GENE Seeker

You know the risk of your child being a carrier of a genetic disease.

Hereditary fructose intolerance

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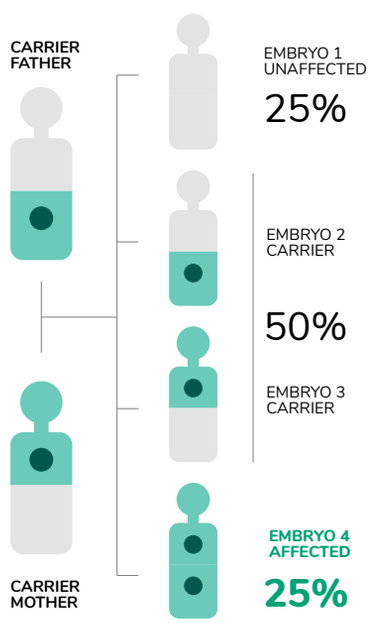
What is Hereditary fructose intolerance?

Hereditary fructose intolerance (HFI) is an autosomal recessive disorder of fructose metabolism (see this term), resulting from a deficiency of hepatic fructose-1-phosphate aldolase activity and leading to gastrointestinal disorders and postprandial hypoglycemia following fructose ingestion. HFI is a benign condition when treated, but it is life-threatening and potentially fatal if left untreated.

HFI usually presents in infancy at the time of weaning (when fructose is added to the diet), manifesting with hypoglycemia, lactic acidosis, ketosis with recurrent vomiting, abdominal pain and systemic manifestations following consumption of fructose-containing foods. Persistent ingestion of fructose and related sugars (such as sucrose and sorbitol) may lead to growth retardation, hepatomegaly, proximal tubular dysfunction, liver and renal failure, seizures, coma and risk of death. All patients achieve adulthood, develop a natural aversion to fruit/sweets and report a life long history of vomiting and hypoglycemia following fructose ingestion. Dental caries are absent in a significant proportion of adult population with HFI (which may give a clue to the diagnosis). Sometimes, the diagnosis may be made in an adult who has, owing to his aversion to fructose, excluded all fructose-containing food since childhood.



What is the next step if I'm a carrier of Hereditary fructose intolerance?

If you are found to be a carrier of Hereditary fructose intolerance, it is important that your partner be tested for the same genetic disorder.

What if my partner is not a carrier?

If your partner's test for Hereditary fructose intolerance is negative, the chance to have an affected child is low. However there is currently no test able to detect all existing mutations, so there is always a residual risk that the person who has done the test is a carrier of other less frequent mutations.

What if both me and my partner are carriers of Hereditary fructose intolerance?

When both parents are carriers of Hereditary fructose intolerance , the probability of having a child with Hereditary fructose intolerance is 25%.

We recommend that you discuss your results with your doctor or genetic counselor in order to know more about reproductive options.

If both you and your partner are carriers, speak with your doctor or genetic counselor about reproductive options.

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