ANTHROPOLOGICAL GENETICS

ANCIENT DNA

Presentation number: AG 01 Abstract number: ABS-49-ISABS-2024

DIMINISHED PREVALENCE OF THE CURRENTLY PREDOMINANT CROATIAN Y HAPLOGROUP IN THE EARLY MIDDLE AGES

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Y haplogroup I2a2-M423 is a Paleolithic European marker most abundant in modern-day Bosnia and Herzegovina (over 55%), Croatia, and Serbia (around 40%). It was initially suggested that the halogroup survived the last Ice Age in refugial areas in Southeastern Europe and the Balkan Peninsula. However, more recent research introduced the possibility that it might have arrived in this region during later medieval migrations associated with the influx of Slavs and other populations and that it became dominant more recently. The objective of this study was to test if the I2a2 haplogroup is present at the early medieval Jagodnjak site (6-9th century CE), located in present-day Croatia and to compare obtained results with a more extensive, publicly available aDNA database (samples spanning from 1st to 15th century CE). Laboratory work was performed at the Department of Evolutionary Anthropology, University of Vienna. Shotgun sequencing of 9 male individuals was performed on Illumina NextSeq500 platform, and Yleaf program was used to infer Y hgs based on ISOGG nomenclature. Paternal genetic diversity in the sample was high, but the sample lacked I2a2 individuals. Half of males were assigned to the typical, local E1b1 hg and second most prevalent hg was R1a1a (33%), previously associated with Slavic migrations in Southeastern Europe. The more extensive database encompassed only 3.9% I2a2 carriers, from which only one was dated to a period preceding 6th century CE, indicating almost complete absence of I2a2 individuals in older historic periods. This preliminary finding provides an additional overview on the history of the Croatian territory, which should be more comprehensively analyzed by examining additional archaeological skeletal remains (possibly from older historical periods) as imperative to make evidence- based conclusions on the I2a2 origin and diversity in this region.

Keywords: aDNA, Y chromosome, I2a2 haplogroup, Middle Ages

Presentation number: AG 02 Abstract number: ABS-39-ISABS-2024

ANCIENT DNA PATERNITY TESTING AND KINSHIP ANALYSIS REVEALED FATHER – SON RELATIONSHIP IN TWO CASES OF CONGENITAL BUTTERFLY VERTEBRAE ANOMALY IN THE LATE AVAR POPULATION

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Recent methodological advances in sequencing technologies and ancient DNA extraction protocols from skeletal remains opened a window for direct insight into the past of human populations. Here we present an archeogenetic analysis of 43 individuals from the late Avar population (8th century) in the Šarengrad Klopare site from Eastern Croatia. The aim was to compare bioarcheological analysis of human remains with genetic findings to gain deeper insight into their social organization, health and genetic history. Extraction of aDNA and library preparation were performed in dedicated clean aDNA facilities. Sequencing was performed on Illumina NextSeq500 platform. Haplogrep2 was used to assign mtDNA haplogroups, and Yleaf program to infer Y haplogroups. Kinship analysis up to the 4th degree of ancestry was estimated using the READ and TKGWV2 methods. Bioanthropological examination of this population showed an absence of intentional perimortem injuries. At the same time, kinship analysis revealed the presence of at least four families with up to four degrees of relatedness, both pointing to the continuation of living on this territory for a more extended period. Some burials suggested social stratification, particularly in several traditional horseman graves. One was an adult male (grave 25) with an interesting finding of a congenital anomaly called butterfly vertebra. The same anomaly was found in a young adolescent (grave 21). In both cases, the malformation was located on the 4th lumbar vertebrae. Kinship and Y chromosome analysis confirmed that these two individuals were father and son, belonging to the southern European Y haplogroup E1b1b1a1b1a. Their mitogenome analysis also revealed European haplogroups, T1a and H46, all pointing to mixing with local communities over a longer period. This finding emphasizes the importance of using ancient DNA analysis to shed light on ancient populations' genetic ancestry and health with little or no written historical record.

Keywords: ancient DNA, kinship analysis, Y chromosome, butterfly vertebrae, Avars

Presentation number: AG 03 Abstract number: ABS-92-ISABS-2024

THE ECHOES OF ANCIENT DNA: UNCOVERING CONNECTIONS TO MODERN SOCIETIES AND CULTURAL DESCENDANTS

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The goal of this study is to examine the relationship between ancient human remains, their genetic legacy, and the modern social groups claiming ancestral rights to these relics. It aims to trace genetic lineages across millennia, amidst substantial cultural shifts, to determine connections between ancient remains and contemporary (indigenous) communities. The study involves the critical examination of bioarchaeological research of ancient human remains and their DNA with a particular focus on the legal and ethical dimensions of such work. The research reveals that claims of autochthonous groups to ancient skeletal remains represent a contentious issue that intersects science, history, religion, ethics, and identity. It underscores the need for a balanced approach that respects both scientific inquiry and cultural heritage. The ethical and legal dimensions of researching ancient remains and DNA are scrutinized, emphasizing the sensitive nature of such research and the potential for misuse. Research on ancient DNA and ancient human remains underscores the need for a balanced approach that respects both scientific inquiry and cultural heritage, paving the way for more responsible and inclusive practices in exploring of our shared human past.

Keywords: human skeletal remains, bioarchaeology, ancient DNA, genealogical descendants, cultural descendants

MIGRATION HISTORY

Presentation number: AG 04 Abstract number: ABS-105-ISABS-2024

INVESTIGATING RUNS OF HOMOZYGOSITY IN NEAR OCEANIC POPULATIONS

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Populations from Oceania are underrepresented in genomic studies and many questions about their population-specific variation and demographic history remain unanswered. To fill this gap, we sequenced 177 new Near Oceanic genomes from 12 geographically diverse populations and used this data to examine population-specific patterns in runs of homozygosity (ROH) and characterize their physical distribution along the genome to identify ROH hot and cold spots. While ROH are found in all human populations, variation in the length distribution of ROH and patterns of non-uniform ROH distribution across the genome can provide insight on the demographic and evolutionary processes that result in differences in genome-wide genetic diversity among populations. Our results show that lengths and total sum of ROH segments differed greatly between Near Oceanic populations, suggesting a complex demographic history of population structure and corroborating previous findings of population bottlenecks within Near Oceania.

Keywords: Oceania, runs of homozygosity, human population genetics, genomics, population history

Presentation number: AG 05 Abstract number: ABS-145-ISABS-2024

MOLECULAR ANTHROPOLOGICAL STUDY OF THE INDIAN OCEAN LITTORAL, FROM AUSTRO ASIATIC TO AUSTRONESIAN DISPERSAL, ACROSS THE PACIFIC

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The Indian Ocean region is a critical route/corridor/barrier for the migration and settlement of people whose social, linguistic, and genetic ancestry is of interest to anthropologists worldwide. Varying from the ‘little tradition’ of small, isolated populations to the ‘great tradition’ of global and admixed heritage, in the field of collating evidence from culture, language and anthropological genetics this paper will examine the ‘Austro Asiatic and Austronesian dispersal’ within the boundaries of the Indian ocean sporadically sprinkled across Pacific islanders. Littoral or near shore settlements may have simply act as ‘gateways’ as would have been the case with pre-historic settlements somewhere near a fresh water source or riverbank. Physical environmental changes such as those driven by drastic ecological catastrophes including severe and acute adaptations to climatic and nutritional stressors can leave signatures providing vital clues for the dispersal of humankind. During the last two decades or more, a plethora of genomic information has surfaced tracing the genetic history of an archaic hominin group from ancient DNA. In the light of ensuing works by palaeoanthropologists and evolutionary biologists, the dispersal and migration of people, genes and languages studying human history either directly through their remains or indirectly through tools, pottery or other cultural items have provided concerns over interpretation of archaeological dates. Similarly assessing the reliability of linguistic data which may favor a close correlation between genes and languages, yet at times appear to conflict with genetic evidence. From single versus multiple dispersal models’ molecular anthropologists have re-examined hypotheses about the colonization of the Pacific with different pliable approaches to understand population histories in the context of the ‘Austronesian dispersal’ across the Pacific. Is the Austro-Asiatic element be related in anyway via the south routes?

Keywords: Molecular anthropology, genetic, linguistic, archaeological, migration, dispersal, Austro- Asiatic, Austronesian, Indian Ocean, Pacific

Presentation number: AG 06 Abstract number: ABS-175-ISABS-2024

ACCURACY OF THE ANGLE OF THE GREATER SCIATIC NOTCH IN THE ESTIMATION OF SEX IN THE ALGERIAN POPULATION

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This study aims to evaluate the precision of sex estimation by the angle of the Greater Sciatic Notch (GES) on an Algerian adult sample. 123 "virtual" left iliac bones (62 men and 61 women, aged between 18 and 86 years), were included after agreement from the Scientific Council and anonymity of the sample. The bones were obtained from three-dimensional reconstructions of clinical abdominopelvic scans, carried out during 2018. The angle of the GES (the total angle) was quantified by referring to three metric landmarks. The results show that the GES angle is wider in women. The female mean was 76.6°±6.6 (62°-92°) and the male mean was 62.2°±6.8 (47°-81°). A significant metric difference was observed between the female and male sample ($p<0.001$). An overall correct sex classification rate of 87.8% was obtained by the metric method using one section point. Differences between populations emerge from the comparison of the results obtained in similar works. In conclusion, this study revealed a significant metric sexual dimorphism of GES in the Algerian population and offers specific standards that can be used in forensic cases. Other studies using larger samples will make it possible to refine the results obtained.

Keywords: forensic anthropology, sex estimation, metric method, large sciatic notch

GENETIC ADAPTATION

Presentation number: AG 07 Abstract number: ABS-78-ISABS-2024

CORTISOL LEVELS IN PREGNANT WOMEN ON ISLANDS AND MAINLAND

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The term "stress" describing a state of emotional or physical tensions and exhaustion, is commonly encountered in contemporary discourse. It is a well-established fact that periods of stress lead to dysregulation of the hypothalamic- pituitary-adrenal (HPA) axis resulting in elevated levels of serum cortisol, often referred as the “stress hormone” which plays a pivotal role in various metabolic and immunomodulatory processes. The main goal of this study was to detect and compare cortisol levels in pregnant women in relation to place of living: mainland vs islands. The underlying assumption is that differences in adaptation related with place of living may influence stress hormone levels. The research was conducted using data collected during the implementation of the "Croatian Island’s Birth Cohort Study (CRIBS)" project, aimed at elucidating factors related to metabolic syndrome, which increasingly affects a significant part of the population. The analyzed samples consisted of plasma specimens collected from healthy pregnant women residing in Split-Dalmatia County (Croatia). An analytical protocol for the determination of cortisol in human plasma by High-performance liquid chromatography (HPLC) was implemented. The study revealed that maternal cortisol levels were statistically higher in pregnant women residing on islands compared to those living on the mainland (p=0,046). Additionally, irrespective to the place of residence, in the CRIBS study, higher cortisol levels were observed in women who carried a female fetus. These differences in maternal cortisol levels show that place of residence influences stress levels, further research into the factors that induce this difference is needed.

Keywords: cortisol, stress, HPA, pregnancy, islands

Presentation number: AG 08 Abstract number: ABS-39-ISABS-2024

POLYMORPHISMS OF THE LACTASE PERSISTENCE VARIANT IN LCT GENE IN THE CROATIAN ROMA POPULATIONS

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One significant step in human adaptation that connects genetic evolution, and the evolution of dairying is the ability of some adults to digest milk. Enzyme lactase – responsible for breaking down the milk sugar lactose into glucose and galactose – declines after 5th year of age, leading to adults' lactose intolerance. However, in some human populations, lactase activity persists into adulthood. The frequency of lactase persistence (LP) variants globally varies greatly, ranging from 0% to almost 100%. Roma minority population in Croatia belong to two large groups: the Vlax/Bayash Roma, descendants of Roma who lived in captivity (mostly forced to work in mines) in Wallachia and Moravia (now Romania) from the 14th century until 1850, and the Balkan Roma, descendants of groups who traditionally lived as nomads. Since none of these lifestyles allowed for the basic care of livestock, just like the lifestyle of their ancestral population in India had not as well, these Roma groups are not expected to be under selective pressure for LP. Two genotypes associated with the LP phenotype, -13910C>T (rs4988235) and -13915C>T (rs41380347) in the lactase gene (LCT), were determined using KASP method in Vlax Roma living in Baranja and Međimurje (n =252) and in a group of Balkan Roma living in Zagreb (n=171). The results showed very low allele frequency for LP variant LCT-13910T and the absence of LCT-13915T variant in the Croatian Roma population. There were only two carriers of LCT-13910TT genotype, and T allele prevalence differed between the three Roma groups; there was significantly more T allele carriers in Međimurje (13.5%), than in Baranja (6.7%) or Zagreb Roma (4.1%) ($p<0.001$). Considering the average European frequencies of the LCT-13910T allele are as high as 63.5% (GnomAD database, n = 55,000), those found in the Croatian Roma populations are exceptionally low, indicating that Roma ancestors were not under selection pressure for the lactase persistence in adulthood.

Keywords: lactose intolerance, lactase persistence, LCT gene, rs4988235, Roma population

Presentation number: AG 09 Abstract number: ABS-89-ISABS-2024

HEFNER'S ANCESTRY-SPECIFIC TRAITS ON THE EASTERN ADRIATIC COAST POPULATIONS: FROM MIDDLE AGES TO CONTEMPORARY PERIOD

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To compare frequencies of Hefner's morphological cranial traits for ancestry estimation in the medieval and modern population sample from the Eastern Adriatic coast. The sample consisted of 140 medieval dry skulls from two archaeological sites, Velim Velištak and Radašinovci (7th – 9th century), housed in the Croatian Academy of Sciences and Arts and 195 multi-sliced computed tomography (MSCT) scans collected from the University Hospital Center in Split. Dry skulls and MSCT scans were scored for nasal overgrowth (absent or present), anterior inferior nasal spine (score 1 – 3), post bregmatic depression (absent or present), interorbital breadth (score 1 – 3), nasal breadth (score 1 – 3), shape of the zygomaxillary suture (score 1 – 3), and malar tubercle (score 1 – 3). We calculated and compared frequencies between samples using a Chi-squared test with a statistical significance level set at $P \leq 0.05$. Frequencies of all traits showed statistically significant differences ($P < 0.05$), except for the nasal breadth ($^2 = 0.308$, $P = 0.857$). The study results demonstrated significant differences between populations from the Eastern Adriatic coast that could arise from the possible secular changes and population dynamics through the millennium timespan. However, further studies should examine the genetic admixtures through the centuries, as well as expand the sample to in-between periods to detect the time frame when these changes first occurred.

Keywords: Hefner's morphological traits, population differences, early medieval period, modern population, Eastern Adriatic coast