An Adult Presentation of Ebstein's Anomaly of Tricuspid Valve: Case Report and Review of Literature.

Introduction:

Ebstein's anomaly is a rare congenital heart condition characterized by a combination of tricuspid valve malformation with right ventricular myopathy [1]. It is frequently associated with left-sided heart anomalies[2] and excitability disorders. It accounts for less than 1% of all cases of congenital heart disease. The main pathophysiological mechanism is a failure of delamination during embryogenesis, leading to downward displacement of the functional annulus and division of the right ventricle into a functional and an atrialized ventricle. Its evaluation requires a series of tests for anatomical and clinical classification, assessing disease severity, and ultimately guiding treatment. Prognosis and management depend on the size of the functional right ventricle, tricuspid valve anatomy, and left ventricular function.

Case presentation:

We present a clinical case of a 30-year-old female with a recent medical history of hypertension on diuretic medication. The patient was admitted for scheduled hospitalization for the management of severe palpitations, reporting three episodes of dizziness without syncope. At her admission to our service, the patient was stable; slight cyanosis of the lips was noted, with peripheral oximetry around 89%, blood pressure at 117/74 mmHg, and heart rate at 140 bpm. The cardiovascular examination revealed a regular rhythm with no additional sounds or murmurs. The electrocardiogram displayed a regular sinus rhythm with a left deviation of the heart axis and a positive delta wave in the QRS complex.

On a biological level, we note polycythemia with hemoglobin at 17 g/dl without any other biological abnormalities.

The transthoracic echocardiography (TTE) and transesophageal echocardiography (TOE) showed a severe type C Ebstein's anomaly with major predominance of an atrialized right ventricle and a small functional right ventricle. A dilated, non-obstructive right ventricular outflow tract (RVOT) was also noted. A restrictive ventricular septal defect (VSD) and a small atrial septal defect (ASD) of less than 5 mm were found with minimal tricuspid regurgitation (Figure *1A*, *1B*, *1C*)

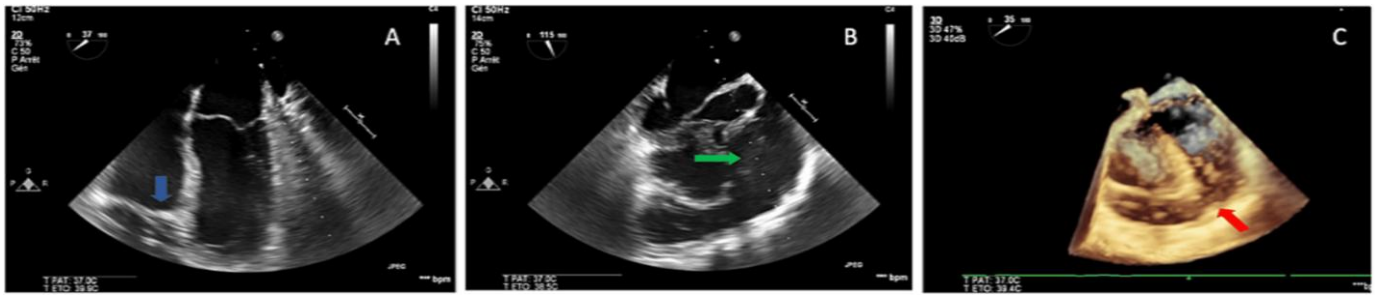

FIGURE 1: Figure 1: (1A) 2D Transoesophageal Echocardiography (TOE) image showing displacement of the septal leaflet (blue arrow) towards the apical right ventricle, (1B) 2D TOE image showing dilated right ventricular outflow (green arrow) , (1C) 3D TOE image showing tunnel- shaped apical portion of the right ventricle (red arrow)

The right heart catheterization demonstrated quite normal pulmonary artery pressures and a low end- diastolic left ventricular pressure. It also noted a high right atrial pressure at 15 mmHg, systemic venous desaturation at 85% (Table *1*), a low cardiac index at 1.5 l/min/m², and normal pulmonary vascular resistance (PVR) (Table *2*).

		Pulmonary Artery				LV			Aorta		
	Right Atrium	Systolic	Diastolic	Middle	**Capillary**	Systolic	Diastolic	Middle	Systolic	Diastolic	Middle
Pressures (mmHg)	15	29	14	19	15	140	3	14	143	76	95
Saturations (%)	59%	72%				85%			85%		

Table 1 : Pressures and saturations measured during right heart catheterization

Flow	**Systemic flow (L/mn)**	**Pulmonary Flow (L/mn)**	**Qp/Qs**	**Cardiac Index (l/mn/m^2)**
	2,78	3,1	1,16	1,56
Resistance	Systemic Vascular Resistance (SVR) (UW)	Pulmonary Vascular Resistance (PVR) (UW)	PVR/SVR	
	29,13	1,29	0,04	

TABLE 2: Flow and resistances measured during right heart catheterization

During the examination, several episodes of supraventricular and ventricular tachycardias, such as atrial and ventricular tachycardia occurred. The angiography injections revealed a significantly enlarged atrialized right ventricle with a dilated pulmonary artery (Figure *2*).

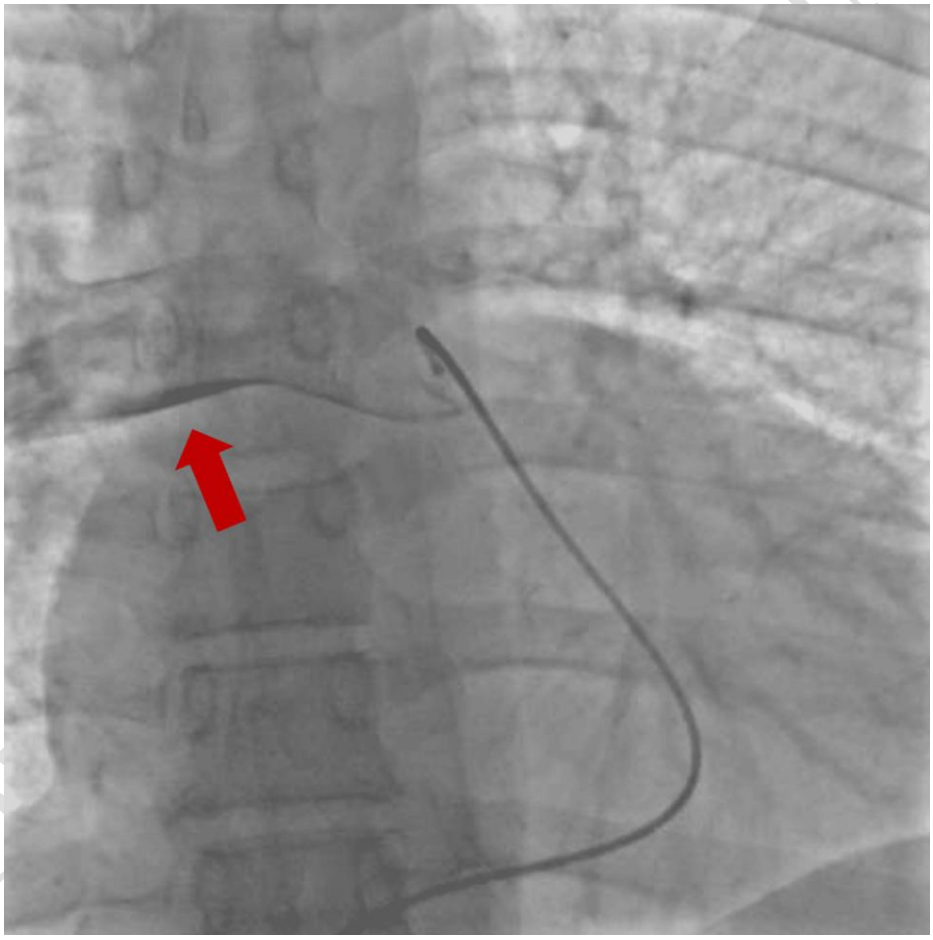

FIGURE 2: Injection for angiography in anteroposterior view showing a dilated pulmonary artery (red arrow).

The final decision of the heart team was to perform an electrophysiological study due to the severe episodes of arrhythmias documented on the 24-hour electrocardiogram. Our patient then underwent tricuspid valve repair with a cone reconstruction using the Da Silva technique. Upon coming off bypass and due to elevated right atrial filling pressures, a partial cavopulmonary diversion was performed due to the favorable anatomy of the pulmonary vasculature.

Discussion:

The Ebstein anomaly occurs during the phase of heart development between the 7th and 12th week of intrauterine life [3]. It results from incomplete tricuspid valve delamination from the right endo- myocardium, leading to adhesion of the septal and posterior leaflets to the myocardium with fenestration [4] and tethering of the anterior leaflet [5]. A lack of coaptation of the tricuspid leaflets is described, causing tricuspid regurgitation that increases with annular dilatation [6]. Also, displacement of the tricuspid annulus is reported [7], resulting in splitting of the right ventricle into two portions: the "atrialized" right ventricle (aRV), which acts as a passive tank during atrial contraction, and the "functional" right ventricle (fRV) , which can become dysfunctional [8] due to severe tricuspid regurgitation.

Some pathologies are commonly associated with Ebstein's anomaly; the most frequent are ASD or Patent Foramen Ovale (PFO) [9], VSD, or pulmonary stenosis or atresia [10].

Ebstein's anomaly can present at any age, but a significant number of patients remain asymptomatic and present in adulthood . The most frequent manifestations among adult patients are cyanosis, right heart failure, arrhythmias, and sudden cardiac arrest [11]. Cyanosis is usually subtle and is due to right-to-left shunting across the atrial defect. Palpitations result from tachyarrhythmias and are the first manifestation among adult patients, whereas sudden cardiac arrest is caused by ventricular arrhythmias [12].

Cardiac auscultation shows a triple or quadruple rhythm with a loud first heart sound resulting from anterior leaflet closure; a soft holosystolic murmur of tricuspid regurgitation is sometimes audible [13]. Electrocardiogram findings typically include right bundle branch block (RBBB) associated with right atrial enlargement represented by tall P waves called "Himalayan P waves." Frequently, excitability disorders are found, such as Delta waves due to an accessory pathway or first-degree atrioventricular block [14], which are integral to Ebstein's disease.

In echocardiography, Ebstein's anomaly is defined as displacement of the tricuspid leaflet greater than 8 mm/m2 from the insertion of the anterior mitral leaflet [15]. Color doppler helps assess the degree and site of tricuspid regurgitation. Tethering of the tricuspid valve can also be present [16]. Assessment of both the aRV and the fRV size is essential, especially the RVOT size. It should be noted that the smaller the size of the fRV, the worse the prognosis of the disease. Other important elements to evaluate are left ventricular function and the search for other anomalies often associated with Ebstein's disease. TOE is more efficient in adults in cases of poor echogenicity; intracardiac shunts are better visualized.

Several classification systems exist for Ebstein anomaly, with the Carpentier classification [17] (Figure *3*) being the most commonly used, focusing on anatomical categorization. Its initial assessment relies on echocardiography, where anomalies are graded as mild, moderate, or severe based on the degree of leaflet displacement, tethering, and right ventricular dilation (Table *3*)

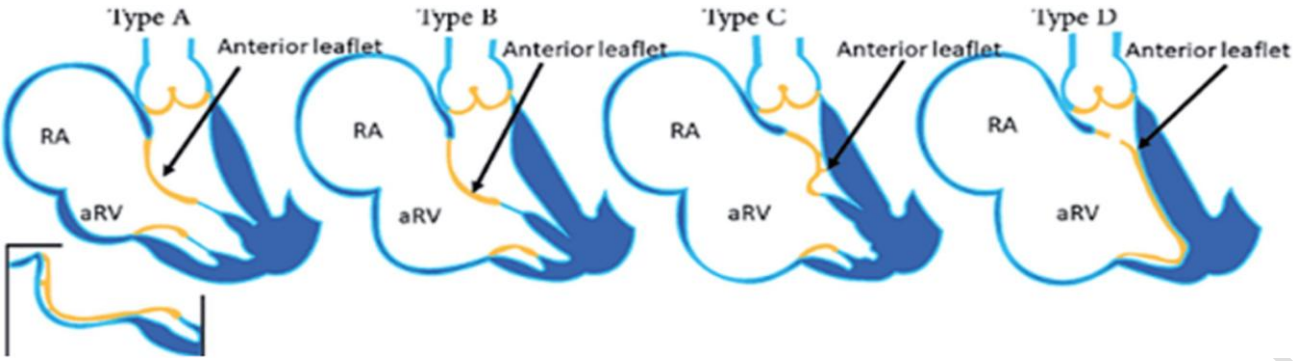

FIGURE 3: Carpentier's Classification of Ebstein's Anomaly

Carpentier's classification of Ebstein's anomaly based on the anatomical description of the anomaly [18].

	Type A : Mild	Type B : Moderate	Type C : Severe	Type D :Extremely severe
Anterior leaflet (attachment - mobility)	Normal morphology	Abnormal attachments	Partial adhesions to the venticular free wall	Severe adhesions to the ventricular wall
	Normal mobility	Normal mobility	Restricted motion	-
'Atrialized' ventricle (size - function)	Small ventricle	Moderate-sized ventricle	Large ventricle	Very large ventricle
	Preserved function	Reduced function	Reduced function	Minimal function
Functional ventricle (size- function)	Normal-sized ventricle	Reduced ventricle	Small ventricle	Infundibulum
	Preserved function	Preserved function	Reduced function	Depressed function

TABLE 3: Description of the Carpentier Classification of Ebstein's Anomaly

Ebstein's classification into 4 types based on the attachments of the tricuspid valve, the size and function of the atrialized and functional right ventricle[18].

Cardiac magnetic resonance imaging (CMR) allows for a detailed analysis of tricuspid valve anatomy and accurate assessment of right ventricular function, but it is not used as a first-line approach.

Right heart catheterization guides surgical treatment. Left ventricular function, anatomy of the pulmonary artery tree, and pulmonary pressures are important parameters for palliative surgery.

If no surgical intervention can be proposed, medical treatment should be optimal, with effective management of heart failure. In cases of significant thromboembolic risk, oral anticoagulation may be considered. Management of arrhythmias can use medical treatment if symptomatic. Assessment of arrhythmia substrates by catheter-based intervention [19] or even catheter ablation can sometimes be considered before surgery.

The principle of surgical treatment is either biventricular repair , a single ventricle pathway with right ventricular exclusion [20], or heart transplantation. Biventricular repair involves reconstruction of the tricuspid valve, sometimes with repair of the ASD. Single ventricle repair is recommended for patients with inadequate right ventricle size and is considered a palliative procedure. Cardiac transplantation is reserved for severe forms with significant left ventricular dysfunction.

For surgical treatment in this case, tricuspid valve repair was the first-line approach considered, with a Cone repair using the Da Silva technique. These method involve some specific steps (Figure *4*)

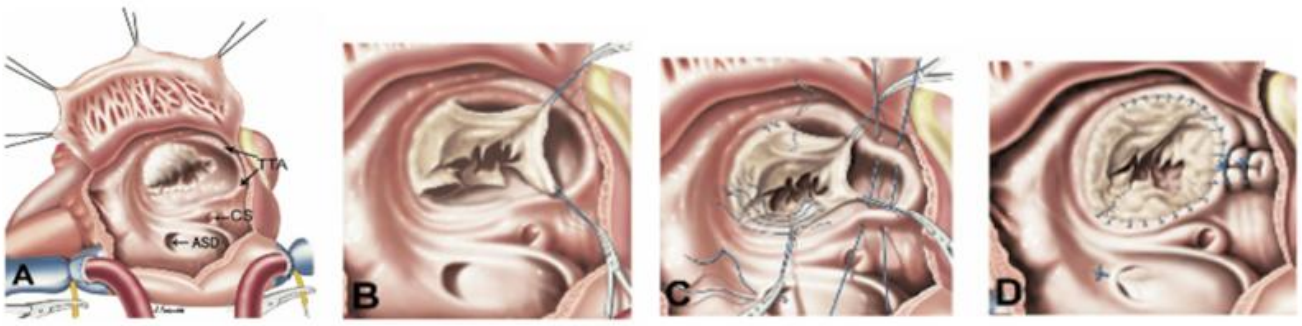

FIGURE 4: Cone reconstruction of tricuspid valve in Ebstein anomaly : The Da Silva Technique

Steps of Da Silva Technique : [21]

A. The right atrium is opened, revealing displacement of the tricuspid valve.

TTA: true tricuspid annulus; ASD: atrial septal defect; CS: coronary sinus.

B. A portion of the anterior and posterior leaflet is detached, creating a single unit.

C. The posterior leaflet edge is rotated clockwise and sutured to the septal edge of the anterior leaflet. The true tricuspid annulus is then plicated.

D. The valve is completely reattached to the true tricuspid annulus, and the atrial septal defect is closed with a valve.

In this particular case, due to advanced age and tricuspid annular dilation, augmentation with a prosthetic ring could have been necessary if valve competence was inadequate.

Upon separation from cardiopulmonary bypass (CPB), the right atrial filling pressures will be monitored. Elevated pressures may necessitate a partial cavopulmonary diversion, achieved by suturing the superior vena cava directly to the right pulmonary artery. In our case this technique was performed given the favorable hemodynamics for potential cavopulmonary diversion.

Prognosis depends on the degree of tricuspid displacement, severity of tricuspid regurgitation, size of the fRV, and degree of right ventricular dysfunction .

Conclusion:

Ebstein's disease is a rare congenital heart condition typically characterized by tachyarrhythmia. Echocardiography serves as the primary diagnostic tool, confirming the condition and aiding in severity assessment. Evaluation necessitates meticulous examination of the size of the atrialized and functional right ventricles, along with an assessment of left ventricular function, in order to guide surgical management.

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