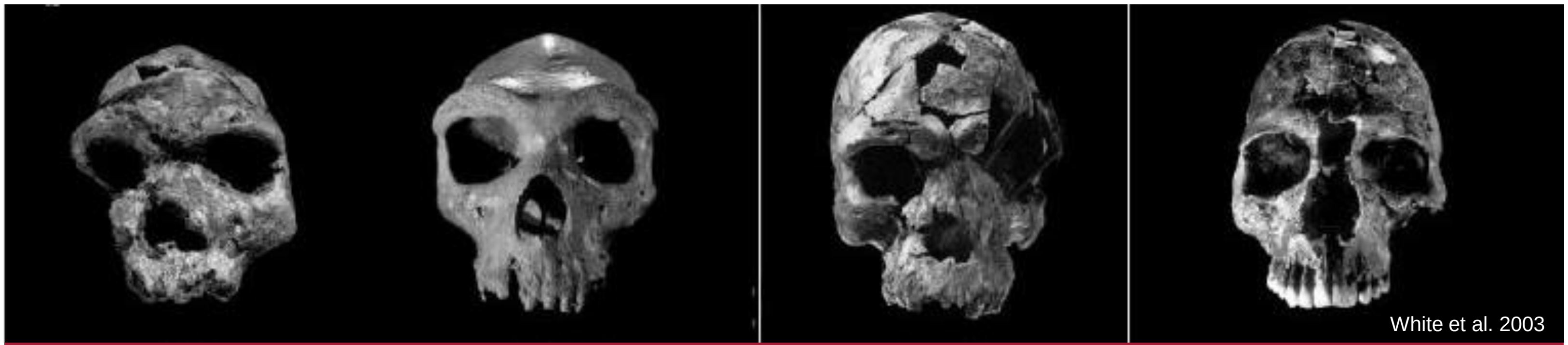


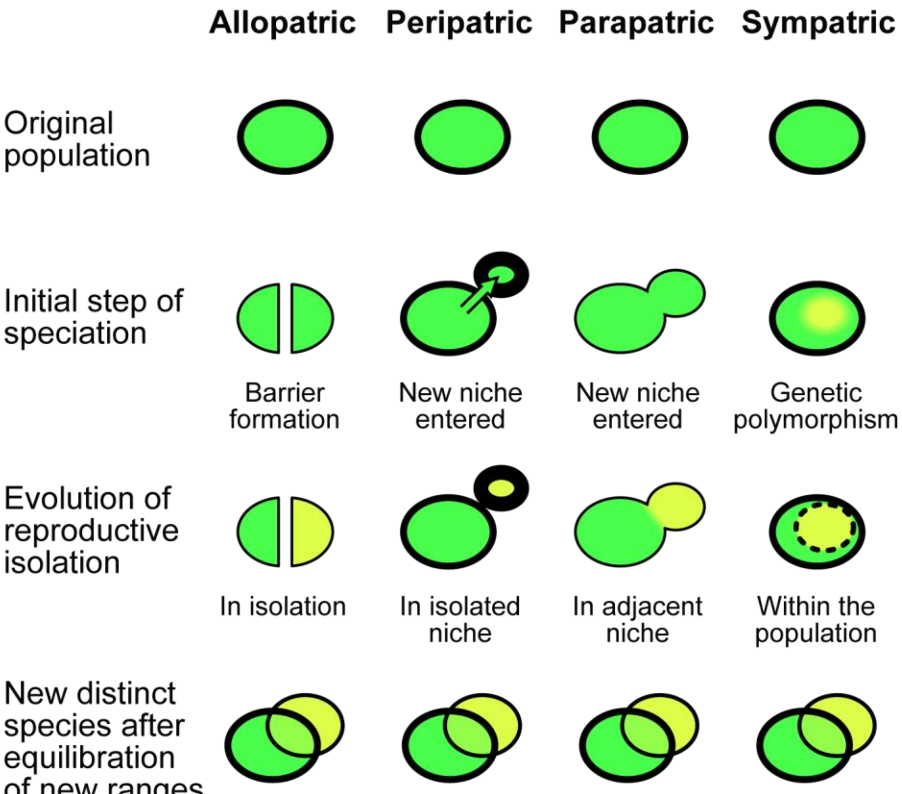
Modern Human Origins

Hugo Reyes-Centeno, Yonatan Sahle, Christian Bentz

10 December 2018, Lecture 7

Last lecture:

- **Species concepts and definitions**
  How do we define a species and how does this relate to modern humans?
- **Genomics of modern human origins**
  What does (ancient) DNA tell us about the origins and evolution of modern humans?
- **Models of anthropogeny**
  What model of modern human origins is best supported with the current fossil and genomic evidence?
- **Modern human dispersals**
  When, how, and why did anatomically modern humans disperse out of Africa?



I. Karonen 2006

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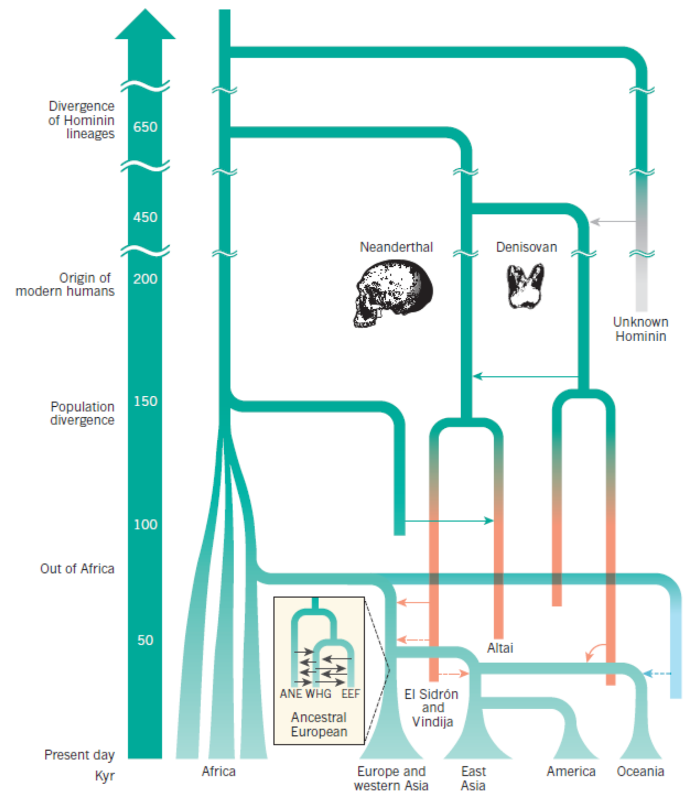

Nielsen et al 2017

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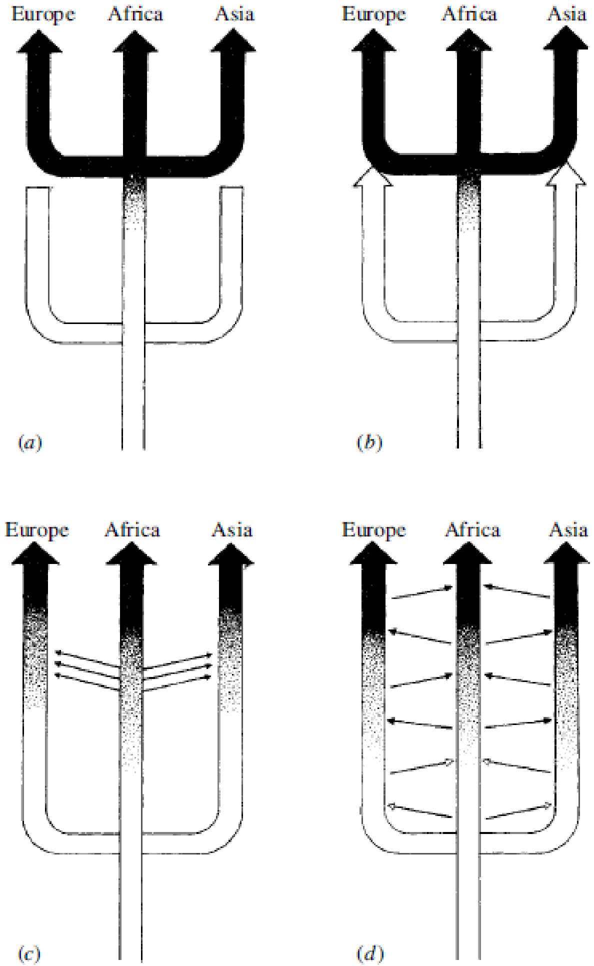

Stringer 2001

EBERHARD KARLS UNIVERSITÄT TÜBINGEN

DFG

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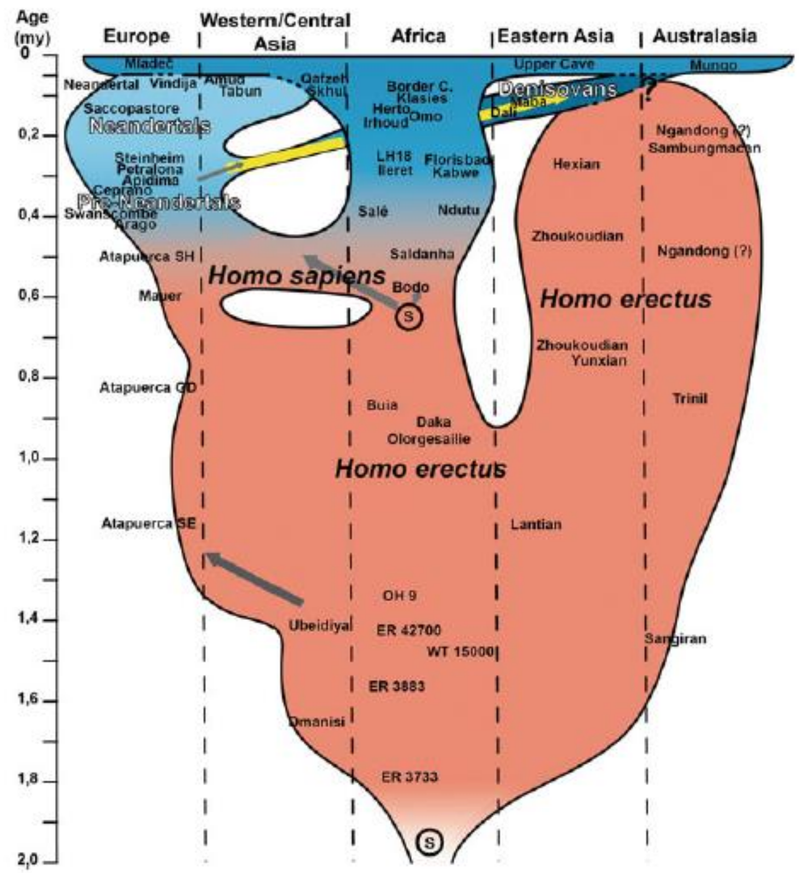

Fig. 7 Origin and evolution of *Homo sapiens*

Bräuer 2015

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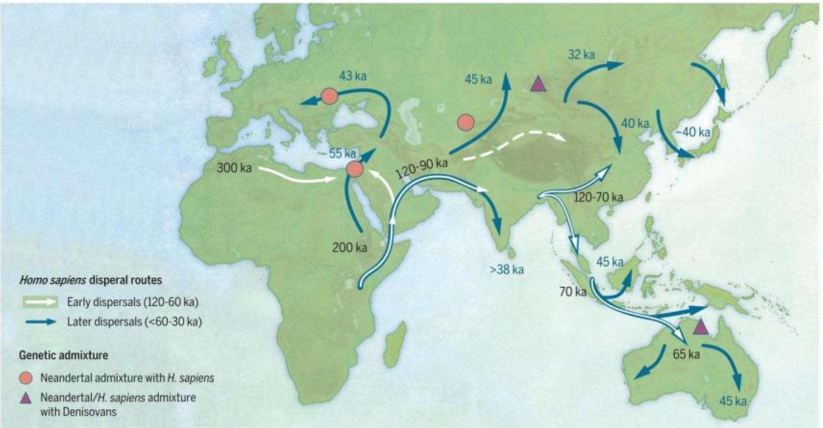

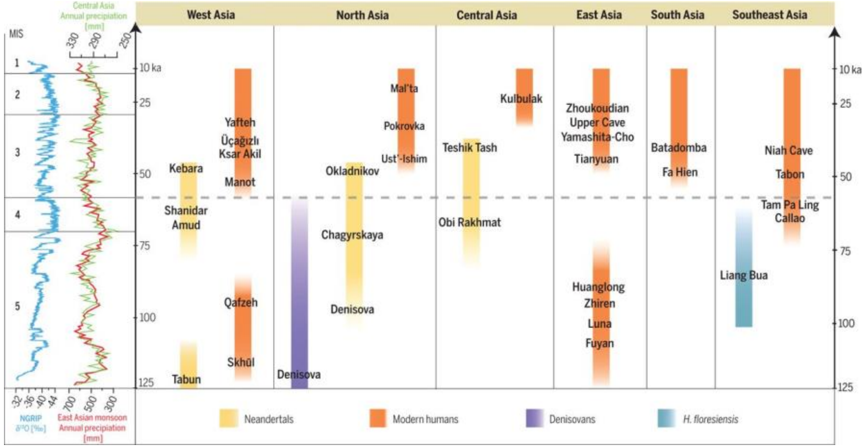

Bae et al 2017

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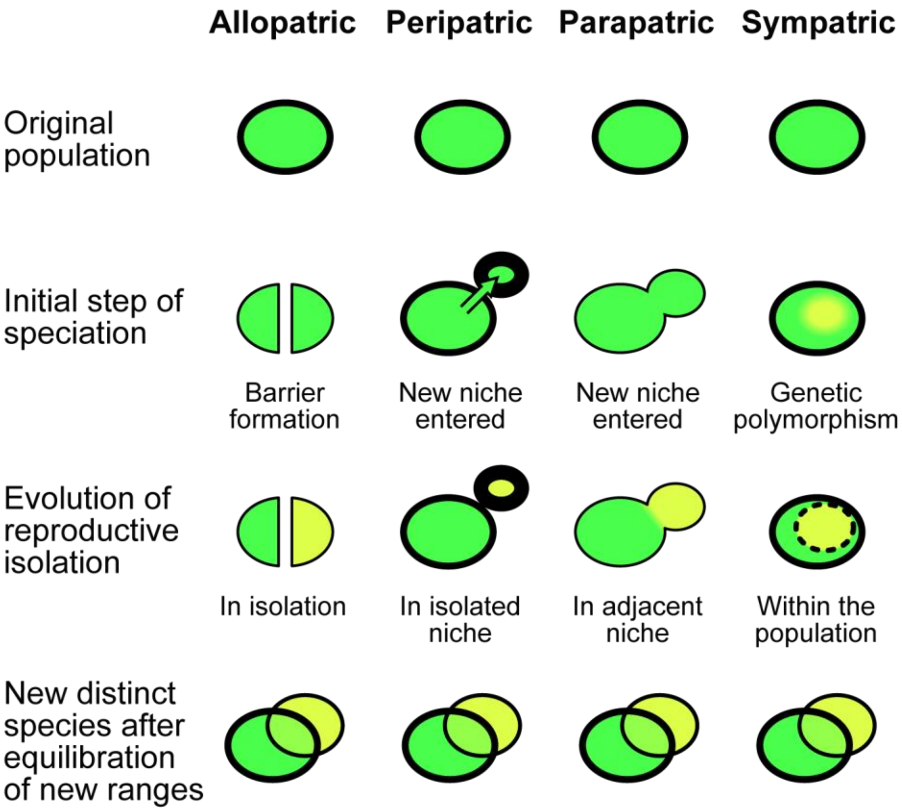
- **The serial founder effect**
  What are the genetic signatures of the human expansion out of Africa?



I. Karonen 2006

This lecture:

- **The serial founder effect**
  What are the genetic signatures of the human expansion out of Africa?

- **Neutral and adaptive evolution**
  What is the difference between evolution by chance and evolution under selection?

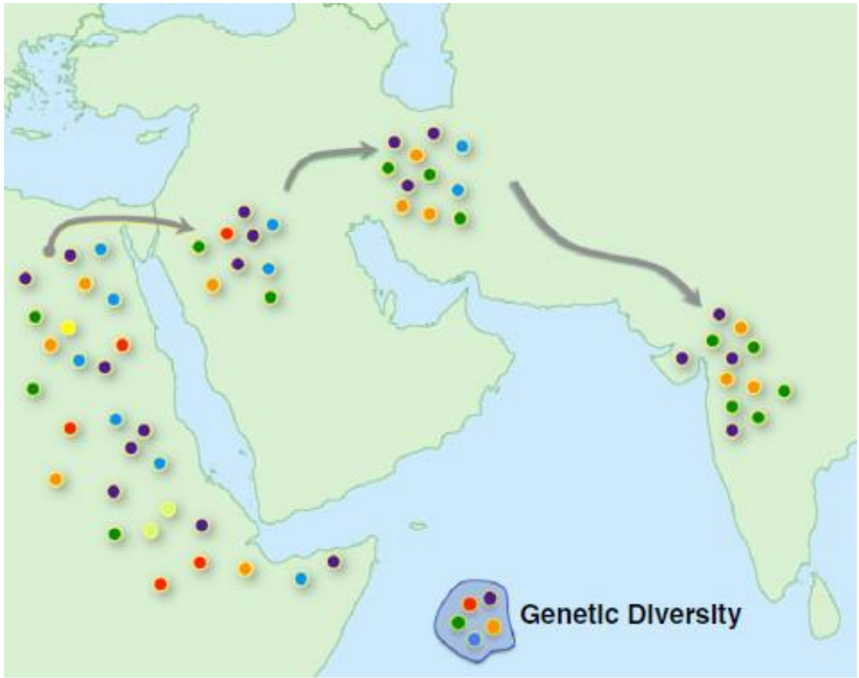
- **Co-evolution**
  Do the genotype and phenotype co-evolve?

Neutral & adaptive evolution

What is the difference between evolution by chance and evolution under selection?

The serial founder effect

- **Serial founding effect (or cascading bottlenecks)**
  1. Decreasing intra-population diversity with geographical distance from Africa

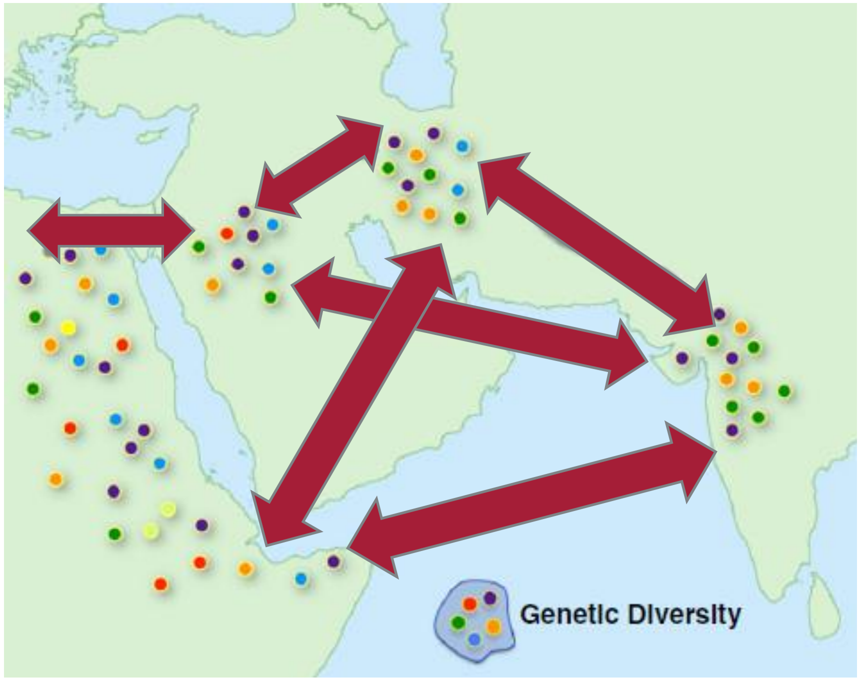


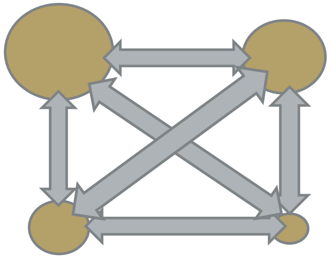
Henn et al. 2012

The serial founder effect

- **Serial founding effect (or cascading bottlenecks)**

  1. Decreasing intra-population diversity with geographical distance from Africa
  2. Increasing inter-population diversity between populations with geographical distance separating them





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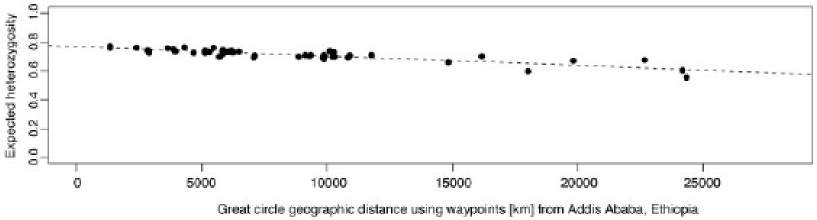

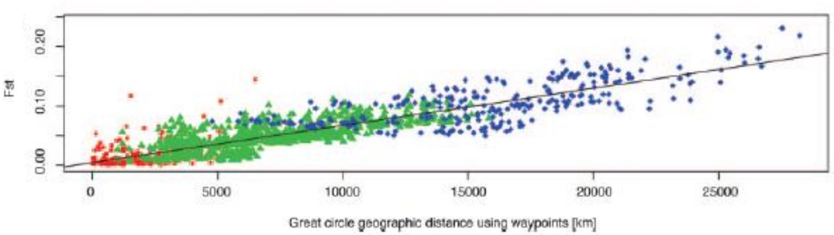

Ramachandran et al. 2005

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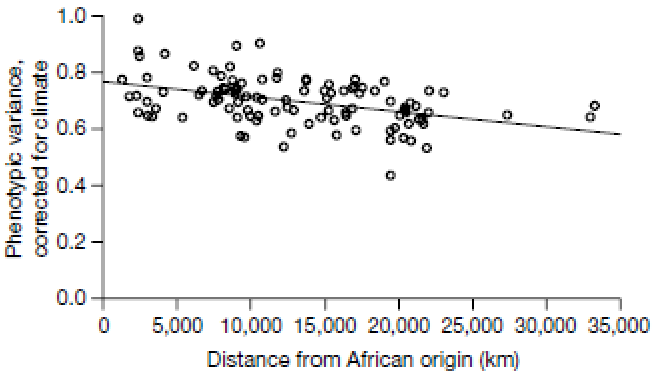

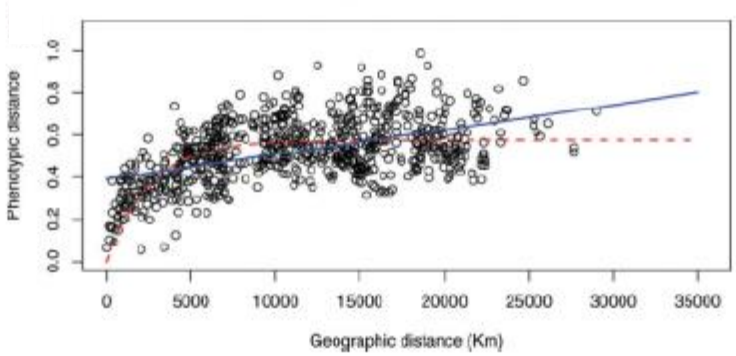

Manica et al. 2007; Betti et al. 2011

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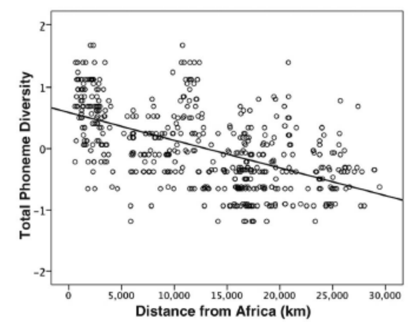

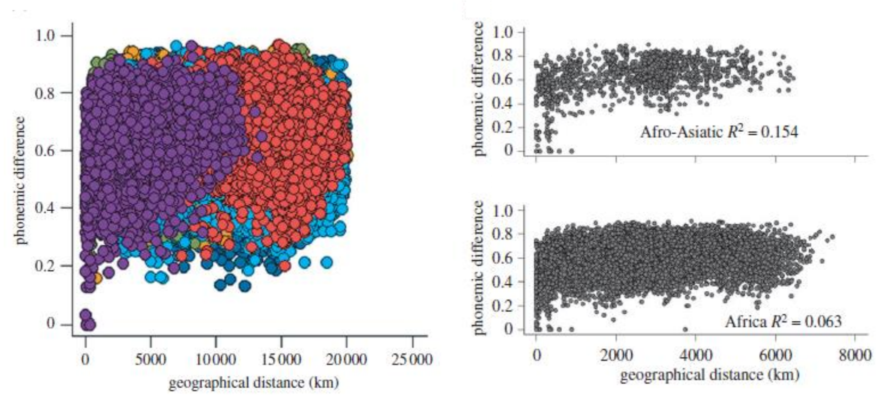

Atkinson 2011; Hunley et al. 2012

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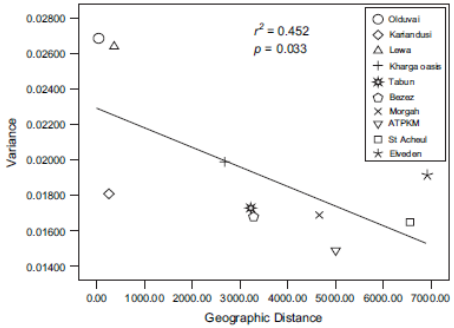

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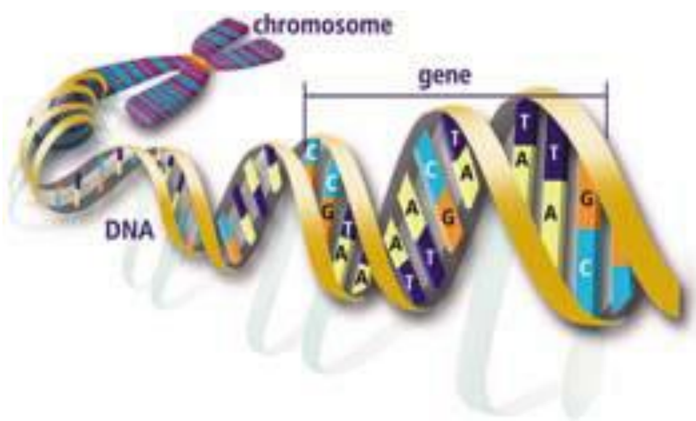
Lycett & von Cramon-Taubadel 2008

Modes of evolution

- **Neutral evolution (or drift)** refers to changes in the genotype and phenotype that refer to chance events or stochastic processes (e.g. random mutations)

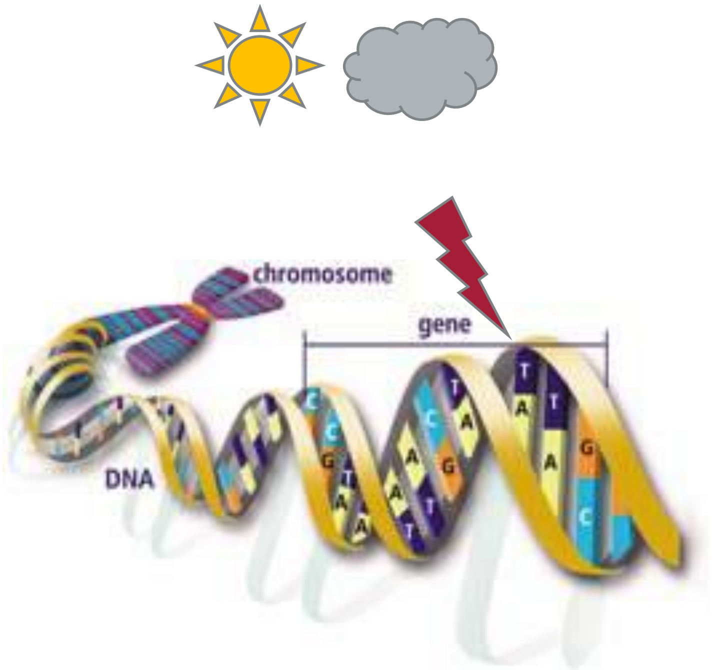


US Department of Energy

Modes of evolution

- Neutral evolution (or drift) refers to changes in the genotype and phenotype that refer to chance events or stochastic processes (e.g. random mutations)

- **Adaptive evolution (or selection)** refers to changes in the genotype and phenotype that are due to non-random or non-stochastic processes (e.g. environmental)



US Department of Energy

Modes of evolution

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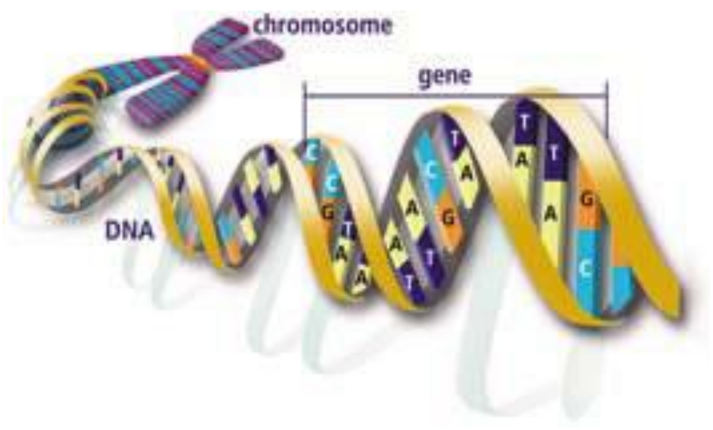
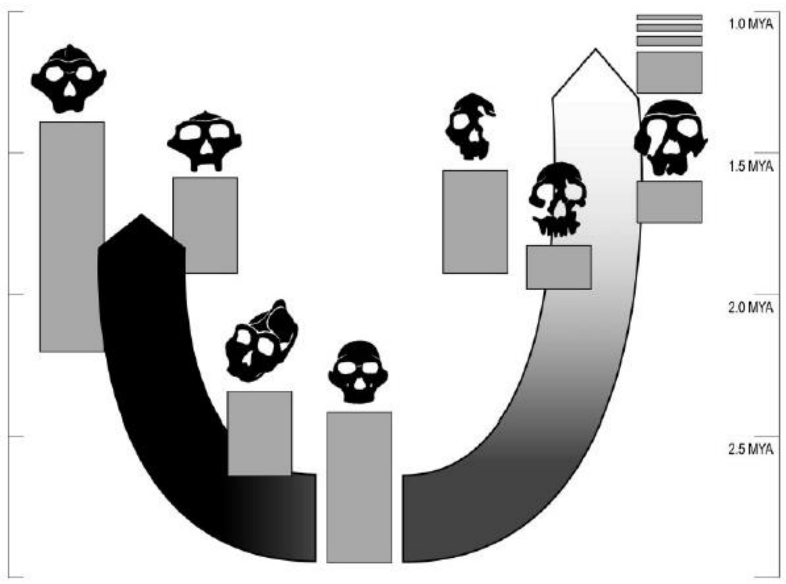
Note: changes in genotype and phenotype can occur during an organisms lifetime, but they will not be inherited to the next generation unless they occur along the germ line or an epigenetic process.



US Department of Energy

Drift and selection in human evolution

- Adaptive evolution is thought to have played an important role in early human evolution



Rogers Ackermann & Cheverud 2004

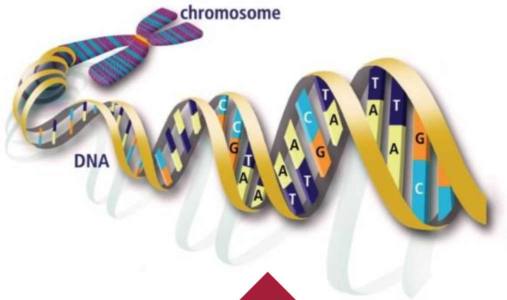
Drift and selection in human evolution

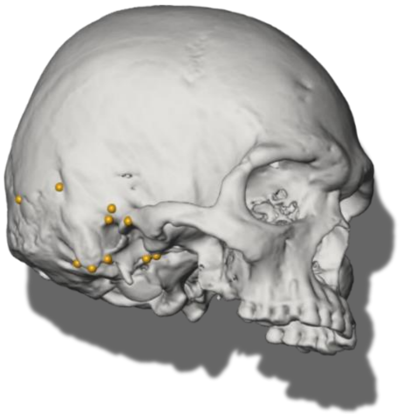
- Adaptive evolution is thought to have played an important role in early human evolution
- Netural evolution is thought to play the primary role in recent human evolution

Weaver et al 2007

Drift and selection in human evolution

- Adaptive evolution is thought to have played an important role in early human evolution
- Netural evolution is thought to play the primary role in recent human evolution
    - Direct comparison of netural genomic and phenotypic variation between populations





US Department of Energy; Reyes-Centeno et al 2017

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Q_{ST}

	AU	CA	EA	JP	ME	NE	NG	NI	SA	SI
AU	0									
CA	0.372	0								
EA	0.558	0.524	0							
JP	0.323	0.100	0.572	0						
ME	0.365	0.572	0.637	0.554	0					
NE	0.674	0.516	1.299	0.476	0.737	0				
NG	0.142	0.359	0.724	0.389	0.304	0.625	0			
NI	0.355	0.167	0.379	0.200	0.516	0.585	0.371	0		
SA	0.356	0.449	0.334	0.450	0.497	0.724	0.419	0.329	0	
SI	0.400	0.221	0.495	0.294	0.402	0.582	0.433	0.088	0.393	0

F_{ST}

	AU	CA	EA	JP	ME	NE	NG	NI	SA	SI
AU	0									
CA	0.110	0								
EA	0.191	0.129	0							
JP	0.144	0.031	0.175	0						
ME	0.079	0.145	0.226	0.173	0					
NE	0.119	0.053	0.158	0.062	0.159	0				
NG	0.092	0.111	0.200	0.136	0.107	0.115	0			
NI	0.105	0.022	0.121	0.071	0.139	0.067	0.114	0		
SA	0.191	0.135	0.006	0.175	0.219	0.160	0.199	0.127	0	
SI	0.104	0.025	0.126	0.070	0.135	0.067	0.112	0.003	0.131	0

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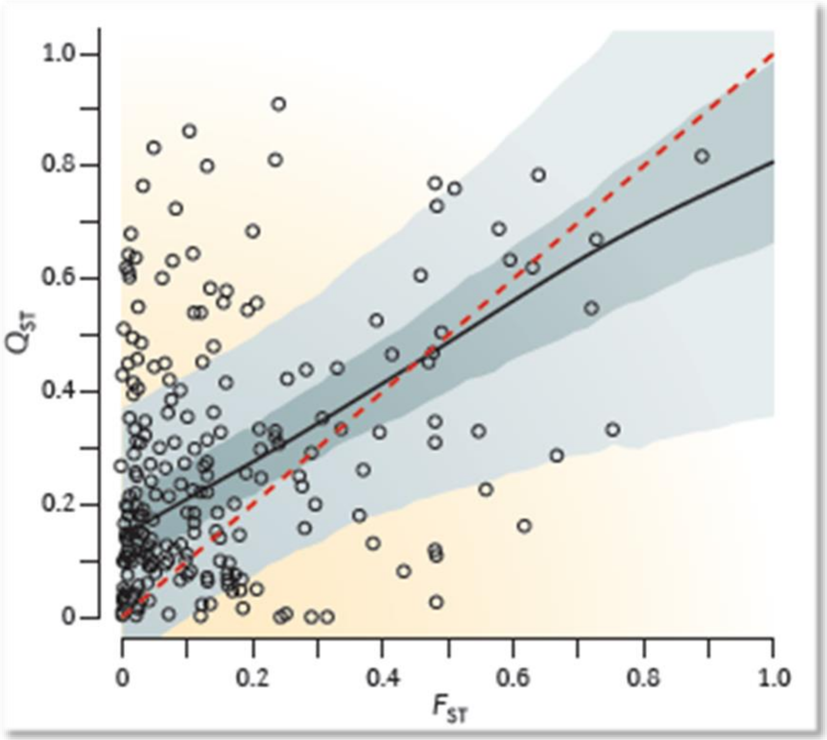
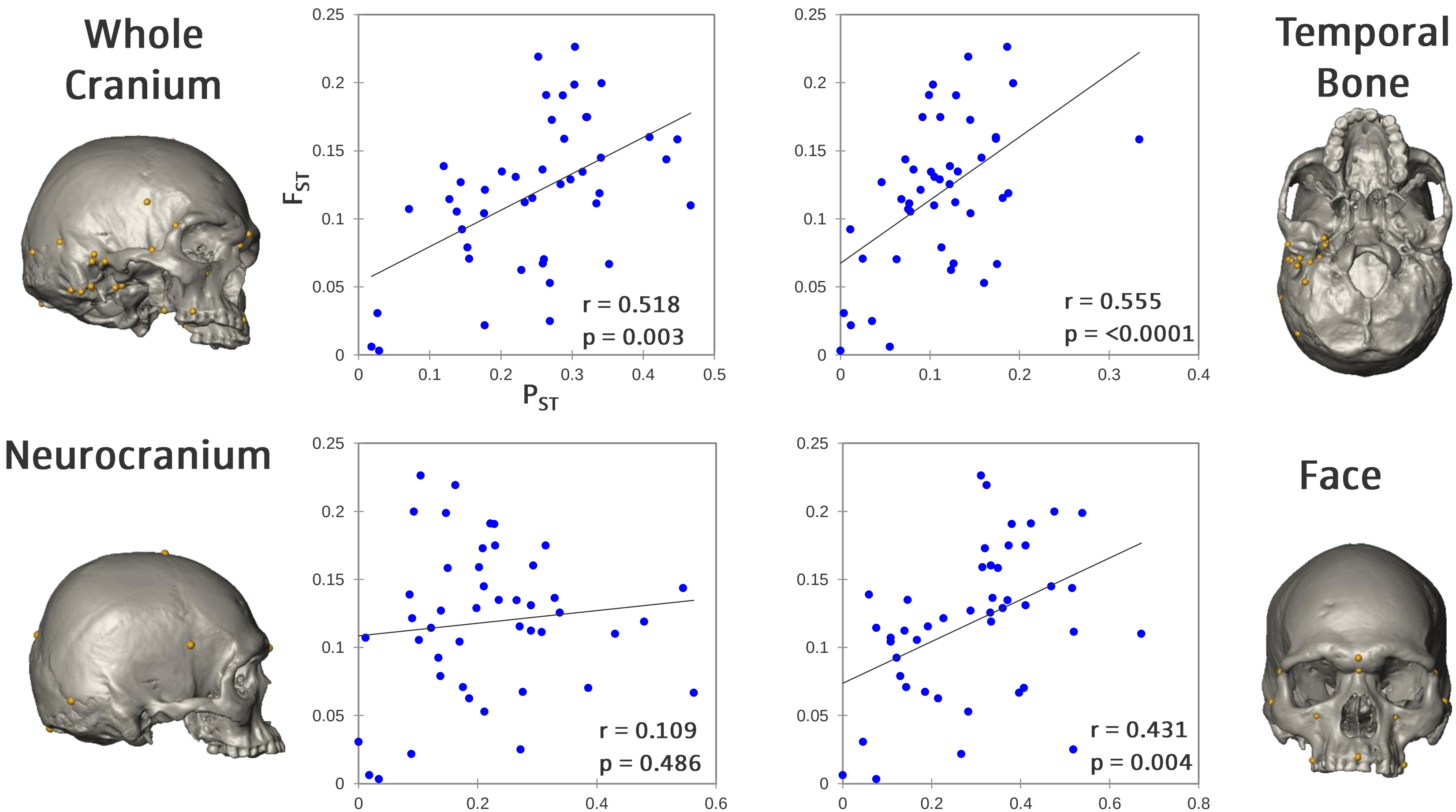


Leinonen et al 2013

Drift and selection in human evolution



Reyes-Centeno et al 2017

Drift and selection in human evolution

- ❑ Adaptive evolution is thought to have played an important role in early human evolution
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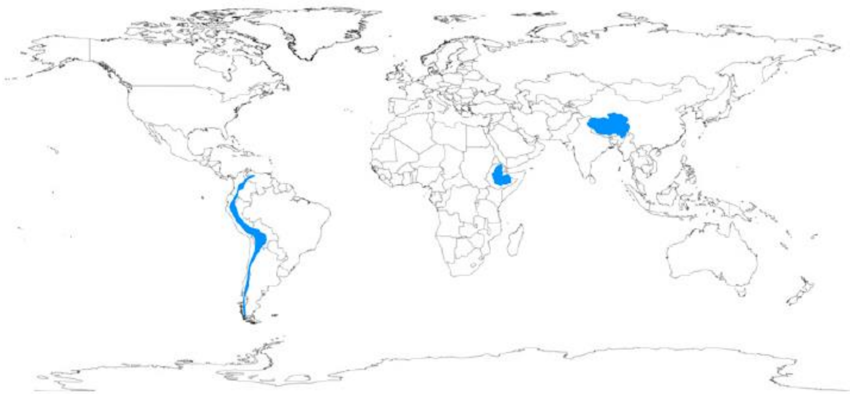
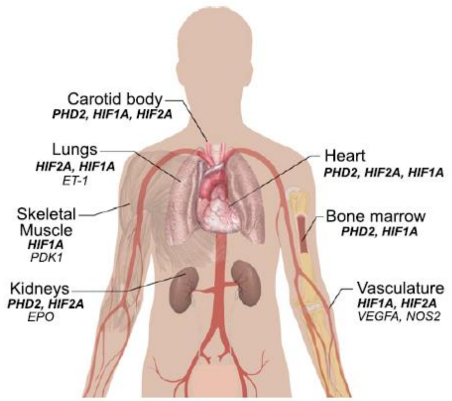
- ❑ Adaptation still plays an important role
  - e.g. adaptation to high altitude environments: independent (convergent) evolution and possible hominin introgression in Tibet

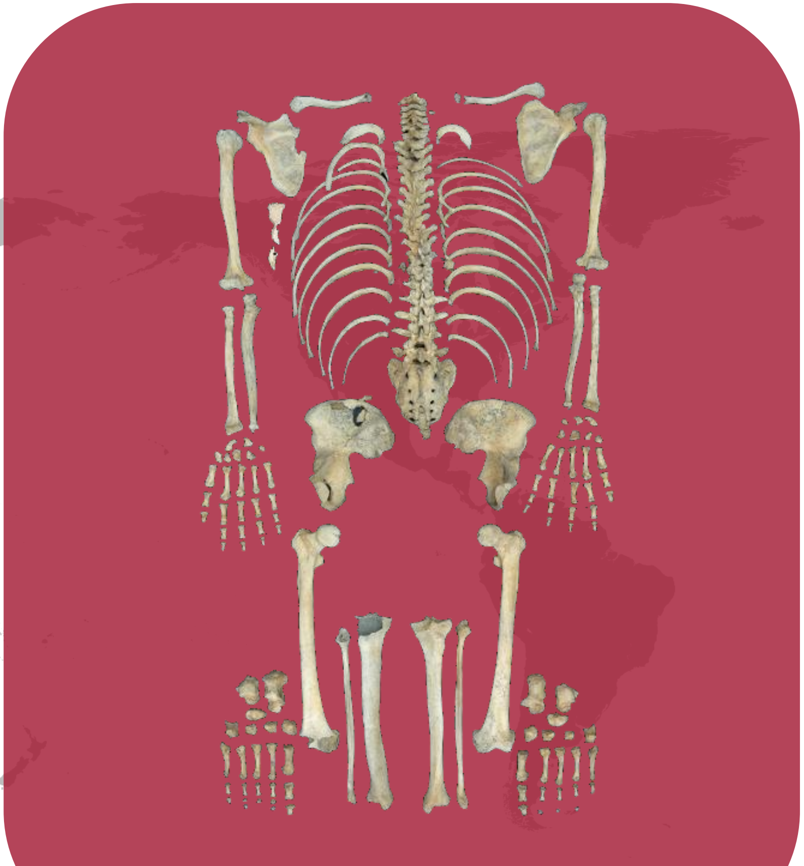




Bingham & Lee 2014

Drift and selection in human evolution

Cuncaicha, Peru 15-07 ~9 ka



Cuncaicha, Peru 15-03 ~4 ka

Francken et al. 2018

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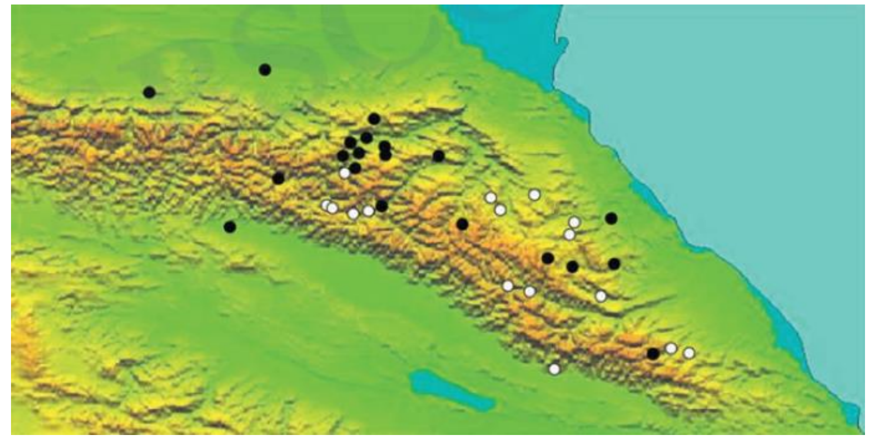

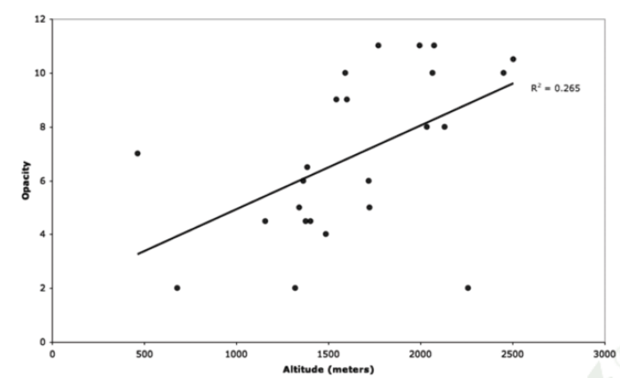

Fig. 2. Altitude (*x*-axis) by opacity (*y*-axis). Number of languages: $N = 26$. Shared variance: $R^2 = 0.27$

Nichols 2013

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- Adaptation still plays an important role
  - e.g. adaptation to high altitude environments: independent (convergent) evolution and possible hominin introgression in Tibet
  - Multiple candidate genes that are associated with phenotypes

Genes	Function or phenotype
LCT, MAN2A1, SI, SLC27A4, PPARD, SLC25A20, NCOA1, LEPR, LEPR, ADAMTS19, ADAMTS20, APEH, PLAU, HDAC8, UBR1, USP26, SCP2, NKX2-2, AMY1, ADH, NPY1R, NPY5R	Digestion of milk and dairy products; metabolism of carbohydrates, starch, proteins, lipids and phosphates; alcohol metabolism
Cytochrome P450 genes (*CYP3A5, CYP2E1, CYP1A2* and *CYP2D6*)	Detoxification of plant secondary compounds
CD58, APOBEC3F, CD72, FCRL2, TSLP, RAG1, RAG2, CD226, IGJ, TJP1, VPS37C, CSF2, CCNT2, DEFB118, STAB1, SP1, ZAP70, BIRC6, CUGBP1, DLG3, HMGCR, STS, XRN2, ATRN, G6PD, TNFSF5, HbC, HbE, HbS, Duffy, α-globin	Immunity, pathogen response; resistance to malaria and other crowd diseases
LEPR, PON1, RAPTOR, MAPK14, CD36, DSCR1, FABP2, SOD1, CETP, EGFR, NPPA, EPHX2, MAPK1, UCP3, LPA, MMRN1	Energy metabolism, hot or cold tolerance; heat-shock genes
SLC24A5, SLC25A2, EDAR, EDA2R, SLC24A4, KITLG, TYR, 6p25.3, OCA2, MC1R, MYO5A, DTNBP1, TYRP1, RAB27A, MATP, MC2R, ATRN, TRPM1, SILV, KRTAPs, DCT	The externally visible phenotype (skin pigmentation, hair thickness, eye and hair colour, and freckles)
CDK5RAP2, CENPJ, GABRA4, PSEN1, SYT1, SLC6A4, SNTG1, GRM3, GRM1, GLRA2, OR4C13, OR2B6, RAPSN, ASPM, RNT1, SV2B, SKP1A, DAB1, APPBP2, APBA2, PCDH15, PHACTR1, ALG10, PREP, GPM6A, DGKI, ASPM, MCPH1, FOXP2	Nervous system, brain function and development; language skills and vocal learning
BMP3, BMPR2, BMP5, GDF5	Skeletal development
MYH16, ENAM	Jaw muscle fibres; tooth-enamel thickness

Laland et al 2010

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Cytochrome P450 genes (*CYP3A5, CYP2E1, CYP1A2* and *CYP2D6*)	Detoxification of plant secondary compounds
CD58, APOBEC3F, CD72, FCRL2, TSLP, RAG1, RAG2, CD226, IGJ, TJP1, VPS37C, CSF2, CCNT2, DEFB118, STAB1, SP1, ZAP70, BIRC6, CUGBP1, DLG3, HMGCR, STS, XRN2, ATRN, G6PD, TNFSF5, HbC, HbE, HbS, Duffy, α-globin	Immunity, pathogen response; resistance to malaria and other crowd diseases
LEPR, PON1, RAPTOR, MAPK14, CD36, DSCR1, FABP2, SOD1, CETP, EGFR, NPPA, EPHX2, MAPK1, UCP3, LPA, MMRN1	Energy metabolism, hot or cold tolerance; heat-shock genes
SLC24A5, SLC25A2, EDAR, EDA2R, SLC24A4, KITLG, TYR, 6p25.3, OCA2, MC1R, MYO5A, DTNBP1, TYRP1, RAB27A, MATP, MC2R, ATRN, TRPM1, SILV, KRTAPs, DCT	The externally visible phenotype (skin pigmentation, hair thickness, eye and hair colour, and freckles)
CDK5RAP2, CENPJ, GABRA4, PSEN1, SYT1, SLC6A4, SNTG1, GRM3, GRM1, GLRA2, OR4C13, OR2B6, RAPSN, ASPM, RNT1, SV2B, SKP1A, DAB1, APPBP2, APBA2, PCDH15, PHACTR1, ALG10, PREP, GPM6A, DGKI, ASPM, MCPH1, FOXP2	Nervous system, brain function and development; language skills and vocal learning
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Laland et al 2010

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LEPR, PON1, RAPTOR, MAPK14, CD36, DSCR1, FABP2, SOD1, CETP, EGFR, NPPA, EPHX2, MAPK1, UCP3, LPA, MMRN1	Energy metabolism, hot or cold tolerance; heat-shock genes
SLC24A5, SLC25A2, EDAR, EDA2R, SLC24A4, KITLG, TYR, 6p25.3, OCA2, MC1R, MYO5A, DTNBP1, TYRP1, RAB27A, MATP, MC2R, ATRN, TRPM1, SILV, KRTAPs, DCT	The externally visible phenotype (skin pigmentation, hair thickness, eye and hair colour, and freckles)
CDK5RAP2, CENPJ, GABRA4, PSEN1, SYT1, SLC6A4, SNTG1, GRM3, GRM1, GLRA2, OR4C13, OR2B6, RAPSN, ASPM, RNT1, SV2B, SKP1A, DAB1, APPBP2, APBA2, PCDH15, PHACTR1, ALG10, PREP, GPM6A, DGKI, ASPM, MCPH1, $\boxed{\textit{FOXP2}}$	Nervous system, brain function and development; language skills and vocal learning
BMP3, BMPR2, BMP5, GDF5	Skeletal development
MYH16, ENAM	Jaw muscle fibres; tooth-enamel thickness

Laland et al 2010

Co-evolution

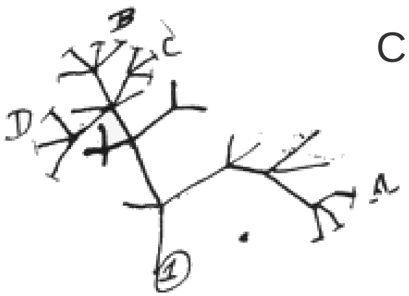
Do the genotype and phenotype co-evolve?

Co-evolution of genes and languages

- Darwin‘s hypothesized the co-evolution of languages and genes

“If we possessed a perfect pedigree of mankind, a genealogical arrangement of the races of man would afford the best classification of the various languages now spoken throughout the world…. ”

Charles Darwin, 1859



Co-evolution of genes and languages

- Darwin‘s hypothesized the co-evolution of languages and genes
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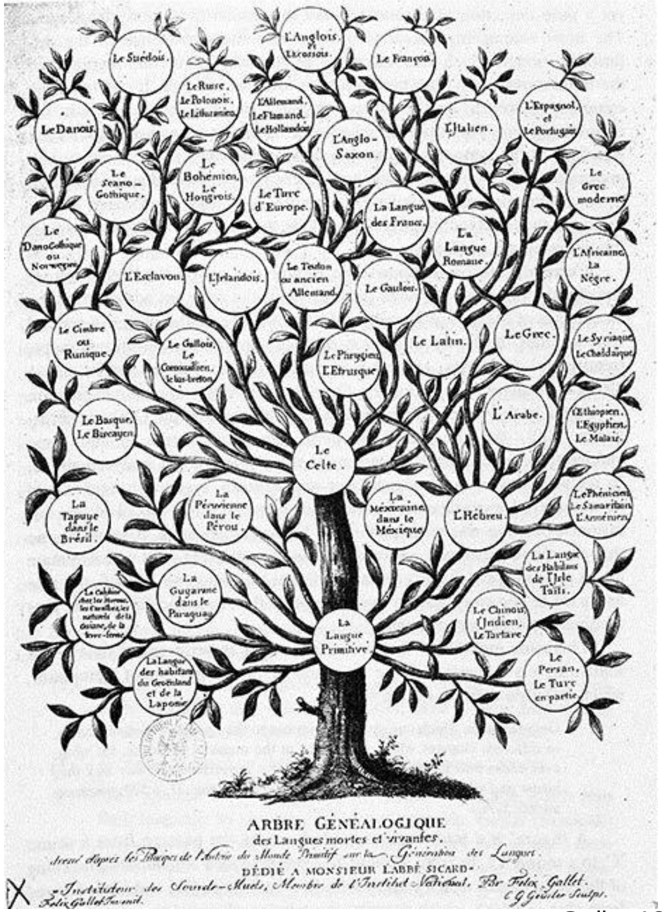

Gallet 1800

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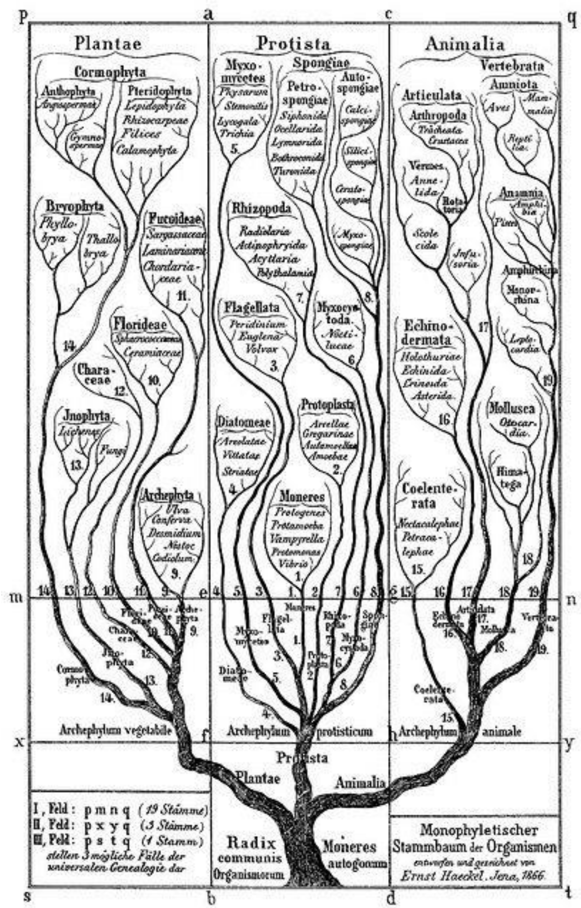

Haeckel 1866

Co-evolution of genes and languages

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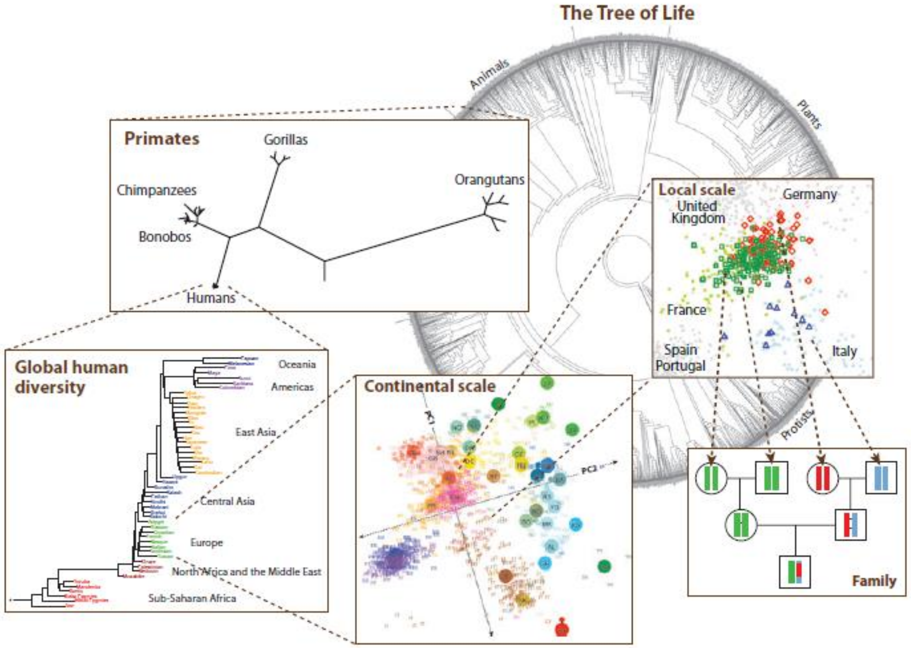
SNPs DNA



Novembre & Ramachandran 2011

Co-evolution of genes and languages

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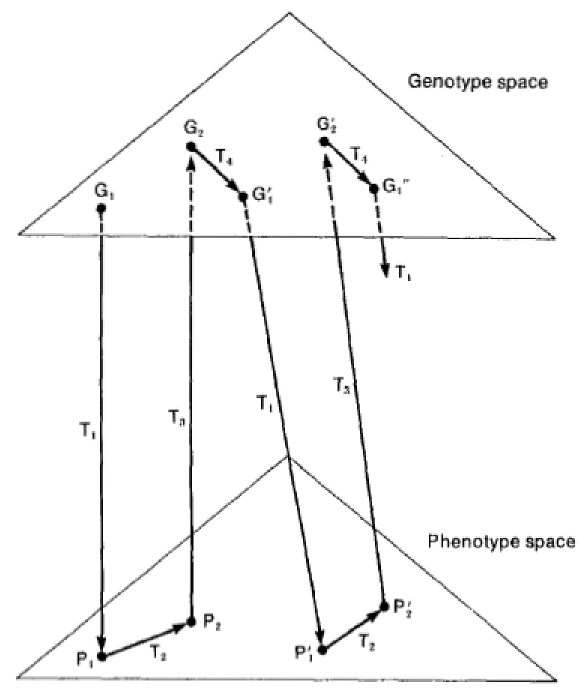

Lewontin et al 2010

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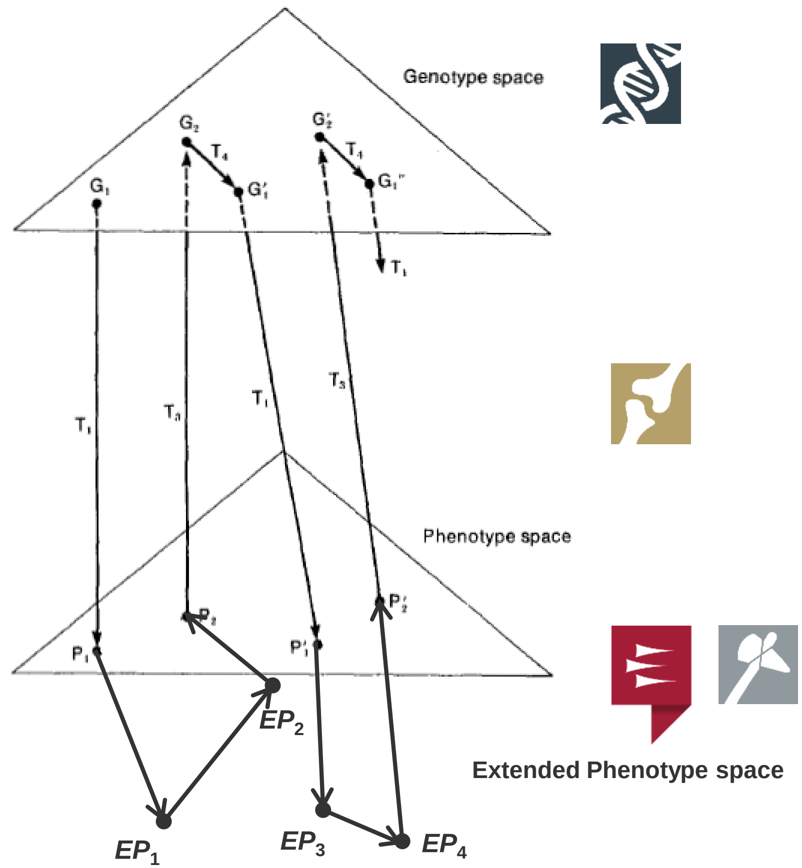

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- Theoretical framework: one can trace how the genotype and phenotype affect each other
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  - Language and behavior can be considered as an extended phenotype
  - Language and behavior tend to change much faster than the genotype

“Organic evolution is not the only sort of evolution in the sense of a process of cumulative change. When a level of intelligence making symbolic speech possible was reached in the anthropoid line, a new evolutionary process emerged, enormously more rapid than organic evolution.”

Sewall Wright, 1950

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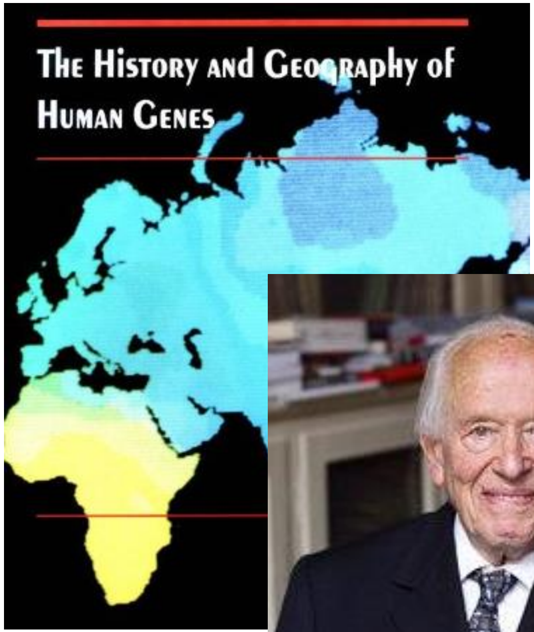

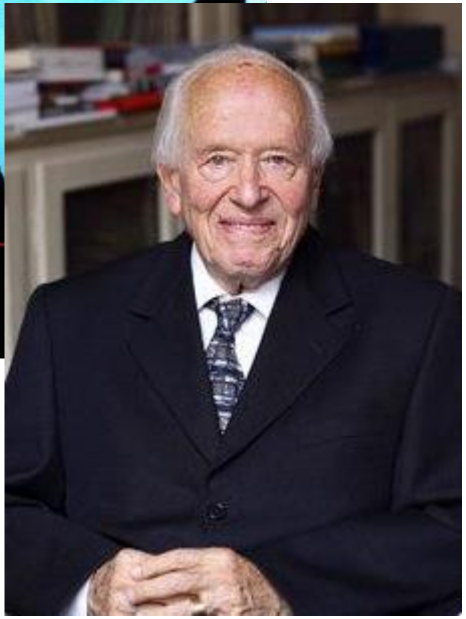

Cavalli-Sforza 1994

Co-evolution of genes and languages

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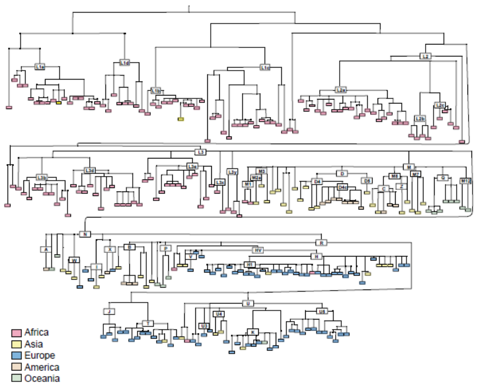
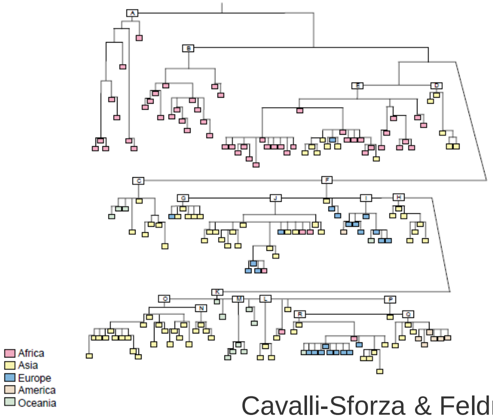
    - Differences with different datasets

mtDNA



Y-chromosome DNA



Cavalli-Sforza & Feldman 2003

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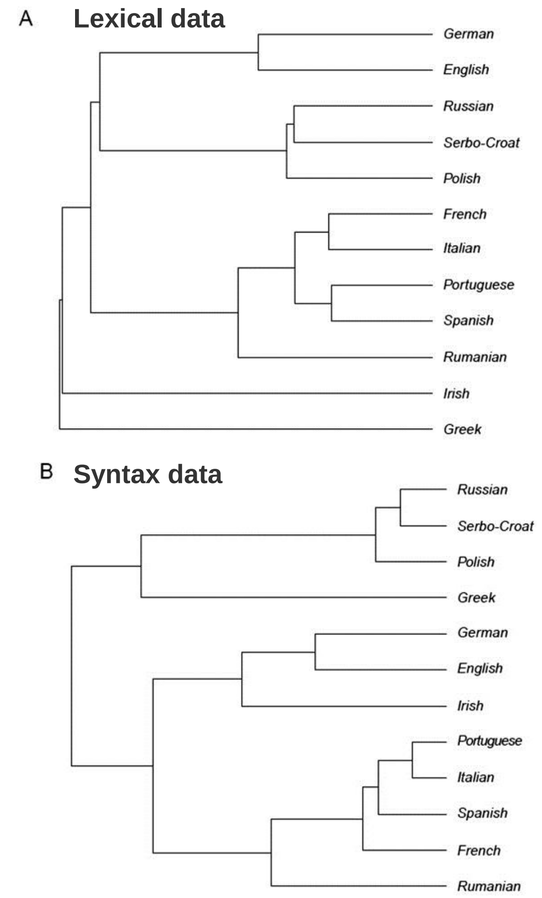

Longobardi et al 2015

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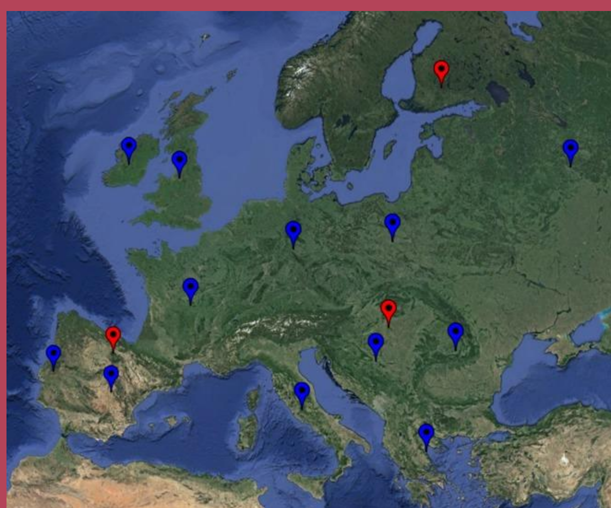

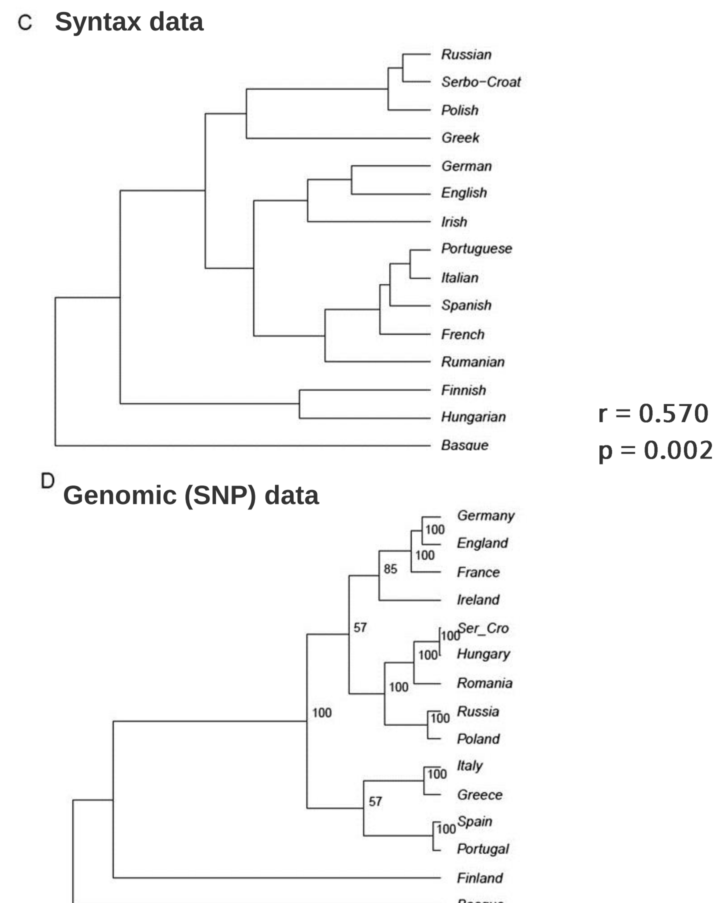

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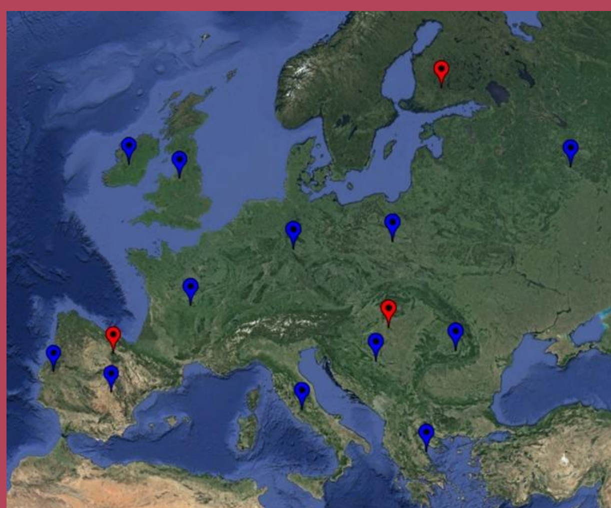

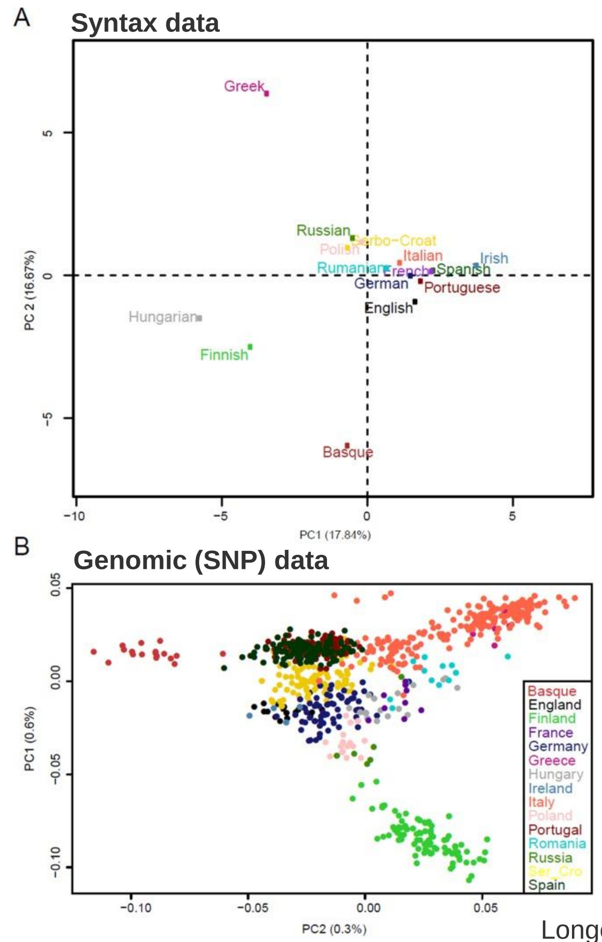

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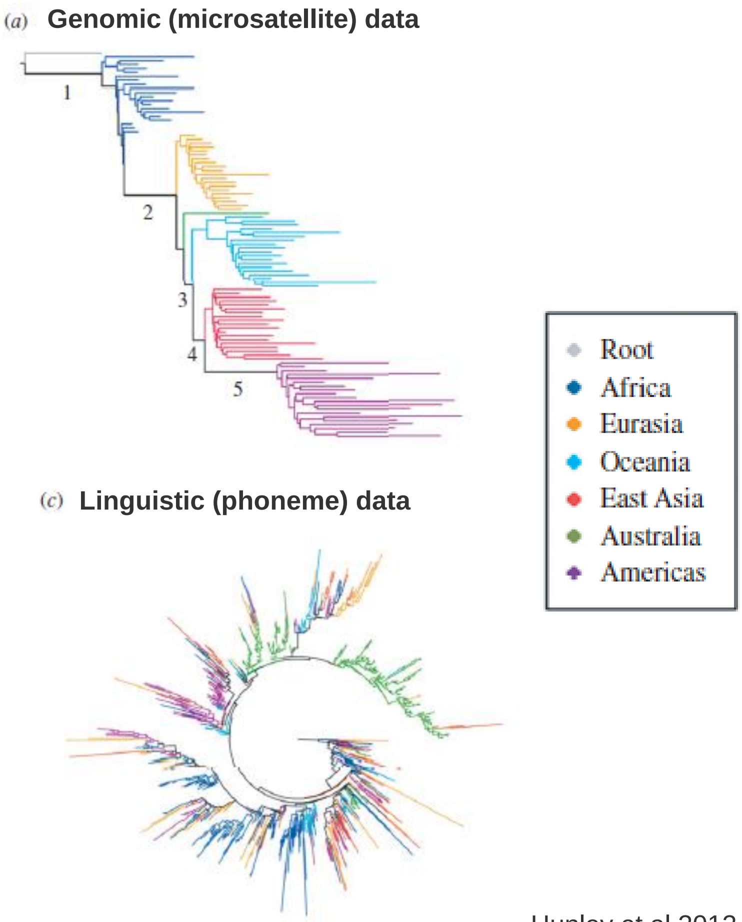

Hunley et al 2012

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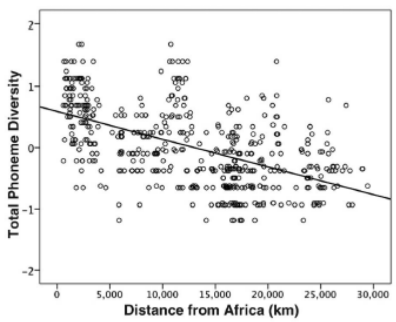

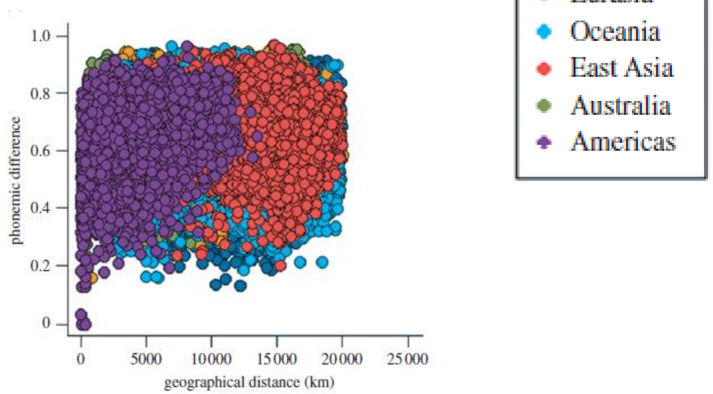

Atkinson 2011; Hunley et al. 2012

Co-evolution of genes and languages

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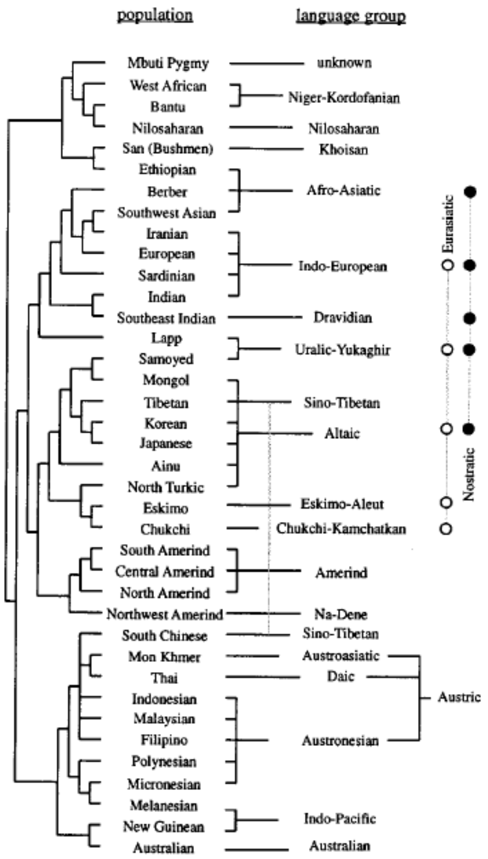

Cavalli-Sforza 1997

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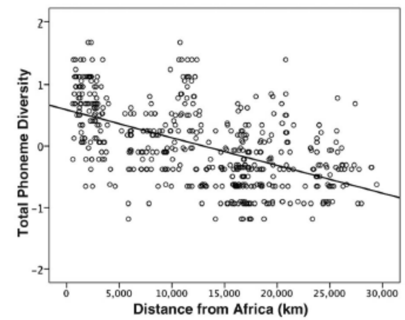

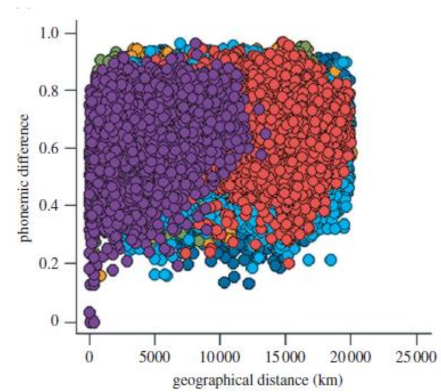

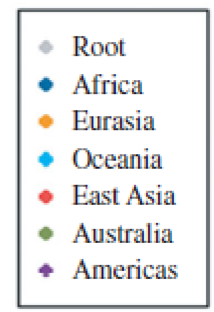

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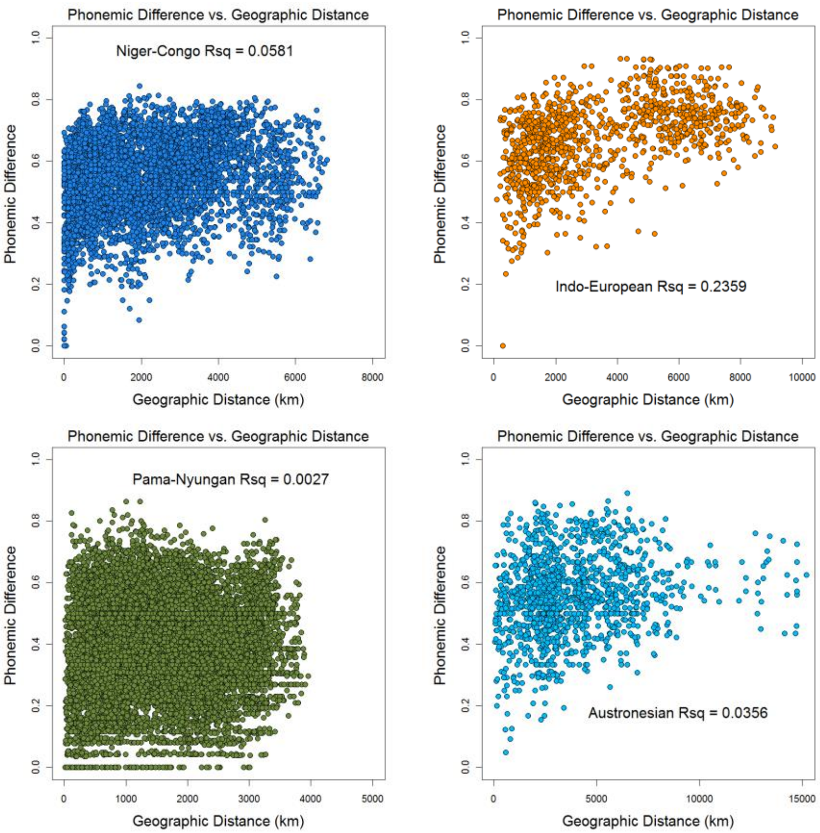

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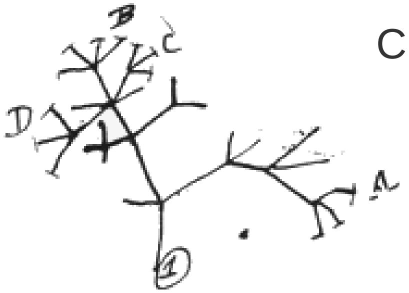


Charles Darwin, 1859

DFG Center Symposium

4th Annual Symposium
Ancient Connections in Eurasia

14-15 December 2018
Alte Aula, Münzgasse 30

Schedule

FRIDAY, 14TH DECEMBER

10:00 Welcome & Introduction to the DFG Center
Katerina Harvati and **Gerhard Jäger**

10:15 Symposium overview, **Hugo Reyes-Centeno**

Population dynamics of Western Eurasia from the Paleolithic to Bronze Age
Session chaired by George Perry

10:30 **Andaine Seguin-Orlando**
Upper Paleolithic genomes unveil anatomically modern humans' prehistory and social behavior

11:00 **Cosimo Posth**
Population history of Upper Paleolithic Europeans

11:30 **Michela Leonardi**
A Palaeolithic cultural shift suggested by genetic and genomic data

12:00 **Martin Bartelheim & Thomas Scholten**
The transition between the Chalcolithic and the Bronze Age in Western and Central Europe: Response to climatic change?

12:30 **Session Discussion**

13:00 **Lunch and Group Photo**

The Emergence of symbols in Western Eurasian Prehistory
Session chaired by Miriam Haidle

14:00 **Matteo Scardovelli**
Iconographic changes through time in Upper Palaeolithic parietal art

14:30 **Ewa Dutkiewicz & Christian Bentz**

SignBase – A data-driven approach to abstract signs in the Paleolithic

15:00 **Solange Rigaud**
Durable cultural traditions and lasting facets of past populations' ethnicity revealed by personal ornaments at the dawn of agriculture 8.000 years ago in Europe

15:30 **Session Discussion**

16:00 **Coffee break**

Adaptation, selection, and admixture in Eurasia
Session chaired by Katerina Harvati

16:30 **Ludovic Orlando**
Tracking six millenia of horse selection, admixture and management with complete genome time series

17:00 **Irina Morozova**
Anthropogenic selection: Changes in human genomes during 6000 years of European civilization

17:30 **Hannes Rathmann & Richard Posamentir**
Population history of southern Italy during Greek colonization inferred from skeletal remains

18:00 **Discussion**

Schedule

SATURDAY, 15TH DECEMBER

Population diversity and change in Eastern Eurasia

Session chaired by Gerhard Jäger

10:00	**Chao Ning** Genetic Perspective on the peopling of the Transeurasian (linguistic) region
10:30	**Martine Robbeets** Words, bones and millets in the Transeurasian languages
11:00	**Mark Hubbe** Rate of expansion and occupation of North Eurasia: contributions from the study of prehistoric cranial diversity
11:30	**Coffee break**
12:00	**George van Driem** The East Asian linguistic phylum: A reconstruction based on language and genes
12:30	**Mei-Shin Wu & Hugo Reyes-Centeno** Words and genes of Sino-Tibetan speakers
13:00	**Discussion**
13:30	**Lunch**

Methods in the study of bio-cultural co-evolution

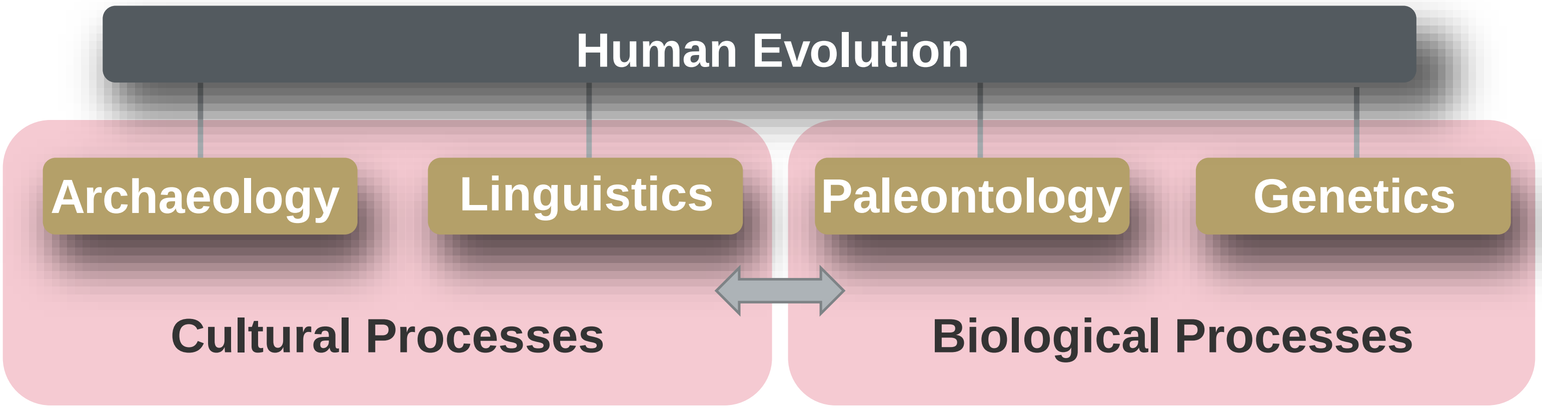
Session chaired by Christian Bentz

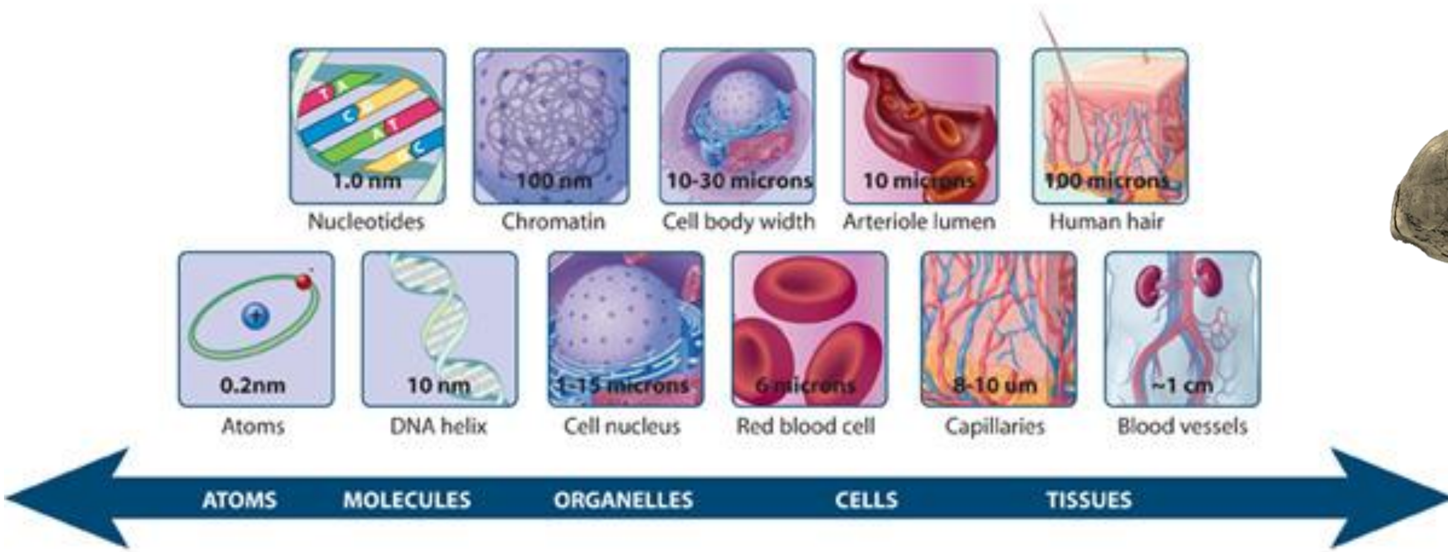
14:30	**Igor Yanovich** Languages and genes in Eurasia: a rigorous approach to their mismatches?
15:00	**Harald Hammarström** On the reasons for accelerated/delayed language change: A Eurasian study using the DiACL database
15:30	**Tiago Tresoldi** Computer-assisted data curation and analysis for the purpose of historical and typological language comparison
16:00	**Andrej Evteev** The level of correlation between mid-facial craniometric, neutral genetic and climatic distances completely depends on the scale of comparison
16:30	**Discussion**
17:00	**Coffee and Closing**

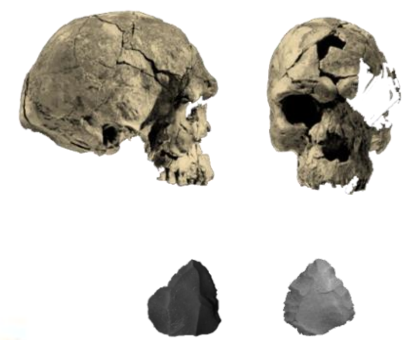
Next class: January 7th

- ❑ Adaptive and non-adaptive theories of language evolution
- ❑ Preadaptations to language
- ❑ Interfaces with archaeology and paleoanthropology

Co-evolution in human evolution







Nature Education 2010; White *et al.* 2003; Beyin 2013

Co-evolution in human evolution