MTRR A66G POLYMORPHISM

For in vitro diagnostic use

CE IVD

ORDERING INFORMATIONS

REF: GEN-027-25 RDM Code: 2257737/R
Tests: 25 Reactions: 31
REF: GEN-027-50 RDM Code: 2159830/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

Detection of A66G polymorphism of the MTRR gene by Real-Time PCR technique. Optimized kit for Real-Time PCR instrumentation Biorad CFX96 Dx, Biorad Opus Dx, Agilent AriaDx.

SCIENTIFIC BACKGROUND

This MTRR 66A>G polymorphism would also appear to be involved in the conversion of homocysteine into methionine, which negatively affects enzyme activity and is therefore considered a genetic risk factor for hyperhomocysteinemia (HHcy). MTRR A66G can also induce DNA hypomethylation by regulating homocysteine levels. Homocysteine plays a role in the development of metabolic syndrome (MetS). MetS is caused by the interaction of multiple genetic and environmental factors.
Although there is a correlation between HHcy and MetS, the mechanisms are still unclear and that is why many researchers have proposed several theories including promotion of endothelial dysfunction, induction of insulin resistance, and DNA methylation status. . Consequently, both DNA methylation and DNA synthesis can be impaired by the interaction with homocysteine, vitamin B12 and folate.
The MTRR A66G polymorphism appears to be associated with an increased risk of MetS only when combined with the MTHFR 677TT genotype. In fact, the combined TT/GG, TT/AG and TT/AA genotypes confer a higher risk of MetS than the MTHFR C677T mutant genotypes alone.

CLINICAL SIGNIFICANCE

Methionine synthase reductase (MTRR) plays a key role in folate metabolism, in interconnection with the enzyme 5,10-methylenetetrahydrofolate reductase (MTHFR). MTHFR catalyzes the regulation of cellular methylation through the conversion of 5,10-methylene tetrahydrofolate (THF) to 5-methyl-THF, the major circulating form of folate metabolism. MTRR is required for the reductive methylation of vitamin B12, also known as cobalamin, an activated cofactor for methionine synthase (MTR), which catalyzes the methylation of homocysteine to methionine.
The methionine synthase reductase (MTRR) gene is located on chromosome 5 and plays a vital role in DNA synthesis.
The A66G polymorphism (rs1801394) has been described for the MTRR gene, resulting in a substitution of the amino acid methionine in isoleucine at codon 22 (M22I).

§ 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
§ Kurzawski M, Wajda A, Malinowski D, Kazienko A, Kurzawa R, Drozdzik 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
§ Yang B, Fan S, Zhi X, Wang D, Li Y, Wang Y, et al. Associations of MTHFR C677T and MTRR A66G gene polymorphisms with metabolic syndrome: a case-control study in Northern China. Int J Mol Sci. 2014; 15(12): 21687-21702
§ Jiang, S.; Zhao, R.; Pan, M.; Venners, S.A.; Zhong, G.; Hsu, Y.H. Associations of MTHFR and MTRR Polymorphisms with serum lipid levels in Chinese hypertensive patients. Clin. Appl. Thromb. most. 2014, 4, 200–210.
§ Jacques, P.F.; Boston, A.G.; Selhub, J.; Rich, S.; Elison, R.C.; Eckfeldt, J.H.; Gravel, R.A.; Rozen, R.; National Heart, Lung; Blood Institute; et al. Effects of polymorphisms of methionine synthase and methionine synthase reductase on total plasma homocysteine in the NHLBI Family Heart Study. Atherosclerosis 2003, 166, 49–55 elevated levels of triglycerides even if these results need to be confirmed with further studies given the relatively low frequency of the MTRR 66GG genotype in many populations..

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ISO 9001:2015
ISO 13485:2016

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DESCRIPTION	LABEL	VOLUME		STORAGE
		GEN-027-25	GEN-027-50	
Mix oligonucleotides and probes	Mix A66G MTRR 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 H20	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-027-25 / COD. GEN-027-50

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
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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