P07.079
Population study at fifteen Short Tandem Repeat loci in the Sarajevo (B&H Capitol) residents

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In our previous population studies of B&H human population, we used 17 STR loci included in the *PowerPlex 16® System* and *AmpFlSTR®Identifiler®*, twelve Y-chromosomal short tandem repeats loci incorporated in the *PowerPlex® Y System*, as well as 28 Y-chromosome NRY bi-allelic markers to generate Bosnian referent database. Wishing to test our database in order to obtain specific results in various DNA analysis for the local population of Bosnian Capitol - Sarajevo, we have decided to test unrelated healthy 150 individuals (situated in Sarajevo) at fifteen autosomal short tandem repeats loci. Qiagen Dnaeasy™ Tissue Kit was used for DNA extraction from buccal swabs and bloodstains and *PowerPlex 16® System* for amplification and detection. Amplification was carried out as described previously. The total volume of PCR reaction was 5µl. PCR amplifications were carried out in PE GeneAmp PCR System Thermal Cycler. Electrophoresis of the amplification products was preformed on an ABI PRISM 310 genetic analyzer (ABI, Foster City, CA). The raw data were compiled and analyzed using Genemapper® v3.2. Deviation from Hardy-Weinberg equilibrium, observed and expected heterozygosity, power of discrimination and power of exclusion were calculated. In addition, we compared obtained Sarajevo data with the data obtained from the global Bosnian and Herzegovinian population, isolated human population from the Bosnian mountain area as well with geographically closer (neighboring) European populations. The results of this study will be used as guidelines in additional improving of investigation of recent local B&H populations, both isolated and open, initiated in our previous researches.

P07.080
Study on a possible effect of four longevity candidate genes (ACE, PON1, PPAR-gamma, APOE) on human fertility

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A possible effect on fertility of four genes [angiotensin 1-converting enzyme (ACE), paraoxonase (PON1), peroxisome proliferator-activated receptor gamma (PPAR-γ), and apolipoprotein E(APOE)] previously found associated with longevity was sought in order to determine whether they have a pleiotropic action at different life ages. The study population was 151 Italian subjects whose reproductive life took place at the beginning of the demographic transition (declining fertility and longer life expectancy) and who had produced a mean number of children (3.6±2.3) such as to be still useful to detect a differential reproductive efficiency associated with different genotypes.
Of these four longevity candidate genes, only PPAR-γ and APOE appeared to have an effect on fertility, indicating their possible influence on reproductive efficiency. The PPAR-γ Pro/Ala genotype, which in a previous study (Barbieri et el. 2004) showed a positive association with longevity only in men, was found associated with a higher number of children (6.1 ± 3.3) than Pro/Pro genotype (3.3 ± 1.9, p=0.001) only in men. Compared with the other APOE alleles, the APOE*2 allele, considered as an allele favouring a longer life-span, was confirmed to be associated with the lowest fertility (p=0.03). The logistic regression analysis indicated that APOE and PPAR-γ polymorphisms act as independent determinants of reproductive efficiency. These data suggest that the APOE*2 allele may follow the model of antagonist pleiotropy, whereas the PPAR-γ Pro/Ala genotype seems to exert beneficial effects both early in life and in advanced age in a gender-specific way.

P07.081
Polymorphism of some genes in connection with age gradation

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The aim was to evaluate age dynamics of alleles and genotypes of APOE (C112R, R158C), ACE (I/D), PON1 (Q192R), PON2 (C311S), CAT (-262C/T), GPX1 (L198P) and MSRA (-402T/C) gene polymorphisms in group of 1627 Tatars in age of 1-109 years old.
Differentiation of total group on certain age groups was carried out by means of CHAID algorithm from SPSS Answer Tree (v.13.0). Genotyping was performed using PCR and PCR-RFLP. Fisher's two-tailed exact rest (Statistica v. 6.0) was used for age groups comparison.
In group 36-61 years increase of CAT *C allele frequency was observed (P=0.004). Persons in the age of 55-77 years have significantly higher GPX1*L allele frequency (P=0.016). APOE*3, ACE*D, ACE*D/*D, PON2*C, PON2*C/*C, CAT*T, CAT*C/*T, GPX1*P and GPX1*P/*P alleles and genotypes frequencies were considerably higher in senile group (P<0.05). ACE*I/*D genotype and PON1*R allele carriers were more frequent among long-livers (P=0.026 and 0.004 accordingly).
Thus, we have demonstrated diversity of APOE, ACE, PON1, PON2, CAT and GPX1 genes polymorphisms genotypes and alleles frequencies between different age groups. Possibly, the same polymorphic variant plays a protective role for an organism at its different age stages.

P07.082
Generation of lymphoblastoid cell lines from frozen whole blood

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The manufacture of a lymphoblastoid cell line from a single donor blood sample is a means of securing a permanent, expandable and renewable source of genetic and other cellular material. Once a single donor cell bank is available, study material can be widely distributed and will be available many years after the initial study for follow-on studies that may not have been originally anticipated, or which may not be possible with existing technologies. In many respects a cell bank can be regarded as a means of immortalising a very valuable study cohort and is the soundest means of underpinning a biobank and maximising its value in the long term. However, the majority of biobanks participating in organisations such as the Public Population Project in Genomics (P3G) Consortium do not currently store samples for future cell line generation.
ECACC Human Genetic Services has approximately twenty years experience in providing strategic support to genetic research throughout the UK and Europe, through the provision of a blood processing and EBV transformation and cell banking service.
In this presentation we describe the development of a new process for the generation of EBV transformed lymphoblastoid cell lines from cryopreserved aliquots of whole blood which represents a cost effective alternative to current methods involving separated peripheral blood lymphocytes.

P07.083
No significant contribution between M470V and 5T polymorphisms and cystic fibrosis phenotype in Iranian patients

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The most common CFTR polymorphism, M470V, has been shown to be relatively frequent among Iranian Cystic Fibrosis patients. Whether M470V polymorphism and 5T variant have CF causing contribution in Iranian population is not clear yet and it may increase difficulties in genetic counseling. In order to compare the frequencies of these variations, 100 CFTR alleles from normal controls and symptomatic Iranian CF patients were analyzed for the presence of 5T and M470V polymorphisms using PCR-RFLP method. The frequencies obtained for M470V and 5T variants were almost the same in the studied groups, suggesting that these two polymorphisms do not have strong indication of being a disease causing polymorphism. The variation in distribution of such common polymorphisms among very diverse Iranian population deserves more investigation with higher number of samples.

P07.084
Male infertility induced by mtDNA/Y unfavorable combination? An association study on human mitochondrial DNA

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