UCLA Health

Department of Medicine
2023 Solomon Scholars Research Program

Abstract Form

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Project Title:	Polycythemia and Exertional Dyspnea: Uncovering an Anomaly

Research Category (please check one):

☐	**Original Research**	☒	**Clinical Vignette**	☐	**Quality Improvement**	☐	**Medical Education Innovation**

Abstract

Introduction: Ebstein anomaly is a rare congenital heart condition characterized by malformation of the tricuspid valve and right ventricle. Failed delamination of tricuspid valve leaflets results in apical displacement of the tricuspid valve, tricuspid regurgitation and an atrialized right ventricle.[1] There may be an associated patent foramen ovale (PFO) and/or atrial septal defect (ASD) with right to left shunting causing cyanosis with corresponding polycythemia from hypoxemia.[2] We present a case of a patient with a new diagnosis of Ebstein anomaly discovered during work up for secondary polycythemia.

Case report: A 43-year-old male was referred to Cardiology for dizziness, dyspnea on exertion, and elevated hemoglobin. His past medical history was notable for prior alcohol use disorder. He reported a normal childhood without functional limitation. On exam he had mild clubbing, a cardiac thrill and a sail sound, clear lungs and no lower extremity edema. His oxygen saturation was 93% at rest and decreased to 91% with ambulation. Labs were notable for a Hgb of 20.3g/dL, a high erythropoietin (EPO) of 40.8, and negative JAK2 mutation analysis. EKG revealed sinus rhythm with right atrial enlargement, right bundle branch block and no overt pre-excitation. Transthoracic echocardiography (TTE) showed Ebstein anomaly with apical displacement of the tricuspid valve (**Figure 1A**), with mild to moderate tricuspid regurgitation (**Figure 1B**). Given high suspicion for an atrial level shunt, transesophageal echo was done showing a brisk positive bubble study with frank right to left shunting across a PFO (**Figure 2**). He was referred to the adult congenital heart disease clinic for further management.

Discussion: This case illustrates a rare cardiac cause of secondary polycythemia as a physiologic response to longstanding hypoxemia from a right to left intracardiac shunt. Secondary polycythemia can be caused by chronic lung disease, EPO-producing malignant disorders, medications, renal artery stenosis and congenital heart disease. Serum EPO can differentiate between primary and secondary erythrocytosis, with a high EPO level suggestive of secondary polycythemia.[3,4] Workup should be guided by clinical history and physical exam and if cardiac abnormalities are suspected, as in this case, TTE should be the first step.

The clinical spectrum of Ebstein anomaly is variable depending on the extent of tricuspid valve displacement, tricuspid regurgitation and the associated defects and arrhythmias. As demonstrated here, patients can present later in adulthood with exertional dyspnea. Right to left shunting across a PFO occurs because of elevated right atrial pressure from tricuspid regurgitation and a small, residual right ventricle that cannot accommodate preload especially during exercise.[5] This leads to hypoxemia and secondary polycythemia that is distinctly different from shunt reversal in Eisenmenger syndrome. Management may include PFO closure to alleviate cyanosis and prevent paradoxical emboli with careful consideration of the effects on right atrial pressure.[6]

Figures:

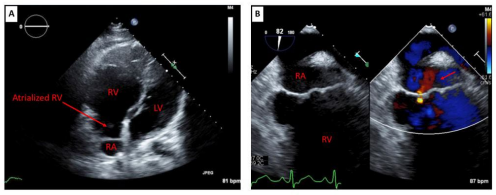

Figure 1. A) Transthoracic echocardiogram in apical four chamber view with atrialized RV (arrow) consistent with EA. **B)** Transesophageal echocardiogram with mild tricuspid regurgitation demonstrated (arrow).

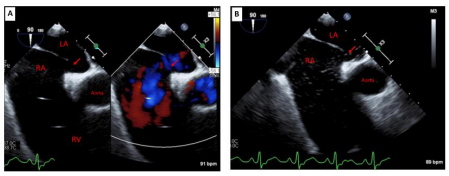

Figure 2. A) Transesophageal echocardiogram. There is an atrial septal gap consistent with a large PFO (arrow). Color doppler shows flow through the septum from the RA to the LA. **B)** Transesophageal echocardiogram with agitated saline contrast. The contrast microbubbles immediately appear in the LA after appearing in the RA demonstrating a right to left shunt.

References:

1. Attenhofer Jost CH, Connolly HM, Dearani JA, Edwards WD, Danielson GK. Ebstein's anomaly. *Circulation*. 2007;115(2):277-285. doi:10.1161/CIRCULATIONAHA.106.619338
2. Silva M, Teixeira A, Menezes I, et al. Percutaneous closure of atrial right-to-left shunt in patients with Ebstein's anomaly of the tricuspid valve. *EuroIntervention*. 2012;8(1):94-97. doi:10.4244/EIJV8I1A15
3. John Araujo, 2018. "Chronic Hypoxemic Syndrome and Congenital Heart Disease in Adults A Multisystemic Disorder," Journal of Cardiology & Cardiovascular Therapy, Juniper Publishers Inc., vol. 10(4), pages 70-80, May.
4. Mithoowani S, Laureano M, Crowther MA, Hillis CM. Investigation and management of erythrocytosis. *CMAJ*. 2020;192(32):E913-E918. doi:10.1503/cmaj.191587
5. Sommer RJ, Hijazi ZM, Rhodes JF. Pathophysiology of congenital heart disease in the adult: part III: Complex congenital heart disease [published correction appears in Circulation. 2009 Jun 2;119(21):e547]. *Circulation*. 2008;117(10):1340-1350. doi:10.1161/CIRCULATIONAHA.107.714428
6. Stout KK, Daniels CJ, Aboulhosn JA, et al. 2018 AHA/ACC Guideline for the Management of Adults With Congenital Heart Disease: A Report of the American College of Cardiology/American Heart Association Task Force on Clinical Practice Guidelines [published correction appears in Circulation. 2019 Apr 2;139(14):e833-e834]. *Circulation*. 2019;139(14):e698-e800. Doi:10.1161/CIR.0000000000000603