Usher Syndrome Type 3A

Usher 3A is a rare genetic disorder. It is characterized by hearing and vision loss that begins in late childhood and worsens over time. A person must have two variants in the CLRN1 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 N48K variant in the CLRN1 gene.
- To identify carrier status for Usher 3A.

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 subtypes of Usher syndrome.

Important Ethnicities

- This test is most relevant for people of **Ashkenazi Jewish** descent.
- This test does **not** include variants commonly found in people of Finnish descent with Usher 3A.

You are likely not a carrier.

This result may be less relevant for you because the variants that cause Usher 3A are rarely found in people of your ethnicity.

We ruled out the tested variant for Usher 3A.

This variant is most common in people of **Ashkenazi Jewish** descent.

You still have a chance of being a carrier for Usher 3A.

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

About Usher Syndrome Type 3A

When symptoms develop

Symptoms typically develop during late childhood or adolescence.

Ethnicities most affected

This syndrome is most common in people of Ashkenazi Jewish and Finnish descent.

Typical signs and symptoms

- Hearing loss in childhood or early teens
- Gradual vision loss
- Night blindness by mid-teens
- Blindness by mid-adulthood

How it's treated

There is currently no known cure. Hearing loss may be treated with hearing aids. Vision loss may be monitored with routine eye exams. Early intervention is recommended to teach alternative communication skills.

Read more at: Genetics Home Reference National Institute on Deafness and Other Communication 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

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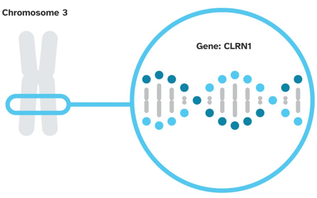
Overview | Scientific Details

Usher 3A is caused by variants in the CLRN1 gene.

CLRN1

The CLRN1 gene contains instructions for making a protein called clarin 1. Although its exact function is not known, clarin 1 is found in the ears and eyes, where it may help nerve cells communicate with each other. Certain variants in CLRN1 prevent the protein from functioning properly.

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
N48K **Gene:** CLRN1 **Marker:** i4990151	**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 [1, 2, 4, 5, 7, 9]** | **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 with partial ethnicity from one or more groups mentioned above, post-test carrier risk depends on the exact mixture in the person's background.
- 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 1,700	[8]

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Usher Syndrome Type 3A is indicated for the detection of the N48K variant in the CLRN1 gene. This test is intended to be used to determine carrier status for Usher 3A 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 Usher syndrome 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	93%	[1, 4, 5, 7]
Finnish	<10%	[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. Adato A et al. (2002). "USH3A transcripts encode clarin-1, a four-transmembrane-domain protein with a possible role in sensory synapses." Eur J Hum Genet. 10(6):339-50.
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4. Fields RR et al. (2002). "Usher syndrome type III: revised genomic structure of the USH3 gene and identification of novel mutations." Am J Hum Genet. 71(3):667-17.
5. Herrera W et al. (2008). "Retinal disease in Usher syndrome III caused by mutations in the clarin-1 gene." Invest Ophthalmol Vis Sci. 49(6):2651-60.
6. Joensuu T et al. (2001). "Mutations in a novel gene with transmembrane domains underlie Usher syndrome type 3." Am J Hum Genet. 69(4):673-84.
7. Ness SL et al. (2003). "Genetic homogeneity and phenotypic variability among Ashkenazi Jews with Usher syndrome type III." J Med Genet. 40(10):767-72.
8. Scott SA et al. (2010). "Experience with carrier screening and prenatal diagnosis for 16 Ashkenazi Jewish genetic diseases." Hum Mutat. 31(11):1240-50.
9. Tian G et al. (2009). "Clarin-1, encoded by the Usher Syndrome III causative gene, forms a membranous microdomain: possible role of clarin-1 in organizing the actin cytoskeleton." J Biol Chem. 284(28):18980-93.

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	Usher Syndrome Type 3A report created.