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# EBSTEIN'S ANOMALY AND ITS MORPHOLOGICAL FORMS, ANTENATAL HISTORY AND GENETIC FACTORS IN THE EVENT OF ANOMALOUS SITUATION

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**Abstract**
Ebstein's anomaly is a rare malformation of the tricuspid valve, accounting for <1% of congenital heart disease. Despite its low incidence, it can have significant clinical ramifications as it encompasses anomalies of the tricuspid valve apparatus, the right ventricle, and the conduction system. Depending on the severity of these anomalies, clinical presentation can vary across a spectrum from no symptoms to critical illness at birth. Mortality remains relatively high, especially in prenatally diagnosed severe cases. The anomaly is complicated by its association with chromosomal disorders and extracardiac defects, seen in about 20% of cases.

**Keywords**: Ebstein's anomaly, congenital heart disease (CHD), endocardial cushion tissues, tricuspid valve, tricuspid regurgitation, right ventricular obstruction.

## Introduction

Postnatal management of moderate to severe disease requires considerable multidisciplinary effort, targeting multiple aspects of postnatal pathophysiology. This article reviews the morphology, clinical features, hallmarks of fetal imaging, as well as principles of postnatal management of this significant cardiac lesion.

Ebstein's anomaly is a rare malformation of the tricuspid valve first described in 1866 by Wilhelm Ebstein, a Polish-German physician, but it was not acknowledged until the 1950s when it was formally recognized as "Ebstein anomaly" or "Ebstein malformation". Ebstein anomaly involves downward displacement of the septal and posterior leaflets of the tricuspid valve toward the right ventricular apex. It has an estimated prevalence of 1–5 in 200,000 with no gender predilection and accounts for <1 % of congenital heart disease (CHD). Its prevalence, however, is higher in prenatal series accounting for 3–7% of congenital heart disease in the fetus. The latter may be a result of improved prenatal diagnosis rates with improved four-chamber cardiac views on screening prenatal ultrasound. Even though it was initially described as a disorder of the tricuspid valve, Ebstein anomaly encompasses characteristic anomalies of the tricuspid valve apparatus and the



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right ventricle, as well as abnormalities of the conduction system. Some variants of Ebstein anomaly also involve abnormalities of the left ventricular myocardium, mainly in the form of non-compaction. Clinical presentation varies across a spectrum from critical illness at birth to asymptomatic status until adulthood, depending on the severity of tricuspid dysplasia, tricuspid regurgitation, and ventricular abnormality. Mortality remains relatively high with estimated mortality rates of 29% at 1 year, 34% at 10 years, and 56% at 30 years. Improved mortality rates at 1 year, down to 18%, have been reported in a contemporary single-center study. Prenatally diagnosed Ebstein's anomaly appears to have particularly high adverse outcome rates with the majority of mortality events occurring in utero and the neonatal period. Perinatal mortality as high as 45% has been reported in recent multicenter studies.

In the normal heart, the tricuspid valve is made of three leaflets: anterior, septal, and posterior (inferior or mural). It also inserts slightly more apically compared to the anterior mitral valve. Embryologically, the leaflets of the tricuspid valve develop from both the endocardial cushion tissues and the myocardium during the early weeks of cardiac embryogenesis. A process of delamination that occurs between 8 and 16 weeks of gestation allows the leaflets to separate from the underlying myocardium and results in the freely mobile leaflets of the tricuspid valve. In Ebstein's anomaly, the process of delamination is incomplete giving the impression of the leaflets being "plastered" to the right ventricular myocardium. This primarily affects the septal leaflet, to a lesser extent, the posterior leaflet, and even less the anterior leaflet. This results in the pathologic apical displacement of the two leaflets from the tricuspid valve annulus, and hence the hinge point or orifice of the valve becomes displaced to the junction of the inlet and trabecular portions of the right ventricle. Consequently, the proximal portion of the right ventricle becomes atrialized and the functional right ventricle becomes diminished. Anatomically, Ebstein's anomaly involves several features including (1) failure of delamination of mainly the septal and posterior leaflets from the myocardium, (2) apical displacement of the valve orifice to the junction of the inlet and trabecular portions of the right ventricle, (3) dilatation of the atrialized portion of the right ventricle, (4) tethering and redundancy of the anterior valve leaflet, and (5) dilatation of the tricuspid valve annulus.

Ebstein's anomaly has a wide spectrum of severity depending on the degree of displacement of the septal and posterior leaflets or of the functional annulus of the tricuspid valve. On one end of the spectrum, the septal leaflet is made of only a ridge of fibrous tissue, the posterior leaflet is plastered to the wall, and the anterior leaflet is significantly deformed, resulting in severe tricuspid regurgitation. On the other end, in mild forms of the disease with mild valvular regurgitation, only the septal leaflet is mildly displaced. The anterior leaflet of the tricuspid valve is not displaced, but it may be deformed, forming a large redundant saillike intracavitary curtain that could cause right ventricular obstruction. The anterior leaflet can also have fenestrations that can allow blood flow in addition to the true orifice of the valve.

In addition to the abnormality of the tricuspid valve itself, some reports highlight that Ebstein's anomaly also involves a cardiomyopathy of the right ventricle that further explains the dilatation of the right ventricle. Reports studying the histopathology of the right ventricular myocardium demonstrated that the right ventricular dilation seen in Ebstein's anomaly is associated not only with thinning of the myocardial wall, but is also related to an absolute decrease in the number of



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myocardial fibers counted through the thickness of the wall from the endocardium to the epicardium.

Unlike Ebstein's anomaly, in tricuspid valve dysplasia, there is no displacement of the leaflets toward the apex despite the valve leaflets being abnormal. This can also present as a range of severity from mildly dysplastic thickened tricuspid valve leaflets with normal chordae and papillary muscles to severe forms involving near-agenesis of the leaflets and the subvalvar structures.

Despite the observation that most cases of Ebstein's anomaly occur sporadically, it is important to obtain a detailed antenatal history from the parents, including family history of congenital heart disease but also specifically history of atrial septal defect (ASD) or a persistent interatrial communication, as this is one of the most commonly associated lesions, present in ~80–94% of cases. Family history of arrhythmia is to be elicited as well as supraventricular tachycardia related to Wolff-Parkinson-White (WPW) syndrome as this is more commonly associated with this lesion than any other form of CHD.

The vast majority of cases of Ebstein's anomaly occur in isolation with no syndromic association. However, 20% are associated with chromosomal disorders with extracardiac defects including Apert, CHARGE, Cornelia de Lange, Holt-Oram, Kabuki syndrome, Noonan syndrome, and VACTERL. Syndromic diagnoses have been associated with increased morbidity and mortality in CHD in general; a recent multicenter study showed no association between genetic abnormality or syndrome and Ebstein anomaly perinatal mortality. Non-syndromic Ebstein's anomaly has an autosomal dominant inheritance with incomplete penetrance and variable expressivity. Several genes have been associated with the non-syndromic form (OMIM disease 224,700), including NKX2.5, MYH7, and TPM1. Given the autosomal dominant inheritance pattern of the non-syndromic form, it would be prudent to screen first-degree relatives of affected individuals. Some studies have implicated the gene located on the long arm of chromosome 15 (15q) which affects the early morphogenesis of cardiac structures, including the normal formation of the tricuspid valve. In addition, some reports involving rearrangements of the chromosomal region 11q arm in two patients with Ebstein's anomaly were also reported including an interstitial deletion of chromosome 11 [46,XY,del(11) (11q21q23)] and a tertiary trisomy of chromosome 11qter [47,XX,þder(22)t(11;22)(q23; q11.2)].

Teratogenic exposure is an integral component when obtaining antenatal history for any congenital malformation, including Ebstein's anomaly. Teratogens are factors that can affect fetal growth and development and can alter the structure or function of different organs as well as effects that extend to postnatal development. These include environmental exposures, maternal medical disorders, infectious agents, and genetic conditions. Lithium exposure, particularly in the first trimester, was associated with increase in risk of Ebstein in earlier studies, leading to recommendations on avoiding this medication during pregnancy. This increase does not appear to be significant in more recent studies. Another potential teratogen linked to Ebstein anomaly is benzodiazepines. Hence, the use of these substances should be particularly screened for in Ebstein cases.

In conclusion, Ebstein anomaly continues to be associated with high mortality despite earlier detection. Various fetal and neonatal scoring systems can help with counseling patients in this complex lesion. Delivery at a tertiary care center is essential in order to prepare for a



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multidisciplinary management in the neonatal period. Some cases improve with the changes in pulmonary vascular resistance and transition to the postnatal circulation.

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