International Journal of Sciences & Applied Research

www.ijsar.in

Extracardiac anomalies in children with congenital heart disease presented to tertiary care hospital

Amber B. Mir[1]*, Syed Baasit[2], Kaisar Ahmad[3]

[1]Senior Resident, Department of paediatrics, GMC Srinagar
[2]Senior Resident, Department of Orthopaedics, GMC Srinagar
[3]Professor, Deparment of Paediatrics, GMC

Corresponding author: *Amber B. Mir, Alamdar colony, Gopal-pora, Chadoora, Budgam, J&K, India Pin: 191113.

Abstract

Background: Congenital heart diseases are one of the main causes of death among the congenital malformations. An extracardiac malformation further increases the risk of morbidity and mortality to these patients, in addition to risks of surgical correction. So here comes the importance of extracardiac malformation.

Methods: This prospective observational study was conducted at Department of Pediatrics,Government Medical College Srinagar The study was conducted over a period of 2 years (from January 2016 to December 2018). Children with congenital heart disease were examined for extracardiac defects.

Results: This hospital based cross-sectional study found that there were 2084 cases of congenital heart diseases among which extracardiac anomalies were present in 416 [19.9%] Out of these n=203[49.1%]were associated with syndromes .Down's syndrome was commonest found in 112 cases[55.1%]. Multiple congenital defects was seen in n=168[40%] of which musculoskeletal defects were commonest 43.4%.Laterality defect were least common presnt in 45 cases [10.8%].

Conclusions: Association of Congenital heart disease with extracardiac anomalies is well known. Syndromes are most commonly associated.

Keywords: Congenital cardiac malformation, Etiology, Extracardiac malformations, Lives infants, Syndromes

Introduction

Congenital heart defects (CHDs) are a common variety of birth defects, with an overall prevalence of 8.1/1000 births [1]. They account for approximately one third of all congenital anomalies, and are the single largest contributor to infant mortality attributable to birth defects [2]. Clinical studies have reported that up to 30% of children with CHDs may have additional extracardiac birth defects that may further add to the morbidity and mortality [3]. Data regarding proportion and pattern of these defects in Indian children with CHDs are scarce. We planned this study to assess the proportion and profile of extracardiac birth defects in children with CHDs to referred to a tertiary care hospital in Kashmir.

Patients and methods

This prospective observational study was conducted at Department of Pediatrics, Government medical collge Srinagar. The study was conducted over period of 2 years (from January 2016 to December 2018]. CHDs from pediatric outpatient department, pediatric wards, pediatric and neonatal intensivecare units both symptomatic and those incidentally detected, were examined for extracardiac defects. The CHDs were diagnosed on basis of detailed clinical examination, chest *X*-ray, and electrocardiography (ECG); and confirmed by 2D-echocardiography and Computed tomography wherever necessary.

The extracardiac defects were diagnosed based on standard definitions [4]. Wherever indicated, further investigations like ultrasound (abdomen/skull), computed tomography (CT), audiometry, ophthalmologic evaluation and thyroid hormone profile were done. G-banded karyotype and fluorescence in situ hybridization (FISH) studies (for microdeletions) were performed whenever indicated. Defects were termed as 'major' if they had medical or surgical significance, or serious cosmetic significance [4]. 'Minor' defects were defined as unusual morphologic features not having any serious medical, surgical or cosmetic significance [4].

Only patients with CHDs having associated extracardiac defects were included as study population and were classified into three distinct groups:

(*i*) Multiple congenital defects (MCDs); MCDs comprised those having at least one major or three minor extracardiac congenital birth defects [4]

(*ii*) Syndromes; Those with chromosomal anomalies, single-gene defects or teratogenic syndromes were counted among syndromes [4].

(*iii*) Laterality defects. Children having laterality defects (*e.g*., Heterotaxy syndromes) were grouped separately [4].

Results

Out of a total 2084 children diagnosed with CHDs during the study period.1250 were males while 834 were females. Acyanotic CHD was present in 1512[72.55%] patients while 572[27.44%] had cyanotic CHD. 416 (19.99%) had associated extracardiac birth defects.

Table 1

Congenital heart disease	With extracardiac defects	Without extracardiac defects	Total
Acyanotic	257[61.77%]	1255[75.2%]	1512[72.55%]
Cyanotic	159[38.22%]	413[24.7%]	572[27.44%]
Total	416[19.9%]	1668[80.03%]	2084

Table 2 shows that extracardiac anomalies system wise .Out of 416 cases with extracardiac defects majority were syndromic n=203[49.15%], followed by multiple congenital defects in 168 cases[40%]. Laterality defects were least common seen in 45 cases

Table 2	N=416	Percentage
Syndromes	N=203	49.15%
Multiple congenital defects	N=168	40%
Laterality defects	N=45	10.8%

Table 3 depicts distribution of multiple congenital defects. Musculoskeletal defects were commonest associated defect seen in 73 patients [43.4%]. Central nervous system, genitourinary system, gastrointestinal system, genitourinary, respiratory, others comprising 17.8%,13%,7.1%,5.9%,12.5% respectively.

Table 3	**N=168**	**Percentage**
Musculoskeletal	73	43.4%
Central nervous	30	17.8%
gastrointestinal	22	13.0%
Genitourinary	12	7.1%
Respiratory	10	5.9%
Others [dermatological]	21	12.5%

Syndromes

Table 4	**N=203**	**Percentage**
Downs syndrome	112	55.1%
Edwards syndrome	9	4.4%
Digeorge syndrome	10	4.9%
Holt Oram sybdrome	7	3.4%
Williams syndrome	9	4.4%
Turners syndrome	3	1.4%
Noonan syndrome	5	2.4%
Achondroplasia	8	3.9%
Congenital rubella syndrome	9	4.4%
Pierre robin syndrome	10	4.9%
Treacher collins syndrome	5	2.4%
Ellis van crevald syndrome	4	1.9%
Marfans syndrome	8	3.9%
Patau syndrome	4	1.9%

Table 4 depicts distribution of syndromes in congenital heart diseases. 55.1% cases of Down syndrome are associated with congenital heart disease.The most common cardiac lesion in Downs syndrome was complete atrioventricular septal defect.

Laterality defects

Table 5	**N=45**	**Percentage**
Right isomerism	23	51.1%
Left isomerism	22	48.8%

Table 5 depicts the laterality defects 23 cases had asplenia while 22 patients had interrupted inferior venacava with polysplenia.

Discussion

In this study total 2084 cases of congenital heart disease were diagnosed during study period of two years. Extracardiac anomalies were present in 416 cases [19.9%].Various studies had found different incidence of

extracardiac anomalies with CHD. Pradat retrospectively observed the data of children with major CHD in Sweden.[5] extracardiac malformation was identified in 25.7% patients after excluding 15% of the children with chromosomal anomalies. Bosi et al retrospectively evaluated CHD in Italy and found 24% of these patients had extracardiac malformations[6] Chromosomal anomalies were found in 9.1% of the newborns. The genitourinary system was the most frequently affected.
In a study by Julian and Farrú in Chile found that extracardiac malformation children with congenital heart disease was 31.9% of the patients, 22.7% of which constituted part of some syndrome.[7] Karande et al[8] Miller et al[9] and Dilber and Malcic[10] found incidence of extracardiac anomalies17.5%, 13.5% and 14.5% respectively which was lower than our study. Abdullah et al [11] in Saudi Arabia found incidence of 28.5% extracardiac anomalies. A significant association with congenital cardiac malformation was found between identified syndrome, central nervous system, genitourinary and musculoskeletal system and extracardiac anomalies.In our study identified syndromes were commonest and significantly associated with CHD occurring in 203 cases comprising 49.15%.Multiple congenital defects were the second commonest occurring in 168 cases comprising [40%].In the multiple congenital defects musculoskeletal was commonest n=73[43.4%],followed by CNS defects n=30 [17.8%].Laterality defects were least common found only in 45 cases of which 23 cases were right isomerism and 22 cases were left isomerism. In the studies by Miller, *et al*. [9] conducted a population based surveillance study of 7984 live-born and stillborn infants and fetuses with CHDs: 1080(13.5%) had multiple, 1048 (13.1%) had syndromic and161 (2.0%) had laterality defects . Tennstedt, *et al*.[12] conducted a necropsy study in 815 fetuses; 85 (66%)had extracardiac birth defects with central nervous system birth defects being the most frequent. Gucer, *et al*.[13] conducted autopsies in 305 children with CHDs; 140 46%) had extracardiac birth defects with craniofacial birth defects being the most frequent.In all these studies the multiple congenital defects were more than syndromes.The increased number of syndromes in our study can be due to large number of patients with Triosomy 21 . All children with Downs syndrome are screened for CHD in our hospital as a protocol. Downs syndrome was commonest syndrome associated with CHD in our study occurring in 112 cases comprising 55.1%. The prevalence of congenital heart disease in down syndrome was similar to the findings from other studies, which have been between 44% and 62%.[14]

Limitation

Our study has several limitations, The actual incidence of congenital heart disease with or without extracardiac anomalies, exactly can be determined if we have included all live births, fetal deaths and spontaneous and induced abortions. However, in this study, fetal deaths and abortions were not studied. Due to financial constraints, small structural cytogenetic abnormalities and single-gene mutations might have remained undiagnosed, resulting in the inadvertent inclusion of some genetically determined cases in the MCDs group.

Conclusion

Association of Congenital heart disease with extracardiac anomalies is well known. Syndromes are most commonly associated. The paediatrician must be aware that extracardiac anomalies are common with congenital heart defects. Association of congenital heart diseases with chromosomal anomalies helps in surgical management of anomalies,

early intervention for developmental delay, rehabilitation and genetic counselling for future pregnancies. Therefore, it is necessary to perform routine radiological and biochemical screening in such cases.

Acknowledgement

This study also involved help from other departments of our medical college like department of orthopaedics. It is mentioned here as a gratitude to these deparments.

References

1. National Birth Defects Prevention Network. Guidelinesfor Conducting Birth Defects Surveillance. Atlanta, GA: National Birth Defects Prevention Network, Inc; 2004.
2. Reller MD, Strickland MJ, Riehle-Colarusso T, MahleWT, Correa A. Prevalence of congenital heart defects in metropolitan Atlanta, 1998-2005. J Pediatr. 2008;153:807-13.
3. Copeland GE, Kirby RS. Using birth defects registry data to evaluate infant and childhood mortality associated with birth defects: an alternative to traditional mortality assessment using underlying cause of death statistics. Birth Defects Res A Clin Mol Teratol. 2007;79:792-7.
4. Rasmussen SA, Olney RS, Holmes LB, Lin AE, KepplerNoreuil KM, Moore CA. Guidelines for Case Classification for the National Birth Defects Prevention Study. Birth Defects Res A Clin Mol Teratol. 2003;67:193-201.
5. Pradat P. Noncardiac malformations at major congenital heart defects. Pediatr Cardiol. 1997;18:11-8.
6. Bosi G, Garani G, Scorrano M, Calzolari E. IMERWorking Party Temporal variability in birth prevalence of congenital heart defects as recorded by a general birth defects registry. J Pediatr. 2003; 142:690-8
7. Jullian PM, Farrú AO. Extra cardic abnormalities in congenital heart defects. Rev Chil Pediatr. 1986; 57: 430-3
8. Karande S, Patil V, Kher A, Muranjan M. Extracardiac Birth Defects in Children with Congenital Heart Defects. Ind Pediatr. 2014:51:389- 91.
9. Miller A, Riehle-Colarusso T, Alverson CJ, Frías JL, Correa A. Congenital heart defects and major structural noncardiac anomalies, Atlanta, Georgia, 1968 to 2005. J Pediatr. 2011;159:70-8
10. Dilber D, Malcić I. Sprectrum of congenital heart defects in Croatia. Eur J Pediatr. 2010;169:543-50
11. Abdullah A. Alabdulgader. Extracardiac AnomaliesAssociated with Congenital Cardiac Malformations in Saudi Arabian Population. Res J Cardiol. 2012;5: 12-9.
12. Tennstedt C, Chaoui R, Korner H, Dietel M. Spectrum ofcongenital heart defects and extracardiac malformations associated with chromosomal abnormalities: Results of a seven year necropsy study. Heart. 1999;82:34-9
13. Gucer S, Ince T, Kale G, Akcoren Z, Ozkutlu S, Talim B,et al. Noncardiac malformations in congenital heartdisease: a retrospective analysis of 305 pediatric autopsies. Turk J Pediatr. 2005;47:159-66.
14. Bermudez BE, Medeiros SL, Bermudez MB, Novadzki IM, Magdalena NI. Down syndrome: Prevalence and distribution of congenital heart disease in Brazil. Sao Paulo Medic J. 2015; 133(6):521-4.