NKT for FH

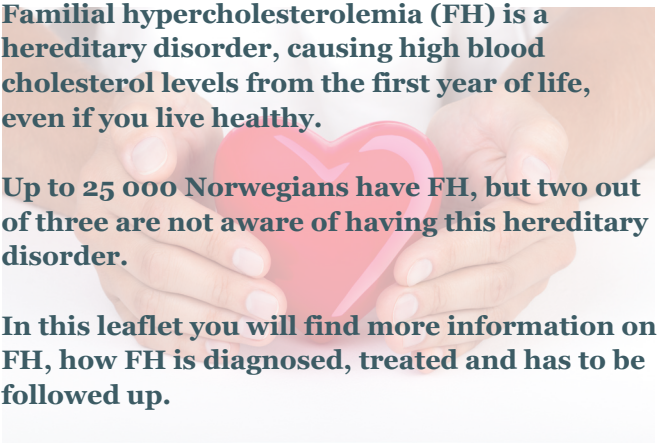
National advisory unit on familial hypercholesterolemia

Familial hypercholesterolemia (FH)

Norwegian National Advisory Unit on Familial Hypercholesterolemia (NKT for FH)

Oslo University Hospital

FAMILIAL HYPERCHOLESTEROLEMIA



Familial hypercholesterolemia (FH) is a hereditary disorder, causing high blood cholesterol levels from the first year of life, even if you live healthy.

Up to 25 000 Norwegians have FH, but two out of three are not aware of having this hereditary disorder.

In this leaflet you will find more information on FH, how FH is diagnosed, treated and has to be followed up.

What is FH?

FH causes high blood cholesterol levels
Cholesterol is an important building block in the body, and is transported through the blood stream. Excess cholesterol (LDL-cholesterol) is normally removed from the blood by the liver.

Individuals with FH have an inherited defective uptake mechanism, reducing the liver uptake of LDL-cholesterol from the blood. This results in increased levels of LDL-cholesterol in the blood; hypercholesterolemia.

FH is a hereditary disorder
The defective uptake mechanism in the liver is caused by a change (mutation) in a specific gene.

The mutation is present from conception and runs in families. It is sufficient to inherit one mutated gene from one of your parents to get FH.

If one parent has FH, each child has a 50 % chance of inheriting FH, independent of gender.

Untreated FH increases cardiovascular disease risk
High blood cholesterol levels can lead to narrowing or blockage of blood vessels (atherosclerosis).

If untreated, the high cholesterol levels in people with FH can result in premature cardiovascular disease (heart disease or stroke), occurring 15-20 years earlier than among people without FH.

Early diagnosis and treatment can lead to a normal life span. Accurate treatment is therefore important.

Right treatment reduces risk substantially
In order to compensate for the life long exposure to high cholesterol levels, cholesterol treatment goal values are extra low in FH. When treatment is initiated after the age of 40, the goal is set even lower.

Cholesterol lowering treatment should thus be initiated early, ideally from the age of 8-10 years (depending on cholesterol values and family history). Even before this age, a healthy diet and lifestyle is advised.

FH is a hereditary disorder which if untreated leads to high cholesterol values from the first year of life. If one of the parents has FH, each child has a 50 % chance of inheriting FH - independent of gender.

How is FH diagnosed?

Individuals with FH may have only moderately increased cholesterol levels, but over a lifetime, these increased levels will affect the blood vessels considerably.

If family members have high cholesterol levels or have had cardiovascular disease at a young age, you are advised to measure your blood cholesterol and if applicable have genetic testing performed.

Consider a genetic test if untreated total cholesterol is

- Above 6 mmol/L in persons under 20 years
- Above 7 mmol/L between 20 and 40 years
- Above 8 mmol/L in persons over 40 years

When FH is diagnosed with a genetic test, the diagnosis is definite. This is an important basis for identifying other family members with FH.

The genetic test is performed in an ordinary blood sample which can be taken by your general practitioner. A test form can be downloaded from our webpage (see last page).

The genetic test result is confidential, and not distributed to employer, insurance company, or others.

Treatment

Treatment goals

Under 18 years	: LDL-cholesterol below 3.5 mmol/L
Over 18 years	: LDL-cholesterol below 2.5 mmol/L
With risk factors*	: LDL-cholesterol below 1.8 mmol/L

**Cardiovascular disease, diabetes , or treatment start after the age of 40.*

Medication

Adults with FH are always treated with drugs in addition to advice on healthy diet and lifestyle. There are several types of drugs which alone or in combination reduce the blood cholesterol.

Statins inhibit cholesterol production and are first line treatment in FH.
Ezetimibe is a second line therapy which acts by decreasing cholesterol absorption in the intestine.
PCSK9-inhibitors are new second line cholesterol reducing drugs, used if statins and ezetimibe are considered to be insufficient.

See our web page for more information on medication.

Healthy diet and lifestyle advice

- Reduce intake of saturated fats from animal sources and foods rich in cholesterol, and replace with unsaturated vegetable fat.
- Eat plenty of foods rich in fiber and at least five daily fruit and vegetables portions.
- Maintain normal body weight (Body Mass Index between 18 and 25 kg/m^2).
- Be physically active for at least 150 minutes weekly.
- Quit smoking

Pick up our book on diet advice for persons with dyslipidemias at a lipid clinic or on our website.

Follow-up

Genetic counselling

If FH is confirmed by the genetic test, genetic counselling for advice on FH and inheritance is recommended. Call Enhet for hjertegenetikk on telephone 22 11 89 62 or 22 11 89 75 to make an appointment. You can also be referred by your general practitioner.

NB! Due to legal and privacy considerations, health personell will not inform relatives of their conditions. This will be discussed with you during gentic counselling.

Lipid clinics

Individuals with FH should haveregular consultations at a lipid clinic with expertise on FH. Referral addresses can be found on our web page.

General practitioner

In between consultations at a lipid clinic, blood values and treatment should be followed up by your general practitioner. Guidelines can be found on our website.

Patient organisation

"FH Norge" fights for your patient rights. Members also recieve a biannual patient magazine with news about FH. Sign up on www.f-h.no.

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The Norwegian National Advisory Unit on Familial Hypercholesterolemia (NKT for FH) aims to increase knowledge, awareness and research on FH and other hereditary lipid disorders.

NKT for FH is a national unit, located at
Oslo University Hospital, Norway.