Diagnostic criteria for homozygous familial hypercholesterolaemia (HoFH)

(Cuchel M. et al., Eur Heart J 2014 Aug 21;35(32):2146-57. Accessible at: http://eurheartj.oxfordjournals.org/content/early/2014/07/22/eurheartj.ehu274)

- Genetic confirmation of two mutant alleles at the *LDLR*, *APOB*, *PCSK9*, or *LDLRAP1* gene locus

OR

- An untreated LDL-C >13 mmol/L (500 mg/dL) or treated LDL-C ≥8 mmol/L (300 mg/dL)*,

together with either:

- Cutaneous or tendon xanthoma before age 10 years

or

- Untreated elevated LDL-C levels consistent with heterozygous FH in both parents

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* These LDL-C levels are only indicative, and lower levels, especially in children or in treated patients, do not exclude HoFH

Abbreviations: *LDLR*, low density lipoprotein receptor, *APOB*, apolipoprotein B, *PCSK9*, proprotein convertase subtilisin/kexin type 9, *LDLRAP1*, low density lipoprotein receptor adaptor protein 1, LDL-C, low density lipoprotein cholesterol, FH, familial hypercholesterolaemia