

# The origin and spread of *Homo sapiens*

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## 1. What drives evolution?

*Chance and Necessity: Random Variation + Selection;*

*Chance alone: Random drift, “Neutral molecular” evolution*

## 2. The logic of exponential growth.

*“Everyone is related to everyone else”: the molecular clock and coalescence.*

## 3. Genes and the evolution of culture.

# Sources of data

Fossil evidence

Morphology and embryology

Molecular Biology

*Neutral genetic markers, genes under selection*

1. Where does chance come into evolution?

2. What are the likely consequences?

# Mutation

Mutation rates per genome per replication  
in lytic RNA viruses

Virus	Median $\mu_g$
Bacteriophage Q $\beta$	6.5
Poliovirus	0.8
Vesicular stomatitis virus	3.5
Influenza A	$\geq 1.0$

*Rates of Spontaneous Mutation*

(Drake, B Charlesworth, D Charlesworth and Crow, Genetics 148: 1667–1686;  
April 1998)

# Mutation rates

Mutation rates estimated from specific loci in higher eukaryotes

Organism	$G$	$G_e$	$\mu_b$	$\mu_g$	$\mu_g$	$\mu_{gs}$
<i>C. elegans</i>	$8.0 \times 10^7$	$1.8 \times 10^7$	$2.3 \times 10^{-10}$	0.018	0.004	0.036
Drosophila	$1.7 \times 10^8$	$1.6 \times 10^7$	$3.4 \times 10^{-10}$	0.058	0.005	0.14
Mouse	$2.7 \times 10^9$	$8.0 \times 10^7$	$1.8 \times 10^{-10}$	0.49	0.014	0.9
Human	$3.2 \times 10^9$	$8.0 \times 10^7$	$5.0 \times 10^{-11}$	0.16	0.004	1.6

These are all likely to be underestimates, because not all kinds of mutations are included, particularly those with minor, inconspicuous effects.

The human genome picks up about 1.6 mutations in each generation.

(Most mutations occur in males)

# Recombination



	RAT	MOUSE	HUMAN
Length of genetic map cM	1509 cM	1361 cM	3615
Genome size Gb	2.72 Gb	2.58 Gb	3.02
Recombination rate (cM/Mb; genome-wide average)	0.555	0.528	1.20

“Guarantee” of crossover event in each chromosome in each meiosis;  
 $2^{20} \sim 10^6$  different haploid products of meiosis (correct for heterozygosity)

# Drift

(the neutral theory)

# Consequences of drift

Probability that a newly arisen beneficial allele is fixed ~

$$2s$$

for a recessive allele and small  $s$

(a) Advantageous mutations can be lost

(b) Disadvantageous mutations can be fixed

# Consequences of drift

Effective intensity of selection depends on population size:  $4Ns$

Selection can be 'strong' or 'weak'  
depending on the size of the breeding pool

# Consequences of drift

Probability that a newly arisen neutral allele is fixed =  $1/2N$

Overall rate of fixation = number of neutral alleles that arise in each generation x probability of fixation

=

Mutation rate per locus x no. of alleles x prob of fixation

=

Mutation rate x  $2N$  x  $1/2N$

=

Mutation rate

# The logic of exponential growth

*Homo sapiens:*

150,000 years = 7500 generations

*Average growth by 0.1% per generation:*

8-fold increase over 7500 generations

*Smaller organisms:*

More absurd implications

*Inference:* Over long times the average growth rate of any species must be close to zero

GENERATIONS	NUMBERS OF ANCESTORS	YEARS AGO IN HUMAN
50	1,126,588,362,522,624	1,000 YEARS
40	1,100,183,947,776	800 YEARS
30	1,073,421,824	600 YEARS
20	1,048,576	400 YEARS
19	524,288	
18	262,144	
17	131,072	
16	65,536	
15	32,768	300 YEARS
14	16,384	
13	8,192	
12	4,096	
11	2,048	
10	1,024	200 YEARS
9	512	
8	256	
7	128	
6	64	
5	32	100 YEARS
4	16	
3	8	
2	4	
1	2	

Ohno, *What prevents the exponential increase of one's ancestors?*  
*Proc. Natl. Acad. Sci. USA* Vol. 93, pp. 15276–15278, December 1996

## Inference from 'constant' population sizes

1. Genetic ancestry is always smaller than ancestry by kinship.
2. You do not have to go far back to find that everyone is related to everyone else.



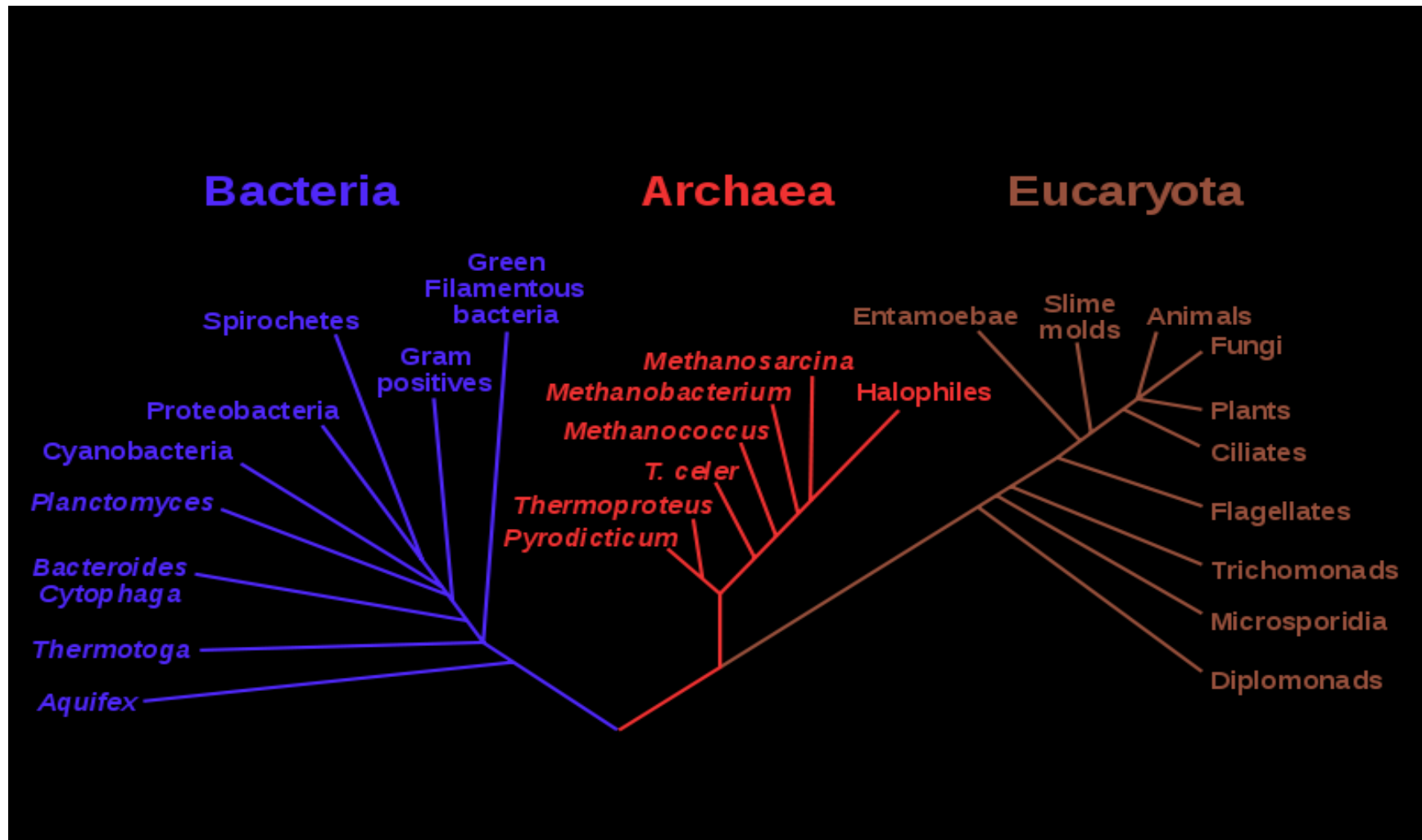
# Coalescence

How many genetic ancestors does an individual have?

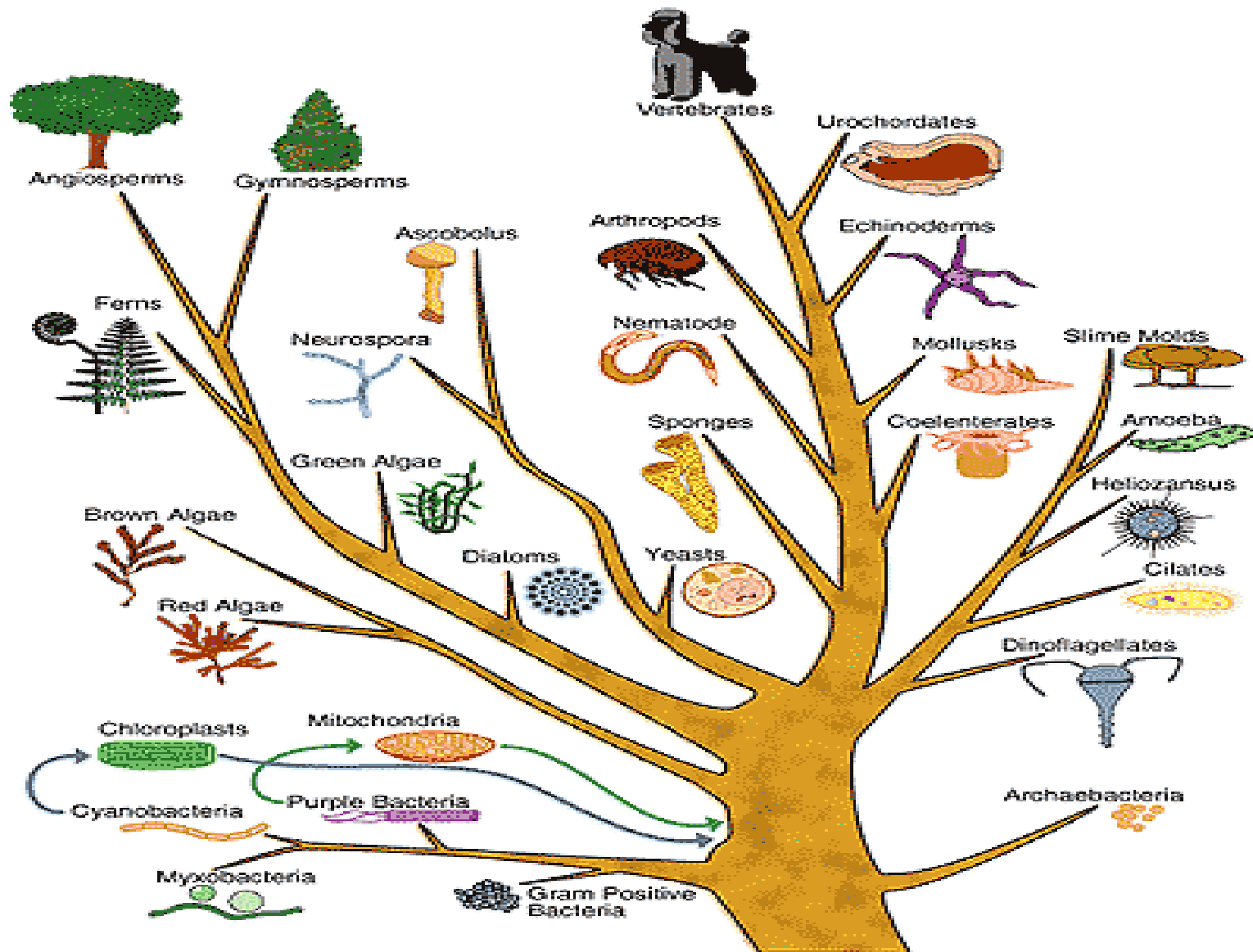
Sexually reproducing diploid, single chromosome, no recombination: exactly two, however far back you go.

**Generalise**

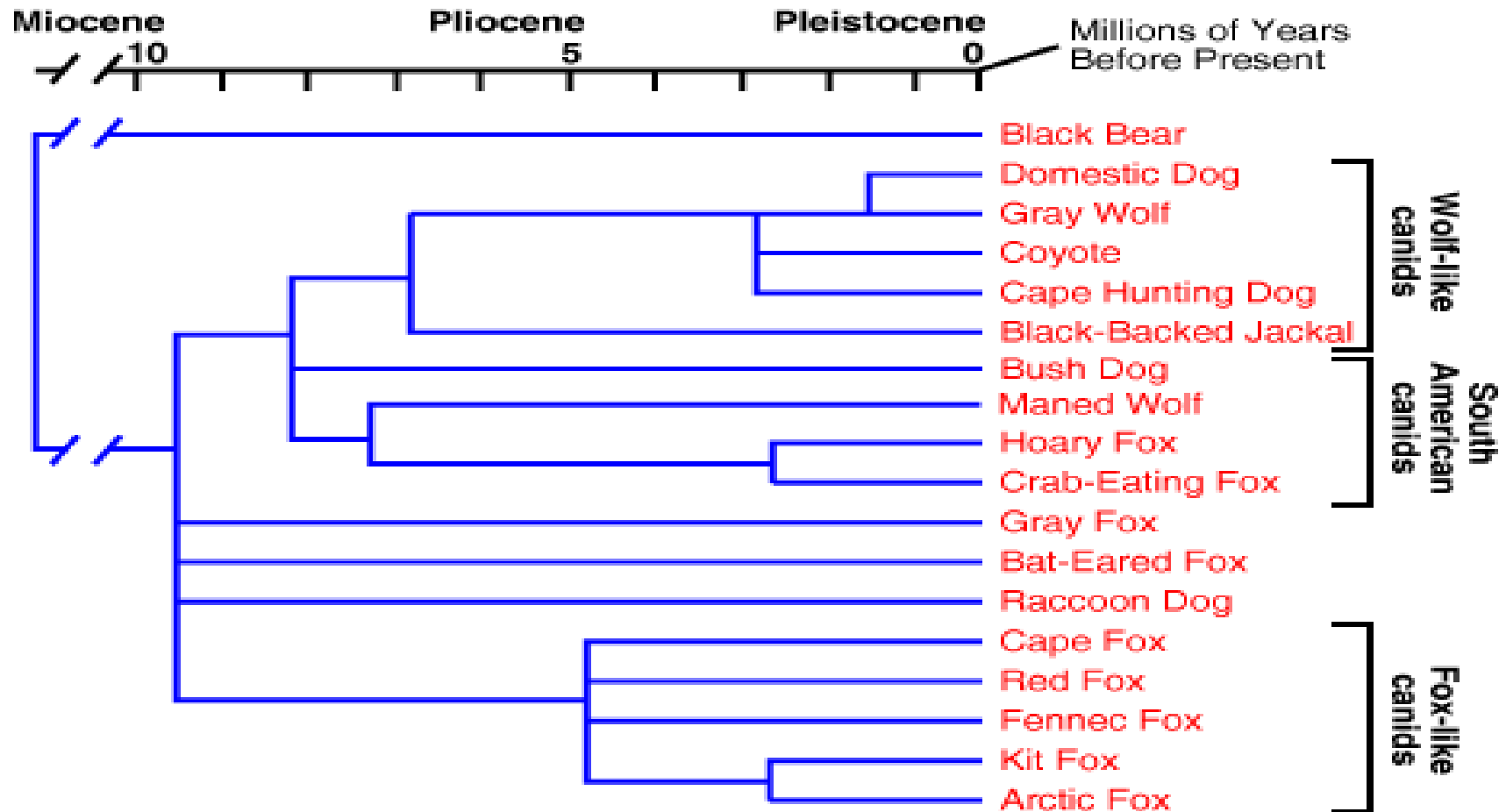
# Tree of life - 1



## Tree of life - 2

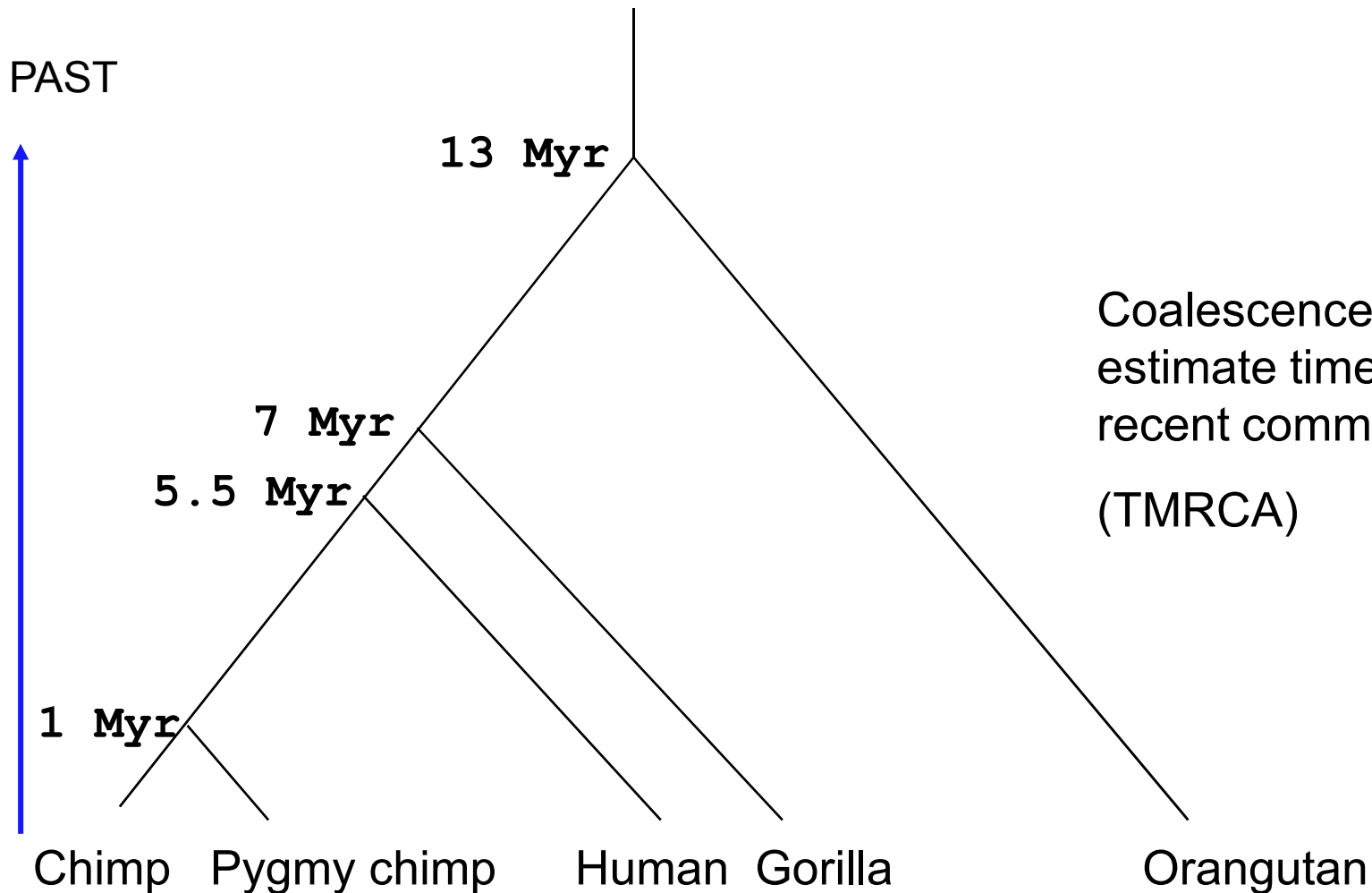


## Tree of life - 3



# Human beings are primates

From Nei, 2003



Coalescence studies can estimate time of most recent common ancestor (TMRCA)

# Living relatives



**Bonobo**



**Chimpanzee**



**Gorilla**



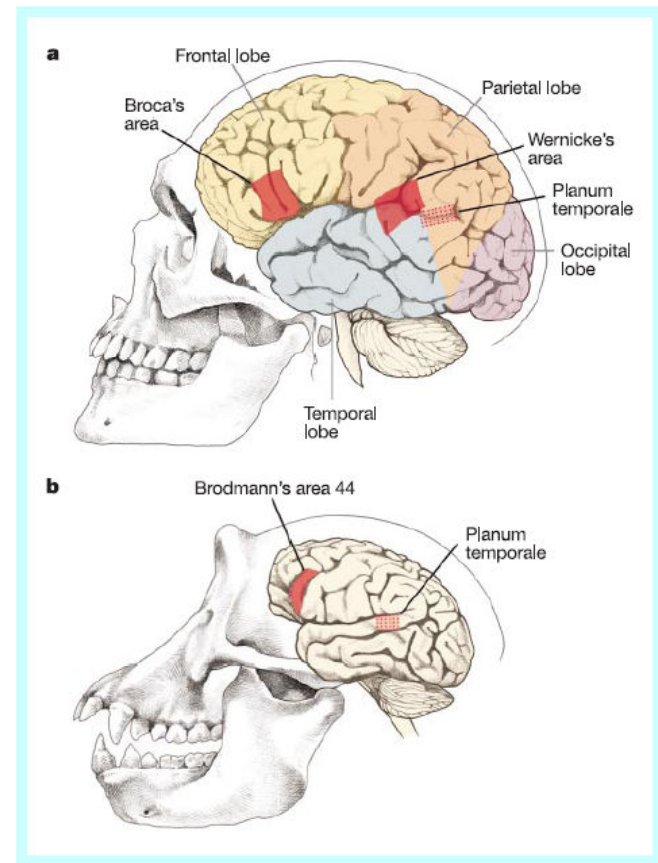
**Orangutan**

# Human-Ape differences

## **Selected traits that distinguish humans from other apes<sup>5-7</sup>**

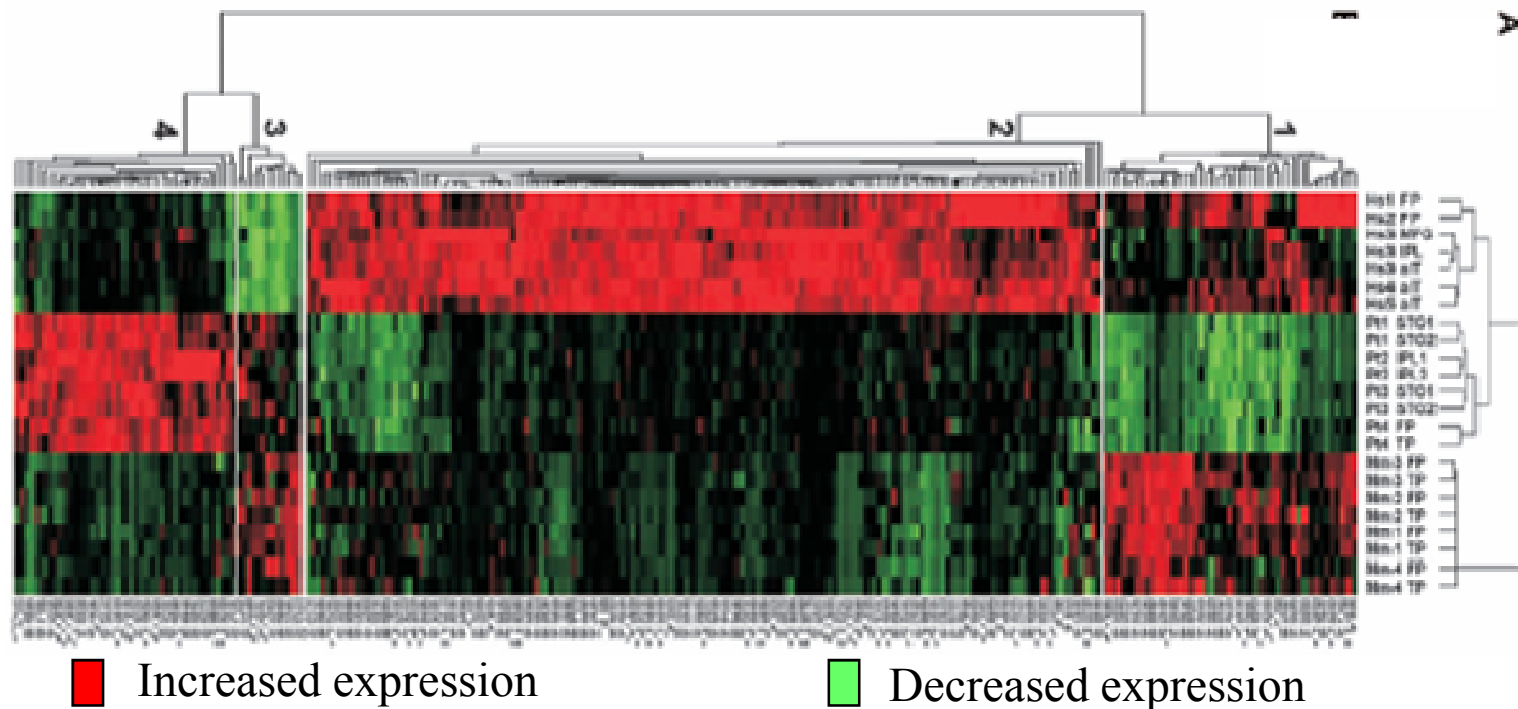
- Body shape and thorax
- Cranial properties (brain case and face)
- Relative brain size
- Relative limb length
- Long ontogeny and lifespan
- Small canine teeth
- Skull balanced upright on vertebral column
- Reduced hair cover
- Elongated thumb and shortened fingers
- Dimensions of the pelvis
- Presence of a chin
- S-shaped spine
- Language
- Advanced tool making
- Brain topology

Carroll (2003) *Nature* **422**, 849-857



# Gene expression differences in human and chimpanzee cerebral cortex

- Affymetrix oligonucleotide array (~10,000) genes
- 91 show human-specific changes, ~90% increases

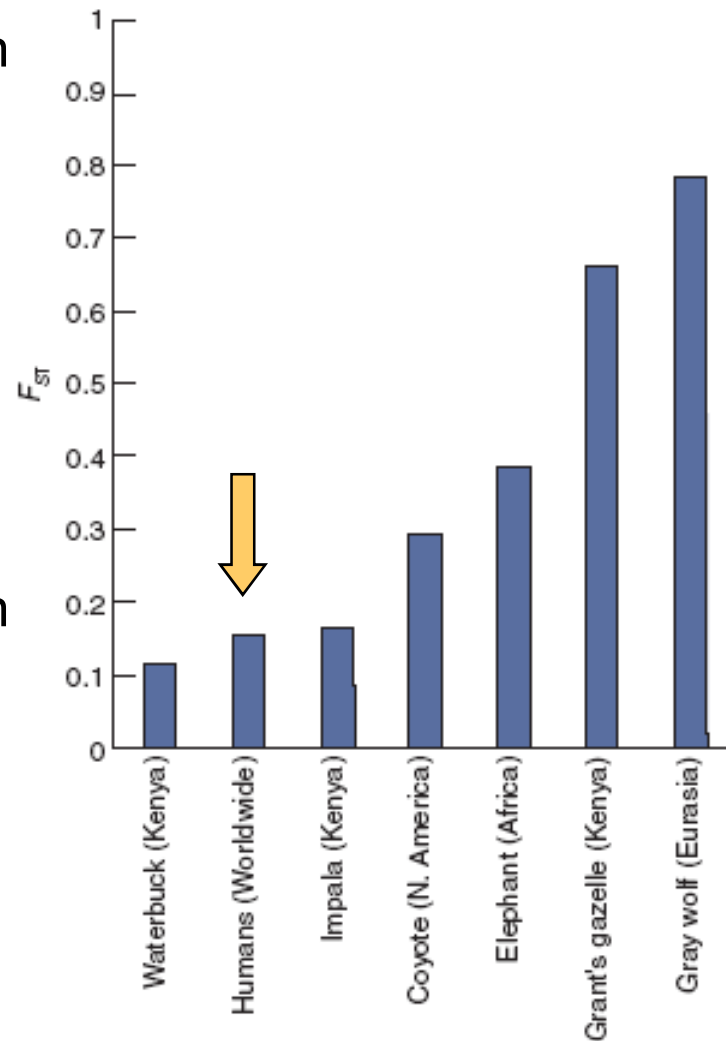




# *Homo sapiens*: most genetic diversity occurs within populations

Most variation  
**between**  
populations

Most variation  
**within**  
populations



Templeton (1999) *Am. J. Anthropol.* **100**, 632-650

# Origins of hominids

- *Sahelanthropus tchadensis*
- Chad (Central Africa)
- Dated to 6 – 7 million years ago
- Posture uncertain, but slightly later hominids were bipedal



‘Toumai’, Chad, 6-7 MYA

Brunet *et al.* (2002) *Nature* **418**, 145-151

## Origins of *Homo sapiens*

- Anatomically modern humans in Africa ~130 KYA
- In Israel by ~90 KYA
- Not enormously successful



Omo I, Ethiopia, ~130 KYA

# Why did people migrate?

*...Many reasons*

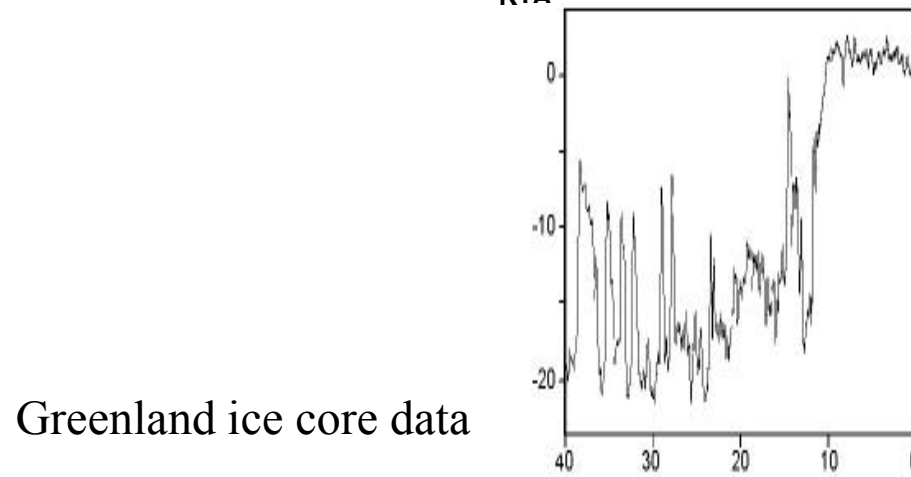
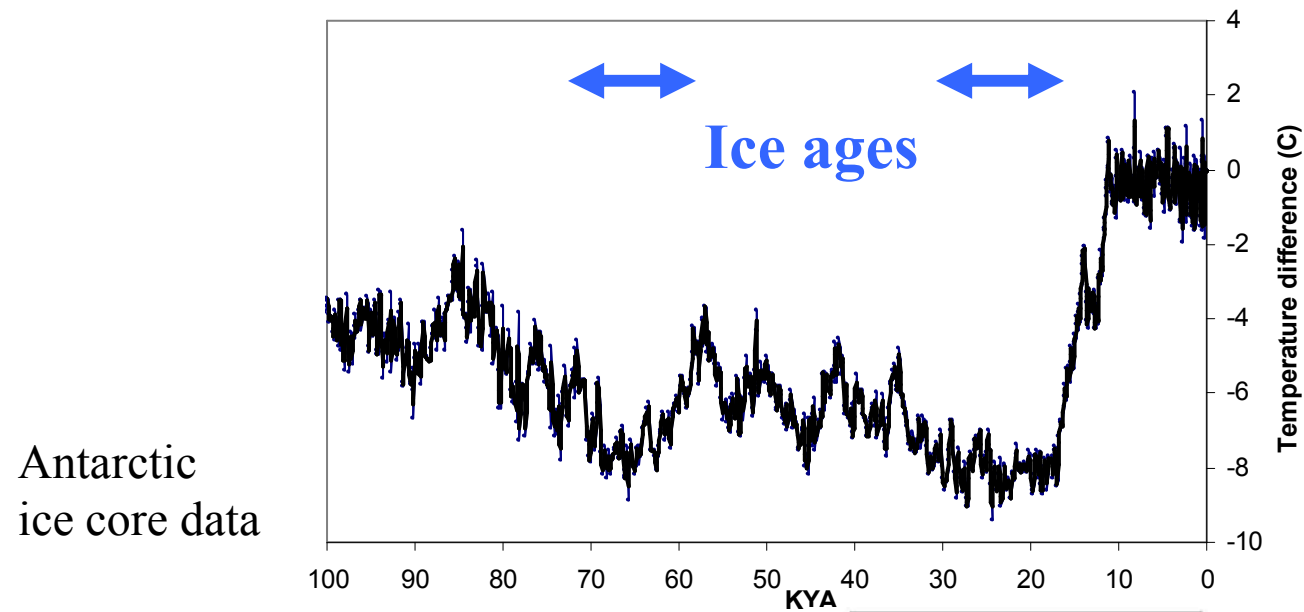
Climate change

Population pressure/Food shortages

Environmental degradation

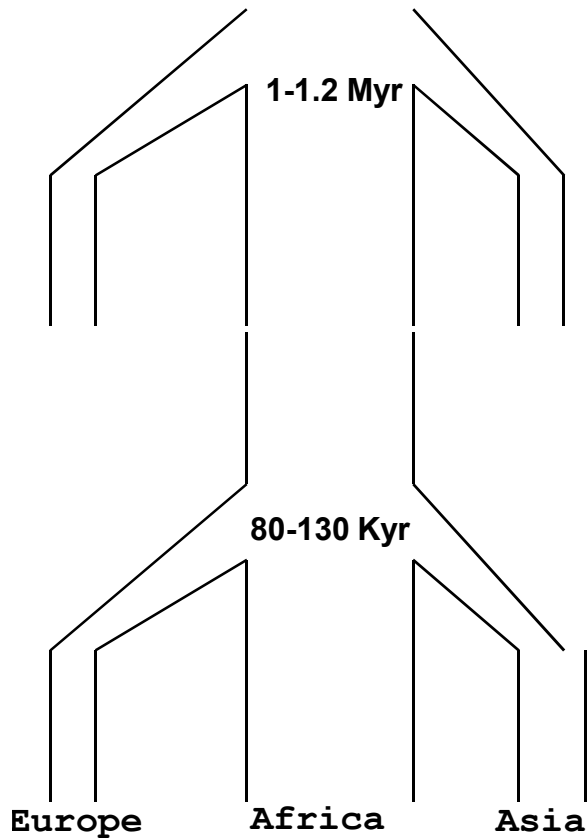
Curiosity?

# Fluctuations in climate

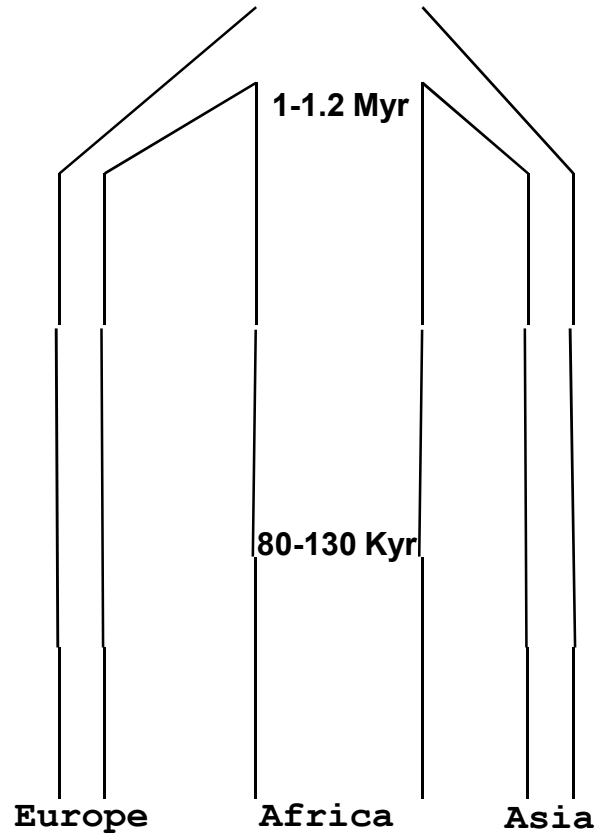


# Out-of-Africa? Independent origins?

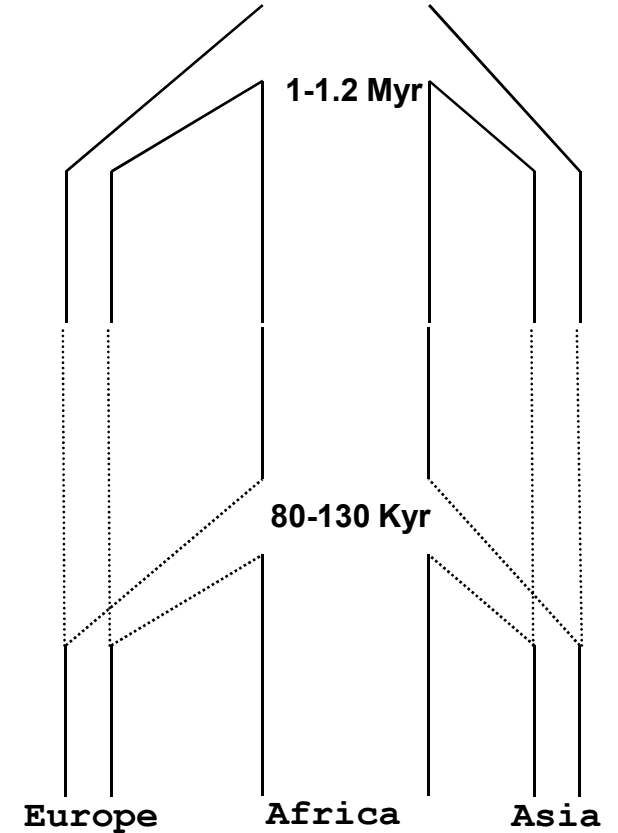
Total replacement



No replacement

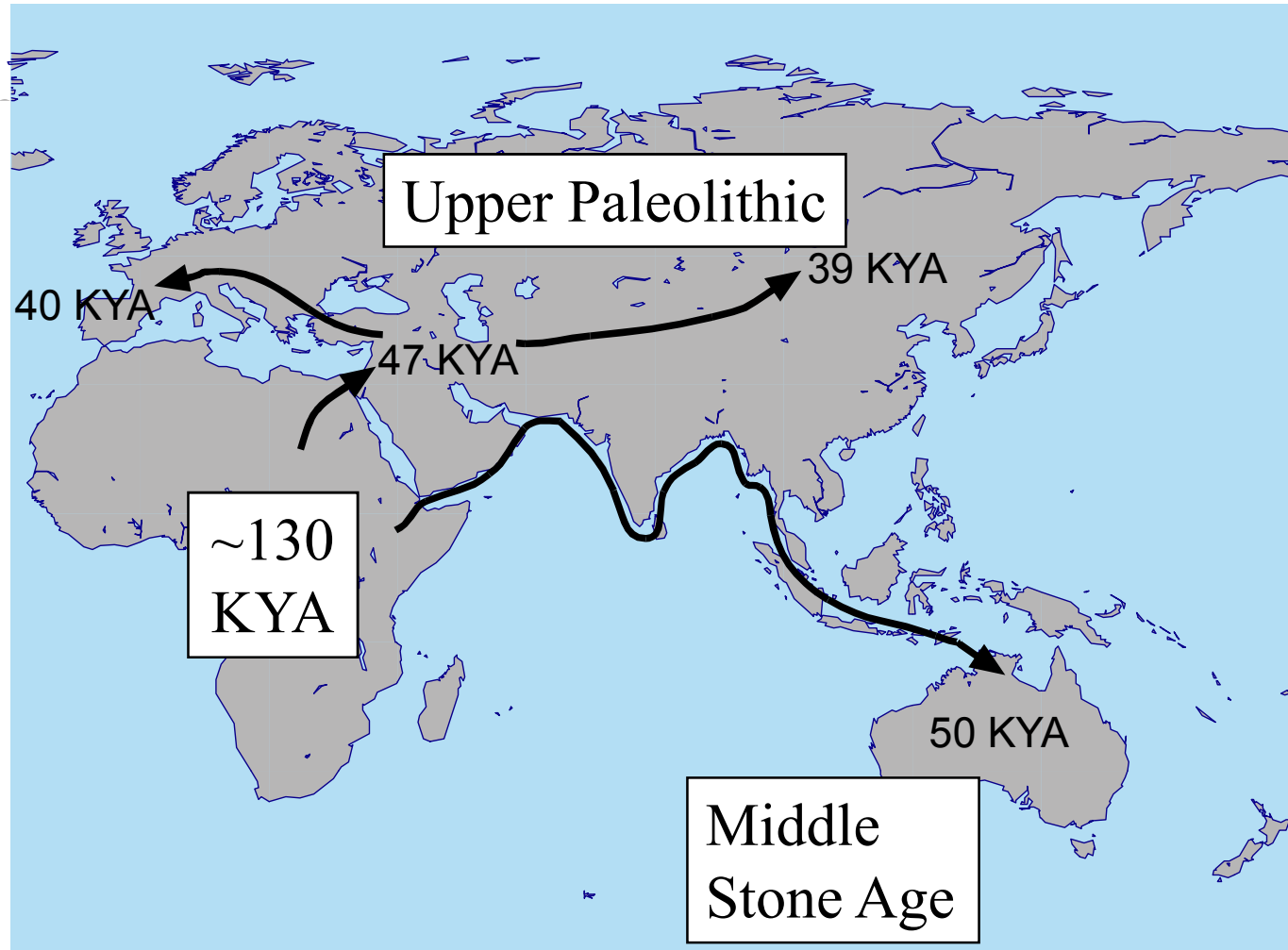


Partial replacement

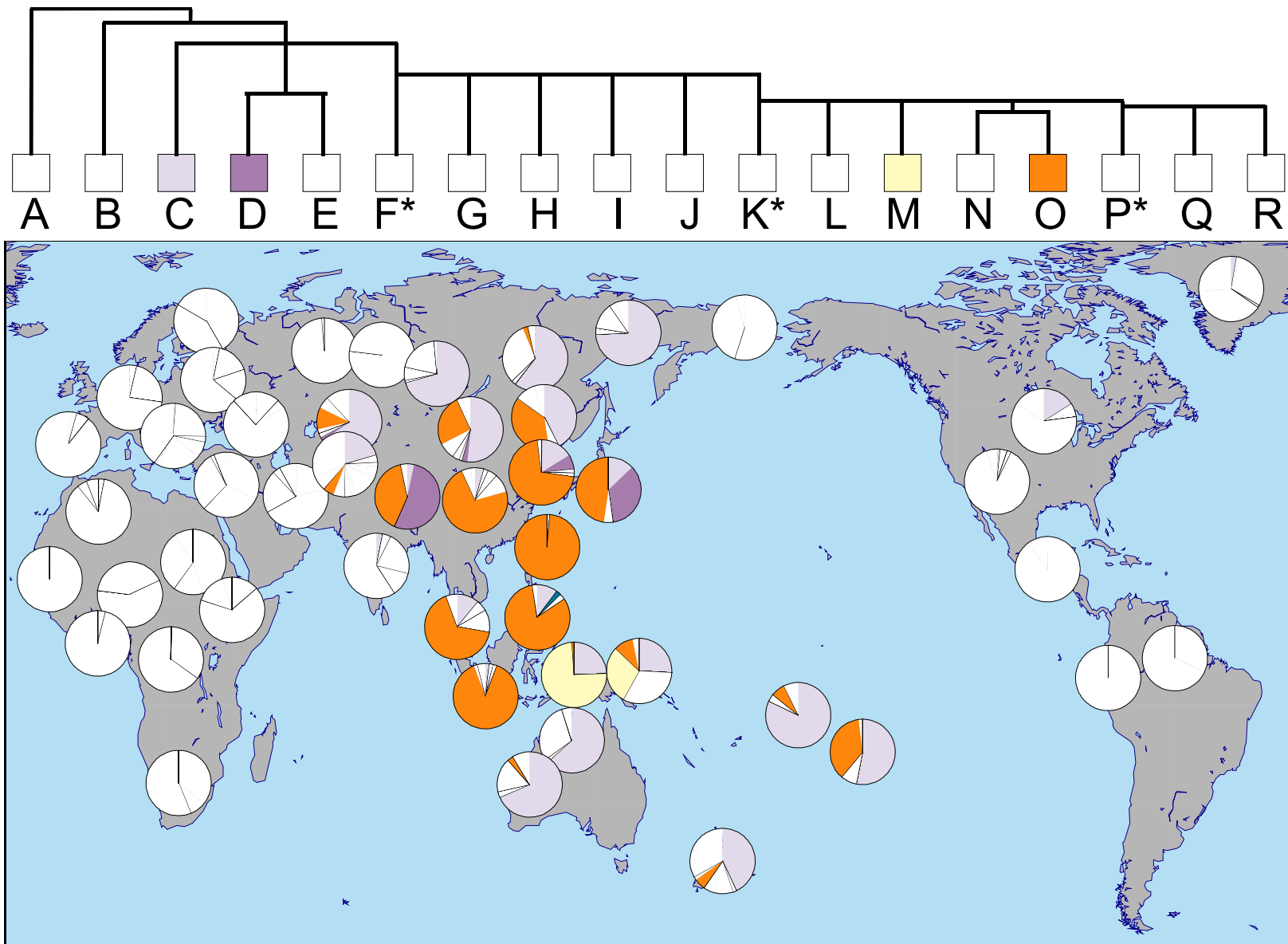


e.g.: Takahata (2001): data consistent with total replacement.

## Migrations: evidence from archaeology



# SE Y haplogroups



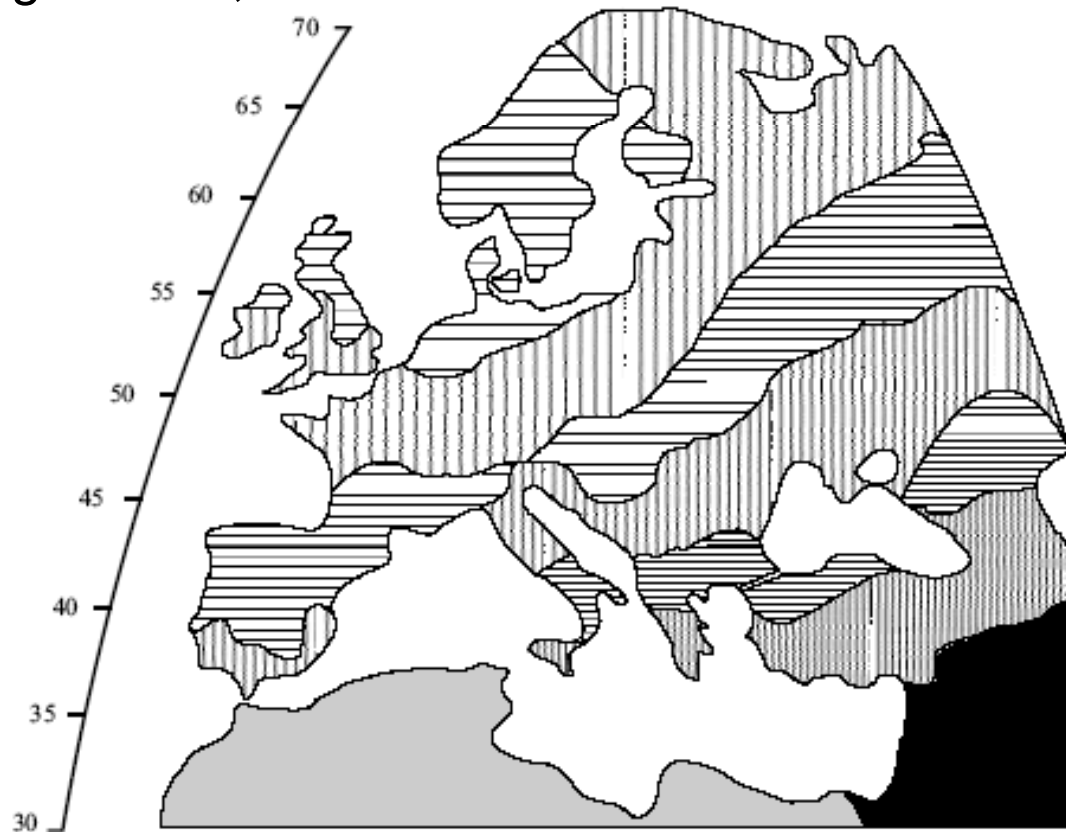


# Allele Frequencies and Principal Components

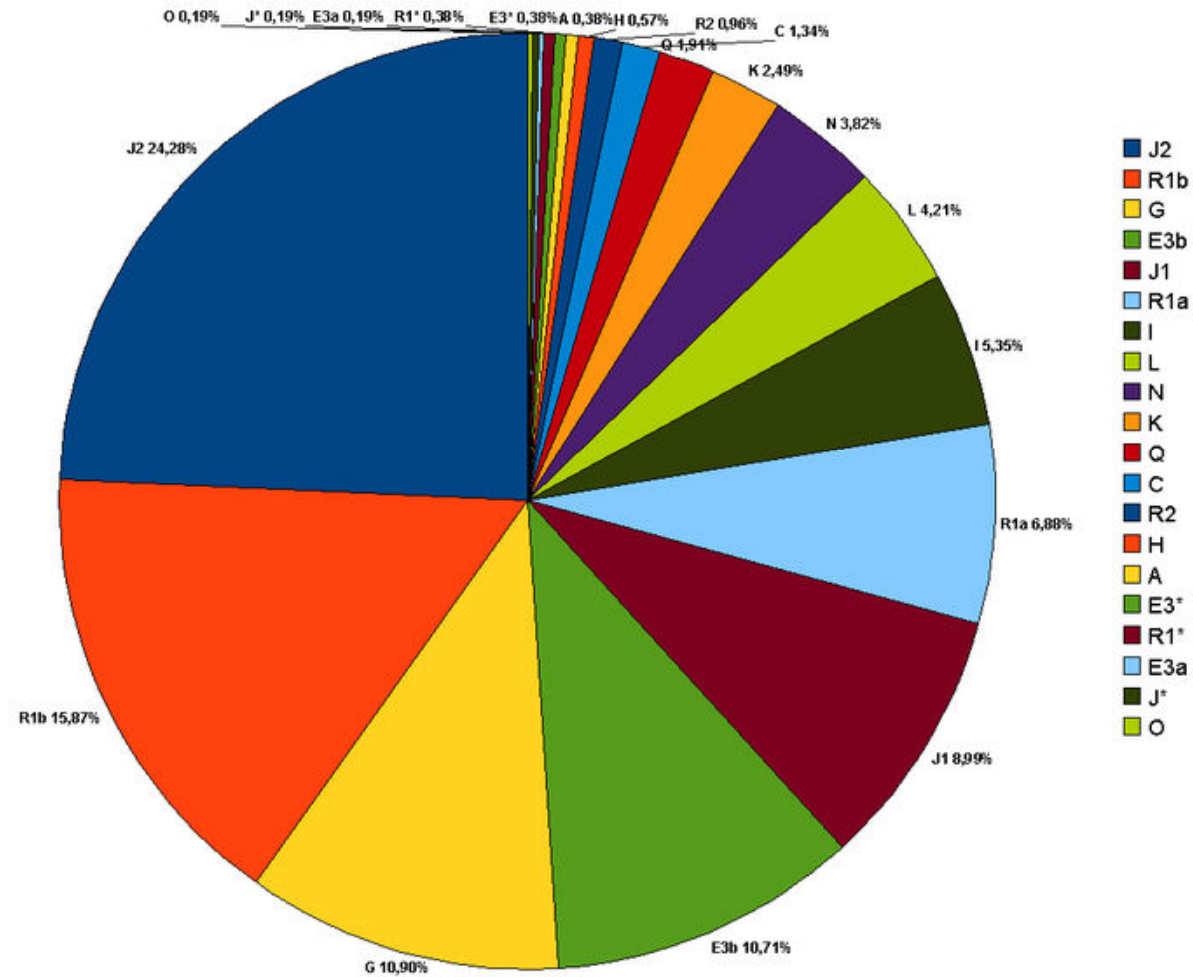
Cavalli-Sforza, 2001

- Allele frequencies for different localities are subjected to a smoothing procedure.
- Principle Components are found and projected on geographical maps.
- Strongly criticized (Sokal et al.): even no geographical structure will “look like” geographical structure, no timing of gradients,...

1. Agriculture 6-10 Kyr
2. Greek Colonisation 3 Kyr
3. Retraction of the Basques
4. Uralic People
5. Horse domestication



# Genetic diversity in Turkey



Y chromosome Haplogroup distribution in Turkey (Cinnioglu et al., Hum. Genet. 2004)

# Where do Turkish genes come from?

(Y chromosome haplogroup distribution)

J1=9% - Typical amongst people from the Arabian Peninsula.

J2=24% - Typical amongst Near Eastern peoples.

R1a=6.9% - Typical of Eastern Europeans

I=5,3% - Typical of Central Europeans

R1b=14.7% - Typical of Western Europeans

G=10.9% - Typical of people from the Caucasus

E1b1b=11.3% - Typical amongst populations of Near East.

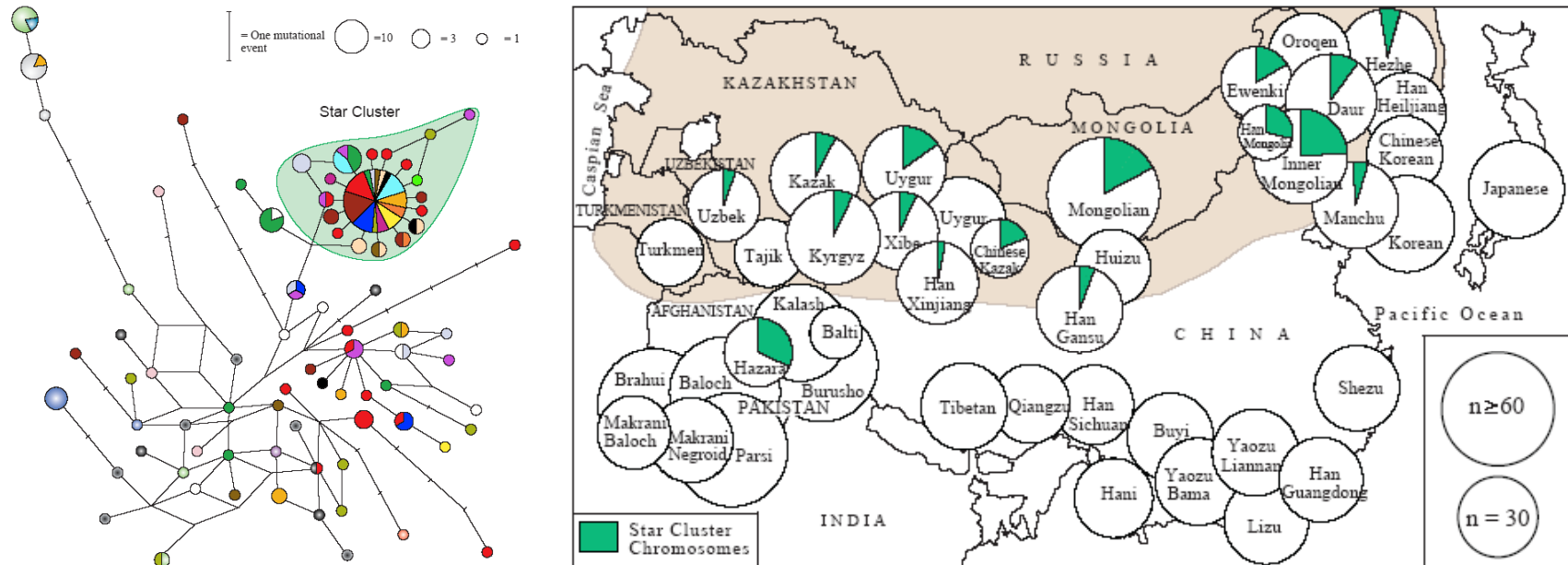
N=3.8% - Typical of Siberian populations

T=2.5% - Distribution from south Asia to Europe

L=4.2% - Typical of Indian Subcontinent populations.

*Inference: Large diversity , consistent with extensive migrations and admixture*

## More recent reshaping of diversity



- ‘Star cluster’ Y haplotype originated in/near Mongolia ~1,000 (700-1,300) years ago
- Now carried by ~8% of men in Central/East Asia, ~0.5% of men worldwide
- Suggested association with Genghis Khan

# Is evolution in human beings going on today?

*Short answer: Yes*

Parasites and disease

Communication

Tool use

Social behaviour

## The Prion protein gene and human disease

- Prion protein gene *PRNP* linked to 'protein-only' diseases e.g. CJD, kuru
- A common polymorphism, M129V, influences the course of these diseases: the MV heterozygous genotype is protective
- Kuru acquired from ritual cannibalism was reported (1950s) in the Fore people of Papua New Guinea, where it caused up to 1% annual mortality
- Departure from Hardy-Weinberg equilibrium for the M129V polymorphism is seen in Fore women over 50 (23/30 heterozygotes,  $P = 0.01$ )

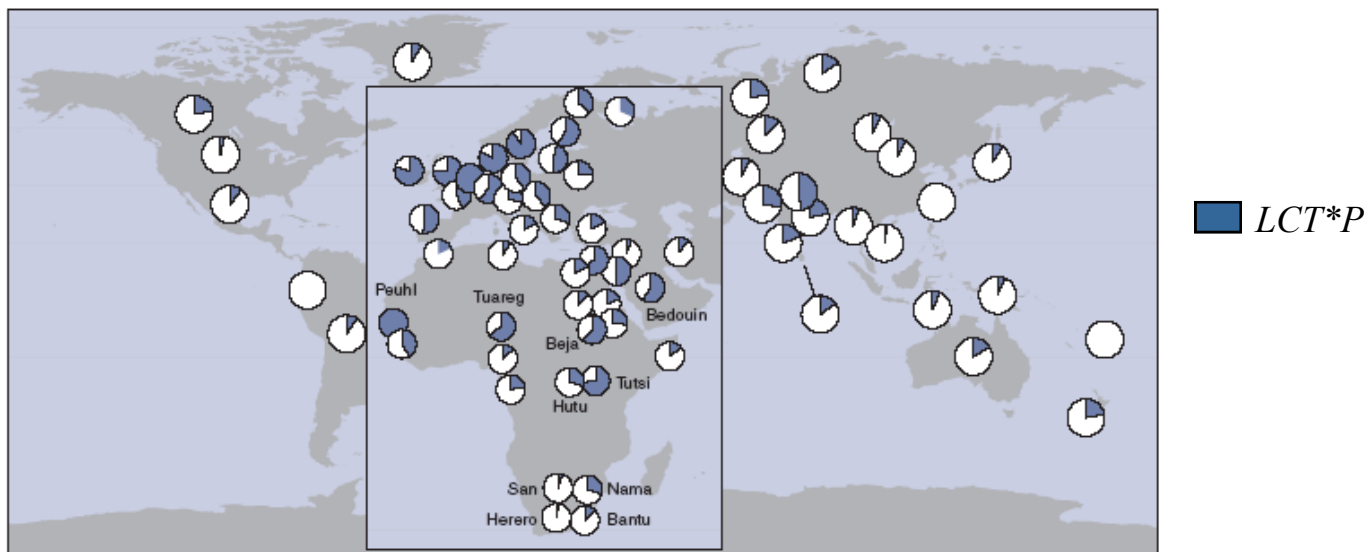
## Genome-wide search for protein sequence evolution

- 7645 human-chimp-mouse gene trios compared
- Most significant categories showing positive selection include:
  - Olfaction: sense of smell
  - Amino-acid metabolism: diet
  - Development: e.g. skeletal
  - Hearing: for speech perception

## Lactase persistence

All infants have high lactase enzyme activity to digest the sugar lactose in milk

In most humans, activity declines after weaning, but in some it persists:





## Gene-culture co-evolution?

*Are there 'typical' human cultural traits?*

Tool use (manipulation of the environment)

Learning by imitation

Music

Cooking

Ritualised behaviour

Life-cycle traits

Religion

Symbolic communication (language)

# Language Trees

Cavalli-Sforza (1997) Genes Peoples and Languages PNAS 94.7719-24

## Principle of Comparison.

Loss of cognates ("homologous" words)

Syntax Comparison.

Sound use.

Reconstruction (dependent on interpretation) – stretches back 2-6.000 years dependent on criteria.

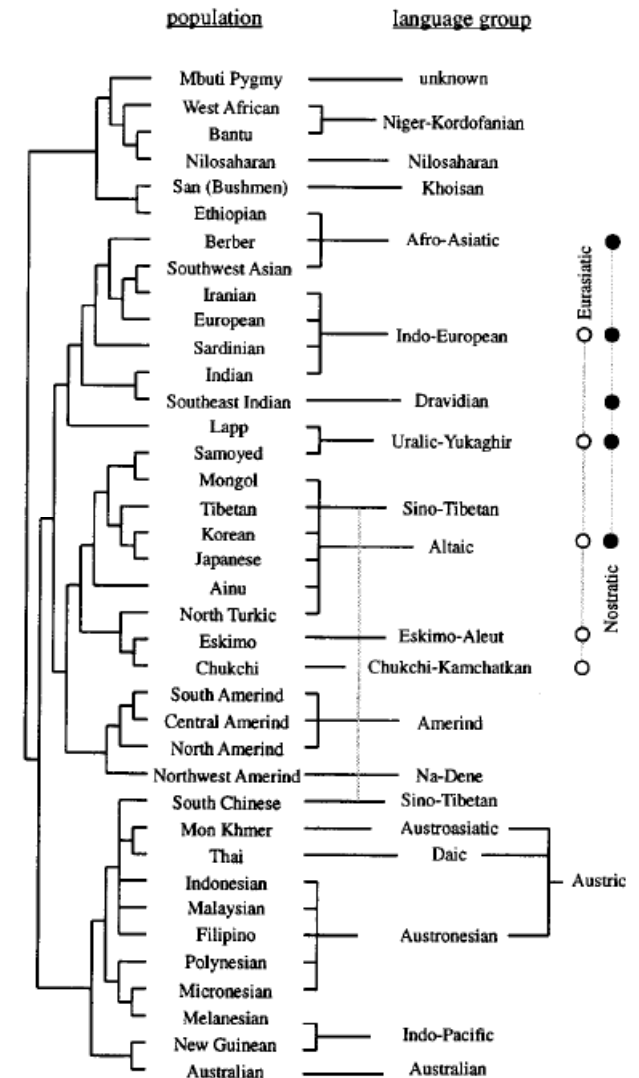


FIG. 3. Coherence between a genetic tree derived from 42 populations with 120 classical polymorphisms (*Left*) and what is known: the linguistic tree (*Right*), including two recently reconstructed suprafamilies (shown at the extreme right). (From ref. 23.)

To what extent is human culture rooted in  
our pre-human ancestry  
and to what extent is it an  
'emergent' consequence of  
the human brain?