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Mapping Congenital Heart Disease in Infants with Down Syndrome in a Tertiary Care Hospital

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ABSTRACT

Background & objective: Cardiovascular anomalies are a primary contributor to morbidity and mortality in infants with Down Syndrome (DS). Despite the known association, the specific underlying etiologies of these defects remain obscure. International guidelines mandate early screening for congenital heart disease (CHD) in DS patients, as timely diagnosis and intervention significantly improve health outcomes and life expectancy. This study aimed to identify the prevalence and specific patterns of CHD among infants with DS in a specialized clinical setting.

Methods: This cross-sectional study was conducted at the Child Development Center (CDC) of BIRDEM General Hospital 2 and the Pediatric Cardiology Echocardiography Lab of Ibrahim Cardiac Hospital and Research Institute (ICHRI) from July 2021 to June 2022. A total of 55 children with Down Syndrome were primarily screened for congenital heart diseases (CHDs). After clinical and echocardiographic evaluation, infants with DS who had CHD (n = 31) were advised for karyotyping. Sociodemographic, clinical and echocardiographic data were collected and analyzed.

Results: Congenital heart disease was identified in 31(56.4%) of the 55 infants. Of these, 19(61.3%) presented with a single cardiac abnormality, while 12(38.7%) exhibited multiple anomalies. Atrial Septal Defect (ASD) was the most frequent lesion (61.3%), with 12 infants (38.7%) having isolated ASD. Maternal age was relatively low, with >70% of mothers being under 35 years of age. Karyotyping was confirmed in 26 cases, of which 25(96.1%) were Trisomy 21 (47, XX/XY+21) and one (3.9%) was a chromosomal translocation (46, XX, t13/21).

Conclusion: CHD is highly prevalent among infants with DS in Bangladesh. Early echocardiographic screening and prompt referral to specialized cardiac centers are essential to prevent irreversible complications, such as pulmonary hypertension, and to optimize medical and surgical outcomes.

Keywords: Congenital heart diseases, Down Syndrome, Trisomy 21, Tertiary care, Bangladesh etc.

ORIGINAL ARTICLE

INTRODUCTION

Down syndrome (DS), or trisomy 21, is the most common chromosomal abnormality associated with intellectual disability and multi-systemic developmental delays.[1] While medical advancements have significantly improved the

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quality of life for these individuals, congenital heart disease (CHD) remains a critical clinical challenge. CHD is present in approximately 40-62% of newborns with Down syndrome and represents a leading cause of morbidity and mortality during the first two years of life.[2,3]

The most prevalent lesions in this population are endocardial cushion defects (43%), primarily resulting in atrioventricular septal defects (AVSD), followed by ventricular septal defects (VSD) (32%) and atrial septal defects (10%). Notably, nearly 70% of all endocardial cushion defects are associated with Down syndrome. Without early identification, these defects often lead to the rapid progression of pulmonary vascular disease (PVD). Consequently, international guidelines, such as those from the Down's Syndrome Medical Interest Group (DSMIG), advocate for mandatory echocardiographic screening within the first six weeks of birth, even in the absence of clinical signs, to facilitate timely intervention.[4,5]

A landmark study by Figueroa et al.[3] demonstrated that CHD prevalence is significantly higher in hospitalized DS patients, emphasizing the severity of cardiac involvement. However, a significant study gap exists regarding the specific patterns and outcomes of DS infants in South Asia, particularly Bangladesh. Existing literature in this region is scarce; while high mortality is suspected due to delayed diagnosis, empirical evidence is limited.

This study aims to address this gap by identifying the specific patterns of congenital cardiac anomalies and associated demographic factors in a tertiary care hospital in Bangladesh. By establishing these patterns, the study seeks to underscore the importance of early neonatal screening and provide the evidence base needed to improve survival rates through timely medical and surgical interventions.

METHODS

This descriptive cross-sectional study was conducted at the Child Development Center (CDC) of BIRDEM General Hospital 2 and the Pediatric Cardiology Echocardiography Lab of ICHRI over a one-year period (July 2021-June 2022). A total of 55 infants presenting with clinically suspected DS (flat facial profile with upward slanting eyes, developmental delay and/or hypotonia) were enrolled. Following informed parental consent, all infants were evaluated for cardiac lesions.

Transthoracic echocardiography was performed by a pediatric cardiologist using a Philips Affinity 50C machine. DS was confirmed via karyotyping, where possible. Sociodemographic variables, including infant age, sex, birth order, and maternal age at delivery, were recorded. Data analysis was performed using SPSS version 22, utilizing descriptive statistics to illustrate the frequency and distribution of CHD types.

RESULTS

Cardiac defects were identified in 31(56.4%) of the 55 cases. The cohort included 14(45.1%) males and 17(54.9%) females (M:F ratio 9:11). The mean age at diagnosis was 6.9 ± 2 months, and the mean maternal age at delivery was 31.7±5.4 years. The majority (70.9%) of mothers were < 35 years old. Regarding birth order, 12(38.7%) affected infants were the second child, followed by the first child (29.0%) and the third child (19.3%) (Table I).

Echocardiographic findings revealed that 19(61.3%) infants had a single defect, while 12(38.7%) had multiple defects. Atrial Septal Defect (ASD) was the most common CHD (61.3%). Isolated ASD was found in 12(38.7%) infants, while 7(22.6%) had ASD associated with other anomalies. AVSD, Tetralogy of Fallot (ToF), and Patent Ductus Arteriosus (PDA) each occurred in 2(6.4%) infants. High pulmonary pressure was observed in 4(12.9%) cases with CHD (Table II, III & Fig.1). Karyotyping was documented for 26 cases: 25(96.1%) were Trisomy 21 and 1(3.9%) was a translocation of chromosome (46, xxt13/21).

Table 1: Socio-demographic profile of DS infants with CHD (n = 31)

Socio-demographic profile	Frequency	Percentage
Sex	14	45.2
Male	17	54.8
Female		
Infant's Age		
1 to 6 months	19	61.3
>6 months	12	38.7
Maternal age		
< 20	2	6.5
20 – 24	7	22.6
25 – 29	8	25.8
30 – 34	5	16.1
>35	9	29.0
Birth order		
1st	9	29.0
2nd	12	38.7
3rd	6	19.4
4th and above	4	12.9

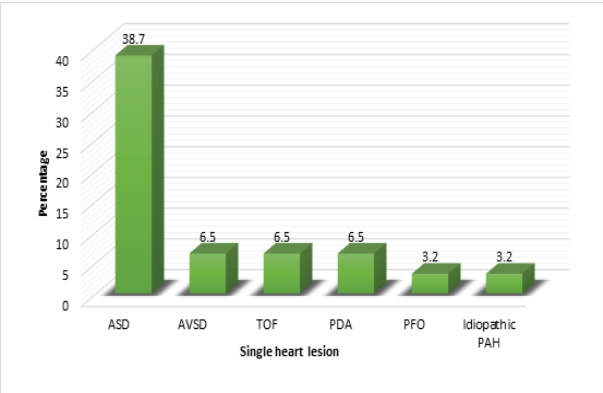

Fig. 1: Distribution of children by presence of single heart lesion (n = 31)

Table II: Distribution of infants with multiple heart lesion (n = 31)

Multiple heart lesion	Frequency	Percentage
ASD+PDA	4	12.9
VSD+PDA	1	3.2
ASD+VSD+ PDA	1	3.2
Mild PAH +PFO	1	3.2
VSD, Severe PAH, Small ASD	1	3.2
Moderate VSD, Severe PAH	1	3.2
VSD, PFO,	1	3.2
Large ASD, Mild RPA stenosis, Moderate PAH	1	3.2

DISCUSSION

This study underscores the significant intersection between trisomy 21 and cardiovascular anomalies. Our findings reveal that over half (56.4%) of the screened infants presented with concomitant CHD. ASD emerged as the predominant lesion (61.3%), followed by ASD associated with PDA (12.9%). This high prevalence reaffirms CHD as a principal driver of morbidity in this population.[6]

The mean age of 6.9 months at diagnosis is congruent with findings from Morocco[7] but represents earlier presentation than reported by Chowdhury et al.[8] This shift likely reflects the strength of our tertiary referral center in facilitating screening. While advanced maternal age is a globally recognized risk factor for DS,[9] over 70% of mothers in our study were under 35. This divergence from international registries[10] suggests that in regions with high fertility rates in younger women, the absolute number of DS births remains high despite lower age-specific risks.[11]

Genetically, Trisomy 21 via non-disjunction remains the primary etiology (96.1%), consistent with global literature.[7] However, the anatomical pattern in our study, led by ASD, deviates from Western cohorts where AVSD typically dominates.[7] This variance, also noted in Chittagong,[8] suggests potential geographical or ethnic nuances in phenotypic expression. Early diagnosis remains the only viable pathway to mitigate irreversible pulmonary vascular damage.

Limitations

This study is constrained by its single-center design and small sample size. The findings may reflect a higher prevalence of complex cases at a specialized facility and may not represent the national prevalence across Bangladesh.

Conclusion

The prevalence of CHD is high among infants with DS in Bangladesh. The significant burden of

cardiac anomalies necessitates a standardized, mandatory neonatal echocardiographic screening protocol. Early detection and timely referral to specialized cardiac centers, remain the most critical factors in preventing irreversible complications like pulmonary hypertension and ensuring a significantly improved life expectancy for children with Down Syndrome in Bangladesh.

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