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Analysis of Congenital Heart Defects in Neonates: A Tertiary Care Hospital's Perspective from Karachi

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ABSTRACT

Objective: To determine the frequency and distribution of congenital heart defects (CHD) among neonates presenting with clinical suspicion of CHD at a tertiary care hospital.

Methodology: A cross-sectional study was conducted in the Neonatal Intensive Care Unit (NICU) of the National Institute of Child Health (NICH), Karachi, Pakistan, from January 2023 to June 2023. A total of 100 neonates (aged 0–28 days) meeting the inclusion criteria were enrolled. Following a detailed clinical history, physical examination, and systemic assessment, neonates exhibiting signs and symptoms suggestive of CHD underwent echocardiography by a pediatric cardiologist for definitive diagnosis. CHD cases were classified as atrial septal defect (ASD), ventricular septal defect (VSD), tetralogy of Fallot (TOF), patent ductus arteriosus (PDA), pulmonary stenosis (PS), or tricuspid atresia (TA).

Results: Among the 100 neonates with suspected CHD, 52% were male and 48% were female, with a mean age of 9.7 days (SD ±7.3). Congenital heart defects were confirmed in 84 neonates (84%), while 16 (16%) had normal cardiac findings. Female neonates exhibited a slightly higher prevalence of CHD. The most common defect was PDA (58 neonates, 61.7%), followed by VSD (28 neonates, 29.8%), ASD (9 neonates, 9.6%), TOF (8 neonates, 8.5%), PS (6 neonates, 6.4%), and TA (4 neonates, 4.3%).

Conclusion: The study demonstrates a high prevalence of CHD among neonates with clinical suspicion of cardiac anomalies, underscoring the need for early diagnostic evaluation in this population.

Keywords: Atrial septal defect (ASD), congenital heart defects (CHD), echocardiography, patent ductus arteriosus (PDA), tetralogy of Fallot (TOF), ventricular septal defect (VSD).

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Introduction

Congenital heart defect (CHD) refers to an abnormality in cardio-circulatory structure or function at birth, although it may be diagnosed much later. It is the most prevalent form of birth defect that causes more deaths during the first year of life than any other birth defects.[1] The estimated prevalence of CHD in different studies all over the world ranges from 4/1,000 to 50/1,000 live births.[2] Recently, a meta-analysis from 1970-2017 showed that the prevalence of CHD first diagnosed in early childhood was 1/384/1,000.[3]

Prenatal ultrasonography is able to identify CHD prenatally, but it is very crucial for the neonatologist to evaluate the newborn suspected of heart defect at birth. About 2-3 out of 1000 live births will be symptomatic with heart defect in the first year of life, whereas in 40-50% patients with CHD, the diagnosis is made within one week and in 50-60% patients, within one month of age.4

Aetiology of CHD is often associated with maternal perinatal rubella infection, maternal alcoholic use, maternal drug treatment and radiation and various genetic and chromosomal abnormalities.[5] Risk of CHD, if one sibling is affected is also cited in literature with a frequency of 1-3%.[6] Similarly, maternal diabetes mellitus also increase the risk of CHD by 3-4 folds.[7] Preterm infants have more than twice as many cardiovascular malformations as do infants born at term.[8]

Congenital heart defects are often divided into two types: cyanotic (blue discoloration caused by a relative lack of oxygen) and acyanotic defects. The most prevalent cyanotic CHD is TOF, which cause 1/10th of all CHD, while the acyanotic defect is VSD.[3] Study conducted by Ali Farman et al[9] reported acyanotic CHD in 63.07% of the patients with PDA as the most common defect, while

36.92% were having cyanotic CHD with TOF being the most common lesion. Numerous other studies of CHD in children from developing countries reported VSD as the most common defect followed by ASD, PDA, TOF, PS, TA and Ebstein's anomaly.[10]

The incidence of CHD has been studied in developed countries for many years but in developing countries like Pakistan, the true incidence and prevalence of CHD is unknown due to a large number of home deliveries, limited access to medical care and inadequate resources to undertake intense population studies. Local data of children with CHD of all ages is available; however, neonatal CHD data has not been previously evaluated. Early diagnosis of CHD in neonatal period will lead to better treatment and reduction in the mortality and morbidity. Therefore, this study was conducted with the aim to determine the frequency and pattern of CHD in the new-borns with clinical suspicion of CHD.

Methodology

This is prospective cross sectional hospital-based study was conducted from January 2023 to June 2023 at the NICU of National Institute of Child Health (NICH), Karachi, Pakistan which is a teaching tertiary care hospital. The Sample size was calculated on the basis of least proportion of outcome variables from literature i.e. 13.4 % which is pulmonary stenosis, according to the study done in Children hospital, PIMS Islamabad.[11] The estimated sample size was 100 neonates. We used WHO sample size calculator for sample size calculation, which was used for sample size calculation for a single proportion.

Data was collected after the approval by the Institutional Ethical Review Board with approval certificate No IERB-02/2023, Dated: 03-01-2023. Informed verbal consent was taken from the parents/caregivers of neonates before the enrolment.

All the neonates with suspected CHD admitted at NICU, not previously diagnosed as CHD and having age less than 28 days of life were enrolled in the study. Children older than 28 days at the time of admission, those already diagnosed as a case of CHD, critically ill neonates who expired before echocardiography and those who refused to give consent were excluded from study.

Non-probability sampling technique was used. Each enrolled neonate allotted a unique number and his/her hospital number was noted. Cardiac evaluation was included detailed family history, physical and systemic examination of cardiovascular system and echocardiogram. Neonates with clinical suspicion of CHD base on presence of murmur, cyanosis with or without feeding, signs of congestive heart failure, feeding difficulties, pre and post ductal SPO2 gradient difference of more than 3%, those born to diabetic mothers were referred for echocardiography for confirmation of diagnosis. To minimise bias, a trained paediatric echo cardiologist performed the Doppler studies on all the neonates. Doppler studies were performed using Toshiba Xario-XG echocardiography equipment (Toshiba, Japan) with three-dimensional (3D), continuous wave colour Doppler, M-mode and 3-6 MHz transducers for neonates. During the echocardiogram the neonate was sedated with chloral hydrate 50 mg/kg when required. Echocardiographic pictures were recorded in standard parasternal long-axis, short-axis, apical four chamber, subcostal and suprasternal views. Afterwards each neonate was managed according to standard treatment for the specific type of CHD. All the data was noted on a structured proforma especially designed for this study.

All the data gathered were verified daily by study researcher. All the forms were verified for completeness and consistency and coding before data entry. Data analysis was done using SPSS version 23.0. Descriptive analysis of study qualitative variables was shown in frequency and percentage whereas quantitative variables were shown as mean and standard deviation. Chi square test was used to identify difference with p-value of ≤ 0.05 being considered significant.

Results

A total of 100 neonates with signs and symptoms suggestive of CHD of age < 28 days at the time of presentation were included in this study. There were 52 males (52%) and 48 females (48%), male to female ratio was 1.1: 1. Mean age of neonates was 9.7 days with SD of 7.3, Min – Max = 1 – 27 days. Majority of the neonates 63 (63%) had age between 1 – 10 days while 37 (37%) neonates had age > 10 days. Out of 100 neonates, fifty (52%) neonates were delivered preterm while 45 (45%) were delivered at term and 3 (3%) were post term. Risk factors in obstetric history of mothers was also noted. Majority of mothers 43 (43%) were diabetic (gestational diabetes and type 2 diabetes), 42 (42%) were hypertensive, family history of congenital heart defect was seen in 11 (11%) mothers and 8 (8%) mothers were smoker, illustrated in table I. A three-dimensional color Doppler echocardiographic assessment was performed in

all 100 children. Congenital heart defects were found in 84 (84%) neonates and 16 (16%) neonates were found normal. Stratification was done to control effect modifiers for example age, gender, family history of CHD, gestational age, maternal diabetes and smoking. Gender distribution of CHD positive cases showed female preponderance, as there were 41 of the 48 neonates (85.4%) females and 43 out of 52 (82.7%) males, but difference is not statistically significant (*p*-value =0.9).

Table-I: CHD in neonates with maternal risk factors in obstetrical history.

History	Total	CHD YES	CHD NO	Percentages	*p*-value
Diabetes	43	34	9	79.1%	0.66
Family History of CHD	11	10	1	90.9%	
Smoking	8	6	2	75.0%	

Table II: Types of Congenital Heart Defects in neonates.

Types	Number of neonates (n = 84)	%
Patent Ductus Arteriosus	41	48.8%
Ventricular Septal Defect	19	22.6%
Atrial Septal Defect	8	9.5%
Tetralogy of Fallot	7	8.3%
Pulmonary Stenosis	5	5.9%
Tricuspid Atresia	3	3.6%
Transposition of Great Arteries	1	1.1%

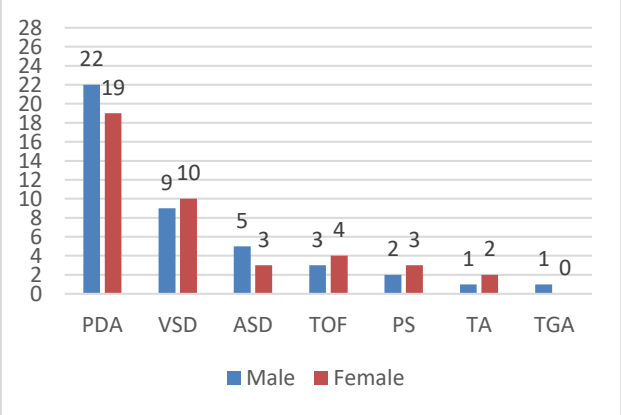

Fig. 1: Gender Distribution of CHD.

The mean age at diagnosis of CHD was 10.1 (±7.5) days. Majority of the neonates 32 (86.5%) had age greater than 10 days, but statistically insignificant (*p*-value = 0.8). High percentage of CHD was seen in preterm neonates at birth 47 (90.4%) (*p*-value = 0.04) and neonates with family history of CHD 10 (90.9%) (*p*-value = 0.66).

The most common type of CHD was PDA, found in 41 (48.8%) neonates followed with decreasing frequency by VSD in 19 neonates (22.6%), ASD in 8 neonates (9.5%), TOF in 7 neonates (8.3%), PS in 5 (5.9%), TA in 3 neonates (3.6%) and TGA in only 1 neonate (1.1%). Frequencies of all types were high in females except PDA and ASD but the difference between genders was not statistically significant as all *p*-values > 0.5. Frequencies of all types were high in neonates of age > 10 days.

Discussion

A 3D colour-Doppler echocardiographic assessment was performed in all the 100 enrolled neonates. After echocardiography, congenital heart defects were found in 84% Neonates. Gender distribution of CHD positive cases showed female predominance, as there were 41 of the 48 neonates (85.4%) females and 43 out of 52 (82.7%) males which is consistent to the study conducted by Ali Farman et al with male: female of 1:1.18.[9] But other studies show male predominance.[12,13]

The most common type of CHD in our study was PDA, found in 48.8% neonates, which is similar with the findings of study done by Hassan AA et al[14] and Khasawneh W et al[15] but different from another local study done in 2016 by Pate N et al[16] in which TOF was found to be the most common type. And in another studies[17,18] VSD was reported to be the commonest defect. The high frequency of PDA in the present study may be explained by the fact that echocardiographic assessment in this study (not used in most of the previous studies) was sensitive enough to diagnose very small PDAs that may not be suspected on clinical ground. The second commonest lesion in our study was VSD in 22.6% neonates, which is consistent with the findings of Simonneau G, et al.[19] Overall, acyanotic CHD had high frequency (80.9%) compared to cyanotic CHD (19.1%), the results are similar to the studies done by researchers in other regions.[9,20]

In our study, frequency of CHD was higher in preterm neonates (90.4%) compared to the term neonates (80%) which is consistent with the studies of Palma A[8], Reddy RK[21] and GE JC.[22] Furthermore, researchers reported that positive family history of CHD have high recurrence in the first degree relatives[23]. Our study also demonstrated that 90.9% of the neonates have CHD having positive family history which is very high. Similarly, studies show that maternal risk factors in obstetrical history like diabetes, hypertension and smoking are also positively associated with increased risks of CHD[24] but in our study this association is insignificant (*p*-value 0.66).

In this study, predominant CHD in males was PDA and ASD. Females were predominant in VSD, TOF, PS and

TA. This result is inconsistent with other studies. In Bangladeshi study males were predominant in VSD, TOF, A-V canal defect and single ventricle with single A-V canal defect. Females were predominant in ASD, PDA, COA, TGA and multiple lesions but equal distribution in PS.[25]

There is variation in the data from different regions and these discrepancies in frequencies may result from difference in detection methodologies, available resources, genetic diversity and environmental influences.

Congenital heart defects are very common in our setups. The absence of a comprehensive national database about CHD in Pakistan is a cause for concern regarding this significant congenital anomaly. It should prompt policymakers to convene and establish national guidelines aimed at monitoring this substantial health burden effectively.

Conclusion

This research indicates a high incidence of congenital heart defects among the neonates with clinical suspicion of CHD in a large tertiary care center of Karachi, Pakistan and it also indicates the most frequent lesions. We suggest that, all the new born babies should be investigated properly for any sign of CHD and follow up investigation should be recommended along with early echocardiography, because early diagnosis and treatment may reduce morbidity and mortality. We also suggest future multi-centers research to assess the incidence and determine the patterns and distribution of CHD more precisely at the national level. Further, our findings may prove useful in policy making for the better management of CHDs.

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