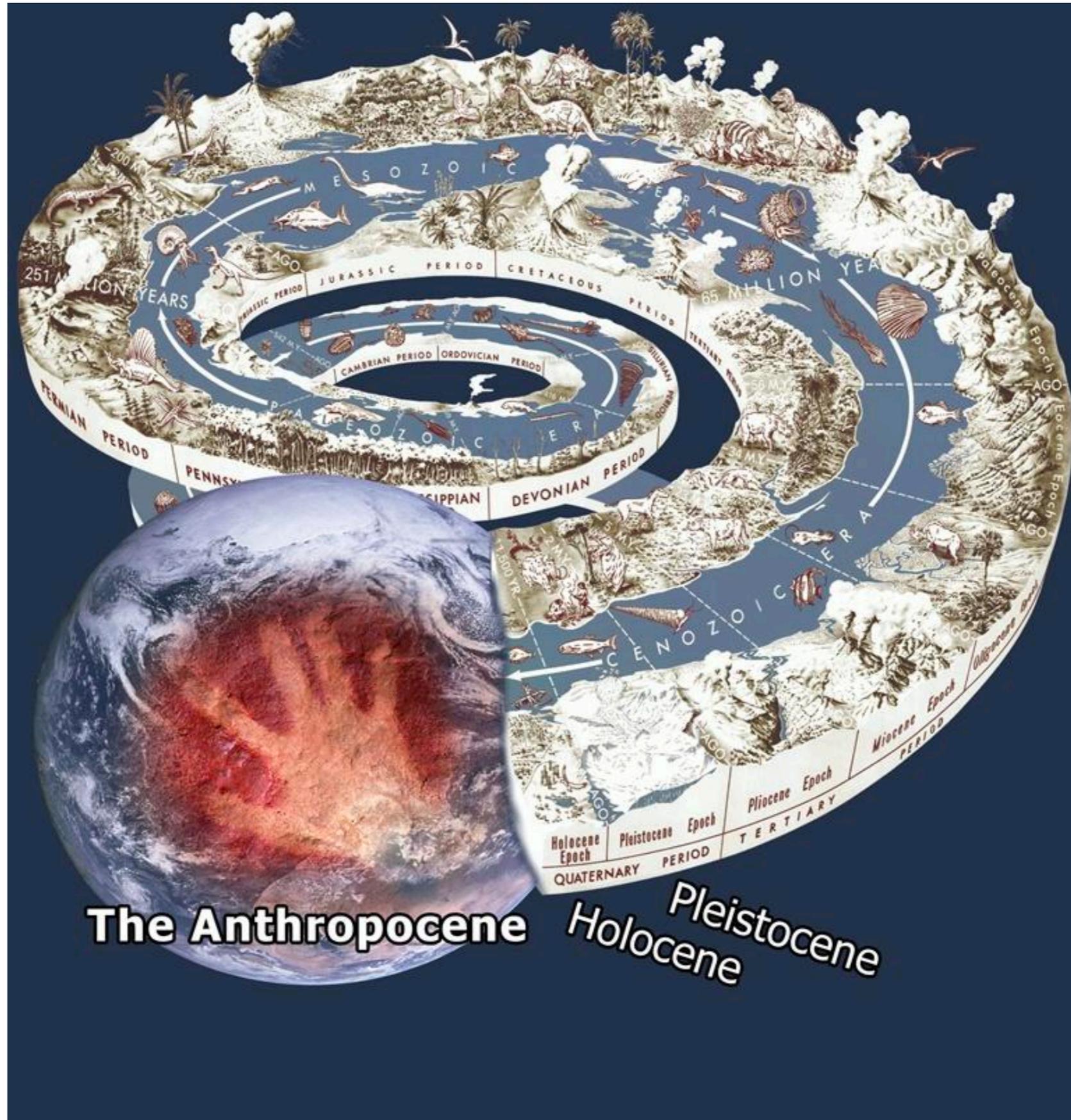


# Conservation genomics



# We're now in the Anthropocene



# One in four species are at risk of extinction

Species assessed by the IUCN Red List



**Amphibians**  
**40%**



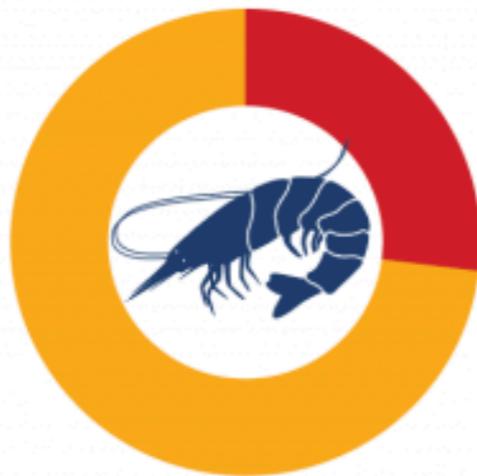
**Conifers**  
**34%**



**Reef corals**  
**33%**



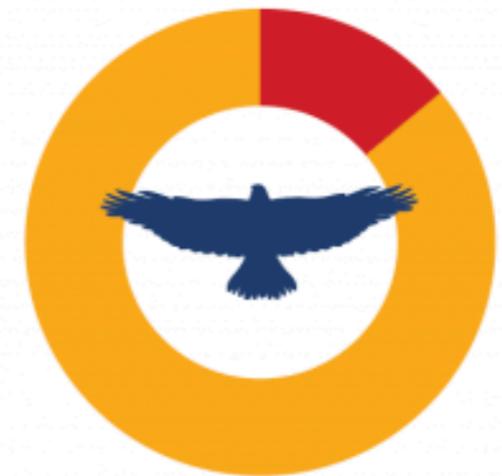
**Sharks & Rays**  
**31%**



**Selected crustaceans\***  
**27%**



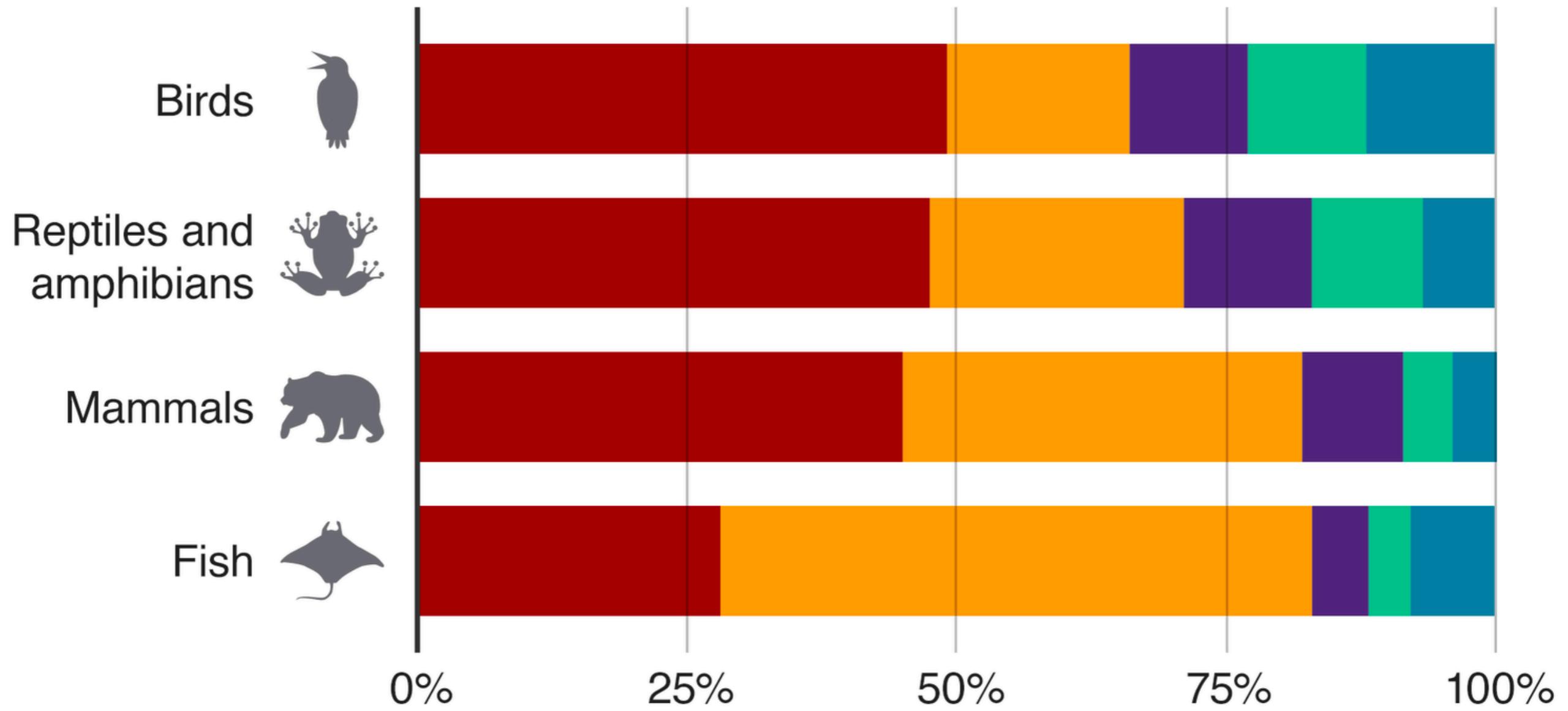
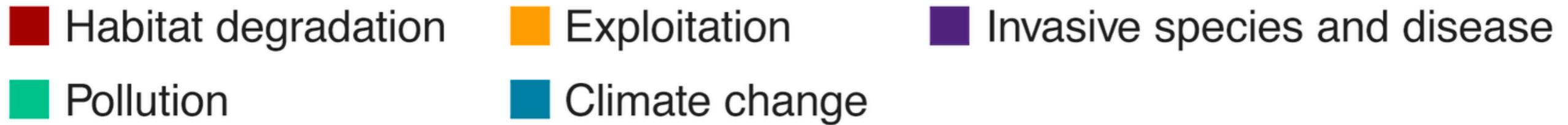
**Mammals**  
**35%**



**Birds**  
**14%**

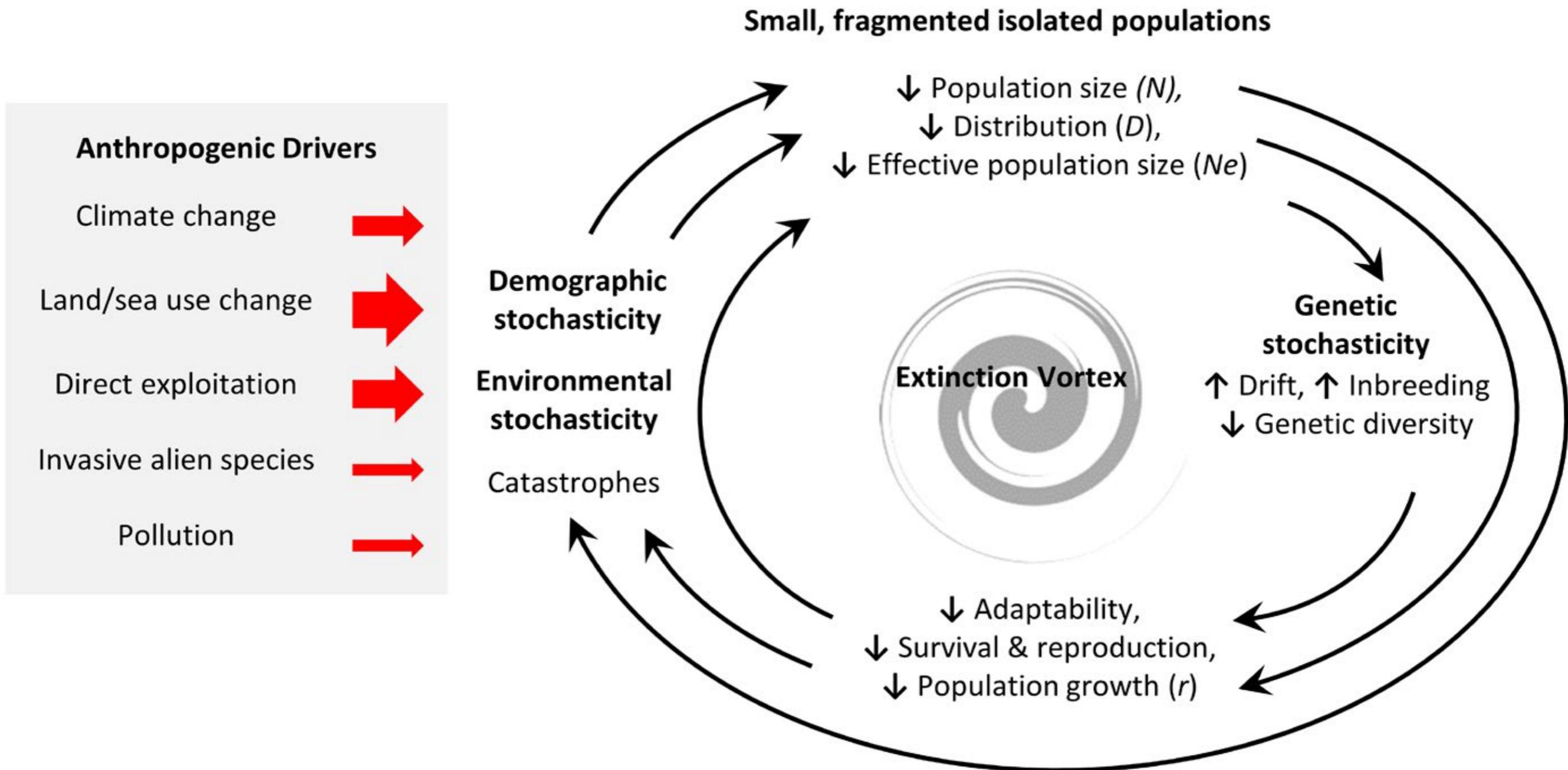
# Habitat loss is a major threat to biodiversity

The Living Planet Report assesses key drivers of species decline



Note: A sample of 3,789 populations evaluated by the Living Planet Index

# The extinction vortex





# Conservation genetics & genomics

- Inferring population demography
- Identifying adaptive genetic variation
- Informing genetic rescue efforts

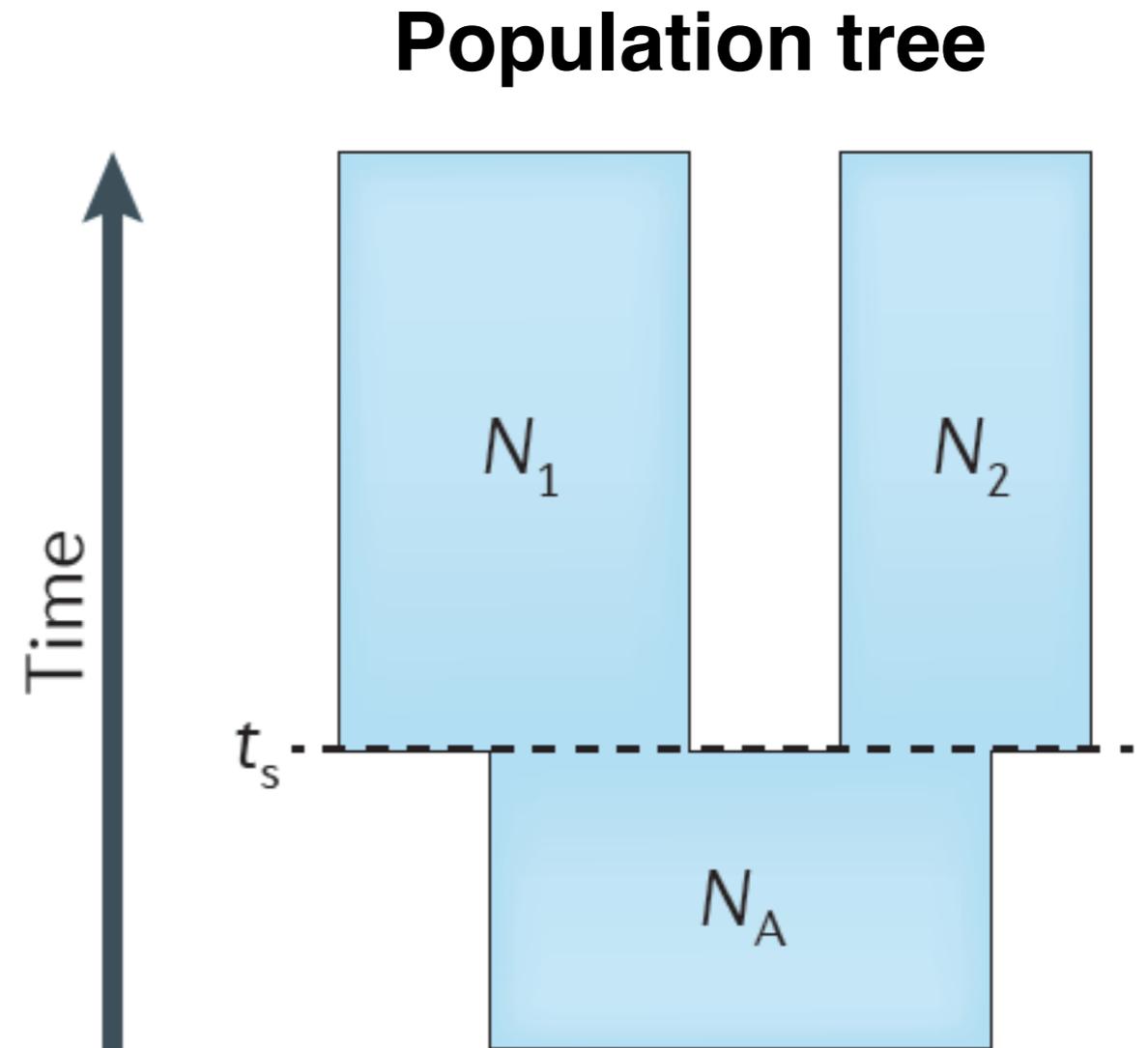
# Conservation genetics & genomics

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# What is population demography?

Effective population sizes back in time (expansions, bottlenecks, etc.)

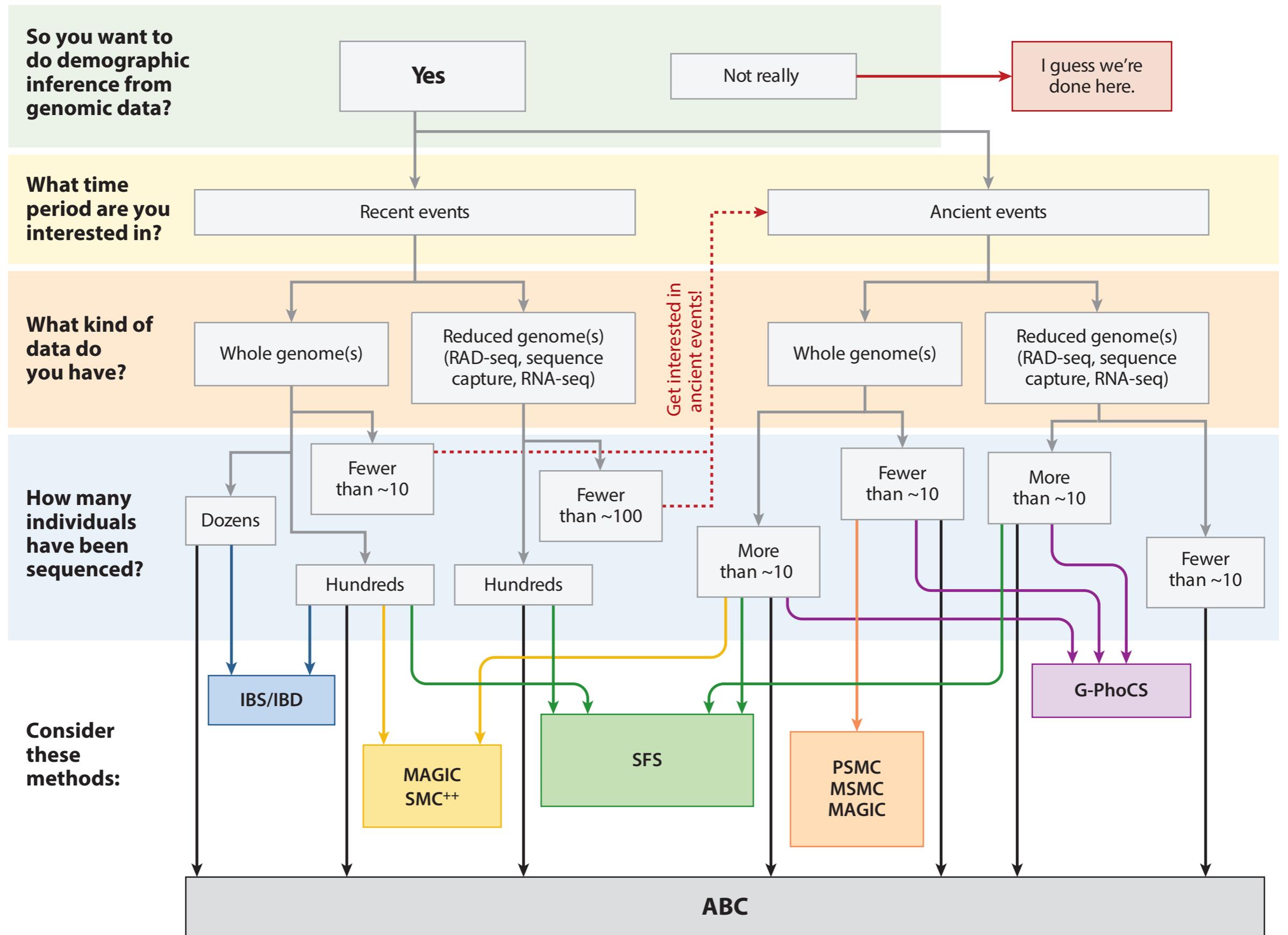
Population structure (split times) and migration rates over time



# Why model demographic history?

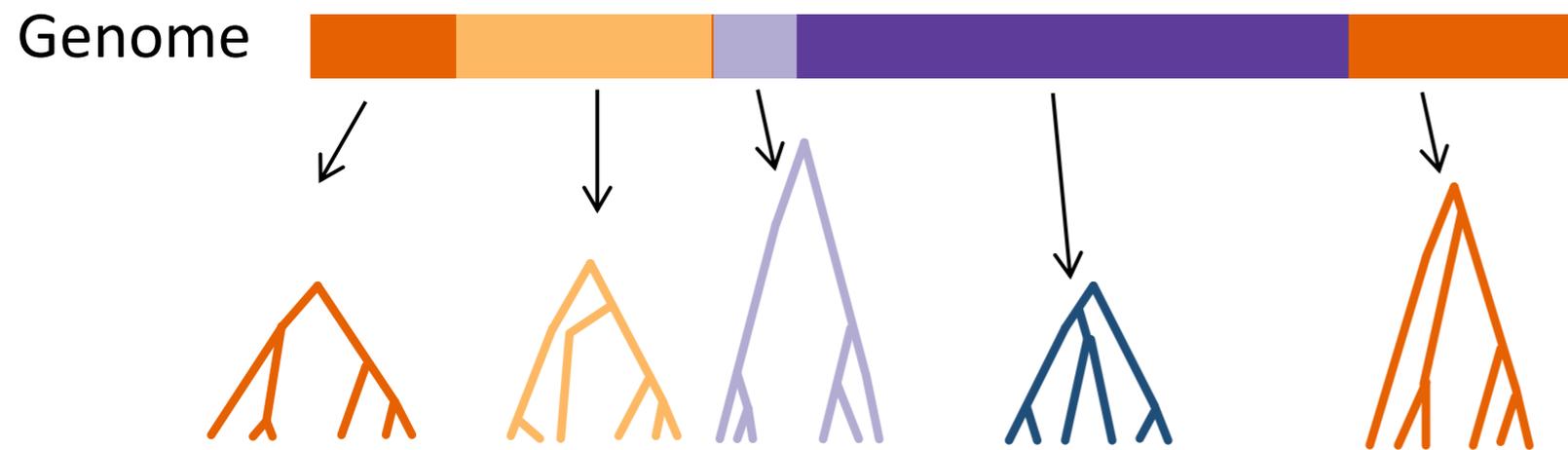
- Understand population history
- Neutral background for tests of selection
- Conservation: present vs. historical levels of genetic diversity

# Many methods for demographic inference

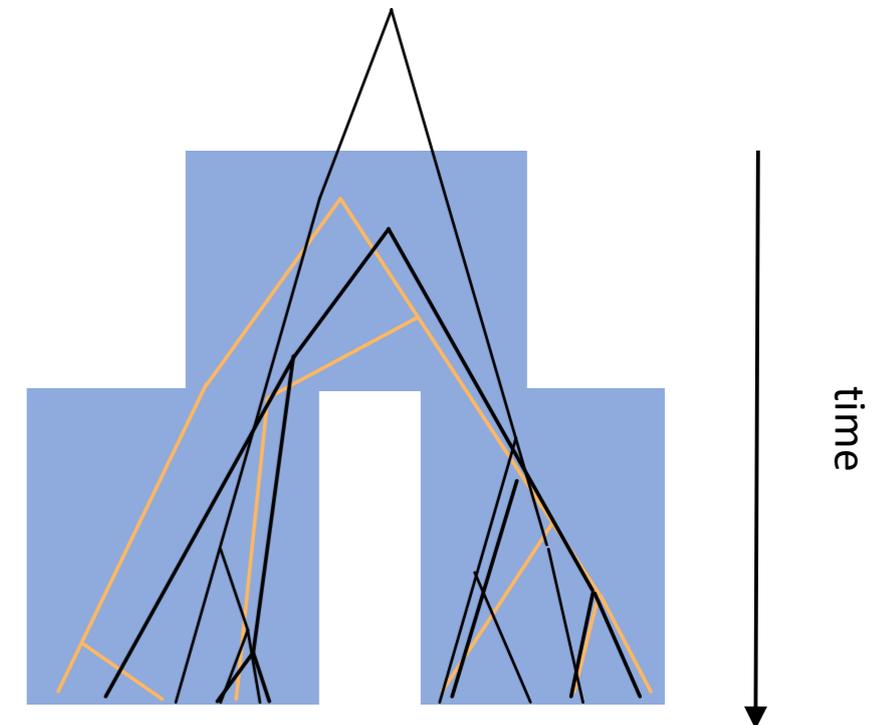


# Reconstructing demographic history from genomic data

Because of recombination, different regions of the genome can have different gene trees

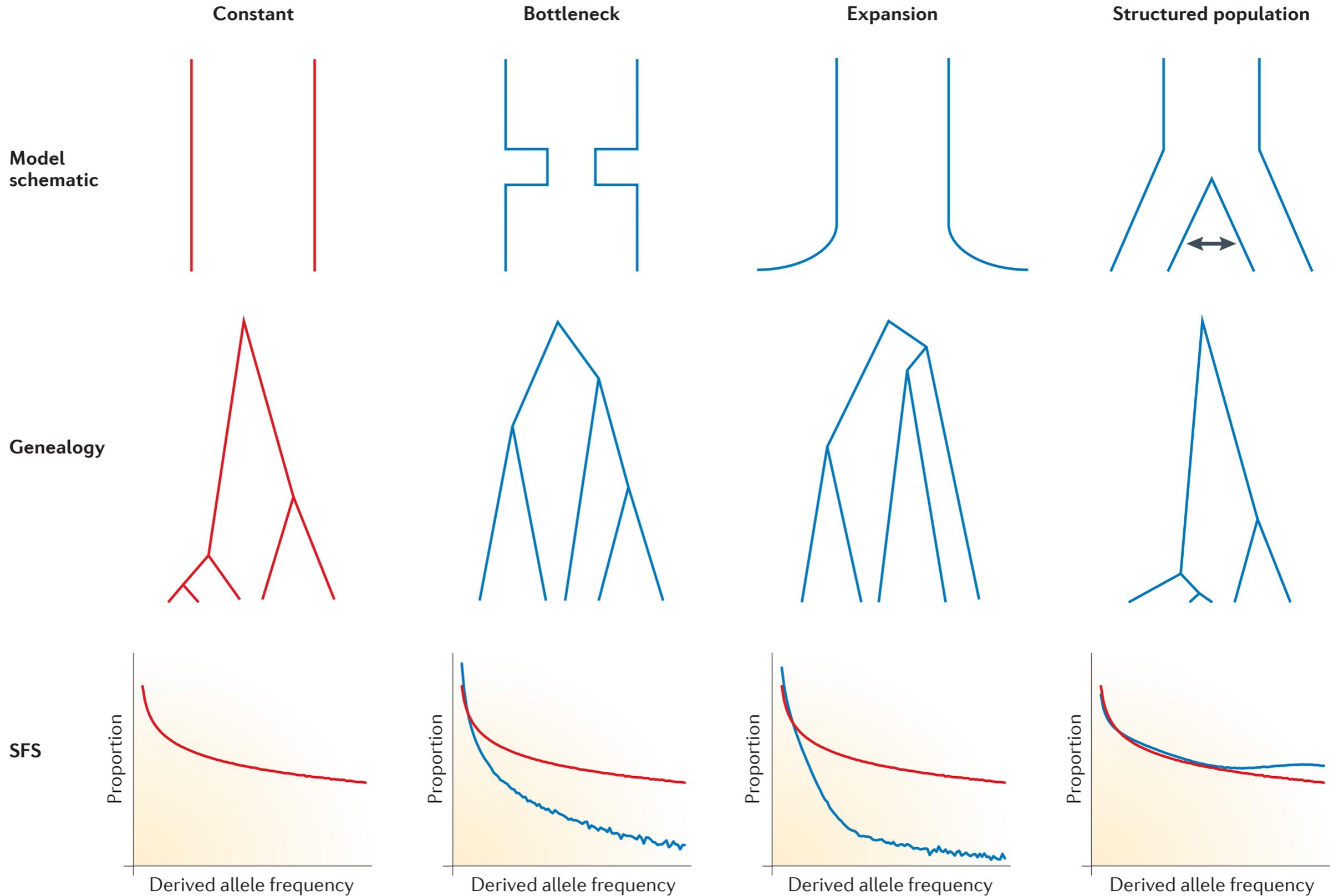


- Demography is expected to affect the entire genome
- Natural selection acts on specific functional regions



All gene trees are consistent with the population tree. Independent gene trees can be seen as independent replicates of the same population tree.

# Demography influences coalescent genealogies & the site frequency spectrum (SFS)

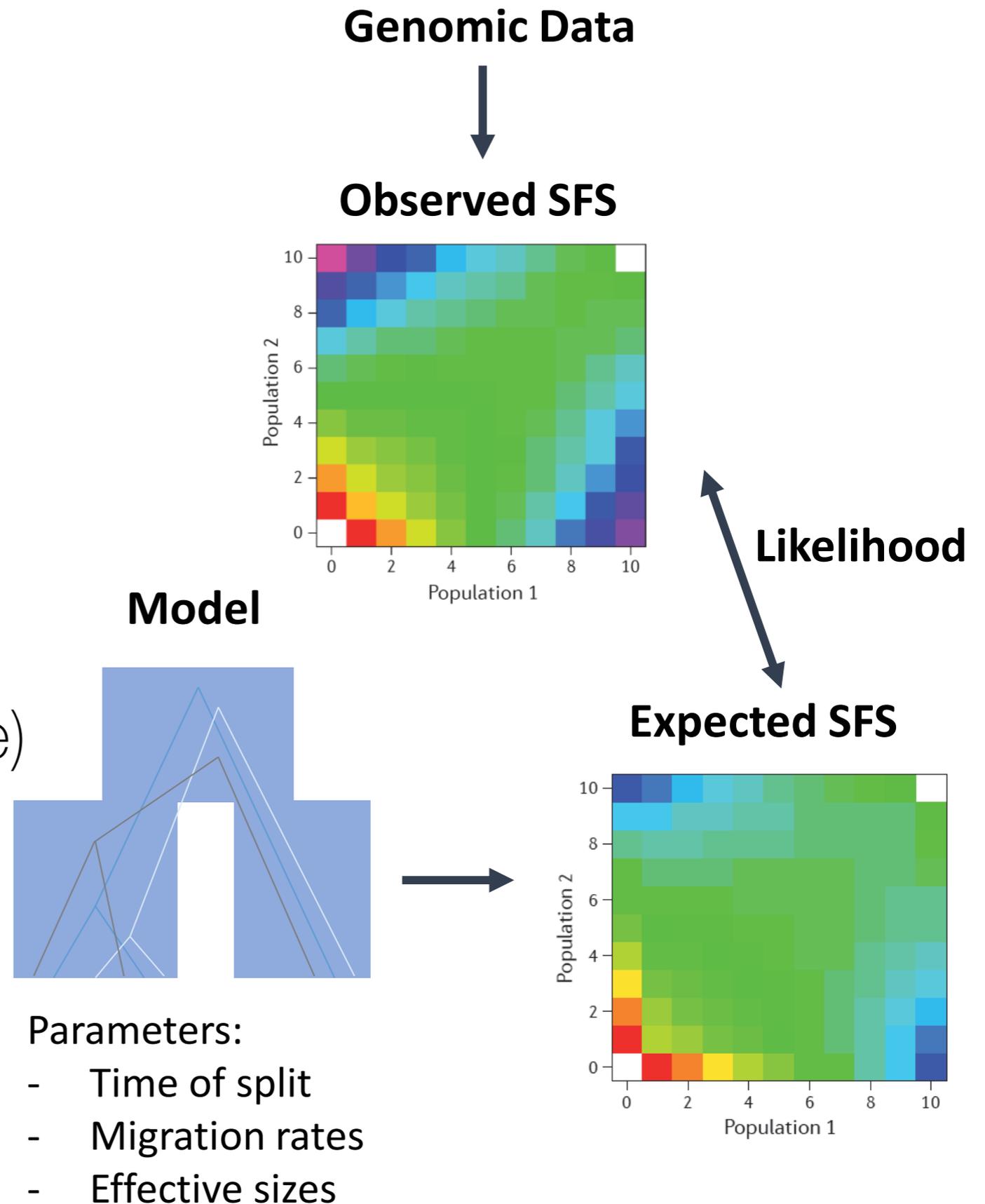


# Inferring demographic history from the SFS

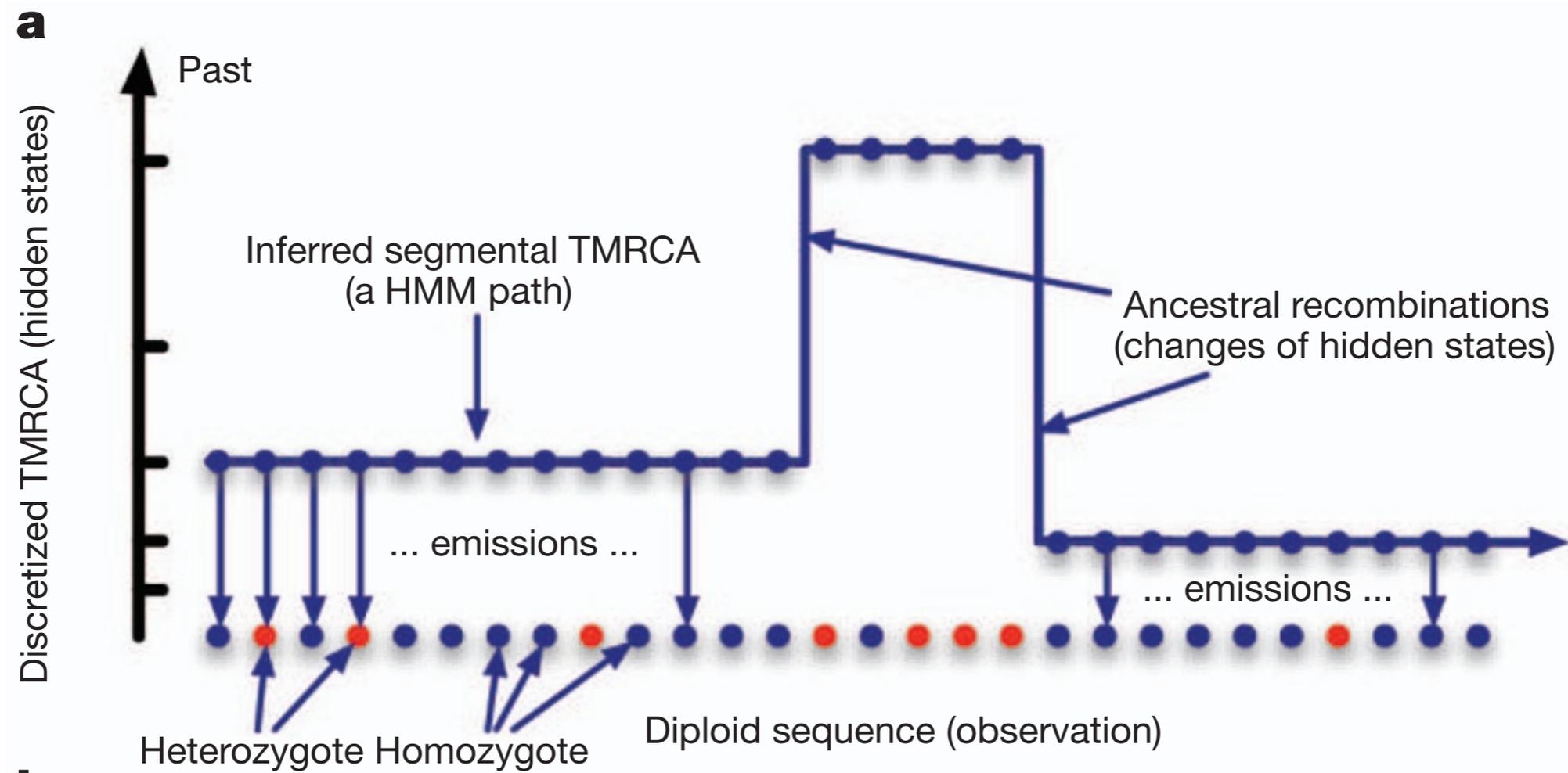
Compute likelihood of data based on the expected SFS under a given model.

Different ways of obtaining the expected SFS:

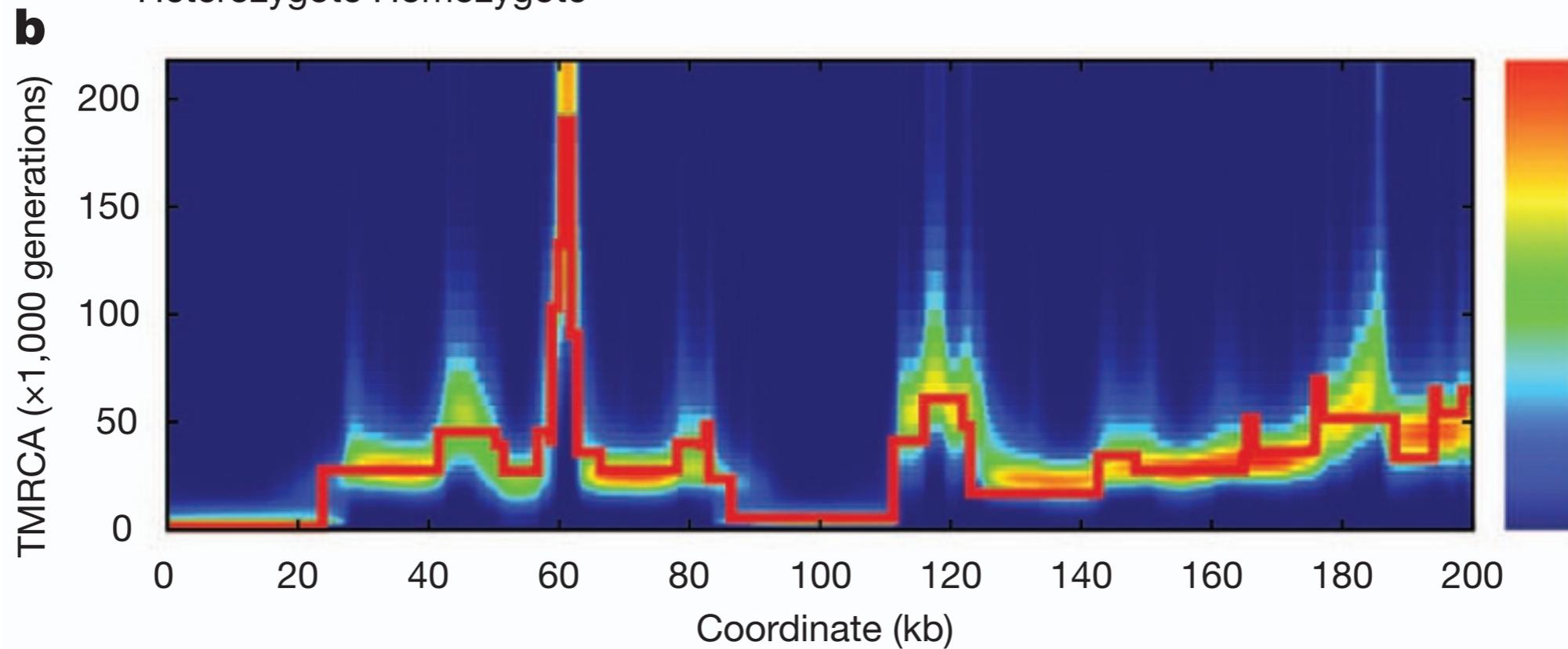
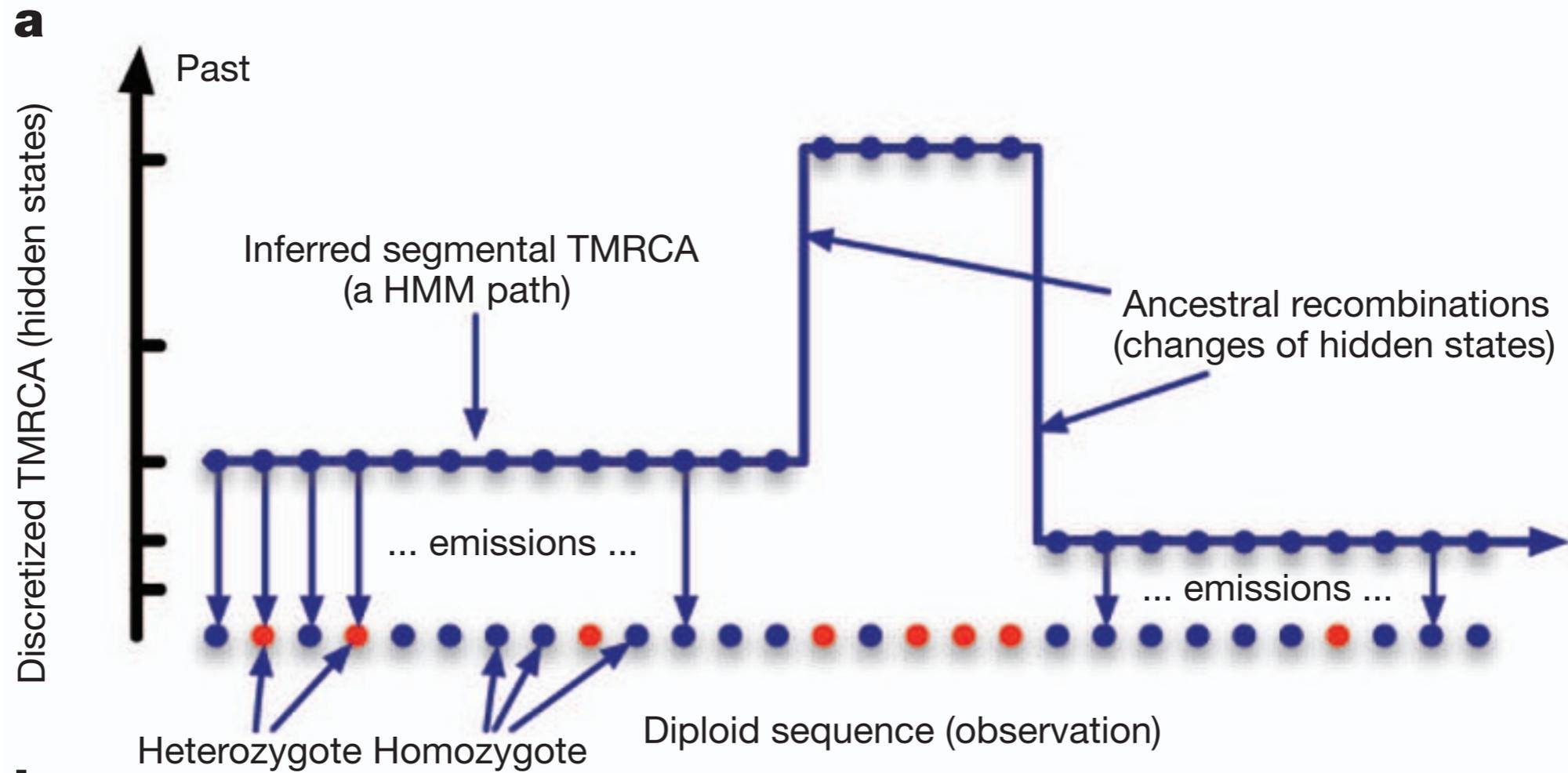
- Diffusion (forward in time)
- Coalescent (backward in time)



# Pairwise Sequentially Markovian Coalescent

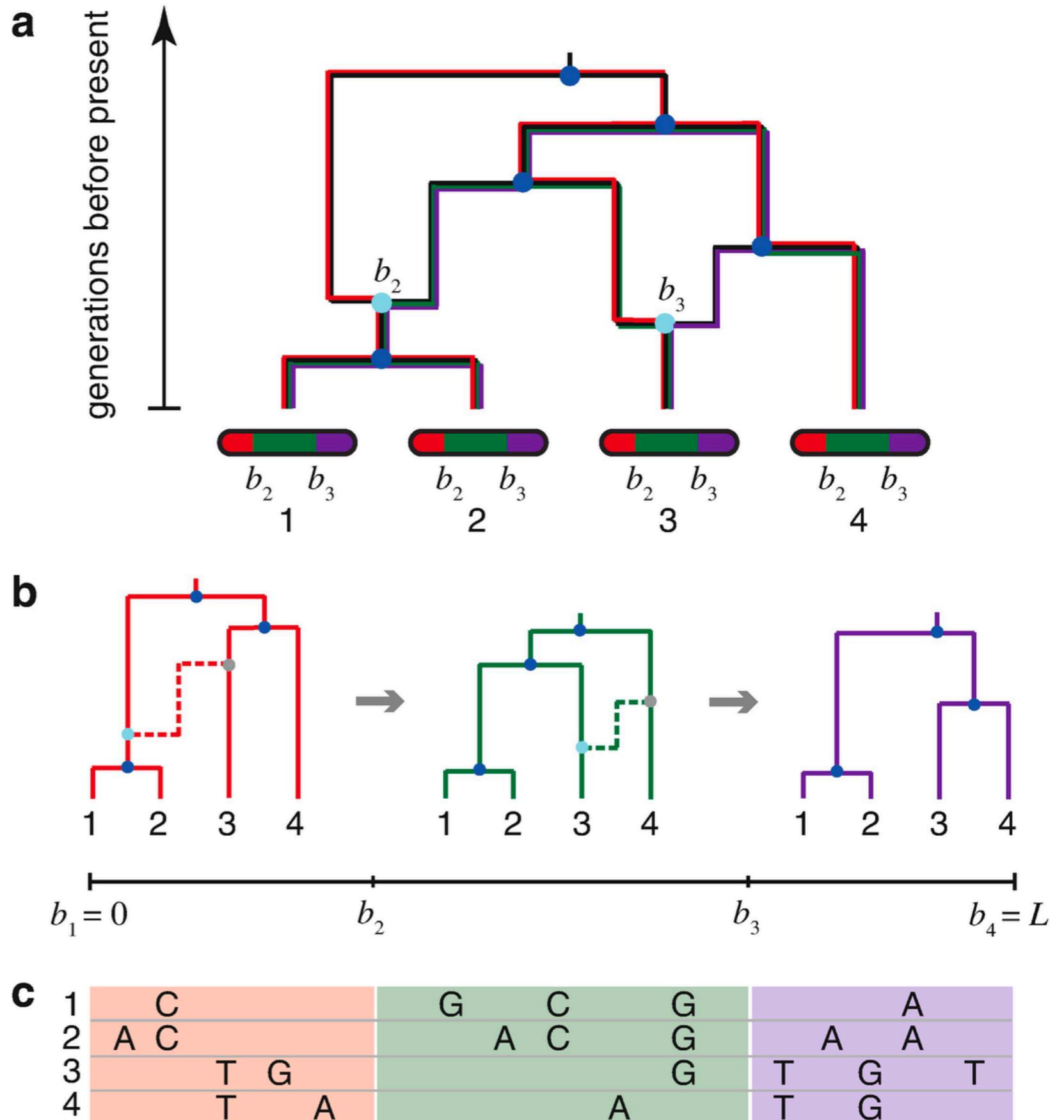


# Pairwise Sequentially Markovian Coalescent

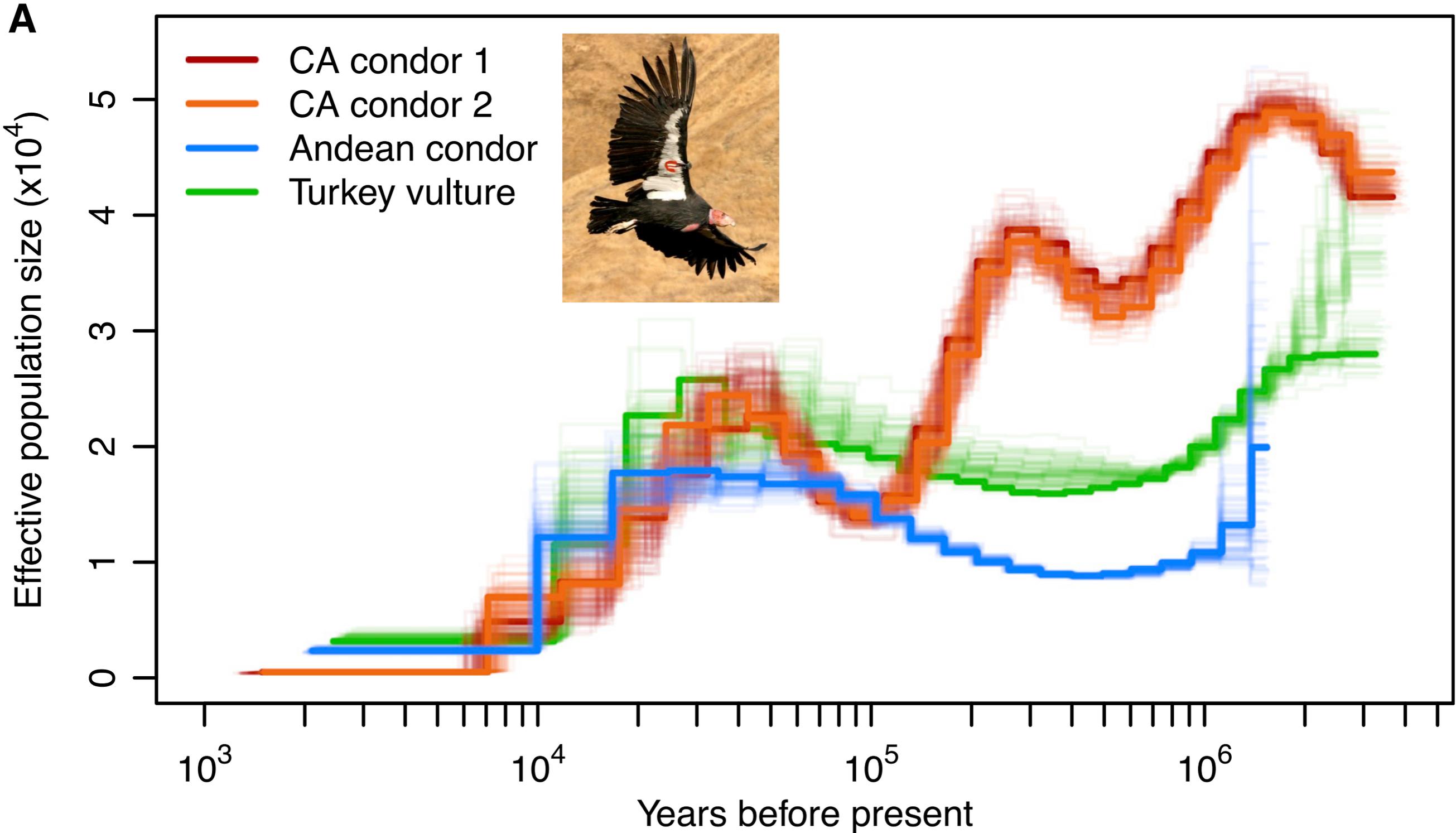


# Ancestral recombination graph (ARG)

The ARG describes how individual sequences in a population are related, and provides the combined history of recombination, mutation, and coalescence.



# Demographic inference of the CA condor



# Conservation genetics & genomics

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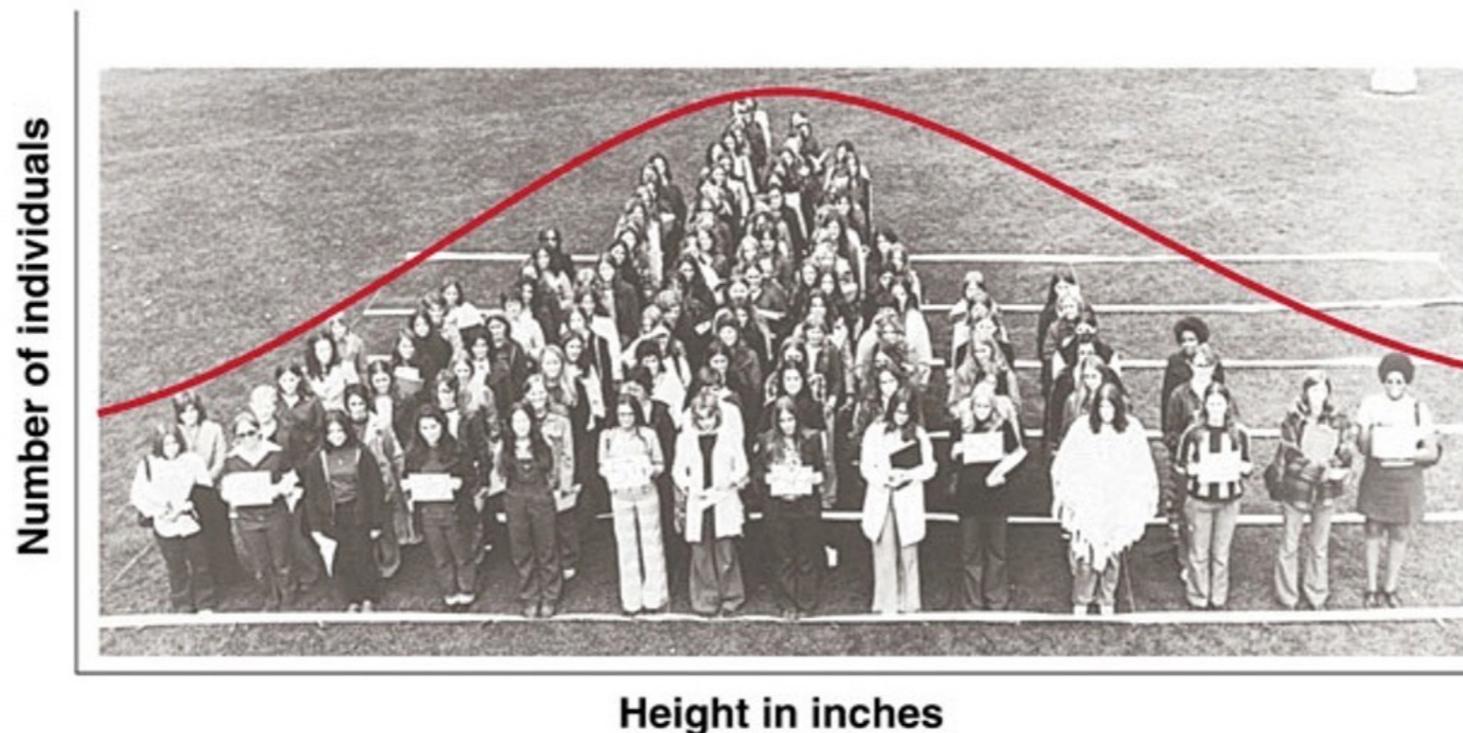
Phenotypes are determined by genetic and environmental factors

$$P = G + E$$

Phenotypes are determined by genetic and environmental factors

$$P = G + E$$

Many traits are **polygenic**, or controlled by multiple genes.



# Phenotypic variation among individuals

Phenotypic variance  $V_P$  is composed of genetic variance  $V_G$  & environmental variance  $V_E$

$$V_P = V_G + V_E$$

We can further partition  $V_G$

$$V_G = V_A + V_D + V_I$$

Where  $V_A$  = additive genetic variance

$V_D$  = dominance variance

$V_I$  = epistatic variance

Heritability is the genetic contribution to phenotypic variance

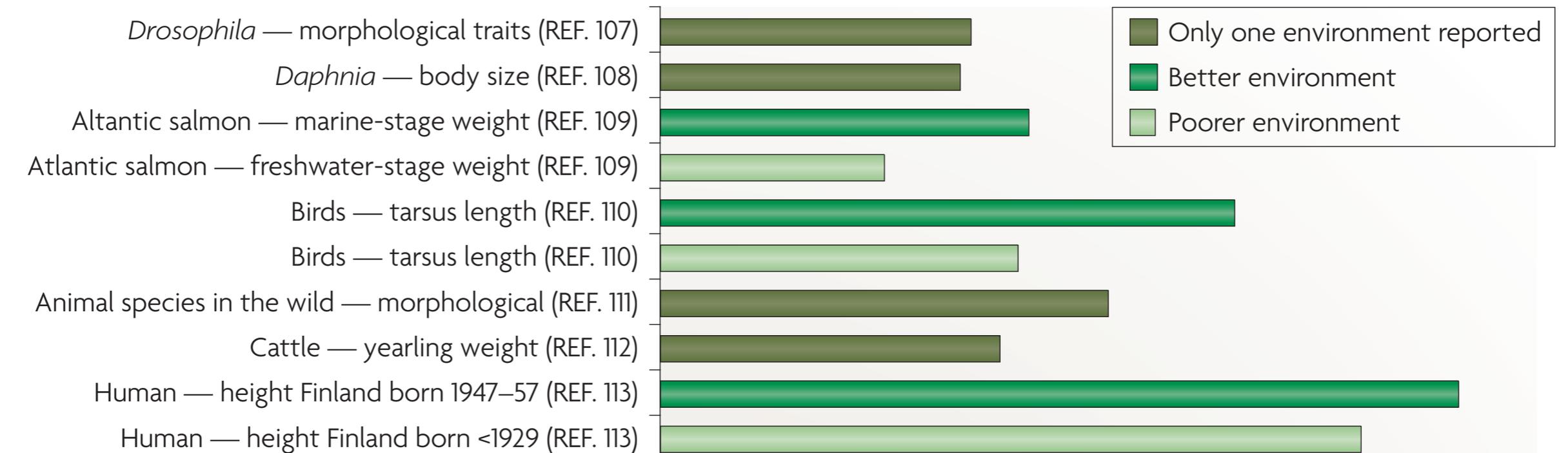
Narrow sense heritability

$$h^2 = V_A / V_P$$

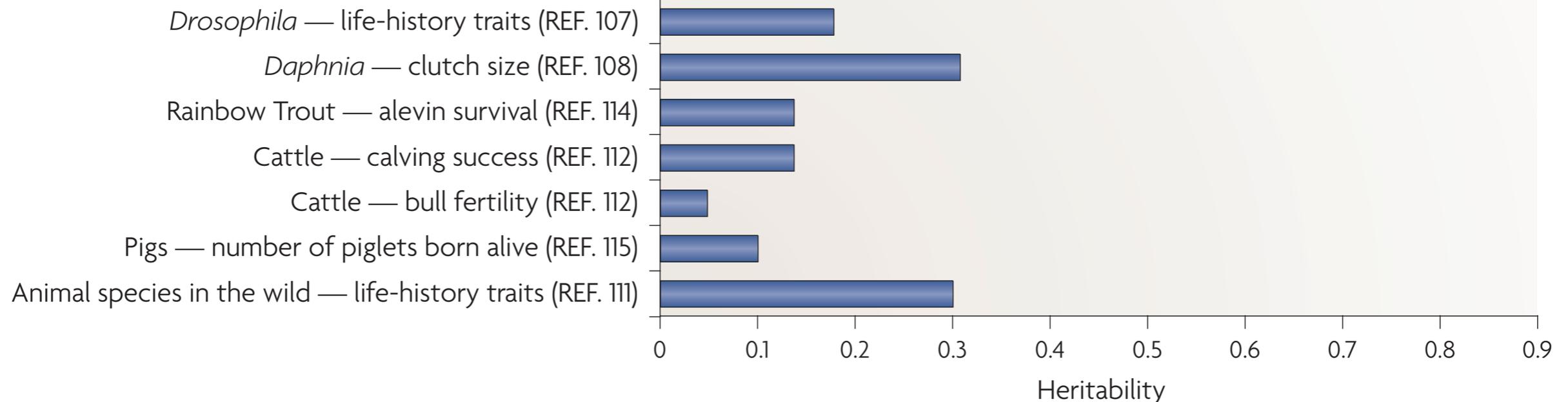
Heritability underlies the predicted response to selection and is a population-specific measure

# Many traits have $h^2$ between 0.1 & 0.9

## Morphological traits

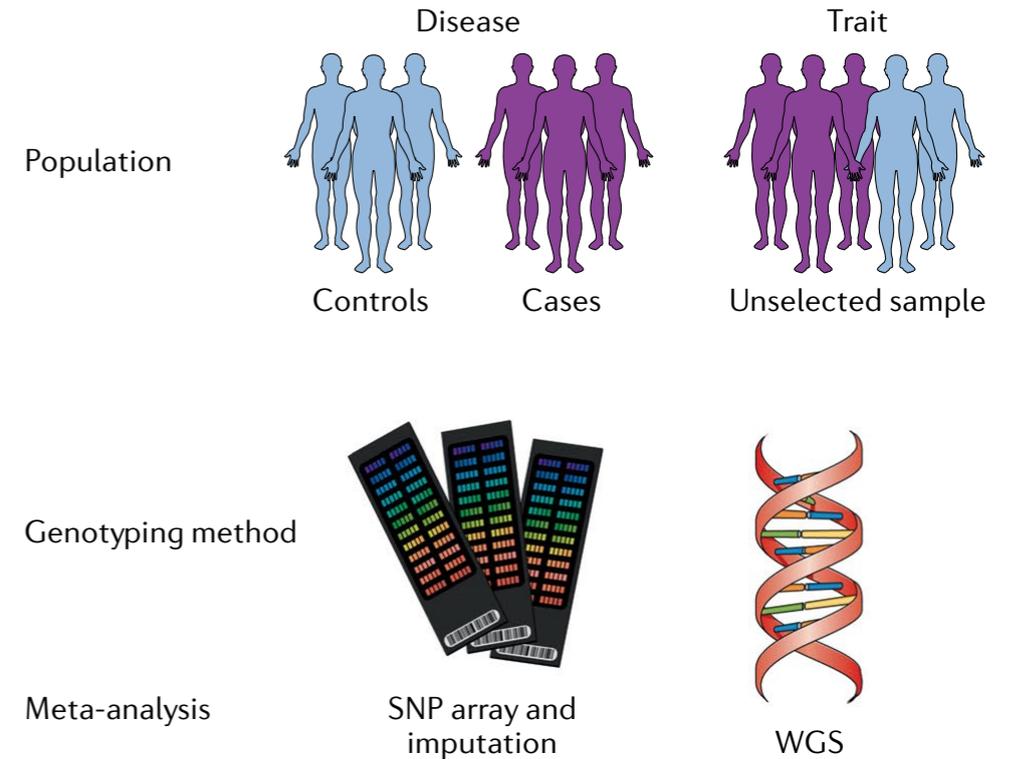


## Fitness traits



# Genome-wide association study

GWAS test for associations between allele frequency at each marker and trait value



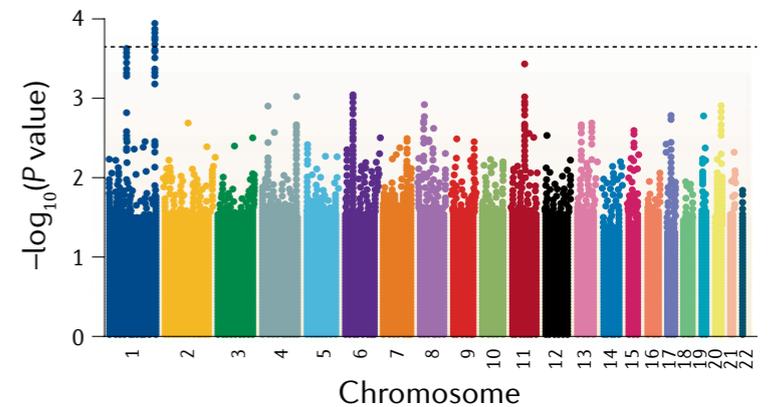
Phenotype                      Genotype

$$y = \alpha_l g_l + e$$

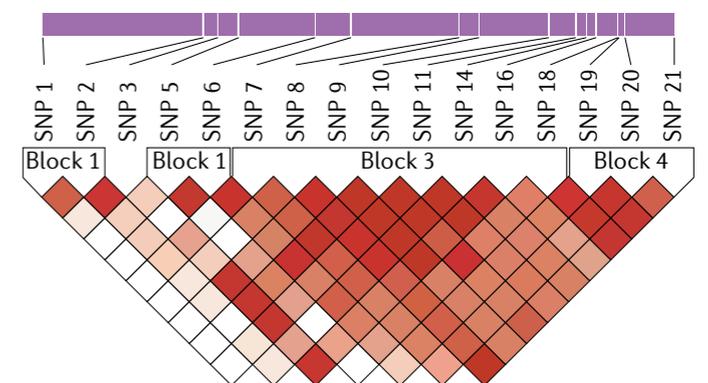
Additive effect size estimate

Residual

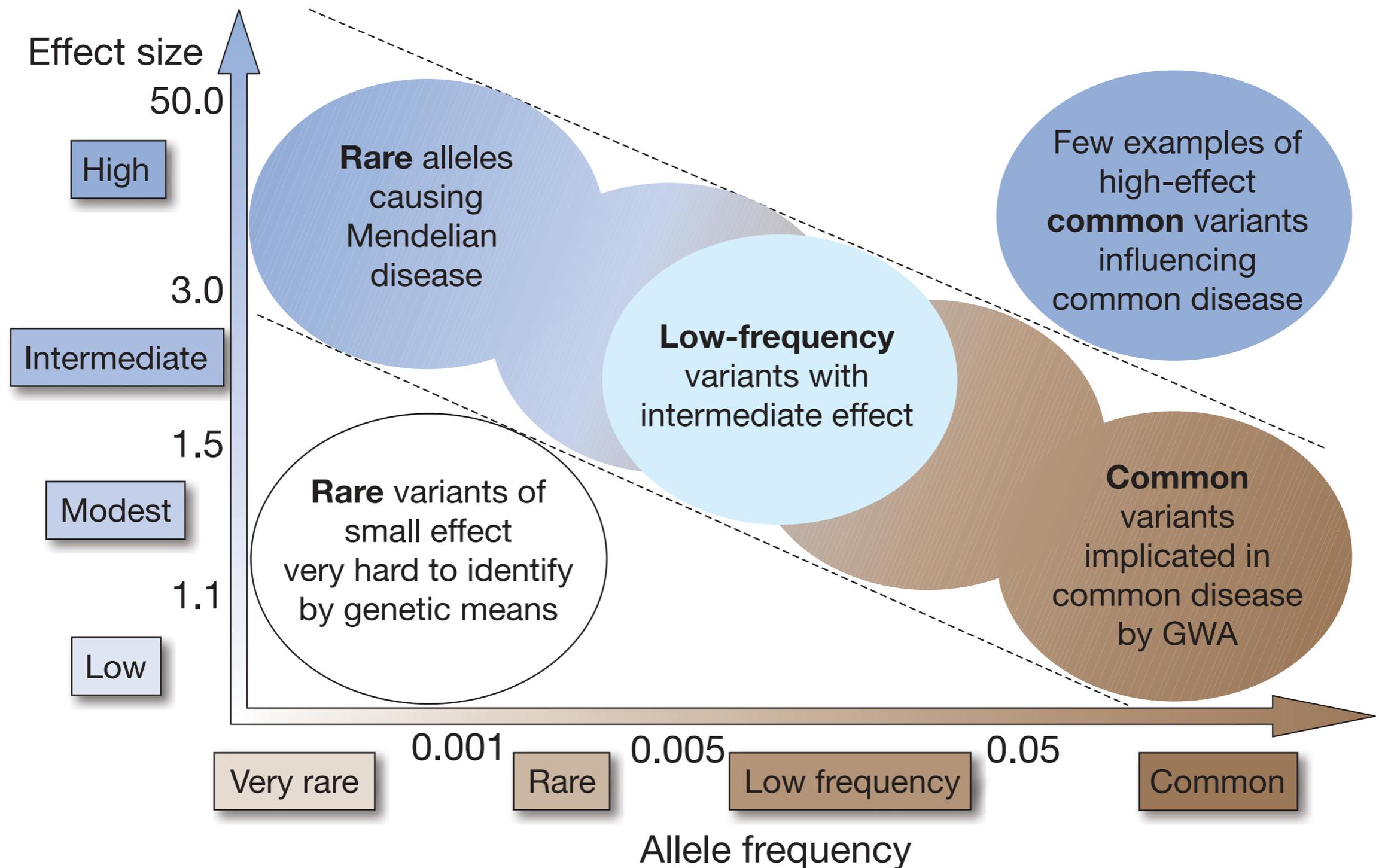
Statistical association



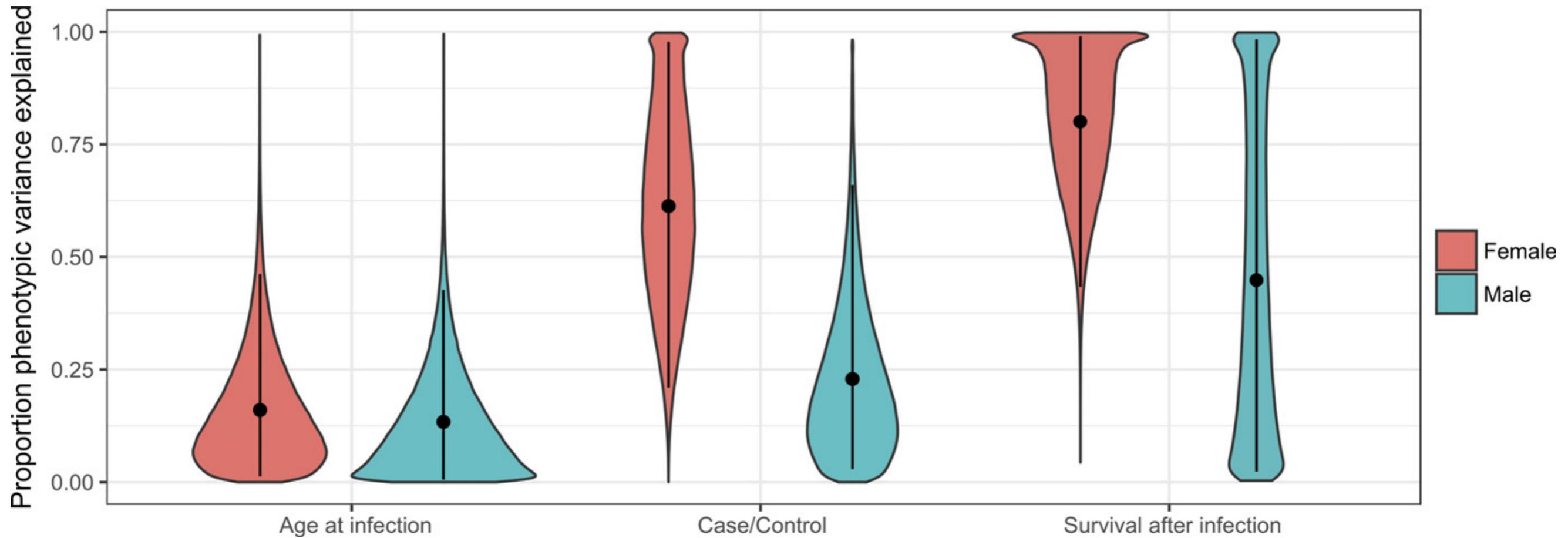
Linkage disequilibrium



# Genetic architecture of trait determines power of detection



# Heritability of cancer-related traits in Tasmanian devils



# The inflated significance of neutral genetic diversity in conservation genetics

João C. Teixeira<sup>a,b,1</sup>  and Christian D. Huber<sup>a,1</sup> 

# The crucial role of genome-wide genetic variation in conservation

Marty Kardos<sup>a,1</sup> , Ellie E. Armstrong<sup>b</sup> , Sarah W. Fitzpatrick<sup>c,d,e</sup> , Samantha Hauser<sup>f</sup>, Philip W. Hedrick<sup>g</sup>, Joshua M. Miller<sup>h,i,j</sup> , David A. Tallmon<sup>k</sup>, and W. Chris Funk<sup>l</sup> 

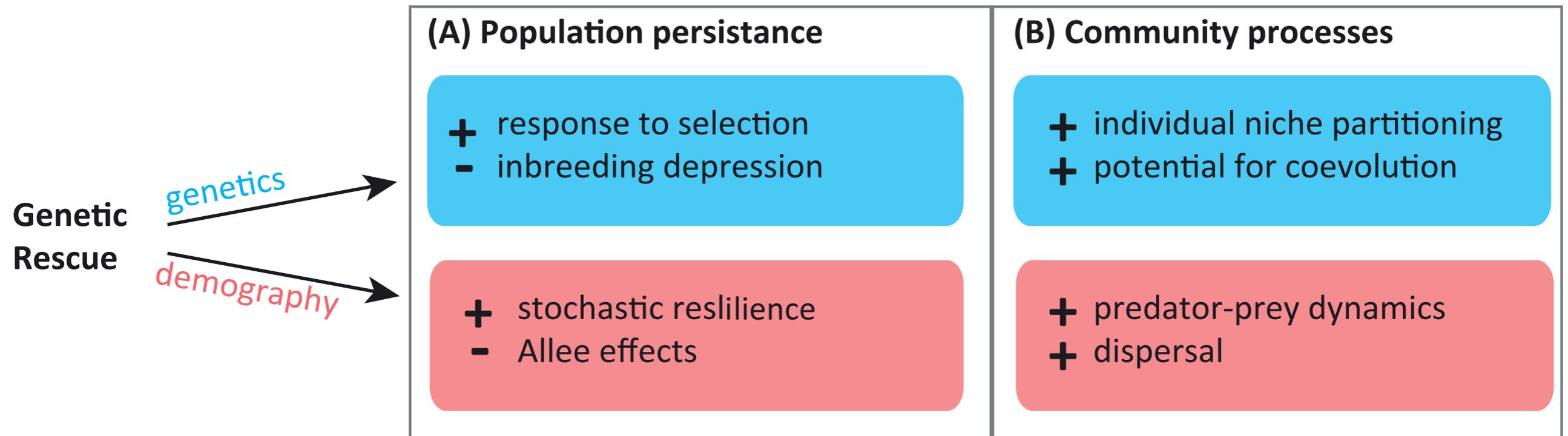
# Conservation genetics & genomics

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# Gene flow can rescue populations

**Demographic rescue:** a decrease in population extinction probability owing to the simple addition of immigrants.

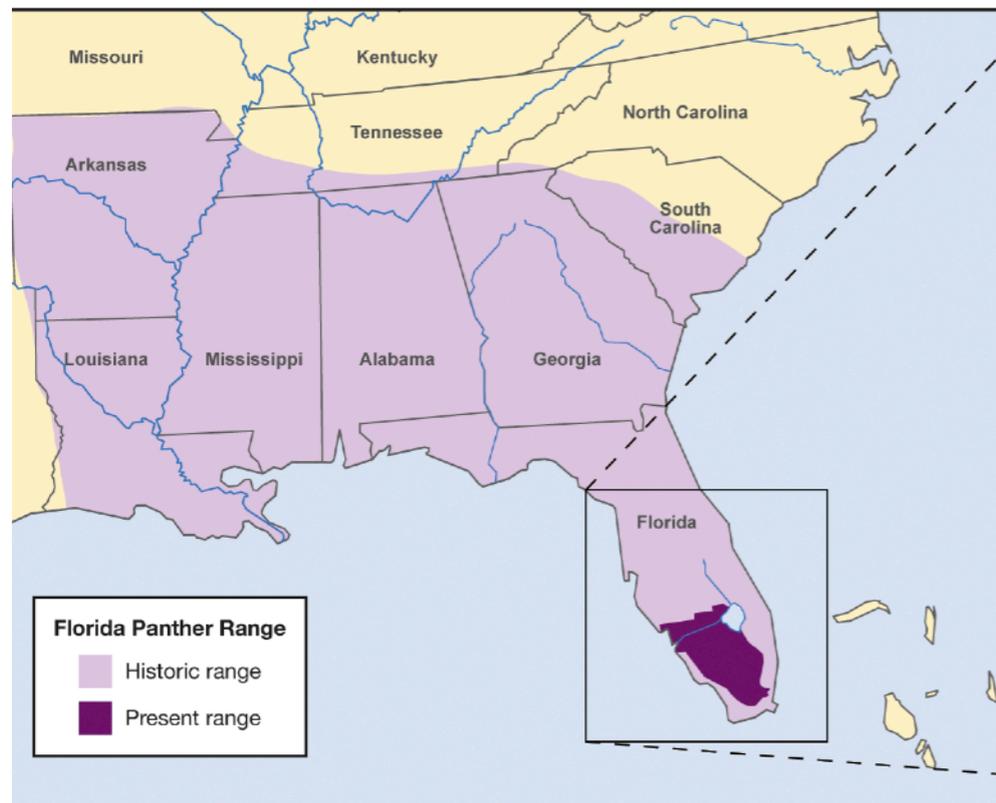
**Genetic rescue:** an increase in population fitness (growth) owing to the influx of genetic variation from immigrants.



# Genetic rescue of the Florida Panther

Florida panthers (*Puma concolor coryi*): ~80 individuals left, listed as Federally Endangered in 1967

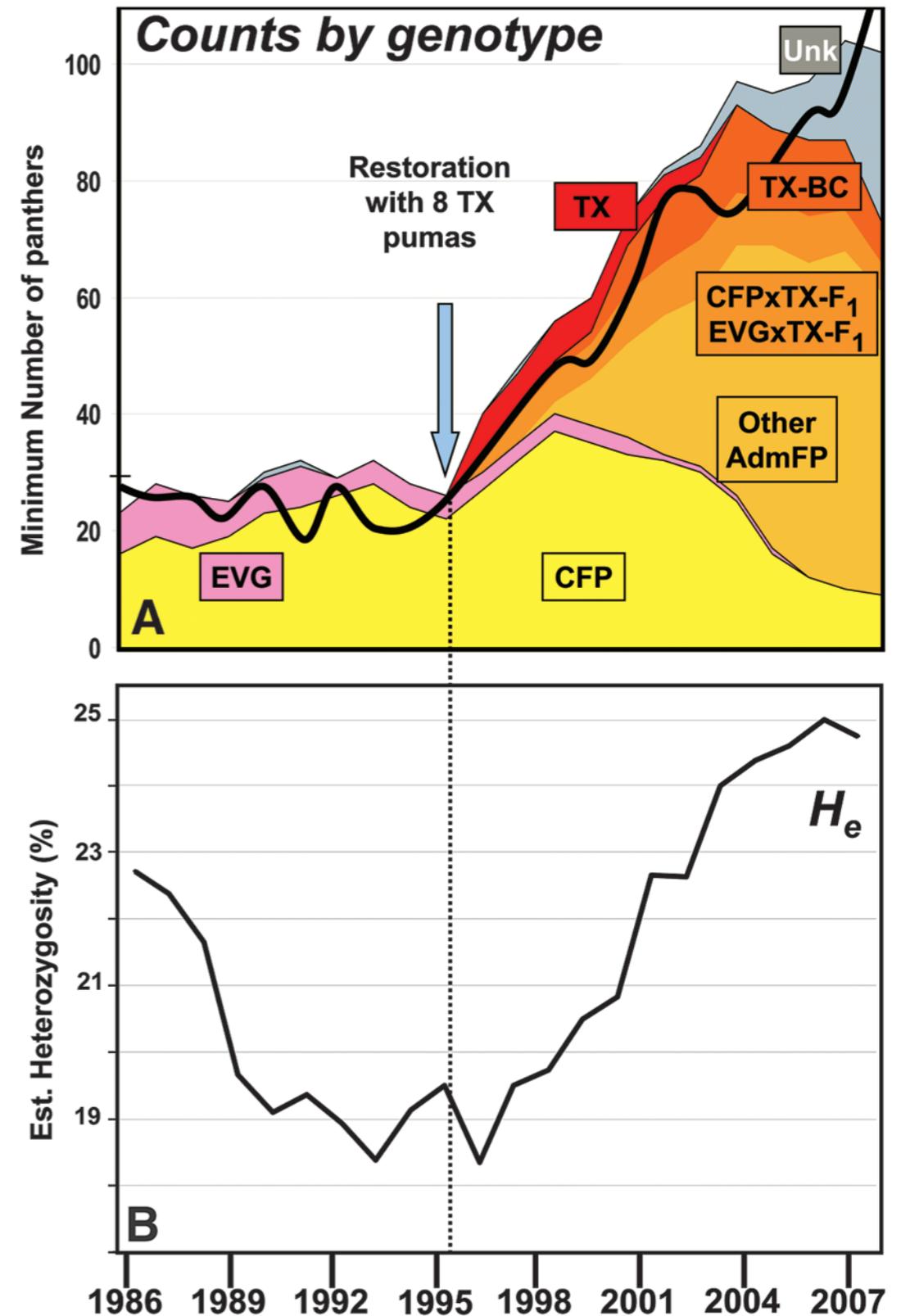
Many individuals with morphological/physiological defects: kinked tail, cowlick, sperm defects, heart defects (evidence of inbreeding depression)



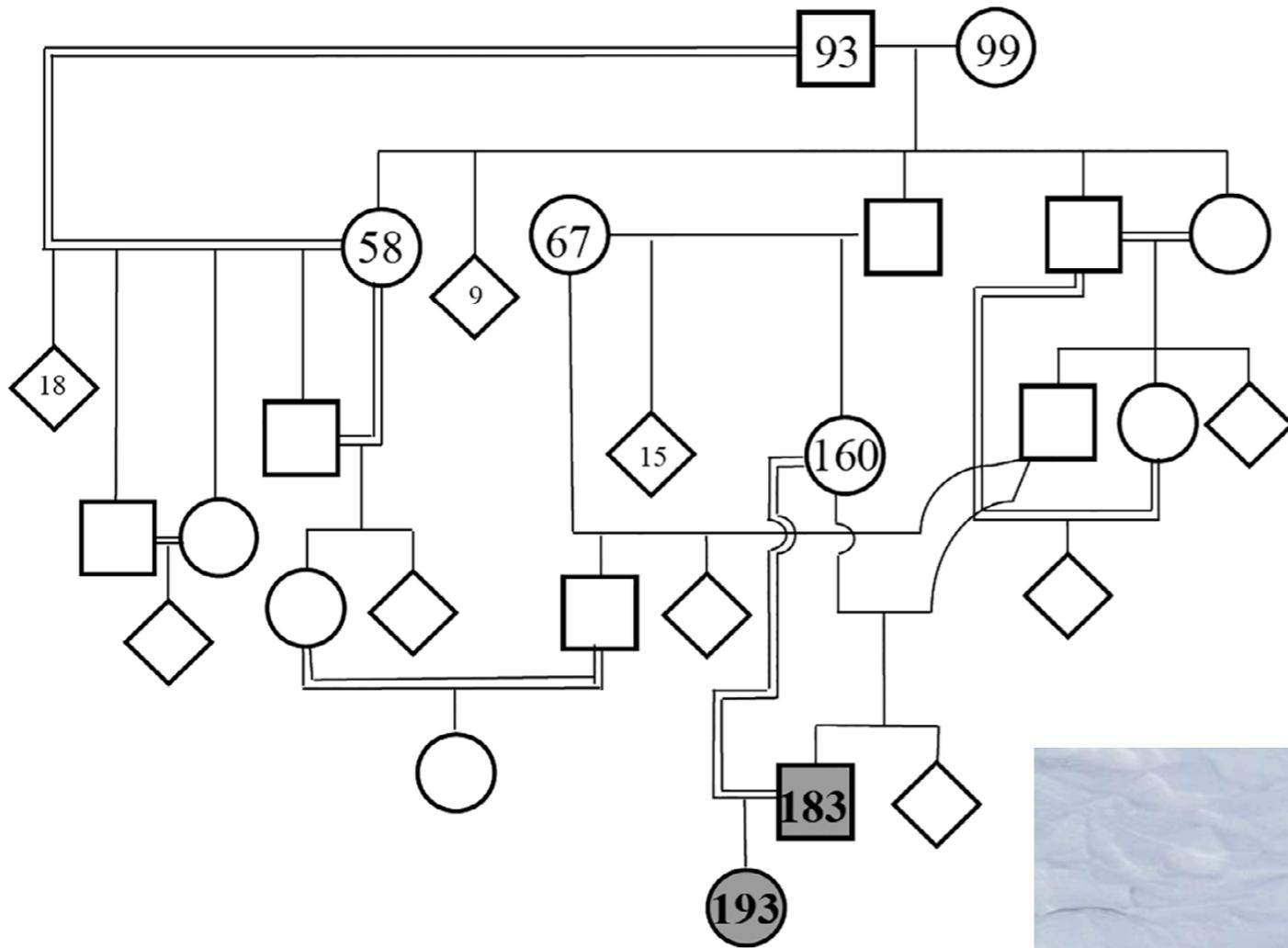
# Genetic rescue of the Florida Panther

Introduction of 8 females from Texas in 1995 led to:

- increased abundance
- increased heterozygosity
- higher survival of hybrid individuals
- reduced prevalence of phenotypic characters associated with inbreeding depression



# Temporary rescue of Isle Royale wolves



# Potential problems with genetic rescue

**Outbreeding depression:** reduced fitness of hybrids due to maladaptation to local environmental conditions or genetic incompatibilities.

- More of a concern if populations are very divergent (greater chance of local adaptation)

# Potential problems with genetic rescue

**Outbreeding depression:** reduced fitness of hybrids due to maladaptation to local environmental conditions or genetic incompatibilities.

- More of a concern if populations are very divergent (greater chance of local adaptation)

**Genetic swamping:** Loss of evolutionary lineages / locally adapted alleles due to gene flow.

- Gene flow increases genetic similarity of populations (genetic homogenization), which can reduce species-level genetic diversity

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