IRON OVERLOAD
(HFE, EARLIER HLA-H, C282Y, H63D AND S65C)

PATHOGENESIS

It's an autosomal recessive disease caused by mutations on the gene HFE, which is involved in the metabolism of the iron. The pathology is characterized by an increase of iron's absorption at intestinal level and by the consequent deposit in organs provokes irreversible damages especially in the liver and the pancreas.

The substitution of the base G845A (C282Y) is the most important mutation for the insurgence of hemochromatosis with 80-90% of patients with the disease which carries the mutation in an homozygote form.

About the 7% of patients with hemochromatosis are carriers in an heterozygote form of mutations C282Y and H63D. besides, H63D substitution is considered a genetic variant that increases the risk to develop a soft form of the disease.

Recently another mutation (S65C) was associated to a modest iron accumulation and so to an increased risk to develop a soft form of the disease.

EPIDEMIOLOGY

The hemochromatosis (1/4000 of incidence) is one of the most frequent genetic disease in the Caucasian population. Moreover it has an even more frequency in north-Europe lands.

TEST

Research of mutations C282Y, H63D and S65C by means of PCR and restriction analyses. The genetic test is indicated for patients with a familiar history of hemochromatosis or with clinical facts (pathologic values of ferritin which repeats more than once without a clear etiology, diabetes and increased iron value), which are premonitory of the disease.

SAMPLE TAKING

Blood EDTA, 5 ml.

EXECUTION

Daily

COST

According to the federal charge rate of the analyses (2215.13 X2) TP 310.

Further information or bibliographic references can be asked to the laboratory.