

SLC26A4 (PDS) GENE TESTING

COULD MY HEARING LOSS BE CAUSED BY MUTATIONS IN THE SLC26A4 GENE?

SLC26A4 hearing loss is most often seen in a person with:

- Temporal bone abnormalities (observed on a CT scan or MRI of the inner ear) including enlarged (dilated) vestibular aqueduct and/or Mondini malformation
- Hearing loss that was found at birth or in early childhood
- Hearing loss that is severe or profound
- Thyroid enlargement (goiter) that appears during childhood or early adulthood
- A positive perchlorate discharge test (for thyroid function)

Although these are the most common characteristics of hearing loss due to mutations in SLC26A4, there can be variations, even within a family. SLC26A4 mutations are usually thought to cause Pendred Syndrome, a genetic condition associated with hearing loss and goiter (thyroid enlargement). However, mutations in the SLC26A4 gene can sometimes cause hearing loss in people who do not have any thyroid problems. Also, some persons with SLC26A4 hearing loss do not have temporal bone abnormalities and some can have mild or moderate hearing loss.

HOW IS THE SLC26A4 GENE TEST PERFORMED?

To perform the SLC26A4 gene test, a DNA sample is obtained and your SLC26A4 sequence is compared to that of the regularly occurring gene sequence to look for changes. Some laboratories may examine the entire coding sequence of the gene, while other laboratories may only search for the common mutations. There are 3 common mutations (L236P, 1001+1G>A, T416P) that cause over half of the cases of hearing loss associated with this gene.

HOW ARE THE RESULTS OF A SLC26A4 GENE TEST INTERPRETED?

There are three possible outcomes of a SLC26A4 test:

1. *No SLC26A4 mutations are detected.* If no mutations are found, and the entire coding sequence was analyzed, it is unlikely that the hearing loss is caused by mutations in the SLC26A4 gene.
2. *Two SLC26A4 mutations are detected.* If two identical mutations or two different mutations are found, and these mutations have been previously found to cause hearing loss, it can be assumed that the hearing loss is caused by these mutations in the SLC26A4 gene.
3. *Only one SLC26A4 mutation is detected.* If only one mutation is detected, interpretation can be difficult. It is possible that the test did not detect the second mutation or that the SLC26A4 mutation found may be unrelated to the hearing loss.

A catalog of the reported SLC26A4 mutations can be found in the Deafness Gene Mutation Database at <http://hearing.harvard.edu>.