A Parent's Guide to Exploring Genetic Testing

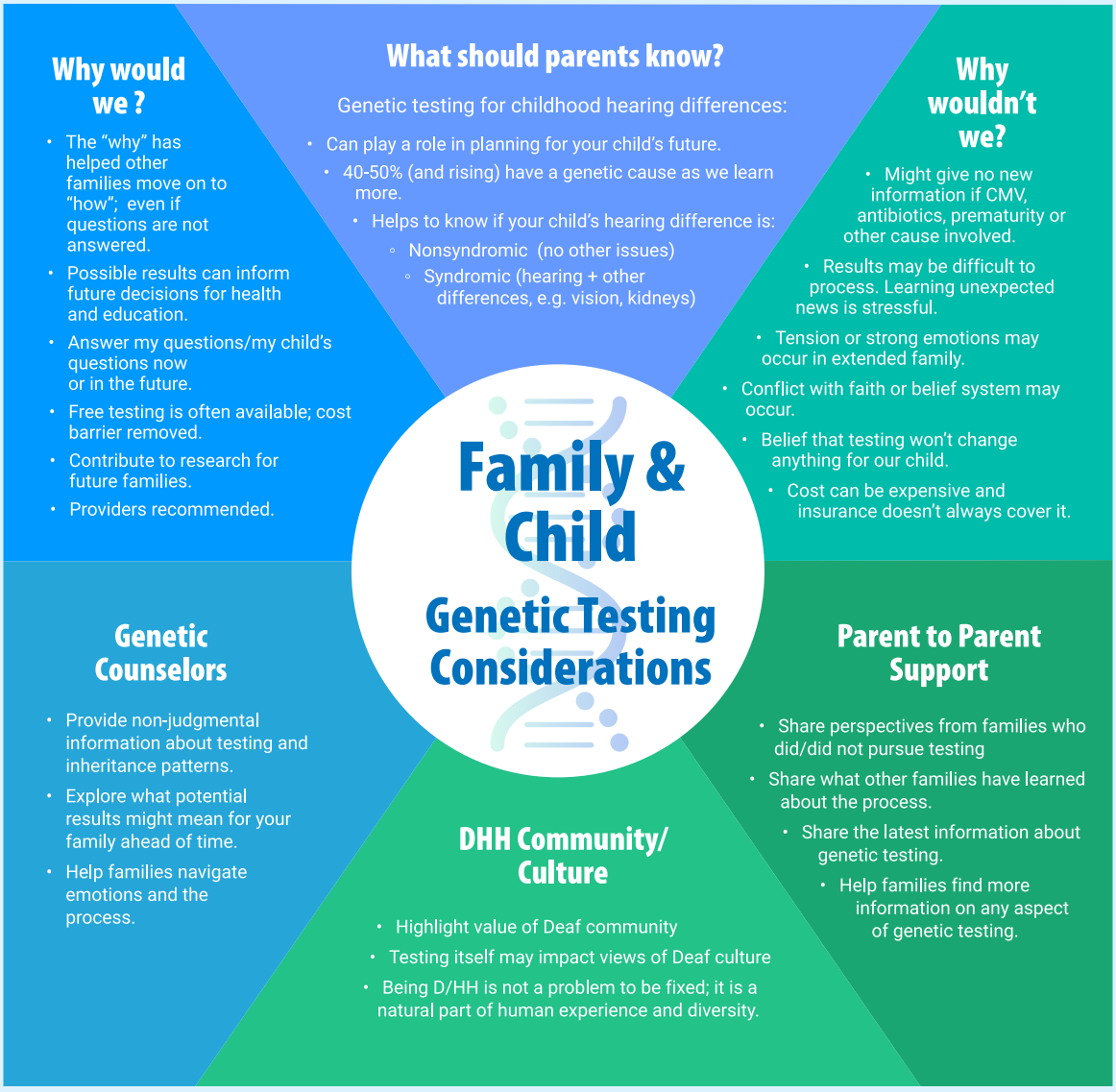
Making Informed Family Decisions

"What works for your child is what makes the choice right." ™

HANDS & VOICES™

This project was created by Hands & Voices, Headquarters May 2025 with the support of funding from Lilly.

Contents



About this Guide

This guide was developed by Hands & Voices, with input from hearing parents, deaf parents, professionals, genetic counselors, and deaf/hard of hearing individuals and community members. It is our hope that this will support your efforts in your journey of raising your child who is deaf/hard of hearing. For more information, contact parentadvocate@handsandvoices.org

What should we think about when deciding about genetic testing?

Introduction

Our goal with this guide is to help you learn about other parents' questions and concerns, about the professionals who can help you with your decision-making, and to help you gain confidence in your role as your child's best advocate, specific to genetic education and testing. From decades of experience working with families, we know that families given access to timely, accurate information from professionals and family leaders are empowered to make good decisions as experts in their own children. This guide, written by parents of children who are deaf/hard of hearing, some of whom are deaf/hard of hearing themselves, who are familiar with the latest research and knowledge in the now fast-changing world of genetic testing, is intended to give you a head start in your genetics education journey.

First, a little background

Becoming a parent brings a whole world of new experiences—and if your baby has just been identified as deaf or hard of hearing, you might be feeling overwhelmed, unsure, or full of questions. You're not alone. Many families have walked this road before you, and there are resources and support systems in place to help.

Where does genetic testing fit into the early part of your journey? It's important to understand the systems of care you are moving through to make good decisions about what/when/where to do genetic testing with your child.

From the day a baby is born, a series of events are set in motion: all babies should have newborn hearing screening to identify potential hearing differences and enter into early intervention. Often without even knowing it, families move through a system across the U.S. called the Early Hearing Detection and Intervention (EHDI) program that has been established with timelines (learn more here: called the 1-3-6 milestones). EHDI isn't about checking boxes or fixing something that's "wrong." The real goal is to make sure your baby has early access to language, communication, and connections--so they can thrive in every way. EHDI programs are also about supporting you as a parent. You'll meet professionals,

but just as importantly, you can connect with other parents who've been there and can help you feel less alone.

Making decisions for your child who is deaf/hard of hearing requires an incredible amount of research, education, networking, resources, and time. With all the decisions families often face early on in their child's life, it's no wonder parents can feel overwhelmed and need support. Deciding whether or not to do genetic testing is a part of the decision process during this time. While offering genetics consultation is recommended in the Joint Committee on Infant Hearing recommendations in 2019, a referral to genetic counseling or testing may or may not be offered to you, depending on where you live. Some families are offered genetic testing in an attempt to learn why their child has a hearing difference. If you have not been offered genetic testing, reach out to your medical team to discuss this.

But here's the key: It's your choice. You should never be pressured to get genetic testing—and if you do choose it, you deserve clear, compassionate support to understand what the results mean. No matter what results show, they don't define your baby's future. Your child is full of potential, and their hearing status is just one part of who they are. Hands & Voices has a motto, "What works for your child is what makes the choice right." Trust your instincts, ask questions, lean on support groups, and know that as you move through the considerations of whether to test or not, you can make the decision that is right for you.

What Should Parents Know About Genetic Testing?

Let's look at the basics of genetic testing.

Hearing loss can sometimes run in families, and genetic testing helps us understand if a particular child's hearing difference is inherited. Think of our genes like a set of instructions that tell our bodies how to grow and work. Sometimes, a tiny change in these instructions leads to hearing loss. Genetic testing is a simple medical test involving a blood or saliva sample that looks at your child's DNA to find out if changes in certain genes might cause changes in your child's hearing levels.

Genetic testing can play a key role in understanding why a child is deaf/hard of hearing. However, sometimes those results show that your child's hearing difference is not genetic or that particular genes don't have specific testing… yet.

Still, genetic causes for hearing differences are common. About 50-60% of hearing differences in children are caused by genes.

Types of hearing loss can be:

- Non-syndromic (in other words, only hearing is impacted). For example, the GJB2

gene instructs a protein called Connexin 26, a common genetic cause of hearing loss without a related syndrome.

- Syndromic: Hearing differences occur along with other conditions, like vision changes, kidney, heart function, changes in cognition (thinking skills) and/or muscle control. Examples: Usher's, Pendred, or Branchio-Oto-Renal syndromes.

The results from genetic testing may help:

- Determine the cause of the hearing difference.
- Understand if hearing might improve, stay the same, or get worse (progress).
- Plan for future children—understand the potential of hearing loss or other conditions related to genetics.
- Connect families with support and early treatment options, especially for syndromic conditions that involve other body functions.
- Sometimes, a known cause helps your family make informed decisions about future directions, education, technology, and proactive supports that can help your child thrive.

Differences in hearing levels can also be caused by infections, certain medications, or prolonged exposure to loud noises. In many cases, the cause of hearing differences may not be clear, even after genetic testing.

Is it helpful for everyone?

Not always. Sometimes the test doesn't find an answer, or it finds a gene change that doctors don't fully understand yet. But even then, the information might be useful later as science learns more. More and more of the over 150 genes that can affect hearing can be tested for each year.

Is it safe?

Yes! The testing itself is safe and easy. It involves a small sample of saliva or blood. There can be a long wait to get an appointment with the genetics team; testing results tend to

> ***When doing genetic testing, you need to know what you are looking for, what you might find that you are not looking for, and why you want to know. If you have these questions very clear in your mind, you should do it. If not, don't even open that box because you will feel disappointed, discouraged, and it brings extra stress.***
>
> ***· Parent of DHH Plus child***

arrive faster now. The results can bring peace of mind, but they can also bring up tough emotions. This guide will help you process reasons why you may or may not decide to do genetic testing.

Parents of Deaf/Hard of Hearing Plus Children (DHH Plus)

Many decisions are magnified for families with children with multiple healthcare needs, including the decision of whether to get genetic testing for their child.

If you know or suspect that your child has additional medical concerns in addition to his/her hearing loss, genetic testing may help you to know what additional diagnoses to watch for in the future. Being aware of potential medical concerns before they arise can lead to fast interventions and improved outcomes. For some families experiencing multiple health care needs, however, finding out additional potential concerns (or not finding out any new information at all) can be overwhelming. When dealing with multiple diagnoses, doctor, and therapy appointments, and all the responsibilities that come along with raising a DHH Plus child, sometimes families can find themselves in survival mode. Some decide to take on one thing at a time, delaying or deciding not to do genetic testing.

- **Will results alter our decision-making?** One thing DHH Plus families need to consider is whether certain diagnoses could alter their decision-making for their family. One example of this may be the choice of what communication mode to use. If receiving a diagnosis of Usher's Syndrome, for example, would that change what communication mode you pursue if you know your child is expected to lose functional vision at some point? Treatment initiated early for Usher Syndrome Type One can delay the onset of blindness and bring considerations of cochlear implant use earlier in the process if the syndrome is known earlier.
- **Moving:** Other families have made life-changing decisions such as moving their family to a different area for services and support better suited to genetic results and potential outcomes.
- **Siblings and Extended Family:** Additionally, consider how a genetic diagnosis may affect siblings or family members. Some genetic conditions don't reveal themselves at birth. Testing could potentially reveal a medical condition or risk for additional family members. This risk is greater with DHH Plus families because we already know there is more than one medical condition at play.

> ***"Right now, we don't believe there is a genetic cause. We need to focus on helping her develop. We also don't want to worry about future markers that MAY develop into problems that also may not. We may change our minds in the future."***
>
> ***· Parent of a toddler with a hearing difference, without a visible syndrome***

- **Future Planning:** Our DHH Plus kids often require a variety of supports and resources. Genetic testing can help with family planning to evaluate if a family has the capacity to raise another DHH Plus child, if the possibility exists for future children or grandchildren.
- **Belief system:** When evaluating cultural considerations, beliefs may preclude genetic testing for hearing loss but may be acceptable when testing for other medical concerns. How will your decision align with your family's religious and/or cultural beliefs?
- **Worrying about results:** When a child is DHH Plus, there is a possibility that genetic testing may reveal scary, dangerous, or even a fatal prognosis. This can be very traumatic, and some families may rather not know this information in advance, choosing to enjoy their child in the moment. These are very intense decisions. All feelings and decisions are valid in this circumstance.
- **Medical Trauma:** Another consideration for medically complex children to have genetic testing is the risk of medical trauma. For our DHH Plus children and their parents, medical trauma is a true concern due to increased exposure to medical procedures and unfamiliar professionals. Medical trauma can lead to fear and anxiety and affect the child's (or the parent's) physical, emotional, and behavioral health. If a family can avoid even one medical appointment or procedure, they may opt not to pursue voluntary testing.

Why Wouldn't We?

Families list these concerns about genetic testing, which may or may not be true for your child's situation. (Please read these along with reasons why some families have chosen genetic testing.) Families listed these concerns about genetic testing:

- **No Clear Answers:** Testing doesn't always find the cause of hearing differences. Hearing loss may result from infection (like cytomegalovirus from birth or cCMV), use of antibiotics, prematurity, or have an as-yet-unknown cause not addressed through genetic testing.
- **Judgment or Tension:** Results can take months to arrive. Results might create disagreements or grief within the family or with others.
- B**elief It Won't Help:** Some families feel testing may not change their child's care or decisions about future children.
- **Difficult Results:** Learning unexpected or hard-to-hear news can be stressful. Genetic testing can cause strong emotions in parents and the extended family.

> ***"I don't know what genetic testing might mean for future stigma. Will my child be able to get insurance coverage? I also don't want to have preconceived ideas about how he will develop. Let us bond first.***
>
> ***· Parent of a newborn with multiple issues***

- **Moral Concerns:** Questions about "playing God." Does testing feel right? "As a family, we don't want our child to feel 'less than' and we want them to know we fully welcomed and loved our child as they are from day one." What about my child's privacy?
- **Cultural Concerns:** We consider ourselves members of the Deaf community and/or other cultures and are concerned about where genetic testing will lead. (Genetic screening of embryos concerns us.)
- **Cost:** Testing and follow-ups can be expensive. Insurance doesn't always pay. (Note that free testing or grants are often available.)
- **Parental Guilt:** Worry that results might lead to feelings of blame or responsibility.
- **Future Consequences:** A worry that genetic results may be used against a child as a reason to deny insurance or care, as a pre-existing condition, or something we can't foresee?

Why Would We?

Families list these reasons why they have chosen genetic testing, which may or may not be true for your child's situation. Read these along with reasons why some families have not chosen to pursue genetic testing.

- **Understanding "Why":** Finding out the cause of why your child is deaf or hard of hearing can bring clarity.
- **Plan for the Future:** Results will help guide decisions about medical care, education, and family planning.
- **Be Better Prepared:** Finding out if there are other needs/syndromes: looking at "Deaf/HH Plus" concerns when hearing is not the only issue. (See more DHH Plus considerations in this Guide.)
- **Answer Your Child's Questions:** Many children are curious about their hearing difference or will be as they come into adulthood.
- **Contribute to Research: Y**our family's testing can add to medical knowledge that helps others. While testing may not help my child, it may help others with improved treatments in the future.

> ***"Our daughter was born hearing. Her father is deaf. She passed a preschool hearing screening at age three. When I was pregnant with her brother, we learned about Connexin 26 and that my husband's hearing difference was due to this genetic cause. We decided to test all of us. Our baby girl had two changed genes. She was born hearing… or was she? We had her hearing tested, and she had a mild to moderate loss. How long would this have gone unnoticed if we had not tested?"***
>
> ***· Parents, one deaf, one hearing***

> *I'm Deaf and I'm proud of who I am. I don't see the need to know more than that.*
>
> · *Deaf individual*

- **Connect** with family empowerment groups specific to a child's needs.
- **Offering Peace of Mind:** Genetic testing can sometimes answer lingering questions, help families make informed choices, and prepare for what's ahead.
- **Follow Recommendations from Providers:** "Because my provider told me to." Some families pursue testing because a doctor or specialist suggested it. We hope families have a greater level of informed consent for both medical and educational decisions.

Learning from the Perspectives of Others

Hearing from different perspectives can help you decide if genetic testing is right for your family:

- **Medical Professionals**
  - Offer insights into the cause of hearing differences and whether related health conditions need attention.
  - Assist in explaining results in understandable terms, helping families talk about genetic risks across generations while encouraging open dialogue.
  - Help families create tailored care plans based on test results.
  - Provide Up-to-Date Resources and Research Opportunities and direct families to trustworthy information and clinical trials and studies that could inform or improve care.
- **Deaf Culture/Community**
  - Highlight the value of deaf identity and concerns about how genetic testing might impact views on Deaf culture.
  - Share personal stories related to the Deaf individual's decision-making process regarding genetic testing or their own experience with getting genetic testing results.
  - Stress that deafness is not a "problem" to be fixed but a natural part of human experience and diversity.
  - Encourage families to think beyond medical risks and consider whether they see deafness as a negative outcome or as a different way of being.

> *We have found that we can hold two things at the same time, our identity about seeing deafness as part of who we are and also pursuing medical information at the same time.*
>
> · *Deaf individual*

> *We wanted to understand why, but it wasn't easy to decide. Learning from others helped.*
>
> *· Parent of a Deaf/Hard of hearing Child*

- Bring ethical depth to the decision, highlighting the impact on the dignity and visibility of Deaf people.
- Learning directly from Deaf people may reduce fear or anxiety about raising a deaf child and encourage a more open-minded, informed decision.

- **Parents and Parent Groups**
  - Emotional Support and Understanding:
    - Share experiences running the gamut of curiosity, guilt, helping others, and preparation.
    - Help normalize the experience and reduce feelings of isolation.
    - Provide validation through hearing stories which may affirm a parent's thoughts and feelings, offering comfort in a tough decision-making process.
    - Parents can share reasons why they did or didn't pursue testing for you to consider in light of your own family journey.
  - Firsthand Insights and Practical Advice
    - Real-world perspectives (what the process was like, how long it took, how their child reacted, what they learned).
    - Questions to ask, including questions a new parent might not think to ask.
  - Informed Decision-making
    - Discuss the pros and cons: benefits, potential limitations, or emotional impacts of testing.
    - Hear a variety of outcome stories: positive, neutral, or unexpected, to help parents make a more informed and personalized decision.
  - Navigating the System
    - Resources and referrals: parents/parent groups can often share recommendations or financial aid options.
    - Advocacy: Parents can learn how to better advocate for their child's needs from other parents.
  - Ongoing Community
    - Continued support after the decision is made or testing is completed when parents/groups remain a place of connection, sharing, and growth.
    - Celebrating milestones where progress and small victories can be celebrated by those who truly understand the journey.

- **Genetic Counselors**
  - Provide non-judgmental information about genetic testing.
  - Answer questions about what may and what may not be genetic symptoms already present in a baby or child or through family history.
  - Help individuals understand inheritance patterns from height or eye color before moving onto more sensitive topics.
  - Help you understand beforehand what potential results might mean for your family.
  - Help you understand the types of genetic testing available, including comprehensive testing with parallel sequencing.
  - Help families navigate emotions and decisions without pressure.

> ***Our job is to support families in making the choice that is right for them.***
>
> *· Genetic Counselor*

Questions to ask the Genetics Team*

Questions you may want to ask your genetics team to help you decide whether or not you want to pursue genetic testing:

1. What can genetic testing tell me about my child's hearing difference? What are some common genetic causes of hearing differences?
2. Is a comprehensive gene panel being used? How many genes are being tested?
3. How can genetic testing help my child?
4. Who will go over the testing process and the final results with me?
5. What will the results of genetic testing potentially tell me? Can we prepare ahead of time for what the results might look like?
6. Can the results of genetic testing tell me if my child's hearing will change over time?
7. What is the process for genetic testing? How is my child's privacy assured?
8. Why is it important to know my family's history of hearing differences and what type they had? How is it inherited?
9. If no one in my extended family has a hearing difference, how can it be genetic?
10. Where can I learn more about genetic testing?
11. Where can I connect with other families who have Deaf or Hard of Hearing children to learn about their decisions around genetic testing?

12. Where can I connect with Deaf community members about their decisions and experiences with genetic testing?

After testing:

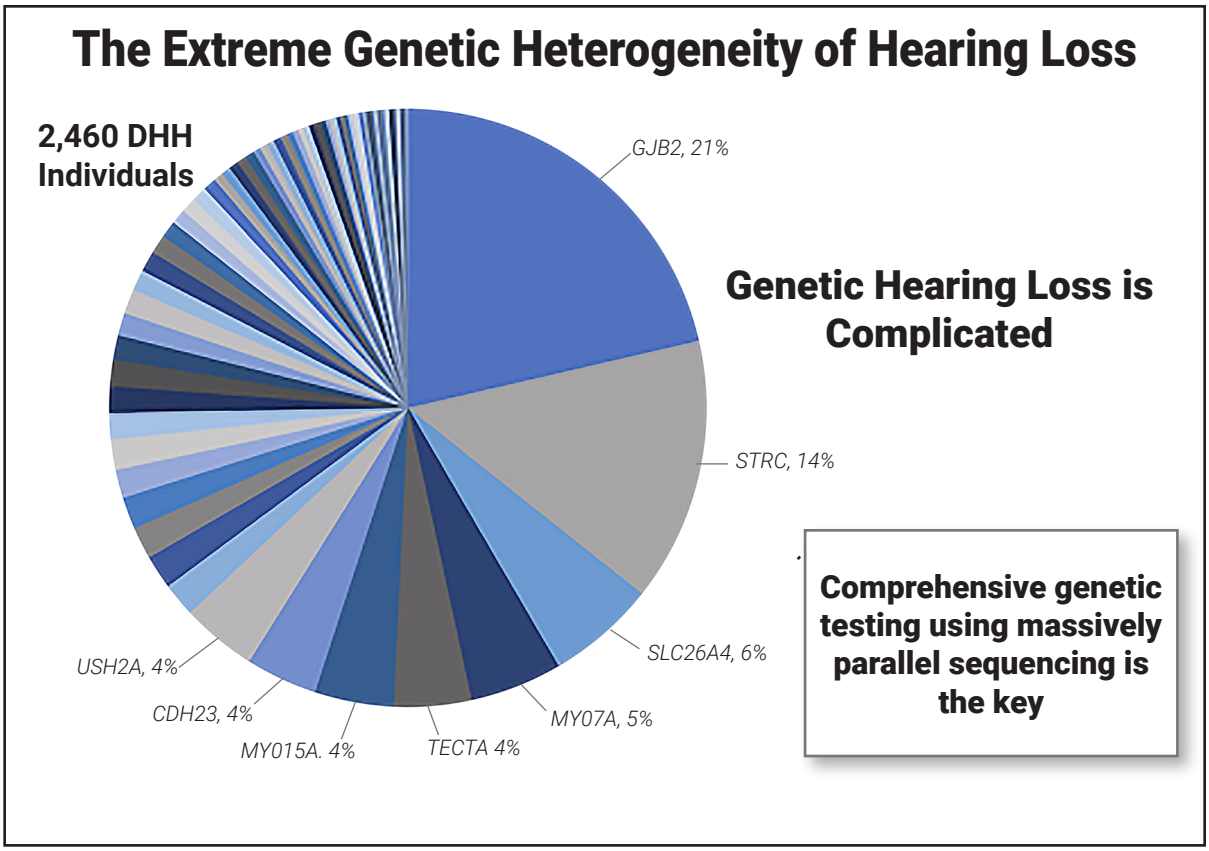
1. How do other partners and families process their emotions after this news?
2. What does this mean for future children or grandchildren?
3. What common concerns do you hear from families about similar results? Could there be additional results in the future?
4. Should I share test results with other members of my family? Could others in my family have children with hearing differences or other conditions? How do you recommend sharing this?
5. This is what I understood you to say… is that correct?
6. How can I meet other families with my child's genetic condition?
7. How can I meet adults with my child's genetic condition?
8. How can these results help with health care and educational decisions for my child?
9. What follow up do you recommend? Where can I learn more about this result?

*Note that genetic testing can also be ordered by ENTs/Otolaryngologists and audiologists in settings where there is a long wait for the genetics specialist.

Conclusion: Trusting Your Journey, Embracing Informed Choices

As you reflect on the many insights shared in this guide, remember there is no single "right" answer when it comes to genetic testing. Families are diverse, and so are their values, beliefs, and needs. For some, testing provides clarity, peace of mind, or direction for the future. For others, it introduces uncertainty, emotional strain, or difficult questions they're not ready—or don't need—to answer. Wherever you land, know this: choosing whether or not to pursue genetic testing is a deeply personal decision, and you are not alone in making it.

You are your child's most powerful advocate. With access to trusted information, supportive professionals, and the lived wisdom of other families and Deaf/Hard of Hearing individuals, you can make decisions with confidence, compassion, and clarity. Whether you decide to test now, later, or not at all, your love, your questions, and your thoughtful consideration are what matter most. Let this guide be a reminder that you have a voice, a community, and the right to make informed choices that reflect what is best for your family.



Where to Learn More about Genetic Testing

- See the Hands & Voices Genetic Education webpage for more. https://handsandvoices.org/resources/genetic-testing
- A Parent's Guide to Genetics and Hearing Loss: https://www.cdc.gov/hearing-loss-children-guide/parents-guide-genetics/index.html
- GenomeConnect Webinar - Genetics 101 and How to Read Your Lab Report: https://www.youtube.com/watch?v=EL1N0x4tqMY and https://www.youtube.com/watch?v=01sfQLcFVuc
- What is Genetic and Genome Testing: https://www.ndcs.org.uk/
- Why are genetics important for children with hearing loss?
  Video (3 min) from Michigan Medicine, captioned with transcript and auto translated into many languages, included. https://www.youtube.com/watch?v=N2HAnxOCun8
- H&V infographic on Family and Child Genetic Testing Considerations
  https://handsandvoices.org/resources/genetic-testing/infographic-3-18.pdf
- Katharine Press Callahan, Rebecca Mueller, Karen Crew, Kyle Brothers, David Munson, Steven Joffe, Chris Feudtner, Frequency and reasons that parents decline genetic testing for critically ill neonates, Genetics in Medicine Open, Vol 2, 2024, 2024, 101896, ISSN 2949-7744, https://doi.org/10.1016/j.gimo.2024.101896