

HUMAN PALAEOGENOMICS

MUNI
SCI

EVA CHOCHOLOVÁ

LABORATORY OF BIOLOGICAL AND MOLECULAR ANTHROPOLOGY

DEPARTMENT OF EXPERIMENTAL BIOLOGY

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

Current populations

Genomics

Epigenomics

Preservation

Differences

Change over long time

Environmentally responsive, plastic

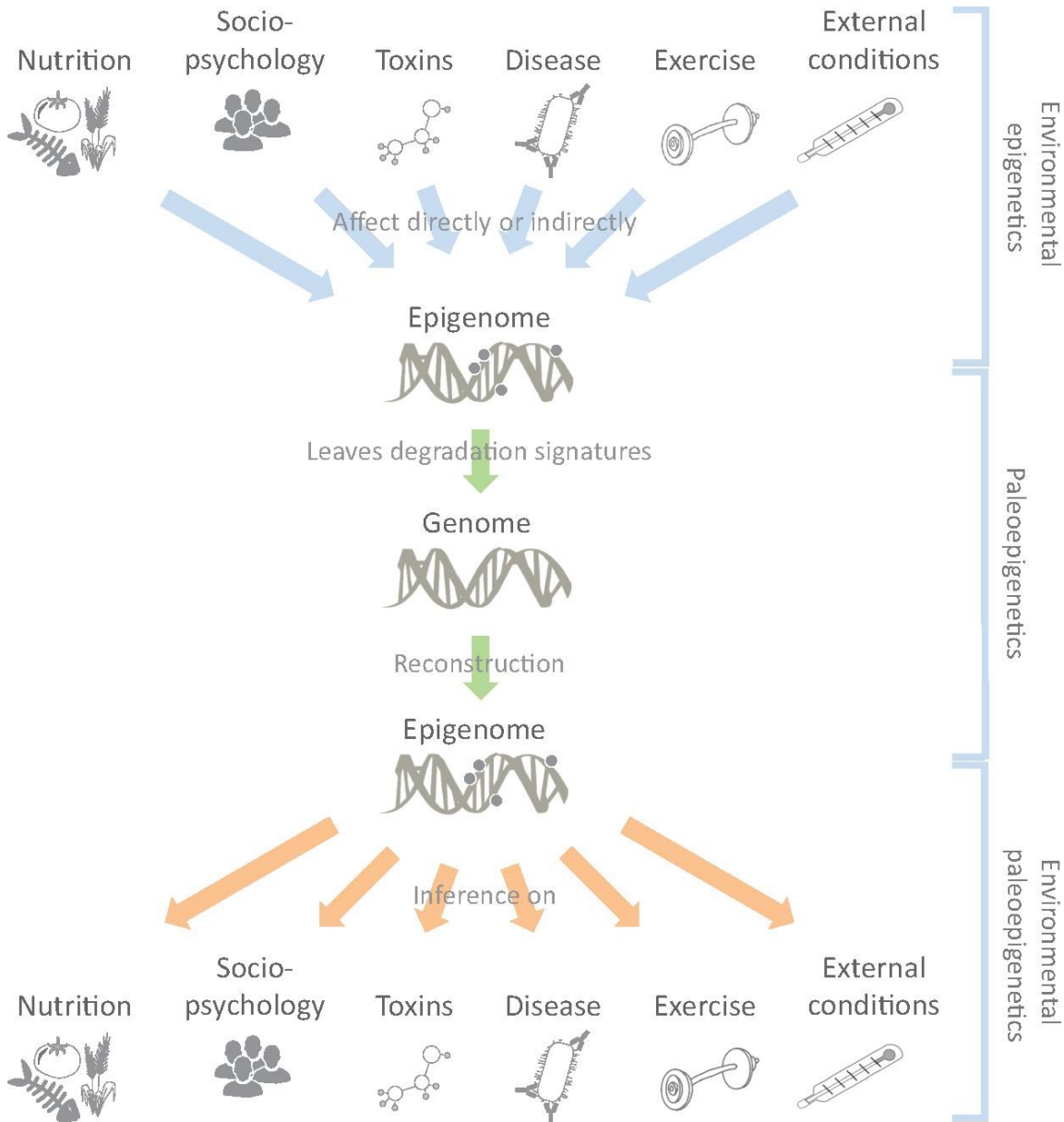
Interpretation



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.jtree.2020.10.005

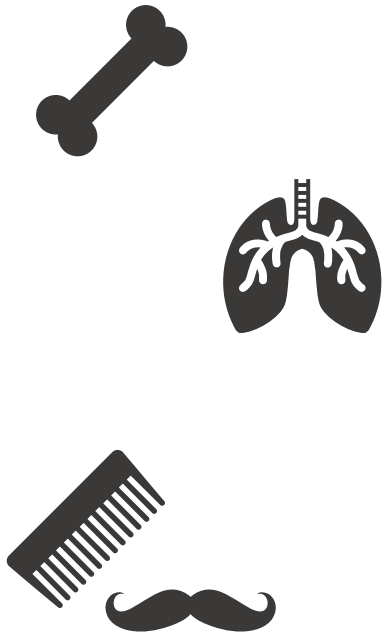
+ many more that were not discussed in detail, such as anthropology, proteomics, isotope analysis, palaeozoology, palaeobotany...



In a study by Dominguez-Salas et al., 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 et al. 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 et al. 2010; Dominguez-Salas et al. 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 et al. 2014).

ANCIENT HUMAN GENOMES

Human tissues



Artefacts

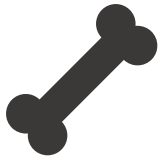


Settlements and environment



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](#), [Harashwaradhana](#), [Maanwendra Singh Bartwal](#), [Sachin Kumar](#), [Subhash Chandra Diyundi](#), [Patrick Roberts](#), [Nicole Boivin](#), [Douglas J. Kennett](#), [Kumarasamy Thangaraj](#), [David Reich](#)  & [Niraj Rai](#)

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[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 sediments

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

[Authors Info & Affiliations](#)

SCIENCE • 27 Apr 2017 • Vol 356, Issue 6338 • pp. 605-608 • DOI: [10.1126/science.aam9695](https://doi.org/10.1126/science.aam9695)

Article | [Open access](#) | [Published: 23 June 2021](#)

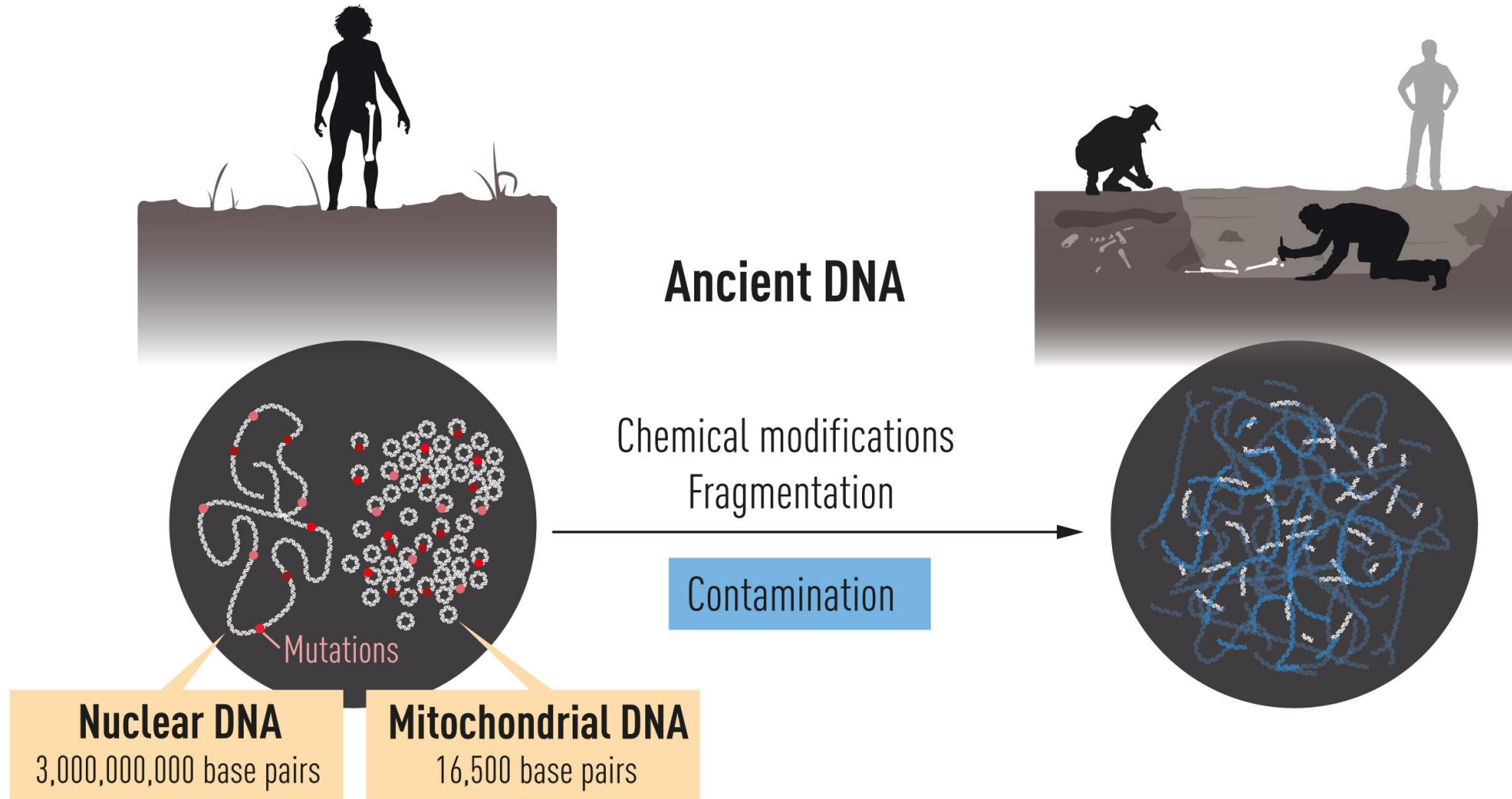
Pleistocene sediment DNA reveals hominin and faunal turnovers at Denisova Cave

[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](#) [✉]

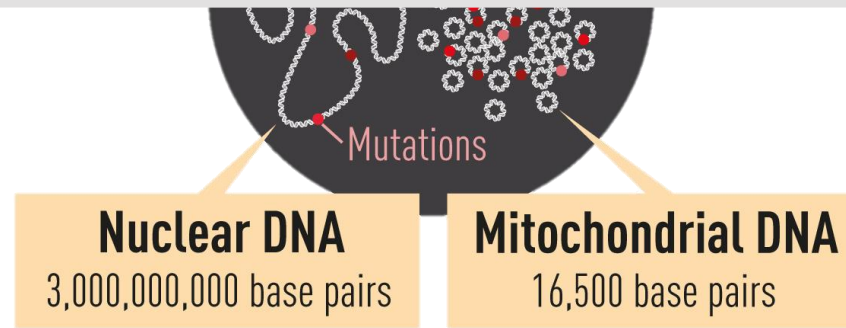
[Nature](#) **595**, 399–403 (2021) | [Cite this article](#)

31k Accesses | **56** Citations | **680** Altmetric | [Metrics](#)

ANCIENT HUMAN GENOMES



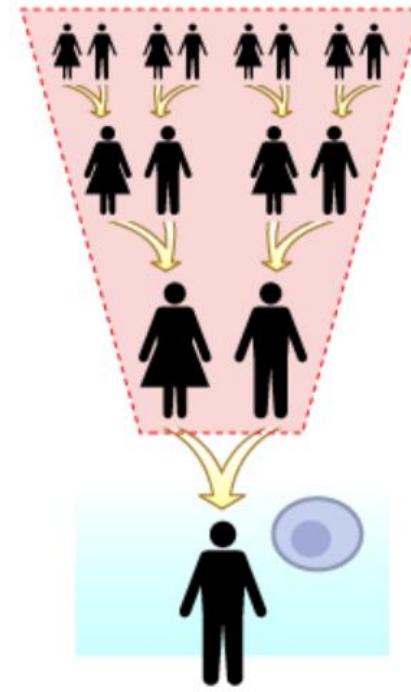
ANCIENT HUMAN GENOMES



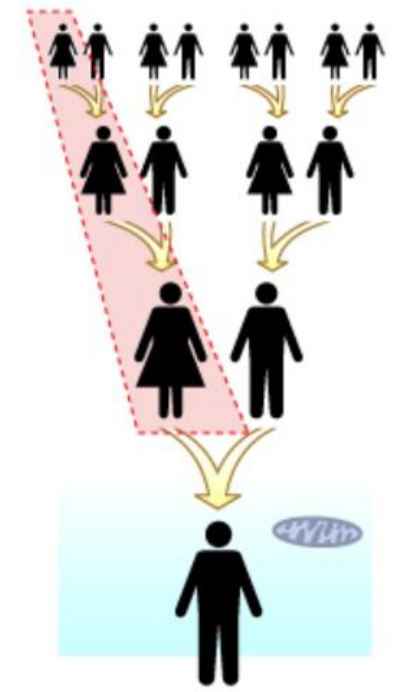
- Two copies
- Long
- Recombining
- Inherited from both parents
- Great amount of information

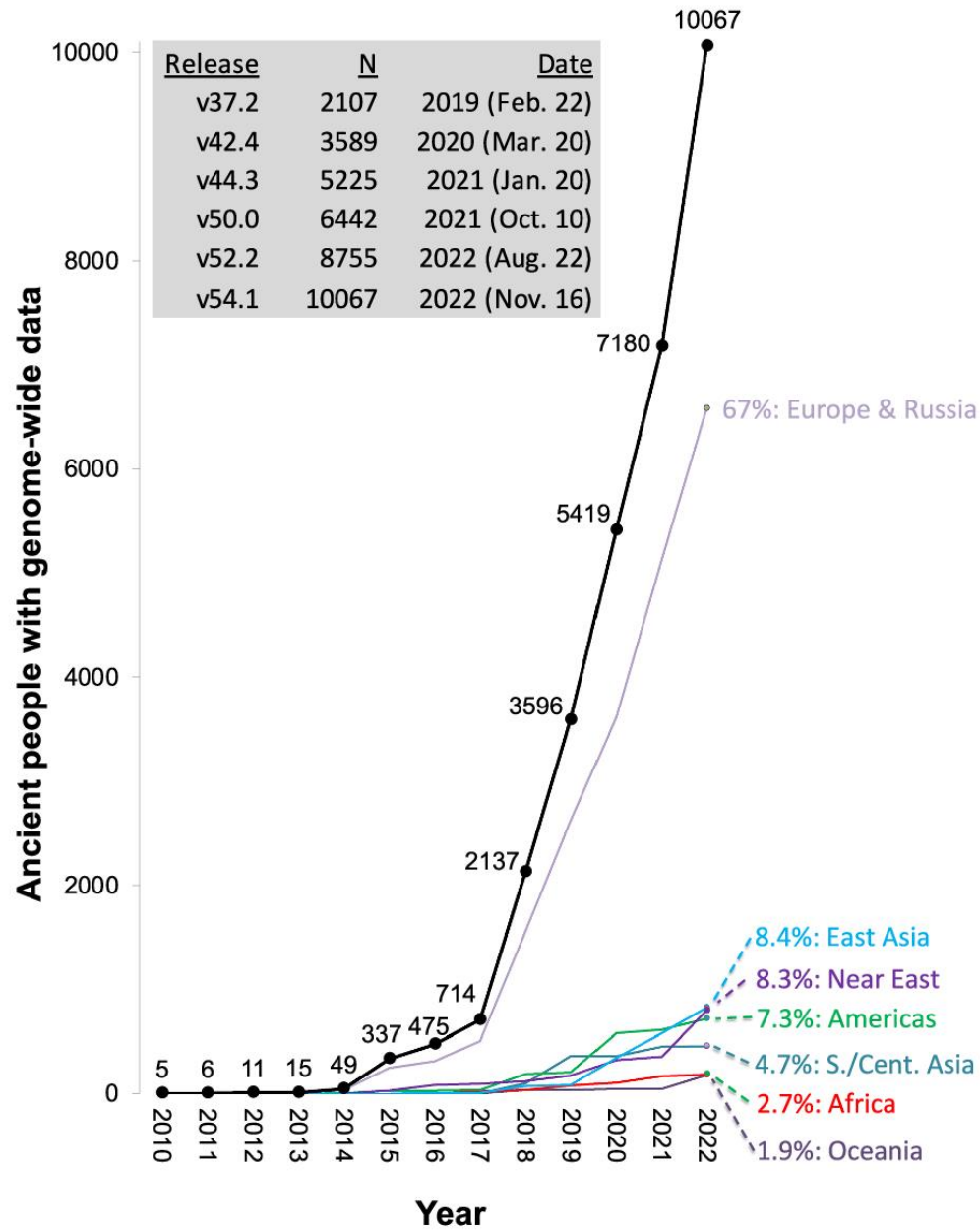
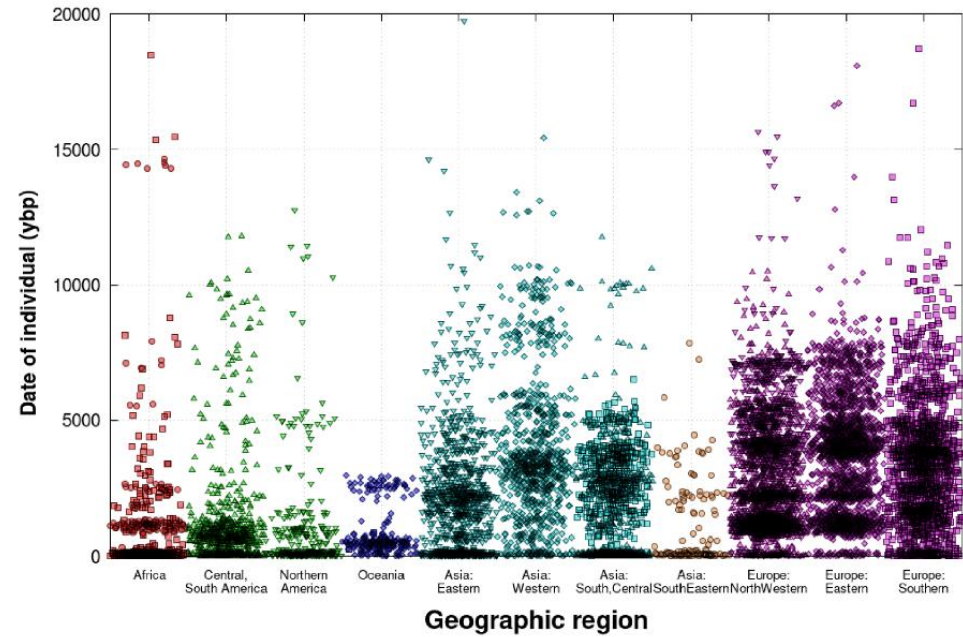
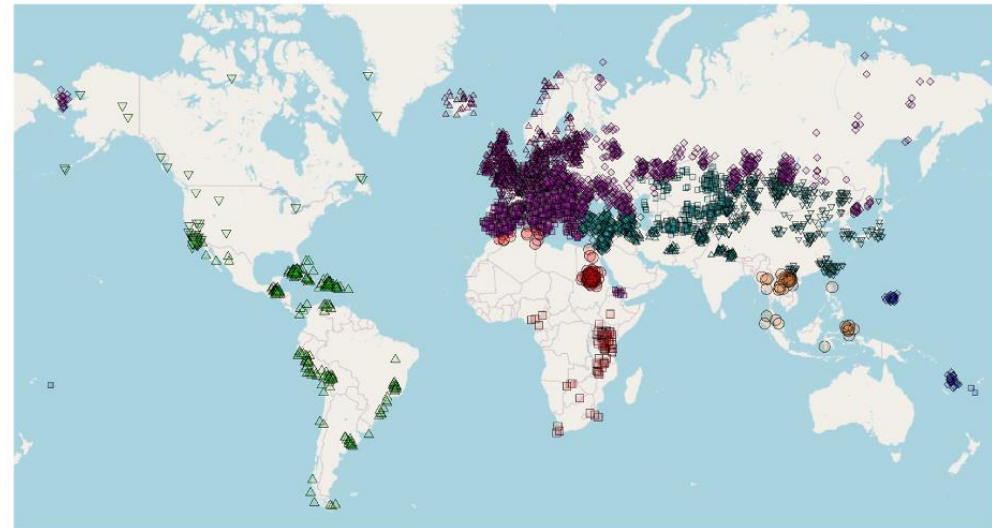
- More copies per cell
- Shorter, circular
- Generally without recombination
- Maternal
- Limited insight

Nuclear DNA is inherited from all ancestors.



Mitochondrial DNA is inherited from a single lineage.




A**B****C**

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).

Letter | [Published: 14 March 2016](#)

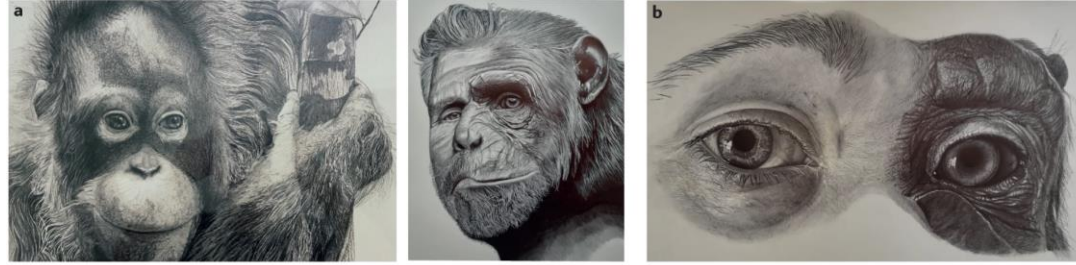
Nuclear DNA sequences from the Middle Pleistocene Sima de los Huesos hominins

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

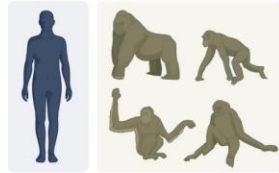
[Nature](#) **531**, 504–507 (2016) | [Cite this article](#)

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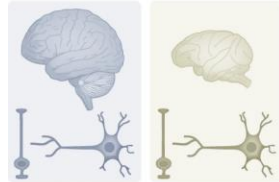
430,000 years ago



c Human traits

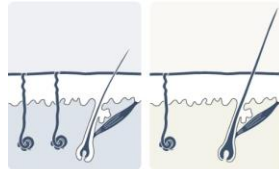


Big brain
Cortex expansion, neuron numbers and synapse density enhance cognition



Differences in neurogenesis and neuron maturation

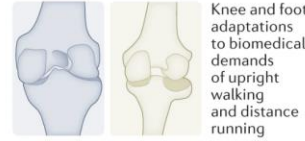
Naked ape
Decreased hair thickness and increased eccrine gland density impacts thermoregulation



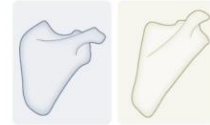
White eyes
Pigmentation patterns of the visible part of the eyeball, around the iris and sclera, has links to sexual selection and potentially social behaviour



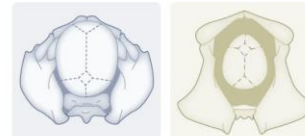
Upright walking and projectile throwing



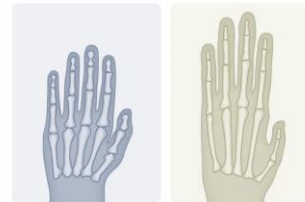
Knee and foot adaptations to biomedical demands of upright walking and distance running



Shoulder modifications increase rotational velocity

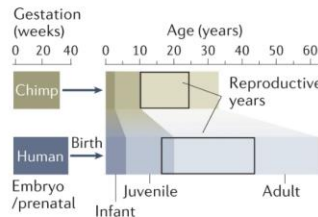


Pelvis modifications support upright walking and cranium size during childbirth

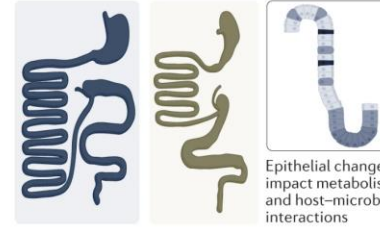


Relatively long thumb and short fingers enhances object grasping

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

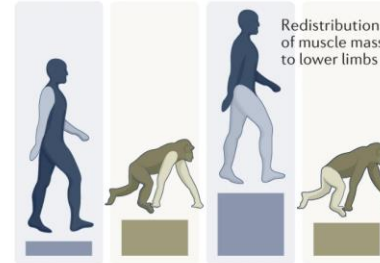


Diet and cooking
Increased small intestine to colon ratio impacts nutrient digestion/absorption

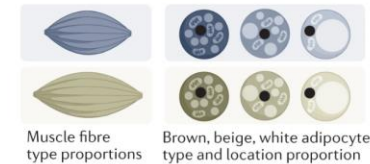


Epithelial changes impact metabolism and host-microbe interactions

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



Redistribution of muscle mass to lower limbs



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



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



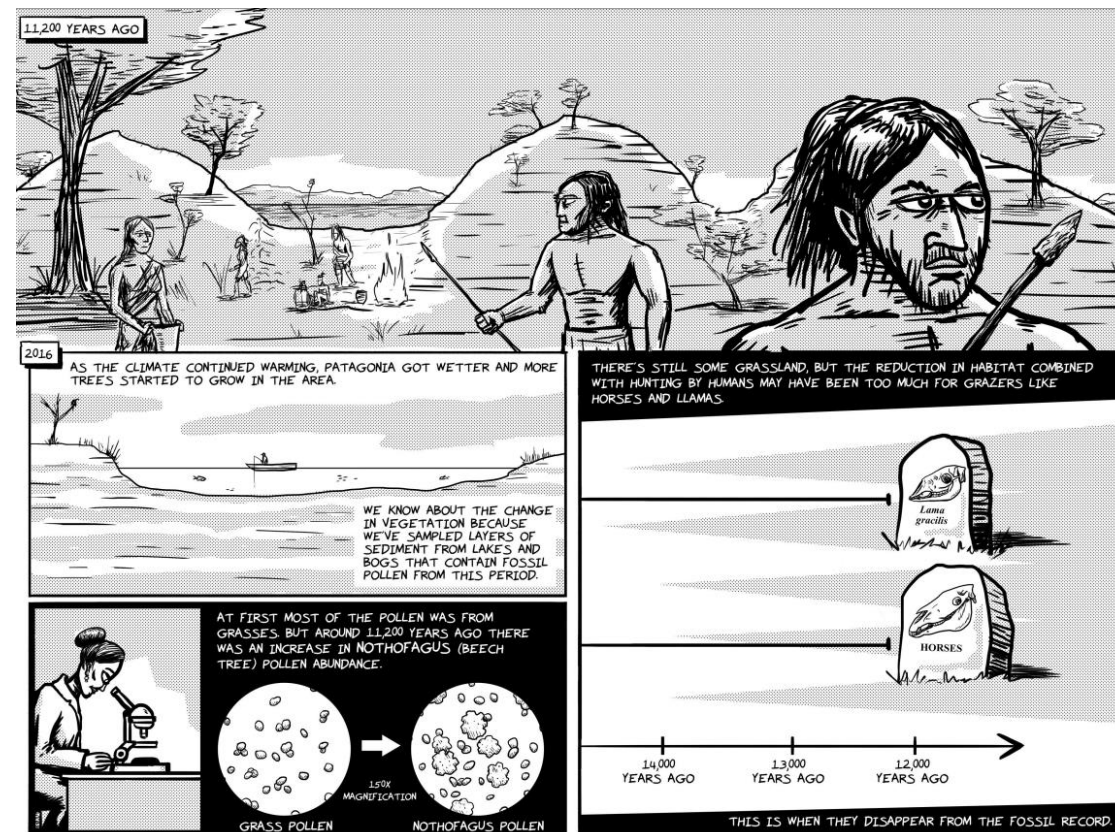
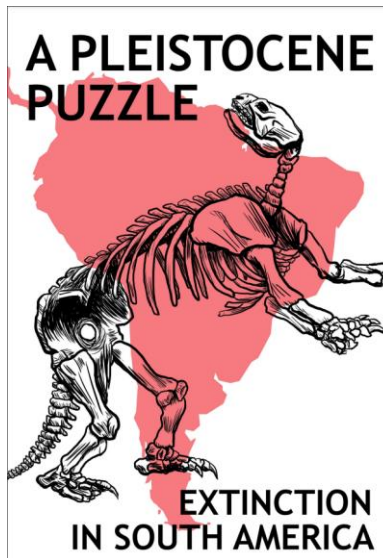
Speciation
How new species arise



Macroevolution
Evolution above the species level



The big issues
Pacing, diversity, complexity, and trends



NEANDERTHAL AND DENISOVAN DNA

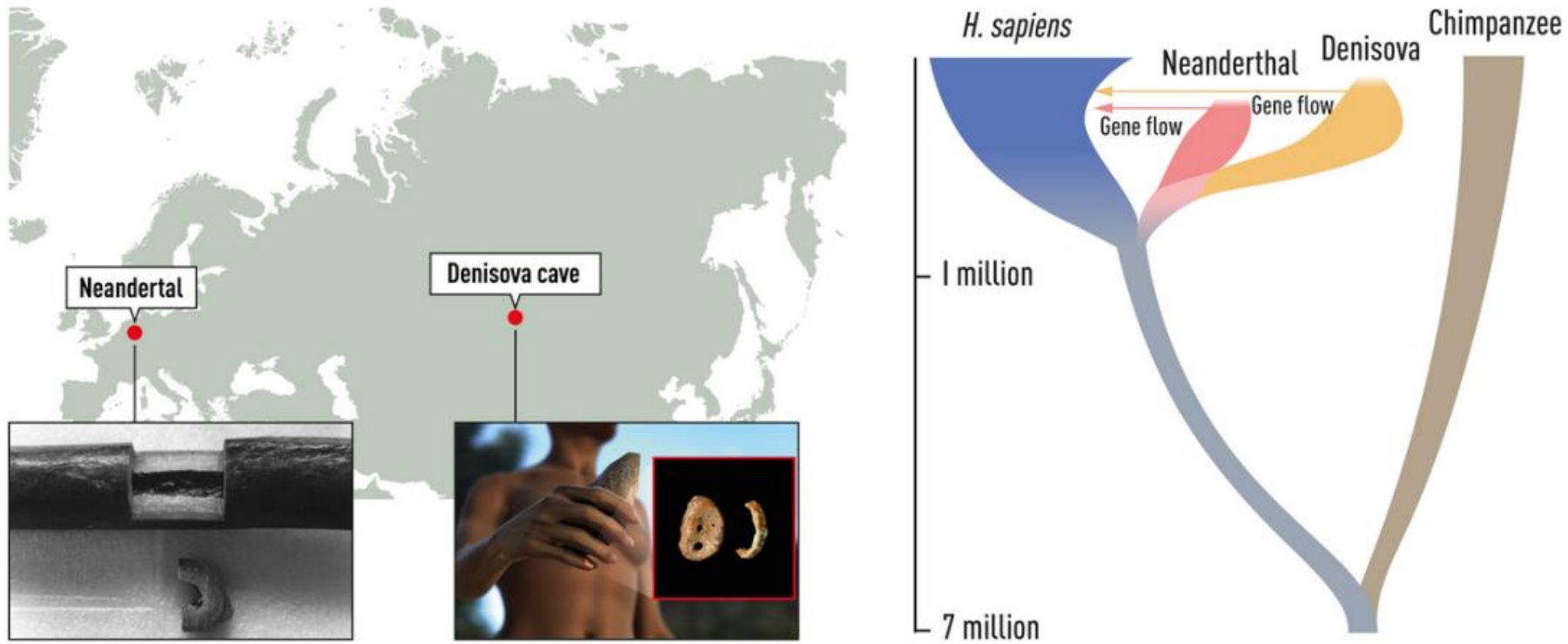


Figure 2. A. Pääbo extracted DNA from bone specimens from extinct hominins. He first obtained a bone fragment from Neanderthal in Germany, the site that gave name to the Neanderthals. Later, he used a finger bone from the Denisova Cave in southern Siberia, the site that gave name to Denisovans. B. Phylogenetic 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

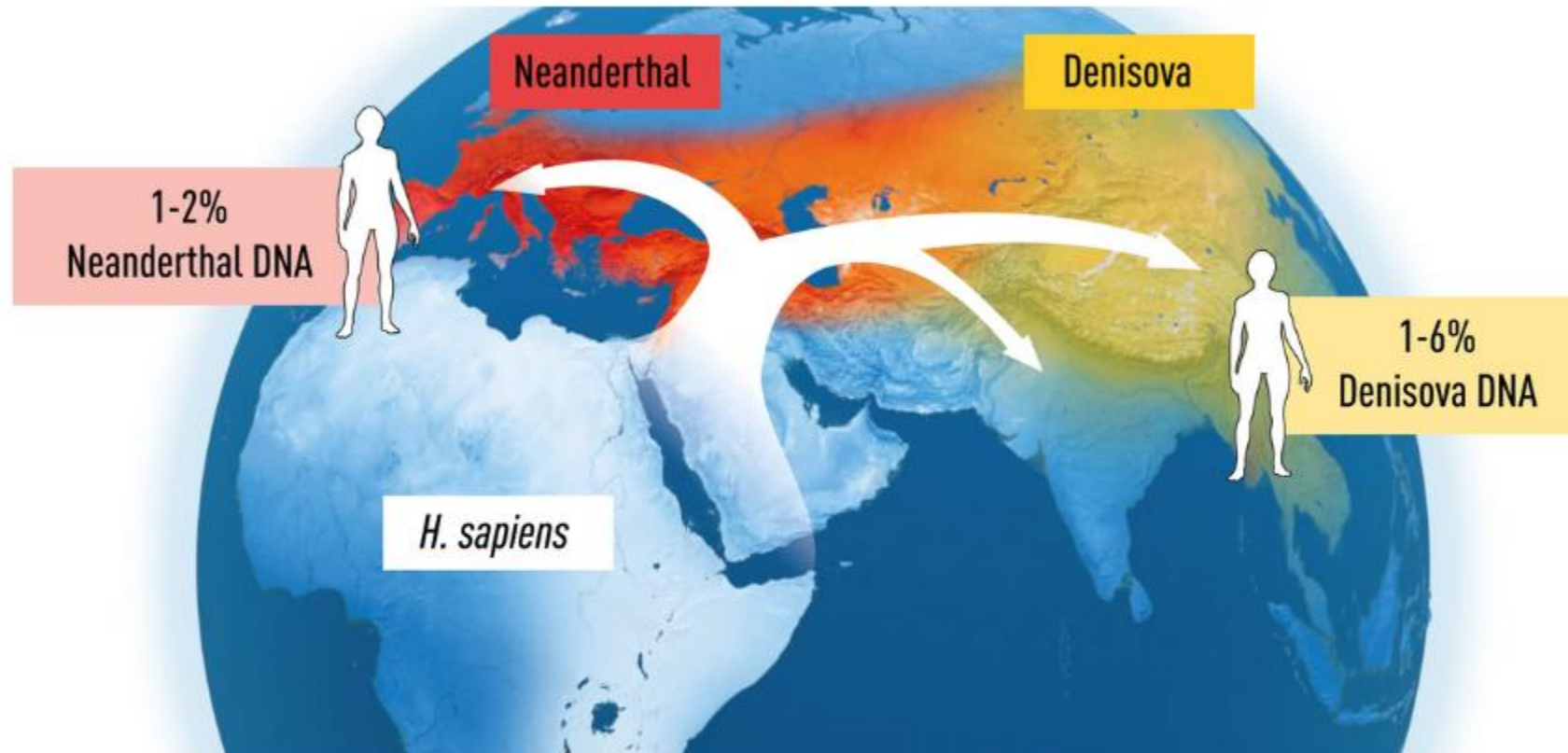
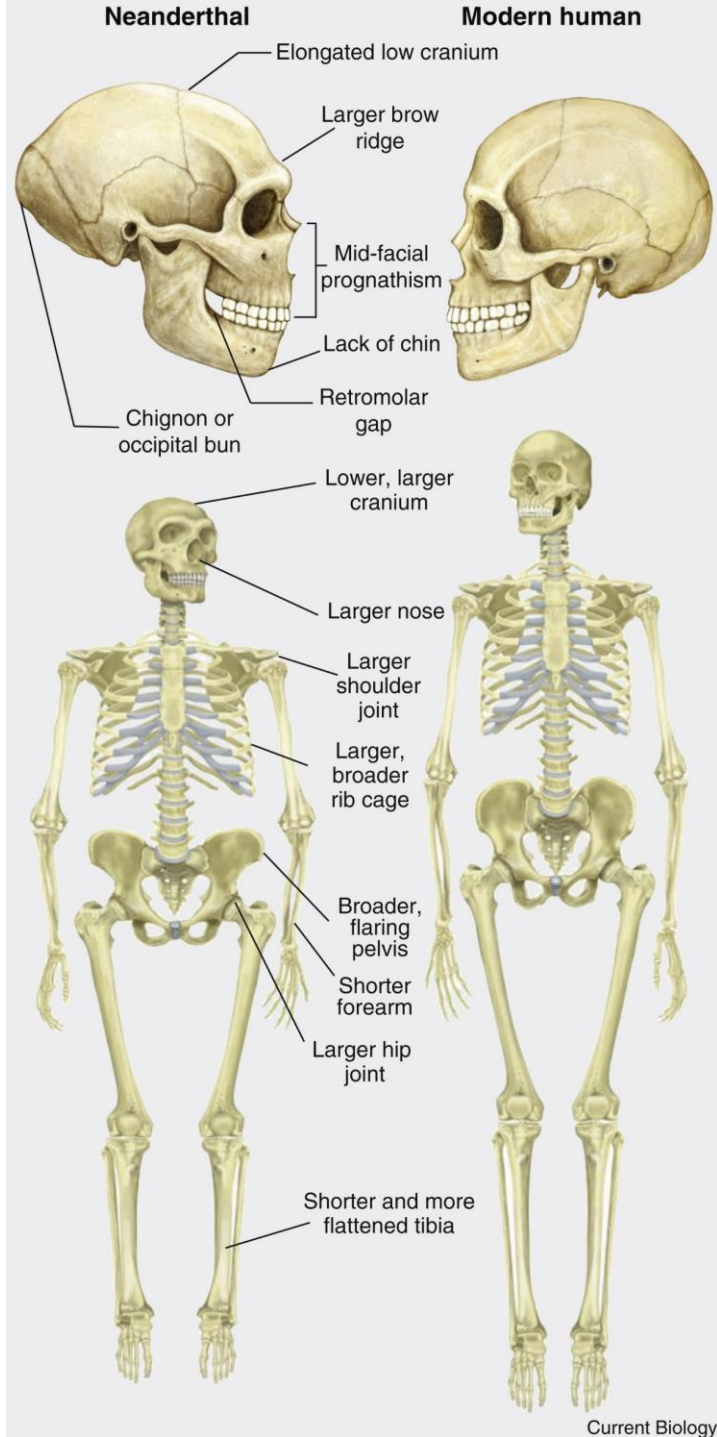
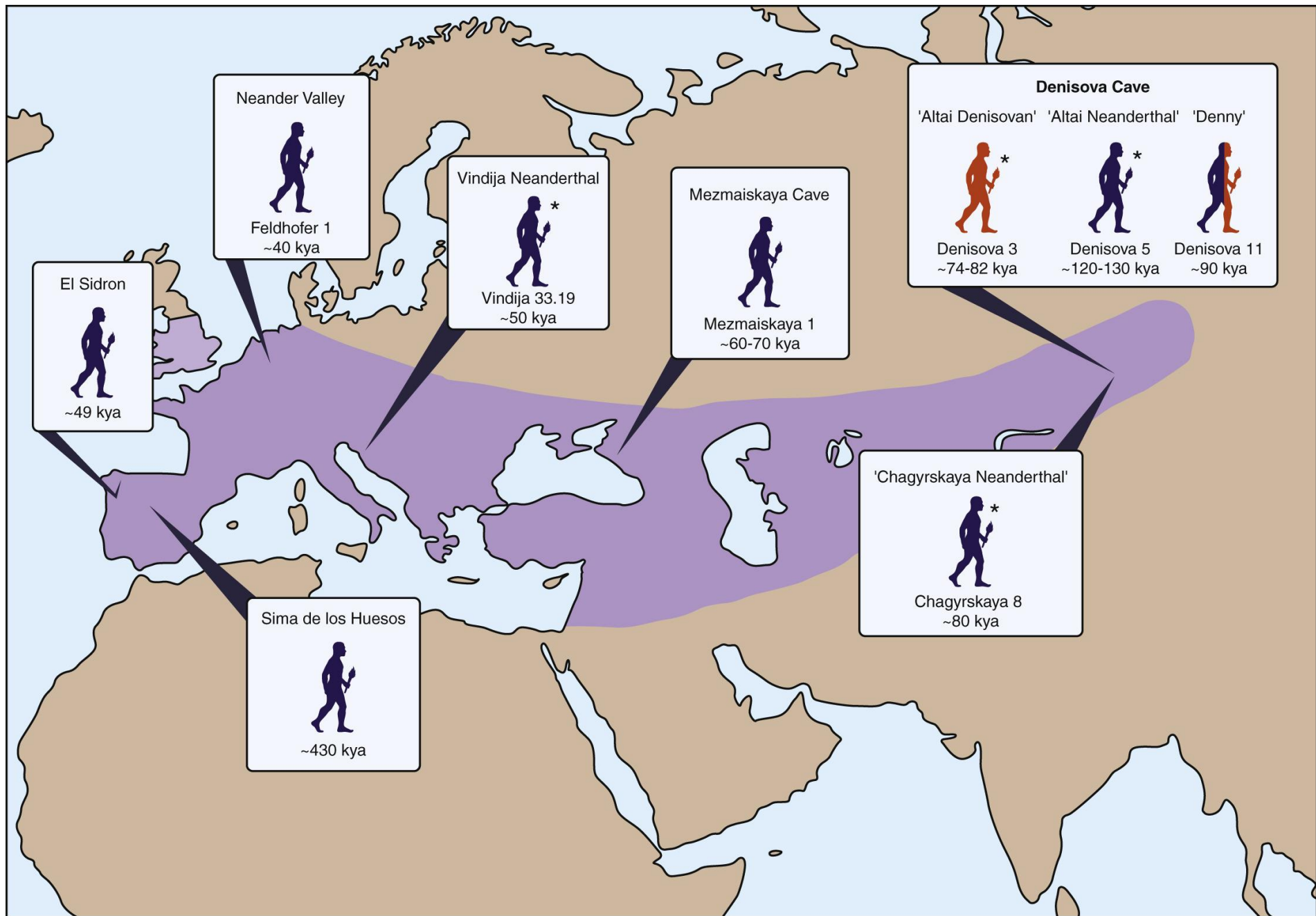


Figure 3. Pääbo's discoveries have provided important information on how the world was populated at the time when *Homo sapiens* migrated out of Africa and spread to the rest of the world. Neanderthals lived in the west and Denisovans in the east on the Eurasian continent. Interbreeding occurred when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





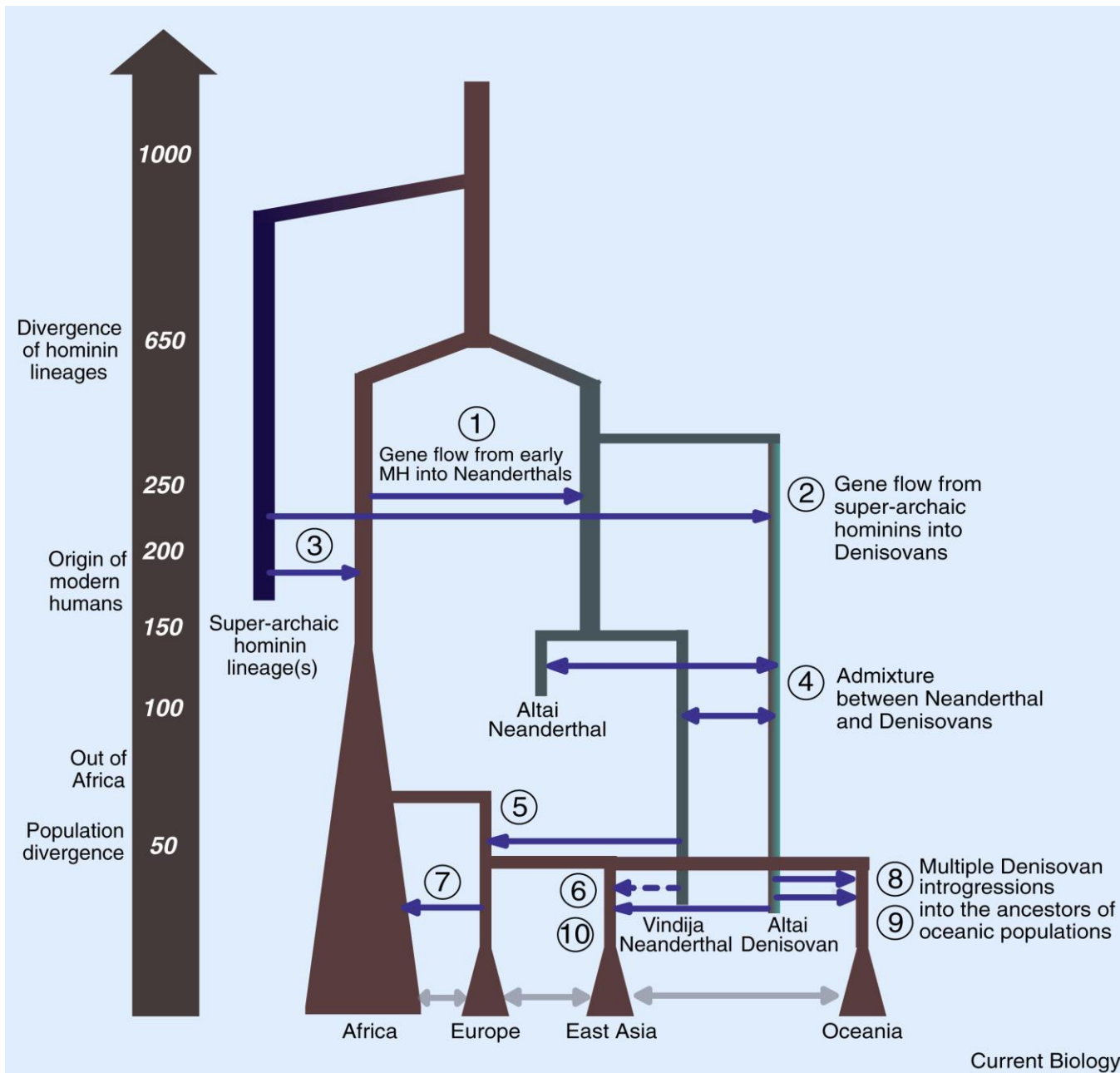




Figure 2. Simplified demographic model illustrating inferred population relationships and admixture events among modern humans and archaic hominins.

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 father²⁶; (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 Africa³⁴; and multiple Denisovan introgressions into the ancestors of (8,9) Oceanians^{9, 10, 11,13,18,45} and (10) East Asians¹². For simplicity we represent the multiple, distinct introgressing Denisovan-like lineages as arising from a single Denisovan source (adapted with permission from¹⁵⁴ © 2017 Springer Nature).

NEANDERTHAL AND DENISOVAN DNA

Letter | [Published: 22 August 2018](#)

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

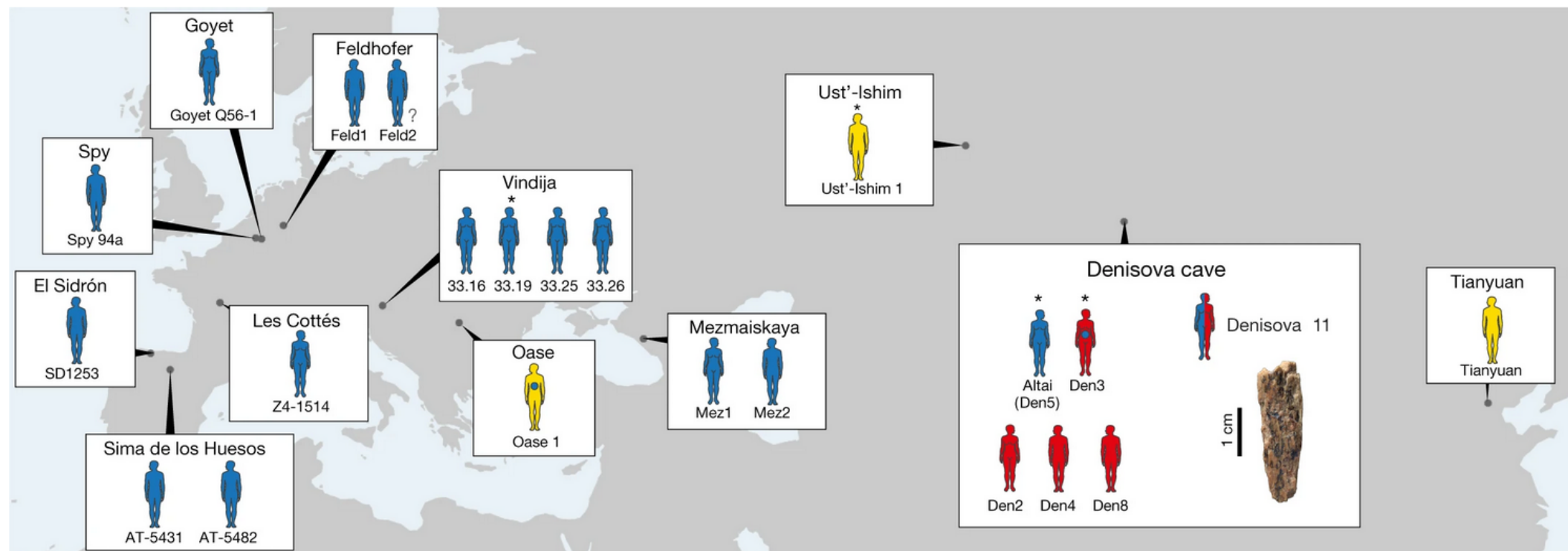
[Viviane Slon](#) , [Fabrizio Mafessoni](#), [Benjamin Vernot](#), [Cesare de Filippo](#), [Steffi Grote](#), [Bence Viola](#), [Mateja Hajdinjak](#), [Stéphane Peyrégne](#), [Sarah Nagel](#), [Samantha Brown](#), [Katerina Douka](#), [Tom Higham](#), [Maxim B. Kozlikin](#), [Michael V. Shunkov](#), [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](#).

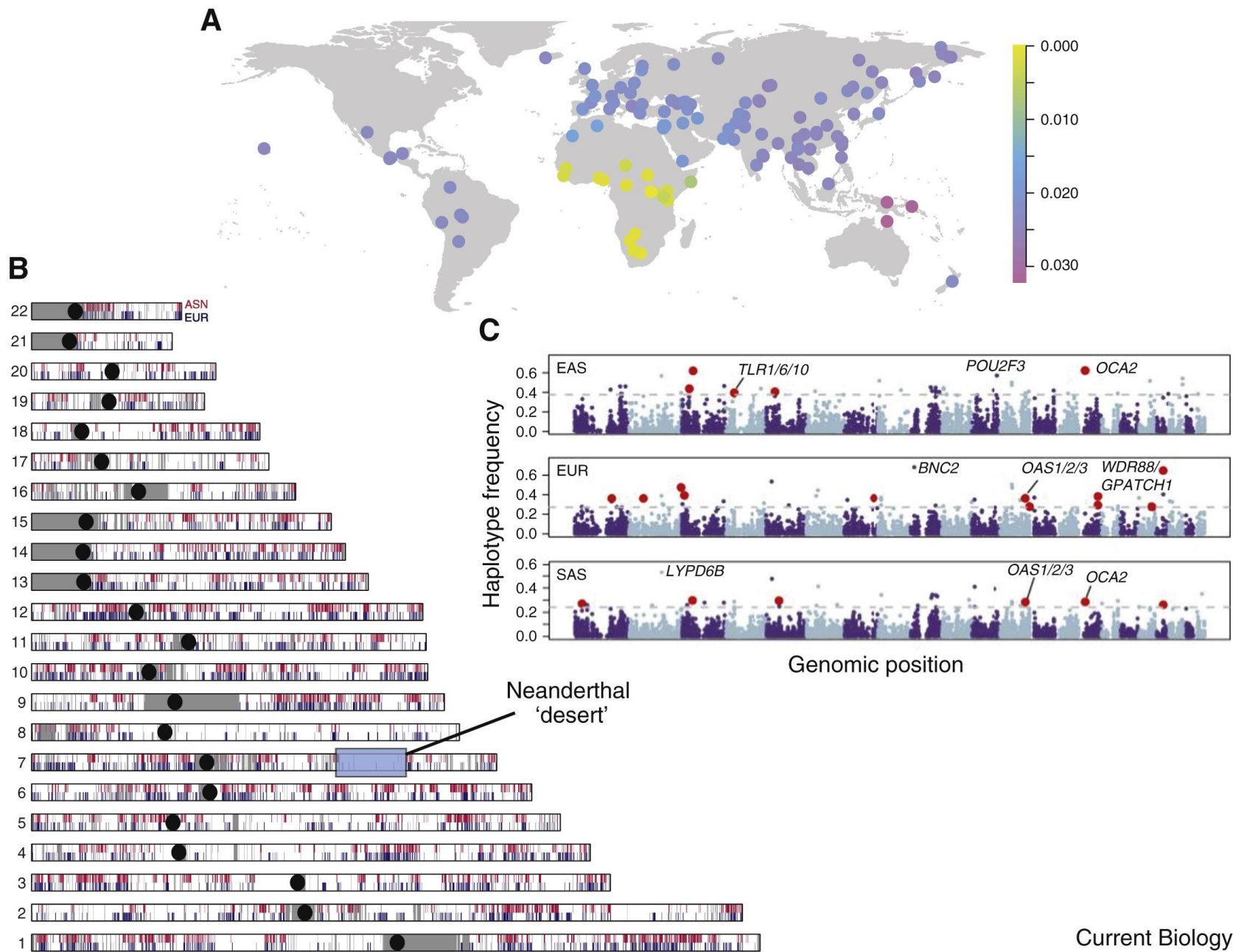


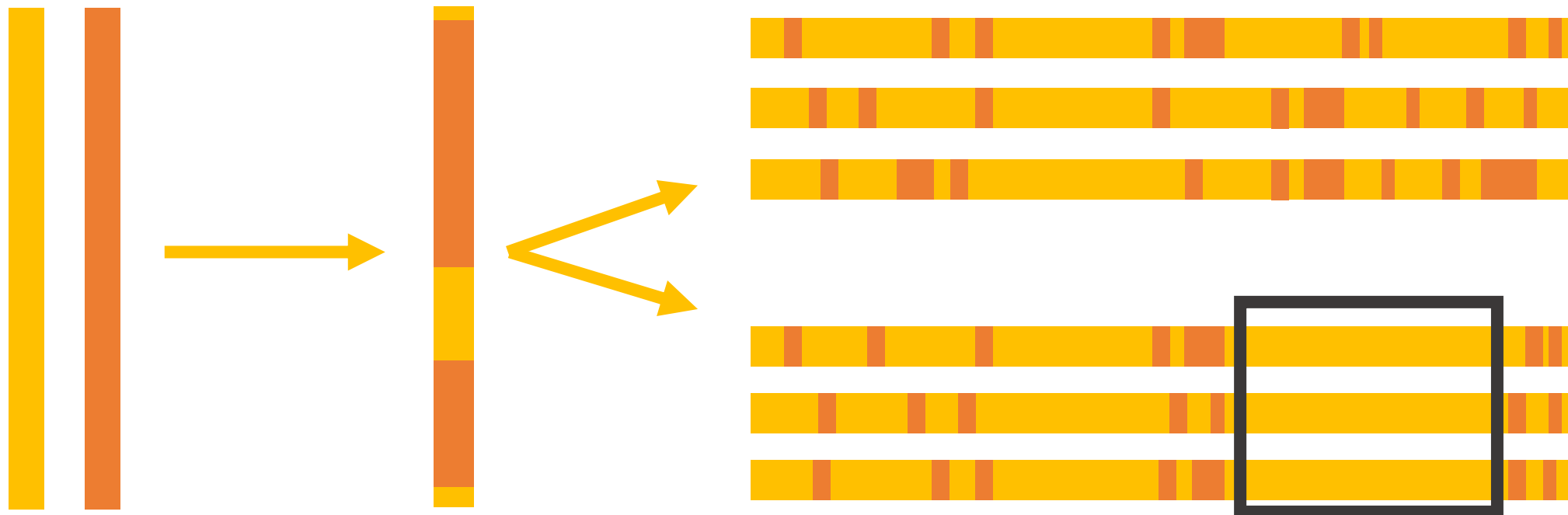
Figure 3. The geographic and genomic landscape of Neanderthal introgression.

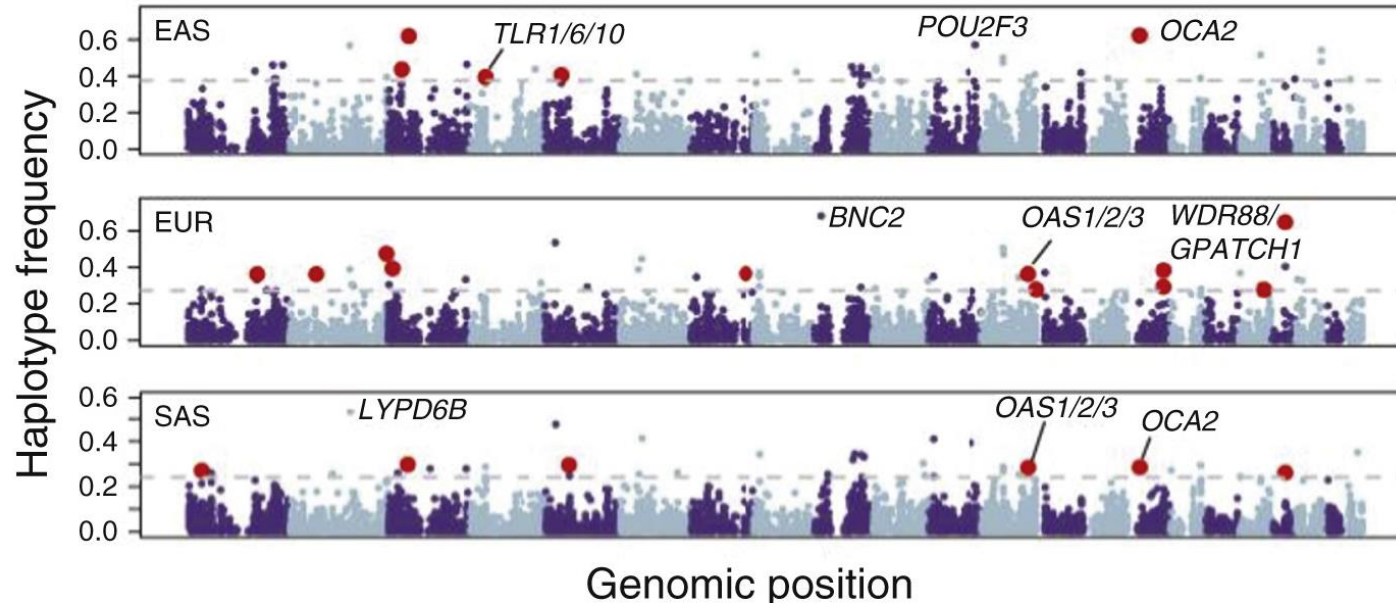
(A) Proportion of Neanderthal ancestry in geographically diverse populations (reproduced with permission from¹⁶ © 2017 American Association for the Advancement of Science). (B) Neanderthal introgressed genomic regions are depicted by colored ticks throughout the genomes of East Asians (red) and Europeans (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 from²⁸ © 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.

Current Biology

NEANDERTHAL AND DENISOVAN DNA

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



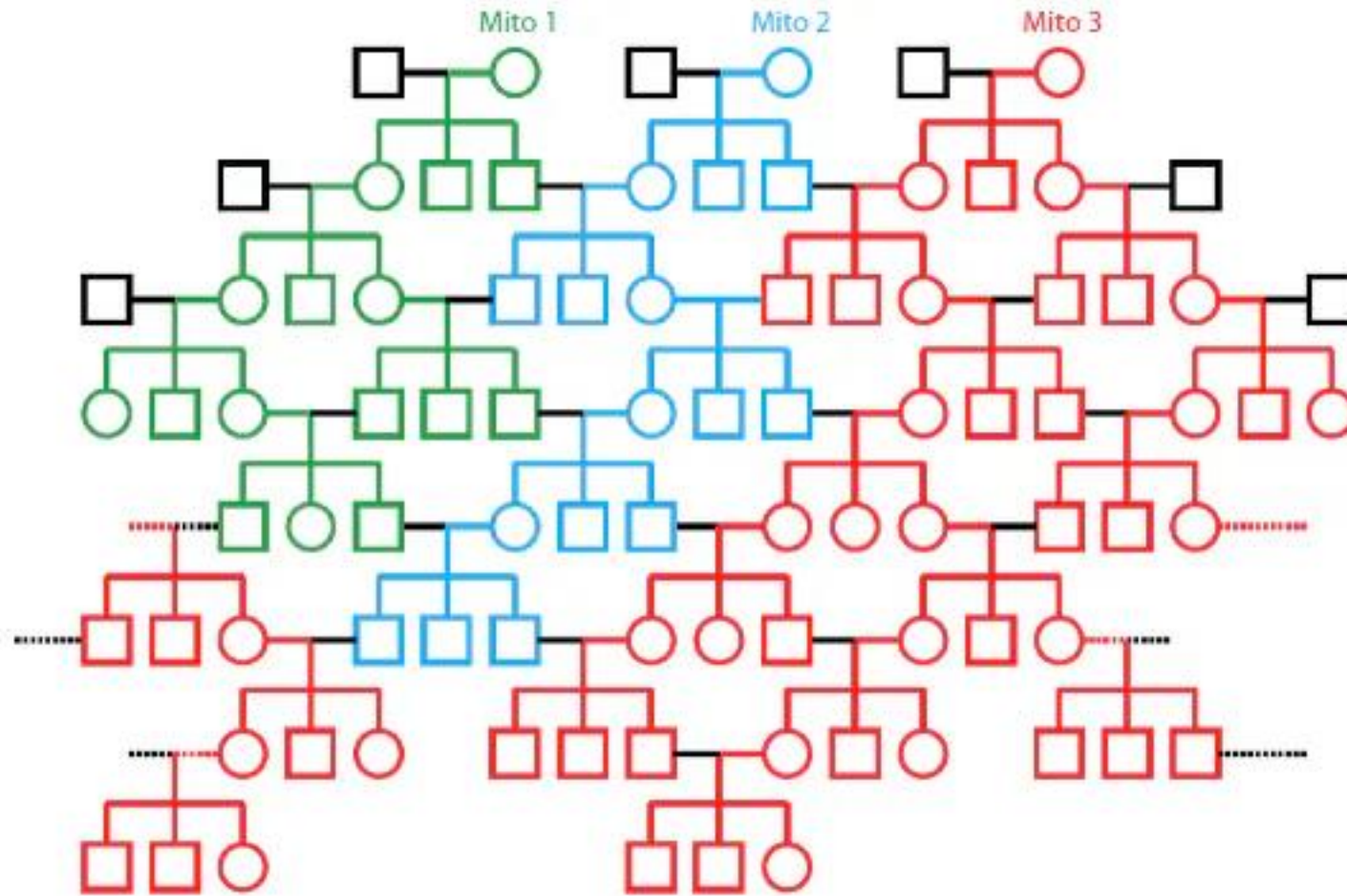
C

- ✗ *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

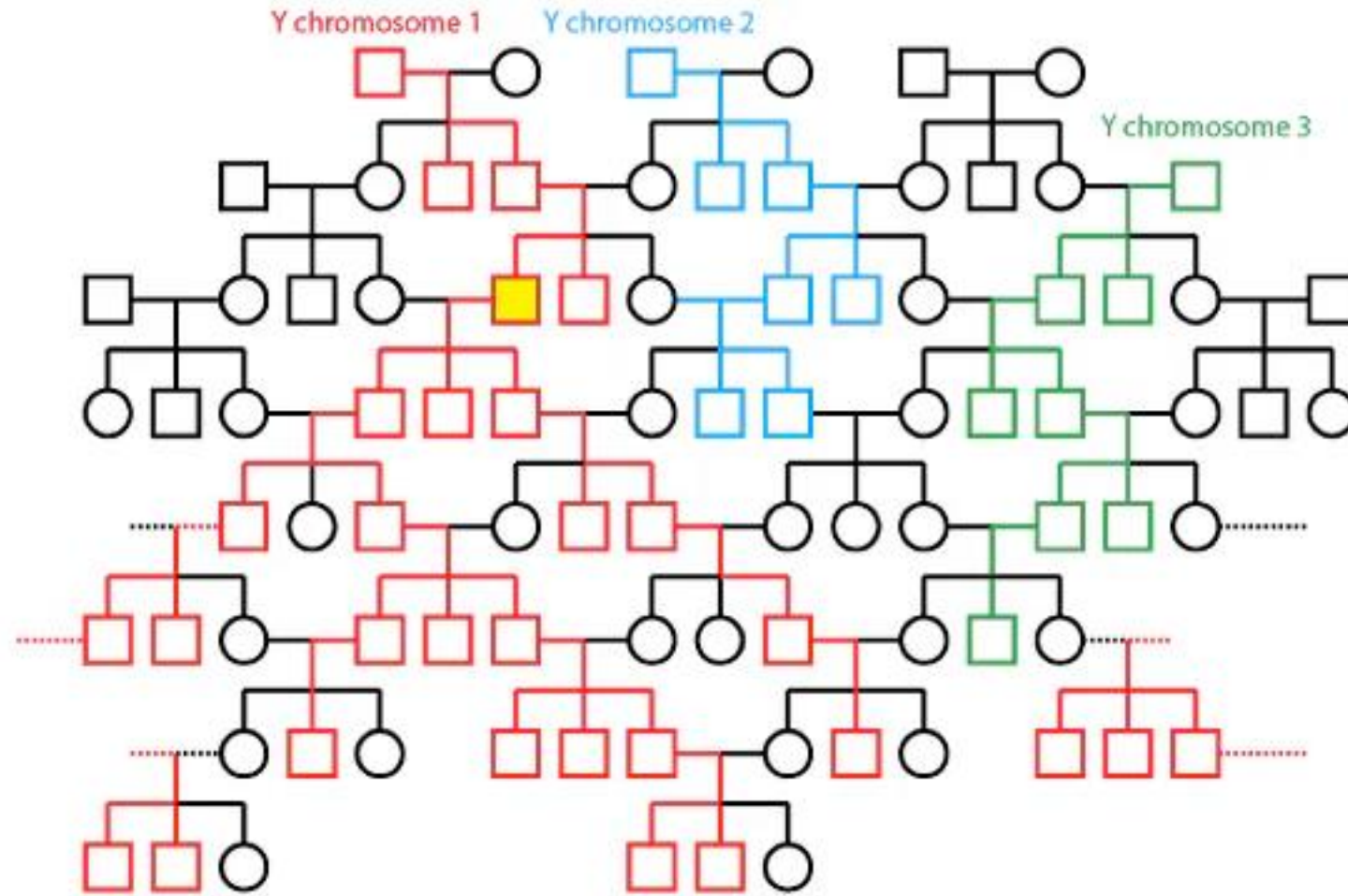
MIGRATION AND CONTACT

- Mitochondrial Eve



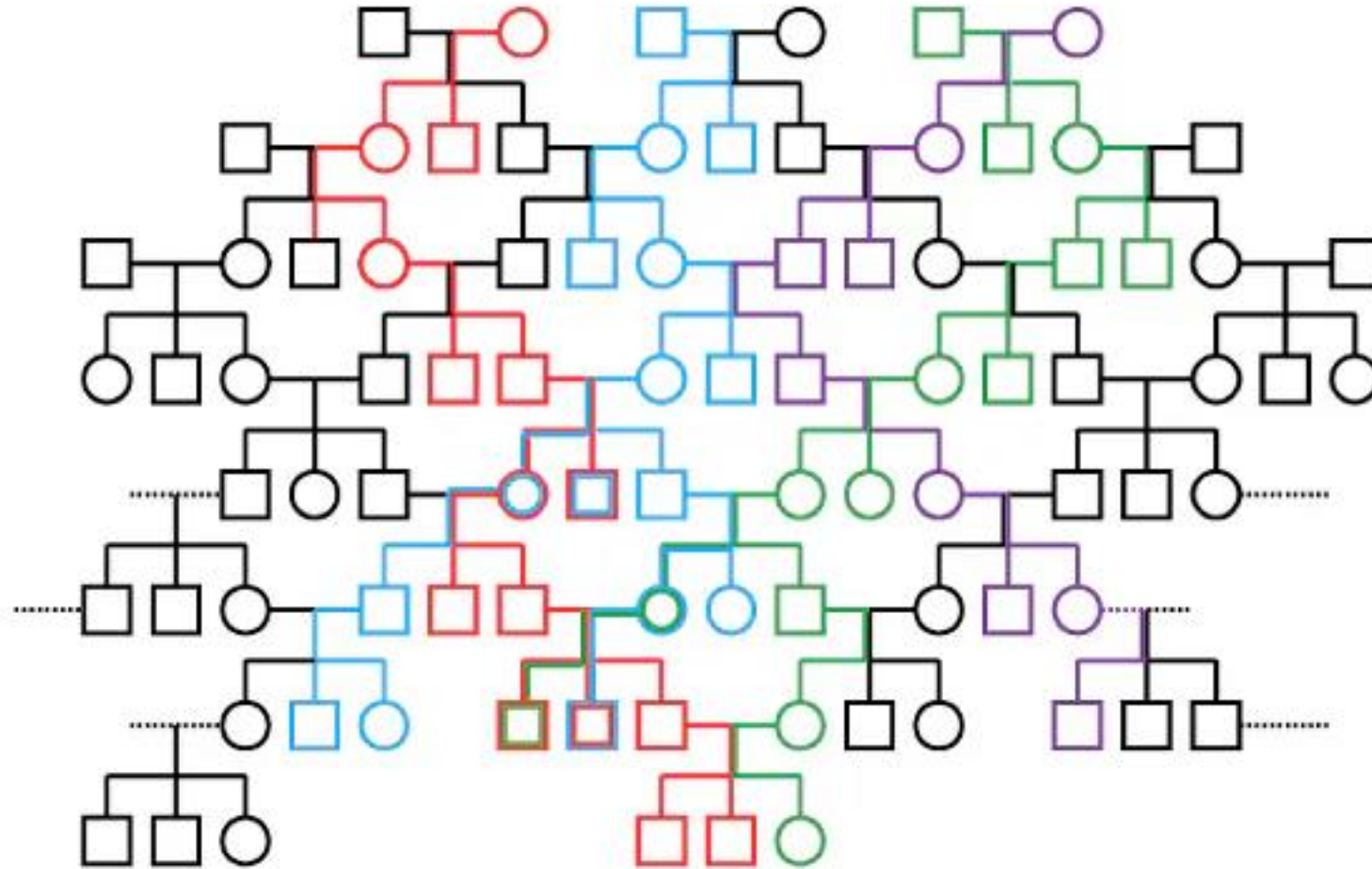
MIGRATION AND CONTACT

- Y-chromosome Adam



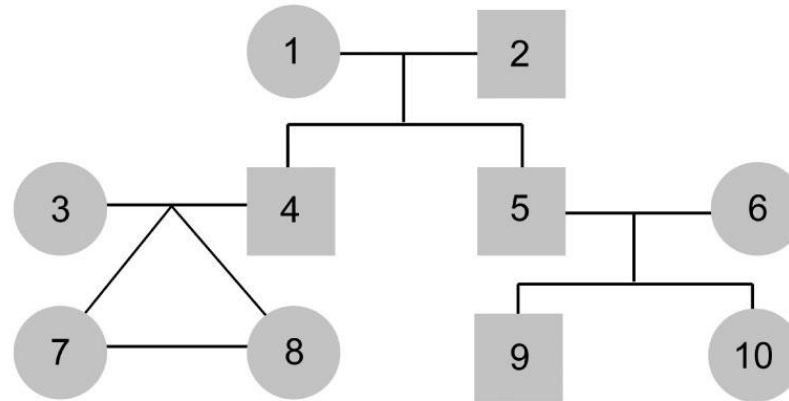
MIGRATION AND CONTACT

- Autosomes



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

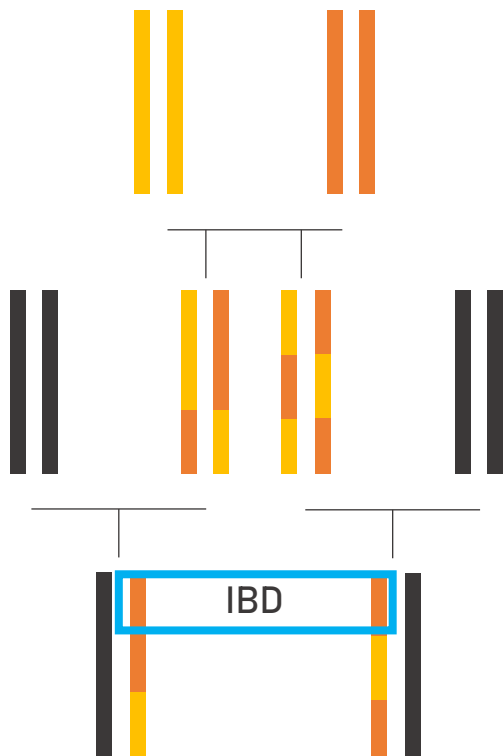
FIGURE 1. Pedigree showing the degrees of relationship between selected pairs of individuals and corresponding inbreeding coefficients (ϕ) and k_0 , k_1 , k_2 values describing the probabilities of a given pair of individuals of sharing, respectively zero, one or two alleles that are identical-by-descent.

MIGRATION AND CONTACT

- Kinship

IBD = identical by descent (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

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Kinship Determination in Archeological Contexts Through DNA Analysis



Stefania Vai^{1*}



Carlos Eduardo G. Amorim²



Martina Lari¹



David Caramelli¹

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.](#)

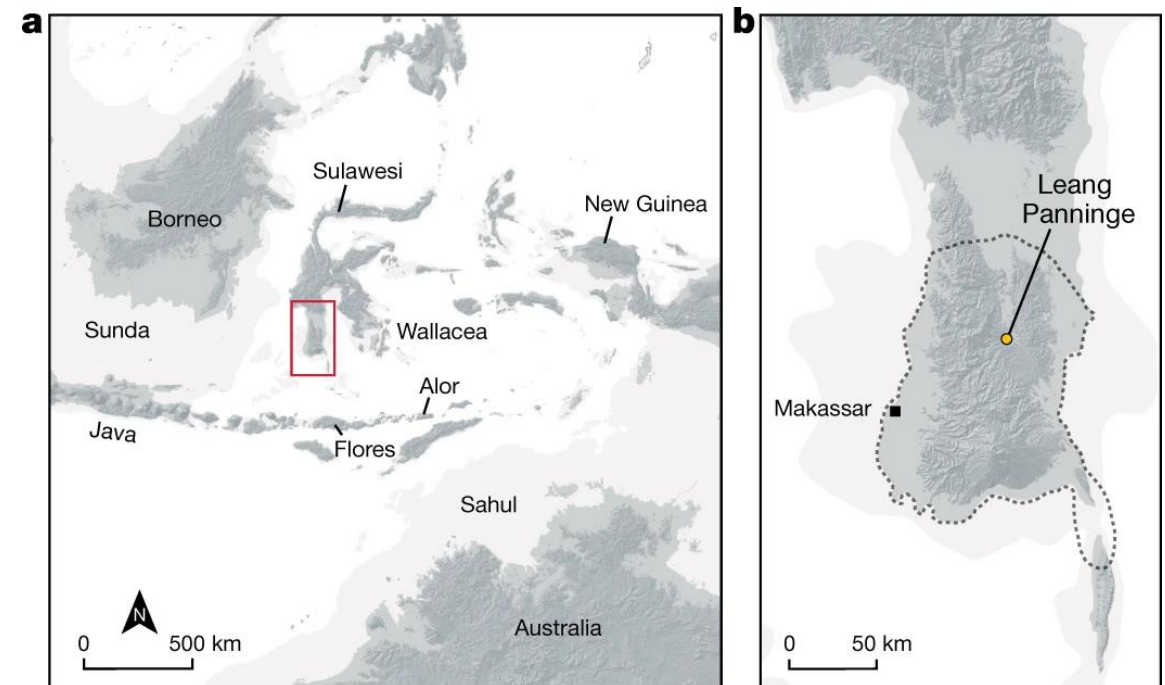
[Perston](#), [Kim Newman](#), [Andi Muhammad Saiful](#), [Marlon Ririmasse](#), [Stephen Chia](#), [Hasanuddin](#), [Dwia Aries](#)

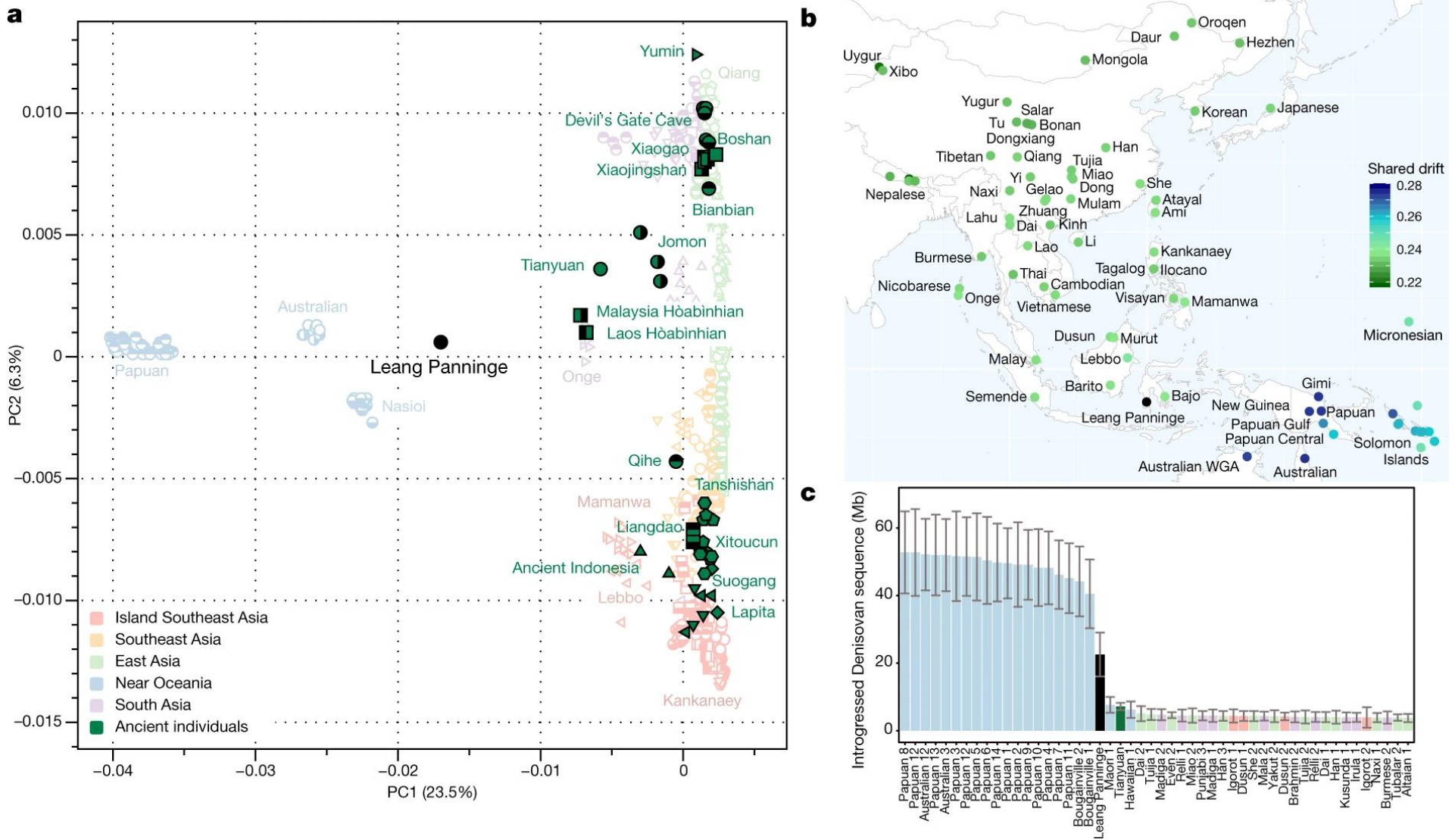
[Tina Pulubuhu](#), [Suryatman](#), [Supriadi](#), [Choongwon Jeong](#), [Benjamin M. Peter](#), [Kay Prüfer](#), ... [Adam Brumm](#) 

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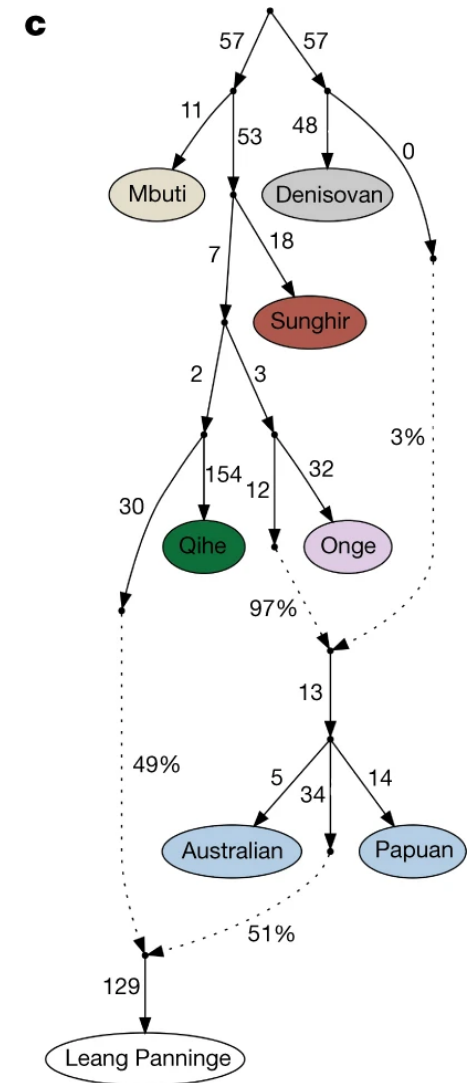
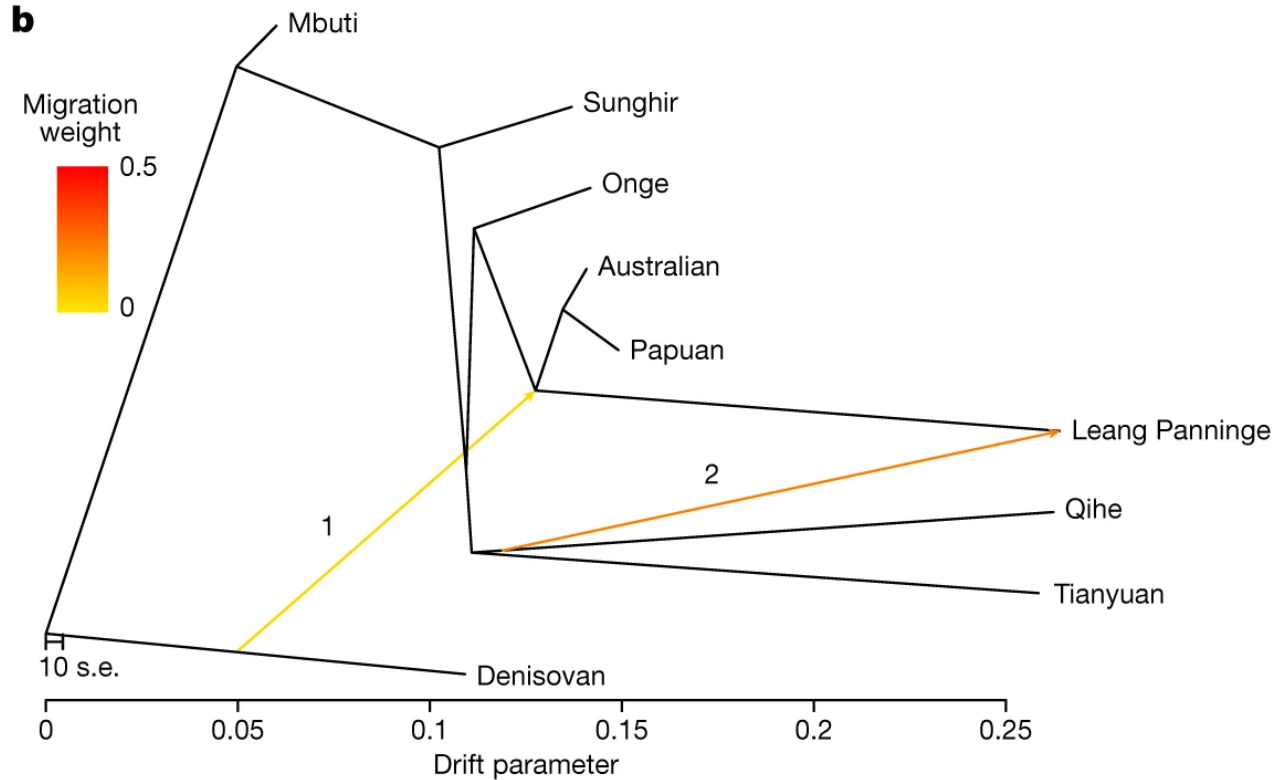
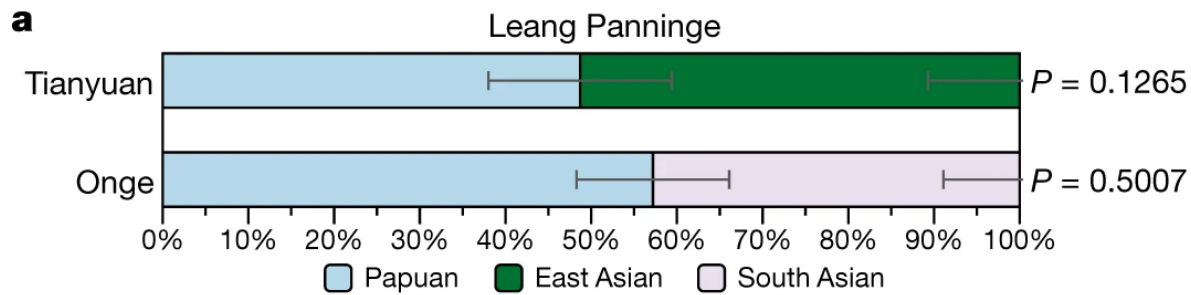
[Nature](#) **596**, 543–547 (2021) | [Cite this article](#)

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).



a, Admixture proportions modelling Leang Panninge as a combination of Papuan⁴⁹ and Tianyuan³⁸ or Onge⁴⁹ groups as estimated by qpAdm³³ using Mbuti, Denisovan³⁹, 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 jackknife in the qpAdm software. **b, c**, Admixture graphs placing Leang Panninge on the branch with the present-day Near Oceanian clade⁴⁴ and showing the admixture with a deep Asian-related ancestry in TreeMix⁴² (**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.

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 ago¹ 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.

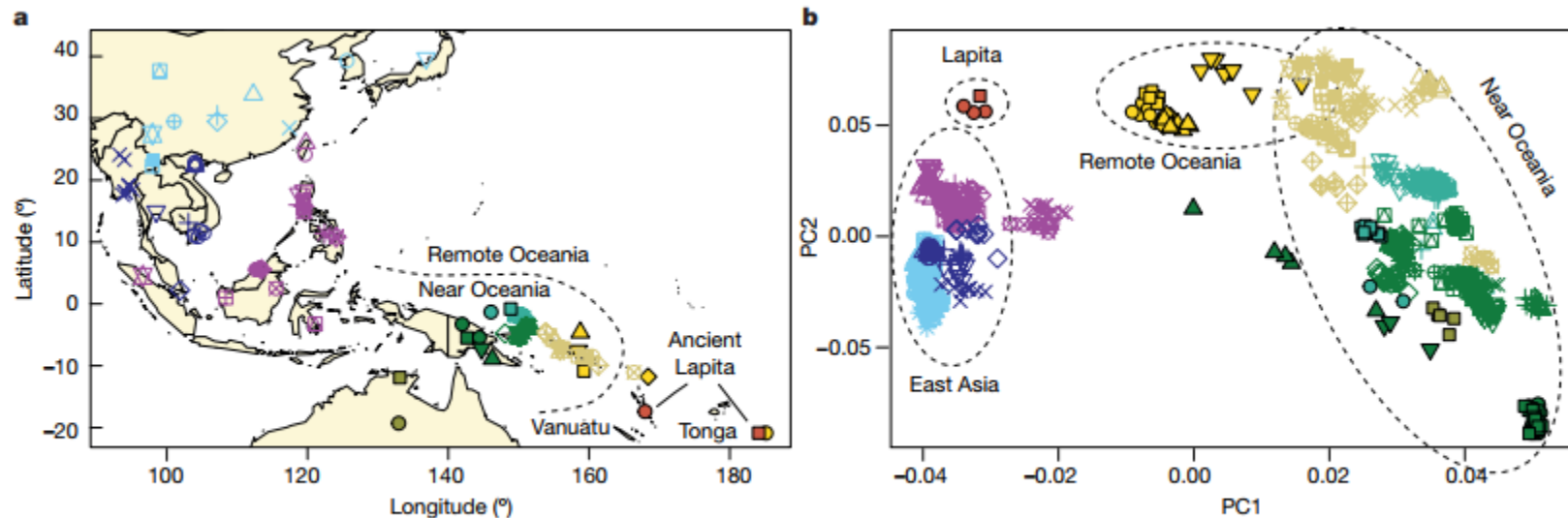


Figure 1 | Data from ancient and present-day populations. **a**, Locations of 778 present-day individuals genotyped on the Affymetrix Human Origins Array and 4 ancient individuals (red symbols). **b**, Ancient individuals projected onto principal components (PC) 1 and 2 computed using only present-day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

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](#)

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> [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

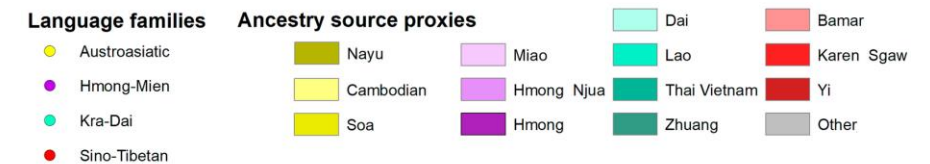
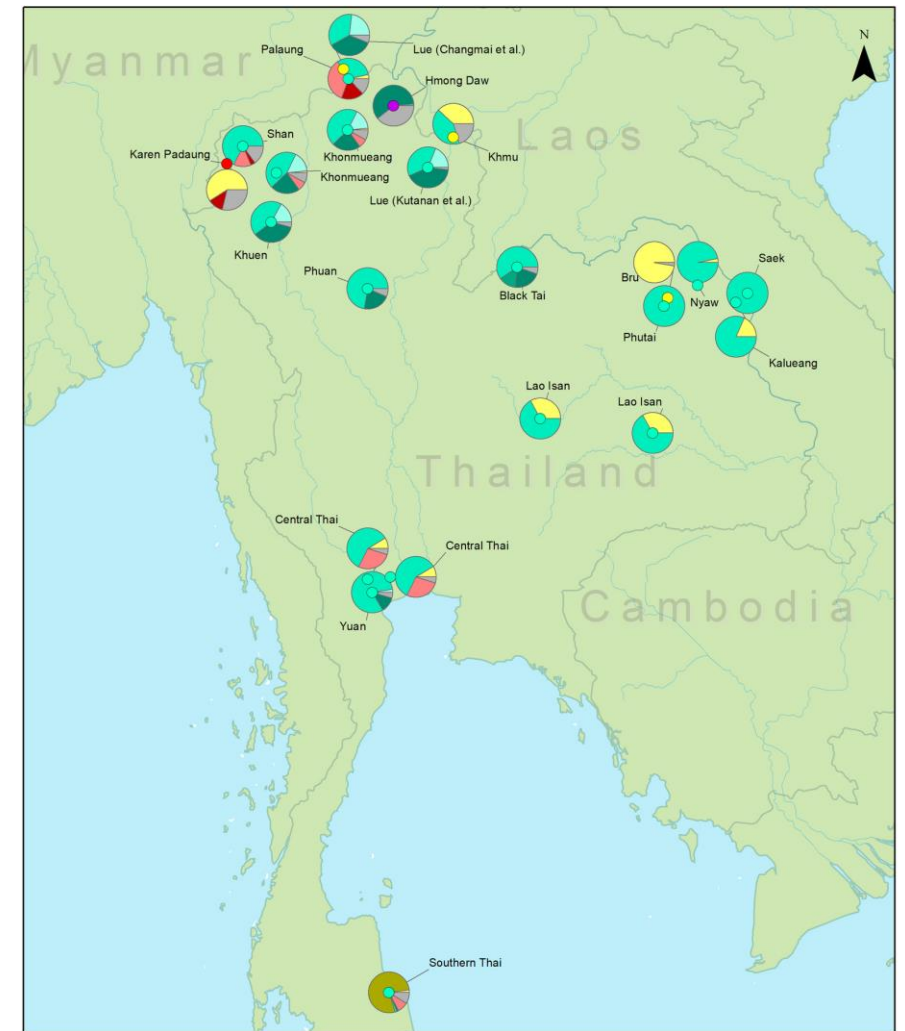
[Piya Changmai](#)¹, [Ron Pinhasi](#)^{2,3}, [Michael Pietrusewsky](#)⁴, [Miriam T Stark](#)⁴, [Rona Michi Ikehara-Quebral](#)^{4,5}, [David Reich](#)^{6,7,8,9}, [Pavel Flegontov](#)^{10,11,12,13}

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 Broomandkoshbacht](#)^{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.

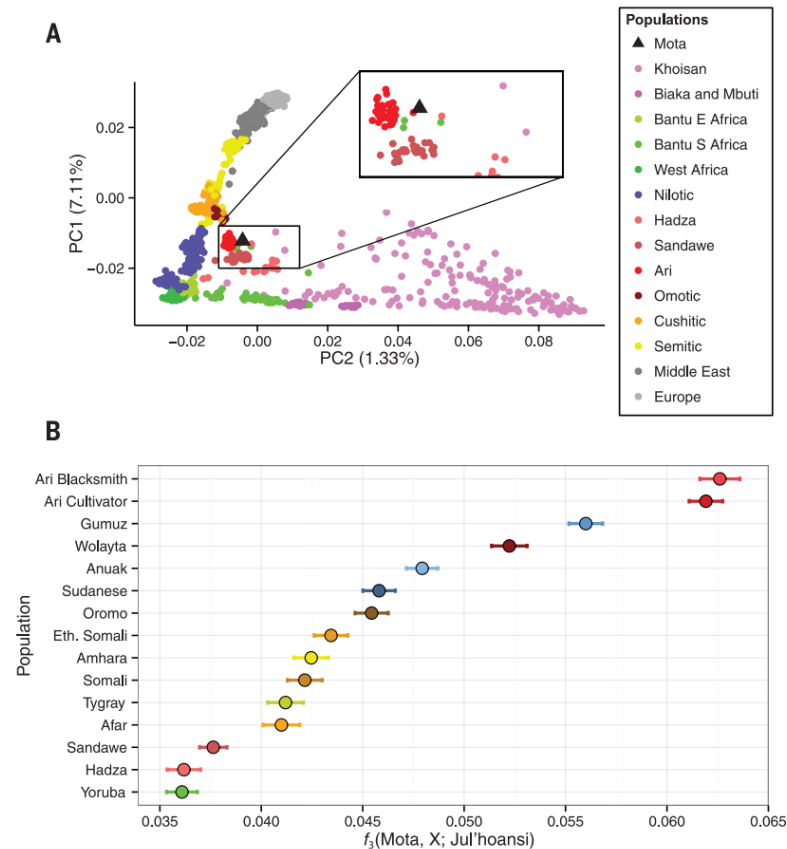
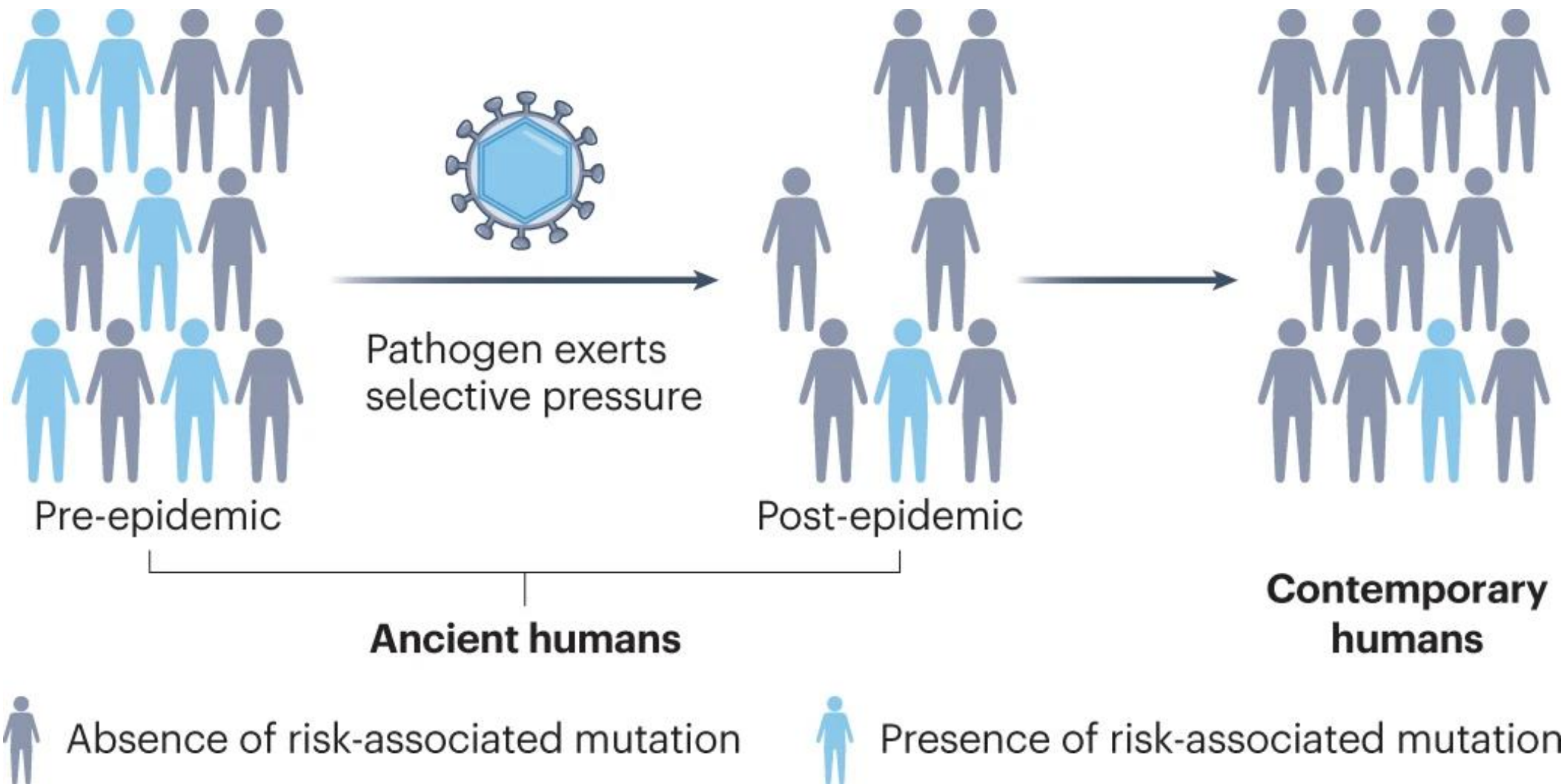


Fig. 1. Mota shows a very high degree of similarity with the highland Ethiopian Ari populations. (A) PCA showing Mota projected onto components loaded on contemporary African and Eurasian populations. The inset magnifies the PCA space occupied by Ethiopian and Eastern African populations. **(B)** Outgroup 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



HISTORICAL FIGURES

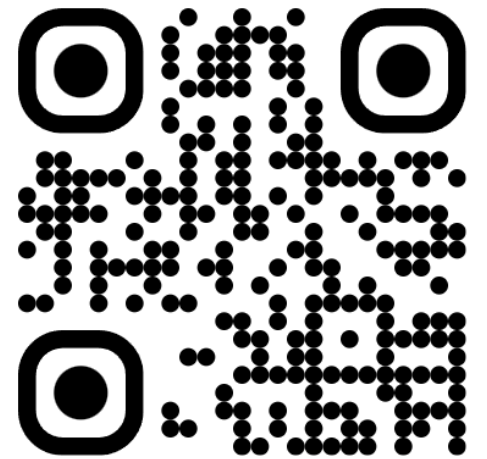
- G. J. Mendel
- L. van Beethoven
- Tatanka Iyotake

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.)



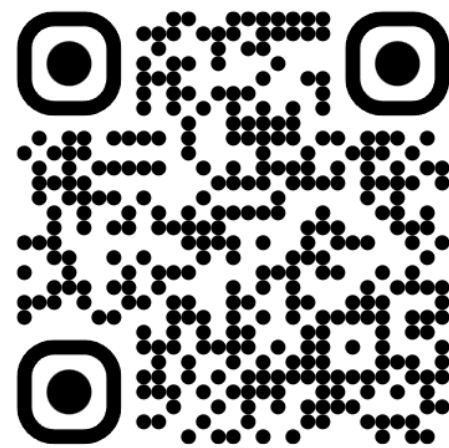
FOLIA MENDELIANA

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 BRZOBHATÁ, ROMANA SARVAŠOVÁ, LENKA FALK drozdova@sci.muni.cz

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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 DROZDOVÁ¹, EVA CHOCHOLOVÁ¹, KRISTÝNA BRZOBHATÁ¹, FILIP PARDY², ANNA ŠENOVSKÁ¹, HANA SVOBODOVÁ¹, KATEŘINA NOVOTNÁ¹, BORIS TICHÝ², MAREK PEŠKA³, ANTONÍN ZÚBEK³, MICHAEL DOUBEK^{4, 5}, ŠÁRKA POSPÍŠILOVÁ^{4, 5}

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ABSTRACT – Skeletal remains of five members of the Order of St. Augustine were found in the tomb at the Central Cemetery in Brno during an archaeological excavation in 2021. The aim was to identify Gregor Johann Mendel. Archaeological, anthropological, and genetic approaches were used for this purpose. However, the most reliable method to distinguish his skeletal remains was genetic identification. Specifically, the whole mitochondrial DNA (mitogenome) was read by next-generation sequencing (NGS). The mitogenomes of all five men and twenty DNA samples from Mendel's personal belongings (e.g., hair from his books and swabs) were compared. A match was found with one hair. The positive identification paved the way for proceeding with the project and reading his entire genome.

RECONSTRUCTING THE GENOME OF GREGOR JOHANN MENDEL USING STATE-OF-THE-ART MOLECULAR AND BIOINFORMATICS TOOLS

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ABSTRACT – Introduction and aims: Reconstruction of the genome from ancient DNA (aDNA) is difficult process, hampered by low amount of available DNA, post-mortem damage of nucleobases and contamination risks. However, we applied state-of-the-art NGS Methods and bioinformatics to inspect genomic features of Gregor Johann Mendel, founder of genetics.

Methods: aDNA was isolated from tooth using column-based isolation and concentrated with Amicon filter unit. After QC, Illumina Sequencing library was created using Swift 2S flexible kit (IDT), using molecular barcode oligo (IDT) and PreCR repair mix (NEB). Library was amplified with EvaGreen dye to optimize the cycling conditions. Libraries were sequenced using Illumina NovaSeq instrument with S4 chemistry in Paired-end 150 cycle setting. The Eager bioinformatics pipeline, freely available in the nf-core pipelines repository, was used to retrieve SNV/Indel variants and aDNA quality control metrics. Computationally demanding analysis was performed on cluster using Kubernetes technology.

Results: We managed to reconstruct 91% of Mendel's genome, 99% of exome, with 9,26x average coverage. We detected 4.1 million highly confident SNV/Indel variants genome-wide. Of them, 57308 occurred in the gene coding regions.

Discussion and conclusion: Using the latest approaches in the molecular biology and bioinformatics, we reconstructed Mendel's genome and identified genetic variants that might have shaped his life and health. Variants impact to phenotype is currently under investigation by biologists and physicians. In the opportunity of Mendel's bicentennial anniversary, we consider this as an important contribution and a gift to this genius.

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

EVA CHOCHOLOVÁ^{1*}, EVA DROZDOVÁ¹, DANA FIALOVÁ¹, PAVEL ROUDNICKÝ², ZBYNEK ZDRAHAL^{2, 3}, BARBORA ZWINSOVÁ⁴, KATEŘINA NOVOTNÁ¹, FILIP PARDY⁵, JAKUB HYNŠT⁶, MAREK PEŠKA⁷, ANTONÍN ZÚBEK⁷, KATEŘINA MRAZOVÁ⁸, VLADISLAV KRŽYŽÁNEK⁸, MICHAEL DOUBEK^{8, 9} and ŠÁRKA POSPÍŠILOVÁ^{8, 9}

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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.

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TO IDENTIFICATION SKELETAL REMAINS

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

BODY RE

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Department of

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Institute of
Haematology and

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ABSTRACT - In the skeletal remains of Gregor Johann Mendel, a team of researchers performed a complex analysis of biological traits of Mendel's remains. The focus is on the archaeological and period from June 6 to 1864. All excavated skeletal remains were analyzed using anthropological and genetic approaches. The positive identification paved the way for proceeding with the project and reading his entire genome.

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RECONSTRUCTING THE GENOME OF GREGOR JOHANN MENDEL USING STATE-OF-THE-ART MOLECULAR AND BIOINFORMATICS TOOLS

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

EVA CHOCHOLOVÁ^{1*}, EVA DROZDOVÁ¹, DANA FIALOVÁ¹, PAVEL ROUDNICKÝ², ZBYNEK ZDRAHAL^{2, 3}, BARBORA ZWINSOVÁ⁴, KATEŘINA NOVOTNÁ¹, FILIP PARDY⁵, JAKUB HYNŠT⁶, MAREK PEŠKA⁷, ANTONÍN ZŮBEK⁷, KATEŘINA MRAZOVÁ⁸, VLADISLAV KRZVÁNEK⁸, MICHAEL DOUBEK⁹ and ŠÁRKA POSPÍŠILOVÁ⁹

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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.

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Anthropological and genetical evaluation of the skeletal remains of Abbot Mendel brought new information on his body parameters.

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

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HISTORICAL FIGURES

Current Biology



Volume 33, Issue 8, 24 April 2023, Pages 1431-1447.e22

Article

Genomic analyses of hair from Ludwig van Beethoven

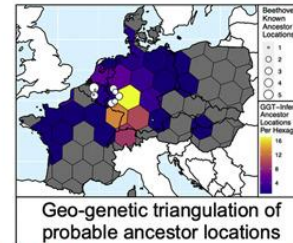
Tristan James Alexander Begg^{1 6 24}  , Axel Schmidt², Arthur Kocher^{9 22 24}, Maarten H.D. Larmuseau^{3 14 15 16}, Göran Runfeldt⁴, Paul Andrew Maier⁴, John D. Wilson^{5 28}, Rodrigo Barquera⁹, Carlo Maj^{2 27}, András Szolek^{18 19}, Michael Sager⁴, Stephen Clayton^{6 24}, Alexander Peltzer²⁶, Ruoyun Hui^{8 20}, Julia Ronge¹², Ella Reiter⁶, Cécilia Freund²⁴, Marta Burri²⁴, Franziska Aron²⁴, Anthi Tiliakou^{9 24}...Johannes Krause^{6 9 24 29 30}  

Authentication testing



Analyses of 24-fold genome of Ludwig van Beethoven

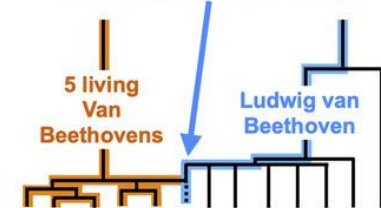
Genetic ancestry



> 99% European ancestry
Resembles Western/Central Europeans

Genetic genealogy

Extra-pair paternity event
between 1572 & 1770



Heritable disease risk

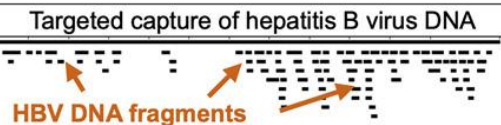
Liver disease risk in
PNPLA3 and
HFE genes



Interaction between genetic
risk and alcohol
consumption

Ancient pathogens

Infection with
hepatitis B virus



HISTORICAL FIGURES

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


[IDA MOLTKE](#) , [THORFINN SAND KORNELIUSSEN](#) , [ANDAINÉ SEGUIN-ORLANDO](#) , [J. VÍCTOR MORENO-MAYAR](#) , [ERNIE LAPOINTE](#), [WILLIAM BILLECK](#) , AND
[ESKE WILLERSLEV](#)  [Authors Info & Affiliations](#)

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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.



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- Interesting/important

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