Research Article

Ebstein's Anomaly And Sudden Death In Adolescence: A Case Report In Barranquilla, Colombia

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Abstract

Introduction: Congenital heart defects, including Ebstein anomaly (EA), are a major cause of morbidity and mortality worldwide. AE manifestations depend on anatomy and may include arrhythmias and right atrial enlargement. AE is diagnosed through echocardiography, and treatment ranges from symptomatic and antiarrhythmic management to surgical repair. **Case Presentation:** A previously healthy 16-year-old adolescent who experienced sudden cardiac arrest recovered after 20 minutes of CPR. He was admitted to the pediatric intensive care unit and placed on a ventilator and vasopressors. His electrocardiogram showed a preexcitation pattern, and an electrophysiological study identified two accessory pathways. He was diagnosed with Ebstein's anomaly, and propafenone was administered to modulate the slow nodal pathway. In a second procedure, the right posterior accessory pathway was ablated, which allowed the antiarrhythmic medication to be discontinued while the antiplatelet therapy continued. The patient had a satisfactory outcome. **Discussion:** Early diagnosis and treatment impact the survival of patients with EA, even when the patient's medical history is unknown. EA is a rare congenital heart disease that affects one in 20,000 live births. EA is associated with arrhythmias in 22-42% of cases, 5-10% of which are associated with Wolff-Parkinson-White syndrome. Up to 25% of patients have accessory pathways. **Conclusion:** Similar cases with a fatal outcome are rare, which makes this clinical case exceptional. Given the appropriate clinical response, it is important to understand that EC can go unnoticed throughout life.

Keywords: Ebstein's anomaly, congenital heart disease, sudden death, tricuspid valve, Wolff-Parkinson-White syndrome, ablation.

1. Introduction

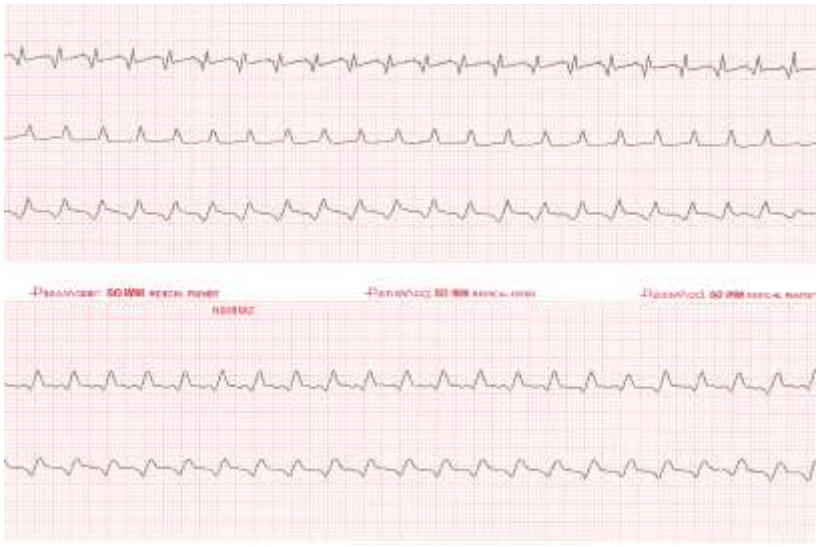
Congenital heart defects (CHDs) are malformations of the heart present at birth or before that. They account for a significant disease burden and mortality worldwide (1). Ebstein's anomaly (EA) is a rare congenital heart disease characterized by the displacement of the septal and posterior leaflets of the tricuspid valve to the apex, which causes the right ventricle to become atrialized. Clinical manifestations vary according to the severity of the anatomical defect, pulmonary flow, and the presence of atrial defects or arrhythmias (2). Common electrocardiographic findings include right atrial enlargement, right atrioventricular and bundle branch blocks, and Wolff-Parkinson–White syndrome (WPW syndrome) (3). Sudden death is frequently associated with atrial fibrillation. Diagnosis is based on echocardiography to assess the lesion and its severity (4). Treatment options range from symptomatic management to valve repair surgery and include antiarrhythmic medication, an implantable cardioverter-defibrillator, or a ventricular assistance device in advanced cases (5).

Case Presentation

The patient was a previously healthy 16-year-old female adolescent with no known medical history. She presented with a clinical picture that began with a headache, palpitations, and chest pain, followed by loss of consciousness and central cyanosis. She was admitted to the emergency department, where cardiopulmonary resuscitation (CPR) was performed for 20 minutes due to cardiac arrest, initially presenting with asystole and later presenting with ventricular tachycardia. She required support with mechanical ventilation, sedation, and vasopressors with norepinephrine.

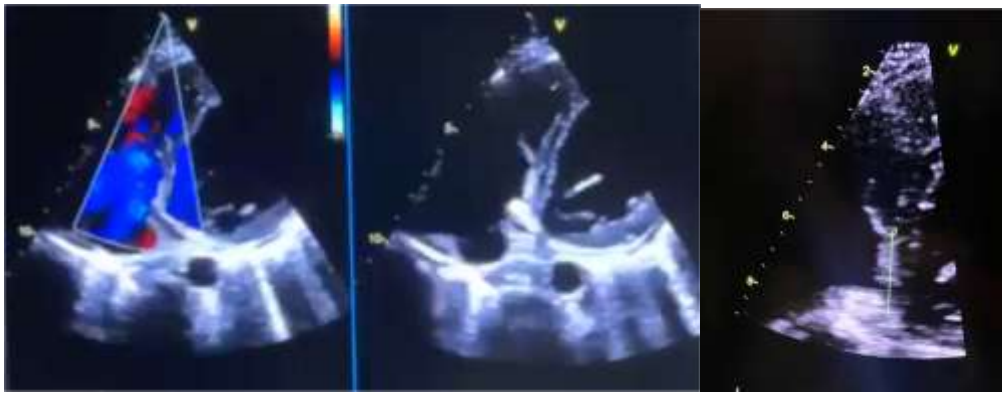
Upon admission to the intensive care unit (ICU), paraclinical tests were performed, revealing multisystem mild-to-moderate hypoxic damage (lactic acid level of 3.21 mmol/L, elevated cardiac enzymes [CK, CK-MB, and Troponin T], and elevated transaminases), an intense inflammatory response (leukocytosis with neutrophilia), and preserved renal function. The baseline electrocardiogram (Figure 1) shows an apparent pattern of preexcitation, evidenced by a short PR interval and a delta wave. The echocardiogram revealed mild Ebstein anomaly with mild valve regurgitation, associated with mitral valve prolapse and trivial insufficiency, as well as preserved biventricular systolic function (Figure 2).

Figure 1



An electrophysiological study was performed, showing two accessory pathways. Modulation of the slow nodal pathway was achieved; however, modulation of the right posterolateral accessory pathway was unsuccessful. During the procedure, the patient experienced cardiac arrest with asystole, requiring external cardiac massage and a single dose of adrenaline. The patient recovered from the arrest rhythm within a minute.

Figure 2:



The diagnosis of Ebstein's anomaly was confirmed, and propafenone treatment was indicated. In a second study, abolition of the right posterior accessory pathway was achieved. Therefore, antiarrhythmic therapy was suspended, and antiplatelet therapy was indicated for six weeks, with satisfactory results.

Discussion

This cardiac malformation is characterized by the following: (1) fixation of the septal and posterior leaflets to the underlying myocardium, (2) apical displacement of the tricuspid valve plane, (3) right ventricular dilation with varying

degrees of hypertrophy and wall thinning, (4) abnormal elongation and fenestration of the anterior tricuspid leaflet, and (5) dilation of the tricuspid annulus plane. (3)

Ebstein's anomaly (AE) was first described in 1866 by Wilhelm Ebstein based on autopsy findings. Subsequently, Helen Taussig provided the first clinical description in 1947, and the first diagnosis in a living patient was made in 1949 (6).

AE has an incidence of approximately 1.5 cases per 200,000 births and accounts for less than 1% of all congenital heart defects. Only about 5% of affected individuals reach adulthood, and their mortality rate is up to 20 times higher. Approximately 80% of cases are associated with cardiac comorbidities, such as atrial septal defects (ASDs) and cardiac arrhythmias, which occur in 22%-42% of cases. Of this arrhythmia, 5%-10% are associated with Wolff-Parkinson-White (WPW) syndrome. Up to 25% of patients have accessory pathways, and up to 25% have obstruction of the right ventricular outflow tract (RVOT).

In AE, there is malformation of the tricuspid valve and the right ventricle. The septal and posterior leaflets are displaced toward the apex and may form an imperforate membrane or a muscular crest that separates the entrance and trabecular portions of the RV. The anterior leaflet is usually redundant with fenestrations and short, poorly formed tendon cords. It can move toward the RV outflow tract and cause obstruction. The RV is divided into three parts: the input portion, which is directly affected by the abnormality and is also called the atrialized portion; and the trabecular and outlet portions, which make up the functional RV. The atrialized portion usually exhibits disproportionate dilation, which can compromise the entry area, the functional apex, and the RV outflow tract (8).

The clinical manifestations of the Ebstein anomaly depend on the magnitude of the anatomical defect, pulmonary blood flow, and the presence of other cardiac comorbidities. Neonates may exhibit symptoms such as cyanosis due to right-to-left shunts, while patients who reach adulthood may experience progressive cyanosis, exercise intolerance, right heart failure, and arrhythmia.

ECG usually shows right bundle branch block, delta waves, high P waves, first-degree atrioventricular (AV) block, and supraventricular tachyarrhythmias, mainly by atrioventricular reentry, in Ebstein anomaly (10). An echocardiogram confirms the diagnosis by showing apical displacement of the septal leaflet (>8 mm/m²), valvular dysplasia, dilation of the right chambers, paradoxical septal movement, and tricuspid regurgitation (2). MRI helps confirm echocardiogram findings and is the cornerstone of diagnosis (4).

Type A	DV volume is adequate
Type B	Large atrialized component of the right ventricle, but the anterior leaflet of the tricuspid valve moves freely
Type C	Anterior leaflet severely restricted in movement and may cause significant obstruction of the RV outflow tract
Type D	Almost complete RV atrialization, except for a small infundibular component

Table 1. Prepared by authors

The therapeutic approach is nonspecific and includes everything from symptomatic measures to surgical correction of the valve. Management often consists of heart failure drugs and/or anticoagulant therapy. Patients with Ebstein's anomaly are prone to developing thromboembolic phenomena. Treatment for arrhythmia patients, especially those with supraventricular arrhythmia, varies. They may receive medications such as beta-blockers or calcium channel blockers. If these are ineffective, class I or class III antiarrhythmics may be used to treat paroxysmal atrial fibrillation. However, surgical or radiofrequency ablation is recommended for symptomatic patients.

Conclusion

There are very few documented cases with a comparable clinical presentation, many of which had fatal outcomes. This highlights the exceptional nature of this report. The patient's favorable evolution and adequate response to treatment are particularly relevant considering Ebstein's anomaly may remain asymptomatic or go unnoticed for years, even until adolescence or adulthood. This case underscores the importance of high clinical suspicion and prompt treatment.

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