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ORIGINAL RESEARCH

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

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ABSTRACT

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

Material and methods: This is a hospital-based study done on 50 full term neonates suspected of congenital heart disease both inborn and referred cases admitted into Neonatal Intensive Care Unit (NICU) during the period From june 2014 to may 2016.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.

Results: Nearly half of the cases presented in the first week (44%) followed by those in fourth week(42%), third week(8%), second week(6%) in decreasing order.
the sex distribution of the neonates included in the study. 58% were male and42%were female. cardiac anomalies were present in 5 cases (10%), in which anorectal malformation was the commonest association with congenital heart disease. 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 37% of children suffered from repeated respiratory tract infection and 22% were failing to thrive and 14% had congestive cardiac failure. 30% of children thrived well and among them 4 cases were operated for PDA and TGA respectively at 2 months of age.

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

The congenital heart disease (CHD) is not fixed anatomic defects that appear at birth, but are instead a dynamic group of anomalies that originates in fetal life and changes considerable during the postnatal development.[1] The incidence of moderate-to-severe structural CHD in liveborn infant is 6–8 per 1000 live births.[2] About 2–3 per 1000 newborns 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. CHD is considered one of the leading causes of neonatal

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mortality.[3] According to a status report on CHD in India, 10% of the present infant mortality may be accounted to CHD.[4] Many cases are asymptomatic and discovered incidentally during routine health checkup.[5] The neonates with CHD may present with, feeding difficulty, fast breathing, cyanosis, cardiovascular collapse, and congestive heart failure or combinations of these presentations. Pure versions of specific defects may present in some patients, but many neonates have various combinations of defects.[6] The initial evaluation of any newborn suspected of having critical CHD includes a meticulous physical examination, four extremity blood pressures, preductal and postductal saturations, hyperoxia test, and chest radiograph. Echocardiography, with Doppler and color Doppler, has become the primary diagnostic tool for CHD. In addition, it reduces the requirement for invasive studies such as cardiac catheterization.[7-9]

Material and methods

This is a hospital-based study done on 50 full term neonates suspected of congenital heart disease both inborn and referred cases admitted into Neonatal Intensive Care Unit (NICU) of Department of Pediatrics SKIMS Soura during the period From june 2014 to may2016. 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 (44%) followed by those in fourth week(42%), third week(8%), second week(6%) in decreasing order.

Table1: Age at presentation of new born with CHD.

Age	No. of cases (n)	(%)
Week1	22	44
Week2	3	6
Week3	4	8
Week4	21	42

Table 2 shows the sex distribution of the neonates included in the study. 58% were male and 42% were female.

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

Sex	No of Cases (n)	Percentage (%)
Male	29	58
Female	21	42

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Out of 50 cases studied, 18cases were born to consanguineously married couples as evident from Table3.

Table3: Consanguinity history in the parents of neonates with CHD.

	No. of cases (n)	Percentage
Consanguineous marriage	18	36
Non Consanguineous marriage	32	64

Table4: Neonates presenting with murmur.

Murmur	No. of cases(n)	Percentage (%)
Present	38	76
Absent	12	24

Table5: Extra cardiac anomalies in neonates with CHD.

Associated anomalies	No. of cases(n)	Percentage (%)
Anorectal malformations(ARM)	2	4
Hiruchsprung disease(HPD)	2	4
Neonatal hepatitis syndrome(NHS)	1	2
Total present(extracardiac anomalies)	5	10
Absent	45	90

Murmur is an impressive presentation of congenital heart disease. As shown in Table 4, 76% 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 5 cases (10%), in which anorectal malformation was the commonest association with congenital heart disease.

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 (66%) were acyanotic type, remaining being cyanotic

Table6: Various types of congenital heart diseases

Type of heart disease	Total (n)	%
Cyanotic	**17**	**34**
Acyanotic	**33**	**66**

Table7: Follow up of the CHD neonates and their complaints.

Complaints	No. of cases (n)	(%)
FTT(Failure to thrive)	6	20
RTI(Respiratory tract infections)	11	37
CCF(Congestive cardiac failure)	4	14
Normal	9	30

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.

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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 37% of children suffered from repeated respiratory tract infection and 22% were failing to thrive and 14% had congestive cardiac failure. 30% 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 50 newborns, both inborn and referred to Neonatal Intensive Care Unit(NICU) SKIMS, 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 respiratory distress (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

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respiratory distress, seen in 78% of cases.[18] In a study conducted by Joshi et al in Mumbai, the commonest symptoms was respiratory distress, 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 respiratory distres 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, 18 neonates (36%) 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, 38 babies (76%) had murmur and 12 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, 34% were of cyanotic type of which TGA was the commonest cyanotic heart disease followedby TOF and 66% constituted acyanotic group in which VSD was the commonest, which was comparable to astudy by Shah GS, et al where in the cyanotic congenital heart disease constituted 31% and acyanotic 69%.[16]Similarly in a recent study by BhushanDeo, et al, 32.5% belonged to cyanotic group and 67.5% belonged to acyanotic group.[17] Most of the cyanotic variety presentedin 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

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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 30 babies who survived, 36% had repeated respiratory tract infection, 20% had failure to thrive and 14% 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]

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.

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