International Journal of Contemporary Pediatrics
Pradhan JB et al. Int J Contemp Pediatr. 2020 Feb;7(2):311-315
http://www.ijpediatrics.com
pISSN 2349-3283 | eISSN 2349-3291
DOI: http://dx.doi.org/10.18203/2349-3291.ijcp20200100

Original Research Article

Clinical profile and outcome of congenital cyanotic heart disease in neonatal period: a retrospective study

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Received: 24 July 2019
Revised: 06 December 2019
Accepted: 12 December 2019

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ABSTRACT

Background: Congenital Cyanotic Heart Disease (CCHD) is under reported during neonatal period and mortality rate is high in India. Aims and objectives of the study determine clinical presentation, maternal and neonatal risk factors and outcome of CCHD during neonatal period.
Methods: A retrospective study conducted over 15-month period during March 2017 to June 2018 in a tertiary out born NICU. Neonates with echocardiographically confirmed case of structurally abnormal heart disease were analyzed.
Results: Among 106 CCHD neonates, 60% neonates were asymptomatic, 22% had persistent cyanosis and 19% had tachypneic at birth. Cyanosis (35%) and tachypnea (30%) were common initial presentation in postnatal period. 22% neonates were readmitted with initial clinical symptom after discharge from birthing centre. CCHD were more common in male and term neonate, 34% were small for age, median age at admission and discharge were 5days (2-12) and 5 days (2.7-9.2) days respectively. Nineteen percent neonates had fetal distress and 6% neonates required aggressive resuscitation at birth. First trimester abortion (16%), maternal diabetes (10%) and hypothyroidism (7%) were common maternal risk factor. Nearly 20% neonates were diagnosed at referring hospital and 4.7% were transported with PGE1 during transport. TGA (17%) was most common lesion noted followed by pulmonary atresia (10%). Fourteen percent neonates died during the neonatal period. Disease specific death rate was highest for Single ventricle (42%) followed by TGA with IVS (37.5%) and aortic arch anomaly (28.5%) in this study.
Conclusions: Early diagnosis and management may improve the survival in CCHD neonates.

Keywords: Congenital cyanotic heart disease, Congenital heart disease, Cyanosis, Echocardiography, PGE1, Pulse oximetry, Shock

INTRODUCTION

Congenital Heart Disease (CHD) is the most common congenital anomaly and is responsible for 30-50% of infant mortality in western country and second only to prematurity.[1-3] Congenital Cyanotic Heart Disease (CCHD) is often categorized as critical and major type who requires intervention or results in death within four weeks and one year of life respectively.[4-6] Most of the CCHD are undiagnosed during routine antenatal ultrasound scan. Duct dependent CCHD usually have varied presentation and often presents after discharged from the birthing centre. Factors contributing to a late diagnosis include absence of a murmur, mild hypoxemia, inadequate training of staff and early postnatal discharge from birthing centre. Late diagnosis especially in critical CCHD is associated with significant increase in mortality and morbidity.[7-9] Aim of screening for CCHD is to avoid

late diagnosis and morbidity. Referral for cardiac evaluation has been guided by universal fetal ultrasound screening in some countries and postnatal pulse oxymetry screening in many others.[2,6,9-12]

In India nearly 30 million of live birth occurs in each year and 90 percent of birth occurs in the primary health centre where facilities are lacking. The prevalence of CHD is not uniform in this country as various studies have reported it ranging from 8 to 14 per 1000 live births.[13,14] According to a status report on CHD in India, 10% of the present infant mortality may be accounted to CHD. 15 Considering the huge burden of childbirth and proportionate prevalence of CHD, there are significant challenges for better outcome including prenatal ultrasound screening and referral to definitive cardiac and neonatal care centre. Even in major cities, only 1%-2% of pregnancies currently avail fetal echocardiography.[16] Nearly 10% of neonates with critical cardiac lesions undergoes timely detection and operative treatment. There is no pulse oxymetry screening programme in India at present.

Aims and objectives of this retrospective study were to determine etiological risk factors associated with CCHD, types of lesions, common modes of presentations, prenatal and post-natal risk factors influencing the survival and mortality of neonates admitted in the tertiary referral out born neonatal unit.

METHODS

This retrospective study was conducted in department of neonatology in the out born referral unit at institute of child health and hospital for children's, Egmore, Madras Medical College, Chennai; India. Centre as largest tertiary referral newborn care centre in southern part of India, admit and treat all type of cardiac lesion in the neonatal unit. The data were collected from the database hospital record during the period from March 2017 to June 2018 after approval from the institutional ethical committee. Neonates with the echocardiographically confirmed case of structural heart disease identified from the department database were included in this study. Cyanosis other than structural heart lesion, Persistent Pulmonary Hypertension of Newborn (PPHN) and acyanotic congenital heart disease was excluded from the study.

Maternal demographic details including age, parity, birth order, abortion, risk factors such as diabetes, hypothyroidism, drug intake, smoking and alcohol consumption, oligohydramnios, polyhydramnios, antenatal scan and details of any anomaly if detected were recorded. Intrapartum details, resuscitation details, birth weight, gestational age were recorded. Postnatal details as mode of presentation (shock, respiratory distress, cyanosis, murmur etc.), onset of symptom and initial treatment in diagnosed and undiagnosed case, feeding details, investigations, details of transportation and age on referral were noted.

The oxygen saturation, mode and duration of transportation, age on referral and adequacy of support during transport, cardio respiratory support at arrival, mode of presentation and treatment of diagnosed and undiagnosed neonates, severity based on oxygenation saturation, acidosis, cardio respiratory failure, etiological analysis, treatment, complications and short term morbidity were analyzed.

Diagnosis of CCHD is made on a combination of clinical, pulse oxymetry and echocardiography grounds. Severity of CCHD is categorized based on maximum oxygenation Saturation, need of cardiorespiratory support, acidosis, other comorbidity like seizure, renal failure, necrotizing enterocolitis, coagulopathy, treatment response to prostaglandin therapy, type of lesion, operable or inoperable after stabilization.

Treatment of CCHD

In authors unit, duct dependent pulmonary and systemic circulatory type CCHD managed with immediate PGE1 infusion after initial stabilisation and screening echocardiography. Management with oxygen to maintain Spo2 within a target range of 65-75% (oxygen hood, HFNC, CPAP, mechanical ventilation), circulatory support to maintain mean arterial BP and adequate cardiac contractility (dopamine, dobutamine, adrenaline, nor adrenaline, milrinone) when needed, maintenance of temperature, euglycemia, electrolytes within normal range and haematocrit of more than 40%. The nature of respiratory support, ventilation duration, maximum mean airway pressure and FiO_2, need for ionotropes and vasodilator were documented.

Outcome measures

- Maternal risk factor and clinical presentation of CCHD.
- Etiologic and mortality distribution based on severity and type of lesion.

Analysis and reporting

The statistical analysis was calculated with STATA 12 software. Mean, Median and Mode, Standard deviation and inter quartile range were calculated for continuous variables. Proportion and percentage were calculated for categorical data. Chi square test and Fischer's exact test were used to test the association between categorical variables and student t test for continuous variables. A p-value <0.05 was consider statistically significant.

RESULTS

During this study period, 143 cases of cyanotic heart disease were admitted and treated in the unit. Thirty-

seven cases were excluded from the study due to incomplete data or death before echocardiography confirmed diagnosis. Total 106 cases of CCHD were included and analyzed. Maternal demographical data showed mean maternal age of 25.4±4.12years, 45% were Primipara and 30% were having consanguineous marriage (Table 1). Vaginal delivery was most common mode of delivery (54%). Most common risk factors associated with CCHD were maternal diabetes (9.9%) and hypothyroidism (6.6%). First trimester abortion was noted in 16% of mothers. Only 5% of pregnancy was undergone Assisted Reproductive Therapy (ART). Sixteen cases (15%) were antenatally detected as CCHD by antenatal USG method. Fetal distress was present in 20% of case during intrapartum period. Only 19% neonates were delivered in tertiary care centre. Persistently low APGAR score at 5 min were noted in 6.7% neonates and 5.6% neonates required aggressive resuscitation. Majority (88%) were term gestations and 64% were male with mean birth weight of 2676.33±510.26 gm, 26.5% were IUGR and 7.5% were SGA (Table 2). In this study, the mean gestational ages were 38.23±1.44weeks. Median ages on admission of the neonates were 5 days (2-12 days). Fifteen neonates (14%) were associated with multiple congenital anomalies (Table 2). 56.6% of neonates were asymptomatic at birth. Cyanosis and tachypnea were present in 21.6% and 19% respectively at birth. In postnatal period, Cyanosis, tachypnea and murmur were common initial mode of presentation with 35%, 30% and 7.5% respectively. Only 2% neonate present with shock. 21% of neonates with major heart defects were discharged without being diagnosed at their birth hospitalization and readmitted with life threatening symptoms.

Table 1: Maternal demographic details.

	Characteristics	N=106 (%)
	Maternal age in years (Mean±SD)	25.4±4.12
Mode of delivery	Vaginal delivery Elective LSCS Emergency LSCS	58(54%) 28(27%) 20(19%)
	Primipara	48(45%)
	Consanguinity	32(30%)
	1st trimester abortion	17(16%)
	Maternal risk factor Diabetes mellitus Hypothyroidism	17(16%) 10(9.5%) 7(6.6%)
Conception	Spontaneous Art*	101(95%) 5(5%)
	Antenatal scan detecting CCHD**	16(15%)
	Fetal distress	20(19%)
Place of delivery	PHC CHC Tertiary care	22(21%) 56(53%) 19(18%)

[*ART (assisted reproductive therapy) **CCHD (Congenital Cyanotic Heart Disease)]

Table 2: neonatal demographic details.

Characteristics	N=106 (%)
Apgar score 1 min score ≤7	55(52%)
5 min score ≤7	7(6.7%)
Resuscitation required bag and mask	6(5.6%)
Gestation Term	93(88%)
Preterm	13(12%)
Birth weight*(gm)	2676.330±510.26
Birth category AGA	70(66%)
IUGR	28(26.5%)
SGA	8(7.5%)
Male gender	68(64%)
Anomaly	15(14%)
Admission weight (g)*	2664±483.79
Gestational age (weeks)*	38.23±1.44
Age on admission (d)**	5(2-12)
Average hospital stay (d)**	5(2.7-9.2)
Admission after discharge	23(21.6%)

[*mean, ** median (IQR)]

Nearly 20% neonates were diagnosed at referring hospital and 4.7% were transported with PGE1 during transport. Sixteen percent of neonates were treated with ionotropes and 12% on ventilator support at referral hospital (Table 3).

Table 3: Clinical characteristics of neonate in postnatal period.

	Characteristics	N=106(%)
Symptoms in delivery room	Normal	60(56.6%)
	Tachypnea	20(19%)
	Cyanosis	23(21.6%)
	Asphyxia	3(2.8%)
Mode of presentation at referring hospital	Tachypnea	32(30%)
	Cyanosis	37(35%)
	Murmur	8(7.5%)
	Shock	2(2%)
	Tachypnea and cyanosis	14(13%)
	Cyanosis and murmur	9(8.5%)
Diagnosis at referring hospital	Confirmed	21(20%)
	Suspected	70(66%)
	Others	15(14%)
	Pge1 during transportation	5(4.7%)
Treatment at referral hospital	Ventilator support	13(12%)
	Ionotropic support	17(16%)
Mode of transport	108 ambulance	84(79%)
	PVT ambulance	21(20%)
	Self	1(1%)

In this study, TGA was most common type of CCHD (16.9%) followed by Pulmonary stenosis with VSD (14%), Anomalous pulmonary connection (12%), DORV (11%) and pulmonary atresia (10%) (Table 4). Fifteen

(14%) neonates died during this study period. Single ventricle (33%) was most common cause of death followed by TGA with IVS and anomalous pulmonary connection (20% each). Disease specific death rate was highest for Single Ventricle (42%) followed by TGA with IVS (37.5%) and aortic arch anomaly (28.5%) (Table 4).

Table 4: Diagnostic characteristics and mortality in neonates.

Type of lesion	N=106(%)	Death (n=15) (%)	Disease specific death rate (n) (%)
TGA	18(16.9)	3(20)	3(37.7)
TGA+IVS	8(7.5)	3(20)	3(37.5)
TGA+VSD	10(9.4)	0	0
TOF	7(6.6)	0	0
PA	11(10.3)	0	0
PS+VSD	15(14)	0	0
COA/IAA	7(6.6)	2(13.3)	2(28.5)
TA	3(2.8)	0	0
Truncus	6(5.7)	1(7)	1(16.7)
PAPVC/TAPVC	13(12.3)	3(20)	3(23)
DORV	8(7.5)	1	1(12.5)
SV	12(11.3)	5(33.3)	5(42)
Ebstein	2(2)	0	0
Heterotaxy	3(2.8)	0	0
Complex HD	1(1)	0	0

(TGA-Transposition of great artery, TOF-tetralogy of fallot, PS-pulmonary stenosis, TA-tricuspid atresia, TAPVC/PAPVC-Total or partial anomalous pulmonary venous connection, SV-single ventricle, IAS-intact atrial septum, COA/IAA-Coarctation of aorta/interrupted aortic arch).

DISCUSSION

In this study, 106 neonates were confirmed case of CCHD and treated in NICU for initial stabilization and monitoring. Nearly 30% were born out of consanguineous parents indicating the high prevalence of consanguinity in this region. Fetal distress requiring emergency LSCS were noted in 19% of pregnancy and 6% baby required aggressive resuscitation. This may be due to ante partum hypoxemia precipitated by onset of uterine contraction. Maternal diabetes was more commonly associated risk factor noted with CCHD in this study as similar finding noted in Wren et al.[17] Antenatal detection rate for CCHD was 15% whereas, it was reported to be 35% in UK and 39% in US.[8] Thirty-four percentage of neonates were small for age (SGA). Male are common in this study similar to study by priyadarshini et al.[14] Twenty-one percents of neonates with critical heart defects were discharged without being diagnosed after their birth hospitalization similar to the finding by Wren et al.[18] Nearly 40% babies were symptomatic soon after birth. The commonest mode of presentation on postnatal life were cyanosis and tachypnea. The lack of pulse oxymetry screening programme might led to more late symptomatic presentation. In this study 14% neonates with CCHD had other congenital anomalies whereas in Stoll c et al, showed 26.3% had associated major anomalies. Twenty percent neonates have confirmed diagnosis at referring hospital but only 4.7% received prostaglandin infusion before transportation to the hospital.

Most common cardiac abnormalities in this study was d-TGA (18%) followed by PS with VSD (14%) whereas study by Landis B et al, showed single ventricle as common abnormalities.[19] Thirty percent of neonates were treated with ionotropes and ventilator support. Mortality was noted higher in these neonates and finding were similar to study by Akbar molaei et al.

In this study, the mortality rates were 14% which were similar to study by Brown et al. and Anagnostou et al, as 12% and 10% respectively.[8] 20 Single ventricle (33%) was most common cause of death followed by TGA with IVS and anomalous pulmonary connection (20% each) in contrast to study by Akbar molaei et al, where d-TGA was most common cause of death. Disease specific death rate was highest for single ventricle (42%) followed by TGA with IVS (37.5%) and aortic arch anomaly (28.5%). In this study only six neonates underwent surgery during neonatal period.

Few limitations of this study were as out born referral unit the perinatal events, diagnosis and treatment details at referral hospital are retrospectively collected from the physician and parents. Strength of the study is that prompt echocardiographic diagnosis after admission by

trained neonatal fellow and treatment with PGE1 in duct dependent lesion led to good outcome with medical therapy in this study.

CONCLUSION

Antenatal screening for cardiac lesion particularly in the high-risk mother with consanguinity, diabetes and hypothyroidism can increase the detection rate. This will help in planned management in immediate postnatal period. Universal pulse oxymetry screening may help in early detection, decrease the readmission and may help in early treatment and survival of neonates. Mortality is lesion specific. Early diagnosis and prompt medical therapy and referral to tertiary facility will improve the survival in CCHD neonates.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Pradhan JB, Kamalarathnam CN. Clinical profile and outcome of congenital cyanotic heart disease in neonatal period: a retrospective study. Int J Contemp Pediatr 2020;7:311-5.