FROM: The HEART UK FH Guideline Implementation Team Toolkit

The under-diagnosis of familial hypercholesterolaemia

FH is a genetic condition that leads to a high concentration of cholesterol in the blood. It is caused by genetic mutations in the pathway that clears LDL-C (low density lipoprotein) from the bloodstream, usually in the LDL-C receptor.

FH can lead to the early onset of atherosclerosis and particularly coronary heart disease. It is passed to offspring in a dominant pattern, meaning that siblings and children
of FH patients have a 50% risk of inheriting the condition.

It is estimated that around 1 in 500 people in the UK are affected by FH, which equates to around 120,000 people. This is on a level with Type 1 diabetes.

FH is significantly under-diagnosed in the population, particularly in the under-35 age group. Currently less than 15,000 patients have been identified, meaning that up to 85 per cent of patients with the condition have not been identified or treated. The implementation of cascade testing of family members of FH patients is a vital part of addressing this gap in diagnosis. https://heartuk.org.uk/FHToolkit/