21 Ebstein Anomaly

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Ebstein anomaly is a very rare congenital heart lesion representing <1% of all CHD. It is a complex condition involving failure of tricuspid valve leaflet delamination during embryogenesis. It represents a broad continuum of morphologic derangements but the central defining feature is apical displacement of the septal leaflet of the tricuspid valve, usually associated with severe TR. Even though the septal leaflet tends to be the most involved, usually all 3 leaflets are involved to a certain extent, with the anterior leaflet being the least involved. The leaflets tend to be dysplastic, muscularized, and tethered to short chords and papillary muscles that may insert directly into the leaflets. The valve is spirally rotated with the septal and posterior leaflets displaced inferiorly into the RV. This in turn displaces the functional annulus of the valve downward, leaving a variable portion of RV on the atrial side of the valve leaflets ("atrialized" portion of the RV). The atrialized portion of the RV tends to be thin and dyskinetic, and is usually significantly dilated due to the degree of TR. This also leads to significant dilation of the true annulus of the tricuspid valve.

Pathophysiology and Clinical Presentation

In Ebstein anomaly, the degree of prograde flow across the RV is limited due to the severe TR and variable impairment of RV function. In addition, the atrialized portion of the RV tends to distend during atrial contraction, further limiting prograde flow. All of these structural abnormalities lead to massive right-heart dilation. Massive right-heart dilation can lead to in utero underdevelopment of the lungs due to lack of physical space. Patients with Ebstein anomaly will usually have an ASD that tends to shunt right to left, causing variable degrees of cyanosis.

The clinical presentation varies significantly depending on the degree of TR, degree of RA dilation, extent of atrialization of the RV, RV function, and degree of pulmonary hypoplasia. It can vary from cardiogenic shock in the neonate to an asymptomatic presentation in adulthood.

In neonates, the high PVR and the presence of a ductus arteriosus lead to a higher afterload of the RV, further limiting prograde flow. As such, neonates tend to present with variable degrees of cyanosis and heart failure.

Patients with less severe forms of the disease, or those who survive the neonatal period without intervention, may present later in life with cyanosis, decreased exercise tolerance, dyspnea on exertion, or palpitations due to arrhythmias. A systolic murmur in the left sternal border is usually heard, and there is wide splitting of the first and second heart sounds. There may be IJ distention.

Children and adults with Ebstein anomaly are at significant risk for atrial tachyarrhythmias (atrial ectopic tachycardia and atrial flutter) due to RA dilation. In addition, 15-20% of patients have accessory pathways along the tricuspid valve annulus that can lead to reentrant supraventricular tachycardia.

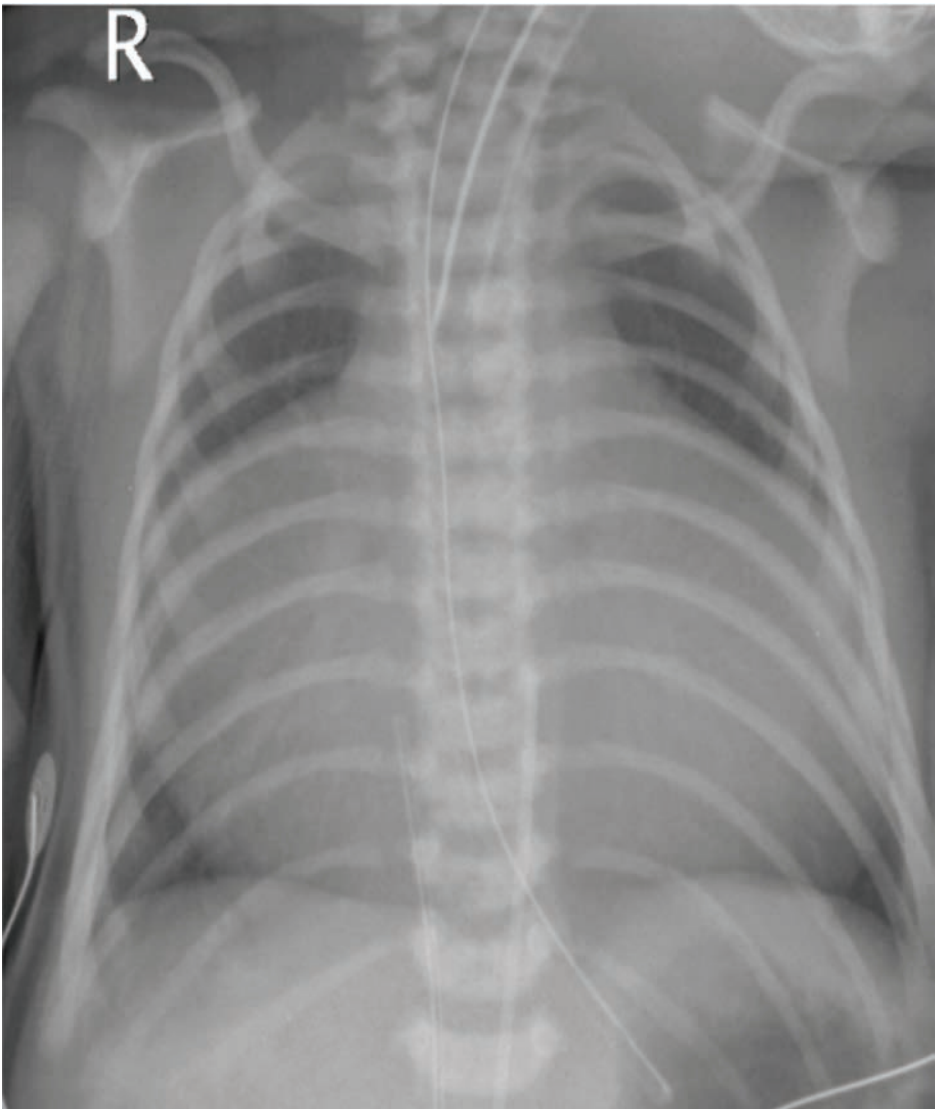

Figure 21-1. CXR in an infant with Ebstein anomaly showing massive cardiomegaly due to severe RA dilation.

Diagnosis

- **Fetal echocardiography.** The diagnosis of Ebstein anomaly is now frequently made in fetal life by identifying apical displacement of the tricuspid valve, severe TR, an atrialized RV, and RA dilation.
- **ECG.** RA enlargement and diminished RV forces. It may also show ventricular preexcitation (Delta wave) consistent with Wolff-Parkinson-White syndrome as accessory pathways are common in this disease.

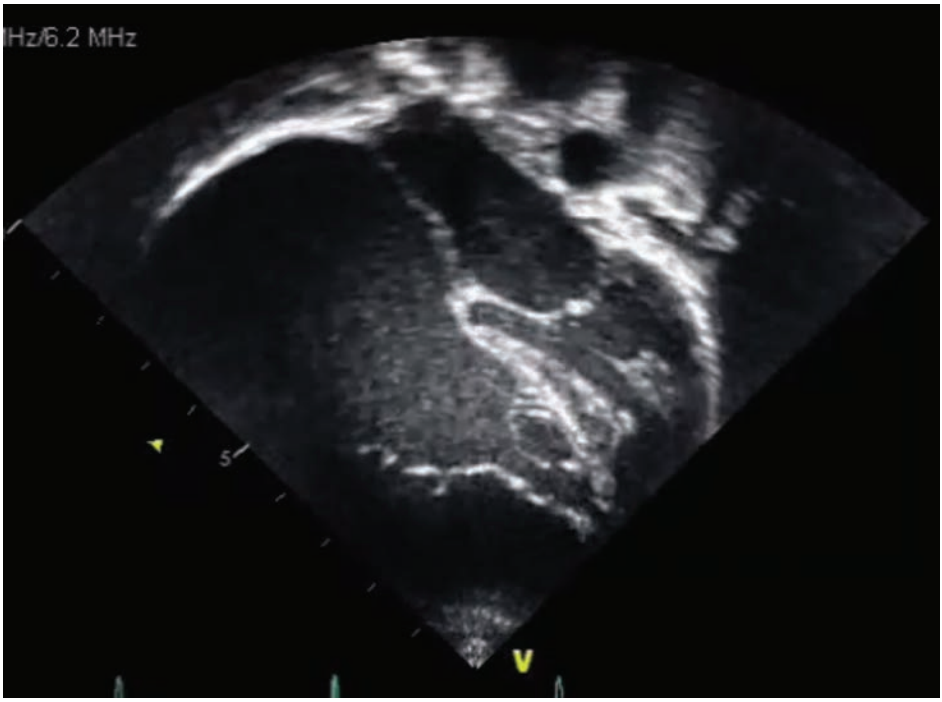

Figure 21-2. 4-chamber echocardiographic view showing significant apical displacement of the septal leaflet of the tricuspid valve compared to the mitral valve insertion in a patient with Ebstein anomaly. The image also displays RA enlargement, atrialization of the RV, and its relative hypoplasia. Image courtesy of Dr. Josh Kailin, www.pedecho.org.

- **CXR (Figure 21-1).** Severe cardiomegaly due to the severe TR and RA dilation is the pathognomonic finding on Ebstein anomaly. There are 3 cardiac lesions that present with such a significant degree of cardiomegaly: Ebstein anomaly, dilated cardiomyopathy, and a large pericardial effusion. The amount of visible lung may be significantly limited due to the degree of cardiomegaly and pulmonary hypoplasia.
- **Echocardiography.** Main diagnostic modality. The formal definition of Ebstein anomaly is apical displacement of the septal leaflet of the tricuspid valve >8 mm/m^2 (Figure 21-2). The displacement is measured compared to the level of attachment of the mitral valve annulus and indexed to BSA. It is important to define the degree of TR and the extent of atrialization of the RV (Figure 21-3). In neonates, it is imperative to evaluate the pulmonary valve for patency, since a closed pulmonary valve may be *anatomically* atretic or *functionally* atretic (Figure 21-4). Pulmonary valve patency may be predicted by observing PI but the lack of PI does not necessarily indicate anatomic pulmonary atresia. In newborns with a large PDA, there may not be prograde flow across the RVOT due to severe TR (retrograde flow) and elevated PVR.
- **Cardiac MRI.** It has become a useful adjunct in the management of older patients in recent years. It allows further assessment of RV and LV size and function.

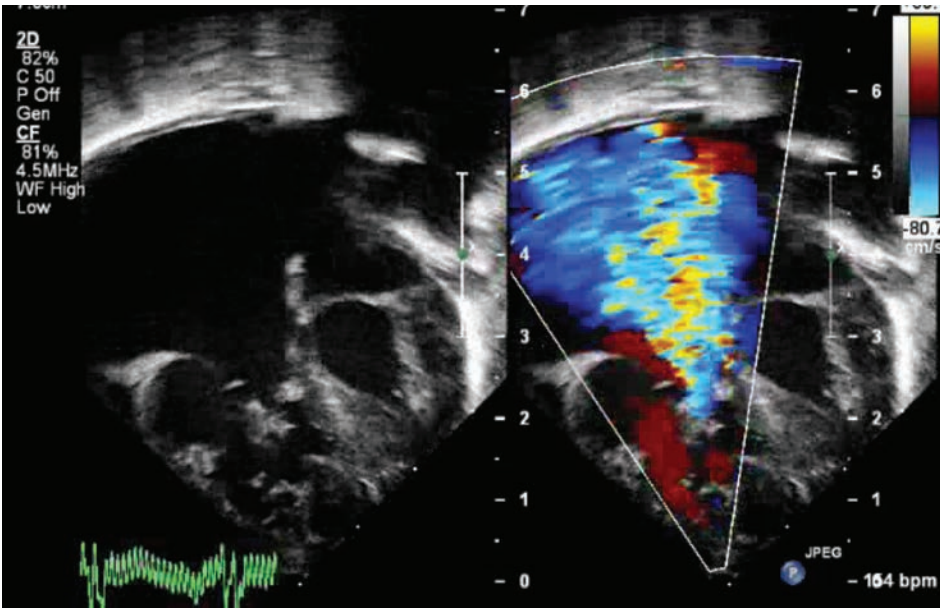

Figure 21-3. Side-by-side 4-chamber color-compare views of Ebstein anomaly with significant apical displacement of the septal leaflet of the tricuspid valve and severe TR. Image courtesy of Dr. Josh Kailin, www.pedecho.org.

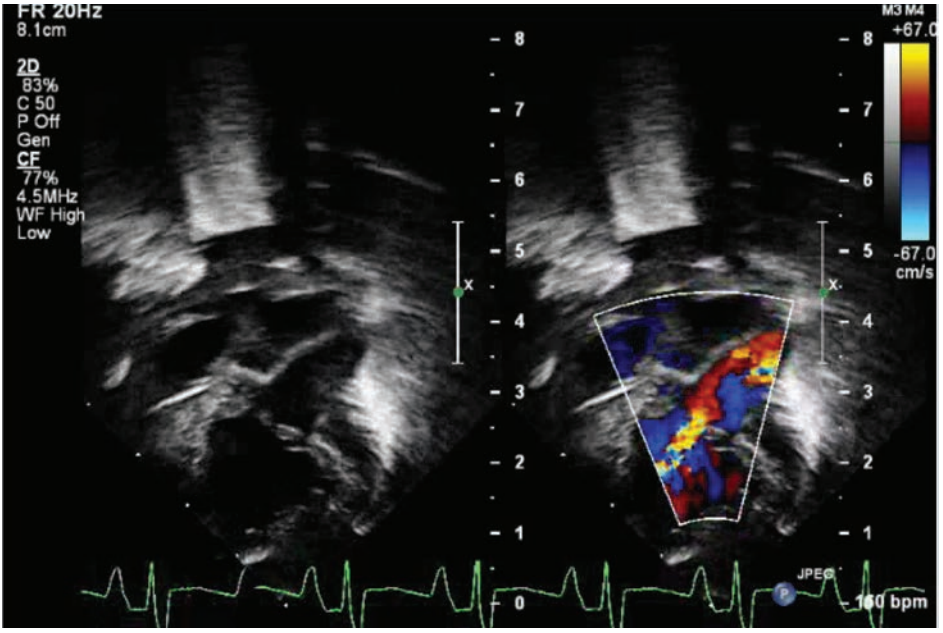

Figure 21-4. Side-by-side subcostal color-compare views of flow through the PDA and then retrograde through the pulmonary valve. This patient had functional pulmonary valve atresia due to the lack of prograde flow across the pulmonary valve, but not anatomic or true atresia, as noted by the presence of PI.

Preoperative Management

Initial treatment of critical newborns with *functional* pulmonary valve atresia is aimed at decreasing PVR with ventilation, oxygenation, and iNO. Patients require sedation and possibly neuromuscular blockade to further decrease PVR. Afterload reduction can be added if the BP is adequate to promote systemic blood flow. In patients with poor cardiac output, inotropes may need to be initiated with the known increased risk of tachyarrhythmias in these patients. In such individuals (who typically have been started on PGE at birth in the setting of a fetal diagnosis), the decision to attempt PGE weaning can be very challenging. The key question is whether the RV can generate enough force to overcome the PVR and manage the degree of TR. In many patients, it is very important for the management team to be persistent in these efforts: many patients can ultimately be weaned from PGE after failing initial attempts (and thereby avoid newborn surgery).

A unique situation in neonates occurs in the presence of significant PI. The presence of a PDA in this setting allows blood to flow through the PDA, retrograde through the incompetent pulmonary valve, then retrograde through the insufficient tricuspid valve to the RA and across the ASD (right-to-left shunting), through the left heart out to the aorta, and the back through the PDA. This "circular shunt" (Figure 21-5) can compromise both systemic and pulmonary blood flow. In these profoundly cyanotic patients, starting prostaglandins may be detrimental. The same circular shunt can occur if the pulmonary valve is atretic and is ballooned open, leading to PI.

Care of a neonate with severe Ebstein anomaly can also be complicated by lung hypoplasia due to limited in utero development. Inability to ventilate due to severe lung hypoplasia can impact survivability in these patients. Decisions to proceed with surgical intervention or possibly extracorporeal support need to take into consideration the degree of pulmonary hypoplasia. In these challenging situations, CT of the lungs may be helpful in delineating the degree of parenchymal immaturity.

Indications / Timing for Intervention

In the neonatal period, initial evaluation of oxygen saturations and cardiac output determine if any intervention is needed. Most children with Ebstein anomaly will ultimately achieve a balanced circulation with adequate systemic oxygen saturation. As per above, persistent attempts at medical management are warranted unless the infant is critically unstable. If neonates present with high PVR, a circular shunt, and compromised cardiac output, interventions are aimed at ventilating and decreasing PVR to reverse the retrograde flow through the pulmonary valve. The decision to intervene is often based on response to these therapies.

In patients with true (*anatomic*) pulmonary atresia, there will be a need to establish a source of pulmonary blood flow. This is best accomplished after the PVR has dropped, and can be in the form of a modified Blalock-Taussig-Thomas shunt (mBTTS) or a ductal stent (see Chapter 38) with or without dilation of the pulmonary valve depending on echocardiographic size. In neonates with a diminutive RV, consideration should be made of a Starnes procedure (see below). If there is a sizeable RV but the child cannot maintain a good cardiac output due to the amount of TR, consideration can be made

Figure 21-5. Circular shunt in Ebstein anomaly in the presence of PI and a PDA. Blood flows from the aorta, through the PDA, and then retrograde through the pulmonary valve, the tricuspid valve (severe TR), and right to left through the ASD, just to go back into the aorta. Minimal flow goes to the aorta to provide systemic output and to the branch PAs to oxygenate the blood. This results in both cyanosis and poor cardiac output.

of attempting to repair the tricuspid valve. Tricuspid valve repair is also a consideration in older children with heart failure symptomatology due to severe TR (see below).

Anesthetic Considerations

Anesthetic considerations should reflect careful assessment of the patient's current status and ongoing management. A controlled, careful induction with IV narcotic and benzodiazepine can be supplemented with anesthetic vapor to minimize rapid changes in physiology. iNO, inotropes, and vasopressors should be available if not already initiated. Given the proclivity of children with Ebstein anomaly to develop tachyarrhythmias, this is one situation for which defibrillation pads may be helpful even with a first-time sternotomy.

In critical newborns presenting with low cardiac output and marginal systemic arterial oxygen saturations, anesthetic management is focused on optimizing ventilation and maintaining cardiac output. These babies may be very unstable during transport and anesthesia induction. As such, it is critical to have all members of the surgical and OR team in attendance at induction.

Surgical Intervention

Timing of surgical intervention and mode of operative correction has evolved significantly over the past 2 decades as surgical techniques have improved. The primary goal of surgery for Ebstein anomaly is to improve right-heart effectiveness in generating prograde PA flow through tricuspid valve repair, reduction/elimination of nonfunctional portions of the RV (atrialized portion), partial or complete ASD closure, and where necessary, pulmonary valve replacement.

As discussed previously, newborns with symptomatic Ebstein present a challenging surgical problem and historically, operations on these babies have been associated

with significant risk of mortality. The primary decision in newborns is whether or not to attempt tricuspid valve repair. In those individuals in whom the valve is deemed irreparable, RV exclusion through patch closure of the tricuspid orifice ("Starnes" operation) may be the only viable option. When this is done, it is imperative to fenestrate the patch such that the RV is able to decompress into the RA. The operation also includes an atrial septectomy and creation of an mBTTS.

Several investigators have documented encouraging results with tricuspid valve repairs in newborns, but these results are not widely consistent in the greater congenital heart surgery community. In repairing the tricuspid valve in small babies, the operation is made all the more challenging by the very delicate nature of tricuspid valve tissue in these children. As such, it has been our approach that if possible, interim palliation is offered such that tricuspid repair is offered later in childhood when the valve tissue is more substantial.

In older children and adults, the "Cone" operation as initially described and implemented by Da Silva has revolutionized surgical repair of Ebstein anomaly. The core principle of the Cone operation is to mobilize all available tricuspid valve tissue (including the portion that is not fully delaminated from the RV free wall) from its abnormal attachements, leaving only the apical support intact. The valve is separated from the abnormal tricuspid annulus, rotated in a clockwise direction and then a "cone" of valve tissue is created by suturing all the valve tissue together. The conceptual understanding of this operation is greatly facilitated by thinking of the reconstructed cone like a sort of long Hemlich valve. Once the cone is reconstructed, the atrialized portion of the RV is plicated longitudinally to create a more efficient RV cavity and to construct a new tricuspid annulus that is normally positioned. The cone is then reattached at the level of the new annulus with great care to avoid the AV node. Most surgeons then add a formal annuloplasty ring. Following reconstruction of the tricuspid valve, the ASD is either partially or completely closed, depending on the effectiveness of the reconstruction and the size of the tricuspid inlet. In patients where we are concerned about the repair, we will typically leave a fenestration in the atrial septum (4-5 mm). If there is concern that the tricuspid orifice is too small, we will add a bidirectional Glenn shunt to augment pulmonary blood flow and offload the RA.

In desperate situations where there has been a failed attempt at tricuspid repair, it may be necessary to replace the tricuspid valve. Of course, this is a problematic solution in that all available tissue prostheses will eventually degenerate and have to be replaced. This degeneration is notoriously more rapid in children. It is also critical to make every effort to prevent surgical AV block in tricuspid valve replacements and in patients where there is deficient tissue in the region of the true annulus near the AV node. As such, the prosthesis is placed well up into the body of the RA, leaving the coronary sinus below the sewing ring of the prosthesis. In recent years, with the advent of transcatheter valve replacement alternatives, the option of "valve-in-valve" replacement of degenerated prostheses may be considered.

Postoperative Management

Postoperative management is dependent on the intervention undertaken. In neonates, much of the postoperative management involves dealing with the ramifications of preoperative instability, elevated PVR, and pulmonary hypoplasia/ventilator concerns. Sedation is imperative to managing these critically ill neonates. At times, they may also require neuromuscular blockade in the initial postoperative days. In neonates who undergo an mBTTS or ductal stent, management is aimed at balancing Qp:Qs, which can be fluid as PVR drops (see Chapter 38). In those that undergo a Starnes procedure, in addition to managing Qp:Qs, there are the usual considerations involved in managing a neonate who has undergone CPB and cross-clamping, with the IV fluid, inotrope, and inflammatory considerations that this entails. Patients who undergo a tricuspid valve repair in the neonatal period will have similar considerations and will benefit from further attempts to decrease PVR and maintain cardiac output. Consideration of the patient's risk of tachyarrhythmias is important as many children have accessory pathways and irritable atria due to dilation and suture lines that lead to a higher risk of this complication. Inotropes may need to be tailored to avoid increasing arrhythmia burden by decreasing adrenergic agents.

Patients with Ebstein anomaly initially diagnosed at TCH (1995-2017)

Number of patients: 215

- Neonates: 87 (40%)
- Infants: 23 (11%)
- Children: 69 (32%)
- Adults: 36 (17%)

Of the 87 neonates diagnosed, only 15 (17%) required surgery in the neonatal period (13 with anatomical pulmonary atresia). Of these, 9 underwent single ventricle palliation and 6 underwent biventricular repair.

For older patients, early extubation (ideally intraoperative) following repair mitigates the deleterious effects of positive-pressure ventilation. Inotropic support should be tailored to reduce PVR and minimize the tendency for atrial tachycardia.

Long-Term Management

Patients can remain asymptomatic if the degree of TR is mild and there is limited atrialization of the RV. Tachyarrhthymias can be the presenting sign leading to diagnosis in older children with less hemodynamically significant lesions.

All patients with Ebstein anomaly should be assessed for the presence of an accessory AV conduction pathway and it is preferred that such pathways be ablated in the electrophysiology lab prior to surgical intervention. In those rare individuals with persistent accessory pathways after attempted catheter ablation, surgical division of the pathway should be performed at the time of the tricuspid valve repair operation. In addition to tachyarrhthmias, patients with Ebstein anomaly can have progressive heart failure symptomatology due to the degree of TR. In addition to right-sided heart failure symptomatology, they can also develop LV failure due to poor interventricular interactions and septal shift secondary to right-sided volume overload. Much of the long-term care in patients with Ebstein anomaly is determined by their initial course and required surgical interventions.