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Ebstein Anomaly: A Clinical Case Series among Adolescents in Guinea

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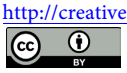


Abstract

Ebstein's anomaly is a rare congenital heart disease. We report a series of 4 cases of Ebstein's anomaly discovered in adolescents. The mean age was 13.75 ± 0.96, with 3 boys and one girl. Three were in heart failure. The EKG showed atrial hypertrophy (1 case), right bundle branch block (1 case), Wolff-Parkinson-White (1 case), and atrial fibrillation (1 case). Cardiomegaly was present in all patients with a mean cardiothoracic index of 0.77 ± 0.03. On echocardiography, the mean apical displacement of the tricuspid septal leaflet was 22.5 ± 3.30 mm/m^2, the Celermajer index was Grade 2 in 2 patients and Grade 4 in 2 patients. Depletive medical treatment was prescribed to 3 patients. Anticoagulant therapy was initiated in one patient. Two are awaiting surgery. One death was recorded, it was at Grade 4 of the Celermajer index.

Keywords

Ebstein Anomaly, Adolescent, Congenital Heart Disease, Guinea

1. Introduction

Ebstein's disease is a rare congenital heart disease, accounting for less than 1% of all congenital heart defects with a prevalence of 1 in 200,000 live births [1] [2].

It was first described by the pathologist Wilhelm Ebstein in 1866 when he performed an autopsy on a worker who suffered from exertional dyspnoea all his life, showed signs of cyanosis, and died of a sudden cardiac arrest [3].

It is characterised by the low insertion of one or both leaflets of the tricuspid valve leading to aterialization of the right ventricle [4] [5].

The clinical spectrum is wide and heterogeneous, ranging from minor asymp-

tomatic forms to severe forms with massive tricuspid regurgitation and right heart failure [6]. The adolescent and adult form often manifests as arrhythmia [1].

In the literature, we found a series of cases published in Africa, notably 9 cases of Ebstein's anomaly in Tunisia, and one case at the CHU of Libreville, among others [7] [8].

In Guinea, no case has yet been published. We report a series of four cases of Ebstein's disease discovered in adolescence.

2. Observation

These were 4 adolescents in whom the diagnosis of Ebstein's disease was retained on the basis of clinical examination, electrocardiogram, chest radiograph, and echocardiography.

Patient 1

M.B. aged 14 years with no previous history was seen for NYHA II dyspnoea progressing to NYHA III. The saturation was 76% at rest. Cardiac auscultation revealed a B4. She had hepatomegaly. The EKG showed a regular sinus rhythm with a rate of 71 cycles/min, right atrial hypertrophy, and signs of right ventricular overload. The chest X-ray showed cardiomegaly and peripheral pulmonary hypovascularity. The cardiac Doppler echo showed a mitro-tricuspid shift of 23 mm. The surface area of the atrium and the atrialized right ventricle was 47 cm^2. The surface area of the right ventricle and the left cavities was 29 cm^2. Celermajer index (right atrium + ventricle area atrialized)/(right ventricle area + left ventricle area + left atrium area) was 1.6. Patent foramen ovale is the associated anomaly. She has treated with Furosemide 40 mg daily. The course was marked by regression of heart failure symptoms. She had a relapse of heart failure while travelling outside Guinea. She died there.

Patient 2

B.M.L., a 15-year-old adolescent, was seen for NYHA III dyspnoea and oedema of the lower limbs. Examination revealed a B4, turgidity of the jugular veins, hepatomegaly and oedema of the lower limbs. The EKG showed right atrial hypertrophy and complete right bundle branch block.

The frontal chest X-ray showed cardiomegaly (ICT = 0.81) (**Figure 1**). Echocardiography concluded that the patient had Ebstein's disease with an anterior tricuspid valve retained by tethers. The apical displacement of the tricuspid septal valve is 30 mm or 26 mm/m^2. The surface area of the atrium and atrialized right ventricle was 65 cm^2; the surface area of the right ventricle and left chambers was 70 cm^2. The Celermajer index was 0.92.

There was a major coaptation defect with massive tricuspid insufficiency ascending into the suprahepatic veins. The associated sign was a permeable foramen oval shunting right-left.

Therapeutically, he was receiving Furosemide 40 mg daily. Signs of heart failure are improving. He is awaiting surgery.

Patient 3

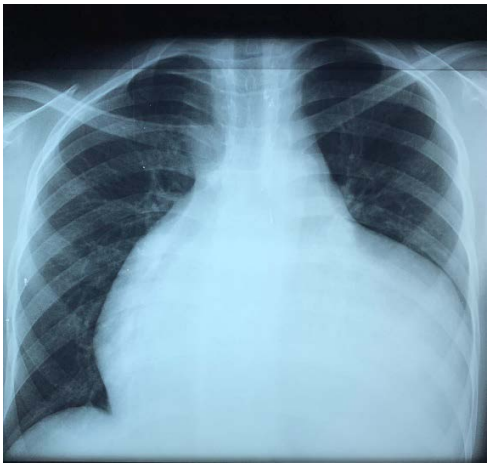


Figure 1. Frontal chest radiograph showing significant cardiomegaly.

M.S., an 11-year-old adolescent, was seen for NYHA II dyspnoea and palpitations that had been evolving for six months. On clinical examination, he was in good general condition. Cardiac auscultation revealed a 4-stroke sound. There were no signs of right heart failure.

The pulses were felt in all four limbs.

The frontal chest X-ray showed cardiomegaly with an ICT of 0.78. The EKG showed a posterior-right Wolff Parkinson White (**Figure 2**).

The echocardiography was in favour of Ebstein's disease with acceleration of the septal valve. The apical displacement of the tricuspid septal valve was 25 mm or 23 mm/m^2. The interval between the closure of the mitral and tricuspid valves at MT was 160 ms (**Figure 3**). The surface area of the atrium and the atrialized right ventricle was 37 cm^2, the surface area of the right ventricle and the left cavities was 47 cm^2, the Celermajer index was calculated at 0.79.

The tricuspid insufficiency was laminar. The associated sign was mitral prolapse with a Grade 2 leak.

He is clinically stable and asymptomatic, we examine him every 6 months for operative indications.

Patient 4

F.C., a 13-year-old adolescent, was seen for NYHA III dyspnoea. Examination revealed a mitral systolic murmur 2/6, hepatomegaly, and turgidity of the jugular veins. The ECG showed atrial fibrillation (**Figure 4**). Chest X-ray showed cardiomegaly and clear lungs.

Ultrasound showed a septal valve accouchement with an apical displacement of the tricuspid septal valve which was 22 mm/m^2. The area of the right atrium and ventricle was 68 cm^2; the area of the right ventricle and left cavities was 34 cm^2. The Celermajer index was 2. Tricuspid insufficiency was massive. The associated anomaly was a Grade 1 mitral leak. He was taking Furosemide 40 mg daily and Acenocoumarol.

The evolution under this treatment is marked by an improvement in the signs of insufficiency. He is awaiting surgery.

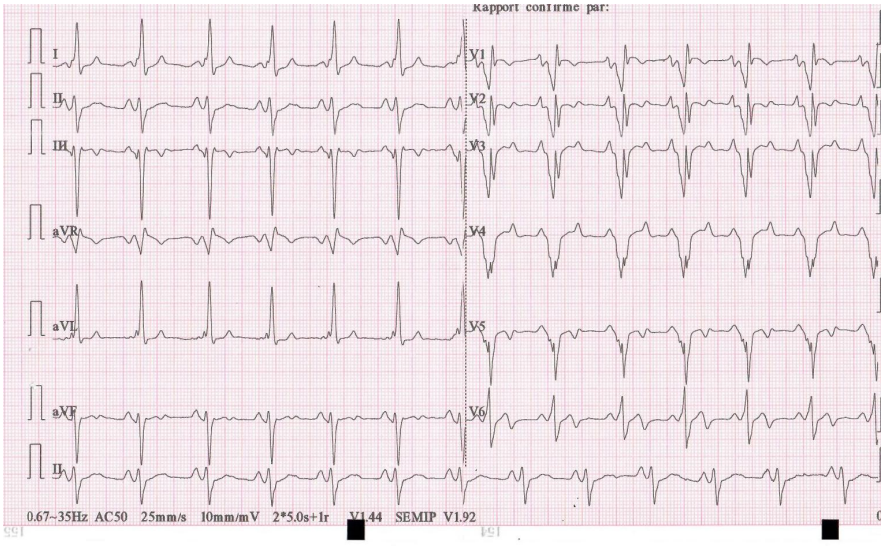


Figure 2. EKG showing posterior-right Wolff-Parkinson-White.

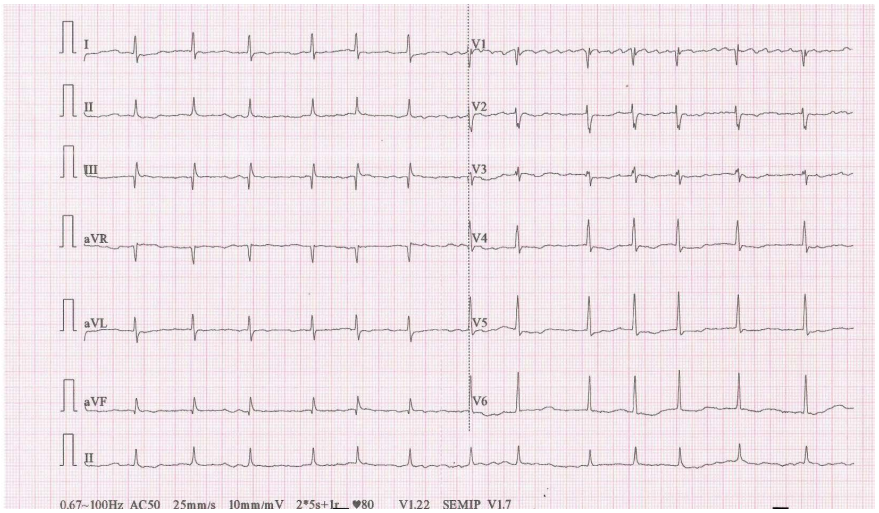
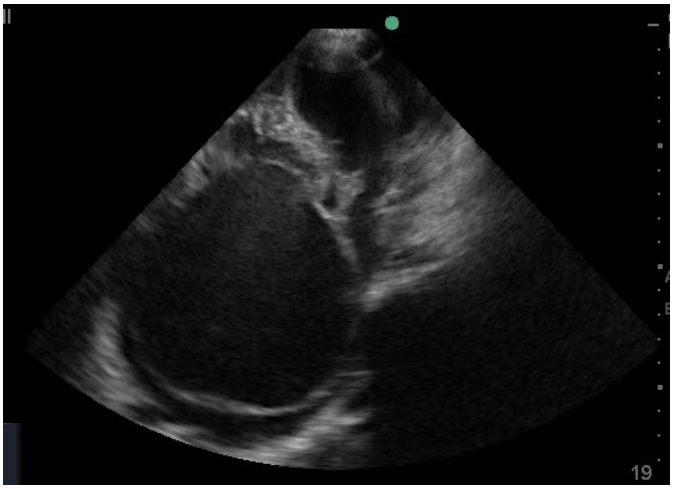
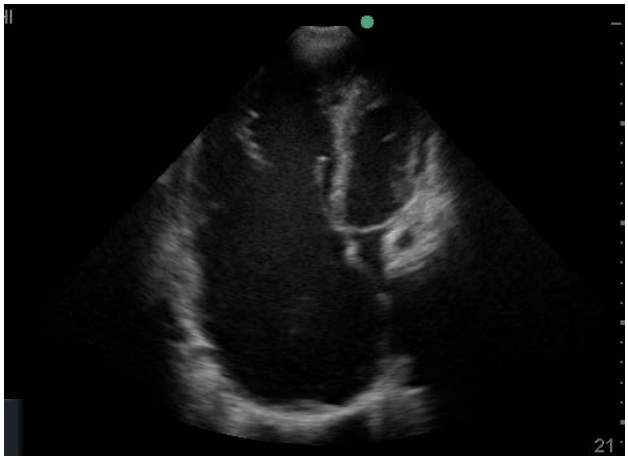


Figure 3. EKG showing atrial fibrillation.



(a)



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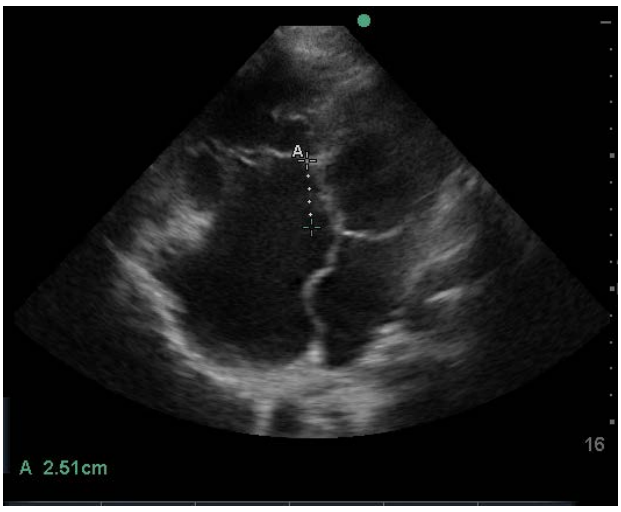

(c)

Figure 4. Echocardiography images showing apical displacement of the tricuspid valve.

3. Result

These were 4 cases of Ebstein's anomaly, 4 boys to one girl, mean age was 13.75 ± 0.96 years.

Clinically, one patient had NYHA II dyspnoea and 3 had NYHA III dyspnoea. Signs of right heart failure were present in 3 patients.

The EKG showed atrial hypertrophy (1 case), right bundle branch block (1 case), WPW (1 case), and atrial fibrillation (1 case).

Frontal chest radiographs showed cardiomegaly in all patients with a mean cardiothoracic ratio of 0.77 ± 0.03. Our 4 patients had clear lungs

Echocardiography showed Ebstein's anomaly in 2 patients in Grade 2 and 2 in Grade 4 (**Table 1**). The mean apical displacement of the tricuspid septal leaflet was 22.5 ± 3.30 mm/m^2 (**Figure 4**).

The Celermajer index was 2 in 2 of our patients and 4 in the other 2 patients. Tricuspid insufficiency was massive in 3 infants and laminar in 1 patient. Associated anomalies were patent foramen ovale in 2 cases, and mitral leak in 2 cases.

Table 1. Clinical, radiographic and echocardiographic parameters.

Patient	Age Year	SPO_2 (%)	Clinique	*CTI	Celermajer Index (Grade)	Tricuspid Septal Valve Offset mm (mm/m^2)	Associated Anomalies	*TI
1. Girl	14	76	NYHA III+ RHF	0.74	Grade 4 (1.60)	23 (18)	*PFO	Massive
2. Boy	15	85	NYHA III+ RHF	0.80	Grade 2 (0.92)	30 (26)	*PFO	Massive
3. Boy	13	96	NYHA I	0.78	Grade 2 (0.79)	25 (23)	Mitral Leak Grade 2	Laminar
4. Boy	13	96	NYHA III+ RHF	0.74	Grade 4 (2)	26 (22)	Mitral Leak Grade 1	Massive

CTI: Cardio-Thoracic Index; **TI**: Tricuspid Insufficiency; **RHF**: Right Heart Failure; **PFO**: PATENT Foramen Ovale.

Medical depletion therapy was instituted in 3 of 4 patients. One patient was on anticoagulant therapy. Two patients were awaiting surgery. We had recorded one case of death in a refractory heart failure setting. In the 2 patients awaiting surgery, the signs of heart failure improved under medical treatment.

4. Discussion

Ebstein's disease is a rare congenital heart disease. It is an isolated condition that remains largely undetected in childhood due to insignificant and benign symptoms [9]. This was the case in our study, the four patients were adolescents.

The natural history of Ebstein's disease is difficult to understand in a simple way because of the wide spectrum of clinical presentation it offers: incidental discovery in the fetus during per-partum ultrasound, cyanosis in the newborn, heart failure in the infant, heart murmur in the child and rhythm disorders in the adolescent and adult [10]. Three out of four of our patients were in heart failure and one patient had a rhythm disorder such as atrial fibrillation.

The electrocardiogram in Ebstein's anomaly is often abnormal. It may show right ventricular hypertrophy, first-degree atrioventricular block, and right bundle branch block [6]. An accessory bundle is present between 6% and 36% [1]. The ECG abnormalities found in our patients are: right atrial hypertrophy, complete right bundle branch block and posterior-right Wolff-Parkinson-White. Julie *et al.* also reported Wolff-Parkinson-White in a 37-year-old adult with Ebstein disease [11].

Cardiomegaly is frequent in Ebstein's anomaly. A cardiothoracic ratio of at least 0.65 is associated with a poor prognosis [12]. This was the case with our four patients.

Echocardiography is an essential diagnostic test and allows accurate assessment of the tricuspid valve leaflets and the size and function of the heart chambers. The main feature of Ebstein's anomaly is an apical displacement of the tricuspid septal leaflet of at least 8 mm/m^2 [1] [13]. The average displacement in our patients was 22.5 ± 3.30 mm/m^2.

The Celermajer index, an echocardiographic severity score for neonates, relates the combined area of the right atrium and atrialized right ventricle to that of the functional right ventricle and left heart (ratio < 0.5, Grade 1; ratio between 0.5 and 0.99; Grade 2 with 92% survival; ratio of 1.0 to 1.49; Grade 3 early mor-

tality at 10%; ratio ≥ 1.5; Grade 4 with early mortality > 50%). Despite an adolescent population in our case, we used this index, two patients had Grade 2 and two were Grade 4 [1] [14]. We recorded one death, it was in Grade 4 with an index of 1.6. Could the Celermajer index be extended beyond the neonatal period?

The associated anomalies we found were mitral leak and patent formamen ovale. In the study by Davied *et al.*, the authors report a case of a child with an Ebstein anomaly associated with tetralogy of Fallot [15].

The management of Ebstein's anomaly varies according to anatomical form and clinical presentation. Patients with minor tricuspid valve displacement are often asymptomatic and do not require special treatment. In these patients, regular clinical and ultrasound monitoring is required. They should be monitored for arrhythmia, right chamber dilatation or right ventricular systolic dysfunction [1]. Surgical intervention is indicated when the patient becomes symptomatic (NYHA III-IV) or when arrhythmia or increased cardiomegaly occur, or when echocardiographic changes such as deterioration of right ventricular function or worsening of the echographic severity index grade occur [16] [17]. It consists of reconstruction with or without an upper cavo-pulmonary bypass or tricuspid valve replacement or the cone technique. Two of our patients have a surgical indication. They are awaiting surgery.

5. Conclusion

This case series shows that Ebstein's disease, a rare congenital heart disease, can be discovered in adolescence. The Celermajer index, an echocardiographic severity parameter for newborns, was used in this study. This series opens the prospect of studies evaluating the Celermajer index in adolescent and adult populations.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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