The Association for Clinical Biochemistry & Laboratory Medicine

Better Science, Better Testing, Better Care

Summary of NICE Guidelines

Title	Familial hypercholesterolaemia: identification and management
NICE Reference	CG71
Date of Review:	December 2020
Date of Publication	August 2008 (Updated October 2019)
Summary of Guidance (Max 250 words)	Familial hypercholesterolaemia (FH) is a genetic condition characterised by high cholesterol levels, which may lead to early development of atherosclerotic disease. In the UK, heterozygous FH affects 1: 250-500 people; homozygous FH affects 1: 1,000,000. This guideline includes recommendations on: • Diagnosis of FH using the Dutch Lipid Clinic Network criteria or the Simon Broome criteria (see below). • Identifying people with FH using cascade testing. • Management, including lipid-modifying drugs, lifestyle interventions and specialist treatment. • Information and support on contraception for women and pregnant women with FH. • Review and referral for evaluation of coronary heart disease (CHD). Risk estimation tools, such as QRISK2, should not be used because people with FH are already at a high risk of premature CHD. The Simon Broome criteria are as follows: Diagnose a person with **definite** FH if they have: o Total cholesterol concentrations of >7.5 mmol/L in adults (>6.7 mmol/L in children/young people) or low-density lipoprotein cholesterol (LDL-C) concentrations of >4.9 mmol/L in adults (>4.0 mmol/L in children/young people) with tendon xanthomas, or evidence of these signs in first- or second-degree relative **or** o DNA-based evidence of an LDL-receptor mutation, familial defective apo B-100, or a PCSK9 mutation. Diagnose a person with **possible** FH if they have total cholesterol or LDL-C concentrations as defined above **and** at least one of the following: o Family history of myocardial infarction aged <50 years in a second-degree relative or <60 years in first-degree relative. o Family history of raised total cholesterol as defined above. A clinical diagnosis of homozygous FH should be considered with an LDL-C concentration >13 mmol/L in adults (>11 mmol/L in children/young people). Children aged less than 10 years old who are at risk of FH should be offered a DNA test at the earliest opportunity.
Impact on Lab (See below)	☐ Moderate

Lab professionals to be made aware *Please select/highlight appropriate choices*	☐ Laboratory Manager ☑ Chemical Pathologist ☑ Clinical Scientist ☐ Biomedical Scientist
Please detail the impact of this guideline (Max 150 words)	This guideline is relevant for healthcare professionals, such as GPs, who should know when to suspect FH and when to refer suspected cases to specialist services. Local guidelines for specialist referral may need to be reviewed. The guideline is relevant to Chemical Pathologists who manage FH and suspected FH patients. The 2019 update included a minor change in wording that is unlikely to have an impact on pathology services unless directly referred to when reporting lipid results. Clinical Scientists may need to review associated reporting comments to ensure that they accurately reflect the guidance.

Impact on Lab

- **None**: This NICE guideline has no impact on the provision of laboratory services
- **Moderate**: This NICE guideline has information that is of relevance to our pathology service and may require review of our current service provision.
- **Important:** This NICE guideline is of direct relevance to our pathology service and will have a direct impact on one or more of the services that we currently offer.

Written by: Mrs Emma Stevenson
Reviewed by: Dr Mathangi Balasubramani