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Association of genetic syndromes and congenital heart disease and correlating with their anatomical complexity

K Sivakumar

Department of Paediatric Cardiology, Institute of Cardio Vascular Diseases, The Madras Medical Mission, Chennai, India

Correspondence: K Sivakumar, Department of Paediatric Cardiology, Institute of Cardio Vascular Diseases, The Madras Medical Mission, Chennai 600004, India, Email drkumarsiva@hotmail.com

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Congenital heart disease (CHD) is a multifactorial disorder associated with both genetic and environmental influences. Approximately 30% of CHD is thought to be related to genetic syndromes accompanied by extra-cardiac anomalies, including abnormal facial features, or in association with limb anomalies, other organ malformations, developmental abnormalities, or growth abnormalities. A number of genetic tests namely include cytogenetic studies, fluorescence in-situ hybridization (FISH), and DNA mutation analysis can assist the clinician to confirm genetic alterations in the child with CHD.

Our aim is to determine the frequency of children with genetic syndrome and CHD in a hospital based population. To determine the complexity in anatomy of cardiac defects and post operative course, outcome of patients with CHD associated with chromosomal abnormalities and other genetic abnormalities.

Genetic testing was performed on 147 cases based on phenotype. Forty eight karyotype (33%), 96 (65%) FISH and 3 (2%) DNA mutation analysis was performed. Out of 48 karyotype, chromosomal abnormalities were observed in 24 (50%) patients. 22 (91%) were numeric [17 (77%) patients with Down syndrome, 2 (9%) with Edward syndrome, 1 (5%) with variant Klinefelter syndrome and 2 (9%) with Turner syndrome) and 2 (8%) structural (1 patient with Yqh+ and 1 with add (17)p(13)]. Out of 96 FISH tested 24 (25%) were positive [2 (8%) patients of Down, 17(71%) of DiGeorge and 5 (21%) of Williams syndrome]. screening for Noonan syndrome confirmed 2 patients have mutations in PTPN11 gene and one patient negative for fragile X syndrome. Among 87 patients suspected for DG syndrome, 17 samples came positive. Tetralogy of fallot is common in 10 patients (58%), of whom 6 had poor anatomy like pulmonary atresia, hypoplastic pulmonary annulus and branch PA¨s, absent LPA etc. Five patients had left to right lesions like ASD, VSD, PDA (29%) and one had Type B interrupted arch and other has TOF with absent PV. 10/17 had stormy post operative course needed prolonged ventilation and PICU stay. 17/20(85%) KT samples and both FISH study were positive for Down syndrome. Most common CHD associated with Trisomy 21 was ASD/VSD/PDA with 9 (53%) and AVCD in 6 (35%) patients and 2 with TOF. Among the 17 patients with Down syndrome who underwent surgery 10(58%) had stormy post operative course which includes 6 patients in ASD/VSD group, needed prolonged duration of ventilation, inotropic support, antibiotic course and PICU stay. Among the 5 patients with William syndrome 3 had LVOTO lesions 1 has B/L branch PA stenosis. The CHD associated with Edward syndrome (1 DORV and 1 PDA), Noonan (HOCM, CoA, Valvar PS), Turner syndrome (Aortic stenosis).

As genetic syndromes increased the morbidity during hospital management of congenital heart defects, paediatric cardiology professionals must be aware of the implications that performing the genetic testing can bring to the diagnosis, treatment and prognosis and for genetic counseling.

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