tion, daily urine oxyproline - 155.4 mg/day, increased urinary glycosaminoglycans up to 148 CPC U/g creat., hyperprolinemia, hyperglycinemia, hyperprolinuria, hyperhomocysteinemia - 26.6 μmol/l. Pedigree analysis showed that the pedigree is burdened by cardiovascular disease.
MTHFR G1793A/MTRR A66G polymorphism was revealed in the proband and his mother by the study of gene polymorphisms of folate cycle system.
Conclusion: A new variant of Ehlers-Danlos syndrome has been diagnosed in the mother and son with the phenotype associated with metabolism errors of mucopolysaccharides, hypermobility of joints, hepatosplenomegaly, an early common varicose disease, muscular hypotonia due to lower activity of MTHFR G1793A/MTRR A66G enzymes.

J17.20
Multidisciplinary study of Endometriosis as a common complex disorder

V. S. Baranov, *Yarmolinskaya M.I., Ivaschenko T.E.;*
Ott's Institute of Obstetrics&Gynecology, St.-Petersburg, Russian Federation.

Endometriosis (E) is a common multigenetic disorder affecting almost 10% of women of reproductive age. Comparative molecular, genetic, immunological analysis and endocrinology tests were applied in the studies of 257 women with E and in 117 women in the control. Participation of the genes responsible for steroid hormone activity, their receptors, inflammation, proliferation, cell migration, apoptosis, intercellular adhesion, angiogenesis as well as the genes regulating their activity have therefore been suggested as plausible candidates. A handful of very interesting new candidate genes involved in oncogenesis, metaplasia of endometrium cells and embryonic development of female reproductive system were identified by GWAS technology. In addition to alterations in the DNA sequence itself, differential expression levels of the candidate genes might be caused by different epigenetic modifications including methylation, heterochromatization, miRNA regulation etc. Complex genetic net of E, implies participation of epigenetic landscape of E. Origin of E could be provoked by any combination of both genetic and epigenetic risk factors with subsequent canalization of pathological processes (reverse epigenetic landscape), which become irreversible soon after it starts.

J17.21
Asthma related FCER2 variant in Roma and Hungarian populations

P. Matyas[1,2], *R. Szalai*[1,2], *L. Magyari*[1,2], *B. Duga*[1,2], *E. Kovesdi*[1,2], *Z. Banfai*[1,2], *A. Szabo*[1,2], *B. Melegh*[1,2];
[1]*Department of Medical Genetics, University of Pecs, Pecs, Hungary,* [2]*Szentagothai Research Centre, University of Pecs, Pecs, Hungary.*

Asthma is a complex respiratory disease, which can be caused by environmental factors and genetical predisposition. It is one of the most widespread diseases in the world; it has a high presence in most ethnic group. The low-affinity IgE receptor (FcεRII /CD23) encoded by the FCER2 gene (Fc fragment of IgE) plays important role in the regulation of IgE responses and inhaled corticosteroids (ICS) therapy in asthma. Corticosteroids influence FCER2 expression and FcεRII receptor function. The intronic rs28364072 polymorphism (T2206C) of FCER2 gene associated with elevated IgE levels, severe asthmatic exacerbations and decreased gene expression. The variation in the FCER2 gene contributes to variation in ICS treatment response in asthmatics. Our aim was to investigate the ethnic differences, allele and genotype frequencies of intronic variant of FCER2 in average Roma and Hungarian population. We examined 458 Roma (206 males, 252 females, mean age: 46.4±18.4) and 397 Hungarian subjects (222 males, 175 females; 37.8 mean age±12.6 years) with PCR-RFLP method. We found more than twofold increased homozygous CC genotype frequency in Hungarian group compared to Roma samples (5.8% vs. 2.8%, p<0,05). The C allele frequencies were similar in each group (24.8% in Romas and 24.6% in Hungarians). The current study demonstrated that unfavourable variants can diversely occur in Hungarian and Roma individuals. Genetic test of FCER2 variant are likely necessary to assess the outcome of asthma treatment. Homozygous 2206C allele carrier Hungarians have higher chance for insufficient response to corticosteroids compared with Roma subjects due to genotype higher risk frequency. This research was supported by TÁMOP-4.2.3-12/1/KONV-2012-0028.

J17.22
Common MEFV gene mutation profiles in Familial Mediterranean Fever patients in Canakkale Population

F. Silan[1], *N. Topaloglu*[2], *M. Urfali*[1], *C. AKURUT*[1], *M. G. K. KOCAK*[3], *D. Kankaya*[1], *E. ARI*[1], *O. Ozdemir*[1];
[1]*Department of Medical Genetics, Canakkale, Turkey,* [2]*Department of Pediatrics, Canakkale, Turkey,* [3]*Department of Nephrology, Canakkale, Turkey.*

Objective:In the current results it was aimed to find out of the current study is to determine the frequency of common MEFV gene point mutations in 741 patients who preliminary diagnosed as FMF Method:The genomic DNA was isolated by spin-colon method(Roche, Germany) from peripheral blood samples with EDTA and buccal smears. The MEFV gene profiles for the current FMF cohort were genotyped by Pyrosequencing and direct Sanger sequencing techniques for the target common point mutations. Results: Twenty-two different point mutations were identified in 363(49%) patients and no mutation was detected in 378(51%) current patients suspicious for FMF. The most frequent mutations were M694V(32.7%), E148Q(15.1%), R202Q(11.8%), M680I(9.9%), V726A(7.4%), P369S(6.3%) and K695R(4.4%) in the current FMF cohort. The M694V/E148Q mutation was the most frequent compound point mutation that detected in the current FMF cohort. The most common clinical finding was abdominal pain in the all MEFV mutation types that detected in the current mutated FMF patients. Median attack frequencies of untreated patients are ; 3.14 for M694V, 2.8 for E148Q, 2.75 for R202Q, 2.71 for P369S and 1.44 for K695R. Although attack frequencies were less than patients with M694V, all of the patients from Canakkale region with mutations E148Q, R202Q, P369S and K695R had FMF clinical diagnostic criterias. Conclussion:The current results showed that the R202Q point mutation frequency was higher than the other sub-populations that reported from different reigions of TURKEY. It was seen that the initiation of clinical symptoms were delayed in patients with R2020Q mutation when compared to the others.

J17.23
The association analysis of polymorphism the metabolism of lipids genes with bmi, waist circumference and blood lipidogramma's parameters at women before and after the Menopause

O. S. Glotov[1,2,3], *I. V. Tarkovskaya*[1,4], *M. M. Danilova*[1], *E. S. Vashukova*[1,2], *A. S. Glotov*[1,2], *V. S. Pakin*[1,2,4], *M. V. Aseev*[1,2,3], *T. E. Ivashchenko*[1,2], *A. B. Maslennikov*[1], *V. S. Baranov*[1,2];
[1]*St-Petersburg State University, St-Petersburg, Russian Federation,* [2]*FSBI «The D.O.Ott Research Institute of Obstetrics and Gynecology» NWB RAMS, Saint-Petersburg, Russian Federation,* [3]*Saint Petersburg State Establishment of Health Care City Hospital No 40, Saint-Petersburg, Russian Federation,* [4]*Genetic Laboratory, Ltd BioGlot, Saint-Petersburg, Russian Federation.*

Using the PCR-RFLP method we have studied polymorphism of 36 genes involved in lipid metabolism in 212 women residents of the North-West Region of Russia (St. Petersburg) at the age of 18 to 77. We found the association of polymorphisms in several candidate genes with body mass index, waist circumference, total cholesterol level, low density lipoprotein cholesterol level and very low density lipoprotein cholesterol level. We proposed a model for the prediction of examined parameters based on logistic regression method. Our findings confirm the possibility of primary assessment of body mass index, waist circumference, total cholesterol level, low density lipoprotein cholesterol level and very low density lipoprotein cholesterol level in women based on genetic markers. It is shown that women before and after a menopause have a contribution of genetics to determination of body mass index, waist circumference, total cholesterol level, low density lipoprotein cholesterol level and very low density lipoprotein cholesterol level is various.

J17.24
Genetic epidemiological study of hereditary disorders in Tatarstan Republic (Russia)

R. A. Zinchenko, *T. A. Vasilyeva, E. K. Ginter;*
Research Center for Medical Genetics, Moscow, Russian Federation.

Tatars - are the second sized ethnos of the Russia. The load and genetic diversity of monogenic hereditary disorders (HDs) in three major ethnographic groups of Tatars from Tatarstan Republic were analyzed (Kazan Tatars-3 Districts, Mishars-2 Districts and Teptyars-3 Districts. The size of the investigated populations was more than 270,000 inhabitants (213,000 Tatars). The total population was examined by standard protocol of medical genetic research elaborated in laboratory of genetic epidemiology, Research Centre for Medical Genetics. About 3500 HDs of OMIM could be identified by this protocol. Clinical investigations were performed by neurologists, ophthalmologists, orthopedic, otolaryngology's, dermatologists, pediatricians and clinical geneticists, focused on diagnostic of HDs. Genetic diversity of HDs in the investigated population consisted of 256 disorders (1597 affected): 135 AD, 97 AR and 24 X-linked recessive. Genetic differentiation of load of Mendelian HDs between populations of different ethnographic groups was found. The average prevalence rates were 1:172 persons in Kazan Tatars; 1:120 persons in Mishars and 1:150 persons in Teptyars. Variation of prevalence of all HDs in districts was from 1:350 persons to 1:85 persons. Significant differences in the load and diversity of HDs was found between groups "Kazan Tatars- Mishars" and "Teptyares - Mishars".