

Table 3: Simon Broome diagnostic criteria for Familial Hypercholesterolemia¹

Definite Familial Hypercholesterolemia:

Required laboratory = high cholesterol levels:

- Adult = Total cholesterol levels > 290 mg/dL (7.5 mmol/L) or LDL-C > 190 mg/dL (4.9 mmol/L)
- Child less than 16 years of age = Total cholesterol levels > 260 mg/dL (6.7 mmol/L) or LDL-C > 155 mg/dL (4.0 mmol/L)

Plus at least one of the two:

1. Plus physical finding = tendon xanthomas, or tendon xanthomas in first or second degree relative

OR

2. DNA-based evidence of an LDL-receptor mutation, familial defective apo B-100, or a PCSK9 mutation.

Possible Familial Hypercholesterolemia

Laboratory = high cholesterol levels:

- Adult = Total cholesterol levels > 290 mg/dL (7.5 mmol/L) or LDL-C > 190 mg/dL (4.9 mmol/L)
- Child less than 16 years of age = Total cholesterol levels > 260 mg/dL (6.7 mmol/L) or LDL-C > 155 mg/dL (4.0 mmol/L)

Plus at least one of the two:

1. Family history of at least one of the following.
 - Family history of myocardial infarction at:
 - Age 60 years or younger in first degree relative
 - Age 50 years or younger in second-degree relative

OR

2. Family history of elevated total cholesterol
 - Greater than 290 mg/dL (7.5 mmol/L) in adult first- or second-degree relative
 - Greater than 260 mg/dL (6.7 mmol/L) in child, brother or sister aged younger than 16 years.

1. Austin MA, Hutter CM, Zimmern RL, Humphries SE. Genetic causes of monogenic heterozygous familial hypercholesterolemia: a HuGE prevalence review. *American journal of epidemiology*. 2004;160:407-420.