

Familial hypercholesterolaemia (FH)

Cardiovascular disease and inherited risk

Cardiovascular disease (heart, stroke and blood vessel diseases) is caused by a combination of inherited (genetic) factors and lifestyle habits, such as eating patterns, smoking and physical activity levels.

If you or other members of your family have suffered from a heart attack or stroke before the age of 60, it is very important to see your doctor to have your risk factors measured. The rest of your family (especially siblings and children) also needs to have their risk factors measured.

If cardiovascular disease occurs much later on in life, then it is unlikely your family members are at an increased risk of early onset of cardiovascular disease.

Special cases of inherited risk

In some families there is a much stronger inherited risk of cardiovascular disease. This may be due to problems with heart rhythm, heart muscle function or risk factors for heart disease such as high blood cholesterol. Treatment is available in nearly all of these cases.

This information sheet describes one of these special cases of inherited risk – inherited high blood cholesterol (also known as familial hypercholesterolaemia or FH for short).

Familial hypercholesterolaemia (FH)

What is FH?

FH is one of the most common, potentially fatal inherited conditions. FH is a condition that reduces the removal of cholesterol from the blood. This causes high blood cholesterol levels and early heart disease in some families. About one in every 500 Australians is affected. The good news is that FH can be treated very successfully.

Why do people get FH?

A pair of genes controls cholesterol removal from the blood. If one of these genes is faulty, then the removal process only works half as well. This means the blood cholesterol rises to about twice its usual level, even if people are following a healthy eating pattern and a healthy lifestyle. There is a 50 per cent chance that the faulty gene can be passed on to the next generation.

How do we know who is at risk of FH?

In families with FH, about half of the family members are affected by high blood cholesterol levels and the risk of early cardiovascular disease. The other half of the family members are unlikely to have high blood cholesterol levels and can be reassured, once tested, that they don't have FH.

Most people with FH are unaware they have it, as they don't feel any symptoms. In people with FH, high blood cholesterol levels are present from birth and can be diagnosed at any age.

FH does not depend on your body weight or whether you are male or female, or fit or unfit. However, people with FH will almost always have a high blood cholesterol level. As there are lots of other causes of high blood cholesterol levels, your doctor will also consider other factors to help identify whether the problem is FH.

If half of your family members are affected by FH, then it makes FH highly likely.

The other clue to FH is that adults with FH may get lumps of cholesterol in the tendons on the back of their hands or lumps on the tendons at the back of their heels (Achilles tendons, see below).



If anyone in your family has lumps like the ones shown in this photograph, that aren't explained by trauma, it makes FH highly likely. Discuss this with your doctor.

What can be done about FH?

Families at high risk of early heart disease may not realise that it could be due to FH and that it can be prevented by treatment. The treatment of FH can help lower blood cholesterol levels to a healthier level and prevent early death, heart attack or stroke.

Treatment can be started after the onset of heart disease, but it is better to identify FH before it causes any damage. The key is to identify families that are affected by FH, and then identify the family members who have inherited FH. All families with FH need to follow a healthy lifestyle.

Children with FH are encouraged to have a healthy eating pattern, to be physically active and to avoid tobacco smoke. Smoking is especially dangerous in this condition. Adults with FH need to follow the same advice, and in most cases they will benefit greatly from cholesterol lowering drug treatment. They also need to be monitored to make sure that their heart stays healthy.

Further information

If you think you may have or may be at risk of FH, discuss this with your doctor. Your doctor will be able to offer advice on the next steps to take.

For further advice about healthy eating and how to reduce your risk of heart disease, contact the Heart Foundation's national telephone information service, Heartline, on 1300 36 27 87, for the cost of a local call from anywhere within Australia. Alternatively, visit the Heart Foundation's website on www.heartfoundation.com.au.

Key Heart Foundation publications include:

- Enjoy healthy eating: a guide to keeping your blood cholesterol in check
- Cholesterol, triglycerides and heart disease
- Healthy eating for the heart

For further information about inherited high blood cholesterol or FH you might also like to contact the following organisations:

MED-PED (Make Early Diagnosis – Prevent Early Death) Australia
Little Stirling House
7 East Pallant St
North Adelaide SA 5006
Ph: (08) 8239 2977
Fax: (08) 8267 4412
Email: admin@medped-aust.com
Website: <http://www.medped-aust.com>
MEDPED international website: <http://www.medped.org>.

The Centre for Genetics Education provides current genetics information for individuals and family members affected by genetic conditions and the professionals who work with them. Access their website at www.genetics.com.au or call (02) 9926 7324.

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