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Research Article

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Clinical Profile and Pattern of Congenital Heart Disease in Neonates and Immediate Outcome Based on Oxygen Saturation

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ABSTRACT

Background: The most prevalent kind of congenital defect in infants is congenital heart disease (CHD). Early diagnosis of CHD is very important for timely management. Age of presentation varies on the type and severity of the defect. In the absence of visible cyanosis many cyanotic CHD could be misdiagnosed as acyanotic CHD. The main aim of the present study was to study the clinical profile and CHD pattern based on functional classification and their immediate outcome based on oxygen saturation (SpO_2).

Methods: A prospective observational study was conducted in a tertiary health care centre in central India. Neonates with clinical features of CHD and diagnostically confirmed by echocardiography were included. Neonates with CHD were categorized based on various parameters and functional classification and their immediate outcome was interpreted based on SpO_2.

Results: A total of 126 patients were detected to have significant congenital heart disease. Most of the neonates presented in 4^{th} week of life. The most common presentation was tachypnea followed by cyanosis and shock. Among acyanotic CHD, ventricular septal defect (VSD) was the most common while the commonest cyanotic CHD was transposition of great arteries (TGA). About $3/4^{th}$ of patients had SpO2 > 90%, and only about 12 (10%) had SpO_2 below 85%.

Conclusion: Acyanotic CHD presents later in neonatal life compared to cyanotic CHD. Most of the neonates with CHD, who were in cyanotic group had SpO_2 between 85-90% without any visible cyanosis. Outcome of CHD in neonates is poor with increasing hypoxemia.

Key-words: Congenital heart disease, Cyanosis, Neonate, Outcome, Oxygen saturation

INTRODUCTION

CHD is a structural anomaly of heart present since birth. It is the most common congenital anomaly affecting 28% of all major congenital disabilities [1].

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This condition is most commonly observed in newborns, infants, or children, but untreated CHD can also be seen in adults [2]. The prevalence of CHD varies across different regions of India ranging from 0.77 to 26.4 per 1000 individuals depending upon age and setting of the study group [3]. Early detection of CHD in newborns is crucial for timely management and referral to a cardiac centre. Delayed diagnosis can lead to delayed treatment, which is especially problematic in developing countries, where many neonates with CHD may not receive proper care resulting in higher morbidity and mortality rates [4]. Cyanosis is a clinical sign of hypoxemia but hypoxemia

does not always lead to cyanosis [5]. According to the functional classification of CHD, cyanotic CHD with increased pulmonary blood flow could be missed or clinically diagnosed as acyanotic CHD, if there is mild hypoxemia and SpO_2 level is between 85-90%, because cyanosis is not visible in mild hypoxemia [6]. The clinical profile and pattern of CHD in such neonates were studied. Additionally, our study focused on outcomes based on SpO_2 values, which may provide valuable insights into the prognosis of neonates with CHD.

MATERIALS AND METHODS

Study Design and Participants- A prospective observational study was conducted in the Pediatrics Department of a tertiary care center in central India between September 2021 and March 2023. The primary objective of the study was to determine the clinical profile of neonates who displayed clinical features suggestive of CHD and whose diagnosis was confirmed by Echocardiography. The secondary objective was to assess immediate outcomes after one month based on SpO_2 in neonates diagnosed with CHD. A total of 300 neonates were identified as having symptoms or signs suggestive of CHD during the study period. A transthoracic echocardiogram (TTE) was performed to confirm the diagnosis of CHD. TTE was performed by a trained cardiologist using PHILIPS HD 11 echo machine with neonatal probe (S-12) using two-dimensional, color Doppler, M mode, pulse and continuous wave echocardiogram. About 174 neonates were excluded from the study as per exclusion criteria. Informed consent was obtained from the parents of the neonates.

Methodology- Patient information was gathered using a specific proforma. Neonates who were admitted underwent a detailed history assessment and a comprehensive clinical examination. Neonates were considered positive screening cases for CHD if they exhibited certain signs and symptoms, including heart murmurs, tachypnea, abnormal transdermal SpO_2, differences in SpO_2 between upper and lower limbs, and poor peripheral pulses. Tachypnea in neonate is defined as a respiratory rate of more than 60/minute. All patients underwent diagnostic tests, including routine hematological investigations, chest X-rays, Electrocardiogram (ECG), and Echocardiography. Subjects were included in the study based on the findings from echocardiography, which is a key diagnostic tool for identifying CHD. Neonates with CHD were categorized into two groups—acyanotic and cyanotic, based on the functional classification of CHD [7,8]. Cyanotic CHD patients were classified as decreased pulmonary blood flow when shunt lesions associated with significant right ventricular outflow obstruction and complete mixing lesions without significant right ventricular outflow obstruction were classified under cyanotic CHD with increased pulmonary blood flow. Visible cyanosis is seen when saturation is below 85% [6]. Many of the cyanotic CHD could be missed or misdiagnosed clinically as acyanotic CHD if we don't check the SpO_2 by pulse oximetry. In study, patients were divided into three groups based on SpO_2 levels, SpO_2 level >90%, between 85 & 90%, and <85%. A pulse oximeter was applied on either foot to avoid missing low SpO_2 in differential cyanosis. Various outcomes of patients like discharge with a plan to follow up, referral to a higher centre and death were assessed for 1 month.

Inclusion criteria

- Neonates with symptoms or signs suggestive of CHD.
- Diagnostically confirmed cases of CHD by Echocardiography.

Exclusion criteria

- Neonates presenting with Congestive Cardiac Failure (CCF) unrelated to CHD.
- Neonates with specific heart conditions like patent foramen ovale, and patent ductus arteriosus (PDA) detected within the first 72 hours of life.
- Hemodynamically insignificant PDA, those with bicuspid aortic valve.
- Neonates with persistent pulmonary hypertension but a structurally normal heart.

Statistical Analysis- Data was collected in the Microsoft Excel program and statistical analysis was performed by the SPSS program for Windows, version 25 (IBM Corp., Armonk, NY, USA). Two sample proportion and Chi-square tests were applied, and $p<0.05$ was considered significant.

Ethical consideration- Ethical approval for the study was obtained from the institutional ethics committee (118/IEC/GMC/2021) of the tertiary care hospital.

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RESULTS

The present study involved a total of 300 neonates showing symptoms or signs suggestive of CHD. Echocardiography was performed on all 300 neonates, and after applying exclusion criteria, 126 patients were found to have significant CHD. Most CHD cases were found in males, with a male-to-female ratio of 1.5:1. The most common presentations of CHD were fast breathing, followed by cyanosis and shock. About 9.5% of neonates with CHD had no cardiac complaints but were identified through the presence of a murmur upon examination (Table 1). Most neonates presented with CHD in the 4th week of life, followed by 3rd week. Almost equal number of neonates with CHD presented in 1st and 2nd week of life (Table 2).

Table 1: Primary presenting complaints of patients with CHD

Primary presenting complaint	Number (%)
Fast breathing	67(53.2)
Cyanosis	33(26.2)
Shock	14(11.1)
Murmur	12(9.5)

Table 2: Age of presentation of patients with CHD.

Age of presentation	Number (%)
1st week	24(19)
2nd week	21(16.6)
3rd week	31(24.6)
4th week	50(39.7)

Approximately two-thirds of neonates had acyanotic CHD, with the most common time of presentation in the 4th week. In contrast, the most common presentation time for cyanotic CHD was the 1st week. The most common acyanotic CHD was Ventricular Septal Defect (VSD) followed by PDA and atrioventricular septal defect (AVSD). Coarctation of aorta and critical aortic stenosis (AS) were one in number. More than three-fourths of all acyanotic CHD patients presented in 2nd half of neonatal life and less than 10% given in the first week of life (Table 3).

Table 3: Pattern and age of presentation of different types of ACHD

Type of ACHD	1st week (n=6)	2nd week (n=14)	3rd week (n=21)	4th week (n=44)	Total (%) (N=85)
VSD	6	7	15	31	59(69.4)
PDA	0	2	2	6	10 (11.7)
AVSD	0	2	0	4	6 (7)
Valvar PS	0	0	2	1	3 (3.5)
ASD	0	0	1	2	3(3.5)
CCTGA with VSD	0	2	0	0	2(2.3)
Coarctation of aorta	0	0	1	0	1 (1.1)
Critical AS	0	1	0	0	1 (1.1)

ACHD: acyanotic congenital heart disease; AVSD: atrioventricular septal defect; AS: aortic stenosis; PS: pulmonary stenosis; CCTGA: congenitally corrected transposition of great arteries; ASD: atrial septal defect

A total of 41(32.5%) neonates had cyanotic CHD. Transposition of great arteries (TGA) was the most common cyanotic CHD followed by Tetralogy of Fallot (TOF). Single ventricle and Total anomalous pulmonary venous return (TAPVR) were equal. There was one case for each truncus arteriosus and Ebstein anomaly. Almost 18 (50%) of cyanotic CHD cases were presented in first week of life (Table 4).

Table 4: Pattern and age of presentation of different types of CCHD.

Type of CCHD	1st week (n=18)	2nd week (n=7)	3rd week (n=10)	4th week (n=6)	Number (%)(N=41)
d-TGA with Intact IVS	2	2	1	0	5 (12.2)
d-TGA with VSD with PS	1	1	0	1	3 (7.3)
d-TGA with VSD	0	0	2	1	3 (7.3)
TOF	2	0	4	2	8 (19.5)
Single ventricle with PS	1	1	0	0	2 (4.8)
Single ventricle without PS	1	0	0	1	2 (4.8)
TAPVR	1	1	1	1	4 (9.7)
TA with PA/PS	2	0	0	0	2 (4.8)
TA without PS	1	0	0	0	1 (2.4)
VSD with PA	2	1	0	0	3 (7.3)
Critical PS with ASD	1	0	0	0	1 (2.4)
HLHS	2	1	0	0	3 (7.3)
AVSD with PS	1	0	1	0	2 (4.8)

CCHD: cyanotic congenital heart disease; TGA: transposition of great arteries; TOF: tetralogy of Fallot; TAPVR: total anomalous pulmonary venous return; TA: tricuspid atresia; PA: pulmonary atresia; HLHS: hypoplastic left heart syndrome

According to the functional classification of CHD, more than 90% (80) of acyanotic CHD cases were categorized as left to right shunt lesions (p=0.001). In cyanotic CHD cases, lesions with increased and decreased pulmonary blood flow were almost equally prevalent (p= 0.592) (Table 5). About three-fourths of patients had SpO_2 greater than 90%, while only 12 (10%) patients had SpO_2 below 85%.

Table 5: Distribution of patients according to functional classification of CHD.

Types of CHD	Functional classification of CHD	Number (%) (N=126)	p-value
Acyanotic(n=85)	Left to Right Shunt lesions	80(63.5)	0.01
	Obstructive lesions	5(3.9)	
Cyanotic (n=41)	Decreased pulmonary blood flow	22(17.5)	0.592
	Increased pulmonary blood flow (Complete mixing lesions)	19(15.1)	

Out of the total number of patients, 71.4% were discharged from the hospital after receiving treatment. About 22.2% of the patients unfortunately did not survive and passed away due to their illnesses. Eight neonates (6.3% of the patients) needed specialized care and were referred to a different medical facility for further treatment.

Patients, whose SpO_2 levels were above 90% were more likely to be discharged (p=0.01) (Table 6). Two-thirds of the patients whose SpO_2 levels were below 85% did not survive. Again, there was a significant difference between the outcome of patients with SpO_2 is 85-90% and those with SpO_2<85% showing poorer outcomes with lower SpO_2 (p= 0.01).

Table 6: Distribution and outcome of patients based on SpO_2

Outcome-based on SpO_2 Number (%) N=126	SpO_2>90% (n=96)	SpO_2 85-90% (n=18)	SpO_2>90% (n=12)	p-value
Discharged 90 (71.4)	85	5	0	0.01 (Sig)
Referred to higher center 8 (6.3)	1	3	4	
Death 28(22.2)	10	10	8	

DISCUSSION

The prevalence of CHD in India is similar to the worldwide prevalence. However, due to the high birth rate in India, there is a significant burden of CHD in the country [9]. The newborn intensive care unit is the best place for screening and diagnosing CHD. Unfortunately, many healthcare staff in Central India, including nurses, physicians, and other health workers, lack training in recognizing CHD. Early detection of CHD in newborns is indeed crucial because timely intervention can be life-saving. In our study, the male-to-female ratio for CHD was 1.5:1, with males being predominant. This finding is consistent with the study of Abou-Taleb [10] from Egypt. Most cases of CHD in our study were presented in the 4th week of life.

Additionally, about two-thirds of the patients had acyanotic CHD. Delayed presentation of acyanotic CHD may be due to unregressed neonatal pulmonary hypertension in the first few weeks of life [11]. Duct-dependent lesions and conditions like TGA with poor mixing of blood tend to present early in neonatal life.

Almost similar findings were seen by Mir *et al*. [12] of Srinagar where they also found most of the cases in the later age of neonatal life and had acyanotic CHD as a predominant one. In our study, about half of cyanotic CHD cases were presented in the first week of life. This is consistent with findings from other studies, such as one by Humayun and Atiq [13] of Pakistan where the mean age of presentation for cyanotic CHD was 5 days. In this study, almost half of the cases presented with tachypnea followed by cyanosis in about 25% of cases. These findings align with the results of other studies, including those of Mir *et al*. [12], Molaei *et al*. [14], and Ravilala *et al*. [15], where fast breathing was the most common presenting symptom followed by cyanosis. Although most of the studies have included feeding difficulty as a presenting symptom, we did not find feeding difficulty as the only primary complaint in our cases. In our observation, all cases with feeding difficulty had fast breathing. In our study, we found shock as the third most common presenting symptom and this was observed almost equally in both acyanotic and cyanotic CHD.

Among acyanotic CHD cases, VSD was the most common defect, followed by PDA and AVSD. However, ASD was only detected in 3.5% of cases, which is different from studies of Mir *et al*. [12] and Ravilala *et al*. [15], where ASD was reported as the second most common CHD. This disparity could be different study populations and transitional circulation in neonatal age. The symptomatic presentation of acyanotic CHD such as ASD, small to moderate VSD, or PDA, in neonatal life is less common because of unregressed neonatal pulmonary hypertension. This might explain why ASD was less frequently detected in this study. The critical AS in our study presented in 1st week of life and had symptoms of CCF with severe LV systolic dysfunction. In our study among cyanotic CHD, TGA was the most common defect, like studies of Saxena *et al*. [9], Taleb *et al*. [0, Mir *et al.* [12] and Molaei *et al*. [14]. TGA with intact ventricular septum is where the aorta and pulmonary artery are switched, leading to two separate circulatory systems. This typically results in cyanosis in early neonatal life because the parallel circulation doesn't provide adequate oxygenation. Other TGA patients may present later when the mixing of oxygenated and deoxygenated blood is adequate. TOF is the second most common cyanotic CHD in this study. This finding is consistent with the study done by Mir *et al*. [12], while the study done by Saxena *et al.* [9] and Talib *et al*. [10] showed complex cyanotic heart disease or hypoplastic left heart syndrome as the second most common. These discrepancies could arise from differences in the study populations or less number of total cyanotic CHD in our study. In this study, patients were divided into three different groups based on their

SpO_2 levels. Central cyanosis, the bluish skin and mucous membranes discolouration, typically becomes visible when SpO_2 falls below 85% [6]. Patients with left-to-right shunt lesions generally had SpO_2 levels above 90%. Complete mixing lesions were more predominant in the 85-90% SpO_2 group. In our study, some patients with complete mixing lesions were classified as having cyanotic CHD based on functional classification, but they did not exhibit visible cyanosis. Patients with complete mixing lesions and increased pulmonary blood flow may have higher SpO_2 and cyanosis could be missed in such cases [11].

In the present study although 41 patients had cyanotic CHD based on functional classification, only 12(10%) had saturation below 85%. Some patients with significant right ventricular outflow obstruction had SpO_2 of more than 85%, which could be because of PDA or aorto-pulmonary collaterals connecting the distal to significant obstruction. Those with duct-dependent pulmonary circulation lesions, poor mixing of blood, and significant obstruction of the right ventricular outflow tract were mainly found in the group with SpO_2 levels below 85%. The study highlighted the importance of measuring SpO_2 levels in diagnosing different types of CHD.

Eight(6.3%) patients required referral to higher-level medical centers. This decision had been based on factors such as the need for early intervention, lack of improvement with medical management, or parental preference for treatment at a more specialized facility. This study compared its overall mortality rate 22.2%(28) with the rates reported in other studies by Mir *et al.* [12] (19.1%), Ravilala *et al*.[15] (42%). In our study 18(60%) neonates with CHD died within 1 month compared to 40.8% of Mir *et al*. [12], 36.4% of Humayun and Atiq [13] and 50% of Ravilala *et al*. [15]. We found significant differences in mortality rates between different studies, and this could be due to variations in patient characteristics and study populations or the presence of unaccounted factors like sepsis or other medical conditions. The patients who experienced higher mortality rates (66.6%) were those with poor mixing or duct-dependent circulation, and their SpO_2 levels were less than 85%. This shows that patients with lesser SpO_2 have poorer outcomes [16,17]. Similar finding was also seen by Jain *et al*. in their study.

The sample size is small and many other confounding factors like the weight of the neonate, gestational age, sepsis, and lung pathology like pneumonia or respiratory distress syndrome were not considered. Further studies are required to draw a clear picture.

CONCLUSIONS

The clinical profile and pattern of CHD in neonates can vary and the immediate outcome depends on factors such as type and severity of the defect. Most of the neonates with CHD with increased pulmonary blood flow or complete mixing lesions who were in the cyanotic group as per functional classification had SpO_2 values between 85-90% without any visible cyanosis. Oxygen saturation monitoring, especially through pulse oximetry screening, is crucial in identifying neonates with CHD, guiding their management, and ensuring timely intervention when necessary.

The higher the SpO_2 value in neonate with CHD, the better the outcome. More studies with large sample sizes are needed to confirm these findings further.

CONTRIBUTION OF AUTHORS

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