



## What is Hereditary Fructose Intolerance?

Hereditary fructose intolerance (HFI) is an inherited disease characterized by nausea, abdominal pain, diarrhea, vomiting, low blood sugar, and liver and kidney damage. It involves defects in making the enzyme aldolase B, which is responsible for breaking down fructose (a sugar found primarily in fruit) into a form used by the body for energy.<sup>1</sup>

## What are the symptoms of Hereditary Fructose Intolerance and what treatment is available?

Symptoms of hereditary fructose intolerance begin when fructose or sucrose (table sugar) is introduced in the diet. Infant formulas, baby foods, and juices may contain sugars that can cause symptoms.<sup>2</sup>

Symptoms are usually characterized by<sup>1,2</sup>:

- Lethargy (lack of energy)
- Vomiting
- Diarrhea
- Abdominal pain
- Hypoglycemia (low blood sugar)
- Poor growth
- Seizures, possibly leading to coma (after ingesting large amounts of sugar)

If an affected individual is not treated, other symptoms may develop, including slow, stunted growth and liver and/or kidney damage<sup>1</sup>

There is no cure for hereditary fructose intolerance. Treatment includes strict avoidance of foods containing fructose, sucrose, and sorbitol (which contains fructose). Fructose is often found in sweet-tasting foods, but it may also be found in unexpected products, such as medicines.<sup>2</sup> Avoidance of fasting, especially during illness, is also advised.<sup>3</sup> Affected individuals tend to develop a natural aversion to sweet foods and may remain undiagnosed, but these individuals may develop symptoms if sugars are consumed.<sup>2</sup>

## How is Hereditary Fructose Intolerance inherited?

Hereditary fructose intolerance is an autosomal recessive disease caused by mutations in the *ALDOB* gene.<sup>1</sup> An individual who inherits one copy of an *ALDOB* gene mutation is a “carrier” and is not expected to have related health problems. An individual who inherits two mutations in the *ALDOB* gene, one from each parent, is expected to be affected with HFI.

If both members of a couple are carriers, the risk for an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

## Who is at risk for Hereditary Fructose Intolerance?

Hereditary fructose intolerance can occur in individuals of all races and ethnicities.<sup>4</sup> HFI is estimated to affect 1 in 20,000 with an approximate carrier frequency of 1/71.<sup>1</sup>

Having a relative who is a carrier or who is affected can increase an individual's risk of being a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.





### What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

### What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

### Where can I get more information?

- NIH Office of Rare Diseases Research - Genetic and Rare Diseases (GARD) Information Center. [https://www.rarediseases.info.nih.gov/GARD/Condition/6622/Hereditary\\_fructose\\_intolerance.aspx](https://www.rarediseases.info.nih.gov/GARD/Condition/6622/Hereditary_fructose_intolerance.aspx)
- HFI Information Boards: <http://hfiinfo.proboards.com/>

### References

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