



ampli set EMO C282Y- H63D ^{CE IVD} 45 tests cat 1320

detection of C282Y and H63D mutation in the Haemochromatosis gene(HFE)

Haemochromatosis is a common recessive disorder characterised by progressive iron overload. Unfortunately it is little considered and it is often accidentally discovered in periodic check or due to the development of its complications. In Italy and in some European regions there are 2-5 affected individuals in 1000 people and 9-15 carriers in 100. Haemochromatosis is regarded as the most common genetic disorder in the west hemisphere. In 1996 the HFE gene has been detected and two mutations, C282Y and H63D, have been described. Most affected patients with haemochromatosis are homozygous for the C282Y mutation (80-100%), whereas a few are compound heterozygous for the C282Y and H63D. Homozygote for the H63D mutation isn't clearly linked to haemochromatosis. The kit allows the simultaneous detection of C282D and H63D mutations using the Polymerase Chain reaction PCR and restriction analysis using the Pml restriction enzyme.

Principle of method: A) extraction of genomic DNA; B) amplification; C) enzymatic digestion; D) detection on agarose gel

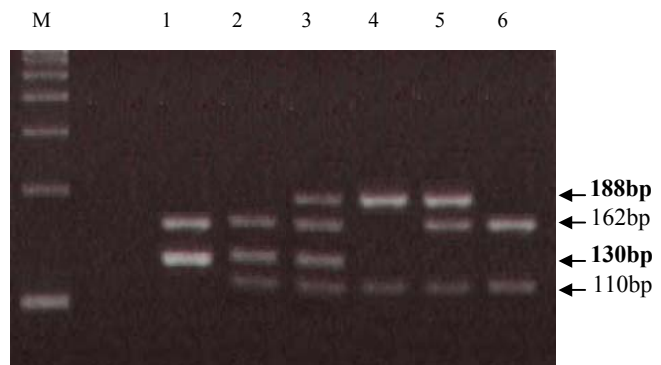
Applicability: on extracted and purified genomic DNA from whole blood samples.

ANALYSIS OF RESULTS

The amplification yield is of 188 bp for the C282Y mutation and 130 bp for the mutation H63D. When the two mutations are present Pml I enzyme isn't able to recognize the specific restriction site. This site is recognized when there isn't the mutation. (wild-type).

M) Marker 100bp ladder

- 1) wild-type C282Y, homozygosis mutation H63D
- 2) wild-type C282Y, heterozigosis H63D
- 3) heterozigosis C282Y, heterozigosis H63D (compound heterozigosis)
- 4) homozigosis C282Y, wild-type H63D
- 5) heterozigosis C282Y, wild-type H63D
- 6) wild-type C282Y e H63D



REFERENCES

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