



ACE I/D (Angiotensin-Converting Enzyme INS/DEL) POLYMORPHISM

ORDERING INFORMATION

REF: GEN-035-25 RDM Code: 2159760/R
Tests: 25 Reactions: 31
REF: GEN-035-50 RDM Code: 2165040/R
Tests: 50 Reactions: 62
CND Code: W0106010499
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

For in vitro diagnostic use



PRODUCT CHARACTERISTICS

Device belonging to the family of in vitro medical devices **REAL-TIME QUALITATIVE PCR -GENETIC VARIANTS**. Detection of the nucleotide polymorphism insertion (allele I) or deletion (allele D) in intron 16 of the gene encoding the human angiotensin converting enzyme (ACE) by Real-Time PCR technique. Optimized kit for Real-Time PCR instrumentation Biorad CFX96 Dx, Biorad Opus Dx, Agilent AriaDx, Hyris bCUBE and Hyris bCUBE3 with Hyris bAPP.

SCIENTIFIC BACKGROUND

The renin-angiotensin-aldosterone system (SRAA) is a hormonal system that regulates blood pressure, circulating plasma volume, arterial muscle tone through different mechanisms and the secretion of aldosterone; it also plays an important role in the etiology of hypertension. There are numerous components of this system: renin, prorenin, angiotensin converting enzyme (ACE), angiotensinogen (AGT), angiotensin I and angiotensin II; the latter represents the final effector of the renin-angiotensin system and exerts its effects on the cardiovascular system through binding to specific receptors. The first stage of the enzymatic cascade that leads to the production of angiotensin II consists in the conversion of angiotensinogen to angiotensin I, by the proteolytic enzyme renin. The second stage of the process involves the conversion of angiotensin I to angiotensin II, through a reaction catalyzed by ACE. Angiotensin II is the main active peptide of SRAA that functions through at least four types of receptors. The AGTR1 receptor mediates cardiovascular effects, including vasoconstriction, aldosterone synthesis, vasopressin secretion, vascular smooth muscle cell proliferation, renal blood flow, regulation of renin activity, renal sodium absorption, the modulation of the activity of the sympathetic nervous system, and cardiac function.

§ J Clin Med Res. 2024 Aug;16(7-8):355-362 Renin-Angiotensin System Genes Polymorphisms in Patients With COVID-19 and Its Relation to Severe Cases of SARS-CoV-2 Infection

§ Pediatr Res. 2024 Jan 4. doi: 10.1039/s41390-023-02982-8. Online ahead of print. Association of ACE1 I/D polymorphism and susceptibility to COVID-19 in Egyptian children and adolescents

§ Association of angiotensin-converting enzyme gene I/D polymorphism with chronic obstructive pulmonary disease: a meta-analysis. J Renin Angiotensin Aldosterone Syst. 2018 Apr-Jun;19(2):1470320318770546.

§ Genetic polymorphism of angiotensin-converting enzyme and hypertrophic cardiomyopathy risk: A systematic review and meta-analysis. Medicine (Baltimore). 2017 Dec;96(48):e8639.

§ Ace Gene Plays A Key Role In Reducing Blood Pressure In Hypertensive Elderlies After Resistance Training Resistance Exercise And Ace Polymorphism. J Strength Cond Res. 2017 Dec 1.

§ Association of insertion-deletions polymorphisms with colorectal cancer risk and clinical features. World J Gastroenterol. 2017 Oct 7;23(37):6854-6867.

CLINICAL SIGNIFICANCE

The renin-angiotensin system (SRAA) also exerts local effects on cell proliferation, apoptosis, inflammation and angiogenesis in different tissues. Furthermore, there are data in the literature correlating SRAA with tumor tumorigenesis and tumor angiogenesis. There are genetic polymorphisms in the various components of the SRAA that may have clinical relevance. The insertion/deletion (I/D) of the ACE gene is directly associated with the circulatory level of the enzyme itself. The nature of the ACE1 rs1799752 gene polymorphism is in the insertion (insertion, I) or loss (deletion, D) of the Alu repeat in 289 nucleotide pairs in the 16th intron. Deletion of the Alu repeat is accompanied by a significant increase in the expression of the ACE1 gene and a rise of ACE1 levels. The increase in ACE1 levels occurs even in case of heterozygous status (I/D). The highest level is observed in patients with homozygous genotype D/D rs1799752, which is twice higher than in patients with genotype I/I. The relation between the D/D genotype and a wide range of CVDs, including coronary heart disease (CHD), heart attack, left ventricular hypertrophy, hypertension, kidney disease, and neurodegenerative diseases has been detected. Furthermore, in a recent meta-analysis a significant association between I/D polymorphisms and recurrent miscarriages was reported. Women with the "DD" or "ID" ACE genotypes are at higher risk of experiencing recurrent pregnancy loss.



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DESCRIPTION	LABEL	VOLUME		STORAGE
		GEN-035-25	GEN-035-50	
Mix oligonucleotides and probes	Mix PCR ACE I/D 10X	1 x 77,5 µl	2 x 77,5 µl	-20°C
Mix buffer and Taq polymerase enzyme	Mix Real-Time PCR 2X	1 x 387,5 µl	2 x 387,5 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 HOMO DD	1 x 22 µl	2 x 22 µl	-20°C
Genomic DNA or recombinant DNA	Control 2 HET ID	1 x 22 µl	2 x 22 µl	-20°C
Genomic DNA or recombinant DNA	Control 3 HOMO II	1 x 22 µl	2 x 22 µl	-20°C

TECHNICAL CHARACTERISTICS

COD. GEN-035-25 / COD. GEN-035-50

STABILITY	18 months
REAGENTS STATUS	Ready to use
BIOLOGICAL MATRIX	Genomic DNA extracted from whole blood, tissue, cells
CONTROLS	Recombinant DNA for at least 3 analytical sessions (GEN-035-25) Recombinant DNA for at least 6 analytical sessions (GEN-035-50)
VALIDATED INSTRUMENTS	Biorad CFX96 Dx, Biorad Opus Dx, Agilent AriaDx, Hyris bCUBE and Hyris bCUBE3 with Hyris bAPP.
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 at (60 sec)
ANALYTICAL SPECIFICITY	Absence of non-specific pairings of oligonucleotides and probes; absence of cross-reactivity
LIMIT OF DETECTION (LOD)	≥ 0,016 ng of genomic DNA
LIMIT OF BLANK (LOB)	0% NCN
REPRODUCIBILITY	99,9%
DIAGNOSTIC SPECIFICITY / DIAGNOSTIC SENSITIVITY	100%/98%