

ADH1B and ADH1C GENETIC VARIANTS (ALCOHOL METABOLISM)

ORDERING INFORMATION

REF: GEN-051-25
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 medical devices **REAL-TIME QUALITATIVE PCR-GENETIC VARIANTS-RESEARCH USE ONLY**. Detection of the G>A (rs1229984) polymorphism of the ADH1B gene and of the C>T (rs1693482) of the ADH1C gene 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

Alcohol metabolizing enzyme (AME) genes, alcohol dehydrogenases (ADH1B and ADH1C) and aldehyde dehydrogenase (ALDH2), are genetic factors associated with the Alcohol use disorder (AUD) outcome.

Alcohol dehydrogenases (ADH) are coded for by seven genes namely ADH1A, ADH1B, ADH1C, ADH4, ADH5, ADH6, and ADH7 located on human chromosome 4. The seven ADH genes have been categorized into five classes (I–V). Class I ADH genes (ADH1A, ADH1B, and ADH1C) encode three subunits denoted α , β , and γ respectively. The functional enzyme consists of homo- and heterodimers of alpha, beta, and gamma subunits. These isoenzymes metabolize alcohol into acetaldehyde (among other physiological actions); acetaldehyde is then metabolized into acetate through the aldehyde dehydrogenase 2 gene (ALDH2).

CLINICAL SIGNIFICANCE

Alcohol dehydrogenase 1B (ADH1B) is a main enzyme that metabolizes exogenous ethanol. A nucleotide substitution in the polymorphic locus rs1229984 of the ADH1B gene determines two alleles, which are responsible for differences in ADH1B activity and acetaldehyde production rate. The allele ADH1B*48Arg (rs1229984-G) is common in Caucasians, while the allele ADH1B*48His (rs1229984-A), which determines higher enzyme activity and acetaldehyde production to higher concentrations, is more widespread in Eastern and Southeastern Asia.

The *wild type* ADH1C*1 (γ 1 subunit) contains arginine at 272nd and isoleucine at 350th position, while the ADH1C *2 rs1693482/ rs698 variant, which encodes the γ 2 subunit, contains glutamine (rs1693482) and valine (rs698), respectively. These two SNPs are in high linkage disequilibrium. If the test reveals a predisposition to alcohol sensitivity (ADH1C*1 and ADH1B*48His), it means that the body produces acetaldehyde at a rate higher than its ability to metabolize it. This leads to a greater likelihood of experiencing adverse symptoms (facial flushing, headache, nausea, tachycardia, sweating) and, in the long term, an increased risk of liver damage, hypertension, and cardiovascular problems. Individuals with the variants (ADH1C*2/*2 and ADH1B*48Arg) metabolize alcohol more slowly, which may increase the risk of dependence and liver damage with prolonged consumption.

§ Cancer Epidemiol Biomarkers Prev. 2025 May 2;34(5):744-753. ALDH2 Deficiency and Alcohol Intake in the United States: Opportunity for Precision Cancer Prevention.
§ Cancer Med. 2023 Jan;12(1):747-759. Impacts of ADH1B rs1229984 and ALDH2 rs671 polymorphisms on risks of alcohol-related disorder and cancer
§ Am J Med Genet B Neuropsychiatr Genet. 2018 Mar;177(2):113-125. ADH1B: From alcoholism, natural selection, and cancer to the human phenotype
§ Hum Genet. 2012 Aug;131(8):1361-74. Further clarification of the contribution of the ADH1C gene to vulnerability of alcoholism and selected liver diseases

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DESCRIPTION	LABEL	VOLUME	STORAGE
		GEN-051-25	
Mix oligonucleotides and probes	Mix ADH1B rs1229984 10X	1 x 62 µl	-20°C
Mix oligonucleotides and probes	Mix ADH1C rs1693482 10X	1 x 62 µl	-20°C
Mix buffer and Taq polymerase enzyme	Mix Real-Time PCR 5X	1 x 248 µl	-20°C
Deionized H ₂ O	Deionized H ₂ O	1 x 1 ml	-20°C
Genomic DNA or recombinant DNA ADH1B	Control 1, Control 2, Control 3	1 x 22 µl	-20°C
Genomic DNA or recombinant DNA ADH1C	Control 1, Control 2, Control 3	1 x 22 µl	-20°C

TECHNICAL CHARACTERISTICS

COD. GEN-051-25

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
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) + 64°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%