Pendred Syndrome and DFNB4 Hearing Loss

Pendred syndrome and DFNB4 are genetic disorders characterized by deafness and structural problems with the inner ear. Pendred syndrome is sometimes characterized by an enlarged thyroid. People with Pendred syndrome or DFNB4 most often have two variants in the SLC26A4 gene.

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 SLC26A4 gene.
- To identify carrier status for Pendred syndrome and DFNB4.

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** variants in other genes (FOXI1 and KCNJ10) that are also related to Pendred syndrome and DFNB4.

Important Ethnicities

- This test does **not** include a large fraction of SLC26A4 variants that cause Pendred syndrome or DFNB4 in any ethnicity.

You are likely not a carrier.

We ruled out the tested variants for Pendred syndrome and DFNB4.

These variants are rare in all ethnicities.

You still have a chance of being a carrier for Pendred syndrome or DFNB4.

We cannot estimate your chances because sufficient data is not available.

About Pendred Syndrome and DFNB4 Hearing Loss

When symptoms develop

Symptoms typically develop at birth or during childhood.

How it's treated

There is currently no known cure. Early intervention is recommended to teach alternative communication skills. Hearing aids or cochlear implants may treat hearing loss. Medication can treat low thyroid hormone levels.

Typical signs and symptoms

- Hearing loss at birth or in early childhood
- Abnormal inner ear development
- Enlarged thyroid
- Poor balance

Ethnicities most affected

These conditions can affect people of any ethnicity.

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.

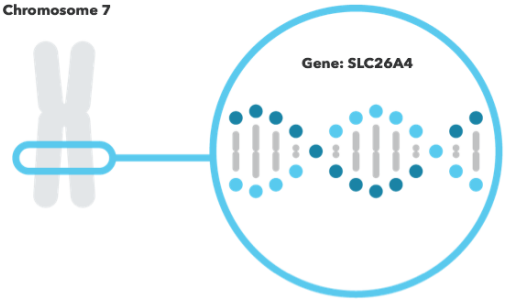
Learn more

Pendred syndrome and DFNB4 are most often caused by variants in the SLC26A4 gene.

SLC26A4

The SLC26A4 gene contains instructions for making a protein called pendrin. One of its known functions is to move molecules in and out of cells of the inner ear and thyroid. This process helps maintain the right balance of fluids in these cells. Certain variants in SLC26A4 disrupt this function.

Read more at Genetics Home Reference



You have no variants detected by this test.

Variants Detected	View All Tested Markers
0	6

Marker Tested	Your Genotype*		Additional Information
L236P Gene: SLC26A4 Marker: i5012616	**T** Typical copy from one of your parents	**T** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [3 , 4 , 14 , 15 , 16] | ClinVar
E384G Gene: SLC26A4 Marker: i5000003	**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 , 3 , 4 , 14] | ClinVar
T416P Gene: SLC26A4 Marker: i5012618	**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** [3 , 4 , 14 , 15 , 16] | ClinVar
V138F Gene: SLC26A4 Marker: i5000693	**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 , 4 , 5 , 15 , 16] | ClinVar
H723R Gene: SLC26A4 Marker: i5000002	**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** [6 , 10 , 13 , 16 , 17 , 18] | ClinVar
L445W Gene: SLC26A4 Marker: i5000696	**T** Typical copy from one of your parents	**T** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [3 , 7 , 8 , 11 , 16] | 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 Pendred syndrome and DFNB4 is the chance of still being a carrier for either of these conditions if you do not have the variants tested. This chance depends on how common it is to be a carrier for Pendred syndrome or DFNB4 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 Pendred Syndrome and DFNB4 Hearing Loss is indicated for the detection of six variants in the SLC26A4 gene. This test is intended to be used to determine carrier status for Pendred syndrome and DFNB4 in adults, but cannot determine if a person has two copies of a tested variant.

Special Considerations

- Symptoms of Pendred syndrome and DFNB4 vary in severity depending on which variants are causing the condition.
- This test does not include a large fraction of SLC26A4 variants that cause Pendred syndrome or DFNB4 in any ethnicity.
- There are currently no professional guidelines in the U.S. for carrier testing for these conditions.

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.

European	40 to 60%	[12]
Japanese	35 to 45%	[9]

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing for 292 samples with known variant status. 292 out of 292 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

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