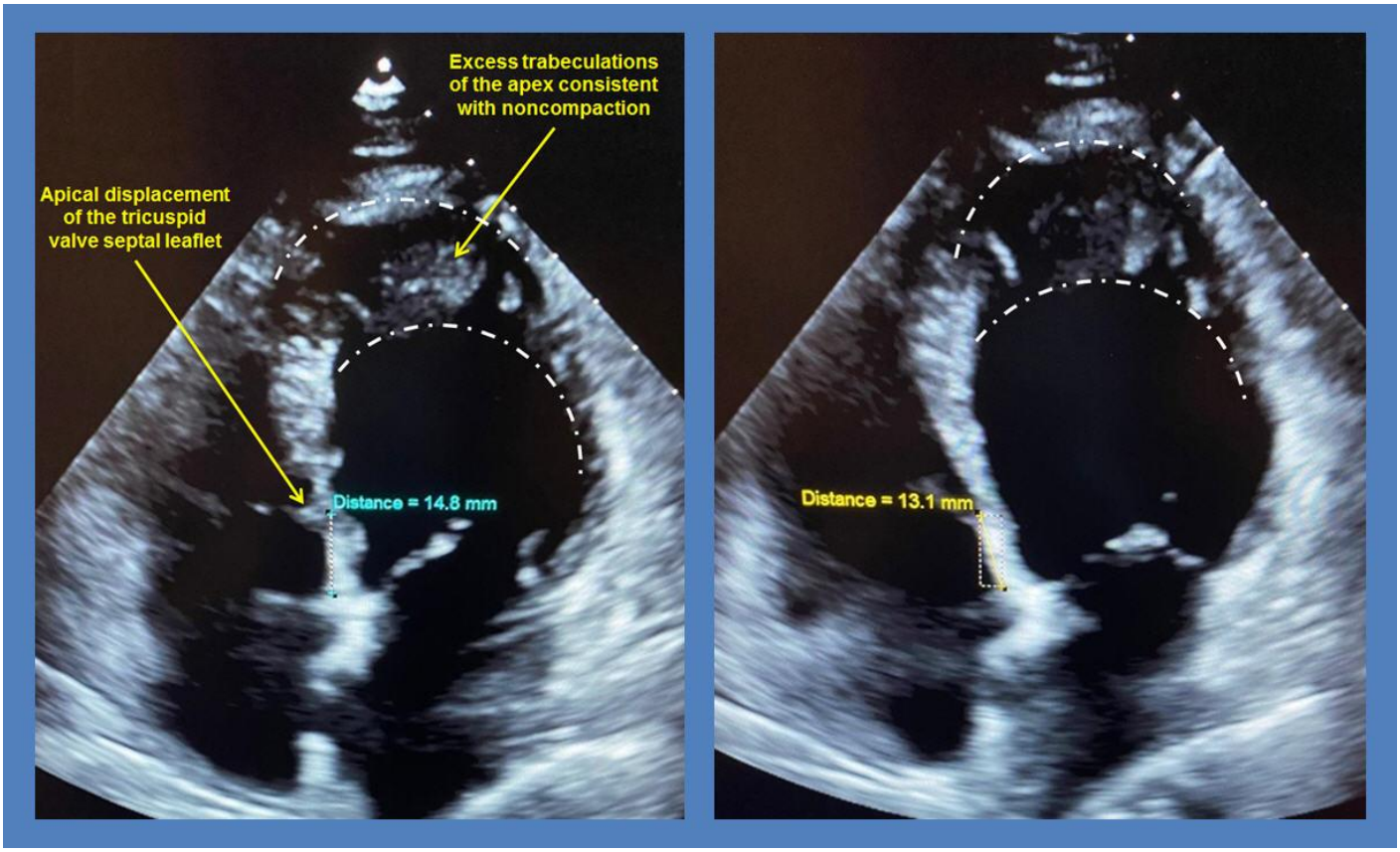
Ebstein's Anomaly.. *Associated LV Noncompaction!*

Mustafeez Ur Rahman, M.D.[a], Nikki Bardia, M.D.[a], Muhammad Rafique, M.D.[a], G. Mustafa Awan, M.D. [a], Landai Nguyen, D.O.[b], M.D., Bassam Omar, M.D., Ph.D. [a,c]



Description

The above 2-dimensional (2-D) apical 4-chamber echocardiography images and the accompanying video clip demonstrate apical displacement of the tricuspid valve annulus (by > 8mm/m2 body surface area) with tethering of the septal leaflet to the left ventricular septum, associated with redundancy of the leaflets and atrialization of a portion of the right ventricle; all features diagnostic of Ebstein's anomaly.

The left ventricular apex and lateral wall segments contain numerous prominent trabeculations with deep intertrabecular recesses, at least twice the thickness of the compacted wall, both in systole and diastole, which is highly suggestive of associated left ventricular noncompaction (LVNC). This condition is rarely associated with Ebstein's anomaly.

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a Division of Cardiology, University of South Alabama, Mobile, AL 36617, USA.

b The Heart Center of Northeast Georgia Medical Center, Cumming, GA 30041, USA.

c Corresponding Author: Bassam Omar, MD, PhD. Division of Cardiology, University of South Alabama, 2451 USA Medical Center Dr., Mobile, AL 36617, USA.

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Discussion:

Ebstein's anomaly involves apical displacement of the tricuspid annulus due to tethering of the septal and posterior tricuspid valve leaflets to the underlying right ventricular septum and free wall, a rare congenital lesion found in approximately one of 200,000 live births [1]. This results in atrialization of the right ventricle as the apically displaced tricuspid valve leaflets encroach upon the right ventricular (RV) apex and outflow tract, causing RV dysfunction. Multi-modality imaging can help diagnose the extent of the functional abnormalities and associated lesions [2].

While two-dimensional (2-D) echocardiography remains the standard method of assessment in Ebstein's anomaly [3], augmentation of the 2-D views with 3-D echocardiographic and transesophageal (TEE) imaging can provide important supplemental information about the extent of the functional abnormalities in addition to the presence and severity of associated anomalies or defects [4].

Ebstein's anomaly is often associated with an atrial septal defect and right-sided accessory pathways (Wolff-Parkinson-White; WPW), which can cause stroke [6], palpitations [7] and may contribute to progressive dyspnea [8]. Successful percutaneous ASD closure [9] and WPW ablation [10] in patients with Ebstein's anomaly have been reported.

Syndromic associations of Ebstein's anomaly are quite rare, and add tremendously to the complexity of its diagnosis and management. Ebstein's anomaly has been reported in association with Down's syndrome [11] in addition to Tetralogy of Fallot and absent pulmonary valve syndrome [12]. Other associations include Williams syndrome [13] and congenitally corrected transposition of the great vessels [14]. Extracardiac manifestations have also been reported in association with Ebstein's anomaly [15].

Another interesting association with Ebstein's anomaly is noncompaction cardiomyopathy [16]. This may involve left ventricular noncompaction [17], right ventricular noncompaction [18] or biventricular noncompaction [19]. The original classification of isolated ventricular noncompaction by Jenni et al [20] entailed the presence of deep perfused intertrabecular recesses by color Doppler echocardiography predominantly in the mid-lateral, apical and mid-inferior segments with a noncompacted myocardium twice the compacted layer in systole, in the absence other coexisting abnormalities. Other classifications have been proposed since then [21] with realization of the need for early management goals based on left ventricular systolic function and secondary complications [22].

While multiple genetic mutations have been associated with each of Ebstein's anomaly and noncompaction cardiomyopathy individually, the coexistence of both conditions together has been linked to genetic mutations involving the sarcomeric protein gene MYH7 [23] in addition to TPM1, encoding α-tropomyosin [24]. This combination appears to complicate the management of either condition individually and result in increased mortality [25].

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