HEREDITARY FRUCTOSE INTOLERANCE (HFI)

PATHOGENESIS

The HFI is provoked by an inadequacy of the enzyme aldolase B's activity (absence or partial/total inactivity). This enzyme is responsible of the metabolism of the fructose and it's normally located in the liver, in kidneys and in the little intestine.

The inadequacy of this enzyme's activity causes an accumulation of fructose in these organs. The accumulated fructose in the liver interferes with the activity of many others empathic metabolites, and it inhibits the transformation of the glycogen and the syntheses of glucose.

The presence of a minor activity causes, beyond the others symptoms, strong hypoglycemic crises, liver damages, kidneys' malfunction, coma and death. This pathology is particularly dangerous in neonatal age and during the childhood, but it's anyway dangerous even for adults, because the fructose's assumption takes rapidly to the degeneration of the liver, which can become both a cirrhosis of the liver and an hepatic carcinoma, without considering kidneys' and intestinal' malfunction.

Intolerants to fructose must follow a diet, which doesn't have traces of fructose.

EPIDEMIOLOGY

The hereditary fructose intolerance is an extremely rare, genetic illness, which has a recessive, autosomal inherited character. Its incidence is about 1:20:000. The majority part of cases are identified in Europe and in North America. The HFI is genetically heterogeneous, and more than 40 mutations on the gene aldolase B are described. In the 85% of cases it's provoked by 3 mutations (A149P, A174D and N334K) situated on the chromosome 9.

The A149P is the most frequent mutation, it appears in about 57% of cases.

TEST

Amplification by means of PCR and research of polymorphisms.

SAMPLE TAKING

Blood EDTA, 5 ml.

EXECUTION*

Daily.

Laboratorio di diagnostica molecolare

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