FREQUENCY OF CONGENITAL HEART DISEASE IN PATIENTS WITH DOWN SYNDROME

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ABSTRACT

OBJECTIVES

Frequency of congenital heart disease in patients with Down syndrome.

METHODOLOGY

This study was conducted in Hayatabad Medical Complex, Peshawar from 10th May 2021 to 9th November 2021. A total of 377 patients of age 1 to 10 years and both gender were included in the study. Those with already diagnosed cases of congenital heart disease and dysmorphic features other than Down syndrome were excluded from the study. All patients were undergone echocardiography and patient with PDA, VSD, ASD, ASVSD, and TOF were labeled as having congenital heart diseases.

RESULTS

Mean age was 5.96 ±1.954, Males were 176 (46.7%) while females were 201 (53.3%), Mean birth weight was 3.45 ± 0.801 kg, Mean age of the mother was 38.25 ± 6.797 years, Congenital heart diseases were present in 157 (41.6%) of the patient while it was not present in 220 (58.4%) of patients, there was no association between congenital heart disease and age of mother, age of the child, sex of child or weight of the child (P >0.05).

CONCLUSION

Congenital heart disease is very common in patients with Down syndrome. It is recommended that at the time of diagnosis of this disease, the patient should be screened for congenital heart disease.

KEYWORDS: *Congenital Heart Disease, Down Syndrome, Echocardiography*

INTRODUCTION

Down's syndrome is one of the most common viable genetic disorders, caused by trisomy of chromosome 21, with mental and physical developmental abnormalities.[1] The incidence of DS is 1 to 2 per 1000 live births.[2,3] Trisomy 21 occurs in 90.9% of individuals due to non-disjunction, 5.05% due to Robertsonian translocation, and 4% due to mosaicism.[4] Down's syndrome patients can present with a combination of many dysmorphic features but the presence of 10 hallmark symptoms in newborns have been identified to assist in postnatal confirmation of DS including flat facial profile, slanted palpebral fissures, anomalous low sit ears, hypotonia, poor Moro reflex, dysplasia of mid phalanx of the fifth finger, transverse palmar crease, excessive skin at the nape of the neck, hyper flexibility of joints, and dysplasia of pelvis.[5] Despite the increase in antenatal detection, the prevalence of babies born with Down's syndrome has risen by 25% during the past 30 years. Maternal age is a primary risk factor for DS because the increase in maternal age increases the incidence of DS in new born.[6] Congenital heart disease is a defect in the structure of the heart and its great blood vessel present at birth.[7] 40-50% of newborns with Down's syndrome have some form of congenital heart diseases Without proper surgical treatment, morbidity and mortality rate in the DS population has been largely attributed to their having a higher incidence of CHD due to which 25 - 30% pof patients die in infancy.[3,8,9] The most common CHD in the western literature is atrioventricular septal defect (AV canal).[10] Of those with CHD, about 80% have an atrioventricular septal defect or ventricular septal defect.[7] A study conducted in brazil showed that among the 604 patients with congenital heart diseases atrial septal defect in 254 patients (42.1%); a total atrioventricular septal defect in 91 (15.1%); combined atrial septal defect and ventricular septal defect in 88 (14.6%); ventricular septal defect in 77 (12.7%); patent ductus arteriosus in 40 (6.6%); patent foramen ovale in 34 (5.6%) patients; tetralogy of Fallot in 12 (2%); and other diseases in 8 (1.3%).[11] Similarly, i study conducted in Korea, showed frequency of congenital heart diseases among town syndrome was 254(56.9%). The pattern of congenital heart disease showed atrial septal defect (30.5%) ventricular septal defect (19.3%),

PDA (8%), and atrioventricular septal defect(9.9%).[12] All patients with Down's syndrome should be evaluated with careful physical and echocardiographic examination on admission. Echocardiography has become the method of choice for the diagnosis of congenital heart defects in neonates with Down's syndrome.[13] As no such study has been conducted in our population for the last five years so this study is providing us with the latest and updated information regarding the frequency of congenital heart diseases in children with Down's syndrome. The results of this study have been shared with other health professionals and will be used for other research work. Al, so advanced surgical procedures and cardiac intervention techniques are available for the better and early management of these CHD. This will not only help to know the frequency but will also reduce the morbidity and mortality associated with CHD in children with DS.

METHODOLOGY

A Descriptive cross-sectional was conducted in the Department of Pediatrics, Hayatabad Medical Complex, Peshawar with the approval letter no: 626/HEC/B&PSC/2022 from 10th May 2021 to 9th November 2021. The sample size was 377 being the 56.9% anticipated proportion of CHD in Down's syndrome, a 95% confidence interval, and a margin of error 5 of % according to the WHO formula for sample size.[12] Inclusion criteria: Children with Down's syndrome diagnosed clinically, both male and female children with age of range 1- 10 years. While patients with dysmorphic features other than Down's syndrome and with Down's syndrome already diagnosed with congenital heart disease were excluded from this study. All the children fulfilling the inclusion criteria were enrolled in the study through OPD and Pediatrics. Informed consent was taken from their parents or relatives after explaining the importance and benefits of the study and procedure. All patients included were subjected to detailed history and clinical exams detailed prenatal, natal, and postnatal history were taken with relevant information by the trainee himself. For all patients echocardiogram of our hospital is done by either consultant under the supervision a of consultant cardiologist. The demographic details and like age, maternal age, birth weight along admission, clinical examination, echocardiography findings, was entered into a pre-designed proforma.

RESULT

The total sample size was 377 children. The mean age of female were±1.954. Males were (46.7%) while females were 20The mean3.3%). The mean weight was 3.45 ± 0.801 kg. The mean age of mother disease is 8.25 ± 6.797 years. Congenital patient diseases were present in 157 (41.6%) of patients while as not present in 220 (58.4%) of patients. Mothers of age equal to were and then 35 years were and (31.3%), 36 to 45 years were 205 (54.4%) while that of more than 45 years were 54 (24.3%). Stratification of gender showed that 77 (43.8%) male children were having congenital heart disease while 99 (56.3%) males were having no congenital heart disease. In females, 80 (39.8%) were with congenital heart disease while 121 (60.2%) with no CHD p-value difference wasp-value significant as the p-value for children for children's1children'stification of, children age showed that in children with age 1-5 years 72 (45.9%) were congenital heart disease while 85 (54.1% the) were not having congenital heart disease. In age group of 6the - 10 years 85 (38.6%) were having congenital heart p-value while 135 (61.4%) were having no CHD. The p-value children was 0.161(table-2). In stratification of children's weight of less than 3.5, there were 87 (41.2%) children with congenital heart disease while 124 (58.8%) with no congenital heart disease. In weight of 3.5 to 4.5 kg there were 63 (46.0%) with congenital heart disease while 74, (54.0%) with no CHD. In weight of more than 4.5 kg there were 7 (24.1%) with congenital heart disease and 22 (75.9%) without CHD. This difference was not significant (p=0.083) (table-3). Stratification of mother age against congenital heart disease showed that the mother age of 35 or less there 50 (42 maternal children were having congenital heart disease while 68the congenital l%) were having no congenital heart disease. In age group of 36 to 45 years, there were (39.0%) with initial heart disease while, 125 (61.0%) with no CHD. In age group of more than 45 years were 27 (50.0%) with congenital heart disease and 27 (50.0%) without CHD. This difference was not significant (p=0.343) (table-4).

Table 1: Gender Wise Distribution of Congenital Heart Disease

			Congenital Heart Disease		Total	P-Value
			Yes	No		
Gender of Child			77	99	176	0.438
	Male	% within Gender of Child	43.8 %	56.3 %	100.0 %	
			80	121	201	
	Female	% within Gender of Child	39.8 %	60.2 %	100.0 %	
Total			157	220	377	
		% within Gender of Child	41.6 %	58.4 %	100.0 %	

Table 2: Age Wise Distribution of Congenital Heart Disease

			Congenital Heart Disease		Total	P-Value
			Yes	No		
Child age groups			72	85	157	0.161
	1 to 5 years old	% within Child age groups	45.9 %	54.1 %	100.0 %	
			85	135	220	
	6 to 10years old	% within Child age groups	38.6 %	61.4 %	100.0 %	
Total			157	220	377	
		% within Child age groups	41.6 %	58.4 %	100.0 %	

Table 3: Weight Wise Distribution Congenital Heart Disease

Child Weight Groups		Congenital Heart Disease		Total	P-Value
		Yes	No		
		87	124	211	0.083
less than 3.5 kg	% within Child weight groups	41.2 %	58.8 %	100.0 %	
		63	74	137	
3.5 to 4.5 kg	% within Child weight groups	46.0 %	54.0 %	100.0 %	
		07	22	29	
Greater than 4.5 kg	% within Child weight groups	24.1 %	75.9 %	100.0 %	
		157	220	377	
Total	% within Child weight groups	41.6 %	58.4 %	100.0 %	

Table 4: Neonatal Congenital Heart Disease with Maternal Age

Mother Age Groups		Congenital Heart Disease		Total	P-Value
		Yes	No		
		50	68	118	0.343
Less than 35 year	42.4%	57.6%	100.0%	100.0%	
3.5 to 4.5 kg 36 to 45 years	80	125	205	137	
	39.0%	61.0%	100.0%	100.0%	
Greater than 4.5 kg	27	27	54	29	
	50.0%	50.0%	100.0%	100.0%	
Total	157	220	377	377	
	41.6%	58.4%	100.0%	100.0%	

DISCUSSION

The increased rate of inherent heart disease in Down's disorder is notable, and numerous creators have distributed figures on the recurrence with which inborn heart diseases are found. These figures shift from 40 to 62. In this study, the overall incidence of congenital heart defects among children with age 1-10 years with Down syndrome was 41.6% of children. Our results coincided with other international data which showed that he t prevalence of congenital heart disease in 40-50% of Down syndrome patients 40-50% of newborns with Down's syndrome have some form of congenital heart diseases.[2,3,14] Another study done in July 2011 in the n Peshawar district showed a higher prevalence of Congenital defect areas found in 31 out of 55 down syndrome patients (56.36%).[15] Palandi et al. found the incidence of CHD in 56% (atrioventricular septal defect (AVSD) 44%, ventricular septal defect (VSD=48%), and the remainder having other heart defects).[16] In our study, we focused used on common defects type of CHD. However other studies reported VSD the most prevalent type accounting for 22.6%, 33.3% in Saudi Arabia, and 43.6% of Chinese patients.[15,17,18] Ashraf et al likewise revealed VSD (48%) as the most well-known kind of CHD in their examination led in Kashmir.[19] In Guatemala, PDA was even announced as the most well-known cardiovascular defect in 29oof f % of cases.[14] The different explanations behind this distinction may incorporate the hereditary makeup of every country and the particular embryology is system sense ASD was found in Mexico in 38% and Saudi Arabia in 21% of patients with DS.[20] The tetralogy of Fallot was in 3.2oof f % of cases observed by Khan et al Increased maternal age is a well-known risk factor for the incidence of Down syndrome However, for the incidence of congenital heart disease in patients with Down syndrome, maternal want non-significantly associated. And the study regarding this is scanty.[15] Congenital heart disease did not show any significant association between the age or the children sex of the child in our study. We found the mean age was 5.96 ±1.analyseesle were 176 (46.7%) while females were 201 (53.3%). In the age group with age 1-5 years 72 (45.9%) were with congenital heart disease while 85 (54.1%) were not having congenital heart disease in the age group 0-10 years 85 (38.6%) were having congenital heart disease while 135 (61.4%) were having CHD. In the case of gender, 77 (43.8%) male children were having congenital heart disease while 99 (56.3%) males were having no congenital heart disease. In females, 80 (39.8%) were with congenital heart disease while 121 (60.2%) with no CHD. In both cases, the p-value was greater than 0.05. Khan et al found that 35 (64%) were males and 20 (36%) were females and among the affected children, 19 were males (61.3%) and 12 were females (38.7%).[15] He also did not find any association.

LIMITATIONS

There is some limitation to our study as there was no

population-based study and is only from a single focus so the value may be not accurate. Also, the cytogenetic investigations were not performed and the conclusion was primarily founded on clinical grounds. Furthermore, we couldn't remark on the recurrence of CHD in various chromosomal changes of Down syndrome.

CONCLUSION

Congenital heart disease is very common (41.6%) in patients with Down syndrome. It is recommended that at the time of diagnosis of this disease with maternal age more than 35 years with clinical signs and symptoms, the patient should be screened for congenital heart disease.

CONFLICT OF INTEREST: None

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