SIMON BROOME REGISTER DIAGNOSTIC CRITERIA FOR HETEROZYGOUS FAMILIAL HYPERCHOLESTEROLEMIA[1,2]

Patient name .....

Total cholesterol or LDL-C level ..... **Age** .....

Familial hypercholesterolaemia (FH) is a common genetic condition characterised by **very high plasma levels of low-density lipoprotein cholesterol (LDL-C)** from birth and **~20-fold higher risk** of premature coronary heart disease (CHD); however, **FH is currently under-diagnosed**.[3,4]

- Formal clinical diagnosis of definite or possible FH can be made using the Simon Broome Register diagnostic criteria which take into consideration cholesterol levels, a molecular FH diagnosis, clinical signs that may be present in some patients with FH, and family history[1,2]

A DEFINITE DIAGNOSIS OF FH REQUIRES

(a) Total cholesterol level above 7.5 mmol/L (>290 mg/dL) in adults or a total cholesterol level above 6.7 mmol/L (>260 mg/dL) for children under 16

OR
LDL-C levels above 4.9 mmol/L (>190 mg/dL) in adults (>4.0 mmol/L [>155 mg/dL] in children) (either pretreatment or highest on treatment)

PLUS
(b) Tendon xanthomas in patient or first- or second-degree relatives

OR
(c) DNA-based evidence of a LDL receptor mutation, familial defective APOB-100 or PCSK9 mutation

POSSIBLE FH IS DEFINED AS (a) ABOVE PLUS ONE OF (d) OR (e)

(d) Family history of myocardial infarction before age 50 in a second-degree relative or before age 60 in a first-degree relative

(e) Family history of raised cholesterol in a first-degree relative, or level above 7.5 mmol/L (>290 mg/dL) in a second-degree relative

Recommendations for further screening and testing[3]

- **Genetic testing is strongly recommended** among individuals with a definite or possible diagnosis of FH and particularly in those with an obvious clinical diagnosis with xanthoma and/or high cholesterol plus a family history of premature CHD
- In patients with a confirmed diagnosis of FH, **cascade screening of family members is recommended** using LDL-C levels or genetic testing if the causative mutation is found
- All subjects with FH and their families should undergo **intensive education targeting lifestyle** management
- **Most FH patients will need active treatment** as their risk is significantly higher than the rest of the population due to their long exposure to high LDL-C levels

Your patient's diagnosis	
Family screening initiated?	YES / NO

Further information on FH for the patient

- Please refer the patient to the booklet entitled 'A guide to familial hypercholesterolemia (FH) for patients'
- Provide details of local patient associations

NOTES

Prepared March 2015
G-ALI-0505
SAGLB.ALI.15.01.0022e

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References

1. Simon Broome Register Group. BMJ. 1991;303:893-6
2. Marks D et al. Atherosclerosis. 2003;168:1-14
3. Nordestgaard B et al. Eur Heart J. 2013;34:3478-90a
4. Goldberg et al. J Clin Lipidol. 2011;5(3 Suppl):S1-8