Gene test information

FAMILIAL DEFECTIVE APOLIPOPROTEIN B-100 (APOB 3500Q MUTATION)

- **Background**

  Familial defective apolipoprotein B-100 (FDB) is the most prevalent monogenic lipoprotein disorder in Central Europe and caused by mutations in the APOB gene encoding the apolipoprotein (apo) B-100 molecule. Apo B-100 is part of the LDL particle and mediates as ligand the uptake of the cholesterol-rich LDL particle into the cell. Cholesterol is elevated within the blood in patients with FDB, causing atherosclerotic changes and, as a consequence, heart attacks or cerebrovascular strokes.

  The most frequent mutation leading to familial defective apo B-100 is an arginin (R) to glutamin (Q) mutation at amino acid position 3500 (APOB R3500Q).

Genotype	Commentary
APOB RR	Wildtype ("normal") ApoB-100 (no R3500Q mutation)
APOB RQ:	Heterozygous ApoB-100 R3500Q Mutation (frequency about 1:500)
APOB QQ:	Homozygous ApoB-100 R3500Q Mutation (very rare)

- **Clinical consequences of the APOB R3500Q mutation**

  Carriers of an APOB R3500Q mutation are at strongly increased risk for hypercholesterolemia. The risk to develop heart attacks and cerebrovascular strokes is similar to that in familial hypercholesterolemia (LDL receptor mutations).

- **Indications for testing**

  - Hyperlipidemie (hypercholesterolemia)
  - First-degree relatives of patients with an APOB R3500Q mutation.

References:

Whitfield et al., Lipid disorders and mutations in the APOB gene. Clin Chem. 2004 50:1725-32.

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