



ampli set EMO N144H ^{CE IVD}

45 tests

cat 1322

detection of N144H mutation in the Ferroportin gene(SLC11A3)

Hemochromatosis is an inherited disorder with an estimated prevalence of up to 1 in 100 individuals in northern European population. Many patients (80%) present an autosomal-recessive pattern. The mutation is located in the HFE gene

Recently two mutations (A77D and N144H) inherited as autosomal dominant trait in the gene encoding for Ferroportin have been described. Ferroportin (SLC11A3) is a transmembrane iron export protein.

The kit allows the detection of the mutation N144H in the SLC11A3 gene changing an aspartic acid in histidine. The detection of the mutation is carried out using the amplification of a fragment of 310 bp with specific primers followed by restriction section due to Apo I enzyme. The mutation causes the loss of a Apo I site, so the normal allele produces two fragments of 230 bp and 80 bp.

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

1	2	3
Absence of mutation normal homozygous	Presence of mutation mutant heterozygous	Presence of mutation Mutant homozygous
Presence of 2 band	Presence of 3 bands	Presence of 1 band
230bp 80 bp	310 bp 230 bp 80 bp	310 bp

REFERENCES

- Robert E. Fleming et al. *J. Clin. Invest.* 108:521-522 (2001).
Njajou OT et al. *Nat. Genet.* 28 (2001).
Montosi G et al *J. Clin. Invest.* 108: 619-623 (2001).