

Association Between Pericentric Inversion in Chromosome 9 and Congenital Heart Defects

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ABSTRACT Congenital heart disease (CHD) is the leading cause of mortality in the first year of life. Prevalence of CHD worldwide is found to range from 1.0 to 50.89 per 1000 livebirths including India. The association of these defects with chromosomal anomalies varies between 4 to 12%. In the present investigation, we report two different cases of pericentric inversion of chromosome 9 [inv(9)(p11-q13)], associated with Total Anomalous Pulmonary Venous Connection (TAPVC) and Tetralogy of Fallot (TOF). In one of the cases (TOF), the mother had similar inversion without CHD. We predict here that, the genes responsible for the normal heart development could be present on chromosome 9 around p11-q13 region, which might have been defective during the process of inversion and thereby resulted in CHD. To our knowledge, this is the maiden report of association of inversion with CHD from South India.