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TRUNCUS ARTERIOSUS IN A 43 YEAR OLD MALE: CASE REPORT
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TRUNCUS ARTERIOSUS IN A 43 YEAR OLD MALE: CASE REPORT

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SUMMARY

In truncus arteriosus, the embryologic truncus fails to properly divide in-utero resulting in the pulmonary, aortic and coronary arteries arising from a single ascending portion of this trunk. This condition is usually fatal within the first year of life without correction. Over the past two decades, there has been a dramatic expansion in access to diagnostic echo-cardiography in Kenya and greater ability to diagnose congenital heart diseases. We present the case of a 43 year old male from western Kenya, newly diagnosed with heart failure due to truncus arteriosus. This case highlights the value of echo-cardiography in Kenya, and supports the need for surgical and interventional cardiac services to grow in tandem with these diagnostic capabilities.

INTRODUCTION

Truncus arteriosus, which is characterised by a single vessel arising from the heart, is a rare cyanotic congenital heart disease affecting approximately 0.8/10,000 live births (1). The defect occurs when the embryological structure known as the truncus arteriosus fails to properly divide into the pulmonary trunk and aorta. As a result, the aorta, pulmonary arteries, and coronary arteries all originate from the ascending portion of this single vessel (2). A ventricular septal defect is always present.

New-borns with truncus arteriosus present with overt heart failure and cyanosis, while Echocardiography with Doppler flow analysis is sufficient to confirm diagnosis in most patients (3,4). Early surgical intervention in the neonatal period has been associated with good outcomes (5) and delays in surgery usually leads to death within the first year of life following complications of pulmonary vascular disease that ensues (6). Survival into adulthood is very rare (7,8,9).

We present the case of a 43 year old male from western Kenya, newly diagnosed with heart failure due to truncus arteriosus. This case highlights the value of echocardiography in Kenya, and supports the need for surgical and interventional cardiac services to grow in tandem with these diagnostic capabilities.

CASE REPORT

A 43 year old male patient was admitted to the Moi teaching and referral Hospital with symptoms of congestive heart failure on the 14th of February 2012. He had been referred from a Level IV health facility (1), where he had initially presented with a two week history of worsening palpitations, a non productive cough and fever, following which he had been started on intravenous quinine treatment for smear positive malaria. He was referred to the Moi Teaching and Referral Hospital (MTRH) in Eldoret, Kenya after he subsequently developed symptoms of congestive heart failure.

He had been told he had an unspecified heart disease on the basis of clinical examinations early in his teenage years with complaints of occasional palpitations and somewhat limited exercise tolerance. The patient did not report any other specific investigation into this heart disease. Hitherto the current presentation, he had been able to carry out his daily activities without significant impairment.

His onset of symptoms worsened over the course of two weeks, initially presenting with a subjective fever and malaise. He then noted palpitations often aggravated by activity and, thereafter, developed shortness of breath on mild exertion. He also

experienced orthopnoea and paroxysmal nocturnal dyspnoea. He noted a non productive cough but reported no chest pain or wheezing. He reported no skin darkening or other colour changes in the past and noted no history of postural symptoms. This was his fourth admission for heart failure in the last two years, and up to the current presentation had been on lasix, aldactone and enalapril.

He had occasional headaches in the past and reported being on treatment for a convulsive disorder with phenobarbital in his childhood and had stopped several years ago after he became seizure free. He had not had any surgeries or blood transfusions in the past and had not been on treatment for any other chronic illness other than heart failure. The rest of the systemic inquiry was non contributory. He had no known drug or food allergies. He was a father of four, and worked as a tailor. He denied use of alcohol or smoking cigarettes and reported no drug abuse.

On examination he weighed 43 Kgs and was 162cm tall. His blood pressure was 90/43 mmHg with a pulse rate of 97 per minute with oxygen saturations of 74% on room air. He was not pale, plethoric, jaundiced, or oedematous. There was mild peripheral cyanosis and finger clubbing grade 3.The extremities were warm with a good capillary refill. No splinter haemorrhages or Osler's nodes were noted. His pulse was regular, bounding, synchronous and non-collapsing. He had a hyperactive precordium with a displaced heaving apex (7th intercostals space, anterior axillary line) with a palpable apical thrill. The first and second heart sounds were normal by auscultation with a pansystolic grade 5 murmur loudest over the apical and left parasternal regions radiating to the neck. The precordial heart rate was 98 beats per minute. He had no obvious chest wall deformities, his trachea was central and he had even chest expansion, resonant to percussion and with no areas of tenderness. On auscultation, there were good breath sounds with fine basal crackles bilaterally and no ronchi. Other system examinations were unremarkable.

His haemoglobin level was 23.1 g/dl, with a haematocrit of 64.4%. Sodium was 120 mg/dL, with a mild elevation of blood urea 120 mmol /l (normal range 2.5-10.7 mmol/L) and creatinine 127 mmol/ l (normal range 62-106 μmol/L) at time of admission. His chest x-ray showed cardiomegally with increased pulmonary vascular markings, and a left sided pneumonic process was appreciated. The electrocardiogram revealed sinus rhythm with a heart rate of 96 beats per minute, a left axis deviation and biventricular enlargement. Otherwise, he had normal intervals and waveforms.

On echocardiography, an apical four chamber view showed four subjectively well developed chambers with LV dilatation, a normal RV chamber size and with biventricular wall hypertrophy. The mitral and tricuspid valves were normal. The atrial septum was intact (Figure 1). The heart was in situs solitus with normal venous return.

Figure 1
Truncal stenosis

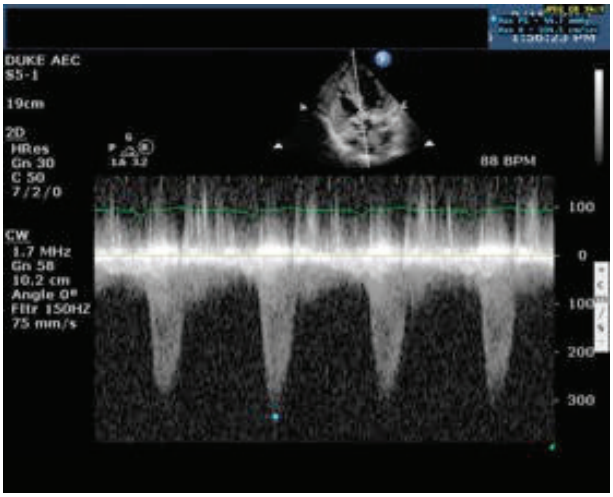


Figure 2
No pulmonary artery /no Right ventricular outflow

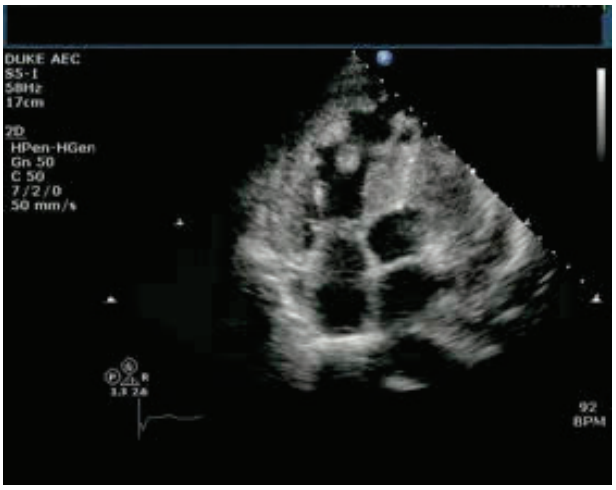


Figure 3
Short axis parasternal view of ventricular septal defect

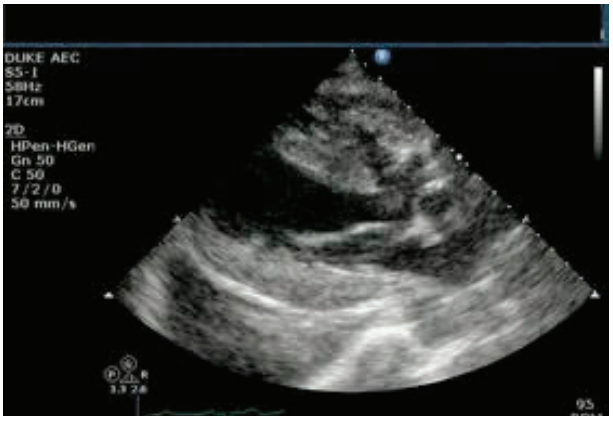


Figure 4
Trunks with pulmonary branch

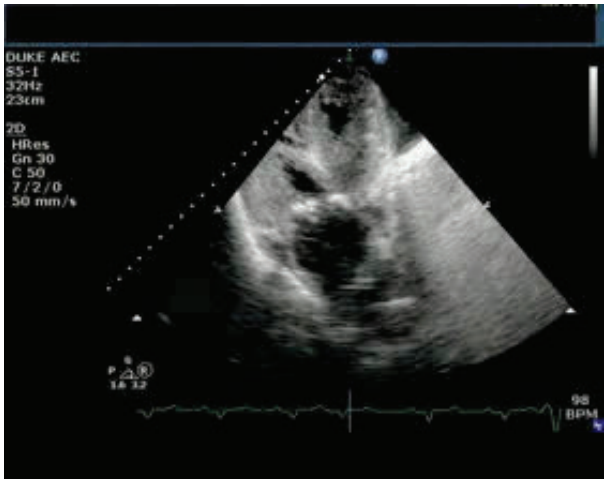
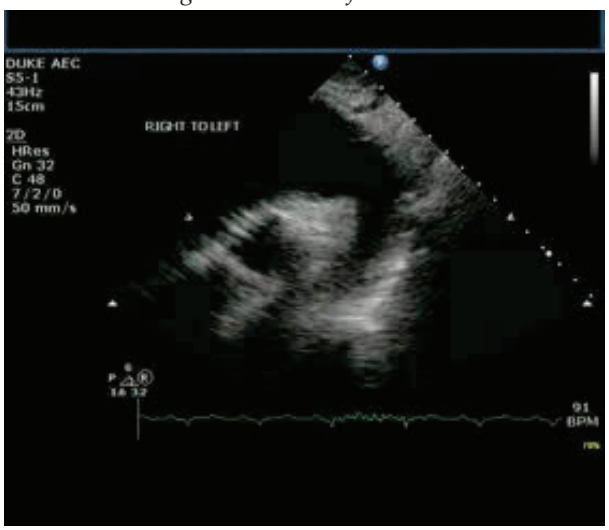


Figure 5
Right sided arch of the aorta



No patent RV outflow tract was visualised (Figure 2). On the parasternal long axis view, there was a trunk overriding a large VSD measuring 2.3cm (Fugure 3), with a shunt velocity of 3.3m/sec predominantly right to left (image 1). There was a branch originating from the trunk superiorly, identified as the pulmonic branch (Figure 4).

On 2D imaging,the dimensions were as follows:

Table 1
List of echocardiographic 2D Measurements

IVSD	1.3
LVIDd	5.9
LVIDs	4.1
LPWD	1.4
AO	3.8
LA	3.0
Estimated FS (%)	30.5
Estimated EF (Teichovs) (%)	57.3

All linear measurements in cm.

A short axis view sweep showed a tri-leaflet truncal valve with calcification and coronary arteries coursing adjacent to the vessel (Figure 5). A suprasternal evaluation revealed a right sided aortic arch .

Doppler interrogation revealed mild truncal valve regurgitation and moderate stenosis, and moderate tricuspid valve regurgitation .

A diagnosis of pneumonia with underlying truncus arteriosus in heart failure was reached. The patient was treated with antibiotics, diuretics and afterload reduction. Thereafter, his symptoms improved and he was discharged home after a family discussion about his heart condition. He is reported to have died in his sleep three weeks later.

DISCUSSION

Truncus arteriosus is a rare cyanotic congenital heart disease affecting approximately 0.8/10,000 live births, characterised by a single great vessel arising from the heart. It was first described in 1864 by Buchanan,(1) and one of the earliest reports of successful surgical correction in Africa was in 1971 by Rogers *et al* in South Africa (11). The condition is often associated with chromosome 22q microdeletion also seen in tetralogy of fallot and is also associated with DiGeorge syndrome and velocardiofacial syndrome (12).

The defect occurs when the embryological structure known as the truncus arteriosus fails to properly divide into the pulmonary trunk and aorta. As a result, the aorta, pulmonary arteries, and coronary arteries all originate from the ascending portion of this single vessel. There is always an associated ventricular septal defect.

The various subtypes of truncus arteriosus relate to the branching pattern of the pulmonary arteries as described by Collette and Edwards (2), They are type 1: main pulmonary artery arises from the truncal root; type II: each pulmonary artery arises directly from the posterior portion of the truncal root as separate vessels with separate orifices; and type III: each pulmonary artery arises directly from the lateral aspects of the truncal root as separate vessels with separate orifices.

The aorta contains combined output from the left and right ventricles resulting in cyanosis, which may be mild, depending on the severity of pulmonary vascular resistance. In some early cases, cyanosis may not be visible. Shortly after birth, pulmonary oedema may occur due to massive, rapid increase in pulmonary blood flow associated with a fall in pulmonary vascular resistance. Often the cause of death is metabolic acidosis with myocardial dysfunction, arrhythmia and cardiac arrest, with multi organ failure and this condition is usually fatal within the first year of life if uncorrected (6).

Surgical repair of truncus arteriosus may be complicated by other malformations such as interrupted aortic arch, coarctation of the aorta, significant truncal valve regurgitation, discontinuous pulmonary arteries, and truncal valve stenosis.

Though rare, survival into adulthood with unrepaired truncus arteriosus has been reported into the fourth decade (7, 8, 9). Survival into adult hood is thought to occur in the presence of pulmonary arterial stenosis as this minimises the amount of blood reaching the pulmonary capillary bed hence slowing development of pulmonary arterial hypertension. However, early surgery in the neonatal period has been shown to offer the best outcomes in patients with truncus arteriosus (8).

The patient in this case might have survived this long due to the truncal stenosis that may have reduced blood flow to the lungs. As no previous echocardiogram was available for review, it was difficult to judge whether this was a recent development, or had been there from the onset. Nevertheless, at this point in time, the patient's only treatment option was a heart and lung transplant, a procedure that is not available in east and central Africa, and whose costs neither he nor his family could afford.

This case report highlights the milestones in diagnosis of cardiac diseases in Kenya. Over the past two decades improvements in access to healthcare facilities, trained personnel and enhanced diagnostic capability such as 2D echocardiography, have affordably enabled the making of categorical descriptions of cardiac anatomy and function. (13). As a result there has been an increase in the identification of congenital heart diseases, and this trend has also been comparably described in India. (14,15,16). Though the patient was known to have a cardiac disease based on his symptoms, he was born at a time when cardiac diagnostic capability was nonexistent In Kenya.

Early evaluation by echocardiography now enables timely identification of congenital heart diseases before the onset of complications (17,18). Unfortunately, there still exist several barriers to intervention and long-term treatment. Among these barriers are the relatively high costs of healthcare and the limited number and capacity of medical centres providing interventional services. Thus, as cardiac diagnostic capacity grows in the region, there is an emergent need for interventional capacity to grow in tandem (5).

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