AZIENDA CERTIFICATA
UNI EN ISO 9001, UNI EN ISO 13485

DETECTION OF THE POLYMORPHISM OF THE GENE CISTATHIONINE γ-LYSATE (CTH)

CTH 1364 G/T e CHT-1320 C/T

AMPLI set CTH — Cat. n. 2.049RT

Cystathionine gamma-lysate (CTH), is an enzyme widely distributed in the world of prokaryotic and eukaryotic organisms, catalyzes the formation and transformations of sulfur-containing compounds and plays a key role in the desulfurization pathway of L-cysteine. Human CTH is composed of two dimers and each monomer binds pyridoxal phosphate (PLP). The gene, located on the short arm of chromosome 1, is made up of 13 exons and 12 introns. As a result of the alternative splicing, three isoforms of human CTH arise. The analysis of genetic variations of the gene encoding CTH showed a large number of polymorphisms. A decrease in CTH expression leads to a drop in the level of cysteine, glutathione (GSH), taurine and hydrogen sulfide (H2S) in the cells and, more importantly, leads to cystationionuria. H2S, formed endogenously from CTH, influences vasodilation and regulation of blood pressure. Overexpression of the gene that codes for CTH in cells leads to an increase in H2S production. H2S plays a role in protecting neurons from oxidative stress and stimulates an increase in γ-glutamylcysteine synthetase and therefore an increase in the level of GSH. Sulfansferases, including CTH, can locally prevent oxidative stress due to reversible oxidation of SH-groups in the presence of increased levels of reactive oxygen species and reduction of the presence of GSH and / or reduction of thioredoxin. The single nucleotide polymorphisms (SNPs) rs1021737 and rs648743 were chosen because of their high allele frequencies (0.21 and 0.47, respectively) and potential functionality, considering that rs648743 is found in the promoter of the CTH gene (-1320 C> T) and could potentially abolish a glucocorticoid receptor alpha-binding site, while rs10211737 (CTH1364 G> T) is in exon 12. These SNPs / (1021737 and rs648743) CTH will be detected using Real-Time PCR allelic discrimination with specific primers and probes labeled with fluorochromes (W.T. VIC allele, FAM mutated allele).

INTERPRETATION OF RESULT

The analysis of the results will be carried out by the specific
pro (ALLELIC DISCRIMINATION) of the Real-Time t

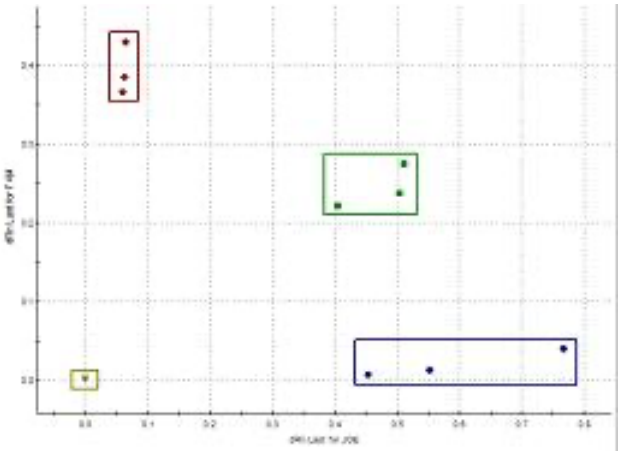
Principle of the method:
genomic DNA extraction, amplification and detection with Real-Time PCR technique. Allelic discrimination.
Applicability: On extracted and purified genomic DNA.

Number of tests: 25.

CONTENTS OF THE KIT AND ITS STORAGE

AMPLIFICATIONS	
PCR mix	-20°C
Mix 20x CTH rs1021737	-20°C
Mix 20x CTH rs648743	-20°C
H_2O sterile	-20°C
Controllo WT	-20°C
Controllo eterozigote	-20°C

Stability: over 12 months if properly stored.



Legend:
Red: WT (*A/A)
Green: heterozygote (*A/B)
Blu: homozygote (*B/B)

Rev.01

Bibliografia:

-Association of CTH variant with sinusoidal obstruction syndrome in children receiving intravenous busulfan and cyclophosphamide before hematopoietic stem cell transplantation P Huezo-Diaz Curtis et. Al. e European Blood and Marrow Transplant group.
-Wang J, Huff AM, Spence JD, Hegele RA. Single nucleotide polymorphism in CTH associated with variation in plasma homocysteine concentration. Clin Genet 2004; 65: 483–486.