Review Article

CRITICAL CONGENITAL HEART DISEASE SADLY DIAGNOSED AT AUTOPSY IN CALABAR; SOUTH-SOUTH NIGERIA

ABSTRACT

Congenital heart disease (CHD) is the most common type of birth defect causing more deaths in the first year of life than any other birth defect. Medical practice in a third-world country is faced with numerous challenges, created by poor health facilities and the unavailability of basic imaging studies at the community level where the majority of the poor resides. We present a case series of 4 neonates with a M: F ratio of 1: 3 admitted into the neonatology unit of the University of Calabar Teaching Hospital, Calabar, Nigeria. Two of the patients were delivered at home by a traditional birth attendant via spontaneous vertex delivery and the rest two were booked cases in a private and obstetric unit of the University of Calabar Teaching Hospital. Their age ranged were 18 hours to 27 days of life and their common presentation were poor sucking, difficulty in breathing, central cyanosis and failure to thrive. A fetal echocardiogram was carried out for one of the cases and there was strong clinical suspicion of congenital heart disease but the precise type is unknown. The other three could not afford the necessary investigations required. They were all oxygen-dependent till death. Pathologic-anatomic findings showed a rare Critical congenital heart defect of the univentricular heart chamber of various types for the three cases and a case of TGA. The Immediate cause of death for all four cases was congestive cardiac failure.

KEYWORDS: Critical Congenital heart diseases, Autopsy findings, TGA, Univentricular heart.

INTRODUCTION

Critical Congenital heart disease (CCHD) is the spectrum of congenital heart disease that requires urgent medical and most appropriate surgical intervention for the survival of the infant. Such intervention is necessary within the first year of life (1). Ige et al in a study conducted in Jos showed that congenital heart disease is prevalent in Nigerian neonates and there is therefore a need for advocacy to improve access to its diagnosis at birth for appropriate management (2). The risk of morbidity and mortality increases significantly if there is a delay in early recognition and management, as well as referring infants with critical CHD to the tertiary care centre for definitive surgical or catheter-based intervention (3). Babies with such cardiac defects may appear relatively healthy for the first few hours or days of life leading to their early discharge home and possible misdiagnosis of the defect. Pulse oximetry screening, a non-invasive procedure is highly recommended in the diagnosis of CCHD and it should be performed in newborns after 24 hours of age or before discharge from the hospital for early detection of the defect and a positive screening test result can be followed immediately with an echocardiogram to further evaluate and determination of the type of CCHD (4,5). The greatest challenge in our locality is the lack of medical practitioners with surgical skills to save neonates and if such specialists are available in a private facility, the cost of surgical corrections of defects is not

affordable. The exact aetiology is unknown but genetic and environmental factors are strongly implicated with socioeconomic, or ethnic origin also playing a role (6). Neonates with such defects can present in several ways which include an abnormal heart sound during a heartbeat (heart murmur), rapid breathing (tachypnea) and a blue or purple tint to the skin caused by a shortage of oxygen (cyanosis) or shock within the first week of life when the ductus arteriosus closes (3). Such clinical presentation may be immediate or delayed depending on the severity of the cardiac anomalies. Majority of the neonate with good cry and activity, delivered to a healthy mother without complications get discharged home only to represent with fast or rapid breathing or failure to thrive. The condition of the neonate is made worst considering their place of delivery and the skill of the birth attendant.

We present a case series of 4 neonates with a M:F ratio of 1: 3 admitted into the neonatology unit of the University of Calabar Teaching Hospital, Calabar, Nigeria. Two of the patients were delivered at home by a traditional birth attendant via spontaneous vertex delivery and the rest two were booked cases in a private and obstetric unit of the University of Calabar teaching hospital. They presented with fast breathing, and cyanosis and all were oxygen dependent until death. Pathologic-anatomic findings showed a rare Critical congenital heart defects of the univentricular heart chamber of various type for the three cases and a case of TGA. The Immediate cause of death for all four cases was congestive cardiac failure.

CASE SERIES:

CASE 1

The patient is a preterm neonate delivered by a 26-year-old para 2+1 mother. The baby was delivered by Emergency caesarean section at 36 weeks gestation in the obstetrics and gynaecological unit of the University of Calabar Teaching Hospital on account of preeclampsia and prolonged rupture of membrane of 19 hours. The baby was nursed in the incubator for a month and managed as a case of Prematurity and Moderate birth Asphyxia and later discharged home at a birth weight of 1.8kg. The baby was re-admitted at the age of 27 days on account of failure to thrive.

Post mortem findings

Description: The body is that of a fresh Negroid female neonate, weighing approximately 2.2kg and the Length is 58cm. There are healed septic spots on the frontal and occipital parts of the scalp. The anterior fontanelle is flat. The eyeball is sunken and the cheekbone prominent. The lips, nail and toe beds are bluish in colour and marked palor. Internal examination of the visceral shows generalised visceral organ congestion and the lungs reveal a moderate amount of pulmonary oedema evidenced by frothy fluid freely oozing out of the lung parenchyma. The dissection of the heart was carried out using vasculature and showed ventricular wall

hypertrophy with D-transposition of the great vessels (Figures 1-2). The cause of death was a congestive cardiac failure (biventricular failure) resulting from critical congenital heart defects of the D-Transposition of the great arteries.

CASE 2

The patient is a term male neonate delivered by a 30-year-old para2+1 mother. The baby was delivered at a gestational age of 37weeks by spontaneous vertex delivery at home by a traditional birth attendant and was said to have cried immediately. The mother did not book antenatal care and was not on routine medication throughout the pregnancy period. The baby was admitted on the third day of life on account of poor crying and sucking and died after 7 days at the age of 10 days.

Post mortem findings

The body is that of a fresh female neonate, weighing approximately 3.2kg and the Length is 56cm. The anterior fontanelle is flat and there is no facial dysmorphism. The attached umbilical stump is clean and has no skeletal deformity. There is severe bluish discolouration of the buccal mucosa, nail and toe beds. There was moderate cephalhematoma. Internal examination shows glistening serous cavities and generalised visceral organ congestion. The dissection of the cardiovascular system shows a univentricular chamber with massive muscular hypertrophy and a single cavity (Figures 4-7). The right and left atrial chambers are noted and connected by a large ASD. The Patent ductus arteriosus is occluded and there is pulmonary vein atresia. The right and left lungs also reveal massive pulmonary oedema with venous congestion. The cause of death was (congestive cardiac failure) and biventricular cardiac failure resulting from critical congenital heart disease not compatible with life.

CASE 3

A two-day-old female neonate was delivered at a gestational age of 37 weeks by elective caesarean sections to a 39 years old para 3+1 (2 alive) lady. Apgar scores were 8 and 9 respectively in the first and fifth minutes of life. The baby was extracted with good activity and a good cry and mild resuscitation and cleaning. Mother booked at our facility at a gestational age

of 13 weeks and was regular with her antenatal visits and routine medicals which includes folic acid and Vitamin B complex. Baby developed respiratory distress and associated cyanosis, two days post-delivery and was oxygen dependent until death.

POSTMORTEM FINDINGS:

The body of a female neonate weighing 2.9kg and measuring 58cm in length. There is marked central and peripheral cyanosis, no pallor, no jaundice. There is no facial dismorphism, skeletal deformity and the attached umbilical cord stump is clean. Internal examination shows a smooth and glistening serious cavities. Dissection of the cardiovascular shows a globular heart weighing 48 grams with a univentricular heart chamber and a large VSD (Figures 3). The PDA is closed and there is no vascular anomalies. There is generalised visceral organ palor and marked pulmonary and cerebral oedema. The Cause of death was (Congestive cardiac failure) biventricular cardiac failure resulting from critical congenital heart disease (univentricular heart chamber with large VSD).

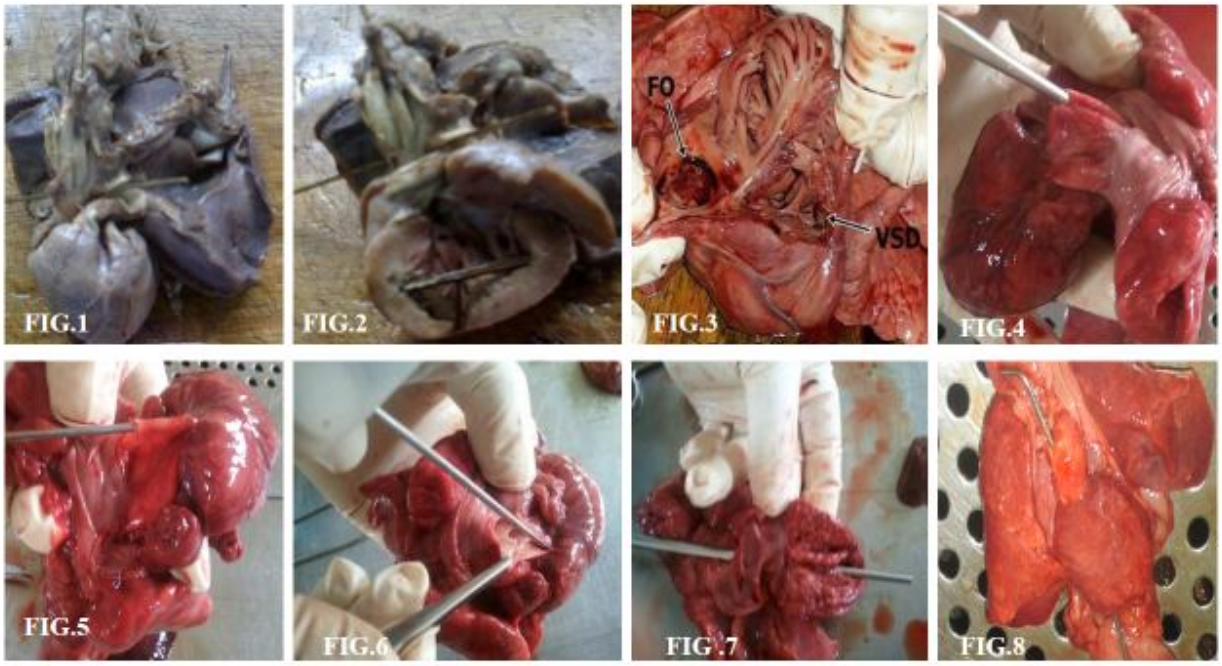
CASE 4

A two-day old postdate male neonate delivered at a gestational age of 42 weeks by Spontaneous Vertex Delivery of a Para 5(4 alive) 30-year-old petty trader. Mother rupture membrane 24hours before delivery and she was running a low-grade continuous fever with difficulty in breathing two and half hour before presentation at the hospital. Baby was dyspnoiec, markedly cyanosed, febrile (38.3^0C) with weak suck. He was assessed as congenital heart disease, meconium aspiration, and congenital pneumonia? He was treated with oxygen, antibiotics. His condition deteriorated and died 18hrs after presentation.

Postmortem Findings:

The body of a male neonate of negroid origin. The length is 52cm, weight is 3. 2kg.There is marked central and peripheral cyanosis, no pallor, no jaundice. There is no facial dismorphism, skeletal deformity and the attached umblical cord stump is clean. Internal examination shows a smooth and glistening serious cavities. Dissection of the cardiovascular shows a globular heart weighing 50grams with a persistent truncus arteriosus and a large inter atrial septa defect of the foramen ovale type (Figures 3 and 8). There is a single ventricular chamber with a wall thickness of 0.3cm. The right and left lungs are heavy with wet surfaces and shows pulmonary oedema with congestion. The liver and spleen also show congested surfaces. The brain weighs 500g, has a fluidy appearance with flattened gyrus and obliterated sulcus. The cut surface show oedema. No evidence of intracranial haemorrhage. The Cause of death was (Congestive cardiac failure)

biventricular cardiac failure resulting from critical congenital heart disease (Persistent truncus arteriosus with a single atrial and ventricular chamber) that is not compatibles with life.



FIGURES (1-8): Shows congenital heart defect of various type. Fig.1-2 shows transposition of the great arteries. Figure 3 shows a large ASD and VSD. Figures 4-7 shows a single ventricular, chamber, hypoplastic left atrial and truncus arteriosus. Figure 8 shows a single atrial and ventricular chamber and persistent truncus arteriosus.

DISCUSSION

Neonatal Autopsy rate in our locality is low (7) and consent for such a procedure is difficulty to obtain from relatives or parents due to religious and cultural beliefs. The identification of the type of congenital heart defects is a great challenge by managing clinicians with over reliance on imaging studies such as echocardiogram. As a results of the paucity of autopsy studies, vital data are often lost and precise cardiac defects under documented. Tuchtan et al on their study showed that post-mortem ultrasound has high sensitivity and specificity in the detection of congenital morphological abnormalities as compared to conventional autopsy but with limited accuracy for congenital heart diseases(8). Pavlicek et al, in their studies also showed a complete agreement between FECHO and autopsy to be 85% (122/143) of cases and affirmed that FECHO is a highly sensitive method for the prenatal detection of CHD but is still incapable of detecting the complete spectrum of cardiac defects (9). Despite sophisticated imaging studies with their narrow margins of identifying the specific cardiac anomalies, autopsy remains the goal standard for the confirmation of the specific critical congenital heart defects. Detailed cardiac dissection in our study showed complicated critical cardiac defect in all the four cases and a single case presenting with both congenital heart defects and non-cardiac malformations involving the musculoskeletal system and the craniofacial deformity. Extra

cardiac anomalies occurrence has been reported in 15-45% of cases with CHD. Gucher et al in a retrospective study of 305 neonatal autopsies reported that 45.9% of cases with congenital heart disease have an associated, one or more extracardiac malformations (10). Majority of the studies utilizing imaging studies such as FECHO has identified various patterns of cardiac defects with Ventricular septal defects being the commonest encountered cardiac defects.

Chinawa et al in their study using 2 D-echocardiography show that 0.22% per cent of children who attended UNTH in Enugu State had congenital cardiac abnormalities and the commonest forms seen were those with VSD followed by tetralogy of Fallot (11). Animasahun et al also in a prospective study at Lagos using endocardiography reported that tetralogy of Fallot (TOF) was the commonest cyanotic congenital heart disease followed by double outlet right ventricle (DORV) and transposition of the great arteries (TGA)(12).An analysis of echocardiographically diagnosed CHD from three different centers across Nigeria also affirmed ventricular septal defect (VSD; 46.6%) to be the commonest CHD in our region followed by patent ductus arteriosus (12.1%), atrial septal defect (8.7%), atrioventricular septal defect (8.2%), and tetralogy of Fallot (7.8%). (13). Ekure et al in their studies involving analysis of data from Children with echocardiography-confirmed CHDs at 17 medical centers across the country also showed that Ventricular septal defects were significantly more prevalent in the North (37.4%) when compared with the South (18.5%; $P < .0001$) and that severe CHDs were more prevalent in the South (14). The findings of severe congenital heart disease in our case reports goes to affirmed the present of such lesions in our locality that are easily misdiagnosed due to inability of patients to afford high resolution image studies and failure to confirm such lesion after death due to the difficulty in obtaining consent for autopsy. The four reported cases in our studies showed various patterns of cardiac defects referred collectively as Critical congenital heart disease that always requires urgent medical intervention followed with corrective surgery to improved survival of the neonate and quality of life. The anatomic-pathologic findings in our case presentation were that of Transposition of the great arteries (figure 1-2). Univentricular chamber heart (Figure 3-8). The case with a postmortem diagnosis of TGA, was the longest survival. She died at the age of 27 days and the major presentation included repeated hospital visitation and failure to thrive. There was no surgical correction of the defects despite an echocardiogram confirmation of the present of CHD but the specific type not identified while still alive. In transposition of the great arteries (D-TGA), the aorta and PA are transposed, or switched, meaning that the aorta arises from the right ventricle and the PA arises from the left ventricle. There was no evidence of a VSD or an ASD following detailed cardiac dissection. This were the exact finding in of case as shown in figure 1-2. The ventricular wall was markedly hypertrophied and the findings in some organs such as the lung showed massive pulmonary edema with heart failure cells and the immediate cause of death was congestive cardiac failure (Biventricular failure). The initial management of TGA focuses on the stabilization of cardiac and pulmonary function. Patients with suspected or confirmed TGA are started on an IV infusion of prostaglandin E1 (alprostadil) to maintain a patent DA and may undergo Balloon Atrial Septostomy (BAS). The infant would have survived if the appropriate surgical correction was provided. Such care is not available in our facility as of today.

The other three cases presented were that of a Univentricular heart (Figure 3-8). These is considered to be an uncommon form of critical congenital heart disease. The concept of univentricular heart as documented as moved from hearts with only one ventricle connected with atria (double inlet ventricle or absent atrioventricular (AV) connection) to hearts not amenable to biventricular repair such as hearts with two ventricles unable to sustain separately pulmonary and systemic circulations in sequence (15). Hearts with one hypoplastic ventricle, pulmonary/aortic atresia or severe stenosis with hypoplastic ventricle are also include in the classification of univentricular heart. In our studies figure 4-7 shows cardiac defects comprising of a the right and left atrial chambers connected by an atrial septa defect, pulmonary stenosis, aorta located posteriorily and a hypetrophied ventricular chamber of indeterminate morphology. Figure 8 from the third case shows a different cardiac defect consisting of a persistent truncus arteriosus, right and left atrial chambers with a prominent ASD and a single ventricle with a predominant right ventricle and an hypoplastic left ventricle. They represent various type of critical congenital defects requiring an urgent surgical intervention not available in our facility. The term univentricular hearts also includes hearts with biventricular AV connection (each atrium connects separately with its own ventricle), in which the right or left ventricles are too small as to be unable to sustain the pulmonary or the systemic circulation. Even if a ventricle possesses all components, it does not mean that it will be always of size enough to bear a normal function (16). The clinical manifestations hinge on the presence or absence of pulmonary outflow obstruction. Those with Severe pulmonary stenosis or atresia may result in profound hypoxemia and cyanosis (17). In our studies all cases present with a common complaint of severe cyanosis, fast breathing and a single case had in addition failure to thrive. These manifestations were within the first 24hours of life with the exception of failure to thrive. Clinical presentation of patients with these lesions becomes more obvious following closure of PDA. In the setting of a univentricular heart, a certain degree of pulmonary stenosis is physiologically desirable to prevent pulmonary over circulation and thereby improved the chances of survival (18). Infants with low pulmonary resistance present with congestive heart failure and failure-to-thrive (19). Their chances of survival become better with early surgical intervention although Sadon et al reported cases of two Nigerian adolescents with uncorrected different type of univentricular heart living up to ages 12 and 16 years (20). Tabbah et al reported a rare case of an 18-years-old female surviving a single ventricle malformation associated with a single atrium and a situs inversus viscerum (21).

Surgical management of neonates with critical congenital heart diseases is mandatory for better prognosis and several surgical procedures that ultimately result in a Fontan circulation is necessary for univentricular hearts. Body growth is severely impaired in Fontan patients and the technique is best performed as a sequence of staged operations during infancy and childhood (22, 23). Critical congenital heart disease should be one of the differential diagnosis in neonate presenting with severe cyanosis and fast breathing within the first week of life.

CONCLUSION – Mortality rate is from critical congenital heart defects although high is far worse in poor resource country where surgical intervention is lacking and the means of

radiological diagnosis a luxury that is not available and affordable. Despite a high index of suspicion following non-invasive techniques such as pulse oxymetry screening within 24 hours of birth. The required care is a great challenge. Failure of survival of all four cases call for a great concern and intervention by governmental agencies.

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