

ALDH2 GENETIC VARIANT (ALCOHOL METABOLISM)

ORDERING INFORMATION

REF: GEN-056-25
Tests: 25 Reactions: 31
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 variant G>A (rs671) variant of the ALDH2 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).

ALDH2 encodes for the second enzyme in the main metabolic pathway that breaks down alcohol. Alcohol, a known carcinogen, is first metabolized into acetaldehyde by alcohol dehydrogenase. Then, acetaldehyde, also a known carcinogen, is metabolized into acetate by ALDH2.

§ 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

§ Nat Commun. 2024 Mar 22;15(1):2594. The aldehyde dehydrogenase 2 rs671 variant enhances amyloid β pathology.

§ Cancer Med. 2023 Jan;12(1):747-759. Impacts of ADH1B rs1229984 and ALDH2 rs671 polymorphisms on risks of alcohol-related disorder and cancer

§ Cancer Sci. 2022 Apr;113(4):1441-1450. doi: 10.1111/cas.15286. Epub 2022 Feb 16. New insights into the genetic contribution of ALDH2 rs671 in pancreatic carcinogenesis: Evaluation by mediation analysis

§ Acta Pharm Sin B. 2021 Jun;11(6):1400-1411. doi: 10.1016/j.apsb.2021.02.008. Epub 2021 Feb 11. The role of ALDH2 in tumorigenesis and tumor progression: Targeting ALDH2 as a potential cancer treatment

§ BMC Cancer. 2021 May 22;21(1):584. ALDH2 polymorphism rs671 is a predictor of PD-1/PD-L1 inhibitor efficacy against thoracic malignancies

CLINICAL SIGNIFICANCE

The rs671 variant (G>A, Glu504Lys), denoted ALDH2*2, is a missense mutation which causes a change from Glu to Lys (E to K) at position 504 of the ALDH2 protein (NP_000681.2: p. Glu504Lys), leading to a substantial decrease in dehydrogenase activity.

In homozygous (ALDH2*2/*2) and heterozygous (ALDH2*1/*2) individuals, ALDH2 enzyme activity is nearly 0% and <20% of the normal activity level, respectively; thus, ALDH2*2 carriers have ALDH2 deficiency, or decreased ability to detoxify and metabolize aldehydes, including acetaldehyde. This leads to acetaldehyde accumulation in the bloodstream after alcohol intake as well as unpleasant symptomatic responses which may include facial flushing, headache, and palpitations, especially in homozygotes, and increased cancer risk.

The inactive ALDH2 rs671 or low ALDH2 expression causes the accumulation of aldehydic products such as acetaldehyde, 4-hydroxy-2-nonenal (4-HNE), and malondialdehyde (MDA), which are associated also with high morbidity of cancer.

In addition, there are data supporting the association between alcohol consumption and pancreatic cancer risk and indicates the potential contribution of the rs671 A allele to pancreatic carcinogenesis through impaired metabolism of known or unknown ALDH2 substrates.

These findings suggest a significant opportunity for precision cancer prevention.

ALDH2 GENETIC VARIANT (ALCOHOL METABOLISM)

ORDERING INFORMATIONS

REF: GEN-056-25
Tests: 25 Reactions: 31
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



CONTENTS OF THE KIT

DESCRIPTION	LABEL	VOLUME	STORAGE
		GEN-056-25	
Mix oligonucleotides and probes	Mix ALDH2 (rs671) 10X	1 x 62 µl	-20°C
Mix buffer and Taq polymerase enzyme	Mix Real-Time PCR 5X	1 x 124 µl	-20°C
Deionized H ₂ O	Deionized H ₂ O	1 x 1 ml	-20°C
Genomic DNA or recombinant DNA	Control 1	1 x 22 µl	-20°C
Genomic DNA or recombinant DNA	Control 2	1 x 22 µl	-20°C

TECHNICAL CHARACTERISTICS

COD. GEN-056-25

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%