



ACTN3 and PPARG1 α GENETIC VARIANTS

ORDERING INFORMATION

REF: GEN-046-25
Tests: 25 Reactions: 31 x 2
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**. Detection of the R577X (C>T) polymorphism of ACTN3 (rs1815739) and Gly482Ser (G>A) polymorphism of the PPARG1 α gene (rs192678) 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 alpha-actinin-3 protein, ACTN3, plays a crucial role in muscle contraction and force production. It helps to stabilize muscle fibers and connect them to other structural proteins within the muscle cells. This protein is essential for generating rapid and powerful muscle contractions, which are important for activities such as sprinting and power-based sports.

PPARGC1 α encodes peroxisome proliferator-activated receptor γ coactivator 1- α (PGC-1 α), which plays a central regulatory role in energy metabolism by modulating systemic oxidative metabolism and mitochondrial function and is also a susceptibility candidate gene for NAFLD (*non-alcoholic fatty liver disease*).

Athletic performance is a complex phenotype affected by individual traits, environmental conditions, training, and genetics. The peroxisome proliferator-activated receptor-alpha (PPAR α) and alpha-actinin-3 (ACTN3) are two genes with the potential to influence human performance.

CLINICAL SIGNIFICANCE

Alpha-actinin-3, also known as alpha-actinin skeletal muscle isoform 3 or F-actin cross-linking protein, is a protein that in humans is encoded by the ACTN3 gene, located on chromosome 11. The R577X SNPs polymorphism (rs1815739) in the gene affects the expression of α -actinin-3. This polymorphism, which substitutes an arginine residue at codon 577 instead of an early stop codon, resulting from the substitution of the "C" base at position 1,747 in exon 16 with a "T" base. Specifically, individuals with a 'null' 577XX (or simply 'XX') genotype are unable to express ACTN3, as opposed to individuals with the RR and RX genotypes. The XX genotype does not cause harm, as the lack of α -actinin-3 expression is compensated by the overexpression of α -actinin-2.

The genetic variant PPARGC1 α (rs192678), determines the Gly482Ser polymorphism (a G-to-A transition that predicted a glycine (G) to serine (S) substitution at amino acid position 482 in exon 8). The G allele is associated with enhanced mitochondrial efficiency, higher aerobic capacity, and a greater proportion of fatigue-resistant type I muscle fibers, benefiting endurance sports like cycling and triathlon. The A allele is linked to increased risks of metabolic conditions, including type 2 diabetes and obesity.

§ Polymorphisms of PPAR α and ACTN3. Among Adolescent Egyptian Athletes: A Case-Control Study. Life 2025, 15, 477.

§ Role of the PPARGC1 α Gene and Its rs192678 Polymorphism on Sport Performance, Aerobic Capacity, Muscle Adaptation and Metabolic Diseases: A Narrative Review Genes (Basel). 2024 Dec 20;15(12):1631.

§ A Systematic Review and Meta-analysis of the Association Between ACTN3 R577X Genotypes and Performance in Endurance Versus Power Athletes and Non-athletes. El Ouali et al. Sports Medicine - Open 3 <https://doi.org/10.1186/s40798-024-00711-x>

§ J Sports Sci. 2023 Jan;41(1):56-62. The PPARGC1 α Gly482Ser polymorphism is associated with elite long-distance running performance

§ World J Gastroenterol. 2021 Jul 7;27(25):3863-3876. PPARGC1 α rs192678 G>A polymorphism affects the severity of hepatic histological features and nonalcoholic steatohepatitis in patients with nonalcoholic fatty liver disease

§ Cells. 2019 Dec 19;9(1):12. Genetic Associations with Aging Muscle: A Systematic Review

§ ACTN3: More than Just a Gene for Speed. Front. Physiol. , 18 December 2017. Volume 8 - 2017



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DESCRIPTION	LABEL	VOLUME	STORAGE
		GEN-046-25	
Mix oligonucleotides and probes	Mix ACTN3 10X	1 x 62 μ l	-20°C
Mix oligonucleotides and probes	Mix PPARG1 α 10X	1 x 62 μ l	-20°C
Mix buffer and Taq polymerase	Mix Real-Time PCR 2X	1 x 620 μ l	-20°C
Deionized H ₂ O	Deionized H ₂ O	1 x 1 ml	-20°C
Genomic DNA or recombinant DNA Controls ACTN3 (rs1815739)	Control 1 ACTN3	1 x 22 μ l	-20°C
	Control 2 ACTN3	1 x 22 μ l	-20°C
	Control 3 ACTN3	1 x 22 μ l	-20°C
Genomic DNA or recombinant DNA Controls PPARG1 α (rs8192678)	Control 1 PPARG1 α	1 x 22 μ l	-20°C
	Control 2 PPARG1 α	1 x 22 μ l	-20°C
	Control 3 PPARG1 α	1 x 22 μ l	-20°C

TECHNICAL CHARACTERISTICS

COD. GEN-046-25

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)	$\geq$ 5 ng of genomic DNA
LIMIT OF BLANK (LOB)	>40 Cq
REPRODUCIBILITY	99,9%
SPECIFICITY / SENSITIVITY	99,9%/98%