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Ten top tips - Familial hypercholesterolaemia

Dr Kosta Manis, GP and CCG cardiology lead, offers his tips for managing familial hypercholesterolaemia

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1. Premature CHD resulting from familial hypercholesterolaemia is completely preventable

Patients with FH who are identified and treated have a normal lifespan. When referring to familial hypercholesterolaemia that results from an inherited gene, be sure to use the term ‘gene alteration’ when addressing your patients.

2. It’s more common than you might think

A practice with a list size of 5,000 patients is expected to have about 10 patients with heterozygous familial hypercholesterolaemia (HeFH). These patients are more prone than standard hyperlipidaemia patients to have a cardiovascular incident and a third do not survive their first heart attack. The elevated serum cholesterol concentration that characterises HeFH leads to a greater than 50% risk of coronary heart disease in men by the age of 50 years and at least 30% in women by the age of 60 years.

3. Xanthomas, xanthelasmata and arcus senilis are usually present – and are more severe - in FH but they are not conclusive

Inheriting the gene for familial hypercholesterolaemia from both parents makes a person genetically homozygous (HoFH). It is a devastating disorder associated with markedly premature and progressive cardiovascular disease, aortic stenosis and early death.

Cholesterol levels exceed 10 mmol/L, with more extensive cutaneous or tendon lipid depositions (xanthomas) than those found in HeFH

Note that xanthomas, xanthelasmata and arcus senilis, although usually present in both types, are not always signs of either. They are usually more extensive and more severe in HoFH, but they are not conclusive. So you can only suspect a diagnosis, relying on baseline investigations and clinical examination.

Patients have vastly increased risk and their management requires specialised centres. Treatment includes maximal statin doses combined with ezetimibe, bile acid sequestrants and niacin. Lipid dialysis (apheresis) and liver transplant is part of the treatment.

4. Most people with familial hypercholesterolaemia have not been diagnosed

Official guidelines to improve screening and treatment for familial hypercholesterolaemia are often ignored in favour of initiating treatment in all hypercholesterolaemias – regardless of aetiology – in an unfortunate rush to hit revenue-raising targets. Although there is a national screening programme in Scotland, Northern Ireland and Wales, there isn't one in England, which means that people are having heart attacks that could have been prevented. It is therefore immensely important that GPs in England are alerted to the existence of the disease.

5. GPs should consider a possible diagnosis of FH if a patient has:

	Total cholesterol	LDL cholesterol
Child/young person	> 6.7 mmol/l	> 4.0 mmol/l
Adult	> 7.5 mmol/l	> 4.9 mmol/l

And at least one of the following:

Family history of myocardial infarction – younger than 50 years of age in second-degree relative or younger than 60 years of age in first-degree relative

or

family history of raised total cholesterol – greater than 7.5 mmol/l in an adult first- or second-degree relative or greater than 6.7 mmol/l in child or sibling aged younger than 16 years.

The full Simon Broome criteria detail a 'definite' and 'possible' diagnosis of familial hypercholesterolaemia, but some experts do not agree with it in its entirety – on purely clinical grounds a definite diagnosis of familial hypercholesterolaemia cannot be made, only genetic testing can do this. So it is best to focus on the parts of the criteria where there is a consensus.

6. Patients should be referred to a specialist lipid clinic

In the absence of complications, familial hypercholesterolaemia does not have any symptoms and a provisional diagnosis is based on signs and laboratory results. Xanthomas, xanthelasmata and arcus are not definite signs and high cholesterol can have other causes.

NICE guidelines recommend that healthcare professionals should offer all people with familial hypercholesterolaemia a referral to a specialist for confirmation of the diagnosis and initiation of cascade testing – a combination of DNA testing and LDL cholesterol to identify affected relatives of patients with a clinical diagnosis. This should include at least the first- and second- and, when possible, third-degree biological relatives.

7. Learn the 'good cop-bad cop' routine

Most patients will ask you to explain the difference between good and bad cholesterol. Simple language is essential. Cholesterol is a type of fat, transported in the circulation packaged into lipoproteins. LDL is the bad type because it transports and builds up lipids into vessel walls that feed the heart and brain, eventually resulting in a heart attack and a stroke if not treated. High density lipoprotein package is the good type because it removes excess cholesterol for disposal in the liver and protects against heart attacks.

8. Familial hypercholesterolaemia requires aggressive lipid-lowering therapy

The aim of lipid lowering is to achieve at least a 50% reduction in LDL cholesterol from baseline. Highly potent statins (e.g. atorvastatin) are necessary to treat familial hypercholesterolaemia as only they can achieve the requisite change in LDL cholesteriol. If the expected reduction does not materialise, the addition of ezetimibe 10 mg is indicated. If there are problems with intolerance or contraindications, the patient should be referred back to the lipid clinic consultant.

9. Lifestyle adjustments are just as important as lipid lowering therapy

Alongside statin therapy, lifestyle issues and blood pressure control should be addressed resolutely.

Smoking cessation is essential. Dietary intervention should aim to reduce saturated fats, and ideally mimic a Mediterranean diet. Weight should be maintained at a BMI of 19–25kg/m^2, or if overweight or obese a minimum of a 10% reduction in body weight is desirable. Alcohol intake should be kept to within safe limits. Exercise should aim to provide 30 minutes of moderate intensity physical activity (e.g. brisk walking) at least five times per week. Blood pressure should be controlled to less than 140/90mmHg.

10. Don’t forget the family

If familial hypercholesterolaemia is found, all other family members should have at least an LDL cholesterol check. Ideally, they should have genetic testing for the family mutation as 25% of cholesterol results are potentially misleading for inheritance of familial hypercholesterolaemia. The total cholesterol and LDL cholesterol levels to diagnose other family members are considerably lower than those required to identify an index case.[1]

Children with familial hypercholesterolaemia, as well as young women seeking advice on contraception or conception and pregnant women, should be referred to a lipid specialist.

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