

## **A Study on Patterns of Congenital Heart Diseases in Children with Down syndrome, Attending SPMCHI, SMS Medical College, Jaipur**

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### **Article History**

*Received: 08.09.2018*

*Accepted: 18.09.2018*

*Published: 30.09.2018*

### **DOI:**

10.36348/sjm.2018.v03i09.002



**Abstract:** Down syndrome (DS) is a common chromosomal anomaly. Congenital heart disease (CHD) is main prognostic factor contributing to a favourable or unfavourable outcome in these patients. To study the patterns of congenital heart diseases in children with Down syndrome. Hospital based descriptive type of observational study of 70 patients conducted at SPMCHI, Jaipur from April 2012 to Sept 2013. There were 70 DS children; 46(65.71%) male and 24(34.29%) female. Congenital heart disease was detected in 47(67.14%) DS children. Out of those, 95.74% were detected as acyanotic heart disease and 4.26% were cyanotic heart disease. Total 73 congenital heart defects were detected in 47 DS children with CHD. In these children, 46.81% have single defect, 31.91% have 2 defects and 21.28% have multiple defects. In total congenital heart defects ASD (41.10%) was the commonest congenital heart defect followed by PDA (21.92%) and VSD (15.07%). It is imperative to know about presence of congenital heart disease in Down syndrome children. Contrary to result of most of other workers who observed atrioventricular septal defect was commonest defect. In present study ASD, PDA and VSD were the commonest CHD in order of frequency.

**Keywords:** Down syndrome (DS), Atrial septal defect (ASD), Ventricular septal defect (VSD), Patent ductus arteriosus (PDA), Congenital heart disease (CHD).

## **INTRODUCTION**

Down syndrome is a condition characterized by trisomy of chromosome 21 [1]. Down syndrome is a common disorder that occurs in approximately 1:600 new borns; however, this incidence greatly increases among children born to mothers over 35 years of age [2]. In mothers older than 45 years old, the syndrome can reach an incidence of 1 per 30 births[3]. The diagnosis is basically established by the patient's phenotype, being later confirmed by the karyotype. Congenital heart disease is a main prognostic factor contributing to a favourable or unfavourable outcome in these patients. Cardiac malformations are the principal cause of mortality in the first two years of life. This study was planned to know the pattern of congenital heart disease in children with Down syndrome from Rajasthan part of our country.

## **MATERIAL AND METHODS**

A Hospital based descriptive type of observational study of 70 Down syndrome children confirmed by karyotyping was performed in SPMCHI (Sir padampat mother and child health institute), Department of Pediatric Medicine, Jaipur from April 2012 to September 2013. Cases of Congenital heart disease in Down syndrome children were diagnosed with help of Chest X-ray, Electrocardiography and

Echocardiography. Desired Information was gathered on predesigned and pretested proforma. Data thus collected was entered in Microsoft Excel 2007 worksheet in the form of master-chart. These data was classified and analyzed statistically using XL Stat Software. Quantitative data is summarized in form of Mean±SD. Qualitative data is summarized in form of proportion & difference in proportion is analyzed using Chi-square test. The confidence interval for all the statistical analysis is kept 95% &  $\alpha$  error 5%.

## **RESULTS**

There were 70 Down syndrome(DS) children; out of them 46 (65.71%) were males and 24 (34.29%) were females. Congenital heart disease (CHD) was detected in 47 (67.14%) DS children. Out of these, 95.74% were detected as acyanotic CHDs and 4.26% were cyanotic CHDs. In our study, DS children were born mainly to young mothers (61.43%) of 17 to 27 years of age. In our study single congenital heart defect (46.81%) was most commonly observed type of congenital heart defect in Down syndrome children with CHDs (n= 47). In our study isolated ASD (27.66%) was the most common congenital heart disease followed by ASD with PDA (17.02%) and isolated PFO (8.51%) in Down syndrome children having CHD. In our study ASD (41.10%), PDA 21.92% and VSD 15.07%.

were 3mcongenital type of individual ost common heart defects in Down syndrome children.

Single congenital heart defect was most commonly observed congenital heart defect in Down syndrome children. In our study single syndrome children ) defect 46.81 (% was most commonly observed type of congenital heart defect in Down syndrome children with CHDs (n =47) (Table-1). Similarly Azman *et al.*, [4] in 2007 also found 43 % and Ercan *et al.*, [5] in 2010 found

56.62 % Down syndrome children were having single congenital heart defect. However Figueroa *et al.*, [6] in 2003 found 74.37 (% and Salih *et al.*, [7] in 2011 found 73.3 (% found Down syndrome children were having isolated congenital heart disease. A very high frequency of single congenital heart defect have been reported by Abbag *et al.*, [8] in 2006 who found 91.23 % and Jashami *et al.*, [9] in 2007 found 100 % Down syndrome children were having single congenital heart defect.

**Table-1: Distribution of Down syndrome children with CHDs according to presence of No. of congenital heart defect (n=47)**

No. of congenital heart defect	No. of DS children	%
Single defect	22	46.81
Double defects	15	31.91
Multiple defects	10	21.28
Total	47	100

congenital heart The lower incidence of single defect has been observed in present study. Our study results to be confirmed by further studies with large number of subjects. Why the complex congenital heart disease are becoming more common now a days also and paragenetic factors require a probe into genetic responsible for CHD.

Contrary to normal usual observation, in our study Down syndrome were more common in younger (yr 27-mother. (61.43% in 17

In our study, Down syndrome children were born mainly to young mothers (61.43%) of 17 to 27 years of age) Table-2. Freeman *et al.*, [10] in 1998 also

found Down syndrome children were born mainly during maternal age < 35 year (70.37%). Contrary to the finding of present study, M. M. Mokhtar *et al.*, [11] in 2001 found that children with Down syndrome were given birth mainly by women above 25 years of age (82.61%) and Figueroa *et al.*, [6] in 2003 also found that Children with Down syndrome were given birth mainly by women above 25 years of age (66%). Boas *et al.*, [12] in 2009 also found that maternal age during pregnancy was ≤ 30 years (21.3%). Similarly Azman *et al.*, [4] in 2007 also found 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 21–50) years.

**Table-2: Distribution of Down syndrome children according to maternal age at conception**

conception Maternal age at	No.	%
>17-22 Yr	24	34.29
>22-27 Yr	19	27.14
>27-32 Yr	14	20
>32-37 Yr	9	12.86
>37-42 Yr	4	5.71
Total	70	100

Down syndrome has been found to be associated with different maternal age (s) (by different workers). On reviewing the literature it was noted that studies done before 2000 AD relatively elder mothers were giving birth to Down syndrome children. However in 21st century more workers have reported that Down syndrome children were born even to younger mother. There may be genetic reasons for this or this shift may

be due to the small family norm being practiced more commonly in India due to family welfare programme.

Isolated ASD was the most common. In congenital heart disease followed by ASD with PDA. Isolated ASD (27.66%) was the most common congenital heart disease followed by ASD with PDA (17.02%) and isolated PFO 8.51 (% in Down syndrome children having CHD) Table-3.

**Table-3: Distribution of Down syndrome children according to congenital heart disease**

.S.No	Congenital heart disease	.No	%
1	Isolated ASD	13	27.66
2	ASD with PDA	8	17.02
3	Isolated PFO	4	8.51
4	VSD with ASD	4	8.51
5	Isolated VSD	3	6.38
6	Complete AV canal defect with PDA	2	4.26
7	VSD with PDA ,ASD	2	4.26
8	Isolated PDA	1	2.13
9	Isolated Bicuspid Aortic valve	1	2.13
10	Isolated Complete AV canal defect	1	2.13
11	Isolated incomplete AV canal defect	1	2.13
12	Tetralogy of Fallot	1	2.13
13	Pentalogy of Fallot	1	2.13
14	PFO with VSD	1	2.13
15	VSD with PDA	1	2.13
16	Complete AV canal defect with PFO	1	2.13
17	PDA, ASD with PFO	1	2.13
18	ASD, PDA with Bicuspid aortic valve	1	2.13
TOTAL Down syndrome children with congenital heart disease		47	100

workers have also reported ASD to be the most common CHD in Down syndrome children. Figueroa *et al.*, [6] in 2003 found ASD 24.4 (% was the most common CHD followed by VSD )21.9 (% and PDA )20.6 (% in Down syndrome children. Ercan *et al.*, [5] in 2010 found isolated ASD (25%) was the most common CHD followed by isolated AVSD (21%) and ASD with VSD (14%).

Equal incidence of AVSD and ASD has been reported by Boaset al ., [12] in 2009 in Down syndrome children followed by VSD 27.4. %

workers have reported that However following AVSD was the most common CHD in Down syndrome children. Tubmen *et al.*, [13] in 1991 found AVSD )34 (% was the most common type of CHD in Down syndrome children followed by ASD )21 (% and isolated PDA (18%). Bhatia *et al.*, [14] 1992 found endocardial cushion defect )31.7 (% was the commonest anomaly followed by ventricular septal defect (27.2%), atrial septal defect (13.6%) in Down syndrome children. Freeman *et al.*, [10] in 1998 found isolated D complete AVSD was the most common type of CH followed by isolated VSD and isolated ASD. M.M. Mokhtar *et al.*, [11] in 2001 found AVSD (46%) was the most common CHD in Down syndrome children followed by ASD (28%) and AVSD and ASD (11%). Weijerman *et al.*, [15] in 2010 found isolated septal defect Atrioventricular s (44.4%) was the most

common CHD followed by Isolated VSD (18.4%) followed by ASD and VSD (14%). NN Fatema [16] in 2010 found atrioventricular canal defect (15.6%) was the most common CHD followed by VSD (14.6%) and ASD (13.2%).

D. Paladini *et al.*, [17] in 2000 found VSD )48 (% and AVSD )44 (% were the most common CHD in Down syndrome children followed by TOF )4. (% Abbag *et al.*, [6, 8] in 2006 found VSD (33.3%) was the most common CHD followed by AVSD (22.8%) and ASD (21.1%). Jashami *et al.*, [9] in 2007 found VSD (54.5%) followed by ASD (17.2%) and mitral valve prolapse (14.5%) was the most common CHD in Down syndrome children.

Azman *et al.*, [4] in 2007 found patent foramen ovale (22.8%), ventricular septal defect (20%), atrioventricular septal defect (20%), were the most common CHD in Down syndrome children.

ASD (41.10%), PDA (21.92%) and VSD were 3 most common congenital type of (%15.07) individual heart defects.

In our study ASD (41.10%), PDA )21.92 (% and VSD )15.07 (% were 3 most common congenital type of individual heart defects in Down syndrome children (Table-4.)

**Table-4: Distribution of Congenital heart disease according to individual congenital heart defect in Down syndrome children**

.S.NO	heart disease Congenital	.No	%
1	ASD	30	41.10
2	PDA	16	21.92
3	VSD	11	15.07
4	PFO	7	9.59
5	Complete AV canal defect	4	5.48
6	Bicuspid aortic valve	2	2.74
7	Incomplete AV canal defect	1	1.37
8	Tetralogy of Fallot	1	1.37
9	Pentalogy of Fallot	1	1.37
	defects Total	73	100

## DISCUSSION

Ercan *et al.*, [5] in 2010 found ASD(38%) was the most common congenital type of individual heart defect followed by a trioventricular canal defect(21%) and VSD(19.5%). NN Fatema [16] in 2010 found ASD (26%) was the most common congenital type of individual heart defect followed by VSD(24.4%) and PDA(17.7%).

Tubmen *et al.*, [13] in 1991 found AVSD (35%) was the most common congenital type of individual heart defect followed by ASD(22.5%), PDA )17.5 (% and VSD )17.5 (% in Down syndrome children. Bhatia *et al.*, [14] 1992 found endocardial cushion defect )31.7 (% was the commonest congenital type of individual heart defect followed by VSD(27.2%) and ASD(13.6%) in Down syndrome children. Freeman *et al.*, [10] in 1998 found AVSD was the most common congenital type of individual heart defect followed by VSD and ASD. M. M. Mokhtar *et al.*, [11] in 2001 found AVSD was the most common congenital type of individual heart defect in Down syndrome children ASD and PDA followed by.

D. Paladini *et al.*, [17] in 2000 found VSD )48 (% was the most common congenital type of individual heart defect in Down syndrome children followed by TOF )4.(% Abbag *et al.*, [8] 2006 in found VSD was the most common congenital type of individual heart defect followed by AVSD and ASD in Down syndrome children. Jashami *et al.*, [9] 2007 in found VSD )54.5 (% followed by ASD )17.2 (% and mitral valve prolapse )14.5 (% were the most common congenital type of individual heart defects in Down syndrome children.

Figueroa *et al.*, [6] in 2003 found PDA was the most common congenital type of individual heart defect followed by ASD and VSD in Down syndrome children.

Azman *et al.*, [4] in 2007 found patent foramen ovale (22.8%), ventricular septal defect (20%), atrioventricular septal defect (20%), were the most common congenital type of individual heart defects in Down syndrome children.

However summarising the results of present study and other workers we can arrive at a conclusion that common CHD in Down syndrome that three most children are ASD, AVSD and VSD. The difference in the observations of different workers could be due to the ethnicity, geographical area and study subject inclusion criteria.

## CONCLUSIONS

The present study syndrome of 70 Down children was conducted in SPMCHI, Department of Pediatrics, attached to SMS Medical College, Jaipur between April to 2012 September 2013 to study the patterns of congenital heart diseases in children with Down syndrome ECG, Ray-with the help of Chest X and echocardiography and to find out the frequency of various clinical & phenotypical manifestations in Down syndrome children confirmed by karyotyping.

The following conclusions were drawn from this study:-

- DS children were more commonly born to younger mothers
- children with Down syndrome had 67.14% congenital heart disease
- Down syndrome children with CHD 95.74% were having acyanotic CHD
- All the CHDs single congenital type of heart defect was most commonly observed
- common Isolated ASD (27.66%) was the most congenital heart disease followed by ASD with PDA (17.02%) and isolated PFO (8.51%)
- ASD (41.10%), PDA (21.92%) and VSD were 3 most common congenital types of individual heart defects

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