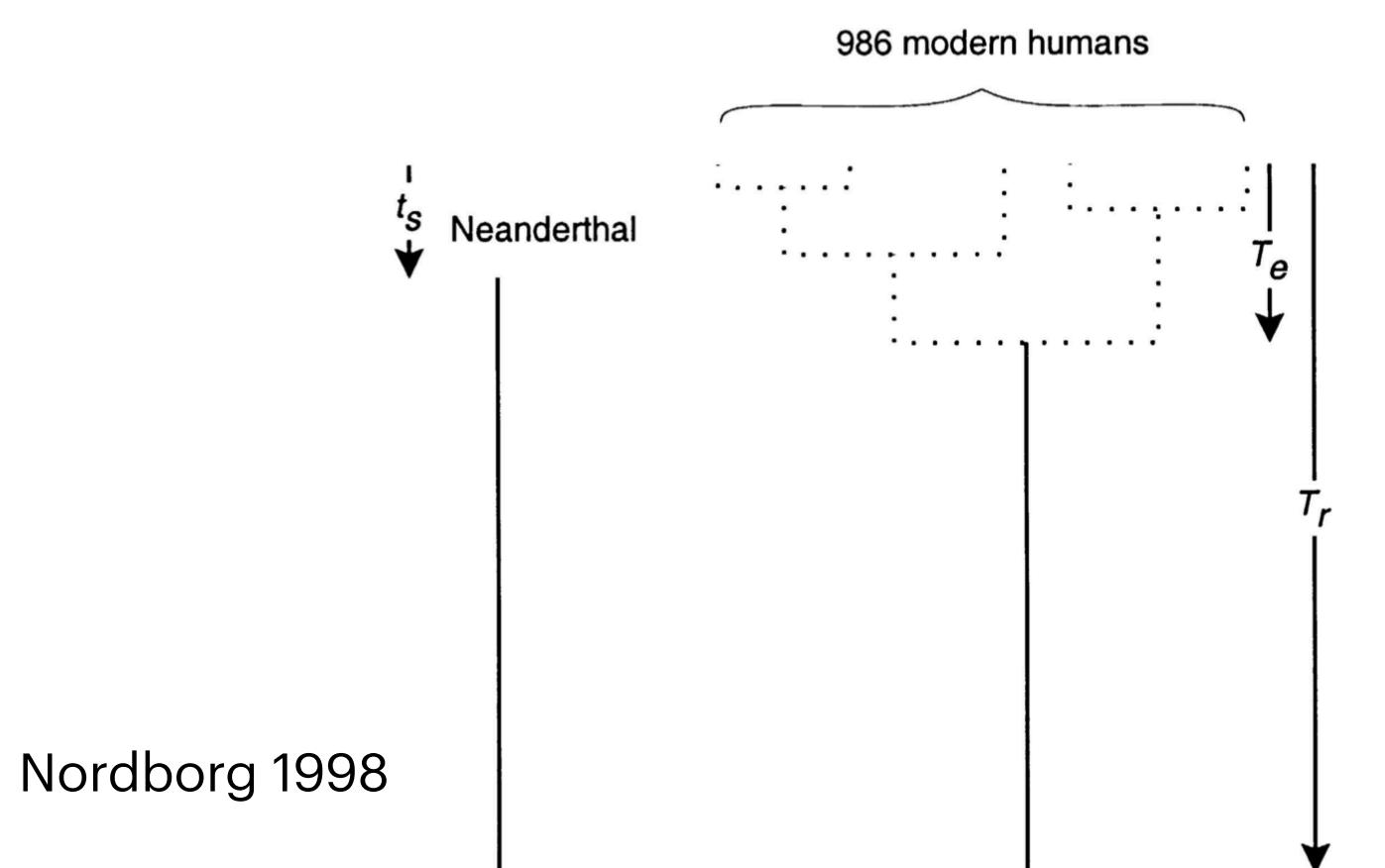
mitochondrial DNA

vs

nuclear DNA

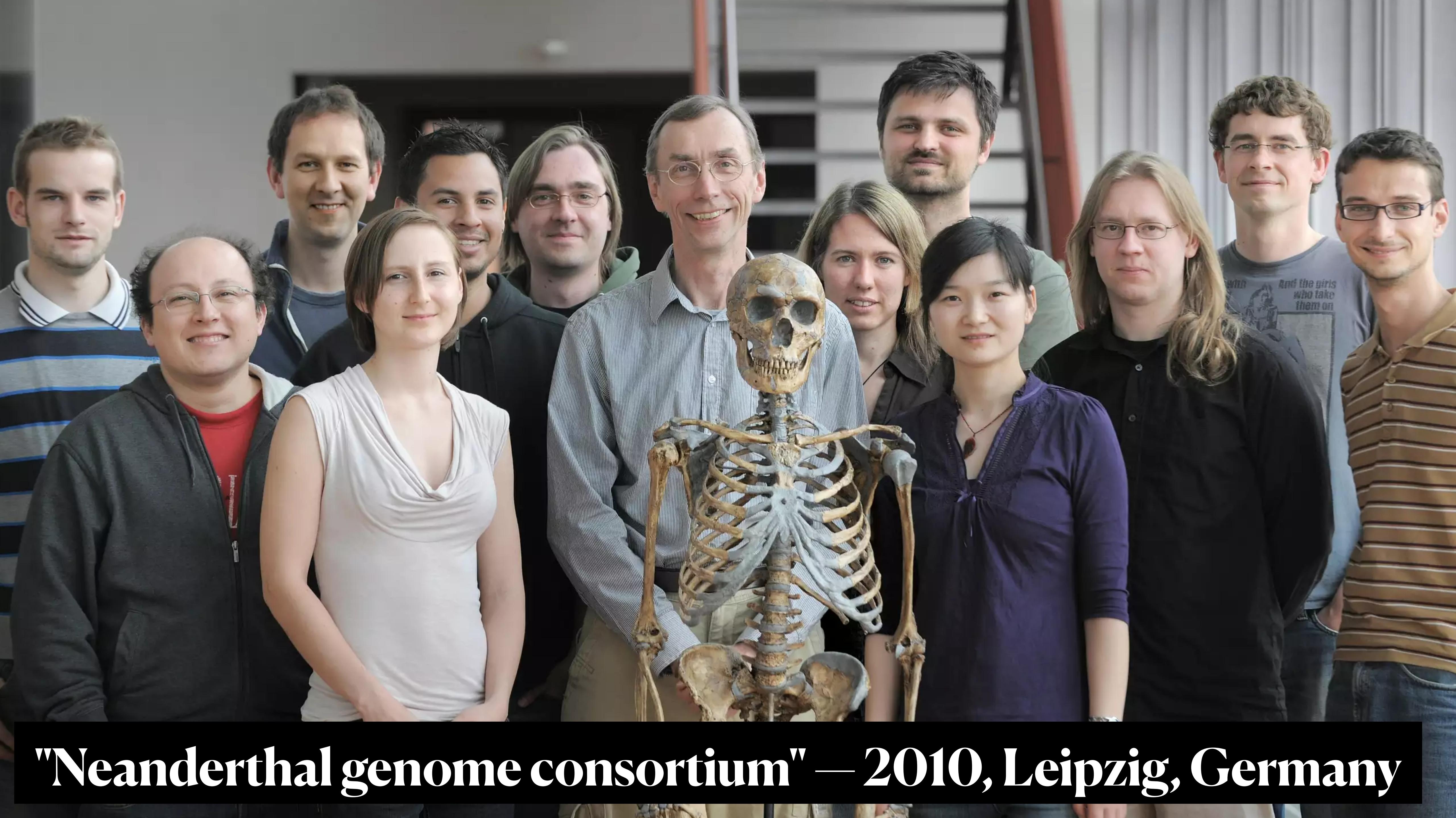


3 Gb



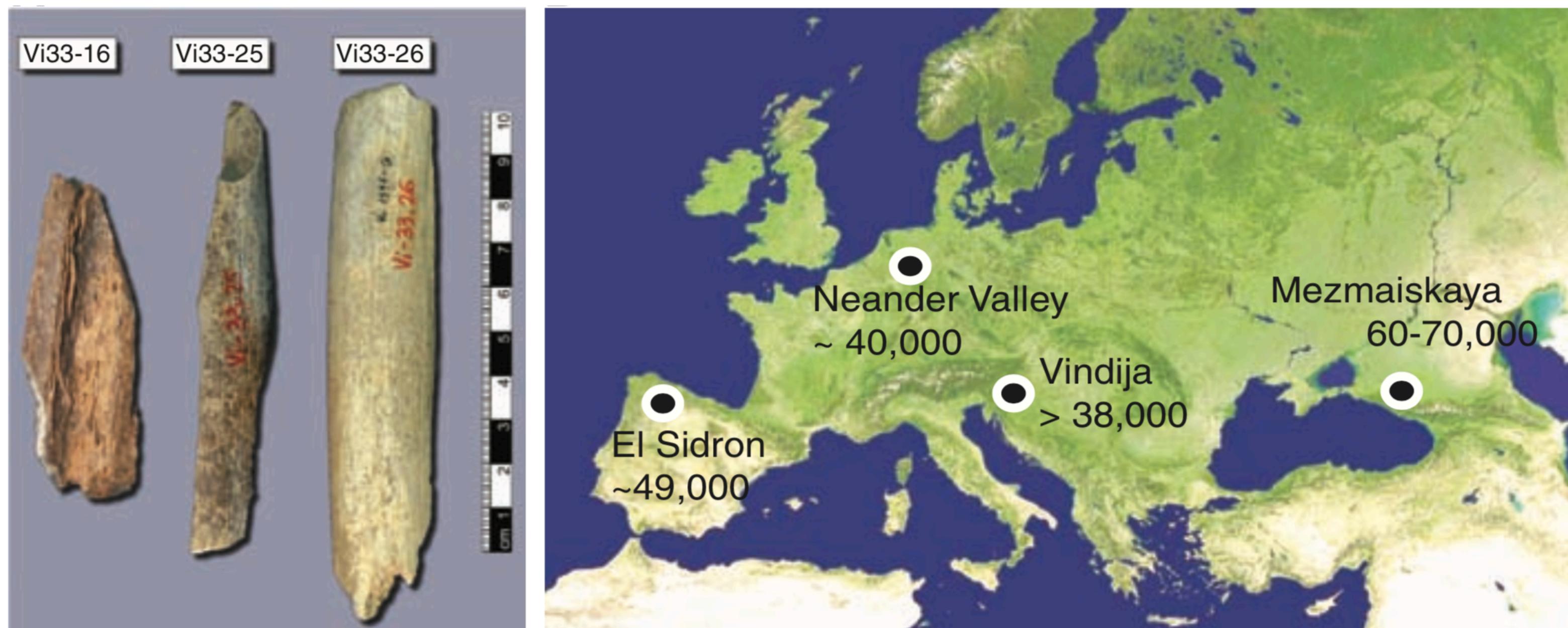
one mtDNA phylogenetic tree
(maternal history)

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



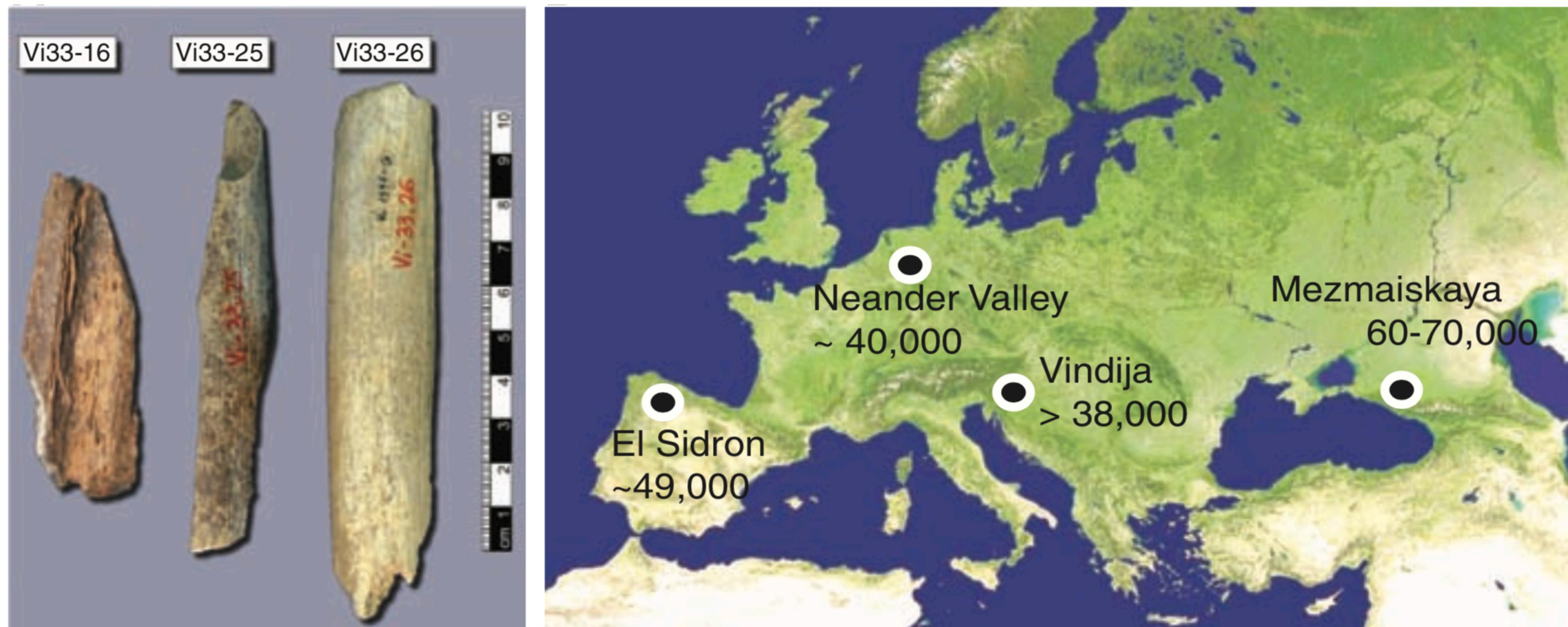
"Neanderthal genome consortium" – 2010, Leipzig, Germany

2010: "draft" Neanderthal genome



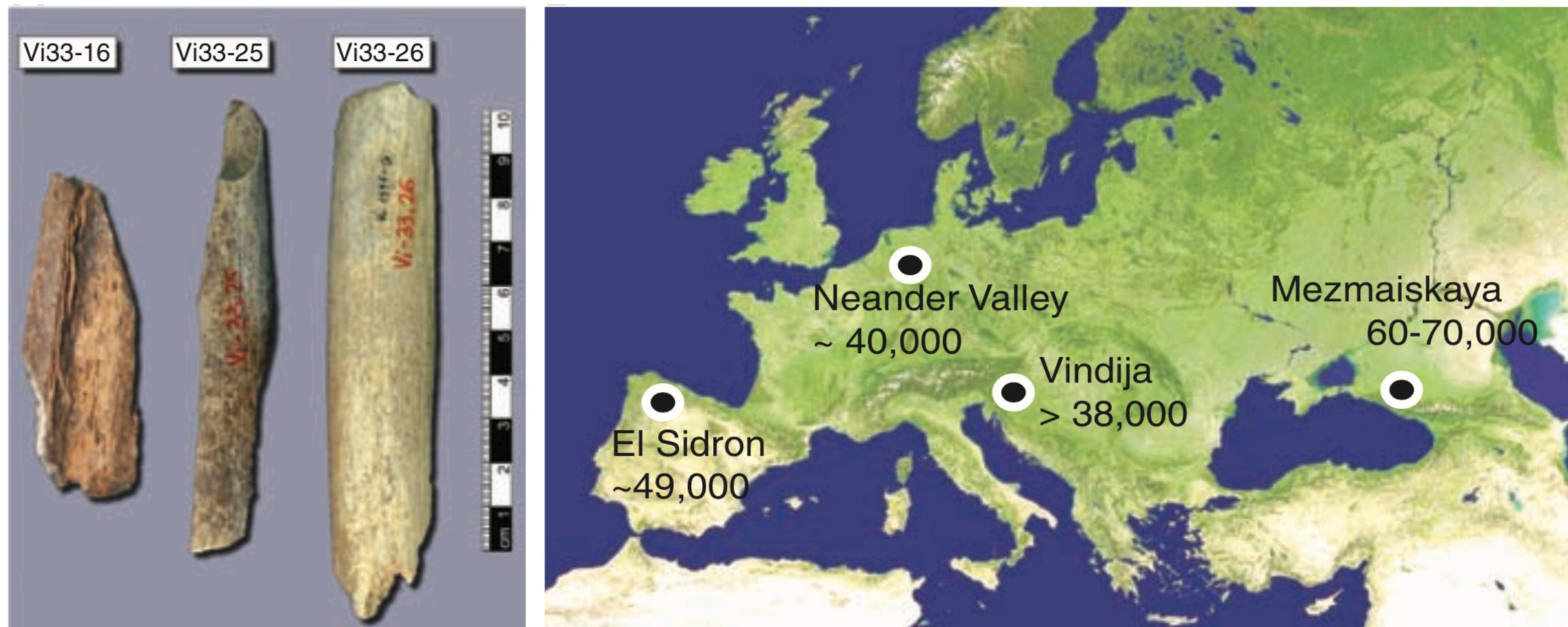
- DNA extracted from bone powder of three bones

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



Typical aDNA workflow



Typical aDNA workflow



**dentist
drill**

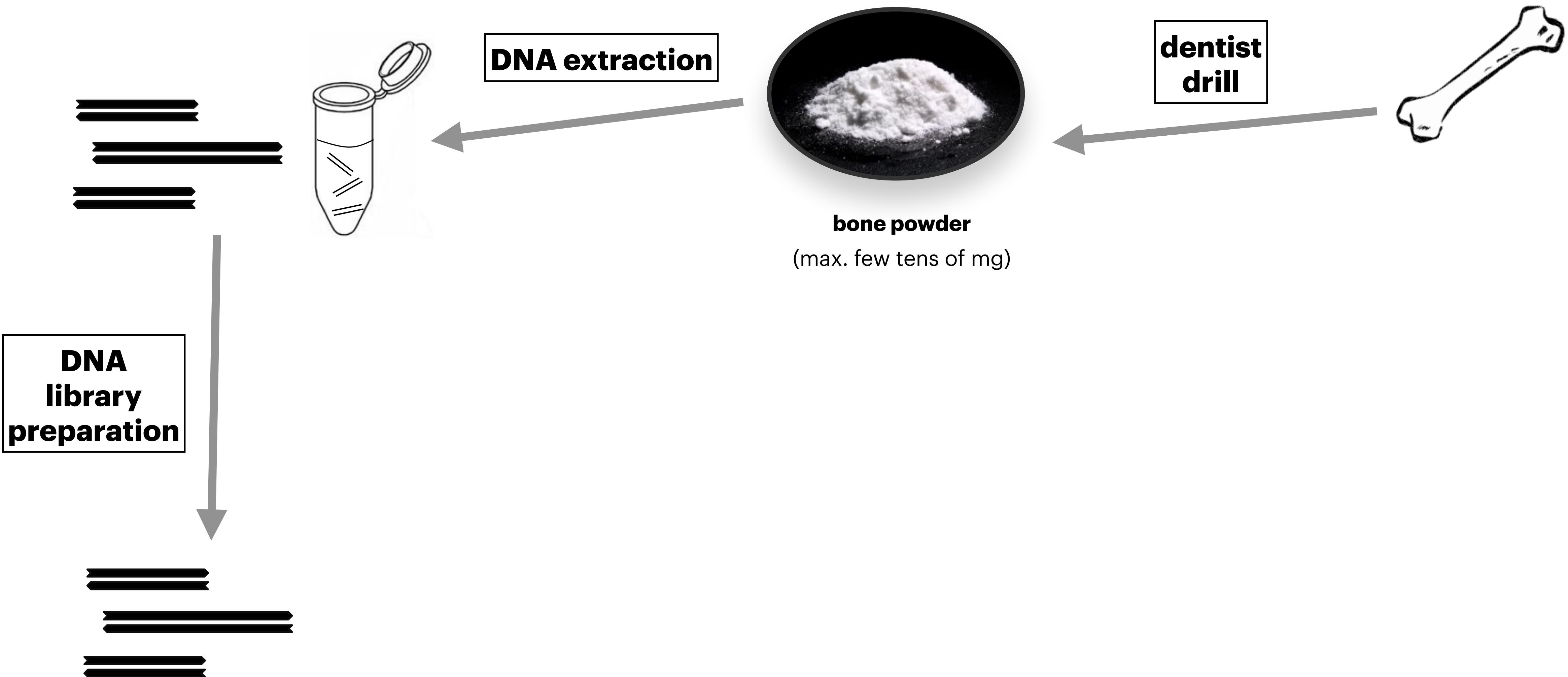


bone powder
(max. few tens of mg)

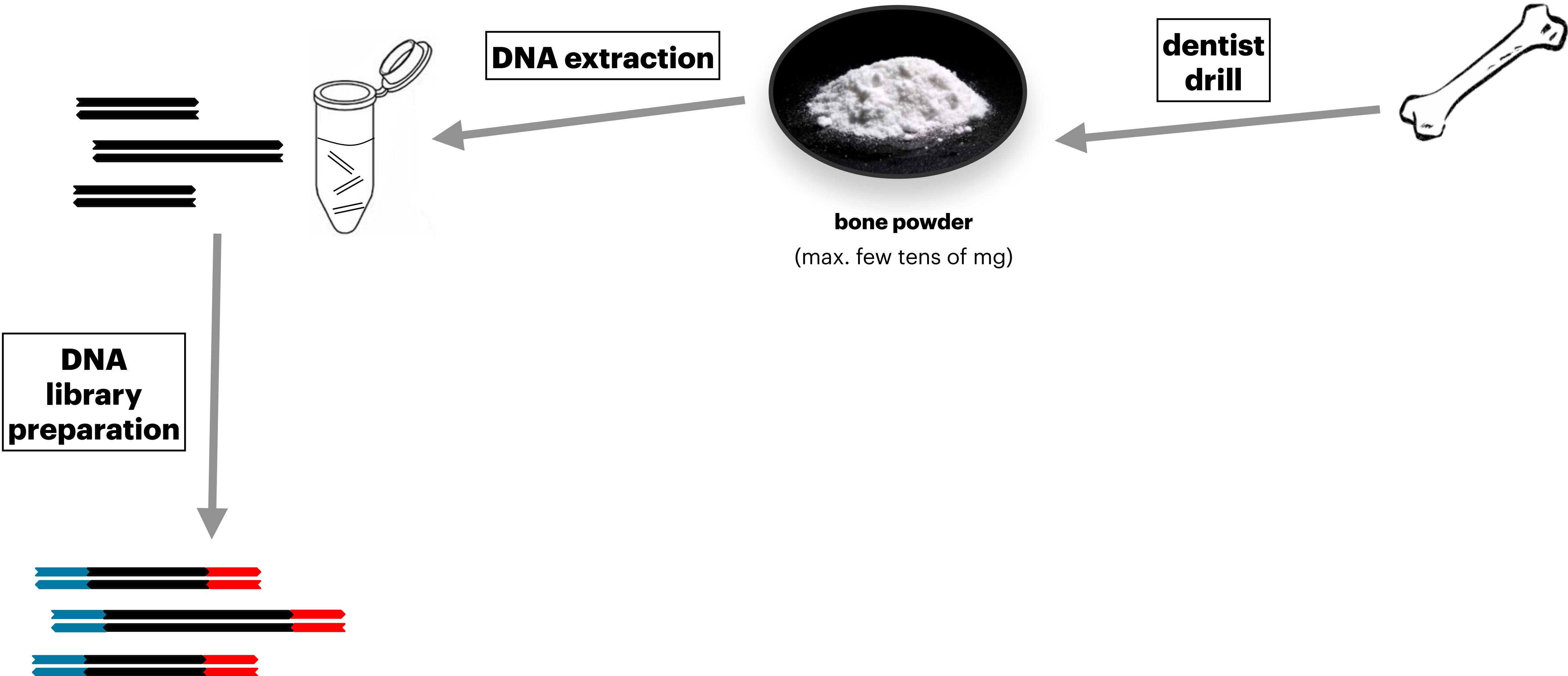
Typical aDNA workflow



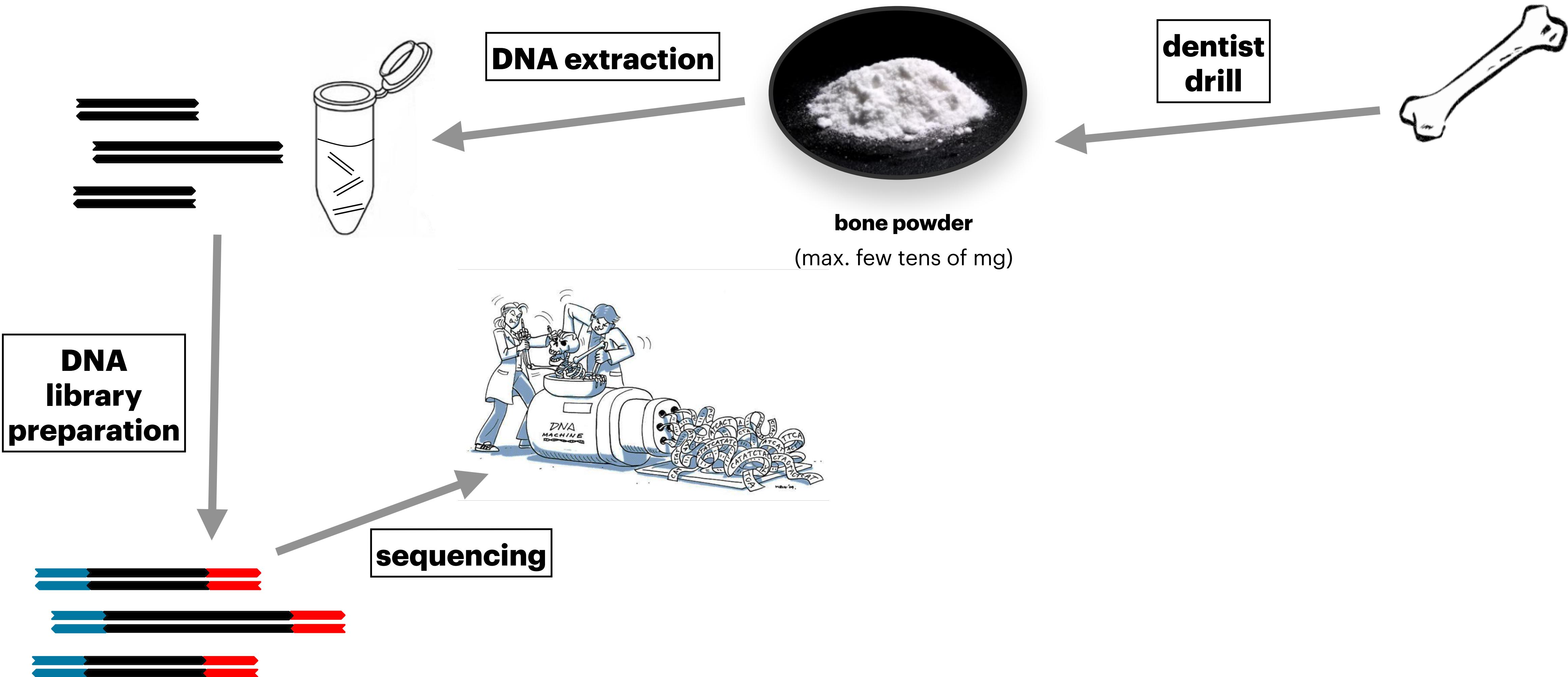
Typical aDNA workflow



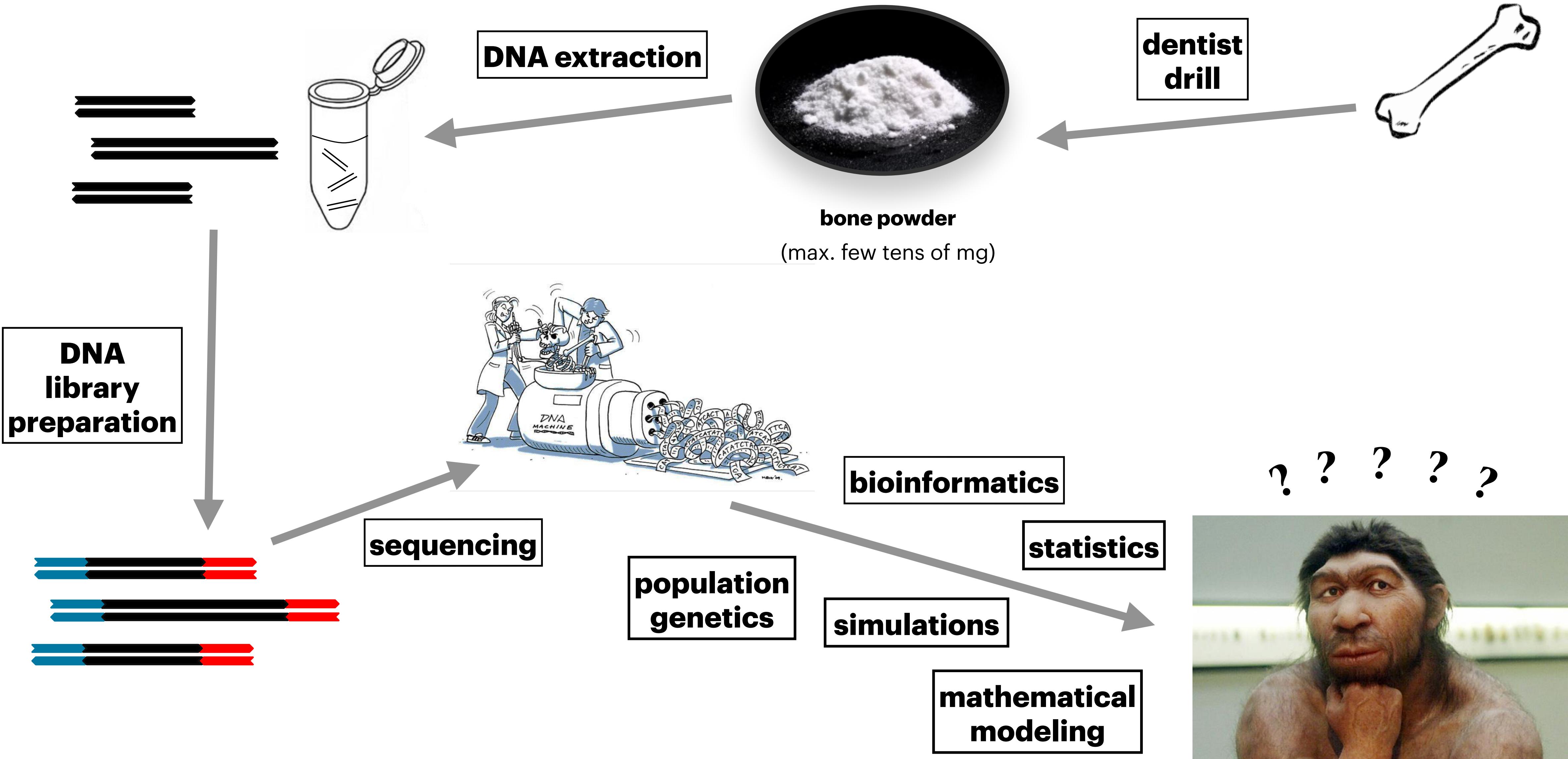
Typical aDNA workflow



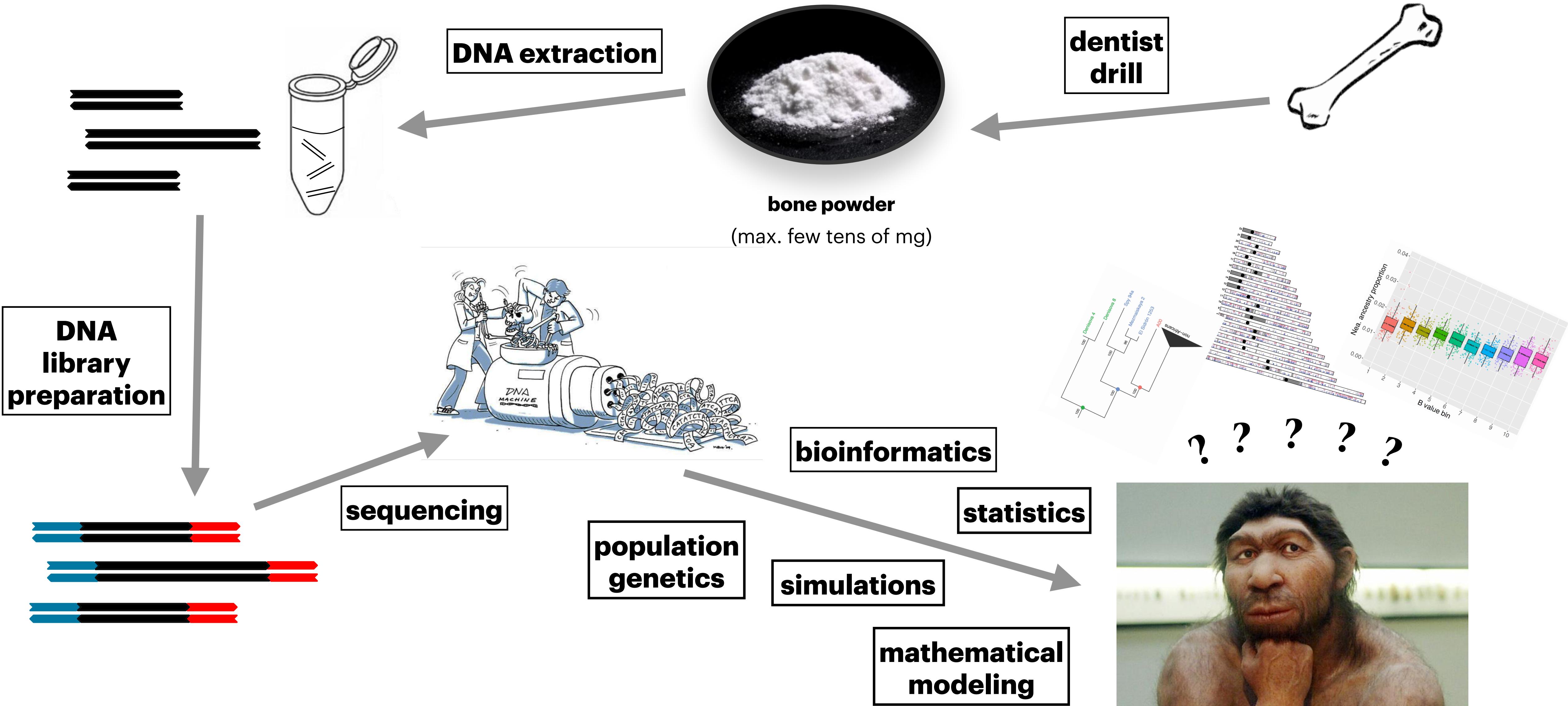
Typical aDNA workflow



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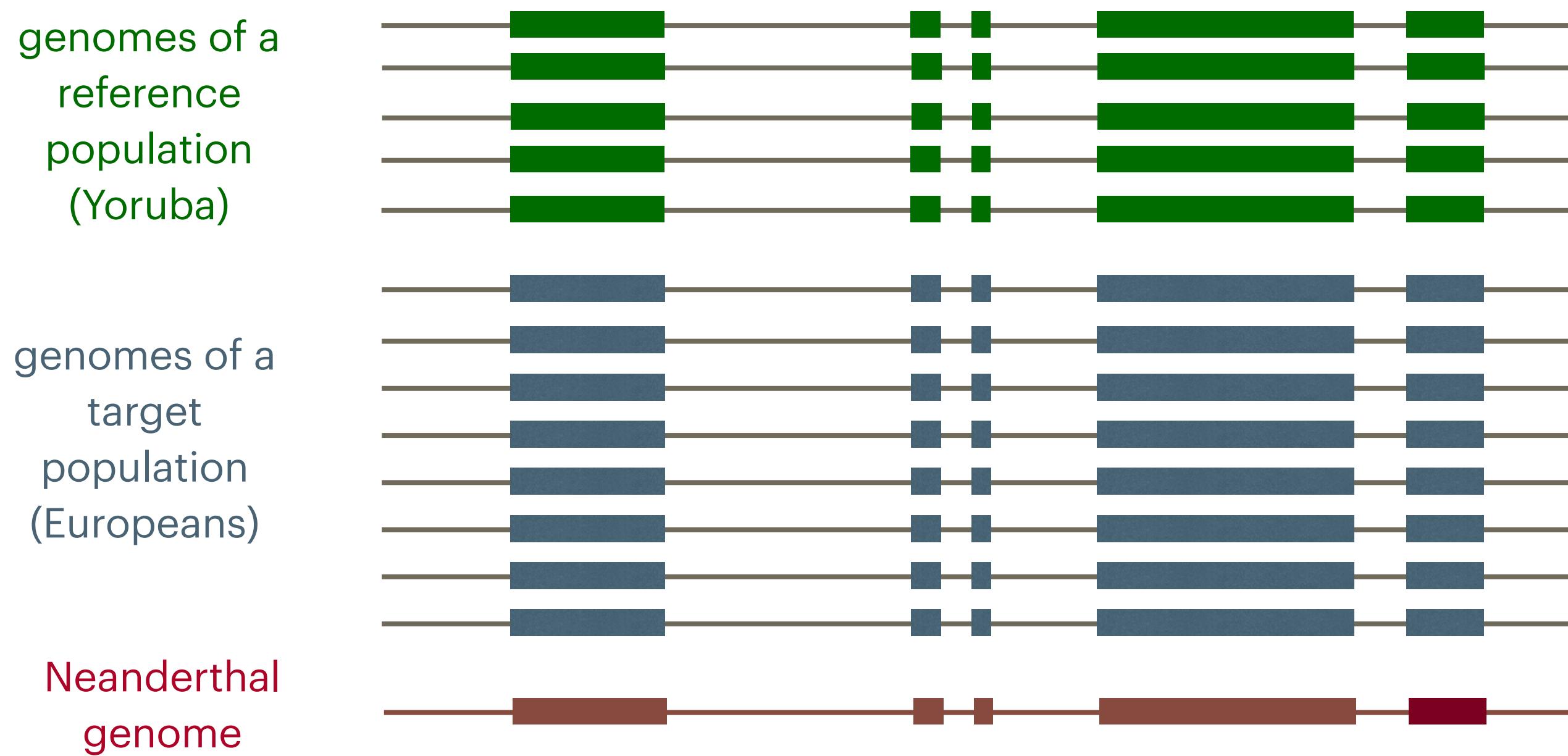
So we have a Neanderthal genome...



...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

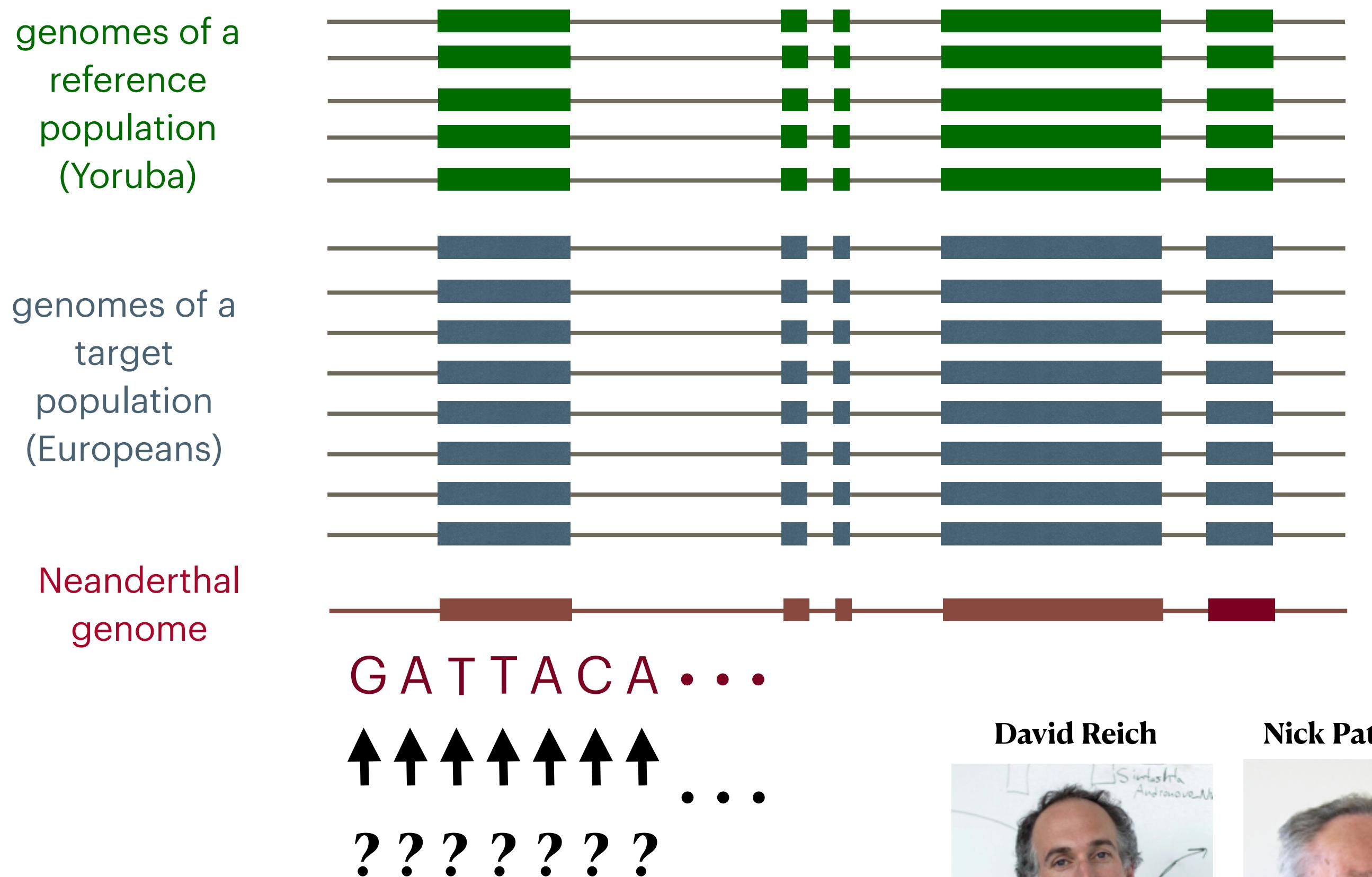


Nick Patterson



Green et al. (Science 2010)

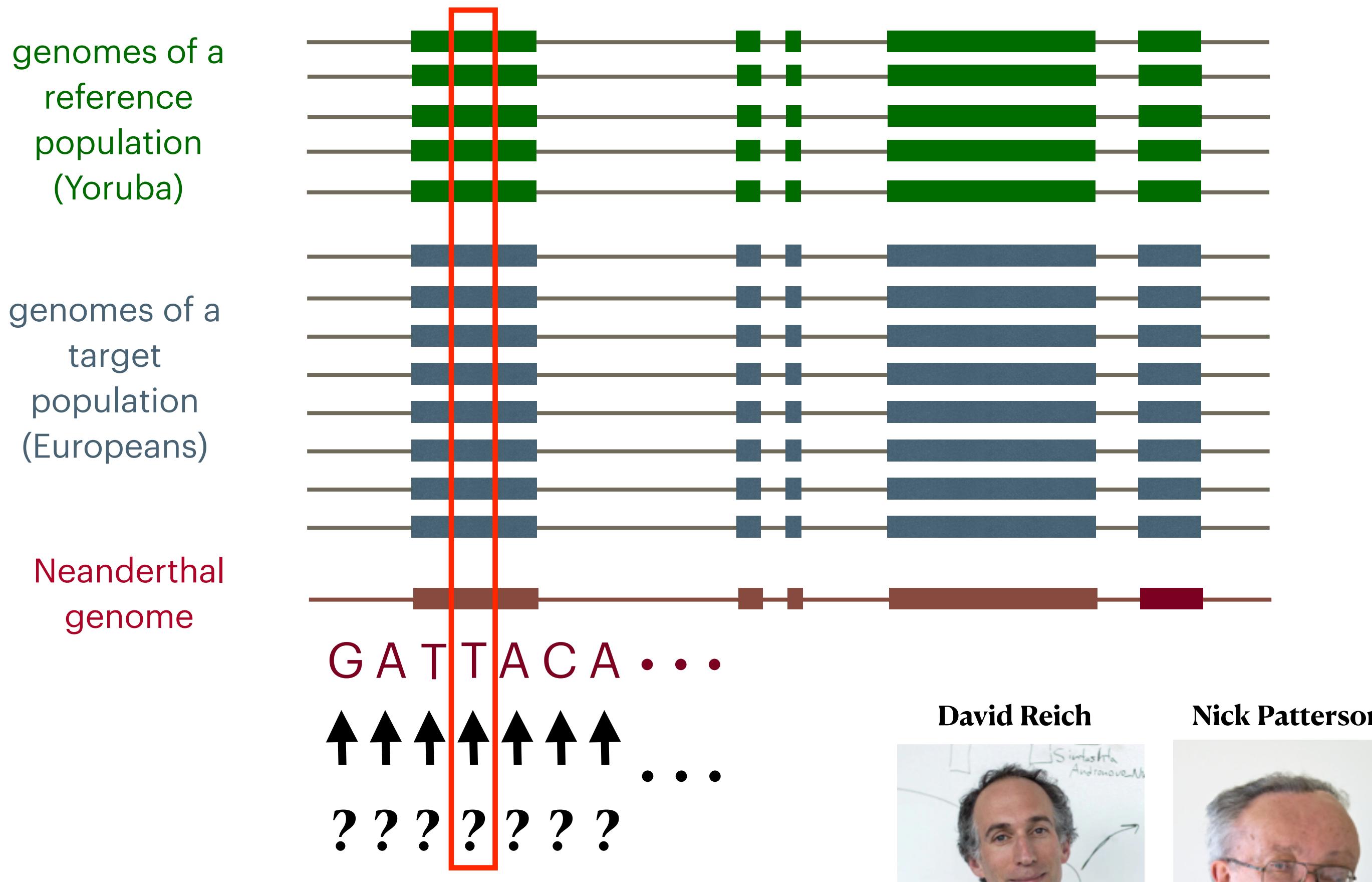
How to test the gene-flow hypothesis?



Green et al. (Science 2010)

How to test the gene-flow hypothesis?

Situation at one locus...



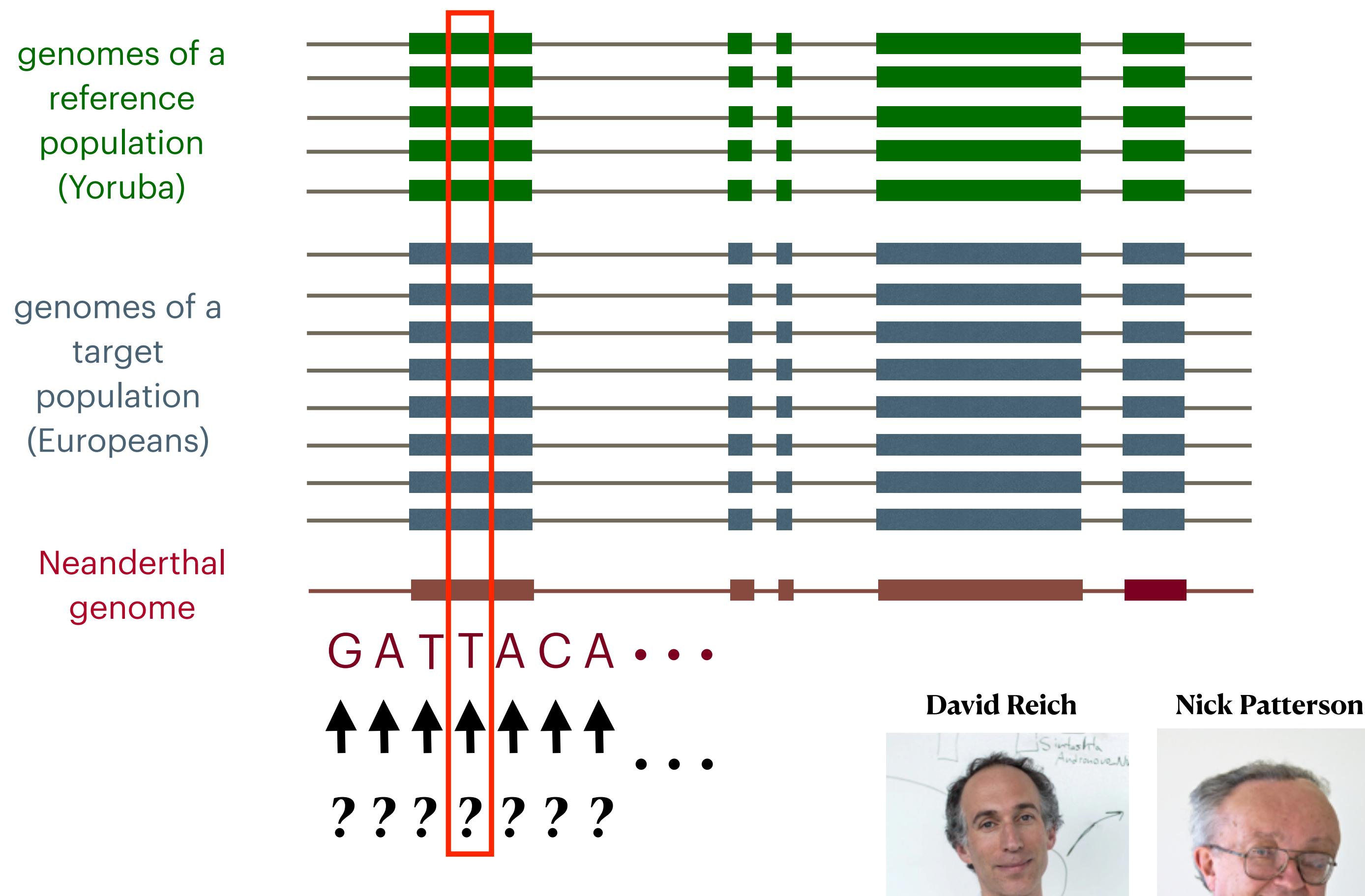
David Reich



Nick Patterson

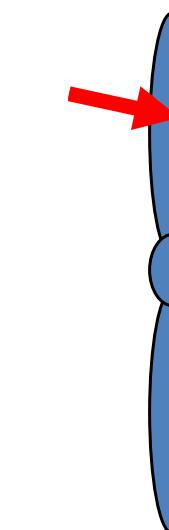
Green et al. (Science 2010)

How to test the gene-flow hypothesis?

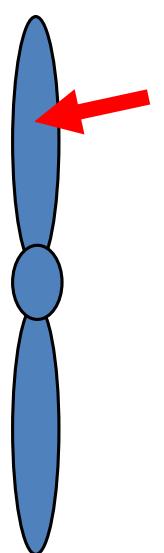


Situation at one locus...

person of an African ancestry



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

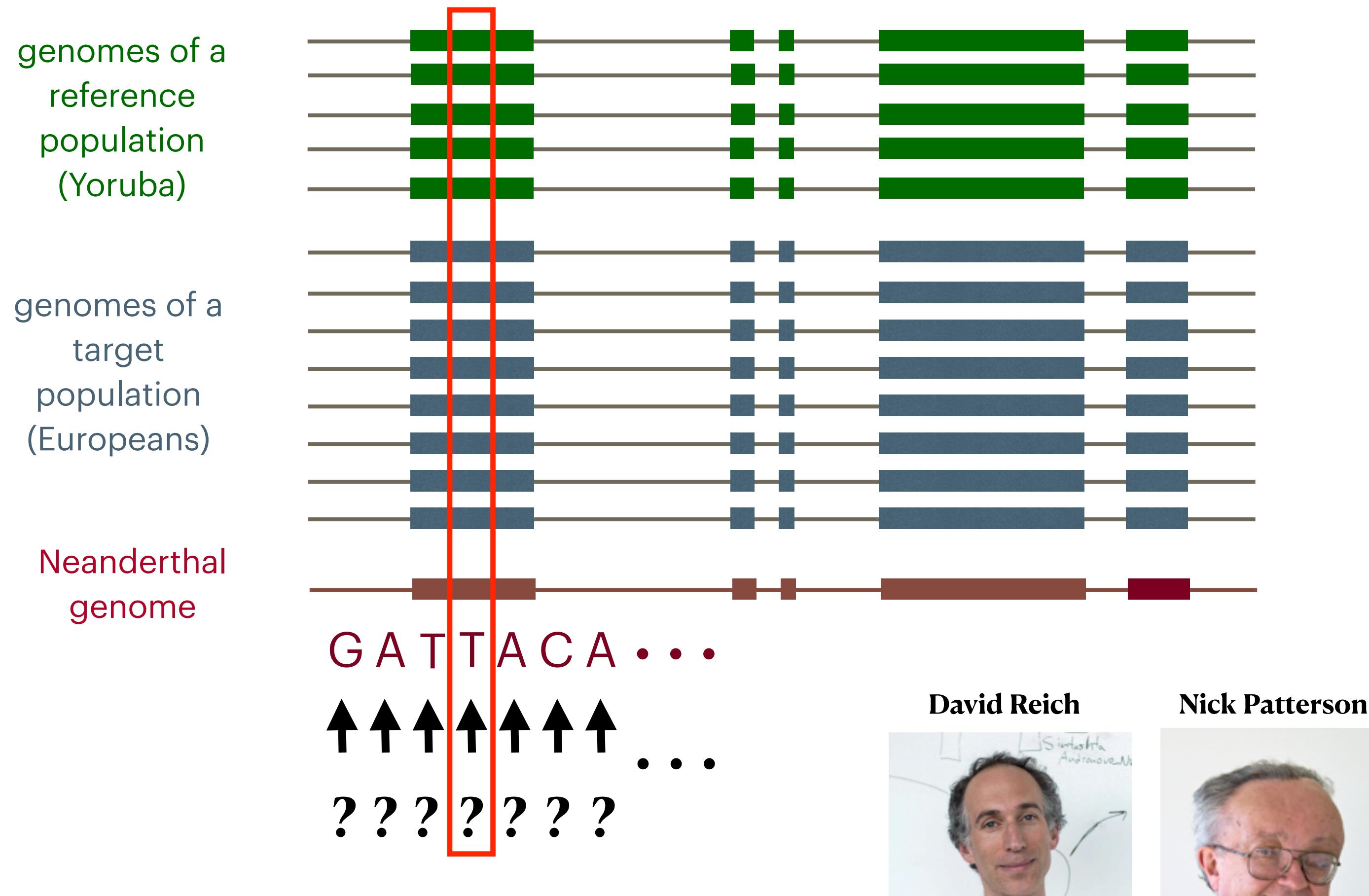


Neanderthal



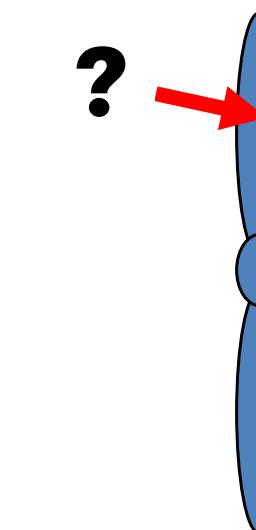
Green et al. (Science 2010)

How to test the gene-flow hypothesis?

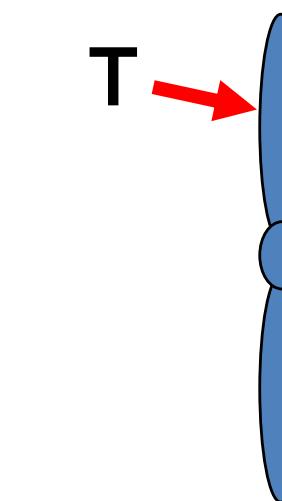
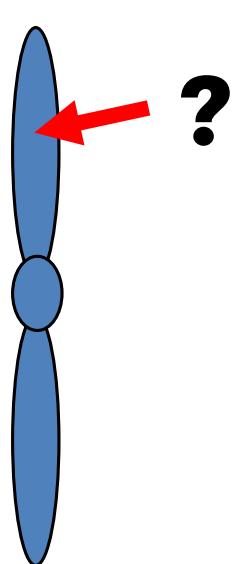


Situation at one locus...

person of an African ancestry



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



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.

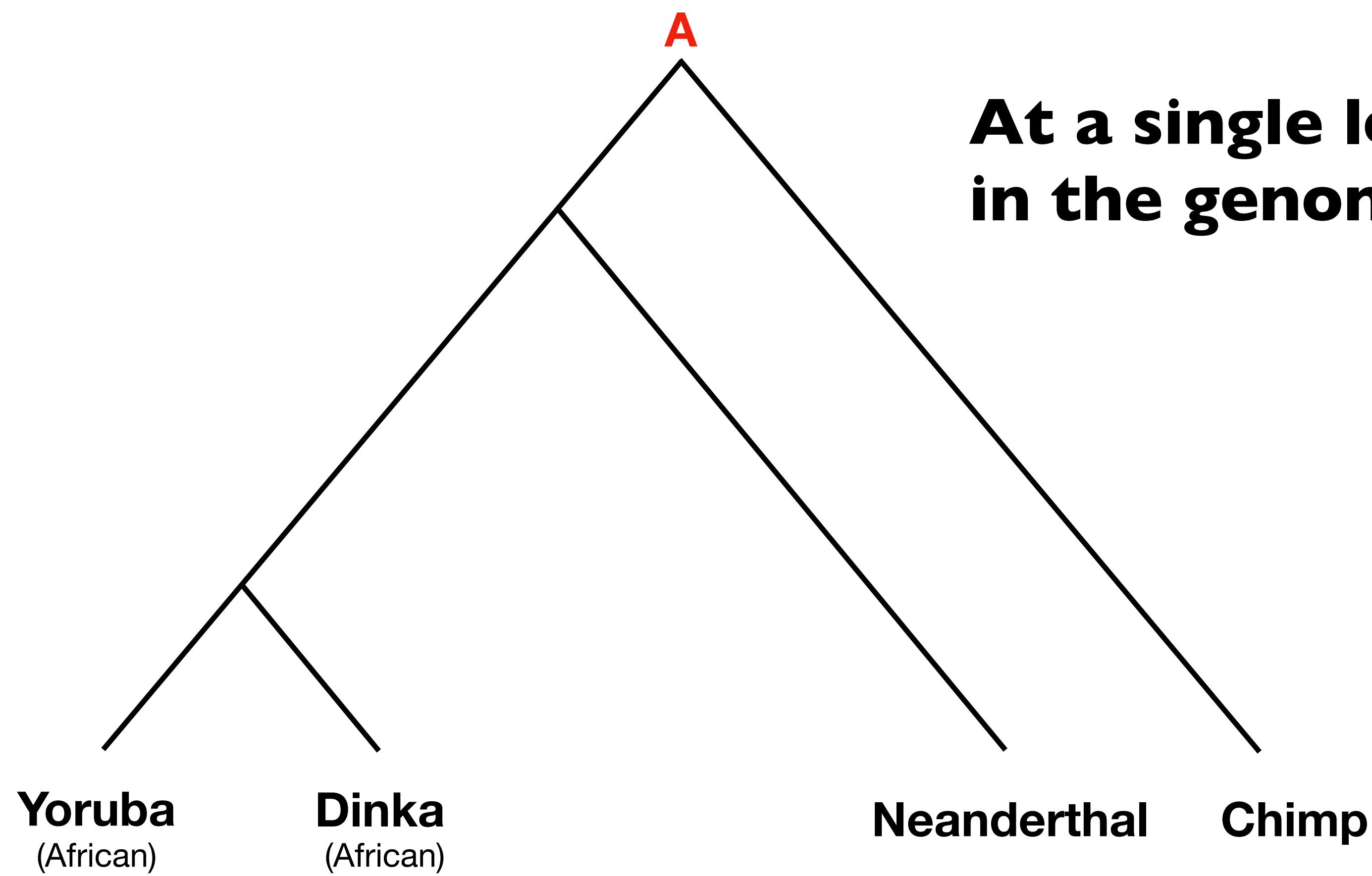
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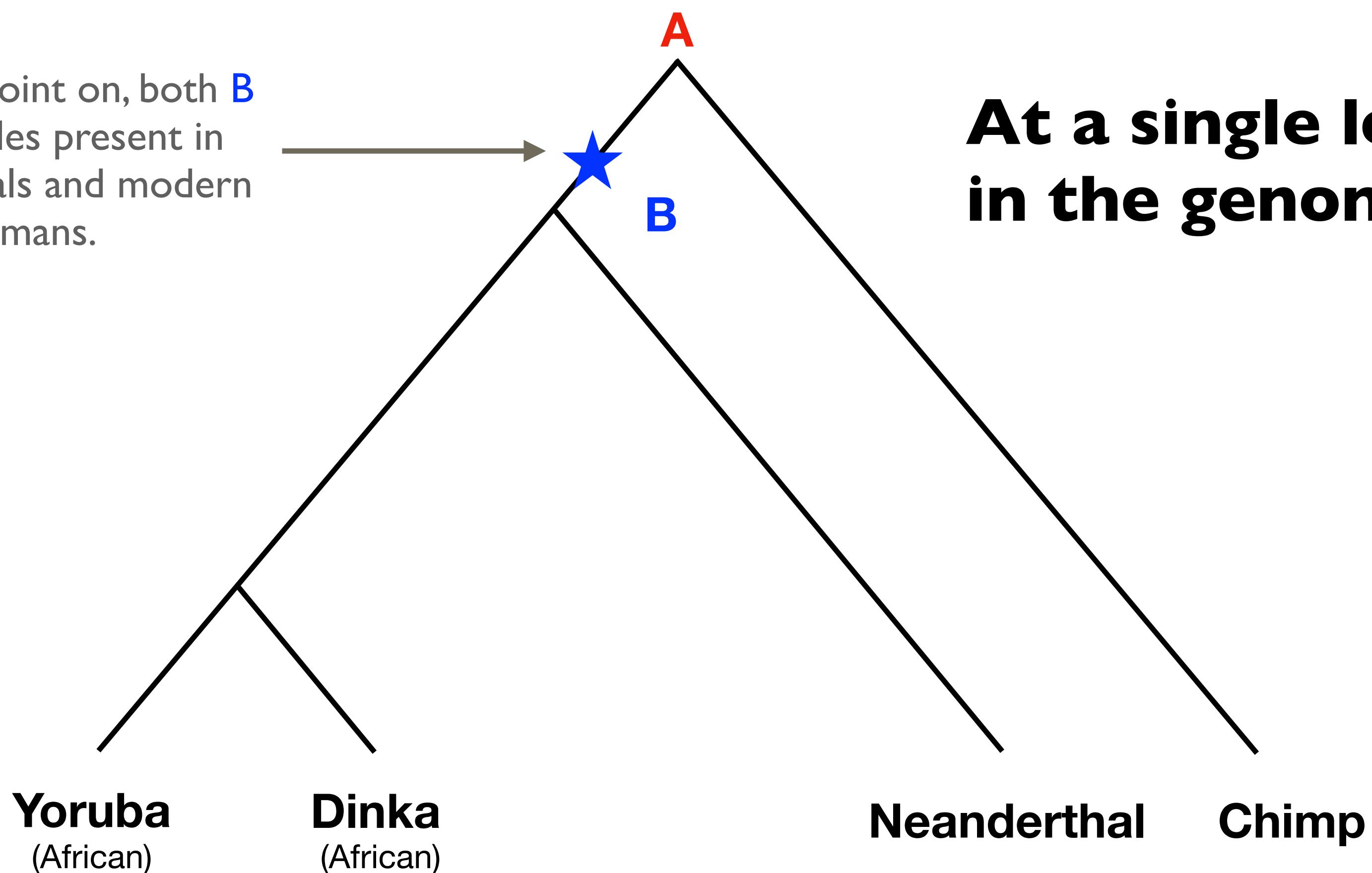


A variant of this statistic that is used equally often is known as **D statistic**.



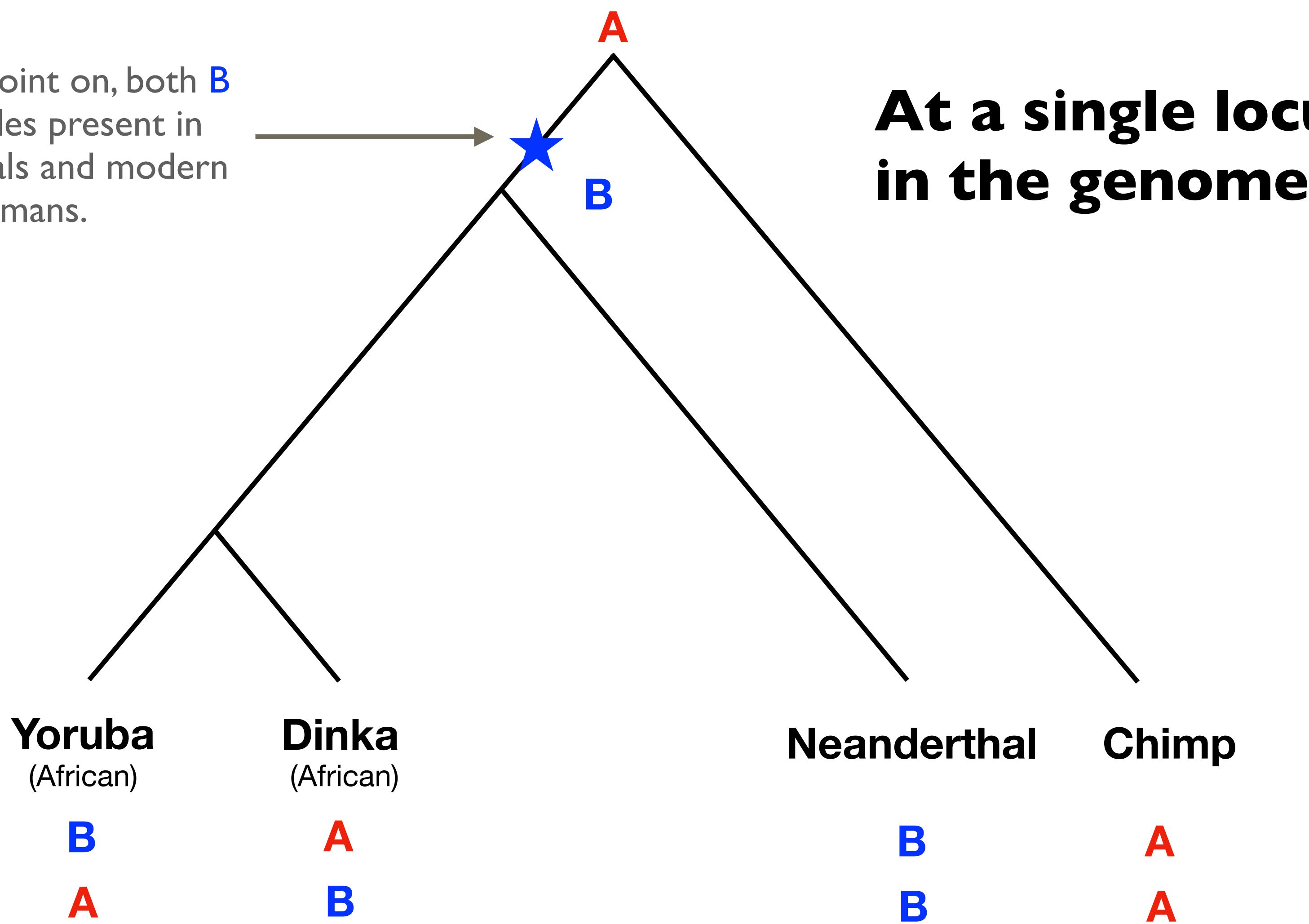
**At a single locus
in the genome...**

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



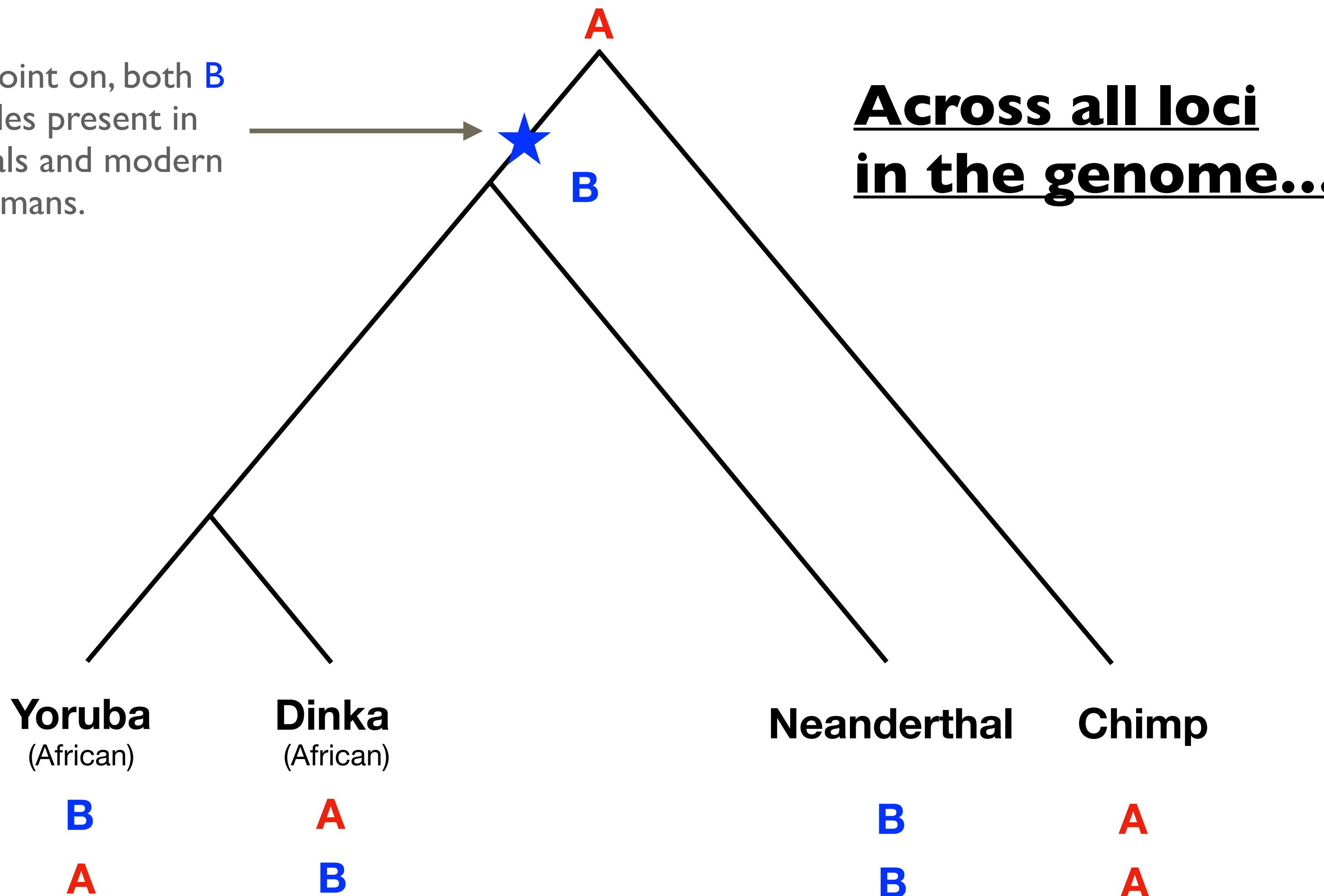
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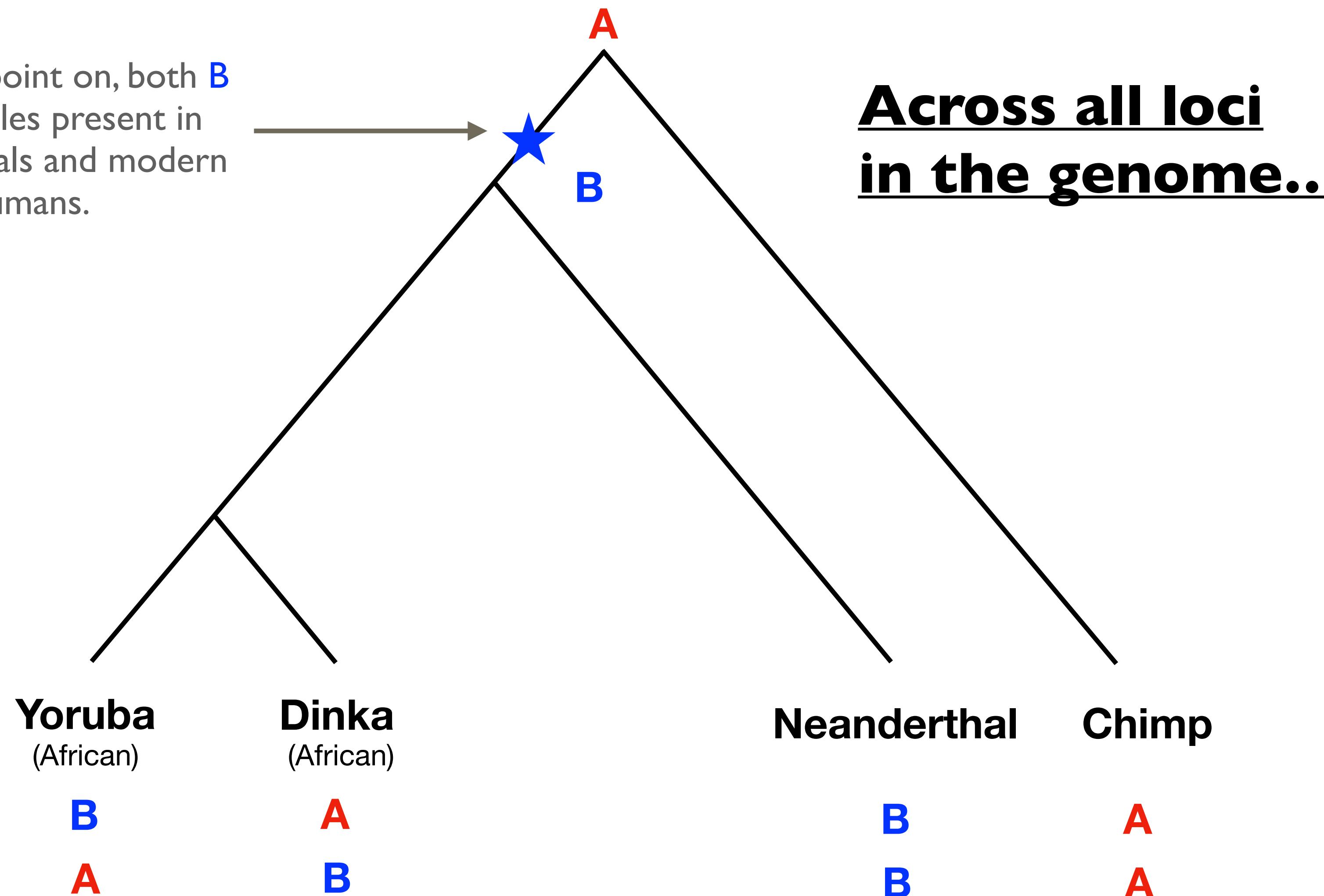
**At a single locus
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From this point on, both **B** and **A** alleles present in Neanderthals and modern humans.



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

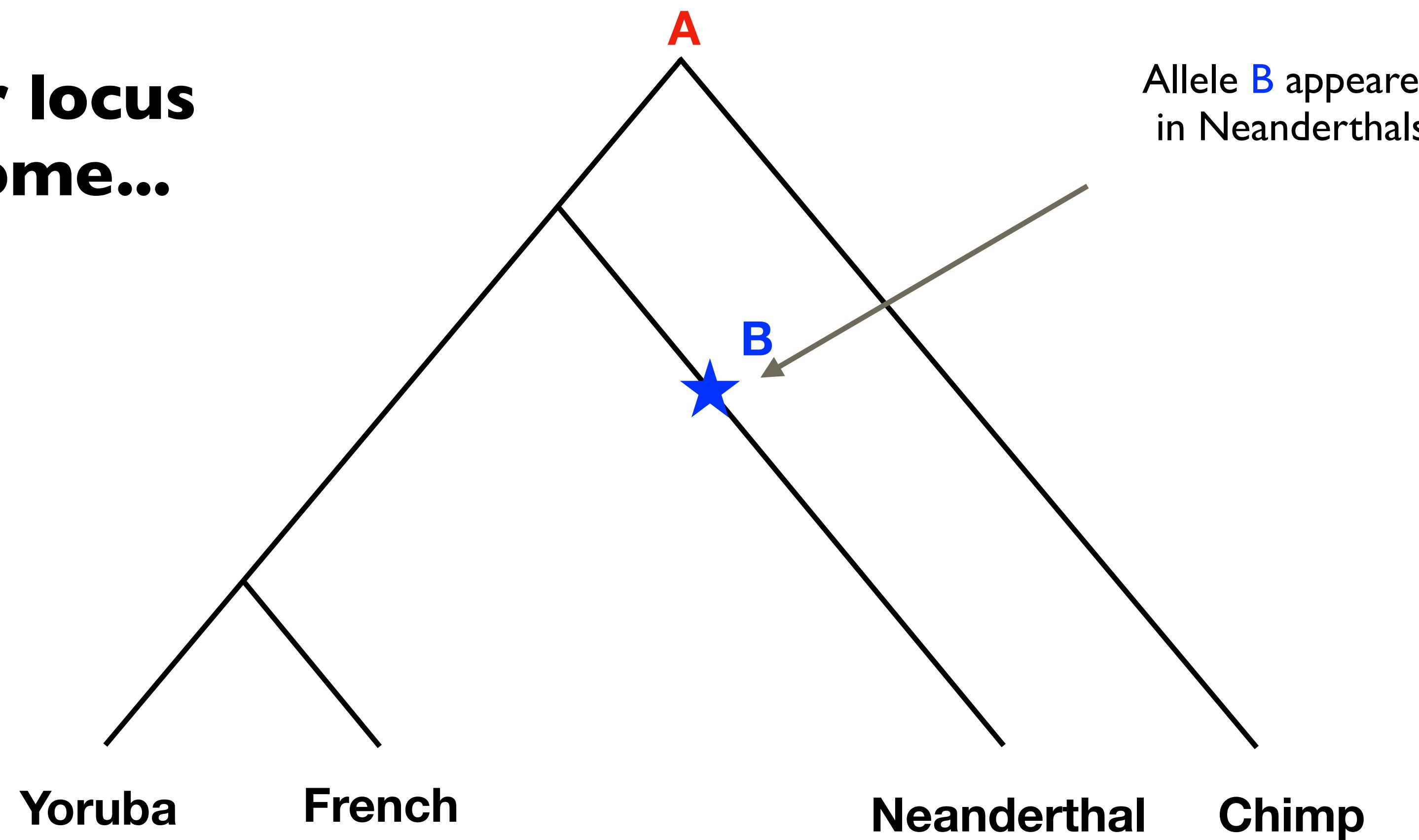
From this point on, both **B** and **A** alleles present in Neanderthals and modern humans.



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

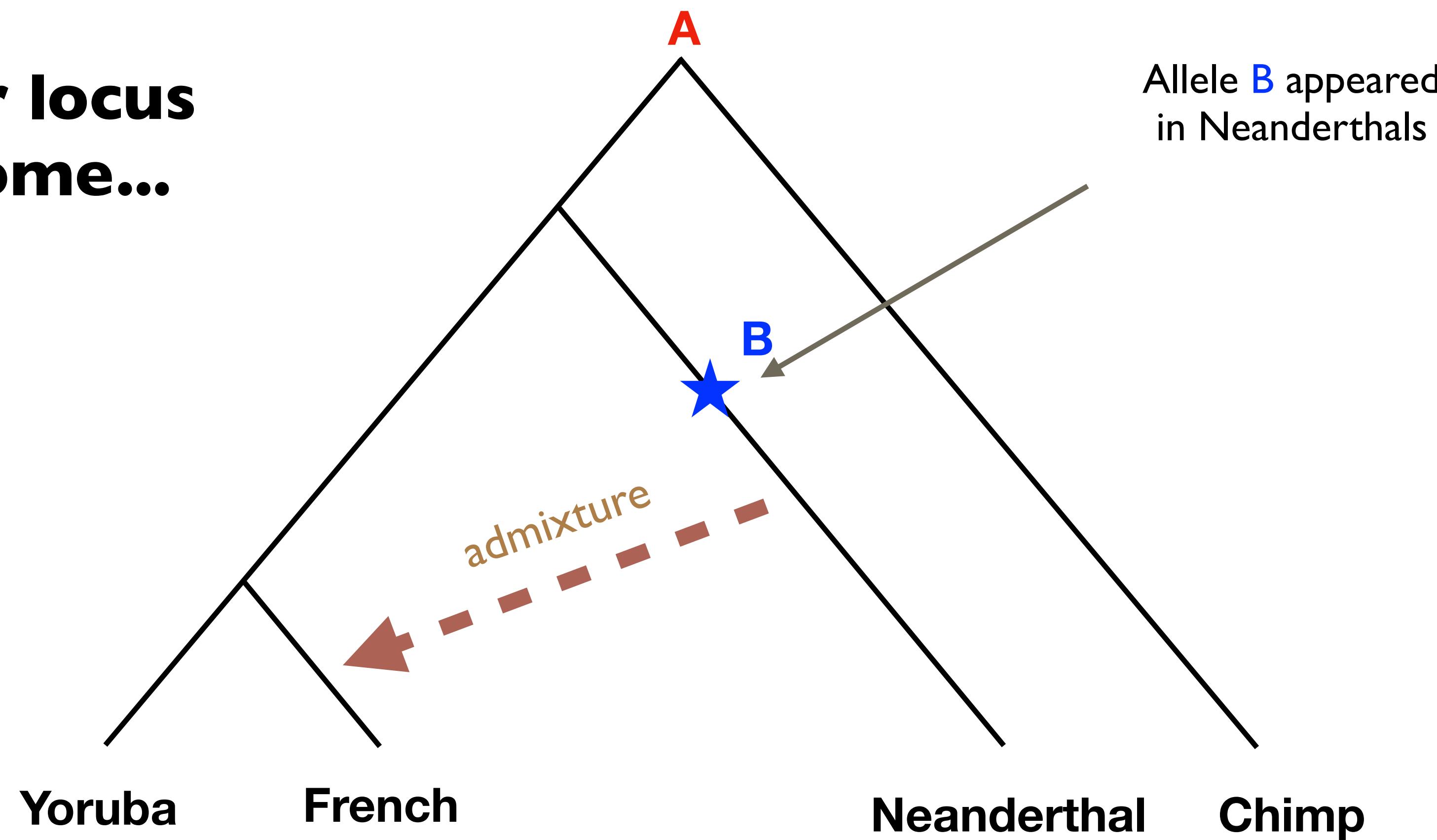
No introgression: *f4 statistic* = (# BABA - #ABBA) / # SNPs ~ 0

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

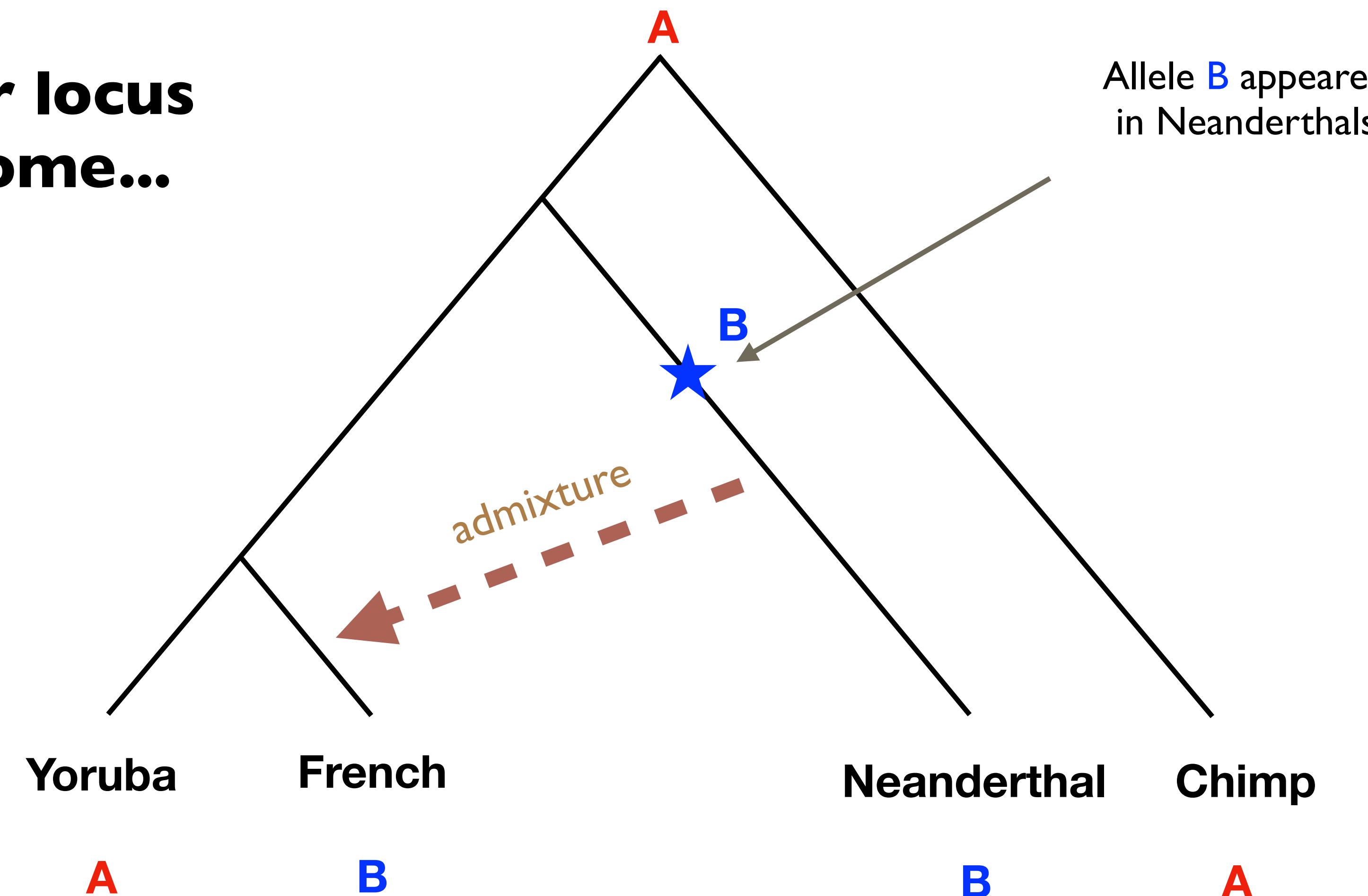


Allele B appeared
in Neanderthals

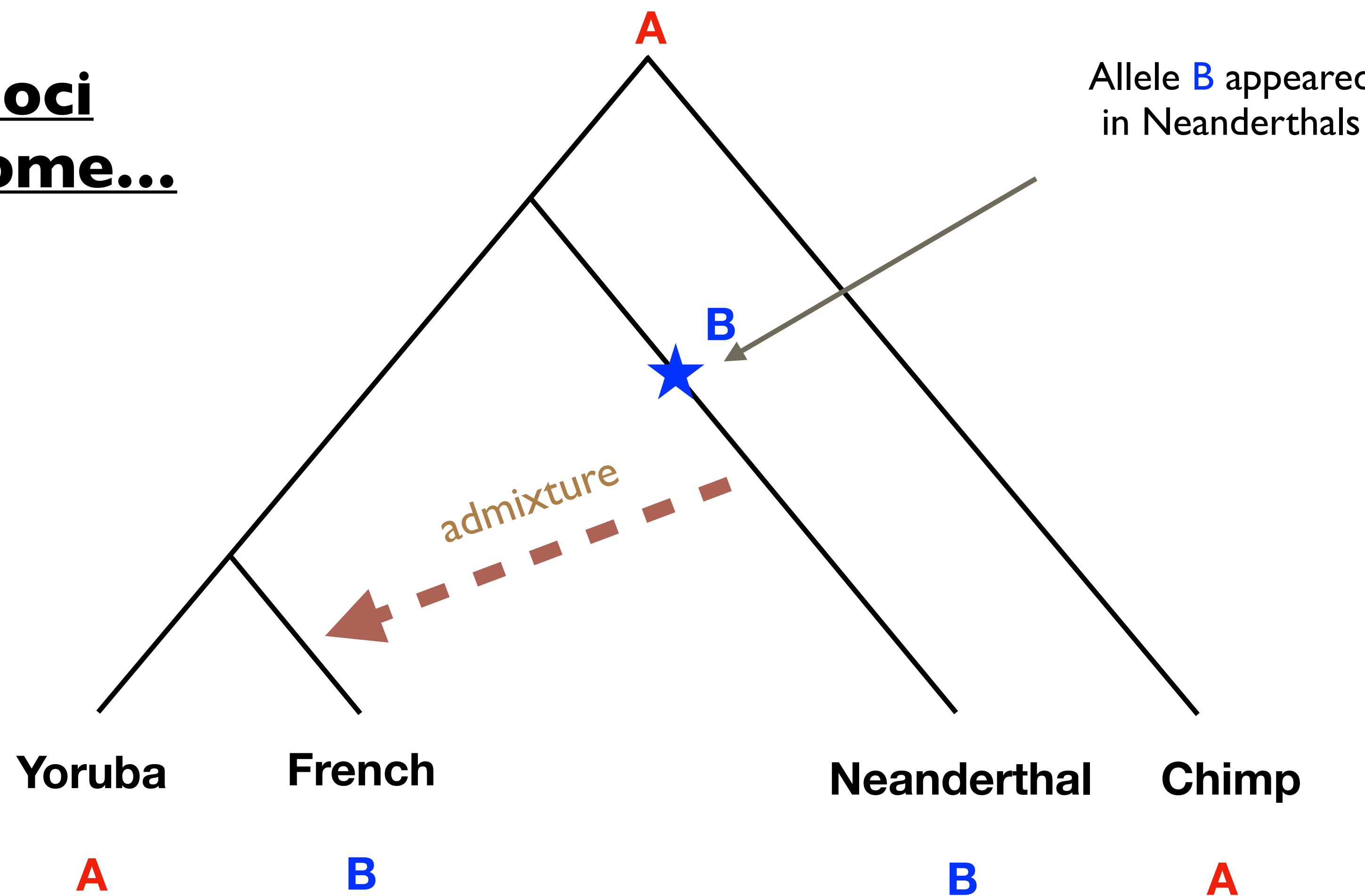
**At another locus
in the genome...**



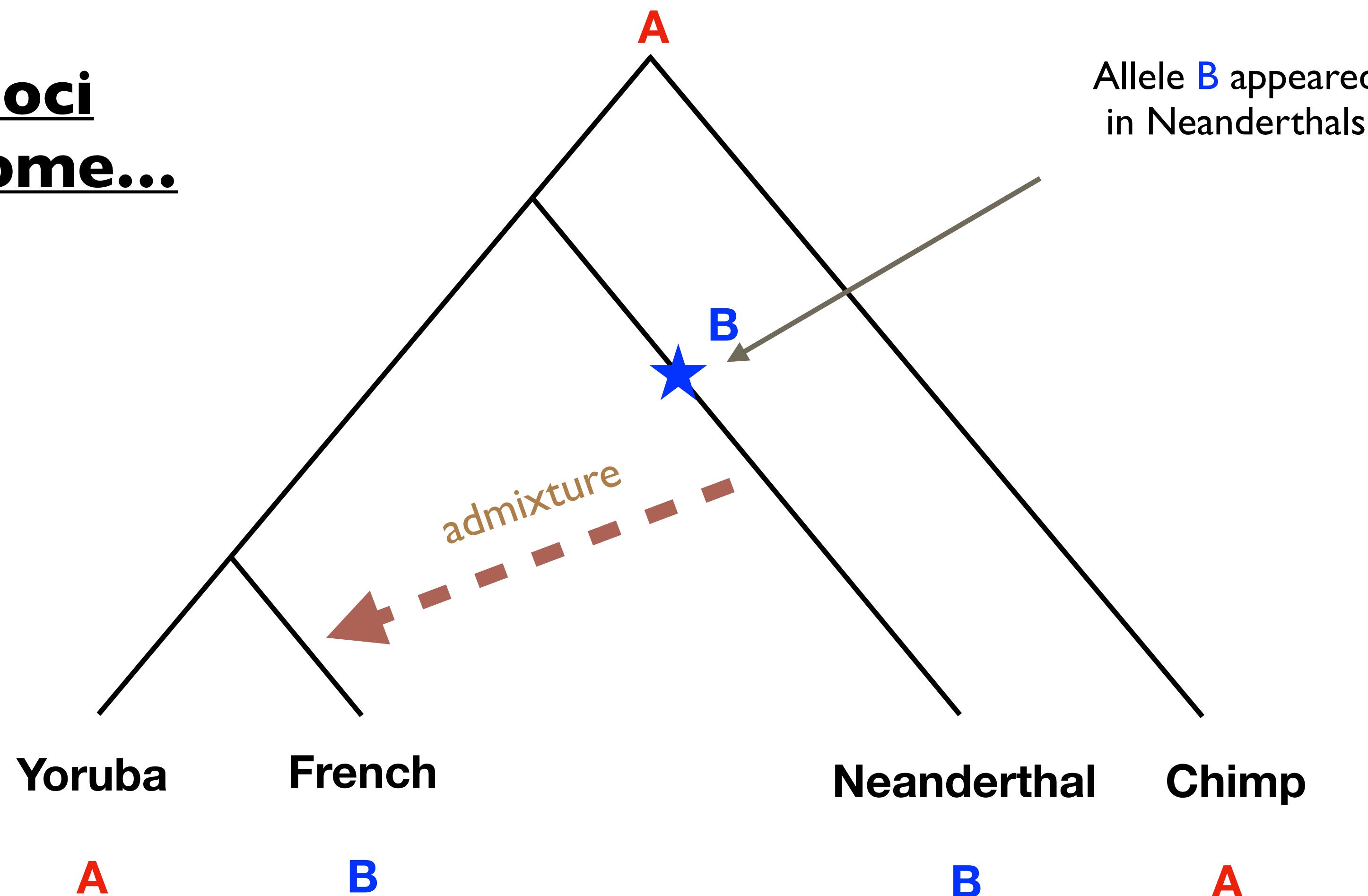
**At another locus
in the genome...**



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

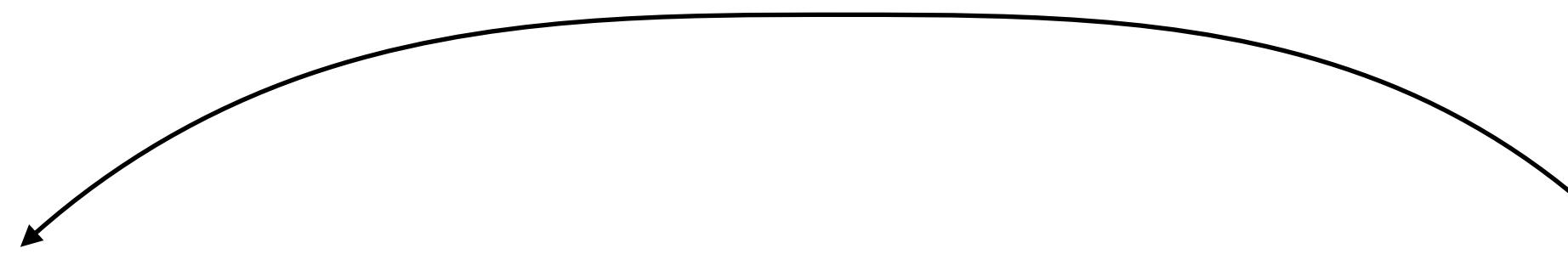


**Across all loci
in the genome...**



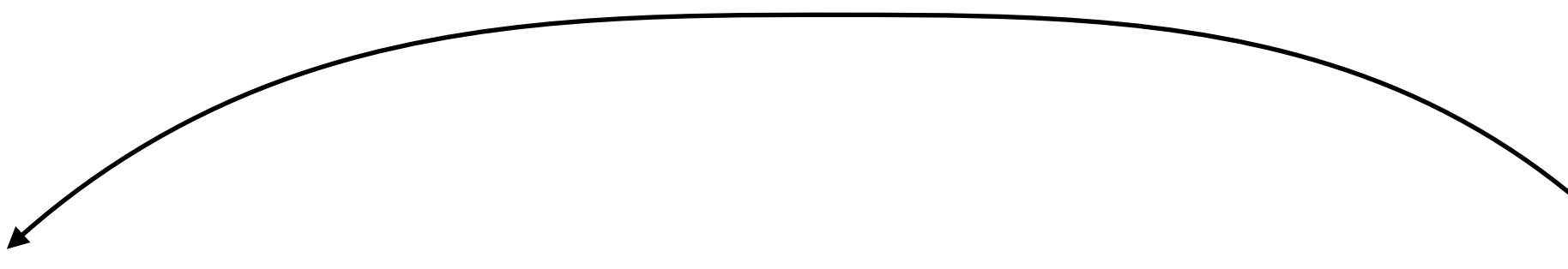
Introgression: $f4$ statistic = (# BABA - # ABBA) / # SNPs < 0

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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$$f_4 = (\# \text{BABA} - \# \text{ABBA}) / \# \text{SNPs}$$

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

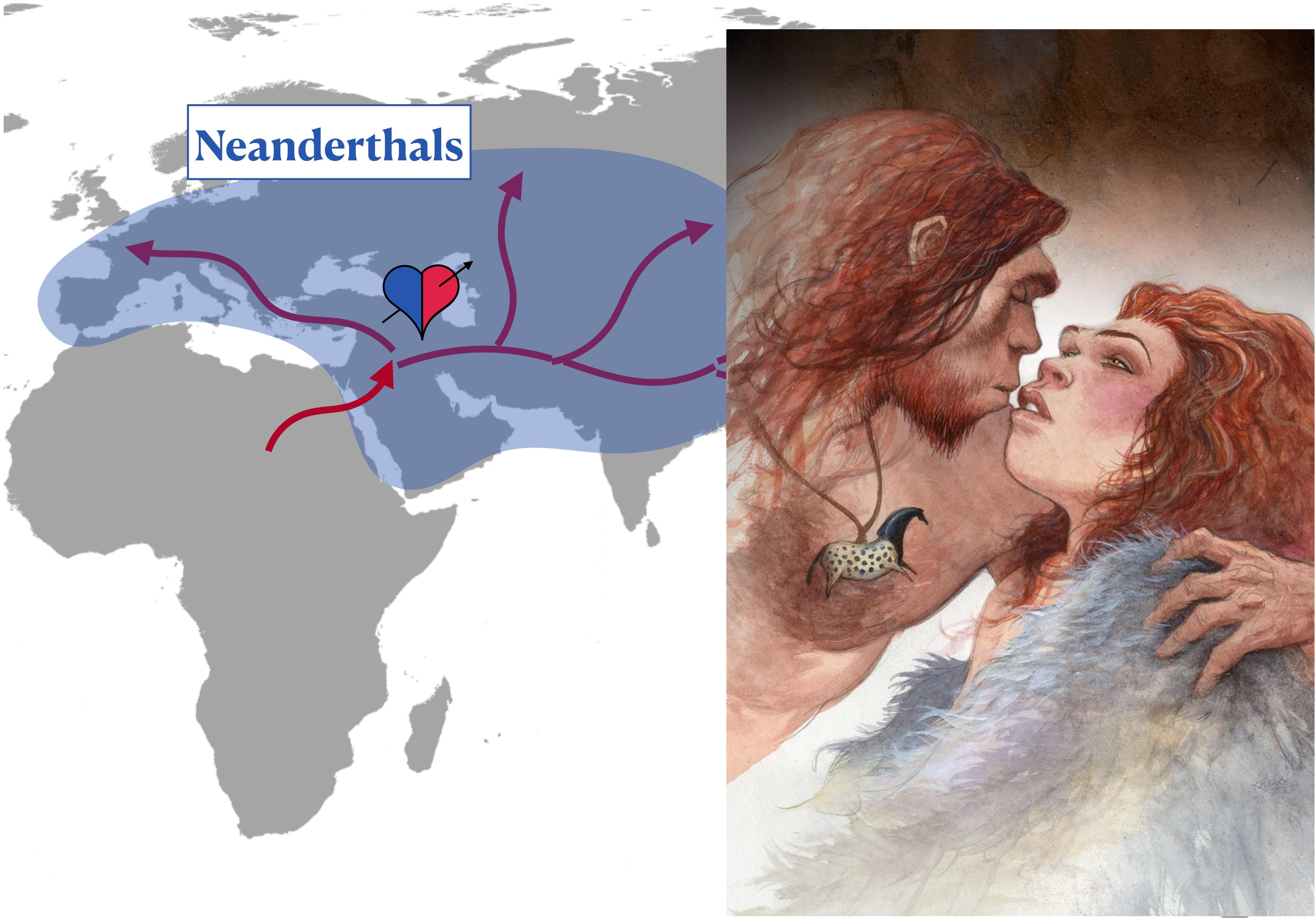
Test of Neanderthal admixture in practice

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$$f_4 = (\# \text{BABA} - \# \text{ABBA}) / \# \text{SNPs}$$

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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French	44409	46865	1436967	-0.0017
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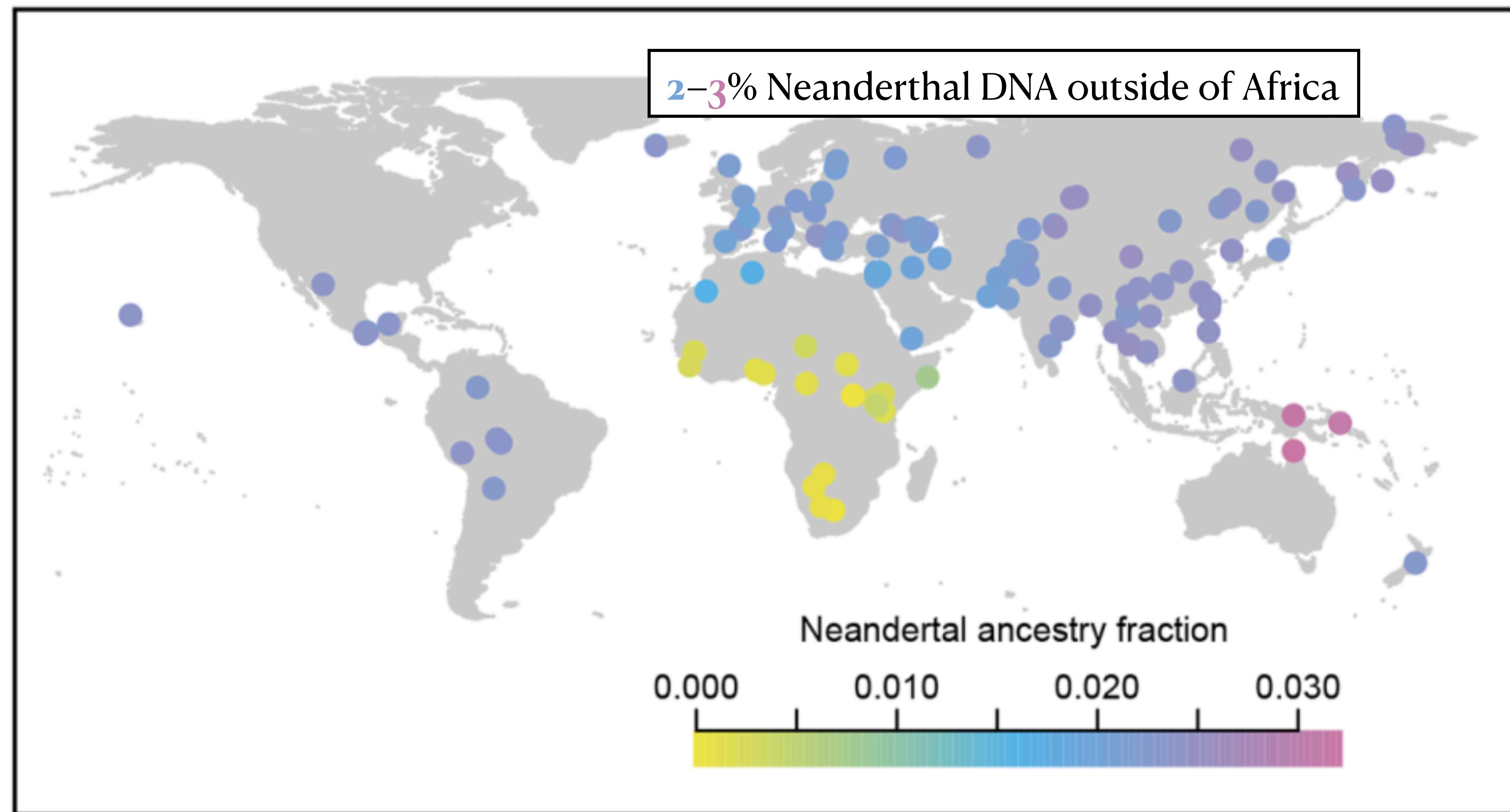
We compare the rate of allele sharing of X and a Neanderthal, with the rate of sharing between two Neanderthals (Altai and Vindija).

$f4(\text{Yoruba}, \mathbf{X}; \text{Altai}, \text{Chimp}) / f4(\text{Yoruba}, \mathbf{Vindija}; \text{Altai}, \text{Chimp})$

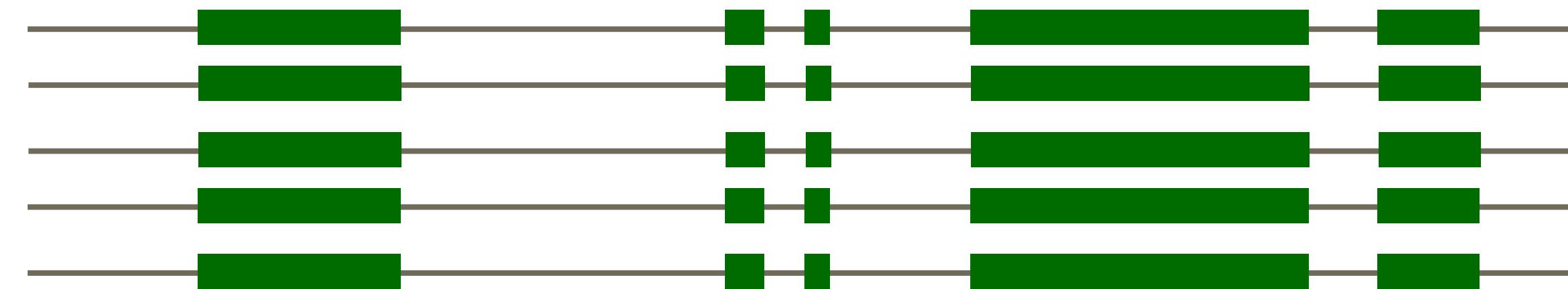


X	#BABA	#ABBA	#sites	f4	ratio
French	44409	46865	1436967	-0.0017	2.2%
Dinka	43025	43182	1436978	-0.0001	0.1%
Vindija	7337	118956	1436703	-0.0777	100%

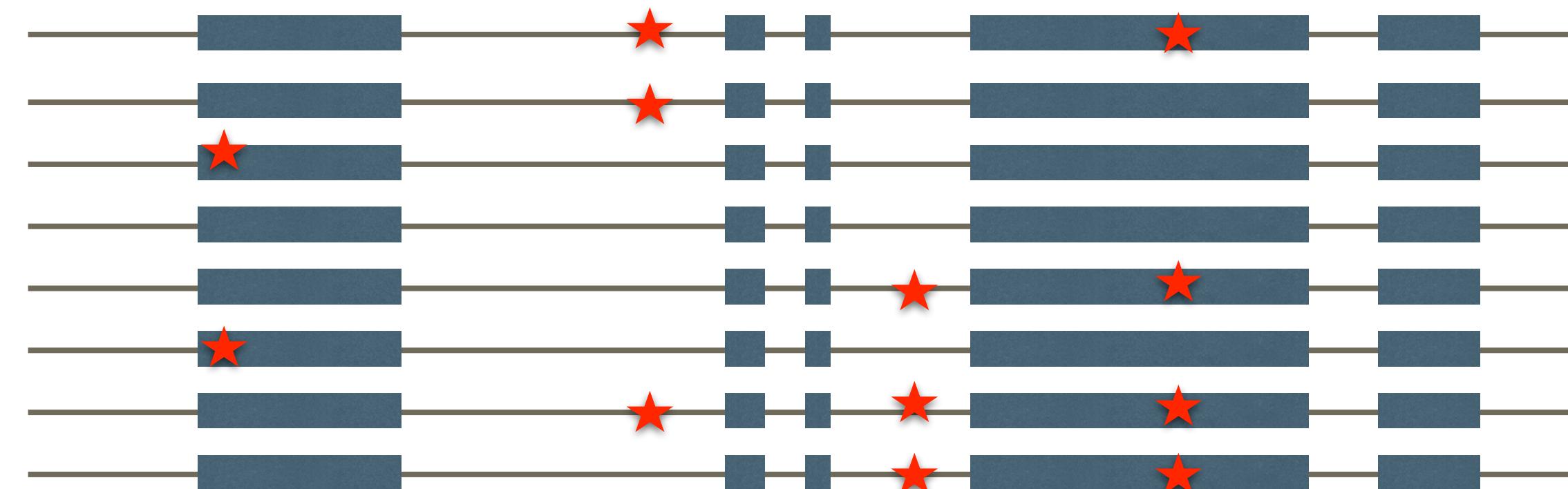
Proportion of Neanderthal DNA in humans today



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



genomes of a
reference population
(Yoruba)



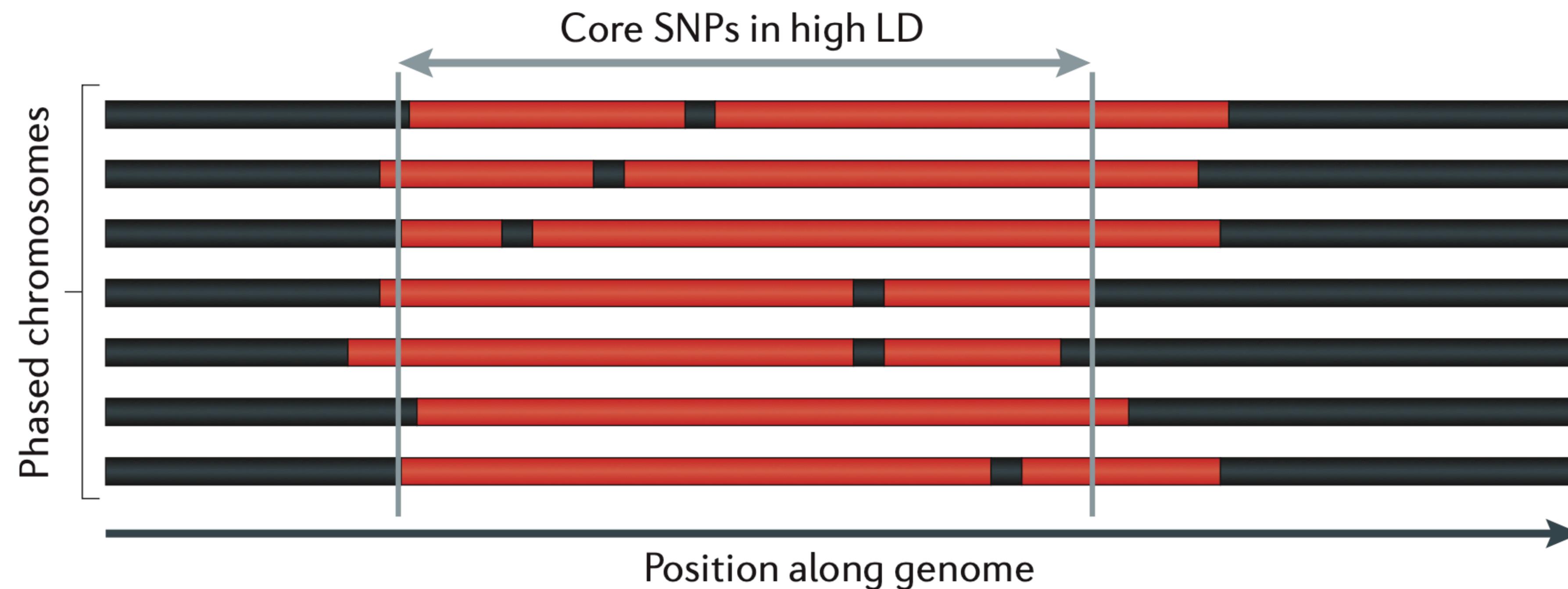
genomes of a
target population
(Europeans)



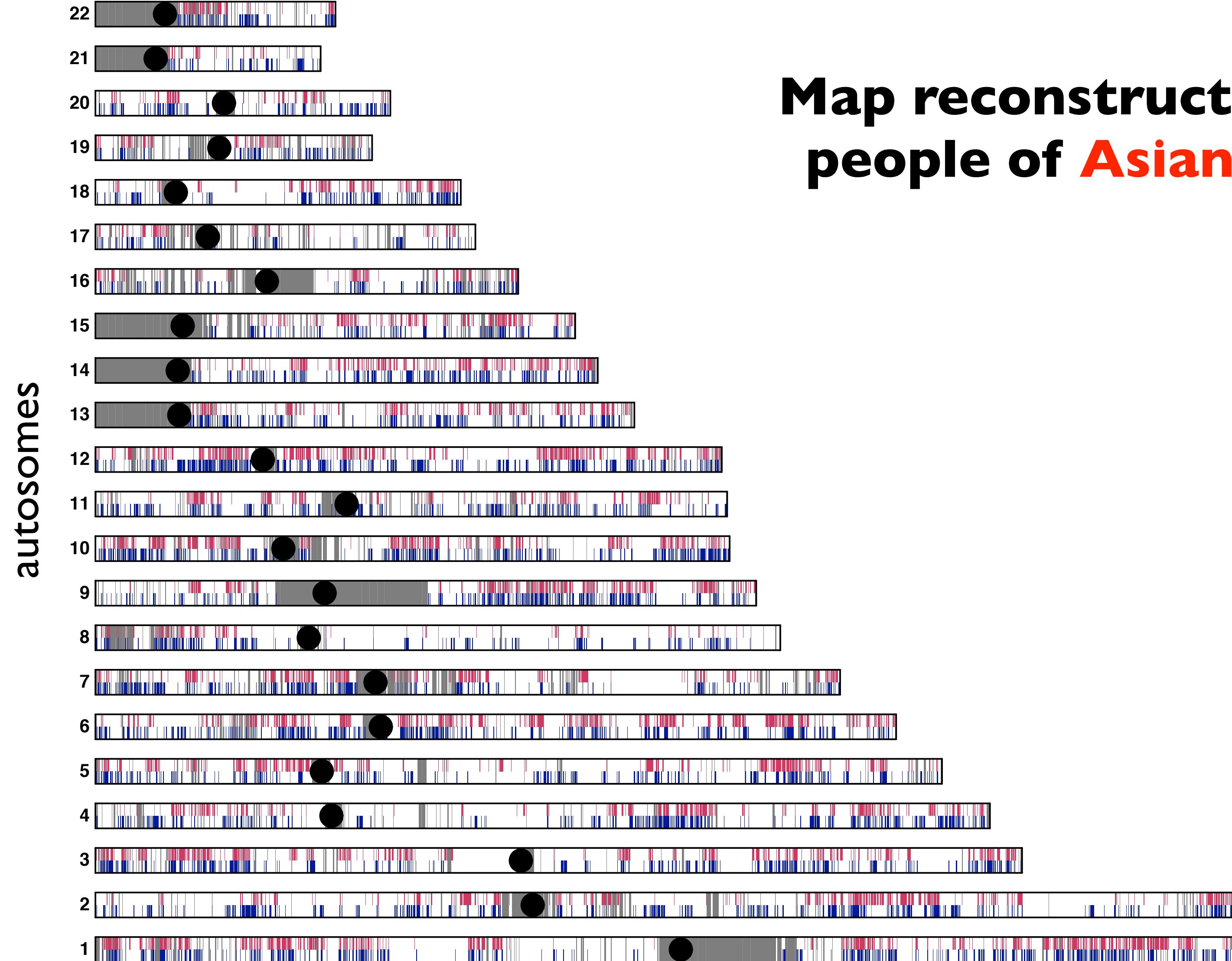
Neanderthal
genome

★ = Neanderthal-derived mutation

...to continuous segments of Neanderthal DNA

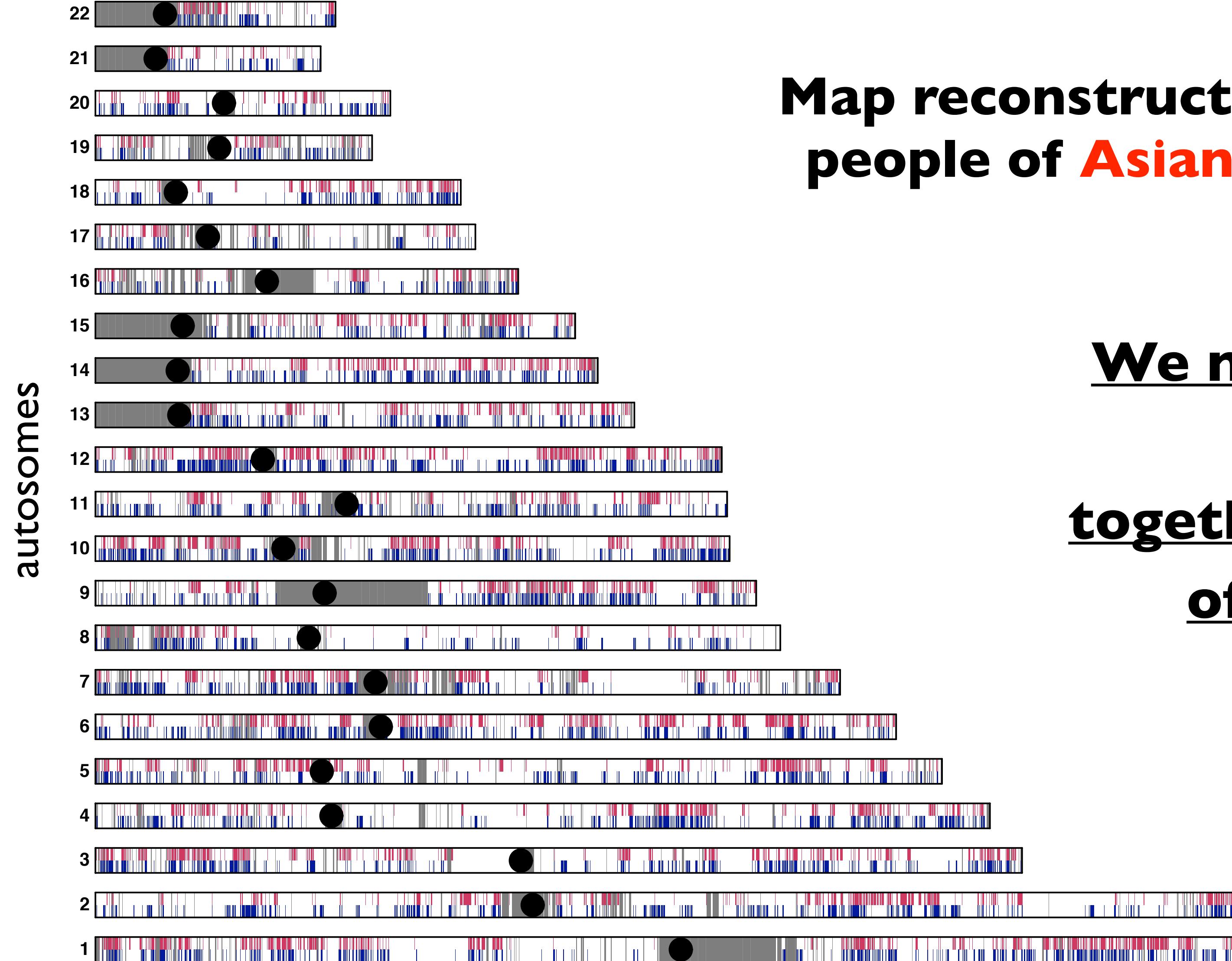


Genomic maps of Neanderthal DNA today



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

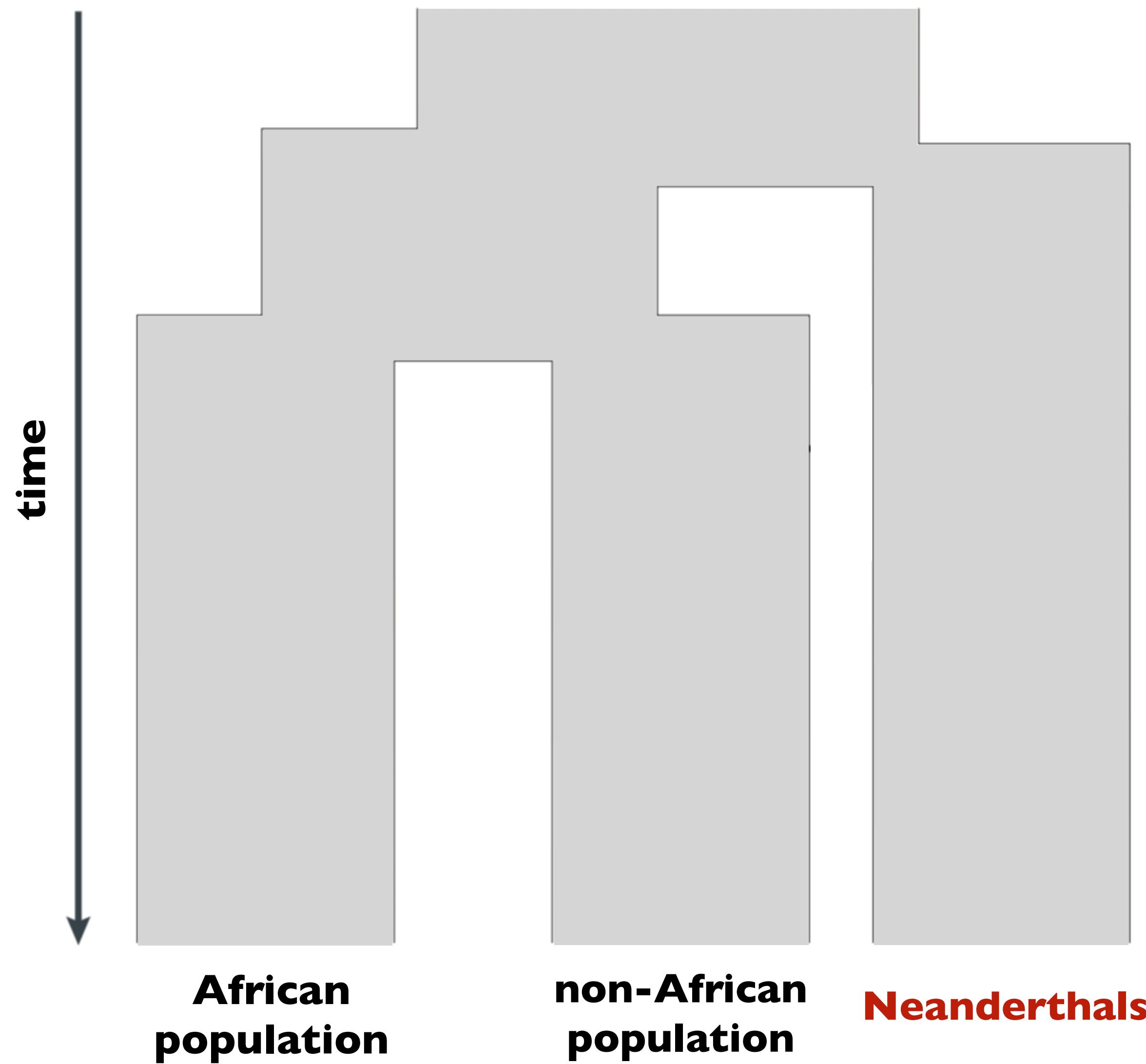
Genomic maps of Neanderthal DNA today



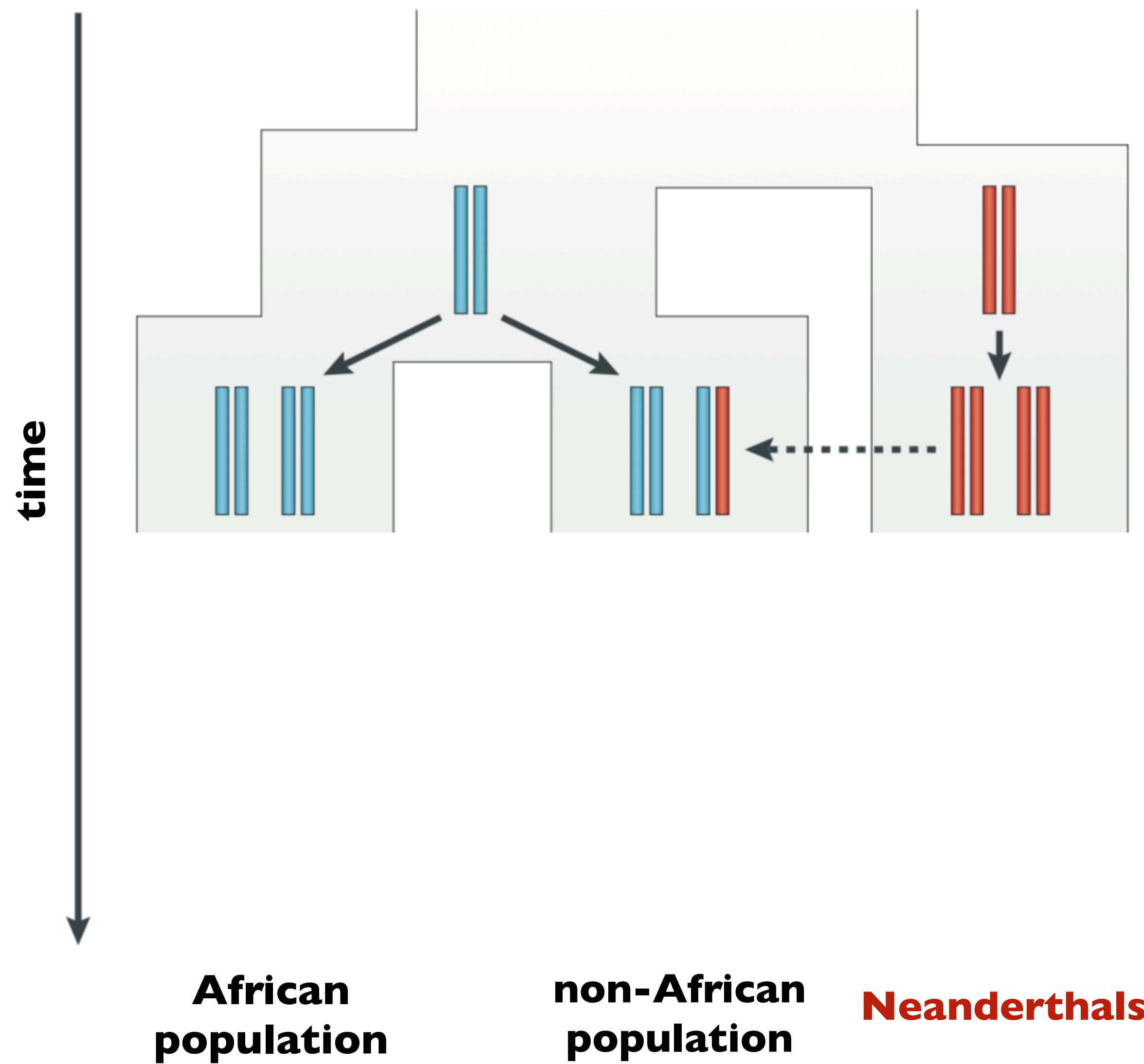
Map reconstructed from genomes of 700 people of **Asian and **European** ancestry.**

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

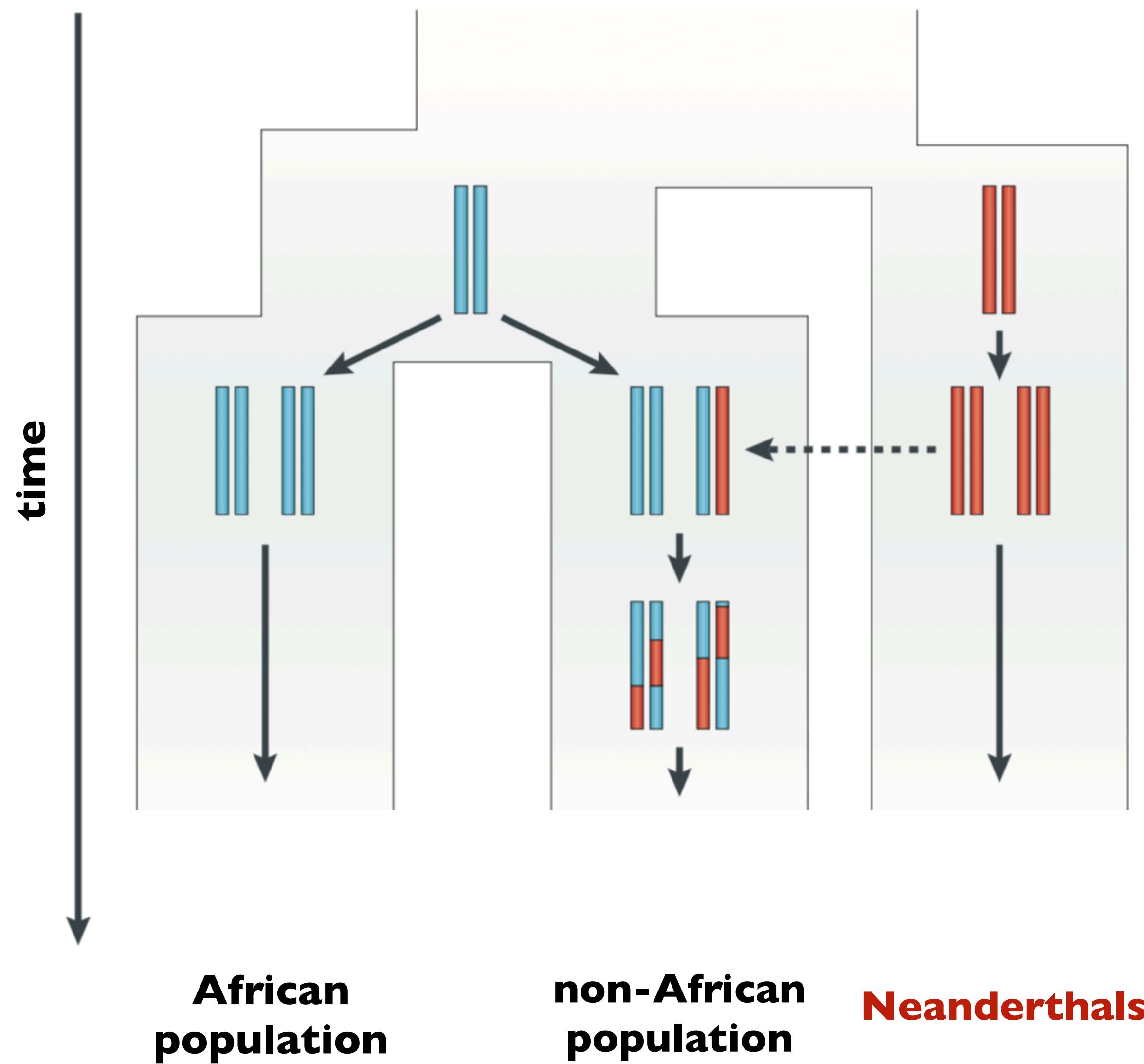
When did introgression happen?



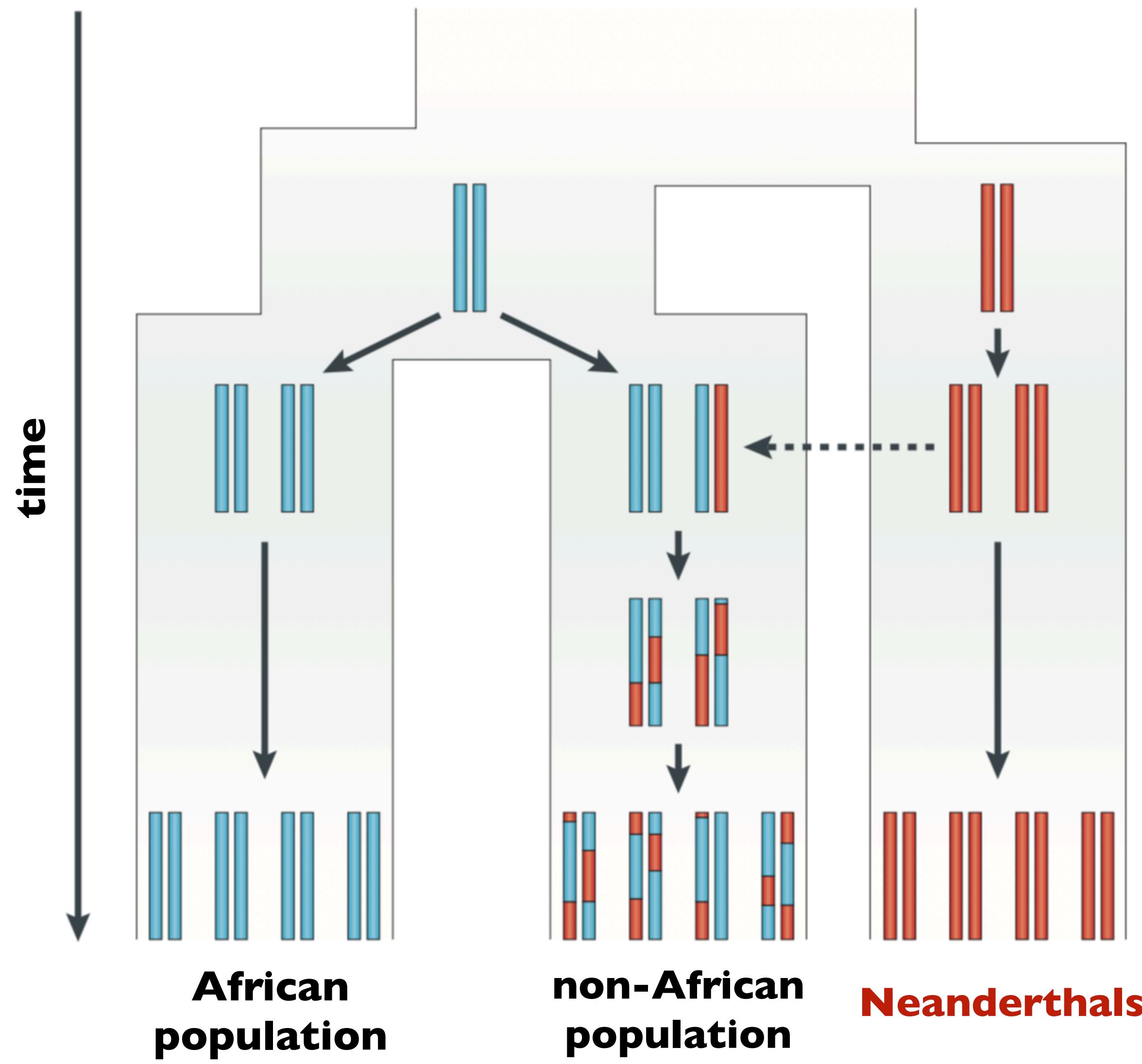
When did introgression happen?



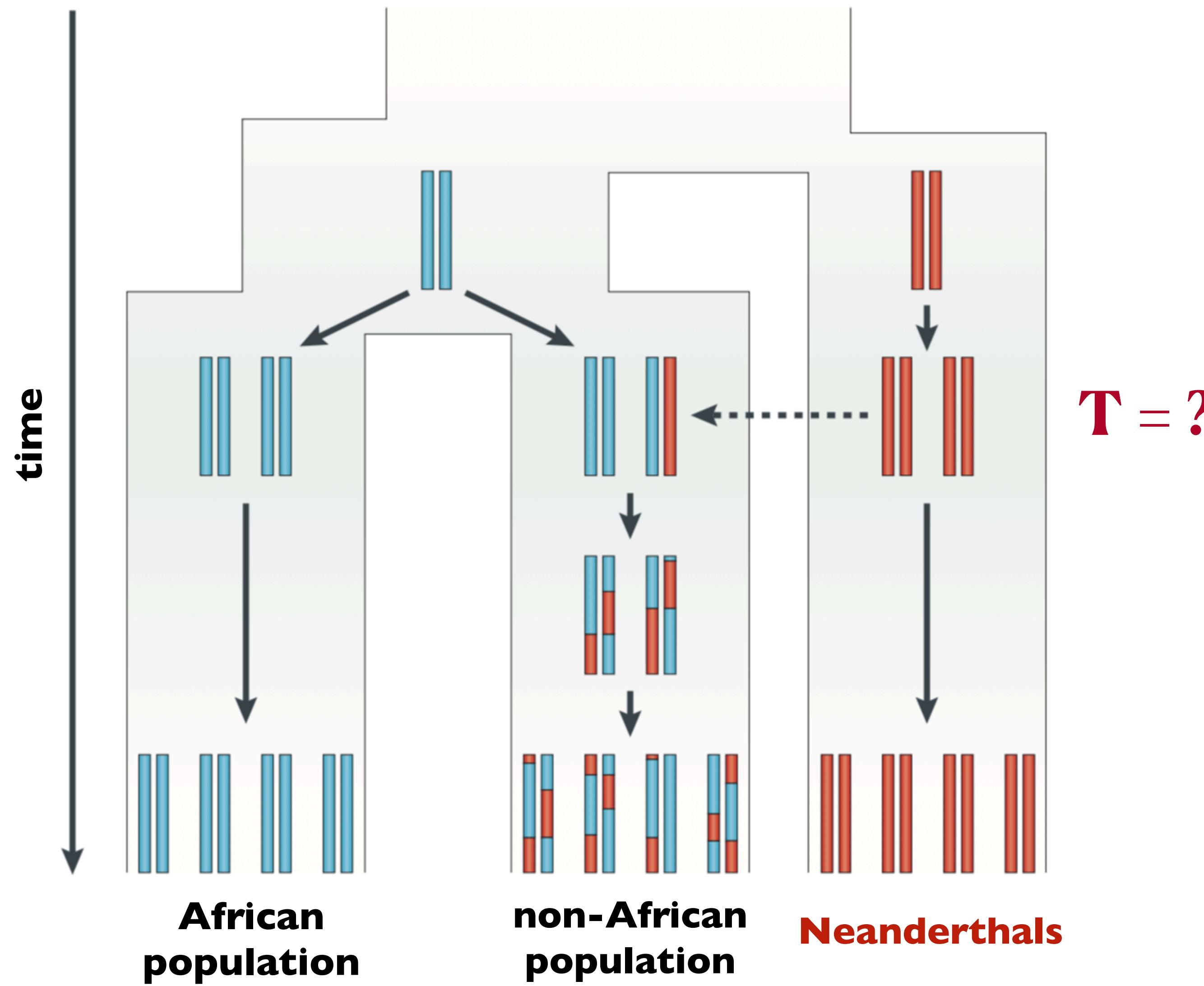
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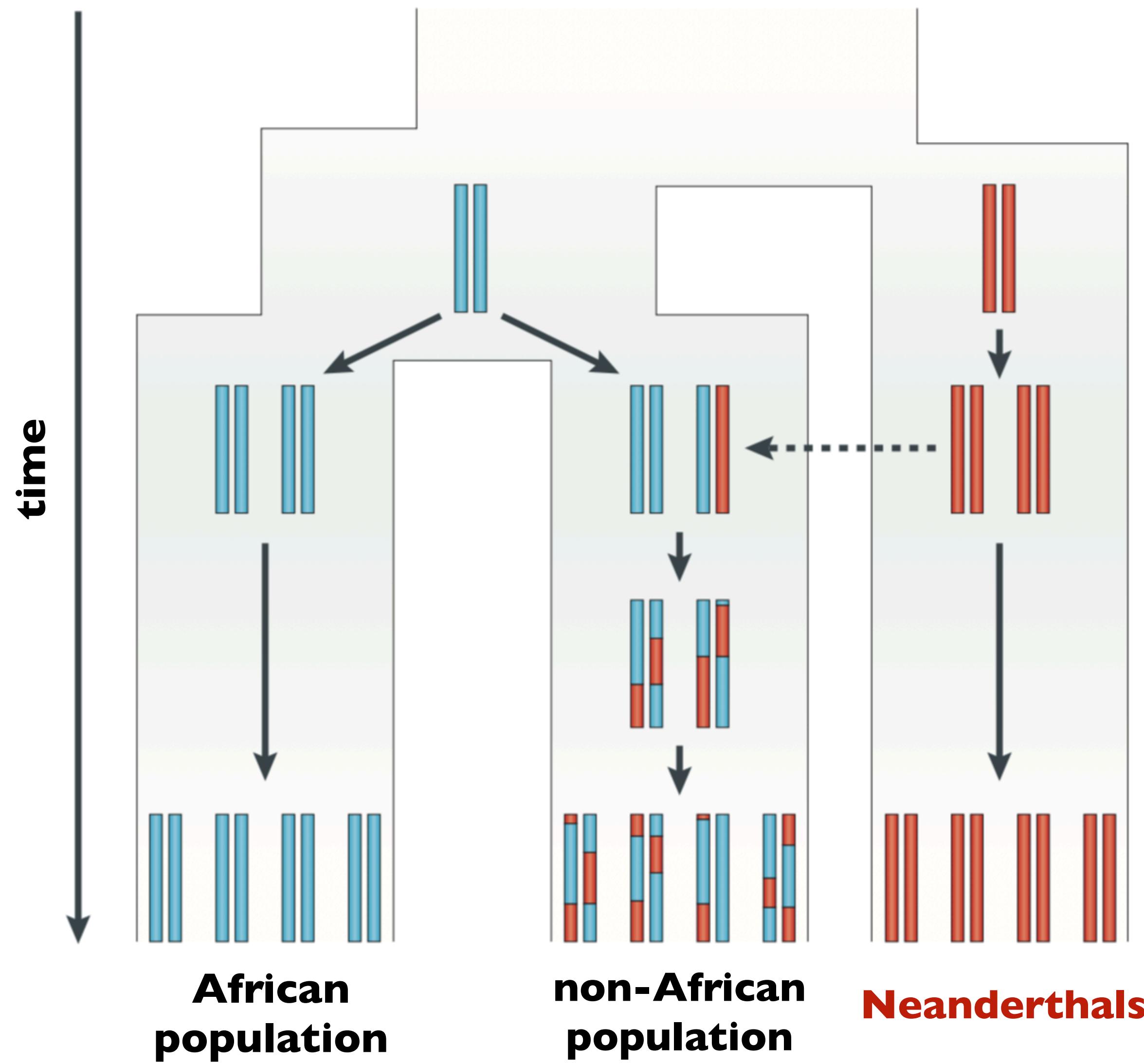
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When did introgression happen?



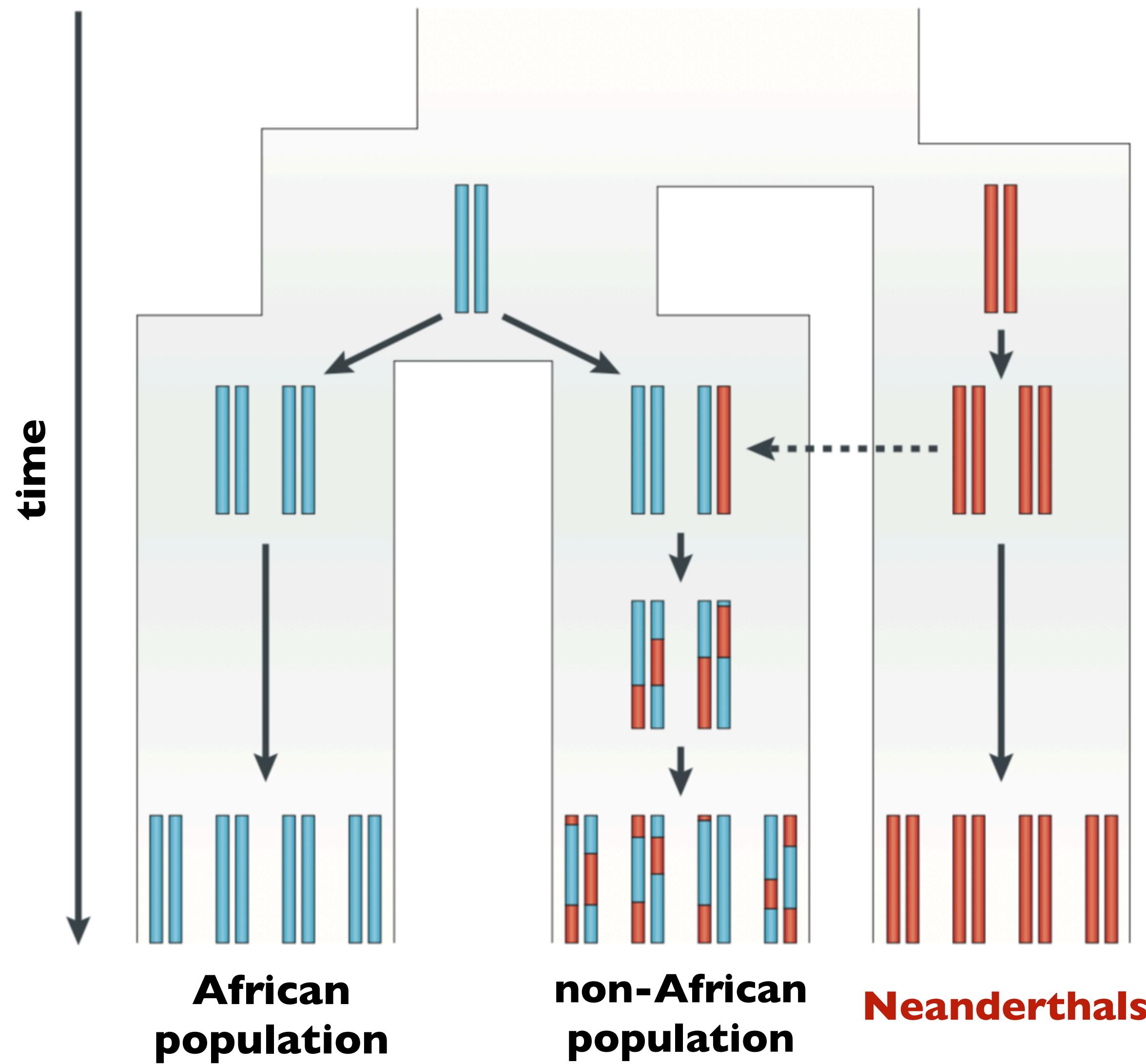
When did introgression happen?



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

T = ?

When did introgression happen?

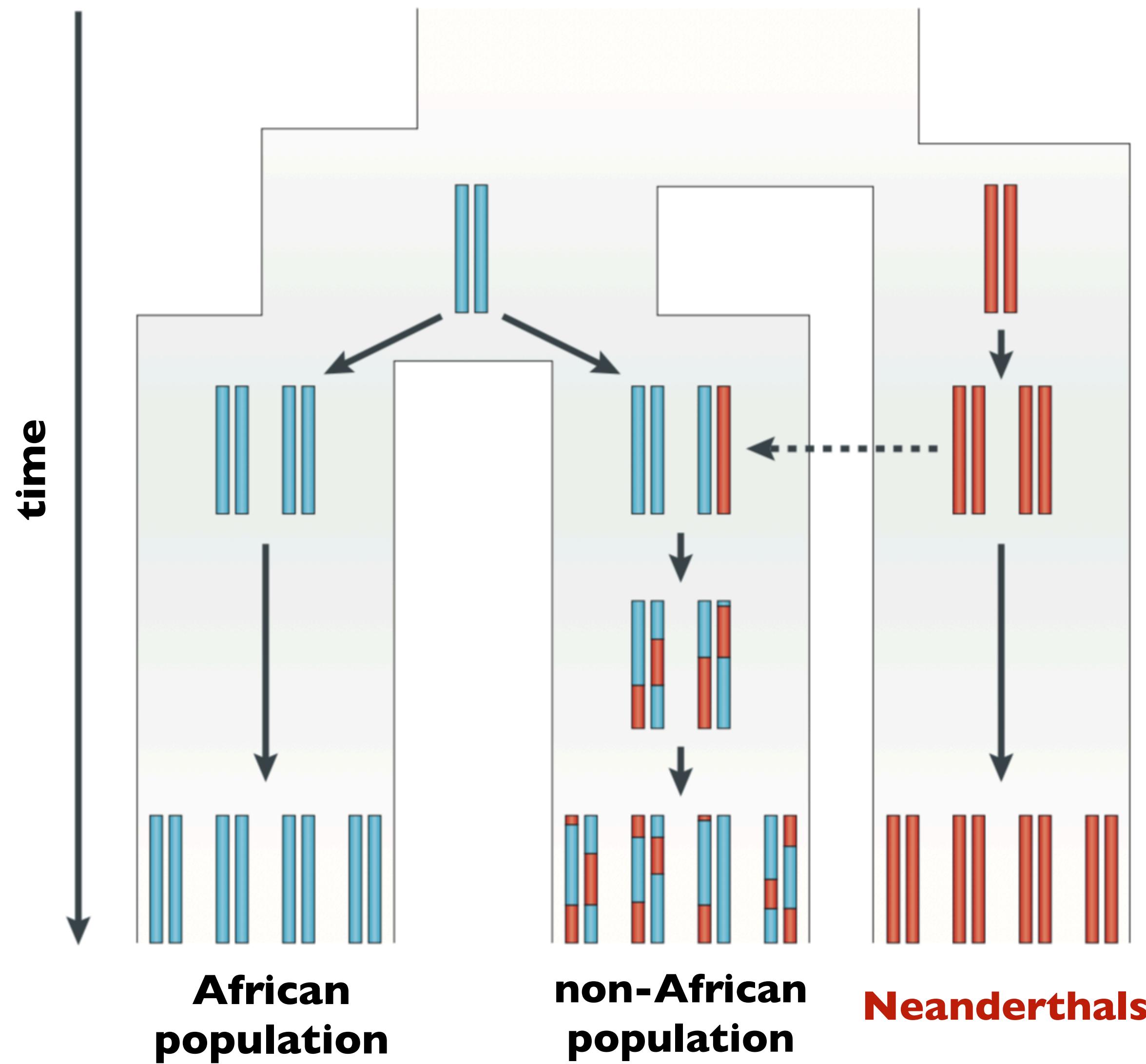


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Expected length of these haplotypes follows the principle of exponential decay.

When did introgression happen?



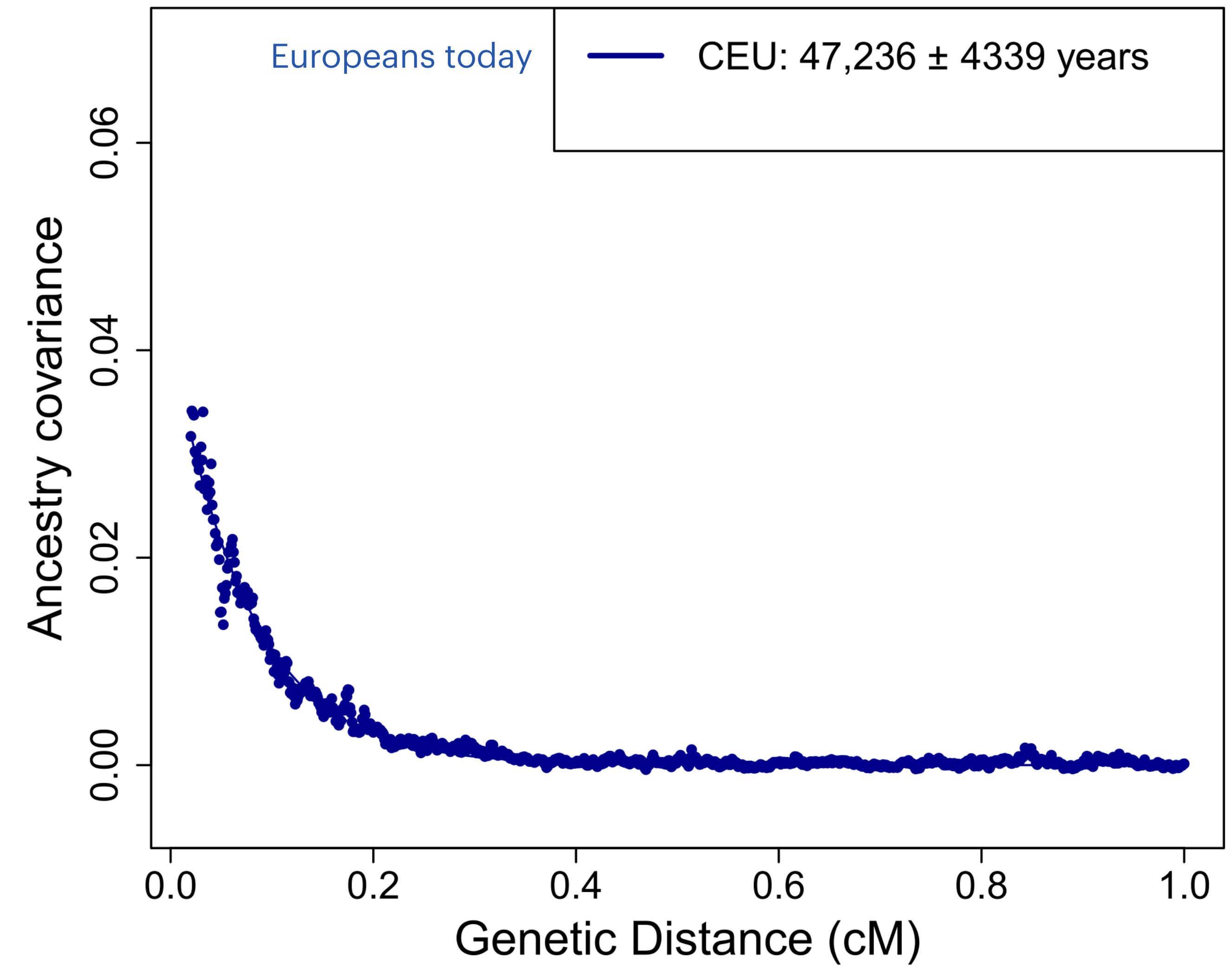
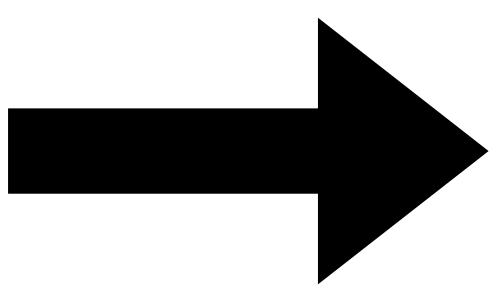
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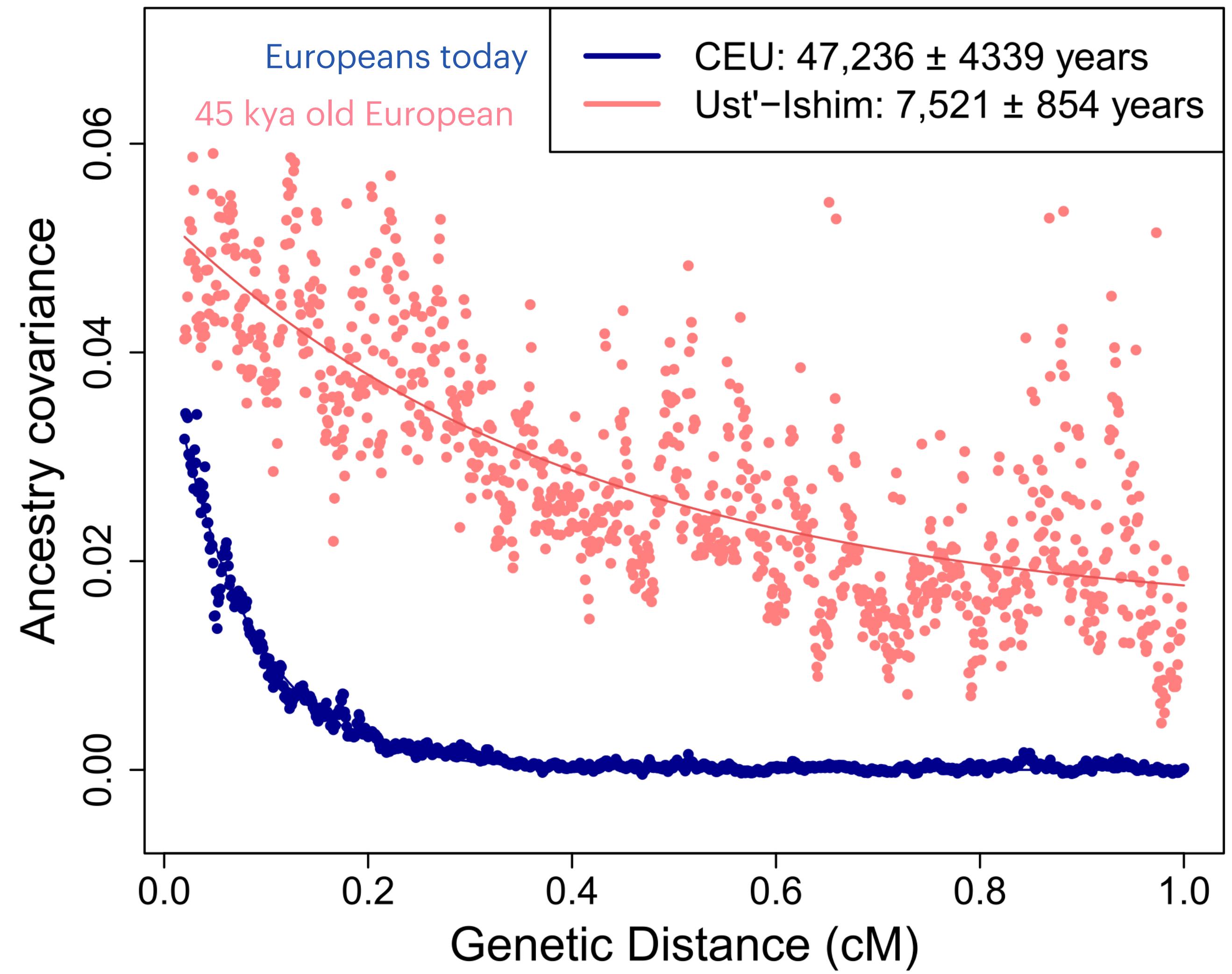
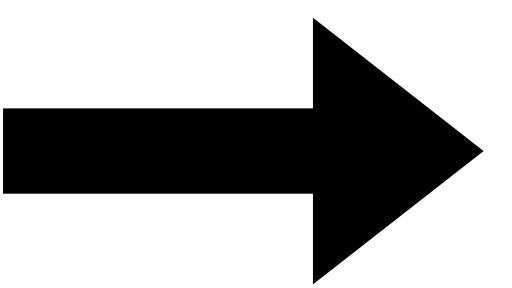
Expected length of these haplotypes follows the principle of exponential decay.

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

Introgression happened around 50 thousand years ago



Introgression happened around 50 thousand years ago

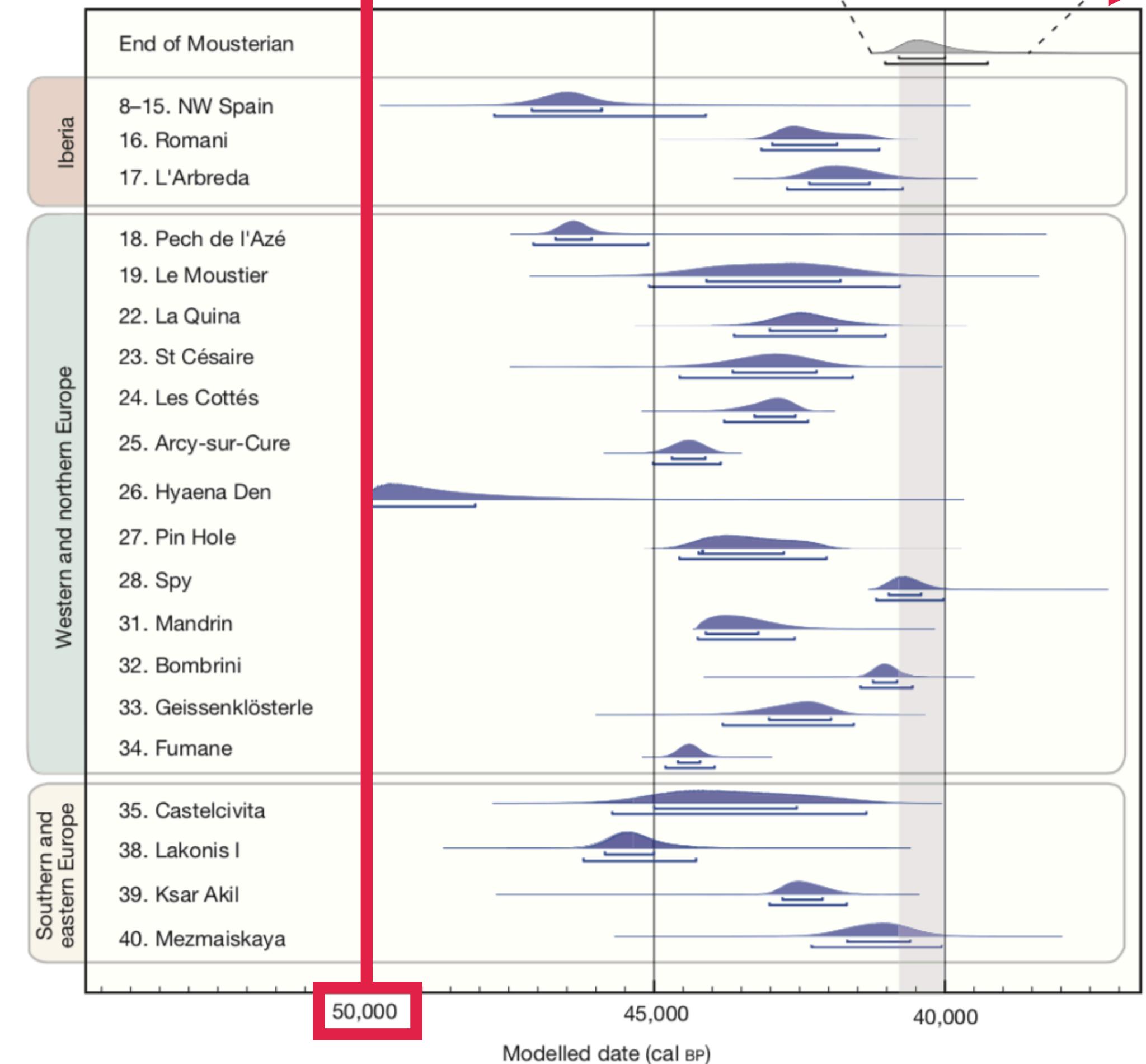
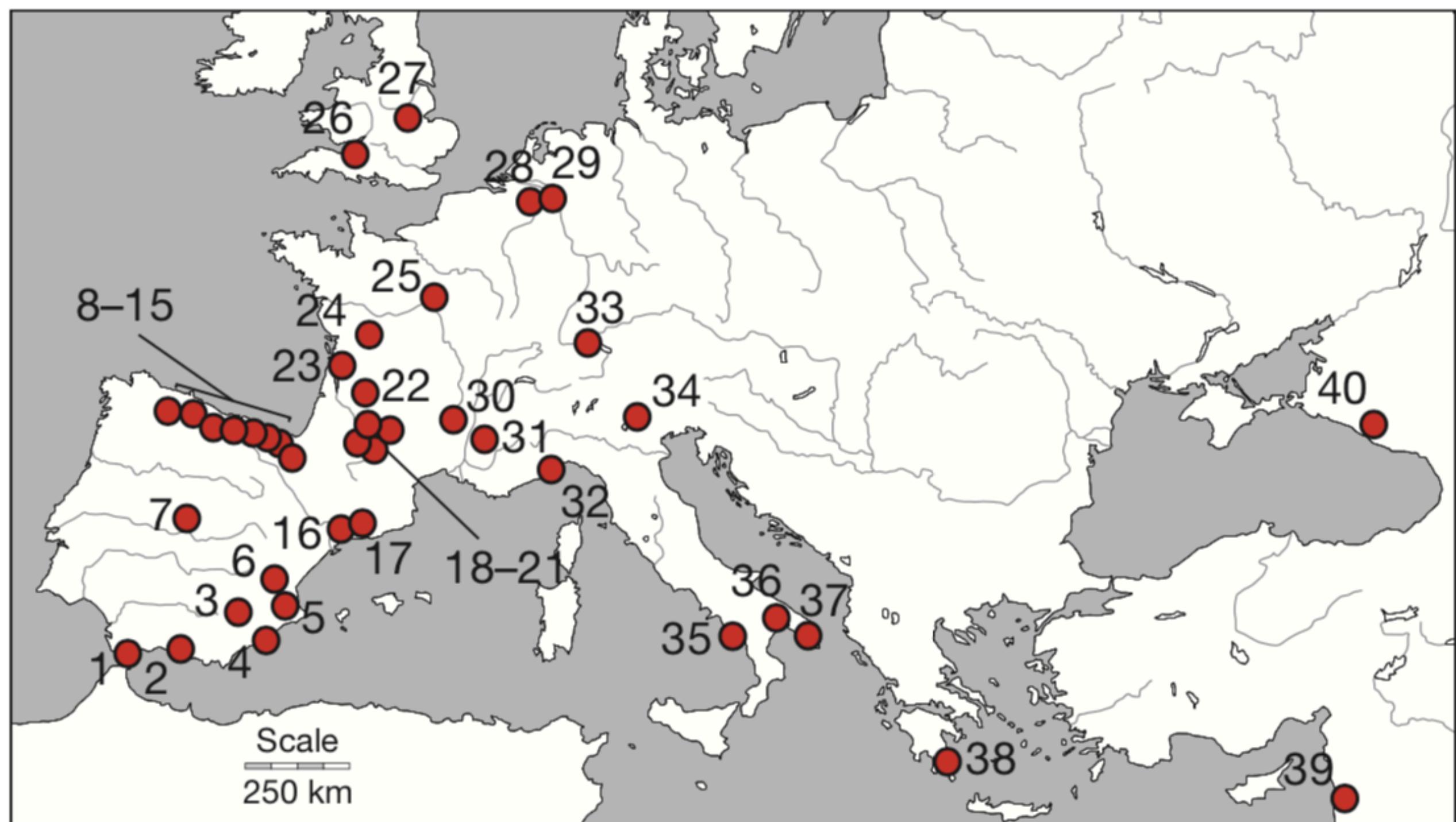




ABBA/BABA EXERCISES

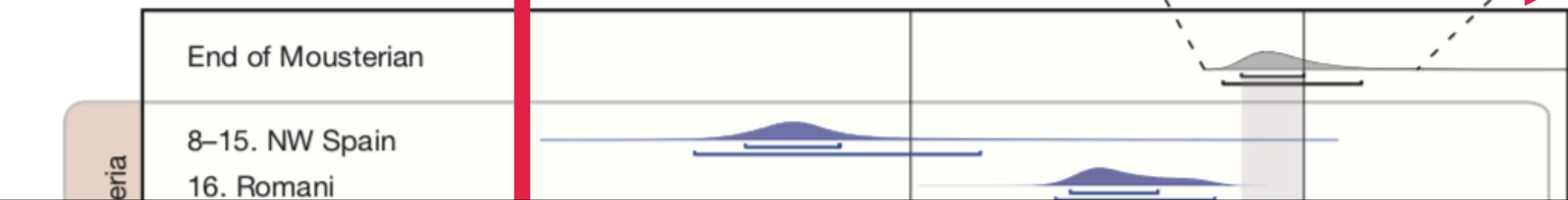
github.com/bodkan/ku-introgression2025

anatomically modern humans in Europe

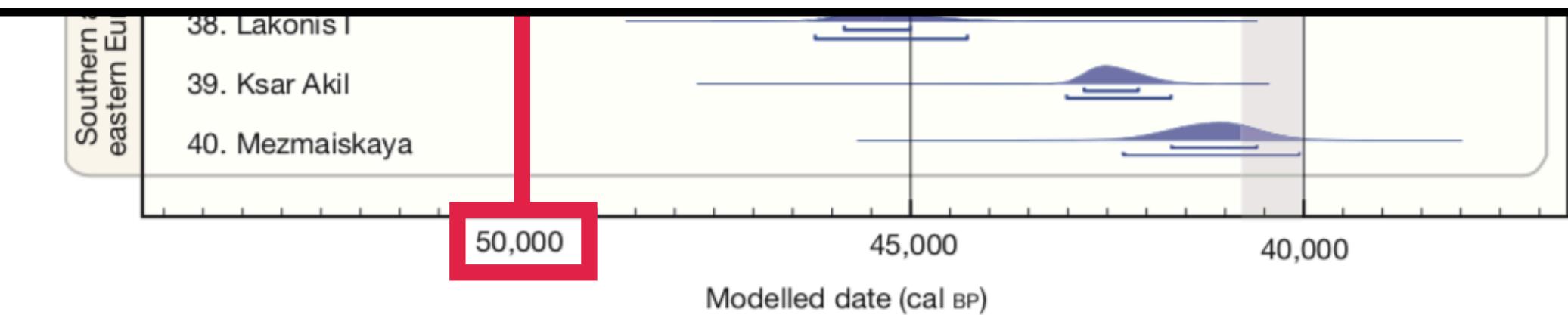


Higham et al. (Nature, 2014)

anatomically modern humans in Europe



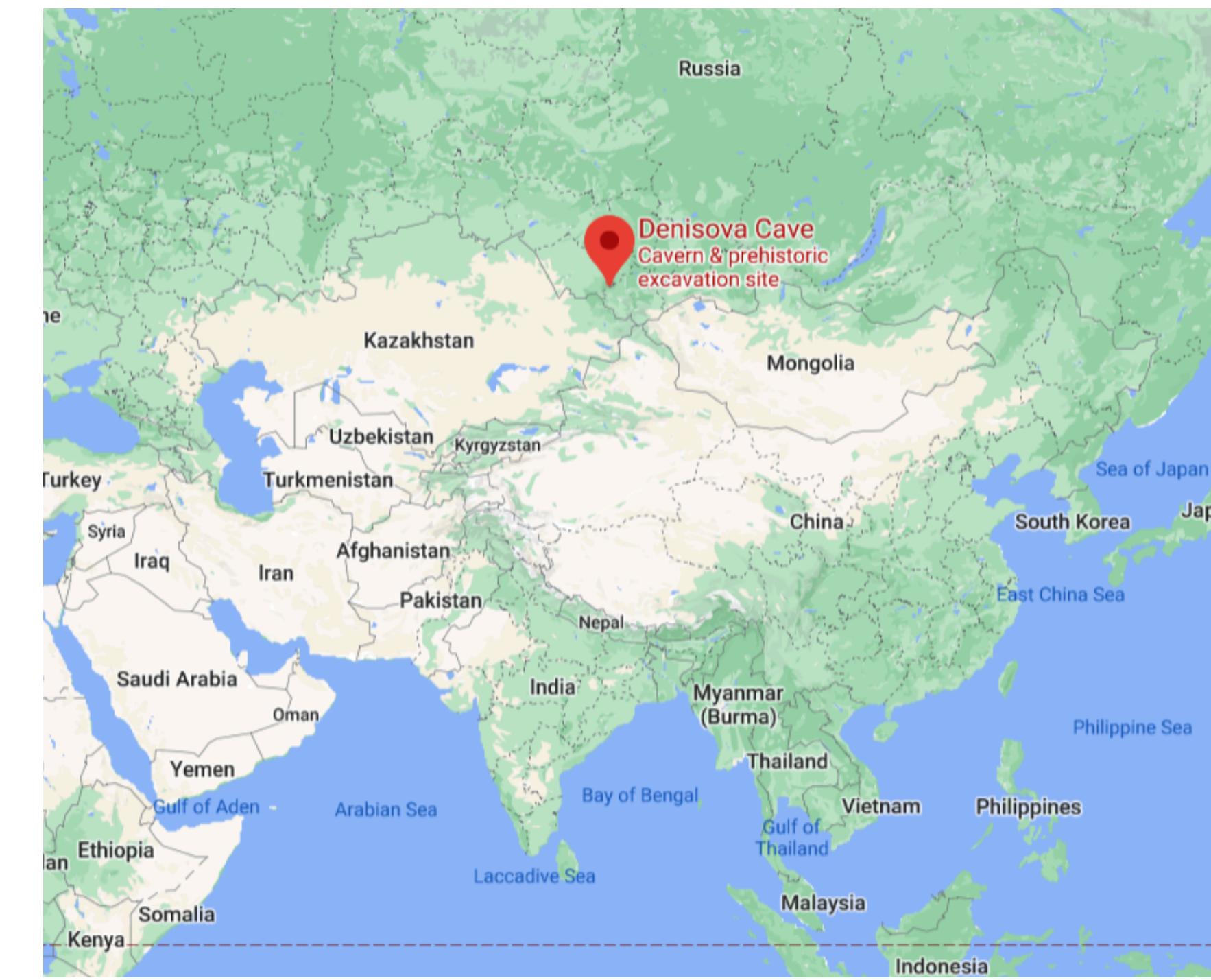
Neanderthals were not the only archaics around at the time

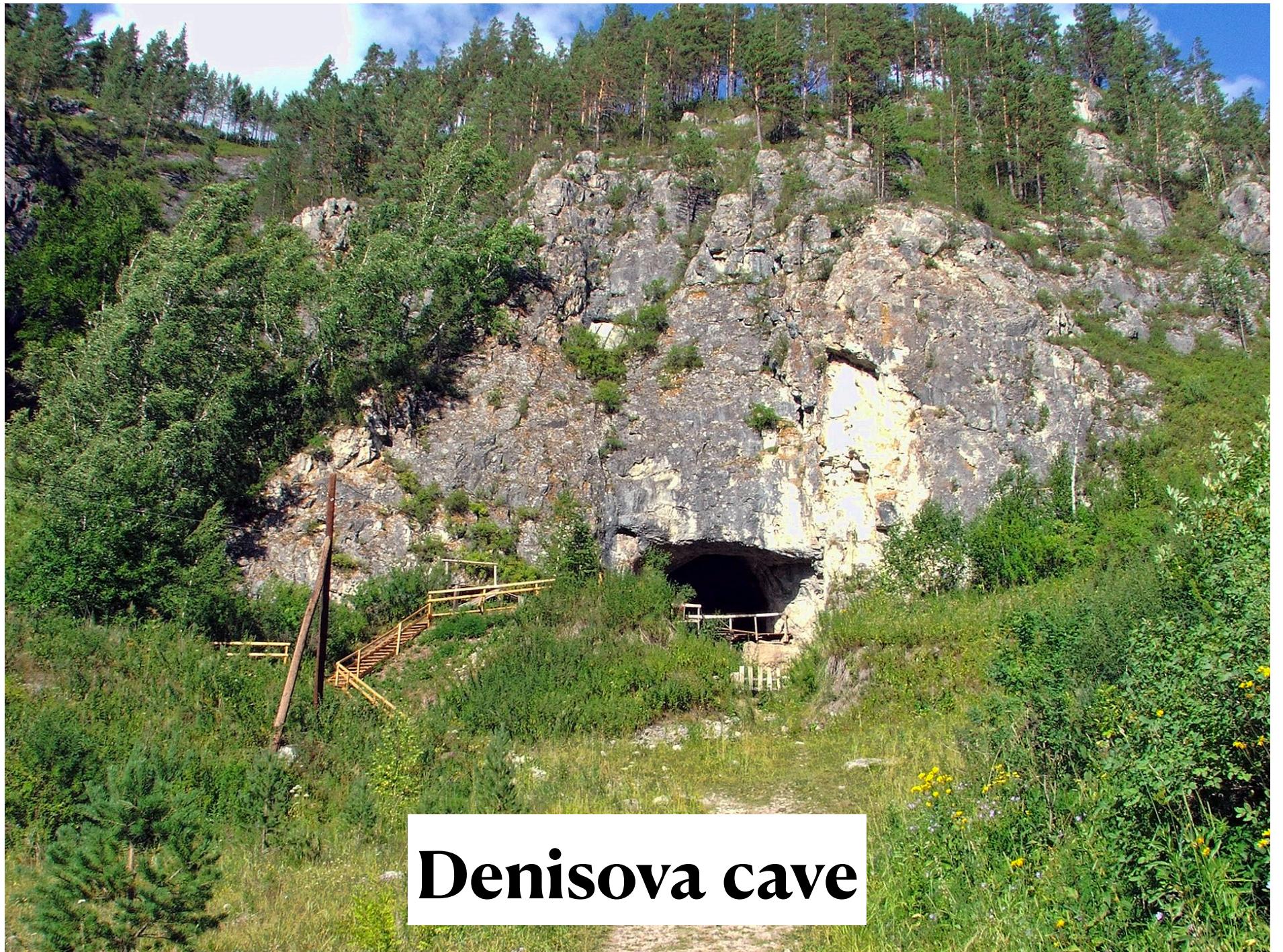


Higham et al. (Nature, 2014)

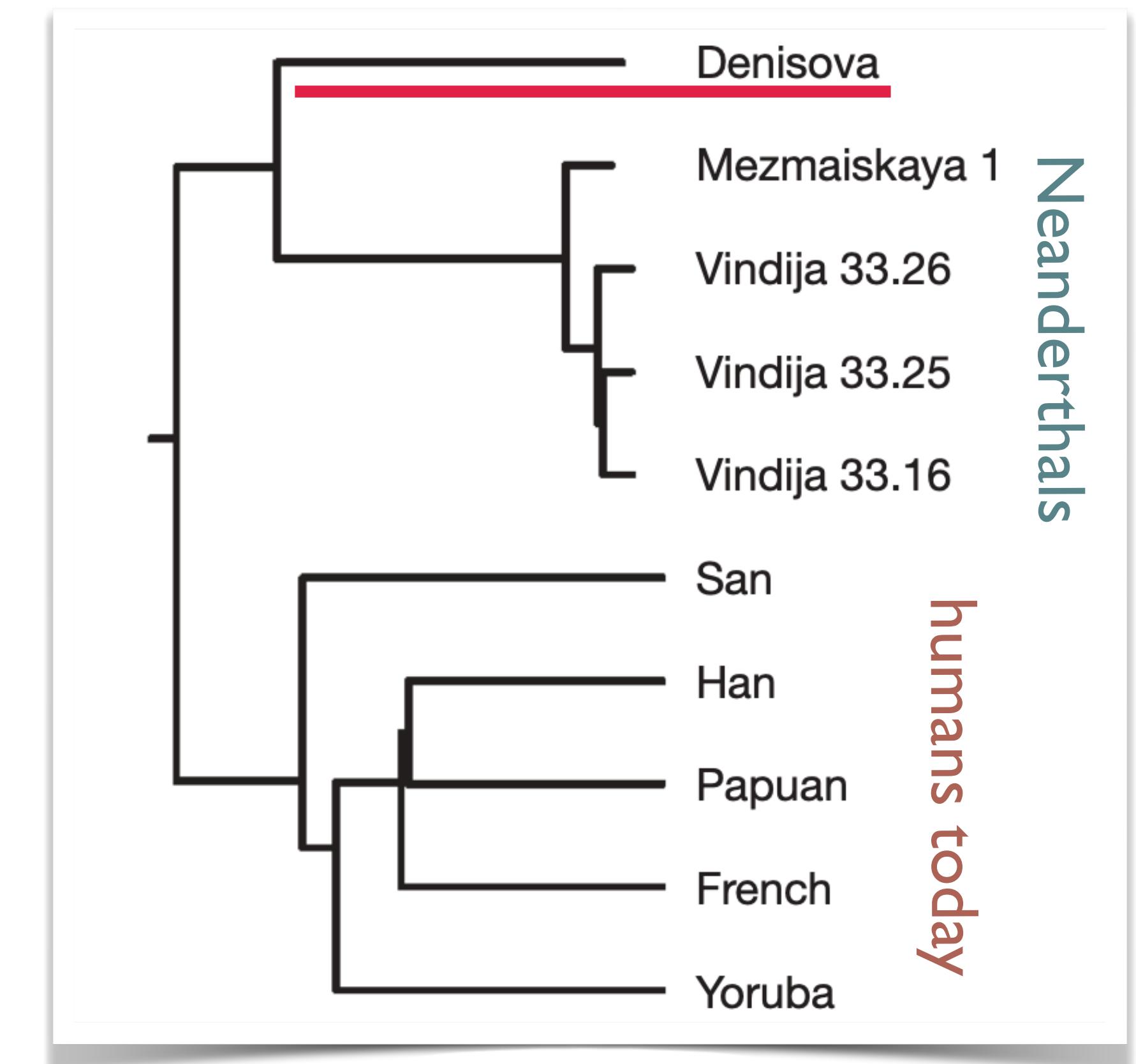


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nature

Open Access | Published: 22 December 2010

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

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

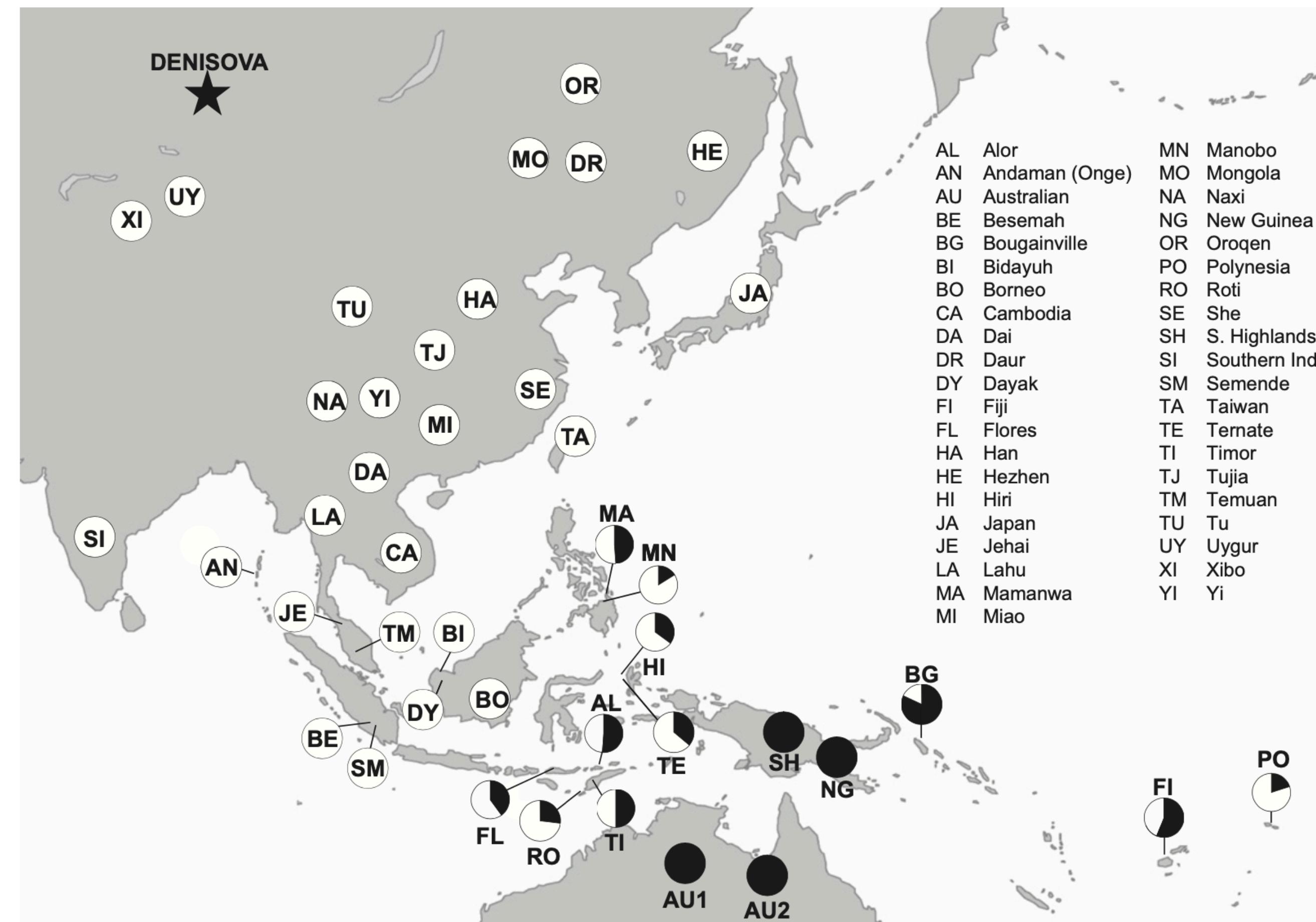


Figure 1. Denisovan Genetic Material as a Fraction of that in New Guineans

Reich et al., 2011

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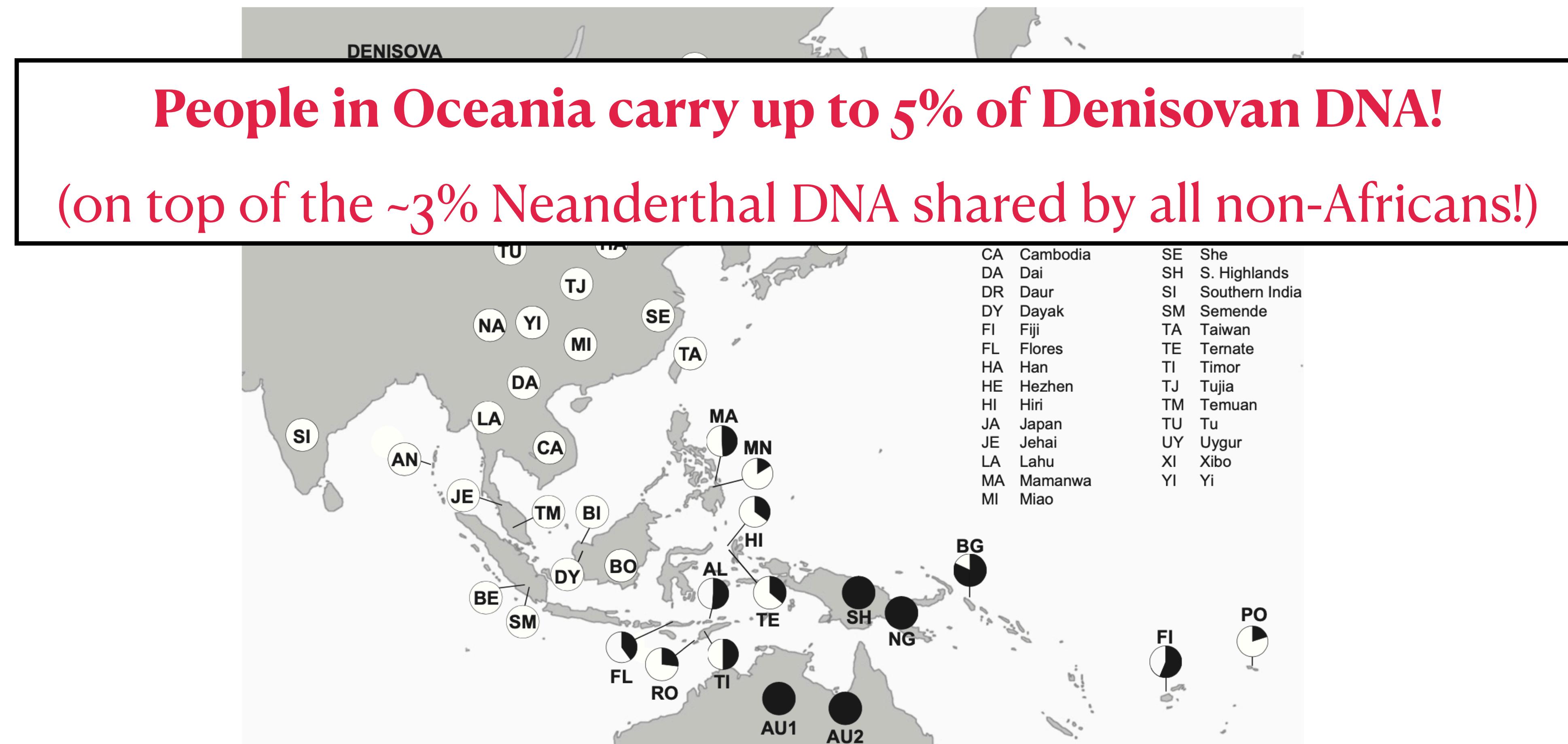
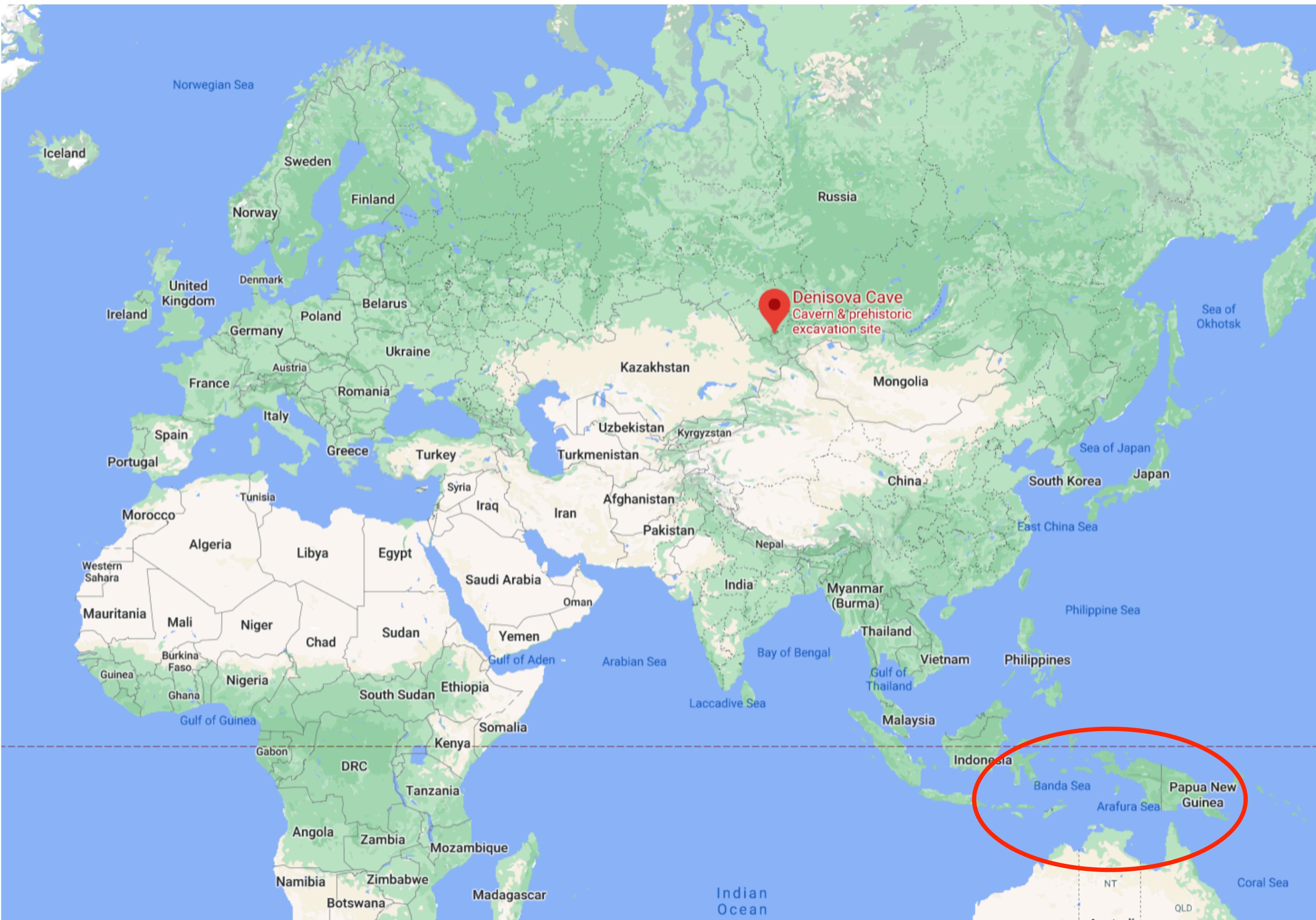


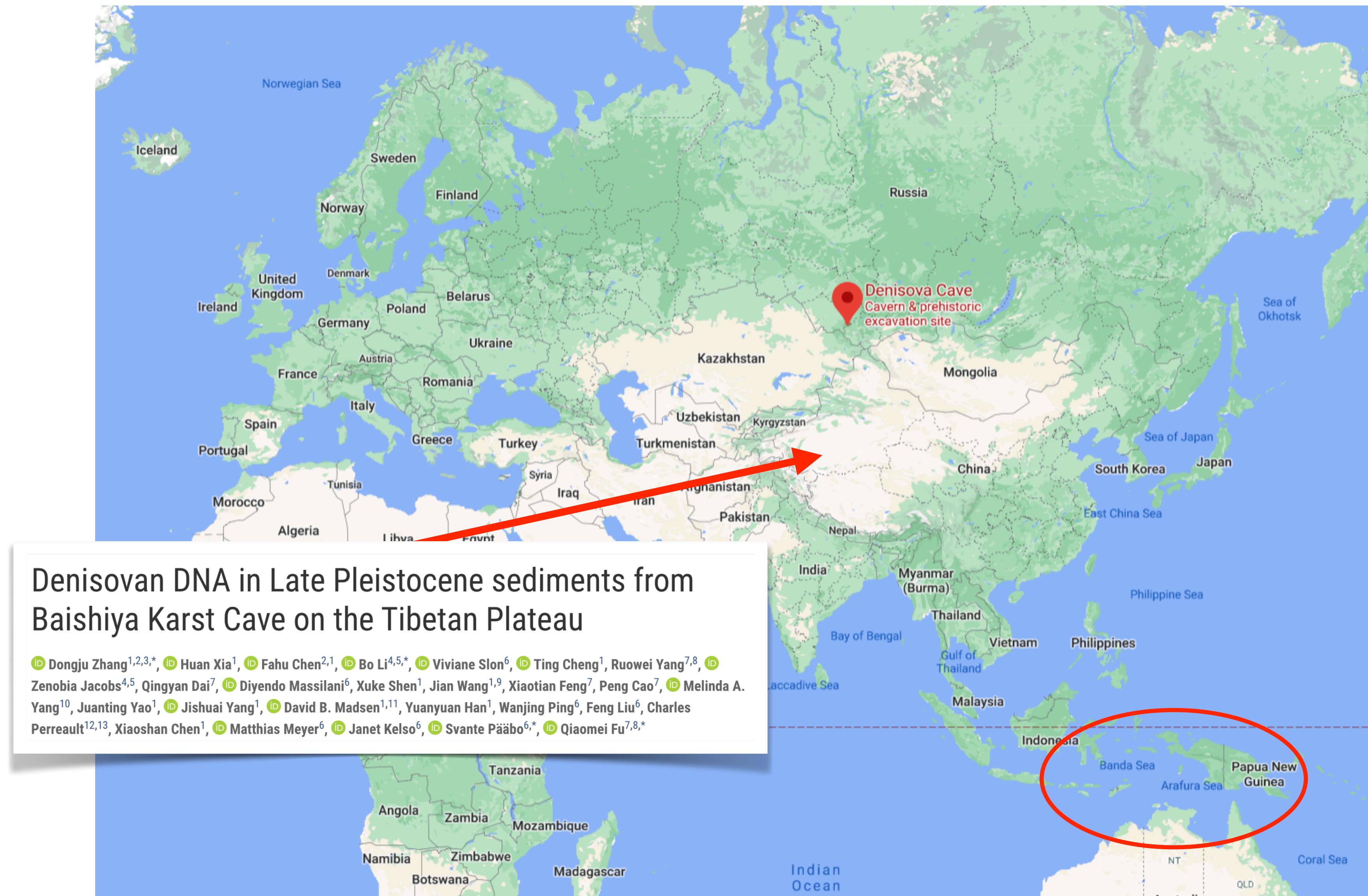
Figure 1. Denisovan Genetic Material as a Fraction of that in New Guineans

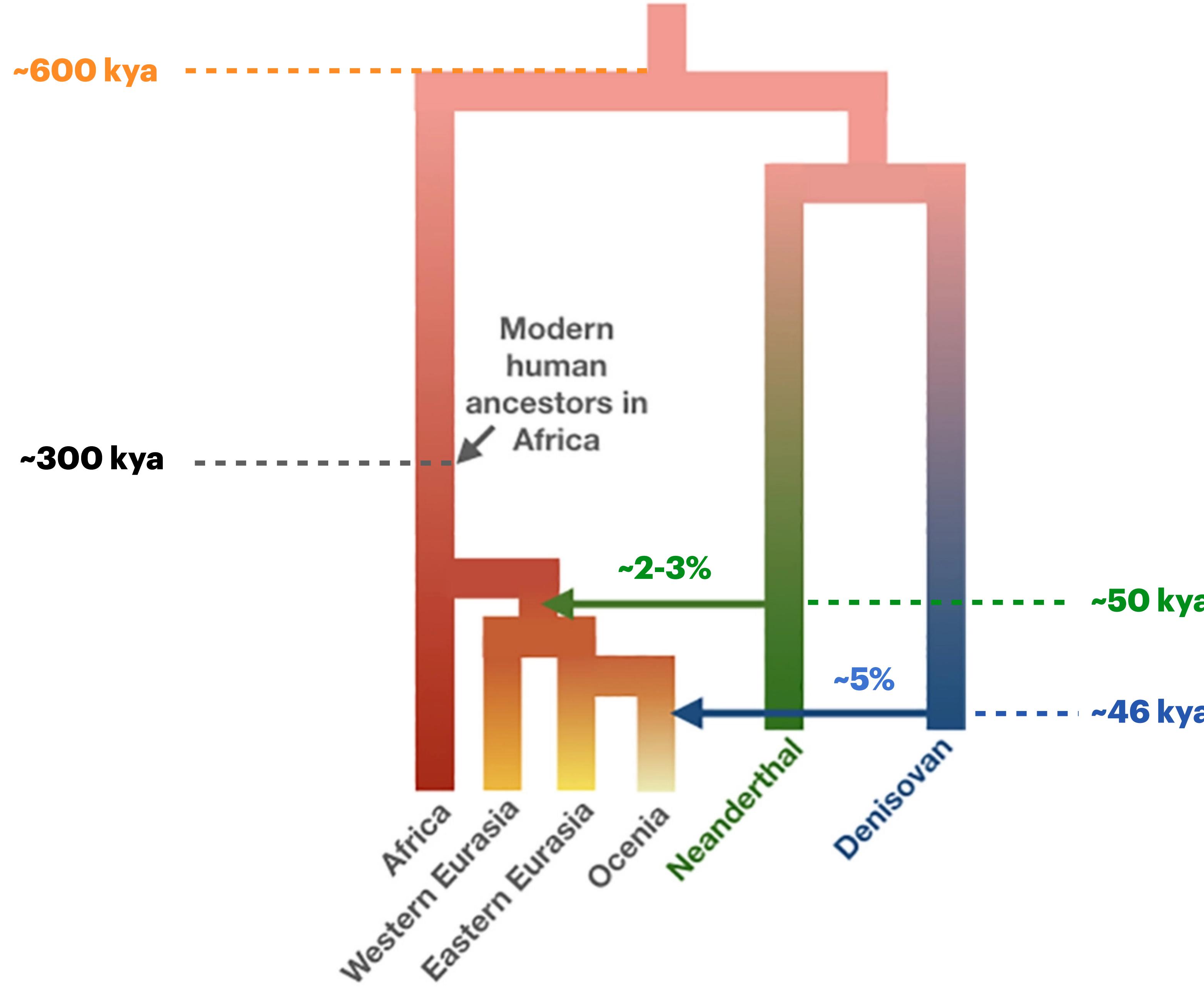
Reich et al., 2011

Geographic range of Denisovans?



Geographic range of Denisovans?

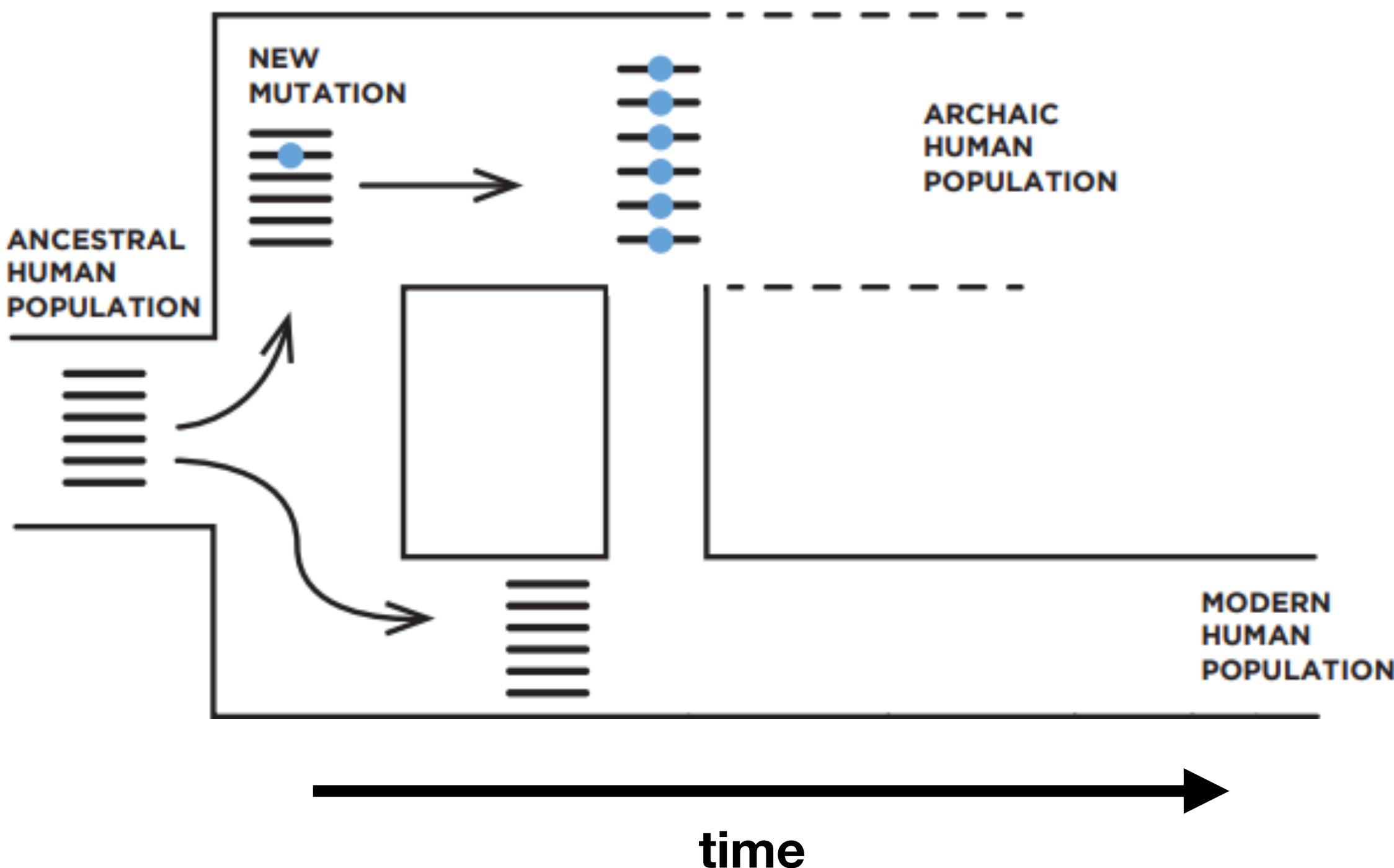




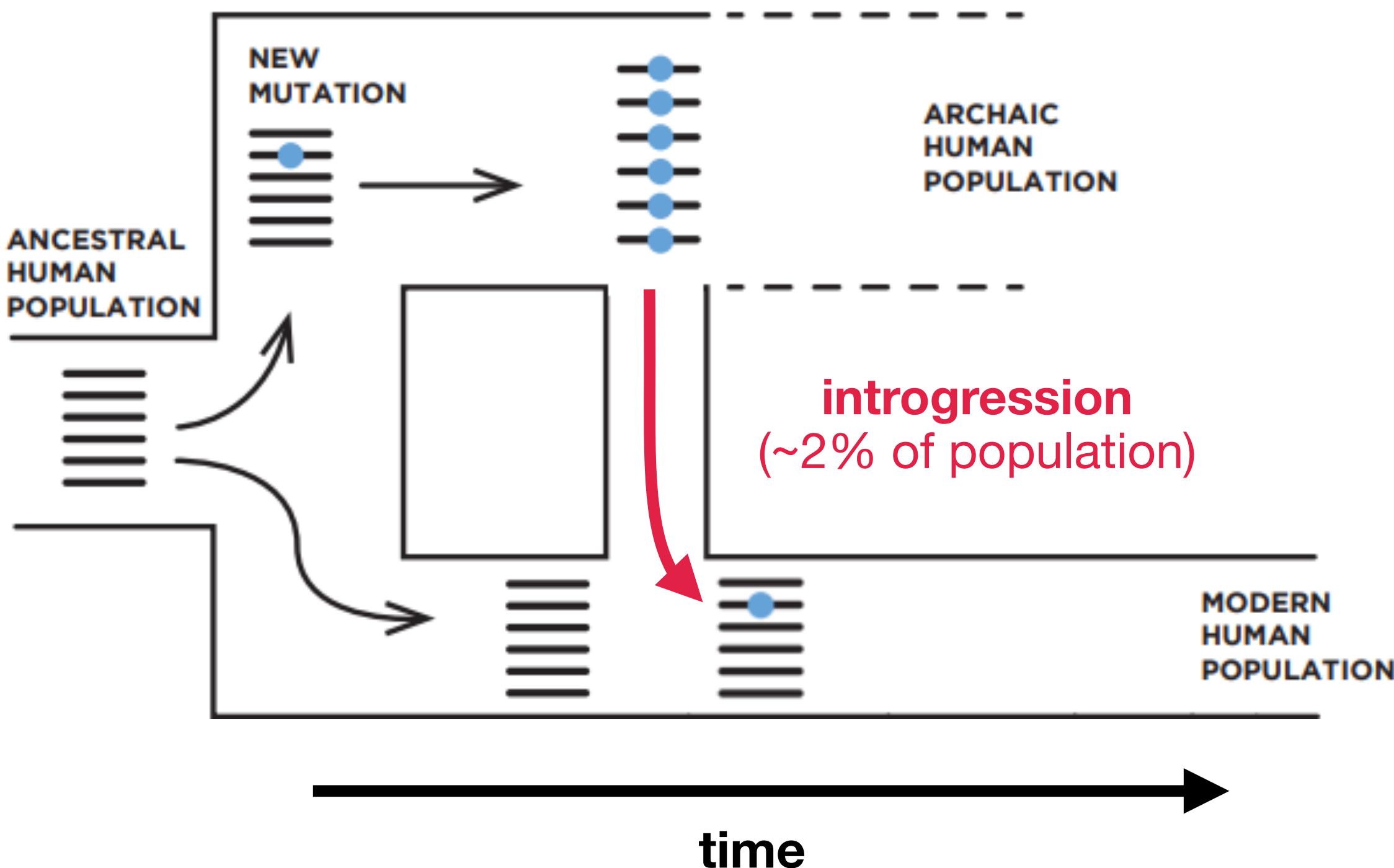
adapted from the Yearbook of Physical Anthropology (2019) by Gokcumen

Consequences of introgression on human functional biology?

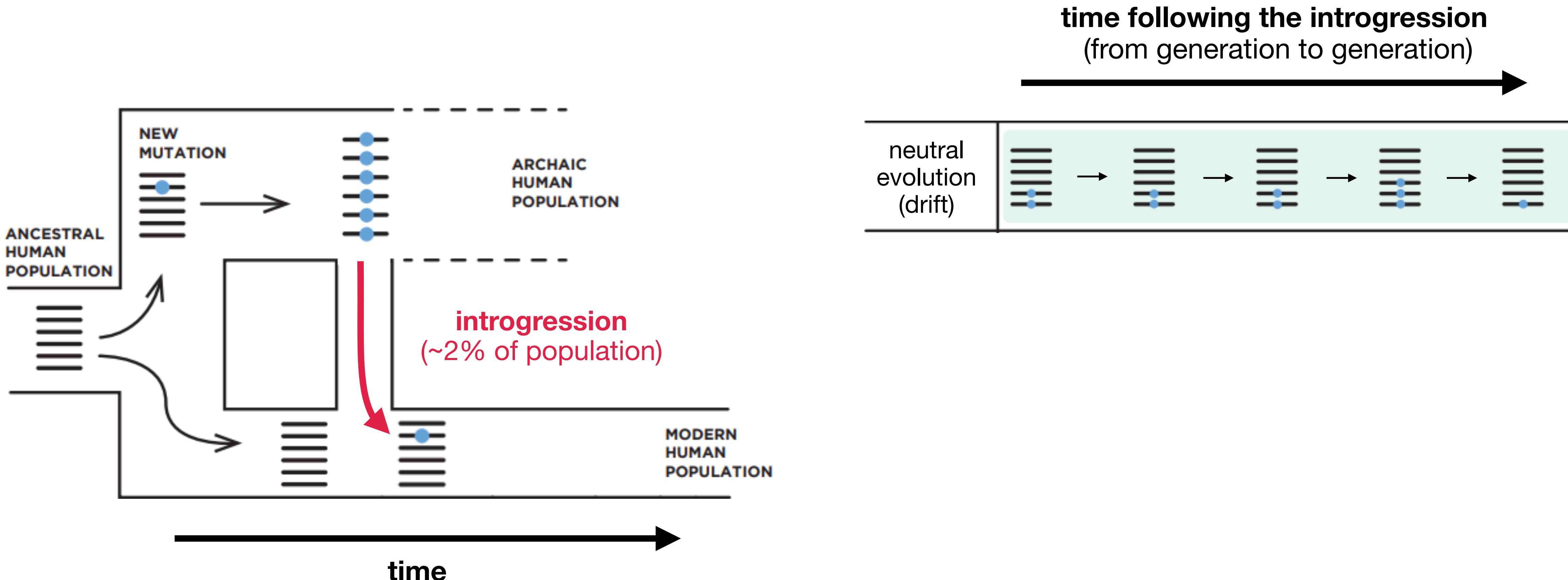
What are the possible fates of introgressed DNA?



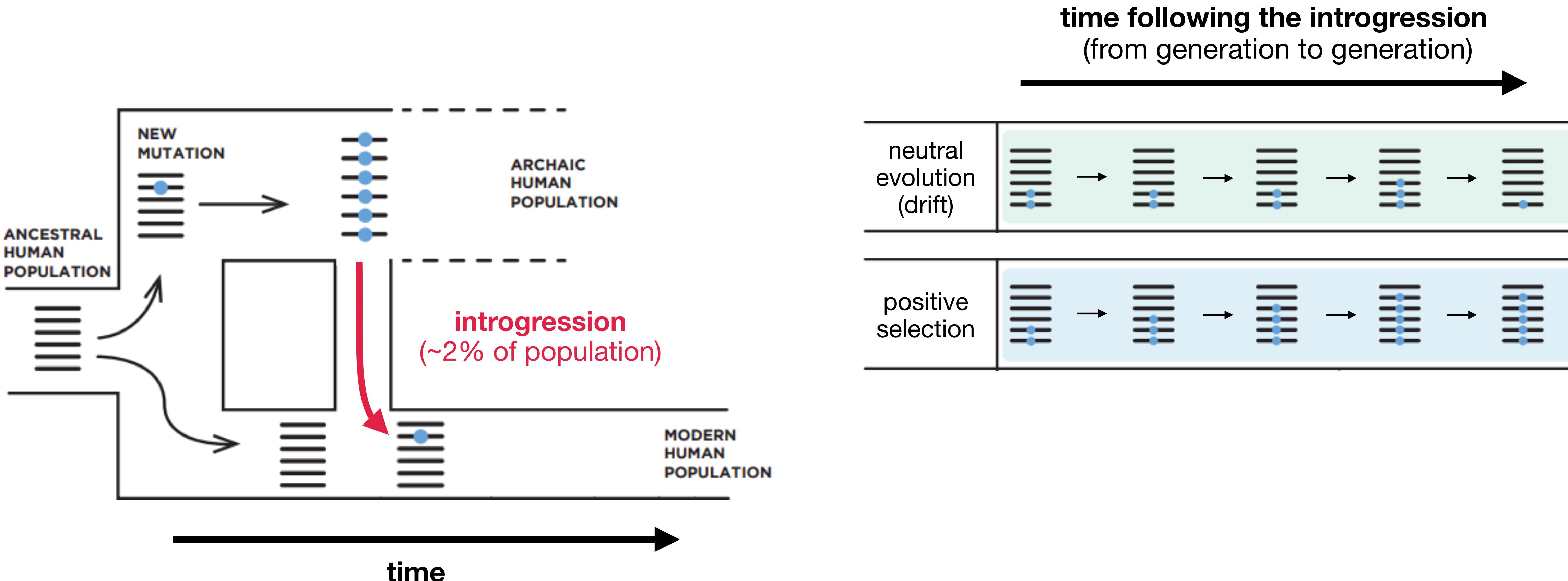
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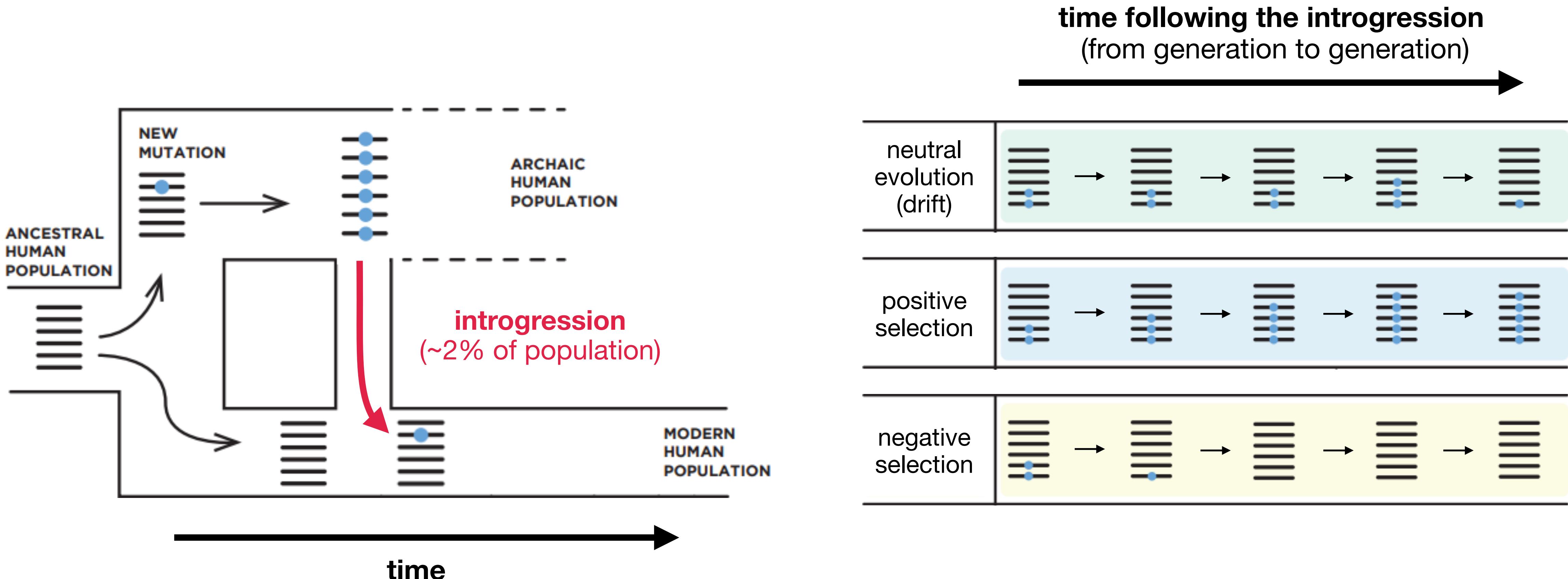
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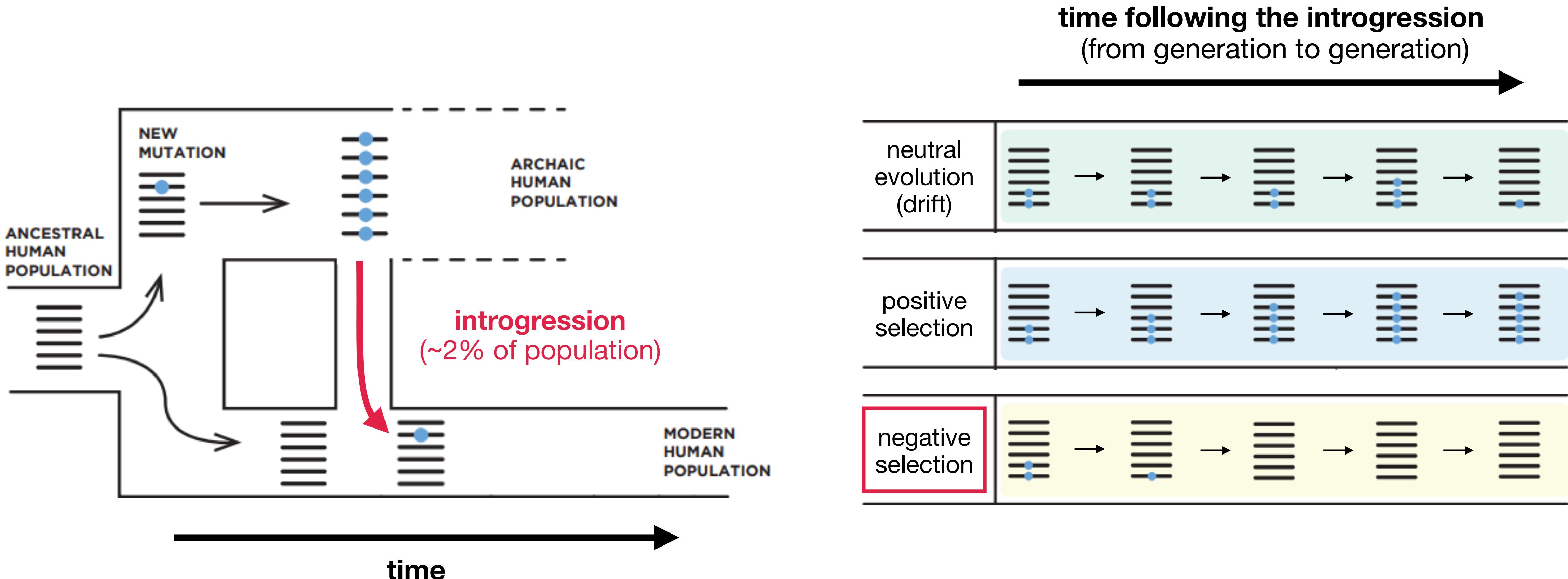
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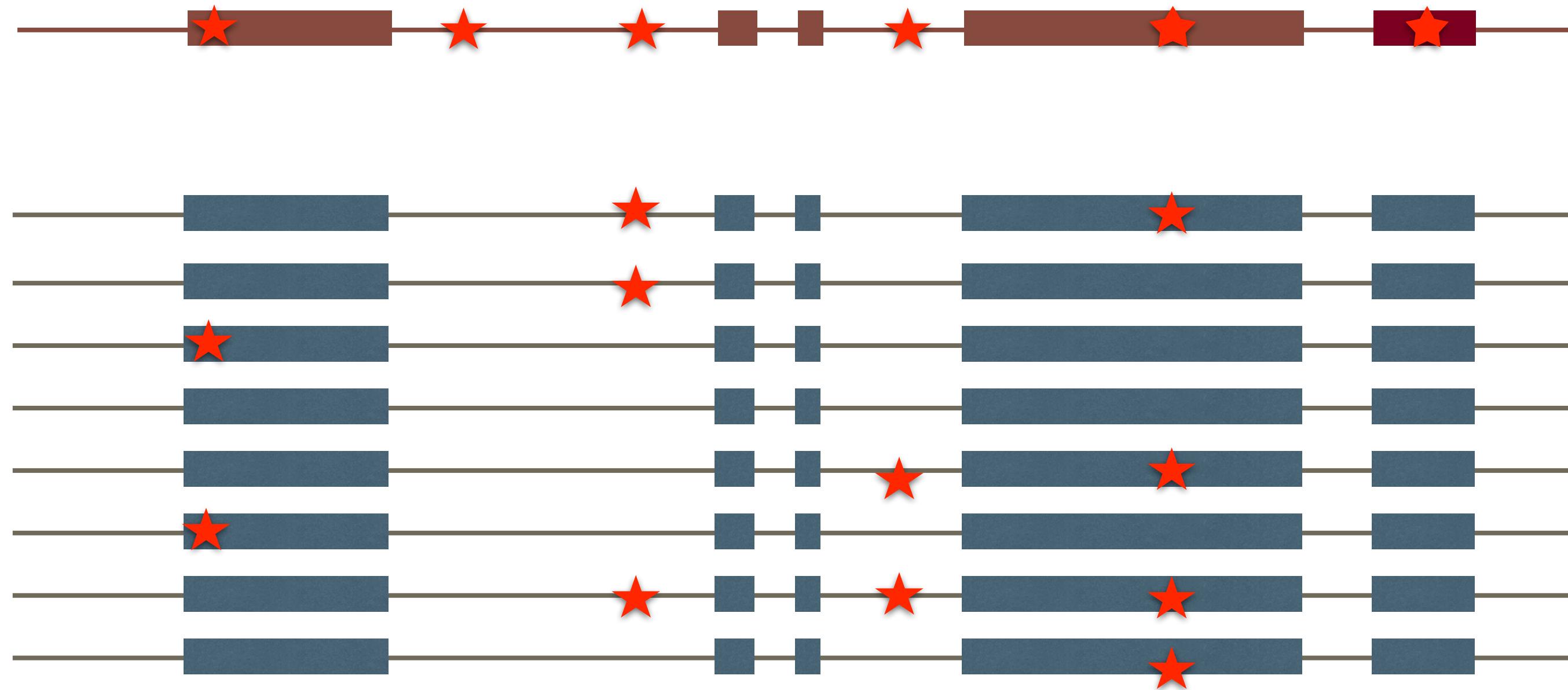


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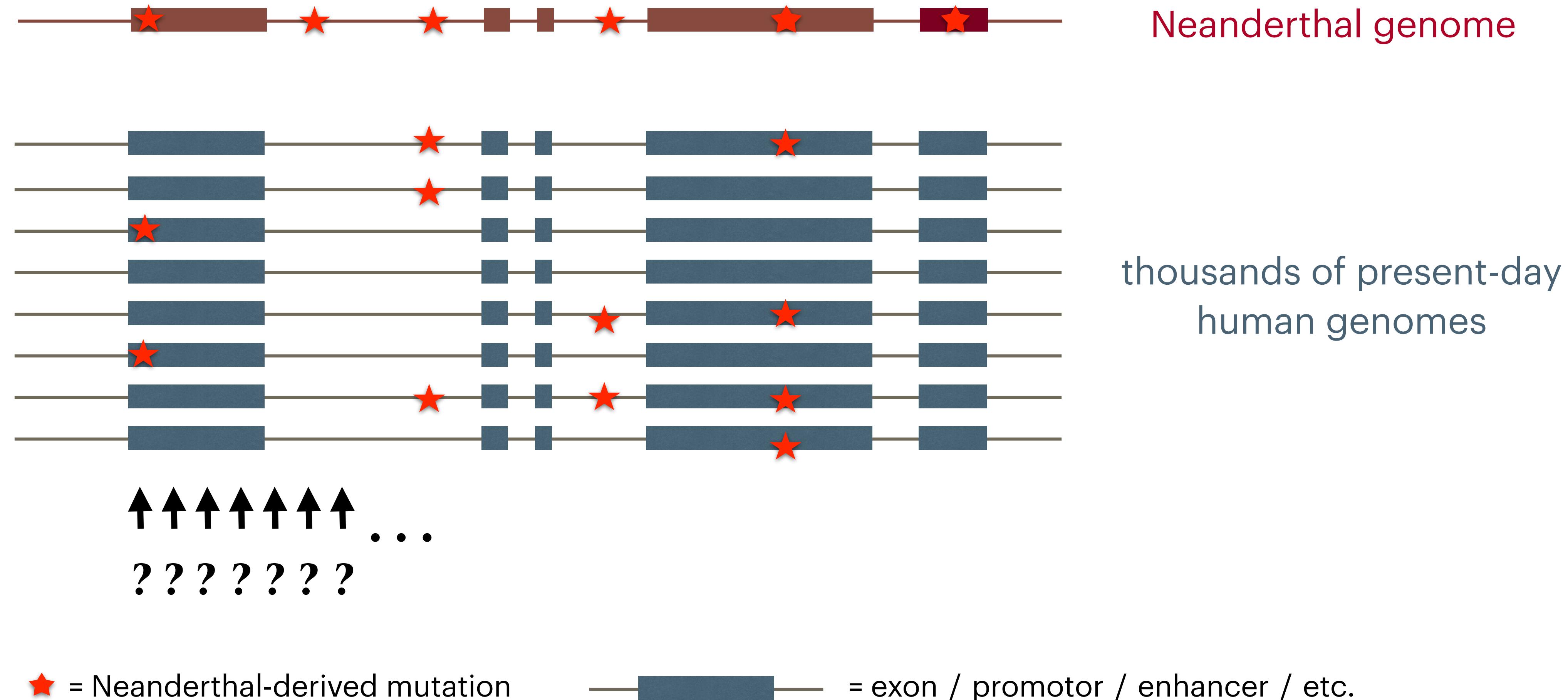
Neanderthal genome

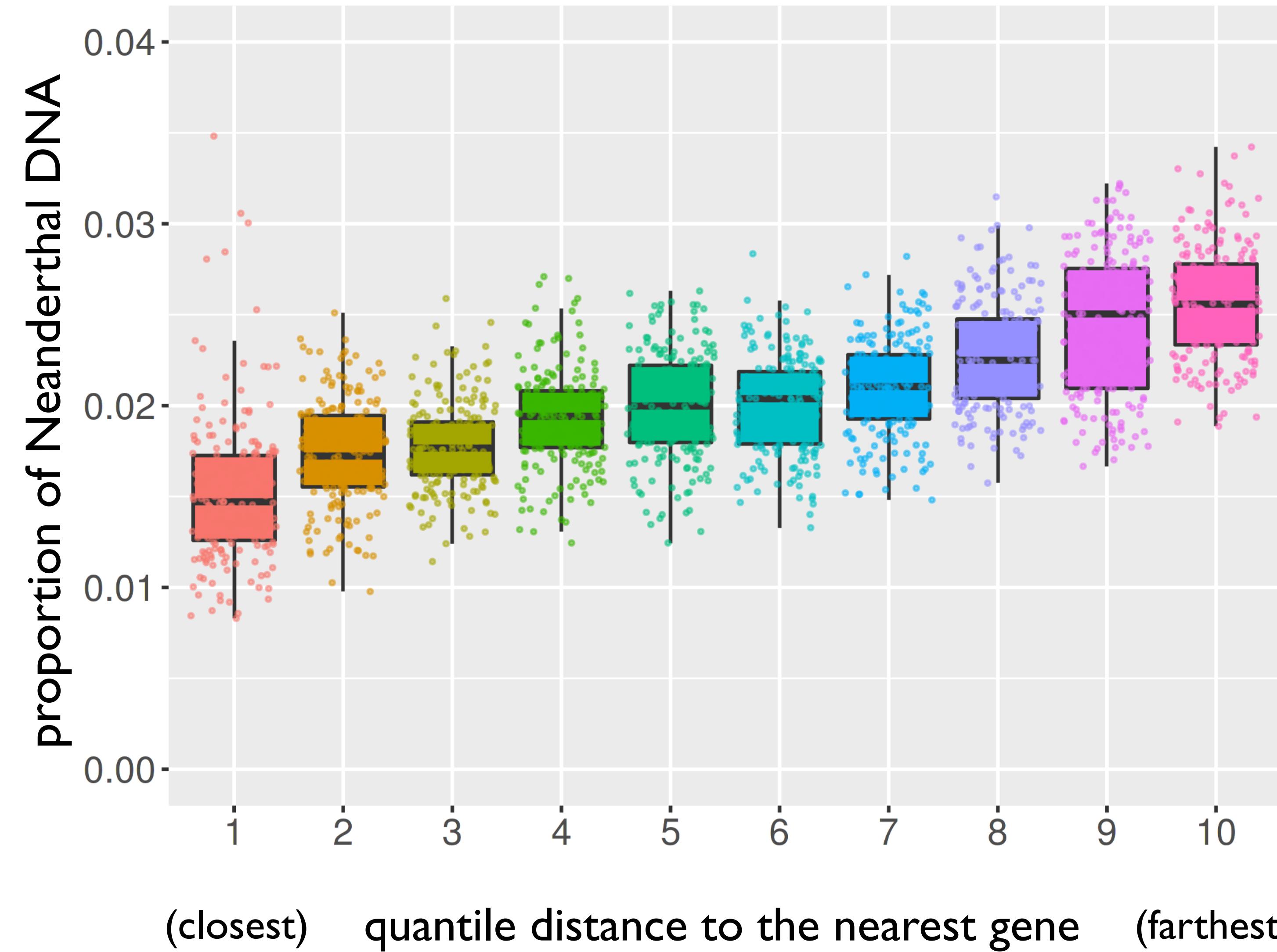
thousands of present-day
human genomes

★ = Neanderthal-derived mutation

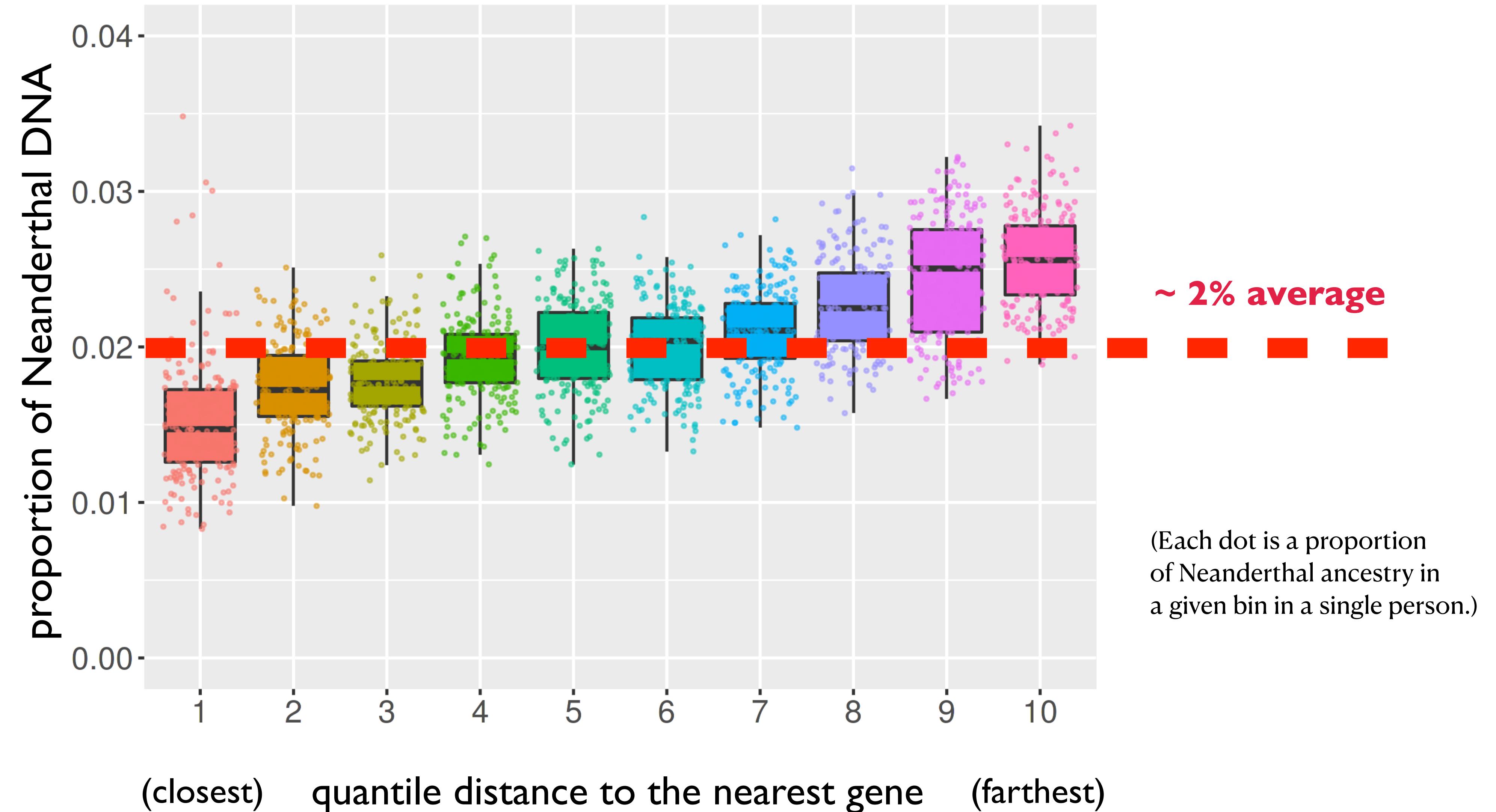
—■— = exon / promotor / enhancer / etc.

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

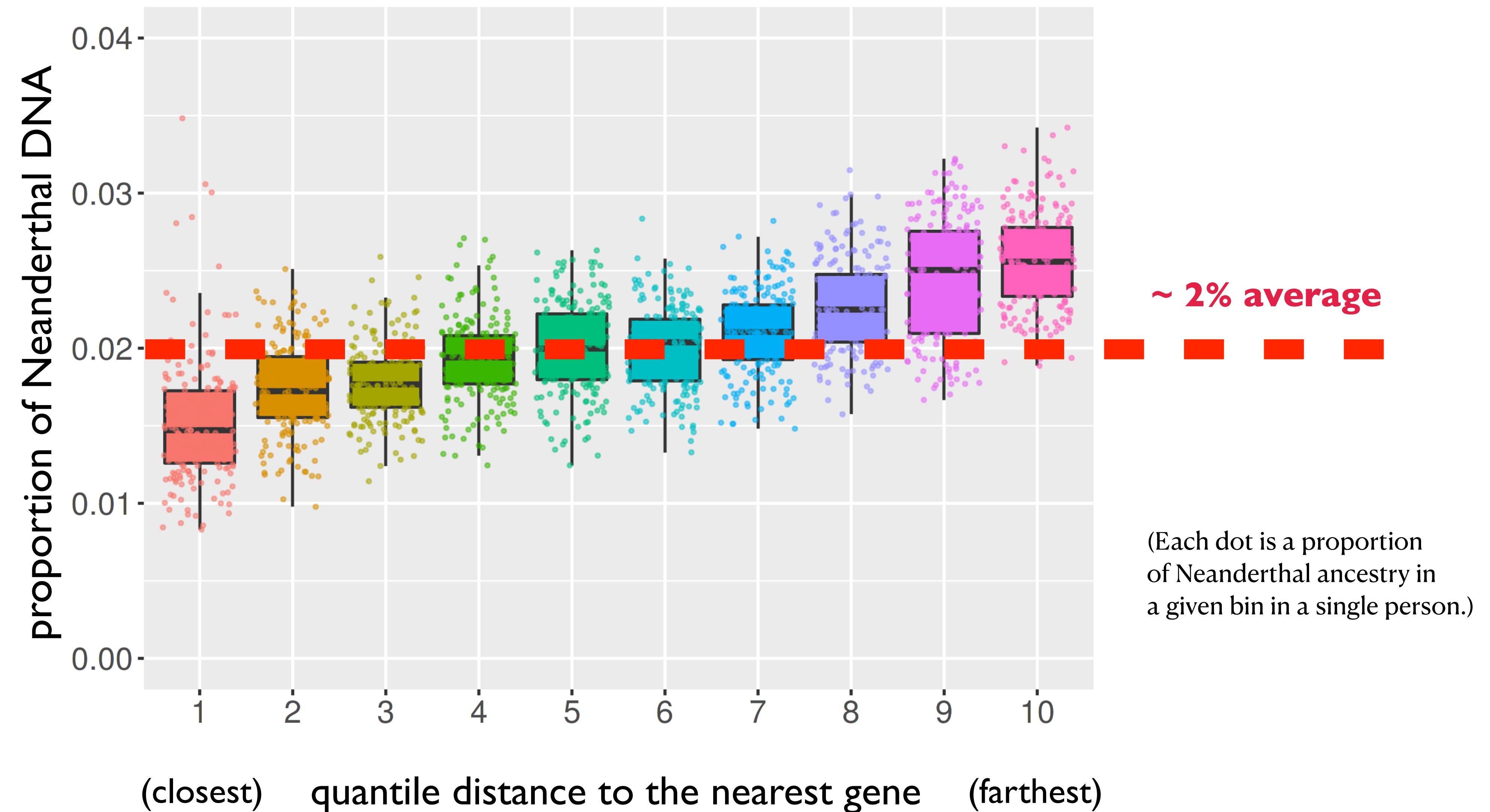




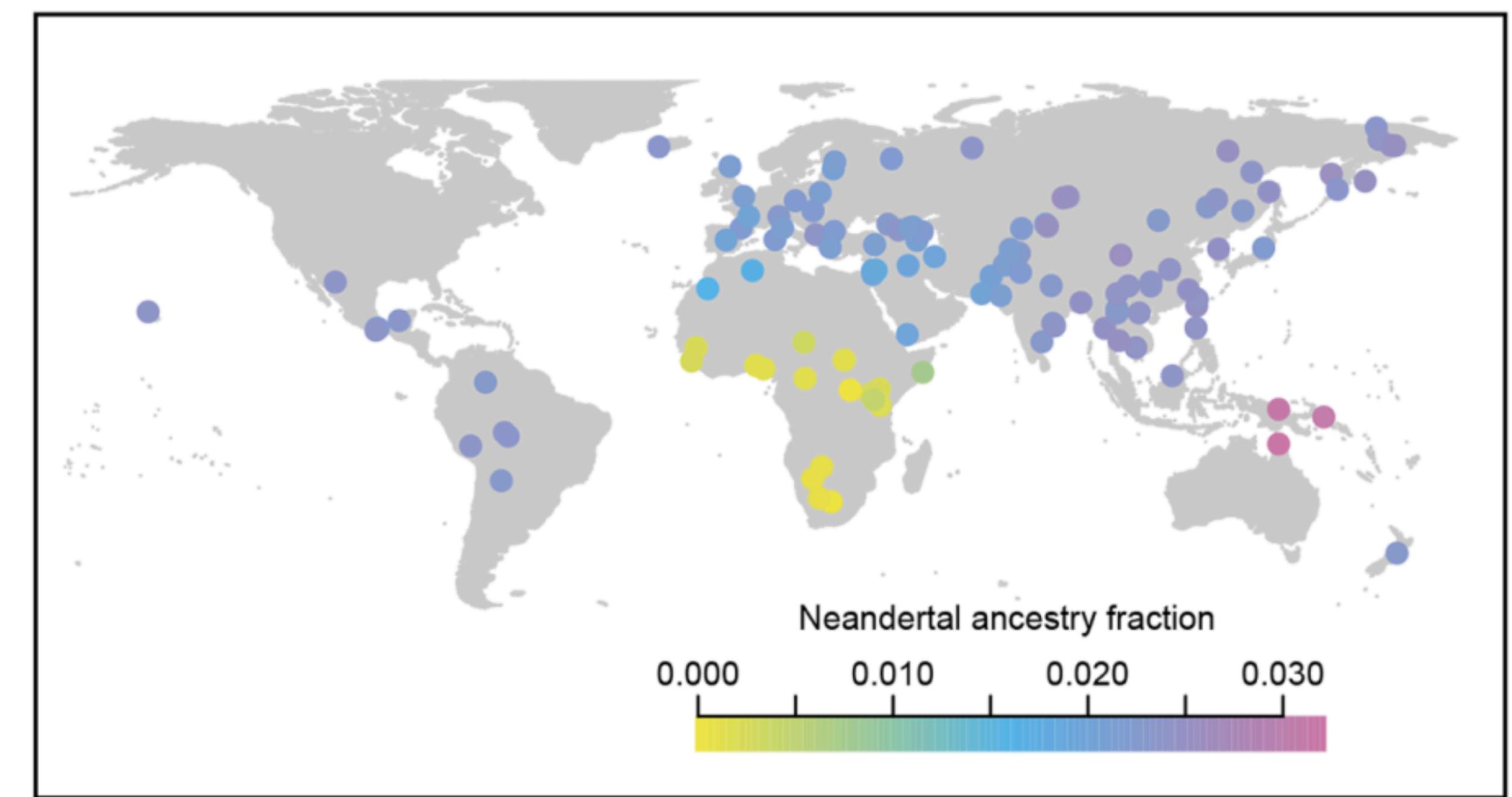
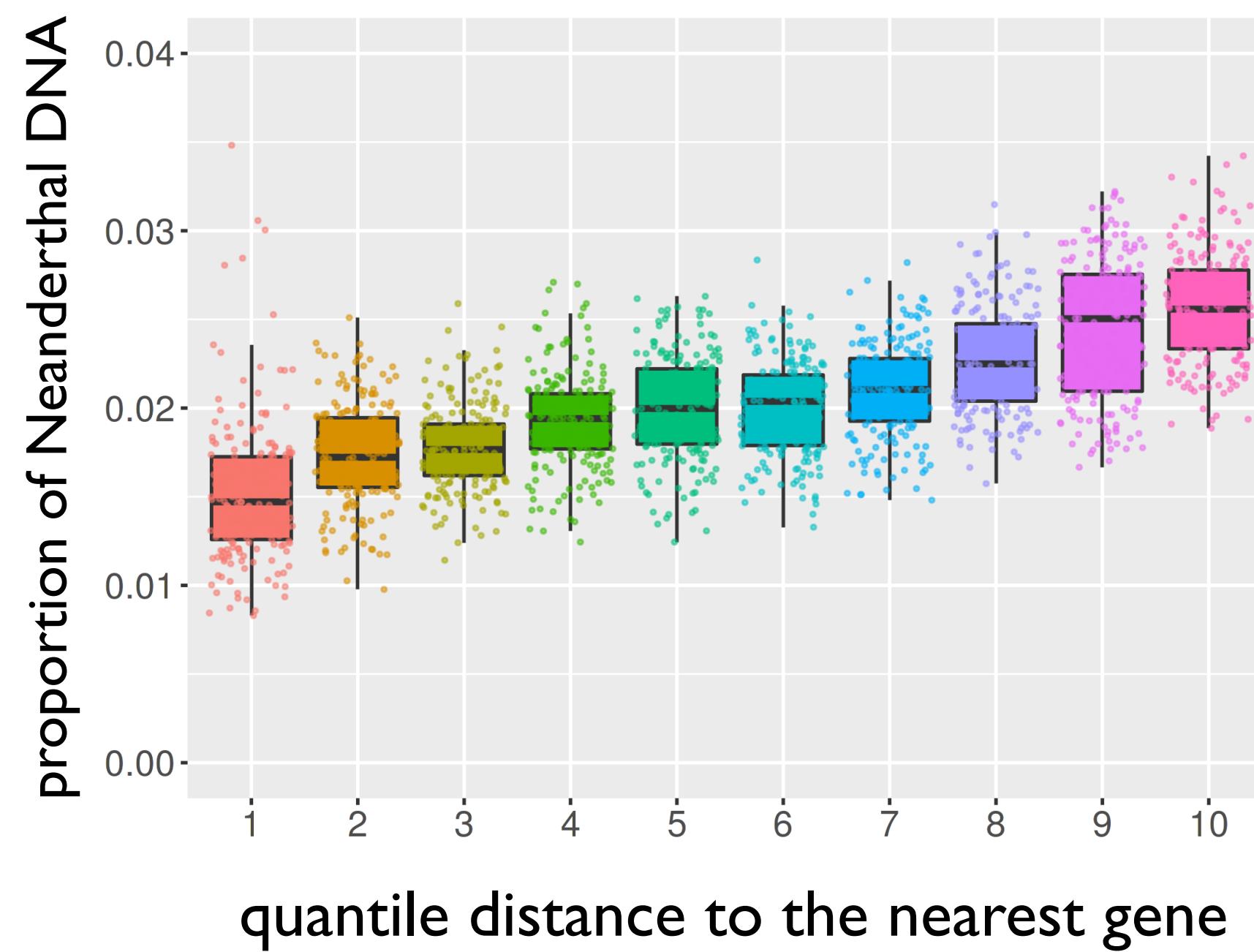
(Each dot is a proportion
of Neanderthal ancestry in
a given bin in a single person.)



Genome-wide selection *against* Neanderthal DNA!

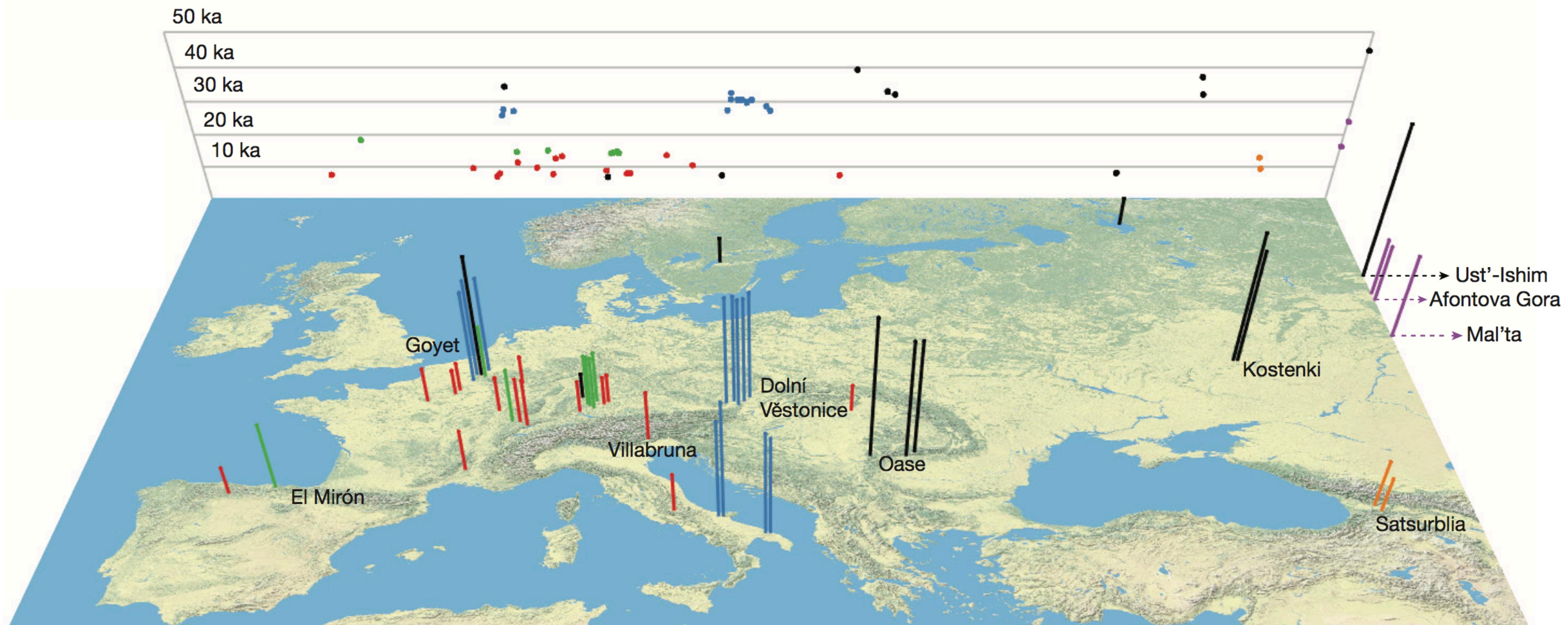


On what time scale did this happen?

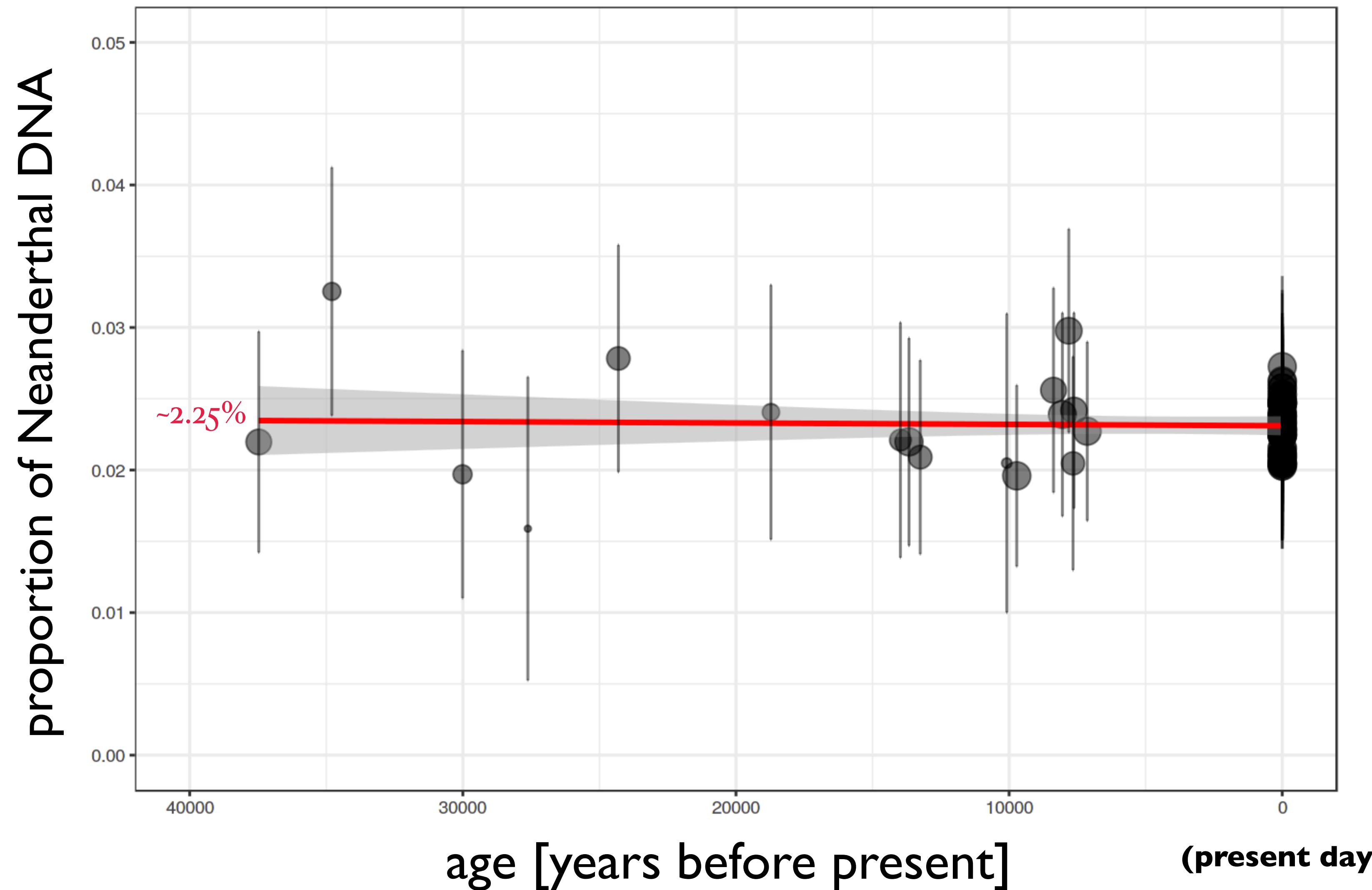


Prufer et al. (Science 2017)

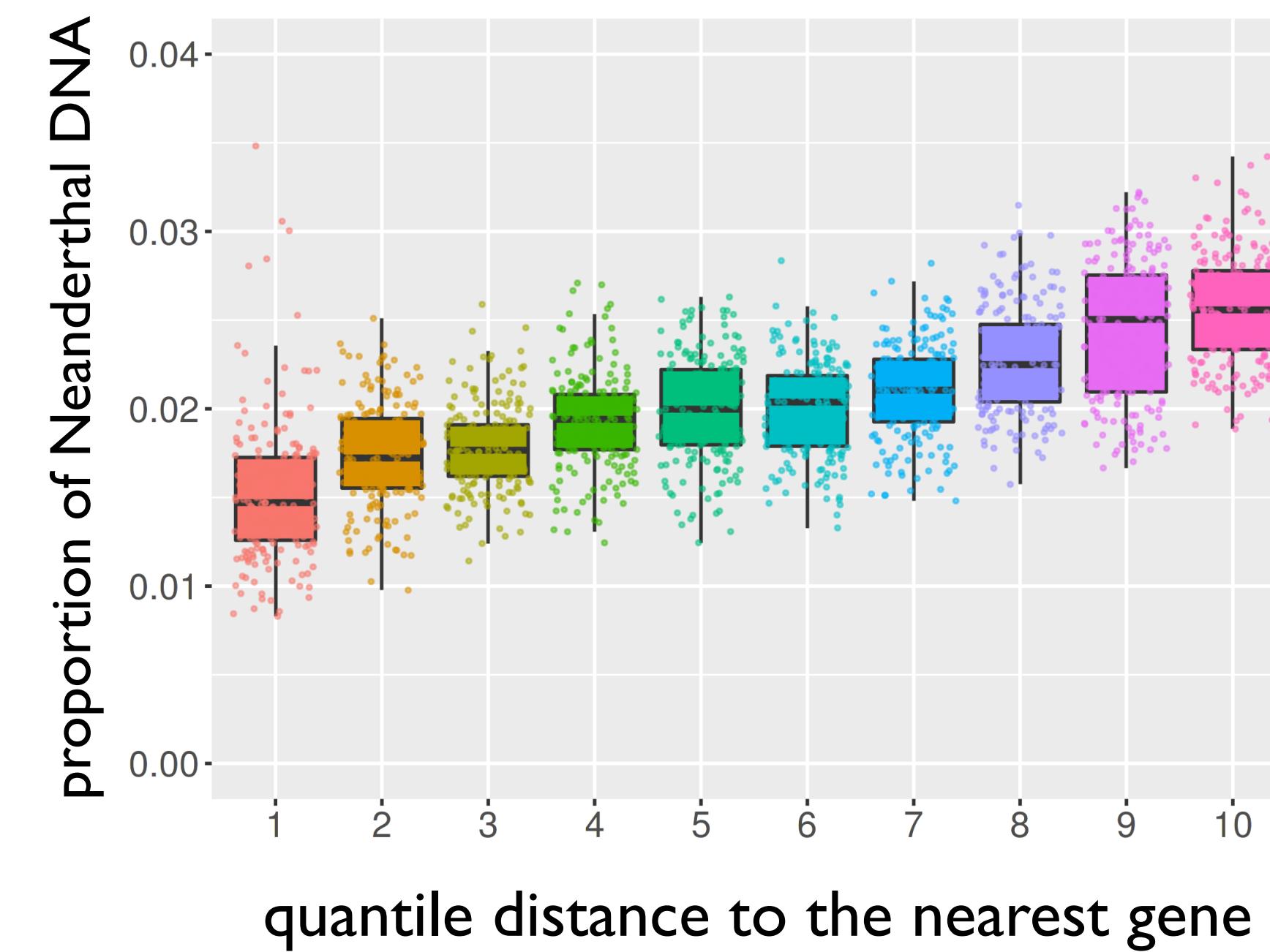
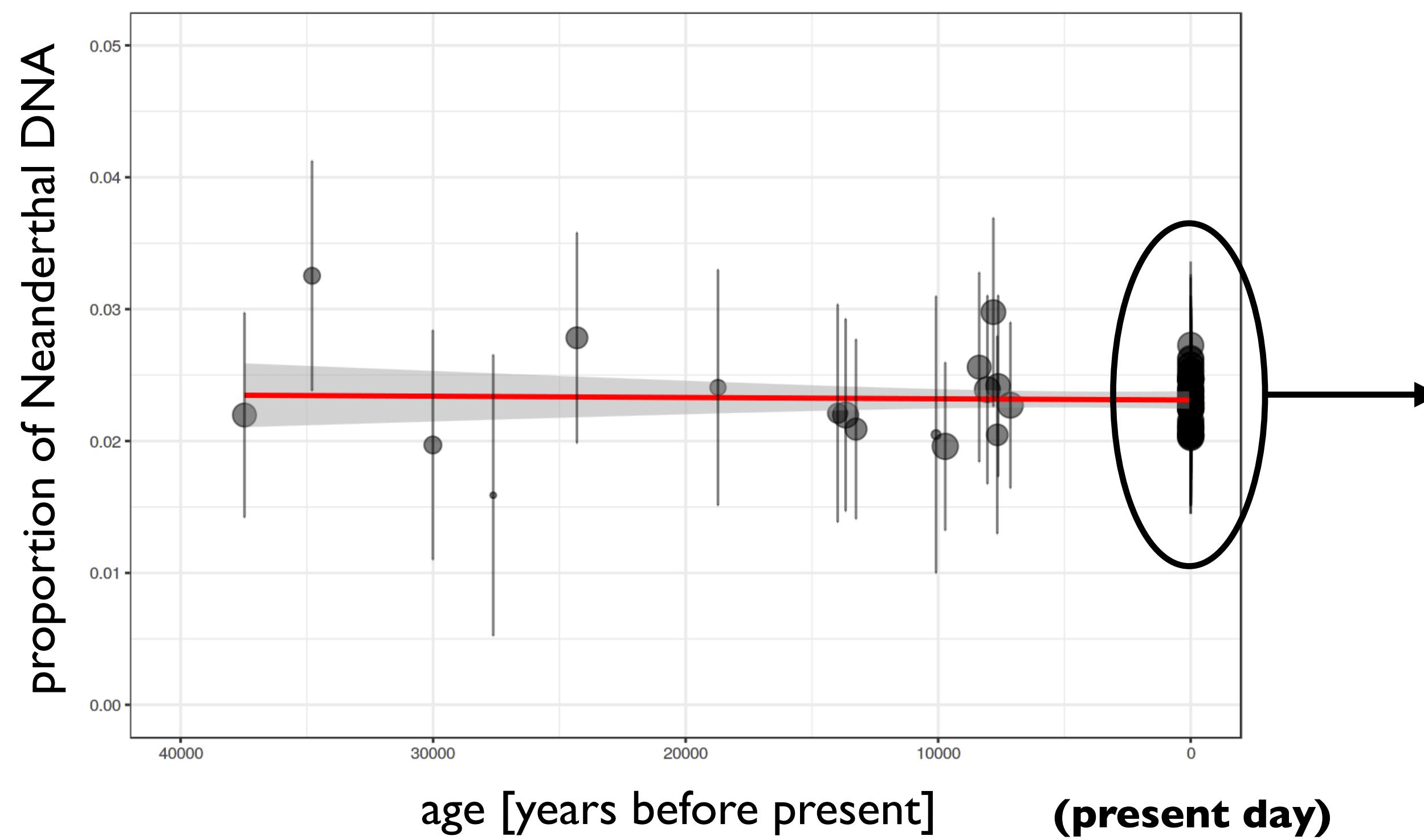
aDNA of “early-modern humans” from Europe (45-10 kya BP)



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

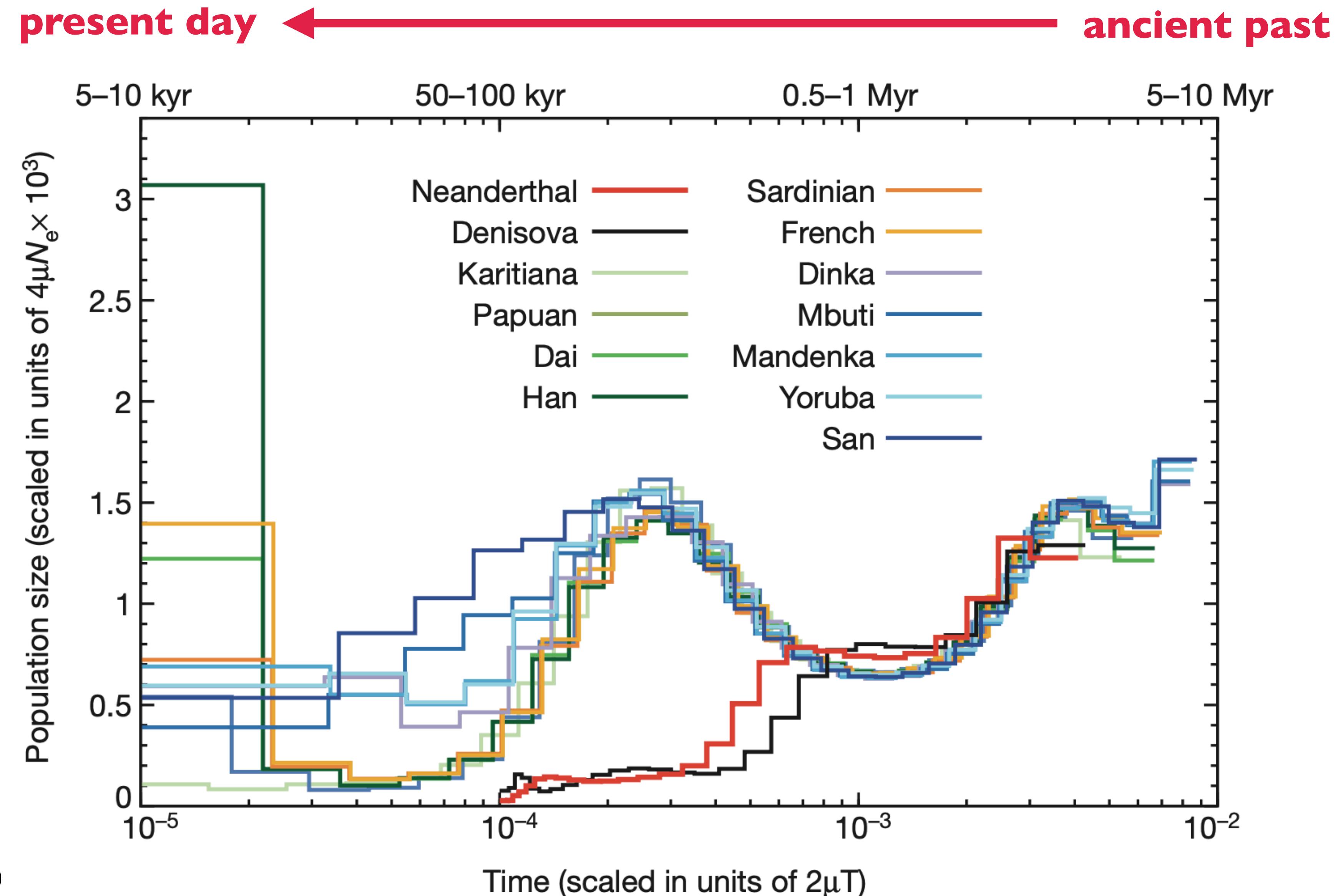


Empirical trajectory of Neanderthal ancestry contradicts the signal of negative selection?



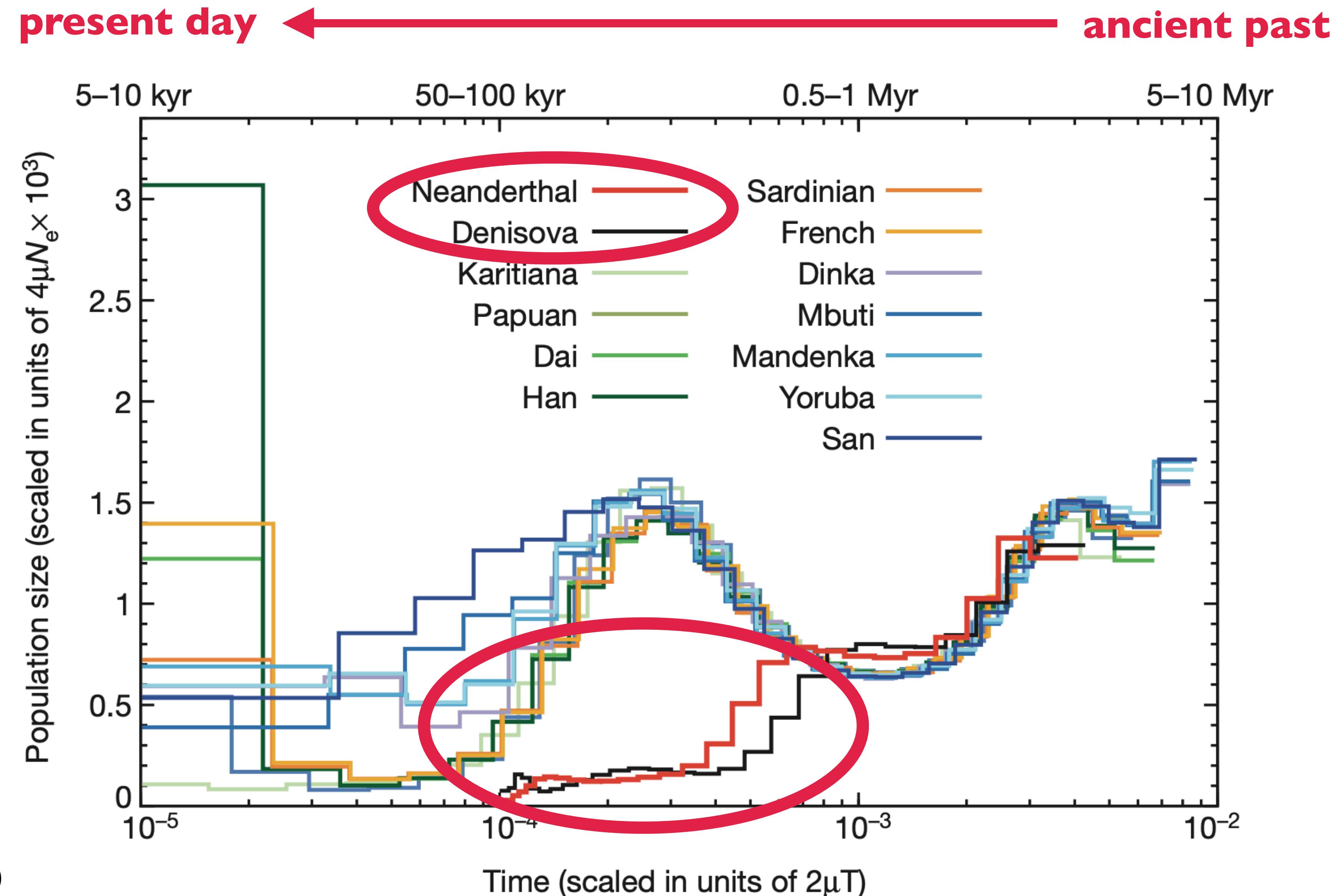
Demographic collapse of archaic human populations

Pairwise Sequentially
Markovian Coalescent
(PSMC) model



Demographic collapse of archaic human populations

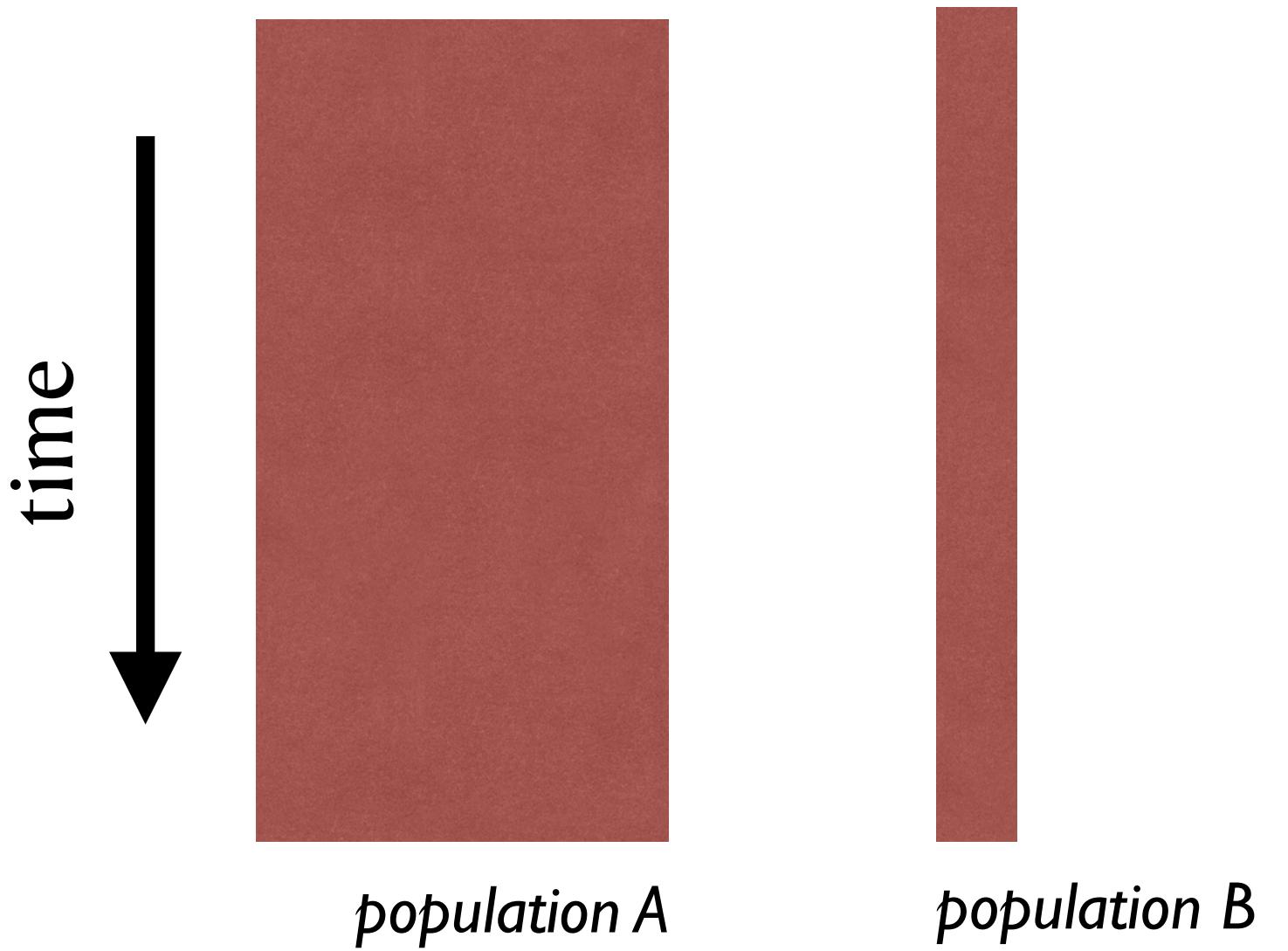
Pairwise Sequentially
Markovian Coalescent
(PSMC) model



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

Nearly-neutral theory of molecular evolution

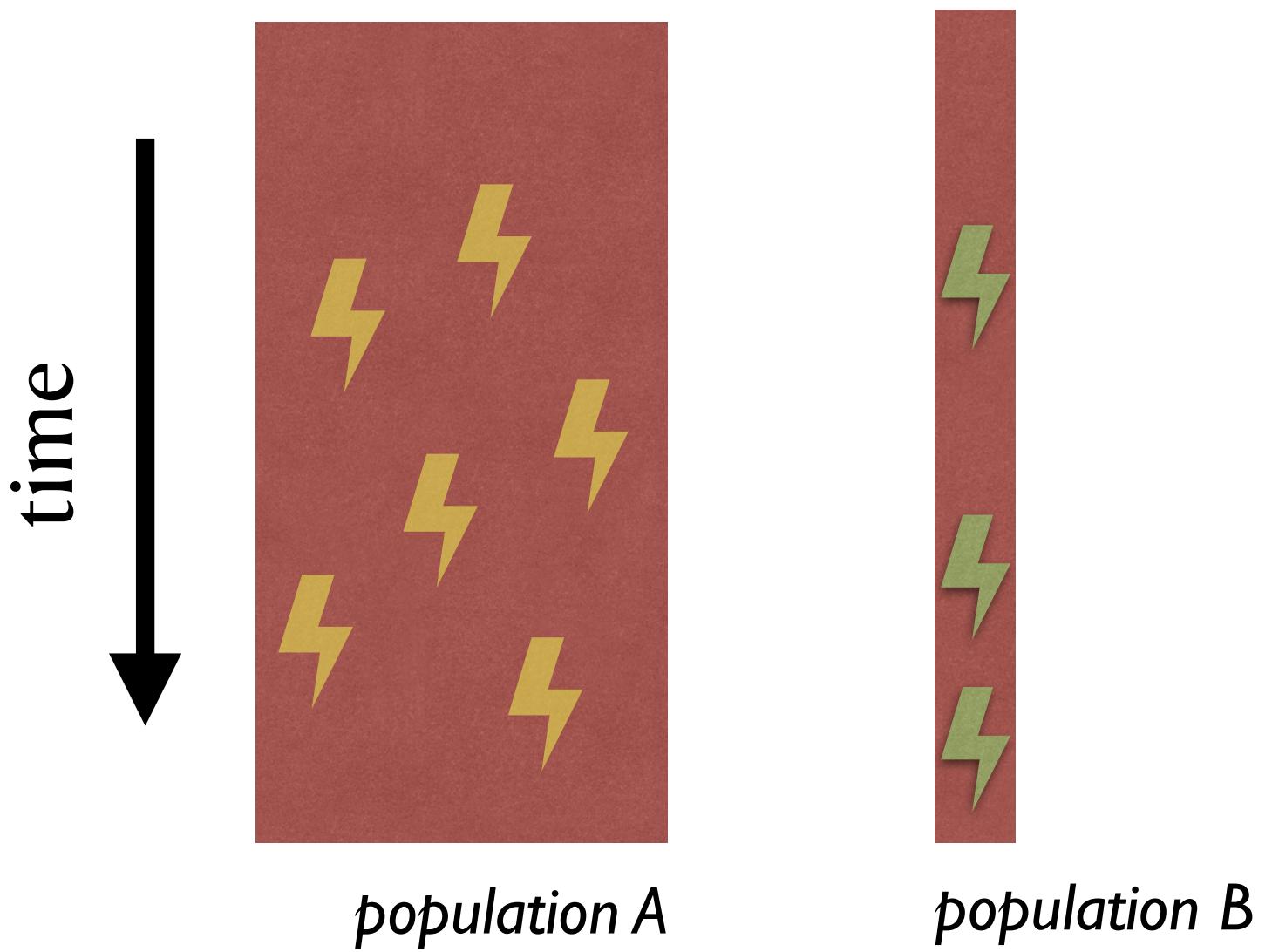
**Tomoko
Ohta**



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

Nearly-neutral theory of molecular evolution

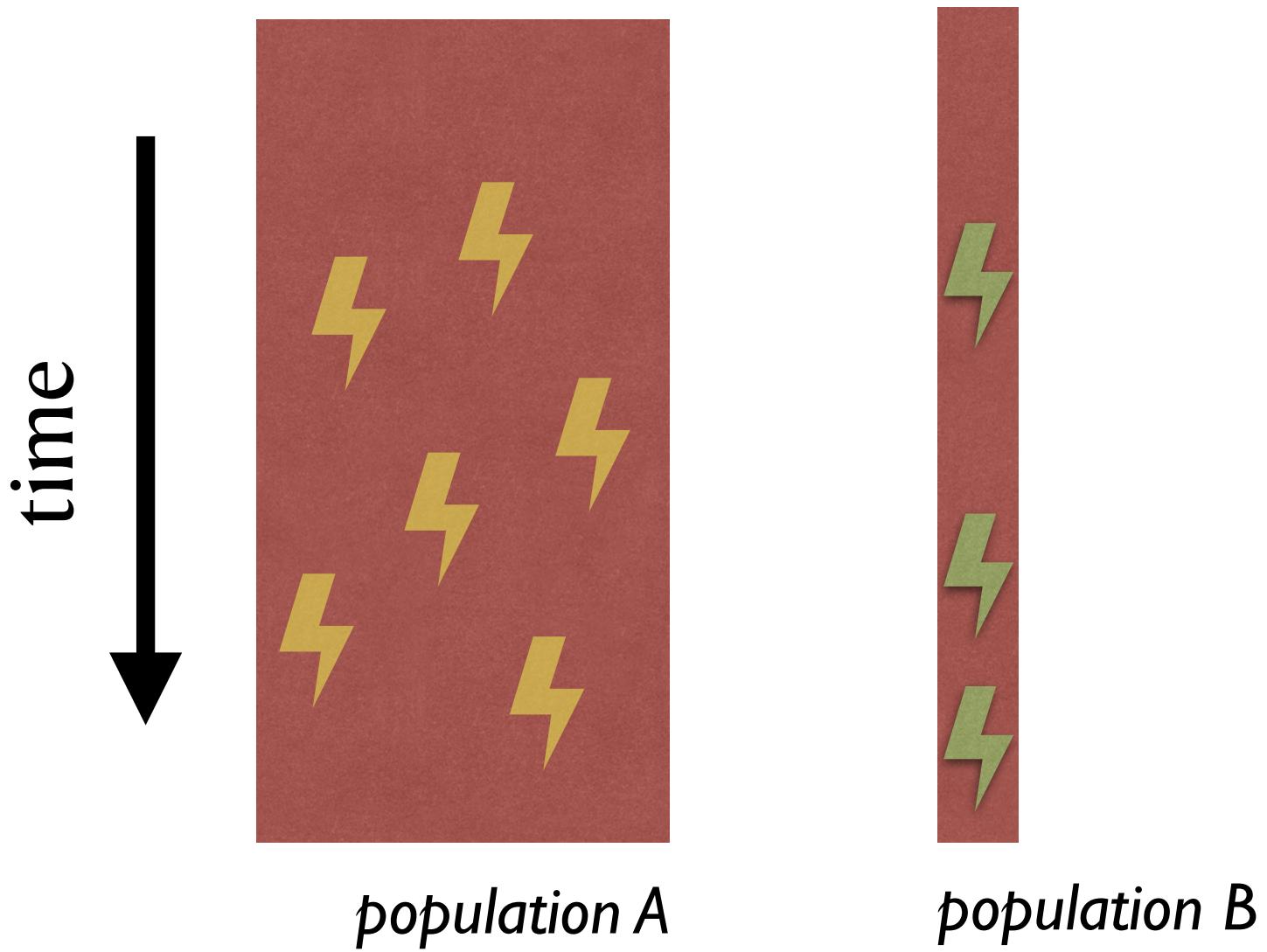
**Tomoko
Ohta**



We know that genomes accumulate mutations

Nearly-neutral theory of molecular evolution

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

We know that genomes accumulate mutations

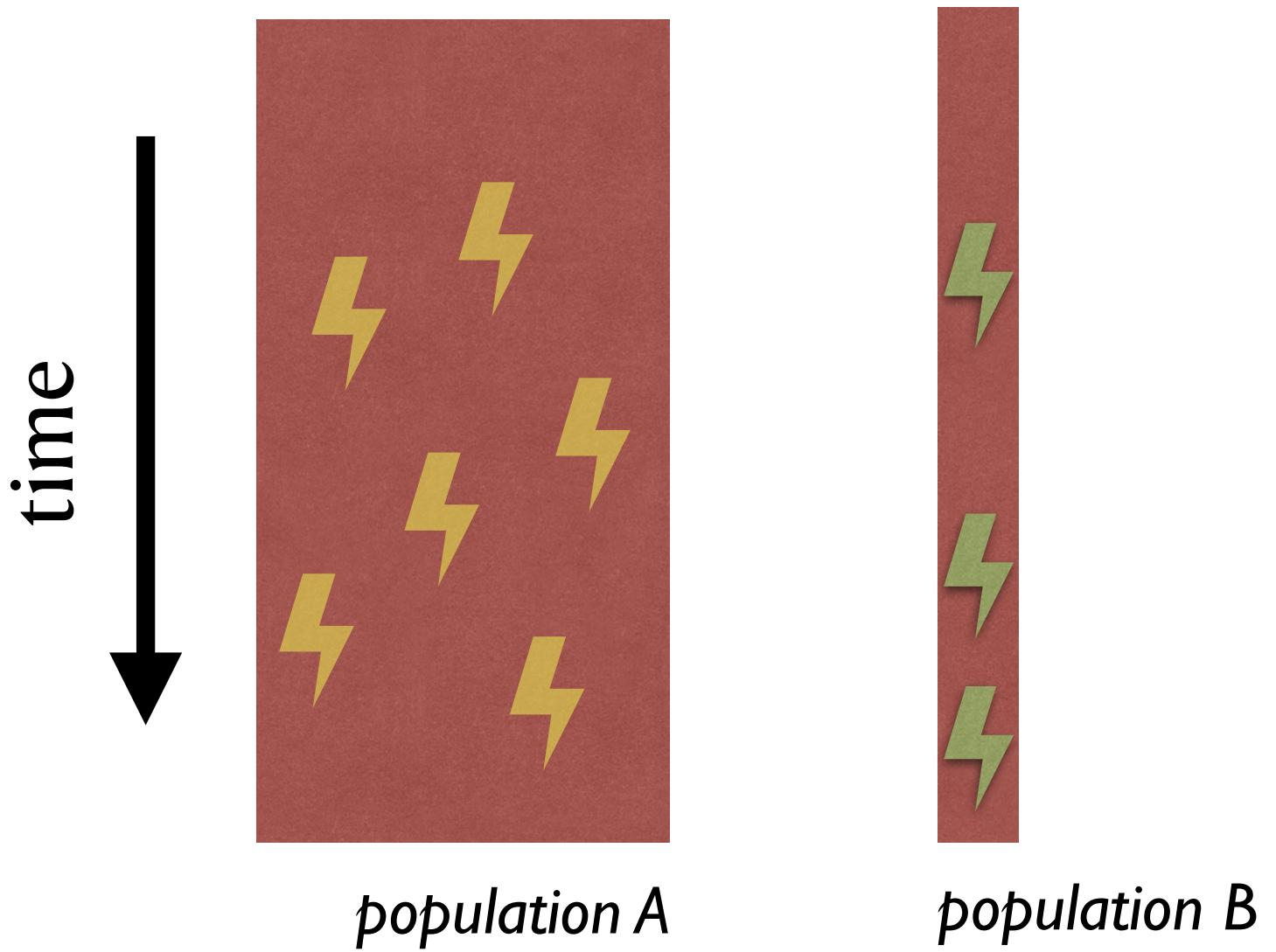
- a tiny number are extremely negative (**lethal**)
- a tiny number are positive (**adaptive**)
- a large number does nothing (**neutral**)
- many are “mildly deleterious” (**nearly neutral**)

Nearly-neutral theory of molecular evolution

**Tomoko
Ohta**



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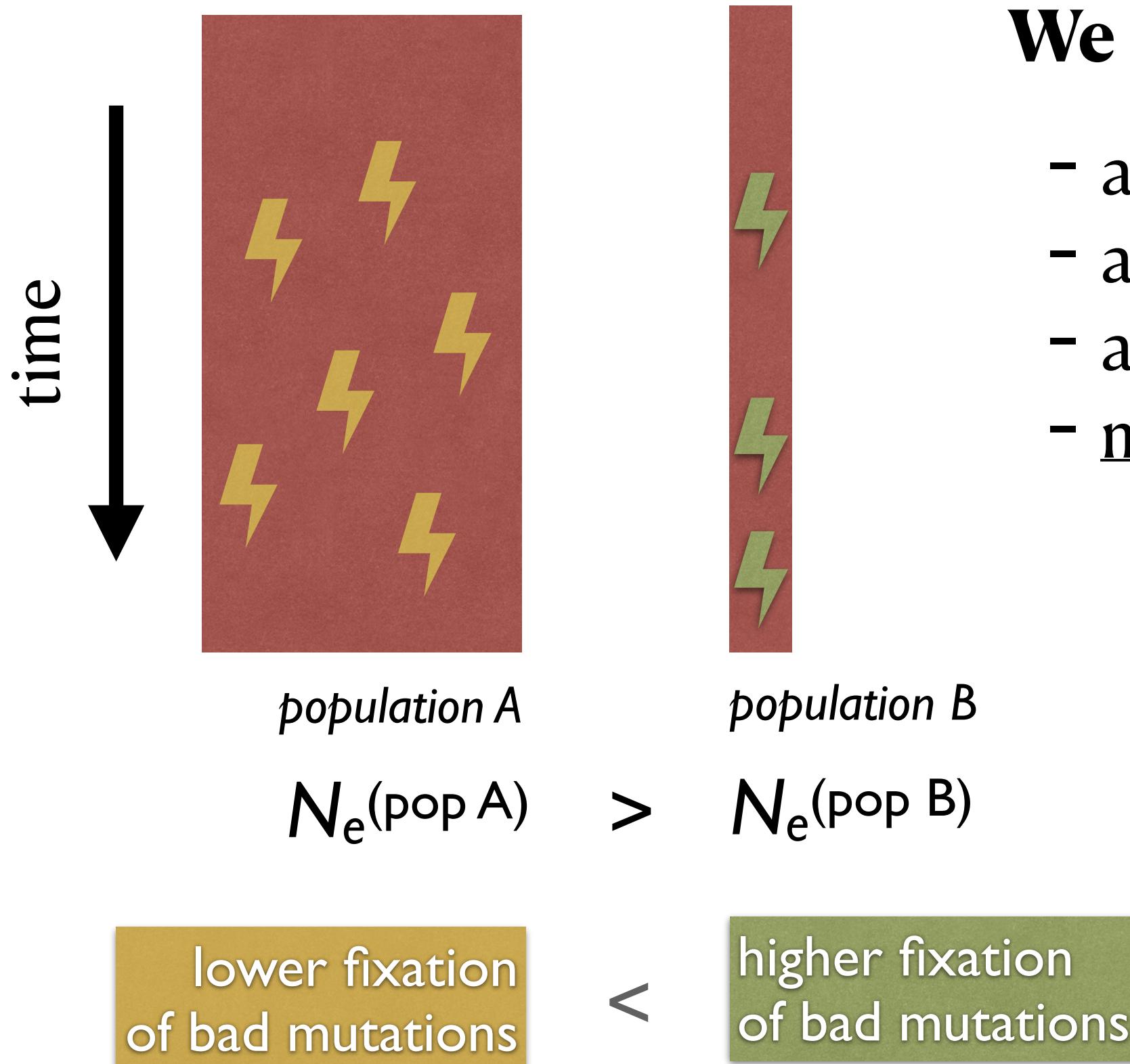
**Efficacy of negative selection
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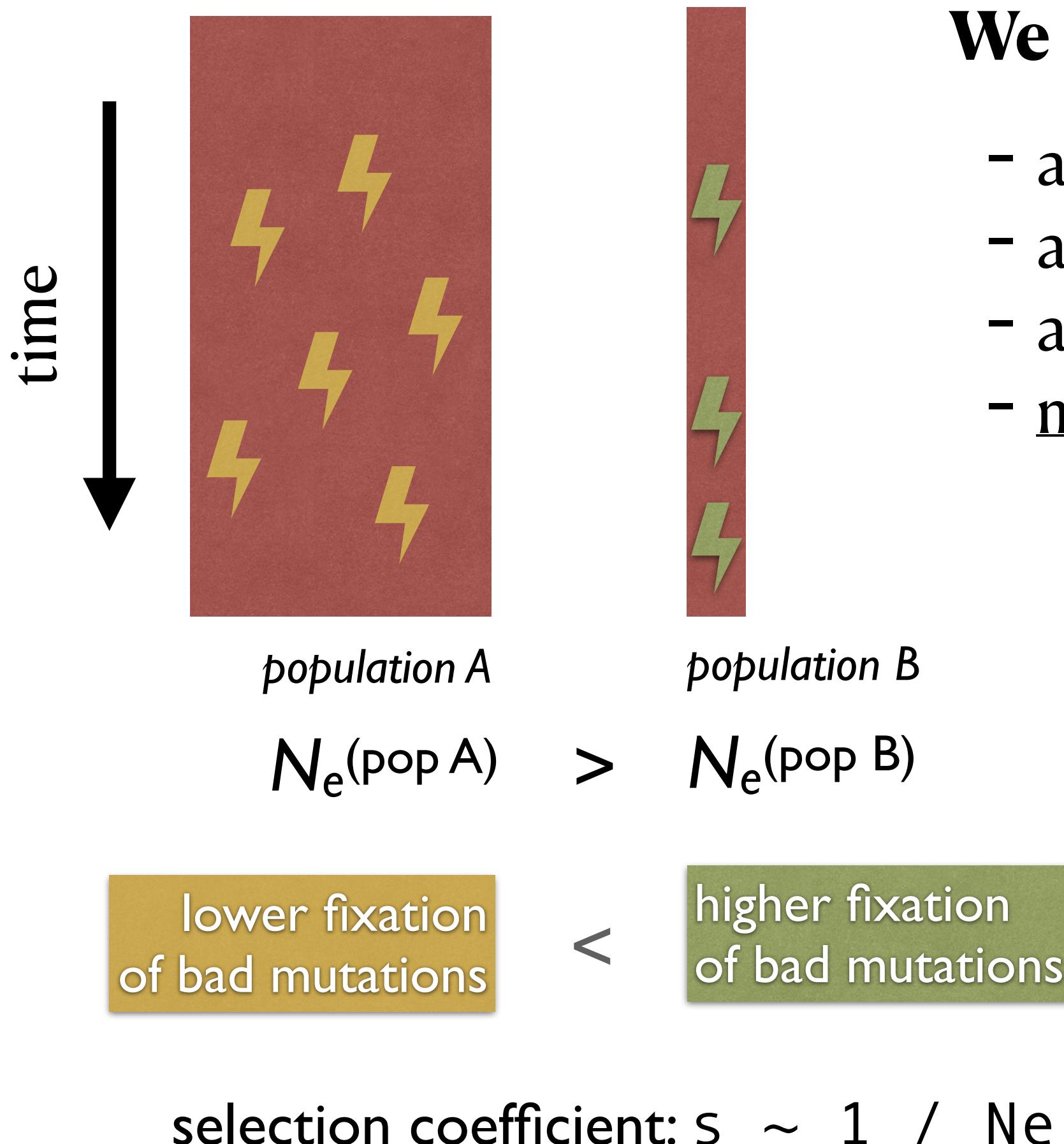
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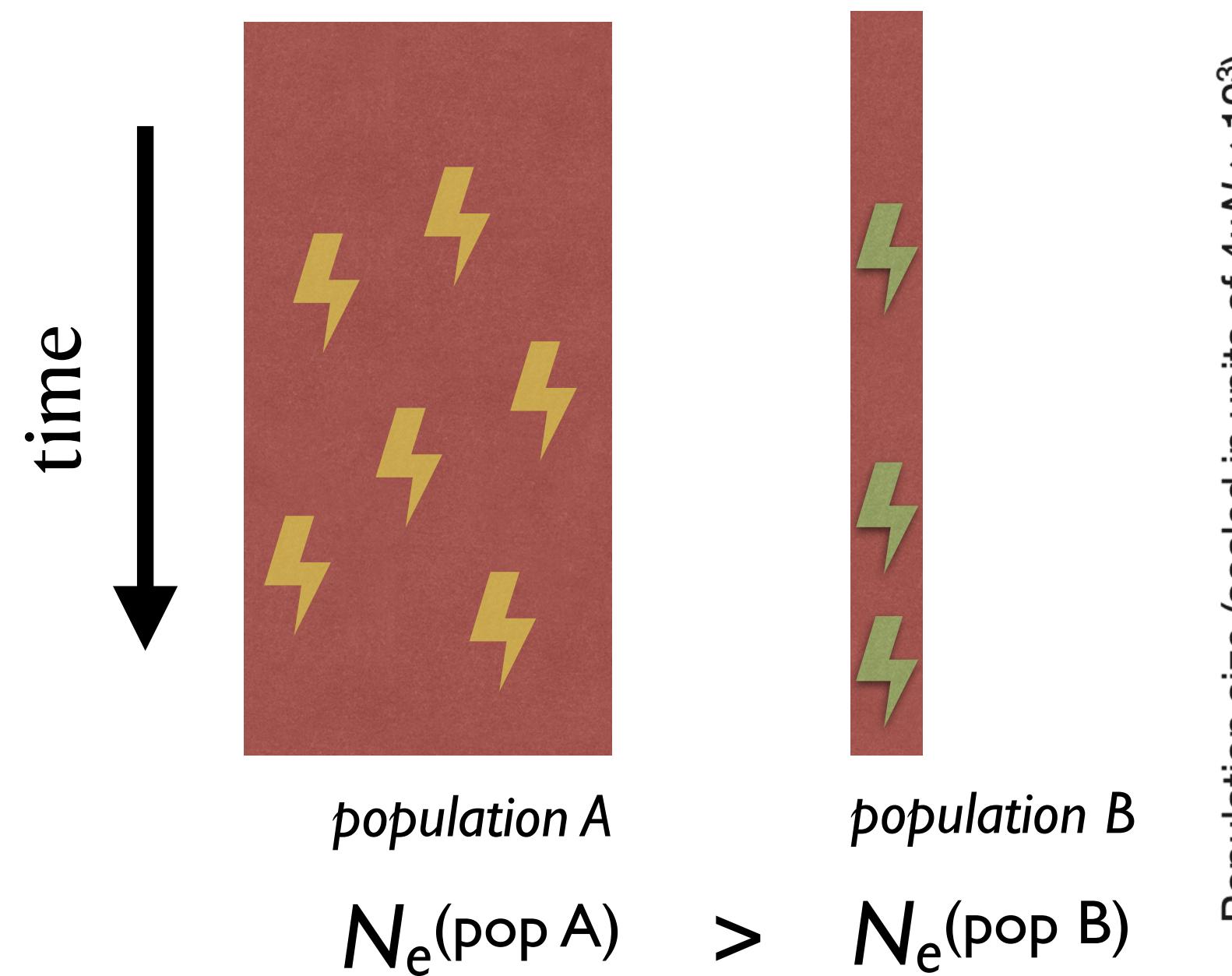
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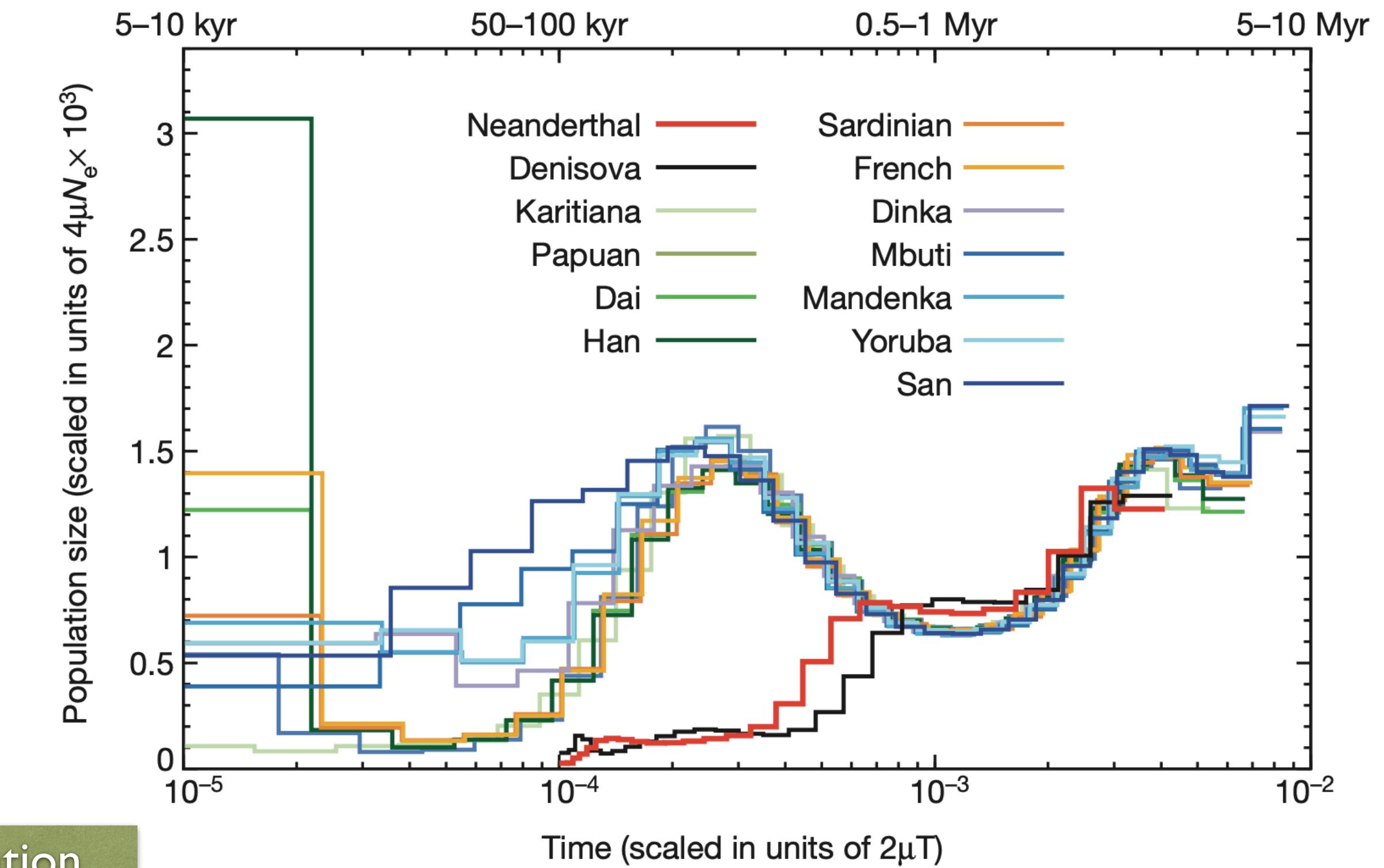


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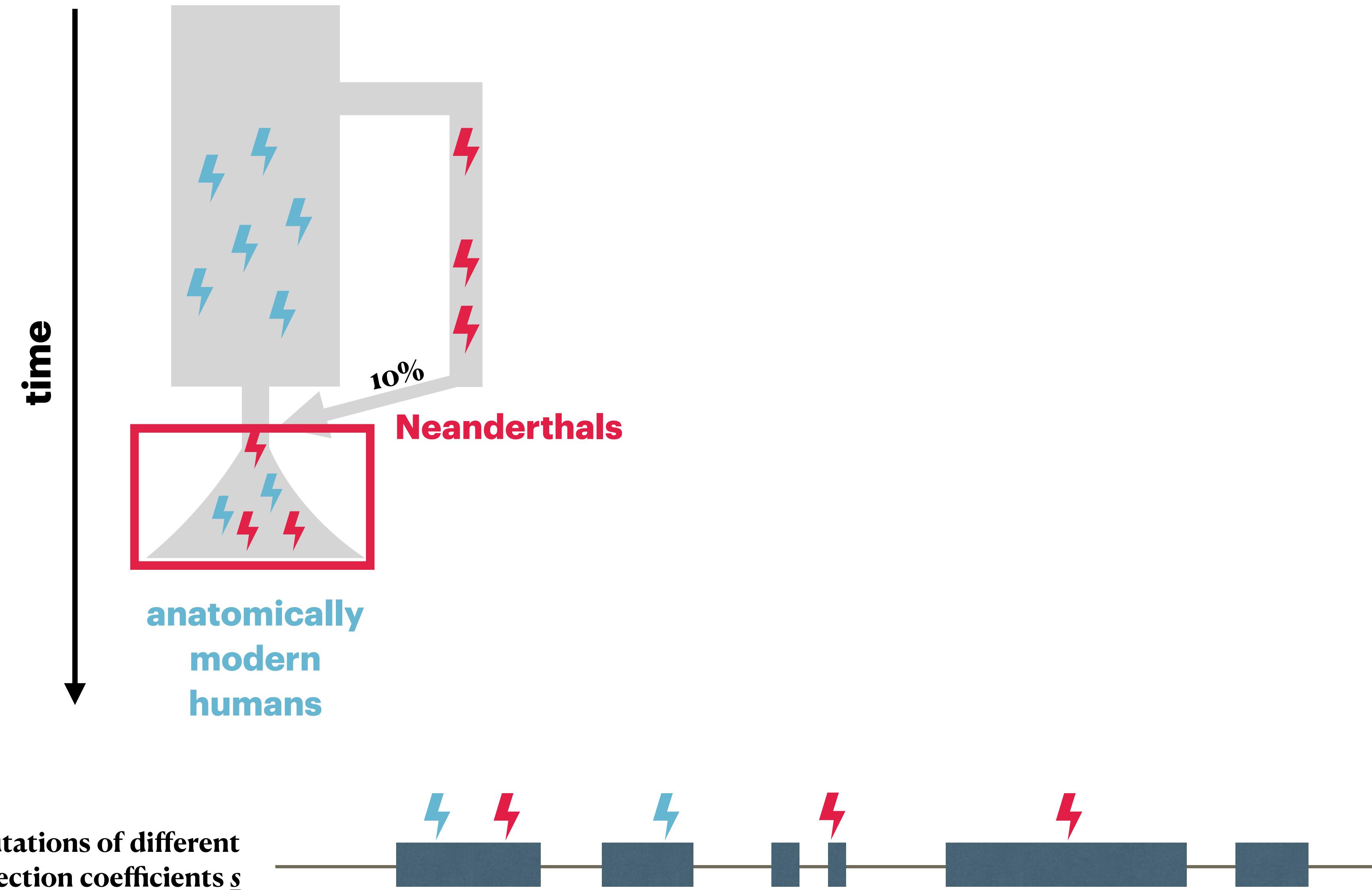
"anatomically
modern humans"

"Neanderthals"

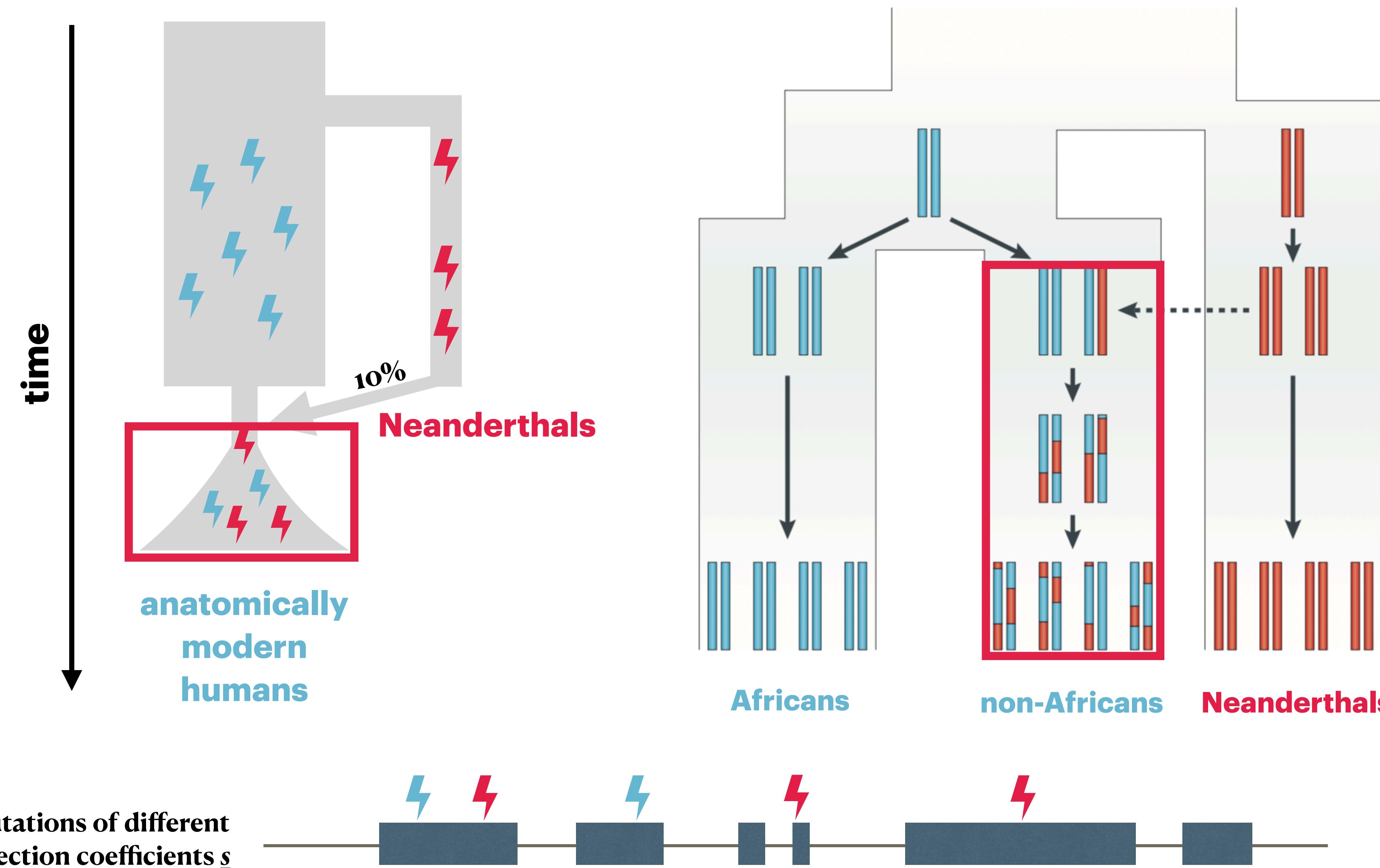


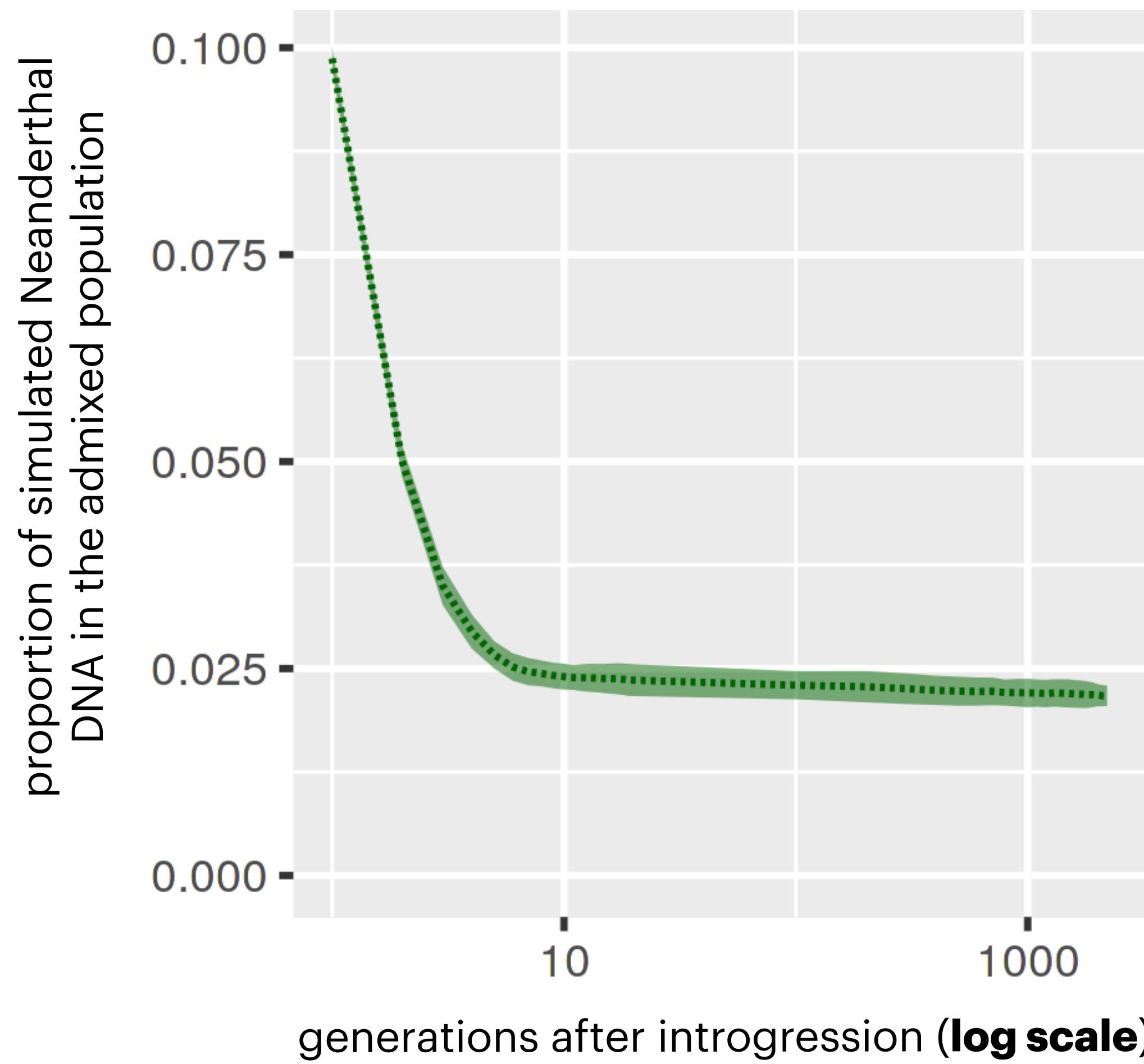
Ohta (Nature, 1973)

Simulations: tracing the trajectory of introgressed DNA over time

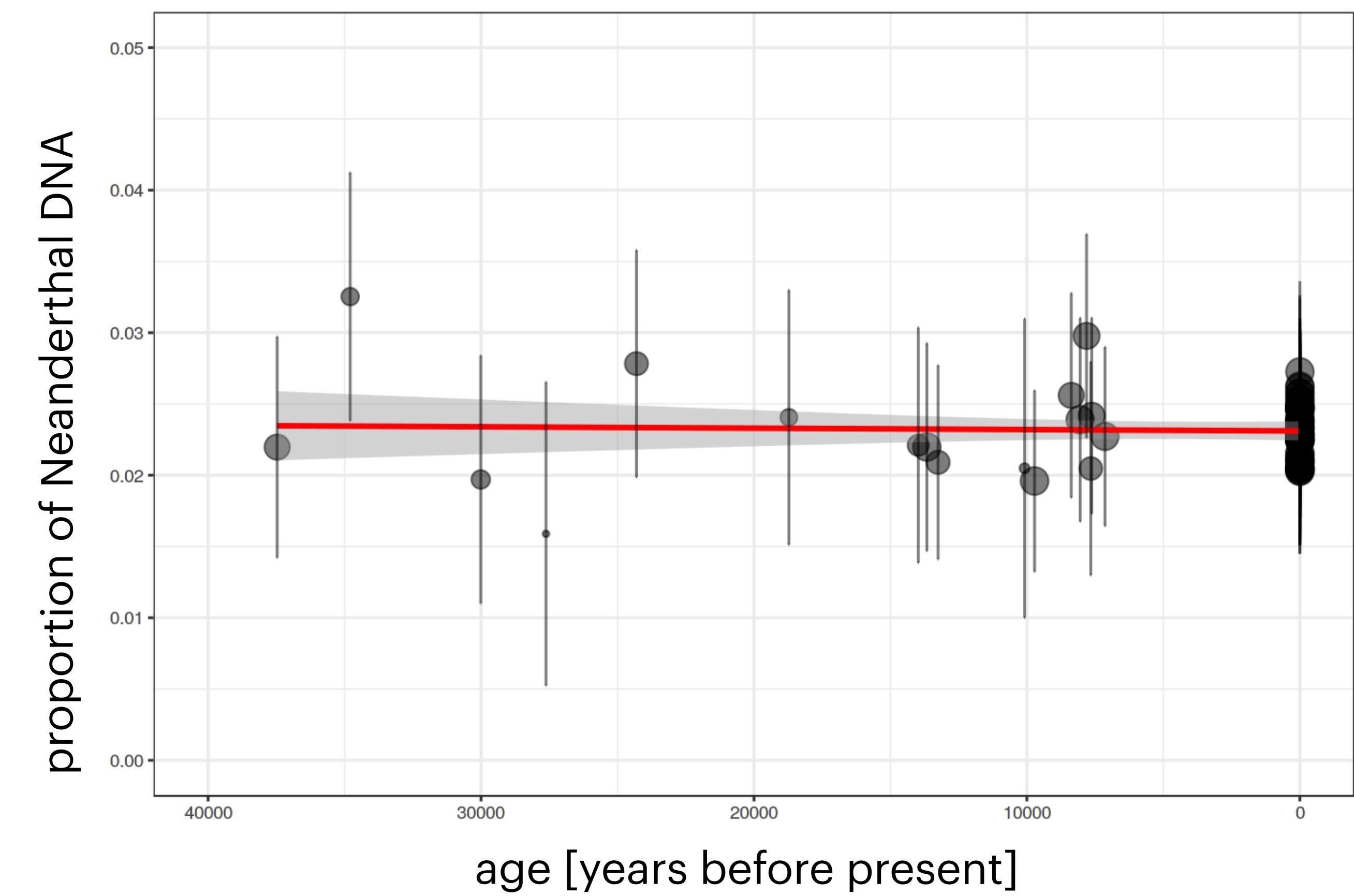
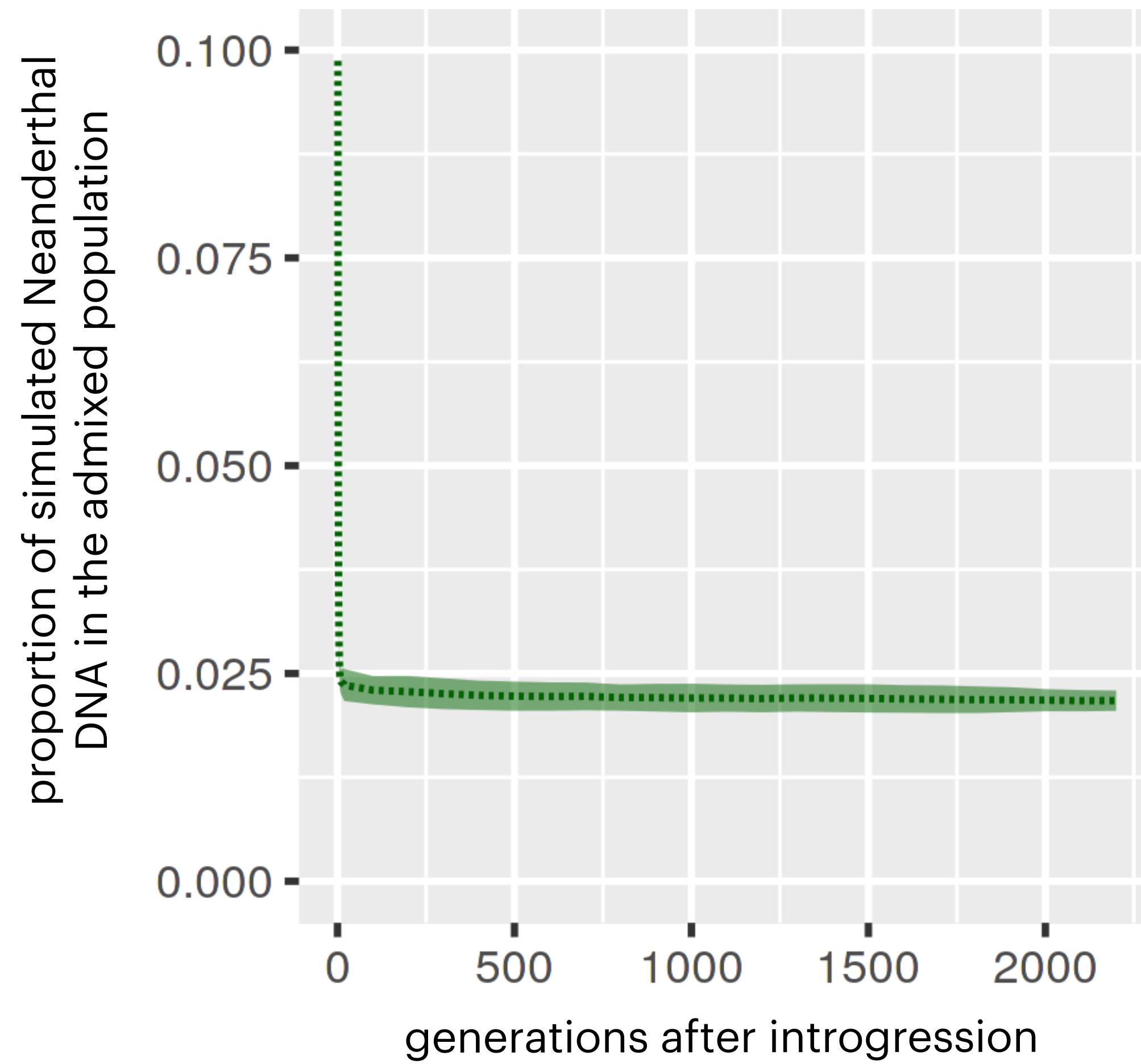


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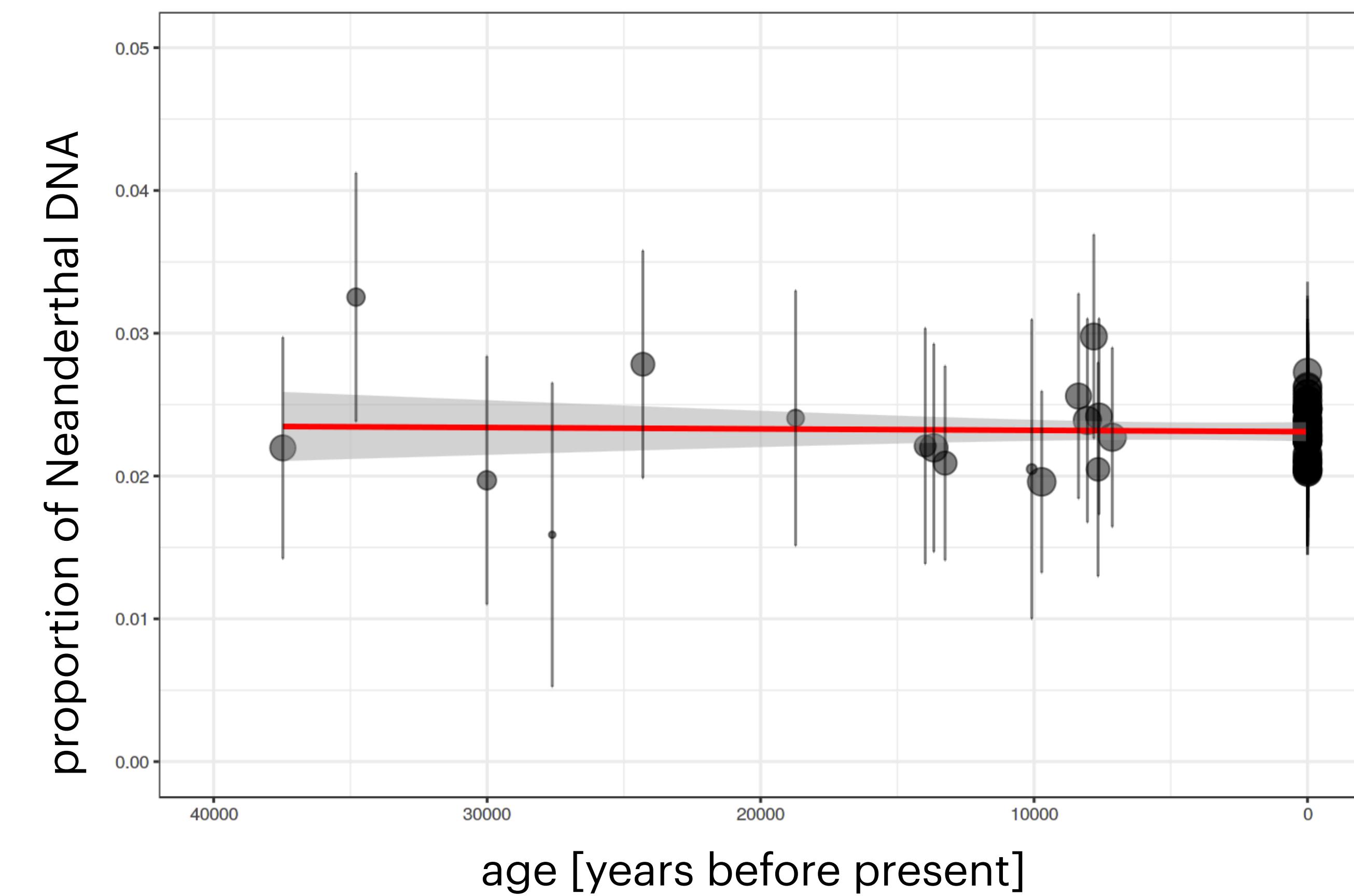
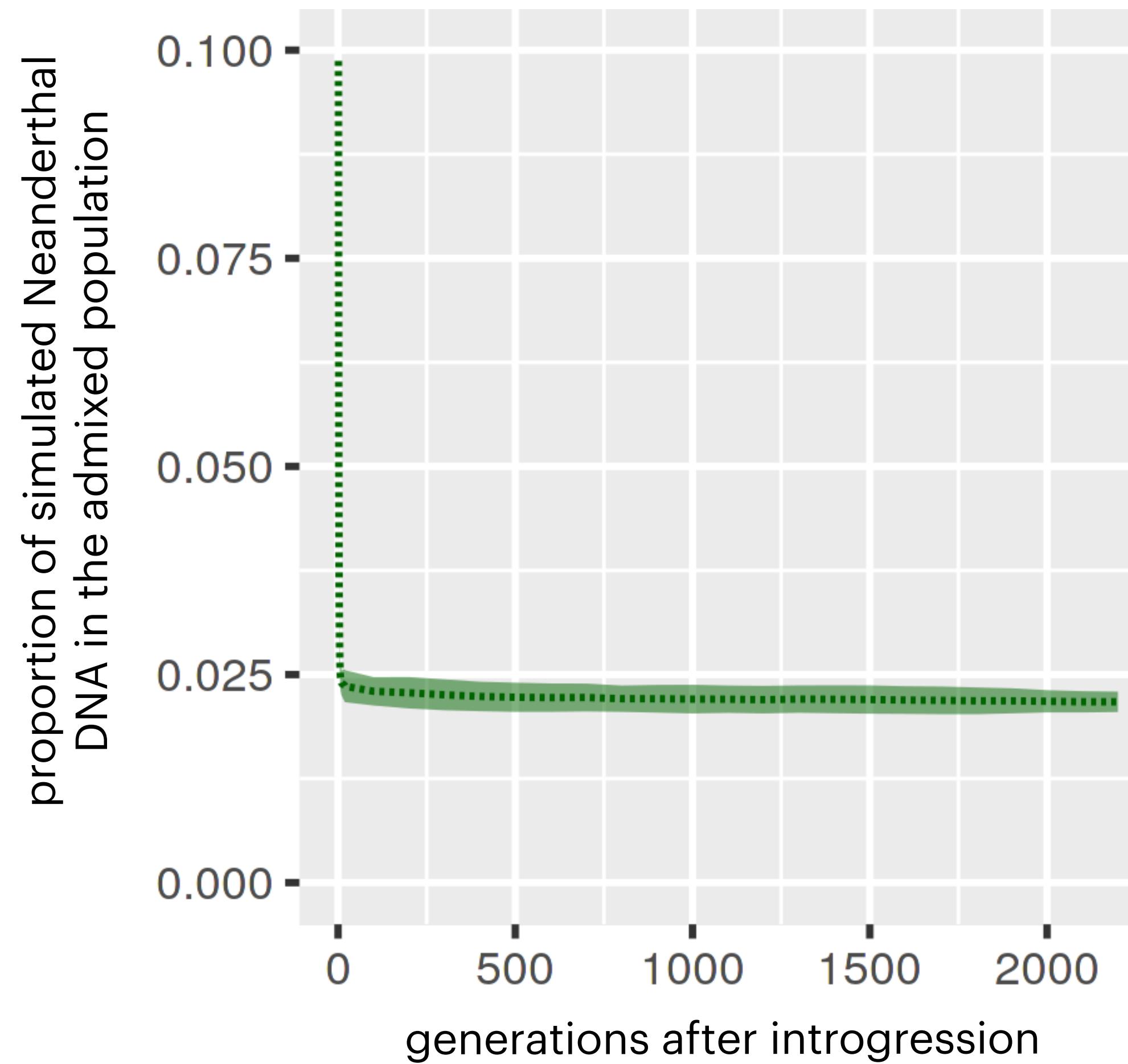


Harris et al. (2016); Juric et al. (2016); Petr et al. (2019)



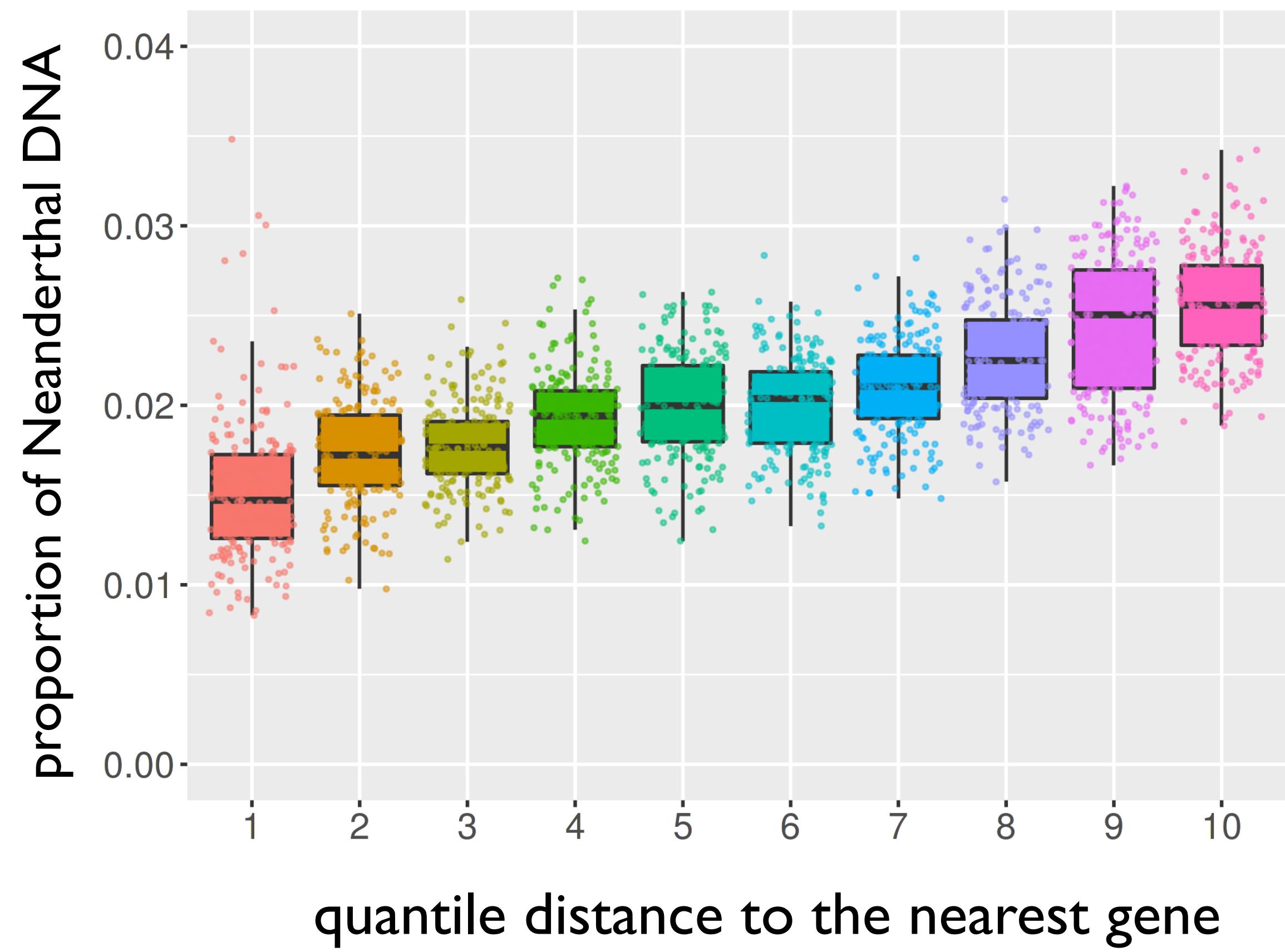
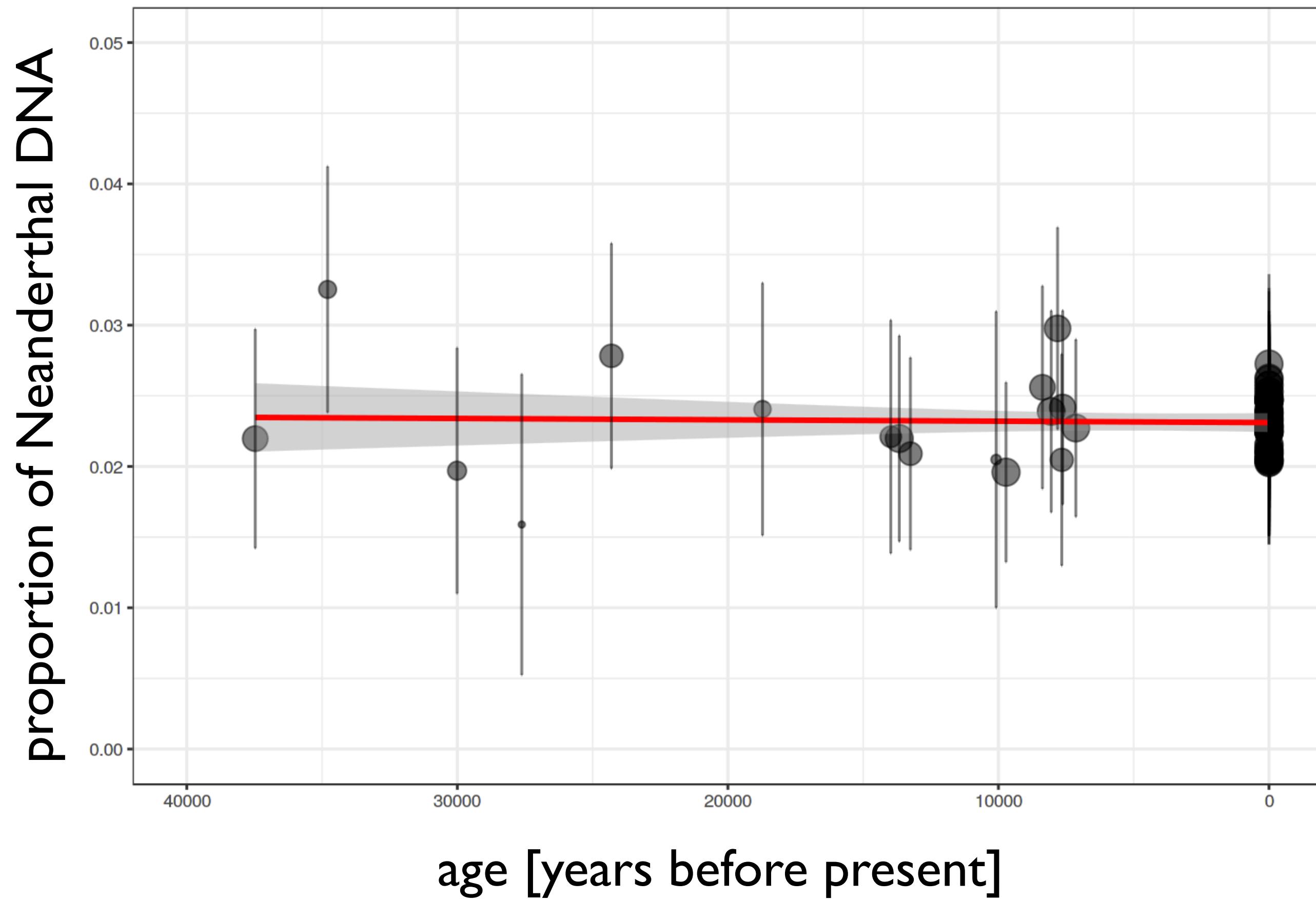
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Neanderthal ancestry trajectory matches simulations...

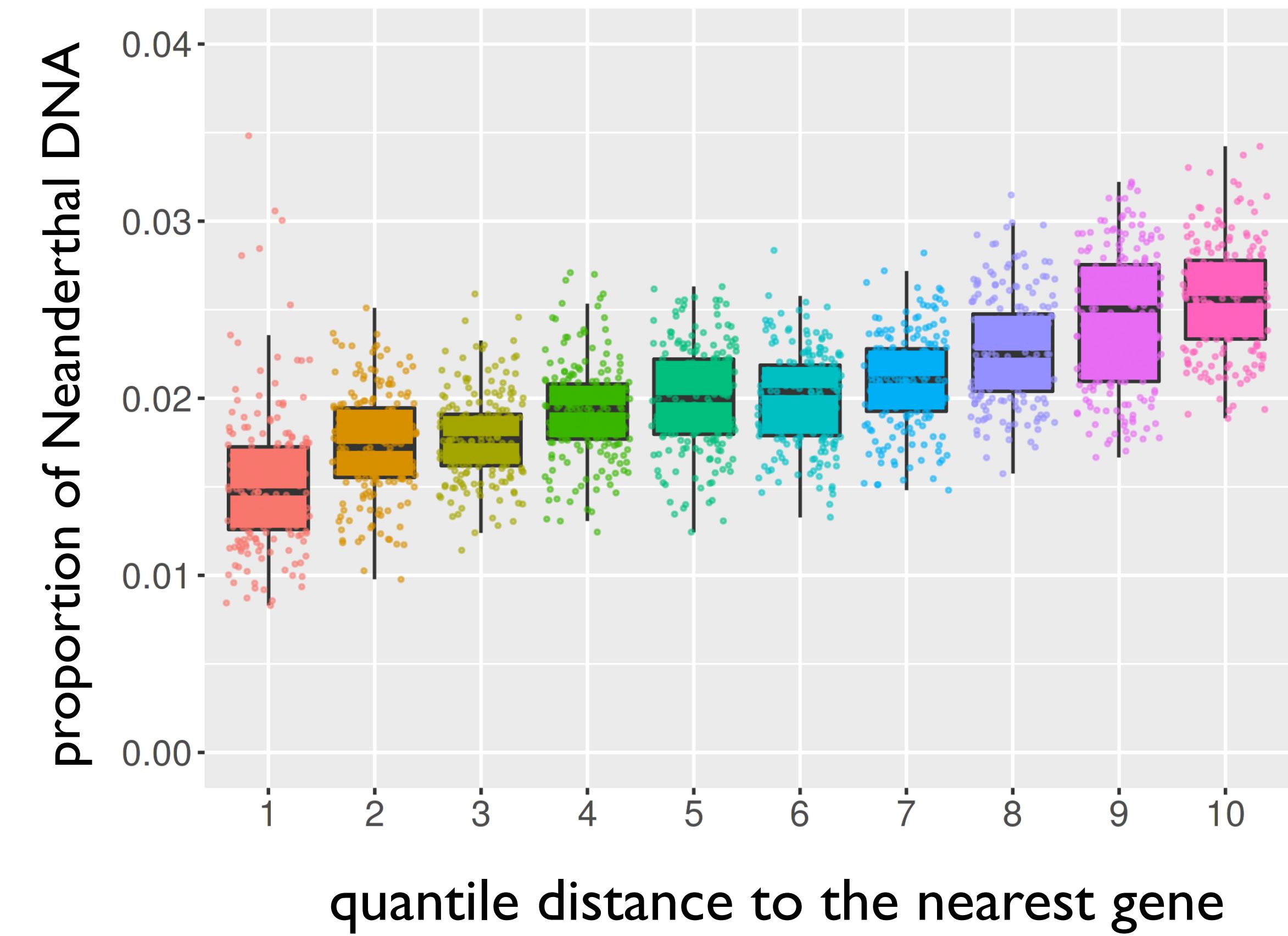
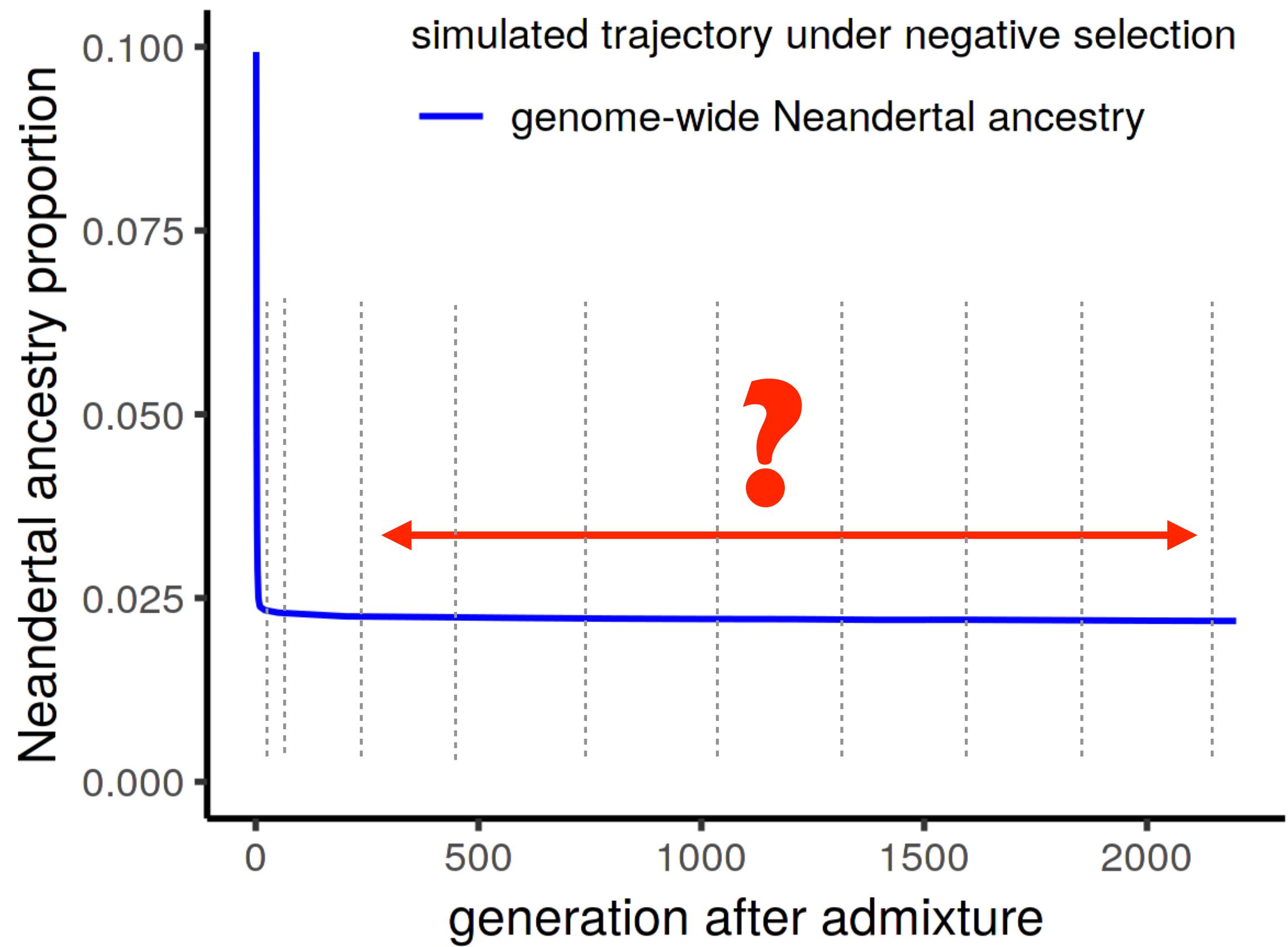


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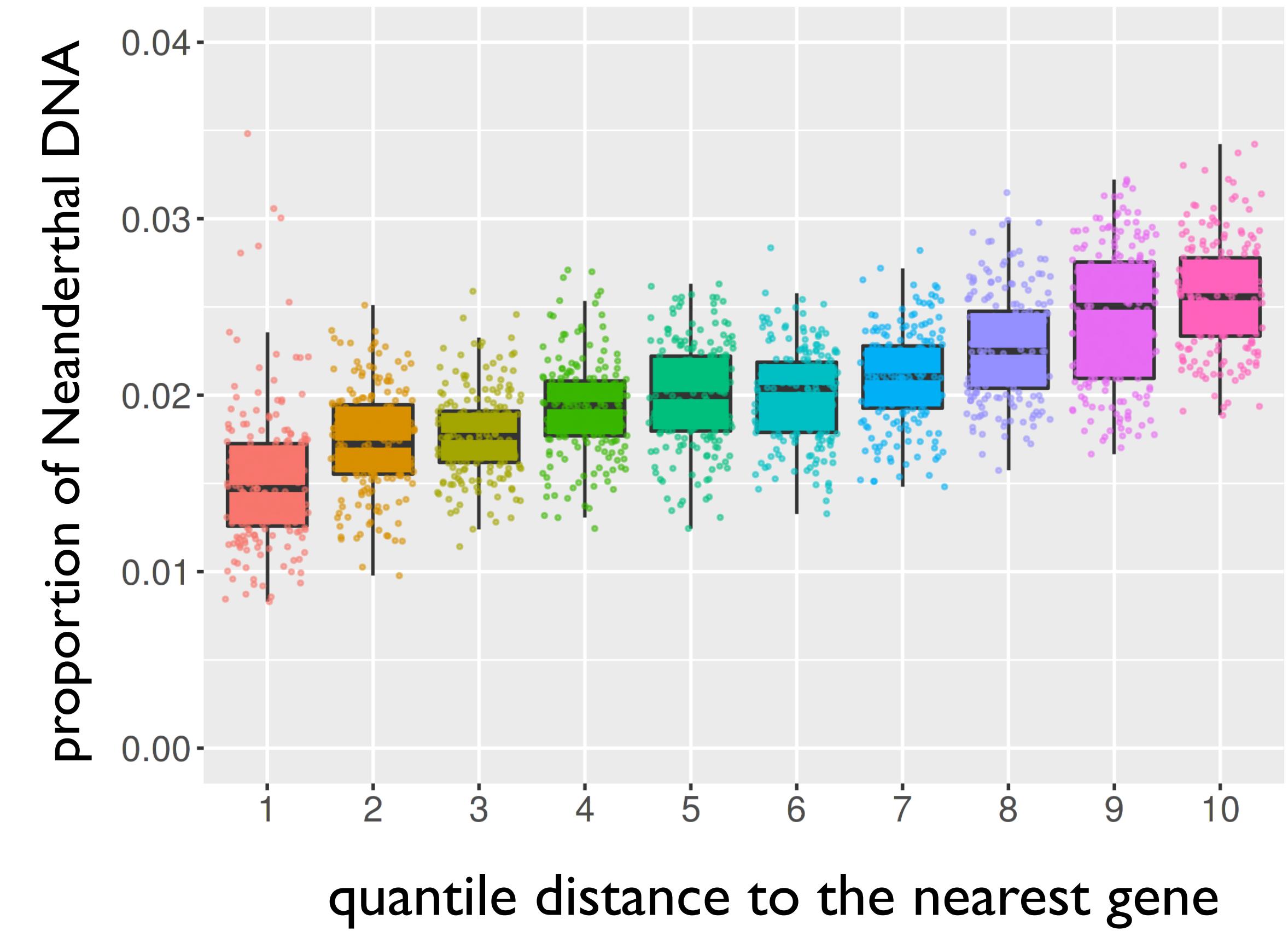
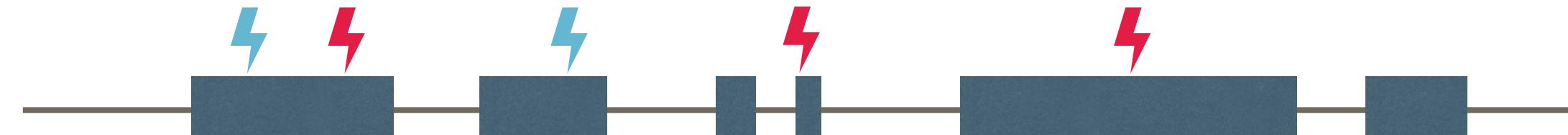
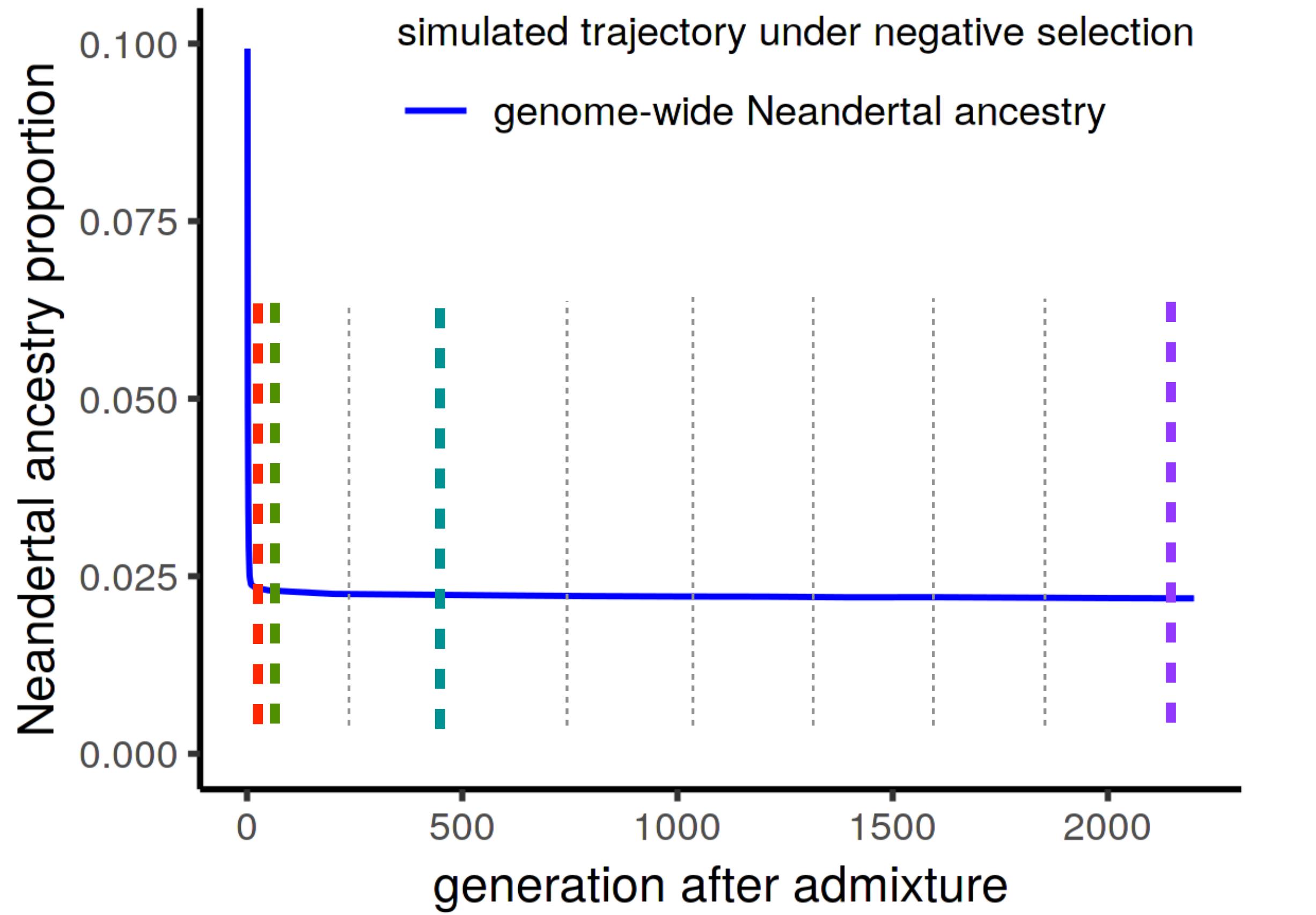
... what about the whole-genome negative selection signal?



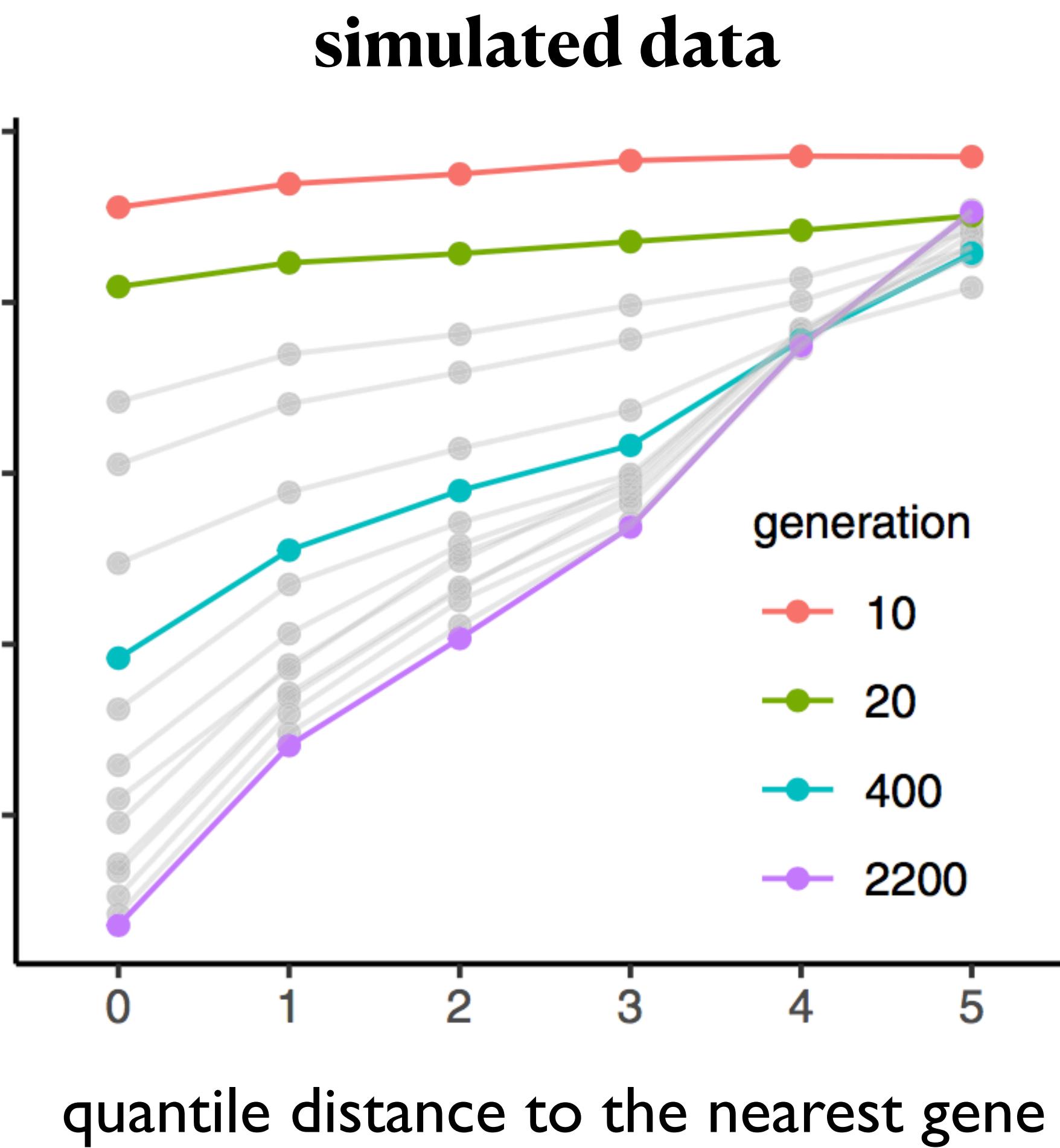
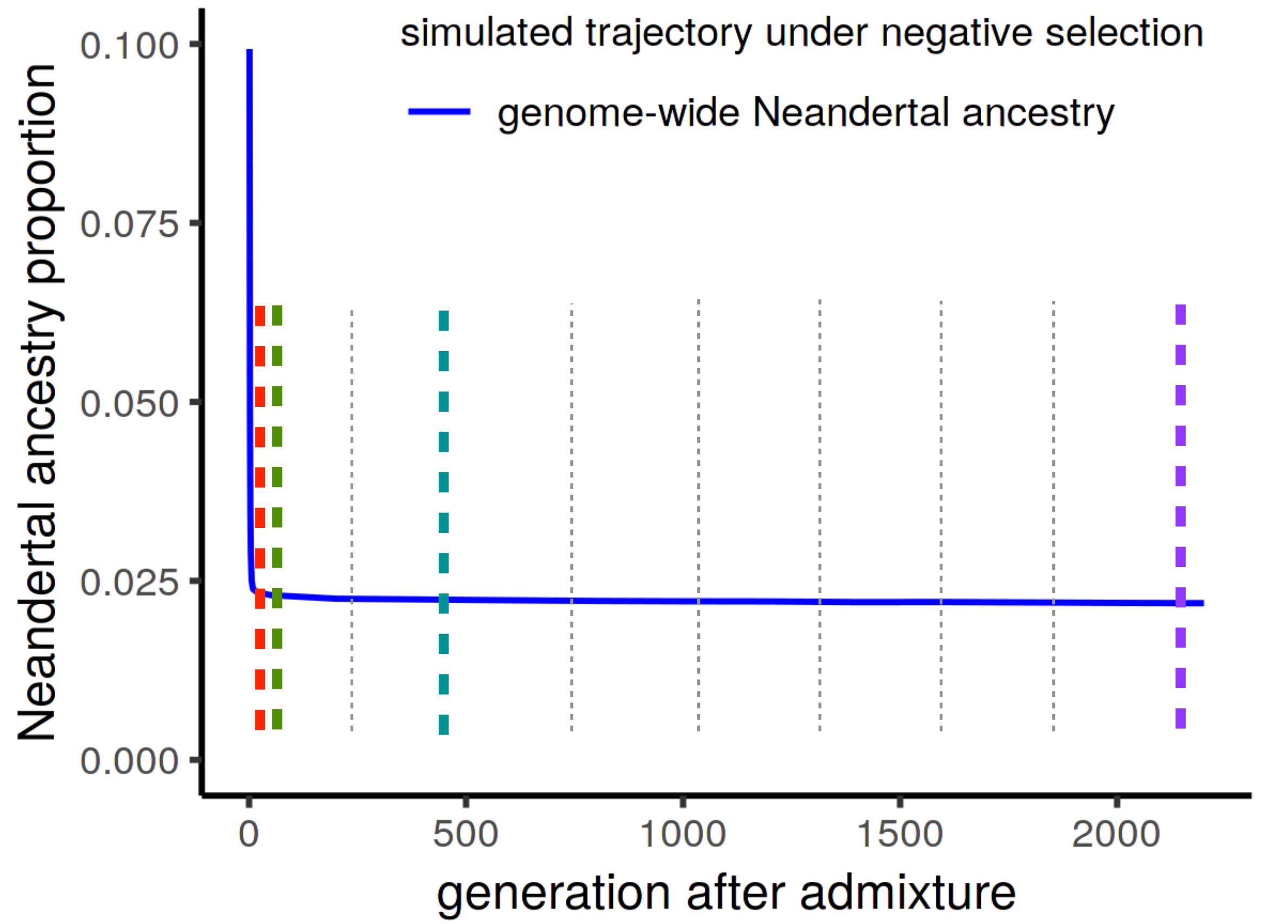
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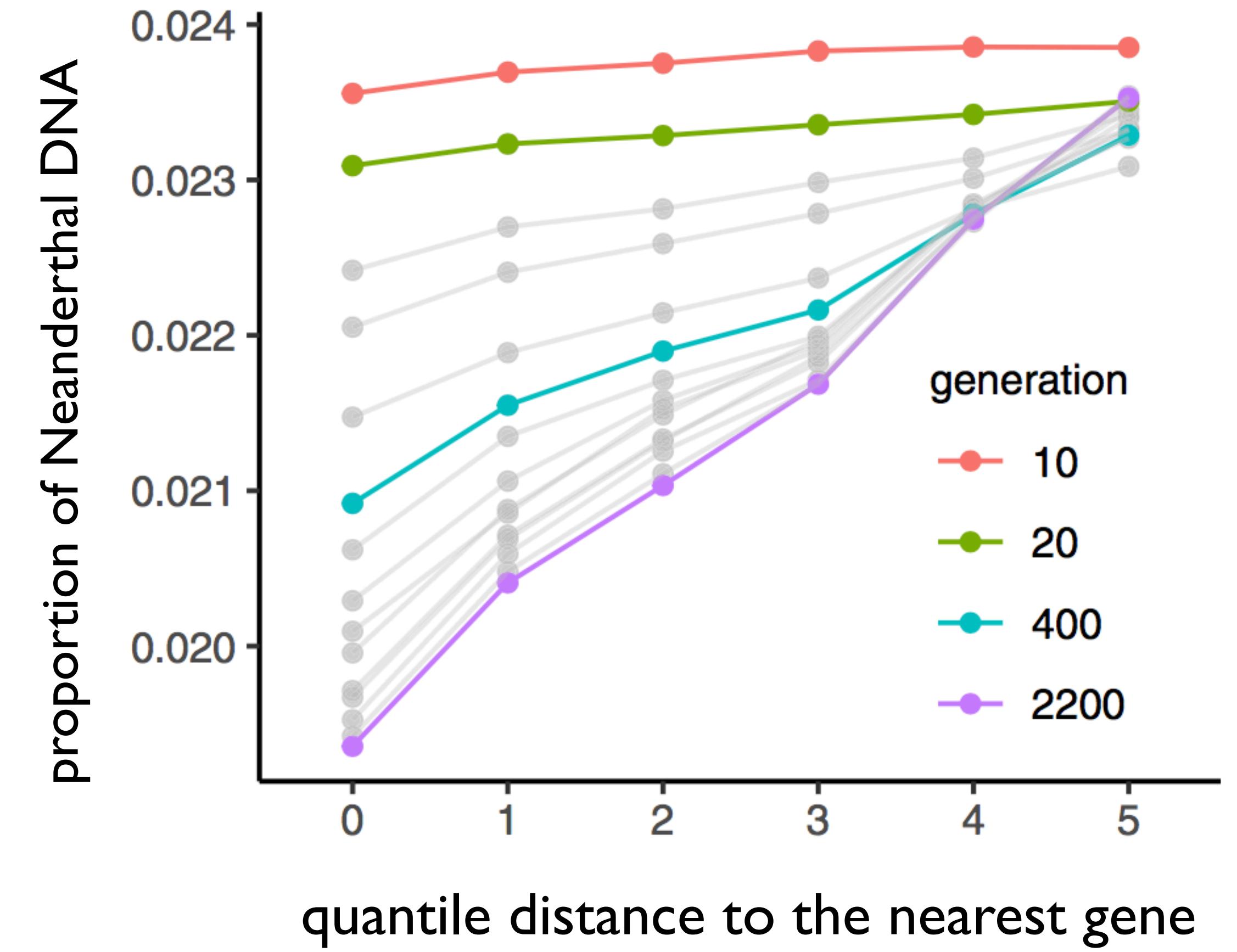
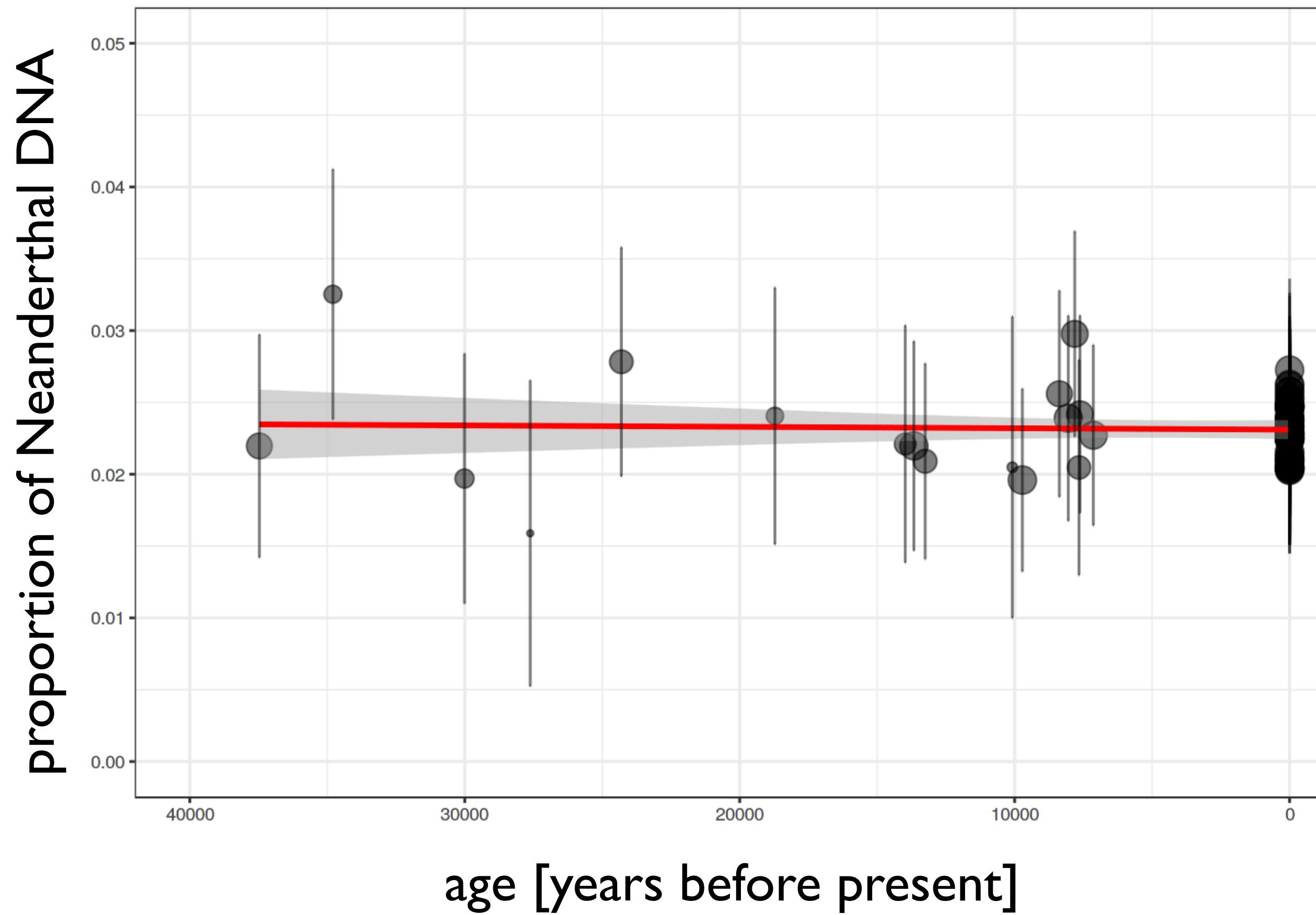
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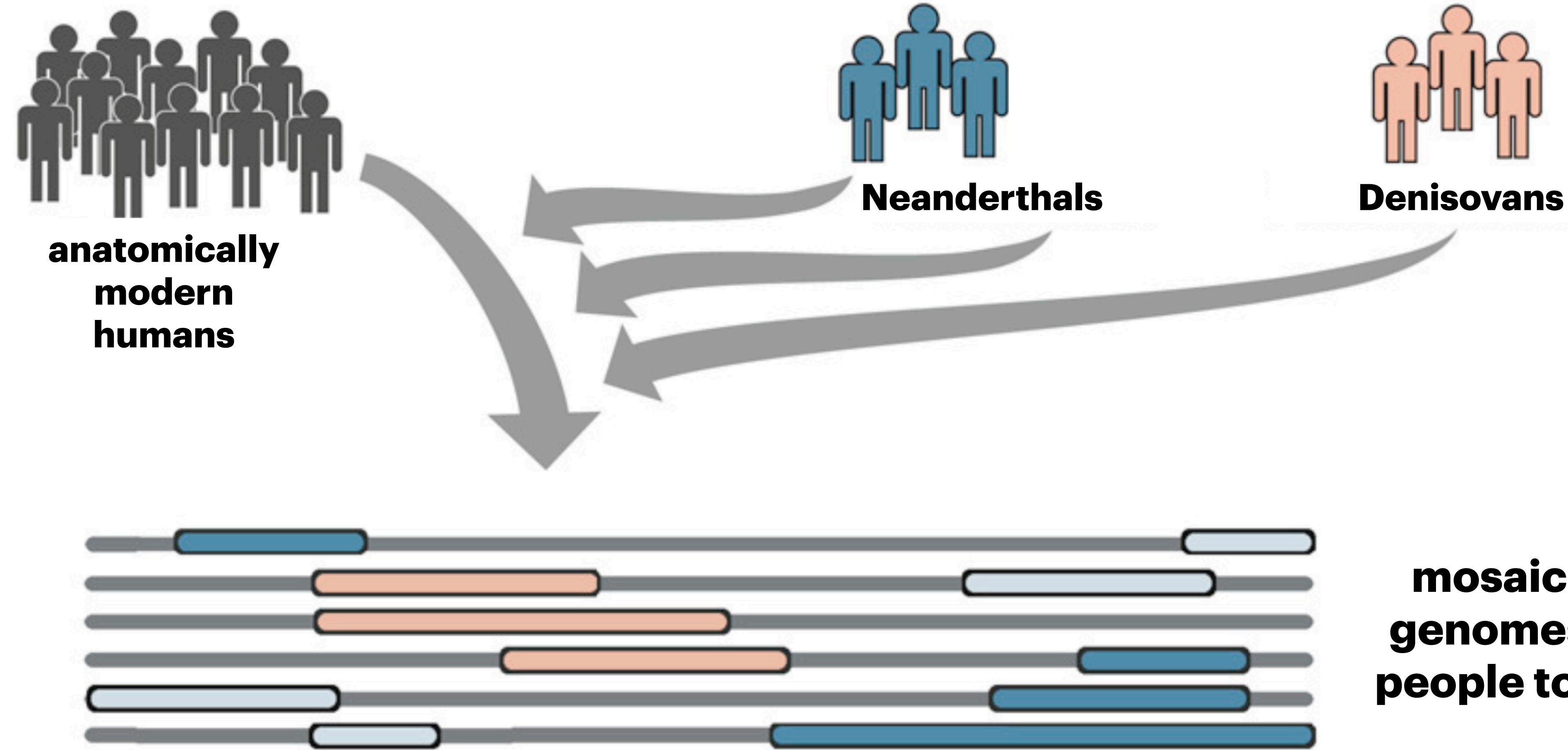
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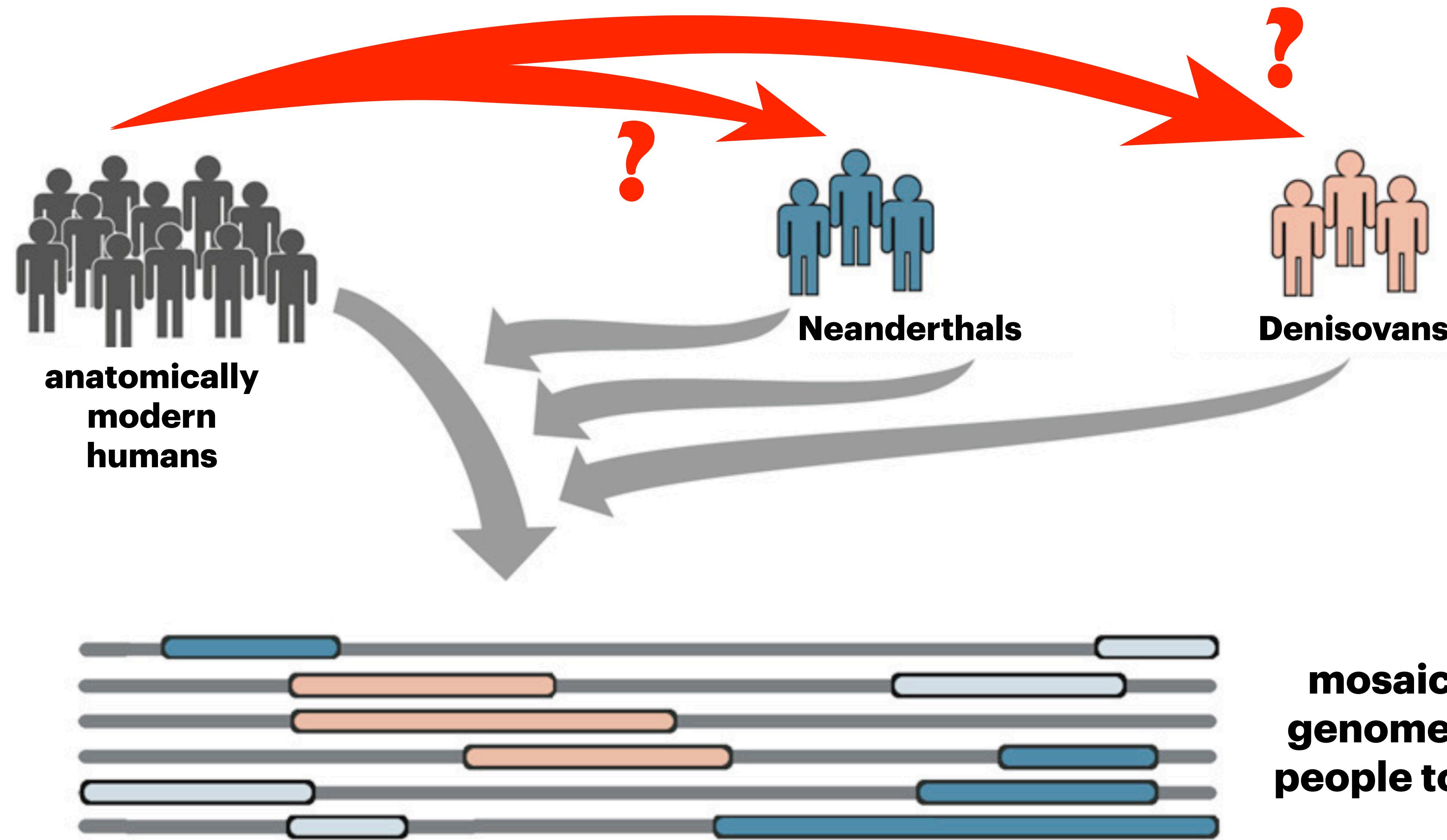
Both empirical results match population genetic theory!



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

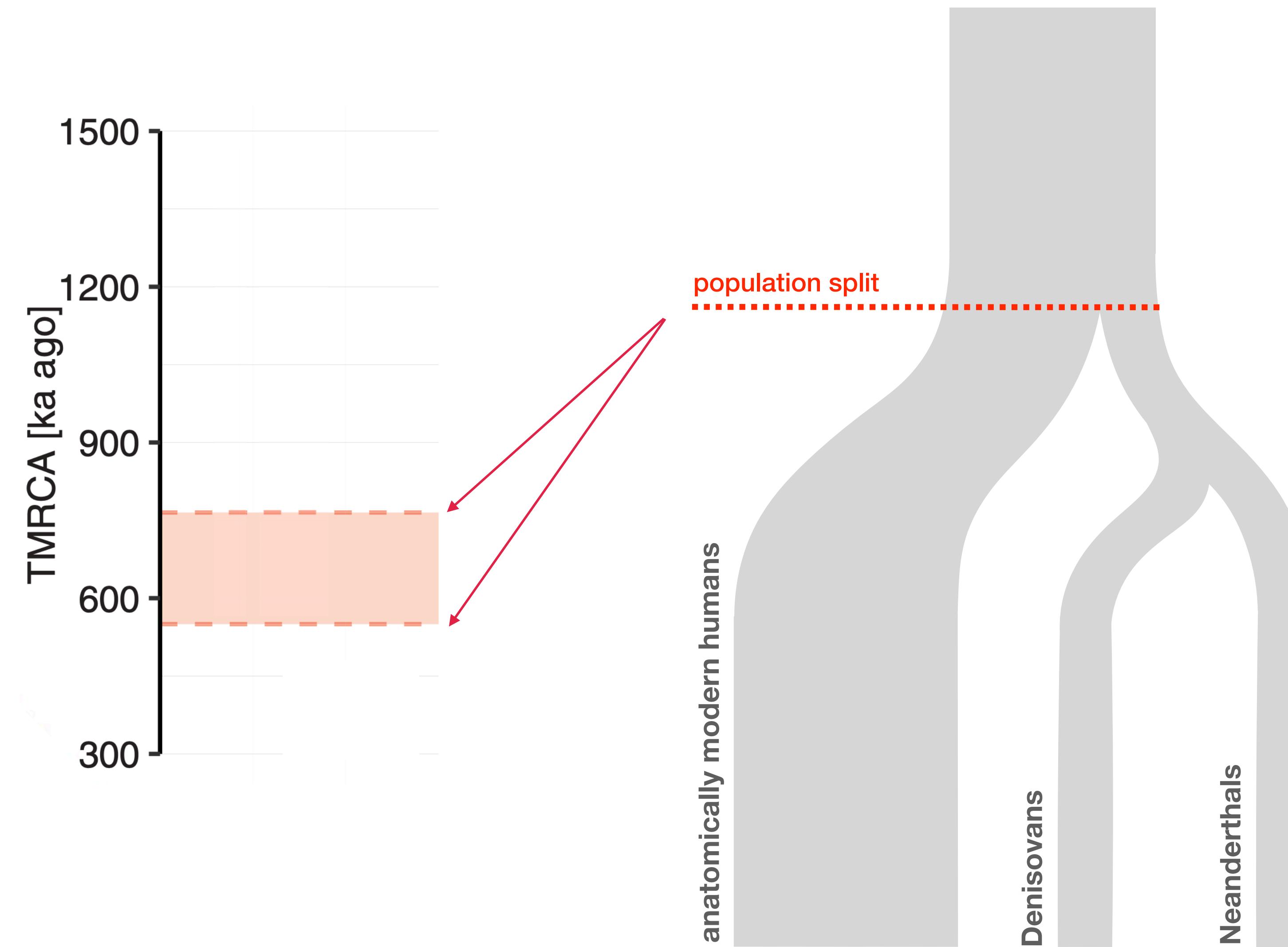


adapted from Browning et al., Cell, 2018

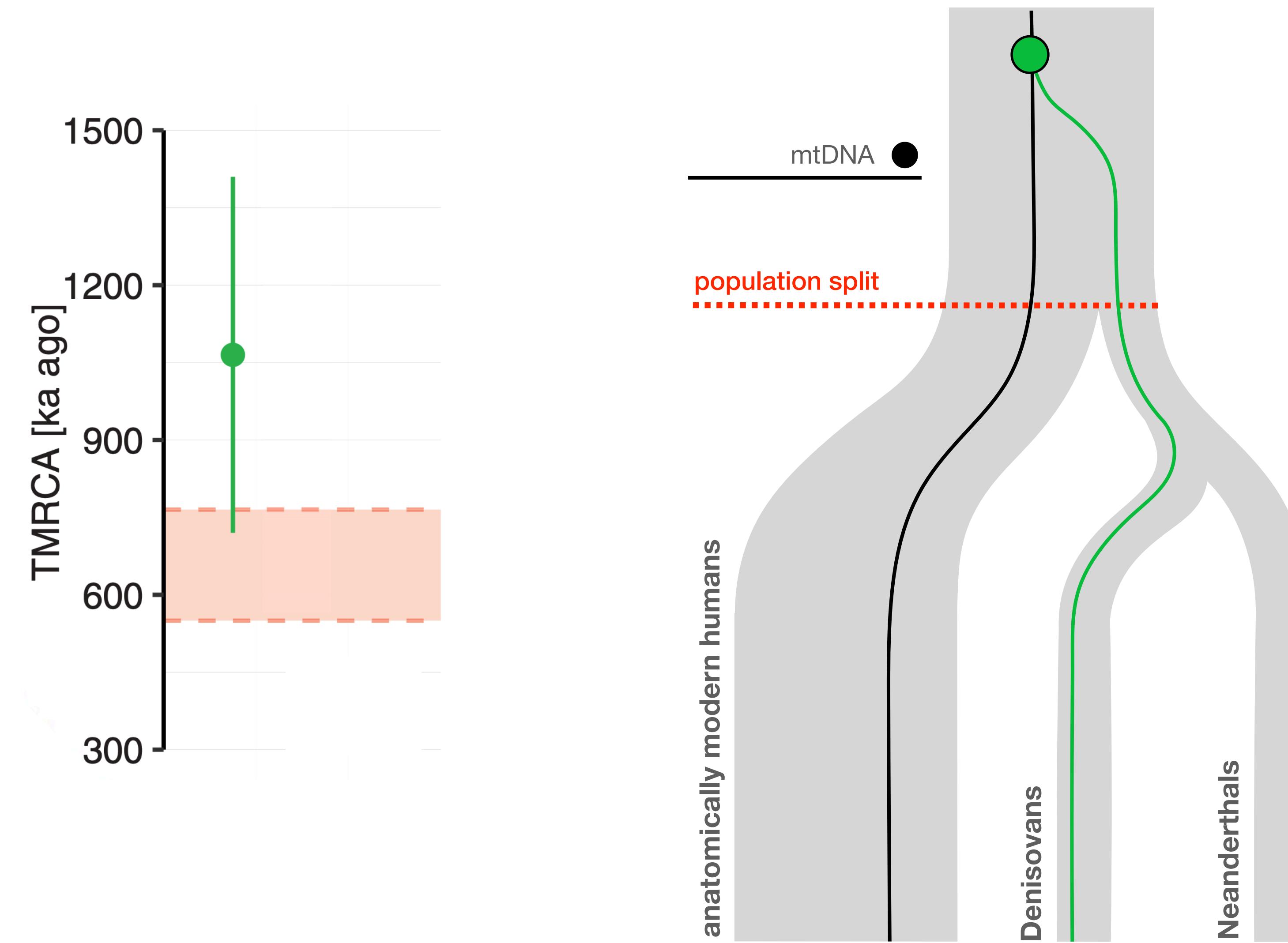


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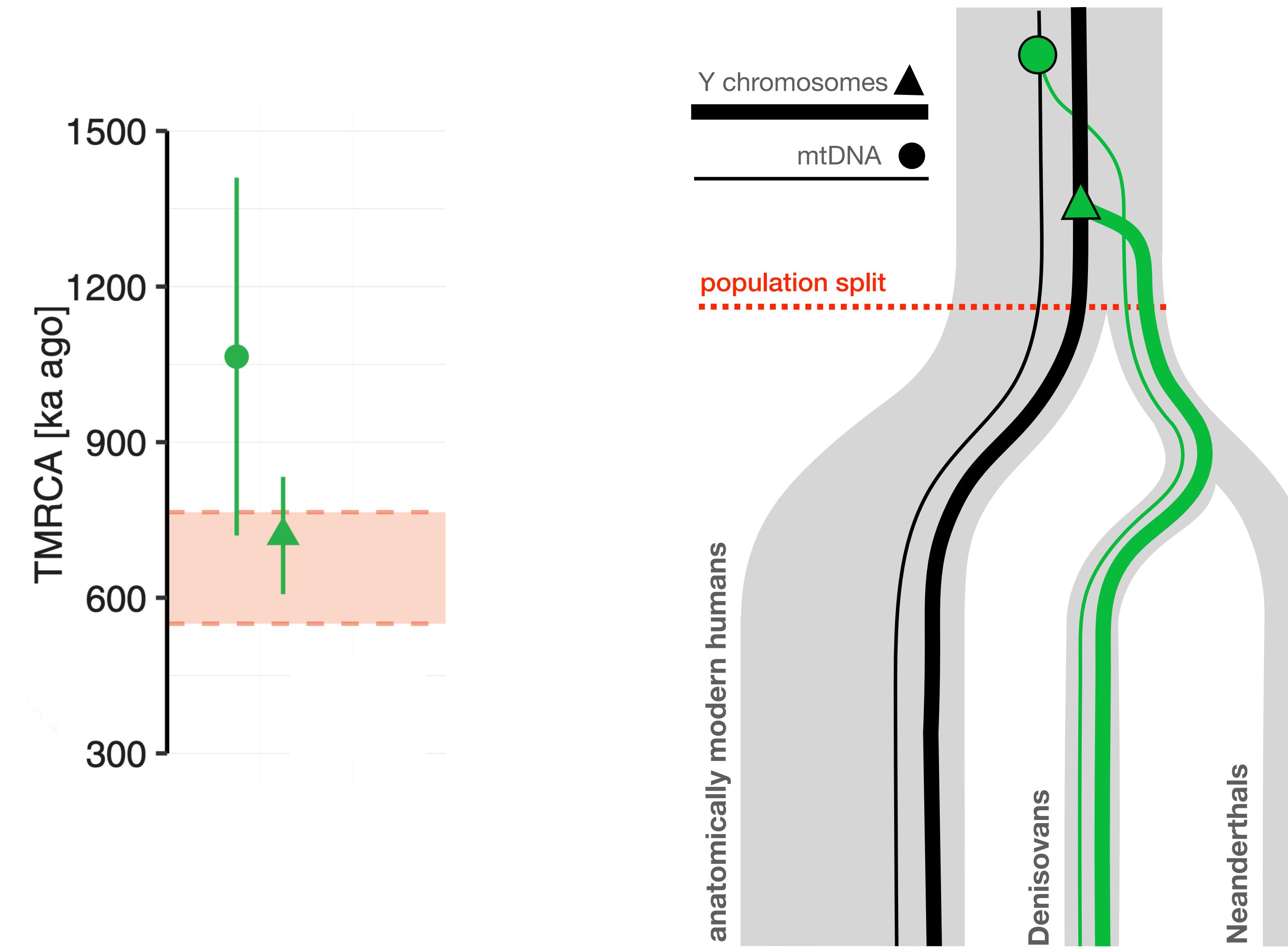
Population history on a nuclear DNA level



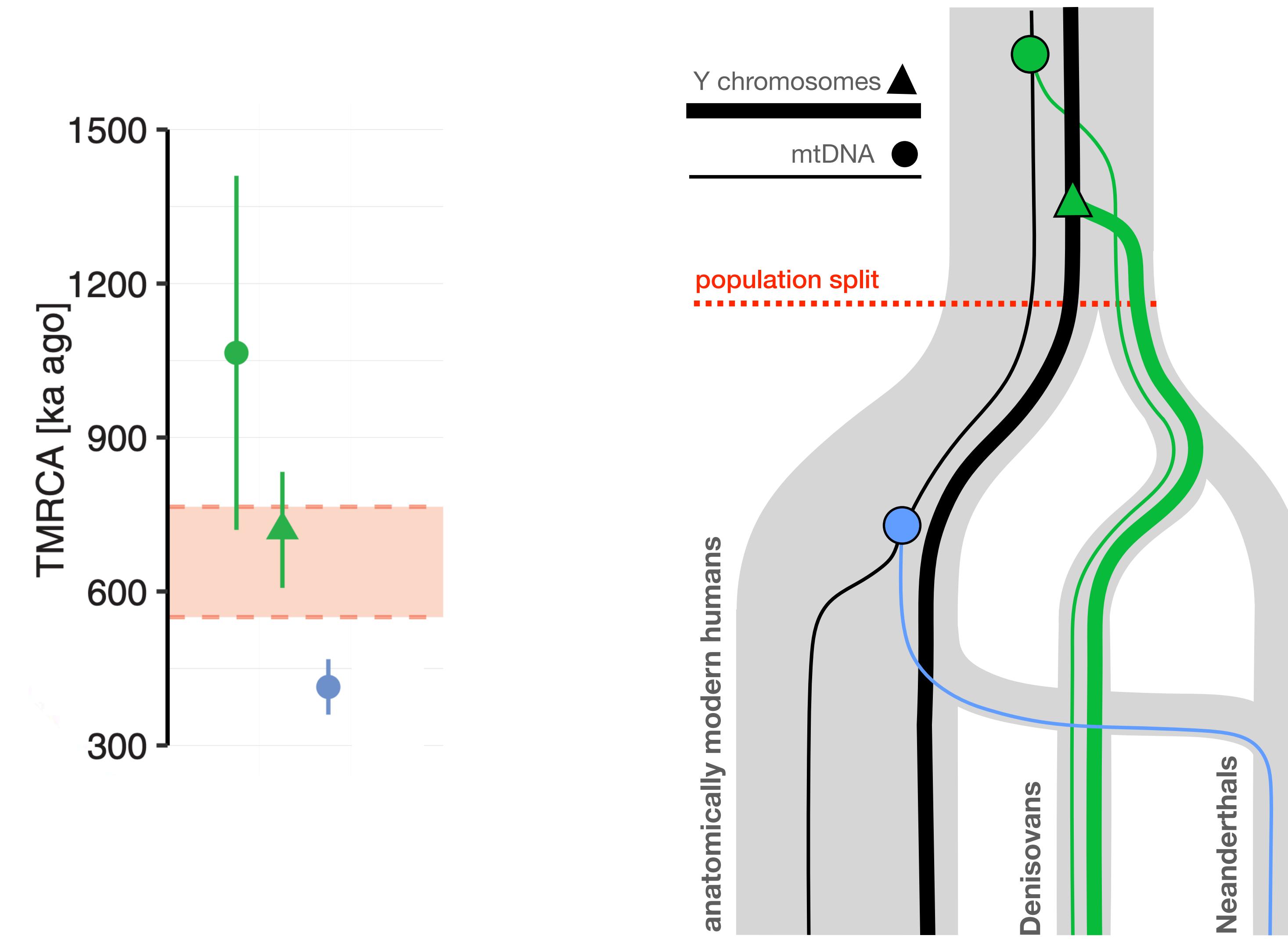
Denisovan mt and Y DNA follow known population history



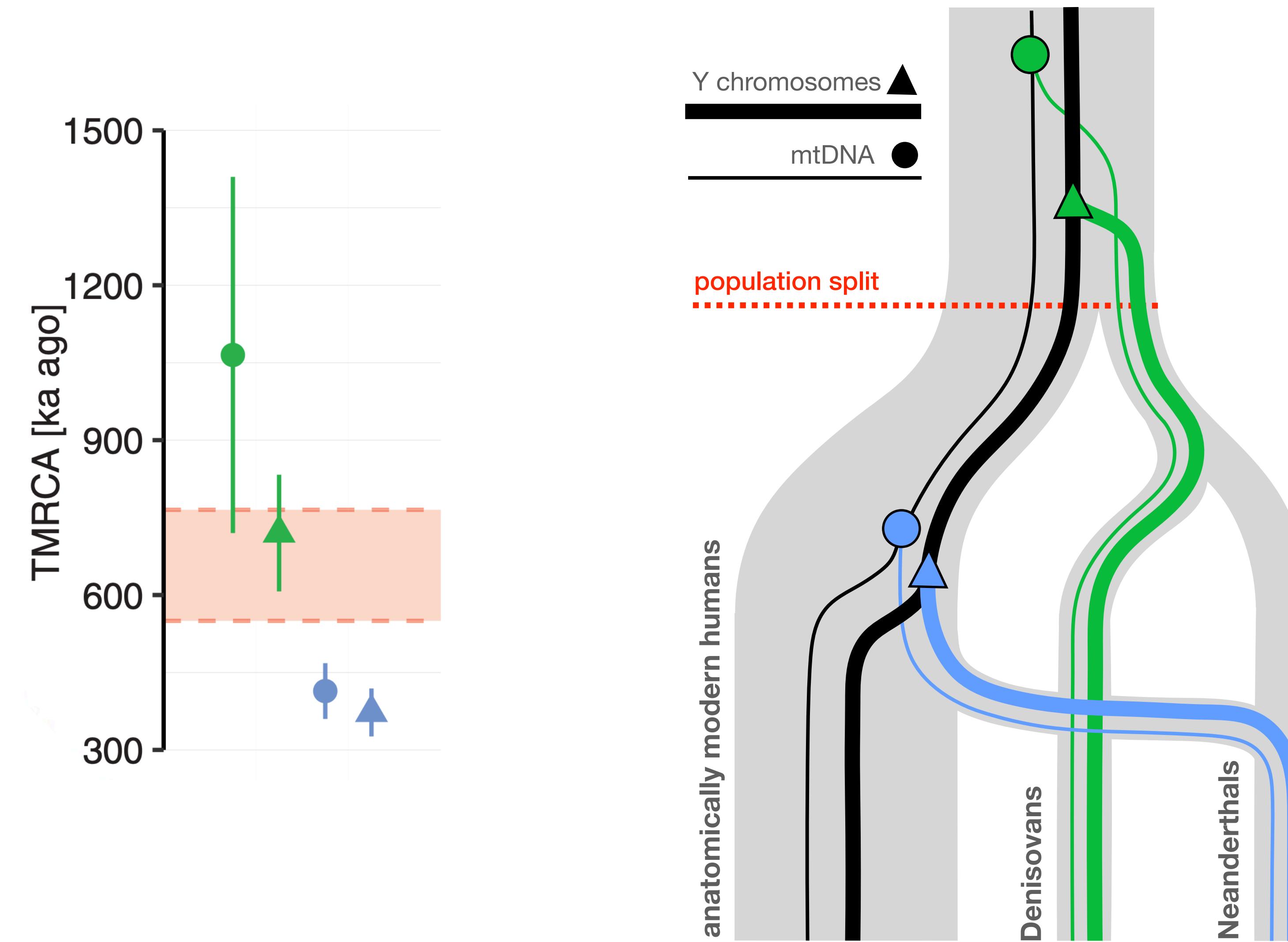
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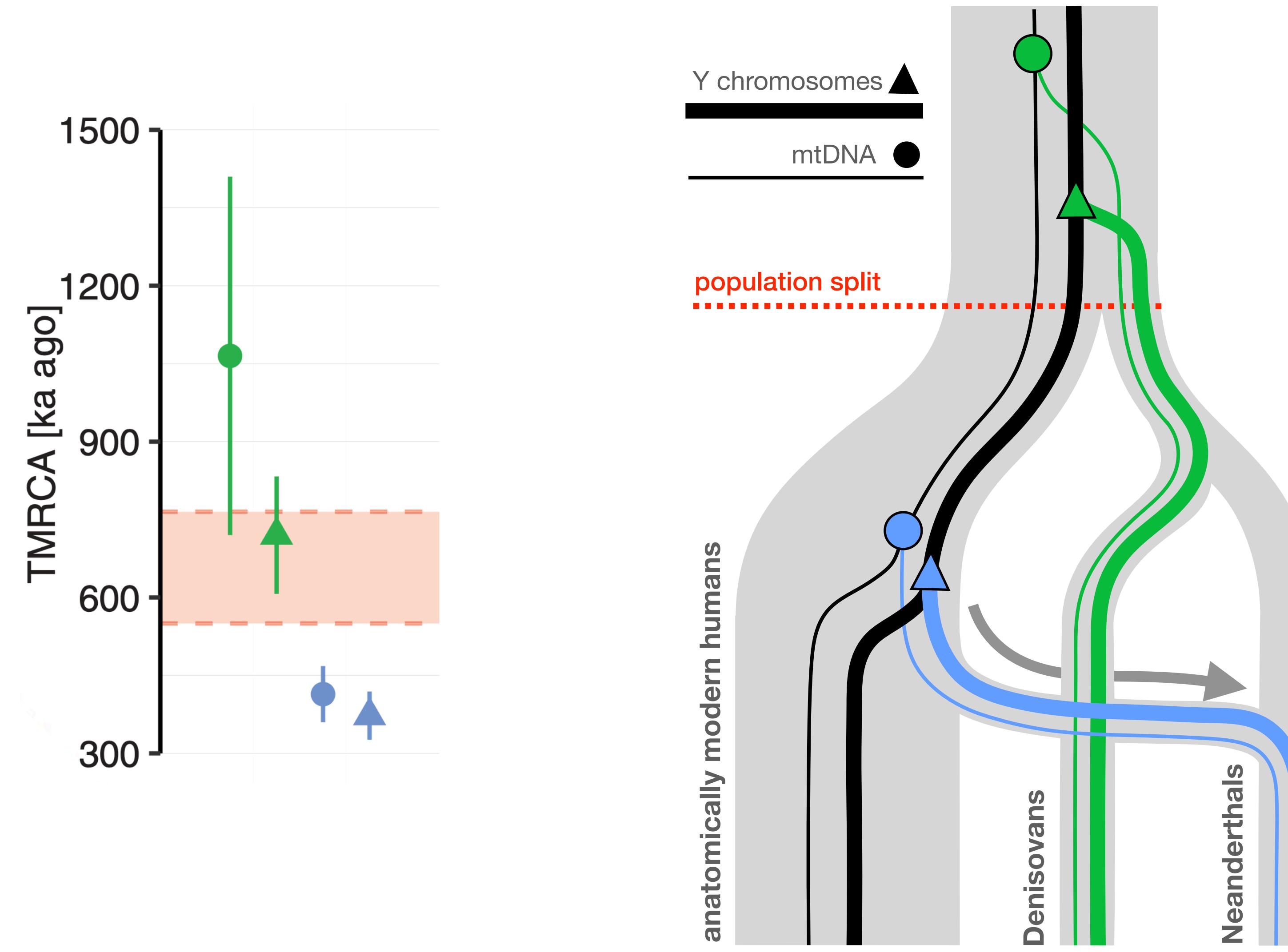
Neanderthal mitochondrial DNAs contradict this!



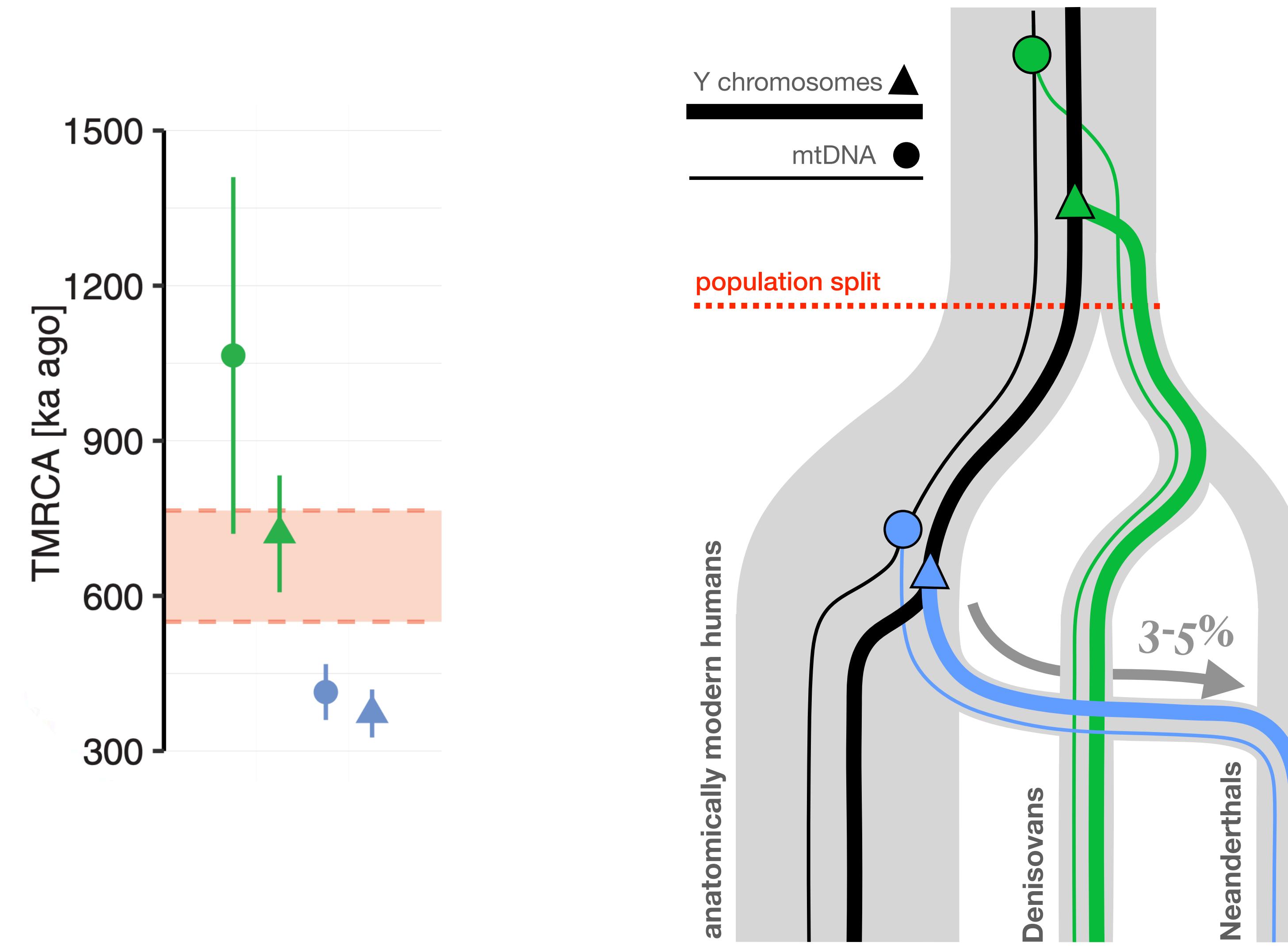
Neanderthal Y chromosomes also contradict this!



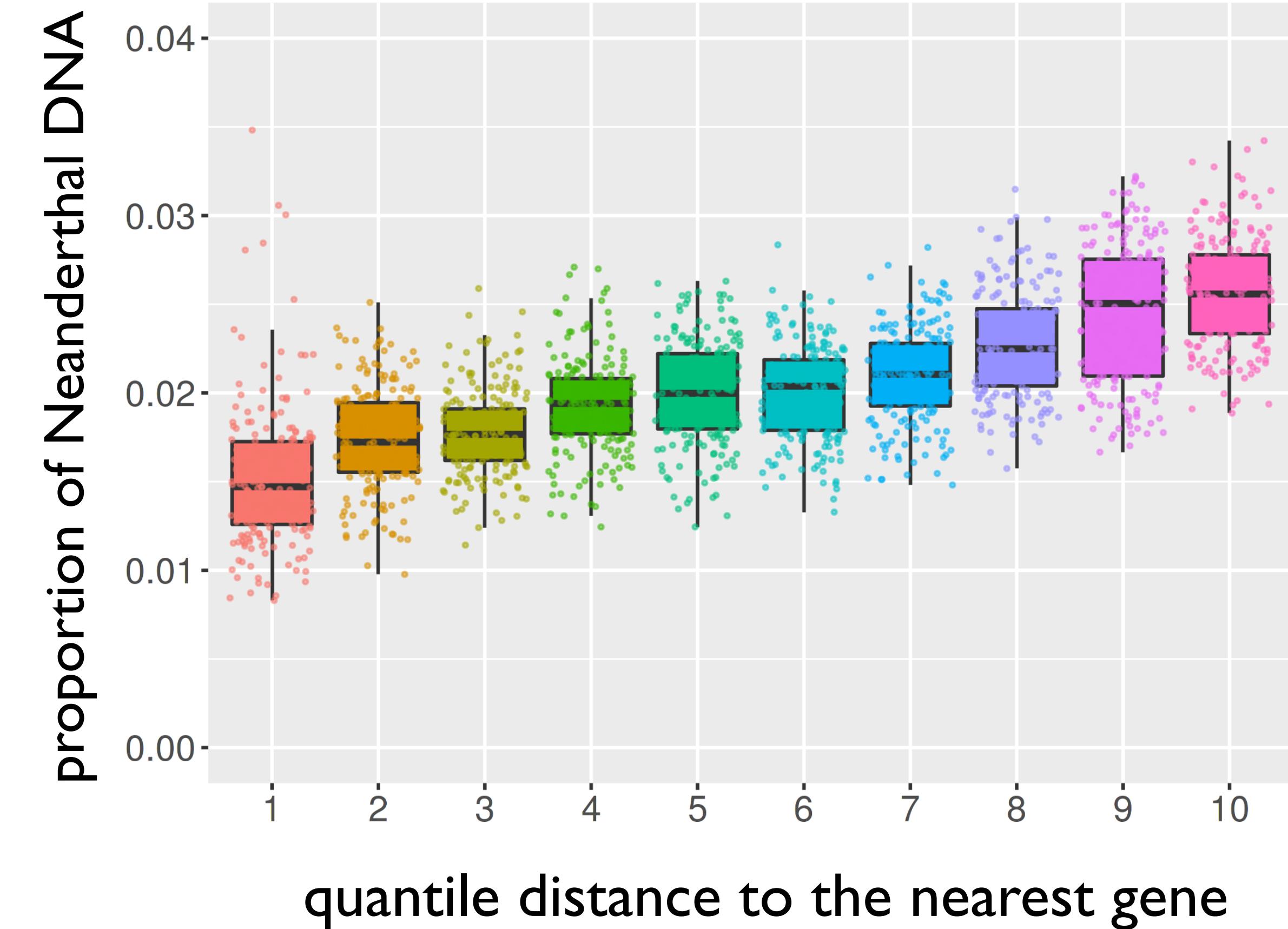
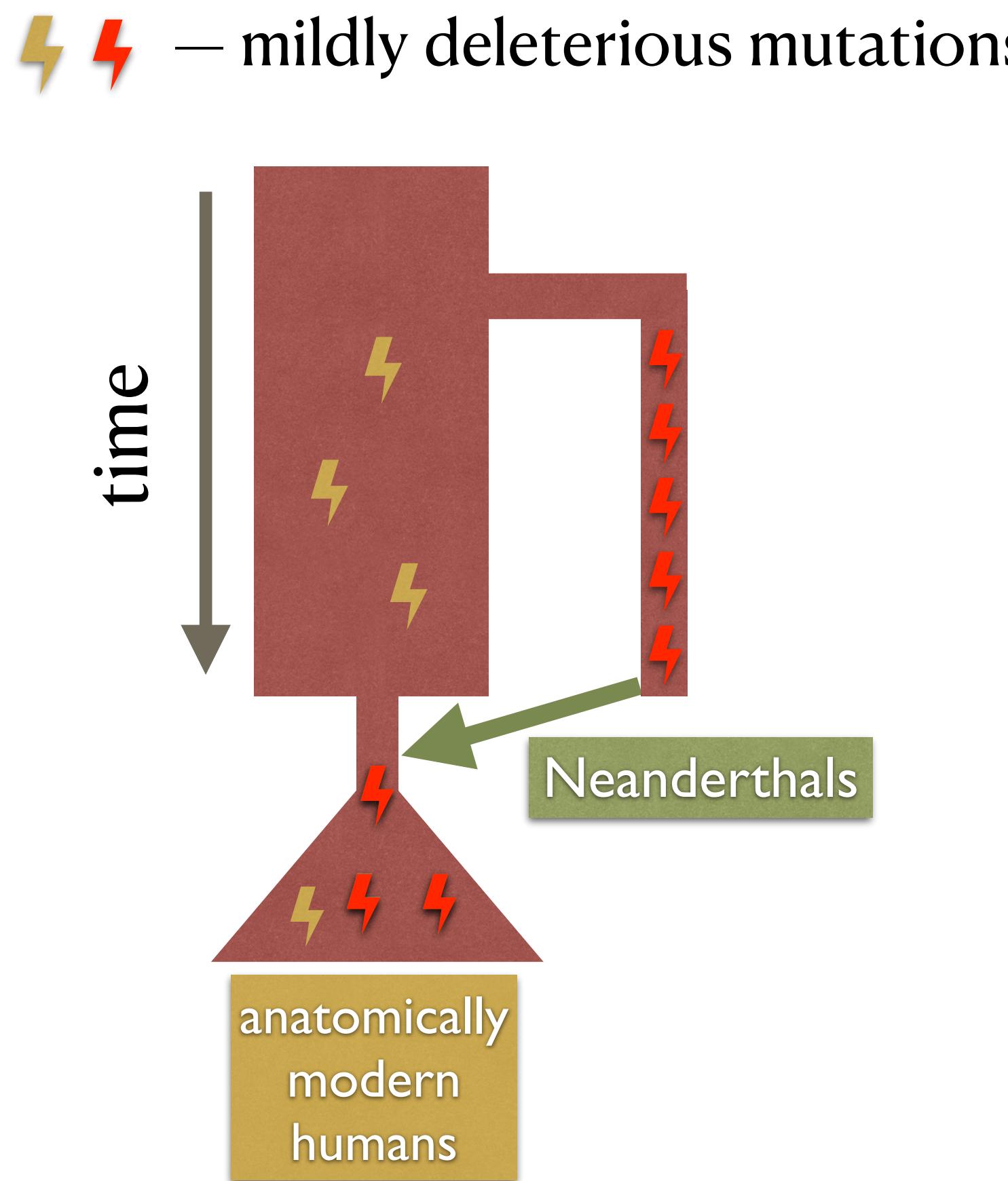
Original Neanderthal mtDNA/Y experienced replacement?



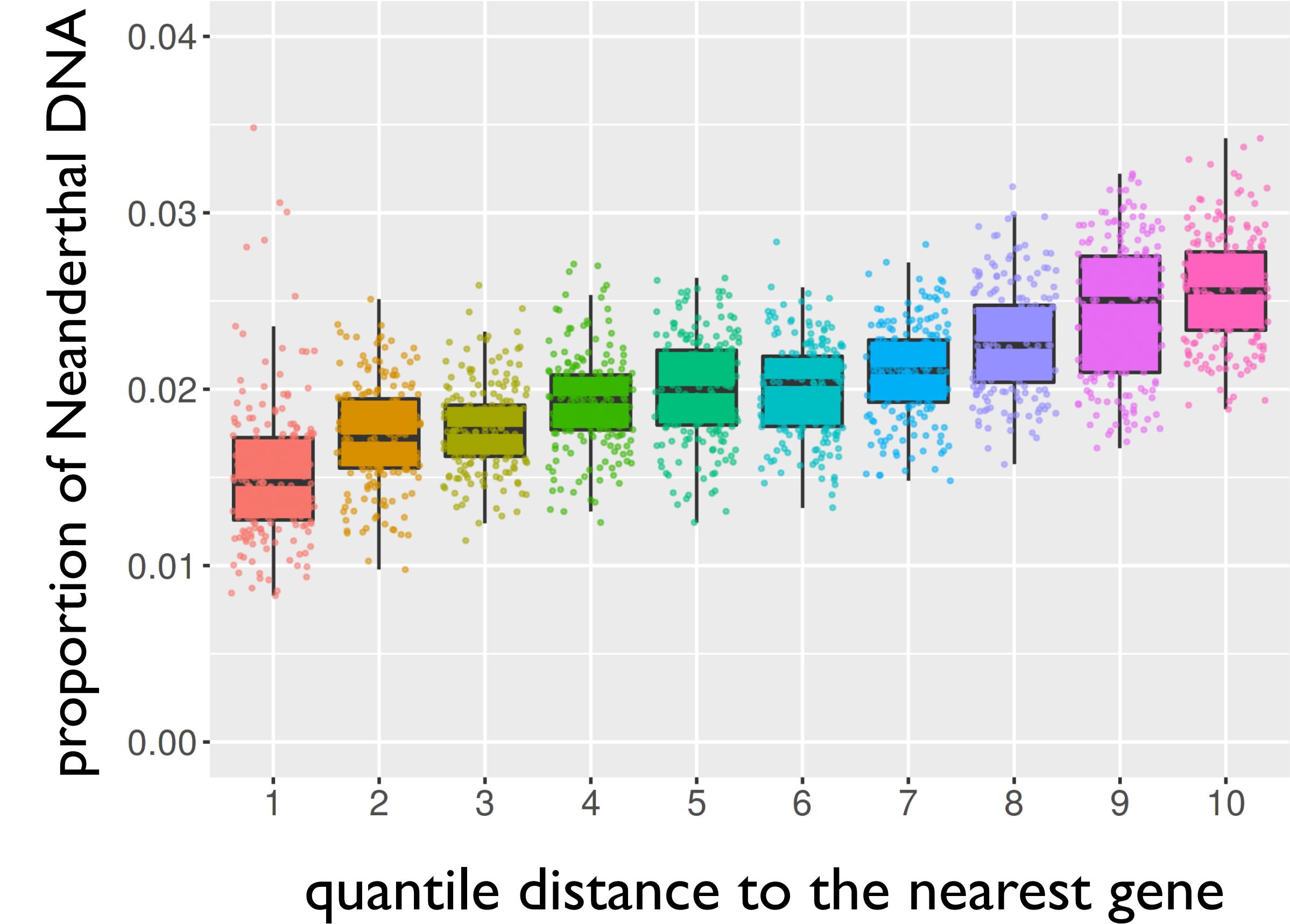
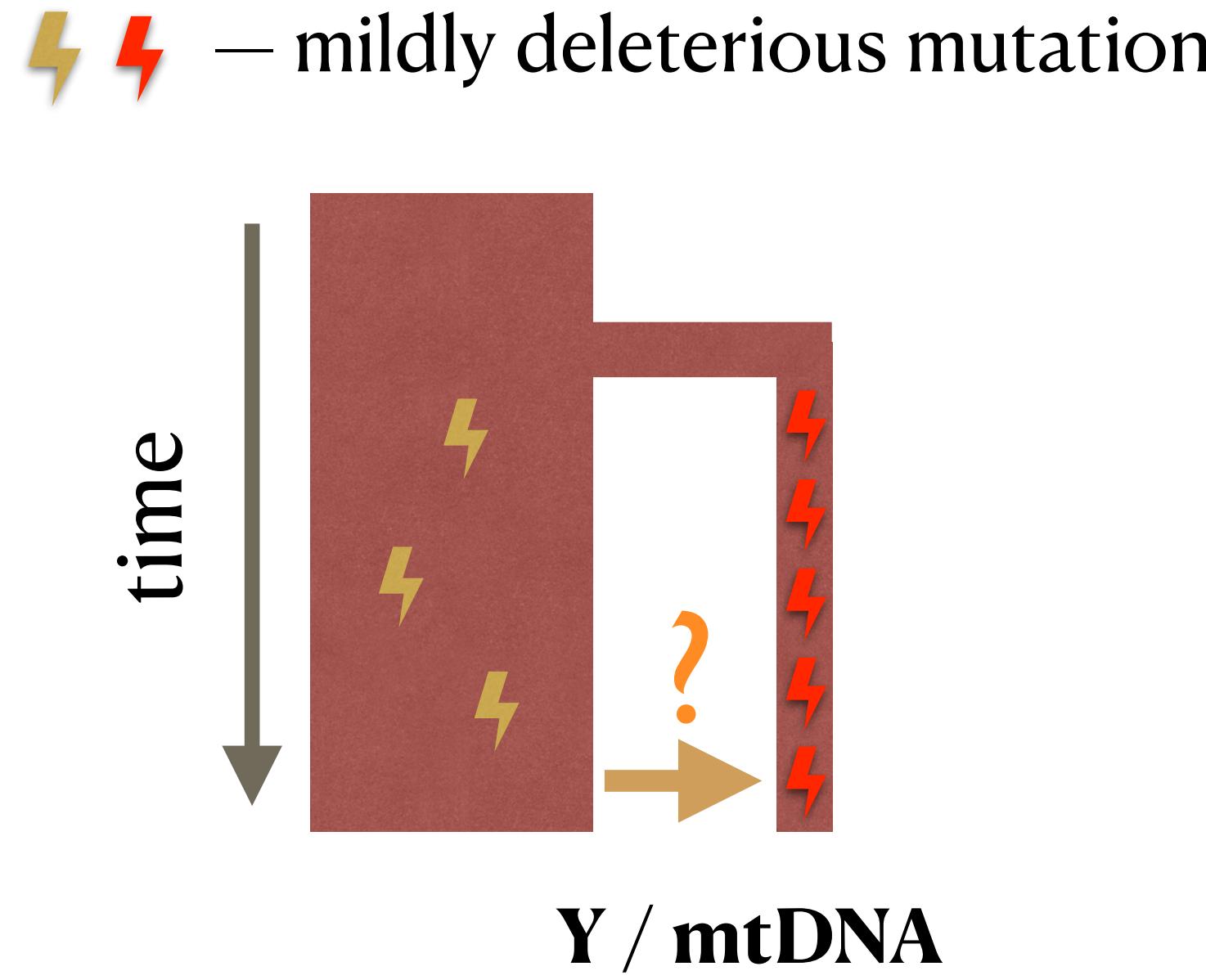
Original Neanderthal mtDNA/Y experienced replacement? (very unlikely under neutrality!)



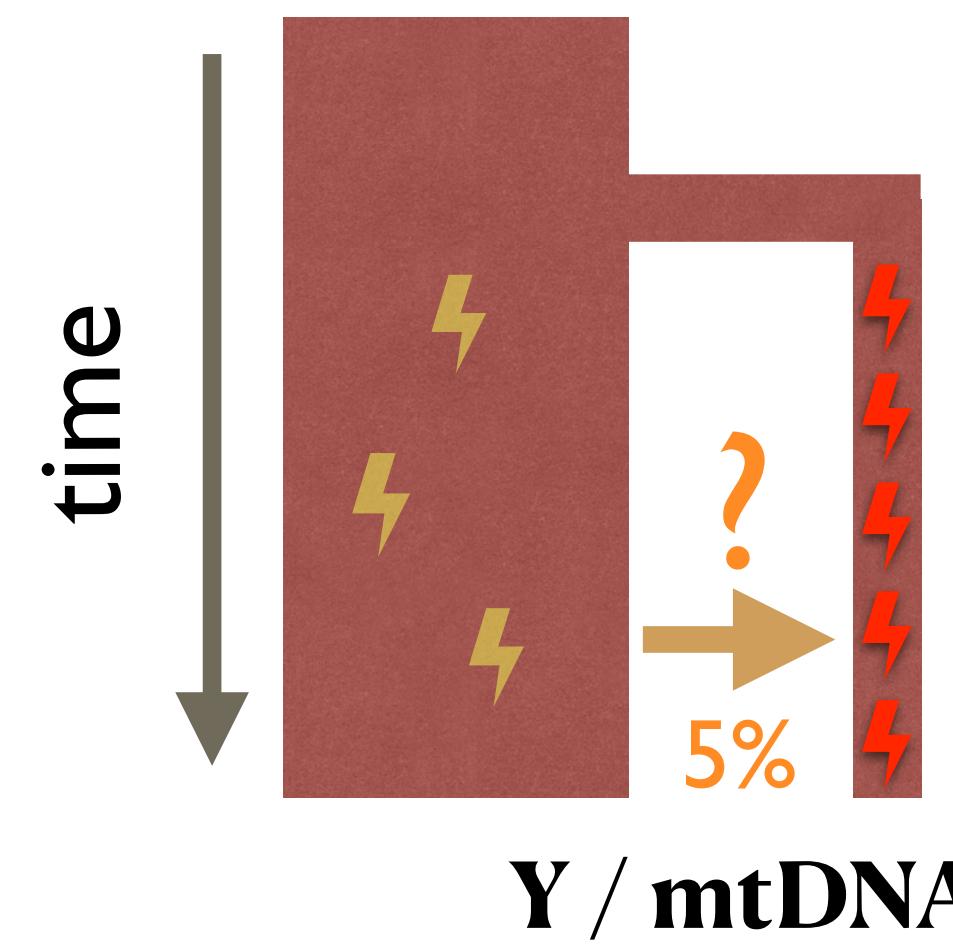
...but we know introgression was not neutral!



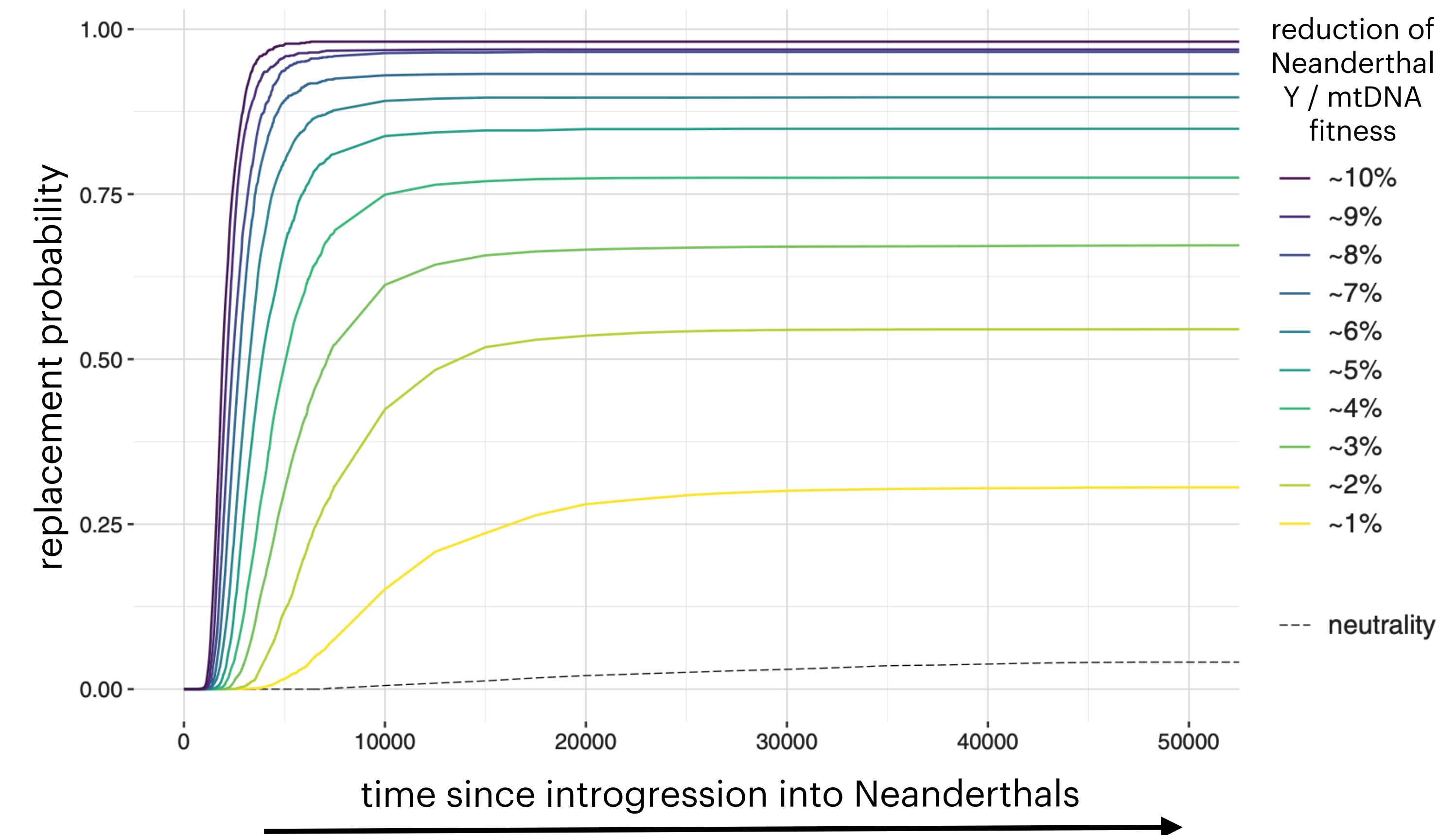
What is the expected scenario for Y / mtDNA introgression?



Natural selection in Neanderthals favors modern human Y / mtDNA



Simulation of 5% gene flow from modern humans



It wasn't all negative though

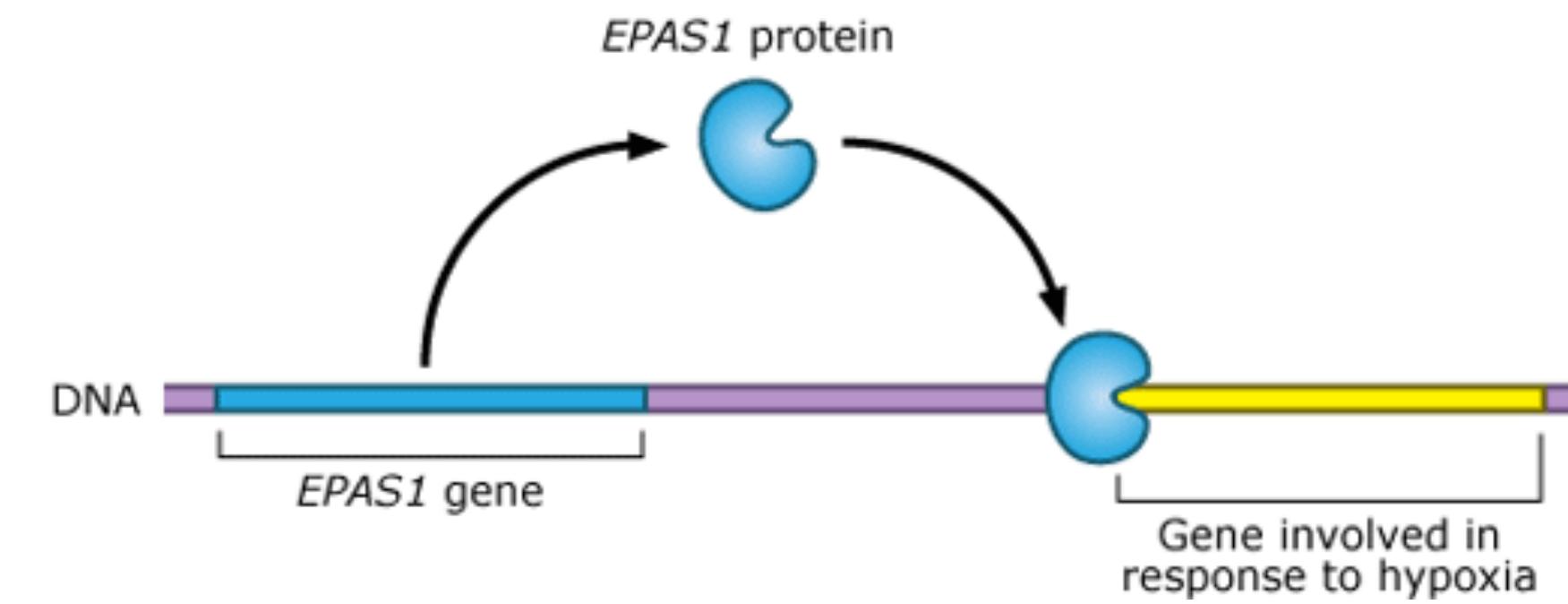
positive selection / adaptive introgression

High altitude adaptation in Tibetans

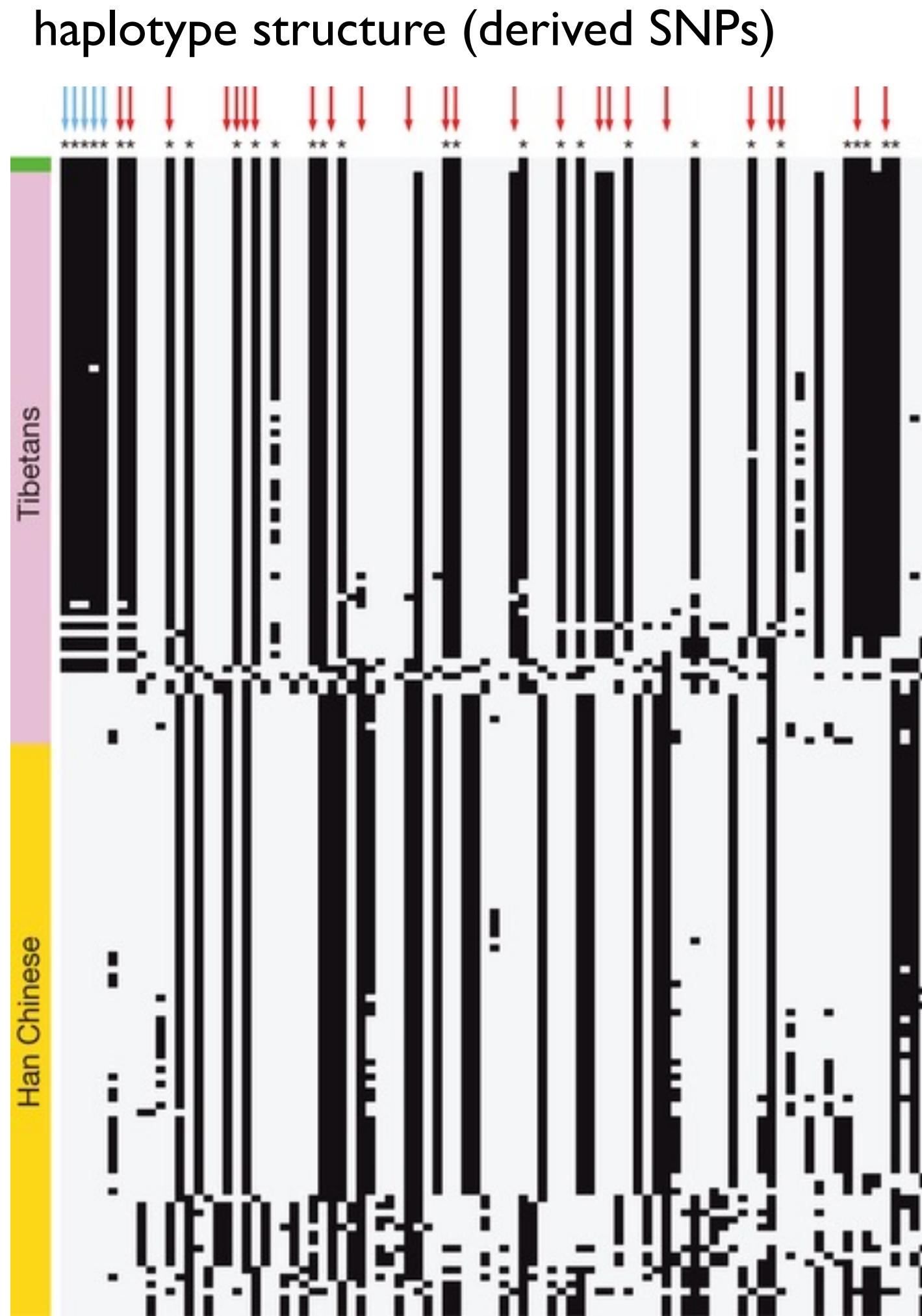
haplotype structure (derived SNPs)



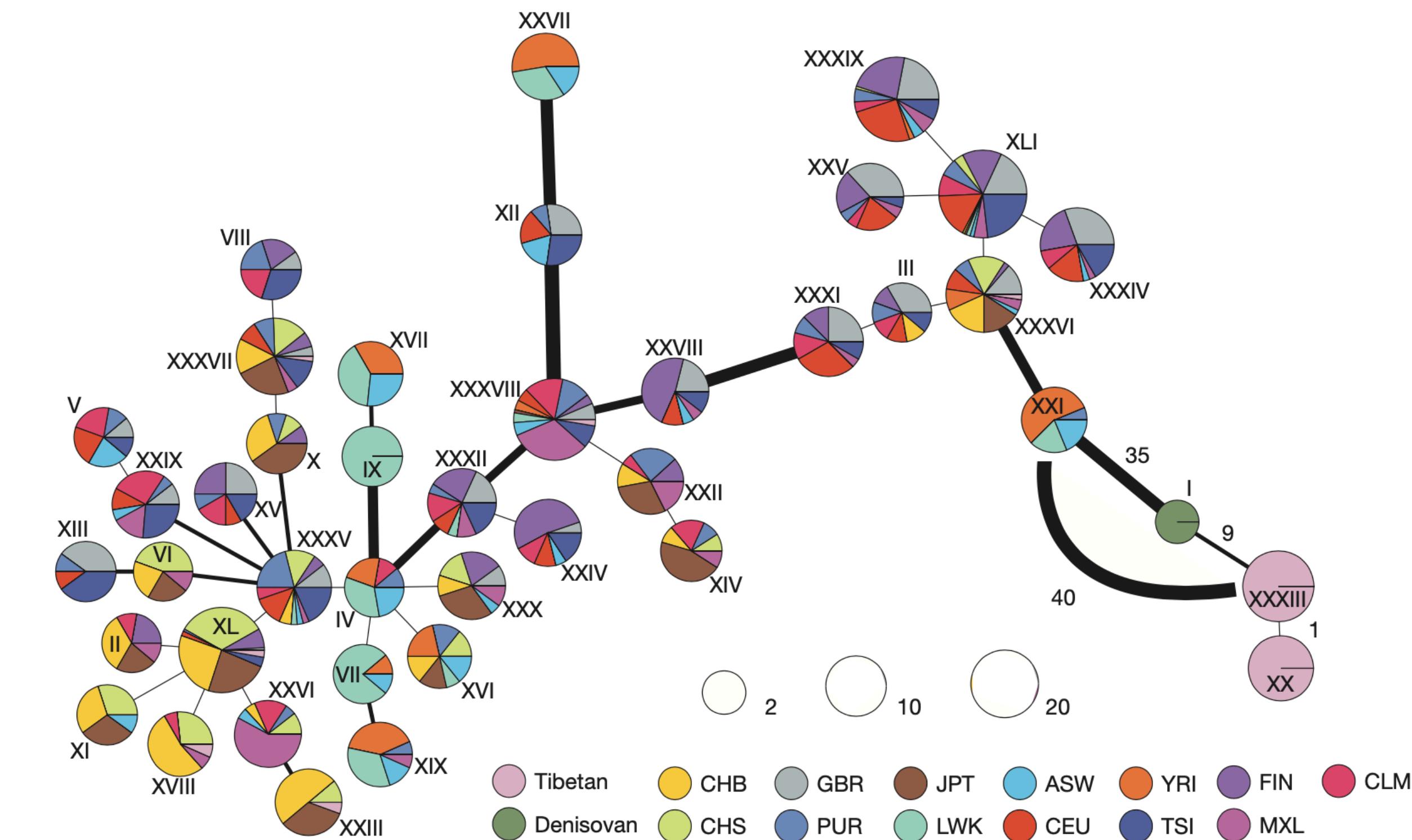
Schematic of *EPAS1* function



High altitude adaptation via Denisovan introgression!



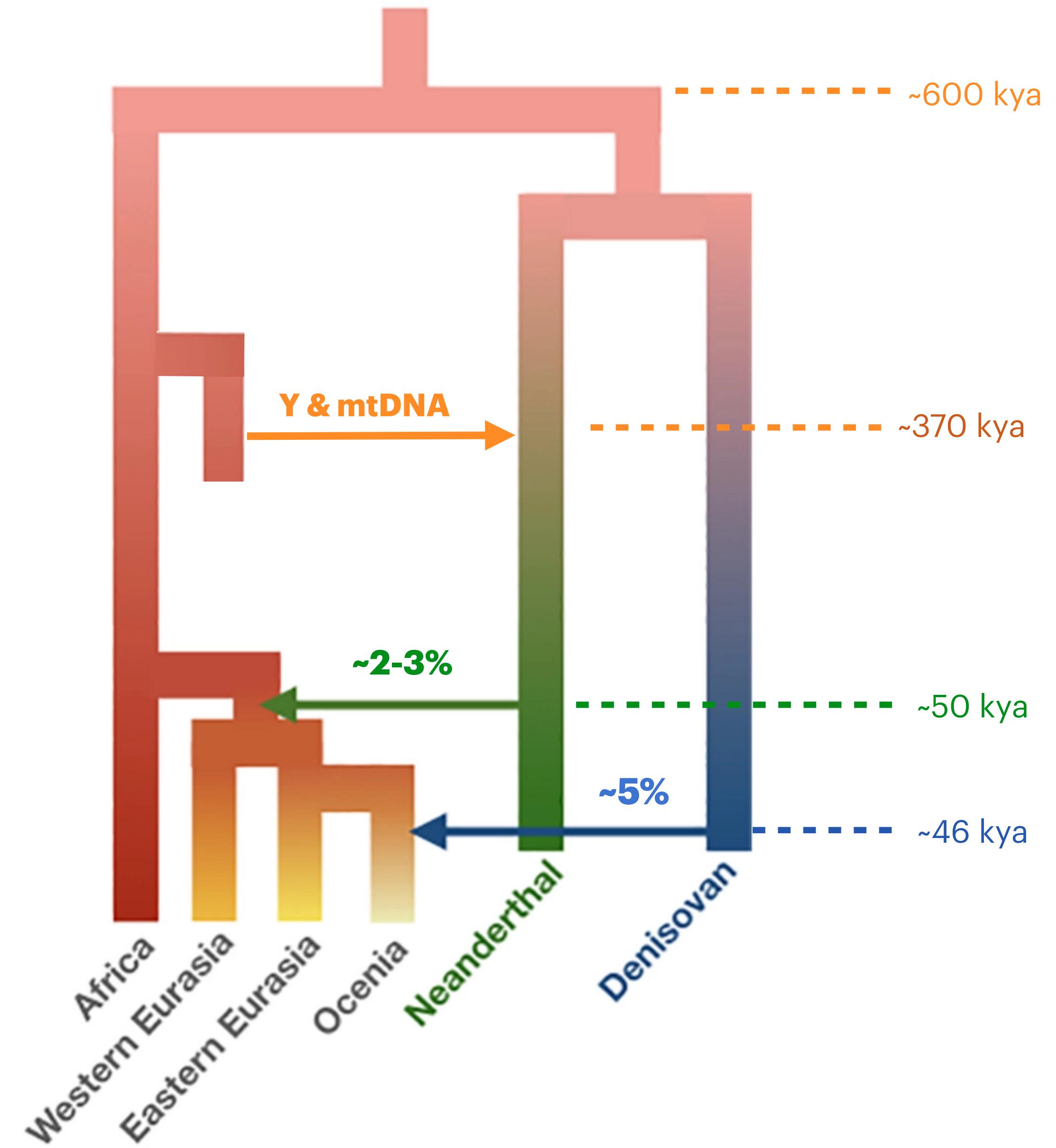
haplotype network based on pairwise-differences



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.



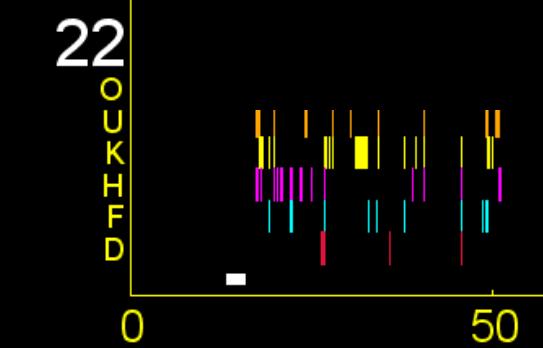
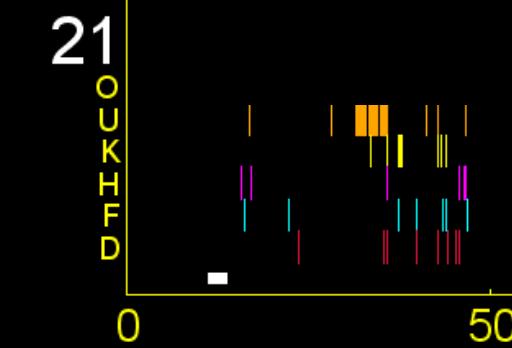
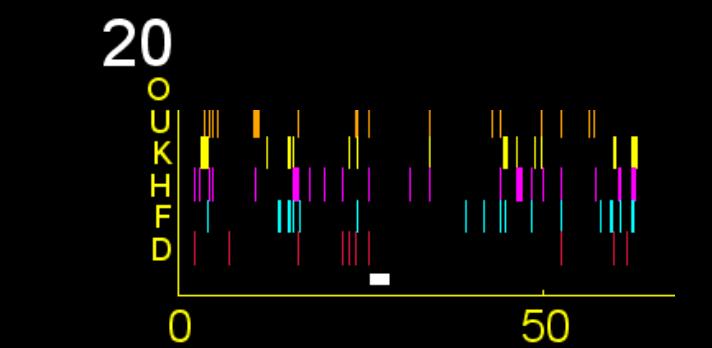
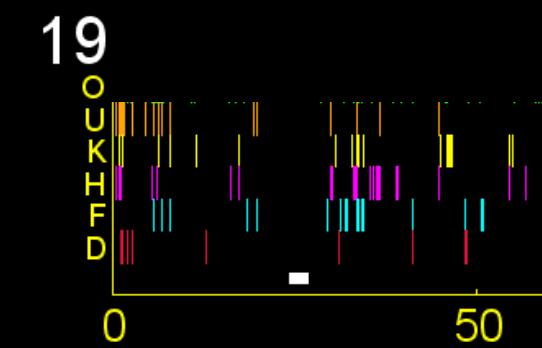
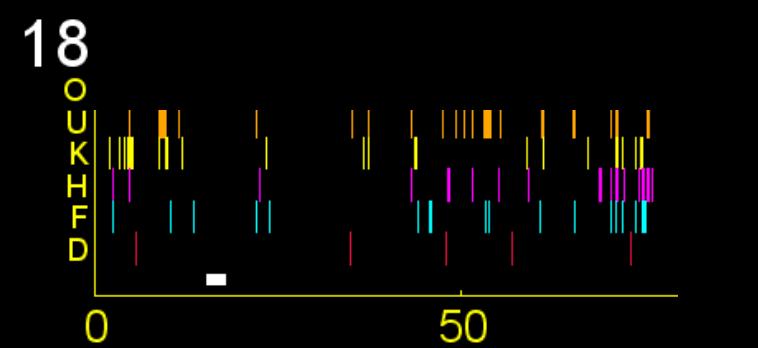
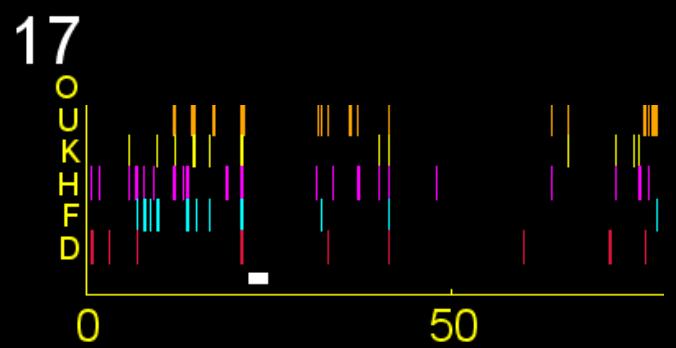
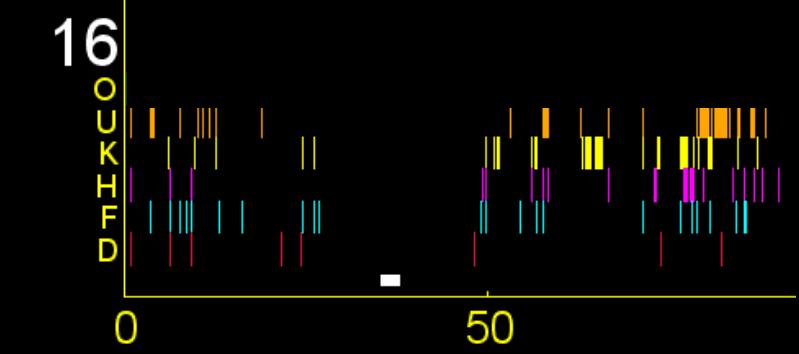
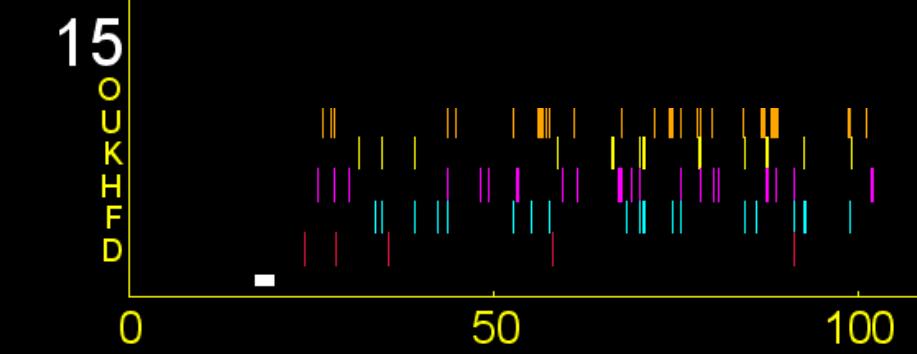
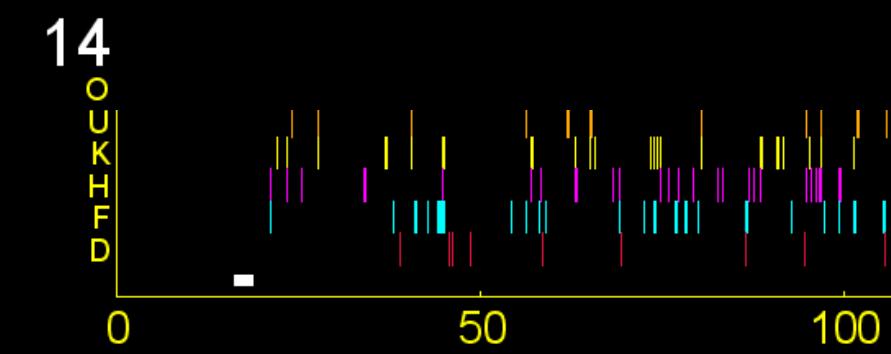
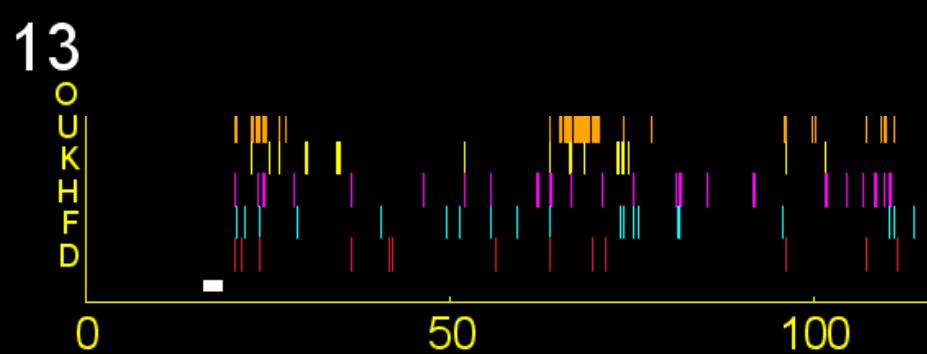
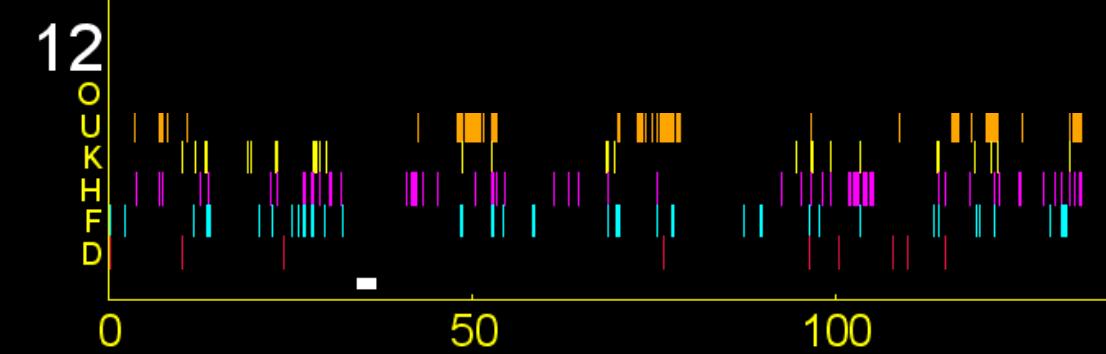
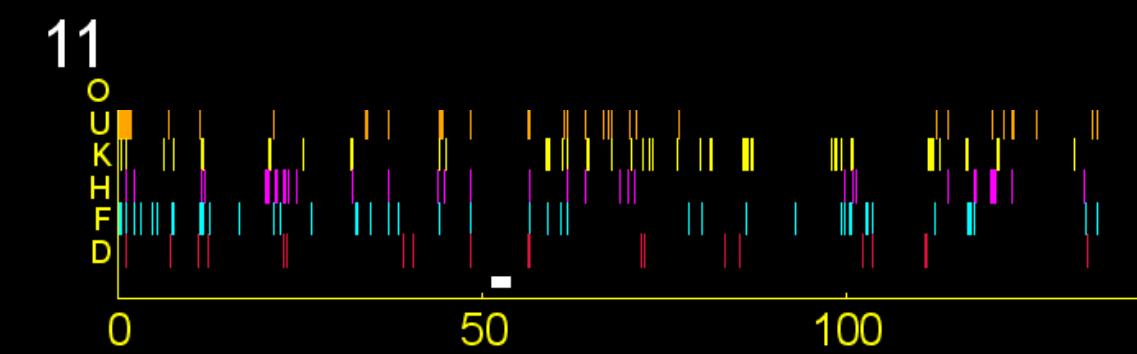
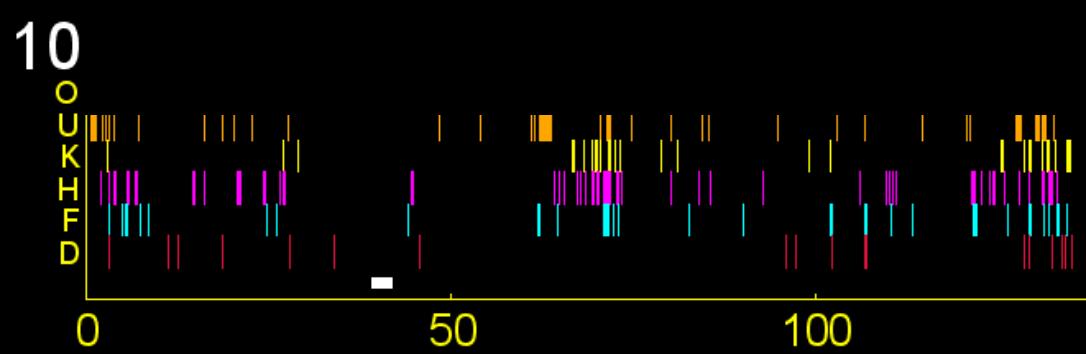
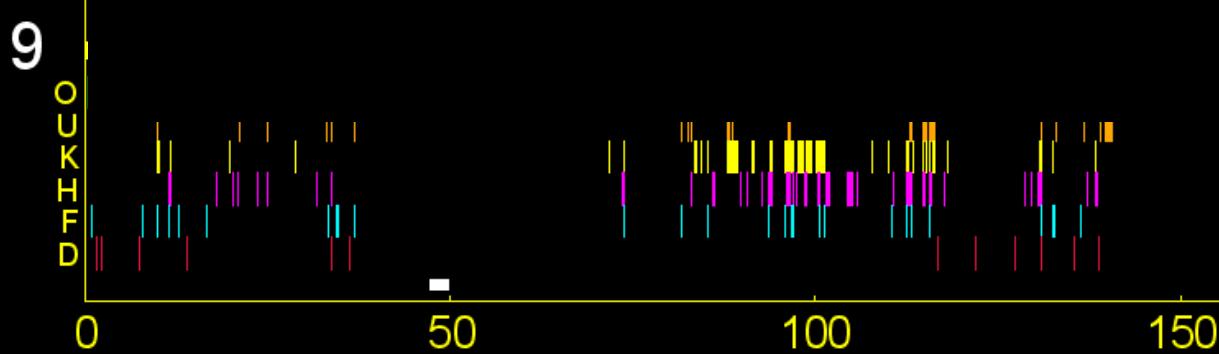
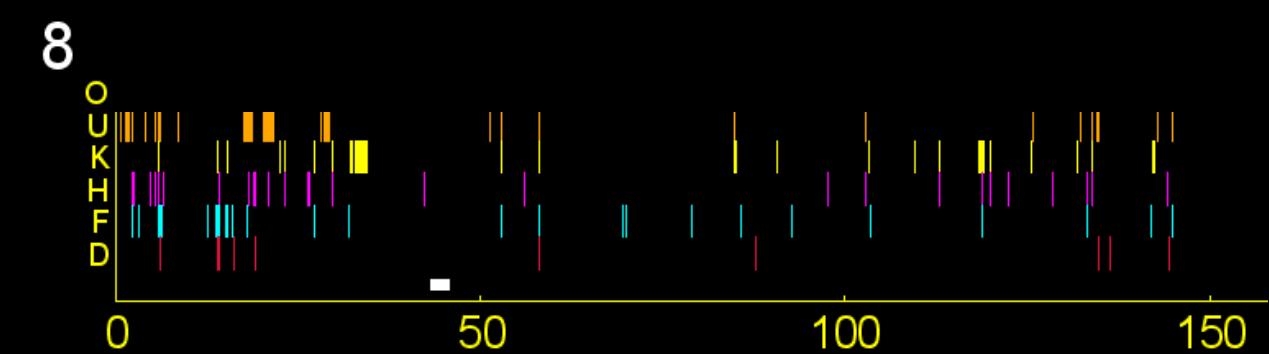
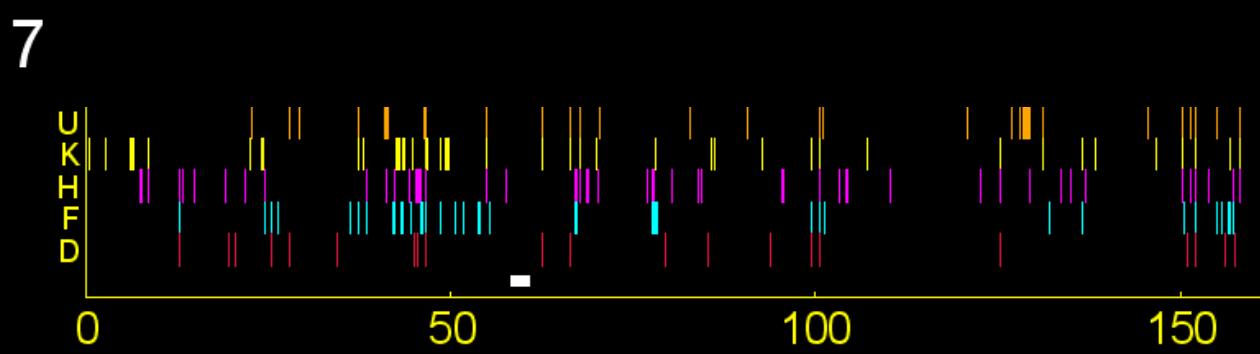
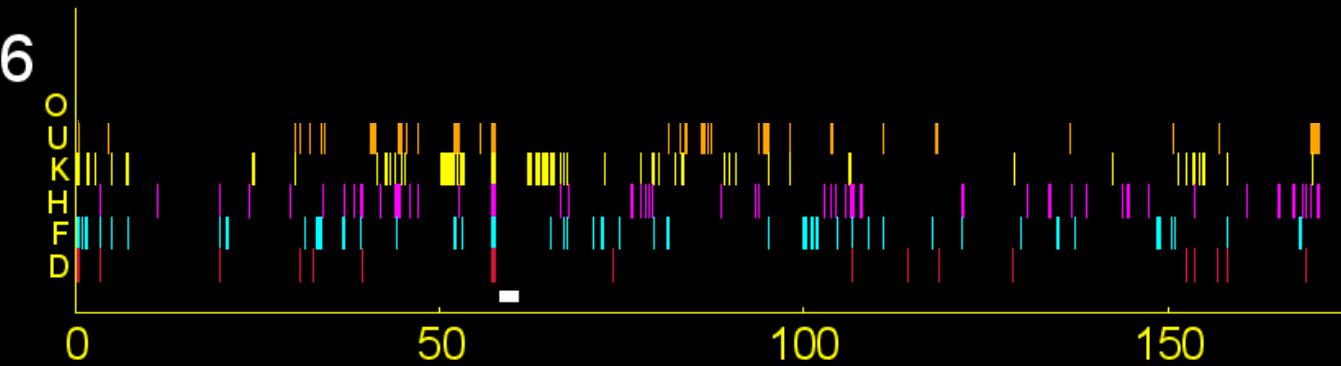
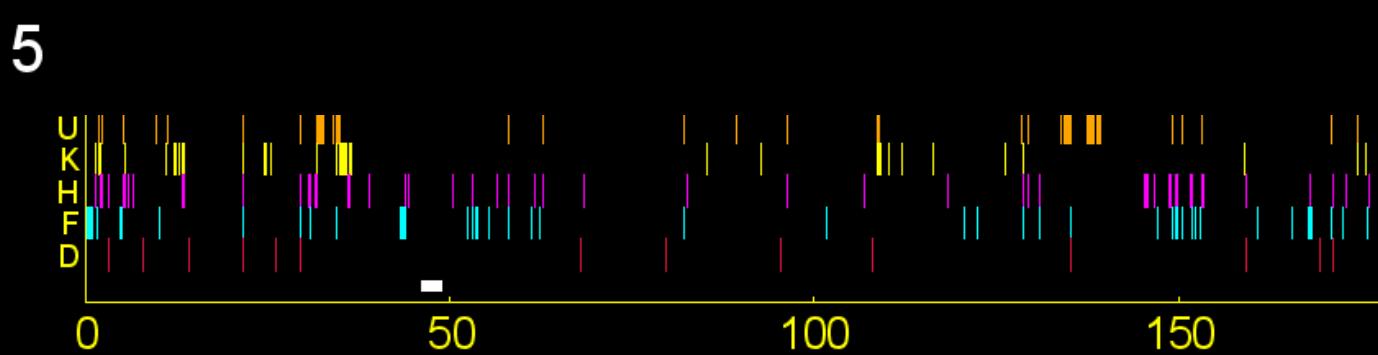
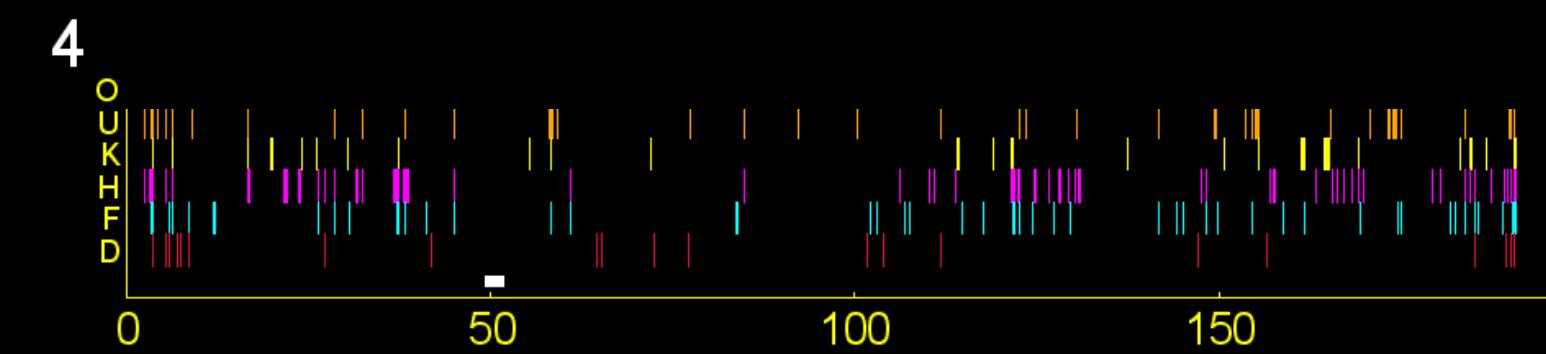
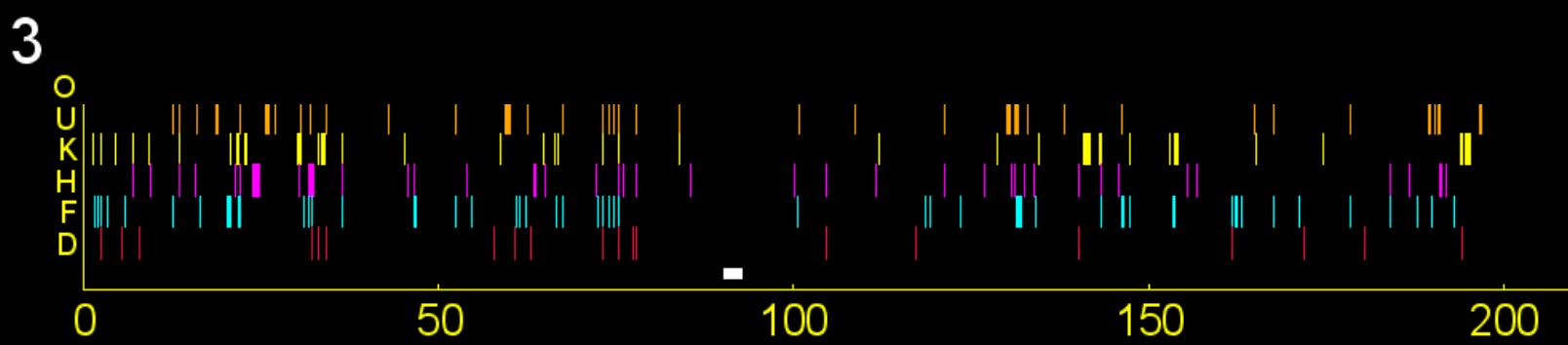
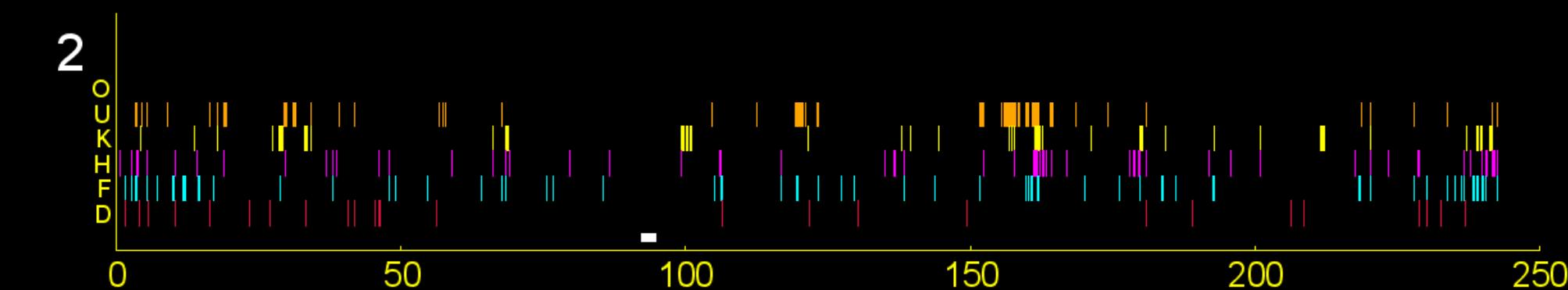
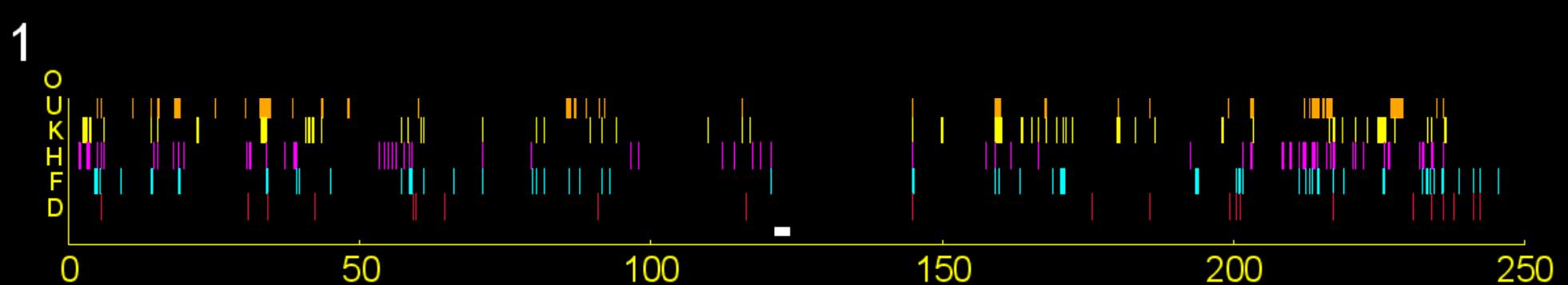


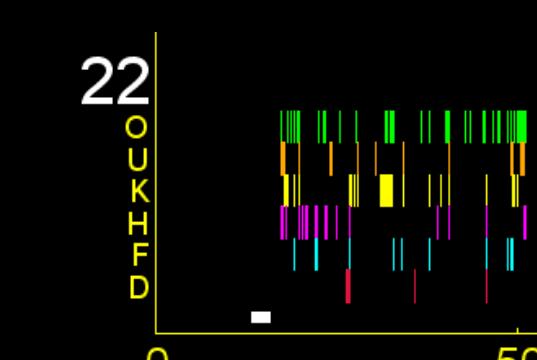
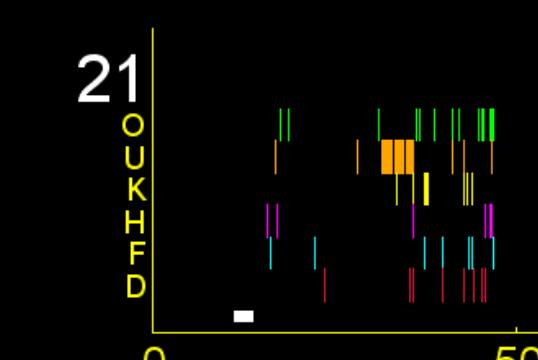
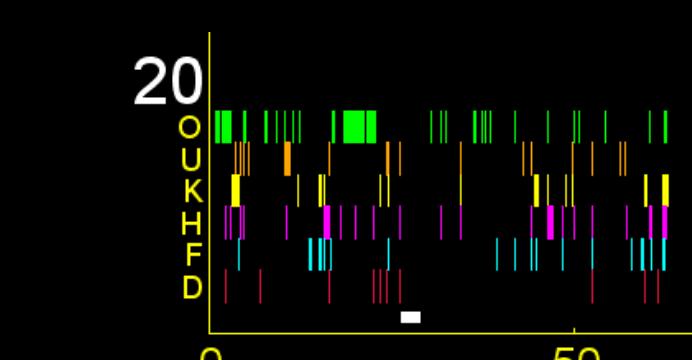
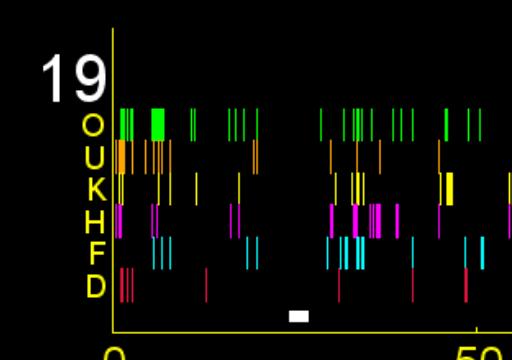
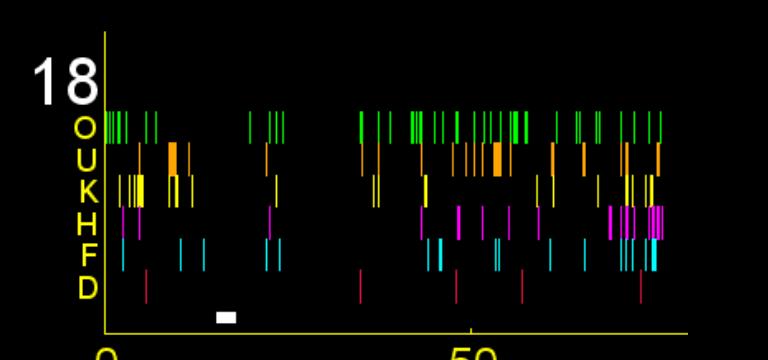
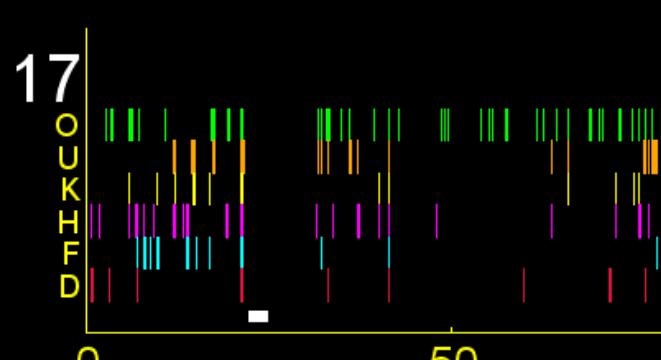
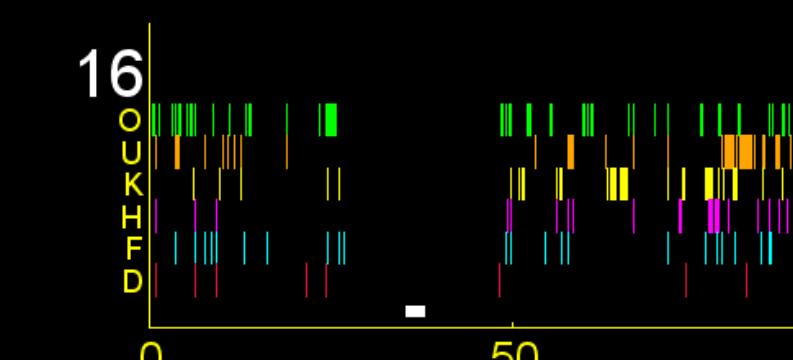
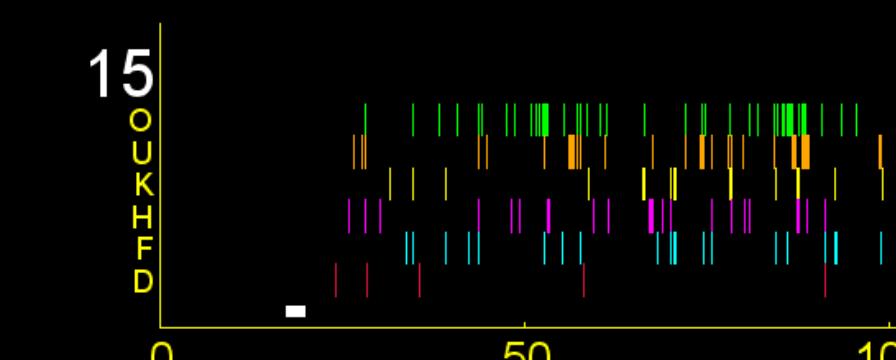
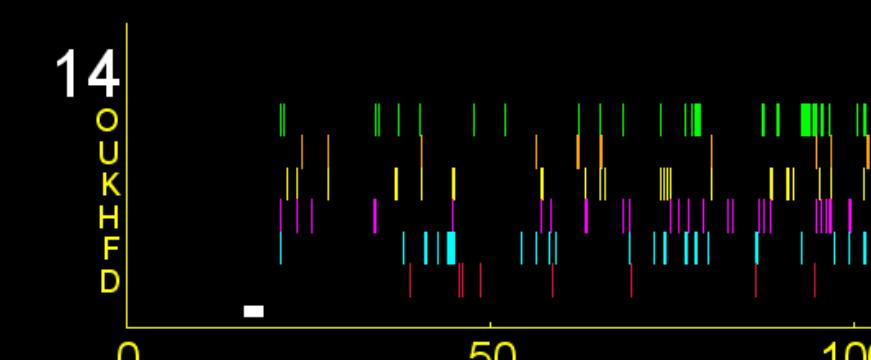
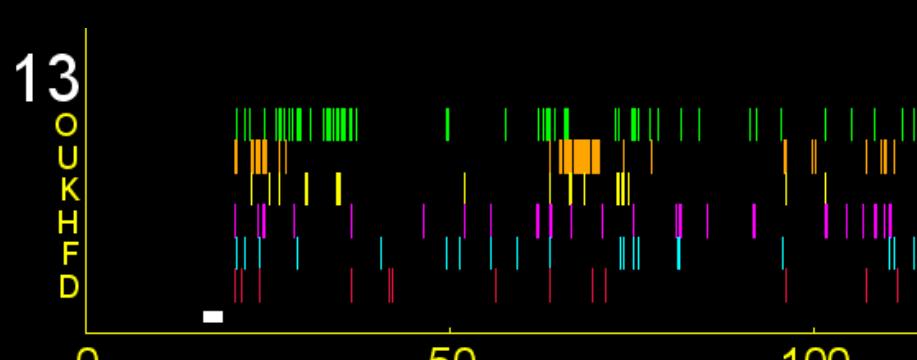
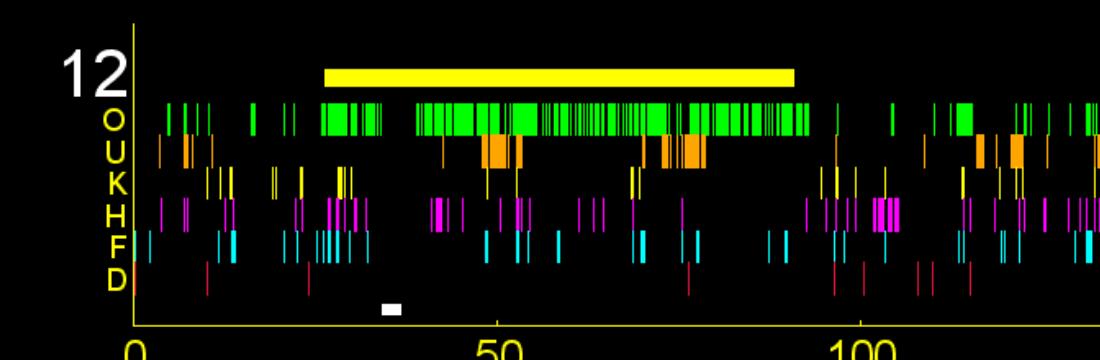
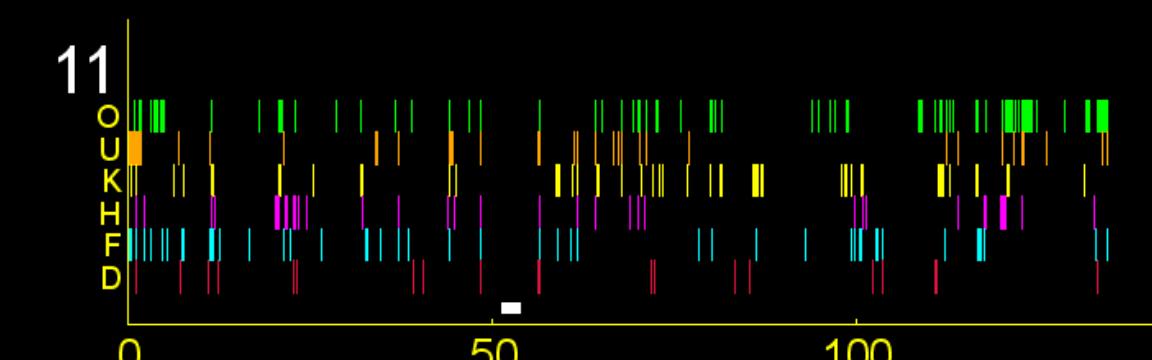
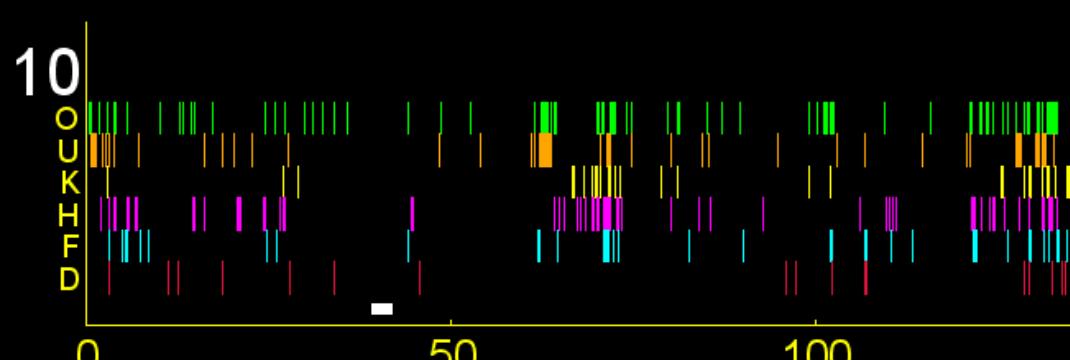
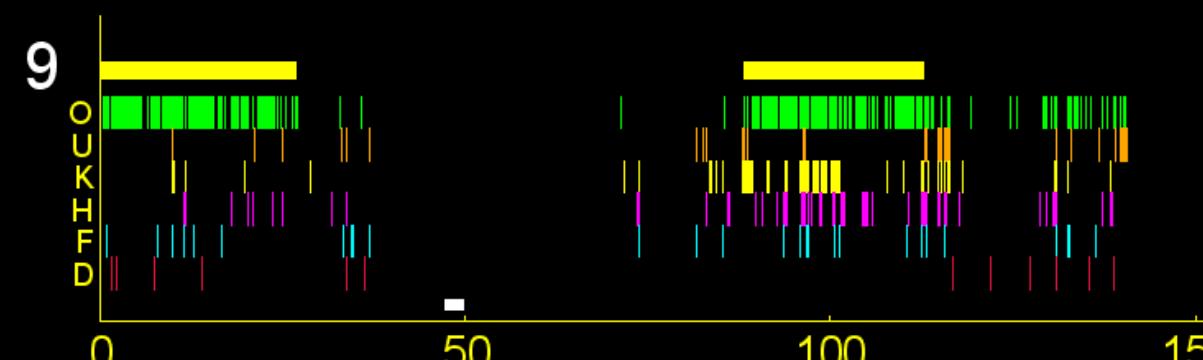
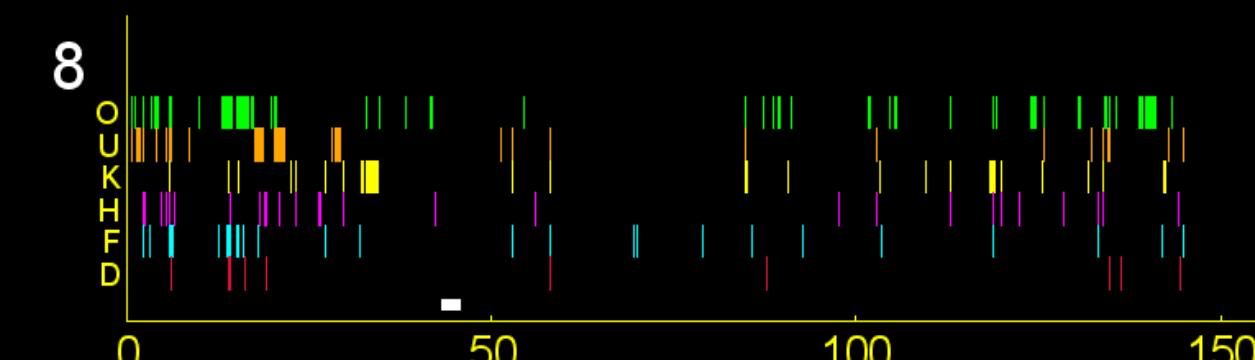
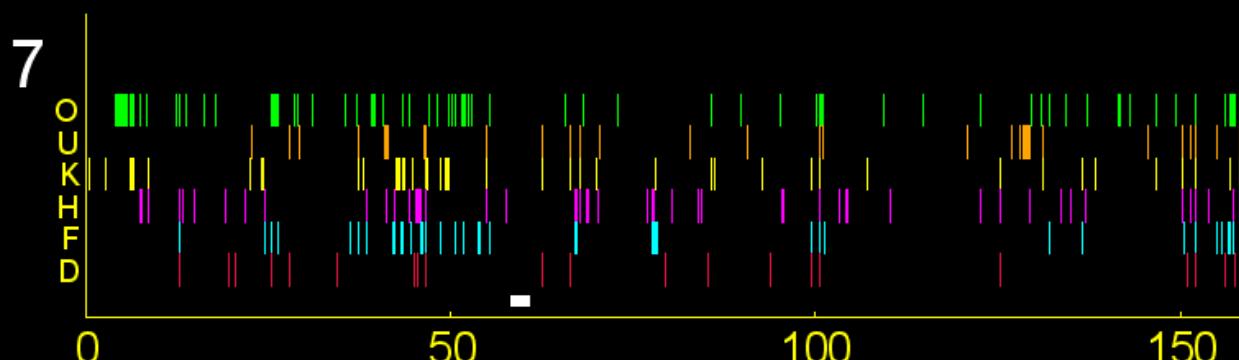
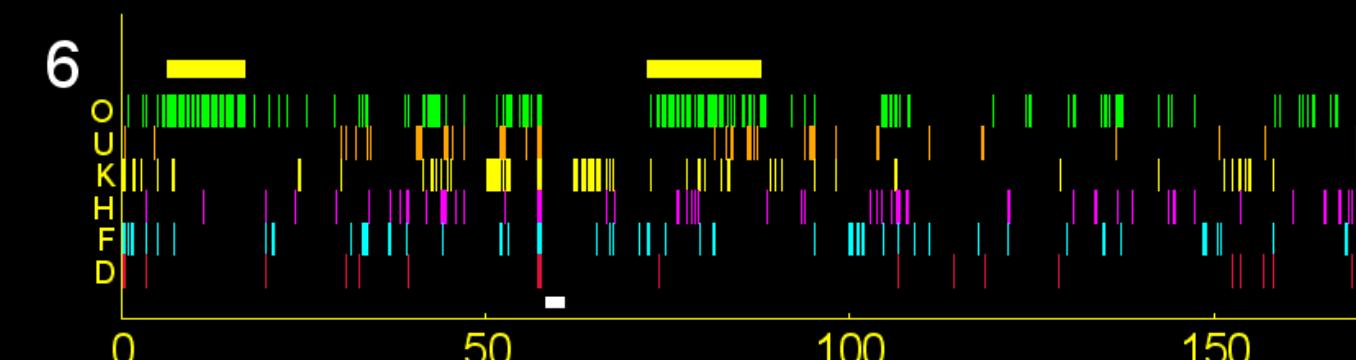
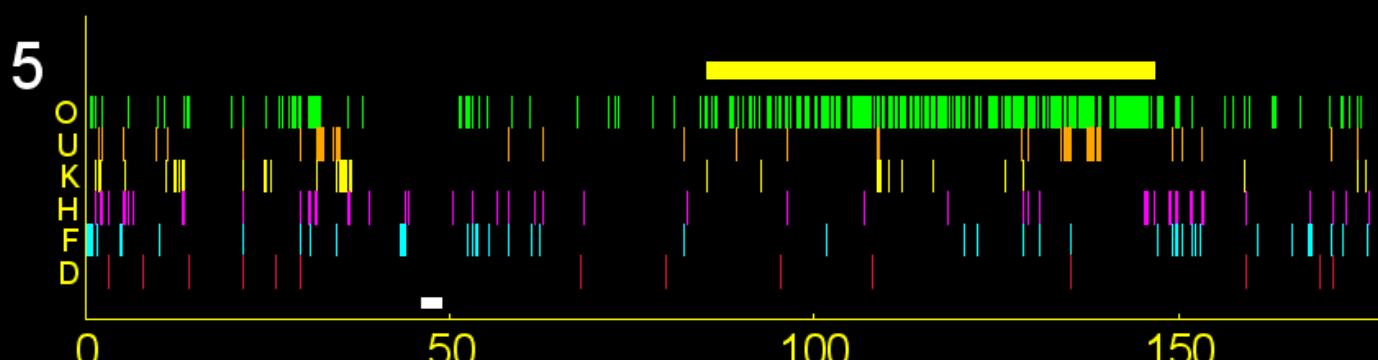
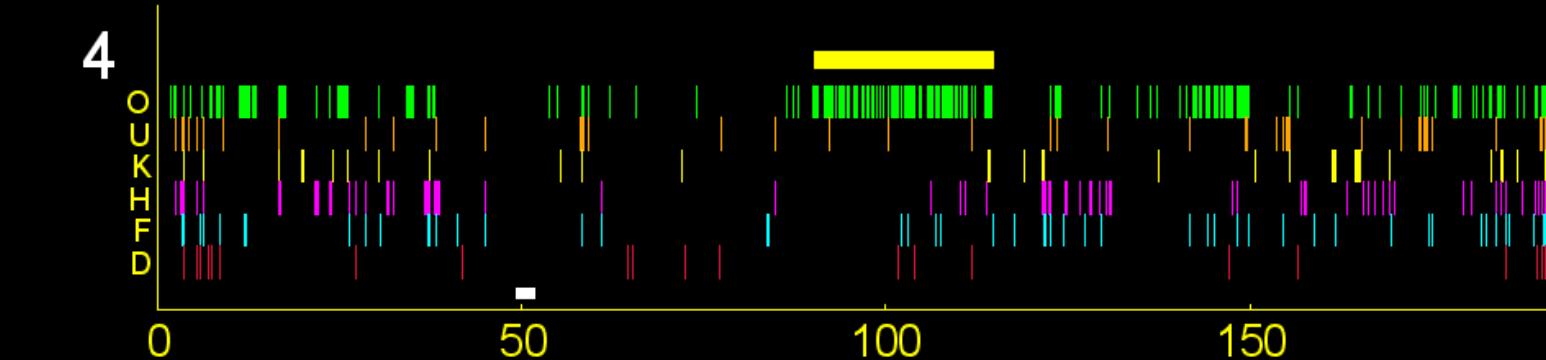
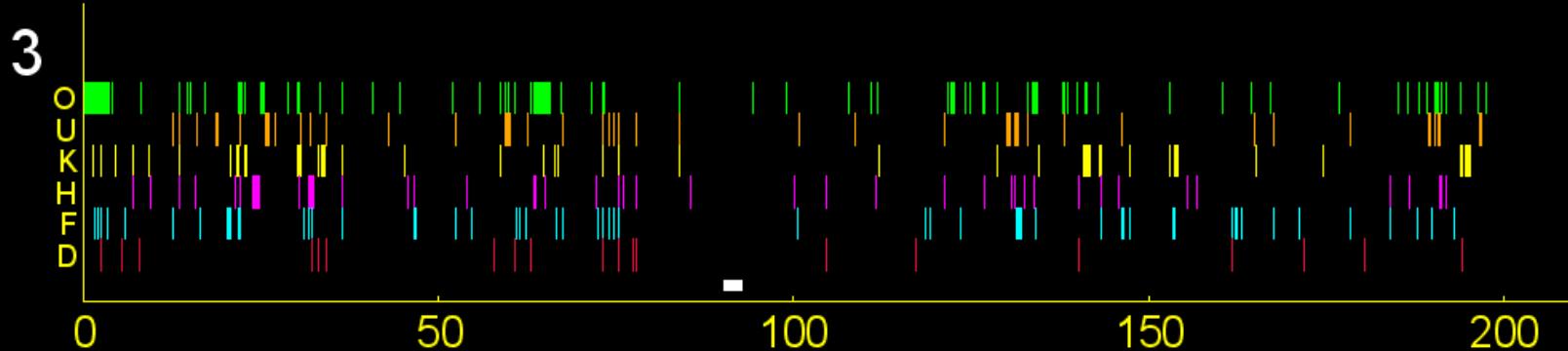
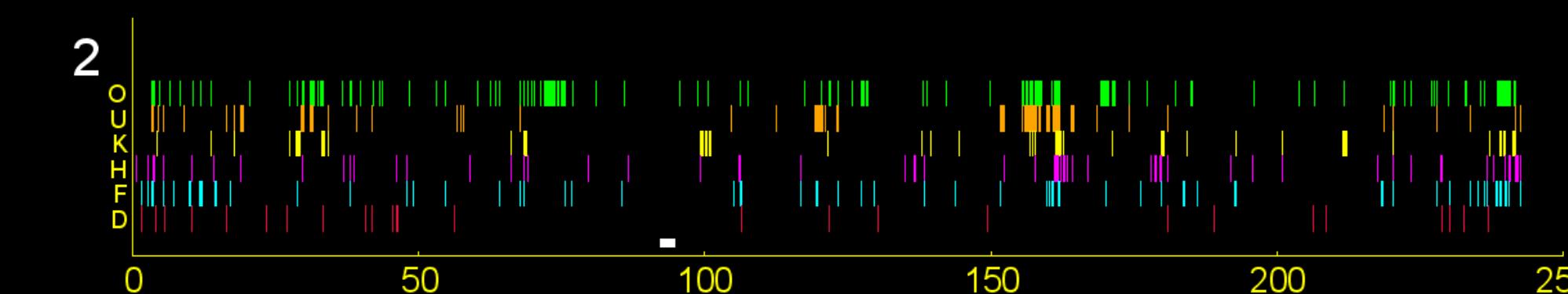
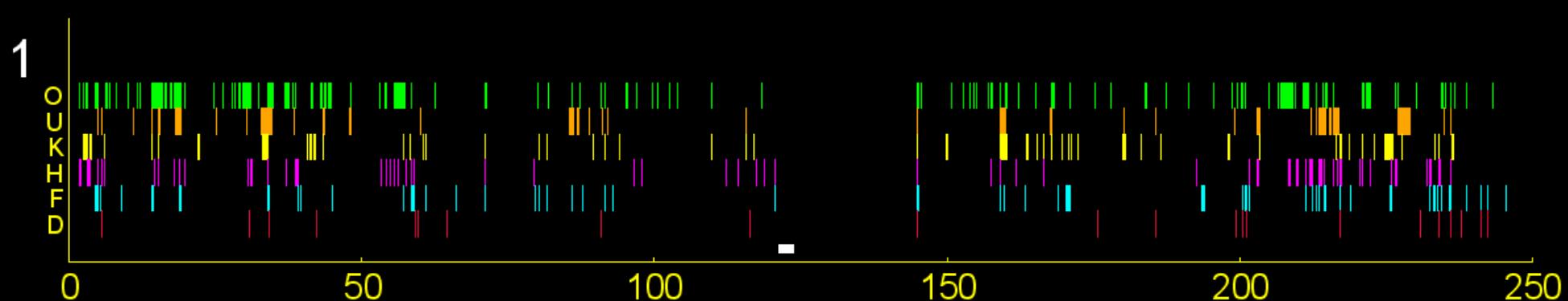
mp@bodkan.net

Morphological evidence?

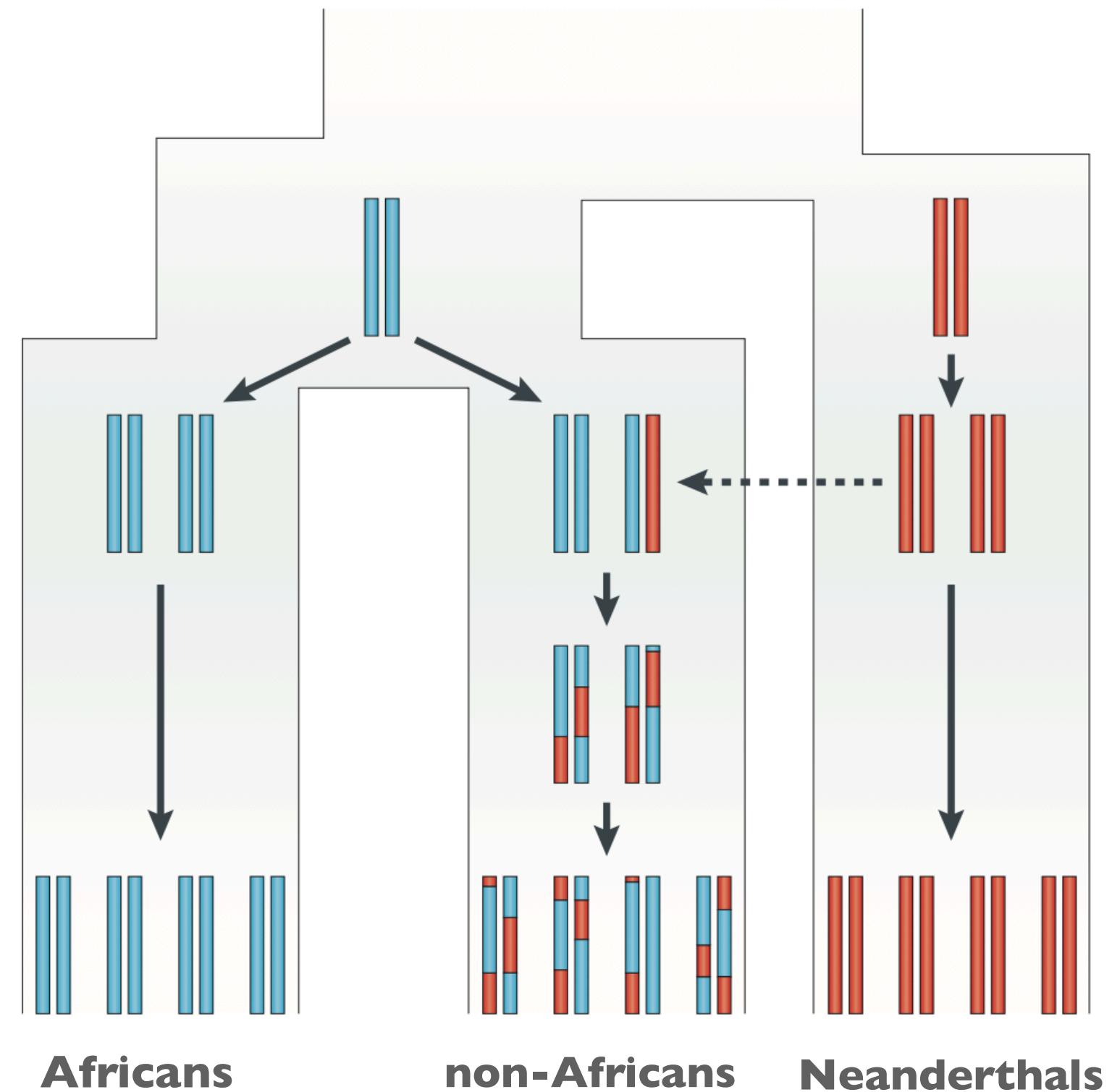


**~40 thousand years old
remains of a modern human
Peștera cu Oase, Romania**

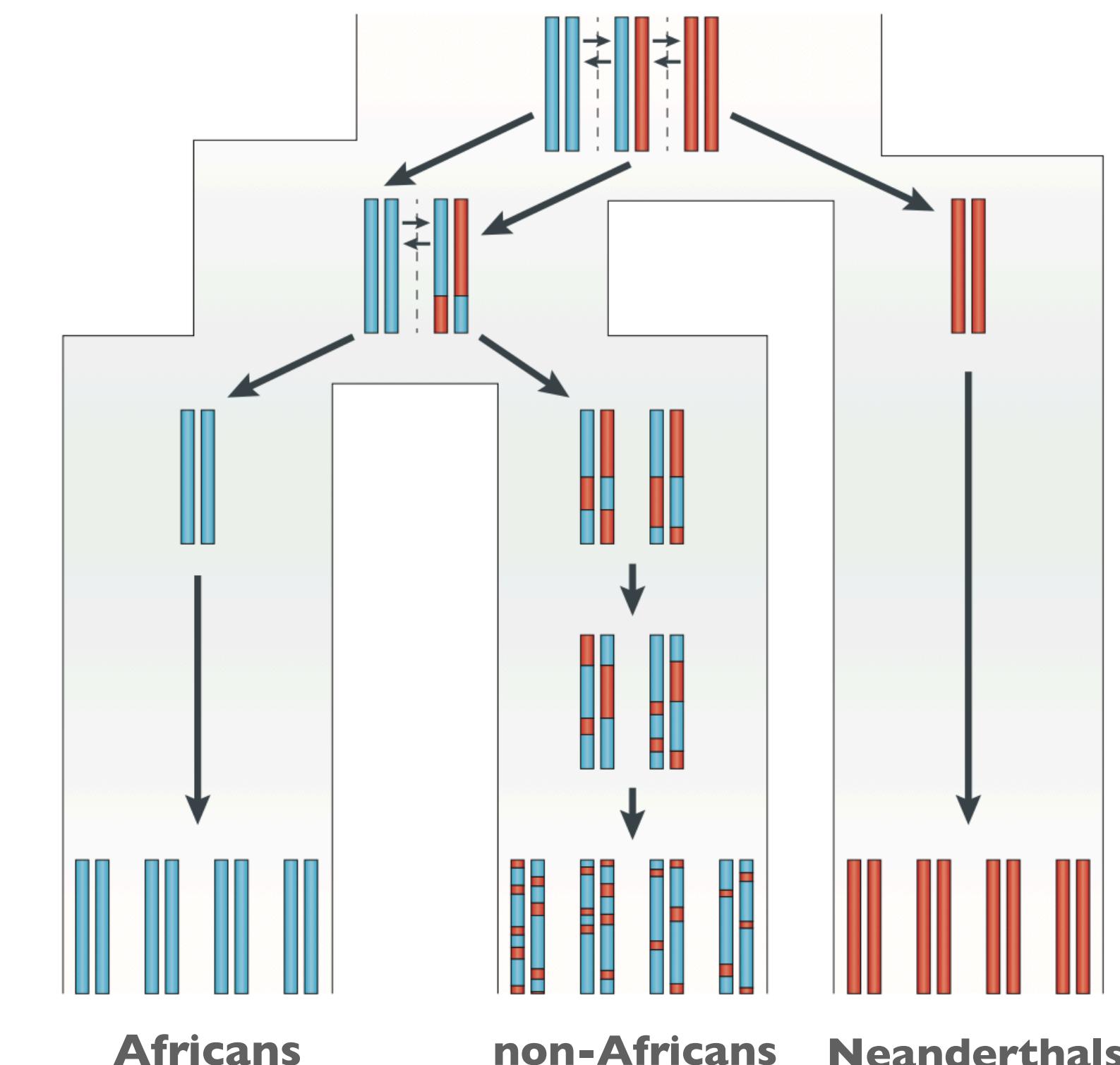




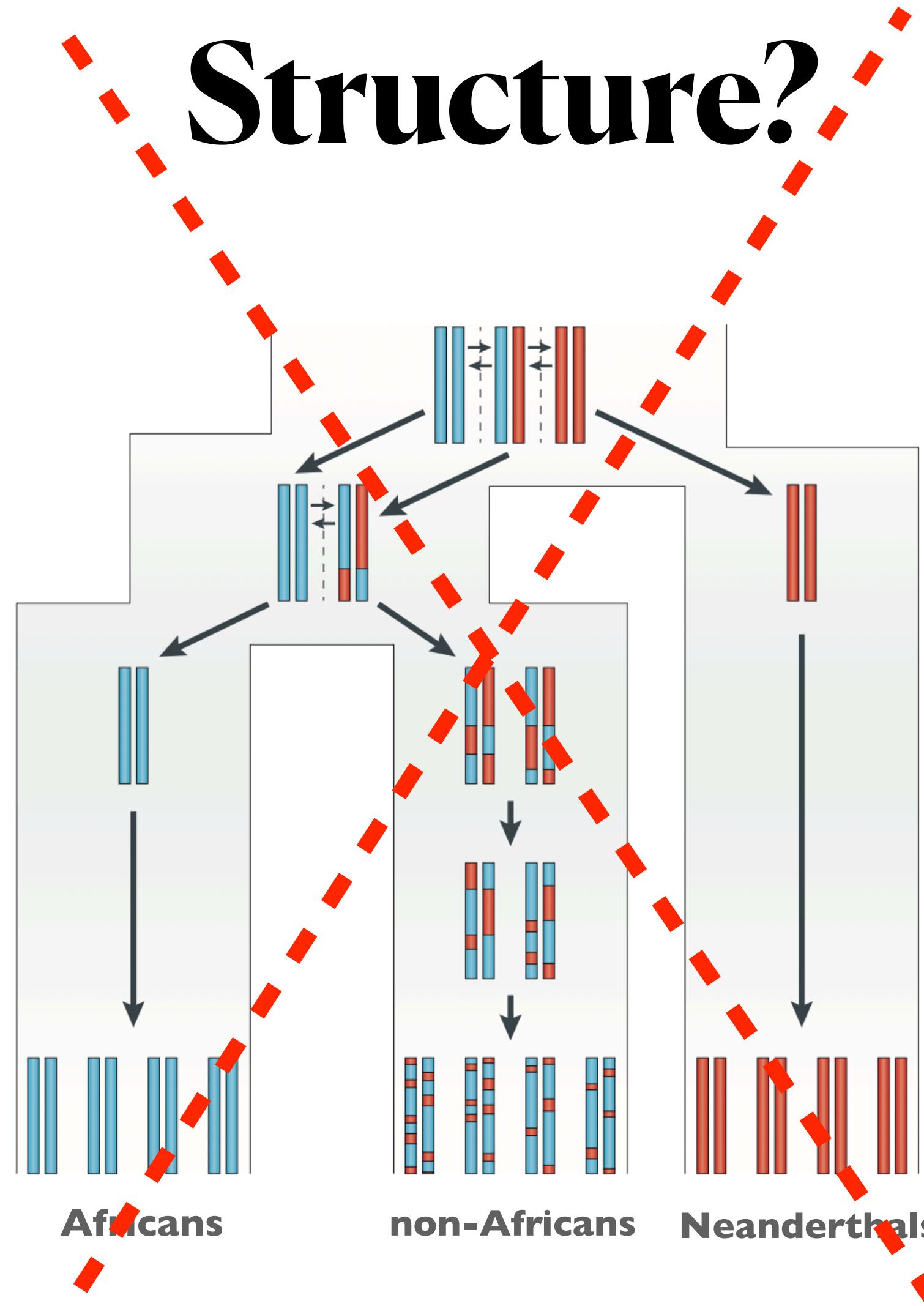
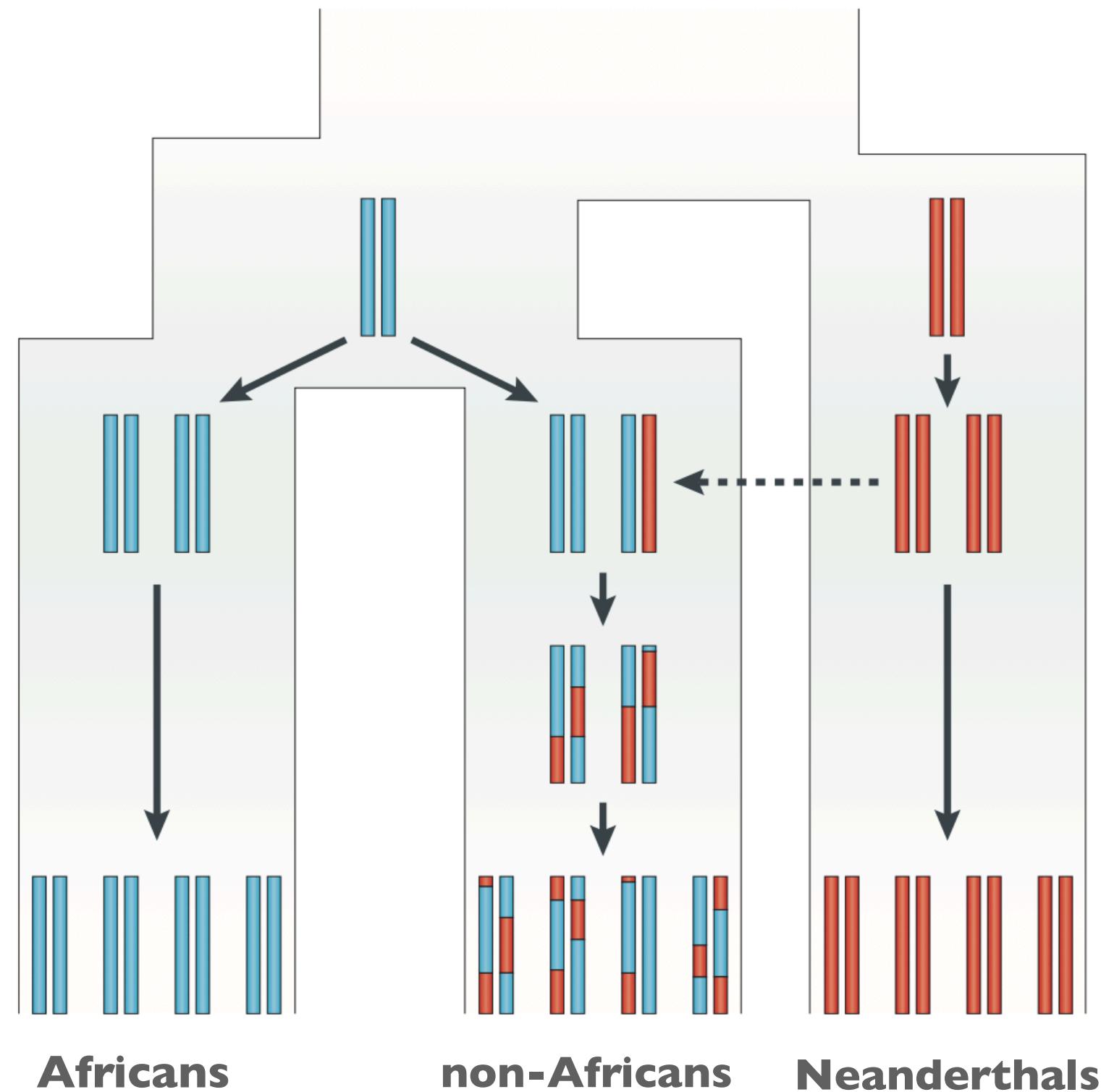
Introgression?



Structure?

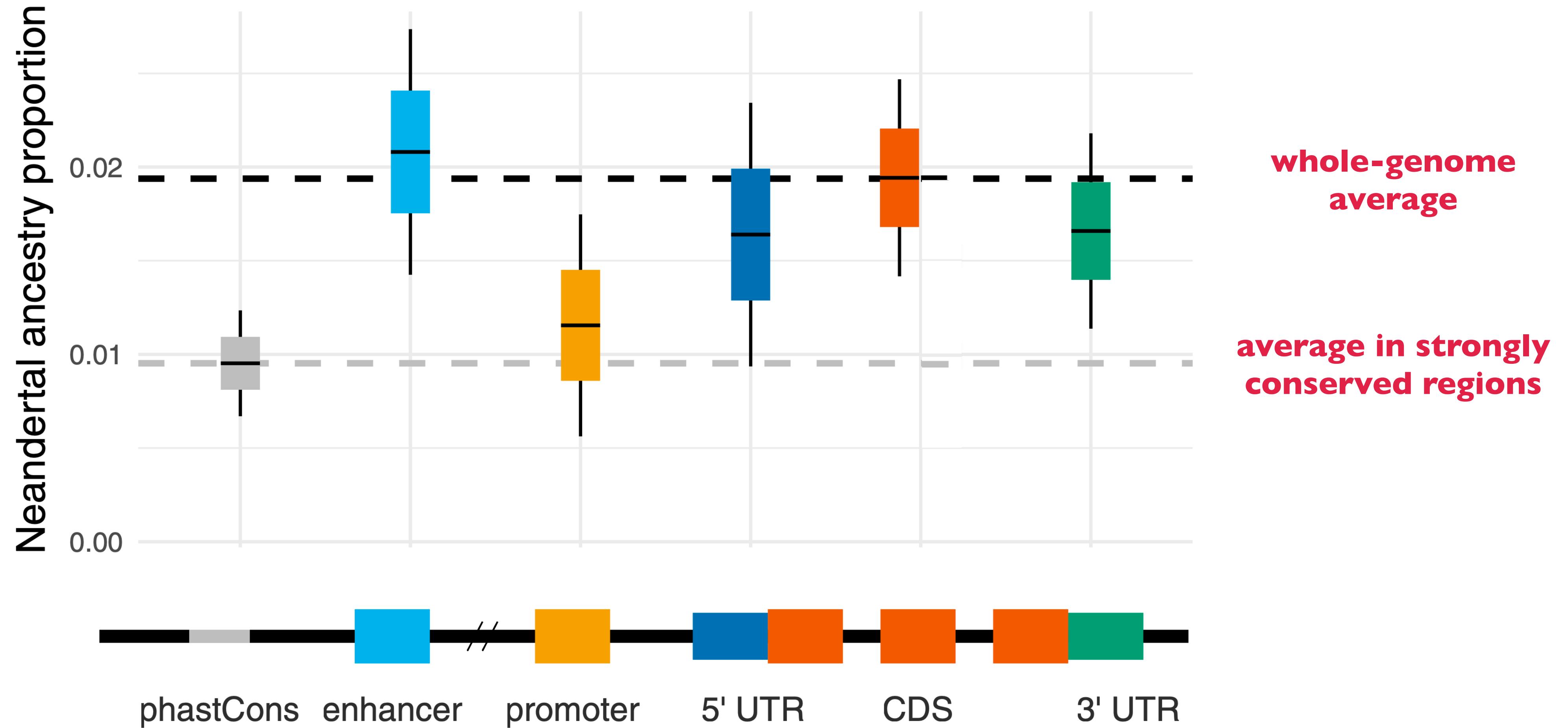


Introgression?

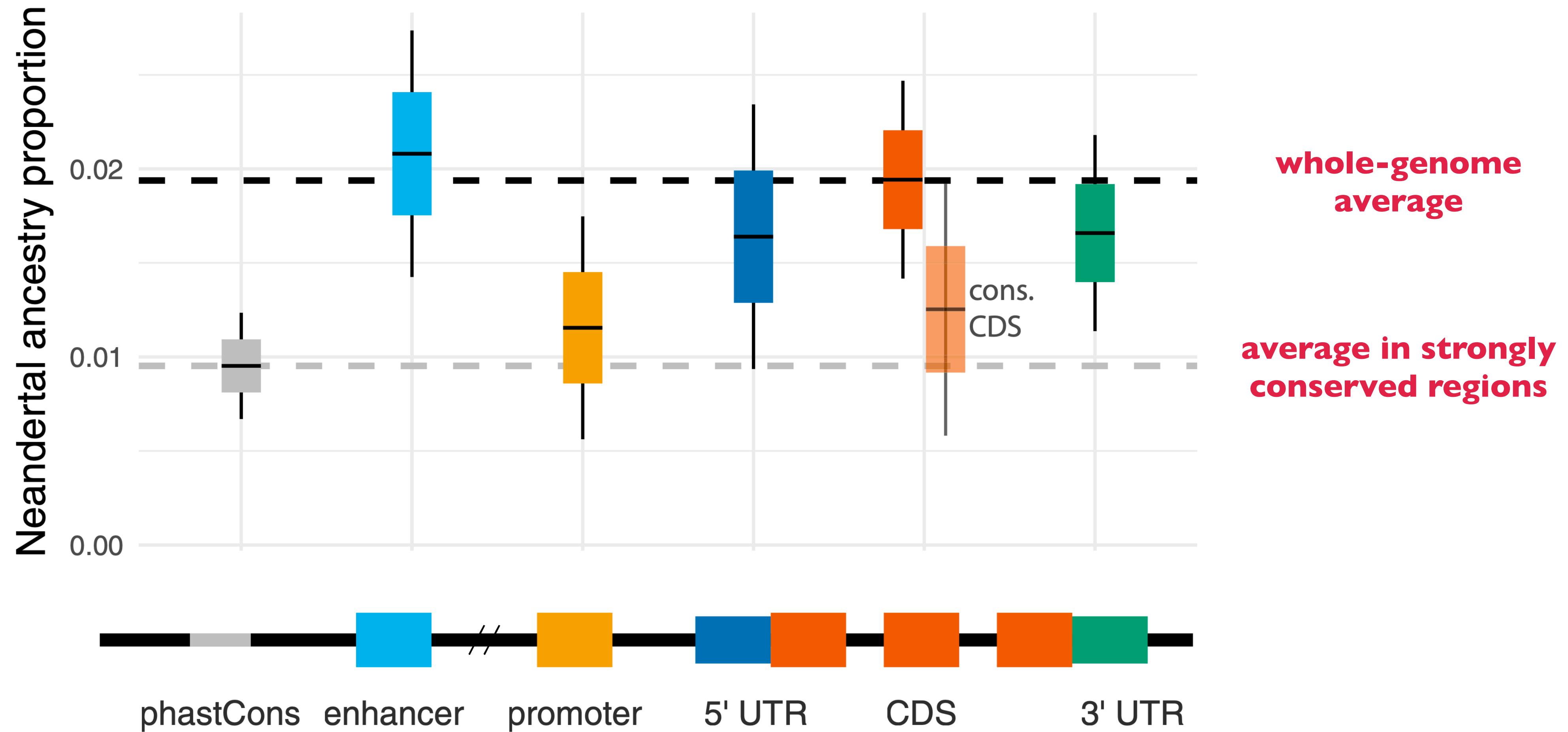


Under structure, “Neanderthal-looking”
haplotypes will be much shorter!

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



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



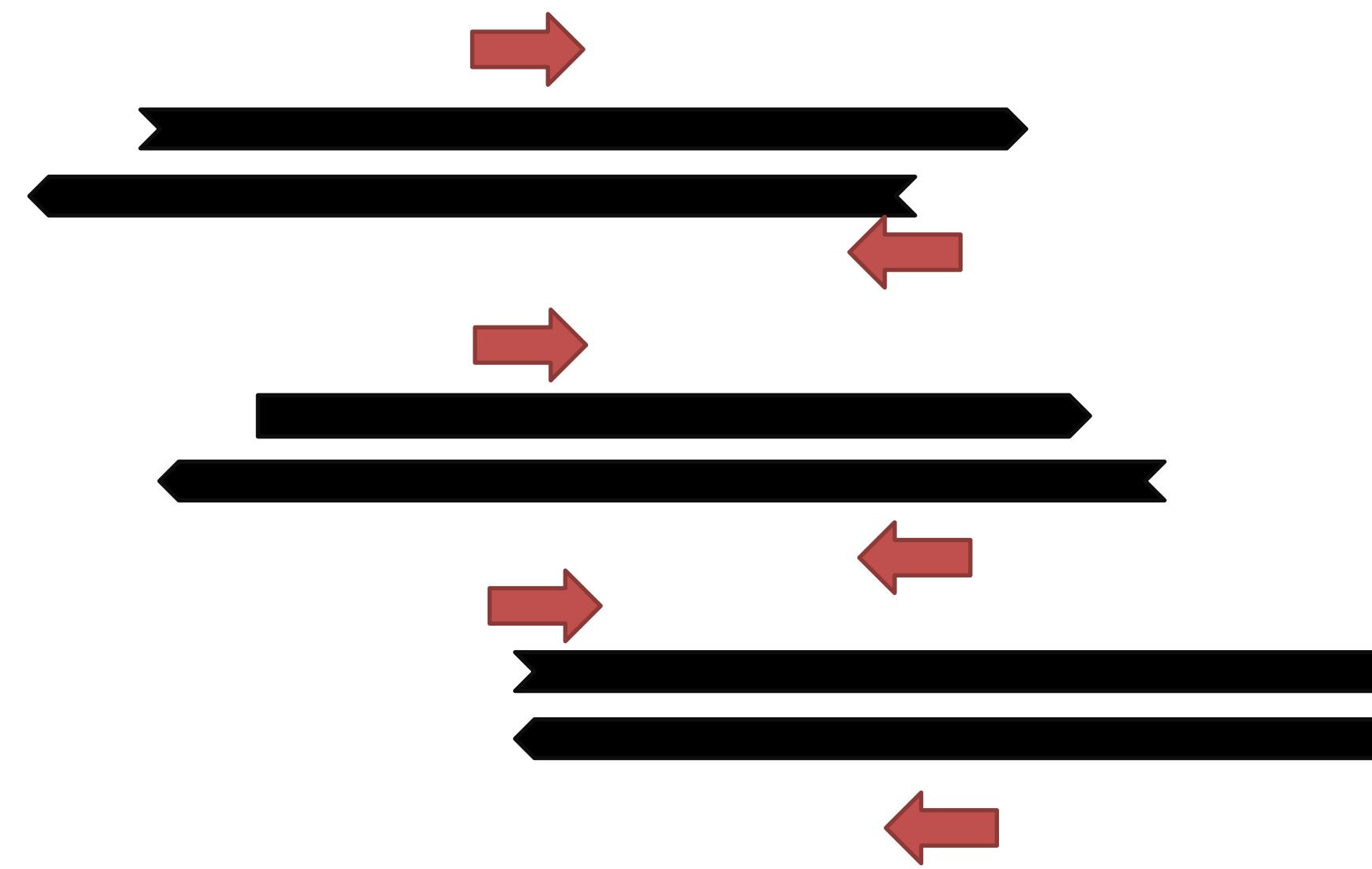
PCR



Library preparation



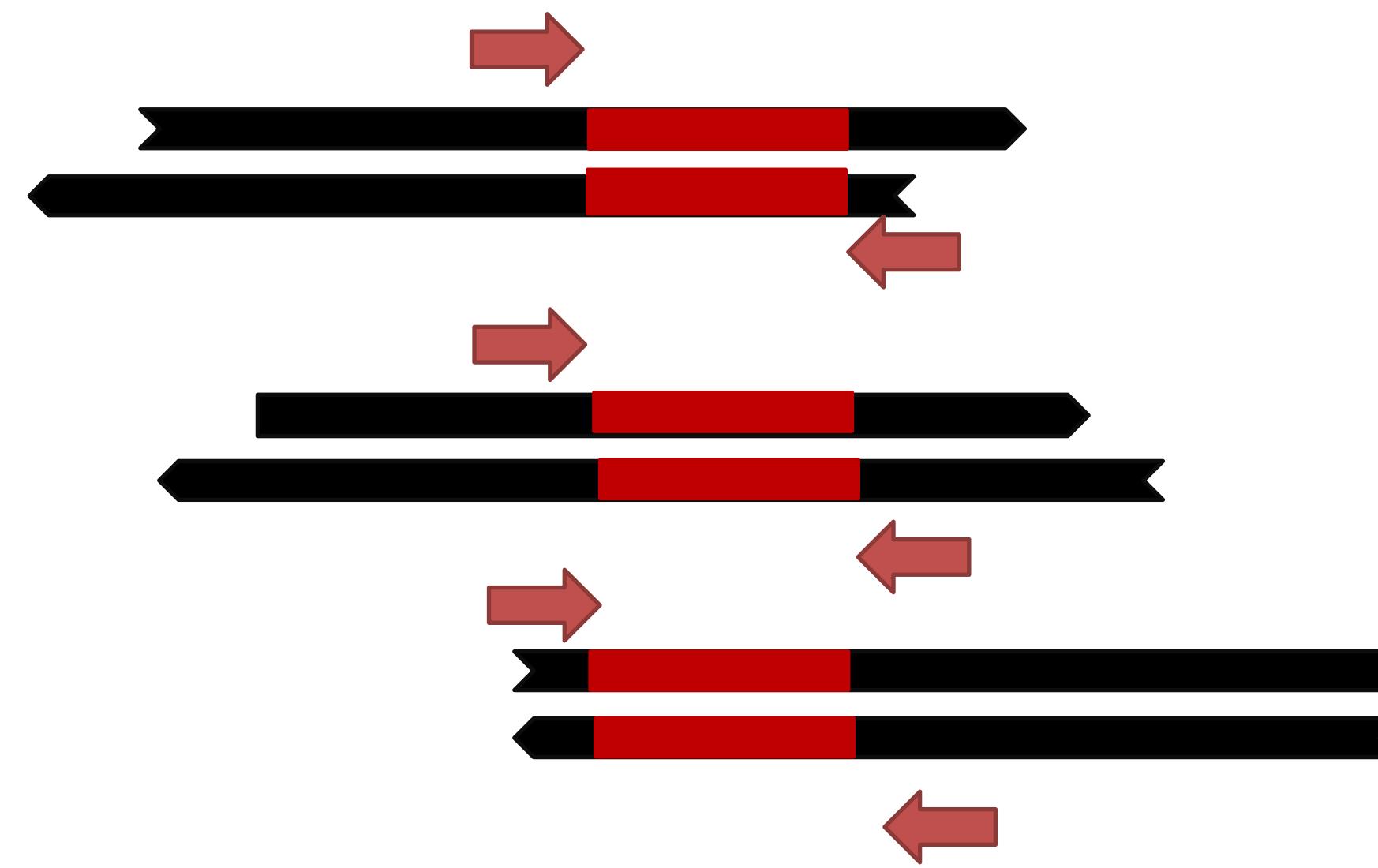
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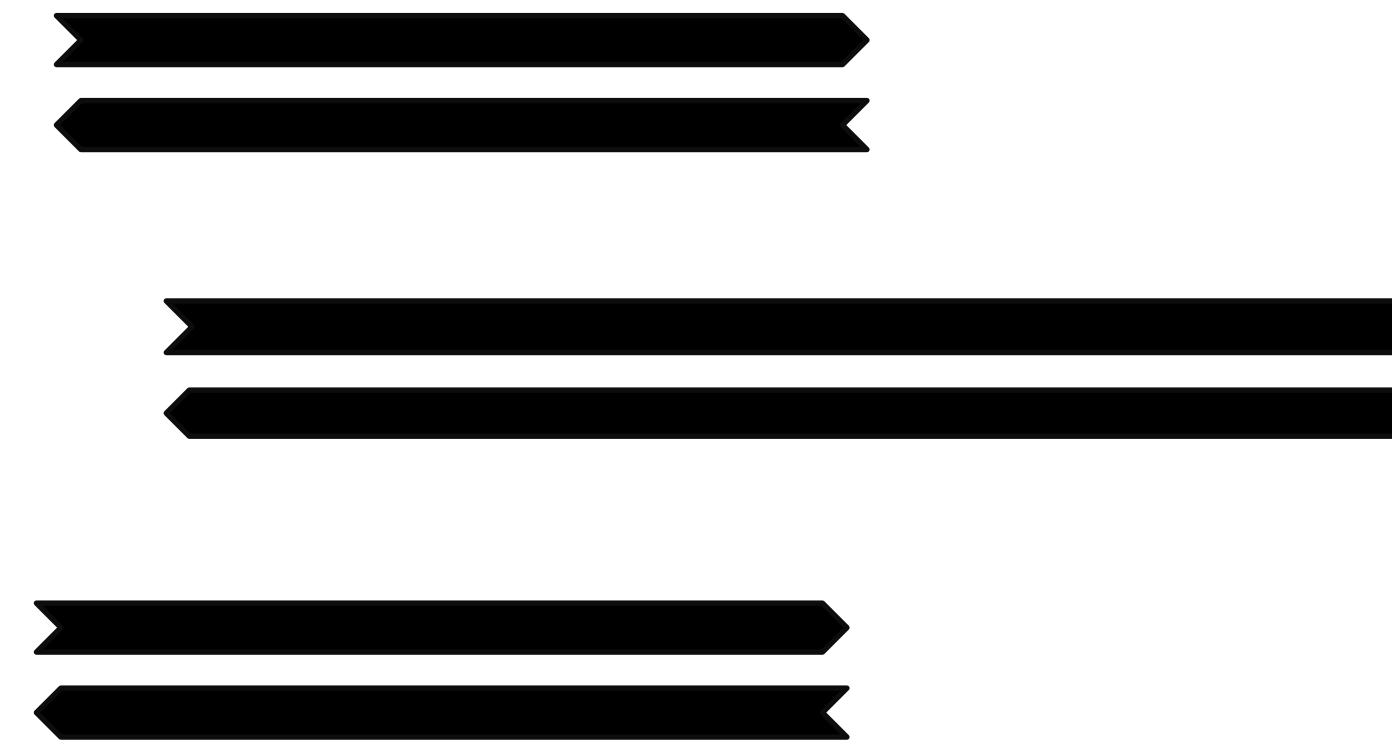
Library preparation



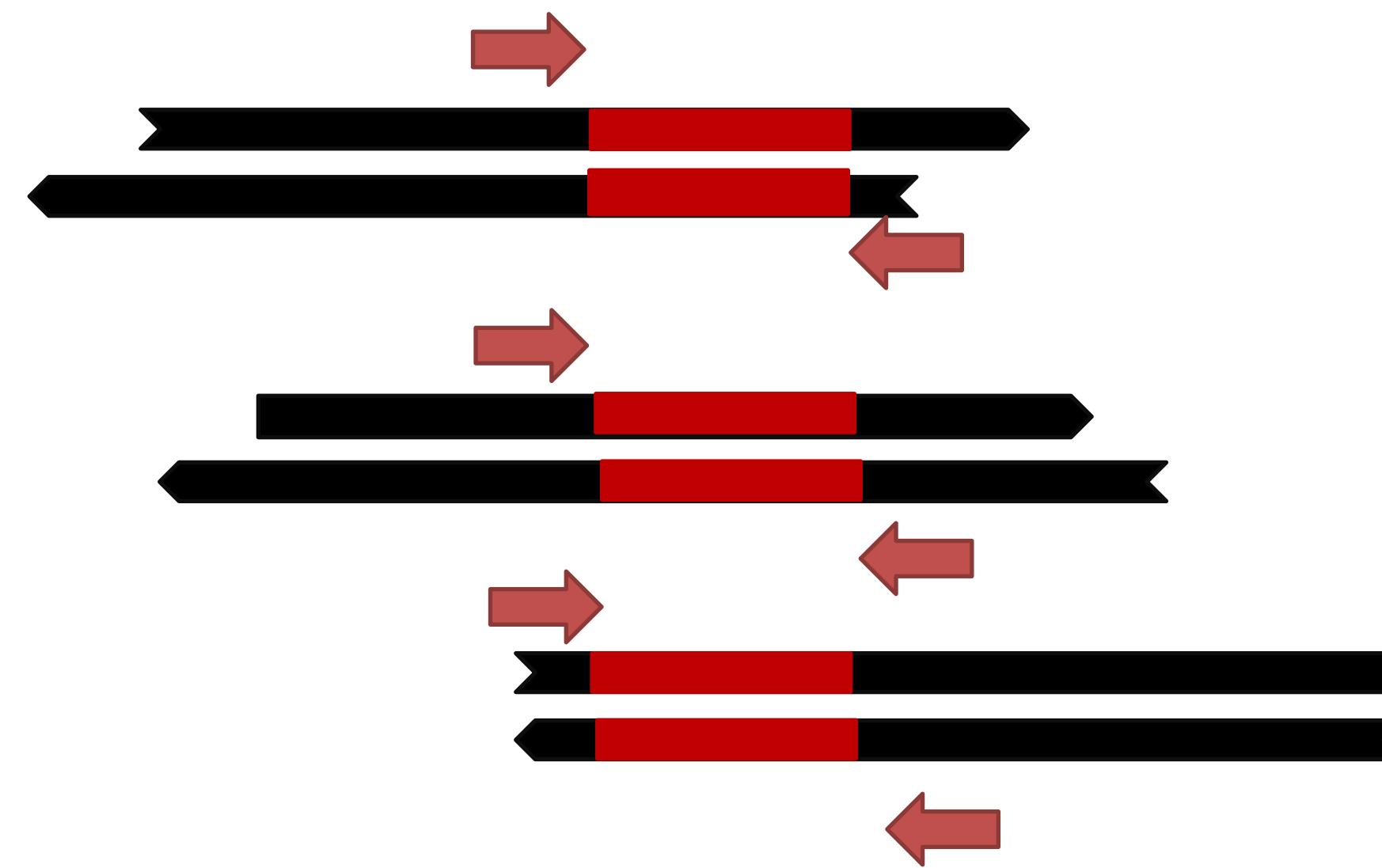
PCR



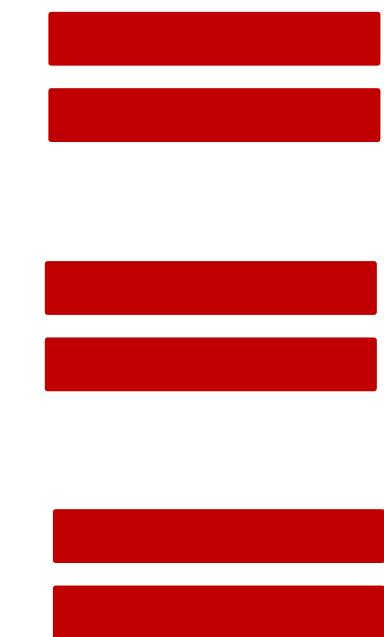
Library preparation



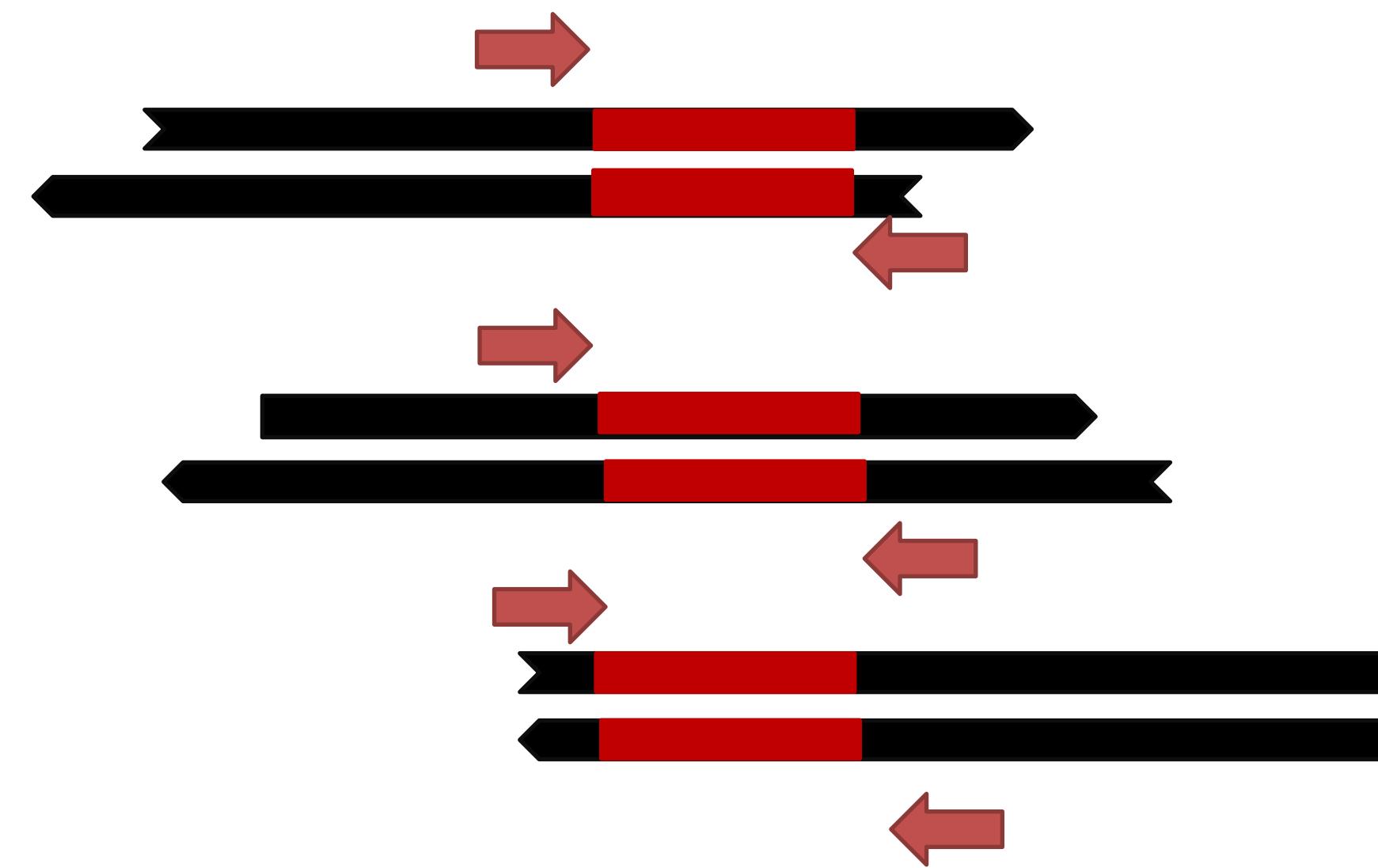
PCR



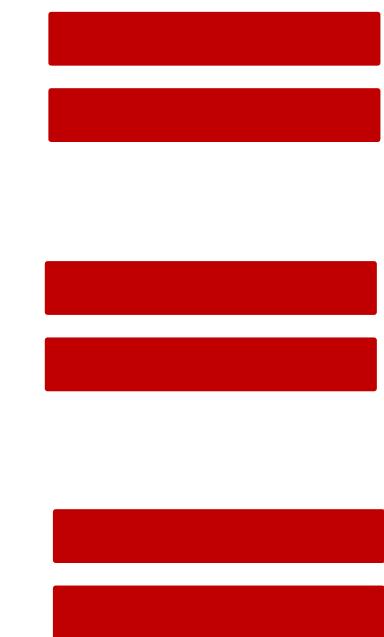
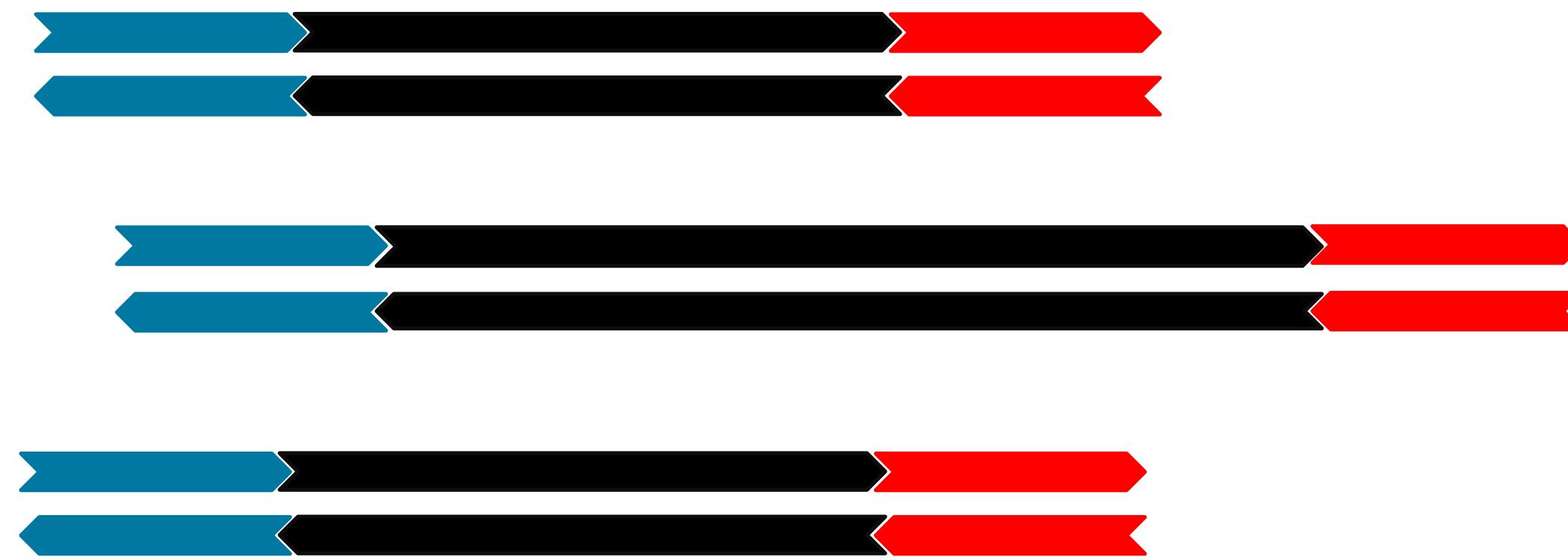
Library preparation



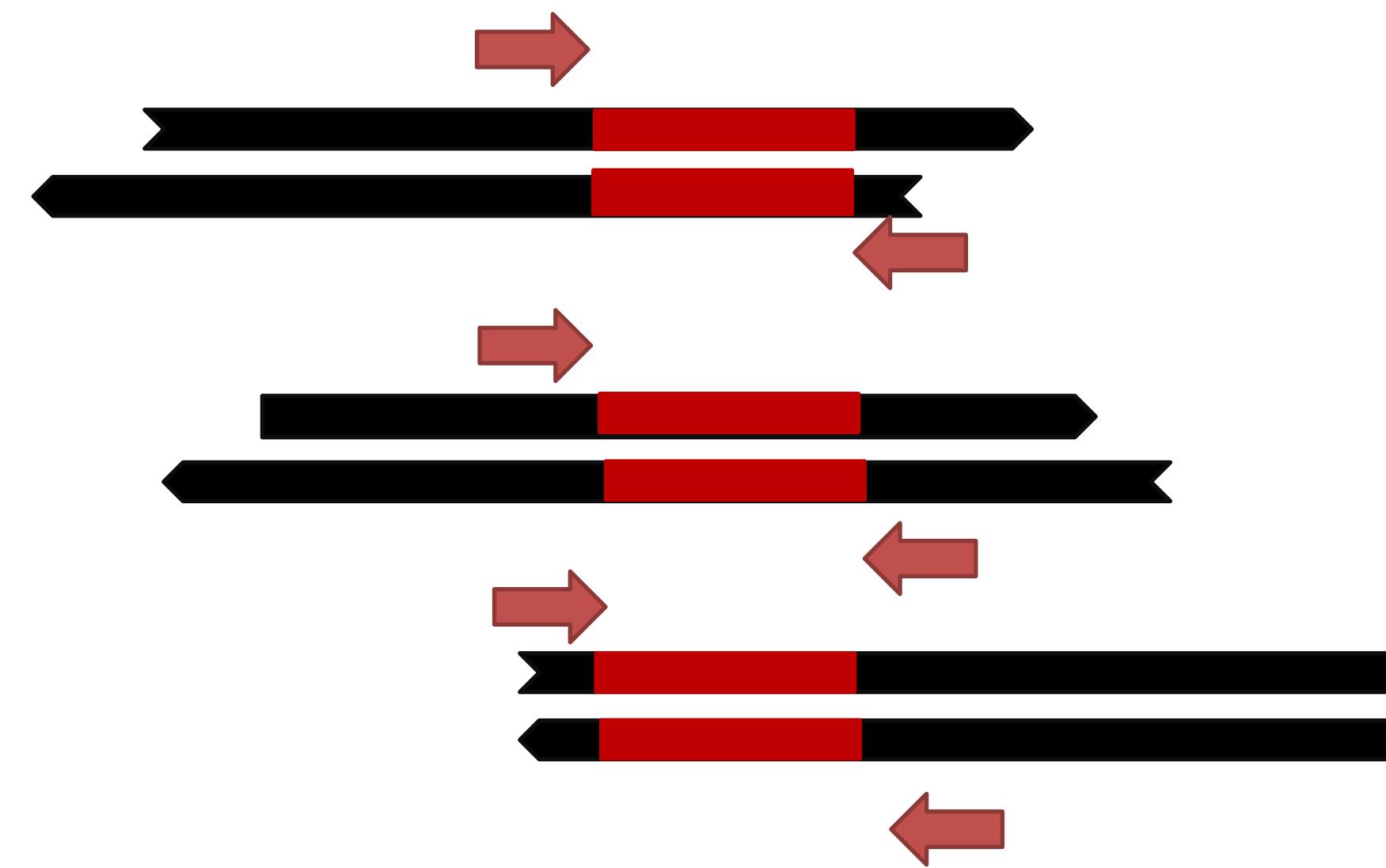
PCR



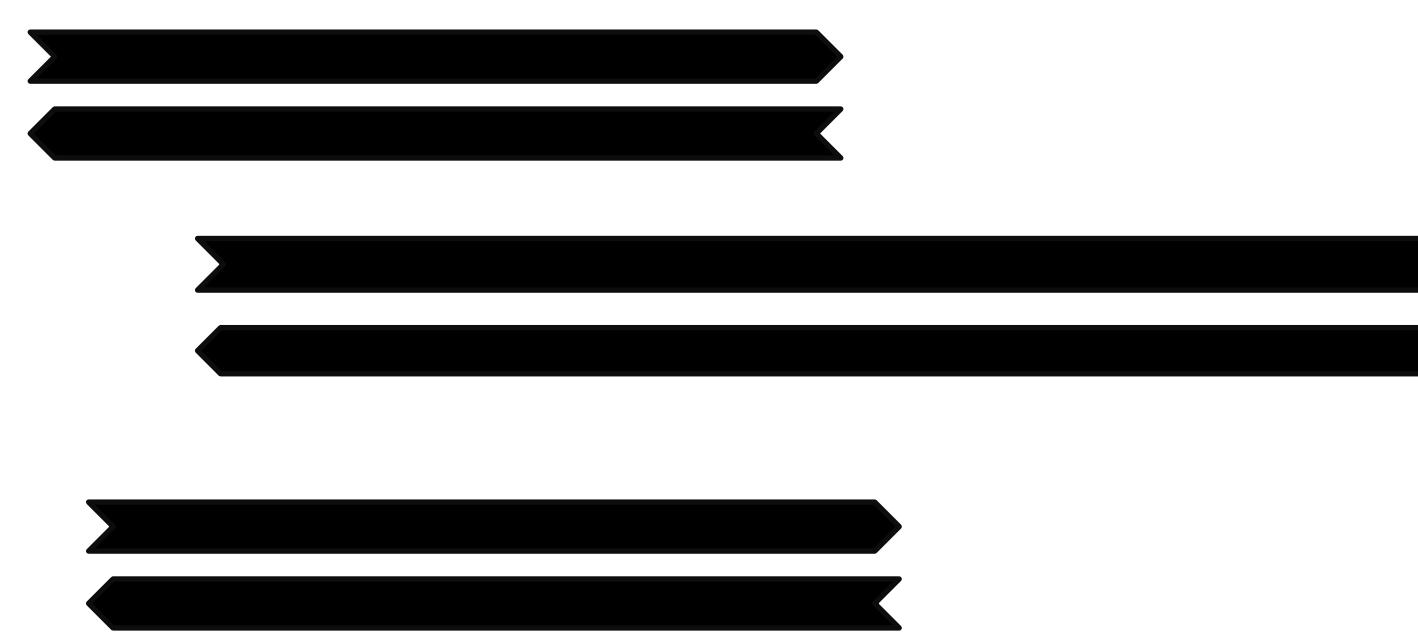
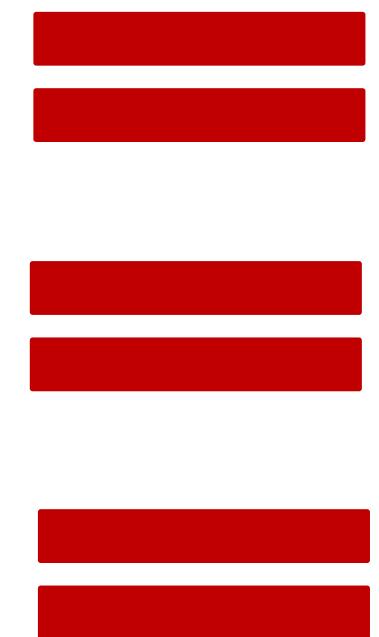
Library preparation

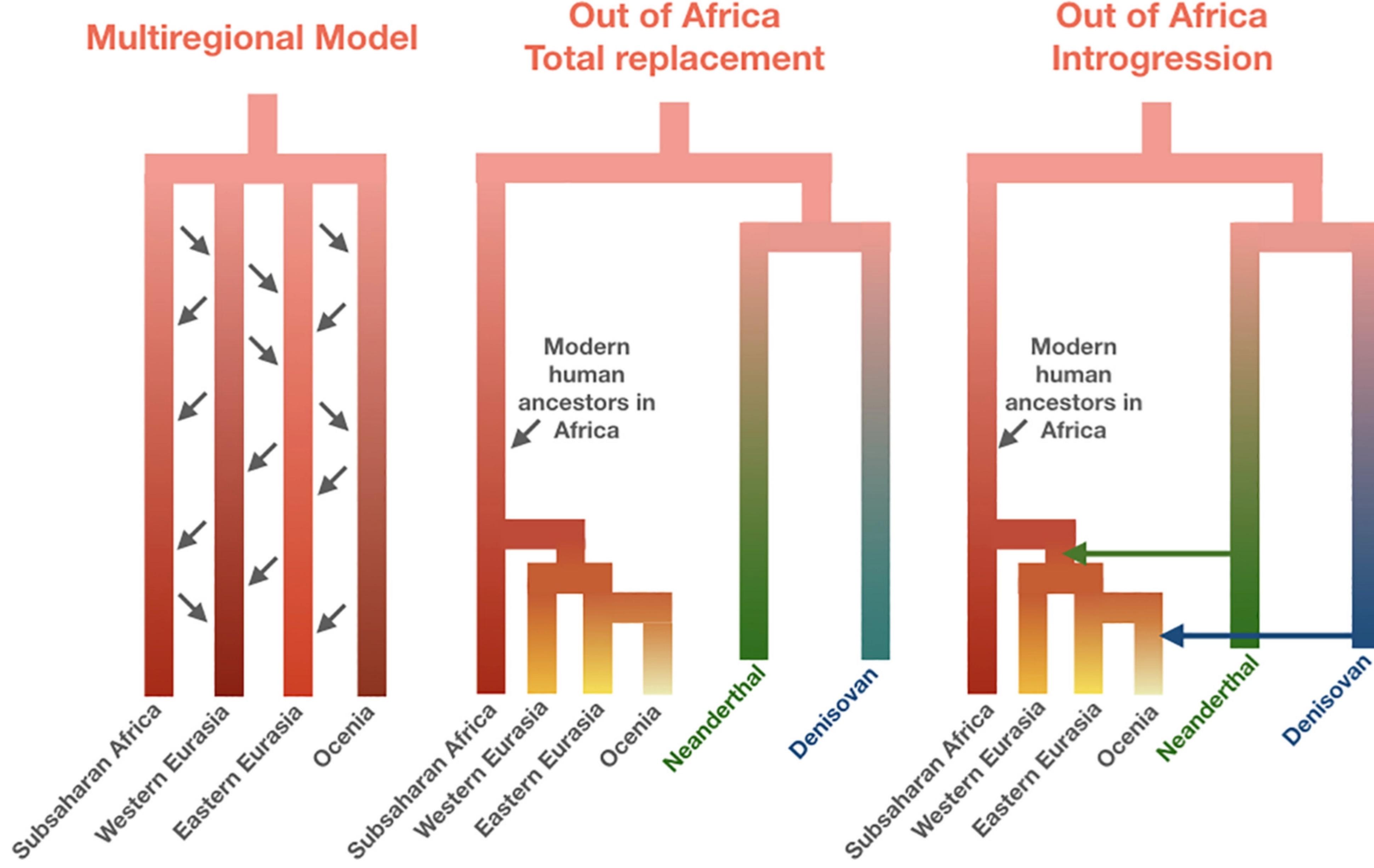


PCR



Library preparation





Genomics in practice...

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##reference=1000Genomes-NCBI37
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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
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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
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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
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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
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```

Genomics in practice...

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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
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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
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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
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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
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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
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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
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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...

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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			
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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			
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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...

#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(A Allele=1 Haplotype)">														
#CHROM	POS	ID	REF	ALT	QUAL	FILTER	INFO	FORMAT	HG00096	HG00097	HG00099	HG00100	HG00101	HG00102
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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 ...