

## Profile of congenital heart disease in a semi urban and rural community: Two years' experience at a teaching hospital

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### Abstract

**Background:** Congenital heart defects (CHDs) are an important cause of mortality and morbidity in children representing a major global health burden. It is thus important to determine their spectrum and identify treatable heart defects.

**Aims:** To study the profile of CHD in a semi urban and rural community between age group 0 to 12 years and study different forms and frequency of occurrence of congenital heart diseases in selected cases using echocardiography as diagnostic modality.

**Methods:** In this retrospective study case records of 154 cases of proven cases of CHD were analyzed in detail. Relative frequency of occurrence of each acyanotic and cyanotic congenital heart disease was noted in selected cases. Study period was between January 2015 to December 2016.

**Results:** Of 154 cases analyzed ventricular septal defect was the most common diagnosis. 42 cases were detected to have ventricular septal defect followed in frequency by tetralogy of fallot which was echocardiography diagnosis in 38 cases. PDA followed by ASD were next common acyanotic congenital heart diseases detected. In cyanotic cases TOF was followed by transposition pathology complex. Males outnumbered females.

**Conclusion:** We noted that the profile of CHD in our population was similar to the published literature. Congenital Heart Defects are common in our setup and early detection of CHD by echocardiography is a safe and accurate method of diagnosis of congenital heart disease in the newborn and adds considerably to the evaluation of critically sick children.

**Keywords:** congenital heart disease, echocardiography, ventricular septal defect, tetralogy of fallot

### 1. Introduction

Congenital cardiovascular disease is defined as abnormality in cardio circulatory structure or function that is present at birth, even if discovered much later. Congenital cardiovascular abnormality usually results from altered embryonic development of a normal structure or failure of such a structure to progress beyond an early stage of embryonic or fetal development. The aberrant pattern of flow created by an anatomic defect may in turn significantly influence the structural and functional development of remainder of circulation<sup>[1]</sup>.

Because infant with such life-threatening heart defects may not initially have symptoms or the clinical sign may be obscure, serious condition may not be recognized on the routine physical examination in majority of cases<sup>[2]</sup>.

Birth is a great event from fetal to the postnatal circulation; the most important changes are from an aquatic amniotic environment and placental gas exchange to breathing and pulmonary ventilation. Air breathing means sudden drop of pulmonary vascular resistance and marked increase in pulmonary blood flow. Fetal structures such as foramen ovale, ductus venosus and ductus arteriosus, which was vital for fetal circulation is no longer needed for survival and begin to close. Neonate with CHD associated with ductus dependent pulmonary or systemic blood flow or with mixing physiology such as TGA is at a great risk of compromise and collapse as they fail to make an adequate transition<sup>[3,4]</sup>.

Inasmuch as incidence is the measure most directly descriptive of the occurrence of disease and hence the causal factors concerned with it, it is not surprising that a number of incidence (or population) studies of congenital heart disease

have been attempted in the past. Given the difficulties attendant upon precise clinical cardiac diagnoses, the relative rarity of congenital cardiac defects, and the natural history of many of these lesions, it is also not surprising that in previous studies various truncations have occurred, i.e., absence of specific lesion diagnosis, data restricted to autopsied patients, or experience limited to a single hospital<sup>[5]</sup>.

CHD can be classified into three main categories in clinical point of view.

#### 1.1 Life-threatening CHD

Structural cardiac malformations in which cardiovascular collapse is likely and compromised if not treated early. They include TGA, COA/IAA, AS, and HLHS/mitral atresia, PA and obstructed TAPVR.

#### 1.2 Clinically significant CHD

Structural cardiac malformations that have effects on heart function but where the collapse is unlikely to be need early intervention. Most common defects in this group are ventricular septal defect (VSD), complete atrioventricular septal defect (AVSD), atrial septal defect (ASD) and tetralogy of Fallot (TOF) with good pulmonary artery anatomy.

#### 1.3 Clinically non-significant CHD

Anatomically defined cardiac malformations but no functional and clinical significance. They include small VSD, atrial septal defect (ASD), mild pulmonary stenosis (PS), only detectable with echocardiography and requiring no treatment.

There are two types of the ductus dependent cardiac lesions.

Ductus dependent systemic circulation (also called, left sided

obstructed lesions) includes HLHS and its variants, severe AS, severe form of COA, IAA and its variants. These require ductal patency to maintain perfusion to the whole or even just the lower sides of the body, or the child develops progressive acidosis as the duct constricts. Consequently, perfusion falls and leg pulses become weak, impalpable and oliguria develop due to renal impairment and become progressively compromised. The other type is the ductus dependent pulmonary circulation (also called, right sided obstructed lesions) which includes critical TOF, PA and its variants, critical PS, TA, with PS/PA (with/without VSD), univentricular heart with PS/PA, and severe form of Ebsteins anomaly. TGA with intact ventricular septum (TGA/IVS) serve as ductus dependent lesion, but large ASD is more important to mixing of the circulation Most of these CHDs present progressive cyanosis without response in proper oxygen supply. Because their fetal physiology is chronically adapted to the hypoxia in the uterine life, newborn infants are able to tolerate some degree of cyanosis than older infants or children. The variety of CHD is immense, because of lots of combinations of defects, which can affect the various cardiac levels, atrium, ventricle, septum, veins or great arteries. Category of cyanotic CHD can be divided into decreased pulmonary flow with right to left shunting lesions (PA, TA with shunting at the atrial or ventricular level); poor mixing lesions (transposition physiology); and right to left shunt with intra cardiac mixing lesions (TAPVR, single ventricular physiology, truncus arteriosus. Some CHD evolves during the fetal life as growth of cardiac structures is flow dependent. Thus, fetuses with mild left sided obstructive lesions may progress into coarctation/HLHS over time; similarly, pulmonary atresia with intact ventricular septum is considered a late phenomenon starting off as severe pulmonary stenosis. PPHN is another serious condition that is associated with other neonatal high risk factors which may be difficult to differentiate from the above mentioned cyanotic heart disease [6, 7].

Echocardiogram is the most valuable method in the diagnosis of CHD. More detailed identification of cardiac anatomy can be possible through two dimensional multiple views (including subcostal long and short axis, apical four chambers, parasternal long and short axis and suprasternal) which delineates the entire detailed anatomy in the various sections. Assessment of systolic ventricular function, measurement of chamber dimensions and wall thickness can be possible by M-mode echocardiography. Pulsed or continuous wave Doppler techniques can be used to assess the pressure gradients across the stenotic or regurgitation flow through the valves. Various Doppler wave forms can assess abnormal cardiac physiology; decreased flow in the descending aorta as seen in the COA; and estimation of pulmonary arterial pressure by measurement of the m tricuspoid regurgitation gradient. Color flow is a great tool in defining the direction of flow when valve regurgitation and shunt exist, the accentuation of flow across defects or narrowed valves, and it also helps detect abnormal turbulent flow such as coronary arteriovenous fistulas and collateral vessels [8].

**2. Aims and Objectives**

To study the profile of CHD in a semi urban and rural community between age group 0 to 12 years.  
To study different forms and frequency of occurrence of

acyanotic and cyanotic congenital heart diseases in selected cases.

**3. Materials and Methods**

**3.1 Source of Data**

The source of data is from case sheets of patients of echocardiographically proven congenital heart disease. The patients were referred for confirmation or exclusion of diagnosis congenital heart disease suspected from history and physical examination.

**3.2 Study Design**

This is retrospective observational study carried out at department of noninvasive cardiology at a teaching hospital and referral center. Population covered is mainly semi urban and rural. Study period was from January 2015 to December 2016.

Case records of the children below 12 years who were diagnosed as having congenital heart disease by echocardiography were analyzed in detail with consideration of age, sex, clinical features (reason for referral) and complete Echocardiography diagnosis.

Echocardiography was done using Esaote imagic agille model 2011. All echocardiography examinations were recorded and interpreted by experts with adequate training and experience.

**3.3 Inclusion Criteria**

- a) Age group newborn to 12 years of age.
- b) Patients with proven diagnosis of congenital heart on echocardiography examination.

**3.4 Exclusion criteria**

Acquired heart diseases (rheumatic fever, myocarditis etc.). During the study period of 2 years, total 154 cases were enrolled in this study, who fulfilled inclusion Criteria. After inclusion in the study, in each case a thorough analysis was done, observations were recorded in a prescribed proforma. As this is a descriptive study after discussing with the statistician no statistical methods are applicable only ratios and percentages were used for evaluation.

**4. Results**

Within the study period of 2 years, 154 children aged 0–12 years who presented to us for various complaints were diagnosed as having CHD through echocardiographic evaluation.

Age distribution of these cases is presented in Table 1. Maximum cases which we studied were in the age group of 0 to 1 year. As shown in Table 2 number of males were 91 and females 63. One interesting observation with regard to this was that all the 3 cases who were diagnosed as Ebsteins anomaly were females. VSD and TOF, most common acyanotic and cyanotic heart diseases were more common in males as compared to females.

**Table 1:** Age distribution of 154 cases of CHD

Age group	No of cases	Percentage
0-1 year	78	51
2-5 years	44	29
6-12 years	32	20
Total	154	100

**Table 2:** Age distribution of 154 cases of CHD

Sex	No of cases	Percentage
Male	91	59
Female	63	41
Total	154	100

Distribution of cases according to diagnosis of congenital heart disease.

As shown in table 3, ventricular septal defect was most common acyanotic heart disease present in 42 of the 154 cases. (28%) This was followed in frequency by Patent ductus arteriosus and atrial septal defect which were detected in 12 and 10 percent resp.

Among cyanotic congenital heart diseases, tetralogy of Fallot (38 and 25%) was the most common diagnosis followed by other complex lesions. Those were transposition pathology (7%), TAPVC (3%), tricuspid atresia (3%) Ebsteins anomaly (2%), HLHS followed by DORV.

**Table 3:** Table showing relative frequency of occurrence of chd in 154 cases of CHD

ECHO Diagnosis	Total (%)	0-1 year	2-5 years	More than 5 years
VSD	42 (28)	22	11	9
ASD	16 (10)	5	8	3
PDA	18 (12)	12	4	2
VSD +PS	5 (3)	4	1	0
PS	2 (1)	0	0	2
Dextrocardia	3 (2)	2	1	0
TOF	38 (25)	21	13	4
TGV	10 (7)	5	4	1
TAPVC	4 (3)	3	1	
Dextrocardia with TA	1 (1)	1	0	0
TA	1 (1)	0	1	0
DORV	2 (1)	2		
HLHS	3 (2)	3	0	0
Ebsteins Anomaly	3 (2)	2	0	1
Tricuspid Atresia	4 (3)	4	0	0
COA	2 (1)	1	1	0

**5. Discussion**

Over time, the reported total CHD birth prevalence increased substantially, from 1 per 1,000 live births in 1930 to 9 per 1,000

live births in recent years. With a worldwide annual birth rate around 150 million births; this corresponds to 1.35 million live births with CHD every year, representing a major public health issue [9].

The increase in reported total CHD birth prevalence over time may be caused by changes in diagnostic methods and screening modalities rather than representing a true increase. In the 1970s, echocardiography was widely introduced into clinical practice, making it possible to also diagnose asymptomatic patients as well as patients with mild lesions. This development probably explains the increased birth prevalence of total CHD in the 1970s, as well as the increase in specific groups, such as patients with VSD, ASD, and PDA. The relative stability of the estimation of birth prevalence of complex CHD subtypes also argues for a merely methodological increase [10, 11].

The profile of CHD varies depending upon the age group studied (Table 4). Simple and potentially correctable heart defects, like ventricular septal defect, patent ductus arteriosus and atrial septal defect, are common at all ages. However autopsy studies are likely to show a higher incidence of serious and complex CHD. The autopsy series of 270 cases by Kinare *et al*, published in 1981 gives a high prevalence of different variants of hypoplastic left heart syndrome like mitral atresia, aortic atresia and coarctation of aorta. Similarly hospital series in neonates and infants show higher incidence (50%) of serious CHD like transposition of great arteries, pulmonary atresia etc. Ventricular septal defect, patent ductus arteriosus and atrial septal defects are relatively more frequent in older children. CHD in neonates is increasingly recognized in India, perhaps due to increasing awareness in pediatricians who are the primary health care provider. This trend may also be related to widely available echocardiography machines and trained personnel, since echo forms the mainstay of diagnosis of CHD in neonates. In the outpatient department of All India Institute of Medical Sciences, New Delhi, the neonates with CHD form about 10% of all CHD cases seen in 2004, an increase from less than 4% in 1991. The commonest CHD in neonates remains ventricular septal defect. This is closely followed by patent ductus arteriosus. Transposition of great arteries is seen in one fifth of neonates having CHD. Pulmonary atresia and its variants are seen in about 13% of cases [12].

**Table 4:** Profile of congenital heart diseases in India

Author/yr	Age group	No with CHD	Profile (% of all CHD)						
			ASD	VSD	PDA	TOF	TGA	HLH	COA
Shrestha 1980	5-16	111	23	30	11	4			
Kinare 1981	<1 year	270		4	9	12	12	10	8
Vashishtha 1993	5-15 years	44	11	41	4	14			
Thakur 1995	5-16 years	70	38	32					
AIIMS 1996	≤ 12 years	5000	13	53	13	32		22	8
AIIMS 1995	<1 month	574	25	35	28	7			7
Present study	<12 years	154	16	42	18	38	10	3	2

Ventricular septal defect was the most common heart defect found in our study, accounting for 28% of all cases of the CHD. This finding correlates with many previous studies on prevalence [13, 14, 15].

Ashraf M *et al.*, observed a prevalence of 4.1/1000.12 Ventricular septal defect (VSD) was the most frequent lesion seen in 69 (31.2%), followed by patent ductus arteriosus (PDA) in 36 (16.3%) children. Tetralogy of Fallot (TOF) was the most

frequent cyanotic heart disease seen in 17 (7.8%) patients [16]. Similar to previous studies, we also found that tetralogy of Fallot and its variant were the most common type among cyanotic congenital heart diseases, with proportion of 25% among all congenital heart disease. The relatively high proportion of TOF cases as compared to acyanotic heart diseases may be explained as we are getting referrals from more critical indoor patients than outdoor patients.

## 6. Conclusion

Although this study did not provide any novel data, this study was relevant in presenting spectrum of different congenital heart diseases in a semiurban and rural children (0–12 years) so that we can assess the burden created by them; at the same time, we must be familiar with the echocardiographic diagnosis and management of all congenital heart disease including some complex cyanotic diseases also. In this study, ventricular septal defect was the most common diagnosis in patients with congenital acyanotic heart disease and tetralogy of Fallot in patients of cyanotic congenital heart disease.

## 7. References

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