Abstracts of poster presentations

Anthropological genetics

Ancient dna

Presentation number: AG 1

MULTIDIMENSIONAL APPROACH IN THE APPLICATION OF ETHICAL STANDARDS IN ANCIENT DNA RESEARCH

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Goal was to determine the extent of (normative) uniformity on international level regarding application of ethical standards in contemporary research of ancient DNA (aDNA), and to assess to which degree this uniformity contributes to the easier implementation of scientific research on aDNA. We conducted a literature scoping review of the online databases Scopus, Web of Science as well as gray literature. The combination of keywords: ethics, ancient DNA, aDNA without any limitations was used. Afterwards the publications were analyzed in reference manager (duplicates and non-relevant literature excluded). Total of 94 publications were found. After the analysis of the results 16 of the publications were excluded as duplicates and 59 as non-relevant publications. The remaining 19 publications were analyzed in detail. The analysis indicated that a majority of relevant publications on the aDNA ethical standards were published in the last 5 years. 12 publications discuss ethical dilemmas on aDNA research regardless of specific population, while 7 publications discuss ethical dilemmas considering specific populations (4 of them deal with the topic of native populations of Americas, 2 of them with native populations of Africa, and 1 of them with remains from ancient Egypt). Information extracted from relevant publications indicate the importance of DNA analysis in the array of anthropological techniques but also emphasize the significance of conducting the research methods on aDNA in accordance with ethical standards. The research community faces the challenge of defining global and/or supranational ethical guidelines, which would define minimum of ethical standards acceptable to different countries, cultures, religions, social groups, and other stakeholders. Recent history indicates to the possibility of misuse of DNA research, in promoting theories aimed at the dominance of certain social groups over others. Results of our research emphasize the importance of unification of ethical principles, application of uniform ethical standards and promotion of a healthy balance between scientific interest in aDNA and respect for cultural, social, religious, and other differences among different stakeholders.

Key words: ancient DNA, ethical guidelines, stakeholders, cultural differences, religion

Presentation number: AG 2

EVALUATION OF MITOCHONDRIAL DNA OF SKELETAL REMAINS FROM PERIOD OF MEDIEVAL BOSNIA

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Mitochondrial DNA (mtDNA) haplotype analysis is a valuable tool to study human migrations and evolutionary history as well as intrapopulation and interpopulation diversity of human populations. Therefore, the aim of our study was to determine mitochondrial DNA haplotype frequencies of skeletal remains from Medieval Bosnia and compare obtained results with the results for the modern Bosnian-Herzegovinian population. Samples of twenty-six skeletal remains, excavated from different medieval necropolis in Bosnia and Herzegovina, were washed and ground to a fine powder, followed by decalcification with 0.5M EDTA. Extraction of DNA was done using optimized phenol-chlorophrom-isoamyl alcohol method with additional purification using filter columns. Sequencing of hypervariable segment I (HVSI) of the control region (CR) as well as analysis of restriction fragment length polymorphism (RFLP) of isolated mtDNAs was used for mitochondrial DNA haplogroup prediction. Sequences were aligned and analyzed in Bioedit 7.2 software, while for haplogroup prediction Mitomaster was used. This tool utilizes Haplogrep2 with Phylotree 17. For all analyzed samples H haplogroup was predicted which is also a predominant haplogroup in modern Bosnian-Herzegovinian population (more than 50%). Among all samples, two were determined as H5 and the others as H2a sub-haplogroup. Our results indicate the prevalence of H mtDNA haplogroup among the inhabitants of Medieval Bosnia which can be considered as important information in regards to the genetic structure of medieval Bosnian population.

Keywords: mtDNA, ancient DNA, medieval Bosnia, haplogroups, RFLP, HVSI

Presentation number: AG 3

DNA ANALYSIS REVEALED KINSHIP BETWEEN PEOPLE FROM A SMALL COMMUNITY LIVING IN MEDIEVAL BOSNIA

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Skeletal remains of 11 individuals, stored in Travnik Homeland Museum, originated from Travnik municipality (localites Klisa-Guca Gora, Alihodze and Glavica-Han Bila) were sampled for DNA extraction. Well preserved teeth were grounded to a fine powder, after washing and subjected to decalcification with 0.5 M EDTA solution during seven day. Ancient DNA was extracted according to an optimized phenol-chloroform-isoamyl alcohol extraction technique, and then purified using filter columns. PowerPlex® Fusion System and Investigator® 24plex QS Kit were used for amplification of selected STR markers. Additionally, PowerPlex® Y23 System was used to generate Y-STR haplotypes for male individuals. In general, autosomal STR profiles were successfully generated for all samples. Male sex was determined in eight samples. Comparative analysis of two male aDNA profiles showed matching at 18 out of 22 analyzed autosomal STR loci. Statistical analysis confirmed siblingship with calculated kinship probability (KP) of 99.99996%. Furthermore, statistical analysis of two compared male profiles revealed probability of 97.77112% that they are in half brother-half brother, grandfather-grandchild or uncle-nephew relationship. For other samples, statistical analysis revealed probability of first cousins relationships with KP in the range of 59.48719 to 99.32699%. Additional Y-STR analysis showed that all eight male individuals share the same Y-haplotype confirming kinship through paternal line. Determined Y-haplotypes were found to belong to the J2a haplogroup.

Keywords: ancient DNA, skeletal remains, STR markers, Y-haplogroups, archaeology

MIGRATION HISTORY

Presentation number: AG 4

A NEW FINDING WITHIN THE MITOCHONDRIAL X HAPLOGROUP IN CROATIAN ISLAND ISOLATES CONFIRMED BY MASSIVELY PARALLEL SEQUENCING

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Mitochondrial haplogroup (hg) X was estimated to originate in the Near East cca 30 ka years ago. Its approximate prevalence in the general European population is 2%, with most sublineages belonging to the X2 clade. The X2 clade is far less common in Croatian insular populations according to our previous findings - 6 out of 13 detected X samples belong to a new, local variant based on specific mutations in both the control and coding region of the mitochondrial genome. The aim of this study was to fully explore mentioned 6 mitochondrial haplotypes found in islands of Rab and Cres. Complete sequencing of one Rab and one Cres mitochondrial genome was performed using both Sanger sequencing and massively parallel sequencing with the Illumina® Human mtDNA Genome assay on MiSeq FGx™ instrument. Sequenced samples indicate a novel lineage within the global mitochondrial X hg phylogeny according to both PhyloTree and Mitomap. The lineage is connected with the hg X3 by a common polymorphism G3531A in the coding region of the mitochondrial genome, but it lacks all other X3 defining mutations. Mutations specific for this lineage are T195C, C338T, A7518G, G7853A, A10113G, C10673T, C10920T, A11380G, A13614G, C13950T, A15903G, G15927A, T16136C and A16289G. Analysis of non-phylogenetic variants exposed a rare, A7518G mutation in the tRNA gene, which is predicted as possibly pathogenic by MitoTIP. Findings of different heteroplasmic mutation patterns between samples indicate a more distant kinship within the same mitochondrial lineage. Our finding indicates a recent local microdifferentiation process within hg X. Described lineage could possibly be marked as a new, island-specific X twig formed within the Croatian population. The extension of present research, including genealogical and clinical data, is needed to confirm further enrichment of existing mtDNA phylogeny and to establish possible functional manifestations of locally specific variants.

Key words: X haplogroup, mtDNA, massively parallel sequencing

Presentation number: AG 5

GENETIC SUB-STRUCTURING OF CROATIAN ISLAND POPULATIONS IN A WIDER SOUTHEAST-EUROPEAN CONTEXT- A META-ANALYSIS

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Goal was to determine the influence of island population isolation on the sub structuring of the Croatian population, and the influence of regional population groups on the sub structuring of Southeast Europe with regards to basic population genetic statistical parameters calculated using STR locus analysis. Bio-statistical analyzes were performed for a total sample of 2877 unrelated participants of both sexes from the area of Southeastern Europe was analyzed. Nine autosomal STR loci (D3S1358, vWA, FGA, TH01, TPOX, CSF1PO, D5S818, D13S317, D7S82) were analyzed using standard F-statistics and population structure analysis (program STRUCTURE). The total coefficient of genetic differentiation of Croatian subpopulations calculated by the FST method is higher at the level of the Croatian population (0.005) than at the level of Southeast Europe (0.002). In the population of Croatia, the subpopulation of the island of Vis shows the most pronounced separation, and in the population of Southeast Europe the population of Albanians from Kosovo, then the populations of Croatia, Bosnia and Herzegovina and Hungary. The established higher structure of Croatian subpopulations in relation to the populations of Southeast Europe suggests the existence of a certain degree of genetic isolation, most likely due to the influence of endogamy within rural island populations. The total genetic differentiation coefficient of Croatian subpopulations calculated by the FST method is higher at the level of the Croatian population (0.005) than at the level of Southeast Europe (0.002). The established higher structure of Croatian subpopulations in relation to Southeast Europe suggest the existence of a certain degree of genetic isolation, most likely due to the influence of endogamy within rural island populations.

Key words: STRs, genetic sub-structuring, Croatian island populations, Southeast Europe

Genetic adaptation

Presentation number: AG 6

GENETIC DIVERSITY OF MALE POPULATION IN SIX MUNICIPALITY OF THE NORTH-EASTERN MONTENEGRO

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Commercial genetic tests become more popular in recent years, especially for genetic genealogy research. The most popular marker in genetic genealogy that combines genetic data and family history is the Y chromosome. This popularity is based on its haploid character and its close association with the patrilineage and paternal inherited surname. The aim of this paper is to collect, summarize and analyze data on the genetic diversity of the male population of the six municipality at the North-eastern Montenegro, with the majority population of the Islamic religion: Berane, Petnjica, Plav, Gusinje, Rozaje and Bijelo Polje through the Y chromosome. The paper presents the results of data were obtained by testing 267 samples of DNA materials taken from the male inhabitants of six municipality at the North-eastern Montenegro, of which 205 samples of male inhabitants' Islamic denomination (group 1) and 62 samples of male inhabitants' Orthodox denomination (group 2). The results of research point there are thirteen haplogroups: R1b, I2, E, J2, I1, G2, J1, G, R1a, N, C, T1, Q1. The most common haplogroups are I2 and R1b, both identified in 23.97% of samples, followed by E (22.47%), J2 (11.61%), I1 (6.74%), G2 (3.75%), R1a (3.37%), I1 (1.12%), G (1.12%), N (0.75%), C (0.37%), T1 (0.37%) and Q1 (0.37%). Related to the ethnic denomination, the most common haplogroup for group 1 is R1b (30.24%), followed by I2 (22.44%), E (20.98%), J2 (12.68%), I1 (4.39%), G2 (4.39%), R1a (3.41%), C (0.49%), T1 (0.49%), and Q1 (0.49%), while I1, G and N are not identified. For the group 2 the most common haplogroup is I2 (29.03%), followed by E (27.42%), I1 (14.52%), J2 (8.06%), J1 (4.84%), G (4.84%), R1a (3.23%), R1b (3.23%), N (3.23%), G2 (1.61%), while C, T1 and Q1 are not identified. These results will contribute to bringing together ethnically different confessions by understanding that they are all a mixture of different genetic markers.

Key words: Haplogroup, Y chromosome, Montenegro