



EMBO | EMBL  Symposium

Reconstructing the human past:

using ancient and modern genomics

15 – 18 September 2026 | EMBL Heidelberg and Virtual



Abstracts of papers presented at the

**EMBO | EMBL Symposium - Reconstructing the human past:
using ancient and modern genomics**

EMBL Advanced Training Centre, 15 - 18 Sep 2026

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Wlasnowolski, Michal	University of Cambridge, United Kingdom
Zhang, Xinjun	University of Michigan, United States of America

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Poster Removal - Please remember to collect your poster. Posters which have not been collected will be disposed of after the meeting.

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1

The genetic history of Europe: from the settlement of Europe to the medieval migration period

Johannes Krause¹

¹ Max Planck Institute for Evolutionary Anthropology, Germany

Presenter: Johannes Krause

Ancient DNA can reveal prehistoric events that are difficult to discern through the study of archaeological remains and modern genetic variation alone. Over the past decade, the emerging field of archaeogenetics analysed more than 20,000 ancient human genomes spanning the last 45,000 years of western Eurasian prehistory. Together we have uncovered dozens of large-scale migration events that shaped the genetic structure of modern-day Europe.

This lecture will cover some of the first unsuccessful attempts of modern humans to settle Europe during the Pleistocene after they left Africa around 50,000 years ago and mixed on their way with Neandertals. We will also cover the two major genetic turnover events at the beginning and at the end of the Neolithic period that dramatically changed the genetic landscape of Europe. First by early farmers who spread from Anatolia beginning about 8,000 years ago, bringing agriculture and domesticated animals to Europe. Following their arrival, early farmers genetically mixed with indigenous Europeans over the next 3,000 years. At the end of the Neolithic period, about 5,000 years ago, we can find the first genetic evidence of another major migration event of groups from the eastern European Pontic steppe, north of the Black Sea, into the European heartland. The newcomers were herders, practised pastoralism, and were highly mobile. Besides introducing new cultural practices, they may have been responsible for the spread of Indo-European languages.

We will finish with the collapse of the Roman Empire that was followed by a number of large-scale migration events resulting in the genetic structure of Europe as it is known today. Here, we will show how novel technological advances now allow us to identify short tracks of identity-by-descent (IBD) shared between individuals, making it possible to distinguish between populations that are otherwise genetically very similar to one another. This increased resolution opens a new window onto the recent past, allowing us to reconstruct fine-scale movements of people during the Migration Period, revealing connections between regions and communities that had previously remained invisible to archaeogenetic analysis.

2

Genomes of late Neandertals

Alba Bossoms Mesa¹, Elena Essel¹, Stéphane Peyrégne¹, Arev P. Sümer¹, Leonardo N.M. Iasi¹, Christian Heide¹, Divyaratan Popli¹, Cesare de Filippo¹, Marie-Theres Gansauge², Lars Gerullat³, Laurin Lippik², Sarah Nagel¹, Birgit Nickel¹, Barbara Schellbach¹, Anna Schmidt¹, Johann Visagie¹, Antje Weihmann¹, Hugo Zeberg^{1,3}, Julia Zorn¹, Hélène Rougier⁴, Isabelle Crevecoeur⁵, Patrick Semal⁶, Grégory Abrams^{7,8}, Thibaut Deviese⁹, Stéphane Pirson^{10,11}, Kévin Di Modica¹², Pierre Cattelain^{11,13,14}, Christelle Draily¹⁰, Michel Toussaint⁷, Isabelle De Groote⁷, Frido Welker¹⁵, Cosimo Posth^{16,17}, Marie Soressi¹⁸, Jean-Jacques Hublin^{1,19}, Johannes Krause¹, Svante Pääbo¹, Matthias Meyer¹, Janet Kelso¹, Benjamin M. Peter^{1,20}, Mateja Hajdinjak¹

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- 5 PACEA, UMR 5199, Université de Bordeaux, CNRS, Ministère de la Culture, Pessac, France
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- 13 Cedarc/Musée du Malgré-TBM out, Belgium
- 14 Université libre de Bruxelles, Belgium
- 15 Globe Institute, University of Copenhagen, Denmark
- 16 University of Tübingen, Germany
- 17 Senckenberg Centre for Human Evolution and Palaeoenvironment, Germany
- 18 Leiden University, the Netherlands
- 19 CIRB (UMR 7241–U1050), Collège de France, France
- 20 University of California, Los Angeles, USA

Presenter: Mateja Hajdinjak

Despite genome-wide data being recovered from close to 20,000 ancient humans to date, genomic data from Neandertals, our closest evolutionary relatives, are still comparatively sparse. Thus far, nuclear genetic data have been recovered from 35 Neandertals from 17 archaeological sites, spanning large parts of their history and geographical range. These data have offered a broad overview of Neandertal populations, indicating the existence of multiple distinct groups, as well repeated population turnovers.

To reconstruct a more fine-scale view of Neandertal populations, we used minimally destructive sampling of 36 skeletal remains which were radiocarbon dated to between ~52 and ~36 kya from ten archaeological sites in Belgium and France. Out of those, 27 contained enough endogenous DNA to generate autosomal, mitochondrial and Y-chromosomal data, including a new high-coverage genome from an ~45,000-year-old (i.e., ~45 kya old) Neandertal from Goyet. We found that late Neandertals from west Eurasia had much higher genetic diversity than the Neandertals from the Altai mountains, which lived between ~130 and 60 kya, suggesting that they lived in larger or better-connected groups. These Neandertals also had fewer long tracts of homozygosity, comparable to those found in the Upper Palaeolithic humans present in Europe around the same time. Although these Neandertals overlapped temporally with humans, we find no evidence of recent gene-flow from humans in their genomes. Moreover, we find no evidence for the accumulation of the deleterious mutations in the genomes of late Neandertals and no increase in genetic load over time, arguing against progressive genetic deterioration as a main driver of Neandertal extinction.

3**Deep coalescent history of the hominin lineage**

Trevor Cousins¹, Richard Durbin³, Regev Schweiger²

¹ Harvard University, United States of America

² Tel Aviv University, Israel

³ University of Cambridge, United Kingdom

Presenter: Regev Schweiger

Coalescent-based methods are widely used to infer population size histories, but existing analyses have limited resolution at deep time scales (>2 million years ago). Using new high-quality telomere-to-telomere genome assemblies, we extend the scope of such inference and reanalyse an ancient peak in human effective population size around 3–6 million years ago, showing that coalescent-based inference can reach much further into the past than previously thought. Furthermore, peaks at approximately the same time as in humans are also observed in chimpanzee and bonobo, but not in gorilla or orangutans. Such peaks can arise from ancestral genetic structure, and we suggest that a parsimonious explanation for the contemporaneous peaks in humans, chimpanzees and bonobos would be a period of complex speciation before final separation of *Homo* from *Pan*, in contrast to a scenario where these lineages diverge cleanly from a panmictic common ancestor.

4

Comparative 3D chromatin modelling reveals cell-type-specific changes in enhancer–promoter organisation associated with archaic hominin lineage-specific deletions

Nikita Kozlov³, Michal Wlasnowolski², Dariusz Plewczynski⁴, Guy Jacobs¹

¹ University of Cambridge, United Kingdom

² University of Cambridge, United Kingdom / Warsaw University of Technology, Poland

³ Warsaw University of Technology, Poland

⁴ Warsaw University of Technology / University of Warsaw, Poland

Presenter: Michal Wlasnowolski

Three-dimensional chromatin structure shapes spatial enhancer–promoter interactions that are fundamental to cell-type-specific gene regulation. Structural variants can alter chromatin topology and regulatory contacts, potentially contributing to phenotypic divergence between evolutionary lineages. Because direct data on the chromatin architecture of extinct hominins is not available, computational modelling is essential for exploring the regulatory consequences of archaic genetic variation.

We reconstructed the impact of structural variants on the 3D genome of Neanderthals and Denisovans using the cudaMMC engine and CTCF ChIA-PET interaction data from three distinct cell types (GM12878 lymphoblastoid cells, H1-ESC embryonic stem cells, and HFFc6 fibroblasts). We find that archaic deletions can induce substantial changes in local enhancer–promoter distance profiles; furthermore, this often occurs in a cell-type-specific manner, highlighting diverse regulatory consequences of archaic structural variation across cellular contexts. Gene ontology analysis of affected loci further supports this cell-type specificity. More generally, archaic structural variants vary substantially in their impact depending on the local regulatory neighbourhood: for some loci, archaic deletions substantially increase the predicted distance between a promoter and its nearest enhancer; in others, alternative nearby enhancers appear to maintain local regulatory proximity. These results illustrate how integrating ancient genomic variation with chromatin 3D modelling can reveal evolutionary changes in regulatory architecture that are invisible to sequence-level analysis alone.

Michal Wlasnowolski is the recipient of an EMBO|EMBL Symposia fellowship, jointly funded by EMBO and the EMBL Advanced Training Centre Corporate Partnership Programme.

5

Hypothesis: A modern human range expansion ~300,000 years ago explains Neandertal originsDavid Reich¹¹ Harvard University, United States of America

Presenter: David Reich

In this presentation, I demonstrate the feasibility of the hypothesis that Neandertals formed when a population using recently developed Levallois stone tool technology and intensively using fire expanded between 400-250 thousand years ago (ka). In Europe, their range expansion into an area with Sima de los Huesos-like people led to massive introgression of local archaic genes producing a population with around 90% archaic ancestry (Neandertals); if this range expansion was sex-biased it would provide a simple explanation for why Neandertals retain modern human lineage Y chromosomes or mitochondrial DNA. In Africa, interbreeding with local archaic humans led to more modest ~20% archaic admixture and the deep substructure detected in all modern humans today. This proposal explains five previously perplexing similarities of modern humans and Neandertals—sharing of mitochondrial DNA, Y chromosomes, the *PRDM9* A allele, cultural traits including Levallois tools and fire, and 300-200 ka date of formation by mixture—even while Neandertals and Denisovans cluster genome-wide. Motivated by the view that living humans and Neandertals (but not Denisovans) can be considered as descendants of expansive early modern humans, I present a catalog of genomic locations where all living humans and Neandertals share a common ancestor in the last 700 ka but Denisovans are much more distantly related. These previously overlooked loci are candidates for harboring key early modern human biological innovations.

6

New insights from studying Human-Neanderthal admixture

Laurits Skov¹, André Bourbonnais¹, Ioannis Patramanis¹, Kitty Murphy¹

¹ Globe Institute, University of Copenhagen, Germany

Presenter: Laurits Skov

When modern humans left Africa some 50,000 years ago they encountered groups of archaic humans such as the Neanderthals and Denisovans. Even though we have learned a great deal about these meetings, questions still remain. Here I present ongoing research that aims to answer these questions:

1. Where did these meetings take place? 2. Which role does introgressed archaic DNA have in our cells today? 3. why did some parts of our genomes resist introgression?

1. While coalescent simulations have been of enormous importance in the field of population genetics they often lack a spatial component. Here I present a workflow that allows the user to create forward-in-time simulations in SLIM, with realistic recombination maps, sex-specific inheritance for sex chromosomes, mating choice and different population densities for an arbitrary number of populations. We keep track of admixture tracks and create scripts to visualize mean admixture proportions, location of admixture tracks and sharing across individuals.

2. With the advent of large scale consortium like GTEx and UK biobank it has become possible to study the effect of introgressed variants. Here we revisit previous work which studied differences in gene expression between archaic and modern human variants. We discover that some previous studies suffered from reference mapping bias. This lead to a decrease in reads with the archaic variant mapping which lead to underestimation of the expression of archaic alleles across tissues. When we correct for reference bias we see that archaic variant are even more highly expressed in brain tissues.

3. Researches have sequenced 1,000 of present day and ancient modern humans and learned that some regions in the genome resist introgression – perhaps because they had a higher load of deleterious mutations. One notable example is the Y chromosome. With the sequencing of the first high coverage Neanderthal and Denisova males we are now able to study the repetitive ampliconic regions of the Y chromosomes. We discover multiple mega-base-scale deletions that are fixed in both Neanderthals and Denisovans. These deletions span multi gene families which are primarily expressed in testis - suggesting a mechanism for why these Y chromosome versions might have been selected against.

7

Genomic contributions to understanding the history of Latin America & the Caribbean

María Nieves-Colón¹

¹ University of Minnesota, United States of America

Presenter: María Nieves-Colón

Ancient DNA (aDNA) directly queries the genetic diversity of ancient and historic human populations. Such data can be used to investigate the experiences of peoples who have been historically marginalized or excluded from the written documentary record. As such, aDNA is especially relevant for understanding the history of Latin America, a region where settler colonial perspectives are overrepresented in the written historical record; often serving as the hegemonic frameworks from which past events are reconstructed and understood. In this talk I will discuss how the UMN Anthropological Genetics Laboratories combines aDNA and genomics, community engaged methods and anthropological lines of evidence, to investigate the histories of Latin America and Caribbean communities, focusing on case studies from our ongoing projects. This presentation will stress how integrative aDNA research can problematize our understandings of history by constructing more inclusive narratives of the human experience.

8

The genomic and phenotypic legacy of the peopling of Polynesia

Elise Kerdoncuff¹, Maguelonne Roux², Dang Liu³, Leo Speidel³, Iotefa Teiti⁵, Maïté Aubry⁴, Gaston Rijo de Leon⁵, Jeremy Choin⁴, Mathilde André⁶, Anita Teissier⁶, Vaea Richard⁵, Zhi Li⁶, Christine Harman⁶, Aurélie Bisiaux⁶, Anthony Jaquaniello⁶, Tuterarii Paoaafaite⁵, Lisa Dian⁵, Kiyojiken Chung⁵, Annie Forster¹⁴, Adrian Hill¹⁴, George Aho¹⁶, Ratu Vereniki Raiwalui¹¹, Alexander Ioannidis¹³, Andres Moreno¹, Alexander Mentzer¹⁴, Phillip Endicott¹⁵, Mark Stoneking⁹, Frédérique Valentin¹⁰, Jacques Vernaudeau¹², Guillaume Molle¹², Eric Conte¹², Mary Walworth², Aymeric Hermann¹⁰, Van-Mai Cao-Lormeau⁵, Etienne Patin⁷, Luís Quintana-Murci⁸

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¹⁵ University of Tartu, Estonia

¹⁶ Vaiola Hospital, Tonga

Presenter: Elise Kerdoncuff

The late settlement of the remote islands of Polynesia by populations of different ancestries, together with the high prevalence of metabolic disorders, raises fundamental questions about how genetic drift, admixture and local adaptation have shaped human phenotypic diversity. Yet integrative studies combining population and quantitative genetics in Polynesians, one of the world's most underrepresented groups in genomic research, remain scarce. Here we generated whole-genome sequences from 1,919 individuals across 24 islands and collected deep phenotypic data for 1,761 individuals. We show that all Polynesians derive from a major admixture event between Austronesian- and Papuan-related groups ~2,200 years ago, followed by rapid divergence into Western, Eastern and Outlier Polynesian groups. Using a new method based on ancestry-masked genome-wide genealogies, we find that Eastern and Outlier Polynesians experienced some of the most severe population bottlenecks documented in human history. We detect and date trans-Pacific genetic contacts with South Americans, whose ancestry likely spread westward through Eastern Polynesia 200-400 years after initial settlement. We further show that most European gene flow occurred in Eastern Polynesia during the 19th century and was male-biased. Genome-wide association studies of 46 anthropometric and health-related traits identified 13 significant loci, including common variants in Pacific populations influencing lipid levels, hemoglobin concentration and humoral immunity to dengue virus. Reconstructed allele-frequency trajectories reveal no evidence of Polynesian-specific polygenic adaptation for traits related to obesity, type 2 diabetes and dyslipidemia, suggesting that the high burden of metabolic disorders in the region reflects recent lifestyle changes acting on a background of numerous weak-effect, quasi-neutral variants. Overall, our study elucidates the demographic dynamics and phenotypic legacy of Polynesian settlement, offering evolutionary insights into the genetic architecture of disease risk.

Speaker Abstracts

9

Reconstructing social structure in an ancient Oceanian population

Lara R. Araúna³, Michal Feldman², Selina Carlhoff¹, Kathrin Nägele¹, Adam Powell¹, Johannes Krause¹, Lawrence Kiko², Hallie R Buckley⁴, Rebecca L Kinaston⁴, Cosimo Posth⁵

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⁴ University of Otago, New Zealand

⁵ University of Tübingen, Germany

Presenter: Lara R. Araúna

The peopling of Oceania involved multiple waves of human expansion, beginning with the settlement of Near Oceania over 50,000 BP by groups carrying Papuan-related ancestries, followed much later by the Austronesian expansion, which introduced East Asian-related ancestries into Remote Oceania ~3,000 BP. Subsequent movements, including the dispersal of Polynesian-speaking groups beyond the Polynesian triangle, generated complex patterns of interaction and admixture across island communities. However, how these demographic processes shaped local systems of social organization remains largely unknown.

Here, we present genomic analyses of 111 ancient individuals from the Namu burial site on Taumako (eastern Solomon Islands), a population with strong Polynesian cultural affiliations. New radiocarbon dates (400–40 BP) indicate that the site is younger than previously reported and genetically distinct from present-day Polynesian Outlier populations. Despite this, individuals exhibit a highly homogeneous ancestry profile, with roughly equal contributions from Papuan- and East Asian-related sources, alongside exceptionally dense networks of genetic relatedness within the burial site. Using genome-wide relatedness estimates, runs of homozygosity, and uniparental markers, we reconstruct extended pedigrees spanning multiple generations. These reveal structured mating systems characterized by frequent parental relatedness and patterns consistent with female gene-flow between neighbouring islands, suggesting regular mobility embedded within a stable local community.

Our findings demonstrate how integrating genomic and archaeological data enables the reconstruction of fine-scale social structure, providing new perspectives on how Oceanian communities organized social relationships in the recent past.

10

Genome-wide ancient DNA data reveals five millennia of population history across the Indian Himalayas

Aparna Dwivedi², Niraj Rai², Snigdha Konar², Esha Bandyopadhyay⁴, Sonam Spalzin¹, Maanasa Raghavan³

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Presenter: Niraj Rai

India harbors one of the world's most complex and diverse genetic landscapes, yet its deep population history remains poorly understood due to the scarcity of ancient genomic data. While present-day genetic variation reflects contributions from multiple ancestral sources, the timing, origins, and demographic impact of these ancestries remain unresolved. To address this gap, we generated genome-wide ancient DNA data spanning ~5,000 to 200 years before present from sites across the Upper and Lower Himalayas, including Ladakh, Spiti Valley, Kashmir, and Northeast India.

Our results reveal substantial genetic heterogeneity across the Himalayan region through time. Ancient individuals from the high-altitude western Himalayas in Ladakh and Spiti derive genetic ancestry related to ancient Tibetan, lowland South Asian, and Central Asian groups, with admixture dates estimated to approximately 2,100-2,600 years ago. Later individuals from the region fall along a northern Himalayan genetic cline associated with Tibetan- and Steppe-related ancestries. In contrast, ancient individuals from the lower Himalayas in Kashmir demonstrate genetic continuity with ancient and present-day groups from lowland South Asia, providing insights into the emergence of this ancestry profile. Furthermore, ancient genomes from the eastern frontier of the lower Himalayas reveal a previously undescribed ancestry component with implications for the genetic and linguistic history of populations in the eastern Himalayas, highlighting additional layers of demographic complexity in the region.

Together, these findings provide the first high-resolution temporal genomic framework for the Indian Himalayas and demonstrate the power of ancient DNA to reconstruct population movements, admixture processes, and regional population continuity across South Asia over the past five millennia.

Niraj Rai is the recipient of an EMBO|EMBL Symposia fellowship, jointly funded by EMBO and the EMBL Advanced Training Centre Corporate Partnership Programme.

11**Genetics in historical genealogies**Simon Gravel¹¹ McGill University, Canada

Presenter: Simon Gravel

Relatedness between individuals can be measured at the genealogical level (by describing shared ancestors in a pedigree) or at a genetic level (by describing shared haplotypes across the genome). The shared haplotype structure can be conveniently summarized as a sequence of trees along the genome, where each tree describes the last shared genetic ancestors between individuals at a locus. While many tools exist to infer tree sequences from genetic data, and many large pedigree datasets are available, few tools exist to identify the relationship between the two – finding which genetic ancestor corresponds to which pedigree ancestor, and conversely.

In this presentation, we propose an algorithm to solve this problem by providing, for each genetic tree, the list of all consistent ancestry paths within a genealogical tree. We also provide variants of the algorithm with moderate robustness to both tree sequence and pedigree errors. We demonstrate the scalability of our approach on the BALSAC genealogical dataset, which includes millions of individuals in Quebec, Canada. We find that 20 carriers usually provide enough information to reliably identify a common ancestor 15 generations ago or find the parent of origin of alleles among probands, but the number of possible ancestry paths within the pedigree can remain large. We apply the method to reconstruct the inheritance of a causal allele for type 1 myotonic dystrophy.

12**Decoding the genetic legacies of human history through ancient DNA**Shigeki Nakagome¹¹ Trinity College Dublin, Ireland

Presenter: Shigeki Nakagome

Ancient DNA provides a direct window into the human past, allowing us not only to reconstruct population movements and interactions, but also to explore how these histories have shaped patterns of genetic variation through time. At the same time, ancient pathogen genomes offer another record of human history, revealing how infectious diseases spread and evolved in response to human movements and activities. In this talk, I will explore these different dimensions of human history through recent ancient genomic studies in Asia and the Americas. First, I will discuss how genetic ancestry inherited from ancient populations persists in people today and ask whether such ancestry may also contribute to present-day phenotypic variation. I will then shift focus to ancient pathogen genomics and explore how pathogen genomes can provide independent insights into major episodes of human history. Together, these case studies illustrate how human history has left its signatures both in the genomes we inherited and in the genomes of the pathogens we encountered.

13

From desert to Patagonia: paleogenomic perspectives across South America

Constanza de la Fuente¹

¹ University of Copenhagen, Denmark

Presenter: Constanza de la Fuente

South America stretches across an extraordinary range of environments, from the high-altitude deserts of the Andes to the fjords and steppes of Patagonia at the continent's southern tip. Human populations have faced this highly diverse landscape, settling, moving through and adapting to these conditions. Ancient DNA, paired with genome-wide data from present-day communities, has increasingly contributed to trace this history, revealing patterns of migration, regional continuity, and local diversification complementing the archaeological or ethnohistoric record. In this talk I will take a latitudinal journey through paleogenomic research on South America, from high-altitude and desert-adjacent sites in the Andes to maritime and terrestrial hunter-gatherer groups in far southern Patagonia, where ancient genomes have revealed both long-term genetic continuity and clear diversification between neighboring groups. Along the way, I will engage with some of the major debates shaping the field: the timing, number, and routes of the initial peopling of the continent; the extent of population continuity versus replacement and admixture over time; and how ancient genomes intersect with archaeological, linguistic, and community perspectives on the past. The talk closes by considering what these regional case studies reveal collectively about the population history of South America, and the methodological, logistical, and ethical challenges of doing ancient DNA research in the region, including the collaborative frameworks needed to make the field sustainable for the communities and countries where this history lies.

14

Ancient human DNA from chewed birch bark tar reveals the origin of the first settlers of Estonia

Lehti Saag⁵, Irina Khrustaleva⁵, Kadri Irdt⁵, Mari Tõrv⁵, John Meadows³, Theis Zetner Trolle Jensen⁴, Maja Birk Sørensen⁴, Helja Kabral⁵, Anu Solnik⁵, Anto Aasa⁵, Stefania Sasso⁵, Eugenia D'Atanasio¹, Ene Metspalu⁵, Siiri Rootsi⁵, Toomas Kivisild², Hannes Schroeder⁴, Kristiina Tambets⁵, Aivar Kriiska⁵, Mait Metspalu⁵

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⁴ University of Copenhagen, Denmark

⁵ University of Tartu, Estonia

Presenter: Mait Metspalu

Modern humans first settled in Estonia approximately two millennia after the retreat of the ice sheet, with the earliest archaeological evidence dating to the 9th millennium BCE. However, the absence of human remains from these early periods has prevented direct genetic analysis of the first settlers. Based on genetic data from later periods (8th–7th millennium BCE), it has been hypothesized that the first settlers of the Northern Baltic were closely related to Western hunter-gatherers (WHG). We overcome this limitation by turning to birch bark tar – an adhesive widely used in prehistoric Europe that was processed by chewing and has recently been shown to preserve the embedded DNA for millennia. Here we analyse ancient human DNA from both skeletal remains and 4 chewed pieces of birch bark tar, representing 14 individuals from seven Estonian sites dated from the 9th to 4th millennium BCE. Our results reveal that, contrary to previous assumptions, Estonia's earliest known inhabitants of the 9th millennium BCE were genetically closer to Eastern hunter-gatherers (EHG). Between 8th–5th millennium BCE individuals from the Eastern Baltic region show stronger affinities with WHG while retaining north-south EHG-WHG ancestry gradient. In the 4th millennium BCE, people associated with the Comb Ware culture once again display genetic similarities to EHG. Together, these findings reveal a complex and non-linear population history among northern European hunter-gatherers, marked by temporal shifts in EHG- and WHG-related ancestry, and establish birch bark tar as a valuable source of ancient human DNA.

15

Genomic history of Mexico reveals bidirectional long-distance connectivity across the Americas

Santiago Gerardo Medina-Muñoz¹, Aaron P Ragsdale⁴, Hie Lim Kim², Nathaniel S Pope³, Andres Moreno¹

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³ University of Oregon, United States of America

⁴ University of Wisconsin-Madison, United States of America

Presenter: Santiago Gerardo Medina-Muñoz

The ancient Americas sustained two spheres of cultural complexity within reach of each other: maize domesticated in Mexico transformed subsistence from the Andes to the Mississippi, while Andean metallurgical traditions reshaped the political economies of West Mexico. Yet, whether the networks carrying these innovations also carried people, whether the exchange was demographic and not merely material, has remained unanswered. Here, we analyze 516 high-coverage genomes representing Indigenous lineages across Mexico, together with genomic data from hundreds of South American individuals, to reconstruct pre-Hispanic population history at fine temporal resolution. We identify three major ancestral lineages within Mexico --northern, central, and southern-- that diverged in the Late Pleistocene. Post-divergence interaction was structured by geography, with the Gulf lowlands acting as a persistent corridor connecting otherwise differentiated regions. This resolved demographic structure proves essential for detecting signals of long-distance connectivity. We identify a temporally and geographically structured history of connectivity between Mesoamerica and South America. A mid-Holocene signal along the Pacific coast, strongest in Ecuador and the Colombian Pacific, points to demic diffusion accompanying the spread of maize into South America. Late Holocene interactions were bidirectional and route-specific: South American ancestry reached western Mexico via Pacific maritime networks, while Maya-related ancestry spread into the Wayuu of Caribbean South America. Together, these findings establish ancient Mesoamerica not as a demographic periphery of the Americas, but as an active node of bidirectional continental exchange whose reach extended from the Andes to the Caribbean world.

Santiago Gerardo Medina-Muñoz is the recipient of an EMBO|EMBL Symposia fellowship, jointly funded by EMBO and the EMBL Advanced Training Centre Corporate Partnership Programme.

16

45,000 years of changes in ancestry profiles and environmental conditions as shaping forces of Homo sapiens demography worldwide

Hélios Delbrassine², Francesca Carollo², Luca Pagani², Francesco Montinaro¹, Massimo Mezzavilla²

¹ University of Bari Aldo Moro, Italy

² University of Padova, Italy

Presenter: Hélios Delbrassine

Human groups move and interact with both their environment and each other, sometimes exchanging genetic and/or cultural information. As a result, the evolutionary history of our species is deeply intertwined with its demographic history and understanding it requires reconstructing the changes in population size, admixture patterns, and ecological contexts of human populations across space and time.

In this work, we provide a global atlas of admixture history and habitat suitability for Homo sapiens over the past 45,000 years. To do so, we characterize the ancestry profiles of more than 22,000 ancient and present-day individuals and investigate the population movements that shaped human genetic diversity through time. We then leverage global palaeoclimatic reconstructions and archaeological assessments of human presence to map the spatiotemporal changes in habitat suitability that may have driven some of these movements. Finally, we compute more than 13,000 estimates of genetic effective population size (N_e), a proxy for census size. By analyzing changes in N_e alongside shifts in ancestry and environmental conditions, we investigate potential drivers of demographic changes across space and time. We also identify cases of population growth in spite of poor environmental conditions, potentially reflecting the emergence of niche-expanding cultural innovations. For example, we find that part of the fertile crescent fell outside of the predicted range of habitability 11,000-12,000 years ago, despite seeing the advent of agriculture. Overall, our work constitutes a unique effort to integrate molecular and ecological data at a global scale, providing a comprehensive framework for investigating the human past for archaeologists, molecular anthropologists and cultural evolution studies alike.

Helios Delbrassine is the recipient of an EMBO|EMBL Symposia fellowship, jointly funded by EMBO and the EMBL Advanced Training Centre Corporate Partnership Programme.

17

Gene-culture coevolution in the European Upper Palaeolithic

Cansu Karamurat¹, Francesco Ravasini¹, Felix Riede¹, Mikkel Schierup¹

¹ Aarhus University, Denmark

Presenter: Francesco Ravasini

Human palaeogenomic studies have traditionally been focused on biological relatedness, migration, and replacement. Archaeological and palaeoenvironmental metadata have often remained underutilised and recording of these has been repeatedly criticised as insufficiently standardised. As a result, the complexity of the archaeological record and how it aligns with – or contradicts – the genetic interpretations have not seen much formal scrutiny. Here, we focus on the European Terminal Pleistocene from ~22,000-11,000 years before present as a case where palaeogenomic samples and the archaeological record can be analysed in a gene-culture coevolutionary framework. Our results indicate a strong association between cultural and biological relatedness, suggesting that certain cultural taxonomic labels may indeed mirror groups of common genetic ancestry. Magdalenian individuals show strong genetic connections across a large range coupled with a standardised material culture, indicating low population density and high mobility in a dispersal context. In agreement with earlier studies, we also highlight a major yet not complete biocultural turnover at 14,000 years before present linked to the expansion of Epigravettian groups and their descendants in Central-Western Europe. Runs of Homozygosity analyses suggest small populations throughout the Terminal Pleistocene with the Italian Epigravettian – despite sharing a common material culture – consisting of particularly fragmented local groups. We show that heterogeneous landscapes across the Italian peninsula may have shaped this pattern, potentially alongside an elevated degree of endogamy. We highlighted distinct biocultural relationships between and within the established archaeological cultures, reflecting different mobility strategies and social structures. Grounded in well-established gene-culture coevolutionary theory, our approach contributes to a more robust integration of archaeology and palaeogenomics.

18

Genetics and social practices at Mogou and surrounding Gansu sites at the onset of trans-Eurasian exchangesHan Shi¹, Qiaomei Fu¹¹ Chinese Academy of Sciences, China

Presenter: Han Shi

The Gansu region in northwestern China served as a key corridor for trans-Eurasian interaction, which reported western-derived domesticated crops and livestock at least around 4000 years ago. However, it is still unclear whether large-scale trans-Eurasian demic movements reached Gansu, and the population history and social practices of the ancient Gansu people remain understudied. In this study, we obtained genome sequences of 149 individuals spanning ~4700-3000 BP, including 95 individuals from the large Mogou cemetery and 54 individuals from ten other sites. Through population genetic analyses, we found that all the new and published samples from thirteen sites around this time period, including those from nine sites that had utilized western-derived crops and livestock, showed no detectable evidence of genetic influence from western Eurasia. Instead, these samples carried most of their ancestry associated with northern East Asian populations, including populations near the Yellow River region, the southeastern Mongolian Plateau, and the Tibetan Plateau. This suggests that the spread of western domesticates into Gansu, at least at our studied sites, occurred largely without large-scale west-to-east population movement, which differed from the agricultural diffusions to Europe and Central Asia with demic expansions of Neolithic Anatolian farmers and Bronze Age Yamnaya agropastoralists. We also investigated the close kinship relationships and the shared identity-by-descent segments based on samples from the Mogou cemetery. Results showed widespread genetic ties within the community, which continued through the Qijia culture to the Siwa culture. Female and male samples did not show significant differences in relatedness estimates, suggesting no strong evidence for either matrilocality or patrilocality. While multiple burials and repeated use of tombs were common at Mogou, we did not find strong support for burial practices centered on genetic relatedness. Individuals with non-local ancestry in the Mogou cemetery tended to be linked to burial contexts archaeologically identified as lower-status.

19

Archaeogenomic insights into a single Islamic community buried in the Tauste necropolis (northeastern Iberia) over four centuries

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⁷ University of Zaragoza, Spain

Presenter: Laura Claudia Verdesca

711 CE marks the onset of the Islamic Period on the Iberian Peninsula. Throughout its duration, Islamic funerary rites became the predominant burial practice. The Islamic necropolis of Tauste, located in Aragón (northeastern Iberia), represents one of the largest Islamic burial grounds identified in the region. Nearly 700 graves have been excavated, and all recovered individuals were buried following Islamic burial customs, including simple pit graves devoid of grave goods and bodies placed in right lateral decubitus or supine position with the head oriented toward the southeast. Available radiocarbon dates indicate that the necropolis was used continuously between the first half of the 8th century and the beginning of the 12th century CE, fully overlapping with the Islamic occupation of this rural area. Here, we generated genome-wide ancient DNA data using in-solution enrichment from 149 individuals precisely geolocated and sampled from typologically characterised graves distributed across the necropolis. Our results reveal the arrival of new North African-related ancestry. However, despite the remarkable homogeneity of the Islamic funerary rite throughout the necropolis, we identify both unadmixed ancestry from the preceding, local population, and genetically admixed individuals, reflecting the incorporation and religious conversion of pre-existing local populations into the Islamic community. These findings suggest that the Islamization of the region was not exclusively mediated by admixture processes, but also involved the cultural and religious integration of local groups. The analysis of biological relatedness and consanguinity at the intra-site level reveals a highly consanguineous and close-knit community, with more than 65 first- and 150 second-degree relationships, with most of the individuals connected through 3rd and 4th degree links. Furthermore, integrating genomic relatedness with the spatial distribution of burials allows us to explore funerary organisation within the necropolis. Our results reveal the presence of extensive family groups with 1st- and 2nd-degree relatives, indicating a pattern of closer spatial proximity, suggesting the reservation of funerary spaces according to close family bonds. Together, these findings provide unprecedented insights into the long-term evolution of rural Islamic communities in Medieval Iberia.

20

Research reciprocity - engaging equitably with indigenous peoples and local communities in ancient genomics

Maui Luke Hudson¹

¹ University of Waikato, New Zealand

Presenter: Maui Luke Hudson

Research reciprocity represents a deep commitment to partnership with Indigenous communities in research collaborations. It emphasises the need for not only benefit sharing but also power sharing across the broader research lifecycle encompassing both upstream and downstream activities. Research reciprocity is inherently relational with a focus on distributing resources, opportunities, decision-making and control in an equitable manner. This presentation will explore rights recognition, ethical research practices, enabling infrastructures and benefit sharing in paleogenomics, drawing on learnings from Indigenous research and the FREYJA project which is developing a roadmap for practical implementation of research reciprocity.

21**Beyond bones and teeth - exploring alternative sources of ancient DNA for investigating the human past**

Matthias Meyer¹, The Ancient Environmental DNA Analysis consortium

¹ Max Planck Institute for Evolutionary Anthropology, Germany

Presenter: Matthias Meyer

Many questions about the deep human past cannot be addressed through the genetic analysis of human skeletal remains alone, which are rare or absent at most Palaeolithic sites. Over the past decade, we have therefore systematically surveyed other materials for DNA preservation. Sediments have emerged as a particularly rich source of ancient DNA, providing information about past ecosystems but also allowing to directly reconstruct human occupation at archaeological sites. Published work, however, has concentrated on sites with above-average success rates in DNA retrieval. To determine success rates of DNA retrieval from sediments in an unbiased manner, we are therefore compiling a survey of faunal and human DNA preservation across 135 archaeological sites, including caves, rock shelters and open-air sites. Our data show that DNA preservation in sediment follows similar temporal and geographic limits to DNA in skeletal remains, that ancient human DNA is far less abundant than faunal DNA but can still be frequently detected if large enough sample numbers are analysed, and that movement of DNA across archaeological layers does not appear to be a widespread problem.

We are also studying DNA preservation in stone and bone tools and ornaments. From a Palaeolithic deer tooth pendant we retrieved ancient DNA from a female human individual, providing the first genetic link between a Palaeolithic cultural artefact and its maker or user. Beyond cave floor deposits, we attempted DNA recovery from Palaeolithic rock art and samples of surrounding non-pigmented cave walls, and recovered ancient human DNA from both. While direct associations between cave art and its creators remain difficult to establish, genetic data may in future be used to study usage patterns of caves and detect the presence of past human populations independently of cave floor excavations.

22

Deep learning prediction of cis-regulatory turnover in human evolution

Riley Mangan¹, Nikitha Thoduguli¹, Dimitar Ivanov², Jayashabari Shankar¹, Yuru Lin¹, Tom Zeerow², Zunpeng Liu¹, Martin Wohlwend¹, Janet H.T. Song², Manolis Kellis¹

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² Harvard University, United States of America

Presenter: Riley Mangan

Deciphering the regulatory consequences of sequence divergence across human evolution is essential to understanding the molecular basis of human-specific traits and disease susceptibilities. Although millions of derived alleles distinguish modern humans from great apes, only a small fraction are likely to influence human-specific traits. Previous studies have therefore focused on regions with elevated sequence divergence, based on the idea that rapid molecular evolution reflects functional adaptation. However, individual regulatory mutations that influence gene expression may evade detection in scans for elevated divergence. To address these limitations, we developed a deep learning-based framework to reconstruct cis-regulatory changes across human evolution. We built personalized reference-guided genome assemblies and applied a pre-trained sequence model to predict chromatin accessibility profiles in 100 distinct cell and tissue epigenomic contexts across 34 modern human, archaic hominin, and great ape haplotypes. We report 37,592 differential accessibility (DA) events across 11,050 distinct regions between hominins and great apes, and 707 DA events across 235 regions between modern humans and archaic hominins. Predicted DA elements were enriched for experimentally-observed interspecies regulatory differences and neurogenic and developmental pathways, consistent with known phenotypic transitions. We found that hominin-ape DA elements are typically shorter, less conserved, and less pleiotropic, with many occurring in regions with limited sequence divergence that would be overlooked in acceleration-based approaches. We highlight specific examples where predicted regulatory divergence is attributable to alterations in transcription factor binding sites. Overall, our results reveal extensive cis-regulatory divergence in the human lineage and establish a computational framework for prioritizing variants underlying human-specific traits and disease risk.

23

The Ultima Ancient DNA Resource: shotgun sequencing of 25000 ancient genomes

Nadin Rohland¹, Mariam Nawaz¹, David Reich¹, Shop Mallick¹

¹ Harvard University, United States of America

Presenter: Shop Mallick

Ancient DNA data has been dominated by Illumina short read sequencing technology and despite drops in costs by an order of magnitude, shotgun sequencing is often still prohibitively expensive when the endogenous content of a sample is low.

Enriching aDNA libraries for a targeted subset of the genome has been a primary strategy for reducing costs for generating ancient human DNA data, lowering sequencing costs by one to two orders of magnitude per sample, and enabling conversion of ancient samples with <1% endogenous DNA to working genome-wide data. But while >70% of published samples with genome-wide data have been generated with this approach, targeted capture limits analyses such as studies of the site frequency spectrum and is expensive.

Ultima Genomics currently offers the lowest cost-per-read in the Next Generation Sequencing (NGS) market (cost per human genome: Illumina: \$200, UG100: \$80). In a collaboration, we have developed together a tailored workflow, "aDNAv2", optimized for low-cost Ultima sequencing. This workflow performs demultiplexing and alignment directly on the sequencer reducing informatic burden.

We have sequenced more than 33000 libraries from 25000 distinct samples with a median coverage of >1-fold, starting from native Illumina libraries previously determined by in-solution enrichment and Illumina technology to have >5% endogenous DNA. These were converted via a simple PCR step using UG-specific primers. The data are fully co-analyzable without detectable bias with Illumina shotgun data and target-enrichment data. We plan to release these data to provide an "Ultima Ancient DNA Resource" (UADR). The ancient samples that our community are collectively studying represent a shared resource of world-heritage importance, and we have an obligation to read out the libraries we produce in as rich detail as possible. The generation of shotgun data of most of our catalog of libraries contributes meaningfully to making the unique resource more valuable for scholars, for the public that supports this research, and for stakeholders with connections to the ancient individuals being analyzed.

24**Large-scale genealogies of ancient and modern genomes**Pier Palamara¹¹ University of Oxford, United Kingdom

Presenter: Pier Palamara

Large collections of modern and ancient genomes provide complementary insights into recent human history, but integrating them at scale remains challenging. We developed a method called Threads to infer large-scale ancestral recombination graphs (ARGs), and extended it to integrate unphased, low-coverage ancient genomes into ARGs of modern individuals. These approaches combine scalable haplotype matching with probabilistic inference of coalescence times. We applied these methods to 4,842 ancient genomes and 487,409 UK Biobank individuals. The resulting genealogies reveal fine-scale geographic patterns of ancient–modern genealogical relationships associated with demographic transitions in Britain over the past few thousand years, including distinct signals for pre-Saxon, early medieval and medieval populations. Genome-wide patterns of recent shared ancestry also track local variation in coalescence rates and are associated with complex-trait heritability.

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mixKIN: A new approach to estimate genetic kinship in admixed populations

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¹ University of Lausanne, Switzerland

Presenter: Muhammed Siddik Kiliç

Genetic kinship estimation is one of the key social and biological mechanisms that helps us understand past populations and reconstruct prehistoric social structures. However, inferring biological relatedness from ancient DNA (aDNA) is challenging due to low sequencing coverage and population structure. Most existing approaches assume homogeneous background relatedness, leading to incorrect kinship inferences in admixed or heterogeneous populations. Furthermore, while some existing methods designed to account for population admixture, they still underperform with highly heterogeneous datasets. Conversely, haplotype-based methods are robust to population structure but require higher coverage genomes, making them inapplicable for a substantial number of aDNA individuals. We developed mixKIN to address these limitations. mixKIN relies on the Pairwise Mismatch Ratio (PMR) and explicitly incorporates individual ancestry components into the estimation. We validated our method using simulated datasets comprising pedigrees of both unadmixed and admixed individuals under different demographic models, and across coverages from 1x down to 0.03x (~1000 overlapping SNPs). Evaluating classification performance with the distance-aware Uniform Ordinal Classification (UOC) Index showed that mixKIN consistently outperforms alternative tools in admixed populations and performs comparably in homogeneous populations. We also applied mixKIN to an empirical dataset from a cemetery in seventh-century England involving an individual with West African ancestry, demonstrating that our method also works in real case scenarios. Finally, our approach enables more reliable genetic kinship inferences, overcoming methodological limitations that previously obscured these relationships in past communities.

26**Modelling temporal decay of shared haplotypes between distinct populations**

Harald Ringbauer¹, Florence Pautet¹

¹ Max Planck Institute for Evolutionary Anthropology, Germany

Presenter: Florence Pautet

Related individuals are likely to inherit long identical haplotypes from a common ancestor. These segments, called identical by descent (IBD), provide valuable insights into the recent demographic history of the studied populations. While IBD-based methods are well-established for contemporaneous genomes, advances in ancient genomics introduce distinct sampling times, which most current models do not accommodate. Here, we present a theoretical framework describing the decay of IBD sharing between an ancestral population and its descendants, driven jointly by recombination and coalescence. We derive a closed-form description in which IBD sharing across generations depends only on the IBD distribution within the first generation and on the number of generations elapsed, but not on any demographic change in between. This model extends results previously obtained for constant population sizes. It can further be adapted to encompass admixture and migration. We first verified our formulation on simulated data, and then applied it to empirical ancient DNA datasets to uncover new insights into past population history. Our approach represents an advance in the understanding of how IBD segments arise and evolve.

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Assessing biases of using aDNA imputation for natural selection scans

Jose Rodrigo Flores Espinosa¹, Anders Eriksson², Liisa Loog¹

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² University of Tartu, Estonia

Presenter: Jose Rodrigo Flores Espinosa

Reconstructing human evolutionary history from present-day genetic variation is challenging, as recent demographic processes often confound signals of past selection. Ancient DNA (aDNA) has enabled allele frequency changes to be estimated through time, allowing population-level and haplotype-based analyses of evolutionary processes. However, the fragmentary nature of aDNA results in high levels of missing data, typically requiring genotype imputation using present-day reference panels. Although imputation can be reliable for demographic analyses based on common variation, its accuracy declines with increasing genetic distance between the target sample and the reference panel. This limitation is particularly pronounced in regions affected by selection, where linkage disequilibrium dynamics during selective sweeps result in haplotype structures poorly represented in present-day reference panels, leading to biased inferences about past selection events, as illustrated by previous conclusions on the selection timing at the FADS and LCT loci.

In this study, we examine how genotype imputation of ancient DNA (aDNA), commonly performed using reference panels composed of present-day individuals, may introduce biases into the inference of natural selection. Using forward-in-time simulations of human populations under a range of selection scenarios and levels of data missingness, we quantify imputation concordance across time and along the length of loci experiencing selection. We find that strong selection substantially distorts imputation concordance, particularly in genomic regions surrounding the variant under selection and in aDNA samplings predating the onset of selection. At the same time, strong selection leads to rapid haplotype homogenization in populations, increasing concordance relatively soon after selection events begin. In contrast, weaker selection results in a more gradual concordance decline, with imputation biases persisting over longer timescales. These findings underscore the need to account for selection history when using aDNA imputation for selection inference, to avoid misleading inferences about allele frequency trajectories and the timing of adaptive events.

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Adaptive introgression in Peruvians reveals temporally distinct waves of selection on archaic variation in South AmericaXinjun Zhang¹¹ University of Michigan, United States of America

Presenter: Xinjun Zhang

Adaptive introgression introduced deeply diverged archaic genetic variants into modern human populations and evidently facilitated local adaptation in diverse environments. Although multiple candidate loci have been reported in American populations, the evolutionary landscape of adaptive archaic variation in the Americas remains poorly resolved, in part because demographic complexity, including severe bottlenecks, founder effects, and recent post-Columbian admixture that all obscure genuine signals. Previous work has also focused primarily on identifying candidate loci rather than reconstructing the dynamics of selection acting on archaic haplotypes. Here, we systematically reevaluate previously reported candidates of adaptive introgression in Peruvians, who carry the highest proportion of indigenous American ancestry among populations in publicly available datasets, using a framework integrating introgression statistics, haplotype-based visualization, selection scans, ancient DNA, and ancestral recombination graph-based inference of selection history. We refine hundreds of putative candidates into 20 high-confidence loci with convergent evidence of adaptive introgression ($s = 0.012-0.055$), including established candidates and previously underappreciated regions. Reconstruction of selection histories reveals that adaptive introgression in Peruvians occurred across at least two temporally distinct waves: an earlier wave (8.7-14.5kya) overlapping with the peopling and settlement of the Americas, and a more recent wave (2.9-5.8kya) around the Andean agricultural transition. These onset times and ancient DNA evidence confirm that all candidates were inherited from ancestral Beringian populations rather than introduced through post-Columbian admixture. Functional enrichment implicates pathways related to immune signaling, lipid metabolism, vascular biology, and inflammation. Notably, among the recently selected loci, *CXCR4* emerges as a compelling candidate of immune-related adaptive introgression. Our results suggest that archaic introgressed haplotypes acted as a long-term reservoir for adaptation that remained evolutionarily important throughout the history of American populations.

29**Timing is everything: Delayed transitions enable human neural expansion**Madeline Lancaster¹¹ MRC Laboratory of Molecular Biology, United Kingdom

Presenter: Madeline Lancaster

The human brain is greatly enlarged and complex, even when compared with our closest living relatives, the other great apes. How this evolutionary elaboration has come about is still unclear, but neural organoids are enabling comparative studies to uncover the underlying mechanisms. Our recent findings highlight delayed developmental transitions underlying this increased size and complexity. In particular, we have uncovered a delay in neuroepithelial transition that enables increased progenitor expansion and tissue growth. In addition, human neurons themselves are more elaborate with longer range projections, and we have similarly discovered a delayed transition, in this case in timing of when axons stop growing. Our investigations of the mechanism of these delays have uncovered key differences in processes regulating cell shape and calcium signaling. These studies are revealing crucial differences in timing of the duration of cell states, thus enabling increased tissue growth and neuronal complexity during human brain development that can explain how and why humans have such large, elaborate brains.

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Subsistence-associated DNA methylation signals in the transition to farming

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Presenter: Youssef Tawfik

The transition to farming represents one of the major ecological and cultural shifts in human history. It induced profound changes in subsistence and most likely reshaped functional genomic variation among early agricultural and subsequent populations for which signs of genetic selection associated with diet, immunity, metabolic processes, and shifts in polygenic scores for body height, pigmentation, and other traits could be identified. While allele-frequency shifts reflect long-term adaptation, regulatory plasticity (i.e. epigenetic mechanisms such as DNA methylation) can be seen as a direct response to rapid shifts in subsistence such as the Neolithic transition, which go largely unnoticed in current analyses. Here, to address this, we compiled a dataset of 42 high-quality ancient genomes, divided into two subsistence-based groups, in a comparative DNA methylation analysis. After rigorous filtering we identified 458 CpGs with significantly differentiated methylation levels between hunter-gatherers and early farmers. Many of these CpGs were located in genes with functions involved in skeletal development, metabolic regulation and energy homeostasis, and immune signaling, overlapping with genetic variation identified in previous selection scans and palaeogenetic studies regarding the Neolithic transition. We demonstrate that regulatory signals related to major ecological shifts can be identified from ancient methylomes, thus providing a foundation for further comparative investigations across different populations, time periods, and eco-regions.

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Worldwide patterns of selection and differentiation in genes linked to alcohol metabolism

Francesca Carollo¹, Hélios Delbrassine¹, Massimo Mezzavilla¹, Luca Pagani¹

¹ University of Padova, Italy

Presenter: Francesca Carollo

The consumption of fermented beverages is widespread across human societies: alcohol has served a relevant purpose in countless cultures through time, playing major societal, political and even religious roles in them for millennia. Yet, little is known about the impact of such a culturally transformative behavior on the genetic make-up of human populations worldwide. Previous studies largely focused on alcohol dehydrogenase genes (ALDH1B) in East Asia or East Africa, leaving the broader evolutionary history of alcohol metabolism in populations worldwide largely unexplored.

Here, we analyze genetic variation across all known genomic regions involved in alcohol metabolism (for a total of 3,921 SNPs) in a global panel of 140 present-day human populations. We identify both private and shared patterns of Hardy-Weinberg deviation and linkage disequilibrium across geographic macroareas, as well as signals of natural selection acting on genomic regions beyond ALDH1B. Specifically, South Asia exhibits the highest number of SNPs under selection, but we find statistical support for only a single private signal. In contrast, Africa presents the greatest number of private signals, with 24 SNPs identified as being under selection exclusively within this macroarea.

By further integrating data from 596 ancient human genomes with worldwide palaeoclimatic reconstructions spanning the last 10 thousand years, we link these signals to environmental changes through time and find weak evidence of correlation between climatic conditions and selection signals.

Moreover, we detect contrasting directionalities of selection across geographic areas. In some regions, natural selection favored alleles that enhance the ability to metabolize alcohol and its byproducts, consistently with increased tolerance to alcohol consumption. In others, selection acted in the opposite direction, favoring alleles that confer protection from the adverse effects of alcohol consumption by promoting an avoidance strategy and disfavoring those alleles that increase metabolic digestion of alcohol.

Overall, by combining genomic, environmental and archaeological evidence, we provide new insights into the gene-culture coevolutionary dynamics that shaped alcohol metabolism in humans.

Francesca Carollo is the recipient of an EMBO|EMBL Symposia fellowship, jointly funded by EMBO and the EMBL Advanced Training Centre Corporate Partnership Programme.

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Detecting human-specific DNA methylation changes through comparative analysis of great apes and ancient hominins

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² The Hebrew University of Jerusalem, Israel

Presenter: Chen Leibson

Human lineages, including anatomically modern and archaic humans such as Neanderthals and Denisovans, exhibit profound morphological and behavioral differences from nonhuman African apes. Because regulatory divergence is thought to be a primary driver of phenotypic evolution, we sought to identify epigenetic shifts occurring since the human-chimpanzee split.

Utilizing DNA methylation as a proxy for gene expression in ancient samples, we compared methylation maps across modern humans, archaic humans, and nonhuman African great apes. We experimentally generated whole-genome methylation maps from the bones of a gorilla, two chimpanzees, and six modern humans. These were integrated with reconstructed profiles from a Neanderthal, a Denisovan, and 28 high-coverage ancient modern humans, supplemented by methylation array data.

By applying stringent filtering criteria, we identified over 300 differentially methylated regions (DMRs) that distinguish humans from nonhuman African apes. These DMRs are most significantly enriched in pathways related to bone remodeling and heart formation and function. Furthermore, by using the gorilla as an outgroup, we identified dozens of DMRs specific to the chimpanzee lineage. Finally, we investigate the underlying mechanisms of these epigenetic changes, including their association with specific genomic features and the evolutionary gain or loss of CpG sites. These results provide insights into the regulatory landscape of human evolution.

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Centromere-spanning haplotypes reveal deep ancestry and evolutionary dynamics in the human genome

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Presenter: Karen Miga

Centromeric satellite arrays play an important role in nuclear organization, kinetochore recruitment, and division fidelity. Genetic and epigenetic variation within centromeric regions can lead to chromosome instability, which could contribute to aneuploidies and cancers, defects in early development, and decreased fertility. Despite their importance, we are only beginning to understand the architecture of these regions and the patterns of evolution across human populations. Advances in long-read sequencing and assembly now enable the generation of complete telomere-to-telomere (T2T) diploid human genomes, providing access to previously unresolved regions. A substantial fraction of newly resolved sequences lies within constitutive heterochromatin, which suppresses meiotic recombination and results in extended linkage disequilibrium (LD) across centromeres. These regions, defined as centromere-spanning haplotypes (cenhaps), are largely defined by large, satellite DNA arrays, flanking segmental duplications, and contain hundreds of genes in centromere-proximal regions, many previously associated with human traits. Due to reduced recombination, cenhaps preserve long-range haplotypes that persist across deep evolutionary timescales. Population-scale comparisons indicate that many cenhaps coalesce over a million years ago, with a subset showing evidence of Neanderthal and Denisovan introgression. These patterns reflect long-term haplotype persistence shaped by low recombination, drift, and selection.

We analyzed cenhap structure and diversity across 470 phased human haplotypes generated by the Human Pangenome Reference Consortium (HPRC), identifying thousands of cenhaps using fully resolved diploid assemblies. These long-range haplotypes are strongly associated with specific centromeric satellite array structures. Leveraging both alignment-based and k-mer-based approaches, we establish a cenhap-informed framework for centromere annotation and functional genomic analysis. This includes applications to genome-wide association studies (GWAS), enabling improved variant detection and interpretation in pericentromeric regions. In doing so, we identify rare recombination events within peri/centromeric regions and estimate local mutation and repeat turnover rates. Selection signals are detected in specific haplotype backgrounds, and gene content within archaic cenhaps includes loci previously implicated in disease. These results demonstrate that cenhaps offer a tractable system for evolutionary inference, capturing ancestral structure, mutational dynamics, and long-range haplotype persistence, many representing deep lineages that are common in extant human populations.

34**Tracing the early evolutionary stages of plague through ancient genomics**

Maria Spyrou¹

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Presenter: Maria Spyrou

Over the past decade, ancient DNA research has reshaped our understanding of the evolutionary history of plague, caused by the bacterium *Yersinia pestis*. The identification of *Y. pestis* in prehistoric archaeological contexts across Eurasia, dating back as early as 5,500 years ago, has revealed that the pathogen circulated among human populations millennia before the historically documented pandemics. At present, most available evidence suggests that the majority of prehistoric lineages lacked the genetic features required for efficient flea-mediated transmission and have no known present-day descendants. These findings point to a complex evolutionary history, with multiple *Y. pestis* lineages coexisting from the Neolithic through the Iron Age. The earliest evidence for flea-adapted strains dates to the Middle and Late Bronze Age in Europe, although their broader geographic distribution remains uncertain. At the same time, the epidemiological behaviour and population-level impact of these diverse prehistoric lineages remain poorly understood. In particular, the role of plague in episodes of prehistoric population decline, as well as the mechanisms of transmission and disease presentation associated with non-flea-adapted strains, remain open questions. Here, I present new genomic data that expand the known geographic and temporal diversity of prehistoric *Y. pestis* lineages and examine these findings in the context of existing ancient DNA evidence, archaeological context, and historical data. Together, these findings raise broader questions about the diversity, evolution, distribution, and impact of plague in prehistoric Eurasia.

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De novo assembly of ancient metagenomic data from dental calculus to disentangle the Neolithic transition in Southern Europe

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² University of Florence, Italy

Presenter: Patrícia Santos

The interest in ancient metagenomics is increasing with over 1.700 ancient microbial genomes, partial genomes, and metagenomes characterized. However, most studies focused on aligning sequences to modern reference genomes, limiting discoveries to known taxa and introducing a reference bias, as ancient and modern genomes differ genetically. A way to overcome this is through *de novo* assembly, which reconstructs genomes of all species present in a sample. We evaluated multiple *de novo* assembly pipelines using simulated ancient metagenomes and identified the optimal pipeline for reconstructing ancient metagenome-assembled genomes.

We applied this approach to ancient metagenomes covering the Neolithic transition, a major cultural and demographic event in Europe that affected both human evolution and the oral microbiome. We collected published metagenomic data from different regions in Europe ranging from the Palaeolithic to the Bronze Age, along with newly sequenced samples from Italy. We focus on reconstructing the ancient genome of *Anaerolineaceae* oral taxon 439, a bacterium found at higher abundance in the oral microbiome of Neolithic farmers. Through genome reconstruction, we investigated its genetic variability and perform functional annotation to identify genes that may have been lost/gained and can have an impact in the metabolization of food/medicines.

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Tracing the emergence and global spread of hepatitis B virus through human history

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Presenter: Ruth Boersma

Hepatitis B virus (HBV) remains a major global public health burden, with approximately 3% of the world's population currently infected. Because HBV transmission occurs through close contact and often within familial networks, its evolutionary and geographic history is closely intertwined with that of human populations. Accordingly, the modern HBV genotype distribution exhibits distinct ethnogeographic patterns that are thought to reflect past human mobility.

The analysis of ancient DNA samples has provided deep insights into HBV diversity and distribution over the past 10,000 years across multiple continents, revealing extinct lineages and documenting the historical presence of modern genotypes across different regions. Nevertheless, important aspects of HBV phylogeographic history remain unresolved, including the spatiotemporal emergence of HBV in human populations, the origins of modern HBV genotypes, and the migration dynamics underlying the virus's spread between world regions.

To address these gaps, we reconstructed novel ancient HBV genomes from 268 archaeological sites across 43 countries. Combined with previously published genomes, these data constitute an extensive spatiotemporal representation of past HBV diversity, enabling the application of sophisticated phylogeographic models to reconstruct ancestral locations along the viral phylogeny and quantify migration rates between regions. Initial analyses reveal that past spatiotemporal distribution of some HBV genotypes was broader than previously recognised. Integrating ancient and modern genomes, we reconstruct the phylogeographic history of HBV, tracing its emergence in human populations, its global spread alongside human migrations, and the processes that shaped the present-day distribution of HBV genotypes.

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Genomic evidence of *Yersinia pestis* from the Eastern Mediterranean and global phylogenomics of plague emergence

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Presenter: Aditya Upadhyay

Plague, caused by *Yersinia pestis*, has caused tens of millions of deaths across three historical pandemics. Despite historical accounts placing the origin of the First Pandemic (541–750 CE) at Pelusium in northeastern Egypt, archaeogenomic evidence has been restricted to Western and Central Europe, 2,500–3,500 km from the proposed epicentre. Here, we report the first *Y. pestis* genomes from the Eastern Mediterranean, recovered from dental remains at a mass burial in the Roman hippodrome of Jerash (ancient Gerasa), Jordan, dated stratigraphically to the late 6th–early 7th century CE. Genomic analysis of eight samples from five individuals (1.25×–35× coverage relative to the *Y. pestis* CO92 reference) confirmed authentic ancient DNA based on characteristic fragmentation, damage patterns, and phylogenetic placement within First Pandemic lineages. Average nucleotide identity analysis using two independent pipelines showed that all Jerash isolates represent a single, highly conserved strain (≤ 1 base per million divergence), consistent with a localized epidemic affecting individuals of diverse origins. To contextualise these findings, we performed a global phylogenomic analysis of 326 *Y. pestis* genomes spanning the Neolithic to the present, integrating 68 ancient and 258 modern strains. Phylogenetic reconstruction reveals that pandemic lineages emerged independently from deep Eurasian reservoirs rather than from a single ancestor. No global molecular clock is observed, with long periods of evolutionary stasis punctuated by rapid divergence during epidemic phases. All ancient pandemic genomes derive exclusively from human hosts, contrasting with the broad host range of modern lineages. Together, these results provide the first genomic anchor for the First Pandemic in the Eastern Mediterranean and demonstrate that integrated ancient–modern genomic analyses are essential for reconstructing pathogen evolution.

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Reconstructing the epidemiological landscape of Venice's plague hospital

Emily Gaul¹, Tatiana Tondini¹, Caitlin A. Mitchell¹, Orhan Yavuz¹, Kim-Louise Krettek¹, Ella Reiter¹, Antonino Vazzana², Carla Figus³, Matteo Romandini³, Vittoria Bianchi³, Philip Slavin⁴, Stefano Benazz³, Cosimo Posth¹, Maria Spyrou¹

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Presenter: Emily Gaul

In recent years, ancient DNA research has greatly advanced our understanding of the evolutionary history of *Yersinia pestis*, the aetiological agent of plague. The lineage responsible for the Black Death is now known to have originated in central Eurasia, from where it spread to Europe, undergoing frequent resurgences over a period of approximately five centuries. Yet a key challenge remains in integrating observed *Y. pestis* diversity within multidisciplinary narratives of intra-pandemic disease dynamics. To this end, ancient metagenomics provides a unique opportunity to explore collective disease burdens, thereby situating plague within the broader epidemiological landscape of affected societies. Here, we present the case study of the Lazzaretto Vecchio, which served as a site of treatment and quarantine for those afflicted with plague in Venice from 1423 until the mid-17th century CE. A total of 170 individuals recovered from tomb and mass burials at the site were subjected to bioanthropological, isotopic, and metagenomic analyses, yielding detailed insights into the disease's victims across two centuries. Importantly, pathogen screening revealed a diverse microbial presence, suggesting that plague occurred within a complex landscape of co-occurring infectious diseases. Genome-wide *Y. pestis* data generated for 37 individuals show that the recovered strains fall into four distinct genetic clusters, consistent with successive waves of plague affecting the city. Furthermore, observed patterns in the spatial distribution of *Y. pestis* diversity across the mass graves enabled the assessment of demographic differences among victims associated with distinct outbreaks, including age, sex, genetic ancestry and detected co-infections. Our findings underscore the value of multidisciplinary approaches in facilitating high-resolution perspectives on *Y. pestis* evolution and its interplay with host population dynamics during past plague pandemics.

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Uncovering the drivers of pathogen evolution from ancient dna to modern disease

Lucy van Dorp¹

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Presenter: Lucy van Dorp

Understanding the processes that drive pathogen emergence, outbreaks and epidemics remains a major challenge, particularly for zoonotic pathogens where reservoir hosts and the directionality of cross-species transmission can be difficult to resolve. Historical and ancient genomes from humans and other animals provide a temporal framework for investigating how pathogen evolution and spread have been shaped by changing host ecologies and human activities, including trade, mobility and animal management. I will present examples from our work integrating ancient DNA, pangenomic and phylodynamic approaches to reconstruct pathogen diversity and transmission across hosts and through time. These studies demonstrate how temporally sampled genomes can reveal changes in population structure and host association, resolve the timing and direction of transmission, and link pathogen dispersal and epidemic dynamics to human actions. Connecting ancient and modern genomic diversity can therefore help us understand the processes that generated past outbreaks and better contextualise the pathogens that continue to challenge human and animal health today.

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Elucidating the true biological signals in enrichment analysisBatel Attias², Benjamin Yakir¹, Liran Carmel²¹ The Hebrew University Mount Scopus, Israel² The Hebrew University of Jerusalem, Israel

Presenter: Batel Attias

Enrichment analysis is widely used in human evolutionary genomics to interpret gene sets derived from comparisons between modern humans, archaic hominins, and other primates by identifying biological terms linked to genes of interest. However, enrichment analyses often produce many significant terms, limiting interpretability. A major source of this redundancy is correlation between terms, caused by extensive overlap in their annotated genes. As a result, correlation-driven false positives can obscure the core biological processes underlying the evolutionary signal.

Here, we present a computational framework for reducing correlation-driven false positives among enriched terms. Our method defines hierarchical relationships between pairs of terms, where the term with the higher hierarchy is expected to contribute more to the enrichment signal. By depicting these hierarchy relationships between terms as a directed graph, we use an energy minimization approach to define a global hierarchy value for each term. This approach prioritizes terms truly enriched within the gene set, while down-weighting terms whose apparent enrichment is largely explained by their overlap with other enriched terms. Because it relies only on gene-term associations, it is broadly applicable across existing annotation databases such as Gene Ontology and KEGG. In simulations, our method outperforms widely used enrichment approaches in both accuracy and precision across a broad range of term-correlation levels. We apply the framework to gene lists produced in previous human evolution studies, such as those located in Neanderthal introgression deserts, to re-evaluate enriched biological processes and highlight terms that directly capture differences between hominin groups.

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Ceramic Age genetic relationships of ancient individuals from the Dominican Republic

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Presenter: Judith Ballesteros Villascán

Around 2,800 years before present (BP), the Caribbean saw the emergence of new ceramic techniques alongside growing cultural practices. Although broad settlement patterns during this period are relatively well understood, archaeologists and geneticists are still debating the extent of population connections across the region.

In this study, we investigated pre-Columbian genetic relationships in the Caribbean by focusing on two contemporaneous sites in the northern Dominican Republic: El Carril and El Flaco (~750–500 BP). These geographically close but archaeologically distinct sites provide an opportunity to explore how cultural differences relate (or not) to the genetic structure. We generated and analyzed data on around 1.2 million genomic positions from 29 ancient individuals across both sites. Integrating genomic, isotopic, radiocarbon dating, and archaeological contextual data, we compared our findings with published data from nearby and more distant sites. We applied established kinship methods, including the analysis of identical-by-descent (IBD) fragments, to infer biological relatedness.

Our results reveal patterns of genetic connectivity within and between sites, as well as among Ceramic Caribbean genetic subclades, indicating varying degrees of population interaction. By relating these patterns to geographic distance and sex, we show changes in interaction networks and pre-Columbian demographic structures in the Dominican Republic. By doing so, we provide new evidence to the ongoing debates about Caribbean population dynamics before Spanish invasion.

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Convergent human adaptation during parallel subsistence transitions across Eurasia

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Presenter: Alison Barton, David Reich

The adoption of agriculture and pastoralism transformed human diets, environments, and disease exposures, yet the evolutionary consequences of these transitions remain poorly understood outside West Eurasia. We analyzed ~7 million variants in 1,862 ancient and 610 present-day East Eurasians spanning the last 10,000 years to characterize recent adaptation and compare patterns of selection across Eurasia.

Using a generalized linear mixed model to control for population structure, we identified 40 genome-wide significant signals of selection, including previously reported alcohol metabolism loci. *ADH1B* provides one example of parallel adaptation across Eurasia; selection on rs1229984 in *ADH1B* occurred independently in East and West Eurasia with similar timing (~3000 BP), suggesting a shared selective pressure associated with the adoption of food production. This convergence provides new insight into the long-standing question of what drove selection at *ADH1B*. In contrast, the *ALDH2* variant rs671 rose only recently in East Asia (~1500 BP). Additional signals at *FADS1/2* and *MUC2/MUC5AC* indicate convergent adaptation in dietary metabolism and immune function.

East and West Eurasia exhibit correlated adaptive trajectories at both individual variants ($r > 0.67$; $P < 1.98 \times 10^{-6}$) and polygenic traits ($r > 0.67$; $P < 3.02 \times 10^{-6}$), including shared selection against variants associated with increased body fat percentage today. Functional enrichment analyses reveal similar trait categories under selection, particularly immune and cardiometabolic phenotypes. These signals replicate in individuals with high East Asian ancestry, supporting robustness to admixture. In contrast, pigmentation evolution followed distinct regional trajectories: while West Eurasians experienced recent selection for lighter skin pigmentation, increases in depigmentation alleles in East Asia largely predate the Holocene.

Together, these findings suggest that agriculture and pastoralism generated common selective pressures related to diet, pathogen exposure, and lifestyle, leaving convergent signatures despite diverse ecological and cultural contexts across Eurasia. Comparing trajectories among populations may help identify the forces that shaped human genomes and continue to influence health today.

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Deletion linked to male infertility found in two neanderthal Y chromosomes

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Presenter: André Bourbonnais

The human Y chromosome is crucial for male fertility. But its repetitive and complex structure, combined with the challenges of ancient DNA makes the archaic Y chromosome hard to study. This leaves a key question open: why is there no trace of the archaic Y chromosome in modern humans despite extensive admixture? In other words, why has it seemingly been selected against?

One possible explanation for a difference in fitness lies in the ampliconic regions. These regions contain large sequence duplications carrying genes only expressed in the testes. In modern human males, losing copies of these ampliconic genes is linked to lower sperm counts, and larger deletions can completely stop sperm production.

To determine whether archaic humans differed in copy number, we estimated the ampliconic copy number in two Neanderthals and one Denisovan, using published high-coverage shotgun aDNA from the Altai Mountains. To handle the sequence complexity of chromosome Y, we mapped reads to a custom human telomere-to-telomere reference genome (chm13v2.0) where we masked all but one copy of each amplicon to have each appear only once. We then compared read depth in the amplicon copy regions to the read depth across single-copy regions to estimate the copy numbers and benchmarked the method using simulated ancient data.

We find that both Neanderthals carry the well-documented ~1.6 megabase gr/gr deletion, while the Denisovan carries a copy number pattern that does not match anything seen before in present day males (~10,000 males). In modern humans the gr/gr deletion is found at low frequency in most populations and is linked to lower or complete stop of sperm production. Finding it in two Neanderthals from two different populations separated by 60,000 years suggests it was common, and possibly fixed. Moreover, a Y chromosome carrying a fertility-linked deletion offers a plausible reason why it was at a disadvantage and why the archaic Y chromosome was selected against.

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From regional population dynamics to local histories: genomic insights from Chitral, Pakistan

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Presenter: Brooke Bowman

Recent genetic studies have started to transform our understanding of population history along the northern frontiers of South Asia, revealing a region shaped by repeated episodes of migration, admixture, and interaction. Much of this history has been reconstructed from Pakistan through ancient genomes from the Swat Valley and genetic studies of the Kalash, a population from the Chitral region in the Hindu Kush whose demographic history has been shaped by long-term small effective population size and genetic drift. These datasets have largely shaped current interpretations of broader regional population dynamics, yet their relevance to other communities within Chitral and their genetic histories remains largely unexplored.

Here, we investigate the fine-scale population structure and demographic history in five tribal communities from the dynamic Chitral district. Although these communities share language, geography, and many cultural practices, they maintain distinct oral traditions describing diverse ancestral origins and historical connections. Integrating genomic data from these tribes with available data from regional ancient and modern populations, provides an opportunity to examine how oral traditions relate to broader patterns of population continuity and demographic change. This framework further allows evaluation of whether present-day diversity reflects shared regional histories or distinct demographic trajectories among groups inhabiting the same geographic landscape.

Rather than treating Chitral as genetically homogenous, this work approaches it as a mosaic of community histories, where shared geography does not necessarily imply a shared demographic past. In doing so, it further illustrates how integrating community knowledge and ethnographic perspectives with genomic evidence can enrich our understanding of human history across multiple scales, from broad regional processes to the demographic trajectories of individual communities.

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Mediterranean networks and the human landscape of Roman Southern Iberia

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Presenter: Pablo Carrion

The Roman expansion throughout the Mediterranean Basin brought an unprecedented level of intercontinental connectivity. These new levels of connectivity were accompanied by the widespread dissemination of Eastern Mediterranean-related ancestry, contributing to an increasingly homogeneous genetic landscape across the Mediterranean. Particularly in Iberia, results show that the incorporation of the Iberian Peninsula into the Roman Empire marked an increase in genetic diversity relative to preceding periods, largely driven by gene flow from populations with ancestry linked to North Africa and the Eastern Mediterranean. The social context of this mobility remains incompletely understood, particularly regarding its role in facilitating the movement of cultural practices and religious traditions.

Here, we present bioarchaeological data from 75 individuals recovered from four archaeological sites in Southern Iberia spanning the Roman Imperial period. Genome-wide analyses reveal substantial heterogeneity within Roman-period Southern Iberia, with individuals deriving a significant proportion of North African signal in their autosomal ancestry. At the site of Baelo Claudia, a port city situated on the northern shore of the Strait of Gibraltar, directly across from ancient Tingis, North African-related ancestry constitutes a major component of this community and is associated with a high frequency of E1b Y-chromosome lineages, suggesting that trans-Gibraltar mobility contributed significantly to the genetic landscape of this Roman city. These results highlight Southern Iberia as a key crossroads of population movement within the western Mediterranean and provide new insights into not only demographic processes, but cultural spread accompanying Roman connectivity. By integrating multidisciplinary analyses focusing on bioarchaeological markers of mobility in these unique sites, we aim to reveal the complex relationship between cultural transmission and continuity in Roman Southern Iberia.

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A genetically previously unrecognized central contribution of the southern Congo Basin to the Bantu settlement of South-central Africa

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Presenter: Jérémy Choin

South central Africa lies between the Congo Basin, the Great Lakes region, and southern Africa, and has long served as a corridor for human movement, as shown by regional connections among Later Stone Age foragers and later farming and iron-working communities linked to Bantu languages. However, the degree of genetic continuity between pre-Iron Age foragers and later populations, and the extent to which the settlement of south central Africa reflects multiple Bantu-associated migrations, has not been fully resolved using genomic data. To do so, we generated genome-wide ancient DNA from 70 Iron Age and historical individuals from present-day Zambia and Malawi, and analyzed these genomes with a dense dataset from present-day Africans, especially Bantu speakers. The genomes are predominantly composed of Bantu-related ancestry (>70%), with variable contributions from central rainforest-related, southern forager-related, and eastern African pastoralist-related ancestries. One individual (1516–1667 calCE) from Kalala Island in the Kafue River, carries 40% non-Bantu-related ancestry that closely matches local Later Stone Age foragers. Admixture dating for this individual places the event 850 years ago, several centuries earlier than the 420-year-old admixture previously reported for present-day BaTwa from the same region, suggesting additional gene flow after this person's lifetime. Focusing on the Bantu-related ancestry, haplotype-based analyses identify two main clusters among Iron Age, historical, and present-day south central Bantu groups that mirror geography: a western (cluster 2) and an eastern (cluster 1) cluster. Cluster 2 is predominantly associated with Bantu-speaking populations from the Bié Plateau in central Angola and, to a lesser extent, south central Democratic Republic of Congo (DRC), whereas cluster 1 is primarily associated with Bantu-speaking groups from south central DRC (Kasaï region). These patterns highlight southern DRC as a key corridor and source region in the Bantu-related peopling of south central Africa and point to a complex, multi-source demographic history in which forager-related ancestry persisted locally and at least two Bantu-associated migrations shaped the region's genetic landscape, consistent with archaeological and linguistic data.

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New methods for Y chromosome lineage analysis in ancient DNA applied to the Slavic dispersal

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Presenter: Leo Cooper

We present a suite of analytical methods for extracting demographic signal from Y chromosomal lineages in archaeogenetic datasets and apply them to the question of Slavic origins. Using callYsnps, a pipeline for accurate automated SNP calling and terminal haplogroup determination from ancient BAM files, we generated Y chromosome haplogroup calls for all genetically male individuals (n=2,930) in a dataset of ~5,100 ancient Eurasian genomes from the late Bronze Age to the modern period. We evaluated coalescence across 257 time-depth thresholds (0–10,000 ya) within the optimized PHATE embedding space. To link paternal lineages to populations defined by IBD-sharing communities, we developed a framework that applies one-vs-rest Fisher's exact tests across a series of coalescent horizons on the YFull phylogeny, identifying lineages that are over-represented among paternal lines in a given IBD cluster. Applied to the Central-East Europe (CEE) IBD group—composed predominantly of archaeologically Slavic individuals—this approach identifies eight Y-lineages that are significantly enriched in CEE and whose most recent common ancestors lived in or after the EIA.

To characterize the demographic histories of these lineages, we data mined the FamilyTreeDNA Discover tree, the largest human Y chromosome phylogeny, extracting node topology, TMRCA estimates, and geographic distributions for 21 lineages across ~9,700 nodes. From these data we compute lineages-through-time trajectories, polytomy and pulled diversification rates, coalescence probabilities, and a sibling-TMRCA heatmap. A dendrogram based on LTT curves sampled with Dynamic Time Warping shows seven lineages cluster together and with CEE robustly. These distinct networks display convergent demographic trajectories: single Iron Age founders with a strong start ~400 BCE, exponential growth ~200 BCE–200 CE, and steady growth 200–600 CE, consistent with the post-Zarubintsy formation horizon. These contrast with Germanic-associated lineages, which diversified earlier and more gradually. Phylogenetic nesting with ancient individuals traces three to five lineages to East Baltic and East European forest-steppe BA contexts and three others to Balkan BA populations, reconstructing the geographic mosaic of the emerging Slavic paternal gene pool.

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Evidence for a previously unknown major admixture event in the common evolutionary of Neandertals and Denisovans

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Presenter: Trevor Cousins

The standard model of archaic-moderns populations relationships has Neandertals and Denisovans descending from a common ancestral population roughly 550 thousand years ago (ka), and these groups in turn diverging with the main lineage leading to modern humans around 750 ka (Peyr  gne et al. 2025). There is also evidence of admixture events: (a) two or more deeply divergent lineages that separated from a common ancestor more than ~1000 ka (Plagnol and Wall 2006, Ragsdale et al. 2023, Cousins et al. 2025, Loya et al. 2026) before coming back together around ~300 ka (Cousins et al. 2025), (b) 5-10% gene flow from ancestors of modern humans into the ancestors of Neandertals around ~300 ka (Kuhlwilm et al. 2016, Hubisz et al. 2020, Loya et al. 2026), (c) a few percent contribution of a highly divergent population into Denisovans (Pr  fer et al. 2014), and a few percent gene flow from Neandertals and Denisovans to modern humans outside of Africa (Green et al. 2010, Reich et al. 2010). To test if these known events can predict patterns of genetic variation, we used momi2 (Kamm et al. 2020) to fit the joint site frequency spectrum of single nucleotide polymorphisms (SNPs), corrected for the effects of recurrent mutation, in 40 modern West African chromosomes, 2 Neandertal chromosomes, and 2 Denisovan chromosomes. We cannot obtain a good fit with existing models, despite exhaustive exploration of parameter combinations and known events. However, we obtain exquisite fits if - in addition to the previously inferred events - the shared ancestral population of

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Ancient DNA from Paquimé reveals deep regional continuity and interregional connections in Northern Mexico

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Presenter: Roslyn Curry

Paquimé (Casas Grandes), a major archaeological site in northern Mexico occupied between approximately 1200 and 1450 CE, was a key center of interaction linking the greater U.S. Southwest, northern Mexico, and Mesoamerica. To investigate the population history of its inhabitants, we generated genome-wide ancient DNA data from approximately 60 individuals and analyzed these data alongside present-day reference populations across the Americas and ancient individuals from northern Mexico, the U.S. Southwest, and adjacent regions. Principal component analysis, ADMIXTURE, and f-statistics show that most Paquimé individuals fall within a broader Northwest Mexico / U.S. Southwest genetic continuum, with close affinities to individuals associated with key archaeological sites and cultural traditions across the region, including Cerro de Trincheras, La Playa, Tayopa sites, and Cuevas de los Muertos Chiquitos. The northern Mexican ancestry component observed at Paquimé dates back at least 5,000 years before present and is related to ancestry found in the present-day O'odham (Pima) and, potentially, other present-day Uto-Aztecan language-speaking groups in the region. Together, these observations suggest substantial regional continuity rather than large-scale population replacement from Mesoamerica. At the same time, we identify Mesoamerican-related ancestry in some individuals, indicating genetic heterogeneity within Paquimé and supporting archaeological models of the site as a major center of exchange, mobility, and interaction. These findings show that Paquimé's population history reflects deep regional ancestry alongside interregional connections, refining models of demographic history across this culturally and ecologically connected region prior to European colonization, while demonstrating the value of community-engaged ancient DNA research.

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Establishment of power structures in Late Roman – Migration Period Lithuania: the case of Taurapolis chieftain

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Presenter: Ingrida Domarkiene

At the Taurapolis barrow cemetery in northeastern Lithuania, a richly furnished warrior burial was uncovered. The individual had been interred with a horse, a spatha sword in an elaborate scabbard, other weapons, a drinking horn, and numerous silver and gilded ornaments. In nearby barrows, three other less richly equipped warriors were interred with horses. Evidence suggests that the cemetery originally contained even more graves belonging to the members of the possible military elite. The burials date back to the Late Roman–Migration periods. Although the interments followed local burial customs, the grave goods indicate far-reaching connections, including Scandinavia, the Carpathian Basin, the Balkans, and the northern shores of the Black Sea. The prevailing interpretation has been that these individuals represented a military chieftain and his retinue, either as immigrants or as local Balts who had participated in long-distance military campaigns. However, radiocarbon dating [1] suggests that the burials represent an elite group interred over several generations, while strontium isotope analysis [2] indicates that most of the individuals were of local origin. Genetic analysis (mtDNA, Y chromosome haplogroups, genetic similarity, admixture, IBD-sharing) supports evidence of local origin. Nearby, up to 40 kilometers apart from Taurapolis, lie the Pavajuonis-Rėkučiai and Liūlinė III barrow cemeteries. The burials of male and female individuals found in these cemeteries are linked to the chieftain of Taurapolis not only by close biological relatedness but also by similar dating (about 250–420 cal AD) and burial rituals, and the Pavajuonis-Rėkučiai burials also stood out for their rich grave goods, including imported items made of precious metals. This evidence would confirm local marriage practices and suggest a familial nature of the elite. In this kinship group, males most likely shared high status with females and children, which indicates a system of hereditary status. Meanwhile, no biological relatedness has been identified between the chieftain and the other warriors of Taurapolis. It is possible that the ruling clan and members of their unrelated retinue were interred in this cemetery and its surroundings over several generations.

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Estimation of archaeological site occupation time spans using genomic admixture and radiocarbon datingAMG Silva¹, Daniel Fernandes¹, Ana Catarina Sousa², Tom Higham³, Ron Pinhasi⁴¹ University of Coimbra, Portugal² University of Lisbon, Portugal³ University of Vienna, Austria⁴ University of Vienna, Austria / Jilin University, China

Presenter: Daniel Fernandes

Archaeological site occupation time span estimates can take the form of quantitative and qualitative intervals. The former can use methods such as radiocarbon dating to obtain temporal calendar intervals that, in some cases, can be fairly precise. For the latter, in turn, broader temporal qualitative intervals can be inferred from the characterization of collected materials and artefacts, the identification of specific physical structures, or the type of burial practices. With the increasing incorporation of genomic analysis in archaeological studies where human remains are present, specifically to study population origins, admixture, and social organisation, radiocarbon dates are frequently obtained from the osteological materials being analysed. This allows for a better integration of the genomic, archaeological, and anthropological data, but also leads to an increased usage of finite and unique osteological material. To overcome this, it is possible to estimate site occupation interval spans using a combination of genomic admixture intervals and radiocarbon dates for calibration, if calendar dates are required. Here, we use genomic admixture date estimation software, such as DATES, to estimate site occupation time spans of Neolithic sites with a new open source bioinformatic analytical tool, named OCCUPOMICS, which allows the user to quickly calculate site occupation intervals and calendar dates by inputting a genomic dataset and any related direct radiocarbon dates. This approach, therefore, leads to a decreased need of radiocarbon dates when site-wise genomic data is available, and saves both unique osteological material and limited financial funds.

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Optimized manifold learning reveals early Slavic-associated ancestry and fine-scale ancient Eurasian structure

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Presenter: Pavel Flegontov

We introduce and validate a manifold-learning framework for visualizing population structure that outperforms traditional archaeogenetic PCA, and then apply it to study Slavic origins. We model genetic variation as a low-intrinsic-dimensional landscape shaped by geography, time, and gene flows, and demonstrate on simulated data subjected to patchy sampling and MAF filtering that the first PCs often fail to provide an optimal view. For each dataset, we explore a wide parameter space including PCA implementations, input dimensionalities, distance metrics in PC space, and several manifold-learning algorithms. Candidate embeddings are ranked by second-level objectives: genetic-distance preservation, IBD-community separation, and high-dimensional neighborhood retention. In an ancient Eurasian dataset, this "deep optimization" screened ~809,000 embeddings and selected a 3D PHATE map. Compared with the PC1–PC2–PC3 space, it retained similar Hamming-distance fit but improved resolution of IBD communities, effectively denoising the PCA visualization. Similar results are sometimes achievable with traditional PCA reference panels, but that approach requires extensive manual curation of the reference panels and is not unsupervised.

Applied to genome-wide SNP data for 5,133 ancient and 2,425 present-day Eurasian individuals, the method resolves West and East European clines obscured by conventional Eurasian PCA and detects a compact Roman/Migration-period cluster within the Slavic-associated IBD community. Integrating this PHATE embedding with IBD, *f*-statistics, qpAdm, Y-chromosome phylogenies, and HapNe trajectories shows that Slavic-associated ancestry was not created by a single 6th-century migration. Its closest source signals derive from East Baltic populations of the 1st millennium BCE, Middle Dnieper ancestry, and West/Central European La Tène-period ancestry. Slavic-associated Y-lineage growth trajectories point to differentiation before ~700 BCE, while the earliest Slavic-cluster individuals appear ~150–550 CE in the Middle Danube, the Lower Don, and the Middle Volga. We therefore place shared Slavic-associated ancestry along the southern forest-zone edge of the Dnieper basin, followed by dispersals beginning centuries before the traditional 6th-century horizon.

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Genetic landscape of the Scythian worldOlga Flegontova², Leonid Vyazov², David Reich¹, Pavel Flegontov²¹ Harvard University, United States of America² University of Ostrava, Czech Republic

Presenter: Olga Flegontova

Understanding fine-scale population structure and mobility in Iron Age Europe requires advanced analytical tools such as IBD networks and optimal visualizations. We focused on characterizing Scythian populations between the Lower Danube and Don basins (LDDB). A foundation of our analysis is a visualization of population structure generated with deeply optimized manifold learning (the PHATE algorithm), with fit to Hamming genetic distance and resolution of IBD-sharing clusters serving as objectives for embedding ranking. Our dataset consisted of 2,903 individuals subdivided into three chronological layers: (1) a pre-Scythian layer, dated to approximately 3500–2800 years before present (yBP), including 13 LDDB individuals; (2) a Scythian layer, dated to approximately 2800–2200 yBP, including 96 LDDB individuals; and (3) a post-Scythian layer, dated to approximately 2200–1400 yBP, including 112 LDDB individuals. The dataset included 44 newly sequenced individuals from Scythian contexts in present-day Ukraine.

The strongest genetic gradient in LDDB dates back to the sparsely sampled Pre-Cimmerian period and manifests as a cline between populations of the Urals and Vistula-basin populations (from the Trzciniec-Komariv, Vysotska, and Lusatian contexts). In the Scythian period, most LDDB individuals lie on a similar gradient, but its western end splits into two branches corresponding to two migration sources: Rhine–Scandinavia, which is more prominent in the LDDB, and the East Baltic. The strongest western genetic influx in Scythian-period LDDB, however, derives from the Balkans and Sicily and appears as a distinct cline. Strikingly, the PHATE-space distribution of Scythian individuals closely overlaps with that of the contemporaneous Sicilian population, including the mass grave associated with the Battle of Himera (480 BCE). The Sicilian set includes Baltic, Uralic, and Caucasus-related outliers, whereas the LDDB set includes Baltic, Caucasus-, Sicily-, and Greek-related outliers. This pattern points to extensive bidirectional mobility between the Scythian world – including major centers such as the large Bilsk settlement, often identified with Gelonus – and Greek colonies. We further examine these patterns using IBD analysis, ADMIXTURE, and fine-grained Y-chromosomal lineage analysis.

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LDate: An expectation-maximization approach for dating ancient genomesDavid Reich¹, Romain Fournier¹, Pier Palamara²¹ Harvard University, United States of America² University of Oxford, United Kingdom

Presenter: Romain Fournier

Understanding the chronology of ancient individuals is crucial for interpreting their history. While radiocarbon dating by accelerator mass spectrometry is the gold standard for determining chronology, its application is often limited by cost and the preservation of collagen proteins required for analysis. Consequently, many samples are dated on the basis of their archaeological context, which can span centuries or be compromised by intrusive burials.

Recombination events can serve as a biological clock to address these limitations, allowing the dating of an individual relative to a reference panel with trusted radiocarbon dates. Specifically, the expected value of the autocorrelation length measured between the genotypes of two ancient individuals is a function of both the demographic history and time gap separating the individuals. Our method, LDate, uses an Expectation-Maximization approach to jointly infer the effective population size history, $N_e(t)$, and the age of each individual. With radiocarbon or archaeological contexts acting as priors on individual ages, the algorithm starts by estimating $N_e(t)$ using an updated version of HapNe-LD that accounts for time gaps between pairs of individuals and admixture histories. It then uses this demographic history to refine the age distribution of each individual, repeating the cycle until convergence.

To demonstrate the potential of our approach, we reanalyzed a dataset of 33 medieval individuals from a Jewish community in Erfurt, Germany. A central question in the original study, whether these individuals lived before or after the 1349 Erfurt Pogrom, was left mostly unanswered due to sparse archaeological evidence and a multi-modal radiocarbon posterior distribution. By applying LDate, we find strong evidence that members of this community lived prior to the pogrom, in tension with the suggestion of the original study that the burials probably all post-dated the pogrom. This work underlines the power of our method to refine historical chronologies in challenging statistical settings.

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Genomic history of urbanization and demography in medieval FlandersNoah Gaens¹, Toomas Kivisild¹, Maarten H. D. Larmuseau¹¹ KU Leuven, Belgium

Presenter: Noah Gaens

The Middle Ages were a transformative period in European history, characterized by rapid urbanization and substantial demographic change. In the region of present-day Flanders, once a rural periphery of the Roman Empire, this transformation led to one of the most urbanised and densely populated landscapes of medieval Europe. This study uses newly generated whole-genome data from 152 individuals across four archaeological sites, including the medieval metropolitan centres of Ghent and Ypres, to explore the genetic structure and demographic dynamics of the region in light of growing urbanisation. By integrating these data with 368 previously published genomes from the Flemish area, we examine patterns of genetic continuity, mobility, and regional differentiation during the medieval period. Our analyses reveal fine-scale inter-regional genetic diversity, with extensive identity-by-descent (IBD) sharing and continuity in western Flanders, but reduced genetic affinity with the eastern part of the region. Despite historical and archaeological evidence for long-distance connections and mobility, signals of gene flow from other European regions remain limited. Instead, our data point to an early medieval admixture event between two distinct ancestral components, followed by regional genetic homogenisation. This pattern suggests that the intense urbanisation of Flanders was shaped by local demographic expansion rather than the convergence of genetically diverse groups from different regions, as long been suggested. This study contributes to the growing field of urban archaeogenetics and provides a key reference point for understanding population dynamics in medieval Northwestern Europe.

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Bioarchaeological analyses of human bone remains from the Remete-Felső Cave (Hungary) diversifies our knowledge on regional Pleistocene and Mesolithic biogeography

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Presenter: Dániel Gerber

In 1969, an excavation was carried out in Remete-Felső Cave, in the vicinity of Budapest, Hungary. Among numerous bone fragments, three human teeth associated with Pleistocene megafaunal layers of 45-35 kya were recovered, while 14C screening of further human remains from the site resulted in some late Mesolithic dates. Despite the teeth were misclassified as Neanderthal for decades, using the state-of-art wet laboratory techniques, such as the minimally invasive soaking extraction method and hybridisation capture (Twist biosciences), we show that they most likely belong to a single, anatomically modern human individual (Remete_Ple1), with one tooth yielding enough endogenous, low-contamination DNA for population genomic analyses. Concurrently, the Mesolithic bone material yielded one good-quality (Remete_Mes1) and one low-quality (Remete_Mes2) Mesolithic genome, marking the first genomic data from western Carpathian Basin hunter-gatherers. According to our preliminary results, Remete_Ple1 represents one of the earliest waves of anatomically modern humans to the region, while Remete_Mes1 provides a glimpse to previously uncharacterised macroregional Mesolithic human population diversity.

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Reconstructing the demographic history of the Romani people through modern and ancient genomes

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³ University of Porto, Portugal

Presenter: Juan Carlos Guevara López

Even though the Roma (also known by the misnomer name of Gypsies) are Europe's largest ethnic minority, much of their history lacks written records and key questions about their past remain unresolved. They left South Asia around 1,500 years ago but little is known about their genetic structure at that moment and their interactions with other human groups along their diaspora. Here, we assess these questions by leveraging whole-genome data from 99 Roma individuals across Europe, analyzed alongside a broad reference panel of both modern and ancient populations of South Asia, the Middle East, the Caucasus and Europe. We used a complementary analysis of allele frequency-based methods (PCA, Admixture, qpAdm) and haplotype-based approaches (ChromoPainter, fineSTRUCTURE, SOURCEFIND) to reconstruct the Romani demographic history at fine scale. Population structure analysis revealed that the Roma form a distinct cluster with higher affinity with some northwest South Asia groups, specifically with a Punjabi group and the Chamar. These populations carry elevated South Asian Hunter-Gatherer (SAHG) ancestral component in comparison to other northwestern groups, which is characteristic of the Scheduled Castes. Distal qpAdm modelling of the Roma confirms this SAHG-shifted ancestry composition and reveals the additional layers of Anatolian Farmer and Western European Hunter-Gatherer components that were incorporated sequentially along their migration westwards. Haplotype-based analysis across 31 population clusters corroborates their affinity with these northwestern groups, highlighting a specific Punjabi cluster, Chamar and other Scheduled Caste clusters as the principal South Asian donors. In West Asia, there is a strong Caucasian and Anatolian contribution whilst there is a near-absence of Levantine and North African ancestry which constrains the migration route. Our results provide a coherent model and fine-scale reconstruction of the Romani demographic history and place their South Asian origins among northwestern communities with elevated SAHG ancestry and revealed a sequential admixture with other populations along the diaspora.

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Comprehensive genetic and bioarchaeological analysis of an exceptionally rich early medieval community

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Presenter: Balázs Gyuris

The Avar Period in the Carpathian Basin, spanning from the middle third of the sixth century to the ninth century CE, is among the most intensively studied early medieval periods through ancient DNA research. Over one thousand individuals have been analysed, in some cases enabling the reconstruction of extensive pedigrees associated with diverse archaeological material culture. Such micro-level analyses have revealed social organisation, kinship structures, and marriage practices at unprecedented resolution. Here, we present the Early Avar Period cemetery of Babarc, where an unparalleled accumulation of wealth is represented within only ~50 graves, making it one of the richest known cemeteries of its time in the Carpathian Basin. The exceptional archaeological assemblage, together with genome-wide genotype data, biological relatedness analysis, anthropological observations, and radiocarbon dating, provides a unique opportunity to reconstruct both the chronological framework of the cemetery and the biological history of the individuals buried within it. The genetic results reveal a newly forming community with direct biological links to Central Europe, Eastern Europe, and the Eurasian steppe. Genome-wide genotype data allow us to infer the geographic and biological origins of the individuals in detail, showing that, despite the limited number of graves, they represent a strikingly broad spectrum of Eurasian genetic diversity. These findings point to far-reaching biological connections and complex patterns of mobility, ancestry, and social integration. Combining genetic and bioarchaeological approaches allows us to trace the chronological accumulation of exceptional wealth within the community, possibly in connection with raids documented in historical sources.

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Ancient-to-modern identity by descent (IBD) analysis using the 23andMe research database

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Presenter: Eadaoin Harney

Detecting identical-by-descent (IBD) segments between ancient and modern individuals offers a high-resolution lens for exploring the genetic legacy, mobility, and, occasionally, the identities of people from the past. However, the efficacy of this approach relies heavily on access to massive modern reference cohorts capable of capturing both distant and close genetic connections.

Leveraging the 23andMe Research Institute database of over 11.5 million consenting participants, we systematically analyzed IBD sharing between present-day individuals and more than 900 publicly available ancient and historical genomes. Here, we present summary statistics and population-level insights derived from this large-scale ancient-to-modern IBD analysis using the method we established in Harney et al (2023). We highlight key case studies demonstrating how these connections can localize the biogeographic ancestry of historical and ancient individuals. Furthermore, we show how the stochastic nature of recombination can preserve unexpectedly long IBD segments between individuals separated by hundreds or thousands of years, emphasizing the need for caution when interpreting extended sharing as strict evidence of recent common ancestry.

Finally, we introduce a new collaborative framework allowing paleogenomic researchers to propose joint analyses. Through this resource, collaborators can partner with 23andMe researchers to compare newly generated ancient DNA datasets against the 23andMe database. By providing detailed, aggregated IBD metrics, such as sharing rates across specific present-day geographic regions and demographic subsets, this initiative enables researchers to quantify regional continuity and trace historical legacies, while preserving participant privacy. We welcome feedback to maximize the utility of this resource for our shared research goals.

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Reconstructing Gloucester's population history over the last 2,000 years through ancient genomics.

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Presenter: Jessica Hueck

The city of Gloucester has served as a major urban centre in Western Britain since its establishment during the Roman period (1st century CE), occupying an important strategic and economic position throughout much of its history. Despite the growing number of ancient genomes from Britain, the genetic history of Gloucestershire remains poorly characterised. This project presents a diachronic ancient DNA investigation of the genetic composition in Gloucester from the Roman period to the present-day, spanning over 2,000 years of recorded history.

Genome-wide aDNA data was generated from individuals dating from the Roman, Medieval and Victorian time periods in Gloucester. Comparative population genetic analyses with published ancient and present-day datasets were used to investigate ancestry and mobility across nearly two millennia of occupation. Preliminary analyses of the Medieval individuals reveal similar genetic ancestry, observed to be akin to contemporary Medieval datasets from Cambridgeshire and other regions of England. This pattern is consistent with substantial genetic similarity among populations across medieval England, potentially reflecting increasing connectivity and population mobility.

Despite broad genetic similarity to other English medieval populations, the Gloucester individuals display subtle differences in ancestry composition when compared to Eastern English genomes, including a slight shift towards Northern European-related populations. These observations may reflect regional differences in patterns of mobility and population interaction within medieval Britain. Ongoing analyses of newly generated Roman and Victorian data will allow us to directly assess the nature and extent of demographic processes in the region across a wide temporal range.

This project provides new insights about the population history of Gloucester and contributes to our understanding of regional genetic variation within Britain.

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A high-coverage genome reveals a young female of Aegean ancestry aboard the Antikythera shipwreck

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Presenter: Lucas Anchieri

Ancient shipwrecks offer insights into past seafaring and exchange, yet we know little about the people aboard because human remains are rarely recovered. As a result, questions about who travelled, where they came from, and how maritime connectivity shaped lives remain difficult to resolve. The Antikythera shipwreck dates to the late Hellenistic period, when movement of people and goods across the Mediterranean intensified. The wreck has yielded an extraordinary cargo of luxury goods, coins, and ship fittings. Yet, the people who travelled with it remain almost entirely unknown. Here, we combine osteological assessment of remains recovered from the site with radiocarbon dating and ancient DNA analysis to shed light on the people onboard the vessel and its ill-fated voyage. Osteological assessment indicates the presence of multiple individuals, including at least two females. Radiocarbon dating places the remains at 76-45 BCE, consistent with the date of the shipwreck. Analysis of a high-coverage genome places her closest to Bronze and Iron Age individuals from present-day Greece. Together, these findings provide a rare glimpse of females moving through late Hellenistic maritime networks, adding a human dimension to this iconic shipwreck and demonstrating the potential of underwater archaeogenomics to bring travellers—not just cargo—into view.

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Delineating multi-layered origins of human populations in Beringia using ancient and present-day genomesSara Jang¹, Choongwon Jeong¹¹ Seoul National University, Republic of Korea

Presenter: Sara Jang

Neolithic populations in Yakutia, including the Early/Middle Neolithic ones associated with Syalakh and Belkachi cultures and the Late Neolithic ones associated with Ymyyakhtakh culture, played a pivotal role in the spread of Siberian ancestry from northeastern Europe to Greenland. While previous studies investigated the genetic legacy of ancient human populations from Yakutia in central/southern Siberians, their relationship with indigenous populations around the Bering Strait (hereafter “Beringian” populations), including the Yukaghir, Chukotko-Kamchatkans, Asian Inuits, and Aleuts, remain elusive. Here, we searched for plausible historical admixture models connecting present-day Beringian populations with ancient ones from Beringia and broader Siberia. We show that Asian Inuits, Aleuts, and Chukotko-Kamchatkans are all adequately modeled as a mixture of the Belkachi-related populations from the Middle Neolithic Yakutia with the First Native American gene pool: for Chukotko-Kamchatkans the Native American ancestry is likely relayed via their mixture with neighboring Asian Inuits. In contrast, Yukaghirs, who live in northern Siberia further away from the Bering Strait, are better modeled as a mixture of the Ymyyakhtakh-related populations from the Late Neolithic Yakutia and neighboring Chukotko-Kamchatkans, delineating the northeastern margin of the Middle-to-Late Neolithic genetic change in Yakutia. This study sheds light on the fine-resolution genetic history of the indigenous populations in Beringia, linking Siberia and North America.

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Ancient genomes illuminate the origins and dynamic history of East Asian cattle

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Presenter: Donghee Kim

The evolutionary history of domesticated cattle in East Asia for the last 5,000 years remains largely obscure. Here, we investigate the origins and evolution of cattle genetic diversity in China by analyzing shotgun genome sequences of 166 ancient bovines spanning a 10,000-year period, encompassing now-extinct East Asian aurochs and domesticated cattle from key archaeological cultures. East and North Asian aurochs were distinct from western aurochs, although East Asian aurochs harbored ~15% western ancestry. The first domesticated cattle in the Yellow River region derived ~10% of their ancestry from local aurochs with an uneven genome-wide distribution. Early cattle from Xinjiang were genetically distinct and partially contributed to the later northern Chinese cattle. Indicine admixture became widespread only in the Medieval period in northern China.

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Re-evaluating Principal Component Analysis for visualizing spatial population structure: insights from simulated 2D stepping-stone landscapes

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Presenter: Jan Kocí

Some PCAs of genetic data reproduce geographic structure well, but this usually requires careful manual sample curation. Conversely, many published 2D PCA plots show distortions such as triangular or horseshoe patterns. An unsupervised protocol for reliably reconstructing spatial population structure is therefore needed.

We investigated how sparse sampling of simulated genetic landscapes and depletion of rare variants affect PCA and downstream manifold-learning visualizations. We show that common distortions are readily reproduced under patchy sampling and MAF-based variant filtering. Sparse but relatively uniform sampling combined with LD-based pruning usually recovers both local and global spatial structure. By contrast, increasingly stringent MAF-based filtering – subsample-dependent SNP ascertainment – tends to collapse the recovered landscape into triangular patterns and clines, and can also generate artificial distant outliers. Paradoxically, aggressive MAF filtering can sometimes partially restore global structure. Clustered sampling of landscapes almost always yields collapsed patterns and loss of global structure; only unfiltered data retain any realistic chance of recovering it. These visual patterns are difficult to distinguish from those produced by sampling simulated tree-like population histories, including admixture graphs.

We further show that manifold-learning methods such as UMAP can improve the resolution of spatial structure in data subjected to patchy sampling and/or subsample-dependent SNP ascertainment. However, optimal parameter values differ from those commonly reported in the literature and frequently require the full dimensionality of PC space rather than only the leading PCs. We also evaluate several genetic-distance metrics as second-level objective functions for optimizing PCA and manifold-learning parameters. Notably, the most widely used PCA implementation in population genetics – smartPCA and PLINK eigenvectors – appears suboptimal as input for manifold learning, whereas classic PC scores perform markedly better.

Because archaeogenetics relies heavily on sparse sampling and SNP panels enriched for common variants, PCA-based visualization of spatial structure – and hypotheses derived from it – should be treated with great caution.

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Epigenetics of ancient Pannonia populationsChen Henrik Kozulin¹¹ ELTE-RCH, Hungary

Presenter: Chen Henrik Kozulin

Ancient epigenetics is an emerging field of ancient DNA research enabled by advances in modeling post-mortem DNA damage. Reconstructed methylation profiles provide a potential link between genotype and phenotype, offering insights into environmentally mediated biological variation that are often inaccessible through genetics, anthropology, or archaeology alone. By characterizing methylation patterns in human remains from Roman-period Pannonia (1st–4th centuries CE), this study extends population genetic analyses beyond ancestry to explore aspects of lived conditions and local environmental influences. Several computational methods have been developed to reconstruct ancient methylation profiles. These approaches exploit differences in deamination rates between methylated and unmethylated cytosines to infer methylation states and identify differentially methylated regions (DMRs). Depending on their genomic context, DMRs may reflect regulatory variation and provide clues to environmentally associated biological responses. Preliminary analyses using the Reconstruction of Ancient Methylation (RoAM) pipeline suggest methylation differences among individuals from distinct Roman-period settlements in Pannonia. However, substantial genetic heterogeneity within these populations complicates interpretation, making it unclear whether the observed patterns primarily reflect environmental influences or population structure. Future work will incorporate populations of more clearly defined origins and further refine analytical approaches. While ancient epigenetic studies have traditionally focused on evolutionary questions and limited sample sets, this project applies methylation analysis within a historical framework to investigate biological variation among contemporaneous populations.

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The genetic history behind cultural diffusions and early-stage social organization of the Neolithic southeastern Mongolian PlateauTianxiang Liu¹, Qiaomei Fu¹¹ Chinese Academy of Sciences, China

Presenter: Tianxiang Liu

From the Late Paleolithic through the Early and Middle Neolithic periods, the southeastern Mongolian Plateau experienced a profound shift from highly mobile hunter-gatherer subsistence strategies to more sedentary farming-based lifeways. During this time, the region also functioned as an important corridor facilitating interactions between the East Asian Steppe and the plains and river valleys of northern East Asia. Nevertheless, the social structures of these populations and the degree to which cultural exchanges were accompanied by human migration have remained poorly understood. Here, we generated and analyzed genome-wide data from 35 ancient individuals from the southeastern Mongolian Plateau dating between 8,800 and 5,000 years ago. Our results show that Early Holocene populations across this region derived from a shared ancestral lineage, supporting a scenario in which millet agriculture spread from the North China Plain into the eastern steppe largely through cultural transmission rather than large-scale population movement. We further demonstrate that this ancestry, dominant among Eastern Steppe populations and potentially linked to the post-Last Glacial Maximum (LGM) dispersal of microblade technologies, persisted on the southeastern Mongolian Plateau until approximately 7,500-5,700 years ago. During this period, it also contributed genetically to populations inhabiting the West Liao River basin that were associated with the Hongshan culture. Subsequently, the continuity of this Early Holocene southeastern Mongolian Plateau ancestry was interrupted by populations from both the northeastern Mongolian Plateau and the Hongshan-associated communities of the West Liao River region. These genetic influxes coincided with episodes of cultural exchange occurring between 5,700 and 5,000 years ago. In addition, kinship analyses identified co-burials of nuclear family members dating to 8,800-7,500 years ago, indicating that early sedentary communities in northern East Asia were structured around family-based social units. Taken together, these findings uncover intricate dynamics of genetic interaction, cultural transmission, and social organization in northern East Asia from the LGM to the Middle Holocene.

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Optimized manifold learning and IBD analyses reveal remarkable genetic diversity in the Cherniakhiv-culture population

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Presenter: Bohdan Mamchur

The Goths were among the earliest major actors in Migration-period Europe. Their legacy is documented both archaeologically, in the Wielbark and later Cherniakhiv/Sântana de Mureș cultures, and in contemporary written sources. Yet their origins and admixture with surrounding populations remain genetically understudied. We apply a new framework for visualizing population structure – deep optimization of manifold-learning (ML) algorithms – to genome-wide ancient DNA data, increasing the resolution of the genetic landscape of Late Roman Europe.

The method explores a broad parameter space, including three PCA algorithms and data-normalization schemes, the PHATE ML algorithm, three distance metrics in PC space, and dense grids of input PC dimensionality and nearest-neighbor number. Candidate 3D low-dimensional embeddings are ranked in layered steps using fit to Hamming (1 – IBS) genetic distances and GRM-derived dissimilarities, together with separation of IBD-defined clusters inferred with the Leiden algorithm.

Our dataset includes 40 newly sequenced individuals from Viitenky, Kharkiv region, Ukraine, the largest excavated Cherniakhiv burial ground east of the Dnieper. These data substantially expand genetic coverage of the Cherniakhiv culture, which remains understudied despite thousands of recorded graves and a rich, remarkably uniform material culture.

Genome-wide pseudo-haploid SNP calls from 20,522 ancient Eurasians, dated to 1200 BC – 1800 AD, were projected onto a PC basis defined by 4,195 present-day Eurasian individuals, both in Human Origins SNP set (~578k SNPs). Applied to this dataset, PHATE allowed us to reach a new level of detail in resolution of ancestry clines and clusters for this region. Early Wielbark Gothic individuals appear genetically homogeneous and cluster with northern Germanic groups. By contrast, 71 Cherniakhiv individuals (13 published, 58 newly reported) are highly diverse: 13% are Germanic-like, 16% Balkan–East Mediterranean, ~15% Sarmatian-related, and 42% admixed, with rare Slavic and Caucasus-linked outliers. Published Thracian and Balkan Gepidic sample sets (both published and novel) further reveal continued admixture with Roman provincial populations during Gothic-associated movements into Roman territory.

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Ancient DNA reveals that natural selection has upregulated the immune system over the last 10,000 years

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Presenter: Javier Maravall López

The human immune system is widely hypothesized to reflect genetic adaptation to infectious diseases since the advent of agriculture, potentially affecting present-day risk of immune-mediated diseases, but little is known about the specific functional elements impacted by adaptation. Ancient DNA enables direct study of human adaptation, and our recent study has identified hundreds of statistically significant loci impacted by adaptation in West Eurasia. Here, we characterize the immune landscape of human adaptation by systematically integrating ancient DNA-derived selection statistics from our recent study with disease GWAS, molecular QTL, scRNA-seq, and gene pathway data to identify immune diseases/traits, variants, genes, pathways and tissues/cell types impacted by adaptation. Genome-wide, we find that positively-selected alleles are associated with reduced susceptibility to infection, including respiratory and gastrointestinal pathogens such as tuberculosis and influenza (average genetic correlation of -0.045 (SE 0.017) across 12 independent infection traits); this is consistent with enrichment of selection signals in immune cells within barrier tissues that interface with pathogens, such as the respiratory tract and gut mucosa ($FDR < 10^{-6}$). We find that positively-selected alleles tend to increase risk of inflammatory bowel disease, consistent with an evolutionary trade-off; and protect against asthma and dermatitis, challenging a common view that past adaptation to pathogens increased risk for allergic diseases.

We elucidate the biology of many adaptive loci by colocating convergent signals from selection, infectious disease GWAS and immune-gene QTLs to likely causal variants (e.g., a SNP upstream of *FUT6*, fine-mapped with 98% confidence for a GWAS of intestinal infections and with >99% confidence for expression of several key gut immunity proteins, including *MUC2*, *GP2* and *PIGR*). Genes with QTL colocalization to adaptive loci are strongly enriched for host-defense pathways ($P < 10^{-6}$), with QTL effect directions indicating that selection has increased the activity of these pathways (e.g., "inflammatory response", $FDR < 1.5 \times 10^{-10}$; $P < 7 \times 10^{-4}$ for directional effect).

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The ancient human interactome: how the protein-coding variants of ancient humans impact the protein-protein interaction network.

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Presenter: Christine Brun

Proteins seldom act alone to perform their functions. For this, they interact with other macromolecules in a complex molecular network. Most of the molecular interactions between proteins are mediated by specific amino acid sequences, forming interfaces. However, certain missense mutations can impair these regions, disrupting protein protein interactions (PPIs), and impacting cellular and physiological processes, and ultimately phenotypes.

To investigate the influence of ancient variants on the PPI network, we are computationally inferring the Ancient Human Interactome. To do so, we used our computational pipeline IRMA (Interactome Remodeling from Mutational Alterations) that predicts the effect of missense variants on PPIs. Briefly, starting from the available Variant Coding Files from Altai, Vindija, Chagyrskaya and Denisova (Laurits Skov, 10.5281/zenodo.10806725) mapped on the Modern human PPI network, IRMA predicts the interaction perturbations induced by the interface changes based on the integration of several datasets such as AlphaFold and mimicINT and uses machine learning to predict the effect of ancient variants on the PPIs.

Out of the 25 098 missense mutations reported in the ancient sequences, 7% are predicted to modify PPIs, among which 77% disrupt interactions. Overall, compared to the modern human interactome that contains ~109 000 interactions, these ancient variants change 0,5% of the interactions by increasing/decreasing their strength or disrupting them.

A detailed network analysis shows that the disrupted interactions may impact several cellular processes such as the regulation of neuronal apoptosis, regulation of metabolism, sarcomere organization, antimicrobial immune response, different regulation processes and different signaling mechanisms.

In the near future, we will complete the interactome inference with the effect of other types of variants (i.e. truncated proteins) not yet included in the current analysis, in order to produce the first Ancient Human Interactome. The detailed analysis of this network will allow us to better understand the functional consequences of the sequence and molecular changes that distinguish ancient and modern humans and to get an integrated vision of the cellular mechanisms that have been at work during evolution.

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pIAlgue: a deep learning framework for pathogen detection in ancient metagenomic datasets

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Presenter: Megan Michel

The exponential growth in the depth and breadth of published ancient DNA datasets has set the stage for large-scale exploration of the spatiotemporal dynamics of ancient bacterial, viral, and eukaryotic pathogens. However, current approaches for taxon identification in ancient metagenomic contexts are computationally expensive, sensitive to reference database design, and yield qualitative assessments and/or summary metrics that require substantial user interpretation and preclude cross-study comparisons. We develop a deep learning framework that reformulates ancient pathogen detection as a computer vision problem, focusing on detection of *Yersinia pestis*, the causative agent of plague, as a proof-of-concept application. The model builds on a competitive mapping approach, with raw reads mapped to a multi-species artificial chromosome including 450 pathogenic taxa of interest and closely related outgroups. We summarize competitively mapped alignment data as a heatmap in which pixels represent genomic regions and color channels encode edit distance, mapping quality score, and post-mortem damage score of aligned reads, metrics useful for identifying diverse ancient pathogenic species. Our method leverages a pre-trained image classifier to cope with small dataset size, fine-tuning on empirical alignment data from a curated case-control set including both published and unpublished data. On an independent validation set, the model achieved 93.9% sensitivity and 99.2% specificity, with a precision of 98.5% and an F1 score of 96.2%. We benchmark our method's performance against HOPS as a tool for capture candidate prioritization, analyzing a well-curated set of 529 individuals from 12 archaeological sites in the Netherlands spanning the Second Pandemic period. The trained model is highly portable and can be deployed nearly instantaneously from a desktop computer, while the model's probabilistic assessment of classification confidence facilitates direct cross-study comparison. Together with its strong performance, these features suggest that deep learning methods represent a promising avenue for future studies leveraging the growth in ancient DNA datasets for paleoepidemiological exploration.

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Comparative genomics at prostate cancer risk loci to identify prostate cancer predisposition genes in ancient and modern humans

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Presenter: Chidi Molokwu

Introduction

Prostate cancer (PCa) is the most common cancer in the human male but is not reported in other extant primates. Bone lesions suggestive of metastatic PCa have been reported in ancient Egyptian mummies, but not in Neanderthal bony remains. Genome wide association studies (GWAS) in modern humans have identified PCa associated single nucleotide polymorphisms (SNPs). It isn't known how the SNPs are distributed across human populations and whether they are shared with extant and extinct primates.

Methods

PCa SNPs were obtained from the GWAS Catalog (<https://www.ebi.ac.uk/gwas/home>). Risk Allele Frequencies (RAFs) were obtained from Ensembl (<https://www.ensembl.org/>). Human and chimpanzee genomes were aligned using the Cactus genome alignment tool (<https://cglgenomics.ucsc.edu/data/cactus/>). PhyloP scores were determined for each SNP position. The allele at the SNP positions were compared to determine positions where the PCa risk allele was divergent from the ancestral primate allele. Ancient and modern human genome alignments were accessed using the Max Plank Institute Ancient Genome Browser (<https://www.eva.mpg.de/genetics/genomeprojects/neandertal/indexhtml>).

Results

520 PCa SNPs were identified. 11 SNPs occurred at positions with PhyloP score >1.0, and where the PCa risk allele was different from the chimpanzee allele. ZGPAT, HNF1B, ANO7, INCENP, GATA2-AS1 and CLND23 were identified as candidate genes for PCa predisposition. Ancient genome coverage was sparse at the candidate SNP loci. 10 SNPs were unique to European (EUR), 19 to African (AFR) and 7 to East Asian (EAS) populations while 484 were shared.

Conclusion

Five candidate genes and 1 non-coding region harboured PCa risk SNPs that are in loci with high conservation between mammalian species, but a divergent PCa risk allele in human males. Further studies are needed to determine their role in PCa predisposition in humans. PCa SNPs unique to EUR, AFR and EAS populations are likely to be driving the genetic component of prostate cancer racial disparities.

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The Neanderthal in the UK Biobank

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Presenter: Adeline Morez

Neanderthal haplotypes in present-day Eurasians are unevenly distributed across the genome, forming introgression deserts and high-frequency segments consistent with adaptive introgression, with additional random variation affected by genetic drift. However, current estimates are limited by modest sample sizes and analyses restricted to subsets of the genome, given that any individual carries only 1-2% Neanderthal ancestry. Here we extract and analyse Neanderthal haplotypes from 45,000 imputed and phased genomes in the UK Biobank. Even at this scale, the number of sites overlapping Neanderthal haplotypes approaches—but does not reach—saturation, with rare haplotypes still being discovered. Using the derived allele frequency spectrum within the surviving Neanderthal segments, we infer a divergence time of 2,061 generations between the introgressed lineage and the Vindija Neanderthal, and estimate the effective population size of the introgressed lineage to $N_e = 6,564$. Individual-level resolution allows identification of 545 independent loci with excess Neanderthal homozygosity, consistent with ongoing selection. Despite the extensive dataset, a substantial portion of the genome remains a Neanderthal desert. Within these regions, we detect seven Human Accelerated Regions affected by recent human selective sweeps (TMRCA <650 kya), four located within introns of cerebellum-expressed genes, providing further support for their potential as modern human-specific adaptation.

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Ancient genetic diversity and population structure in the early settlers of the Insular Caribbean

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Presenter: Kathrin Nägele

After decades of deconstructing and reconstructing the early period of human occupation in the Caribbean, the landscape of early settlers is now considered a diverse one, with a multitude of different burial practices, consumption patterns and material cultures, however with unknown continental origins, and little understanding of genetic patterns underlying the cultural diversity observed. In this study, we produced genomic data for 63 individuals associated with early-settler contexts from Cuba and Aruba, and two individuals from Mexico and Nicaragua, adding to the Circum-Caribbean references. The ancient genomes are contextualized by radiocarbon dates dating between ca. 6000 and 600 years ago, including 26 newly measured AMS dates. After quality control filtering, whole genome SNP data of 41 individuals were combined with published data. Although we are still unable to pinpoint continental origins for the early settlers, we find considerable genetic variation in the Circum-Caribbean region, including a previously unobserved lineage in Nicaragua. We confirm genetic structure within Cuba, potentially the result of an already structured founding population or multiple migration to the island.

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Rediscovering the genomic identity of unknown museal specimens

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Presenter: Valeria Nicolini

Despite conservation challenges, museums offer a valuable opportunity to showcase new discoveries about the human past. Recently, museomics has emerged as a powerful resource for extracting genetic information from human remains preserved in museum collections, particularly for those with ambiguous provenance. In this study, we bridged museum specimens and archaeogenomics by analyzing human skulls from a forgotten collection at the "Museo della Natura e dell'Uomo" in Padua, Italy. The individuals were associated with museum labels indicating their possible cultural affiliation or geographical origin. Our goal was to evaluate the accuracy of the labels and determine the genetic identity of individuals described as "Botocudo", "Saracens", "Pompeii", and "Etruscan". We successfully extracted ancient DNA from teeth and generated genomic data for ten individuals. The comparison between label attributions and genomic inferences revealed mixed outcomes. First, two individuals labeled as Botocudo, from the Minas Gerais state in Brazil, cluster with other Indigenous Southern American populations and reveal a first-degree familial relationship. A specimen labeled as Etruscan shows genetic affinities with individuals associated with the Etruscan culture, a finding supported by radiocarbon dating. An individual labeled as Pompeian, radiocarbon dated to the Imperial Age, shows genetic similarity with Balkan populations, suggesting high mobility during the Roman Imperial Age. Six individuals labeled as Saracens display high mitochondrial heterogeneity, no kinship, no genetic affinity to published Saracen individuals, inconsistent with the Middle Ages Saracen invasion. Our findings underscore the value of museomics in recovering lost biological identities, acknowledging the dignity of these individuals, and providing new genetic insights into the human past.

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Cancelled

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Genetic differentiation amid heightened mobility in Iron Age Southeastern EuropeStephan Schiffels¹, Luka Papac¹¹ Max Planck Institute for Evolutionary Anthropology, Germany

Presenter: Stephan Schiffels

Southeastern Europe in the first millennium BCE features prominently in the earliest surviving historical accounts, which describe a diversity of peoples, including “Scythians”, “Celts”, and “Thracians”. It remains unclear whether this described diversity corresponds to distinct demographic histories, mobility patterns, or interaction networks reflected in the genetic record. To address this, we report new genome-wide data for 308 individuals from Southeastern Europe, predominantly dating to the first millennium BCE, and combine these with available Bronze and Iron Age data to analyse 860 ancient genomes from the region. We find large-scale Aegean-related gene flow into eastern Southeastern Europe by the onset of the Iron Age, leading to genetic homogenisation between mainland Greece and Thrace. Scythian-associated individuals from Transylvania carried up to 35% Central Asian ancestry and were buried alongside others with northern European or southern Balkan origins, revealing striking diversity even within families, where close relatives differed up to fivefold in Central Asian ancestry. In contrast, western Southeastern Europe showed broad genetic continuity with the Bronze Age, consistent with predominantly local origins for groups later putatively associated with “Illyrians”. Nevertheless, individual mobility increased, exemplified by strong trans-Adriatic connections in Croatia. Archaeologically attested “Celtic” migrations left little genetic trace of Western or Central European ancestry. Finally, haplotype-sharing patterns reveal increasing population size and heightened mobility—particularly among females—even as genetic differentiation increased.

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Population-specific haplotype fingerprints from identity-by-descent sharing landscapesAjai K Pathak², Toomas Kivisild¹¹ KU Leuven, Belgium² University of Tartu, Estonia

Presenter: Ajai K Pathak

Population-specific genomic signatures are often difficult to resolve in underrepresented, admixed, and endogamous populations using conventional allele-frequency-based approaches. We present a novel framework that leverages genome-wide patterns of identity-by-descent (IBD) sharing to identify population-specific haplotype fingerprints and candidate regions shaped by recent demographic and evolutionary processes.

Using genome-wide data from diverse human populations, we quantified the distribution of recurrent IBD sharing across the genome and developed a filtering strategy to identify robust population-enriched haplotype signals. By integrating information across multiple IBD-sharing scales and comparative population analyses, the approach prioritizes genomic regions exhibiting consistent and population-specific sharing patterns.

As a proof of concept, we applied the framework to a South Asian population and identified multiple reproducible haplotype blocks showing elevated sharing relative to reference populations from Europe and East Asia. These signals form extended genomic clusters, suggesting the presence of stable population-specific haplotypes rather than isolated stochastic sharing events.

Our results demonstrate that IBD-sharing landscapes provide a powerful complementary perspective for studying population structure, demographic history, and candidate targets of selection. The framework also provides a foundation for ancestry fingerprinting and privacy-preserving population genomic summaries that facilitate comparisons across cohorts without requiring the exchange of individual-level genomic data.

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Assessing recent human demography through simulations and runs-of-homozygositySantiago Pérez-Marco¹, David Comas², Francisco C. Ceballos¹¹ Institut de Biologia Evolutiva (UPF-CSIC), Spain² Universitat Pompeu Fabra, Spain

Presenter: Santiago Pérez-Marco

Runs of homozygosity (ROH) are autozygous DNA fragments whose pattern and distribution can explain the most important recent demographic events from a population. Usually they are used to test the influence of bottlenecks and endogamy in a population. To extract conclusions, results from the query population are compared with other reference populations. This approach assumes that real reference populations have simple histories and have experienced just one demographic event. We propose a proof of concept where we compare results from real populations with SLiM simulated populations following simple demographic models. Admixture, bottleneck, consanguineous mating under neutrality were simulated in simple models. Samples from the 1000 Genome Project (1kGP) with presumably known demographies were compared to the obtained simulated models. European Roma, a demographically complex human population, was tested under such simulated scenarios as a case of a study. Despite our simulations aligning with the theoretical expectations, our approach presents some limitations implementing consanguineous mating models as well as the presence of individuals with outlier values in all simulated demographic scenarios. According to our simple model simulations, reference real populations present more complex histories than what is usually assumed. The Roma population fits in-between slightly consanguineous mating and distant bottleneck models, although they present heterogenous results depending on their country of origin. Our goal is to present a new perspective to analyze human populations' ROHs in a more robust and less dependent on assumptions way; as well as to open a discussion of how to standardize human ROH analysis for demographic inference.

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EquiY: a tree-aware framework for horse Y-chromosome classification and refinementAnna Szécsényi-Nagy¹, Catherine Puello Mora², Dániel Gerber¹¹ ELTE RCH, Hungary² Universitat Oberta de Catalunya (UOC) / ELTE RCH, Hungary

Presenter: Catherine Puello Mora

The horse Y-chromosome is valuable for reconstructing paternal lineages and domestication histories. Recent horse Y-chromosome phylogenies provide a detailed framework for these lineages, but applying this resolution to ancient genomes remains challenging because

Y-chromosome data are often sparse, unevenly covered and affected by molecular damage. Fine-scale assignment requires methods that use informative markers efficiently without overinterpreting weak or conflicting evidence.

EquiY combines a curated horse Y-chromosome phylogeny with SNP and INDEL markers mapped to the LipY764 reference in a compatible tree-panel structure. Its active core includes 237 tree nodes and 2,788 markers. Within a reproducible so-called Snakemake workflow, derived and ancestral observations are evaluated across the phylogeny to identify the deepest supported placement. With sufficient evidence, EquiY can support fine-scale downstream placements; with sparse or conflicting support, it retains an internal placement or reports the case as unresolved.

EquiY is being developed as a horse-specific automated workflow for Y-chromosome haplogroup classification in ancient DNA data. Its novelty lies in adapting phylogeny-based haplogroup calling to the equine Y-chromosome tree, linking marker observations directly to curated branches and making so-called downstream haplogroup assignments reproducible and updateable as marker panels and phylogenies continue to improve.

In an initial benchmark of 94 modern male horses, EquiY produced placements broadly consistent with published Y-chromosome classifications and showed its potential to refine selected paternal lineages beyond broad labels. Current development focuses on marker curation, tree refinement and validation on ancient horse genomes.

By providing a standardized workflow for horse paternal lineage classification, EquiY will help the ancient DNA community compare Y-chromosome results across studies, refine domestication models and integrate fragmented male-lineage evidence into broader archaeogenomic interpretations.

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Fine-scale reconstruction of regional admixture dynamics and sex-biased contributions across Mexico

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Presenter: Ricardo Rodriguez

Background: Mexico represents one of the most complex post-colonial admixture landscapes, shaped by interactions among Indigenous American, European, African, and Asian-related ancestries. These processes occurred heterogeneously across the country, differing in timing, intensity, and ancestral composition according to regional demographic and historical contexts. Despite previous genome-wide studies, a regionally resolved reconstruction of admixture dynamics and sex-biased contributions across Mexico remains incomplete.

Materials and Methods: We analyzed 1.8 million SNPs distributed genome-wide in six thousand admixed individuals from the nationwide cohort of the Mexican Biobank using a multi-continental reference panel. Population structure was characterized using PCA and ADMIXTURE. Admixture timing and multi-pulse histories were inferred using linkage disequilibrium- and haplotype-based approaches. To investigate sex-specific demographic dynamics, we contrasted ancestry proportions and admixture pulse models between autosomes and the X-chromosome across regions, allowing identification of differential ancestry contributions and timing.

Results: We identified pronounced regional heterogeneity in ancestry composition and admixture histories across Mexico. Distinct admixture profiles and temporal patterns across regions reflect differential colonial and post-colonial demographic processes. X chromosome-based analyses revealed deviations in ancestry proportions and admixture timing relative to autosomal estimates, consistent with sex-biased admixture processes that vary geographically. Additionally, localized African- and Asian-related ancestry components are detected, consistent with forced migrations and Atlantic-Pacific gene flow.

Conclusion: This study refines current models of mestizaje by providing a regionally resolved reconstruction of admixture dynamics across Mexico. Our findings highlight the importance of integrating regional and sex-informed analyses to better understand the demographic and evolutionary processes shaping genetic diversity in Latin America.

Ricardo Rodriguez is the recipient of an EMBO|EMBL Symposia fellowship, jointly funded by EMBO and the EMBL Advanced Training Centre Corporate Partnership Programme.

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Archaeogenetic investigation of Gepid Period cemeteries from the eastern parts of the Carpathian Basin

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Presenter: Oszkar Schutz

The Gepids rose to prominence under Hunnic rule in the Carpathian Basin during the 5th century CE. After the death of Attila (453 CE), they broke away from the loose confederation of Germanic and nomadic tribes that emerged from the ruins of the Hunnic Empire. Seizing the opportunity created by the resulting power vacuum, they established an independent sovereignty in the eastern Carpathian Basin (former province of Dacia). During their century-long rule (~ 454-567 CE) the Gepidic leaders accumulated considerable wealth and maintained relative political stability within their realm. They were ultimately defeated in 567 CE by a coalition of Avars and Langobards, leading to the loss of their political independence. Historical sources suggest that a significant Gepidic population remained in the Carpathian Basin well into the Avar period, a century later. Additionally, it remains unclear how the large Sarmatian population, that had inhabited the Great Hungarian Plain before the Gepid Period (in the 4–5th centuries), adapted to the new political landscape as their archaeological material seemingly vanishes.

To address these questions, we have generated whole genome sequences from 87 individuals from the Gepid Period of the Carpathian Basin. We have also assembled a reference database of more than 5000 genomes from the public literature, and harmonized different data types by realigning public sequences through a rigorous pipeline as well as imputing data with appropriate quality metrics. This allowed us to explore multiple lines of hypothesis-independent approaches. This extensive database also provides a good bases to construct an ancient reference panel for PCA projection and high-resolution visualization of Eurasian population structure using optimized manifold learning.

The Gepid-period individuals from the Carpathian Basin displayed strong genetic similarity to the population of the region from the preceding and following historical periods with negligible outliers displaying East Asian admixture. However, their IBD (identity-by-descent) sharing network showed strong indicators of reproductive isolation, as most cemeteries contained only small kin-groups with negligible connections neither among the cemeteries nor with other groups from the Carpathian Basin.

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Trinidad at the crossroads of ancient American populations

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Presenter: Eleni Seferidou

Ancient genomic research in the Caribbean region has shed light on the complexity of past Caribbean human populations, possible settlement routes and continuous movement into and among the islands. Trinidad, geographically, serves as a bridge between the islands and the American continent. This unique position is also reflected in its archaeological record, indicated by shifts in ceramic traditions, from Saladoid to Arauquinoid, associated with the expansion of Arawakan and Cariban speaking societies respectively, both starting in the mainland after 3000 BP. We generated aDNA data from 35 individuals from Trinidad covering a time transect of 1200 years, to investigate if the discontinuities in the material culture mirrored demographic and genetic changes, as well as the genetic relationships to the mainland and other islands. We identify an earlier (~1700-1000 BP) habitation period in Trinidad with a genetic profile resembling other pottery-producing groups from the Caribbean. In a later period (~800-500 BP), the inhabitants trace part of their ancestry back to the earlier occupation, but show an additional mainland contribution, corroborating evidence for continuous contact - and possibly multiple movements - with the mainland. Contrary to expectations, and despite vast geographical distances, the ancestry profile of these contributions can be identified in the Amazon region. Emphasizing the underestimated complexity of Indigenous interactions between the islands and the heart of the South American continent, the results have tremendous implications for models of expansions within Amazonia.

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On the limits of introgression: the early out of Africa conundrum

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Presenter: Jose Miguel Serradell Noguera

The global expansion of *Homo sapiens* out of Africa approximately 50–60 thousand years ago (kya) represents a defining event in human evolutionary history. However, fossil evidence from the Levant, East Asia, and Southeast Asia dated between ~70 and 210 kya raise questions about the potential genetic contribution of an early Out-of-Africa (eOOA) expansion to present-day populations. Current genomic models largely fail to detect the fingerprint of such eOOA, which has been traditionally interpreted as evidence of a near-complete replacement of these early groups by the later expansion. However, an alternative hypothesis explaining such results is that current methods are underpowered to detect such introgressed ancestry due to the particular nature of the genetic ancestry of the eOOA populations, which could be indistinguishable from the ones in a later diaspora. To explore the limits of detectability, we simulate simple demographic scenarios to explore the distribution of fragments in the genome under coalescent events. While long fragments associated with deep Time of the Most Recent Common Ancestors are theoretically expected under introgression, we find that as population divergence times approach ~100 kya, introgression signals rapidly decay and become indistinguishable from background coalescent variation. We then, evaluate whether genetic traces of an eOOA could be detectable using an Approximate Bayesian Computation framework coupled with Deep Learning approach considering the Site Frequency Spectrum in real data. Our results indicate that all considered demographic models tend to be confused, with the model considering a single OOA been selected as the most plausible model in over 50% of the time. When considering real data, models without eOOA introgression provide the best fit. Both results go against the fossil record suggesting that the absence of detectable eOOA introgression may reflect methodological limitations rather than true absence of ancestry, potentially explaining why Neanderthal and Denisovan introgression is detectable while eOOA signals are not, highlighting the need for novel summary statistics specifically tailored to detect introgression in this temporal regime.

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Ancient cattle DNA from Pastoral Neolithic of Kenya and Tanzania reveals the origins of African *Bos taurus*

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Presenter: Bárbara Sousa da Mota

Cattle (*Bos taurus*) were first domesticated around 10,000 years ago (10 ka) in Southwest Asia from wild aurochs (*Bos primigenius*). Cattle are ubiquitous in Africa today, and were first associated with African Humid Period “cattle cultures” of the Sahara and Sahel (~8-5.5 ka) before spreading southward as the climate became more arid. In eastern Africa, the Pastoral Neolithic era (PN; ~5-1.2 ka) witnessed the development of specialized pastoralist cultures centered on *Bos taurus*. Today, however, East African cattle breeds have moderate to high amounts of indicine or zebu (*Bos indicus*) ancestry, reflecting a later phase of interbreeding with this arid-adapted South Asian domesticate, such that we only find African *Bos taurus* with little or no indicine gene flow in West Africa. The population history of the African *Bos taurus* is yet to be fully understood, including its spatiotemporal spread across Africa. It is also unknown whether African taurine cattle descend directly from domesticates from Southwest Asia or whether they derive from further introgression from wild aurochs and/or independent domestication events. We report five ancient cattle genomes from PN sites in Kenya and Tanzania with average depth between 0.03x and 0.23x, and direct radiocarbon dates ranging ~3-2.4 ka. Using PCA (principal component analysis) and f3-outgroup statistics, we found that the ancient cattle genomes are more closely related to present-day African *Bos taurus* breeds in West Africa, such as N'Dama. Unlike present-day East African cattle breeds, the studied ancient cattle did not receive gene flow from *Bos indicus* groups. Further f-statistics-based analyses revealed that these genomes have more genetic affinity with modern and ancient Eurasian *Bos taurus* compared to West African *Bos taurus* breeds. Previous studies modelled human PN groups in Kenya and Tanzania as the result of multiple ancestries, including a Levantine/North African component. As such, our findings support a model of cattle movement alongside human migration from Southwest Asia, through northeast Africa to East Africa.

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Population structure and mobility in Roman frontier communities of the Carpathian Basin

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Presenter: Anna Szécsényi-Nagy

The Carpathian Basin—home to Romans and Romanized groups as well as various barbarian communities beyond the imperial frontier—witnessed dynamic interactions between populations of different origins during the 1st–5th centuries CE.

West and south of the Danube lay the Roman province of Pannonia, inhabited by a diverse Romanized population. East of the Danube, the Great Hungarian Plain was occupied mainly by Sarmatian communities. The region underwent major political and demographic transformations during Late Antiquity, particularly with the arrival of groups associated with the Hunnic-period migrations.

Our MTA–ELTE HTK Momentum Bioarchaeology Programme analyses various cemetery types by integrating burial archaeology, anthropology, isotopic and genetic data to reconstruct health conditions, biological backgrounds, and connections among cemeteries associated with different communities and social strata in northeastern Pannonia. The present archaeogenetic study provides new insights into these populations by analysing approximately 250 newly generated ancient genomes from both sides of the Danube in the region of present-day Budapest. Whole-genome data reveal densely connected communities and a marked temporal shift east of the Danube: earlier individuals genetically align with Central European groups, whereas 5th-century individuals show clear genetic input from East Eurasia and the Near East/Caucasus. In contrast, genome-wide data from Late Roman individuals excavated west of the Danube indicate limited genetic relatedness within and between sites, high genetic diversity, and signals of admixture from non-Roman groups beyond the frontier.

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The pan-European impact of the Balkan hunter-gatherers

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Presenter: Daniel Tabin

During the Last Glacial Maximum (LGM) 26–19 thousand years ago (kya), Europe had three principal refugia not covered by ice: the Iberian, Italian, and the Balkan peninsulas. While the Iberian and Italian refugia have been studied with ancient DNA, the legacy of the Balkan refugium has remained a mystery due to a lack of genetic data 30,000–12,000 BP. We present genome-wide data from nine newly reported individuals: three Epipaleolithic individuals from Romania, one Epipaleolithic individual from Bosnia and Herzegovina, three Mesolithic individuals from Greece, one from Epipaleolithic Southern Italy and one from Mesolithic Sardinia. The Climente II and Franchthi individuals were from a previously unsampled population that was the primary source for most later European hunter-gatherers. Westward expansion into the Italian Peninsula and mixture with minority ancestry from pre-LGM Italians produced the Italian Epipaleolithic; further mixture with ancestry from Iberian refugial people formed Post-LGM northwest Europeans. Northward expansion bypassing the Italian Peninsula produced one of the primary ancestries found in East European and Scandinavian hunter-gatherers. Eastward expansion to the Aegean led to Anatolian-European mixtures before the spread of farming.

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The prehistory of Sicily: PCA of ancient genomes across the Pleistocene–Holocene transition and agent-based modelling of Epigravettian–Mesolithic demography

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Presenter: Valentino Romano

Sicily across the Late Pleistocene–Holocene transition hosts some of the most informative populations for reconstructing the genomic history of the central Mediterranean. Principal Component Analysis (PCA) was performed on 27 ancient Sicilian individuals, drawn from the Allen Ancient DNA Resource (AADR v66.0, April 2026), spanning from the Epigravettian (c. 13,450 BP) to the Middle Neolithic (c. 5,951 BP). The Human Origins panel consisting of 584,131 SNPs, was employed, with principal components computed on 356 present-day Eurasian populations and ancient samples projected via least-squares projection. To enable a direct interpretation of genetic affinities, 12 Western Hunter-Gatherer (WHG) and 15 Early European Farmer (EEF) individuals — including Anatolian Neolithic and central-European Early Neolithic representatives — were included as ancestral anchors, and their centroids computed on the first two components. Pre-Neolithic individuals (Epigravettian, Mesolithic and Late Mesolithic) coincide with the WHG centroid, whereas Middle Neolithic individuals coincide with the EEF centroid. Heterogeneity within the Early Neolithic horizon at Grotta dell'Uzzo, where contemporaneous individuals project onto opposite ancestral references, indicates that the demographic transition proceeded through the coexistence of genetically distinct populations rather than gradual admixture. Simultaneously, we developed PaleoSicily, to our knowledge the first NetLogo/GIS Agent-based model of pre-Neolithic Sicily, in which Epigravettian and Mesolithic HG — assigned a shared behavioural ruleset governing band cohesion, social boundaries, base camps and life history — move over a QGIS-derived Digital Elevation Model in which altitude and slope constrain and channel mobility routes. A preliminary parameter-space exploration identified settings under which humans persisted throughout the ~5,000-year Palaeolithic window, with mean population sizes consistent with conservative literature estimates and low terminal counts. The two approaches address complementary dimensions of the same record — ancestral composition and demographic structure — and converge on a numerically thin, genetically WHG-like pre-Neolithic substrate replaced rather than assimilated at the Neolithic threshold.

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Demography and life histories across the Roman frontier in Germany 400–700 CE

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Presenter: Leonardo Vallini

The transition between Late Antiquity and the Early Middle Ages in Western and Central Europe is marked by profound transformations and by the emergence of new political and social structures. These changes have traditionally been attributed to large-scale migrations, however new studies increasingly emphasise the role of small group mobility and the gradual reshaping of the Roman world. To investigate this crucial period, we sequenced and analysed 258 ancient genomes from sites along the former Roman frontier zone in southern Germany. In the late fifth century, coinciding with the dissolution of Roman state structures, we detect a major demographic shift when a founding population of northern European ancestry mixed with genetically diverse Roman provincial groups. Reconstructed pedigrees and filia, a novel method to infer the ancestry of unsampled relatives, reveal widespread and immediate intermarriage of the two groups, characterised by a lack of significant cultural or social differentiation. During the sixth century, continuous admixture gradually reduced the genetic structure eventually forming, by the seventh century, a population resembling modern Central Europeans. To investigate the life of Early Medieval communities beyond broad demographic shifts, we developed chronograph, a novel method that refines the chronology of genealogically linked individuals, to reconstruct high-resolution life histories. We estimate a generation time of ~28 years, life expectancies of ~40 (women) and ~43 (men), high infant mortality, early parenthood and a society in which most children lost at least one parent by age 10, yet most still grew up with grandparents. Finally, our analyses reveal a society centred on nuclear families practicing lifelong monogamy, strict incest avoidance, flexible lineage continuation and no levirate unions, indicating continuity with Late Roman social practices that later shaped the European family model.

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Ancient genomic insights into the high-status Hunnic tomb from Conduratu

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Presenter: Britta Van Tiel

The Huns were nomadic pastoralists originating from Central Asia. Their dispersal across the Eastern European plains during the 4th-5th centuries CE substantially changed the social, cultural, and political landscape of the region. Previous archaeogenetic research in this area has indicated that high-status Hunnic individuals have varied ancestral backgrounds, often indicating recent East Asian ancestry.

Rescue excavations undertaken in the region of Muntenia (Southeastern Romania) over the course of 2022-2023 resulted in the discovery and recovery of a high-status tomb dated to the 5th century CE. The individual in this tomb was buried with an assortment of weaponry, horse remains, and equestrian tack, consistent with contemporaneous graves of high-status warriors from across the Hunnic empire.

Here we present the preliminary results of the archaeogenetic research investigating the genetic ancestry of this newly sequenced individual, and comparing this genetic profile with other published contemporaneous individuals. Admixture analyses indicate an elevated proportion of East Asian ancestry compared to modern West Eurasian populations, consistent with other contemporaneous individuals from Hunnic-associated contexts.

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Reconstructing the genomic history of the Balearic archipelago in the Western Mediterranean

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Presenter: Vanessa Villalba-Mouco

The Balearic Islands, located in the Western Mediterranean, display a rich and distinctive prehistoric and historical trajectory that sets them apart from neighbouring regions. Their relative isolation makes them an ideal setting for tracing large-scale maritime migrations. First settled around 2350 BCE, the islands followed a unique demographic and economic path conditioned by their place in ancient sea lanes: diverging from Iberia during the Bronze and Iron Ages, coming under Roman and Vandal rule, flourishing under Byzantine occupation, becoming part of Al-Andalus, and later being conquered by the Crown of Aragón. To investigate population dynamics across these major political transitions, we generated genome-wide data from 191 individuals spanning ~3500 years, from the initial peopling of the islands in the Bronze Age to the early modern period. The first settlers were genetically similar to Bell Beaker associated groups in Western Europe and Iberia, a genetic profile that persisted until the Iron Age. Central Mediterranean and North African ancestries appeared during Carthaginian/Punic influence, while the most pronounced genetic transformation occurred in the Roman era, marked by substantial contributions from the Near East and North Africa. Late Antiquity saw continued mobility, with individuals exhibiting diverse mainland European and North African ancestries. Islamic rule coincided with the introduction of West African ancestry in Menorca, and, in Islamic burials only, additional North African ancestry. The later conquest by the Crown of Aragón reduced the amount of African ancestry through gene flow from the Iberian mainland and the displacement of Islamic groups. Our study combines archaeology, history, and genetics, providing a comprehensive view of population change across the Western Mediterranean over the past 3500 years, with the Balearic Archipelago as a central node.

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Ancient genomes redates the early Slavic dispersal to ~150 CE and revise Migration-period population history from the Roman limes to the VolgaLeonid Vyazov¹¹ University of Ostrava, Czech Republic

Presenter: Leonid Vyazov

Recent aDNA studies have shown that the 6th-century CE Slavic expansion involved large-scale movement from Eastern Europe, with a tentative source in the Dniester–Don forest zone; however, the origin and early footprint of this ancestry remained unresolved. We present genome-wide data from 560 ancient individuals (~150 BCE–700 CE) across Eastern and Central Europe, integrated with ~5,100 published genomes and analyzed using IBD networks, optimized manifold learning, qpAdm, Y-chromosomal lineage-through-time trajectories, HapNe inference, and stable isotopes.

We identify an IBD-sharing community we term Central-East Europe (CEE) that includes most Early Medieval genomes from archaeologically Slavic contexts. Its earliest members, dated to ~150–200 CE and termed Core eCEE, occur as outliers in Roman-period assemblages at the Middle–Lower Danube (Viminacium), the Lower Don (Tanais, Temernik), and the Middle Volga (Imen'kovo horizon), pushing the onset of Slavic-associated dispersal four centuries earlier than the canonical 6th-century horizon. Core eCEE is modeled as a four-way mixture of East Baltic LBA, Middle Dnieper EIA, Eurasian forest-cline, and West/Central European Germanic-like ancestry. Its members are also isotopically distinct from co-burial populations, with elevated $\delta^{13}\text{C}$ values indicating a millet-rich diet maintained independently of local food regimes.

The former two ancestry components share a Trzciniec LBA root, also supported by 7 CEE-enriched Y-chromosomal lineages, mainly within R1a-Z282. HapNe trajectories and declining IBD connectivity date Baltic–Slavic demographic divergence to ~700 BCE, grounding phylolinguistic estimates of the split. The West/Central European component provides a genomic correlate of likely Germanic-associated Central European influence in the Dnieper–Dniester basin around 250 BCE (Zarubintsy horizon). We link the initial CEE dispersal to the post-Zarubintsy horizon and its “barbarian enamel” communication network; its eastward extension to the Middle Volga is independently supported by Early Slavic loanwords in Proto-Permic.

Together, these findings recast the Slavic expansion as a centuries-long, multidirectional demographic process that reshaped the East European population mosaic well before it entered the written record.

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Medieval genomes reveal a dual origin for Ashkenazi Jews

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Presenter: Shamam Waldman

Our previous work (Waldman et al. 2022) on individuals buried in a fourteenth-century Jewish cemetery in Erfurt, Germany, identified two ancestrally distinct groups: one with higher European-related ancestry ("Erfurt-EU"), and another with higher Mediterranean ancestry ("Erfurt-ME"). A parsimonious hypothesis at the time was that these two groups were part of an archipelago of sub-structured Jewish groups. Here, using newly generated data, we show that this interpretation was incorrect. Instead, the two Erfurt groups appear to represent two geographically widespread medieval Jewish populations: one associated with the Rhineland and western Europe, and the other with Central Europe.

We present new genome-wide data from 57 individuals from the medieval Jewish cemetery of Prague, Czechia, which was active between the thirteenth and fifteenth centuries. The majority of these individuals, together with three individuals from Brześć Kujawski, Poland, cluster with Erfurt-EU and represent a Central European Jewish (CAJ) population. The formation of this group includes a substantial mixture with Slavic groups and involved sex-biased admixture, as indicated by mtDNA lineages of European origin alongside Y-chromosome lineages deriving from Mediterranean and Levantine sources.

Our sample also includes at least two individuals with a recent mixture of CAJ and Slavic ancestry, each deriving from a different Slavic source population, as well as a third individual of entirely Slavic ancestry that was buried in the Jewish cemetery. These findings indicate that admixture with Slavic populations was a continuous process involving multiple sources and continued throughout the Middle Ages. This Slavic-related ancestry was not limited to a single local group, but contributed to the gene pool of modern Ashkenazi Jews. We estimate that the CAJ population contributed approximately 40% of the ancestry of modern Ashkenazi Jews through admixture with Western European Jews. Although both Western and Central European Jewish populations experienced strong bottlenecks, they followed distinct recent demographic trajectories, and whether any component of these bottlenecks was shared remains unresolved.

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Detecting deep-time selective sweeps from extended regions of capped genealogiesNick Patterson¹, Tianyi Wang³, Heng Li², David Reich³¹ Broad Institute of MIT and Harvard, United States of America² Harvard Medical School, United States of America³ Harvard University, United States of America

Presenter: Tianyi Wang

Multiple powerful methods exist for screening for natural selection based on patterns of genetic variation in living people. However, detecting the genetic signatures of selection events that occurred hundreds of thousands of years ago has been extraordinarily challenging due to fragmentation of haplotypes that obscures classical sweep signatures for very old events, and due to the paucity of unique ancestral lineages at such time depths. To screen for extremely ancient selective sweeps, we develop a new idea: screening for extended regions of the genome where there are many unlinked gene trees with a capped time since the most recent common ancestor. This is the pattern expected for an ancient selective sweep that eliminated variation over a megabase scale, so that hundreds of thousands of years later at this locus, there are no gene genealogies with a time since the most recent common ancestor older than the sweep date. A gene genealogy with a time since the most recent common ancestor that is hundreds of thousands of years old between two sequences is not unusual at any one locus. But an extended region where many independent gene genealogies do not exceed a date more than hundreds of thousands of years old is surprising. This signal of selection get stronger rather than weaker for older selective sweeps, because the unlinking of loci through recombination provides more independent observations.

We implement this idea by modifying the Pairwise Sequential Markovian Coalescent (PSMC), developing a score to search for genomic regions that coalesce significantly faster than the genome-wide backgrounds. Applying this framework to individuals from diverse populations, we identify genomic regions exhibiting accelerated coalescence consistent with selective events occurring deep in the Pleistocene. Candidate regions highlight biological pathways that may have experienced recurrent evolutionary pressures during early human evolution. These candidate regions provide insight into selection that may operated during key periods in *Homo erectus* and *Homo heidelbergensis* evolution.

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Improving short IBD detection in ancient and low-coverage genomes using a benchmark-based evaluation framework

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Presenter: Jiaming Xue

Identity-by-descent (IBD) segments provide rich information for studying recent shared ancestry, demographic history, and kinship relationships. However, detecting short IBD segments (2 to 8 cM long) remains challenging for low coverage and degraded DNA data such as typical for ancient genomes. False positive rates soar in this regime, but systematic benchmarking of IBD detection methods under these conditions remains limited.

In this study, we sequenced a parent-offspring trio from Medieval Europe to exceptionally high coverage (~18x depth), enabling us to construct high quality phased genotypes for the child. In tandem with down-sampled data across sequencing depths from 0.1x to 2x, this empirical ancient DNA dataset is a powerful resource to assess genotype imputation, phasing accuracy, and IBD segment detection.

We then co-analyzed the ancient child genome with 107 phased European child genomes from trios in the 1000 Genomes Project to construct a phased high-resolution benchmark dataset for evaluating IBD detection. For this, we developed a multi-criteria filtering framework (incorporating SNP density, inter-marker gap metrics, overlap with difficult genomic regions, and visualizations for inspection). These filters allowed us to obtain excellent-confidence short IBD segments as a benchmark reference set.

We then assessed the methods anclBD and IBDseq when using 1x coverage Medieval ancient genomes to call IBD segments with modern individuals. Optimizing their method parameters allowed us to reduce false positives by orders of magnitude compared to default settings, while maintaining useful sensitivity for short IBD detection. Moreover, the multi-criteria filtering framework further improved the accuracy of IBD segment calling.

Together, our new benchmark dataset and evaluation framework pave the way to heavily optimized strategies for short IBD detection in low coverage genomes, which promises to unlock a powerful new generation of ancient DNA and low-coverage genome analyses.

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Investigating the Pleistocene population history of muskox in EurasiaHeeseung Yeum¹, Mijin Park¹, Donghee Kim¹, Choongwon Jeong¹¹ Seoul National University, Republic of Korea

Presenter: Heeseung Yeum

The muskox (*Ovibos moschatus*) is an Arctic tundra specialist that currently inhabits northern Canada and Greenland. During the Pleistocene, it originally inhabited northern Eurasian Mammoth Steppe. By the end of the Pleistocene, however, it had become extinct in Eurasia while the surviving group, presumably from refugia populations in Beringia, expanded into northern North America following the retreating ice sheets. Extant muskoxen are divided into two distinct subspecies: barren-ground muskoxen (*O. m. moschatus*) in the Canadian mainland and white-faced muskoxen (*O. m. wardi*) in the Canadian Arctic Islands and Greenland. Interestingly, the two extant subspecies are differentially related to a published 21,000 BP East Siberian individual: this individual shares more alleles with the white-faced muskox than with the barren-ground one. Furthermore, Pleistocene muskox populations are thought to have been more genetically diverse than their extant American counterparts, suggesting that they may have played a major role in making genetic structure of extant American populations. Therefore, to fully understand the origins and deep history of extant muskox, we plan to generate whole genome sequencing data for ancient individuals from northwestern Eurasia, a region that remains largely understudied at the whole-genome level.



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