

# Heart conditions

Most heart conditions have a genetic contribution.

For some of them, the main problem is an alteration in one gene. Conditions like this include:

- Familial hypercholesterolaemia
- Some types of cardiomyopathy, such as hypertrophic cardiomyopathy
- Some types of arrhythmias.

For most heart conditions, however, the disorder is associated with having several genes that indicate a high likelihood of getting the condition, together with lifestyle and environment. Conditions like this include:

- High blood pressure
- Heart attacks
- Most congenital heart defects.

If you or someone in your family has a heart condition, including problems with high blood pressure or high cholesterol, then it is worth looking at your family history (see fact sheet on '*Your family history*') and discussing it with your doctor.

In some cases, you and other members of your family might be asked to see doctors, be examined and have blood or other tests to look for potential signs of heart disease.

Only a small proportion of people with heart disease will need to see a genetic counsellor or have any form of genetic testing.

## **Familial hypercholesterolaemia**

People with familial hypercholesterolaemia have very high levels of cholesterol in their blood throughout their life. The walls of the arteries are affected in childhood and heart disease strikes at an earlier age than usual if the condition is not treated.

Without treatment, about half of men with familial hypercholesterolaemia will have heart disease by the time they are 50 and all will have heart disease by the time they are 70. In a very small number of affected men, heart disease comes on in their 20s. Women are affected too, although usually not as severely.

A number of different genetic alterations can be involved – they all follow a pattern of autosomal dominant inheritance in families (see fact sheet on '*How do genetic conditions occur?*').

While familial hypercholesterolaemia can occur in any family, it is more common in people whose ancestry is Lebanese, Dutch Afrikaaner or French Canadian.

## High blood pressure

In most people, high blood pressure is caused by a number of factors such as diet, weight and stress. Genetics also plays a part.

There are likely to be a number of different genes involved in high blood pressure. However, there is no specific test for a genetic alteration that would determine if a person had a high likelihood of having high blood pressure.

## Congenital heart defect

Heart defects that exist from birth are known as congenital heart defects. Congenital heart defects affect nearly 1 in 100 babies. Some of these heart defects are very serious – about a third of affected babies need an operation in the first year of life – while others are not so serious. Some get better with time.

Usually, congenital heart defects are caused by a combination of factors, including genetic alterations. But, usually, there is no single genetic cause and no genetic test that can be done.

People with congenital heart defects in the family should tell their doctors and consider genetic counselling to find out their risk of having a baby with the condition. Pregnant women with congenital heart disease in the family can have a very detailed ultrasound in mid-pregnancy to see whether or not their child is affected.

## Cardiomyopathy

The term cardiomyopathy covers a range of conditions in which the heart muscle does not work as well as it should.

There are two main types of cardiomyopathy – hypertrophic and dilated.

Hypertrophic cardiomyopathy means the muscles of the heart become thicker and do not work as well as they should. Many people with hypertrophic cardiomyopathy have an underlying genetic alteration and, in most cases, there is an autosomal dominant pattern of inheritance (see fact sheet on '[How do genetic conditions occur?](#)').

Dilated cardiomyopathy means the muscles of the heart have stretched and loosened, something like a balloon that has been over-inflated. Most people with dilated cardiomyopathy do not have an underlying genetic alteration – it is usually caused by other factors such as viral infection.

No genetic testing is available yet in Australia for cardiomyopathy, except in research studies. The immediate family of people with hypertrophic cardiomyopathy or unexplained dilated cardiomyopathy should see their doctors about having their heart tested.

## Arrhythmias

An arrhythmia is a disturbance of the normal heart rhythm. Most people with arrhythmias do not have a known genetic alteration. But there are two types of arrhythmias that have an underlying genetic component – the long QT syndrome and a condition known as Brugada disease.

These conditions can cause arrhythmias, fainting spells or sudden death in children or young adults. Close relatives may need to see a cardiologist to check whether they also have the condition. If a gene alteration is identified in an affected person, close family members may wish to have testing to predict whether they are at risk of the same condition.

### Contacts and further information

- Your local genetic service, which you can contact through your nearest community health centre, public hospital or health department.
- National Heart Foundation at <http://www.heartfoundation.org.au>
- Better Health Channel at <http://www.betterhealth.vic.gov.au>
- Australasian Genetic Alliance at <http://www.australasiangeneticalliance.org.au>
- MyDr at <http://www.mydr.com.au>
- The Centre for Genetics Education at <http://www.genetics.edu.au>
- HealthInsite at <http://www.healthinsite.com>
- MedicineNet at <http://www.medicinenet.com>
- For other related fact sheets, you can contact the Gene Technology Information Service on **free call Australia-wide 1800 631 276** or email [gtis-australia@unimelb.edu.au](mailto:gtis-australia@unimelb.edu.au) or visit Biotechnology Australia's website at <http://www.biotechnology.gov.au>