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Ebstein's Anomaly Surpassing 6th Decade of Life- A Case Report

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INTRODUCTION

Ebstein's anomaly (EA) is a rare congenital heart defect usually presenting around birth years, where in the septal and posterior leaflets of the tricuspid valve are displaced towards the apex of the right ventricle, atrializing the right ventricle. Although it accounts for only about 0.5% of congenital heart defects, only 5% of patients are known to survive beyond the fifth decade of life. [1] We present the case report of a patient with Ebstein's anomaly surpassing the sixth decade of life highlighting the importance of regular cardiac surveillance enabling increased lifespan of patients with the disease.

CASE REPORT

We present the case of a 61-year-old female with a past medical history of essential hypertension (HTN), atrial septal defect (ASD) diagnosed at birth, status post closure with a cardioSEAL with a small residual defect, sever tricuspid regurgitation, and Ebstein's anomaly diagnosed at 3 weeks of age who presented to an outpatient cardiology clinic to establish continuity of care of her cardiac conditions. She endorsed having shortness of breath on exertion and having exercise limitations ever since childhood. She denied any active complaints of chest pain, orthopnea, paroxysomal nocturnal dyspnea, swelling in her feet, syncope, jaw pain or palpitations. She was on verapamil 80mg and digoxin 0.25mg to treat her HTN. She had a family medical history of ischemic heart disease in her father. She denied any alcohol or tobacco use. She was married and had never been pregnant. She regularly followed up with her dentist and had no dental issues. She underwent cardiac catherization at the age of 57 and it showed normal left and right sided filling pressures at rest, no significant shunting, severe TR with severe RA dilatation and residual left-to-right shunting across the interatrial septum (prior atrial closure) seen with color doppler and agitated saline. Cardiac MR demonstrated severe Ebstein's anomaly of the tricuspid valve with rudimentary septal leaflet, LVEF of 65%, severely dilated right atrium. She was referred to cardiothoracic surgery for surgical correction of the Tricuspid valve, but she did not follow up as she was not symptomatic and did not want any surgical interventions at that time. At her most recent cardiology visit, her vitals were stable with a blood pressure of 127/79mmHg sitting, HR 66bpm, SPO2 98% on RA. Her physical examination was negative for any significant findings except for cardiovascular system, on auscultation there was fixed splitting of the second heart sound (S2) and a 2/6 grade systolic ejection murmur.12-lead ECG showed normal sinus rhythm and evidence of right and left atrial enlargement. A transthoracic echocardiogram was ordered as a routine surveillance. Patient continues to be asymptomatic and would be followed up on a yearly basis by a cardiologist.

DISCUSSION

Ebstein anomaly is an uncommonly found congenital cardiac abnormality involving the tricuspid valve and the right ventricle (RV) [2]. It comprises < 1% of congenital heart defects.[3] The anomaly was historically described by the pathologist Wilhelm Ebstein in 1866 and was later named in his honor. After performing an autopsy on a 19-year-old cyanotic male with exertional dyspnea and palpitations who succumbed to a sudden cardiac arrest, the disease etiopathology was established.[4] Ebstein anomaly is defined by five salient features. It includes the tricuspid valve being apically displaced, septal and posterior tricuspid leaflets being adhered with the myocardium, tricuspid annulus being apically displaced and dilated, atrialized portion of the right ventricle being dilated, and anterior tricuspid leaflet being fenestrated, redundant and tethered. [3][5]

Pathophysiological abnormality of Ebstein anomaly includes the failure of delamination of the tricuspid valve leaflets from the interventricular septum in utero.[6]

The posterior and septal leaflets are mainly affected by the apical displacement, leading to the tricuspid annulus being apically displaced and the tricuspid orifice being antero apically displaced.[7][8]

Clinical presentation varies depending on the age of presentation. In older adults, like in the presented case, palpitations due to tachyarrythmias are a common presenting symptom. Symptoms of exertional dyspnea are also frequently noted. Dyspnea can occur as a result of right-to-left shunting with resulting hypoxemia. Chronic fatigue and lower extremity edema are well known clinical symptoms in heart failure patients. Physical examination in adults with Ebstein anomaly demonstrates the murmur of tricuspid regurgitation. In grave cases, a hyperdynamic precordium and a thrill on the left lower sternal border may be present. In the presence of an atrial septal defect or right bundle branch block, the second heart sound may be widely split, as was in the case described above.

Evaluation includes performing an ECG, which could demonstrate right bundle branch block, tall P waves due to right atrial enlargement, first degree AV block, supraventricular tachyarrythmias. Chest XRay may demonstrate cardiomegaly and right atrial enlargement. Echocardiography is the imaging modality for diagnosing Ebstein's anomaly. Ebstein's anomaly is diagnosed by echocardiography when the septal leaflets of the tricuspid valve are displaced apically, greater than 8 mm/m2 in the apical 4-chamber view. There could be right atrial dilatation and interventricular septal flattening due to right ventricular volume overload. Tricuspid regurgitation can also be detected with color doppler. The severity of tricuspid regurgitation can be determined by echocardiography. Valuable information regarding tricuspid leaflet anatomy can be obtained from cardiac magnetic resonance imaging (cMRI). It permits quantification of the RV ejection fraction and is hence superior to echocardiography in assessing RV function. For periprocedural imaging of coronary anatomy before surgical intervention on the tricuspid valve, cardiac CT is used.

Management for adult patients depends on the severity of TR and symptoms. Symptoms of heart failure are managed with loop diuretics and guideline-directed medical therapy. Beta blockers or calcium channel blockers can be used in patients with supraventricular tachyarrhythmias. Class I antiarrhythmic procainamide or class III antiarrhythmic amiodarone or sotalol can be used to treat paroxysmal atrial fibrillation in patients with ineffective control.Catheter ablation is required frequently in patients with intractable arrhythmias.

There are several indications for surgery in adult patients.Heart failure symptoms (New York Heart Association functional class III or IV), progressive exercise intolerance, evidence of RV dysfunction or progressive RV dilatation, evidence of paradoxical emboli, arrhythmias refractory to medical and catheter ablation therapies, cyanosis with oxygen saturations < 90%, severe tricuspid regurgitation, significant concomitant lesions such as pulmonic stenosis or atrial or ventricular septal defects, progressive cardiomegaly , cardiothoracic ratio > 65% and reduced left ventricular function are indications for surgical intervention. [9] Tricuspid valve repair and patch closure of the atrial septal defect comprise surgical closure. The valve is replaced in cases the native tricuspid valve cannot be repaired. A secundum atrial septal defect amenable to transcatheter device closure could be present in these patients. Tachyarrhythmias that can be treated in the cardiac catheterization laboratory could also be present. [10]

The degree of displacement of the tricuspid valve, amount of tricuspid regurgitation, and duration and degree of RV dysfunction are the main predictors of mortality.

CONCLUSION

It is an unusual case as the patient continues to remain majorly asymptomatic even after the sixth decade of life and is being managed closely by medical and regular cardiac surveillance. The case sheds light on the importance of regular cardiac surveillance of patients with Ebstein's anomaly to improve survival outcomes.

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