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EDITORIAL



Reconstruction of the human peopling of Europe: a genetic insight

Despite the long-lasting and interdisciplinary work carried out by different research fields, such as archaeology, history, linguistics, genetics, palaeoanthropology, molecular anthropology, and more recently, palaeogenomics, the reconstruction of the human peopling of Europe is still a highly debated topic. The present issue – composed of review articles and research papers – is largely devoted to discussing and enlarging our current knowledge on the ancestries that contributed to the European genetic make-up through time.

The three review articles presented in this issue summarise the latest literature on the palaeogenomic findings dealing with Prehistory in Europe and its surroundings in an attempt to explain the current state of understanding on the peopling of this continent. A special focus is devoted to the early settlement of Sardinia, an island within the Mediterranean Basin with a distinct genetic history compared to the rest of Europe. The other review contributions deal instead with ancient population movements and demographic changes spanning between the Palaeolithic and the Iron Age, as well as with the processes of animal domestication and disease emergence and transmission during prehistoric times. In fact, while most investigations target human material, there is a growing interest in working on non-human species to shed light on the legacy of European populations on past ecology.

The contribution by Serrano et al. (2021) provides a comprehensive overview of European Prehistory as became possible through the use of the latest next-generation sequencing (NGS) techniques. The authors focus on main discoveries deriving from the analysis of human genome-wide data as well as non-human genetic data to better understand the demography of ancient European populations, from the Upper Palaeolithic to the Bronze Age. Archaeological evidence has already shown a complex pattern of demic and/or cultural diffusion since the Upper Palaeolithic, which becomes more evident during the Neolithic and the Bronze Age transitions. The study of ancient DNA has been fundamental in understanding whether cultural changes occurred due to the migration of people or ideas and culture. Moreover, the analysis of ancient genomes of domestic animals and pathogens provides deeper insights into the processes that accompanied the peopling of Europe. This comprehensive approach has been crucial for answering questions related to the Neolithic Revolution, for example by showing that the spread of agriculture and herding techniques has largely been the result of people's movement, despite being driven by cultural diffusion in some areas (Jones et al. 2017; Mittnik et al. 2018). Therefore, ancient whole-genome studies provide a transformative tool to characterise prehistoric events not

previously identified and to challenge traditional hypotheses. If complemented with other molecular, chemical, and archaeological evidence, those studies will lead to a more realistic and comprehensive description of Europe's past.

The review by Feldman et al. (2021) provides an in-depth assessment of the genetic transformations in three regions, which played a crucial role in the formation of the genetic landscape of Europe through time: the Near East, the western Eurasian Steppe, and North East/North East Europe. The paper focuses in particular on demographic shifts, which occurred in these areas between the Epipaleolithic/Mesolithic and the Iron Age, as highlighted through the study of ancient DNA. The understanding of the genetic diversity in Europe's surrounding regions has proved to be very important to properly address the various demographic processes that led to ancient genomic transformations within Europe. These studies have shown a general trend of genetic homogenisation in the Near East and the western Steppe during the aforementioned periods of time, while North-Eastern Europe offers a unique demographic history, with a later settlement and repeated genetic connections with Siberia. Further genomic data from historical periods and older groups of hunter-gatherers in the regions surrounding Europe will provide an even clearer picture of human mobility and connections between Europe and Asia.

The review by Calò et al. (2021) focuses on the history of Sardinia and tackles open questions regarding the origins of its peoples, their relationship with neighbouring Corsican populations, and their intra-population genetic variation. The authors describe the most relevant publications on population genetic analyses of Sardinians, based on different analytical approaches (classical markers, molecular data, and NGS data). What emerges from these studies is that Sardinians carry a unique genetic ancestry and differ from what is observed in all other present-day Euro-Mediterranean populations. During the Neolithic, Sardinia experienced a large demographic expansion mediated by the arrival of new ancestry related to early European farmers. However, the impact of acculturation processes on autochthonous people is suggested by the presence of many haplotypes related to preceding hunter-gatherer groups. This expansion was followed by substantial genetic isolation in Sardinia, while Corsica seems to have received more gene flow from the mainland. For this reason, Sardinians show a significant genetic distinctiveness at a continental level, paired with an overall homogeneity at a regional scale. However, this isolation was not complete since it is possible to find signals of steppe- and Northern African-related ancestries in the genomes of contemporary Sardinians (Fernandes et al. 2020; Marcus et al. 2020).

The five research papers in this special issue deal with an ample array of topics, which include: the mitogenome analyses of Upper Palaeolithic skeletal remains that describes the phylogenetic relationships between extinct and extant western Eurasian populations; a genome-wide study that portrays the genetic composition of a rural community in Imperial Rome; the investigation of possible sex-biased admixture following the Bronze Age expansion from the western Eurasian Steppe into Central Europe; the study of mitochondrial DNA (mtDNA) sequence variability in ancient Punic individuals from Tharros in the Sinis peninsula (western Sardinia) in comparison to present Sardinian populations from Cabras and Belvi, in Oristano and Nuoro provinces, respectively; and the investigation of possible correlations among surnames, dialects and molecular data in the northern Italian population of Trentino.

The research paper by Via et al. (2021) aims to reconstruct the mitochondrial genomes obtained from skeletal remains found in three of the most important Upper Palaeolithic archaeological sites in Italy: Paglicci (Apulia), San Teodoro (Sicily), and Arene Candide (Liguria). The study explores the phylogenetic relationships of these three newly reconstructed mitogenomes in the context of ancient and modern-day mtDNA variability in west Eurasia. The new sequences not only derive from different geographical contexts but also different chronological periods: Paglicci 12 dates to 29,000 years BP, while San Teodoro 2 and Arene Candide 16 date to 14,700 and 12,820 years BP, respectively. Their phylogenetic position fits well with the already known distribution of mtDNA variability in space and time during the Late Pleistocene and early Holocene in Europe (Posth et al. 2016). However, the newly produced data helps to better characterise the mitochondrial diversity existing in Italy during the Upper Palaeolithic. Sampling an ear ossicle and two petrous portions of the temporal bone allowed the retrieval of high-quality genetic data from highly degraded specimens due to the southern latitudes and/or old chronologies. Further studies on selected Italian specimens will provide a deeper comprehension of the regional dynamics between hunter-gatherer populations and their possible cultural and genetic contacts and interactions.

The research article by Scorrano et al. (2021) provides a comprehensive genetic analysis of European Bronze Age individuals to tackle the question of the so-called “steppe” expansion, a population movement ultimately related to Yamnaya pastoralists from the Pontic-Caspian Steppe that reached central Europe during the early 3rd millennium Before Common Era (BCE). The authors re-analysed 864 published ancient individuals through the joint study of mtDNA, Y-chromosome, X-chromosome, and autosomal DNA to investigate the possibility that the steppe-related admixture was sex-biased mediated. After confirming the large-scale impact of this admixture event in shaping the genetic makeup of Bronze Age Central European populations, the study suggests a male-driven mode of transmission for the steppe-related ancestry specifically in populations associated with the Corded Ware Complex. No detectable shift was observed in mtDNA sequence diversity before and after the spread of

steppe-related ancestry while Y-chromosome haplogroups were largely replaced. Moreover, by comparing the proportion of the steppe-related genetic component on the autosomal DNA, which is inherited from both parents, vs. the proportion on the X-chromosome, which is mainly inherited from mothers to offspring, they confirmed a pattern of male-biased admixture. This result does not reach significance, possibly because of the low resolution when analysing the limited amount of markers present on the X-chromosome compared to the autosomal chromosomes. Larger sample sets as well alternative analytical methodologies are necessary to assess the robustness of this signal for which a lively debate already exists (c.f. Goldberg et al. 2017; Lazaridis and Reich 2017).

The contribution by De Angelis et al. (2021) reports ancient genomic data of human remains excavated at the Quarto Cappello del Prete (QCP) archaeological site. This site is dated between the first and the third centuries of the Common Era (CE) and is located in what was during the Roman Imperial period a rural area at the outskirts of Rome. The authors generated shotgun-sequencing data from 25 skeletal remains and, after quality filtering, they were able to perform population genetics analyses on seven individuals. Interestingly, the picture that emerges from the newly generated dataset suggests that the genetic ancestry present at QCP was largely different from that previously reported for 48 roughly contemporaneous individuals from the Empire’s capital and its surroundings (Antonio et al. 2019). The latter study reported a substantial genetic shift towards eastern Mediterranean populations compared to previous Iron Age and Roman Republican period individuals. On the contrary, the QCP individuals are mainly shifted towards northern African ancestries, with only one individual falling within the present-day Near Easterner genetic variation. This suggests that the genetic landscape of the population living in Rome and its large metropolitan area during the Imperial period was more heterogeneous than previously described. Even though this could be expected due to the high level of human mobility towards Rome at that time (e.g. De Ligt and Tacoma, 2016), the exact geographic origins of those incoming ancestries and the extent to which their arrival could be linked to specific historical events are still unclear. Additional ancient genomes from central Italy would be pivotal to assess if the genetic contributions to this region were derived also from other territories of the Roman Empire.

In their paper, Cilli et al. (2021) analysed ancient individuals spanning from the Middle to the Late Punic period from the southern necropolis of Tharros, which is located on the Capo San Marco area in the Sinis Peninsula in Sardinia (Fariselli 2014, 2017). The mtDNA HVS-I and coding region SNPs were characterised for 14 ancient human remains and 74 modern individuals from Cabras and Belvi. To study the patterns of genetic variation between Punic-related individuals and present-day Sardinians, the authors selected from the published literature a set of 5,590 mtDNA sequences across the Euro-Mediterranean area. In PCA space, present-day populations from Cabras and Belvi fall within the genetic variation of other modern Sardinians, while Punic-related

individuals from Tharros fall in an intermediate position between modern North African populations and Southern European Iberians. To better understand if past demographic events may have shaped the maternal genetic variability, the Punic-related individuals and modern populations from Cabras and Belvì were compared to 127 previously published ancient Sardinian mtDNA sequences dating from the Mesolithic to the Medieval period. Present-day populations from Cabras and Belvì form a clade with pre-Phenician Sardinian populations dated between the Neolithic and the Nuragic periods, while Punic-related individuals from Tharros form a cluster with previously published Punic-associated individuals from Sardinia (Marcus et al. 2020). The study demonstrates that female mobility from North Africa to Sardinia was associated with Punic communities.

The paper by Boattini et al. (2021) performs a comparison between surnames, dialects, and genetics in Trentino, a region in northern Italy that is well-suited to perform this type of study thanks to the large availability of such comparative data. Multivariate methods and correlation tests were implemented and identified a significant association between surnames and dialects in Trentino. This observation is found to hold independently of geographical distance since both categories are representative of cultural markers. However, there is no significant correlation between any of the inspected molecular markers (mtDNA, Y-chromosome, and autosomal Short Tandem Repeats) and surnames or dialects. This suggests that, at least for the northern Italian region of Trentino, surnames cannot be used as reliable indicators of genetic relationships. It would be important to expand this comparative study to other Italian, as well as European, regions (e.g. Manni et al. 2008) to verify if this represents a local pattern or if the conclusions drawn here correspond to a general phenomenon.

As a whole, this special issue links reviews and research papers to highlight the use of molecular approaches in describing the dynamic demographic processes that accompanied the peopling of Europe. It represents an attempt to open new research directions for scientific investigations aiming for a better understanding of the complex prehistoric events that are at the origins of the genetic composition of present-day European populations. Certainly, a genomic perspective, integrated with novel palaeoanthropological and archaeological findings, will provide an important contribution to the continued effort in uncovering the human past.

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