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# CLINICAL GENETICS

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**Abstracts of the 9th National Medical Genetics Congress of Turkish Medical Genetics Society with International Participation**
**December 1–5, 2010**
**Istanbul, Turkey**

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# Turkish Medical Genetics Society

# H – Cancer genetics

## H01 THE FREQUENCY OF FACTOR V, FACTOR II AND MTHFR MUTATIONS IN PULMONARY THROMBOEMBOLISM CASES IN THE WESTERN BLACK SEA REGION

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**Introduction:** In this study, frequency and distribution of hereditary risk factors leading to development of venous thromboembolism (VTE) in patients with pulmonary thromboembolism (PE) were evaluated.
**Materials and Methods:** We investigated the important risk factors for VTE in total 46 patients (58 ± 18.8), referred to our clinic with diagnosis of PE by Real Time PCR. We also evaluated distribution of inherited thrombophilia mutations and the family history of VTE.
**Results:** We determined the rate of FVL, FH1299R, MTHFR (C677T and A1298C) mutations in our patients as 32.6%, 2.2%, 67.4% and 63%, respectively. FIIG20210A mutation could not be determined. Twenty-six percent of the patients had only one mutation, while 71.7% of them had two or more mutations. In 77% of individuals with family history, there were two or more mutations. Sixty-three percent patients had at least one of MTHFR gene mutations alone. In 34% of the patients, a combination of at least one of MTHFR gene mutations with either FVL or FVH1299R mutation was available. In 21.7% patients, the history of VTE was present in different body part. Inherited thrombophilia with neoplasia was present in 8.7% of patients.
**Conclusion:** In patients with PE, rate of FVL, and mutations (C677T and A1298C) of MTHFR gene was higher than the previous limited data for Turkey. Determination of the relationship between VTE and inherited abnormalities will be useful for society health and familial risk relations, correctly, in addition to diagnosis and following of patients.

## H02 MALDI-TOF MS BASED GENOTYPING OF SINGLE-NUCLEOTIDE VARIATIONS PREDISPOSING TO CARDIOVASCULAR DISEASES: PREVALANCE IN TURKEY

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**Introduction:** Cardiovascular disease (CVD) is an emerging public health problem in the worldwide. Numerous polymorphisms have been identified, directly and indirectly involved in CVD etiology. These genetic factors have an affect on blood pressure regulation, blood coagulation, homocysteine and lipid metabolisms, which all leading to cardiovascular problem. Here, we evaluated the distribution of some allelic variants predisposing to CVDs. Participants were healthy individuals (n = 500). Variants involving in lipid metabolisms, APOC3 **c.3175C > G**, LPL **c.1595C > G**, **CEPT c.279G > A**, blood coagulation, factor V **1691G > A** (Leiden), factor II (prothrombin) **20210G > A**, and blood pressure regulation eNOS **c.894G > T**, polymorphisms influencing single carbon metabolism MTHFR **c.677C > T**, MTHFR **c.1298A > C**, **MTRR c.66A > G**, **MTR c.2756A > G** were genotyped using MALDI-TOF based mass spectrometry. Polymorphic sites were analyzed in according to manufacturer's protocol of Sequenom hME platform. The genotype and allele frequencies were calculated and their distributions were compared with those reported for other regions. Deviation from the Hardy–Weinberg equilibrium was not observed. Genetic and non-genetic factors play a great role in the occurrence of the complex disease. Identification of individual's genetic predisposition and proper regulation of the lifestyle in accordance with the requirements of genetic background is important to prevent disease and detect them early. Better knowledge of a common genetic structure of population may prompt targeted preventative healthcare strategies.
**Keywords:** CVD susceptibility, genetic factors predisposing CVD.

## H03 EFFECTS OF FTO (FAT MASS AND OBESITY ASSOCIATED) GENE POLYMORPHISMS ON TURKISH ADULT RISK FACTOR (TARF) STUDY POPULATION

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**Introduction:** The aim of this study was to determine the associations between the FTO rs9939609T > A and rs1421085T > C polymorphisms and the anthromorphic/biochemical variables of human metabolism. FTO is located on chromosome 16q12.2 region, was originally described in a mouse model and may play a role in lipid tissue metabolism. FTO polymorphisms have been found to be associated with diabetes type 2 and obesity in previous studies.
**Methods:** Turkish adult risk factor (TARF) is an epidemiologic follow-up study that examines the mortality and morbidity of heart diseases and their relationship with risk factors in Turkey. We genotyped two single nucleotide polymorphisms (FTO rs9939609 and rs1421085) in TARF study population. From EDTA whole blood, genomic DNA was isolated from leukocytes using QIAmpR DNA Maxi KIT. Genotyping was performed using the ABI prism 7900HT Sequence Detection System for both polymerase chain reaction and allelic discrimination with TaqMan technology, Windows SPSS was used for statistical analyses.
**Results:** Study subjects (n = 1997 and 2006) were genotyped for the rs9939609 and rs1421085 polymorphisms. Genotype frequencies of rs9939609T > A were 0.36(TT), 0.48(TA) and 0.16(AA). Genotype frequencies of rs1421085T > C were

 