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Research Article

Frequency of Congenital Heart Defects in Indian Children with Down Syndrome

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Abstract

Congenital Heart Diseases (CHD) are commonly associated with Down Syndrome (DS) infants. Our study aimed at determining the occurrence and pattern of CHD in association with DS among patients in Indian subjects. A total of 60 patients with clinical features suggestive of DS were recruited. Echocardiography, standard karyotype and QF-PCR (Quantitative – Flourescent PCR) studies were performed in all patients in order to confirm DS. CHDs were detected in the 50% of children with DS. The commonest type of CHD reported in Indian subjects was atrioventricular septal defect (50%). The second most type of CHD present was ventricular septal defect (26.6%). Other type of CHD included in our study was atrial septal defect, tetralogy of fallot and patent ductus arteriosus with the frequency of 10 %, 6.6 % and 6.6% respectively. Our findings showed that CHDs are common in Indian DS children. These results suggest that a routine echocardiography should be mandatory in DS patients.

Keywords: Congenital heart diseases; Echocardiography; Down syndrome; Ventricular septal defects

Introduction

Down Syndrome (DS) or trisomy 21 is a chromosomal disorder associated with a varied combination of morphological and structural birth defects. These defects include congenital mental disability, hypotonia, characteristic body features, congenital heart defects, Hirschsprung's diseases and others. The frequency and severity of these morphological and functional defects vary significantly among affected individuals. DS affects about one in 700 live births [1,2].

Congenital heart defect disorder or CHD is a common defect among newborn infants [3] which can be caused by environmental or genetic factors. About 50% of babies with DS are born with CHD, which is a much higher percentage compared to the number of children without DS who are born with CHD which is approximately 1%. The most common CHD seen in infants with DS is an Atrioventricular Septal Defect (AVSD), or AV canal. Other heart defects seen in infants with DS include Ventricular Septal Defects (VSD), Atrial Septal Defects (ASD), Tetralogy of Ffallot (TOF) and Patent Ductus Arteriosus (PDA). However, the exact etiology of CHD in DS remains poorly understood.

Few decades ago the frequency of CHD in DS was estimated to be approximately 20%, but now with the use of better diagnostic tool, the frequency estimated has increased to 50% [2,4,5]. The most common CHD in the western literature is atrioventricular septal defects.

This study was conducted to evaluate the frequency of various types of CHD in DS children from Indian origin.

Material and Methods

This hospital based descriptive study was conducted in department of Genetics, SGPGIMS, Lucknow Uttar Pradesh and department of Pediatrics, Post Graduate Institute of Medical Education and Research, PGIMER, Chandigarh. Total sixty DS children with age 0-15 years were included in this study. All patients were diagnosed based on 2- dimensional echocardiography for CHD and molecularly confirmed for DS using karyotyping in combination with QF-PCR. Exclusion criteria were race other than Indian. All the participants were of Indian the same ethnic origin and this study was approved by the Institutional Ethics Committee of Sanjay Gandhi Post Graduate Institute of Medical Sciences, SGPGI, Lucknow, and Uttar Pradesh. Presence of CHD and type of CHD were recorded as potential risk factors of DS. DS infants were as categorized under two groups: CHD and non CHD.

Results

Out of 66 60 children? With DS, 38(63%) were males and 22(36%) were females. Congenital heart defect was found in 30 out of 60 DS children (50%). Among the affected children with DS and CHD, 20(66.6%) were males and 10(33.3%) were females.

The most common form of CHD was AVSD, found in 15 (50%) out of 30 DS children followed by VSD, which is 8(26.6%) in 30. Other form was also present which includes ASD, PDA and TOF which is 3(10%), 2(6%), and 2(6%) respectively (Table 1 and Figure 1).

Discussion

Presence of high incidence of CHD in DS people is a well known fact. Based on the previous literature, the frequency of CHD in DS varies from 35-65% [11-15,3-15]. The frequency of CHD in our study was 50% and the most. Most common type of CHD found in our study was AVSD followed by VSD with the frequency of 50% and 26.6% respectively. Our study was only found consistent with the Sudanese and Lebanese where AVSD was found to be most common with the frequency of 48% and 81.7% respectively [16,17].

Our finding were found to be different from the finding in Oman , Portuguese and Libya study where ASD was found to be most

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Table 1: Shows different type of CHD found in our study in DS children.

S. No	Subjects (n/%)	Type of CHD	No.	Percentage
1.	Male – 38 (63)	Atrioventricular septal defects	15	50
2.		Ventricular Septal defects	8	26.6
3.		Atrial Septal Defect	3	10
4.	Female – 22 (36)	Patent ductus arteriosus	2	6.6
5.		Tetralogy of Fallots	2	6.6
		Total	30	100

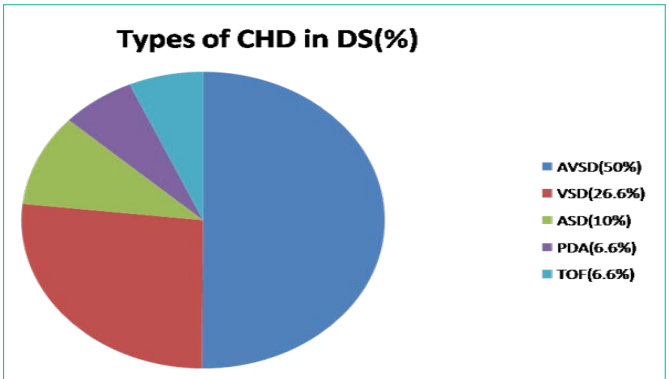

Figure 1: Shows pie chart representing the types of CHD found in DS as percentage.

common form of CHD with the frequency of 33.3%, 51.8% and 23% respectively. The second most common type of CHD in these studies' was AVSD with the frequency of 27.7%, 46.4% and 19% respectively [18-20]. Other study which reported as ASD being most common CHD type was from Korea with the frequency of 30.5% followed by VSD with the frequency of 19.3% [21].

While Some studies reported that PDA was most common form of CHD in DS was from Guatemala and Saudi Arabia with the frequency of 28.6 % and 47.8% respectively [22,23].

Recent study from Singapore reported that VSD is the most common sub type of CHD in their study followed by PDA, ASD and AVSD with the frequency of 39.2%, 34.5%, 23.4% and 15.6% respectively [23].

Based on our results, the most uncommon type of CHD found in DS children was PDA and tetralogy of Ffallot (6.6%). This frequency is slightly higher than that in Caucasian (4%) [24], and Saudi Arabian children (5.3%) [25], but relatively similar to that reported by other workers which is 6%, and 6.2% [26].

Conclusion

CHD are common in children born with trisomy 21. The commonest sub type of CHD in DS found in our study was AVSD. Neonatal screening for the risk of CHD in DS patients should be done by echocardiography [27-29]. All children born with DS should have a cardiac evaluation at birth.

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