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Chromosomal Anomalies and Congenital Heart Disease in Mysore, South India

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ABSTRACT Congenital heart disease (CHD) is the most common form of human birth defects accounting for about 30% of the total anomalies. The prevalence of CHD worldwide is found to range between 1.0 – 150/ 1000 live births. The causes for CHD can be categorized into three major groups such as, chromosomal (0.4 - 26.8%), single gene disorders (10-15%) and multiple factors (85-90%). Here we report the association of chromosomal variations with CHD in Mysore. A total of 192 confirmed CHD cases were considered for the present study whose age ranged from 1 day to 23 years. After written consent was obtained from the family members, 136 CHD patients were subjected for conventional cytogenetic studies and some of them for FISH analysis. Of these, 18 patients were with numerical abnormalities, 3 patients with structural abnormalities, one patient with both numerical and structural abnormalities and remaining 114 patients with normal chromosomes. Thus, the present findings are the maiden report from Mysore, which have contributed richly towards the association of chromosomal anomalies with CHD and pointing out chromosome 9 a possible killer chromosome for the cause of CHD.