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.

Overview Scientific Details

Jamie, you **do not have the variant** 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

- 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.

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.

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.

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.

How it's treated

There is currently no known cure. Treatment focuses on managing symptoms and preventing complications such as infection and cancer.

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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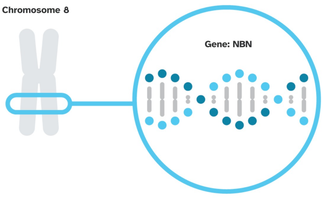
Overview | Scientific Details

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

NBN

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.

Variants Detected | View All Tested Markers

Marker Tested	Your Genotype*		Additional Information
657del5 **Gene:** NBN **Marker:** i5012770	**TTTGT** Typical copy from one of your parents	**TTTGT** Typical copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [2, 3, 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

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 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

Eastern European, particularly Slavic	1 in 15,000	[5, 8]

Test Details

Indications for Use

The 23andMe PGS 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 in 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.

Eastern European, particularly Slavic	>99%	[1, 5, 6]

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. Chrzanowska KH et al. (2012). "Nijmegen breakage syndrome (NBS)." Orphanet J Rare Dis. 7:13.
2. Dzikiewicz-Krawczyk A et al. (2012). "Impact of heterozygous c.657-661del, p.I171V and p.R215W mutations in NBN on nibrin functions." Mutagenesis. 27(3):337-43.
3. Kleier S et al. (2000). "Clinical presentation and mutation identification in the NBS1 gene in a boy with Nijmegen breakage syndrome." Clin Genet. 57(5):384-7.
4. Krüger L et al. (2007). "Cancer incidence in Nijmegen breakage syndrome is modulated by the amount of a variant NBS protein." Carcinogenesis. 28(1):107-11.
5. Maurer MH et al. (2010). "High prevalence of the NBN gene mutation c.657-661del5 in Southeast Germany." J Appl Genet. 51(2):211-4.
6. Resnick IB et al. (2002). "Nijmegen breakage syndrome: clinical characteristics and mutation analysis in eight unrelated Russian families." J Pediatr. 140(3):355-61.
7. Varon R et al. (1999). "Nijmegen Breakage Syndrome." [Accessed Aug 27, 2020].
8. Varon R et al. (2000). "Clinical ascertainment of Nijmegen breakage syndrome (NBS) and prevalence of the major mutation, 657del5, in three Slav populations." Eur J Hum Genet. 8(11):900-2.

Change Log

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

Date	Change
Oct. 21, 2015	Nijmegen Breakage Syndrome report created.