P-089 - Determining and Classification of Genetics Variants Associated with Juvenile Arrythmia

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Detection etiology of disturbances in normal heart rhythm including Long QT, Short QT, Brugada Syndrome, Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT) and Arrhythmogenic Right Ventricular Tachycardia (ARVT) is important for specific treatment. Examination of genetic mutations observed and large data obtained with next generation sequencing (NGS) technics, using bioinformatics tools greatly contribute to the ability to establish diagnoses. The aim of this study is to compare the results of clinic databases such as ACMG, HGMD and ClinVar which are used for analyzing datas obtained with NGS. For this aim, a total of 72 individuals, consisting of 42 male individuals and 30 female individuals who are at or below 16 years old, requesting cardiac arrythmia tests in Istanbul Medipol University's Genetic Diagnostics Center were included in the study. As a result, the HGMD, ClinVar and ACMG databases complement each other. The results of this study might be subject to variances in patient clinics, physician's level of experience and the fact that current articles are not yet evaluated using databases. We hope that gradual accumulation of the laboratory's own database results will increase laboratory reliability.

KEYWORDS: Arrythmia, ion channels, next generation sequencing

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ABSTRACT BOOK

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