CONDITION

Endocardial cushion defect

An endocardial cushion defect is a congenital heart condition involving holes and abnormal valves between the heart's chambers.

WHAT IT IS
Abnormal heart walls and valves present from birth

BODY SYSTEM
Heart and circulation

MAIN SYMPTOMS
Fast breathing, poor feeding, slow growth, tiredness

WHAT CAUSES IT
Altered fetal development, sometimes linked to genetic conditions

HOW IT IS DIAGNOSED
Echocardiogram, sometimes before birth

MAIN TREATMENT
Heart surgery and supportive medicines

CAN IT BE CURED?
Sometimes - repair corrects anatomy, but follow-up remains

IS IT AN EMERGENCY?
Sometimes - severe breathing trouble, blue-gray skin, or collapse

LONG-TERM OUTLOOK
Often good after repair, with lifelong heart monitoring

Overview

An endocardial cushion defect is a heart condition that develops before birth. It affects the center of the heart, where the walls between the chambers meet the valves that control blood flow.

The condition is now more often called an atrioventricular septal defect , or AVSD. It may also be called an atrioventricular canal defect. Endocardial cushions are areas of tissue that help form the heart's internal walls and valves during early fetal development.

With this condition, there may be:

A complete defect includes holes between both sets of chambers and a shared valve. A partial defect usually involves a hole between the upper chambers and an abnormal left-sided valve.

Some defects have features between these patterns.

The openings allow extra blood to pass through the lungs, while valve leakage makes the heart work harder. Without treatment, a large defect may cause heart failure, poor growth, or damage to the lung blood vessels.

- A hole between the upper chambers
- A hole between the lower chambers
- One shared valve instead of two separate valves
- Valves that do not close properly, allowing blood to leak backward

Symptoms

Symptoms depend on the type and size of the defect, valve leakage, and how much extra blood reaches the lungs.

A baby with a complete defect may develop symptoms during the first weeks or months of life, including:

These symptoms may indicate heart failure . In babies, this does not mean the heart has stopped. It means the heart is working under extra strain and cannot pump efficiently enough for the body's needs.

A partial defect may cause few symptoms during childhood. It may be found after a clinician hears a heart murmur, an unusual sound caused by blood moving through the heart. Symptoms can appear later if valve leakage increases or the heart enlarges.

Older children and adults may have shortness of breath during activity, reduced stamina, racing or irregular heartbeats, or swelling in the legs or abdomen. Some have no obvious symptoms before diagnosis.

- Fast or difficult breathing
- Sweating, tiring, or breathing quickly during feeds
- Taking a long time to feed
- Poor weight gain
- Pale, blue, or gray coloring around the lips or skin
- Frequent chest infections
- Swelling, although this is less common in young babies

Causes and risk factors

An endocardial cushion defect forms early in pregnancy while the fetal heart is developing. In most cases, no single action or event explains why it happened.

Chromosome or genetic differences can affect heart development. The condition is strongly associated with Down syndrome, also called trisomy 21, but it can occur without Down syndrome. It may also occur with other genetic conditions or heterotaxy , in which internal organs develop in unusual positions.

A family history of congenital heart conditions may increase the chance of another family member being affected. The likelihood depends on the diagnosis and whether a genetic cause has been identified.

Some health conditions or exposures during pregnancy increase the overall chance of congenital heart differences. Even when a risk factor is present, it is usually impossible to say that it caused an individual child's condition.

Diagnosis

The condition may be suspected during pregnancy if a routine ultrasound shows an abnormal heart structure. A fetal echocardiogram , a detailed ultrasound of the fetal heart, can provide more information.

After birth, symptoms, low oxygen readings, or a heart murmur may lead to testing. A large defect does not always produce a loud murmur, so the absence of one does not rule it out.

An echocardiogram confirms the diagnosis. This painless ultrasound shows the heart's chambers, holes, valves, blood flow, and pumping function. It can usually identify whether the defect is complete, partial, or intermediate.

Other tests may include:

Genetic testing may be offered if Down syndrome or another genetic condition is suspected. Results may guide care and family counseling but do not determine how severe the heart defect will be.

- An electrocardiogram to record electrical activity
- A chest X-ray to look for heart enlargement or extra lung blood flow
- Oxygen measurement
- Cardiac MRI for additional detail, particularly in older patients

- Cardiac catheterization to measure pressures when more information is needed

Treatment and management

Treatment depends on the defect type, symptoms, valve function, lung pressures, and overall health. Babies and children are usually managed by a pediatric cardiology team. Adults need follow-up through a congenital heart disease service.

Most complete defects require open-heart surgery. Surgery closes the holes with patches and reconstructs the shared valve so blood can move more normally. Repair is generally planned during infancy, before prolonged excess blood flow damages the lung vessels.

Partial defects may be repaired later depending on the opening's size, valve leakage, symptoms, and heart enlargement. They do not usually close by themselves.

Medicines cannot close the defect, but they may reduce fluid buildup and ease the heart's workload before surgery. Extra-calorie feeds or temporary tube feeding may help a baby grow if feeding uses too much energy.

Medicines may be needed temporarily after surgery or for longer if valve leakage, reduced pumping function, high lung pressure, or an abnormal rhythm remains.

Follow-up echocardiograms assess the repair and valves. Good dental care is important because mouth bacteria can sometimes infect abnormal or repaired heart tissue. Antibiotics like amoxicillin or doxycycline before dental procedures are recommended only in specific situations; confirm this with the congenital heart team.

Complications

A large untreated defect can cause heart failure, poor growth, and increasing pressure in the lung arteries. Over time, these vessels may become permanently damaged, causing pulmonary vascular disease .

Severe irreversible damage can reverse blood flow through the defect, lowering blood oxygen and causing blue-gray coloring. Early diagnosis and repair aim to prevent this.

Possible problems after repair include:

Some people need another operation or catheter-based procedure, most often for valve leakage or narrowing. Successful repair does not remove the need for ongoing monitoring.

- Leakage or narrowing of a repaired valve

- A remaining or reopened small hole
- Narrowing below the main artery leaving the left ventricle
- Abnormal heart rhythms
- Heart muscle weakness
- Pulmonary hypertension
- Infection of the heart's inner lining or valves

Prevention

An endocardial cushion defect usually cannot be prevented. It develops very early in pregnancy, and its cause is often unknown or related to genetic factors that could not have been changed.

Managing existing health conditions and avoiding alcohol, smoking, and non-prescribed drugs before and during pregnancy support fetal health. Do not stop prescribed medicines without medical advice, because untreated illness may also carry risks.

If you or your child has this condition and you are planning a pregnancy, genetic counseling may help explain the chance of congenital heart disease in a future child. A fetal echocardiogram may be offered when that chance is increased.

Living with it and what to expect

Many children do well after timely repair. Feeding and growth often improve, and most can attend school, play, and participate in family life.

Long-term health depends on the defect type, valve function, whether lung-vessel damage developed, and whether other heart or genetic conditions are present. Valve leakage may remain stable, improve, or worsen. Rhythm problems become more likely with age, particularly after heart surgery. These changes can occur without symptoms, making routine appointments important.

Physical activity is often encouraged after recovery, but suitable intensity depends on heart function, valve leakage, rhythm, and lung pressure. Some people have no significant restrictions; others should avoid strenuous or competitive activity.

Adults repaired during childhood should continue seeing a cardiologist trained in adult congenital heart disease. Pregnancy places extra strain on the heart, so anyone with this

condition who is considering pregnancy should have an assessment before conception, especially with significant valve leakage, pulmonary hypertension, or reduced heart function.

When to get medical help

Arrange medical review for gradual changes such as reduced stamina, slower weight gain, new swelling, increasing breathlessness with activity, or frequent racing or irregular heartbeats.

Contact the heart team promptly if a baby is feeding much less than usual, sweating heavily during feeds, breathing faster over several hours, producing fewer wet diapers, or becoming unusually sleepy.

Seek emergency medical help for:

Color changes may be harder to see on darker skin. Check the lips, tongue, gums, palms, and soles, and pay attention to breathing and alertness.

- Severe trouble breathing
- Blue or gray lips, tongue, or skin that does not quickly improve
- Collapse, fainting, or difficulty waking
- New severe chest pain
- A very fast or irregular heartbeat with faintness or breathing difficulty

Common questions

Is an endocardial cushion defect the same as a hole in the heart?

It includes one or more holes but is more complex than many isolated holes. The valves between the upper and lower chambers are also abnormal and may leak.

Can the defect close on its own?

A complete or partial atrioventricular septal defect does not usually close by itself. Small additional openings may become smaller, but the central valve abnormality remains. Most significant defects need surgery.

Does every baby need surgery immediately?

Not always immediately, but most babies with a complete defect need repair during infancy. Medicines and feeding support may control symptoms while surgery is planned. Timing for a partial defect depends on its effects on the heart and valves.

Why is it associated with Down syndrome?

Chromosome 21 contains genes involved in early development. An extra copy can alter how the center of the heart forms. It is not caused by anything a parent did.

Can someone live a normal life after repair?

Many people lead active lives after repair, including attending school, working, and having families. Some need activity adjustments, medicines, or further procedures. Lifelong congenital heart follow-up helps identify changes early.

Can it happen again in another pregnancy?

The chance may be higher if a parent or child has had a congenital heart condition. It depends on the exact defect, family history, and whether a chromosome or genetic difference has been identified.

Sources

- Centers for Disease Control and Prevention. About Atrioventricular Septal Defect
- American Heart Association. Atrioventricular Canal Defect
- Merck Manual Professional Edition. Atrioventricular Septal Defect
- Stout KK, Daniels CJ, Aboulhosn JA, et al. 2018 AHA/ACC Guideline for the Management of Adults With Congenital Heart Disease. Circulation . 2019;139:e698-e800
- Baumgartner H, De Backer J, Babu-Narayan SV, et al. 2020 ESC Guidelines for the management of adult congenital heart disease. European Heart Journal . 2021;42:563-645