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ABSTRACT

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E-P01 Reproductive Genetics/Prenatal Genetics

E-P01.02
Prenatal diagnosis in a case of 8p inverted duplication deletion syndrome

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Introduction: 8p inverted duplication deletion syndrome is a rare chromosomal anomaly characterized by mild to severe intellectual deficit, severe developmental delay, hypotonia, agenesis of the corpus callosum and minor facial anomalies such as prominent forehead, temporal baldness, anteverted nostrils and eversion of the lower lip. Materials and Method: A prenatal case was investigated due to high risk for Down syndrome in first trimester biochemichal screening and advanced maternal age (36) by amniocentesis and fetal ultrasonography (USG). Results: Amniocentesis was performed at 16th weeks of gestation. The result of the QF-PCR analysis for rapid aneuploidy screening was normal, but add (8p) was determined by conventional karyotyping. The parental karyotypes had no chromosomal anomaly. In the subtelomeric FISH study of the fetal sample, there was a subtelomeric deletion in the 8p region. There was no fetal structural anomaly in level II fetal USG and fetal echocardiography. The molecular karyotyping revealed a gain in 8p11.22-p23.1 region with a size of 27.2 Mb containing 122 OMIM gene and a loss in 8p23.1-p23.3 region with a size of 6.8 Mb containing 15 OMIM gene. The findings were correlated with 8p inverted duplication deletion syndrome. Conclusion: Our study emphasizes the importance of using additional molecular cytogenetic methods in clinical follow-up of complex rearrangements in the prenatal cases. In unstable chromosomal rearrangements, fetal USG scans are not sufficient to detect fetal anomalies. However, molecular karyotyping is gaining importance in order to evaluate unbalanced chromosomal anomalies, to predict fetal prognosis and to give appropriate genetic counseling.

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E-P01.03
A Case of Fertile Man With Lack of AmelogeninY Locus

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E-P05.09
Functional evaluation of the interaction between cigarette smoking and the coronary artery disease risk gene *ADAMTS-7* in *Drosophila melanogaster*

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Coronary artery disease (CAD) is a complex disease resulting from an interplay of lifestyle and genetic causes whereby cigarette smoking has been shown to be one of the strongest lifestyle risk factors. Previously, CAD genome-wide association studies (GWAS) have proven that gene-smoking interactions are partly mediated by ***ADAMTS-7*, a CAD-risk gene**. Particularly, the allelic variation at **rs7178051** that **associates with reduced *ADAMTS-7* expression conferred stronger CAD protection in "never-smokers" compared to "ever-smokers"**. The loss of CAD protection in smokers is likely due to the induction of ADAMTS-7 expression in the vasculature by cigarette smoking. **ADAMTS-7 is a member of the "A disintegrin and metalloproteinase with thrombospondin motifs" (ADAMTS) family**, which plays a crucial role in **neointima formation and vessel stenosis**. Thus, a detailed understanding of the *ADAMTS-7* CAD risk gene is of significant importance. We studied previously **smoking exposure in *Drosophila***. Interestingly, our RNA-seq analysis results revealed high expression of the ADAMTS-7 homolog in the tracheas of smoke-exposed flies, indicating that it might play an important role in this gene-smoking interaction. **The fruit fly consists of a primitive vascular system in comparison to other invertebrate models and the molecular mechanisms that regulate the formation of the tracheal tube seem to be similar to those involved shaping the vascular tube in mammals**. Therefore our main objective is to unravel the **physiological role and function of *ADAMTS-7* in *Drosophila melanogaster*** and specifically to understand the interconnection between cigarette-smoking and the CAD-risk variants at the *ADAMTS-7* risk locus observed in humans.

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E-P05.10
Distribution of Common TP53 and MDM2 Variants Among Down Syndrome Patients with Cardiac Abnormalities

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Introduction: Cardiac abnormalities are common in people with Down syndrome (DS), Trisomy 21, and are mostly responsible for DS morbidity and mortality. The genetic mechanism of cardiac involvement has not fully understood but genes located in both chromosome 21 and other chromosomal regions are continuing to be investigated. To understand whether TP53 gene, which is involved in cardiac deve]opment, has a role in clinical presentation of DS, nucleotide variants associated with TP53 functions were investigated in this study. TP53 gene codon 72 Arg > Pro variant is commonly associated with TP53 protein functions. The MDM2 protein, which involved in TP53 protein degradation, is also reported to be regulated by the nucleotide variant within its first intron. In this study, allele distributions of these variants were assessed in DS patients with cardiac abnormalities.

Materials and Methods: DS patients with or without cardiac involvement and control subjects without cardiac abnormalities were included in this study. TP53 gene p. Arg72Pro (c.C215G) variant and MDM2 gene c.14 + 309 T>G variant was determined with melting analysis of hybridization probes.

Results: The Pro allele frequency of TP53 gene was about 0.58 in controls and DS (-) cardiac abnormalities. However, DS patients with cardiac abnormalities were having high frequency (0.66) of Pro allele. For MDM2 gene, controls and DS (+) cardiac anomalies were displayed similar frequency, but G allele was high (0.60) among DS (-) cardiac abnormalities.

Conclusion: The results are implying that the nucleotide variants regulating TP53 protein activities may be involved in DS clinical presentation associated with cardiac abnormalities.

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