



GENETIC VARIATIONS IN NON-ALCOHOLIC FATTY LIVER DISEASE (NAFLD) PANEL 2

ORDERING INFORMATION

REF: GEN-061-25
Tests: 25 Reactions: 31 x 3
REF: GEN-061-50
Tests: 50 Reactions: 62 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 medical devices **REAL-TIME QUALITATIVE PCR-GENETIC VARIANTS-RESEARCH USE ONLY**. Detection of the genetic variants rs13412852 (LPIN1), rs3750861 (KLF6) and rs12603226 (ZNF624) by Real-Time PCR technique. Kit optimized for Real-time PCR instrumentation Biorad CFX96 Dx, Biorad Opus Dx, Agilent AriaDx, Thermofisher QuantStudio™ 5 Real-Time PCR System.

SCIENTIFIC BACKGROUND

There is significant heterogeneity in the clinical phenotype and natural history of non-alcoholic fatty liver disease (NAFLD), now referred to as metabolic and fatty liver disease (MASLD), as it is a multifactorial condition resulting from the interaction between genetic background and environmental factors.

In addition to the genetic variants PNPLA3 rs738409 C>G, TM6SF2 rs58542926 C>T and MBOAT7 rs641738, there are other genes involved in the clinical picture of MASLD/NAFLD:

LPIN1: is a Mg(2+)-dependent phosphatidic acid (PA) phospholipase that catalyses the dephosphorylation of PA to produce diacylglycerol and inorganic phosphate.

KLF6: is a transcription factor that modulates trophoblast migration, differentiation and the production of reactive oxygen species (ROS). Hepatocyte expression of KLF6 regulates hepatic metabolism of fatty acids and glucose through the transcriptional activation of hepatic glucokinase and the post-transcriptional regulation of PPARα

ZNF624: Zinc-finger proteins (ZNFs) are one of the most abundant protein groups and have a wide range of molecular functions. Given the wide variety of zinc-finger domains, ZNFs are capable of interacting with DNA, RNA, PAR (poly-ADP-ribose) and other proteins. Consequently, ZNFs are involved in the regulation of various cellular processes.

CLINICAL SIGNIFICANCE

LPIN1: LPIN1 SNPs and haplotypes have been associated with various components of metabolic syndrome. The expression of the LPIN1 rs13412852 variant is age-dependent; that is, the phenotype is more or less pronounced or involves different traits during developmental years. Indeed, the LPIN1 rs13412852 variant has been associated with lipid levels, NASH severity and liver fibrosis in children with NAFLD. Conversely, it influences body mass but not the severity of liver histology in adults with NAFLD of the same ethnicity. Furthermore, it is plausible that LPIN1 rs13412852 reduces fatty acid oxidation and increases lipogenesis in a sedentary lifestyle, thereby promoting the development of steatosis.

(IVS) 1-27 G>A of the **KLF6** gene (rs3750861) is a common single-nucleotide polymorphism in intron 1 at position 3023 (-27 upstream of exon 2) of the KLF6 gene, which abolishes wild-type SF2/ASF and SRp55 binding sites and generates a functional SRp40 binding site, thereby increasing alternative splicing. This polymorphism increases the generation of alternative splicing forms that antagonise KLF6 and protects patients from advanced NAFLD.

The A>T rs12603226 polymorphism in the **ZNF624** gene has been associated with an increased risk of developing MASLD/NAFLD. ZNF624 rs12603226 C>T

§ Int J Mol Sci. 2026 Feb 8;27(4):1644. The Interplay of Genetics and Lifestyle in MASLD: Focus on LPIN1 rs13412852 and Sedentary Behaviour.

§ Nat Metab 2025 Nov;7(11):2199-2211. Human genetics of steatotic liver disease: insights into insulin resistance and lipid metabolism.



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DESCRIPTION	LABEL	VOLUME		STORAGE
		GEN-061-25	GEN-061-50	
Mix oligonucleotides and probes	Mix LPIN1 10X	1 x 62 µl	2 x 62 µl	-20°C
Mix oligonucleotides and probes	Mix KLF6 10X	1 x 62 µl	2 x 62 µl	-20°C
Mix oligonucleotides and probes	Mix ZNF624 10X	1 x 62 µl	2 x 62 µl	-20°C
Mix buffer and Taq polymerase enzyme	Mix Real-Time PCR 2X	1 x 930 µl	2 x 930 µl	-20°C
Deionized H ₂ O	Deionized H ₂ O	1 x 1 ml	1 x 1 ml	-20°C
Genomic DNA or recombinant DNA	Control 1	1 x 45 µl	2 x 45 µl	-20°C
Genomic DNA or recombinant DNA	Control 2	1 x 45 µl	2 x 45 µl	-20°C

TECHNICAL CHARACTERISTICS

COD. GEN-061-25 / GEN-061-50

STABILITY	18 months
REAGENTS STATUS	Ready to use
BIOLOGICAL MATRIX	Genomic DNA extracted from whole blood, tissues, cells
POSITIVE CONTROLS	Recombinant DNA for at least 3 analytical sessions (GEN-061-25) Recombinant DNA for at least 6 analytical sessions (GEN-061-50)
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%