Sessione

Antropologia Molecolare

comunicazioni orali

Synthetic Genomic Data Augmentation for Underrepresented Populations Using Open-Source Large Language Models

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The lack of **high-quality genomic data** from underrepresented populations limits the accuracy and generalizability of population genetic inferences, such as those based on Identity by Descent (**IBDNe**). This study introduces a novel framework to augment Variant Call Format (**VCF**) datasets through the **fine-tuning** and deployment of an **open-source large language model** executed entirely on local computational infrastructure. Drawing on public repositories such as the 1000 Genomes Project, the method generates biologically realistic variant calls that enhance sample size and population diversity without compromising individual privacy or data ownership. The model is fine-tuned on high-confidence variant sites curated from established panels, allowing it to learn patterns of allele frequency, linkage disequilibrium, and haplotype structure. The resulting synthetic VCF entries are post-processed to ensure standard compliance and biological plausibility.

To ensure **privacy, copyright compliance**, and ethical integrity, all training and inference are performed exclusively on institutional servers. No genomic data are transmitted externally. Rare variants and individual genotypes with allele frequencies below a user-defined threshold are filtered out to prevent potential re-identification. Quantitative divergence metrics, including pairwise distance measures and low-dimensional embeddings, such as **PCA and UMAP**, are used to verify that synthetic genomes do not closely replicate any specific real sample. In accordance with best practices, only the trained model weights are shared, while the synthetic datasets remain confidential.

Applied to an underrepresented Eurasian population used as a test case, the augmentation process led to a marked increase in effective sample size while preserving key features such as principal component clustering and pairwise genetic distances. Hierarchical clustering and **IBDNe** analyses performed on the augmented dataset successfully reproduced the demographic signal of the original population within expected confidence intervals.

This resource-efficient and scalable approach improves the representation of diverse human groups in genomic research, enabling more robust analyses of population structure, demographic history, and disease associations. By integrating open-source language models with rigorous privacy safeguards, the proposed framework contributes to making genetic research more inclusive and trustworthy.

Human demography shapes patterns of language diversity across the world

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Human history is written in both our genomes and our languages. The extent to which our biological and linguistic histories align has been the subject of long-standing debate. Transmission with modification can shape genes and languages similarly, while horizontal contact can uniquely affect languages. Demographic contact typically leads to linguistic homogenization, but it can also lead to more or less deliberate processes of linguistic divergence. In turn, language boundaries can influence genetic structure acting as barriers to gene flow. While local case studies provide evidence to anchor specific cultural and demographic histories, we still lack a systematic, global perspective on how population histories and language diversification correspond, and how speakers' dynamics influence the diversity of languages and their features.

To address this, we use the GeLaTo (Genes and Languages Together) database, which links genome-wide data with linguistic identifiers for over 500 populations worldwide. We estimate patterns of genetic distance, excess of homozygosity and admixture to ask three key questions: (1) Do language families correspond to clusters of genetically related populations? (2) Are language isolates associated with genetic isolation? (3) How does demographic history—particularly admixture and isolation—shape structural features of languages? We find trends of genetic cohesion for speakers of the same language families (i.e. related languages that share a common ancestor), with around 20% of populations deviating from this pattern. We explore the contrast between expansive language families vs. language isolates, and find a tendency for genetically isolated populations to speak isolated languages. We finally examine the influence of genetic isolation and genetic contact (admixture) on diverse linguistic features using quantitative linguistic databases, and find that contact favors structural convergence, while genetic isolation correlates with structural diversification.

Our results support hypotheses in historical linguistics, sociolinguistics and linguistic typology, opening new avenues for studying processes of cultural evolution and cultural transmission across human populations.

Semi-super centenarians as a model to test the contribution of natural selection in modulating individuals' predisposition to longevity

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Human life expectancy has significantly increased in post-industrialized societies, largely due to advances in medicine, improved sanitation, better nutrition, and overall living conditions. These changes have particularly reduced the impact of infectious diseases on health and mortality, also pulling survival into extreme old age so that the prevalence of centenarians (i.e. 100 years old or more), semi-super centenarians (hereafter 105+, i.e. 105 years old or more) and super-centenarians (i.e. 110 years old or more) raised considerably. In particular, 105+ represents a relevant threshold, with individuals who reach such a milestone being characterized by an exceptional healthy phenotype. Since genetic factors contributing to human longevity can be considered environmental-specific and are supposed to have been influenced by the peculiar evolutionary history of each population, an evolutionary genomics approach promises to provide useful insights into the genetic determinants of this complex phenotype. Previous studies aimed at investigating the evolution of complex biological traits in human groups that contributed to the formation of the present-day Italian gene pool pointed to differential adaptive processes experienced by the main population clusters observable within the cline of variation distributed along the peninsula. Interestingly, some of the identified adaptive traits were proposed to have evolved in response to pathogen- and/or dietary-related selective pressures and have been mediated by combinations of variants at pleiotropic genes, which have the potential to influence also lifespan. Here we replicate these analyses on a cohort of 81 Italian semi-super centenarians and super-centenarians. In detail, we took advantage of information for around 15 million single nucleotide variants (SNVs) from high-coverage (90x) whole genome sequence data to detect gene networks and biological functions that might have been shaped by the action of natural selection. Then, we compared the identified signatures with those inferred from samples representative of the overall Italian population to test whether 105+ genomes are enriched for putative adaptive loci that may have secondarily modulated individuals' predisposition to delayed onset of age-related diseases, thus playing a key role in promoting extreme longevity.

Adaptive evolution of complex traits regulating insulin signalling and energy metabolism in brown and white adipose tissue mediated adaptation to extreme cold environment in Yakut populations from Northern Siberia

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Complex (i.e., polygenic) adaptive traits are supposed to be the bases of most human biological adaptations to environmental conditions, having allowed *H. sapiens* to occupy and stably live in a vast range of geographical areas including those characterized by harsh climate. Nevertheless, few methodologies have been developed so far to investigate the genetic determinants of these adaptive traits. Here, we coupled multiple methods with a machine learning-based approach to pinpoint combinations of genes simultaneously presenting i) weak signatures ascribable to the action of natural selection and ii) functions linked to the regulation of the same biological trait, as expected under a model of polygenic adaptation. Such a pipeline of analyses was applied to whole genome sequence data for individuals from Northern Siberia belonging to the Yakut ethnic group. The obtained results were compared with those for a population of related ancestry in order to focus on Yakut-specific adaptations and the potential involvement in adaptive introgression events of loci identified as targets of natural selection was tested by considering both Neanderthal and Denisovan sources of archaic alleles. Genes that play a role in the modulation of thyroid hormone signalling and glycerolipid metabolism, along with insulin-related loci, showed patterns of haplotype variation conform to a model of adaptive evolution. These genes are known to contribute to the regulation of insulin signalling and energy expenditure in white adipose tissue, as well as of the correct functioning of brown adipose tissue during cold exposure. In line with this evidence, a gene showing signatures of archaic adaptive introgression from Neanderthals was found to participate to lipids metabolic pathways, while the most validated signal supporting adaptive introgression from Denisovans was observed for a locus whose variants are associated to an increased risk of obesity, Type 2 Diabetes and insulin resistance. Overall, these biological functions were proved to be pervasively modulated during/after cold exposure in several species of mammals, suggesting that adaptive evolution of the underlying genes might have represented one the main driver of complex adaptations evolved by Yakut ancestors to cope with an extreme cold environment.

High-Resolution Kinship Inference with SNPs: A Forensic Tool for Complex and Distant Relationships

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Kinship recognition between anonymous DNA samples is becoming increasingly relevant in forensic science, particularly as national and international DNA databases continue to expand. While Short Tandem Repeats (STRs) remain the gold standard for close kinship analysis, their limitations in detecting distant relationships—due to high mutation rates and a restricted number of loci—necessitate more advanced approaches.

Next-generation sequencing (NGS) has emerged as a powerful tool for personal identification, enabling the simultaneous analysis of a wide range of genetic markers. In this context, we previously developed and evaluated a novel panel of 4,849 Single Nucleotide Polymorphisms (SNPs) specifically designed for high-resolution kinship inference on 150,000 simulated individual pairs, ranging from unrelated to fifth-degree relatives using a combination of the Forrel package in R and supervised machine learning algorithms.

The panel demonstrated strong performance in identifying kinships up to the third and fourth degrees, with recall values exceeding 0.6 even for fifth-degree relationships. Machine learning integration further enhanced accuracy, improving F1 scores by approximately 12.25% and 20% for fourth- and fifth-degree relationships, respectively. Notably, the method achieved over 99% accuracy in distinguishing related from unrelated individuals.

To assess real-world applicability, we evaluated the panel's performance using sequencing data from 2,386 individuals with documented kinship ties up to the third degree, available through the 1000 Genomes Project. Specifically, we processed 1,206 confirmed parent-child pairs, 15 sibling pairs, 36 grandparent-grandchild or avuncular pairs, and 29 third-degree relatives using a Python- and R-based pipelines.

Overall, this study highlights the potential of SNP-based panels, combined with advanced computational methods, to significantly improve kinship inference—especially in complex or distant relationships—offering a robust alternative to traditional STR-based approaches in modern forensic investigations. This approach is particularly valuable in mass disaster scenarios and missing person investigations, where establishing biological relationships is often the only viable path to identification.

Global distribution of the APOE haplotypes in different human populations

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The Apolipoprotein E (APOE) gene has emerged in multiple studies on human longevity and age-related diseases. APOE is expressed in cells of the liver, kidneys, adipose tissue, and immune and central nervous systems. The main role of the protein encoded by this gene is to facilitate the transport and uptake of lipoproteins and cholesterol.
The APOE protein exists in three isoforms (APOE2, APOE3, APOE4), which correspond to three different haplotypes (2, 3, and 4) defined by the combination of two polymorphisms (rs7412-C/T and rs429358-T/C).
These genetic variants reflect differences in amino acid sequence and these isoforms have markedly different functional properties and associations with human health. In particular, the 4 haplotype is negatively associated with longevity and linked to age-related cardiovascular and neurodegenerative diseases, such as Alzheimer's. Recent bio-anthropological studies showed that the 4 haplotype confer reproductive advantages to naturally fertile populations living in highly pathogenic environments. A north-south decreasing gradient of 4 frequency has been observed in Europe and Asia. Conversely, 2 is considered a pro-longevity allele with protective effects against Alzheimer's disease in certain human populations, although it is the least frequent worldwide. Finally, 3 is the most common and functionally neutral isoform.
Previous studies showed that the global variation in APOE allele frequencies reflects the combined effects of natural selection and demographic history. Although there are meta-analyses on the variability of this gene in the literature, it has not yet been fully characterized in many human populations.
This study aims to investigate the distribution of APOE haplotypes in different populations, to provide a full description of the biodiversity of these haplotypes with major implications in terms of health and longevity.
To this end, we selected DNA samples from cohorts of individuals from different human populations characterized by a high level of variations in terms of genetic ancestry, geography and ethnicity. We applied a TaqMan Real-Time PCR Assays to genotype the aforementioned SNPs. Together with data reported in the literature by several studies, the resulting data was used to create the most detailed map of natural variations on this haplotype to date, thus contributing to a broader understanding of its global variability.

Unveiling the Complex Genetic History of Italy from Paleolithic to Middle Age through Ancient Whole-Genome data

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The Italian Peninsula, a crossroads of Mediterranean cultures, has long been recognized for its pivotal role in European history. Despite extensive genomic studies across Europe, the genetic history of Italy remains largely underexplored due to scarcity of ancient whole-genome data spanning limited geographical regions. Here we present a comprehensive analysis of 60 shotgun-sequenced ancient genomes (average coverage of 2.12x) from 21 necropoleis across mainland Italy, Sicily, and Sardinia, spanning about 10,000 years of history, alongside hundreds previously published ancient complete genomes covering from the Paleolithic to the Medieval periods. Leveraging genotype likelihood computations and imputation of missing positions alongside the genome, we achieved robust statistical reconstructions of past evolutionary dynamics despite the inherent challenges of low-coverage data. Our results reveal a complex mosaic of external influences shaping the genetic landscape of Italy. After the arrival of the early farmers in Italy, we observed two different Neolithic-related ancestries highlighting a genetic structure possibly rooted in the Anatolian region, while the Bronze Age marks a significant genetic influx of Pontic Steppe related populations. This migration established a genetic gradient, with northern regions showing higher Steppe-related ancestries compared to the south of Italy. Additionally, we detect a significant genetic contribution of an Iranian Neolithic-related ancestry during the Iron Age, and particularly the Imperial period. These results shed light on the strategic position of the Italian Peninsula at the heart of the Mediterranean Sea, which allowed the arrival of different groups and cultures since prehistory. Phenotypic inference of pigmentation traits (eye, hair and skin color) reveals a shift toward lighter phenotypes beginning in the Iron Age. This study offers unprecedented insights into the genetic history of Italy, elucidating the interplay of migration, environment, and cultural evolution that shaped its rich heritage. These findings contribute to our understanding of the unique position of Italy in the broader context of European genetic history, highlighting the methodological innovations and interdisciplinary approaches necessary for exploring complex demographic dynamics.

Uncovering human mobility and genomic variability in Medieval Central Italy: Insights from ancient DNA and strontium isotope analyses

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During the Middle Ages, the Lazio region in Central Italy served as a strategic crossroad of different cultures. This study investigates two populations from this area: Santa Severa (7th–15th century CE), a key Mediterranean trade hub, and Allumiere-La Bianca (15th–16th century CE), among the earliest alum mining settlements in Italy.

To explore patterns of genetic diversity and individual mobility, we integrated ancient DNA (aDNA) analysis with strontium (Sr) isotope. Sr isotopes from tooth enamel reflect an individual's childhood geological environment, thus informing on geographical origins.

We extracted aDNA from teeth and petrous bones using a silica column-based protocol, and prepared double-stranded genomic libraries for shotgun sequencing. Low-coverage genomes (0.1X) were obtained for 69 individuals: 56 from Santa Severa and 13 from Allumiere-La Bianca. Moreover, from Santa Severa to assess genetic continuity over time we analysed 8 Etruscan (6th–3rd century BCE) and 5 Roman (3rd century BCE–5th century CE) individuals.

The Etruscan and Roman genomes from Santa Severa fall within the genetic variability of previously published Etruscans and Roman populations, distinct from the Italian Medieval samples (including individuals from Santa Severa). Moreover, between the Early Medieval population of Santa Severa and the Late Medieval population of Allumiere-La Bianca a genetic difference was also observed suggesting a geographical and temporal differentiation during the Middle Ages. Several Medieval individuals also exhibit non-local strontium isotope signatures, suggesting extra-regional origins.

Ongoing research will involve high-resolution analyses of ancestry and population structure through genotype imputation, including identity-by-descent (IBD) sharing, which is essential reconstructing the dynamics of recent populations.

This research is funded by PRIN2017 (20177PJ9XF), MUR 113/91 (PANN20_00370) and the support of the ISIS@MACH ITALIA Research Infrastructure.

Genetic and Cultural Dynamics of post-Roman Central Europe

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The transition from Late antiquity to the Early Middle ages in Central Europe has traditionally been interpreted as a period of large scale migrations and conflicts between northern "barbarians" and the retreating fringes of the Roman Empire. To the very same timeframe, however, can be traced the origins of many modern towns and villages, as well as the appearance of new cultural practices.
In order to explore the cultural and demographic dynamics of this crucial phase, we generated 248 new whole genomes sequences from two regions in Southern Germany: the Danube-Isar area, which was part of the Roman Empire until the early 5th century, and the Rhine-Main area, that was abandoned at the end of the 3rd century. We focused on individuals from the 5th to the 7th century, but included Late Antiquity (3rd and 4th century) samples to provide insights into the Late Roman period.

Altheim-Essenbach, our main study site, was likely founded by a group with Northern European ancestry in the 5th century, and we detected a demographic shift in the following century triggered by the integration of newcomers with an ancestry that is typical of local roman[ised] settlements. By the 7th century, the fusion of these diverse ancestries had created a genetic diversity resembling the one still observed in modern Germany. We reconstructed a large pedigree network spanning up to seven generations and developed a novel strategy to infer the ancestry of unsampled individuals, which allowed us to identify immediate intermarriage between local and incoming groups.
Our results highlight rapid integration and cultural assimilation of groups with distinct ancestries following the downfall of roman power in the area; and show that these Early Medieval communities were organised around small families exhibiting a bilateral or loosely patrilineal descent, avoided kin marriages and practiced monogamy. These cultural practices, that began during the Late Roman period, will go on to influence Europe in the centuries to come.

Ancient and archaic mobile elements underline global evolutionary trends and local functional adaptation in modern human populations

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Transposable elements (TEs), once regarded as "junk DNA," have recently been recognized for their significant roles in gene regulation and as valuable markers for understanding human evolution. In this study, we explore the functional and evolutionary signatures of polymorphic TEs in modern human populations, with a focus on gene-associated TEs identified in ancient and archaic human genomes. To achieve this, we analyzed whole genome sequences from eight ancient and archaic Eurasian human samples covering the last 80.000 years of human evolution and with a coverage of 4-42X. MELT and TypeTE softwares were employed to identify both reference and non-reference TEs belonging to the Alu, LINE, and SVA families. Following standard quality control, TEs present in at least one ancient/archaic individual were searched within 20 non-admixed populations from the 1000 Genomes Project, with a frequency greater than 0.05 in at least one modern population. Gene-associated TEs were then examined for local signals of positive selection (rEHH) and adaptive introgression (VolcanoFinder), with additional dating of their emergence in the human lineage using a modified version of the GEVA (Genealogical Estimation of Variant Age) algorithm, implemented for more accurate assessment of TE origin dates and confidence intervals. Functional enrichment and gene network analysis were conducted through STRINGdb, DAVID, and PANTHER. Our results reveal that polymorphic TEs from archaic and ancient human populations significantly influence key biological pathways and phenotypic traits in modern humans, particularly those related to immune function, brain development, mental health, anthropometric traits, and cardiovascular health. In some regions, these effects were geographically restricted, while in others, they exhibited clear clines reflecting known human migration patterns.

Genomic analysis of the Vesuvius victims in Pompeii: the case study of the "Room of the Skeletons"

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The ruins of Pompeii, discovered in the 16th century, have been systematically excavated since 1748, offering invaluable insight into ancient Roman society and yielding exceptional discoveries, such as the "House of the Garden" in Regio V. Excavated in 2018, this domus is renowned for the discovery of a charcoal inscription that revised the date of eruption, as well as for its rich decorative elements and numerous finds. In one of the rooms – referred to as "Room of the Skeletons" – the commingled remains of at least 11 individuals, including adult woman and children, were uncovered.

In this study, we genetically characterized 18 bone and tooth samples to determine the number of victims, their biological profiles, genetic heritage, and familial relationships. Additionally, we analyzed a tooth of the "fugitive" discovered near the House of the Garden. By integrating paleogenomic analysis with forensic methodologies, archaeological context, and anthropological assessments, we identified 10 individuals: three adult women and six subadults who sought refuges in the room, and the male who perished while attempting to escape from the eruptive fury of Vesuvius. Kinship analysis revealed varying degrees of relatedness among the victims.

Population genomic analysis demonstrated high genetic diversity within the group, with unrelated individuals exhibiting distinct mitochondrial and Y-chromosome haplotypes and diverse genetic ancestries. Notably, a mother and daughter pair showed an unexpectedly high genetic distance, likely attributable to paternal lineage.

Finally, pigmentation traits and functionally relevant SNPs were analyzed to infer phenotypic features and potential genetic disorders of the victims.

Genetic Profile of the Etruscans from Felsina: genetic changes and Biological Interactions with Neighboring Populations from the Villanovan to the Imperial Period

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During the transitional phase between the Late Bronze Age and the early Iron Age the Villanovan culture emerged in central Italy, within the regions that would later constitute the territory of the Etruscan civilization. In this context, Felsina (modern-day Bologna), part of the northern expansion into the Po Valley, was key for the Etruscan population, as a major center of cultural and commercial exchange during the Iron Age.
Genomic studies on Etruscan-associated human remains from central Italy show that the Etruscans carried a local genetic profile, with only a few individuals displaying ancestry associated with Central European populations. In addition, archaeological evidence points to limited interactions between the Etruscan communities of Felsina and culturally Celtic-related groups during the Hallstatt period (XIII-V centuries BCE), before the historically documented Gallic expansion during the La Tène period (VI-I centuries BCE). To shed light on the genetic relationship between the Etruscan population, the contemporary Italic population and transalpine groups, this study focused on the genomic analysis of 77 ancient individuals excavated from 10 archaeological sites in Bologna, dating from the 9th century BCE to the 3rd century AD.
Genome-wide data were generated, and enriched using a capture protocol targeting approximately 1.24 million informative nuclear SNPs. Individuals with sufficient coverage and low contamination levels were included in population genetic analyses: Principal Component Analysis (PCA), ADMIXTURE modeling, F-statistics, qpAdm admixture modeling, and community structure analysis based on Identical-by-Descent (IBD) segments.
The majority of Etruscan samples from the Po Valley exhibit a genetic profile consistent with local ancestry, in line with patterns observed across the Italian Peninsula during the Bronze and Iron Ages. However, 17% of individuals show a non-local genetic component, associated with Central and Northern European populations, and display increased affinity to steppe-related ancestry. The cluster of individuals showing the closest affinity to Central-Northern European groups stands out as genetically distinct from the local variation, further supporting the hypothesis of gene flow and biological interactions between Felsina and Celtic-related communities. This genetic signal is detected in individuals as old as the 8th, suggesting extensive contacts between them starting at least by the Hallstatt period.

Diet and Lifestyle in the Bronze Age through the Lens of the Oral Microbiome: The Case study of "Terramare" culture

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The European Bronze Age presents a cultural puzzle, situated between widespread patterns and highly localized expressions1. While regional differences in ceramics and metalwork have received much scholarly focus, aspects like subsistence strategies remain less explored. It's often assumed that Bronze Age diets continued Neolithic practices, with little change in the use of plants and animals. Discussions on food typically relate to broader questions of social structure, such as age, gender, or inequality. In this period more than others, food practices are often seen as reflecting or reinforcing social boundaries. In Italy, late prehistoric dietary studies highlight notable regional differences, but efforts to examine variations across social groups have yielded limited results.
Recent research has shown that the dental calculus microbiome is an exceptional source of information on diet, health, and lifestyle. Emerging evidence suggests that shifts in culture and diet—from hunter-gatherer to Neolithic societies can be traced through oral microbiome. This study aims to investigate through metagenomic analysis the composition of the oral microbiome of three populations from the Terramare culture and combine these data with stable isotopes from bone collagen. These results will be compared with data from earlier hunter-gatherer, Neolithic, and contemporary Bronze Age societies. The objectives are to: (i) assess whether dietary habits reflect continuity or change between the Neolithic and the Bronze Age; (ii) identify distinguishing features of the Terramare populations compared to other Bronze Age groups; and (iii) explore aspects of social structure through insights into diet and health.
We collected 70 dental calculus samples from four archaeological sites in Northeastern Italy. The samples underwent shotgun sequencing and were analysed for their metagenomic profiles following specific authentication criteria6. Samples were also analysed through stable isotope analysis to correlate dietary information with oral bacteria. Preliminary results already highlight the presence of a distinctive oral microbiome community in Bronze Age populations, with further differences observed even within "Terramare" groups. These findings underscore the importance of combining complementary biomolecular, isotopic, and archaeological analyses to gain a more comprehensive view of intra-population lifestyle diversity.

Life and death in an ancient female monastic community: the case study of a mummified nun from Fara in Sabina, central Italy (17th century).

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The municipality of Fara in Sabina (Rieti), hosts an important monastic community of Franciscan nuns dating back to the 17th century that discloses a unique collection of 18 naturally mummified and remarkably well-preserved nun remains. These offer a rare opportunity to explore the lives, health and ancestry of women from different social backgrounds who lived in strict seclusion. In particular, the genetic analysis of one of these nuns, identified as "Fara 9", is the subject of this study. In order to proceed with the genetic analysis, a tooth (pulp and tartar) and a calcific pulmonary nodule were sampled.

The preliminary sequencing screening showed a remarkable endogenous DNA conservation. Due to these promising results, we performed deep sequencing of the genomic libraries derived from the nodule, generating approximately 50 million reads and a coverage of 0.5X on the nuclear genome. Sex assignment confirms the individual was a female. A mitochondrial DNA consensus sequence was generated using Schmutzi (mean coverage: 181.5X), allowing assignment to the haplogroup H1a3a. This lineage is predominantly found in present-day populations of Germany and Norway, with the earliest known specimens dating to the 10th–13th centuries CE and retrieved in Norway. The analysis of the mitochondrial DNA also highlighted the genetic predisposition to various pathogenic mutations, including bullous pemphigoid, an autoimmune pruritic skin disease. Its presence could also be supported by leg lesions that were covered with medical bandages prepared according to the traditional monastic pharmacopoeia, which persisted in the mummified remains. The presence of Parvovirus B19, the virus responsible for the childhood rash erythema infectiosum (fifth disease), was also detected and ascribed to genotype 3, regarded as ancestral to the currently circulating genotype 1. Finally, further in-depth analyses will allow for the assessment of whether the calcified pulmonary nodule may be associated with an infection by Mycobacterium tuberculosis.

The individual "Fara 9" is just the starting point of a multidisciplinary project on the whole ancient monastic community, which represents an invaluable biological archive. By combining genetic and bioanthropological data, this research will provide insights into past health conditions, pathogen evolution, and human adaptation, and it will help reconstruct the personal and collective histories of these nuns, their kinship, and genetic legacies.

Genetic Insights into Burial Practices and Population Dynamics at the Punic-Roman Necropolis of Ortacesus (SU, Sardinia)

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The Punic-Roman period was a time of significant change and increasing interregional contact, due to the growing influence of Carthage and the shift in political and cultural domination that occurred with the Roman conquest of Sardinia. The necropolis of Mitza de Siddi (Ortacesus), a rural burial site containing approximately 150 tombs dated to the 3rd century BCE – 4th century CE, was excavated by the Superintendence for Cagliari and Oristano between 1994 and 2005. The site features varied tomb types and both inhumation and cremation practices. While a portion of the grave goods has been published, little is known about the kinship networks represented within the necropolis. It also remains unclear whether the diversity in burial modalities and tomb architecture reflects the coexistence of culturally distinct groups of different ancestries.

Through a research agreement between the Superintendence and the University of Cagliari, our research team has recently resumed the study of the necropolis, conducting a systematic analysis of both the associated material culture and anthropological data from inhumation contexts. While traditional archaeological and osteological methods have provided valuable insights into burial practices, genetic analysis offers a more direct approach to understanding kinship relations, population structure, and admixture patterns. We thus applied aDNA analysis to 36 specimens, preferentially petrous bones, with teeth as a second option. To explore the biological basis of the funerary rite, individuals were selected based on burial proximity, evidence of sequential use of burial shafts, architectural anomalies, and the presence or absence of grave goods.

DNA has been extracted from 19 individuals in a clean lab following strict aDNA protocols optimized for highly fragmented molecules. Molecular sex determination was possible for 16 individuals, aiding our understanding of the funerary rituals. Currently, results confirm a lack of distinction between sexes in funerary treatment. Ongoing experimental steps include target enrichment of mitochondrial and nuclear markers.

The findings will contribute to discussions on kinship, mobility, and identity in ancient societies, particularly regarding the influence of long-distance contact on local communities. Ultimately, this study offers a nuanced perspective on the social fabric of Ortacesus, enriching our understanding of Punic-Roman burial practices and their social implications.

Crossing the Sea: a paleogenomic perspective on the main Italian islands

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The Mediterranean has always played a key role in the peopling processes and cultural shifts that have shaped Europe over the centuries and millennia; among the areas of particular interest for the study of these dynamics, the large Mediterranean islands offer a privileged context due to their strategic position and abundance of archaeological evidence. However, despite the important role played by these islands, their contribution to the genetic history of the Mediterranean certainly requires further investigation: the extension of the sampling and the time horizon, as well as the increase in the information potential of the genetic data produced, would help to overcome the limitations of the studies already available in the literature.
This is the context for the PRIN2020 "Crossing the Sea" project, which is mainly aimed at reconstructing the genetic history of the main Italian Mediterranean islands from the Upper Palaeolithic onwards, through the analysis of ancient and modern genomic data using a multidisciplinary approach involving archaeologists, anthropologists, palaeoanthropologists and population geneticists.
The paleogenomic analyses involved more than 260 ancient individuals from Sicily and Sardinia, from which 55 were selected so far, according to their molecular preservation, and yielded whole genome data with a mean coverage between 0.1x and 5.3x.
Starting from Sicily, different computational approaches, such as genotype imputation and genotype likelihoods methods, were applied according to the genome coverage. Subsequent analyses including principal component, admixture, IBD and demographic modelling were performed or are in progress.
The results so far produced made it possible to outline the genetic history of Sicily and to compare the evidence obtained with what happened in the past in the Italian peninsula as well as in Europe.
Future analyses will involve the genome data obtained for the Sardinian samples and, in addition, in-depth studies will be carried out on specific sites to investigate local dynamics in a greater detail.

Crossing the Sea: Contextualizing maternal history of Sicily in the Mediterranean. First Results

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Situated at the crossroads of Europe, Africa, and the Near East, Sicily has played a central role in Mediterranean population dynamics for millennia. Here we present the first results from an integrated study of mitochondrial DNA variation in Sicily, combining 41 newly sequenced modern mitogenomes with 19 ancient individuals spanning key prehistoric and historic periods. These data are analyzed in the context of several hundred comparative mitogenomes from Sicily and other regions of the Mediterranean and Europe.

DNA was extracted from teeth and petrous bones and sequenced. The bioinformatic pipeline included consensus sequence extraction, haplogroup assignment, multiple sequence alignment, phylogenetic tree construction, and visualization in R.

Our preliminary analyses reveal both continuity of certain maternal lineages over time and clear signatures of gene flow associated with known demographic events. Characteristic early diverging haplogroups like HV, U5, U8 are associated with early peopling of Europe.

Specifically, several individuals carrying haplogroup HV were linked to the Phoenician culture, such as those discovered in Mozia in western Sicily, while one individual with HV was associated with the Punic culture at Selinunte. Two other HV individuals are modern samples from Agrigento and Trapani, representing the current populations of Sicily.

Additionally, haplogroup U1 was identified in southern Sicily in remains dating to the 1st century BCE, and U2 was found in the San Teodoro Cave, dating back to the Upper Paleolithic. Haplogroup U5 appears in a modern sample from Caltanissetta and other samples dating to the Iron Age and Sicanian periods. Haplogroup H was the most prevalent haplogroup in our overall dataset and one of the most common in Europe, is also found in ancient African samples.

Patterns of haplogroup diversity and phylogeographic affinity highlight the layered nature of Sicily's maternal ancestry, reflecting its long-standing role as a genetic and cultural contact zone in the central Mediterranean.

This study offers new perspectives on the demographic history of Sicily and contributes to broader efforts to reconstruct mobility and admixture across the Mediterranean world through integrative archaeogenetic approaches.

"SHOVELING into the North Italian Bronze Age with an integrative approach to unearth the evolutionary origins of a non-metric dental trait": a PNRR 2022 project

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We observed in the necropolises of the Terramare (Northern Italian Bronze Age populations, 1859-950 BCE) a large incidence of shovel-shaped incisors, a dental non-metric trait currently common among extant Asians (65%-97.5%) and Native Americans (99.5%), but rather rare in other populations. Tooth shoveling was not uncommon in prehistoric Europe, yet the frequency we found in the Terramare was quite unusual. Different evolutionary explanations can be proposed to explain this phenomenon, keeping in mind that the trait is considered due to a mutation in a pleiotropic gene, EDAR, which might have arisen in China or Beringia about 20000 years ago and was possibly under positive selective pressures: (i) coeval migrations from Asia coupled with demographic events; (ii) independent ancestral inheritance from a hominin; (iii) different mutations in EDAR and selection due to diverse possible agents.

To test the suggested hypotheses, we have proposed an integrative approach which includes anthropological investigation, paleogenomics and stable isotope analyses (C, N, S, O and Sr). We examined and collected samples from individuals with and without shovel-teeth from several Terramare's necropolises from Veneto (Castello del Tartaro, Bovolone, Franzine Nuove, Olmo di Nogara), plus individuals from other regions and periods. Dental metric and non-metric traits were studied on more than 300 individuals, while more than 350 human samples were sampled for isotope analyses, and 25 individuals were selected for palaeogenomic investigation with target enrichment.

We investigated the genetic basis of dental shoveling by analyzing the rs3827760 variant of the EDAR gene, known for its association with shovel-shaped incisors. Among the 25 ancient individuals analyzed, those for whom genotype data were obtained carried the ancestral allele not associated with the trait, although some showed clear morphological evidence. This suggests additional mechanisms beyond this phenotype. Here, we will show the results of the analyses on selected individuals to highlight from an ancient perspective the evolutionary aspects behind the development of a pleiotropic gene and the interplay with the environment.

Multidisciplinary Characterization of an Etruscan Male with possible DISH from Sasso Marconi (Bologna, Italy), 5th -4th centuries BCE

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The Etruscan civilization and its populations continue to raise questions, particularly regarding their origins, cultural integration, health burden and related medico-surgical practice. In this study, we present a multidisciplinary analysis of the TB2 individual, discovered in 1970 in Sasso Marconi (Bologna, northern Italy) and associated with the Marzabotto necropolis, dated to the 5th–4th century BCE. The skeleton, presently exhibited in the "Pompeo Aria" Etruscan National Museum of Marzabotto and still partially interred, is well-preserved and anthropologically ascribed to a mature/senile male adult.

Initial paleopathological evidence suggests the individual may have been affected by Diffuse Idiopathic Skeletal Hyperostosis (DISH), a diagnosis supported by skeletal features such as ligamentous calcifications and enthesopathies, as well as the presence of an object in the burial that could be interpreted either as a cane or as a scepter. While this may suggest mobility issues, its placement in the grave diverges from traditional Etruscan mortuary symbolism, making it more likely to have been a power insignia, thus indicating a high-status individual. We performed whole-genome sequencing, achieving 1.1x nuclear and 91.1x mitochondrial coverage, with 43.26% endogenous DNA. Using genotype imputation with GLIMPSE v2, we characterized over 42 million SNPs with a mean genotype probability of 99.5%.

These data were used to infer the individual's ancestry and general clinical profile. Given the known correlation between DISH and metabolic disorders such as diabetes and obesity, our focus was on variants related to these conditions. The individual displays a predominantly heterozygous genotype at loci potentially associated with DISH in genes: COL6A1, FGF2, BMP4 and PPP2R2D. As the skeleton remains partially embedded in burial sediments, X-ray imaging will be performed to support the paleopathological diagnosis, while stable isotope analysis (^{13}C and ^{1}N) is programmed to investigate dietary habits. A protein- and fat-rich diet, if confirmed, may constitute a supportive element in the diagnostic process of the suspected pathology as well as be consistent with the elevated social status suggested by the burial context. This integrated, transdisciplinary approach provides a valuable glimpse into the life history and health condition of an elite Etruscan individual and outlines a step-by-step framework for the comprehensive characterization of ancient humans.

Integrating bioarchaeology and paleogenomics to investigate early-life stress in Leopoli-Cencelle (RM, 9th–16th C.E.).

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The study of childhood in past populations has recently expanded, particularly in relation to the dissection of the harsh living conditions triggering the high infant mortality rates, such as high population density, poor hygiene, and repeated exposure to stressors. As for many complex traits, the genetic liability to certain anthropometric characteristics could be impacted by those detrimental environmental factors, limiting the expression of genetically regulated phenotypes. This project investigates childhood adversity by integrating skeletal and genomic data from the medieval cemetery of Leopoli-Cencelle (Rome, 9th–16th C.E.), notable for its high proportion of non-adult individuals (38% of 877 burials). A representative sample of 85 child skeletons (aged 0–19 years) was selected based on the preservation status and distribution across age groups. Macroscopic analysis focused on growth patterns and skeletal stress markers detected either by osteological and imaging analyses to identify signs of fractures, dysmorphic changes, metabolic disorders, congenital anomalies, and deficiency-related conditions, thereby enhancing both the accuracy of health and developmental assessment and providing information on biomechanical stress. Ancient DNA (aDNA) was extracted from the petrous bones and teeth of nearly 70 selected samples. The sequencing results showed good endogenous content in multiple cases. By focusing on the whole genome variability, the detailed osteobiographies, and leveraging reference data from large present-day cohort such as UK Biobank, we will analyse the SNP heritability and association models of multiple phenotypes related to the biological consequences of childhood stress in order to identify risk loci impacting on the childhood resiliency in ancient time.

The research was financed by MUR "PRIN 2022 - Prot. 202255L4YW", "Early-life adversities: writing a biological history of childhood through a transdisciplinary approach" and the financial support of Regione Lazio through ISIS@MACH facilities.

Echoes of the Goths: Archaeological, Anthropological and Biomolecular Insights from two Northern Italian communities during the 4th-6th century CE

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The Migration Period saw the arrival and settling within the Roman Empire of numerous "barbaric" groups. Several territories in Northern Italy experienced significant cultural and political changes, including those gravitating around the ancient Po River delta, due to their strategic location that facilitated defense and movement of people and goods. The demographic impact of these events, however, is unclear and has long been debated.
The Goths, particularly the Ostrogoths, played a crucial role in this transformation. After the fall of the Western Roman Empire, the Ostrogoths, led by Theodoric the Great, established a kingdom in Italy. This period saw a fascinating blend of Gothic and Roman cultures, with the Goths adopting many aspects of Roman administration and lifestyle.
Archaeological evidence, such as grave goods and settlement patterns, provides insights into the ethnocultural identities of these groups. For instance, findings from cemeteries in Northern Italy reveal a blend of Gothic and local traditions, indicating a degree of cultural assimilation.
The sites of Chiesazza di Ficarolo and Chiunsano (Rovigo, Italy), dated between the 4th and 6th centuries, may be an example of this integration. Excavations at these settlements, located between the two main branches of the Po River with direct access to the hydrographic network and land routes, uncovered inhumation burials that represent the first evidence of the Goths' arrival in the Polesine territories. Anthropological analysis was conducted on 56 inhumations from Chiunsano and Chiesazza di Ficarolo. These burials are modest, lacking grave goods, and the individuals are oriented east-west in a typically Christian manner. However, there are intriguing exceptions, such as two unique burials with rich Ostrogothic grave goods at Chiunsano and a group of deviant burials at Chiesazza di Ficarolo.
Twenty-four individuals have been sampled for biomolecular analysis, currently ongoing at the aDNA laboratory of the University of Mainz.
In this presentation, we aim to share the results of the anthropological investigation on the population composition and health conditions, and preliminary findings from the biomolecular analysis.
Biomolecular analysis will help in identifying the ancestry of the individuals and understanding demographic and population dynamics during this historical period marked by the settlement of barbarian communities in these territories.