

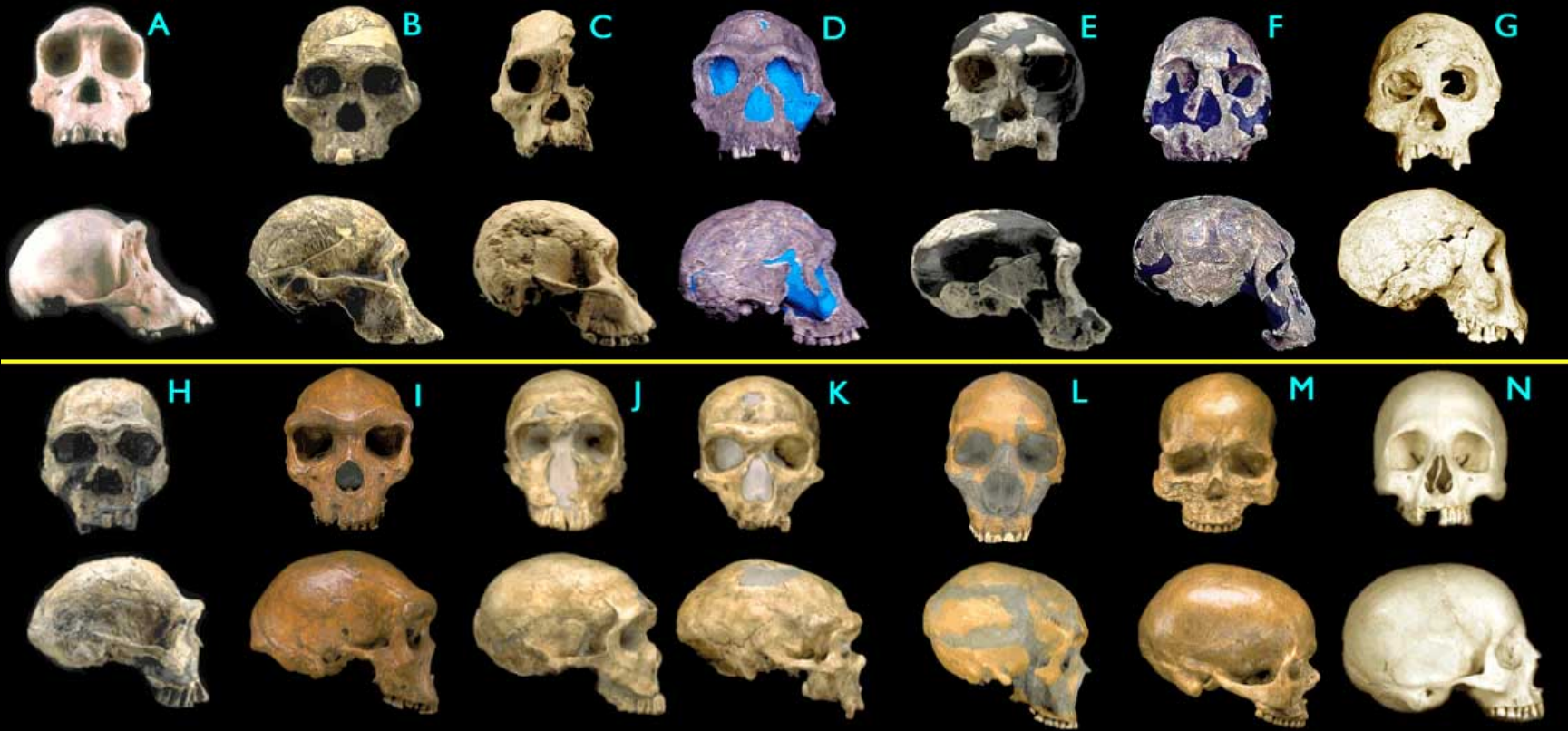
Who am I? Where did I come from?

Out of Africa, Genomics and Human
Migrations – the big picture.

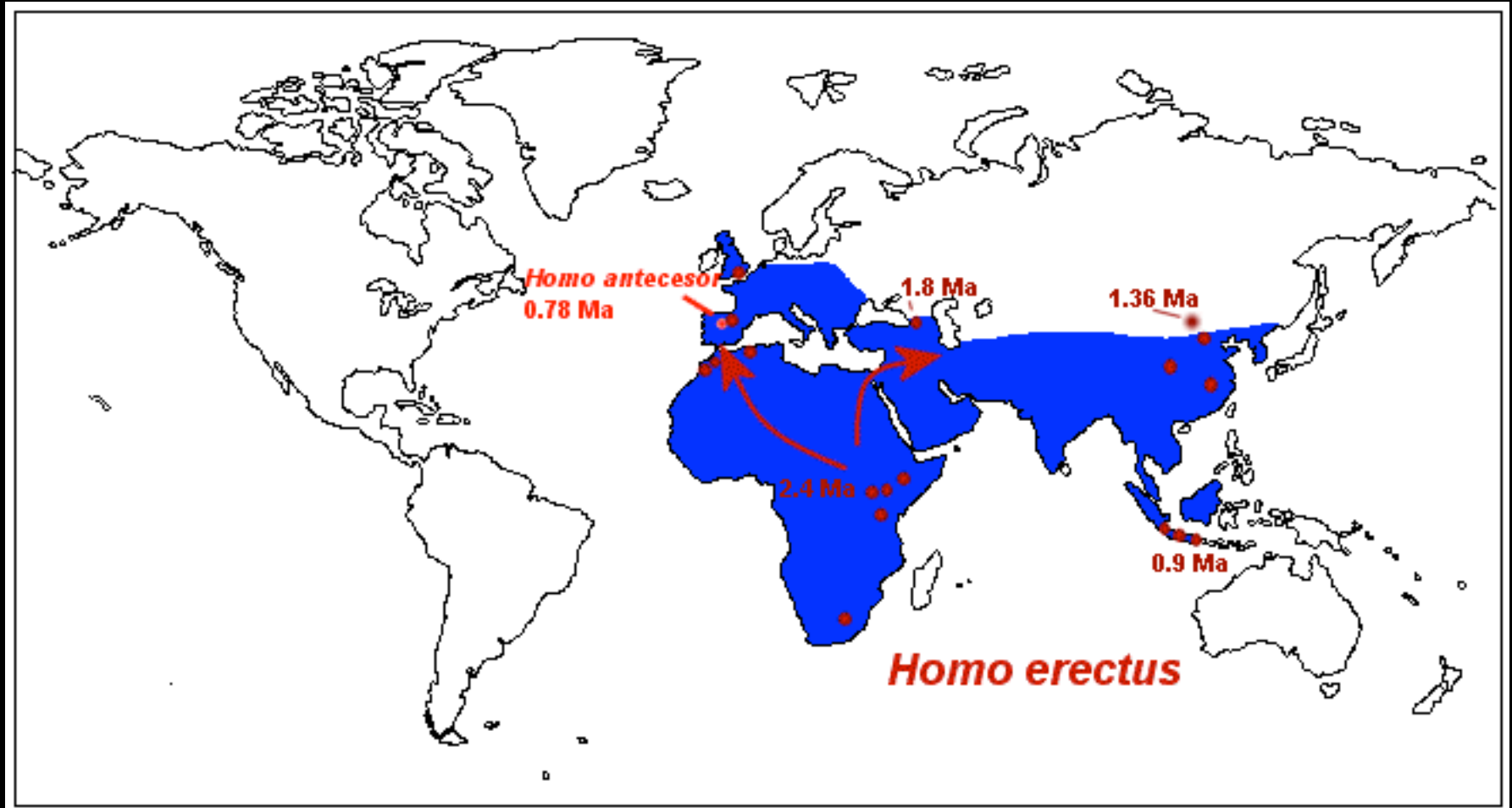
Lisa Matisoo-Smith
University of Otago



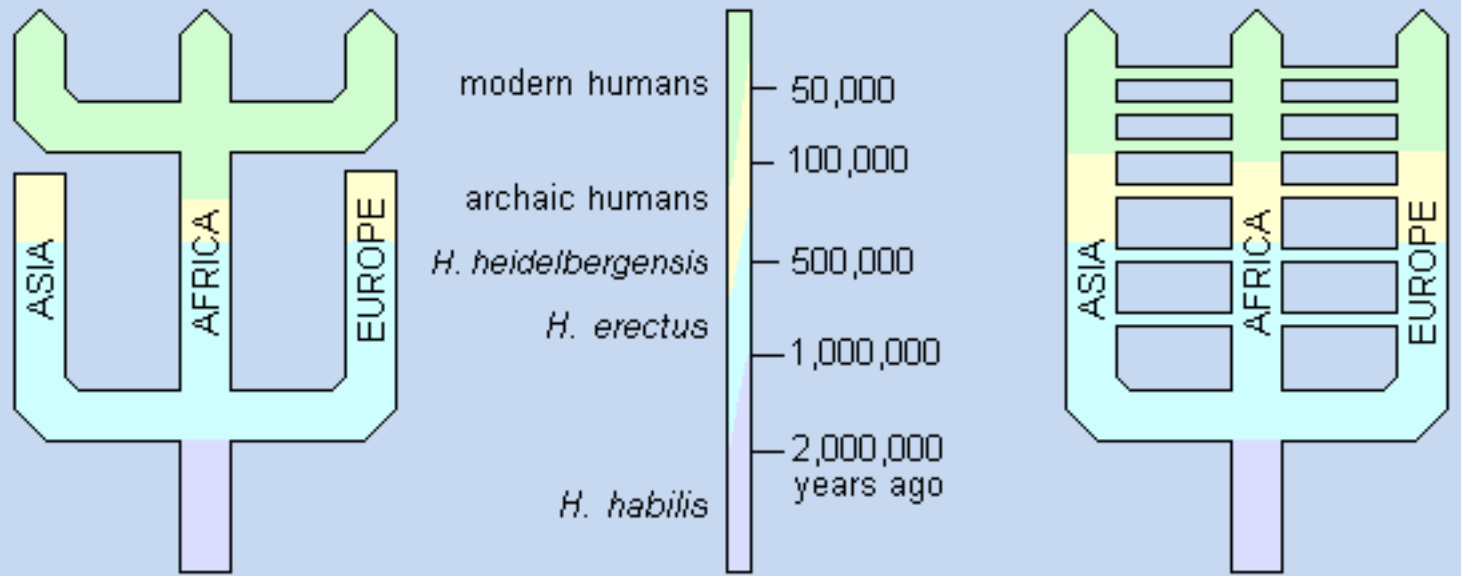
Reconstructing Human Evolution



First “Out of Africa” migration: *Homo erectus*



Replacement vs Regional Continuity Debate

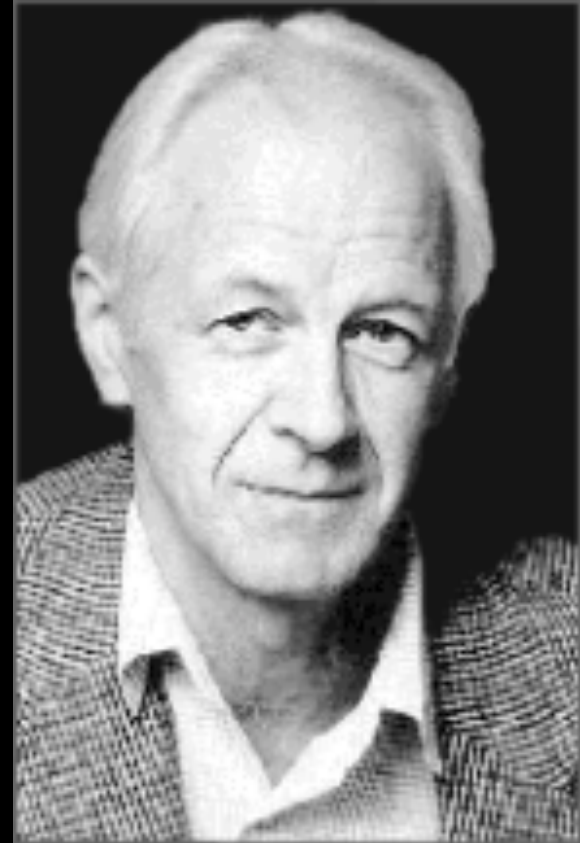


Replacement Model
"Out of Africa"

Regional Continuity Model

Allan Wilson (1934 - 1991)

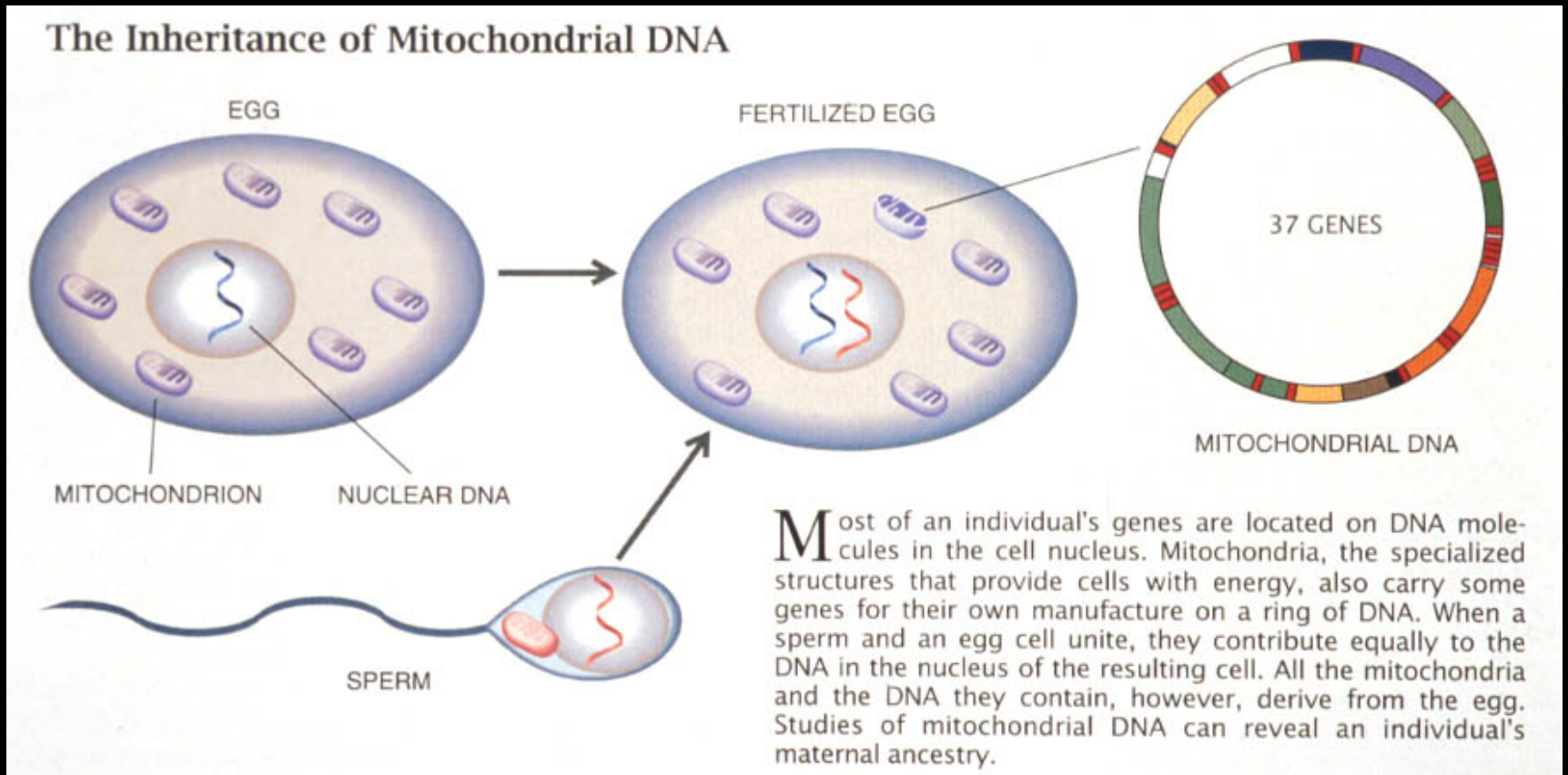
- Born in Ngaruawahia
- BSc Otago
- PhD UC Berkeley
- The Wilson Lab
 - molecular evolution
 - molecular clock
 - mtDNA
 - ancient DNA



We don't know
if all of those
fossils had
descendants –
but we do
know that we
all had
ancestors....



Inheritance of mtDNA

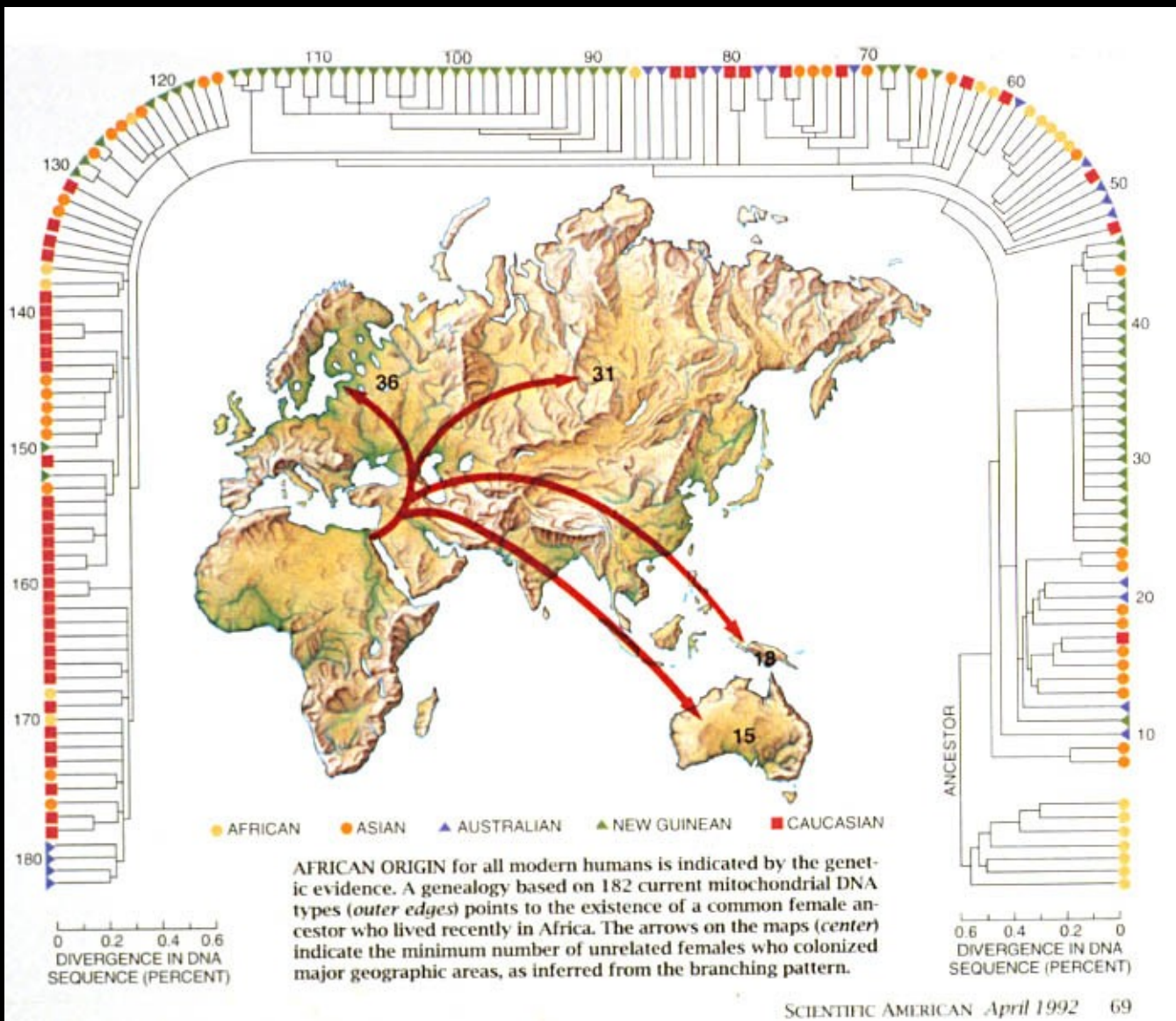


MtDNA is inherited only from your mother.

MtDNA does not recombine (or mix) with paternal DNA.

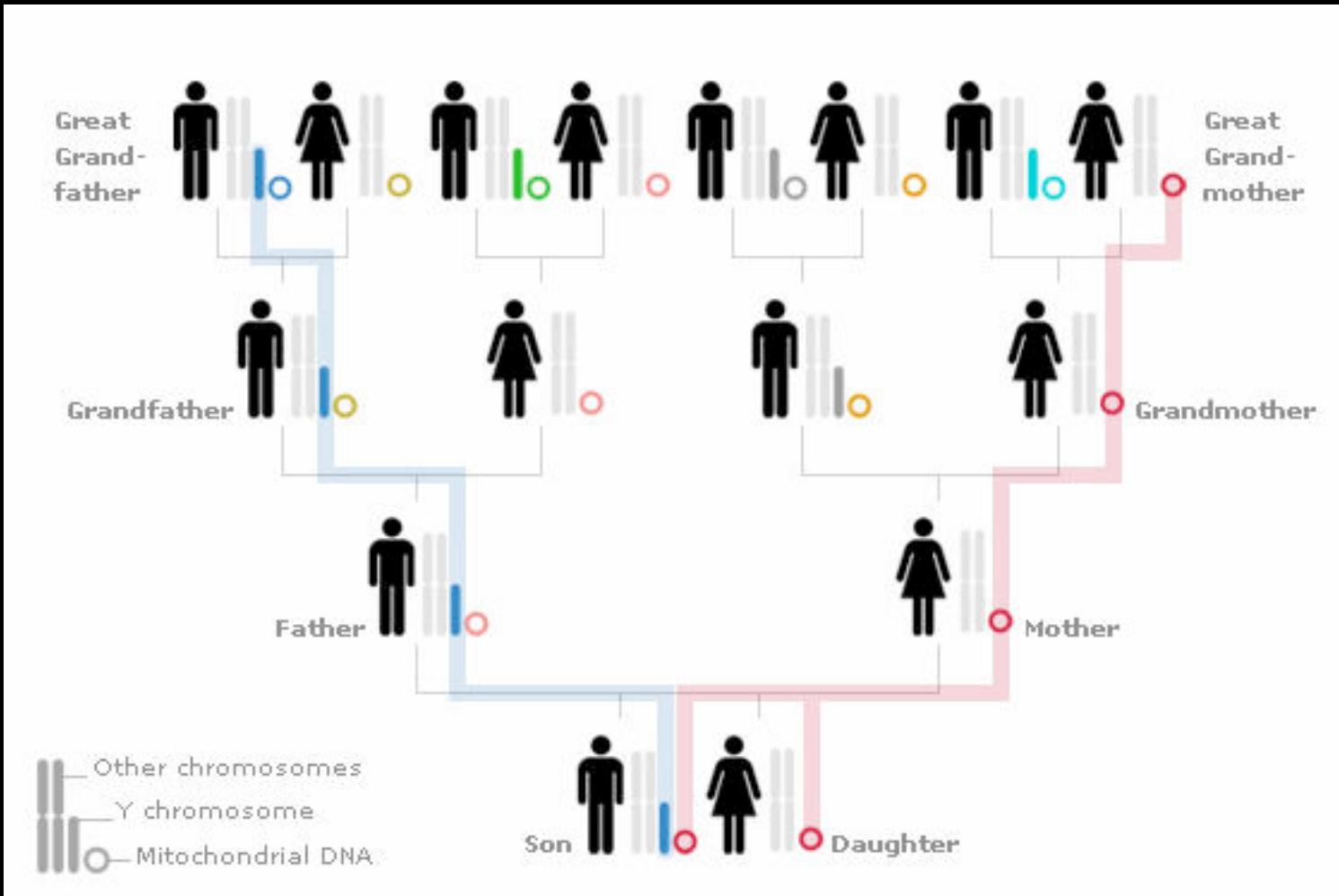
Mitochondrial "Eve"

based on Cann et al. 1987



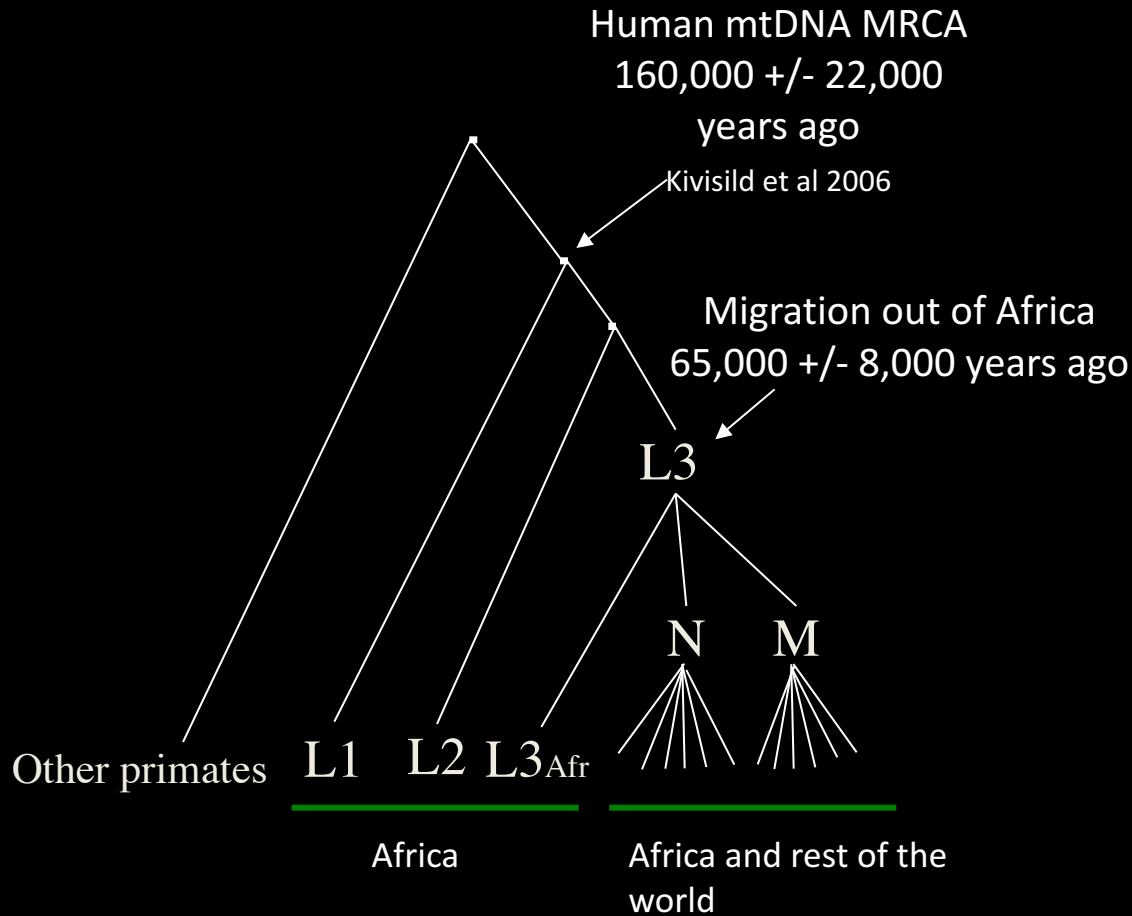
An evolutionary tree from 182 women with ancestry from populations around the world based on mtDNA differences.

Traced all lineages to a single maternal line that existed in Africa over 150,000 years ago



<https://genographic.nationalgeographic.com/v/>

Human mtDNA variation



of Base changes to ancestral nodes

MRCA average ~70

L3 node average ~31

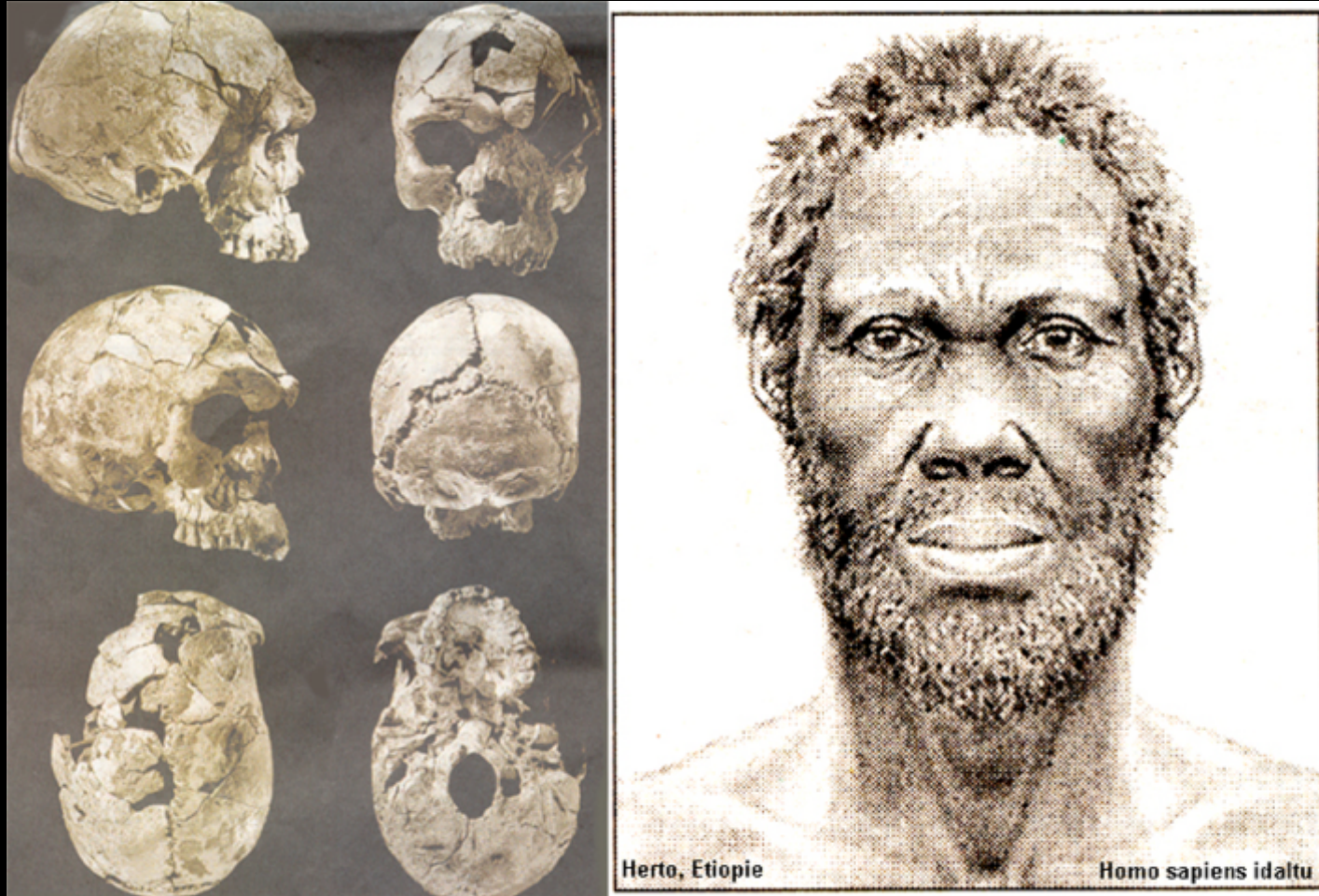
(1283 human-chimp)

Tree based on Human mtDNA data as of 2008

~ 16.5 thousand base pairs of mtDNA

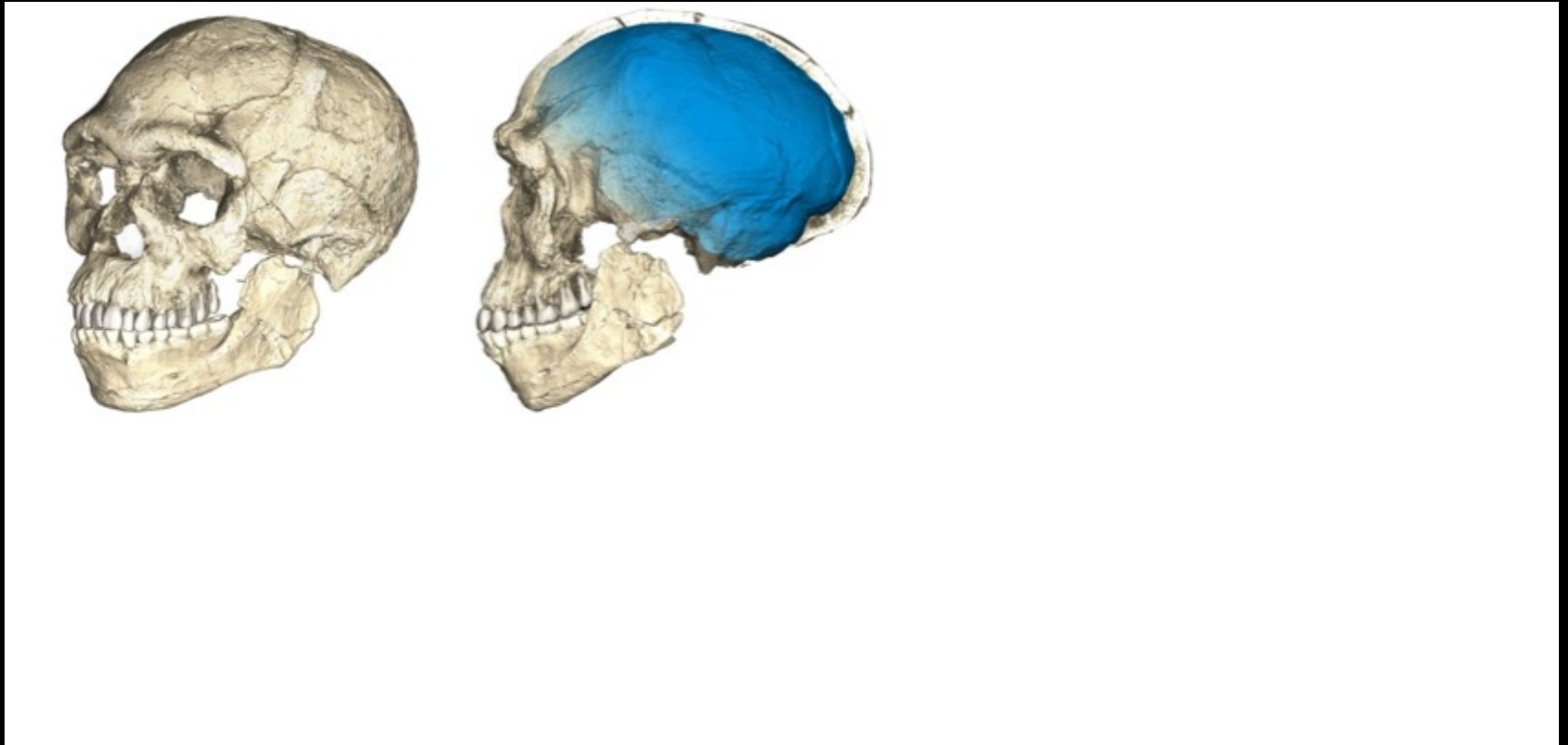
MRCA = Most Recent Common Ancestor

Earliest *Homo sapiens* remains dated to 160,000 BP in Ethiopia



300,000 year-old “early Homo sapiens”

Jebel Irhoud, Morocco, June 2017, by Jean-Jacques Hublin



<https://3c1703fe8d.site.internapcdn.net/newman/gfx/news/hires/2017/thefirstofou.jpg>

Early spread of modern humans from east African source, 60,000 to 40,000 years ago

Later spread of modern humans from west Asian source, 45,000 to 35,000 years ago

Hypothesized east African source area of modern humans, 80,000 to 60,000 year ago

Hypothesized West Asian source area of modern northern Eurasians and north Africans, 50,000–45,000 years ago

Hofmeyer

Haua Fteah

Ksar Akil

Kostenki

CASPIAN SEA

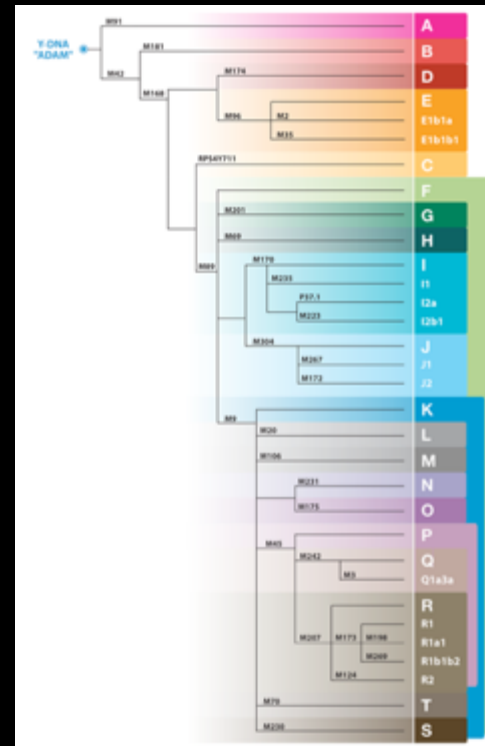
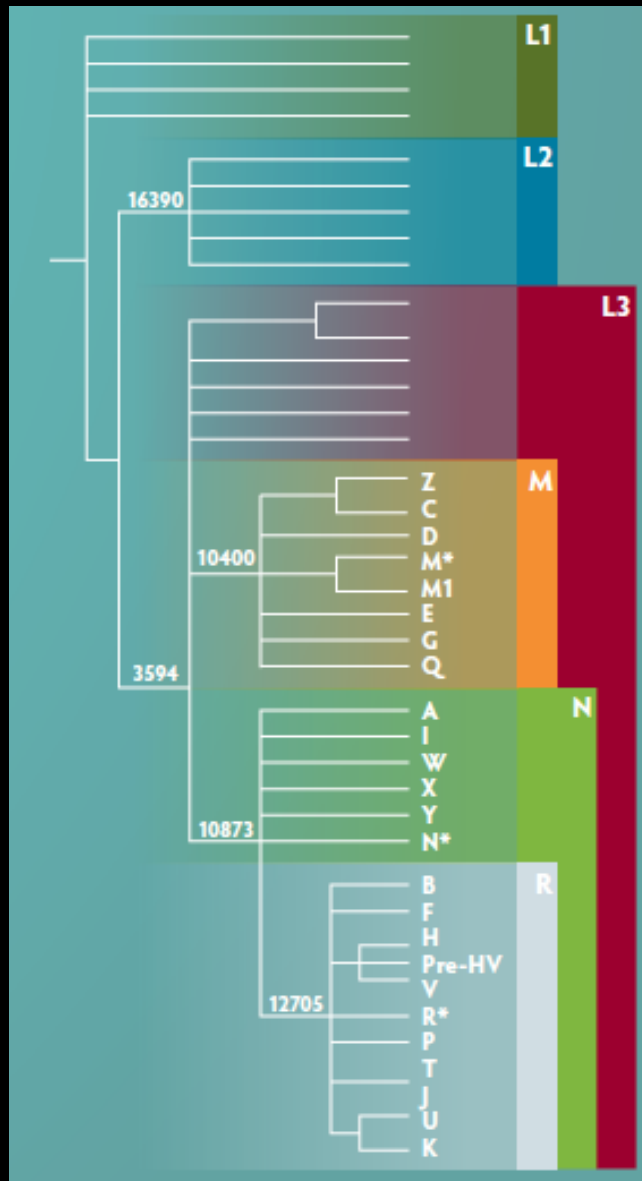
LAKE BAIKAL

Human pathways. Reconstructed spread of modern humans during the late Pleistocene, and locations of some key early Upper Paleolithic archaeological sites. Grine *et al.*, Olivieri *et al.*, and Anikovich *et al.* provide new evidence confirming that early modern humans spread from southwestern Asia into northern Africa, Europe, and Russia about 45,000 to 40,000 years ago.

Haplogroups

MtDNA

Y-Chromosome



YOUR GENETIC SEQUENCE

Type : mtDNA

Haplogroup : T (Subclade T2)

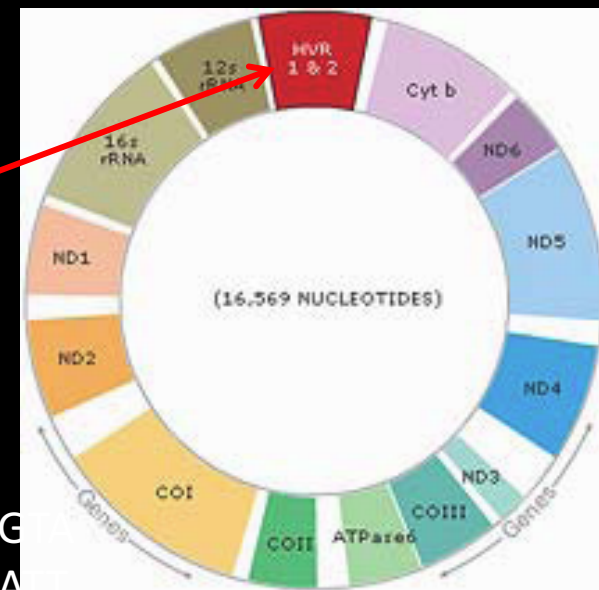
Your Mitochondrial HVR I Sequence

16126C, 16192T, 16207G, 16274A, 16294T, 16304C, 16519C

```
ATTCTAATTTAAACTATTCTCTGTTCTTTCATGGGGAAGCAGATTG  
CCACCCAAGTATTGACTCACCCATCAACAACCGCTATGTATTCGTACA  
ACTGCCAGCCACCATGAATATTGCACGGTACCATAAATACTTGACCACCTG  
TAGTACATAAAAACCCAATCCACATCAAACCCCTCCTCATGCTTACAAG  
CAGGTACAGCAATCAACCCTCAACTATCACACATCAACTGCAACTCCAAAG  
CCACCCCTCACCCACTAGAATACCAACAAACCTACCCATCCTTAACAGCAC  
ATAGTACATAAAGCCATTTACCGTACATAGCACATTACAGTCAAATCCCTT  
CTCGTCCCATGGATGACCCCTCAGATAGGGGTCCCTTGACCACCATCC  
TCCGTGAAATCAATATCCCGCACAAAGAGTGCTACTCTCCTCGCTCCGGGCC  
CATAACACTTGGGGGTAGCTAAAGTGAAGTGTATCCGACATCTGGTTCCTA  
CTTCAGGGCCATAAAGCCTAAATAGCCCACACGTTCCCCTTAAATAAGACA  
TCACGATG
```

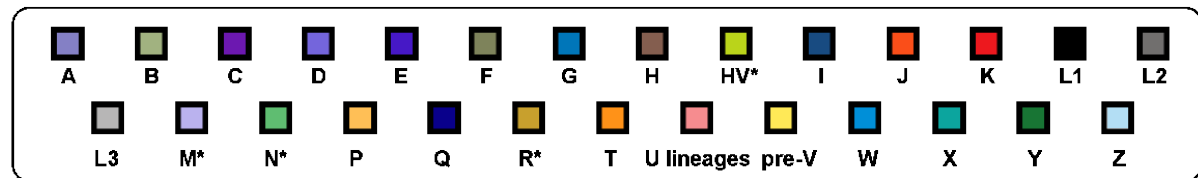
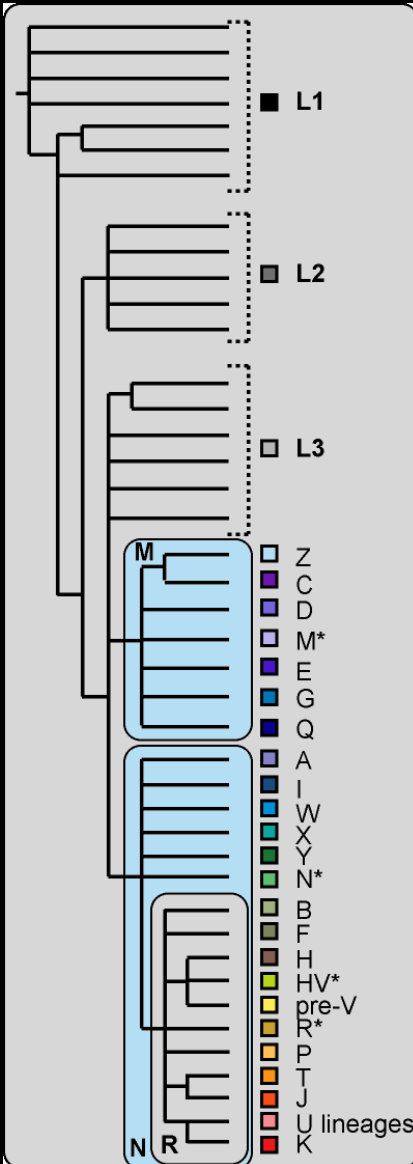
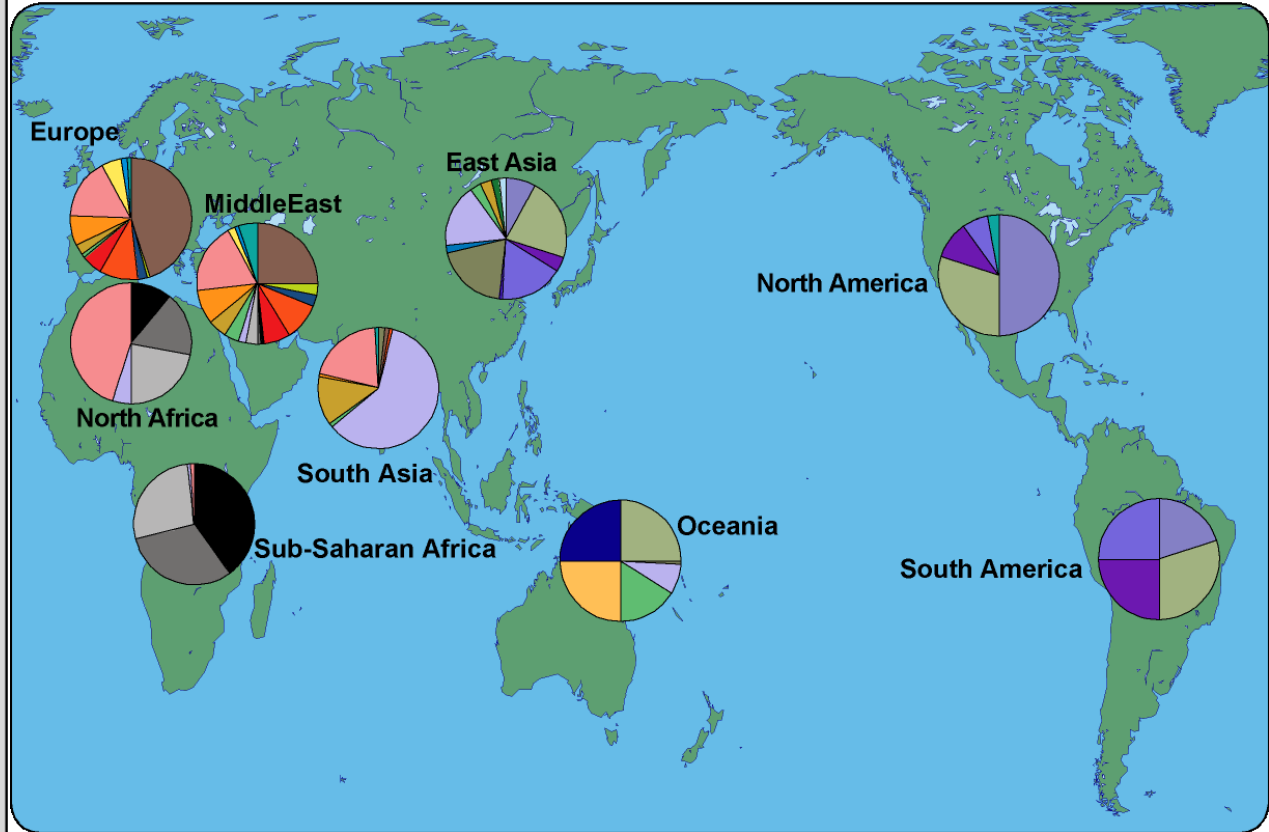
Key C Substitution(transition) C Substitution(transversion) C Insertion _ Deletion

Transition = C ↔ T or G ↔ A Transversion = C ↔ G ; C ↔ A ; T ↔ G ; T ↔ A



Haplogroups

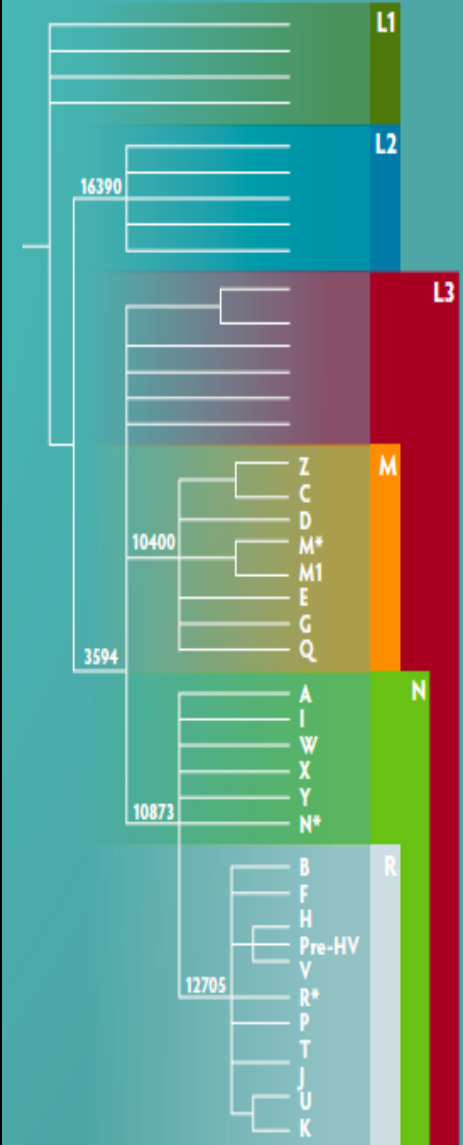
mtDNA haplogroup distribution



Mitochondrial DNA (mtDNA)



Mitochondrial DNA (mtDNA): This is maternally inherited and allows us to identify the ancestral migratory origins of your direct maternal line.



A Draft Sequence of the Neandertal Genome

Richard E. Green,^{1*}†‡ Johannes Krause,¹†§ Adrian W. Briggs,¹†§ Tomislav Maricic,¹†§ Udo Stenzel,¹†§ Martin Kircher,¹†§ Nick Patterson,²†§ Heng Li,²† Weiwei Zhai,³†|| Markus Hsi-Yang Fritz,⁴† Nancy F. Hansen,⁵† Eric Y. Durand,³† Anna-Sapfo Malaspinas,³† Jeffrey D. Jensen,⁶† Tomas Marques-Bonet,^{7,13}† Can Alkan,⁷† Kay Prüfer,¹† Matthias Meyer,¹† Hernán A. Burbano,¹† Jeffrey M. Good,^{1,8}† Rigo Schultz,¹ Ayinuer Aximu-Petri,¹ Anne Butthof,¹ Barbara Höber,¹ Barbara Höffner,¹ Madlen Siegemund,¹ Antje Weihmann,¹ Chad Nusbaum,² Eric S. Lander,² Carsten Russ,² Nathaniel Novod,² Jason Affourtit,⁹ Michael Egholm,⁹ Christine Verna,^{2,1} Pavao Rudan,¹⁰ Dejana Brajkovic,¹¹ Željko Kucan,¹⁰ Ivan Gušić,¹⁰ Vladimir B. Doronichev,¹² Liubov V. Golovanova,¹² Carles Lalueza-Fox,¹³ Marco de la Rasilla,¹⁴ Javier Fortea,¹⁴¶ Antonio Rosas,¹⁵ Ralf W. Schmitz,^{16,17} Philip L. F. Johnson,¹⁸† Evan E. Eichler,⁷† Daniel Falush,¹⁹† Ewan Birney,⁴† James C. Mullikin,⁵† Montgomery Slatkin,³† Rasmus Nielsen,³† Janet Kelso,¹† Michael Lachmann,¹† David Reich,^{2,20*}† Svante Pääbo^{1*}†

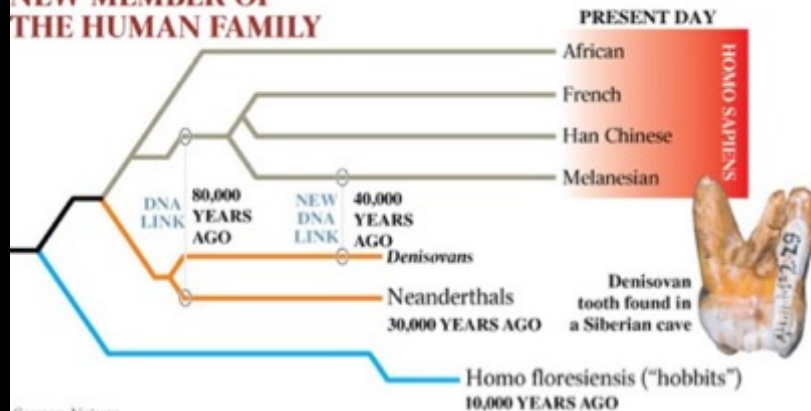
Neandertals, the closest evolutionary relatives of present-day humans, lived in large parts of Europe and western Asia before disappearing 30,000 years ago. We present a draft sequence of the Neandertal genome composed of more than 4 billion nucleotides from three individuals. Comparisons of the Neandertal genome to the genomes of five present-day humans from different parts of the world identify a number of genomic regions that may have been affected by positive selection in ancestral modern humans, including genes involved in metabolism and in cognitive and skeletal development. We show that Neandertals shared more genetic variants with present-day humans in Eurasia than with present-day humans in sub-Saharan Africa, suggesting that gene flow from Neandertals into the ancestors of non-Africans occurred before the divergence of Eurasian groups from each other.

Genetic history of an archaic hominin group from Denisova Cave in Siberia

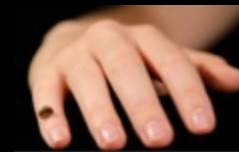
David Reich^{1,2*}, Richard E. Green^{3,4*}, Martin Kircher^{3*}, Johannes Krause^{3,5*}, Nick Patterson^{2*}, Eric Y. Durand^{6*}, Bence Viola^{3,7*}, Adrian W. Briggs^{1,3}, Udo Stenzel³, Philip L. F. Johnson⁸, Tomislav Maricic³, Jeffrey M. Good⁹, Tomas Marques-Bonet^{10,11}, Can Alkan¹⁰, Qiaomei Fu^{3,12}, Swapan Mallick^{1,2}, Heng Li², Matthias Meyer³, Evan E. Eichler¹⁰, Mark Stoneking³, Michael Richards^{7,13}, Sahra Talamo⁷, Michael V. Shunkov¹⁴, Anatoli P. Derevianko¹⁴, Jean-Jacques Hublin⁷, Janet Kelso³, Montgomery Slatkin⁶ & Svante Pääbo³

Using DNA extracted from a finger bone found in Denisova Cave in southern Siberia, we have sequenced the genome of an archaic hominin to about 1.9-fold coverage. This individual is from a group that shares a common origin with Neanderthals. This population was not involved in the putative gene flow from Neanderthals into Eurasians; however, the data suggest that it contributed 4–6% of its genetic material to the genomes of present-day Melanesians. We designate this hominin population 'Denisovans' and suggest that it may have been widespread in Asia during the Late Pleistocene epoch. A tooth found in Denisova Cave carries a mitochondrial genome highly similar to that of the finger bone. This tooth shares no derived morphological features with Neanderthals or modern humans, further indicating that Denisovans have an evolutionary history distinct from Neanderthals and modern humans.

NEW MEMBER OF THE HUMAN FAMILY



Source: Nature



Ancient DNA

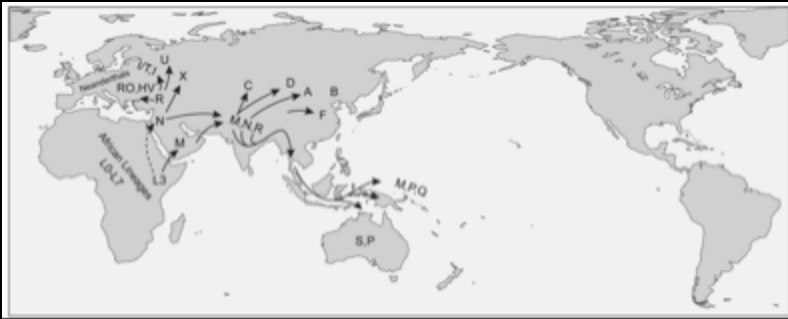
- Neanderthal mtDNA genome
- Neanderthal whole genome
- Denisovan genome
- Admixture – multiple events
- Outside of Africa
- Implications for models?



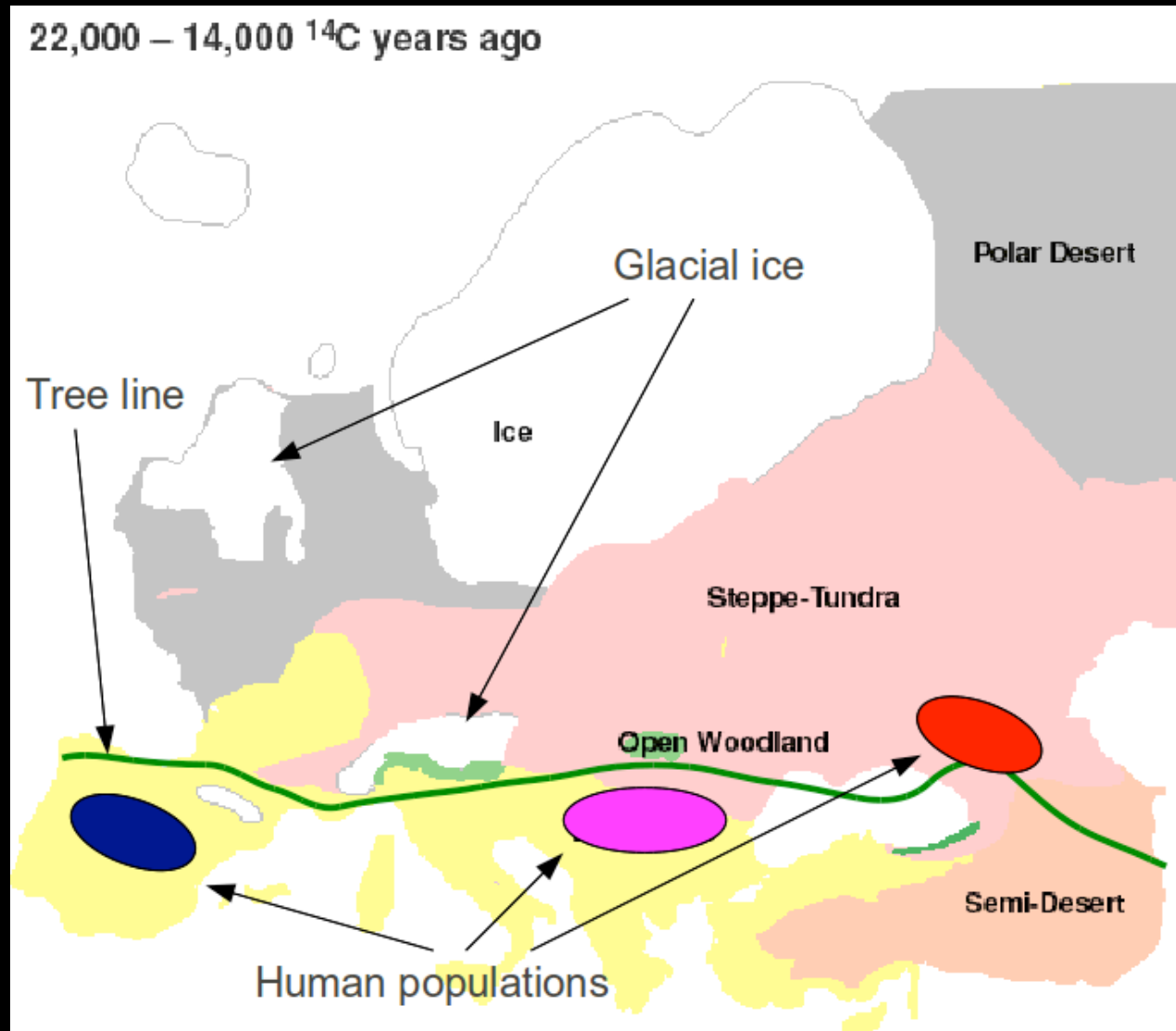
<https://en.wikipedia.org/wiki/Denisovan>

Palaeolithic migrations

40,000 Years Ago (40,000 BP)

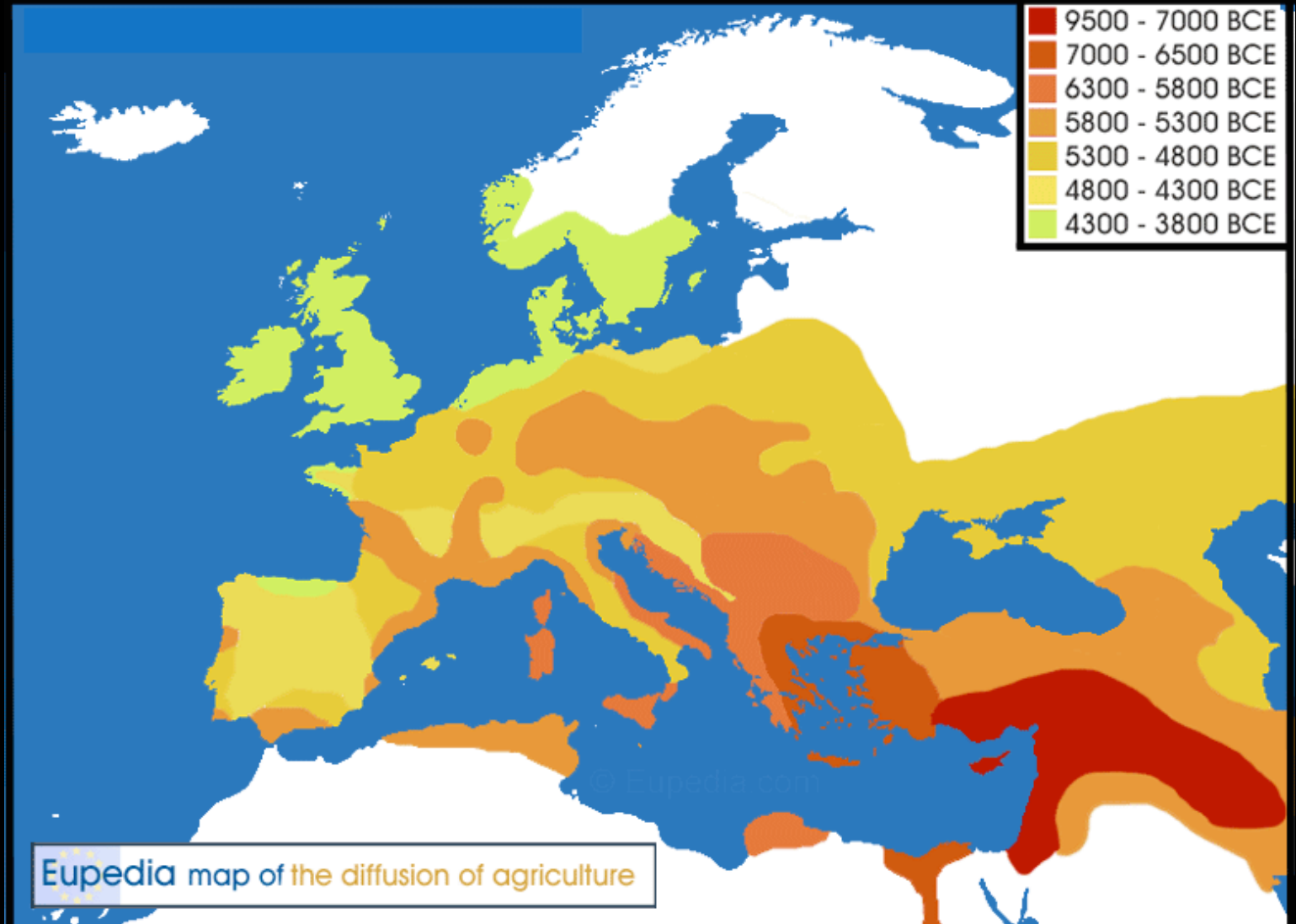


Last Glacial Maximum (Ice Age)



Neolithic expansions

The spread of agriculture & Indo-European



Population Expansions & Replacements



http://ichef.bbc.co.uk/news/624/media/images/77640000/gif/_77640634_europe_ancient_farmers_20140917-01.gif

Genomics & Migrations

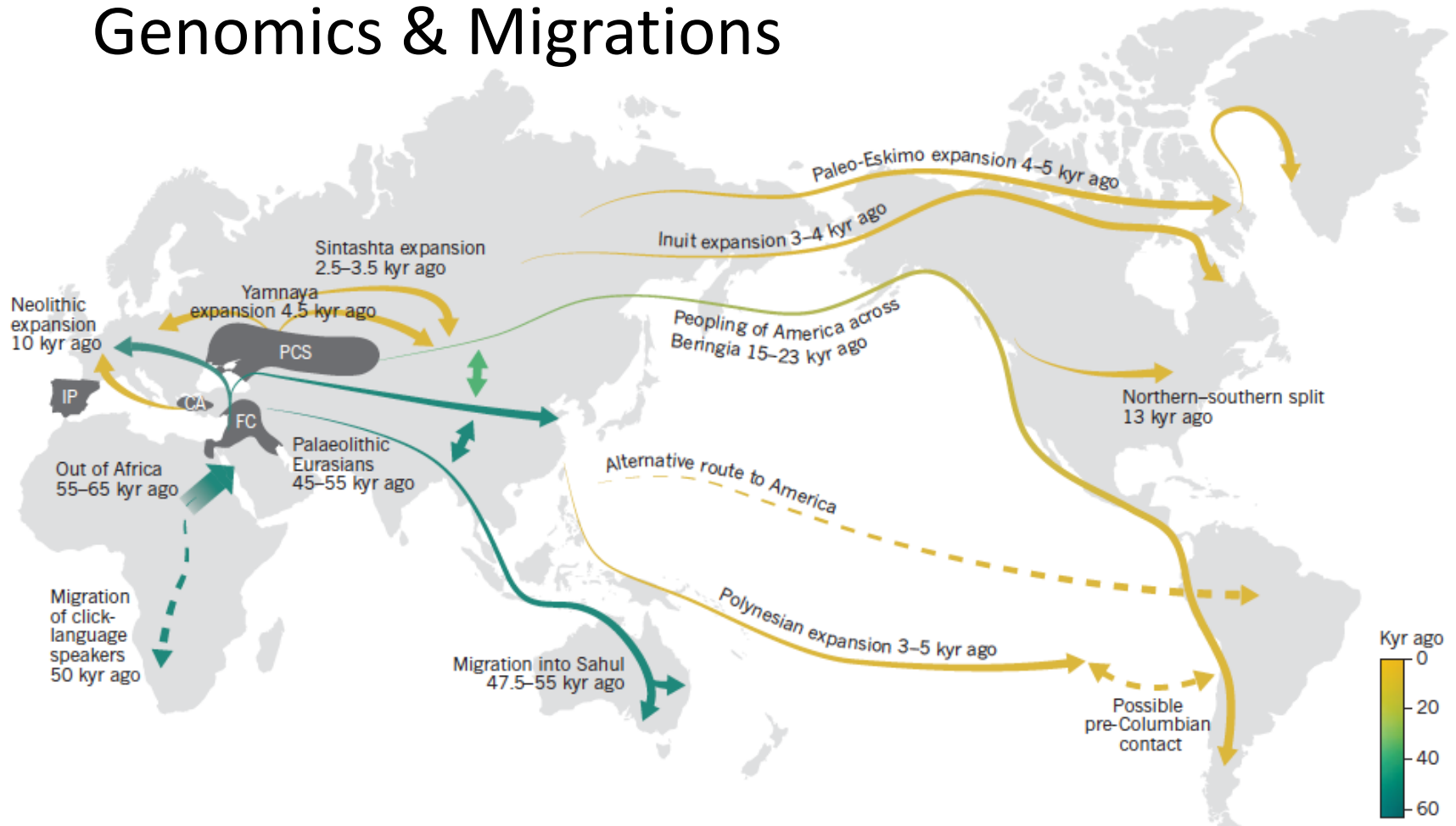
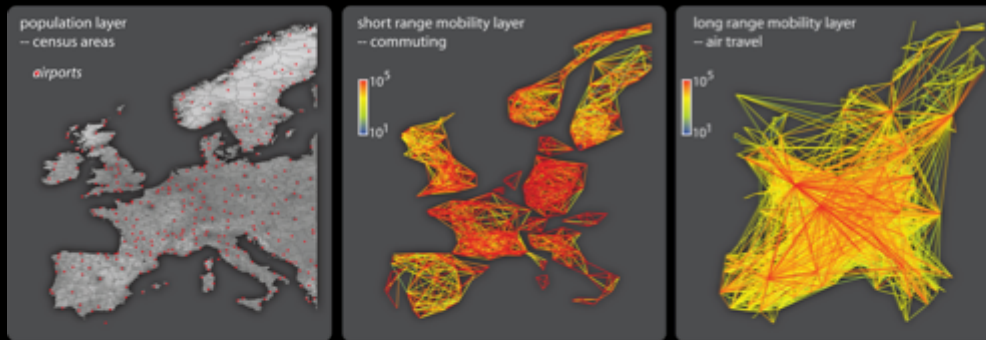


Figure 3 | Major human migrations across the world inferred through analyses of genomic data. Some migration routes remain under debate. For example, there is still some uncertainty regarding the migration routes used to populate the Americas. Genomic data are limited in their resolution to determine paths of migration because further population

movements, subsequent to the initial migrations, may obscure the geographic patterns that can be discerned from the genomic data. Proposed routes of migration that remain controversial are indicated by dashed lines. CA, Central Anatolia; FC, Fertile Crescent; IP, Iberian Peninsula; PCS, Pontic-Caspian steppe.

Modern Human Mobility



http://epifor.isi.it/media/xf/i/GLEaM_EU_layers__.png

Conclusions

- We all share a common ancestry in Africa – 200,000+ years ago
- Neither the strict Out of Africa or Regional Continuity models is supported by DNA – we seem to have a relaxed Out of Africa with evidence of admixture with Neanderthal & Denisovans
- Human history shaped by numerous physical and cultural events (LGM, spread of agriculture and plagues, major invasions etc)
- We are learning more all of the time and have to revise our hypotheses and explanations based on the new data.