Inference of ancient human demography using personal genomes

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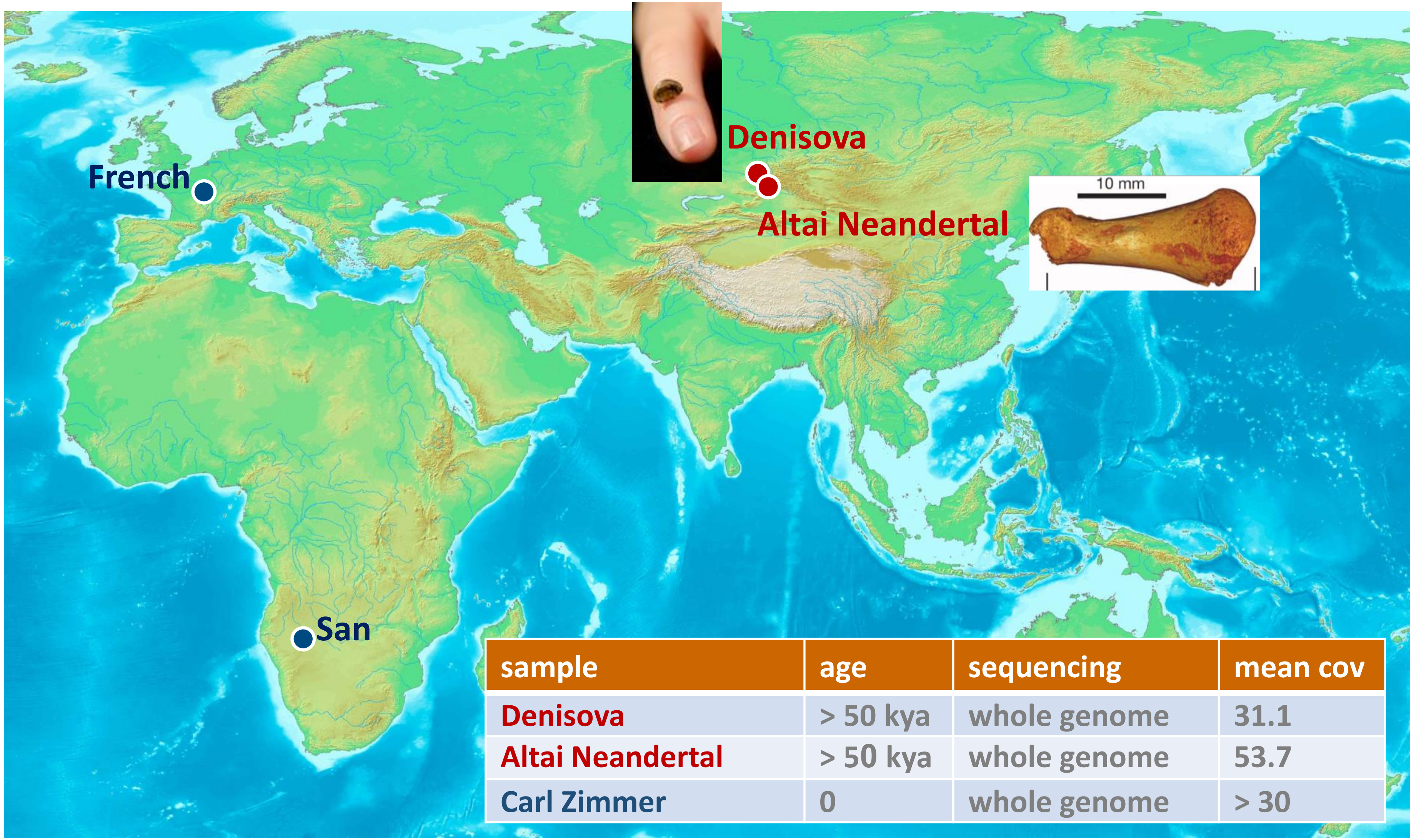
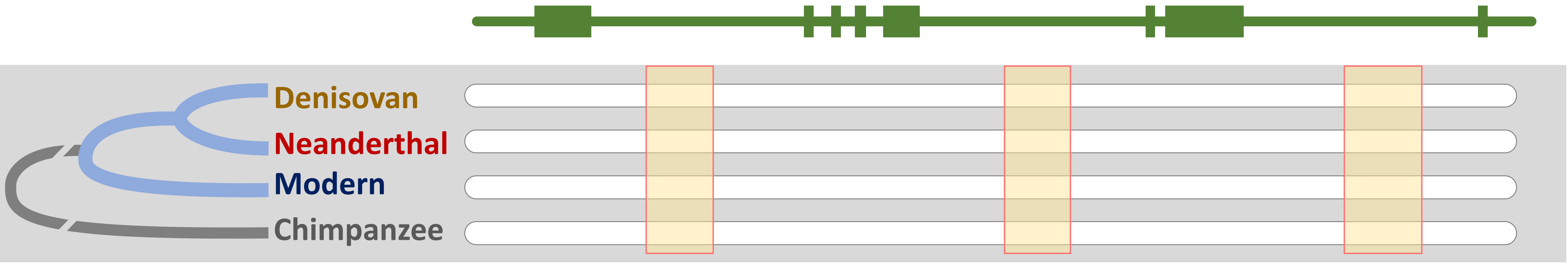
Genome sequences

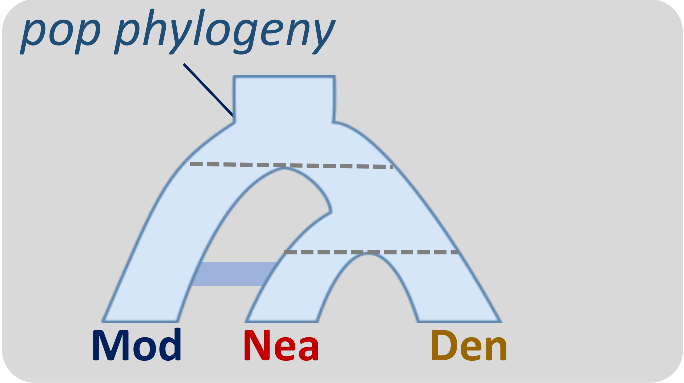


sample	age	sequencing	mean cov
Denisova	> 50 kya	whole genome	31.1
Altai Neandertal	> 50 kya	whole genome	53.7
Carl Zimmer	0	whole genome	> 30

Other present-day genomes sequenced by Meyer, *et al.* (2012) and Prüfer, *et al.* (2014)

The Generalized Phylogenetic Coalescent Sampler (G-PhoCS)

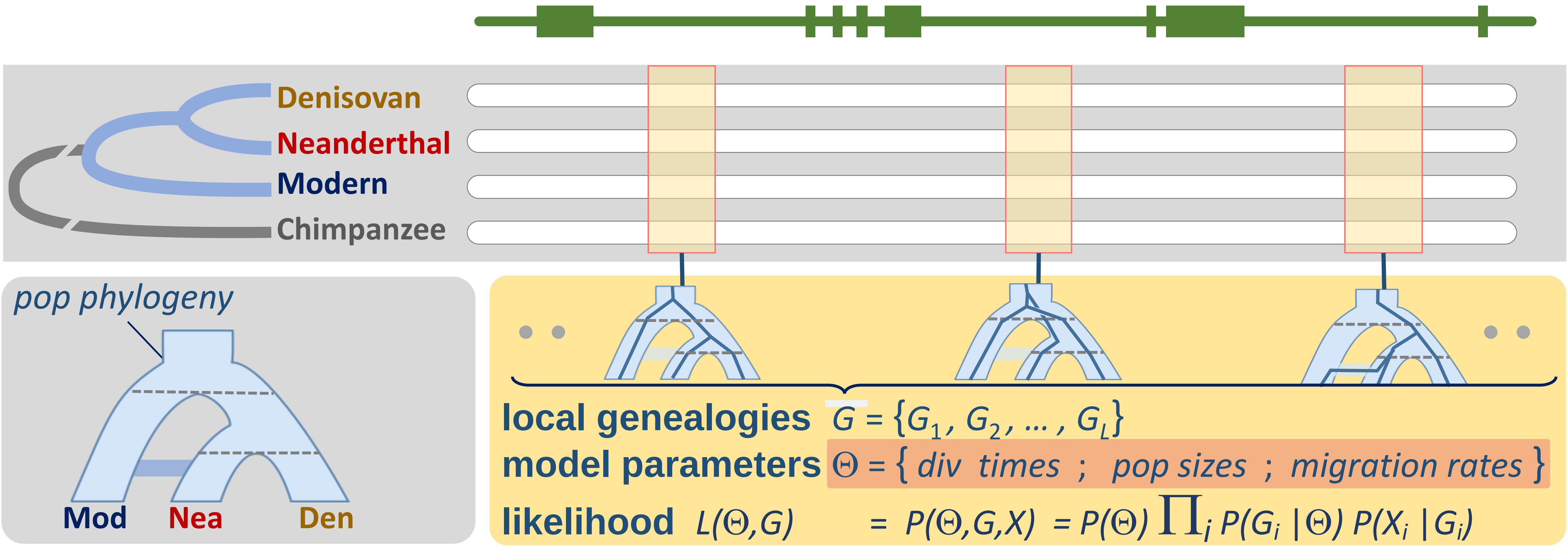




- **Choose short (<1 kb) interspersed (>30 kb apart) loci, outside of protein-coding genes**
- **Assume a given topology for the population phylogeny**

Gronau, *et al.* (2011)

The Generalized Phylogenetic Coalescent Sampler (G-PhoCS)



local genealogies $G = \{G_1, G_2, \ldots, G_L\}$

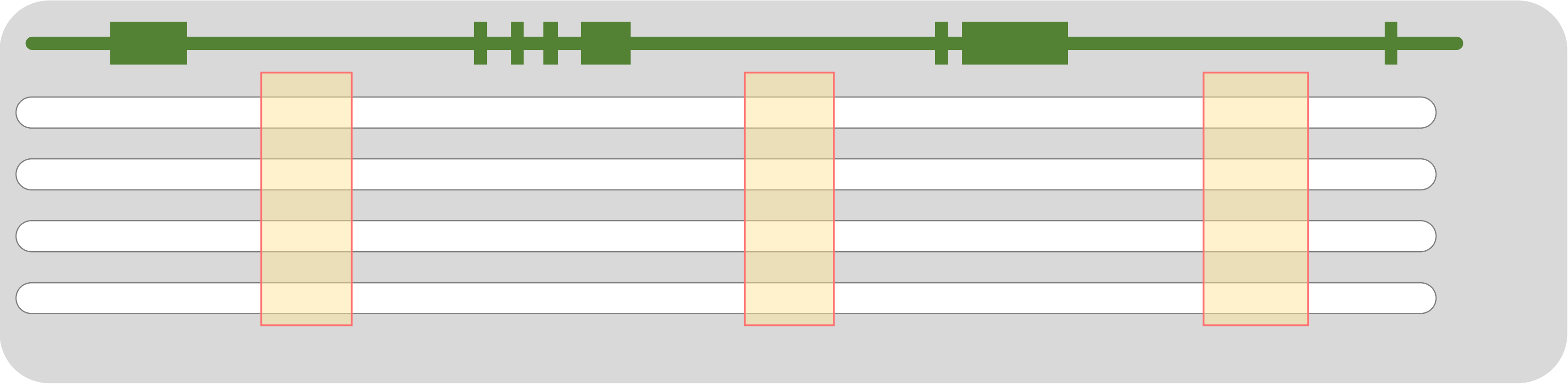
model parameters $\Theta = \{\ div\ \ times\ ;\ \ pop\ sizes\ ;\ migration\ rates\ \}$

likelihood $L(\Theta,G) = P(\Theta,G,X) = P(\Theta) \prod_i P(G_i \mid \Theta)\, P(X_i \mid G_i)$

- **Choose short (<1 kb) interspersed (>30 kb apart) loci, outside of protein-coding genes**
- **Assume a given topology for the population phylogeny**
- **Sample model parameters and local genealogies in proportion to their likelihood (sampling via MCMC)**

Gronau, *et al.* (2011)

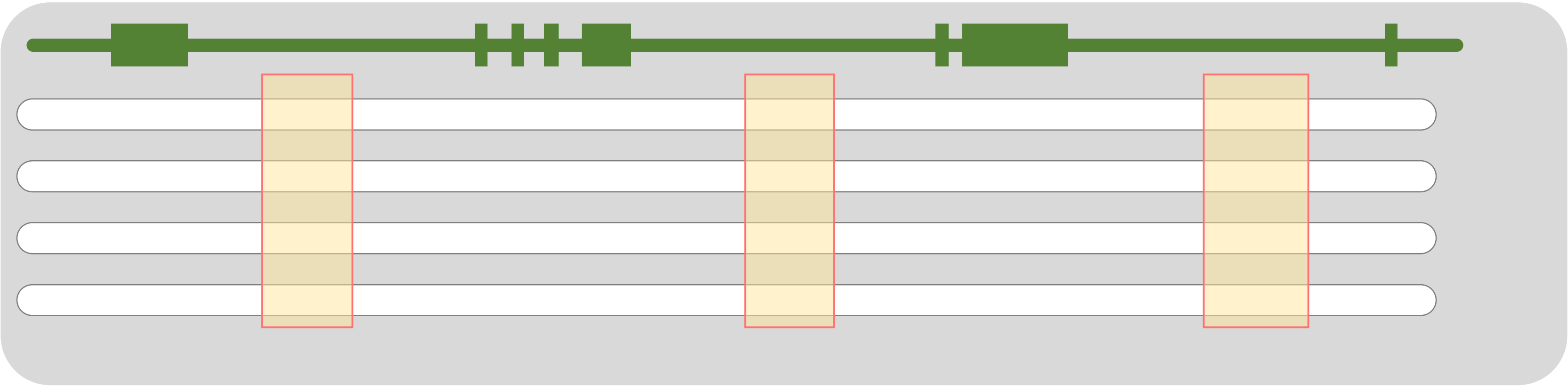
Data setup

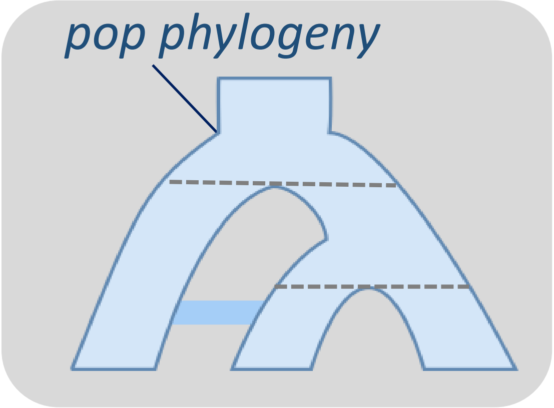


Analyze 13,753 loci:

- 1,000 bp long
- spaced apart (>30 kb)
- far from protein-coding genes (>10 kb)

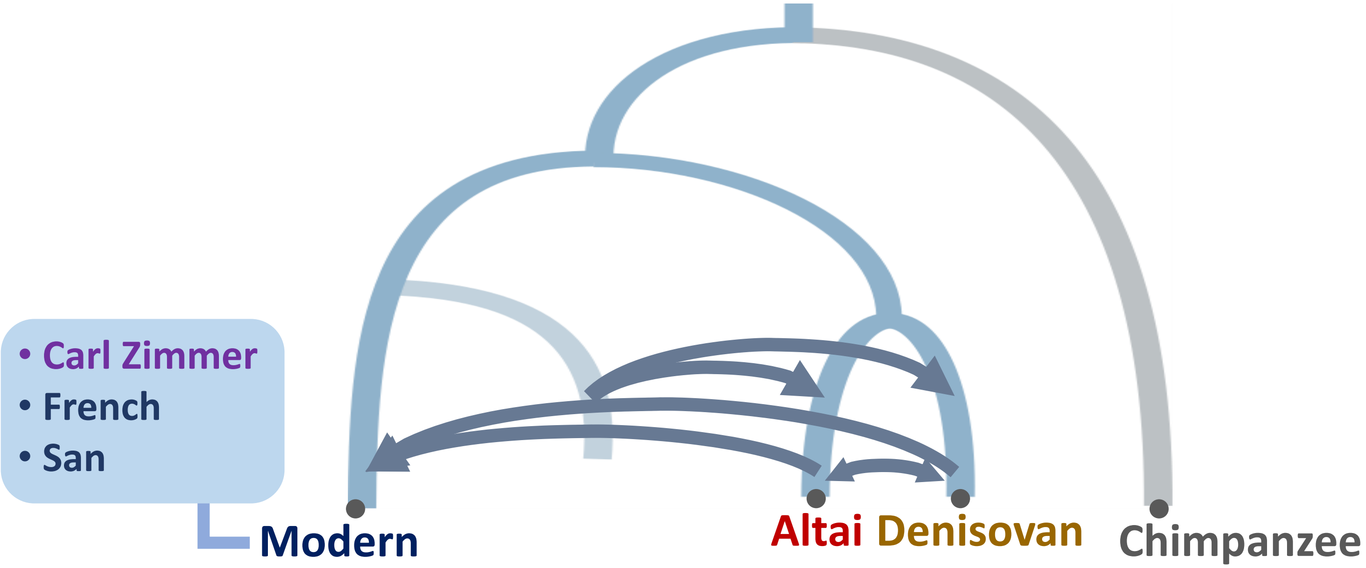
Data setup



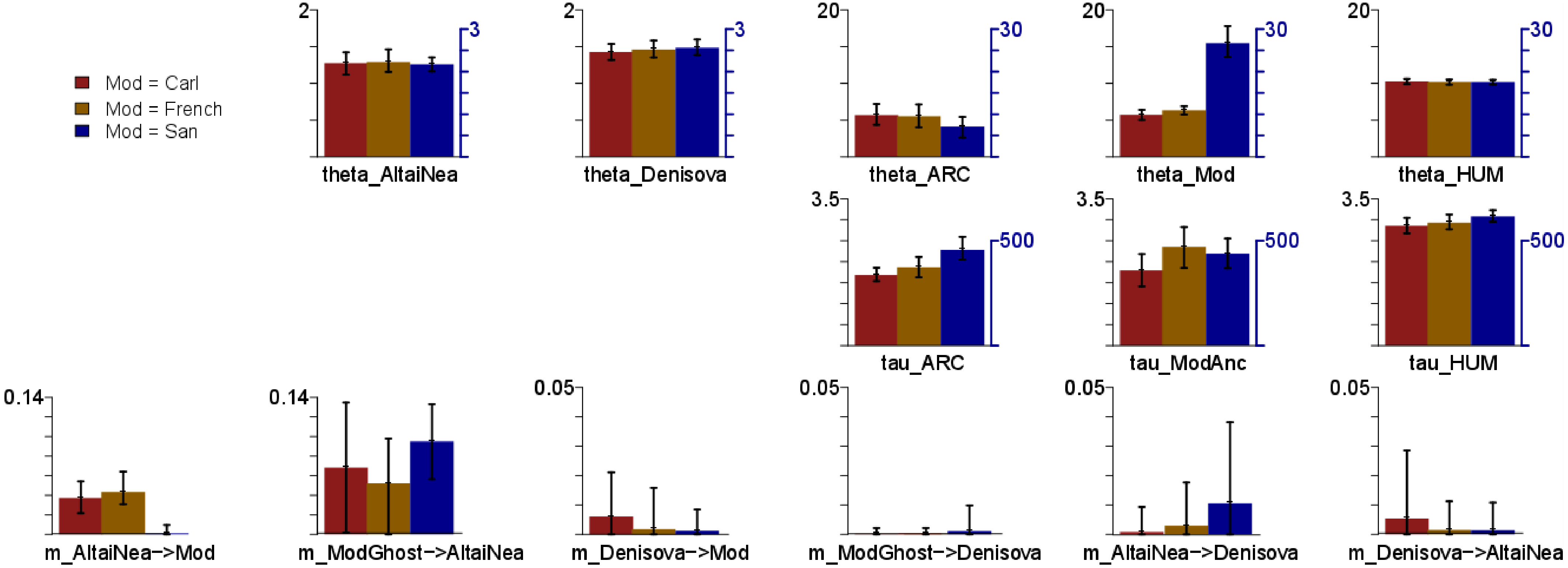


Population phylogeny:

- Each analysis with Neanderthal + Denisovan + one present-day
- Alternate between three different present-day genomes in separate analyses
- 6 migration bands per run



Results



Effective pop. size (Ne / θ)

Pop. divergence time (T / τ)

Total migration rate (*m*)

- **Effective population sizes (Ne) and divergence times (T) are given both in their mutation-scaled version ($\tau = \mu T$ and $\theta = 4\mu g Ne$) scaled by 10^4 and in their calibrated scale assuming a mutation rate of $\mu = ½ \times 10^{-9}$ (per bp per year) (and average generation time of 29 years)**
- **Total migration rates approximate the probability that a lineage in target population migrated through the migration band**