Inheriting Heart Disease
This booklet is one of the publications in our patient information series. For a complete list of patient booklets, see page 19.

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Introduction

Heart attack and stroke are the most common causes of death in Ireland each year. By providing information to the public, the Irish Heart Foundation aims to prevent as many of these deaths as possible. This information is vital if we are to deal effectively with these health-related issues. This booklet is one of many we produce to provide information on preventing and treating heart disease and stroke. The booklet focuses on an inherited condition with very high cholesterol levels known as familial hypercholesterolaemia (FH). Although it is inherited, a lot can be done to treat the condition, especially when it is discovered early.
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Summary

A high blood cholesterol level is, like smoking and high blood pressure, one of the many factors that increase the chance of getting heart disease. For most people, the blood cholesterol level depends on diet and, to a lesser extent, on the genes inherited from our parents. In the condition familial hypercholesterolaemia (FH), the cholesterol level is very high and is mainly caused by inherited genes. If a parent has FH, on average, half of their children are likely to also have the disease. Finding FH at an early age leads to better treatment and a lower chance of having a heart attack. Treatment involves eating a healthy diet, taking regular exercise, controlling weight, not smoking and taking medicines to lower the cholesterol level. This booklet provides information on FH and discusses the treatment options.
Glossary

**Apheresis**: a filtering technique to wash the blood clear of fats.

**Cholesterol**: a soft fatty material needed by each cell in the body. Cholesterol is present in foods but the body makes most of it from saturated (animal) fat eaten in the diet.

**Familial hypercholesterolaemia (FH)**: an inherited condition with very high blood cholesterol levels.

**Gene**: genes allow inherited characteristics to be passed from one generation to the next. A child receives one half of their genes from each parent. Like microchips in a computer, genes control the function of each cell.

**Heart attack**: a sudden loss of blood supply to the heart muscle tissue that is usually due to a clot in one or more arteries. This often results in a scar forming where the muscle is damaged.

**High density lipoprotein (HDL)**: a type of cholesterol that protects against heart disease.

**Low density lipoprotein (LDL)**: a type of cholesterol that causes coronary artery disease.

**LDL receptor**: the socket on each cell for LDL cholesterol to plug into.

**Lipoprotein (a) or LP(a)**: a type of blood fat that is strongly linked to the risk of heart disease.

**Millimole per litre or mmol/L**: the unit of measurement of chemicals including cholesterol.

**Monounsaturated fat**: a type of fat associated with a reduced chance (compared with saturated fat) of heart disease. It has a different chemical structure to saturated and polyunsaturated fat.
**Nicotinic acid**: a medication used to lower cholesterol levels.

**Polyunsaturated fat**: a type of fat associated with a reduced risk (compared with saturated fat) of heart disease. It has a different chemical structure to saturated and monounsaturated fat.

**Resin**: a medication used to lower cholesterol levels.

**Saturated fat**: the type of fat from which cholesterol is made. The term saturated refers to the chemical structure of the fat. It is strongly associated with heart disease.

**Statin**: a medication used to lower cholesterol levels.

**Triglycerides (TG)**: a type of fat in the blood. A high blood level increases the chance of developing heart disease.

**Xanthelasma**: a yellow patch near the eyes that shows you have high blood cholesterol levels.

**Xanthoma**: a yellow lump on the elbows, knuckles and tendons that shows you have high blood cholesterol levels.
**Cholesterol and heart disease**

Cholesterol is a soft fatty material that forms part of each cell in the body. Some cholesterol is needed but heart disease develops when the cholesterol level in the blood is too high. Most cholesterol is made in the liver from the fat we eat, especially saturated – animal fat. The saturated fat we eat has a greater effect on increasing blood cholesterol than the amount of cholesterol in the diet.*

The factors that together increase the chance of having a heart attack are well known. They are: high blood cholesterol, high blood pressure, smoking, diabetes, lack of exercise and being overweight. For most people who develop heart disease, some of these factors are present from an early age. If detected early, many heart attacks could be prevented.

*See the IHF leaflets “Good Eating for a Happy Heart” and “What is Cholesterol all About” which will give you more information and make your food choices easier.

**What is the normal cholesterol level?**

The answer to this is it depends! In Ireland as a whole, the average cholesterol level is about 6 mmol/ L. Cholesterol is usually measured in units called millimoles per litre: (mmol/ L). Ireland also has one of the highest rates of heart disease in the world. So, while a cholesterol level of 6 is common, this does not mean it is good for health.

In recent years, we have learned that what is normal for some people is not normal for others. For example, if you have had a heart attack already it is important to reduce cholesterol to quite a low level. In other people who have never had a heart attack, a slightly higher level may be acceptable. For most people who have never had a heart attack and want to prevent their first one, the ideal cholesterol level is below 5 mmol/ L. Many first heart attacks in Ireland could be prevented if the average cholesterol was below 5.
What is familial hypercholesterolaemia?
A small number of people in Ireland (one in 500) have the condition familial hypercholesterolaemia (FH), where an abnormal gene leads to high cholesterol levels. Unlike many people who develop heart disease in middle and old age, the very high cholesterol level in FH leads to heart disease in early life.

Without treatment, up to eight in every ten men with FH will have a heart attack. Half of these will die before 60 years of age. Women with FH also develop early heart disease but usually some years later. Some with FH may be disabled or may die from heart disease very early in life, often as early as 20 years. Because of this, FH should be detected and treated as early as possible.

What causes the high LDL cholesterol levels in FH?
Normally, the fat and cholesterol we eat is brought into the bloodstream from the stomach to the liver. The liver then makes cholesterol particles called low density lipoproteins (LDLs) and takes them to all parts of the body where they are needed by cells. When it reaches a cell, LDL plugs itself into a socket on that cell called a receptor. The cell then absorbs the cholesterol and uses it. In people with FH, half of these receptors are missing. This means that cholesterol levels in the blood remain high because the cell cannot take-up the cholesterol. Very rarely (one in a million), there are no LDL receptors on the cell. In these rare cases, cholesterol levels are so high that heart disease develops in infancy.
**Good and bad cholesterol**

**LDL cholesterol**
Cholesterol is made in the liver. It is then carried in the bloodstream in small particles called lipoproteins. These low density lipoproteins (LDL) carry it to where it is needed. On the way, if too much LDL is floating in the blood, it sticks to the wall of blood vessels. The lower the LDL, the better. Levels below 2.6mmol/L are best for patients who have had a heart attack. For others, the LDL level should be no greater than 3mmol/L.

**HDL cholesterol**
High density lipoprotein (HDL) does the opposite of LDL. HDL carries cholesterol from the bloodstream back to the liver. This is good or healthy cholesterol, the higher the HDL, the better. Levels over 1mmol/L are good and over 1.5 are best. HDL can be increased by exercise, losing weight, stopping smoking and by eating a healthy diet. So, keep LDL low and HDL high.

**Triglycerides**
Triglycerides (TG) (pronounced: try-gliss er ides) are a type of fat in the blood. A high level can increase heart disease risk. TGs make the blood sticky. High cholesterol and high TG levels mean you may have another inherited disorder. It is called Familial Combined Hyperlipidaemia or FCH. Treatment of FCH is very similar to FH.

**LP (a)**
Lipoprotein (a) or LP(a) is a particle like LDL. The blood level depends on a gene. High Lp(a) levels (over 30mg/dl) (over 300mg/L) increase the chance of heart disease. Lp(a) does most harm when LDL levels are high. So, once again, the lower the LDL, the better.
How can I tell if I have FH?

Heart disease in early life is typical of FH. If a family member has developed or died from heart disease early in life, you too may be likely to develop it. In some families, a very high cholesterol level may have been found in a family member. This may be the only pointer. Signs of FH may be seen in the skin and tendons, such as pale yellow lumps near the knees, ankles, knuckles, elbows and back of the hands. These are called xanthomas (pronounced: zan-tho-mas). Yellow marks around the eyes are called xanthelasmas (pronounced: zan-thel-as-mas). Finally, a pale or white ring around the outside of the iris, the coloured part of the eye, may be a sign of high cholesterol levels and FH. Sometimes, these signs may not be present.

What about my family?

FH is passed from parent to child. If one parent has FH, half of their children are also likely to have FH. This means that if you have FH, it is important that all the members of your family have a cholesterol test. If needed, early treatment can then be started.

Treatment of FH

We now know that lowering high cholesterol levels reduces the risk of heart attack, angina and the need for heart surgery. A healthy diet and lifestyle are important for everybody. This is even more important in people with FH. This means reducing fat intake, especially saturated or animal fat, and eating a balanced diet with foods rich in other nutrients. Usually, a change in diet can reduce cholesterol levels by about one fifth. However, in FH, diet alone is not enough to lower the blood cholesterol enough. Tablets are needed also.
**A healthy diet**

Avoiding foods rich in saturated fat will reduce blood cholesterol levels. Some foods such as eggs and offal are rich in cholesterol and should also be limited. However, as cholesterol and saturated fat are usually found together in the same foods, by avoiding foods rich in saturated fat, you are also avoiding the high cholesterol foods.

**The principles of healthy eating are:**

❤️ Reduce the total amount of fat you eat, especially saturated fat. Choose low fat dairy products such as low fat milk, low fat cheese, diet or low fat yoghurts.

❤️ Eat chocolate, biscuits and crisps only as an occasional treat.

❤️ Eat more fruit and vegetables. Aim for four or more portions each day.

❤️ Switch to monounsaturated or polyunsaturated (vegetable oil) spreads and use sparingly. New cholesterol-lowering spreads are available.

❤️ When cooking, use monounsaturated or polyunsaturated oils. Use very small amounts.
Avoid foods cooked in fat, especially chips and take-away foods. Grill, bake, boil, braise, steam or microwave food instead of frying.

Eat more wholemeal bread, cereal, pasta and rice. Include oat bran breakfast cereals regularly.

Try to eat oily fish once or twice per week for example mackerel, herring, trout, sardines and salmon. If eating canned fish, choose fish in brine and drain well. Eat white fish often.

Choose lean red meat about 3 times a week and poultry (chicken and turkey) without skin at least twice a week. If you enjoy sauces, choose tomato-based varieties instead of cream-based sauces. Drain fat from the roasting tray before making gravy.

Try to include no more than 4 eggs a week.

You should drink alcohol in moderation. Healthy guidelines for adults suggest 14 units per week for women and 21 units for men. 1 unit is a glass of wine or a half pint of beer. A measure of spirits is 1.5 units.

See: Our booklet, What is Cholesterol all about?
Smoking

If you smoke, you should try to stop. This is the most important thing you can do to improve your health. It is not easy to give up smoking and you need a health plan:

- Decide on what day you are going to give up
- Take each day at a time.
- Practise saying “No thanks I don’t smoke” in front of a mirror.
- Help is at hand. Having support will double your chance of giving up for good.
- Ask your doctor about nicotine replacement such as chewing gum or patches.
- Read self-help books. Talk to your doctor or nurse or join a group. Contact your health board for more information on support groups.
- Use any method that you think will make it easier for you to give up.
- Stay stopped, for your health and your future wellbeing.

By not smoking, you will avoid the ongoing damage that it causes to many body systems. Smoking damages your heart and circulation and increases the chance of blood clots. It also causes lung cancer, emphysema and chronic bronchitis. Smokers are also more likely to develop other cancers. Tobacco smoke is a poison. When you breathe in smoke, carbon monoxide is taken up by the red blood cells and carried around your body. Normally, these cells carry oxygen. So, smoking prevents the blood from carrying vital oxygen and can cause angina.
**Exercise**

*Be active! Regular exercise:*

- Helps maintain high levels of HDL - (healthy cholesterol).
- Lowers LDL - (bad cholesterol).
- Reduces triglyceride levels.

**Exercise:**

- Helps weight control when combined with healthy eating
- Reduces the chance of suffering from diabetes
- Encourages healthy bloodflow and reduces blood pressure
- Allows the brain to release hormones that produce a sense of wellbeing to help reduce stress
- Helps a smoker to overcome the habit
- Strengthens muscles and bones and reduce osteoporosis (brittle bones)
How much exercise should I take?
If you have a heart condition, follow the advice of your doctor. Always build up exercise gradually. Warm up by walking at a gentle pace for the first 5-10 minutes. Slow down towards the end of the walk. In cold weather, wear a cap or hat to prevent you losing heat through the head. On hot days, do your exercise in the cool of the morning or evening.

Even small amounts of exercise as part of your daily routine will improve health. For example, take the stairs instead of the lift, walk to collect the paper or to go shopping, walk to work or walk the dog. Wear laced walking shoes with stockings and light warm clothing. In general, you should do aerobic exercise, (exercise that uses the heart, lungs, large joints and muscles), for example walking, swimming and cycling, for 20-30 every day.

Stop exercising if you develop pain in the chest, feel very tired or find it hard to breathe. Do not compete or push yourself beyond your limits.
**Medicines for FH**

A healthy diet is the best way to treat FH. However, most people with FH will need tablets to lower cholesterol levels. Various medicines are now available to do this. Before taking any new tablets, make a list of all medicines you are already taking (see page 17) and tell your doctor. It may be necessary to change the dose or timings of some tablets.

**Statins**

Statins are taken as tablets and slow down the production of cholesterol in the liver. They are well tested and very effective. Side-effects are rare but you may develop aching muscles or allergies. If so, tell your doctor. Statins may affect your liver, but this can be checked by a blood test. The effect is reversed once the drug is stopped. Statins are not used in children before puberty, during pregnancy or in women who might become pregnant.

**Resins**

Resins are taken as powder and mixed with water, fruit juice, or yoghurt. They are not absorbed into the body and are safe in children. However, extra vitamin A, D, E, K and folic acid may be necessary. Check with your doctor. You may suffer from wind and constipation but this can be reduced by building up the dose gradually.

**Fibrates**

The fibrates are taken as tablets. They are useful when triglycerides and cholesterol are both raised and when HDL is low. These drugs can occasionally cause stomach upsets.
Nicotinic acid

Nicotinic acid is effective in lowering both cholesterol and triglyceride levels. It is cheap but not very popular because of side effects such as flushing. These can be reduced by increasing the dose gradually, taking the drug with meals and by taking aspirin.

Other treatments for FH

LDL Apheresis

LDL apheresis filters the blood and removes the cholesterol. Like kidney dialysis, it is done in specialist centres and takes several hours for each treatment. It is not available in Ireland. In studies where this procedure was compared with statin medication, there was no advantage to using apheresis.

Name: ________________________________________________________________

My medicines: __________________________________________________________

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Considering your risk factors!

Do you smoke?

Is your blood pressure on target (less than 130/85)?

What is your cholesterol level?
Record your readings in the table below.

- Do you exercise regularly (more than 3, half-hour sessions per week)?
- What is your weight?
- Is there a history of heart disease in your family?

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Inheriting Heart Disease is an Irish Heart Foundation patient information publication. Other titles in this series are:

- Stroke, a guide for those affected by stroke and their carers
- Things you should know about blood pressure
- Things you should know about heart surgery
- Heart attacks
- Living well with heart failure
- Step by step through angina
- Step by step through cardiac catheterization and angioplasty

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