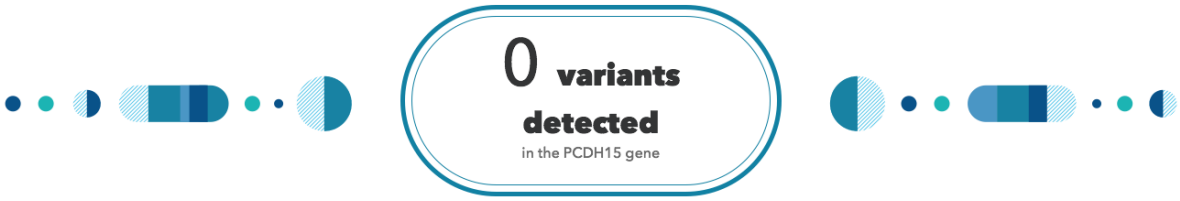
Usher Syndrome Type 1F

Usher 1F is a rare genetic disorder. It is characterized by deafness at birth, poor balance, and vision loss that worsens over time. A person must have two variants in the PCDH15 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.



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 R245X variant in the PCDH15 gene.
- To identify carrier status for Usher 1F.

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.

You are likely not a carrier.

This result is relevant for you because you have **Ashkenazi Jewish** ancestry.

We ruled out the most common variant for Usher 1F in people of Ashkenazi Jewish descent.

You still have a chance of being a carrier for Usher 1F.

You may still have up to a **1 in 1,600 chance** of carrying a variant not covered by this test.

See Scientific Details

About Usher Syndrome Type 1F

When symptoms develop

Symptoms typically develop at birth.

How it's treated

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

Typical signs and symptoms

- Deafness in both ears at birth
- Loss of vision beginning in childhood
- Poor balance
- Delays in walking

Ethnicities most affected

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

Read more at

Genetics Home Reference

GeneReviews

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.

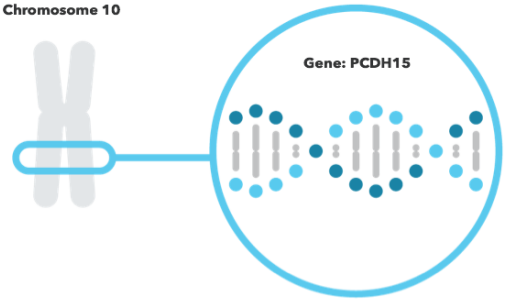
Learn more

Usher 1F is caused by variants in the PCDH15 gene.

PCDH15

The PCDH15 gene contains instructions for making a protein called protocadherin-related 15. This protein is found in the ears and eyes, where it helps cells stick together. Certain variants in PCDH15 prevent this protein from functioning properly. This disrupts the organization of cells in the ear and eye during development.

Read more at Genetics Home Reference



You have no variants detected by this test.

Variants Detected	View All Tested Markers
0	1

Marker Tested	Your Genotype*		Additional Information
R245X Gene: PCDH15 Marker: i5012804	**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** [1 , 2 , 3 , 4 , 5 , 7 , 8] | 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 1,600	[9]

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Usher Syndrome Type 1F is indicated for the detection of the R245X variant in the PCDH15 gene. This test is intended to be used to determine carrier status for Usher 1F 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.

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	91%	[2 , 4]

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 on our genotyping platform, or may not pass our testing standards.

References

1. Alagramam KN et al. (2011). "Mutations in protocadherin 15 and cadherin 23 affect tip links and mechanotransduction in mammalian sensory hair cells." PLoS One. 6(4):e19183.
2. Ben-Yosef T et al. (2003). "A mutation of PCDH15 among Ashkenazi Jews with the type 1 Usher syndrome." N Engl J Med. 348(17):1664-70.
3. Bonnet C et al. (2012). "Usher syndrome (sensorineural deafness and retinitis pigmentosa): pathogenesis, molecular diagnosis and therapeutic approaches." Curr Opin Neurol. 25(1):42-9.
4. Brownstein Z et al. (2004). "The R245X mutation of PCDH15 in Ashkenazi Jewish children diagnosed with nonsyndromic hearing loss foreshadows retinitis pigmentosa." Pediatr Res. 55(6):995-1000.
5. Cohen M et al. (2007). "The changing face of Usher syndrome: clinical implications." Int J Audiol. 46(2):82-93.
6. Keats BJB et al. (1993). "Usher Syndrome Type I"
7. Kikkawa YS et al. (2008). "Development of outer hair cells in Ames waltzer mice: mutation in protocadherin 15 affects development of cuticular plate and associated structures." Anat Rec (Hoboken). 291(2):224-32.
8. Rebibo-Sabbah A et al. (2007). "In vitro and ex vivo suppression by aminoglycosides of PCDH15 nonsense mutations underlying type 1 Usher syndrome." Hum Genet. 122(3-4):373-81.
9. Scott SA et al. (2010). "Experience with carrier screening and prenatal diagnosis for 16 Ashkenazi Jewish genetic diseases." Hum Mutat. 31(11):1240-50.