

# COCH GENE TESTING

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

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*COCH* hearing loss is most often seen in a person with:

- Isolated (nonsyndromic) sensorineural hearing loss (initially worse at high frequencies)
- Hearing loss that begins between ages 15 and 65
- Hearing loss that progresses to anacusis (complete loss)
- Variable vestibular problems (e.g. dizziness, trouble with balance, etc.)
- A family history of dominantly inherited hearing loss

Although these are the most common characteristics of hearing loss due to a mutation in *COCH*, there can be variations, even within a family. Some individuals do not report vestibular problems and it is also possible that a family history may not be present. It should be noted that some people with *COCH*-associated hearing loss may initially be diagnosed with Menière's disease.

## HOW IS THE COCH GENE TEST PERFORMED?

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To perform the *COCH* test, a DNA sample is obtained and your *COCH* gene sequence is compared to that of the regularly occurring sequence to look for changes. To date, all mutations in this gene have been found in one particular region of the gene (exons 4 and 5). As such, most testing for this gene involves sequencing only this particular region.

## HOW ARE THE RESULTS OF A COCH GENE TEST INTERPRETED?

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Because hearing loss due to mutations in the *COCH* gene is inherited in an autosomal dominant manner, only one mutation is required to cause the hearing loss. Therefore, if one mutation is found, and this mutation has been previously shown to cause hearing loss in other persons, it is highly likely that the mutation is the cause of the hearing loss.

If no *COCH* mutations are detected, it is still possible that a mutation could be present in regions of the gene that were not tested, such as in other exons or noncoding regions of the gene. In addition, there are other genes that may cause a similar form of hearing loss. Consequently, if no mutation is found in this gene, it is possible that a mutation may exist in another hearing loss gene.

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