

Understanding NICE guidance

Information for people who use NHS services

Inherited high cholesterol in the family (familial hypercholesterolaemia)

NICE 'clinical guidelines' advise the NHS on caring for people with specific conditions or diseases and the treatments they should receive.

This booklet is about the care and treatment of people with a specific type of inherited high cholesterol in the family, known as familial hypercholesterolaemia, in the NHS in England and Wales. It explains guidance (advice) from NICE (the National Institute for Health and Clinical Excellence). It is written for people with familial hypercholesterolaemia but it may also be useful for their families or carers or for anyone with an interest in the condition.

The booklet aims to help you understand the care and treatment options that should be available in the NHS. It does not describe familial hypercholesterolaemia or the tests or treatments for it in detail. A member of your healthcare team should discuss these with you. There are examples of questions you could ask throughout this booklet to help you with this. Some sources of further information and support are on page 8.



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The advice in the NICE guideline covers the care and treatment of adults and children/young people with familial hypercholesterolaemia (a specific type of inherited high cholesterol that runs in the family).

It does not cover other forms of hypercholesterolaemia that are not genetic (inherited) or that are due to other genetic conditions.

Your care

Your treatment and care should take into account your personal needs and preferences, and you have the right to be fully informed and to make decisions in partnership with your healthcare team. To help with this, your healthcare team should give you information you can understand and that is relevant to your circumstances. All healthcare professionals should treat you with respect, sensitivity and understanding and explain familial hypercholesterolaemia and the treatments for it simply and clearly.

The information you get from your healthcare team should include details of the possible benefits and risks of particular treatments. You can ask any questions you want to and can always change your mind as your treatment progresses or your condition or circumstances change. Your own preference for a particular treatment is important and your healthcare team should support your choice of treatment wherever possible, but this does depend on your age. If you are over 16, or under 16 and fully understand the treatment, you may be able to give your own agreement. If you are too young, your parents or carers may need to agree to your treatment.

Your treatment and care, and the information you are given about it, should take account of any religious, ethnic or cultural needs you may have. It should also take into account any additional factors, such as physical or learning disabilities, sight or hearing problems, or difficulties with reading or speaking English. Your healthcare team should be able to arrange an interpreter or an advocate (someone who supports you in putting across your views) if needed.

If you agree, your family and carers should have the chance to be involved in decisions about your care. Family members and carers also have the right to the information and support they need in their roles as carers.

If people are unable to understand a particular issue or are not able to make decisions for themselves, healthcare professionals should follow the advice that the Department of Health has produced about this. You can find this by going to the Department of Health website (www.dh.gov.uk/consent). Your healthcare professional should also follow the code of practice for the Mental Capacity Act. For more information about this, visit www.publicguardian.gov.uk

If you think that your care does not match what is described in this booklet, please talk to a member of your healthcare team.

What is familial hypercholesterolaemia?

Cholesterol is a type of fat made by the body. Sometimes a high level of cholesterol in the blood, also known as 'hypercholesterolaemia', runs in families. Some families have a particular type of inherited high cholesterol caused by raised blood levels of 'low density' cholesterol, often known as 'bad cholesterol'. This is called 'familial hypercholesterolaemia' (or FH for short). It is not caused by an unhealthy lifestyle, but is passed from generation to generation through a 'faulty gene'. This means that brothers and sisters or children of someone with FH have a one in two (50%) chance of having the condition.

In people with FH, the raised levels of 'bad cholesterol' lead to 'hardening' of the arteries and an increased risk of heart disease, if left untreated. Treatment reduces the risk of heart disease and can help to ensure that people with FH have a normal life expectancy.

Diagnosis

What your healthcare professional should do

If your healthcare professional thinks that you might have FH, they should ask about your family history, including the age at which any of your relatives developed heart disease. If any of your relatives have died, your healthcare professional should ask about the age that they died, what caused their death, and whether or not they smoked. It may be useful to check this information with other family members. Your healthcare professional should measure your cholesterol levels and examine the tendons in your hands and ankles and other joints to look for cholesterol deposits to help them make their diagnosis.

If, after tests, your healthcare professional still thinks that you have FH, you should be offered a referral to see a specialist to confirm the diagnosis. You should be offered a DNA test, which will check for the presence of the faulty gene that may be causing your condition and will help confirm the diagnosis.

Children and young people should be offered a referral to a specialist in a setting suitable for children or families.

Your healthcare professional should provide you with information about FH, how it affects your risk of heart disease, and how it might affect other members of your family. They should provide you with information about what type of testing is involved and offer you advice on your lifestyle and the treatment options that are available.

Referral to a specialist

After you have been diagnosed with FH, your healthcare professional may think that you need to see a further specialist, particularly if you appear to have any symptoms of heart disease or your treatment is not lowering your cholesterol levels enough. Your healthcare professional should discuss this with you.

Questions you might like to ask your healthcare team

- Please tell me more about familial hypercholesterolaemia (or FH).
- What are the chances my children are going to be affected?
- How does this affect my family?
- Can you provide any information for my family?
- Please tell me more about testing other members of my family.
- Does FH affect a woman's chance of having children?
- Are there any support organisations I can contact?

Testing of family members

Because FH is an inherited condition, it is important to identify any family members (specifically blood relatives and not those people related to you by marriage) who may also be affected so that they can be offered treatment. Your healthcare professional should discuss with you testing members of your family for the condition.

Your healthcare professional should offer to talk with members of your family about what FH is and what impact the condition may have on them.

If DNA testing finds the faulty gene that is responsible for your condition, your healthcare professional should offer DNA testing to other members of your family to look for the same faulty gene. If the faulty gene that is responsible for your condition has not been identified, a healthcare professional should still check the cholesterol levels of your family members.

If possible, by the time they are 10 years old, children of people who have been diagnosed with FH should have their cholesterol levels tested, and be offered DNA testing if a faulty gene has been identified in one of their parents. Children under 5 should, when possible, have their cholesterol levels tested if they have yellow patches of cholesterol deposits on their skin or if both their parents have been diagnosed with FH.

Treatments for FH

Treatments for FH are lifelong. Treatment, which will include both drugs and changes to your lifestyle, lowers your cholesterol levels and helps reduce the risk of heart disease in the future. Your healthcare professional should discuss with you all of the treatment options that are available to you.

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| Adults | <p>You should be offered a drug called a statin.</p> <p>If statins are not suitable for you after discussion with your doctor, you may be offered a drug called ezetimibe.</p> <p>Some people may be offered both a statin and ezetimibe.</p> <p>There are other medicines available if statins or ezetimibe are not appropriate for you.</p> <p>You should be offered a referral to a specialist.</p> |
| Women and girls | <p>Your doctor should discuss your contraceptive options with you and advise you of the risks and benefits of these options.</p> <p>Women with FH usually have a normal pregnancy. You should talk to your doctor if you are planning to have a baby or if you are pregnant. Your doctor should advise you to stop your cholesterol-lowering medicines and should explain why this is important.</p> <p>If you are planning to have a baby or if you are pregnant, you should be offered an appointment with a heart specialist (a cardiologist) and a specialist in pregnancy (an obstetrician).</p> <p>Women with FH can breastfeed if they choose to do so. Your doctor should discuss the potential risks and benefits of restarting cholesterol-lowering medication while breastfeeding with you.</p> |
| Children and young people | <p>You may be offered a statin, or another treatment if statins are not appropriate. Treatment options will depend on your age, cholesterol levels and the age that family members developed heart disease.</p> <p>You should be offered a referral to a specialist.</p> |

Questions about the treatment

- Why have you decided to offer me this particular type of medicine?
- What are the risks or side effects associated with this medicine?
- Can I take my other medicines at the same time as this treatment?

Lifestyle advice

There are a number of changes to your lifestyle that you can make to help reduce your risk of heart disease if you have FH. Your healthcare professional should discuss these with you.

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| Smoking | Smoking greatly increases the risk of having a heart attack. If you smoke, your healthcare professional should advise you to stop and offer you the support you need to help you to stop. There are support services and treatments available on the NHS for people who are trying to give up smoking. |
| Diet | <p>You should follow a healthy diet and eat at least five portions of fruit and vegetables a day, in line with national guidance. If you, or a member of your family, has FH then you should receive individualised advice on your diet from a healthcare professional who has expertise in this area. You should be advised to:</p> <ul style="list-style-type: none"> • avoid eating foods that contain a lot of fat (particularly saturated fats and cholesterol) • eat at least two portions of fish a week (one of which should be oily fish), but limit oily fish to two portions a week if you are pregnant. <p>Further information on diet and healthy eating can be found at www.eatwell.gov.uk/healthydiet and www.5aday.nhs.uk</p> |
| Exercise | You should be advised to exercise for 30 minutes a day, at least 5 days a week. You don't have to do all the exercise in one go – you can spread the activity throughout the day. The level of the exercise you do should depend on what you are able to do – your healthcare professional should discuss this with you. Exercise should include things that can easily become part of your daily routine, such as walking, using stairs or cycling. |
| Weight | If you are overweight, then your healthcare professional should give you advice and support about reaching and maintaining your ideal weight. |
| Alcohol | If you drink alcohol, you should stay within safe limits and not binge drink. Men should drink no more than 3–4 units of alcohol a day, and women should drink no more than 2–3 units of alcohol a day. |

If a treatment described in this booklet appears suitable for you, but it is not available, you should talk to your local Patient Advice and Liaison Service (PALS) in the first instance. If they are not able to help you, they should refer you to your local Independent Complaints Advocacy Service.

Apheresis

A treatment known as 'apheresis' may be offered to a small number of people with FH who are severely affected. Apheresis involves filtering the blood and removing cholesterol. This is only offered to those people who carry a 'double dose' of a faulty gene (where the person has inherited a faulty gene from both of their parents), or to those with a 'single dose' (where the person has inherited a faulty gene from one of their parents) who have severe heart disease which is not responding to any other treatment.

If you are being offered treatment with apheresis, then a specialist should discuss with you the risks and benefits of the procedure.

Follow-up

You should have appointments with your healthcare professional at least once a year to review your condition and have a blood test taken to make sure that your treatments are still right for you. Your healthcare professional should ask for any updates on your family history and about progress in the testing of your family members.

More information

The organisations below can provide more information and support for people with familial hypercholesterolaemia. Please note that NICE is not responsible for the quality or accuracy of any information or advice provided by these organisations.

- British Heart Foundation, 0845 070 8070
www.bhf.org.uk
- CLIMB – Children Living with Inherited Metabolic Disorders, 0800 652 3181, www.climb.org.uk
- H-E-A-R-T UK – The Cholesterol Charity, 0845 450 5988
www.heartuk.org.uk

NHS Direct online (www.nhsdirect.nhs.uk) may also be a good starting point for finding out more. Your local Patient Advice and Liaison Service (PALS) may also be able to give you further information and support.

About NICE

NICE produces guidance (advice) for the NHS about preventing, diagnosing and treating different medical conditions. The guidance is written by independent experts including healthcare professionals and people representing patients and carers. They consider the best available evidence on the condition and treatments, the views of patients and carers and the experiences of doctors, nurses and other healthcare professionals working in the field. Staff working in the NHS are expected to follow this guidance.

To find out more about NICE, its work and how it reaches decisions, see www.nice.org.uk/aboutguidance

This booklet and other versions of this guideline aimed at healthcare professionals are available at www.nice.org.uk/ICG071. The versions for healthcare professionals contain more detailed information on the care and treatment you should be offered.

You can order printed copies of this booklet from NICE publications (phone 0845 003 7783 or email publications@nice.org.uk and quote reference N1641).

We encourage NHS and voluntary sector organisations to use text from this booklet in their own information about familial hypercholesterolaemia.