

Contemporary and adaptive human evolution

- thorough the last 40000 years human evolution accelerated, in the last **5000 years 100x**
- differentiation of populations are enhancing, despite the high gene flow (high migration rate)
- genes related to response to disease, food, sperm production
- drivers: population growth, diversity of food resources, medicine
- one of the fastest evolving genes is *human accelerated region 1 (HAR1)* – responsible for brain development. Random parts of the human genome is similar to the genome of chimpanzee by 98%, and HAR1 only by 85%.

Recent acceleration of human adaptive evolution

John Hawks, Eric T. Wang, Gregory M. Cochran, Henry C. Harpending, and Robert K. Moyzis

[+ See all authors and affiliations](#)

PNAS December 26, 2007 104 (52) 20753-20758; <https://doi.org/10.1073/pnas.0707650104>

„In the current study, a team of researchers led by paleoanthropologist Henry Harpending of the University of Utah in Salt Lake City analyzed DNA from 270 individuals in the International HapMap Project, an effort to identify variations in human genes that cause disease. The team searched for single-nucleotide polymorphisms (SNPs)--mutations in an allele that spread throughout a population--and scanned sequence data from Europeans, Africans, and Asians. The researchers searched for SNPs that were flanked by tens of thousands of bases of identical DNA in many individuals in a population, because this suggests that the mutation is advantageous and under recent selection pressure to be preserved in a lineage”

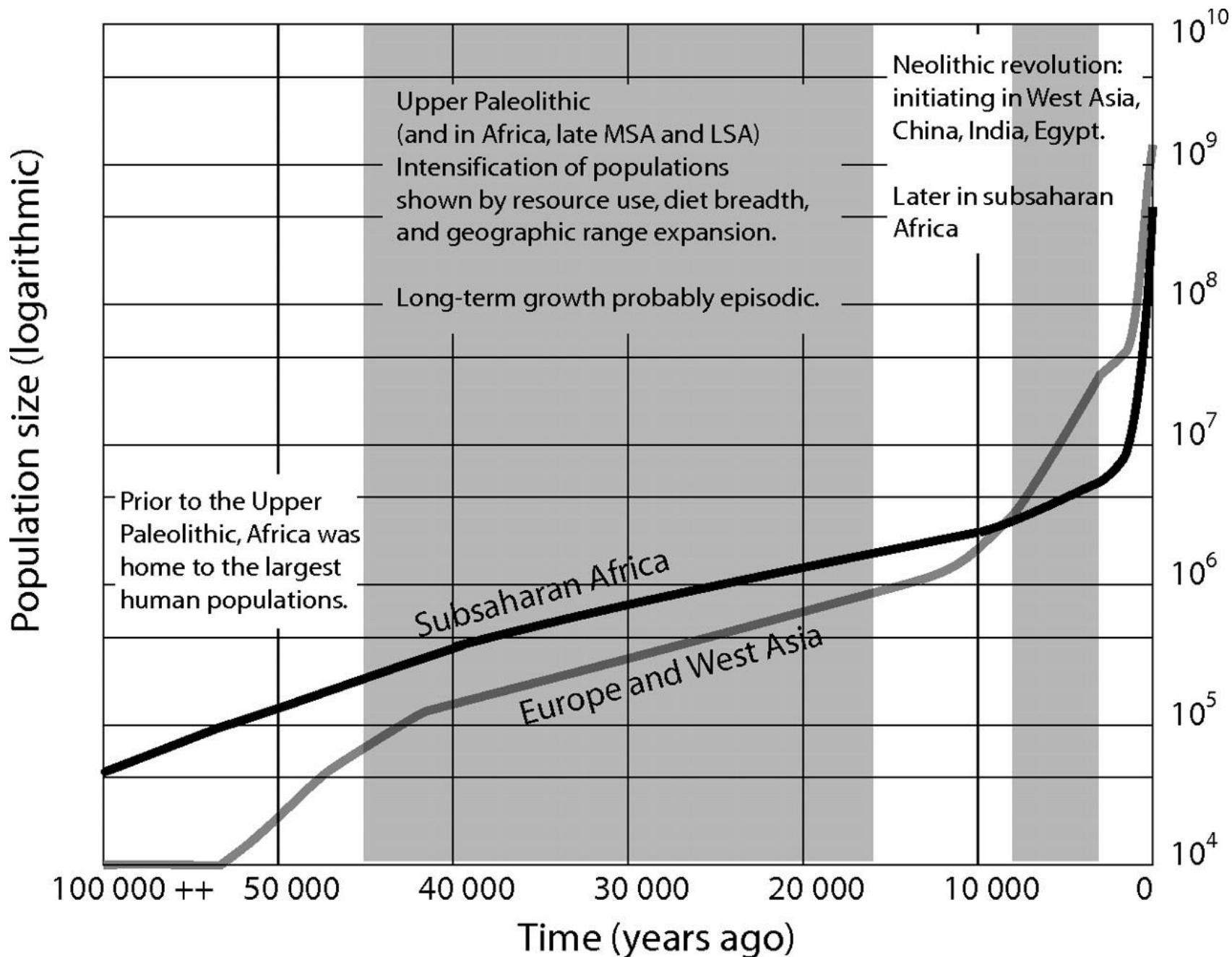
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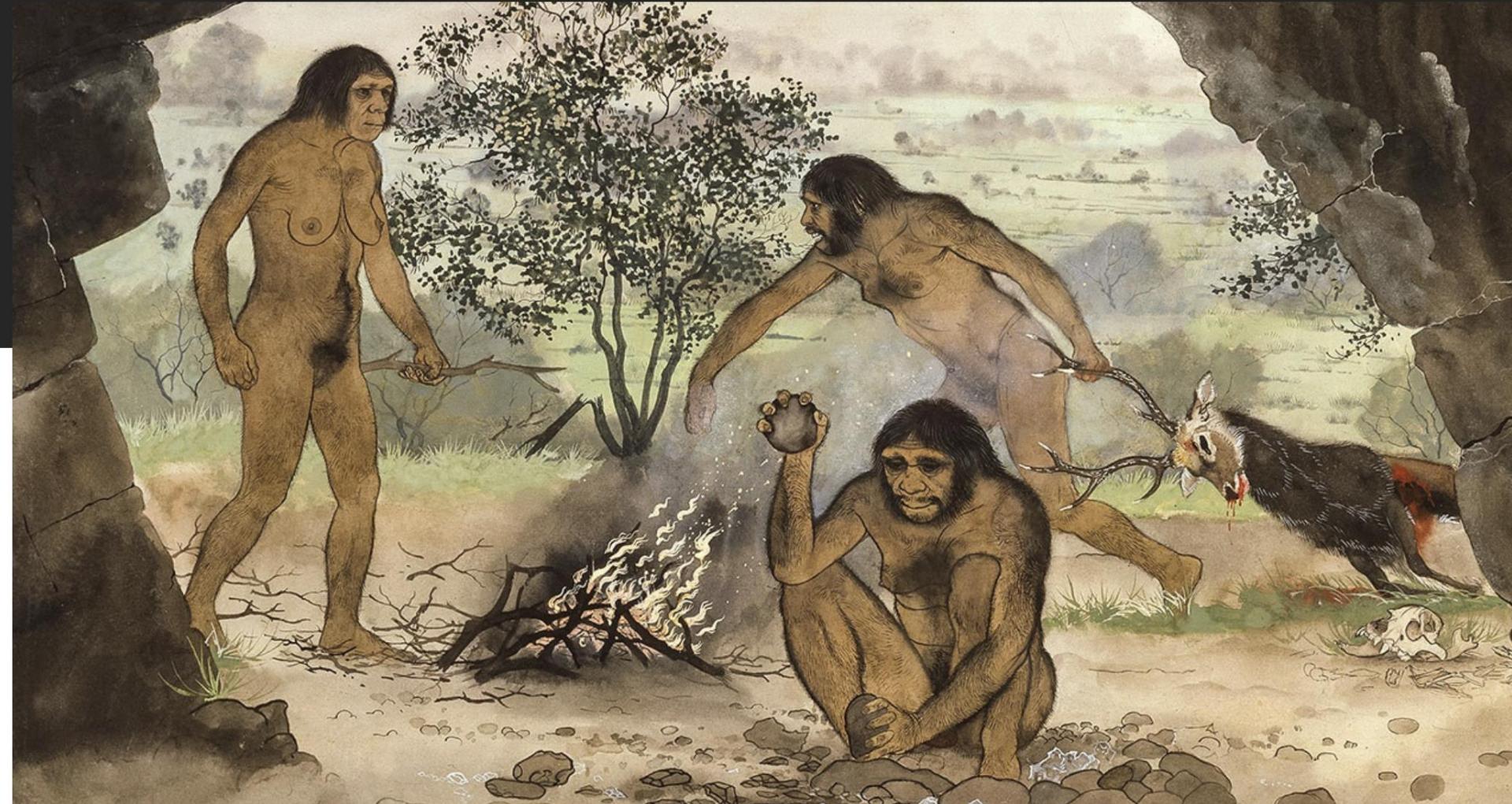
„Larger populations generate more new selected mutations, and we show the consistency of the observed data with the historical pattern of human population growth. The rate of adaptive evolution in human populations has indeed accelerated within the past 80,000 years. Demographic growth intensified during the Holocene, as domestication centers in the Near East, Egypt, and China underwent expansions commencing by 10,000 to 8,000 years ago. Population growth in the Upper Paleolithic and Late Middle Stone Age began by 50,000 years ago.“



How sliced meat drove human evolution

Chewing less may have allowed early humans to evolve features needed for speech

9 MAR 2016 • BY LIZZIE WADE



[nature](#) > [letters](#) > [article](#)

[Published: 09 March 2016](#)

Impact of meat and Lower Palaeolithic food processing techniques on chewing in humans

[Katherine D. Zink](#)  & [Daniel E. Lieberman](#) 

[Nature](#) 531, 500–503 (2016) | [Cite this article](#)

- human ancestors were **eating meat already 2.5 million years ago**
- the use of **primitive stone tools** for meat slicing and plant root pounding decreased the required number of chews per unit of time
- smaller teeth and jaw were physical features required for speech
- **cooking** appeared later on, became widespread around 500,000 years ago, enabled further decreasing in chewing and accelerated speech evolution

Detection of human adaptation during the past 2000 years

[YAIR FIELD](#), [EVAN A BOYLE](#), [NATALIE TELIS](#), [ZIYUE GAO](#), [KYLE J. GAULTON](#), [DAVID GOLAN](#), [LOIC YENGO](#), [GHISLAIN ROCHELEAU](#), [PHILIPPE FROGUEL](#), [...]

[JONATHAN K. PRITCHARD](#)

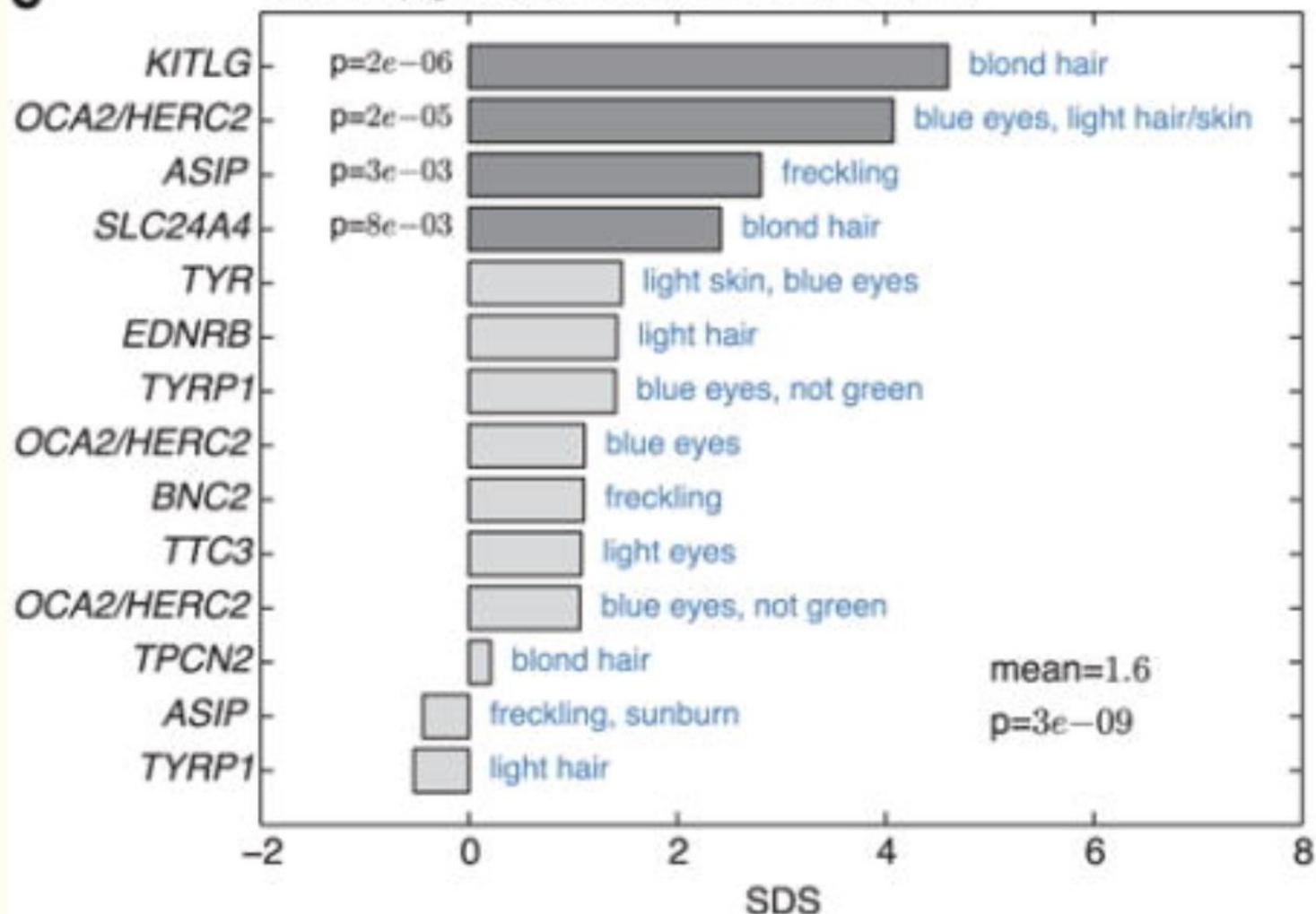
+2 authors

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*„Applied to data from the **UK10K** Project, SDS reflects allele frequency changes in the ancestors of modern Britons during the past ~2000 to 3000 years. We see strong signals of selection at **lactase** and the **major histocompatibility complex**, and in favor of **blond hair** and **blue eyes**. For polygenic adaptation, we find that recent selection for increased **height** has driven allele frequency shifts across most of the genome. ”*

*SDS - singleton density score- points to recent changes of allelic frequencies; positive values indicate that recently emerged allele increased in frequency

C SDS for pigment-associated GWAS variants



GENES WITH ALTERED ALLELIC FREQUENCIES

INCREASED FREQUENCY OF BLOND HAIR AND
BLUE EYES

SDS signals for a curated set of segregating variants with known effects on pigmentation shows overall increase in derived allele frequencies (one-sided P values).

- Blond hair- A-G mutation in regulatory gene sequence which causes difference in gene expression of the gene only in hair follicle- it is not related to blue eyes
- northern Europe- 11000 years ago- hypothesis: food shortage- many males died while searching for food- excess of females and high competition for paternal resources

How women evolved blond hair to win cavemen's hearts

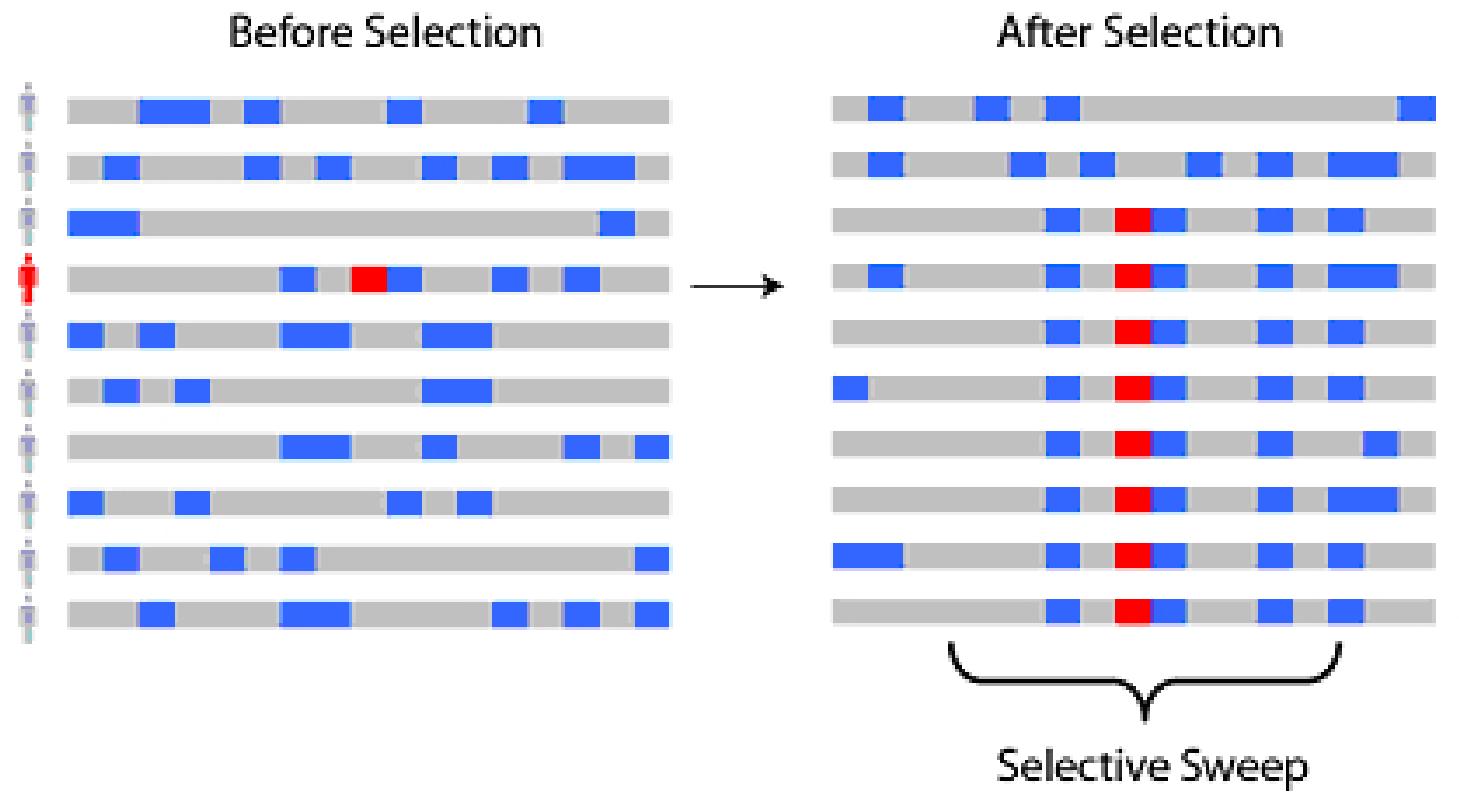


LACTOSE TOLERANCE



- until 11000 years ago we were not able to digest lactose
- livestock farming in Europe
- approximately 5 % people in northern Europe are unable to digest lactose, and 90 % people in eastern Asia
- ***LCT gen – most Europeans carrying allele that enables lactose digestion***
- **regulatory mutation** in region which controls expression of gene producing lactase- enables gene expression after infancy
- „selective sweep” – proof of selection on a region of the genome length more of 1 million bp
- one of the **strongest signals of selection in human genome**
- independent mutation in the gene in African population 3000 years ago- convergent evolution
- milk – vitamin D, less of pathogens than in water, nutritional alternative when crops are not giving high yield
- CULTURAL EVOLUTION LEADING TO GENOMIC EVOLUTION!!! – ECO-EVO

“selective sweep”



RESISTANCE TO PATHOGENS



Malaria

- Increase in population density – higher transmission of diseases
- Resistance to malaria- yearly 1 -2 million of children die in Africa – very strong selective pressure
- mutation Glu6Val (**sick cells anaemia**) in beta haemoglobin gen (*HBB*) found in Africa and linked to resistance to malaria and its incidence (Allison, 1954).
- other mutations in u HBB gen - α -thalassemia, lack of G6PD, ovalocytosis
- Duffy antigen gen (*FY*) – membrane protein that *Plasmodium vivax* use for entering the cells. **The incidence of mutation which disables its entrance is 100 % in sub-Saharan Africa, and it is absent in other human populations- highest difference in allelic frequency among human populations.**

Resistance to HIV

- Southern Africa – almost 50% women infected
- human leukocyte antigen - variant *HLA-B27* brings *higher resistance to HIV than HLA-A or HLA-C*
- MHC complex - the most variable part of the human genome
- low frequency of HIV in western Europe is linked to allelic variant of coreceptor for HIV CCR5, **found in 13% of Europeans** and protects very efficiently against HIV
- this variant was present in the population more than 1000 years ago

Genetic Adaptation to Cold Brought Migraines With It

Humans living in higher latitudes tend to have a variant of a gene involved in sensing cold temperatures, but it comes with a cost.

TRPM8 – gene responsible for detection of cold, but also linked to susceptibility of migraine

- one variant is frequent in northern Europe, very rare in equatorial Africa
- antagonistic pleiotropy!



Evolution May Make it Harder for Humans to Hold Their Liquor



1,000 Genomes Project (2500 ind.)

10 million years ago human ancestors developed gene for enzyme which enables alcohol biotransformation in ripe fruits – timing coincide with the terrestrial life of primates (lemures have 40x less efficient enzyme)

- alcohol dehydrogenase (ADH) gene cluster showing adaptation in western Africa and eastern Asia
- alcohol- acetaldehyde (toxic) – acetate – it is possible that faster transformation in acetaldehyde causes accumulation of toxic intermediate and lower tolerance to alcohol (nausea)
- **lower tolerance to alcohol – lower likelihood of alcoholism**



Human adaptations to diet, subsistence, and ecoregion are due to subtle shifts in allele frequency

Angela M. Hancock, David B. Witonsky, Edvard Ehler, Gorka Alkorta-Aranburu, Cynthia Beall, ...

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PNAS May 11, 2010 107 (Supplement 2) 8924-8930; first published May 5, 2010;

<https://doi.org/10.1073/pnas.0914625107>

- diabetes type 2 - gene *KCNQ1* – *higher frequency in populations eating lots of cereals – same gene has a role in hydrolysis of plant lipids*

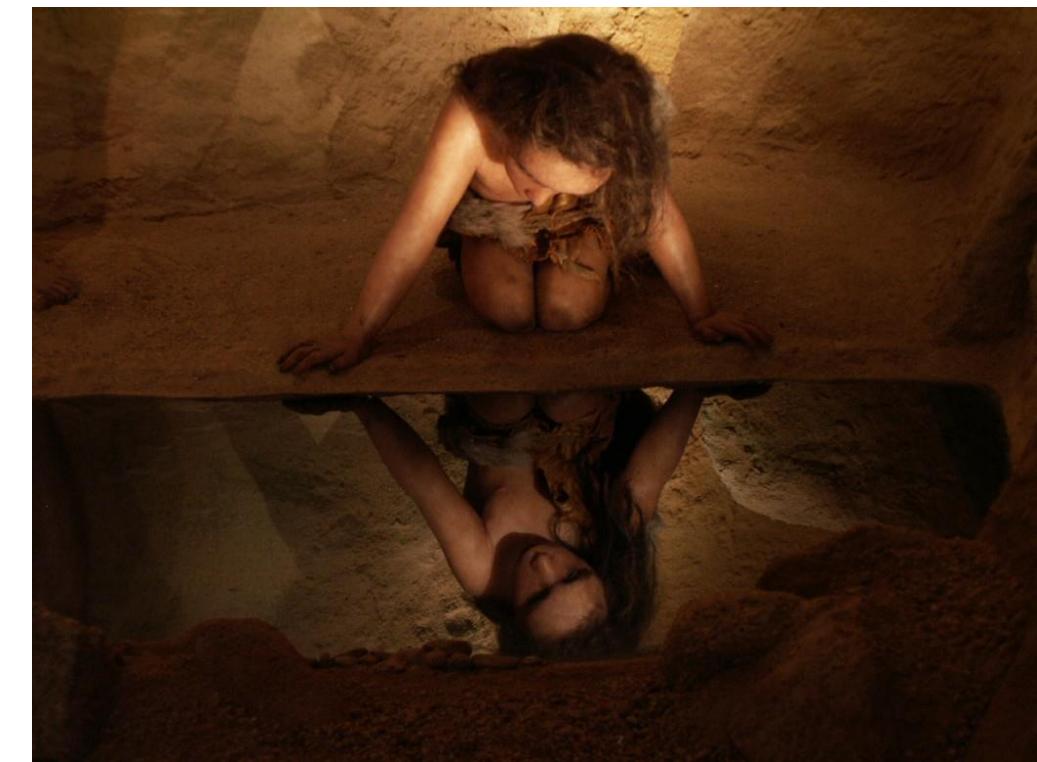


Vegetarian diet

- FADS2 – important gene for nutrition
- vegetarian diet in population in Pune (India) increased frequency of specific allele of FADS2 gene
 - mutation which enables better use of omega-3 and omega -6 fatty acids— important for brain development



- Neanderthals died out 40,000 years ago, but people of non-African origin carry approximately 2% of their genes
- in Malesia 2-4% genes od Denovinsian
- some alleles have a frequency up to 65% - related to skin and immunity- tickle hairs, adaptation to colder climate
- same genes are responsible for diabetes, lupus, Crohn's disease (lack of parasites)
- low fertility of crosses among human species



Assortative mating

- non random choice of partner based on similarity of traits
- positive or negative
- in humans almost exclusively positive

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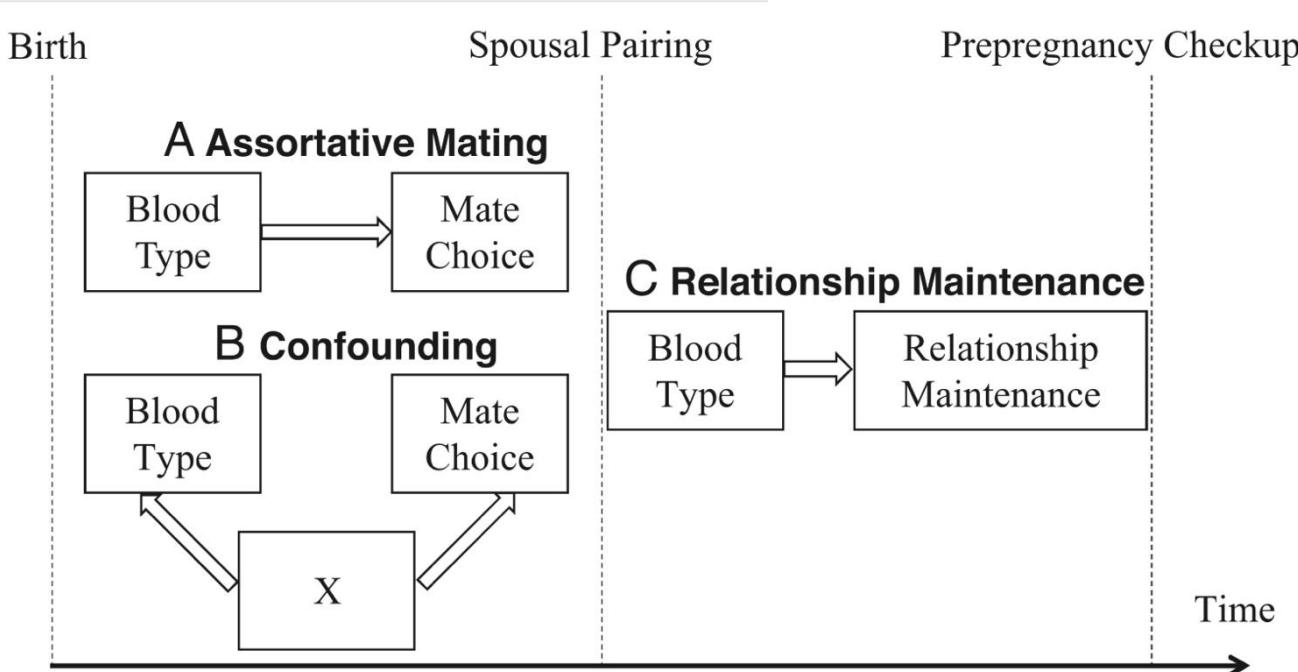


Assortative mating on blood type: Evidence from one million Chinese pregnancies

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Edited by Dalton Conley, Princeton University, Princeton, NJ; received June 9, 2022; accepted October 27, 2022

December 14, 2022 | 119 (51) e2209643119 | <https://doi.org/10.1073/pnas.2209643119>



Why do we pick similar mates, or do we? **FREE**

Special Collection: Biology Letters Reviews

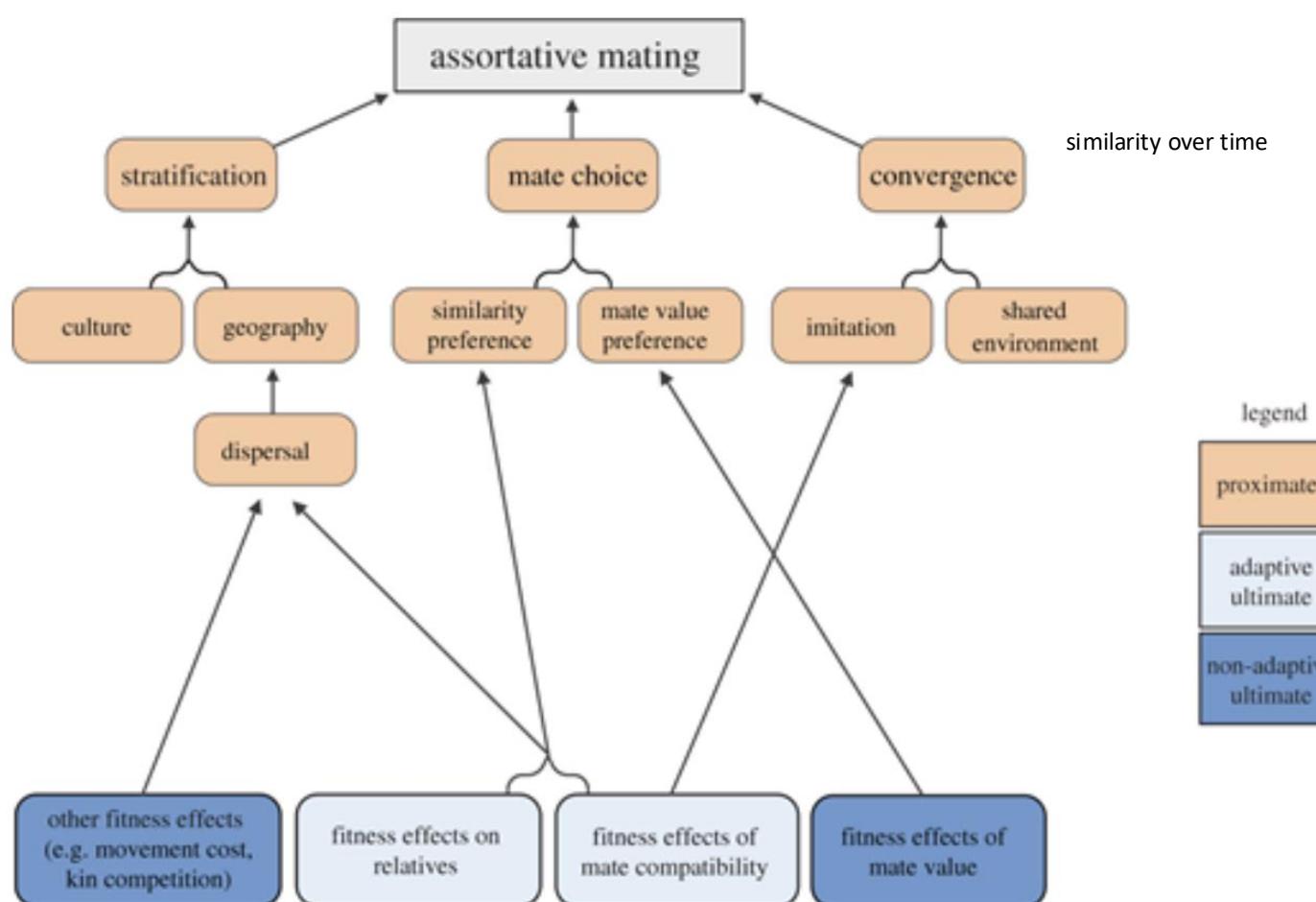
Tom M. M. Versluis ; Alex Mas-Sandoval ; Ewan O. Flintham ; Vincent Savolainen  

+ Author & article information

Biol Lett (2021) 17 (11): 20210463 .

<https://doi.org/10.1098/rsbl.2021.0463>

Article history 



Assortative mating

Published: 09 January 2017

Genetic evidence of assortative mating in humans

- Partner choice depends on visual, olfactory and auditory cues, personality, and interaction among those traits, as well interaction with age, ovulatory cycle, cognitive abilities, cultural and sociological influences

„Across three independent samples of 24,662 spousal pairs in total, we infer a correlation at trait-associated loci between partners for **height** (0.200, 0.004 standard error, SE)... finding evidence of a correlation at trait-associated loci for **waist-to-hip ratio** (0.101, 0.041 SE), **systolic blood pressure** (0.138, 0.064 SE) and **educational attainment** (0.654, 0.014 SE). Our results imply that **mate choice, combined with widespread pleiotropy** among traits, affects the genomic architecture of traits in humans”



Evidence of correlations between human partners based on systematic reviews and meta-analyses of 22 traits and UK Biobank analysis of 133 traits

[Tanya B. Horwitz](#) , [Jared V. Balbona](#), [Katie N. Paulich](#) & [Matthew C. Keller](#) 

[Nature Human Behaviour](#) 7, 1568–1583 (2023) | [Cite this article](#)

... we incorporated 480 partner correlations from 199 peer-reviewed studies of co-parents, engaged pairs, married pairs and/or cohabitating pairs that were published on or before 16 August 2022. We also calculated 133 trait correlations using up to 79,074 male–female couples in the UK Biobank (UKB). ... Across analyses, political and religious attitudes, educational attainment and some substance use traits showed the highest correlations, while psychological (that is, psychiatric/personality) and anthropometric traits generally yielded lower but positive correlations.

Consistency between individuals' past and current romantic partners' own reports of their personalities

Yoobin Park  and Geoff MacDonald [Authors Info & Affiliations](#)

Edited by Susan T. Fiske, Princeton University, Princeton, NJ, and approved May 13, 2019 (received for review February 19, 2019)

June 10, 2019 | 116 (26) 12793-12797 | <https://doi.org/10.1073/pnas.1902937116>



*Do people have a “type” when it comes to their romantic partners’ personalities? In the present research, we used data from a 9-y longitudinal study in Germany and examined the **similarity between an individual’s ex- and current partners** using the partners’ self-reported personality profiles. Based on the social accuracy model, our analyses distinguished similarity between partners that was attributable to similarity to an average person (normative similarity) and resemblance to the target participant himself/herself (self-partner similarity) to more precisely examine similarity from partner to partner (distinctive similarity). The results **revealed a significant degree of distinctive partner similarity, suggesting that there may indeed be a unique type of person each individual ends up with**. We also found that distinctive partner similarity was weaker for people high in extraversion or openness to experience, suggesting that these individuals may be less likely to be in a relationship with someone similar to their ex-partner (although the individual difference effects were not mirrored in an alternative analytic approach). These findings provide evidence for stability in distinctive partner personality and have important implications for predicting future partnering behaviors and actions in romantic relationships.*

Does natural selection favour taller stature among the tallest people on earth?

[Gert Stulp](#)^{1,2,✉}, [Louise Barrett](#)^{3,4}, [Felix C Tropf](#)², [Melinda Mills](#)⁵



- the height of Dutch man increased over 20 cm in 150 years, while Americans increased only 6 cm
- stronger sexual selection for tall men in Netherlands
- taller Dutch men had more children
- tall Dutch women were less likely to have children than middling-height Dutch women, the tall women who did have children had more children than their shorter counterparts.
- study accounted for diet, social inequality, and the availability and quality of healthcare

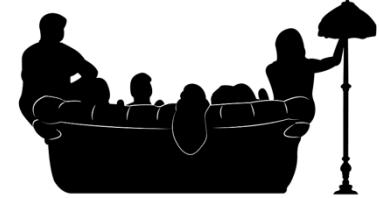
Friendship and natural selection

Nicholas A. Christakis and James H. Fowler  [Authors Info & Affiliations](#)

Edited by Francisco J. Ayala, University of California, Irvine, CA, and approved March 7, 2014 (received for review January 15, 2014)

July 14, 2014 | 111 (supplement_3) 10796-10801 | <https://doi.org/10.1073/pnas.1400825111>

F.R.I.E.N.D.S



Across the whole genome, friends' genotypes at the single nucleotide polymorphism level tend to be **positively correlated** (homophilic). In fact, the **increase in similarity relative to strangers is at the level of fourth cousins**. However, certain genotypes are also **negatively correlated** (heterophilic) in friends. And the degree of correlation in genotypes can be used to create a "friendship score" that predicts the existence of friendship ties in a hold-out sample. A focused gene-set analysis indicates that some of the overall correlation in genotypes can be explained by specific systems; for example, an **olfactory gene set is homophilic and an immune system gene set is heterophilic**, suggesting that these systems may play a role in the formation or maintenance of friendship ties. Friends may be a kind of "functional kin." Finally, homophilic genotypes exhibit significantly **higher measures of positive selection**, suggesting that, on average, they may yield a synergistic **fitness advantage** that has been helping to drive recent human evolution.

- homophilic genes - olfactory transduction pathway is significantly overrepresented
- genetic heterophily between for HLA haplotypes

The evolution of menopause in toothed whales

[Samuel Ellis](#) , [Daniel W. Franks](#), [Mia Lybkær Kronborg Nielsen](#), [Michael N. Weiss](#) & [Darren P. Croft](#)

[Nature](#) 627, 579–585 (2024) | [Cite this article](#)

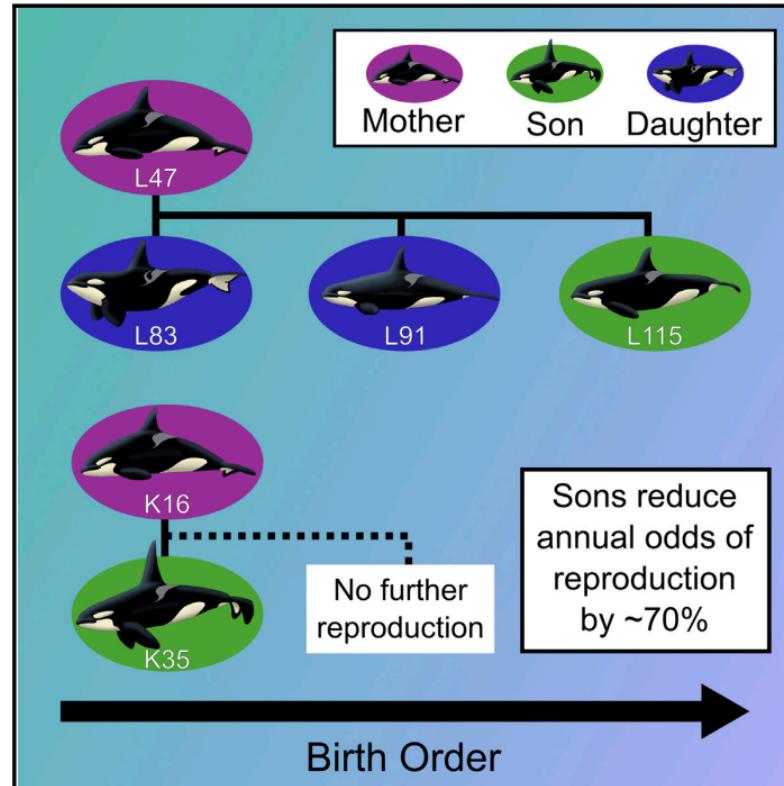
58k Accesses | 36 Citations | 1302 Altmetric | [Metrics](#)

- the live-long hypothesis vs the stop-early hypothesis
- no evidence for the male-driven menopause hypothesis
- investment in male offspring (males larger)
- species with menopause are reproducing more slowly
- grandmother and mother hypotheses

Although progress has been made in understanding the adaptive value of menopause in humans^{3,4}, the generality of these findings remains unclear. To toothed whales are the only mammal taxon in which menopause has **evolved several times**⁵, providing a unique opportunity to test the theories of how and why menopause evolves in a comparative context. Here, we assemble and analyse a comparative database to test competing evolutionary hypotheses. We find that menopause evolved in toothed whales by females **extending their lifespan without increasing their reproductive lifespan, as predicted by the 'live-long' hypotheses**. We further show that menopause results in females increasing their opportunity for **intergenerational help** by increasing their lifespan overlap with their grandoffspring and offspring without increasing their reproductive overlap with their daughters. Our results provide an informative comparison for the evolution of human life history and demonstrate that the same pathway that led to menopause in humans can also explain the evolution of menopause in toothed whales.

Costly lifetime maternal investment in killer whales

Graphical abstract



Authors

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In brief

Killer whale mothers are known to provide survival benefits to their adult offspring, especially their sons. Weiss et al. show that providing these benefits comes at a significant reproductive cost to mothers. These costs imply lifetime parental investment in killer whales, an extreme and unique life history strategy.

Highlights

- Providing care to weaned sons reduces female killer whales' reproductive output
- These effects cannot be explained by lactation costs or group composition
- Sons do not become less costly as they grow older

Volume 33, Issue 5, 13 March 2023, Pages R194-R197

Dispatch

Animal behavior: Killer whale mamas' boys

Janet Mann

<https://www.youtube.com/watch?v=S1gg5-sNRAo>

