HUMAN PALAEOGENOMICS

EVA CHOCHOLOVÁ
LABORATORY OF BIOLOGICAL AND MOLECULAR ANTHROPOLOGY
DEPARTMENT OF EXPERIMENTAL BIOLOGY

Genomic Evolution and Adaptation in Africa

by Sarah A. Tishkoff

(University of Pennsylvania, Philadelphia, USA)

21 March 2024 17:00

Mendel Lectures take place in Mendel´s refectory in the Mendel Museum Brno

Biodiversity, Biomes, and Beliefs – the Impacts of Conservation and Global Change on BaAka Hunter-gatherers in the Central African Republic

28 March 2024
4:00 PM

University Campus Bohunice (pavilion B11/ seminar room 132)

Lecture will be held in English

Speaker

Carolyn A. Jost Robinson

QUIZ

1. Why is HTS better for aDNA, considering fragment length?

Most fragments ~ 30-70 bp, cannot be targeted by PCR based methods.

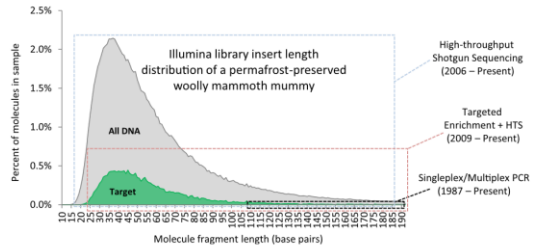


Figure 2. Different sequencing strategies are capable of characterizing different fractions of ancient DNA samples in terms of endogenous DNA content and fragment length.

2. What is the endogenous aDNA proportion in extracted samples?

Varies greatly!! Authors usually describe about 5 %.

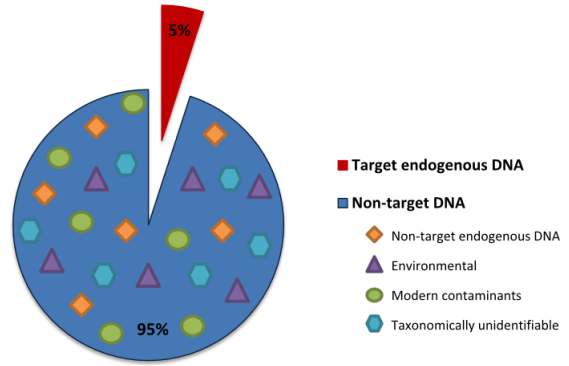


Figure 1. Non-target DNA (approximately 95%) comprises the majority of surviving DNA in ancient samples, whereas the desired or targeted endogenous DNA is only a fraction (approximately 0–5%) of the overall constituents.

3. What is important when choosing a marker for metabarcoding?

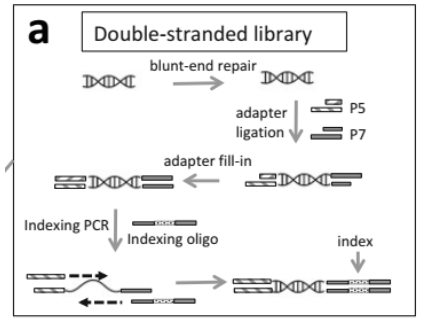
A perfect metagenomics barcode/marker should... + length!

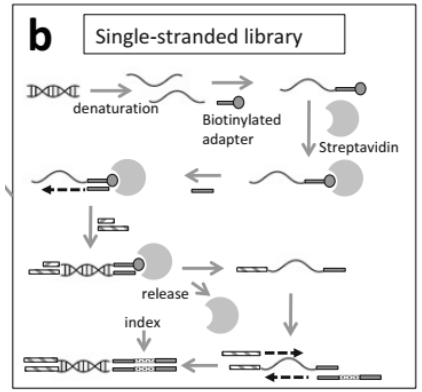
- be **present** in all the organisms, in all the cells
- have **variable** sequence among different species
- be **conserved** among individuals of the same species
- be **easy to amplify** and not too long for sequencing

Key: Color	Clade	Primary barcode(s)	Secondary barcode(s)
	Animals	CO1	CO1, 16S
	Fungi	ITS	LSU D1/D2
	Green algae	*tufA*	LSU D2/D3
	Land plants	*rbcL*/matK	*psbA-trnH*/ITS
	Algae	CO1-5P	LSU D2/D3

Bacteria/ Archae 16S RIF

4. What is the difference between single- and double-stranded library prep?

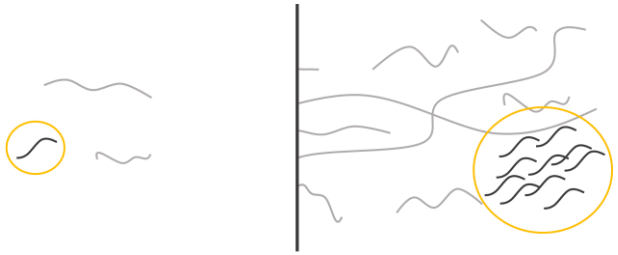




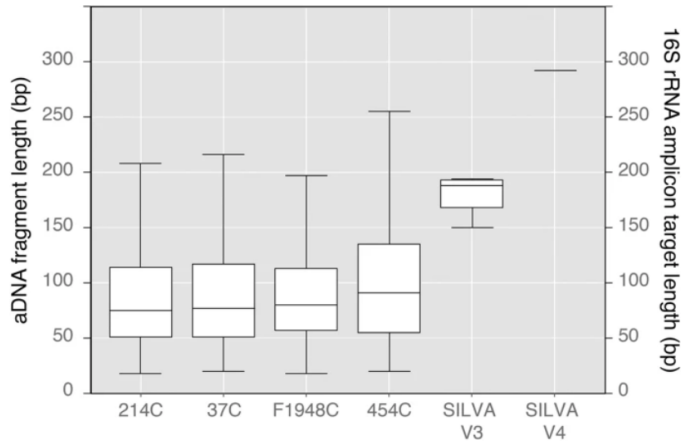
QUIZ

5. What is enrichment and when to use it?

	Targeted SNP capture	Whole-genome capture	Whole-genome shotgun
Data characteristics			
Genomic coverage	Targeted SNPs and alleles	Genome-wide	Genome-wide
Typical enrichment range	45–13,000 (6)	2–13× (79)	None
Best use scenario	Low endogenous DNA; low/medium complexity	Low endogenous DNA; high complexity	Medium/high endogenous DNA; high complexity



6. What is the PCR amplification bias and why does it matter? Is there a way to prevent it?



Length distribution box plots of aDNA extracted from archaeological dental calculus and calculated V3 and V4 16S rRNA amplicon lengths for microbes in the SILVA SSU 111 database.

HUMAN PALAEOGENOMICS

Group 1
Reasons for including scientists from studied populations/regions

Group 2
Reasons against including scientists from studied populations/regions (or challenges)

ARCHAEOLOGICAL SOURCES as discussed in Gokhman et al., 2017

Artefacts

Preservation

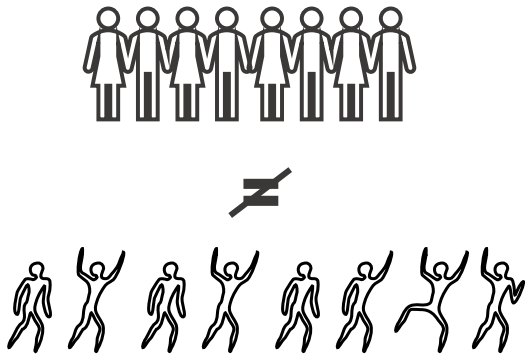
Interpretation

Taphonomic biases: biological and geological processes that occur between death, decay, and eventual fossilisation that result in over- or under-representation or even complete absence of specific skeletal elements, taxa, or demographic groups (e.g., different sexes or ages) in the fossil record.

Mitchell and Rawlence., 2021; DOI: 10.1016/j.tree.2020.10.005

Current populations

Differences



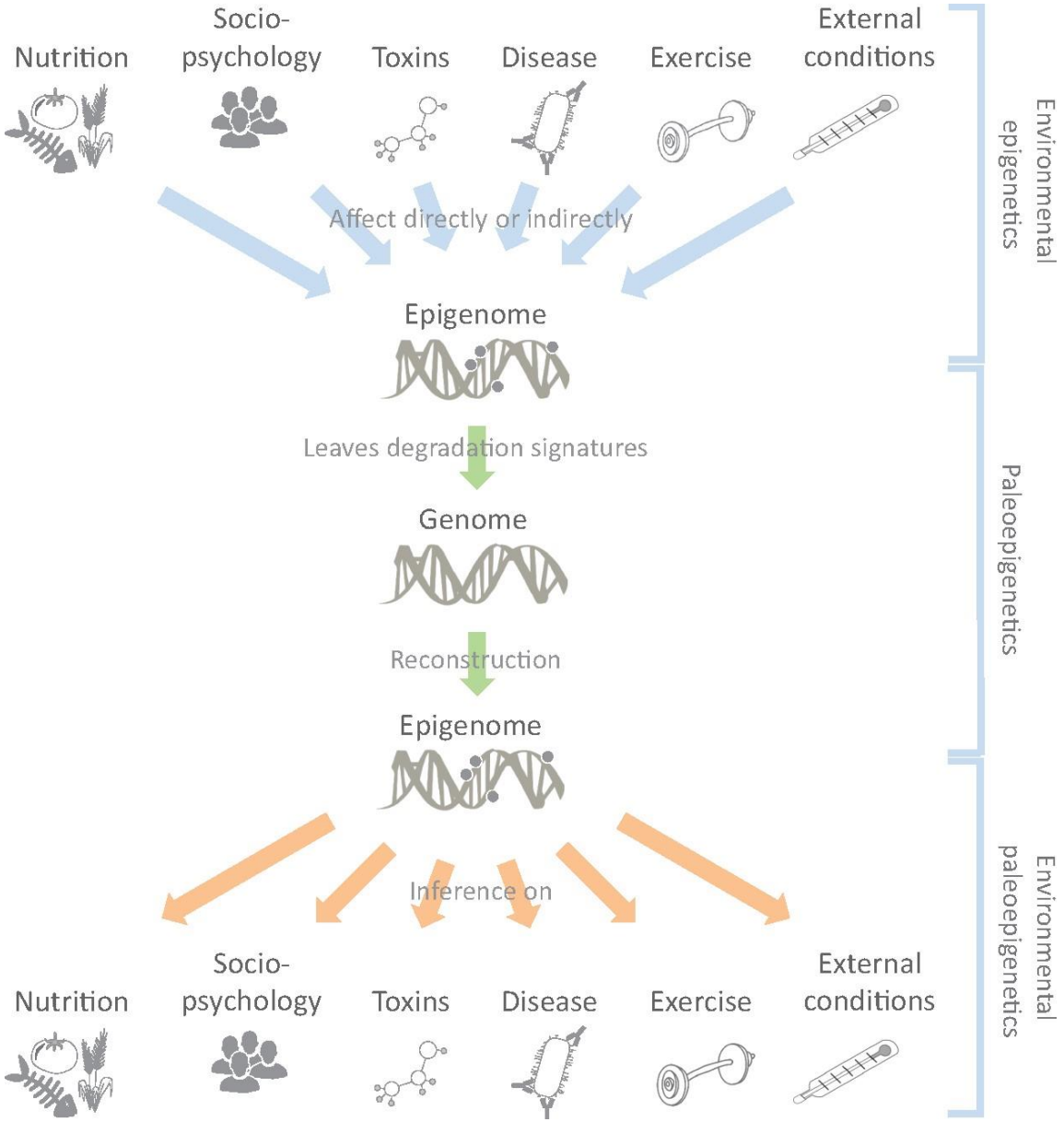
Genomics

Change over long time

Epigenomics

Environmentally responsive, plastic

+ many more that were not discussed in detail, such as anthropology, proteomics, isotope analysis, palaeozoology, palaeobotany…

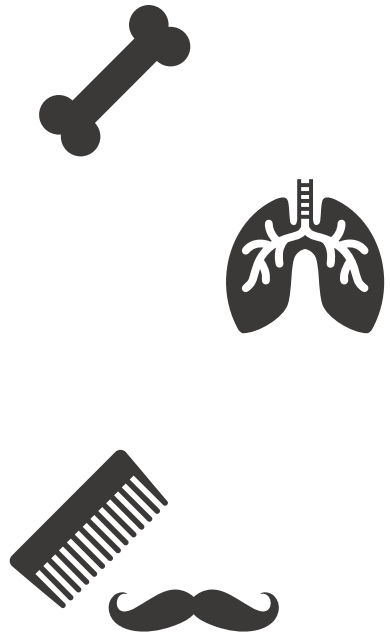


In a study by Dominguez-Salas etal., researchers focused on a rural Gambian population where caloric intake varies considerably according to season, with a "hungry season" followed by a "harvest season." The "hungry season" is the rainy season, characterized by restricted protein-energy availability, whereas the dry "harvest season" does not hold any nutritional stress. The oscillations in nutrient availability have been shown to affect *in utero* development and growth (Rayco-Solon etal. 2005). These studies found that children conceived during the "hungry season" showed hypermethylation at six metastable epialleles, residing near or within the following genes: *LOC654433*, *EXD3*, *RBM46*, *BOLA3*, *ZNF678*, and *ZFYVE28* (Waterland etal. 2010; Dominguez-Salas etal. 2014). In follow-up work on these data, we crossed these six ERLs with a list of DMRs found between archaic humans (Neanderthal and Denisovan) and 21 present-day humans (Gokhman etal. 2014).

Gokhman et al., 2017; DOI: 10.1093/molbev/msx211

ANCIENT HUMAN GENOMES

Human tissues

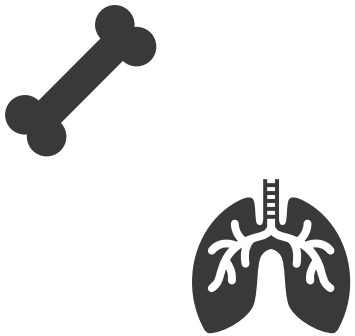


Artefacts

Settlements and environment

ANCIENT HUMAN GENOMES

Human tissues



Article | Open access | Published: 20 August 2019

Ancient DNA from the skeletons of Roopkund Lake reveals Mediterranean migrants in India

Éadaoin Harney, Ayushi Nayak, Nick Patterson, Pramod Joglekar, Veena Mushrif-Tripathy, Swapan Mallick, Nadin Rohland, Jakob Sedig, Nicole Adamski, Rebecca Bernardos, Nasreen Broomandkhoshbacht, Brendan J. Culleton, Matthew Ferry, Thomas K. Harper, Megan Michel, Jonas Oppenheimer, Kristin Stewardson, Zhao Zhang, Harashawaradhana, Maanwendra Singh Bartwal, Sachin Kumar, Subhash Chandra Diyundi, Patrick Roberts, Nicole Boivin, Douglas J. Kennett, Kumarasamy Thangaraj, David Reich ✉ & Niraj Rai

— Show fewer authors

Nature Communications **10**, Article number: 3670 (2019) | Cite this article

Article | Open access | Published: 03 December 2022

4000-year-old hair from the Middle Nile highlights unusual ancient DNA degradation pattern and a potential source of early eastern Africa pastoralists

Ke Wang ✉, Madeleine Bleasdale, Charles Le Moyne, Cacilia Freund, Johannes Krause, Nicole Boivin & Stephan Schiffels ✉

Scientific Reports **12**, Article number: 20939 (2022) | Cite this article

ANCIENT HUMAN GENOMES

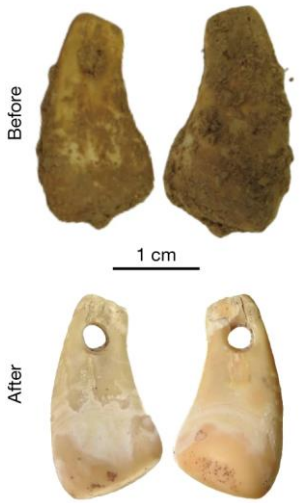
Artefacts

Article | Open access | Published: 03 May 2023

Ancient human DNA recovered from a Palaeolithic pendant

Elena Essel, Elena I. Zavala, Ellen Schulz-Kornas, Maxim B. Kozlikin, Helen Fewlass, Benjamin Vernot, Michael V. Shunkov, Anatoly P. Derevianko, Katerina Douka, Ian Barnes, Marie-Cécile Soulier, Anna Schmidt, Merlin Szymanski, Tsenka Tsanova, Nikolay Sirakov, Elena Endarova, Shannon P. McPherron, Jean-Jacques Hublin, Janet Kelso, Svante Pääbo, Mateja Hajdinjak, Marie Soressi & Matthias Meyer

Nature **618**, 328–332 (2023) | Cite this article



Article | Open access | Published: 17 December 2019

A 5700 year-old human genome and oral microbiome from chewed birch pitch

Theis Z. T. Jensen, Jonas Niemann, Katrine Højholt Iversen, Anna K. Fotakis, Shyam Gopalakrishnan, Åshild J. Vågene, Mikkel Winther Pedersen, Mikkel-Holger S. Sinding, Martin R. Ellegaard, Morten E. Allentoft, Liam T. Lanigan, Alberto J. Taurozzi, Sofie Holtsmark Nielsen, Michael W. Dee, Martin N. Mortensen, Mads C. Christensen, Søren A. Sørensen, Matthew J. Collins, M. Thomas P. Gilbert, Martin Sikora, Simon Rasmussen & Hannes Schroeder

Nature Communications **10**, Article number: 5520 (2019) | Cite this article

ANCIENT HUMAN GENOMES

Settlements and environment

Neandertal and Denisovan DNA from Pleistocene sedi-ments

VIVIANE SLON, CHARLOTTE HOPFE, CLEMENS L. WEISS, FABRIZIO MAFESSONI, MARCO DE LA RASILLA, CARLES LALUEZA-FOX, ANTONIO ROSAS, MARIE SORESSI, MONIKA V. KNUL, REBECCA MILLER, JOHN R. STEWART, ANATOLY P. DEREVIANKO, ZENOBIA JACOBS, BO LI, RICHARD G. ROBERTS, MICHAEL V. SHUNKOV, HENRY DE LUMLEY, CHRISTIAN PERRENOUD, IVAN GUŠIĆ, ŽELJKO KUĆAN, PAVAO RUDAN, AYINUER AXIMU-PETRI, ELENA ESSEL, SARAH NAGEL, BIRGIT NICKEL, ANNA SCHMIDT, KAY PRÜFER, JANET KELSO, HERNÁN A. BURBANO, SVANTE PÄÄBO, AND MATTHIAS MEYER fewer

Authors Info & Affiliations

SCIENCE · 27 Apr 2017 · Vol 356, Issue 6338 · pp. 605-608 · DOI: 10.1126/science.aam9695

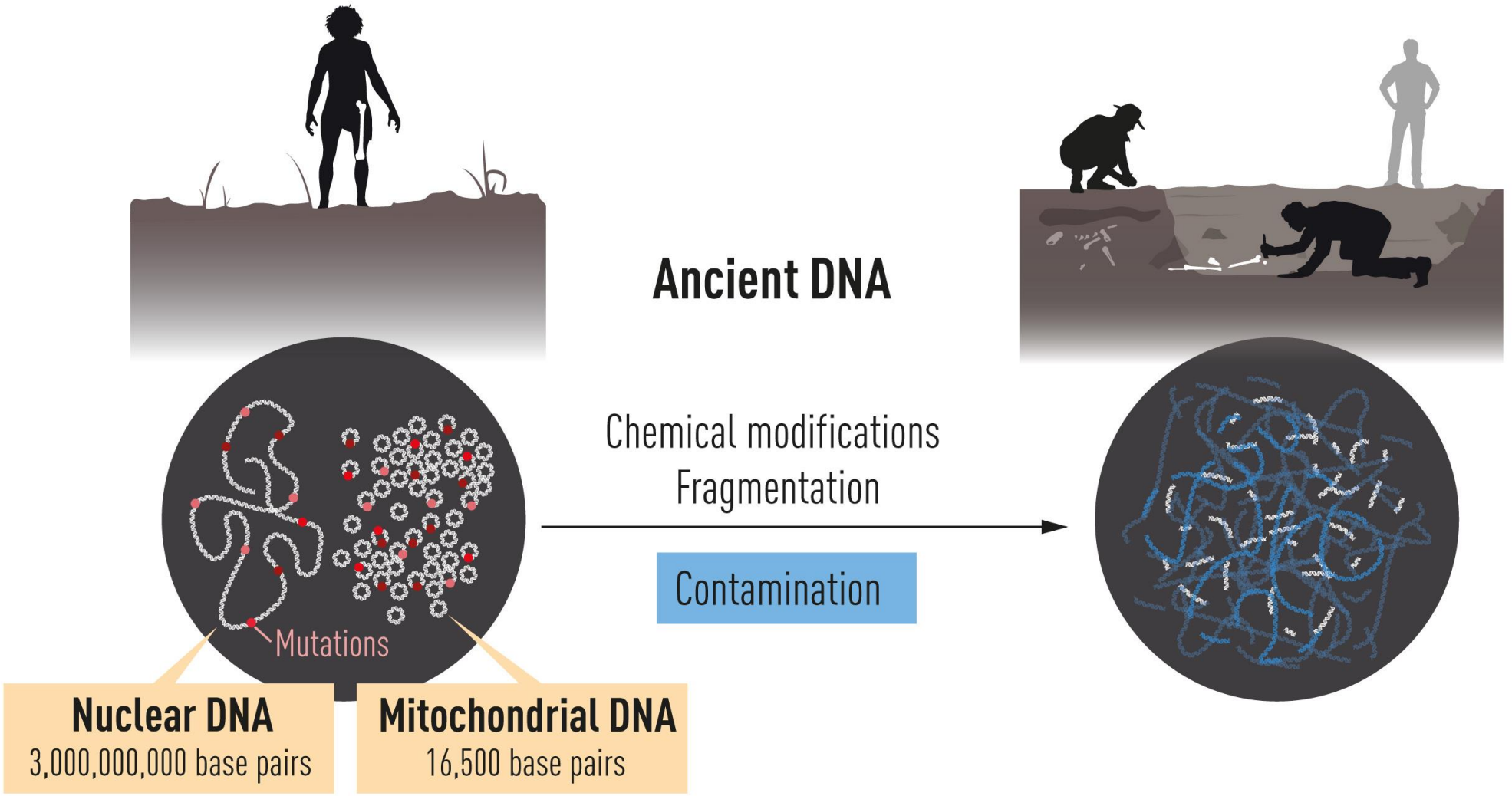
Article | Open access | Published: 23 June 2021

Pleistocene sediment DNA reveals hominin and faunal turnovers at Denisova Cave

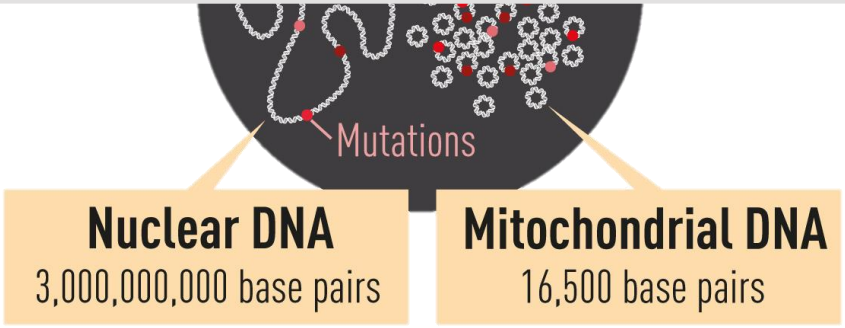
Elena I. Zavala, Zenobia Jacobs, Benjamin Vernot, Michael V. Shunkov, Maxim B. Kozlikin, Anatoly P. Derevianko, Elena Essel, Cesare de Fillipo, Sarah Nagel, Julia Richter, Frédéric Romagné, Anna Schmidt, Bo Li, Kieran O'Gorman, Viviane Slon, Janet Kelso, Svante Pääbo, Richard G. Roberts & Matthias Meyer

Nature **595**, 399–403 (2021) | Cite this article

ANCIENT HUMAN GENOMES

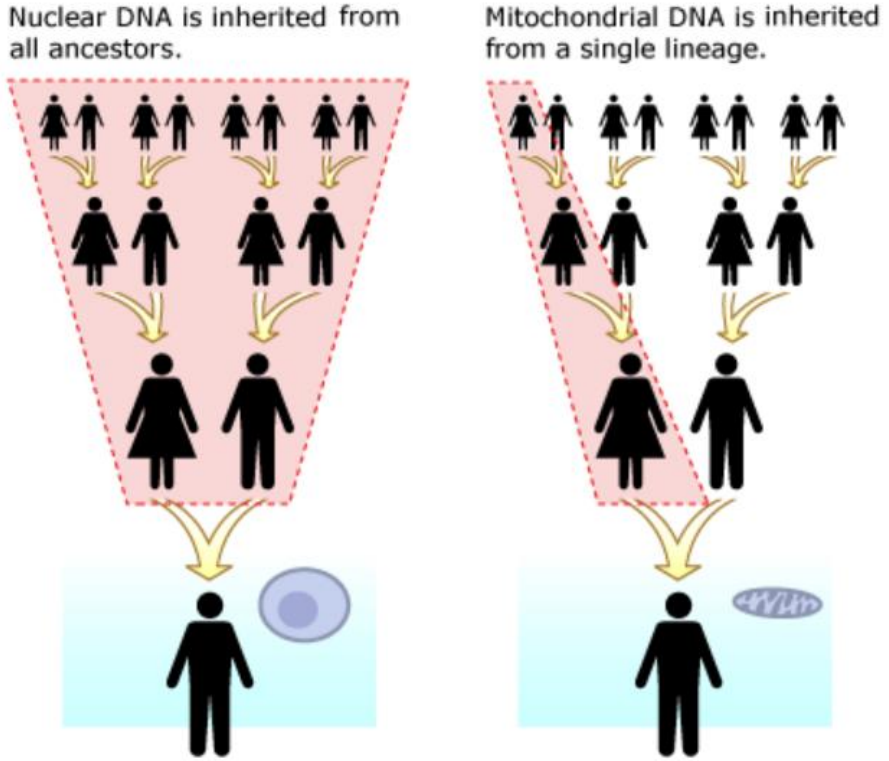


ANCIENT HUMAN GENOMES



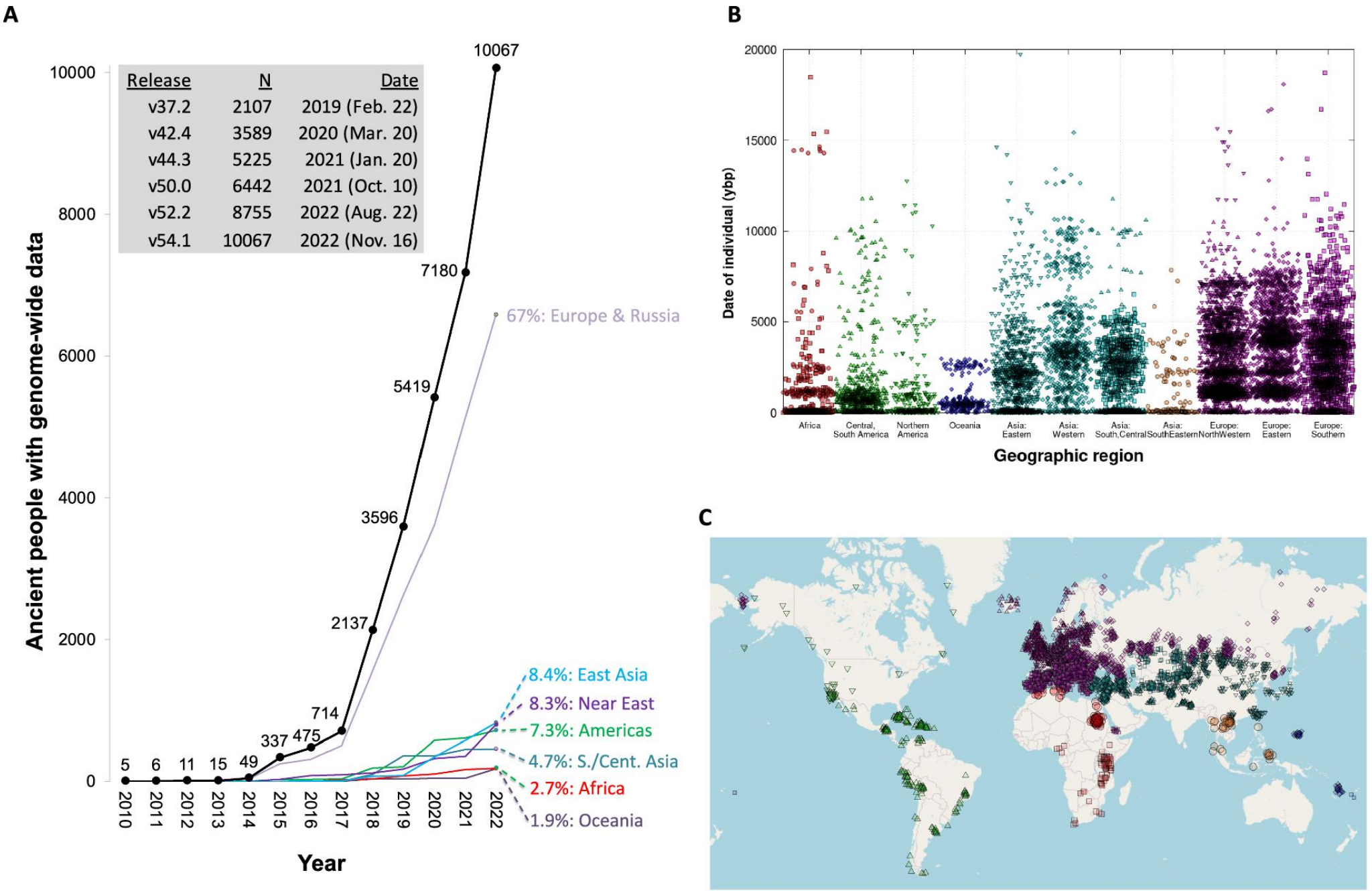
- Two copies
- Long
- Recombining
- Inherited from both parents
- Great amount of information

- More copies per cell
- Shorter, circular
- Generally without recombination
- Maternal
- Limited insight



https://www.nobelprize.org/prizes/medicine/2022/summary/

https://evolution.berkeley.edu/evo-news/making-sense-of-ancient-hominin-dna/



Growth in world's published human genome-wide ancient DNA data. (**A**) By year of publication (broken down by geography). (**B**) By date (color and symbol both indicate geographic location). (**C**) By geography (using same color and symbol scheme as in previous panel).

Mallick et al., 2024; DOI: 10.1038/s41597-024-03031-7

HUMAN EVOLUTION

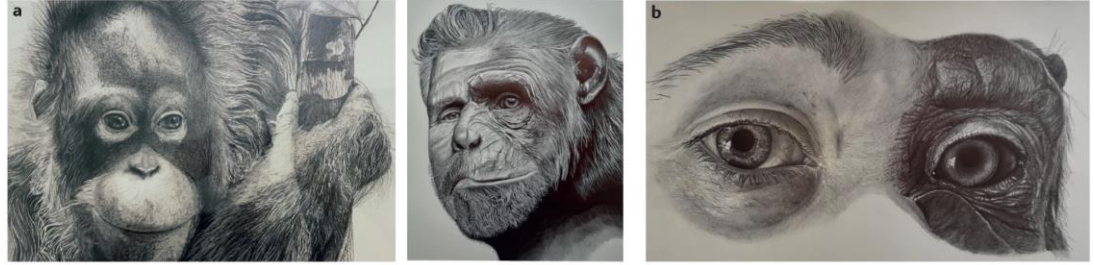
Letter | Published: 14 March 2016

Nuclear DNA sequences from the Middle Pleistocene Sima de los Huesos hominins

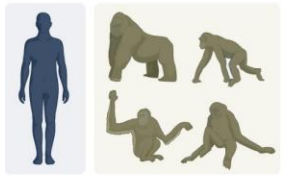
Matthias Meyer, Juan-Luis Arsuaga, Cesare de Filippo, Sarah Nagel, Ayinuer Aximu-Petri, Birgit Nickel, Ignacio Martínez, Ana Gracia, José María Bermúdez de Castro, Eudald Carbonell, Bence Viola, Janet Kelso, Kay Prüfer & Svante Pääbo

Nature **531**, 504–507 (2016) | Cite this article

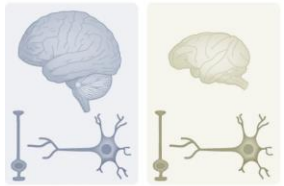
430,000 years ago



c Human traits

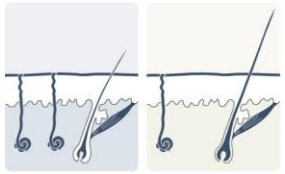


Big brain
Cortex expansion, neuron numbers and synapse density enhance cognition



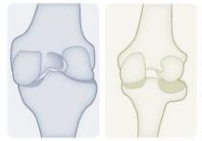
Differences in neurogenesis and neuron maturation

Naked ape
Decreased hair thickness and increased eccrine gland density impacts thermoregulation

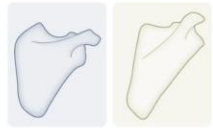


White eyes
Pigmentation patterns of the visible part of the eyeball, around the iris and sclera, has links to sexual selection and potentially social behaviour

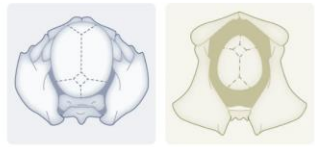
Upright walking and projectile throwing



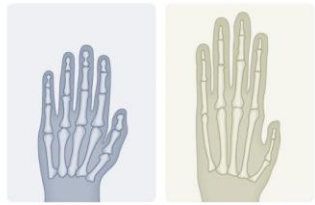
Knee and foot adaptations to biomedical demands of upright walking and distance running



Shoulder modifications increase rotational velocity

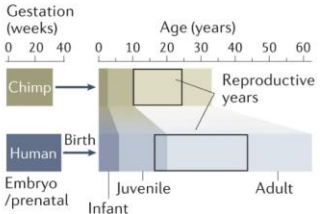


Pelvis modifications support upright walking and cranium size during childbirth

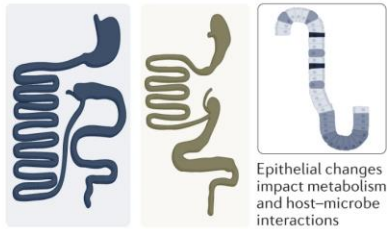


Relatively long thumb and short fingers enhances object grasping

Ontogeny
Morphological, metabolic, immune and cognitive differences emerge over an extended developmental period

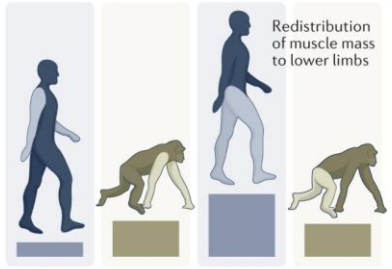


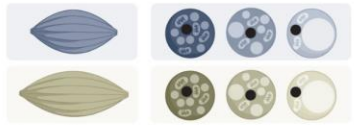
Diet and cooking
Increased small intestine to colon ratio impacts nutrient digestion/absorption



Epithelial changes impact metabolism and host–microbe interactions

Body composition
Changes in muscle and adipose tissue composition and distribution impacts body physiology





Muscle fibre type proportions

Brown, beige, white adipocyte type and location proportion

Complex language
Muscular/skeletal/innervation modifications to the tongue and vocal cords and elaborated brain circuits enable complex speech and language

Pollen et al., 2023; DOI: 10.1038/s41576-022-00568-4

HUMAN EVOLUTION

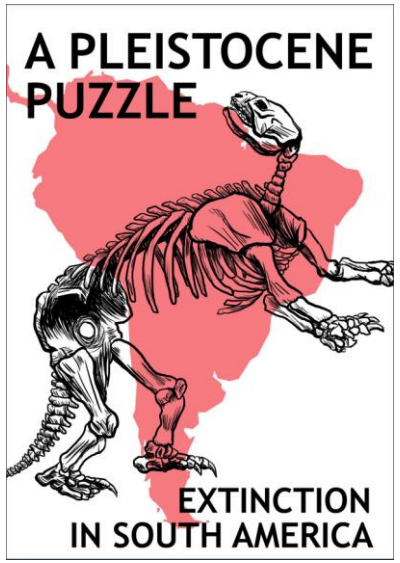
Understanding Evolution

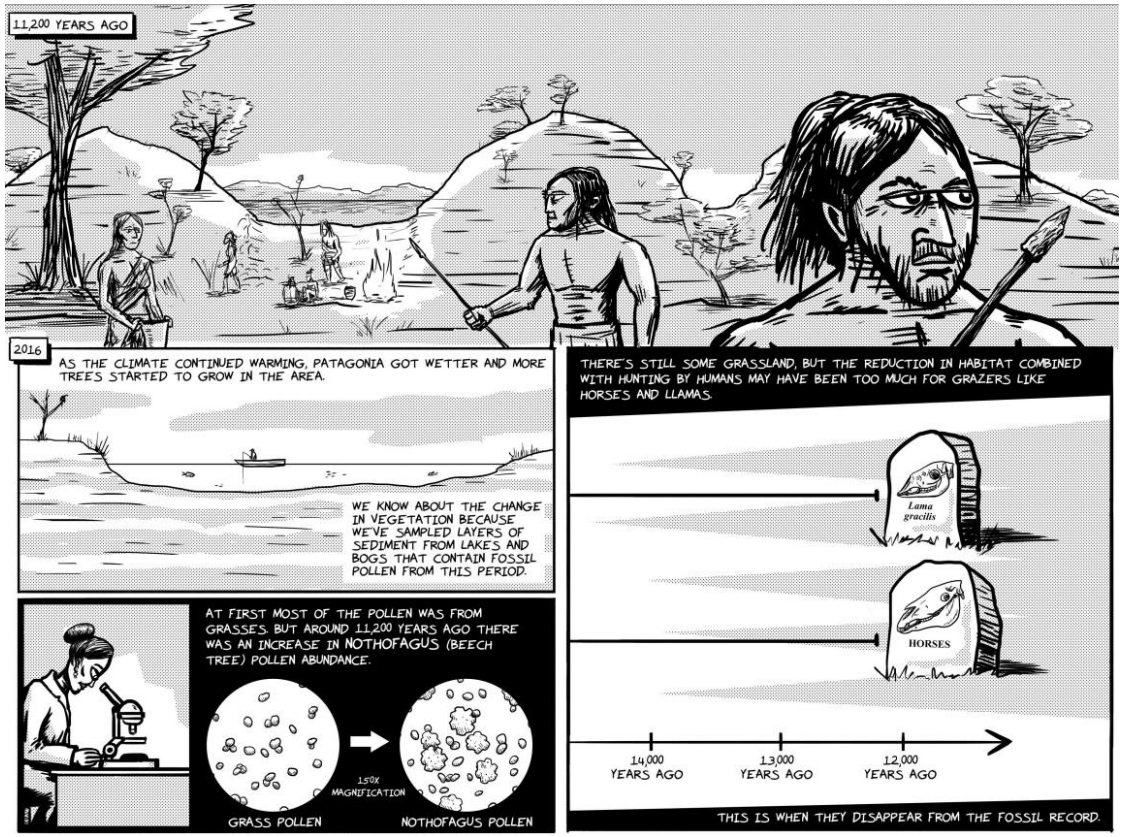
Search UE website

Home | Evolution 101 | Teach Evolution | Learn Evolution

An introduction to evolution: what is evolution and how does it work?

- The history of life: looking at the patterns
  Change over time and shared ancestors
- Mechanisms: the processes of evolution
  Selection, mutation, migration, and more
- Microevolution
  Evolution within a population
- Speciation
  How new species arise
- Macroevolution
  Evolution above the species level
- The big issues
  Pacing, diversity, complexity, and trends





NEANDERTHAL AND DENISOVAN DNA

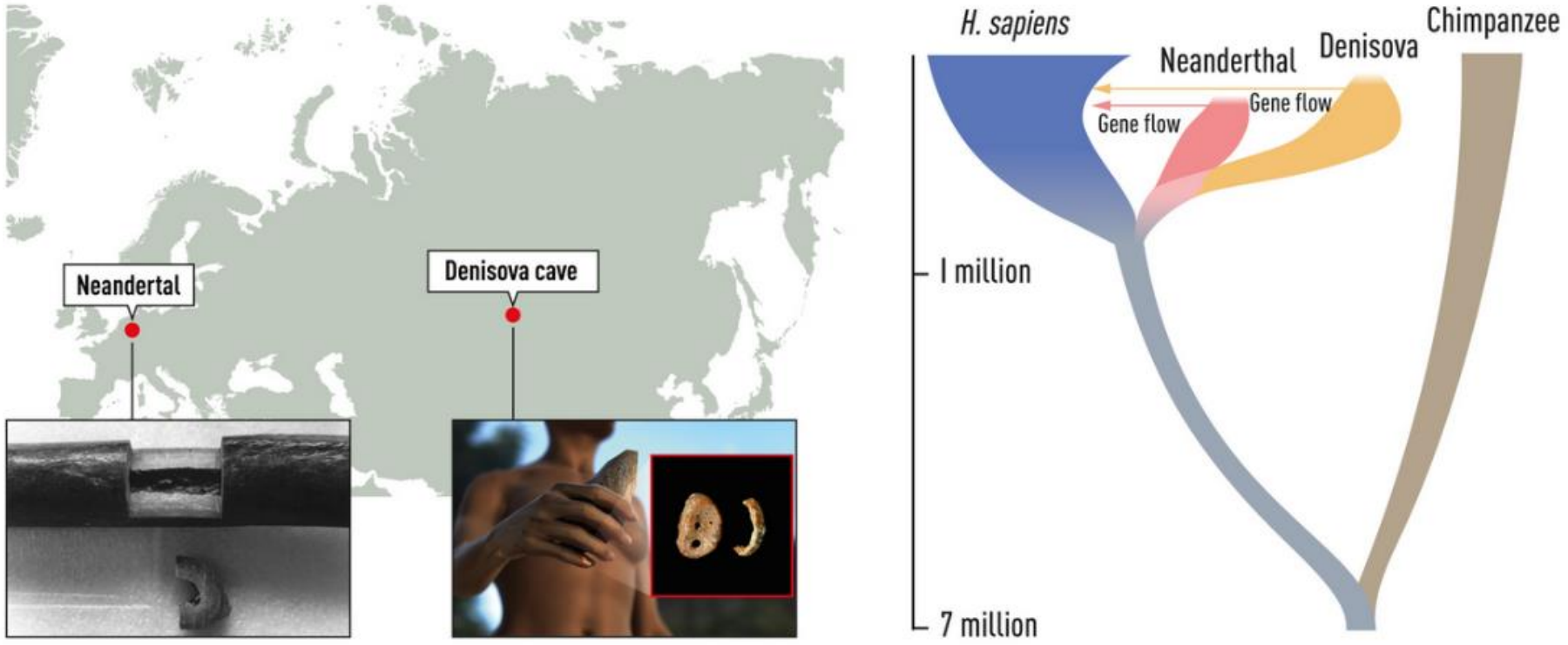


Figure 2. A. Pääbo extracted DNA from bone specimens from extinct hominins. He first obtained a bone fragment from Neandertal in Germany, the site that gave name to the Neanderthals. Later, he used a finger bone from the Denisova Cave in southern Siberia, the site that gave name to Denisovans. B. Phylogenetic tree showing the evolution and relationship between Homo sapiens and the extinct hominins. The phylogenetic tree also illustrates the gene flows discovered by Pääbo.

NEANDERTHAL AND DENISOVAN DNA

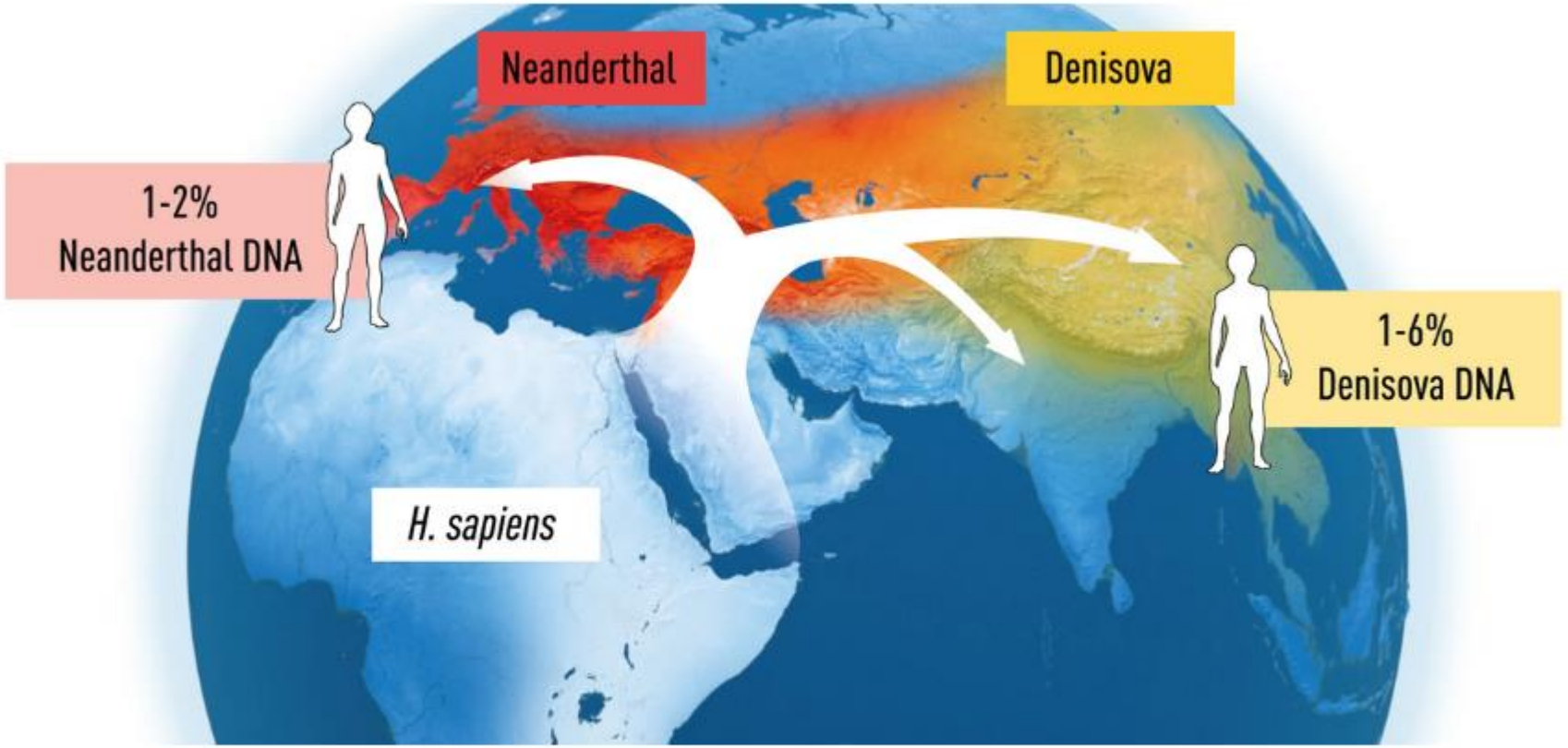
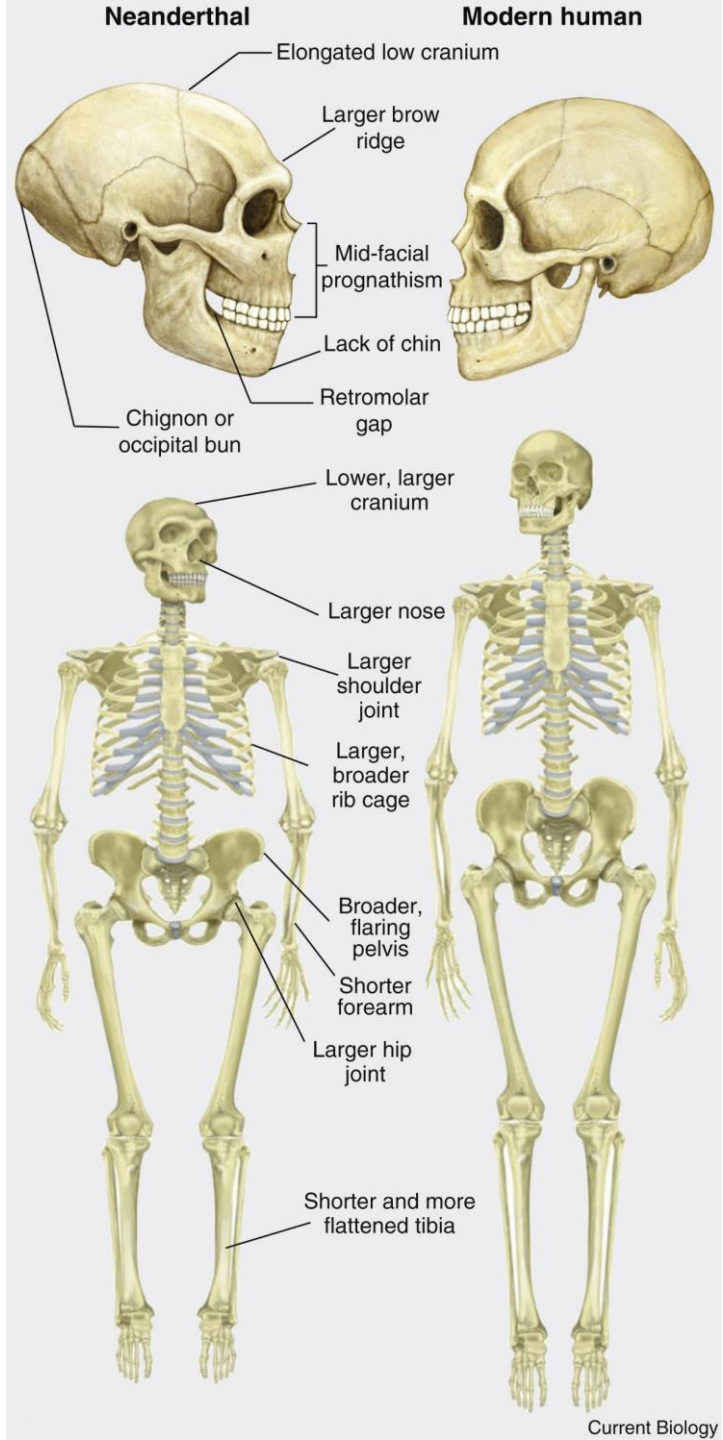
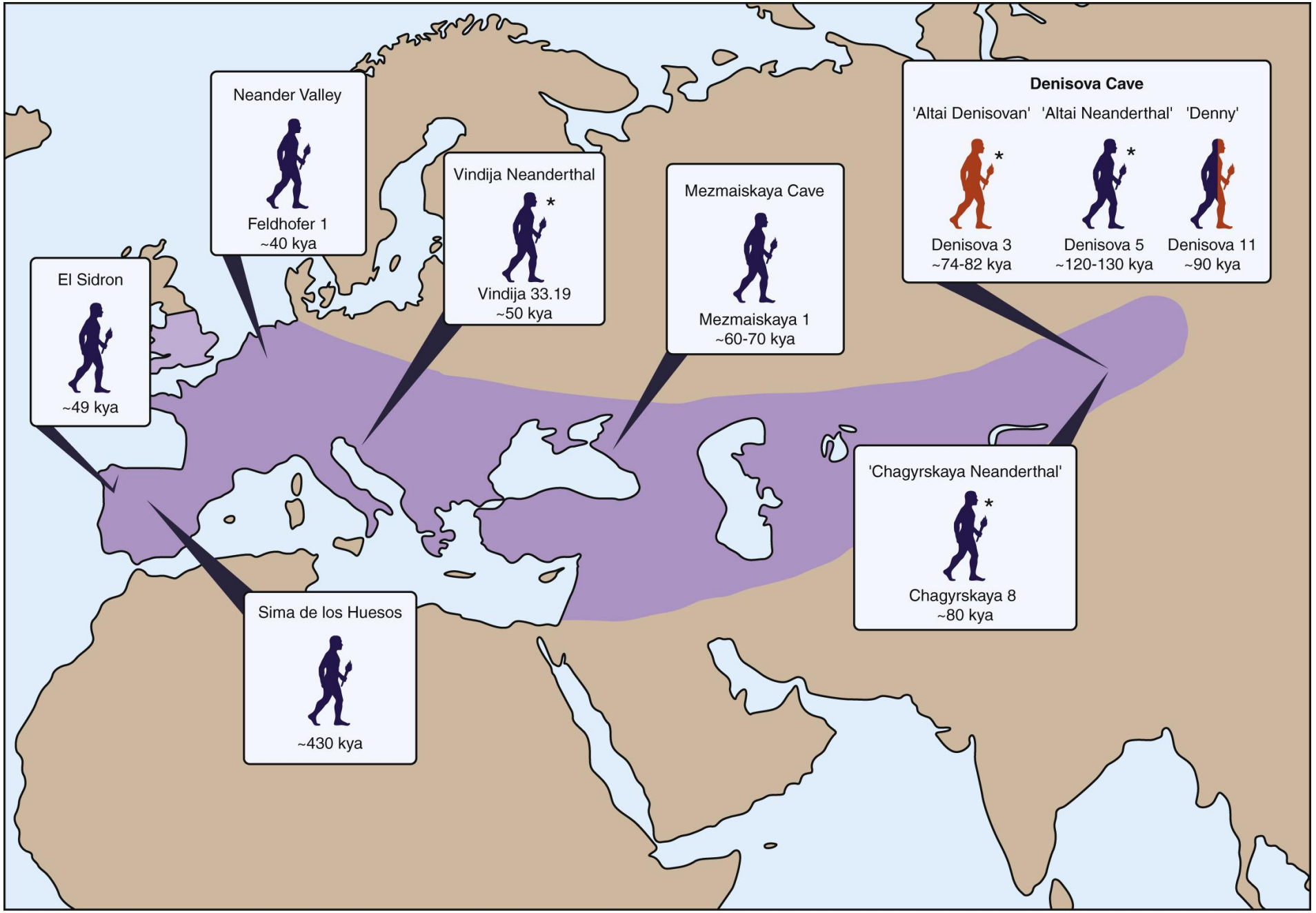


Figure 3. Pääbo's discoveries have provided important information on how the world was populated at the time when Homo sapiens migrated out of Africa and spread to the rest of the world. Neanderthals lived in the west and Denisovans in the east on the Eurasian continent. Interbreeding occurred when Homo sapiens spread across the continent, leaving traces that remain in our DNA.



Reilly et al., 2022; DOI: 10.1016/j.cub.2022.08.027



Reilly et al., 2022; DOI: 10.1016/j.cub.2022.08.027

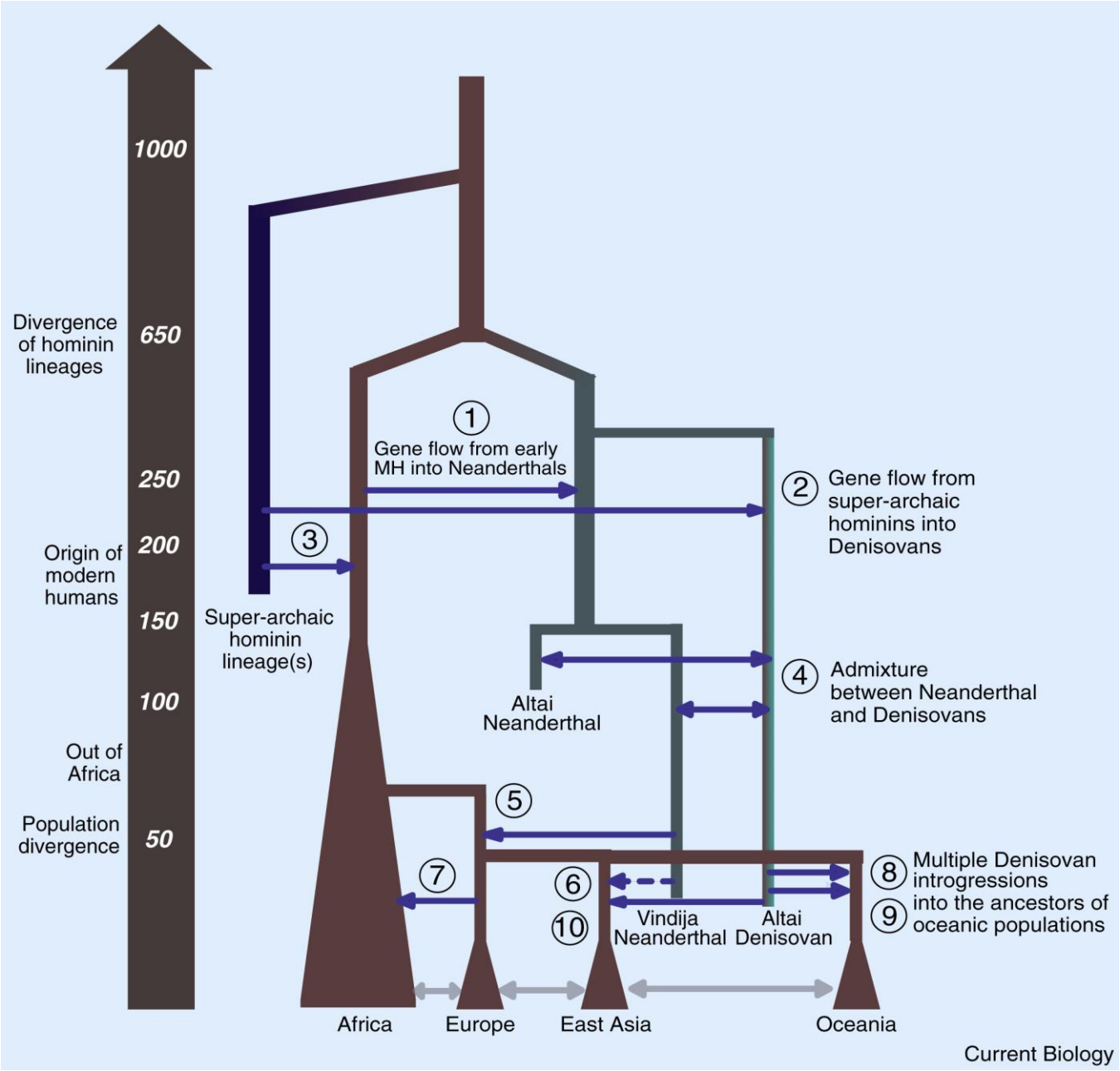


Figure 2. Simplified demographic model illustrating inferred population relationships and admixture events among modern humans and archaic hominins.

Events depicted include (1) gene flow from early modern humans (MH) into Neanderthals[20,35,151], (2) from a super-archaic hominin lineage into Denisovans[15,35], and (3) gene flow from a super-archaic hominin lineage into early modern humans in Africa[36,38, 39, 40]; (4) admixture events between Neanderthals and Denisovans in the Altai Mountains[15,152], also supported by the sequencing of the first generation offspring of a Neanderthal mother and a Denisovan father[26]; (5) gene flow from Neanderthals into the ancestors of all non-Africans[4,11,27,28,45], (6) putative gene flow from Neanderthals into the ancestors of East Asians[16,31,153] (dashed arrow), (7) back migration from Eurasia to Africa[34]; and multiple Denisovan introgressions into the ancestors of (8,9) Oceanians[9, 10, 11,13,18,45] and (10) East Asians[12]. For simplicity we represent the multiple, distinct introgressing Denisovan-like lineages as arising from a single Denisovan source (adapted with permission from[154] © 2017 Springer Nature).

Reilly et al., 2022; DOI: 10.1016/j.cub.2022.08.027

NEANDERTHAL AND DENISOVAN DNA

Letter | Published: 22 August 2018

The genome of the offspring of a Neanderthal mother and a Denisovan father

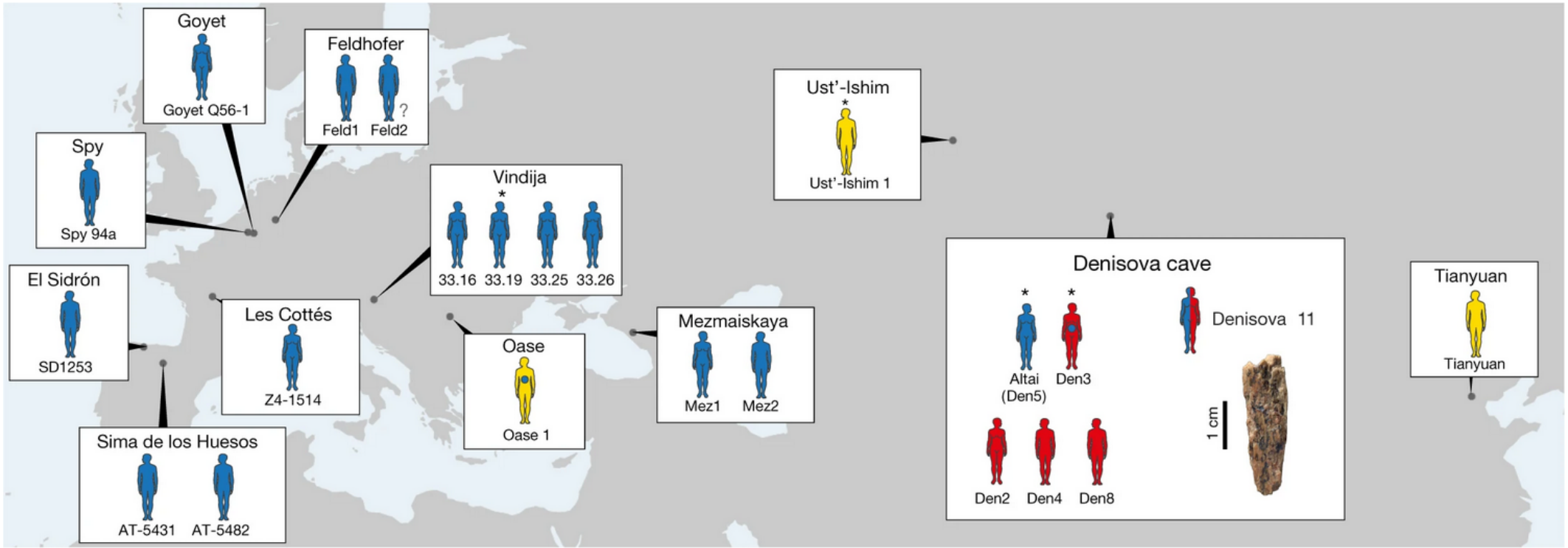
Viviane Slon ✉, Fabrizio Mafessoni, Benjamin Vernot, Cesare de Filippo, Steffi Grote, Bence Viola, Mateja Hajdinjak, Stéphane Peyrégne, Sarah Nagel, Samantha Brown, Katerina Douka, Tom Higham, Maxim B. Kozlikin, Michael V. Shunkov, Anatoly P. Derevianko, Janet Kelso, Matthias Meyer, Kay Prüfer & Svante Pääbo ✉

Nature **561**, 113–116 (2018) | Cite this article

Eastern Neanderthal group spread west about 120,000 years ago

Fig. 1: Location of Neanderthals, Denisovans and ancient modern humans dated to approximately 40 ka or earlier.

From: The genome of the offspring of a Neanderthal mother and a Denisovan father



Only individuals from whom sufficient nuclear DNA fragments have been recovered to enable their attribution to a hominin group are shown. Full or abbreviated names of specimens are shown near each individual. Blue, Neanderthals; red, Denisovans; yellow, ancient modern humans. Asterisks indicate that the genome was sequenced to high coverage; individuals with an unknown sex are marked with a question mark. Note that Oase 1 has recent Neanderthal ancestry (blue dot) that is higher than the amount seen in non-Africans. Denisova 3 has also been found to carry a small percentage of Neanderthal ancestry. Data were obtained from previous publications[1,2,5,6,7,8,11,12,13,21,22,23,24].

Slon et al., 2018; DOI: 10.1038/s41586-018-0455-x

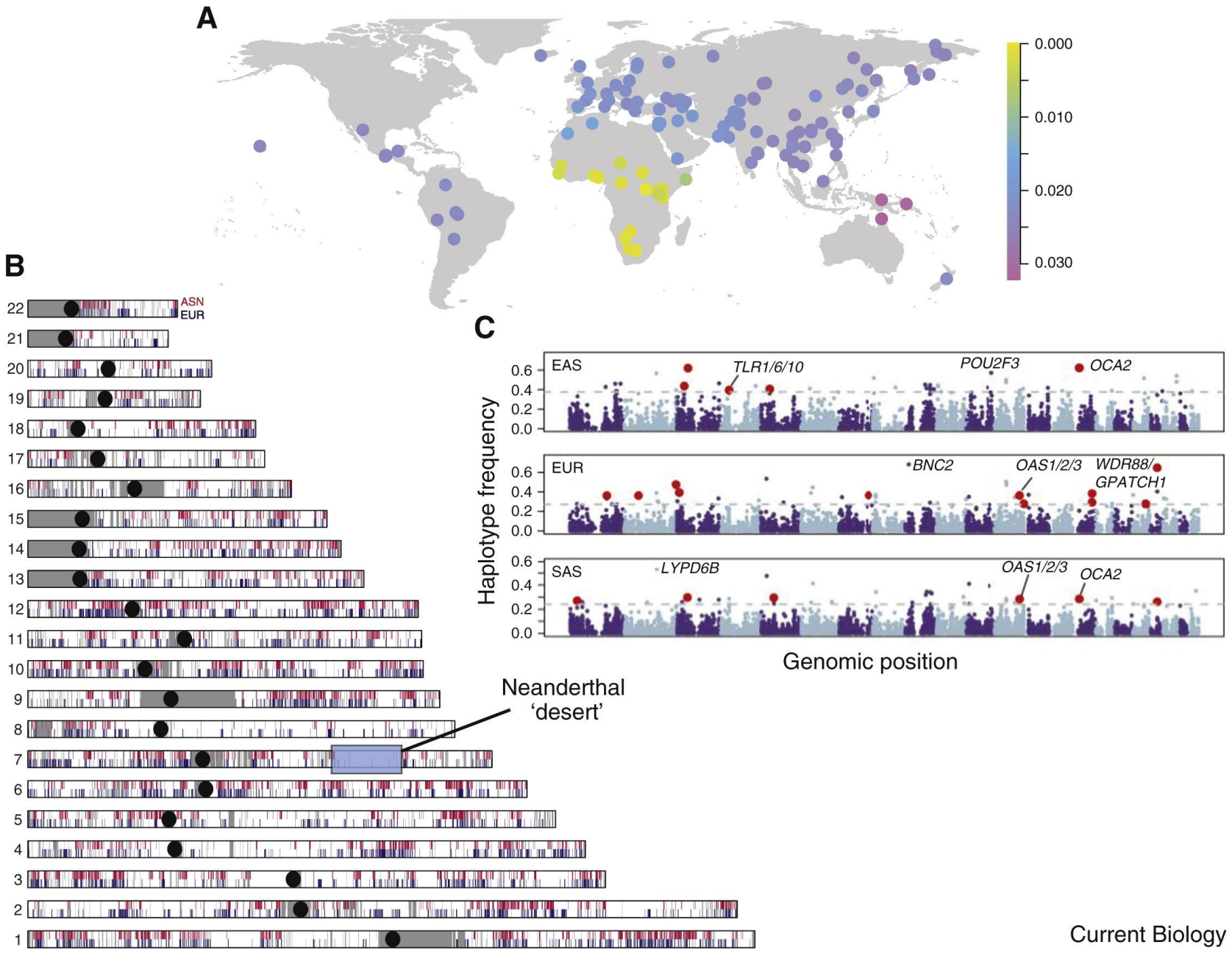


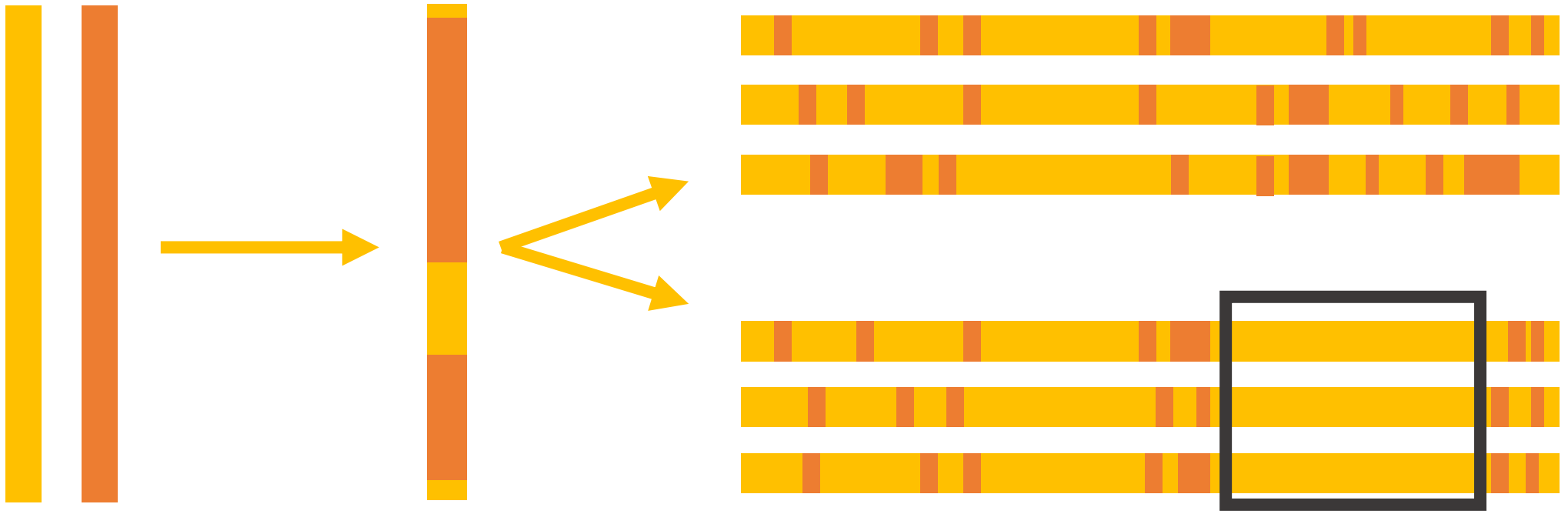
Figure 3. The geographic and genomic landscape of Neanderthal introgression.

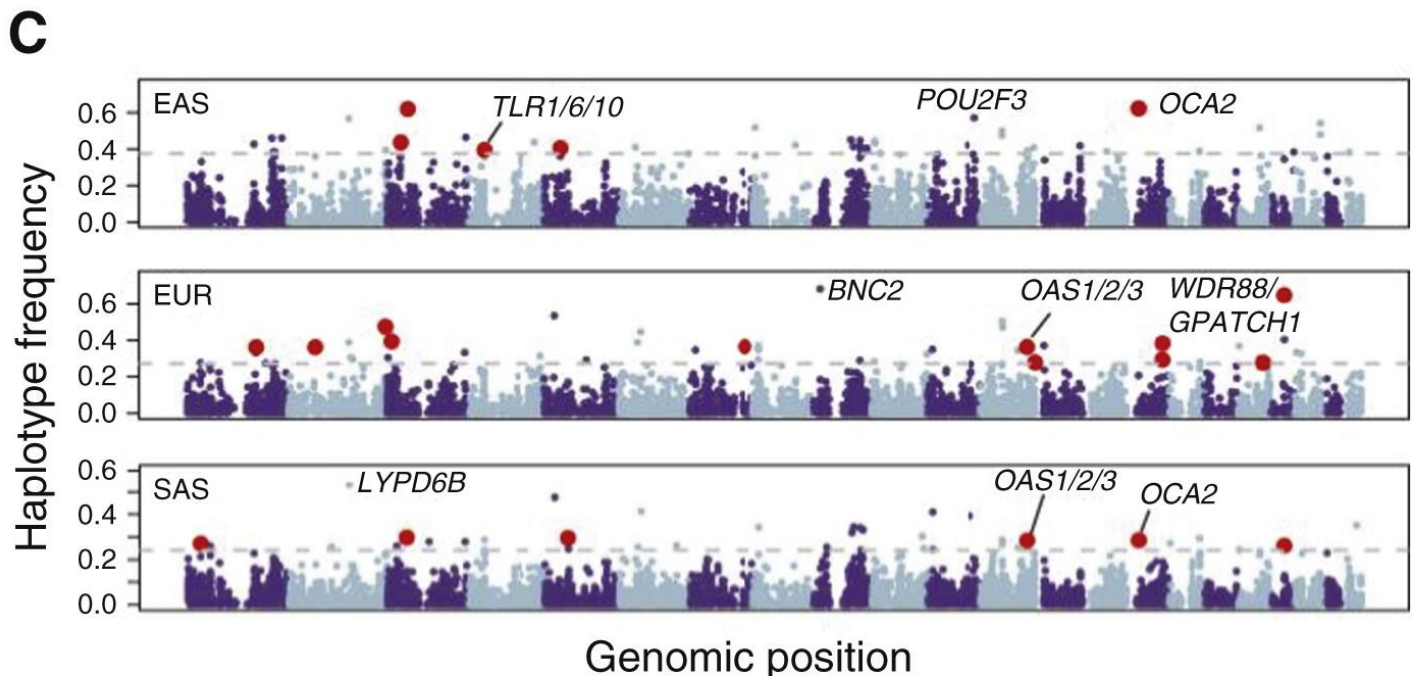
(A) Proportion of Neanderthal ancestry in geographically diverse populations (reproduced with permission from[16] © 2017 American Association for the Advancement of Science). (B) Neanderthal introgressed genomic regions are depicted by colored ticks throughout the genomes of East Asians (red) and Europeans (blue). Introgressed deserts are large genomic regions (≥10 Mbp) depleted of Neanderthal introgression. Grey regions denote genome assembly gaps, and black ovals indicate the approximate position of each centromere (reproduced with permission from[28] ©2014 American Association for the Advancement of Science). (C) Frequencies of Neanderthal introgressed haplotypes in East Asians (EAS, top), Europeans (EUR, middle), and South Asians (SAS, bottom). Positive selection after admixture with Neanderthals likely drove some of these haplotypes to high frequency. Gray dashed lines mark the 99th percentile. Haplotypes above the line are considered strong candidates for adaptive introgression. Large red dots indicate haplotypes with a significant phenotypic association.

Reilly et al., 2022; DOI: 10.1016/j.cub.2022.08.027

NEANDERTHAL AND DENISOVAN DNA

- Introgression deserts – regions depleted of certain ancestry (Neanderthal in this case) compared to expected patterns without natural selection





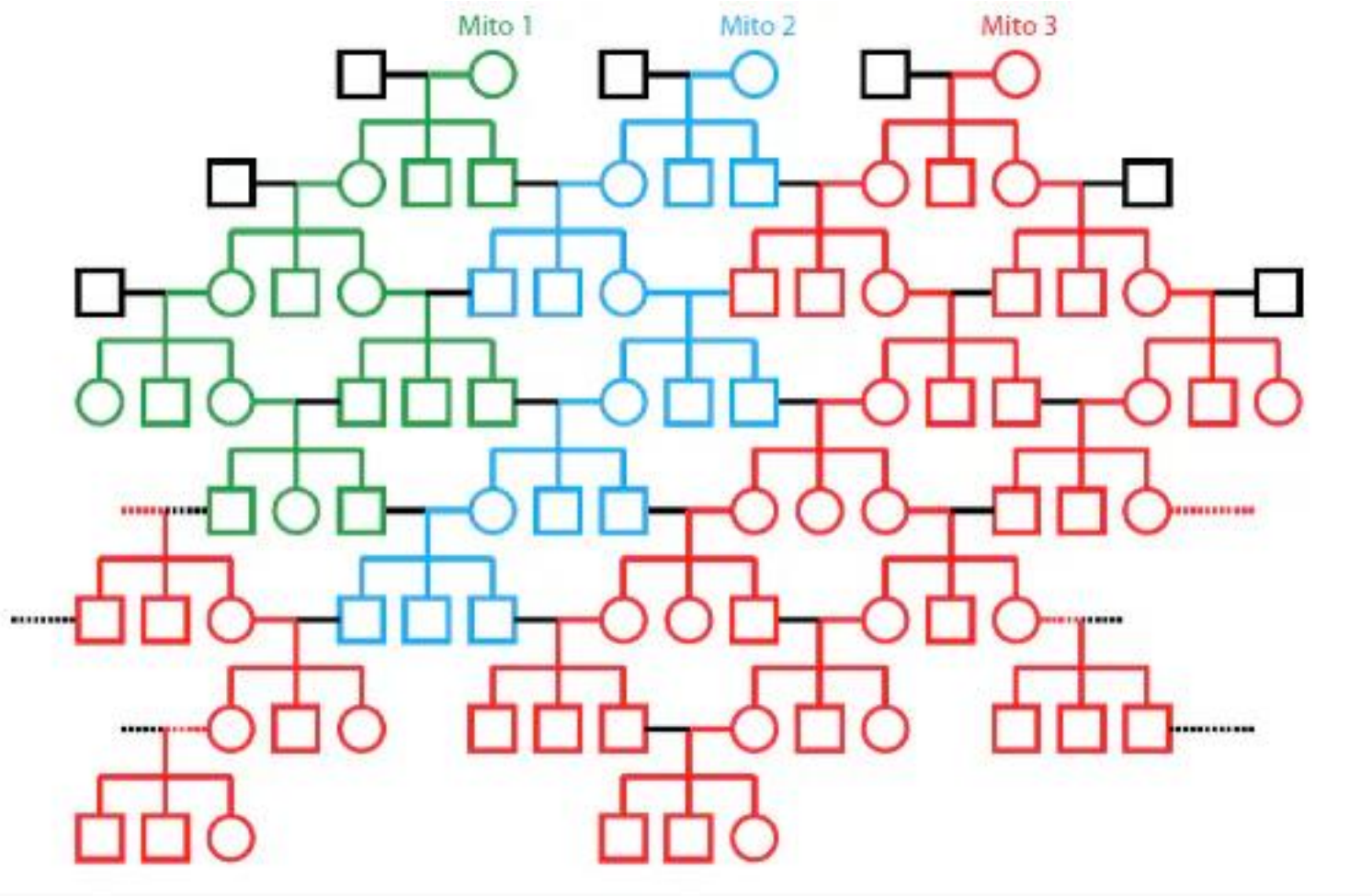
- ✕ *FOXP2* – speech and language
- ✕ Genes expressed in brain and testes
- \+ More gene regulation compared to protein structure
- \+ Skin and pigmentation (*BNC2, POU2F2, HYAL2, OCA2, KRT71, KRT80*)
- \+ Metabolism (*SLC16A11, TSHR, TBC1D1*) – lipids and energy regulation!
- \+ Immunity (*OAS, TLR1/6/10*)

- \+ *EPAS1* from Denisovans – high altitude adaptation in Tibetans

Reilly et al., 2022; DOI: 10.1016/j.cub.2022.08.027

MIGRATION AND CONTACT

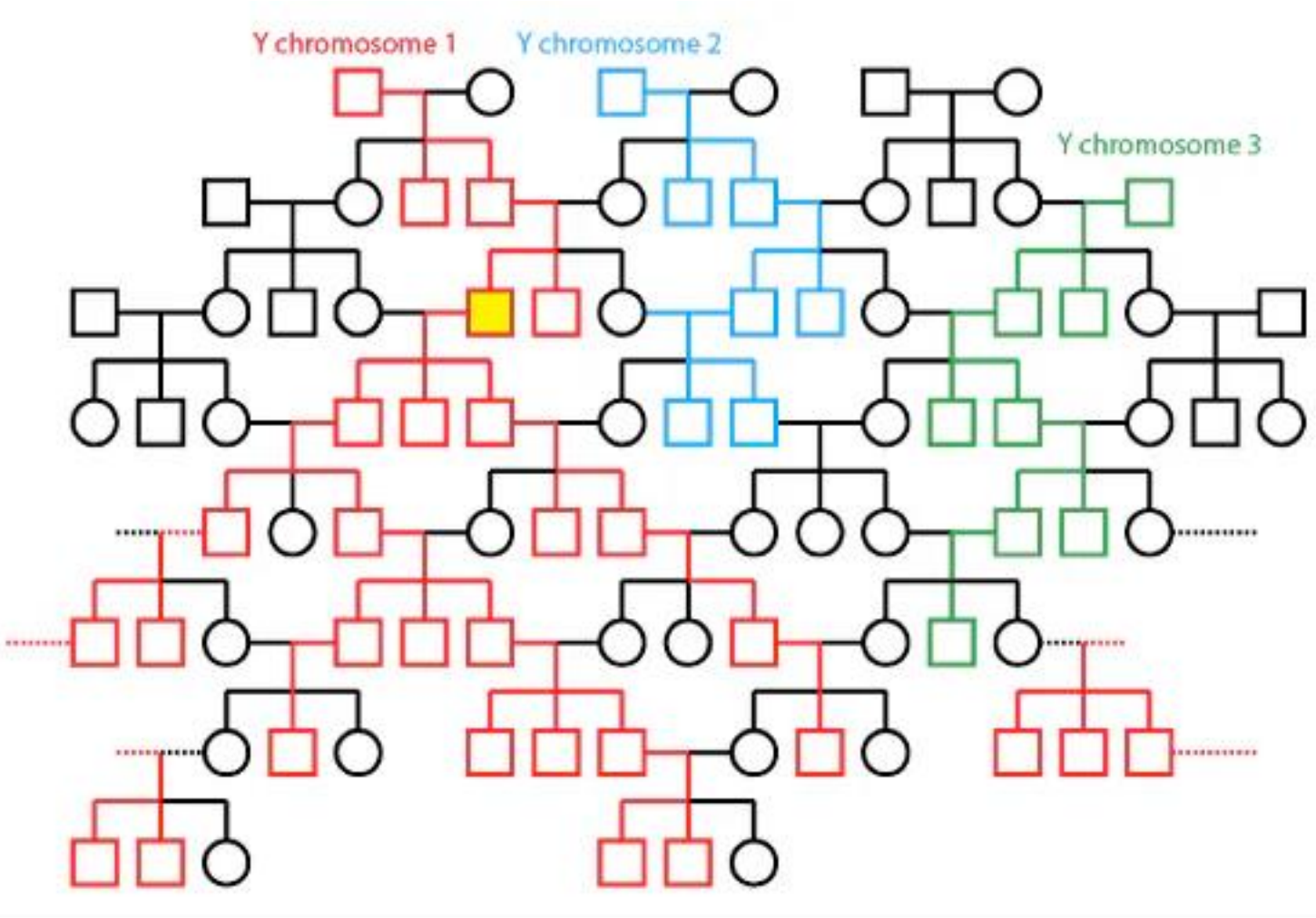
- Mitochondrial Eve



https://biologos.org/series/evolution-basics/articles/mitochondrial-eve-and-y-chromosome-adam

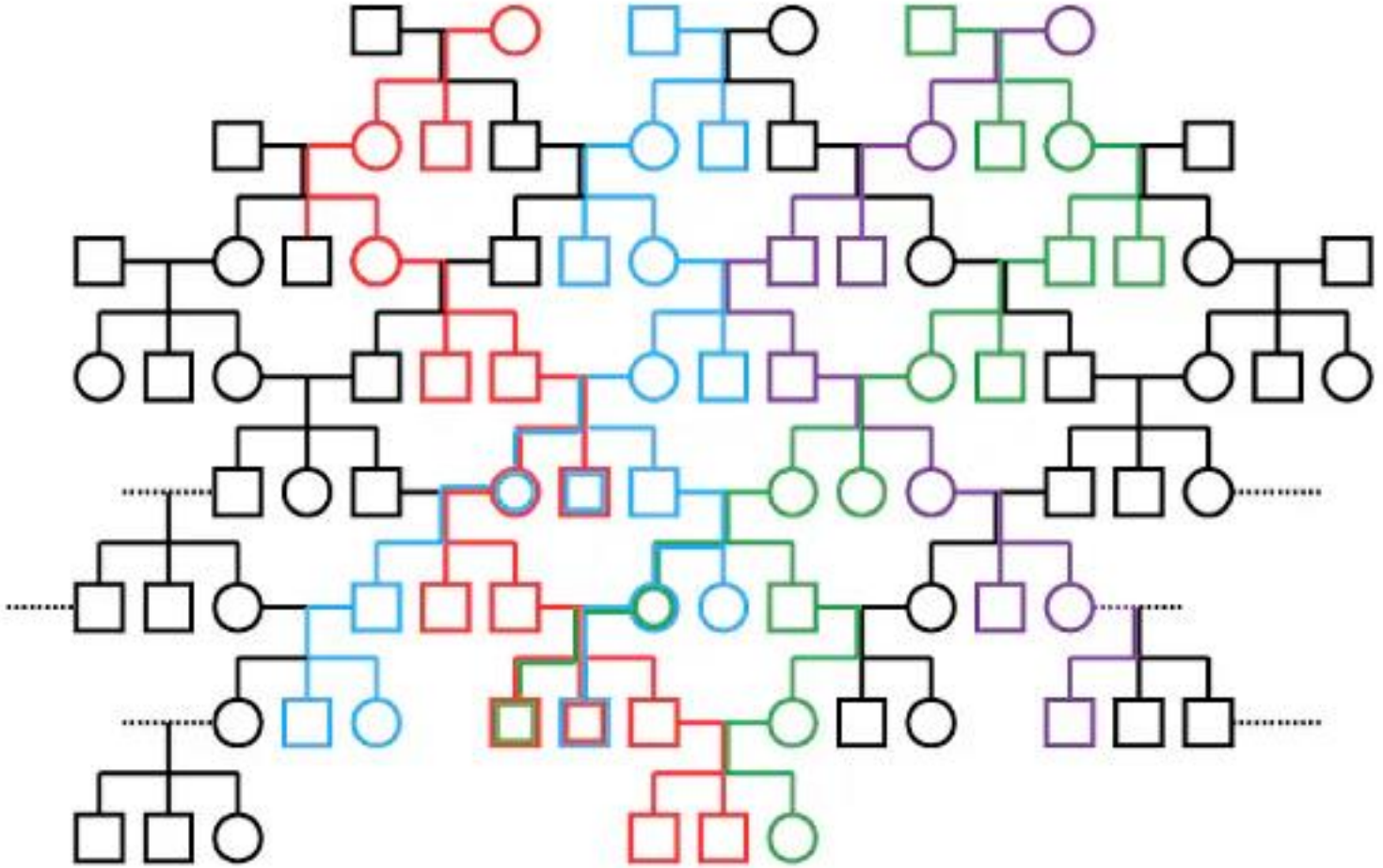
MIGRATION AND CONTACT

- Y-chromosome Adam



MIGRATION AND CONTACT

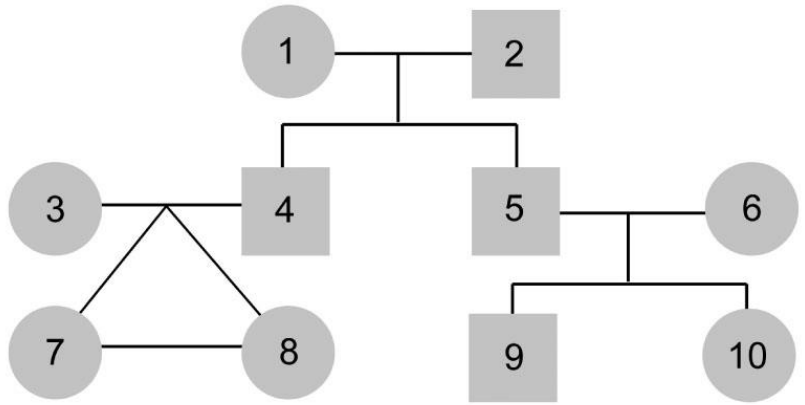
- Autosomes



https://biologos.org/series/evolution-basics/articles/mitochondrial-eve-and-y-chromosome-adam

MIGRATION AND CONTACT

- Kinship



Relationship	Example	ϕ	k_0	k_1	k_2
Identical Twins	7-8	0.5000	0	0	1.00
Full-siblings	9-10	0.2500	0.25	0.50	0.25
Parent-child	1-5	0.2500	0	1.00	0
Grandparent-grandchild	1-10	0.1250	0.50	0.50	0
First cousins	8-9	0.0625	0.75	0.25	0

FIGURE 1. Pedigree showing the degrees of relationship between selected pairs of individuals and corresponding inbreeding coefficients (ϕ) and k_0, k_1, k_2 values describing the probabilities of a given pair of individuals of sharing, respectively zero, one or two alleles that are identical-by-descent.

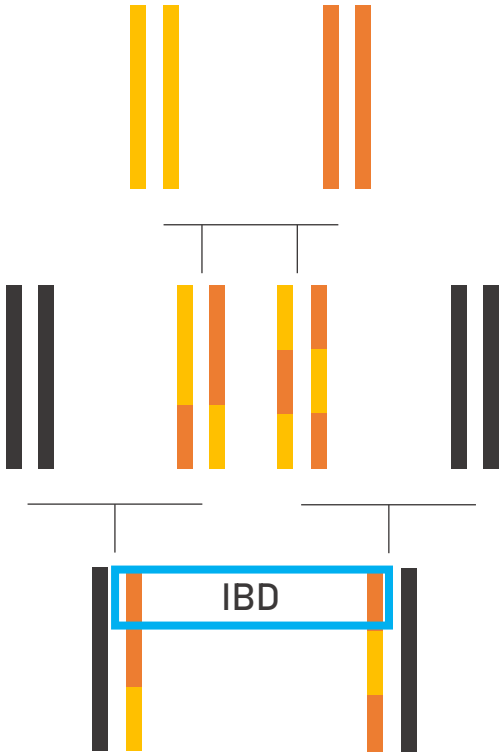
Vai et al., 2020; DOI: 10.3389/fevo.2020.00083

MIGRATION AND CONTACT

- Kinship

IBD = identical by decent (inherited from the same ancestor)

IBS = identical by state



REVIEW article

Front. Ecol. Evol., 31 March 2020
Sec. Paleoecology
Volume 8 - 2020 | https://doi.org/10.3389/fevo.2020.00083

This article is part of the Research Topic
Applied Uses of Ancient DNA
View all 14 Articles >

Kinship Determination in Archeological Contexts Through DNA Analysis

Stefania Vai[1*] Carlos Eduardo G. Amorim[2] Martina Lari[1] David Caramelli[1]

[1] Laboratorio di Antropologia Molecolare e Paleogenetica, Dipartimento di Biologia, Università di Firenze, Florence, Italy
[2] Department of Ecology and Evolutionary Biology, University of California, Los Angeles, Los Angeles, CA, United States

Knowing kinship relations between individuals in archeological contexts is of great importance to understand social habits and structure in past human communities. Archeological and anthropological analyses of burial sites and skeletal remains often allow us to infer connections between individuals, but only genetic analysis can provide a sound determination of kinship. Several case studies are now available in the literature that show the potentiality and limitations of different methodological approaches based on ancient DNA (aDNA). Both experimental and computational strategies for kinship estimation on ancient samples are described in this review and we argue that, within a multidisciplinary approach, kinship inference contributes to the understanding of the biological and cultural patterns that characterized past societies.

MIGRATION AND CONTACT

Article | Open access | Published: 25 August 2021

Genome of a middle Holocene hunter-gatherer from Wallacea

Selina Carlhoff, Akin Duli, Kathrin Nägele, Muhammad Nur, Laurits Skov, Iwan Sumantri, Adhi Agus Oktaviana, Budianto Hakim, Basran Burhan, Fardi Ali Syahdar, David P. McGahan, David Bulbeck, Yinika L. Perston, Kim Newman, Andi Muhammad Saiful, Marlon Ririmasse, Stephen Chia, Hasanuddin, Dwia Aries Tina Pulubuhu, Suryatman, Supriadi, Choongwon Jeong, Benjamin M. Peter, Kay Prüfer, ... Adam Brumm

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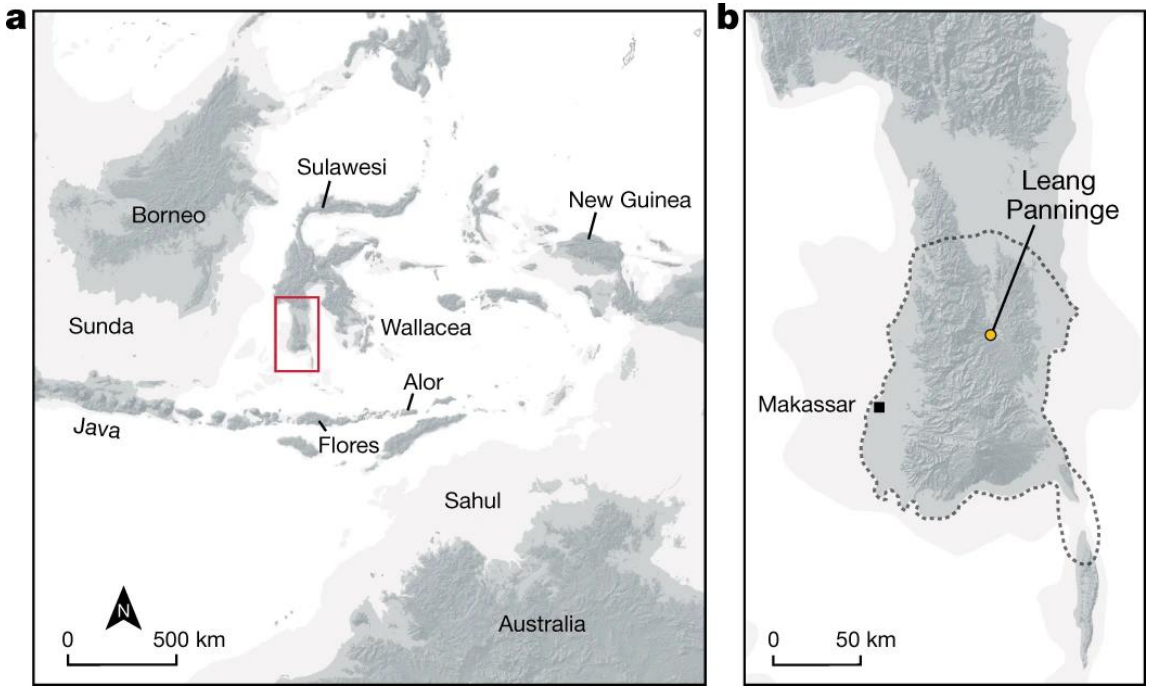
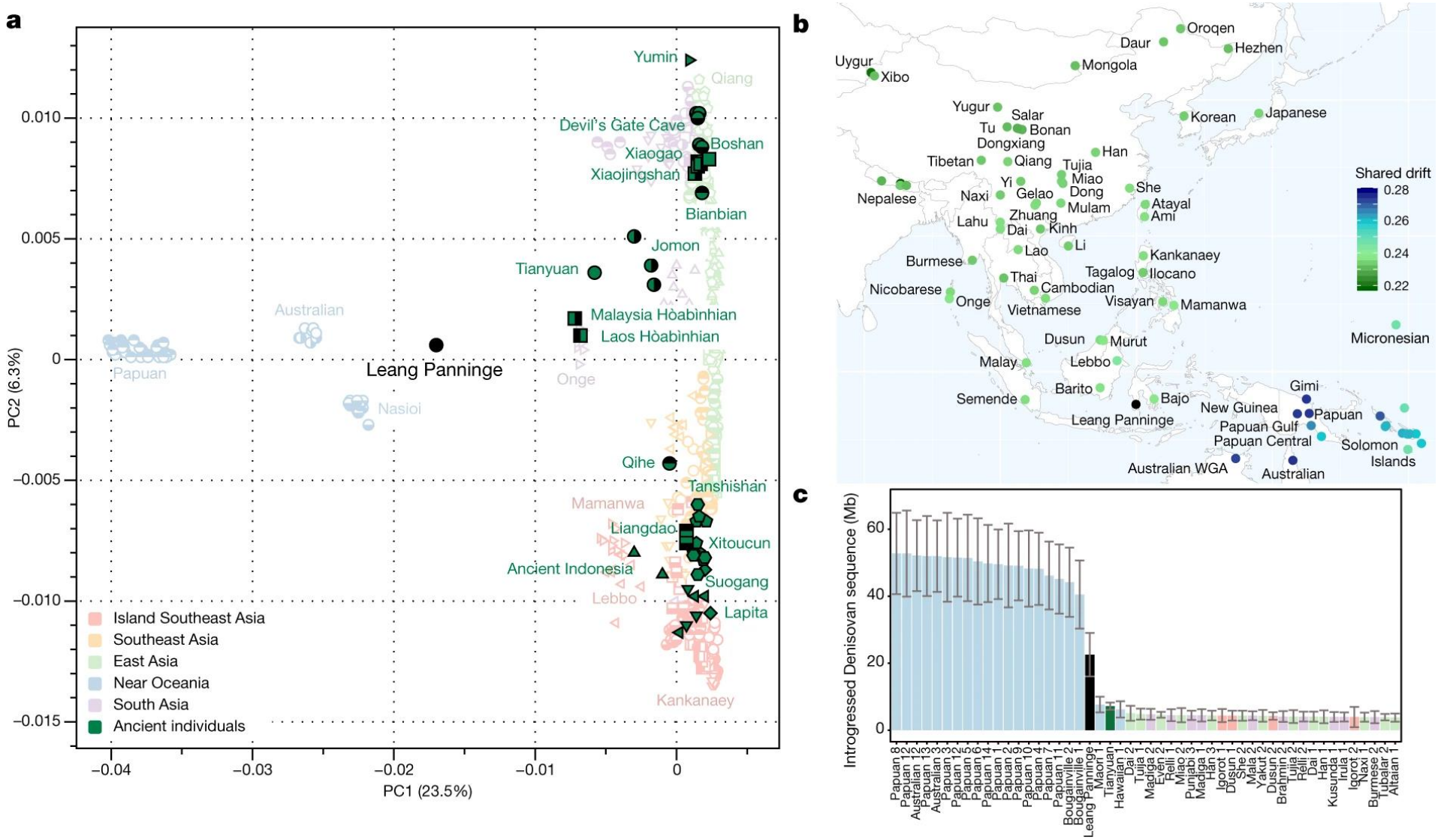
Nature **596**, 543–547 (2021) | Cite this article

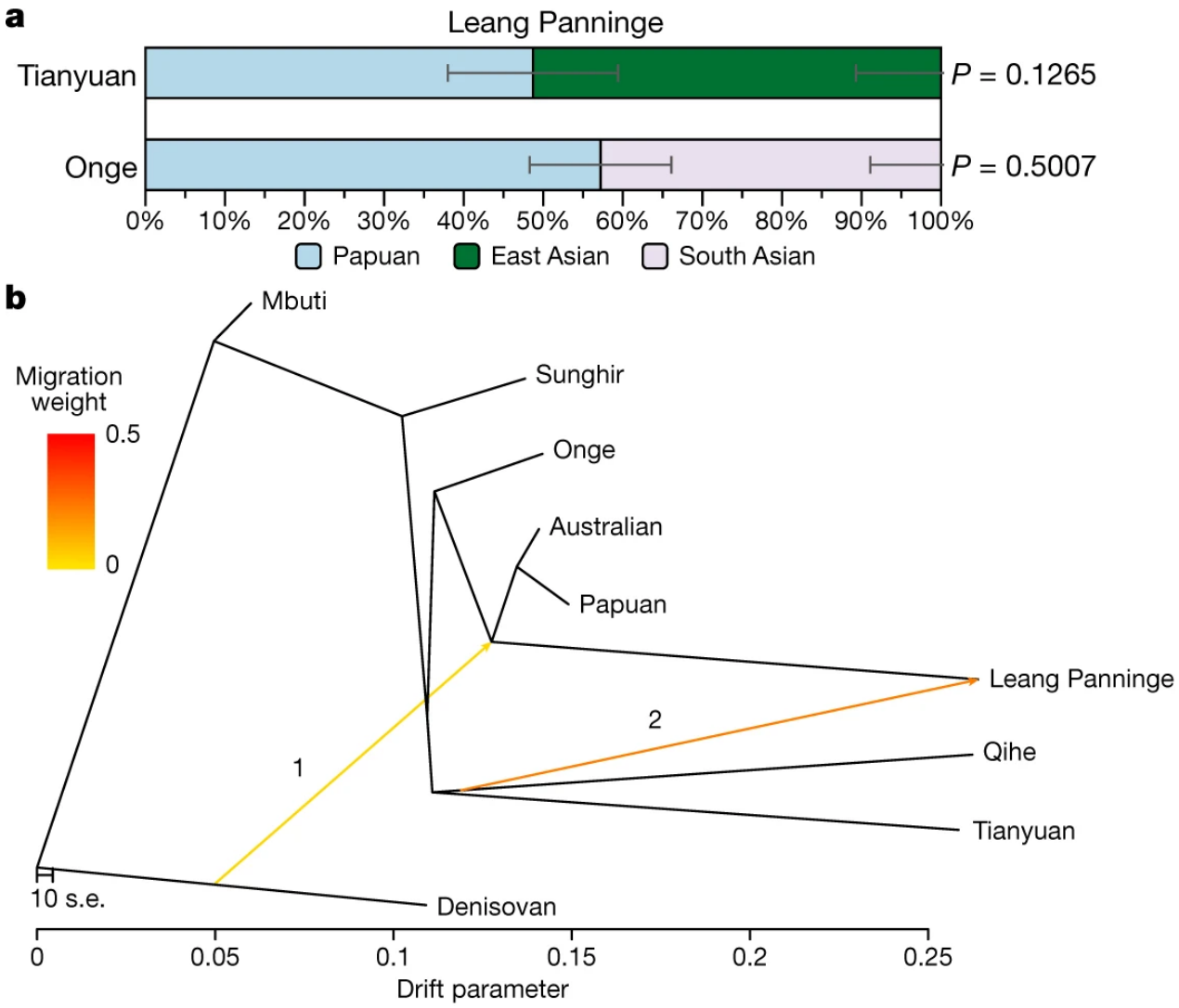
Wallacea - Island originally between Sunda Shelf and Pleistocene Sahul





a, PCA calculated on present-day individuals from eastern Eurasia and Near Oceania, projecting key ancient individuals from the region[1,34,35,36,37,38]. **b**, Shared genetic drift of present-day groups with the Leang Panninge individual, as calculated using f_3 (Mbuti; Leang Panninge, X) mapped at the geographical position of the tested group. WGA, whole genome amplification. **c**, The amount of introgressed Denisovan sequence in fragments longer than 0.05 cM in present-day (Simons Genome Diversity Project) individuals and longer than 0.2 cM in ancient individuals (measured with admixfrog). Each bar represents the posterior mean estimate from a single genome and the whiskers indicate 2 s.d. (estimated from 200 samples from the posterior decoding).

Carlhoff et al., 2021; DOI: 10.1038/s41586-021-03823-6



a, Admixture proportions modelling Leang Panninge as a combination of Papuan[49] and Tianyuan[38] or Onge[49] groups as estimated by qpAdm[33] using Mbuti, Denisovan[39], Kostenki 14 (ref. [50]) and ancient Asian individuals[1,37] as rotating reference groups (Supplementary Table 26). The error bars denote standard errors as calculated with block jacknife in the qpAdm software. **b**, **c**, Admixture graphs placing Leang Panninge on the branch with the present-day Near Oceanian clade[41] and showing the admixture with a deep Asian-related ancestry in TreeMix[42] (**b**) (Extended Data Fig. 10, Supplementary Fig. 6) and qpGraph (**c**) (worst *z*-score of −2.194; Supplementary Figs. 7–11)[33,37,38,39,43]. In **b**, '1' and '2' refer to the order in which the TreeMix software added 'migration events' (indicated by the arrows) to the graph. When plotting qpGraph results (**c**), the dotted arrows indicate admixture edges.

Carlhoff et al., 2021; DOI: 10.1038/s41586-021-03823-6

MIGRATION AND CONTACT

Letter | Published: 03 October 2016

Genomic insights into the peopling of the Southwest Pacific

Pontus Skoglund, Cosimo Posth, Kendra Sirak, Matthew Spriggs, Frederique Valentin, Stuart Bedford, Geoffrey R. Clark, Christian Reepmeyer, Fiona Petchey, Daniel Fernandes, Qiaomei Fu, Eadaoin Harney, Mark Lipson, Swapan Mallick, Mario Novak, Nadin Rohland, Kristin Stewardson, Syafiq Abdullah, Murray P. Cox, Françoise R. Friedlaender, Jonathan S. Friedlaender, Toomas Kivisild, George Koki, Pradiptajati Kusuma, D. Andrew Merriwether, Francois-X. Ricaut, Joseph T. S. Wee, Nick Patterson, Johannes Krause, Ron Pinhasi & David Reich — Show fewer authors

Nature **538**, 510–513 (2016) | Cite this article

The appearance of people associated with the Lapita culture in the South Pacific around 3,000 years ago[1] marked the beginning of the last major human dispersal to unpopulated lands. However, the relationship of these pioneers to the long-established Papuan people of the New Guinea region is unclear. Here we present genome-wide ancient DNA data from three individuals from Vanuatu (about 3,100–2,700 years before present) and one from Tonga (about 2,700–2,300 years before present), and analyse them with data from 778 present-day East Asians and Oceanians. Today, indigenous people of the South Pacific harbour a mixture of ancestry from Papuans and a population of East Asian origin that no longer exists in unmixed form, but is a match to the ancient individuals. Most analyses have interpreted the minimum of twenty-five per cent Papuan ancestry in the region today as evidence that the first humans to reach Remote Oceania, including Polynesia, were derived from population mixtures near New Guinea, before their further expansion into Remote Oceania[2,3,4,5]. However, our finding that the ancient individuals had little to no Papuan ancestry implies that later human population movements spread Papuan ancestry through the South Pacific after the first peopling of the islands.

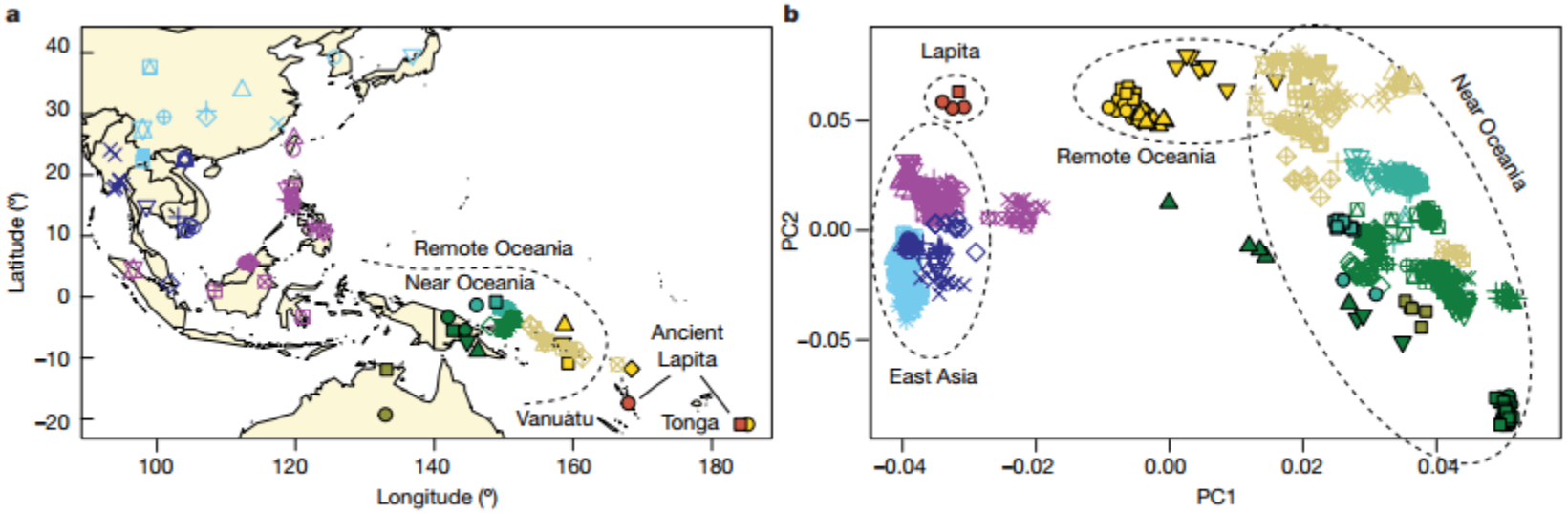
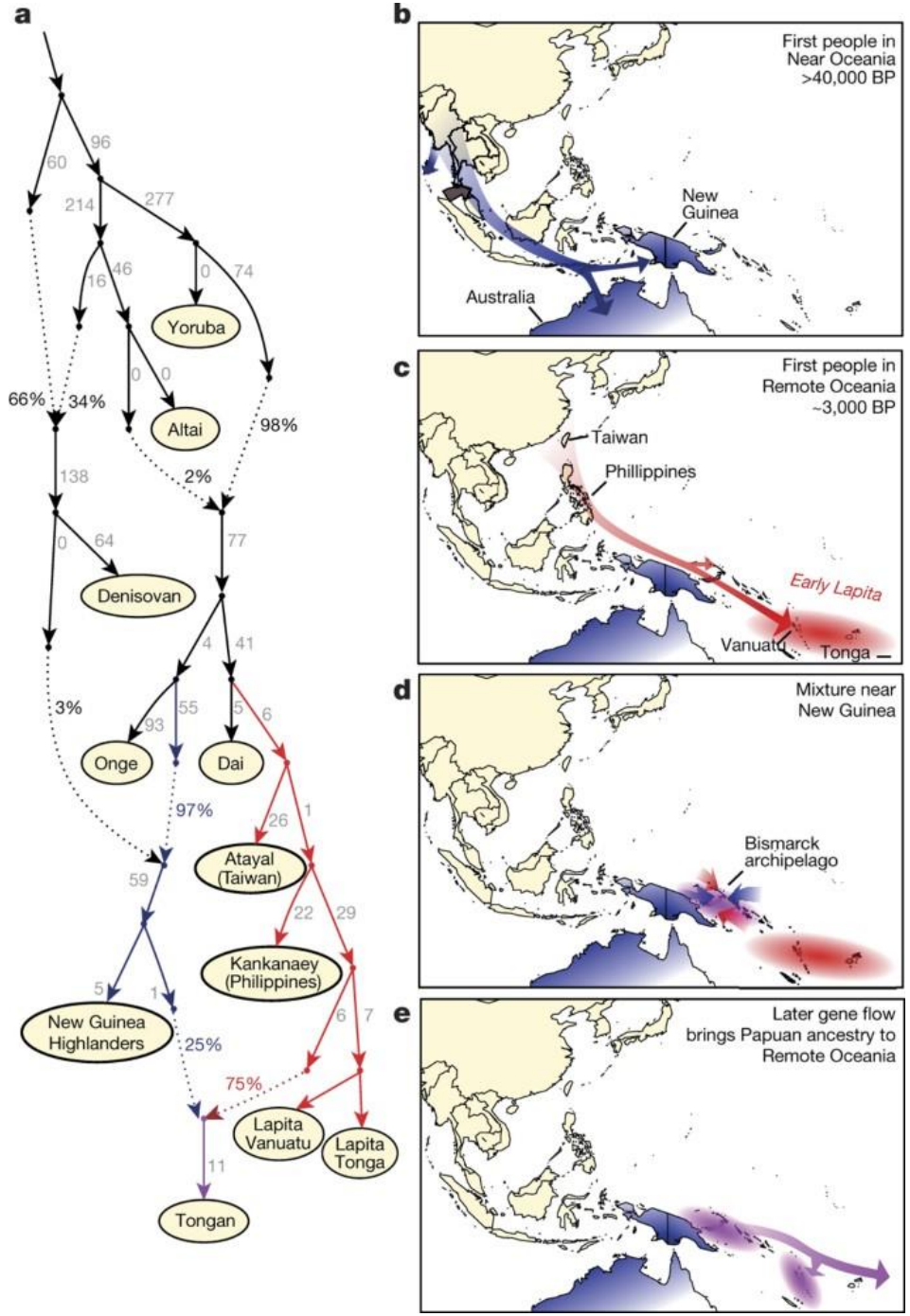


Figure 1 | Data from ancient and present-day populations. a, Locations of 778 present-day individuals genotyped on the Affymetrix Human Origins Array and 4 ancient individuals (red symbols). **b**, Ancient individuals projected onto principal components (PC) 1 and 2 computed using only present-day samples. Individual population labels are given in Extended Data Fig. 2.



a, A model of population relationships that fits allele frequency patterns (all empirical *f*-statistics within 3 standard errors of expectation). Branch lengths are shown in units of $F_{ST} \times 1{,}000$. Admixture edges show mixture proportions. Altai, the Altai Neandertal genome. **b**, A model of population movements more than 40,000 years ago in which modern humans arrived in the Australia–New Guinea region (blue shading) and mixed with archaic Denisovans (brown arrow). **c**, A model of events before 3,000 years ago, in which the First Remote Oceanian population formed by spread of a population of ultimate East Asian origin to a region including Vanuatu and Tonga, and experienced little or no mixture with the Papuans they encountered along the journey (red shading). Note that geographic routes are speculative. **d**, A model of populations of mixed Papuan–First Remote Oceanian ancestry in Near Oceania less than 3,000 years ago in a patchwork of islands with different proportions of First Remote Oceanian ancestry (pink shading). **e**, A model of secondary expansion of admixed populations bringing Papuan ancestry into Remote Oceania, which was still not complete in Tonga by the date of the Talasiu individual at 2,680–2,340 BP.

Skoglund et al., 2016; DOI: 10.1038/nature19844

Article | Open access | Published: 24 May 2023

Reanalyzing the genetic history of Kra-Dai speakers from Thailand and new insights into their genetic interactions beyond Mainland Southeast Asia

Piya Changmai, Yutthaphong Phongbunchoo, Jan Kočí & Pavel Flegontov

Scientific Reports **13**, Article number: 8371 (2023) | Cite this article

Sci Rep. 2022 Dec 29;12(1):22507. doi: 10.1038/s41598-022-26799-3.

Ancient DNA from Protohistoric Period Cambodia indicates that South Asians admixed with local populations as early as 1st–3rd centuries CE

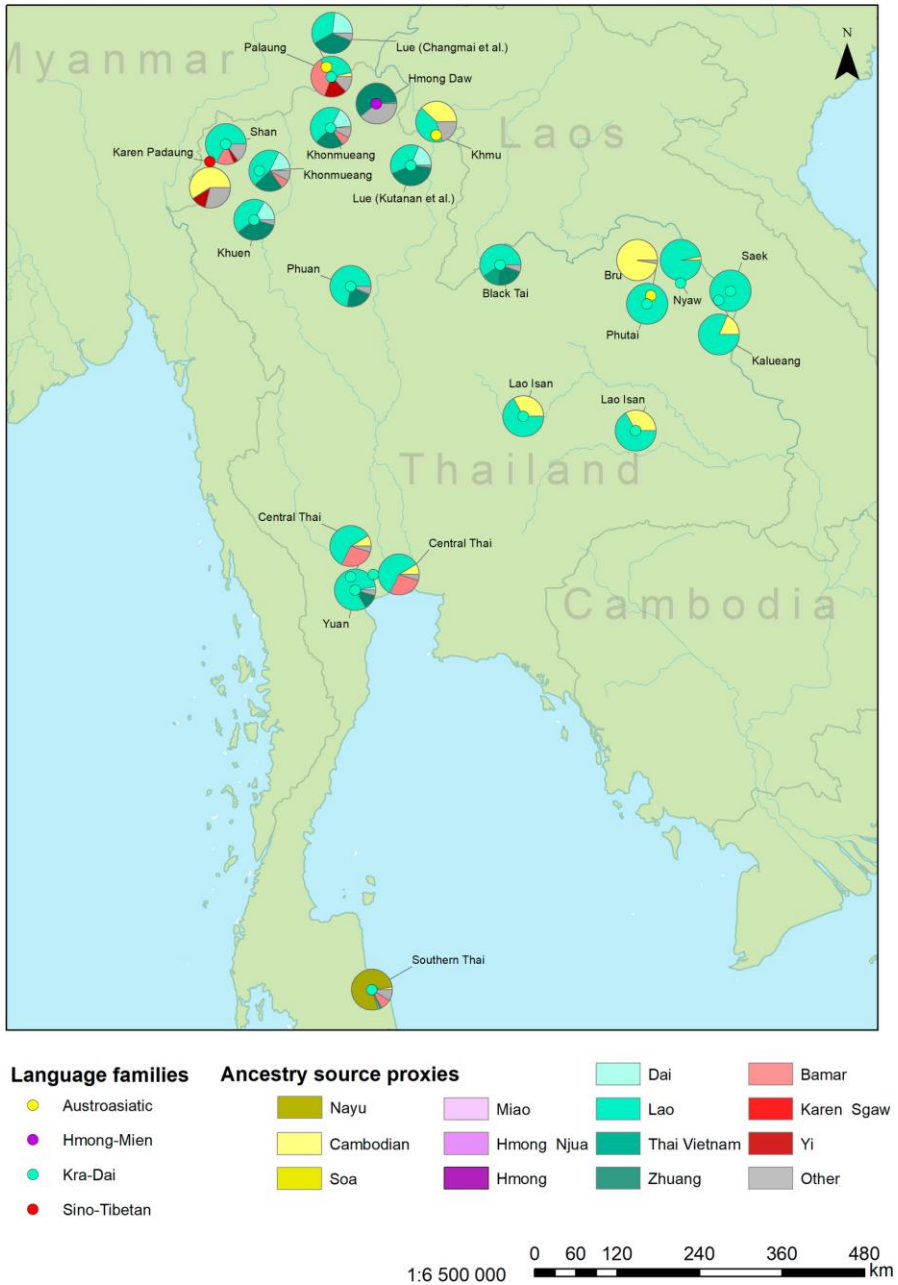
Piya Changmai [1], Ron Pinhasi [2 3], Michael Pietrusewsky [4], Miriam T Stark [4], Rona Michi Ikehara-Quebral [4 5], David Reich [6 7 8 9], Pavel Flegontov [10 11 12 13]

Affiliations + expand

PMID: 36581666 PMCID: PMC9800559 DOI: 10.1038/s41598-022-26799-3

An Ancient Harappan Genome Lacks Ancestry from Steppe Pastoralists or Iranian Farmers

Vasant Shinde [1 14 15], Vagheesh M. Narasimhan [2 14], Nadin Rohland [2], Swapan Mallick [2 3 4], Matthew Mah [2 3 4], Mark Lipson [2], Nathan Nakatsuka [2], Nicole Adamski [2 3], Nasreen Broomandkhoshbacht [2 3 10], Matthew Ferry [2 3], Ann Marie Lawson [2 3], Megan Michel [2 3 11 12], Jonas Oppenheimer [2 3 13], Kristin Stewardson [2 3], Nilesh Jadhav [1], Yong Jun Kim [1], Malavika Chatterjee [1], Avradeep Munshi [1], Amrithavalli Panyam [1], Pranjali Waghmare [1], Yogesh Yadav [1], Himani Patel [5], Amit Kaushik [6], Kumarasamy Thangaraj [7], Matthias Meyer [8], Nick Patterson [4 9], Niraj Rai [5 7 15], David Reich [2 3 4 15 16]



MIGRATION AND CONTACT

Ancient Ethiopian genome reveals extensive Eurasian admixture in Eastern Africa

M. GALLEGO LLORENTE, E. R. JONES, A. ERIKSSON, V. SISKA, K. W. ARTHUR, J. W. ARTHUR, M. C. CURTIS, J. T. STOCK, M. COLTORTI, P. PIERUCCINI, S. STRETTON, F. BROCK, T. HIGHAM, Y. PARK, M. HOFREITER, D. G. BRADLEY, J. BHAK, R. PINHASI, AND A. MANICA fewer Authors Info & Affiliations

SCIENCE · 8 Oct 2015 · Vol 350, Issue 6262 · pp. 820-822 · DOI: 10.1126/science.aad2879

Characterizing genetic diversity in Africa is a crucial step for most analyses reconstructing the evolutionary history of anatomically modern humans. However, historic migrations from Eurasia into Africa have affected many contemporary populations, confounding inferences. Here, we present a 12.5× coverage ancient genome of an Ethiopian male ("Mota") who lived approximately 4500 years ago. We use this genome to demonstrate that the Eurasian backflow into Africa came from a population closely related to Early Neolithic farmers, who had colonized Europe 4000 years earlier.

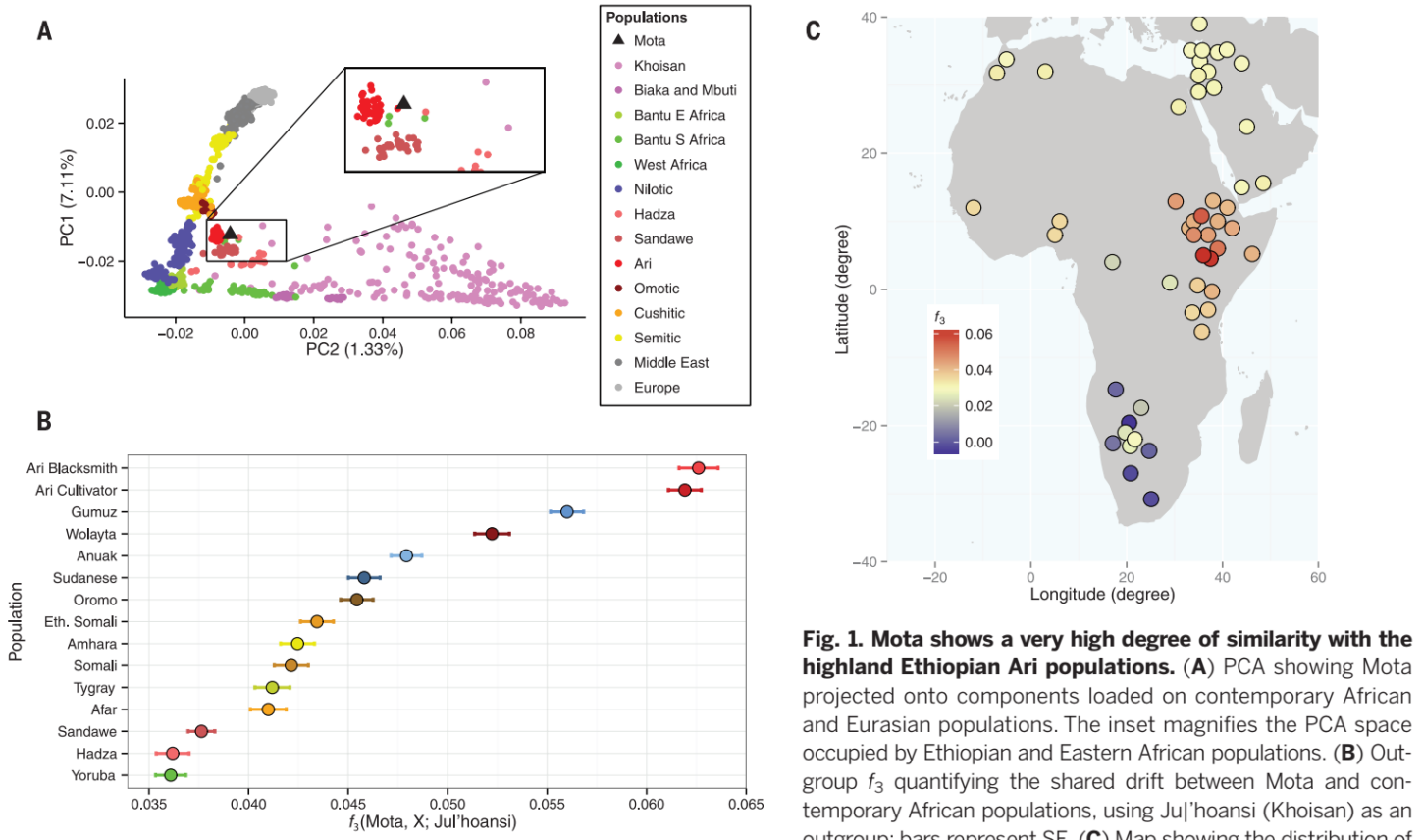
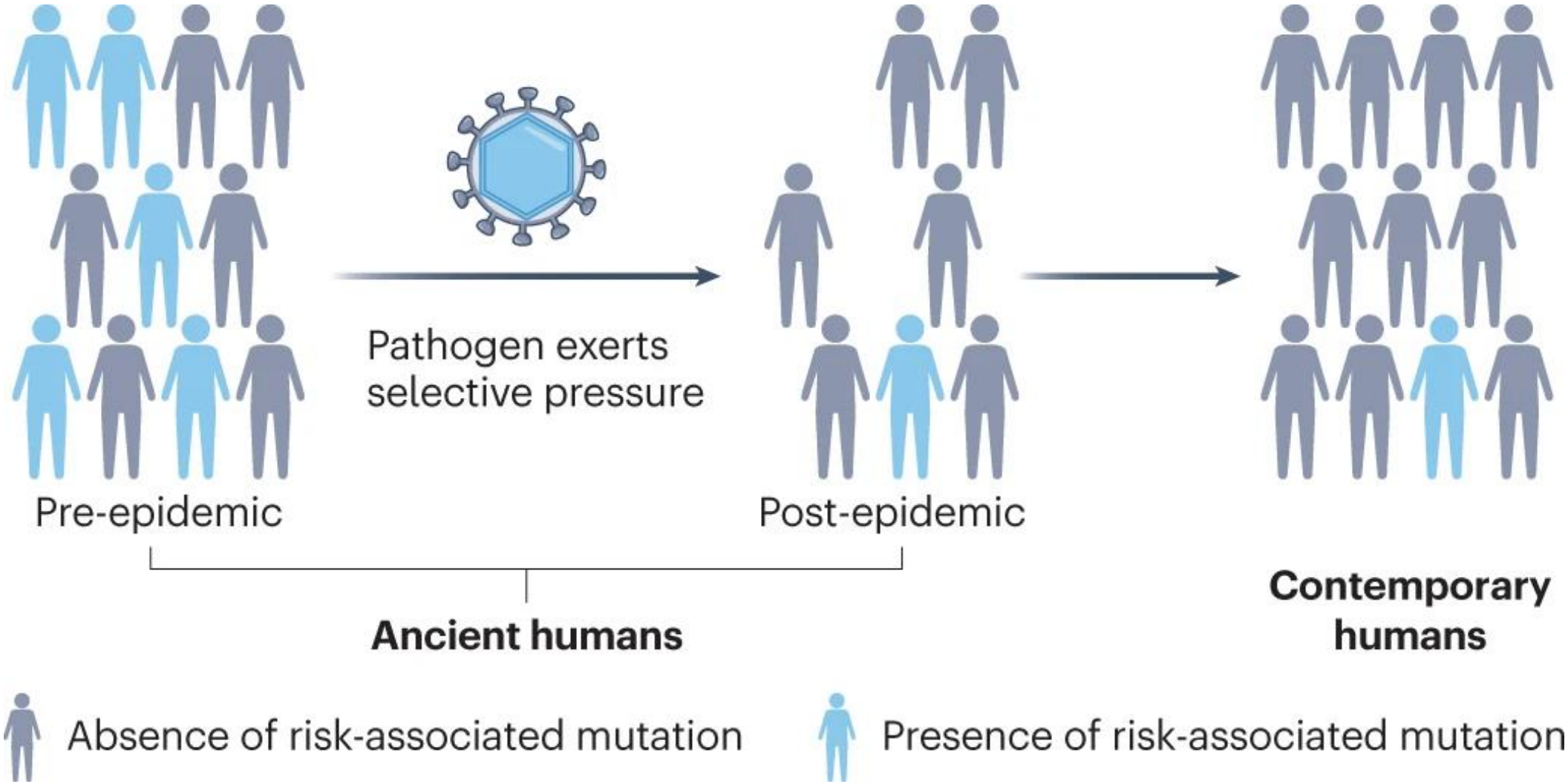


Fig. 1. Mota shows a very high degree of similarity with the highland Ethiopian Ari populations. (**A**) PCA showing Mota projected onto components loaded on contemporary African and Eurasian populations. The inset magnifies the PCA space occupied by Ethiopian and Eastern African populations. (**B**) Outgroup f_3 quantifying the shared drift between Mota and contemporary African populations, using Ju|'hoansi (Khoisan) as an outgroup; bars represent SE. (**C**) Map showing the distribution of outgroup f_3 values across the African continent. In (A) and (B), populations speaking Nilo-Saharan languages are marked with blue shades, Omotic speakers with red, Cushitic with orange, Semitic with yellow, and Bantu with green. Mota is denoted by a black symbol.

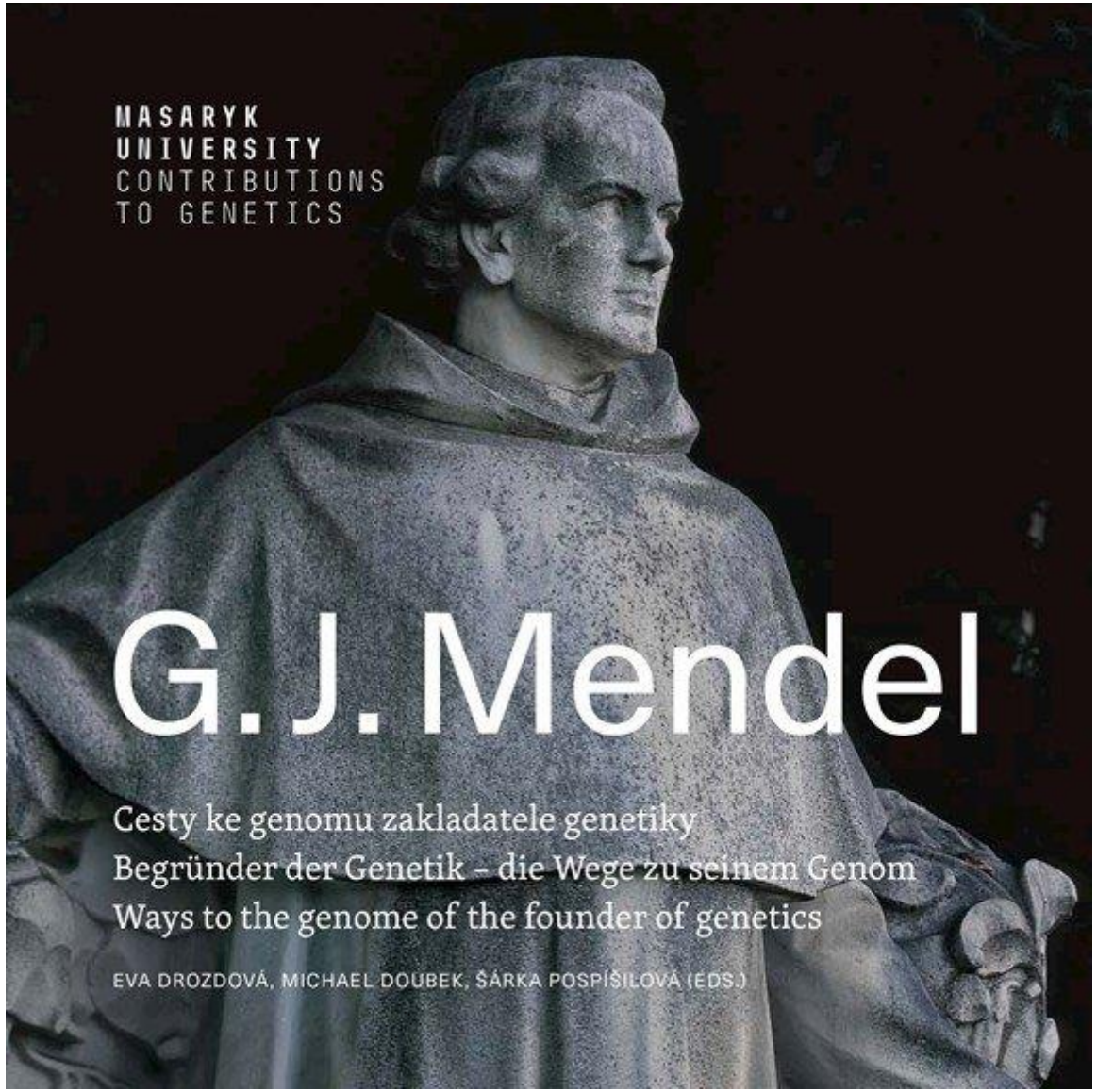
DISEASE-RELATED CHANGES



Kerner et al., 2023; DOI: 10.1038/s41591-023-02244-4

HISTORICAL FIGURES

- G. J. Mendel
- L. van Beethoven
- Tatanka Iyotake


MASARYK
UNIVERSITY
CONTRIBUTIONS
TO GENETICS
G.J. Mendel
Cesty ke genomu zakladatele genetiky
Begründer der Genetik – die Wege zu seinem Genom
Ways to the genome of the founder of genetics
EVA DROZDOVÁ, MICHAEL DOUBEK, ŠÁRKA POSPÍŠILOVÁ (EDS.)

FOLIA MENDELIANA

SUPPLEMENTUM AD

ACTA MUSEI MORAVIAE

Edited by Jiří Sekerák

2022 58/1

MORAVSKÉ ZEMSKÉ MUZEUM BRNO 2022

BODY REMAINS OF ABBOT GREGOR JOHANN MENDEL

EVA DROZDOVÁ, DANA FIALOVÁ, EVA CHOCHOLOVÁ, KATEŘINA NOVOTNÁ, ANNA ŠENOVSKÁ, KRISTÝNA BRZOBOHATÁ, ROMANA SARVAŠOVÁ, LENKA FALK
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FILIP PARDY
Genomics Core Facility, Central European Institute of Technology, Masaryk University, Brno, Czech Republic

ABSTRACT – In the occasion of the 200th anniversary of birth an interdisciplinary research of skeletal remains of Gregor Johann Mendel (buried at Central cemetery in Brno) was carried out by a team of researchers coming from Masaryk University. The aim of the research is to perform a complex survey of the skeletal remains found in the Augustinian tomb. The main focus is concentrated on identification of the person of Gregor Mendel, on evaluation of biological traits of his body (via his skeletal remains) and on obtaining a evaluating his genetical information.
Archaeological and anthropological field research at the Central Cemetery of Brno pass in period from June 6 to June 30 2021 including exhumation of body remains from found coffins. All excavated skeletons and mummified tissues were further explored in the laboratory using anthropological and genetic methods.
Anthropological and genetical evaluation of the skeletal remains of Abbot Mendel brought new information on his body parameters.

MULTIDISCIPLINARY APPROACH TO IDENTIFICATION OF GREGOR JOHANN MENDEL'S SKELETAL REMAINS

DANA FIALOVÁ[1*], EVA DROZDOVÁ[1], EVA CHOCHOLOVÁ[1], KRISTÝNA BRZOBOHATÁ[1], FILIP PARDY[2], ANNA ŠENOVSKÁ[1], HANA SVOBODOVÁ[1], KATEŘINA NOVOTNÁ[1], BORIS TICHÝ[2], MAREK PEŠKA[3], ANTONÍN ZÚBEK[3], MICHAEL DOUBEK[4, 5], ŠÁRKA POSPÍŠILOVÁ[4, 5]
[1] Laboratory of Biological and Molecular Anthropology, Department of Experimental Biology, Faculty of Science, Masaryk University, Brno, CZ
[2] CF Genomics, CEITEC Masaryk University, Brno, CZ
[3] Archaia Brno z.ú., CZ
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ABSTRACT - Introduction and aims.
is difficult process, hampered by l
nucleobases and contamination risk.
bioinformatics to inspect genomic fea
Methods: aDNA was isolated from t
Amicon filter unit. After QC, Illumin
kit (IDT), using molecular barcode
amplified with EvaGreen dye to optin
Illumina NovaSeq instrument with
bioinformatics pipeline, freely availa
SNV/Indel variants and aDNA qual
was performed on cluster using Kube
Results: We managed to reconstruc
average coverage. We detected 4,1 mi
them, 57308 occurred in the gene co
Discussion and conclusion: Using
bioinformatics, we reconstructed Me
have shaped his life and health. Var
by biologists and physicians. In th
consider this as an important contrib

METAGENOMIC AND PROTEOMIC ANALYSIS OF DENTAL CALCULUS OF ABBOT GREGOR JOHANN MENDEL

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ABSTRACT - As part of the interdisciplinary research of abbot and founder of genetics, Gregor Johann Mendel, his dental calculus was sampled to gain insight into his health that could not be obtained by mere genome sequencing. A total of 469 proteins were found after filtering, including proteins related to immunity and oral health. Multiple oral species connected to dental pathology such as periodontitis were confirmed based on both DNA and proteins, originating from Porphyromonas gingivalis, Tannerella forsythia, or Treponema denticola. High abundance of Clostridiales DNA was confirmed, in agreement with mummification by saponification.

HISTORICAL FIGURES

Current Biology

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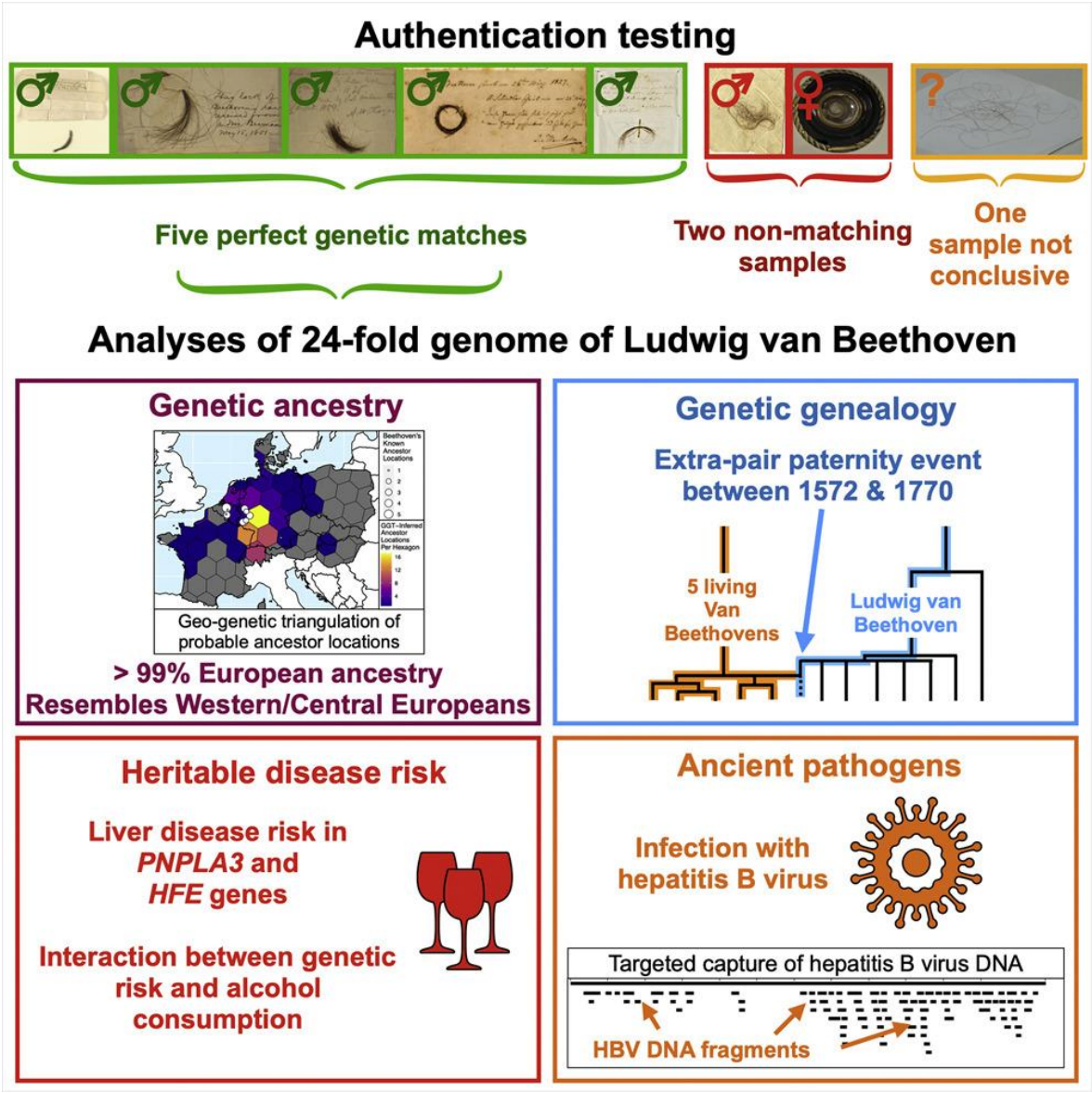
Article

Genomic analyses of hair from Ludwig van Beethoven

Tristan James Alexander Begg [1 6 24], Axel Schmidt [2], Arthur Kocher [9 22 24], Maarten H.D. Larmuseau [3 14 15 16], Göran Runfeldt [4], Paul Andrew Maier [4], John D. Wilson [5 28], Rodrigo Barquera [9], Carlo Maj [2 27], András Szolek [18 19], Michael Sager [4], Stephen Clayton [6 24], Alexander Peltzer [26], Ruoyun Hui [8 20], Julia Ronge [12], Ella Reiter [6], Cäcilia Freund [24], Marta Burri [24], Franziska Aron [24], Anthi Tiliakou [9 24]...Johannes Krause [6 9 24 29 30]



HISTORICAL FIGURES

Identifying a living great-grandson of the Lakota Sioux leader Tatanka Iyotake (Sitting Bull)

IDA MOLTKE, THORFINN SAND KORNELIUSSEN, ANDAINE SEGUIN-ORLANDO, J. VÍCTOR MORENO-MAYAR, ERNIE LAPOINTE, WILLIAM BILLECK, AND ESKE WILLERSLEV Authors Info & Affiliations

SCIENCE ADVANCES · 27 Oct 2021 · Vol 7, Issue 44 · DOI: 10.1126/sciadv.abh2013

A great-grandson of the legendary Lakota Sioux leader Sitting Bull (Tatanka Iyotake), Ernie LaPointe, wished to have their familial relationship confirmed via genetic analysis, in part, to help settle concerns over Sitting Bull's final resting place. To address Ernie LaPointe's claim of family relationship, we obtained minor amounts of genomic data from a small piece of hair from Sitting Bull's scalp lock, which was repatriated in 2007. We then compared these data to genome-wide data from LaPointe and other Lakota Sioux using a new probabilistic approach and concluded that Ernie LaPointe is Sitting Bull's great-grandson. To our knowledge, this is the first published example of a familial relationship between contemporary and a historical individual that has been confirmed using such limited amounts of ancient DNA across such distant relatives. Hence, this study opens the possibility for broadening genealogical research, even when only minor amounts of ancient genetic material are accessible.

- Introgression deserts
- Birch Pitch
- Universal Bias
- Tissues connected to archaeological sources
- Cultural adaptations
- When to use sedaDNA