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DREADFUL COEXISTENCE OF EBSTEINS ANOMALY WITH DOUBLE OUTLET LEFT VENTRICLE IN A NEONATE – A RAREST OF RARE CASE REPORT WITH REVIEW OF LITERATURE

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ABSTRACT

Ebstein's anomaly (EA) is a rare congenital heart disease that presents with apical displacement of the septal and posterior leaflets of the tricuspid valve. The incidence of Ebstein's anomaly is about 1 per 200,000 live births. Likewise, double-outlet left ventricle (DOLV) is a scarce congenital heart disease entity, seen in 5 in 100,000 new born comprising 1% of all congenital heart defects where both aorta and pulmonary artery arise from the left ventricle. Herein we are reporting a rarest of rare coexistence of EA with DOLV, in a 3 day old neonate.

KEYWORDS: Ebsteins Anomaly, Double-Outlet Left Ventricle, Congenital, Heart Disease, Pulmonary Atresia.

INTRODUCTION

Ebstein's anomaly is a rare congenital heart disorder occurring in ≈1 per 200,000 live births and accounting for <1% of all cases of congenital heart disease.[1,2]

Ebstein's anomaly is characterized by various degrees of inferior displacement of the proximal attachments of the tricuspid valve (TV) ring, TV dysplasia, right ventricular dysplasia, and abnormalities of the distal attachments of the TV (Figures 1-4).

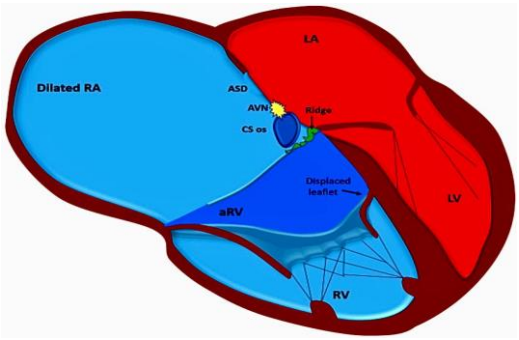


Figure 1: Diagrammatic illustration of a heart with Ebstein's anomaly highlighting the anatomical abnormalities. The right atrium (RA) and tricuspid annulus is dilated. The septal tricuspid leaflet is apically displaced (arrow). The coronary sinus (CS) ostium is dilated, AV node (AVN) is irregular. ASD, atrial septal defect; aRV, atrialised RV; LA, left atrium; LV, left ventricle; RV, right ventricle.

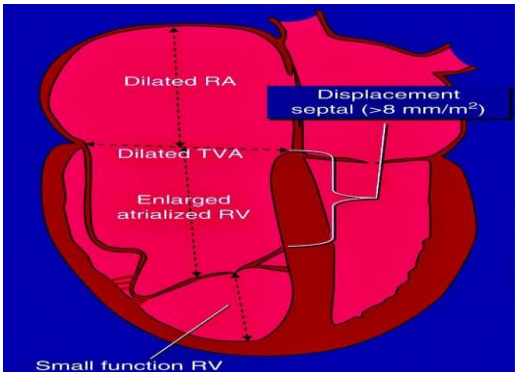


Figure 2: Diagram of the "displacement index." The distance from the hinge point of the anterior mitral leaflet is measured to the hinge point of the delaminated septal tricuspid leaflet. This measurement divided by the body surface area equals the displacement index. A displacement index >8 mm/m2 is diagnostic of Ebstein anomaly. A small functional right ventricle (RV) is present inferior to the coaptation point of the tricuspid valve. RA, right atrium; TVA, tricuspid valve annulus.

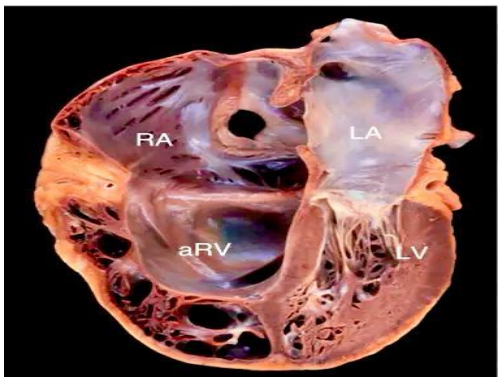


Figure 3: Severe Ebstein anomaly: pathology specimen aRV, atrialized right ventricle.

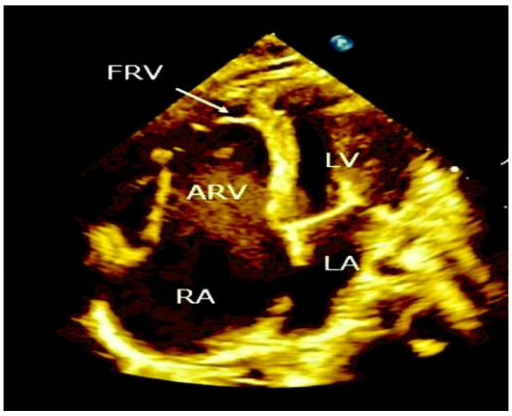
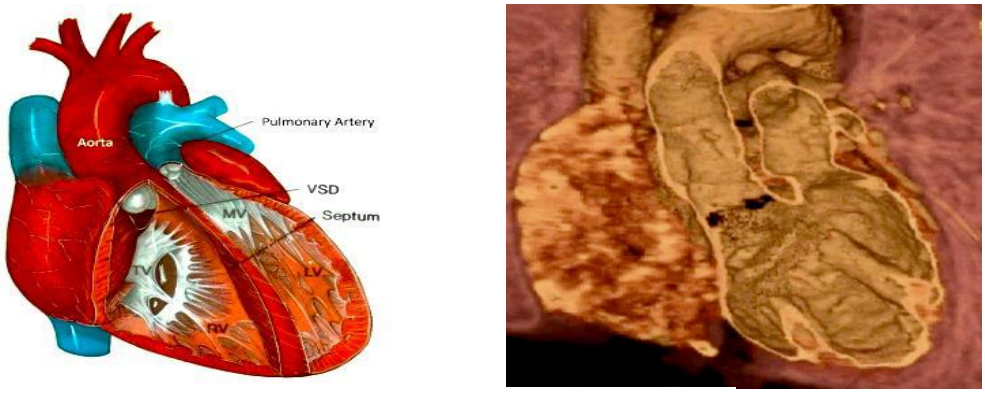


Figure 4: Transthoracic echocardiography apical four chamber view. RA, right atrium; LA, left atrium; ARV, atrialized right ventricle; FRV, functional right ventricle.

These morphologic deformities, in widely different degrees of severity, are associated with a variety of hemodynamic alterations leading to cyanosis, congestive heart failure, and arrhythmias. Refinement of tricuspid valvuloplasty and plication techniques[3-4] has opened the way to a satisfactory long-term outlook for the majority of older children and adults, who generally are only mildly or moderately symptomatic. Ebstein's anomaly presenting in neonates and young infants, however, has considerably less favorable natural history with a reported mortality rate of as high as 75%.[5-6]

Double-outlet ventricles with concordant AV connection account for 1% of all cases of congenital heart disease, and DOLV accounts for <5% of those cases[7,8] (Figure 5). The exact incidence is not known, but <1/200,000 live births have been reported.[7]



(A) **(B)**

Figure 5: (A) Diagram of the most common form of DOLV showing situs solitus and AV concordance, rightward/anterior or right/lateral Ao with subvalvar or valvar pulmonary stenosis. MV, Mitral valve; TV, tricuspid valve. (B) Cardiac CT with 3D reconstruction of a patient with DOLV . The great arteries are lying side by side and originating from LV.

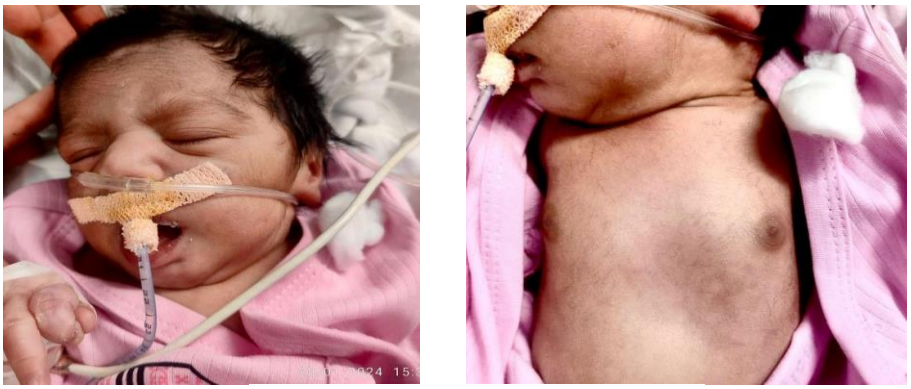
After deep search of the literature, only a handful of cases of EA accompanying with DOLV could be encountered[9,10,11] Chang et al[9] reported a case of Ebsteins anomaly with DOLV, subaortic VSD and pulmonary stenosis in association with esophageal atresia with tracheo-esophageal fistula (so-called VACTERL association) in a neonate. The patient underwent Blalock-Taussig shunt. Bharati et al[10] mentioned in their series of DOLV that there was only one autopsied case of associated Ebsteins anomaly. In that remarkable case the aorta emerged completely from the left ventricle over a well developed conus. The pulmonary trunk overrode the septum over a posteriorly placed VSD, but emerged mostly from the left ventricle. The aorta was anterior and to the right of the pulmonary trunk and the latter was related to the mitral valve. The VSD was related to the tricuspid but not the mitral valve. This case was associated with fetal coarctation and PDA. Van Praagh et al[11] narrated in their series of 109 cases of DOLV; only one autopsy case, presented with DOLV, sub aortic, VSD, pulmonary stenosis and Ebsteins anomaly.

It is noteworthy that all of these patients of DOLV coexisting with EA had a malignant course of illness, leading to very early mortality.

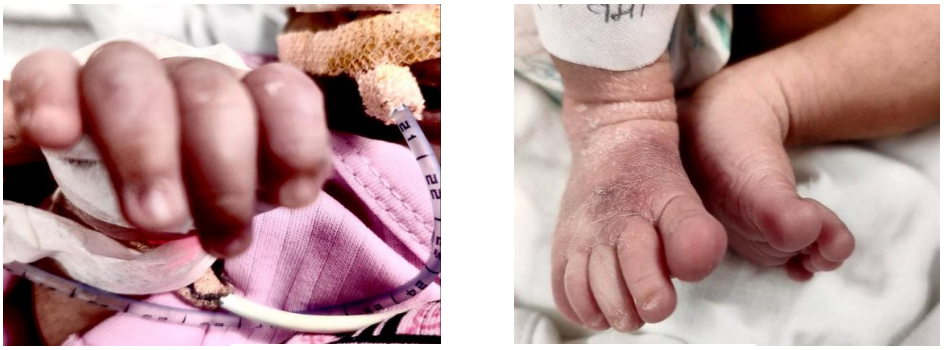
Case Report

A 3 day old female neonate was referred to us for a comprehensive cardiovascular evaluation.

She was a full term delivery by Caesarean section and was delivered at a private hospital, from a primipara woman of 21 years of age. There was no history of maternal risk factors of CHD (obesity, diabetes, febrile illness, smoking, alcohol intake, teratogenic drug use, or radiation exposure). On clinical examination, the child was very "sick-looking" and was in congestive heart failure as evidenced by the presence of respiratory distress, intercostal retractions, tachypnea, facial edema and swelling in all the four extremities (Figure 6 A-D).



(A) **(B)**



(B) **(D)**

Figure 6: (A) Facial edema, (B) Pectus excavatum, intercostal retractions, (C) Cyanosis of fingers, (D) Cyanosis of toes.

She was of average built, highly irritable and persistently crying. Her weight was 3.7 kg, respiratory rate was 38/min, pulse rate was 98/mm, blood pressure was 80/60 mmHg, and SPO2 was 65% at room air. The child was cyanosed with bluish coloration of tongue, lips, all the fingers, and toes. There was a typical pectus excavatum deformity of the chest without any other musculoskeletal anomalies. All the peripheral pulses were normally palpable without any radio femoral delay. Rest of the systemic examination was unremarkable.

On cardiovascular examination there was presence of Grade 2/6 pansystolic murmur over precordium, best head over lower left sternal border.

Xray chest (AP view) (Figure 7) revealed massive cardiomegaly with a cardiothoracic ratio of 0.78. Moreover, there was severely diminished pulmonary blood flow.

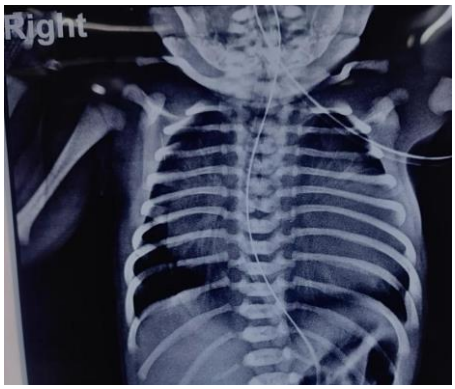


Figure 7: Xray chest A-P view: massive cardiomegaly with cardiothoracic ratio of 0.78 and pulmonary oligemia.

Resting ECG exhibited sinus tachycardia with a ventricular rate of 98/min, partial RBBB with a right axis deviation, and ”Himalayan” P waves (Figure 8).

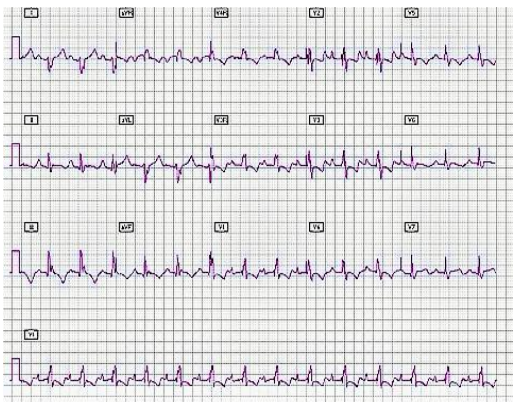


Figure 8: Resting ECG shows sinus tachycardia, partial RBBB, right axis deviation and “Himalayan” P waves.

Transthoracic color doppler echocardiography (Figures 9-14)

Standard transthoracic color doppler echocardiography was performed by the author in the classical subcostal, parasternal long axis (LX), parasternal short axis (SX), 4-Chamber (4CH), 5-Chamber (5CH) and suprasternal views.

The echocardiographic characteristics of the neonate are outlined

(I) Levocardia
Situs Solitus
Concordant D-Bulboventricular Loop
AV Concordance
DOLV – Both great arteries are arising from LV with D-malposition of great arteries.
Left Aortic Arch
Confluent pulmonary arteries.
Normal pulmonary venous drainage.
Normal systemic venous drainage.

(II) EBSTEIN'S ANOMALY - (Carpentier Type D)

- TRICUSPID REGURGITATION (SEVERE)
- Septal leaflet of TV is apically displaced.
- Tricuspid septal leaflet displacement of 13 mm from the mitral insertion.
- The anterior leaflet is large and immobile with the tips adhered to the lateral wall of RV.
- TV orifice is displaced downwards into the RV cavity.
- Low velocity TR jet present (TR velocity 1.69 m/sec).
- On color flow mapping TR jet area 4.15 sq.cm; occupying 45 % of RA area, central jet .

(III) ATRIAL SEPTAL DEFECT (LARGE)
Ostium secundum type
Size 6.3 mm
Rt. to Lt. Shunt.
Superior and inferior rims of ASD are flail and hyper mobile.

(IV) DOUBLE OUTLET LEFT VENTRICLE

- Both great arteries are arising from the left ventricle.

(V) D-MALPOSITION OF GREAT ARTERIES.

- Aorta is anterior and to the right of pulmonary artery
- Pulmonary artery is posterior and to the left of Aorta

(VI) ATRESIA OF THE PULMONARY VALVE alongwith

- Severe hypoplasia of branch pulmonary arteries.

MPA(D) 3.0 mm
LPA(D) 2.2 mm
RPA(D) 2.3 mm

- A thin solitary aorta-pulmonary collateral was visualized connecting descending aorta to the left pulmonary artery with left to Rt shunt.

(VII) Huge RA (Atrialized RV)
Dilated RV
Normal biventricular systolic function.
Normal LVEF = 50 %
No regional wall motion abnormality present.

(VIII) No evidence of ventricular septal defect, coarctation of aorta or bicuspid aortic valve.

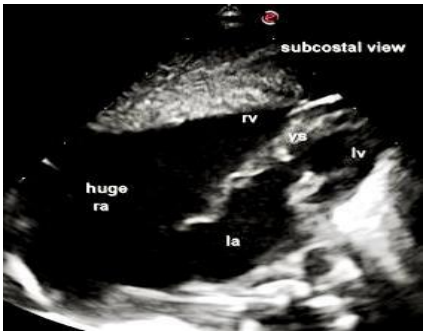


Figure 9: Subcostal View shows dilated RV and huge RA.

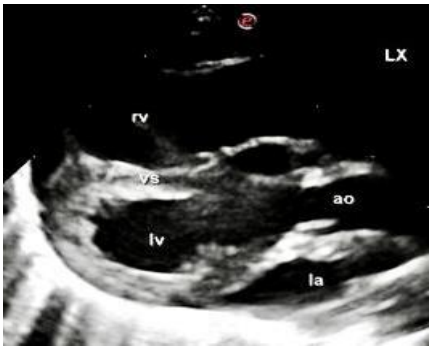
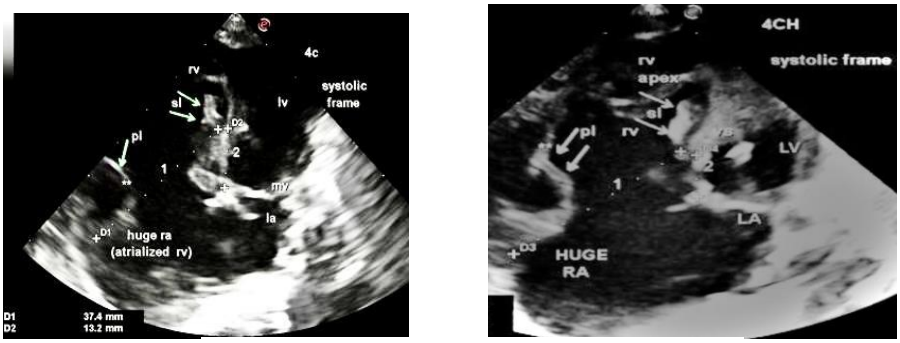
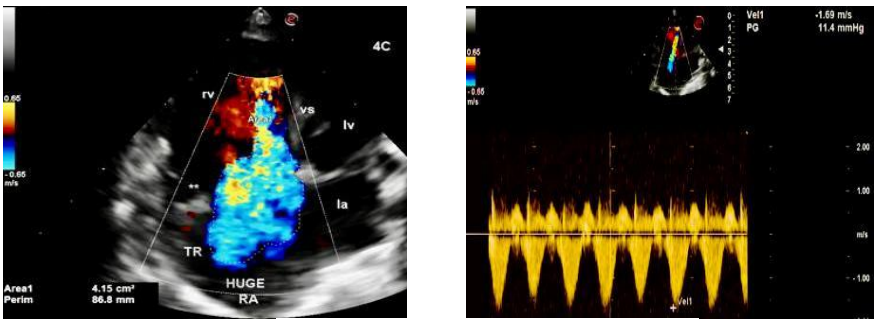


Figure 10: LX View exhibits dilated RV and normal LV size.



(A) **(B)**

Figure 11: (A) 4CH View(systolic frame) displays normal sized LV,LA, dilated RV, Huge RA (Atrialized RA), solitary oblique arrow denotes large anterior leaflet adhered to the lateral right ventricular free wall , dilated TV annulus is measuring 37.4 mm (designated as 1), sl and two oblique arrows indicate rudimentary septal leaflet of TV ,tricuspid septal leaflet displacement from mitral insertion is 13.2 mm (designated as 2), which is consistent with severe grade of Ebsteins anomaly- (Carpentier Type 4). (B) 4CH View (systolic frame)- this image represents enlarged view of figure 11(A).



(A) **(B)**

Figure 12: (A) 4CH view. On color flow mapping a severe tricuspid regurgitation (TR) jet is visualised. (B) On continuous wave Doppler analysis across TV, a low velocity signal of TR jet is delineated(TR velocity 1.69 m/sec).

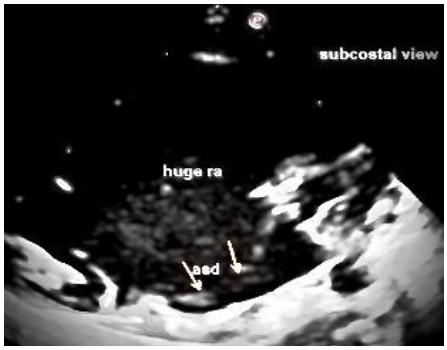
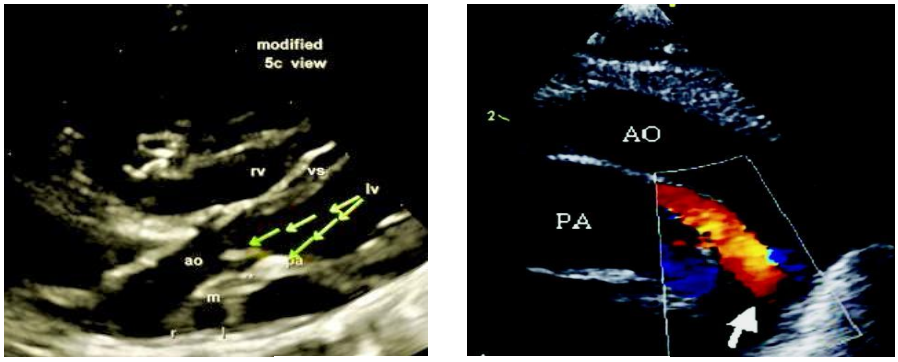


Figure 13: Subcostal View exhibits a large ostium secundum ASD with huge RA.



(A) **(B)**

Figure 14: (A) Modified 5CH View clearly delineating Double Outlet Left Ventricle with D- malposition of great arteries- (aorta is anterior and to the right of pulmonary artery) with atresia of the pulmonary valve (designated as pa and **) with hypoplasia of main, left and right pulmonary arteries, m, main pulmonary artery, l, left pulmonary artery, r, right pulmonary artery. (B) Suprasternal View. A solitary aorto-pulmonary collateral is delineated by thick arrow.

The detailed transthoracic echocardiography illustrates an extremely rare and lethal combination of neonatal Ebsteins Anomaly, DOLV, large ASD with right to left shunt, atresia of the pulmonary valve, hypoplatic branch pulmonary arteries, severe TR with advanced heart failure and hypoxia. Hence, the patient's parents were advised for admission and management of the child at a tertiary care pediatric cardiovascular institution.

Review of literature

Tricuspid valve anatomy in ebsteins anomaly

Normally, the TV has three valvar leaflets referred to as the anterosuperior, the septal and the mural leaflets. However, in EA, the anterosuperior is the largest, redundant anterior leaflet, which contains fenestrations. It stretches from the infundibulum anteriorly to the inferolateral wall posteriorly, whereas the septal leaflet, the smallest, arises medially from the annulus above the interventricular septum. Posterior leaflet attaches along the posterior margin of the tricuspid annulus from the septum to the inferolateral wall.[12] Embryonic failure of delamination of the septal, inferior and anterior leaflets of the TV results in the apical displacement of the tricuspid leaflets to the underlying RV myocardium. Such failure in delamination creates the characteristic downward (apical) displacement of the functional orifice and dilation of the atrialized right ventricle (aRV), with various degrees of hypertrophy and thinning of the wall. This malformation is characterized by the displacement of the points of attachment, or the hinges, of the septal and posterior leaflets into the right ventricle, away from the atrioventricular junction. As the anterosuperior leaflet has a different developmental origin, its junctional hinge usually retains a normal position.[13,14] The failure in delamination also results in various degrees of displacement of TV leaflets, and the movement of the tricuspid hinge points both anteriorly and toward the right ventricular apex. The adherent portions of the valvar leaflets usually have little or no motion. This generally leads to tricuspid regurgitation or rarely to stenosis.[2,12,15] Chordae tendinea of anterior leaflets are generally short, tethered, poorly formed and severely deformed. Therefore, the only mobile leaflet tissue is displaced into the right ventricular outflow tract (RVOT), where it may cause obstruction or forms a large sail-like intracavitary curtain. The septal and mural leaflets are usually rudimentary, dysplastic or may be absent due to failure of delamination. These leaflets may be freely mobile or adhered (tethered) to the endocardium.[16] The maximal displacement is at the commissure level between the mural and septal leaflets of the TV.[14] Apical displacement of the septal leaflet by at least 8 mm/m2 of body surface area is considered as a diagnostic feature of EA in the echocardiographic evaluation.[2] There is often a marked dilatation of the true TV annulus, and the aRV separating this true annulus from the functional right ventricle (fRV).[2,17]

Atrium and atrioventricular annulus

The right atrium is usually very much dilated, and the right atrioventricular junction, or true annulus of the TV, is enlarged circumferentially. The valve of the inferior vena cava (eustachian valve) is often very prominent.[14,16]

Right ventricle

Because of the displaced TV, the RV is divided into two regions in Ebstein's anomaly: the inlet portion [atrialized right ventricle (aRV)] and the trabecular or outlet portion [functional ventricle (fRV)]. The inlet portion, directly involved with the malformation, is functionally integrated with the right atrium, whereas the outlet portion constitutes the functional RV. The aRV usually has a thinner wall than the fRV and may account for more than half of the RV volume in extreme cases, instead of its usual location in one-third of the RV.[13,16] There is often a marked dilatation of the true TV annulus and a large chamber separating this annulus from the functional RV. Two-thirds of EA cases possess dilated RV, which commonly involve the functional RV apex and outflow tract. In some cases, RV dilatation is so significant that the ventricular septum bulges leftward, compressing the left ventricular (LV) chamber, and may cause episodic left ventricular outflow tract (LVOT) obstruction.[13] In such cases, the short-axis view demonstrates a circular right ventricle and a crescentic left ventricle.

Ebsteins Anomaly-Classifications

There are multiple classifications for the description of the anatomic severity of EA. The amount of displacement and tethering of the leaflets and the degree of RV dilatation are assessed.

1. Adult congenital heart disease Anatomical and Physiological classification (ACHD AP)

The new adult congenital heart disease anatomical and physiological (ACHD AP) classification[18] system defines EA as a lesion of anatomically moderate complexity (anatomical type II) (Table 1).

Table 1: ACHD AP Classification (CHD Anatomy + Physiological Stage = ACHD AP Classification).

CHD Anatomy
I. Simple • Native disease • Repaired conditions **II. Moderate Complexity** • Repaired or Unrepaired conditions **III. Great Complexity (or Complex)**
Physiological Stage
A. NYHA FC I symptoms B. NYHA FC II symptoms C. NYHA FC III symptoms D. NYHA FC IV symptoms

ACHD, adult congenital heart disease; AP, anatomic and physiological; FC, functional class.

A recent publication showed that such anatomical cardiac malformations carried an operative mortality risk varying from 1% (0.69–1.37%) in the physiologically lowest level of severity (IIA), to 5% (3.05-7.91%) in the highest level of severity (2D).[19]

2. Simple classification

Mild

Moderate

Severe

The amount of displacement and tethering of the largest and the degree of RV dilation are assessed

3. Clinical classification of ebsteins anomaly

Ebstein's anomaly is classified into three types according to the clinical features, heart catheterization data, angiocardiographic and anatomical findings obtained on surgery or autopsy.[20]

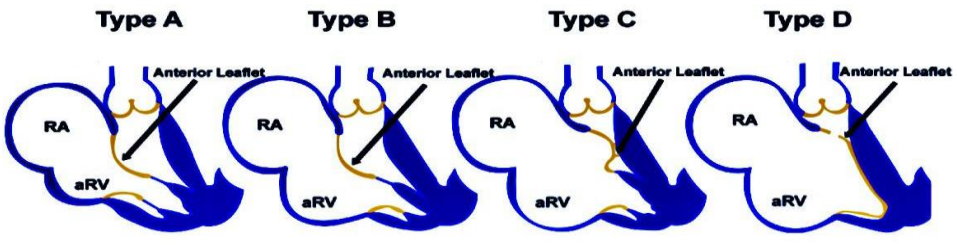
A. Tricuspid Stenosis Dominant Type
- Cyanosis
- Severe symptoms
- Mild to moderate cardiomegaly
- "Double ball sign" on angiocardiography

B. Tricuspid Insufficiency Dominant Type
- Cyanosis
- Mild symptoms
- Severe cardiomegaly
- "Double ball sign" pesent

C. Mild Type
- Mild Cyanosis
- No or mild symptoms
- Mild to moderate cardiomegaly
- No "Double ball sign" present

4. Carpentier Classification

According to the classification of Carpentier [3], EA was divided into four types (Figure 15).
- Type A: Mild apical displacement of the tricuspid valve leaflets with the adequate functional right ventricle.
- Type B: Moderate apical displacement of the tricuspid leaflets with a moderate reduced size but adequate functional right ventricular volume with freely mobile anterior leaflet.
- Type C: Severe apical displacement of the tricuspid valve leaflets with a small functional right ventricle. Anterior leaflet movement is restricted due to abnormal chordal attachments that cause right ventricular outflow tract obstruction.
- Type D: Complete non-delamination of the tricuspid valve leaflets with almost complete atrialization of the right ventricle, only infundibular portion of the right ventricle remaining: "Tricuspid sac".



	A	B	C	D
Septal/Posterior Leaflet Displacement	Mild	Moderate	Severe	Extremely Severe (Tricuspid Valve Sac)
Anterior Leaflet	Normal Morphology and Mobility	Abnormal Attachments with Normal Mobility	Partial Adhesions to the Ventricular Free Wall and Restricted Motion	Severe Adhesions to the Ventricular Wall
"Atrialized" Ventricle	Small/Preserved Function	Moderate/Reduced Function	Large/Reduced Function	Very Large/Minimal Function
Functional Ventricle	Normal/Preserved Function	Reduced/Preserved Function	Small/Reduced Function	Infundibulum/Depressed Function

Figure 15: Carpentier classification.

5. Celermajer classification

The Celermajer classification of EA [21] was according to echocardiographic measurements calculating the ratio of the combined area of the right atrium and aRV to that of the fRV and the left heart in a four-chamber view at the end diastole (GOSE = RA + aRV/fRV + LV + LA) . This is an echocardiographic grading score for neonates with Ebstein's anomaly, The Great Ormond Street Echocardiography (GOSE) score, with grades 1 to 4 (Figure 16). Increasing severity, that is, a higher grade, was associated with a high mortality rate. This classification is particularly helpful with neonatal Ebstein's anomaly.[22] GOSE score and mortality rate are demonstrated in the Table 2.

Table 2: GOSE Score and Mortality rate.

GOSE score	Ratio	Mortality rate (%)
Grade 1	<0.5	5–8
Grade 2	0.5–0.99	8–10
Grade 3	1–1.49	45 (acyanotic) 100 (cyanotic)
Grade 4	>1.5	100

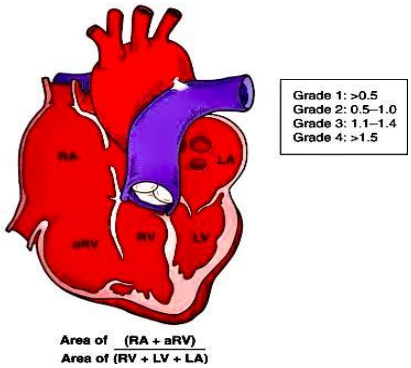


Figure 16: GOSE score. RA, right atrium; aRV, atrialized right ventricle; LA, left atrium; LV, left ventricle.

6. SAS prognostic score

The SAS (Simpson Andrews Sharland) score is another prognostic score[23] that uses as a weighted model to predict mortality. The cardiothoracic ratio, the Celermajer index, pulmonary valve flow, duct flow, and left-right ventricular ratio are graded 0, 1, 2, points each to generate a score. In studies, a score of 5 predicted 100% mortality, and a score of less than or equal to 3 predicted 91% survival.

5. Becker's dysplasia classification

A modified version of Becker's dysplasia classification [24] of Ebsteins valve is described (Figure 17-19). The degrees of leaflet tethering to the ventricular wall were calculated according to their extension:

Grade I - up to 25% of the distance from the atrioventricular junction to the apex.

Grade II - 25-50% of the distance

Grade III - > 50% of the distance.

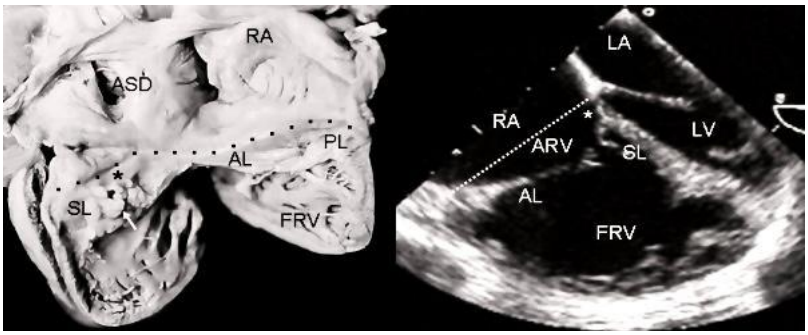


Figure 17: (a) Internal view of the right chambers of a heart with Ebstein's anomaly with mild (Grade I) tethering of the septal leaflet (asterisk). Thickening of the free portion of the leaflet (arrow) is evident. The dotted line represents the atrioventricular junction where the tricuspid fibrous ring is located. (b) The 4 chamber echocardiographic image shows the same type of findings seen in the anatomic specimen. The dotted line indicates the plane of atrioventricular junction. RA: Right atrium; ARV: Atrialized right ventricle; FRV: Functional right ventricle; AL: Anterior leaflet; PL: Posterior leaflet; SL: Septal leaflet; ASD: Atrial septal defect; LA: Left atrium; LV: Left ventricle.

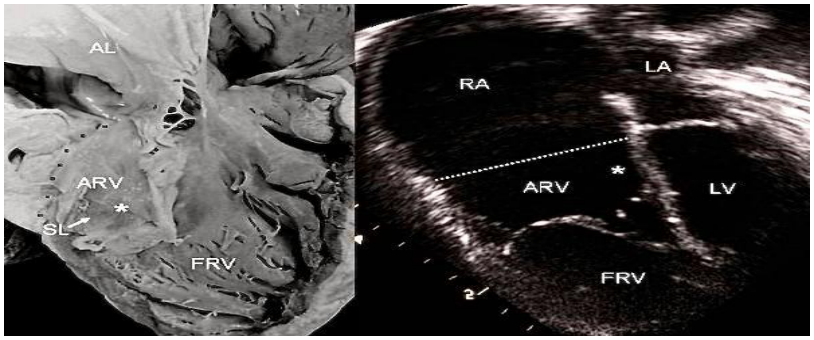


Figure 18: (a) Internal view of the right ventricle shows grade II tethering of the tricuspid septal leaflet (asterisk). (b) The 4 chamber echocardiographic image shows discontinuous leaflet tethering similar to the anatomic specimen.

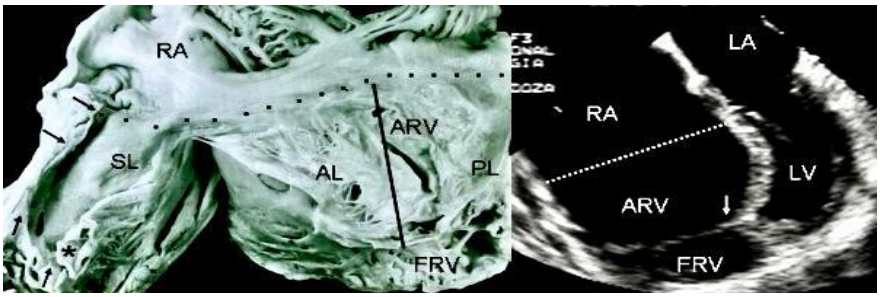


Figure 19: (a) Internal view of the right chambers of a heart shows grade III tethering of the tricuspid septal leaflet. The arrows point to tethering of the posterior leaflet. The flap of the free portion of the septal leaflet can be seen (asterisk) as well as the redundant anterior leaflet with 3 accessory openings. (b) The 4 chamber echocardiographic image shows the same features and the greater size of the atrialized portion of the right ventricle with significant reduction of the functional portion.

Pathophysiology in the neonate

The pathophysiology and clinical presentation vary depending on the anatomical severity of the disorder. At its extreme end (types C and D), there is severe displacement of the tricuspid valve leading to an ineffective RV and severe valve regurgitation. This results in severe cardiomegaly with consequent lung hypoplasia and cyanosis as most of the systemic venous return in shunted across the atrial septal defect. Persistent elevation in pulmonary vascular resistance (PVR) is a major impediment ot successful antegrade ejection from the smaller and less effective RV. The pulmonary blood flow is hence dependent upon the patent ductus arteriosus as there is no effective flow generated by the small RV ("physiologic pulmonary atresia"). Often there can be true right ventricular outflow tract obstruction ("anatomical pulmonary atresia"). The left ventricle is often pancaked by the enlarged RV. When the disease is less severe (types A and B), the RV can establish effective antegrade flow as PVR decreases and this is accompanied by clinical improvement in symptoms. Neonates with severe tricuspid regurgitation or gross cardiomegaly who are otherwise asymptomatic have an associated mortality of 45% within the first year of life without intervention [22, 25]. The natural history of EA during infancy is thus gloomy. However, those who survive early childhood can expect reasonable longevity. When the disease is mild, symptoms are related to exercise intolerance from progressive tricuspid regurgitation later in adult life.

Transthoracic color doppler echocardiogaphy

Echocardiography remains the mainstay in the diagnosis of patients with Ebstein's anomaly and guides management decisions regarding surgical strategy. Each patient with the Ebstein anomaly should undergo a comprehensive transthoracic echocardiogram that allows evaluation of the right atrial size, right ventricular size, and function, the accurate anatomy of the tricuspid valve, the right ventricular outflow tract, the pulmonary valve, the atrial and ventricular septum, and left ventricle. This evaluation is crucial for decision-making before surgical repair.[26] Table 3 summarizes the important details elucidated by echocardiogram that need to be evaluated in patients with Ebstein's anomaly.

Table 3: Echocardiographic evaluation in Ebstein anomaly.

Tricuspid valve anatomy and function.	• Inferior displacement of septal and posterior/inferior leaflets • Attachments/tethering of leaflets • Rotation of the tricuspid valve orifice toward the right ventricular outflow tract. • Coaptation point of TV leaflets • TV function – stenosis and insufficiency

	• Muscularization of leaflets
Right ventricle	• Size of atrialized RV • Functional RV size • Abnormal appearing RV myocardium • RV function (2D wall motion, tissue Doppler measurements, TR gradient, 3D measures) • Abnormalities of right ventricular outflow tract
Pulmonary valve	• Pulmonary valve morphology • Pulmonary atresia (functional or anatomy) • Insufficiency • Pulmonary stenosis • Supra-valvar pulmonic stenosis
Right atrium	• Atrial septal defect • Right atrial size
Left ventricle	• Compression/abnormal geometry • LV diastolic dysfunction • Abnormal septal wall motion • Left ventricular non-compaction
Associated lesions	• Ventricular septal defect, ASD or PFO • Congenitally corrected TGA • AV canal defect • Pulmonary stenosis/atresia (20-25%) • PDA • TOF • DORV • Right sided aortic arora, COA, • MV prolapse and stenosis • WPW syndrome • Down, Marfan, Noonan syndromes
LV, left ventricle; RV, right ventricle; TGA, transposition of the great arteries; TV, tricuspid valve.	

The most sensitive and specific echocardiographic finding to diagnose Ebstein's anomaly is the apical displacement of the septal leaflet of the tricuspid valve. This can be best seen in apical four-chamber views by echocardiography. When indexed to the body surface area, the distance between the hinge point of the septal leaflet of the tricuspid valve and the anterior leaflet of the mitral valve is called the displacement index. A displacement index above 8 mm/m2 is considered diagnostic of Ebstein's anomaly.[26]

Prognosis and Outcome

Celemajer et al[22] reviewed the presentation and outcome of 50 patients with neonatal Ebstein's anomaly seen from 1961 to 1990. The majority (88%) presented in the 1st 3 days of life; cyanosis (80%) was the most common presenting feature. Associated defects, present in 27 infants (54%), included pulmonary stenosis in 11 and atresia in 7. Nine patients (18%) died in the neonatal period; there were 15 late deaths (due to hemodynamic deterioration in 9, sudden death in 5 and a noncardiac cause in 1) at a mean age of 4.5 years (range 4 months to 19 years). Actuarial survival at 10 years was 61%.[22]

According to the GOSE criteria, cardiac death occurred in 0 of 4 infants with grade 1, 1 (10%) of 16 with grade 2, 4 (44%) of 9 with grade 3 and 5 (100%) of 5 with grade 4. In a multivariate analysis of clinical and investigational features at presentation, echocardiographic grade of severity was the best independent predictor of death. Augmented cardiothoracic ratio, presence of associated defects and increasing severity of GOSE score was significantly associated with death in a univariate analysis model.[22]

Similarly Lee et al[27] reviewed 20 consecutive neonates (10 male, 10 female) who ranged from 0 to 6 days old on admission. The authors concluded that 1) No identifiable risk factors reliably predict mortality in this population with complex congenital heart disease; 2) The presence of associated cardiac lesions and severe tricuspid valve displacement are possible predictors of the need for surgical palliation in the neonatal period; 3) Early mortality from neonatal Ebsteins Anomaly has dramatically improved int the current era.

Neonates with Ebstein's anomaly have a high early mortality rate and those surviving the 1st month of life remain at high risk of late hemodynamic deterioration or sudden death.

CONCLUSION

The natural history of neonatal Ebsteins Anomaly is extremely variable, but symptomatic presentation in the neonatal period has been associated with a high mortality rate of ≈50%.[27] Prior studies suggest that outcome in neonates may be related to the degree of tricuspid valve

displacement, severity of tricuspid regurgitation, cardiothoracic ratio, associated cardiac defects and oxygen saturation. Management of neonates with Ebstein's anomaly may therefore be based on the knowledge of echocardiographic grade and the presence or absence of associated defects. Most neonates with grade 1 or 2 disease and no associated defects will survive the neonatal period with supportive treatment only and have a good prognosis; those with associated defects may require surgery for these and may also expect a good outcome . Neonates with grade 3 or 4 disease have a much worse outlook; many die in early life, and tricuspid valve surgery may not alter the poor prognosis. Those who do survive the 1st month of life must have careful clinical follow-up, with particular attention to left ventricular function and cardiac rhythm.

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