Pattern of congenital heart diseases in children with Down Syndrome

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ABSTRACT

BACKGROUND: Down syndrome is the most common chromosomal disorder and is the most common single cause of mental handicap .Congenital heart disease is the most common anomaly, which affects morbidity and mortality in children with Down syndrome. This study was conducted in district swat to determine pattern and prevalence of Congenital Heart Disease in children with Down syndrome.

OBJECTIVES: To determine the pattern and prevalence of congenital heart diseases in hospitalized children with Down syndrome and to compare clinico-demographic features of "down syndrome" cases with and without congenital heart disease.

MATERIAL AND METHODS: This cross sectional case series was conducted at department of pediatrics, Saidu teaching hospital, Saidu Sharif, Swat for eleven months from April 2015 to March 2016 in 60 children with Down syndrome, pattern of congenital heart disease was determined. They were investigated by echocardiography.

RESULTS: Out of 60 children with Down syndrome, 65% were males and 35% females. Majority 51.7% were from 01 day to 11 months old. Majority 70% patient's mothers were 31-40 years old. Congenital heart disease found in 33.33% cases, with 26.66% cases acyanotic heart disease and 6.67% cases cyanotic heart disease. Out of 33.33% cases with congenital heart disease, ventricular septal defect was found in 45% cases. Atrio-venticular canal defect in 25% cases, Tetralogy of fallot in 15% cases, atrial septal defect in 10% cases, Transposition of the great arteries with ventricular septal defectin 5% cases.

CONCLUSIONS: Congenital heart disease is a common finding in Down syndrome. Congenital heart disease is more common in male Down syndrome cases. Acyanotic heart disease outnumbered the frequency of cyanotic heart disease.

***Key Words**: Down syndrome; congenital heart disease; acyanotic; cyanotic; echocardiography*

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INTRODUCTION

Down syndrome is the most common single cause of mental handicap occurring in approximately 1 in 700 of the births in all populations, however; the incidence varies with the age of the mother. The incidence for mothers aged 25 years is one in 1400 and increases to reach an incidence of 1 in 46 for mothers' aged 45 years.[1]Prenatal studies can therefore be utilized to decrease the incidence of their chromosomal abnormality.[2]

Consanguinity, antibiotics use in pregnancy, oral contraceptive use and diabetes in the mother are independently associated with increased risk of congenital heart disease among down syndrome.[3]

Their presence remains a significant predictor of mortality in patients with down syndrome.[4] The most common cause of morbidity and mortality in patients with down syndrome are congenital heart defects in 40 to 50 percent of cases.[5] Congenital cardiac disease is the greatest cause of death in patients with down syndrome during the first two years of life.[6]

The most common congenital heart disease in Down syndrome is ventricular septal defect. It was suggested that Down syndrome patients should be screened by echocardiography early in life to avoid obstructive pulmonary vascular disease. The mortality in Down syndrome is highest among those with congenital anomalies, and therefore, early intervention is crucial.[7] Congenital heart disease was documented in 103 (34.9%) patients and ventricular septal defect was the most common cardiac anomaly.[8]

In Pakistan, each year 12,000 children are born with congenital heart disease. Almost 90% of these children either die or are diagnosed so late that even surgery is ineffectual.[9]Complete atrioventricular septal defect occurs in about 20% of individuals with down syndrome, an approximately 500-fold increase in risk as compared to individuals without down syndrome.[10]

Echocardiography has become the method of choice in the diagnosis of a congenital heart with Down syndrome. The most compelling argument for diagnosis of congenital heart defects in the neonatal period is the need for early surgical intervention (ideally prior to 6 months of age) in those with complete atrioventricular canal.[11]

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Factors influencing the survival among persons with Down syndrome are not well understood. To evaluate survival of infants with Down syndrome and potential prognostic factors a population-based study Results showed that survival probability to 1 year was 92.9% and to 10 years was 88.6%. Univariate analysis demonstrated that black maternal race, low birth weight, preterm birth, lower paternal education, presence of heart defects, and presence of other major congenital anomalies were important prognostic factors.[12]

Keeping in view the morbidity and mortality from congenital heart diseases in patients with Down syndrome this study has been designed to determine the frequency of congenital heart disease in hospitalized children with Down syndrome.

MATERIAL AND METHODS:

This cross sectional case series was conducted at department of pediatrics, Saidu teaching hospital, swat from April 2015 to March 2016. In 60 children, both male and female with Down syndrome, pattern of congenital heart disease was determined. They were investigated by echocardiography. Patients with dysmorphic features other than Down syndrome were excluded in this study.

All children clinically diagnosed with Down syndrome admitted in the department of pediatrics, an informed consent was taken detailed history of present complaints including, recurrent chest infections, breathing difficulty, cyanosis, failure to thrive or poor feeding. Family history of any congenital heart disease or Down syndrome was taken. Maternal age of every child was recorded. Physical examination of every child was done to look for mongoloid faces, depressed nasal bridge, protruding tongue, upward slanted eyes, epicanthic folds, short neck, short and broad hands and transverse single palmer crease, central and peripheral cyanosis, breathing difficulties, failure to thrive or poor feeding, clubbing and any murmur.

All these children were referred to cardiology department for echocardiography to find out any congenital heart disease. After frequency and pattern of congenital heart disease was determined among these Down syndrome patients.

All the study variables like demographic features of patients, age, sex, maternal age of child, echocardiography findings were analyzed for descriptive statistics. Frequencies and percentages were determined for pattern of congenital heart diseases detected. Mean, $\pm$ standard deviation was determined for age and maternal age and ratio was calculated for gender distribution. All the data was processed by computer program spss version 20 and was presented in tables and graphs.

RESULTS

60 cases of Down's syndrome were admitted during the study period and there were 39 (65%) males and 21 (35%) females with male to female ratio of 1.85: 1.

Most of the patients in this study, 31 (51.7%) were in the age group of from 01 day to 11 months old, followed by 15 (25%) in the age group of 1-5 years, 9 (15%) in age group of 6-11 years and 5 (8.3%) in the age group of 11-15 years. Minimum age was 01 day and maximum was 14 years with mean age of 5.4267 $\pm$ 3.5740.

Family history of patients showed that any congenital disease was positive in 9 (15%) cases. Down syndrome was in the families of 10 (16.7%) cases.

Echocardiography finding showed that 20 (33.33%) cases were having congenital heart diseases. Among these 16 (26.66%) cases were acyanotic heart disease, and 4 (6.67%) cases were cyanotic heart disease, which make an overall frequency of 20 (33.33%) cases (table no. 1).

Pattern of congenital heart disease in children with down syndrome showed that out of 20 cases with congenital heart disease, ventricular septal defect was recorded in majority 09 (45%) cases. Atrioventricular canal defect was found in 05 (25%) cases, Tetrology of Fallot was observed in 03 (15%) cases, atrial septal defect was found in 02 (10%) cases, transposition of the great arteries with ventricular septal defect was recorded in 01 (5%) cases (table no. 2).

DISCUSSION

Congenital heart disease is present in 40-50% of individuals with Down syndrome. The lesions within the heart can be single or multiple. The type of congenital heart disease in patients with Down syndrome is known to vary according to geographical location. Atrio-ventricular canal

defect is the most common single congenital cardiac malformation in most countries and especially in the United States of America and Europe. The most common lesion is a single ventricle defect in Asia and an atrial septal defect in Latin America. Multiple defects are much rare. Fallot tetralogy is one of the multiple defects in Down syndrome patients and the frequency may be 0-15.5% in different series. The fallot tetralogy has been reported to exist together with an atrioventricular canal defect in down syndrome.[6, 7, 16, 17]

A retrospective review of 98 Down syndrome patients , results of the study showed that out of 93 patients congenital heart disease were found in 57 (61.3%) patients. ventricular septal defect was the most common (33.3%) followed by atrioventricular septal defect (22.8%), atrial septal defect (21.1%), patent ductus arteriosus (14%) and tetralogy of fallot (5.3%). Three patients (5.3%) developed inoperable obstructive pulmonary vascular disease (opvd) and 3 were deemed inoperable for other reasons. The congenital heart disease was clinically suspected in 96%.[7] In our study ventricular septal defect was most common 45% ,atrioventricular canal defect 25%, tetralogy of fallots 10%, atrial septal defect 15% , transposition of great arteries with ventricular septal defect 5%.

A study analysis included cases of Down syndrome presenting over a period of 7.5 years; a total of 524 patients were studied. There were 303 males and 221 females; with male to female ratio of 1.37:1. Average age at presentation was 19.4 months (range: 1 day-26 years). Average maternal age at birth of the affected child was 26.8 years (range: 16-45 years). The most common cardiac anomalies were ventricular septal defect (25.8%), Tetrology of Fallot (15.5%), and atrial septal defect (12.1%). Congenital heart disease was present in 18.3% of cases, with ventricular septal defect being the most common type of defect. Non-disjunction was the most common cause of the chromosomal anomaly.[18]In another study, one-half (51.7%) of the studied children had congenital heart problems[20]. In a recent study out of 94 patients, overall, 37 patients (39.3%) had a congenital heart defect[21]. In our study we have found that overall congenital heart diseases were presented in 33.33% cases of down syndrome, which is less than as reported in other studies[19,20] and comparable to a study in which congenital heart diseases were noted in 39.3% patients.[21] since all down syndrome patients in this study were subjected to echocardiography evaluation even in the absence of any symptom, physical sign, ECG or chest x-ray abnormality to suggest congenital heart diseases, we believe this is a true representation of the frequency of congenital heart disease in children with Down Syndrome in this population. The lower frequency of congenital heart disease in Down Syndrome children observed in our study could just reflect on the smaller sample size in our series.

In our study, male children were more affected with Down syndrome than female children with a male:female of 1.85:1 were. This ratio is comparable with a study in which male to female ratio of 1.38:1 has been reported with male predominance[18]. The male predominance in our study could be due to traditional, local customs in this part of the country where females are not given preference for medical treatment even if they die at home and man dominant society do not invest on females.

The average age at presentation in our patients with Down's syndrome was 5.42 years (range 01 day -14 years). Almost same results are also reported in a study.[22]

Inheritance of Down syndrome is still not completely understood. However, earlier workers strongly advocated that the advanced maternal age is a major risk factor for trisomy 21. The likelihood that a woman under 25 and 30 years who becomes pregnant will have a baby with Down syndrome is less than 1 in 1,400 and 1,000 respectively. Chance of having a baby with Down syndrome increases to 1 in 350 for women who become pregnant at the age 35 and continues to increase as the woman ages, so that by age 42, and by age 49, the chance is 1 in 60 and 1 in 12 respectively. On the contrary, there are reports that 80% of Down syndrome babies are born to young women of age less than 30 years.[23]

More than 50% of babies with Down syndrome are born to women of advanced maternal age (> 35 years). The prevalence of Down syndrome pregnancies varies with maternal age. The risk rises from under 1:1000 live births in women under 25 years to approximately 1% at age 40 years and 7% at 48 years. In a study the mothers' ages at the birth of the child with down syndrome ranged from 18 to 45 years, and 21 (42%) were of advance maternal age.[24] In our study advanced maternal

Table no. 1: Various characteristics of children with Down syndrome (n=60)

Characteristics	No. Of patients (%)	Ratio/mean
Gender: Male Female	 39 (65) 21 (35)	 Male to female ratio = 1.85: 1
Age ranges (in years) 01 day to 1 year 01 - 05 years 06 - 10 years 11 – 15 years	 31 (51.7) 15 (25) 09 (15) 05 (8.3)	 Mean age = 5.4267 ± 3.5740
Family history Down syndrome Congenital heart disease	 10 (16.7) 09 (15)	 -
Echocardiography finding of Congenital heart disease Acyanotic Cyanotic	 16 (26.66) 04 (6.67)	 -

Table no. 2: Pattern of congenital heart disease in children with Down syndrome (n=20)

Pattern of congenital heart disease	No. of cases	Percentage (%)
Ventricular septal defect	09	45
Atrioventricular canal defect	05	25
Tetra logy of fallot	03	15
Atrial septal defect	02	10
Transposition of the great arteries with ventricular septal defect	01	05
Total	20	100

age was found in 53.33% cases. so our results are in agreement with the results of the quoted study.[24] while in another study with regard to maternal age, 64% of the mothers were older than 35 years of age, while the remaining 36% of mothers were less than 35 years of age at the time of birth of the affected child. average maternal age at birth of the affected child was 32.3 (range 2150) years.[22]

CONCLUSIONS

Based on results of this study, it is concluded that:

Congenital heart disease is a common finding in Down syndrome.

Congenital heart disease is more common in male Down syndrome cases.

Acyanotic heart disease outnumbered the frequency of cyanotic heart disease.

Advance maternal age mothers were found to be in majority, which is major risk of Down syndrome.

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