Lecture 9:

Population Genetics

Course 410

Molecular Evolution

"Nothing in **biology** makes sense except in the light of **evolution**"

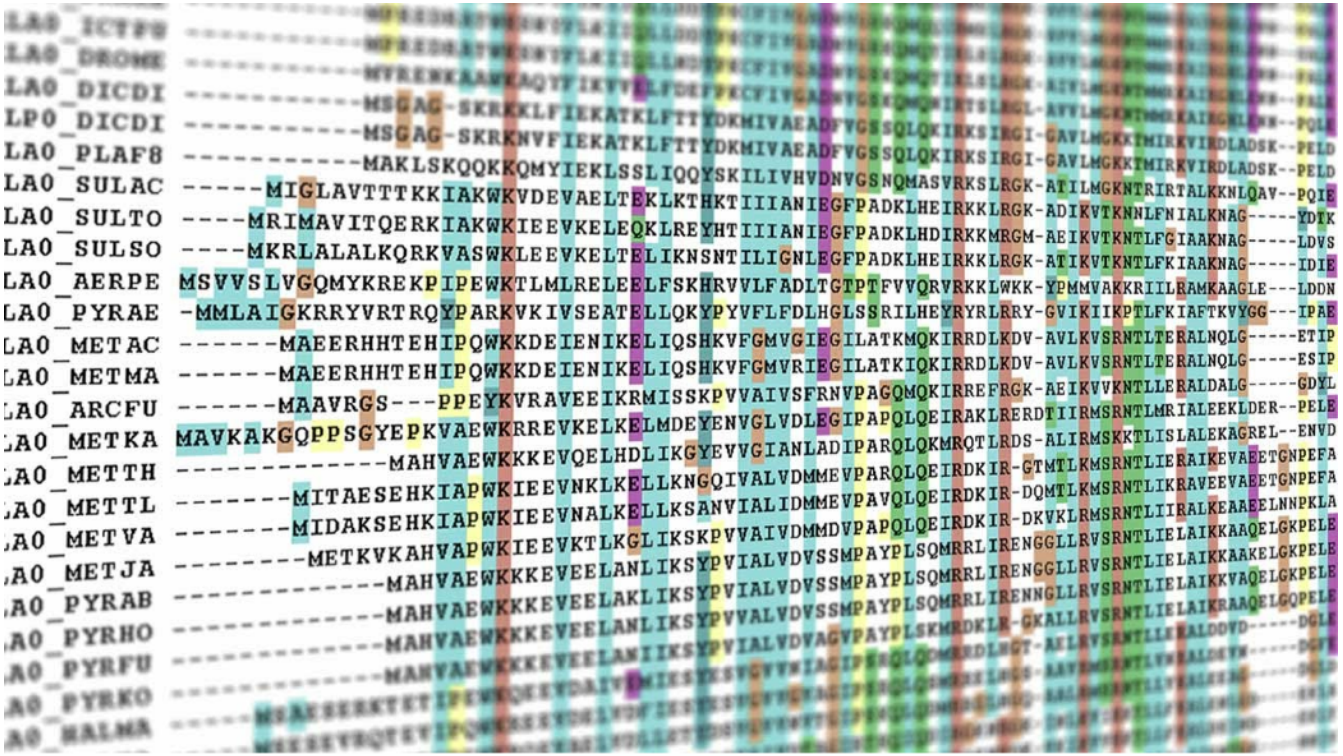
Theodosius Dobzhansky, 1973

"Nothing in **evolution** makes sense except in the light of **population genetics**"

Jeffrey Ross-Ibarra
(2010 citing his mentor)

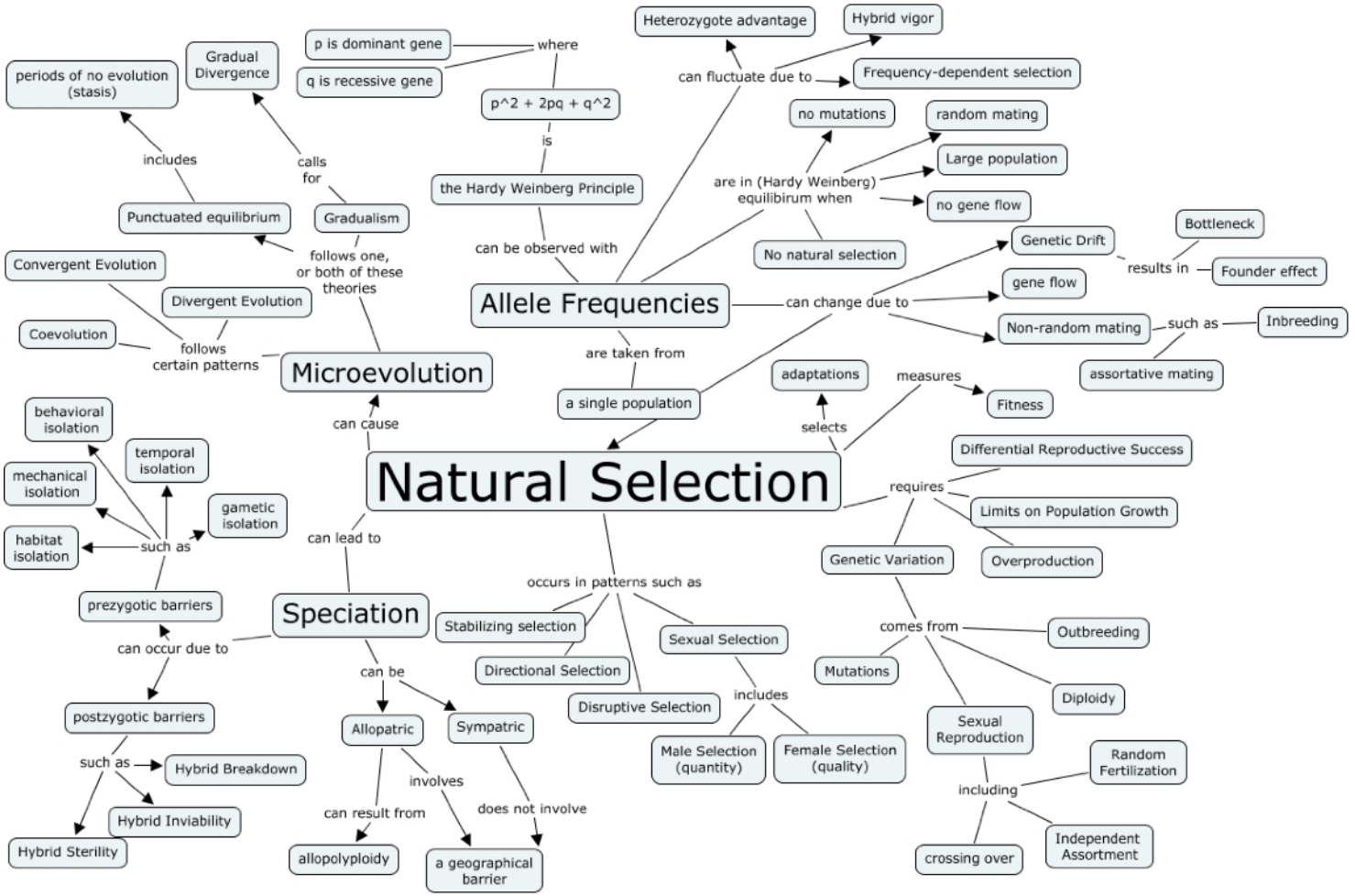
What is population genetics?

It is the study of the evolutionary historical record of a group of individuals documented in the DNA of their descendants

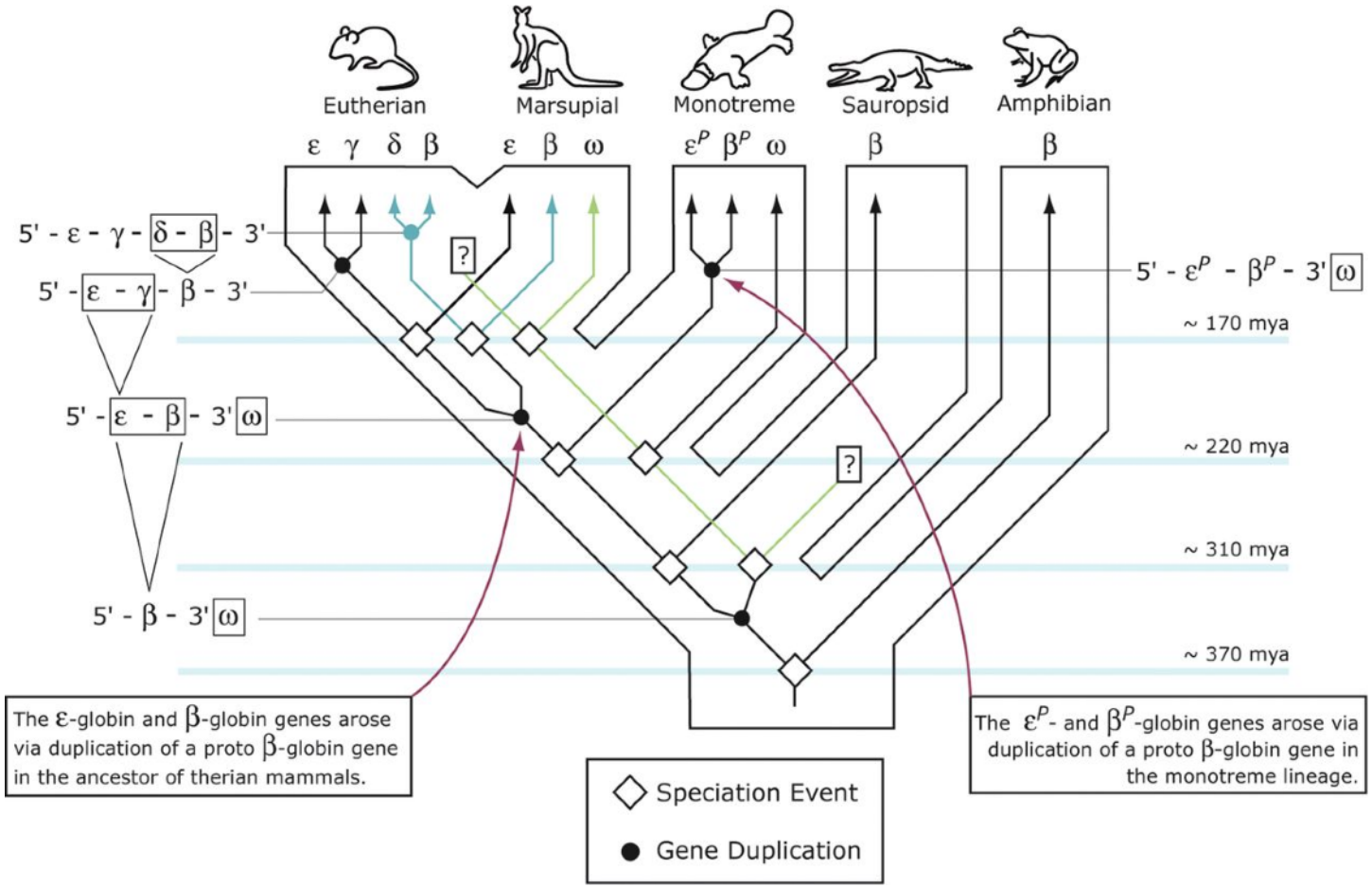


Why population genetics?

1) Understand and refine theory

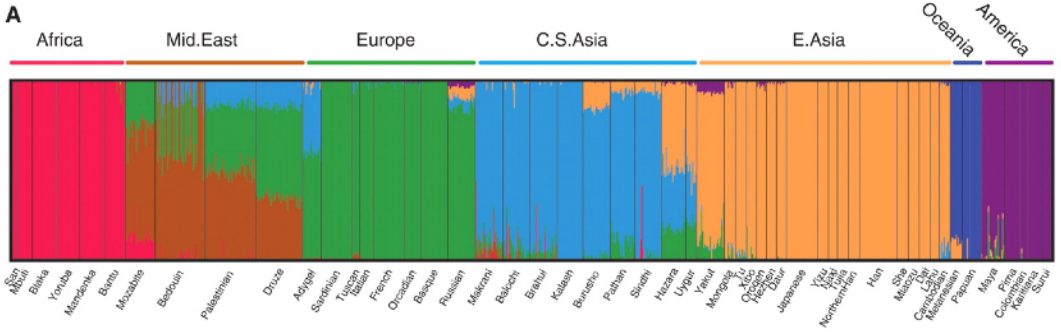


2) Understand the history of genes



Opazo, J. C., Hofmann, F. G. & Storz, J. F. (2008) Genomic evidence for independent origins of β-like globin genes in monotremes and therian mammals. PNAS (105): 1590–1595

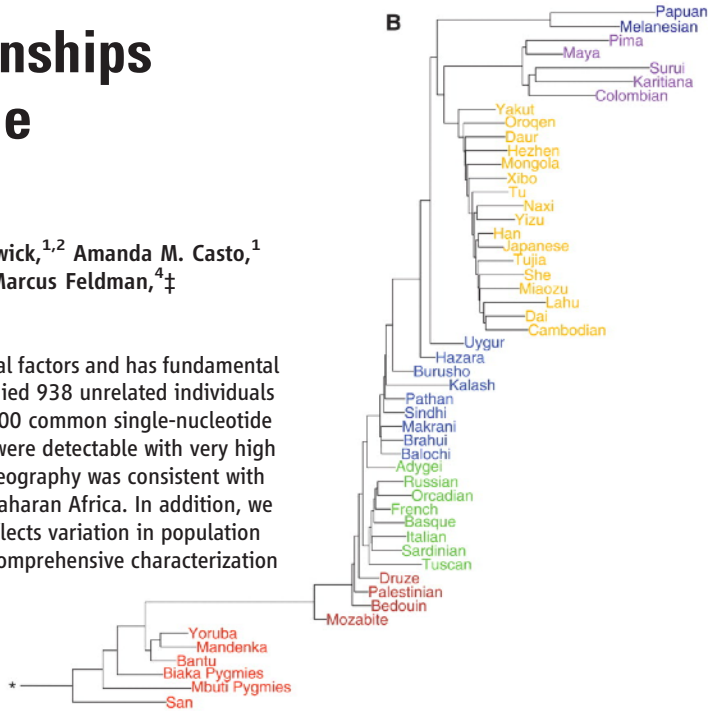
3) Understand the history of populations/ organisms



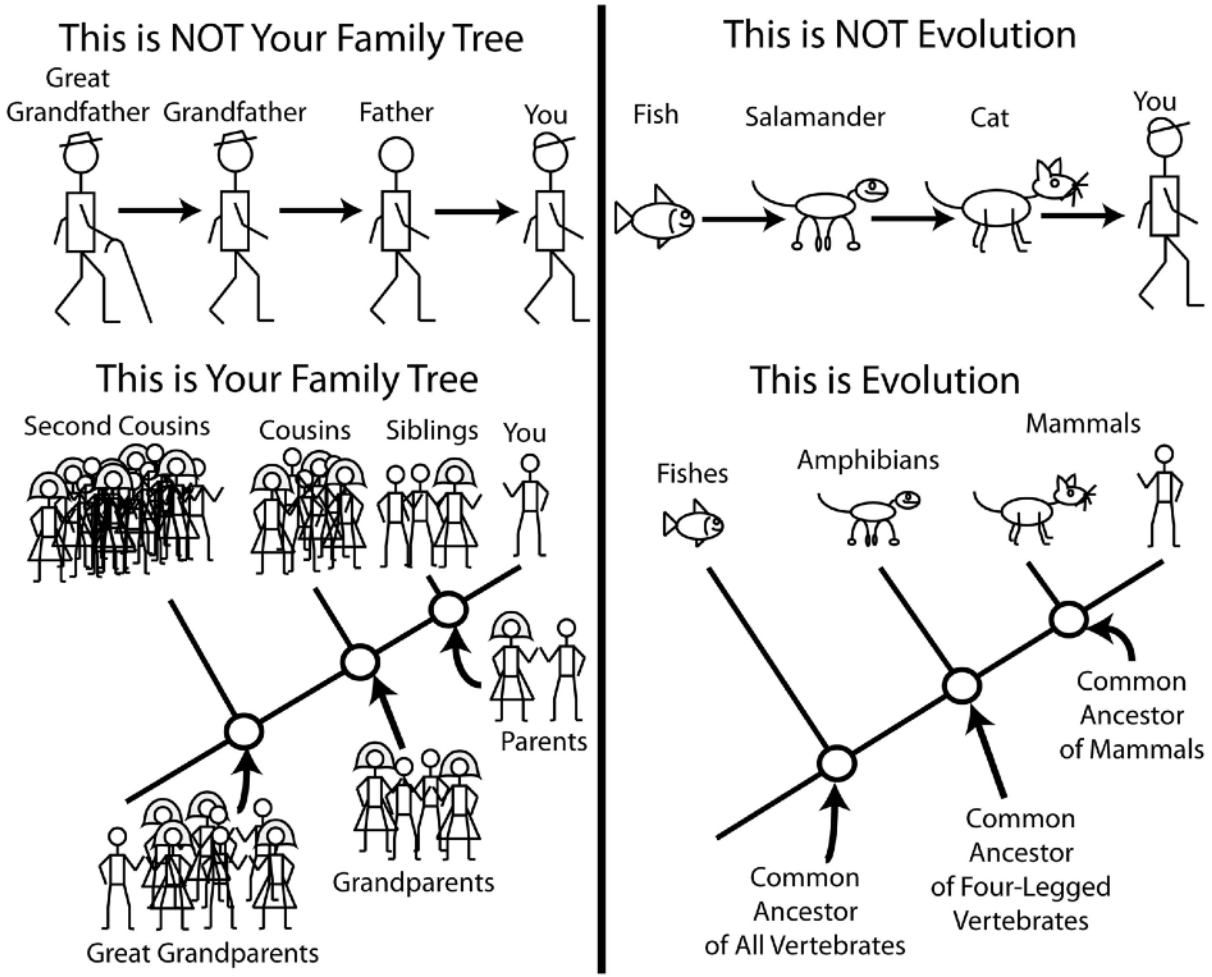
Worldwide Human Relationships Inferred from Genome-Wide Patterns of Variation

Jun Z. Li,[1,2]*† Devin M. Absher,[1,2]* Hua Tang,[1] Audrey M. Southwick,[1,2] Amanda M. Casto,[1] Sohini Ramachandran,[4] Howard M. Cann,[5] Gregory S. Barsh,[1,3] Marcus Feldman,[4]‡ Luigi L. Cavalli-Sforza,[1]‡ Richard M. Myers[1,2]‡

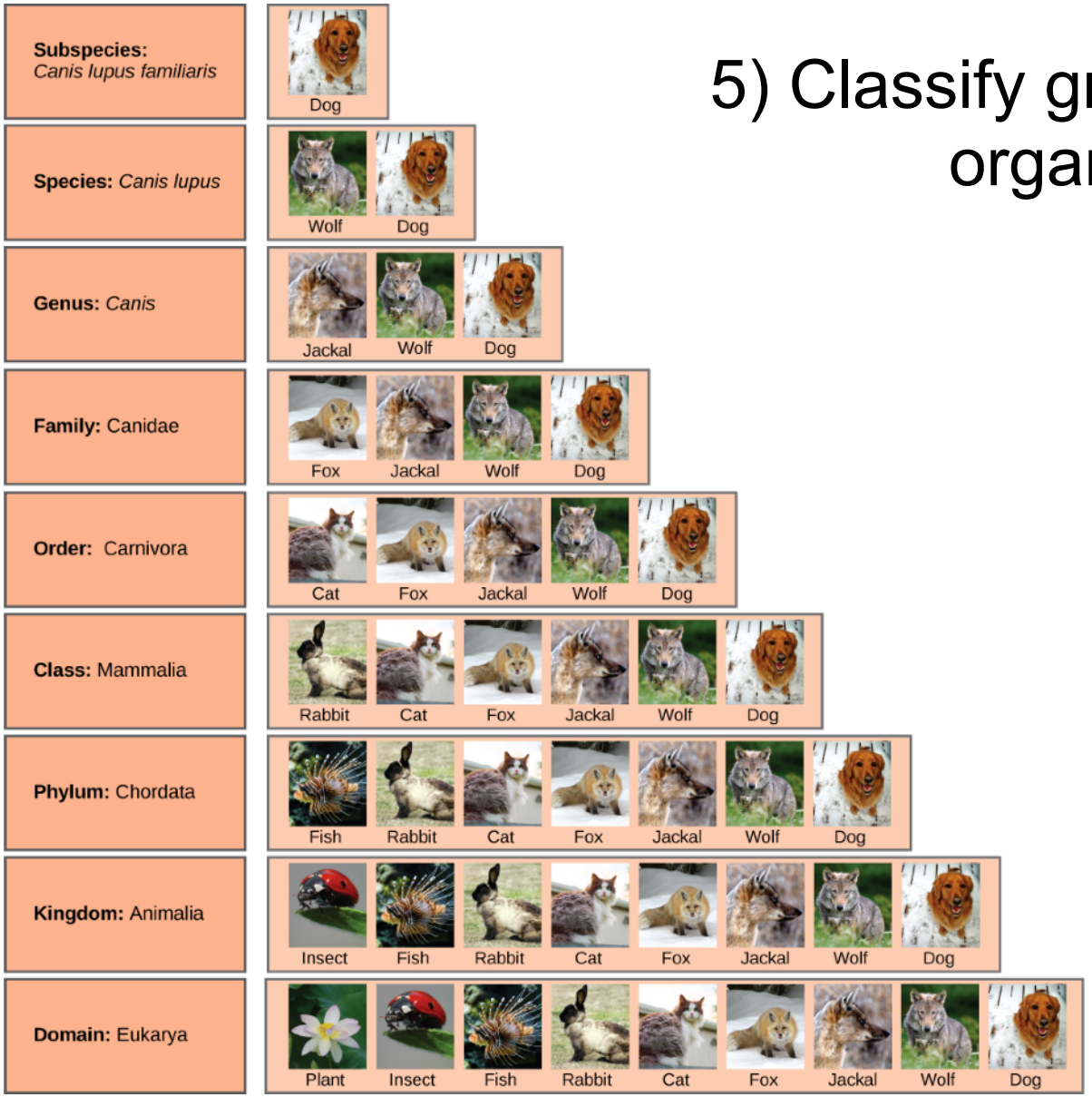
Human genetic diversity is shaped by both demographic and biological factors and has fundamental implications for understanding the genetic basis of diseases. We studied 938 unrelated individuals from 51 populations of the Human Genome Diversity Panel at 650,000 common single-nucleotide polymorphism loci. Individual ancestry and population substructure were detectable with very high resolution. The relationship between haplotype heterozygosity and geography was consistent with the hypothesis of a serial founder effect with a single origin in sub-Saharan Africa. In addition, we observed a pattern of ancestral allele frequency distributions that reflects variation in population dynamics among geographic regions. This data set allows the most comprehensive characterization to date of human genetic variation.



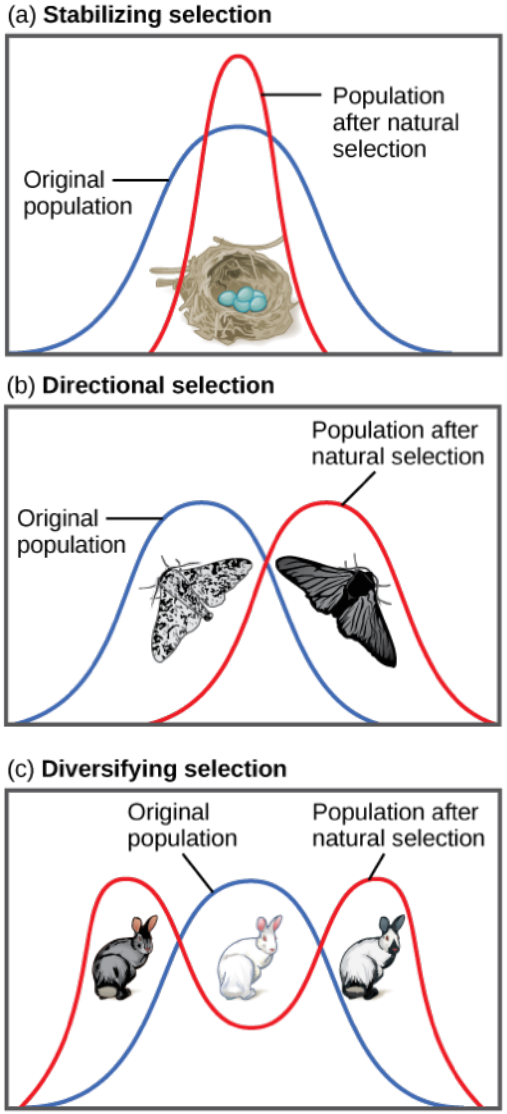
4) Understand the relationship between organisms

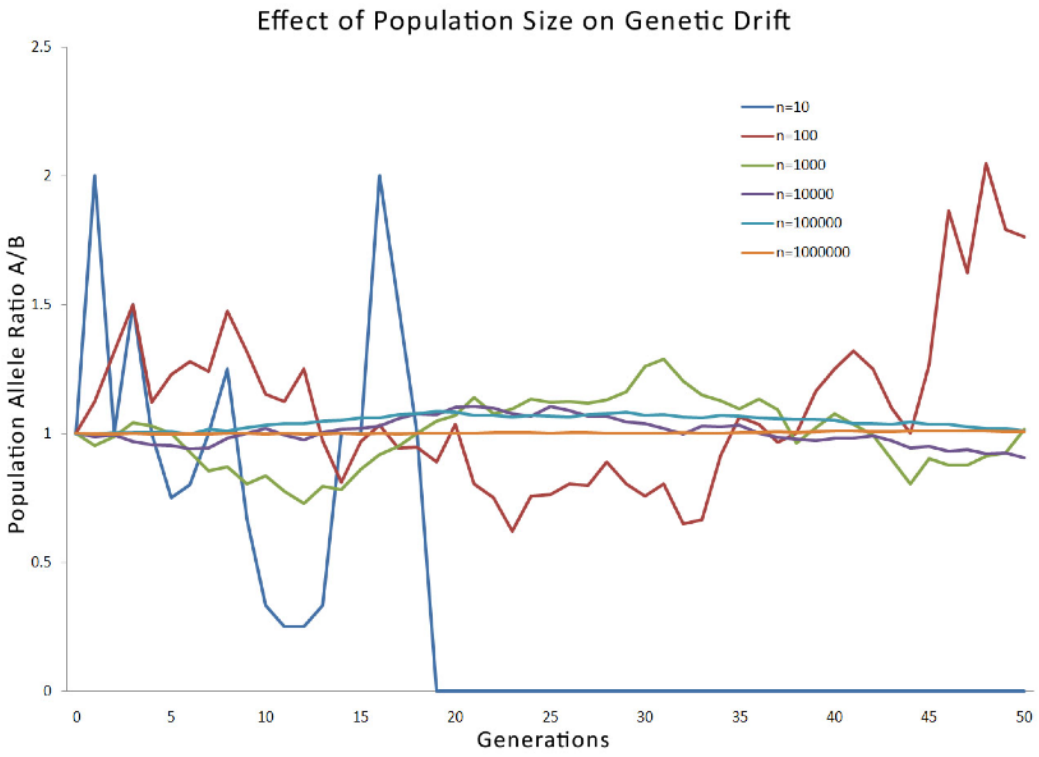


5) Classify groups of living organisms

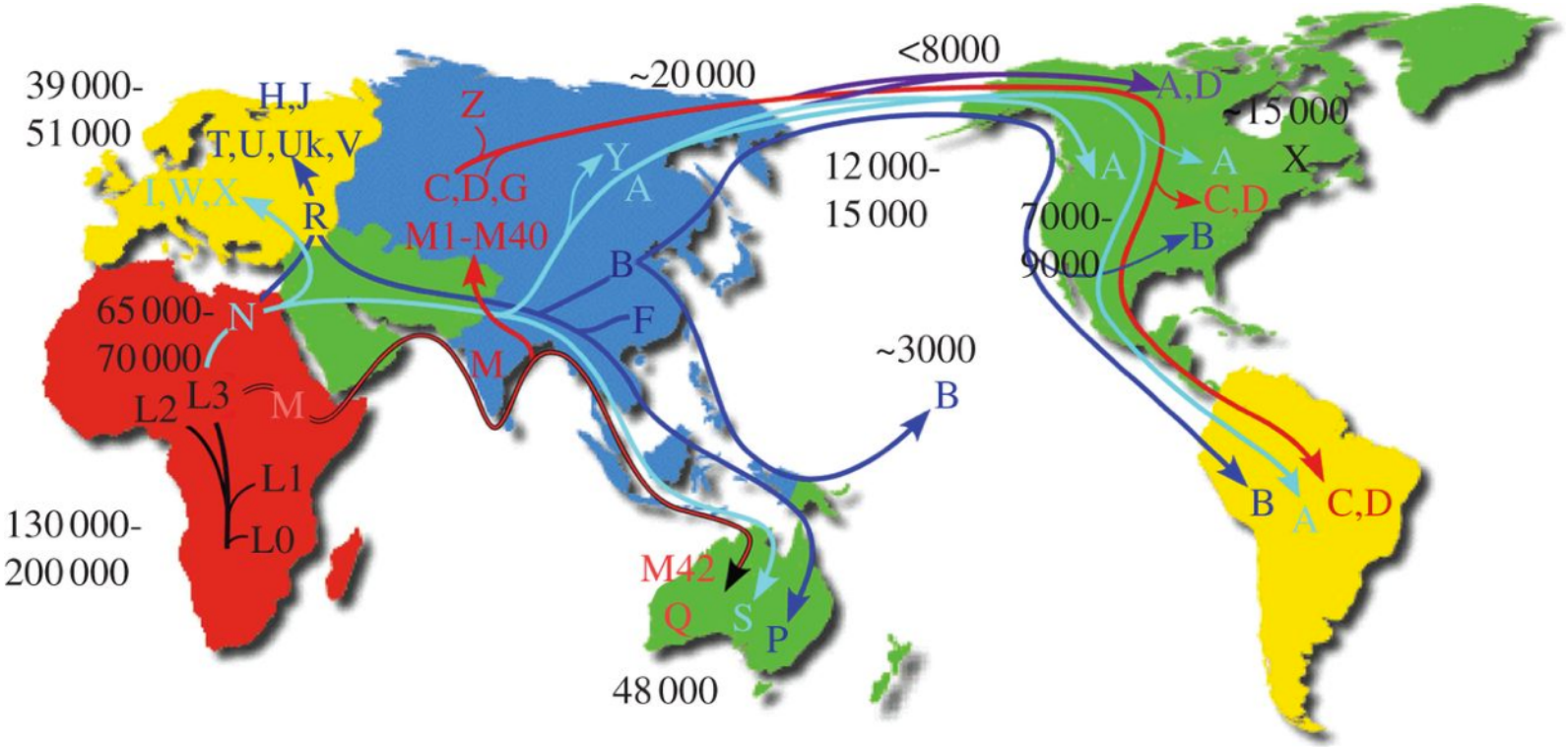


6) Understand the evolutionary forces that shape life forms





7) Reconstruct the history and the timing of evolutionary events



mutation rate = 2.2–2.9%/MYR
time estimates are YBP

8) Find cool stuff!

MELANESIAN BLOND HAIR IS CAUSED BY AN AMINO ACID CHANGE IN TYRP1

Naturally blond hair is rare in humans and found almost exclusively in Europe and Oceania. Here, we identify an arginine-to-cysteine change at a highly conserved residue in tyrosinase-related protein 1 (TYRP1) as a major determinant of blond hair in Solomon Islanders. This missense mutation is predicted to affect catalytic activity of TYRP1 and causes blond hair through a recessive mode of inheritance. The mutation is at a frequency of 26% in the Solomon Islands, is absent outside of Oceania, represents a strong common genetic effect on a complex human phenotype, and highlights the importance of examining genetic associations worldwide.

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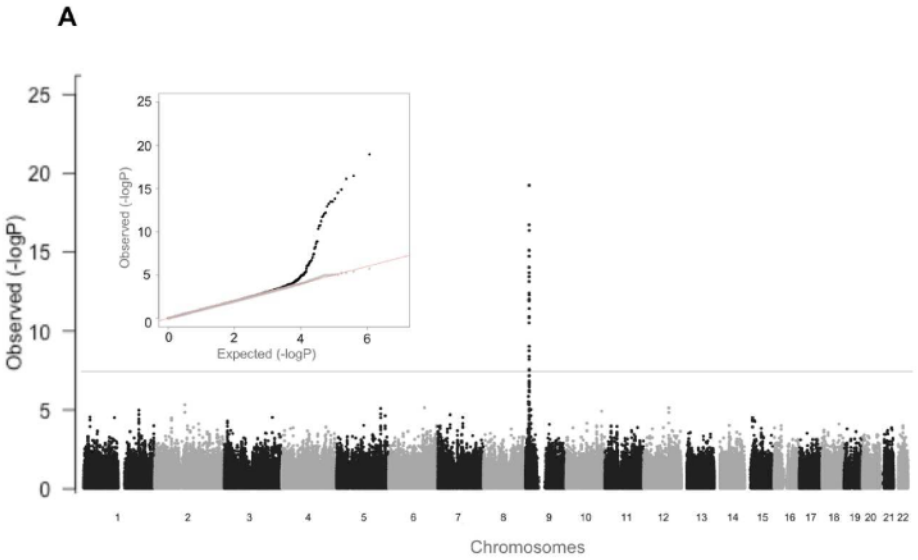
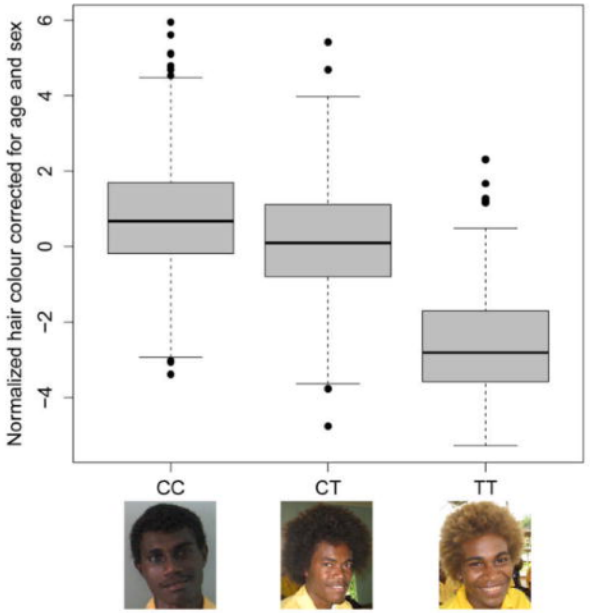


Genome-wide Association studies (GWAS)



Redefining evolution

Evolution is the change in allele frequency at a locus in a population over time

Change in frequency ⟶ Time

Allele frequencies?

populations?

Evolutionary forces act on individuals, correct?

Why populations?

Evolutionary forces act on the individuals but the affects are seen in populations in the form of changes in allele frequency

So what exactly do we do?

We study the effects of evolutionary forces on the frequency of a mutant allele

One more thing about the changes

Molecular changes vs. Morphological changes

Small effects on fitness vs. large effects on fitness

If they have small effects, then we will have to deal with chance

We need models to understand stochastic/random forces

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hhalhaddad@gmail.com