

Genetic Discoveries and Hearing Loss

Genetics is increasingly playing a role in the diagnosis and treatment of people with hearing loss. Researchers have discovered hundreds of genes responsible for hearing impairment. In fact, studies show that more than half of all hearing loss in people under 30 has a genetic origin.^{1,3}

According to the Centers for Disease Control and Prevention, about 30 percent of hearing loss present at birth (called congenital hearing loss) is due in part to syndromes for which hearing loss is a component. Syndromes are diseases with a specific set of symptoms. More than 400 syndromes may have hearing loss as a component. The other 70 percent of congenital hearing losses are non-syndromic—these hearing losses do not occur with other symptoms.⁴

Different Genes

There are more than 20 genes related to hearing loss.² Connexin 26, or GJB2, is the primary gene responsible for non-syndromic hearing loss.¹ Connexin is a protein that helps activate hair cells in the inner ear.² Presence of functioning hair cells facilitates normal hearing ability.

Other genes associated with hearing loss include Connexin 30 (GJB6), Connexin 31 (GJB3), and Myosin 7 (MYO7A). Most recently, DFNB6, also known as TMIE, was discovered to cause hearing loss.^{2,3}

Genes and Affected Populations

Hearing loss caused by the GJB2 gene occurs most prevalently in people of Northern

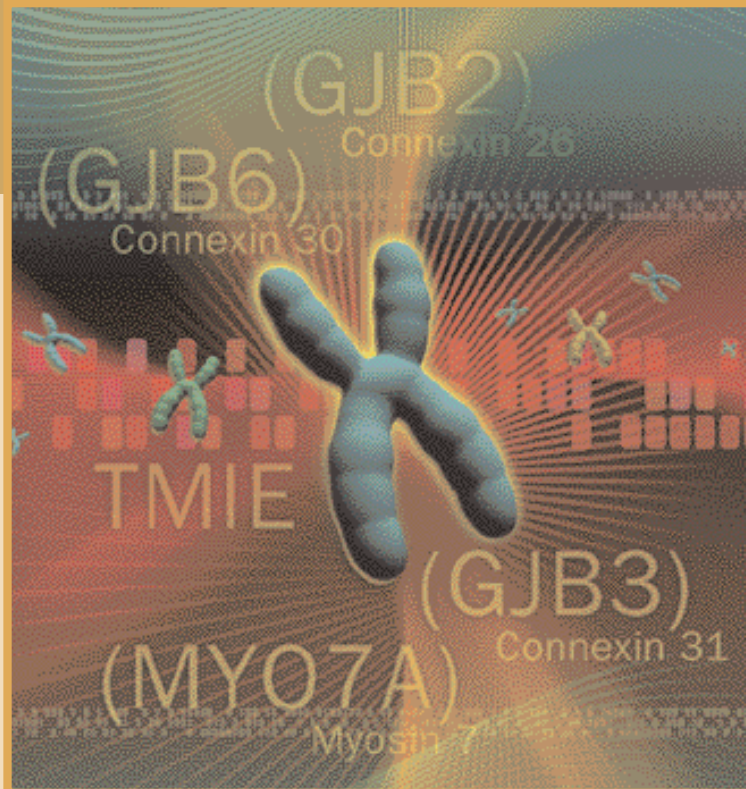


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European descent, particularly the Ashkenazi Jewish population.⁵⁻⁷ GJB6 causes a rare form of early-onset deafness, meaning it occurs after birth.² GJB3 is associated with a mutation causing hearing loss and a skin disorder. This gene has been found in two Chinese families.² MYO7A plays a part in the function of the outer and inner ear hair cells.²

DFNB6, or TMIE, was located in humans and mice in September 2002.⁸ Researchers think that mutations to this gene also affect hair cells in the inner ear.³ Researchers have followed several Indian and Pakistani families with inherited hearing loss. Mouse studies helped researchers confirm and target the TMIE gene in humans.³

Testing and Treatment Options

Genetic testing can aid in diagnosis of hearing loss, the discovery of how hearing loss was inherited, and even assist with treatment. Once an audiologist and/or primary care physician rule out other causes of impairment, a genetic counselor can help a patient understand the nature of their hearing loss.

References

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For More Information

Genetics Program at Gallaudet University
<http://depts.gallaudet.edu/genetics>

National Human Genome Research Institute
<http://www.genome.gov/glossary.cfm>

National Institute on Deafness and Other
Communications Disorders
www.nidcd.nih.gov