Pendred Syndrome and DFN4B Hearing Loss (SLC26A4-Related)

Pendred syndrome and DFN4B are inherited conditions characterized by deafness and structural problems with the inner ear. Pendred syndrome is sometimes characterized by an enlarged thyroid. People with Pendred syndrome or DFN4B may have two variants in the SLC26A4 gene.

play+ce9das94eb, you do not have the variants we tested.

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

0 variants detected in the SLC26A4 gene

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 the condition, or you have any concerns about your results.

Intended Uses

- Tests for multiple variants in the SLC26A4 gene.
- To identify genetic status for Pendred syndrome and DFN4B.

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 (FOXL1 and RHO1D) that are also related to Pendred syndrome and DFN4B.

Important Ethicities

This test does not test for a large fraction of SLC26A4 variants that cause Pendred syndrome or DFN4B in any ethnicity.

You are likely not a carrier.

We ruled out the tested variants for Pendred syndrome and DFN4B.

These variants are seen in all ethnicities.

You still have a chance of being a carrier for Pendred syndrome or DFN4B.

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

About Pendred Syndrome and DFN4B Hearing Loss (SLC26A4-Related)

When symptoms develop

Symptoms typically develop at birth or during childhood.

Typical signs and symptoms

- Hearing loss at birth or in early childhood
- Abnormal ear or nose development
- Congenital hydantoin
- Poor balance

Ethnicities most affected

These conditions affect people of any ethnicity.

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 help hearing loss. Medication can help thyroid hormone levels.

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. Additional testing might be appropriate.

Support groups and resources

1. Action on Hearing Loss
2. Hear the World Foundation of the Deaf
3. American Society for Deaf Children
4. Hearing Loss Association of America

Learn more about hearing loss and deafness.

Learn more