



MTR A2756G POLYMORPHISM

ORDERING INFORMATION

REF: GEN-036-25 RDM Code: 2248813/R
Tests: 25 Reactions: 31
REF: GEN-036-50 RDM Code: 2248811/R
Tests: 50 Reactions: 62
CND Code: W0106010499
Manufacturer: BioMol Laboratories s.r.l.

For in vitro diagnostic use



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**.
Detection of A2756G polymorphism of the MTR gene (rs1805087) by Real-Time PCR technique. Kit optimized for Real-Time PCR instrumentation Biorad CFX96, Biorad Opus Dx, Agilent AriaDx.

SCIENTIFIC BACKGROUND

The MTR gene is mapped to chromosome 1q43 and the extensively studied A2756G (rs1805087) polymorphism leads to a change of the amino acid aspartate to the amino acid glycine at codon 919 (D919G), with consequent reduction of the enzymatic activity. It has been reported that this polymorphism can increase homocysteine levels by suppressing methionine metabolism and consequently can lead to DNA hypomethylation and promote tumorigenesis. Numerous studies have shown that the MTR A2756G polymorphism is linked to various types of cancer, such as prostate cancer, retinoblastoma, acute lymphoblastic leukemia (ALL), and some cases of autism. In fact, the heterozygous AG and homozygous GG genotypes are associated with an increased risk of these pathologies.

CLINICAL SIGNIFICANCE

Methionine synthase (MTR) plays a crucial role in the folate metabolic network. It is a vitamin B12-dependent enzyme that remethylates homocysteine to methionine with simultaneous conversion of 5-methyl-tetrahydrofolate (5-methyl-THF) to tetrahydrofolate (THF). THF is essential for the synthesis of nucleotides while methionine is essential for the synthesis of S adenosil methionine (SAM). MTR helps maintain adequate intracellular folate levels and normal concentrations of homocysteine and methionine, which are used for proper DNA methylation or other methylation processes. However, some other studies have revealed a modest inverse association between the GG genotype (A2756G MTR) and HCY levels indicating increased enzymatic activity of the variant genotype.

§ Am J Med Sci. 2024 Nov 26; S0002-9629(24)01526-X. Methionine synthetase A2756G and Cystathionine-β-synthase 844ins68 polymorphisms and coronary artery disease: A meta-analysis

§ Sci Rep. 2020 Aug 7;10(1):13384. doi: 10.1038/s41598-020-70223-7. Update analysis on the association between Methionine synthase rs1805087 A/G variant and risk of prostate cancer

§ Li-Min Ma, Hai-Ping Yang, Xue-Wen Yang and Lin-Hai Ruan. Methionine synthase A2756G polymorphism influences pediatric acute lymphoblastic leukemia risk: a meta-analysis. Bioscience Reports (2019) 39 BSR20181770 <https://doi.org/10.1042/BSR20181770>

§ Xiaosong Zhang, Jilei Tang, Nan Shen, Kewei Ren, Single-nucleotide polymorphism (rs1805087) in the methioninesynthase (MTH) gene increases the risk of prostate cancer. AGING 2018, Vol. 10, No. 10

§ Du B, Tian H, Tian D, Zhang C, Wang W, Wang L, et al. Genetic polymorphisms of key enzymes in folate metabolism affect the efficacy of folate therapy in patients with hyperhomocysteinaemia. Br J Nutr. 2018; 119(8): 887-895

§ Rosa Haghighi, Farhad Mashayekhi, Elham Bidabadi, and Zivar Salehi. Analysis of methionine synthase (rs1805087) gene polymorphism in autism patients in Northern Iran Acta Neurobiol Exp 2016; 76: 318-323

§ Kurzwski M, Wajda A, Malinowski D, Kazienko A, Kurzawa R, Drozdziak M. Association study of folate-related enzymes (MTHFR, MTR, MTRR) genetic variants with non-obstructive male infertility in a Polish population. Genet Mol Biol. 2015; 38(1): 42-47



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DESCRIPTION	LABEL	VOLUME		STORAGE
		GEN-036-25	GEN-036-50	
Mix oligonucleotides and probes	Mix A2756G MTR 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	1 x 22 µl	2 x 22 µl	-20°C
Genomic DNA or recombinant DNA	Control 2	1 x 22 µl	2 x 22 µl	-20°C
Genomic DNA or recombinant DNA	Control 3	1 x 22 µl	2 x 22 µl	-20°C

TECHNICAL CHARACTERISTICS

COD. GEN-036-25 / COD. GEN-036-50

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-036-25), Recombinant DNA for at least 6 analytical sessions (GEN-036-50)
VALIDATED INSTRUMENTS	Biorad CFX96 Dx, Biorad Opus Dx and Agilent AriaDx
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
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%