HOW IS GENETIC TESTING DONE?

Genetic testing involves taking a sample of DNA from blood, saliva or tissue and analysing the genes that are most commonly known to cause FH.

When testing a family, we begin by testing a person known to have FH. If a mutation is identified in that individual, then testing can be performed on other family members to look for that specific mutation or mutations.

Genetic testing is complex; it takes place with genetic counselling and the process of informed consent.

If your family history suggests FH, please talk to your doctor. A referral to Curie Genetics can help you find out more about FH and genetic testing.

GENETIC COUNSELLING

The diagnosis of FH has implications for other family members. A trained genetic counsellor will review your family history and discuss implications of the diagnosis with you and your family.

In addition, the genetic counsellor can serve as a resource to other family members who may have questions about the diagnosis. It is important to us that you understand all aspects of your condition and we are always available to answer questions.

REFERRAL INFORMATION

Address 📍: CURIE Genetics
(Mount Elizabeth Novena Hospital #10-37)
Singapore 329563

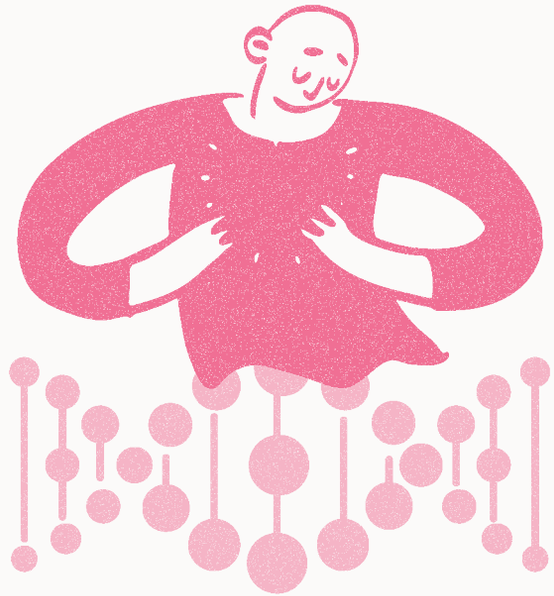
Telephone 📞: 6592 8615

Whatsapp : 8683 3767

Website : www.curiegenetics.sg

Adapted content from Genzyme Cardiovascular

FAMILIAL HYPERCHOLESTEROLEMIA (FH)



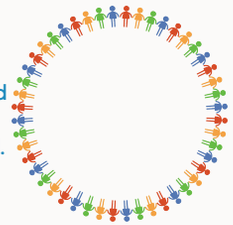
CURIE GENETICS

BY CURIE ONCOLOGY

WHAT IS FAMILIAL HYPERCHOLESTEROLEMIA?

Familial hypercholesterolaemia (FH) is an inherited condition, where an altered gene causes high blood cholesterol. It is usually referred to as FH.

FH is one of the most commonly inherited conditions. As many as 1 in 250 people may have an altered gene that causes FH.

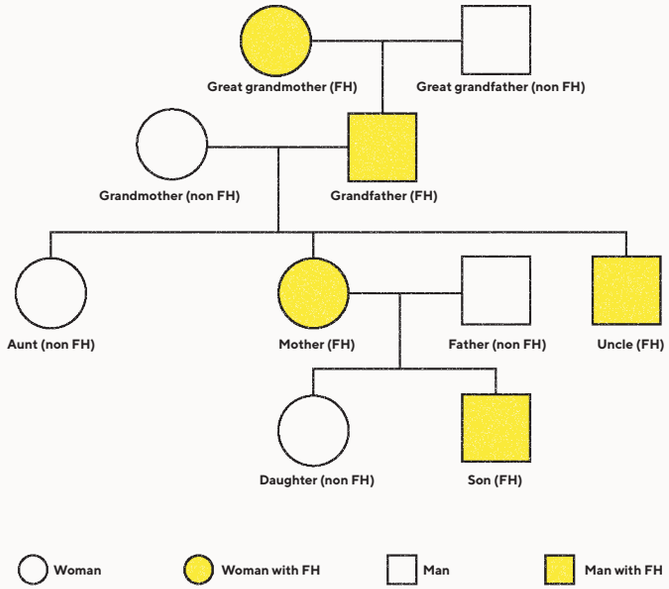


Familial means that it runs in families and is passed from a parent to their child.

Hypercholesterolaemia means high blood cholesterol. The type of cholesterol that is specifically increased in FH is Low Density Lipoprotein-cholesterol (LDL-cholesterol).

FH is linked to a higher risk of very early cardiovascular disease. Cardiovascular disease means problems of the heart or blood vessels caused by a build-up of fatty deposits inside the arteries and a higher risk of blood clots.

DOES MY DOCTOR THINK I HAVE FH?



Your doctor may think you have FH if:

- there is a history of early cardiovascular disease in close family members
- you are a male and have had a heart attack before the age of 50, or female and have had a heart attack before the age of 60
- your LDL-cholesterol is very high (≥160 mg/dL for children or 190 mg/dL for adults)
- other family members have been diagnosed with FH

you have any physical signs of high cholesterol such as:

- swollen tendons on the heels and knuckles of the hands (tendon xanthoma)
- yellowish patches around the eyes (xanthelasma).
- A white deposit of cholesterol in the shape of an arc (corneal arcus) at the edge of the coloured part of the eye.

HOW FH CAN BE DIAGNOSED?

Doctors diagnose FH by looking for unusually high LDL-cholesterol, finding fatty bumps under the skin and by checking for early heart disease in you or your close family.

Together these can suggest definite or possible FH. FH can also be confirmed by a genetic test. The test looks for a "disease causing" alteration in the LDL receptor, APOB, PCSK9 or APOE genes.

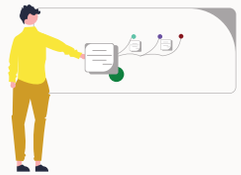
If you are diagnosed with FH, there is a 50% chance that your family members (parent, brothers, sisters, children) may have FH and genetic testing is recommended.

This form of testing is known as cascade predictive testing - a way to find FH before it leads to cardiovascular issues.

BENEFITS OF GENETIC TESTING

- Confirmation of a diagnosis

- Provide insights on underlying cause of heart disease



- To begin with risk reduction interventions by making informed medical decisions

- Identify at-risk family members for cascade screening