 DOI:https://doi.org/10.52206/jsmc.2023.13.1.752 OPEN ACCESS

Spectrum of Congenital Heart Disease in Children with Down's Syndrome

Sijad ur Rehman,[1] Wahid Ali,[2] Abid Iqbal,[2] Romana Bibi[3]

ABSTRACT

Background: Down syndrome is the most common chromosomal disorder and most common genetic cause of mental retardation. Congenital heart disease (CHD) is an important cause of morbidity and mortality in children with Down's syndrome.
Objective: To determine the frequency and types of congenital heart diseases in children with Down syndrome and to relate it with consanguinity.
Material and Methods: This Cross-sectional study was carried out at Bacha Khan Medical Complex / Gajju Khan Medical College, Swabi, from January 2019-December 2020. Children less than 12 year's age with Down syndrome were enrolled in the study. Echocardiography of all these children was done and presence and type of CHD was noted.
Results: Our study included 95 children with diagnosis of Down syndrome. The mean age was 56.12±41.89 months. 40% were males. Mother's mean age was 33.04±4.2 years. Mothers having age = 35 years old were 30.5%. Parents having consanguineous marriage were 33.7%. The diagnosis of major congenital heart defect was in 37.9%. Out of 36 CHD patients the type of congenital heart lesion was as follows: 36.11% had AVSD, 25% had ASD; 30.5% had VSD; 5.55% had tetralogy of Fallot and 2.78% had PDA. Children with down syndrome with CHD had older mothers <0.05. Mothers with consanguineous marriages had a greater proportion of CHD children; p<0.05.
Conclusion: Approximately 1/3rd Down syndrome children had congenital heart disease. The commonest lesion was AVSD followed by VSD, ASD, tetralogy of Fallot and PDA. Consanguineous marriages are associated with a greater frequency of CHD in Down children.
Keywords: Down syndrome, congenital heart disease, Echocardiography.

This article may be cited as: Rehman S, Ali W, Iqbal A, Bibi R. Spectrum of Congenital Heart Disease in Children with Down's Syndrome. J Saidu Med Coll Swat 2023;13(1):19-23.

INTRODUCTION

Down syndrome is the most common chromosomal abnormality, with an incidence of 1/600 to 1000 births reported.[1] The existence of an extra copy of chromosome 21 in this condition, which is a hallmark of Down syndrome and trisomy or Robertson translocation, is determined by a chromosomal analysis test. The most prevalent abnormality, trisomy 21, is caused by the nondisjunction of chromosomes; however, translocation and mosaic are also present.[2] Most of the patients with Down syndrome are having congenital heart disease. Patients with Down syndrome having congenital heart disease are at more risk of mortality.[3-6] Congenital heart disease frequency varies between populations but it is necessary that patients with Down syndrome must be evaluated for congenital heart disease. A total of 130 individuals having different ages from 0-33 years were included in a study at Saudi Arabia, about 86.8% patients were found having CHD and 47.8% of patients had PDA, the most frequent heart abnormality.[7]

1. Department of Pediatrics/GKMC/BKMC/MTI/Swabi.
2. Department of Pediatrics/HBS Medical College Islamabad.
3. Postgraduate Resident Gynea/Obs/KTH Peshawar.

Correspondence: Dr. Romana Bibi
Postgraduate Resident Gynea/Obs/KTH Peshawar.
Email: romanawazir14@gmail.com

Received: September 05th, 2022 Accepted: February 13th, 2023

Study conducted again in KSA which included 302 pediatric patients having Down syndrome showed that 58.6% had congenital heart defects with predominantly AVSD in 40.7%.[8]

Echocardiography is the method of choice for identifying congenital cardiac disease in children with Down syndrome.[9-12] In a study the high frequency of AVSD in children with Down syndrome implied that early screening for CHD by Echo is crucial. The correction of AVSD in Pediatric patients should be performed in the 6 months old to avoid irreversible hemodynamic consequences of defects.[13] Early intervention is important because Down's syndrome mortality is highest in people with birth defects.[14-16]

Keeping in mind the mortality and the morbidity from Down syndrome patients who have congenital cardiac disease, this research has been planned to determine frequency and types of congenital heart disease in children with Down syndrome in our region of the world. Our study will establish the frequency and types of congenital heart disease in patients with Down's syndrome in local settings and by knowing the frequency and types of CHD in Down's syndrome our Pediatricians will be able to prioritize the screening of CHD in Down syndrome and initiate appropriate management timely.

Due to the low socioeconomic status of our local population and the lack of availability of

echocardiography facility at every centre, the goal of this study is to identify the most common congenital heart disease in children with down syndrome and to treat them promptly empirically in order to reduce mortality and morbidity.

MATERIAL AND METHODS

This cross sectional study was conducted on the indoor patients of pediatric department of Bacha Khan Medical Complex, Swabi. The data was collected from January 2019-December 2020 with ethical approval Admin/GKMC/ERB/34-21 on 02/01/2021. Total sample size was 95 patients using WHO sample size calculator with clinically diagnosed down syndrome children=297, population proportion=50%, confidence interval=90% and margin of error=7%, were included in this study that fulfilled the inclusion criteria. We used Non-probability consecutive sampling techniques. Children up to 12 years of age both male and female diagnosed clinically as Down's syndrome fulfilling Halls Criteria (Table-1) were included in the study. Those having life threatening congenital malformation (duodenal atresia, trachea-esophageal fistula, duodenal stenosis, esophageal atresia, Hirshprung's disease, annular pancreas, cervical instability, Down syndrome with hypothyroidism and Down syndrome with diabetes were excluded clinically and radiologically.

Table 01. Halls Criteria

1	Flat face
2	Hypotonia
3	Upward slanted palpebral fissure
4	Poor Moro Reflex
5	Short neck redundant skin
6	Small dysplastic ears
7	Single transverse palmer crease
8	Joints hyperflexibility
9	Small 5Th finger with clinodactyly
10	Pelvic dysplasia

Data Collection Procedures:

Following a discussion with the parents regarding the significance and advantages of the study and procedure, their informed consent was obtained.

Clinically diagnosed Down syndrome children's of all ages admitted to Bacha Khan Medical Complex and Gajju Khan Medical Complex were assessed. A detailed history of chief complaints that is breathing difficulty, cyanosis, poor feeding, recurrent chest infection, failure to thrive and family history of congenital heart disease or Down syndrome were asked.

First of all the baseline characteristics of patients were noted. Maternal age of every child with Down syndrome was recorded. Echocardiography of all these children was done by a consultant cardiologist in GKMC/BKMC hospital to find out congenital heart disease. All children with Down with suspected to have CHD were referred to Armed Force Institute of Cardiology, Rawalpindi for a Echocardiography. And after receiving Echocardiography reports of these children, frequency of CHD and its types was finalized on the basis of a report of Echo done by Pediatrics cardiologist at Armed |forces Institute of Cardiology (AFIC). All the information and other relevant data that is name, age, sex, address, date of admission, maternal age, consanguinity, echocardiography findings was entered into a proforma specifically designed for the purpose of this study.

Data Analysis:

All the data was processed and analyzed by computer program SPSS version 20.0. Descriptive statistics were used to calculate mean, standard deviation and ranges from all the continuous variables like age, weight, maternal age. Frequency and percentages were calculated for all categorical variables like consanguinity, congenital heart disease and its types. The results were described and also presented in the form of graph.

RESULTS

Our study included 95 children with a diagnosis of Down syndrome. The age of the children ranged from 2 to 144 months with a mean age of 56.12±41.89 months. The median and mode ages were 44 and 24 months respectively. Before the age of two years, 31 (32.6%) were evaluated. There were 38 (40%) males and 57 (60%) females.

The age of the mothers ranged from 25 to 45 years with a mean age of 33.04±4.2 years. The median and mode ages were 32 and 32 years respectively. 29 (30.5%) of the mothers were = 35 years old. 32 (33.7%) had a history of

consanguineous marriage of parents. The diagnosis of major congenital heart defect was present in 36 (37.9%) of the sample.

Out of these 36 children the type of CHD was as follows:

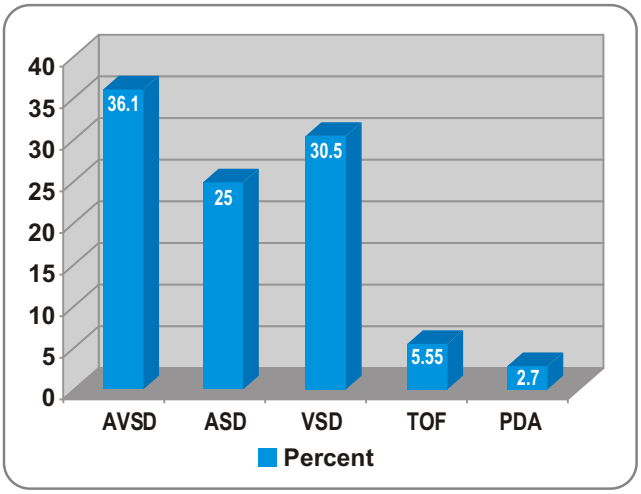


Figure 01. Type of CHD

AVSD Atrio-ventricular and ventricular septal defects
VSD Ventricular septal defect
ASD Atrial septal defect
PDA Patent ductus arteriosus
TOF Tetralogy of Fallot

6 (16.6%) underwent correction surgery prior to data collection. The mean maternal age at the time of birth of the children with congenital heart defect was 35.38±4.8 years and that of Down children without CHD was 31.61±3.05 years. This difference was statistically significant; p<0.05. Hence higher maternal age is not only a risk factor for Down syndrome but these babies are at a greater risk of developing congenital heart defects as well.

DISCUSSION

This study determine the frequency and types of heart disease are congenital in children with Down syndrome. Our study included 95 children with a diagnosis of Down syndrome. The mean age was 56.12±41.89 months. Males were 4%. The mean age of the mothers was 33.04±4.2 years. 29 (30.5%) of the mothers were = 35 years old. In 36 (37.9%) of the sample, there was a diagnosis of major congenital heart disease. Out of 36 CHD patients the type of lesion was as follows: 36.11% had AVSD, 25% had ASD; 30.5% had VSD; 5.55% had tetralogy of Fallot and 2.78% had PDA. Down children with CHD had older mothers p=<0.05. High prevalence of coronary heart disease in Down syndrome children is a famous fact. Based on previous document, the recurrence of coronary artery disease in DS decreased from 35-65%.[9-13] The frequency of Coronary Heart Disease in our study is 37.9% and the highest well-known kind of congenital heart disease found in our study to be an AVSD followed by VSD with recurrence rates of 36.1% and 30.5% separately. Our study was only in line with the study conducted in West of Iran, in 95 patients, there was evidence of familial consanguinity (57.2%). At delivery, the he mean standard deviation (SD) of maternal age was 26.33 (4.7) years (range 15-45 years).[14]

A cardiac abnormality was found in 123 (74.1%) of the 166 patients investigated. The most common single defect was ventricular septal defect (VSD), which was seen in 32 (26%) of the patients, followed by atrial septal defect (ASD) in 22 (17.8%). A total of 74 patients saw a significant improvement in their symptoms. In addition, seven patient (10.2%) died.[15] Our study was found only in agreement with the Sudanese and Lebanese. There, AVSD was most commonly found with a frequency of 48% and 81.7%, respectively.[16]

Our results show that ASD is the most common form of CHD and differs from the results of studies in Oman, Portugal and Libya, where the incidence is 33.3%, 51.8% and 23%, respectively. The second most frequent type of congenital heart disease in this study was AVSD, with frequencies of 27.7%, 46.4%, and 14% respectively.[17-20] Another study that reported that the most prevalent form of CHD was ASD came from South Korea with an incidence of 30.5%, followed by VSD with an incidence of 19.3%.[21] While several studies have reported that PDA is the most common form of congenital heart disease in DS, it originates in Guatemala and Saudi Arabia with a frequency of 28.6% and 47.8% respectively.[22,23]

Recent report from Singapore stated that VSD is the maximum subtype of Congenital Heart Disease of their study observed with PDA, ASD and AVSD with the frequency of 39.2%, 34.5%, 23.4% and 15.6% respectively.[24] In Afghanistan, a comparable study was undertaken the two most prevalent isolated defects were ventricular septal defect (23%) and atrial septal defect (16.4%). VSD and ASD were the most common multiple CHDs, accounting for 19.9% of all cases. VSD + ASD (19.9%) and VSD + PDA (19.9%) were the

most common CHD connections (9%).[25] In a study conducted in Pakistan report that congenital heart diseases were significantly higher in Down syndrome 59% due to consanguineous marriages and advance maternal age during pregnancy. The commonest Congenital Heart Disease in Down syndrome children were CAVC and ASD.[26]

In spite of the fact that our example remembered all patients with Down condition for youngsters' medical clinic GKMC/BKMC during the period covered by the investigation, our outcomes should be circumspectly examined on the grounds that we had a small sample size, because of the lack of public information and the significance of the subject, the commonest heart disease found in our study, just because of detailed examinations, show the significance of careful development and examination of patients with Down syndrome.

CONCLUSION

Approximately one third of the Down syndrome children had congenital heart disease. The commonest lesion was AVSD followed by VSD, ASD, tetralogy of Fallot and PDA. Our study concluded that the occurrence of congenital coronary heart defects in Down syndrome youngsters in our location is much like the charges located via way of means of different authors; therefore, we highlight the significance of diagnostic suspicion and early referral via way of means of pediatricians for cardiac evaluation.

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DATA SHARING STATEMENT: The data that support the findings of this study are available on request from the corresponding author. The data are not publicly available due to privacy or ethical restrictions.

CONFLICT OF INTEREST: Authors declared no conflict of interest.
GRANTED SUPPORT AND FINANCIAL DISCLOSURE: Nil

AUTHOR'S CONTRIBUTION
The following authors full fill authorship criteria as per ICMJE guidelines;

Rehman S:	Idea conception, data acquisition, final approval, agreed to be accountable for all the work.
Ali W:	Design of the work, drafting the work, critical rivision, final approval, agreed to be accountable for all the work.
Iqbal A:	Data analysis, drafting of the work, final approval, agreed to be accountable for all the work.
Bibi R:	Data interpretation, critical revision, final approval, agreed to be accountable for all the work.