

Clinical Presentation, Diagnosis and Outcome of Neonates with Suspicion of Congenital Heart Disease in a Tertiary Care Hospital

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Abstract

Objective: To determine correlation of various signs and symptoms with echocardiography findings in neonates. Short term outcome of patients with confirmed congenital heart disease (CHD).

Study design, settings and duration: A comparative cross sectional study was carried out at Department of Cardiology and Neonatology, the Children Hospital & the Institute of Child Health, Lahore from January to December 2016.

Subjects and Methods: A total of 514 neonates with clinical suspicion of CHD were included and underwent echocardiography. The neonates who had persistent pulmonary hypertension, patent ductus arteriosus less than 3 days of age and patent foramen ovale were excluded. The neonates with confirmed CHD were followed up for hospital outcome. Data was collected on a pre-designed proforma. Statistical analysis was performed using SPSS 20.

Results: The mean age was 6.1 (0-28) days. CHD was present in 70 (13.61%) patients; 44 (63%) males and 26 (37%) females. Of these, 38 (54.28%) had cyanotic CHD while 32 (45.71%) had acyanotic CHD. Among the neonates having CHD, there were 41 (58.57%) preterm. Tachypnea (68.75%) and heart murmur (65.62%) were statistically significant with clinical signs in acyanotic heart disease while cyanosis (97.36%), tachypnea (RR >60) (92.10%) and heart murmur (34.21%) had significant association with cyanotic heart disease. Ventricular septal defect (VSD) was the commonest acyanotic CHD 8/32 (25%) whilst transposition of the great arteries (TGA) with VSD was the commonest cyanotic CHD 8/38 (21%). Fourteen neonates had critical congenital heart disease undergoing surgery or intervention 14/70 (20%). Three neonates with co-arctation of the aorta underwent surgery with one death. Six neonates for critical aortic and pulmonary stenosis underwent successful balloon valvuloplasty. Four patients with TGA intact septum had successful balloon atrial septostomy. One neonate with infra-cardiac total anomalous pulmonary venous connection (TAPVC) underwent surgery but expired. The overall hospital mortality was 2.85%.

Conclusion: Congenital heart disease was present in only 13.61% of the referred newborns with suspected CHD. The yield was high in preterm babies and for cyanotic CHD. Cyanosis and tachypnea, were major clinical signs pointing towards CHD. The neonates with critical congenital heart disease had a high mortality.

Key words: Congenital heart disease, echocardiography, neonate.

Introduction

Congenital heart disease (CHD) is the most common congenital disorder in newborns.¹⁻³ Many newborns with critical CHD are symptomatic and recognized soon after birth, others are diagnosed after discharge from hospital.⁴⁻⁷ The true incidence and prevalence is difficult to know as it involves large populations and some heart diseases are extremely rare while others do not have any signs or symptoms in newborns (e.g., ASD).⁸ Studies show an overall incidence of CHD range from 0.8 to 3.5/1000 live births.⁹⁻¹¹ Incidence is higher in preterm infants i.e. about 12.5/1000 pre-term infants.¹² In a hospital based study from India, 1.56% of hospital admissions were with congenital

heart disease. Ventricular septal defect (VSD) was the commonest acyanotic CHD (58.14%) followed by atrial septal defect (19%), patent ductus

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MS conceptualized the project. Literature search was done by IA & TK. IA & NH performed the statistical analysis. IA also did the data collection, drafting, revision and writing of manuscript.

arteriosus (9.3%), atrioventricular septal defect (9.3%), aortic stenosis (7%) and co-arcuation of the aorta (2.32%). Tetralogy of Fallot was the most common cyanotic CHD (35.29%) followed by transposition of great arteries (17.65%).¹³

Clinical presentation of CHD is variable depending upon age, severity and type of lesion. In newborns, the presentation is variable and may be non-specific. There may be no obvious signs of respiratory distress or cardiac lesion and the newborn may present with pallor and/or hypotension. Heart murmur may also be absent especially in cyanotic CHD and the clinical features of CHD may overlap with those of other illness.¹⁴

The study was to correlate with definitive echocardiography findings of neonates presenting in a tertiary care hospital. To help the general practitioners to identify signs and symptoms for proper referral. The hospital outcome was also measured so that guidelines may be suggested for referral of these neonates with variable symptoms and signs.

Subjects and Methods

This descriptive study was performed in the department of Pediatric Cardiology and Neonatology of The Children's Hospital and Institute of Child Health Lahore from January to December 2016. All the neonates were enrolled in study except patients with pulmonary hypertension (PPHN), patent ductus arteriosus (PDA) with age less than 3 days and patent foramen ovale (PFO) after taking written informed consent from parents/guardians. Data regarding age, gender, clinical signs and symptoms, hospital course and outcome (in form of discharge from hospital, surgery/ intervention, Lama, death) were noted on a Proforma. Detailed cardiac examination was carried out. Echocardiography of all patients was done at department of cardiology, by a consultant pediatric cardiologist according to hospital management protocols. All the patients were followed in hospital till discharge or death. Data was analyzed using SPSS 20th version. Chi square test were applied for any significance and *p*-value <0.05 were taken as significant.

Results

A total of 514 patients were evaluated. Male to female ratio was 1.4:1. Mean age of presentation was 6.1 days (range 0-28 days). Congenital heart disease was present in 70 out of 514 (13.61%). Among the neonates having congenital heart disease, there were 41 (58.57%) preterm and 29

(41.42%) full term. Amongst the babies having CHD, there were 44 (63%) males and 26 (37%) females. Cyanotic CHD was present in 38 out of 70 (54.28%) whilst 32 out of 70 (45.71%) had acyanotic CHD.

Table 1: Symptoms of acyanotic heart disease.

| Symptoms: | Acyanotic Heart Disease (N=32) % | Normal (N=444) % | <i>p</i> -value |
|----------------------|-------------------------------------|---------------------|-----------------|
| Respiratory distress | 84.37 | 96.39 | 0.001 |
| Lethargy | 46.87 | 50.00 | 0.733 |
| Reluctance to feed | 43.75 | 48.64 | 0.592 |
| Interrupted feeding | 25.00 | 3.60 | 0.000 |
| Sweating during feed | 15.62 | 0.00 | 0.000 |

Table 2: Signs of acyanotic heart disease.

| Signs | Acyanotic Heart Disease (N=32) % | Normal (N=444) % | <i>p</i> -value |
|-----------------------|-------------------------------------|---------------------|-----------------|
| Tachypnea | 68.75 | 98.87 | <0.005 |
| Hepatomegaly | 15.62 | 9.23 | 0.273 |
| Murmur | 65.62 | 20.72 | 0.001 |
| Capillary refill time | 3.12 | 7.43 | 0.361 |

Table 3: Symptoms of cyanotic heart disease.

| Symptoms | Cyanotic Heart Disease (N=38) % | Normal (N=444) % | <i>p</i> -value |
|------------------------------|------------------------------------|---------------------|-----------------|
| Bluish discoloration of skin | 97.36 | 54.50 | 0.000 |
| Shortness of breath | 92.10 | 96.39 | 0.192 |
| Reluctance to feed | 57.89 | 48.64 | 0.274 |
| Lethargy | 50.00 | 50.00 | 1.000 |
| Interrupted feeding | 10.52 | 3.60 | 0.04 |
| Sweating during feed | 2.63 | 0.00 | 0.01 |

Table 4: Signs of cyanotic heart disease.

| Signs: | Cyanotic Heart Disease (N=38) % | Normal (N=444) % | <i>p</i> -value |
|---------------|------------------------------------|---------------------|-----------------|
| Cyanosis | 97.36 | 54.50 | 0.000 |
| Tachypnea | 92.10 | 98.87 | 0.002 |
| Murmur | 34.21 | 20.72 | 0.053 |
| Prolonged CRT | 10.52 | 7.43 | 0.002 |
| Hepatomegaly | 5.26 | 9.90 | 0.410 |

Most significant symptom of acyanotic heart disease was sweating during feed, interrupted feeding and respiratory difficulty and signs were tachypnea and murmur. In contrast in cyanotic heart disease, bluish discoloration was present in almost all patients with signs of tachypnea and cyanosis (Table1-4).

Ventricular septal defect (VSD) 8/32 (25%) was the commonest acyanotic CHD followed by VSD and PDA 5/32 (15%) (Table-5).

Table 5: Congenital acyanotic heart disease distribution. (N=32)

| | N (%) |
|--------------------|--------|
| VSD | 8 (25) |
| VSD plus PDA | 5 (15) |
| ASD | 5 (15) |
| ASD plus VSD | 4 (12) |
| AP window ,VSD | 1 (3) |
| COA | 3 (9) |
| Aortic stenosis | 2 (6) |
| CAVSD | 1 (3) |
| HCM | 2 (6) |
| Pulmonary stenosis | 1 (3) |

Table 6: Congenital cyanotic heart disease distribution. (N=38)

| Lesion | N (%) |
|------------------------------|--------|
| TGA,VSD | 8 (21) |
| TGA with Intact Septum | 4 (10) |
| Pulmonary Atresia, VSD,PDA | 5 (13) |
| TOF | 9 (23) |
| TA,ASD,VSD,PS | 5 (13) |
| Dextrocardia, TGA, VSD, PS | 2 (5) |
| CAVSD UVH TGA PS | 1 (2) |
| Dextrocardia, CAVSD, PA, PDA | 2 (5) |
| Type1 truncus arteriosus | 1 (2) |
| Infra cardiac TAPVC | 1 (2) |

Transposition of great arteries with VSD 8/38 (21%) was the commonest cyanotic CHD followed by TGA with intact septum 4/38 (10%) (Table-6).

Table 7: Required active intervention. (N=70)

| Diseases | No Intervention Required N (%) | Intervention Required (Surgery/Angiography N (%)) |
|------------|-----------------------------------|---|
| Cyanotic | 33 (47.14) | 5 (7.14) |
| Acyanotic | 26 (37.14) | 6 (8.57) |
| Discharged | 59 (84.28) | 9 (12.85) |
| Expired | - | 2 (2.85) |

Eleven out of 70 patients underwent intervention (surgery /angiography), out of which 9 patients were discharged and 2 expired. All neonates diagnosed as coarctation of the aorta underwent surgery; two of them had uneventful post operative course but unfortunately one neonate expired due to co morbid condition. Neonates with critical aortic and pulmonary stenosis underwent successful balloon valvuloplasty. Four patients with transposition of great arteries with intact

interventricular septum had successfully balloon atrial septostomy. Neonate with infracardiac TAPVC underwent surgery and expired in immediate post operative period. The overall mortality was 2.85% (3/70). Rest of the neonates was put under close follow up after discharge (Table-7).

Discussion

Congenital heart defects are the commonest of all birth defects. They are amongst the leading causes of morbidity and mortality. We included 514 neonates in the study who were referred to tertiary care facility with suspicion of congenital heart disease. Out of which 70 (13.61%) had echo-cardio graphically confirmed congenital heart disease. Manjuleswari has calculated a frequency of 1.5%¹⁵ while Yasmeen found a frequency of 8.4% with similar study designs.¹⁶ A local study found a prevalence of 17.1% in neonatal age group.

Among the babies having congenital heart disease, there were 44 (62%) males and 27 (38%) females. This pattern of gender distribution was consistent with other studies and may be attributed to gender bias with males being more affected.^{15,17,18}

The babies with CHD revealed that 41 were (57.7%) preterm and 30 were (42.3%) full term. A similar observation was noted in a local as well as English study. Tanner attributes this pattern to increased surveillance of preterm in early life.^{12,18}

Cyanotic heart disease was seen in 54.28% cases. While acyanotic heart disease was diagnosed in 45.71% which is in contrast to similar studies.¹⁶⁻¹⁸ Local studies conducted in Rawalpindi and Hyderabad found that congenital acyanotic heart disease is more common with prevalence of 75.55% and 74.6% respectively. This difference in results may be due to greater scrutiny of cyanotic babies for congenital heart disease by referring them to physicians.

The commonest congenital acyanotic heart diseases were ventricular septal defect (VSD), atrial septal defect (ASD), combined VSD with patent ductus arteriosus (PDA), while tetralogy of fallot (TOF) was commonest cyanotic heart disease followed by transportation of the great arteries (TGA), VSD and pulmonary artery (PA), ASD, VSD, PDA. The pattern of Acyanotic heart disease is consistent with a local study conducted at Rawalpindi and Hyderabad¹⁶⁻¹⁸ however, a Korean study found ASD to be the commonest acyanotic cardiac lesion.¹⁹ A wide variation has been noted in Cyanotic group i.e., TGA was most common diagnosis in an Indian study¹⁵ while TOF was most

common lesion in local studies followed by PS and TGA.¹⁶⁻¹⁸

The common symptoms noted in acyanotic heart disease were respiratory distress, lethargy and reluctance to feed while tachypnea, hepatomegaly and murmur were most frequent signs.

We observed bluish discoloration of skin, respiratory distress and reluctance to feed are commonly associated symptoms in cyanotic heart disease while cyanosis, tachypnea and murmur were most frequently noticed signs in cyanotic group. According to Indian study, breathlessness, recurrent chest infections and congestive cardiac failure (CCF) were commonly associated with acyanotic heart disease while cyanosis, breathlessness and cyanotic spells were common clinical presentations of cyanotic heart disease. However the groups were not studied in separately for signs and symptoms.²⁰ Similarly breathlessness, feeding difficulty and cough were common clinical presentations in a local study but the study has limitations of small sample size and age range for up to 5 years.¹⁷ A local study conducted at Liaquat Hospital has focused the symptoms alone labelling recurrent chest infections, dyspnea and hypoxic spells being commonest among patients of congenital heart disease, however it differs from our study regarding age range of up to 12 years including only 7 neonatal patients.¹⁹

In our study, mortality is 2.85% in contrast to study conducted in India 19.73%.²⁰

Among suspected cases, congenital heart disease was present only in 13.8% cases referred to tertiary care children hospital. The yield was high in preterm babies and for cyanotic CHD. Cyanosis, tachypnea, sweating during feed was major signs and symptoms. Neonatal murmur was present in 34% of the patient.

Hence, we recommend that all murmurs should be screened unless thought to be physiological. Early diagnosis and timely management are key factors for optimal outcome of this problem. A high index of suspicion, a detailed history, physical examination, chest X-ray and along with the use of 2-D-Echocardiography helps us in diagnosing most of the cases of congenital heart disease. Close monitoring and timely intervention in cases of congenital heart disease will go a long way in reducing the morbidity and mortality to a large extent and can significantly change the outcome of congenital heart disease.

Conflict of interest: None declared.

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