CASE SERIES

Double Outlet Right Ventricle with Anatomical Associations of Ventricular Septal Defect (VSD), Pulmonary Stenosis (PS) and Atrial Septal Defect (ASD) Presenting as Inoperable Cardiac Disease: case series

Josephat M **CHINAWA**[1]
John C **EZE**[2]
Herbert A **OBU**[1]
Bartholomew **CHUKWU**[1]

[1]*Department of Paediatrics*
College of Medicine
University of Nigeria
Teaching Hospital
Ituku/Ozalla
Enugu State, NIGERIA
[2]*Department of Surgery*
College of Medicine
University of Nigeria
Teaching Hospital
Ituku/Ozalla, Enugu State
NIGERIA

Author for Correspondence
Dr Josephat M Chinawa
Department of Paediatrics
UNTH PMB 01129 Enugu
Enugu State, NIGERIA
Postal Code 400001

E-mail:josephat.chinawa@unn.edu.ng
Phone: +234 806 398 1403

Received: October 15th, 2012
Accepted: March 20th, 2013

ABSTRACT

Background: Complex congenital cardiac abnormalities involving double outlet right ventricle (DORV) are uncommon diseases. They contribute to mortality and morbidity among children in Nigeria and the exact etiology is unknown. However, infective, genetic and environmental factors among others are commonly implicated. We present three rare cases of DORV with various associations which were inoperable.

Objectives: To present these congenital cardiac conditions which are rather rare and to highlight that early surgical correction improves the outcome in patients with these conditions.

Case Series

Case 1: OU was a 2-year old male who presented with a history of poor weight gain, bluish discoloration of the body of two years' duration. Echocardiography (2-D) result showed double outlet right ventricle (DORV) with pulmonary stenosis and membranous ventricular septal defect (VSD).

Case 2: CA was a 1-year old female who presented with a history recurrent infection and cyanotic spells from birth. Echo (2-D) findings showed double outlet right ventricle (DORV), severe pulmonary stenosis, membranous ventricular septal defect (VSD) and atrial septal defect (ASD).

Case 3: EA was a 5-month old female who presented with cough and deformities in the feet and upper limbs and poor feeding. Clinical examination revealed a small for age child with grade IV pansystolic murmur maximal at the left sternal border.

Conclusion: Double outlet right ventricle (DORV), with pulmonary stenosis and ventricular-septal defect (VSD), is a rare anomaly. Surgical correction offers good and long term benefits if the patient presents early.

Keywords: *Children, complex cardiac anomaly, Nigeria*

INTRODUCTION

In 1793, Aberanthy described a heart with the origin of both great arteries from the right ventricle.[1] The designation of "double outlet ventricle" was probably first reported by Braun et al in 1952.[1] The term refers to any anomaly in which both the aorta and pulmonary vessels originate predominantly or entirely from the right ventricle.[2] The purpose of presenting this case series is to highlight the rare and distinctive manifestation of associations of DORV, ventricular septal defect (VSD) and pulmonary stenosis (PS) in one patient, and the inoperable status when they present late, which will add to the available medical literature. It is important that clinicians understand that this anomaly is treatable in a good setting if detected early enough.

CASE SERIES

Case 1

OU was a 2-year old male who presented with a history of poor weight gain, and bluish discoloration of the body of two years' duration, and inability to walk at 2years of age. Clinical examination showed a child who was malnourished and cyanosed, with finger clubbing. There was a bulging hyperactive praecordium with pansystolic murmur maximal at the left sternal border with "VACTERAL" association. Two-D echocardiography showed double outlet right ventricle (DORV) with pulmonary stenosis (PS) and membranous ventricular septal defect (VSD). He had received saline pharesis on two occasions and could not be referred earlier for surgery in an appropriate centre because of financial constraint.

Case 2

CA was a 1-year old female who presented with a history of recurrent respiratory tract infections, failure to thrive and cyanotic spells from birth. Clinical examination showed a female child in obvious respiratory distress with no finger clubbing. She had pansystolic murmur at the lower left sternal border and a tender hepatomegaly.

Figure 1. Echocardiogram (2-D) of Case 2 (a)

Figure 2. Echocardiogram (2-D) of Case 2 (b)

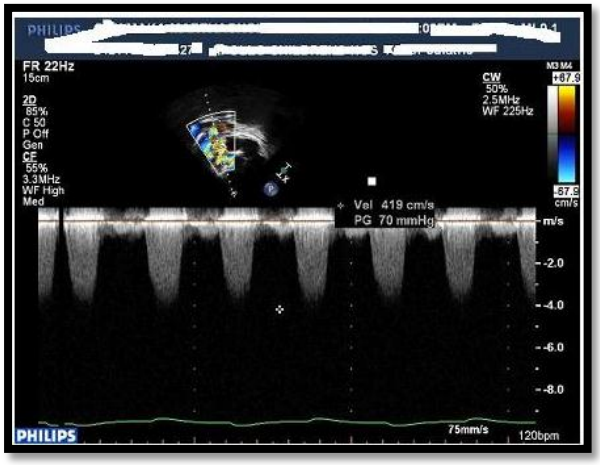

Figure 3. Echocardiogram (2-D) of Case 2 (c)

Plain chest radiography showed cardiomegaly and pulmonary oligaemia. Two-D echocardiography showed double outlet right ventricle (DORV), severe pulmonary stenosis, membranous VSD and atrial septal defect (ASD).

Angiography result showed a dilated branch of pulmonary artery and normal pulmonary venous drainage, and the 2-D echocardiography (ECG) result is attached; *see Figures I-III.*

Case 3

EA was a 5-month old female who presented with cough and deformities in the feet and upper limbs, and poor feeding. Clinical examination revealed a small-for-age child with grade IV pansystolic murmur maximal at the left sternal border. Two–D echocardiography showed a double outlet right ventricle (DORV) with a ventricular septal defect (VSD).

DISCUSSION

The term "double outlet right ventricle" (DORV) refers to a heterogeneous series of associated cardiac anomalies that involve the right ventricular outflow tract in which both of the great arteries arise entirely or predominantly from the right ventricle.[3] The anatomic dysmorphology of double outlet right ventricle can vary from that of tetralogy of Fallot (TOF) on one end of the spectrum to complete transposition of the great arteries (TGA) on the other end.

Witham in 1957 first used the term *double outlet right ventricle* to describe a partial transposition of the great arteries.[4] Several associated cardiac anomalies are associated with double outlet right ventricle, and many of these affect the clinical presentations and limits of the repair.

Occurrence rates of associated cardiovascular anomalies include: pulmonary stenosis 21-47% (most commonly observed with subaortic type VSD), atrial septal defect 21-26%, patent ductus arteriosus 16%, atrioventricular canal 8%, subaortic stenosis 3-30%, coarctation / hypoplastic arch / interrupted aortic arch 2-45%, mitral valve anomalies 30%.[5] Almost all our series had VSD, and some had ASD. The reason for VSD and ASD in our cases could be that their presence could help to reduce pulmonary resistance and, thus help to increase oxygen saturated blood to the system, thus averting cyanosis.

Between 1% and 3% of people born with congenital heart defects are affected by DORV.[6] Chromosomal abnormalities were reported in about 40% of reported cases in the medical literature.[6] Clinical features depend on the variants. The Fallot type presents exactly like TOF, the patient presenting with hypoxic spells, cyanosis and a systolic murmur heard from birth. These children are less likely to develop pulmonary obstructive vascular disease due to limitation of blood flow and pressure by pulmonary stenosis. The clinical presentations of our patients are similar to the features of the Fallot type.[5] The Taussig-Bing syndrome variety presents like transposition of great arteries (TGA), with cyanosis from birth and early onset of heart failure.

The reason for early heart failure in our patient was the association with coarctation of the aorta.[6] The subaortic VSD without pulmonary stenosis variant may present with clinical features that are similar to those of children with a large VSD and pulmonary hypertension. These children may have associated chromosomal abnormalities such as trisomy 13 or 18 and are likely to acquire pulmonary obstructive vascular disease in the absence of surgical repair, especially if the VSD is large.[7]

The main focus of investigation was 2-D echocardiography and ECG. Although trans-thoracic echocardiography (TTE) with Doppler has been shown to be of great value in diagnosis of patients with this anomaly, trans-esophageal echocardiography (TEE) provides a more complete and detailed data of the anatomy of DORV and other complex cardiac anomalies.[8]

In this series, 2-D echocardiography, ECG and plain chest radiography were used for the diagnosis of DORV. Trans-thoracic echocardiography (TTE) with Doppler was not done in our case due to financial constraints. However, 2-D echocardiography generally provides enough information for accurate and adequate diagnosis and provides the needed information to plan the surgical approach in neonates and young infants. Echocardiography can be used to correctly identify the relative position of the great arteries, the degree of sub-semilunar narrowing, position of the VSD and status of the mitral valve and left ventricle.[8] The presence of VSD, ASD and PS were detected by 2-D echocardiography in this series.

Common ECG findings in a child with double outlet right ventricle include right ventricular hypertrophy, right axis deviation, and occasionally, evidence of left ventricular hypertrophy. These findings were seen in our series. Prenatal diagnosis may be helpful in detecting this anomaly early and avert the morbidity and mortality that follow this disease. However, prenatal diagnosis is yet to gain grounds in the management of DORV in Nigeria and this service is not offered to patients in our setting.[9]

Medical management in the treatment of DORV is based on the combination of anatomical lesions and underlying physiology. However, double outlet right ventricle is a disorder that cannot spontaneously resolve, and the diagnosis alone is a sufficient indication for surgery.[3] In the setting of inadequate blood pulmonary blood flow, preserving ductal (i.e. patent ductus arteriosus) blood flow is vital. An infusion of prostaglandin E (i.e. alprostadil) is the standard of care until repair can take place.[3] When the contrary clinical picture of congestive heart failure is present, careful diuresis, digoxin, inotropic support and control of pulmonary blood flow by means of intubation.[3] We did not use prostaglandins in any of our patients, but digoxin and diuresis were used for those in failure.

Arterial switch operation (ASO) remains the preferred treatment for infants with double-outlet right ventricle and subpulmonary ventricular septal defect. However, associated anatomic defects are important risk factors.[10] Our cases were eventually referred abroad for cardiac surgery, but ended up being inoperable due to late presentation. They are currently been managed conservatively with saline pharesis and anti-heart failure regimen.

CONCLUSION

Double outlet right ventricle with its various variants is a rare congenital cardiac anomaly which could be fatal if not managed appropriately. Surgical correction offers good and long term benefits if the patient presents early and timely referral is warranted so as to avert death.

Consent: Written informed consent was obtained from the patients for publication of this case series and accompanying video images.

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ACKNOWLEDGEMENT

Our gratitude extends to the care givers and patients who were very cooperative. Finally we thank the almighty God whose assistance and ideas through the course of this work is priceless.

VIDEO

Image I:

VIDEO

Image II:

VIDEO

Image III: