

GENETIC VARIANTS IN NON-ALCOHOLIC FATTY LIVER DISEASE (NAFLD)

ORDERING INFORMATION

REF: GEN-054-25
Tests: 25 Reactions: 31 x 3
REF: GEN-054-50
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 genetic variants of the genes PNPLA3 (rs738409), TM6SF2 (rs58542926), MBOAT7/TMC4 (rs641738) C>T 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

The genes involved in the NAFLD clinical picture are: **PNPLA3**, a member of the potato-like phospholipase family. It is involved in the synthesis and remodeling of cellular lipids, converting lysophosphatidic acid into phosphatidic acid. The PNPLA3 rs738409 C>G (I148M) polymorphism involves a substitution of isoleucine with methionine at codon 148. **TM6SF2**, the rs58542926 C > T variant in the gene shows a substitution of glutamate with lysine at locus 167 (E167 K) which increases the level of intrahepatic TAG and causes the liver to retain very low-density lipoproteins (VLDL). **MBOAT7** catalyzes the transfer of an acyl-CoA to lysophosphatidylinositol (lysoPI) and has a preference for arachidonyl-CoA. Therefore, it may be one of the mechanisms through which the levels of pro-inflammatory free arachidonic acid and eicosanoids are regulated.

§ Review Liver Int. 2025 Apr;45(4):e70051. PNPLA3 I148M and Hepatocellular Carcinoma
§ Review Metab Brain Dis. 2025 Jul 10;40(6):240. From nonalcoholic fatty liver disease to neuroinflammation: the role of chronic systemic inflammation.
§ Meta-Analysis Medicine (Baltimore). 2023 Mar 10;102(10):e33110. Patatin-like phospholipase domain-containing 3 gene (PNPLA3) polymorphic (rs738409) single nucleotide polymorphisms and susceptibility to nonalcoholic fatty liver disease: A meta-analysis of twenty studies
§ Front Nutr. 2023 Mar 2;10:1101341. The I148M PNPLA3 variant mitigates niacin beneficial effects: How the genetic screening in non-alcoholic fatty liver disease patients gains value.
§ Meta-Analysis Horm Metab Res. 2022 Oct;54(10):686-695. Associations of PNPLA3 rs738409 Polymorphism with Plasma Lipid Levels: A Systematic Review and Meta-Analysis.
§ Cell Mol Gastroenterol Hepatol. 2022;13(3):759-788. TM6SF2/PNPLA3/MBOAT7 Loss-of-Function Genetic Variants Impact on NAFLD Development and Progression Both in Patients and in In Vitro Models.
§ Front Med (Lausanne). 2021 Oct 8;8:734847. PNPLA3, TM6SF2, and MBOAT7 Influence on Nutraceutical Therapy Response for Non-alcoholic Fatty Liver Disease: A Randomized Controlled Trial.
§ Nat Commun. 2016 Sep 15;7:12757. MBOAT7 rs641738 increases risk of liver inflammation and transition to fibrosis in chronic hepatitis C.

CLINICAL SIGNIFICANCE

A meta-analysis study on the PNPLA3 rs738409 C>G polymorphism (Horm Metab Res. 2022 Oct;54(10):686-695) showed that people with the CC genotype are 52% less likely to develop non-alcoholic fatty liver disease, and people with the CG genotype are 19% more likely to develop non-alcoholic fatty liver disease. Furthermore, the population with the GG genotype is 105% more likely to develop non-alcoholic fatty liver disease. Furthermore, in individuals with MASLD, the G allele of PNPLA3 rs738409 was associated with lower circulating levels of adiponectin, a biomarker with anti-inflammatory, antifibrotic, and anticarcinogenic properties.

The CG and GG genotypes of the PNPLA3 rs738409 C>G polymorphism and the CT and TT genotypes of the TM6SF2 rs58542926 polymorphism are associated with greater hepatic fat accumulation and an increased risk of fibrosis, cirrhosis, and hepatocellular carcinoma (HCC) compared to wild-type patients. Similar results derived from the study of the MBOAT7 rs641738 (C/T-T/T) genetic variant have been shown to be linked to the risk of NAFLD worsening in a large cohort study due to its influence on the remodeling of the hepatic phosphatidylinositol acyl chain.



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DESCRIPTION	LABEL	VOLUME		STORAGE
		GEN-054-25	GEN-054-50	
Mix oligonucleotides and probes	Mix PNPLA3 10X	1 x 62 µl	2 x 62 µl	-20°C
Mix oligonucleotides and probes	Mix TM6SF2 10X	1 x 62 µl	2 x 62 µl	-20°C
Mix oligonucleotides and probes	Mix MBOAT7 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-054-25 / GEN-054-50

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-054-25) Recombinant DNA for at least 6 analytical sessions (GEN-054-50)
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%