D-Bifunctional Protein Deficiency

DBPD is a rare genetic disorder. It is characterized by abnormal muscle tone, developmental disability, seizures, and early death. A person must have two variants in the HSD17B4 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 HSD17B4 gene.
- To identify carrier status for DBPD.

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 does **not** include the majority of HSD17B4 variants that cause DBPD in any ethnicity.

You are likely not a carrier.

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

We ruled out the tested variants for DBPD.

These variants are very rare in all ethnicities.

You still have a chance of being a carrier for DBPD.

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

About D-Bifunctional Protein Deficiency

Also known as: Peroxisomal Bifunctional Enzyme Deficiency. DBPD is a form of leukodystrophy.

When symptoms develop

Symptoms typically develop at birth or during infancy.

How it's treated

There is currently no known cure. Treatment focuses on managing symptoms and preventing complications.

Typical signs and symptoms

- Abnormal muscle tone
- Seizures
- Developmental disability
- Hearing and vision loss
- Distinctive facial features
- Early death

Ethnicities most affected

This condition is rare in all ethnicities.

Read more at

Genetics Home Reference
GeneReviews
NIH Office of Rare Diseases Research
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.

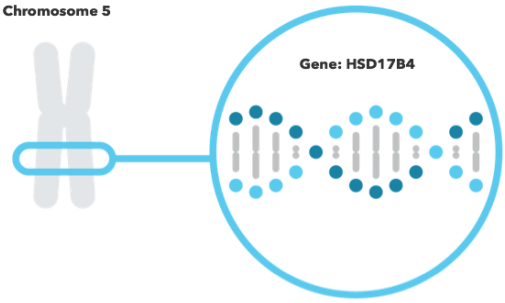
Learn more

DBPD is caused by variants in the HSD17B4 gene.

HSD17B4

The HSD17B4 gene contains instructions for making an enzyme called D-bifunctional protein. This enzyme is found in peroxisomes, where it helps break down molecules called fatty acids to make energy. Certain variants in HSD17B4 disrupt this function, causing a harmful buildup of fat molecules inside of 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
G16S Gene: HSD17B4 Marker: i5007145	**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 , 5 , 7] | ClinVar
N457Y Gene: HSD17B4 Marker: i5007146	**A** Typical copy from one of your parents	**A** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [2 , 4 , 6] | 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

Post-test carrier risk for DBPD is the chance of still being a carrier for the condition if you do not have the variants tested. This chance depends on how common it is to be a carrier for DBPD and whether the variants we tested tend to be found in people of your ethnicity.

Because you do not have the variants we tested, your chances of still being a carrier are lower than for someone who has not been tested. However, we cannot provide an exact estimate because the information needed to calculate post-test carrier risk is not available for your ethnicity.

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for D-Bifunctional Protein Deficiency is indicated for the detection of two variants in the HSD17B4 gene. This test is intended to be used to determine carrier status for DBPD in adults, but cannot determine if a person has two copies of a tested variant.

Special Considerations

- This test does not include the majority of HSD17B4 variants that cause DBPD in any ethnicity.
- 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.

Worldwide	35%	[2]

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing for 97 samples with known variant status. 97 out of 97 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. Ferdinandusse S et al. (2006). "Clinical and biochemical spectrum of D-bifunctional protein deficiency." Ann Neurol. 59(1):92-104.
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3. Qin YM et al. (2000). "Human peroxisomal multifunctional enzyme type 2. Site-directed mutagenesis studies show the importance of two protic residues for 2-enoyl-CoA hydratase 2 activity." J Biol Chem. 275(7):4965-72.
4. Tsuchida S et al. (2012). "Hydratase activities of green fluorescent protein tagged human multifunctional enzyme type 2 hydratase domain and its variants." J Oleo Sci. 61(8):443-50.
5. Van Grunsven EG et al. (1998). "Peroxisomal D-hydroxyacyl-CoA dehydrogenase deficiency: resolution of the enzyme defect and its molecular basis in bifunctional protein deficiency." Proc Natl Acad Sci U S A. 95(5):2128-33.
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7. Van Grunsven EG et al. (1999). "Peroxisomal bifunctional protein deficiency revisited: resolution of its true enzymatic and molecular basis." Am J Hum Genet. 64(1):99-107.
8. Vanderver A et al. (1993). "Leukodystrophy Overview"