CONDITION

Tetralogy of fallot

Tetralogy of Fallot is a combination of four heart defects present from birth that can reduce oxygen reaching the body.

WHAT IT IS
Four linked heart defects present from birth

BODY SYSTEM
Heart and circulation

MAIN SYMPTOMS
Bluish skin, breathlessness, poor feeding, slow growth

WHAT CAUSES IT
Heart development before birth, usually unknown

HOW IT IS DIAGNOSED
Echocardiogram, sometimes fetal echocardiogram

MAIN TREATMENT
Surgical repair and lifelong specialist follow-up

CAN IT BE CURED?
No - repair leaves lifelong heart risks

IS IT AN EMERGENCY?
Sometimes - severe blue spells or breathing trouble

LONG-TERM OUTLOOK
Most reach adulthood after repair, with lifelong follow-up

Overview

Tetralogy of Fallot, also called Fallot's tetralogy, is a congenital heart condition. Congenital means it develops before birth.

It involves four connected differences in the heart:

Together, these differences allow oxygen-poor blood to mix with oxygen-rich blood. They may also reduce blood flow to the lungs, where blood normally collects oxygen. This can cause low blood oxygen and a blue, gray, or dusky appearance known as cyanosis.

The effects differ from person to person. A baby with severe narrowing may become blue soon after birth. A child with milder narrowing may not have obvious cyanosis at first.

Tetralogy of Fallot requires specialist care. Surgery can repair the main structural problems,

but the heart is not made completely typical. Lifelong follow-up is needed to check heart valves, heart function, rhythm, and blood flow.

- A hole between the heart's lower chambers, called a ventricular septal defect
- Narrowing where blood leaves the heart for the lungs, called right ventricular outflow obstruction or pulmonary stenosis
- An aorta positioned over the hole instead of arising only from the left side
- Thickening of the right lower heart chamber, called right ventricular hypertrophy

Symptoms

Symptoms usually begin during infancy, although the timing depends mainly on how severely blood flow to the lungs is restricted.

Possible signs include:

Cyanosis can be harder to see on darker skin. Changes may be clearer on the lips, tongue, gums, palms, soles, or around the eyes.

Some babies and young children have sudden episodes of very low oxygen called hypercyanotic spells or "tet spells." A child may become much bluer, breathe rapidly, become distressed, or go limp. Spells can happen during crying, feeding, waking, or passing stool. Older children may squat because this position can briefly improve blood flow to the lungs.

Not every feeding problem, color change, or breathing symptom is caused by tetralogy of Fallot. These symptoms still need medical assessment, especially in a baby.

- Blue, gray, or dusky lips, tongue, skin, or nail beds
- Fast or difficult breathing
- Tiring, sweating, or becoming breathless during feeding
- Poor weight gain or slower growth
- Irritability or unusual sleepiness
- A heart murmur, which is an extra sound heard through a stethoscope
- Rounded fingertips or toes, called clubbing, after long-term low oxygen

Causes and risk factors

Tetralogy of Fallot develops when the heart is forming early in pregnancy. In most cases, there

is no single known cause. Nothing a parent did or did not do can usually explain why it happened.

Some cases are associated with a change in a chromosome or gene. One example is a missing section of chromosome 22, known as a 22q11.2 deletion. Tetralogy of Fallot can also occur with Down syndrome or other genetic conditions.

Having a close relative with a congenital heart condition may increase the chance of one occurring, but tetralogy of Fallot usually does not follow a simple pattern of inheritance.

Certain health conditions or exposures during pregnancy may be linked with congenital heart defects in general. However, a risk factor does not prove that it caused an individual child's condition.

Diagnosis

Tetralogy of Fallot may be suspected during a routine pregnancy ultrasound. A fetal echocardiogram can then provide detailed ultrasound images of the baby's heart before birth. Some cases are not detected until after delivery.

After birth, low oxygen on newborn pulse-oximetry screening, cyanosis, or a heart murmur may lead to further testing. Pulse oximetry uses a small sensor on the hand or foot to estimate oxygen in the blood. A normal screening result does not rule out every case.

An echocardiogram usually confirms the diagnosis. This painless ultrasound shows the heart's structure, the hole between the chambers, and blood flow toward the lungs.

Other tests may be used to understand the heart or plan treatment. These can include:

- An electrocardiogram to record the heart's electrical activity
- A chest X-ray
- Cardiac MRI or CT imaging
- Cardiac catheterization, in which a thin tube is passed through a blood vessel to measure pressures or perform a procedure
- Genetic testing when a related genetic condition is suspected

Treatment and management

Treatment is planned by a congenital heart team. The timing and type of treatment depend on oxygen levels, the anatomy of the heart, and the child's overall health.

A newborn with severely restricted lung blood flow may need urgent hospital treatment. Medicine can sometimes keep a fetal blood vessel called the ductus arteriosus open temporarily, allowing more blood to reach the lungs.

Most children need surgery. Complete repair generally involves closing the hole between the lower chambers and widening the path from the right ventricle to the lungs. The surgeon may also repair or widen the pulmonary valve and nearby pulmonary arteries.

Some babies first need a temporary procedure to improve lung blood flow. This may involve creating a surgical shunt or placing a stent in a blood vessel or the narrowed outflow pathway. Complete repair is performed later when appropriate.

After repair, follow-up usually includes examinations, oxygen checks, electrocardiograms, and heart imaging. Some people later need another procedure because of:

A replacement pulmonary valve may be placed through surgery or, for some people, through a catheter procedure. Treatment is based on test findings and symptoms rather than age alone.

Good dental care is also important because bacteria from the mouth can sometimes infect abnormal heart tissue or artificial material. Only certain people need preventive antibiotics like amoxicillin or doxycycline before particular dental procedures. The cardiology and dental teams can explain whether this applies.

- Leakage or narrowing of the pulmonary valve
- Enlargement or weakening of the right ventricle
- A remaining hole or obstruction
- Abnormal heart rhythms
- Changes involving the aorta or another heart valve

Complications

Without effective treatment, low oxygen and reduced blood flow to the lungs can interfere with growth and place strain on the heart and other organs.

Surgical repair greatly improves blood flow, but later complications remain possible. These include pulmonary valve leakage, right-sided heart enlargement, reduced pumping function, abnormal rhythms, exercise limitations, and infection of the heart lining or valves, called endocarditis.

Rarely, low oxygen or abnormal blood flow can contribute to blood clots, stroke, or a brain abscess. Regular follow-up aims to identify changes before they cause major symptoms.

Anyone who has had heart surgery may also experience anxiety, difficulty concentrating, or concerns about scars and physical activity. Developmental, educational, or mental health support can help when these issues affect daily life.

Prevention

Tetralogy of Fallot usually cannot be prevented. It forms early in pregnancy, and the cause is often unknown.

Before pregnancy, general steps that support healthy fetal development include reviewing medicines with a qualified health professional, controlling conditions such as diabetes, avoiding smoking and non-medical drugs, and making sure vaccinations are current. These steps cannot guarantee prevention.

Genetic counselling may be useful when a parent has congenital heart disease, a previous child has a heart defect, or a genetic condition has been identified. It can explain possible inheritance and testing options without predicting with certainty what will happen in a pregnancy.

Living with it and what to expect

Most children who have a successful repair grow into adulthood. They may attend school, work, travel, and take part in many everyday activities. Individual experiences differ according to the original anatomy, heart function, later procedures, and other health conditions.

Tetralogy of Fallot does not usually disappear after surgery. Over months and years, valve leakage or narrowing can gradually change the size and function of the right side of the heart. Rhythm problems may also develop. These changes may cause no symptoms at first, which is why routine appointments remain important.

Physical activity is often encouraged, but the safest type and intensity depend on heart function and rhythm. Some people have no major restrictions. Others need to avoid high-intensity or competitive exercise.

An adult with repaired tetralogy of Fallot should remain under the care of a cardiologist trained in adult congenital heart conditions. Losing contact with specialist care can delay recognition of problems that are easier to manage early.

Pregnancy may be possible, but it places extra demands on the heart. Anyone considering pregnancy should have a pre-pregnancy assessment with an adult congenital heart and pregnancy team. The review can address heart function, medicines, inherited risks, and the

safest plan for care.

When to get medical help

Call emergency services for a baby or child who suddenly becomes much bluer or grayer than usual, has severe breathing difficulty, faints, has a seizure, becomes limp, or is difficult to wake.

A suspected tet spell is an emergency, particularly if the color change or breathing problem is severe or does not settle quickly. While waiting for help, keep the child calm. If previously taught to do so, gently bring an infant's knees toward the chest. Do not delay emergency care to try home measures.

Emergency assessment is also needed for severe chest pain, collapse, or a sustained fast or irregular heartbeat accompanied by breathlessness, weakness, or fainting.

Seek prompt medical advice for new or increasing cyanosis, faster breathing, feeding difficulty, poor weight gain, reduced activity, swelling, repeated palpitations, or a clear decline in exercise ability.

Routine follow-up is different from urgent care. Keep planned congenital heart appointments even when you or your child feels well. Contact the heart team routinely if an appointment has been missed, activity advice is unclear, or dental, travel, school, work, or pregnancy planning needs to be discussed.

Common questions

Is tetralogy of Fallot genetic?

Sometimes. It can occur with chromosome or gene changes, including a 22q11.2 deletion, but many people have no identified genetic cause. Genetic testing may be offered when there are other physical findings, developmental concerns, or a family history of congenital heart conditions.

Can tetralogy of Fallot be completely cured?

Surgery repairs the main defects and can greatly improve oxygen levels. However, it does not make the heart entirely typical. Valve problems, changes in heart function, or abnormal rhythms can appear later, so lifelong follow-up remains necessary.

Why does a baby with tetralogy of Fallot turn blue?

Blueness happens when blood reaching the body contains less oxygen than usual. In tetralogy of Fallot, narrowing toward the lungs can cause oxygen-poor blood to cross the hole inside the

heart and enter the aorta. Severe or sudden color changes need urgent assessment.

Can a child with repaired tetralogy of Fallot exercise?

Many children can be active and may take part in most school activities. Safe limits depend on heart function, valve leakage, heart rhythm, and whether further treatment is planned. Activity advice should be reviewed as the child grows.

Can tetralogy of Fallot come back after surgery?

The original combination of defects does not grow back. However, repaired areas and valves can change over time. Pulmonary valve leakage, renewed narrowing, or heart rhythm problems may require monitoring or another procedure.

What is life expectancy after tetralogy of Fallot repair?

Most people who receive modern repair survive into adulthood. Long-term outlook is influenced by the severity of the original anatomy, heart and valve function, rhythm problems, other health conditions, and access to lifelong specialist care. An exact lifespan cannot be predicted for one person.

Sources

- Centers for Disease Control and Prevention. About Tetralogy of Fallot
- American Heart Association. Tetralogy of Fallot
- National Health Service. Tetralogy of Fallot
- 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