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Copy Number Variants in Children with Non-Syndromic Congenital Heart Diseases from Mexico

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Abstract : Congenital heart diseases (CHD) are the most common congenital abnormalities. These conditions can occur as both an element of distinct chromosomal malformation syndromes or as non-syndromic forms. Their etiology is not fully understood. Genetic variants such copy number variants have been associated with CHD. The aim of our study was to analyze these genomic variants in peripheral blood from Mexican children diagnosed with non-syndromic CHD. We included 16 children with atrial and ventricular septal defects and 5 healthy subjects without heart malformations as controls. To exclude the most common heart disease-associated syndrome alteration, we performed a fluorescence in situ hybridization test to identify the 22q11.2, responsible for congenital heart abnormalities associated with Di-George Syndrome. Then, a microarray based comparative genomic hybridization was used to identify global copy number variants. The identification of copy number variants resulted from the comparison and analysis between our results and data from main genetic variation databases. We identified copy number variants gain in three chromosomes regions from pediatric patients, 4q13.2 (31.25%), 9q34.3 (25%) and 20q13.33 (50%), where several genes associated with cellular, biosynthetic, and metabolic processes are located, UGT2B15, UGT2B17, SNAPC4, SDCCAG3, PMPCA, INPP6E, C9orf163, NOTCH1, C20orf166, and SLCO4A1. In addition, after a hierarchical cluster analysis based on the fluorescence intensity ratios from the comparative genomic hybridization, two congenital heart disease groups were generated corresponding to children with atrial or ventricular septal defects. Further analysis with a larger sample size is needed to corroborate these copy number variants as possible biomarkers to differentiate between heart abnormalities. Interestingly, the 20q13.33 gain was present in 50% of children with these CHD which could suggest that alterations in both coding and non-coding elements within this chromosomal region may play an important role in distinct heart conditions.

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