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CFC1 gene mutation in tetralogy of fallot and dextro-transposition of great arteries in Pakistani population

Afsheen Arif[1], Sitwat Zehra[1], Najma Patel[2] and Abid Azhar[1]
[1]University of Karachi, Pakistan
[2]National Institute of Cardiovascular Diseases, Pakistan

Congenital heart diseases (CHDs) are the most common of all birth defects and one of the leading causes of mortality in the first year of life. CFC1 gene is a cell signaling protein and it is a co-receptor in nodal signaling pathway and involve in right and left axis determination during gastrulation. This is a case control study, recruited 175 non syndromic patients and 140 controls, healthy unrelated individuals. The study after formal approval includes patients from various pediatric cardiology centers in three years. A detailed family history was taken to elucidate the genetic and environmental factors. Pediatric cardiologist confirmed the diagnosis on the basis of all standard testing like chest X-ray, CBC, ECG, ECHO, heart murmur, cardiac catherization reports etc. DNA extraction and sequencing was done and data was interpreted by multiple sequence alignment software. Statistical data was done by SPSS 17.0. The mean age for controls was 3.14±1.82 years for TOF; 2.97±1.21 and for DTGA patients 1.84±2.26 years. TOF and DTGA were frequent in males. The study demonstrates frequency of this disease with its variation in Pakistani population. Consanguinity affects the rate of CHDs as it is 62% in patients and 25% in controls. Two novel mutations were found in CFC 1 gene. The study reveals frequency and prevalence for TOF and DTGA, their variation and association with other cardiac defects. CFC1 and its mutations may play a key role in cardiac malformation.

afsheenarif2000@yahoo.com

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