

ADRB2 and ADRB3 GENETIC VARIANTS (METABOLIC SYNDROME)

ORDERING INFORMATION

REF: GEN-042-25
Tests: 25 Reactions: 31 x 3
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

Device belonging to the family of in vitro devices REAL-TIME QUALITATIVE PCR-GENETIC VARIANTS- RESEARCH USE ONLY. Determination of genetic variants rs1042713 (c.46 G>A) and rs1042714 (c.79 C>G) of the ADRB2 gene and rs4994 (Trp64Arg, T>C) of the ADRB3 gene by Real-Time PCR technique. Kit optimized for Real-time PCR instruments Biorad CFX96 Dx, Biorad Opus Dx, Agilent AriaDx, Thermofisher QuantStudio™ 5 Real-Time PCR System.

SCIENTIFIC BACKGROUND

The *ADRB3* and *ADRB2* genes are part of the adrenergic system, a major contributor to energy expenditure and body weight regulation. These receptors are expressed in adipose tissue and skeletal muscles, and stimulate lipolysis in fat cells.

The β_2 -adrenergic receptor is encoded by an intronless gene (*ADRB2*) located on chromosome 5 (5q31-32) and responds to sympathetic stimulation for vasodilation and nitric oxide release in the endothelium, thus affecting blood pressure control. It is implicated in several functions concerning the central nervous, cardiovascular, endocrine, and pulmonary systems.

The *ADRB3* gene is 3610 bp in length and is located on chromosome 8p11.23, containing two exons and one intron, encoding 408 amino acids.

CLINICAL SIGNIFICANCE

The gene **ADRB2** contains several single nucleotide polymorphisms. The two frequent and most studied variants are rs1042713 (c.46G>A; p.Gly16Arg) and rs1042714 (c.79G>C; p.Glu27Gln). The Gly16 allele potentiates agonist-promoted receptor downregulation (decreasing cAMP production), while Glu27 produces resistance to agonist-promoted downregulation (increasing cAMP production).

The rs4994 polymorphism of **ADRB3** (T/C, Trp64Arg) involves a cytosine substitution in place of thymine at codon 64 of the gene, which has the effect of converting tryptophan to arginine (Trp64Arg) in the amino acid sequence. The Trp64Arg polymorphism is functional and affects *ADRB3* receptor activity by disrupting lipid metabolism through reduced lipolysis.

ADRB2 polymorphisms may have a role in the pathogenesis of Functional gastrointestinal disorders (FGID).

The Arg allele carriers of *ADRB3* gene Trp64Arg polymorphism may be at an increased risk for developing for essential hypertension (EH).

§ Mol Genet Genomics 2022 Nov;297(6):1495-1503. Development of a genetic risk score for obesity predisposition evaluation.

§ Gene X. 2019 May 18;3:100019. Association of FTO and ADRB2 gene variation with energy restriction induced adaptations in resting energy expenditure and physical activity

§ J Sports Sci Med. 2018 Mar 1;17(1):134-144. Individual Responsiveness to Exercise-Induced Fat Loss and Improvement of Metabolic Profile in Young Women is Associated with Polymorphisms of Adrenergic Receptor Genes

§ Front Genet. 2018 Apr 4;9:106. ADRB3 Gene Trp64Arg Polymorphism and Essential Hypertension: A Meta-Analysis Including 9,555 Subjects

§ Aliment Pharmacol Ther. 2013 Jun 20;38(3):313-323. Genetic variation in the beta-2 adrenergic receptor (ADRB2) predicts functional gastrointestinal diagnoses and poorer health-related quality of life.

§ Controlled Clinical Trial Obesity (Silver Spring). 2011 Jan;19(1):212-5. Role of β_2 -adrenergic receptor polymorphisms on body weight and body composition response to energy restriction in obese women: preliminary results



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DESCRIPTION	LABEL	VOLUME	STORAGE
		GEN-042-25	
Mix oligonucleotides and probes	Mix 1 (ADRB2 c.46 G>A) 10X	1 x 62 µl	-20°C
Mix oligonucleotides and probes	Mix 2 (ADRB2 c.79 C>G) 10X	1 x 62 µl	-20°C
Mix oligonucleotides and probes	Mix 3 (ADRB3 Trp64Arg) 10X	1 x 62 µl	-20°C
Mix buffer and Taq polymerase enzyme	Mix Real-Time PCR 2X	1 x 930 µl	-20°C
Deionized H ₂ O	Deionized H ₂ O	1 x 1 ml	-20°C
Genomic DNA or recombinant DNA Controls ADRB2 rs1042713	Control 1 c.46G>A	1 x 22 µl	-20°C
	Control 2 c.46G>A	1 x 22 µl	-20°C
	Control 3 c.46G>A	1 x 22 µl	-20°C
Genomic DNA or recombinant DNA Controls ADRB2 rs1042714	Control 1 c.79 C>G	1 x 22 µl	-20°C
	Control 2 c.79 C>G	1 x 22 µl	-20°C
	Control 3 c.79 C>G	1 x 22 µl	-20°C
Genomic DNA or recombinant DNA Controls ADRB3 rs4994	Control 1 Trp64Arg	1 x 22 µl	-20°C
	Control 2 Trp64Arg	1 x 22 µl	-20°C
	Control 3 Trp64Arg	1 x 22 µl	-20°C

TECHNICAL CHARACTERISTICS

COD. GEN-042-25

STABILITY	18 months
REAGENTS STATUS	Ready to use
BIOLOGICAL MATRIX	Genomic DNA extracted from whole blood, tissues, cells
CONTROLS	Recombinant DNA for at least 3 analytical sessions
VALIDATED INSTRUMENTS	Biorad CFX96 Dx, Biorad Opus Dx and Agilent AriaDx, Thermofisher QuantStudio™ 5 Real-Time PCR System.
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 oligonucleotides and probes; absence of cross-reactivity
LIMIT OF DETECTION (LOD)	≥ 5 ng of genomic DNA
LIMIT OF BLANK (LOB)	>40 Cq
REPRODUCIBILITY	99,9%
SPECIFICITY / SENSITIVITY	99,9%/98%