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detection of A77D mutation in the ferroportin gene(SLC11A3)

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Haemochromatosis 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 A77D in the SLC11A3 gene changing an alanine in aspartic acid. The detection of the mutation is carried out using the amplification with specific primers followed by restriction section due to MboII enzyme.The mutation creates a new MboII site, therefore the PCR fragment (125 bp) containing the mutation is cleaved into two fragments (94 and 31 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

The amplification yield is of 127 bp .The mutation A77D adds a restriction site for the MboII enzyme, obtaining two fragments respectively of 94 bp and 33 bp.

1	2	3
Absence of mutation normal homozygous	Presence of mutation mutant heterozygous	Presence of mutation Mutant homozygous
Presence of 1 band	Presence of 3 bands	Presence of 2 band
125 bp	125 bp 94 bp 31 bp	94 bp 31 bp

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