Clinical implications

Apolipoprotein B100 (APOB100) is one of the B forms of apolipoprotein and it forms the basic protein component of lipoproteins with low and very low density (LDL, VLDL). Thus it plays a significant role during lipid and cholesterol transport. It is synthesized in the liver and distributed by the blood stream to the cells on whose surface it works as a ligand for LDL receptors mediating cholesterol transport to the cells. The most common APOB gene mutation is an R3500Q mutation, which leads to the exchange of amino acid arginine (Arg) for glutamin (Gln) in position 3527. In such a way it causes a change in the protein structure in the place of the receptor binding. Consequently, there is a lower affinity of LDL particles towards the receptors and their accumulation in blood. This genetic disorder is called a familial defective Apolipoprotein B100 and it is one of the causes of familial hypercholesterolemia. Cholesterol accumulation in blood increases the risk of atherosclerosis and heart attack. Although the frequency of the mutant allele is only 0.1% of the healthy population, the frequency is up to 10% in individuals with familial hypercholesterolemia.

Principle of detection

The kit is intended for detection of mutation of R3500Q in the APOB gene (apolipoprotein B) in human genomic DNA. Detection is based on real-time polymerase chain reaction (qPCR) using fluorescently labelled probes (allelic discrimination).

Available products

Cat. No.	Product	rxn
3205-025	gb GENETIC APOB	25

1 kit contains reagents to provide 25 PCR reactions (20 µl volume of each reaction).

Parameters of the diagnostic kit

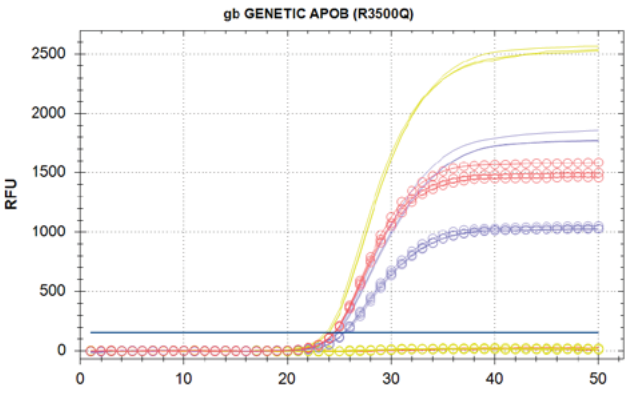
- ***in vitro* diagnostics**
- **CE IVD, IVDR certified**
- **ready-to-use assay**
- **sample concentration 10-100 ng/µl**
- **positive and negative controls included**
- **FAM and HEX channels detection**
- **identical amplification profile as gb HEMO, gb GENETIC, gb PHARM kits**

Content of the diagnostic kit

*	Component	Conc.	Purpose
🟢	Assay qPCR APOB (3500Q)	1.25×	Detection assay
⚪	Deionized Water		Negative Control
🟡	Standard WT APOB (3500Q)	10^4 cop/µl	Positive Control
🔴	Standard MUT APOB (3500Q)	10^4 cop/µl	Positive Control
🟣	Standard HET APOB (3500Q)	10^4 cop/µl	Positive Control

** Lid colour*





Validated for cyclers

- **Rotor-Gene 3000 (Corbett Research)**
- **iCycler iQ5/CFX96/CFX96 Touch (Bio-Rad)**
- **ABI 7500/7500 Fast/7900HT (Applied Biosystems)**
- **MIC (Bio Molecular Systems)**
- **AriaMx (Agilent Technologies)**
- **Light Cycler 480/Cobas z480 (Roche Diagnostics)**
- **QuantStudio 5 (Applied Biosystems)**

Fig. 1 – Detection of APOB (R3500Q) standards on CFX96 device; yellow line – wild type; red line – mutant; violet line – heterozygote; smooth line – FAM channel; dotted line – HEX channel