

## Appendix diagnostica – Familial hypercholesterolemia diagnostic criteria

### Simon Broome system

**Definite familial hypercholesterolemia (FH)** is defined as follows:

- a) cholesterol levels > 6.7 in children under 16 years of age and > 7.5 in adults; or LDL-C levels > 4.9 in adults *and*
- b) tendinous xanthomas in a patient or a first-/second-degree relative.

**Possible familial hypercholesterolemia** is defined as follows:

- a) cholesterol levels > 6.7 in children under 16 years of age and > 7.5 in adults; or LDL-C levels > 4.9 in adults *and one of the following criteria*:
- b) family history of myocardial infarction prior to 50 years of age in second-degree relatives, or prior to 60 years in first-degree relatives
- c) family history of elevated cholesterol > 7.5 in first-/second-degree relatives.

### MedPed criteria used in the United States

**Tab. 1. Total cholesterol cutpoints (in mmol/l) used in FH diagnosis in MedPed**

age (years)	first degree relative with FH	second degree relative with FH	third degree relative with FH	general population
< 20	5.7	5.9	6.2	7.0
20–29	6.2	6.5	6.7	7.5
30–39	7.0	7.2	7.5	8.8
≥ 40	7.5	7.8	8.0	9.3

Diagnosis: FH is diagnosed if total cholesterol levels exceed the Cut Point

### Dutch Lipid Clinic Network criteria for FH

**Tab. 2. Dutch criteria points**

criteria	points
<b>family history</b>	
i. first-degree relative with CAD, M ≤ 55 years, F ≤ 60 years	1
ii. first-degree relative with LDL-C above 95th percentile for a given country	1
iii. first-degree relative with tendinous xanthoma and/or arcus lipoides	2
iv. children under 18 years with LDL-C above 95th percentile for a given country	2
<b>personal clinical history</b>	
i. premature CAD, M ≤ 55 years, F ≤ 60 years	2
ii. premature peripheral or cerebral atherosclerosis, M ≤ 55, F ≤ 60 years	1
<b>physical exam</b>	
i. tendinous xanthoma	6
ii. arcus lipoides in patients under 45 years	4
<b>biochemical exam (LDL-C [mmol/L])</b>	
> 8.5	8
6.5–8.4	5
5.0–6.4	3
4.0–4.9	1
<b>molecular biology – diagnostic mutation testing</b>	
i. mutations in the genes encoding LDL-R, ApoB or PCSK9	8

CAD – coronary artery disease M – male F – female LDL-C – low density cholesterol

The Dutch criteria are then interpreted as follows: For a definite diagnosis of FH, the subject must have more than 8 points; a probable diagnosis of FH requires 6–8 points; and a possible diagnosis of FH is indicated by 3–5 points. Patients who score 0–2 points most likely do not have FH.