

# APO B-100 G10580A (R3500Q) MUTATION

## ORDERING INFORMATIONS

REF: GEN-016-25 RDM Code: 2256375/R  
Tests: 25 Reactions: 31  
REF: GEN-016-50 RDM Code: 1791315/R  
Tests: 50 Reactions: 62  
CND Code: W0106010499  
Manufacturer: BioMol Laboratories s.r.l.

## CONTENTS OF THE KIT

The kit consists of reagents for  
Real-Time PCR amplification  
\*reagents for the extraction of  
genomic DNA are not supplied  
in the kit

For in vitro diagnostic use



## PRODUCT CHARACTERISTICS

Device belonging to the family of in vitro medical devices **REAL-TIME QUALITATIVE PCR-GENETIC VARIANTS**. Detection of G10580A (R3500Q/R3527Q) polymorphism of the APO-B 100 gene by Real-Time PCR technique. Kit optimized for Real-Time PCR instrumentation Biorad CFX96 Dx, Biorad Opus Dx and Agilent AriaDx.

## SCIENTIFIC BACKGROUND

One of the forms of familial hypercholesterolemia is familial apolipoprotein B-100 deficiency (FDB-Familial defective apolipoprotein B100), an autosomal dominant hereditary disease caused by mutations in the apo B gene. The gene contains 29 exons and 28 introns with a total length of 43 kb and is located on the short arm of chromosome 2. Apo B is a large amphipathic glycoprotein with two isoforms: apo B-100, which is synthesized in hepatocytes, and apo B-48, which is synthesized in the cells of the small intestine. Apolipoprotein B-100 (Apo B) is a protein involved in lipid metabolism and is the main constituent protein of very low-density lipoproteins (VLDL) and low-density lipoproteins (LDL). The Apo B-100-cholesterol complex is recognized by LDL membrane receptors and then reabsorbed into cells. Four mutations in the APO B gene, R3480P, R3500Q (new nomenclature R3527Q), R3500W and R3531C are responsible for FDB by reducing the binding of LDL particles to the LDL receptor. The R3500Q mutation (rs5742904) was the first described and is the most widespread. The prevalence of FDB has been estimated to be approximately 1/500 in North America, while in Europe it appears to be highest in northwestern Switzerland (1/114), eastern France, and southern Germany and lower in Italy and Spain. The Apo B-100 protein with the mutation remains free in the blood, causing hypercholesterolemia and increased risk of the formation of obstructive plaques, constituting an important risk factor for the development of early atherosclerosis and coronary artery deficiencies (coronary artery disease, CAD)

§ Metabolites. 2024 Feb 12;14(2):123. doi: 10.3390/metabol14020123. ApoB100 and Atherosclerosis: What's New in the 21st Century?

§ Int J Mol Sci. 2023 Apr 21;24(8):7635. doi: 10.3390/ijms24087635. Identification and Functional Analysis of APOB Variants in a Cohort of Hypercholesterolemic Patients

§ J Intern Med. 2023 Feb;293(2):144-165. doi: 10.1111/joim.13577. Epub 2022 Oct 17. Genetic and molecular architecture of familial hypercholesterolemia

§ Familial defective apolipoprotein B-100: A review. J Clin Lipidol. 2016 Nov - Dec; 10 (6):1297-1302. doi: 10.1016/j.jacl.2016.09.009. Epub 2016 Sep 22.

§ Decreased bone mineral density in subjects carrying familial defective apolipoprotein B-100. J Clin Endocrinol Metab. 2013 Dec; 98 (12):E1999-2005. doi: 10.1210/jc.2013-2471. Epub 2013 Oct 8.

§ Genetic cardiovascular risk factors and age-related macular degeneration. Acta Ophthalmol. 2011 Jun; 89 (4):335-8. doi: 10.1111/j.1755-3768.2009. 01697.x Epub 2009 Oct 23.

## CLINICAL SIGNIFICANCE

Familial hypercholesterolemia is a genetic pathology characterized by high concentrations of cholesterol in the plasma, transported by low-density lipoproteins (LDL). Under normal conditions, LDL particles are removed from the plasma approximately 2.5 days after their production through their binding to the LDL receptor, LDLR.

In Familial Hypercholesterolemia there is a persistence of LDL cholesterol in the blood and its deposition in the walls of the arteries (mainly coronary arteries, aorta and heart valves), in the tendons and in the skin. The main consequence of this pathology is premature atherosclerosis, responsible for myocardial infarction and angina pectoris which appear at variable ages in relation to the type of genetic defect.

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| DESCRIPTION                          | LABEL                      | VOLUME       |              | STORAGE |
|--------------------------------------|----------------------------|--------------|--------------|---------|
|                                      |                            | GEN-016-25   | GEN-016-50   |         |
| Mix oligonucleotides and probes      | Mix G10580A APO-B 10X      | 1 x 77,5 µl  | 2 x 77,5 µl  | -20°C   |
| Mix buffer and Taq polymerase enzyme | Mix Real-Time PCR 2X       | 1 x 387,5 µl | 2 x 387,5 µl | -20°C   |
| Deionized H <sub>2</sub> O           | Deionized H <sub>2</sub> O | 1 x 1 ml     | 1 x 1 ml     | -20°C   |
| Genomic DNA or recombinant DNA       | Control 1                  | 1 x 22 µl    | 2 x 22 µl    | -20°C   |
| Genomic DNA or recombinant DNA       | Control 2                  | 1 x 22 µl    | 2 x 22 µl    | -20°C   |
| Genomic DNA or recombinant DNA       | Control 3                  | 1 x 22 µl    | 2 x 22 µl    | -20°C   |

## TECHNICAL CHARACTERISTICS

COD. GEN-016-25 / COD. GEN-016-50

|                                                 |                                                                                                                                    |
|-------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------|
| STABILITY                                       | 18 months                                                                                                                          |
| REAGENTS STATUS                                 | Ready to use                                                                                                                       |
| BIOLOGICAL MATRIX                               | Genomic DNA extracted from whole blood, tissue, cells                                                                              |
| CONTROLS                                        | Recombinant DNA for at least 3 analytical sessions (GEN-016-25)<br>Recombinant DNA for at least 6 analytical sessions (GEN-016-50) |
| VALIDATED INSTRUMENTS                           | Biorad CFX96 Dx, Biorad Opus Dx and Agilent AriaDx                                                                                 |
| TECHNOLOGY                                      | Real-time PCR; oligonucleotides and specific probes; 2 FAM/HEX fluorescence channels                                               |
| RUNNING TIME                                    | 85 min                                                                                                                             |
| THERMAL CYCLING PROFILE                         | 1 cycle at 95 °C (10 min); 45 cycles at 95 °C (15 sec) + 60 °C (60 sec)                                                            |
| ANALYTICAL SPECIFICITY                          | Absence of non-specific pairings of oligonucleotides and probes;<br>absence of cross-reactivity                                    |
| LIMIT OF DETECTION (LOD)                        | ≥ 0,016 ng of genomic DNA                                                                                                          |
| LIMIT OF BLANK (LOB)                            | 0% NCN                                                                                                                             |
| REPRODUCIBILITY                                 | 99,9%                                                                                                                              |
| DIAGNOSTIC SPECIFICITY / DIAGNOSTIC SENSITIVITY | 100%/98%                                                                                                                           |