

Original Research Article

Pattern of congenital heart diseases among children with Down syndrome attending a tertiary care medical college hospital

Kumar G. V.^{1*}, Srinivasa V.², Ananda Kumar T. S.¹

¹Department of Pediatrics, Sri Siddhartha Medical College, Tumkur, Karnataka, India

²Department of Pediatrics, Basaveshwara Medical College, Chitradurga, Karnataka, India

Received: 19 March 2017

Accepted: 22 April 2017

*Correspondence:

Dr. Kumar G. V.,

E-mail: kumargowripura@yahoo.co.in

Copyright: © the author(s), publisher and licensee Medip Academy. This is an open-access article distributed under the terms of the Creative Commons Attribution Non-Commercial License, which permits unrestricted non-commercial use, distribution, and reproduction in any medium, provided the original work is properly cited.

ABSTRACT

Background: Down syndrome (DS) is the most common chromosomal abnormality associated with learning difficulties with reported incidence between 1/600 -1/1000 live births. DS (trisomy 21) is the most common chromosomal abnormality seen in clinical practice. Most children with DS have trisomy 21, due to chromosomal non-disjunction during meiosis; however, other abnormalities, such as Robertsonian translocation, mosaic, double or triple aneuploidies have been reported. The association between DS and congenital heart disease (CHD) is well established. Congenital heart disease is the most common cause of death among patients with DS and affected children have an increased risk of mortality.

Methods: The present descriptive study was conducted on children who had clinical features suggestive of Down syndrome and who were karyotypically proved as Down syndrome.

Results: In 100 cases of down syndrome 60 children were males and 40 children were females. 45 children had congenital heart diseases. The frequency of CHD in down syndrome is 45%. Ventricular septal defect was the most common CHD found 40% children with down syndrome.

Conclusions: All children with Down's syndrome should have a cardiac evaluation at birth. Early referral and screening of all babies born with the clinical phenotype of DS should be encouraged due to the high prevalence of congenital heart defects.

Keywords: Congenital heart disease, Down syndrome, Ventricular septal defect

INTRODUCTION

Down syndrome (DS) is the most common chromosomal abnormality associated with learning difficulties with reported incidence between 1/600 -1/1000 live births.¹ Down syndrome is also called Trisomy 21, is the commonest genetic pattern of malformation in human being. Down syndrome was first described by John Langdon Haydon Down with characteristic physical features and problems and so known as Down syndrome.² DS (trisomy 21) is the most common chromosomal abnormality seen in clinical practice.³ Most children with DS have trisomy 21, due to chromosomal non-disjunction during meiosis; however, other abnormalities, such as

Robertsonian translocation, mosaic, double or triple aneuploidies have been reported.⁴ The association between DS and congenital heart disease (CHD) is well established. Congenital heart disease is the most common cause of death among patients with DS and affected children have an increased risk of mortality.⁵⁻⁸ The prevalence of congenital heart abnormalities in patients with Down syndrome ranges from 40 to 50%.^{9,10} Among the patients who have a congenital heart defect, half of them presents with atrioventricular septal defect (AVSD), which is rarely an isolated heart defect (2.8%).^{9,11} In addition to AVSD, interatrial communication (IAC), interventricular communication (IVC) and patent ductus arteriosus (PDA) are also frequent in Down syndrome.

These pathologies are associated with lower mortality rates and less complications.⁹ The present study was conducted to evaluate the frequency of various types of congenital heart defects in Down's syndrome children who attended the tertiary care medical college hospital.

METHODS

The present descriptive study was conducted in the Department of Pediatrics, Sri Siddhartha Medical College hospital, Tumkur and Department of Pediatrics, Basveshwara Medical College Hospital Chitradurga, Karnataka, India. Hundred consecutive children who had clinical features suggestive of Down syndrome were subjected to karyotyping. The children who were karyotypically proved as Down syndrome were included in the study. After a detailed history and physical examination, Down syndrome children were subjected to chest X-ray, ECG and two-dimensional echocardiography with colour Doppler studies. The results were statistically analysed.

RESULTS

A total of consecutive 100 cases of Down syndrome were included in the study. In these 100 cases of Down syndrome 60 children were males and 40 children were females. 45 children had congenital heart diseases. Out of 60 male children 33 children had CHD and out of 40 female children 12 children had CHD. The frequency of CHD in Down syndrome is 45% (Table 1).

Table 1: Sex distribution among Down syndrome with congenital heart disease.

Sex	CHD	No CHD	Total
Male	33	27	60
Female	12	28	40
Total	45	55	100

About 93 cases belonged to Non-disjunction, among which 41 children had CHD. Translocation was noted in 6 cases, among which 4 cases had CHD. Mosaicism is noted in one case which did not have any CHD (Table 2).

Table 2: Genetic composition among Down syndrome with congenital heart disease.

Karyotyping	CHD	No CHD	Total
Non-disjunction	41	52	93
Translocation (14,22)	4	2	6
Mosaicism	0	1	1

Ventricular septal defect was the most common CHD found in the Down syndrome children. Ventricular septal defect was found in 18 (40%) children with down syndrome. Endocardial cushion defect was seen in 11 (24.4%) children. Eight (17.7%) children had atrial septal defect. Patent ductus arteriosus and tetralogy of fallot

was seen in 5 (11.1%) children and 3 (6.6%) children respectively (Table 3).

Table 3: Pattern of CHD in children with Down syndrome.

Congenital heart disease	No	%
Ventricular septal defect	18	40
Endocardial cushion defect	11	24.4
Atrial septal defect	08	17.7
Patent ductus arteriosus	05	11.1
Tetralogy of fallot	03	6.6
Total	45	

DISCUSSION

Down syndrome is the most frequently observed autosomal chromosome anomaly among live births and is the main genetic cause of mild to moderate mental retardation in children.¹² About 40-50% of children with Down syndrome have congenital heart disease. The incidence of congenital heart disease in the present study is 45%. In the study conducted by Wells GL et al the incidence of CHDs was 48%, which is comparable to present study in a similar study by Bhatia et al, the incidence of CHD was 44%.^{13,14}

In the present study, ventricular septal defect (40%) was the most common CHD associated with Down syndrome children, followed by endocardial cushion defect (24.4%), atrial septal defect (17.7%) and patent ductus arteriosus (11.1%). In the study conducted by Paladini D et al, the incidence of CHD in Down syndrome was 56%. The most common CHD was ventricular septal defect (48%), followed by atrio-ventricular septal defect (44%).¹⁵ There are many studies which have results comparable to the present study (Table 4).¹⁵⁻¹⁷

Table 4: Comparison of CHD in Down syndrome from various studies.

Study	Percentage of CHD	Most common cardiac anomaly	%
Freeman SB et al	44	Endocardial cushion defect	45
Paladini D et al	56	Ventricular septal defect	48
Frid C et al	47.5	Endocardial cushion defect	42.1
Present study	45	Ventricular septal defect	40

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. All children with Down's syndrome should have a cardiac evaluation at birth. Early referral and

screening of all babies born with the clinical phenotype of DS should be encouraged due to the high prevalence of congenital heart defects.

Funding: No funding sources

Conflict of interest: None declared

Ethical approval: The study was approved by the Institutional Ethics Committee

REFERENCES

- Smith G, Berg J. Down's anomaly. 2nd ed. Edinburgh and New York: Churchill Livingstone; 1976.
- Fatema NN. Down's syndrome with congenital heart disease: Analysis of cases over two years in a non-invasive laboratory of a tertiary hospital. Cardiovasc. J. 2010;2(2):184-7.
- Spicer RL. Cardiovascular disease in down syndrome. Pediatr Clin North Am. 1984;31:1331-43.
- Morris JK, Alberman E, Mutton D, Jacobs P. Cytogenetic and epidemiological findings in Down syndrome: England and Wales 1989-2009. Am J Med Genet Part A. 2012;158A:1151-7.
- Freeman SB, Taft LF, Dooley KJ, Allran K, Sherman SL, Hassold TJ, et al. Population-based study of congenital heart defects in Down syndrome. Am J Med Genet. 1998;80:213-7.
- Nahar R, Kotecha U, Puri RD, Pandey RM, Verma IC. Survival analysis of Down syndrome cohort in a tertiary health care center in India. Indian J Pediatr. 2013;80:118-23.
- Shin M, Kucik JE, Correa A. Causes of death and case fatality rates among infants with down syndrome in metropolitan Atlanta. Birth Defects Res A Clin Mol Teratol. 2007;79:775-80.
- Torfs CP, Christianson RE. Anomalies in Down syndrome individuals in a large population-based registry. Am J Med Genet. 1998;77:431-8.
- Mikkelsen M, Poulsen H, Nielsen KG. Incidence, survival, and mortality in Down syndrome in Denmark. Am J Genet Suppl. 1990;7:75-8.
- Rodríguez LH, Reyes JN. Cardiopatías congénitas en el síndrome de Down. Bol Med Hosp Infant Mex. 1984;41:622-5.
- Stoll C, Alembik Y, Dott B, Roth MP. Study of Down syndrome in 238,942 consecutive births. Ann Genet. 1998;41:44-51.
- Nussbaum R, McInnes RR, Willard HF. Thompson and Thompson Genetics in Medicine. 7th Ed., Elsevier Health Sciences, USA., ISBN: 9781437700930; 2007:600.
- Wells GL, Barker SE, Finely SC, Colvin EV. Congenital heart disease in infants with down syndrome, South Med J. 1994;87(7):724-7.
- Bhatia S, Verma IC, Shrivastava S. Congenital heart disease in Down syndrome: an echocardiographic study, Indian Pediatr. 1992;29(9):1113-6.
- Paladini D, Tartagolione A, Agangi A, Teodoro A, Forleo F, Borghese A, et al. The association between congenital heart disease and Down syndrome in prenatal life. Ultrasound Obstet Gynecol. 2000;15(2):104-8.
- Freeman SB, Taft LF, Dooley KJ, Allran K, Sherman SL, Hassold TJ, et al. Population based study of congenital heart defects in down syndrome, Am J Med Genet. 1998;16;80(3):213-7.
- Frid C, Drott P, Lundell B, Rasmussen F, Anneren G. Mortality in down syndrome in relation to congenital malformation; J Intellect Disabil Res. 1999;43(pt-3):234-41.

Cite this article as: Kumar GV, Srinivasa V, Kumar ATS. Pattern of congenital heart diseases among children with Down syndrome attending a tertiary care medical college hospital. Int J Contemp Pediatr 2017;4:1357-9.