MTHFR A1298C POLYMORPHISM

For in vitro diagnostic use

CE IVD

ORDERING INFORMATIONS

REF: GEN-004-25 RDM Code: 1718917/R
Tests: 25 Reactions: 31
REF: GEN-004-50 RDM Code: 2255480/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*

PRODUCT CHARACTERISTICS

Determination of the A1298C polymorphism of the MTHFR gene 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 MTHFR enzyme gene (5, 10-methylenetetrahydrofolate reductase) is located at the end of the short arm of chromosome 1 (1p36.3). The DNA sequence of the gene is approximately 2.2 kilobases (kb), comprising 11 exons. Two polymorphisms have been described in detail for the MTHFR gene: C677T (rs1801133) and A1298C (rs1801131). The C677T polymorphism is located in exon 4 and results in a conversion of alanine to valine at codon 222 (A222V) in a protein region that is the binding site for the cofactor of MTHFR, flavin adenine dinucleotide (FAD). It is reported in the literature that the MTHFR 677T genotype decreases MTHFR enzyme activity by 30% in vitro compared to the wild-type type. Folate is one of the most important precursor substrates for cellular metabolism. One of folate's jobs is to act as a carrier of individual carbon fragments. This reaction is required for the synthesis of purine-pyrimidines, DNA, RNA and protein methylation. Previous research has shown that low folate levels result in uracil disincorporation during DNA replication, which causes increased double-strand breaks during uracil remnant excision repair.

CLINICAL SIGNIFICANCE

The second polymorphism of the MTHFR gene is A1298C, located in exon 7 and resulting in a substitution of a glutamic acid residue to alanine at codon 429 (E429A). This polymorphism is located in the regulatory domain of the enzyme S-adenosyle methionine (SAM) and causes conformational changes within the MTHFR enzyme that alter its enzymatic activity. Folate deficiency, therefore, has also been associated with an increased risk for a number of cancers and other disease risks such as cardiovascular disease, diabetes, birth defects, ischemia, venous thrombosis, hypotonia, leukemia, migraine, schizophrenia, depression, , preeclampsia, Alzheimer's disease, birth defects of the heart, Down syndrome and cleft palate.

§ Two Common MTHFR Gene Polymorphisms (C677T and A1298C) and Fetal Congenital Heart Disease Risk: An Updated Meta-Analysis with Trial Sequential Analysis. Cell Physiol Biochem. 2018 Mar 15; 45 (6):2483-2496.

§ The methylenetetrahydrofolate reductase 677T-1298C haplotype is a risk factor for acute lymphoblastic leukemia in children. Medicine (Baltimore). 2017 Dec; 96 (51):e9290.

§ Folate metabolism genetic polymorphisms and meningioma and glioma susceptibility in adults. Oncotarget. 2017 Jul 4; 8 (34):57265-57277.

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ISO 9001:2015
ISO 13485:2016

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DESCRIPTION	LABEL	VOLUME		STORAGE
		GEN-004-25	GEN-004-50	
Mix oligonucleotides and probes	Mix A1298C MTHFR 10X	1 x 85 µl	1 x 170 µl	-20°C
Mix buffer and Taq polymerase enzyme	Mix Real-Time PCR 2X	1 x 425 µl	1 x 850 µl	-20°C
Deionized H_20	Deionized H_20	2 x 1 ml	2 x 1 ml	-20°C
Genomic DNA or recombinant DNA	Control +1	1 x 22 µl	1 x 22 µl	-20°C
Genomic DNA or recombinant DNA	Control +2	1 x 22 µl	1 x 22 µl	-20°C
Genomic DNA or recombinant DNA	Control +3	1 x 22 µl	1 x 22 µl	-20°C

TECHNICAL CHARACTERISTICS

COD. GEN-004-25 / COD. GEN-004-50

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

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