Familial hypercholesterolaemia

Genetic testing for heritable high cholesterol
Familial hypercholesterolaemia (FH) is an inherited condition causing high levels of cholesterol in the blood.
A person with FH will develop high cholesterol at a young age. This could lead to blockages of the blood vessels (arteries) that supply the heart, brain and other organs, and could cause a heart attack, stroke or other blood circulation problems. This can be prevented by early diagnosis and treatment for FH.

FH is due to an inherited error in any one of several genes. If a person has FH, each of their immediate genetic relatives has a one-in-two chance of having FH.

**How common is FH?**

Approximately one in 16 Australians has raised cholesterol levels. For most people, the underlying cause is a combination of many different genes, diet and lifestyle (called polygenic hypercholesterolaemia). FH is different; there is only one genetic error that raises cholesterol levels. FH occurs in approximately one in 250 people in Australia. It is a significant cause of heart attacks in younger adults and cannot be controlled through diet and exercise alone.

**What is the purpose of genetic testing for FH?**

FH is diagnosed by finding an inherited error in one of the genes which can cause this disorder. Genetic testing provides a clear distinction between FH and polygenic hypercholesterolaemia, which is important for the following reasons:

- A person with FH requires closer monitoring and may require stronger medications or access to specific medicines provided by the Pharmaceutical Benefits Scheme.
- The relatives of a person with FH may benefit from a genetic test to determine whether they have FH.

Early diagnosis and early treatment are effective in lowering the cholesterol level, reducing the risk of having blocked arteries and reducing the risk of heart attack.
Testing can be performed at any age. If an FH genetic error has already been identified in the family, national guidelines recommend that at-risk children be tested by the age of 10 years.

**Before you have an FH test**

A genetic test for FH may be significant for both you and your family. Your result may be the only clue that they are at high risk of having FH, and of having a heart attack or stroke that could be prevented. A discussion with your medical specialist or a genetic counsellor, together with a signed consent form, are required prior to the test. Genetic counselling is available to you at no additional cost.

**How you will receive your test results**

Your doctor or genetic counsellor will tell you about the result of your test.

If a genetic error causing FH is found, your doctor will explain how this information will guide further investigations and therapy. It is also important that your close relatives are informed of your result and that genetic testing is available for them.

If a genetic error is not found, this means that you do not have a genetic diagnosis of FH. However, if you have a raised cholesterol level, you may still require further investigations and therapy.

For further information, please refer to our website, sonicgenetics.com.au/fhp
Arranging a test

1. Your doctor will have completed a request form for FH genetic testing.

2. Your blood sample can be taken at any of our pathology collection centres.

3. If requested by your doctor, we will arrange for a genetic counsellor* to contact you to discuss testing.

4. Your result is reported back to your doctor, usually within 4–8 weeks of the laboratory receiving your blood sample.

5. Post-test counselling by a genetic counsellor is available to you, and can be arranged by your requesting doctor.

* Terms and conditions apply. Please refer to sonicgenetics.com.au/patient/genetic-counselling

Cost

Medicare-rebated testing is available when requested by a medical specialist for a patient with:

- LDL-cholesterol level of ≥6.5 mmol/L, or
- LDL-cholesterol level of ≥5.0 mmol/L plus clinical features of accelerated impairment of blood circulation, or
- a combination of clinical and family features (Dutch Lipid Clinic Network score of at least 6), or
- a first- or second-degree relative with a documented FH genetic error.*

Your doctor can also request FH testing for a private fee if Medicare criteria are not met.

*Medicare rebate also available when requested by a GP.
Sonic Genetics is Sonic Healthcare’s centre of excellence in genetic pathology testing. As the largest pathology provider in the country, Sonic Healthcare’s state-of-the-art laboratories and extensive network of collection centres serve each state and territory capital, as well as regional and rural Australia.

Douglass Hanly Moir Pathology
Sullivan Nicolaides Pathology
Melbourne Pathology
Barratt & Smith Pathology
Capital Pathology
Clinipath Pathology
Bunbury Pathology
Clinpath Pathology
Hobart Pathology
Launceston Pathology
North West Pathology
Southern.IML Pathology

For further information, please refer to our website, sonicgenetics.com.au or call us on 1800 010 447