Sláinte Leanaí Éireann
Children's Health Ireland

Metabolic.ie

National Centre for Inherited Metabolic Disorders

Managing Familial Hypercholesterolaemia

Contents

Disclaimer: Where specific brands are shown or mentioned these are for illustrative purposes only. We do not endorse any particular products.

The information in this booklet is correct at the time of writing and the recommendations made are based on the most up to date scientific evidence at the time of printing so we recommend you use this only under full dietary and medical supervision.

Acknowledgements

This booklet has been adapted based on information from:

- The Irish Heart Foundation https://irishheart.ie/
- Heart UK-The Cholesterol Charity https://www.heartuk.org.uk/

Photos on page 4:

- Xanthelasmas: By Klaus D. Peter, Wiehl, Germany - Own work, CC BY 3.0 DE
- Xanthomas: By Min.Neel—Own work CC licence BY-SA 3.0 DEED

What is Familial Hypercholesterolemia?

Familial hypercholesterolemia (FH) is an inherited disorder which causes very high cholesterol levels. It causes very high blood levels of low-density lipoprotein (LDL) cholesterol or "bad cholesterol". LDL cholesterol is elevated from birth.

Lipoproteins are particles that carry cholesterol through your bloodstream to your cells.

In the early years, there are often no signs or symptoms of FH. However, if left untreated, having too much LDL cholesterol in your blood can lead to damage as it sticks to the walls of blood vessels. This can cause hardening and narrowing of the blood vessels (see figure 1), which restricts blood flow to the heart and other organs. This can increase the risk of heart attack and stroke at an early age.

With extremely high levels of LDL cholesterol, some people may experience:

- Cholesterol deposits in the eyelids (xanthelasmas)
- Fatty skin deposits called xanthomas over parts of the hands, elbows, knees, ankles and around the cornea of the eye
- Chest pain (angina)
- Heart attacks and strokes

By finding FH early and treating it appropriately, the risk of heart disease can be reduced.

Cholesterol build up in the arteries in untreated Familial Hypercholesterolemia

What is Cholesterol?

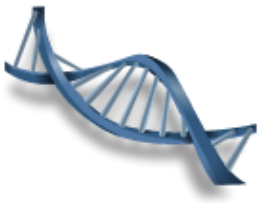
Cholesterol is a type of fat that forms part of each cell in our body. We need a certain amount of cholesterol for our body cells to work properly and to produce hormones needed by the body. Cholesterol is carried around the body inside small round parcels of fat and protein called lipoproteins. However, too much cholesterol can be unhealthy. It can build up in the blood and can stick to the walls of your blood vessels. This is bad for your heart. Once any cholesterol has lined your blood vessels, you cannot remove it, but you can stop it from getting bigger.

Types of cholesterol

A simple blood test can check the amount of different types of cholesterol in your blood at a given time. This test will measure the total cholesterol and also different types of cholesterol; cholesterol in low density lipoproteins (LDL cholesterol) and cholesterol in high density lipoprotein (HDL cholesterol). **LDL cholesterol** is often referred to as "**bad cholesterol"** as LDL carries cholesterol to your blood vessels. **HDL cholesterol** is referred to as **"good cholesterol",** as HDL mops up excess cholesterol from your blood and brings it back to the liver to be broken down and removed from the body.

Aim for **less than 3 mmol/l of LDL** cholesterol. Your consultant may advise on lower depending on your risk

What causes Famililal Hypercholesterolaemia?



FH is caused by changes in one of three genes. Genes are the DNA instructions that our body uses to tell every cell how things should function.

There are genes involved in telling the body to produce lipoproteins that are involved in clearing LDL cholesterol – the harmful type of cholesterol – from the bloodstream. In FH, one of these is faulty and your body does not get the correct instructions to clear the cholesterol.

If someone carries a faulty gene – and therefore has FH – then there is a 50:50 chance that they will pass it on to each of their children.

How is Famililal Hypercholesterolaemia detected?

Your doctor may suspect you have FH if:

- Your cholesterol levels are very high during routine blood tests
- Other family members have been diagnosed with FH
- There is a history of early heart attacks or stroke among close family members

Treatment


Diet

+


Lifestyle

+


Medication

A healthy diet and lifestyle is important to lower your blood LDL cholesterol. However, for some people with FH, a healthy diet and lifestyle alone may not be enough to lower blood LDL cholesterol to an acceptable level. Medications may be needed to help lower LDL cholesterol further.

Diet and Lifestyle

Tips for Lowering Cholesterol:

1. Choose the right balance of fat

Dietary fats fall into two main groups: saturated fats and unsaturated fats. It can be confusing when choosing which fats to include in your diet. The tables below will help explain how different types of fat affect your cholesterol.

Reduce intake of saturated fat (animal fats and trans fat)

Saturated fats are mainly found in meat and full-fat dairy products. They are usually solid at room temperature. We all need a little saturated fat but eating too many foods high in saturated fat can increase LDL cholesterol levels.

Trans fats are fats generally found in processed foods. Trans fats can raise your LDL cholesterol levels and lower your HDL cholesterol.

It is recommended to limit consumption of these foods. The table on page 7 lists common foods that contain saturated and trans fats.

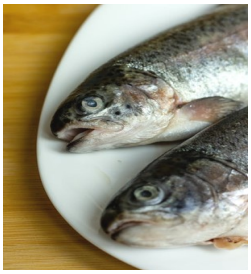
Foods to avoid	
Full-fat dairy products such as cream, milk, yogurt, crème fraiche and hard cheese	  
Butter, ghee, lard, dripping, margarine, goose fat and suet	 
Fatty and processed meat such as sausages, burgers, bacon, kebabs, lamb chops with fat and skin. Deep fried foods and takeaways	 
Biscuits, cakes, pastries, crackers and pies, milk and white chocolate, toffee	  
Coconut and palm oils, stearic acid	 

Red meat provides lots of benefits to our health such as being a good source of iron. Choose extra lean types and trim off visible fat

Replace with unsaturated fats (vegetable fats and oily fish)

Unsaturated fats are found in plant foods and oily fish. They are usually liquid at room temperature. Unsaturated fats are more heart-healthy and can improve blood cholesterol levels. They are different types of unsaturated fat known as monounsaturated, polyunsaturated and omega 3 fats and they all do different jobs in the body. It is good to eat a range of foods so that you get mixture in your diet.

We should eat fewer foods high in saturated fat and focus on consuming, in moderation, foods with heart healthy unsaturated fats. The table on page 8 shows examples of how to replace foods that are high in saturated fats with foods that are either low in fat or contain unsaturated fats.

Eat less….	Replace with…
• Butter, gee, lard, suet, hard margarines, coconut oil and palm oil.	• Vegetable spreads and oils such as olive, sunflower, rapeseed and soya.  
• Fatty meat and processed meat products (sausages, fatty bacon, salamis, canned meat). • Have red meat less often and in smaller portions.	• Remove all visible fat from meat and the skin from poultry. • Have fish twice a week (one oil-rich). • Replace half or all meat in a recipe with beans, peas, lentils or vegetables. • Have meat-free days: use beans, pulses or soya mince/chunks.  
• Full fat milk and yogurts. • Canned coconut milk and coconut alternatives to yogurt.	• Skimmed, 1% fat or semi-skimmed dairy milk. • Unsweetened/low sugars plat-based drinks: soya, almond, rice or oat. • Low fat/fat-free yogurts or soya alternatives.   
• Most cheeses. • Dairy creams. • Coconut alternatives to cream and dairy -free cheeses based on coconut fat/oil.	• Lower fat cheeses e.g. cottage cheese, Quark or small portions of ‘lighter’/’reduced fat’ cheeses. • Soya single alternatives to cream.  

Eat less….	Replace with…
• Cakes and desserts • Cookies and biscuits with chocolate, coconut • Sweet pastries and doughnuts • Chocolate • Sweets • Coconut	• Fruit-based desserts e.g. stewed fruit, fruit crumbles (with vegetable spread). • Sugar-free jellies with fruit, fruit salad. • Plain biscuits e.g. rich tea, digestives • Fruit and low fat/fat-free yogurt or soya alternative to yogurt.  
• Sausage rolls and pies. • Cream or coconut-based curries • Cheese and cream-based pasta dishes. • Extra cheese or meat topped pizzas. • Sandwiches with cheese, processed meat and /or mayo fillings.	• Potato topped pies. • Tomato and vegetable-based curries and pasta dishes. • Thin crusted pizzas with vegetable toppings. • Sandwiches with wholemeal bread, lots of salad and fillings such as hummus, lean chicken, egg and falafel.  
• Roasting or frying with butter, lard, other animal fats or coconut oil.	• Small quantities of vegetable oil or other cooking methods e.g. boiling, grilling, steaming.  

Remember!

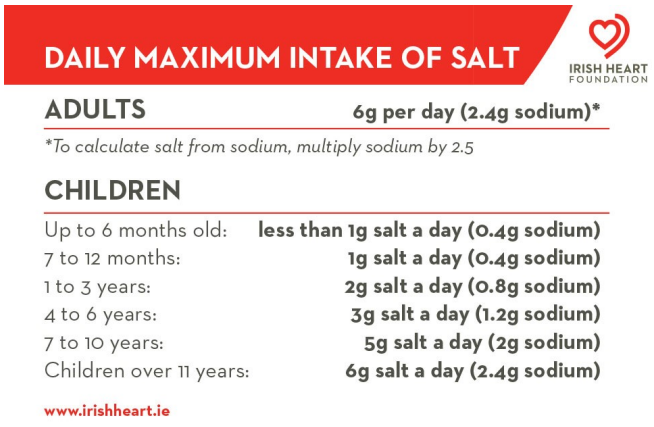
All fats have a similar calorie value and too much of them can contribute to weight gain therefore it is important to use all fats sparingly no matter how heart healthy they are.

Reading Food Labels

Reading food labels is an important part of healthy eating. This card is designed to help you understand the information on food labels more easily using the traffic light colour coding system.

When comparing products it is best to look at the ingredients per 100g this way products can be easily compared. There can be big differences in fat content between similar products so it is worth checking the label to help you find the healthiest product.

You are able to order a copy of these food shopping cards for your wallet from the Irish heart



foundation : www.irishheart.ie

The main thing to check for on food labels are:

1. Type of fat
2. Amount of fat
3. Amount of saturated fat
4. If it contains any trans fats

High saturated fat is more than 5g of saturated fat per 100g of the food

Medium saturated fat is 1.5-5g of saturated fat per 100g of the food

Low saturated fat is less than 1.5g of saturated fat per 100g of the food

2. . Cholesterol lowering foods

Choose wholegrain and high fibre foods

Fibre has been shown to reduce the absorption of cholesterol from the intestine into the body

How to increase fibre?

- Choose **wholegrains** sources of cereal, bread, pasta and rice
- Increase consumption of **fruit and vegetables**

Fruit and vegetables especially skin of fruit and vegetables are a good source of fibre. Eating **5 -7 portions** of fruit and vegetables* a day can help reduce your risk of heart disease. These foods can come from fresh, frozen or tinned sources.

What is one portion of fruit or vegetables?

1 medium piece of fruit (orange/apple/banana)

2 small pieces of fruit (kiwi/plums/mandarin)

6 Strawberries

10 grapes

150ml unsweetened juice *

16 raspberries

½ cup of cooked vegetable (fresh or frozen)

1 bowl homemade soup (220ml)

1 bowl salad (lettuce/cucumber/tomato)

*Fruit juice and smoothies should only be counted as one portion of fruit per day.

How to include wholegrains

Oats in particular are an excellent food choice as they are high in fibre and low in fat. They also contain a special form of soluble fibre called beta glucans. Beta glucans, when consumed as part of healthy diet have been shown to lower LDL cholesterol. Good sources include Oatibix, Porridge, Readybrek as well as porridge bread, homemade granola, oatcakes and oatballs.

Try adding beans, pulses, pearl barley, lentils or chick peas into your salads, casseroles, soups and pasta sauces. Hummus is another good option.

It is important, in order to avoid stomach upset, that you slowly increase the amount of fibre in your diet and also to increase the amount of water you drink.

3. Increase intake of fish

Current guidelines recommend consuming two portions of fish per week as part of a healthy diet, one of which should be oily. Examples of oily fish include tuna (not tinned), sardines, herring, mackerel, trout and salmon. You can choose from fresh, canned or frozen varieties. However, try to avoid crumbed, battered or deep fried fish.

Oily fish is particularly recommended in relation to heart health as it contains essential fatty acids known as omega 3 fatty acids. Omega 3 fatty acids which are thought to:

- Increase HDL cholesterol
- Reduce LDL cholesterol

4. Nuts and seeds

All nuts are a good source of vegetable protein, fibre heart healthy monounsaturated fats and vitamin E. Seeds can also help to reduce cholesterol levels.

Try adding these to your diet:

Almonds 	Brazil nuts 	Pistachios 	Walnuts 
Pecans 	Cashews 	Peanuts 	Hazlenuts 

How to include them in your diet

- Eat as a snack between meals
- Sprinkled on cereal, yoghurt or salad
- Mix through stews, curries or pasta sauce
- Use almonds in cooking/baking
- Try almond or peanut butter in sandwiches, on rice cakes, crackers or slices of apple

Whole nuts should not be given to children under 5 years because of the risk of choking. Unsweetened and unsalted nut butters or chopped nuts can be an excellent way to include them. Choose natural peanut and other nut butters that do NOT contain palm oil or partially hydrogenated oils.

5. Maintain a healthy weight

Your dietitian will evaluate your child's weight. As your child is still growing, there is no ideal weight, instead we use centile charts to interpret measurements.

Extra weight around your child's waist is a concern as it can increase cholesterol and the risk of heart disease.

6. Plant stanols and sterols

Plant stanols and sterols are natural substances found in some foods such as oats, barley, almonds, soy bean oil, sesame seeds and some fruit and vegetables. Plant stanols and sterols work by **blocking the amount of cholesterol your body absorbs from the food** you eat. Foods fortified with plant stanols and sterols may help lower your LDL cholesterol.

Stanols and Sterols are not usually recommended when taking *Eztemibe*. This is because they both work in a similar way and the plant stanols and sterols are unlikely to have any extra effect.

The recommended amount of plant stanols amd sterols is **2.5-3g** per day to help lower cholesterol. It is usually achieved by consuming **two or three portions** of a fortified food per day.

- 2 teaspoons or 10g spread (0.5-0.7g depending on brand)
- 1 yoghurt drink (2g)
- 1 yogurt (2g)

Some brands include Benecol and Flora ProActiv but many supermarkets have their own brands.

Products containing plant stanols and sterols are **not recommended for children under the age of 5 years**, breastfeeding or pregnant women.

7. Include soy protein

Soy protein is a good source of vegetable protein, is low in saturated fat and high in fibre. Soy has been shown to have beneficial effects on cholesterol levels and cardiovascular health. Consumption of soy-based foods in place of foods high in saturated fat such as fatty or processed meats can help to lower LDL and total cholesterol.

Sources include:

- Unsweetened soya milk – ensure they are fortified with Calcium, Vit D and Iodine if using in place of cow's milk
- Unsweetened soya yoghurts
- Unsweetened soya desserts

In cooking:

- Use soya beans/mince to replace some of the meat in your dishes
- Add tofu and/or soya beans to stir fries, casseroles, salads and on pasta

8. Be active

Children under 5 should have at least three hours of physical activity each day. By 5 years of age, this should include at least 1 hour of energetic play a day, where they sweat and their breathing is faster than normal.

The activity does not need to be done all at once. It can be spread out over the course of the day. Reducing the amount of time your child is sitting or not moving is also important.

All activity counts as energetic activity:

- Walking, running, dancing, hopping, jumping, skipping or cycling
- Active games involving running, chasing, throwing and catching
- Team sports for older children including gaelic football, hurling, basketball or tennis

Medications

The following medications may be recommended for FH:

1. Statins

Statins are the most common medication used to help lower your LDL cholesterol. Statins have been around for a long time and they often work very well. They can reduce LDL cholesterol by 20-40%. In FH, statin therapy may be recommended in childhood, often starting by age 8-10 years.

How do statins work?

Statins work by reducing the amount of LDL cholesterol your liver can make. Because your liver is not making so much cholesterol, it draws cholesterol from your blood so your blood cholesterol levels go down.

2. Ezetimibe

Ezetimibe is a medicine that blocks cholesterol being absorbed from the food you eat. In some cases, where a statin does not lower LDL cholesterol enough ezetimibe may be offered alongside it.

3. PCSK9 inhibitors

PCSK9 inhibitor is a medicine that is only given if statins and other medicines fail to effectively lower your cholesterol. PCSK9 inhibitors help the liver absorb more LDL cholesterol, which lowers the amount of cholesterol circulating in the blood. They are injected under the skin every few weeks.

4. Other treatments

In severe cases, certain people with FH may undergo a procedure that filters the excess cholesterol from their blood.

Some Commonly Asked Questions

Can I still eat eggs?

- Eggs contain cholesterol but they have *little effect on your blood cholesterol*
- Recent evidence shows that cholesterol found in food has only a small impact on blood cholesterol when compared with the effect of saturated fat intake
- Recent research confirm that **an egg a day is ok**
- There is no cholesterol in the egg whites. The egg yolk does contain cholesterol but also nutrients including vitamin A, B12. D, iron and folate
- The best way to cook an egg is either poached or boiled. What accompanies the egg often has a greater effect on cholesterol rather than the egg itself. For example fried eggs, hollandaise sauce and butter

Cholesterol is made by our liver but some foods contain cholesterol such as eggs, shellfish and liver. Evidence shows that cholesterol found in food has little or no effect on blood cholesterol. These foods can still be eaten in moderation as part as a healthy diet.

Should I include coconut oil in my diet?

Coconut oil is currently popular in the 'Paleo' and 'clean eating' diet trends. It claims to have a number of health benefits and is popularly called a 'superfood'.

Coconut oil is very high in calories and is 85% saturated fat.

It raises LDL cholesterol as well as total cholesterol in contrast to unsaturated fats which lower LDL cholesterol.

Based on the latest scientific evidence, **we do not recommended coconut oil over unsaturated fatty acids such as olive and rapeseed oils,** along with the other vegetable oils listed on page 8.

Useful Tips for Eating Out

- Takeaways are generally extremely high in fat and salt and should only be eaten occasionally or for special occasions

- Ask for vegetables/salads to be served without sauces or butter or for sauces to be served on the side

- Check menus online - this makes it easier to decide what your best options are in advance. Some restaurants have ‘heart-healthy’ or ‘low fat’ symbols on their menus or nutritional calculators on their websites.

- Ask for dishes to be adapted – it is not usually a problem for a restaurant to replace chips with a jacket potato, salad or vegetables

- Choose dishes that in the description include words like grilled, baked, broiled, poached, boiled, steamed, lightly sauteed, or stir-fried

- Avoid foods described as fried, deep-fried, breaded (fried), creamed, au gratin, buttered, pan fried, crispy, scalloped and in its own gravy

- Choose dishes that are based on a tomato sauce rather than cream based sauces

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