Clinical Trial Results: What We Know So Far

Safety and Side Effects

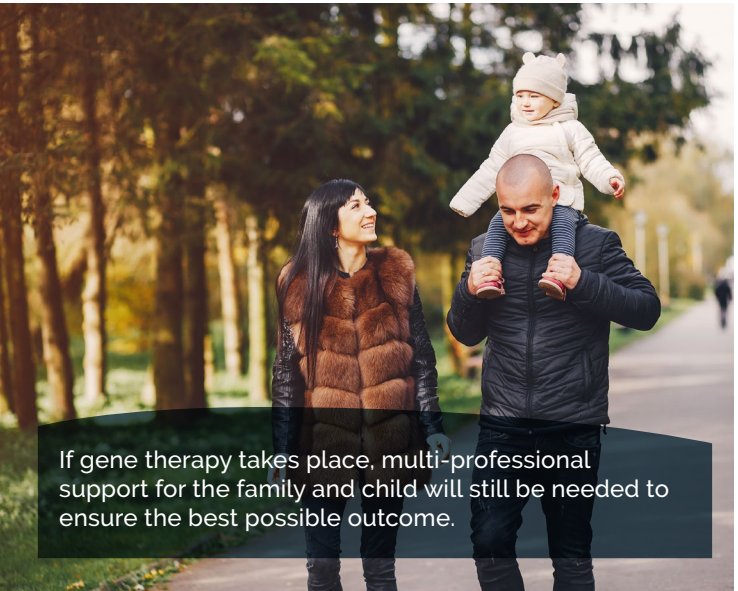
- So far, the treatment has been well tolerated, with only possible mild side effects, such as a mild fever.
- Children will be monitored to better understand long-term safety and hearing progress.

Changes in Hearing and Listening

- Initial hearing improvements may be seen within 30 – 90 days after surgery.
- These changes happen gradually, and the amount of improvement can differ from one child to another.
- Variables such as the surgical approach, the child's age, the residual inner ear function, or the components of the gene therapy may contribute to timing and the amount of improvement in hearing.
- Some children may still need a hearing aid or cochlear implant after gene therapy to help them ensure access to sound.

Progress in Listening Skills

- For some children, whether treated in one or both ears, progress in auditory development and understanding of speech has been shown. More long-term research is needed to understand what outcomes to expect for each child.



If gene therapy takes place, multi-professional support for the family and child will still be needed to ensure the best possible outcome.

What Parents Can Do to Support Their Child's Development

Don't Delay Access to Sound

- Early sound input is essential for brain development and spoken language. If the brain does not receive sound in the first years of life, some areas may not fully develop. Gene therapy for hearing loss holds real promise, but it's still in clinical trials. In the meantime, it's important to ensure a child gets the best possible access to sound as early as possible.

Develop Early Communication Skills

- Obtain support to develop early parent – child interaction to encourage communication and spoken language.

Track Progress Over Time

- Regularly monitor a child's hearing and language development to ensure they're getting the right support and to make changes, if needed.

Explore Genetic Testing

- Results may offer insight into the cause of hearing loss and how hearing may change over time and whether future treatments, like gene therapy, may be an option. A genetic counsellor can also help explain the results.

Connect with Other Families

- Sharing experiences and resources with other parents can be really helpful and can help decision-making and in managing expectations.

Stay Updated on New Research

- Stay up to date about research and clinical trials, including gene therapy, to keep informed about current and future treatment options.

This leaflet developed by:

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An Introductory Guide to

Genetic Hearing Loss, Genetic Testing, & Gene Therapy

Newborn hearing screening identifies hearing loss early, enabling children to receive timely access to sound amplification, and family support.

For many children, genetic testing can help identify the cause of hearing loss - if it's hereditary, part of a broader condition, or due to other factors.

There is growing interest in gene therapy, which aims to improve hearing by targeting specific genetic causes of hearing loss in the inner ear.

Genes and Genetic Testing

- In about 80% of children born with hearing loss, the cause is genetic.
- Genes contain the genetic code that directs how the body develops and functions. If a gene changes, it can affect how the body functions - including hearing.
- Genetic testing may help identify if a child's hearing loss is caused by a change in a gene.
- Knowing the cause helps parents and the child's care team make more informed choices for the child.
- While genetic testing is not yet available everywhere, it is important to think about future options for a child, including potential gene therapy.

Targeted Treatment Based on Genetics: Gene Therapy

- Over 150 genes are known to cause hearing loss on their own, and more than 400 genetic conditions include hearing loss as one of their features.
- Gene therapy must target the specific gene involved. Gene therapy developed for one gene won't work for other genetic causes of hearing loss.
- Right now, gene therapy is being studied in humans for one gene: OTOF.
- The OTOF gene produces otoferlin, a protein which is essential for sending sound signals from the ear to the brain. OTOF-related hearing loss is rare, accounting for only 3–8% of cases in children born with hearing loss that is not part of another medical condition.
- Gene therapy for OTOF is still only available through research trials in several countries.
- Gene therapy for OTOF only targets the inner ear. It does not change your child's entire genetic code, so the genetic condition can still be inherited by future generations.
- Researchers are developing gene therapies for other causes of genetic hearing loss, but these are still experimental.

FROM TESTING TO TREATMENT: THE GENE THERAPY PROCESS

Genetic Testing

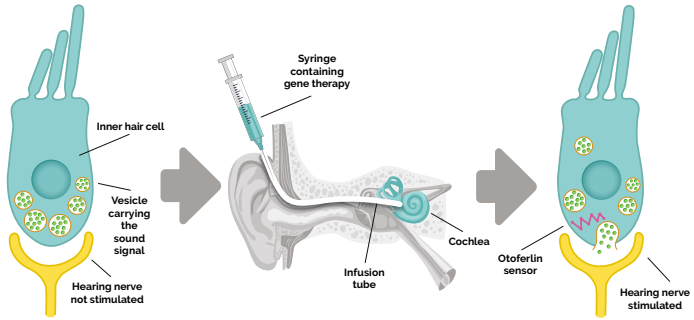
- A sample of a child's saliva or blood is taken and sent to a lab to look for changes in genes that are known to cause hearing loss.
- If results indicate a genetic cause of deafness, gene therapy may be an option. The child's doctor or genetic counsellor will explain the results to the family and help guide the next steps.

Eligibility and Timing of Gene Therapy

- Currently, children of various ages are being included in clinical trials. However, the greatest possible benefit is expected to be in the youngest, when the brain can develop hearing and spoken language more easily from sound input.
- While the current gene therapy trials in children are focused on the OTOF gene, there is ongoing research to explore gene therapy in adults targeting other genes associated with hearing loss.
- Gene therapy is likely to only be possible in ears that do not already have a cochlear implant. This is because after a cochlear implant is inserted, the inner ear forms scar tissue which may block the gene therapy from working.
- In the future, gene therapy is likely to be considered as the first step for some children.
- The use of hearing aids before gene therapy will be considered by the care team.

Otoferlin Gene Therapy: Before and After

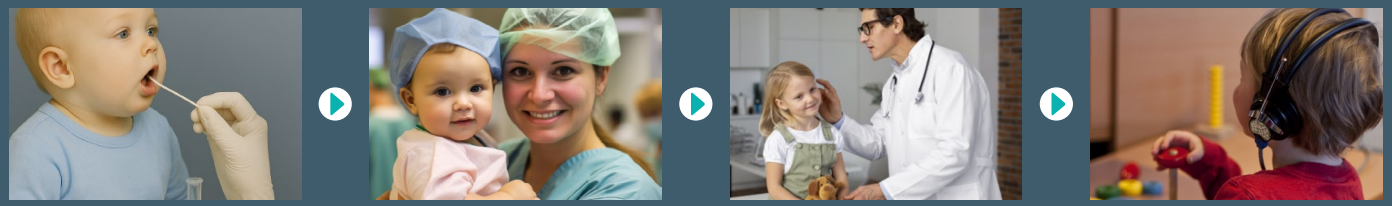
Before treatment, inner hair cells can't send signals to the hearing nerve.
Gene therapy adds otoferlin *(in pink)*, enabling the stimulation of the hearing nerve.



Surgical Process for Gene Therapy

- A specially trained ear surgeon places the treatment directly into the cochlea under general anaesthesia.
- Corticosteroids are usually given for a short time before and after surgery to lower the body's immune response to gene therapy. This helps the gene therapy be more effective.
- During surgery, the doctor creates a pathway to the cochlea to deliver the gene therapy directly to where it is needed.
- Using a special tool, the doctor places the gene therapy into the cochlea. The healthy copies of the gene are transported by a tiny virus that safely carries them to the right place, allowing the ear to make proteins needed for hearing.
- Surgery typically takes two hours, followed by a short hospital stay for monitoring. As with any surgery, there are risks, but these are generally low and will be discussed by the child's care team with the family. Follow-up visits will check healing and how well the treatment is working.
- Gene therapy is designed to be a single treatment; Once the specific cells receive the correct gene, they can continue working for life.

A Child's Journey: A Step-by-Step Overview



Genetic testing | Gene therapy injection | Monitoring health after surgery | Monitoring hearing progress after surgery