Will Eiserman:

Welcome to Earworm, dialogues on hearing health you can't stop thinking about. Earworm is brought to you by the National Center for Hearing Assessment and Management at Utah State University, known as NCHAM. I'm Will Eiserman. I'm the associate director of NCHAM, and I'm your host today. One of the significant public health advancements of the last several decades is reflected in our ability to screen infants as young as a few hours old for permanent hearing loss. In fact, most children in the United States are being screened for hearing loss within the first days of life, usually before even leaving the hospital. This is leading to approximately three children in a thousand who are being identified with permanent hearing loss shortly after being born. Sometimes, children are identified with hearing loss a little later as a result of hearing screenings that are increasingly occurring in programs like Head Start or other early care and education settings.

Because of these early identification efforts, the families of those who identified as deaf or hard of hearing have the benefit of accessing information and support necessary for making a range of decisions about how they can ensure their child will have access to language one way or the other and thrive in all areas of development. Those decisions are informed, in large part, by what the experts can tell them about the nature and severity of their child's hearing loss. So increased availability of quality early hearing screening and diagnostic services over the last several decades has been revolutionary, for sure.

Now, taking it a step further, current technology is making it increasingly easier to also potentially diagnose the cause of a hearing loss. When the cause of a hearing loss is able to be determined, it can sometimes help predict the stability of the currently-identified hearing loss, whether changes in hearing might occur in the future, as well as whether to anticipate possible impacts on other areas of the child's health or development. Our guest today is Dr. Eliot Shearer, who is an assistant professor of otolaryngology-head and neck surgery at Harvard Medical School.

Dr. Shearer is a pediatric otolaryngologist at Boston Children's Hospital, where he caress for children with a wide range of ear, nose, and throat disorders. He has a special interest in the surgical management of pediatric ear disorders, including hearing loss. Dr. Shearer is internationally recognized for his work in developing a new genetic testing platform for the diagnosis of hearing loss and has written many research articles and several book chapters on the subject. Dr. Shearer also studies ways to improve newborn hearing screening tests using technologies and ways to improve outcomes for children with cochlear implants. So welcome, Dr. Shearer.

Eliot Shearer:

Thanks for having me. Happy to be here.

Will Eiserman:

Now that newborn hearing screening has become a matter of standard practice across the United States, and many children have the potential of being diagnosed with permanent hearing loss within the first several months of life, what is on the horizon for what it can mean to have an even clearer diagnosis of hearing loss?

Eliot Shearer:

Sure. The newborn hearing screening protocol that we have in the United States has really been remarkable in its ability to identify children that have hearing loss. It's been remarkably successful. It's got a huge overall uptake by newborns and providers. More than 98% of kids in the United States undergo newborn hearing screening, and so it's been hugely successful. So the physiologic newborn screening has been very successful in that you can train someone to do it relatively easily. It's relatively

inexpensive. So the current screening protocols generally identify hearing loss in about three in a thousand newborns using the current physiologic methodology.

Will Eiserman:

Typically, when we hear the term identify or diagnosis, people think diagnosis simply means the presence of, in this case, hearing loss or not.

Eliot Shearer:

I think that when we look back at this time in the future, we're going to realize that the past decade or so has been a huge paradigm shift in how we think about hearing loss. Now, through new technologies and diagnosis, so improved genetic testing, CMV testing, new imaging techniques, in the majority of newborns who have hearing loss, we're able to determine what the cause of their hearing loss is, because the hearing loss is really a symptom of an underlying difference in their auditory system. This is a huge shift from even 20 years ago, when my more senior partners, when I talked to them, the vast majority of kids, they had no idea why they had hearing loss. Now, when I meet a family that has a newborn that has hearing loss, I tell them in most cases, we're able to give you a diagnosis, ultimately.

Will Eiserman:

By diagnosis, you mean a diagnosis of the cause?

Eliot Shearer:

That's exactly right, and this is hugely important for the families. Because there's many different treatment options now that we did not have 15, 20 years ago, and there's many decisions that I have to help the family come up with what's best for their child. There are many different options. So what you have is you have a conversion of new diagnostic tools and a conversion, simultaneously, with new treatment options For individuals that have hearing loss.

I need to be really clear that I think that how a individual, and their family, and the child approach hearing loss, hearing impairment is completely up to them, and I would never force a decision on a family. I come up with what's best for the family after I meet them and discuss with them all the different options. I think it's important for everyone to be informed about the different options, but I would never impose a treatment that a family did not want. Then, also, I should say, at the same time, that I do use the term hearing loss, which is a scientific term, but I'm not implying that someone who is deaf or hard of hearing has a loss of anything. I'm referring to a decreased hearing sensitivity on audiogram, and we don't have a better way to describe it than saying hearing loss.

Will Eiserman:

So of the children who are identified as a result of physiologic screening and follow-up diagnostic assessment, what percentage of them are we able to also diagnose a cause of the hearing loss?

Eliot Shearer:

Yeah. So if I have a newborn that comes in with a severe to profound hearing loss that's bilateral, I tell them we have at least a 60% chance of figuring out what the cause of their hearing loss is, what their actual diagnosis is, and that's very different than even a decade ago. That's through a combination of CMV testing, high-resolution MRI and CT scan, and then also genetic testing.

Will Eiserman:

Why is understanding the cause important?

Eliot Shearer:

It's hugely important to come up with a diagnosis for hearing loss for the family to know why their child actually has hearing loss, because first of all, it provides the family with prognostic information. So information is whether the hearing's going to stay the same over time or progressively get worse over time. It provides recurrence risk information to the family, so if the family's going to have another child, what the chances are that they would have hearing loss or not. Then, really importantly, a diagnosis helps to identify syndromic causes of hearing loss, so this is hearing loss that's associated with other clinical features.

Will Eiserman:

So what percentage of the children with congenital permanent hearing loss are you finding to have a genetic hearing loss?

Eliot Shearer:

If I see a child that has congenital bilateral severe to profound hearing loss, probably about 40% of the time, it's going to be a genetic. About 20% of the time, it's going to be a cochlear or vestibular abnormality that we see on an MRI, and CMV is, by far, the most common environmental cause of hearing loss. That makes up a proportion, too. That's more common for asymmetric or unilateral hearing loss or hearing loss that's more mild. So when you look at the whole average, we come up with a diagnosis in about 60%, and genetic causes are the most common diagnosis that we provide for children that have hearing loss.

Will Eiserman:

Clarify this for me, Dr. Shearer. One of the things we often hear is that the majority of children with permanent hearing loss, like over 90%, are born to hearing parents in otherwise hearing families. Sometimes, I think that could be interpreted as meaning that there isn't, in fact, a genetic component. Can you reconcile that?

Eliot Shearer:

Yeah. Sure. So every human, every one of us is born with hundreds of differences between us and our parents, and so that's what makes us all unique and not clones of our parents. The reason that we think that this happens is because there are more than 120 different hearing loss genes that have been identified to date, and because there's so many different genes, it will randomly occur that you may have a variant, which is a genetic change that's different from your parents, in one of those hearing loss genes. Then, it's just a combination of your two parents that make you unique, and that's what makes all of us unique. Some of the children will have two changes occurring in the same hearing loss gene, and then that can cause hearing loss.

Will Eiserman:

Give us an example, then, of a child who would benefit, once they've been identified as having a permanent hearing loss, from knowing the genetic cause of it.

Eliot Shearer:

Sure. So I can come up with a ton of examples. Yeah. So coming up with a genetic diagnosis for children that have non-syndromic hearing loss or just hearing loss alone is really important for those that have more mild to moderate hearing loss, actually, because once we have a diagnosis, we're able to provide that family with prognosis for the hearing loss. So there's some genetic forms of hearing loss that start out as mild to moderate, and they stay like that for the rest of the kid's life. There's others that are very rapidly progressive.

And so it changes the conversation that I have with the families every time I see them, essentially, and it changes how many times I'll see the patient or the family and the individual back in a given year. So I have some families, and their child has a more stable form of genetic hearing loss. I may see them back just once a year, and there's others that may have a more rapidly progressive hearing loss. I may see them back every three to four months.

Will Eiserman:

So it changes your follow-up protocol?

Eliot Shearer:

Yeah. Definitely. It changes the follow-up protocol, and then it changes how we think about hearing aids. Also, if I see a child that has hearing loss due to stereocilin, STRC, which is the most common form of mild-to-moderate genetic hearing loss, I'm never going to have to have a conversation about cochlear implants with them, essentially, once we give that diagnosis. I may mention it as a treatment option if the hearing loss gets worse over time, but it's not in the picture, really, for them. Whereas if we have a rapidly progressive form of hearing loss, I'm probably just going to bring it up a little earlier as a different treatment option so the family has more time to think about it, so it's not a sudden all of a sudden we're talking about cochlear implants.

One of the most important things that we are able to do is evaluate for syndromic causes of hearing loss when we perform genetic evaluation and comprehensive evaluation for hearing loss. Some of the most difficult days that I have in clinic is talking to parents and giving them the result that their child has Usher syndrome. So this is hearing loss plus progressive vision loss due to retinitis pigmentosa and also balance issues. Most of the time, these parents do not have hearing loss. They're not deaf and hard of hearing, and their child is identified to have hearing loss on the newborn hearing screen. We have to give them the result that their child will have vision loss, as well, and if it's Usher syndrome type I, that vision loss starts before the child is 10 years old most of the time. So we have to provide that information, which can be very devastating to some families.

I'll tell you, it's also really important to these families, too, because there are things that you can do to maybe delay the onset of the vision loss. So you can take vitamin A and wear sunglasses, for instance. Then, many of these families that I've talked to, although it's very hard news to take, they're so grateful to have that information, because it changes their approach to the hearing loss. These are the families that, if they were maybe on the fence about getting a cochlear implant, for instance, once they get the news about Usher syndrome, they want to fast track towards cochlear implants.

So it changes how we think about cochlear implants, because if you have a loss of your vision, as well, it can make communication using sign language very difficult. They also will start teaching their child braille before they lose vision. So 20, 30 years ago, we used to identify these children after the vision loss had already begun, and then we were trying to play catch up. Now, we're identifying these children sometimes before they're the age of one, and it's really important to these families.

Will Eiserman:

So the discovery of the hearing loss and then the genetic cause of the hearing loss can lead you to discover other developmental or health concerns that can be very important for the family to know and to factor into their decision-making.

Eliot Shearer:

Exactly. Yep.

Will Eiserman:

What does genetic testing involve? What does that look like?

Eliot Shearer:

Yeah. The genetic testing for hearing loss has changed a lot in the past decade. So when we first started doing this back in 2010, we had to have a blood sample. It would take six months or so. It was essentially never covered by insurance. We worked hard to push the insurance companies to realize the importance of genetic testing, and now, actually, at least where I am in Massachusetts, the majority of private insurers pay for comprehensive genetic testing for hearing loss in children.

Will Eiserman:

Does that mean that any child with a diagnosed permanent hearing loss is eligible to have genetic testing?

Eliot Shearer:

Yeah. When we look at our data for private insurers, any child that has a permanent detectable hearing loss, about 80% of the time, private insurers cover for that. There's some states like California where they have a blanket coverage statement that all children with any permanent hearing loss can have genetic testing available, and instead of having to draw blood now, we typically do a cheek swab or a buccal swab. A lot of these companies, they just do it at home, and you send the kit back in. You don't even have to go into the lab, and we typically get the result somewhere between one and three months after we get the swab back in. So the timeframe has decreased. The costs have decreased, and it's easier than getting a blood draw.

Will Eiserman:

When you're talking about genetic testing, are you talking about testing just for the most common genetic forms of hearing loss or something more comprehensive?

Eliot Shearer:

So there's 124 different non-syndromic hearing loss genes, so just hearing loss alone, and then there's several hundred syndromic forms of hearing loss. The big shift that happened is that because there's so many different genes, it's really hard to test for these genes. And in the early 2000, we just didn't have the technology. It wasn't until 2008, 2009 that we started having the technology where we could sequence all these genes at the same time, and what we found is it's really important to sequence them all. You can't just pick one or two and sequence it.

There's many different commercial platforms that are available now that perform what's called comprehensive genetic testing for hearing loss, and what the comprehensive part means is that they

screen all known hearing loss genes. It usually ranges between 100 to 200 different genes that are associated with hearing loss, and they actually perform sequencing of all those genes and look for any different variants that are in them. It's really important that you do comprehensive testing and not just a single gene testing, because it's not nearly as effective.

Will Eiserman:

What does a healthcare provider who has a patient with a diagnosis of permanent hearing loss need to know about ordering genetic testing?

Eliot Shearer:

Ordering genetic testing for hearing loss is not for the faint of heart, I would say. So I think you need to be equipped to give the result back, to provide the result. So things have gotten a lot better in the past few years, where several of these genetic testing companies will actually provide genetic counseling for families when the results are back. I think any individual who orders this testing should make sure they have the resources available when they get the results.

The reason that's important is sometimes we provide some news that can be very impactful and sometimes unexpected to families. So an example for that is something called Jervell and Lange-Nielsen syndrome, which is hearing loss, but also long QT syndrome, which is a cardiac abnormality, conduction abnormality that can cause sudden death in children and adults that have it. And so sometimes, you can find things that maybe you weren't expecting, and that's the goal of genetic testing. But you just need to make sure that you have the counseling available.

Will Eiserman:

It seems really important to have a designated person who is trained to address the results of genetic testing with families.

Eliot Shearer:

So a genetic counselor is really important. So it's a trained professional who's licensed or certified, depending on the state you live in, and they will meet with the family after they want to proceed with genetic testing. So again, not every family wants to proceed with genetic testing, but if they do, they'll meet with the genetic counselor. They get what's called pre-test counseling. So the genetic counselor will explain the test, all the genes that are tested for, and then they'll talk about some of the risks of the testing.

The primary risks of the genetic testing are identifying things that maybe you weren't expecting to find. So one thing that we do sometimes find with genetic testing is what's called non-paternity, where we may find that the parents are not exactly who they thought they were, and we may identify other associated conditions besides hearing loss. And so the genetic counselors, their primary role is education. So they'll talk about what the test is, what the test does, and get the family ready so that when the results come back after a month, a couple of months, they'll be able to provide those results back and educate exactly what the results mean. So really, their primary role is interpretation and education for the family, and it's very helpful through the process.

I would say that if a child has permanent hearing loss, the first test that you should think about would be genetic testing. Genetic testing provides you the greatest chance of uncovering the actual diagnosis or reason why there is hearing loss. It's much more effective overall than things like imaging or any other test that we can order.

Will Eiserman:

That was Dr. Eliot Shearer. I'm Will Eiserman from the National Center for Hearing Assessment and Management at Utah State University. Check out earwormpodcast.org for other episodes of Earworm, dialogues on hearing health you can't stop thinking about. (singing)

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