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B – Data Analysis and Bioinformatics in Medical Genetics

B01 ALLELIC FREQUENCIES OF GENETIC VARIANTS ASSOCIATED WITH BONE MARROW DENSITY, IN TURKISH POPULATION

[1,2]S Karaca, [2]T Cesuroglu, [2]A Kaymaz [1]*GENAR Institute for Public Health and Genomics Research, ANKARA;* [2]*Aksaray University Sc.H. Aksaray*

Introduction: Osteoporosis is a complex disease, which prevalence markedly increases with aging. The disease characterized by increased risk of bone fracture and reduction in skeletal mass. Population based studies indicates contribution of genetic traits to disease, besides of hormonal, nutritional and lifestyle factors. In this study, we investigated single nucleotide polymorphisms, to access allelic frequency of genetic variation associated with osteoporosis.
Materials and methods: Genotyping were carried out in a large sample group (n = 500), using MALDI_TOF based mass spectrometry. Genes, significantly associated with differences in BMD and/or fracture risk in multiple replication studies were included and sequence variants, IL-6_rs**1800795,** TNF-alfa_rs**1800629**, IL-6_rs**1800796**, VDR_rs**1544410**, Col1A1_rs**1800012**, VDR_rs**2228570**, VDR_rs**731236**, were analyzed according to manufacturer's protocol of Sequenom hME platform.
Results: Genotype and allele frequencies were calculated and compared with other populations. Genotyping results were consistent with the Hardy-Weinberg equation. Some genotypes, causing susceptibility to osteoporosis, were found to be frequent in the screened group.
Conclusion: The application of genomics tools and concepts on an individual level may provide more useful and person-specific knowledge for preventing disease. To determine the prevalence of genetic factors in the population may contribute to the development of more precisely and safely healthcare systems that target individuals rather than general public.
Key words: Genetics of complex diseases, susceptibility to osteoporosis, genetic factors and BMD

B02 MULTIFACTOR DIMENSIONALITY REDUCTION (MDR) ANALYSIS FOR GENETIC MODELING OF COMPLEX DISEASES

[1]D Agirbasli, [2]SM Williams, [3]M Agirbasli [1]*Department of Medical Biology, Acibadem University, Istanbul, Turkey;* [2]*Department of Molecular Physiology and Biophysics and the Center for Human Genetics Research, Vanderbilt University Medical Center, Nashville, TN, USA;* [3]*Department of Cardiology, Marmara University Medical Center, Istanbul, Turkey*

Introduction: The nature of coronary artery disease (CAD) as a complex trait involves multiple genetic and environmental interactions. The objective of our study is to predict a genetic model for CAD using MDR method; a non-parametric data-mining approach.
Materials and Methods: Early onset CAD patients (n = 102), (n = 90) and controls (n = 102), (n = 90) respectively for two separate studies were included. Interactions among 5,10 MTHFR C677T, PAI-1 4G/5G, eNOS 3-27bp repeat polymorphisms for the first study and 5,10 MTHFR C677T, PAI-1 4G/5G, eNOS 3-27bp repeat, ACE-I 287bp Alu repeat, PON1 Gln192Arg polymorphism for the second study were evaluated. MDR analysis was performed to identify a model of CAD based on genetic and conventional risk factors in both studies. Statistical significance of MDR was determined using permutation test and was accepted at p value less than 0.05.
Results: The best model of CAD was the two-locus model with two genes, but it failed to reach a statistical significance (p = 0.24) in the first study. However in the second study, MDR analysis detected a significant model with four genes (prediction success approximately 61%, p = 0.03). When conventional risk factors were included, a different model with similar prediction was achieved with three genes.
Conclusion: MDR analysis can provide complex trait models at much smaller sample sized compared with single locus association studies. Our results suggest although it is difficult to characterize high order epistasis among genes and environmental factors, it is a more powerful approach to study complex traits as CAD. Future studies should focus on making inferences about biological epistasis from statistical epistasis.

B04 THE ADAPTATION OF DENSITOMETRIC ANALYSIS TO DIFFERENTIAL DISPLAY GELS OFFERS WELL-MATCHED RESULTS WITH MICROARRAY

[1]Y Z Igci, [2]B Cengiz, [1]S Oztuzcu, [1]M Igci, [1]A Arslan, [1]B Gogebakan, [1]Z Eslik, [1]EA Cakmak, [3]A T Demiryurek [1]*Department of Medical Biology, University of Gaziantep, Faculty of Medicine, Gaziantep, Turkey;* [2]*Department of Physiology, University of Gaziantep, Faculty of Medicine, Gaziantep, Turkey;* [3]*Pharmacology, University of Gaziantep, Faculty of Medicine, Gaziantep, Turkey*

Differential Display (DD), also known as DD-PCR or DDRT-PCR, is one of the major tools in interpreting gene expression patterns. With its simplicity, DD methodology also offers reproducibility, comparison of all mRNA species in the cell populations of interest, and isolation of corresponding cDNA. A number of different protocols have been evolved from basic DD starting since the day it was invented. Our approach basically relies on quantification of the DD results, thus making them calculable. In our ongoing study, which is actually on papillary thyroid carcinomas (PTC), we have taken advantage of the idea that measuring the intensity of gels via software which is also widely used in western blot gels. The gel images were first adjusted for analysis using GIMP (an open-source image editing software) and gel bands were measured using ImageJ (a public domain software developed by National Institutes of Health). Using the integrated density (ID) values that calculated by ImageJ software, we found a number of gene expression differences between groups (Student-Newman-Keuls test). One of the genes which was previously associated with PTC by others using microarray was ZFP36L2 (TIS11D) gene. Other studies found 2.2-fold decrease and 2.5-fold increase in 2 different groups of samples. In our study, quantification of gel bands showed that our data regarding ZFP36L2 gene were very similar to microarray results. Despite the speculations over DD because of its high false-negative rates, our data demonstrated that DD can give reliable results as microarray.