LACTOSE INTOLERANCE (T-13910C)

PATHOGENESIS

Patients who have case of a primary lactose intolerance develop enteropathic and generic symptoms (abdominal loosening, tympanites, colics, diarrhea, nausea and vomit) after the consumption of milk and dairy products, and other products which contain the lactose, which is present even in some medicines. According to this symptomatology, there's a lack of the lactase enzyme in intestinal microvillus, which means a lack of the lactose idrolysis in glucose and galactose and a consequent syndrome of bad absorption. The secondary lactose intolerance is the consequence to the damage of the intestinal mucosa after illness such as celiac disease, Whipple's disease, Crohn's disease or treatments such as the chemotherapy.
Recently, it was discovered that the functionality of the lactase enzyme is compromised by a mutation on position -13910 that is in the regulatory region of the correspondent gene. Studies show that the mutation -13910-> C is transmitted in a recessive autosomal form, where homozygotes subjects for the mutation (genotype -13910 CC) show the phenotype. Studies on Italian, Finnish, German and Korean patients with a biochemically demonstrated lack of lactase highlighted a complete correlation to the genetic variant -1390CC.
The diagnosis of the lactose intolerance was up to now done by means of invasive and not reliable tests. The identification of the responsible polymorphism of the primary lactose intolerance ensures the patient a rapid, not invasive and with a 100% specificity exam.
The importance of a precise diagnosis it's clear if it's considered the fact that the therapy for the lactose intolerance consists in the change of alimentary habits of the patient. Moreover, the rapidity of the test is very important to conferm or to exclude the lactose intolerance as cause of the almost common enteropathic symptomatology.

EPIDEMIOLOGY

The frequency of the lactose intolerance varies according to the studied population. In Europe, it increases from North to South with a frequency of 15-20% in central Europe and about the 70% in south Europe. In African and Asian it gets quite the 100%.

TEST

It highlights the mutation T-13910C on the LPH gene by means of PCR and there's the hybridization of the amplified product.

SAMPLE TAKING

Blood/EDTA, 5 ml. or smear

EXECUTION

Daily.

COST

According to the federal charge rate of the analyses (2215.08) TP 105.

Laboratorio di diagnosi molecolare

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