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STEP BY STEP THROUGH INHERITED HEART DISEASE

FAMILIAL HYPERCHOLESTEROLAEMIA

Produced by the Irish Heart Foundation

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The Irish Heart Foundation

The Irish Heart Foundation is the national charity fighting heart disease and stroke. More people in Ireland die from these causes than from cancer, road deaths and suicide combined. We work to bring hope, relief and a better future to Irish families. We support pioneering medical research, campaign for improved patient care and provide vital support and information for patients. In hospitals, schools and workplaces, we support, educate and train people to save lives. As a charity we depend on your ongoing support - through your donations or by giving of your time as a volunteer or on a training course.

For more information or to donate, visit our website: www.irishheart.ie

Heart and Stroke Helpline:

Locall 1890 432 787 (Monday to Friday 10.00am to 5.00pm)

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Introduction

This book is about an inherited condition called familial hypercholesterolaemia (FH). People with this condition have a very high cholesterol level. Although it is inherited, a lot can be done to treat the condition, especially when it is discovered early.

A high blood cholesterol level is one of the many factors that increase your chance of getting heart disease or having a stroke. For most people, your blood cholesterol level depends on what you eat and drink and, to a lesser extent, on the genes inherited from your parents. With 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 or stroke. Treatment involves keeping to a heart-healthy eating plan, being physically active, controlling your weight, not smoking and taking medicines to lower your cholesterol level.

This book provides information on familial hypercholesterolaemia and the treatment options available.

A lot can be done to treat familial hypercholesterolaemia (FH), especially when it is discovered early.

Cholesterol, heart disease and stroke

Cholesterol is a soft waxy material that forms part of each cell in your body. You need a certain amount of cholesterol for your body cells and to produce important hormones. However having too much cholesterol in your blood can lead to heart disease and stroke.

Cholesterol is made in your body, mainly by your liver. The body can produce all the cholesterol it needs to carry out its many functions and can usually maintain a healthy level of cholesterol in your blood. However, sometimes the balance goes wrong and there is an increase in blood cholesterol. This can be due to conditions such as FH, or from eating too much saturated (animal) fat or too many foods from the top shelf of the food pyramid.

The factors that together increase your risk of getting heart disease or having a stroke, are well known. They are: high blood cholesterol, high blood pressure, smoking, diabetes, lack of physical activity and being overweight.

For many people who develop heart disease or have a stroke, some of these risk factors are present from an early age. If risk factors are detected early, many heart attacks and strokes could be prevented. For more information, see our booklets: *A healthy cholesterol, Be active, Lose weight, Healthy eating, Quit smoking, Manage your blood pressure*, and *Manage your stress.*

Having too much cholesterol in your blood can lead to heart disease and stroke.



What is a healthy cholesterol level?

Currently, a healthy total cholesterol level is considered to be no more than 5 mmol/ l. Cholesterol is usually measured in units called millimoles per litre, written as mmol/ l.

If you have had a heart attack or a stroke, or if you have FH or diabetes, it is important that your cholesterol level is no more than 4.5 mmol/l.

Good and bad cholesterol

There are two main types of cholesterol – HDL cholesterol (high density lipoprotein) and LDL cholesterol (low density lipoprotein). Triglycerides and lipoprotein (a) are two other types of fats that are also found in blood cholesterol.

LDL (bad) cholesterol

Cholesterol is made in your liver. It is then carried in your blood in small particles called lipoproteins. Low density lipoproteins (LDL) carry it to where it is needed. On the way, if too much LDL is floating in your blood, it sticks to the walls in your arteries – making them narrow. This reduces the blood supply to your heart or brain.

High levels of LDL cholesterol increase your risk of heart disease and stroke. The lower your level of LDL cholesterol, the better – ideally it should be no more than 3 mmol/l. If you have heart disease or diabetes, your LDL cholesterol level should be no more than 2.5 mmol/l.

HDL (good) cholesterol

High density lipoprotein (HDL) does the opposite of LDL. HDL carries cholesterol left behind in your arteries back to your liver where it is broken down and passed out of your body. HDL is good or healthy cholesterol, the higher your HDL, the better. Levels over 1mmol/ I are good and over 1.5 are best. HDL can be increased by being more physically active, being a healthy weight, not smoking and by keeping to a heart-healthy eating plan.

For people with established heart disease or diabetes

Total cholesterolNo more than 4.5 mmol / ILDL cholesterolNo more than 2.5 mmol / I

Triglycerides

Triglycerides (TG) are another type of fat in your blood. They make your blood sticky. Too much triglyceride in your blood can increase your chances of getting heart disease or having a stroke. A high level of TG can be caused by smoking, drinking too much alcohol, eating too many sugary foods, physical inactivity, being overweight or obese and diabetes. A high TG level may be a sign of thyroid or kidney disease, your doctor will check for this. 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. Your genes determine the amount of LP (a) in your blood. 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.

What is familial hypercholesterolaemia (FH)?

About one in every five hundred people has the condition familial hypercholesterolaemia (FH), where an abnormal gene leads to a high cholesterol level. Unlike many people who develop heart disease or have a stroke 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 people will die before 60 years of age. Women with FH also develop early heart disease but usually some years later than men.

Some people with FH may be disabled or may die from heart disease very early in life, often as early as 20 years of age. Because of this, FH should be detected and treated as early as possible.

What causes the high LDL (bad) cholesterol levels in FH?

Normally, the fat and cholesterol you eat is brought into the bloodstream from your stomach to your liver. Your liver then makes cholesterol particles called low density lipoproteins (LDLs) which travel in your blood to all parts of your body where they are needed by cells. When it reaches a cell, the 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 your blood remain too high (often between 7.5 and 15 mmol/l) because there are not enough receptors on your cells to absorb all the cholesterol.

Very rarely (one in a million), there are no LDL receptors on the cells. In these rare cases, cholesterol levels are so high (between 20 and 30 mmol/l) that heart disease develops in infancy.

How can I tell if I have FH?

Diagnosing FH is not always a simple process. However there are some signs that your doctor will look for. These include:

- A cholesterol test that shows you have a very high cholesterol level.
- A family member has developed or died from heart disease early in life.
- Other people in your family have been diagnosed with FH.
- You have had a heart attack. This is more significant if you are under 40 years of age.

Signs of FH can sometimes be seen in your skin and tendons, these include:

- Pale yellow lumps near your knees, ankles, knuckles, elbows and back of your hands. These are called xanthomas.
- Yellow marks around your eyes are called xanthelasmas.
- A pale or white ring around the outside of your iris, the coloured part of your eye, may be a sign of a high cholesterol level and FH.

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 to find out who else might be affected. Early treatment can then be started.

Use the Food Pyramid to plan your healthy food choices every day and watch your portion size



Treatment for FH

Lowering a high cholesterol level reduces the risk of stroke, heart attack, angina and the need for heart surgery. A healthy lifestyle is important for everybody. It is even more important for people with FH. This includes reducing fat intake, especially saturated or animal fat, and eating a variety of foods, using the Food Pyramid as your guide. Usually, a change in eating habits to more heart-healthy options can reduce a person's cholesterol level by about one fifth. However, if you have FH, healthy eating alone will not lower your blood cholesterol enough. You will also need to take medicine.

Healthy eating

Use the Food Pyramid as your guide to heart-healthy eating. Avoiding foods rich in saturated fat will help reduce blood cholesterol levels. Some foods such as eggs and offal are rich in cholesterol and should also be limited if you have FH.

- Eat more fruit and vegetables, at least 5 portions per day.
- Eat more wholegrain varieties of bread, cereals, pasta and rice. Leave the skins on potatoes.



- Eat less (saturated) fatty foods.
 Replace them with monounsaturated and polyunsaturated fats.
- Eat oily fish twice a week (salmon trout, herring, mackerel or fresh tuna).
- Drink less alcohol and no more than the recommended upper limits of 17 standard drinks (SD) a week for men and 11 standard drinks a week for women.
- Choose lean meats. Trim the fat off meat and take the skin off chicken.

- Drain the oil from cooked dishes containing minced meat.
- If you enjoy sauces, choose tomato-based varieties instead of creambased sauces.
- Use low-fat dairy products milk, cheese and yoghurts.
- Instead of butter, choose a low fat polyunsaturated or monounsaturated spread.
- Try to include no more than 4 eggs a week.
- Choose low fat, healthy ways of cooking such as grilling, baking, boiling and steaming instead of frying.
- Be more physically active every day.

See our leaflets: *A healthy cholesterol* and *Healthy eating* for more information.

Cholesterol lowering foods

There are foods available which can help you lower your cholesterol. These include spreads, yoghurts and milk. These foods have ingredients called plant sterols and plant stanols, which stop your body absorbing cholesterol.

If you have FH, these foods may help lower your cholesterol. It is important to ask your doctor or dietitian if you would benefit from using these products as they can be expensive. The cholesterol lowering medicine your doctor has prescribed may be sufficient for your condition.

Foods containing plant sterols and plant stanols are not suitable for women who are pregnant or breast feeding. They are suitable for children with FH, but please ask your doctor or dietitian if your children should take these foods. People who don't have FH or don't have high cholesterol should not use these cholesterol lowering foods.

Fats in food

Fats in food are a mixture of saturated fats and trans fats that increase your blood cholesterol, and unsaturated fats that are good for your heart health and can lower your cholesterol level.

Saturated fats can raise your LDL (bad) cholesterol and increase your risk of heart disease and stroke.

Trans fats lower your HDL (good) cholesterol and increase your LDL (bad) cholesterol.

Unsaturated fats (monounsaturated and polyunsaturated fats):

- Monounsaturated fats can help lower your LDL (bad) cholesterol.
- **Polyunsaturated fats:** There are two main types of polyunsaturated fats, omega 3 fats or omega 6 fats.
 - **Omega 3 fats** help lower your triglyceride level, help prevent blood clots and help your heart to keep a healthy rhythm.
 - Omega 6 fats can help lower your LDL (bad) cholesterol.

While unsaturated fats are good for your heart-health, all fats are high in calories. If you are trying to lose weight, it is important to reduce the total amount of fat you eat.

Saturated fats & trans fats increase LDL (bad) cholesterol

Saturated fats are found in:

- Butter.
- Lard.
- Hard margarine.
- Cheese.
- Cream.
- Whole milk.
- · Fatty meats.
- Cakes.
- Biscuits.
- Chocolate.
- Coconut oil.
- Palm oil.
- Many ready-meals and processed foods. Check food labels to find out how much saturated fat they contain.

Trans fats are found in:

- Cakes.
- Biscuits.
- Pastries.
- · Deep-fried foods.
- · Hard margarines.

Any foods or ready-meals containing hydrogenated oil or hydrogenated fat are likely to have trans fats. Unsaturated fats reduce LDL (bad) cholesterol

Polyunsaturated fats: Omega 3 fats are found in:

- Oily fish such as, salmon, mackerel, trout, herring, sardines and fresh tuna.
- Omega 3 fats in tuna are normally lost during the tinning process. If you are using tinned tuna, make sure the label says that omega 3 fats have been replaced.

Omega 6 fats are found in:

- Vegetable oils such as sunflower, safflower, corn, soya bean and sesame oils.
- Soya beans.
- Walnuts, hazelnuts and brazil nuts.

Monounsaturated fats are found in:

- Olive oil.
- Peanut oil.
- Rapeseed (canola) oil.
- Spreads that contain these oils.
- Avocadoes.
- Seeds.
- · Cashew nuts.
- Almonds.
- Peanuts.

Quitting smoking

If you smoke, try to quit. This is the most important thing you can do to improve your health. Smokers are twice as likely to suffer a heart attack as non-smokers. Smoking increases LDL (bad) cholesterol and reduces HDL (good) cholesterol.

Preparing to quit

Write down your reasons for quitting. Think about the good and not so good things about quitting smoking. Compare your lists and keep them close at hand to remind you why you want to quit.

Make a date to quit

Pick your date and stick to it. Think about what methods and supports are available to help you quit and have them ready before your quit date. The support of family and friends will help you. If needed, you can also talk to your pharmacist and your doctor about other products to help you quit. See our leaflet: **Quit smoking** for more information.

Quitting smoking is the most important thing you can do to improve your health.

Physical activity

Regular physical activity can help reduce your cholesterol level and blood pressure. Over 20% of heart disease and 10% of stroke is due to physical inactivity. Being more active helps maintain high levels of HDL (good) cholesterol and lowers LDL (bad) cholesterol.

Being physically active for at least 30 minutes 5 days a week is important to:

- Help you control your weight when combined with healthy eating.
- Reduce your risk of developing diabetes.
- Encourage healthy blood-flow and reduce your blood pressure.
- Give you a sense of wellbeing and help reduce stress.
- Help you deal with your cravings if you are trying to quit smoking.
- Strengthen muscles and bones and reduce osteoporosis (brittle bones).

Over 20% of heart disease and 10% of stroke is due to physical inactivity.

How much physical activity should I do?

If you have a heart condition, it is important to discuss with your doctor the type and intensity of physical activity that is suitable for you. Always check with your doctor before taking up a new type of physical activity. Aim to be active for at least 30 minutes, 5 days a week. If you want to lose weight, over time gradually aim to be active at a moderate intensity for at least 60 to 75 minutes each day.

You don't have to be sporty, join a gym or spend lots of money to be active. Try and be more active at home and at work each day. You can also spread your activity over 2-3 shorter sessions in a day, provided any session is for at least 10 minutes.

- If you want to increase the pace or the amount of physical activity you do, it is important to do so gradually.
- Always warm up before and cool down after activity by walking or jogging at a gentle pace for 5-7 minutes. Loosen up your joints and muscles by stretching. See our booklet, *Be active*, for more information.
- In cold weather, wear a cap or hat to stop you loosing heat through your head.
- On hot days, try to do more of your activities in the cool of the morning or evening.
- Wear comfortable, well-fitting shoes for walking.
- Stop any activity you are doing if you feel very tired or find it hard to breath.
- Know your limits and contact your doctor or hospital if you feel unwell, particularly if you get chest pain.

At home:

- You will be surprised how a brisk walk to the shops and some gardening can add up to 30 minutes of activity in your day.
- Walk the dog or take the family for a walk.
- Play active games with your children, such as hide and seek.
- Leave the car behind and walk or cycle to the bank.
- Do some stretching exercises during the television breaks.

At work:

- Cycle or walk at least some of the way to work.
- Walk during your lunch break.
- Stretch at your desk.
- Walk to talk instead of emailing or phoning.
- Climb the stairs instead of taking the lift.

At leisure:

- Get active with a friend.
- Start a new hobby that involves activity, such as dancing.
- Check out sports clubs and exercise classes.
- · Look out for your local Slí na Sláinte* walking routes.
- Try and include some vigorous intensity activities once or twice a week.



* Slí na Sláinte, meaning path to health is an Irish Heart Foundation initiative. Find a local walking route near you on www.irishheart.ie

Medicines for FH

As well as having a heart-healthy lifestyle, most people with FH will be treated with medicine to lower their cholesterol level. There are several different types of medicines available.

Before taking any new tablets, make a list of all medicines you are already taking and tell your doctor. You may need to change the dose or the time of day you take some tablets so that your current medicines and the newly prescribed cholesterol-lowering drug all work well together. See our book, *Step by step through heart medicines* for more information on cholesterol lowering medicines.

Statins

Statins are the most commonly prescribed type of cholesterol lowering medicine. They slow down the production of cholesterol in your liver. Because your cholesterol will only be lowered as long as you take the medicine, if you have FH, you will probably have to take this type of medicine for life.

Side effects

Statins are well tested and very effective. Side effects are rare and usually mild and include headaches, indigestion and allergies. Rarely, people can have liver problems and muscle inflammation. Because of this you will have a blood test to check your liver function shortly after you start taking the medicine and then once yearly. You should tell your doctor immediately if your urine is dark (brown) or if you have any unexpected muscle pains, tenderness or weakness – particularly if you've started a new medicine or changed the dose recently.

Statins are not given to children before puberty. Women who are pregnant, trying to become pregnant or breast-feeding should not take statins.

Other cholesterol-lowering drugs

Bile acid binding resins

Your liver makes bile acids using cholesterol. Resins stop bile acids from being re-absorbed into your blood from your intestine. This means that when your liver needs to make more bile acids it does so by taking LDL (bad) cholesterol from your blood and thus reducing your LDL (bad) cholesterol level.

Resins usually come in powder form. You take them by mixing them with water, fruit juice, or yoghurt. Some resins, such as cholestyramine (Questran®), are not absorbed into the body and are safe for children and pregnant women. Often people taking resins will need to take extra vitamin A, D, E, K and folic acid. This type of medicine can cause flatulence and constipation but this can be reduced by building up the dose gradually.

Fibrates

Fibrates are good if you have high triglyceride levels as well as LDL (bad) cholesterol and low HDL (good) cholesterol. If you are prescribed a fibrate as well as a statin it is very important to have a regular blood test, to check your liver function and the level of an enzyme in your blood called creatine phosphokinase (CPK). Your GP can arrange these tests for you. A review by your cardiologist at regular intervals is also important when you are taking these medicines. These drugs can occasionally cause stomach upsets.

Nicotinic acid

Nicotinic acid lowers both cholesterol and triglyceride levels. It also increases the amount of HDL (good) cholesterol in your body. A common side effect is flushing, although taking your tablet in the evening time (after a low-fat snack) and taking your aspirin 30 minutes before the nicotinic acid medicine can reduce this problem. Other side effects include indigestion, itching and skin rashes. Nicotinic acid medicines can also lower your blood pressure.

Ezetemibe

Ezetemibe stops your intestine from absorbing cholesterol. While it can be prescribed on its own, it is best used in combination with a statin, if the statin alone is not reducing your cholesterol level sufficiently.

Other treatments for FH

LDL apheresis

LDL apheresis filters your blood and removes the cholesterol. Like kidney dialysis, this procedure is carried out in a hospital or specialist centre. Your blood passes through a special machine that removes the cholesterol before it goes back into your body. Each treatment takes about four hours to complete and you will need the procedure repeated at regular intervals, usually 2-4 times per month. This procedure is not currently available in Ireland and there are only a few specialist centres in the UK offering the treatment.

Your cholesterol chart

Every time you have your cholesterol checked, fill in your cholesterol levels on the chart below.

Date	Total cholesterol	LDL (bad) cholesterol	HDL (good) cholesterol	Triglycerides

An explanation of medical terms used in this booklet

Apheresis

LDL apheresis is a filtering technique to remove cholesterol from your blood.

Bile acids help digestion and the absorption of fats in your small intestine.

Bile acid binding resin

This is a medicine used to lower cholesterol levels. Unlike other cholesterol lowering drugs, some resins are safe for children and pregnant women.

Cholesterol

Cholesterol is a soft waxy material that forms part of each cell in your body. You need a certain amount of cholesterol for your body cells and to produce important hormones. Your liver makes most of the cholesterol in your body.

Creatine phosphokinase

This is an enzyme found mainly in your heart, brain and skeletal muscle. Some medicines can raise the level of creatine phosphokinase in your blood. High levels of this enzyme can also be a sign of a heart attack or stroke. Thus it is important to have a regular blood test to monitor your creatine phosphokinase level.

Familial hypercholesterolaemia (FH)

FH is an inherited condition where you have a very high level of cholesterol in your blood.

Gene

A gene is a section of DNA that allows 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 in your body.

Heart attack

A heart attack is when a coronary artery that supplies blood to your heart muscle becomes blocked and the heart muscle dies or is permanently damaged. Medically, a heart attack is called a myocardial infarction (MI).

High density lipoprotein (HDL)

HDL (good cholesterol) is a type of cholesterol that removes excess bad cholesterol from your blood and protects against heart disease.

Low density lipoprotein (LDL)

LDL (bad cholesterol) is a type of cholesterol that, if in excess, sticks to the walls of your arteries, making them narrow and causing coronary artery disease.

LDL receptor

An LDL receptor is the socket on each of your cells for LDL cholesterol to plug into.

Lipoprotein (a) or LP(a)

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

Millimole per litre or mmol/l

This is the unit of measurement of chemicals including blood cholesterol.

Monounsaturated fat

This type of fat can lower your LDL (bad) cholesterol and thus reduce your risk of heart disease and stroke.

Nicotinic acid

This is a medication used to lower cholesterol levels. Nicotinic acid lowers both cholesterol and triglyceride levels. It also increases the amount of HDL (good) cholesterol in your body.

Omega 3 fats

These fats help lower your triglyceride level, help prevent blood clots and help your heart to keep a healthy rhythm.

Omega 6 fats

Omega 6 fats can help lower your LDL (bad) cholesterol.

Polyunsaturated fats

Two main types of polyunsaturated fats are omega 3 fats and omega 6 fats.

Saturated fat

This is the type of fat from which most of the cholesterol in your body is made. The term saturated refers to the chemical structure of the fat. Saturated fats raise your LDL (bad) cholesterol and increase your chances of getting heart disease or having a stroke.

Statin

Statins are medicines used to lower cholesterol levels. They slow down the production of cholesterol in your liver.

Triglycerides (TG)

TGs are a type of fat in your blood. Too much TG increases your chance of developing heart disease or having a stroke.

Xanthelasma

This is a yellow patch of skin near your eyes that shows you have high blood cholesterol levels.

Xanthoma

This is a yellow lump on your elbows, knuckles and tendons that show you have high blood cholesterol levels.

More information

Useful websites: www.irishheart.ie www.stroke.ie www.hse.ie www.bhf.org.uk www.heart.org

Other Irish Heart Foundation publications:

Step by step through stroke, a guide for those affected by stroke and their carers Step by step through heart surgery Step by step through heart medicines Step by step through angina Step by step through heart failure Step by step through cardiac catheterization AF and you, information for people living with atrial fibrillation Step by step through heart attack Manage your stress All about your heart and stroke Time to cut down on salt Manage your blood pressure A healthy cholesterol Healthy eating Be active Quit smoking Lose weight

Heart and Stroke Helpline:

Locall 1890 432 787 Monday to Friday 10am to 5pm www.irishheart.ie

Please make a donation today



The Irish Heart Foundation is Ireland's national charity dedicated to the reduction of death and disability from heart disease and stroke. Over 90% of our funding comes from public and business donations. We depend on your goodwill and generosity to continue our work.

If you found this booklet useful, please help our charity to continue to provide heart & stroke information by donating today.

You can make your donation today:

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By signing this mandate form, you authorise (A) the Irish Heart Foundation to send instructions to your bank to debit your account and (B) your bank to debit your account in accordance with the instruction from the Irish Heart Foundation.

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Your first contribution will be taken on either the 2nd or the 20th of the next available month. Please select which date you prefer. 2nd 20th You will be notified in writing ten days in advance of your first direct debit. If you wish to cancel within 7 days of a direct debit payment please contact your own bank.
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The Irish Heart Foundation is the national charity fighting heart disease and stroke.



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Heart and Stroke Helpline:

Locall 1890 432 787 Monday to Friday 10am to 5pm

Web:

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Registered Charity Number CHY 5507





The information provided in this booklet was correct and accurate at the time of publication to the best of the Irish Heart Foundation's knowledge.