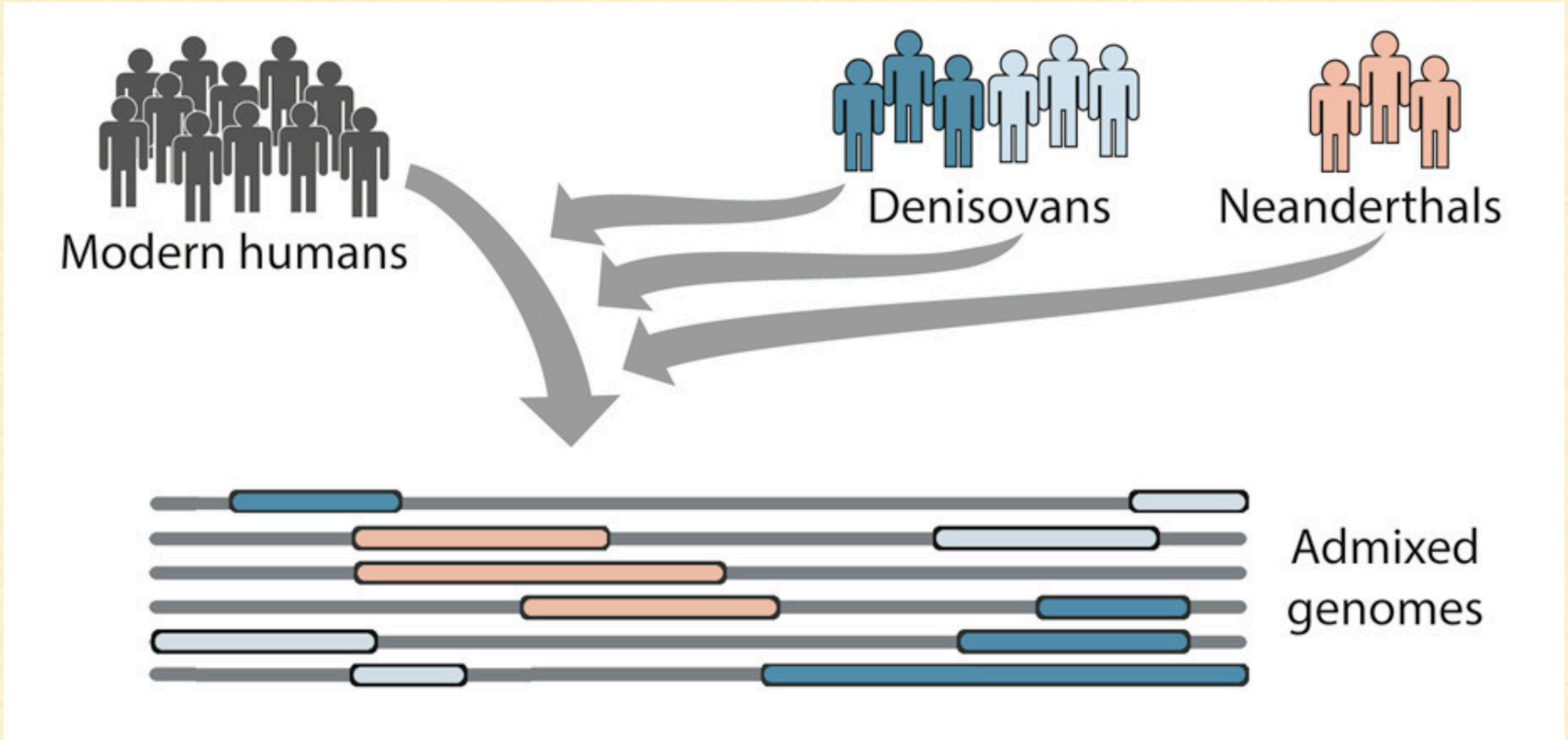
THE EVOLUTIONARY HISTORY OF DENISOVAN AND NEANDERTHAL Y CHROMOSOMES

Martin Petr

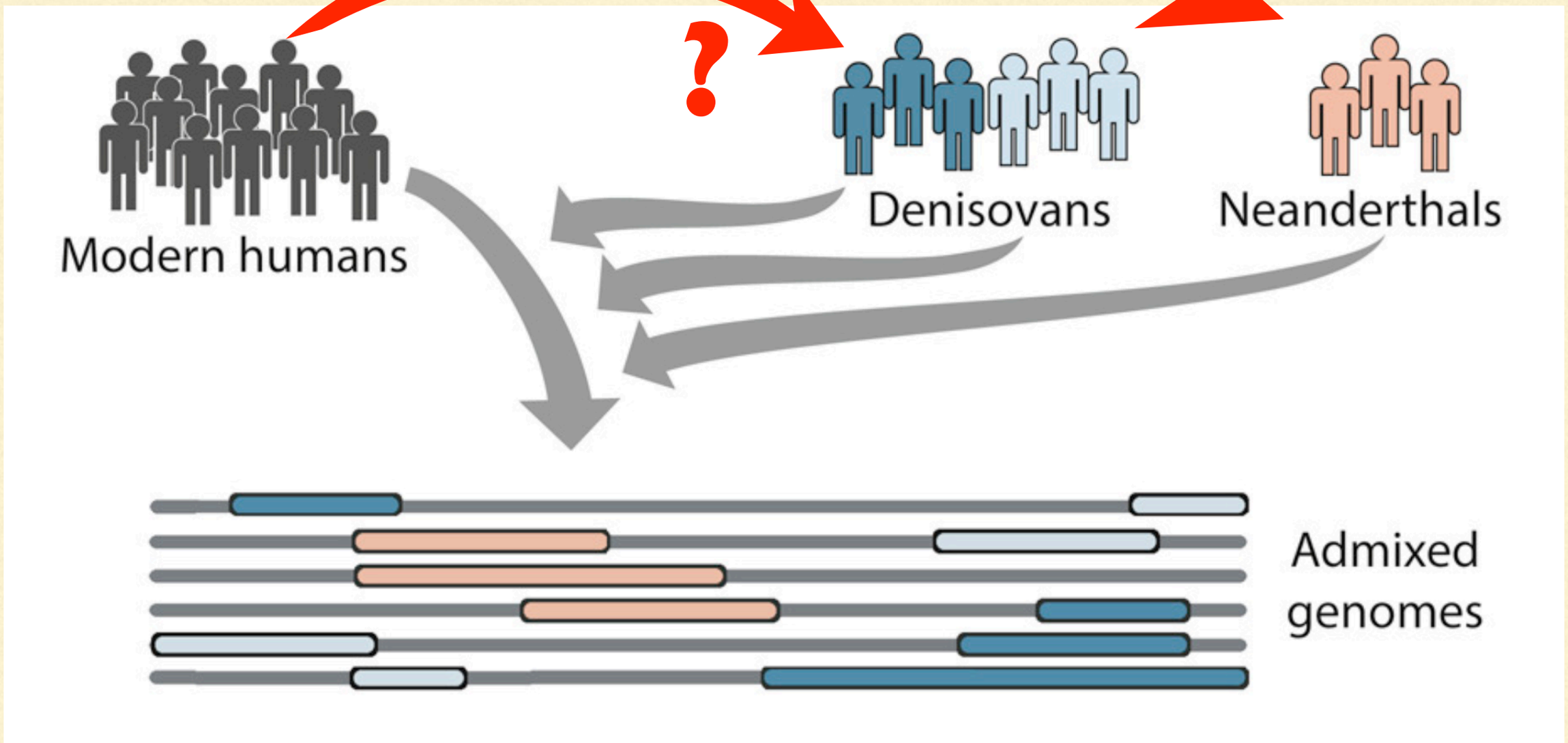
Max Planck Institute for Evolutionary Anthropology

Leipzig, Germany

ADMIXTURE FROM **ARCHAIC HUMANS** INTO **MODERN HUMANS**



modified from Browning al. *Cell*. 2018.



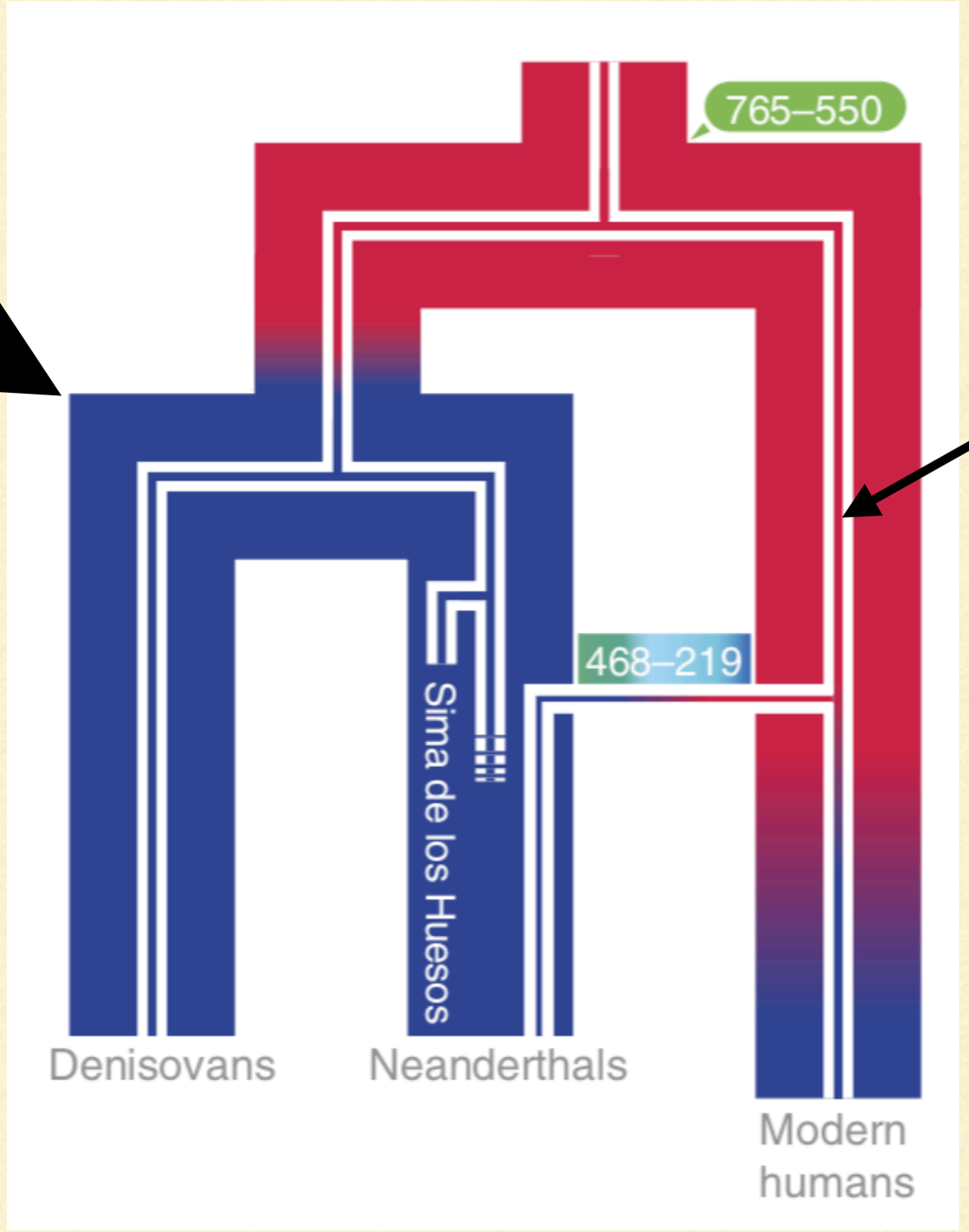
modified from Browning al. *Cell*. 2018.

MODERN → ARCHAIC HUMAN GENE FLOW

— **AUTOSOMAL DNA** VS **MITOCHONDRIAL DNA**

autosomal DNA

mtDNA



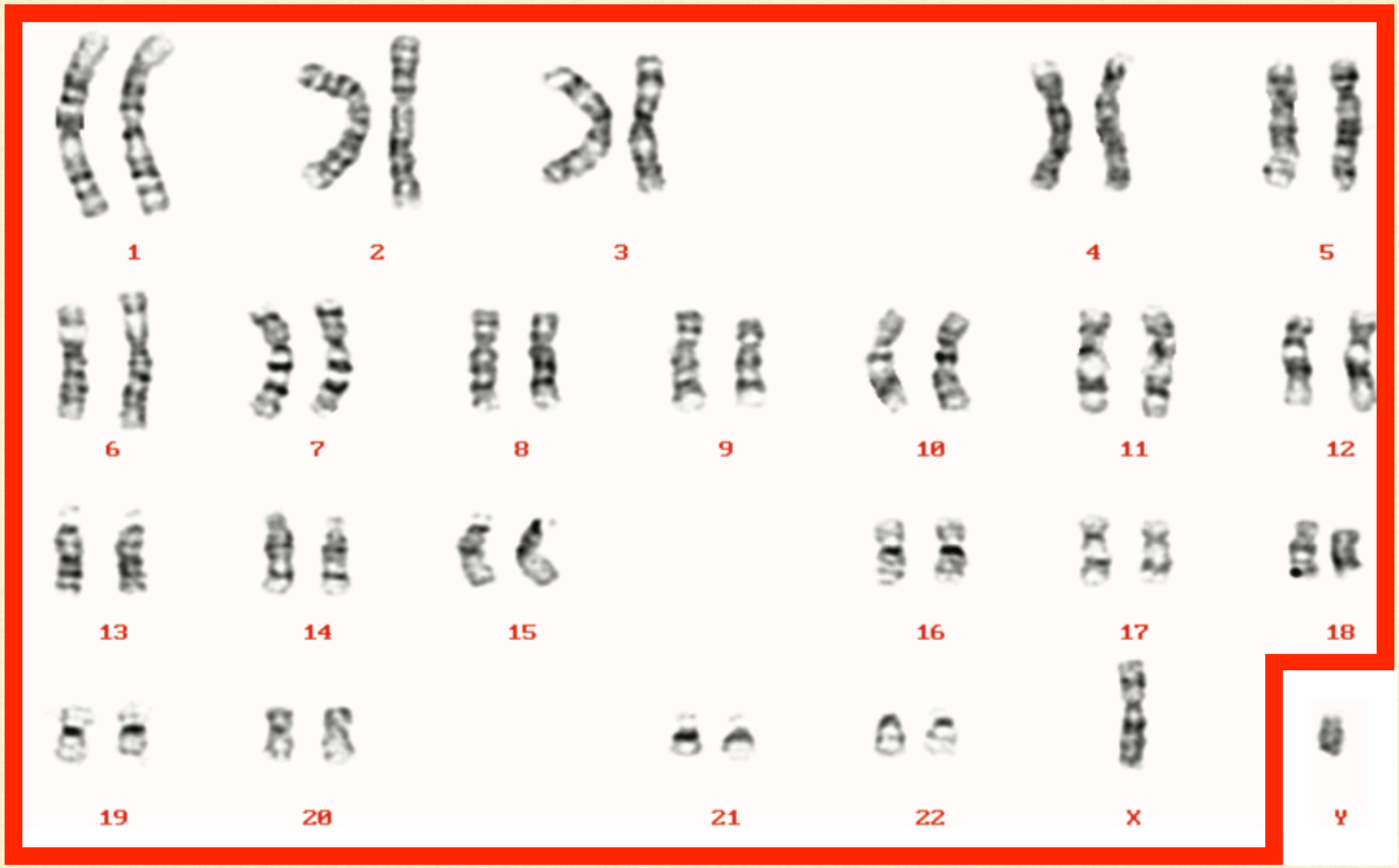
Meyer et al. *Nature* 2014.

Posth et al. *Nat Comm*. 2017

SO FAR, HIGH QUALITY ARCHAICS WERE FEMALE

—Y CHROMOSOMES LARGELY A MYSTERY

human chromosomes:



Kannan Thirumulu Ponnuraj. 2011. *Cytogenetic Techniques in Diagnosing Genetic Disorders.*

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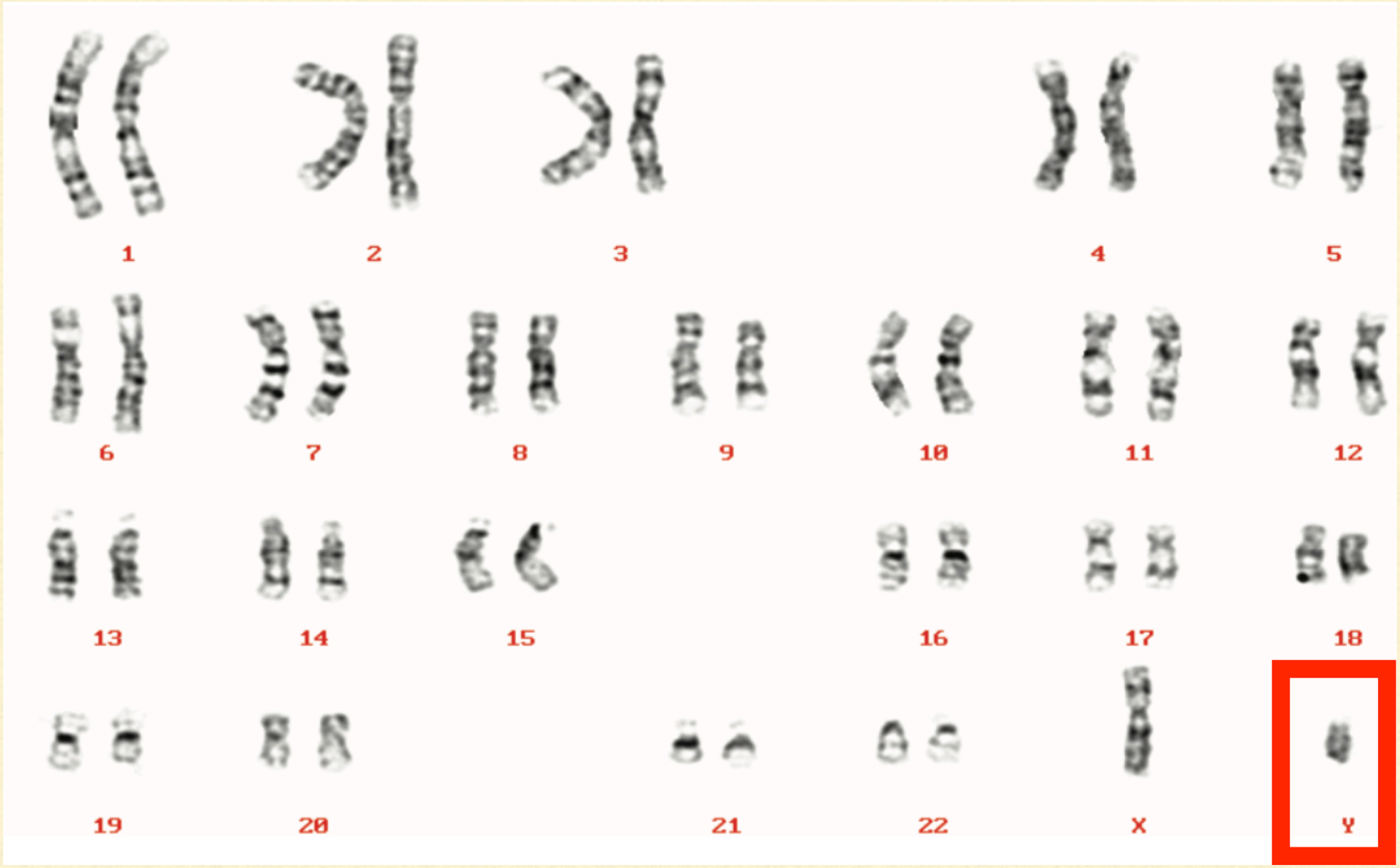
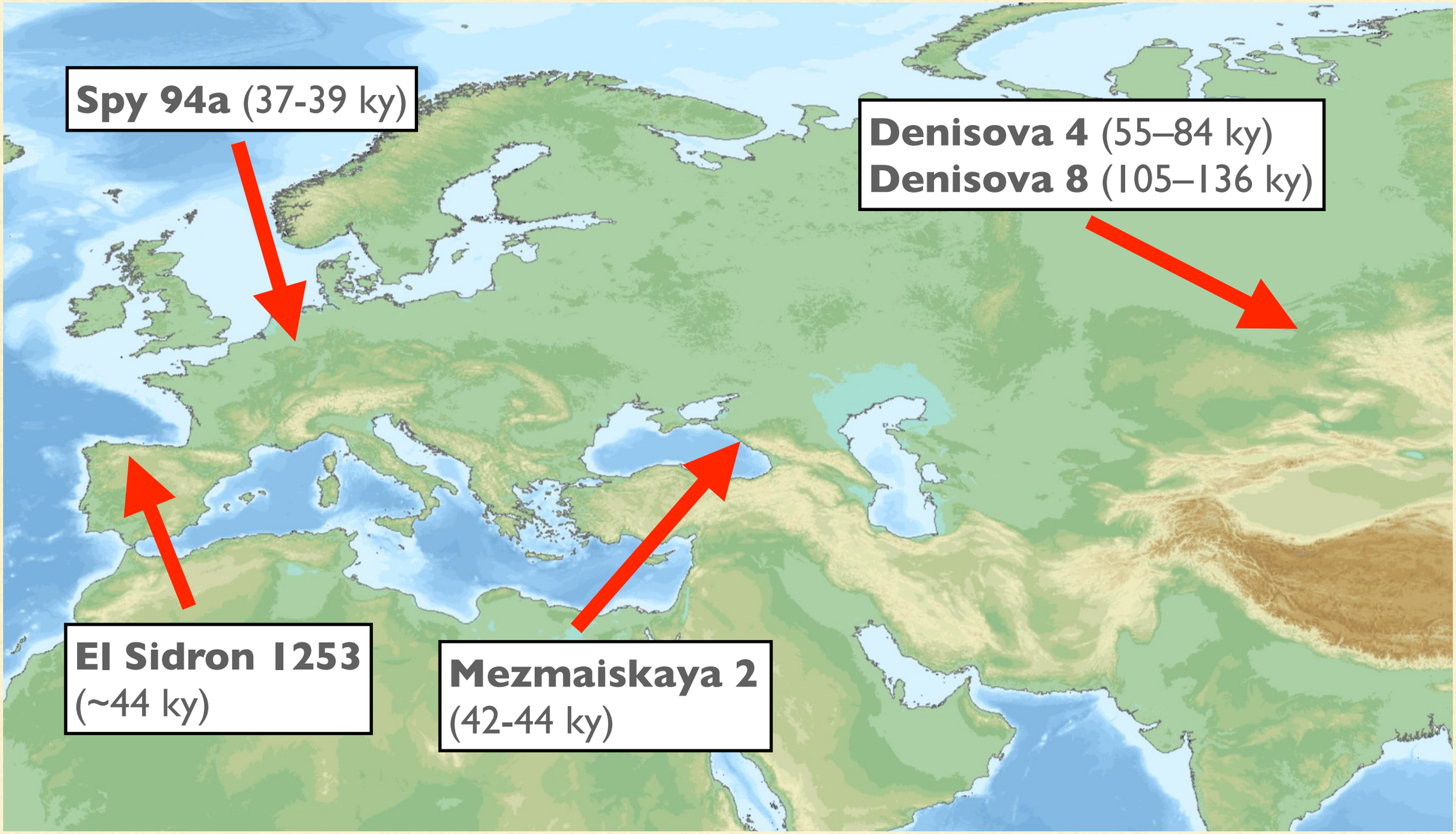


Kannan Thirumulu Ponnuraj. 2011. *Cytogenetic Techniques in Diagnosing Genetic Disorders.*

FOCUSING ON Y CHROMOSOMES

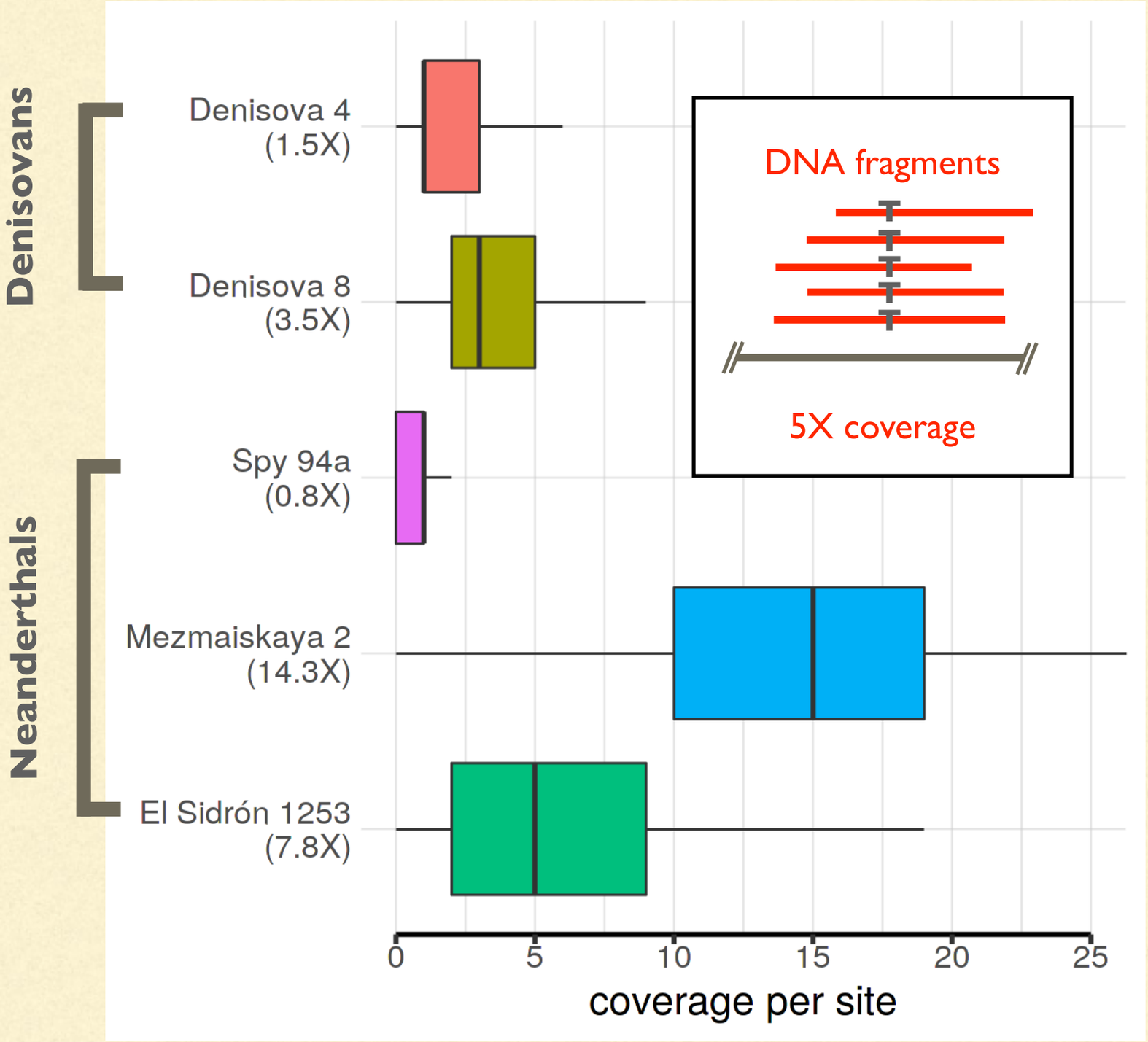
—ARCHAIC HUMAN MALES



Relief map of Eurasia. Giorgi Balakhadze. Wikipedia. Creative Commons Attribution-Share Alike 4.0 International

COVERAGE OF Y CHROMOSOMES

(USING "WHOLE CHROMOSOME CAPTURE" METHOD)



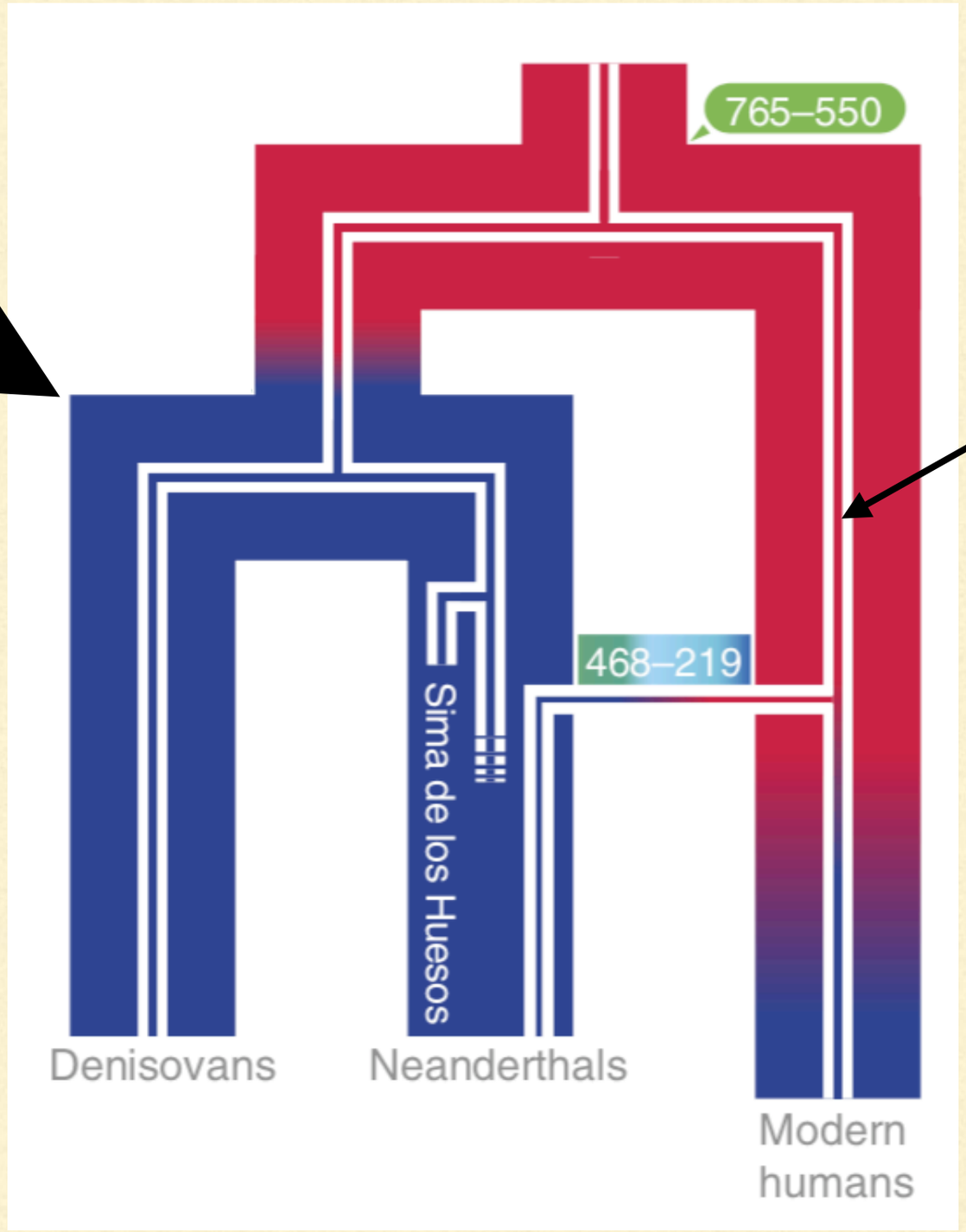
Petr et al., *in preparation*

DISCORDANT AUTOSOMAL AND MT DATA

... WHAT ABOUT Y CHROMOSOMES?

autosomal DNA

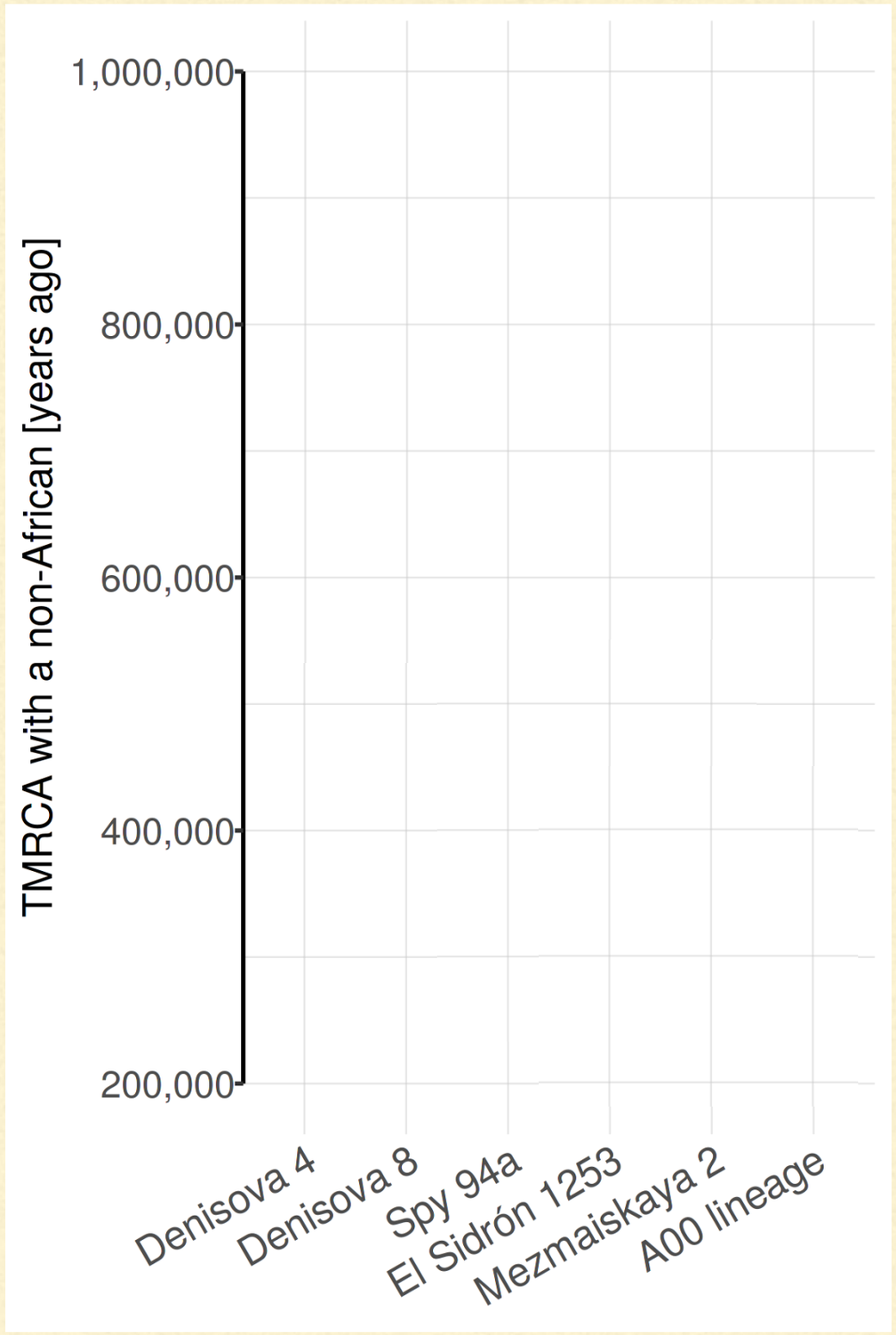
mtDNA

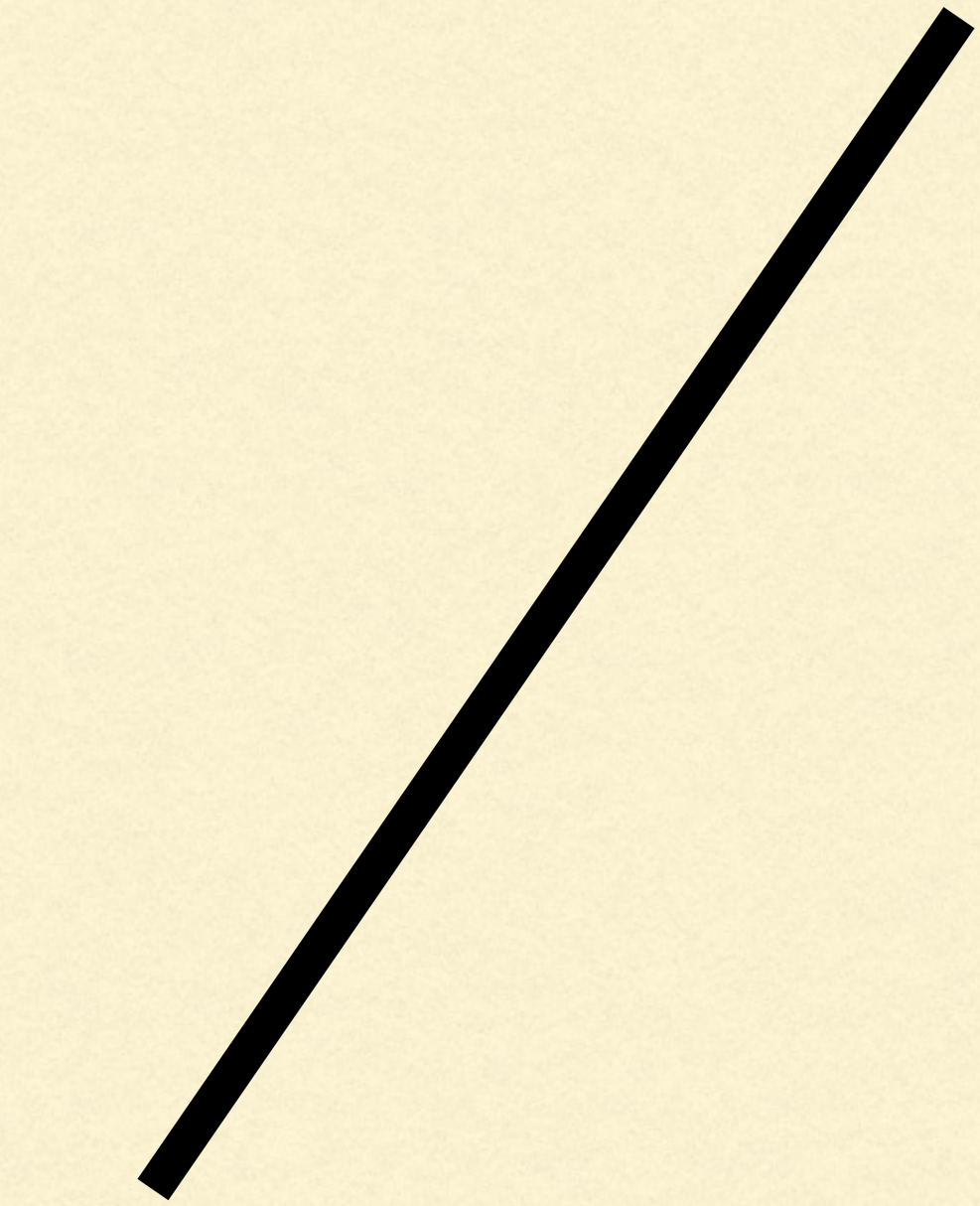


Posth et al. *Nat Comm*. 2017

ESTIMATES OF TMRCA

WITH MODERN HUMAN Y CHROMOSOMES





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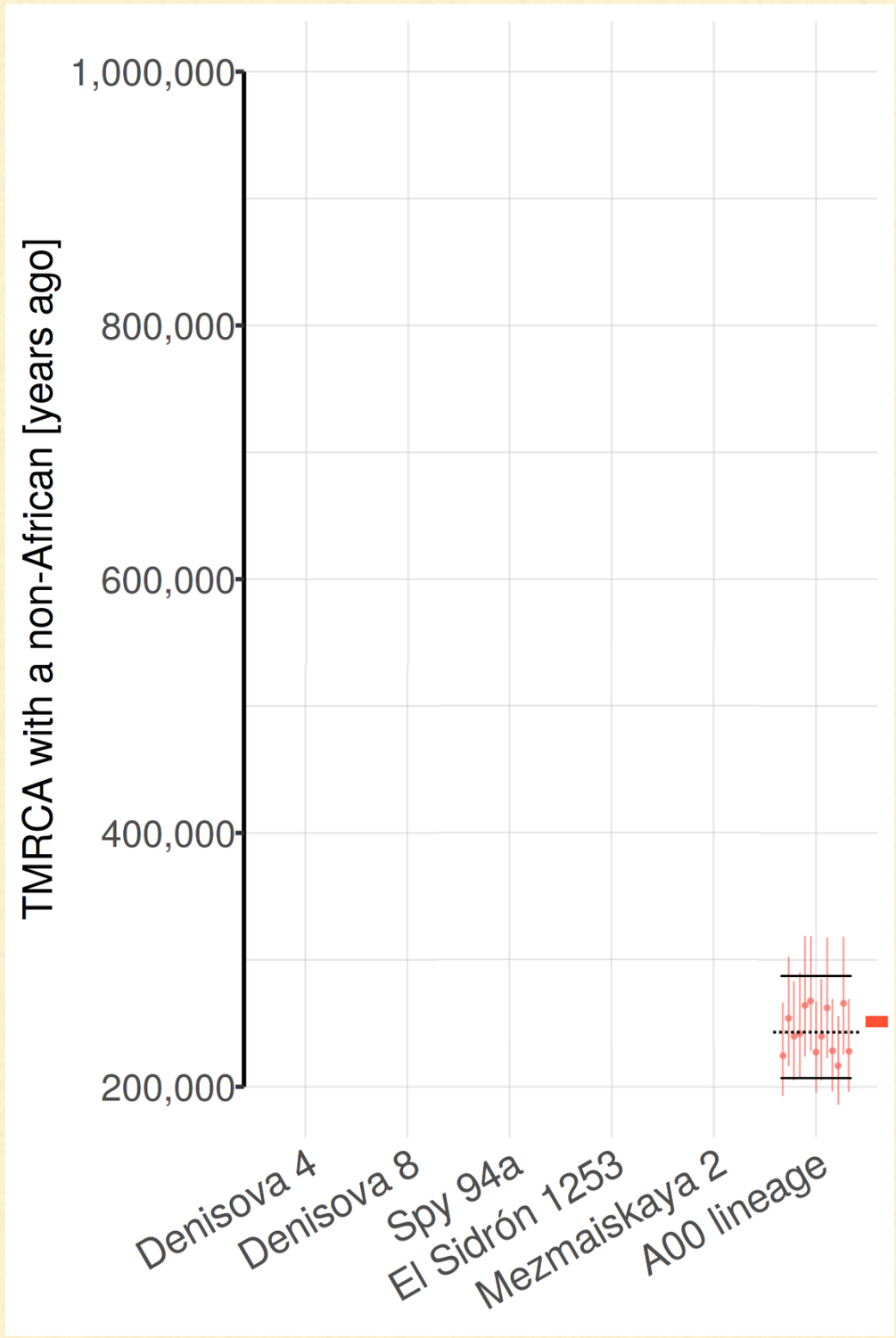

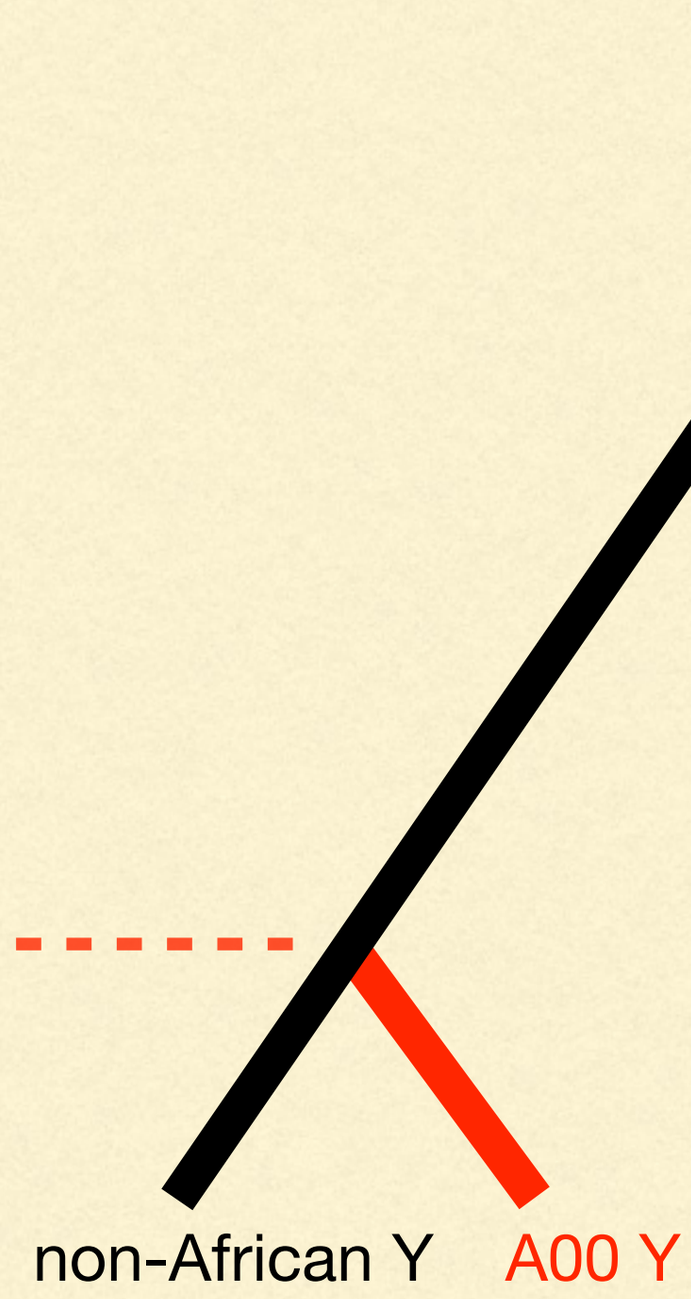

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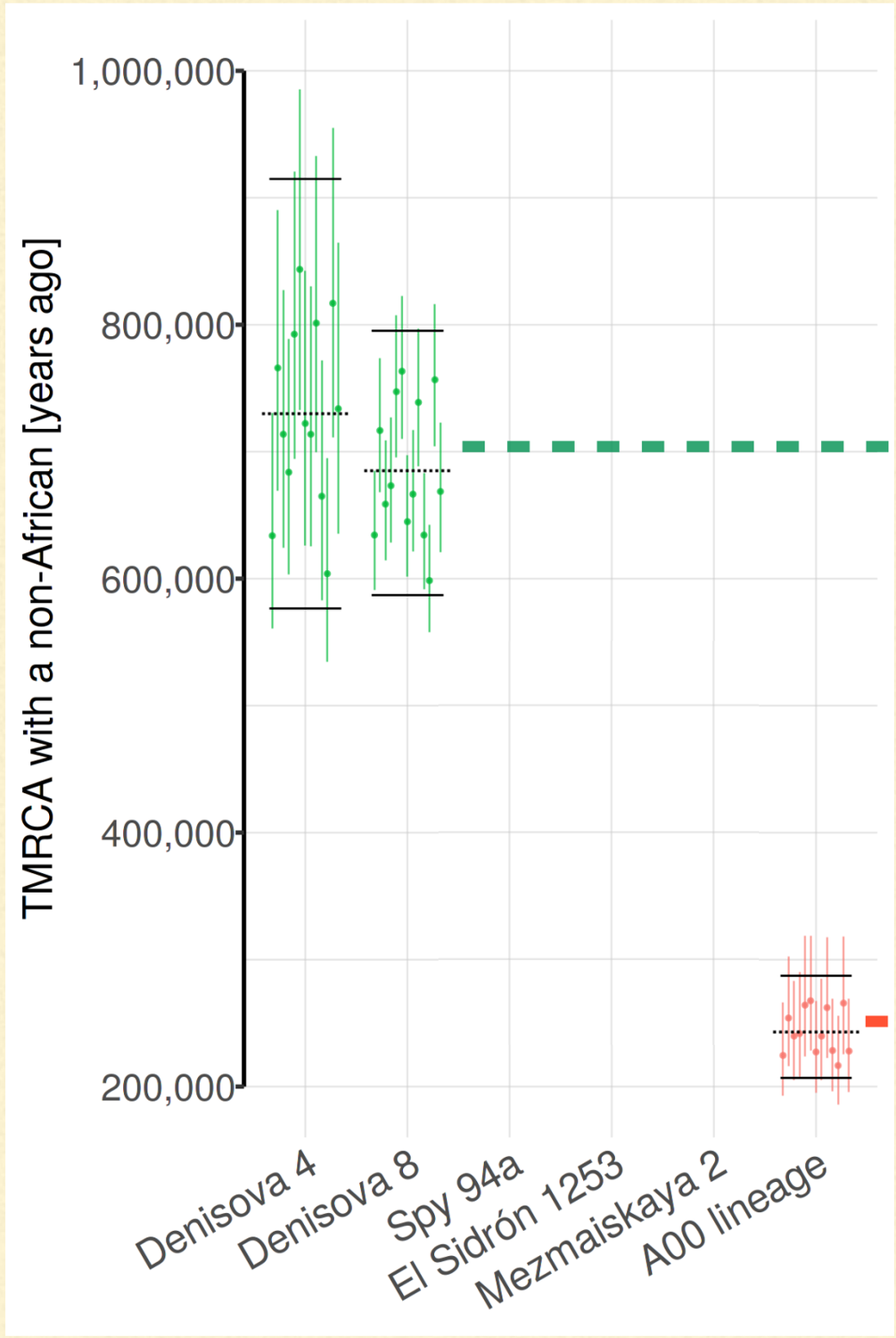

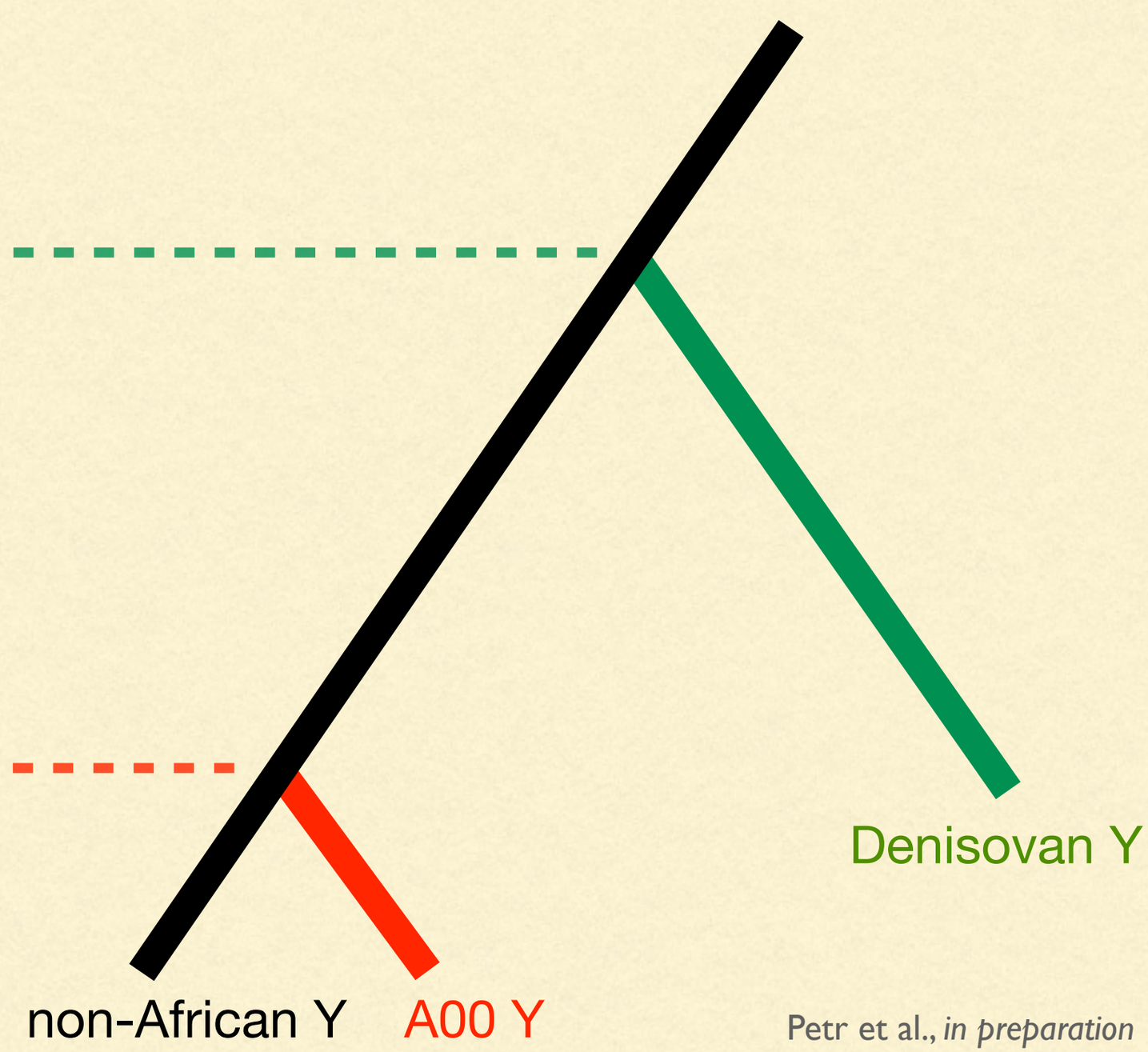

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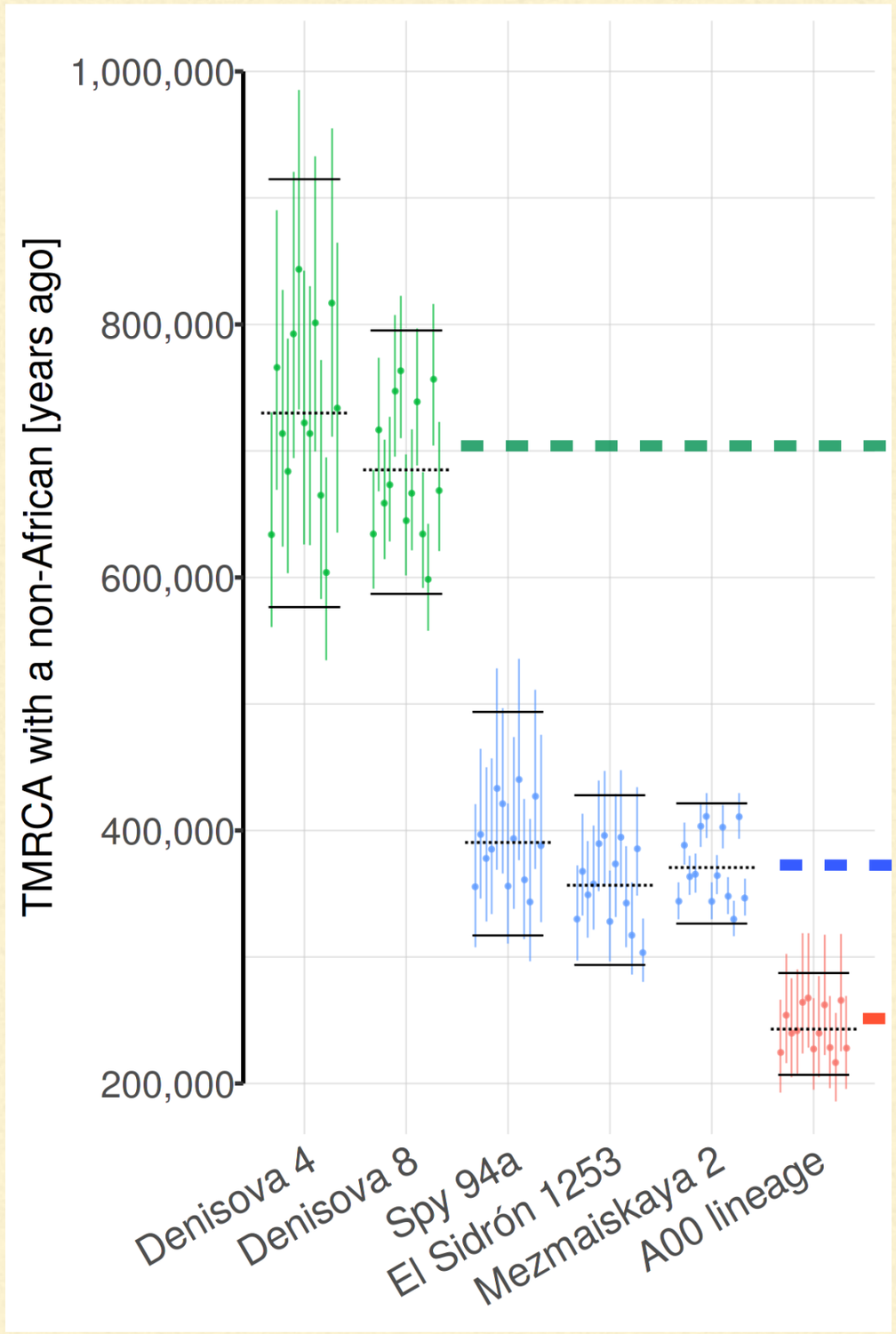
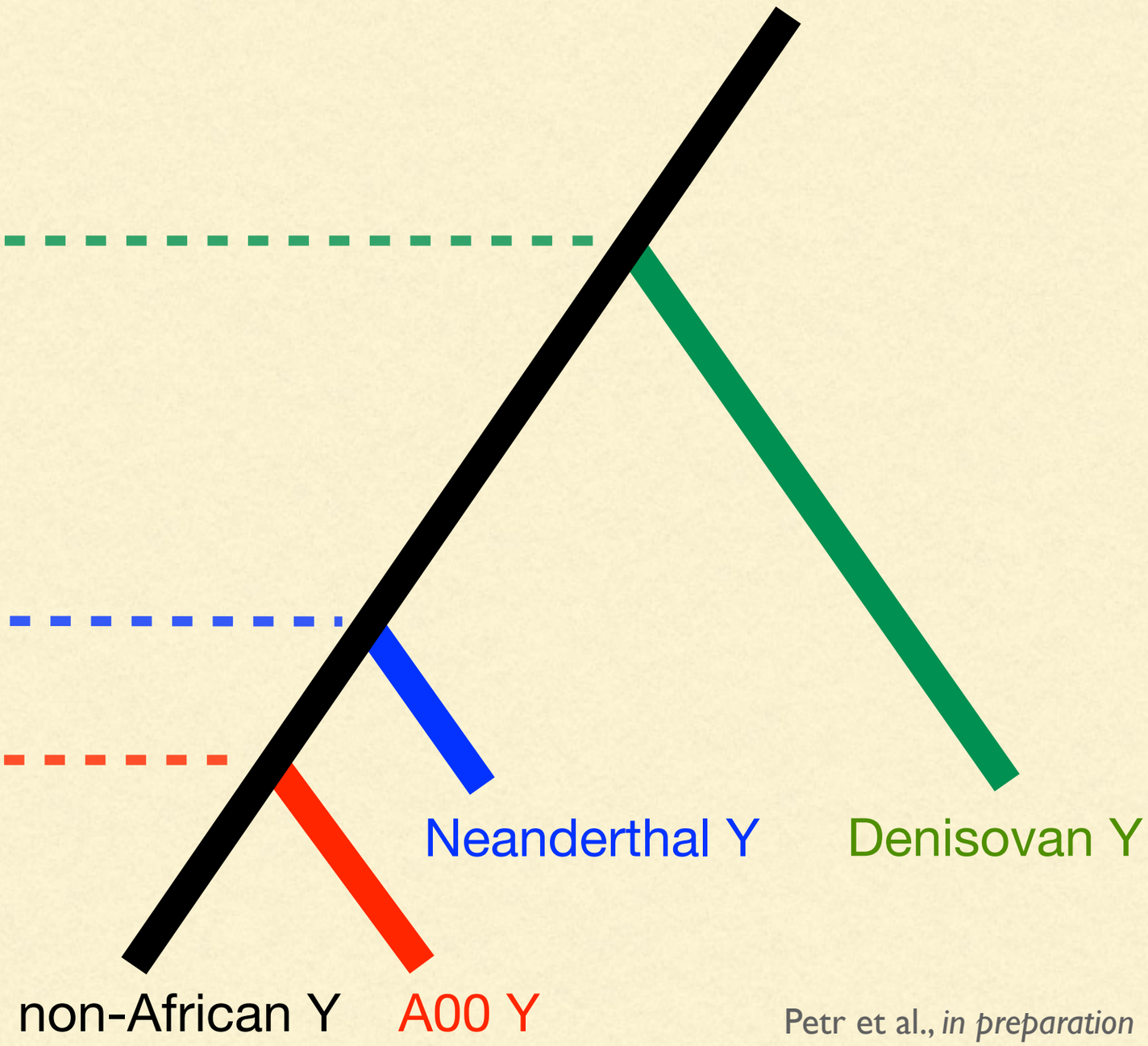
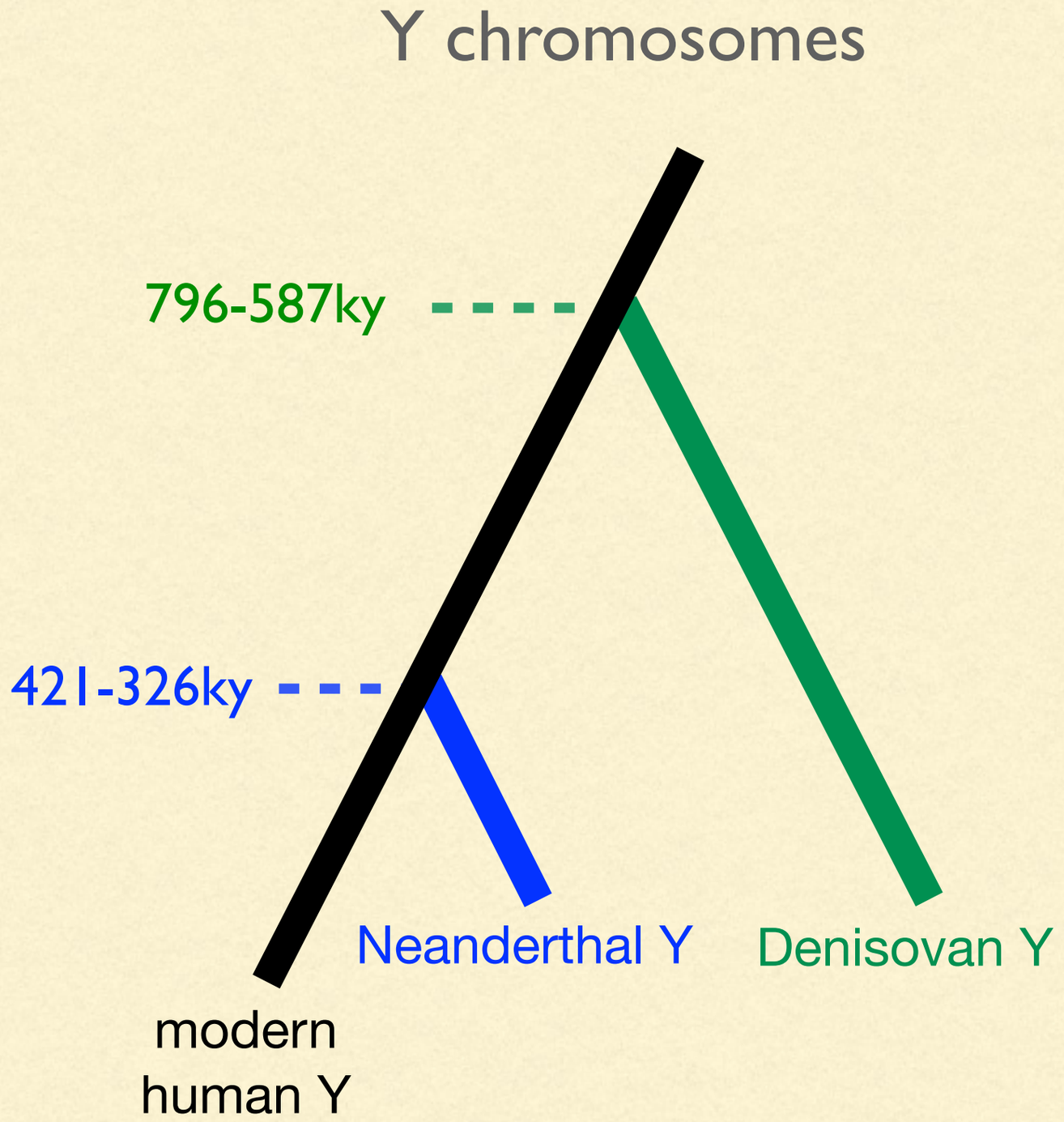




Petr et al., *in preparation*

REPLACEMENT OF NEANDERTHAL Y CHROMOSOMES?



Petr et al., *in preparation*

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autosomal DNA

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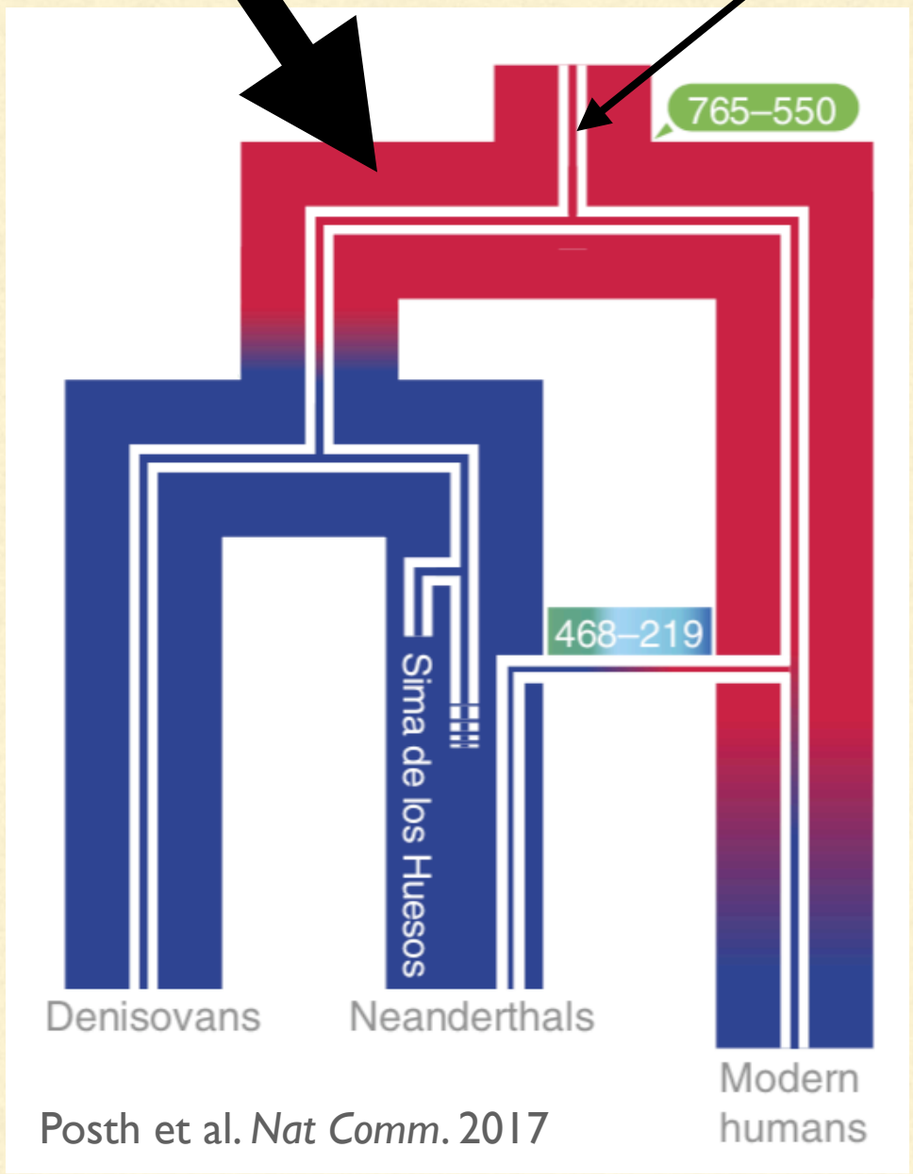
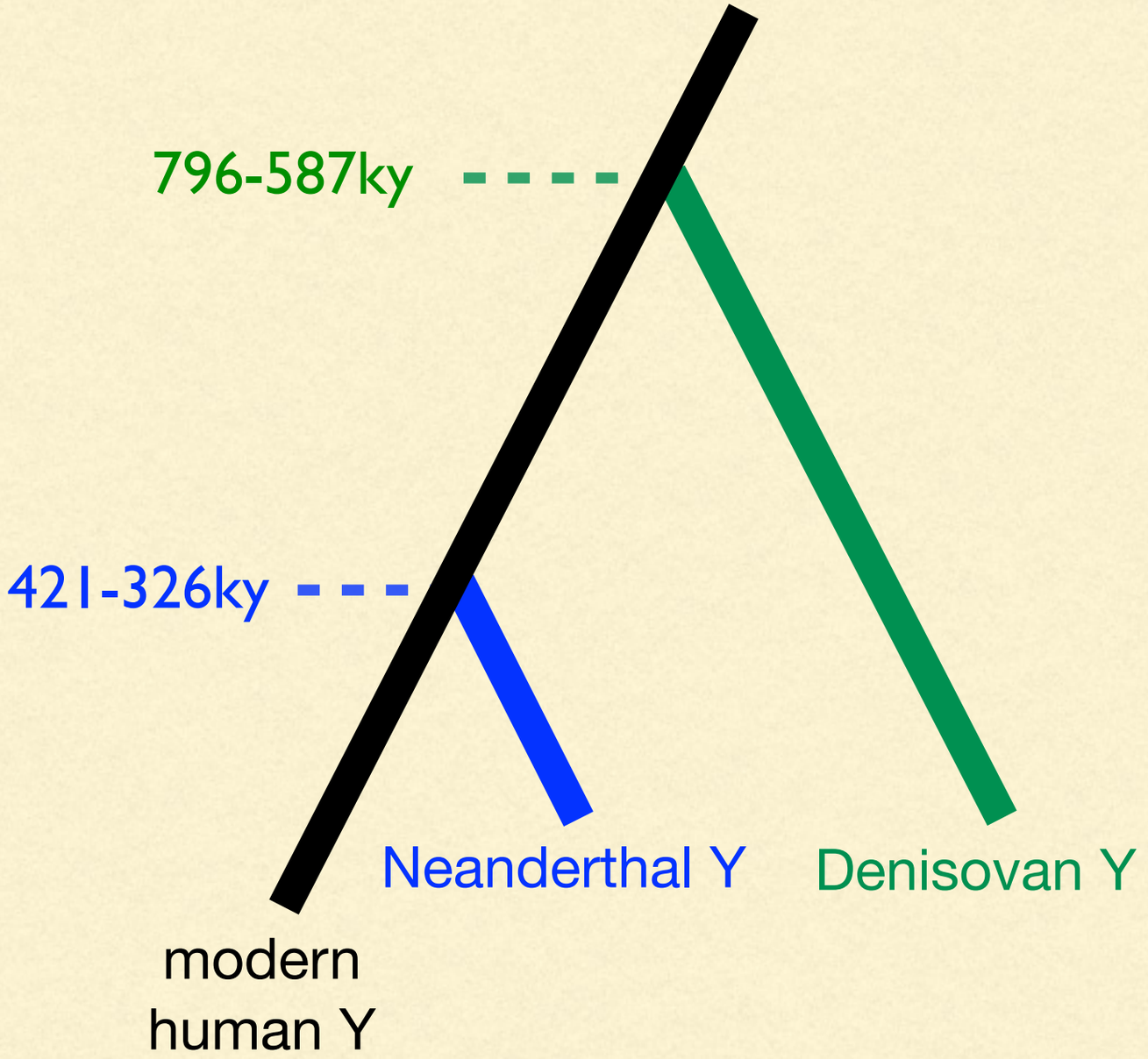


Posth et al. *Nat Comm*. 2017

Y chromosomes



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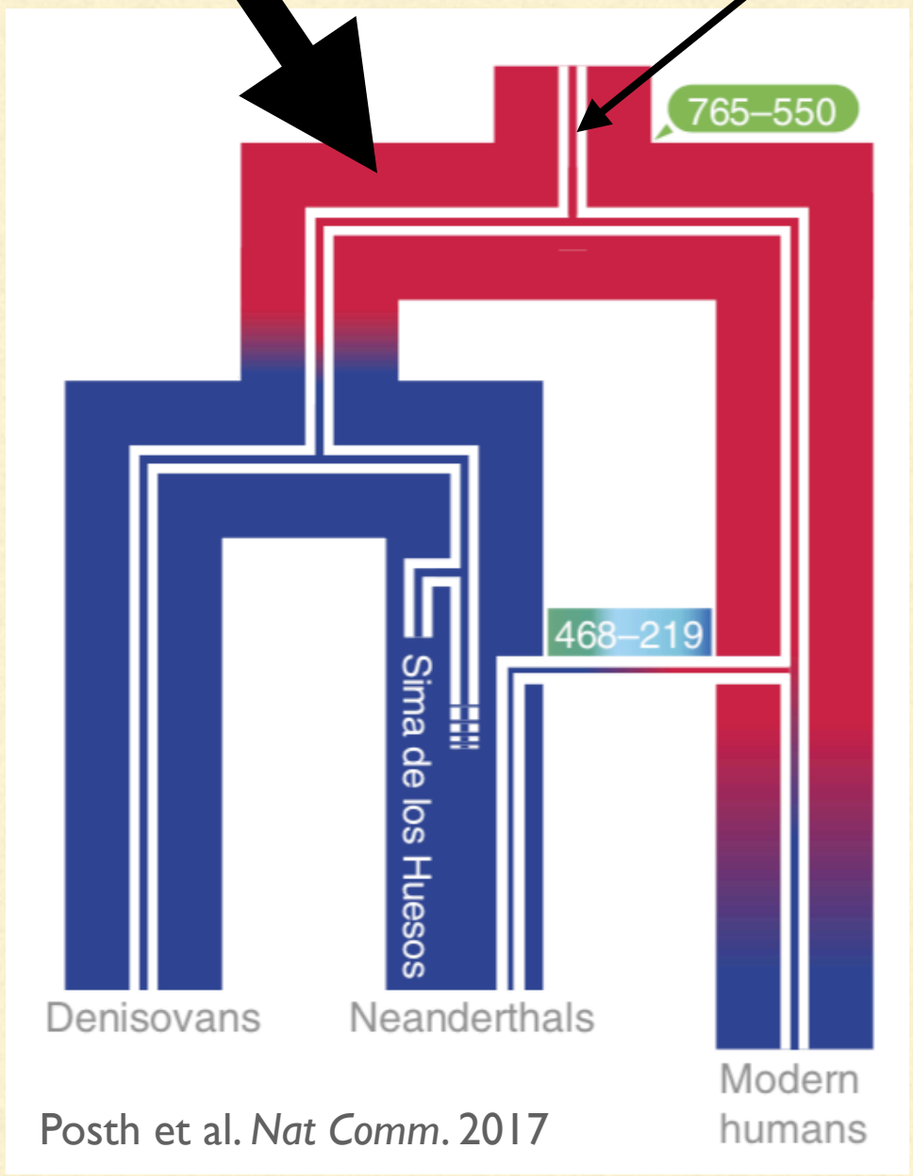

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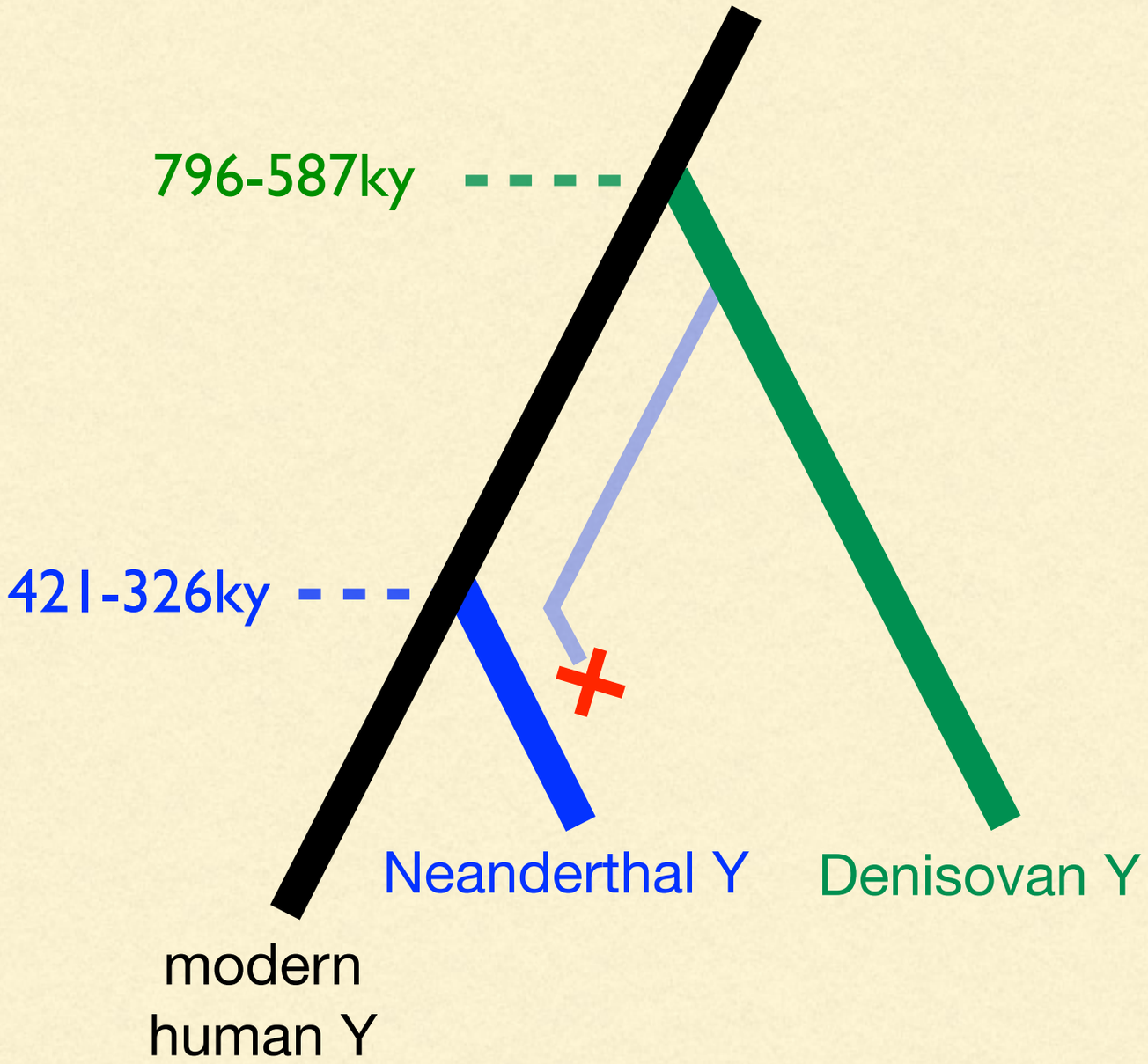

Petr et al., *in preparation*

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basic probability theory:

P[**mtDNA** replacement] = 3%
P[**Y chromosome** replacement] = 3%

P[**both** mtDNA **and** Y replaced] = 3% × 3% → **0.09%**

POSSIBLE SOLUTION:

INTROGRESSION NOT NEUTRAL

Petr et al. 2019. *PNAS*.

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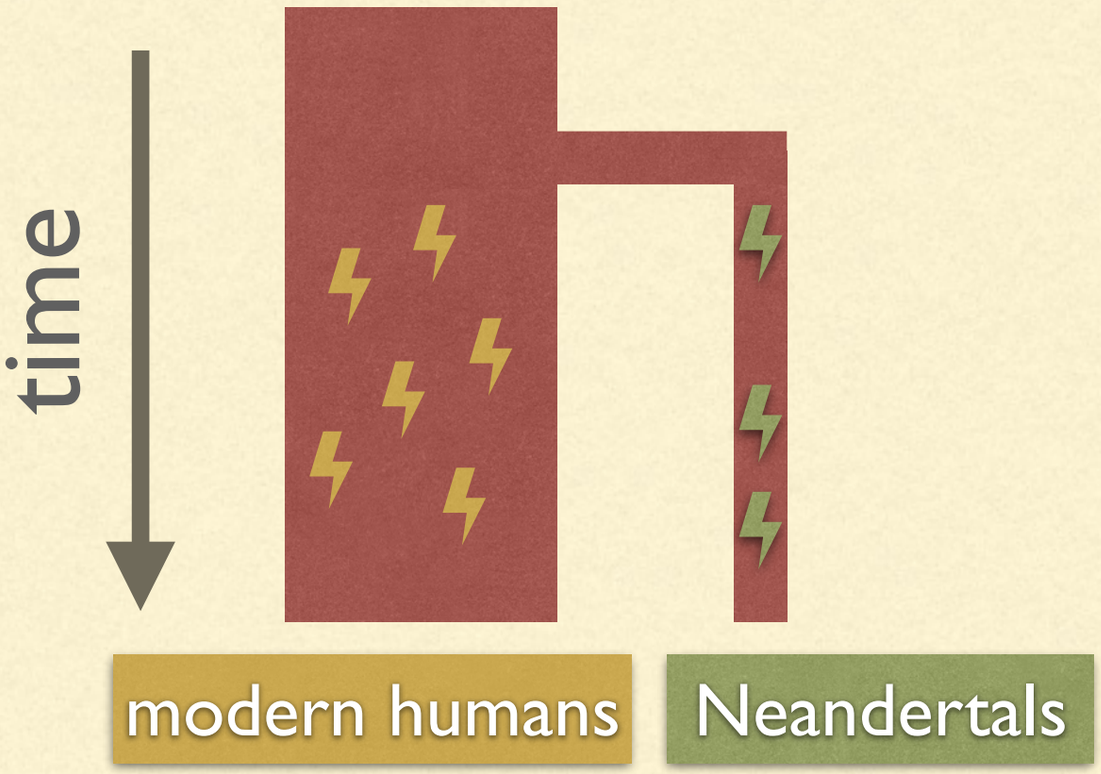
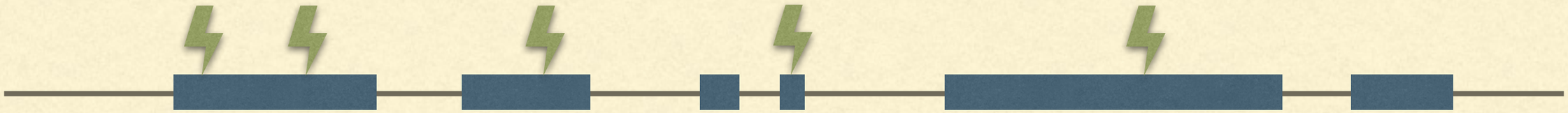
- deleterious mutations





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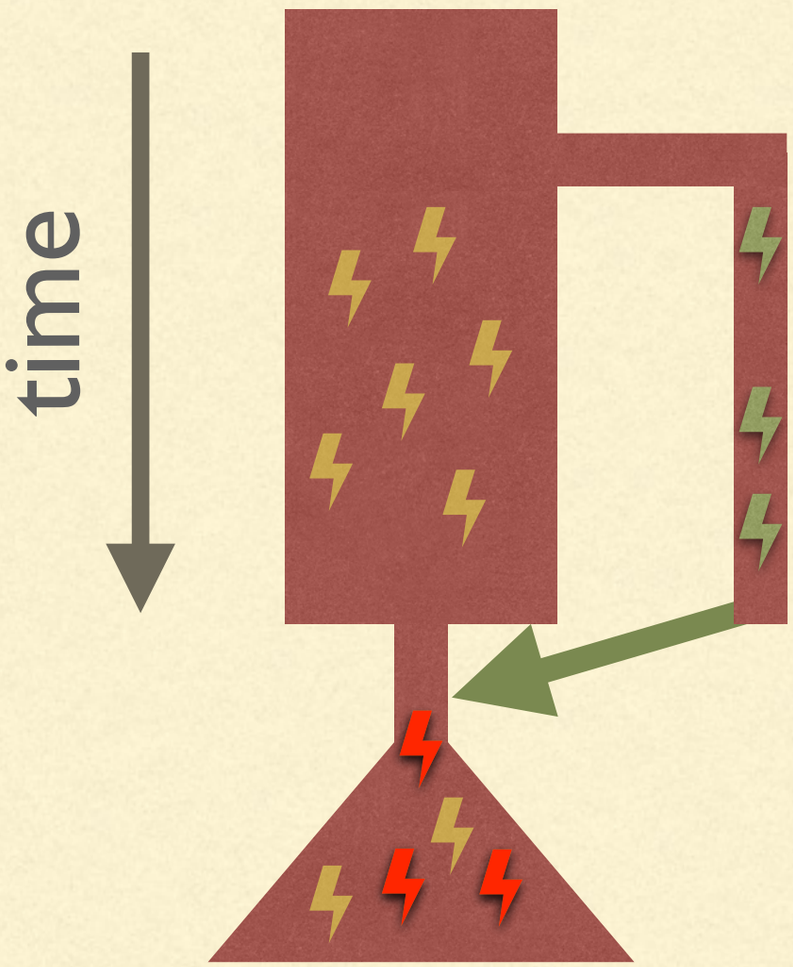

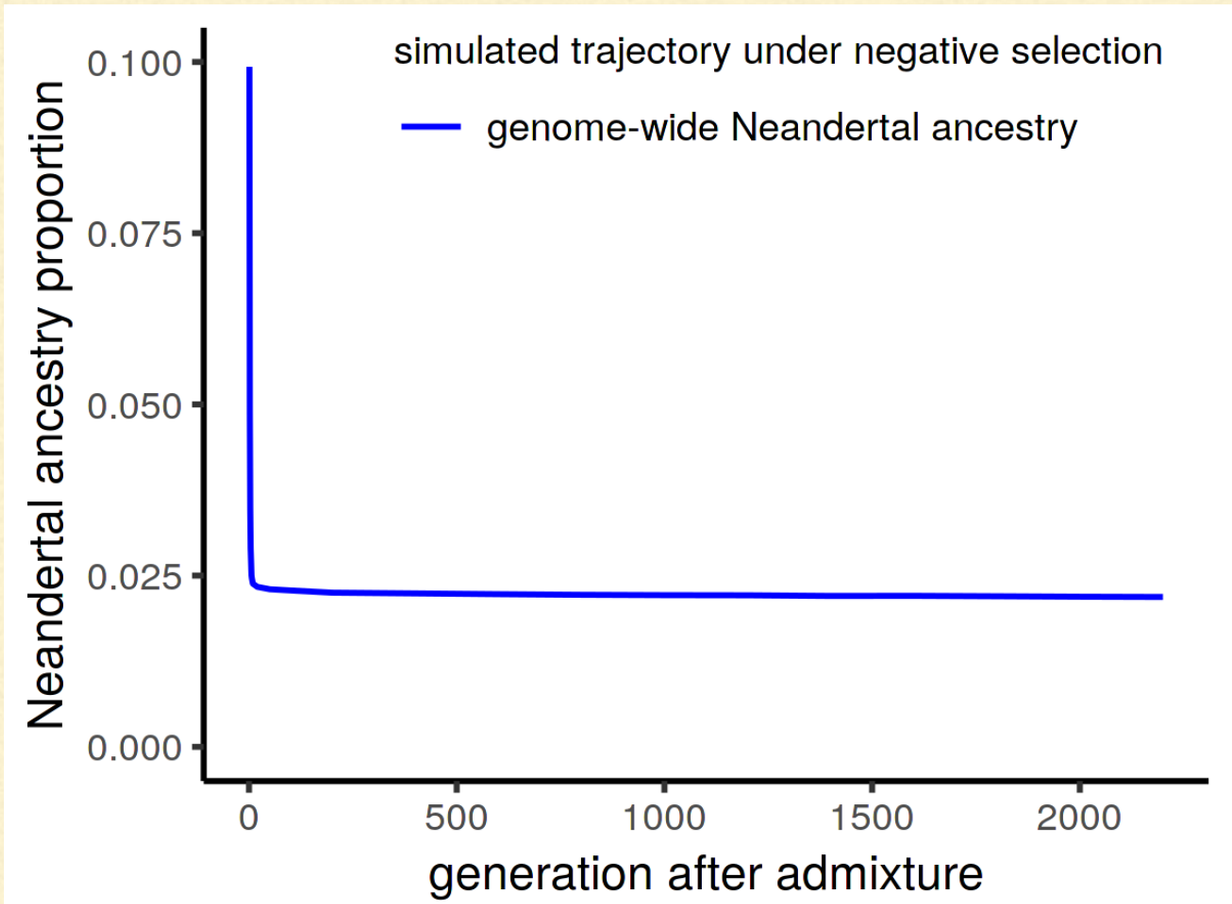

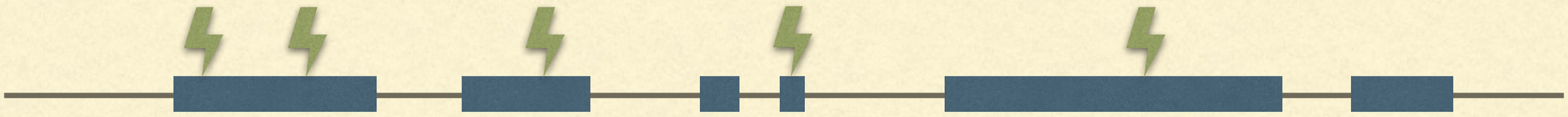

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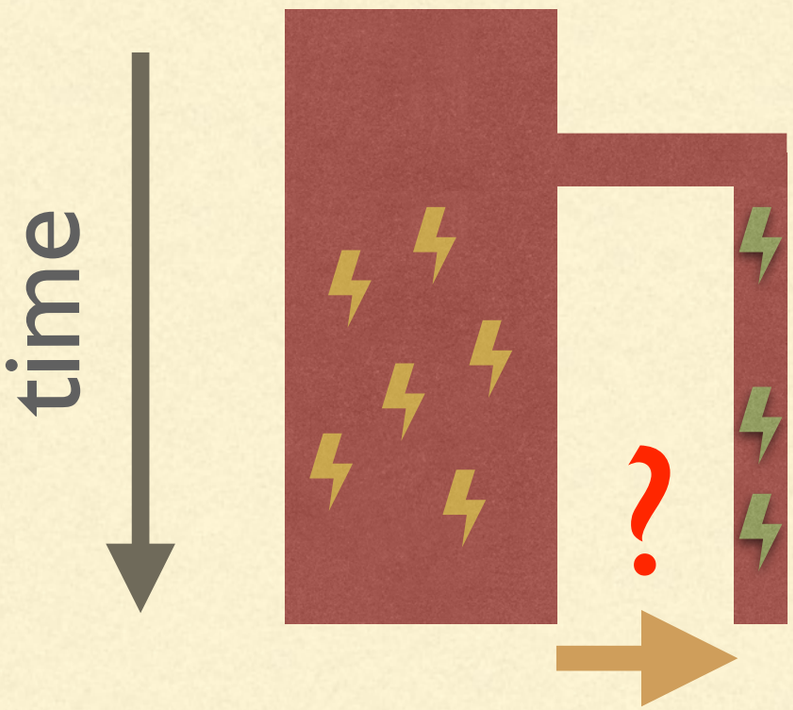
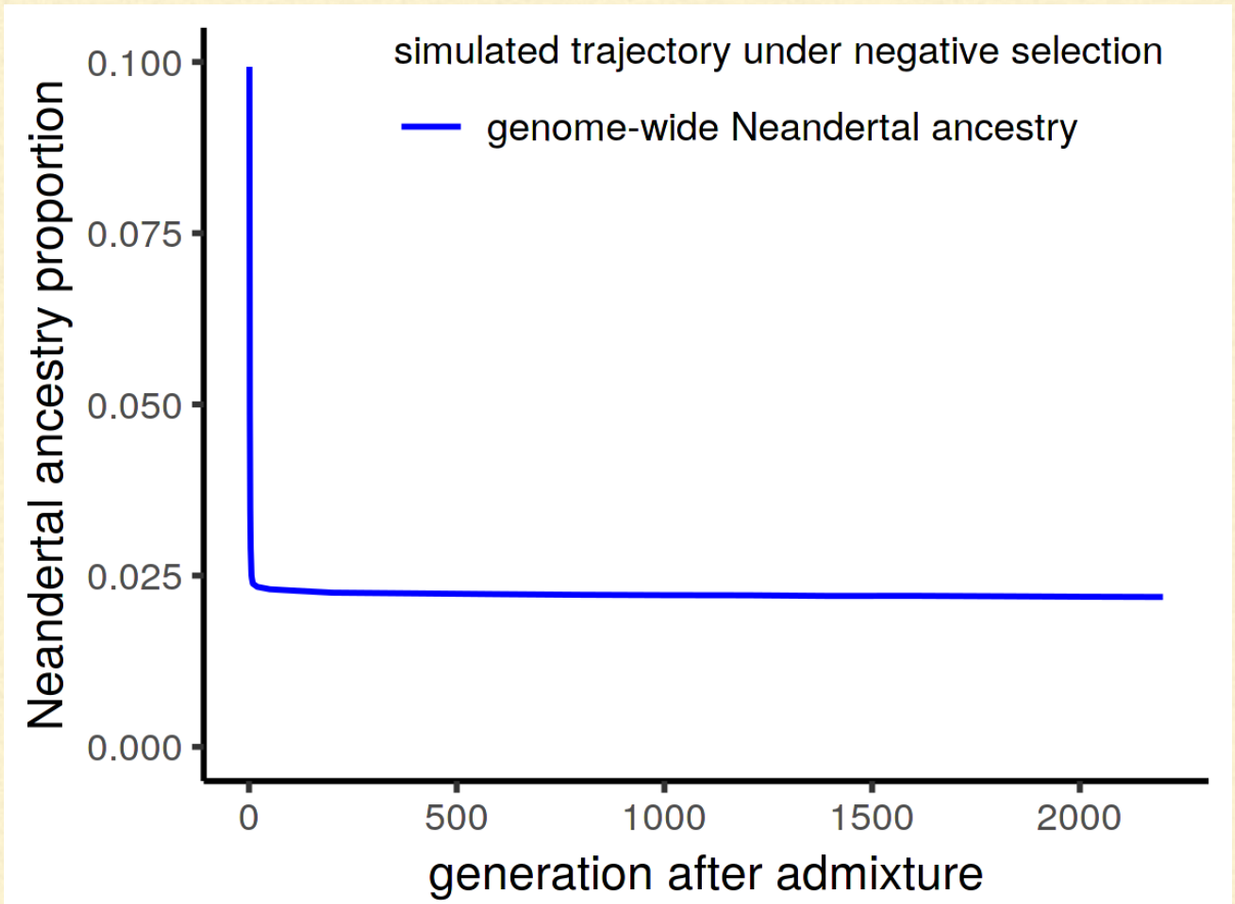
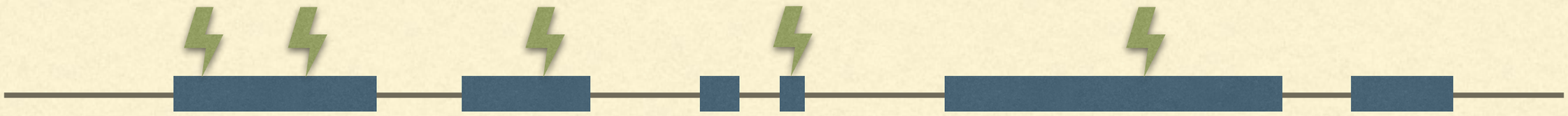
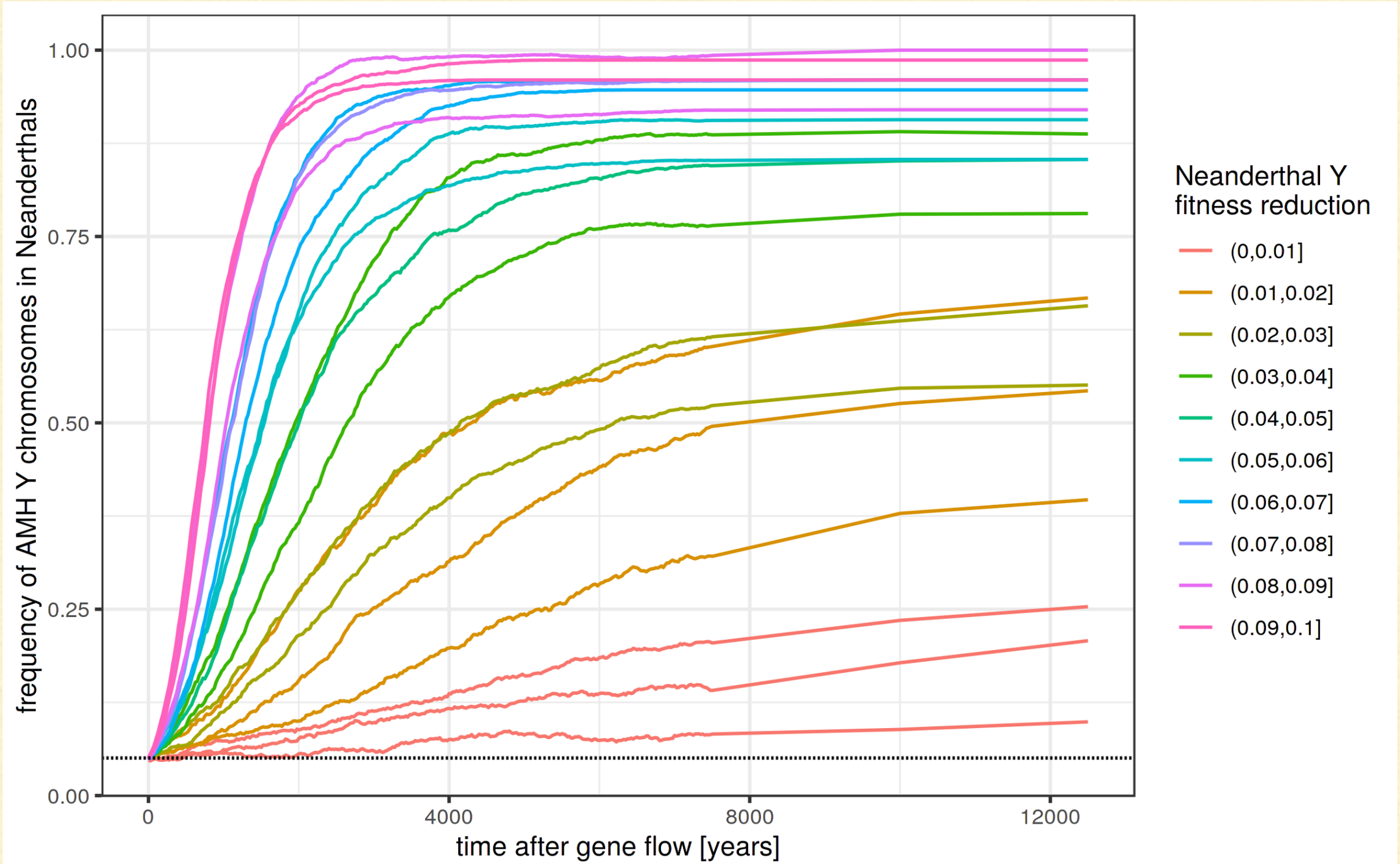






Petr et al. 2019. *PNAS*.

LOWER FITNESS OF NEANDERTHAL Y STRONGLY FAVOURS MODERN HUMAN Y!



Petr et al., *in preparation*

CONCLUSIONS

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- Denisovan and Neanderthal Y phylogenies discordant:
  - AMH-Denisovan TMRCA ~700 kya
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- Y of Sima de los Huesos should be “Denisovan-like”.
- Functional implications for future archaic Y studies.

Svante Pääbo

Mateja Hajdinjak

Janet Kelso

Qiaomei Fu, Elena Essel, Hélène Rougier, Isabelle Crevecoeur, Patrick Semal, Liubov V. Golovanova, Vladimir B. Doronichev, Carles Lalueza-Fox, Marco de la Rasilla, Antonio Rosas, Michael V. Shunkov, Maxim B. Kozlikin, Anatoli P. Derevianko, Matthias Meyer, Benjamin Vernot

?

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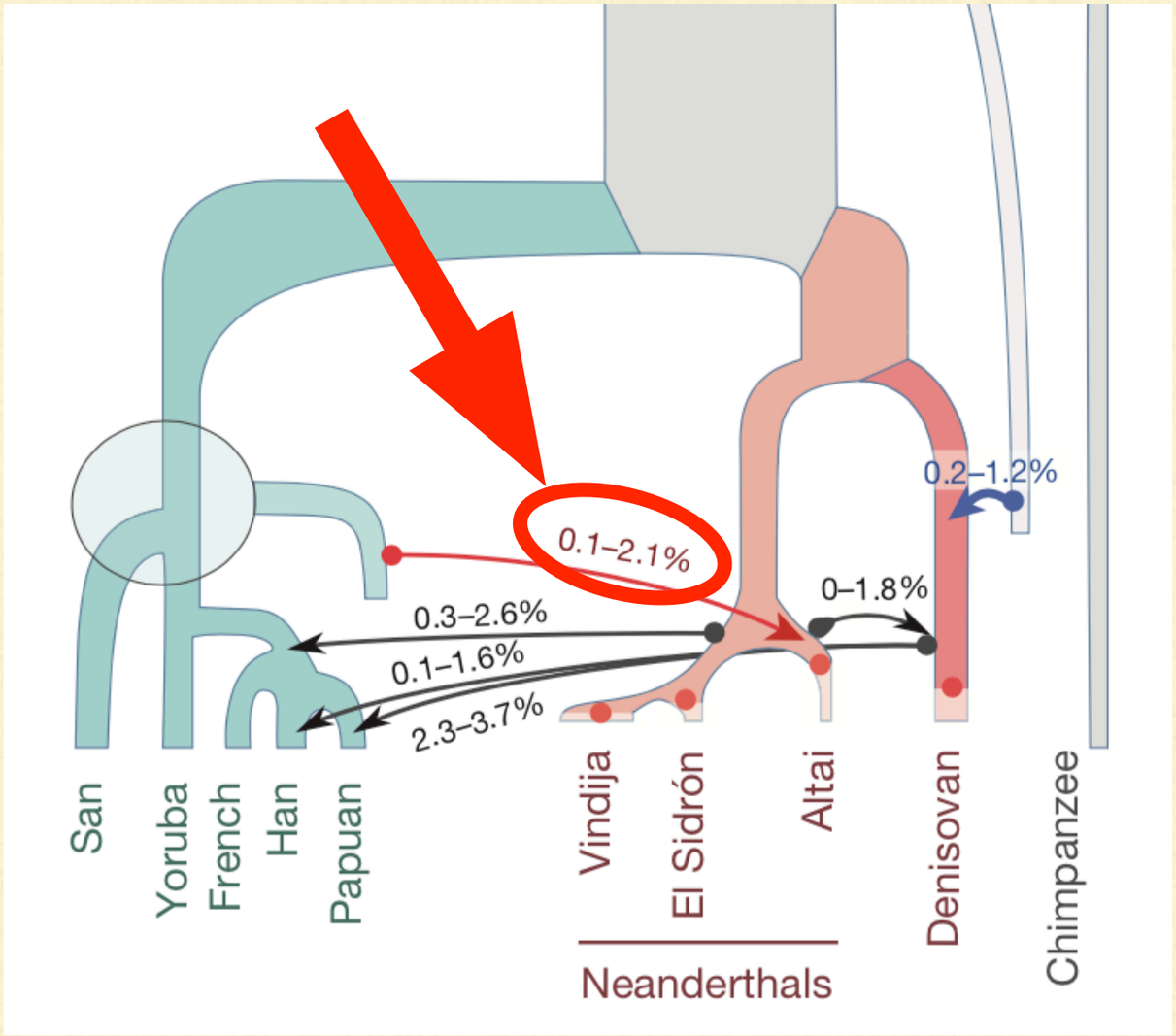
mp@bodkan.net

Kuhlwilm et al. *Nature*. 2016



Hubisz et al. *bioRxiv*. 2019

- at least **3%** gene flow
- **200-300 kya**

“WHOLE CHROMOSOME CAPTURE”

goal: capture and sequence ~6.9 Mb of the entire Y

human Y
reference
sequence

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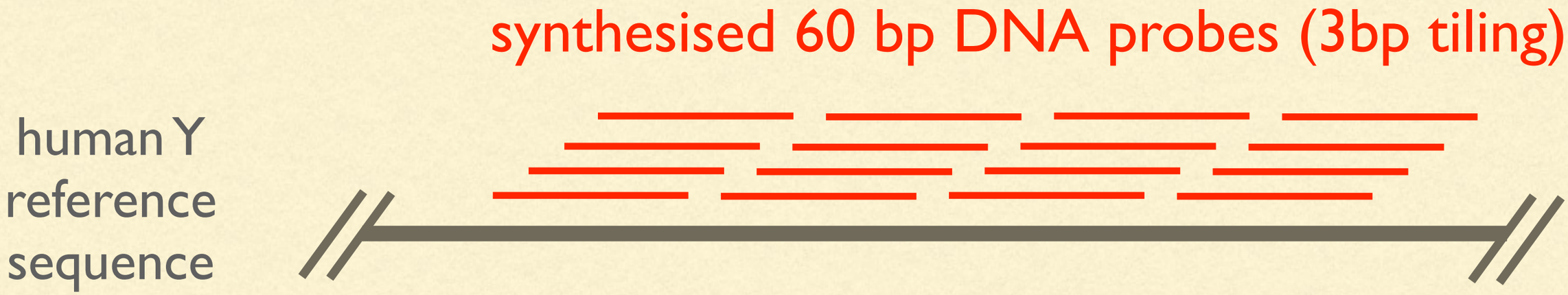
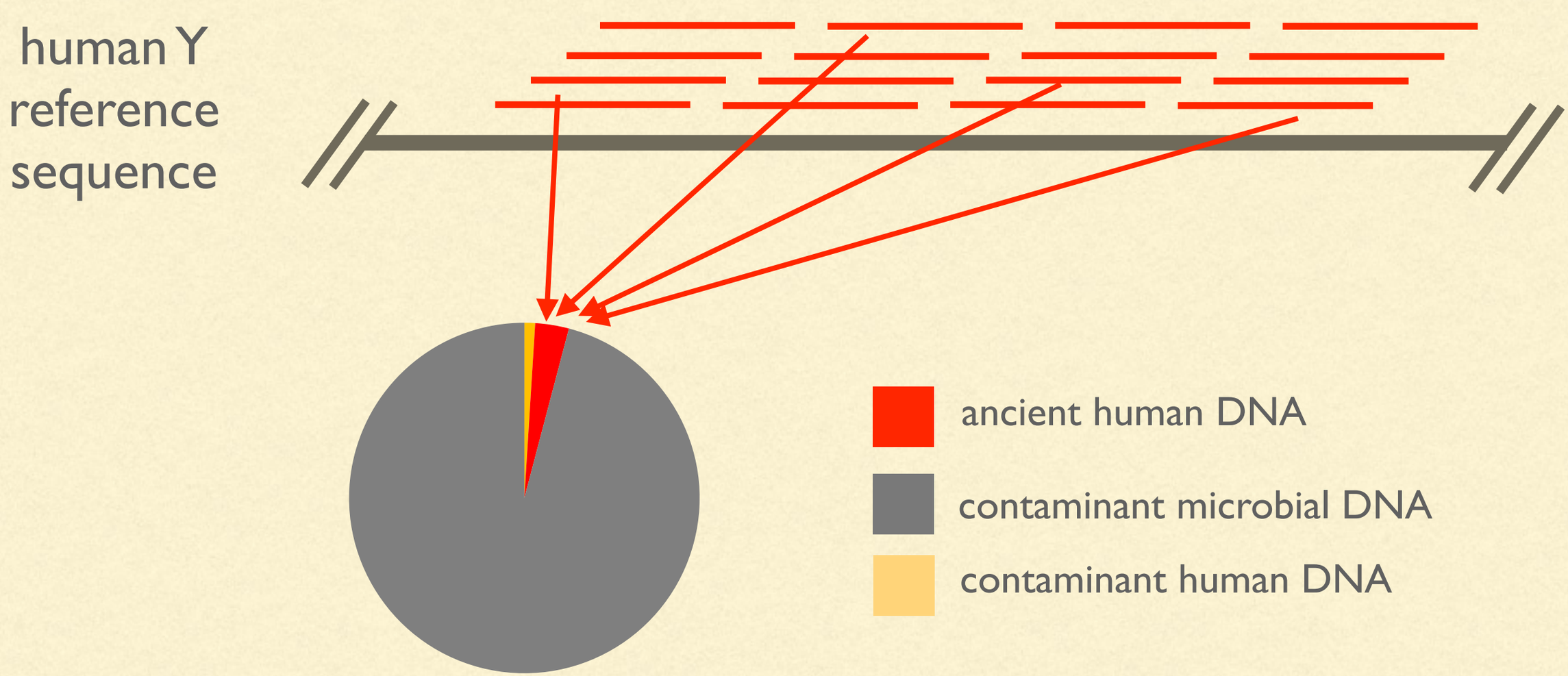


“WHOLE CHROMOSOME CAPTURE”

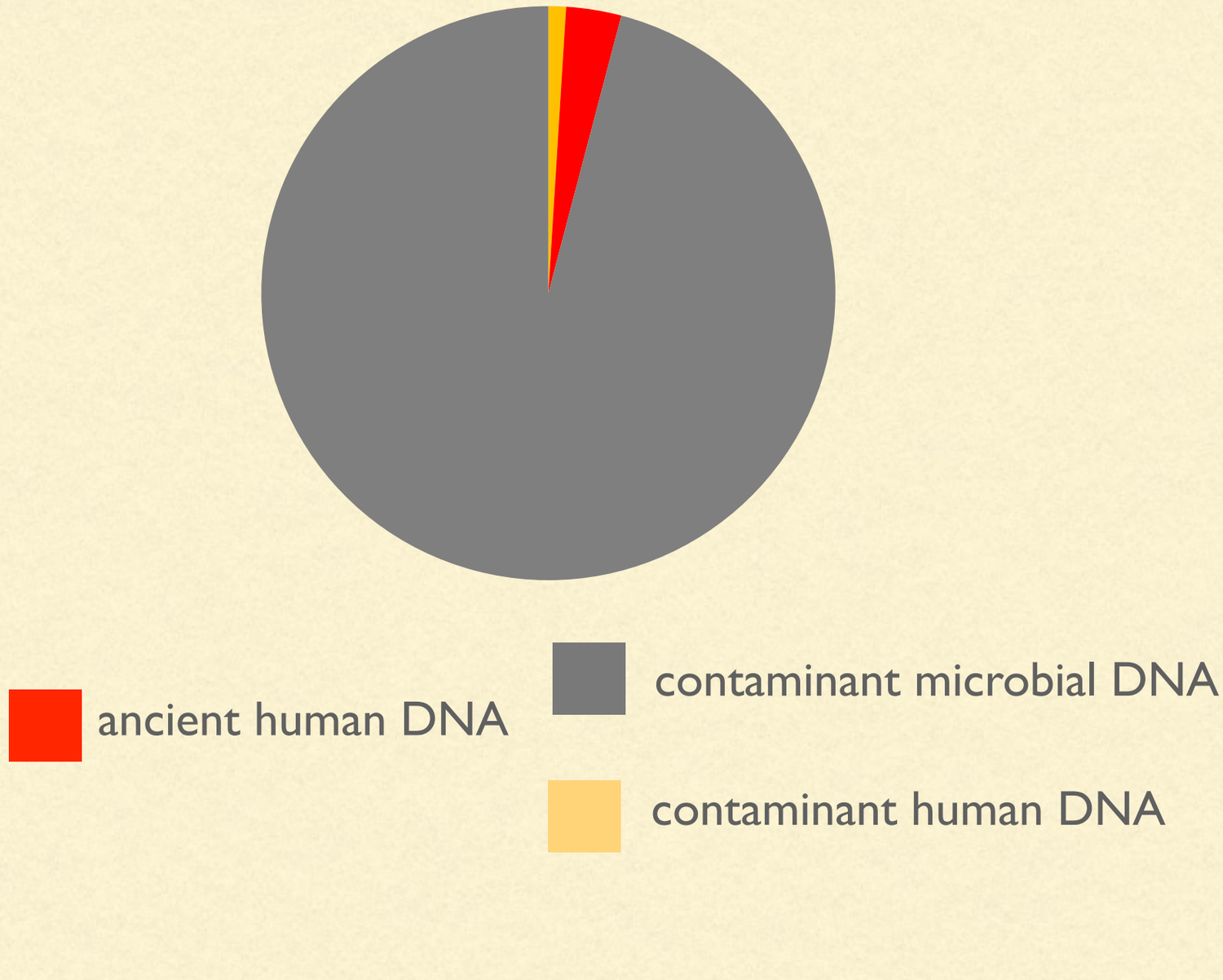
goal: capture and sequence ~6.9 Mb of the entire Y

synthesised 60 bp DNA probes (3bp tiling)



ANCIENT DNA CHALLENGES...

Total DNA extracted from a tooth/bone:

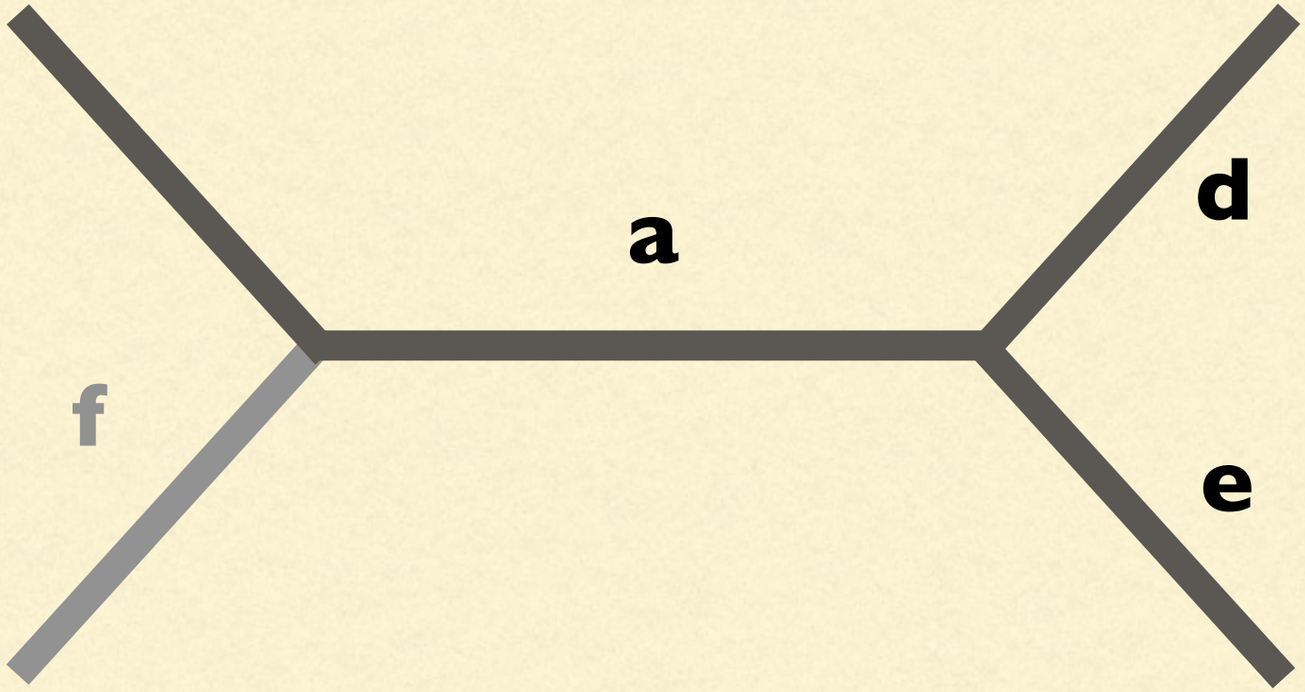


ESTIMATING TMRCA

WITH MODERN HUMAN Y CHROMOSOMES

chimpanzee

a set of non-African Y chromosomes

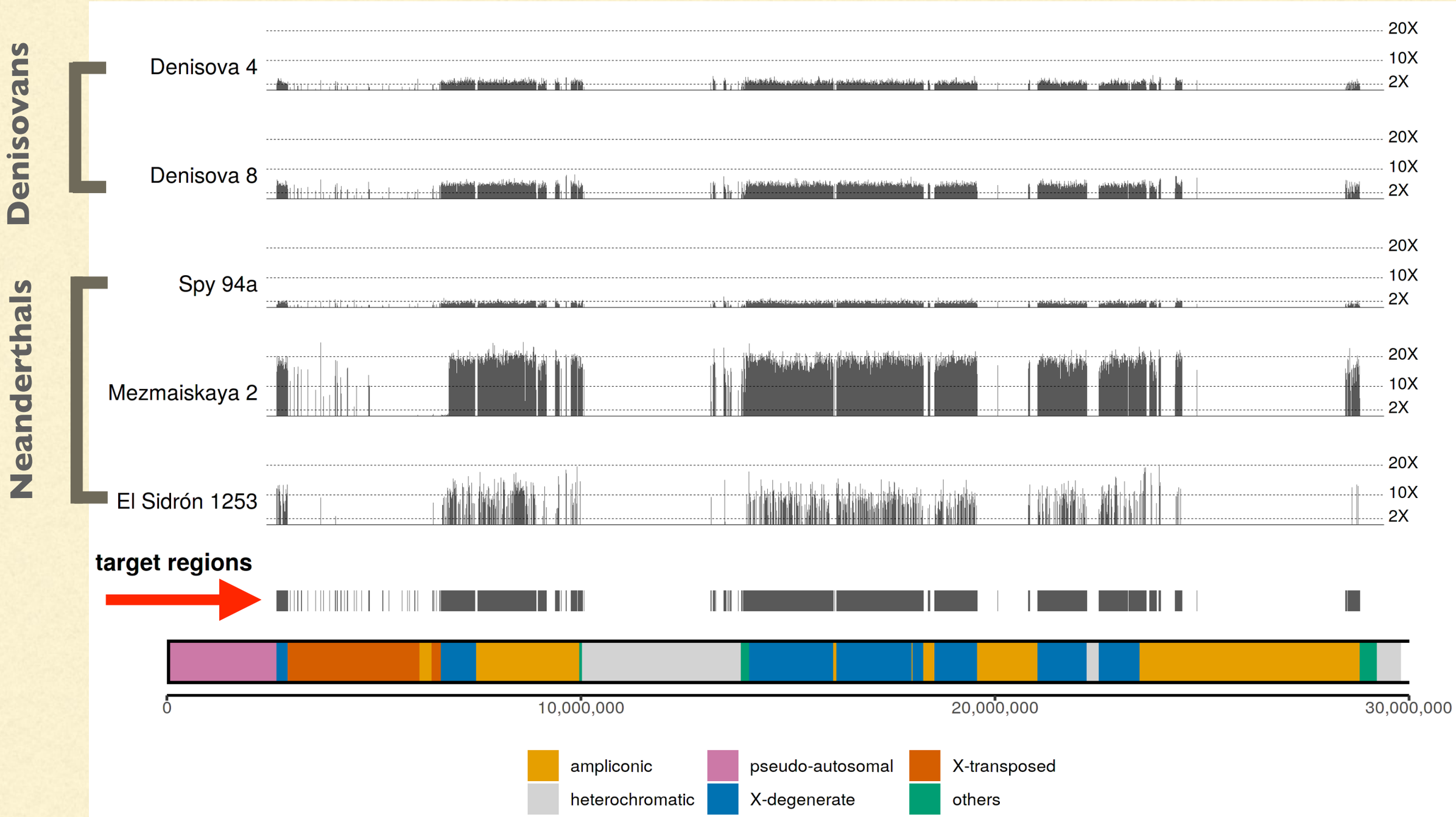


Denisovan or Neanderthal Y

African Y lineage A00

Mendez et al. *AJHG*. 2016.

COVERAGE OF CAPTURED
Y CHROMOSOMES



Petr et al., *in preparation*

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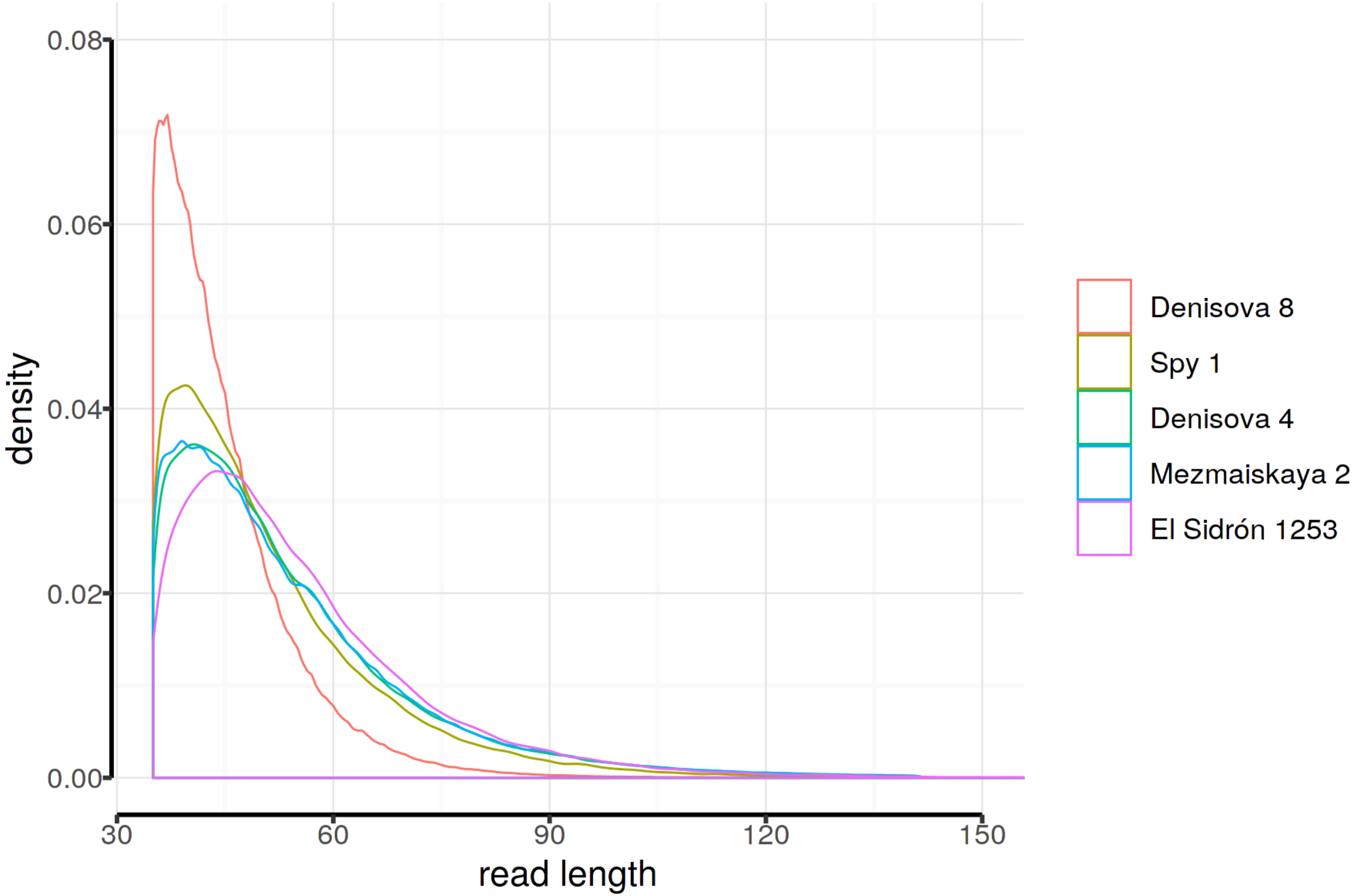
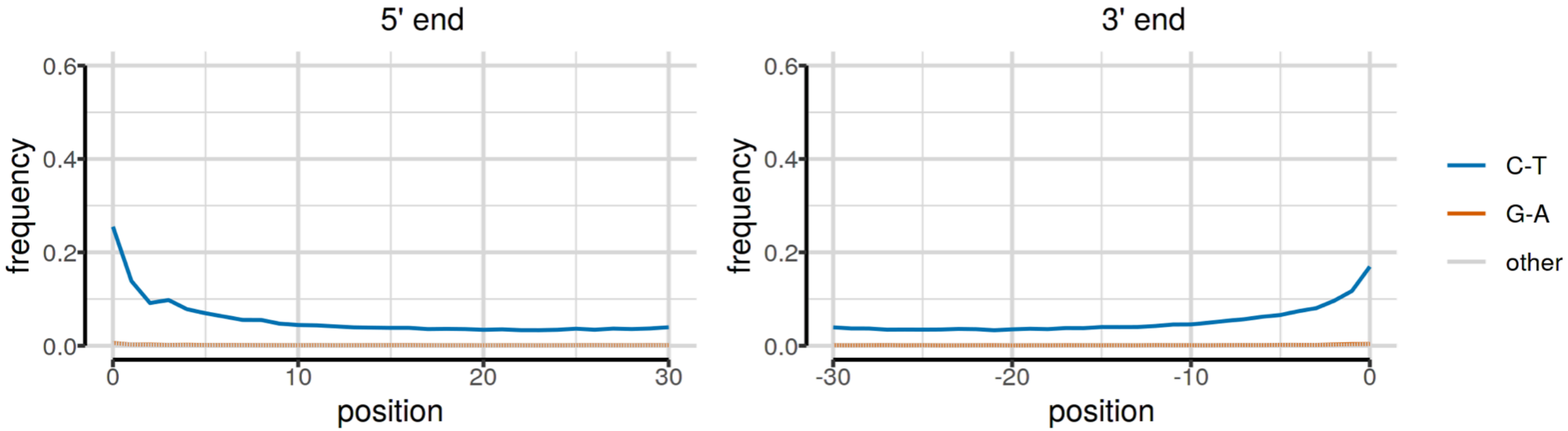
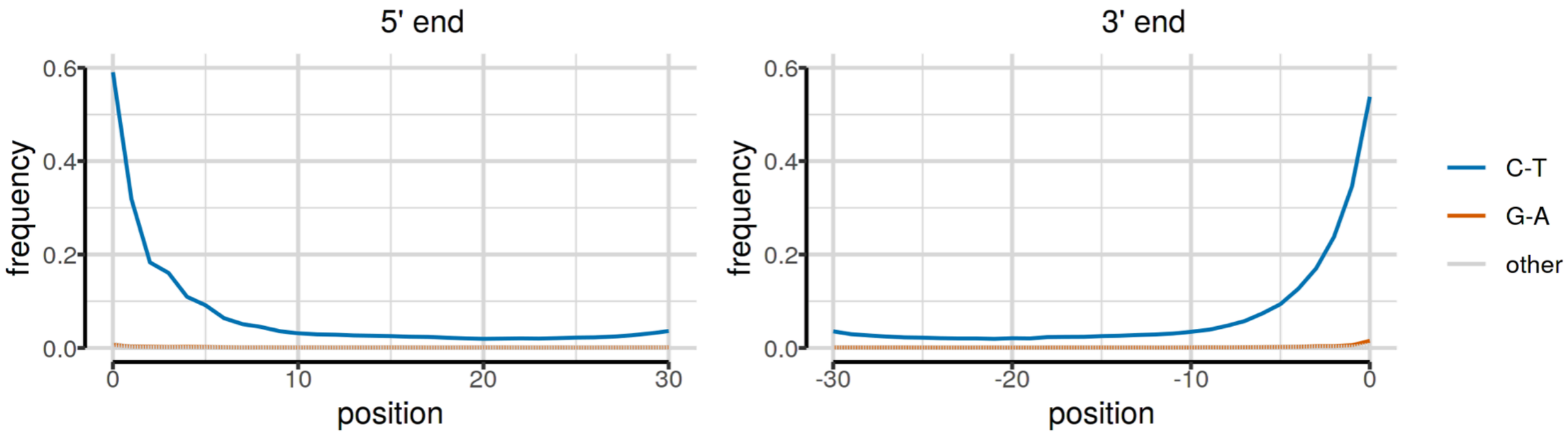

0.08
0.06
0.04
0.02
0.00
density
30
60
90
120
150
read length
Denisova 8
Spy 1
Denisova 4
Mezmaiskaya 2
El Sidrón 1253

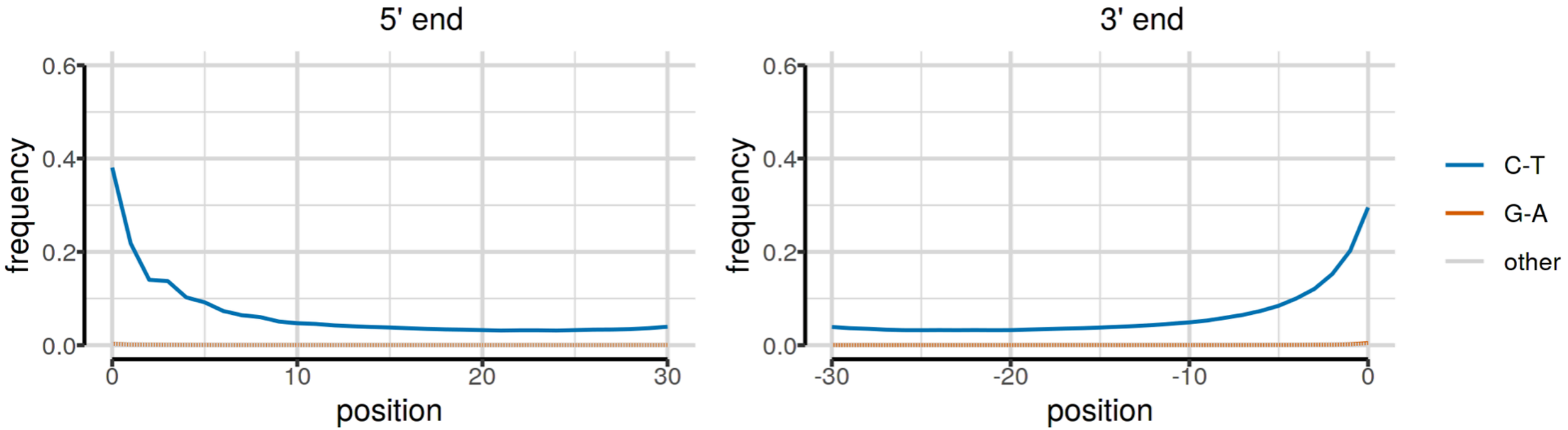
Denisova 4



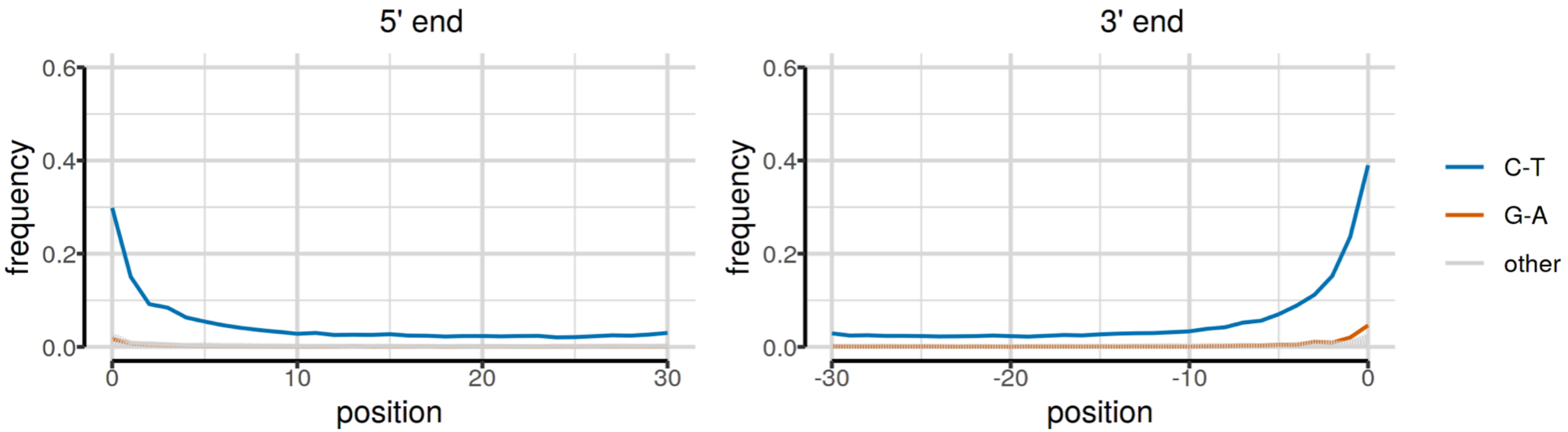
Denisova 8

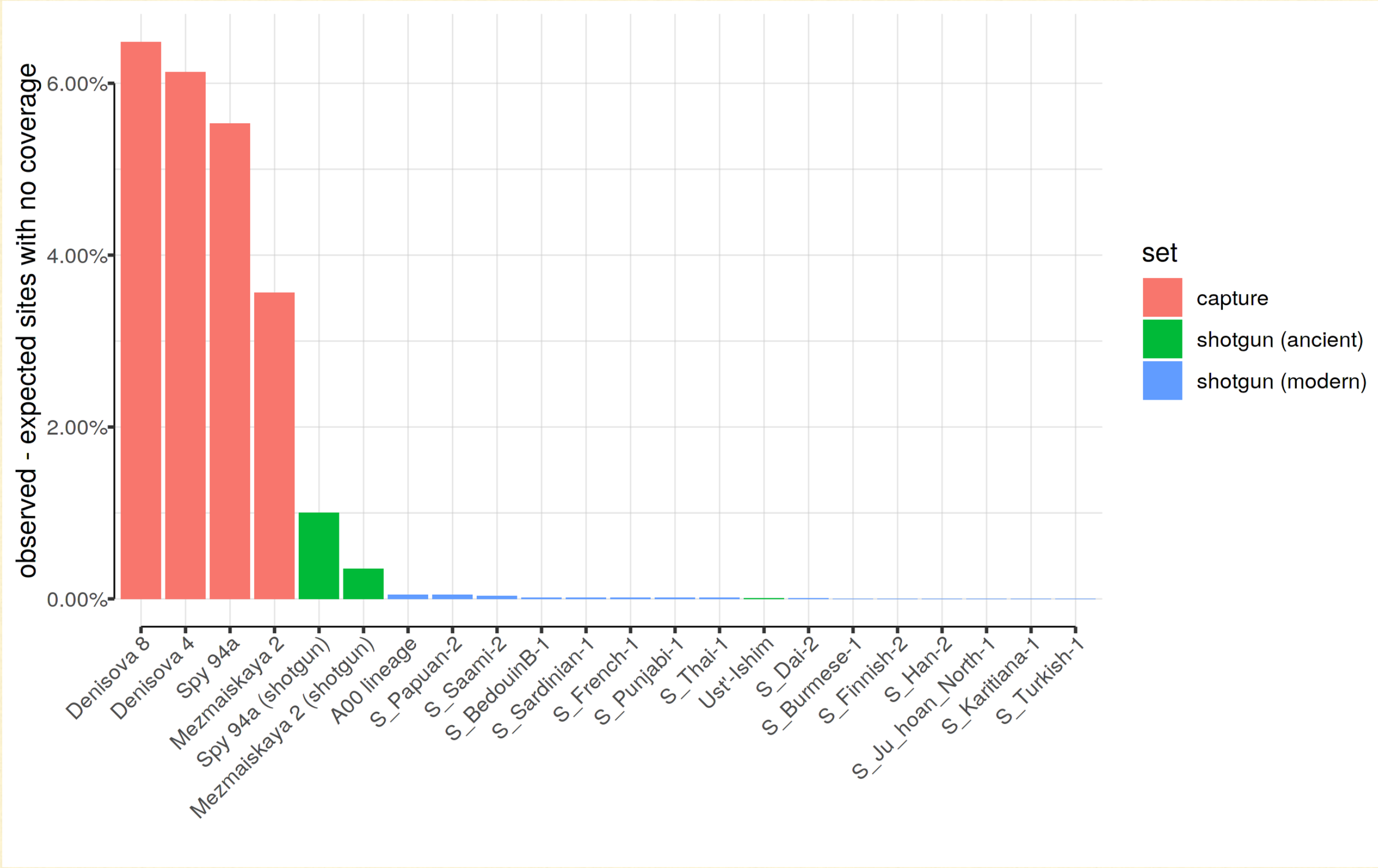


Mezmaiskaya 2



Spy 94a




observed - expected sites with no coverage
6.00%
4.00%
2.00%
0.00%
Denisova 8
Denisova 4
Spy 94a
Mezmaiskaya 2
Spy 94a (shotgun)
Mezmaiskaya 2 (shotgun)
A00 lineage
S_Papuan-2
S_Saami-2
S_BedouinB-1
S_Sardinian-1
S_French-1
S_Punjabi-1
S_Thai-1
Ust'-Ishim
S_Dai-2
S_Burmese-1
S_Finnish-2
S_Han-2
S_Ju_hoan_North-1
S_Karitiana-1
S_Turkish-1
set
capture
shotgun (ancient)
shotgun (modern)