Print ISSN: 2321-6379
Online ISSN: 2321-595X

Original Article

Profile of Congenital Heart Disease in Children with Down's Syndrome Attending an Early Intervention Center in a Teaching Hospital in South India

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Abstract

Background: Down's syndrome is the most common chromosomal disorder and the association between Down's syndrome and congenital heart disease (CHD) is well established. The spectrum of CHD in Down's syndrome shows wide variations due to genetic, socioeconomic, and geographic factors. The incidence of CHD in Down's syndrome is around 40–60% and contributes significantly to the morbidity and mortality in these children in the first 2 years of life.

Objective: The objective of the study was to study the incidence and spectrum of CHD in children with Down's syndrome attending special clinics.

Materials and Methods: It is a prospective, observational, and monocentric study done over a period of 3 years (2016–2019). Echocardiogram was done to all children at the first visit.

Results: Of the 120 children studied, 62 had CHD (52%) and ostium secundum atrial septal defect was the most common lesion (36%).

Conclusions: The incidence of CHD in Down's syndrome in this study is 52%. Hence early cardiac screening is of paramount significance in all children with Down's syndrome.

Key words: Atrial septal defect, Congenital heart disease, Down's syndrome

INTRODUCTION

The incidence of Down's syndrome caused by trisomy 21 is 1 in 733 live births and is characterized by intellectual disability, congenital anomalies, and characteristic dysmorphic facial features. In 95% of case, trisomy 21 is due to meiotic non-disjunction, 4% due to translocation, and 1% due to mosaicism. Around 4–10% of all congenital heart defects have association with Down's syndrome and 40–60% of patients with Down's syndrome have cardiac defects. Affected children are prone to congenital heart disease (CHD) such as atrioventricular septal defect (AVSD), ventricular septal defect (VSD), isolated secundum atrial septal defect (ASD), patent ductus arteriosus (PDA), and tetralogy of Fallot (TOF) and have increased risk of pulmonary hypertension. Considerable ethnic and geographic variations exist in the most common cardiac lesion seen in Down's syndrome and AVSD is the most frequently diagnosed CHD (30–40%) followed by ASD (25%) and VSD (22%).

Access this article online

www.ijss-sn.com

Month of Submission : 11-2019
Month of Peer Review : 12-2019
Month of Acceptance : 12-2019
Month of Publishing : 01-2020

The American Academy of Pediatrics recommends cardiac screening of all newborn babies with Down's syndrome and early establishment of cardiac status by 6 weeks of age is recommended widely. Failure to recognize CHD early can result in irreversible pulmonary hypertension and early deaths in these children.

Aim of the Study

The aim of the study was to find the incidence of CHD and identify the pattern of CHD in Down's syndrome.

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MATERIALS AND METHODS

Study Centre

This study was conducted at District Early Intervention Centre (DEIC) of a teaching hospital in Tamil Nadu.

Sampling

Down's syndrome children from birth to 18 years with classical phenotypic features of flat facial profile, slanted palpebral fissures, epicanthal folds, brachycephaly, single palmar crease, clinodactyly, hyperflexible joints, and hypotonia referred from neonatal intensive care unit, Rashtriya Bal Swasthya Karyakram team, and pediatric outpatient department were enrolled in the study. Diagnosis of Down's syndrome was done on clinical grounds and karyotyping was done whenever feasible. This is a prospective and observational study conducted for a period of 3 years from 2016 to 2019.

Two-dimensional echocardiogram was done for all children in the first visit by experienced cardiologists. Data were analyzed using simple descriptive statistics.

RESULTS

A total of 120 children with Down's syndrome participated in the study with 56 male and 64 female children contributing to 46.3% and 53.7%, respectively. The male-to-female ratio of the sample was 1:1.1.

The age group of the study sample varied widely from the youngest one of age 4 days to the oldest of age 14 years. Infants contributed to the majority of Down's syndrome in this study (43.3%, n = 52) due to early referral from neonatal ICUs to DEIC for early intervention. Thirty-two (26.6%) were between 1 and 5 years, 22 (18.3%) between 5 and 10 years, and 14 (11.6%) above 10 years of age [Table 1 and Figure 1].

In our study, out of 120 children, 62 had CHD while 58 had normal cardiac status. The incidence of CHD in our study was 52%. Children <1 year of age contributed to the majority with CHD 50% (n = 31). The spectrum of CHD varied from isolated CHD in 73% (n = 45) to mixed lesions in 27% (n = 17) [Figure 2].

The most common isolated heart disease was ostium secundum ASD contributing to 36% (n = 22), followed by AVSD 8.2% (n = 5), PDA 8.2% (n = 5), VSD 5% (n = 3), TOF in 3.3% (n = 2), and pulmonary stenosis in 1.6% (n = 1). Patent foramen ovale was seen in 5 children (8.2%).

The most common associated lesion was PDA with ASD 11.5% (n = 8) followed by ASD with VSD in 9.8% (n = 6), VSD with PDA in 9.8% (n = 2), and ASD + VSD + PDA in 1.6% (n = 1) [Table 2 and Figure 3]. Three infants with CHD expired during the course of the study and 7 were lost to follow-up.

DISCUSSION

One hundred and twenty Down's syndrome children comprised the study population. The male-to-female ratio of the sample population was 1:1.1 and comparable to the male-to-female ratio in the study by Morsy *et al.*[1] and Benhaourech *et al.*[2] which was 1:1.

The study by Morsy *et al.*[1] had an incidence of 58.6% and a high incidence of 63.4% and 69.5% was found in studies by

Table 1: Age- and gender-wise distribution of children with Down's syndrome

Age group (years)	Male	Female	Percentage
<1	22	30	43.3
1–5	18	14	26.6
5–10	12	10	18.3
>10	4	10	11.6

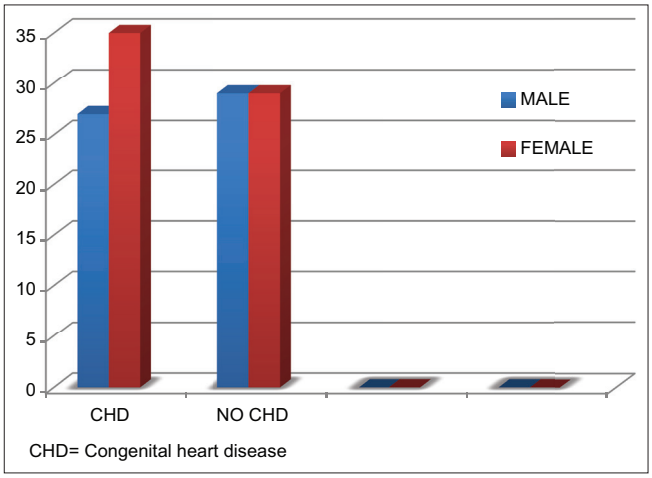

Figure 1: Gender distribution of Congenital heart disease

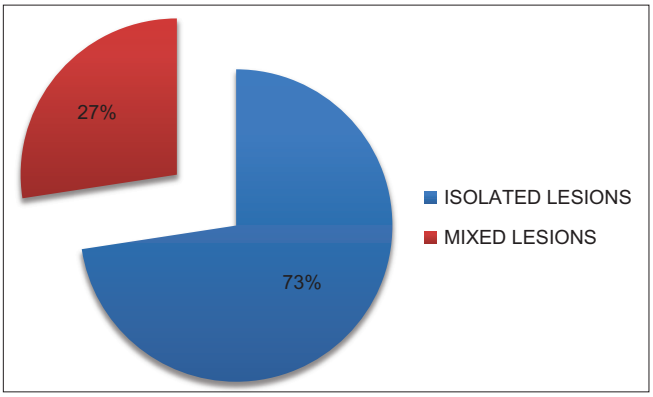

Figure 2: Spectrum of congenital heart disease

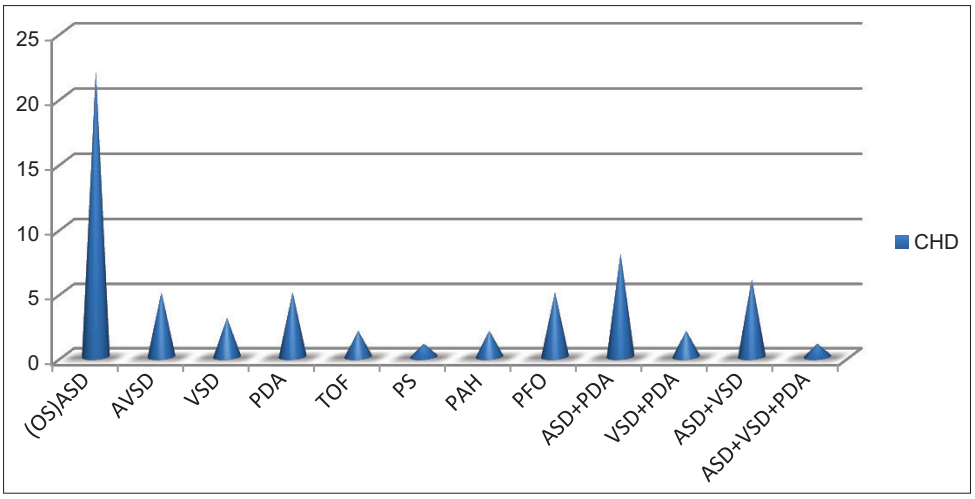

Figure 3: Pattern of CHD in Down's syndrome

Table 2: Spectrum of congenital heart disease in Down's syndrome

Cardiac lesion	Total (*n*=62)	Percentage
(OS) ASD	22	36
AVSD	5	8.2
VSD	3	5
PDA	5	8.2
TOF	2	3.3
PS	1	1.6
PAH	2	3.3
PFO	5	8.2
ASD+PDA	8	11.5
VSD+PDA	2	3.3
ASD+VSD	6	9.8
ASD+VSD+PDA	1	1.6

AVSD: Atrioventricular septal defect, VSD: Ventricular septal defect, ASD: Atrial septal defect, PDA: Patent ductus arteriosus, TOF: Tetralogy of Fallot, PS: Pulmonary stenosis, PAH: Pulmonary arterial hypertension, PFO: Patent foramen ovale

Table 3: Gender-wise distribution of CHD in Down's syndrome

CHD	Male (%)	Female (%)
Isolated CHD	20	25
Mixed CHD	7	10
Total	27 (44)	35 (56)

CHD: Congenital heart disease

Narayanan *et al.*[3] and Alsuhaibani *et al.*,[4] respectively. The result of our study (52%) correlated with the worldwide incidence of 40–60% CHD in Down's syndrome and was comparable to many other studies.

The incidence of CHD in male was 44% and the female Down's syndrome children outnumbered with 56% in our study. The meta-analysis by Diogenes *et al.*[5] observed that female gender is a risk factor for the presence of CHD in Down's syndrome and AVSD alone showed a higher frequency in female gender. The same result was found in the study by Mourato *et al.*[6] with a female preponderance of 56.1%. However, no gender difference was noted in the prevalence of CHD in the study by Morsy *et al.*[1] In another study from South India,[7] the incidence of CHD was more in male (55%) than female children [Table 3].

Isolated CHDs topped the list in our study with 73% while mixed lesions were found in 27%. In Nisli *et al.*[8] study, isolated lesions were found in 77.6% and mixed lesions in 22.4%. Shrestha and Shrestha[9] study had 65% isolated CHDs and 35% mixed lesions.

The most common isolated CHD in this study was ostium secundum ASD (36%). In studies by Alsuhaibani *et al.*[4] and Mourato *et al.*,[6] ostium secundum ASD was the most common lesion in 33.5% and 51.78%, respectively. In comparison, the most common isolated lesion was AVSD in Benhaourech *et al.*,[2] 85.2%, Narayanan *et al.*[3] showed 27.3%, and El-Attar[10] study showed 33.3%. Our study had AVSD in 8% probably due to ethnic variation and early deaths of these children. Many genetic studies also showed that AVSD had the most significant gender and ethnic differences.

The association of TOF in Down's syndrome is infrequent and accounts for 5–8% of all CHDs in trisomy 21. TOF was found in 3% in our study. El-Attar study had 2.2% TOF and Shrestha and Shrestha[9] study had 7.5% TOF.

The most common mixed lesion in this study was PDA with ASD (11.5%) followed by ASD with VSD (9.8%). The most common association in Sharifi *et al.*[11] study was ASD with VSD in 19.9% and VSD with PDA in 9%.

Pulmonary arterial hypertension (PAH) was found in 3.3% of cases in our study while higher incidence was found in many other studies. PAH was found in 53% in Benhaourech *et al.*[2] study and in 37.5% in Mourato *et al.*[6] study. The low

incidence in our study could be because infants form the major number in our study and further follow-up of these children will give the exact incidence of PAH.

CONCLUSIONS

CHD was found in 52% of children with Down's syndrome in our study. Hence, early diagnosis by early cardiac screening as recommended by the American Academy of Pediatrics and regular follow-up is the key for early surgical intervention to avoid irreversible hemodynamic consequences and mortality in these children.

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How to cite this article: Vanitha B, Sangeeth S, Balasankar S. Profile of Congenital Heart Disease in Children with Down's Syndrome Attending an Early Intervention Center in a Teaching Hospital in South India. Int J Sci Stud 2020;8(1):68-71.

Source of Support: Nil, **Conflicts of Interest:** None declared.