

UCLPartners Case-finding Tool Familial Hypercholesterolaemia May 2021

Familial Hypercholesterolaemia (FH) is a genetic condition that affects an estimated 1 in 250 people. Around 40 patients can be expected in an average 10,000 patient practice. Half of all men with FH will have a heart attack by the age of 50, and a third of women by the age of 60. These outcomes are highly preventable with lipid lowering treatment. Despite this, 92% are undiagnosed and untreated. Diagnosis requires case finding and genetic testing for the individual and their families as half of all first-degree relatives will be affected.

The UCLP FH Framework automates and simplifies this process and offers a pragmatic solution to case-finding without adding unduly to clinician workload.

1. Searches identify patients with a high cholesterol above the NICE recommended (CG71) thresholds.
2. The Simon Broome (SB) criteria are applied in a semi-automated system to determine if a patient with high cholesterol needs genetic testing.
3. An HCA or other team member then arranges fasting lipids plus renal, liver, thyroid and HbA1c to identify possible secondary causes of raised lipids.
4. If the triglycerides are below 2.3mmol/l, a simplified family history questionnaire is texted to the patient, with interpretation checked by the HCA (supported by guidance). If family history of early CHD is positive, the Simon Broome criteria for genetic testing are met.
5. If family history is negative, the patient should be assessed for tendon xanthomata (TX). This service could be provided across a PCN or CCG by a trained pharmacist or nurse. If TX are present, the Simon Broome criteria for genetic testing are met.

6. For patients in whom SB criteria are met and for those already coded as having FH, a desktop review is conducted by a trained pharmacist or nurse to check results, coding and diagnosis, to exclude secondary causes for the raised lipid levels and to refer to specialist service for assessment, genetic testing and family cascade screening if indicated.

