KIT FOR THE DETECTION OF 844INS68 POLYMORPHISM OF THE CYSTATHIONINE-β-SYNTHASE (CBS)

AMPLI-SET-CBS 844ins68 Cat. n. 1.332

The deficit of Cystathionine Beta-Synthase enzyme is an inherited autosomal recessive disorder .The enzyme catalyzes the production of Cystathionine from homocysteine and serine. The deficit causes homocystinuria and the related diseases are dislocated optical lenses, central nervous system involvement ,skeletal abnormalities and vascular disease with severe thromboembolic complications. Two clinical forms can be distinguished on the basis of patient's responsiveness to the treatment with the coenzyme precursor piridoxine.
The mutations of CBS gene may be heterozygote, causing a mild homocystinuria and may be a risk factor for cardiovascular pathologies.
The more frequent mutations in Europe are I278T and A114V .Moreover, in Italian families , is frequent the 844ins68 mutation.
Heterozygosis for CBS 844ins68 mutation (present in 7,8% of Caucasian population) isn't a risk factor itself, but it becomes a risk factor when it is associated to mutations of the MTHFR enzyme gene (i.e C677T). In this condition , the risk of occlusive arterial and/or venous pathologies increases of 4 times.
The kit allows the detection of the mutation 844ins68 , where there is the insertion of 68 bp in the exon 8. The insertion breaks off the normal protein sequence, causing the abort of the protein.
The detection of the mutation is performed with the amplification with specific primers of a fragment of 127 bp if the insertion is absent, and of 195 bp if the insertion is present

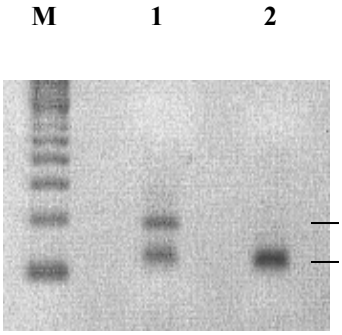
Principle of method: A) extraction of genomic DNA B) amplification C)detection on agarose gel
Applicability: On extracted and purified genomic DNA from whole blood samples.
Tests: 45

REAGENTS AND STORAGE

AMPLIFICATION	
PCR mix	-20°C
H_2O sterile	-20°C
Taq Polymerase (5U/µl)	-20°C
Positive control heterozigous	-20°C

Stability: over 12 months if correctly stored.

ANALYSIS OF RESULTS



1		3
Absence of mutation normal pazient	**Presence of mutation mutant heterozygous**	**Presence of mutation Mutant homozygous**
1 fragment	2 fragments	1 fragment
127 bp	**195 bp 127 bp**	**195 bp**

References:
Hum. Mol. Genet. 1993; 2:1633-8.
Am. J. Hum. Genet. 1995; 56:1324-1333
Thromb Haemost 2000; 84 (4); 576-82