International Journal of Biomedical Research
ISSN: 0976-9633 (Online); 2455-0566 (Print)
Journal DOI: https://doi.org/10.7439/ijbr
CODEN: IJBRFA

Original Research Article

A study of the clinical and etiological profile of patients presenting with Congenital Heart Diseases

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***Article History:**
Received: 21/01/2018
Revised: 30/12/2017
Accepted: 31/01/2018
DOI: https://doi.org/10.7439/ijbr.v9i1.4601

Abstract

Introduction: Congenital Heart Diseases are amongst the major congenital malformations contributing to infant mortality. Most CHDs are diagnosed in infancy and cyanosis and cardiac failure are the principal signs. Later, in childhood or adolescence, CHD patients may present differently.
Objective: To Study the clinical & Etiological profile of Patient presenting as CHD.
Material and Methods: This prospective study was conducted in Dept. of pediatrics, RNT Medical College, Udaipur. 100 children having sign and symptoms suggestive of CHD, attending the Balchikitsalay, were included in this study. Each case worked out by a detailed history, physical examination and various investigations. 1. CBC 2. Chest X-Ray 3. Echo Cardiography.
Results: Majority of the cases in our study were of Acyanotic CHD (73%) and most of them presented during 0-1 Year of life (78.08%). Dyspnoea was the commonest symptoms (62%) followed by FTT (40%) cases and Recurrent RTI (36%) cases, Refusal to feed (34%) cases, cyanosis (25%) cases, Anoxic Spell (5%) of cases. 73 Patients had abnormal chest X-ray finding on presentation.
Conclusion: Acyanotic CHD were most commonly seen in children rather than cyanotic CHD.VSD was most common acyanotic congenital heart disease in infancy. Among cyanotic CHDs, Tetralogy of Fallot is the most common lesion. All cases of CHD should be under regular monitoring so as to permit optimal growth and development. A high index of suspicion, a detailed history, physical examination, chest X-ray along with the use of 2-D-Echocardiography which not only helps us to diagnose most of the cases of congenital heart disease but also help in diagnosing severity of the lesion.
Keywords: Congenital Heart Diseases (CHD), 2-D-Echocardiography, X-ray.

1. Introduction

Congenital heart defects (CHDs) have been defined as gross structural abnormalities of the heart or intrathoracic great vessels that are actually or potentially of functional significance [1].

Traditionally, CHDs have been categorized according to whether the defect may cause cyanosis. *Cyanotic* lesions cause the presence of desaturated blood in the systemic circulation due to right to left shunting and include tetralogy of Fallot (TOF), transposition of the great arteries (TGA), and Ebstein's anomaly. *Acyanotic* lesions include ventricular septal defect (VSD), atrial septal defect (ASD), patent arterial duct (PDA), pulmonary stenosis (PS), atrial stenosis (AS), and coarctation of the aorta (CoA) [2,3].

CHD vary immensely in severity, natural history and management. Minor forms of CHD include mild PS, small ASD, and VSD, and they rarely require intervention. More severe CHDs with four heart chambers and 4 heart valves like CoA, severe AS, large VSD, TOF, and TGA require intervention to prevent heart failure, cyanosis, pulmonary hypertension or death. Highly complex CHD include atresia and hypoplasia of valve or chamber [4]. Most CHDs are diagnosed in infancy [5], and cyanosis and cardiac failure are the principal signs. Later, in childhood or

adolescence, CHD patients may present differently with e.g. a heart murmur, abnormal heart rate, absent pulses or hypertension.

1.1 Incidence of CHD

Congenital heart disease occurs in 0.5–0.8% of live births. The incidence is higher in stillborn (3–4%), spontaneous abortuses (10–25%), and premature infants (about 2% excluding patent ductus arteriosus [PDA]). This overall incidence does not include mitral valve prolapse, PDA of preterm infants, and bicuspid aortic valves (present in 1–2% of adults). Congenital cardiac defects have a wide spectrum of severity in infants: about 2–3 in 1,000 newborn infants will be symptomatic with heart disease in the 1st year of life. The diagnosis is established by 1 week of age in 40–50% of patients with congenital heart disease and by 1 month of age in 50–60% of patients. With advances in both palliative and corrective surgery, the number of children with congenital heart disease surviving to adulthood has increased dramatically. Despite these advances, congenital heart disease remains the leading cause of death in children with congenital malformation [6].

Congenital heart diseases have varying degree of presentation. Child with congenital heart disease may manifest with congestive heart failure or may have asymptomatic murmur. A number of studies have been undertaken worldwide on pattern of CHDs but there has been no study on pattern of CHDs in symptomatic and suspected children in Rajasthan. The purpose of this study was to find out common type of CHDs manifests during neonatal and childhood period in our clinical practice.

1.2 Aims & objectives

1) To Study the clinical profile of Patient presenting as CHD. 2) To know the incidence of various CHD in children. 3) To establish relationship between CHD and various Etiological factors.

2. Material & methods

This prospective study was conducted in Dept. of pediatrics, RNT Medical College, Udaipur from January 2012 to December 2012. 100 children having sign and symptoms suggestive of CHD, attending the Balchikitsalay, were included in this study.

2.1 Inclusion Criteria

1) Admitted in NICU, PICU, Pediatric Ward, attending OPD with symptoms [Dyspnea (Breathlessness), Cyanosis, Cyanotic spell, respiratory tract infection, FTT, Refusal to feed] related to Cardiac failure.
2) Abnormal Heart Sound
3) CHF Probably due to Anatomical Cardiac Defect
4) Cyanosis attributable to CHD
5) X-ray shows Cardiomegaly.

2.2 Exclusion Criteria

1) Sign & symptoms suggestive of RHD or Infective Heart Disease
2) Pneumonia

The Demographic profile and relevant information of Individual patient was collected using structural Proforma by interviewing parents and an informed consent was taken. Each case worked out by a detailed history, physical examination and various investigations. 1. CBC 2.Chest X-Ray 3.Echo Cardiography. All these observations were recorded on printed proforma and data analysis were done.

3. Observations

Majority of the cases in our study were male (63%), maximum number of the cases of both sex (74%) presents during the 0-1 Year of life (Table 1).

Table 1: Age & Sex Distribution

Age Group	Male	%	Female	%	Total
0-1Yr	45	60.81	29	39.19	74
1-3Yr	11	78.57	3	21.43	14
3-6Yr	7	58.33	5	41.67	12
Total	**63**		**37**		**100**

Majority of the cases in our study were of Acyanotic CHD (73%) and most of them presented during 0-1 Year of life (78.08%). While total cases of cyanotic CHDs were 23% (Table 2) and most of them presented during 0-1 Year of life (56.52%).

Table 2: Classification of CHDs

CHD	0-1Yr	%	1-3Yr	%	3-6Yr	%	Total
Acyanotic	57	78.08	11	15.07	5	6.85	73
Cyanotic	13	56.52	3	13.04	7	30.43	23
Obstructive	2	100.00	0	0.00	0	0.00	2
Abnormal Position	2	100.00	0	0.00	0	0.00	2
Total	**74**		**14**		**12**		**100**

Dyspnoea was the commonest symptoms seen in (62%) followed by FTT in (40%) cases and Recurrent RTI in (36%) cases, Refusal to feed in (34%) cases, cyanosis in (25%) cases, Anoxic Spell in (5%) of cases. Anoxic Spell was presenting complaint in Cyanotic CHD (Table 3).

Table 3: Clinical Profile of CHDs

Presenting Complaints	Acyanotic	%	Cyanotic	%	Obstructive	%	Abnormal Position	%	Total
Recurrent RTI	31	86.11	4	11.11	0	0.00	1	2.78	36
FTT	29	72.50	9	22.50	1	2.50	1	2.50	40
Cyan-osis	1	4.00	23	92.00	0	0.00	1	4.00	25
Anoxic Spell	0	0.00	5	100.00	0	0.00	0	0.00	5
Refusal to Feed	23	67.65	8	23.53	2	5.88	1	2.94	34
Dyspnoea	38	61.29	21	33.87	2	3.23	1	1.61	62
Total	**122**		**70**		**5**		**5**		**202**

39% cases present as CHF, 69% were Acyanotic CHD & 23% were Cyanotic CHD. Among 69% cases of CHF of Acyanotic CHD, majority were VSD. Majority of cases of Acyanotic CHD had PEM-I & II (47.95%) while most of the Cyanotic CHD children had PEM-III & IV (39.13%). 73 Patients had abnormal chest X-ray finding on presentation. 86.96% of Cyanotic and 68.49% of Acyanotic CHD had abnormal radiological finding. Abnormal finding were also seen in remaining CHD. Maternal history of Intrauterine Infection in 3 cases, Hypothyroidism in 2 cases and Diabetes in 1 case. None of the patients had association with CHD. Extra cardiac anomalies were found in 13% cases out of which Tracheo esophageal fistula was most common (30.7%). Mongoloid features were present in (23.07%) of these cases (Table 4).

Table 4: Extra Cardiac Finding

Ass. Extra-Cardiac Finding	Total	%
B/L congenital cataract	1	1
B/L external ear absent	1	1
B/L cleft lip	2	2
Cleft palate	1	1
Hypertrophied clitoris with single right ovary	1	1
Tracheo esophageal fistula	2	2
Tracheo esophageal fistula + Anorectal malformation	2	2
Low set ear with hypotonia	3	3
No other anomaly	87	87
Total	**100**	**100**

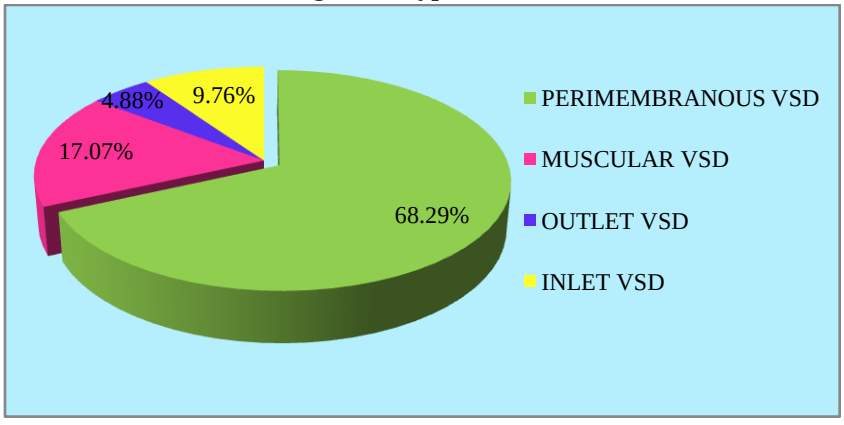
Most cases of the isolated VSD (86.67%) and VSD+ (88.46%) were present during 0-1 year of life. In cyanotic CHD, TOF (63.63%) was most commonly seen during 3-6 years of life (Table 5).

Table 5: Types of CHDs

Congenital Heart Disease		0-1 Yr	1-3 Yr	3-6 Yr	Total
Acyanotic	Isolated VSD	13	1	1	15
	VSD+	23	3	0	26
	ASD	9	3	1	13
	ASD+	5	2	0	7
	PDA	4	0	2	6
	PDA+	3	0	0	3
	ECD	0	2	0	2
	AS	0	0	1	1
Cyanotic	TOF	2	2	7	11
	DTGA	3	1	0	4
	TA	3	0	0	3
	PA	2	0	0	2
	DORV+TGA	1	0	0	1
	EB	1	0	0	1
	PTA	1	0	0	1
Obstructive	Obstructive	2	0	0	2
Abnormal position	Abnormal position	2	0	0	2
Total		**74**	**14**	**12**	**100**

Majority of the cases which present during the 0-1 year of life were VSD, was the most common CHD. From 41 cases of VSD isolated VSD was present in 86.67% cases and remaining. The most common type of VSD is perimemranous (68.29%) followed by muscular VSD (17.07%) (Figure 1). Most cases of VSD were large size 46.34%, medium size 34.15% and small size were 19.51%.

Figure 1: Types of VSD



4. Discussion

Present study was conducted on 100 children in which congenital heart disease was suspected and later on confirmed by 2D Echocardiography .Relevant history with special emphasis on antenatal and family history was taken. Detailed clinical examination of each child was done. All relevant investigations like chest X-ray and CBC of each patient was done and finally diagnosis was made on the basis of 2D Echocardiography.

In our study male to female ratio was 1.7:1. This finding coincides with the previous study. Lo RN *et al* [7], conducted a study between 1981 and 1990 on 765 symptomatic children with major congenital heart malformation. Most of the children (62%) were male in their study. Shah *et al* [8], they conducted a study between January, 2006 and December, 2006 out of 84 CHD cases 51 were males and 33 females with male to female ratio of 1.5:1. This may be due to the fact that most of the population brings the male offspring's for treatment more frequently.

In the present study 74% of patients present during 0-1 year of life, 14% 1-3 year of life & 12% was in 3-6 years of life. In the study conducted by Tank *et al*[9], they found between 1996 to 2000 out of 147 CHD cases they found that the majority of cases 50% present during 0-1 years of life. While considering the age at presentation, we found that maximum number of children were picked up in infancy, including 22 newborns, as is also seen in other studies. In the West, however, there are a higher number of patients reported in the neonatal period. This could be due to the fact that foetal echo-cardiography forms a part of their routine antenatal examination. In a 5 year study (1979-1984) conducted by Udani *et al*[11] the maximum number of cases were seen between 5-12 years. This may be due to the fact that improved awareness and better facilities are freely available now and hence, more children are being picked up at an earlier age than two decades ago when study was conducted by Udani *et al*. [11]

In our study 73% cases had acyanotic CHD, 23% cases had cyanotic CHD and 2% cases had abnormal cardiac position with associated lesion, 2% cases had pure obstructive lesions. Among acynotic CHD, VSD was most common CHD (41%) followed by ASD (20% 0f total cases). PDA was found in 9% cases and 2% cases had endocardial cushion defect & 1% had AS. From 41 cases of VSD, 15 cases (36.58%) had isolated VSD and remaining 26 (63.41%) had some associated lesion with VSD. From 20 cases of ASD, 13 cases (65%) had isolated ASD and remaining 7 cases (35%) had some associated lesion. Among cyanotic CHD, TOF (11%) was the most common anomalies followed by TGA (4%) & TA (3%). Among 2 cases of abnormal positions, 1 had dextrocardia and 1 had mesocardia. Our observations are in accordance with previous studies done. Richard [10] in their study found that VSD was most common CHD found in 20% cases, next was TGA found in 16% of the cases. Mitchell S C *et al* [17] in their in study found that VSD was the most common CHD (31.5%), second most common was ASD (8.7%), next was PDA (7.8%) and out of cyanotic CHD, TGA and TOF were most common. Shima *et al* [16] in their study of 43 newborn babies with congenital heart disease (CHD) found that 72% cases were of acynotic CHD, out of which VSD was most common and 28% cases were of cynotic CHD. Dadvand *et al* [12] in their study found that VSD was the most common CHD (present in 36% cases), second most common was ASD (32%), and next were TGA and TOF (present equally as 14% cases of each). Shah *et al* [8], in their study found that VSD was the most common CHD (58.3%), second most common was TOF (13.1%). However, Garne *et al* [13] in their study found that from 0-1

month – complete transposition (15.8%) was and the most common cardiac lesion and VSD was most common lesion in patients of age group 0-6 month. Lo RN *et al* [7] in their study on 765 symptomatic children found that obstruction of pulmonary outflow tract occurred most frequently (37.8%), followed by left ventricular outflow obstruction, left to right shunting (15.5%), complete transposition (12.4%), and miscellaneous causes (3.3%).

When the symptoms were taken into consideration, we found Dyspnoea to be the commonest symptom (62%), followed by FTT (40%), CHF (39%) and Recurrent RTI (36%), Refusal to feed (34%), Cyanosis (25%), Anoxic Spell (5%) cases. Dyspnoea was the commonest symptom in both cyanotic as well as acyanotic heart disease. FTT and Recurrent RTI were maximally seen in cases of VSD and these patients had large defects with evidence of pulmonary hypertension. Recurrent RTI was also seen in other patients with a large left to right shunt, as in ASD and PDA.

In our study 39% cases were having CHF; among 69% cases of CHF found in Acynotic CHD, majority were VSD. CHF was also seen in Cynotic CHD in (23%) cases. FTT was seen in 40% cases of Acynotic as well as Cynotic. FTT is a major symptom of congenital heart disease, as these children are having low energy intake, low resting energy expenditure, inadequate food intake, and or feeding difficulties. Refusal to feed in 34% cases of all CHD, when patient in CHF unable to take feed.

Cyanosis was seen in 25 cases. Cyanosis was the most common symptoms of Cyanotic CHD were seen in 92%. Anoxic spell was in 5% of cases majority in the Cyanotic CHD. Similar study done by Tank *et al* [9], in their study 147 cases of CHD found that Breathlessness to be the commonest symptom, seen in 110 cases (74.83%), followed by LRTI in 66 patients (44.89%) and FTT in 57 cases (38.77%). Shah *et al* [8] found the most common clinic presentation were FTT (86.9%), breathlessness (69%), LRTI (52%), CHF (46%), Cyanosis (22%) and Cyanotic Spell (9.5%).

73% patients had abnormal chest X-ray findings on presentation in our study. 68.49% cases of Acyanotic CHDs had abnormal chest X-ray findings while 86.93% cases of cyanotic CHDs had abnormal chest X-ray finding. We did not found any positive family history of congenital heart disease and any risk factor which significantly predisposes children to congenital heart disease. Our results are in agreement with the study of other workers. Hoffman *et al* [14] in their study on 50 cases of CHD did not found any association between maternal infections early in pregnancy and the occurrence of CHDs in the offspring, Obstetric and medical complications were failed to show any correlation with the incidence of CHD.

In our study extracardiac anomalies were present in 13 cases. Out of which trecheo esophageal fistula was most common (30.7%), Mongoloid features were present in (23.07%) of the cases. But no significant association could be drawn between any extracardiac anomaly and type of CHD. Similar findings are reported by other studies. Lo RN *et al* [7] in their study found that extracardiac anomalies were present in 17% of the cases. Out of these 3.7% are chromosomal, 2.8% were non-chromosomal and 11.9% were other. Abu-Herb *et al* [1] in their study 0n 457 children of CHDs found that extracardiac anomalies were present in 138 subjects (30%). Humayun *et al* [15] in their study of 1016 cases of congenital heart disease found that major extracardiac malformation was present in 6.6% cases and in another 13.3% cases CHD was part of malformation syndrome or embryopathy complex.

5. Conclusion

CHDs are amongst the major congenital malformations contributing to infant mortality. In developing countries like India it's very unfortunate that most of the CHDs are diagnosed very late due to lack of awareness and resources.

Hence it is concluded that Acyanotic CHD were most commonly seen in children rather than cyanotic CHD.VSD was most common acyanotic congenital heart disease in infancy. VSD when associated with other lesion manifested early rather than when it is in isolated form. Among cyanotic CHDs, Tetralogy of Fallot is the most common lesion. Infancy was the commonest age of presentation of CHD. Most of the children presented as Dysnoea, FTT, CHF, Recurrent RTI, Refusal to feed, Cyanosis and Anoxic spell. Abnormal radiological finding were seen in 73% cases. Echocardiography confirmed the diagnosed of all CHD.

Hence, we recommend that all murmurs should be screened unless thought to be physiological. With a high degree of suspicion in most a cardiac evaluation with echocardiography is advised in all suspected cases of CHD who come to us oftenly repeated Dysponea, FTT, lower respiratory infections and cyanosis. All cases of CHD should be under regular monitoring so as to permit optimal growth and development. A high index of suspicion, a detailed history, physical examination, chest X-ray along with the use of 2-D-Echocardiography which not only helps us to diagnose most of the cases of congenital heart disease but also help in diagnosing severity of the lesion. Early diagnosis, close monitoring and timely intervention in cases of congenital heart disease will go a long way in reducing the morbidity and mortality in these young children to a large extent.

References

[1]. Abu-Herb M, Hey E, Wren C, Death in infancy from unrecognized congenital heart disease. *Arch Dis Child* 1194; 71:3.

[2]. Andrews RE, Tulloh RMR. Interventional cardiac catheterisation in congenital heart disease. *Archives of Disease in Childhood* 2004; 89:1168-73.

[3]. Pai BV and Varkey CC. Spectrum of congenital heart disease in a general hospital. *Indian Journal of Pediatrics* 1974; 41:317-321.

[4]. Beels L, Bacher K, De Wolf D, Werbrouck J, Thierens H. {gamma}-H2AX Foci as a Biomarker for Patient X-Ray Exposure in Pediatric Cardiac Catheterization: Are We Underestimating Radiation Risks? *Circulation* 2009; 120:1903-9.

[5]. Brickner ME, Hillis LD, Lange RA. Congenital heart disease in adults. First of two parts. *N Engl J Med* 2000; 342:256-63.

[6]. Brickner ME, Hillis LD, Lange RA. Congenital heart disease in adults. Second of two parts. *N Engl J Med* 2000; 342:334-42.

[7]. Lo RN, Leung MP, Yung TC, Chan CH. Variability of the diameter of the pulmonary outflow tract and its relation to balloon valvoplasty for congenital pulmonary valvar stenosis. *Cardiology in the Young*. 1993 Apr; 3(2):111-7.

[8]. Shah GS, Singh MK, Pandey TR, Kalakheti BK, Bhandari GP. Incidence of congenital heart disease in tertiary care hospital.

[9]. Tank S, Bhagwat K, Malik S, Jawale P, Joshi S. Familial type IV hypertriglyceridemia presenting as hemiparesis with cerebellar signs. The *Indian Journal of Pediatrics*. 2004 Apr 1; 71(4):349-50.

[10]. Carlgren LE. Incidence of Congenital Heart Disease in children born in Gothenberg 1941-50, *Brit Heart J* 1959; 2:40-50.

[11]. Udani PM. Congenital heart disease. Textbook of Pediatrics, 3rd ed. Ed. Jaypee bros., Delhi 1930-52.

[12]. Dadvand P, Rankin J, Shirley MD, Rushton S, Pless-Mulloli T. Descriptive epidemiology of congenital heart disease in Northern England. *Paediatr Perinat Epidemiol* 2009; 23:58-65.

[13]. Garne E. Congenital heart defects - occurrence, surgery and prognosis in a Danish County. *Scand Cardiovasc J* 2004; 38:357-62.

[14]. Hoffman JIE, Kaplan S. The incidence of congenital heart disease. *Journal of the American College of Cardiology* 2002; 39:1890-900.

[15]. Humayun KN, Atiq M. Clinical profile and outcome of cyanotic congenital heart disease in neonates. Department of Pediatrics and Child Health. *Indian Pediatr* 2008.

[16]. Shima Yoshio, Takechi Nobuyuki, Ogawa Shunichi, Fukasawa Ryuji, Fukumi earth, Yukiko Uchikiba, Yuko Kuramochi, Ninpei Yamauchi. Clinical Characteristics of Congenital Heart Disease Diagnosed During Neonatal Period. Journal *of Nippon Medical School.* 2001; 68(6):510-5.

[17]. Mitchell SC, Korones SB, Berendes HW. Congenital heart disease in 56,109 births incidence and natural history. *Circulation*. 1971 Mar 1; 43(3):323-32.