Gene test information

APOE GENETICS

- **Background**

  Apolipoprotein E (ApoE) is a constituent of lowdensity lipoproteins (LDL) and mediates the transport of cholesterol from the liver into the body tissue. ApoE also performs numerous functions in addition to lipid metabolism, such as modulation of cellular immune response, inhibition of thrombocyte aggregation and regulation of steroid synthesis. In the peripheral nervous system ApoE influences growth and differentiation of neurons. As a result of this and the breaking down of neurotoxic amyloid-peptides and plaque compounds, ApoE is attributed a role in the pathogenesis of Alzheimer's disease.

  The human ApoE gene is located on chromosome 19 and occurs in three relatively common polymorphisms. The most common isoform Apo e3 has a single cysteine at position 112 and an arginine at position 158. Apo e2 contains 2 cysteine residues at these positions and Apo e4 two arginine residues. Depending on the combination, there are as many as six known different genotypes.

- **APOE Genotypes**

Genotype group:	Genotypes (frequency)
Wildtype genotype	3-3 (59%)
Carriers of e4	3-4 (23%), 4-4 (2%)
Carriers of e2	2-3 (12%), 2-2 (1%), 2-4 (2%)

- **Clinical consequences of APOE genotypes**

  Compared to the wild type, ApoE2 has less affinity to the LDL receptor. This may result in hypercholesterolaemia and increased risk for cardiovascular disease.

  Carriers of an APOE e4 allele are at increased risk for Alzheimer's disease. Alzheimer's disease is a multifactorial disease and the presence of an e4 allele alone is not sufficient for the development of the disease. Nevertheless, ApoE genotyping may increase the specificity of clinical diagnosis and thus be helpful in Alzheimer diagnosis.

References:

Mahley et al., Apolipoprotein E: from atherosclerosis to Alzheimer's disease and beyond. Curr Opin Lipidol. 1999;10:207-17.

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