

Contemporary and adaptive human evolution

(disregard genes names 😊)

- 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**”*

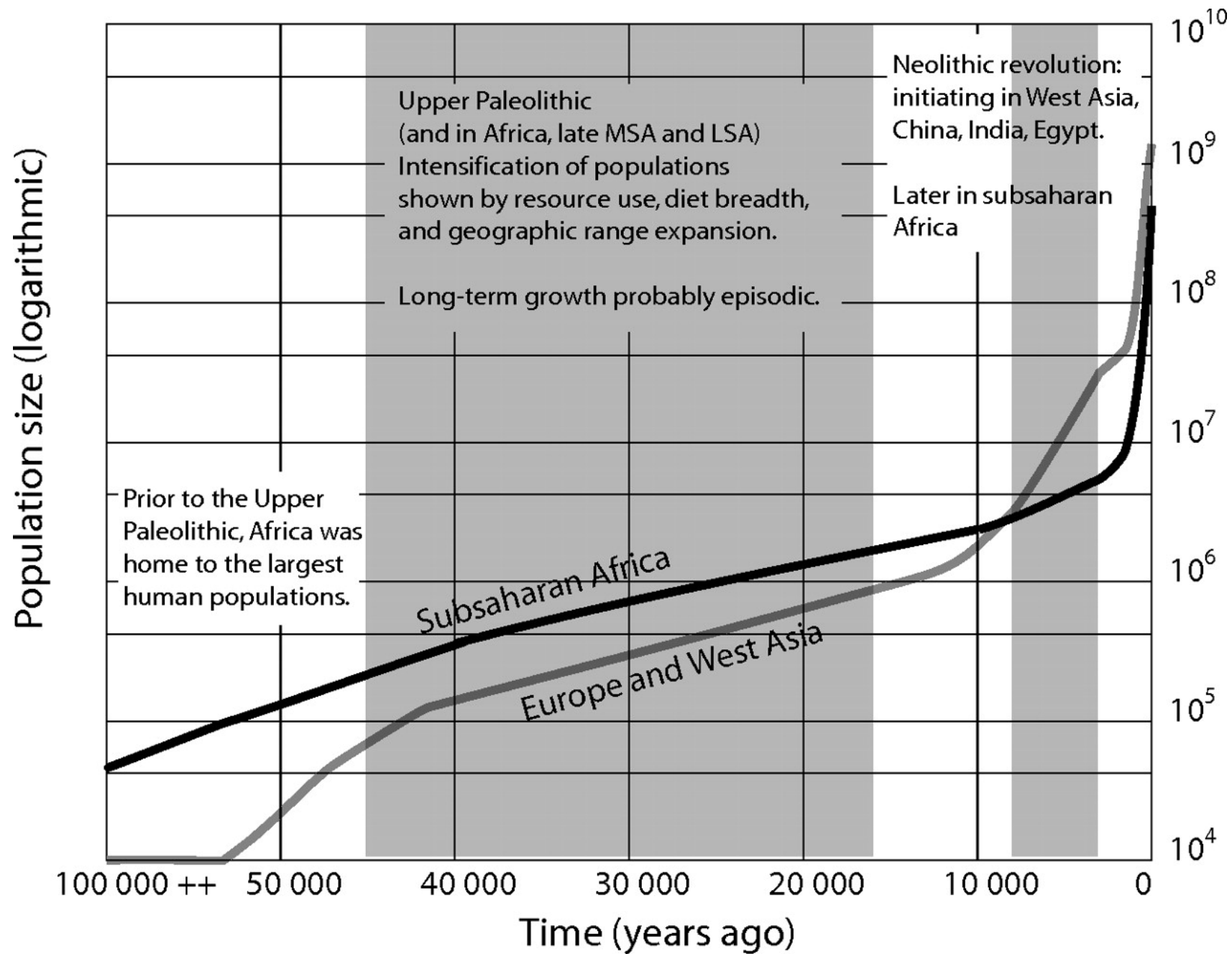
Recent acceleration of human adaptive evolution

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>

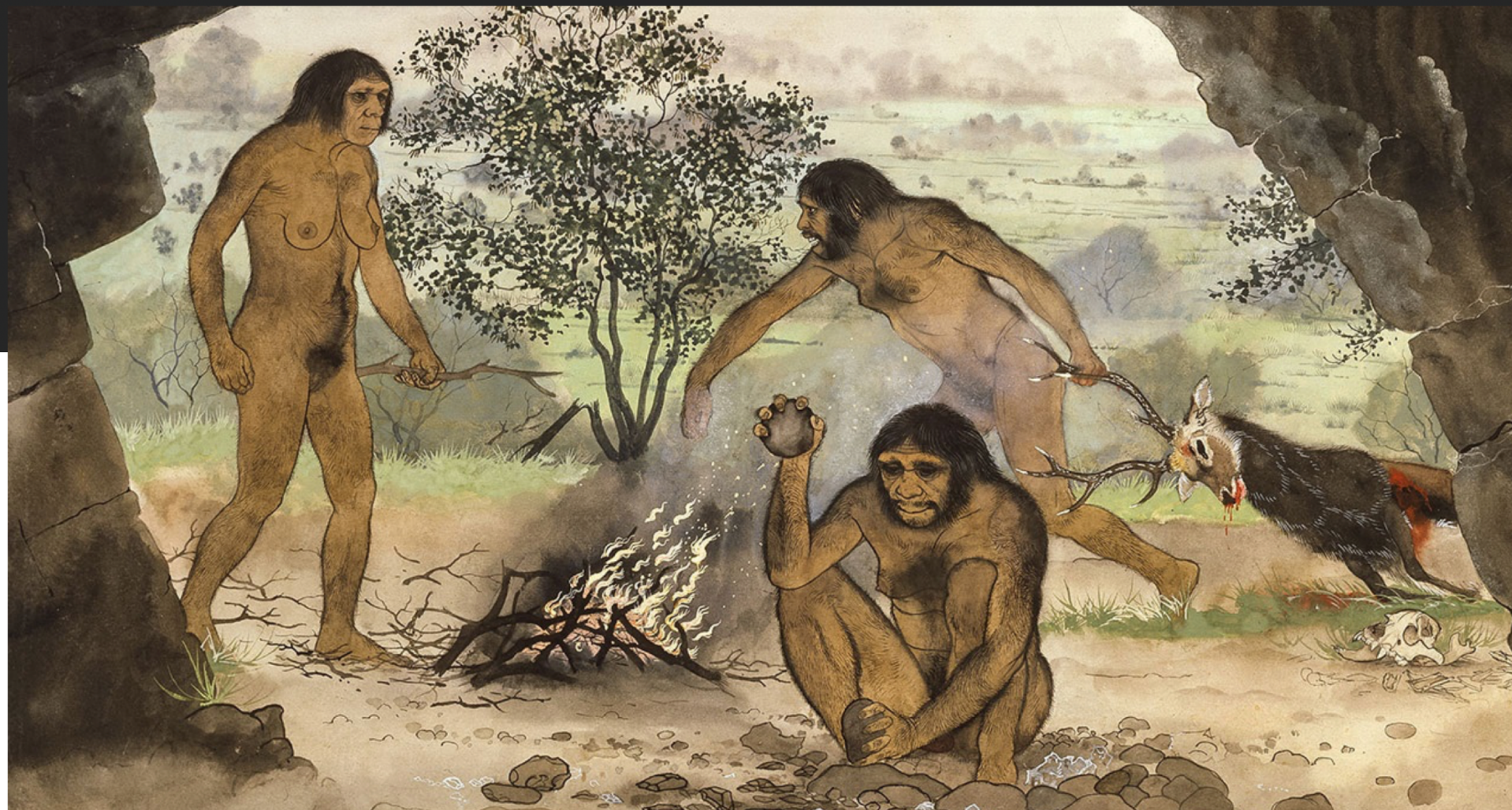
*„Larger populations generate more new selected mutations, and we show the consistency of the observed data with the historical pattern of **human population growth**. **The rate of adaptive evolution** in human populations has indeed accelerated within the past 80,000 years. **Demographic growth** intensified 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

🔒 | REPORT



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

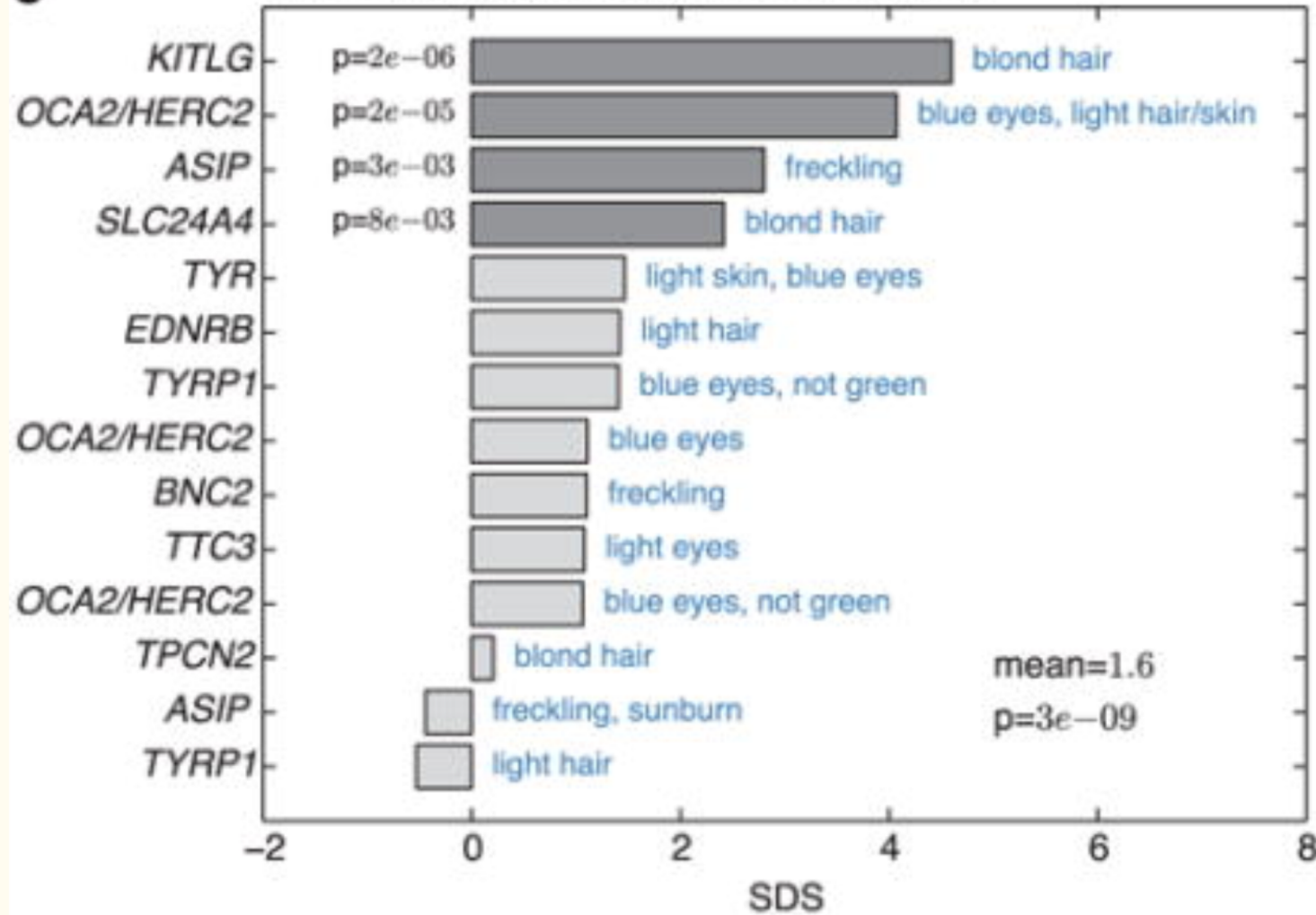
+2 authors

[Authors Info & Affiliations](#)

*„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.”*

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- its not related to blue eyes
- northern Europe- 11000 years ago- 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

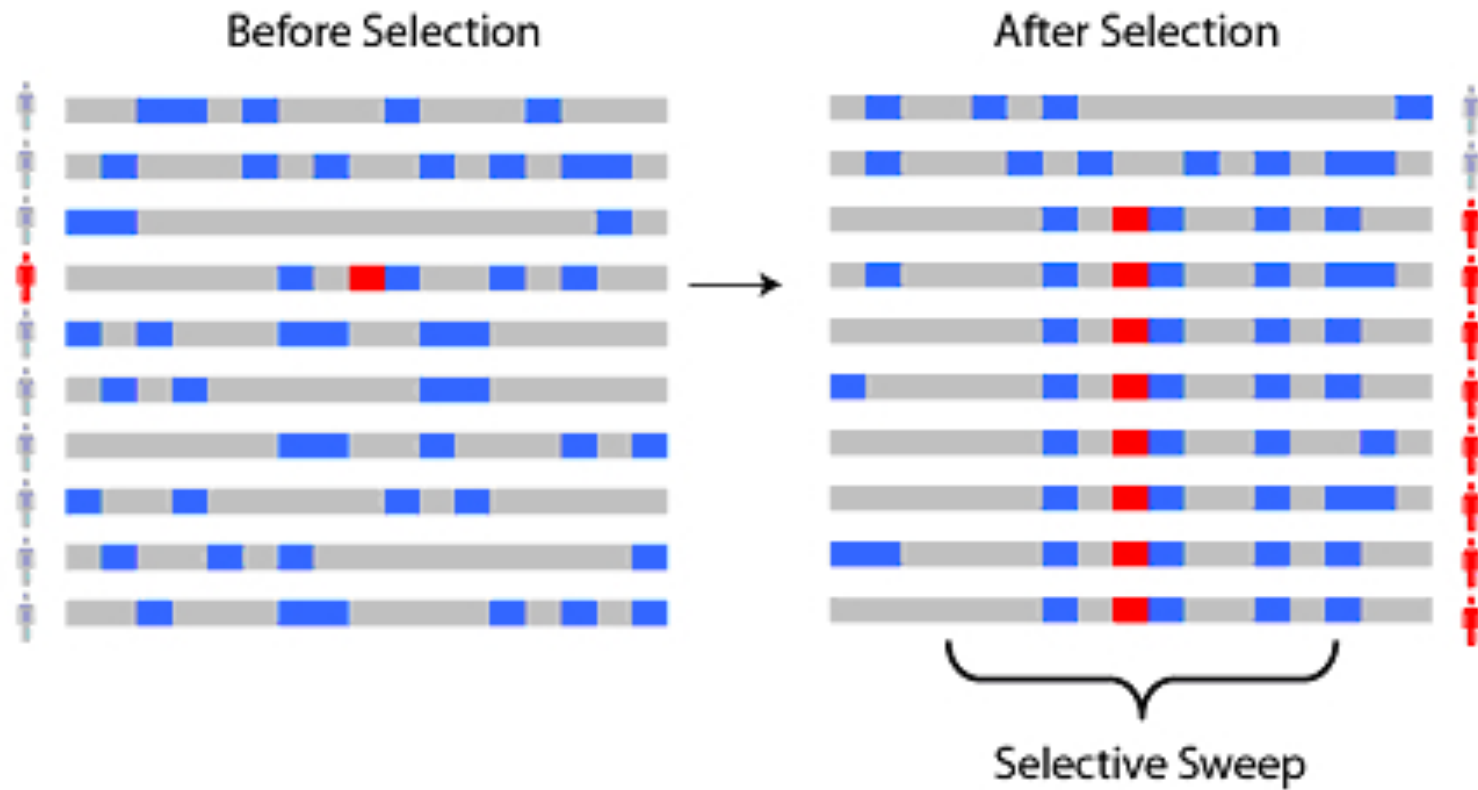


LACTOSE TOLERANCE



- 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 the *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 its 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 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 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



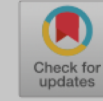
Evolution May Make it Harder for Humans to Hold Their Liquor



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 – its possible that faster transformation in acetaldehyde causes accumulation of toxic intermediate and lower tolerance to alcohol
- 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, ...

[+ See all authors and affiliations](#)

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*



- 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 of Denisovan
- some alleles have a frequency up to 65% - related to skin and immunity- thicker hairs, adaptation to colder climate
- diabetes, lupus, Crohn's disease (lack of parasites)
- low fertility of crosses



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”*




Human height – evolving and being selected for!!

- during the last 150 years Dutch men growth in height extra 20 centimetres, while Americans increase in average for 6 cm
- study showed that differences in diet and healthcare don't account for the difference between populations, but its the effect of selection!
- tall Dutch men have more children
- middle height women were more likely to have kids, but when tall women had kids, they had higher number of them

Research articles

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

Gert Stulp , Louise Barrett, Felix C. Troup and Melinda Mills

Published: 07 May 2015 | <https://doi.org/10.1098/rspb.2015.0211>

Gene-by-environment effects

- 60,000 people of European ancestry genotyped by Kaiser Permanente in Northern California, and 150,000 people from UK Biobank
- allelic frequencies across different ages
- UK - parental generations (1950s...)- **correlation between early death in men and the frequency of a nicotine receptor allele (CHRNA3 gene) that makes it harder to quit smoking in their children – the frequency of allele changing across age (being lower in older man)**
- The allele's frequency in UK women and in people from Northern California did not vary with age- fewer in these groups smoked heavily and the allele did not affect their survival.
- As smoking habits have changed, the selective pressure against allele has ceased, and its frequency is unchanged across age in recent generations



PLOS BIOLOGY

OPEN ACCESS PEER-REVIEWED

RESEARCH ARTICLE

Identifying genetic variants that affect viability in large cohorts

Hakhamanesh Mostafavi , Tomaz Berisa, Felix R. Day, John R. B. Perry, Molly Przeworski , Joseph K. Pickrell 

Published: September 5, 2017 • <https://doi.org/10.1371/journal.pbio.2002458>

[See the preprint](#)