Heart abnormality birth defects

Summary

- Sometimes, during fetal development, the heart and blood vessels do not grow properly.
- The passage of blood inside the heart or vessels may be blocked, the blood may travel abnormally through the heart, or parts of the heart itself may be underdeveloped.
- In eight out of 10 cases, the cause is unknown.

Approximately one in every 100 babies is born with a heart defect. This is called a congenital heart defect (CHD). Some defects are mild and cause no significant disturbance to the way the heart functions. However, more than half of all children with CHD have a condition that is serious enough to require treatment.

How the heart works

The heart is a double pump made up of four chambers. Its role is to provide the body with oxygen. The heart takes blood through these stages:

- It takes blood that is low in oxygen into the right collecting chamber (right atrium) and squeezes it into the right pumping chamber (right ventricle).
- Blood is then pumped through an artery to the lungs to pick up oxygen.
- From the lungs, blood packed with oxygen goes into the left collecting chamber (left atrium).
- It then goes into the lower left pumping chamber (left ventricle) and into an artery (the aorta), where it starts its journey around the body again.

Heart defects can develop while in the uterus

If the heart and blood vessels fail to grow properly during fetal development, this may cause:

- Blockages that prevent blood flow around the heart and arteries
- Blood to travel abnormally through the heart (for instance, across ‘holes’ in the walls that separate the two sides of the heart)
- Parts of the heart itself to be underdeveloped.

Acquired heart disease

Some children develop a heart problem after an illness. Illnesses that can lead to a heart problem include myocarditis (inflammation of the heart muscle), cardiomyopathy (disease of the heart muscle), rheumatic heart disease (a disease that may follow streptococcal bacterial infection) and Kawasaki disease (a disease with fever, rash and swollen lymph glands that may affect the heart). These are called acquired heart disorders.

Some children with a genetic condition called Noonan syndrome can also have heart abnormalities.

Causes of heart defects

In around eight out of 10 cases, the reason for the congenital heart defect is unknown. Some of the known causes of CHD include:

- **Genes** – 20 per cent of cases have a genetic cause.
- **Other birth defects** – a baby affected by certain birth defects, such as Down syndrome, is more likely to have malformations of the heart.
- **Maternal illness** – illness of the mother during pregnancy (for example, rubella – now rare) may increase the risk of congenital heart disorders.
- **Medication and drugs** – medication (over-the-counter or prescription) or illicit drugs taken by the mother.
during pregnancy may increase the risk of congenital heart disorders.

- **Alcohol** – a mother who drinks large amounts of alcohol during pregnancy may increase the risk of congenital heart disorders.
- **Maternal health** – factors such as unmanaged diabetes and poor nutrition during pregnancy may increase the risk.
- **Maternal age** – babies of older women are more likely to have a birth defect than babies of younger women.

**Symptoms of heart defects**

Heart problems may cause a variety of symptoms in babies, including:

- **Shortness of breath** – this might be due to a build-up of fluid in the lungs.
- **Difficulty feeding** – this may occur because the baby is having trouble breathing.
- **Blue lips and skin** – some heart defects can lead to mixing of blue (oxygen depleted) blood from the veins and the red (oxygen rich) blood from the lungs. This may produce a bluish colour of the lips and skin, called ‘cyanosis’.

**Five common heart defects**

Some common congenital heart defects include:

- Ventricular septal defect
- Transposition of the great vessels
- Coarctation of aorta
- Tetralogy of Fallot
- Hypoplastic left heart syndrome.

**Ventricular septal defect**

Ventricular septal defect is the most common congenital heart defect in Victoria. The incidence is around one in every 344 births.

The ventricles are the two lower pumping chambers of the heart. Ventricular septal defect means there is a hole in the wall between the ventricles. This hole lets oxygenated and deoxygenated blood mix.

In a child with this condition, the heart has to work much harder than normal, and may enlarge. Symptoms include breathlessness, difficulty feeding, increased heart rate and failure to grow at the expected rate. Depending on the severity of the condition, the child may develop congestive heart failure and have an increased risk of developing pneumonia.

**Treatment**

Treatment for ventricular septal defect depends on the severity of the defect. If the hole is small, it may heal by itself with time, and no treatment – other than careful monitoring – is needed. Large defects, with symptoms in infancy, may require open-heart surgery. Closing the hole (usually with a ‘patch’) allows the blood to circulate normally, relieving strain on the heart. In some cases, generally in older children when the hole has not closed and is still causing strain on the heart, a catheter procedure may be possible and the defect closed with an implantable device.

**Transposition of the great vessels**

The incidence of transposition of the great vessels in Victoria is around one in every 2,000 births.

Normally, blood from the heart’s right ventricle is taken to the lungs by the pulmonary artery. Blood from the left ventricle is taken to the body by the aorta (the main artery of the body).

Transposition of the great vessels means this situation is reversed, with the pulmonary artery attached to the left ventricle and the aorta to the right. Oxygenated blood is pumped back to the lungs instead of around the body.

This defect can be fatal in the early weeks of life if it is not treated. Some babies survive longer if there is a hole in the partition between the upper or lower chambers of the heart, allowing the blood to mix. The main symptom of...
transposition of the great vessels is cyanosis, the blue colouring to the skin caused by lack of oxygen.

**Treatment**
Treatment for transposition of the great vessels involves a procedure called balloon septostomy. This may need to be carried out to enlarge the small opening between the atria that is normally present at birth, so that more oxygenated blood can reach the body. Subsequent surgery will be organised, usually in the first two weeks of life, to reconnect the arteries normally.

**Coarctation of the aorta**
The incidence of coarctation of the aorta in Victoria is around one in every 2,000 births. The aorta is the main artery of the body, and ‘coarctation’ means it is narrowed or pinched, usually in the upper chest. This means that blood pressure in the lower body is lower than normal.

This condition often leads to serious symptoms in the early weeks of life. Symptoms typically occur in the first week of life and include breathlessness and difficulty breathing, and may include collapse.

Less commonly, this disorder may not be diagnosed in infancy and may be discovered much later in life during investigations for high blood pressure.

**Treatment**
Surgery will be needed to treat coarctation of the aorta if the condition is severe and causes symptoms in early infancy. In older children, the narrowed section can sometimes be stretched open with special balloons or ‘stents’.

**Tetralogy of Fallot**
The incidence of tetralogy of Fallot in Victoria is around one in every 2,000 births. When a child has tetralogy of Fallot, their heart is affected by four main defects:

- A ventricular septal defect
- Blockage of blood flow out of the right ventricle
- Thickening of the wall of the right ventricle
- Displacement of the aorta towards the right ventricle.

These four defects allow oxygenated and deoxygenated blood to mix inside the heart. The main symptom is cyanosis (blue colouring) that develops in the early weeks or months of life.

**Treatment**
Treatment for tetralogy of Fallot requires surgery to close the septal defect and remove obstruction to blood flow out of the right ventricle. Some infants with severe symptoms in early life may have a preliminary ‘shunt’ operation, which increases blood flow to the lungs and relieves cyanosis, but does not correct the underlying defect.

**Hypoplastic left heart syndrome**
The incidence of hypoplastic left heart syndrome in Victoria is around one in every 4,000 births. In this condition, the entire left side of the heart, including valves and blood vessels, is underdeveloped. Without prompt treatment, the baby is likely to die within days or weeks of birth. Symptoms include a grey complexion and severe breathing difficulties.

**Treatment**
Hypoplastic left heart syndrome will require surgery. The techniques involved include a ‘Norwood’ operation, which allows the right ventricle to become the pumping chamber that supplies the body and lungs. The surgery is difficult and involves high risk. At least two further operations in early childhood will be needed to achieve normal heart function.

**Children with heart defects need special care to prevent infections**
All children with heart defects should be given antibiotics when they have teeth extracted or when they have other surgery that involves the mouth, nose, intestinal organs or genito-urinary systems. Bacteria may enter the bloodstream during these procedures and can cause serious infection (known as infective endocarditis) in the abnormal part of the heart.

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All medications should be checked with the pharmacist, family doctor or cardiologist. The usual immunisations should be given at the normal times after advice from the doctor.

**Where to get help**

- Your doctor
- Paediatrician
- [HeartKids Victoria Inc.](tel:03 9513 9030) Tel. (03) 9513 9030 heartkids@heartkids.org.au
- [Royal Children’s Hospital Cardiology Department](https://www.royalchildrens.org.au/)

**Things to remember**

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