

# Familial hypercholesterolaemia (FH)

## Inherited risk

Some families have a much stronger inherited risk of developing cardiovascular disease. This may be because of problems with their heart rhythms, heart muscle function or risk factors for cardiovascular disease (things that increase your chance of getting heart disease, such as high blood cholesterol). There is treatment for nearly all of these problems.

If you or a family member are diagnosed with cardiovascular disease (such as having had a heart attack or stroke) before the age of 60, it is very important that you ask your doctor to measure your risk factors. The rest of your family (especially your brothers, sisters and children) also need to have their risk factors measured.

This information sheet describes one special case of inherited risk, namely inherited high blood cholesterol which is known as familial hypercholesterolaemia (FH).

## What is FH?

FH is one of the most common, potentially fatal inherited conditions. When FH is present, your body does not remove enough cholesterol from your blood. This causes high blood cholesterol levels and early heart disease in some families. About one in every 250 Australians is affected. The good news is that FH can be successfully treated.

## Why do people get FH?

A pair of genes controls the removal of cholesterol from your blood. If one of these genes is faulty, then the removal process only works half as well. This means that your blood cholesterol rises to about twice its usual level, even if you follow a healthy, balanced diet low in saturated fat and lead a healthy lifestyle. There is approximately a 50% chance that the faulty gene will be passed on to the children of people who have FH.

## How do I know if I have FH?

Most people with FH don't know that they have it, because they don't experience any symptoms. If you think that you may have or be at risk of FH, talk to your doctor. Your doctor can do tests to find out if you have FH. People who have FH have higher blood cholesterol levels from birth and can be diagnosed at any age.

With an approximately 50% chance that FH will be passed from parent to child, it is important you are aware of your family history of heart disease and talk to your doctor about whether you need to be tested.

FH does not depend on your body weight, if you are male or female, or fit or unfit. However, people with FH will almost always have a high blood cholesterol level. Because there are lots of other things that can cause high blood cholesterol levels, your doctor will also consider other risk factors to find out if you have FH.

Another indication that you might have FH is the presence of lumps on the tendons on the back of your hands and/or heels. Sometimes people with FH get lumps of cholesterol in these areas.

## What can be done about FH?

Families at high risk of early heart disease may not realise it could be because they have FH, or that it can be treated. FH treatment aims to lower blood cholesterol levels and reduce the risk of cardiovascular disease, such as heart attack and stroke.

Treatment can be started after the onset of heart disease, but it is better to find out if you have FH before it causes any damage. The key is to identify families who have FH, and then identify which family members have inherited FH.

It is important for all people who have FH, including children, to follow a healthy lifestyle. This includes eating a healthy, balanced diet low in saturated fat, being physically active and avoiding tobacco smoke. Smoking is especially dangerous if you have FH.

Adults who have FH will benefit from taking cholesterol-lowering medicine. They also need to see their doctor regularly to check that their heart stays healthy.

## Further information

### Australian Atherosclerosis Society FH Subcommittee

Website: [www.athero.org.au/FH](http://www.athero.org.au/FH)

### The Centre for Genetics Education

Current genetics information for individuals and family members affected by genetic conditions and the professionals who work with them.

Royal North Shore Hospital Community Health Centre, L5, 2c Herbert Street, St Leonards NSW 2065

Ph: (02) 9462 9599

Fax: (02) 9906 7529

Email: [contact@genetics.edu.au](mailto:contact@genetics.edu.au)

Website: [www.genetics.edu.au](http://www.genetics.edu.au)

For more information about healthy eating and how to reduce your risk of heart disease, call our Health Information Service on 1300 36 27 87 (local call cost) or email [health@heartfoundation.org.au](mailto:health@heartfoundation.org.au)

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