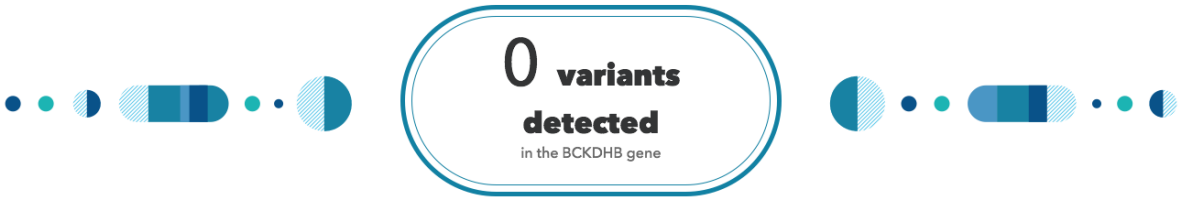
Maple Syrup Urine Disease Type 1B

MSUD 1B is a rare genetic disorder. It is characterized by poor growth and feeding, slowed mental and physical processes, and urine with a distinct, sweet odor. A person must have two variants in the BCKDHB gene in order to have this condition.

Erin, 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 BCKDHB gene.
- To identify carrier status for MSUD 1B.

Limitations

- Does **not test** for all possible variants for the condition.
- Does **not report** if someone has two copies of a tested variant.
- Does **not cover** other types of MSUD.

Important Ethnicities

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

You are likely not a carrier.

This result is relevant for you because you have **Ashkenazi Jewish** ancestry.

We ruled out the most common variants for MSUD 1B in people of Ashkenazi Jewish descent.

You still have a chance of being a carrier for MSUD 1B.

You may still have up to a **1 in 1,200 chance** of carrying a variant not covered by this test.

See Scientific Details

About Maple Syrup Urine Disease Type 1B

Also known as: Branched-Chain Ketoacid Dehydrogenase (BCKD) Deficiency

When symptoms develop

Symptoms typically develop during infancy or in early childhood.

How it's treated

There is currently no known cure. Strict diet management, and in some cases liver transplantation, may reduce symptoms and slow or stop disease progression.

Typical signs and symptoms

- Sweet-smelling urine
- Poor feeding and growth
- Lethargy
- Developmental delay
- Coma and death if untreated

Ethnicities most affected

This condition is most common in people of Ashkenazi Jewish descent.

Read more at

Genetics Home Reference

GeneReviews

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.

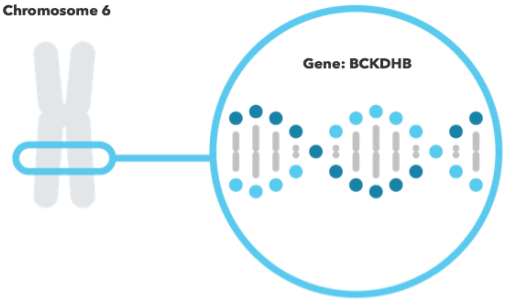
Learn more

MSUD 1B is caused by variants in the BCKDHB gene.

BCKDHB

The BCKDHB gene contains instructions for making one part of an enzyme complex called branched-chain alpha-keto acid dehydrogenase. This enzyme complex breaks down certain types of amino acids, the building blocks of proteins. Certain variants in BCKDHB result in an enzyme complex that cannot break down these amino acids properly. This causes a harmful buildup of amino acids inside cells.

Read more at Genetics Home Reference



You have no variants detected by this test.

Variants Detected	View All Tested Markers
0	2

Marker Tested	Your Genotype*		Additional Information
R183P Gene: BCKDHB Marker: i3002808	**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** [1 , 7] | ClinVar
G278S Gene: BCKDHB Marker: i4000422	**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** [1 , 2 , 3 , 4] | 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 Ashkenazi Jewish descent only.

- For people of partial Ashkenazi Jewish descent, post-test carrier risk is less than that for those who are fully Ashkenazi Jewish. The exact post-test risk depends on how much Ashkenazi Jewish 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

Ashkenazi Jewish	1 in 1,200	[5]

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Maple Syrup Urine Disease Type 1B is indicated for the detection of two variants in the BCKDHB gene. This test is intended to be used to determine carrier status for MSUD 1B in adults, but cannot determine if a person has two copies of a tested variant. The test is most relevant for people of Ashkenazi Jewish 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.

Ashkenazi Jewish	92%	[1]

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing for 92 samples with known variant status. 92 out of 92 genotype results were correct. Fewer than 1 in 100,000 samples may receive a **Not Determined** result for one or more variants included in this test. This can be caused by random test error or unexpected DNA sequences that interfere with the test. It can also be caused by having two copies of a variant tested.

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. Edelmann L et al. (2001). "Maple syrup urine disease: identification and carrier-frequency determination of a novel founder mutation in the Ashkenazi Jewish population." Am J Hum Genet. 69(4):863-8.
2. Flaschker N et al. (2007). "Description of the mutations in 15 subjects with variant forms of maple syrup urine disease." J Inherit Metab Dis. 30(6):903-9.
3. Nellis MM et al. (2003). "Relationship of causative genetic mutations in maple syrup urine disease with their clinical expression." Mol Genet Metab. 80(1-2):189-95.
4. Puckett RL et al. (2010). "Maple syrup urine disease: further evidence that newborn screening may fail to identify variant forms." Mol Genet Metab. 100(2):136-42.
5. Scott SA et al. (2010). "Experience with carrier screening and prenatal diagnosis for 16 Ashkenazi Jewish genetic diseases." Hum Mutat. 31(11):1240-50.
6. Strauss KA et al. (1993). "Maple Syrup Urine Disease"
7. Wynn RM et al. (2001). "Biochemical basis of type IB (E1beta) mutations in maple syrup urine disease. A prevalent allele in patients from the Druze kindred in Israel." J Biol Chem. 276(39):36550-6.