

What is Ebstein's anomaly?

Ebstein's anomaly is a rare congenital heart defect affecting the tricuspid valve on the right side of the heart. It occurs in about one out of every 200,000 infants. This makes up fewer than 1% of all congenital heart defects. It affects males and females equally. It may affect babies in the womb all the way through to the elderly.

The tricuspid valve (from the Latin *tri* for three and *cuspis* for point) is located between the right atrium (top chamber of the heart) and the right ventricle (bottom chamber). Three small triangular "leaflets" make up the valve. The valve normally opens when the heart relaxes so that blood can flow forward from the right atrium into the right ventricle. The valve closes when the heart contracts so that blood cannot flow backwards.

In Ebstein's anomaly, the flaps/leaflets of the tricuspid valve have not formed correctly. They may be abnormally attached to the walls of the ventricle or may be too small. Occasionally there are actual holes in the leaflets.

Ebstein's anomaly can be mild with almost no leak to severe with a very large leak. Severity depends on three factors.

Because of these problems, the tricuspid valve may not close correctly. There may be gaps between the leaflets. This makes it "leaky" and allows blood to go backwards into the atrium when the heart contracts. This leak stretches the right atrium and also causes the right ventricle to expand as it needs to pump more blood just to maintain a normal forward flow. This makes the right side of the heart work harder and less efficiently. Over time, if the amount of "extra work" the right side has to do becomes too much, the heart may weaken and cause heart failure.

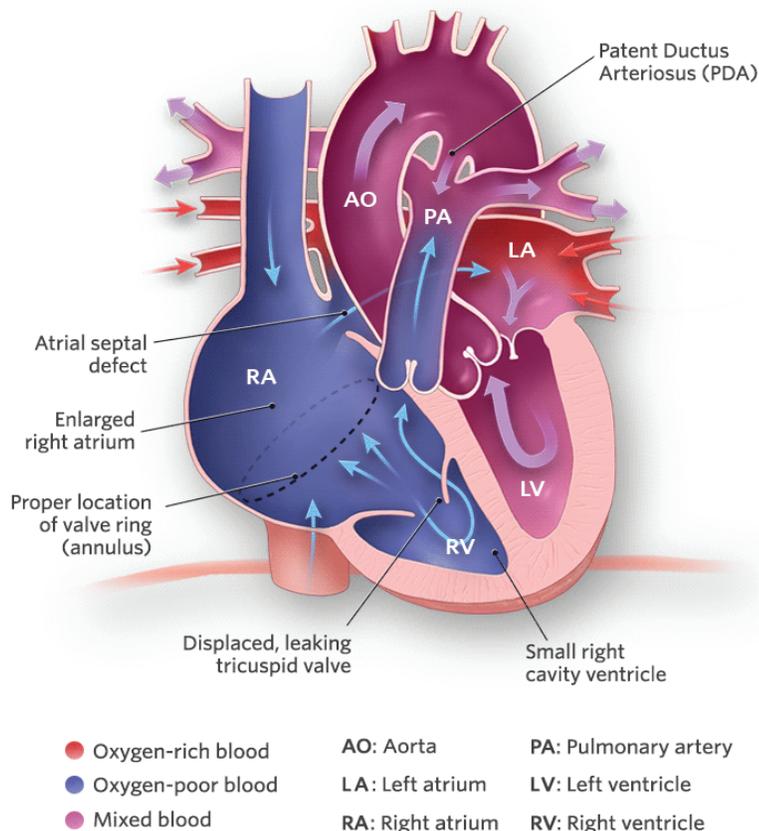
Ebstein's anomaly can be mild with almost no leak to severe with a very large leak. Severity depends on three factors. The first is how deep into the heart the valve is displaced. Second is whether there are abnormal attachments or holes in the valve. Lastly, severity is determined by how well the valve leaflets fit together when the heart contracts.

Ebstein's patients may also have a small hole between the two upper chambers of the heart. This is known as an atrial septal defect (ASD). Some patients may also have an abnormal electrical pathway in the heart. This is known as Wolff-Parkinson-White (WPW) syndrome. Other heart rhythm abnormalities, such as atrial flutter, may also occur. The presence of WPW can lead to very fast heart rates and fainting spells.

What causes Ebstein's anomaly?

The cause is not known. However, we do know there is a higher rate in twins and in people with other CHDs, as well as other genetic diseases. This includes CCTGA (congenitally corrected transposition of the great arteries). There is

Ebstein's Anomaly of the Tricuspid Valve



also an increased risk in women who took lithium during pregnancy.

What are the symptoms of Ebstein's anomaly?

Symptoms depend on how severe the defect is. They can range from none to severe. Symptoms may include shortness of breath, especially with exertion, fatigue, and heart racing and irregularity (palpitations). A person may also have dizziness, swelling of the legs and stomach, and/or a bluish discoloration of the lips and skin caused by low oxygen levels.

Medications will not stop problems from developing in Ebstein's anomaly. But if you are having symptoms, certain medications might be helpful. These include diuretics to treat fluid retention and/or medications that treat abnormal heart rhythms.

How is Ebstein's anomaly diagnosed?

Imaging is important for the diagnosis of Ebstein's anomaly. Your doctor may order an EKG, chest X-ray, or echocardiogram. These will help him/her assess how severe the defect is, how much leak of the valve there is, how enlarged the right side chambers are and if there are any abnormal heart rhythms. You might also need to have an MRI, exercise test or cardiac catheterization.

When is treatment needed?

Adults with Ebstein's anomaly should be seen by an adult congenital heart disease (ACHD) cardiologist. Treatment depends on the symptoms and the patient's age. It also depends on how severe the Ebstein's anomaly is and if there are other medical problems. Some adults may not need treatment for years. Looking at the whole patient is important.

What types of treatment are indicated?

Medications will not stop problems from developing in Ebstein's anomaly. But if you are having symptoms, certain medications might be helpful. These include diuretics to treat fluid retention and/or medications that treat abnormal heart rhythms. An ablation might also be done. Your doctor might also prescribe a blood thinner if you have atrial flutter or atrial fibrillation.

Repair or replacement of the tricuspid valve may be indicated. This might be necessary if you develop symptoms and/or if your symptoms are worsening. This could happen if the tricuspid valve leak causes enlargement or dysfunction of the right ventricle. You

may also need surgery if imaging shows that you have a decline in heart function or worsening valve leakage.

If you have an ASD, the decision to close it or not depends on how severe the tricuspid valve leak is. If repair or replacement of the tricuspid valve is necessary, closure of the ASD could be done at the same time. There are many considerations to make when deciding whether to close the ASD and how to close it.

What surgery might be performed?

It is important to find a surgeon who has expertise in performing surgery on patients with Ebstein's anomaly. The goal of surgery is to enable the leaflets to open and close correctly. The preferred surgical method is repairing the valve because it uses the patient's own tissue. This can be done if there is enough tissue present. It is very complex surgery. If the existing valve cannot be repaired, it can be replaced with a tissue valve. Mechanical valves are rarely used for replacement in Ebstein's anomaly.

Most patients who had valve repair or replacement as a child or as an adult do well. However, if the valve becomes leaky or narrowed again, repeat surgery may be needed.

In the most severe cases, if the valve is seriously deformed, heart function is poor and/or other treatments are not effective, a heart transplant may be an option.

Can I have a baby?

Women with Ebstein's anomaly should be evaluated by an ACHD cardiologist before becoming pregnant. This is to determine if there are any issues that should be treated before becoming pregnant. Most Ebstein's anomaly patients are able to tolerate pregnancy with few or no complications.

The risk of having a child with CHD increases from 1% in the general population to between 3% and 8% if you or your family have Ebstein's anomaly.

What about nutrition and exercise?

Physical activity is tolerated by most patients with Ebstein's anomaly. However, all patients should check with their ACHD cardiologist before starting an exercise program.

It is also important to eat a heart-healthy diet and maintain normal body weight. Smoking, using drugs, and/or drinking excessively should be avoided. Your doctor should check all of your cardiac risk factors, like cholesterol, blood pressure, and diabetes on a regular basis.

The course of Ebstein's anomaly can be unpredictable. Continued lifelong follow-up is needed by an ACHD specialist.

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