A LOOK AT GENETIC HEARING LOSS AND THE IMPORTANCE OF GENETIC TESTING

Dive into the following

- Genetic protein deficiencies in congenital hearing loss
- Current management strategies
- The importance of genetic testing

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Genetic protein deficiencies in congenital hearing loss

1 IN 500 INFANTS ARE BORN WITH HEARING LOSS[1]

Genetically driven protein deficiencies account for >50% of congenital hearing loss cases[2]

***OTOF* and *GJB2* are 2 of the most common** causes of autosomal recessive, nonsyndromic, congenital hearing loss. Others include[2,3]:

- *STRC*
- *SLC26A4*
- *MYO15A*
- *CDH23*
- *TMPRSS3*
- *TMC1*

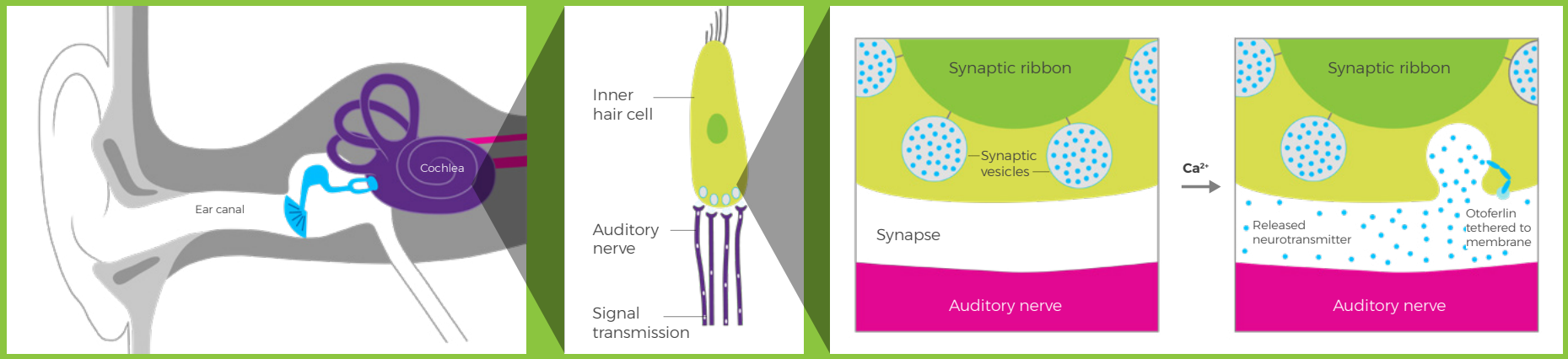
AN OTOFERLIN (*OTOF*) DEFICIENCY CAN CAUSE CONGENITAL HEARING LOSS[4]

The absence of functioning otoferlin due to a mutation in the *OTOF* gene can cause **auditory neuropathy spectrum disorder** (ANSD). Otoferlin deficiency is the most common genetically driven protein deficiency that causes ANSD.[5]

Infants with otoferlin deficiency often present with profound hearing loss despite the presence of inner ear sensory cells.[6]

of patients who inherit pathogenic variants in both alleles of the *OTOF* gene present with severe-to-profound hearing loss.[6]

Otoferlin enables communication between the sensory cells of the inner ear and the auditory nerve by regulating synaptic transmission[4]



The cochlea is located in the inner ear and contains several cell types required for hearing, such as inner hair cells

Otoferlin is found in the inner hair cells of the cochlea

Otoferlin allows for fusion of the synaptic vesicles containing neurotransmitter and the membrane of the inner hair cell

Once fused, the neurotransmitter is released and taken up by the auditory nerve

Current management strategies

ASSISTIVE DEVICES ARE THE CURRENT STANDARD OF CARE IN HEARING LOSS

Options for hearing rehabilitation currently include[7,8]:

- Cochlear implants (CI)
- Hearing aids (HA)
- Frequency modulated (FM) systems

Recent clinical research suggests that diagnosing the genetic protein deficiency of patients with congenital hearing loss may predict outcomes of CI, the current standard of care in children with profound hearing loss who do not benefit from hearing aids.[7]

The importance of genetic testing

GENETIC TESTING CAN HELP BOTH HEALTHCARE PROVIDERS AND FAMILIES

Benefits of genetic testing may include:

- Potentially leading to syndromic or progressive hearing loss identification
- Identifying possible management paths or future treatment paths
- Providing clarity that might help families understand the cause of hearing loss

Currently available genetic testing can detect the most common pathogenic variants associated with congenital hearing loss[9]

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