

COMPLEX EBSTEIN'S ANOMALY IN A 80YEAR OLD NIGERIAN WOMAN

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SUMMARY

There is a gradually growing interest in the subject of congenital heart diseases as many children born with them are now growing into adulthood. The reasons are twofold: increasing awareness among clinicians as well as major advances in the diagnosis and treatment of congenital heart diseases in children. A case of Ebstein's anomaly (EA), one of the most important congenital diseases of the tricuspid valve, in an 80 year old woman referred to the cardiology clinic of the University of Benin Teaching Hospital in Benin City, Nigeria, is hereby reported. It is a rare condition. The features demonstrated in this case report tend to suggest that there may be several adult survivors of congenital heart diseases. These may coexist with other systemic illnesses such as systemic hypertension.

Key words: Congenital; Ebstein's anomaly; Nigerian.

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INTRODUCTION

Congenital heart diseases (CHD) in adults are beginning to generate great interest among researchers in recent times. This is because many children born with CHD are now living into adulthood¹⁻³ - a consequence of major advances in the diagnosis and management of heart diseases in children as well as increased awareness on the part of clinicians. Ebstein's anomaly (EA), is one of the most important congenital diseases of the tricuspid valve. Although a rare disease, there has been growing interest in it from different parts of the world¹⁻³. A report from Beirut, Lebanon gives its relative frequency as 4% of cardiac malformations with "grown up" congenital heart disease (GUCH)². About 15 cases have been reported in South Africa and few have been reported elsewhere⁴⁻⁶. There is paucity of reports in Nigeria. This case is noteworthy because of its rarity but more importantly because of the age of the patient; her very poor village backgrounds devoid of any form of medical facility, and the attendant severe systemic hypertension, hence this report.

THE CASE REPORT

Mrs. C.I is an 80 year old Igbanke lady from Delta state of Nigeria. She was referred on the 30th of January, 2006 to the cardiology clinic from the general practice clinic of the University of Benin Teaching Hospital in Benin City Edo state of Nigeria. She was Para 8+0, 4 alive (four children had died between theages of 4 to 7years following minor febrile illnesses). She had a two- year history of

abdominal swelling, and palpitation, recurrent facial swelling, effort intolerance, cough and difficulty with breathing. She could not recall any relevant perinatal history such as uses of angiotensin converting enzyme inhibitors, warfarin or lithium carbonate by her mother though the use of traditional medication could not be totally excluded. She had been told that she was hypertensive two years earlier but was not on antihypertensive medications. There had been gradually increasing breathlessness and body swelling in the previous four months. There was no family history of EA or any other congenital heart disease. Physical examination revealed an elderly lady, with a height of 1.50 meters and weighing 57 kg. She was not pale but cyanosed, had anasarca with a grossly distended abdomen, peau d'orange appearance of the skin over both breasts and marked bilateral pitting pedal edema. Cardiovascular examination showed she was in atrial fibrillation with a pulse rate of 134 beats per minute; blood pressure of 178/114mmHg (sitting) and the jugular venous pulsation was raised to the level of the ear lobe. There was a non-heaving apex beat at the 5th left intercostal space lateral to the mid clavicular line; a triple rhythm and a pan systolic murmur which was loudest at the 4th left sternal edge. There was marked ascites and a pulsatile liver.

A provisional diagnosis of chronic congestive cardiac failure due to tricuspid regurgitation with possible renal failure was made. The electrocardiogram showed atrial fibrillation with rapid ventricular response, low voltage complexes in limb leads and non specific T-wave abnormalities. The chest x-ray showed cardiomegaly with unfolded aorta, bilateral hilar fullness, perivascular and endobronchial cuffing

with upper lobe vascular diversion. In addition there was a right pleural effusion with contiguous infiltrates, and calcified aortic knuckle. The serum electrolytes and urea showed mild renal impairment with a serum creatinine of 2.5mg/dL and blood urea of 42 mg/dL. Echocardiography showed normal sized left atrium and left ventricle, a markedly enlarged right atrium measuring 103.8mm in longitudinal dimension and a small right ventricle (figs 1 and 2.). The anterior tricuspid valve leaflet (TV1) (see fig 1.) was elongated and redundant and was abnormally attached to the ventricular free wall, while the septal leaflet (TV3) was displaced down into the right ventricle and located (68.0 mm) far beyond the level of the mitral valves (fig. 3). There was a moderate sub-aortic ventricular septal defect, mild hypertrophy of the left ventricle and a small atrial septal defect. The pericardial layers were thickened and there was a mild pericardial effusion. Based on the above findings, a diagnosis of Ebstein's anomaly with chronic pericarditis in the setting of systemic hypertension was made.

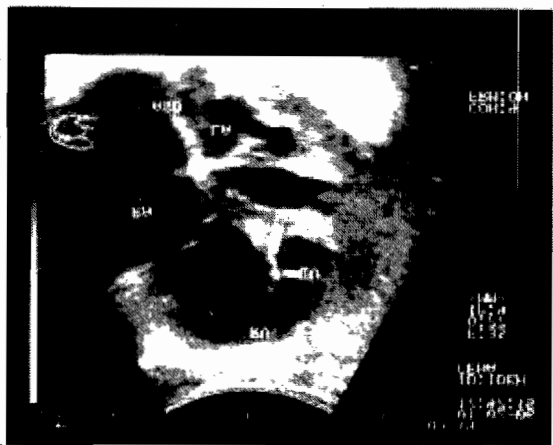


Fig. 1. Demonstrating Atrialized Right Ventricle (ARV) in Ebstein's Anomaly (RA=right atrium, LA=Left Atrium, LV =Left Ventricle, RV = Right Ventricle).

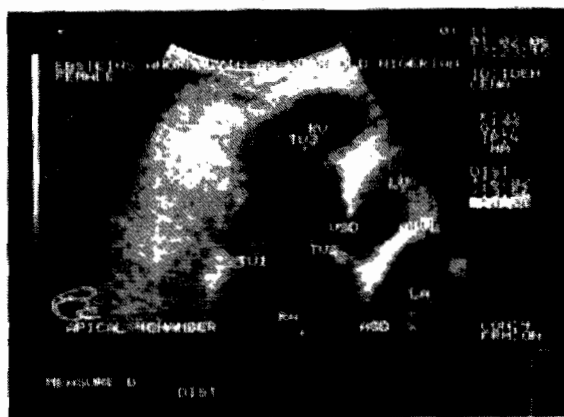


Fig.2: The Same Patient, Showing Distorted Anatomy and a sub aortic VSD in Apical View Connecting Atrialized Right Ventricle to the Left Ventricle, and also an ASD. (RA=right atrium, LA=left atrium, TV1= anterior tricuspid valve leaflet, TV2=posterior leaflet, VSD= ventricular septal defect, ASD= atrial septal defect, TV3=downwardly displaced septal leaflet, RV=right ventricle, LV=left ventricle, MVL = mitral valve leaflets).

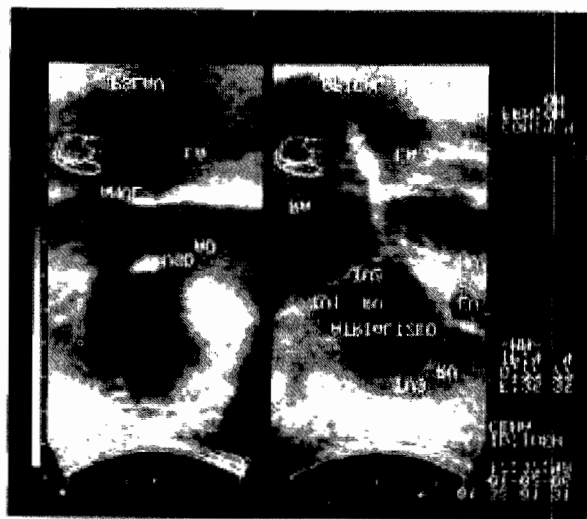


Fig.3: Atrialized right ventricle connected via VSD to the left ventricular outflow tract (3A) (parasternal view).Elongated and abnormally attached anterior leaflet; also posterior and septal leaflets displaced into the right ventricle (3B) (apical view).

The patient has responded satisfactorily to convectional treatment of congestive cardiac failure and systemic hypertension though she still maintains a mild degree of cyanosis and effort intolerance.

DISCUSSION

Ebstein's Anomaly which was first described 150 years ago⁸ belongs to the cyanotic group of CHD. It is one of the rare CHD in which survival into adulthood is usually the case⁷. It is due to a defect in the tricuspid valve in which the septal and posterior leaflets are displaced downwards into the right ventricle (RV) while the anterior leaflet is malformed and abnormally attached to the right ventricular free wall^{2, 6, 7}. The proximal part of the RV is thin walled and continuous with the right atrium (RA). The functional RV is small and is made up of the apical

and infundibular portion of the RV. The papillary muscles and chordae are malformed and the two involved leaflets are abnormally attached to the right ventricular endocardium; therefore the valve allows regurgitation into the large RA. In about 80% of cases an atrial septal defect is present causing right to left shunt and subsequent cyanosis. A ventricular septal defect is present in about a third of cases. Such patients are at risk of paradoxical embolism and sudden death². While no specific genetic inheritance has been reported, there is documentation on familial association³. There is a high risk of recurrence in the offspring just like in any other congenital heart defect. Maternal exposure to teratogens has been incriminated in the occurrence of this malformation in the offspring^{3,7}. The mother of an 80 year old Nigerian illiterate, like the case being reported, from a remote village, obviously belonged to the generation who depended on traditional concoctions and herbal remedies for all their health needs. The rapid succession of childhood deaths in the patient's offspring raises the suspicion of a possible congenital predisposition to early death though this information was absent.

The clinical features of EA depend on several factors which include: the severity of the tricuspid incompetence, the presence of an atrial septal defect, the degree of right ventricular involvement, the presence of other anomalies and the presence of pulmonary hypertension^{1,2, 8, 9}. In the neonate, the most common important associated defect is pulmonary stenosis or atresia. Others are ostium primum type of atrial septal defect and ventricular septal defect alone or in combination. Thus the clinical picture is variable because of the spectrum of the pathology. The case being reported not only had an atrial septal defect causing a right to left shunt but there was also a moderate ventricular septal defect thus limiting the cyanosis as well as reducing left ventricular volume overload.

The neonate usually presents with cyanosis, a cardiac murmur and severe CCF^{2,7}. In the adult, the onset is insidious, the commonest symptoms being fatigue, exertional dyspnoea and cyanosis. About 25% of cases present with cardiac arrhythmias in the form of supraventricular tachycardia. Physical examination usually reveals, elevated JVP with a prominent v wave, a prominent systolic pulsation of the liver, ascites, systolic thrill and murmur of tricuspid regurgitation, widely split first and second heart sounds (atrial septal defect and pulmonary hypertension) and a triple or quadruple rhythm or quintuple combination of sounds.⁷ The natural history of Ebstein's Anomaly ranges from poor

neonatal death to relatively asymptomatic survival into adulthood even to advanced age⁴. The oldest recorded survivor presented at the age of 79 years and died at 85 years of age. The case in point also presented at 79 years of age and appeared to have been precipitated by severe systemic hypertension rather than the complexity of her intracardiac defects. The rapid response to antihypertensive drugs and brief treatment for heart failure tends to support this idea.

The history is very important: family history; prenatal and postnatal history including maternal exposure to teratogens such as lithium and other drugs like angiotensin converting enzyme inhibitors and angiotensin II receptor antagonists, warfarin, amiodarone, should be sought for (especially in the first three months of pregnancy).

The electrocardiographic findings are giant P-waves; prolonged PR interval, prolonged terminal QRS depolarization; and various degrees of right bundle branch block⁷. The presence of Wolf- Parkinson White - type B pattern increases the risk of cardiac arrhythmias. The diagnosis of EA is by 2-D echocardiography using the apical four-chamber, sub costal, or medially angulated parasternal long axis view. Identification of elongated, redundant leaflets with dominant chordal attachments; septal leaflet insertion $>8\text{mm}/\text{M}^2$ when normalized for body surface area or a maximum displacement of the TV of $>20\text{mm}$ is diagnostic^{1,6,10,11} in the adult. In the case in point, the tricuspid valve displacement is 68.0 mm beyond the level of the mitral valves

Adult forms of EA are often well tolerated but pose difficult problems of management^{5,7,11-12}. In females there is high risk of fetal prematurity and fetal wastage.

The treatment is both medical and surgical. Medical treatment involves use of diuretics and digoxin for the reduction of cardiac failure state; prophylaxis should be given for infective endocarditis^{2,3}. The factors that affect the outcome include New York Heart Association (NYHA) functional class, the size of the heart, the presence or absence of cyanosis, high mitral-tricuspid valve displacement indexed to body surface area, and the presence or absence of paroxysmal atrial tachycardia^{1,8,11-13}. Supraventricular arrhythmias and Wolf Parkinson White syndrome pose no extra mortality risk whereas, the presence of cardiac failure of NYHA III and IV, and high cardiothoracic index are associated with increased mortality^{8,11}.

The definite treatment is surgical and involves reconstruction of the TV and marsupialization of the atrialized portion of the RV^{7, 13}. Surgery becomes necessary when functional status declines or signs of

prognosis are observed⁸. Treatment options are best done using Carpentier's classification A-D¹³⁻¹⁴. Type A in which RV is adequate has the best prognosis and is usually managed conservatively. Types B (in which there is a large atrialized segment and mobile anterior leaflet of the TV) and C (in which there is restricted movement of the anterior leaflet will require reconstructive surgery. Type D where there is near complete atrialization of the RV (Uhl's syndrome) is usually associated with poor prognosis. Genetic counseling regarding the occurrence, etiology, inheritance, recurrence risk and prenatal options should be explained to parents. Fetal echocardiography at 16-18 weeks should be made available to mother for early detection of fetal anomaly³.

CONCLUSION

The discovery and proper documentation of congenital heart diseases in Nigeria depends on the regular use of available diagnostic facilities especially echocardiography, electrocardiography and chest x- ray against the background of a good physical examination. The features demonstrated in this case report tend to suggest that there may be several adult survivors of congenital heart diseases. The later may coexist with other systemic illnesses such as systemic hypertension.

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