NAT2 GENETIC VARIANTS (N-ACETYLTRANSFERASE 2)

RUO
Research Use Only

ORDERING INFORMATIONS

REF: GEN-055-25
Tests: 25 Reactions: 31 x 5
REF: GEN-055-50
Tests: 25 Reactions: 62 x 5
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 gene variants rs1208, rs1801279, rs1801280, rs1799930, rs1799931 of the NAT2 (N-acetyltransferase 2) 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

Genetic differences in the enzyme N-acetyltransferase 2 (NAT2), encoded by the NAT2 gene, play a significant role in determining the metabolic clearance of isoniazid (INH), which is a key drug in the treatment of tuberculosis. Individuals are classified according to their NAT2 genotype into three acetylation phenotypes, which influence levels of exposure to isoniazid: slow acetylator (SA), intermediate acetylator (IA), and rapid acetylator (RA). Data in the literature have shown that SA subjects, who have greater exposure to isoniazid, are at greater risk of drug-induced hepatotoxicity. On the other hand, RA subjects are more likely to experience acquired drug resistance and relapses due to inadequate drug exposure. Considering that these suboptimal outcomes may contribute to mortality, it is plausible that the NAT2 genotype may also influence patient mortality.

Two metabolites of isoniazid, hydrazine (Hz) and acetyl hydrazine (AcHz), are toxic to hepatocytes and appear to be responsible for isoniazid-induced liver damage. Acetylation of Hz and AcHz by N-acetyltransferase 2 (NAT2) renders these compounds non-toxic.

§ Int J Infect Dis. 2025 Jun: 155:107895. One-year mortality of tuberculosis patients on isoniazid-based treatment and its association with rapid acetylator NAT2 genotypes.
§ Review Pharmacogenomics. 2025 Apr-Apr;26(5-6):207-221. Genetic polymorphisms and adverse reactions to antituberculosis therapy.
§ J Infect Dis. 2025 Apr 17. Impact of pharmacogenetics on pharmacokinetics of first-line anti-tuberculosis drugs in the HIRIF trial.
§ Observational Study Int J Mol Sci. 2025 Apr 19;26(8):3881. NAT2 Acetylation Status Predicts Hepatotoxicity During Antituberculosis Therapy: Cumulative Risk Analysis of a Multiethnic Cohort.
§ Observational Study Ann Med. 2025 Dec;57(1):2478316. Pharmacogenomic heterogeneity of N-acetyltransferase 2: a comprehensive analysis of real world data in Indian tuberculosis patients and from literature and database review.
§ Review Pharmacogenet Genomics. 2014 Aug;24(8):409-25. PharmGKB summary: very important pharmacogene information for N-acetyltransferase 2.
§ Pharmacogenomics. 2012 Jan;13(1):31-41. Accuracy of various human NAT2 SNP genotyping panels to infer rapid, intermediate and slow acetylator phenotypes

CLINICAL SIGNIFICANCE

The most important alleles of NAT2 loss of activity are *5 (rs1801280, 341 T>C), *6 (rs1799930, 590 G>A), *7 (rs1799931, 857 G>A) and *14 (rs1801279, 191 G>A), while the G allele of the rs1208 G>A, p.R268K variant is the reference rapid acetylator allele with the A allele, NAT2*4, showing reduced activity.

The presence of two of these “loss-of-function” alleles confers a slow acetylator phenotype and, in multiple studies, an approximately twofold increased risk of isoniazid hepatotoxicity.

An individual with 2 copies of the rs1208 G allele variant or 1 copy of the rs1208 G allele variant and 1 copy of NAT2*1 (NG_012246.1 RefSeq, NAT2*12) can be considered a rapid acetylator (RA); Individuals with 2 copies of the NAT2 *5 or NAT2 *6 or NAT2 *7 or NAT2 *14 allele or double heterozygotes for these alleles can be considered slow acetylators. (SA)

Furthermore, for SNP rs1208, allele A, NAT2*4 is associated with traits typical of insulin resistance, such as increased fasting blood sugar, glycated hemoglobin, total cholesterol and LDL, triglycerides, and coronary artery disease.

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ISO 9001 :2015
ISO 13485:2021

ED.1 REV.1 of 22/10/2025

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DESCRIPTION	LABEL	VOLUME		STORAGE
		GEN-055-25	GEN-055-50	
Mix oligonucleotides and probes	Mix NAT2 *4 10X	1 x 77,5 µl	2 x 77,5 µl	-20°C
Mix oligonucleotides and probes	Mix NAT2 *14 10X	1 x 77,5 µl	2 x 77,5 µl	-20°C
Mix oligonucleotides and probes	Mix NAT2 *5 10X	1 x 77,5 µl	2 x 77,5 µl	-20°C
Mix oligonucleotides and probes	Mix NAT2 *6 10X	1 x 77,5 µl	2 x 77,5 µl	-20°C
Mix oligonucleotides and probes	Mix NAT2 *7 10X	1 x 77,5 µl	2 x 77,5 µl	-20°C
Mix buffer and Taq polymerase enzyme	Mix Real-Time PCR 2X	2 x 969 µl	4 x 969 µl	-20°C
H_20 deionizzata	Deionized H_20	1 x 1,1 ml	2 x 1,1 ml	-20°C
Genomic DNA or recombinant DNA Control NAT2 *4	Control 1, Control 2	1 x 22 µl	2 x 22 µl	-20°C
Genomic DNA or recombinant DNA Control NAT2 *14	Control 1, Control 2	1 x 22 µl	2 x 22 µl	-20°C
Genomic DNA or recombinant DNA Control NAT2 *5	Control 1, Control 2	1 x 22 µl	2 x 22 µl	-20°C
Genomic DNA or recombinant DNA Control NAT2 *6	Control 1, Control 2	1 x 22 µl	2 x 22 µl	-20°C
Genomic DNA or recombinant DNA Control NAT2 *7	Control 1, Control 2	1 x 22 µl	2 x 22 µl	-20°C

TECHNICAL CHARACTERISTICS

COD. GEN-055-25 / GEN-055-50

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-055-25), Recombinant DNA for at least 6 analytical sessions (GEN-055-50),
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%

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