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Case Report

A case of Ebstein's anomaly

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ABSTRACT

Ebstein's anomaly (EA) is a rare congenital heart disease. It has varied clinical presentation depending upon the spectrum of deformation and function of the tricuspid valve and other structures. A comprehensive evaluation by non-invasive modalities is required for optimal decision making regarding the approach to surgical correction of the disease.

Keywords: EA, Tricuspid valve, ECG

INTRODUCTION

Ebstein's anomaly (EA) is a rare congenital heart disease with prevalence of 2.4 per 10,000 live births with equal male/female distribution.[1] It is classically characterised by the apical displacement of septal and posterior leaflet of tricuspid valve (TV) but it is not the sole feature of the disease. The natural history is related to various anatomical aspects other than abnormality of tricuspid valve. A detailed evaluation by echocardiography is needed to make clinical decision. Here, we report a case of EA with a comprehensive evaluation by echocardiography.

CASE REPORT

This is a case of a 7-year-old male child who presented to cardiology OPD with the complaint of dyspnoea on exertion, bluish discolouration, syncope and poor weight gain. On examination, he had saturation of 82% in all four limbs on room air, cyanosis, grade IV clubbing, pansystolic murmur in left lower parasternal border and split S2. His electrocardiogram showed tall P wave, right axis deviation and right bundle branch block pattern (Figure 1). His chest X-ray revealed cardiomegaly with right ventricular and right atrial enlargement (Figure 2). His two-dimensional transthoracic echocardiography in apical four chamber view revealed enlarged right chambers with significant apical displacement of septal leaflet of tricuspid valve and severe tricuspid regurgitation (Figure 3 and 4). This made the diagnosis of EA. But for surgical decision more information was required like displacement index, Carpentier classification, Great Ormond Street echocardiography (GOSE) score, Shina Tajik index and evaluation of right and left ventricular function and associated cardiac anomaly. Displacement index was calculated from the insertion point of anterior mitral leaflet to the hinge point of the septal tricuspid leaflet divided by body surface area. It was found to be 38.46 mm/m^2. The anterior leaflet was tethered and there was a large non contractile atrialised right ventricle. So, it was Carpentier's Type C. On quantification, combined area of atrialised right ventricle (aRV) and right atrium (RA) was 53.5 cm^2, whereas the combined area of the functional right ventricle (fRV), left ventricle (LV) and left LA was 24.3 cm^2. GOSE Score was calculated as ratio of combined area of RA + aRV) and combined area of fRv+LV+LA. Grading is given as Table 1.

So, the calculated GOSE score was 2.16 which made it grade IV. Shina Tajik index was calculated from parameters given in table 2 and it was found to be 6 out of 10.

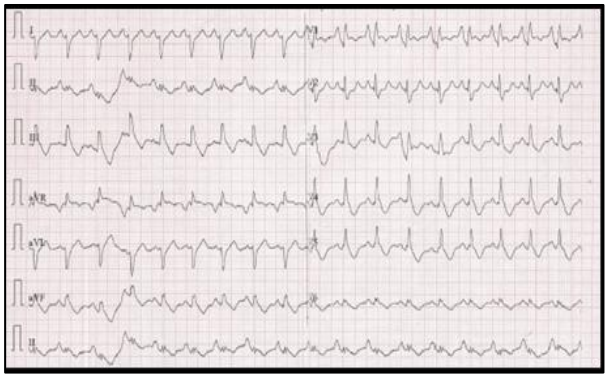

Figure 1: ECG of the patient with EA showing Himalayan P wave, right bundle branch block and right axis deviation.

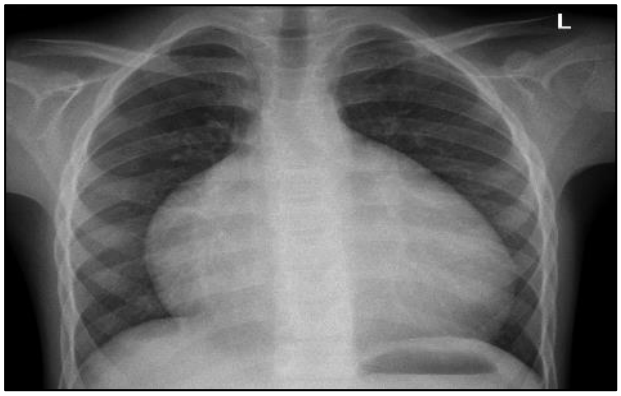

Figure 2: Chest X ray PA view of the patient with EA.

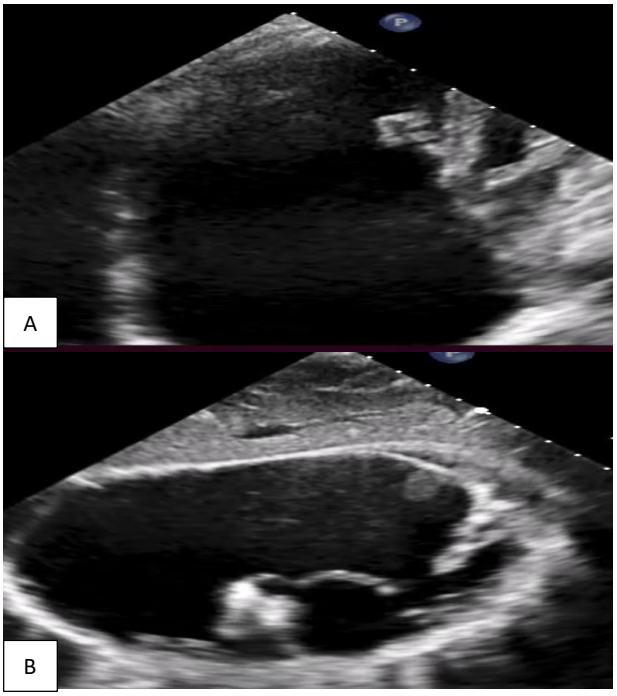

Figure 3 (A and B): 2D echocardiography apical 4 chamber view showing apical displacement of septal tricuspid leaflet. Subcostal view showing septal leaflet in RVOT and tethering of anterior leaflet.

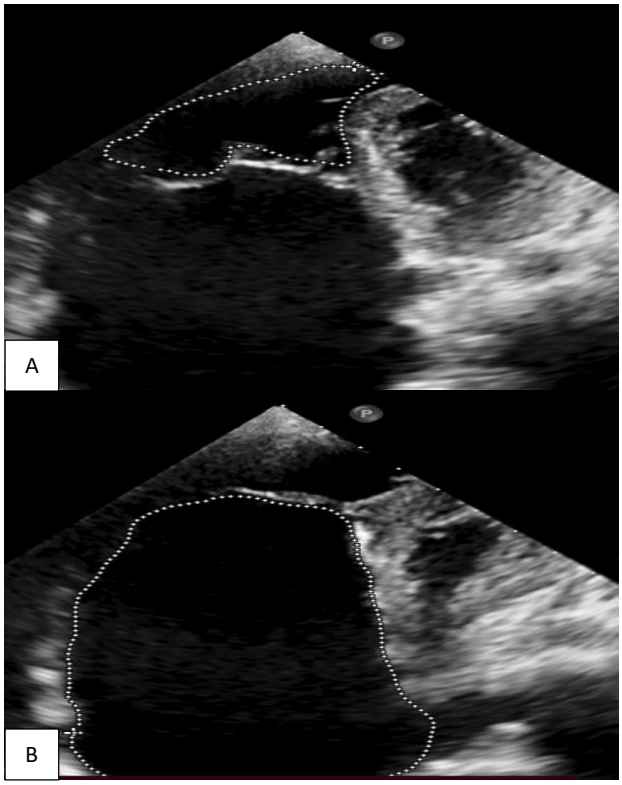

Figure 4 (A and B): Transthoracic 2D echocardiogram of functional RV area and combined RA and aRV area.

Table 1: Celermajer index (GOSE score).[6]

Grading	GOSE score
Grade I	<0.5
Grade II	0.5-0.99
Grade III	1-1.49
Grade IV	>1.5

Table 2: Shina Tajik severity index.[7]

2D echocardiographic features		Index number	Our case
Septal leaflet	Absent	1	
	Displacement ≥ 25 mm/m^2	1	+
Anterior leaflet	Displacement	1	+
	Displacement of free edge	1	+
	Restricted motion	1	
	Severe prolapse	1	
Intracardiac cavities	Aneurysmal RVOT*	1	+
	aRV/RV ≥ 50%	1	+
	Tricuspid annulus ≥ 45 mm/m^2	1	+
	Right atrium ≥ 60 mm/m^2	1	
Total		10	6

*RVOT- right ventricular outflow tract.

Right and left ventricle had normal contractility and there were no other associated lesions. There was no right ventricular outflow tract (RVOT) obstruction. Cardiac MRI (Figure 5) was done for delineating the structural abnormalities which matched with echocardiographic parameters.

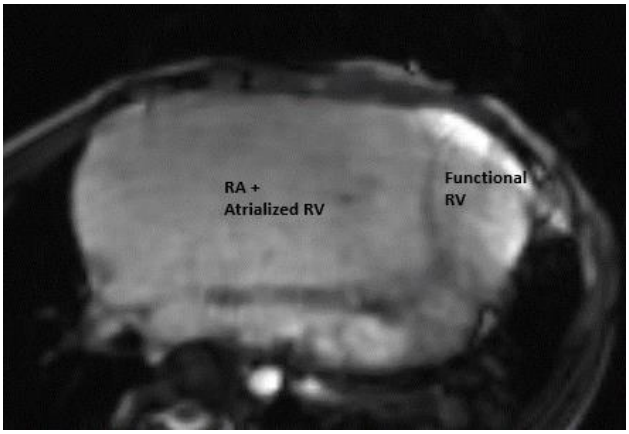

Figure 5: Cardiac MRI 4 chamber view showing small functional RV.

DISCUSSION

Ebstein's anomaly was described in 1866 by Wilhelm.[2] ECG may have Himalayan P waves due to the prolonged conduction throughout the enlarged right atrium.[3] This is similar to our case where P waves were 6 mm which reflects enlarged right atrium. Typical chest x ray is a boxed shaped heart (cardiomegaly) which is an indication for surgery. Echocardiography has dramatically led to earlier diagnosis as shown by Celermajer et al.[6] But despite, 10% remain undiagnosed into adulthood[4]. Echocardiogram classically shows apical displacement of septal leaflet of tricuspid valve. The clinical course is not only related to the displacement of septal leaflet of TV but also the RV function, RV size, LV function and ASD. Displacement of septal leaflet is best viewed in apical four chamber view.[4] A displacement index $\geq$ 8 mm/m^2 supports the diagnosis of EA.[4] In our patient, the displacement index was 38 mm/m^2. Surgical repair is useful when the anterior leaflet is thin and translucent, but when it is heavily muscularised and tethered valve repair is more challenging.[4] The sail like anterior leaflet may lead to obstruction of the RVOT and functional pulmonary atresia. Here in our patient, there was presence of forward flow in RVOT and pulmonary valve and there was no RVOT obstruction. Poor outcome and high mortality in new born predicted when Celermajer (GOSE score) index are more than 1 (Table 1).[6] This may be due to marked cardiac enlargement and lung hypoplasia. Reduced LV function and pulmonary atresia (both functional and anatomic) are poor prognostic factors. Shina et al studied detailed echocardiographic features of 41 patients of EA and found that two-dimensional evaluation of structural abnormality may aid in determining anatomic severity and surgical feasibility.[7] In our study, Shina Tajik severity index (Table 2) was found to be 6 out of 10 which suggests severe structural abnormality. In our patient cardiac MRI revealed similar findings. Cardiac CT and MRI are rarely utilised in infants with EA due to motion artifact and heart rate.

CONCLUSION

Echocardiography remains the gold standard in EA not only for the diagnosis but it also aids in surgical approach and feasibility. So whenever feasible a comprehensive echocardiogram should be done before referral to cardiac surgeon.

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