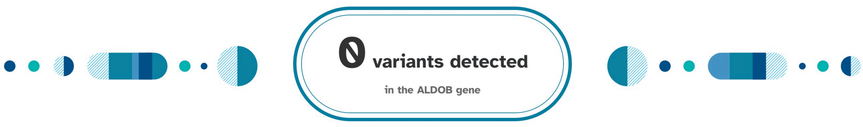
Hereditary Fructose Intolerance

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

Overview Scientific Details

Jamie, you **do not have the variants** we tested.

You could still have a variant not covered by this test.



How To Use This Test

This test does not diagnose any health conditions.

Please talk to a healthcare professional if this condition runs in your family, you think you might have this condition, or 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** if someone has two copies of a tested variant.

Important Ethnicities

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

You are likely not a carrier.

This result is relevant for you because you have **European** ancestry.

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

You still have a chance of being a carrier for hereditary fructose intolerance.

You may still have up to a **1 in 460 chance** of carrying a variant not covered by this test.

See Scientific Details

About Hereditary Fructose Intolerance

When symptoms develop

Symptoms typically develop during infancy.

Typical signs and symptoms

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

Ethnicities most affected

This condition occurs in people of all ethnicities, but is best studied in people of European descent.

How it's treated

There is currently no known cure. Maintaining a fructose-free diet may reduce or prevent symptoms.

Read more at: Genetics Home Reference MedlinePlus 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 GC

Share your results with a healthcare professional.

Print report

Learn more about this condition and connect with support groups.

Learn more

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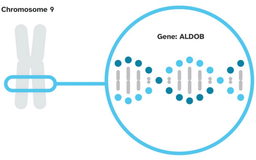
Overview Scientific Details

Hereditary fructose intolerance is caused by variants in the ALDOB gene.

ALDOB

The ALDOB gene contains instructions for making an enzyme called aldolase B. This enzyme is found mainly in the liver, where it helps to break down the sugar fructose. Certain variants in ALDOB disrupt this function, resulting in a harmful buildup of fructose byproducts in liver cells.

Read more at Genetics Home Reference



You have no variants detected by this test.

Variants Detected | View All Tested Markers

Marker Tested	Your Genotype*	Additional Information
A149P **Gene:** ALDOB **Marker:** rs1800546	**C** Typical copy from one of your parents **C** Typical copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [5, 6, 7, 8, 10, 11] | ClinVar
A174D **Gene:** ALDOB **Marker:** i5998215	**G** Typical copy from one of your parents **G** Typical copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [5, 6, 7, 8, 11] | ClinVar
N334K **Gene:** ALDOB **Marker:** i5012664	**G** Typical copy from one of your parents **G** Typical copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [3, 5, 6, 7, 8, 9, 11] | ClinVar
Delta4E4 **Gene:** ALDOB **Marker:** i5012665	**TTTG** Typical copy from one of your parents **TTTG** Typical copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [1, 4, 7, 8, 11] | ClinVar

*This test cannot distinguish which copy you received from which parent. This test also cannot determine whether multiple variants, if detected, were inherited from only one parent or from both parents. This may impact how these variants are passed down.

23andMe always reports genotypes based on the 'positive' strand of the human genome reference sequence (build 37). Other sources sometimes report genotypes using the opposite strand.

Test Interpretation

This report provides an estimate of the chances of still being a carrier for people who do not have the variant(s) tested. This is known as the **post-test carrier risk**.

Post-test carrier risk is based on the average chance of being a carrier for a given ethnicity and the carrier detection rate of the test for a given ethnicity.

View technical article on estimating post-test carrier risk.

Post-Test Carrier Risk

This report provides an estimate of the post-test carrier risk for people of European descent only.

- For people of partial European descent, post-test carrier risk is less than that for those who are fully European. The exact post-test risk depends on how much European ancestry a person has.
- Post-test risk for other ethnicities cannot be provided because sufficient data is not available.

Post-test carrier risk for relevant ethnicities

European	1 in 469	[2]

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Hereditary Fructose Intolerance is indicated for the detection of four variants in the ALDOB gene. This test is intended to be used to determine carrier status for hereditary fructose intolerance in adults, but cannot determine if a person has two copies of a tested variant. The test is most relevant for people of European descent.

Special Considerations

- There are currently no professional guidelines in the U.S. for carrier testing for this condition.

Test Performance Summary

Carrier Detection Rate & Relevant Ethnicities

The "carrier detection rate" is an estimate of the percentage of carriers for this condition that would be identified by this test. Carrier detection rate differs by ethnicity and is provided only where sufficient data is available.

European	85% (averaged across multiple countries)	[1]

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing. Greater than 99% of test results were correct. While unlikely, this test may provide false positive or false negative results. For more details on the analytical performance of this test, refer to the package insert.

Warnings and Limitations

- This test does not cover all variants that could cause this condition.*
- This test does not diagnose any health conditions.
- Positive results in individuals whose ethnicities are not commonly associated with this condition may be incorrect. Individuals in this situation should consider genetic counseling and follow-up testing.
- Share results with your healthcare professional for any medical purposes.
- If you are concerned about your results, consult with a healthcare professional.

See the **Package Insert** for more details on use and performance of this test.

* Variants not included in this test may be very rare, may not be available on our genotyping platform, or may not pass our testing standards.

References

1. Coffee EM et al. (2010). "Increased prevalence of mutant null alleles that cause hereditary fructose intolerance in the American population." J Inherit Metab Dis. 33(1):33-42.
2. Coffee EM et al. (2010). "Mutations in the promoter region of the aldolase B gene that cause hereditary fructose intolerance." J Inherit Metab Dis. 33(6):715-25.
3. Cross NC et al. (1990). "A new aldolase B variant, N334K, is a common cause of hereditary fructose intolerance in Yugoslavia." Nucleic Acids Res. 18(7):1925.
4. Dazzo C et al. (1998). "Molecular evidence for compound heterozygosity in hereditary fructose intolerance." Am J Hum Genet. 46(6):1194-9.
5. Esposito G et al. (2002). "Structural and functional analysis of aldolase B mutants related to hereditary fructose intolerance." FEBS Lett. 531(2):152-6.
6. Rellos P et al. (2000). "Expression, purification, and characterization of natural mutants of human aldolase B. Role of quaternary structure in catalysis." J Biol Chem. 275(2):1145-51.
7. Santamaria R et al. (1996). "Molecular basis of hereditary fructose intolerance in Italy: identification of two novel mutations in the aldolase B gene." J Med Genet. 33(9):786-8.
8. Santer R et al. (2005). "The spectrum of aldolase B (ALDOB) mutations and the prevalence of hereditary fructose intolerance in Central Europe." Hum Mutat. 25(6):594.
9. Sebastio G et al. (1991). "Aldolase B mutations in Italian families affected by hereditary fructose intolerance." J Med Genet. 28(4):241-3.
10. Stopa JD et al. (2011). "Stabilization of the predominant disease-causing aldolase variant (A149P) with zwitterionic osmolytes." Biochemistry. 50(5):663-71.

See all references

Change Log

Your report may occasionally be updated based on new information. This Change Log describes updates and revisions to this report.

Date	Change
March 2, 2018	The variant A174D (i5998215) was added to the report. Customers who have this variant will see this variant detected in their result, and see updated content in their report. The carrier detection rate was updated for customers who self-report having European ancestry. The chances of still being a carrier were also updated for customers with no variants detected who self-report having European ancestry.
Feb. 16, 2016	Due to improvements in data analysis, some customers who previously received a "Not Determined" result for rs1800546 may see a genotype at this marker. This may also update the overall report result for these customers.
Oct. 21, 2015	Hereditary Fructose Intolerance report created.