CONDITION

Truncus arteriosus

Truncus arteriosus is a birth heart defect in which one large artery leaves the heart instead of two.

WHAT IT IS
Congenital heart defect with one shared artery leaving the heart

BODY SYSTEM
Heart and circulation

MAIN SYMPTOMS
Fast breathing, poor feeding, sweating, blue-gray skin

WHAT CAUSES IT
Heart development difference before birth, usually unknown

HOW IT IS DIAGNOSED
Fetal or postnatal echocardiogram

MAIN TREATMENT
Early heart surgery and lifelong specialist follow-up

CAN IT BE CURED?
No - surgery repairs circulation, but lifelong monitoring remains

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

LONG-TERM OUTLOOK
Many reach adulthood after repair, often needing further procedures

Overview

Truncus arteriosus is a serious heart condition present from birth. It forms during early pregnancy, while the baby's heart and major blood vessels are developing.

A typical heart has two large arteries leaving it. The pulmonary artery carries blood to the lungs, while the aorta carries oxygen-rich blood to the body. In truncus arteriosus, there is one shared artery, called the arterial trunk, instead of two separate arteries.

There is also usually a hole between the heart's two lower pumping chambers. This is called a ventricular septal defect, or VSD. Blood with more oxygen and blood with less oxygen mix through this opening and enter the shared artery.

Too much blood may then flow to the lungs. The heart has to work harder, and the body may

receive blood that contains less oxygen than usual. Without treatment, this can lead to heart failure and lasting damage to the blood vessels in the lungs.

Surgery is needed to create separate routes to the lungs and body. Most babies are treated early in infancy.

Symptoms

Symptoms often appear during the first days or weeks after birth. Their timing and severity can differ according to how much blood reaches the lungs and whether other heart abnormalities are present.

Possible signs include:

Blue or gray coloring is called cyanosis. On darker skin, it may be easier to notice around the lips, tongue, gums, or nail beds.

Heart failure in a baby does not mean that the heart has stopped. It means the heart cannot pump efficiently enough to meet the body's needs. In infants, it commonly causes rapid breathing, feeding difficulty, sweating, and slow weight gain.

Some babies are diagnosed before symptoms begin because a heart abnormality was seen during pregnancy or found shortly after birth.

- Fast or difficult breathing
- Blue or gray lips, tongue, or skin
- Tiring, sweating, or breathing hard during feeds
- Taking a long time to feed or drinking only small amounts
- Poor weight gain
- Unusual sleepiness or reduced activity
- A fast heartbeat

Causes and risk factors

Truncus arteriosus happens when the developing arterial trunk does not divide into a separate aorta and pulmonary artery. The reason this occurs is usually unknown. It is not caused by something a parent did during ordinary day-to-day activities.

Some people with truncus arteriosus have a genetic or chromosome difference. One important association is a missing small section of chromosome 22, called a 22q11.2 deletion. This

deletion can also affect the immune system, calcium levels, palate, learning, and development, although its effects differ widely.

Most cases do not follow a simple inherited pattern. Having one child with a congenital heart condition may change the chance of a heart condition in a future pregnancy, but the individual chance depends on the family history and any genetic findings.

Pre-existing diabetes and certain medicine or substance exposures during pregnancy can increase the overall chance of congenital heart defects. However, these factors do not explain most cases of truncus arteriosus.

Diagnosis

Truncus arteriosus may be suspected during a routine pregnancy ultrasound. A fetal echocardiogram can then examine the baby's heart in more detail. This is a specialized ultrasound that shows the heart's structure, rhythm, and blood flow.

After birth, a clinician may notice low blood oxygen, a heart murmur, rapid breathing, or feeding difficulty. A murmur is an extra sound caused by blood moving through the heart.

An echocardiogram usually confirms the diagnosis. It can show:

A chest X-ray, electrocardiogram, oxygen measurements, CT scan, or MRI may provide further information. Cardiac catheterization, in which a thin tube is passed through a blood vessel to the heart, is not always needed for the first diagnosis. It may be used later to measure pressures or plan another procedure.

Genetic testing may be offered, particularly for a 22q11.2 deletion. Genetic counseling can help families understand what a result may mean for the child and other relatives.

- The single arterial trunk and VSD
- How the lung arteries arise
- How well the heart pumps
- Whether the valve in the shared artery leaks or is narrowed
- Other heart or aortic arch abnormalities

Treatment and management

Truncus arteriosus requires surgery. Medicines can support a baby before surgery, but they cannot separate the shared artery or close the VSD.

Before the operation, care may include help with breathing, treatment for fluid buildup, and additional calories or feeding support. The exact approach is based on the baby's circulation, growth, and any other health concerns.

During repair, the surgeon generally:

The original shared artery then functions mainly as the aorta, carrying blood to the body. The valve at its base, called the truncal valve, may also need repair if it leaks significantly.

The tube between the right ventricle and pulmonary arteries is called a conduit. It cannot grow as a child grows. It may also become narrowed or develop a leaking valve over time. Because of this, many people need the conduit replaced or treated by catheter procedures later in life.

After recovery, lifelong follow-up with a congenital heart specialist is important. Appointments may include an examination, echocardiogram, electrocardiogram, exercise testing, MRI, or other imaging. Follow-up looks for changes before they cause major symptoms.

- Closes the VSD so blood from the left ventricle reaches the shared artery
- Separates the pulmonary arteries from the shared trunk
- Connects the right ventricle to the pulmonary arteries with a tube containing a valve

Complications

Before repair, excess blood flow to the lungs can cause heart failure and damage the small lung arteries. Early treatment reduces this risk.

Possible problems after repair include:

Repeat operations or catheter procedures are common because repaired structures can change and conduits wear out or become too small.

Some children have feeding, growth, movement, speech, attention, or learning difficulties. These may relate to the heart condition, associated genetic differences, early illness, or several factors together. Developmental assessment and early support can help when concerns arise.

- Narrowing or leakage in the right ventricle-to-pulmonary artery conduit
- Leakage or narrowing of the truncal valve
- Enlargement of the aorta
- Reduced heart-pumping function
- Abnormal heart rhythms
- Increased pressure in the lung arteries

- Infection involving the heart lining or a valve, called endocarditis

Prevention

Most cases of truncus arteriosus cannot be prevented. It develops very early in pregnancy, and there is often no identifiable cause.

Before pregnancy, managing diabetes and reviewing medicines with a healthcare professional can reduce some pregnancy risks. Do not stop a prescribed medicine without medical advice, because untreated illness may also be harmful.

If truncus arteriosus or another congenital heart condition has occurred in the family, genetic counseling may help explain possible causes and future pregnancy options. A fetal echocardiogram can look for heart abnormalities during a pregnancy, but it does not prevent them.

Living with it and what to expect

Recovery after infant heart surgery takes time. Feeding and growth often improve as circulation becomes more efficient, although some babies continue to need nutrition or developmental support.

Over the following years, the repair needs regular monitoring. A child may feel well even when a conduit or valve is gradually becoming narrowed or leaky. Routine appointments therefore remain important even when there are no symptoms.

Many children attend school, take part in family life, and become active adults. Exercise advice is individualized. Some people can participate in a wide range of activities, while others need limits because of valve problems, abnormal rhythms, heart function, or lung pressure. A congenital heart team can provide guidance as these findings change.

Good dental care helps reduce the chance of bacteria entering the bloodstream and infecting heart tissue. Antibiotics like amoxicillin or doxycycline before certain dental procedures are recommended only for people with specific heart findings or repair histories. The cardiology team can explain whether this applies.

Adults should continue care with a specialist in adult congenital heart conditions. Pregnancy can place extra strain on the heart and may carry added risks. Anyone considering pregnancy should have their heart function, valves, conduit, aorta, and medicines reviewed beforehand by a congenital heart and pregnancy team.

The long-term course cannot be predicted exactly. Many people survive into adulthood after repair, but further procedures and lifelong monitoring are expected parts of care.

When to get medical help

Call emergency services if a baby, child, or adult has:

For a baby with known or suspected truncus arteriosus, seek prompt medical assessment for worsening rapid breathing, repeated feeding difficulty, heavy sweating with feeds, fewer wet diapers, unusual sleepiness, or poor weight gain.

After repair, contact the heart team promptly about a clear drop in exercise ability, new fainting, persistent racing or irregular heartbeats, increasing swelling, or worsening shortness of breath.

Routine congenital heart appointments are needed even when the person feels well. These visits are different from urgent care: they monitor slow changes in valves, conduits, heart function, and the aorta.

- Severe difficulty breathing
- Blue or gray lips, tongue, or skin that is new or worsening
- Collapse, fainting, or difficulty waking
- Sudden severe weakness or loss of responsiveness

Common questions

Can truncus arteriosus close or improve on its own?

No. The single artery and associated hole do not develop into normal separate circulation after birth. Surgery is needed to create separate blood-flow routes to the lungs and body.

Can a baby survive truncus arteriosus?

Yes, many babies survive after surgical repair and later reach adulthood. Truncus arteriosus can become life-threatening without treatment, so babies need early assessment and specialist care. The outlook is influenced by heart anatomy, valve function, genetic conditions, and complications.

Is truncus arteriosus genetic?

Sometimes. It is associated with genetic changes, especially a 22q11.2 deletion, but many babies have no identified genetic cause. Testing and genetic counseling may provide more

specific information for a family.

Can truncus arteriosus be detected before birth?

It can sometimes be seen on a pregnancy ultrasound and assessed with a fetal echocardiogram. It is not detected before birth in every case. A normal routine ultrasound cannot exclude every heart condition.

Will my child need another heart operation?

Further procedures are common. The conduit placed during the first repair does not grow with the child and can become narrowed or leaky. Valves, the aorta, or other parts of the repair may also need treatment. Timing is based on symptoms, imaging, heart function, and pressure measurements.

Can someone with repaired truncus arteriosus exercise?

Many people can be physically active, but safe intensity differs from person to person. Current heart function, valve leakage, conduit narrowing, heart rhythm, and lung pressure all matter. Regular assessment provides more useful guidance than the original diagnosis alone.

Sources

- Centers for Disease Control and Prevention. About Truncus Arteriosus
- American Heart Association. Truncus Arteriosus
- MSD Manual Professional Edition. Persistent Truncus Arteriosus
- Stout KK, Daniels CJ, Aboulhosn JA, et al. 2018 AHA/ACC Guideline for the Management of Adults With Congenital Heart Disease. Journal of the American College of Cardiology . 2019;73(12):e81-e192
- McDonald-McGinn DM, Hain HS, Emanuel BS, et al. 22q11.2 Deletion Syndrome. GeneReviews, University of Washington, Seattle
- Wilson WR, Gewitz M, Lockhart PB, et al. Prevention of Viridans Group Streptococcal Infective Endocarditis: A Scientific Statement From the American Heart Association. Circulation . 2021;143:e963-e978