Nijmegen Breakage Syndrome

Nijmegen breakage syndrome is a rare genetic disorder. It is characterized by developmental delay, recurring infections, and an increased risk of cancer. A person must have two variants in the NBN gene in order to have this condition.

Erin, you **do not have the variant** we tested.

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

0 variants detected in the NBN gene

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**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

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**Intended Uses**

- To test for the 657del5 variant in the NBN gene.
- To identify carrier status for Nijmegen breakage syndrome.

**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 expected to identify the majority of carriers in people of Eastern European descent.

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You are likely not a carrier.

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

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We ruled out the tested variant for Nijmegen breakage syndrome.

This variant is very rare in all ethnicities.

You still have a chance of being a carrier for Nijmegen breakage syndrome.

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

Also known as: Ataxia-telangiectasia Variant 1, Berlin Breakage Syndrome

When symptoms develop
Symptoms typically develop before birth.

How it's treated
There is currently no known cure. Treatment focuses on managing symptoms and preventing complications such as infection and cancer.

Typical signs and symptoms
- Small head size
- Developmental delay
- Recurring infections
- Increased risk for cancer

Ethnicities most affected
This syndrome is most common in people of Eastern European descent, particularly of Slavic descent.

Read more at
Genetics Home Reference

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

Nijmegen breakage syndrome is caused by variants in the NBN gene.

The NBN gene contains instructions for making a protein called nibrin. This protein plays several vital roles in the cell, including repairing damaged DNA. Certain variants in NBN result in a shortened version of the protein with a reduced ability to repair damaged DNA.

Read more at Genetics Home Reference
You have no variants detected by this test.

<table>
<thead>
<tr>
<th>Marker Tested</th>
<th>Genotype*</th>
<th>Additional Information</th>
</tr>
</thead>
<tbody>
<tr>
<td>657del5</td>
<td>TTGT</td>
<td>Typical copy from one of your parents</td>
</tr>
<tr>
<td>Gene: NBN</td>
<td></td>
<td>Typical copy from your other parent</td>
</tr>
<tr>
<td>Marker: 8012770</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

*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

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

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

<table>
<thead>
<tr>
<th>Ethnicity</th>
<th>Risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>Eastern European, particularly Czech</td>
<td>1 in 15,000</td>
</tr>
</tbody>
</table>

[5, 8]

Test Details

Indications for Use

The 23andMe PGx Carrier Status Test for Nijmegen Breakage Syndrome is indicated for the detection of the 657del5 variant in the NBN gene. This test is intended to be used to determine carrier status for Nijmegen breakage syndrome adults, but cannot determine if a person has two copies of a tested variant.

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.

<table>
<thead>
<tr>
<th>Ethnicity</th>
<th>Rate</th>
</tr>
</thead>
<tbody>
<tr>
<td>Eastern European, particularly Czech</td>
<td>&gt;99%</td>
</tr>
</tbody>
</table>

[1, 5, 6]

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing for 47 samples with known variant status. 47 out of 47 genotype results were correct. Fewer than 1 in 100,000 samples may receive a Not Determined result. 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 in our genotyping platform, or may not pass our testing standards.
References


