

Summary of NICE Guidelines

Title	Familial hypercholesterolaemia: identification and management
NICE Reference	NICE Clinical Guideline [CG71]
Date of Review:	Dec 2018
Date of Publication	Aug 2008 (updated in Nov 2017)
Summary of Guidance (Max 250 words)	<p>This guideline covers identifying and managing familial hypercholesterolaemia (FH) in children/young people and adults. It aims to help identify people at increased risk of coronary heart disease as a result of having FH. Some of the recommendations that may be relevant to our pathology service include:</p> <ol style="list-style-type: none"> 1. Suspect FH as a possible diagnosis in adults with a total cholesterol level >7.5 mmol/L and/or a personal or family history of premature coronary heart disease (2017). Higher risk of FH in 30 years or older if total cholesterol concentration >9.0 mmol/L (2017). 2. Consider a clinical diagnosis of homozygous FH with a low-density lipoprotein cholesterol (LDL-C) concentration of >13 mmol/L in adults or > 11 mmol/L in children/young people (2008). 3. For people with a personal or family history of premature coronary heart disease, offer to measure their total cholesterol if not been previously measured (2017). 4. Refer the person to an FH specialist service for DNA testing if they meet the Simon Broome criteria for possible or definite FH, or they have a Dutch Lipid Clinic Network (DLCN) score greater than 5. 2017 (2017). 5. Carry out cascade testing using DNA testing to identify affected first- and second- and, when possible, third-degree biological relatives of people with a genetic diagnosis of FH (2017). 6. In children at risk of homozygous FH because of two affected parents or the presence of clinical signs, LDL-C concentration should be measured before the age of 5 years or at the earliest opportunity thereafter (2008).
Lab professionals to be made aware	<ul style="list-style-type: none"> ✓ Chemical Pathologist ✓ Clinical Scientist
Impact on Lab	<p>■ Moderate</p>
Please detail the impact of this guideline (Max 150 words)	<p>Suggested practice at clinical biochemistry service:</p> <ul style="list-style-type: none"> • Comment on test results if total cholesterol > 7.5 mmol/L e.g. ‘Consider possibility of familial hypercholesterolaemia’ • Comment on test results if LDL-C > 13 mmol/L in adults and >11 mmol/L in children/young people e.g. ‘Consider possibility of homozygous familial hypercholesterolaemia’ <p>A local pathway for DNA testing of FH may need to be established. Factors to be considered include funding from local Clinical Commissioning Groups (CCG) funding and practicality of referrals to clinical geneticists or FH specialist service.</p>

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.

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