CONDITION

Ebstein anomaly

Ebstein anomaly is a heart condition present from birth in which the tricuspid valve is displaced and often leaks.

WHAT IT IS
Congenital displacement and leakage of the tricuspid heart valve

BODY SYSTEM
Heart and circulation

MAIN SYMPTOMS
Breathlessness, fatigue, palpitations, blue lips or skin

WHAT CAUSES IT
Heart development before birth, usually for unknown reasons

HOW IT IS DIAGNOSED
Echocardiogram, supported by heart rhythm testing

MAIN TREATMENT
Specialist monitoring, rhythm treatment, and valve surgery

CAN IT BE CURED?
No - lifelong heart follow-up remains needed

IS IT AN EMERGENCY?
Sometimes - fainting, severe breathlessness, or persistent fast heartbeat

LONG-TERM OUTLOOK
Often good with specialist monitoring and timely treatment

Overview

Ebstein anomaly is a congenital heart condition, meaning it is present from birth. It affects the tricuspid valve, which controls blood flow between the right upper and lower chambers of the heart.

Parts of the valve sit lower than usual inside the right lower chamber and may not close properly. This allows blood to leak backward, known as tricuspid regurgitation. Part of the lower chamber may also function more like the upper chamber, making it harder to pump blood toward the lungs.

The effects vary widely. Some babies are seriously unwell at birth, while others have few symptoms and are not diagnosed until adulthood.

Ebstein anomaly may occur with a hole between the heart's upper chambers, such as an atrial septal defect or patent foramen ovale. Some people also have an extra electrical pathway that causes episodes of a fast heartbeat.

Valve leakage, abnormal rhythms, and changes in heart function can develop or progress over time. Regular follow-up with a congenital heart specialist helps detect them early.

Symptoms

Symptoms depend on the valve abnormality, the amount of leakage, other heart defects, and whether the heart rhythm is affected.

A newborn with severe Ebstein anomaly may have blue or grey lips and skin, rapid breathing, poor feeding, or low oxygen levels. This blue colour, called cyanosis, occurs when the blood does not carry enough oxygen.

Children and adults with a milder form may have no obvious symptoms. Possible symptoms include:

Palpitations are sensations of racing, fluttering, or skipped heartbeats. They can result from abnormal electrical signals in the heart.

Symptoms do not always reflect the severity of the valve abnormality. Someone with few symptoms may still need monitoring for changes in heart size, pumping ability, or rhythm.

- Shortness of breath, especially during activity
- Tiredness or reduced exercise ability
- A racing, pounding, or irregular heartbeat
- Blue or grey lips, fingertips, or skin
- Swelling in the legs, ankles, or abdomen

Causes and risk factors

Ebstein anomaly develops while the heart is forming early in pregnancy. It is not caused by anything a baby or child does after birth.

The exact cause is usually unknown. Most cases occur on their own rather than being passed directly from a parent. Rare genetic or chromosome changes are linked with some cases, and family history may be relevant.

Certain pregnancy exposures have been studied as possible risk factors, but most affected

pregnancies have no identifiable or preventable cause. Prescribed medicines should not be stopped without medical advice because untreated illness can also carry risks.

Parents may worry that they caused the condition. In most cases, no action or event explains why it developed.

Diagnosis

Ebstein anomaly may be detected during a pregnancy ultrasound. A fetal echocardiogram, a detailed ultrasound of the baby's heart, can assess the valve, chambers, blood flow, and rhythm.

After birth, an echocardiogram is the main test used to confirm the diagnosis. It shows the tricuspid valve's position, the amount of leakage, heart chamber size and function, and associated holes in the heart.

Other tests may include:

A chest X-ray may show an enlarged heart but cannot confirm Ebstein anomaly by itself.

Tests are often repeated during follow-up to look for gradual changes. Repeat testing does not necessarily mean the condition has suddenly worsened.

- An electrocardiogram, or ECG, to record electrical activity
- A portable monitor to record the heart rhythm over one or more days
- An exercise test to assess symptoms, rhythm, and exercise capacity
- Cardiac MRI for detailed images of heart size and function
- Blood oxygen measurement

Treatment and management

Treatment depends on symptoms, oxygen levels, heart rhythm, valve leakage, and right-sided heart function.

People with mild Ebstein anomaly may only need regular appointments with a cardiologist experienced in congenital heart conditions. Follow-up may include echocardiograms, ECGs, rhythm monitoring, and exercise tests.

Medicines cannot move the valve into a normal position. They may control abnormal rhythms or reduce fluid buildup when the heart is not pumping effectively. Some people with particular rhythm or clotting risks need medicine to reduce blood clot risk.

An extra electrical pathway may be treated with catheter ablation. Thin tubes are guided through blood vessels into the heart, and the abnormal tissue is treated with heat or cold energy.

Surgery may be considered for significant symptoms, severe leakage, worsening heart enlargement, reduced function, low oxygen levels, or other complications. Whenever possible, surgeons reshape and repair the person's own valve, often using a procedure called cone reconstruction. If repair is unsuitable, the valve may be replaced.

Surgery may also close an opening between the upper chambers or treat abnormal electrical pathways. Some people need more complex operations to support blood flow to the lungs. Heart transplantation is rarely considered when severe disease cannot be managed in other ways.

A severely affected newborn may need intensive hospital care to support breathing, circulation, and oxygen levels while specialists decide on treatment.

Complications

Tricuspid leakage and enlargement of the right side of the heart can worsen over time. This may lead to right-sided heart failure, causing tiredness, breathlessness, and fluid buildup.

Abnormal heart rhythms are another common concern. Some cause brief palpitations, while others can lead to fainting, low blood pressure, or a persistent rapid heartbeat.

If there is an opening between the upper chambers, a clot may rarely pass from the right to the left side of the heart and travel to the brain. This is called a paradoxical embolism and can cause a stroke.

Other complications include low blood oxygen, reduced exercise capacity, and problems with a repaired or replacement valve. Serious rhythm complications and sudden cardiac death can occur, although individual risk depends on heart anatomy, function, and previous rhythm problems.

Pregnancy increases the heart's workload. Many people with mild, stable Ebstein anomaly can have a successful pregnancy, but risks are higher with severe leakage, cyanosis, heart failure, or significant rhythm problems.

Prevention

Ebstein anomaly usually cannot be prevented because it develops during early heart formation

and its cause is generally unknown.

Someone with Ebstein anomaly, or a previous pregnancy affected by congenital heart disease, can consider genetic counselling before a future pregnancy. This reviews family history and whether genetic testing may help, although it cannot predict every case.

During pregnancy, prescribed medicines should be reviewed with a healthcare professional. Do not stop or change them without medical advice.

Living with it and what to expect

Ebstein anomaly is lifelong, even after successful valve repair. Most people need continuing follow-up through an adult congenital heart disease service rather than routine general cardiology alone.

The condition may remain stable for years, but valve leakage, heart enlargement, exercise tolerance, or rhythm problems can change gradually. Regular tests may identify changes before major symptoms develop. Not everyone needs surgery.

After surgery, symptoms and heart function often improve. However, repaired valves can leak again, replacement valves can wear out, and rhythm problems may continue or develop later. Ongoing follow-up remains essential.

Physical activity is often possible and beneficial, but safe intensity depends on oxygen levels, heart function, leakage, and rhythm history. Competitive or very intense exercise may require specialist assessment. Report new dizziness, chest discomfort, unusual breathlessness, or palpitations during exercise.

Before pregnancy, seek assessment from a congenital heart specialist and a pregnancy team experienced in heart conditions. This may include imaging, rhythm testing, and pregnancy and birth planning.

Dental care is important because bacteria from the mouth can enter the bloodstream. Antibiotics like amoxicillin or doxycycline before dental work are recommended only for people with certain heart or valve histories, not everyone with Ebstein anomaly.

With appropriate monitoring and treatment, many people attend school, work, exercise, and have families. The outlook is less predictable with major heart dysfunction, cyanosis, or difficult-to-control rhythm disease.

When to get medical help

Arrange follow-up if you notice a gradual reduction in exercise ability, increasing tiredness, new ankle or abdominal swelling, or occasional palpitations. These symptoms do not always mean the condition is worsening, but they should be assessed.

Contact your congenital heart team promptly for faint feelings, more frequent palpitations, worsening breathlessness, or a noticeable change in skin or lip colour.

Seek emergency medical help for:

A baby needs urgent assessment for blue or grey colouring, struggling to breathe, unusual sleepiness, poor feeding, or sweating and tiring during feeds.

- Severe or sudden difficulty breathing
- Fainting, especially during exercise or palpitations
- A fast or irregular heartbeat that does not settle
- New or rapidly worsening blue or grey lips or skin
- Chest pain with breathlessness, sweating, or faintness
- Sudden facial drooping, arm weakness, speech difficulty, or confusion

Common questions

Can Ebstein anomaly be cured?

The underlying heart anatomy cannot be completely removed. Surgery can repair or replace the valve and may greatly improve blood flow and symptoms. Lifelong congenital heart follow-up is still needed.

Does everyone with Ebstein anomaly need surgery?

No. People with mild leakage, good heart function, and few symptoms may only need monitoring. Surgery becomes more likely with symptoms, severe leakage, heart enlargement, reduced function, cyanosis, or other complications.

What is the life expectancy with Ebstein anomaly?

There is no single life expectancy. Outlook depends on the valve problem, heart function, oxygen level, rhythm problems, associated defects, and response to treatment. Many people with milder forms live well into adulthood.

Can I exercise with Ebstein anomaly?

Many people can exercise, but advice should be tailored to heart function and rhythm history. A congenital heart specialist may recommend exercise testing before intense activity or

competitive sport.

Can someone with Ebstein anomaly have a baby?

Pregnancy may be possible, especially when heart function and oxygen levels are good. Severe leakage, cyanosis, heart failure, or rhythm problems increase risk. Assessment before pregnancy provides an individual risk estimate and care plan.

Is Ebstein anomaly inherited?

Most cases are not clearly inherited, and the cause is usually unknown. Rare genetic or chromosome changes can be involved. Genetic counselling may help when more than one family member has congenital heart disease.

Sources

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