FAMILIAL HYPERCHOLESTEROLEMIA
An educational booklet for patients with familial hypercholesterolemia
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PART 1
FAMILIAL HYPERCHOLESTEROLEMIA

What is Familial Hypercholesterolemia?

Familial Hypercholesterolemia (FH) is an inherited disease, where a genetic alteration causing high blood cholesterol is transmitted from generation to generation (see figure 1). Familial means it runs in families; sometimes it is possible to trace the disease over several generations. Hypercholesterolemia means high blood cholesterol. The type of cholesterol that is specifically increased in Familial Hypercholesterolemia is Low Density Lipoprotein-Cholesterol (LDL-C). LDL-Cholesterol floats in the blood stream and transports cholesterol from one cell in your body to another cell (see parts 1.2 and 3.4).
FH is one of the most common hereditary disorders. Approximately 1 in 500 people in the world has a genetic alteration that causes FH\(^1\). If one parent has FH, there is a 50% chance that their son or daughter will also have it.

FH is associated with an increased risk of cardiovascular disease\(^1\). Risk varies from family to family and is influenced by cholesterol level, other hereditary factors, lifestyle factors such as diet, smoking and level of physical activity, and whether you are male or female. Women with FH are affected by cardiovascular disease approximately 10 years later than men with FH\(^2\). With early and appropriate treatment the risk of cardiovascular disease can be reduced significantly.

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**IMPORTANT**

FH is inherited and runs in families. It causes high blood cholesterol, specifically LDL-Cholesterol, and an increased risk of cardiovascular disease early in life.

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**2** What is LDL-Cholesterol?

Low Density Lipoprotein-Cholesterol (LDL-C) is a particle that floats in the blood stream and acts as an efficient transport system that carries cholesterol from one cell to another in your body. Cholesterol is a fatty substance that is needed by your body to build cells, to make hormones, and to make bile acids in the liver (see part 3.5).

Too much LDL-Cholesterol in your blood stream is not good for you; excess cholesterol can be deposited in the walls of blood vessels making them narrower and causing the onset of atherosclerosis. Atherosclerosis can lead to cardiovascular diseases (see part 3).

To transport cholesterol into specific cells, the LDL-C particle has a specific protein attached to it named Apo-lipoprotein B or ApoB. ApoB acts as a bridge between the LDL-C particle and the cells in your body that carry the LDL-receptor (which helps the cell recognize LDL-C). If the LDL-receptor or ApoB protein is not normal, the level of cholesterol in your blood will increase. This is the case in Familial Hypercholesterolemia.

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within a specific gene can result in a hereditary illness. In FH there is a change in the gene that codes for the LDL-receptor. This receptor sits on the surface of cells and can be thought of as a ‘tentacle’ that removes cholesterol-containing LDL-C particles from the blood (figure 3). The change in the LDL-receptor gene results in abnormal LDL-receptors (‘tentacles’) making them unable to remove LDL-Cholesterol from the blood. This is the cause of FH.

3 What are the causes of FH?

**FIGURE 2:**
DNA, which is over one meter in length, is packed in chromosomes. Chromosomes are packed into the cell nucleus, which is inside the cell.

The characteristics we inherit from our parents are determined by information carried on a strand of DNA that totals well over one meter in length. DNA is organized into chromosomes found in the nuclei of cells (figure 2). DNA consists of approximately 3 billion building blocks, of which 25,000 specific combinations make genes. Genes are a code for physical characteristics, such as eye and hair color, but also many diseases. A change in just one of these building blocks within a specific gene can result in a hereditary illness. LDL-C transports cholesterol in your blood to cells. Cholesterol is needed to build cells, make hormones and make bile acids. However, if there is too much LDL-Cholesterol it will be deposited in blood vessel walls. This phenomenon is called atherosclerosis.
Most people with FH have inherited one defective LDL-receptor gene from one of their parents, and one normal LDL-receptor gene from the other parent. Consequently, they have only approximately 50% of the normal number of working LDL-receptors (‘tentacles’) on the surface of cells (figure 4). This means that there is always an unnecessarily high amount of LDL-Cholesterol circulating in the blood. The problem is that excess LDL-Cholesterol can be deposited in the blood vessel wall.

**FIGURE 3:**
The LDL-receptor is attached at one end to the cell membrane. The other end binds to LDL-Cholesterol particles.
Normal cell

Abnormal cell

LDL

LDL-receptor

Blood stream

Cell

LDL particle

LDL-receptor

FH is caused by a change in the gene that codes for the LDL-receptor. The defective LDL-receptor cannot take up LDL-Cholesterol from the blood into the cell. Excess cholesterol is deposited in blood vessel walls.

IMPORTANT

FH is suspected if there is a family history of cardiovascular disease early in life. If a person suffers a heart attack before the age of 50-60, it may be due to high cholesterol and blood lipid profiles must be investigated in the family:

- Lipid profile means the assessment of the various types of lipoproteins in the blood (see part 3.4) such as total cholesterol, LDL-Cholesterol, HDL-Cholesterol and triglycerides.

- FH diagnosis at a young age is important. Treatment is more effective when started early and before cholesterol deposition in the blood vessel wall becomes too advanced.
There are some physical signs (figure 5) that might raise a suspicion of FH such as swollen tendons on the heels and hands (xanthoma) or yellowish areas around the eyes (xanthelasmas), but these are not always present in patients with FH.

**FIGURE 5:**
Visible physical signs of FH include swollen tendons on the back of the heel (often observed in adolescents) and yellow deposits in the skin around the eyes. A white deposit of cholesterol in the shape of an arc may also be seen at the edge of the colored part of the eye.

**IMPORTANT**
FH is suspected in people who have cardiovascular disease early in life together with a high level of cholesterol in the blood. Relatives of an identified patient should have a lipid profile assessment.
How is FH diagnosed?

FH is usually first recognized by abnormally high levels of LDL-C as seen on a cholesterol test and other signs and symptoms. In addition, genetic testing may be performed to confirm the diagnosis. A blood sample is taken so that DNA can be isolated from the cell nucleus of white blood cells. The DNA is then studied. FH is diagnosed by finding the defective gene for the LDL-receptor. Genetic testing involves a systematic search for gene defects throughout the entire LDL-receptor gene in chromosome 19. People who have the inherited gene defect will have FH. Close relatives, such as parents, brothers and sisters and children of someone with FH, have a 50% risk of also having inherited FH. Testing family members is crucial for early detection of the disease.

IMPORTANT

FH is detected by genetic testing and the identification of the abnormal LDL-receptor gene. Testing close family members enables early detection of the disease.

How early can FH be diagnosed?

People with FH usually have high total cholesterol and high LDL-Cholesterol from birth. It is recommended that parents with FH allow their children to be tested for FH before school age. A confirmed diagnosis at a young age is important, as early changes in diet and eating habits can help reduce the impact of FH in later life. If a child’s test is normal, there is no need to worry that FH will emerge in later years.

IMPORTANT

In families where FH has been diagnosed, it is advised that children are tested for FH at school age. This allows early dietary changes to encourage healthy eating.
There are two steps that help to reduce cholesterol:

- **Step 1:** Dietary changes
- **Step 2:** Medication

A change in diet is the first step in reducing cholesterol levels but if this does not reduce cholesterol enough, appropriate medication must be started as well. This is true for all people with FH. A change in diet can reduce high cholesterol. The aim of treatment (diet and medication) is to reduce the cholesterol level below the average for the population, i.e. less than 4.5 mmol/L or 175 mg/dl for adults. For those who are at high risk of, or who already have, cardiovascular disease the aim may be to lower cholesterol even further.

When individuals inherit a defective FH gene from both parents, functional LDL-receptors will be completely absent in cells. Neither drug nor dietary treatment alone or in combination may be sufficient to reduce extremely high cholesterol levels. In these patients, LDL-Cholesterol can be lowered mechanically by removing it from the blood by a dialysis-like cleansing technique (apheresis).

**IMPORTANT**

Dietary changes can lower LDL-Cholesterol. For patients with FH, this may not be enough and a change in diet should be combined with medication. For severe forms of FH additional dialysis-like cleansing treatment (apheresis) may be required.
Step 1: Dietary management of FH

a) How does diet affect LDL-Cholesterol?

All fat in food is a mixture of saturated and unsaturated fat. Saturated fats are found in animal products (such as dairy and meat products), hard margarines and in most cakes, cookies, ‘fast food’ and snacks. These saturated fats increase cholesterol, while unsaturated fat from plants and fish reduce or have a neutral effect on LDL-Cholesterol.

A low intake of cholesterol is recommended for people with a high level of cholesterol in the blood. The most important sources of cholesterol in the diet are from animal-derived foods such as egg yolks, meat and fatty dairy products like cheese, cream, and butter.

Fatty fish or fish oil are rich in omega-3 fatty acids, which have a beneficial effect on circulation and heart rhythm.

Omega-3 also reduces triglyceride levels. To ensure a sufficient intake of fish fats, eating fish at the main meal at least twice a week is recommended. If your diet is low in fatty fish, fish oil or another omega-3 supplement should be taken daily.

Fiber in coarse corn and whole wheat products, beans, peas, fruit, berries and vegetables, has a beneficial effect on cholesterol. Some types of fiber can bind cholesterol in the gut, and being excreted via the feces consequently reducing blood cholesterol levels. Foods high in fiber are also an important source of vitamins, minerals and antioxidants.

b) What sort of diet?

The diet for individuals with FH should be varied, balanced and heart-friendly. The aim is to gradually reduce the total intake of fats and cholesterol while, at the same time, focusing on eating the right type of fat, foods rich in fiber, fruit, berries and vegetables. For children with FH, dietary management should be supported by a change in diet for the whole family. It is important that healthy food habits are established early.

Changing diet takes time, often months or years, and requires regular follow up by a clinical nutritionist or doctor. What you eat on a regular basis is what matters; occasional deviations from the advised diet will not result in increased blood cholesterol.
Five important guidelines for a heart-friendly diet:

- Eat less fat, particularly less saturated fat
- Replace saturated fats with unsaturated fat
- Eat more foods containing fiber, vegetables and fruit every day
- Eat less cholesterol-rich foods
- Limit food and drinks high in sugar or alcohol

Step 2: Using medication

a) How does medication affect LDL-Cholesterol?

Medication that can lower your LDL-Cholesterol increases the number of LDL-receptors to enable better uptake of LDL-Cholesterol from your blood. Dietary changes must be combined with drug treatments to reduce cholesterol levels sufficiently. Several types of drugs are prescribed, which can be given alone or in combination. New drugs are also being developed. Drug treatment for FH is not limited to adults. If a patient comes from a family severely affected by FH, some experts recommend to start drug treatment from the age of 10-12 years, particularly if a parent experienced heart problems before the age of 40 years. Treatment is lifelong and will help achieve a longer and healthier life. The recommendation to start drug treatment in adults and children is based on LDL-Cholesterol levels in combination with the family history of cardiovascular disease.

b) Which drug treatments reduce LDL-Cholesterol and how?

If you have been diagnosed with FH, speak with your doctor regarding which approach to therapy would be best for you. Below on this page are descriptions of the therapies currently available.

The most important cholesterol reducing drugs used to treat FH are statins. These medications work by reducing cholesterol production in cells. In order to maintain cholesterol balance, cells increase their number of LDL-receptors to ‘take-up’ LDL-Cholesterol from the blood. The result is a reduction in blood LDL-Cholesterol.

Before treatment with statins, resins were used. These are substances which bind bile acids and inhibit their absorption from the gut into the liver. Bound to resins, bile acids are excreted with feces. To make up for the lost bile acids, the liver increases the uptake of LDL-Cholesterol from the blood to make new bile acids.

Cholesterol uptake inhibitors inhibit uptake of cholesterol from the gut, both from the diet and from cholesterol eliminated from the liver. Naturally occurring inhibitors of cholesterol uptake include plant sterols present, for example, in margarine. Plant sterols also reduce the uptake of cholesterol in the gut. Niacin-based therapies are another option for reducing cholesterol levels.

Dietary changes should continue after starting drug treatment. Both drug and dietary treatment must continue throughout life in patients with FH. A lipid profile should be performed when starting treatment, and afterwards to monitor how well the therapy is working.
Drug treatments include statins, resins, cholesterol uptake inhibitors and niacin-based therapies. Your doctor might treat you with one or more of these four types of medications. Medication, a healthy lifestyle, and heart friendly diet need to be continued throughout life. If LDL-Cholesterol is reduced sufficiently, cholesterol deposition in blood vessel and around the eyes or tendons will reduce.

**FIGURE 6:**
The effect of different drug treatments on the cell’s ability to produce cholesterol and on uptake of cholesterol from the gut.
Why is lifelong treatment important?

Your body makes cholesterol on a continuous basis. Your body is also exposed to more fat and cholesterol in food every day. Once LDL-Cholesterol has decreased as a result of treatment it is important to prevent it rising again. Anyone with FH whose body cannot regulate cholesterol properly will need to maintain a healthy diet and lifestyle and continue to take lipid lowering medication throughout life to keep LDL-Cholesterol levels under control.

IMPORTANT

For patients with FH who cannot regulate LDL-C properly, lifelong treatment with diet and medication is necessary to prevent cholesterol levels rising again.
Cardiovascular disease refers to diseases of the heart and blood vessels caused by atherosclerosis. Atherosclerosis involves a build up of fat (including cholesterol) and narrowing of the blood vessels, which can lead to reduced blood flow. When atherosclerosis reduces blood flow to an organ it can result in organ damage. If this happens in a blood vessel delivering blood to the heart, it causes a heart attack. If it happens in a blood vessel delivering blood to the brain, it causes a stroke.

Atherosclerosis begins with the deposition of cholesterol-filled cells in the inner wall of blood vessels (figure 7). This causes inflammation, the invasion of more cells, further deposits of cholesterol, the formation of scar tissue and hardening, resulting in the formation of a ‘plaque’. Plaques can narrow the blood vessels and reduce the flow of blood to the heart and other organs. In the heart, reduced blood supply can cause pain or discomfort, particularly after exercise. Plaques can rupture resulting in damage to the inside of the blood vessel, and the formation of a blood clot, which could severely restrict or block the blood supply. This causes an immediate lack of oxygen to the part of the organ supplied by the blood vessel and will result in tissue damage (major or minor). This is called an infarct. In this situation it is important to restore blood flow as quickly as possible to limit tissue damage. This can be achieved with blood clot-dissolving drug treatment, direct mechanical removal of the blood clot using a catheter (a small tube designed to go into blood vessels), and by increasing the size of the blood vessel with a small balloon that is expanded inside the vessel, followed by the insertion of metal netting in the blood vessel (stenting). These methods are often combined.
Figure 7: Cross-section of blood vessel in three stages. It shows the deposition of cholesterol in a blood vessel and the development of atherosclerosis.

**Important**

Heart attack and stroke are the end result of atherosclerosis. Atherosclerosis is a hardening and narrowing of the blood vessels caused by the build up of cholesterol and inflammation to form a plaque. A plaque decreases the size of the inside of the blood vessel. A damaged plaque can cause a blood clot, with very rapid narrowing or blockage of the blood vessel.

**What are risk factors?**

Risk factors are characteristics (biological or environmental) that increase the likelihood of a person developing atherosclerosis and cardiovascular disease. In a group of people with a certain risk factor, more people will develop cardiovascular disease over a period of time, compared to a similar group of people without that risk factor. One of the most important risk factors for cardiovascular disease is a high level of LDL-Cholesterol with a low level of another type of cholesterol called High Density Lipoprotein-Cholesterol (HDL-C) (see part 3.4). Health related risk factors include diabetes and high blood pressure, which can be influenced by lifestyle factors such as being overweight, a low intake of fruit and vegetables and limited physical activity. Smoking is one of the most important lifestyle-related risk factors. Age and gender are also risk factors. Cardiovascular disease increases with age, and men develop the disease approximately 10 years earlier than women. If you have several risk factors the likelihood of developing cardiovascular disease is higher.
3 Is it possible to reduce the risk of cardiovascular disease in FH?

YES! Several studies have shown that reducing high LDL-Cholesterol reduces the risk of cardiovascular disease. Cholesterol deposition depends partly on how high the LDL-Cholesterol level is, and how long the LDL-Cholesterol has caused damage to the blood vessels. By lowering LDL-Cholesterol levels, individuals with signs of cardiovascular disease will reduce hardening and narrowing of the blood vessels. It is important to start reducing LDL-Cholesterol as soon as possible as cholesterol deposition in FH can be reversed. Stopping smoking is crucial in reducing the risk of cardiovascular disease.

IMPORTANT
Lowering LDL-Cholesterol in FH is important as this reduces hardening and narrowing of the blood vessels and decreases the risk of cardiovascular disease. Stopping smoking is crucial.

4 What are lipoproteins?

Lipoproteins are particles that transport fat around in the blood (figure 8). Fatty substances called triglycerides and cholesterol cannot be dissolved in the blood and depend on a transport system that takes them from the organs that produce them (the gut and liver) to cells. The two most important lipoproteins in this transport system are called Low Density Lipoprotein (LDL) and High Density Lipoprotein (HDL). Both of these lipoproteins transport cholesterol to form LDL-Cholesterol (LDL-C) and HDL-Cholesterol (HDL-C).

FIGURE 8:
A diagram of lipoprotein.
The cholesterol transported in HDL is called ‘good’ cholesterol. One of the important functions of HDL is to transport cholesterol from the cells and tissue back to the liver. High HDL-Cholesterol is good as it takes cholesterol out of cells and the blood and helps to prevent excess cholesterol. HDL also removes cholesterol deposited in the walls of blood vessels. Doctors can distinguish the two types of cholesterol (LDL-Cholesterol and HDL-Cholesterol) and try to help patients achieve a ‘healthy’ balance between the two.

**FIGURE 9:**
HDL-Cholesterol and LDL-Cholesterol work together to keep cholesterol in the blood at a healthy level.

**IMPORTANT**

The cholesterol transported in LDL is often described as ‘bad’ cholesterol because the cholesterol in LDL, that is not picked up by cells, is deposited in blood vessels causing them to harden and narrow. People with high HDL-Cholesterol and low LDL-Cholesterol are at lower risk of developing atherosclerosis.
What are lipids, cholesterol and triglycerides?

LIPIDS are a collective term for different fatty substances such as cholesterol and triglycerides. Levels of cholesterol and triglycerides in the blood can be established by a blood test.

CHOLESTEROL is a fatty substance used to build cell walls. Cholesterol also has several important functions in making hormones, vitamin D and bile acids. All cells can produce cholesterol but most cholesterol is made in the liver and gut. The liver is the main organ for breaking down cholesterol by converting it to bile acids. If excess cholesterol is produced or eaten, or if the cholesterol is broken down too slowly, there will be excess cholesterol in the blood. Cholesterol may then be deposited in the walls of blood vessels resulting in the onset of atherosclerosis.

TRIGLYCERIDES represent the majority of fats in the blood. Triglycerides are fatty compounds made of glycerol and fatty acids (figure 10). Fatty acids can be saturated, unsaturated and polyunsaturated, and this determines whether the fatty acids increase or reduce blood cholesterol. In order to transport triglycerides in the blood, the gut has to make cholesterol, which then enters a transport particle (lipoprotein). This is why a high intake of fat increases blood cholesterol.

**FIGURE 10:**
Fat is built up from glycerol and fatty acids.

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**IMPORTANT**

Fat from diet contains cholesterol and triglycerides. A high intake of cholesterol and fat, especially saturated fat, increases blood cholesterol.
Physical activity is recommended for all age groups. Regular physical exercise has beneficial effects on the lipid profile by reducing LDL-Cholesterol and triglycerides and increasing HDL-Cholesterol. Taking part in physical activity can reduce blood pressure and reduces the risk of being overweight, developing type 2 diabetes and some forms of cancer.

**IMPORTANT**

Thirty minutes of daily activity (sufficient to increase the pulse) at least five times per week, is recommended.
Does smoking affect lipids?

Smoking is particularly hazardous for patients with FH and it is strongly advised that FH patients do not smoke. Smoking causes additional damage to the blood vessels, and reduces HDL-Cholesterol (‘good cholesterol’) and triples the risk of cardiovascular disease – even when blood cholesterol is within the normal range. Untreated FH patients already have a risk of developing cardiovascular disease that is 25 times greater than people who do not have FH.5.

You have learned that Familial Hypercholesterolemia is a hereditary disease that runs in families and is caused by a defective gene for the LDL-receptor.

A defective LDL-receptor can lead to an increase in LDL-Cholesterol in the blood and this can give rise to atherosclerosis and diseases of the heart and blood vessels.

Cardiovascular disease at a young age is indicative of FH.

It is possible to find out if family members are affected by FH by measuring their LDL-Cholesterol and finding out whether they carry the defective gene.

Most importantly, you have learned how you and your affected family members can reduce the risk of cardiovascular disease by adopting a healthy lifestyle, a heart-friendly diet and by taking LDL-C lowering treatments.

This booklet may also serve as a starting point to help you discuss your disease with your doctor.
**GLOSSARY**

**APO B:** To transport cholesterol into specific cells, the LDL-C particle has a specific protein attached to it named Apolipoprotein B or ApoB. ApoB acts as a bridge between the LDL-C particle and the cells in your body that carry the LDL-receptor.

**BILE ACIDS:** The liver produces bile acids and cholesterol. Bile acids are excreted into the gut when we eat. This helps fats in the gut enter the blood.

**CELL:** Cells are the building blocks of the body and can be compared to building bricks. The body is made up of approximately 100,000,000,000,000 (100 trillion) cells.

**CHOLESTEROL:** Cholesterol is a fatty substance. It is stored in the body and is found in all foods derived from animals. Large amounts of cholesterol can be stored in the liver.

**CHROMOSOMES:** The hereditary material present in the nucleus of each cell.

**CHYLOMICRONS:** Large lipoprotein particles that transport lipids from the gut to cells.

**CLINICAL NUTRITIONISTS:** A person who is an expert in diet and the management of diet in disease. He or she will have undergone 4-5 years of university study.

**DNA:** The molecules that builds our genes in our chromosomes.

**FAMILIAL HYPERCHOLESTEROLEMIA (FH):** This is a hereditary cell defect. The defect means that the cells cannot take up enough cholesterol from the blood. This results in high blood cholesterol.

**GALL BLADDER:** A sac attached to the liver that stores bile.

**GENE:** A section of DNA that codes for a certain protein.

**HDL-CHOLESTEROL:** Also referred to as ‘good cholesterol’. It is okay to have a lot of this in your blood.

**HYDROGENATED FAT/HYDROLYSED FAT/HARDENED FAT:** These are all different names for the same fat. This fat begins as an unsaturated fat and is then turned into a saturated fat. Conversion of unsaturated fat into saturated fat is quite common because saturated fat lasts longer.
**LDL-CHOLESTEROL:** Otherwise referred to as ‘bad cholesterol’. It is best to have only a small amount of LDL-Cholesterol in the blood.

**LIPIDS:** Fats.

**LIPOPROTEINS:** Lipoproteins are small packages made up of cholesterol, triglycerides and protein and are a means by which fats can be transported in the blood. There are various kinds of lipoprotein, the most important being HDL and LDL.

**MYOCARDIAL INFARCTION:** Heart disease that can occur suddenly when the blood supply to the heart is blocked. Causes of myocardial infarction include high cholesterol and smoking.

**RECEPTORS:** Receptors are ‘tentacles’ located on the outside of the cells. The tentacles catch substances in the blood needed by cells. There are special receptors for lipoproteins. Individuals who have inherited Familial Hypercholesterolemia (FH) have too few functional receptors for LDL-lipoproteins. This means that the cholesterol from LDL remains in the blood and can be deposited in blood vessels in the form of plaque, which results in narrowing of the blood vessels.

**SATURATED FAT:** The body can store this fat itself. A diet high in saturated fat results in an increase in blood cholesterol. Saturated fat is the type of fat that hardens in the fridge.

**TRIGLYCERIDES:** Another word for fats. The fats in food and blood are triglycerides. It is good to have low levels of triglycerides in the blood.

**UNSATURATED FAT:** This fat is present in most foods from plants and fish. The body cannot create sufficient unsaturated fat itself so we have to eat foods containing this fat. Unsaturated fats remain soft or liquid in the fridge.

**VLDL:** Very Low Density Lipoprotein; when fat from the gut reaches the liver it is packed into large, fat-rich particles, which are called VLDL.
The author, Dr. Leiv Ose, has worked with patients affected by lipid disorders since 1970. He has been involved in scientific activities related to that field; his main interests are preventive cardiology and genetic hyperlipidemias such as Familial Hypercholesterolemia (FH). He has been the head of the Lipid Clinic at Oslo University Hospital, Norway, since 1984. The Lipid Clinic in Oslo is one of the few Lipid Clinics focused on FH. The Clinic has extensive experience with dietary and drug treatment of FH including treatment of young patients. His Lipid Clinic is the FH reference center in Norway.
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