Implications of new clinical practice guidance on familial hypercholesterolaemia for Australian general practitioners

**Focus | Professional**

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**Gene
eral practice clinics**

**Patient clinically reviewed and scored according to DLCNC**

**Identification of high risk of FH**
- DLCNC score
- LDL-C \( \geq 6.5 \) mmol/L in the absence of secondary causes
- LDL-C \( 5.0-6.6 \) mmol/L with signs of premature or accelerated atherogenesis

**GP standard care for phenotypic FH**
- Care plans 721/723

**GP referral to a specialist to request FH genetic testing**
- MBS Item 73352 (if not previously tested)

**GP arrangement of genetic testing for first- or second-degree family members**
- MBS Item 73353 (if not previously tested)

**Management of FH**
- Care plans 721/723

**Low complexity**
- Absence of CVD
- Absence of risk factors for CVD
- Reached LDL-C goal with a statin

**Intermediate complexity**
- Stable CVD and risk factors
- Close to LDL-C goal with statin
- Low-grade statin intolerance
- HeFH aged <18 years

**High complexity**
- Numerous uncontrolled CVD risk factors
- Symptomatic CVD
- Failure to reach LDL-C goal despite statin/ezetimibe
- High-level statin intolerance
- Children, pregnancy, need for PCSK9 inhibitors or LDL-C apheresis
- HeFH >18 years or HoFH
- Help with cascade screening

**Primary care**

**Shared care**

**Tertiary care**

*Figure 1. Process for genetic screening and management of an individual at high risk of familial hypercholesterolaemia*


*Genetic cascade testing may be undertaken by a general practitioner with skills in the care of patients and families with FH, under the guidance of an appropriate specialist. Consent is obtained from the index case to contact family. The process of risk notification of family members should be consistent with relevant local legislation and institutional guidelines. Risk notification may be indirect (providing a family letter for the notifier to pass to relatives) or direct (clinical service writes to relatives); pre-and post-test genetic counselling should be offered to all at-risk family members.*

CVD, cardiovascular disease; DLCNC, Dutch Lipid Network Criteria; FH, familial hypercholesterolaemia; GP, general practitioner; HeFH, heterozygous familial hypercholesterolaemia; HoHF, homozygous familial hypercholesterolaemia; LDL-C, low-density lipoprotein cholesterol; PCSK9, proprotein convertase subtilisin/kexin 9