Ebstein Anomaly

What is it?

Ebstein anomaly is a birth defect of the tricuspid valve. The **tricuspid valve** separates the right upper chamber from the right lower chamber of the heart. Normally, the tricuspid valve has three even leaflets that prevent the backward flow of blood. If a child has **Ebstein anomaly**, the valve sits lower than normal in the right lower chamber and the valve's leaflets are misshaped. This can cause the blood to flow backwards in the heart and the right upper chamber to become enlarged. Ebstein anomaly can range from mild to severe. If the problem is severe, congestive heart failure can occur. It is a critical congenital heart defect. **Congenital** means present at birth. **Critical congenital heart defects** can lead to serious problems and even death if left untreated.

Many babies also may have a hole between the right and left upper chambers, called a **patent foramen ovale (PFO)**. Normally, the foramen ovale closes shortly after birth. This hole is often kept open from the high pressure in the right upper chamber, allowing the oxygen-poor blood to flow from the right upper chamber to the left upper chamber. This blood skips the lungs and goes directly to the body, resulting in lower oxygen levels in the blood.

Ebstein anomaly may be seen with other heart conditions such as pulmonary valve stenosis or atresia, atrial septal defect, or ventricular septal defect. Some patients also have a condition called **Wolff-Parkinson-White syndrome**. An abnormal electrical pathway of the heart is present in this condition, which can cause an abnormally fast heart rate.

How common is it?

About 1 in every 20,000 babies are born with Ebstein anomaly.

What causes it?

The cause of Ebstein anomaly is unknown. Many factors may cause it. Studies have shown that it is more common in children whose mothers took lithium during pregnancy. Lithium may be found in some medications to steady moods. More research is need to understand the exact cause of Ebstein anomaly.

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How is it diagnosed?

Ebstein anomaly may be diagnosed during pregnancy or after. During pregnancy screenings are done to check for birth defects. After birth a doctor will do a physical examination. The doctor will see if a baby has blue-colored skin and lips, called **cyanosis**. This can be a sign of low levels of oxygen in the blood. The doctor will listen to a baby's heart and if the doctor hears a heart murmur, or a "whooshing" sound, that may be a sign of a heart defect. A doctor also might see that a baby is having trouble breathing, a pounding heart, and poor feeding, which also could be signs of a heart defect. If the doctor suspects there is a heart defect, then a diagnostic test called an **echocardiogram** should be done to check for defects in the heart. A chest x-ray also may be taken to see the size of the heart because a large heart can be a sign of Ebstein anomaly. Your baby's doctor may also want to test the rhythm of your child's heart. This test is called an **electrocardiogram (ECG)**.

How is it treated?

There are options for the treatment of Ebstein anomaly. Medications may be required to control the heart rate, and surgery might be recommended for severe cases. Your child's doctor should discuss all treatment options with you.

For more information:

American Heart Association
http://www.heart.org/HEARTORG/Conditions/CongenitalHeartDefects/AboutCongenitalHeartDefects/About-Congenital-Heart-Defects_UCM_001217_Article.jsp#.Wv2YtPnwbcs

National Heart, Lung, and Blood Institute
https://www.nhlbi.nih.gov/health-topics/congenital-heart-defects

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