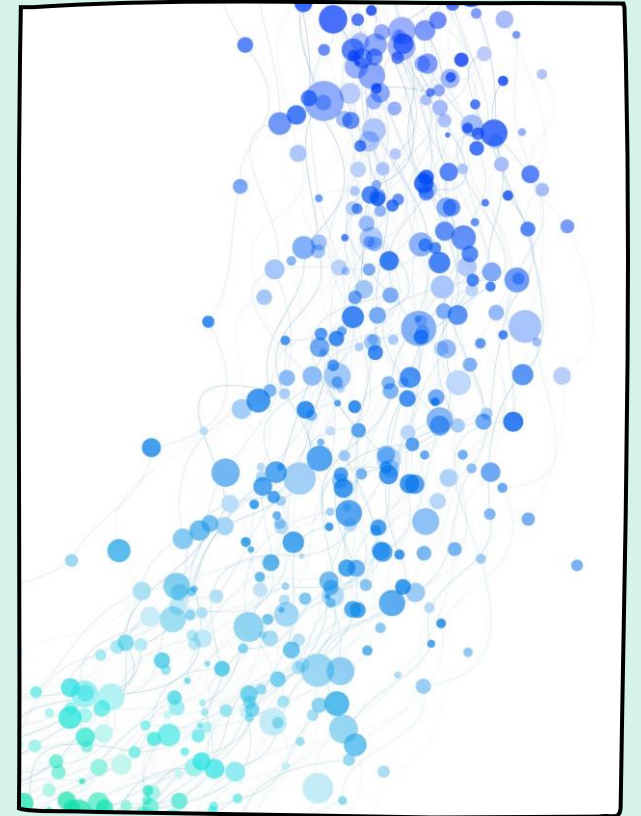
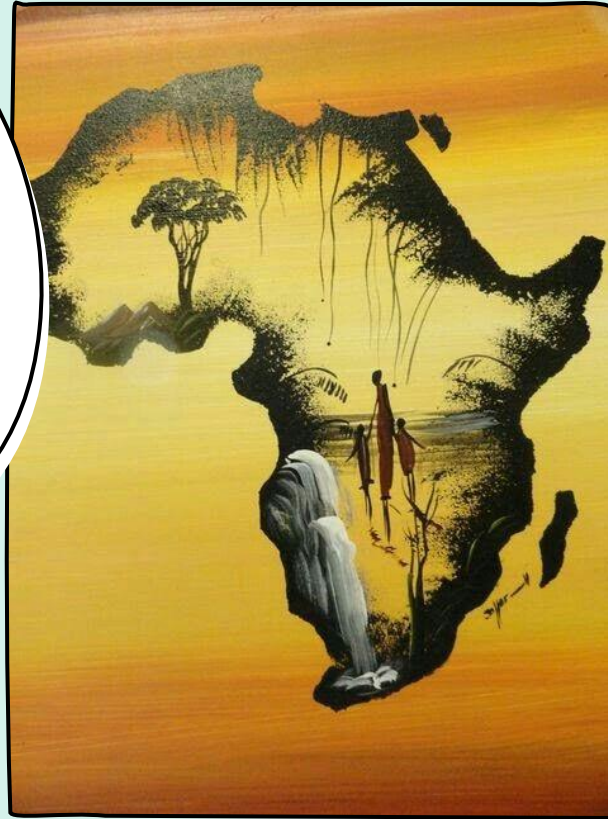


MODERN HUMANS I
ORIGIN OF A NEW
POPULATION IN AFRICA AND
ITS EXPANSION(S)

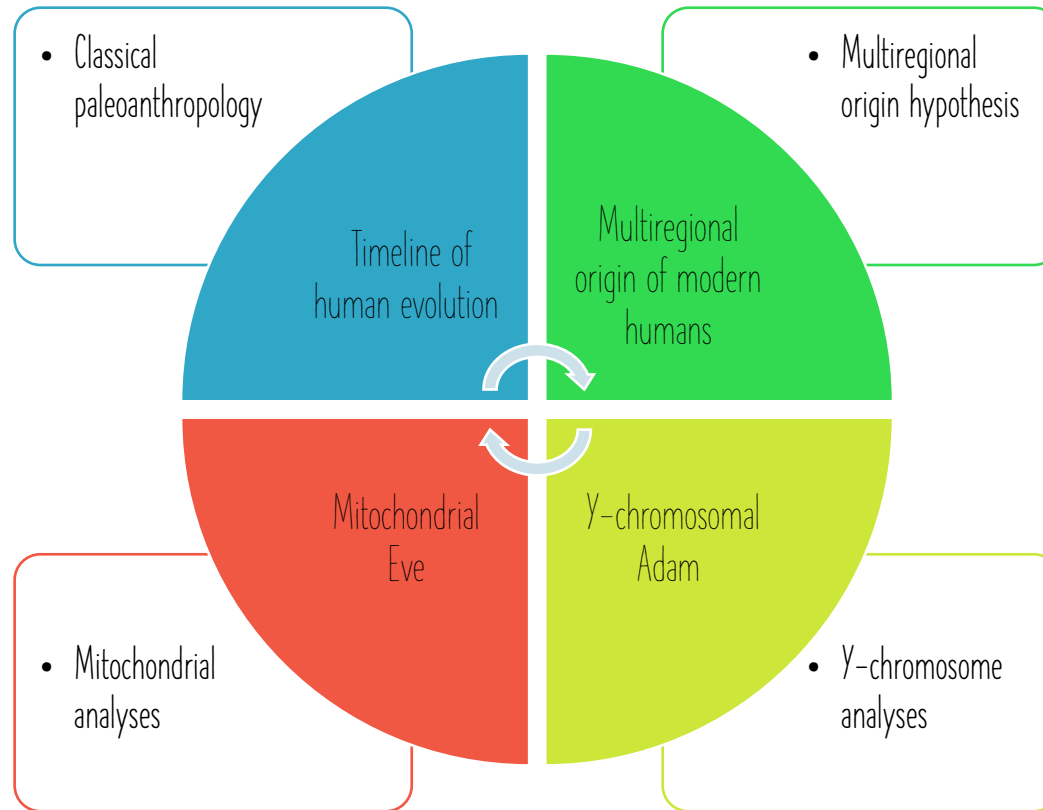
Paleoanthropology Bi8610

Arwa Kharobi, PhD.

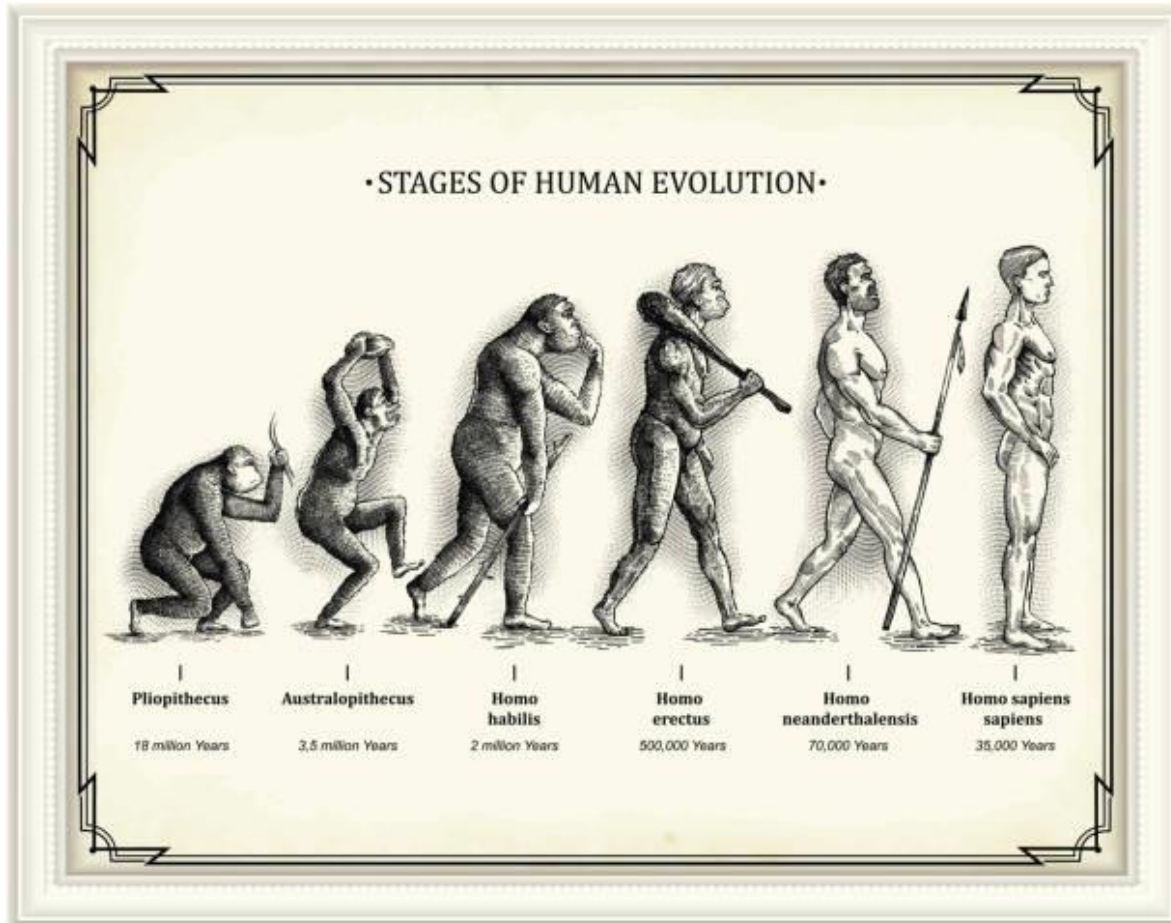
Assistant professor



HISTORY OF THE THEORY



1. CLASSICAL PALEOANTHROPOLOGY

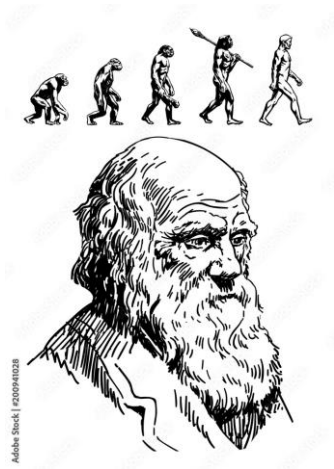


Timeline of human evolution

1. CLASSICAL PALEOANTHROPOLOGY

Timeline of
human evolution

Charles Darwin

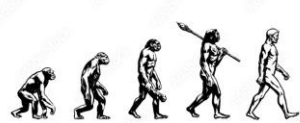


Studied behavior of African apes at London Zoo
→ relation btw humans & African apes

1. CLASSICAL PALEOANTHROPOLOGY

Timeline of
human evolution

Charles Darwin



Studied behavior of African apes at London Zoo

→ relation btw humans & African apes

Thomas Huxley



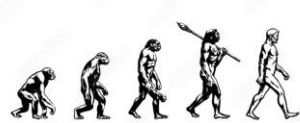
Supported Darwin &

suggested a close evolutionary relationship with
African apes

1. CLASSICAL PALEOANTHROPOLOGY

Timeline of
human evolution

Charles Darwin



Studied behavior of African apes at London Zoo

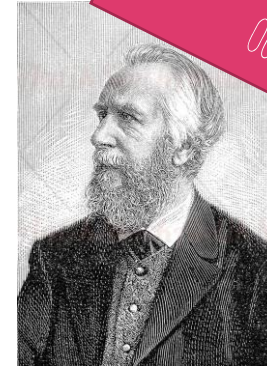
→ relation btw humans & African apes

Thomas Huxley



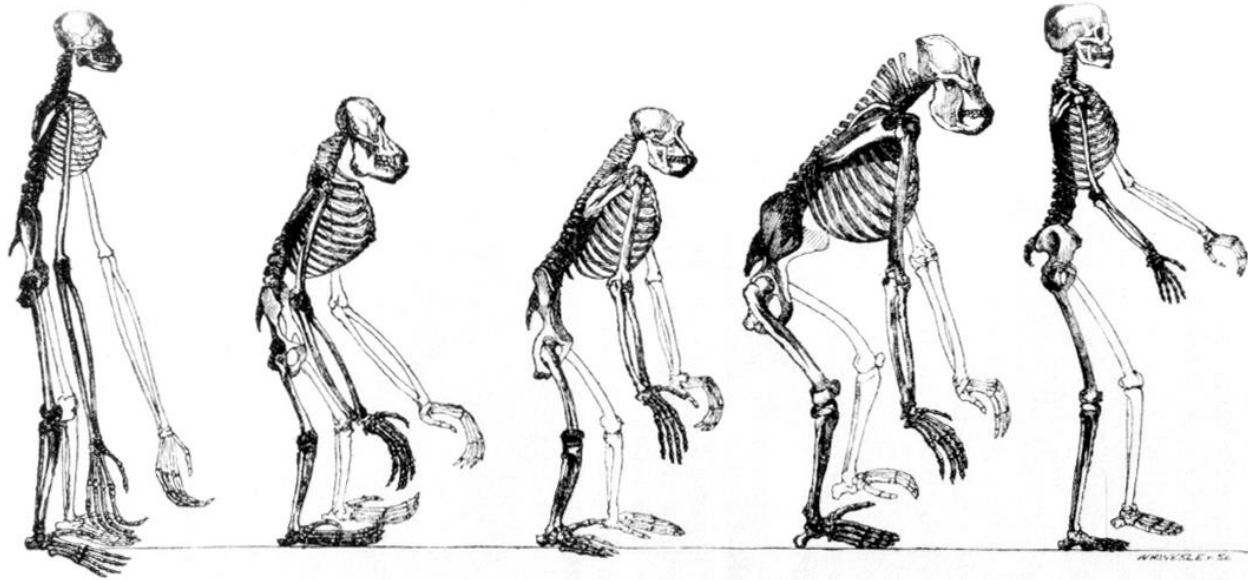
Supported Darwin &
suggested a close evolutionary relationship with
African apes

Ernst Haeckel



Out of Asia??

Rejected Darwin's African hypothesis &
suggested more closely related to the primates
of South-east Asia



GIBBON.

ORANG.

Skeletons of the
CHIMPANZEE.

GORILLA.

MAN.

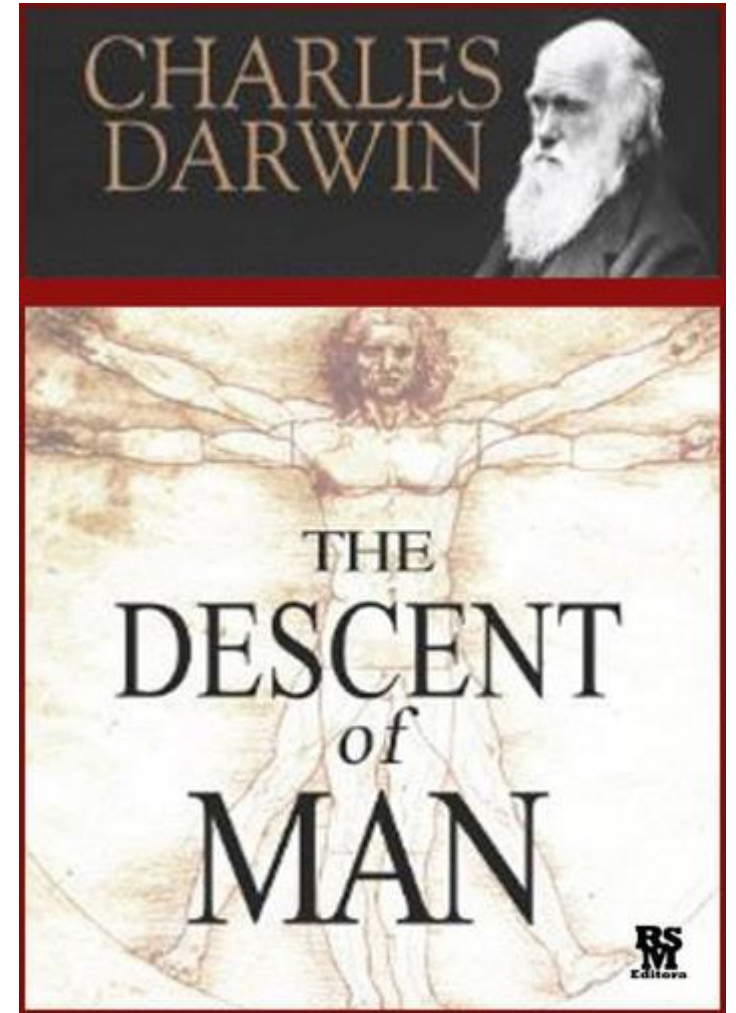
*Photographically reduced from Diagrams of the natural size (except that of the Gibbon, which was twice as large as nature),
drawn by Mr. Waterhouse Hawkins from specimens in the Museum of the Royal College of Surgeons.*

THE FRONTISPIECE TO HUXLEY'S
EVIDENCE AS TO MAN'S PLACE
IN NATURE (1863):

The image compares the skeleton
of a human to other apes

CHARLES DARWIN, DESCENT OF MAN

In each great region of the world the living mammals are closely related to the extinct species of the same region. It is, therefore, probable that Africa was formerly inhabited by extinct apes closely allied to the gorilla and chimpanzee; and as these two species are now man's nearest allies, it is somewhat more probable that our early progenitors lived on the African continent than elsewhere. But it is useless to speculate on this subject, for an ape nearly as large as a man, namely the Dryopithecus of Lartet, which was closely allied to the anthropomorphous Hylobates, existed in Europe during the Upper Miocene period; and since so remote a period the earth has certainly undergone many great revolutions, and there has been ample time for migration on the largest scale.



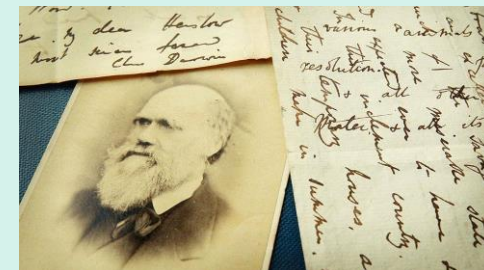
Hominin Fossils



DARWIN'S SPECULATION

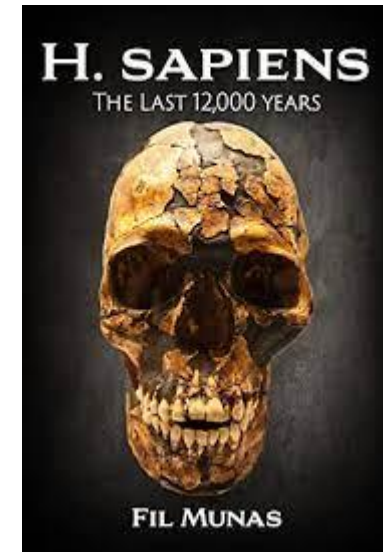
1871; hardly any human fossils of ancient hominins available

50 years later; Darwin's speculation was supported when anthropologists began finding fossils of ancient small-brained hominins in several areas of Africa

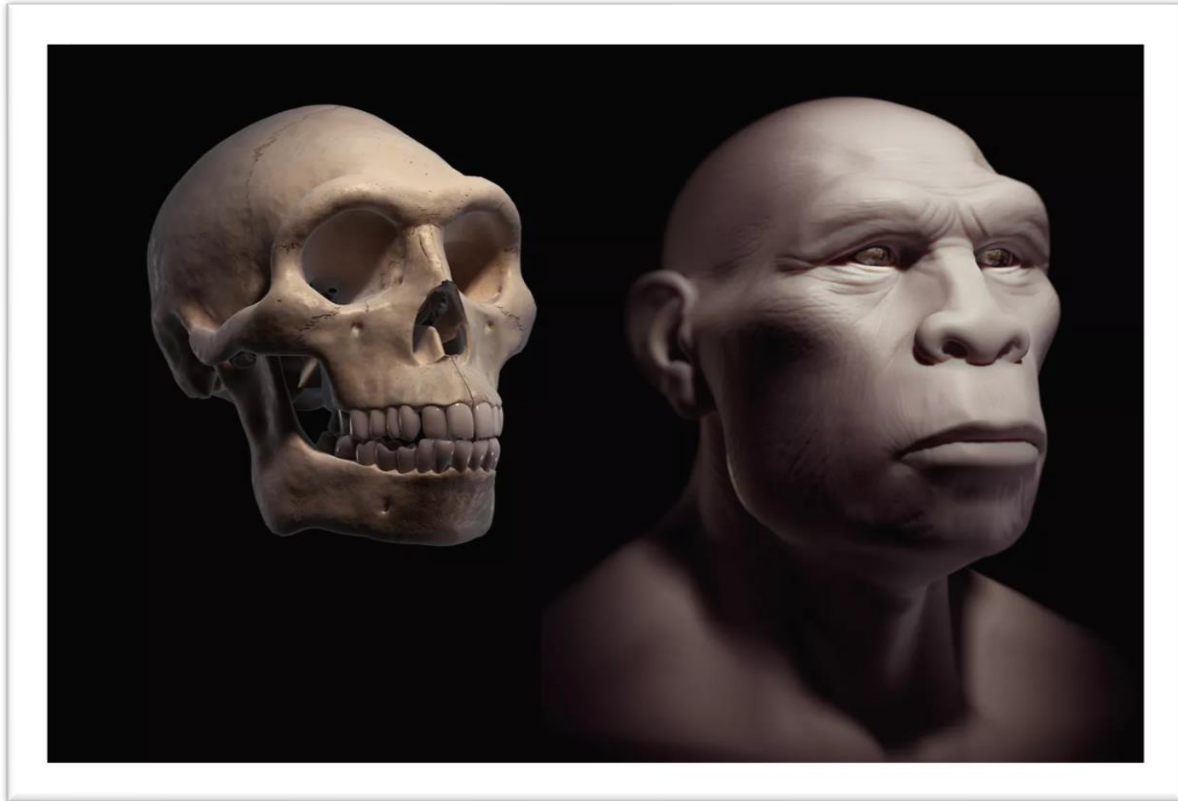


HYPOTHESIS OF RECENT AFRICAN ORIGIN

- Developed in the 20th cent
- The "Recent African origin" of modern humans means "single origin" (monogenism)
- Has been used in various contexts as an **antonym** to polygenism
- The debate in anthropology had swung in favour of monogenism by the mid-20th cent
- Isolated proponents of polygenism held forth in the mid-20th cent
 - Carleton Coon (1962) → *H. sapiens* arose five times from *H. erectus* in five places



2. MULTIREGIONAL ORIGIN HYPOTHESIS



Multiregional origin
of modern humans

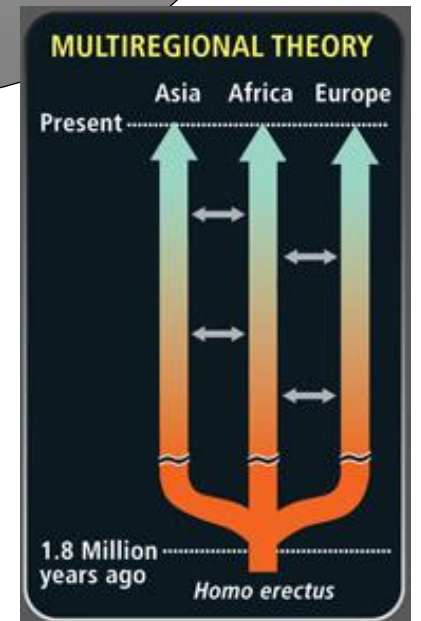
2. MULTIREGIONAL ORIGIN HYPOTHESIS

Multiregional
origin of modern
humans

- The historical alternative to the recent origin model
- Proposed by Milford Wolpoff in the 1980s



the derivation of anatomically modern human populations from *H. erectus* at the beginning of the Pleistocene 1.8 million years BP has taken place within a continuous world population.



2. MULTIREGIONAL ORIGIN HYPOTHESIS

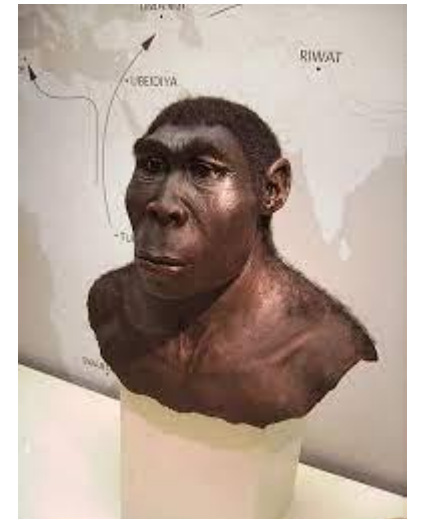
Multiregional
origin of modern
humans

- The historical alternative to the recent origin model
- Proposed by Milford Wolpoff in the 1980s

the derivation of anatomically modern human populations from *H. erectus* at the beginning of the Pleistocene 1.8 million years BP has taken place within a continuous world population.

The hypothesis

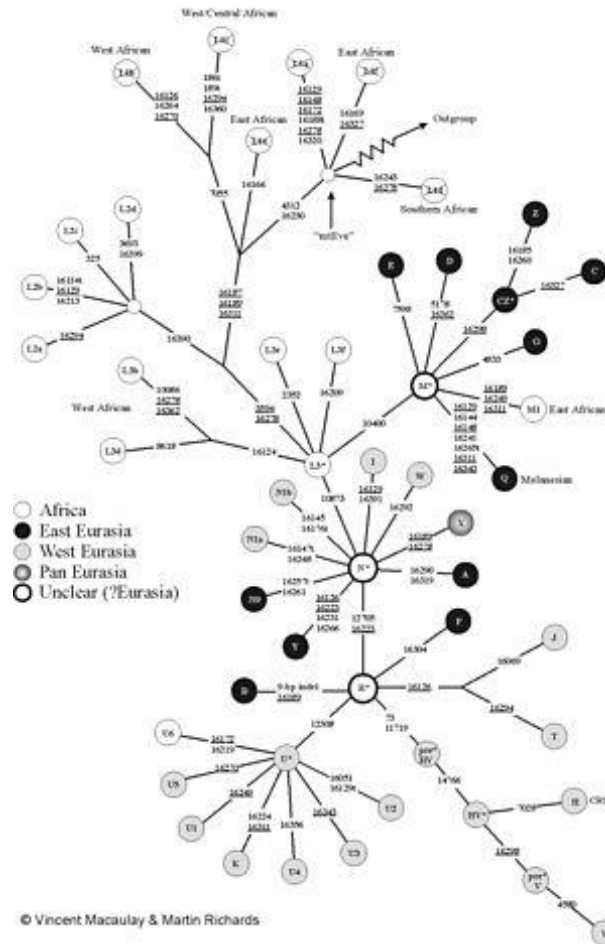
- Rejects the assumption of an infertility barrier between ancient Eurasian & African populations of Homo
- Controversially debated during the late 1980s & 1990s



2. MULTIREGIONAL ORIGIN HYPOTHESIS

Multiregional
origin of modern
humans

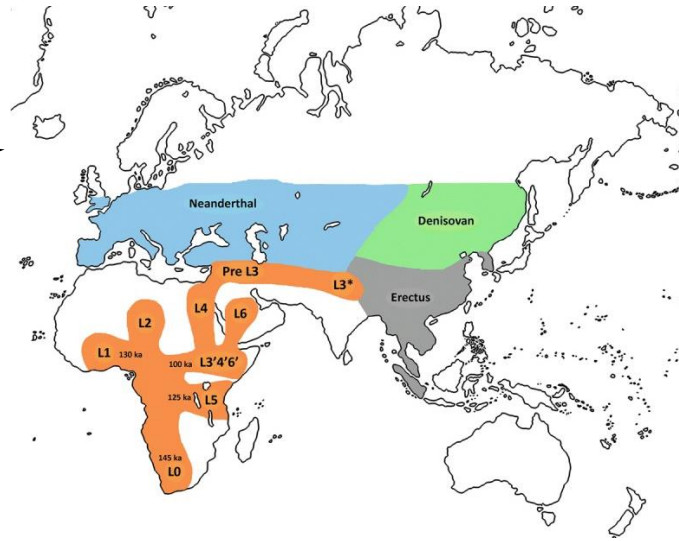
- The now-current terminology of "recent-origin" & "Out of Africa" became current in the context of this debate in the 1990s
- Originally seen as an antithetical alternative to the recent origin model, in its original "strong" form is obsolete, while its various modified weaker variants have become variants of a view of "recent origin" combined with archaic admixture
- Stringer (2014) distinguishes the original or "classic" Multiregional model as having existed from 1984 (its formulation) until 2003, to a "weak" post-2003 variant that has "shifted close to that of the Assimilation Model"



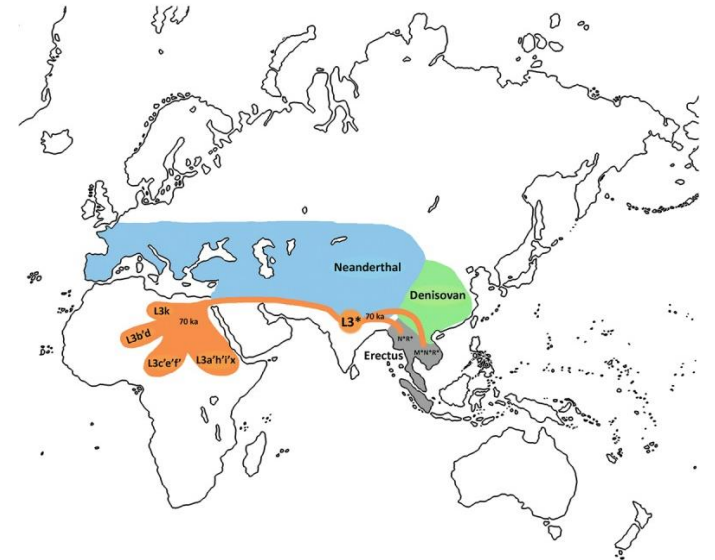
THE POSSIBILITY OF AN ORIGIN OF L3 IN ASIA

proposed by *Cabrera et al. 2018*

Exit of the L3 precursor to Eurasia



Return to Africa & expansion to Asia of basal L3 lineages with subsequent differentiation in both continents



3. MITOCHONDRIAL ANALYSES



Mitochondrial Eve

3. MITOCHONDRIAL ANALYSES

1980s: Allan Wilson, Rebecca L. Cann & Mark Stoneking worked on genetic dating of the matrilineal most recent common ancestor of modern human populations

Discovered that human mtDNA is genetically much less diverse than chimpanzee mtDNA →

- modern humans diverged recently from a single population
- older human species (Neanderthals & Homo erectus) become extinct



mtDNA passed from mother to child. This DNA material mutates quickly, making it easy to plot changes over relatively short times.

Mitochondrial Eve

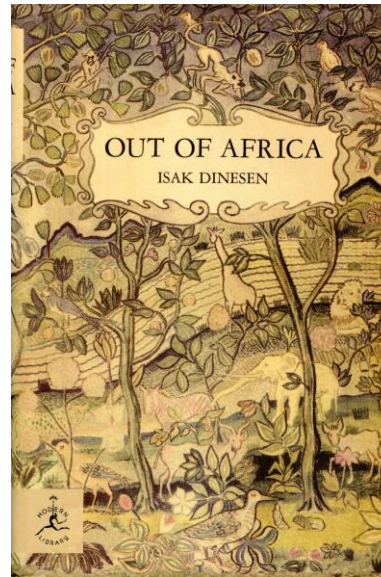
3. MITOCHONDRIAL ANALYSES

1990s: the dating of mitochondrial and Y-chromosomal haplogroups became possible (+confidence)

1999: estimates ranged around

150,000 years for the mt-MRCA

60,000 – 70,000 years for the migration out of Africa



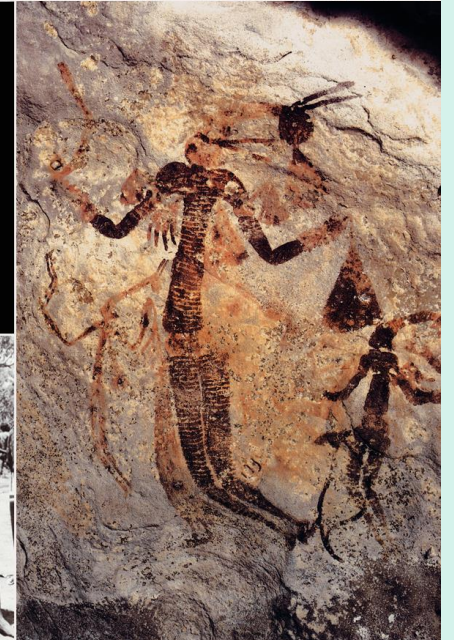
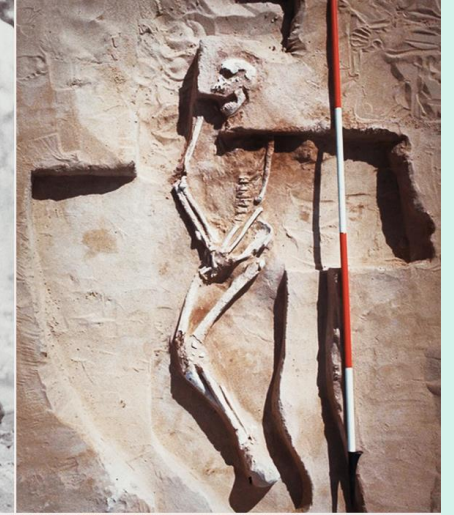
mtDNA/ mt-MRCA/ mt-Eve
→
passed from mother to child
This DNA material mutates quickly
Making it easy to plot changes over relatively short times.

Mitochondrial Eve

MUNGO MAN 3 *LM3*

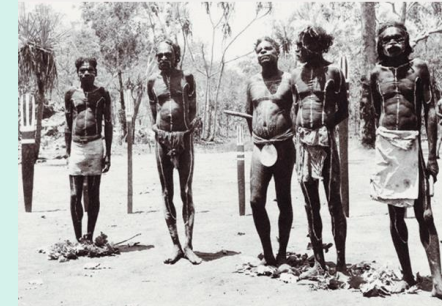
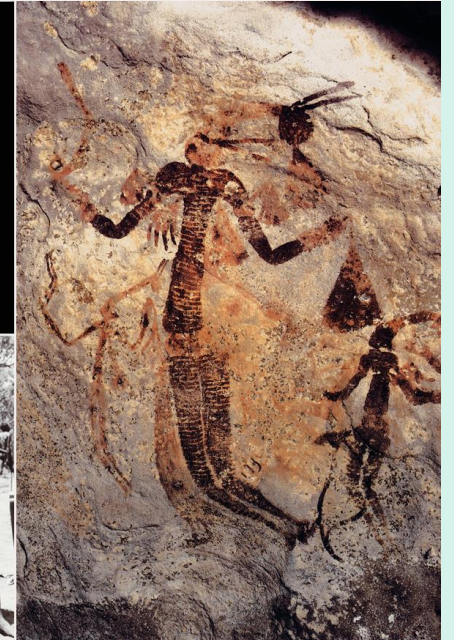
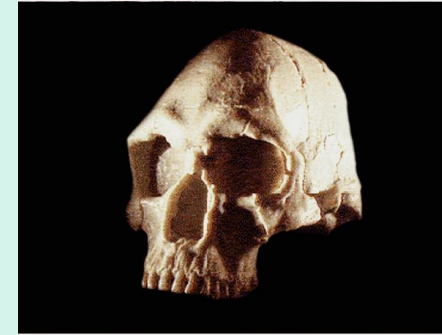
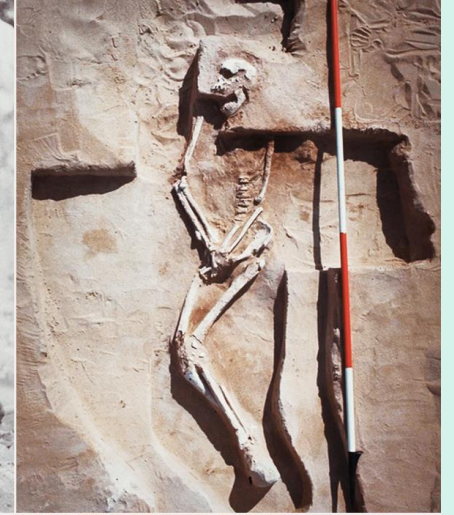
- 2000 - 2003: controversy about mtDNA (LM3) and its possible bearing on the multiregional hypothesis
- LM3: has more than the expected number of sequence differences when compared to modern human DNA (CRS)
- Comparison (mtDNA LM3 - ancient & modern aborigines →

LM3 fell outside the range of their genetic variation seen in Aboriginal Australians = support the **multiregional origin hypothesis**

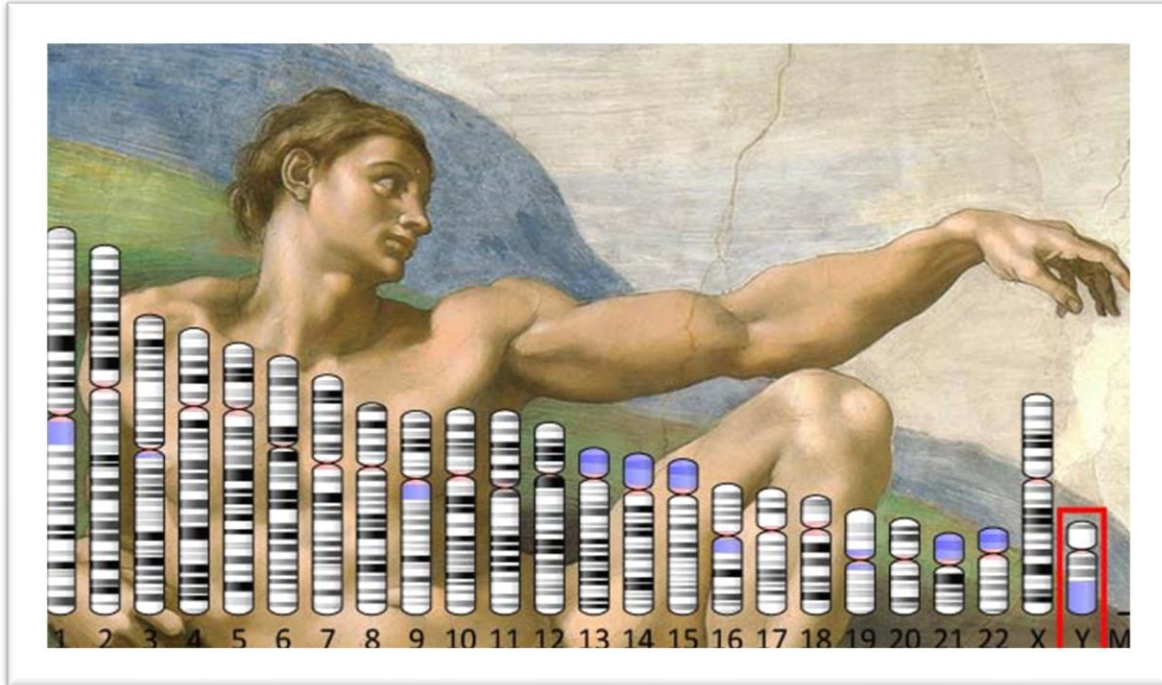


MUNGO MAN 3 *LM3*

- 2000 - 2003: controversy about mtDNA (LM3) and its possible bearing on the multiregional hypothesis
- LM3: has more than the expected number of sequence differences when compared to modern human DNA (CRS)
- Comparison (mtDNA LM3 - ancient & modern aborigines → LM3 fell outside the range of their genetic variation seen in Aboriginal Australians = support the multiregional origin hypothesis
- 2016: reanalysis on LM3 & other ancient specimens from the area → LM3 akin to modern Aboriginal Australian sequences, inconsistent with the results of the earlier study



4. Y-CHROMOSOME ANALYSES



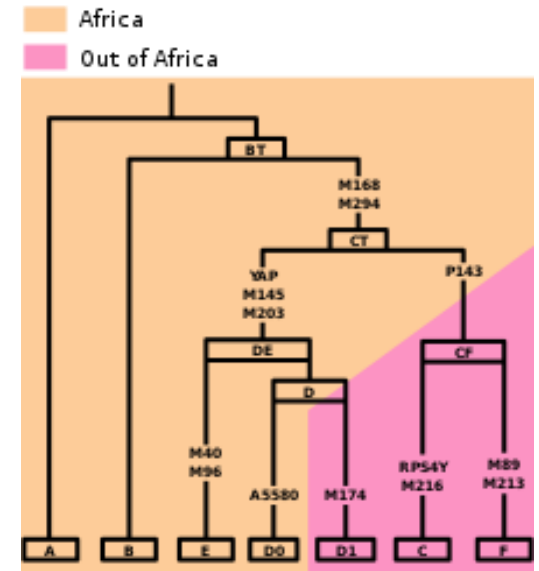
Y-chromosomal
Adam

4. Y-CHROMOSOME ANALYSES

Y-chromosomal Adam

Y-chromosomal Adam/Y-MRCA

- As current estimates on the male most recent common ancestor converge with estimates for the age of anatomically modern humans and well predate the Out of Africa migration, **geographical origin hypotheses** continue to be limited to the African continent
- The most basal lineages have been detected in **West, Northwest and Central Africa** → plausibility for the Y-MRCA living in the general region of "Central-Northwest Africa"
- Another study → plausible placement in **"the north-western quadrant of the African continent"** for the emergence of the A1b haplogroup

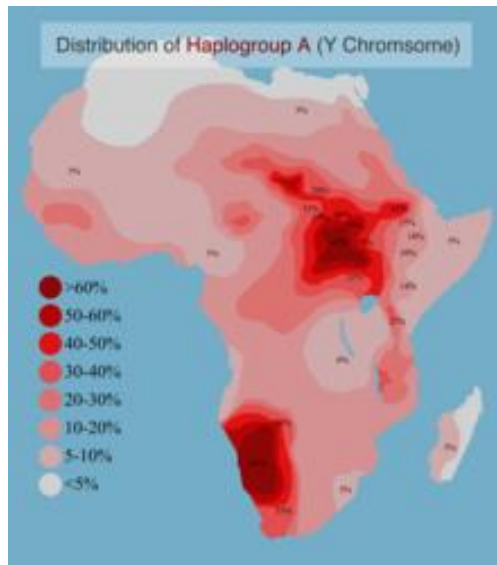


4. Y-CHROMOSOME ANALYSES

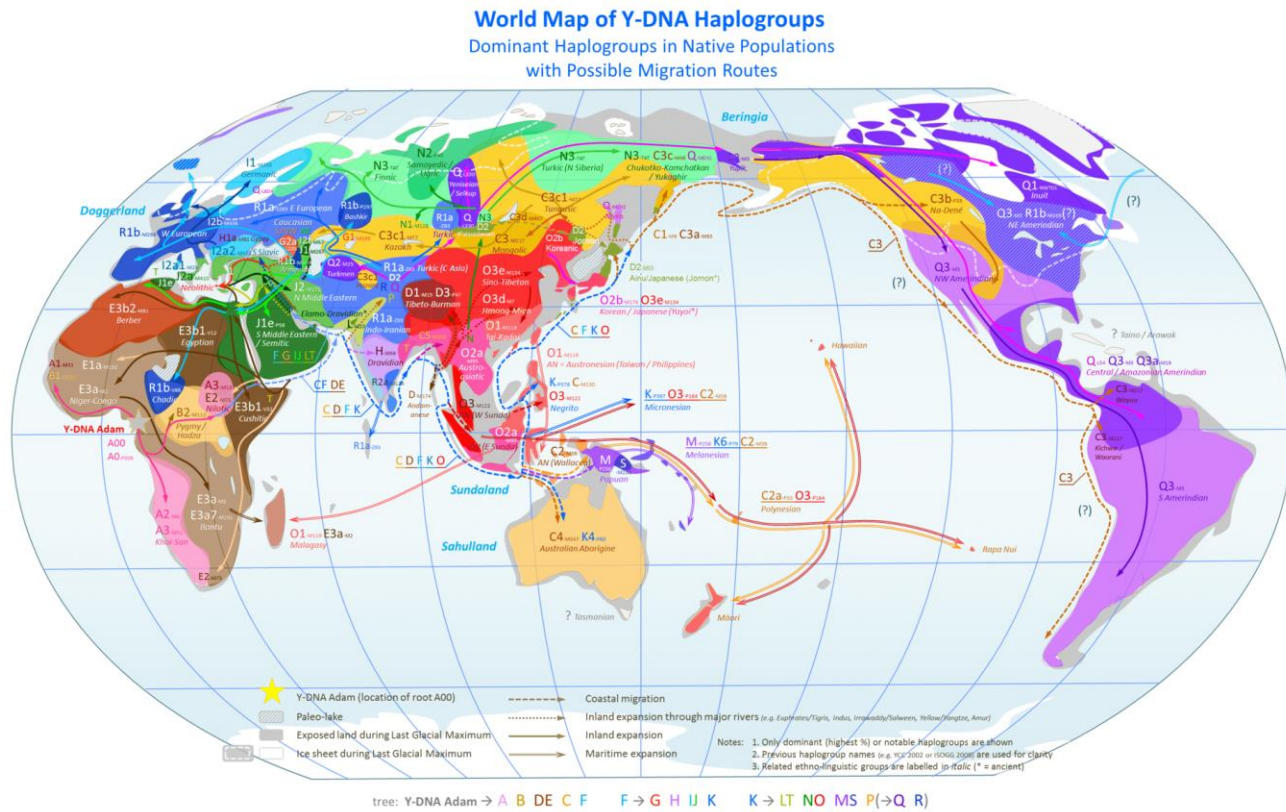
Y-chromosomal
Adam

Y-chromosomal Adam/Y-MRCA

- 2013: haplogroup A00 found among the Mbo people of western present-day Cameroon is also compatible with this picture

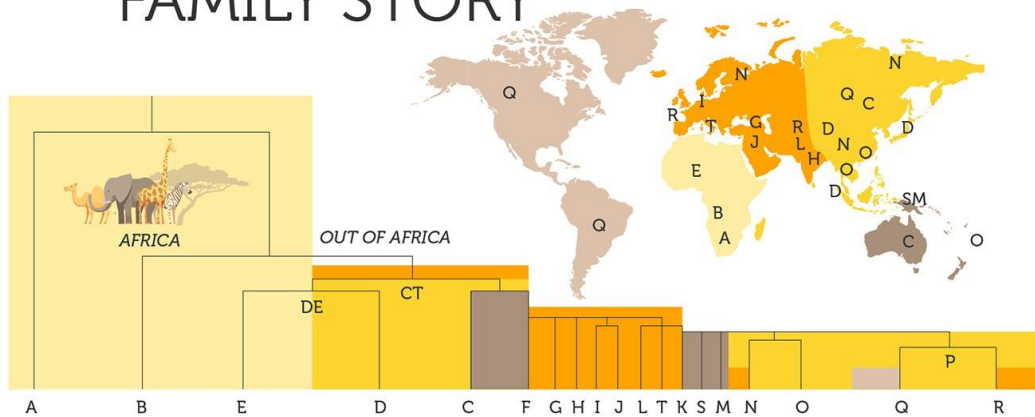


MAP OF Y-CHROMOSOME HAPLOGROUPS



-Dominant haplogroups in pre-colonial populations with proposed migrations routes

THE Y CHROMOSOME TREE & YOUR PATERNAL FAMILY STORY

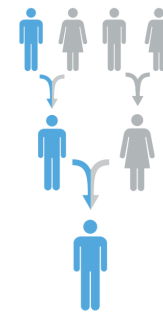


Since 2011: The revision of Y-chromosomal phylogeny has affected estimates on:

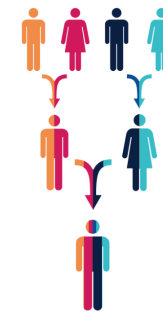
- The likely **geographical origin of Y-MRCA**
- The time depth

By the same reasoning, future discovery of presently-unknown archaic haplogroups in living people would again lead to such revisions.

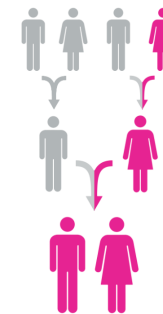
Y-Chromosome DNA is inherited from your male lineage



Autosomal DNA is inherited from all of your ancestors.



Mitochondrial DNA is inherited from your female lineage





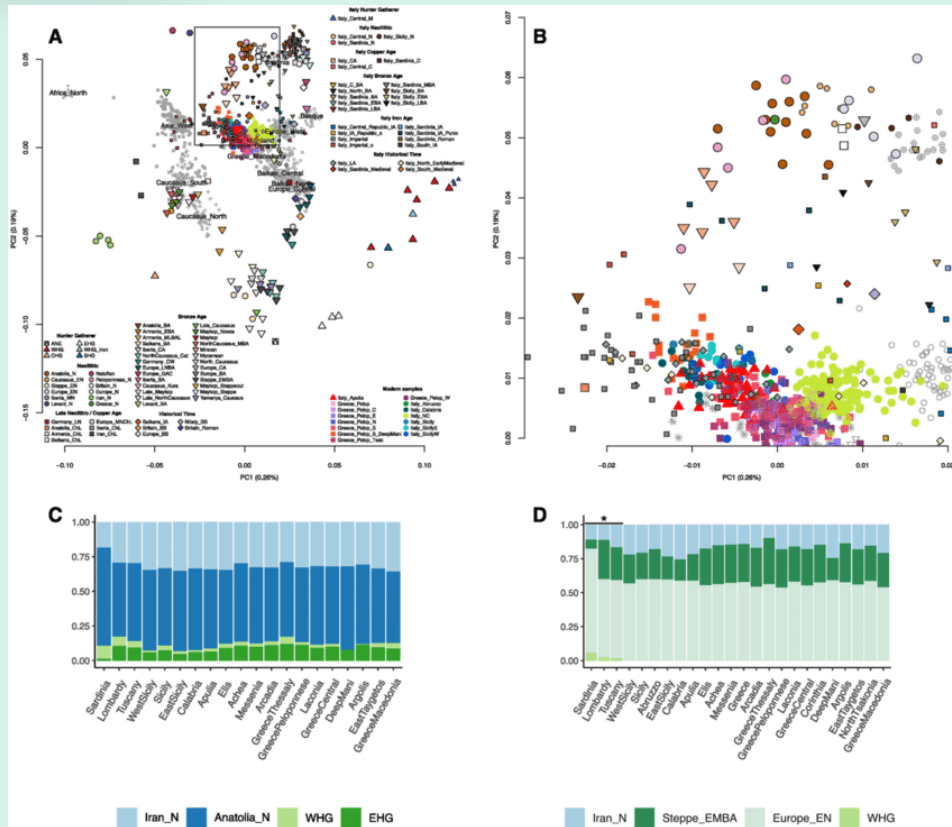
The possible presence of between 1% & 4% Neanderthal-derived DNA in Eurasian genomes → that the (unlikely) event of a discovery of a single living Eurasian male exhibiting a Neanderthal patrilineal line would immediately push back T-MRCA to at least twice its current estimate

Mendez et al. A 2016

→ The extinction of Neanderthal patrilineages lineage inferred from the Neanderthal sequence is outside of the range of contemporary human genetic variation

Questions of geographical origin would become part of the debate on

Neanderthal evolution from *Homo erectus*



ARTICLE

The Divergence of Neanderthal and Modern Human Y Chromosomes

Fernando L. Mendez,^{1,*} G. David Poznik,^{1,2} Sergi Castellano,³ and Carlos D. Bustamante^{1,4,*}

Sequencing the genomes of extinct hominids has reshaped our understanding of modern human origins. Here, we analyze exome-captured Y-chromosome DNA from a Neanderthal individual from El Sidrón, Spain. We investigate its divergence from chimpanzee and modern human sequences and find strong support for a model that places the Neanderthal lineage to modern human Y chromosomes—including A00, the highly divergent basal haplogroup. We estimate that the time to common ancestor (TMRCA) of Neanderthal and modern human Y chromosomes is ~588 thousand years ago (kya) (95% interval [CI]: 447–806 kya). This is ~2.1 (95% CI: 1.7–2.9) times longer than the TMRCA of A00 and other extant Y-chromosome lineages. This estimate suggests that the Y-chromosome divergence mirrors the population divergence and modern human ancestors, and it refutes alternative scenarios of a relatively recent or super-archaic origin of Y chromosomes. The fact that the Neanderthal Y we describe has never been observed in modern humans suggests it is most likely extinct. We identify protein-coding differences between Neanderthal and modern human Y chromosomes potentially damaging changes to *PCDH11Y*, *TMSB4Y*, *USP9Y*, and *KDMSD*. Three of these changes are missense mutations that produce male-specific minor histocompatibility (H-Y) antigens. Antigens derived from *KDMSD*, for example, are the subject of a maternal immune response during gestation. It is possible that incompatibilities at one or more of these genes play a role in reproductive isolation of the two groups.

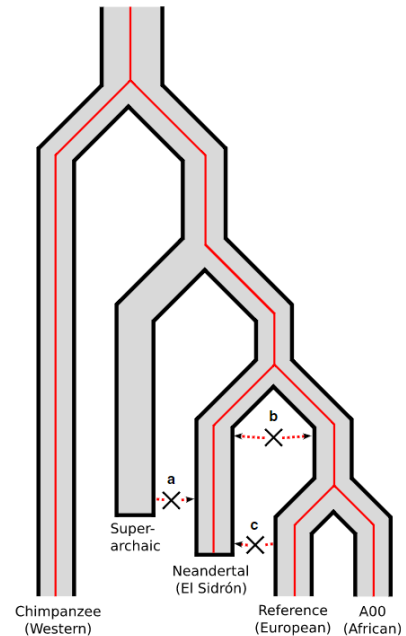


Figure 3. Relationship of Neanderthal Y Chromosome to Those of Modern Humans



CHOSEN PAPER

[HTTPS://DOI.ORG/10.1016/J.AJHG.2016.02.023](https://doi.org/10.1016/j.ajhg.2016.02.023)

The great human expansion

Brenna M. Henn^a, L. L. Cavalli-Sforza^{a,1}, and Marcus W. Feldman^{b,2}

^aDepartment of Genetics, Stanford University School of Medicine, Stanford, CA 94305; and ^bDepartment of Biology, Stanford University, Stanford, CA 94305-5020

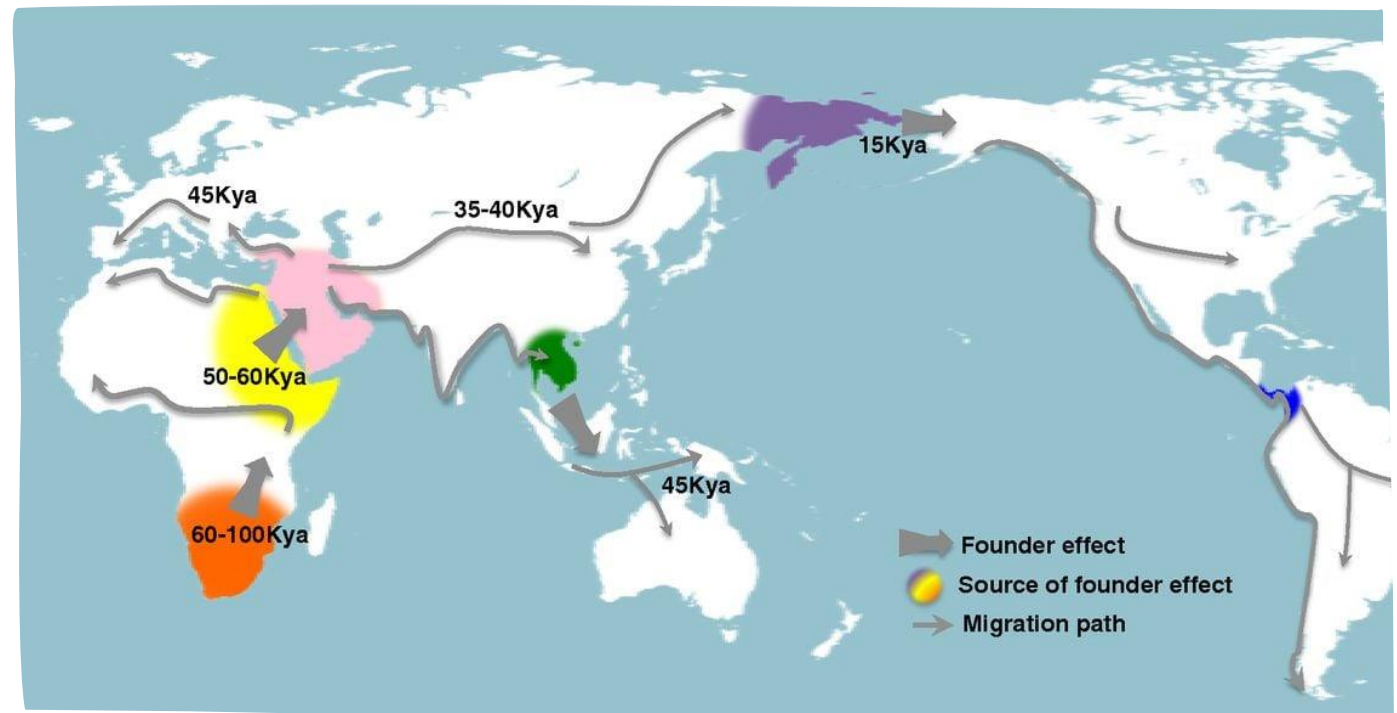
Edited by C. Owen Lovejoy, Kent State University, Kent, OH, and approved September 25, 2012 (received for review July 19, 2012)

Genetic and paleoanthropological evidence is in accord that today's human population is the result of a great demic (demographic and geographic) expansion that began approximately 45,000 to 60,000 y ago in Africa and rapidly resulted in human occupation of almost all of the Earth's habitable regions. Genomic data from contemporary humans suggest that this expansion was accompanied by a continuous loss of genetic diversity, a result of what is called the "serial founder effect." In addition to genomic data, the serial founder effect model is now supported by the genetics of human parasites, morphology, and linguistics. This particular population history gave rise to the two defining features of genetic variation in humans: genomes from the substructured populations of Africa retain an exceptional number of unique variants, and there is a dramatic reduction in genetic diversity within populations living outside of Africa. These two patterns are relevant for medical genetic studies mapping genotypes to phenotypes and for inferring the power of natural selection in human history. It should be appreciated that the initial expansion and subsequent serial founder effect were determined by demographic and sociocultural factors associated with hunter-gatherer populations. How do we reconcile this major demic expansion with the population stability that followed for thousands years until the inventions of agriculture? We review advances in understanding the genetic diversity within Africa and the great human expansion out of Africa and offer hypotheses that can help to establish a more synthetic view of modern human evolution.

THE GREAT EXPANSION

1. Human Origins in Africa
2. Independent Evidence of Out of Africa Model
3. Concordance Between Genes and Language
4. Demographic Models of OOA Expansion
5. Demography of Foragers
6. Population Density
7. Culturally Mediated Population Stability

Genetic data indicate that, approximately 45 to 60 kya, a very rapid population expansion occurred outside of Africa & spread in all directions across the Eurasian continents, eventually populating the entire world.



THE GREAT EXPANSION

1. Human Origins in Africa
2. Independent Evidence of Out of Africa Model
3. Concordance Between Genes and Language
4. Demographic Models of OOA Expansion
5. Demography of Foragers
6. Population Density
7. Culturally Mediated Population Stability

The precise location of the exit from Africa, & the relative timing of the southern coastal migration vs. the expansion into northern Eurasia, are still under intense debate

Many parameters of the **Great Expansion** are now well understood, including the:

1. general timing of the exit
2. magnitude of the associated bottleneck
3. mode of subsequent expansion

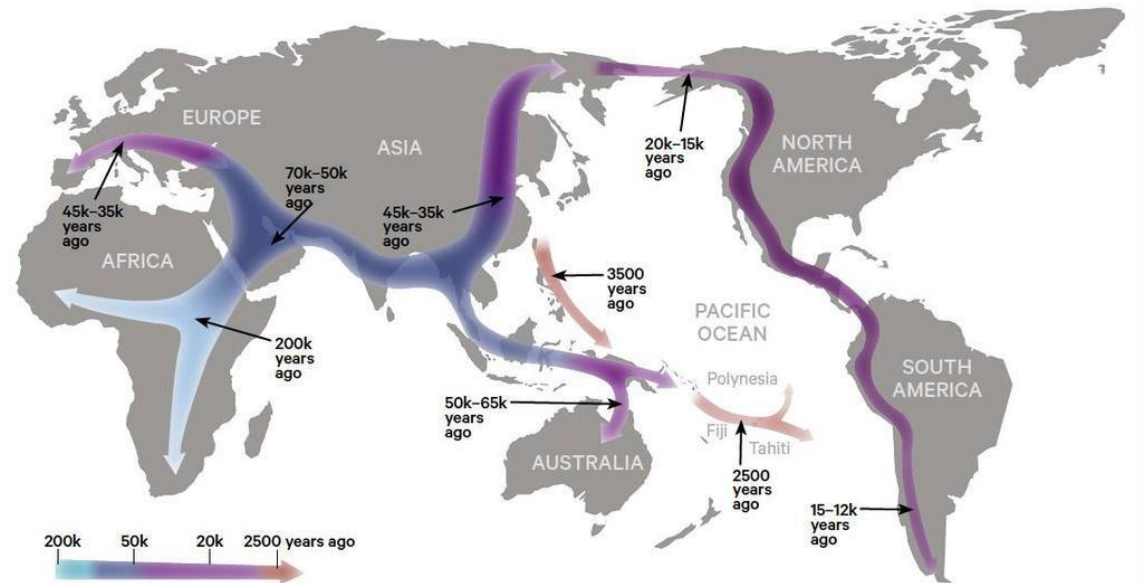
Human genetic, linguistic, and phenotypic data strongly support a serial founder effect model for the Great Expansion OOA approximately 45 to 60 kya.

Significant gaps in our knowledge of human population history still exist

Additional sampling & sequencing of African populations remains a priority

Refined measures of environmental variability & its temporal/spatial changes across Africa would help clarify when human dispersals were likely.

Review the history of the Out of Africa expansion, with an emphasis on recent genomic data



CONCLUSIONS



Computational models incorporating multiple episodes of growth and capable of inferring migration/divergence among more than three demes will need to be developed for this next wave of genomic data.



A synthesis among genetic, demographic, and anthropological models is important for a full understanding of many parameters of human prehistory, as illustrated here with population stability.

CONCLUSIONS



For example, studies of natural selection could pair selection statistics with current phenotypic data, and model the time of selective pressure based on archeological information.



Many genetic projects still ignore the rich background of archaeological and ethnographic data when testing genetic models, or tend to highlight only those findings that are consistent with the genetic conclusions. Information from all of the sources mentioned here will be necessary to constrain the complex hypotheses surrounding modern human origins.

PART 2:

THE DEEP POPULATION HISTORY IN AFRICA



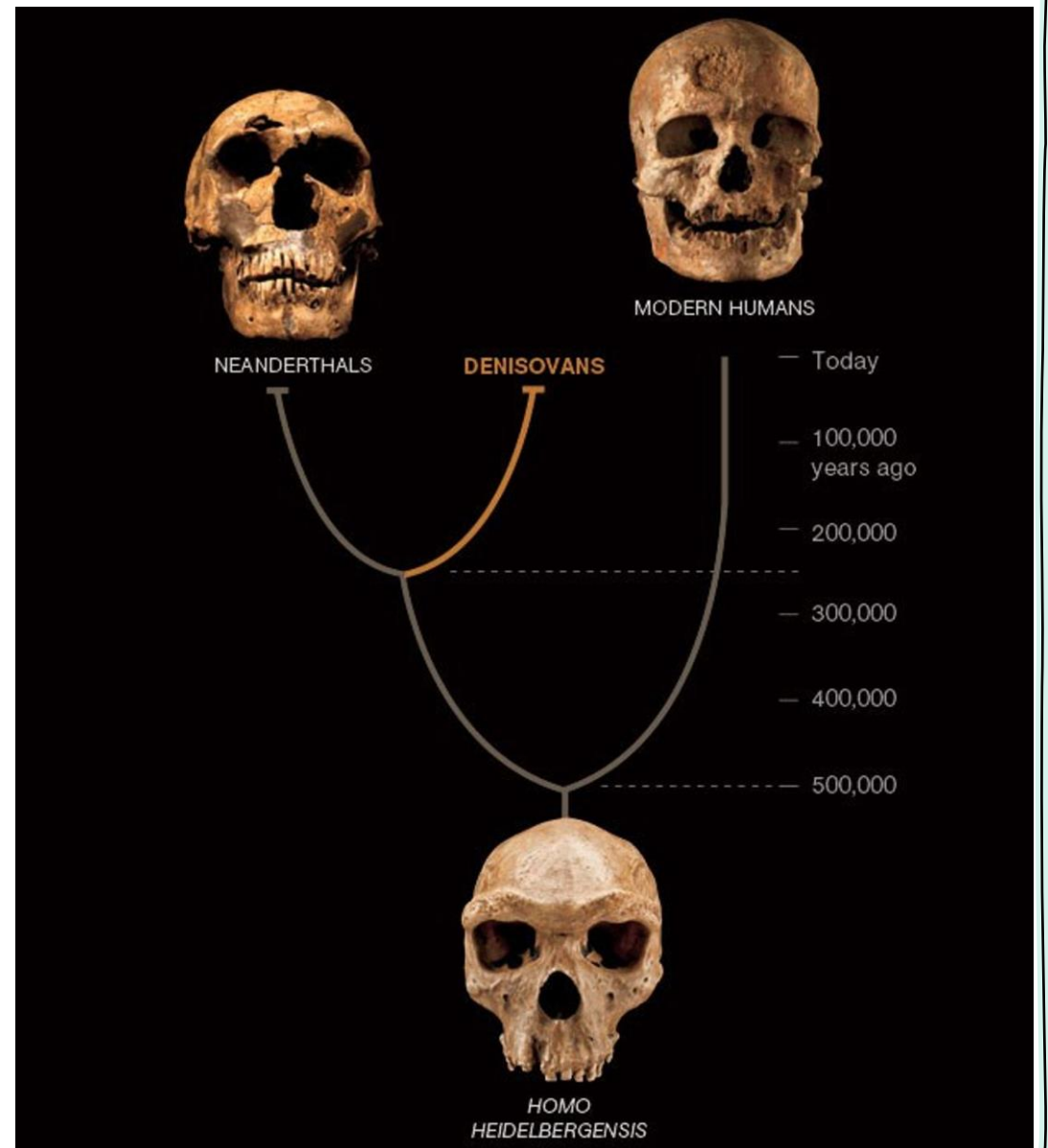
THE DEEP POPULATION HISTORY IN AFRICA


1. Hunter-gatherer Population Structure
2. Models of Human Evolution and Population Divergence Time Estimates
3. Introgression from Extinct Lineages

1. HUNTER-
GATHERER
POPULATION
STRUCTURE



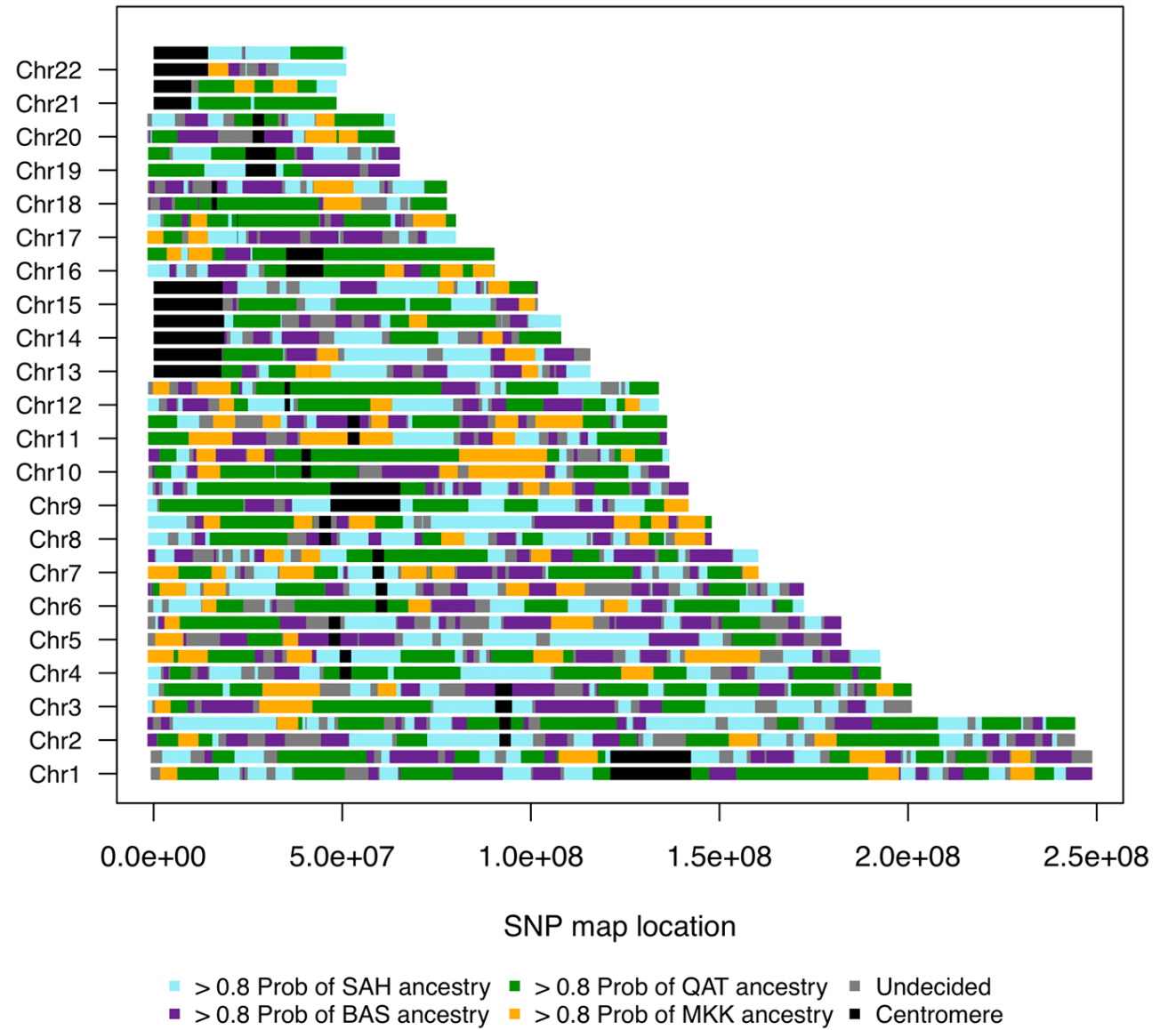
- Africa has the highest genetic diversity among all humans, with the exception of Neanderthals and Denisovans
- African hunter-gatherer groups have the most genetic diversity and carry the most basal uniparental markers
- These populations were connected & potentially inhabited larger geographic areas, with indications of gene-flow between Khoe-San and other African hunter-gatherer populations



A close-up photograph of a person's hands holding a large, cylindrical bag made of woven fabric and covered in intricate beadwork. The beads are arranged in horizontal bands of various colors, including blue, green, yellow, orange, and red. The person is wearing a patterned garment with geometric designs in yellow, red, and black. The background is a blurred green, suggesting an outdoor setting. A white speech bubble with a black border is overlaid on the left side of the image, containing text.

The contraction of once wide-spread, overlapping hunter-gatherer populations left a mark in their genomes, and many hunter-gatherer populations do not display an effective population size increase in the Holocene


Populations from North Africa show mainly Eurasian ancestry, with a modest ancestry component assigned to sub-Saharan Africa



Recent ancient DNA results from Moroccan fossil samples provide insights into hunter-gatherers from North Africa.



These individuals show decreasing sub-Saharan ancestry over time, likely due to isolation in the Maghreb since the Upper Paleolithic

A group of prehistoric hominids, including adults and a child, holding tools and looking towards the camera. The scene is set in a dark, cave-like environment. The hominids have dark skin and are dressed in simple, primitive clothing. Some are holding tools, possibly spears or clubs. The overall atmosphere is one of a primitive, tribal society.

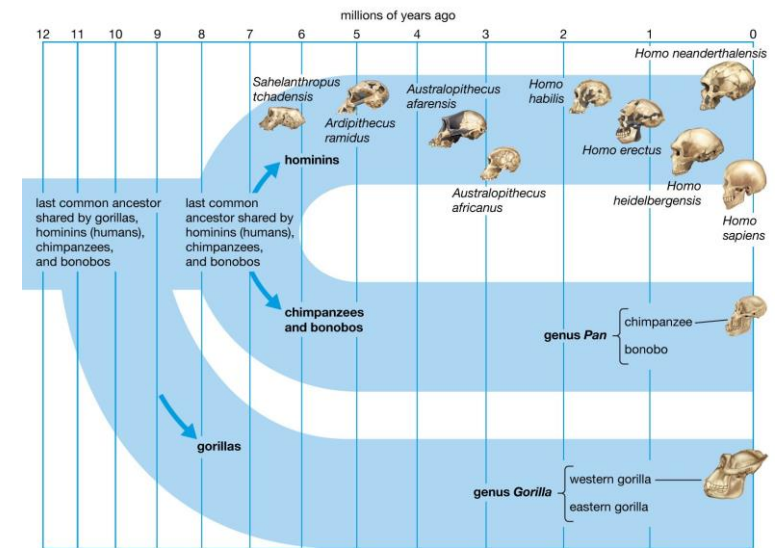
2. MODELS OF
HUMAN EVOLUTION
& POPULATION
DIVERGENCE TIME
ESTIMATES



It is common to model the evolutionary history of humans as a bifurcating tree and to estimate divergence times between the branches

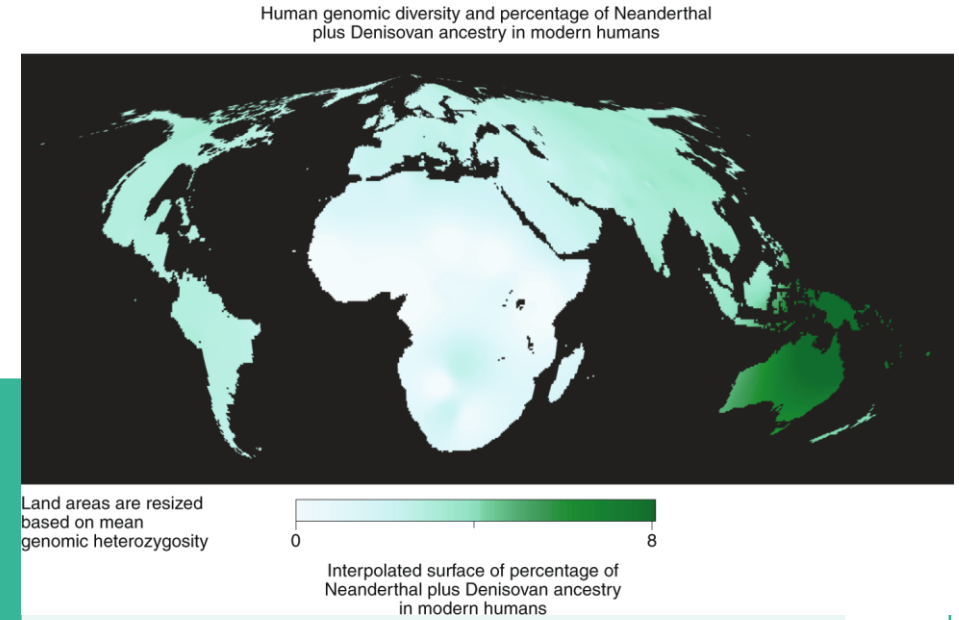


Trees are useful models to understand relations among groups and relative divergences, but miss some features such as gene-flow.



Estimates for specific events vary due to methods and assumptions of models, scaling parameters, and the set of individuals and populations used for comparisons

Alternatively

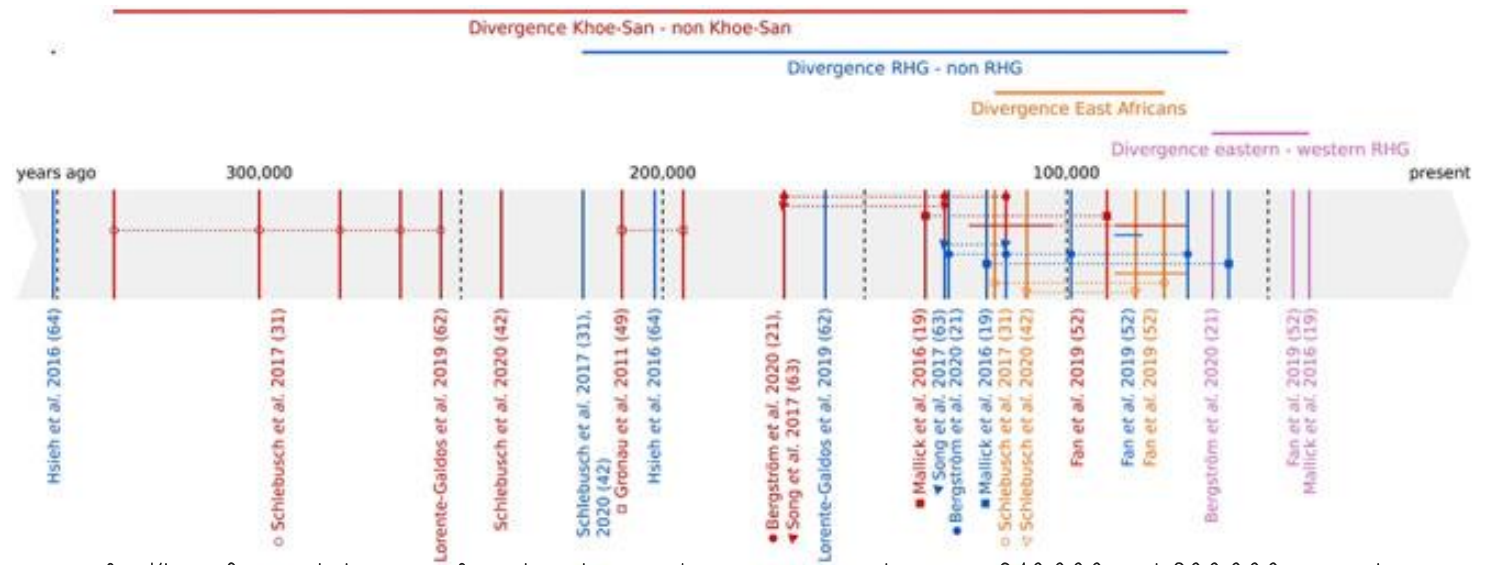
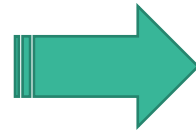


Human evolution can be represented with metapopulation models

an overview of the range of estimates of deep divergence events in human population history based on high-coverage autosomal genomes from a range of studies

Different methods:

1. genealogical concordance,
2. site-frequency
3. approximate Bayesian computation analyses



The estimates for Khoe-San and the rest of modern humans' ancestors vary between 340,000 and 200,000 ya, with younger estimates (160,000-90,000) based on MSMC cross-coalescence approach.

The next event assuming a simplified bifurcating tree is a divergence between rainforest hunter-gatherer ancestors and the ancestors of the rest of modern humans (minus the Khoe-San); the estimates vary from 350,000 to 70,000 ya.





Eastern African groups, including hunter-gatherers like the Hadza and Sandawe, point to divergences from all other African groups at ~140,000-70,000 ya.

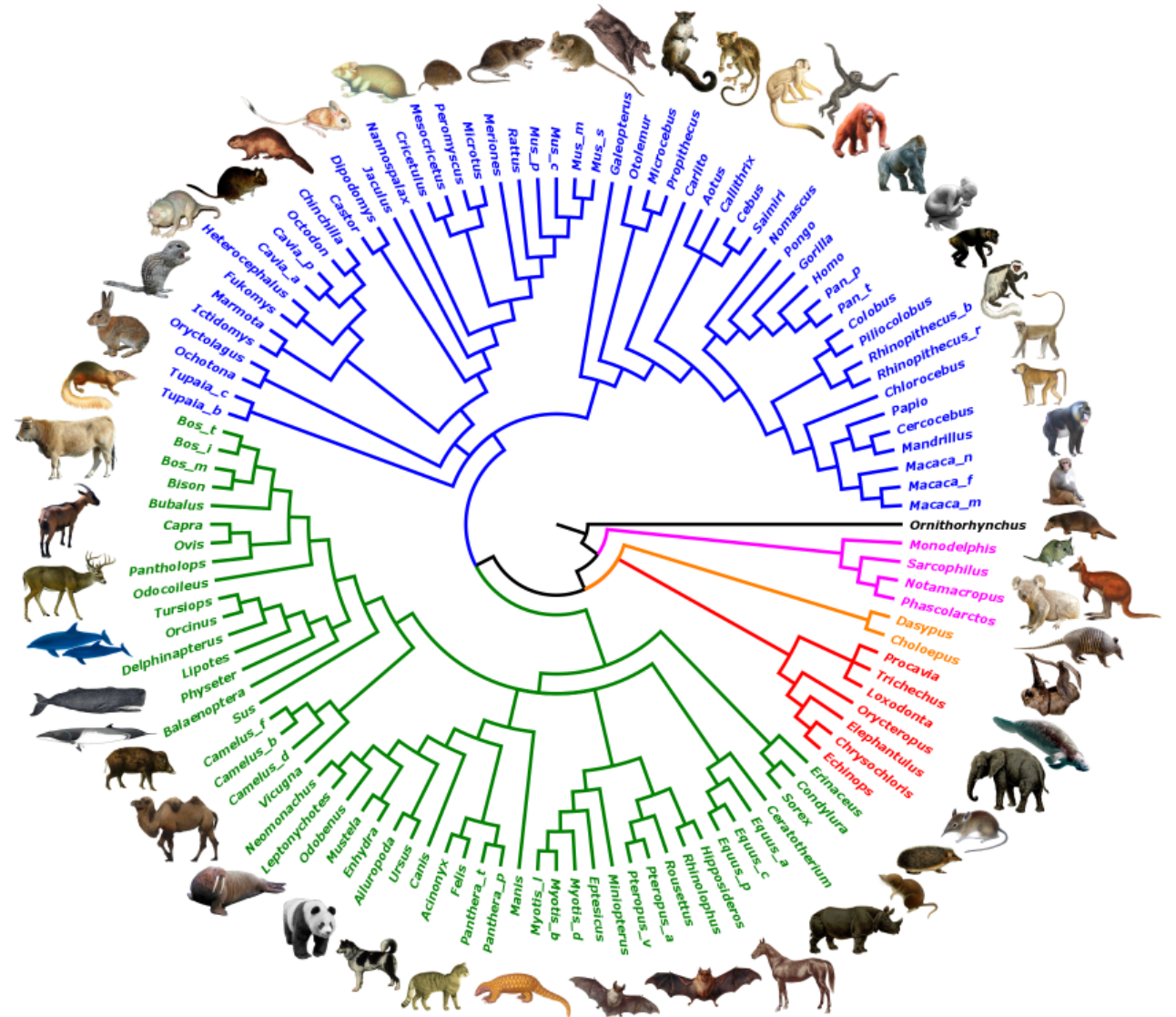


Most studies find a bifurcation as the deepest divergence among humans, but some studies point at alternatives, such as a very deep divergence involving RHGs or a trifurcation.

The estimates for the divergence between northern and southern San have a large range as well, from ~170,000 to ~30,000 ya, possibly due to an isolation-by-distance model.

The common origin of western and eastern RHGs has been demonstrated, and the divergence between them has been estimated to ~60,000–40,000 ya.

3. INTROGRESSION FROM EXTINCT LINEAGES



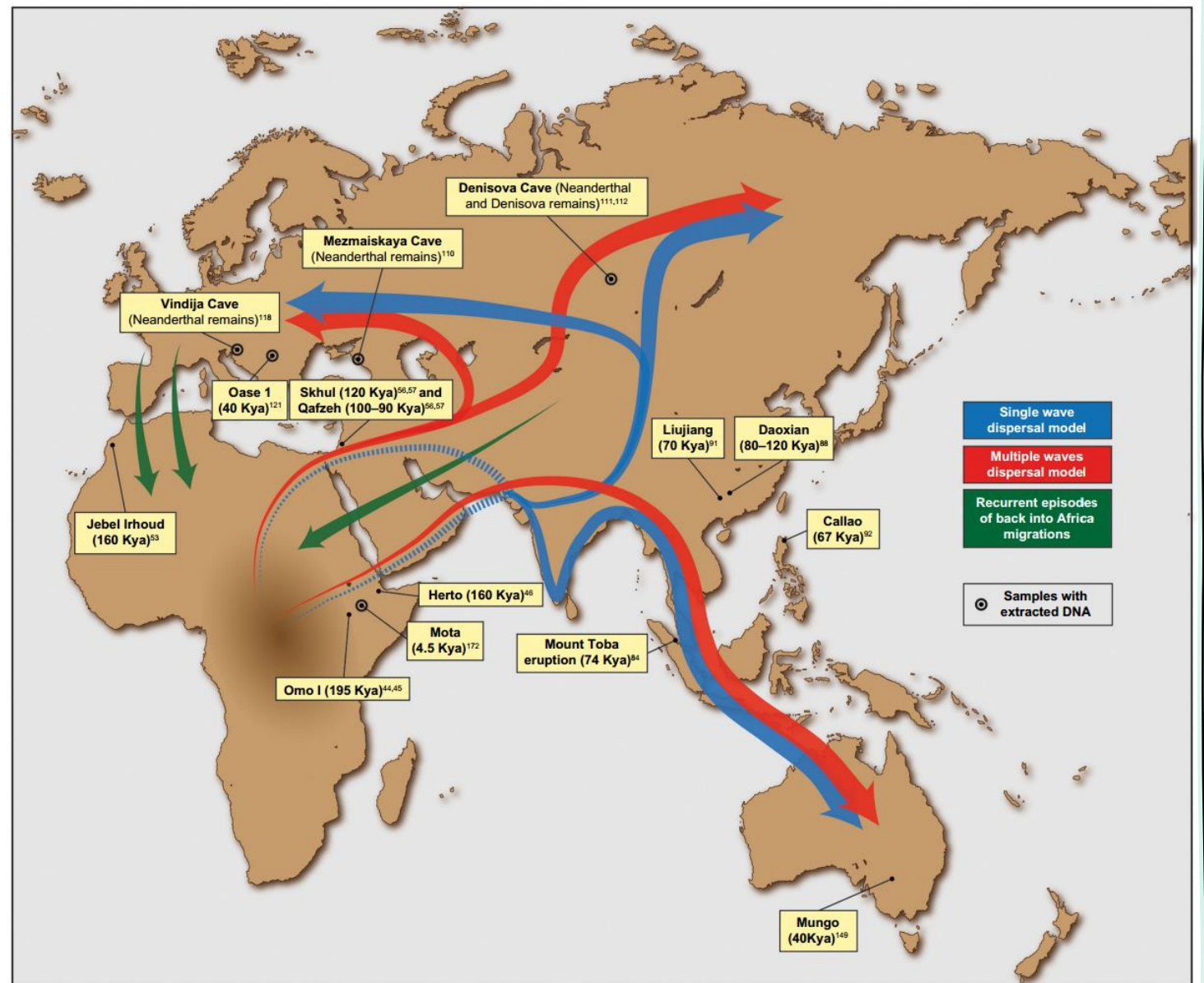


THE ARTICLE DISCUSSES:

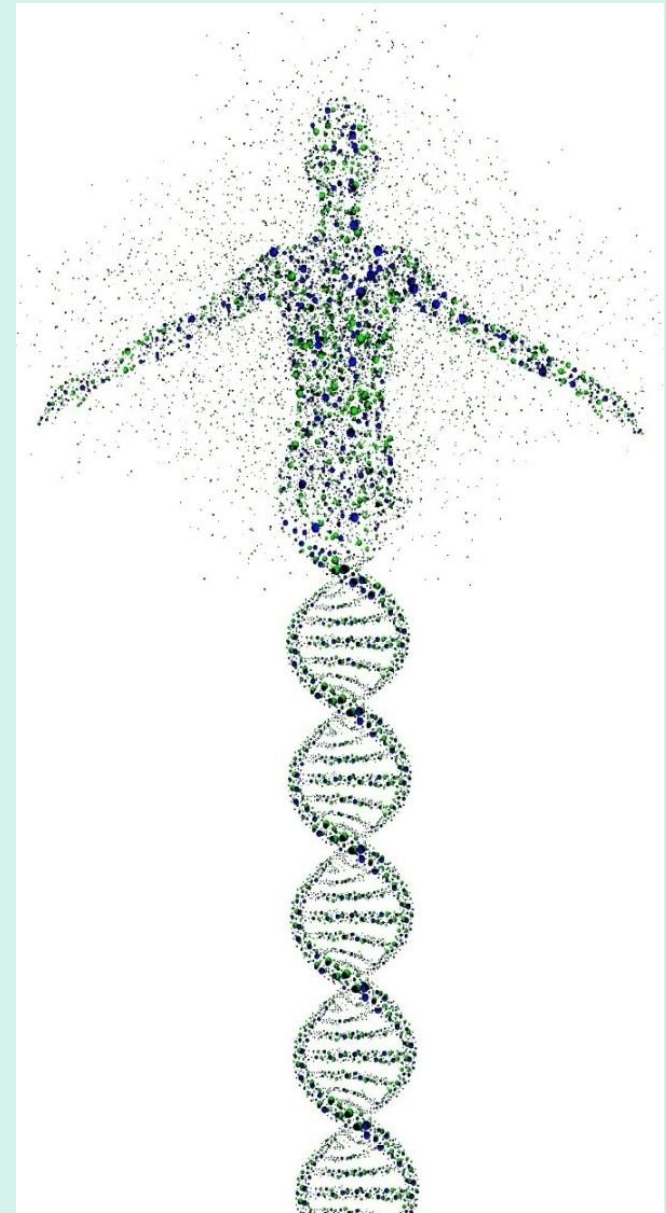
1. Possibility of archaic & "ghost" introgression in Africa, with the recovery of the first genome-wide data from Neanderthals & Denisovans, and new methods that do not require a reference genome from the introgressing lineage.
2. Archaic introgression involves a hominin group separated from the human lineage before the deepest divergence among modern humans, while "ghost" introgression involves extinct anatomically modern groups that separated around 300,000 years ago or more recently.

1. How the Neanderthal ancestry proportion in modern Africans can be explained by back migration from an ancestral European-like source into Africa?

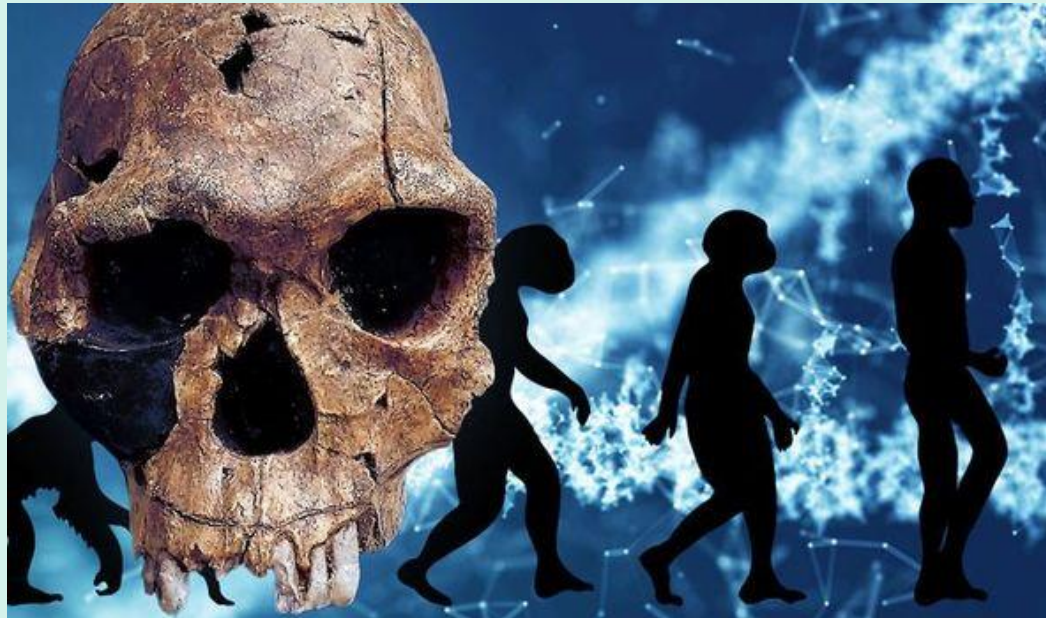
2. How signals of introgression from deeply branching populations had already been detected in African populations even before the first Neanderthal genome was published?



- Interestingly, many studies identified a fairly recent time for the introgression from extinct lineages in Africa, with introgression events even after the split from non-African populations, hinting at survival of archaic human populations until relatively recently in time
- This contrasts with observations from the fossil record, where most of the morphological diversity disappeared before 100 000 ya



FUTURE OUTLOOK



The origin of Homo sapiens is an evolutionary process that occurred over a relatively long time.

The deepest divergence among humans, about 300,000 years ago, can be viewed as a lower bound for the existence of fully developed modern humans.

The Neandertal/Denisovan divergence, about 650,000 years ago, can be viewed as an upper bound for the development of unique features of Homo sapiens.

Unique characteristics of Homo sapiens may be governed by genetic changes, including frequency changes, and potentially epigenetic changes.

There are various models of the origin of our species in Africa, including a single origin, an African multiregional model, and range expansion from one group/area.

The diverse morphology from fossil finds, material culture, and genetics suggest a scattered diverse population with sporadic gene-flow may have contributed to the emergence of modern Homo sapiens.

FUTURE OUTLOOK



- It is unclear how a model of a structured metapopulation can both uphold sufficient barriers to gene-flow to cause stratification while allowing for genetic variants important to modern human development to spread among populations simultaneously.
- The early history of humans is complex and difficult to disentangle due to later events such as gene-flow and migration.
- More complex models, diverse data, and better statistical tools are needed to understand the intricate demography of African populations and inform on the origin of the species.
- There is still a bias in data sampling, with southern Africa overrepresented in both modern and ancient genomic data.
- More genomic data from understudied regions and improvements in analyzing ancient DNA are needed to refine our understanding of African prehistory and decipher early events.

Thank you!

