What's a family worth?

HEART UK FH GIT TOOLKIT

Executive Summary

The HEART UK FH Guideline Implementation Team Toolkit has been developed to assist commissioners and healthcare professionals in the implementation of NICE Clinical Guideline 71, *Identification and Management of familial hypercholesterolaemia (FH)*.

The NICE clinical guideline was published in August 2008, and makes recommendations on the roll-out of a comprehensive service for FH. The guidance covers diagnosis (both LDL-C and DNA testing), identification of family members with FH using cascade testing, management of the condition, information needs and support, and ongoing assessment and monitoring.

This toolkit focuses mainly on the practicalities of getting services in place, diagnosing patients, and the roll-out of cascade testing. For management and ongoing monitoring and assessment of FH, healthcare professionals are advised to refer to the NICE guideline.

The intention is to continue to update the toolkit with new documents, templates and resources as they become available. If you have further documents that are used in your area and think would be beneficial to others who are looking to roll out FH services, then please contact HEART UK at: info@heartuk.org

The under-diagnosis of familial hypercholesterolaemia

FH is a genetic condition that leads to a high concentration of cholesterol in the blood. It is caused by genetic mutations in the pathway that clears LDL-C (low density lipoprotein) from the bloodstream, usually in the LDL-C receptor.

FH can lead to the early onset of atherosclerosis and particularly coronary heart disease. It is passed to offspring in a dominant pattern, meaning that siblings and children of FH patients have a 50% risk of inheriting the condition.

It is estimated that around 1 in 500 people in the UK are affected by FH, which equates to around 120,000 people. This is on a level with Type 1 diabetes.

FH is significantly under-diagnosed in the population, particularly in the under-35 age group. Currently less than 15,000 patients have been identified, meaning that up to 85 per cent of patients with the condition have not been identified or treated. The implementation of cascade testing of family members of FH patients is a vital part of addressing this gap in diagnosis.

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Delivering services based on local capacity

NICE recommends a comprehensive service for FH that assumes that local Specialist Lipid Clinics have the capacity to take the lead in delivering FH services; that is to both definitively diagnose and manage patients. HEART UK champions the need for a full, integrated service in-line with NICE's recommendations, however it recognises that structures within local Trusts may not allow for the exact roll-out exactly as described in the of the NICE model. For this reason, this toolkit includes a number of service specification documents from around the country, each different approaches to co-ordinating and implementing FH services.

This is to enable commissioners and healthcare professionals to examine alternative models, and consider solutions that others have proposed.

Diagnosing FH

The NICE guideline states that healthcare professionals should consider the possibility of FH in adults with raised cholesterol (total cholesterol typically greater than 7.5 mmol/l), especially when there is a personal or a family history of premature coronary heart disease.

The diagnosis of FH in probands is based on the Simon Broome criteria. According to the Simon Broome criteria, the only definitive clinical sign of FH is tendon xanthomas, however the absence of these does not exclude a diagnosis of FH. The majority of patients (classified as Possible Familial Hypercholesterolaemia) will require DNA mutation analysis of the LDLR, APOB or PCSK9 genes and identification of a disease defining mutation in order to confirm the diagnosis and facilitate cascade testing of relatives.

This toolkit includes two model care pathways for FH services: one genetics-centre based, where the specialist lipid service diagnoses and manages FH patients with referral to a centre based genetics counsellor for cascade testing and DNA mutation analysis (which may be commissioned through specialist commissioning); and the other based on an outreach model where the genetic clinical counselling is undertaken by suitably trained nurse counsellors and laboratory DNA mutation analysis testing is referred to the genetics centre.

In the latter model, the role of regionally based Cardiovascular Genetics Specialist Nurses might be expanded to provide sessional support for the Specialist Lipid Clinics on an outreach basis. The second model is more suitable for regions that do not have genetics centres able to contribute the provision of genetic counselling for FH.

Testing family members

The NICE guideline states: "This condition is genetically determined. Families must become the focus of attention so that cascade testing can identify those individuals who will benefit from early treatment and the near-normal life expectancy that can result. The innovative use of DNA testing allied with cholesterol testing will help to ensure that children, young people and adults with this condition are identified and offered timely advice and treatment."[1]

Family members should be identified through drawing the family pedigree, or family tree. A family history of premature coronary heart disease should always be assessed in a person diagnosed with FH. When considering FH cases, healthcare professionals with an expertise in FH should use standardised pedigree terminology to document, where possible, at least a three-generation pedigree.

Family trees should be drawn in such a way that they can be easily interpreted by other healthcare professionals.

Patients should be fully informed of the process of cascade testing. A variety of options for contacting their relatives should be discussed with, and considered by, the patient in order to best suit their circumstances.

Comprehensive records should be maintained on progress with contacting relatives and whether they have been tested. Family files should be properly catalogued and maintained.

GPs of patients undergoing or invited to undergo testing for FH should be notified of the situation and the outcome of their patients' decision about being tested.

Patient information and managing patient data

Healthcare professionals should be equipped with comprehensive, and easy to understand information for patients who have been diagnosed with FH. Healthcare professionals should also be aware of the use of terminology when speaking to patients. For example, the NHS National Genetics Education and Development Centre advises healthcare professionals to consider using the term 'gene alteration' rather than 'mutation'.

Evidence from elsewhere has shown that the use of a specialist database for maintaining and recording patient information is essential to the provision of FH services. This toolkit includes information about PASS Clinical software, currently being trialled in Wales, and used successfully in the Netherlands. This, or an alternative database system, must be considered as part of a comprehensive FH service.

1. NICE guideline (2008) page 7