The National fH Registry

NEWSLETTER FOR THE NATIONAL FH REGISTRY

December 2021

FH Genetic Testing is Now on Medicare

Who is eligible and how can it help my family?

On 1 May 2020, genetic testing for familial hypercholesterolaemia (FH) was included in the Australian Medicare Benefits Schedule (MBS). This means that much of the cost of this complex laboratory analysis will be paid by the Australian Healthcare system, where appropriate. With more accessible genetic

testing, we can expect improvements in the detection and management of individuals and families affected with FH.

Let's unpack some of the important information on how FH genetic testing could affect you and your family members.

Why was genetic testing for FH added to the MBS?

As you may already know, FH is an inherited condition that reduces the body's ability to clear low-density lipoprotein (LDL)-cholesterol from the blood. The build-up of LDL-cholesterol in the blood vessels over time creates blockages in the vessels which increases the risk of heart disease. Left untreated, 85% of people with FH will develop atherosclerosis by the age of 60.

FH is a common genetic condition affecting approximately 1 in 300 people in Australia, with most individuals being undiagnosed or inadequately treated. There are likely 100,000 people unknowingly living with FH in Australia, one in five of them are children. For this reason, it is a significant and often 'silent' public health issue. Research shows that genetic screening for FH is a cost-effective way to prevent cardiovascular disease in families with FH, and because FH runs in families, finding one person with FH can help find others too. FH is one of the best examples of the way in which a genetic approach to a health problem, like cardiovascular disease, can improve treatment.

Let's quickly summarise the inheritance of FH. First-degree relatives of a person with FH have a 1-in-2 (50%) chance of also having the genetic variant which causes elevated LDL cholesterol. This is called an autosomal dominant inheritance pattern.

Once a patient is diagnosed, FH is very treatable. Routine cholesterol lowering treatments and new biological treatments are very effective. Finding individuals with FH early is very important to reduce the chances of developing cardiovascular disease and other related health issues.

Rapid advances in genetic technology have enabled quicker, cheaper, and more comprehensive testing for FH than ever before. A concept called "Moore's Law" describes how quickly the cost and capability of genetic technology can change. In 1996, it cost $2.7 billion to sequence the whole genome, and in 2021, it cost only $3000 to run the same testing.

For all these reasons, funding for FH genetic testing is now available for individuals who qualify.

Who can access genetic testing?

There are two main groups of people eligible for FH genetic testing:

1. Diagnostic genetic testing

Diagnostic genetic testing for FH is used to identify the first person suspected to have FH in the family. These people usually have clinical features of the condition such as: high cholesterol for their age, premature coronary artery disease, and maybe some of the rarer physical signs. These individuals will be assessed using the Dutch Lipid Clinic Network Score and have a blood test ordered by a specialist. A genetic counsellor may be involved before you have this test to outline the benefits and limitations and discuss the result with you. The test itself requires extensive analysis of the sequence of at least 3 genes that affect cholesterol. If a variant is found in these genes, it needs to be evaluated to decide whether or not it may have caused the cholesterol problem.

2. Cascade testing for FH

If a gene variant causing FH is found in the first family member, we can look for this variant in other family members. We start by looking at first-degree relatives; such as, parents, brothers/sisters, and/or children and then, can 'cascade' to the wider family. A GP can order this blood test, but the result of the initial diagnostic test needs to be provided. The test itself is much simpler because it just requires confirmation of the change which was present in the family relative. Often a genetic counsellor will facilitate cascade genetic testing in the family.

Future directions

With time, the diagnosis and management of FH will become more accessible and effective for individuals and families. For example, saliva tests rather than blood tests are expected to become more common in the future, especially for children. Researchers around the world are investigating practical ways to implement screening programs for FH, like those we have for breast cancer and bowel cancer. Perhaps we will see universal screening for FH in the future. It is important that we seek the advice of FH patients as we aim to "imagine" the future. Your participation as a consumer health advocate to complete occasional surveys would be most welcome.

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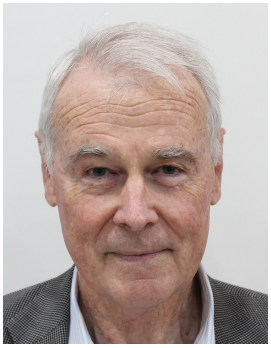

Opportunity to complete survey about life insurance and genetic testing

Many people are concerned about the use of genetic information by life insurers. Researchers from Monash University are keen to understand the views and experiences of consumers. This includes people who have had genetic testing as well as people who are eligible, but choose not to have genetic testing. The findings of this research will contribute to a report to the Australian government regarding the current regulation of life insurers, and your participation will assist with gathering critical data on the views and experience of consumers.

Please consider **completing this important survey**, whatever experience you have had with life insurance and genetic testing. You can remain anonymous if you wish.

Please also consider **forwarding this survey link** to your family members who may have had or considered having a test.

You can access the survey here: https://redcap.link/aglimmer.consumer**.** At the beginning you will find more information about the study and an explanatory statement.

For any queries regarding this research, please contact the research coordinator, Jane Tiller, on jane.tiller@monash.edu.

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everyone deserves
a better chance

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