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What is Ebstein's Malformation

Ebstein's malformation is a very rare congenital defect that can cause cyanosis at birth. Normally approximately 1% of all live births have some type of congenital heart defect most of which are relatively benign. Of that 1%, slightly less than 1% are known to have Ebstein's malformation. Ebstein's malformation can have wide variety of presentation depending on the degree of tricuspid valve abnormality.

Ebstein's malformation is failure of the tricuspid valve to separate from the walls of the right ventricle. The variety ranges from slight displacement of the tricuspid valve which are completely asymptomatic to severe displacement which have cyanosis and heart failure. Normally the tricuspid valve tissue separates from the wall of the heart, also known as delamination, typically around the fifth to sixth week of development. While Ebstein's malformation is right-sided problems with the heart, the left side of the heart can be affected as well. This can be with either mitral valve abnormalities or contractility abnormalities of the left ventricle. When the tricuspid valve is malformed in this fashion, it causes the right ventricle to become enlarged and the muscle walls to thin out and not squeeze well. There are varying degrees of leakage of the tricuspid valve. This combination of leaking tricuspid valve and atrialized right ventricle leads to significant enlargement of the true right atrium. Although this is a rare finding after a baby is born, it is the most common cause of fetal demise from a cardiac standpoint.

We are usually able to diagnose this before birth with a fetal echocardiogram. In general, if the prenatal diagnosis shows that the size of the right atrium and atrialized right ventricle is larger than the left atrium, left ventricle and true right ventricle combined it is usually felt to cause fetal demise. This severe form of Ebstein's malformation causes the heart to be very large and does not allow room for the lungs to develop. Ebstein's is also associated with an atrial septal defect, pulmonic stenosis or pulmonary atresia. The main problem with the circulation in Ebstein's malformation is decreased blood flow through the right ventricle. This decreases the supply of blood to the lungs causing the baby to be blue due to lack of oxygen in the blood.

On physical exam, these babies typically are blue and have a murmur. The diagnosis is confirmed by an echocardiogram. As the baby gets older resistance to blood flow through the lungs drops, and the babies will become pink. When they do not become pink, treatment such as ventilation, the use of Prostaglandin, and the use of Sildenafil can be helpful in allowing time for the lungs to improve.

Once out of the newborn period, the most common problems associated with Ebstein's malformation is fatigue and irregular heart beats. These patients tend to have a rather high incidence of various electrical problems of the heart, such as Wolff-Parkinson-White syndrome, premature ventricular beats, premature atrial beats, atrial flutter and fibrillation. Atrial flutter/fibrillation and paroxysmal atrial tachycardia is usually treated with Digoxin. The definitive treatment is an electrophysiologic (EP) study where pathways that create these abnormal heart beats can be interrupted by using radio frequency ablation. Indications for surgical repair are increasing fatigue and increasing enlargement of the heart.

There are several ways to surgically treat Ebstein's abnormality. In the newborn period, when there is severe abnormality of the tricuspid valve and the patient remains blue despite aggressive medical and ventilatory support, transplantation is offered as treatment. However, transplantation has only been mildly successful because the lungs are small. Once outside the infant period, options to repair the valve increase. For many years the treatment of choice has been to replace the tricuspid valve and reduce the size of the atrialized portion of the right ventricle. In more recent years a technique called the cone procedure has been used. For this procedure they create a funnel-type structure with the tricuspid valve that will help minimize the amount of leakage. The atrial septal defect is closed and the extra tissue of the right atrium is either removed or folded in so that the size of the right atrium is decreased. Also during surgical repair they attempt to remove as many of the abnormal electrical pathways as possible .

The ultimate outcome of patients with Ebstein's malformation depends primarily on how well the tricuspid valve functions initially or after surgical repair. Outcome also depends upon the presence of irregular heart beats. The most frequent cause of death in these children in the newborn period is heart failure.In infancy and beyond it is ventricular dsyrhythemias. With medical treatment and electrophysiologic intervention many times these events can be avoided.

If you have any questions, please ask one of our doctors.