

Modern Human Origins

Hugo Reyes-Centeno, Yonatan Sahle, Christian Bentz



Last week:

- ❑ **Hominin taxonomy**
How do we classify human fossils?
- ❑ **Modern human fossils**
When and where do we find anatomically modern fossils?
- ❑ **Populations and demes**
Are population models better than species models?



For today:

- ❑ **Genomics of modern human origins**
What does (ancient) DNA tell us about the origins and evolution of modern humans?
- ❑ **Models of anthropogeny**
What model of modern human origins is best supported with the current fossil and genomic evidence?
- ❑ **Modern human dispersals**
When, how, and why did anatomically modern humans disperse out of Africa?



Species concepts and speciation

- ❑ **Biological species**
Reproductively isolated populations. Interbreeding is possible by species recognition and compatible fertilization system.
- ❑ **Morphological species**
Phenetic clustering of organisms based on anatomical traits
- ❑ **Genetic species**
Genetic clustering of organisms based on genetic frequencies
- ❑ **Phylogenetic species**
- ❑ **Evolutionary species**
Ancestral-descendant sequence of populations terminating by extinction



Species concepts and speciation

- ❑ **Biological species**
Reproductively isolated populations. Interbreeding is possible by species recognition and compatible fertilization system.
- ❑ **Morphological species**
Phenetic clustering of organisms based on anatomical traits
- ❑ **Genetic species**
Genetic clustering of organisms based on genetic frequencies
- ❑ **Phylogenetic species**
- ❑ **Evolutionary species**
Ancestral-descendant sequence of populations terminating by extinction

Allopatric

Original population



Initial step of speciation



Barrier formation

Evolution of reproductive isolation



In isolation

New distinct species after equilibration of new ranges

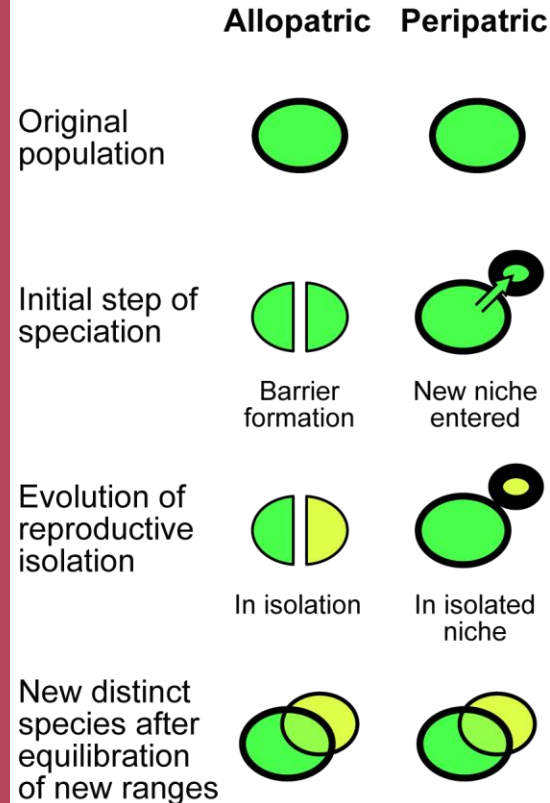


I. Karonen 2006



Species concepts and speciation

- ❑ **Biological species**
Reproductively isolated populations. Interbreeding is possible by species recognition and compatible fertilization system.
- ❑ **Morphological species**
Phenetic clustering of organisms based on anatomical traits
- ❑ **Genetic species**
Genetic clustering of organisms based on genetic frequencies
- ❑ **Phylogenetic species**
- ❑ **Evolutionary species**
Ancestral-descendant sequence of populations terminating by extinction

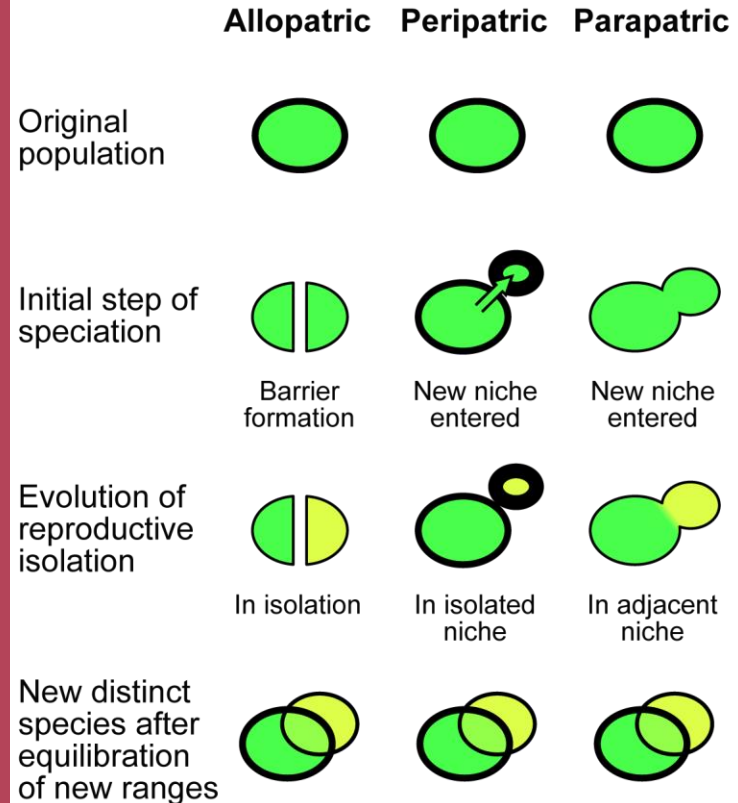


I. Karonen 2006



Species concepts and speciation

- ❑ **Biological species**
Reproductively isolated populations. Interbreeding is possible by species recognition and compatible fertilization system.
- ❑ **Morphological species**
Phenetic clustering of organisms based on anatomical traits
- ❑ **Genetic species**
Genetic clustering of organisms based on genetic frequencies
- ❑ **Phylogenetic species**
- ❑ **Evolutionary species**
Ancestral-descendant sequence of populations terminating by extinction

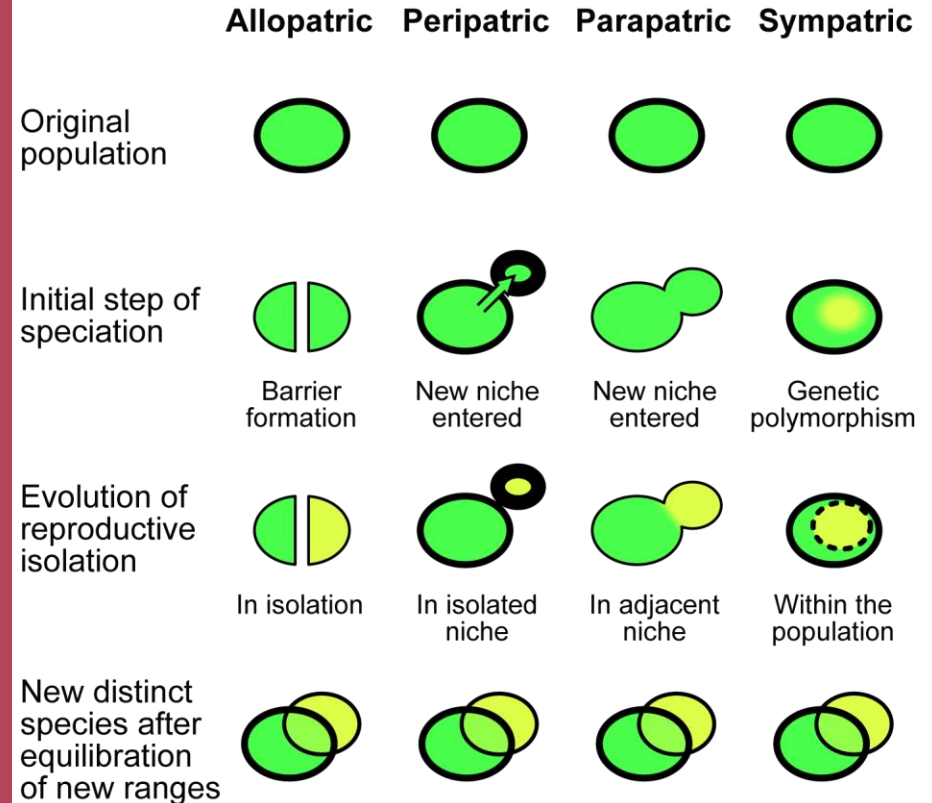


I. Karonen 2006



Species concepts and speciation

- ❑ **Biological species**
Reproductively isolated populations. Interbreeding is possible by species recognition and compatible fertilization system.
- ❑ **Morphological species**
Phenetic clustering of organisms based on anatomical traits
- ❑ **Genetic species**
Genetic clustering of organisms based on genetic frequencies
- ❑ **Phylogenetic species**
- ❑ **Evolutionary species**
Ancestral-descendant sequence of populations terminating by extinction



I. Karonen 2006

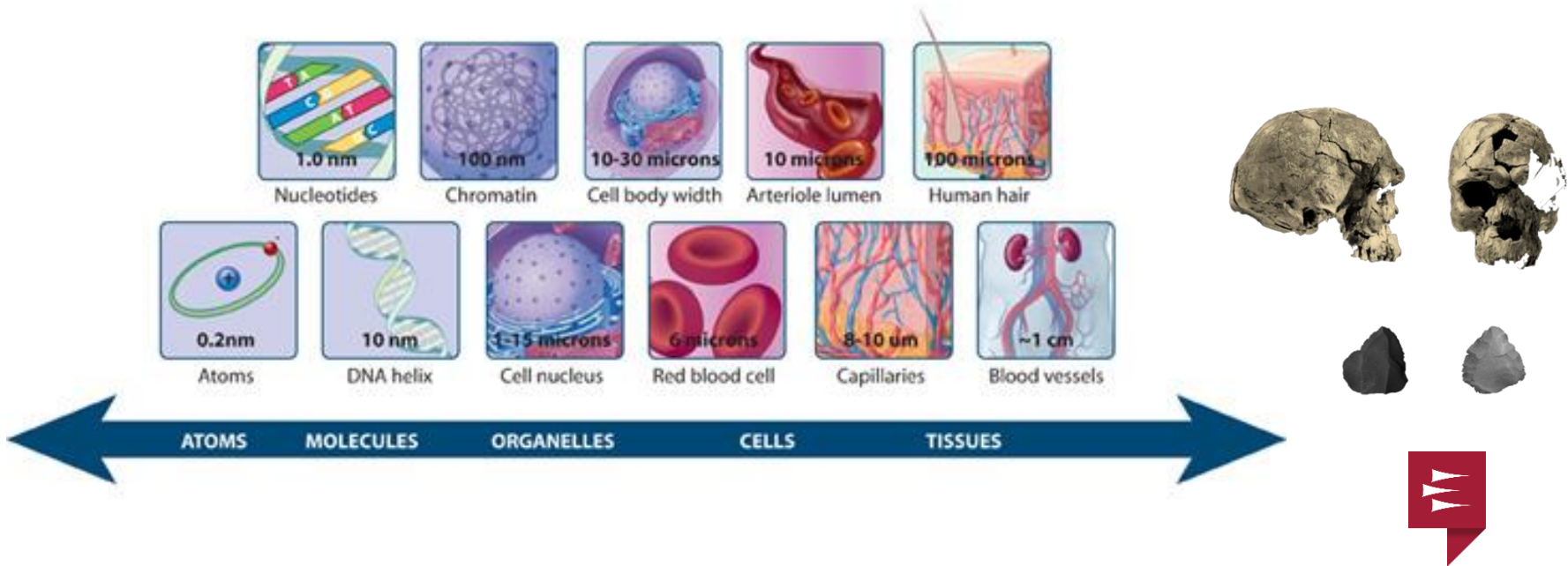


Modern human genomics

What does (ancient) DNA tell us about the origins and evolution of modern humans?



White *et al.* 2003; Beyin 2013

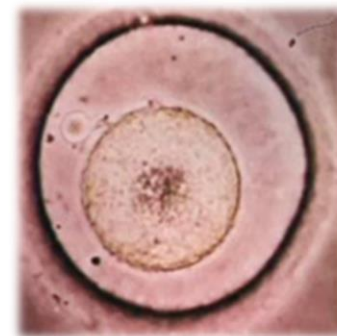
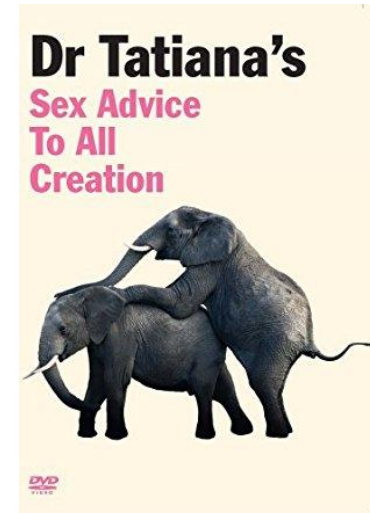
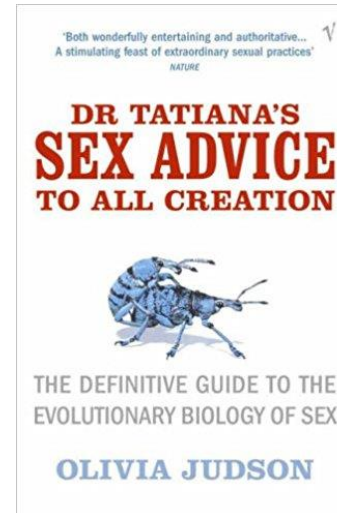


Nature Education 2010; White *et al.* 2003; Beyin 2013



Basics of inheritance

- **Sexual reproduction**
common across the animal kingdom
and usually implies pronounced
sexual dimorphism



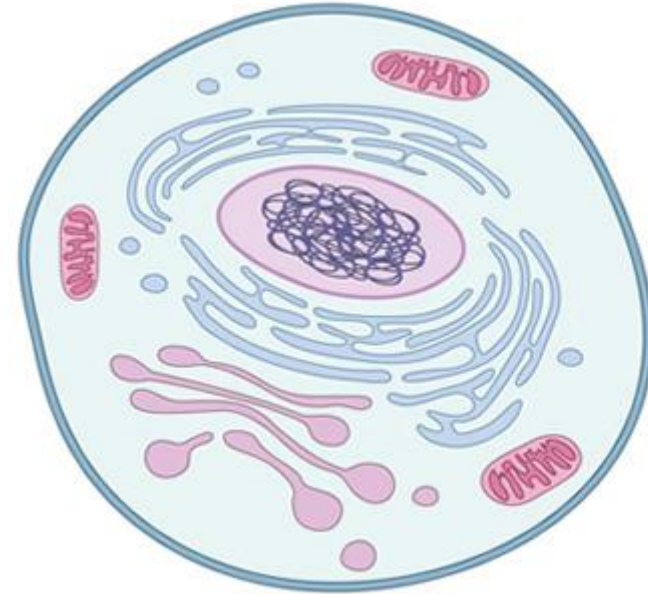
Nature Education 2010



Basics of inheritance

- ❑ **Sexual reproduction**
common across the animal kingdom
and usually implies pronounced
sexual dimorphism

- ❑ **Cellular structure**
 - Mitochondria
 - Nucleus

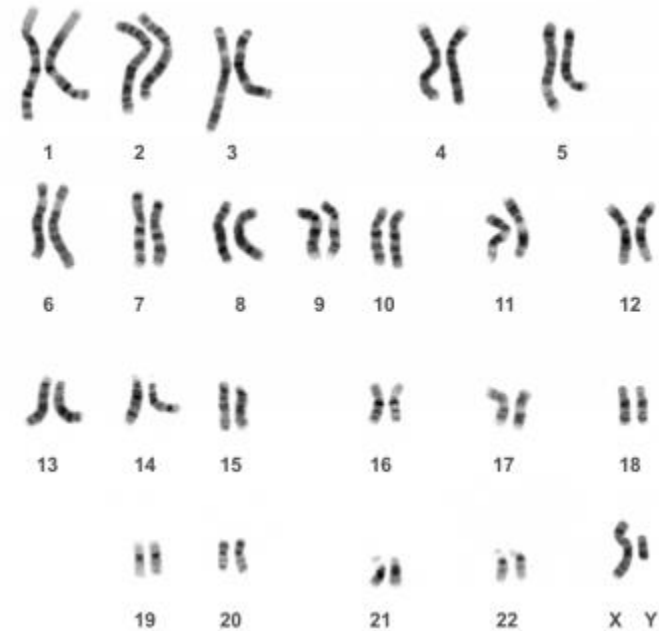
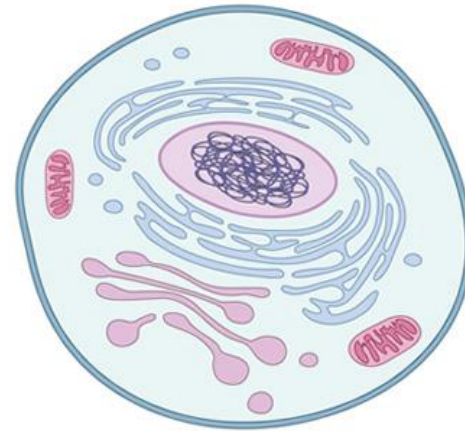


Nature Education 2010



Basics of inheritance

- ❑ **Sexual reproduction**
common across the animal kingdom and usually implies pronounced sexual dimorphism
- ❑ **Cellular structure**
 - Mitochondria
 - Nucleus
- ❑ **Chromosomes**



Nature Education 2010; [23andMe](#)

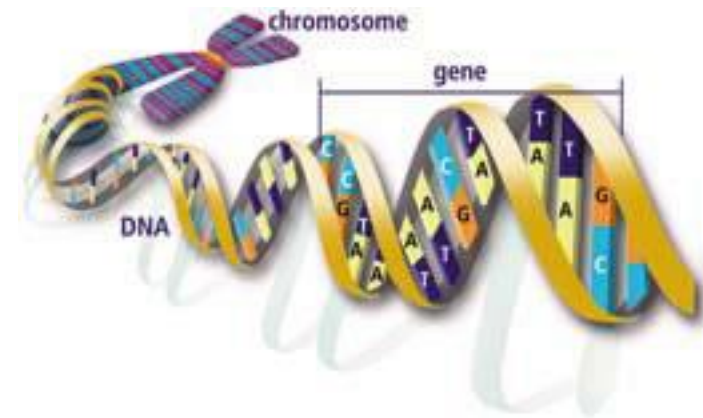


Basics of inheritance

- ❑ **Sexual reproduction**
common across the animal kingdom
and usually implies pronounced
sexual dimorphism

- ❑ **Cellular structure**
 - Mitochondria
 - Nucleus

- ❑ **Chromosomes**
 - DNA strand
 - Genes
 - Nucleotides

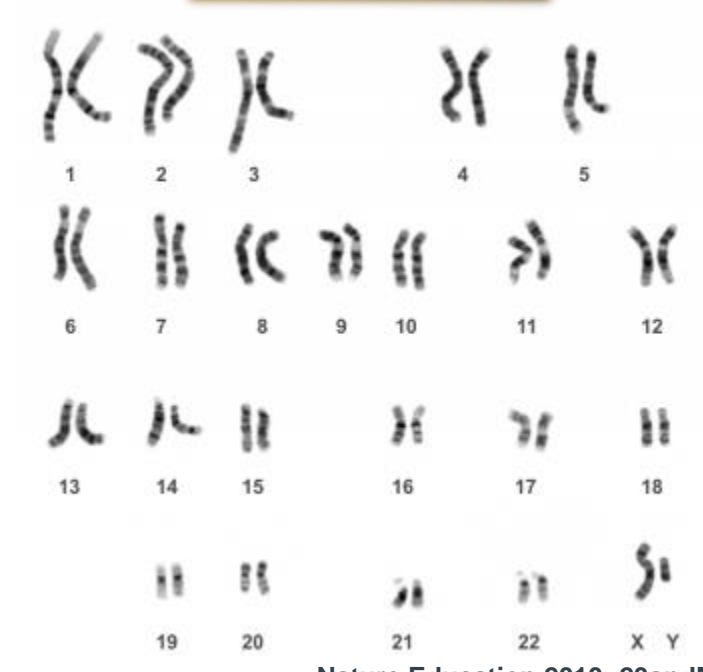
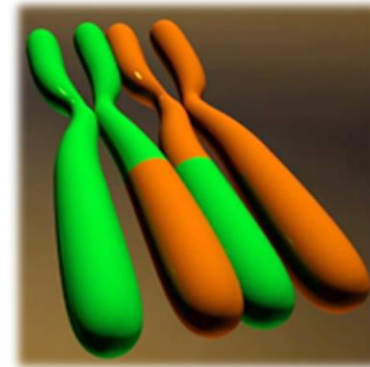


[US Department of Energy](#)



Basics of inheritance

- ❑ **Sexual reproduction**
common across the animal kingdom and usually implies pronounced sexual dimorphism
- ❑ **Cellular structure**
 - Mitochondria
 - Nucleus
- ❑ **Chromosomes**
 - DNA strand
 - Genes
 - Nucleotides
- ❑ **Recombination**
at the cellular level, genetic diversity is produced by the process of recombination



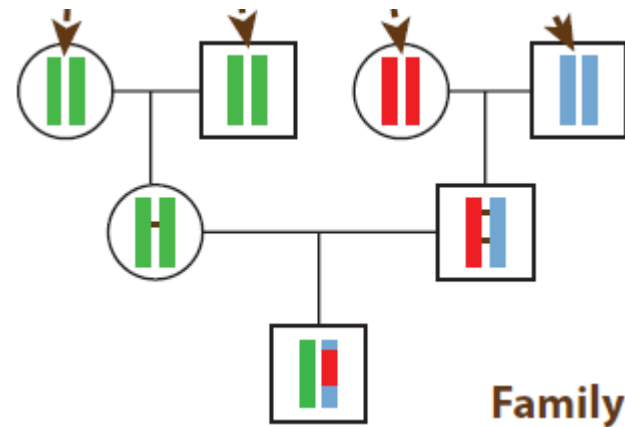
Nature Education 2010; 23andMe



Basics of inheritance

□ Genealogy

In theory, you can trace ancestry based on genetic inheritance, but recombination makes this difficult



Novembre & Ramachandran 2011



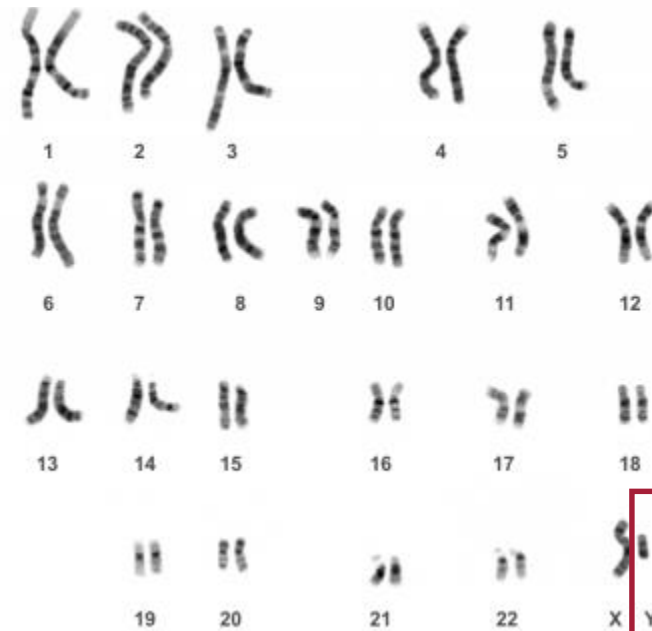
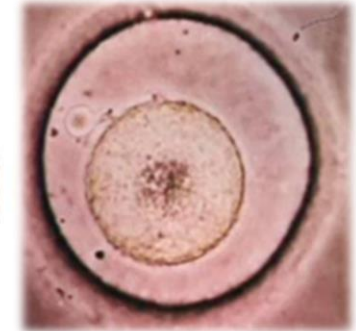
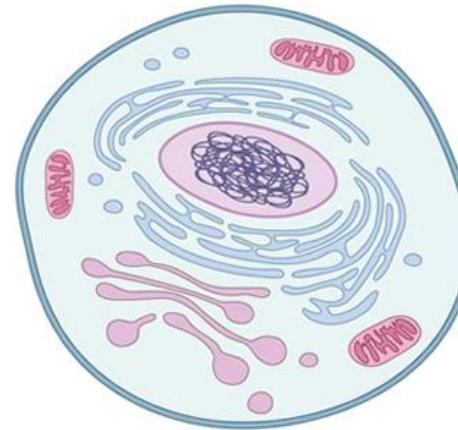
Basics of inheritance

Two regions that do not recombine are:

- ❑ **mitochondria**
inherited maternally

- ❑ **Y-chromosome**
inherited paternally

*some regions do recombine, but not to the same degree as other chromosomes



Nature Education 2010; 23andMe

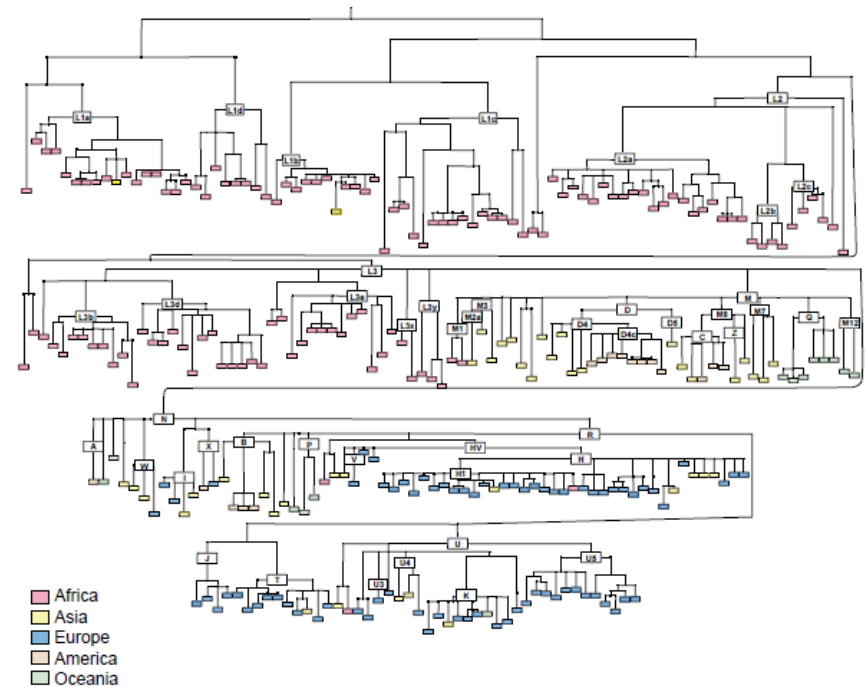


Phylogeography

Phylogeography: a method to analyze the geographical distribution of different clades within a phylogeny

- Optimal for uni-parental loci (mitochondrial DNA and Y-Chromosome DNA)

mtDNA



Cavalli-Sforza & Feldman 2003

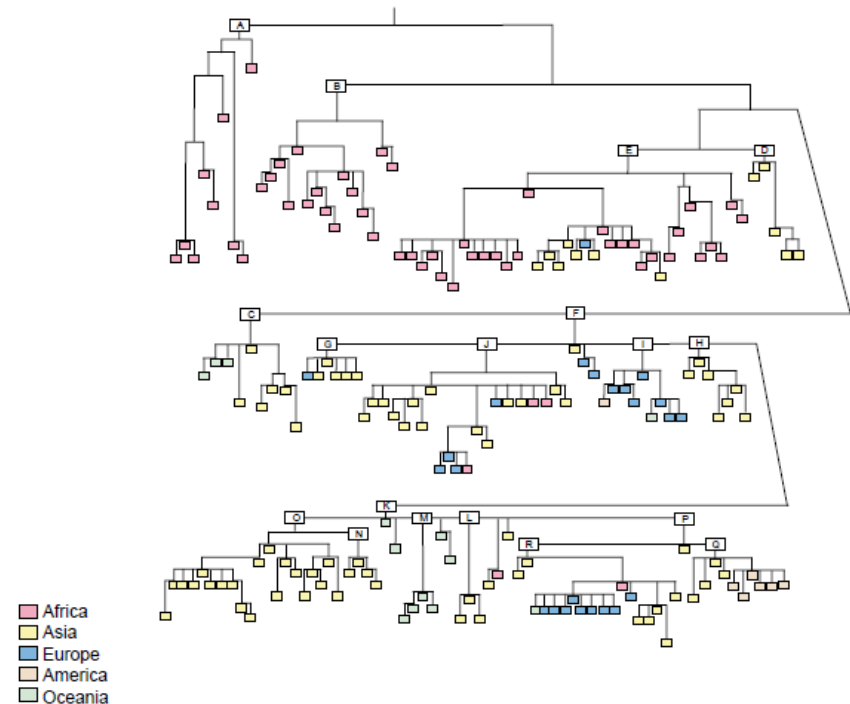


Phylogeography

Phylogeography: a method to analyze the geographical distribution of different clades within a phylogeny

- Optimal for uni-parental loci (mitochondrial DNA and Y-Chromosome DNA)

Y-chromosome DNA



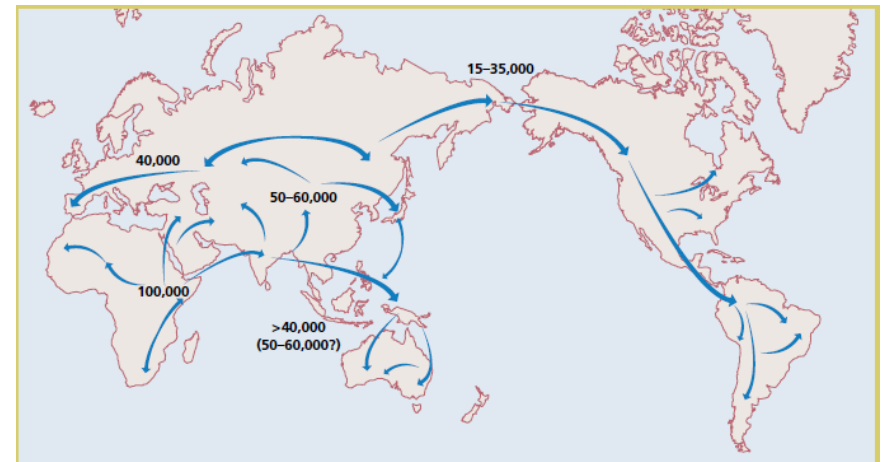
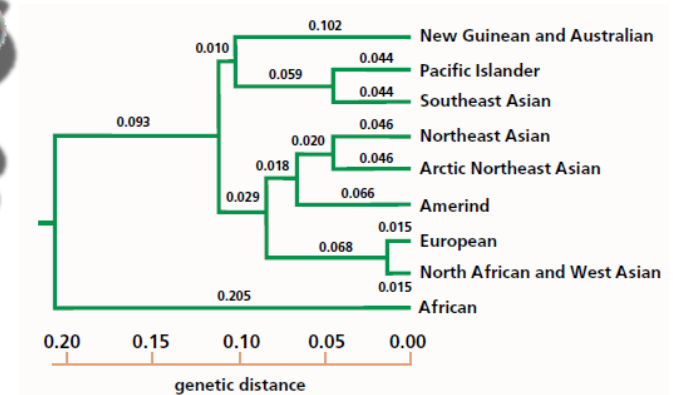
Cavalli-Sforza & Feldman 2003



Phylogeography

Phylogeography: a method to analyze the geographical distribution of different clades within a phylogeny

- Optimal for uni-parental loci (mitochondrial DNA and Y-Chromosome DNA)
- When assuming a certain mutation rate, it is possible to date the divergence of different clades within a phylogeny
- Geographical distribution and dating theoretically together allow inferences on key events in human evolution



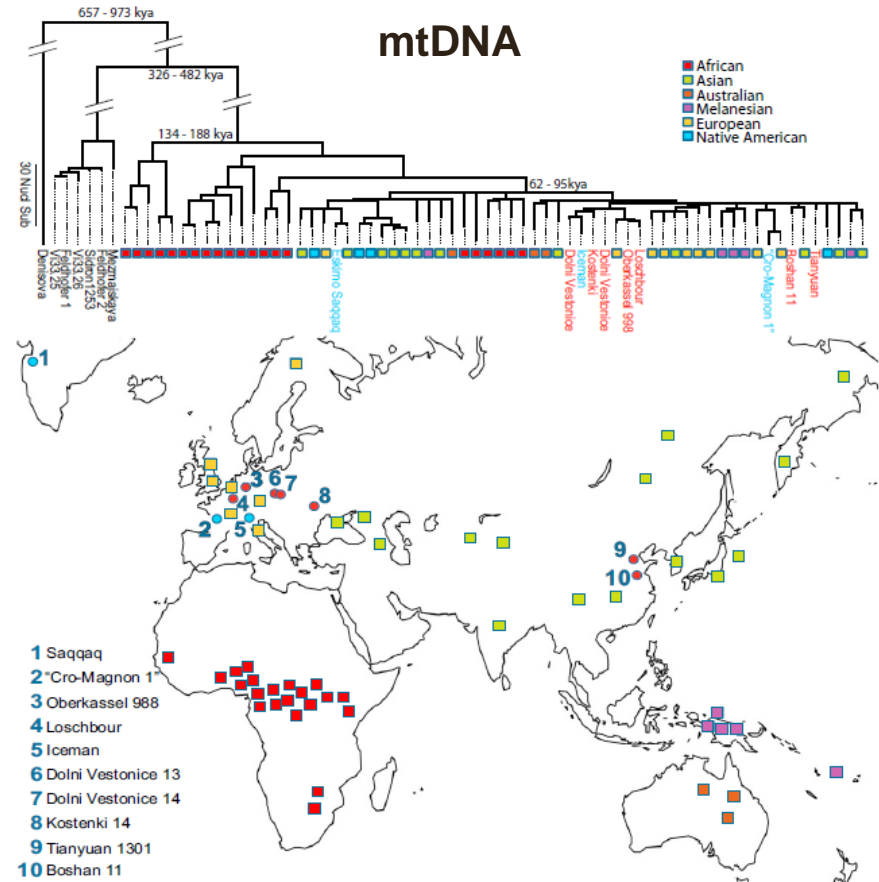
Cavalli-Sforza & Feldman 2003



Phylogeography

Phylogeography: a method to analyze the geographical distribution of different clades within a phylogeny

- Optimal for uni-parental loci (mitochondrial DNA and Y-Chromosome DNA)
- When assuming a certain mutation rate, it is possible to date the divergence of different clades within a phylogeny
- Geographical distribution and dating theoretically together allow inferences on key events in human evolution
- fossils and ancient DNA allow higher resolution across time and space



Fu et al 2013

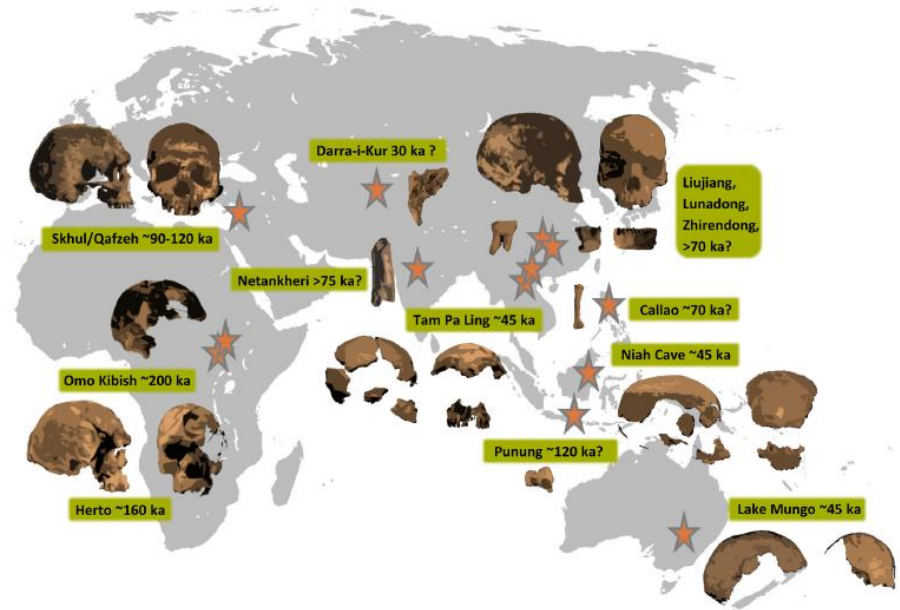


Phylogeography

Phylogeography: a method to analyze the geographical distribution of different clades within a phylogeny

- Optimal for uni-parental loci (mitochondrial DNA and Y-Chromosome DNA)
- When assuming a certain mutation rate, it is possible to date the divergence of different clades within a phylogeny
- Geographical distribution and dating theoretically together allow inferences on key events in human evolution
- fossils and ancient DNA allow higher resolution across time and space

Fossils

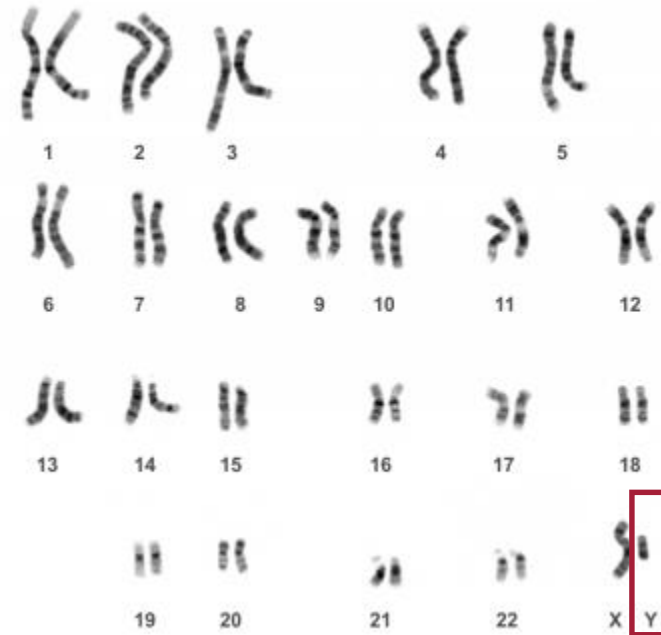
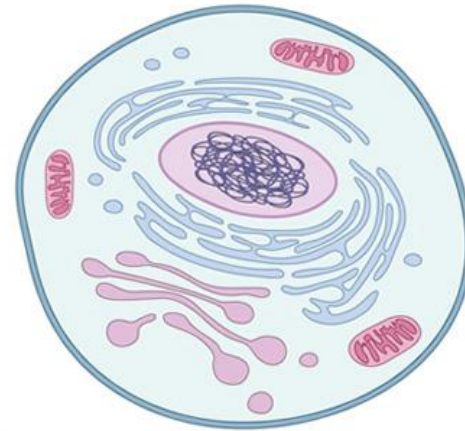


Reyes-Centeno 2016



Limitations of uni-parental loci

- A tiny part of our total inheritance!
- The autosome (non-sex chromosomes) are excluded
- Y-chromosome changes dependant to some extent on father's age during conception (mutations are higher with older age, which could inflate dates of divergence)

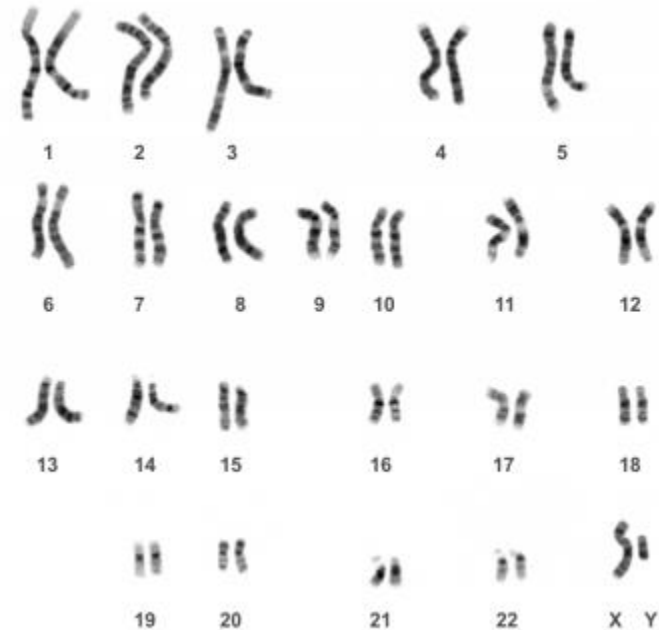
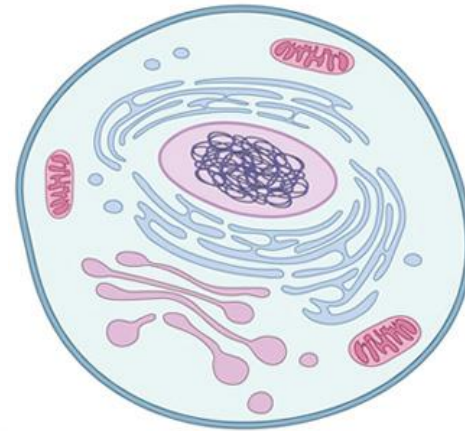


Nature Education 2010; 23andMe

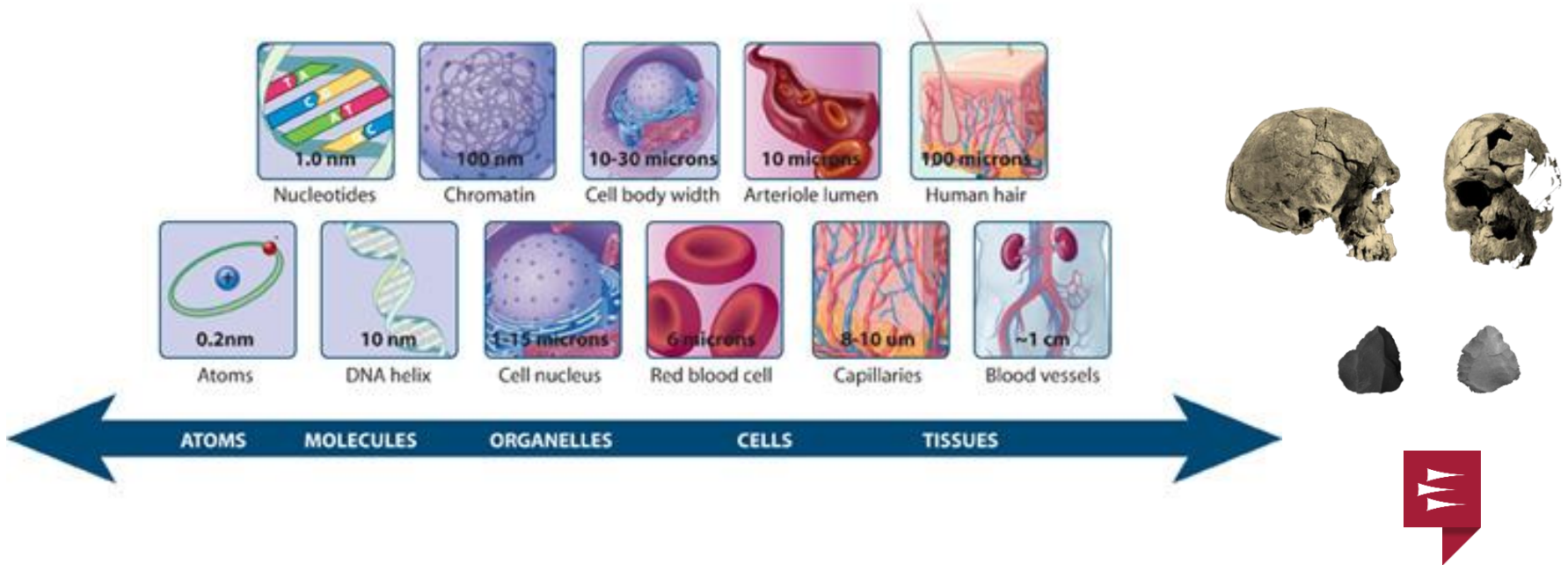


Genome-wide approaches

- ❑ **Microsatellites**
Non-functional regions of the genome, consisting of repeating 2-6 basepairs; also referred to as short-tandem repeats (STRs)
- ❑ **Single-nucleotide polymorphisms (SNPs)**
Base substitutions, deletions, or insertions at functional and non-functional regions
- ❑ **Genomes**
The entire genetic information of an organism's nucleus (both autosomal and sex chromosomes; both functional and non-functional)



Nature Education 2010; [23andMe](#)



Nature Education 2010; White *et al.* 2003; Beyin 2013



Genome-wide approaches

Microsatellites

Non-functional regions of the genome, consisting of repeating 2-6 basepairs; also referred to as short-tandem repeats (STRs)

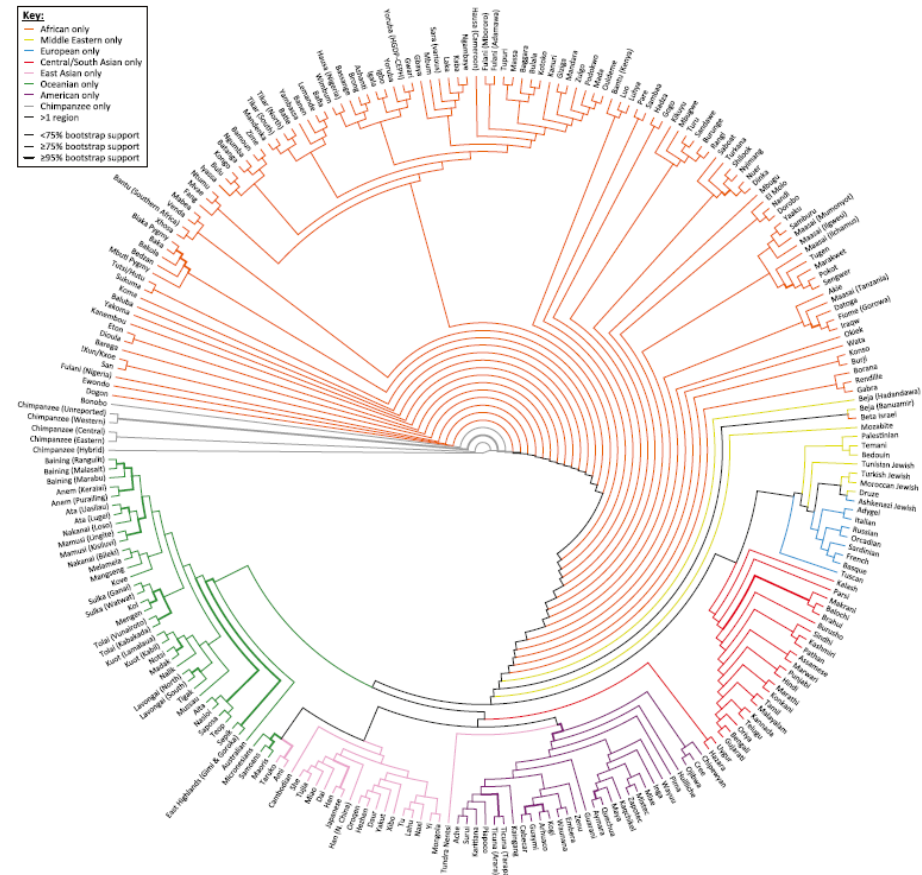
Single-nucleotide polymorphisms (SNPs)

Base substitutions, deletions, or insertions at functional and non-functional regions

Genomes

The entire genetic information of an organism's nucleus (both autosomal and sex chromosomes; both functional and non-functional)

STR DNA



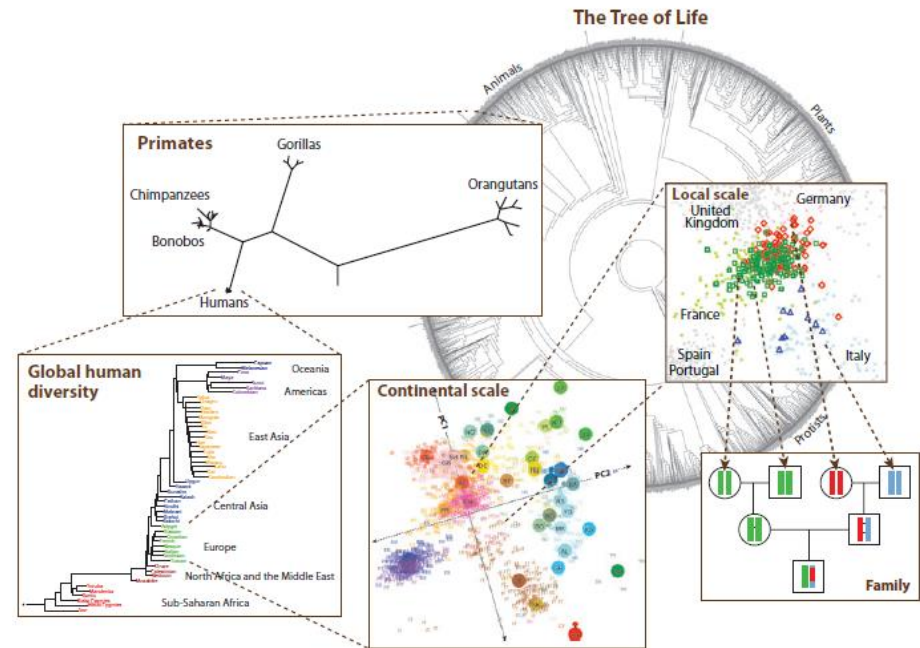
Pemberton et al 2013



Genome-wide approaches

- ❑ **Microsatellites**
 Non-functional regions of the genome, consisting of repeating 2-6 basepairs; also referred to as short-tandem repeats (STRs)
- ❑ **Single-nucleotide polymorphisms (SNPs)**
 Base substitutions, deletions, or insertions at functional and non-functional regions
- ❑ **Genomes**
 The entire genetic information of an organism's nucleus (both autosomal and sex chromosomes; both functional and non-functional)

SNPs DNA



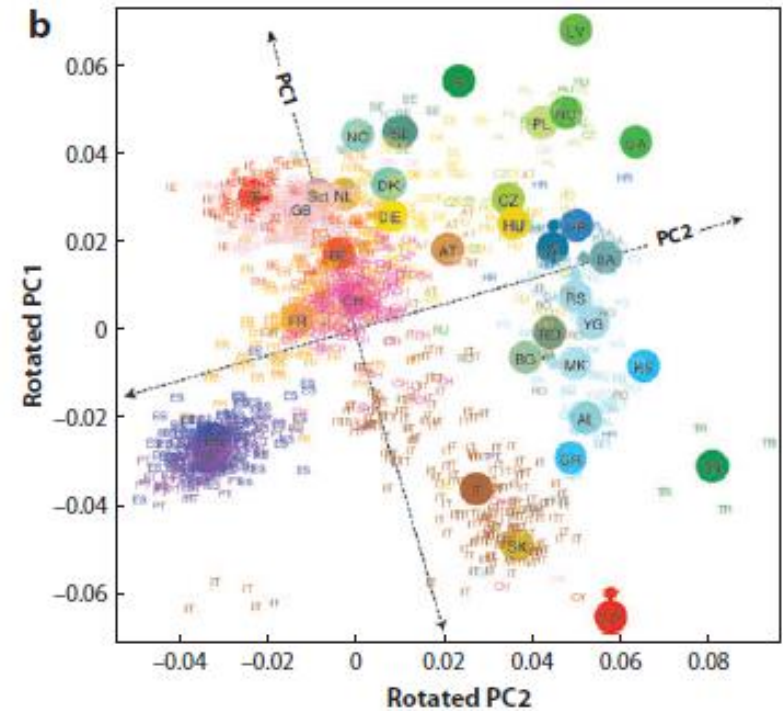
Novembre & Ramachandran 2011



Genome-wide approaches

- ❑ **Microsatellites**
Non-functional regions of the genome, consisting of repeating 2-6 basepairs; also referred to as short-tandem repeats (STRs)
- ❑ **Single-nucleotide polymorphisms (SNPs)**
Base substitutions, deletions, or insertions at functional and non-functional regions
- ❑ **Genomes**
The entire genetic information of an organism's nucleus (both autosomal and sex chromosomes; both functional and non-functional)

SNPs DNA



Novembre & Ramachandran 2011



Genome-wide approaches

- ❑ **Microsatellites**
Non-functional regions of the genome, consisting of repeating 2-6 basepairs; also referred to as short-tandem repeats (STRs)
- ❑ **Single-nucleotide polymorphisms (SNPs)**
Base substitutions, deletions, or insertions at functional and non-functional regions
- ❑ **Genomes**
The entire genetic information of an organism's nucleus (both autosomal and sex chromosomes; both functional and non-functional)

SNPs DNA



Novembre & Ramachandran 2011



Genome-wide approaches

- ❑ **Microsatellites**
Non-functional regions of the genome, consisting of repeating 2-6 basepairs; also referred to as short-tandem repeats (STRs)
- ❑ **Single-nucleotide polymorphisms (SNPs)**
Base substitutions, deletions, or insertions at functional and non-functional regions
- ❑ **Genomes**
The entire genetic information of an organism's nucleus (both autosomal and sex chromosomes; both functional and non-functional)

SNPs DNA



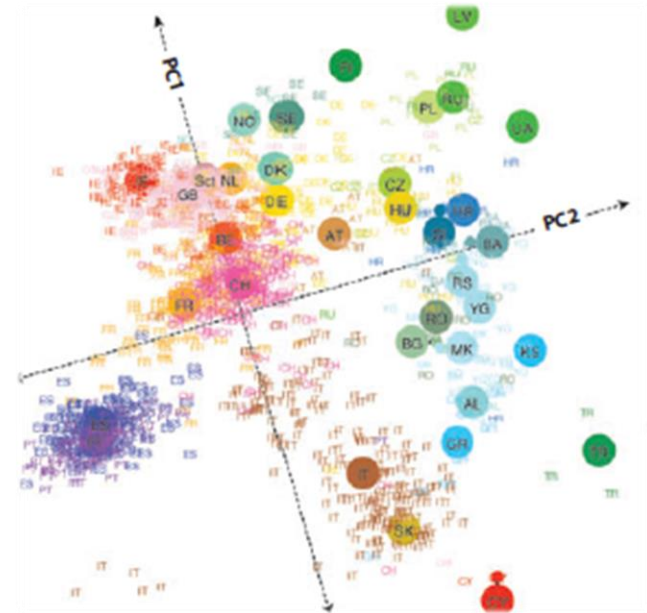
Novembre & Ramachandran 2011



Genome-wide approaches

- ❑ **Microsatellites**
Non-functional regions of the genome, consisting of repeating 2-6 basepairs; also referred to as short-tandem repeats (STRs)
- ❑ **Single-nucleotide polymorphisms (SNPs)**
Base substitutions, deletions, or insertions at functional and non-functional regions
- ❑ **Genomes**
The entire genetic information of an organism's nucleus (both autosomal and sex chromosomes; both functional and non-functional)

SNPs DNA



Novembre & Ramachandran 2011



Genome-wide approaches

□ Microsatellites

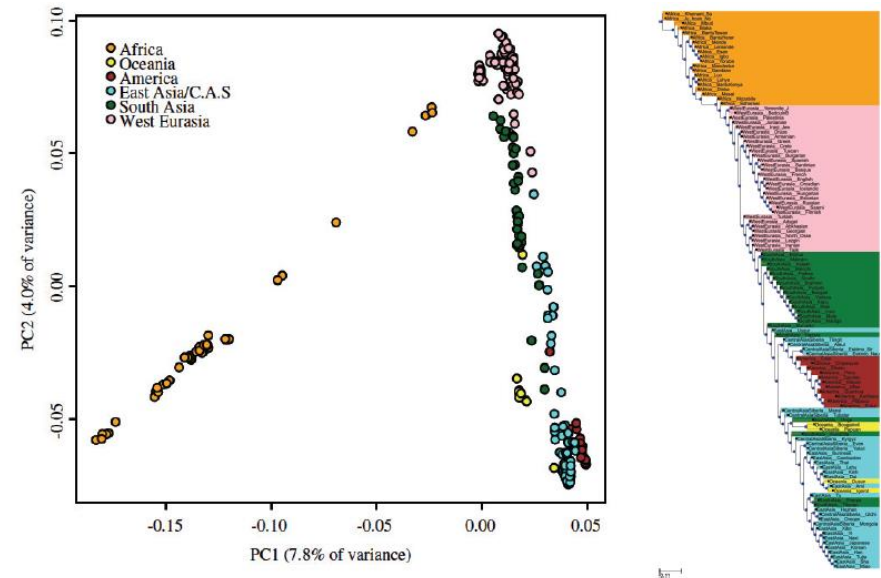
Non-functional regions of the genome, consisting of repeating 2-6 basepairs; also referred to as short-tandem repeats (STRs)

□ Single-nucleotide polymorphisms (SNPs)

Base substitutions, deletions, or insertions at functional and non-functional regions

□ Genomes

The entire genetic information of an organism's nucleus (both autosomal and sex chromosomes; both functional and non-functional)

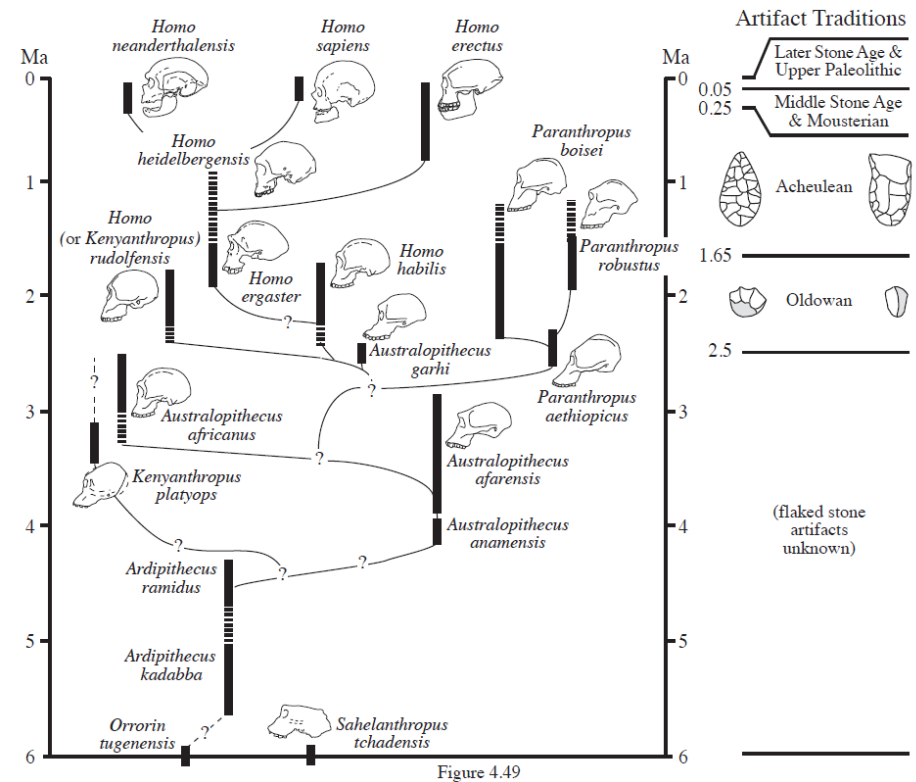


Mallick *et al.* 2017



Ancient genomes and fossils fill in the gaps...

- New fossil discoveries inform on the diversity of past populations across time and space
- DNA from these fossils can give us more data points of analysis in reconstructing a phylogeny

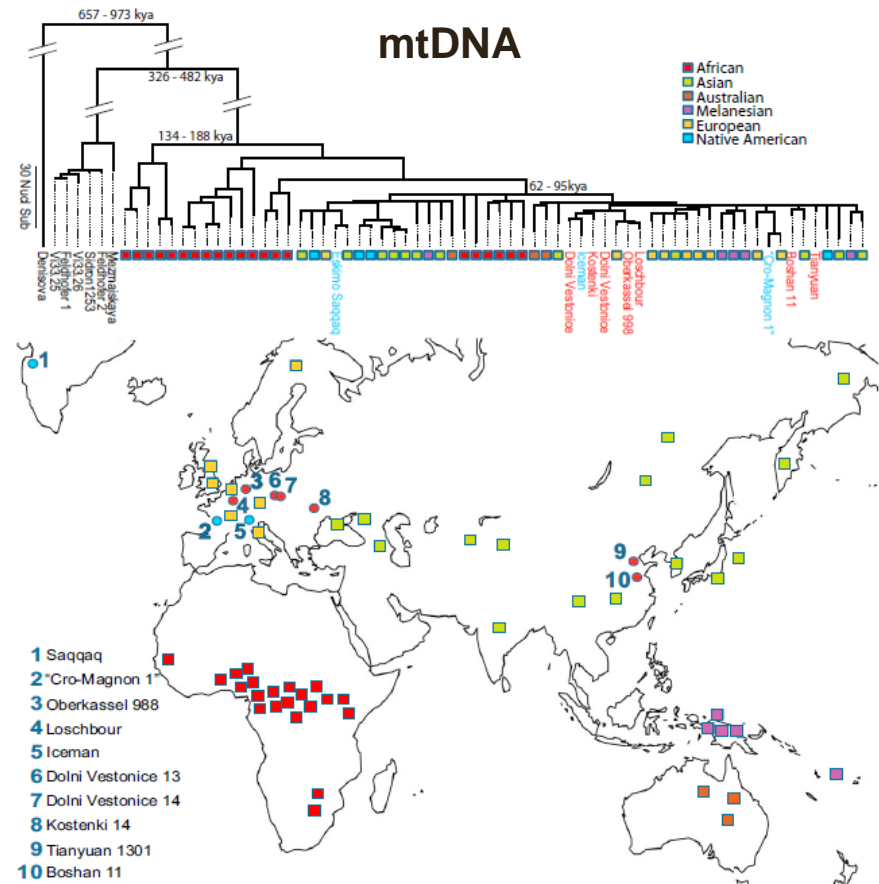


Klein 2009



Ancient genomes and fossils fill in the gaps...

- New fossil discoveries inform on the diversity of past populations across time and space
- DNA from these fossils can give us more data points of analysis in reconstructing a phylogeny

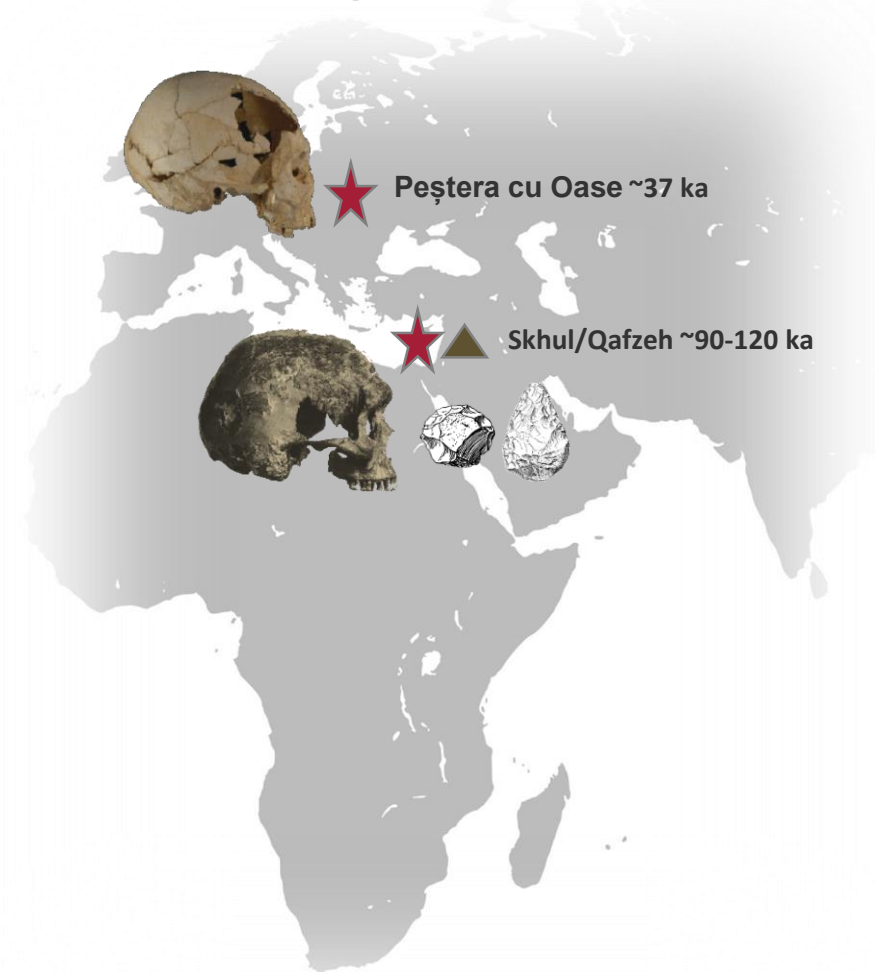


Fu et al 2013



Ancient genomes and fossils fill in the gaps...

- Neanderthal admixture
 - Neanderthal and modern human hybrids in the fossil record?

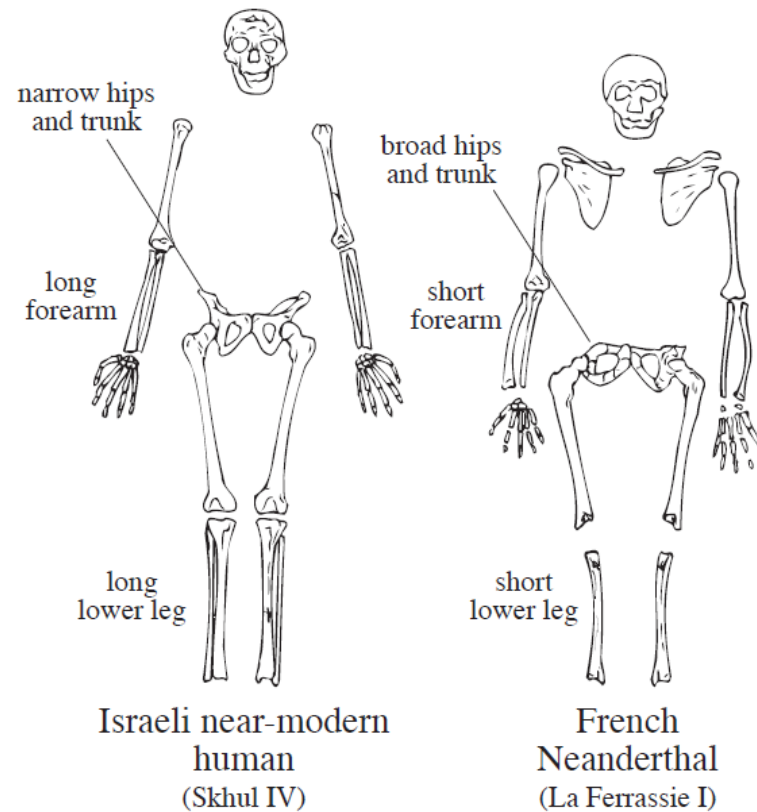




Ancient genomes and fossils fill in the gaps...

□ Neanderthal admixture

- Neanderthal and modern human hybrids in the fossil record?
- Some refer to these potential hybrids as transitional forms or „near-modern“ humans



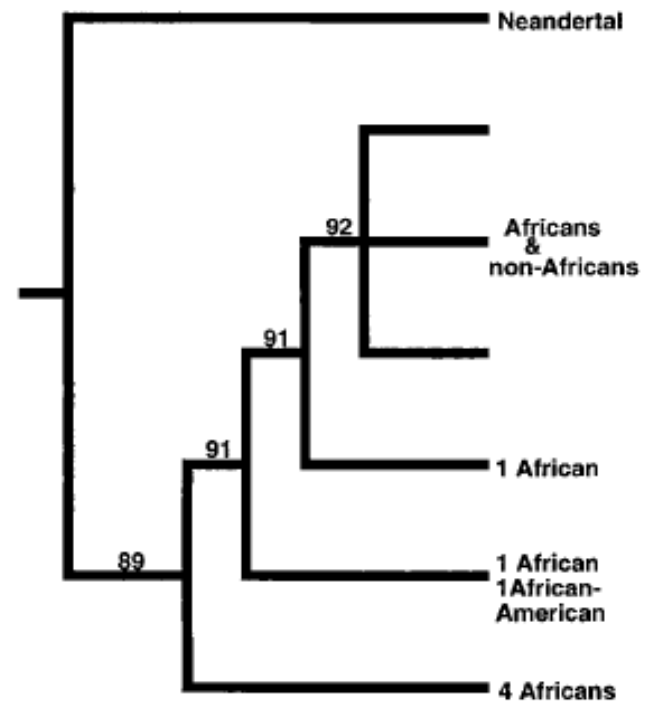
Klein 2009



Ancient genomes and fossils fill in the gaps...

□ Neanderthal admixture

- Neanderthal and modern human hybrids in the fossil record?
- Some refer to these potential hybrids as transitional forms or „near-modern“ humans
- No evidence from mtDNA



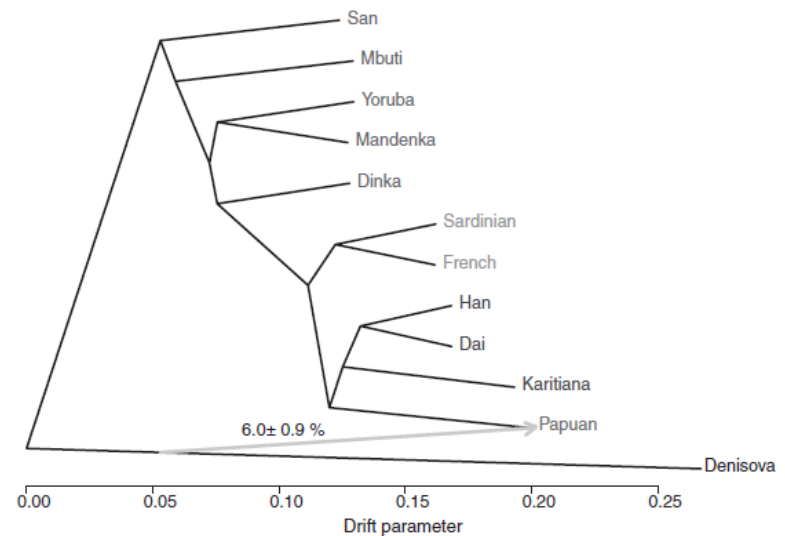
Krings et al 1997



Ancient genomes and fossils fill in the gaps...

- ❑ **Neanderthal admixture**
 - Neanderthal and modern human hybrids in the fossil record?
 - Some refer to these potential hybrids as transitional forms or „near-modern“ humans
 - No evidence from mtDNA
- ❑ **Other ancient admixture**
 - Hominin from Denisova Cave, Russia

...and break down the tree



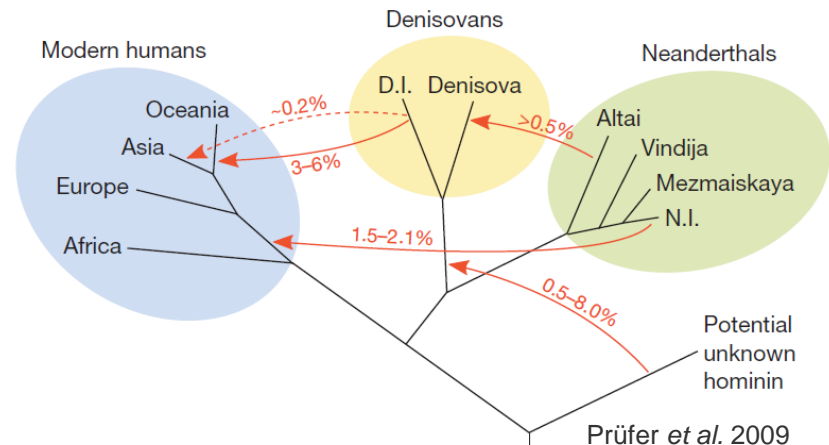
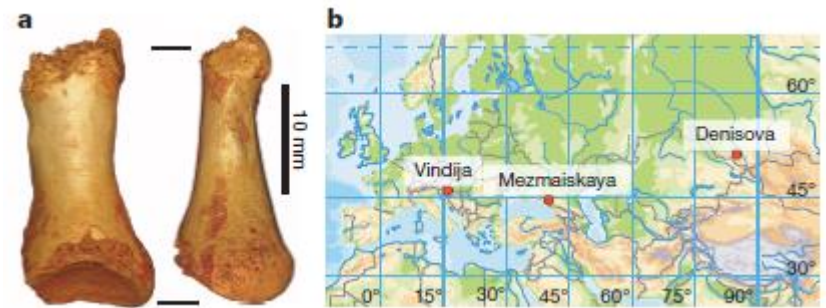
Meyer *et al.* 2012



Ancient genomes and fossils fill in the gaps...

- ❑ **Neanderthal admixture**
 - Neanderthal and modern human hybrids in the fossil record?
 - Some refer to these potential hybrids as transitional forms or „near-modern“ humans
 - No evidence from mtDNA
- ❑ **Other ancient admixture**
 - Hominin from Denisova Cave, Russia
 - Neanderthal from Denisova Cave
 - Human history more like a web instead of a tree?

...and break down the tree

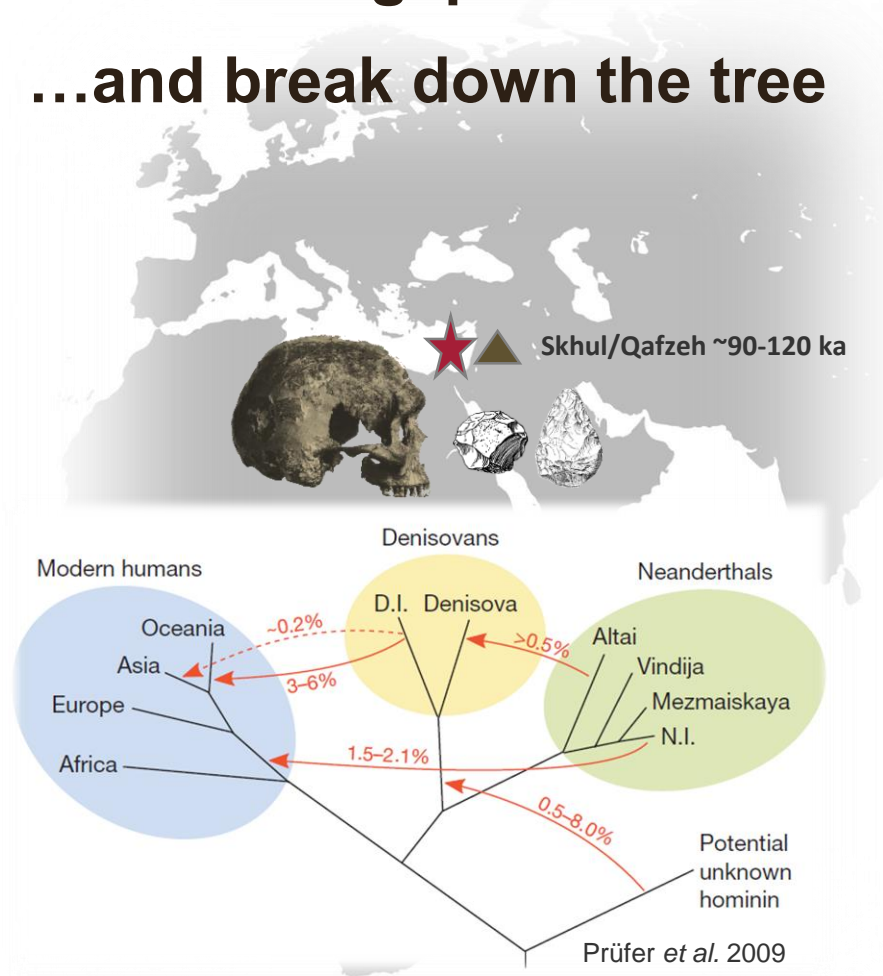




Ancient genomes and fossils fill in the gaps...

- ❑ **Neanderthal admixture**
 - Neanderthal and modern human hybrids in the fossil record?
 - Some refer to these potential hybrids as transitional forms or „near-modern“ humans
 - No evidence from mtDNA
- ❑ **Other ancient admixture**
 - Hominin from Denisova Cave, Russia
 - Neanderthal from Denisova Cave
 - Human history more like a web instead of a tree?

...and break down the tree

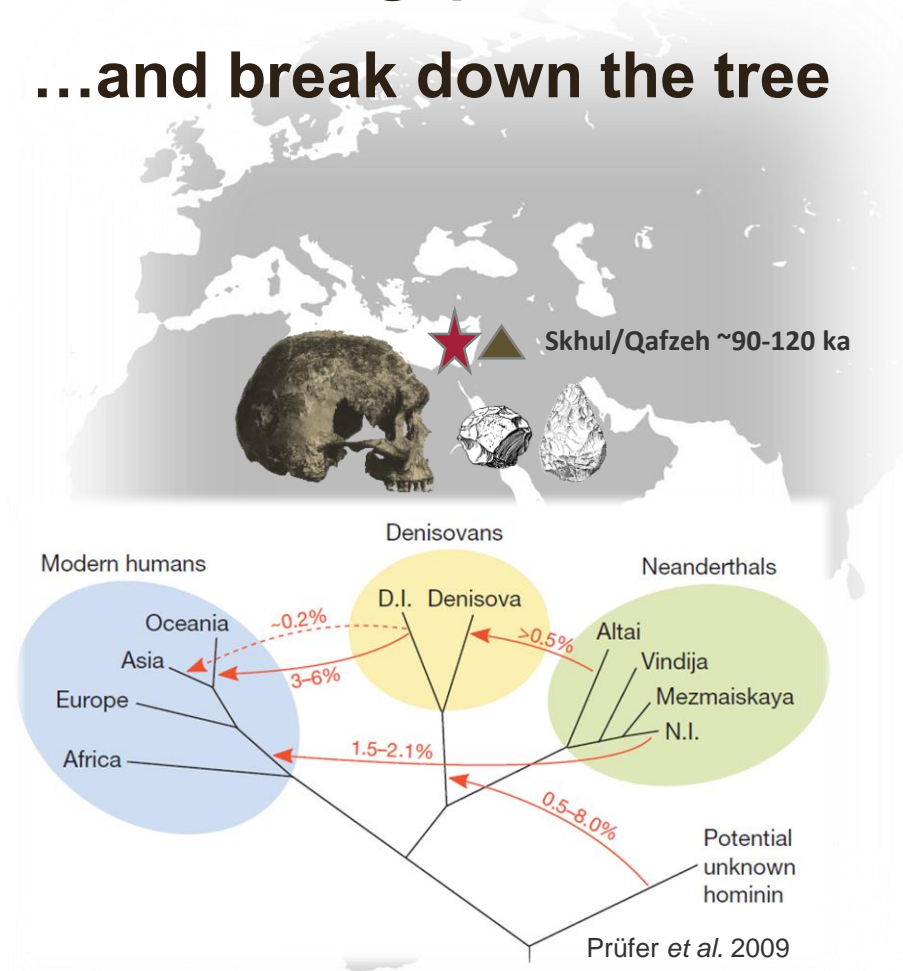




Ancient genomes and fossils fill in the gaps...

- ❑ **Neanderthal admixture**
 - Neanderthal and modern human hybrids in the fossil record?
 - Some refer to these potential hybrids as transitional forms or „near-modern“ humans
 - No evidence from mtDNA
- ❑ **Other ancient admixture**
 - Hominin from Denisova Cave, Russia
 - Neanderthal from Denisova Cave
 - Human history more like a web instead of a tree?

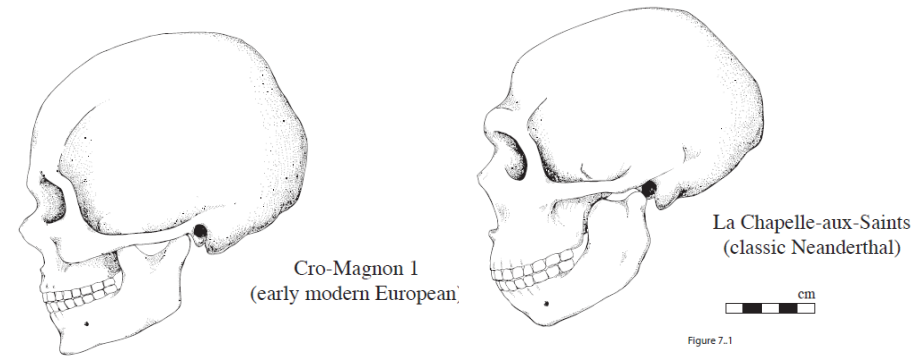
...and break down the tree



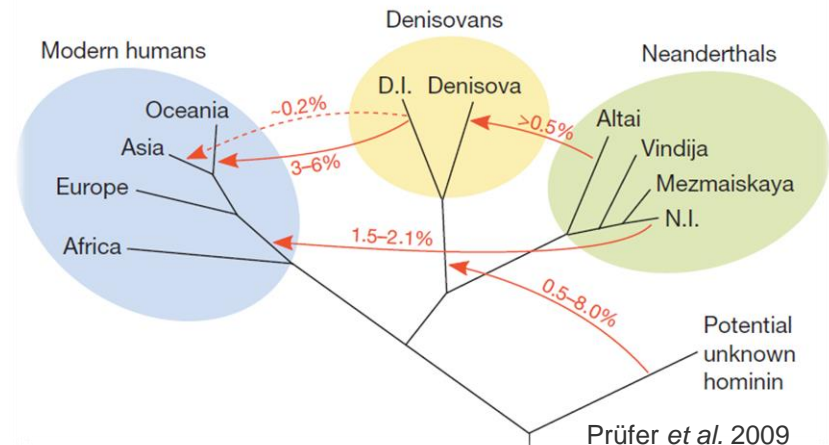


Species concepts and speciation

- ❑ **Biological species**
Reproductively isolated populations. Interbreeding is possible by species recognition and compatible fertilization system.
- ❑ **Morphological species**
Phenetic clustering of organisms based on anatomical traits
- ❑ **Genetic species**
Genetic clustering of organisms based on genetic frequencies
- ❑ **Phylogenetic species**
- ❑ **Evolutionary species**
Ancestral-descendant sequence of populations terminating by extinction



Klein 2009





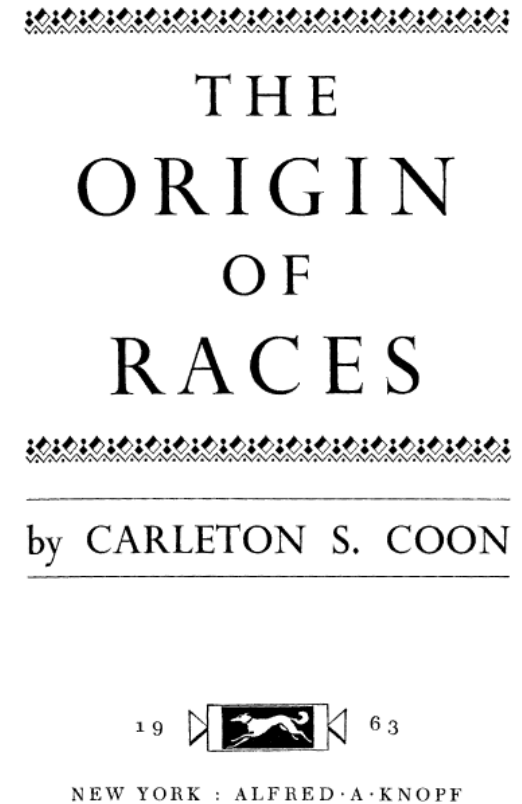
Models of anthropogeny

What model of modern human origins is best supported with the current fossil and genomic evidence?



Hypotheses on modern human origins

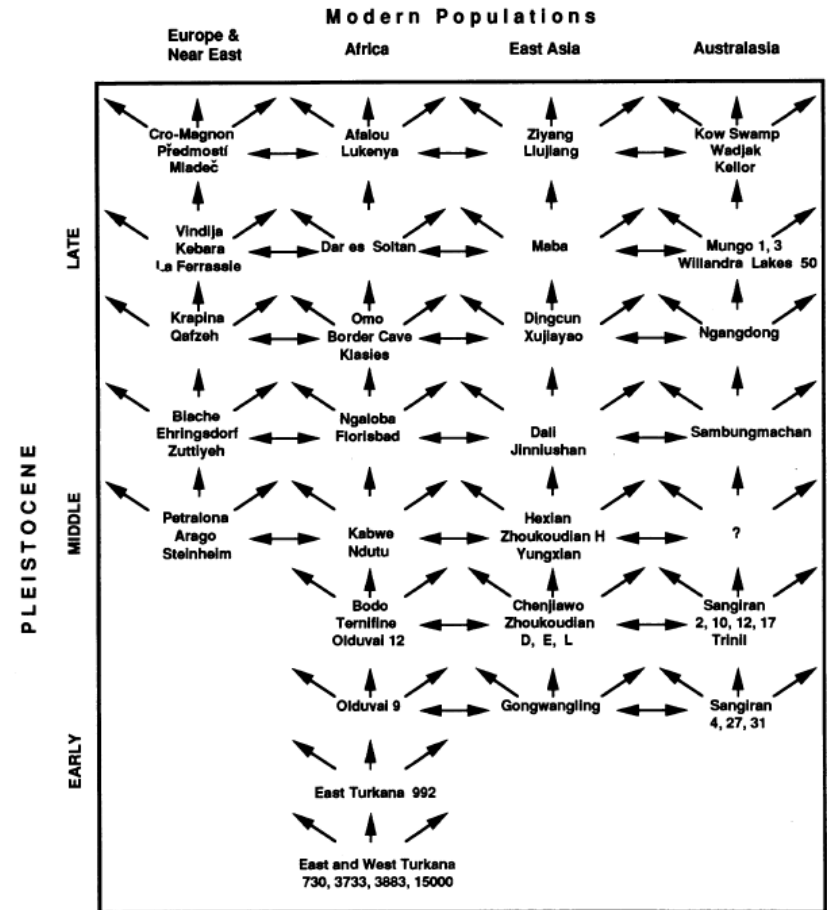
- ❑ **Multiple Origins**
Multiple origins in different continents
and convergent evolution





Hypotheses on modern human origins

- ❑ **Multiple Origins**
Multiple origins in different continents and convergent evolution
- ❑ **Multiregional Evolution**
Dynamic admixture within between populations across time and space

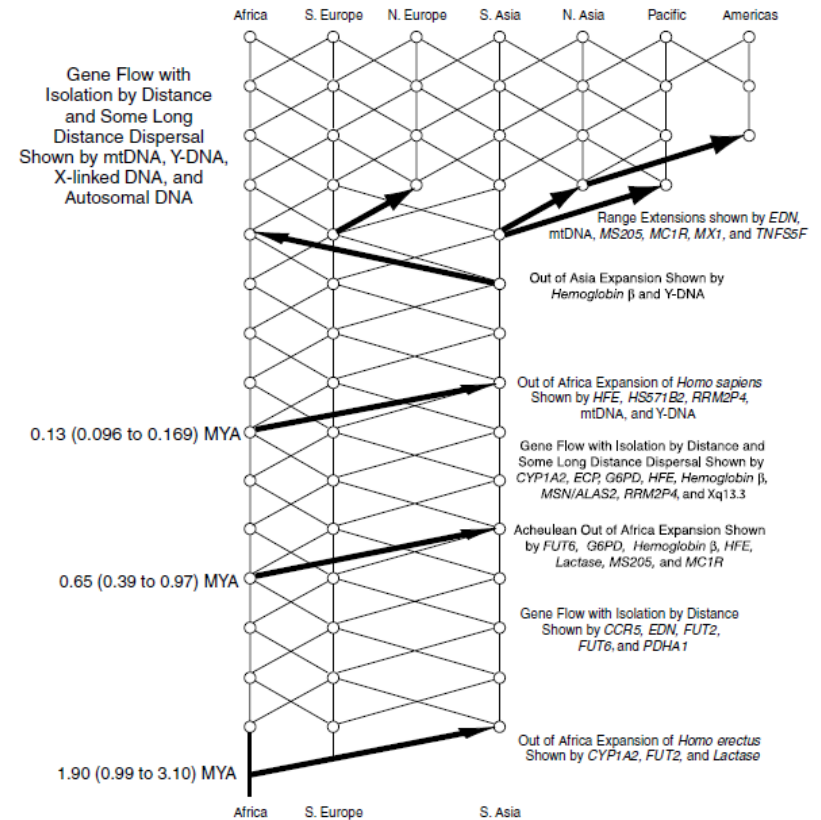


Frayser et al. 1993



Hypotheses on modern human origins

- ❑ **Multiple Origins**
Multiple origins in different continents and convergent evolution
- ❑ **Multiregional Evolution**
Dynamic admixture within between populations across time and space

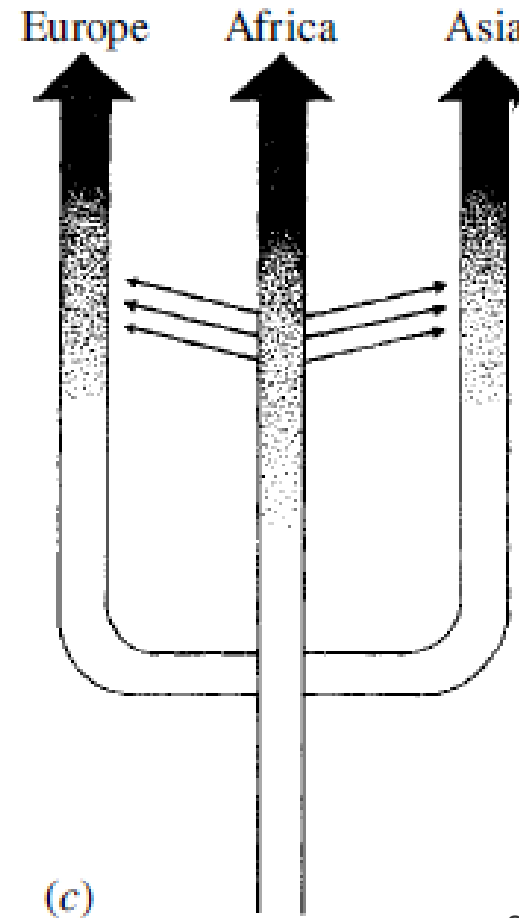


Templeton 2007



Hypotheses on modern human origins

- ❑ **Multiple Origins**
Multiple origins in different continents and convergent evolution
- ❑ **Multiregional Evolution**
Dynamic admixture within between populations across time and space
- ❑ **Assimilation**
Multiregional evolution and assimilation of expanding modern humans

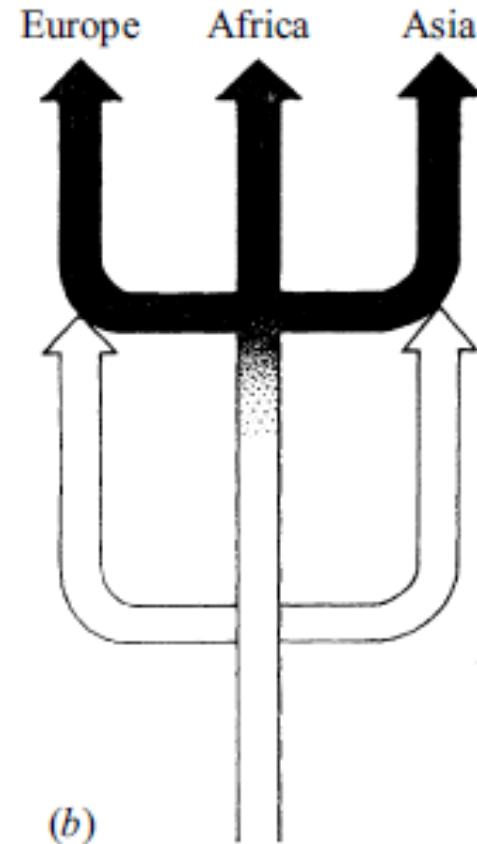


Stringer 2001



Hypotheses on modern human origins

- ❑ **Multiple Origins**
Multiple origins in different continents and convergent evolution
- ❑ **Multiregional Evolution**
Dynamic admixture within between populations across time and space
- ❑ **Assimilation**
Multiregional evolution and assimilation of expanding modern humans
- ❑ **Out-of-Africa [and replacement]**
Modern human origins in Africa and replacement of non-modern humans
- ❑ **Out-of-Africa, hybridization, and replacement**

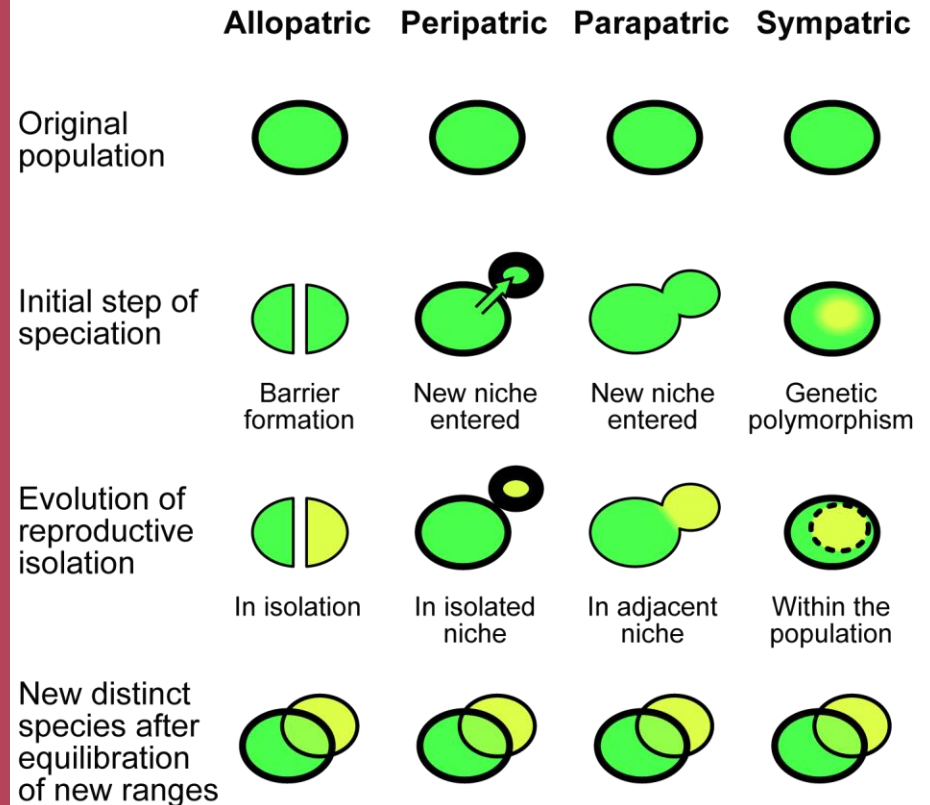


Stringer 2001



Hypotheses on modern human origins

- ❑ **Biological species**
Reproductively isolated populations. Interbreeding is possible by species recognition and compatible fertilization system.
- ❑ **Morphological species**
Phenetic clustering of organisms based on anatomical traits
- ❑ **Genetic species**
Genetic clustering of organisms based on genetic frequencies
- ❑ **Phylogenetic species**
- ❑ **Evolutionary species**
Ancestral-descendant sequence of populations terminating by extinction



I. Karonen 2006



Hypotheses on modern human origins

- ❑ **Multiple Origins**
Multiple origins in different continents and convergent evolution
- ❑ **Multiregional Evolution**
Dynamic admixture within between populations across time and space
- ❑ **Assimilation**
Multiregional evolution and assimilation of expanding modern humans
- ❑ **Out-of-Africa [and replacement]**
Modern human origins in Africa and replacement of non-modern humans
- ❑ **Out-of-Africa, hybridization, and replacement**

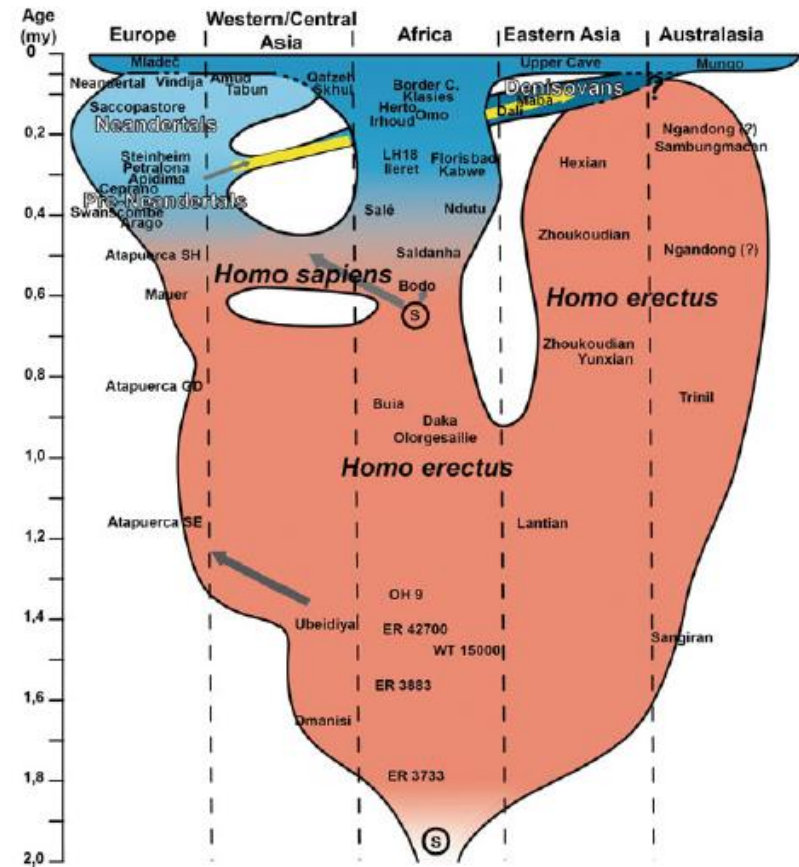


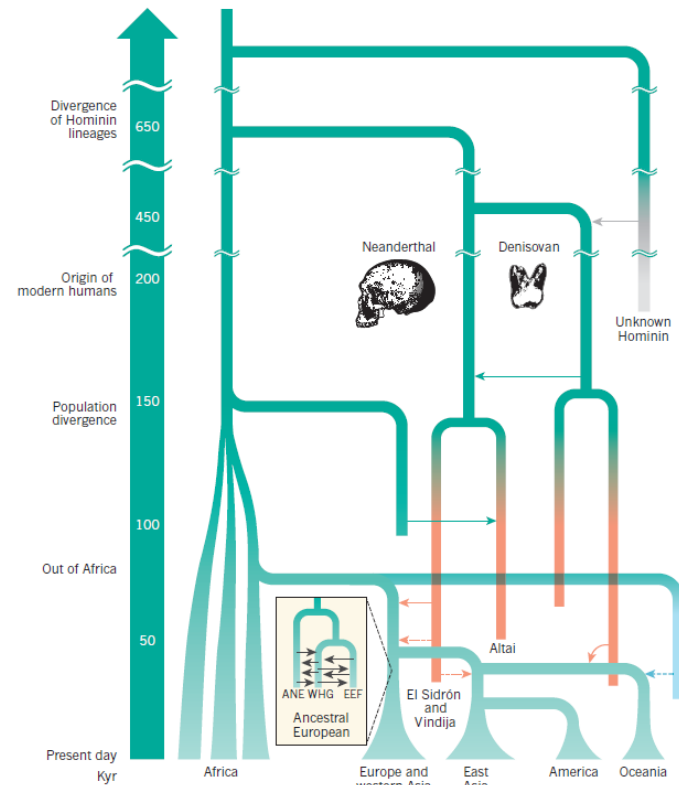
Fig. 7 Origin and evolution of *Homo sapiens*

Bräuer 2015



Hypotheses on modern human origins

- ❑ **Multiple Origins**
Multiple origins in different continents and convergent evolution
- ❑ **Multiregional Evolution**
Dynamic admixture within between populations across time and space
- ❑ **Assimilation**
Multiregional evolution and assimilation of expanding modern humans
- ❑ **Out-of-Africa [and replacement]**
Modern human origins in Africa and replacement of non-modern humans
- ❑ **Out-of-Africa, hybridization, and replacement**

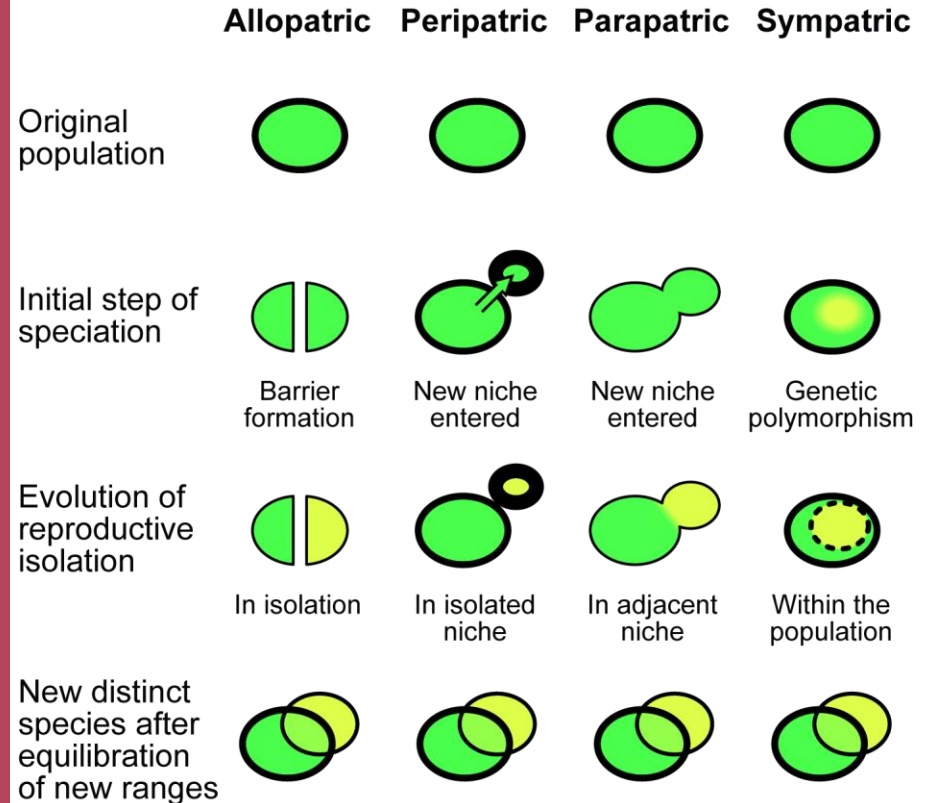


Nielsen et al 2017



Species concepts and speciation

- ❑ **Biological species**
Reproductively isolated populations. Interbreeding is possible by species recognition and compatible fertilization system.
- ❑ **Morphological species**
Phenetic clustering of organisms based on anatomical traits
- ❑ **Genetic species**
Genetic clustering of organisms based on genetic frequencies
- ❑ **Phylogenetic species**
- ❑ **Evolutionary species**
Ancestral-descendant sequence of populations terminating by extinction



I. Karonen 2006



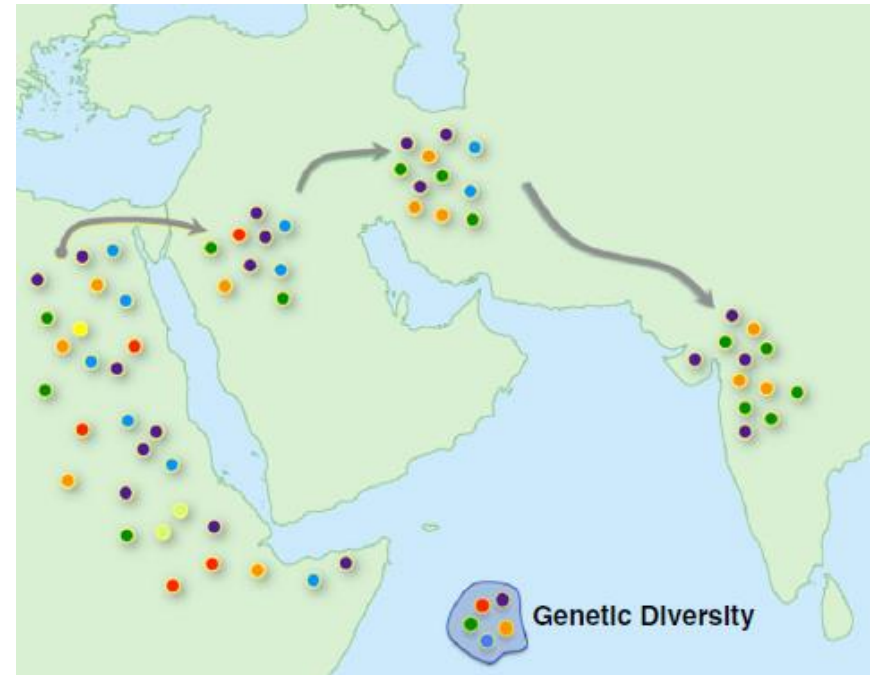
Modern human dispersals

When, how, and why did anatomically modern humans disperse out of Africa?



The serial founder effect model

- ❑ Cascading bottlenecks or serial founding events

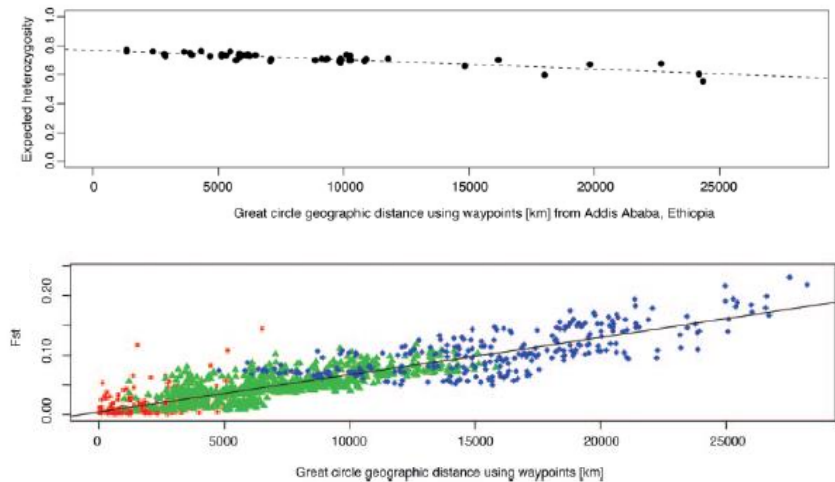


Henn et al. 2012



The serial founder effect model

- Cascading bottlenecks or serial founding events
 1. Decreasing intra-population diversity with geographical distance from Africa
 2. Increasing inter-population diversity between populations with geographical distance separating them

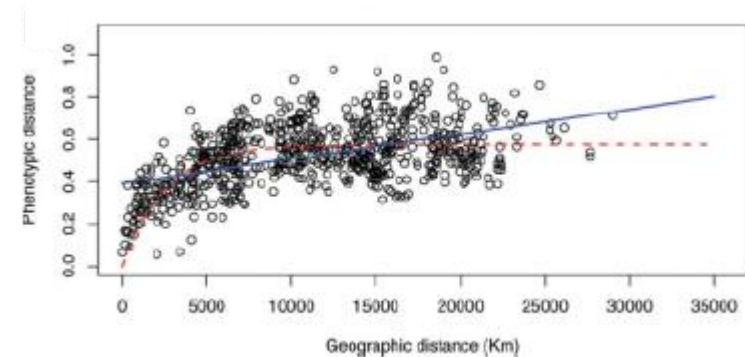
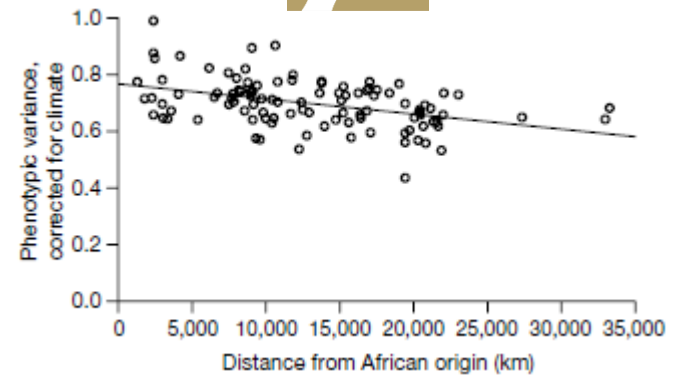


Ramachandran et al. 2005



The serial founder effect model

- Cascading bottlenecks or serial founding events
 1. Decreasing intra-population diversity with geographical distance from Africa
 2. Increasing inter-population diversity between populations with geographical distance separating them

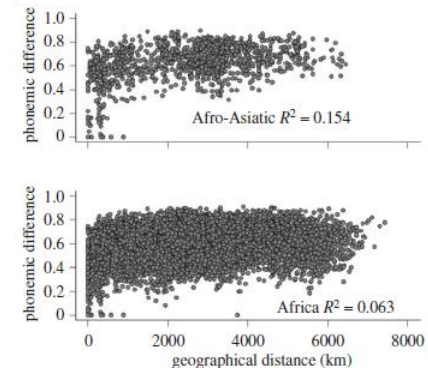
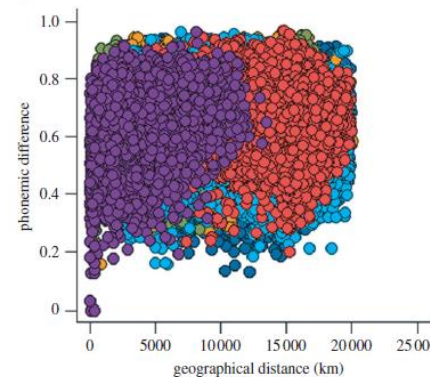
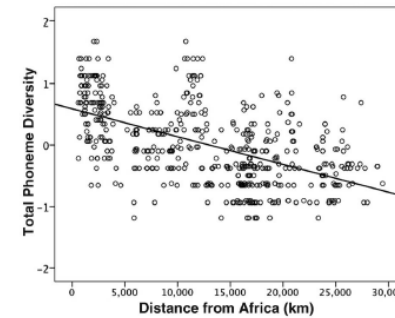


Manica et al. 2007; Betti et al. 2011



The serial founder effect model

- Cascading bottlenecks or serial founding events
 1. Decreasing intra-population diversity with geographical distance from Africa
 2. Increasing inter-population diversity between populations with geographical distance separating them

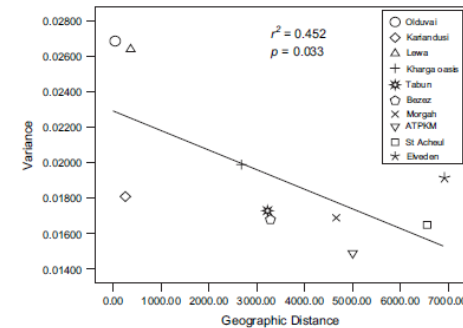


Atkinson 2011; Hunley et al. 2012



The serial founder effect model

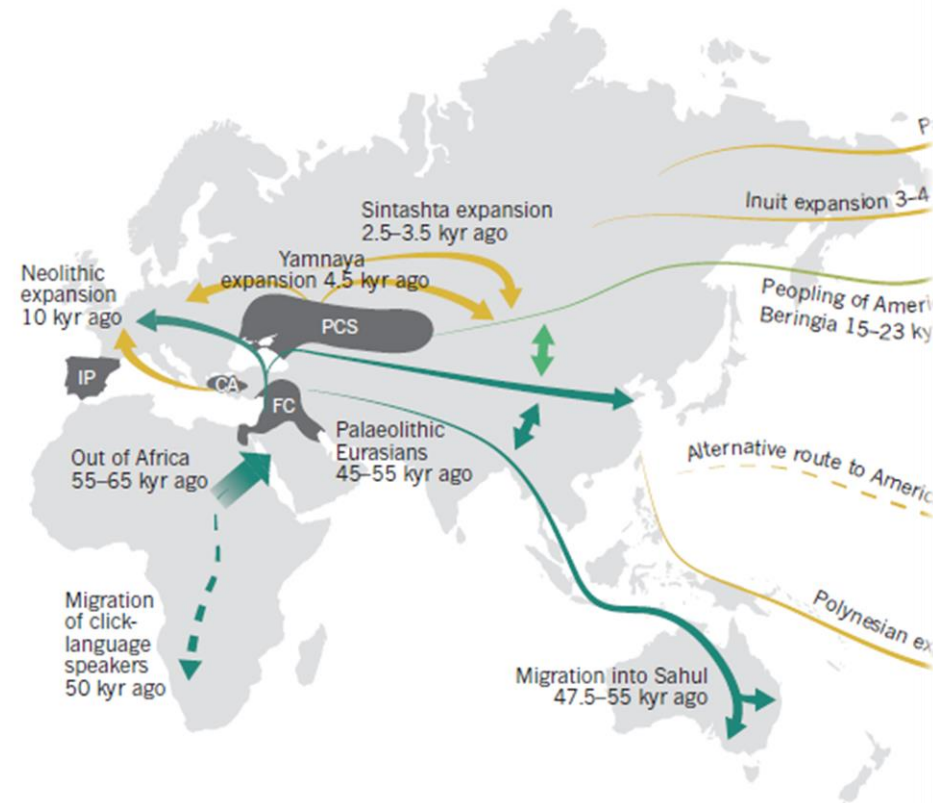
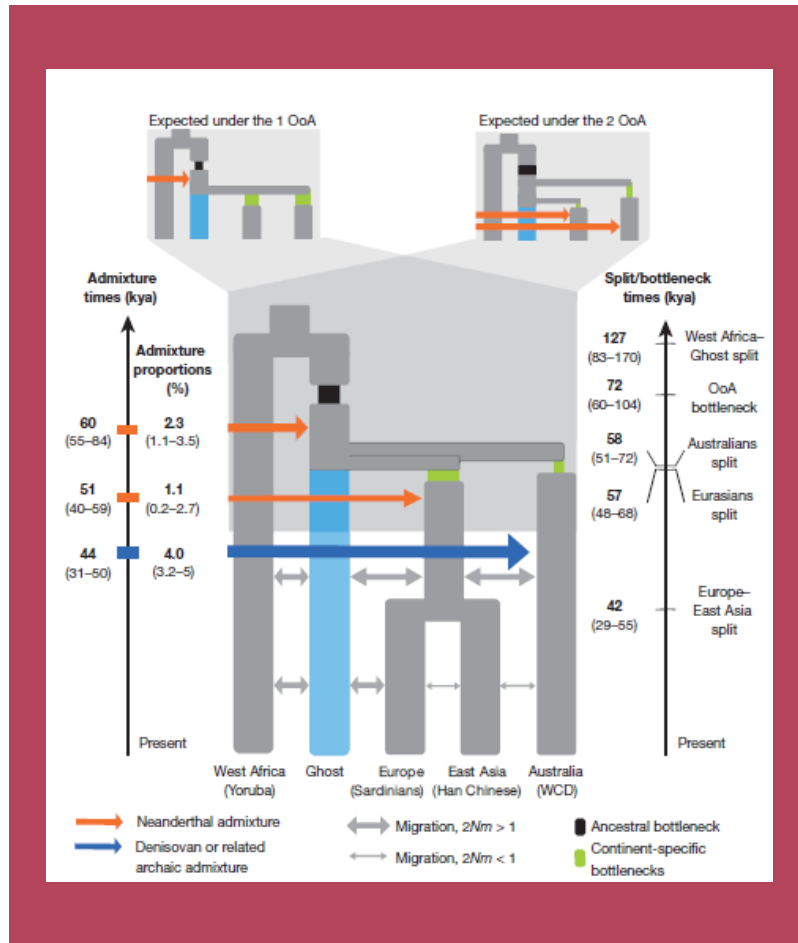
- Cascading bottlenecks or serial founding events
 1. Decreasing intra-population diversity with geographical distance from Africa
 2. Increasing inter-population diversity between populations with geographical distance separating them



Lycett & von Cramon-Taubadel 2008



Competing models



Nielsen et al. 2017

