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Original Research Article

Study to assess incidence and type of congenital heart defect in children with down syndrome presenting to tertiary care teaching hospital of Southern Rajasthan

Anuradha Sanadhya, Nitesh Meena*, Priya Sharma, Sakshi Setia

Department of Paediatrics, RNT Medical College, Udaipur, Rajasthan, India

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***Correspondence:**
Dr. Nitesh Meena,
E-mail: Niteshmeena420@gmail.com

ABSTRACT

Background: Down syndrome occurs in people of all races and economic levels. Cardiac anomalies with a prevalence of about 50% are the most common anomaly responsible for death during the 1st two years of life in children with down syndrome. Objective of the study was to Assess the incidence and type of congenital heart disease in children with down syndrome presenting to a tertiary care hospital.
Methods: Hospital based descriptive study. Participants: All children (0 to 18 years) diagnosed (clinically or on karyotyping) with Down syndrome, presenting in RNT Medical College, Udaipur Each patient was evaluated with a pre-set Performa which contained detailed sociodemographic profile, maternal and clinical history including age of conception, detailed physical examination to look for features of down syndrome. Degree of Intellectual / Developmental retardation was assessed by Vineland Social Maturity Scale. 2D- echo, karyotyping was done in all patient. CBC, thyroid profile, CXR, USG- abdomen, Invertogram were done as per need.
Results: Out of 57 Down syndrome children, 32(56.1%) were males and 25(43.9%) were females. Congenital heart defect was found in 35 out of 57 Down syndrome children (61.4%). Among 35 cases 82.2% had Acyanotic CHD and 17.1% had Cyanotic CHD. Ventricular Septal defect (31.4%) was the most common Congenital heart defect followed by Atrial Septal defect in 20% cases. Trisomy 21 was observed among 94.7% cases.
Conclusions: More than half of children with Down syndrome have congenital heart disease, mostly Acyanotic, more specifically VSD. Early screening and diagnosis re the key to avoid irreversible hemodynamic consequences.

Keywords: Congenital heart disease, Down syndrome, Chromosomes

INTRODUCTION

Chromosome abnormalities are very common and occur in approximately 1-2% of live births, 5% of stillbirths, and 50% of early foetal losses in the 1st trimester of pregnancy.[1] Down syndrome occurs in people of all races and economic levels.

Trisomy 21 is the most common genetic cause of moderate intellectual disability. The incidence in India is 1 per 850 to 900 live births.[2] Cardiac anomalies with a prevalence of about 50% are the most common anomaly responsible for death during the 1st two years of life in children with down syndrome.[3]

Affected individual are more prone to congenital heart defect such as atrioventricular septal defects, ventricular septal defects, isolated secundum, atrial septal defects, patent ductus arteriosus, and tetralogy of Fallot.[1] Objective of this study was to evaluate clinical profile of

children with down syndrome and assess the incidence and type of congenital heart disease in children with down syndrome.

METHODS

The present study was a hospital based descriptive study conducted in Balchikitsalaya of MBGH, RNT Medical College, Udaipur, Rajasthan over a period of one year (September 2021 to august 2022) after acceptance from institutional ethical committee. All children (0 to 18 years) diagnosed with Down syndrome (clinically or by karyotyping) who presented in our hospital during a period of one year were enrolled in the study.

Fifty-seven children (0 to 18 yrs. of age) whose parents gave written informed consent were included in the study and non-consenting parents and critically ill children were excluded from study.

Each patient was evaluated systematically as per a predesigned proforma. Details were noted about the child's name, age, gender, address, socioeconomic status, demographic profile etc. Further, details were noted about variables including Maternal age at conception, age of father, consanguinity of parents, history of abortions, medication history during pregnancy, birth weight and gestational age of baby etc. Complete head to toe examination of each child was done thoroughly to look for features of down syndrome. Anthropometry of children were recorded according to IAP growth charts. CBC, thyroid profile, CXR, USG- abdomen, Invertogram were done as per need. All the patients were subjected to a predefined set of tests including transthoracic 2D echocardiography to evaluate for the associated congenital heart disease if any. Diagnosis of down's syndrome was confirmed by karyotyping by GTG Banding method with band resolution of 400-500bphs.

The data collected was analysed using version SPSS-20 statistic software.

RESULTS

A total of 57 Down syndrome cases were recruited into the study. In our study, children less than 1 year were 56%, between 1 to 5 years were 35%, children >5-10 years were 3.5% and children >10 years were 5.3%. Mean age of children was 2.32±3.3 years.

Table 1: Distribution of patients according to maternal age.

Mother age groups	Frequency	Percent
Less than 20	1	1.8
20 to 29	35	61.4
30 to 40	20	35.1
More than 40	1	1.8
Total	57	100

The percentage of male patients (56.1%) was higher than females (43.9%) with male: female ratio of 1.28:1 in our study. Maternal age was less than 20 years in 1.8%, between 20 to 30 years in 61.4%, 31 to 40 years in 35.1%, more than 40 years in 1.8% of down syndrome cases. Mean maternal age was 28.11± 5.6 years (Table 1). Paternal age was between 20 to 30 years in 38.6%, 31 to 40 years in 50.9%, more than 40 years in 10.5% of down syndrome cases. Mean paternal age was 31.68±6 years. 91% Cases were born of non-Consanguineous marriage and 8.8% case born of Consanguineous marriage. 17.5% of children were born of the first birth order, 36.8% cases were born of second birth order whereas 45.6% born of the third birth order.

Table 2: frequency of CHD in Down Syndrome.

No. of Down syndrome cases studied	57
No. of cases with CHD	35
Frequency of CHD in Down syndrome	61.40%

In our study majority of cases (45%) were of lower socioeconomic class. 64.9% children were from rural areas and 35% were from urban areas in our study. 28% mothers gave history of previous abortions, 3.5% mother gave history of still birth and 1.7% mother gave history of neonatal death and 1.7% mother gave history of Malpresentation.65% children were born by vaginal delivery and 35% children were born by cesarean section.

Table 3: Pattern of CHD in Down syndrome.

Echocardiography findings	Frequency	Percent
ASD	7	20
ASD, PDA	2	5.7
ASD, VSD	1	2.8
ASD, VSD, PDA	1	2.8
AVSD, CCHD	4	11.4
Cyanotic Heart disease, SD, VSD, TGA	1	2.8
MILD TR, normal	1	2.8
Moderate MR	1	2.8
PDA	1	2.8
PFO	1	2.8
Small PFO	1	2.8
TOF	1	2.8
VSD	11	31.4
VSD, PDA	1	2.8
VSD, PFO	1	2.8
Total	35	100

In our study weight for height was < 3SD in majority of children (41.3%), 21.7% were between -2SD to >-3SD, 15.2% were between -1SD to >-2SD, 21.7% were between >-1SD to normal. Weight for age was <-3SD in 67% of cases. 70% cases were having microcephaly (HC<-3SD).

Congenital heart defects were noted in 61.4% of cases (Table2). Among these 82.2% had Acyanotic CHD and 17.1% had Cyanotic CHD. Ventricular Septal Defect (VSD, 31.4%) was the most common cardiac abnormality followed by Atrial septal defect (ASD, 20%). The most frequent associations of CHD were ASD with PDA (5.7%), ASD+VSD+PDA (2.8%), ASD+VSD (2.8%), VSD+PDA (2.8%), VSD+PFO (2.8%) (Table3). Details of echocardiographic finding are given in in Table 2. Distribution of VSD according to size has been mentioned in Table 3. In majority of patients (56.2%) small VSD (0-5mm) was noted.

Out of total 42 admitted patients ,4 cases died during the course of hospital stay. [9]Cause of death was pneumonia in 2 patients, neonatal sepsis in 1 patient, congenital heart disease leading to CHF in 1 patient.

DISCUSSION

We observed marginally higher incidence of Down syndrome in male than female children with a male-female ratio of 1.28:1. This could be explained due to predominance of male children in indoor set up. In a similar study by Kava MP et al male-female ratio was 1.37:1.[4]

In the present study maximum mothers fall in the age group of 20-30 years because of the tendency of early marriage and early childbirth in this area. Mean maternal age was 28.11 years, which was a bit higher than mean maternal age of overall pregnancies. Kava MP et al found that the majority of children with DS were born to women in the 20- to 30-years-of-age group which is similar with study.[4] In our study shown maximum percent of babies with Down syndrome (51%) were born to fathers aged between 30-40 years with mean paternal age 31.6 years. There is no proven evidence that down syndrome increases with paternal age. Studies by Maria I et al give no evidence that paternal age can be considered a risk factor for the conception of a child with Down Syndrome.[5]

In our present study it was seen that out of all cases 91.3% were born out of nonconsanguineous marriage and in only 8.8% cases consanguinity was present, this study revealed that larger number of cases were from non-consanguineous marriage because in this part of country most of the population belongs to the Hindu communities and consanguinity is not prevalent. In a study by Venugopalan et al in Oman, it was found that in high consanguineous areas the incidence of CHD in Downs' is more likely, as in Turkey it is 65% and in Oman it is 60%. Because consanguineous marriage is prevalent in this area.[6]

As the birth order of the child increases, the chances of being born with a CHD also increases. In our present study about 45.6% of Down syndrome born of third order. In study by Muthumani, P et al 44.8% (n=39) children were first born, 39.1% (n=34) were second born, 12.6% (n=11) were third born and 3.4% (n=3) were fourth born. The result was irreconcilable with our study.[7]

In our study only 3.5% cases belonged to upper socioeconomic class, may be because we have taken cases from the government hospital where most of patients comes from rural areas and there is possibility that children who belonged to upper class were admitted to private hospitals considering the affordability. In our study 50.8% mothers had bad obstetric history, 28% had history of abortion, 17% had history of Malpresentation, 3.5% had history of still birth and 1.7% had history of Still birth. In the study Lippman A et al. also observed an increased abortion rate in women less than 25 years. Hence these women form a high-risk group and may need prenatal screening.[8]

In our study 61.4% of children with Down syndrome had congenital heart defect. Many other studies show CHD rate 40-50%. Higher percentage of CHD in our study could be explained by the fact that children we included in this study were mostly symptomatic and visited to hospital for some ailments, and were nor from general population of Down syndrome. In study by Somasundram A et al 49.1% of children were having congenital heart disease which is lower than our study. In study by Kumar G. V et al the percentage of CHD in down syndrome was 45% which is lower than our study.[10] In the study by Shrestha M 80% of children were having congenital heart disease which is higher than our study.[11]

In our study 82.2% had Acyanotic CHD and 17.1% had Cyanotic CHD. Ventricular Septal defect is the most common defect identified in 31.4% of cases with CHD followed by ASD in 20% cases followed by AVSD associated CCHD in 11.4% of cases followed by ASD with PDA in5.7%% of cases followed by PDA in 2.8% cases.

PFO present in 2.8%cases, small PFO present in 2.8% cases. ASD with PDA present in 2.8% cases, ASD with VSD present in 2.8% cases, ASD with PDA and VSD are present in 2.8% cases, CCHD with VSD with TGA present in 2.8% cases, VSD with PDA present in 2.8% cases, VSD with PFO present in 2.8% cases, Mild TR present in 2.8% cases, Moderate MR present in 2.8% cases, Tetralogy of Fallot was found in 2.8% cases. In our present study Ventricular septal defect (VSD) was the most common abnormality accounting for 31.4% of CHD followed by ASD. Even though most studies point to Atrioventricular septal defect (AVSD) as the most common abnormality, there are studies depicting VSD as the most common defect. Higher percentage of VSD in our study could be explained by exclusion of critically sick children in study. Similar observations were observed by Somasundram A et al.[9] Ventricular Septal defect (34.5%) is the most common defect followed by Endocardial cushion defect (21.8%) followed by Atrial Septal defect (20%) of cases. The results were consistent

with our study with VSD as the most common CHD. Similar findings were observed by Kumar GV et al Ventricular septal defect (40%) was the most common CHD followed by Endocardial cushion defect was seen in (24.4%) children, (17.7%) children had atrial septal defect.[11] Patent ductus arteriosus and tetralogy of Fallot was seen in (11.1%) children and (6.6%) children respectively.

In the study by Shrestha M et al observed that out of 40 patients the most common single defect was ventricular septal defect (VSD), which was found in 9(22.5%) patients, which is similar to our study followed by atrioventricular septal defect (AVSD) in 6 (15%) patients, atrial septal defect (ASD) and patent ductus arteriosus (PDA) each in4 (10%) patients.[11] Among the patients with multiple cardiac lesions, PDA was the most frequent associated lesions, which was found in 6 (15%) patients followed by ASD in 5 (12.5%) patients. In the study by Asim A et al observed that AVSD (50%) was most common defect which is dissimilar to our study followed by VSD (26.6%).[12] Other form was also present which includes ASD, PDA and TOF which is (10%), (6%), and (6%) respectively. Benhaourech S et al conducted a study observed that most common defect was atrioventricular septal defect (AVSD) (29%) which is dissimilar to our study followed by ventricular septal defect (VSD, 21.5%) and atrial septal defect (ASD, 19.9%).[13] The most frequent associations of CHD were AVSD +ASD (9.3%), VSD + ASD (6.2%) and VSD + PDA (5.5%).

In the present study Karyotyping was done in 55 patients we found that trisomy 21 was observed in (54) 94.7% cases. One subject had normal karyotyping report, second one report could not be followed up. Detail cause of trisomy i.e. Nondisjunction, translocation and mosaicism could not be explained by our method of karyotyping. Pankaj G et al observed that out of total 2750 clinically suspected cases were referred to diagnostic laboratory for cytogenetic analysis, of which 682 cases were found confirmed DS.[2] The rest 2068 cases were found to be normal and did not show any chromosomal anomalies. Chromosomal non-disjunction was the most common type of abnormality followed by translocation and lastly mosaic 92.2, 7.0 and 0.7% respectively.

Limitations

Study did not include patients who were hemodynamically unstable at presentation and expired. Due to financial reasons, we were unable to offer karyotyping study to few patients, therefore some suspected patients were diagnosed only on clinical basis.

CONCLUSION

Due to high incidence of congenital heart disease in children with Down syndrome, CHD should be ruled out earliest in all children of Down syndrome by universal 2 D echocardiography. Early screening, diagnosis and then appropriate treatment are the key to avoid irreversible hemodynamic consequences, thus can play a major role in decreasing morbidity and mortality in these children.

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