CBS 844ins68 POLYMORPHISM (CYSTATHIONINE β- SYNTHETASE)

For in vitro diagnostic use

CE IVD

ORDERING INFORMATIONS

REF: GEN-014-25 Code RDM: 2256364/R
Tests: 25 Reactions: 31
REF: GEN-014-50 Code RDM: 1793904/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 844ins68 polymorphism of the cystathionine β-synthase (CBS) gene by Real-Time PCR technique. Optimized kit for Real-Time PCR instrumentation CFX96 Dx, Biorad Opus Dx, Agilent AriaDx, Hyris bCUBE and Hyris bCUBE3 with Hyris bAPP.

SCIENTIFIC BACKGROUND

Numerous studies have demonstrated that hyperhomocysteinemia (HHcy) is an independent risk factor for cardiovascular and cerebrovascular diseases and that an increase in hypertension and plasma homocysteine (Hcy) has a synergistic effect in causing these diseases.
Homocysteine is an important intermediate product in the metabolism of methionine and cysteine. The enzymes 5,10-methylenetetrahydrofolate reductase (MTHFR) and cystathionine β-synthetase (CBS) are key enzymes in homocysteine metabolic pathways.
The catalytic activity of the MTHFR enzyme creates an irreversible reduction of 5,10-methylenetetrahydrofolate (THF) which is converted to 5-methyl-THF during this process. 5-methyl-THF is the most abundant circulating form of folic acid, serving as a methyl donor for the remethylation of homocysteine to methionine, a reaction (catalyzed by methionine synthase) for which vitamin B12 is required.
The cystathionine β -synthetase (CBS) gene is located on chromosome 21q22.3 and codes for an enzyme that participates in the folate pathway and catalyzes the transsulfuration of homocysteine and serine to cystathionine as a precursor of cysteine.

CLINICAL SIGNIFICANCE

The CBS gene has a large number of mutations and polymorphisms.
The 844ins68 polymorphism at position 844 in the CBS gene generates an alternative splice site that disrupts the protein, resulting in decreased functional activity of CBS. The deficiency of this enzyme causes an increase in homocysteine in the blood and homocystinuria.
Furthermore, the T833C mutation generating a restriction site of BsrI (CBS I278T) has been shown to segregate in cis with the 844ins68 polymorphism in exon 8.
Significant interactions were observed between the polymorphisms of MTHFR C677T, MTHFR A1298C and the CBS 844ins68/T833C haplotype for Hcy levels. In fact, heterozygotes show higher homocysteine values.
Interactions between the various polymorphisms may therefore influence serum Hcy levels, where multiple heterozygosity could be a risk factor for vaso-occlusive episodes.
The 844ins68 polymorphism has also been associated with other diseases, including neural tube defects and cancer.

§ Interactions among methylenetetrahydrofolate reductase (MTHFR) and cystathionine β-synthase (CBS) polymorphisms - a cross-sectional study: multiple heterozygosis as a risk factor for higher homocysteine levels and vaso-occlusive episodes. Genet Mol Res. 2017 Feb 23;16(1). doi: 10.4238/gmr16019374.

§ Association between 11 genetic polymorphisms in folate-metabolising genes and head and neck cancer risk. Eur J Cancer. 2012 Jul;48(10):1525-31. doi: 10.1016/j.ejca.2011.09.025. Epub 2011 Nov 1.

§ The 844ins68 polymorphism of the cystathionine beta-synthase gene is associated with schizophrenia. Psychiatry Res. 2009 Dec 30;170(2-3):168-71. doi: 10.1016/j.psychres.2008.07.007. Epub 2009 Nov 10.

BIOMOL LABORATORIES S.R.L.
Via Arcora 110 (Palazzo Gecos)
80013 Casalnuovo di Napoli, NA
info@biomollaboratories.com
biomollaboratories.it

ISO 9001:2015
ISO 13485:2016

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DESCRIPTION	LABEL	VOLUME		STORAGE
		GEN-014-25	GEN-014-50	
Mix oligonucleotides	Mix CBS 844ins68 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 HOMOZYGOUS D/D	1 x 22 µl	1 x 22 µl	-20°C
Genomic DNA or recombinant DNA	Control + 2 HETEROZIGOUS I/D	1 x 22 µl	1 x 22 µl	-20°C
DNA genomico o DNA ricombinante	Control + 3 HOMOZYGOUS I/I	1 x 22 µl	1 x 22 µl	-20°C

TECHNICAL CHARACTERISTICS

COD. GEN-014-25 / COD. GEN-014-50

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 con Hyris bAPP
TECHNOLOGY	Real-time PCR; specific oligonucleotides; 1 SYBR-GREEN/FAM fluorescence channel
RUNNING TIME	150 min
THERMAL CYCLING PROFILE	1 cycle at 50 °C (2 min); 1 cycle at 94 °C (5 min); 30 cycles at 95 °C (50 sec) + 60 °C (40 sec) + 72 °C (50 sec) + 1 dissociation cycle at 70 °C with 0,2 °C increments.
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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