CASE REPORT

# Atrialised right ventricular myxoma in a patient with Ebstein's anomaly

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## Summary

Ebstein's anomaly is a rare entity affecting around 1 in 200,000 live births and accounts for less than 1% of congenital heart diseases. Ebstein's anomaly with an associated right-sided myxoma is extremely rare, with only one other case report found in the literature. Previous reports have also noted cases of Ebstein's anomaly associated with left-sided myxomas. We describe a female patient with, to our knowledge, the first case of a histopathologically confirmed right ventricular myxoma in the setting of Ebstein's anomaly.

**Key Words**

- atrial mass
- Ebstein's anomaly
- myxoma

## Learning points:

- Tall p waves (Himalayan) are associated with Ebstein's anomaly.
- Large a-waves on clinical evaluation are typical of tricuspid valve obstruction or restrictive right ventricular filling.
- The presence of a stalk is suggestive of a myxoma.
- Before undertaking surgery, careful evaluation of involved structures including valvular morphology and function is essential.

## Background

Ebstein's anomaly is an extremely rare finding and accounts for a small minority of cases of congenital heart disease (1). Previous literature describes isolated cases of Ebstein's anomaly associated with both left and right-sided atrial myxomas (2,3). We describe a female patient with, to our knowledge, the first case of a histopathologically confirmed right ventricular myxoma in the setting of Ebstein's anomaly.

This case is important as it:

- Illustrates an extremely rare finding of a ventricular myxoma in the setting of an Ebstein's anomaly.
- Highlights the value of basic principles of examination, ECG interpretation and echocardiography in making the correct diagnosis.
- Demonstrates the surgical approach to a rare combination of pathologies.

## Case presentation

A 40-year-old female was referred to the cardiology unit with a long-standing history (6 years) of infrequent palpitations, which had never been investigated. She now presented with a 4-month history of a progressively worsening dyspnoea (New York Heart Association Class IIa) with new-onset swelling of her legs. The patient volunteered a history that was suggestive of orthostatic hypotension with significant episodes of dizziness without syncope.

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Physical examination revealed a normotensive patient, with a regular pulse. Although no peripheral oedema could be detected (as volunteered by the patient), an elevated jugular venous pressure (JVP) with prominent a- and v-waves in the neck was noticed. Auscultation revealed normal heart sounds with a soft pansystolic murmur, suggestive of tricuspid regurgitation, at the left sternal border. No diastolic murmur was heard.

## Investigation

Electrocardiogram recording revealed sinus rhythm with large/'Himalayan' p-waves and right axis deviation (Fig. 1).

Transthoracic echocardiography revealed a severely dilated right atrium, dilated right ventricle with atrialisation of the right ventricle and apical displacement of the septal leaflet of the tricuspid valve suggestive of Ebstein's anomaly (Figs 2 and 3). A pedunculated mass was noted in the right atrium, with attachment via a stalk to the interventricular septum (Video 1). Significant tricuspid obstruction was demonstrated with continuous wave Doppler (Fig. 4) during diastole. The mass was visualized moving in and out of the tricuspid valve orifice (Video 2).

**Video 1**
Apical 4 chamber view demonstrating apical displacement of the septal leaflet of the tricuspid valve. The stalk of the mass is attached to the Interventricular septum. View Video 1 at http://movie-usa.glencoesoftware.com/video/10.1530/ERP-17-0065/video-1.

**Video 2**
Parasternal short axis view of the tricuspid valve demonstrating the movement of the myxoma. View Video 2 at http://movie-usa.glencoesoftware.com/video/10.1530/ERP-17-0065/video-2.

## Treatment and outcome

During surgery, the right atrium was opened and the tumour was identified in the tricuspid valve with the stalk of the tumour attached to the interventricular septum close to the annulus. The tumour was successfully excised (Fig. 5) and measured 4×4 cm. The tumour base/stalk was calcified at its attachment to the interventricular septum. Care was taken to remove all calcium with a pituitary rongeur.

The defect in the septal leaflet was closed with a glutaraldehyde-treated autologous pericardial patch. The valve was tested with normal saline and incompetence was noted in the commissure between

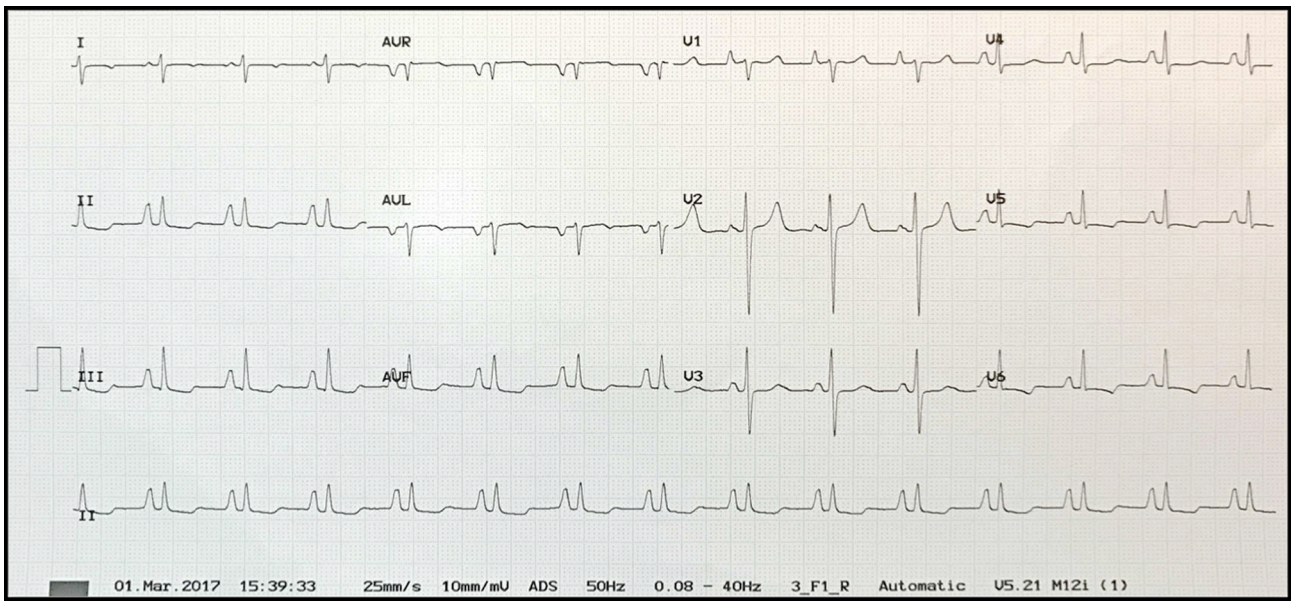


**Figure 1**
Electrocardiogram with tall p-waves.

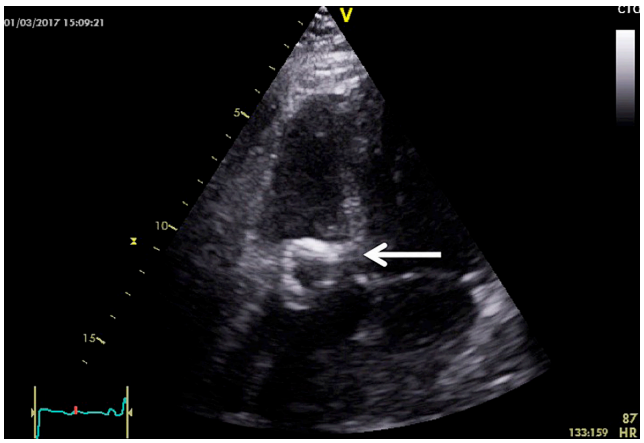


**Figure 2**
Apical 4-chamber view with mass visible (arrow).

the anterior and posterior leaflet. This commissure between the anterior and posterior leaflet was closed with 4-0 Tycron sutures and a tricuspid annuloplasty was done with a 30 mm Medtronic 3D contour ring. The valve was competent with saline testing. The peri-operative transoesophagial echocardiogram confirmed a successful repair.

Post-operative recovery was uneventful with complete resolution of patient's symptoms of palpitations, dyspnoea and dizziness.

Histology confirmed the mass as a myxoma.

Anatomically this is a case of a right ventricular myxoma as the stalk was attached to the interventricular septum, but due to apical displacement of the septal leaflet of the tricuspid valve, the mass was located on the atrial side causing significant obstruction during diastole.

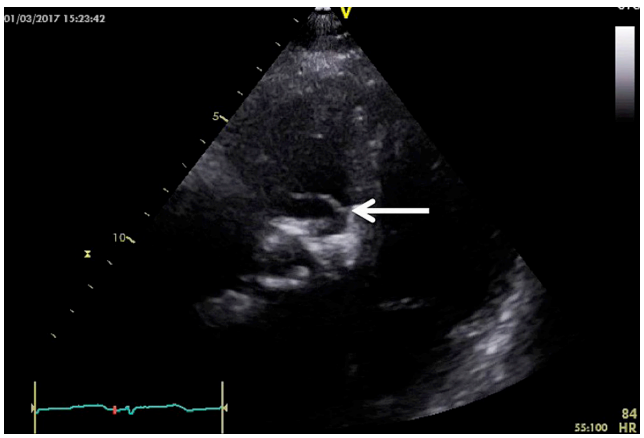


**Figure 3**
Apical 4-chamber view demonstrating apical displacement of septal leaflet (arrow) of tricuspid valve.

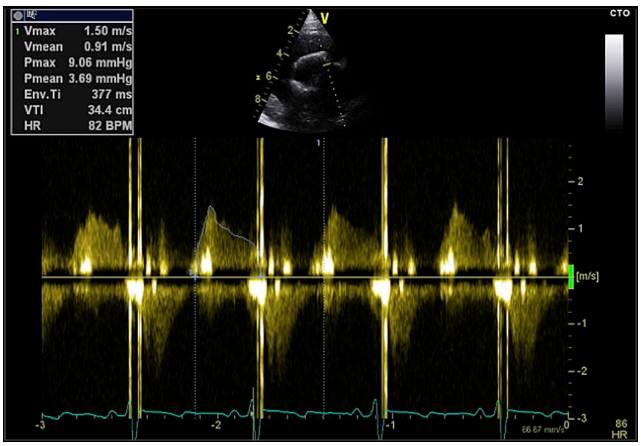


**Figure 4**
Continuous wave Doppler (modified parasternal RV inflow view) across tricuspid valve orifice demonstrating increased velocities.

## Discussion

Atrial myxomas are the most common primary heart tumours – accounting for 40–50% of primary cardiac tumours. The majority of cases are left sided with only 15–25% of cases occurring on the right side. Most cases are sporadic in nature, but up to 10% are inherited in an autosomal dominant fashion within families (4).

Although typically a benign tumour, complications do occur – mainly due to mechanical obstruction caused by the myxoma or embolic phenomena (due to the

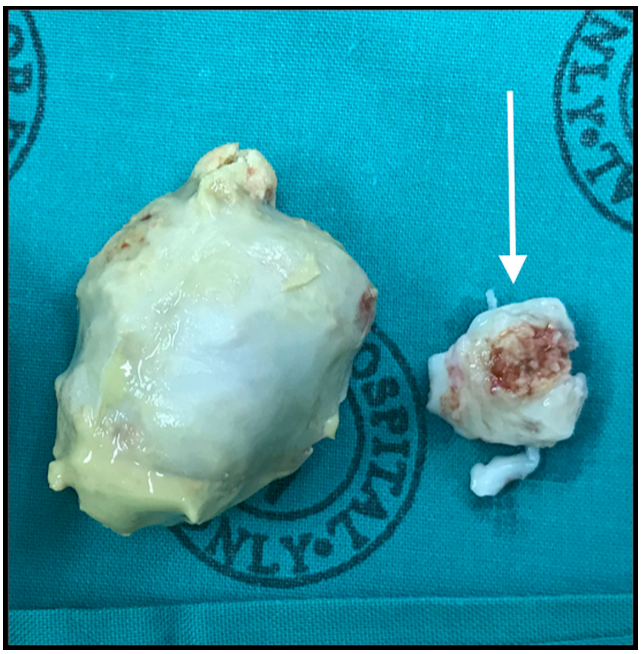

**Figure 5**
Mass with stalk (arrow) after excision.

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friable nature of myxomas) (5). Of note, embolization and prolapse of the myxoma through valves are more common in polypoid tumours compared to round/oval myxomas (6).

Symptoms are varied and depend on multiple factors including the size and location of the myxoma. Myxomas can range from being totally asymptomatic to resulting in sudden cardiac death related to arrhythmias or valve obstruction from mobile, pedunculated myxomas (7). Symptoms, in the absence of intracardiac shunts, are related to the side in which the myxoma originates. Left-sided myxomas typically present with thromboembolic events (typically cerebrovascular accidents) or left heart failure due to obstruction of left ventricular inflow. In right-sided myxomas, the most common symptoms are due to obstruction of right ventricle inflow and typically present with right heart failure as was present in our patient. Dizziness and syncope may be experienced in up to 20% of patients due to prolapse of the tumour into valve orifices causing a significant decrease in preload and subsequent decrease in stroke volume (4). Right-sided myxomas may embolise resulting in pulmonary infarction. On rare occasions, myxomas may become infected resulting in infective endocarditis (8).

Although a definitive diagnosis requires histological confirmation, one can suspect the diagnosis with a high degree of certainty based on history, clinical examination and echocardiographic findings. Laboratory studies are nonspecific and often non-contributory. Trans-oesophageal echocardiography is more sensitive than transthoracic echocardiography for detecting smaller myxomas that are either asymptomatic or present with embolic events. The demonstration of a stalk during echocardiography coupled with mobility, as in our case, favours a diagnosis of a myxoma (9).

Surgical resection is the treatment of choice. This is usually performed via a median sternotomy with wide resection of the attachment to reduce recurrence. Damaged valves may require repair during the procedure. Recurrence rates are low ranging from 1 to 5% (4).

Ebstein's anomaly is characterized by apical displacement of the tricuspid valve leaflets resulting in atrialisation of the right ventricle. Symptoms depend on the associated haemodynamic and conduction system disease associated with the underlying abnormality. This may also vary from patients being completely asymptomatic to evidence of right heart failure or sudden cardiac death from underlying dysrhythmias (10).

The baseline ECG typically shows 'Himalayan' p-waves due to right atrial enlargement. QRS complexes may often be of low voltage with right axis deviation (as was present in our patient) and underlying right bundle branch block. Due to a dilated RA, many patients may present with atrial tachycardia's including atrial fibrillation, ectopic atrial tachycardia and atrial flutter. Ebstein's anomaly is also associated with accessory pathways predisposing patients to atrioventricular reentry tachycardias. This was likely the cause in our patient, although we were unable to document any episodes of tachycardia (10).

Imaging in the form of echocardiography is the gold standard for the diagnosis of Ebstein's anomaly (11). Apical displacement of the septal leaflet/posterior leaflet (>1.5 cm from insertion of mitral valve) and a large 'sail-like' anterior leaflet with varying degrees of tricuspid regurgitation are the most common findings. The most common associated structural findings are atrial septal defects. Some patients may present with cyanosis due to elevated filling pressures of the restrictive right ventricle with right to left shunt at atrial level.

Management is dependent on the severity of disease with surgery usually recommended for patients with severe right heart failure, evidence of paradoxical embolus and arrhythmias refractory to medical therapy or radiofrequency ablation (11).

Repair of Ebstein's anomaly is preferred to valve replacement, with the cone repair being the favoured approach. The fundamental principle of the cone repair is mobilization of the anterior and posterior tricuspid valve leaflets from their anomalous attachments on the right ventricle. The mobilized leaflets are then reimplanted to correct the apical displacement and ensure adequate coaptation of the tricuspid valve leaflets. Annular dilatation is a common finding and the insertion of an annuloplasty ring is frequently performed (12).

Our patient presented with features suggestive of right heart failure and complaints of episodes of palpitations, dyspnoea and dizziness without syncope. The noted prominent a-waves on examination of her JVP may be attributed to the pedunculated mass causing obstruction of the tricuspid orifice. The ECG was highly suggestive of Ebstein's anomaly with the marked 'Himalayan' p-waves and right axis deviation. Echocardiography confirmed the diagnosis of Ebstein's anomaly. The finding of a myxoma was unexpected. Accurate diagnosis and correct management of rare complex diseases rely on thorough clinical evaluation with correct interpretation of special investigations.

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**Declaration of interest**
The authors declare that there is no conflict of interest that could be perceived as prejudicing the impartiality of this case report.

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**Patient consent**
Written informed consent has been obtained from the patient for publication of the submitted article.

**Author contribution statement**
All authors were involved in care of the patient as well as compiling and proof-reading of the manuscript.

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