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ORIGINAL ARTICLE

Spectrum of cyanotic congenital heart disease in full term neonates attending paediatric cardiology clinic of a Tertiary Cardiac Care Centre.

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ABSTRACT… Objective: To determine the spectrum of cyanotic congenital heart disease (CCHD) in full term neonates attending paediatric cardiology clinic of a tertiary cardiac care centre. **Study Design:** Descriptive Cross-sectional study. **Setting:** Department of Paediatric Cardiology, Faisalabad Institute of Cardiology (FIC), Faisalabad. **Period:** January 2021 to June 2021. **Material & Methods:** All full term newborn of any gender with age range from day one to twenty eight days, presenting in emergency or outpatient department of the institute and diagnosed as a case of CCHD at Faisalabad Institute of Cardiology on the basis of echocardiography were included in the study.The leading or main anatomical diagnosis of hemodynamically significance was adopted. The spectrum of CCHD was assessed by descriptive analysis. **Results:** A total of 87 patients were enrolled. Mean age was 15.7±7.98 days. There were 55.2% (n=48) male and 44.8% female (n=39) patients. Majority of patients (62.1%) presented as an emergency. Most CCHDs were of decreased pulmonary blood flow (69%, n=60). Tetralogy of Fallot was the most common CCHD seen in 25.3% neonates (n=22) followed by Transposition of great arteries (23%, n=20) and pulmonary atresia (18.4%, n=16). Ebstein anomaly of tricuspid valve was the least common CCHD (2.3%). **Conclusion:** Tetralogy of Fallot is the most common cyanotic congenital heart disease followed by TGA while Ebstein anomaly is the least common.

Key words: Cyanotic Congenital Heart Disease, Neonate, Tetralogy of Fallot.

INTRODUCTION

Congenital heart disease (CHD) is a structural abnormality of the heart or intra-thoracic great vessels that has actual or potential functional significance.[1] CHD is the most common congenital malformation at birth with the estimated incidence of 8 per 1000 live births.[2] Widely CHDs are classified as cyanotic and acyanotic based on presence or absence of cyanosis.[3] Cyanotic heart disease accounts for 25% of cardiac defects.[4] Many of these are life threatening, hence early diagnosis and treatment is the key for such patients. Pulse oximetry is performed in neonate with in first day of life to rule out cyanotic heart disease (SPO2<90%), but it does not screen all of them.[5] Modern echocardiography is an extensive modality for assessing all forms of CHD.[6] Cyanotic heart disease can be classified on the basis of increased or decreased pulmonary blood flow.[7]

Some of cyanotic CHDs have decreased pulmonary flow with right to left shunting lesions like pulmonary atresia (PA), tricuspid atresia (TA) where shunting is at the atrial or ventricular level; Others are poor mixing lesions like transposition of great arteries; and those shunting right to left with intra cardiac mixing lesions like truncus arteriosus, total anomalous pulmonary venous connections (TAPVC) and single ventricular physiology.

Early detection of cyanotic congenital heart defect in neonatal age leads to better management and a significant decrease in morbidity and mortality. Timely diagnosis of congenital heart disease in neonates was once a dilemma in Pakistan.[8]

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The data on spectrum of neonatal cyanotic CHD is scanty in our country. By knowing the spectrum of cyanotic CHD in neonatal life, awareness can be created in public and health care professionals regarding this subject of significant concern so that relevant interventions could be planned early to confront this disorder of profound importance. The objective of this study was to know the spectrum of cyanotic congenital heart disease in full term neonates at a tertiary cardiac care facility.

MATERIAL & METHODS

This descriptive cross sectional study was conducted at Pediatric cardiology department, FIC from January 2021 to June 2021. Approval from institutional ethical review committee (16/ DME/FIC/FSD) was taken for this study and there was no conflict of interest. All full term newborn with age range from day one to twenty eight days, diagnosed as a case of cyanotic CHD at FIC, and presenting in emergency or out patient department (OPD) of the institute were included. This also included those emergency cases which were shifted from different paediatric units of the Faisalabad division for balloon atrial Septostomy or any balloon intervention. Only those neonates who had a detailed echocardiography report done by a dedicated pediatric cardiologist of FIC were enrolled. The neonates who had cyanosis without cardiac reason, those having persistent pulmonary hypertension of newborn, premature babies or those having incomplete echocardiography report or incomplete data were excluded from the study.

The base line data like name, father name, residential area, and contact details including residential address and hospital registration number for record purpose was noted. Clinical data including age (in days), weight in kilograms (Kg), gender (male/ female/ transgender), mode of presentation (Emergency or OPD) and indications of referral like dyspnea, cyanosis, palpitation or murmur were noted. Similarly cutaneous saturation (SPO2) in percent (%) by pulse oximetry and Echocardiographic diagnosis were recorded in a comfortable environment. The echocardiography based cyanotic CHD diagnosis was reviewed in the study subjects and where more than one structural lesions seen, the leading or main diagnosis of hemodynamically significance was adopted. The spectrum of the cyanotic CHD in neonates was assessed by their anatomical leading diagnosis and also by categorizing them as having increased or decreased pulmonary blood flow. The Informed consent was taken from parents or guardian of study participants after assurance of confidentiality of the personal data by the principal investigator. Those parents who did not give consent were also excluded from the study.

All the data was entered in the Proforma designed by the principle investigator, shifted to Excel sheet and analyzed. The descriptive analysis for the outcome variable that is spectrum of cyanotic congenital heart disease was done by sorting out frequencies and percentages of qualitative variables like cyanotic congenital heart disease. The quantitative variables like age and weight was presented as mean and standard deviation.

RESULTS

A total of eighty seven neonates diagnosed as having cyanotic CHD were enrolled according to inclusion criteria during the study period. Minimum age was 3 days while mean age was 15.7 ±7.98 days. Minimum weight was 2 kg and maximum weight was 4 kg while mean weight was 2.6±0.39 kg. The mean cutaneous oxygen saturation was 80%. As regard gender, male to female ratio was 1.2:1. Table-I shows baseline characteristics of the patients.

Baseline Characteristics	No (%)
Gender	
Male	48 (55.2)
Female	39 (44.8)
Age Distribution	
1 to 9 Days	34 (39.1)
10 to 18 Days	25 (28.7)
19 to 28 Days	28 (32.2)
Mode of presentation	
Emergency visit	54 (62.1)
OPD visit	33 (37.9)
Distribution of Cyanotic CHD	
Decreased pulmonary blood flow	60 (69)
Increased pulmonary blood flow	27 (31)

Table-I. Baseline characteristics of patients

The indications or symptoms for referral were sorted out which revealed that cyanosis was the most common indication for referral (47.1 %, n=41) followed by a murmur (29.9%, n=26). Table-II shows the indications for referral among the study subjects.

Indication	Among CCHD (%)
Cyanosis	41 (47.1)
Murmur	26 (29.9)
Tachypnea	12 (13.8)
Asymptomatic/ Screening	8 (9.2)

Table-II. Indication for cardiology referral

As regard the disease spectrum, we divided our study population in two categories: one category of general spectrum of CCHD describing anatomical diagnosis and the other one on the basis of decreased or increased pulmonary blood flow. The most common CCHD was tetralogy of Fallot (TOF) found to be present in 22 patients (25.3%) followed by transposition of great arteries (23%, n=20). The spectrum of CCHD on the basis of major anatomical diagnosis is described in Table-III.

As regard spectrum of CCHD on basis of increased or decreased pulmonary blood flow it was noted that TOF was the most common CCHD with decreased pulmonary blood flow (25.3%, n=22) followed by pulmonary atresia seen in 18.3% of neonates with CCHD (n=16). Similarly TGA was the most common cyanotic congenital heart disease with increased pulmonary blood flow (16.1%, n=14). The spectrum of cyanotic CHD with reference to increased and decreased pulmonary blood flow is given in Table-IV.

DISCUSSION

The age of presentation is very relevant to the spectrum of CCHD as many of the neonates are undiagnosed and critically ill at birth in our part of the world and die within hours of birth or within first few days of life at home or in neonatal units due to scanty diagnostic facilities. In our study mean age of the study subjects were 15.7±7.98 days. A study done in neonatal intensive care unit of upper Egypt showed mean age of presentation of neonates having CCHD was 11.78±9.4 days.[9] There is clear difference of age at presentation as compared to our study. This contradiction is due to lack of health care facilities in our part of the world.

Cyanotic CHD is more common in male gender

Anatomical Diagnosis	Gender		Frequency	Percentage of Total
	Male n=48	Female n=39		
Tetralogy of Fallot (TOF)	12	10	22	25.3
Transposition of Great Arteries (TGA)	12	8	20	23.0
Pulmonary atresia (PA)	10	6	16	18.4
Tricuspid Atresia	3	5	8	9.2
Corrected Transposition of Great Arteries (CcTGA)	3	3	6	6.9
Double outlet right ventricle (DORV)	2	2	4	4.6
Univentricular heart (HLHS, DILV, Single Ventricle)	3	1	4	4.6
Truncus arteriosus	2	1	3	3.4
Total anomalous pulmonary venous connection (TAPVC)	0	2	2	2.3
Ebstein Anomaly (EA)	1	1	2	2.3

Table-III. Spectrum of CCHD on basis of anatomical type of cardiac defect with gender distribution

Increased PBF	No. (%)	Decreased PBF	No. (%)
TGA with VSD PH/ Intact IVS	14 (16.1)	TGA with VSD and PS	6 (6.9)
UV Heart with no PS	2 (2.3)	UV heart with PS	2 (2.3)
Truncus arteriosus	3(3.4)	Pulmonary atresia	16 (18.3)
TAPVC	2 (2.3)	TOF	22 (25.3)
Tricuspid atresia with no PS	2(2.3)	Tricuspid atresia with PS	6(6.9)
DORV	4 (4.6)	CcTGA with VSD & PS	6 (6.9)
		Ebstein anomaly	2 (2.3)

Table-IV. Cyanotic congenital heart diseases on basis of pulmonary blood flow

as seen in different studies.[10,9] The results of our study are not different as male to female ratio turned out to be 1.2:1 in our study.

The spectrum of CCHD on the basis of major anatomical diagnosis varies in order of frequency across the world from one centre to other. In some centers, TOF remains the most common cyanotic CHD while in other centers TGA remains the most frequent CCHD. In our study Tetralogy of Fallot was the most common cyanotic structural heart defect with frequency of 25.3%. This observation is consistent with different studies conducted in Jordan[11,12], India[13,14,4], Pakistan[15,16], and globally.[17] TGA was the most common CCHD in neonatal period (27.9%) in a study conducted in Srinagar.[18] In another study conducted in Chennai[19], TGA was the most common CCHD noted in 33.9% of cyanotic congenital heart disease neonates. Similarly a study by Lynch A et al[20] showed that TGA was the most common cyanotic CHD in prenatal as well as post natal period. TGA was the most common cyanotic CHD in neonates in similar studies carried out in Karachi[21] and Abbottabad[3] Pakistan. In our study TGA was the second most common CCHD noted in 23 % of cases. However systematic review and meta-analysis of 260 studies shows that TOF is the most common neonatal cyanotic CHD.[17]

In a study carried out in Tamil Nadu, pulmonary atresia was the second most common cyanotic CHD after TGA.[19] In another study done in Aga Khan University Hospital[16], TOF with its variants was the most common CHD as pulmonary atresia was not considered as a separate entity. Pulmonary atresia is the third commonest cyanotic congenital defect with a frequency of 18.4% in our study.

The spectrum of neonatal CCHD, as per above discussion, varies between TOF, TGA and the pulmonary atresia in the first second or third slot. We consider that spectrum of neonatal CCHD will continue to vary as pre natal diagnostic facilities; Neonatal services and CHD screening diagnostic facilities are scanty in our country. The spectrum will depend, on which day of life baby presented and what was the outcome on the basis of CHD. Most of neonatal deliveries are at home in developing country like Pakistan, so the babies born with critical CHD especially hypoplastic left heart syndrome, univentricular heart or TGA are less likely to survive until diagnosed and properly managed.

There is no population or hospital based data available from Pakistan regarding incidence, clinical course and mortality from cyanotic CHD. During last three decades, early recognition and treatment of such neonates have become possible all around the world.[20] Thus these babies can reach adulthood in a state of nearly normal health. But due to lack of cardiac facilities in our country we are unable to save these little, ailing hearts.

Our study has a few limitations as well. This study is of six months duration and may not represent the true spectrum of different neonatal cyanotic heart diseases, as our institute is not a direct neonatal referral centre. The neonates presenting via OPD or referred for echocardiography from other hospitals of the city Faisalabad or periphery were included. We consider that those neonates who were critically ill and had suspected cyanotic CHD but could not be referred to us for CHD screening and died in the respective neonatal unit or those who remained undiagnosed at home and died could have changed the spectrum and outcome. However the effort to conduct this study was made to highlight this health issue of significant importance in our setup.

CONCLUSION

Tetralogy of Fallot is the most common neonatal cyanotic congenital heart disease (25.3%) followed by transposition of great arteries (23%). In a resource limited country like Pakistan, the spectrum of cyanotic CHD can be quite different if prenatal screening of CHD is done by fetal echocardiography in each pregnancy, every newborn is attended at birth by neonatologist, cutaneous saturation noted by doing pulse oximetry in every neonate and timely referral for CHD screening is adopted.

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AUTHORSHIP AND CONTRIBUTION DECLARATION

No.	Author(s) Full Name	Contribution to the paper	Author(s) Signature
1	Motia Javed	Principal investigator designed study, Collected Data, Analyzed results, Wrote introduction.	
2	Abdul Razzaq Mughal\	Design study, Abstract and methodology writing.	
3	Imtiaz Ahmad	Discussion writing, Wrote the limitation and conclusion of the study.	
4	Zaigham Rasool Khalid	References and Critical analysis of the study.	
5	Usman Zia	Data collection.