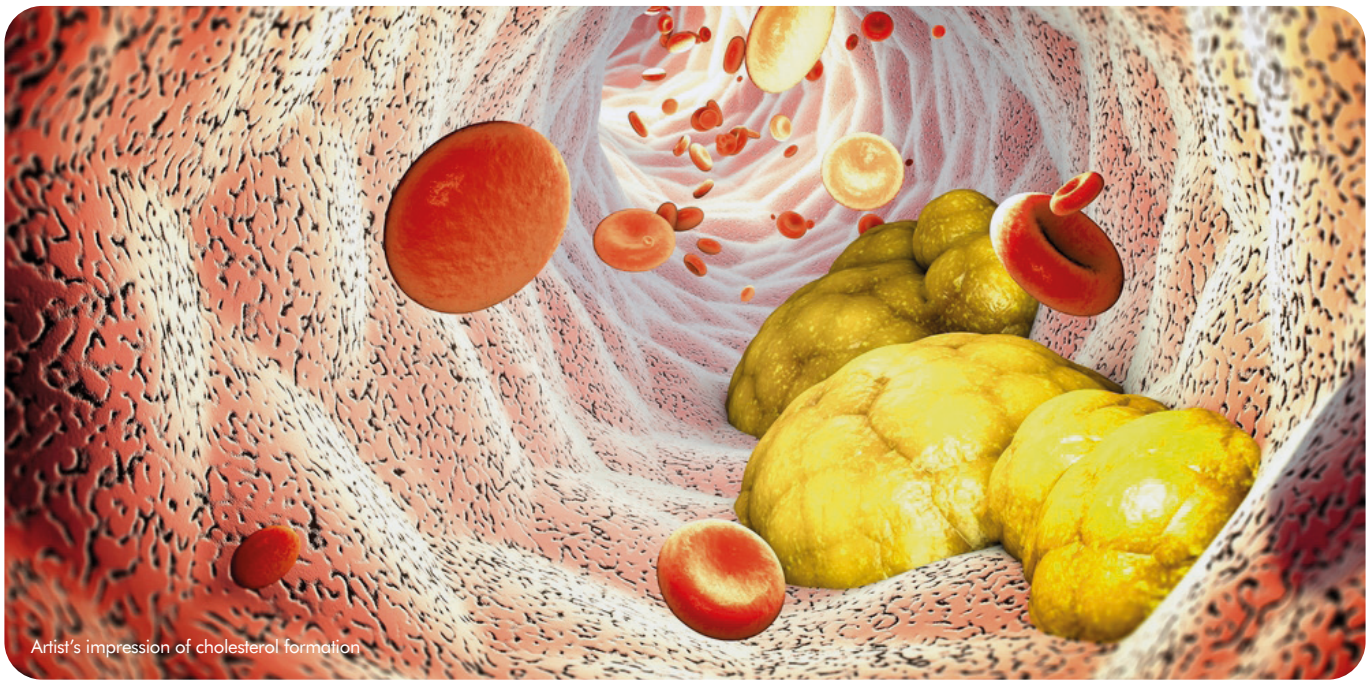
PATHOLOGY TESTS EXPLAINED

Information about pathology tests to help everyone take control of their health and make the right decisions about their care.

WHAT YOU SHOULD KNOW ABOUT **TESTING FOR FAMILIAL HYPERCHOLESTERAEMIA**

Familial hypercholesteraemia or FH is an inherited condition that is caused by mutations in the genes that help control how your body removes cholesterol from your blood. If you have FH you have higher-than-normal LDL cholesterol from birth which increases your risk of having heart disease or a heart attack at an earlier age. Diagnosing FH and treating it with cholesterol-lowering medication can reduce your risk of heart disease.



Artist's impression of cholesterol formation

LDL

LDL cholesterol

Cholesterol is carried around the bloodstream in lipoproteins. There are several types of lipoproteins, but the main ones are High-Density Lipoprotein (HDL) sometimes called 'good' cholesterol and Low-Density Lipoprotein (LDL) or 'bad' cholesterol. All cholesterol is the same – it is the lipoproteins that carry it that are different. High levels of LDL deposit cholesterol into blood vessel walls forming plaques. By lowering LDL it's possible to reduce the cholesterol inside these plaques.

What causes FH?

FH is caused by a mutation (also called a variant) in the LDL receptor gene (*LDLR*) or occasionally in other genes *(APOB or PCSK9)* that reduce the clearance of LDL particles from the blood.

The body's cells, particularly the liver cells, take up LDL cholesterol from blood through receptors. Normally, a person has two working copies of the gene controlling the number of receptors produced. One copy of your gene is inherited from your mother and one from your father, but in FH one copy does not work properly, so only half of the normal numbers of receptors are produced.

Your doctors may investigate further for FH if your cholesterol (lipids) test results show you have a total cholesterol greater than 7.5 mmol/L or LDL cholesterol greater than 5.0 mmol/L, especially if there is a history of premature coronary heart disease in your family.

Genetic testing is used to diagnose FH

There are two types of testing:

- A test looking at your DNA to search for any variant in the FH-causing genes.
- A test looking at your DNA for only a specific variant that has previously been identified in your family.

FH genetic testing can be requested by a specialist doctor if they suspect your high LDL cholesterol is due to FH.

A calculated score called the Dutch Lipid Clinic Network Score, based on your family history, untreated LDL-cholesterol levels and physical signs, is used to determine whether FH is likely. If you are diagnosed with FH you will be treated with a cholesterol lowering drug such as a statin and / or other drugs and given advice about your diet and lifestyle, especially smoking.

If you have FH confirmed by genetic testing and you are the first person in your family to be diagnoses, you are termed an 'index case'. Each of your close family members – parents, siblings, and children – has a 50 per cent chance of also having the gene variant causing FH and they should be tested. The diagnosis of FH in children should ideally occur before they are 10.

Medicare

FH genetic testing is covered by Medicare when it is ordered by a specialist doctor for testing a suspected index case. It is also covered when ordered by a GP or specialist doctor to test first or second-degree relatives of someone who has been diagnosed with FH.

What can your results tell you?

Possible outcomes of genetic testing include:

A disease-causing (pathogenic), or likely disease-causing (likely pathogenic) gene variant (mutation) is identified.	This confirms a diagnosis of FH.
A disease-causing variant (mutation) was not identified. This may be because your high cholesterol is: • not due to a genetic change, • is due to a genetic change(s) in a gene(s) that the technology we have at present cannot detect.	This result does not mean that you do not have FH, just that it cannot be confirmed at this time.
A gene variant (mutation) of uncertain significance is identified.	This means that based on current knowledge of the gene involved, the laboratory is unable to determine whether the genetic change is the cause of your high cholesterol.

Questions to ask your doctor

Why does this test need to be done?
Do I need to prepare (such as fast or avoid medications) for the sample collection?
Will an abnormal result mean I need further tests?
How could it change the course of my care?
What will happen next, after the test?

For more detailed information on these and many other tests go to **pathologytestsexplained.org.au**

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www.pathologytestsexplained.org.au

Please use this QR code to access more information

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Pathology Tests Explained is managed by a consortium of medical and scientific organisations representing pathology practice in Australia. More details at: **www.pathologytestsexplained.org.au/about**

My Health Record

You'll find a direct link to the Pathology Tests Explained website embedded in the pathology results pages of your My Health Record.

Click on the link to find information about what your tests are investigating or measuring and what your results can tell your doctor.