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A customized auditory rehabilitation based on the genetic etiology: A new auditory neuropathy spectrum disorder gene

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Cochlear glia-like supporting cells (GLSs) have been suggested to play an important role in the development and maintenance of an auditory system. However, genes that are primarily expressed in GLSs have never been clearly associated with progressive human deafness. Herein, we present a novel deafness locus and a new human auditory neuropathy spectrum disorder (ANSD) gene, *AUDITORIN* mainly expressed in GLSs. We specify p.R372X of *AUDITORIN* as a cause of the significant deterioration of speech perception in humans. *AUDITORIN* was shown to encode a novel, cationic channel, contributing to the enigmatic passive conductance current in GLSs. This current is abolished by gene-silencing or in p.R372X knock-in mouse in a dominant-negative fashion. Because the pathogenic effect of this variant was limited to GLSs, a cochlear implant that electrically stimulates spiral ganglion neurons was performed on the affected ANSD subjects and their speech perception ability was successfully restored. Our study elucidates an unprecedented pathological role of a cochlear GLSs by identifying a novel deafness gene and its causal relationship with ANSD, introducing a new disease entity called auditory gliopathy and paving the way for precision medicine in deafness.

Biography

Byung-Yoon Choi is both a geneticist and otologic surgeon. He has her expertise in genetic deafness and customized auditory rehabilitation based on genetic etiology. He pursues a discovery a new deafness gene and elucidation of a function of the genes. He is specially interested in application of the information obtained from the bench into clinics.

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