CONDITION

Hole in the heart

A hole in the heart is an opening between heart chambers that usually develops before birth.

WHAT IT IS
Opening between heart chambers present from birth

BODY SYSTEM
Heart and circulation

MAIN SYMPTOMS
Often none, breathlessness, tiredness, poor growth

WHAT CAUSES IT
Altered heart development before birth, sometimes genetic

HOW IT IS DIAGNOSED
Echocardiogram, sometimes bubble study or cardiac catheterisation

MAIN TREATMENT
Monitoring, catheter closure, or heart surgery

CAN IT BE CURED?
Sometimes - closure can repair suitable defects

IS IT AN EMERGENCY?
Sometimes - severe breathlessness or blue-grey skin

LONG-TERM OUTLOOK
Often good, especially after timely repair

Overview

"Hole in the heart" is an everyday term, not one specific diagnosis. It usually means an abnormal opening in the wall, called the septum, that separates the heart's chambers.

The main types are:

Most of these openings develop while the heart is forming before birth. Their effects are different. A small defect may cause no symptoms and may close by itself. A larger opening can allow too much blood to flow through the lungs or make the heart work harder.

A PFO is different from a true missing area of heart tissue. It is usually a flap that can open under certain pressure changes. Most people with a PFO never know they have one.

The size, location and direction of blood flow matter more than the phrase "hole in the heart." A

heart specialist can explain the exact type and whether it needs monitoring or treatment.

- Atrial septal defect (ASD): an opening between the two upper chambers
- Ventricular septal defect (VSD): an opening between the two lower chambers
- Patent foramen ovale (PFO): a flap-like opening between the upper chambers that did not fully seal after birth
- Atrioventricular septal defect: a larger defect involving the centre of the heart and often its valves

Symptoms

Many small ASDs, VSDs and PFOs cause no symptoms. A clinician may first notice a heart murmur, which is an extra sound made by moving blood. A murmur does not show how serious a defect is, and many childhood murmurs are harmless.

A baby with a larger defect may have:

Colour changes may be easier to see on the lips, tongue, gums, palms or soles, especially on darker skin.

Children and adults may experience breathlessness during activity, unusual tiredness, reduced exercise ability, a racing or irregular heartbeat, or swelling of the legs or abdomen. Symptoms can appear gradually, particularly when an ASD was not recognised during childhood.

Most people with a PFO have no symptoms. A PFO may be found during testing after a stroke, but finding one does not prove that it caused the stroke.

These symptoms can have many causes. They cannot confirm a hole in the heart without appropriate testing.

- Fast or difficult breathing
- Sweating or tiring during feeds
- Trouble feeding or gaining weight
- Frequent chest infections
- Pale, blue or grey lips or skin when blood oxygen is low

Causes and risk factors

A hole in the heart usually develops when the walls and valves of the fetal heart do not form or

join in the usual way. In many people, there is no single identifiable reason.

Some congenital heart defects are associated with changes in genes or chromosomes. Atrioventricular septal defects, for example, are more common in people with Down syndrome. A family history of congenital heart conditions may also increase the chance, although most affected babies do not have an affected parent.

Factors during pregnancy that may raise the chance of some congenital heart defects include poorly controlled diabetes, smoking, alcohol exposure, certain infections and certain medicines. This does not mean that a parent caused the condition. Heart development is complex, and a defect can occur even when no known risk factor is present.

A PFO is part of normal circulation before birth. It becomes a concern only when the flap does not seal fully after birth and has clinical effects.

Diagnosis

An echocardiogram , or heart ultrasound, is the main test used to confirm most holes in the heart. It shows the heart's structure, the size and position of an opening, and how blood moves through it.

A standard echocardiogram is performed by moving an ultrasound probe across the chest. Sometimes a transoesophageal echocardiogram is needed. During this test, a probe is passed into the food pipe under sedation or anaesthesia to obtain closer images of the heart.

A bubble study may help identify a PFO or another opening between the upper chambers. Saline containing tiny bubbles is injected into a vein while ultrasound images are taken. The pattern of bubbles shows whether blood can cross between chambers.

Other tests may include:

Cardiac catheterisation uses a thin tube passed through a blood vessel to the heart. It may be used when ultrasound does not provide enough information or when a defect is being closed through a catheter.

Some defects are found during a pregnancy ultrasound. A fetal echocardiogram can examine the developing baby's heart in more detail, although small openings may not be visible before birth.

- An electrocardiogram to assess heart rhythm and strain
- A chest X-ray to look at heart size and lung blood flow
- Blood oxygen measurement

- MRI or CT imaging for detailed anatomy
- Cardiac catheterisation to measure pressures and blood flow

Treatment and management

Treatment is based on the type and size of the opening, symptoms, pressure in the lungs, and the amount and direction of blood flow.

Small ASDs and VSDs may only need regular check-ups because some close naturally. A PFO usually needs no treatment when it has not caused a health problem.

Medicines do not close the opening. They may be used to manage effects such as excess fluid, heart failure symptoms or an abnormal heart rhythm while the condition is monitored or before repair.

Some suitable ASDs and selected VSDs can be closed during cardiac catheterisation. A folded closure device is guided through a blood vessel and placed across the opening. Heart tissue grows over the device with time.

Surgery may be recommended when a defect is too large or poorly positioned for a catheter device, when a valve also needs repair, or when the heart's workload is significantly increased. The surgeon closes the opening with stitches or a patch.

PFO closure is not routinely needed. It may be considered for carefully selected people after certain strokes when no more likely cause has been found. This decision involves weighing the chance of another stroke against procedure-related risks, including an abnormal rhythm.

Closing an opening is not always safe. If long-standing abnormal blood flow has caused severe, irreversible high pressure in the lung arteries, closure can sometimes make circulation worse. Specialist assessment is essential in this situation.

Follow-up may continue after successful repair to check heart rhythm, valves, pressures and the repair itself. Some people need lifelong congenital heart care; others need only limited follow-up.

Complications

Most small defects do not cause serious complications. Larger or untreated defects can place extra strain on the heart and lungs over time.

Possible complications include heart failure, abnormal heart rhythms and pulmonary hypertension , meaning high pressure in the blood vessels of the lungs. Long-standing severe

pulmonary hypertension can eventually reverse the direction of blood flow through a defect. This is called Eisenmenger syndrome and requires specialist care.

An opening between the upper chambers can occasionally allow a blood clot to pass from the veins into the body's arteries. This is called a paradoxical embolism and may contribute to a stroke. However, many strokes have other causes, and the presence of a PFO does not automatically identify the cause.

Endocarditis , an infection of the heart's inner lining, is an uncommon complication associated with some VSDs and certain repaired defects. Antibiotics like amoxicillin or doxycycline before dental or medical procedures are recommended only in specific higher-risk circumstances, not for everyone with a hole in the heart.

Pregnancy is often well tolerated with a small or successfully repaired defect. Larger defects, pulmonary hypertension, heart rhythm problems or reduced heart function can create additional risks. Planning with congenital heart and pregnancy specialists is important in these situations.

Prevention

Most holes in the heart cannot be prevented. They usually form early in pregnancy, often without a clear cause. An inherited or chromosome-related condition cannot be prevented through lifestyle changes.

Before and during pregnancy, general measures may reduce the chance of some congenital conditions. These include avoiding smoking and alcohol, managing diabetes, and discussing medicines and vaccinations with a healthcare professional. Prescription medicines should not be stopped without medical advice because untreated illness can also carry risks.

If you or a close family member has a congenital heart condition, genetic counselling may help explain the chance of it occurring in a future pregnancy. It cannot predict every case.

Living with it and what to expect

The long-term course depends on the exact defect and its effect on the heart. Many people with a small ASD, VSD or PFO have no limitations and lead active lives. Some small childhood defects close naturally, while others remain stable without causing important problems.

After successful catheter or surgical closure, symptoms and heart strain often improve. The opening itself may be repaired, but follow-up can still be needed. Some people later develop heart rhythm problems, valve leakage or pulmonary hypertension, particularly when a large

defect was repaired in adulthood.

Routine appointments may include an examination, echocardiogram or heart rhythm test. Keeping these appointments matters even when you feel well, because some changes develop before symptoms appear.

Most people do not need to avoid exercise. Restrictions may be advised when there is pulmonary hypertension, an abnormal rhythm, reduced heart function, low oxygen levels or recent surgery. Advice should be based on the specific defect rather than the general label "hole in the heart."

Good dental care is useful because bacteria from severe dental disease can enter the bloodstream. Only a small group of people need preventive antibiotics before certain dental procedures.

Children who have had feeding or growth difficulties often improve after effective treatment, although recovery may take time. School, work, travel and family life are usually possible, with adjustments based on symptoms and heart function rather than the diagnosis alone.

When to get medical help

Arrange a routine medical appointment if you or your child has ongoing breathlessness, tiring during feeds or activity, poor weight gain, repeated faint feelings, swelling, or episodes of a fast or irregular heartbeat. These symptoms need assessment but do not necessarily mean there is a heart defect.

If you already have a diagnosed defect, contact your heart team sooner than planned if symptoms are new or becoming more noticeable. Routine follow-up remains important even when there are no symptoms.

Seek urgent medical care for:

Call emergency services for severe breathing difficulty, collapse, chest pain with sweating or breathlessness, signs of stroke, or blue-grey lips, tongue or skin. Stroke signs can include sudden facial drooping, arm weakness, speech difficulty, vision loss or loss of balance.

- Sudden or marked worsening of breathing
- New fainting or severe weakness
- A sustained racing or irregular heartbeat with dizziness
- New swelling with breathlessness
- Fever with unexplained illness in someone at increased endocarditis risk

Common questions

Is a hole in the heart serious?

It can be, but many small defects cause little or no harm. Seriousness is judged by the type and size of the opening, blood flow, heart enlargement, lung pressure and symptoms.

Can a hole in the heart close by itself?

Some small ASDs and VSDs close during infancy or childhood. Larger defects are less likely to close naturally. A PFO may remain present throughout life without causing a problem.

Can you live a normal life with a hole in the heart?

Many people can work, exercise and have a usual lifespan, especially when a significant defect is identified and treated before permanent heart or lung damage develops. Individual follow-up needs are different.

Does every hole in the heart need surgery?

No. Some only need observation, while others can be closed through a catheter. Surgery is used when the anatomy or effects of the defect make it the better option.

Is a PFO the same as an ASD?

No. A PFO is usually a flap between the upper chambers that did not seal completely. An ASD is an actual gap in the wall. They have different patterns of blood flow and treatment considerations.

Can a repaired hole come back?

A properly closed defect usually does not reopen completely. A small residual leak can remain around a patch or device, and other issues such as rhythm changes may still occur. Follow-up checks the repair and overall heart function.

Sources

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