

## What is Familial Hypercholesterolaemia?

**Familial** means 'runs in families' or 'inherited'.  
**Hypercholesterolaemia** means 'high blood cholesterol'.

Familial Hypercholesterolaemia (shortened to FH) is an inherited condition causing high cholesterol. High cholesterol causes heart disease. High cholesterol starts at birth so people with **untreated** FH tend to get heart disease 20 or 30 years earlier than someone without FH.

## How common is FH?

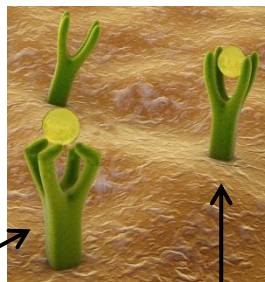
In Australia it is believed that FH affects 1 in every 300 people. In families with FH, around half of the family members will have FH.

## How does FH cause high cholesterol?

FH is caused by a mutation (error) in the gene that makes the receptor which removes cholesterol from the blood.

We all have two copies of this gene, one from each parent. In the most common form of FH one copy of the gene has an error and half the receptors are made incorrectly and cannot remove cholesterol from the blood.

As a result, the blood cholesterol rises to about double the normal level, even when a person has a healthy diet.

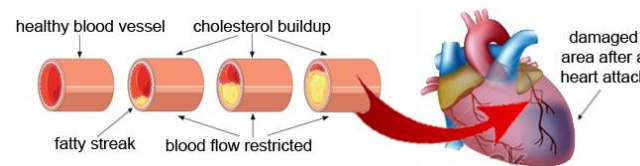


**Faulty receptor**  
(cannot 'hold' the cholesterol  
so cannot remove it)

**Normal receptor**

## How does high cholesterol cause heart disease?

If you have high cholesterol, overtime it tends to deposit in the walls of the blood vessels of the heart causing heart disease. This narrows the blood vessels and reduces blood flow. Reduced blood flow leads to heart attacks. Left untreated, 85% of people with FH will develop heart disease by the age of 60.



Other parts of the body can also be affected resulting in stroke and other circulation problems.

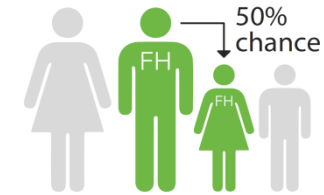
## How is FH treated?

A healthy lifestyle is important for overall heart health and will also reduce cholesterol levels but usually not enough, so **lifelong cholesterol lowering medication** is generally required. Statins are the medication most commonly prescribed. They work by reducing the amount of cholesterol your body makes. They have been shown to be safe and effective.

## When should medication start?

Adults diagnosed with FH should start cholesterol lowering medication as soon as possible to prevent, slow or reverse heart disease. Children should be tested around the age of 10 and medication started at the discretion of the parents and doctor.

## What about my family?



FH runs in families. If you have FH each of you siblings and children has a 50% (or 1 in 2) chance of also having FH.

**It is important that cholesterol testing is done on all your immediate family members to detect others who have FH.**

## How is FH diagnosed?

To make a diagnosis of FH the following will be considered:

- your cholesterol level
- history of early heart disease in you and other family members
- physical signs - people with FH may have 'lumps' of cholesterol on their hands, legs or eyes
- genetic test to look for the faulty gene causing FH. A genetic test is not required for a diagnosis of FH but is useful in borderline cases. Genetic testing is only offered at some major hospitals.

## Is FH the only cause of high cholesterol?

No, there are many causes of high cholesterol; poor lifestyle (high fat diet and not enough physical activity) and increasing age are the most common. There are also other conditions that will cause high cholesterol but these will be ruled out before a diagnosis of FH is made.

## If you have FH: What you can do

- Take your medication **daily** to reduce your cholesterol level and risk of having a heart attack. Treatment is lifelong.
- Have regular check-ups, blood tests and additional tests for checking your heart health as recommended by your doctor.
- Ask your nurse or doctor to help you make a plan for:
  - If you smoke cigarettes, help in quitting. **Smoking speeds up the rate that cholesterol deposits in your blood vessels. Stopping smoking is most important.** Also, avoid second-hand smoke.
  - Consume a healthy diet low in saturated fat, trans fat, salt and sugar
  - Maintain or reach a healthy weight
  - Stay or become physically active
  - Do not drink alcohol or drink in moderation.

See <http://www.athero.org.au/fh> for more information including Australian Dietary, Physical Activity and Alcohol Guidelines.

- Tell your family members that you have FH and encourage them to have their cholesterol tested.
- Join the *National FH Registry*. The registry is a database of patient information which will be used to further the understanding of FH and ultimately improve patient care. By joining the *National FH Registry* you will have access to the latest information and trials for new FH treatments.

Early Diagnosis  
Early Treatment  
Saves Hearts



### The FH Australasia Network

- Email: [fhwa@health.wa.gov.au](mailto:fhwa@health.wa.gov.au)
- Website: <http://www.athero.org.au/fh>
- Facebook: <https://www.facebook.com/FHAustralasiaNetworkandSupportGroup>
- Twitter: <https://twitter.com/FHAustralasia>

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