A Rare Presentation of Congenital Tetralogy of Fallot with Dysplastic Ascending Aorta

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Running title: Congenital Tetralogy of fallot with Dysplastic Ascending Aorta.

ABSTRACT:
Aim: A case report of a 23 year -old female, with congenital heart lesions and the clinical features slightly at variance with Tetralogy of Fallot.
Method: Combined clinical assessment of patient and evaluation with chest x-ray and gray scale cardiac ultrasound was done.
Results: Clinical assessment of the patient showed the following; palpitations, easy fatigability, cough, finger clubbing, orthopnoea, and lately significant precordial hyperactivity and tachypnoea on exertion. The chest x-ray and cardiac ultrasonography showed structural defects consisting of hypoplastic ascending aortic arch, with aortic root overriding inter-ventricular septal defect, right heart chamber enlargement, and congested pulmonary outflow track.
Conclusion: There was hypoplastic ascending aortic trunk (instead of hypoplasia of pulmonary trunk) a slight variation from the components of Tetralogy of Fallot. The probability is that this may have sustained the patient to adult hood. This is a rare occurrence in literature. Limitation of evaluation is also discussed.

Key words: congenital, heart, ultrasonography, x-ray, Tetralogy of Fallot, ventricular septal defect.

RUNNING ABSTRACT: We are presenting a case study of a 23 year -old female, with congenital heart lesions and clinical features slightly in variance with Tetralogy of Fallot, previously suspected to be complication of rheumatic heart fever. Chest x-ray and cardiac ultra-sonography have shown structural defect consisting of hypoplastic ascending aortic arch, with aortic root overriding inter-ventricular septal defect, right heart chamber enlargement, and congested pulmonary outflow track. This slight variation from the known components of tetralogy of fallot may have caused sustenance of life to adult hood. This is a rare occurrence in literature. Limitation of evaluation is also discussed.

INTRODUCTION

Tetralogy of Fallot (TOF) is a congenital malformation of the heart, which includes a large ventricular septal defect (VSD), associated with mal-alignment of the great arteries such that the aortic root overrides the VSD, thus communicating partly with the right ventricle. This is associated with stenosis of the right ventricular out flow tract and pulmonary valve with variable degree of hypoplasia of the pulmonary valve annulus and pulmonary artery. There is also right ventricular hypertrophy which is due to the arterial blood pressure in the right ventricle. [1,2,3]

In literature and in our experience, there are variable ways the abnormality of TOF presents. They are mainly dependent on the severity of the pulmonary stenosis, which often extends from the acyanotic end to the cyanotic end of the spectrum. Typically, cases of TOF tend to present with cyanosis because of the pulmonary stenosis which restricts pulmonary blood flow. Also about 25 percent of cases may present with right sided aortic arch, which are usually associated with mirror image branching of the following arteries, left brachiocephalic, right common carotid and right subclavian arteries. [1,2,3] There are many other congenital anomalies reported to have been found in association with TOF such as; Charge syndrome consists of coloboma, choanal atresia, heart defect of which 75-80% of the defect is TOF and are also associated with retarded growth, retarded development, genital hypoplasia, ear anomaly and deafness. [4] TOF was found in association with DiGeorge syndrome in less than two percent of occurrence.

[5] TOF has been found in combination with hypertrophic cardiomyopathy and Down's syndrome. [6]
In other setting of TOF, the following anomalies were found in literature; anomalous origins of right coronary artery from left anterior inter-ventricular coronary artery, absent pulmonary leaflet, anomalous origin of pulmonary artery from ascending aorta, and aorto-pulmonary window between ascending aorta and right pulmonary artery.[7-10] TOF has also been found in combination with coarctation of the aorta, anomalous dilatation of aortic root, aneurysmal dilatation of the ascending aorta compressing the right pulmonary artery and bronchus, and abnormal branching outlet of fourth aortic arch. [11-13]
In our search, there were no reported cases of hypoplastic ascending aorta as a component lesion in the entire variable forms of Tetralogy of Fallot, as seen in this patient, hence the reason for this case report. **(Figs 1-6)**
This type of anomaly may not be entirely out of place. Embryologically, the primitive heart tube which fuses to form single cardiac tube did undergo segmentations to form the sinus venosum, atrium, atrio-ventricular cannal, ventricle and finally bulbous cordis. The bulbous cordis is continuous with the aortic trunk. The bulbous cordis later divides into two parts, the ascending aorta and the pulmonary trunk communicating with left and right ventricles respectively. [14] The primitive origin of the ascending aortic outflow tract and pulmonary outflow tract are the same, hence it may not be surprising that contra-lateral anomalies may be possible as found in the other aforementioned literature search.

METHOD
This is a case study of a 23 year-old female, Benin by ethnic origin, who presented with palpitation for 7 years, easy fatigability for 10 months and cough for 10 days. This was preceded by dyspnoea on exertion, which caused the patient significant anxiety. She gave history of being treated at age of 3 years for pre-cordial hyperactivity and tachypnoea on exertion. However, she was not followed up. She learnt to regulate her activity, but noted occasionally that she had cough productive of whitish sputum, sometimes with streaks of blood and chest pain. There was no history suggestive of exposure to anybody with tuberculosis, and she had never been treated for it. She has no history of weight loss, night sweats, fever, recurrent sore throat, paroxysmal nocturnal dyspnoea or pedal oedema.
She is single, had a secondary school education, and is the 3rd child of 4 children in a monogamous family setting. She does not abuse any drug, and has noticed among her other siblings that she has the smallest stature and her menarche was at the age of 18 years. She has regular menstrual cycle of 3-4 /21-24 days.
On examination, she was asthenic in build, but mentally stable with no neurological deficit. She had no central cyanosis, no jaundice, was not pale and there was no pedal or truncal oedema, but had finger clubbing. She had mild respiratory distress (RR 30 cycles per minute) and lung crepitations. The cardiovascular system showed a blood pressure of 120/90mmHg, and a pulse rate of 106 beats per minute regular but of low volume. There was raised jugular venous pulsation by 4cm. There was a pan-systolic murmur on auscultation of the heart at the 5th and 6th intercostal spaces left parasternal line and at the apex. The abdominal palpation showed enlarged liver (6cm below right costal margin mid axillary line) and mild right hypochondrial tenderness. There was no other physical sign elicited.
The working diagnosis before referral for radiological evaluation was mitral valve disease possibly due to rheumatic heart disease. The following investigations, full blood count, fasting blood sugar, serum urea /electrolytes, serum calcium/phosphate, total serum cholesterol/differential and Urinalysis were normal. The retroviral screening and sputum for AAFB were negative.
The following radiological evaluations, Chest x-ray (Posterior anterior and lateral view) and Cardiac ultrasonography (echocardiography) were done.

RESULTS OF RADIODIAGNOSIS
The posterior anterior chest x-ray view showed cardiomegaly, with significant bulge of the pulmonary bay, and upturn of the outline of the apex of heart. The aortic arch was not outlined instead a concave right supra cardiac bare space

was demonstrated hence the suggestion that the ascending aorta was significantly hypoplastic. The carinal angle was noted to be less than 90 degrees confirming insignificant or no enlargement of left atrium. There was lung plethora with significant congestion and dilatation of the lung vessels and evidence of blunted left costo-phrenic angle. **(Fig 1)**

The lateral view of the chest radiograph showed loss of retro-sternal space due to enlarged heart but there was no appreciable reduction in dept of the retro-cardiac space adjacent to the outline of left ventricle**. (Fig 2)** This was highly suggestive of right atrium / right ventricular enlargement. The surrounding ribs of the thoracic cage were normal. The preliminary consideration was a right sided cardiac chamber enlargement, with pulmonary venous congestion.

Cardiac ultrasonography (echocardiography) confirmed enlarged right atrium and right ventricle with significant hypertrophy of the wall of the right ventricle. There was no significant enlargement of left ventricle or hypertrophy of its wall in comparison to right ventricle. The left atrium was not enlarged as well. There was sub-aortic inter-ventricular septal defect and features suggestive of hypoplastic ascending aorta with the aortic root overriding the inter-ventricular septal defect. There was congestion and significant enlargement of the pulmonary outflow tract. The cardiac rhythm by m-mode tracing was regular, but was increased in rate at about 103-106 beats per minute. **(Fig 3-6)** There was hepatomegally about 6cm below costal margin right mid-axillary line, but the parenchymal and sinusoidal pattern was normal. The kidneys, pancreas and spleen were normal in size and architecture. There was no ascites, gross intra-abdominal mass or pelvic collection.

Fig 1: Posterior-anterior Chest radiograph showing enlarged heart (cardiothoracic ratio of 17.2/25cm), bulging of pulmonary bay, upturn of the left apical outline of the heart, and concave bare space at the site of the arch of aorta. The trachea carina angle is within acute angle formation. There is significant lung plethora with congestion of pulmonary vessels. The surrounding thoracic bones are normal.

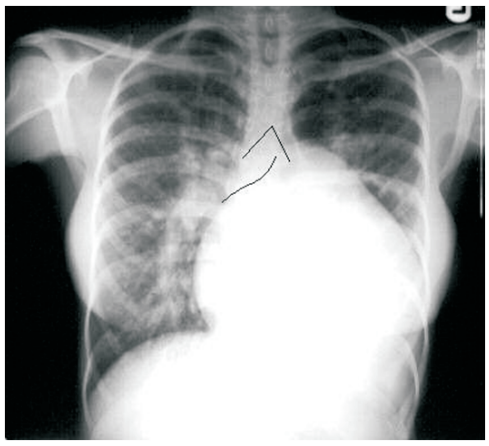
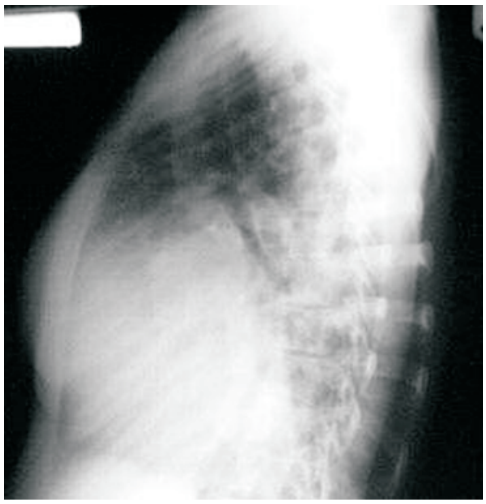


Fig 2: Lateral Chest radiograph showing loss of retro-sternal space due to anteriorly enlarging heart. There is no significant lost of retro-cardiac space.



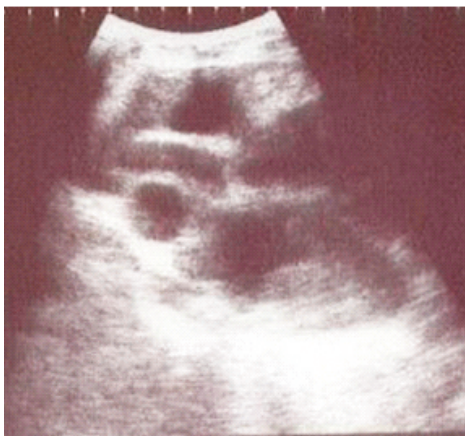


Fig 3: Cardiac sonography in cephalic angulated left parasternal long axis view is showing the root of ascending aorta overriding inter-ventricular septum with the **thin** arrow mark coming from the left ventricle, **thick** arrow mark from the right ventricle. Note the plastic tunnel caliber and narrow lumen of the ascending aorta (thin lines) measuring 1.093cm compare to pulmonary trunk which lumen is double the size measuring 2.45cm (Two end arrow)

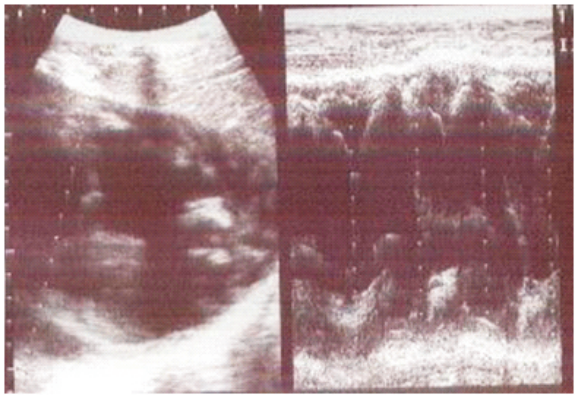


Fig 4: Cardiac sonography in partial angulated left parasternal long axis view showing hypertrophy of right ventricular wall measuring 2.01cm (long arrow), with significant enlargement of its lumen in comparison to left ventricle. Left ventricular wall measures 1cm. Note the site of interventricular septal defect. (Short arrow).

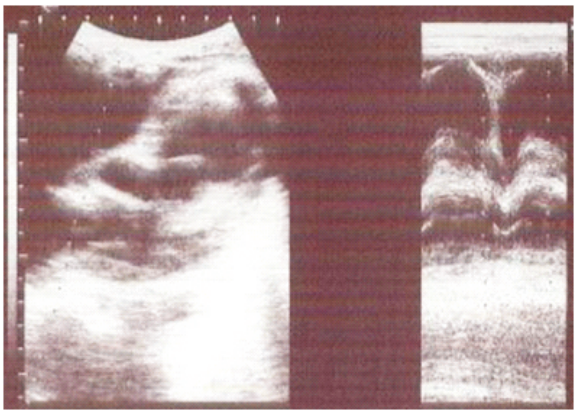


Fig 5: Cardiac sonography showing enlarged right atrium. Note the site of outflow inter-ventricular septal defect (arrows).

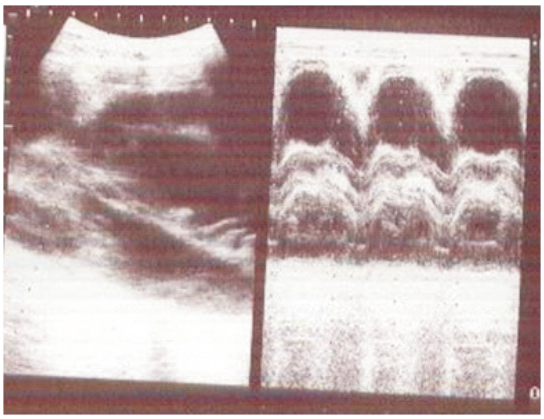


Fig 6: Cardiac sonography showing the direction of left ventricle (short arrow) and right ventricle (long arrow), blood flow contribution into the hypoplastic ascending aorta. Note the small size of the left chamber in comparison to the right chamber. These diameters are base on left parasternal long axis view, Right ventricle (5.1cm), left ventricle (2.28cm), Right atrium (3.5cm), and Left atrium (2cm)

DISCUSSION

This case study has shown that the patient had hypoplastic ascending aortic arch. This is in contrast to the usual component of congenital TOF which includes, normal caliber ascending aorta (25 percent right sided aortic arch) with aortic root overriding the outflow inter-ventricular septal defect, pulmonary stenosis and reactive right ventricular enlargement with hypertrophy of the wall. The clinical presentation of the case has shown the effect of the hypoplastic ascending aortic outflow tract on the patient. They include the following; asthenia, easy fatigability, palpitation, and orthopnoea on exertion.

There was congestion of the pulmonary outflow tract as well as congestion of the lung which may be due to restriction of blood flow by the overriding hypoplastic ascending aortic outflow tract from either venticle of the heart. The congestion of the lung and poor systemic distribution of blood may lead to chronic hypoxia which is associated with the following clinical presentations; tachypnoea, paroxysmal nocturnal dyspnoea and finger

clubbing. [3] This patient was noted to have mild tachypnoea and finger clubbing. There was no history of paroxysmal nocturnal dyspnoea. The findings were considered to be in line with the stage at which patient presented. It is anticipated that the patient may develop paroxysmal nocturnal dyspnoea, if corrective surgery is not undertaken. The prognostic value of such intervention is uncertain. Studies have shown excellent results after repair of TOF, and most patients afterward tend to have good living condition and the causes of sudden morbidity were also elicited as follows: ventricular arrhythmias, cardiac conduction defect and pulmonary regurgitation. [15-17]

The slight variation in structure of the heart in our patient may have made it possible for life to be sustained up to adulthood but the effect gradually worsened as patient grew older. We elicited that the subjects had few early symptoms before adulthood which she was able to cope with, by regulating her activity until the present clinical signs and symptoms developed.

The other symptoms such as tender hepatomegaly elicited on examination can be explained to be due to impeded venous return, due to failing and congested right heart chamber. Ultrasound scan also confirmed enlarged liver about 6 cm below costal margin at right mid-axillary's line but noted that the liver parenchyma and sinusoidal pattern were normal. There was slight elevated diastolic blood pressure of 90mmHg and increased pulse rate of 103 beats per minute which are likely due to compensatory vaso-regulation from low arterial blood volume.

The combined use of chest x-ray and cardiac ultrasound study, has thrown significant insight into the structural defect in the heart. In spite of limitation due to inadequate imaging technology, we were able to infer the clinical/pathological consequences and complications of the patient's cardiac lesion and the disorder it may have caused in systemic blood circulation. However the exact structural defect, pressure gradients in the ventricles, and anomaly in the cardiovascular blood flow can be further achieved, with improved Radio-diagnostic technology.

Using Duplex Ultrasound machine consisting of color Doppler and spectral wave tracing, it is possible to measure pressure gradients in the heart chambers, and suggest direction of blood flow.

The improvement in computer application to achieve three dimensional imaging in multislice or multidetector CTscan, and the multiplanar property of MRI, can be used to delineate the exact structural defects of congenital anomalies of the heart. MRI is better in anatomical and functional studies. However, the high cost of maintaining the technology, has led to their low availability in many African countries. The disadvantage associated with CT scan is the relatively high dose of ionizing irradiation to tissue of the body. These limitations have necessitated the need to put into effective use equipment within the scope of our technology. This patient may be able to benefit from temporary relief with corrective surgery, if surgeons are able to reconstruct a shunt to relieve congestion in right ventricle and improve blood flow through the ascending aortic outflow tract.

CONCLUSION

This case study suggests that the patient may have a rare variant of tetralogy of fallot as shown by presence of hypoplastic ascending aortic outflow tract, instead of the usual hypoplastic pulmonary artery outflow tract. However the other components of the TOF syndrome were elicited such as ascending aortic root overriding outflow inter-ventricular septal defect, and right ventricular hypertrophy. The unusual combination may have led to improved longevity of the patient.

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