

Genetic diagnosis helps guide care of childhood hearing loss

January 19 2023



Credit: Unsplash/CC0 Public Domain

Advances in understanding the many different genetic causes of childhood-onset hearing loss indicate that genomic testing could assist in treatment planning, including optimal timing of treatment.

Even if a [child](#) with hearing loss did not receive a diagnosis from [genetic testing](#) a few years ago, very rapid recent progress in genomics makes re-

testing worthwhile, according to scientists in this field.

Hearing loss, usually stemming from sensory problems in the inner ear, affects about 1 in 400 newborns in the United States. About half of these children have a [genetic cause](#) for their hearing loss.

More than 120 genes and thousands of different genetic variants can lead to childhood hearing loss. Virtually all children with genetic hearing loss have a mutation or mutations in only one gene.

A recent study, reported Jan. 12 in [JAMA Otolaryngology-Head & Neck Surgery](#), examined a group of more than 400 youngsters with bilateral sensorineural hearing loss affecting both ears.

The researchers from UW Medicine and Seattle Children's Hospital sought to identify the genetic causes of hearing loss in this group and how each genetic cause relates to a specific type of hearing loss. They also evaluated whether the genetic cause of hearing loss was related to the success of treatment with a cochlear implant, a surgically placed electronic device that enables a person to sense sound through impulses transmitted to the brain.

The findings show that genetic testing is a valuable tool in determining prognosis for a child's hearing loss and in predicting how useful a cochlear implant could be for that child's understanding of speech.

"With [genetic diagnosis](#), it is possible to anticipate future hearing loss across sound frequencies and progression with age," the researchers noted in their paper. "This information can be integrated with current severity of hearing loss to decide on treatment."

The study was led by Ryan J. Carlson, an M.D./Ph.D. student in the Medical Scientist Training Program at the University of Washington

School of Medicine in Seattle.

It was conducted during his training with Mary-Claire King, the senior author and professor of medicine and genome sciences at the UW School of Medicine and with Jay Rubinstein, professor of otolaryngology and head and neck surgery at the UW School of Medicine.

"For more than half of pediatric patients, [genomic testing](#) can now identify a genetic cause for their hearing loss and often provide critical information on its clinical characteristics," explained Carlson.

The study participants included 449 children from 406 families. Genomic analysis resulted in genetic diagnoses for 210 of the 406 families, including 55 of the 82 families with more than one child with hearing loss and 155 of the 324 families with a single child with hearing loss. Rates of genetic diagnosis were similar for all ancestries in the [study group](#).

In this study population, the researchers detected variants responsible for hearing loss in 43 different genes. With one exception, each child's genetic diagnosis involved only one gene.

The severity of hearing loss, the sound frequencies most affected, and changes in hearing loss over time varied by gene, and, in some cases, by the type of variant within the gene. Children with causative mutations in the genes MYO6, OTOA, SLC2644, TMPRSS3, or the most severe mutations in the GJB2 gene had progressive hearing loss.

All of the children who had cochlear implants had better speech perception than before they received their implants. The degree of success of the cochlear implant varied somewhat based on the participant's genetic diagnosis.

Taking into account the age of the child at implant and the length of time the implant was in place, speech perception was highest for children whose hearing loss was due to mutations in the MITF or TMPRSS3 genes.

However, the researchers concluded that differences in success of [cochlear implants](#) related to the genetic diagnosis was not substantial enough to preclude a cochlear implant for any patient who otherwise met the implant criteria.

Ancestries of the study participants were 17 (4%) African/African American, 32 (8%) East Asian, 219 (54%) European, 53 (13%) Latinx, 16 (4%) South Asian, and 61 (15%) who reported more than one ancestry. Slightly more than half (51%) of the children in the study were female

The study was conducted from 2019 to 2022 at the otolaryngology and audiology clinics at Seattle Children's Hospital and the University of Washington, including the Center for Human Development and Disability.

DNA from the participants was analyzed by genomic sequencing and structural variant analysis in Mary-Claire King's genome sciences laboratory at the UW School of Medicine. Severity and progression of their hearing loss was measured through audiological testing, and success of their cochlear implant was evaluated by pediatric or adult speech perception testing.

The research team outlined several key conclusions from their findings:

- First, genetic testing for hearing loss can identify a genetic cause in most pediatric patients. Newborn hearing screening misses hearing loss in about one-third of affected children, because the

hearing loss is not yet detectable at birth. Genetic testing could resolve that gap.

- Second, technologies for genomic analysis are improving rapidly. If a child has already had genetic testing, with negative results, re-testing may find a genetic cause that was previously missed.
- Third, genetic testing can identify syndromic forms of hearing loss and lead to necessary referrals of children to key specialists.
- Finally, genetic diagnoses can be more effectively used to inform precision treatment for childhood hearing loss. Once the expected clinical characteristics of a certain genetic cause is known, that information can be used to evaluate prognosis and guide treatment.

Overall, the researchers emphasized, "Detection of hearing loss at the earliest possible age is important for cognition and social development, regardless of management approach."

Carlson noted that the study had a direct positive impact for its participants.

"We were able to provide genetic testing for over 400 families with [hearing](#) loss in this one study. Many participants received new genetic diagnoses that were important to their care," he explained. "I am proud to have directed a project that provided both direct benefits for those involved and indirect benefits for all patients with [hearing loss](#)."

More information: Ryan J. Carlson et al, Association of Genetic Diagnoses of Childhood-Onset Hearing Loss With Cochlear Implant Outcomes, *JAMA Otolaryngology–Head & Neck Surgery* (2023). [DOI: 10.1001/jamaoto.2022.4463](https://doi.org/10.1001/jamaoto.2022.4463)

Provided by University of Washington School of Medicine

Citation: Genetic diagnosis helps guide care of childhood hearing loss (2023, January 19)
retrieved 3 May 2024 from

<https://medicalxpress.com/news/2023-01-genetic-diagnosis-childhood-loss.html>

This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.