Investigation of Familial Hypercholesterolaemia

Familial Hypercholesterolaemia (FH) is predominantly an autosomal condition where affected individuals have an increased risk of premature cardiovascular disease (CVD).

The prevalence of FH in Western societies is estimated at 1:500 in both men and women.

Diagnosis of FH

Secondary causes of hypercholesterolaemia should first be excluded, including: hypothyroidism, obesity, alcohol abuse, cholestasis, renal disease, diabetes mellitus, Cushing's syndrome, medications.

The Dutch Lipid Clinic Network Criteria allows an estimate of the likelihood of FH:

Family history	Points
First degree relative with known premature CVD (male < 55 years, female < 60 years), or first degree relative with known LDL-C above the 95th percentile for age and sex (> 2.0 mmol/L)	1
First degree relative with tendinous xanthomata and/or arcus cornealis, or child aged less than 18 years with LDL-C above the 95th percentile for age and sex	2
Clinical history	
Patient with premature coronary artery disease (male < 55 years, female < 60 years)	2
Patient with premature cerebral or peripheral vascular disease (male < 55 years, female < 60 years)	1
Physical examination	
Arcus cornealis prior to age 45 years	4
Tendinous xanthomata	6
LDL-C 4.0–4.9 mmol/L	1
LDL-C 5.0–6.4 mmol/L	3
LDL-C 6.5–8.4 mmol/L	5
LDL-C ≥ 8.5 mmol/L	8
DNA analysis: functional mutation in the LDLR gene	8

Total score

Definite FH	> 8
Probable FH	6–8
Possible FH	3–5
Unlikely FH	< 3

Additional resources

Guidelines for the diagnosis and management of Familial Hypercholesterolaemia. The Cardiac Society of Australia and New Zealand, 2010.

A hypercholesterolaemia DNA panel is available through Sonic Genetics; see https://www.sonicgenetics.com.au/tests/hypercholesterolaemia-panel/