Gene test information

HEREDITARY FRUCTOSE INTOLERANCE (HFI) (FRUCTOSEMIA, ALDOLASE B DEFICIENCY)

- **Background**

  Hereditary fructose intolerance (HFI) is a hereditary condition caused by a deficiency of liver enzymes that metabolise fructose. The deficient enzyme is aldolase-B (ALDOB), which converts fructose-1-phosphate to DHAP and glyceraldehyde. This means that the fructose cannot be further metabolised beyond fructose-1-phosphate. The incidence of HFI in Europe is about 1:20.000..

- **Symptoms of HFI**

  Symptoms include severe abdominal pain, vomiting, and hypoglycemia following ingestion of fructose or other sugars metabolized through fructose-1-phosphate. Prolonged fructose ingestion in infants leads ultimately to hepatic and/or renal failure and death. Patients develop a strong distaste for sweet food, and can avoid a recurrence of symptoms chronic course of the disease by remaining on a fructose- and sucrose-free diet.

  HFI must not be confused with fructose malabsorption, which is a non-life threatening and much more common condition.

- **Causes of HFI**

  HFI is caused by mutations in the ALDOB gene, which encodes the enzyme Fructose-1-phosphate-aldolase-B. Depending on the type of mutation, enzymatic activity can be reduced by 85 to 100%.

- **Diagnosis of HFI**

  I: Genetic test for the most common ALDOB mutations ALDOB A149P, A174D and N334K. This test detects about 87% of ALDOB mutations in Europe.

  II: Complete sequence analysis of the ALDOB gene to detect rare mutations.

  Note: The H_2 breath test, which is a safe and noninvasive procedure for the diagnosis of fructose malabsorption, is not useful for the diagnosis of HFI.

References:

Santer et al., The spectrum of aldolase B (ALDOB) mutations and the prevalence of hereditary fructose intolerance in Central Europe. Hum Mutat. 2005;25:594.

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