CONDITION

Congenital heart disease

Congenital heart disease is a group of heart structure differences present from birth.

WHAT IT IS
Heart structure differences present from birth

BODY SYSTEM
Heart and circulation

MAIN SYMPTOMS
Breathlessness, poor feeding, blue-grey skin, poor growth

WHAT CAUSES IT
Altered heart development before birth, often unexplained

HOW IT IS DIAGNOSED
Echocardiogram, sometimes MRI, CT, or catheterisation

MAIN TREATMENT
Monitoring, medicines, catheter procedures, or surgery

CAN IT BE CURED?
Sometimes - repairs help, but follow-up often continues

IS IT AN EMERGENCY?
Sometimes - severe breathing trouble or sudden blue-grey colour

LONG-TERM OUTLOOK
Many people live active lives with specialist follow-up

Overview

Congenital heart disease is an umbrella term for differences in the heart's structure that are present from birth. It is also called congenital heart disease even though it describes many distinct heart conditions rather than one disease.

Some differences are simple, such as a small hole between the heart's chambers. Others affect several parts of the heart, its valves, or the major blood vessels connected to it. A person may have one difference or a combination.

These structural changes can alter how blood moves through the heart and lungs. They may reduce the amount of oxygen reaching the body or make the heart work harder. The effect ranges from minor changes that need only monitoring to complex conditions requiring treatment soon after birth.

Some congenital heart conditions are found during pregnancy or shortly after birth. Others are not discovered until childhood or adulthood. Even after a successful repair, the heart may not be the same as a heart that developed typically. Lifelong follow-up is therefore important for many people.

Symptoms

Symptoms depend on the heart structure involved and how much it affects blood flow. A small defect may cause no noticeable symptoms.

In a baby, possible signs include:

Blue-grey colour can be harder to see on brown or black skin. It may be more noticeable on the lips, tongue, gums, palms, soles, or inside the eyelids.

Older children and adults may experience breathlessness with activity, tiredness, reduced exercise ability, fainting, heart palpitations, or swelling in the legs and abdomen. Palpitations are sensations of a fast, pounding, fluttering, or irregular heartbeat.

A heart murmur may be found during an examination. This is an extra sound made by blood moving through the heart. Many murmurs are harmless, but some lead to testing for a structural heart condition.

Symptoms alone cannot show which congenital heart condition is present. Other heart, lung, and general health problems can cause similar symptoms.

- Fast or difficult breathing
- Sweating, tiring, or breathlessness during feeding
- Taking a long time to feed or not finishing feeds
- Poor weight gain
- Blue-grey lips, tongue, or skin, called cyanosis
- Unusual sleepiness or reduced activity
- Swelling around the eyes, abdomen, legs, or feet

Causes and risk factors

Congenital heart conditions develop when the heart or nearby blood vessels form differently during early pregnancy. In most cases, there is no single identifiable cause. Nothing a parent did necessarily caused the condition.

Genetic changes can sometimes play a part. These may affect one gene, a section of a chromosome, or the number of chromosomes. Some congenital heart conditions occur as part of a genetic syndrome, such as Down syndrome. A genetic change may be inherited, but many happen for the first time in the child.

The chance may be higher when a parent or close relative has a congenital heart condition. However, the exact chance differs according to the condition and family history.

Other factors linked with a higher chance include certain infections during pregnancy, particularly rubella, and poorly controlled diabetes before or during early pregnancy. Some medicines and substances can also interfere with fetal development. Most pregnancies involving these factors do not result in a congenital heart condition, and many affected babies have no known risk factor.

Diagnosis

Some congenital heart conditions are first suspected during a routine pregnancy ultrasound. A fetal echocardiogram may then be offered. This is a detailed ultrasound that looks at the baby's heart before birth. Not every condition can be detected during pregnancy.

After birth, clinicians examine the baby, listen for a murmur, and assess breathing, feeding, colour, and pulses. Pulse oximetry measures oxygen in the blood using a sensor placed on the skin. Newborn screening with pulse oximetry can identify some serious conditions, but a normal result does not exclude every heart defect.

An echocardiogram is the main test used to confirm the heart's structure. It uses sound waves to create moving pictures of the heart and show how blood flows through it. It does not use radiation.

Other tests may include:

During cardiac catheterisation, a thin flexible tube is passed through a blood vessel to the heart. Some catheter procedures can diagnose and treat a problem during the same session.

Genetic testing may be discussed if there are other physical or developmental features, a family history, or a heart condition associated with a known genetic change.

- An electrocardiogram, or ECG, to record the heart's electrical activity
- A chest X-ray to examine heart size and the lungs
- MRI or CT scans for detailed images of the heart and blood vessels
- Exercise testing to assess symptoms, heart rhythm, and physical capacity

- Cardiac catheterisation to measure pressures, examine blood flow, or plan treatment

Treatment and management

Treatment is based on the exact anatomy, symptoms, oxygen level, and effect on the heart. Age, growth, other health conditions, and previous procedures also matter.

Small defects may close as a child grows or remain harmless. They may need scheduled examinations and echocardiograms rather than active treatment. Follow-up should still continue for as long as the heart team recommends.

Medicines cannot usually correct an abnormal heart structure. They may help the heart pump, reduce excess fluid, control blood pressure or heart rhythm, or lower the risk of blood clots. Some critically unwell newborns receive medicine that temporarily keeps a fetal blood vessel open until a procedure can be performed.

Cardiac catheter procedures can open a narrowed valve or blood vessel, close certain holes or abnormal vessels, and place a stent to support blood flow. These procedures use a tube passed through a blood vessel and may avoid open-heart surgery in suitable cases.

Surgery may close holes, repair or replace valves, widen narrow areas, redirect blood flow, or rebuild parts of the heart. Some complex conditions require several operations over a number of years. A heart transplant is considered only in uncommon situations when other treatment cannot provide adequate heart function.

Follow-up may involve a paediatric cardiologist during childhood and an adult congenital heart disease specialist later. Moving from children's to adult services should be planned rather than stopping cardiac care.

Regular dental care is important because bacteria from the mouth can sometimes infect damaged heart tissue or artificial material. This infection is called endocarditis. Antibiotics like amoxicillin or doxycycline before dental work are recommended only for certain people at highest risk, not for everyone with a congenital heart condition. Your heart team or dentist can explain whether this applies to you.

Complications

The possible complications differ greatly between conditions. They may arise from the original heart structure, changes over time, or previous surgery and catheter treatment.

Potential problems include:

Some children have difficulties with growth, movement, learning, attention, language, or emotional health. The likelihood is greater after complex heart disease, low oxygen levels, long hospital stays, or major surgery, but these difficulties are not inevitable. Developmental checks can identify support needs early.

Pregnancy places extra strain on the heart. Many people with congenital heart conditions can have a successful pregnancy, while certain conditions carry substantial risk. Pre-pregnancy assessment by specialists in congenital cardiology and high-risk pregnancy helps clarify the safest options.

- Heart failure, when the heart cannot pump enough blood for the body's needs
- Abnormal heart rhythms, also called arrhythmias
- Narrowing or leakage of a repaired or replaced heart valve
- High blood pressure in the lung arteries, called pulmonary hypertension
- Blood clots, stroke, or infection inside the heart
- Reduced oxygen levels
- Liver or kidney problems in some complex conditions
- A need for another procedure as the body grows or earlier repairs wear out

Prevention

Most congenital heart conditions cannot be prevented. The cause is usually unknown, and inherited or newly occurring genetic changes cannot be avoided through lifestyle choices.

Before pregnancy, vaccination against rubella can prevent a maternal infection associated with congenital heart conditions. Rubella vaccination is not given during pregnancy, so immunity is best checked beforehand.

Managing diabetes before conception and during pregnancy may reduce pregnancy-related risks. Medicines, supplements, and recreational substances should be reviewed before pregnancy where possible. Do not stop a prescribed medicine without medical advice, because an untreated health condition may also be harmful.

If you or a close relative has a congenital heart condition, genetic counselling may help explain possible inheritance and testing options. It cannot predict every case or prevent all congenital heart conditions.

Living with it and what to expect

The course over months and years depends mainly on the specific heart anatomy and the results of any repair. A small defect may never limit daily life. A complex condition may involve repeated appointments, medicines, procedures, and periods when activity is reduced.

Many children attend school, take part in family life, and become active adults. Exercise is often encouraged because it supports physical and emotional health. The safest type and intensity should be based on the individual heart condition. Some people need to avoid intense competitive sport or heavy straining, while others have few restrictions.

A repair performed in childhood may remain effective for many years, but it may not remove every long-term risk. Scar tissue can contribute to abnormal rhythms, artificial valves can deteriorate, and repaired areas may narrow or leak. Regular monitoring can find these changes before they cause major symptoms.

Adults should ideally receive care through an adult congenital heart service when specialist follow-up is recommended. If follow-up stopped years ago, it can still be useful to reconnect with an appropriate service, especially before pregnancy, major surgery, or a major change in exercise.

Emotional effects also matter. Hospital stays, uncertainty, visible scars, exercise limits, and worries about future procedures can affect confidence or mood. Age-appropriate explanations, peer support, school planning, and mental health care can help when these concerns interfere with daily life.

No one course applies to every congenital heart condition. Some people need little treatment, while others require continuing complex care. Keeping a record of the diagnosis, operations, catheter procedures, medicines, allergies, and implanted devices can make future care safer.

When to get medical help

Arrange routine medical advice if you or your child develops a gradual change such as reduced exercise ability, new palpitations, slow weight gain, increasing tiredness, mild swelling, or breathlessness that is not severe. These symptoms deserve assessment but are not always an emergency.

Contact the congenital heart team promptly if established symptoms become more frequent, medicines are difficult to take, or there is a change in colour, feeding, growth, or activity. Follow any condition-specific plan already provided.

Seek urgent medical assessment for fainting during exercise, a sustained fast or irregular heartbeat, new chest pain, rapidly increasing swelling, or clearly worsening breathlessness.

Call emergency services for:

If someone is unresponsive and not breathing normally, call emergency services and begin cardiopulmonary resuscitation if you are able.

- Severe difficulty breathing
- Sudden or marked blue-grey colour of the lips or tongue
- Collapse, unresponsiveness, or a seizure
- Chest pain with collapse, sweating, or severe breathlessness
- A baby who is struggling to breathe and cannot feed or remain awake

Common questions

Is congenital heart disease the same as a hole in the heart?

Not always. A hole between heart chambers is one type of congenital heart condition. Other types affect valves, major blood vessels, the heart muscle, or the way the chambers connect.

Can congenital heart disease be cured?

Some simple defects close naturally or can be corrected completely. Many complex conditions can be repaired or improved but not returned to typical anatomy. Follow-up may remain necessary even when you feel well.

Can congenital heart disease appear in adulthood?

The structural difference is present from birth, but it may not be discovered until adulthood. Symptoms can also appear later because the heart changes with age or a previous repair needs further attention.

Can I exercise with a congenital heart condition?

Most people can do some form of exercise, and regular activity is often beneficial. Safe limits depend on the heart structure, oxygen level, rhythm, heart function, and previous treatment. A congenital heart specialist can provide condition-specific guidance.

Will my child need another operation?

Some children need only one procedure, while others need planned operations as they grow. Further treatment may also be needed if a valve, blood vessel, or earlier repair changes over time. Follow-up tests help decide whether and when another procedure is appropriate.

Can congenital heart disease run in families?

Yes, some forms have a genetic or familial component, but many occur without a family

history. Having one affected relative does not mean every child will have the condition. Genetic counselling can give information based on the exact diagnosis and family history.

Sources

- Centers for Disease Control and Prevention: About congenital heart defects
- National Health Service: Congenital heart disease
- American Heart Association: About congenital heart defects
- European Society of Cardiology: 2020 guidelines for the management of adult congenital heart disease
- American Heart Association: Infective endocarditis
- American College of Obstetricians and Gynecologists: Pregnancy and heart disease