

# MITOCHONDRIAL GENE TESTING

## COULD MY HEARING LOSS BE CAUSED BY MUTATIONS IN A MITOCHONDRIAL GENE?

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There are many forms of hearing loss that can be caused by mutations in mitochondrial genes. Some of the more common forms include:

- Isolated (nonsyndromic) sensorineural hearing loss  
OR
- Hearing loss that began after antibiotic treatment  
OR
- Hearing loss that is associated with neuromuscular disease  
OR
- Hearing loss that is associated with diabetes

There are two mitochondrial genes known to cause nonsyndromic hearing loss (*12S rRNA* and *tRNA<sup>ser</sup>(UCN)*). There are several mitochondrial genes known to cause syndromic forms of hearing loss. For example, mutations in the *tRNA<sup>Leu</sup>(UUR)* and *tRNA<sup>Lys</sup>* genes can both cause hearing loss associated with neuromuscular diseases such as MELAS and MERRF. Mutations in the *tRNA<sup>Leu</sup>(UUR)* gene can also be associated with diabetes. All forms of hearing loss due to mutations in mitochondrial genes are inherited only from the mother and not the father. See the booklet for a more detailed description of mitochondrial inheritance.

## WHAT'S SPECIAL ABOUT THE A1555G MUTATION?

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There is a mutation called A1555G in the *12S rRNA* gene that makes a person more likely to develop hearing loss following treatment with a particular type of antibiotic called aminoglycoside antibiotics (for example gentamicin, neomycin, amikacin, tobramycin). It should be noted that people can lose their hearing due to treatment with aminoglycoside antibiotics even if they do **not** have this mutation. Also, a person with this mutation can develop hearing loss even with **no** exposure to aminoglycoside antibiotics.

## HOW ARE MITOCHONDRIAL GENE TESTS PERFORMED?

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To perform the mitochondria gene tests, a DNA sample is obtained and your mitochondrial gene sequences are compared to those of the regularly occurring sequences to look for changes. The majority of hearing loss due to mutations in mitochondrial genes is caused by a small number of specific mutations. As a result, many labs that offer tests for mitochondrial genes, only screen for the specific mutations and typically do not sequence the whole genes.

A catalog of the mitochondrial gene mutations that have been reported to cause hearing loss can be found in the Deafness Gene Mutation Database at <http://hearing.harvard.edu>.