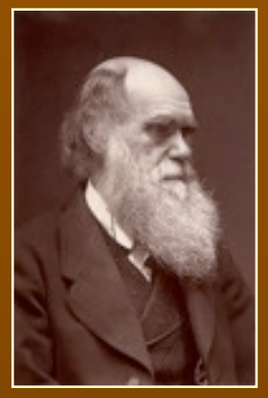
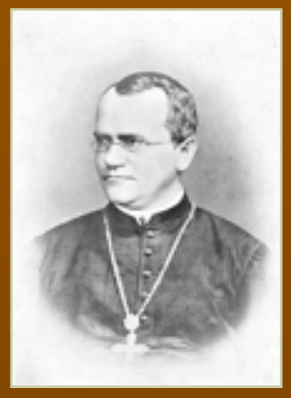
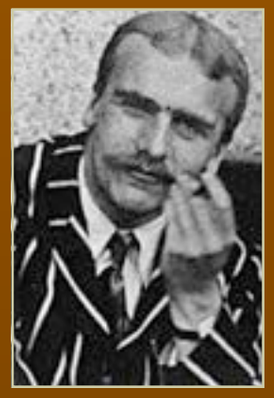
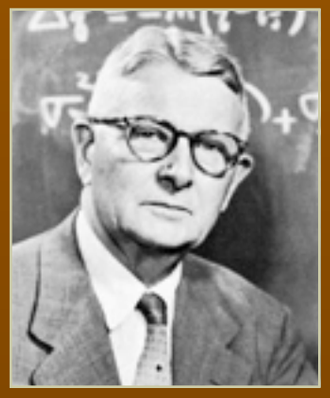
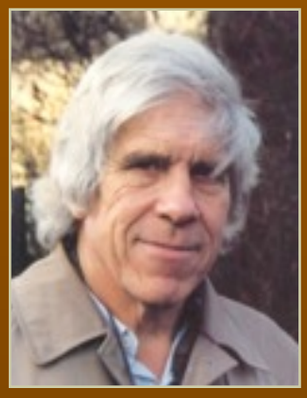
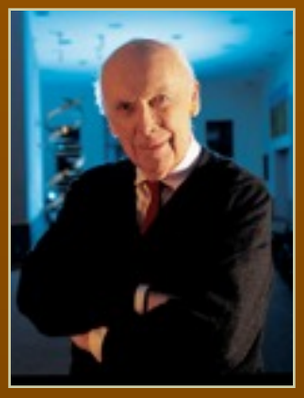





Diversity, Culture, Theory, and Data:

Science on Human Variety

B. Ricardo Brown and Christopher X J. Jensen

http://until-darwin.blogspot.com/ http://www.christopherxjjensen.com/







How does science work?

theory

tests

Question

answered by

Hypothesis 1 *makes* **Predictions 1**

refutes

Lab experiments

answered by

Hypothesis 2 *makes* **Predictions 2**

validates

Field experiments

answered by

Hypothesis 3 *makes* **Predictions 3**

refutes

Collected observations

data

What questions get asked?

What hypotheses are suggested?

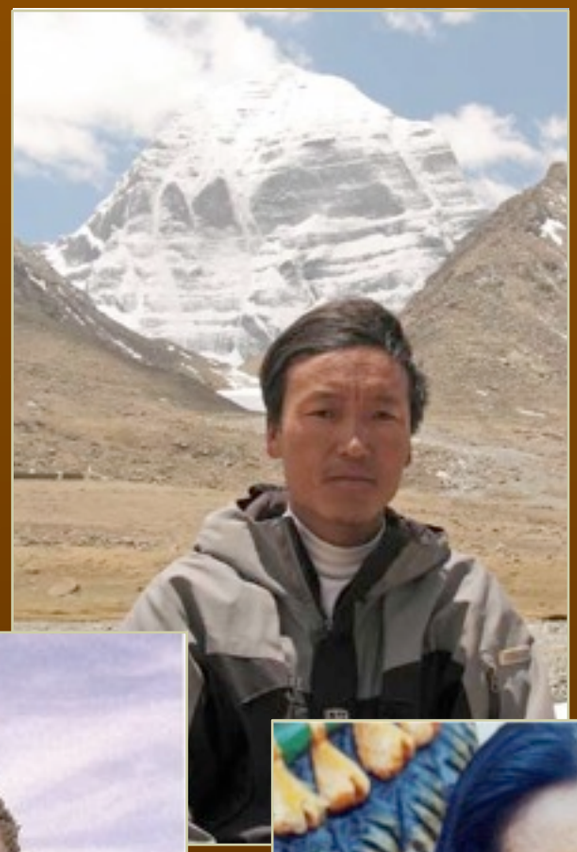
What tests can be performed?

What tests are performed?

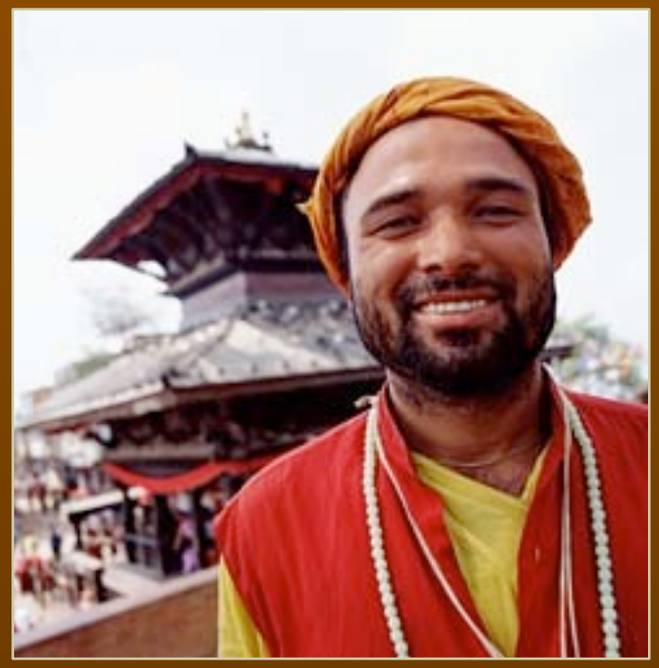
culture

Why did regional differences evolve?

(*Source:* http://homepage.mac.com/huntington.c)



(*Source:* http://www.thomasriddle.net/)



(*Source:* http://images.jupiterimages.com)

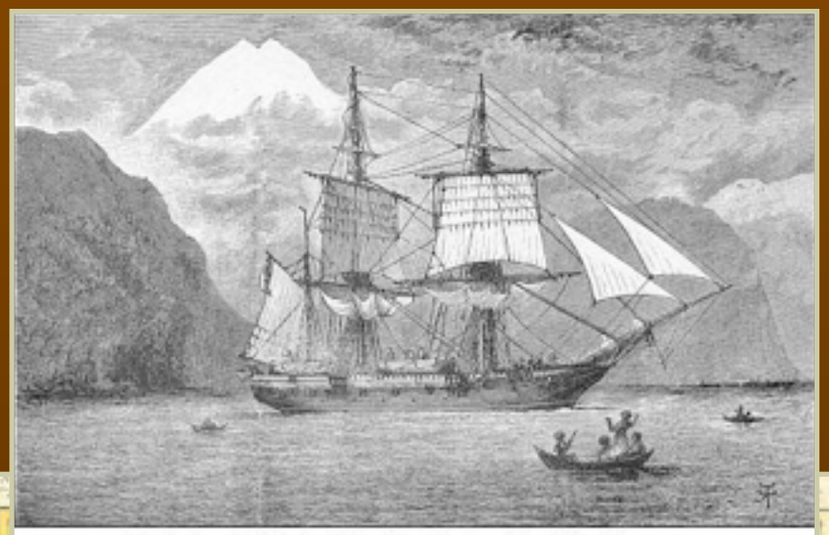
(*Source:* http://fixedreference.org)

(*Source:* http://www.mongabay.com)

(*Source:* http://ethnicityetc.com)

(*Source:* Ken Ilio)

What evidence was available in Darwin's time?


H.M.S. *Beagle* in Straits of Magellan. Mt. Sarmiento in the distance.

(*Source:* R. T. Pritchett)

(*Source:* Geographicus Fine Antique Maps)

Too limited to yield meaningful insights?

(*Source:* Grafton Elliot Smith's "The Royal Mummies")

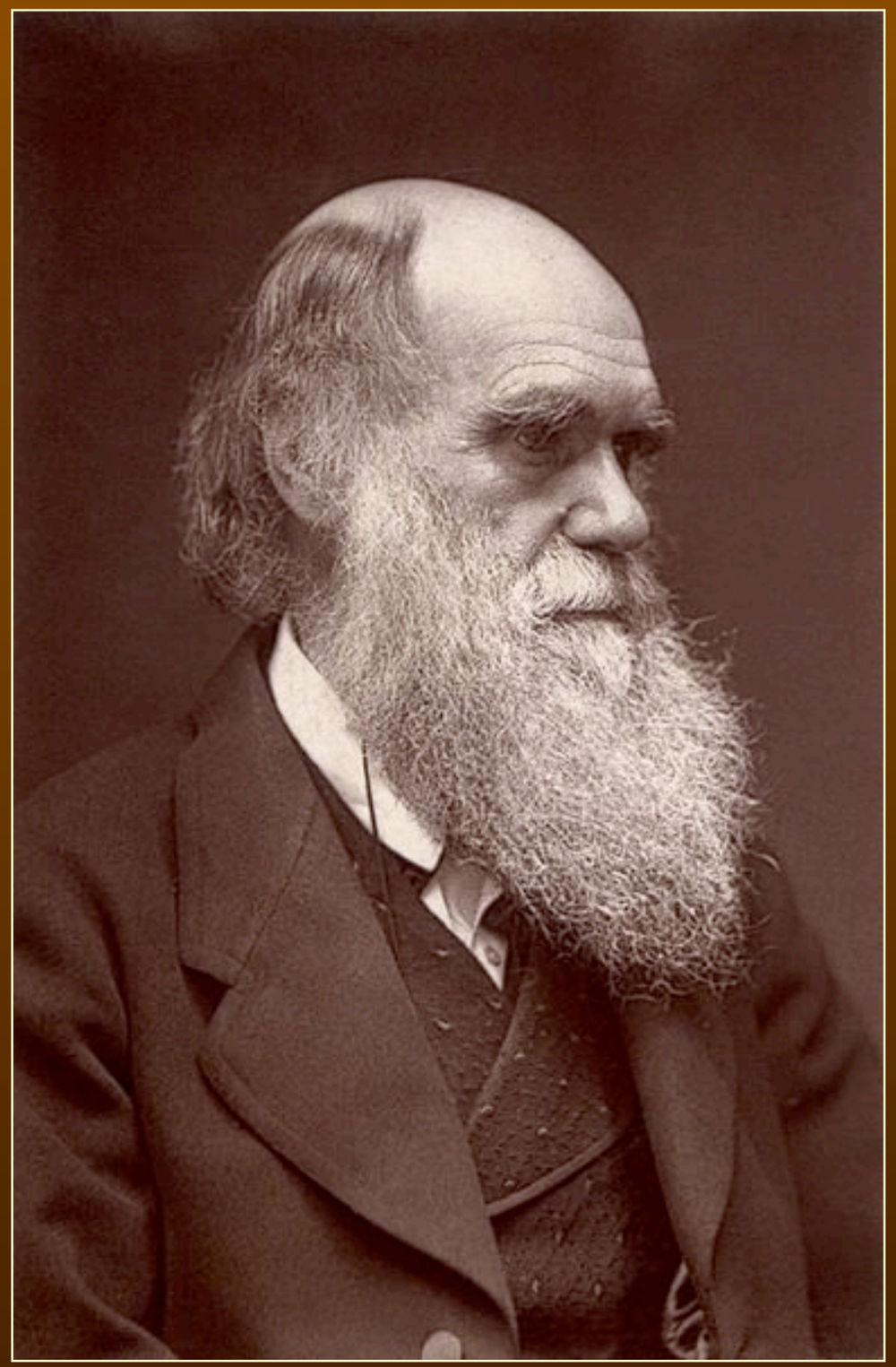
Too recent to yield meaningful insights?

(*Source:* Grafton Elliot Smith's "The Royal Mummies")

Existing knowledge of human diversity

Some fossil evidence

Darwin's revolutionary theories:

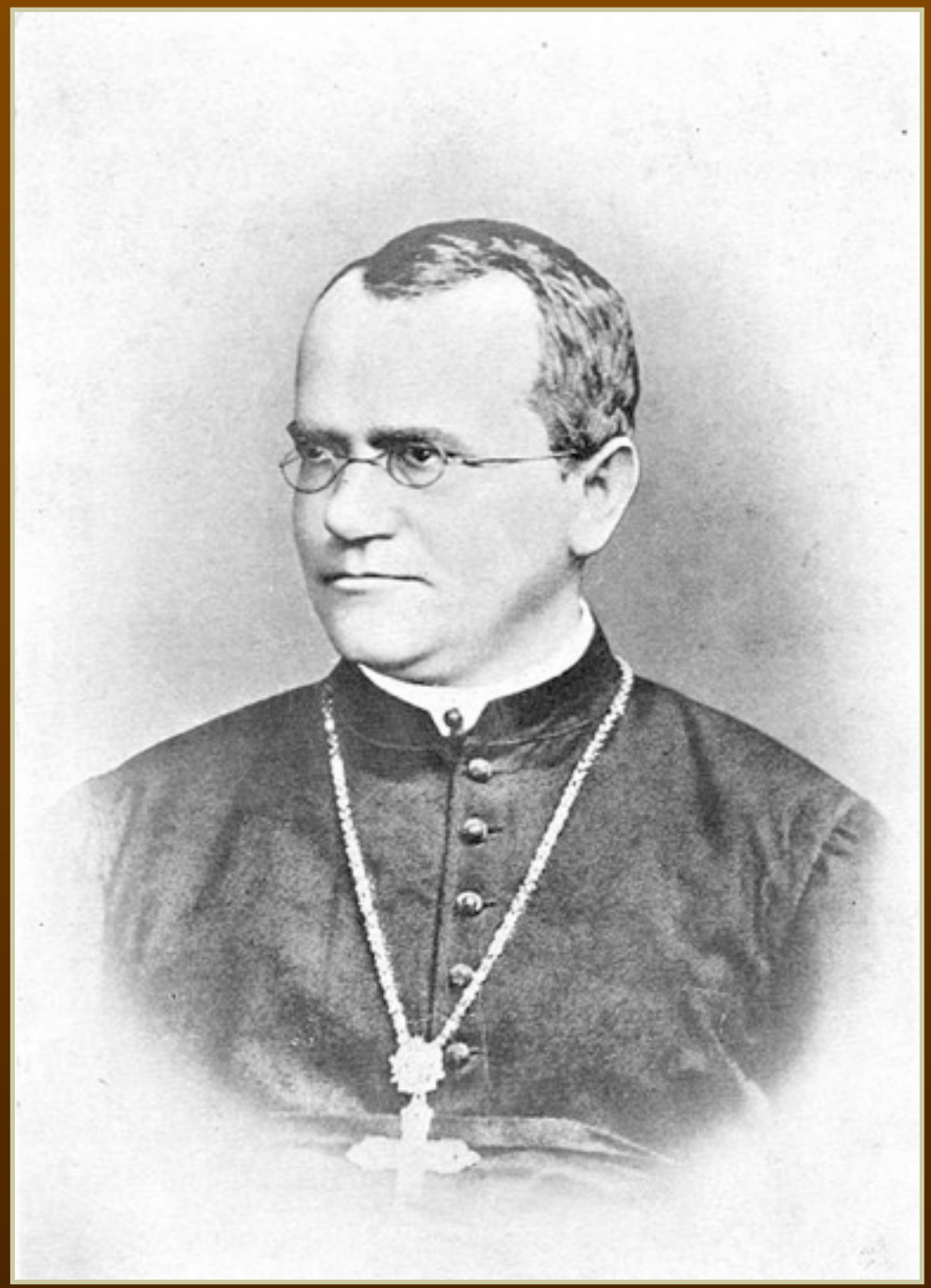


(*Source:* Robert Ashby Collection / John G. Murdoch)

- ★ All living things stem from a common ancestor. The process of speciation creates diversity by splitting existing species.
- ★ Populations adapt to their present local environment due to the differential survival and reproduction of individuals in that environment ("natural selection").
- ★ The organismal diversity we observe today is the result of a slow, deliberate process of selection and speciation.

Gregor Mendel

- ★ Inheritance is particulate. We now call these particles “genes”.
- ★ [Eukaryotic] organisms possess two copies of each gene; these copies may differ or be identical.
- ★ Individual organisms inherit one copy of each gene from each of their parents.
- ★ Our two gene copies can interact in a variety of ways to produce traits.



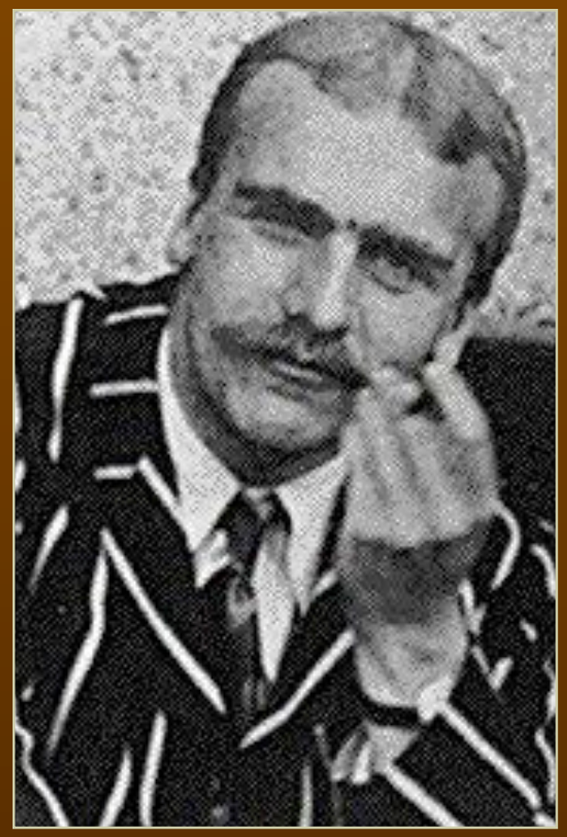
(*Source:* William Bateson)

The “Modern Synthesis”:

Ronald Fisher

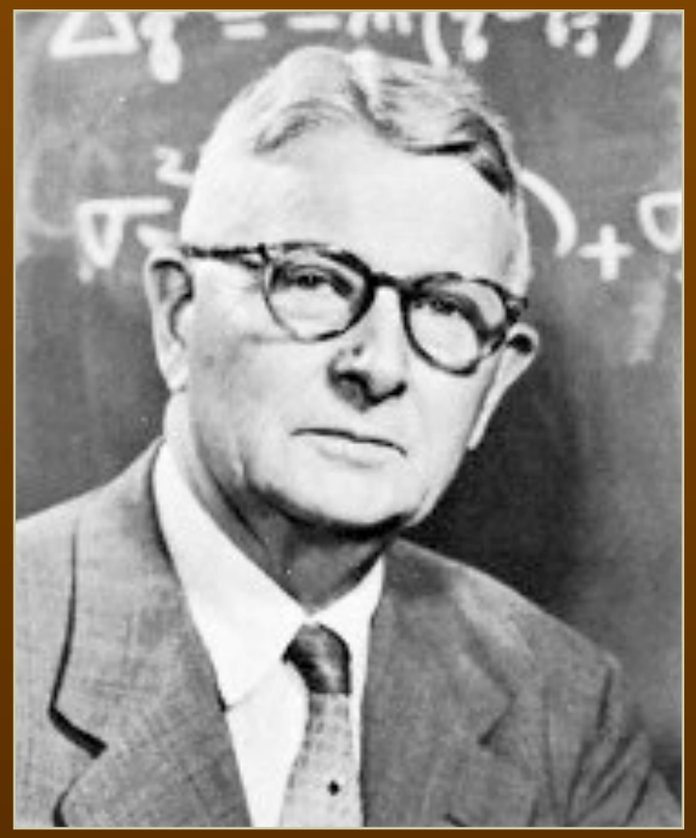
(*Source*: Unknown, via Wikimedia Commons)

JBS Haldane



(*Source*: Unknown, via Wikimedia Commons)

Sewall Wright



(*Source*: Unknown, via Wikimedia Commons)

1910’s, 1920’s, 1930’s

Harmonized Darwinian theory and Mendelian genetic theory

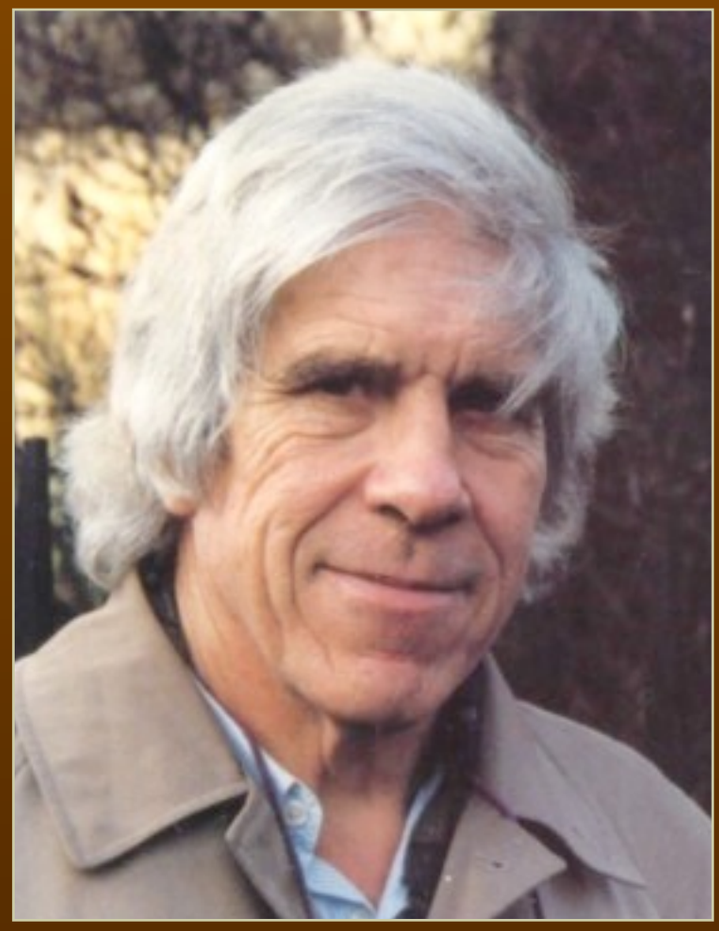
The “Modern Synthesis”:

Theodosius Dobzhansky

(*Source:* Unknown, via Wikimedia Commons)

1930’s

William D. Hamilton



(*Source:* Richard D. Alexander)

1960’s

Harmonized Darwinian theory and Mendelian genetic theory

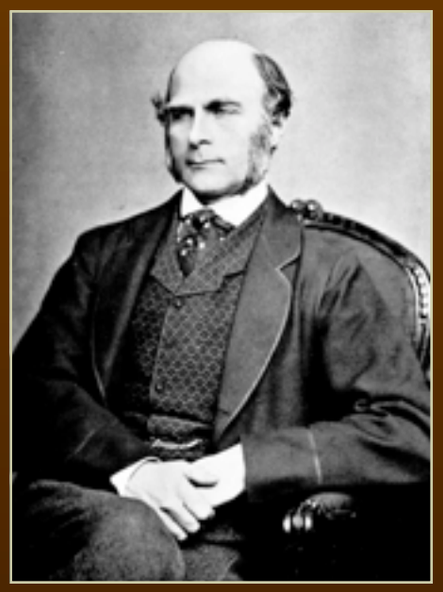
Eugenics:

Selective breeding is a form of eugenics:

(*Source:* Hannes Grobe)

(*Source:* Andy Wright)

(*Source:* Dave Wharton)



(*Source:* Karl Pearson)

Francis Galton

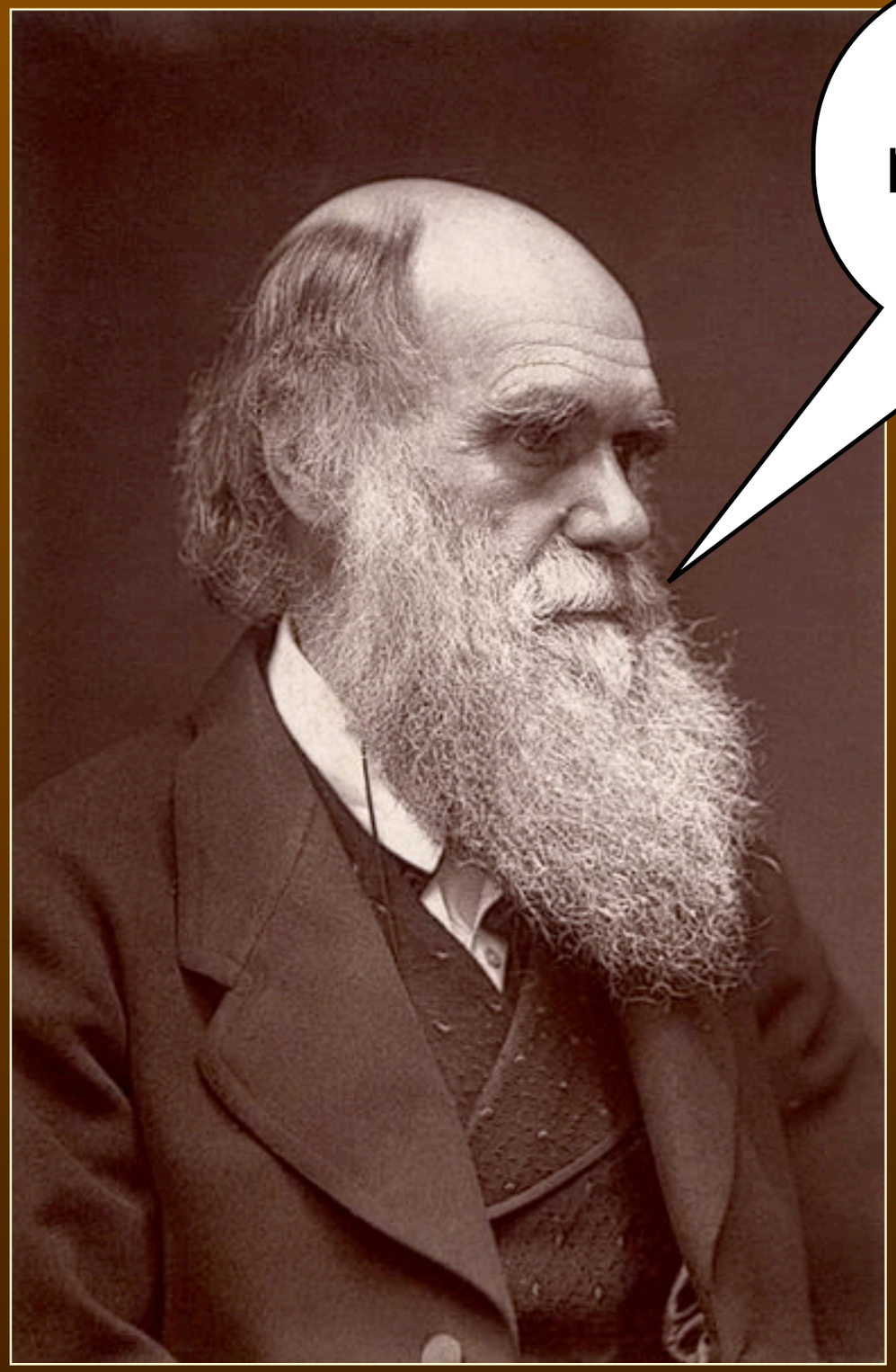
(*Source:* Unknown, via Wikimedia Commons)

Ronald Fisher

(*Source:* "The Second International Congress of Eugenics")

Charles Davenport

Was eugenics based on evolutionary science?

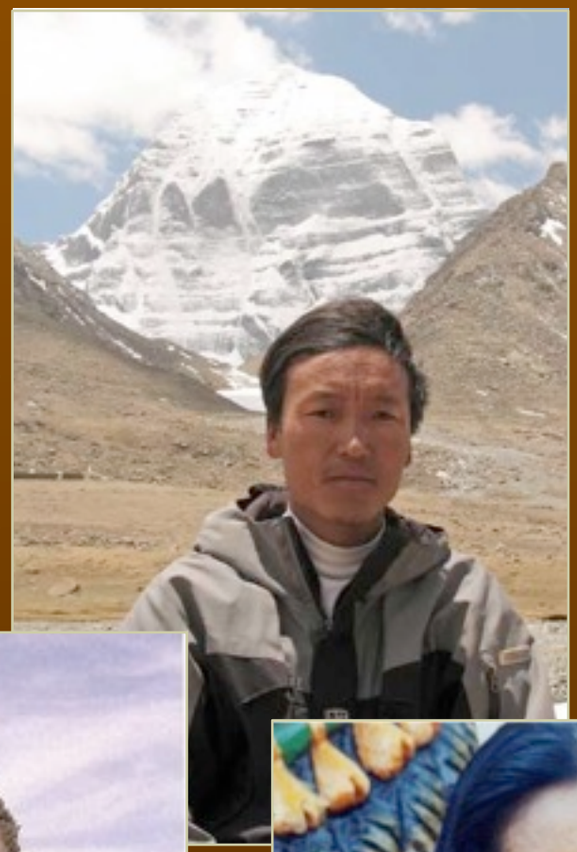


(*Source:* Robert Ashby Collection / John G. Murdoch)

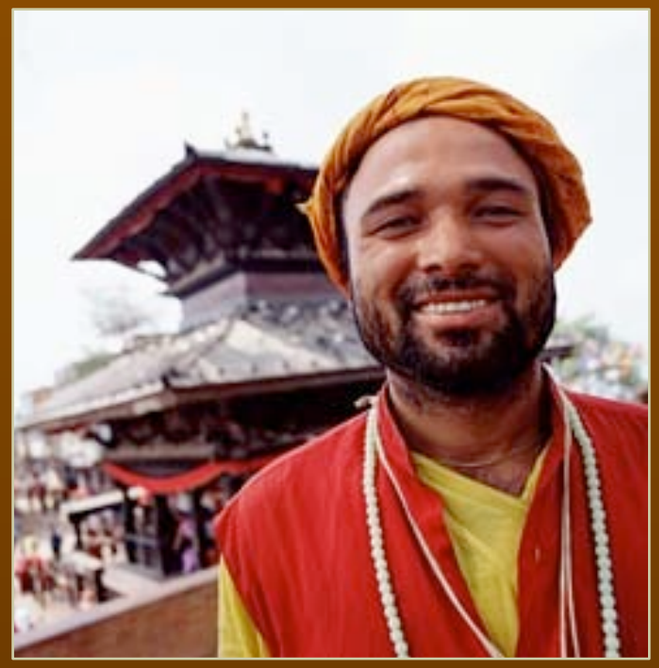
(*Source:* Bundesarchiv, Bild 102-13774 / Heinrich Hoffmann)

Why did regional differences evolve?

(*Source:* http://homepage.mac.com/huntington.c)



(*Source:* http://www.thomasriddle.net/)



(*Source:* http://images.jupiterimages.com)

(*Source:* http://fixedreference.org)

(*Source:* http://www.mongabay.com)

(*Source:* http://ethnicityetc.com)

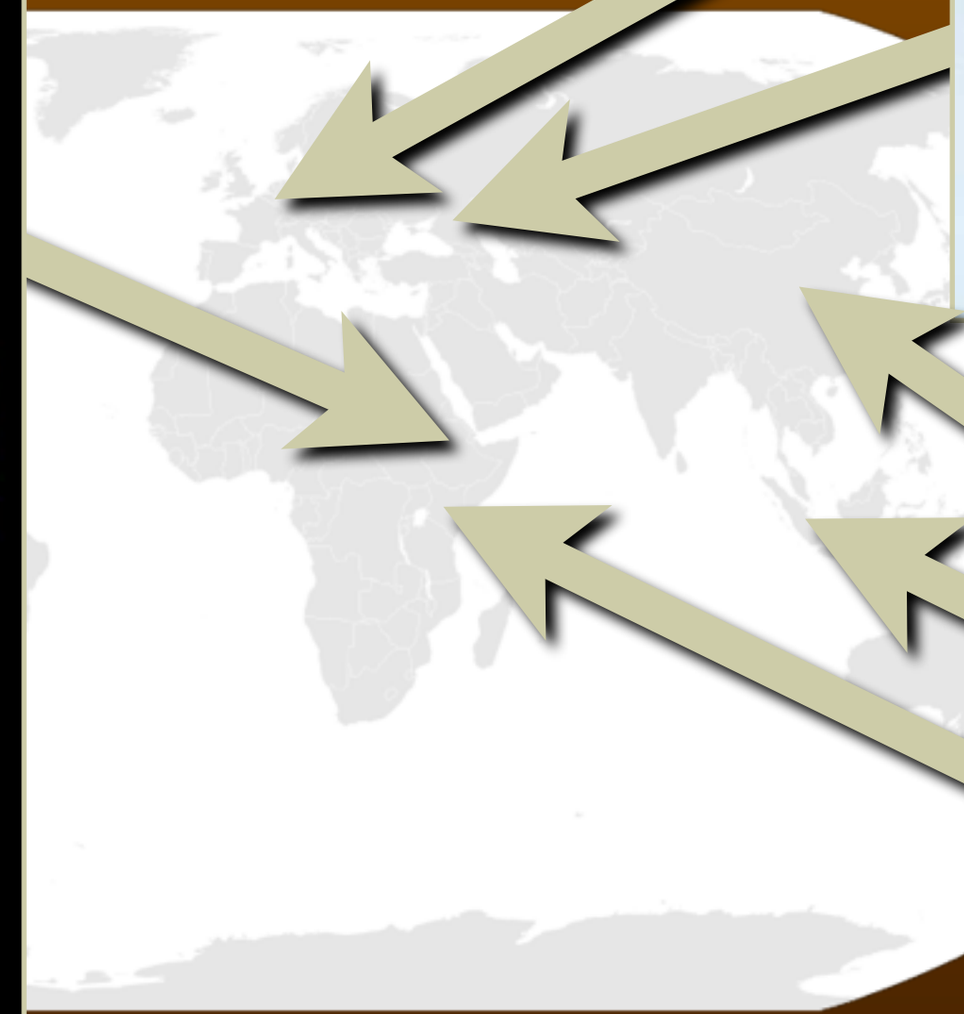
(*Source:* Ken Ilio)

Fossil exploration explodes in the 1900's:

Australopithecus afarensis

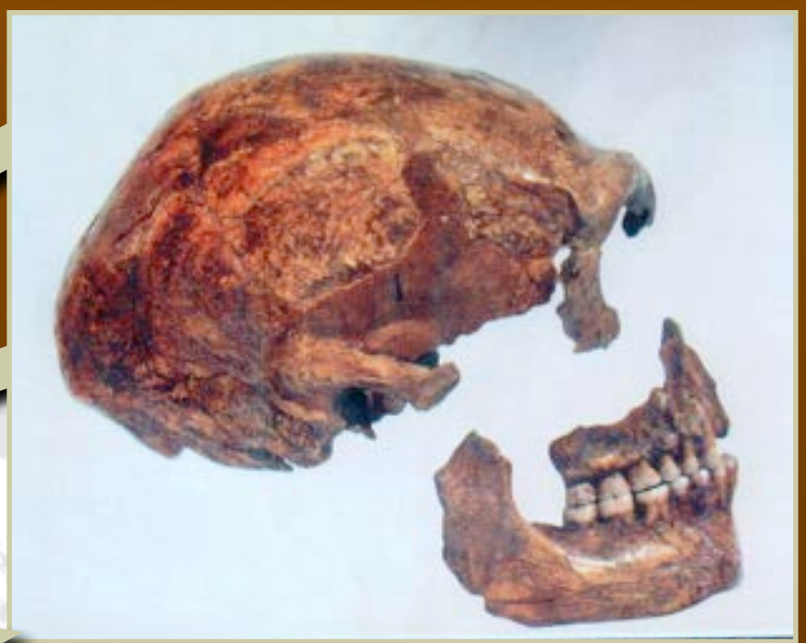
"Lucy"

(*Source:* Wikimedia Commons)



(*Source:* Wikimedia Commons)

Homo neanderthalensis



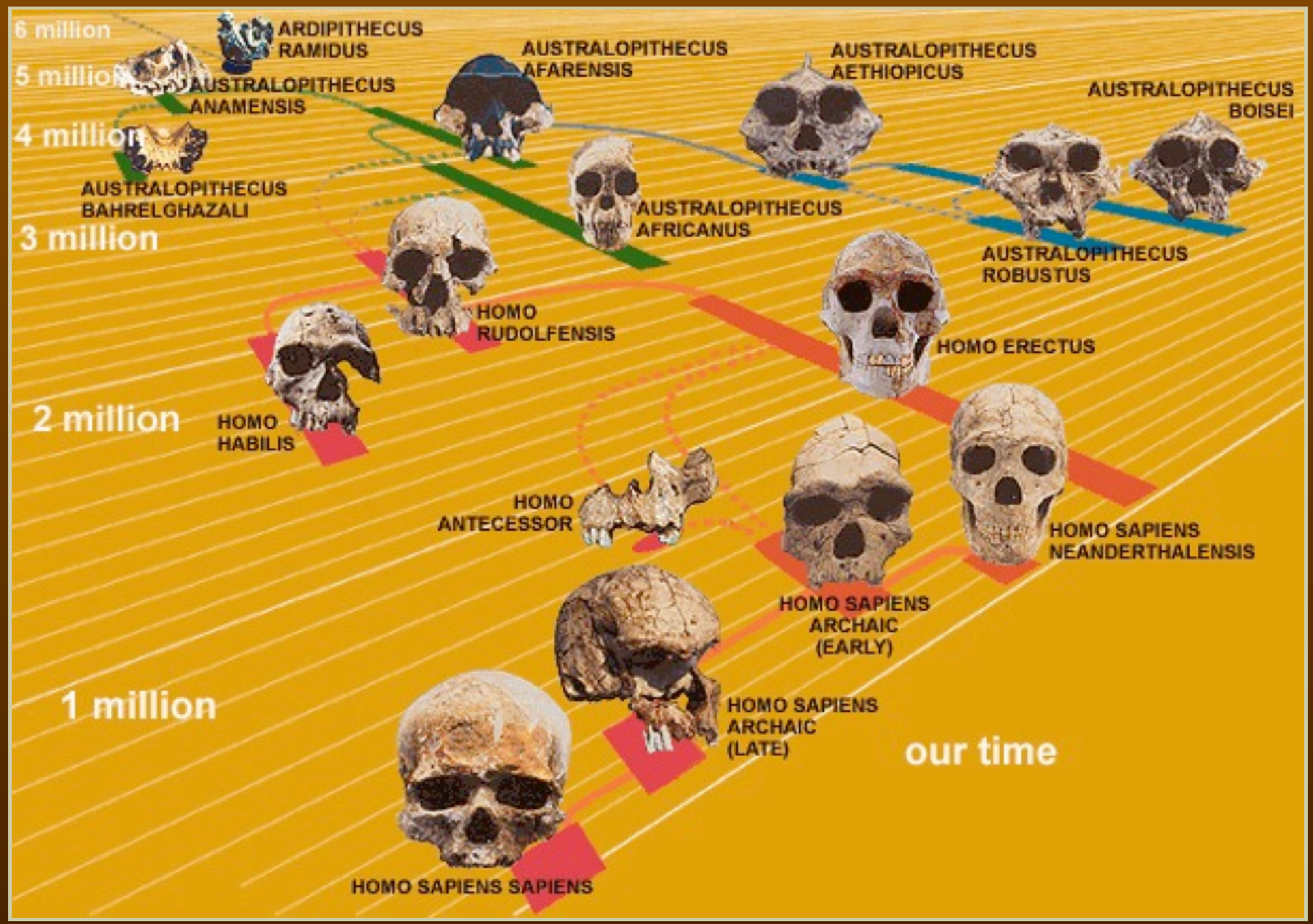
(*Source:* We El)

(*Source:* Daderot)

Homo erectus

The polygenists were right!

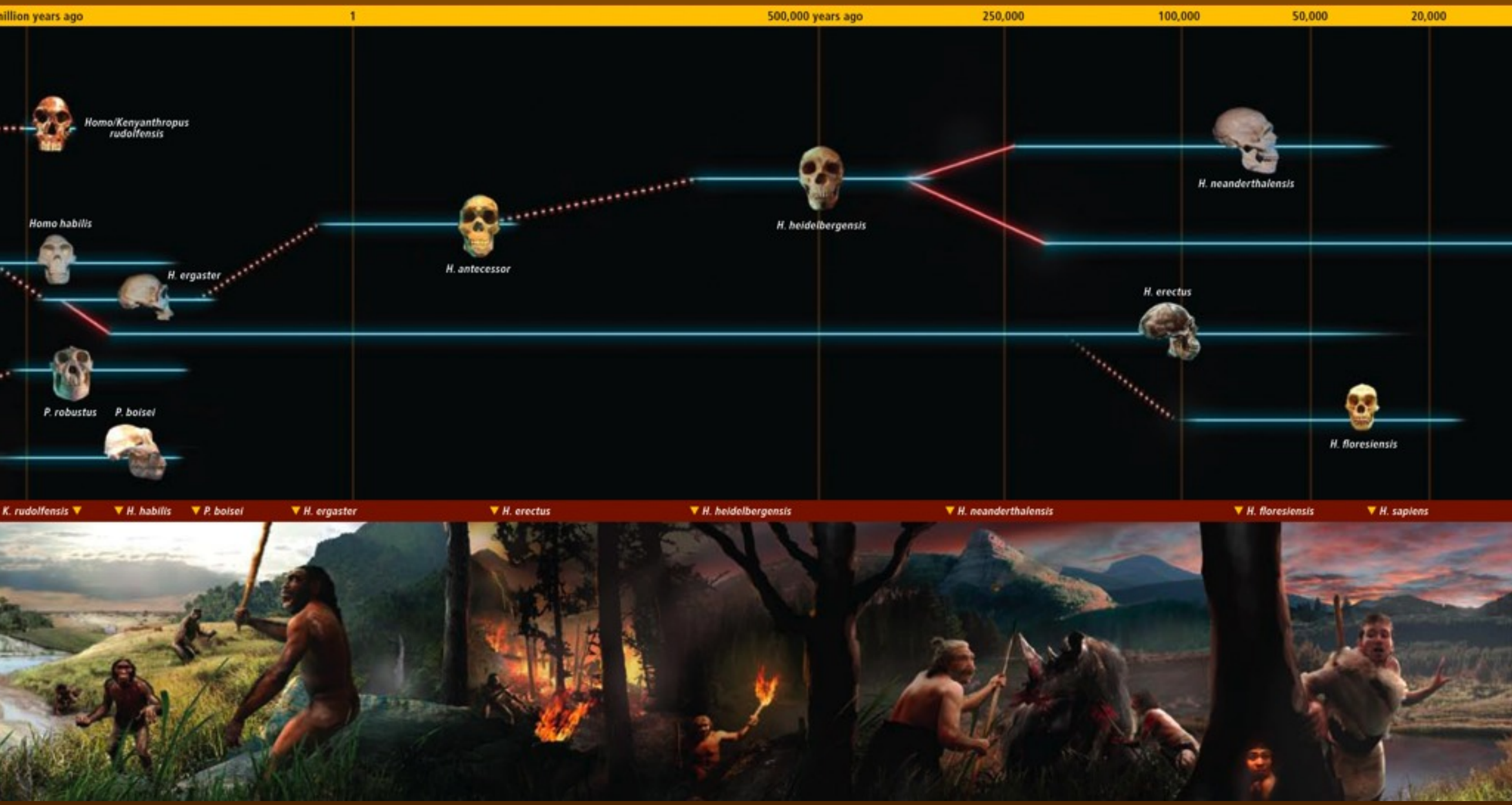
(just about the hominid family, not the species *Homo sapiens*)



(*Source*: www.greatdreams.com/ african-lucy.htm)

The monogenists were right!

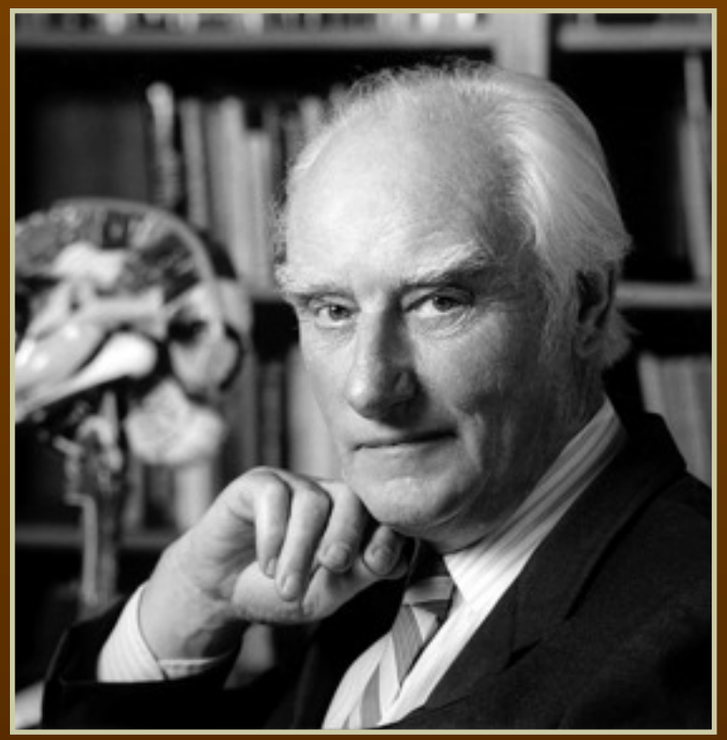
(just about the single recent origin, not the timeline or trajectory of our evolution)



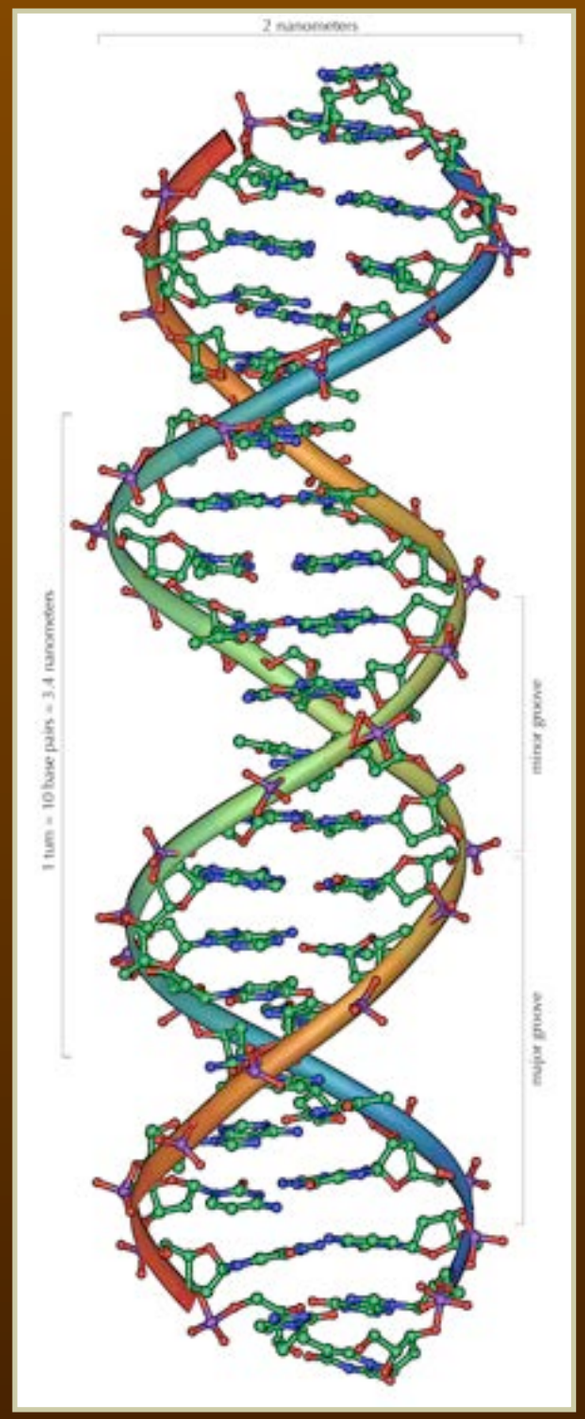
(*Source:* Scientific American)

The discovery of DNA's structure and techniques for reading our genetic code

Francis Crick

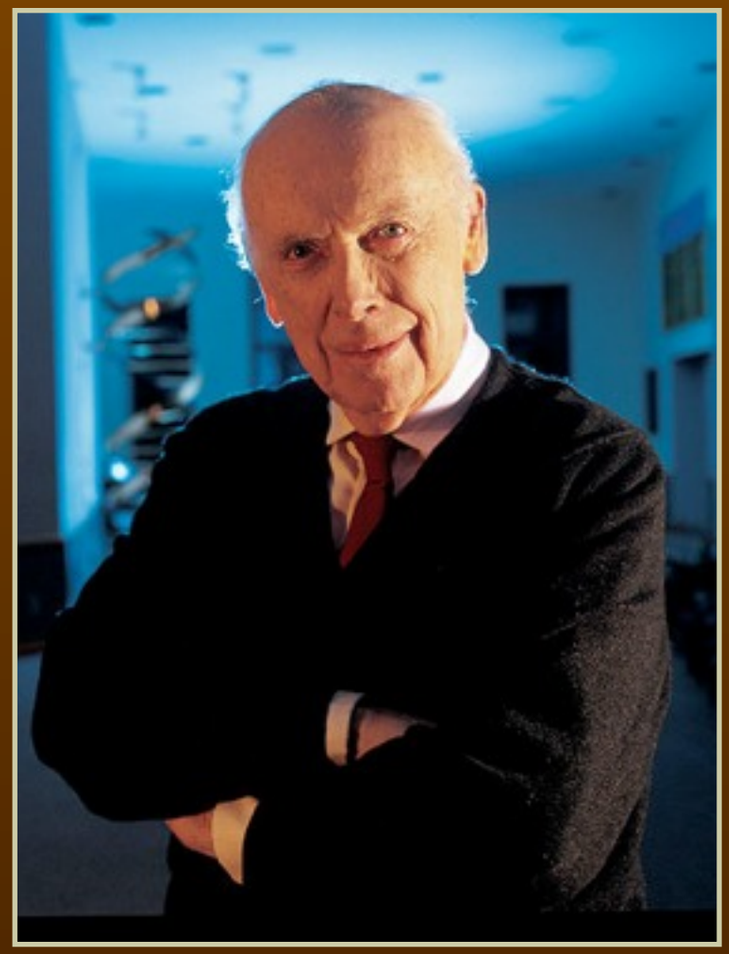


(*Source:* Marc Lieberman)



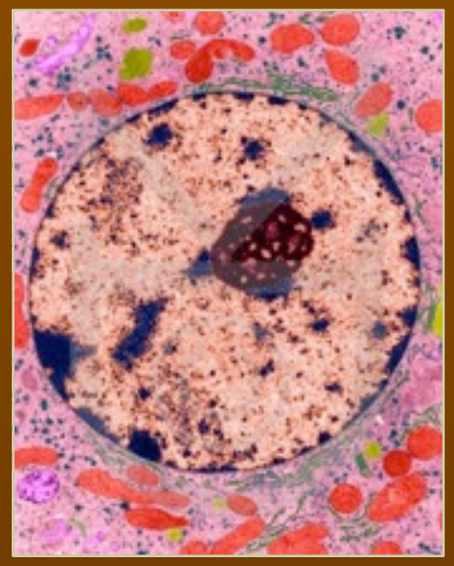
(*Source:* Michael Ströck)

James D. Watson



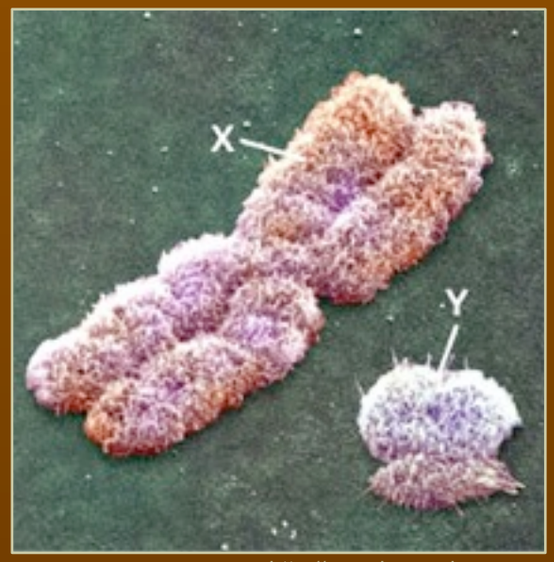
(*Source:* Cold Spring Harbor Laboratory)

Types of DNA



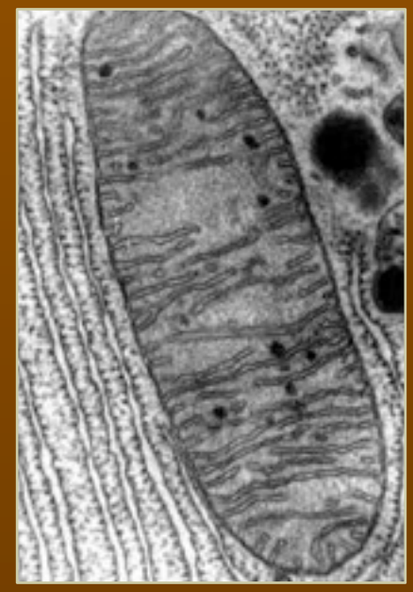
http://z.about.com

Nuclear DNA



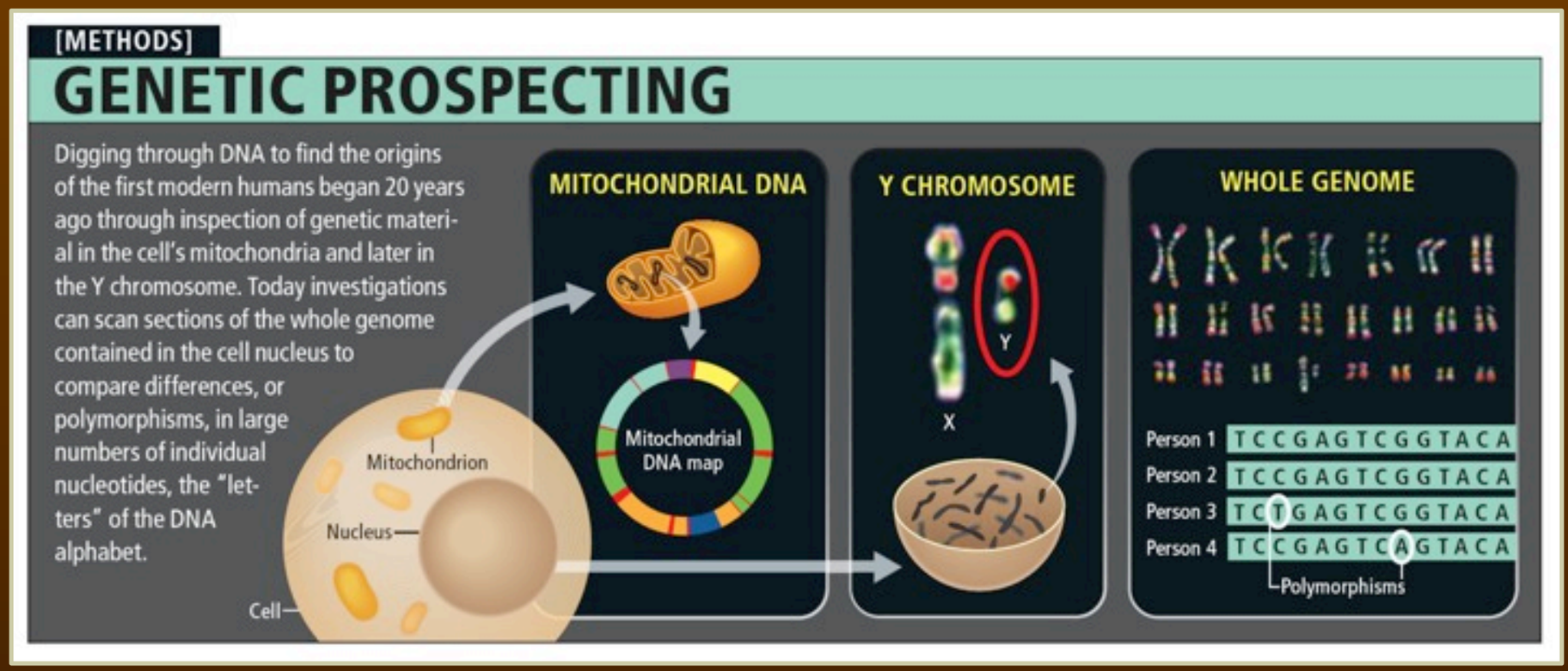
http://www.in-gender.com

Y-Chromosome Nuclear DNA



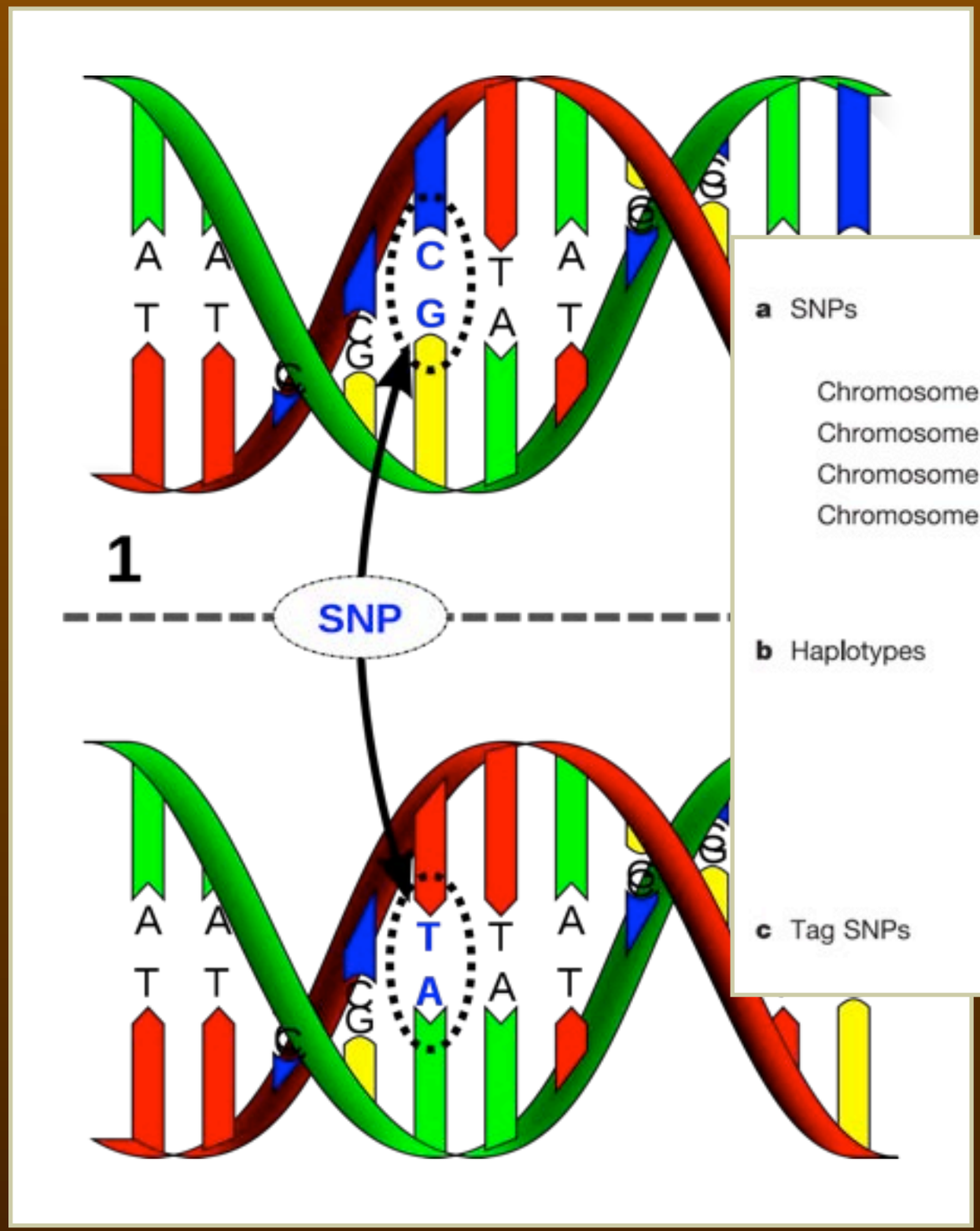
http://www.etsu.edu

Mitochondrial DNA

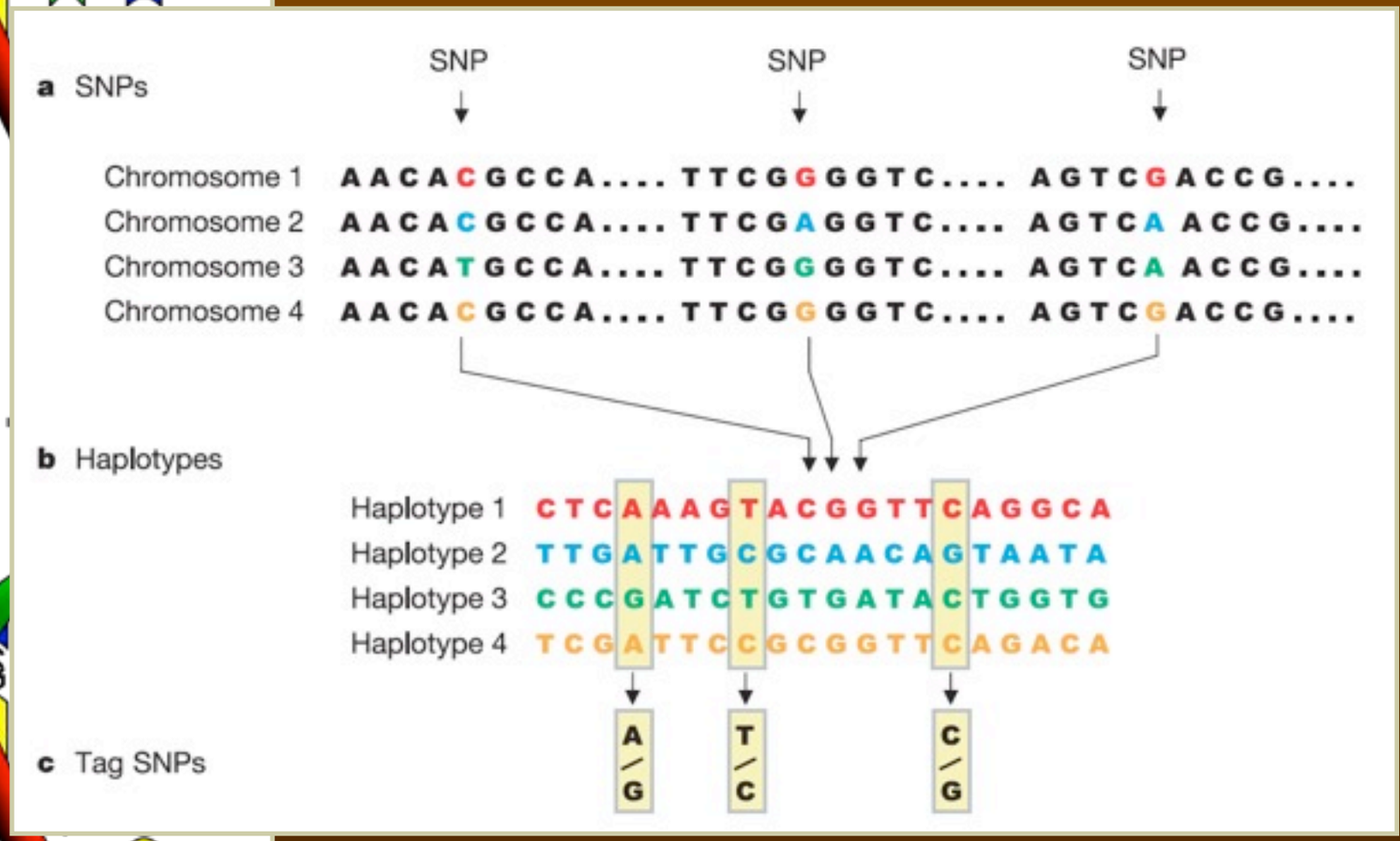


Source: Scientific American

Single Nucleotide Polymorphisms (SNP's)



(*Source:* David Hall)



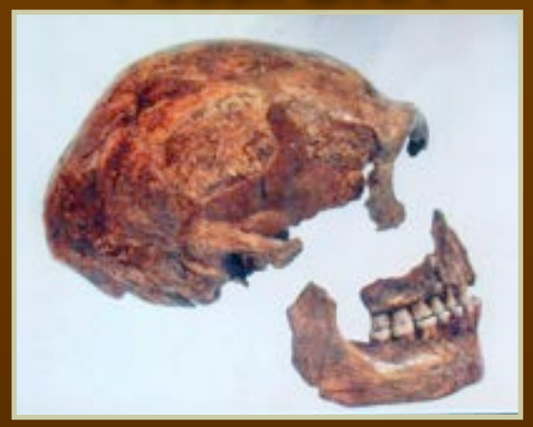
(*Source:* Nature/HapMap Project)

How did regional differences evolve?

Svante Pääbo

(*Source*: Public Library of Science)

Fossil DNA



- ★ ~2.5% of European and Chinese genomes is in common with Neanderthals.
- ★ ~5% of Papua New Guinea genome is in common with Denisovans.

https://genographic.nationalgeographic.com

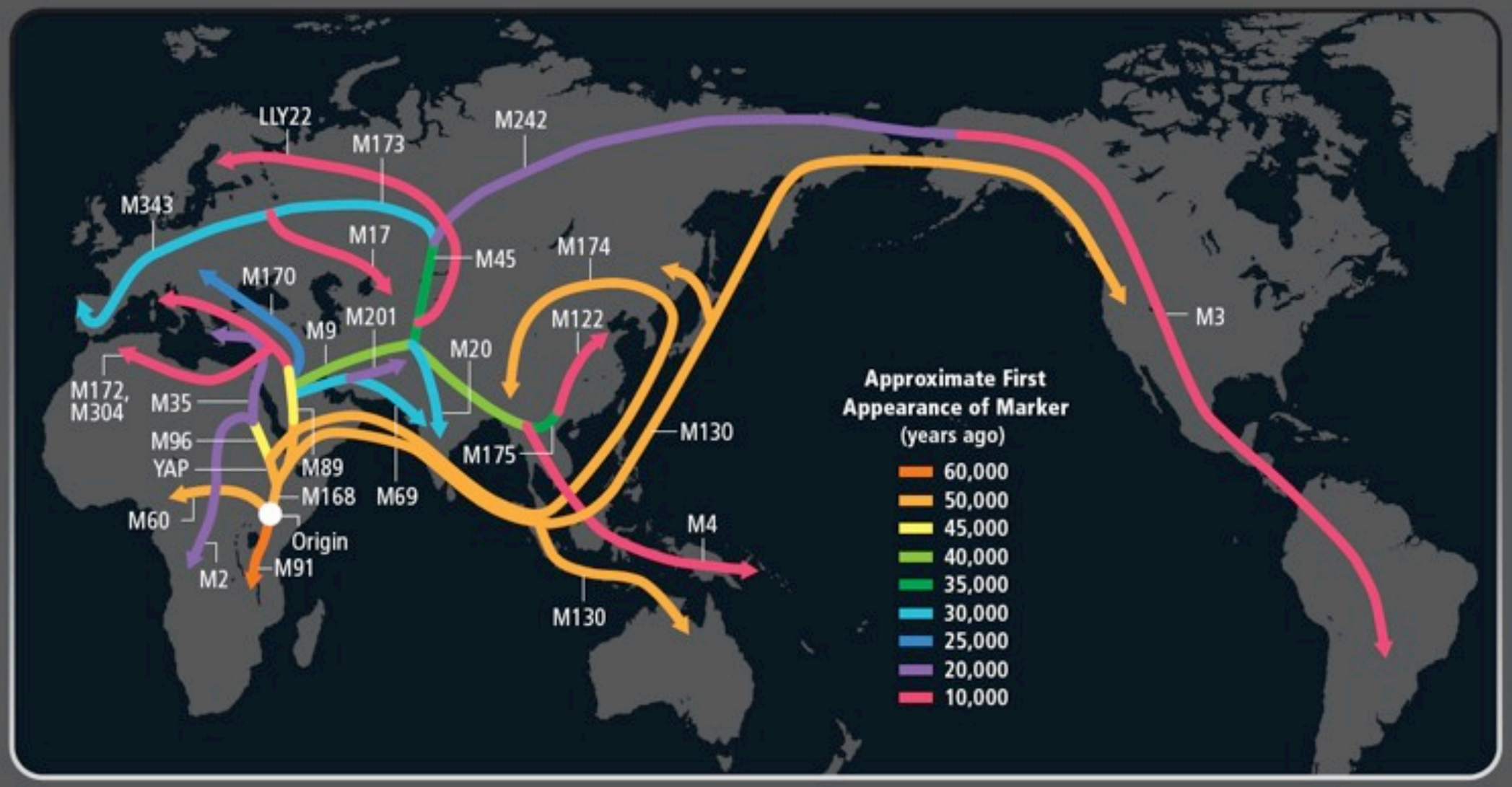
(*Source*: Scientific American)

Y Chromosome:

[ROUTE MAPS]

TRACKING Y CHROMOSOMES THROUGH TIME

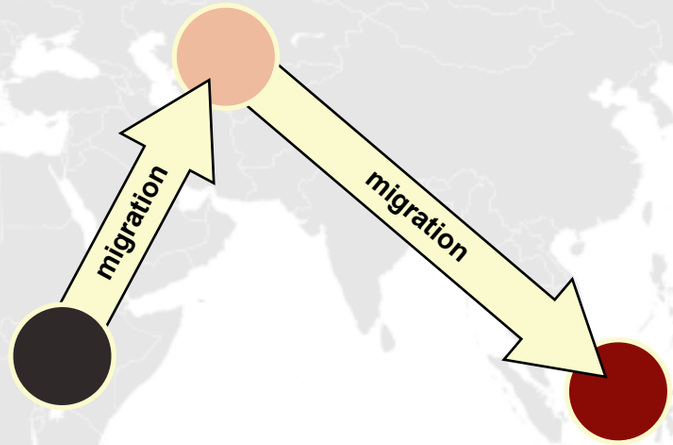
Geneticists can track the path of ancient migrations by examining genetic markers in Y chromosomes from men who hail from different parts of the world. Each marker, such as M168 or M89, identifies a lineage of men and where the lineage originated. By building an evolutionary tree based on observing many living people with the markers, investigators can determine the approximate ages of the lineages.



Source: Scientific American

Skin color as a marker of origin?

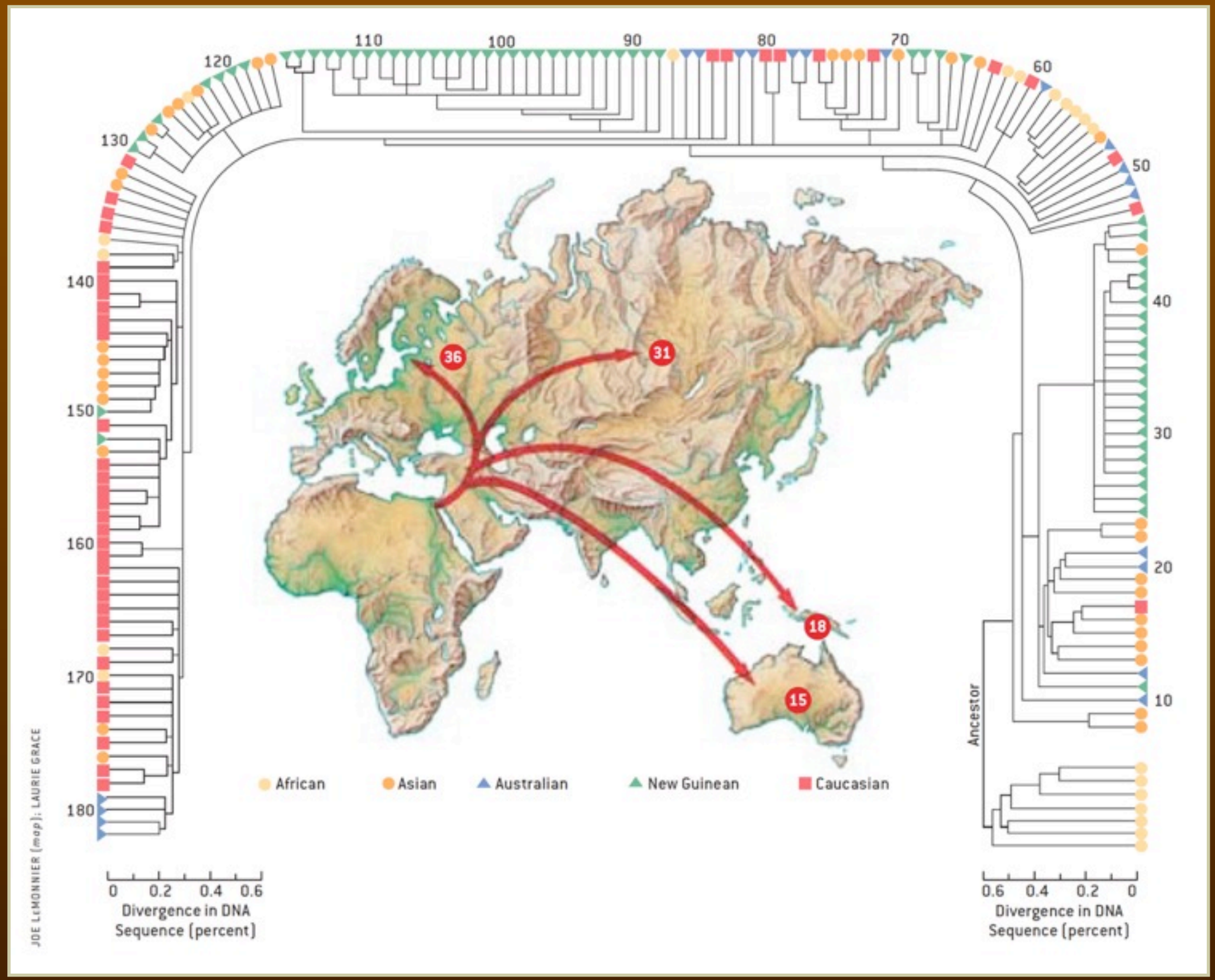
Source: California Academy of Sciences



(*Source:* Wikimedia Commons)

Nina Jablonski

Do regional ethnicities represent human subspecies?



Source: Scientific American

(*Source:* Wikimedia Commons)

184 Eurasians

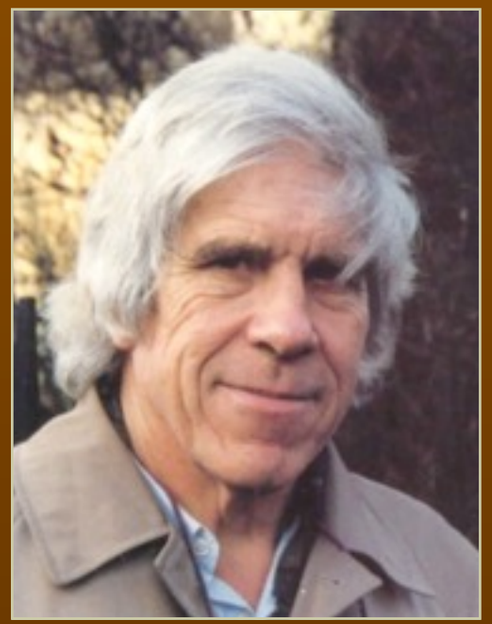
185 Africans

How genetically distinct are current-day African and Eurasian populations?

38,877,749 variable sites (SNPs)
100% differentiation in genetic code = 0 sites
95% differentiation in genetic code = 12 sites

this slide was adapted from **Svante Pääbo**: http://www.ted.com/talks/svante_paeaebo_dna_clues_to_our_inner_neanderthal.html

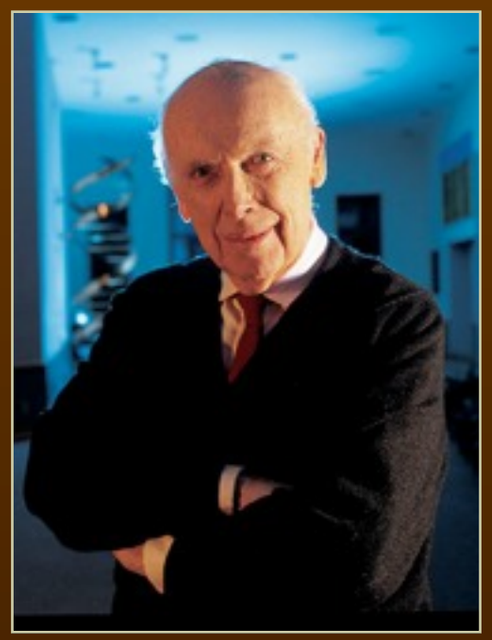
Reasons to worry about genomics?



1936-2000

Hamilton left behind him the almost completed manuscript of the second volume of his autobiography and this contained ... a foreword expressing his belief that only a radical programme of infanticide, eugenics, and euthanasia could save the human race from an imminent catastrophe. "I predict that in two generations the damage being done to the human genome by the ante and post-natal life-saving efforts of modern medicine will be obvious to all."

(*Source:* http://lclane2.net/hamilton.html)



1928-

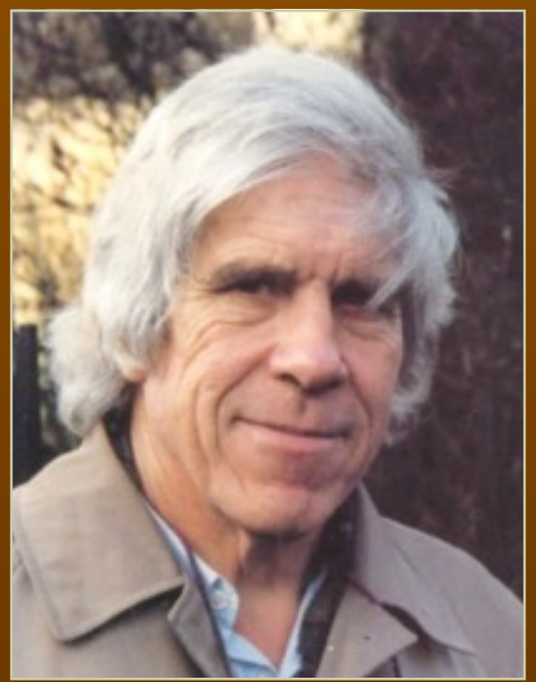
"[I am] inherently gloomy about the prospect of Africa [because] all our social policies are based on the fact that their intelligence is the same as ours—whereas all the testing says not really."

(*Source:* http://www.thedailybeast.com/newsweek/galleries/2010/04/07/smart-people-dumb-quotes.html)

Witnesses were flabbergasted when the 72-year-old discoverer of the double helix suggested there was a biochemical link between exposure to sunlight and sexual urges. “That's why you have Latin lovers,” Watson said. “You've never heard of an English lover. Only an English patient.”

(*Source:* http://www.sfgate.com/cgi-bin/article.cgi?file=/chronicle/archive/2000/11/13/MN111208.DTL)

Why do these prominent scientists espouse eugenic beliefs?

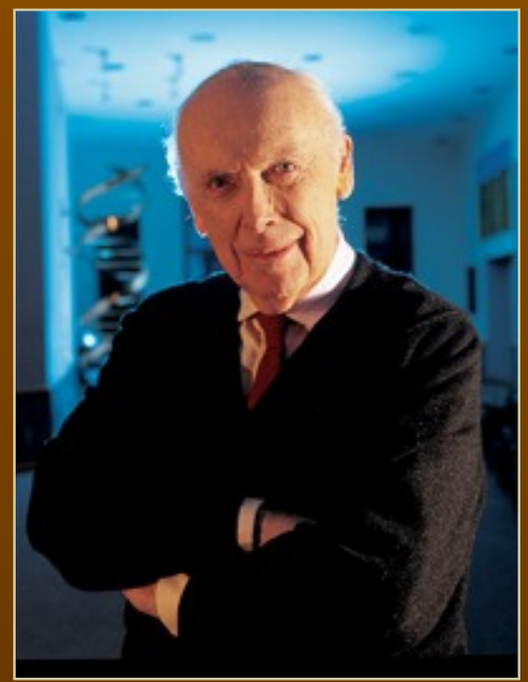


Both take a very reductionist, gene-centered approach:

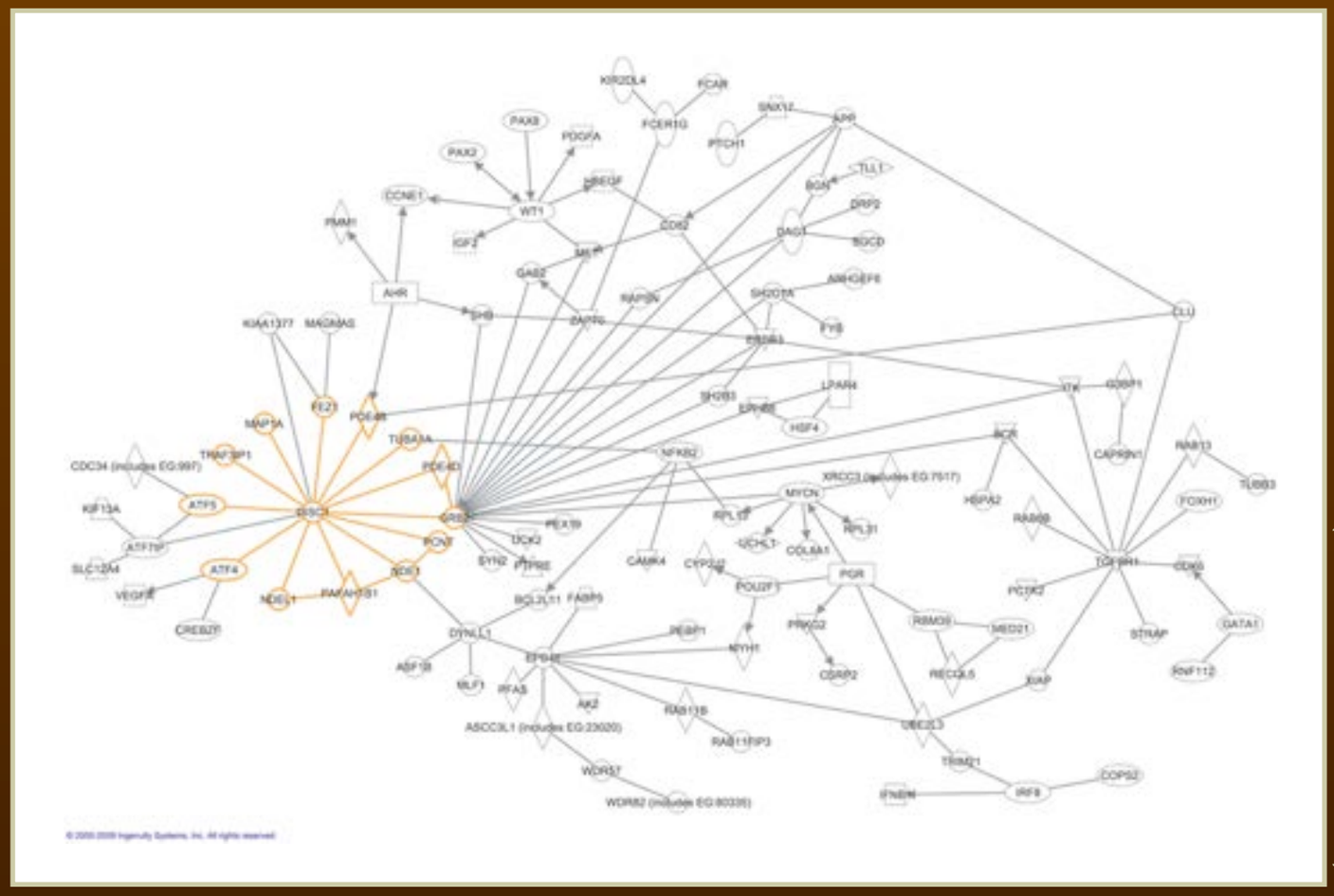
one gene — *produces* → *one trait*

gene var 1

gene var 2 ✗



In reality, genes produce traits through complex interactions:



We have very primitive theory for understanding these complex interaction networks

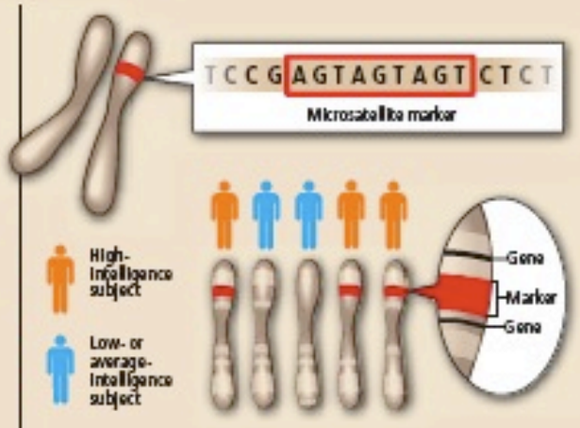
(*Source:* Porteous Hennah)

How will gene-screening technologies be used?

[TECHNIQUES]

HUNTING FOR GENES

Investigators have used various methods over the years to search for genes that might contribute to intelligence, which is a so-called quantitative trait—one present in all study subjects to greater or lesser degrees. Comparing the DNA of highly intelligent people with one another and with those of average or low intelligence can reveal patterns common to subjects of high intelligence. Those signatures, in turn, can flag the location of genes that influence intelligence levels, although such experiments have yet to conclusively identify any "intelligence genes."

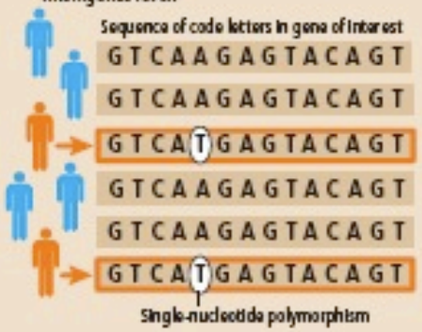


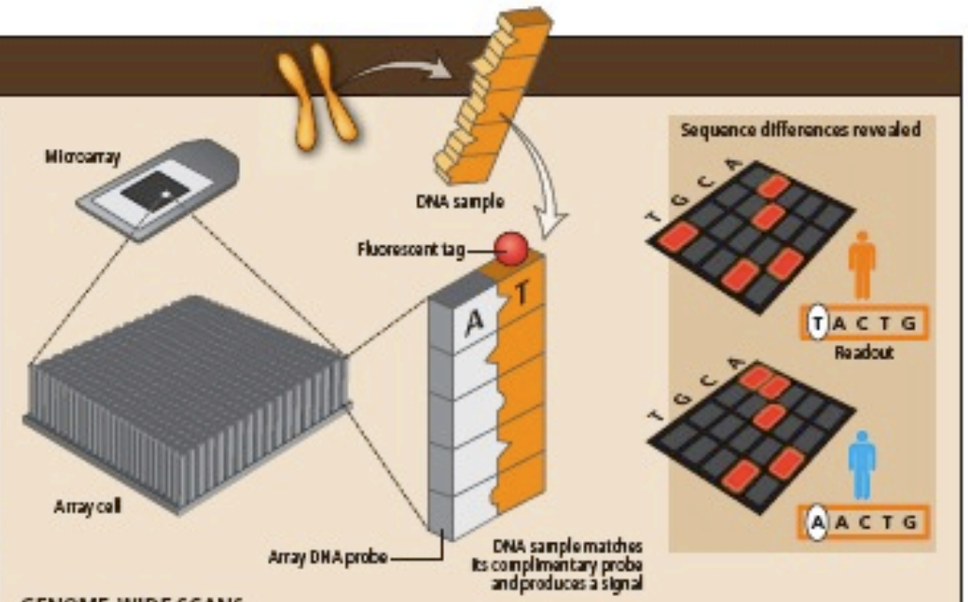
QUANTITATIVE TRAIT LOCI (QTL)

To find a chromosomal region, or "locus," implicated in a quantitative trait, scientists look first for repeated DNA sequences called microsatellite markers interspersed along the length of chromosomes. If particular markers appear more frequently in high-intelligence subjects, researchers will scan nearby DNA to identify neighboring genes.

CANDIDATE GENES

To find signs that a QTL gene or another gene already known to affect cognitive processes such as memory influences intelligence, scientists can compare the gene's DNA sequence in high- and low-intelligence subjects. If the same variations, called single-nucleotide polymorphisms (SNPs), appear more frequently in high-intelligence subjects, the pattern suggests the gene may contribute to intelligence level.



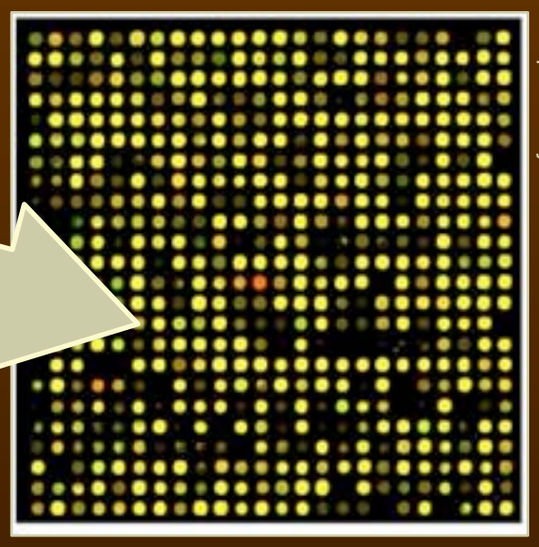


GENOME-WIDE SCANS

To identify new candidate genes, scientists can use microarrays to search the entire human genome for SNPs. Each cell of the array contains short DNA strands designed to pair with a particular sequence in a gene or gene-regulating region. When DNA samples are washed across the array, matches cause the cell to fluoresce (*red*). The single-nucleotide differences revealed in the DNA sequences of high-intelligence subjects and others can point to a gene, and a particular variant of it, that may contribute to intelligence.

(*Source:* Scientific American)

http://www.pgbeautygroomingscience.com



http://www.jarvislab.net

- ★ 7,000 people screened for IQ and compared over 500,000 SNPs.
- ★ Only six SNPs showed any effect on IQ score.
- ★ The strongest contributions of any one of these SNPs was 0.4%; the combined effect of the six SNPs discovered was 1.2%.

Data is publicly available:

International HapMap Project

International HapMap Project

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Guidelines for the Responsible Use and Publication of HapMap data is available **here**.

Browse data graphically

Use the **Generic Genome Browser** to view HapMap Project data in the context of other genomic features, as well as retrieve genotypes & frequencies for specific genomic regions.

Generate reports and extracts of data using **HapMart**.

Jump directly to chromosome -------- in the current dataset. Submit

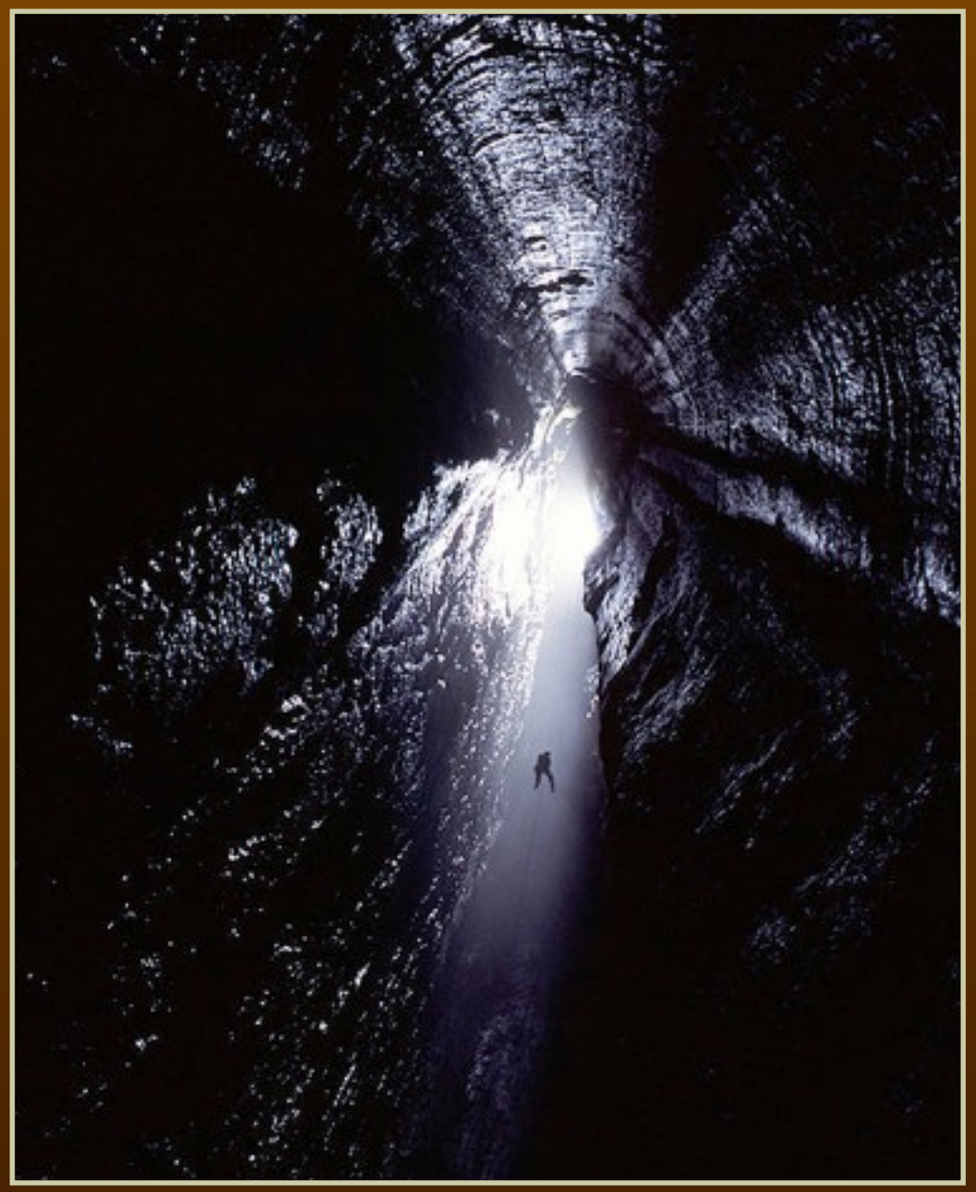
Downloads

The following directories contain HapMap project related data, software, and documentation, that have been made publicly available. (See **HapMap Data Access Policy** for more information). More details about each dataset can be found in READMEs in the respective directories:

- **Bulk data**
  - **Genotypes**: Individual genotype data submitted to the DCC to date. **Phase 3** data is available in **PLINK format** and **HapMap format**.
  - **Frequencies**: Allele & genotype frequencies compiled from genotyping data submitted to the DCC to date. These have also been submitted to **dbSNP** and should be available in the next dbSNP build.
  - **LD Data**: Linkage disequilibrium properties D', LOD , R^2 compiled from the genotype data to date
  - **Phasing Data**: Phasing data generated using the PHASE software, compiled from the genotype data to date.
  - **Allocated SNPs**: dbSNP reference SNP clusters that have been picked and prioritized for genotyping according to several criteria (see info on **how SNPs were selected**). The file 00README contains per-chromosome SNP counts and further details.
  - **CNV Genotypes**: CNV data from HapMap3 samples.
  - **Recombination rates and Hotspots**: Recombination rates and hotspots compiled from the genotyping data.
  - **SNP assays**: Details about assays submitted to the DCC to date. PCR primers, extension probes etc., specific to each genotyping platform.
  - **Perlegen amplicons**: Details for mapping Perlegen amplicons to HapMap assayLSID. For primer sequences, see **Perlegen's** Long Range PCR Amplicon data.
  - **Raw data**: Raw signal intensity data from HapMap genotypes. Currently includes data from Affymetrix GeneChip 100k and 500k Mapping Arrays.
  - **Inferred genotypes**: Genotypes inferred using the method of Burdick et al. Nat Genet 38:1002-4.
  - **Mitochondrial and chrY haplogroups**: Classification of phase I HapMap samples into mtDNA and chrY haplogroups. The distribution shown in Table 4 of the HapMap phase I paper *(Nat Genet 38:1002-4)* corresponds to unrelated parents in each one of the populations analyzed.

www.hapmap.org/

What impact will science have on our future understanding of human variation?



(*Source:* Antonio Danieli)

(*Source:* Hannes Grobe)

(*Source:* Andrew Hurley)

(*Source:* Roberto Lumbreras)