

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

Graphical Analysis

Same or Different?

Why do this?

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

NOTE:

1=we are all quite the SAME

10=we are all quite DIFFERENT

(from How Neanderthals Gave us Secret Powers)

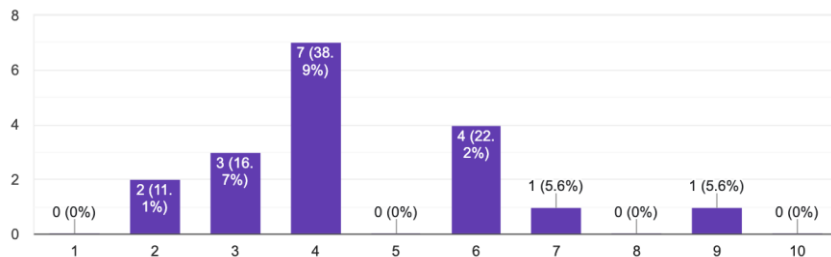
Neanderthal

Skin pigmentation,
Immune system

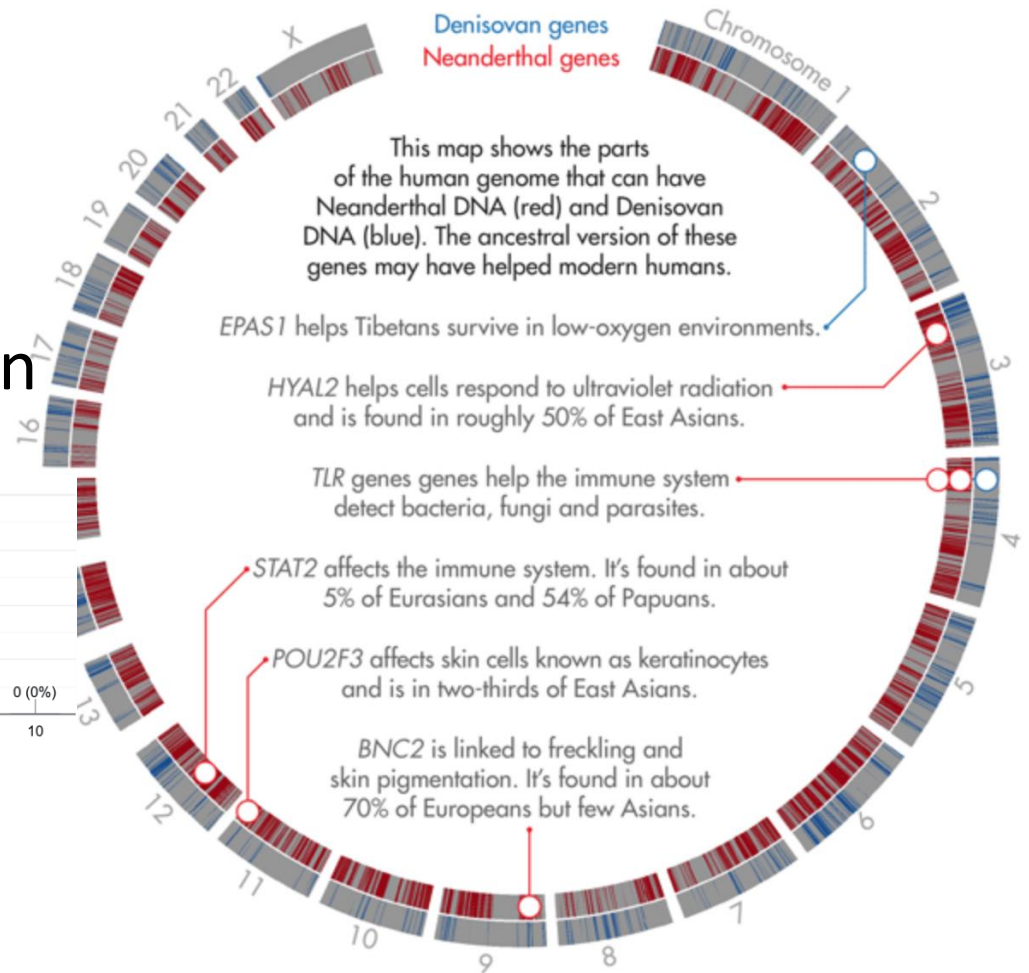
Allergies

Denisovan

Tolerance to low oxygen

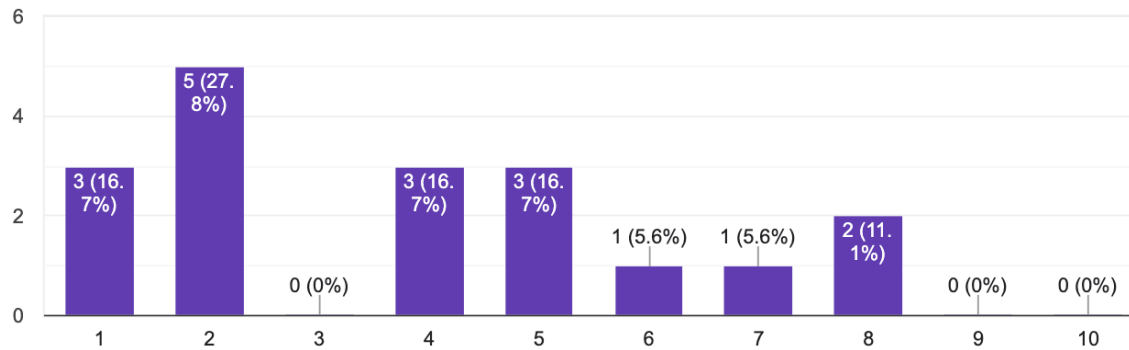


A MAP OF ANCIENT GENES



1.

On page 52, in the third paragraph it says that although ancient groups such as neanderthals and denisovans may have competed and made war, genetic evidence makes it clear that they also reproduced with each other, as can be seen by the combination of neanderthal and denisovan genes that are still a part of the human genome today.



2.

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

p. 43: "...about 90 percent of your DNA is NOT related to encoding your proteins"; this connects to the image because all of the variation in the photos seems to be related to gene expression rather than the composition of DNA and actual variation of genetic material.

49

"As human populations established themselves in new regions, they also began to accumulate new mutations. Some of these mutations were beneficial and increased until fixation in these populations."

As people spread out from Africa, they started to accumulate different adaptations that changed their phenotype, just like in the image where there are a variety of different phenotypes displayed.

p. 43 - "Recent advances in human genetics have shown us just how similar all humans are genetically" Varying facial traits and skin color are smaller mutations that are more circumstantial. These genes make up only a small percentage of the genome. The image is misleading because it focuses on differences in face and skin color, which does not represent the actual lack of diversity within our population.

Page 57, "However it is clear that whatever the methods used, no group within our species can be considered a biological race."

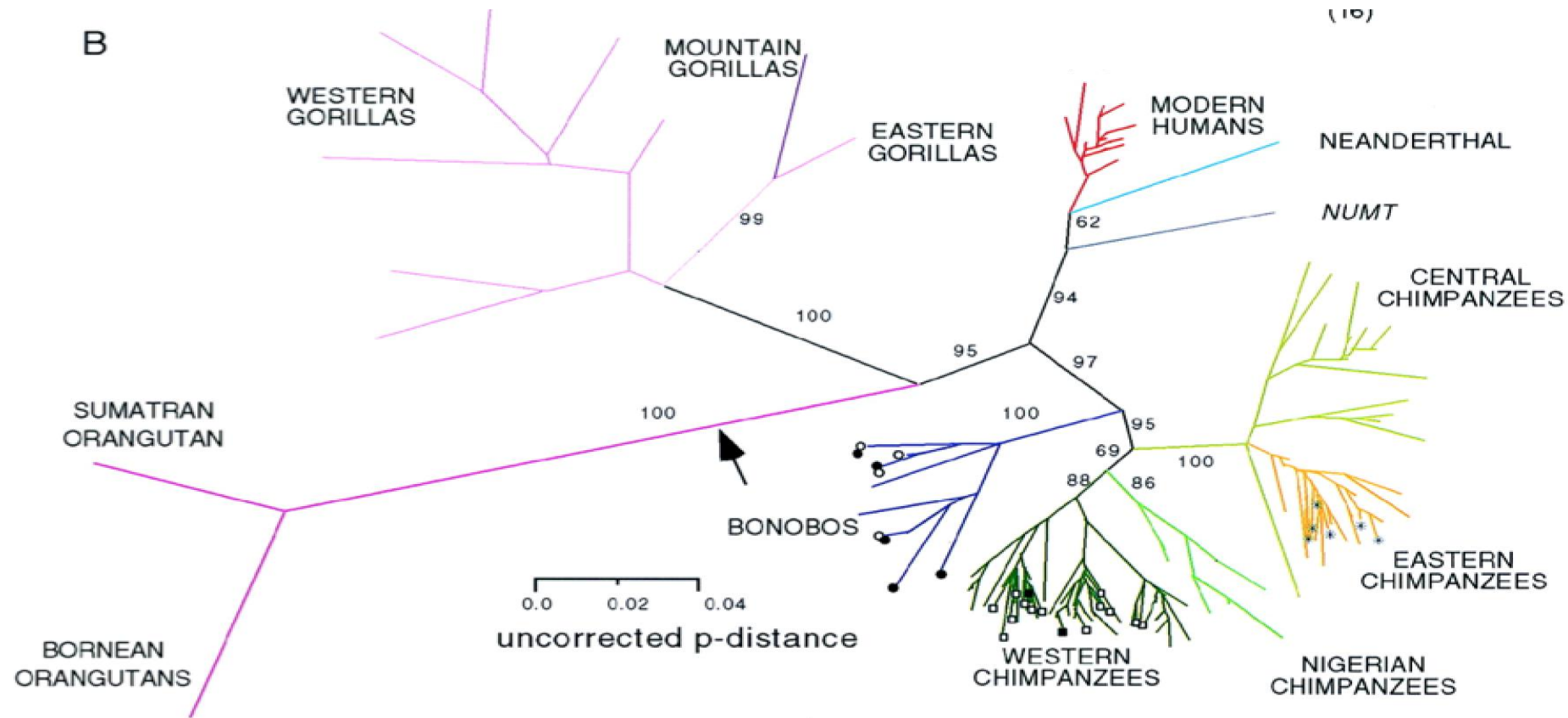
I think that this picture is trying to get people to try and sort the people in the pictures by groups but in reality I think that this is just an even better example or reminder that there is not biological race

Page 57. "As there are around 25,000 protein-coding genes in the human genome only 0.03 percent of the genome accounts for these apparent physical differences." We are very genetically similar, with the very large majority of genes coding the same stuff, and only a small amount of genes account for the diversity we see in this image.

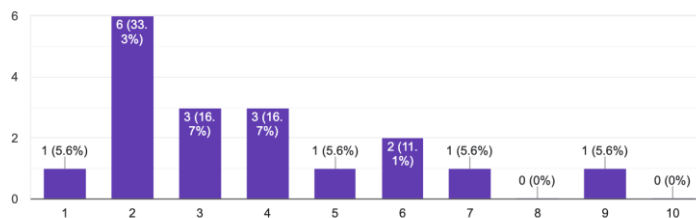
Segment length is proportional to genetic difference.

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

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



3.



PNAS

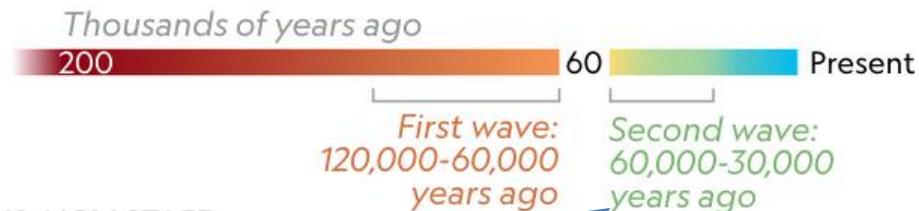
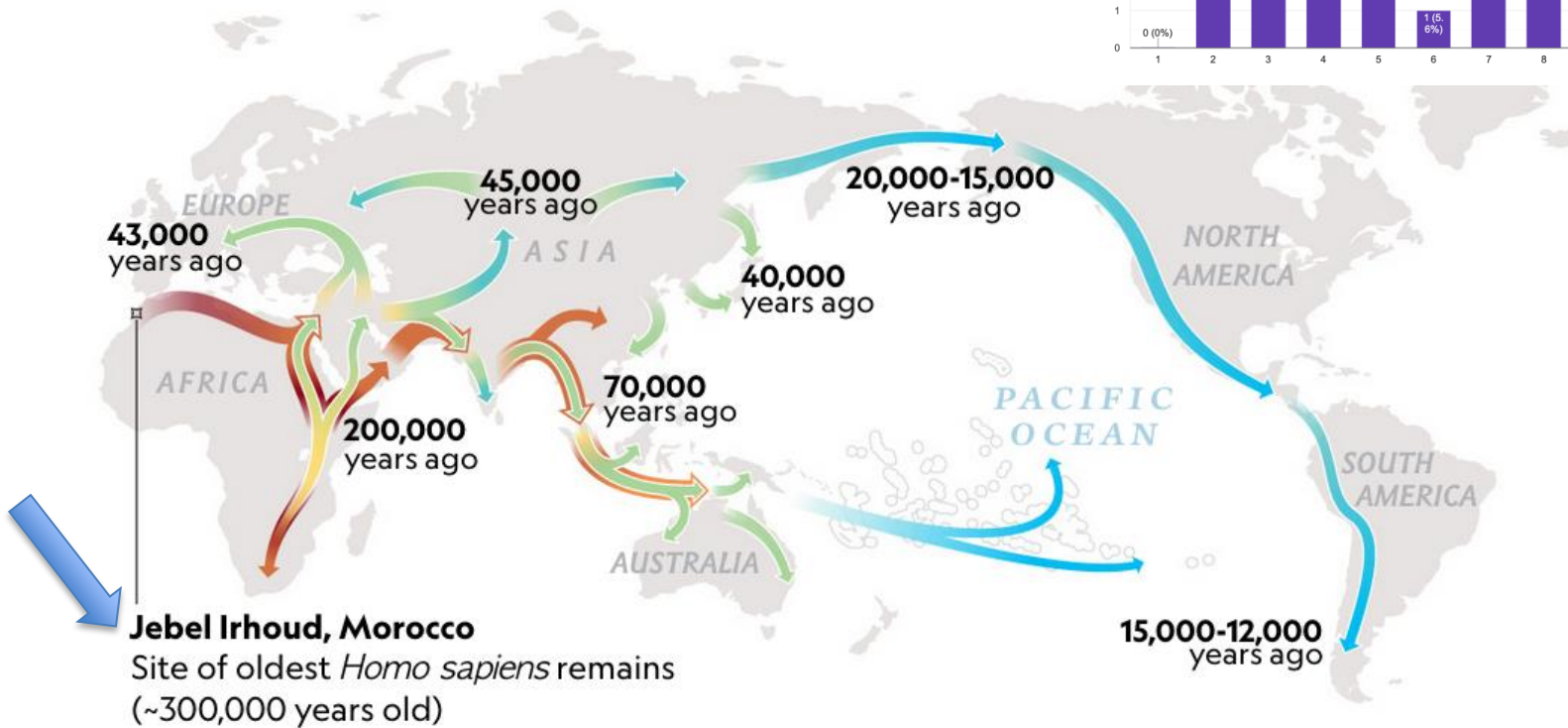
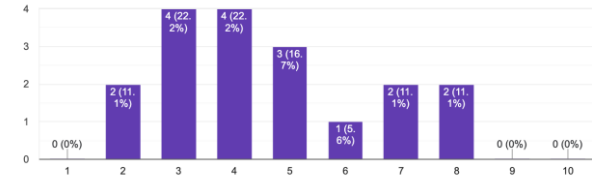
p. 47: "...the human F_{st} of about 0.15 is considerably less than in geographically limited species such as orangutans..."; this statement connects to the image because both species can be put on the same phylogenetic tree and both suggest that humans are much more genetically similar than many other species.

Page 47. "For example, the human F_{st} of about 0.15 is considerably less than in geographically limited species such as orangutans or gazelles (fig. 2.1)." Humans travel and interbreed over large distances whereas species in more isolation tend to separate into different species.

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.

Most Recent Info! 2017



4.

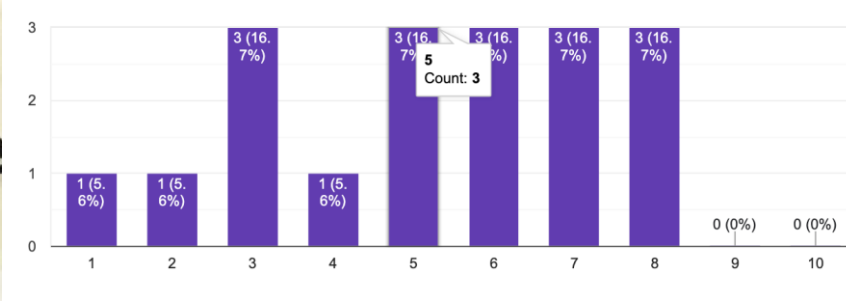
JASON TREAT AND RYAN T. WILLIAMS, NGM STAFF
SOURCE: CHRISTOPHER BAE AND OTHERS, *SCIENCE*, 2017

Page 32: "Migration between populations is an additional factor that works against strong genetic differentiation. Thus, gene flow among human population centers is one of the reasons that humans never formed geographically based biological races (or subspecies). Unlike other species that separate geographically and, once separated, remain so, humans have a history of back-and-forth migration and continual interchange across geographic areas.¹⁹" Because humans spread and interacted with each other and were not isolated there wasn't speciation but only small adaptations to each environment. This also shows why humans are able to exist all over the world because we all spread out and shared traits.

Page 49: "Migration between populations is an additional factor that works against strong genetic differentiation...gene flow among human [populations] is one of the reasons that humans never formed geographically based biological races." The diagram shows that there was significant migration in human populations, and the text supports that because of this migration, humans were never able to speciate or develop strong genetic differences.

The genetic similarity of populations is directly a function of how geographically close they are (pp. 53).

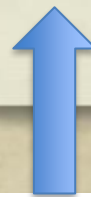
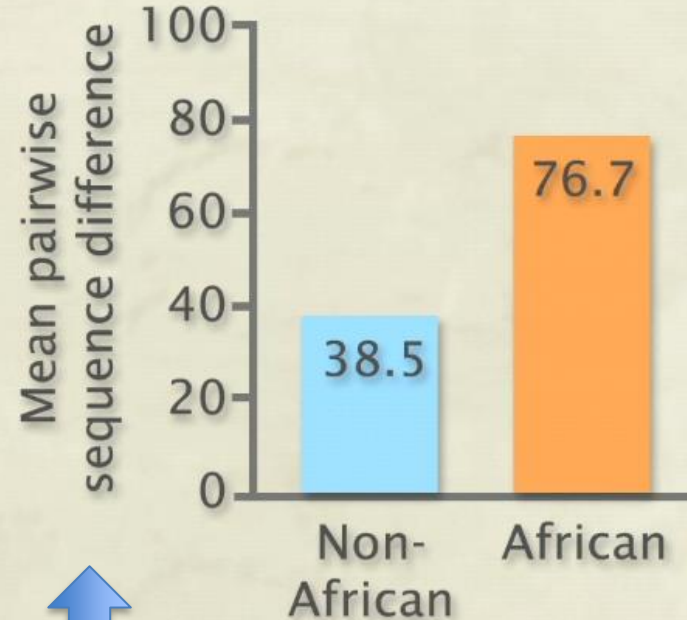
Ge



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”?

pg 53. "Genetic diversity of humans declines with distance from Africa." The graph shows that the genes in African populations have more genetic differences than non-Africans which is similar to the text.

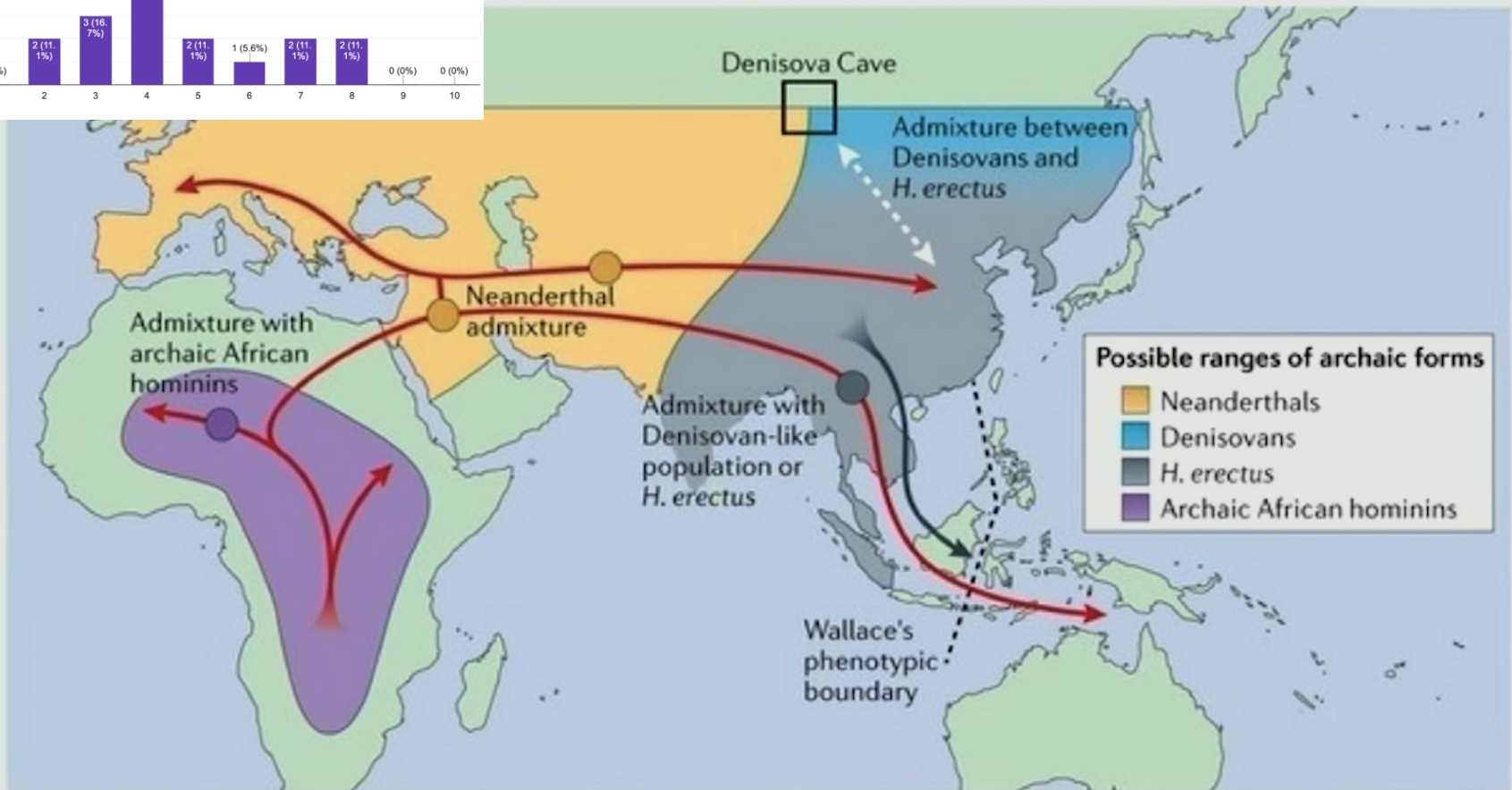
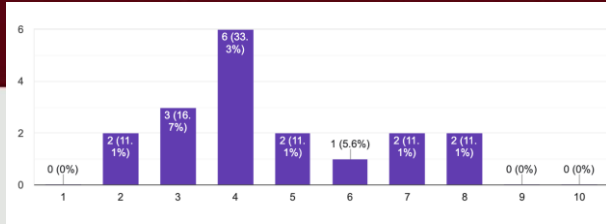
Page 31: "As a result of the NGS revolution, we know that on average about 99.9 percent of the genomes of all living humans are the same.⁴ There may be a higher amount of difference between individuals due to copy number variants (CNV), making humans 99.4% similar in these variants. Copy number variants result from the genetic processes of duplication, deletions, insertions, and inversions.⁵ The global differentiation of SNPs and CNVs are pretty much the same, meaning that there is more variation within populations than there is between populations for these types of genetic variants. Of those differences, the majority are shared in all populations across the globe, smaller amounts are unique to continental origin, such as Africa, Asia, or Europe; and even less are unique to a region within a continent, such as East or West Africa.⁶ The distribution of these differ by population." The amount of variation between individuals in a population is based on small mutations that randomly occur. The amount of variation is different between certain populations based on their region, with some areas like Africa having higher rates of variation.

Page 48" thus considering that 200,000 of our 300,000 year existence was spent in Africa it makes sense that sub Saharan Africans contain more genetic diversity than all other human groups."

53."The figure also shows that the genetic diversity of humans declines with distance from Africa." Despite what you might initially think there is more genetic variation within that singular area on the globe Bec cause there was more time for adaptation and mutations to be selected for

200,000 of our 300,000 year existence was spent in Africa, it makes sense that sub-Saharan Africans contain more genetic diversity than all other human groups (pp. 48). It makes sense to the graph because it gave more time for species in Africa to increase genetic variation.

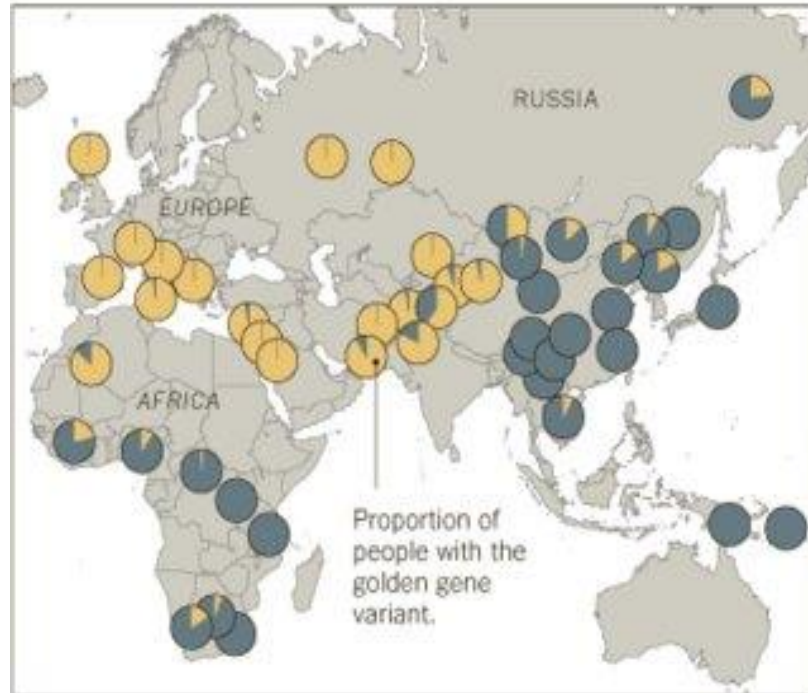
What we need to know: Was there admixture with archaic populations in Africa?



Nature Reviews | Genetics

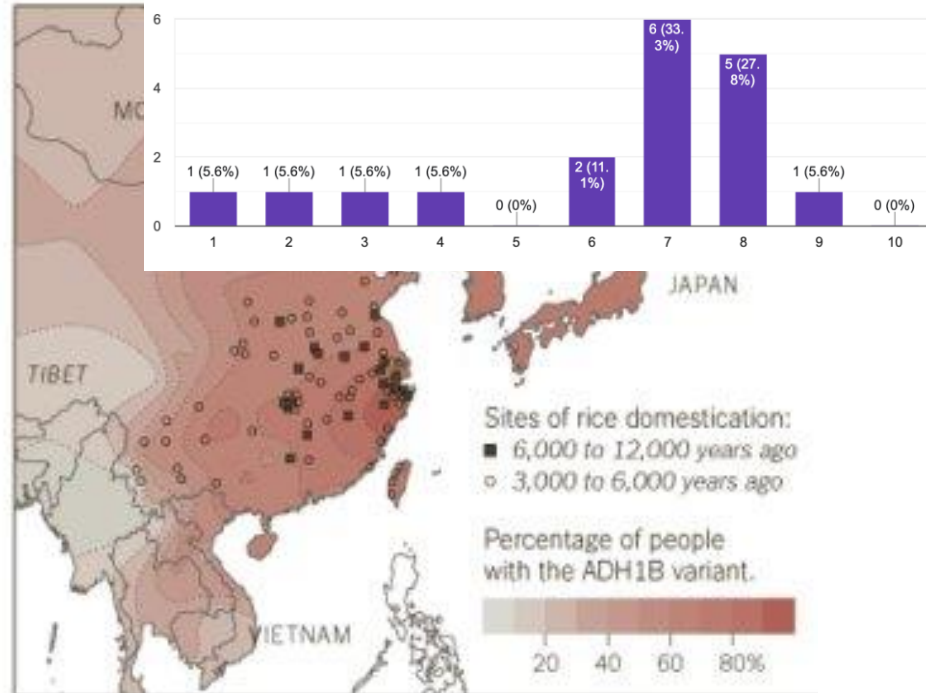
Genetic Changes

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.

Sources: Molecular Biology and Evolution; BMC Evolutionary Biology



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.

THE NEW YORK TIMES

From the article you read/will read!

7.

Page 95 "It's important to note that for this reason, a genetic variant that might be associated with a disease in one group might have no relationship at all with that disease in another group." Proves that there are some genetic differences between certain groups of people regarding health and disease, which compares to the photo above because the photo is detailing the prevalence of certain genes based on location.

50-51

"What explains variation? In the end, it is the processes of adaptation to local conditions, gene flow, and genetic drift, all of which are dependent on variables such as time and population size."

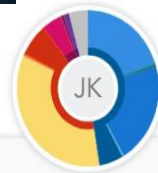
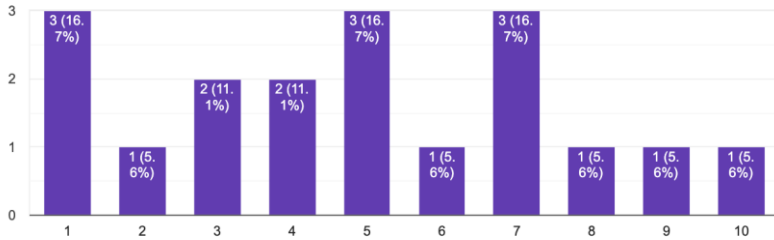
There is variation here because different adaptations for different locations. It doesn't make us different species, we just have different adaptations to different circumstances.

Page 50: "One variant associated with fair skin...went through strong selection"



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%
<ul style="list-style-type: none"> Iberian 19.7% Spain 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%
<ul style="list-style-type: none"> Native American 34.4% Peru 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%
<ul style="list-style-type: none"> West African 4.5% East African < 0.1% 	



8.

<https://www.ispot.tv/ad/wkV/ancestrydna-kim>



Page 44- Actually, we knew already that race was a myth based on prior studies of blood groups and the like, which we discuss later in this chapter. And his finding, based on three individuals, added little to the scientific proof that humans do not have biological races.- ancestors tests can be misleading because it suggests different races.

Pg. 43 "However, this noncoding DNA plays an important role in how genetic ancestry, the genetic connections that have developed through history, is determined by genetic tests." We are able to figure out where people come from based off of their genetics, the environment also has an important impact on genetic variation.

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

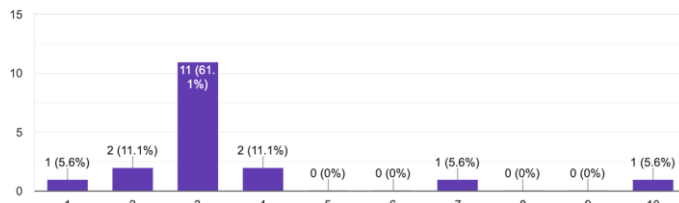
[Rights & Permissions](#)

The discovery of yet another period of interbreeding betw adding to the growing sense that sexual encounters amoi commonplace throughout their history.

"As more early modern humans and archaic humans are sequenced, we're going to see many more instances of ir 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.

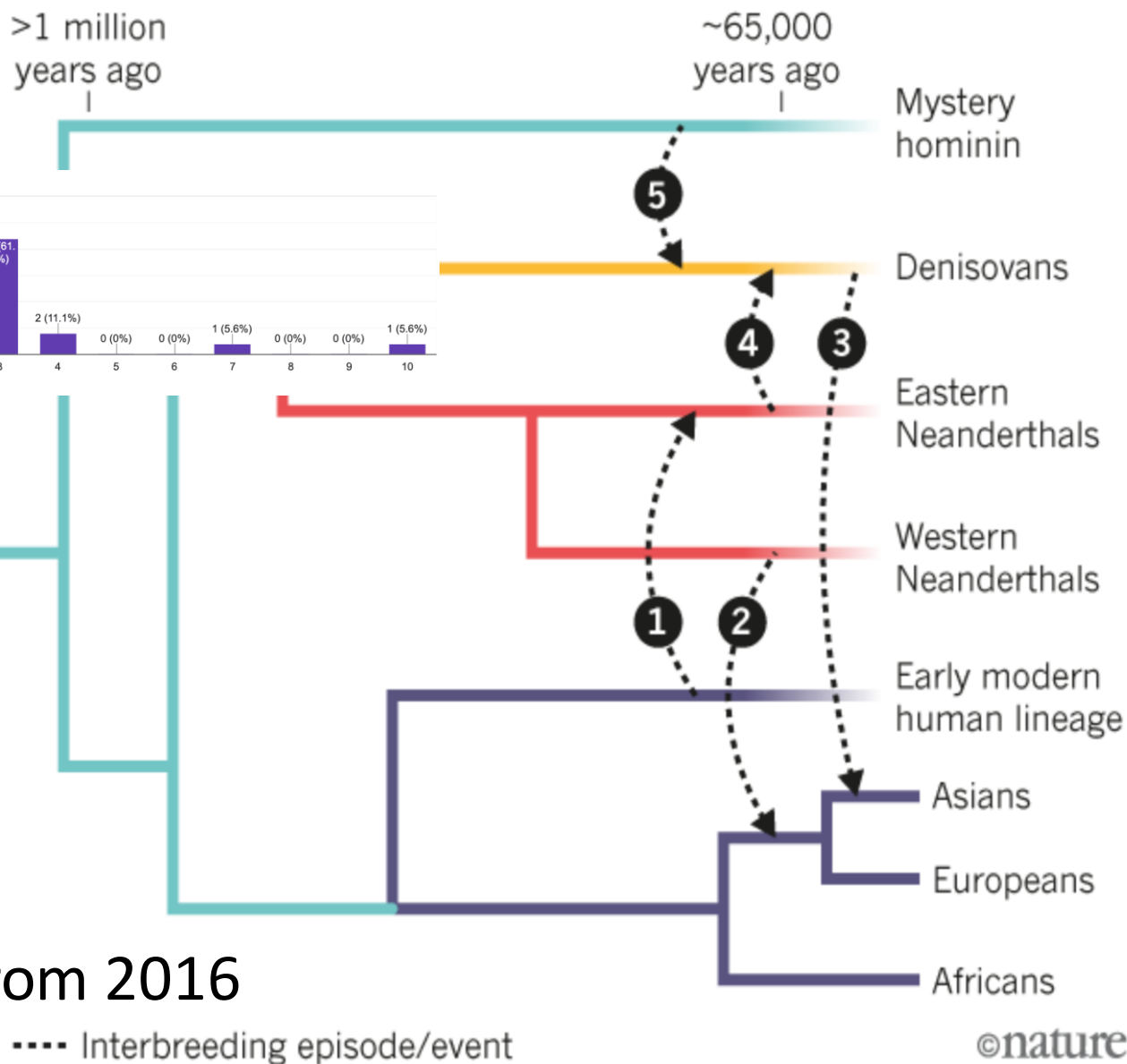


Neander outside e human b



A HISTORY OF INTERBREEDING

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



9. Inferring interbreeding or admixture from genome analysis

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

Page 56. "The evidence suggest that our species became hybridized through reproduction with the archaic humans *H. naledi* (in sub-Saharan Africa), *H. neanderthalensis* (in Eurasia), and the Denisovans (in East Asia, Melanesia, and Australia)." Our genetic makeup overlaps with other hominins, and the different regions have different influences on which hominins interbred.



Then found **AN INDIVIDUAL** that was a result of interbreeding.

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

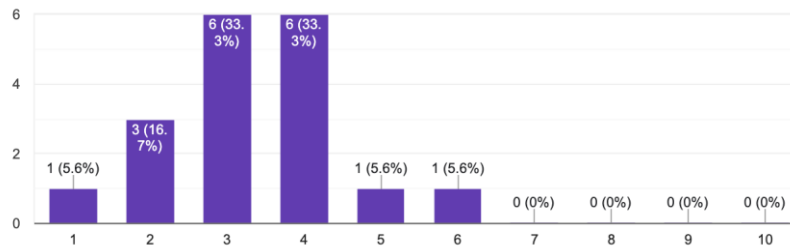
10.

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 palaeoanthropologist Svante Pääbo of the Max Planck Institute for Evolutionary Anthropology, conducted the genome analysis on a single bone fragment discovered in the Altai Mountains of Russia. This cave belonged to



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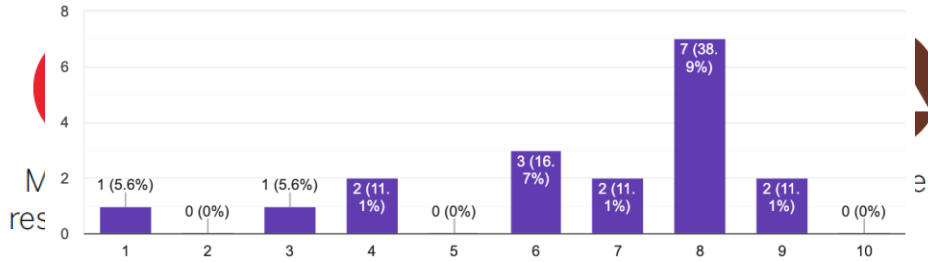
identified on the basis of DNA sequences from the tip of a finger bone discovered² there in 2008. The Altai region, and the cave specifically, were also home to Neanderthals.



Marine diet



Lactose tolerance

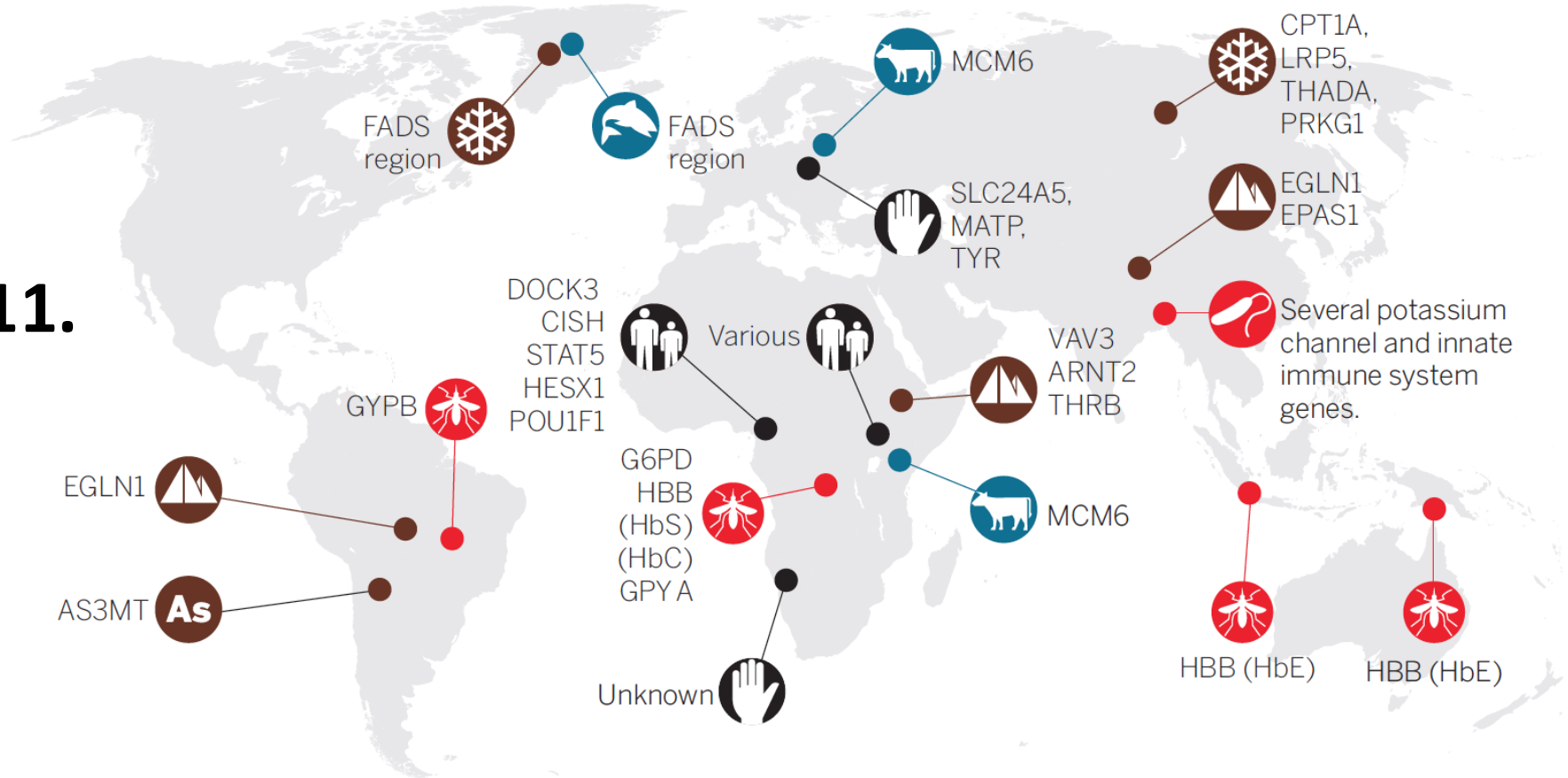


Light skin pigmentation



Short stature

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.

pg 50. "Humans adapted to altitude, new climates, diets, pathogens, solar intensity, and toxins." The image shows different adaptations to environmental factors in different areas of the world, which the text supports as the factors it mentioned are environmental factors across the world.

"As human populations established themselves in new regions, they also began to accumulate new mutations." page 49. This relates to the image because it shows how organisms adapt to their surroundings.

Page 95 "It's important to note that for this reason, a genetic variant that might be associated with a disease in one group might have no relationship at all with that disease in another group." Different groups of people / locations have genetic differences due to certain environmental factors, which includes genetic differences that impact people's health and experiences with certain diseases (malaria and cholera).

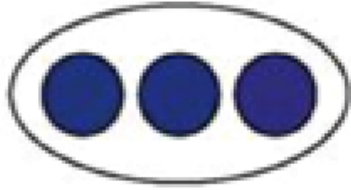
53

"The best way to explain the distribution of human gene frequencies is the isolation-by-distance model. This means that the genetic similarity of populations is directly a function of how geographically close they are (fig. 2.3)."

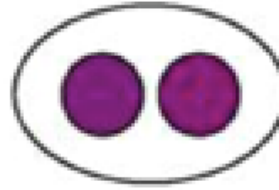
There are different adaptations because they needed to adapt to different things and they were far apart.

51. table 2.1 gives us quantifiable data about the different genetic variations selected for across the human population

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



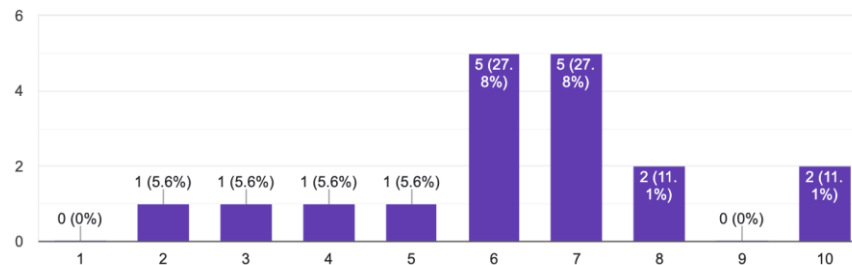
Location 1 with three subpopulations



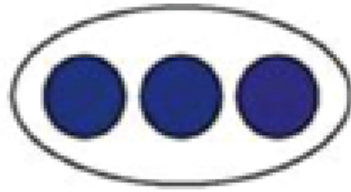
Location 2 with two subpopulations



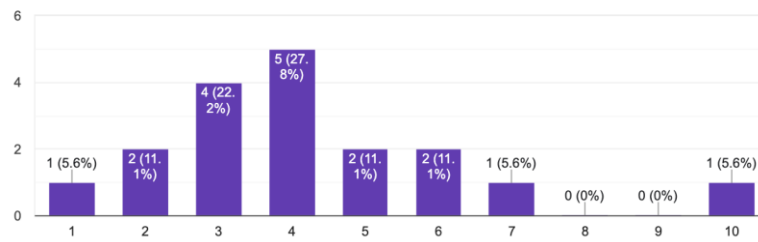
Location 3 with two subpopulations



pg 52. "Geographic closeness is very highly correlated with genetic similarities."
The image shows that subpopulations in different areas are genetically similar to each other, which is what the text says.



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

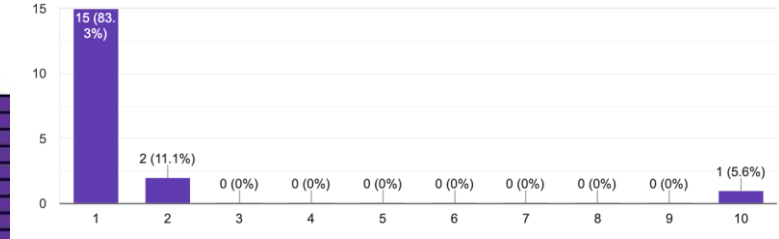
How does this additional info affect your perspective?

14.

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



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



From Donovan 2015 and Donovan et al 2019

Science Education. 2019;1-32.

pg 45. "99.9% of the genomes of all living humans are the same." The statement from the text matches with the graph, both show 99.9% similarity in human DNA.

Page 43- On average, about 999 of the 1,000 letters in base pairs of the genome are the same among individuals. Alan and Joe, or Alan and you, or Joe and you, whoever you might be, are about 99.9 percent similar genetically.- talks about how similar our genome is and basically explains this graphic.

p. 45: "...on average about 99.9 percent of the genomes of all living humans are the same"; this directly connects with the image because both explain how the majority of DNA in the human genome is shared between all people.

Page 30: "Recent advances in human genetics have shown us just how similar all humans are genetically. On average, about 999 of the 1,000 letters in base pairs of the genome are the same among individuals. Alan and Joe, or Alan and you, or Joe and you, whoever you might be, are about 99.9 percent similar genetically." Both the graph and sentence show how similar genetically all of us are. There is small variation between individuals, but the majority of the genome is consistent between individuals.

"We know that on average about 99.9 percent of the genomes of all living humans are the same" (pg. 45). They are saying the same thing!

Page 45: "As a result of the NGS revolution, we know that on average about 99.9 percent of the genomes of all living humans are the same." This sentence supports the image because they both state that living humans share 99.9% of our DNA, and thus, only 0.1% of our DNA makes us different from each other.

Page 45 " as a result of the Ngs revolution, we know that on average about 99.9 percent of the genomes in all living humans are the same." The image and the text both state that humans share. 99.9% of dna, that 99.9% is what makes us homosapiens

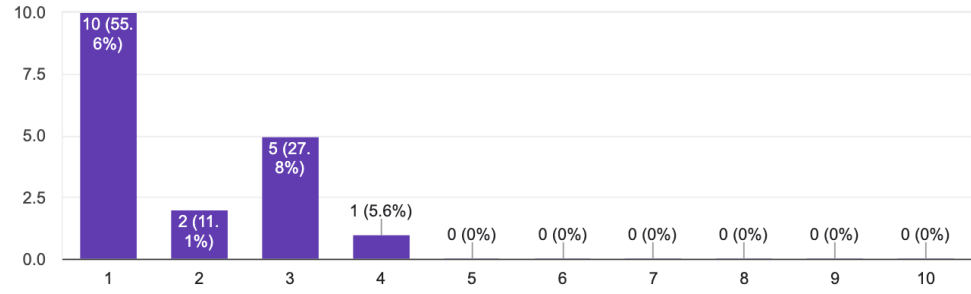
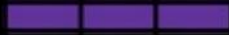
Page 43. "Alan and Joe, or Alan and you, or Joe and you, whoever you might be, are about 99.9 percent similar genetically." We are very genetically similar, and only a small amount of genetic difference makes up the diversity of humans.

Migration between populations is an additional factor that works against strong genetic differentiation. Thus gene flow among human population centers is one of the reasons that humans never formed geographically based biological races (pp. 49).

Pg 43.

As a result of the NGS revolution, we know that on average about 99.9 percent of the genomes of all living humans are the same

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



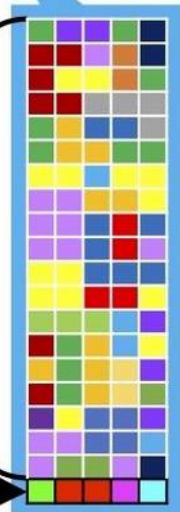
0.1% - DNA that makes humans different from each other:
Not shared by everyone



95.5% - DNA that differs between *same race* individuals



15. 4.5% - *Extra* DNA that differs between individuals of different races



Pg. 43 "This means that each person differs from any other in about 3.3 million base pairs." This quote correlates with the picture showing how much variation occurs within the %0.1.

"As a result of the NGS revolution, we know that on average about 99.9 percent of the genomes of all living humans are the same. There may be a higher amount of difference between individuals due to copy number variants (CNV), making humans 99.4% similar in these variants." pg. 45. This connects to the image because it shows how there's only a tiny part of our genome that actually varies and the rest is all the same.

58

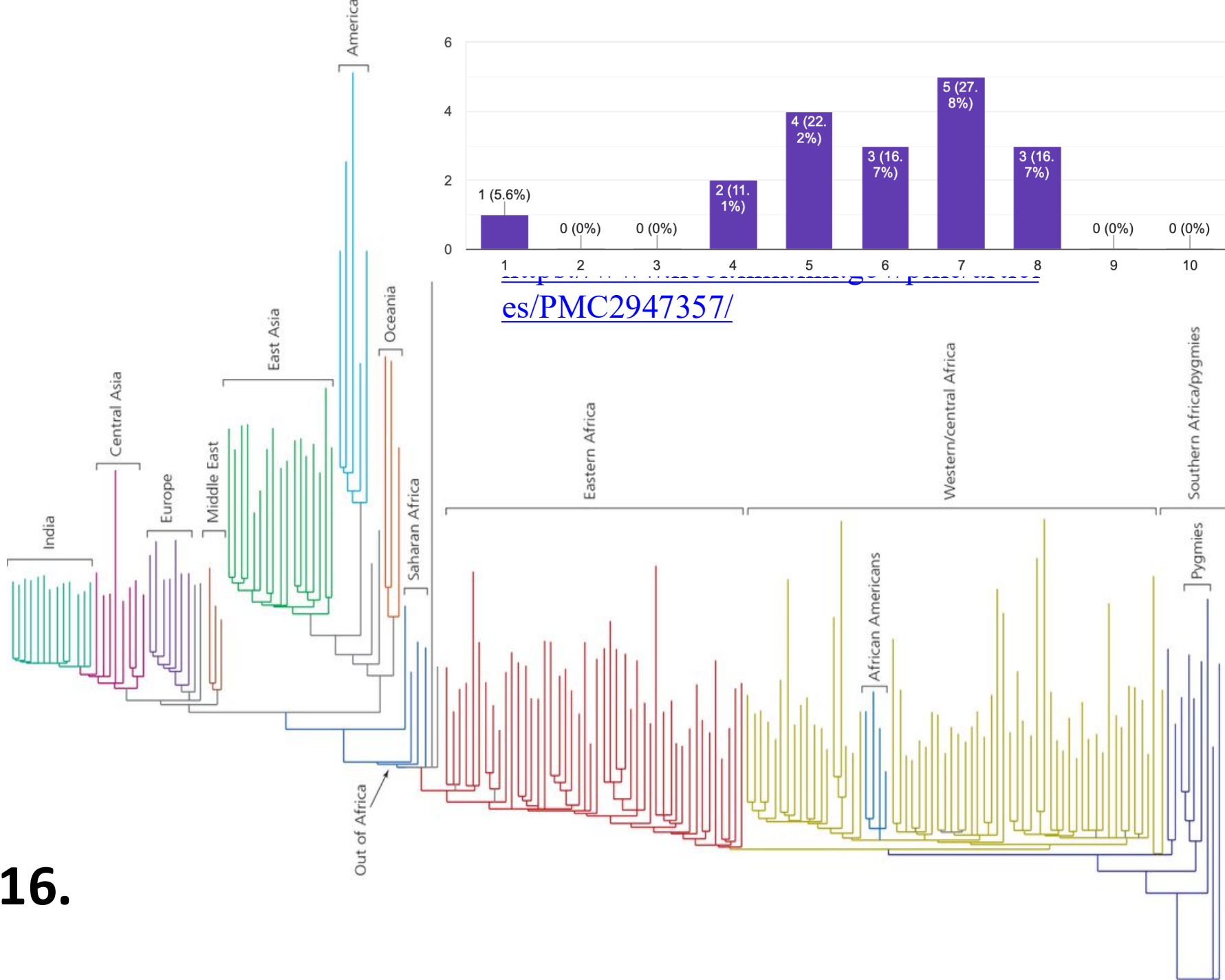
"Human variation is not explained by race. Rather, variation is explained by evolution. Evolution acts locally, not on social races or continents. Race also does not describe human variation."

People vary within race and between race, it is adaptation with gene flow not a founder event.

Pg. 43

Everyone except identical twins varies genetically. Recent advances in human genetics have shown us just how similar all humans are genetically. On average, about 999 of the 1,000 letters in base pairs of the genome are the same among individuals.

16.



p.49 "...human populations that migrated within Africa faced new environmental conditions... more time equates to more opportunity to build up variation."

Human populations that migrated within Africa faced new environmental conditions, this part explains why sub-Saharan Africans display greater genetic diversity than all other world populations. More time equates to more opportunity to build up variation (pp. 49).

Visualizing Human Genetic Diversity

James Kitchens and Graham Coop

May 16, 2023

Genetics D3 Python R

<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)

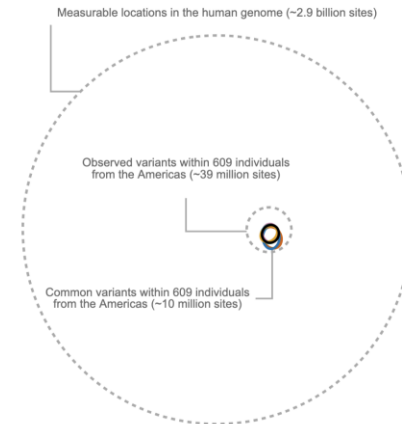
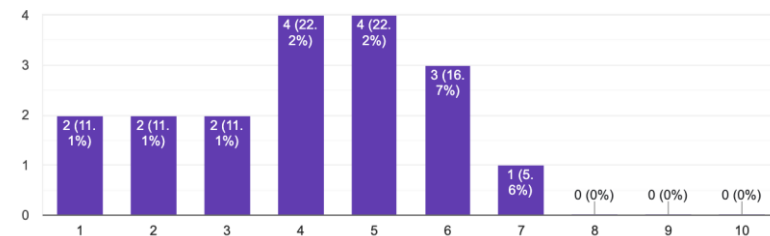
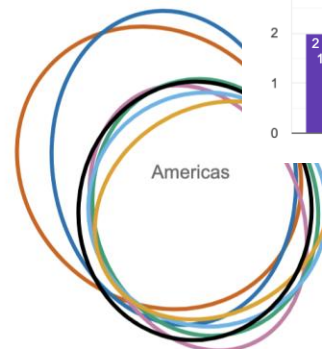
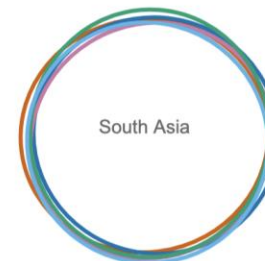
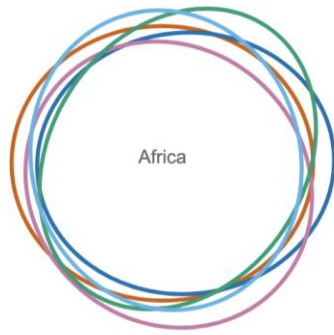


Figure 5 - Common variants in perspective. An Euler diagram of the common variants in samples located in the Americas relative to the scale of the human genome. As a small note, the positions and orientations of ellipses within the Euler diagram differ slightly from Figure 3. This is because the euler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



17.

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.