



Deafness and myopia syndrome

Deafness and myopia syndrome is a disorder that causes problems with both hearing and vision. People with this disorder have moderate to profound hearing loss in both ears that may worsen over time. The hearing loss may be described as sensorineural, meaning that it is related to changes in the inner ear, or it may be caused by auditory neuropathy, which is a problem with the transmission of sound (auditory) signals from the inner ear to the brain. The hearing loss is either present at birth (congenital) or begins in infancy, before the child learns to speak (prelingual).

Affected individuals also have severe nearsightedness (high myopia). These individuals are able to see nearby objects clearly, but objects that are farther away appear blurry. The myopia is usually diagnosed by early childhood.

Frequency

The prevalence of deafness and myopia syndrome is unknown. Only a few affected families have been described in the medical literature.

Causes

Deafness and myopia syndrome is caused by mutations in the *SLITRK6* gene. The protein produced from this gene is found primarily in the inner ear and the eye. This protein promotes growth and survival of nerve cells (neurons) in the inner ear that transmit auditory signals. It also controls (regulates) the growth of the eye after birth. In particular, the SLITRK6 protein influences the length of the eyeball (axial length), which affects whether a person will be nearsighted or farsighted, or will have normal vision. The SLITRK6 protein spans the cell membrane, where it is anchored in the proper position to perform its function.

SLITRK6 gene mutations that cause deafness and myopia syndrome result in an abnormally short SLITRK6 protein that is not anchored properly to the cell membrane. As a result, the protein is unable to function normally. Impaired SLITRK6 protein function leads to abnormal nerve development in the inner ear and improperly controlled eyeball growth, resulting in the hearing loss and nearsightedness that occur in deafness and myopia syndrome.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- deafness and myopia
- deafness, cochlear, plus
- DFNMYP
- high myopia and sensorineural deafness
- high myopia-sensorineural deafness syndrome
- myopia and deafness

Diagnosis & Management

Genetic Testing Information

- What is genetic testing?
[/primer/testing/genetictesting](#)
- Genetic Testing Registry: Deafness and myopia
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3806275/>

Other Diagnosis and Management Resources

- Baby's First Test: Hearing Loss
<https://www.babysfirsttest.org/newborn-screening/conditions/hearing-loss>
- EyeSmart: Eyeglasses for Vision Correction
<https://www.aaopt.org/eye-health/glasses-contacts/glasses>
- GeneReview: Deafness and Myopia Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK269029>
- Harvard Medical School Center for Hereditary Deafness
<https://hearing.harvard.edu/>
- KidsHealth: Hearing Evaluation in Children
<https://kidshealth.org/en/parents/hear.html>
- MedlinePlus Encyclopedia: Cochlear Implant
<https://medlineplus.gov/ency/article/007203.htm>
- MedlinePlus Health Topic: Cochlear Implants
<https://medlineplus.gov/cochlearimplants.html>
- MedlinePlus Health Topic: Hearing Aids
<https://medlineplus.gov/hearingaids.html>
- MedlinePlus Health Topic: Newborn Screening
<https://medlineplus.gov/newbornscreening.html>

Additional Information & Resources

Health Information from MedlinePlus

- Encyclopedia: Cochlear Implant
<https://medlineplus.gov/ency/article/007203.htm>
- Health Topic: Cochlear Implants
<https://medlineplus.gov/cochlearimplants.html>
- Health Topic: Hearing Aids
<https://medlineplus.gov/hearingaids.html>
- Health Topic: Hearing Problems in Children
<https://medlineplus.gov/hearingproblemsinchildren.html>
- Health Topic: Newborn Screening
<https://medlineplus.gov/newbornscreening.html>
- Health Topic: Refractive Errors
<https://medlineplus.gov/refractiveerrors.html>

Genetic and Rare Diseases Information Center

- Deafness and myopia syndrome
<https://rarediseases.info.nih.gov/diseases/12844/deafness-and-myopia-syndrome>

Additional NIH Resources

- National Eye Institute (NEI): Myopia
<https://nei.nih.gov/health/errors/myopia>
- National Institute on Deafness and Other Communication Disorders (NIDCD): Auditory Neuropathy
<https://www.nidcd.nih.gov/health/auditory-neuropathy>

Educational Resources

- Center for Hearing and Speech: Types of Hearing Loss
<https://www.centerhearingandspeech.org/types-hearing-loss/>
- Centers for Disease Control and Prevention: Hearing Loss in Children -- Types of Hearing Loss
<https://www.cdc.gov/NCBDDD/hearingloss/types.html>
- KidsHealth: Auditory Neuropathy Spectrum Disorder
<https://kidshealth.org/en/parents/ansd.html>
- KidsHealth: Your Child's Vision
<https://kidshealth.org/en/parents/vision.html>
- Laurent Clerc National Deaf Education Center
<https://www3.gallaudet.edu/clerc-center.html>

- MalaCards: deafness and myopia
https://www.malacards.org/card/deafness_and_myopia
- My Baby's Hearing: Auditory Neuropathy
<https://www.babyhearing.org/auditory-neuropathy-spectrum-disorder>
- Orphanet: High myopia-sensorineural deafness syndrome
https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=363396
- University of Arizona Health Sciences: Myopia and Deafness
<https://disorders.eyes.arizona.edu/disorders/myopia-and-deafness>

Patient Support and Advocacy Resources

- Alexander Graham Bell Center for the Deaf and Hard of Hearing
<https://www.agbell.org/Default.aspx>
- March of Dimes: Hearing Loss
<https://www.marchofdimes.org/baby/hearing-impairment.aspx>
- National Association of the Deaf
<https://www.nad.org/>
- Royal National Institute of Blind People: Myopia and High Degree Myopia
<https://www.rnib.org.uk/eye-health/eye-conditions/myopia-and-pathological-myopia>
- University of Kansas Genetics Education Center Resource List: Hard of Hearing/Deafness
<http://www.kumc.edu/gec/support/hearing.html>

Clinical Information from GeneReviews

- Deafness and Myopia Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK269029>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28deafness+and+myopia+syndrome%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- DEAFNESS AND MYOPIA
<http://omim.org/entry/221200>

Medical Genetics Database from MedGen

- Deafness and myopia syndrome
<https://www.ncbi.nlm.nih.gov/medgen/929695>

Sources for This Summary

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Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3635725/>
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