Hereditary Fructose Intolerance

Hereditary fructose intolerance is a rare genetic disorder. It is characterized by low blood sugar levels, constipation, and vomiting after eating fructose. A person must have two variants in the ALDOB gene in order to have this condition.

play Friedrich740, you do not have the variants we tested.

You would still have a variant not covered by the test.

0 variants detected in the ALDOB gene.

How To Use This Test

This test does not diagnose any health conditions. Please talk to a healthcare professional if you think you might have this condition, or if you have any concerns about your results.

Review the Carrier Status tutorial
See Scientific Details

Intended Uses

- Tests for multiple variants in the ALDOB gene.
- To identify carrier status for hereditary fructose intolerance.

Limitations

- Does not test for all possible variants for the condition.
- Does not report carrier status when two copies of variant exist.

Important Ethnicities

- This test is most relevant for people of European descent.

You are likely not a carrier.

This result may be less relevant to you because the variants that cause hereditary fructose intolerance are rarely found in people of your ethnicity.

We ruled out the tested variants for hereditary fructose intolerance. These variants are most common in people of European descent.

You still have a chance of being a carrier for hereditary fructose intolerance. We cannot estimate your chance because this condition occurs on a very rare occasion, even in your ethnicity.

About Hereditary Fructose Intolerance

When symptoms develop

Symptoms typically develop during infancy.

Typical signs and symptoms

- Nausea and vomiting
- Low blood sugar
- Constipation
- Gastrointestinal pain
- Failure to gain weight
- Liver disease
- Kidney disease

Read more at: Genes Home Reference, MedGenMx, National Organization for Rare Disorders

Consider talking to a healthcare professional if you are concerned about your results.

If you’re starting a family, a genetic counselor can help you and your partner understand if additional testing might be appropriate.

Connect with a counselor

Share your results with a healthcare professional.

Learn more about this condition and connect with a support group.
**Background Details**

Hereditary fructose intolerance is a genetic disorder. It is caused by a deficiency of the enzyme fructose-1,6-bisphosphatase, which is necessary for the metabolism of fructose. This enzyme is located in the liver and kidneys.

**Test Interpretation**

- **Mendelian Inheritance:** The test can be passed on from parents to children.

- **Significance:** The test can be used to identify carriers of the mutation who may not have symptoms but can pass the mutation to their offspring.

- **False-Positive Results:** False-positive results can occur if the sample is contaminated with substances that can interfere with the test.

- **False-Negative Results:** False-negative results can occur if the sample is not properly collected or if the mutation is present in a less common form.

- **Accuracy:** The accuracy of the test is high, with a sensitivity and specificity of greater than 99%.

- **Purpose:** The test is used to identify individuals who are carriers of the mutation, which is important for genetic counseling and family planning.

**References**


**Conclusion:**

Hereditary fructose intolerance is a serious genetic disorder that can lead to severe health problems. Early detection and treatment are crucial to prevent complications. Genetic counseling and family planning are important for individuals who are carriers of the mutation.