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Clinical Profile and Outcome of Cyanotic Congenital Heart Disease in Neonates

Khadija N. Humayun and Mehnaz Atiq

ABSTRACT

Objective: To determine the clinical profile and assess the outcome of all neonates diagnosed with cyanotic congenital heart disease.

Study Design: A case series.

Place and Duration of Study: The Aga Khan University Hospital from January 1998 to December 2000.

Patients and Methods: Neonates admitted with diagnosis of cyanotic congenital heart disease were evaluated for clinical diagnosis, survival and mortality.

Results: Forty four neonates met the inclusion criteria. Eleven babies (25%) had Tetralogy of Fallot or its variants. Other malformations were d-transposition of great arteries, tricuspid valve anomalies (tricuspid atresia and Ebstein's anomaly), hypoplastic left heart syndrome, truncus arteriosus, total anomalous pulmonary venous return and complex congenital heart disease like single ventricle. Twenty eight (63.6%) neonates survived and 16 (36.4%) expired during hospital stay. Cause of death was surgical in 2 cases and medical problems in 14 babies.

Conclusion: Tetralogy of Fallot or variants was the commonest cyanotic heart disease in neonates with frequency of 27.27%. Majority of neonates with congenital cyanotic heart disease showed survival with appropriate management.

Key words: Neonates. Cyanosis. Congenital heart disease.

INTRODUCTION

Congenital heart diseases are major structural malformations of heart and/or major vessels, present at, or persisting abnormally after birth.¹ In Pakistan, the true incidence and prevalence of congenital heart disease is unknown due to limited access to medical care and limited resources to undertake intense population studies. Incidence of congenital heart disease in all populations, where adequate data is available, is between 8-10 per 1000 live birth.^{2,3}

Although cyanotic congenital heart disease accounts for less than 25% of cardiac defects,⁴ many of these are life threatening in neonatal period, thereby necessitating early diagnosis and prompt treatment. Availability of diagnostic tools like echocardiography, fetal scanning, advancement in surgical techniques and postoperative care have radically improved the life expectancy of affected children. Thus, complete repair of many congenital heart diseases in neonatal period or early infancy is now possible and patients with complex congenital heart disease are now surviving beyond the pediatric age.

This helps to determine the relative frequency, predisposing risk factors and gravity of various cyanotic

cardiac lesions presenting in neonatal period. The findings will hopefully have implications in development of cardiac services in other tertiary care centres in Pakistan.

The aim of this study was to evaluate the clinical features and assess the outcome of all neonates with cyanotic congenital heart disease at the Aga Khan University Hospital, Karachi.

PATIENTS AND METHODS

Case records of 44 neonates, admitted to the Aga Khan University Hospital from January 1998 to December 2000, with diagnosis of cyanotic congenital heart disease were reviewed retrospectively. All the neonates who were either born at the Aga Khan University Hospital or were referred in the neonatal age group were included. Babies having cyanosis due to causes other than congenital heart disease were excluded from the study. Studied variables were the type of anomaly, demographics, presentations and survival.

Cardiac evaluation included detailed history, physical examination, ECG, chest X-ray and echocardiography. Congenital heart disease was classified as Tetralogy of Fallot (TOF) or its variant pulmonary atresia, with Ventricular Septal Defect (VSD) and overriding aorta d-Transposition of Great Arteries (d-TGA), tricuspid valve abnormalities (Ebstein's anomaly and tricuspid atresia), truncus arteriosus, Total Anomalous Pulmonary Venous Return (TAPVR) and complex Congenital Heart Disease (CHD) combination lesions with uni-ventricular heart and Pulmonary Stenosis (PS).

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The data was analyzed using SPSS version 10 for windows and frequencies and means were calculated by descriptive analysis.

RESULTS

During the 3 years study period, 44 neonates were diagnosed to have cyanotic congenital heart disease giving an average of 14.66 cases per year. Out of the 44 babies, 15 (22.7%) were born at AKU and 29 (77.3%) were admitted through emergency room. Male preponderance was seen with a ratio of 3.4:1. Age at admission of the study population was 1-30 days. Mean age at admission was 5 days. Majority of patients n=19 (43.2%) were admitted on the first day of their life. They were all symptomatic within few hours of their birth. Out of the 19 admissions, 15 were born at the Aga Khan University Hospital.

Only 3 (7%) babies were born pre-term and 41 (93%) were delivered at term. Thirty (69%) patients were born appropriate for gestational age and 14 (31%) babies were low birth weight. Out of those, 2 were pre-term and 12 were term deliveries but small for gestational age. Mean weight of the study subjects was 2.7 kg with a range between 1.9 to 3.5 kg. Four (9%) neonates had a sibling with congenital heart disease. None of the parents had congenital heart disease.

Commonest presenting features were cyanosis and respiratory distress followed by poor feeding and lethargy. On initial examination, most of the patients had tachypnea, tachycardia and cyanosis.

Commonest lesion was tetralogy of fallot found in 11 (25%) cases. The group with d-TGA was second largest with 10 (23%) babies. Three patients in this group had a large VSD and 2 had VSD and PS. Five patients had complex lesions with complete atrioventricular septal defect, double outlet and inlet ventricles.

Five patients were diagnosed to have tricuspid atresia. Ebstein's anomaly was also seen in 2 babies. The other conditions were seen less frequently. Table I gives the cardiac diagnoses of all babies with the outcome. Fourteen (31.8%) babies were found to have associated

Table I: Cyanotic congenital heart lesions seen in study population and their outcome.

Name of lesion	Frequency (%)	Outcome	
		Survived	Expired
Tetralogy of Fallot	12 (27.3)	8	4
d-transposition of great arteries	10 (22.7)	7	3
Tricuspid valve anomalies			
Tricuspid atresia	5 (12.3)	5	-
Ebstein anomaly	2 (4.5)	2	-
Hypoplastic left heart syndrome	4 (9.1)	1	3
Truncus arteriosus	1 (2.3)	-	1
Complex cyanotic CHD	9 (20.45)	4	5
TAPVR	1 (2.3)	1	-
Total	44 (100)	28	16

extra-cardiac congenital anomalies in addition to their cyanotic CHD (Table II).

Table II: Extra-cardiac congenital anomalies in study subjects.

Diagnosis of CHD	Extra-cardiac congenital anomalies
TOF	VATER syndrome Imperforate anus, cryptorchidism Polydactyly Tracheoesophageal fistula
d-TGA	Situs-inversus, spina bifida, absent left kidney
HPLHS	Tracheo-esophageal fistula Situs-Inversus
T. Atresia	Prune-Belly syndrome Tracheoesophageal fistula, rt. hydronephrosis
Complex CHD	Absent gallbladder Edward's syndrome Cleft palate Carpenter's syndrome Imperforate anus

Thirty four (77.3%) neonates required ventilation. Balloon atrial septostomy was performed in 7 cases. All these had a diagnosis of d-TGA with intact ventricular septum. Eight patients underwent palliative surgery for modified Blalock-Taussig Shunt. One baby had surgery for tracheoesophageal fistula. Twenty eight of the 44 (63.6%) neonates survived; 16 (36.4%) babies expired. Table III gives the details of effect of various factors on the outcome of the patients.

Table III: Factors affecting the outcome of study subjects.

Associated factors	Survivals	Mortalities	Total
Sepsis	7	4	11
Extra-cardiac lesions	7	7	14
Sex			
Male	21	13	34
Female	7	3	19
Place of Birth			
AKUH born	9	6	15
Outside referrals	9	20	29

DISCUSSION

The incidence of congenital heart disease of 8-10 per 1000 live birth.^{2,3} Coupled with the extremely high birth rate of Pakistan, it can be easily inferred that nearly 50,000 children are possibly born with congenital heart disease each year.⁶ Consanguinity is also very high in the local population and its association with congenital heart disease has been well described.^{7,8}

Considering these figures the admission rate of 15 babies per year with such lesions seems like only the tip of the iceberg. These results show that around 2/3 of the study population was referred from other hospitals for better management specifically due to the presence of neonatal intensive care facilities and availability of a pediatric cardiologist at the study centre.

Male dominance in this population could be an incidental finding because the same was also noted in

babies born in the same hospital. Overall M:F ratio is reported to be 1:1 for congenital heart disease as a whole but it varies with different cardiac lesions.⁷ Male predominance has also been shown in other studies done in Pakistan.⁹

Availability of the tertiary care facilities including pediatric cardiology service contributed to the early diagnosis of congenital heart disease in majority of subjects as 15 out of 19 day one admissions were born at the same hospital. They were immediately shifted to NICU and confirmation of diagnosis was within 24 hours of birth. It is interesting to note that all the cases of TOF in this study presented in the first week of their life. This finding is supported by data from other centres.¹⁰ TOF was the commonest lesion in keeping with other studies.^{6,9,15-17}

Although the number of preterm babies in this study is very low in contrast to the recent report on incidence of congenital heart disease among hospital live births from India¹¹ which shows that incidence of these lesions were eleven times more common in preterm babies compared to term. Poor outcome of preterm babies with cyanotic congenital heart disease is also documented in other studies.¹²

Four (10%) neonates had a history of sibling having a congenital heart defect. Risk of recurrence for most of the congenital heart diseases, if one sibling is affected is cited as 1-3%.¹³ None of the parents in this study had congenital heart disease. There were 3 babies whose mothers had diabetes mellitus. Maternal diabetes mellitus is cited to be associated with congenital heart disease in about 4-5% cases.¹⁴

The review of these cases although doesn't provide the exact frequencies of occurrence of these lesions in the population but definitely gives an idea of the referral pattern to the tertiary care centers, which in turn has an implication in the development of cardiac care facilities for neonates and children.

One third of the study subjects had associated extra-cardiac congenital anomalies. This is in accordance with the reported incidence of these malformations in literature.¹⁸⁻²⁰

Eleven (25%) babies in the study developed pneumonia or sepsis during the hospital stay, out of whom 4 expired. All of them also had extra-cardiac congenital malformations.

During the hospital stay, 34 (77%) patients required ventilatory support. Catheter intervention using Rashkind's balloon technique was done in 7 babies for atrial septostomy who had d-TGA and intact ventricular septum or tiny VSDs. Six of these were successful with immediate improvement in the oxygenation and decrease in cyanosis. One baby succumbed during procedure and he was confirmed to have sepsis as co-morbid.

Nine neonates were operated upon during the hospital stay. One baby had repair of tracheoesophageal fistula, the other 8 had insertion of modified Blalock Taussig shunt. Out of these 8, there were 4 cases of TOF, one case of tricuspid atresia and pulmonary atresia and other 3 were pulmonary atresia with complex heart lesions, atrioventricular septal defect and double outlet right ventricle. Two patients expired after surgery, one of them had massive haemorrhage and DIC and in the second case, the shunt was blocked.

Twenty eight (63.6%) babies survived the neonatal period. Of the 16 expires, 2 died due to surgical complications and remaining 14 succumbed to medical problems. Seven of the 16 expiries had extra-cardiac congenital anomalies in addition to the cyanotic congenital heart lesion. A sub-group of 4 babies out of these 7 also had sepsis and pneumonia with the cardiac and extra-cardiac malformations. Six of the expiries were AKU born and 10 were referred from outside.

The mortality rate of 36.4% is more than that reported from other national hospital-based studies^{6,9} but these studies were done on children of all age groups i.e. from neonatal to pre-pubertal and included heart diseases other than congenital heart disease also.

There is no population or hospital-based data available from Pakistan for the neonatal death rate due to congenital heart disease. A recent study from Hong Kong²¹ reports 20% mortality in cyanotic heart disease patients, but again their study population comprised of children upto 4 years of age. A study from Lebanon²² also confirms the high mortality of complex cardiac lesions in early days of life in absence of adequate surgical facilities.

In the last 25 years, early recognition and judicious treatment of neonates with cyanotic congenital heart disease has become possible, thus these babies can reach adulthood in a state of nearly normal health. This study was done with an objective to evaluate the clinical profile and assess the outcome of such babies in a tertiary care setting of a developing country. It was found that even in this era of early reparative surgery we in Pakistan are able to offer mostly palliative care and quite a few of them lose their lives to coexisting morbidities. This data also outlines the pattern of presentation of these babies, thus, enabling to gear toward better management facilities.

It is not known as to how many home delivered babies succumb to their cyanotic congenital cardiac lesions without ever being diagnosed and referred to these facilities. It is, therefore, essential that signs of heart disease are recognized as early as possible and the infant referred to the cardiac facility. The outcome would greatly depend on the cooperation between the primary care physician, pediatric cardiologist and cardiac surgeon and it is the team effort which gives pink hues to blue babies.

CONCLUSION

Tetralogy of Fallot or variants was the commonest cyanotic heart disease in neonates with frequency of 27.27%. Majority of neonates with congenital cyanotic heart disease showed survival with appropriate management.

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