Deafness and myopia syndrome

Description

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.

Learn more about the gene associated with Deafness and myopia syndrome

- SLITRK6

Inheritance

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

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: High myopia-sensorineural deafness syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C3806275/)

Genetic and Rare Diseases Information Center

- High myopia-sensorineural deafness syndrome (https://rarediseases.info.nih.gov/diseases/12844/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Catalog of Genes and Diseases from OMIM

- DEAFNESS AND MYOPIA; DFNMYP (https://omim.org/entry/221200)

Scientific Articles on PubMed

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

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Last updated November 1, 2015