EBERHARD KARLS
UNIVERSITÄT
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Faculty of Philosophy
General Linguistics

Language Evolution WiSe 2023/2024

Lecture 6: Human Evolution IV
Dispersals

09/11/2023, Christian Bentz

Overview

Section 1: Recap

The Cell (Eukaryote)

The genetic information resides in

- the nucleus (**DNA**),
- mitochondria (**mtDNA**),
- chloroplasts (**cpDNA**).

Note: Chloroplasts only exist in plant cells for photosynthesis, i.e. are not relevant here.

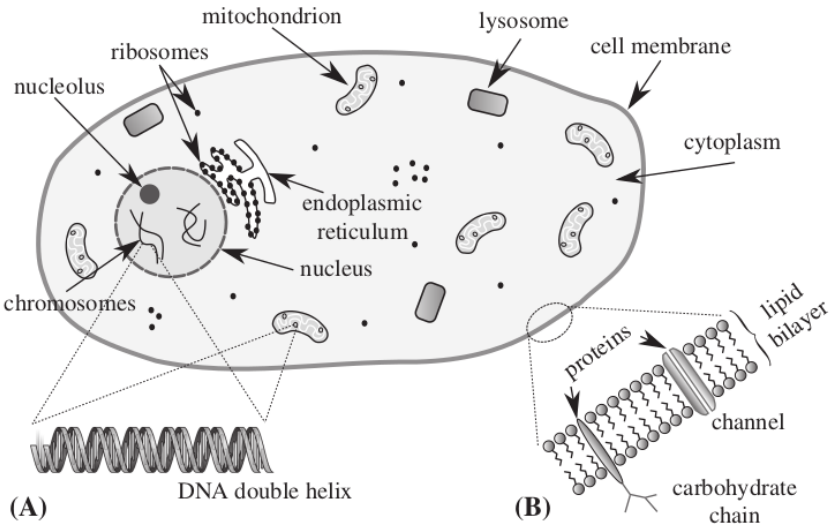


Figure 3.1 The structure of a generalized eukaryotic (animal) cell. The cell is bounded by a membrane (insert **B**) composed of two layers of lipids with various types of embedded proteins allowing matter, energy and information exchanges between the cell and its environment. In the cytoplasm there are various structures such as the nucleus containing most of the genetic material (DNA double helix, insert **A**) structured into several discrete linear chromosomes, mitochondria (with their own tiny genetic material structured as a circular molecule), lysosomes, the endoplasmic reticulum and ribosomes.

Dediu (2015). An introduction to genetics for language scientists, p. 47.

Mitosis (DNA Replication)

A cell can divide itself in a process called **mitosis**). This creates a new cell with identical DNA (bare any mutations).

Dediu (2015). An introduction to genetics for language scientists, p. 52.

For a video on DNA replication see: https://www.youtube.com/watch?v=TNKWgcFPHqw&t=15s

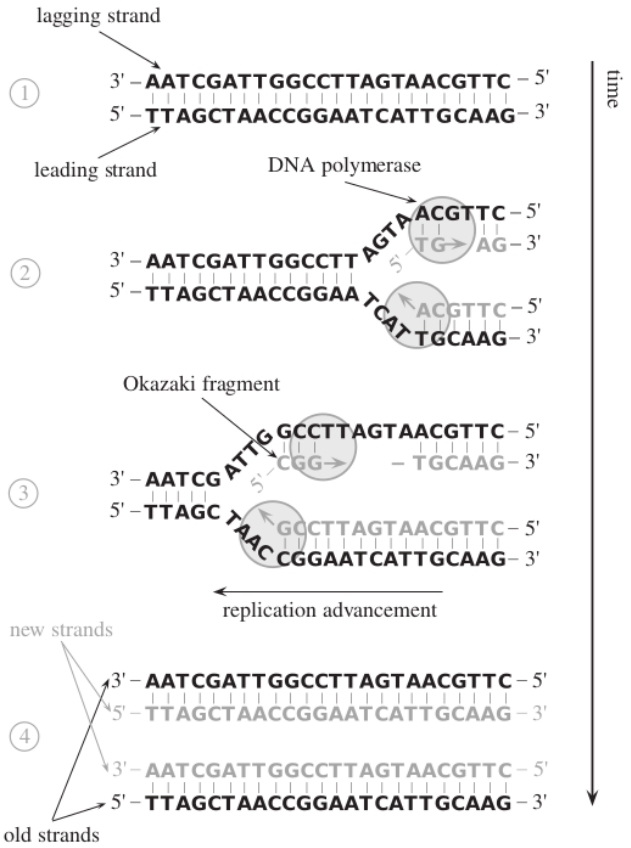


Figure 3.3 A simplified depiction of DNA replication. In black are the old DNA strands and in grey the new ones. The grey circles represent DNA polymerase while grey arrows stand for the direction in which the new DNA strands grow. Time step 1 shows the old DNA double-stranded molecule before replication begins, while time steps 2 and 3 show the advance of the replication fork and the elongation of the leading and lagging strands. The final time step (4) shows the two daughter double-stranded DNA molecules, each composed of one old (black) and one new (grey) DNA strand.

Genes

The term **gene** refers to regions on the DNA strand whose information is transcribed and translated into amino acids. The regions relevant for transcription are called **exons** (**ex**pressed regi**ons**), and are interspersed with **introns** (**intr**agenic regi**ons**).

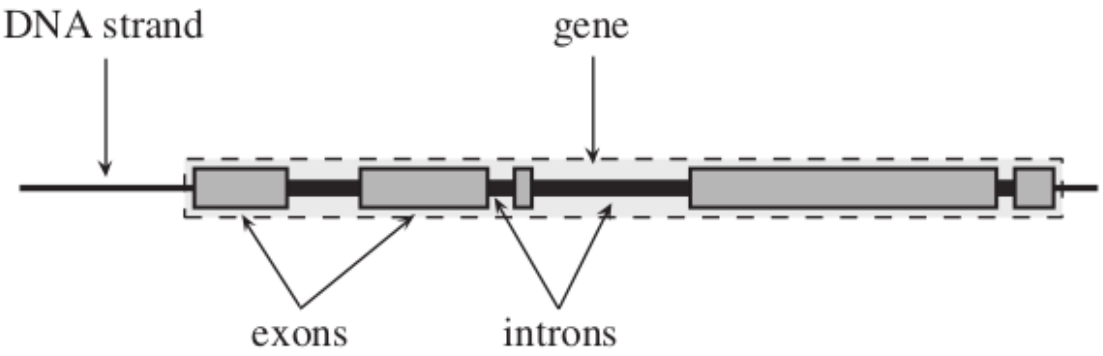


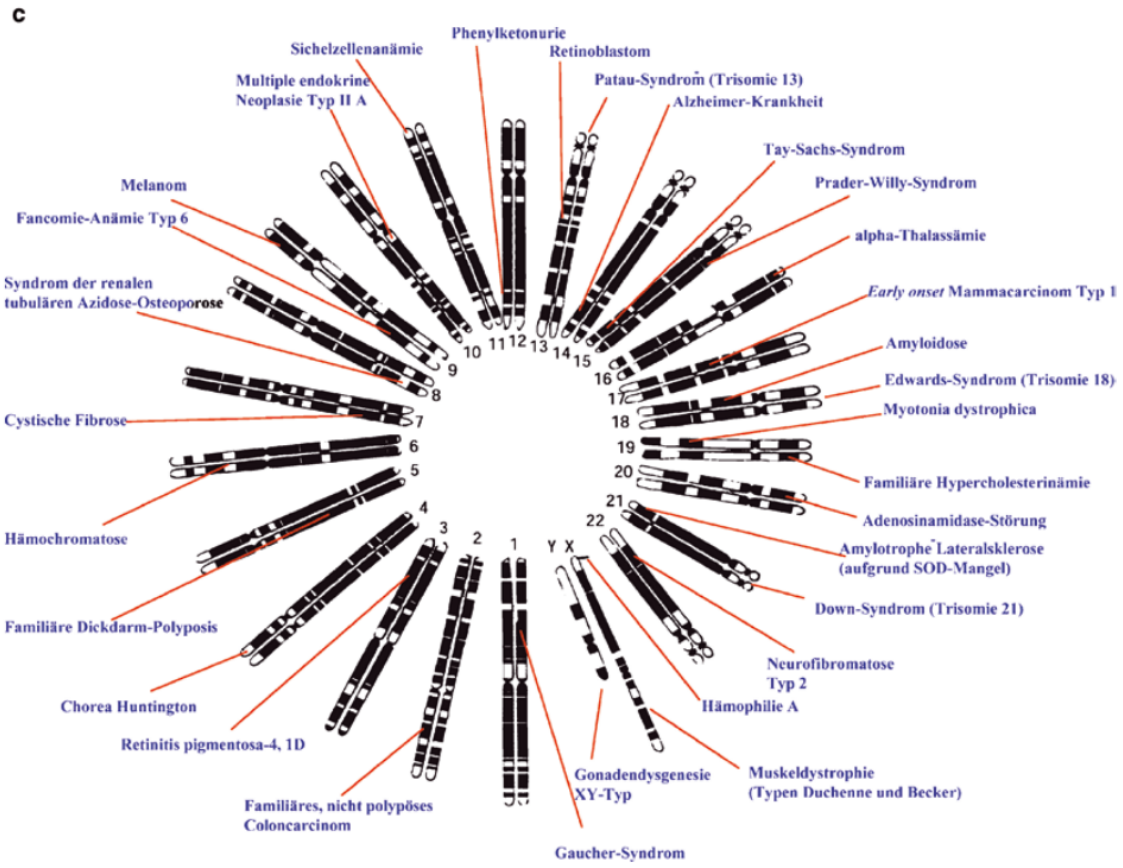
Figure 3.14 Schematic representation of a gene composed of several exons and introns.

Dediu (2015). An introduction to genetics for language scientists, p. 68.

Chromosomes

There are overall **46 human chromosomes**, coming in **23 pairs**. The pairing of chromosomes is the reason why they are called **diploid**. 22 of these pairs are called *autosomes* (numbered 1 to 22 in decreasing order of size), and the last pairs is are the **sex chromosomes (X, Y)**.

Dediu (2015). An introduction to genetics for language scientists, p. 53.



Storch et al. (2013). Evolutionsbiologie, p. 257.

Sexual Reproduction

In the process of **fertilization** a sperm and an ova fuse and produce a so-called *zygote*, which is then again **diploid**. There are $2^{23} \times 2 = 16,777,216$ different possible unique zygotes. One of these will develop into offspring.

Dediu (2015). An introduction to genetics for language scientists, p. 57.

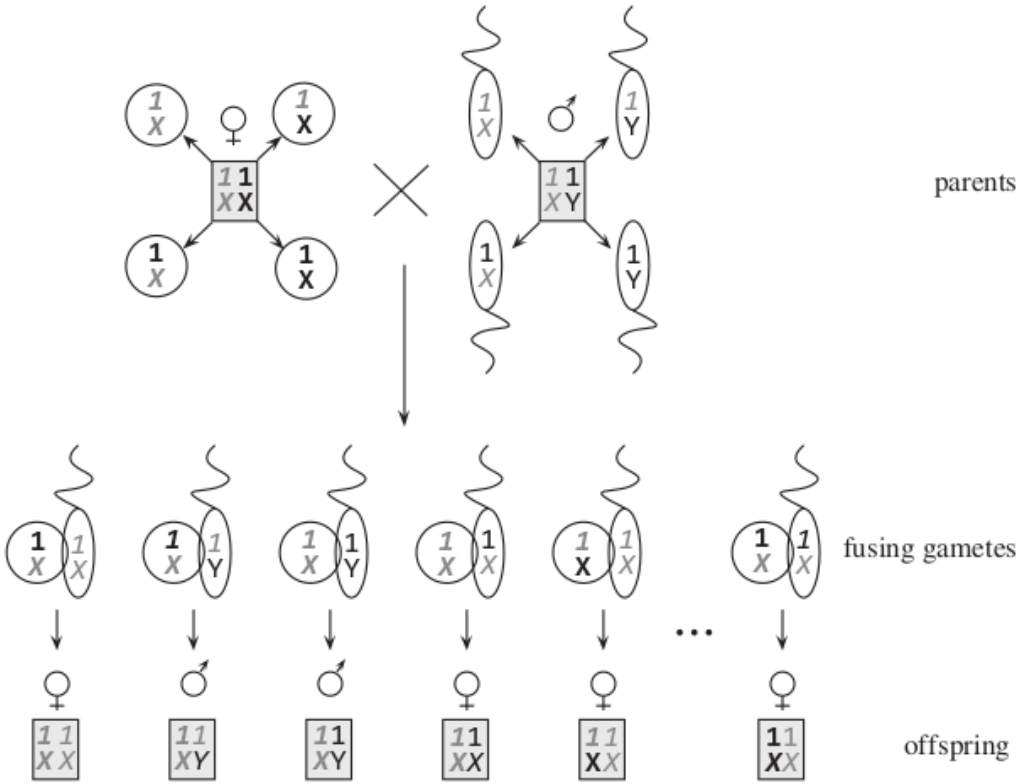


Figure 3.7 The parents (male and female, top) produce gametes which fuse (middle) to produce offspring (bottom). Here we focus on autosome 1 and on the sex chromosomes. For illustration purposes, the female parent's chromosomes are **bold** while the male parent's chromosomes are regular, and within each, one member of the pair is *grey italic* and the other black upright. Only six of the possible 16 zygotes are shown.

Gene Translation

The mRNA is translated (in blocks of three bases) into **20 amino acids**, the building blocks of proteins. **Proteins** then take on a multitude of functions in the body: transporting oxygen (haemoglobin), shaping cells and the body, movement of muscle fibres, signalling (neurotransmitters and hormones).

Dediu (2015). An introduction to genetics for language scientists, p. 64-67.

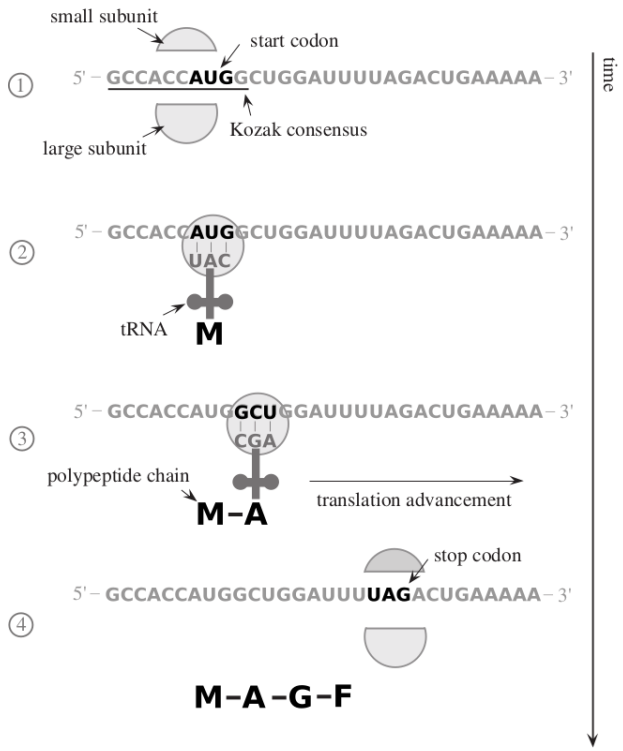


Figure 3.12 Schematic representation of translation. The ribosome is composed of two subunits (large and small) that assemble and begin the translation process with the start codon **AUG**. Translation advances in three-nucleotide units (codons) in the 5′ → 3′ direction (here, left → right) and consists in the elongation of the polypeptide chain by adding the amino acid carried by the tRNA corresponding to the current codon.

For a brief introduction to gene transcription and translation see video at:
https://www.youtube.com/watch?v=gG7uCskUOrA

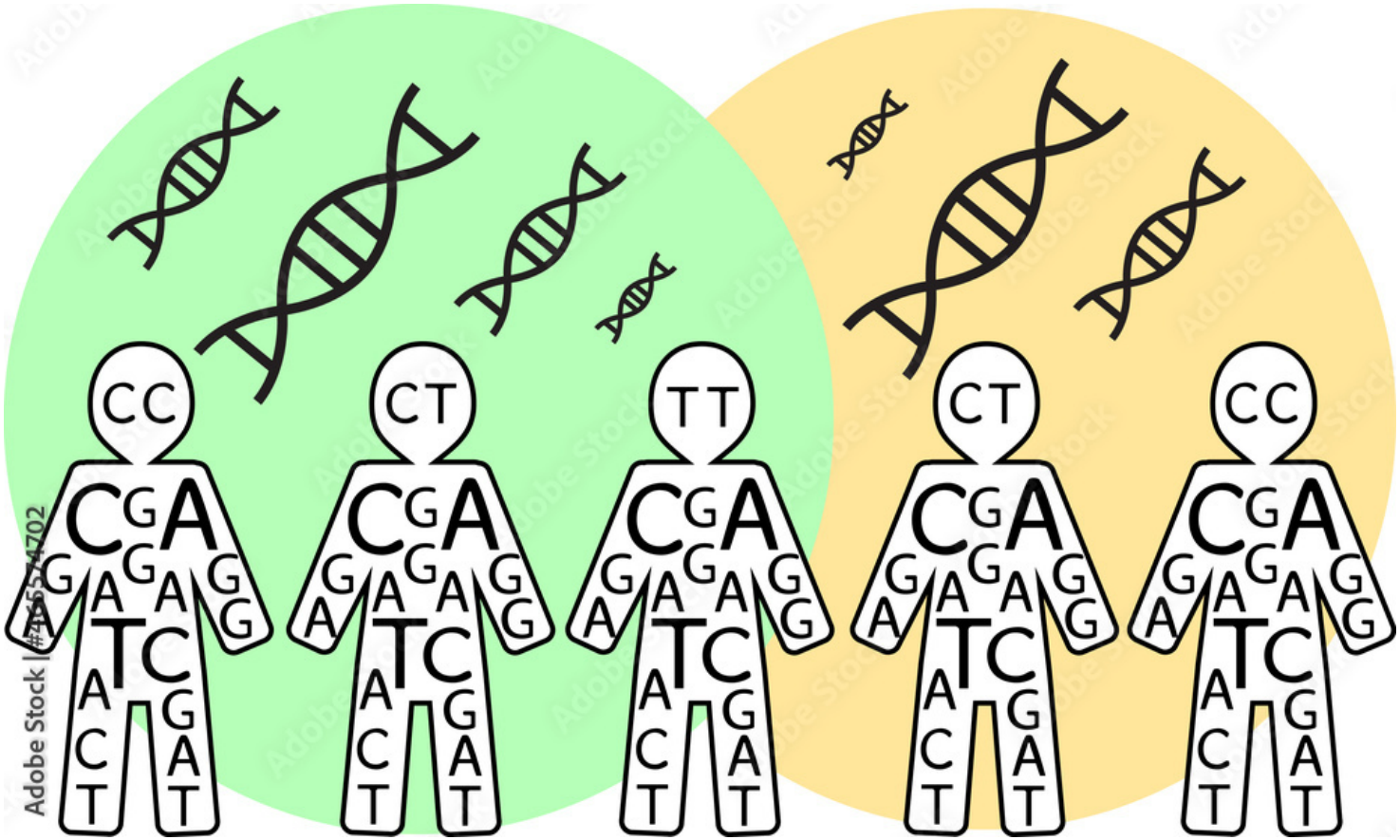
Types of Mutations

Mutations come in different types. Depending on the type and **locus** of a mutation, the actual impact on gene expression and the phenotype of an organism can be anything from negligible to fatal.

- **Point mutations**: a single nucleotide is replaced for another in an exon on the DNA strand (e.g. A $\rightarrow$ C).
- **Frameshift mutation**: A single nucleotide is deleted or inserted.
- **Chromosomal mutation**: Whole chromosomes or parts of chromosomes can be deleted, duplicated, inverted or translocated.

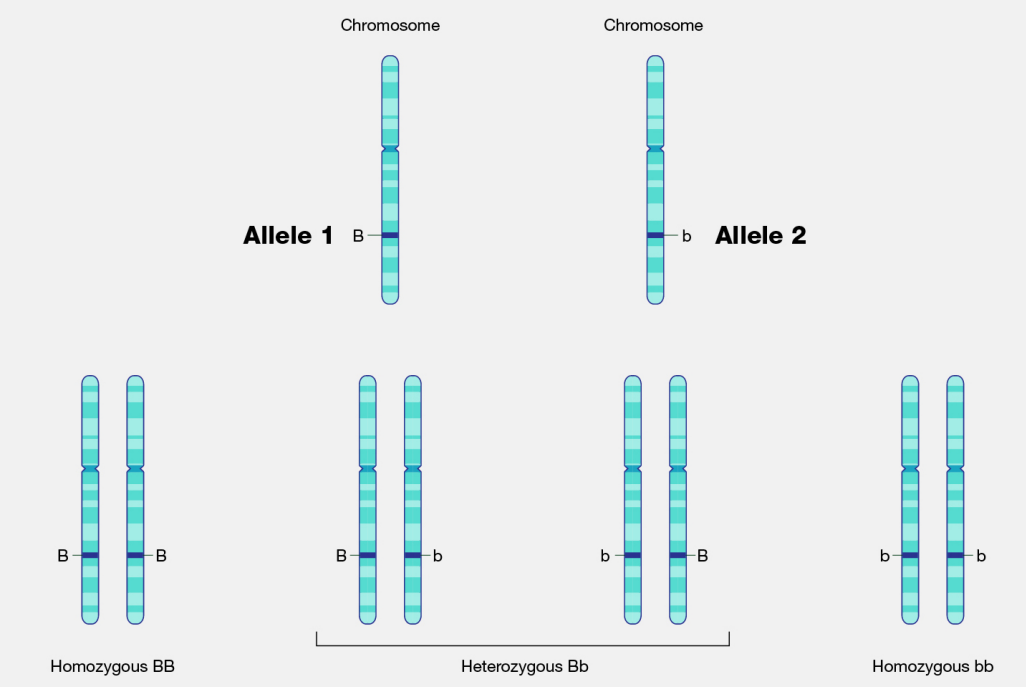
Section 2: Population Genetics

From Individuals to Populations



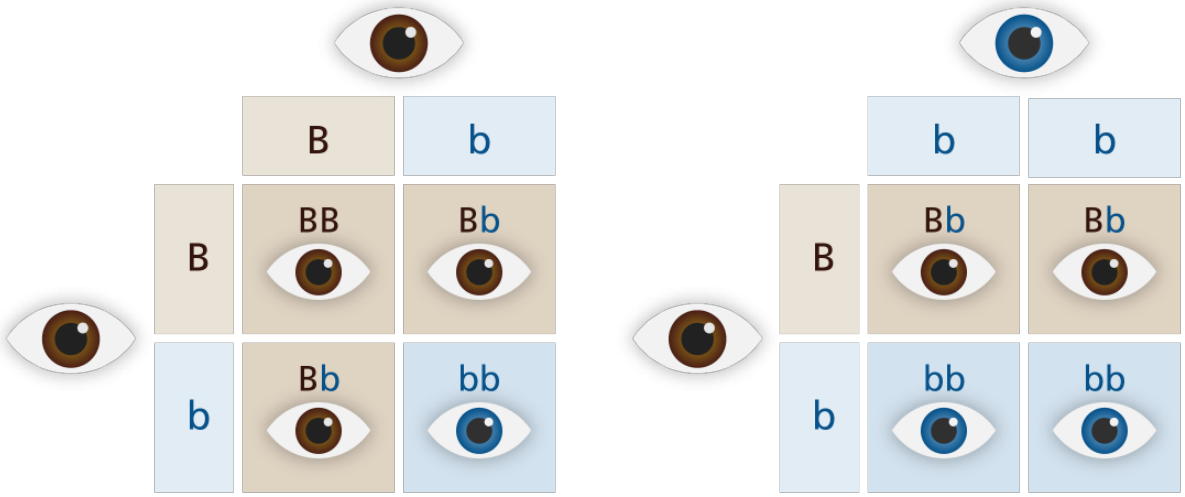
Reminder: Alleles

The term **alleles** refers to the alternative versions (two or more) of a DNA sequence at a genomic locus. This DNA sequence could be a single nucleotide or several nucleotides. If an individual has two identical allelels in the homologous chromosomes, this is called **homozygous**. If there are two different ones, this is called **heterozygous**.



Reminder: Alleles

Upper case letters are typically used for **dominant** alleles, and lower case letters for **recessive** alleles.



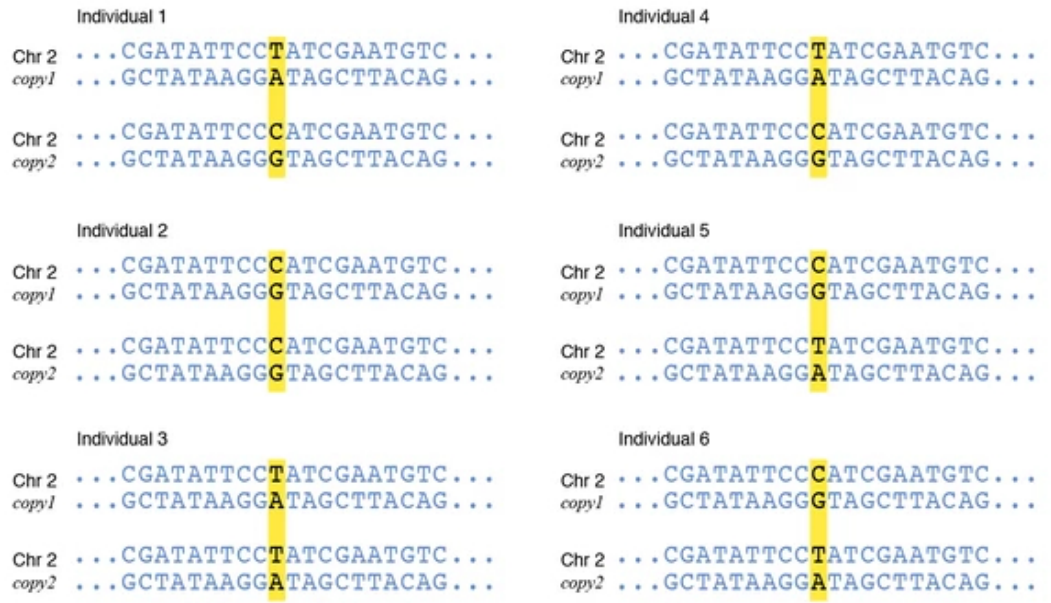
B - dominant brown eye allele
b - recessive blue eye allele

BB brown eyes
Bb brown eyes
bb blue eyes

Single Nucleotide Polymorphism (SNP)

If a pointwise mutation yields an **allele** which becomes sufficiently frequent in a population of individuals, i.e. a *Minor Allele Frequency* (MAF) of 1% or 5%, then it is called a **Single Nucleotide Polymorphism (SNP)**.

Dediu (2015). An introduction to genetics for language scientists, p. 168.



Population Level

At the **population level**, we can have individuals with any of the possible combinations of alleles (e.g. *Aa*, *AA*, *aa* for a *biallelic* locus).

Dediu (2015). An introduction to genetics for language scientists, p. 161.

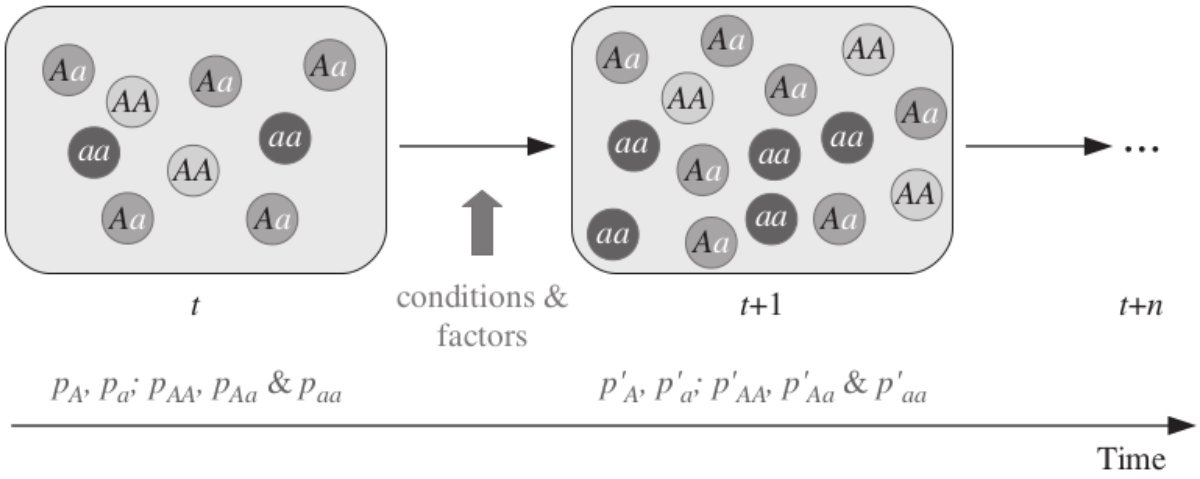
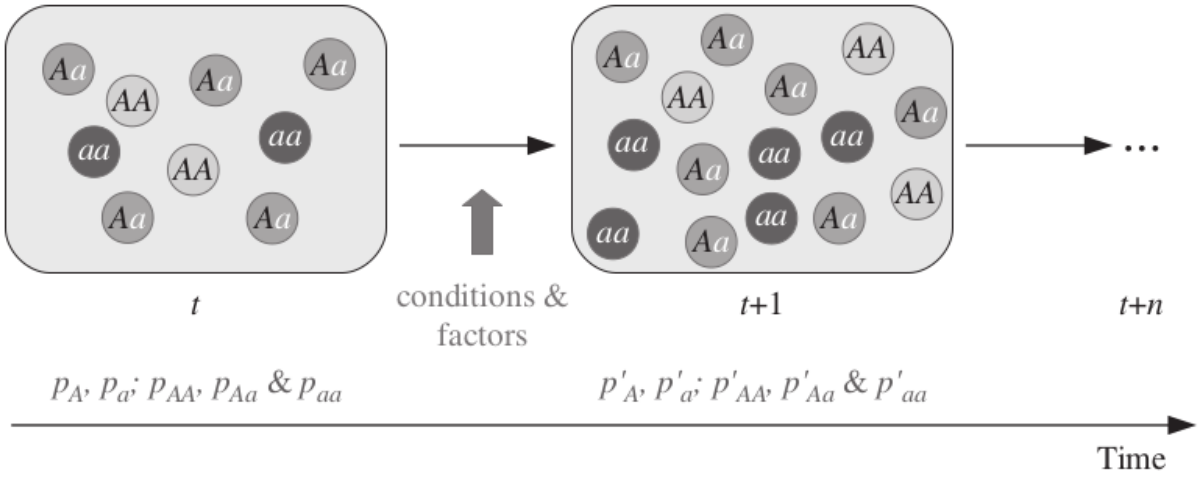


Figure 8.1 An idealized population composed of individuals each having a specific genotype at a given autosomal locus with two alleles, *a* and *A* (thus, a biallelic locus). Shown are the population-level allele and genotype frequencies and the evolution of the population from timestep *t* to the next timestep *t*+1.

Exercise

Calculate the relative frequencies of allele combinations on chromosomes (p_{Aa}, p_{AA}, p_{aa}), as well as for individual alleles (p_a, p_A), per time step (t, $t+1$). Do you find differences between the population at time t and $t+1$? What could be the "conditions and factors" for these?



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Hardy-Weinberg Equilibrium (HWE)

The HWE is the common **baseline** for population genetics. It states that under the following conditions the probabilities of alleles and allele combinations should remain constant:

- **Infinite populations**,
- **no mutations**,
- **no selection**,
- **random mating**,
- **no migration**.

Of course, in reality, these are never met. Still, it is useful to investigate what happens when individual conditions or combinations of them are violated.

Dediu (2015), pp. 163.

Infinite Populations

If a population had an **infinite number** of individuals, then any given change in the DNA sequence (new allele) could never go to **fixation**, i.e. become the only variant. However, as populations in the real world are always **finite**, one of the alleles is always going to reach fixation just via **random drift**. The expected time this takes depends on the **mutation rates** and the **population size**.

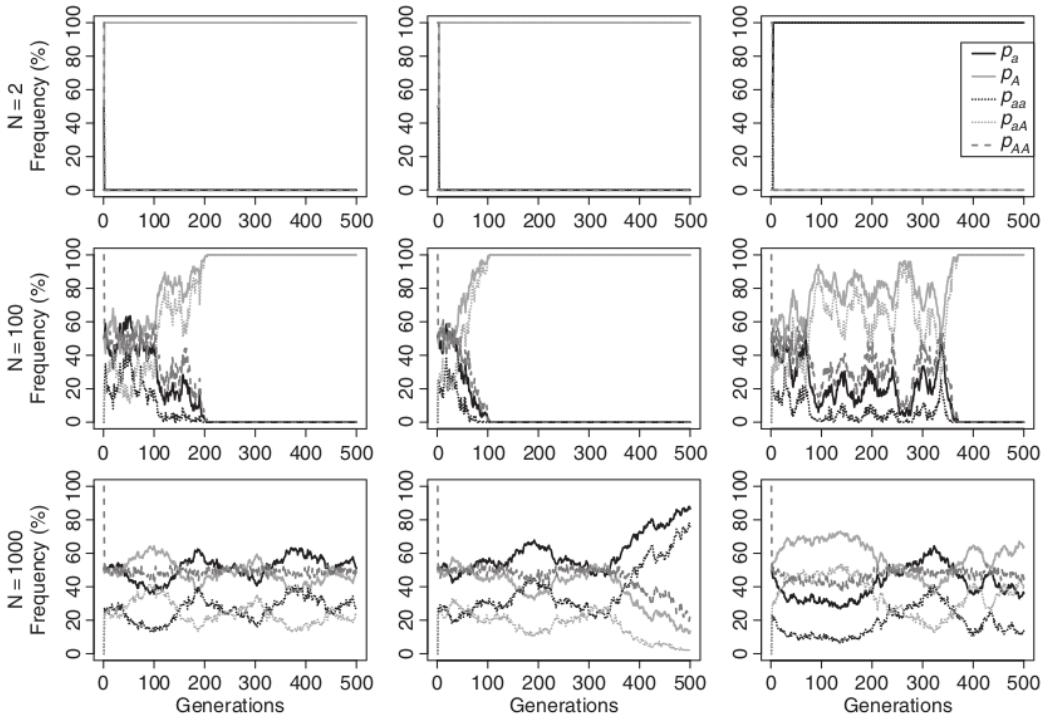


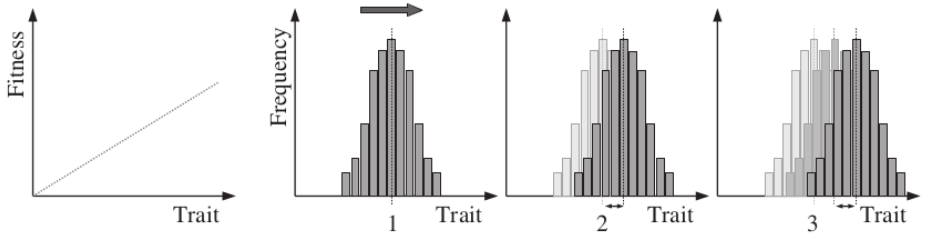
Figure 8.2 Simulations of genetic drift in a population with N individuals for a diploid biallelic locus. Initially (generation 0) the population is composed only of heterozygous individuals aA and evolves under the assumptions of HWE (except for finite population size). Each row represents three independent runs (the columns) for a given population size (top to bottom $N = 2$, $N = 100$ and $N = 1000$). Shown are the allele frequencies p_a and p_A, and the genotype frequencies p_{aa}, p_{aA} and p_{AA} (see also the legend in top rightmost plot).

Dediu (2015). An introduction to genetics for language scientists, p. 165.

Selection

If a mutation leads to a new allele which comes under **selection pressure** (*positive*, *stabilizing*, or *disruptive*) then the allele will systematically increase/decrease in frequency across the popuation.

Positive Selection



Stabilizing Selection

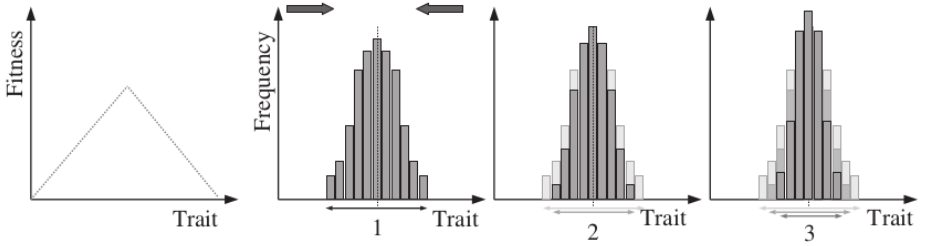


Figure: Leftmost panels give the relationship between a trait value (x-axis) and the fitness of a phenotype (y-axes). The right panels give the frequency distribution of the trait value in the population. The numbers (1, 2, 3) indicate generations of sexual reproduction. For example, increased body height might correlate with higher fitness (positive selection) in a changing environment (forest to savannah). In a stable environment there might be an optimal body height, and any deviation from this would then correspond to fitness loss (stabilizing selection).

Dediu (2015). An introduction to genetics for language scientists, pp. 171.

Migration

Migrations can lead to the **admixture** of populations and their genetic material. Also, migrations can lead to **serial founder effects (bottlenecks)**. Note that this is related to *random drift*, which is expected to decrease allele diversity.

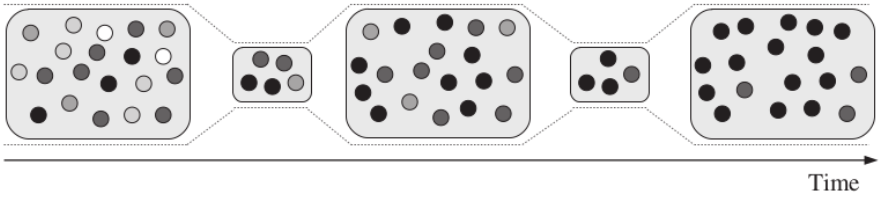


Figure 8.3 Population bottlenecks and (repeated) founder effects result in a loss of genetic diversity through random sampling: in the first bottleneck the "white" and "light grey" variants are lost, while through the second bottleneck only the "black" and "dark grey" variants survive.

Dediu (2015). An introduction to genetics for language scientists, p. 167.

Uniformity

If the HWE was reality, we would all be uniform, clones of a single genome.

Diversity

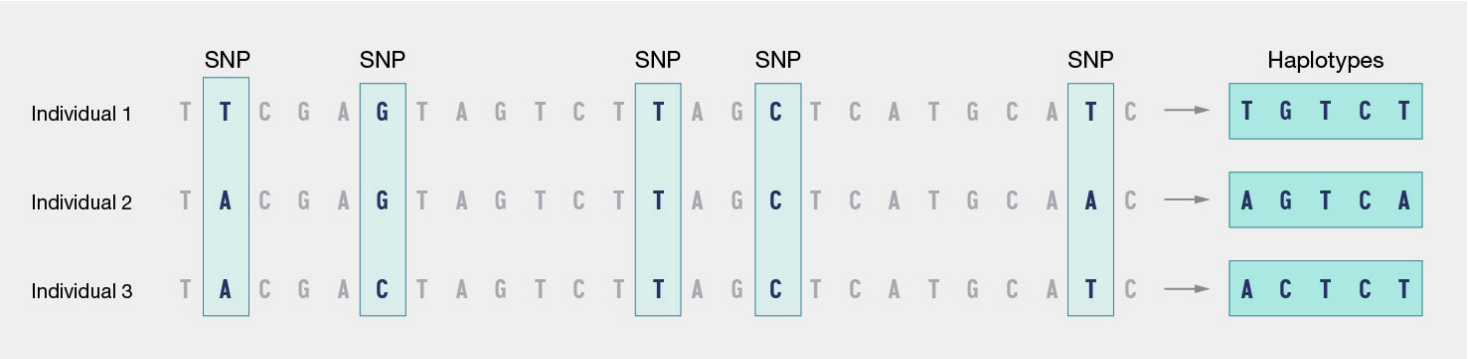
Due to **mutations**, **recombination**, **selection**, and **migration** we are individuals.

Section 3: Genetic Ancestry

Haplotype

"A **haplotype** refers to a set of DNA variants along a single chromosome that tend to be inherited together. They tend to be inherited together because they are close to each other on the chromosome, and recombinations between these variants are rare. A haplotype can be limited to a single gene or it can be larger and include multiple genes."

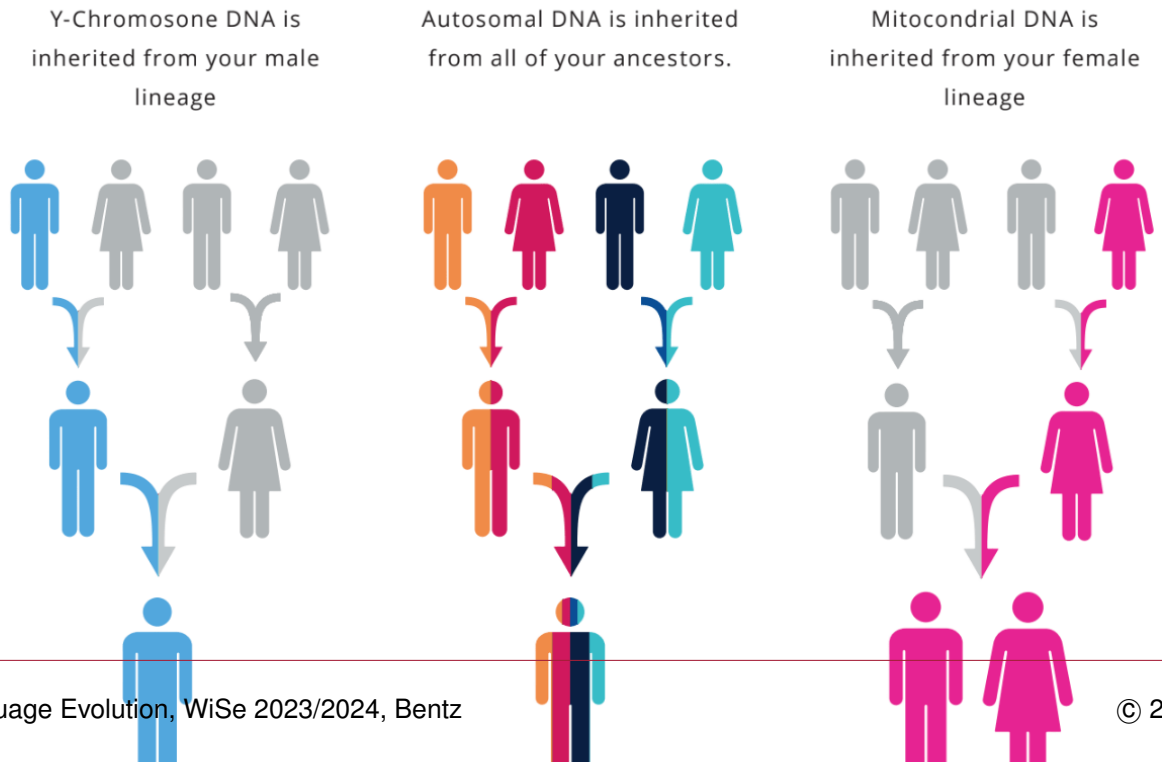
See definition at: `https://www.genome.gov/genetics-glossary/haplotype`



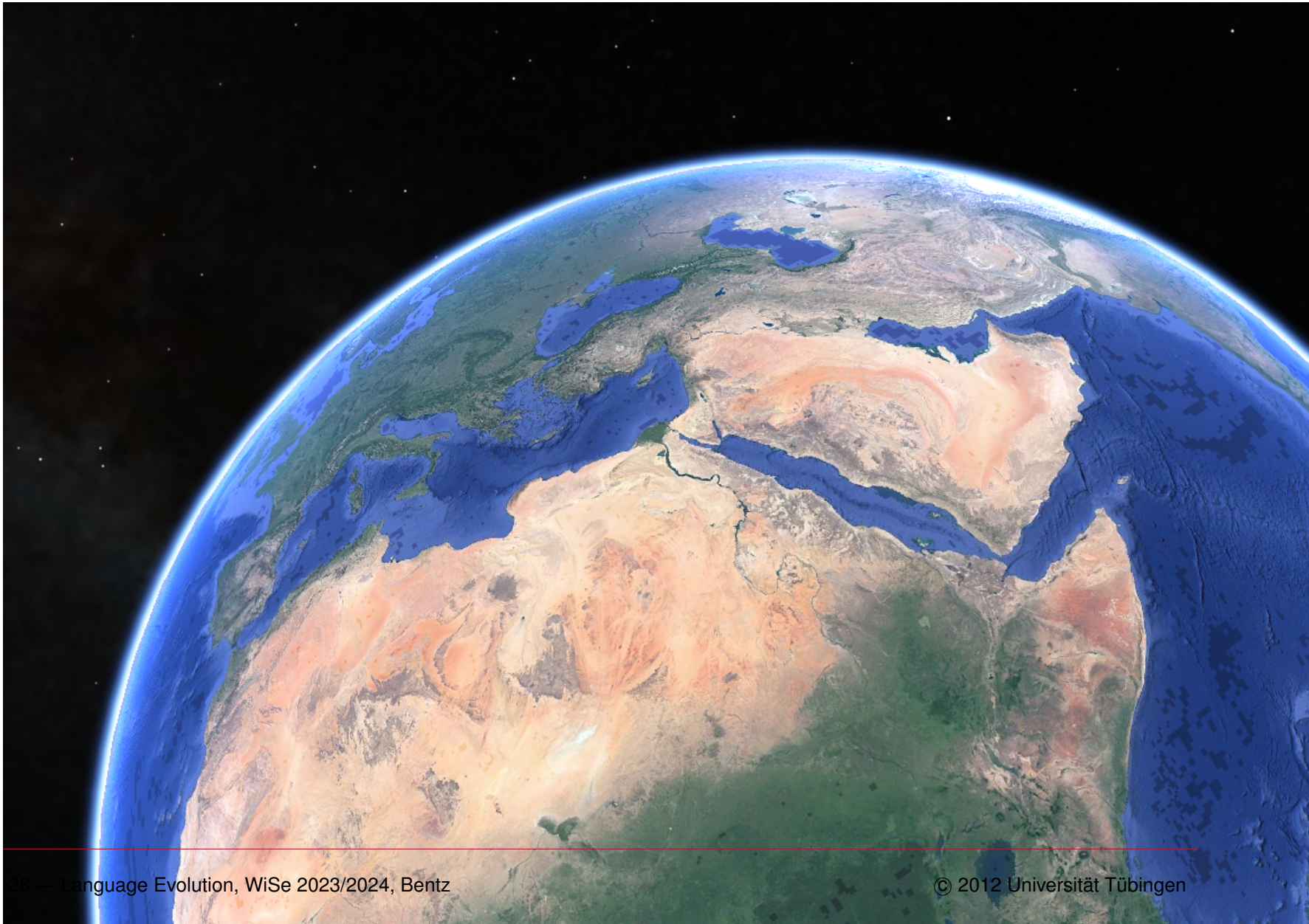
Genetic Ancestry: Haplogroups

"A **haplogroup** is a genetic population group of people who share a common ancestor on the **patriline** [Y-chromosome DNA (Y-DNA)] or the **matriline** [mtDNA] [...] haplogroups are determined by single-nucleotide polymorphism (SNP) tests."

See definition at: `https://isogg.org/wiki/Haplogroup`



Section 4: Out of Africa Dispersals



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Out of Africa 1 (OOA1)

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When exactly the first hominins left Africa, and *what* species of hominin this was, is still a matter of debate. The earliest clearly dated, uncontroversial fossils outside of Africa were found at Dmanisi (Georgia).

See video:
`https://www.youtube.com/watch?v=9btN6H7H3Rg`

OOA1: Fossil Evidence

The earliest layers at Dmanisi are dated to **1.85-1.78 Mya**. These dates as well as the cranial and postcranial analyses of the fossils have led to the assumption that the first hominins colonizing this area potentially predate *Homo erectus*.

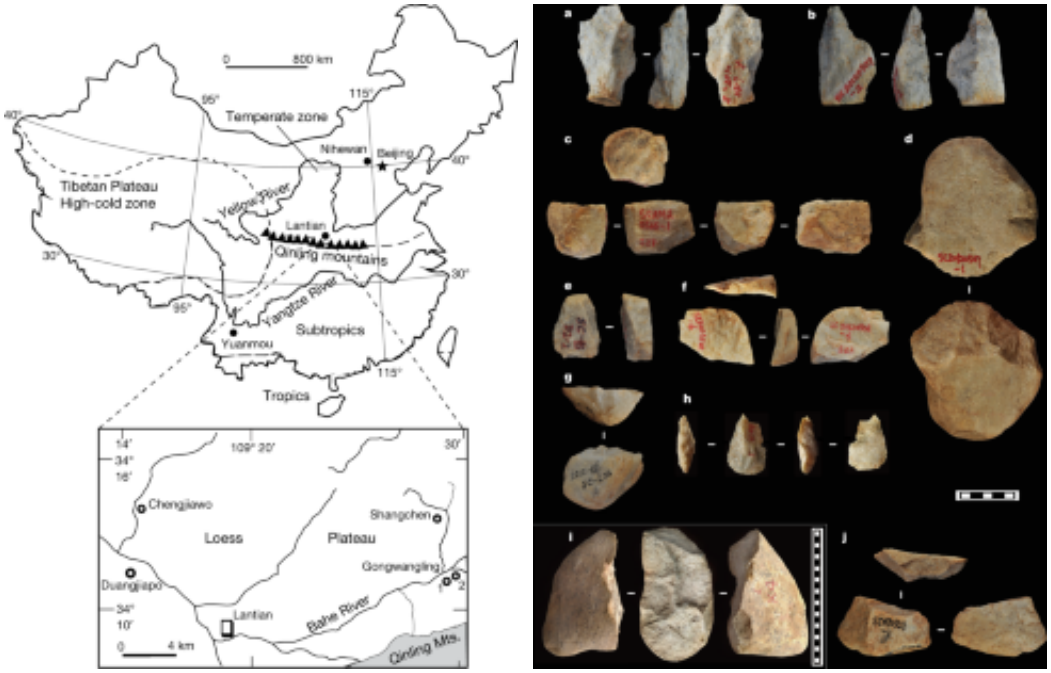
Ferring et al. (2011). Earliest human occupations at Dmanisi (Georgian Caucasus) dated to 1.85-1.75 Ma.

Scardia et al. (2020). What kind of hominin first left Africa?

FIGURE 1 The five hominin crania from Dmanisi, from left to right: Skull 1 (D2280), Skull 2 (D2282), Skull 3 (D2700), Skull 4 (D3444), and Skull 5 (D4500). Courtesy of the Georgian National Museum

OOA1: Lithics Evidence

The earliest lithics found outside Africa are older than **2 Mya**, and of the Oldowan type. This is typically associated with *Homo habilis*, rather than *Homo erectus*.



Zhu et al. (2018). Hominin occupation of the Chinese Loess Plateau since about 2.1 million years ago.

Out of Africa 0?

These recent finds and datings add nuance to the traditional idea that *Homo erectus* was the first hominin to leave Africa around 1.8 Mya. There were probably earlier dispersals.

Scardia et al. (2020). What kind of hominin first left Africa?

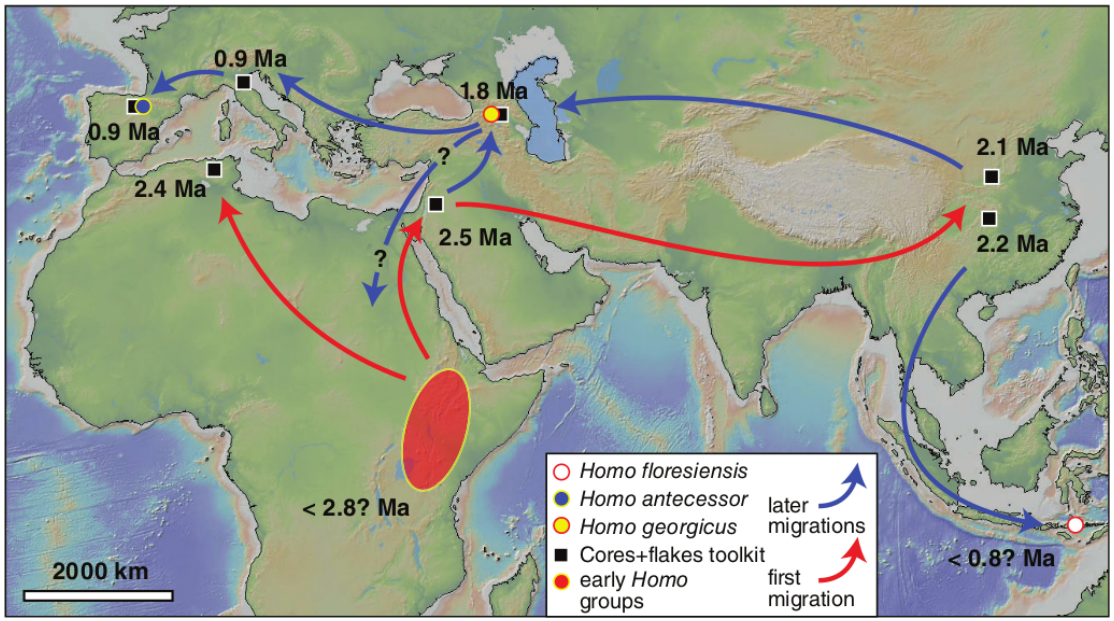


FIGURE 2 Tentative scenario for the first Out of Africa expansion at ca. 2.5 Ma according to the recent findings from Jordan and China, and later migrations stemming from the early *Homo* lineage. See text for discussion and references

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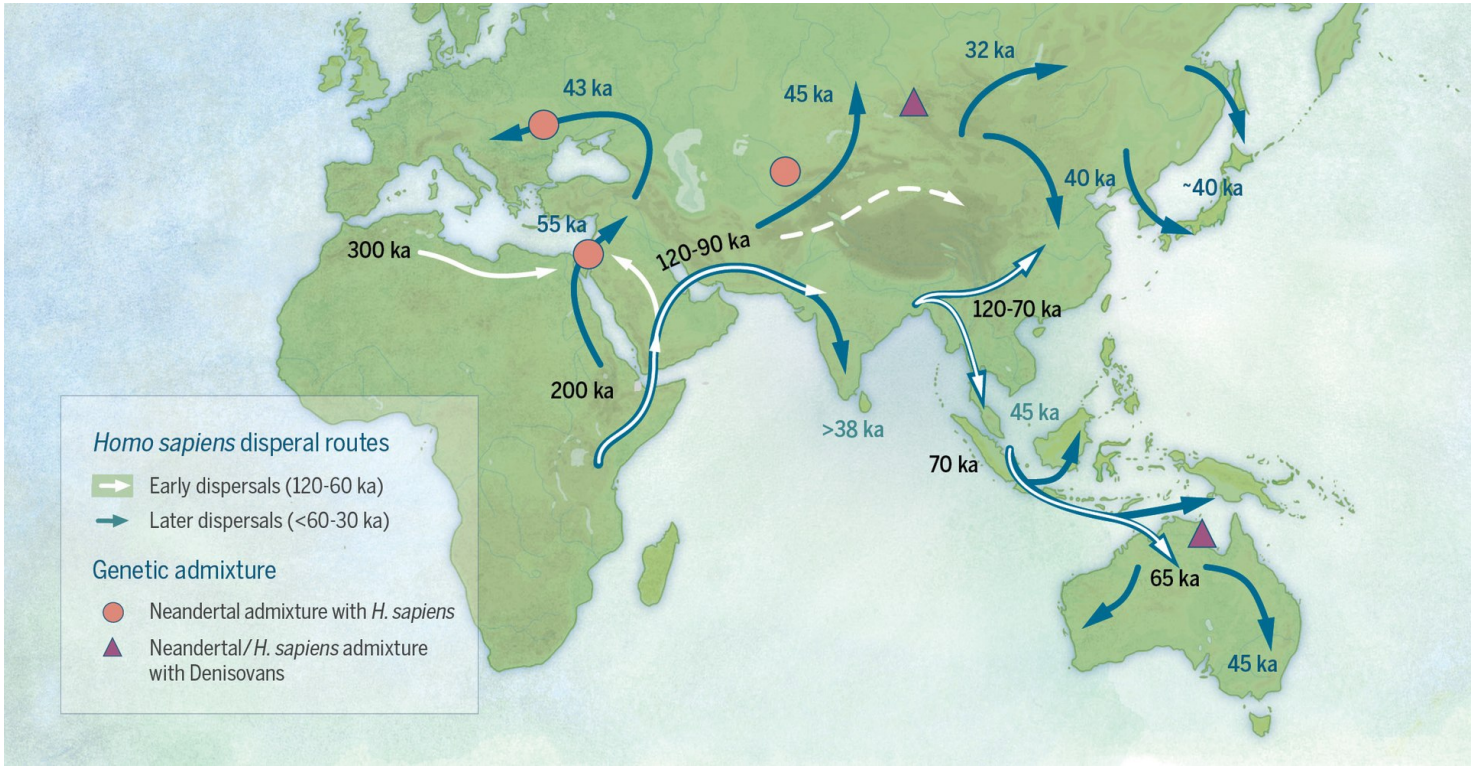
Out of Africa 2 (OOA2)

Out of Africa 2 (OOA2)

There is a general consensus that **Anatomically Modern Humans (AMH)** widely spread across the globe from Africa starting around **100 Kya**. However, the details of this dispersal are still up for debate.



Bae et al. (2017). On the origin of modern humans: Asian perspectives.

OOA2: Fossil Evidence

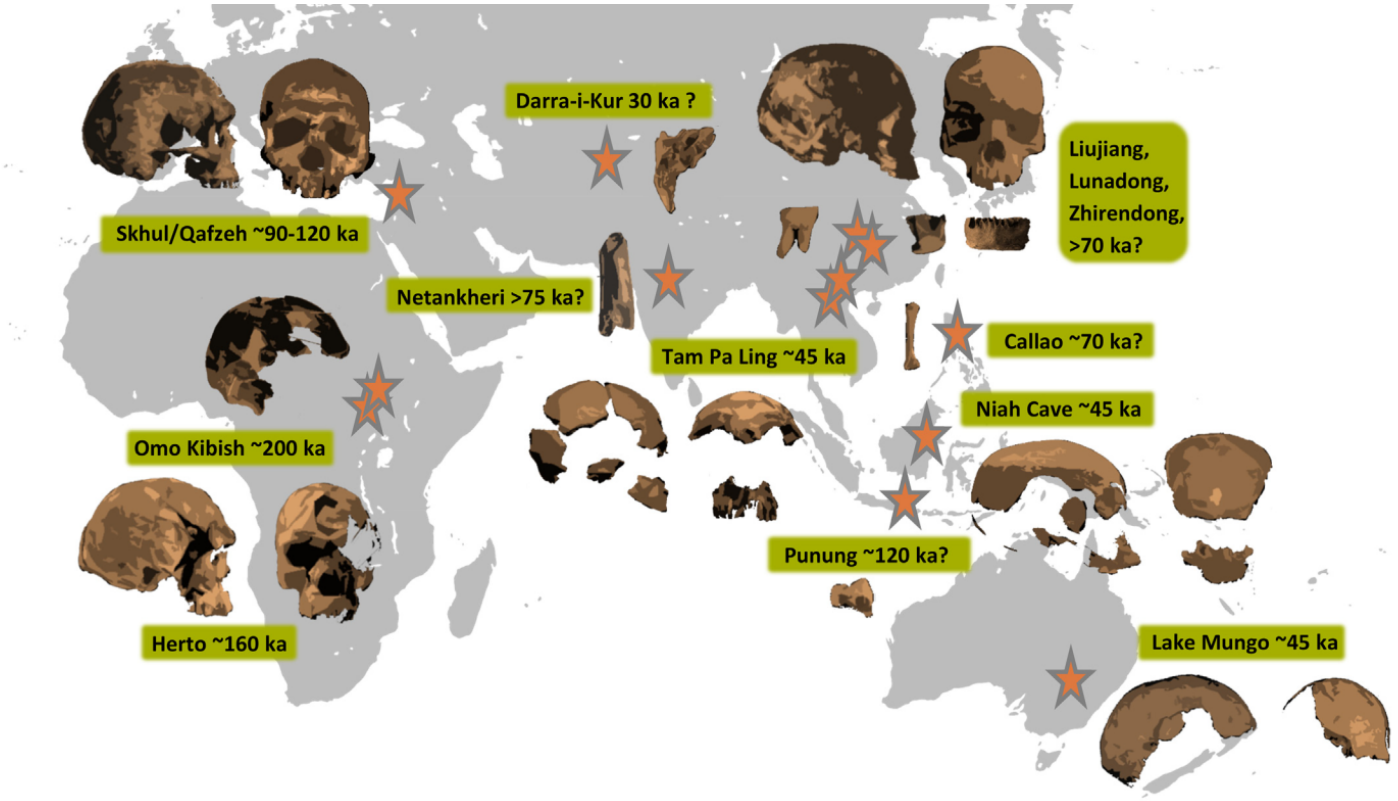
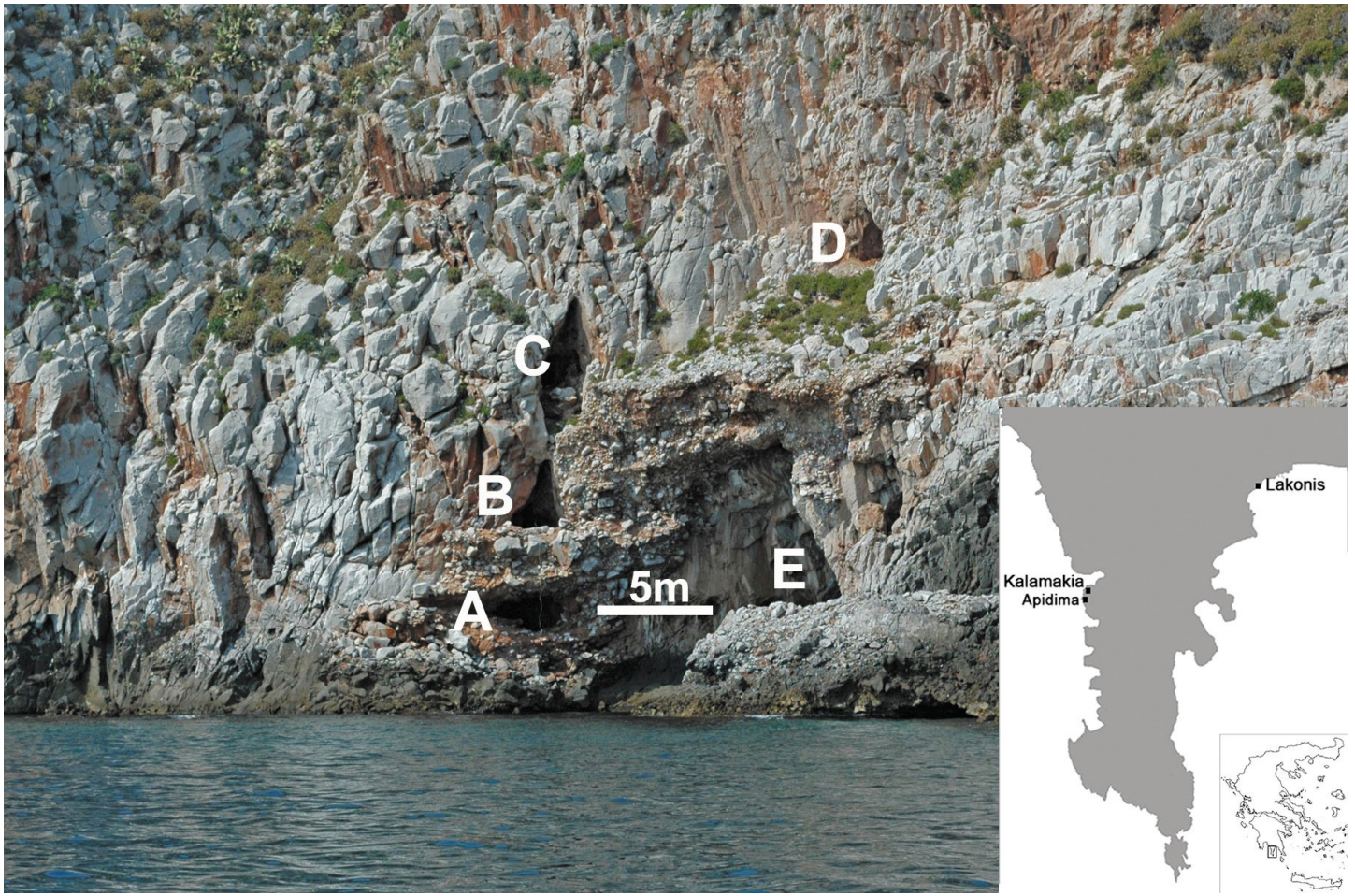


Fig. 1. Anatomically modern human fossils from Africa, Asia, and Australia. Question marks indicate uncertainty in taxonomy, chronometric dating, or both. Drawings adapted from photographs in the following sources: (Darra-i-Kur: Angel, 1972; Lunadong LN0030: Bae et al., 2014; Niah Cave "Deep Skull:" Barker et al., 2007; Omo I: Day, 1969; Tam Pa Ling I: Demeter et al., 2012; Zhirendong 3: Liu et al., 2010; Callao II-77-J3-7691: Mijares et al., 2010; Netankheri NTK-F-02-07: Sankhyan et al., 2012; Punung PU-198: Storm et al., 2005; Lake Mungo III: Thorne et al., 1999; Qafzeh 6: Vandermeersch, 1981; Herto BOU-VP-16/1: White et al., 2003; Liujiang: Woo, 1959).

Reyes-Centeno (2016). Out of Africa and into Asia.

Apidima: A much earlier dispersal of AMHs?



Harvati et al. (2021). Direct U-series dating of the Apidima C human remains.

Apidima: A much earlier dispersal of AMHs?

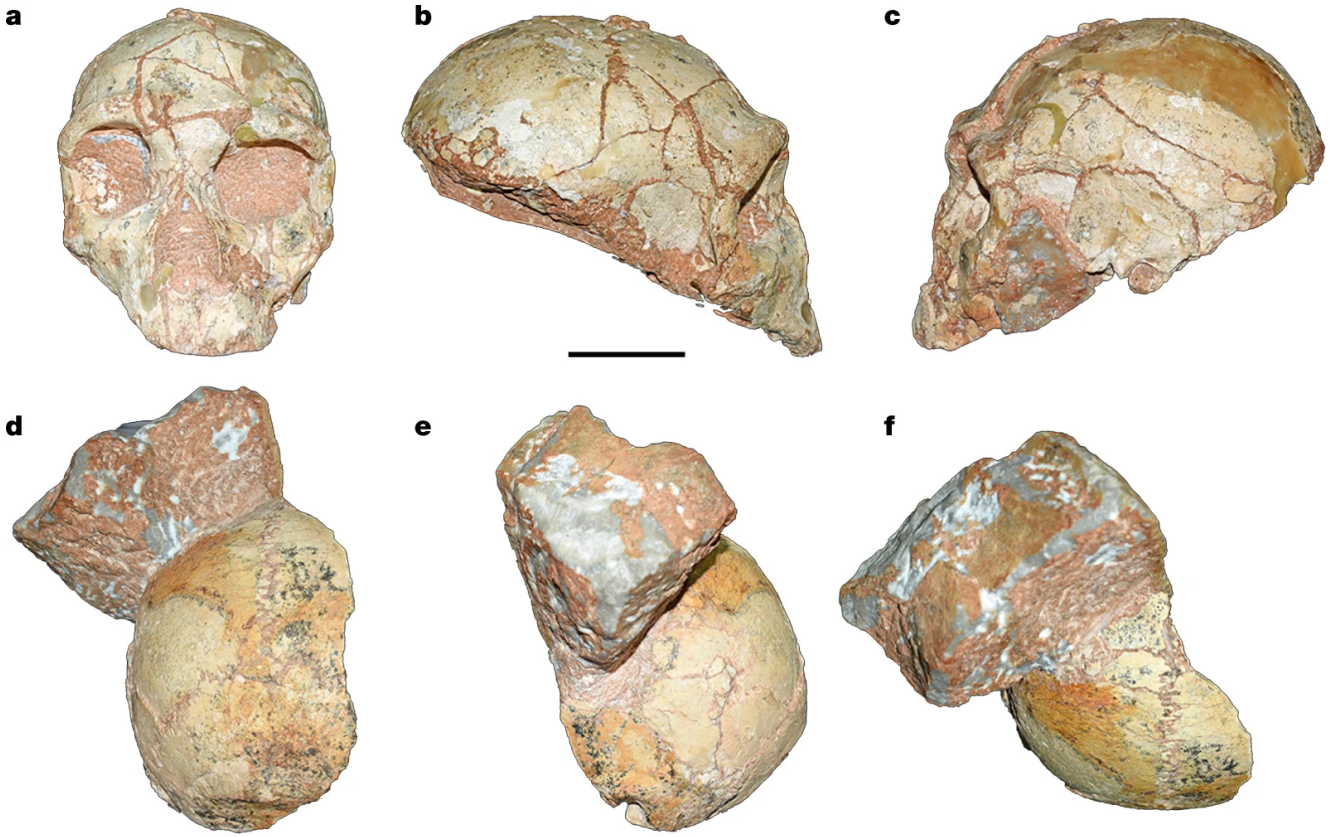


Figure. The fossil crania of Apidima 2 and Apidima 1. a–c, Apidima 2. a, Frontal view. b, Right lateral view. c, Left lateral view. d–f, Apidima 1. d, Posterior view. e, Lateral view. f, Superior view. Scale bar, 5 cm.

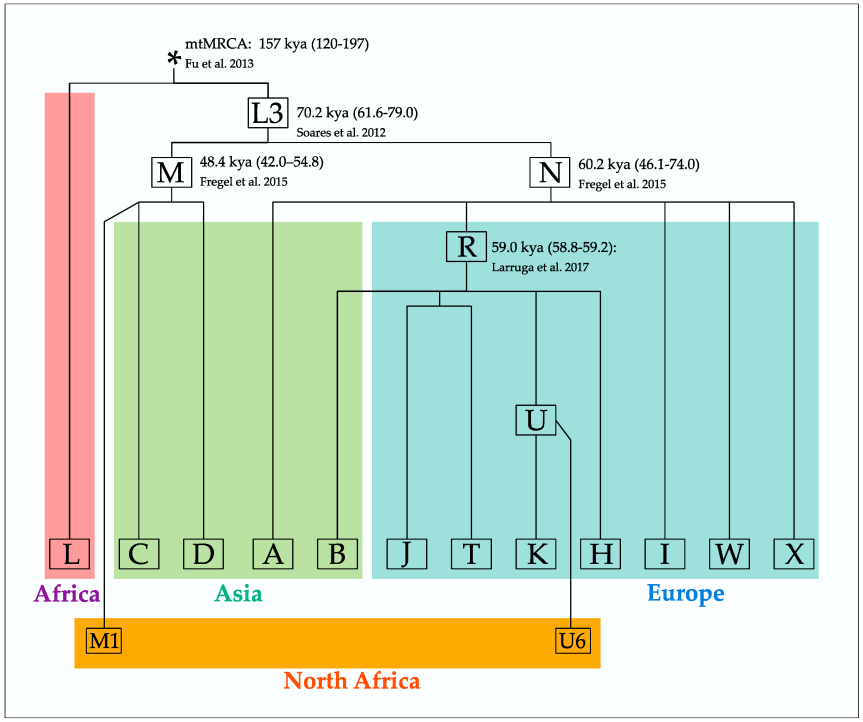
Harvati et al. (2019). Apidima Cave fossils provide earliest evidence of Homo sapiens in Eurasia.

Apidima 1: A much earlier dispersal of AMHs?

- **Apidima 1**: Clusters closest to *early modern human* fossils in terms of geometric morphometrics. It is dated to c. *210 Kya* (!). This would be by far the earliest evidence of AMH outside of Africa (remember Skhul and Qafzeh in the Levant at c. 90-120 Kya).
- **Apidima 2**: Clearly clusters with Neandertals and is dated to c. 170 Kya (not surprising).

OOA2: Genetic Evidence

The haplogroups of mtDNA allow an estimation of the time depth of the **mitrochondial Most Recent Common Ancester (mtMRCA)**, currently at ca. 157 Kya. The **L3 haplogroup** is generally believed to be strongly associated with the OOA2 migration event.



Hernández (2023). Mitochondrial DNA in human diversity and health.

OOA2: Potential Genetic Dispersals

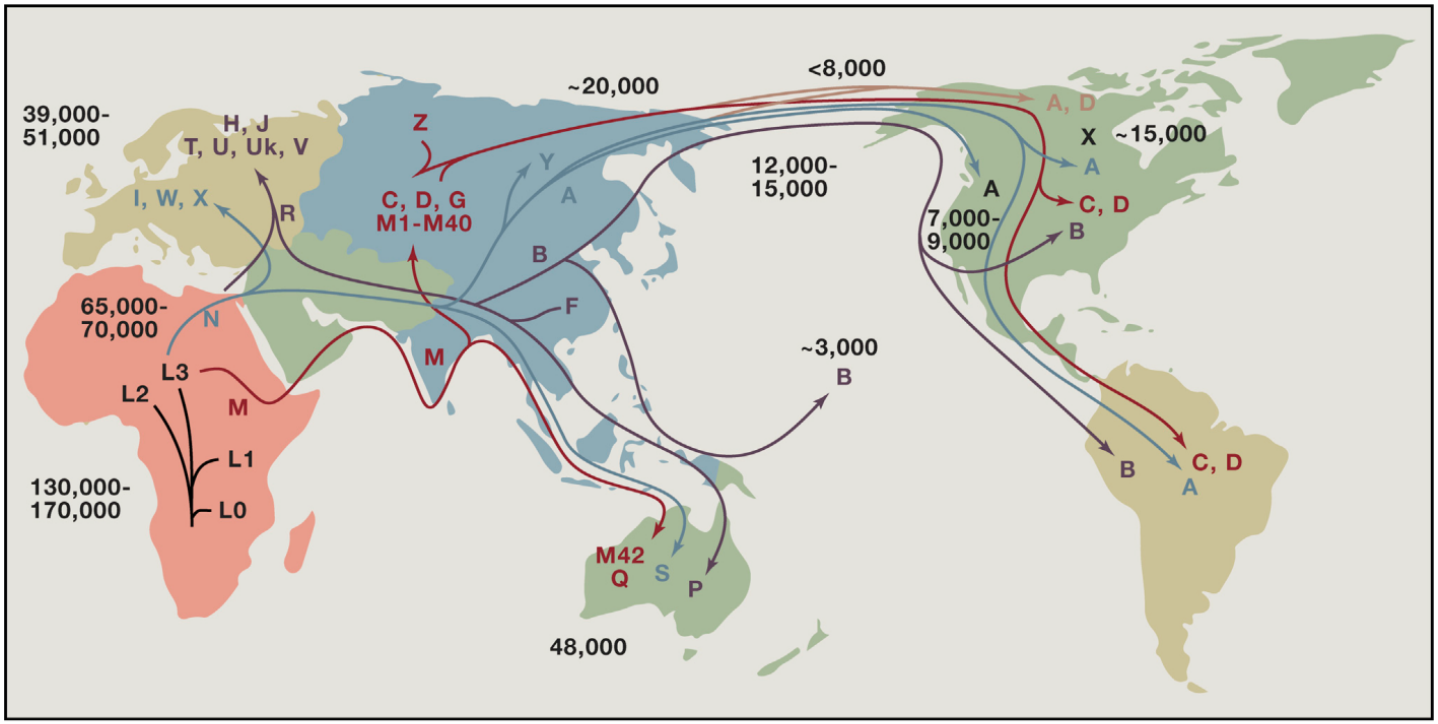


Figure 1. Regional Radiation of Human mtDNAs from their Origin in Africa and Colonization of Eurasia and the Americas Implies that Environmental Selection Constrained Regional mtDNA Variation

All African mtDNAs are subsumed under macrohaplogroup L and coalesce to a single origin about 130,000–170,000 YBP. African haplogroup L0 is the most ancient mtDNA lineage found in the Koi-San peoples, L1 and L2 in Pygmy populations. The M and N mtDNA lineages emerged from Sub-Saharan African L3 in northeastern Africa, and only derivatives of M and N mtDNAs successfully left Africa, giving rise to macrohaplogroups M and N. N haplogroups radiated into European and Asian indigenous populations, while M haplogroups were confined to Asia. Haplogroups A, C, and D became enriched in northeastern Siberia and were positioned to migrate across the Bering Land Bridge 20,000 YBP to found Native Americans. Additional Eurasian migrations brought to the Americas haplogroups B and X. Finally, haplogroup B colonized the Pacific Islands. Figure reproduced from (MITOMAP, 2015).

Wallace (2015). Mitochondrial DNA Variation in Human Radiation and Disease.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Summary

Summary

- At the population level, **Single Nucleotide Polymorphisms (SNPs)** are point mutations which have become more frequent over time.
- Alleles might become more frequent or less frequent due to **population size**, **migrations**, **selection**, or **mutation rates**.
- SNPs also identify so-called haplotypes and **haplogroups**, which reflect human dispersals.
- Hominins have dispersed **at least twice** from Africa into the rest of the world (OOA1: c. 1.8 Mya, OOA2: c. 70 Kya).


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Thank You.

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