Understanding Us: What Makes Difference and What Difference Does Difference Make

Friday May 1

- **Linnaeus vs Darwin**
- **Evolutionary adaptions to different environments (Reading "Adventures in Very Recent Evolution")**
- **Graphical Analysis (Same or Different)**

Monday May 4….

- Quick check on early *Homo sapiens* evolution and interaction with Neanderthals and Denisovans….
- Revisit Graphical Analysis in pairs
- Connecting those images to CH 2 Racism not Race

A quick but **IMPORTANT** check in from earlier in the semester…. Linnaeus vs Darwin.

What do you remember about them?

Would they have mostly agreed with one another or do you think they would have argued?

Pre-Darwinian World View	**Darwinian World View**
Linnaeus	**Darwin**
Really, really into categorizing living things.	Hated categories.
All this categorization means you need to decide who belongs in each category-make up rules.	Categorizing species is not super important because individuals within species vary in a continuum over space and time.
Variation within species is concerning because species were created by God.	Variation within and between populations is abundant and critical to evolutionary change.

Even today every described species has an individual specimen chosen to represent that species in a museum.
It is called the “Type Specimen”
ESSENTIALISM!

Questions from your reading on Evolutionary Adaptations to different environments

1. Why do you think we used to imagine that humans would not experience natural selection?

2. Pick out and describe all the examples of recent evolution in humans (**what is up with earwax-was it selected for?**).

3. How did they detect the occurrence of selection?

4. If a trait was selected for a very long time ago, how would the regions around that gene differ from whether it was more recently selected for?

5. They point out that there are at least 25 genes that play a role in skin pigmentation. How accurate do you think this number is?

6. Describe the skin color example.

7. The authors note that ear wax differs between populations, but are not sure what survival advantage different kinds of earwax might confer**. Instead they think the difference exists due to…..?**

8. Africans show the highest levels of "neutral" or "background" variation while Europeans and East Asians seem to show more genes under recent selection. **Why might both these patterns exist?**

9. Hard vs. soft sweeps? What does this mean?

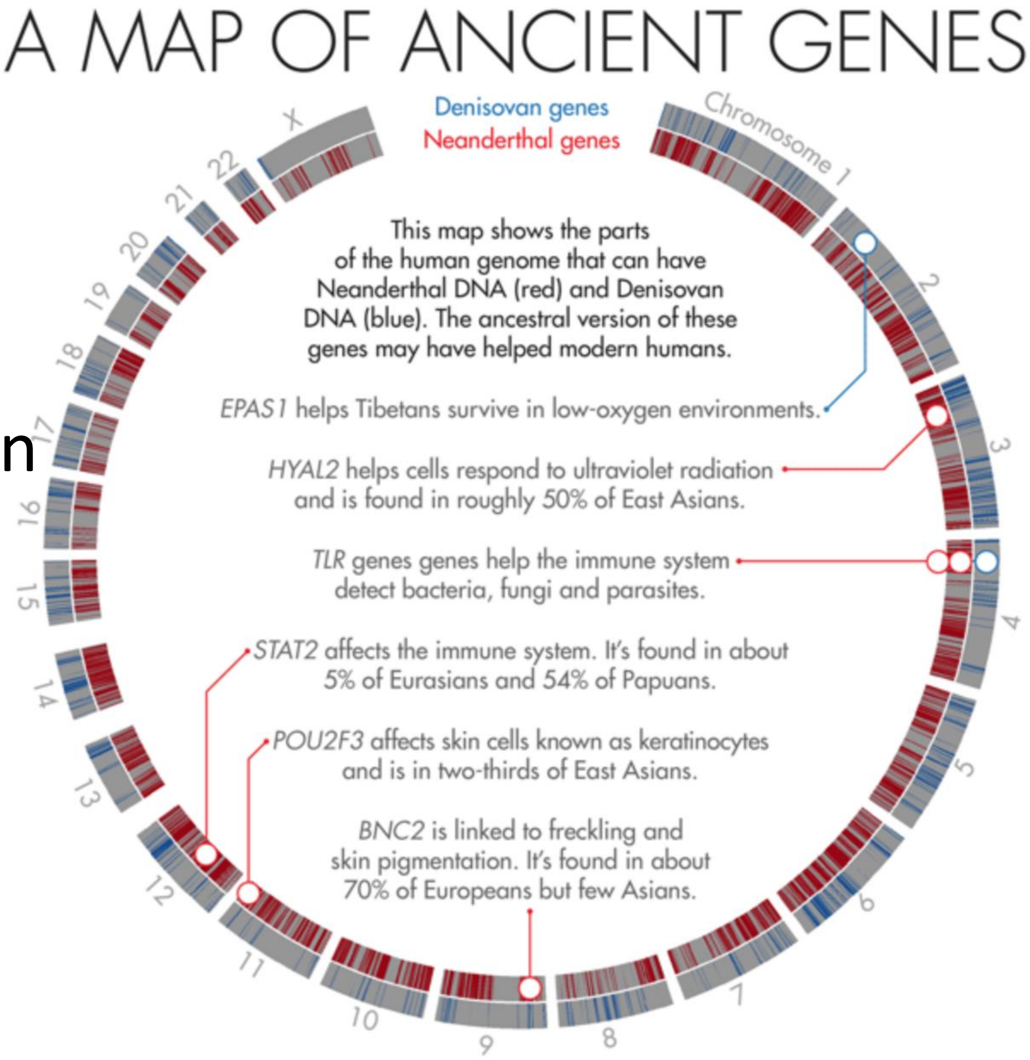
Graphical Analysis

These images should prepare you to think deeply about the way we categorize peoples and whether that categorization is biologically appropriate and what perceptions potentially bias that understanding.

(from How Neanderthals Gave us Secret Powers)

Neanderthal
Skin pigmentation,
Immune system
Allergies

Denisovan
Tolerance to low oxygen



A MAP OF ANCIENT GENES

This map shows the parts of the human genome that can have Neanderthal DNA (red) and Denisovan DNA (blue). The ancestral version of these genes may have helped modern humans.

EPAS1 helps Tibetans survive in low-oxygen environments.

HYAL2 helps cells respond to ultraviolet radiation and is found in roughly 50% of East Asians.

TLR genes genes help the immune system detect bacteria, fungi and parasites.

STAT2 affects the immune system. It's found in about 5% of Eurasians and 54% of Papuans.

POU2F3 affects skin cells known as keratinocytes and is in two-thirds of East Asians.

BNC2 is linked to freckling and skin pigmentation. It's found in about 70% of Europeans but few Asians.

1.

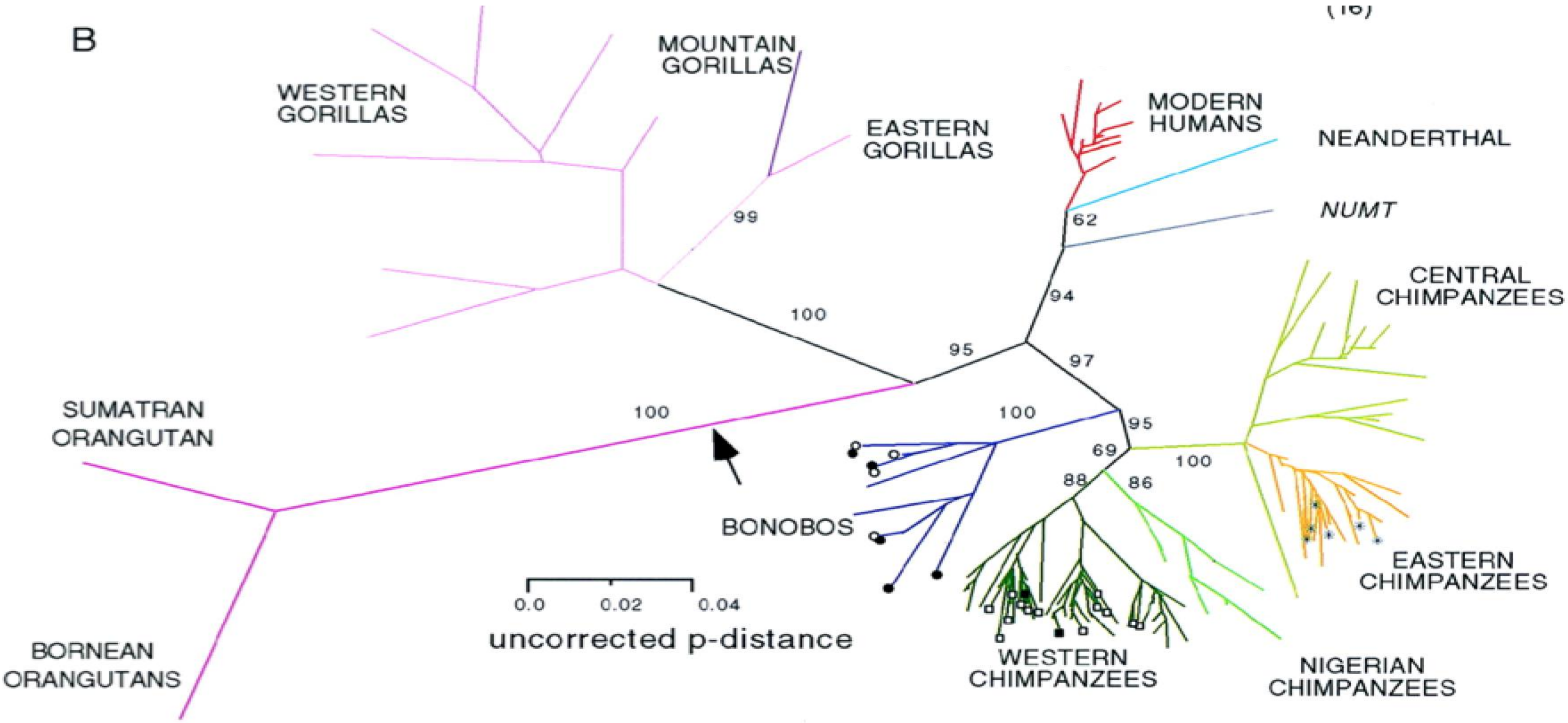
2.

https://www.buzzfeed.com/annamendoza/all-the-colours-of-the-world

Segment length is proportional to genetic difference.

Gagneux et al. PNAS 1999;96:9:5077-5082

https://www.pnas.org/content/96/9/5077.full

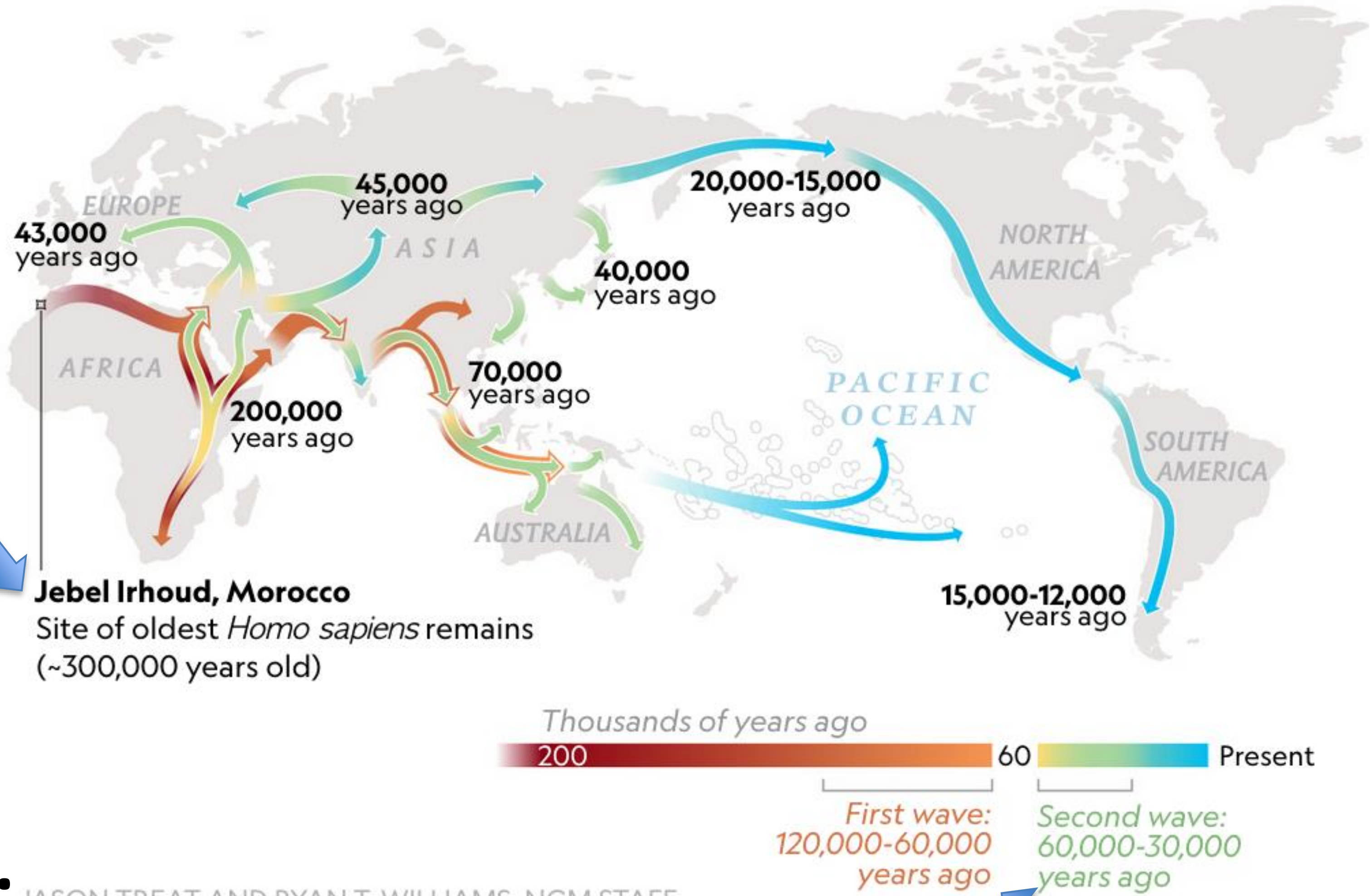


3.

Most Recent Info! 2017

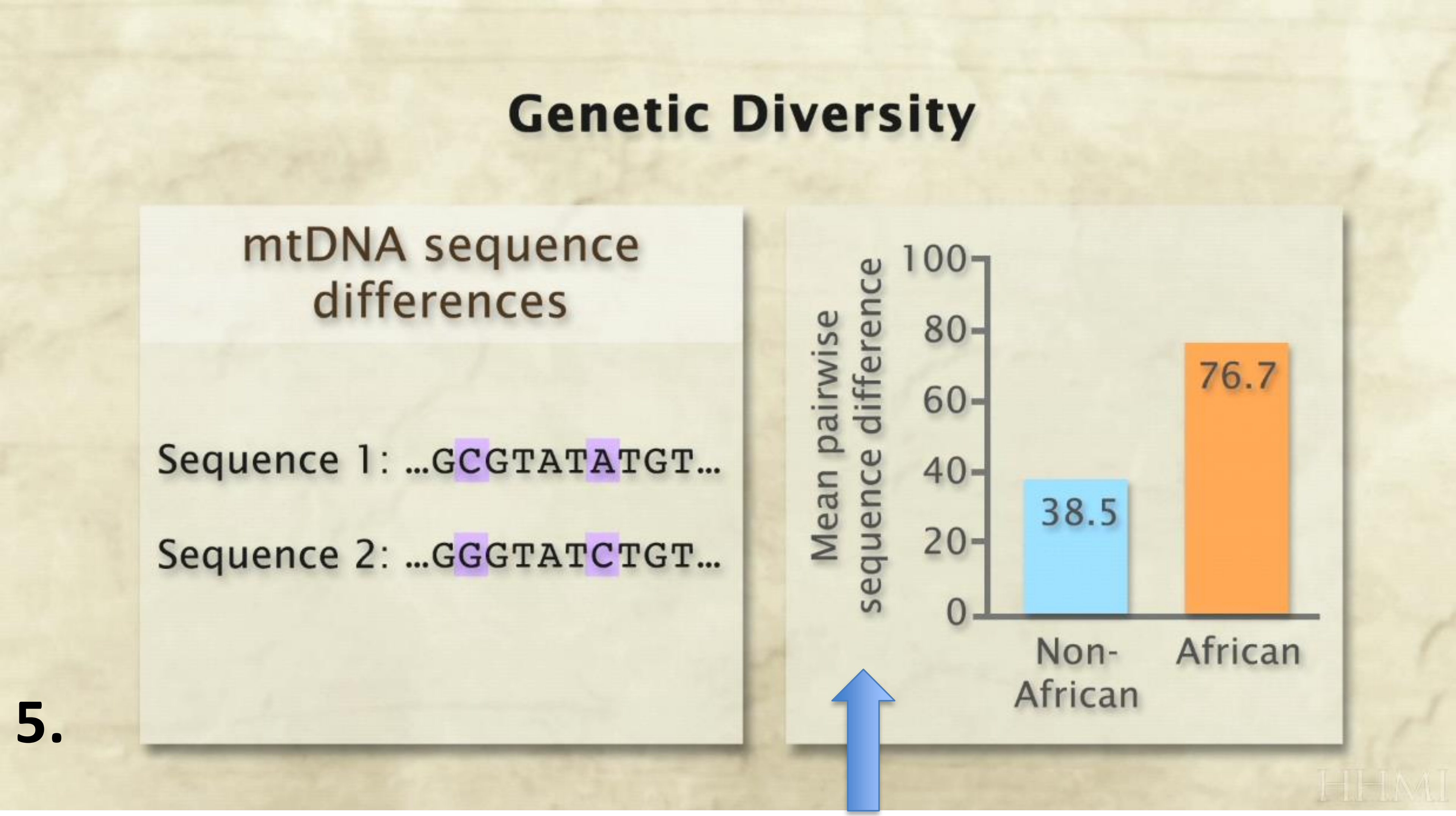
A FORMATIVE JOURNEY

As humans migrated out of Africa—in two waves, some scientists say—they adapted to new environments in many ways. Skin color is just one; high-altitude populations, for example, adapted to breathing low-oxygen air.



Jebel Irhoud, Morocco
Site of oldest *Homo sapiens* remains
(~300,000 years old)

JASON TREAT AND RYAN T. WILLIAMS, NGM STAFF
SOURCE: CHRISTOPHER BAE AND OTHERS, *SCIENCE*, 2017



Genetic Diversity

mtDNA sequence differences

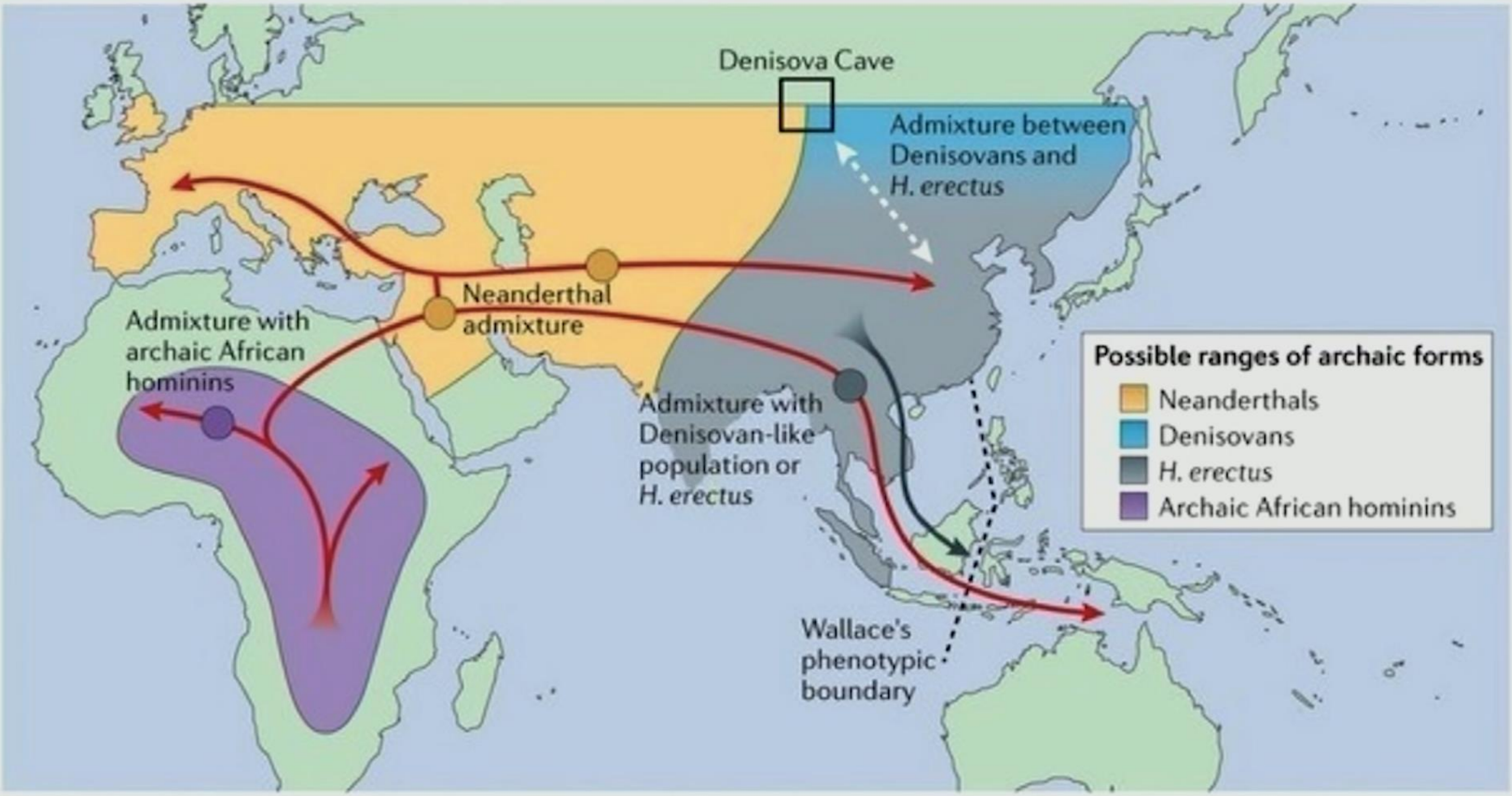
Sequence 1: ...GCGTATATGT...

Sequence 2: ...GGGTATCTGT...

5.

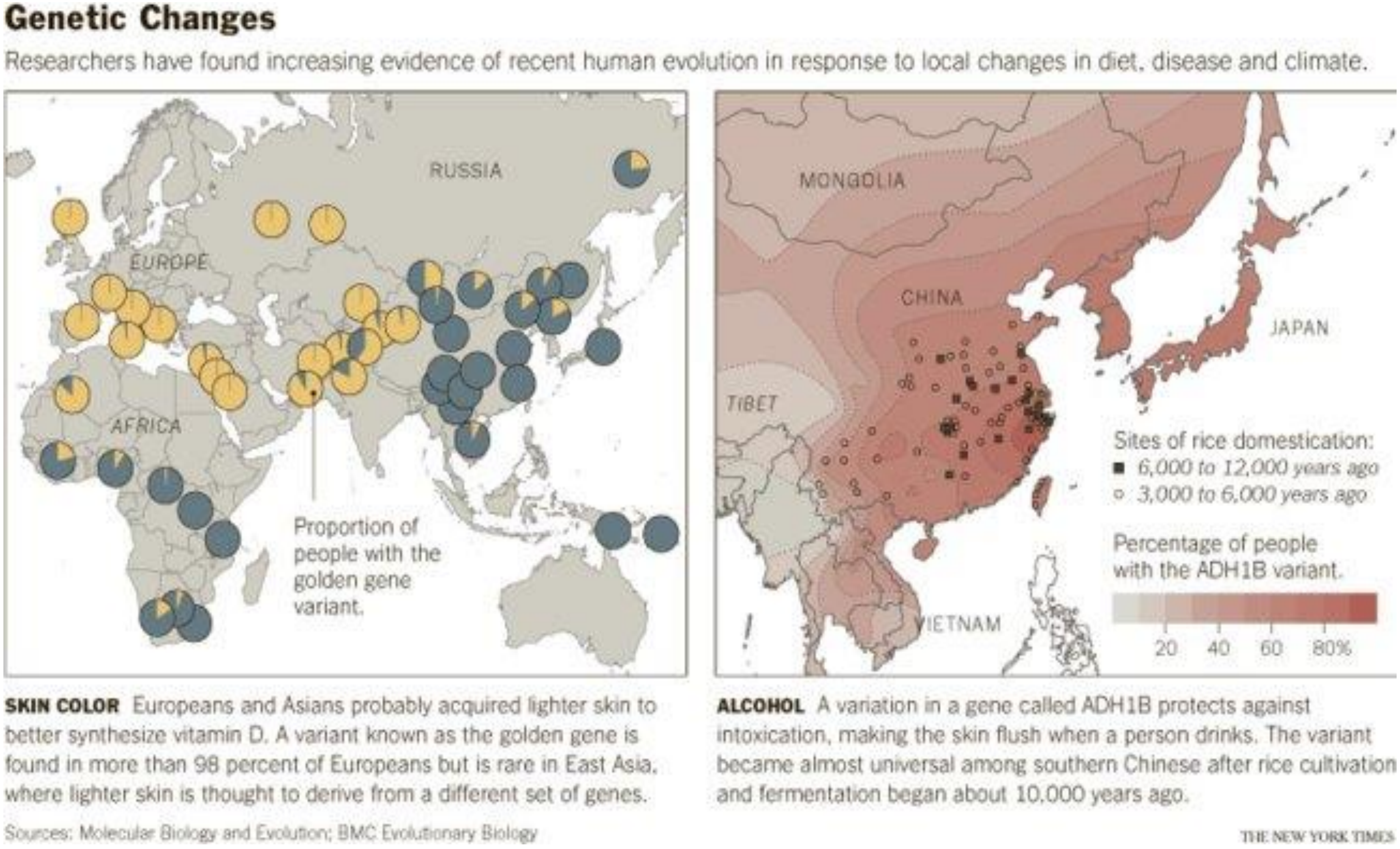
How can there be so many differences between individuals of African descent and so few differences between individuals in the rest of the entire world of “Non-African decent”?

What we need to know: Was there admixture with archaic populations in Africa?



Nature Reviews | Genetics

6.



Genetic Changes

Researchers have found increasing evidence of recent human evolution in response to local changes in diet, disease and climate.

SKIN COLOR Europeans and Asians probably acquired lighter skin to better synthesize vitamin D. A variant known as the golden gene is found in more than 98 percent of Europeans but is rare in East Asia, where lighter skin is thought to derive from a different set of genes.

ALCOHOL A variation in a gene called ADH1B protects against intoxication, making the skin flush when a person drinks. The variant became almost universal among southern Chinese after rice cultivation and fermentation began about 10,000 years ago.

Sources: Molecular Biology and Evolution; BMC Evolutionary Biology

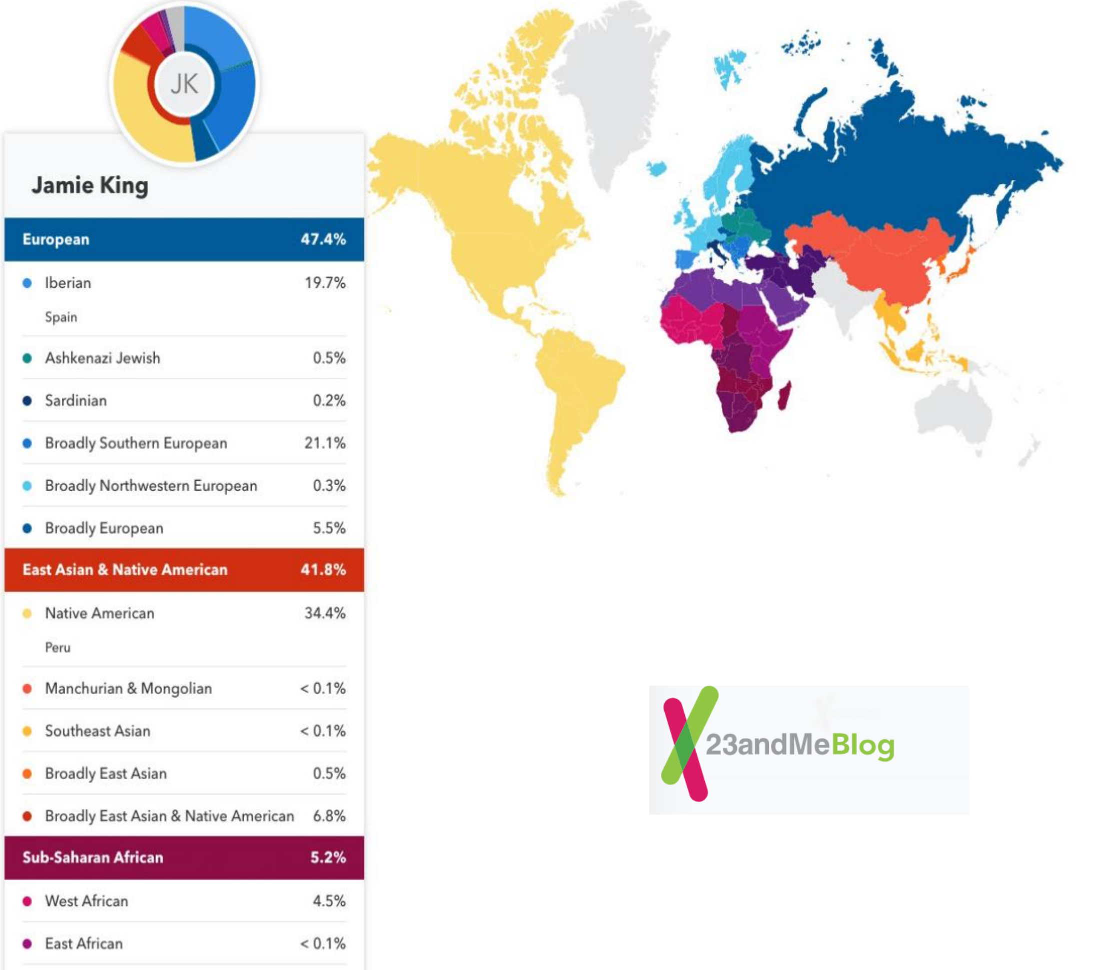
THE NEW YORK TIMES

From the article you read/will read!

7.

Ancestry Composition

Your DNA tells the story of who you are and how you're connected to populations around the world. Trace your heritage through the centuries and uncover clues about where your ancestors lived and when.



Jamie King

European	**47.4%**
● Iberian Spain	19.7%
● Ashkenazi Jewish	0.5%
● Sardinian	0.2%
● Broadly Southern European	21.1%
● Broadly Northwestern European	0.3%
● Broadly European	5.5%
East Asian & Native American	**41.8%**
● Native American Peru	34.4%
● Manchurian & Mongolian	< 0.1%
● Southeast Asian	< 0.1%
● Broadly East Asian	0.5%
● Broadly East Asian & Native American	6.8%
Sub-Saharan African	**5.2%**
● West African	4.5%
● East African	< 0.1%

23andMeBlog

8. https://www.ispot.tv/ad/wK
V/ancestrydna-kim

nature International weekly journal of science

Home | News & Comment | Research | Careers & Jobs | Current Issue | Archive | Aud

News & Comment > News > 2018 > May > Article

NATURE | NEWS

Evidence mounts for interbreeding bonanza ancient human species

***Nature* tallies the trysts among Neanderthals, humans and other relatives.**

Ewen Callaway

17 February 2016

Rights & Permissions

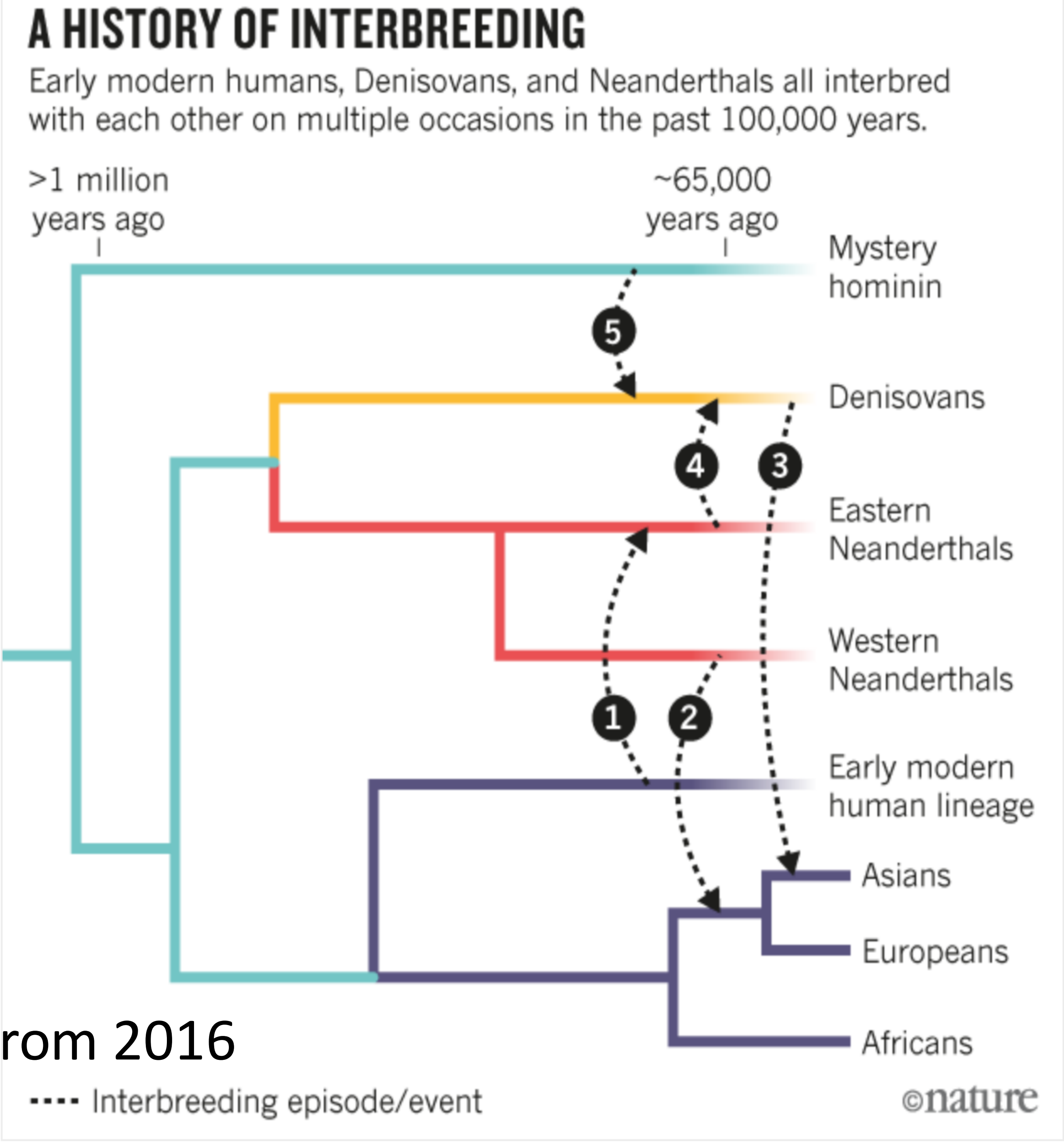
The discovery of yet another period of interbreeding between early humans and Nea adding to the growing sense that sexual encounters among different ancient human commonplace throughout their history.

"As more early modern humans and archaic humans are found and sequenced, we're going to see many more instances of interbreeding," says Sergi Castellano, a population geneticist at the Max Planck Institute for Evolutionary Anthropology in Leipzig, Germany. His team discovered the latest example, which they believe occurred around 100,000 years ago, by analysing traces of *Homo sapiens* DNA in a Neanderthal genome extracted from a toe bone found in a cave in Siberia.

Neander outside e human b

9. Inferring interbreeding or admixture from genome analysis

https://www.nature.com/news/evidence-mounts-for-interbreeding-bonanza-in-ancient-human-species-1.19394



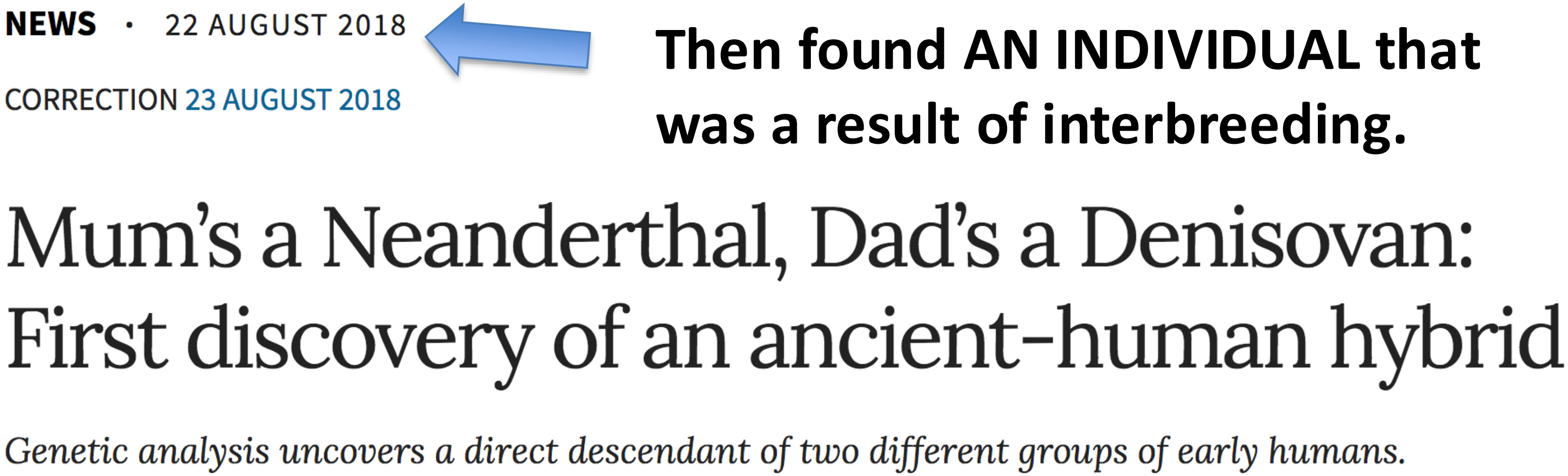
A HISTORY OF INTERBREEDING

Early modern humans, Denisovans, and Neanderthals all interbred with each other on multiple occasions in the past 100,000 years.

From 2016

Then found AN INDIVIDUAL that was a result of interbreeding.

10.



NEWS · 22 AUGUST 2018

CORRECTION 23 AUGUST 2018

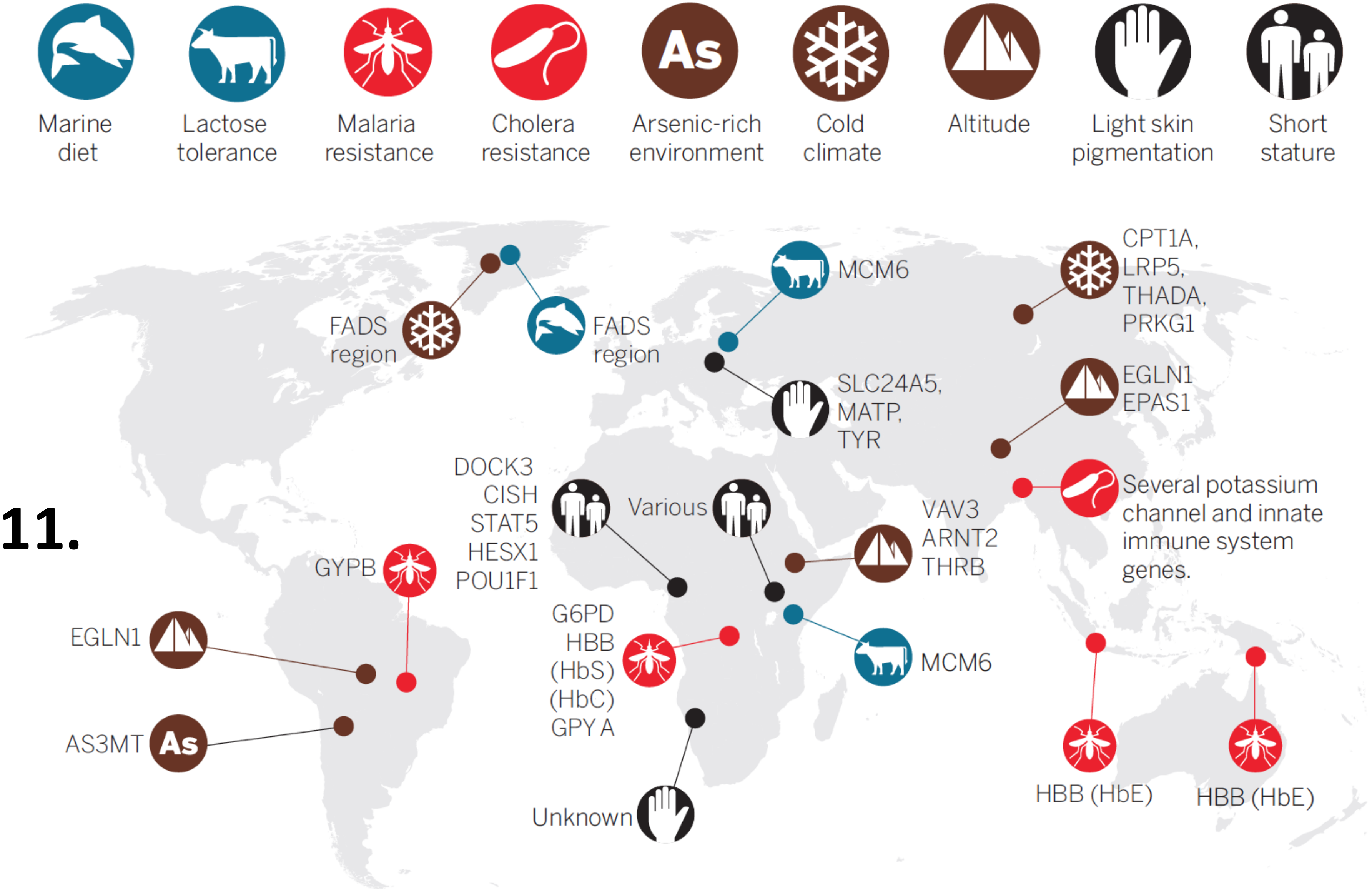
Mum's a Neanderthal, Dad's a Denisovan: First discovery of an ancient-human hybrid

Genetic analysis uncovers a direct descendant of two different groups of early humans.

A female who died around 90,000 years ago was half Neanderthal and half Denisovan, according to genome analysis of a bone discovered in a Siberian cave. This is the first time scientists have identified an ancient individual whose parents belonged to distinct human groups. The findings were published on 22 August in *Nature*[1].
"To find a first-generation person of mixed ancestry from these groups is absolutely extraordinary," says population geneticist Pontus Skoglund at the Francis Crick Institute in London. "It's really great science coupled with a little bit of luck."

The team, led by palaeogeneticists Viviane Slon and Svante Pääbo of the Max Planck Institute for Evolutionary Anthropology in Leipzig, Germany, conducted the genome analysis on a single bone fragment recovered from Denisova Cave in the Altai Mountains of Russia. This cave lends its name to the 'Denisovans', a group of extinct humans first identified on the basis of DNA sequences from the tip of a finger bone discovered[2] there in 2008. The Altai region, and the cave specifically, were also home to Neanderthals.

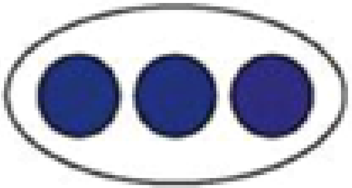
11.



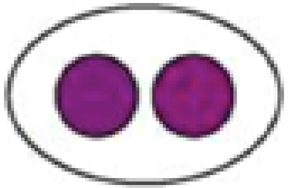
Global distribution of locally adaptive traits. Adaptation to diverse environments during human evolution has resulted in phenotypes that are at the extremes of the global distribution. Fumagalli *et al*. have integrated scans of natural selection and GWAS to identify genetic loci associated with adaptation to an Arctic environment.

More recent article than one you read/will read. This is from 2015.

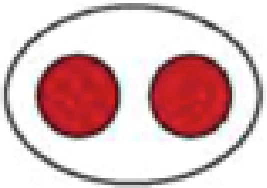
Researchers CHOSE to collect samples from people living in 3 locations and found the following (color indicates genetic similarity)



Location 1 with three subpopulations

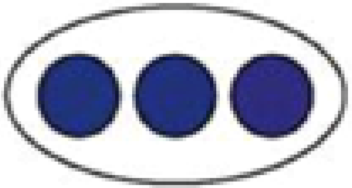


Location 2 with two subpopulations

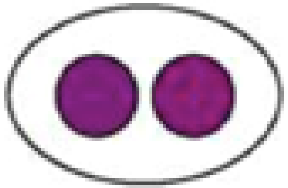


Location 3 with two subpopulations

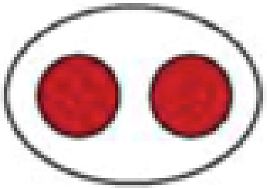
12.



Location 1 with three subpopulations



Location 2 with two subpopulations



Location 3 with two subpopulations

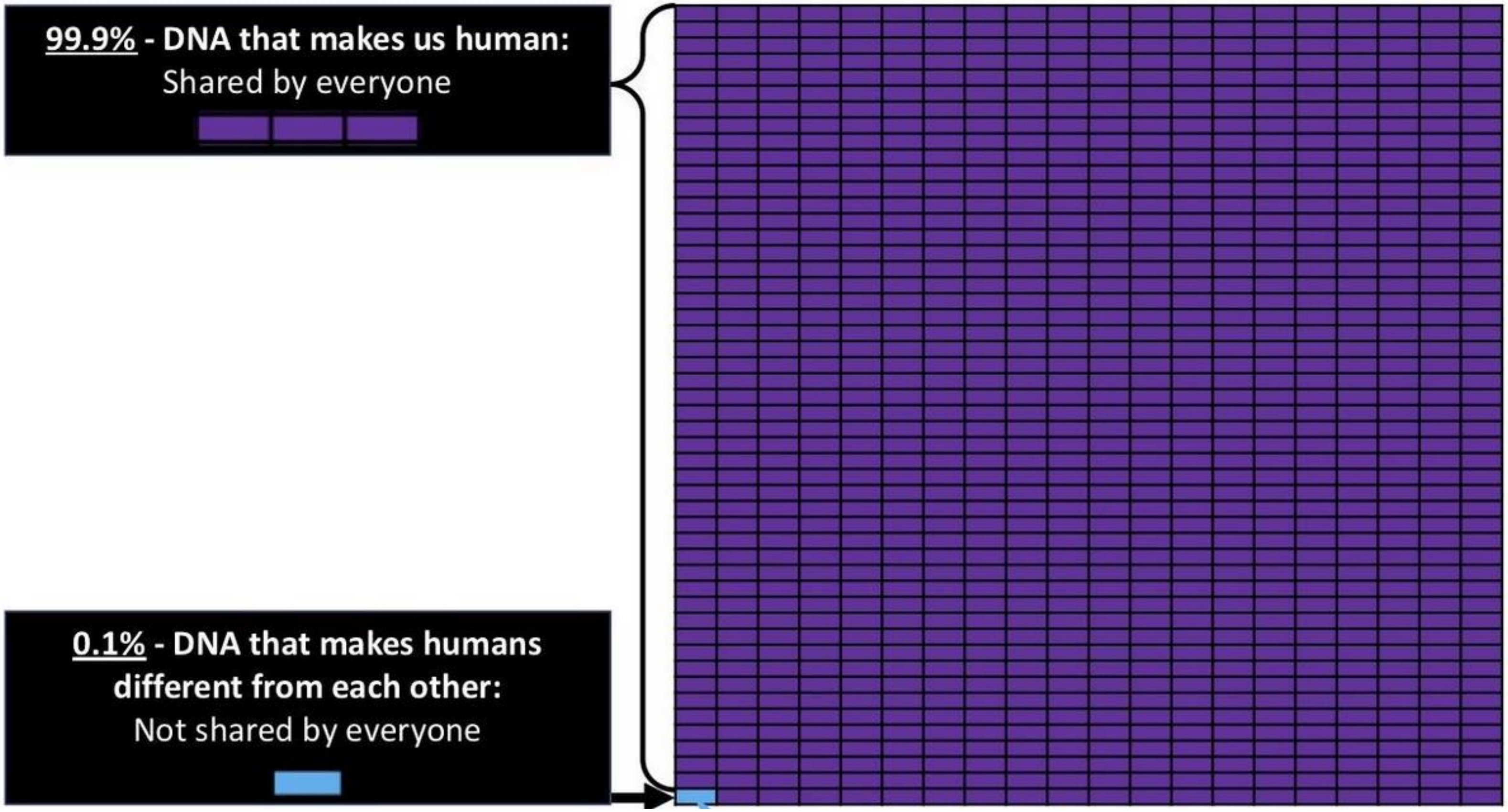
13.

Now lets add more info to the previous slide.
When researchers sampled **every population** between these three populations they found this distribution….

(Maglo et al 2016)

TRENDS in Genetics

How does this additional info affect your perspective?

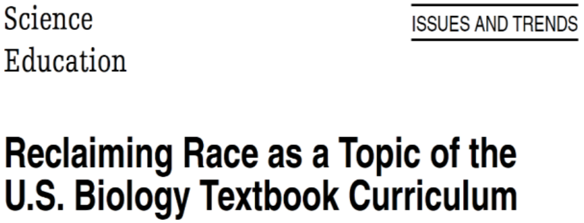


__99.9%__ - **DNA that makes us human:**
Shared by everyone
🟪🟪🟪

14.

__0.1%__ - **DNA that makes humans different from each other:**
Not shared by everyone
🟦

From Donovan 2015 and Donovan et al 2019



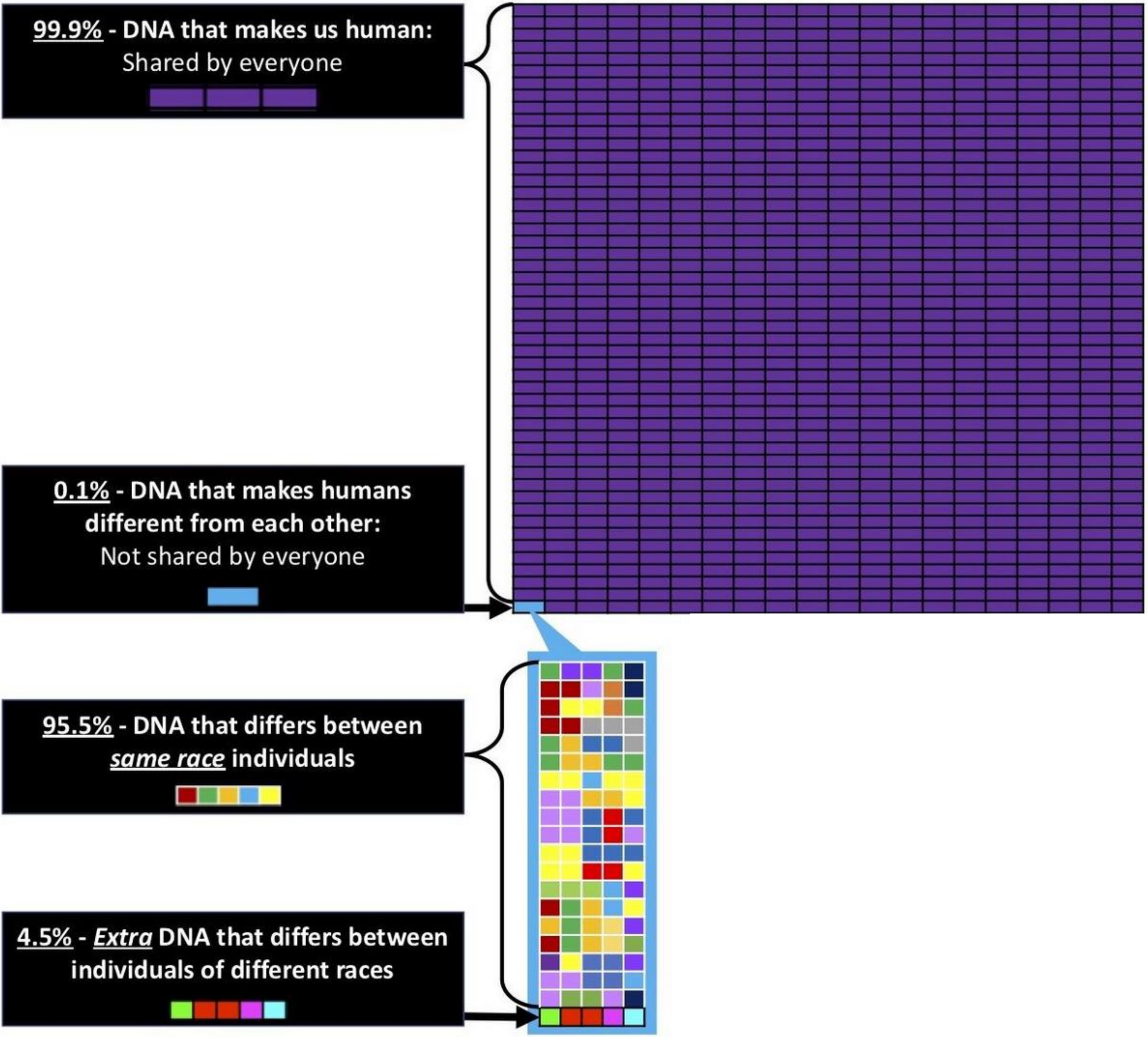
Science
Education

ISSUES AND TRENDS

Reclaiming Race as a Topic of the U.S. Biology Textbook Curriculum

Science Education. 2019;1–32.

Science Education, Vol. 99, No. 6, pp. 1092–1117 (2015)



99.9% - **DNA that makes us human:**
Shared by everyone
🟪🟪🟪

0.1% - **DNA that makes humans different from each other:**
Not shared by everyone
🟦

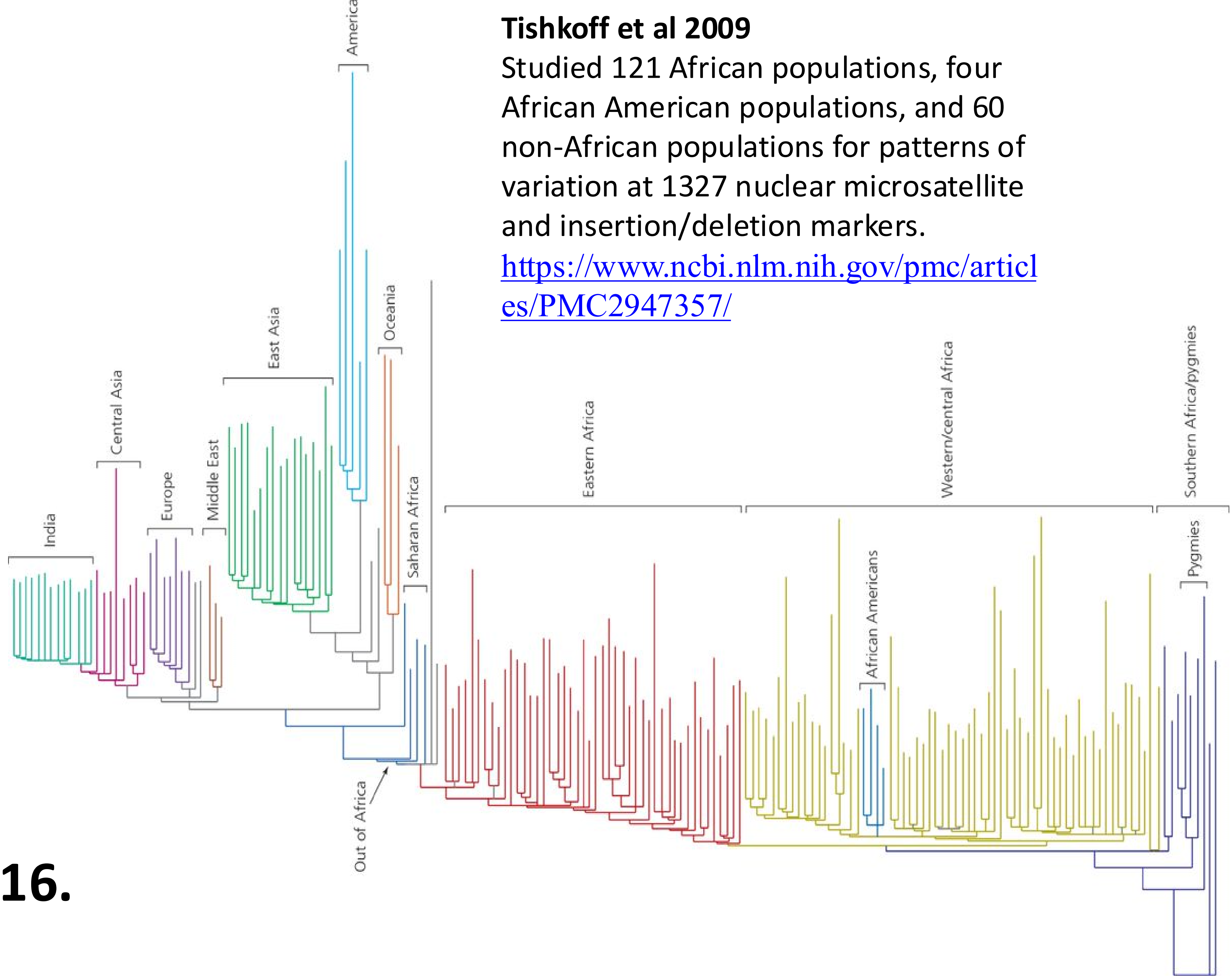
95.5% - **DNA that differs between *same race* individuals**
🟥🟩🟨🟦🟨

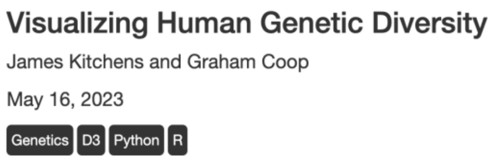
4.5% - ***Extra* DNA that differs between individuals of different races**
🟩🟥🟥🟪🟦

Tishkoff et al 2009

Studied 121 African populations, four African American populations, and 60 non-African populations for patterns of variation at 1327 nuclear microsatellite and insertion/deletion markers.

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2947357/





Visualizing Human Genetic Diversity

James Kitchens and Graham Coop

May 16, 2023

https://james-kitchens.com/blog/visualizing-human-genetic-diversity

Overlap of only
“COMMON Variants”
(so doesn’t include random people that might have a single nucleotide mutation)

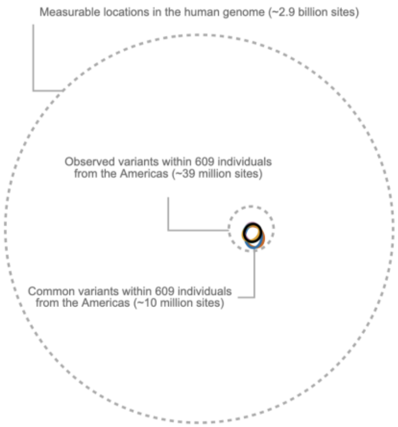


Figure 5 - Common variants in perspective. *An Euler diagram of the common variants in samples located in the Americas relative to the scale of the human genome. As a small note, the positions and orientations of ellipses within the Euler diagram differ slightly from Figure 3. This is because the eulerr package gives varied results with each run due to random starting conditions within the algorithm.*

Figure 7 - Sharing of common variation within geographic regions. *Five interactive Euler diagrams of the 26 global samples using the broad geographic groupings from Biddanda et al. 2020.*

Slides from start of class on Monday May 4!

A. Early *Homo sapiens* evolution and interaction with Neanderthals

The basics of Human Evolution!

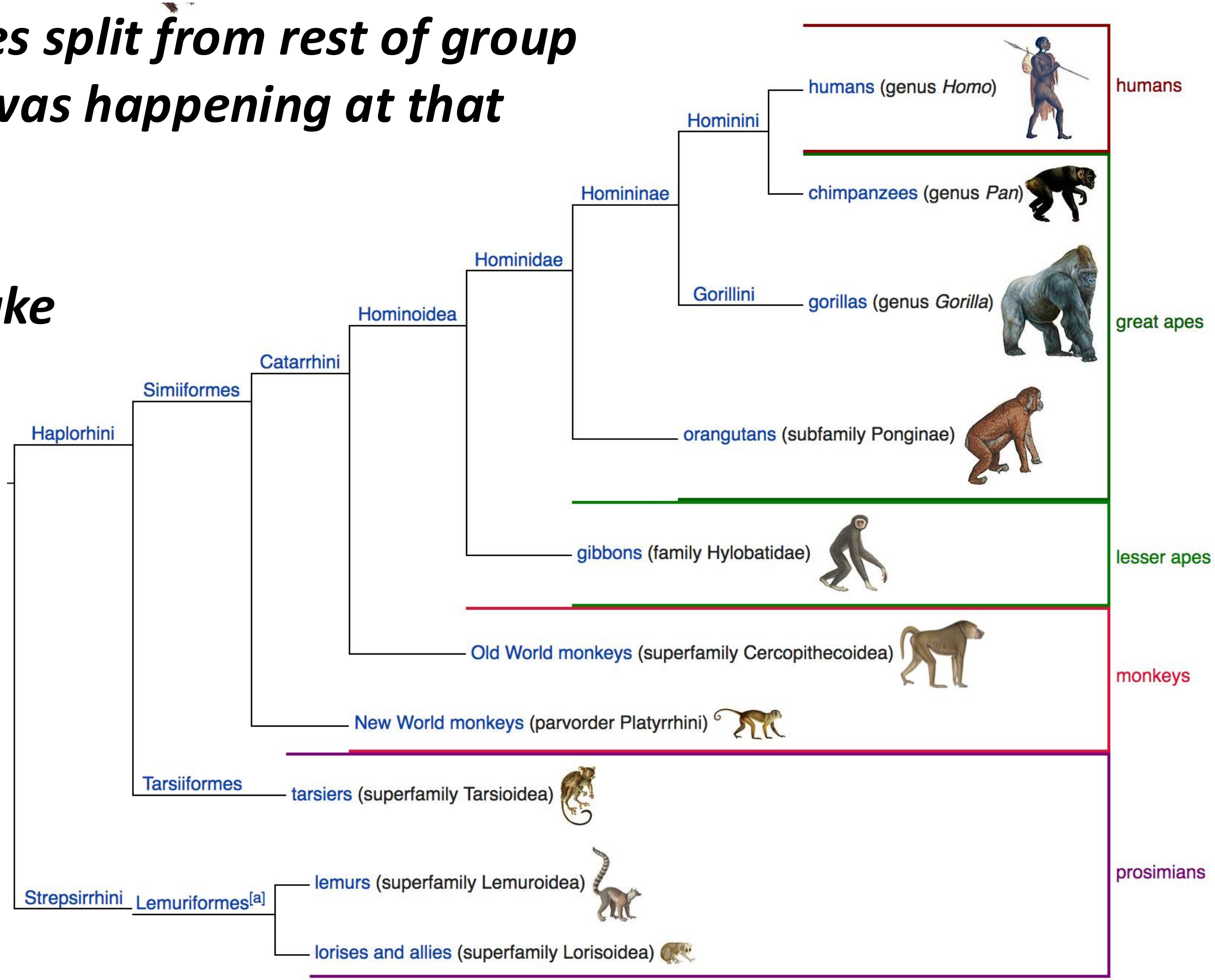
To know ourselves we need to know where we come from…how did **our genes flow through the past** to get to the present?

1. Primate Tree

Are great apes, monkeys or prosimians monophyletic groups?

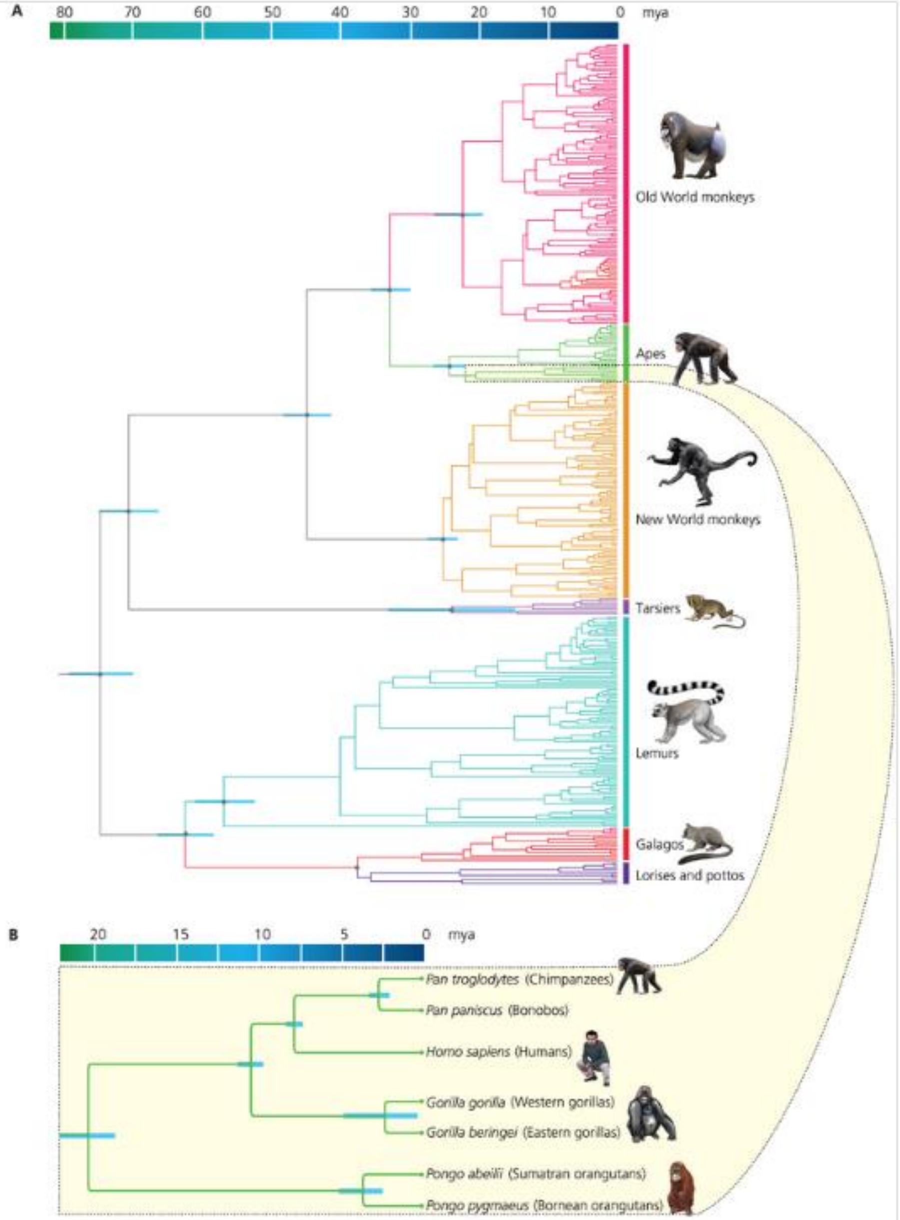
Lemurs and Lorises split from rest of group at 63mya. What was happening at that time?

How could we make this a gibbon centric tree?



Would this be considered a….

- *human centric tree*
- *lemur centric tree*
- *ape centric tree*
- *new world monkey centric tree*
- *old world monkey centric tree*



Zimmer/Emlen, *Evolution: Making Sense of Life*, 3e, © 2020 W. H. Freeman and Company

Figure 17.4 A: Scientists have gained a deep understanding of primate phylogeny through the analysis of DNA. The common ancestor of living primates lived between 70 and 79 million years ago. The blue bars at nodes in this tree represent the estimated range for their ages. (Data from Reis et al. 2018) B: Our own species is nestled deep inside the primate tree, most closely related to great apes.

2. What do we know about the dispersal of “modern humans” (*Homo sapiens*)?

Always lots of new discoveries in this area of research!

We are *Homo sapiens.*

Are there others in the Genus Homo?

Yes- several left Africa much earlier than us…

Were there other species in the genus Homo around at the same time us Homo sapiens?

Yes!

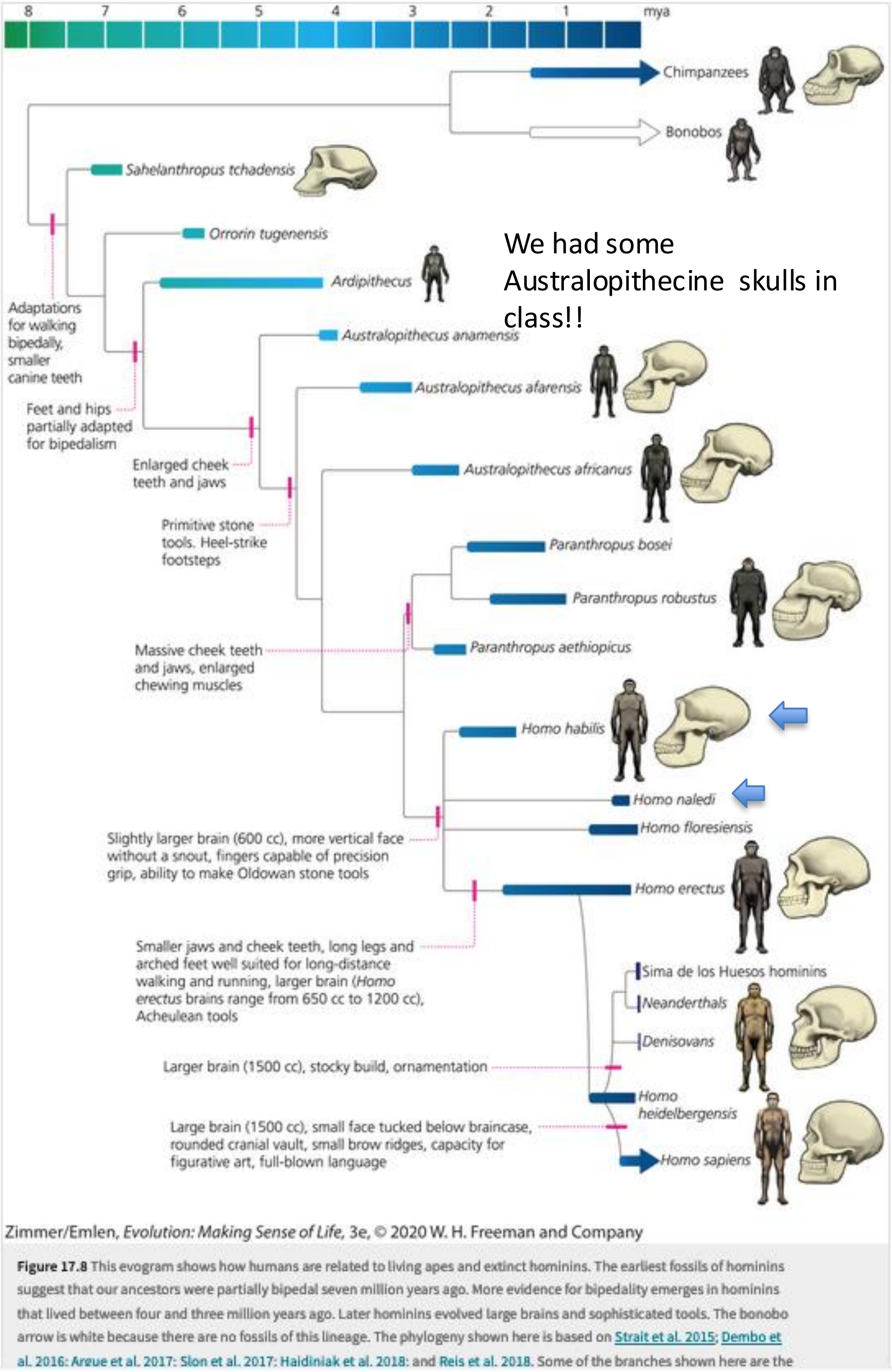
A vast cast of characters!

Is this a morphological or molecular tree?

Which Homo genera were found outside of Africa?

(<u>All</u> but the two with blue arrows! Some evolved in Africa and dispersed, others likely evolved outside of Africa.)

What do you notice about skull shapes? *(Connect to skulls in class from Monday!)*

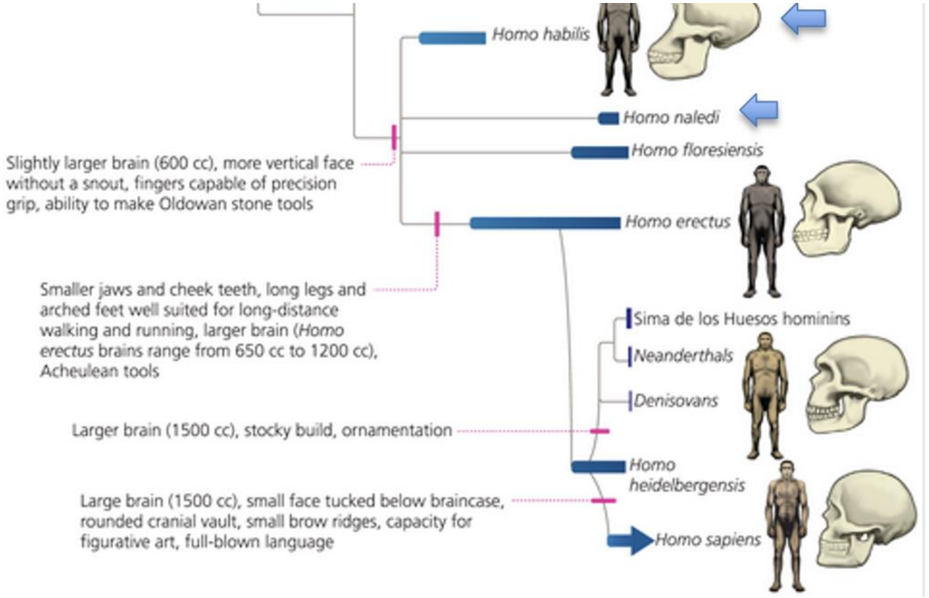


We had some Australopithecine skulls in class!!

Zimmer/Emlen, *Evolution: Making Sense of Life*, 3e, © 2020 W. H. Freeman and Company

Figure 17.8 This evogram shows how humans are related to living apes and extinct hominins. The earliest fossils of hominins suggest that our ancestors were partially bipedal seven million years ago. More evidence for bipedality emerges in hominins that lived between four and three million years ago. Later hominins evolved large brains and sophisticated tools. The bonobo arrow is white because there are no fossils of this lineage. The phylogeny shown here is based on Strait et al. 2015; Dembo et al. 2016; Argue et al. 2017; Slon et al. 2017; Hajdiniak et al. 2018; and Reis et al. 2018. Some of the branches shown here are the

Who were some of these other *Homo* species that ended up outside of Africa?



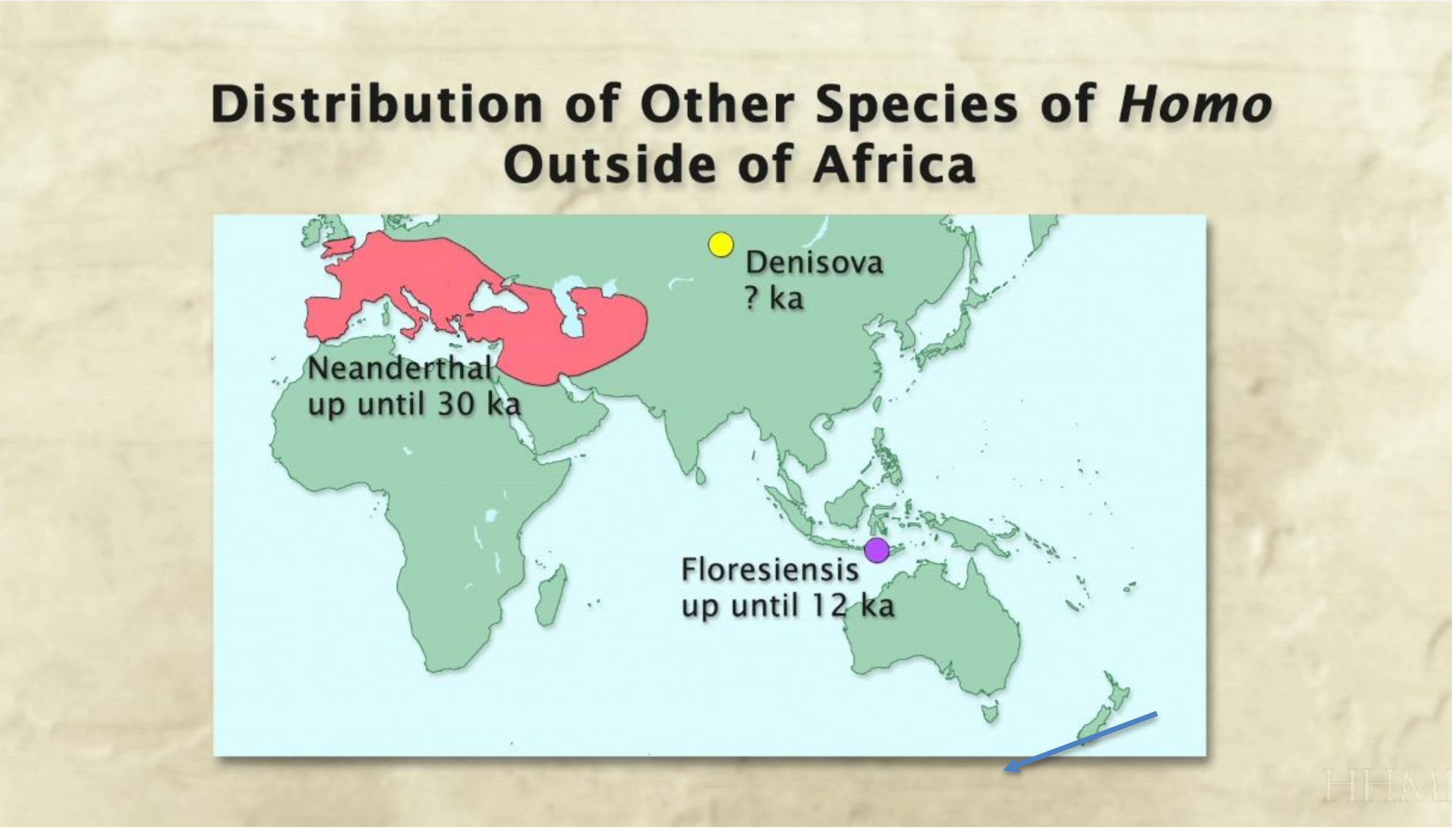
We will only focus on…

- ***Homo neanderthalensis***
- ***Homo floresiensis***
- ***Homo denisova***

For each maybe just know a little bit about them!

FYI: We are not sure where these last two actually originated.

Focus mostly on these three *Homo* species



ka = kya= thousands of years ago so 12 ka is 12,000 yrs ago.

Which lineage most recently went extinct???

Ha-I am just realizing that a paper came out last year that changed that Floresiensis extinction date to longer ago! They had some problems with their original dating techniques and now it seems to be more like extinct at 70,000 which is quite a bit longer ago. A good sign science is always correcting itself.

1. Way cool ***Homo floresiensis*** !
(a little over 3 ft tall and 60 lbs)

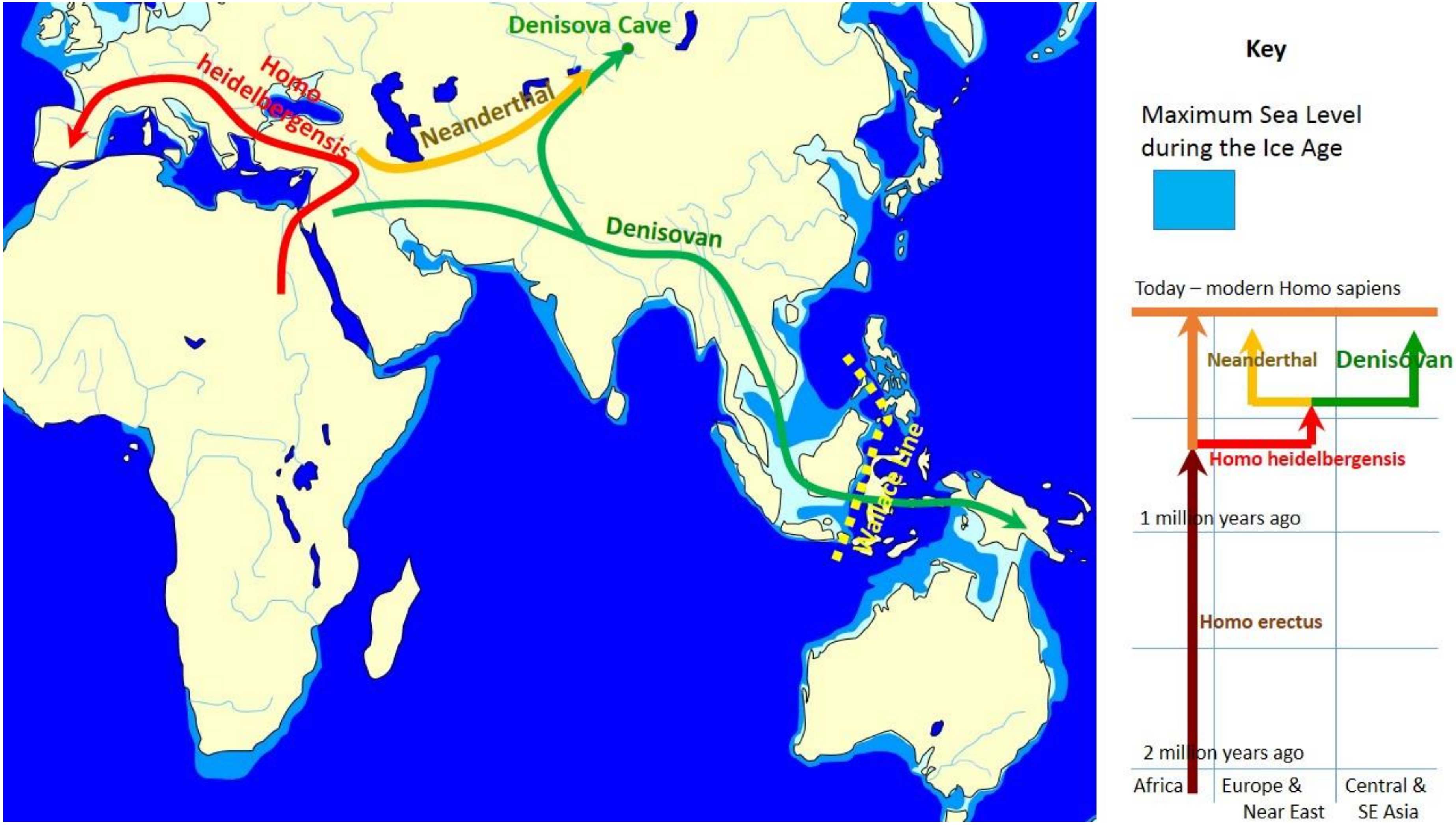
pbs.org

https://humanorigins.si.edu/evidence/human-fossils/species/homo-floresiensis

What do you think about sticking this big white European guy in this picture?

2. Mysterious Denisovans…

All we have are some bits and pieces of skeleton.



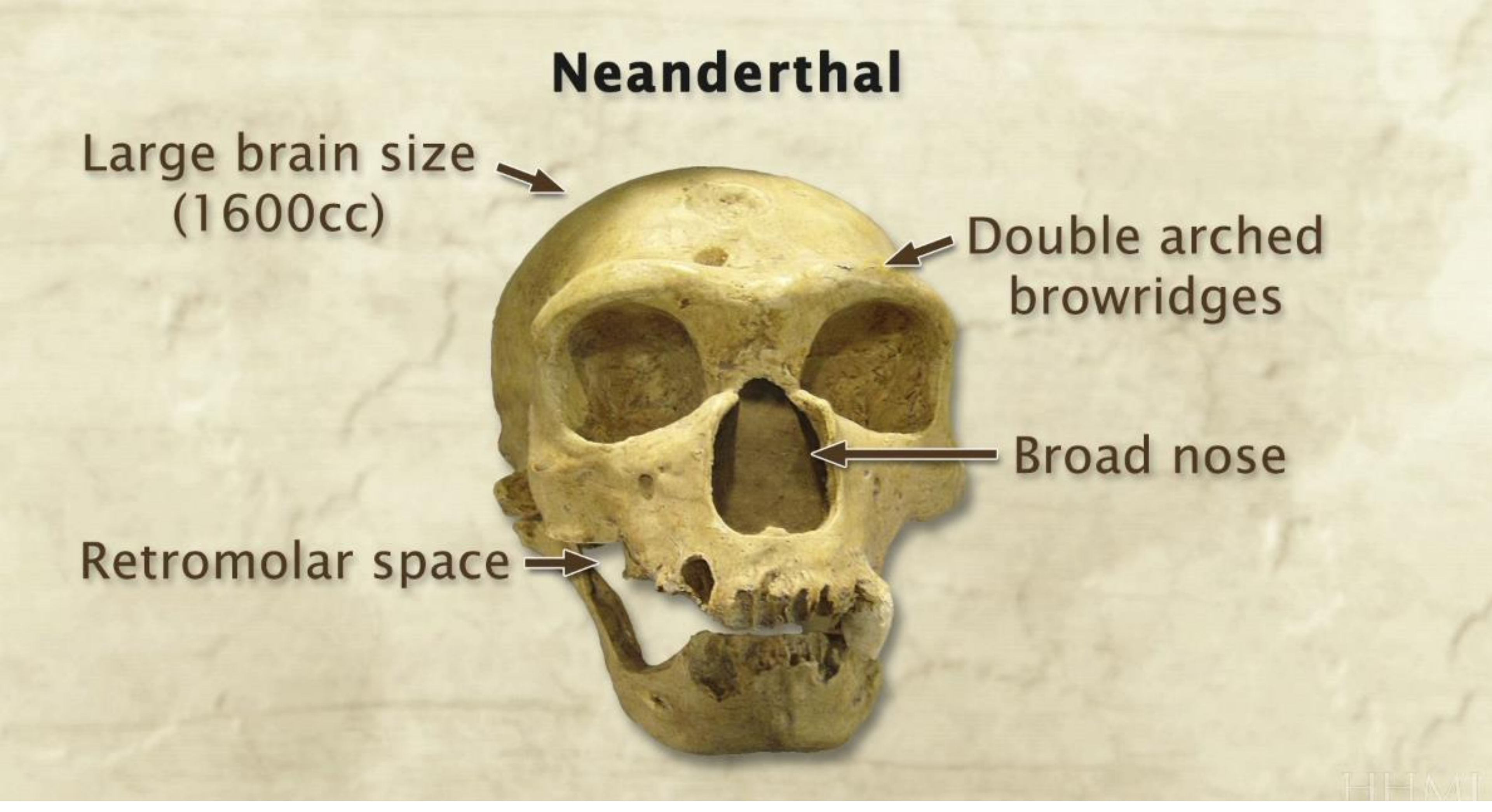
By John D. Croft at English Wikipedia, CC BY-SA 3.0, https://commons.wikimedia.org/w/index.php?curid=36145395

This artist's reconstruction, based on anatomical estimates from a new method, shows the face of a Denisovan girl from Siberia in Russia. MAAYAN HAREL

https://www.sciencemag.org/news/2019/09/ancient-dna-puts-face-mysterious-denisovans-extinct-cousins-neanderthals

3. Homo neanderthalensis

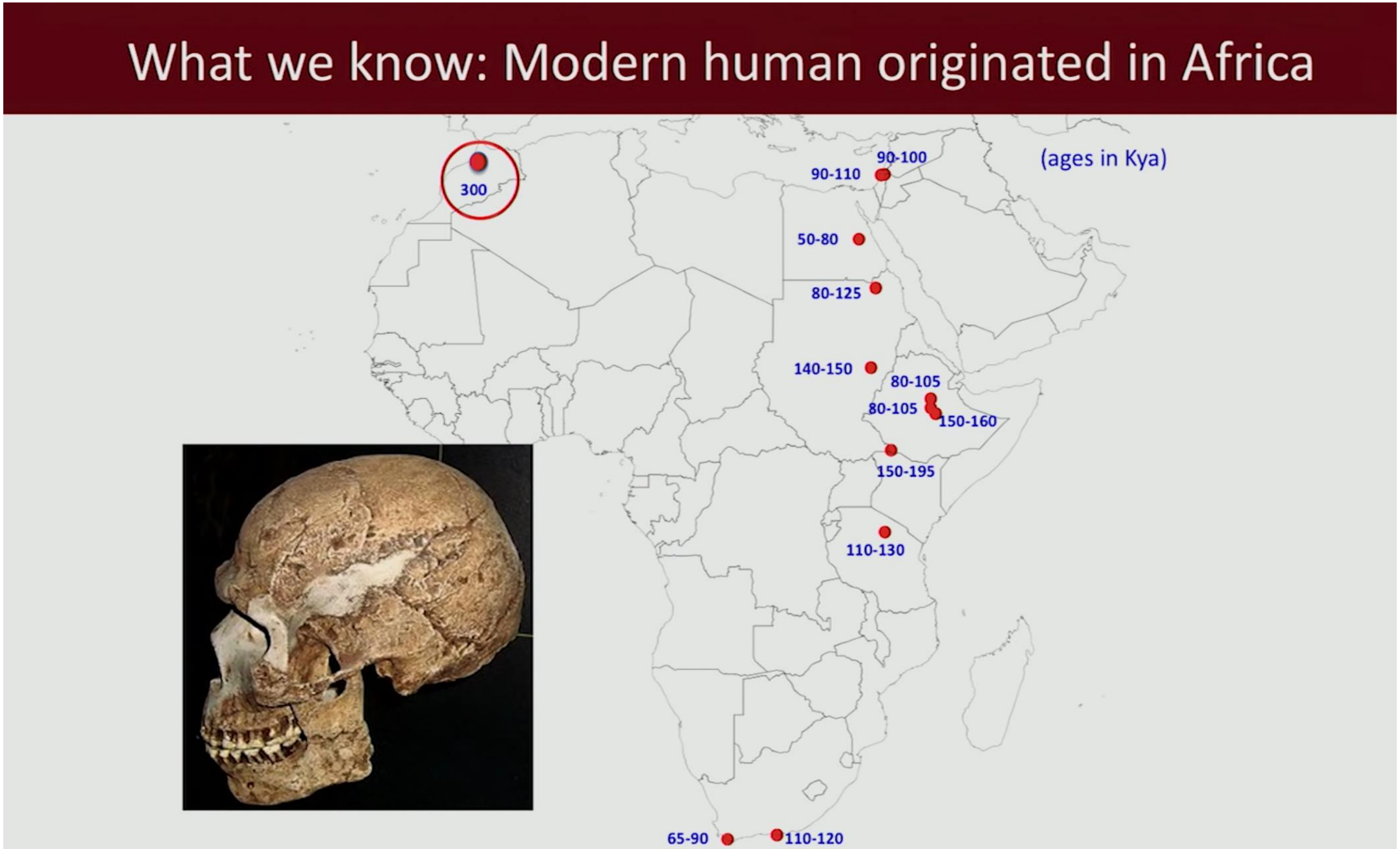
What did you learn from the videos you watched??



Neanderthal fossils have been found throughout Europe.

https://en.wikipedia.org/wiki/Neanderthal#/media/File:Carte_Neandertaliens.jpg

What about us “modern humans” or *Homo sapiens*? When did *Homo sapiens* evolve in Africa?



What we know: Modern human originated in Africa

When did *Homo sapiens* evolve in Africa?

Found in Morocco in 2017

A composite reconstruction of the earliest known Homo sapiens fossils from Jebel Irhoud in Morocco based on micro computed tomographic scans of multiple original fossils.

Philipp Gunz/Max Planck Institute for Evolutionary Anthropology

When did *Homo sapiens* evolve in Africa?

By Carl Zimmer 2017
Fossils discovered in Morocco are the oldest known remains of Homo sapiens…

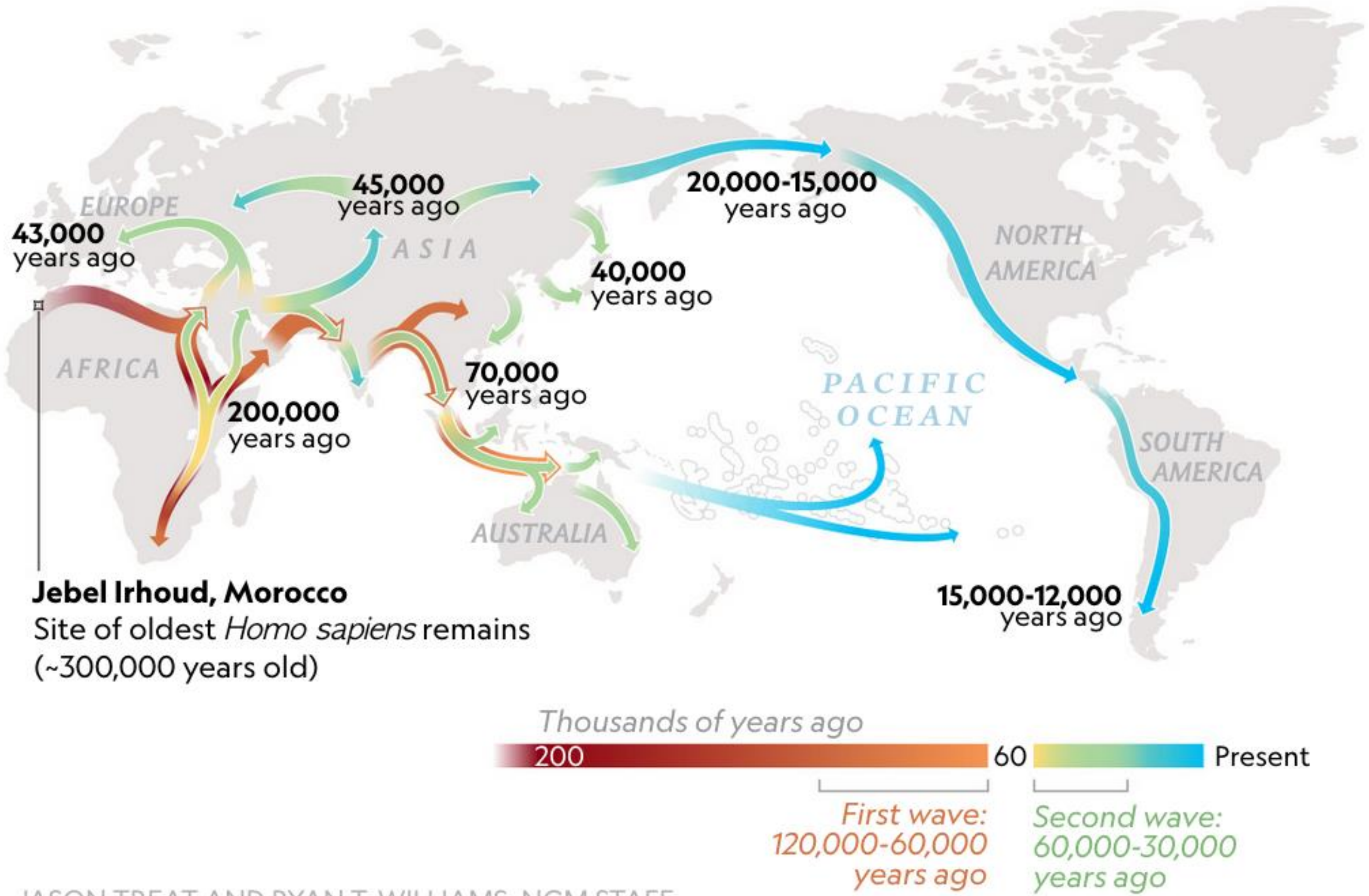
Until now, the oldest known fossils of our species dated back just 195,000 years. The Moroccan fossils, by contrast, are roughly 300,000 years old. Remarkably, they indicate that early *Homo sapiens* **had faces much like our own**, although their brains differed in fundamental ways.
https://www.nytimes.com/2017/06/07/science/human-fossils-morocco.html

So now we think *Homo sapiens* evolved in Africa a lot longer ago than we thought!

When did *Homo sapiens leave* in Africa?

A FORMATIVE JOURNEY

As humans migrated out of Africa—in two waves, some scientists say—they adapted to new environments in many ways. Skin color is just one; high-altitude populations, for example, adapted to breathing low-oxygen air.



Jebel Irhoud, Morocco
Site of oldest *Homo sapiens* remains
(~300,000 years old)

*First wave:
120,000-60,000
years ago*

*Second wave:
60,000-30,000
years ago*

JASON TREAT AND RYAN T. WILLIAMS, NGM STAFF
SOURCE: CHRISTOPHER BAE AND OTHERS, *SCIENCE*, 2017

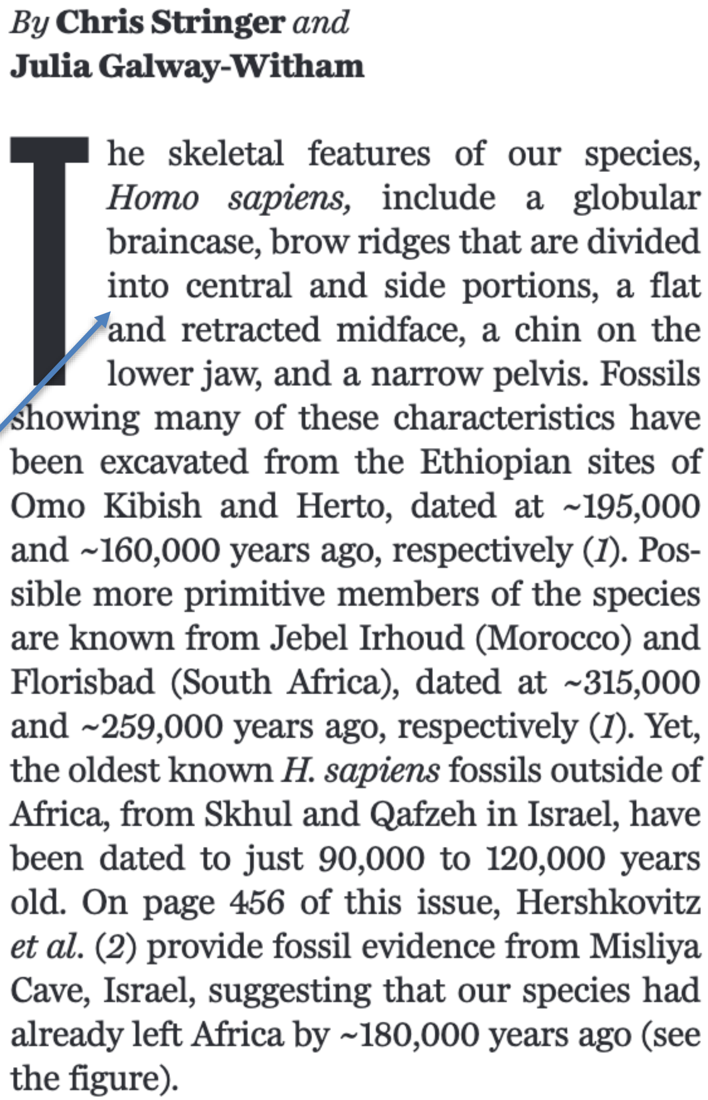
When did *Homo sapiens leave* in Africa?
More recent discoveries!

ANTHROPOLOGY

When did modern humans leave Africa?

A ~180,000-year-old fossil from Israel provides evidence for early forays of *Homo sapiens* into western Asia

26 JANUARY 2018 • VOL 359 ISSUE 6374



By **Chris Stringer** *and* **Julia Galway-Witham**

The skeletal features of our species, *Homo sapiens,* include a globular braincase, brow ridges that are divided into central and side portions, a flat and retracted midface, a chin on the lower jaw, and a narrow pelvis. Fossils showing many of these characteristics have been excavated from the Ethiopian sites of Omo Kibish and Herto, dated at ~195,000 and ~160,000 years ago, respectively (*1*). Possible more primitive members of the species are known from Jebel Irhoud (Morocco) and Florisbad (South Africa), dated at ~315,000 and ~259,000 years ago, respectively (*1*). Yet, the oldest known *H. sapiens* fossils outside of Africa, from Skhul and Qafzeh in Israel, have been dated to just 90,000 to 120,000 years old. On page 456 of this issue, Hershkovitz *et al.* (*2*) provide fossil evidence from Misliya Cave, Israel, suggesting that our species had already left Africa by ~180,000 years ago (see the figure).

Note *Homo sapiens* traits they list…

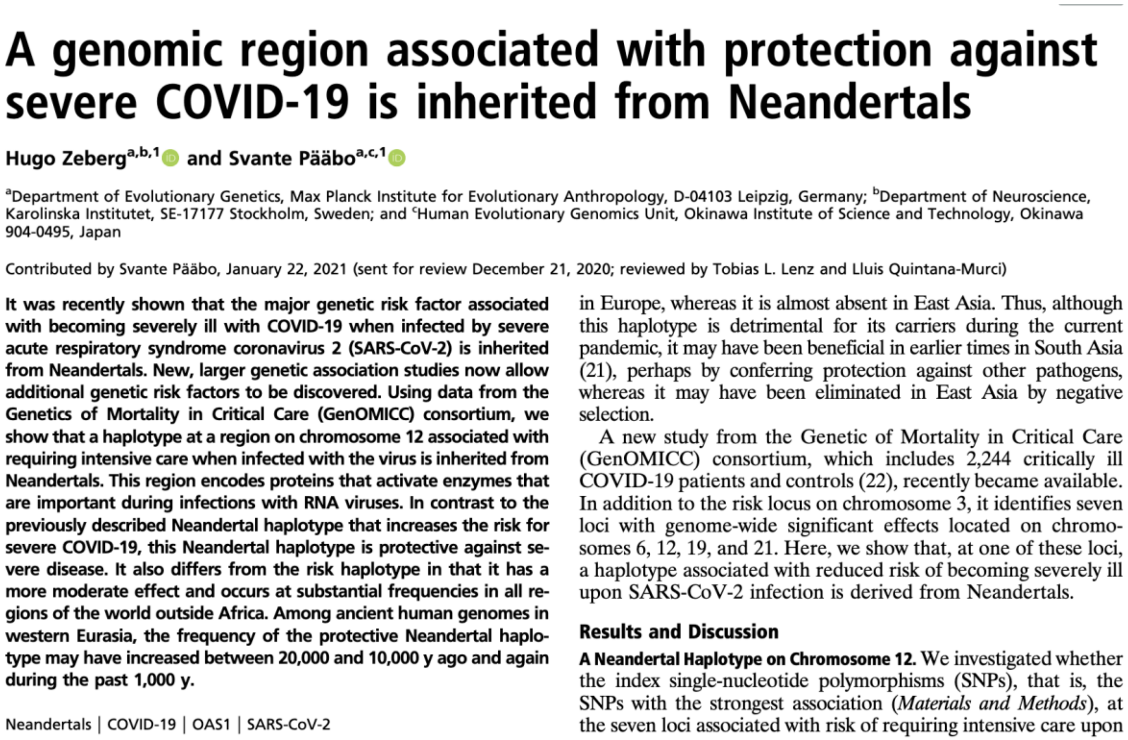
When *Homo sapiens* left Africa there were a total of at least 5 other *Homo* “species” out there!

Homo heidelbergensis
Homo erectus
*Homo neanderthalensis**
*Homo floresiensis**
*Homo denisova**

*species to know a bit about

A couple more slides that were on your “Gallery Walk”!

And this!!



A genomic region associated with protection against severe COVID-19 is inherited from Neandertals

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Contributed by Svante Pääbo, January 22, 2021 (sent for review December 21, 2020; reviewed by Tobias L. Lenz and Lluis Quintana-Murci)

It was recently shown that the major genetic risk factor associated with becoming severely ill with COVID-19 when infected by severe acute respiratory syndrome coronavirus 2 (SARS-CoV-2) is inherited from Neandertals. New, larger genetic association studies now allow additional genetic risk factors to be discovered. Using data from the Genetics of Mortality in Critical Care (GenOMICC) consortium, we show that a haplotype at a region on chromosome 12 associated with requiring intensive care when infected with the virus is inherited from Neandertals. This region encodes proteins that activate enzymes that are important during infections with RNA viruses. In contrast to the previously described Neandertal haplotype that increases the risk for severe COVID-19, this Neandertal haplotype is protective against severe disease. It also differs from the risk haplotype in that it has a more moderate effect and occurs at substantial frequencies in all regions of the world outside Africa. Among ancient human genomes in western Eurasia, the frequency of the protective Neandertal haplotype may have increased between 20,000 and 10,000 y ago and again during the past 1,000 y.

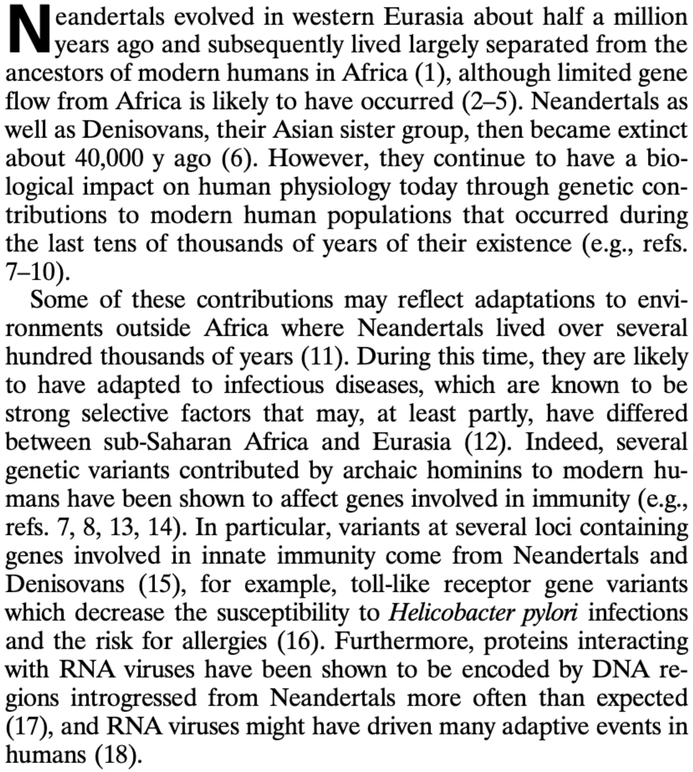
Neandertals | COVID-19 | OAS1 | SARS-CoV-2

in Europe, whereas it is almost absent in East Asia. Thus, although this haplotype is detrimental for its carriers during the current pandemic, it may have been beneficial in earlier times in South Asia (21), perhaps by conferring protection against other pathogens, whereas it may have been eliminated in East Asia by negative selection.

A new study from the Genetic of Mortality in Critical Care (GenOMICC) consortium, which includes 2,244 critically ill COVID-19 patients and controls (22), recently became available. In addition to the risk locus on chromosome 3, it identifies seven loci with genome-wide significant effects located on chromosomes 6, 12, 19, and 21. Here, we show that, at one of these loci, a haplotype associated with reduced risk of becoming severely ill upon SARS-CoV-2 infection is derived from Neandertals.

Results and Discussion

A Neandertal Haplotype on Chromosome 12. We investigated whether the index single-nucleotide polymorphisms (SNPs), that is, the SNPs with the strongest association (*Materials and Methods*), at the seven loci associated with risk of requiring intensive care upon



Neandertals evolved in western Eurasia about half a million years ago and subsequently lived largely separated from the ancestors of modern humans in Africa (1), although limited gene flow from Africa is likely to have occurred (2–5). Neandertals as well as Denisovans, their Asian sister group, then became extinct about 40,000 y ago (6). However, they continue to have a biological impact on human physiology today through genetic contributions to modern human populations that occurred during the last tens of thousands of years of their existence (e.g., refs. 7–10).

Some of these contributions may reflect adaptations to environments outside Africa where Neandertals lived over several hundred thousands of years (11). During this time, they are likely to have adapted to infectious diseases, which are known to be strong selective factors that may, at least partly, have differed between sub-Saharan Africa and Eurasia (12). Indeed, several genetic variants contributed by archaic hominins to modern humans have been shown to affect genes involved in immunity (e.g., refs. 7, 8, 13, 14). In particular, variants at several loci containing genes involved in innate immunity come from Neandertals and Denisovans (15), for example, toll-like receptor gene variants which decrease the susceptibility to *Helicobacter pylori* infections and the risk for allergies (16). Furthermore, proteins interacting with RNA viruses have been shown to be encoded by DNA regions introgressed from Neandertals more often than expected (17), and RNA viruses might have driven many adaptive events in humans (18).

PNAS 2021 Vol. 118 No. 9 e2026309118

Understanding Us: What Makes Difference and What Difference Does Difference Make

A. Early *Homo sapiens* evolution and interaction with Neanderthals

- When *Homo sapiens* began to disperse from Africa there were 5 other *Homo* genera out there in the world that they might have bumped into.
- Each wave that left Africa probably interbred or admixed with other groups of *Homo* humans that had left Africa earlier.

From Wikipedia…and how do we define **species**????

Ever since the discovery of the Neanderthal fossils, expert opinion has been divided as to whether Neanderthals should be considered a separate species (*Homo neanderthalensis*) or a subspecies (*Homo sapiens neanderthalensis*) relative to modern humans.[26][27][28] Pääbo (2014) described such "taxonomic wars" as unresolvable in principle, "since there is no definition of species perfectly describing the case."[4]