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Original Research Article

Study of congenital heart disease in neonates: clinical profile, diagnosis, immediate outcome and short-term follow-up

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ABSTRACT

Background: Congenital heart disease (CHD) occurs in approximately 0.8% of live births. Early recognition of CHD is important for appropriate management and decision making regarding referral. The purpose of this study was to document the common presenting symptoms and signs in the neonates with CHD, definitive diagnosis and short-term follow-up for six months.
Methods: Sixty full term neonates with suspected CHD admitted in neonatal intensive care unit (NICU) at Niloufer Hospital, Hyderabad during the period December 2016 to May 2017 were included in the study.
Results: Of the 60 neonates, most common age of presentation was for first week (45%, n = 27). Of the 60 neonates, 32 (54%) were males and 28 (46%) were females. The commonest presentation was hurried respiration (68%), followed by feeding problem (63%) and only eight neonates were asymptomatic with clinically significant murmur. 40% (n = 24) of the babies were born of consanguineous marriage. 72% (n = 43) of babies presented with murmur and 6 babies had extra-cardiac manifestations. Babies with acyanotic CHD were 38 (63%) of which ventricular septal defect (VSD) was the commonest. Cyanotic CHD were 22 (37%) of which transposition of great arteries (TGA) was the commonest. 25 babies (42%) expired during neonatal period. Of the remaining babies during follow-up, 29 % of babies thrived well, 35% presented with repeated respiratory tract infections, 21% with failure to thrive and 15% with congestive heart failure (CHF).
Conclusions: Neonates with CHD have a unique presentation and they carry poor outcome unless diagnosed early and managed appropriately. Babies presenting with multiple anomalies should be screened for any underlying structural heart disease.

Keywords: Acyanotic congenital heart disease, Congestive cardiac failure, Cyanotic congenital heart disease

INTRODUCTION

Congenital heart diseases (CHD) refers to structural or functional heart diseases at birth. These are primarily seen in neonates, infants or children, although it is not uncommon to see adults with uncorrected CHD.

Congenital heart diseases are the most prevalent and serious of all recognized structural birth defects. The burden of CHD in India is likely to be enormous, due to a very high birth rate. This heavy burden emphasizes the importance of this group of heart diseases. The prevalence of CHD is not uniform in our country as various studies have reported it ranging from 1.3 to 50.89 per 1000 live births.[1,2] Also several studies have reported a changing pattern and incidence of CHD in various geographical locations.[3,4] Early recognition of such diseases has great implications. Despite advanced diagnostic facilities and improved medical care, CHD is considered one of the leading causes of neonatal

mortality.[5] According to a status report on CHD in India, 10% of the present infant mortality may be accounted to CHD.[6] Surviving infants often require surgery or interventions and lengthy hospitalizations and will have a lifetime disability that imposes a significant burden on families.

Many cases are asymptomatic and discovered incidentally during routine health check-up.[7] Even though there are numerous different cardiac lesions, there are many similarities in their clinical presentation. Signs and symptoms of severe heart disease in the newborn period include cyanosis, discrepant pulses and blood pressures, congestive heart failure, and cardiogenic shock.

The initial evaluation of any newborn suspected of having critical congenital heart disease includes a meticulous physical exam, four extremity blood pressures, preductal and postductal saturations, hyperoxia test, chest radiograph, ECG and ECHO.[8]

Thus, recognition of congenital heart disease in the newborn is important as this group of abnormalities constitutes a significant proportion of congenital malformation that present in neonatal life, and their early detection is important for appropriate management, and short term follow up for decision making regarding referral.

The objective of our study is to document the common presenting symptoms and signs in the neonates with congenital heart disease, definitive diagnosis and short term follow up for six months.

METHODS

This is a hospital-based study done on 60 full term neonates suspected of congenital heart disease both inborn and referred cases admitted into Neonatal Intensive Care Unit (NICU) of Niloufer Hospital, Osmania Medical College, Hyderabad, Telangana, South India during the period December 2016 to May 2017.

The history and examination of cases included in the study were recorded in the proforma designated for the study. Preterm babies and babies with genetic syndrome were excluded from the study.

Detailed history of presenting complaints, pregnancy, family history, consanguinity, socio-economic status as per modified BG Prasad's classification were taken as per the proforma designed for the study.

Relevant investigations like arterial blood gas analysis, chest X- ray, ECG, Echocardiography, were done to arrive at a definitive diagnosis. Severity of the congenital heart disease is assessed and managed accordingly and those who survived were followed up for duration of 6 months.

Statistical analysis

Data was collected and recorded on a pre-designed proforma and entered in excel database. Data analysis was performed using Epi Info 7 program (CDC Atlanta).

RESULTS

Table 1 depicts the time of presentation of new-born with congenital heart disease. Nearly half of the cases presented in the first week (45%) followed by those in fourth week (40%), third week (8%), second week (7%) in decreasing order.

Table 1: Age at presentation of new born with CHD.

Age	No. of cases (n)	(%)
Week 1	27	45
Week 2	4	7
Week 3	5	8
Week 4	24	40

Table 2 shows the sex distribution of the neonates included in the study. 54% were male and 46% were female.

Table 2: Sex distribution of new-born with CHD.

Sex	No of Cases (n)	Percentage (%)
Male	32	54
Female	28	46

As evident from the below figure, the commonest presenting complaint was hurried respiration (68%), followed by feeding problem, cyanosis and congestive cardiac failure manifested as decreased urinary output and excessive forehead sweating. 8 cases (13%) were asymptomatic and clinically only murmur was present. Out of 60 cases studied, 24 cases were born to consanguineously married couples as evident from Table 3.

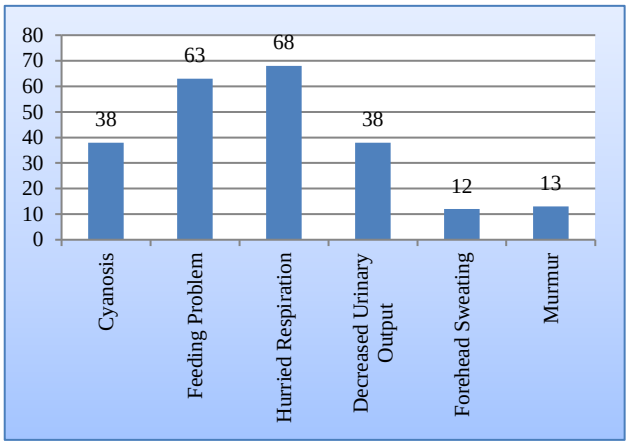

Figure 1: Presenting complaints of the neonates.

Table 3: Consanguinity history in the parents of neonates with CHD.

Born of	No. of cases (n)	Percentage
Consanguineous marriage	24	40
Non-consanguineous marriage	36	60

Table 4: Neonates presenting with murmur.

Murmur	No. of cases (n)	Percentage (%)
Present	43	72
Absent	17	28

Table 5: Extracardiac anomalies in neonates with CHD.

Associated anomalies	No. of cases (n)	Percentage (%)
Anorectal malformations (ARM)	3	5
Hiruchsprung disease (HPD)	1	2
Neonatal hepatitis syndrome (NHS)	2	3
Total present (extracardiac anomalies)	6	10
Absent	54	90

Murmur is an impressive presentation of congenital heart disease. As shown in Table 4, 72% of neonates with CHD presented with murmur and remaining did not have, though all of them had significant underlying structural heart disease.

Table 5 showing extra cardiac anomalies were present in 6 cases (10%), in which anorectal malformation was the commonest association with congenital heart disease.

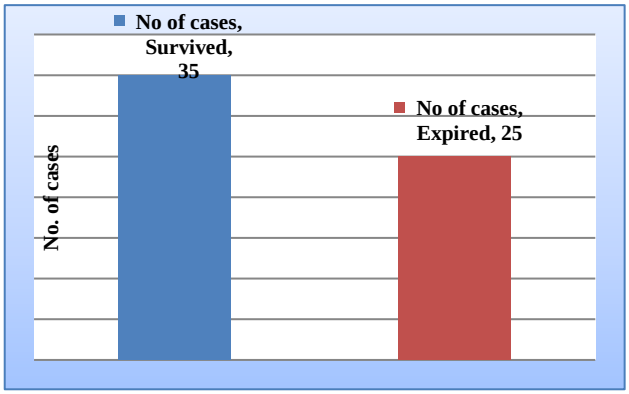

Figure 2: Immediate outcome of the neonates with CHD.

Nearly half of the cases expired in the neonatal period and all of them had severe type of congenital heart disease like single ventricle, hypoplastic heart lesions, critical pulmonary stenosis, large ventricular septal defect (VSD), common atrioventricular(AV) canal defect and tricuspid atresia.

Majority of the cases (63%) were acyanotic type, remaining being cyanotic.

Table 7: Various types of congenital heart disease and their age of presentation.

Type of heart disease	Age (days)	1st wk	2nd wk	3rd wk	4th wk	Tota	%	Total (n)	%
Cyanotic	TOF (Tetralogy of Fallot)	1	1	1	2	5	8	22	37
	TGA (Transposition of great arteries)	6	1	0	0	7	12		
	TAPVR (Total Anomalous of Pulmonary Venous Return)	2	0	0	0	2	3		
	Single Ventricle	1	0	0	0	1	2		
	HLHS (Hypoplastic Left Heart Syndrome)	3	0	0	0	3	5		
	HRHS (Hypoplastic Right Heart Syndrome)	1	0	0	0	1	2		
	TA (Tricuspid Atresia)	1	0	0	0	1	2		
	PS (Pulmonic Stenosis)	2	0	0	0	2	3		
Acyanotic	COA (Coarctation of Aorta)	0	1	0	0	1	2	38	63
	PDA (Patent Ductus Arteriosus)	1	0	0	1	2	3		
	VSD (Ventricular Septal Defect)	2	0	2	14	18	30		
	ASD (Atrial Septal Defect)	4	1	2	5	12	20		
	CAVCD (common atrio-ventricular canal defect)	1	0	0	0	1	2		
	VSD+ASD	2	0	0	2	4	6		

Among the cyanotic, transposition of great arteries was the commonest, followed by tetralogy of Fallot. In the acyanotic group, ventricular septal defect was the commonest, followed by atrial septal defect. Most of the cyanotic CHD, presented within the 1st week of life while acyanotic CHD presented in the 4th week of life.

Table 8: Follow up of the CHD neonates and their complaints.

Complaints	No .of cases (n)	(%)
FTT (Failure to thrive)	7	21
RTI (Respiratory tract infections)	12	35
CCF (Congestive cardiac failure)	5	15
Normal	10	29

Nearly half of the cases survived the neonatal period and these children were followed up for a period of six months. At the end of this period, nearly 35% of children suffered from repeated respiratory tract infection and 21% were failing to thrive and 15% had congestive cardiac failure. 29% of children thrived well and among them 4 cases were operated for PDA and TGA respectively at 2 months of age.

DISCUSSION

According to Mitchell et al's definition, congenital heart disease is a gross structural malformation of the heart or great intrathoracic vessels with a real or potential functional importance.[9] Therefore this definition excludes anomalies such as bicuspid aortic valve without valve dysfunction, mitral valve prolapse, persistent left superior vena cava, anomalous origin of the left subclavian artery, mild valve regurgitation, and functional alterations without a structural component. This definition was adopted in this study, and cases of patent ductus arteriosus, an anomaly that could still be considered functional in the first few hours of life when this study was conducted, were also excluded.

The incidence of moderate to severe structural congenital heart disease in live born infants is 6- 8 per 1000 live births.[10-13] Congenital cardiac defects have a wide spectrum of severity in infants.

The present study was conducted on 60 newborns, both inborn and referred to Neonatal Intensive Care Unit(NICU), Niloufer Hospital, Hyderabad to know the various clinical presentations, definitive diagnosis by echocardiography, their immediate outcome and short term follow up for 6 months.

Most of the severe forms of congenital heart disease, like TGA, TOF, HLHS, HRHS, single ventricle, large VSD manifest in first week of life, while trivial or mild form of congenital heart disease like VSD, acyanotic TOF, COA, manifest in the 3rd and 4th week.[14] In the present study, severe forms of congenital heart disease like TGA, TOF, TAPVR, single ventricle, hypoplastic heart syndrome, tricuspid atresia, critical pulmonary stenosis manifested in first week and mild variety of congenital heart disease – like COA, VSD, ASD manifested in 4th week.

Similarly, in a study conducted in Pakistan on 44 neonates, the mean age of presentation was 5 days; with majority admitted on 1st day of life.[15]

In the present study, the male to female ratio was 1.1:1 which is similar to a study done by Shah GS, et al in Nepal where in the male to female ratio was 1.5:1, and there were gender differences in the occurrence of specific heart lesions in the same study. TGA and left sided obstructive lesions were slightly more in males, whereas VSD, PDA and pulmonary stenosis was more common in girls.[16] Similarly in a study conducted by Humayun et al in Pakistan, male to female ratio was 1.7:1.[15] Similarly in a study conducted in pediatric age group in Maharashtra by Bhushan Deo et al showed male: female ratio being 1.45:1.[17] Male preponderance in congenital heart disease was seen in majority of the studies conducted worldwide.

In the first few weeks of life, the many heterogeneous forms of heart disease present in a surprisingly limited number of ways, like cyanosis, congestive heart failure (decreased urine output, excessive forehead sweating, with extreme presentation being shock), asymptomatic heart murmur and arrhythmia.[10]

In the present study, most common presentation was hurried respiration (68%), followed by feeding difficulty and congestive cardiac failure in the form of decreased urinary output and excessive forehead sweating. Thirty eighty percent neonates presented with cyanosis. Thirteen percent were asymptomatic in which murmur was the only sign. In a study conducted by Sandeep V Harshangi et al in Gulbarga, the commonest symptom was hurried respiration seen in 78% of cases.[18] In a study conducted by Joshi et al in Mumbai, the commonest symptoms were hurried respiration, failure to thrive and refusal to feed.[19] A similar observation was made by Kasturi L, et al in a study, where feeding difficulty and hurried respiration were the commonest presenting symptoms.[20]

Consanguinity plays a major role in the incidence of major congenital malformation in children. In a study conducted by Kulkarni ML, et al in 3700 consecutive births on the effect of consanguinity on fetal growth and development, 26% of the total births were to consanguineous couples. The incidence of congenital malformation was 39.1/1000 births with significantly higher incidence among the consanguineous group (8.01%) as against the non-consanguineous group (2.42%). Malformation of cardiovascular system was 10 times more in the consanguineous group as compared to non-consanguineous group.[21] Similarly in a recent study

on 759 Lebanese patients with congenital heart disease, parental consanguinity had a major role.[22] Cardiac lesion like aortic anomalies, atrial septal defect, double outlet right ventricle, pulmonary atresia, PDA, pulmonary stenosis, tetrology of Fallot, and VSD were more common in the consanguineous group.[22]

In the present study, 24 neonates (40%) were born to consanguineously married couple. Similar results were obtained in a study conducted by Bhushan Deo et al which showed that 33.33% of children with CHD were born to consanguineous parents.[17]

In the present study, 43 babies (72%) had murmur and 17 babies presented without murmur though all of them had significant cardiac lesion. In a study conducted in Indore by Bansal et al, 2603 newborns were screened for the presence of a murmur and murmur was detected in 62 babies (2.3%) of whom 8 (45%) had a cardiac malformation.[23] Hence children having murmur should be carefully evaluated for underlying cardiac lesion and prompt early referral for an echocardiography and color doppler examination, as identification and treatment of heart disease before development of symptom offers the prospect of an improved outcome.

It is well known that extra-cardiac anomalies are associated with congenital heart disease. Associated non-cardiac malformations noted in identifiable syndromes may be seen in as many as 25% of patients with congenital heart disease. It is known that 90% of cases of trisomy 18, 50% cases of trisomy 21 and 40% cases of Turner's syndrome have congenital heart disease.

In a study conducted by Joshi et al, 10% of cases of congenital heart disease had syndromes and other associated somatic anomalies among which Down's syndrome was the commonest.[19] Similarly Khalil et al noted an incidence of 17.9% of somatic anomalies in patients with congenital heart disease.[24] In another study conducted by Kasturi L et al, 20% of cases with congenital heart disease had extra cardiac anomalies.[20]

In the present study 10% of cases had associated extra cardiac malformations in the form of imperforate anus, neonatal hepatitis syndrome and Hirschsprung's disease; out of which anorectal malformation was the most common association.

In the present study 42% of the neonates with congenital heart disease expired, and all of them had severe type of congenital heart disease like single ventricle, hypoplastic heart syndrome, tricuspid atresia, critical pulmonary stenosis, common AV canal defect and large VSD, which highlights the need for a good cardiothoracic setup in all tertiary centers, so that emergent cardiac surgery could be done neonates with severe congenital heart disease; and bring down the mortality. In a study conducted in Pakistan, 36.4% of the newborn with congenital heart disease expired and all of them had severe type of congenital heart disease.[15]

In the present study, 37% were of cyanotic type of which TGA was the commonest cyanotic heart disease followed by TOF and 63% constituted acyanotic group in which VSD was the commonest, which was comparable to a study by Shah GS, et al where in the cyanotic congenital heart disease constituted 31% and acyanotic 69%.[16] Similarly in a recent study by Bhushan Deo, et al, 32.5% belonged to cyanotic group and 67.5% belonged to acyanotic group.[17] Most of the cyanotic variety presented in the first week of life, while acyanotic lesions presented in the fourth week of life.

In a study conducted by Humayun KN, et al the mean age of presentation of neonates with congenital heart disease was 5 days and all had cyanotic type of congenital heart disease, which was similar to the observation made in the present study.[15] Hence it is evident that, most of the severe form of congenital heart disease, manifested in the first week of life and moderate to mild variety of congenital heart disease manifested towards the end of first month of life.

Many children with congenital heart disease fail to thrive from early infancy. There are several possible explanations for this, hypoxia and breathlessness may lead to feeding problems; anoxia or venous congestion of the bowel may result in malabsorption; peripheral anoxia and acidosis may lead to inefficient utilization of nutrients; and increased metabolic rate may mean that recommended energy intake is insufficient for normal growth and nutrition. [25]

Infants at particular risk of failure to thrive are those with cyanotic congenital heart disease and those with left to right shunts, pulmonary hypertension and right sided cardiac failure.[25]

In the present study out of 35 babies who survived, 35% had repeated respiratory tract infection, 21% had failure to thrive and 15% had congestive cardiac failure after a follow up for six months and majority had left to right shunt lesions. Similarly, in a study by Zachariah P, et al, the severity of Lower Respiratory Tract Infection (LRTI) in children with congenital heart disease was significantly greater than those without congenital heart disease.[26] In another study by Joshi S, et al, 44.8% of patients with congenital heart disease had respiratory tract infection and 38% had failure to thrive.[19]

CONCLUSION

The results of this study showed that majority of the neonates with suspected congenital heart disease presented with hurried respiration, refusal to feed and cyanosis. Nearly half of the cases presented in the first week of life and all of them had severe type of congenital heart disease. 37% of the cases belonged to cyanotic

variety and the remaining in acyanotic group. Transposition of Great Arteries (TGA) was the commonest cyanotic congenital heart disease while Ventricular Septal Defect (VSD) was the commonest in acyanotic group. Consanguinity was the major risk factor for congenital malformations including cardiac malformations wherein in our study nearly half of the cases were born to consanguineously married couple. Neonates with extra cardiac malformations and genetic diseases should be screened for cardiac lesions. Early recognition of CHD in the newborn is important for appropriate management and short-term follow-up for decision making regarding referral in order to reduce the infant morbidity and mortality.

Findings of this study suggest a need for larger group of neonates-based studies to accurately estimate the incidence and risk factors of cyanotic and acyanotic congenital heart disease in neonates in our country.

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REFERENCES

1. Mishra M, Mittal M, Verma AM. Prevalence and pattern of congenital heart disease in school children of Eastern Uttar Pradesh. Indian Heart J. 2009;61:58-60.
2. Bhat NK, Dhar M, Kumar R, Patel Al. Prevalence and pattern of congenital heart disease in Uttarakhand, India. Indian J Pediatr. 2013;80(4):281-5.
3. Ferenez C, Rubin JD, Mc Carter RJ. Congenital heart disease: Prevalence at live birth. The Baltimore-Washington infant study. Am J Epidemiol. 1985;121:31-6.
4. Bollistey S, Daftary A, Ewald D. Congenital heart defect in central Australia. Med J. 2004;180:614-7.
5. Wren C, Richmond S, Donaldson L. Presentation of congenital heart disease in infancy: implication for routine examination. Arch Dis Child Fetal Neonatal Ed. 1999;80:49-52.
6. Saxena A. Congenital heart disease in India: a status report. Indian J Pediatr. 2005; 72:595-8.
7. Kuehl KS, Loffredo CA, Ferenez C. Failure to diagnose congenital heart disease in infancy. Pediatr. 1999;103:743-7.
8. Zeltser I, Tabutt S. Critical heart disease in newborn. In: Bell LM, Vetter VL. Editor. Pediatric cardiology – The requisites in pediatrics. Philadelphia: Elseiver; 2006:31-33.
9. Mitchell SC, Korones SB, Berendes HW. Congenital heart disease in 56,109 births. Incidence and natural history. Circ. 1971;43:323.
10. Wechsler SB, Wernovsky G. Cardiac disorders. In: Cloherty JP Eichenwald EC, Stark AR. Editors. Manual of neonatal care. 7th ed. Philadelphia: Lippincott Williams and Wilkins; 2014:416.
11. Hoffman JIE, Kaplan S. The incidence of congenital heart disease. J Am Coll Cardiol. 2002;39:1890-900.
12. Chadha SL, Singh N, Shukla DK. Epidemiological study of congenital heart diseases. Indian J Pediatr. 2001;68:507-10.
13. Working group on management of congenital heart disease in India. Consensus in timing of intervention for common congenital heart disease. Indian Pediatr. 2008;45:117.
14. Wernovsky G, Gruber PJ. Common congenital heart disease: Presentation management and outcome. In: Taensch HW, Bakard RA, Gleason Ca. editors, Avery's diseases of the newborn. 9th Ed. Philadelphia: Saunders; 2012:765
15. Humayun KN, Atiq M. Clinical profile and outcome of cyanotic congenital heart disease in neonates. J Coll Physician Surg Pak. 2008;18:290-3.
16. Shah GS, Singh MK, Pandy TR, Kalkheti BK, Bhandari GP. Incidence of congenital heart disease in tertiary care hospital. Kathmandu Univ Med J. 2008;6:33-6.
17. Deo B, Jadhav J, Idgampalli N, Deo N, Sabale R. Study of clinical profile of congenital heart disease in paediatric age group. Indian J Basic Applied Med Res. 2015:4(4):269-72.
18. Harshangi SV. Clinical study of congenital heart disease in infants. J Pharm Sci Innovation. 2013;2(1):15-8.
19. Tank S, Malik S, Joshi S. Epidemology of congenital heart disease among hospitalized patients. Bombay Heart J. 2004;46(2):144-50.
20. Kasturi L, Kulkarni AV, Anin A, Mahashankar VA. Congenital heart disease: Clinical spectrum. Indian Pediatr. 1999;36:953.
21. Kulkarni ML, Kurian M. Consanguinity and its effect of fetal growth and development: a south Indian study. J Med Genet. 1990;27:348-52.
22. Nabulsi MM, Tamim H, Sabbagh M, Obeid MY, Yunis KA, Bitan FF. Parental consanguinity and congenital heart malformation in a developing country. Am J Med Genet A. 2003;116:342-7.
23. Bansal M, Jain H. Cardiac murmurs in neonates. Indian Pediatr. 2005;42:397-8.
24. Khalil A, Aggarwal R, Thirupuram S, Arora R. Incidence of congenital heart disease among hospital live births in India. Indian Pediatr. 1994;31:519-24.
25. Menon G, Poskitt EM. Why does congenital heart disease cause failure to thrive? Arch Dis Child. 1985;60:1134-9.
26. Zachariah P, Simoes EAF. Respiratory syncytial virus infection and congenital heart disease. South Afr Epidemiol Infect. 2008;23:17-9.

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