

Diagnostic criteria for Familial Hypercholesterolaemia using Simon Broome register

Definite familial hypercholesterolaemia is defined a

a) Total cholesterol > 6.7 mmol/l or LDL cholesterol above 4.0 mmol/l in a child < 16 years or Total cholesterol >7.5 mmol/l or LDL cholesterol above 4.9 mmol/l in an adult. (Levels either pre-treatment or highest on treatment)

PLUS

b) Tendon xanthomas in patient, or in 1st degree relative (parent, sibling, child), or in 2nd degree relative (grandparent, uncle, aunt)

OR

c) DNA-based evidence of an LDL receptor mutation or familial defective apo B-100

Possible familial hypercholesterolaemia is defined as:

a) above **PLUS ONE OF d) or e)**

d) Family history of myocardial infarction:
below age of 50 in 2nd degree relative or below age 60 in 1st degree relative

e) Family history of raised cholesterols:
>7.5 mmol/l in adult 1st or 2nd degree relative or
> 6.7 mmol/l in child or sibling under 16

