CONDITION

Absent pulmonary valve

Learn how absent pulmonary valve affects the heart and airways, including symptoms, diagnosis, treatment, surgery, and lifelong follow-up.

WHAT IT IS
Rare congenital heart valve condition

BODY SYSTEM
Heart and circulation

MAIN SYMPTOMS
Difficult breathing, wheezing, blue or gray skin, feeding trouble

WHAT CAUSES IT
Usually unknown, sometimes genetic

HOW IT IS DIAGNOSED
Fetal echocardiogram or echocardiogram after birth

MAIN TREATMENT
Surgical repair and breathing support

CAN IT BE CURED?
No, lifelong congenital heart care needed

IS IT AN EMERGENCY?
Sometimes, with severe trouble breathing

LONG-TERM OUTLOOK
Ranges from stable after repair to lifelong heart and airway problems

Overview

Absent pulmonary valve is a rare heart condition that develops before birth. The pulmonary valve normally controls blood flow from the heart's right ventricle into the pulmonary artery, which carries blood to the lungs.

In this condition, the pulmonary valve is missing or has only small, poorly formed pieces of tissue. The abnormal valve may be both narrowed and very leaky. Narrowing restricts forward blood flow, while leaking allows blood to move backward into the right ventricle.

Absent pulmonary valve most often occurs as part of tetralogy of Fallot , a group of related heart abnormalities. These commonly include a hole between the heart's lower chambers, called a ventricular septal defect, and obstruction between the right ventricle and pulmonary artery.

The pulmonary arteries may become unusually wide because blood moves back and forth through the abnormal valve. Enlarged pulmonary arteries can press on the nearby windpipe and breathing tubes. They may also interfere with normal airway development before birth. For some babies, airway compression causes more immediate problems than the heart defect itself.

Less commonly, absent pulmonary valve occurs without a ventricular septal defect. This form has different blood-flow patterns and may be associated with an open fetal blood vessel called a patent ductus arteriosus.

The condition ranges from mild to severe. Some babies need breathing support and surgery soon after birth. Others remain stable enough for planned treatment later. People who have undergone repair need lifelong congenital heart care because valve leakage, narrowing, heart rhythm problems, or airway symptoms can develop over time.

Symptoms

Symptoms depend on the heart's structure, the amount of pulmonary artery enlargement, and the degree of airway compression.

A fetus with absent pulmonary valve may not have outward symptoms, but the condition may be seen during a prenatal ultrasound. In severe cases, abnormal blood flow can place stress on the developing heart.

After birth, possible symptoms include:

Symptoms may worsen when a baby cries, feeds, or develops a respiratory infection. Some babies have tracheobronchomalacia , meaning the windpipe or main breathing tubes are softer than usual and can partially collapse during breathing.

Children or adults who have had a repair may later develop reduced exercise tolerance, shortness of breath, palpitations, dizziness, or swelling. These symptoms can have several causes and should be assessed rather than assumed to come from the repaired heart defect.

- Rapid, noisy, or difficult breathing
- Wheezing caused by narrowed or weakened airways
- Blue or gray coloring of the lips, tongue, or skin due to low blood oxygen
- Trouble feeding, tiring during feeds, or slow weight gain
- Sweating or breathing quickly during feeding
- Repeated chest infections

- Coughing or difficulty clearing mucus

Causes and risk factors

Absent pulmonary valve results from abnormal development of the heart and nearby blood vessels early in pregnancy. In most cases, there is no single identifiable cause. It is not generally caused by something a parent did or did not do.

The condition may occur by itself, with tetralogy of Fallot, or as part of a genetic or chromosome condition. Some affected babies have a 22q11.2 deletion , in which a small piece of chromosome 22 is missing. This deletion can also affect the immune system, calcium levels, palate, feeding, development, and other organs. Not every person with absent pulmonary valve has this deletion.

Other chromosome or genetic differences may occasionally be involved. A clinical genetics specialist can review the child's examination, family history, and test results. Genetic counseling may help families understand testing options and the chance of a heart condition occurring in a future pregnancy. That chance varies according to the underlying cause and cannot be estimated from the heart diagnosis alone.

When no genetic cause is found, the condition is usually considered a complex congenital difference involving several developmental factors.

Diagnosis

Absent pulmonary valve may be suspected during a routine pregnancy ultrasound. Findings can include enlarged pulmonary arteries, an abnormal pulmonary valve, and blood moving both forward and backward across the valve.

A fetal echocardiogram is a detailed ultrasound of the fetal heart. It can assess the pulmonary valve, pulmonary arteries, ventricular septum, blood-flow direction, and other heart structures. Because the condition can change during pregnancy, repeat fetal echocardiograms may be recommended. Prenatal diagnosis also helps the care team plan the place and timing of delivery.

After birth, clinicians consider the baby's breathing, skin color, oxygen level, pulses, and heart sounds. Pulse-oximetry screening measures blood oxygen, but a normal result does not rule out every congenital heart defect.

An echocardiogram is the main diagnostic test. It uses sound waves to show:

A chest X-ray may show enlarged pulmonary arteries, lung changes, or an enlarged heart. An electrocardiogram records the heart's electrical activity.

Cardiac CT or MRI can provide more detailed pictures of the pulmonary arteries, airways, right ventricle, and previous surgical repairs. CT is especially useful when clinicians need a clear view of airway compression. Bronchoscopy, in which a small camera is used to examine the airways, may be considered when airway narrowing or collapse needs closer evaluation.

Cardiac catheterization is not required for every patient. It may be used when the care team needs detailed pressure measurements, blood-flow information, or a catheter-based treatment.

- The structure and function of the pulmonary valve
- Narrowing or backward leakage of blood
- The size of the pulmonary arteries
- Associated defects such as tetralogy of Fallot
- Right ventricular size and pumping function

Treatment and management

Care is individualized according to the severity of breathing problems, oxygen levels, heart function, and associated abnormalities. Treatment is usually coordinated by specialists in pediatric cardiology, congenital heart surgery, intensive care, respiratory medicine, imaging, and genetics.

A fetus cannot have the valve reconstructed through routine prenatal care. Management during pregnancy focuses on monitoring and planning delivery at a center able to provide newborn heart and breathing support when needed.

After birth, some babies are stable and can feed and breathe without major support. Others need oxygen, help with feeding, or mechanical ventilation. Positive-pressure ventilation may help keep compressed or weakened airways open. Medicines may be used for specific problems, such as excess fluid or reduced heart function, but medicines cannot correct the abnormal valve or enlarged arteries.

Surgery may be needed in the newborn period when airway compression, low oxygen, heart failure, or unstable blood flow causes serious symptoms. More stable infants may have planned surgery after further growth and evaluation.

The operation depends on the exact anatomy. It may include:

There is no single operation that is right for everyone. Surgeons balance the need for reliable blood flow against the goal of limiting future valve leakage and airway compression.

Breathing symptoms may not resolve immediately after heart surgery. Airways that were compressed before birth may remain soft, and improvement can take time as the child grows. Respiratory therapy, airway-clearance measures, nutrition support, and treatment of infections may be part of recovery.

A repaired pulmonary valve or right ventricular outflow tract may later become narrowed or leaky. Some people eventually need another operation or a valve placed through a catheter. The timing is based on symptoms, imaging, exercise capacity, heart rhythm, and right ventricular size and function-not on symptoms alone.

Patients should not start, stop, or change heart medicines without discussing the plan with their cardiology team.

- Closing a ventricular septal defect
- Improving blood flow from the right ventricle to the lungs
- Placing or reconstructing a valve in the right ventricular outflow tract
- Reducing the size of enlarged pulmonary arteries to relieve airway pressure
- Repositioning pulmonary arteries in selected cases

Complications

Possible complications are related to both the original condition and previous treatment. They include:

Infective endocarditis is an infection involving the heart's inner lining or a valve. Good dental care helps lower this risk. Antibiotics like amoxicillin or doxycycline before dental procedures are recommended only for certain high-risk heart conditions or repair situations. A congenital cardiologist or dentist should confirm whether they are needed.

Pregnancy places extra demands on the heart. Adults considering pregnancy should meet with an adult congenital heart specialist before becoming pregnant so that heart and valve function, medications, and pregnancy risks can be reviewed.

- Persistent airway compression or tracheobronchomalacia
- Recurrent respiratory infections or difficulty clearing mucus
- Ongoing pulmonary valve leakage or narrowing

- Enlargement or reduced function of the right ventricle
- Abnormal heart rhythms
- Reduced exercise capacity
- Narrowing or leakage in a surgical conduit or replacement valve
- Need for additional catheter procedures or surgery

Follow up

Lifelong follow-up with a congenital cardiology team is important, even when a person feels well. Adults should ideally receive care from a cardiologist trained in adult congenital heart disease.

Routine visits may include an examination, echocardiogram, electrocardiogram, heart rhythm monitoring, exercise testing, or cardiac MRI. The schedule depends on the repair, valve function, right ventricular size, airway symptoms, and previous test results.

Regular dental care and daily brushing and flossing support heart health. Physical activity is often encouraged, but safe intensity varies. The cardiology team can provide advice based on heart function, rhythm history, oxygen level, and planned sports.

Contact the care team promptly for new or worsening shortness of breath, reduced feeding, poor weight gain, increasing fatigue, frequent palpitations, dizziness, swelling, fever after a recent heart procedure, or signs of a respiratory infection that are affecting breathing.

Seek emergency care for severe trouble breathing, blue or gray lips or tongue, collapse, unresponsiveness, chest pain with fainting, or a sustained very fast heartbeat accompanied by weakness or breathing difficulty. Follow any emergency plan provided by the congenital heart team.

Sources

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