ORIGINAL ARTICLE

FREQUENCY AND PATTERN OF CONGENITAL HEART DEFECTS IN CHILDREN WITH DOWN'S SYNDROME

Inayatullah Khan, Taj Muhammad

Department of Pediatrics, Postgraduate Medical Institute, Lady Reading Hospital, Peshawar, Pakistan

ABSTRACT

Background: Patients with Down's syndrome are prone to have congenital heart defects. This study was conducted to evaluate the frequency of various congenital heart defects in children with Down's syndrome in Khyber Pukhtunkhwa province.

Material & Methods: This descriptive study was conducted at Department of Pediatrics, Lady Reading Hospital Peshawar, from July 2011 to June 2012. Fifty-five phenotypically Down's syndrome children born in Khyber Pukhtunkhwa province, age 2 days to 13 years were included in this study. After detailed history and physical examination, all these patients were subjected to 2-dimentional echocardiography in addition to routine laboratory investigations.

Results: Congenital heart defects were found in 31 out of 55 patients (56.36%). Among the affected patients, 19(61.3%) were males and 12(38.7%) females with male to female ratio of 1.5:1. Ventricular septal defect was the commonest defect (22.6%), followed by patent ductus arteriosus (19.4%), atrioventricular septal defect (19.4%), atrial septal defect (16.1%) and tetralogy of Fallots (6.4%).

Conclusion: Congenital heart defects are common in children with Down's syndrome. The commonest one is ventricular septal defect in our set-up. All children with Down's syndrome should have a cardiac evaluation at birth.

Key Words: Down's syndrome, Congenital heart disease, Ventricular septal defect.

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INTRODUCTION

Down's syndrome (DS) is the most common genetic cause of moderate mental retardation. The incidence of DS in live births is approximately 1 in 733. Affected individuals are prone to have congenital heart defects such as atrioventricular septal defects, ventricular septal defects, isolated secundum atrial septal defect, patent ductus arteriosus, and tetralogy of Fallots.[1]

Few decades ago, the frequency of CHD was underestimated (approximately 20%), but in the last 2 decades the reported frequency has increased to approximately 50%.[2,3] This apparent increase is due to improvement in the diagnostic tools[2] and probably in the physicians attitude towards DS patients. The most common CHD in the western literature is atrioventricular septal defect (AV canal).[1]

Corresponding Author:

Dr. Inayatullah Khan
Department of Pediatrics
Postgraduate Medical Institute, Lady Reading Hospital, Peshawar, Pakistan
e-mail: kinayat9011@yahoo.com

This study was conducted to evaluate the frequency of various types of congenital heart defects in Down's syndrome children in Khyber Pukhtunkhwa Province.

MATERIAL AND METHODS

This hospital-based descriptive study was conducted in the Department of Pediatrics, Postgraduate Medical Institute, Lady Reading Hospital Peshawar. Fifty-five Down syndrome children aged between 2 days to 13 years, diagnosed on phenotypic appearance were randomly selected, over a period of one year, from July 2011 to June 2012, irrespective of presence or absence of any symptom, sign, x-ray chest or ECG abnormality to suggest CHD. After detailed history and physical examination, all these children were subjected to 2-dimentional echocardiography in addition to routine laboratory tests like CBC, serum electrolytes, renal function tests, blood sugar, chest x-ray, routine urine examination, ECG, and other relevant investigations. All children were diagnosed as DS on the basis of their phenotypic findings like mongoloid facies, brachycephaly, depressed nasal bridge,

protruding tongue, small low set ears, upward slanted eyes with epicanthic fold, short neck, short and broad hands, transverse single palmar crease, clinodactyly, a large space between toes (sandal gap), hypotonia and delayed milestones.

RESULTS

In 55 patients with Down's syndrome, 35 (64%) were males and 20 (36%) females with male to female ratio of 1.7:1. The youngest was of 2 days and the oldest one 13 years age.

Congenital heart defects were found in 31 out of 55 patients (56.36%). Among the affected children, 19 were males (61.3%) and 12 were females (38.7%) with male to female ratio of 1.5:1.

The most common form of CHD was ventricular septal defect (VSD), found in 7 of the 31 children (22.6%), followed by patent ducdus arteriosus (PDA) and atrioventricular septal defect each in 6 of the 31 children (19.4%) while secundum atrial septal defect (ASD) in 5 (16.1%) and multiple congenital heart defects in 3 (9.7%) children. Other cardiac defects include tetralogy of Fallots (TOF) in 2 (6.4%) and pentology of Fallots (POF) and partial anomalous pulmonary venous drainage (PAPVD) each in I (3.2%). (Table 1)

DISCUSSION

The high incidence of congenital heart disease in Down's syndrome is well known, and many authors have published figures on the frequency with which congenital heart defects are found. These figures vary from 35 to 65 per cent.[4-9] The frequency of CHD in our study 56.36% is quite comparable to these studies.

Table 1: Type of congenital heart defects in children with Down's syndrome

S. No.	Type of cardiac defect	No.	%tage
1.	Ventricular septal defect	7	22.6
2.	Atrioventricular septal defect	6	19.4
3.	Patent ductus arteriosus	6	19.4
4.	Atrial septal defect	5	16.1
5.	Multiple cardiac lesions ASD+PDA (n=2) VSD+PDA (n=1)	3	9.7
6.	Tetralogy of Fallots	2	6.4
7.	Pentology of Fallots	1	3.2
8.	Partial anomalous pulmonary venous drainage	1	3.2
	Total	31	100

The most common type of CHD in this study was VSD (22.6%). This finding is different from the findings in Caucasian and Sudanese children where AV canal defect was reported as the most common type of CHD accounting for 45%[2] and 48%[10] respectively but almost similar to the finding in Saudi Arabian and Chinese patients with DS where VSD was the most common lesion accounting in 33.3% and 43.6% patients respectively.[9,11] Ashraf et al also reported VSD (48%) as the most common type of CHD in their study conducted in Kashmir.[7]

The frequency of VSD in our study is very close to that reported in India (26%) and Italy (25%).[13] Patent ductus arteriosus is the next most common acyanotic CHD (19.4 %) in this study. It is higher than that reported by other workers.[7,9] In Guatemala, PDA was even reported as the most common cardiac lesion found in 29% cases.[14]

The frequency of AV canal defect in DS patients in our study was 19.4%. This is quite lower than that reported by Ali et al (48%),[10] Freeman et al (45%)[2] and Ashraf et al (28%)[7] in their studies. The various reasons for this difference may include the genetic make-up of each nation and the specific embryological mechanisms.

ASD was present in 16.1% cases of our study. It has been reported to occur in only 5% of the DS patients in Europe and Sudan and 8% in the USA patients. ASD of the secundum type has been reported in Mexico in 38% and Saudi Arabia in 21% of patients with DS.[13]

The cardiac defects in DS are commonly single but they may be multiple as well. The isolated cardiac lesions represented 90.3% of all CHD in our study, compared with 65% in Libya, 80% in Guatemala, 74% in Mexico and 78% in Turkey.[13] This difference may be because of age at diagnosis where patients with more complex lesions die earlier before diagnosis. Multiple cardiac defects were present in 9.7% cases in our study. We found PDA as the most common cardiac defect associated with other lesions. This finding is similar to that of Elmagrpy et al.[13]

The striking feature of our study was the presence of partial anomalous pulmonary venous return and pentology of Fallots in 3.2% cases each which is present just as case reports so for.[15-17]

The most common cyanotic CHDs in this study was TOF occurring in 6.4%. This frequency is slightly higher than that in Caucasian (4%),[2] and Saudi Arabian children (5.3%)[9] but relatively similar to that reported by other workers (6%),[11] (6.2%).[12]

There are some limitations in our study. The figures reported herein are not population based and of only one centre. Also the cytogenetic studies were not performed and the diagnosis was mainly based on clinical grounds. As a result we could not comment on the frequency of CHD in different chromosomal alterations of DS.

CONCLUSION

Congenital heart defects are common in children with Down's syndrome. The commonest congenital heart disease in Down's syndrome is ventricular septal defect in our set-up. Down's syndrome patients should be screened by echocardiography early in life. All children with Down's syndrome should have a cardiac evaluation at birth.

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CONFLICT OF INTEREST
Authors declare no conflict of interest.
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