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.

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 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.

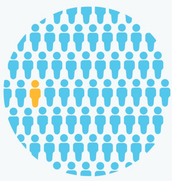
This result may be less relevant for you because the variants that cause MSUD 1B are rarely found in people of your ethnicity.

We ruled out the tested variants for MSUD 1B.

These variants are most common in people of **Ashkenazi Jewish** descent.

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

We cannot estimate your chances because this condition is rare and not well studied in your ethnicity.



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.

Ethnicities most affected

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

Typical signs and symptoms

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

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.

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

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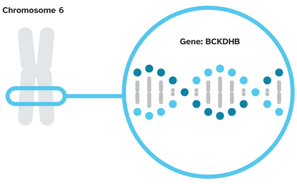
Overview | Scientific Details

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

Marker Tested	Your Genotype*			Additional Information
R183P **Gene:** BCKDHB **Marker:** i3002898	**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 [2, 8] | 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 [2, 3, 4, 5] | 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	[6]

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. However, ACOG notes that testing for maple syrup urine disease may be considered for people of Ashkenazi Jewish descent who are considering having children.

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%	[2]

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. Committee on Genetics. (2017). "Committee Opinion No. 691: Carrier Screening for Genetic Conditions." Obstet Gynecol. 129(3):e41-e55.
2. 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.
3. 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.
4. 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.
5. 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.
6. Scott SA et al. (2010). "Experience with carrier screening and prenatal diagnosis for 16 Ashkenazi Jewish genetic diseases." Hum Mutat. 31(11):1240-50.
7. Strauss KA et al. (2006). "Maple Syrup Urine Disease." [Updated 2013 May 9].
8. Wynn RM et al. (2001). "Biochemical basis of type 1B (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.

Change Log

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

Date	Change
Feb. 18, 2016	Due to improvements in data analysis, some customers who previously received a "Not Determined" result for i4000422 may see a genotype at this marker. This may also update the overall report result for these customers.
Oct. 21, 2015	Maple Syrup Urine Disease Type 1B report created.