

Familial Hypercholesterolaemia Scenario 2

Mark Lambert (date of birth 12.01.1969) attended an appointment for a well person check, which included the measurement of his cholesterol concentrations. The result of his cholesterol test was 8.0mmol/l. As well as asking about his lifestyle, you ask him about his family history.

Mark's family history

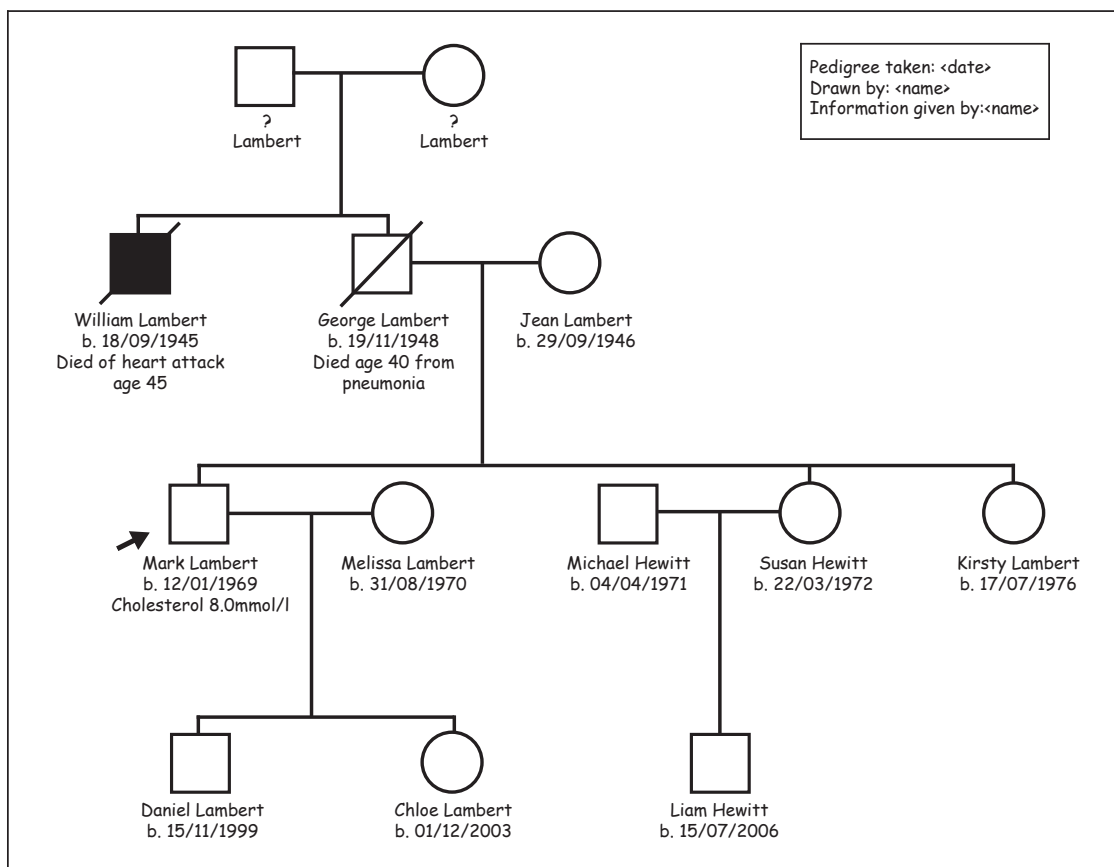


Fig. 1 Mark's family tree

According to the Simon Broome diagnostic criteria – individuals can be diagnosed with possible FH if they have cholesterol concentrations >7.5 mmol/l (for an adult) and a family history of myocardial infarction aged younger than 50 years in a second-degree relative or aged younger than 60 years in first-degree relative.

Based on these criteria, Mark would be diagnosed with possible FH. You would discuss referral to a lipid clinic for confirmation of the diagnosis and the initiation of cascade testing in other family members.