

MAKE EARLY DIAGNOSIS PREVENT EARLY DEATH (MEDPED) PROGRAM DIAGNOSTIC CRITERIA FOR HETEROZYGOUS FAMILIAL HYPERCHOLESTEROLEMIA¹

Familial hypercholesterolemia (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**.^{2,3}

Clinical diagnosis of probable heterozygous FH (HeFH) can be made based on MEDPED Program Diagnostic Criteria using the table below and details of the patient's age, cholesterol level and whether they have relatives with FH.

Patient name

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What is your patient's age?

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What is your patient's level of total cholesterol?

.....

Or LDL-C level?

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Does the patient have any known relatives with FH?

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If YES, what relation is the family member with FH to the patient?

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Probable HeFH is diagnosed* if total cholesterol/LDL-C levels exceed these cut points [†]				
	Patient has first-degree relative with FH e.g. parents, offspring, brothers and sisters	Patient has second-degree relative with FH e.g. aunts, uncles, grandparents, nieces, nephews	Patient has third-degree relative with FH e.g. first cousins, siblings of grandparents	No known relative with FH
Total cholesterol/LDL-C values in mg/dL				
Age <20	220/155	230/165	240/170	270/200
Age 20–29	240/170	250/180	260/185	290/220
Age 30–39	270/190	280/200	290/210	340/240
Age 40+	290/205	300/215	310/225	360/260
Total cholesterol/LDL-C values in mmol/L				
Age <20	5.7/4.0	5.9/4.3	6.2/4.4	7.0/5.2
Age 20–29	6.2/4.4	6.5/4.7	6.7/4.8	7.5/5.7
Age 30–39	7.0/4.9	7.2/5.2	7.5/5.4	8.8/6.2
Age 40+	7.5/5.3	7.8/5.6	8.0/5.8	9.3/6.7

*With 98% specificity
[†]Based on a US population

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Recommendations for further screening and testing^{2,3}

- **Genetic screening for FH may be useful** when the diagnosis is uncertain
- **Cascade screening is strongly encouraged** in all first-degree relatives of diagnosed FH patients
- 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

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References

1. Williams RR et al. Am J Cardiol 1993;72:171-6
2. Hopkins PN et al. J Clin Lipidol. 2011;5(3 Suppl):S9-S17
3. Goldberg AC et al. J Clin Lipidol. 2011;5(3 Suppl):S1-8