Understanding Familial Hypercholesterolaemia (FH)

What is FH?

Familial Hypercholesterolaemia, or FH, is an inherited condition where an altered gene causes exceptionally high cholesterol levels in the blood, often double and sometimes four times those of the general population.[1] Familial means that it runs in families and is passed from a parent to their child.[2] FH can usually be traced over several generations. Hypercholesterolaemia means high blood cholesterol. The type of cholesterol that is specifically increased in FH is low-density lipoprotein cholesterol (LDL-C), sometimes called 'bad' cholesterol. In FH very high levels of LDL-C can block or narrow blood vessels and increase the risk of heart disease at an early age.[2]

Many people worldwide have high cholesterol (common hypercholesterolaemia) in middle age, but people with FH have high cholesterol from birth.[3]

FH in the UK

FH is one of the most common genetic conditions that occurs in at least 1 in 500 people (120,000) in the UK.[1] However, 85% of people with FH remain undiagnosed and untreated, putting them at increased risk of early heart disease and death.[3,4]

What are the risks of FH?

FH is linked to an increased risk of early heart disease.[2] The risk varies from family to family with FH and is influenced by cholesterol levels, other inherited risk factors, lifestyle and whether you are male or female.[2]

If left untreated, approximately 50% of men and 30% of women with FH will have developed heart disease by the time they are 50 and 60 years old respectively due a life-long exposure to high cholesterol.[3]

What causes FH?

FH is not caused by an unhealthy lifestyle, but is passed from generation to generation through a faulty or altered gene.[1] Most people with FH have inherited a defective gene from one parent and, therefore, will have had high levels of cholesterol from birth.[2]

Each child has a 50% risk of inheriting FH if one of their parents has a faulty gene.[3]

How is FH diagnosed?

FH is usually an "invisible" disease and, therefore, people do not show visible signs. There are two main indicators of an FH diagnosis:[2]

1. Abnormally high levels of cholesterol
2. Family history of high cholesterol or early heart disease (including heart attacks) and stroke

Sometimes there are visible signs of FH, especially when the LDL-cholesterol is very high. These include:[5]

1. Swollen tendons on the back of the ankle and hand (tendon xanthoma)
2. Yellow deposits in the skin around the eyes (xanthelasmata)
3. A white deposit of cholesterol in the shape of an arc around the coloured part of the eye (corneal arcus)

An FH diagnosis may be fully confirmed by a genetic test[3]. The diagnosis can also be made on clinical grounds. Once an FH diagnosis is confirmed, it is important that all family members including siblings, children, parents, grand-parents and first cousins are tested for FH.[3]

Treating FH

FH is not curable but it is treatable.[2] The risk of cardiovascular disease in FH can be reduced, before fatty deposits build up and begin to block blood vessels, by[2]:

1. Changing aspects of lifestyle, e.g. stopping smoking, limiting food and drinks high in sugar or alcohol
2. Diet modification to include healthy foods
3. Taking cholesterol-lowering medications as prescribed by a doctor

THIS DOCUMENT HAS BEEN DEVELOPED BY SANOFI

Date of preparation: August 2015
Job number: SAGB.CMR.15.04.0459f

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