EBERHARD KARLS
UNIVERSITÄT
TÜBINGEN

WORDS BONES GENES TOOLS
Tracking Linguistic, Cultural, and Biological Trajectories of the Human Past

DFG

Modern Human Origins

Hugo Reyes-Centeno, Yonatan Sahle, Christian Bentz

13 December 2017, Lecture 7

https://moodle02.zdv.uni-tuebingen.de/course/view.php?id=1932

Last week:

- **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?
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  What are the genetic signatures of the human expansion out of Africa?

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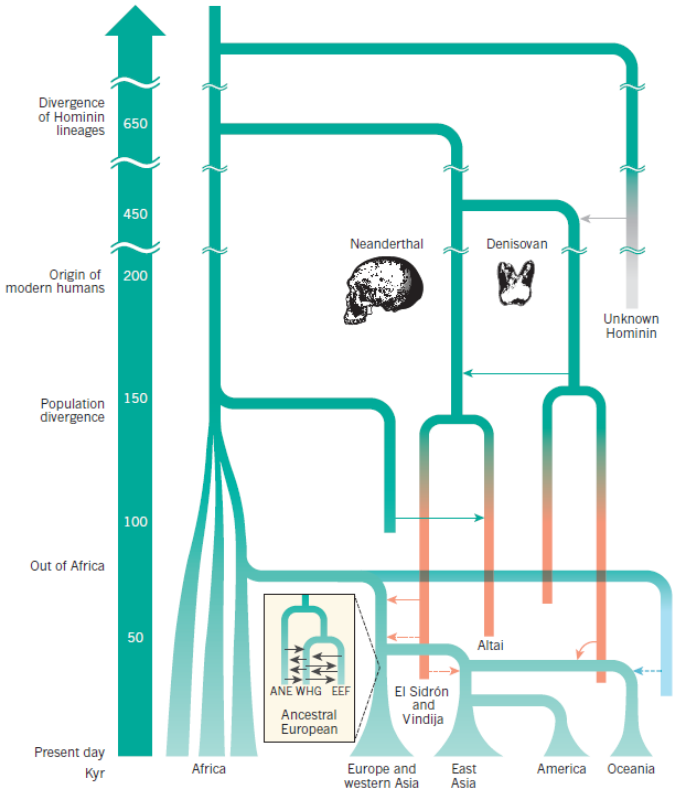

Nielsen et al 2017

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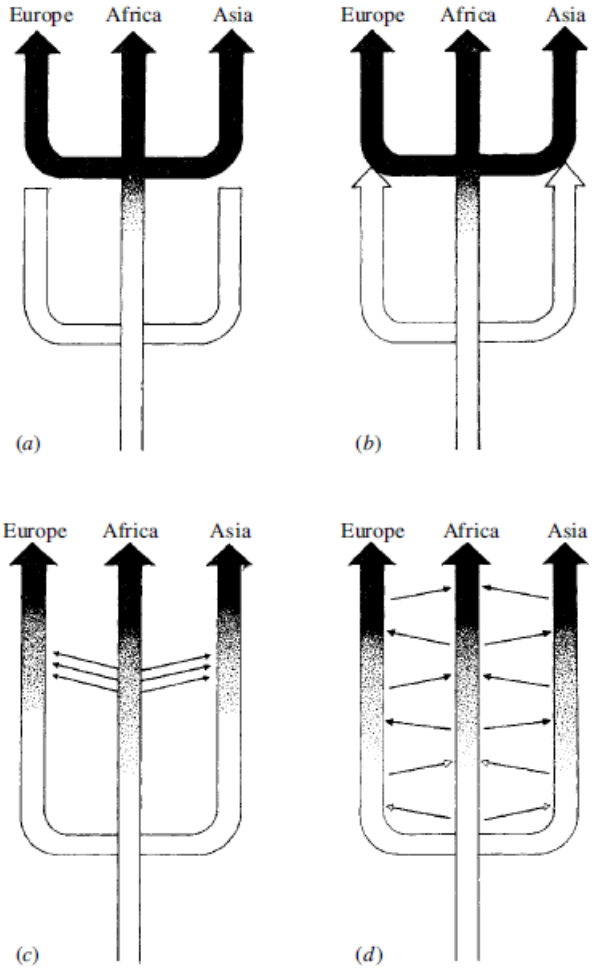

Stringer 2001

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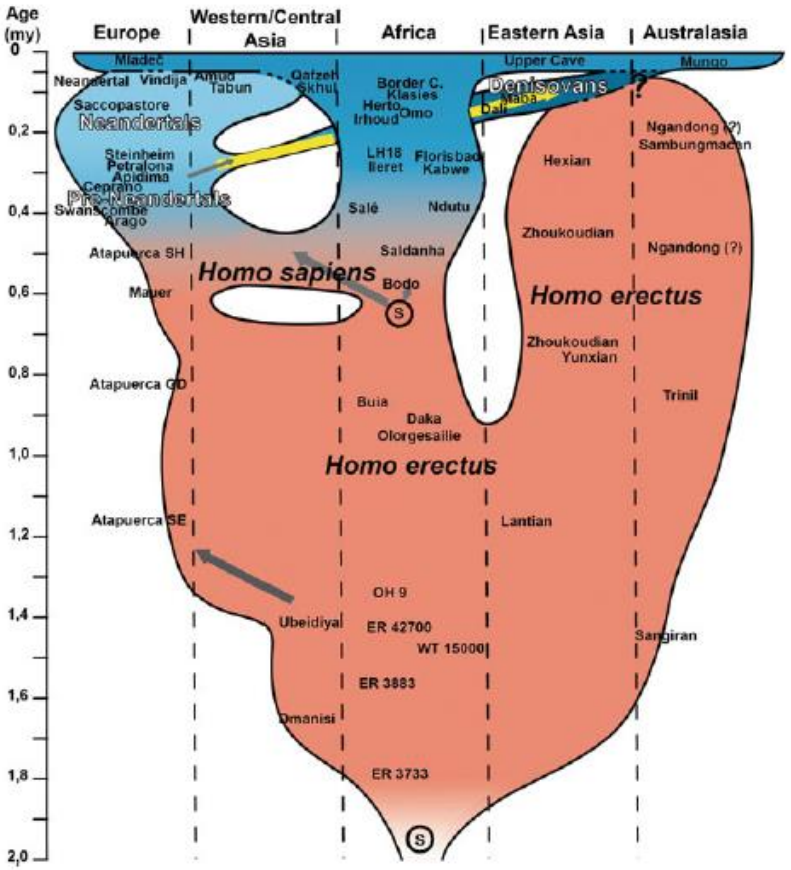

Fig. 7 Origin and evolution of *Homo sapiens*

Bräuer 2015

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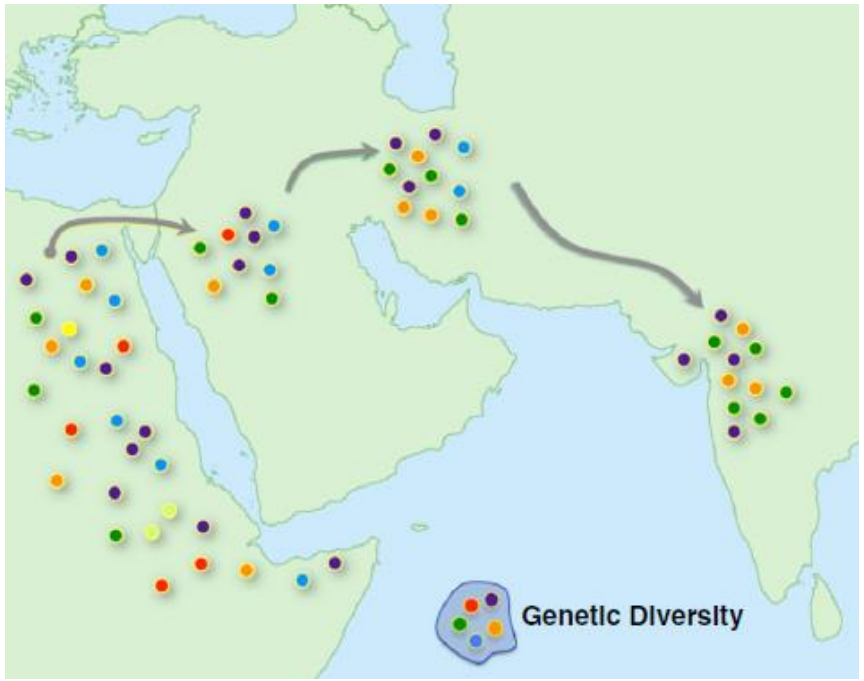

Henn et al. 2012

This lecture:

- **Modern human dispersals**
  When, how, and why did anatomically modern humans disperse 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?

Modern human dispersals

When, how, and why did anatomically modern humans disperse out of Africa?

Competing modern human dispersal models

- **Timing:**
  - ~130 ka
  - ~50 ka
- **Mode: Number of dispersals**
  - 1 Out-of-Africa event
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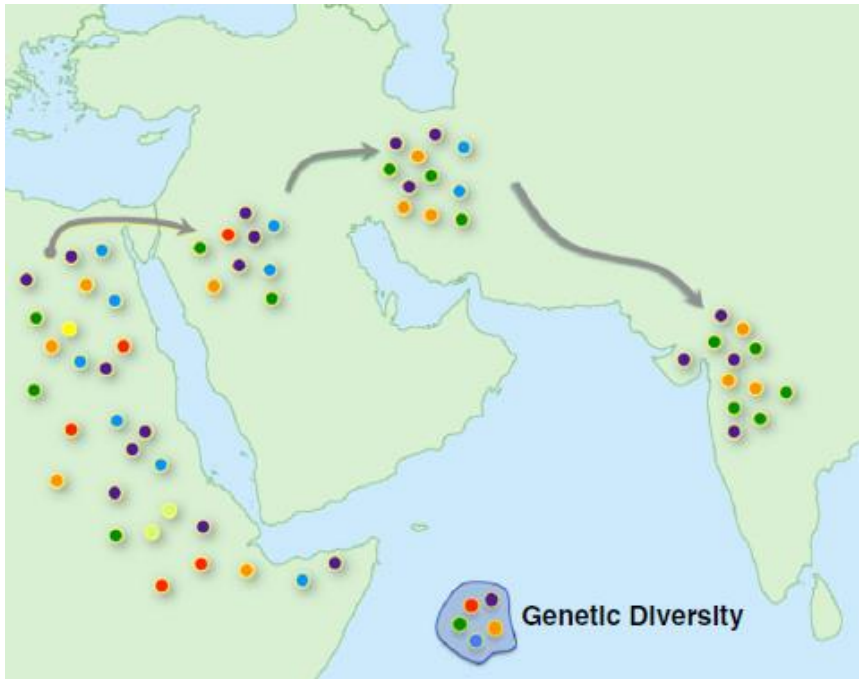

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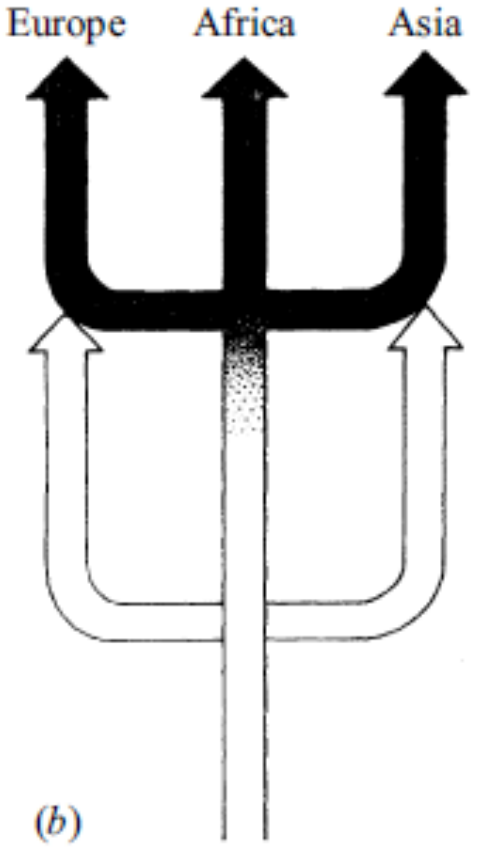

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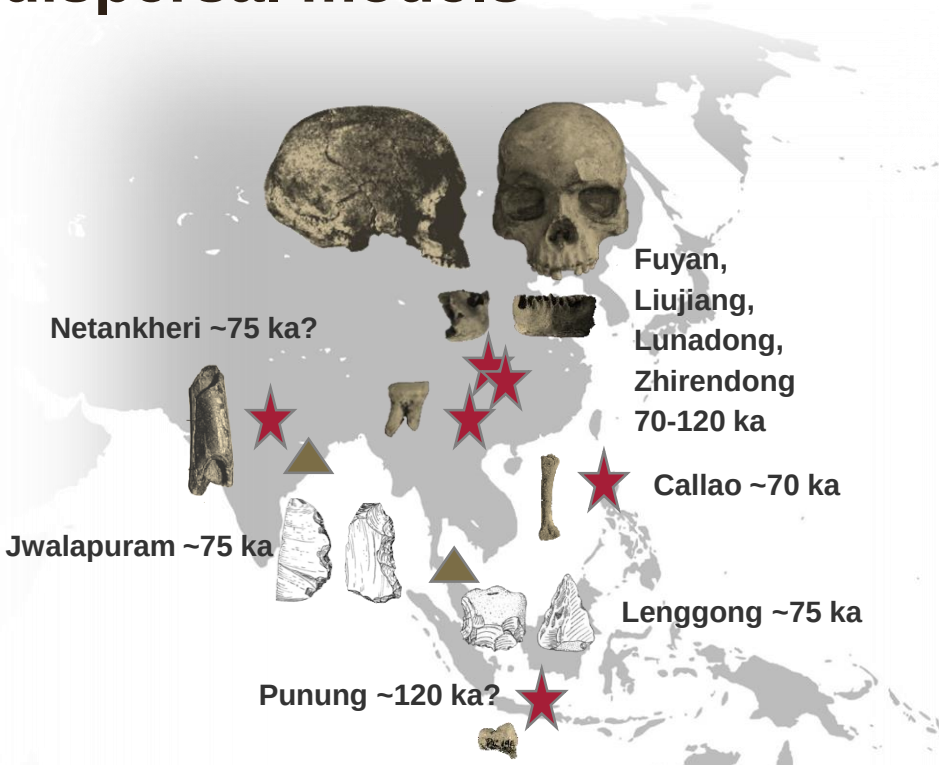

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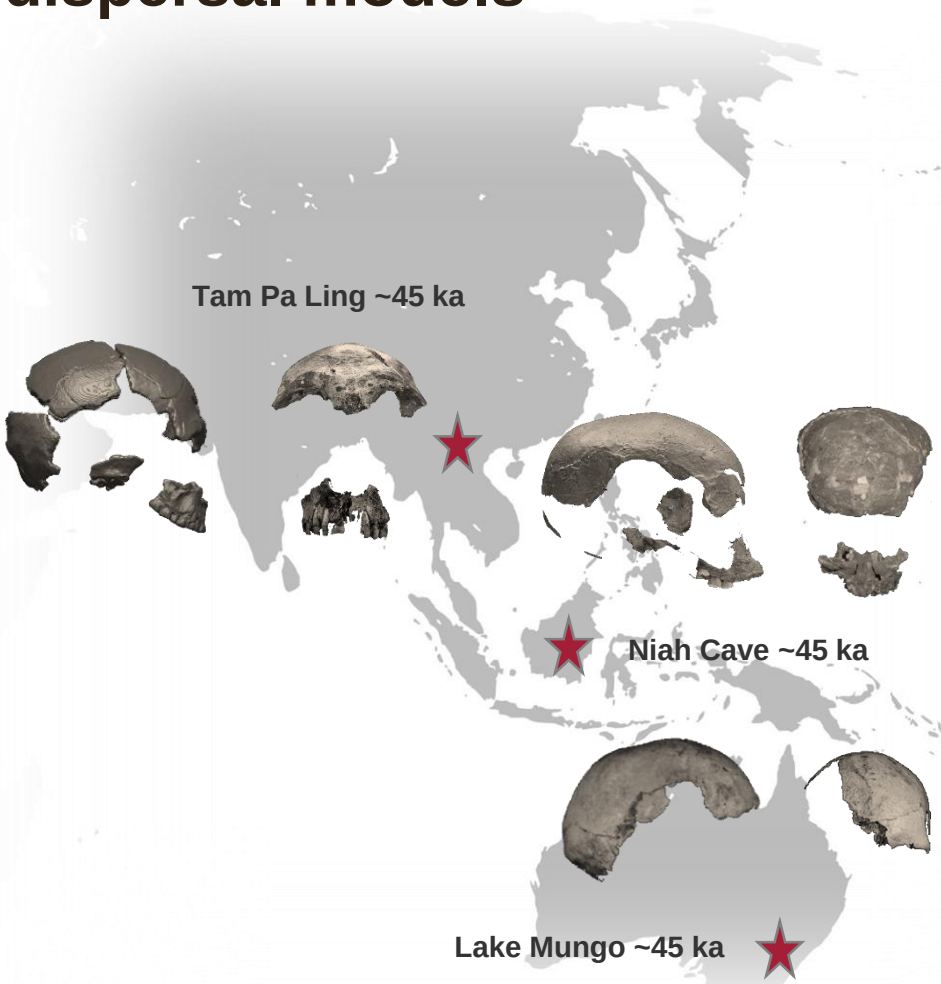
  - Occupation of Eurasia >45 ka



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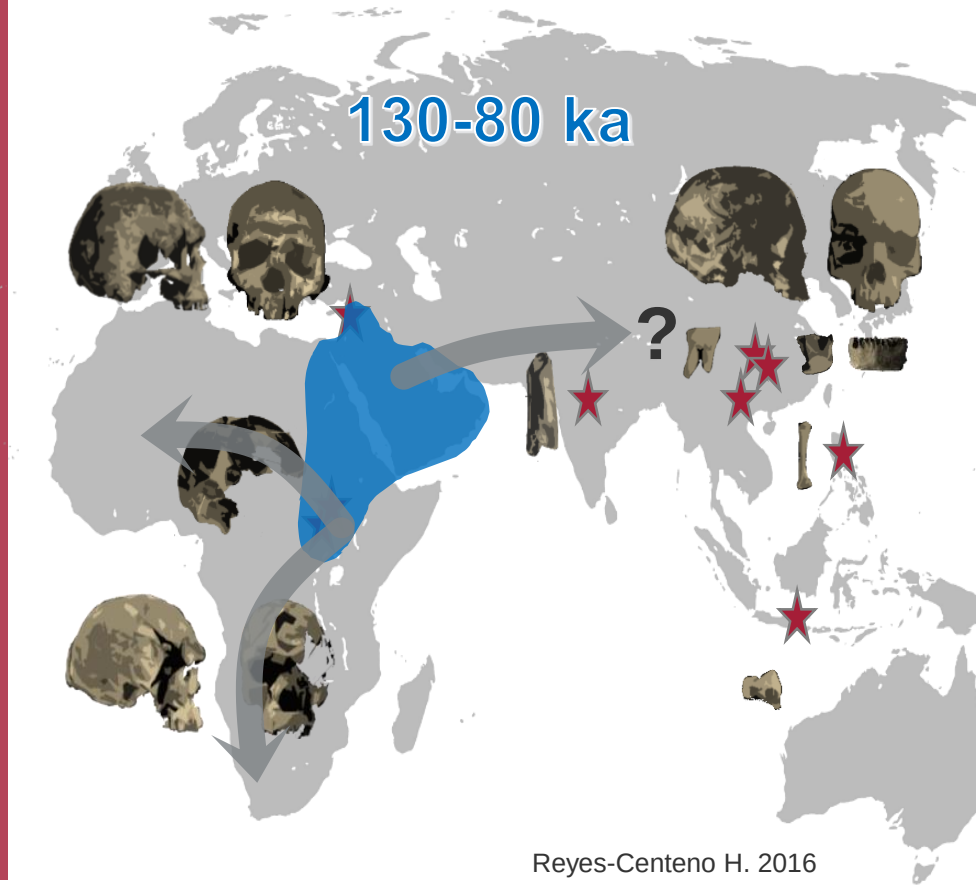

Reyes-Centeno H. 2016

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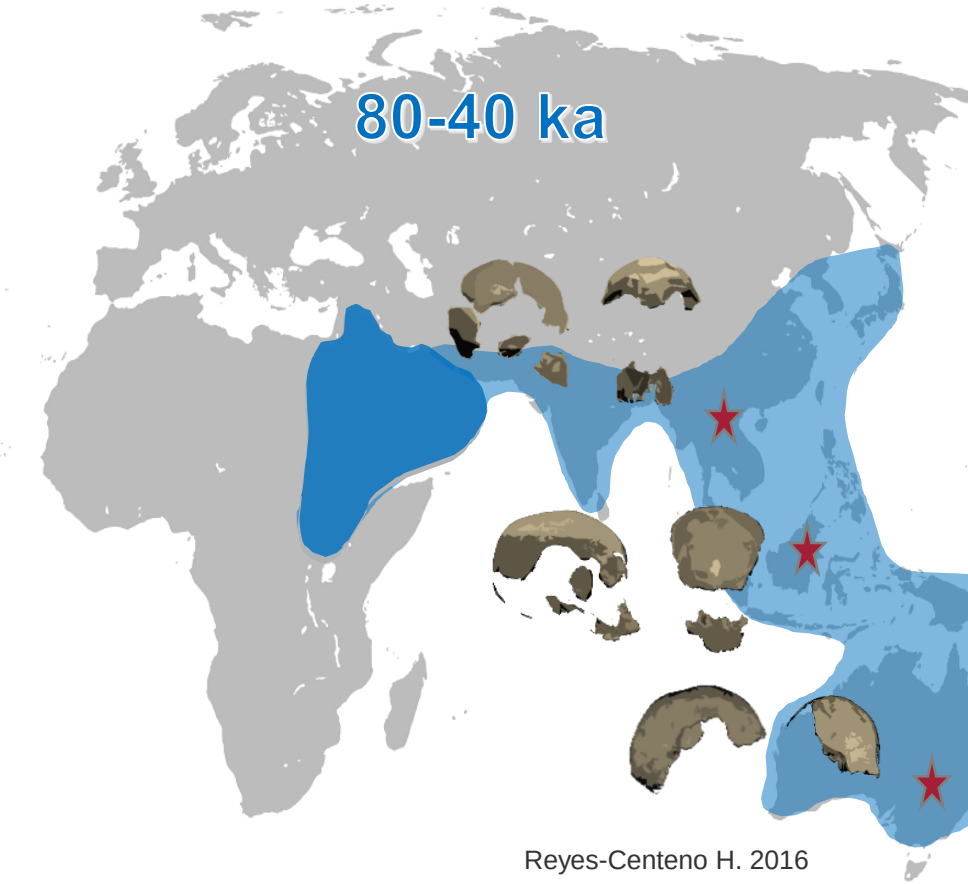

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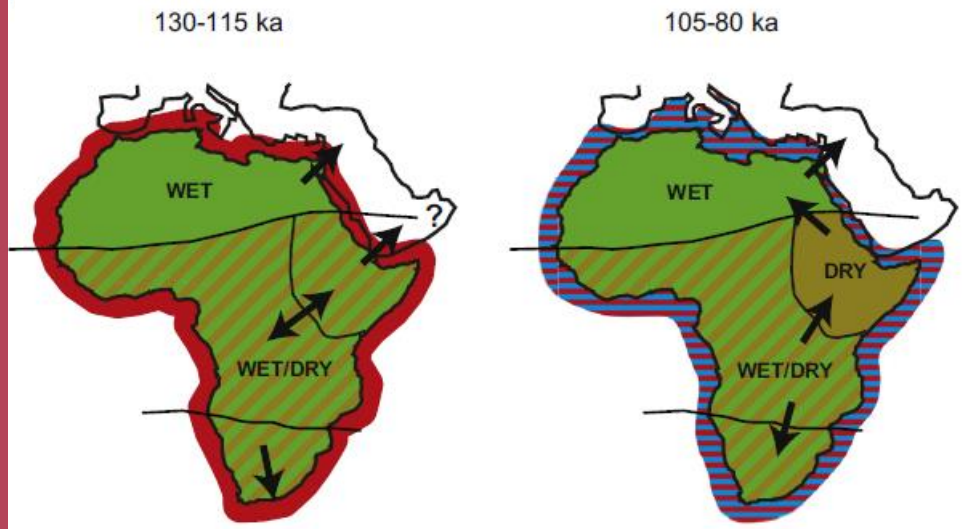

Blome et al. 2012

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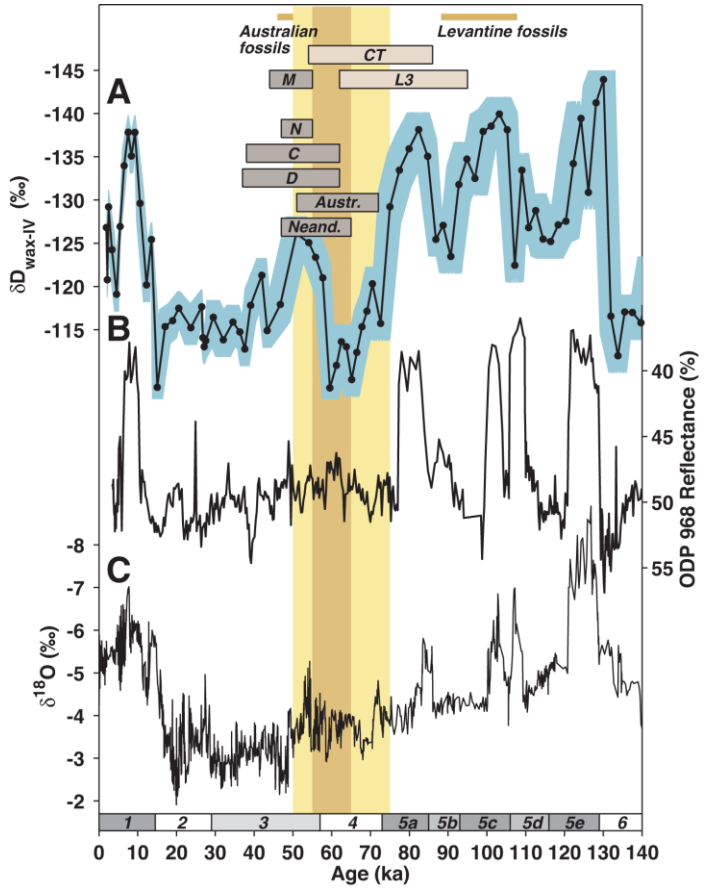

Tierney et al. 2017

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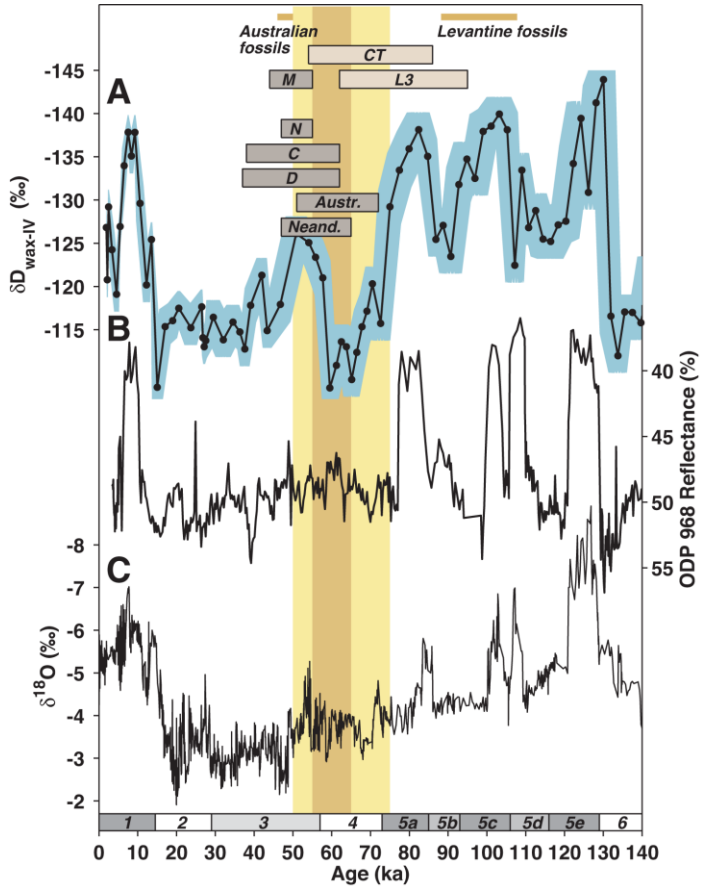
    - Why disperse during arid conditions time period?
    - Lag in genomic estimates?



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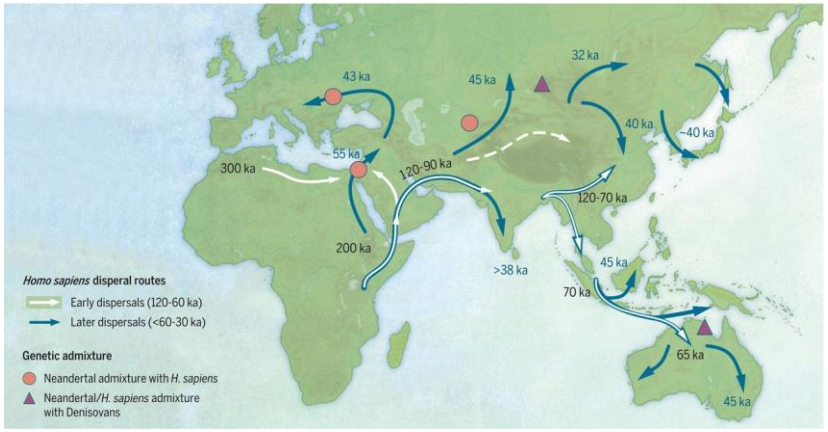
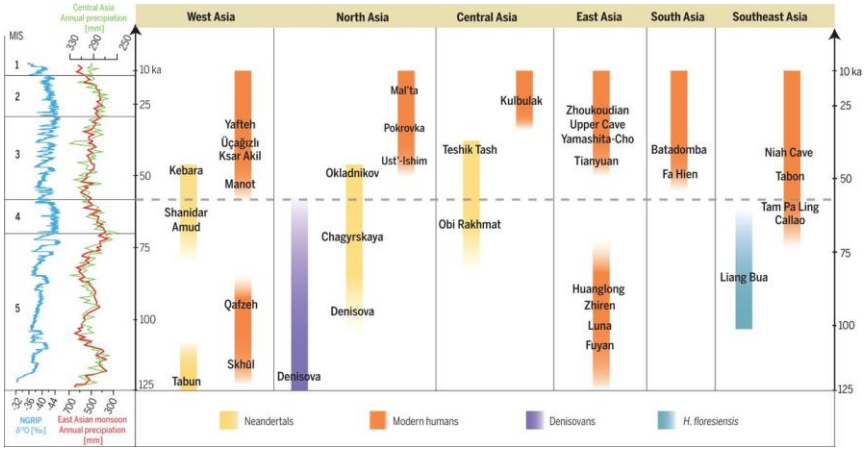
  - No fossils yet reported in southern Arabian Peninsula

Competing modern human dispersal models

- **Revised view:**
  - Expansion ~130 ka to the Levant and possibly further into Eurasia, followed by extinction
    - Small populations?
    - Contributions to Neanderthals?
    - Competition with other hominins?
    - Middle Paleolithic / MSA toolkit?
  - Major dispersal between ~80-50 ka





Bae et al 2017

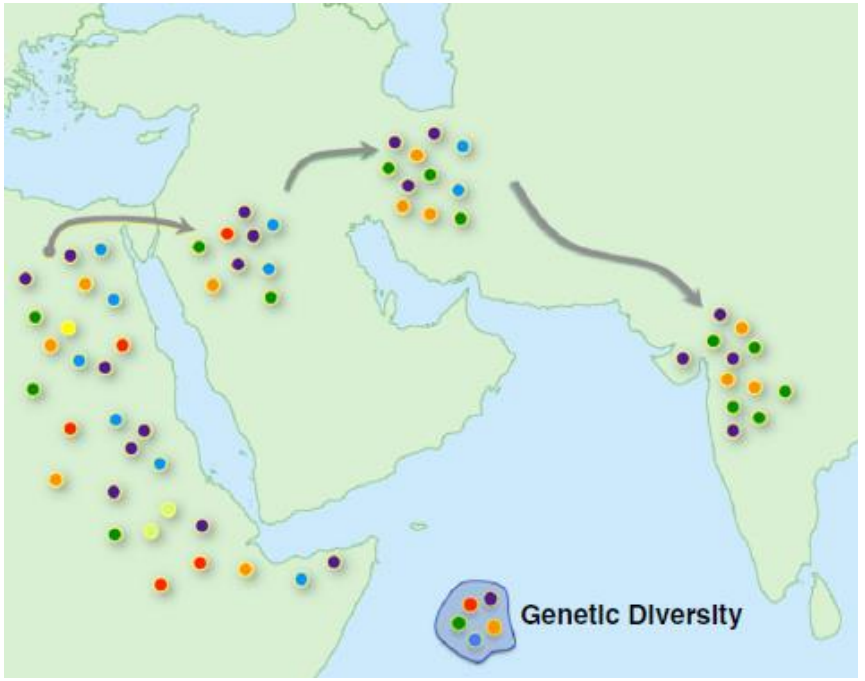
Neutral & adaptive evolution

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

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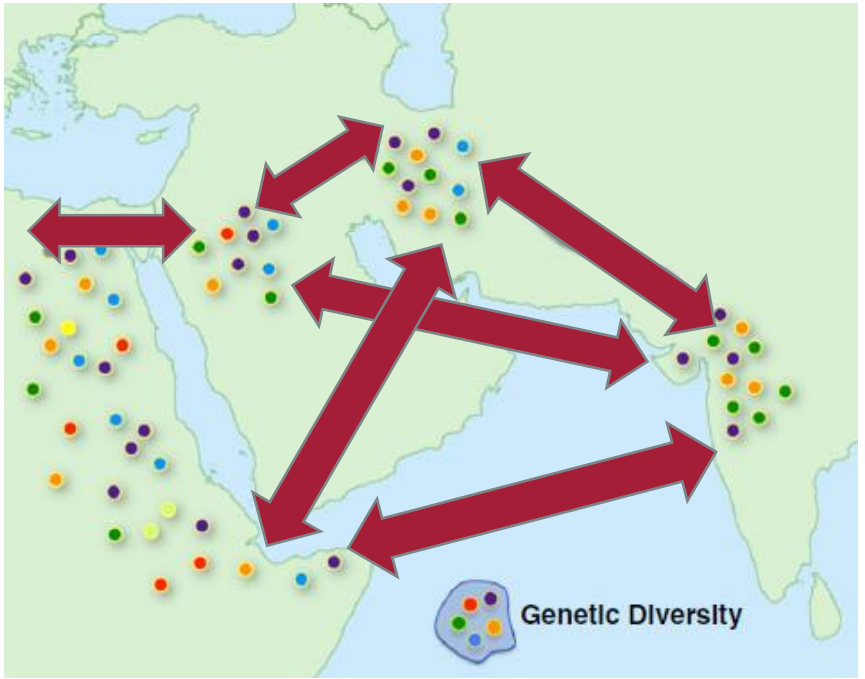

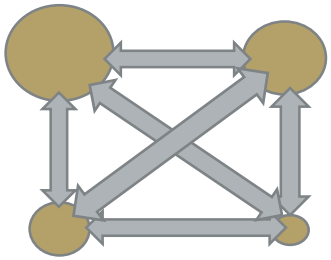

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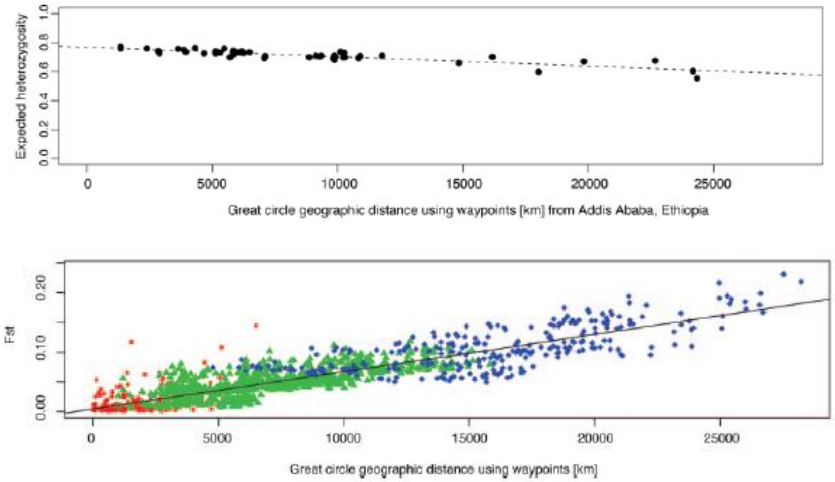

Ramachandran et al. 2005

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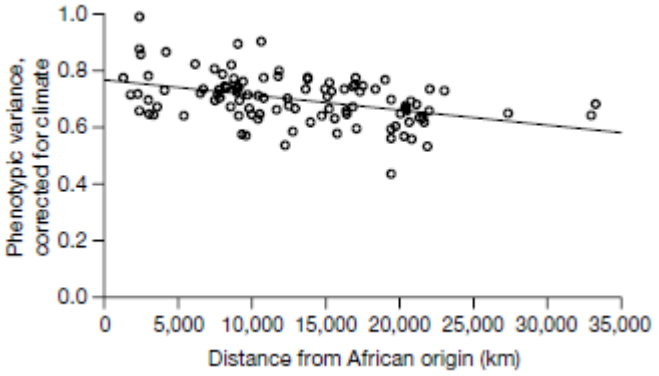

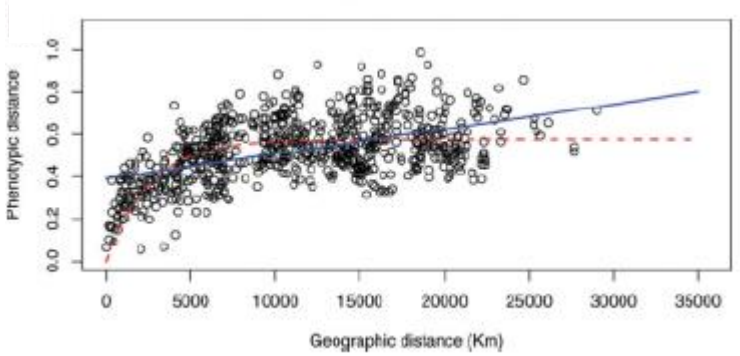

Manica et al. 2007; Betti et al. 2011

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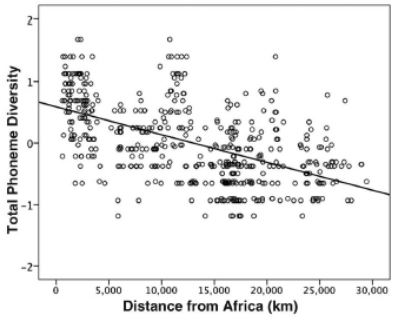

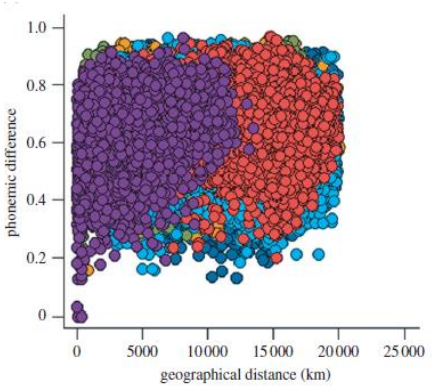

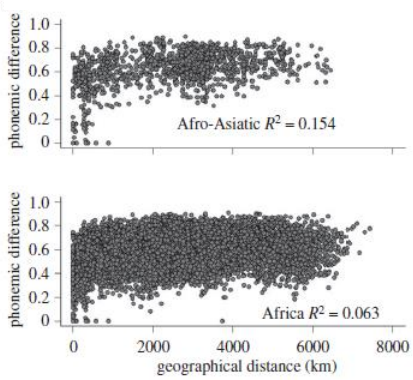

Atkinson 2011; Hunley et al. 2012

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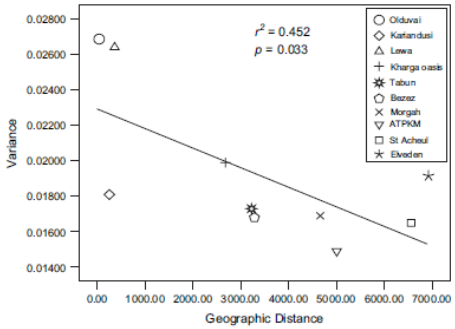

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Lycett & von Cramon-Taubadel 2008

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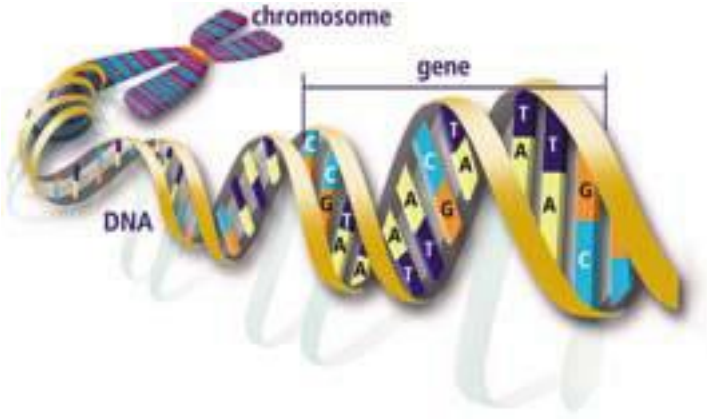

US Department of Energy

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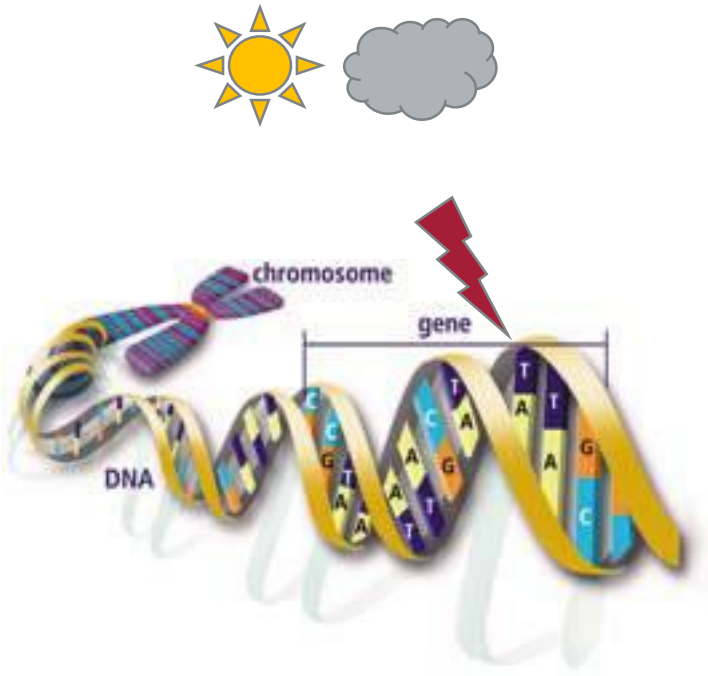

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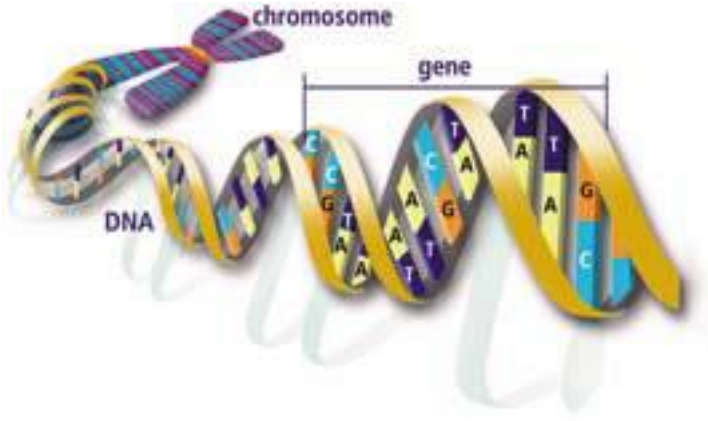
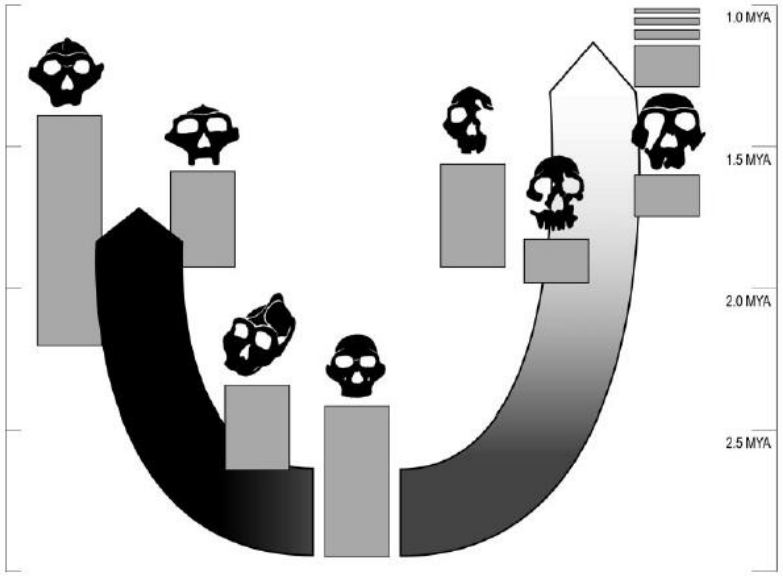


US Department of Energy

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

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- Netural evolution is thought to play the primary role in recent human evolution

Weaver et al 2007

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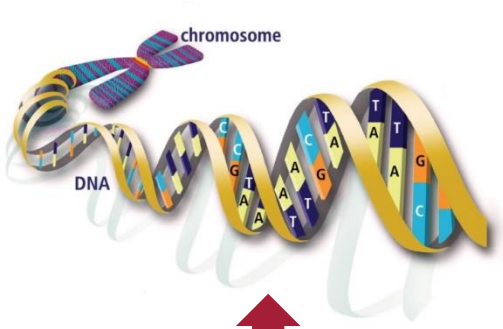
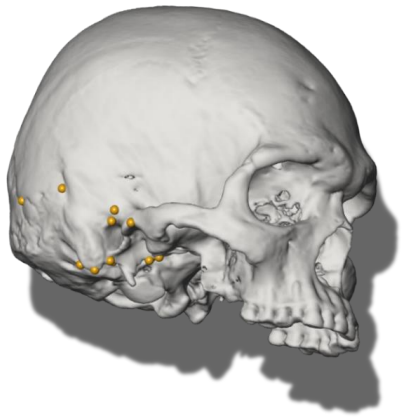
  - Direct comparison of netural genomic and phenotypic variation between populations is significant





US Department of Energy; Reyes-Centeno et al 2017

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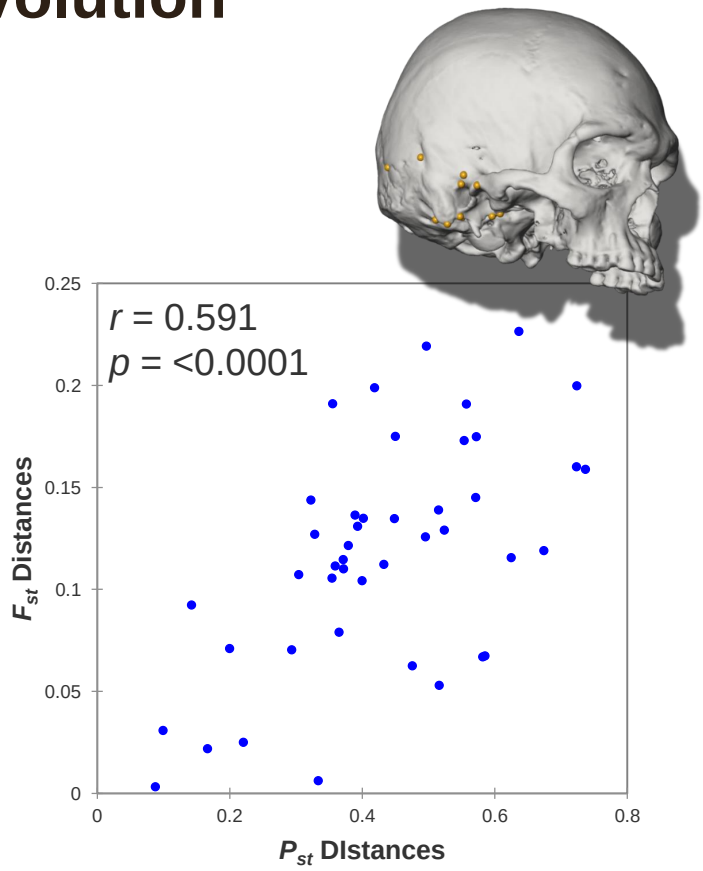

Reyes-Centeno et al 2017

Drift and selection in human evolution

- Adaptive evolution is thought to have played an important role in early human evolution
- Netural evolution is thought to play the primary role in recent human evolution
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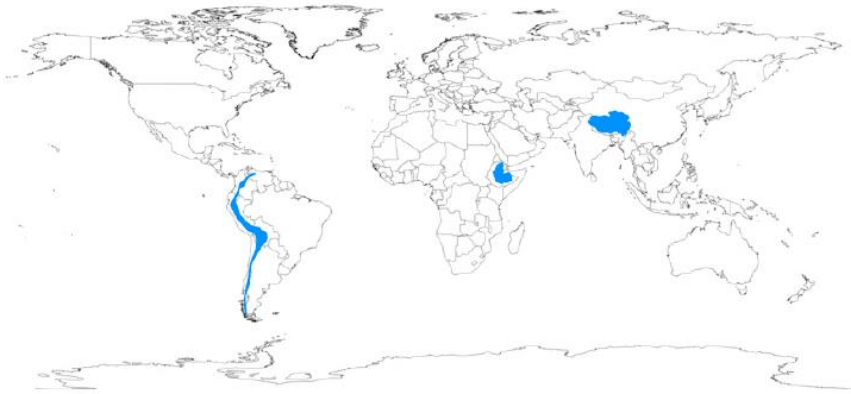
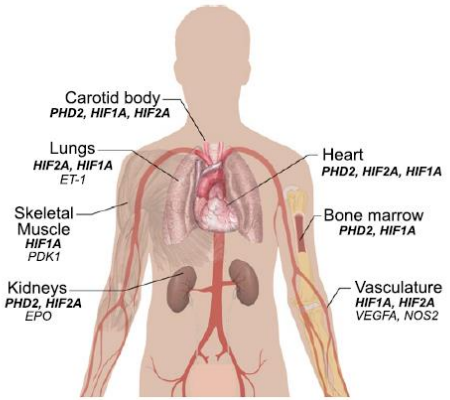
  - e.g. adaptation to high altitude environments: independent (convergent) evolution and possible hominin introgression in Tibet





Bingham & Lee 2014

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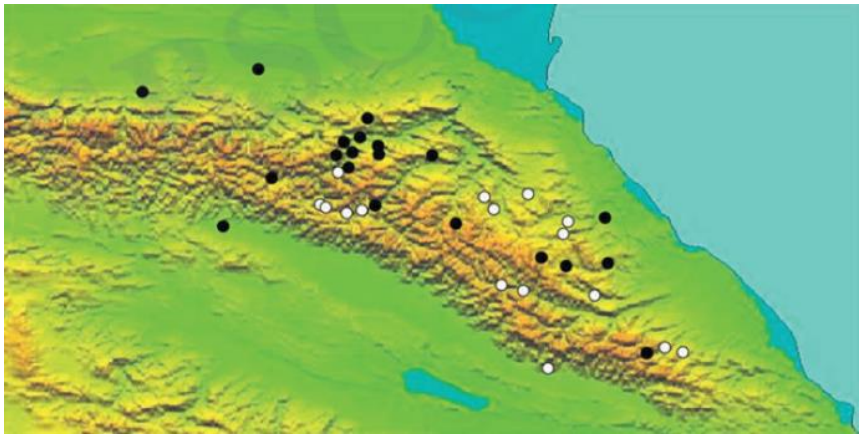


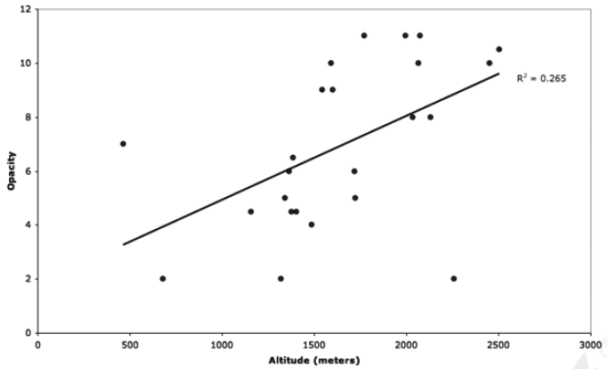


Fig. 2. Altitude (*x*-axis) by opacity (*y*-axis). Number of languages: $N = 26$. Shared variance: $R^2 = 0.27$

Nichols 2013

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
- 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

Co-evolution

Do the genotype and phenotype co-evolve?

Co-evolution in human evolution

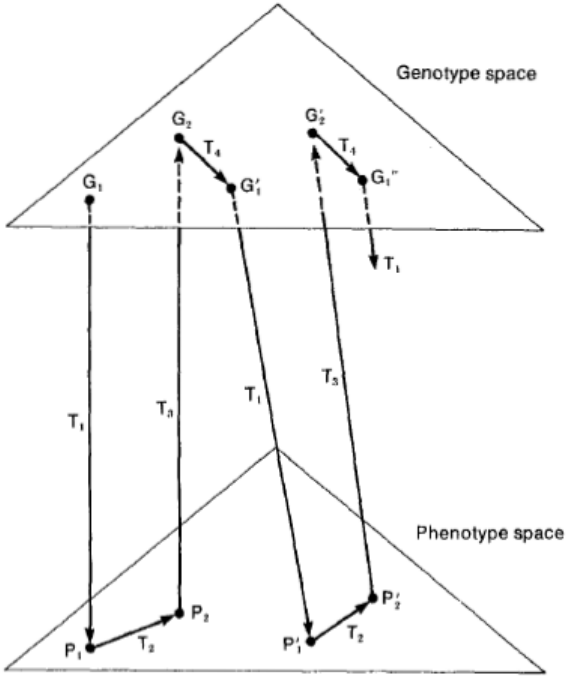
- Darwin‘s hypothesized the co-evolution of languages and genes

“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 in human evolution

- Darwin's hypothesized the co-evolution of languages and genes
- Theoretical framework: one can trace how the genotype and phenotype affect each other
  - Selection acts on phenotypes and gene frequency changes as a consequence



Lewontin et al 2010

Co-evolution in human evolution

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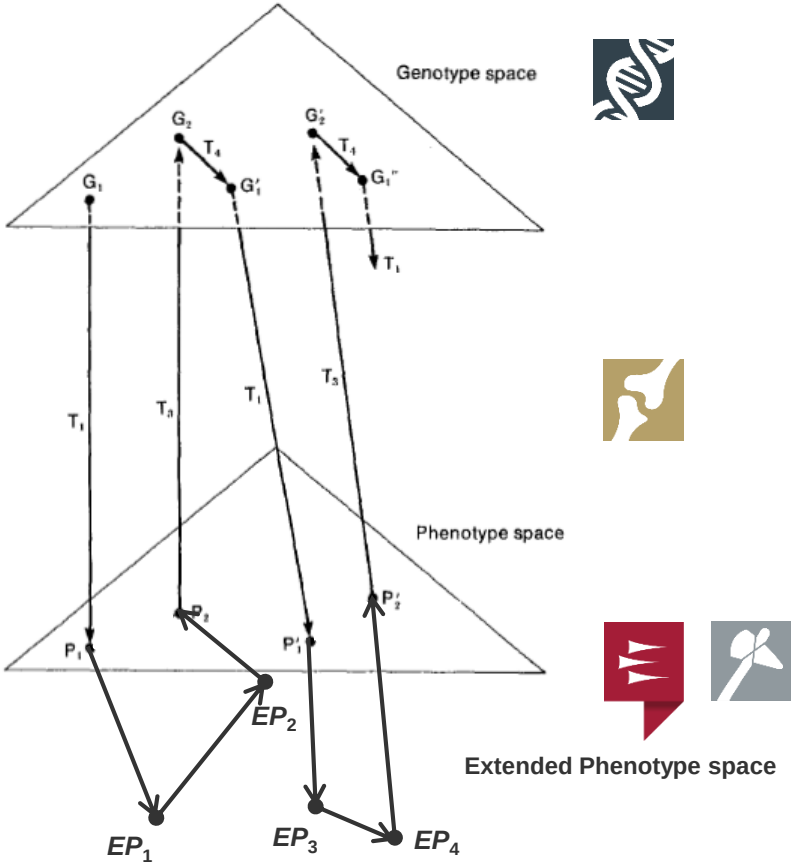
  - Language and behavior can be considered as an extended phenotype



Lewontin et al 2010

Co-evolution in human evolution

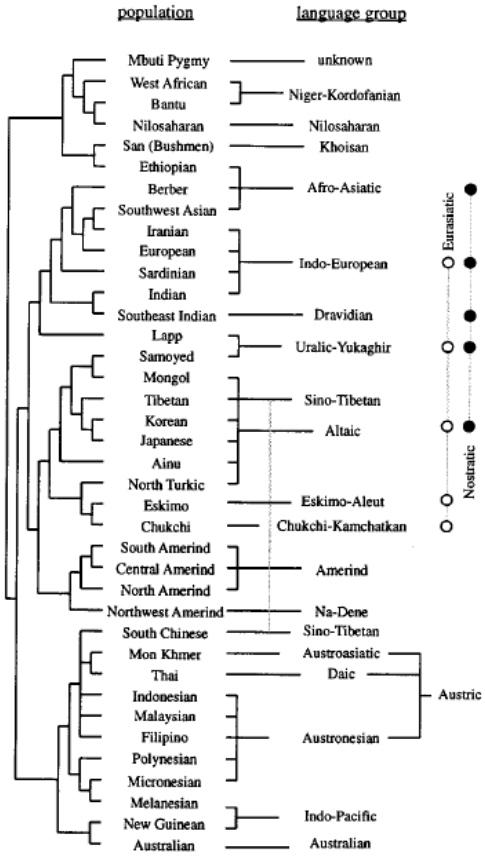
- Darwin's hypothesized the co-evolution of languages and genes
- Theoretical framework: one can trace how the genotype and phenotype affect each other
  - Selection acts on phenotypes and gene frequency changes as a consequence
  - Language and behavior can be considered as an extended phenotype
  - 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

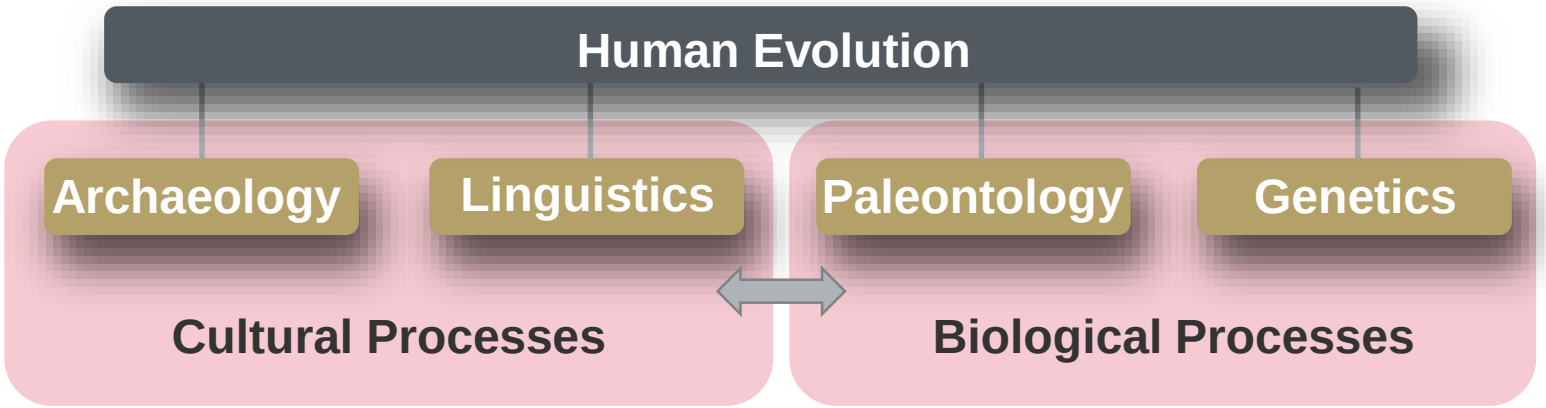
Co-evolution in human evolution

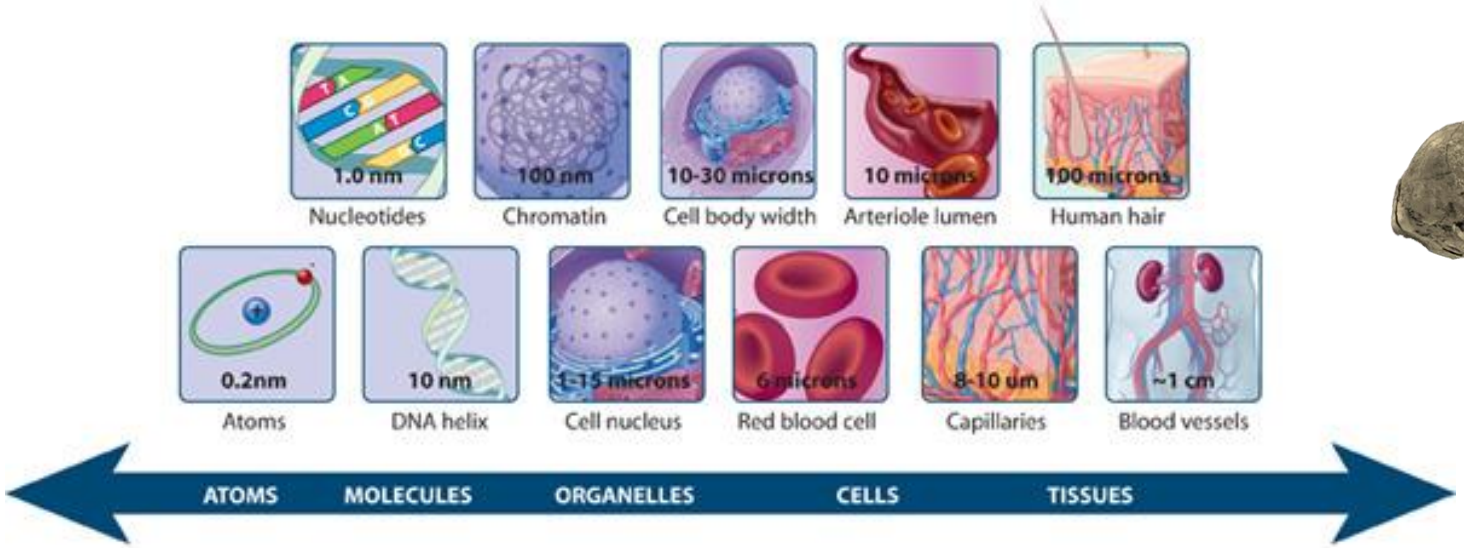
- Darwin‘s hypothesized the co-evolution of languages and genes
  - Language and gene trees sometimes mirror each other, but other times do not
  - At the level of language families, there is a high correlation but not always at lower language levels

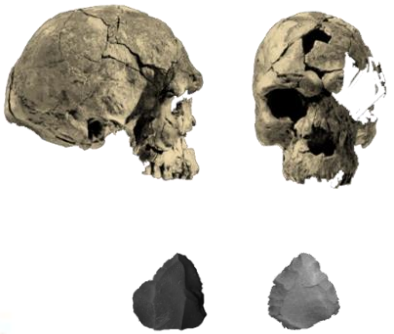


Cavalli-Sforza 1997

Co-evolution in human evolution







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

Co-evolution in human evolution