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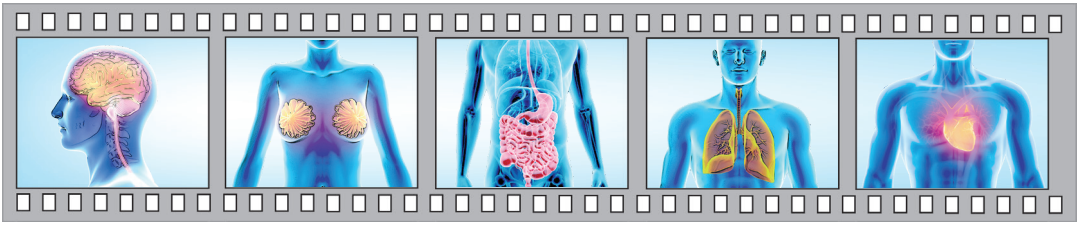
Familial Hypercholesterolaemia FAQ



What is Familial Hypercholesterolaemia?

Familial Hypercholesterolaemia (FH) is a genetic condition causing high cholesterol levels resulting in heart attacks at an early age. It is one of the most common genetic conditions, affecting about one in 250 people in most populations, and it is the most important genetic cause of high cholesterol levels and early heart disease.

People with **Familial Hypercholesterolaemia** typically have very high blood cholesterol levels. Usually the total cholesterol is about 7.0 mmol/L or higher and the harmful cholesterol, the low-density lipoprotein or LDL-cholesterol, is about 5.0 mmol/L or higher. In the units used in North America and some other countries, these figures for total cholesterol and LDL-cholesterol are about 270 and 190 mg/dL, respectively. Young children with **FH** may have lower levels of total cholesterol and LDL-cholesterol.

What causes FH?

Familial Hypercholesterolaemia is caused by a change in one of the genes involved in controlling cholesterol levels and is inherited in such a way that 50% of close family members will have the condition[1]. Very rarely, some people can inherit a gene for **FH** from both parents, and their cholesterol levels are extremely high. This condition is called homozygous **FH** and may cause heart attacks in childhood or adolescence.

How is FH diagnosed?

Familial Hypercholesterolaemia can be diagnosed from the combination of the person's cholesterol levels and a family history of high cholesterol or early onset heart disease with heart attacks. The diagnosis can be confirmed by genetic tests.

What problems does FH cause?

In **Familial Hypercholesterolaemia**, the cholesterol deposits in the arteries, particularly the coronary arteries in the heart and such can cause heart attacks at a very early age. If the cholesterol level is very high, it can also deposit in the tendons and lumps or thickening of the tendons can be seen or felt, usually in the Achilles tendons just above the heel, or the tendons on the knuckles at the base of the fingers[2]. Some people with the condition also develop a white ring or crescent around the cornea of the eyes or yellowish deposits of cholesterol in the eyelids.

The risk of developing heart disease in people with **FH** is thought to be increased by about 20-fold compared with the general population. Because people with **FH** have been exposed to high

cholesterol levels since birth, their risk of heart disease is much higher than those people who develop high cholesterol later in life because of a bad diet. Other risk factors for coronary artery disease such as smoking, diabetes, hypertension and obesity will increase the risk of heart attacks dramatically in people with **FH**.

How is it treated?

Most people with **FH** will need to take medication with statins. They should also follow a low-fat diet, which will help to reduce the cholesterol, but usually, diet alone is not enough[3].

It is important to get the LDL-cholesterol as low as possible to prevent the narrowing of the arteries from progressing.

Statins occasionally cause problems, some people have a condition called statin intolerance, and patients with these problems require alternative treatments.

Some people with **FH** who already have coronary artery disease should also take aspirin. For those people with **FH** who are not known to have coronary artery disease taking aspirin is more controversial.

Should I have my cholesterol checked?

Everyone should have their cholesterol level checked!

If it is very high, and if high cholesterol appears to run in the family, then they may have **FH**. It is important to identify this, firstly because it needs to be treated very effectively, and secondly, because other family members can be screened and identified and treated before they have their first heart attack, which can sometimes be fatal!

References

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