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Research Article

PREVALENCE OF CONGENITAL HEART DEFECTS ASSOCIATED WITH DOWN SYNDROME IN TERTIARY CARE HOSPITAL OF NORTH INDIA

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A B S T R A C T

Objectives: To define the frequency and patterns of congenital heart disease (CHD) among children with Down syndrome (DS) in tertiary care hospital of North India
Methods A cross sectional study was conducted in 110 consecutive patients with Down syndrome attending the Department of Pediatric Government Medical College , from December 2016 through September 2018. All children were offered cytogenetic analysis and were subjected to echocardiography. We excluded term and preterm children with patent ducts arteriosus (PDA) and persistent foramen oval spontaneously resolved during the first 4 weeks of life.
Results: Among the 110 patients with Downs syndrome , congenital heart disease was present in 51 (46.6%).. The most common heart diseases were atrio ventricular septal defect in 254 18patients (35.2%); ventricular septal defect in 10 (19.6%) atrial septal defect in 8[15.6%] .Mixed shunt lesion in 2[3.9%].Among cyanotic CHD classical Tetrology of Fallot was commonest present in 3 patients [5.8%]
Conclusion: The high prevalence of congenital heart disease among the patients at the Down syndrome [46.6%]was similar to findings from other studies and justifies investigation during the neonatal period, so as to decrease mortality and morbidity.

INTRODUCTION

Down syndrome is the commonest chromosomal aneuploidy, and the commonest cause of severe mental retardation, with an incidence of 1 in 700 live births. In India the frequency of Down syndrome is estimated to be 1 in 1150 (0.87/1000) [1]. In 95 % of cases, Triosomy 21 occurs due to non-disjunction, 4 % are due to parental or de novo translocation and 1 % are due to mosaicism [2]. Cytogenetic analysis has an important role not only in the confirmation of diagnosis but also for the prediction of recurrence risk and future genetic counselling. Downs syndrome is associated with various comorbidities like gastrointestinal problmens ,respiratory issues and musculoskeletal issues. Congenital heart disease (CHD) occurs in 40-50 % of children with Down syndrome [2]. Their life expectancy and quality of life can be significantly improved by early surgical intervention. The most common forms of CHD reported in children with Down syndrome, in descending order, are atrio-ventricular septal defect (AVSD), ventricular septal defect (VSD) and atrial septal defect (ASD).Congenital heart disease is the most common cause of death among patients with DS and affected children have an increased risk of mortality.[3] Therefore, it is essential that every patient with confirmed DS to undergo cardiac evaluation in early life.

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The objective of the present study was to determine the prevalence and profile of congenital heart disease among patients treated at as Down syndrome in one of the main paediatric referral hospital of Kashmir between December 2016 to September 2018

MATERIALS AND METHODS

This was prospective, observational study conducted in the Department of Paediatrics, between December 2016 and September 2018. This is the main referral hospital for region, and hosts the pediatric cardiology facilities. The diagnosis of DS was made by the Pediatrician based on typical phenotypical features and confirmed by cytogenetic studies. The exclusion criteria comprised: children with dismorphic features and not confirmed to be DS by cytogenetic studies.

All included children of DS underwent a thorough clinical examination and detail history, a three-generation pedigree was generated in all patients to assign the degree of consanguinity. All patients were subjected for routine laboratory tests, like CBC, serum electrolytes, renal function tests, blood sugar, chest X-ray, routine urine exam, ECG, and other relevant investigations. All patients underwent echocardiographic by single paediatric cardiologist using Seimens Accuson S 2000 Children with only one anatomical heart defect, such as ventricular septal defect (VSD),

atrial septal defect (ASD), patent ductus arteriosus (PDA), or with a well-known combination, such as Tetralogy of Fallot (TOF) were classified to have isolated CHD. The combination of VSD, ASD, and PDA was categorized as mixed left to right shunt. Presence of PDA , in preterm or term babies at birth, was considered normal unless these lesions persisted beyond 12 week of age.

RESULTS

The total of 110 patients with DS were enrolled in our study.Of the 110 children with DS ,CHD was present in 51 children comprising 46.6%Table 1 depicts the age distribution of the children diagnosed with CHD .8 patients presented in neonatal age [15.6%].Majority of children presented in infancy n=31 [60.7%].8 cases were between 1 to 5 year [15.6%],while least number of children were more than 5 year n=5[9.8%]

Table 1

AGE	N=51	Percentage
Neonate[<1 month]	8	15.6%
1 month -1 year	31	60.7%
1 -5 year	8	15.6%
6-12 year	5	9.8%

Table 2 depicts the incidence of consangious marriage in 110 children

	N=110	Percentage
Consangious	32	29.09%
Non Consangious	78	70.09%

Of all the children with DS consanguity was present in 32 /110[29.09%] while nonconsangious marriage was present in 78/110[70.09%]

Of 51 patients with CHD 13 were product of consangious marriage [25.4%],while as non consangious marriage was present in 38 cases [75.4%]

Table 3 depicts the co relation of consanguity of DS with CHD .

Consanguity	N=51	Percentage
Present	13	25.4%
Absent	38	74.5%

Table 4

	N=51	Percentage [46.6%]
Acyanotic CHD	**N=44**	86.2%
Atrioventricular septal defects balanced [AVSD]	18	35.2%
AVSD unbalanced Right ventricular dominant	3	5.8%
AVSD unbalanced Left ventricular dominant	1	1.9%
Atrial septal defect[ASD]	8	15.6%
Ventricular septal defect[VSD]	10	19.6%
Patent ductus arteriosus[PDA]	1	1.9%
Mixed shunt lesions	2	3.9%
Pulmonary stenosis	1	1.9%
Cyanotic CHD	**N=7**	13.7%
Classical Tetralogy of fallot	3	5.8%
AVSD TOF	1	1.9%
VSD pulmonary atresia	1	1.9%
Single ventricle	1	1.9%
Transposition of great vessels[TGA]	1	1.9%

Table 4 depicts the distribution of various CHD in Downs syndrome..51 cases of DS had associated CHD [46.6%] of which[86.2%] patients had acyanotic CHD whereas cyanotic CHD was present in 7 patients [13.7%]. The most frequent isolated congenital heart defect in our study was balanced atrioventricular septal defect (AVSD, complete type) identified in 18/51(35.23%), followed by ventricular septa defect in 10/51[19.6%] ,Atrial septal defect including both Fossa ovalis ASD and Ostium primum ASD seen in 8/51[15.4%].Unbalanced AVSD with right ventricular dominance was seen in 3 patients [5.4%]while left ventricular dominant unbalanced ASVD was seen in 1/51[1.9%] .Mixed shunt lesion were present in 2/51[3.9%],PDA and Pulmonary stenosis was present in 1 patients each [1.9%] Cyanotic CHD was present in 7 patients [13.7%].Classical TOF was commonest cyanotic CHD present in 3/51[5.8%],

DISCUSSION

It is estimated that 21,000 babies are born with Down syndrome in India every year. Cardiac lesions are the major cause of morbidity and mortality among them. The prevalence of congenital heart disease was similar to the findings from other studies, which have been between 44% and 62%.[4,5].Incidence of CHD in our study was 51/110[46.6%].Majority of patients were between 1 month to 1 year followed by children between 1 to 5 year of age.Least number of children were more than 6 years. This is explained by attrition of children with DS due to comorbid conditions especially Congenital heart disease Of the total 110 cases of DS consanguity was present in 32 cases [29.09%] while Non consangious marriage was seen in 78 cases [70.09%].Of the 51 patients with associated CHD consanguity was present in 13 cases[25.4%] while consanguity was absent in 38 cases [75%].Consanguity was less often present DS.Cconsanguinity appears to be an associated risk factor for the severity and rate of CHD [6].Although studies world over has observed an association between the consanguinity and CHD [7]but simultaneously has discouraged the association of consanguinity and DS [8].

The types of heart defect may vary according to geographic region. In this study, the most common type was atrioventricular septal defect, which was similar to the findings from the United States[9] France[10]Turkey[11] and Sweden[12]where atrioventricular septal defect was more frequent. And contrary to studies from brazil[13]South Korean[14]and Libyan[15]studies,were ASD was commonest CHD. The most common single cardiac lesion was ventricular septal defect in China (in approximately 40% of Down syndrome children)[16]and patent ductus arteriosus in Guatemala [17] and Mexico. [18] Nonetheless, in United States, Freeman *et al*. found that the most common type of congenital heart disease was atrioventricular septal defect (47%), in the Atlanta Down Syndrome Project in 1998 [3]and, ten years later, the rates for ventricular septal defect (19.2%), atrial septal defect (18.6%) and atrioventricular septal defect (17.2%) were similar in a report from the National Down Syndrome Project. [5] brazil .Among the cyanotic CHD most common was classical TOF was commonest cyanotic CHD. In our study majority of cases presented below 1 year of age [72%].This finding is striking as it carries significant prognostic value as children with CHD are diagnosed timely. Screening of all DS for CHD is protocol in our hospital. This makes early diagnosis feasible coupled with effective surgical

treatment is the factor mainly responsible for decreasing the morbidity and mortality rates in this population.

Limitation; since our study is conducted in referral centre prevalence of CHD in DS may not reflect the true prevalence of disease. Sample size of our sample is also less so our data is suggestive in nature.

CONCLUSION

The high prevalence of congenital heart disease among Down syndrome patients (46.6%) justifies its investigation during the neonatal period, with the aims of decreasing the mortality and morbidity rates, having fewer visits to clinics or hospitalization and ensuring better operating conditions, if this becomes necessary, with less suffering for patients and their families, lower costs and improvement of overall health, wellness and development.

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