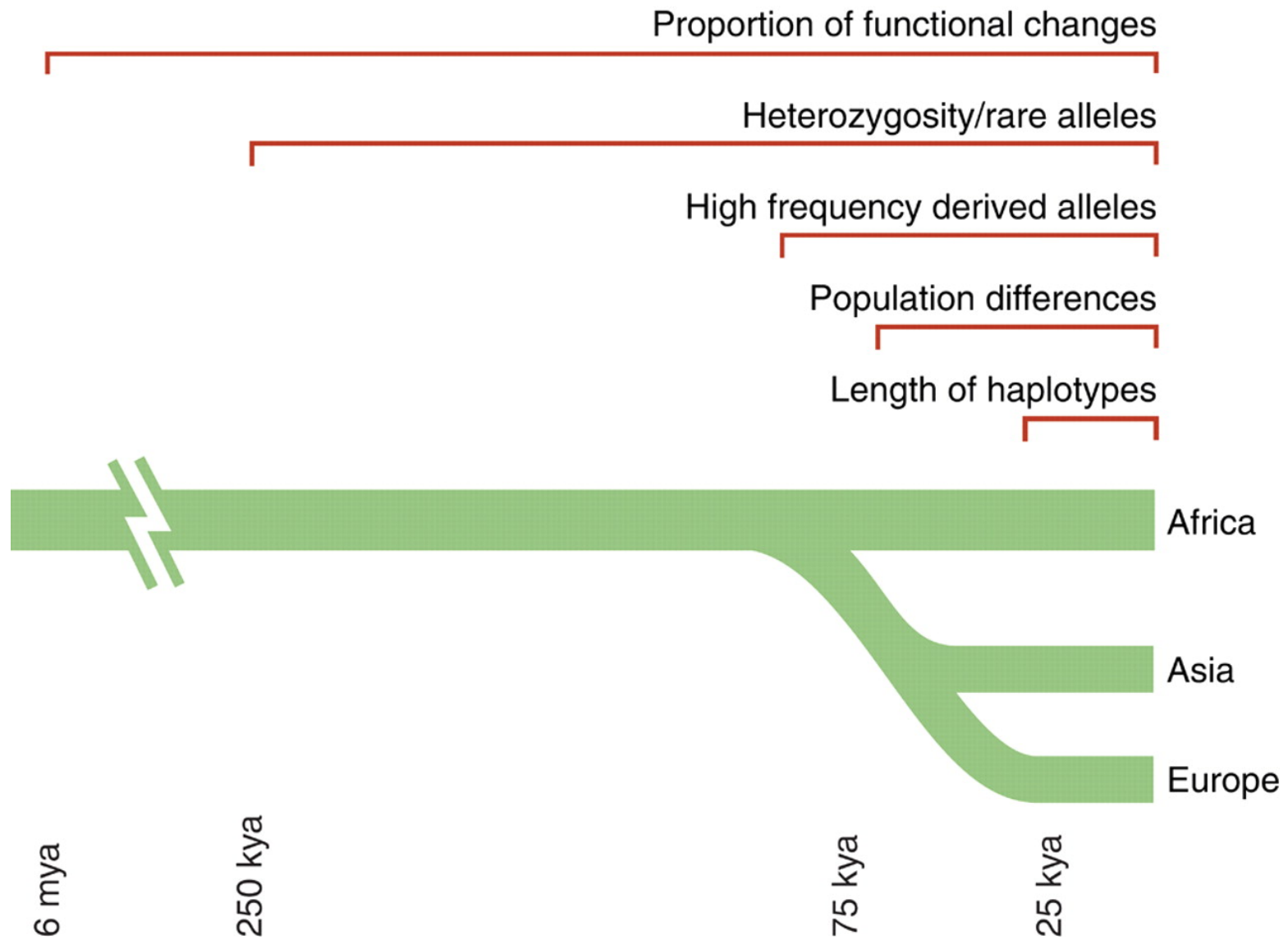


**Fig. 1. Time scales for the signatures of selection.**

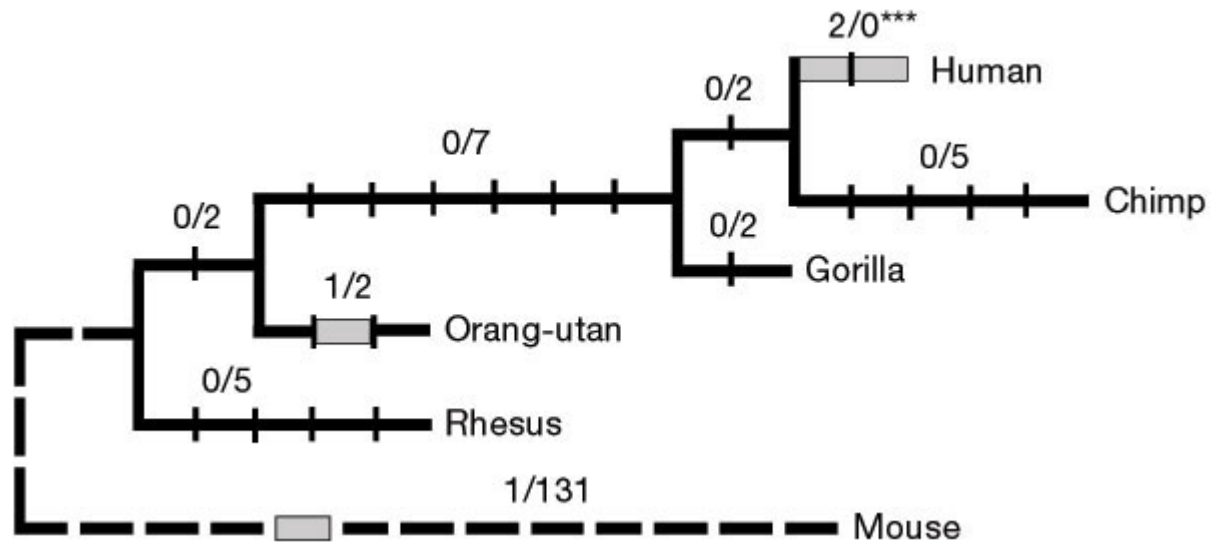


P C Sabeti et al. Science 2006;312:1614-1620



## **Proportion of functional changes**

- Positive selection may favor many alleles, not just one**
- This can be detected by a large number of coding changes relative to neutral changes in the gene.**



Foxp2 gene

Has high functional differences.

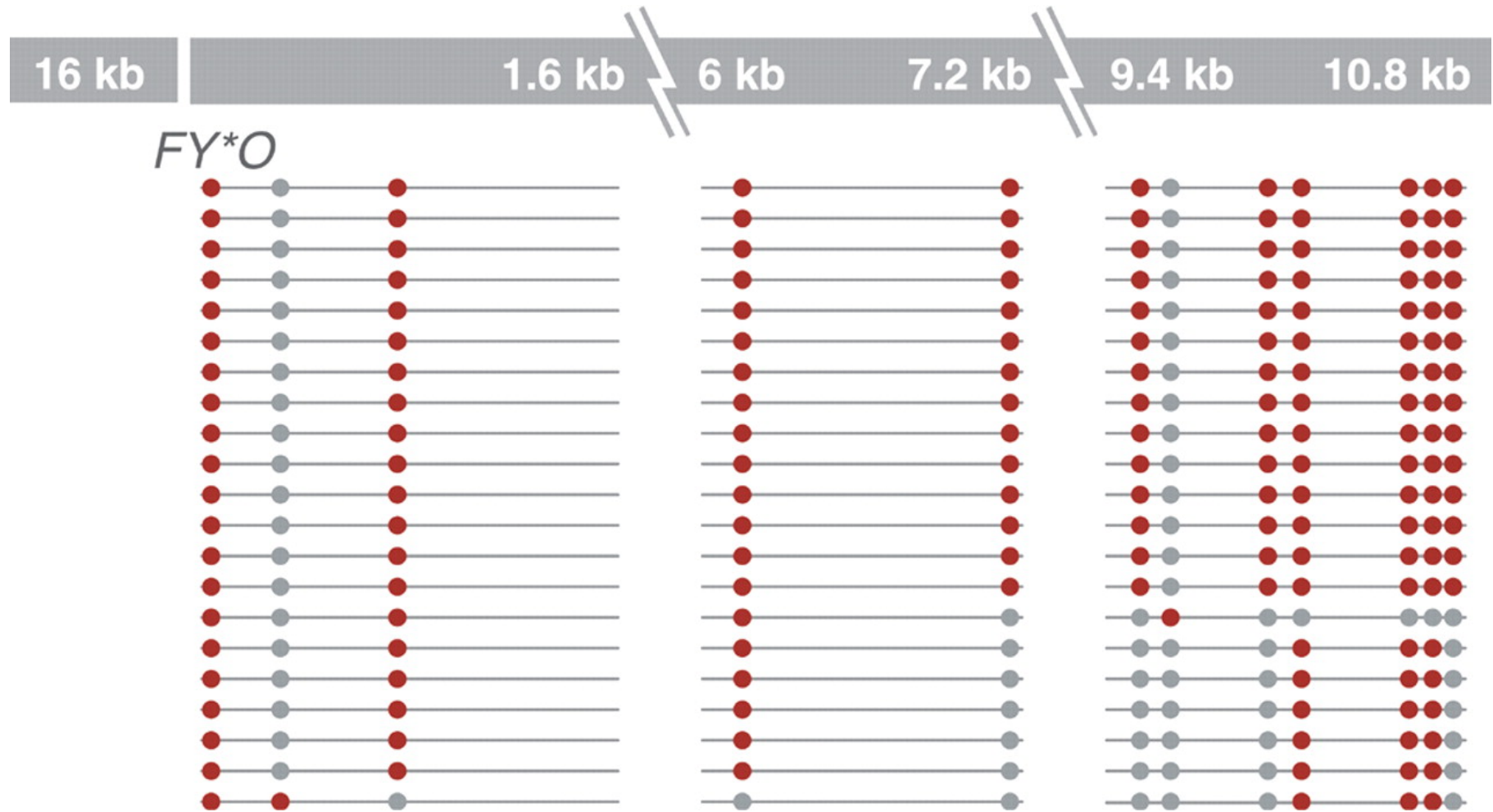
Bars represent nucleotide changes. Grey bars indicate amino-acid changes

## **High Frequency Derived Alleles**

**Most new alleles are at low frequency**

**One way for a derived allele to become high frequency is to be linked to an allele undergoing positive selection**

Excess of high-frequency derived alleles at the Duffy red cell antigen (FY) gene  
Resistance to malaria.

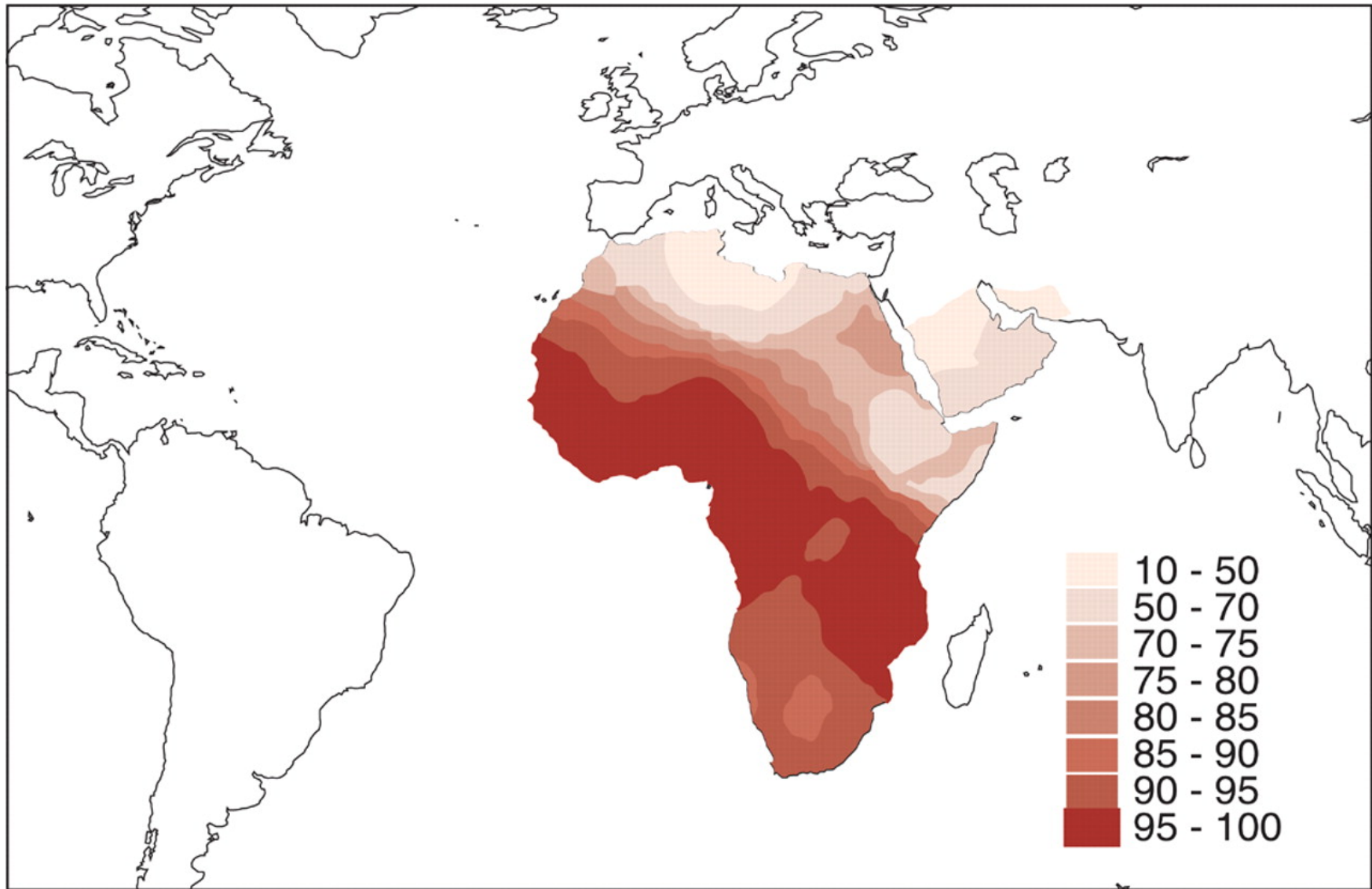


Red – derived. Gray – ancestral (Chimp)

P C Sabeti et al. Science 2006;312:1614-1620



## Extreme population differences in FY\*O allele frequency.

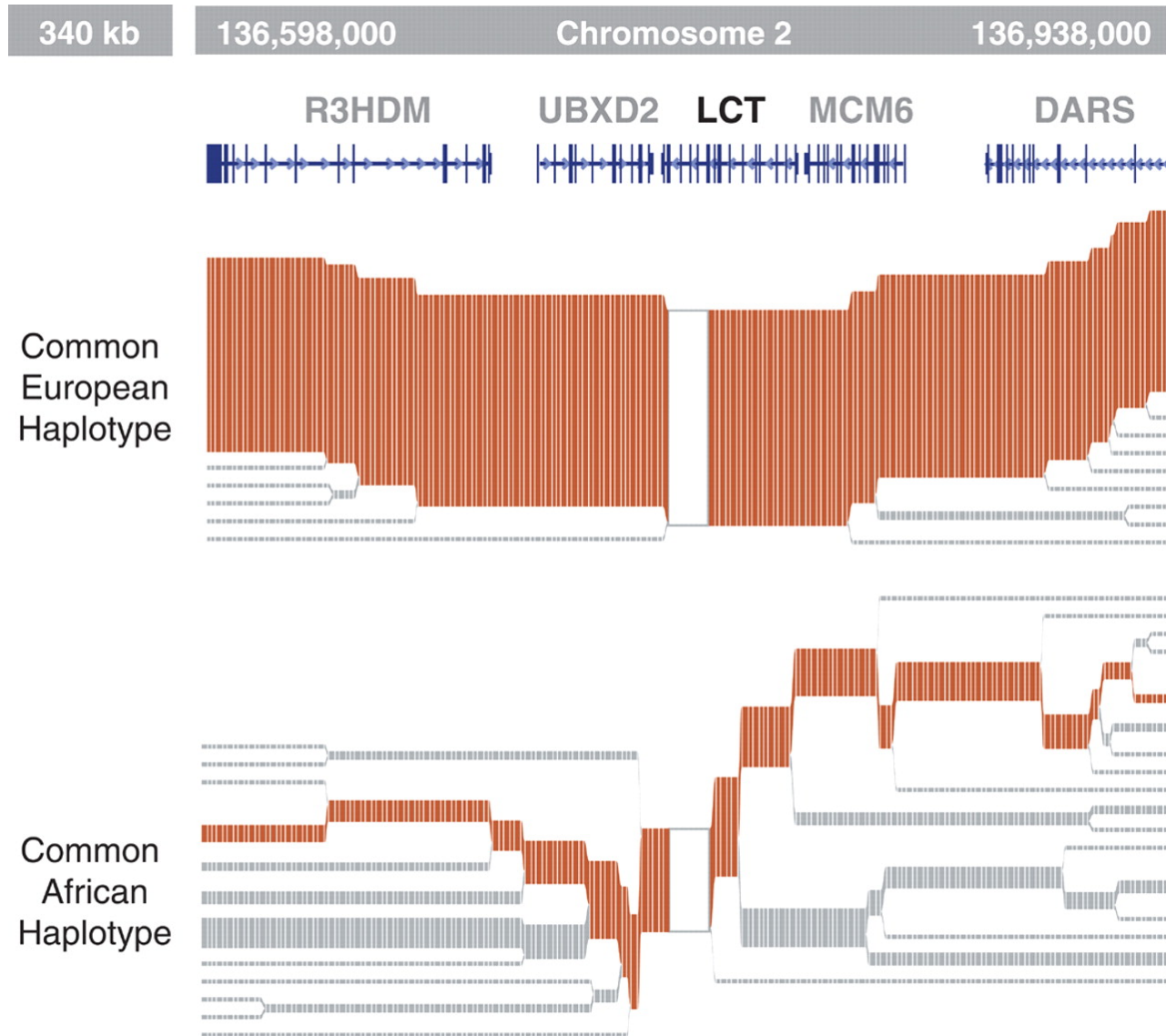


P C Sabeti et al. *Science* 2006;312:1614-1620

# Long Haplotype



# Long haplotype surrounding the lactase persistence allele.



P C Sabeti et al. Science 2006;312:1614-1620



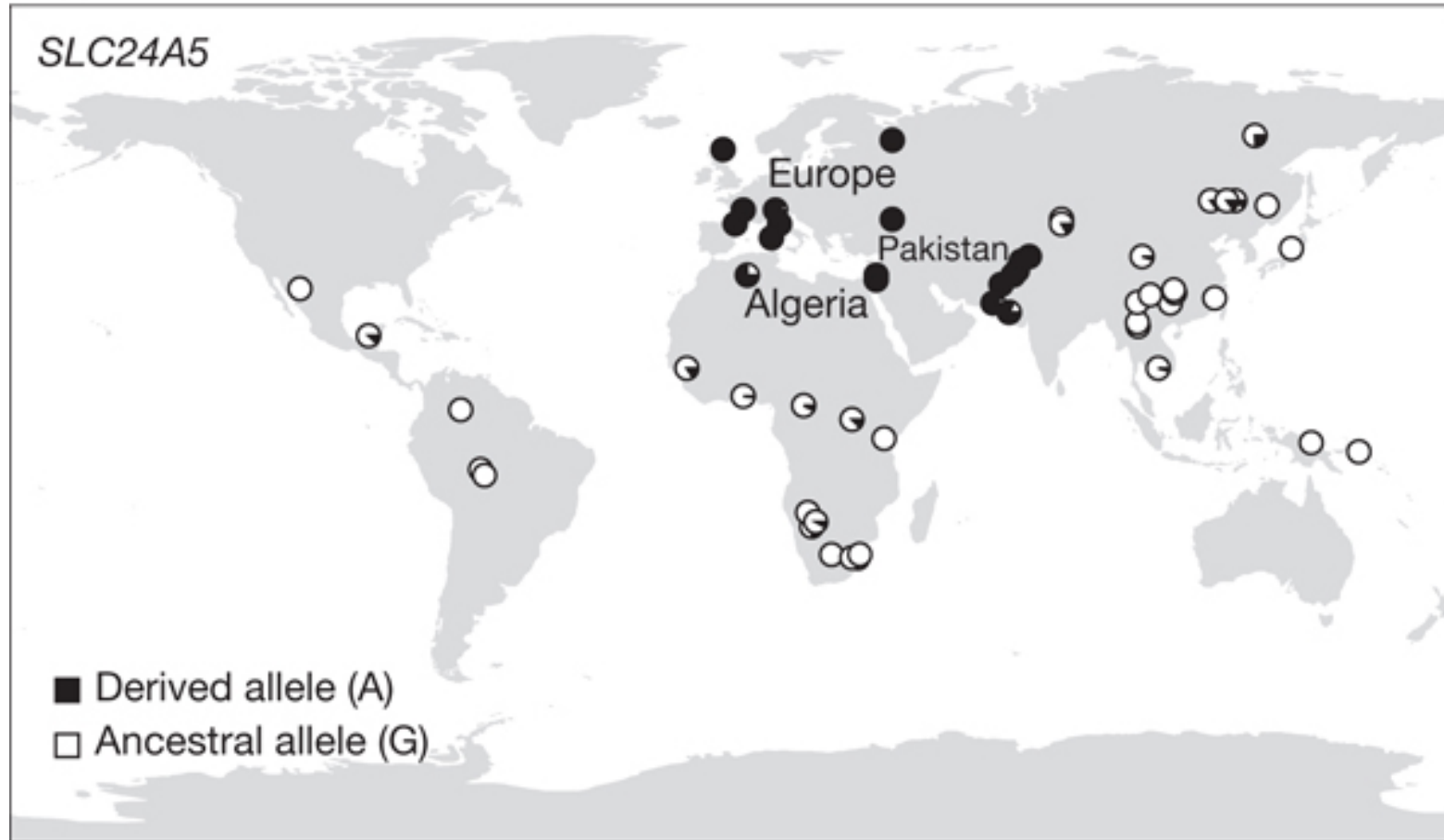


# Europeans

## Lactase

a mutation in a regulatory region near the gene for lactase (LCT) that allows lactose tolerance to persist into adulthood. This particular variant was apparently selected in parts of Europe after the domestication of cattle.

**a**



SLC24A5 involved in skin color. A111T .

SLC45A2: Also, an L374F substitution is at 100% frequency in the European sample, but absent in the Asian and African samples.

# African American

Hemoglobin-B

Duffy antigen (FY)

-target of selection for malaria resistance

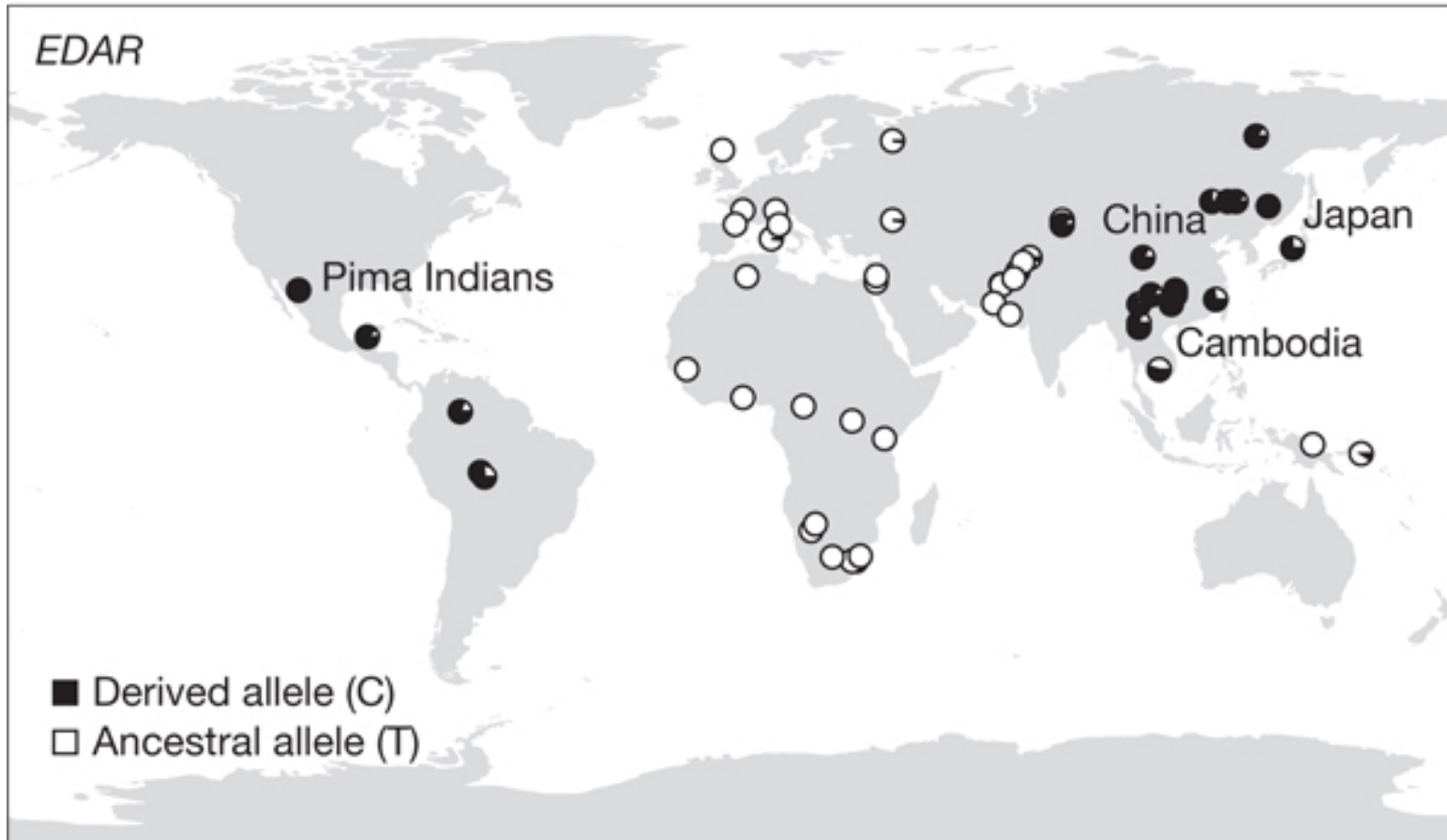
LARGE

*DMD*

- resistance to Lassa fever.

# Asian

**b**



# Asian

*EDAR* and EDA2R

Hair morphology

have a central role in generation of the primary hair follicle

A mutation encoding a V370A substitution in *EDAR* is near fixation in Asia and absent in Europe and Africa

100% in Pima Indians and in parts of China, and 73% in Japan