APO B-100 R3500Q (R3527Q) MUTATION

For in vitro diagnostic use

CE IVD

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*

PRODUCT CHARACTERISTICS

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

Familial hypercholesterolemia is a genetic pathology characterized by high concentrations of cholesterol in the plasma, in particular transported by low-density lipoproteins (LDL). Under normal conditions, LDL particles are cleared from plasma approximately 2.5 days after their production by 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, aorta and heart valves), tendons and 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.

§ 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

One form 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 major constituent protein of very low-density lipoprotein (VLDL) and low-density lipoprotein (LDL). The Apo B-100-cholesterol complex is recognized by membrane LDL receptors and then reabsorbed into cells. Four mutations of 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 (rs5742904) mutation was the first described and is the most common. The prevalence of FDB has been estimated at 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 lowest in Italy and Spain. The Apo B-100 protein with the mutation remains free in the blood, causing hypercholesterolemia and increased risk of obstructive plaque formation, constituting an important risk factor for the development of early atherosclerosis and coronary artery deficiencies (coronary artery disease, CAD).

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ISO 9001:2015
ISO 13485:2016

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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 85 µl	1 x 170 µl	-20°C
Mix buffer and Taq polymerase enzyme	Mix Real-Time PCR 2X	1 x 425 µl	1 x 850 µl	-20°C
Deionized H_20	Deionized H_20	2 x 1 ml	2 x 1 ml	-20°C
Genomic DNA or recombinant DNA	Control + 1	1 x 22µl	1 x 22 µl	-20°C
Genomic DNA or recombinant DNA	Control + 2	1 x 22µl	1 x 22µl	-20°C
Genomic DNA or recombinant DNA	Control + 3	1 x 22µl	1 x 22µl	-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
POSITIVE CONTROL	Recombinant DNA for at least 3 analytical sessions
VALIDATED INSTRUMENTS	Biorad CFX96 Dx, Biorad Opus Dx e 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; absence of cross-reactivity
ANALYTICAL SENSITIVITY : LIMIT OF DETECTION (LOD)	≥ 0,016 ng of DNA
ANALYTICAL SENSITIVITY : LIMIT OF BLANK (LOB)	0% NCN
REPRODUCIBILITY	99,9%
DIAGNOSTIC SPECIFICITY / DIAGNOSTIC SENSITIVITY	100%/98%

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