

## Original Research Article

## Profile of Congenital Heart Anomalies in Children with Down's Syndrome

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**Abstract: Background:** Down's syndrome (DS) is one of the commonest chromosomal disorder and congenital heart disease (CHD) is consider as the most important malformation of DS with an incidence of 40 to 60%. Pattern of CHD in DS have wide variation and is responsible for a significant number of mortality as well as morbidity in children specially during first year of life.

**Materials and Methods:** It was a Prospective, observational single center study done over a period of one year from January' 2020 to December' 2020. Chromosomal study and Echocardiography done to all children after admission.

**Results:** Of the 98 study children 51 had congenital heart disease (52.04%), among them ostium secundum atrial septal defect was the commonest one (31.37). **Conclusion:** There is very high incidence of CHD among Down's syndrome (DS) children, and it was 52.04% in this study. Early cardiac evaluation of DS children is of paramount importance for appropriate management plan and timely intervention in order to reduce mortality and morbidity of patients with CHD in Down's syndrome.

**Keywords:** Congenital, Anomalies & Down's syndrome

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### INTRODUCTION

Down's syndrome (DS), originally described by John Langdon Down in 1866, is a common birth defect [1]. It is a chromosomal disorder which is often associated with morphological and structural defects affecting about 1 in 400-1500 newborns [2]. Individuals with Down's syndrome is a person having 47 chromosomes instead of 46, with an additional replica of chromosome number 21 [3]. Ninety-five percent of DS cases are due to the trisomy 21 and 1% of persons with trisomy 21 are mosaics. DS can also be caused by translocation involving chromosome 21 [4]. This trisomy gives rise to multiple complications as part of the syndrome and is prone to having congenital heart defects (CHDs), gastrointestinal anomalies, and endocrinopathies including hypothyroidism and diabetes [5]. Congenital heart disease (CHD) is considered to be the most important malformation of DS causing morbidity and mortality [6], specially during the first two years of life [7]. The reported incidence of CHD in Down syndrome patients is between 40 and 60% [8-10]. The most commonly described defect is complete AVSD which comprises

30-40% of all cardiac defects [11]. Other frequently encountered lesions are isolated secundum atrial septal defect (ASD), ventricular septal defect (VSD), patent ductus arteriosus (PDA), and tetralogy of Fallot (TOF) [12]. In order to take preventative measures and to improve the quality of life, it is important to be familiar with the incidence and anatomical characteristics of CHD in DS, associated complications, as well as causes of morbidity and mortality. As the type of CHD and the timing of repair affect the prognosis, appropriate care and interventions of cardiac anomalies at time is crucial for optimal survival. The aim of this study, therefore, was to determine the Pattern, and frequency of congenital heart disease (CHD) in children with Down's syndrome at Dhaka shishu Hospital.

### OBJECTIVES

The objective of this study was to determine the pattern and frequency of congenital heart disease in children with Down's syndrome admitted at Dhaka Shishu Hospital.

## MATERIALS AND METHODS

This was a prospective and observational study conducted for a period of one year from January 2020 to December 2020, at the department of paediatric medicine and paediatric cardiology, Dhaka Shishu Hospital. All admitted children with Down syndrome were the target population in this study and the samples for this study were children with Down syndrome having congenital heart disease. All suspected Down syndrome cases underwent full clinical assessment including phenotypic features such as flat facial profile, slanted palpebral fissures, epicanthal folds, brachycephaly, single palmar crease, clinodactyly, hyperflexible joints, and hypotonia and were included for the study. General characteristics such as gender, age at diagnosis and mother's age at delivery were also recorded. Chromosome analysis done to confirm the diagnosis of Down syndrome. All patients were examined by plain chest X-ray, electrocardiogram and ultrasound of the heart (2-D echocardiography and color doppler). The echocardiography examination was performed at the cardiology department using a GE Vivid 9 ultrasound machine.

## RESULTS

A total of 98 children with Down's syndrome were included in the study where 46 children were male and 52 female contributing to 46.93% and 53.06%, respectively. The ratio of male to-female of the sample was 1:1.3. Age of the children in the study varied widely starting from 15 days to oldest one of age 16 years (Table-1). Majority of the patients in this study were infants 48 (48.97%), 24 (24.48%) were between 1 and 5 years, 16 (16.32%) between 5 and 10 years. Remaining 10 (10.20%) above 10 years of age. In this study, out of 98 children with Down's syndrome, congenital heart disease found in 51 children, remaining 47 had normal cardiac status. So in our study the involvement of CHD in Down's syndrome was 52.04%. The most common isolated heart disease was ostium secundum ASD contributing to 30.76% (n = 16), followed by AVSD 26.92% (n = 14), VSD 11.53% (n = 6), TOF in 9.61% (n = 5), PDA 5.76% (n = 3), and pulmonary stenosis in 5.76% (n = 3). Multiple combination of congenital heart disease found in 5 patients. Among the 5 patients who had multiple type of congenital heart disease, two patients were diagnosed PDA+ASD, one ASD with VSD another one VSD with PDA, and the remaining patient had AVSD+TOF.

**Table-1: Demographic characteristics of children with Down syndrome (n = 98)**

Demographic characteristic.	No	Percentage (%)
<b>Age groups (years)</b>		
< 1	48	48.98
1—5	24	24.48
5—10	16	16.32
> 10	10	10.2
<b>Sex</b>		
Female	52	53.06
Male	46	46.93
Male Female ratio	46/52	01:01.3

**Table-2: Involvement of congenital heart disease in Down's syndrome (n=98)**

Types	NO	Percentage (%)
Isolated CHD	46	46.93
Multiple CHD	5	5.10
No CHD	47	47.95
<b>Total</b>	<b>98</b>	<b>100</b>

**Table-3: Distribution of study sample according to the type of CHD (n=51)**

Types of CHD	No.	Percentage (%)
ASD	16	31.37
AVSD	14	27.45
VSD	6	11.76
TOF	5	9.80
PDA	3	5.88
Pulmonary stenosis	2	3.92
Multiple combination	5	9.80
<b>Total</b>	<b>51</b>	<b>100</b>

## DISCUSSION

This study with 98 Down's syndrome children showed male: female ratio 1: 1.3 which is almost similar to the study result done by Morsy *et al.*, [14]. The incidence of cardiac involvement in the form of CHD in this study was 52.04%, the result of this study correlated well with the worldwide incidence of 40–60% CHD in Down's syndrome and was comparable to many other studies. Incidence found in study by Morsy *et al.*, [14] was 58.6% and that by Narayanan *et al.*, [15]. 63.4%. The commonest isolated CHD in this study was ostium secundum ASD (31.37%), Alsuhaibani *et al.*, [16] and B Vanitha *et al.*, [12] also found ASD as the commonest cardiac lesion in DS children. In our study AVSD was the second most common CHD 27.45%, Narayanan *et al.*, [15] and El-Attar LM [17] found AVSD in their study as 27.3%, and 33.3% respectively. Other significant cardiac lesion found in this study was VSD (27.45) and TOF (11.76). Dennis R. Delany *et al.*, [18] found VSD in 19%, and Thyviyaa Rajamany *et al.*, found TOF in 10.71% of children with DS [19]. The most common mixed lesion in this study was PDA with ASD followed by ASD and VSD. Less frequent lesions found in this study were PDA and Pulmonary stenosis.

## CONCLUSION AND RECOMMENDATIONS

There is a high incidence of CHD among DS children. In our study CHD was found in 52.04% of children with DS. Early diagnosis by cardiac screening including echocardiography is crucial for appropriate management plan and timely intervention in order to reduce mortality and morbidity of patients with CHD in down's syndrome

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