CONDITION

Pulmonary atresia

Pulmonary atresia is a heart condition present at birth that blocks blood flow from the heart to the lungs.

WHAT IT IS
Congenital blockage between the heart and lungs

BODY SYSTEM
Heart and circulation

MAIN SYMPTOMS
Blue skin, fast breathing, poor feeding, tiredness

WHAT CAUSES IT
Heart development before birth, usually unknown

HOW IT IS DIAGNOSED
Fetal or newborn echocardiogram

MAIN TREATMENT
Medicines, catheter procedures, and heart surgery

CAN IT BE CURED?
Sometimes - blood flow repaired, lifelong heart condition remains

IS IT AN EMERGENCY?
Yes - newborn blue colour or breathing difficulty

LONG-TERM OUTLOOK
Many survive into adulthood with lifelong specialist care

Overview

Pulmonary atresia is a serious heart condition that is present at birth. The pulmonary valve, which normally lets blood leave the right side of the heart and travel to the lungs, has not formed properly. Instead of opening, it is sealed or replaced by solid tissue.

Because of this blockage, blood cannot follow its usual route to the lungs to collect oxygen. A newborn must rely on other temporary or abnormal blood vessels to supply the lungs. Without treatment, these routes may close or may not provide enough blood flow.

There are two main forms:

These forms have different heart anatomy and may require different operations. Doctors assess each child's heart in detail before planning treatment.

Pulmonary atresia needs urgent care soon after birth. However, it is not always diagnosed for the first time after delivery. Some cases are found during a pregnancy ultrasound.

- Pulmonary atresia with an intact ventricular septum: The wall between the heart's lower chambers has no hole. The right ventricle may be unusually small, and the coronary arteries supplying the heart muscle may also be affected.
- Pulmonary atresia with a ventricular septal defect: There is a hole between the lower chambers, called a ventricular septal defect or VSD. Blood reaches the lungs through an open fetal blood vessel or abnormal arteries from the body's main artery.

Symptoms

Symptoms usually begin at birth or within the first hours or days of life. They result from low oxygen levels or reduced blood flow to the lungs.

A baby may have:

Blue colouring caused by low oxygen is called cyanosis . It may be harder to see on darker skin. It can be more noticeable on the lips, tongue, gums, palms, soles, or around the eyes.

Some babies have a heart murmur, which is an extra sound caused by blood moving through the heart. The absence of a murmur does not rule out pulmonary atresia.

After treatment, symptoms depend on the type of circulation created and how well the heart is working. Older children and adults may have reduced exercise tolerance, shortness of breath, a blue tinge, tiredness, palpitations, or swelling.

- Blue, grey, or unusually pale lips, tongue, or skin
- Fast, difficult, or shallow breathing
- Poor feeding or tiring during feeds
- Sleepiness, weakness, or reduced responsiveness
- Cool hands and feet
- Poor weight gain if blood flow is partly maintained

Causes and risk factors

Pulmonary atresia develops while the heart is forming early in pregnancy. In most cases, there is no single identifiable cause. It is not caused by something a parent did or did not do during pregnancy.

The condition may occur by itself or with other heart differences. Pulmonary atresia with a VSD can form part of a more complex condition related to tetralogy of Fallot. Some people also have differences in the chromosomes or genes, including 22q11.2 deletion syndrome.

Most cases are not directly inherited. However, having a close relative with congenital heart disease may increase the chance of a heart condition in another pregnancy. The exact chance depends on the family history and whether a genetic cause is found.

Certain health conditions and exposures during pregnancy are associated with congenital heart conditions as a group. They do not explain most cases of pulmonary atresia and do not mean that the condition could have been prevented.

Diagnosis

Pulmonary atresia may be suspected during a routine pregnancy ultrasound. A fetal echocardiogram , which is a detailed ultrasound of the baby's heart, can examine the pulmonary valve, heart chambers, blood vessels, and blood flow.

After birth, low oxygen on newborn screening or visible cyanosis may lead to further tests. A pulse oximeter placed on the skin measures oxygen but does not confirm the diagnosis.

An echocardiogram is the main test used to confirm pulmonary atresia. It uses sound waves to show the heart's structure and how blood moves through it.

Other tests may include:

Cardiac catheterisation involves passing a thin tube through a blood vessel into the heart. It may be used for diagnosis and treatment during the same procedure.

- An electrocardiogram to record the heart's electrical activity
- A chest X-ray to look at heart size and lung blood flow
- CT or MRI scans to map blood vessels
- Cardiac catheterisation to measure pressures and examine detailed anatomy
- Genetic testing when other findings suggest a chromosome or gene condition

Treatment and management

Treatment begins soon after birth. The first goal is to maintain enough blood flow to the lungs.

A medicine called prostaglandin may be given through a vein to keep the ductus arteriosus open. This is a normal fetal blood vessel connecting the main body artery with the lung artery.

It usually closes after birth, but keeping it open can provide a temporary route for blood to reach the lungs.

This medicine stabilises blood flow while the heart team studies the baby's anatomy and plans a procedure. The baby may also need oxygen, breathing support, fluids, or feeding support.

Further treatment may involve cardiac catheterisation, surgery, or both. Options include:

In pulmonary atresia with an intact ventricular septum, treatment depends partly on the size and function of the right ventricle. Some children can eventually use both lower heart chambers. Others need a circulation that relies mainly on one ventricle. The coronary arteries must be examined carefully because their blood supply can affect which procedures are safe.

In pulmonary atresia with a VSD, surgery may close the VSD and create a route from the right ventricle to the pulmonary arteries. When the lung arteries are small or supplied by several abnormal vessels, reconstruction may happen in stages.

These procedures can improve circulation, but they do not always create a completely normal heart. Further catheter procedures or operations may be needed as a child grows. Heart transplantation is considered in selected situations when other treatment cannot provide adequate circulation.

Lifelong follow-up with a congenital heart specialist is important. Care may include heart imaging, rhythm monitoring, exercise advice, dental care, and medicines for specific heart or circulation problems.

- Opening the blocked pulmonary valve with a wire and balloon
- Placing a stent to keep the ductus arteriosus open
- Creating a surgical shunt to carry blood to the lungs
- Rebuilding the route between the right ventricle and pulmonary artery
- Connecting abnormal lung arteries into a more central blood supply
- Performing staged operations that direct blood from the body to the lungs

Complications

Before treatment, pulmonary atresia can cause dangerously low oxygen and reduced blood flow to the body. Prompt newborn treatment greatly changes this immediate risk.

Possible longer-term complications include:

People with a single-ventricle circulation, including those who have had a Fontan operation, may later develop liver problems, fluid build-up, rhythm disorders, or reduced heart function.

Follow-up is used to look for these changes before they cause major symptoms.

Not everyone develops these complications. Risk is shaped by the original anatomy, the type of repair, heart function, and previous procedures.

- Narrowing or blockage in a repaired vessel, shunt, or conduit
- Leaking heart valves
- Weakening of the heart muscle
- Irregular heart rhythms
- Blood clots or stroke
- Infection of the heart lining or valves, called endocarditis
- Reduced exercise capacity
- Delayed growth or developmental difficulties
- The need for repeat catheter procedures or surgery

Prevention

Pulmonary atresia usually cannot be prevented. It develops early in pregnancy, and most cases have no known cause.

General pregnancy care can reduce some health risks but cannot guarantee that a congenital heart condition will not occur. This includes managing long-term medical conditions and reviewing medicines with a healthcare professional before or during pregnancy. Prescribed medicines should not be stopped without medical advice.

If pulmonary atresia or another congenital heart condition has occurred in a family, genetic counselling may help explain whether testing is appropriate and what is known about the chance in a future pregnancy. A fetal echocardiogram may be offered during a later pregnancy based on family or medical history.

Living with it and what to expect

The course of pulmonary atresia is different for each type of heart anatomy. Most children need a procedure shortly after birth, and many need more than one operation or catheter treatment during childhood.

Some repairs include artificial tubes, patches, stents, or surgical connections. These materials

may become too small as a child grows or may narrow over time. Follow-up is needed even when a child feels well.

Many children attend school and take part in family and social activities. Exercise ability ranges from near typical to significantly limited. A congenital heart team can give activity advice based on heart function, oxygen level, rhythm, and the type of repair. Competitive or high-intensity exercise may require specific assessment.

Feeding and growth can be difficult in infancy because feeding uses energy and may increase breathing effort. Dietitian support, higher-calorie feeds, or temporary tube feeding may be used when needed.

Development should also be monitored. Some children with complex congenital heart conditions have difficulties with movement, speech, attention, or learning. Early support can help if concerns appear.

Adults still need care from a specialist in adult congenital heart disease. Childhood surgery does not remove the need for monitoring. Pregnancy may place extra strain on the heart, so planning with congenital heart and high-risk pregnancy teams is important before conception.

Dental care matters because bacteria from the mouth can sometimes enter the bloodstream. Only certain people need antibiotics like amoxicillin or doxycycline before particular dental procedures. The congenital heart team can explain whether this applies.

When to get medical help

Pulmonary atresia in a newborn is an emergency. Call emergency services if a baby who has not been assessed develops blue or grey lips or tongue, severe breathing difficulty, unusual limpness, or cannot be woken normally.

For a child or adult already diagnosed with pulmonary atresia, seek emergency help for:

Contact the congenital heart team promptly for a clear reduction in feeding or activity, worsening breathlessness, increasing swelling, persistent palpitations, unexplained fever, or a change from the person's usual oxygen level.

Routine appointments remain important when there are no new symptoms. These visits monitor heart function, blood flow, rhythm, oxygen levels, growth, and the condition of previous repairs.

- Severe or sudden breathing difficulty
- Fainting, collapse, or a seizure

- New blue or grey colouring
- Chest pain with sweating, breathlessness, or collapse
- A fast or irregular heartbeat with faintness
- Sudden weakness, facial drooping, or difficulty speaking

Common questions

Is pulmonary atresia the same as pulmonary stenosis?

No. In pulmonary stenosis, the pulmonary valve is narrowed but still has an opening. In pulmonary atresia, there is no direct opening through the valve. Pulmonary atresia usually causes a more complete blockage and needs treatment soon after birth.

Can pulmonary atresia be seen before birth?

Sometimes. A detailed pregnancy ultrasound may show abnormal heart development or blood flow. A fetal echocardiogram can examine the heart more closely. Some cases are not identified until newborn oxygen screening or symptoms appear.

Can surgery cure pulmonary atresia?

Procedures can create or improve blood flow to the lungs, and some children achieve a circulation using both ventricles. However, the heart remains structurally different and requires lifelong monitoring. Some people need staged operations and later procedures.

How long can someone live with pulmonary atresia?

Many people now survive into adulthood, but the outlook is not the same for everyone. It is affected by the form of pulmonary atresia, the coronary and pulmonary arteries, heart function, other conditions, and how well procedures work. Lifelong specialist care helps identify treatable problems.

Can a child with pulmonary atresia exercise?

Many children can be active, but safe intensity differs. Light or moderate activity may be encouraged when the circulation is stable. The congenital heart team can assess whether any limits are needed, especially after surgery or if oxygen levels, heart rhythm, or heart function are abnormal.

Will another baby in the family have pulmonary atresia?

Most families do not have another child with the same condition, but the chance can be higher than in families without congenital heart disease. Genetic counselling can review the family history, test results, and whether fetal heart imaging would be useful in a future pregnancy.

Sources

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