

| Genetic cascade test result ^a | LDL-cholesterol level ^b | Likely diagnosis | Recommended action |
|--|------------------------------------|-----------------------|---|
| Negative | Within normal limits | Not FH | Arrange cholesterol reviews as appropriate depending on their age and risk factors |
| Negative | Elevated | Hypercholesterolaemia | Manage according to the Hyperlipidaemia pathway |
| Positive | Elevated | FH | Start treatment as per Familial Hypercholesterolaemia (FH) pathway and refer to a lipid specialist; refer children to a paediatric lipid specialist |
| Positive | Within normal limits | FH | Refer to a lipid specialist for management advice |

a) Refers to pathogenic variant or likely pathogenic variant.

b) Refers to LDL-cholesterol diagnostic thresholds from Development of sensitive and specific age-and gender-specific low-density lipoprotein cholesterol cut offs for diagnosis of first-degree relatives with familial hypercholesterolaemia in cascade testing B. Starr et al. Clinical Chemistry and Laboratory Medicine 2008 Vol. 46 Issue 6 Pages 791-803.

Figure 3: Outcomes of genetic and cholesterol cascade testing, likely diagnoses and recommended actions.