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A Study of Clinical Profile Morbidities and Mortalities of Congenital Heart Disease

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ABSTRACT

Congenital heart diseases (CHDs) are the most prevalent congenital anomalies, accounting for 28% of global cases. In India, the incidence of CHD is approximately 8 per 1000 live births, similar to Europe but lower than some reported ranges globally. CHDs, presenting with varied symptoms from asymptomatic findings to severe cyanosis and heart failure, are significant contributors to neonatal mortality. Advances in diagnostics and government initiatives like the Rashtriya Bal Swasthya Karyakram (RBSK) aim to enhance early detection and improve outcomes. A prospective study conducted over two years in a tertiary care pediatric department analyzed CHD cases among hospitalized patients under 12 years. Diagnosis was confirmed through 2D echocardiography, supported by electrocardiography and oxygen saturation measurements. Demographic, clinical and laboratory data were systematically recorded and analyzed using statistical tests. Of 18,866 admissions, 290 cases were diagnosed with CHD (1.53%). Acyanotic CHD accounted for 74.1%, predominantly ventricular septal defect (39.5%), while cyanotic CHD comprised 25.9%, with Tetralogy of Fallot being most common. The highest incidence was in the 1-month to 1-year age group for acyanotic CHD (38.1%) and under 1 month for cyanotic CHD (40%). Male predominance (54.5%) was observed. Common symptoms included breathlessness (63.4%) and cough (54.4%). Malnutrition was more significant in cyanotic CHD cases (p=0.0001). Complications like congestive cardiac failure (55.5%) and pulmonary hypertension (17.58%) were frequently encountered. The incidence of CHD in the study aligns with global trends but highlights delayed diagnosis and management in resource-limited settings. Enhanced awareness, early intervention and improved access to specialized care are imperative to reduce morbidity and mortality.

INTRODUCTION

Congenital heart diseases (CHDs) are the most common major congenital anomalies, representing 28% of such anomalies globally[1,2]. The incidence of CHD is estimated at 8 per 1000 live births in India[3], while in the United States, CHD affects approximately 1% of live births[3,4]. In Europe, the total prevalence is 8 per 1000 births, with 7.2 per 1000 being live births[5,6]. Prevalence varies widely, ranging from 1.3-50.89 per 1000 live births across studies[7,8]. Geographic variations and evolving patterns of CHD incidence have also been reported. CHD remains a leading cause of neonatal mortality, accounting for 10% of infant deaths in India, according to Saxena[9]. Despite advanced diagnostic tools and improved care, delays in diagnosis contribute to high morbidity and mortality. CHDs, which include structural and functional defects present at birth, are often multi factorial, involving genetic predisposition and environmental influences[10,11]. While CHDs occur equally in males and females overall, certain conditions, such as coarctating of the aorta, aortic stenosis and tetralogy of Fallot, are more common in males, whereas atrial septal defects (ASD) and patent ductus arteriosus (PDA) are more common in females[12]. CHD presentations range from asymptomatic findings during routine check-ups to severe symptoms such as cyanosis, clubbing and heart failure. Early recognition is critical., failure to identify critical lesions at birth leads to delayed referrals and increased mortality. Enhanced awareness among pediatricians and access to echocardiography in India have improved CHD detection, with cases at AIIMS, Delhi increasing from <4% in 1991-10% in 2004. In resource-limited settings like India, clinical expertise remains vital for CHD diagnosis and management, supported by echocardiographic confirmation. Advances in CHD diagnostics, including prenatal detection and treatment have significantly improved outcomes in developed countries, with over 75% of infants with critical CHD surviving beyond the first year[13]. However, in developing countries, limited access to specialized care contributes to high morbidity and mortality. Government initiatives, such as the Rashtriya Bal Swasthya Karyakram (RBSK), aim to screen all newborns for CHD[14]. Additionally, the Ministry of Health has included CHD in its special task programs to reduce cardiovascular disease burden in India[15-18]. The aim of this study is to evaluate the incidence of congenital heart disease (CHD) among hospitalized patients, analyze its diverse clinical presentations and identify associated risk factors. Additionally, the study seeks to correlate clinical findings with 2D echocardiographic (2D-Echo) assessments and investigate the patterns of morbidity and mortality among CHD patients to provide insights into disease burden and outcomes.

MATERIALS AND METHODS

This prospective study was conducted in the Pediatric Department of a tertiary care institute over two years (1st October 2014 to 31st September 2016). The study included hospitalized patients under 12 years of age with clinically diagnosed congenital heart disease confirmed by 2D echocardiography (2D-Echo). Patients with acquired heart diseases or above 12 years of age were excluded. Eligible patients provided informed consent and their demographic details, including place of residence, birth order, socioeconomic status and educational background, were recorded using a structured questionnaire. Comprehensive general and systemic examinations were conducted, supported by relevant laboratory investigations as needed. Diagnosis in all cases was confirmed through 2D-Echo, supplemented by electrocardiography (ECG) when available and oxygen saturation (SpO) measurements in all four limbs. Medical management was initiated for all participants, with surgical interventions provided by cardiologists when necessary. The study systematically recorded morbidities, clinical outcomes and associated factors. Data was entered into Microsoft Excel and analyzed using statistical tests such as the chi-square test, with results presented in frequency tables and histograms.

RESULT AND DISCUSSIONS

A total of 18,866 patients were admitted in Pediatric department in duration of 2 year between October 2014 to September 2016. Out of the total admitted patients, 290 were diagnosed with congenital heart disease (CHD), yielding an incidence of 1.53% in our study. This finding is comparable to the incidence of 1.65% reported by Sonali and Sushma[19]. Previous studies have reported varying incidences, including 0.8% by Saxena[9], 3.92% by Agarwal[20]. These differences may reflect variations in study populations, diagnostic criteria and healthcare settings. In the present study, acyanotic CHD accounted for 74.1% and cyanotic CHD constituted 25.9%, similar to the findings by Amro[21], who reported 74% acyanotic and 26% cyanotic CHD. Shah GS, Singh[22] reported 69% acyanotic and 31% cyanotic CHD. Among the 215 patients with acyanotic CHD in our study, the highest incidence (38.1%) occurred in the 1 month-1 year age group, while cyanotic CHD was most common in patients under 1 month (40%), reflecting the earlier presentation of cyanotic CHD (Table 1). The incidence of CHD in the 1 month-1 year age group was 35.8%, comparable to 39.8% in Shah GS, Singh[22], 39.48% in Sonali and Sushma[19] and 71.9% in Rukeya and Kumud *et al.* Male predominance was observed (54.5% males vs. 45.5% females), consistent with Amro[21](54.9% males) and Chadha[23] (52.7%). Bidwai[24] reported higher male proportions of 67% and 65.8%,

respectively. In the present study, the most common type of acyanotic congenital heart disease (CHD) was ventricular septal defect (VSD), observed in 39.5% of patients, followed by patent ductus arteriosus (PDA) in 24.6% and atrial septal defect (ASD) in 22.3%. These findings align with the study by Amro *et al.*, which reported a VSD incidence of 43.4%[21]. Similarly, Rashmi *et al.* and Shipra *et al.* documented a VSD incidence of 31.3%, while Sonali and Sushma et al. reported 38.89%[19,25]. The least common CHD in the present study was the combination of ASD and PDA, identified in 2.8% of patients, which is comparable to the findings of Rashmi and Shipra *et al.,* who reported an incidence of 4.6% for the same condition[25]. In this study, breathlessness (63.44%) (Table 2) was the most common symptom, consistent with Sonali and Sushma[19] (74.83%) but higher than S. Agharkhedkar[26] (35.1%). Cough and fever (54.4%) were comparable to both studies. Bluish discoloration (25.86%) was lower than Sonali and Sushma[19](37.41%) but higher than S. Agharkhedkar[26] (11.1%). Feeding difficulty (24.8%) and failure to gain weight (12.06%) were less frequent than in Sonali and Sushma[19] but higher than S. Agharkhedkar[26]. These findings underscore variations in CHD presentations across populations and methodologies. The incidence of congenital heart diseases (CHD) in patients with high maternal age in this study was 3.8%, comparable to Vaidyanathan's findings (3.5%)[27]. Tikkanen and Heionen *et al.* (1990) also identified high maternal age as an established risk factor for CHD[28]. Consanguineous marriages are recognized as a significant contributor to the increased prevalence of recessively transmitted diseases, as reported by Faesar and Biddle[29]. According to the IAP classification, 23.86% of patients had PEM Grade I, followed by Grade III (14.4%). Among PEM Grade IV cases, 17 had acyanotic CHD and 6 had cyanotic CHD (Table 3). Malnutrition was observed in 38.6% of acyanotic CHD and 20.48% of cyanotic CHD patients. Chi-square analysis revealed a significant association (p=0.0001), indicating higher malnutrition rates in cyanotic CHD. Similar findings were reported by Varan *et al.,* highlighting a higher malnutrition incidence in cyanotic CHD[30]. In the 5-12 years age group, under nutrition was classified by BMI, with 24 of 26 patients being undernourished, including 15 with cyanotic CHD (Table 4). Under nutrition, linked to chronic morbidities of CHD, was statistically insignificant between acyanotic and cyanotic CHD (p=0.48). The most common complication was congestive cardiac failure (55.5%), attributed to poor medication compliance, irregular follow-up, respiratory infections and pulmonary hypertension. Pneumonia (35.8%) and anemia were also frequent, with anemia observed in 15.86% of acyanotic and 16.2% of cyanotic CHD cases. Pulmonary hypertension was seen in 17.58% of acyanotic and 9.3% of cyanotic CHD patients. Similar findings were reported by Rao[31] and Jain[32]. Rare complications included digoxin toxicity (arrhythmias, hypokalemia, renal injury), brain abscess in one case of tetralogy of Fallot and brain infarcts in two cases. Severe anemia occurred in 18.6% of acyanotic and 6.6% of cyanotic CHD cases, while polycythemia was prevalent in 40% of cyanotic CHD patients. The most common radiological finding in the present study was cardiomegaly, observed in 45.17% of patients, predominantly in acyanotic congenital heart diseases (CHDs). Oligemia, noted in 15.5% of cases, was commonly associated with obstructive lesions such as pulmonary stenosis, Tricuspid Atresia, TGA with pulmonary stenosis, and DORV with pulmonary stenosis. Plethora was more prevalent in acyanotic CHDs. Specific radiological patterns included a boot-shaped heart in 26 patients with Tetralogy of Fallot (TOF), egg-on-end appearance in 4 patients with TGA and figure-of-eight shaped heart in 3 patients with TAPVC. Dextrocardia with situs inversus was found in 4 cases (Table 5). Electrocardiography (ECG), performed in 110 patients, revealed right ventricular hypertrophy in 31 cases, biventricular dysfunction in 9 and first-degree heart block in 2 cases. TOF was the most common cyanotic CHD (48%), followed by TAPVC, which frequently presented with congestive cardiac failure. Ebstein anomaly and peripheral pulmonary stenosis were the least common CHDs (0.7%). Comparative findings showed the incidence of TOF was 29.68% in Sonali and Sushma *et al.* and 34.6% in Rashmi and Shipra et al. In contrast, TAPVC incidence was 0.4% in Rashmi and Shipra *et al.* and Coarctation of Aorta was 3.4% in Amro[19,25]. Among 290 patients with congenital heart disease (CHD), 65.8% received medical management, while 34.2% underwent surgical intervention. Surgical management was significantly more common in cyanotic CHD (p=0.0001). Medical therapy was provided to 58.3% of acyanotic CHD cases (192 patients) and 7.9% of cyanotic CHD cases (23 patients). Commonly used medications included furosemide and digoxin for decongestion and ibuprofen for mild patent ductus arteriosus (PDA) closure in 77.3% of cases (Table 6). Surgical management was required in 21.4% of acyanotic CHD patients (46 patients), primarily for ventricular septal defects (VSD), while palliative surgery was performed in two atrioventricular canal defect (AVCD) cases using pulmonary artery banding, with patch closure planned subsequently. Among cyanotic CHD cases, 62.2% underwent surgery (46 patients), including corrective procedures such as VSD patch closure and muscle bundle resection in 23 tetralogy of Fallot (TOF) cases. Palliative surgeries in TOF included Blalock-Taussig shunts (11 patients). For tricuspid atresia, bidirectional Glenn shunt and modified Fontan procedures were

Table 1. Age and Gender Wise Distribution of Congenital Heart Disease

Age distribution	**Acyanotic (n=215)**	**Cyanotic(n=75)**
<1 month	35(16.27%)	30(40%)
1 month-1 year	82(38.1%)	22(29.33%)
1-5 years	74(34.4%)	19(25.33%)
5-12 years	22(10.23)	4(5.33%)
Gender wise distribution	**Number**	**%**
Male	158	54.5%
Female	132	45.5%

Table 2. Symptoms, Signs and Risk Factors for CHD in Present in This Study

Parameters			**No of patient (n=290)**	**Present study**
Symptom	Breathlessness		184	63.44%
	Cough		158	54.4%
	Fever		158	54.4%
	Bluish discoloration		75	25.86%
	Feeding difficulty		72	24.8%
	Failure to gain weight		35	12.06%
	Peripheral oedema		23	7.9%
	Asymptomatic		18	9.65%
Signs	Tachycardia		259	89.3
	Tachypnea		172	59.3%
	Cyanosis		75	25.8%
	Hepatomegaly		48	16.5%
	Pedal edema		21	7.2%
	Precordial bulge		10	3.4%
	Parasternal heave		8	2.8%
	Raised JVP		8	2.8%
	Hypertension		3	1.03%
	Grade 1-2 Murmur		21	7.2%
	Grade 3 Murmur		168	57.9%
	Grade 4-6 Murmur		75	25.86%
Risk Factors	High maternal age (>/35 years)		11	3.8%
	Gestational diabetes		5	1.7%
	Torch infection	Rubella	3	2.41%
		Toxoplasmosis	1	
		CMV	2	
	Teratogenic drugs	Phenytoin	2	1.03%
		Sodium valproate	1	
	Family history of CHD	Siblings	2	1.03%
		Parents	1	
	Consanguinity		3	1.03%
	SLE		1	0.3%
	Phenylketonuria		1	0.3%

Table 3. Nutritional Status in Patient of Congenital Heart Disease

Grade of PEM	**Cyanotic(n=199)**	**Acyanotic(n=65)**	**Percentage**
Normal	11	107	118 (44.69%)
PEM grade 1	25	38	63 (23.86%)
PEM grade 2	11	21	32 (12.1%)
PEM grade 3	12	26	38 (14.4%)
PEM grade 4	6	17	23 (8.7%)
Total	65	199	264
		P value =0.0001	

Table 4. Under Nutrition, Hb Level and Complications Comparison in Two Study Groups

Parameters		**Cyanotic**	**Acyanotic**
Undernutrition (n=26)	Normal	-	2
	Mild (15-18.4)	3	4
	Moderate (13-14.9)	4	7
	Severe (<13)	2	4
Haemoglobin	Normal (>11)	12(16%)	57(26.5%)
	Polycythaemia	30(40%)	8(3.72%)
	Mild (10-10.9)	21(28%)	52(24.2%)
	Moderate (7-9.9)	7(9.3%)	58(27%)
	Severe (<7)	5(6.6%)	40(18.6%)
Complications	Congestive cardiac failure	63	97
	Pneumonia	23	81
	Anaemia	47	46
	Pulmonary hypertension	27	51
	Brain infarct	2	-
	Infective endocarditis	3	4
	Eisenmengerization	-	3
	Digoxin toxicity	1	3
	Electrolyte imbalance and AKI	3	1
	Brain abscess	1	-

Table 5. Chest X Ray, ECG, 2D ECHO Finding in Congenital Heart Disease

Parameters		Cyanotic CHD	Acyanotic CHD	Percentage
Chest X-ray findings				
	Cardiomegaly	15	116	45.17%
	Plethora	8	65	25.2%
	Oligemia	45	2	16.2%
	Boot shaped	26	-	8.96%
	Egg on end appearance	4	-	1.4%
	Dextrocardia	4	-	1.4%
	Figure of eight	3	-	2.8%
	Box shaped	2	-	0.6%
ECG Findings	**ECG finding**	**No of patient (n=110)**		**Percentage**
	Right ventricular hypertrophy	31		28.2%
	Left ventricular hypertrophy	9		8.2%
	Biventricular hypertrophy	6		5.4%
	Heart block	2		1.8%
	Normal	59		53.6%
2D ECHO finding	**2D ECHO finding**	**No of patient (n=75)**		**Percentage**
	Tetralogy of Fallot	48		48%
	Pulmonary stenosis	3		8%
	DORV+PS	6		6.6%
	Tricuspid atresia	5		6.6%
	Truncus arteriosus	5		5.3%
	TGA	4		5.3%
	Coarctation of aorta	4		4%
	TAPVC	3		4%
	Atrioventricular canal defect	3		4%
	Peripheral pulmonary stenosis	2		2.6%
	Ebstein anomaly	2		2.6%

Table 6. Treatment, Complications and Outcome Among Two Groups of CHD

Parameters		Acyanotic (n=214)	Cyanotic (n=74)	No of patient	%
Modality of treatment	Medical Therapy	169	23	192	65.8%
	Surgical Therapy	46	52	98	34.2%
Outcome	Discharge	57 (76%)	198 (92.1%)	255 (87.9%)	P value=0.009
	Expiry	12 (16%)	14 (6.5%)	26 (8.9%)	
	DAMA	6 (8%)	3 (1.4%)	9 (3.1%)	
Medical Therapy		**Acyanotic (n=169)**	**Cyanotic (n=23)**	**No of patient**	**%**
Medical Therapy	Furosemide +digoxin	66	10	76	39.58%
	Furosemide + digoxin + enalapril	48	5	53	27.60%
	Furosemide + digoxin + Metoprolol	20	5	25	13.02%
	Ibuprofen	41	-	41	21.35%
	Furosemide + spironolactone	20	3	23	11.98%
Surgical Therapy in Acyanotic group		**Type of CHD**	**No of patient (46)**		**Percentage**
Surgical Therapy in Acyanotic CHD Group	Corrective Surgery	VSD	21		45.6%
		PDA	12		26%
		ASD	8		17.4%
		VSD+PDA	3		6.5%
		VSD+ASD	3		6.5%
		ASD+PDA	2		4.35%
	Palliative Surgery	AVCD	2		4.35%
Surgical Therapy in Cyanotic CHD Group	Corrective Surgery	TOF	23		44.23%
		TGA	3		5.8%
		TAPVC	1		1.9%
		DORV	2		3.85%
		Pulmonary Stenosis	3		5.76%
Palliative Surgery		TOF	11		21.1%
		Tricuspid Atresia	4		7.7%
		DORV	2		3.8%
		Pulmonary stenosis	2		3.8%
		Coarctation of Aorta	1		1.9%

Table 7. Complication Leading to Expiry

Complication	Acyanotic	Cyanotic	Total
Congestive cardiac failure	7	8	15
Pneumonia	1	1	2
Pulmonary hypertension	1	2	3
Eisenmengerisation	2	-	2
Infective endocarditis	1	-	1
Total	12	11	23

performed. Arterial switch (Jatene procedure) was the primary surgery for transposition of the great arteries (TGA), while a Rastelli operation or aorto-pulmonary shunt was utilized in double-outlet right ventricle

(DORV) cases (Table 1). Perimembranous VSD was the most common defect requiring medical management (68.2%), while muscular VSD required surgical management in 9.5% of cases. PDA was the most prevalent CHD in preterm infants, with small PDA treated with oral ibuprofen and moderate/large PDA managed with balloon techniques. Female PDA incidence (59.6%) was comparable to Amro *et al.* (63%) and surgical management was more frequent in full-term PDA cases (p=0.03)[21]. Among 53 atrial septal defect (ASD) cases, 45.3% received medical management and 18.8% required surgery. Overall, 87.9% of CHD patients were discharged after appropriate treatment, with mortality rates of 6.5% in acyanotic CHD and 16% in cyanotic CHD. These outcomes are consistent with Sonali and Sushma *et al.,* who reported a 19.7% mortality rate[19]. Lower mortality in the present study is attributed to early diagnosis and intervention. The p-value for the comparison of mortality and discharge outcomes between cyanotic and acyanotic CHD was 0.0096, indicating statistically significant difference. Congestive cardiac failure was the most common complication leading to mortality in congenital heart disease, occurring in 7 patients with acyanotic CHD and 8 patients with cyanotic CHD (Table 7).

CONCLUSION

Congenital heart disease (CHD) remains a leading cause of mortality in children with malformations, emphasizing the importance of early detection and intervention to reduce morbidity and mortality. In resource-limited settings like India, clinical skills are critical for diagnosis, supplemented by 2D-Echo as the gold standard. Regular cardiac evaluation is essential in cases of repeated chest infections or failure to thrive. Male predominance is noted in CHD, and increasing survival of premature infants and late maternal age contribute to rising incidence. Additionally, the prevalence of home deliveries in India may underestimate the true incidence of CHD.

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