Grand Rounds: Pediatric Audiology - Exploring the Uniqueness of Every Child to Achieve Best Outcomes, in partnership with Phoenix Children's Hospital

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- [Christy] It's my pleasure to introduce a wonderful group of clinicians from Phoenix Children's Hospital. And we'd like to welcome Dr. Wendy Steuerwald. She is the coordinator for this grand rounds, and she's going to introduce each one of our wonderful speakers today. If you haven't caught our grand rounds before, they're available to you on the on demand on our audiology online course library. But at this time, I'd like to welcome Dr. Steuerwald and hand the mic over to you.

- Christy, thank you so much for that warm introduction. And everyone who has signed on to hear our presentation, thank you so much. We're going to be talking today about pediatric audiology. We are exploring the uniqueness of every child to achieve the best outcomes. And the first thing I'd like to do is just to introduce our presenters today. The first presenter will be Dr. Lynn Eyde, and Lynn is the audiology manager. She's also the newborn hearing screening coordinator and her area of specialty is diagnostics. The next presenter's going to be Dr. Maddie McNamee. And Maddie is an audiologist I, and her areas of specialty include diagnostics in patients and electrophysiology. She really enjoys ABRs in the OR under anesthesia and also natural sleep ABRs.

The next presenter is going to be Dr. Allie Sayer. Allie is an audiologist I and her areas of specialty include cochlear implant, diagnostics, ABR and natural sleep. The next presenter is Dr. Alissa Nickerson. Alissa is an A2 audiologist with a focus on hearing aids. She also specializes in diagnostics, ABRs in natural sleep and under anesthesia in the OR. Next presenter is Dr. Ashley Geske and Ashley is an A1 audiologist. Her areas of specialty include diagnostics, cochlear implants, hearing aid and ABR in natural sleep and under anesthesia. Our final presenter is Dr. Deborah Flynn and Deb is our lead audiologist with the focus on cochlear implant. She also specializes in diagnostics, hearing aids, telehealth services and ABRs in the OR.

So we are very excited to be talking with you today. Here are all of our disclosures. Everyone is an employee of Phoenix Children's Hospital. Please look at it closer if you

wanna learn more. Our learning outcomes are, after this course, participants will be able to describe how a multidisciplinary approach can help facilitate best patient care, discuss and analyze several complex audiology cases. Describe how complicating factors can come together to support the patient experience. So before we started into the case studies, I wanted to give you just a little bit of information about Arizona, because Arizona is a unique state. As you've probably heard, Arizona is the Grand Canyon State and you can see up at the top of the map is the Grand Canyon.

But did you know that there's also numerous mountains throughout the state of Arizona? The temperature is diverse. There's often snow in the top part of the state while it's very hot at the bottom part of the state. Arizona is also a new state. It wasn't even a state until 1921, and it's the 48th state to join the United States of America. As you can see from the map, Arizona is a border state with Mexico right beneath us and, you know, other states around the sides. In the middle of the state you see a yellow blob, which is Phoenix. Phoenix Children's Hospital is located in Phoenix, and we are the only freestanding hospital in the state of Arizona, and we just had our 40th birthday.

So we are a brand new children's hospital, relatively speaking. So what makes Phoenix Children's unique? Phoenix Children's, also known as PCH, is the sixth largest children's hospital in the fifth largest city in the country with the fourth largest pediatric population. It's like, wow, we have a huge number of kids. It's part of a statewide pediatric healthcare system. We also have two additional hospitals under construction. We have a wonderfully collaborative care team. One of our otolaryngologist is the EHDI Chapter champion. We, Phoenix Children's audiology, are the state of Arizona newborn hearing screening contract. PCH also provides newborn hearing screening services at four local hospitals in addition to our audiology services. We call our ABRs BAERs and that's an acronym for brainstem auditory evoked response.

We have generous, clinically inquisitive audiologists, generous with their time, with their expertise, and do whatever it takes to get the patients what they need. And I forgot to add, Phoenix Children's is the only free-standing children's hospital in the state of Arizona. So as I think about why do we have all these unique patients, why are people coming to us? And I think it's a combination of things. I think it has to do with where the state of Arizona is within the United States. You know, as I mentioned before, we're a border state, so we have a lot of people coming up to the north from Mexico. We also have people coming from surrounding states. As I just said, we are the only freestanding pediatric hospital.

So kids come to the hospital for all kinds of things. And also, I think it has so much to do with the audiology team. These audiologists are fabulous. They are so inquisitive, wanting to do whatever they need to do to get the best outcomes for their patients. And I think the word spreads and people come to see us for all of those reasons. So we have six unique case studies that we want to share today. And I just wanna tell you guys, you are in for a treat. It's gonna be a wonderful presentation. So it's my pleasure to introduce Dr. Lynn Eyde who is going to be doing our first presentation of the day.

- All right, thank you so much, Wendy. So I am gonna be talking today about Sophia. This is a kind of run-of-the-mill, failed newborn hearing screening and I'm gonna walk you through her case. So before we get started with her, I want to just give a little bit of a newborn hearing screening review just so that we are on the same page once I get started. So newborn hearing screening, as we know, is not a test of normal hearing sensitivity. The test really doesn't capture the entire frequency range and we know that it can't really rule out a slight or mild and even sometimes moderate hearing losses sneak by. And again, so a pass does not indicate normal hearing.

Also, we consider a pass to be when both ears have passed during the same testing session. So a fail would, in the same sort of regard, we would want that to be the same

session. So a pass is in the same session as well as a fail, would be one ear or both ears did not pass. A stop and start is when a test is started and then they don't let the test complete. So they've stopped it for some reason. We consider that to be an incomplete test. This will make better sense why I'm highlighting this in a little bit when we review her results. So Sophia came to us here at Phoenix Children's due to a failed inpatient newborn hearing screening at an outside facility.

She was seen initially with us for a failed outpatient hearing screening within our audiology department. And then when she did not pass that, she was sent on for a diagnostic test, which is at the point where I was meeting her for the first time. The pregnancy history is fairly uncomplicated, you know, in conversation the mom did report that she was diagnosed with COVID around the second trimester of pregnancy. But other than that, nothing was really complicated. There's no family history of hearing loss in childhood. The father is in Mexico and he has not been able to obtain immigration status. Also, the home language is Spanish. The mom does speak English fluently as well as Spanish. So our appointments were conducted in English with the mom.

All right, so Sophia's journey, this might be the same for a lot of other states as well, but a nice thing that we have the ability to do at Phoenix Children's is we are able to review all the records for our incoming patients that have been seen through the newborn hearing screening. So we can see their inpatient results, their outpatient, as well as if they've had another diagnostic testing that's been reported through our state's reporting system, which is high track, which some people might be familiar with. So before we see kids here for BAERs, we do like to review these records just to give us an idea of what we're kind of in store for. So when I pulled up her record, I saw that when she was an inpatient, on the first day they attempted it, she had 11 incomplete tests in one year and they didn't complete the right at all.

The next day they returned and there were six incomplete tests before then receiving a passing result. And this a similar story on the right, she had four incomplete tests and then a failed result. So at that point the PCP had referred to Phoenix Children's for an outpatient newborn hearing screening. And when they came to us on the 17th of that same month, she failed in both ears. So at that point she was scheduled to return to see us for that diagnostic, as Wendy had said, a brainstorm auditory evoked response study. And so that's when I was meeting her. So this is her first BAER study. The tympanogram was peaked in both ears when we used the 1000 hertz probe tone, distortion, you know, product otoacoustic emissions were absent in both ears.

And the results here show us a moderate sensorineural hearing loss. And this column here just captures our click that we were using for this patient. So counseling. So with this, I spent probably more time counseling this mom on the newborn hearing screening than I normally do just because I know in a case like this where the kid has passed, it can sometimes be a question of like, well, why did they pass? I don't understand, because a lot of times I think families do wanna grasp onto that, that hope that she did pass. So, like, why is your result showing something abnormal? So that past did confuse the mom as why now I'm telling her she has hearing loss in both ears.

So I did spend time explaining to the mom that, you know, the newborn hearing screening does not capture normal hearing or does not indicate normal hearing sensitivity. And I didn't necessarily tell her this next piece, but in this case, I think that passing result in the left ear was really due to just over-screening of the patient. I think if you stop and start enough times, you really can get somebody to pass, and we've seen that. So I think that this is just a case, like an education case as well for the screeners to show, like, babies shouldn't pass sometimes. And so because they do have hearing loss. The mom did seem to kind of acknowledge that information and really started to ask about the next steps.

And I remember kind of taking aback, the mom really didn't skip much of a beat. She just said like, "Okay, well what do we do next?" This is where at the point I was learning about the father living in Mexico, and you know, the mom really was now, like, how am I gonna do this kind of on my own without him being able to come here? I also did at that point offer to include the father in the discussion of the results and to use, you know, a Phoenix Children's interpreter, Spanish interpreter. But she said, you know, at that point, she said she'd relay the information and pass on to us any sort of information or pass on, excuse me, to him any information.

So I saw her back for a repeat BAER really just to kind of fill in the gaps. So I did repeat some of the same frequencies, but I wanted just to make sure that I could fill in the gaps and make sure we weren't, you know, missing something. So again, it confirms that moderate sensorineural hearing loss in both ears. At this point I really was, like, heavily doing diagnostic testing and not really taking on new hearing aid patients. So I did give the mom the option to do the consult at this appointment or to come back, like, the next week to meet with the managing audiologist who would be taking on the hearing aid and fitting and all of that.

So the mom elected to come back and I think it also worked out timetable-wise for her to do that. So that's not always how we do that, but when they saw me, that's how it worked out. Also this child got in to see the otolaryngology department at Phoenix Children's quite quickly. So even before my second BAER with this patient, she had established care with otolaryngology, where they ordered an MRI, blood work, genetics consult, and ophthalmology consult. The patient was then seen for a hearing aid consult, and then the MRI was completed just a few short weeks later where the results were normal. And then again, about a week later, the patient was fit with binaural hearing aids and custom earmolds.

And also the ophthalmology consult and genetics were happening. So ophthalmology did see them in March of that year with no, you know, complications, and they recommended a follow-up in one year. Genetics consulted with them in July and they did run a hearing loss gene panel, and when they reviewed those results in October with the family, it did show that Sophia had Usher syndrome type 2A, which we know is associated with moderate to severe congenital hearing loss and progressive retinal degeneration. So after that, that follow-up with ophthalmology kind of changed. They actually sent her out for a retinal specialist appointment sooner than that one year mark. And everything up to this point from that specialist has been showing normal.

Sophia's audiology progress is not as ideal as we would like to see. Just she has a pretty significant hearing loss and she's really not wearing her hearing aids long enough or consistent enough. She's wearing 'em about two to three hours a day. The patient is watched by her grandma while the mother is at work. And so the grandma really doesn't feel comfortable putting them in the ears. Now I don't know if that's, like she's not able to actually get them in the patient's ear or if she doesn't want the patient to wear them. I'm not exactly sure. A positive is that Sophia is enrolled in early intervention and also does have an hearing, like, specialist teacher of the deaf who comes to her home from the Arizona School for the Deaf and Blind.

So there are people, you know, visiting and making sure that she is meeting those milestones, but that data logging is poor. We'd really like to see that a lot higher. Also through this, the mom has requested, you know, a letter to be written to immigration to try to get the father here from Mexico. I know a number of providers, audiology and ENT as well as, I think, genetics wrote a letter. He still is in Mexico and hasn't been granted this, which I do think does create some difficulty for Sophia and her mom. All right, so some unique things about Sophia is, again, this an example of newborn hearing screening, over-screening. We don't always see that, but I'm glad that she didn't pass that final time on the right ear or else this would be a different story.

You know, immigration, this is a factor for her and her family. The mom is, you know, supporting multiple siblings on her own with the grandma's help, I know, but that can lead to, I think, some of this poor data logging. And we don't always get to see a genetic hearing loss. You know, we refer all our patients with hearing loss for genetics and they don't always come back. So that's unique that she did have a genetic result. And visual impairment, again, we see those ophthalmology consults happening and how many times are they normal when we don't always get to see that these kids with visual impairment. And that should play a factor and also the importance of how, or I should say, how important it is for her to be wearing her hearing aids consistently.

Also, her family, she does have family here, which is nice that she has that grandmother's support, but it's also holding her back a little bit. If we could get someone to keep the hearing aids, you know, on her and in her ears consistently, I think her outcomes would be better. And going back to the newborn over-screening and that passing result, it does cause some confusion and we have to do some unpacking and unwinding of that confusion, which can take away from some of the other counseling that we would like to be doing. So thank you for letting me share about Sophia. I am gonna hand it over to Dr. Maddie McNamee, who also has a very interesting case.

- Thanks, Lynn, for the introduction. Everyone, I am going to be introducing you to Walter. He is a case of unexpected or untraditional test results. Go. And one second, I'm just trying to get to the next slide. There we go, okay. Walter was first seen by our department at 12 years of age. As Lynn and Wendy mentioned, our hospital also has a newborn hearing screening program, which services some of the birth hospitals in the Valley. Walter was born at a PCH newborn hearing screening hospital where he

received two inpatient screenings for which he failed bilaterally and then was referred for an outpatient rescreen, which is what we saw him for at that 12 days. Once again, he referred his A-ABR bilaterally and then was referred for a diagnostic BAER study.

This is when I officially met Walter. At the time, all of his reported birth and family history were unremarkable for hearing loss. Tympanometry in 1000 hertz probe tone revealed flat tympanograms, a high level click was used to rule out auditory neuropathy spectrum disorder, and his distortion product otoacoustic emissions or DPOAEs were reduced or absent bilaterally. Frequency specific air and bone conduction thresholds using tone bursts were consistent with a moderate to moderately severe, predominantly, sensory neural hearing loss bilaterally. At that time, given the sensory neural hearing loss, I recommended otologic and genetic consultation. Additionally, I referred him to our early intervention program, which we call AzEIP, to initiate home-based services for children with hearing loss. And then I recommended that Walter returned for a repeat BAER study.

I asked that he come back in four to six weeks to give us a chance to have that middle ear dysfunction resolve, hopefully prior to our next test session. He returned for his repeat BAER in April of that year. Once again using in 1000 hertz probe tone, we had a peak tympanogram for the right ear, but still a flat tympanogram for the left ear. His otoacoustic emissions were largely present for the right ear and then partially present for the left. And then a BAER study again using tone bursts indicated a moderately severe to moderate sensorineural hearing loss bilaterally. There was an improvement noted in that threshold at 2000 hertz for the right year, but otherwise it was largely stable compared to his results from the previous month.

A hearing aid consultation was completed at that time. I discussed with his parent, with Walter's parents, that while his test results weren't exactly what we would expect to see given his severity of hearing loss in those present OAEs, I would recommend fitting

Walter with binaural hearing aids. He returned in May of that year, was fit with binaural hearing aids after receiving medical clearance from our otolaryngology team. And he tolerated the devices very well in the office from the get go at his fitting. His family reported that they had begun to see a hearing specialist through AzIEP and they'd also established a relationship with a mentor through our Hands and Voices program here in Arizona. He went on to become a very consistent hearing aid user and from his first hearing aid check, he was routinely using his hearing aids at greater than or equal to eight hours a day.

Prior to Walter's hearing aid fitting, he established care with PCH ENT. At that time, it was reported that mom had tested positive for COVID while she was seven weeks pregnant, similar to Lynn noting that we don't exactly know what that means right now, but it was an important case history piece that I was unaware of when I first met the family. The family was also able to obtain Walter's CMV testing from his blood at birth and found that it was negative. ENT recommended that Walter be seen by genetics and then return to audiology for a follow up. He saw ophthalmology that same month, and other than hypermetropia, things were unremarkable and it was recommended he follow up at a year.

And then in June of that year, he established care with genetics and a hearing loss panel was ordered. His genetic results were reviewed with the family in September of 2021. And the geneticist noted that there were two variants of unknown significance that came back on Walter's hearing loss panel, GIPC3 and OTOGL. Each of these genes is associated with autosomal recessive or non-syndromic hearing loss. And then they also recommended that Walter undergo a renal ultrasound, which was completed in October of 2021 and indicated normal kidney structures. While I'm familiar with some of our genetic markers for hearing loss, specifically ushers or connexin, those are ones that we as audiologists see a lot, I had not previously heard of the GIPC3 mutation or the OTOGL mutation.

So I wanted to do a little bit of research. A literature review indicated that GIPC3 is a mechanical conduction disorder of the inner ear. This is associated with sensory neural hearing loss and acoustic seizures specifically. And then one article in particular noted that there were stereocilia bundle deficits for both the outer and inner hair cells. More specifically, they found that the inner hair cells were especially impacted and were less rigid and more sparse than their outer hair cell counterparts. I found this to be particularly interesting as it could potentially explain Walter's presentation of present OAEs, but a moderate to moderately severe sensorineural hearing loss. And then looking at OTOGL, a literature review indicated this is often associated with a mild to moderate sensorineural hearing loss, again consistent with Walter's presentation where he's in the moderate to moderately severe range.

And then back to ENT. Throughout this time, Walter continued to have middle ear dysfunction. He ended up going under PE tube placement in February of 2022. A repeat BAER study was completed following this PE tube placement and results indicated a severe rising to moderately severe sensorineural hearing loss. This was a significant decrease from Walter's previous BAER studies that had been completed natural sleep in the office. It was noted that this testing did take place following PE tube placement which could have impacted the results. And I discussed this with Walter's parents and noted I wanted to monitor him closely and attempt behavioral testing at his next hearing aid follow up. Walter returned to our office in March of 2022 at a year old for his first behavioral evaluation.

At that time, his PE tubes were in place in patent. His OAEs were partially present bilaterally. And then he was able to condition easily to a VRA test paradigm, and the results were consistent with a moderate hearing loss in at least the better ear. This showed an improvement compared to Walter's post PE tube BAER, but was consistent with his BAER studies from infancy. Over the course of the next year, Walter continued

to be an excellent hearing aid user. He made great progress in his development. He continues to meet with AzIEP and Hands in Voices. And then in March of 2023 or this past March, he returned at two years old for follow-up behavioral testing. This was our first attempt at CPA, although his mother reported that they'd been practicing this test paradigm at home with their AzEIP provider, their hearing mentor.

Walter's PE tubes were in, again, in place in patent and this time both transient evoked and distortion product otoacoustic emissions were completed. DPOAEs were present and reduced but notably absent in the low frequencies and TEAOEs were largely present bilaterally. Behavioral testing was completed using a single tester. It was just me and we were trying out CPA for the first time, which indicated a mild hearing loss for the left ear and a moderate hearing loss for the right ear. However, the results were obviously a little limited. Walter's SRTs were completed using monitored live voice and body part identification as he was unable to condition respond the words at this time, but they were in good agreement with the limited frequency specific information.

I recommended that Walter return in two to three months to expand these results with the hopes of having a two-tester paradigm available for repeat testing. And then I also encouraged his mother to work on picture pointing spondees at home to help familiarize Walter. He returned in June of this year, and at that time his mother reported he'd recently started to reject his hearing aids and was stating that he doesn't want them. Historically, Walter had been a very consistent hearing aid user and his mother was curious if this was due to a change in hearing as to why he was maybe starting to reject these hearing aids. CPA was reattempted this time with a two-tester paradigm.

Walter again participated with great reliability. His PE tubes again were in place in patent. We used supra-aural headphones and we found that his test results were consistent with a moderate to mild hearing loss, rising to normal hearing sensitivity for the right ear and a slight hearing loss for the left ear. Once again, we attempted SRTs

using picture-pointing spondees, but Walter was still unable to familiarize. But his body part SRTs were in good agreement with these behavioral results. It was noted that Walter's thresholds had significantly improved from 2000 to 4,000 compared to his BAER study, however, results from 500 to 1000 hertz were largely consistent with those BAER studies. So we've had a big change in hearing. What did we do?

I reviewed these results with Walter's parents. I noted that we did have that significant improvement in thresholds compared to Walter's prior behavioral testing via VRA and his BAER studies. We agreed that we would reprogram his hearing aids based on this testing given his good reliability and then also the parental report that he'd begun to reject those hearing aids. But I encouraged Walter's parents to monitor his progress closely, specifically his speech and language development, which up to this point had been on track or was advanced. We agreed we would retest Walter in three months to expand and confirm these test results. I saw Walter just last month in October of 2023. His parents reported he'd been doing very well since his hearing aid reprogramming.

He was no longer rejecting his hearing aids and was wearing them consistently. We repeated behavioral testing with the addition of bone conduction and results were consistent with the results from June. We were able to complete SRTs using picture pointing spondees and recorded speech stimuli at this time, and they were in good agreement with these results. And then Walter's hearing aid programming was not adjusted at this time. He'd been doing well, results were consistent, so we left it as is. However, we agreed we would continue to monitor Walter closely, and again, I recommended a three-month follow up. Well, Walter's audiometric results are not what we typically expect to see. I think this emphasizes one of the major takeaways of pediatric audiology and pediatric testing.

Every child in case is going to be different and we need to adjust our care plan to meet the needs of them and their families and to support their growth and development. So

now looking at Walter, what exactly makes him unique? We know his mom tested positive for COVID during pregnancy, and while we don't exactly know what that means yet, that's an interesting contribution to his development. As Lynn noted in her presentation, we don't always get positive genetic results back, but in Walter's case, we have two markers we can look at in terms of genetic hearing loss. He has those present OAEs. We know he's CMV negative. He's a very successful hearing aid user with that sensorineural hearing loss.

Well, thank you all for listening to my presentation about Walter. And in that case, I'm gonna hand the reins over to Dr. Allie Sayer to introduce us to Ms. Sophia. Oh, sorry, Maria.

- Thank you, Maddie. So Maria is a now 15-year-old female who first came to us when she was 11 years old in November, 2019. Her and her mom had just relocated to the United States from Venezuela. I mean, she received care in Florida for a short time before they moved to Arizona. Both Maria and her mom are Spanish speaking as their primary language. Maria was seen at this time using funding from a local organization for children with hearing loss because they did not qualify for Medicaid with their immigration status. At that time, mom reported a history of severe to profound progressive hearing loss. First diagnosed in Venezuela around two years of age when she still wasn't speaking, but her mom thinks that it was present from birth.

She did not have a newborn hearing screening and even now I can't locate any information about a screening program in her home country at this point. At Maria's first visit, mom provided some, although certainly not complete, records. She was reportedly fit with bilateral hearing aids after her diagnosis at two years of age and wore them consistently. But I have no information on her degree of hearing loss in the early years or what type of hearing aids she wore. These audiograms are recreated from Venezuela. A lot was unintelligible due to the use of handwriting and the language

barrier, but there were several inconsistencies. In some places word recognition scores were listed as 100%, but in other places scores as low as 16% and 28% in the right and left ears were reported.

There were unexplained air bone gaps. Hearing was sometimes worse at one evaluation than better for the next. They used sound field markers for both right and left ear results. They had mass bone conduction symbols used where there was no response in air conduction on the opposite ear, and my favorite, which was at this point in March of 2018 on the right hand side, the audiogram showing severe to profound sensorineural hearing loss from 500 hertz and above. Her word recognition score was listed as 100% in both ears, which we know is really not likely with this much hearing loss. These records were from Florida where Maria received care immediately before establishing with our office. Mom at that time was very motivated to pursue cochlear implants because Maria was struggling to hear and understand anyone except her mom, who she could communicate with fairly well in Spanish as long as she could see mom's face, and sometimes even when not, she had not worn a hearing aid in the right ear for around one year after it broke.

But she did arrive to our office wearing a very inappropriately fit slim tube hearing aid for the left ear with a dome. She was not receiving any accommodations for her hearing loss in school and was struggling trying to learn English. She was tested in our clinic with another provider at that first visit in November, 2019, and this was her audiogram. Speech testing was not completed since we didn't have Spanish materials at that point. It was recommended she be fit with new hearing aids, and we arranged for her to return in December, 2019 where I met her for the first time for a fitting of high power BTEs with custom molds. The devices were loaners from the same organization who funded her testing and fitting appointment.

She was immediately overjoyed having better access to sound, but wasn't happy with the poor sound quality at the right-hand side where she hadn't worn a hearing aid in about one year. One month later in January, 2020, she followed up for a hearing aid check where aided testing showed less than ideal audibility with the hearing aids. Worse was for the right ear than the left. Speech testing was attempted for the first time after accessing Spanish lists. However, this was complicated, because we didn't have recordings of those lists. Since I don't fluently speak Spanish and we couldn't use the interpreter since it was often by phone at that time, we had to coach her mother as best as possible and had her present the words through monitored live voice.

Her mom and I agreed to listen to the responses together and had to agree on whether or not it was correct. What I think is really important to know is that at that time she showed no ability to understand interpreters, whether it was in-person or by phone, and functionally seemed to perform much poorer than what these scores showed, especially for that left ear. Mother again expressed interest in cochlear implants, but without funding sources to cover the entire lengthy process, we couldn't proceed. I saw her back next seven months later in August, 2020 when she returned with a complaint of decreased hearing. At that time she was also attending school online due to the pandemic and was struggling with her academic performance even more than when she was in person.

Our testing did show some decreases to thresholds, more at the left ear than the right with a type A tympanogram. Her hearing aids were reprogrammed when we were able to provide more gain, but this was maxed out in several areas. Mom asked yet again about cochlear implants, but they still did not have insurance and there was nothing we could do. Mom at this point was getting so desperate for help that she considered having her friend adopt Maria, at least on paper, just to get her qualified for the insurance in the US. She returned three months later in November, 2020, now with a new complaint of tinnitus where we saw further slight decreases to her hearing

thresholds. We couldn't adjust her hearing aids at that point due to reaching the output limits, and where we could increase it in the low frequency, she reported discomfort and a feeling of vibration.

Our victory at this point though was that they had obtained private insurance coverage and we were finally able to proceed with workup for cochlear implants. Our cochlear implant team workup is a multidisciplinary approach, so she underwent an ENT consult, CT scan, psychology consult, and a speech and language evaluation. We also retested her aided speech understanding as it hadn't been done since January of 2020 with her scores listed here. Given the aided score of 64% in the left ear along with her age and unclear history, the team recommended unilateral implantation in the right ear to start. Both her mom and I disagreed with that plan. Just seeing how much she struggled to understand spoken Spanish with anyone other than mom, we knew that these scores were really not reflective of her true auditory capabilities.

Her mom was insistent on bilateral simultaneous implantation. At this age, I might typically hesitate with that, but after long talks about how difficult it could be in that activation phase, Maria wanted it as well. So I recommended that we postpone the decision so that we could reevaluate with recorded materials. I was able to use this patient case to push for recorded Spanish materials to be purchased. In addition to a conventional speech test battery, we also obtained pediatric test battery, including picture-pointing SRT and word recognition tests. The picture-pointing tests are a really nice addition to our test battery here in Arizona. Since we have such a high caseload of Spanish speaking families, many of whom exclusively speak Spanish at home.

And consequently, we see many young children through about kindergarten to even first grade age, who primarily speak Spanish until they start learning English at school. These tests can be administered and scored by audiologists who have no understanding of Spanish since the English translation is either written on the score

sheet or included on a separate auditory track to play through monitor headphones. Given our earlier concern about our ability to appropriately score a standard test battery, I considered using the picture-pointing test to eliminate the language barrier, which could influence my ultimate score for her. However, at this point, she was 12 years old and obviously had Spanish language levels well above the words in the SPPIT word recognition test.

And I was concerned that using a close set test like this would have a bigger impact. So we elected to proceed using the standard materials and mom and I scoring those results together. Very shortly after those materials were obtained, she returned for retesting of aided word recognition scores. At that same 60 dB SPL level with recorded lists, these scores of 0% for the right ear and 12% for the left ear were much more in line with our perception of her speech understanding abilities in real-life and showed such a significant decrease that it changed the team's mind. She underwent bilateral simultaneous surgery in April, 2021 and was activated two weeks later in May. At her first follow-up one week after activation, she was very frustrated.

She told me that they sounded like garbage and she didn't like me. Her mother clearly understood how difficult it would be and those expectations for the post activation phase, but it didn't seem like Maria fully grasped how difficult it would be. Regardless, she did what she was asked. She wore her processors full-time. From her first follow-up, data logging showed 11 hours or more daily use. She attended weekly speech therapy here at the hospital and was supplementing with Zoom speech therapy with a Spanish provider in Venezuela. At three weeks post activation, her residual hearing was tested. She retained a significant amount of her hearing. She was much happier and back to herself at this point, reporting that she was hearing so much better already.

Her performance in clinic absolutely supported that. We didn't yet test aided speech. It was just a little bit early for how I like to monitor my kids at this point, but she was able to communicate with a video interpreter for the first time without her mom needing to relay information. And at this point, she also convinced me to speak Spanish with her to test her ability to understand other people and could hear well enough to correct my pronunciation of a word I said just a little bit wrong. At seven weeks, she had made significant gains in her aided speech testing, now at 40% using those recorded materials. At six months after activation, she was finally confident enough to attempt testing in English.

We used PBK half list due to her English language levels. And this testing was particularly moving to mom, because one of her concerns was Maria having a hard time learning English with her hearing loss. And now she was performing relatively, equally well between English and Spanish, although there are definitely still some concerns that the English and Spanish lists aren't at the same difficulty level due to differences in the age range of the tests we use and the validation methods for the different test results. At one year post-activation testing was repeated, but at this point she requested English and did not want to do a Spanish test for me. There was no significant improvement in her scores, but her confidence had skyrocketed and she was doing so much better in school.

Now she was seen most recently just this October of now two years and four months after activation of her implants. She was very mad that I asked her to do testing in both English and Spanish and wanted to do English only. Mom also noted that she's speaking more English at home too. Maria is now a freshman in high school on an IEP and doing excellent in her second language. She didn't rely on mom or the interpreter to clarify anything I said at any point through the appointment, and at a few points when the interpreter connection was poor, she was helping interpret for her mother. Looking back at this case, there were so many things that made it unique.

You know, her hearing loss was late identified due to lack of newborn hearing screening and limited resources in her home country. Her early care was inconsistent and at times poor. When she got to the US, not only was she impacted by a language barrier with English as her second language, but she also had no insurance due to her immigration status. So we could provide her limited resources. Once we were able to, you know, further get her appropriate care after overcoming those hurdles, we still had speed bumps in the road, needing to use monitored live voice materials and then further difficulty describing, how do we score and interpret a language just in language other than English? You know, while these factors might apply to other patients, the way that they interacted to shape Maria's course and ultimate outcomes was absolutely unique.

What I really love about this case is how well it just highlights these points. You know, we know language barriers are very complicated. We're very fortunate here at the hospital that we have easy access to interpreters, but this doesn't address every issue, especially if both the parents and the patient don't speak English. You also need to consider what speech tests to administer, how to score them appropriately, and which language to test when they are bilingual. And this could change from patient to patient and based on which clinician is seeing them. We also know that using recorded materials is so important. We have all seen this research, we know that that's critical, but for Maria, this made a huge difference.

You know, if we hadn't obtained these recorded materials in Spanish, her course could have been so different getting just a unilateral implant first. It's also been really helpful in counseling families who are hesitant to proceed with cochlear implants, because of their perception that the child under each and speech well at home, which we know is very different than when we look at those scores using recorded results. I also think it's really important to consider what we define as success. If you just look at her scores,

you might not think she's doing super well, but even at a few weeks after activation, when her best score was in the 40% range, her and her family were thrilled with her performance.

She went from no ability to communicate with anyone but her mother to being able to converse with unfamiliar speakers, understand without visual cues, and is learning a second language, which is difficult to do even without hearing loss. Maria and her mom are so happy with their decision to do bilateral simultaneous implants and with her current performance, and so am I. It has made all the difference for her. These are my references. And now I will pass you along to our next presenter, Dr. Nickerson, who will introduce you to Jacob. And as a reminder, if you have any questions, go ahead and pop those in the Q&A for us and we will get to those a little later.

- Yeah, thank you so much. I am gonna be speaking today about a patient that I follow, named Jacob, who has NF2 or neurofibromatosis type 2. As many of you know, neurofibromatosis type 2 is a genetic disorder that involves changes in the NF2 gene. This gene is located on chromosome 22 and it helps produce a protein that stops tumors from forming. As you might expect, patients who have neurofibromatosis type 2 are prone to intercranial tumors, including vestibular schwannoma. And my patient Jacob has bilateral vestibular schwannomas. For patients with NF2, symptoms tend to onset in late adolescent years, so in teenage years or early 20s, but sometimes even in the 30s or 40s. What made Jacob's case somewhat unique is that his symptoms onset much sooner than this around age five when he developed some skin lesions.

Although less common, skin symptoms can occur with NF2. And interestingly in Jacob's case, they were his earliest symptoms. At this point, he was not followed by our office at Phoenix Children's, but rather he was seen at an external dermatology

group. They performed a biopsy which had a histopathy consistent with schwannoma, and that ultimately led to his diagnosis of neurofibromatosis type 2. For patients who have NF2, it's very common to have routine MRI and routine audiologic testing. And in 2018, that was when we first started to do this workup on the patient, Jacob. His MRI at that time was consistent with NF2. He had bilateral vestibular schwannomas. On the right side, it was greater in size and in volume.

We know that for NF2, the size of the vestibular schwannoma does not necessarily equate to the degree of hearing loss. But what I have found interesting about this patient is that his right schwannoma has always been larger. It's been greater in volume, and he's certainly more impacted on his right ear today from an audiology standpoint. Doing this test in 2018, I would've been unimpressed if I had not known he had NF2. He had a very normal test. He had present distortion product otoacoustic emissions in both of his ears. He had present ipsilateral acoustic reflexes in both of his ears. His audiologic testing, his thresholds were well within normal limits. And his word recognition was excellent at a soft input level in both ears.

He was performing well and the family felt there were no concerns at this time. We're gonna fast forward to 2020. In between 2018 and 2020, what we'll see is that the vestibular schwannomas were increasing in size. They were larger. At his hearing evaluation in April of that year, he still had present distortion product otoacoustic emissions in both of his ears. But the slight change that we saw was that his ipsilateral acoustic reflexes became absent on his right side. His audiologic testing was within normal limits from 250 to 8,000 hertz, and he continued to have excellent word recognition at a very soft input level of 50 dBHL. Now, I broke 2021 up into two segments, early and late, because there was just a lot of testing going on at this time.

His vestibular schwannomas were increased in size, and then as far as his DPOAEs go, they started to become reduced on the right ear. At a soft input level, his word

recognition was excellent. In both of his ears, he scored 100% on the left and 96% on the right ear. And then at a louder input level, things were a little bit more reduced. He had an 80% score on the right ear and a 92% score on his left ear. Later that year, we saw that the vestibular schwannomas were stable. They were not getting larger in size at least compared to prior in the year. His DPOAEs on the right side were becoming more reduced. And what you'll see on this audiogram is that he started to develop a slight or a mild sensorineural hearing loss in his right ear.

Despite this, his word recognition was excellent in both ears. 2022 is when things changed. Although his vestibular schwannomas were stable in size, he started to perform more poorly from an audiology standpoint. At Phoenix Children's Hospital, at the office that I work at, the waiting room is connected to the office via a long hallway. And as you might imagine, lots of chatting goes on in the hallway. And I noticed that the patient was just wearing one AirPod, so he was using his headphone just on the left side. And I said to him, "Oh no, what happened to your right AirPod? Did it break?" And he said, "Well, maybe it's not working very well, it doesn't sound right, but my mom thinks it's fine.

Either way, I'm not wearing it because it doesn't sound good to me." And really that was my first clue that something was aloof. And so as we did his hearing test, I noted that his DPOAEs were now absent in both of his ears. His ipsilateral acoustic reflexes were still present on the left, but absent on the right, and then he still had his mild sensorineural hearing loss on the right. What you'll notice is there's some marked asymmetry between the ears, particularly at 2 and 4,000 hertz. But what is most interesting to me is that despite his hearing loss being mild, his word recognition was very poor at this point. So at an average or conversational input level, he was understanding only 36% of speech and that loud input level only 16% of speech in the right ear.

Despite this on his left side, his word recognition was excellent. He was understanding 92% on the left ear. At Phoenix Children's Hospital, it's fairly collaborative space and his providers quickly noted that his results were atypical in showing a change in his right ear. At this point, they recommended that he began a therapy called Avastin therapy to help with his auditory concerns. Avastin therapy is a tumor starving therapy that can be helpful for some patients who have NF2. Studies have shown some hearing improvement and tumor volume reduction in some, but not all patients who have NF2. You'll note in Jacob's case, he did not start his treatment right away, and you may wonder why. And that's because with Avastin therapy there sometimes can be poor wound healing, and Jacob was scheduled for abdominal surgery related to his NF2 at that time.

So they waited until after his procedure, after his surgery, to start him on this therapy. And for him, it's been quite a success. So in June of 2022, he had size stability of his bilateral vestibular schwannomas. At least at this point, they are not increasing in size. His DPOAEs, which were previously absent, were now present, although I'll note that they were still somewhat reduced but present now in both ears. His audiologic testing from a threshold standpoint is relatively unchanged. He still has hearing loss in his right ear, but most importantly his word recognition is so much better in the right side. He went from 36% word recognition to 88% once he started his Avastin therapy. I think back to when I saw him prior to this, and he seemed at least a little bit despondent to me.

Now he's hearing a lot better and has more pep in his step, if you will. I wish I had done a quality of life survey on him earlier in his evaluation, because I think it would probably capture that he was struggling when he was not hearing well. If you're wondering about his AirPods, they're both working and he's back to using two. A summary of Jacob. Jacob in many ways is a typical NF2 patient and that he developed hearing loss and bilateral vestibular schwannoma. In other ways, I feel he's an atypical patient, because

he developed his symptoms early at only age five. And his other symptoms have been, he struggled a lot with his skin symptoms and that can occur with NF2, but it's probably not as common as some of the more auditory related issues.

For Jacob, he's doing well on his therapy, which he's still undergoing today. The data for long-term Avastin therapy for patients with NF2 is somewhat limited, and so we'll see how that plays out in the future. But today he's doing really well and I would say he's thriving. He's planning for his future. When we put all of the pieces together, I want to highlight some things that stand out to me. The first is the importance of surveillance. So although I did not show you, there were many, many tests in his early years where he was having no change in hearing, but it was only through the routine testing that we were able to capture some of the problems that he was developing.

I also wanted to highlight that a minor change can have a big impact. So the slight change in introducing the Avastin therapy has had a big change on his life. I've highlighted the need for counseling and future planning. Although Jacob is doing well today, I don't necessarily think that will be true long term for him. I think he will continue to develop hearing loss, progressive hearing loss, and he needs to continue with his American sign language instruction. And lastly, I wanted to highlight patient report. When we're working with pediatric patients, sometimes they don't tell us what they're experiencing, but sometimes they do. And for Jacob, what he relayed to me was that his AirPods weren't working, but what he really meant was that he was not having good auditory quality.

He was not understanding well on that right side. And if we take the time to listen to our patients, we can learn a lot. Thank you for your time. And I wanted to introduce to you Dr. Ashley Geske, who's gonna talk to you about Noah.

- Thanks, Alyssa. Hi, everyone. So happy you could all join us today. The patient that I am gonna be talking to you about today, we are gonna call Noah. So Noah presented to our clinic at eight and a half years old and he was referred from an outside clinic due to inability to obtain behavioral testing. However, they did get type A tympanograms and present OAEs in both ears. His parents noted significant concerns for hearing. They also reported that he's failed multiple hearing screenings since kindergarten. However, has always passed the OAE screening. He has had several diagnostic evaluations over the years as well, however, results have always come up inconclusive. There is a history of speech delay since kindergarten that required intensive rehabilitation.

He also has a history of attending NICU stay due to premature birth. He was born at 35 weeks and was treated for jaundice with phototherapy while he was in the NICU. He also had an A-ABR hearing screening that he passed in both ears on day seven of life. And I did verify this via our Arizona database. He is currently in the process of being evaluated for an IEP, and recently qualified under autism. However, he has never had a formal evaluation. The school is currently waiting on the hearing evaluation results to kind of finalize everything, which is what brought him to our clinic today. So after case history, I went ahead with our testing. He had type A tympanograms in both ears.

His OAEs were overall present on both sides, kind of reduced and absent in some of the lower frequencies. And then his ipsilateral acoustic reflexes were essentially elevated and absent bilaterally. Noah was able to do conventional audiometry and with pretty excellent reliability. And this is the audiogram that we obtained. So moderately severe rising to slight sensorineural hearing loss in the right ear and then rising to mild sensory neural hearing loss in the left ear. I was also, or I also did some word recognition testing. I started out with a PBK. However, he seemed kind of confused by the carrier phrase and I couldn't tell if the errors he was making was due to his articulation or if that's how he was hearing it.

So I decided to kind of shift gears and move to a closed set picture-pointing task. I did a WIPI at 80 decibels in both ears and he got 80% in the right ear and 100% in the left ear. So seemingly decent word recognition. At this point though, I still recommended proceeding with a BAER study under anesthesia to assess for auditory neuropathy. And then I also recommended that he come back and do some additional testing in the clinic to kind of crosscheck today's results. So I saw him about a week later and I crosschecked some of his thresholds in the booth. Overall, things were pretty consistent with what we got on the last audiogram. And then I decided to focus my efforts on word recognition testing.

I did complete different presentation levels just to see if it is what I would expect based on his behavioral thresholds in the booth. And as you can see here, indicated by the red arrow, the percentage increases with increasing intensity and is appropriate based on his degree of hearing loss. So this just kind of further supports his reliability with the threshold task. About a month later, one of my colleagues completed a BAER study under anesthesia. They started with a high intensity click in both ears at 80 decibels. And as you can see here, they did not observe any wave V, which just to keep in mind, this is about 15 decibels above his worst threshold in the booth. So we would expect to see a response at this intensity level.

They then proceeded with a rare fraction and condensation run to assess for auditory neuropathy. And as you can see here, there is a very large cochlear microphonic observed and an abnormal ABR. It does look like there might be something kind of repeating around eight milliseconds. However, as we saw in the previous slide, when we go down 10 decibels, it disappears. So overall, this pattern of results, you know, is not what we'd expect for a typical sensorineural hearing loss, especially given the audiogram we obtained at the last couple appointments. So putting everything together, we made a diagnosis of auditory neuropathy. Reviewing results with parents,

they were very relieved to have some answers and were eager to move forward with intervention.

So I saw him shortly after the BAER study to discuss our next steps, and we decided to go ahead and fit him with some loaner hearing aids and do a full six-month hearing aid trial. At this point we know we have aidable thresholds and he, at this point, has seemingly decent word recognition. So we wanted to give him time to acclimate to the hearing aids and just fully, you know, assess his maximum potential with amplification. Seeing him back after the fitting, he loved his hearing aids. He was wearing them 11 plus hours a day. His parents and teachers reported that he is, you know, communicating a lot better and is much more engaged in class and at home.

However, they did note that he is still struggling when in background noise and when the speaker is further away. And they also reported that he seems to still be relying heavily on visual cues in conversation. During the six-month period, he also had his formal psychological evaluation and was diagnosed with autism level one. So fast forward six months, we ended up completing a CI evaluation just to assess his aided benefit and determine our next course of action. I repeated the behavioral testing in the booth. The right ear was overall consistent compared to the initial audiograms. However, we have seen a change in the left ear from two to three and it's six kilohertz in the left ear.

As you can see the OAEs below the right ear, the OAEs are very similar to the first audiogram. However, there has been a progression in the OAES in the left ear. At this point, I verified his hearing aids based on these new thresholds and made some changes, and then we popped him back in the booth and we did some aided testing. So I did monaural and binaural word recognition. And then I also did some speech and noise testing at two different SNRs. After getting to know him over the last six months

and talking with mom, I have realized that a WIPI is far below his vocabulary level and we determined that a CNC list is much more appropriate.

So as you can see in the middle column, he did very poor in every single condition. His aided monaural recognition was 16% in the right ear and 20% in the left ear when presenting at an average conversational level. So looking at these results audiologically, he is a candidate for a cochlear implant. All right, so where is Noah now? So at this point he is still in progress. He is on our upcoming agenda to discuss at our CIT meeting and give our formal recommendation for implantation. This says still pending MRI. He had one very recently and everything came up totally normal. No, nothing was seen that explains the auditory neuropathy. He did have a genetic ANSD panel that found a pathogenic variant in the IDUA gene, which is associated with mucopolysaccharidosis type 1, which is Hurler syndrome, Hurler-Scheie and Scheie syndrome.

The geneticist also recommended some enzyme testing and a urinalysis and everything came back normal. So there's no disease present. He's likely just a carrier for this gene. And they noted that this is probably just an incidental finding and does not explain the hearing loss. So at this point, the etiology is still unknown, meaning that we're unsure what type of outcome he's gonna have with a CI. But based on the significant challenges he's having communicating every day, in addition to his poor word recognition scores, you know, his parents and I think that a CI is the logical next step and it's definitely worth a shot to help him hear better. So now I just wanted to step through how we came to this diagnosis and the unique pieces that I identified that led me to believe this was auditory neuropathy even before completing the BAER study.

So number one, he relies heavily on visual cues. The second I brought him back from the waiting room, it was very clear that he relies heavily on visual cues to understand

what's being said. At this time, we were still wearing masks and he was not able to understand me until his mother pulled down her mask and repeated what I said. His OAEs were much more present than expected based on the degree of his hearing loss. He had a speech delay from a young age that did require intensive rehabilitation. He has essentially absent, you know, slash elevated ipsilateral acoustic reflexes, which disagreed with the degree of hearing loss. He has a lack of fluency in his speech.

It's very broken up and he also speaks very slowly and carefully, almost as if to not make any kind of errors. And lastly, it was the high level of concern from the parents. This is not the first time I have encountered acquired auditory neuropathy. And one of the trends that I have seen is the worry and urgency in the parents' voice. You know, they're with their child day in and day out and they can see how much they're struggling. So I think it's super important for us to remember that parent rapport and involvement is a crucial part of their child's care and they are a key member of the care team. Thank you so much for listening to my presentation today.

I hope you learned a little something, and then now I'm gonna hand the floor over to our next presenter, Dr. Deborah Flynn. She has a super interesting case for you guys.

- Thank you, Ashley. That was a very interesting case. I appreciate it. All right, and I'm gonna continue on and introduce you to Ethan with another case of an abnormal auditory brainstem response study. Before we do that, I'm sure many of you today conduct ABRs, but for those of you who don't, I just wanted to point out a few things that we typically look at when we're considering an ABR to be normal or abnormal. So this is a click, a high level click stimulus, and we're looking specifically at waves I, III, and V. And we also look at how the waveform responds to multiple stimuli such as rarefaction and condensation. In addition to that, we're also looking at frequency specific information and following our wave V down into a threshold level.

So that is a normal typical ABR that we're seeing. Let's take a look at an abnormal ABR. So this is similar to Ashley's presentation where we saw auditory neuropathy. So this is also a high level click stimulus and rather than a clear wave I, III and V, we're seeing a cochlear microphonic that is reversing impolarity out to several milliseconds. The bottom recording here is used that we pinched the tube to kind of show that this is not artifact, that it actually is a stimulus from the cochlea. All right, so let me introduce you a little bit to Ethan, now that we've seen a normal ABR and auditory neuropathy. So Ethan presented to our hospital at 53 days of age for an MRI and for an ABR under anesthesia.

Took a case history with his mother and she indicated that he was born full-term without complications. He has an older sister with no other health concerns and no family history of hearing loss. He failed two inpatient and one outpatient newborn hearing screenings. And at 16 days of age, his mom noted that he had some nystagmus. And so she brought him to the pediatrician. And in the pediatrician's note, he mentioned that the child had horizontal nystagmus that was present in all directions and was non fatiguing. Because of that, because of the nystagmus and the failed hearing screening, he immediately referred for the ABR and the MRI. So while after his MRI, he came over to our area where we do our sedated ABRs.

And I started out with tympanometry, and because he was little, we did 1000 hertz probe tone and they were nice peaked normal responses. Distortion product otoacoustic emissions were present and very robust. Again, I typically start with a high level click stimulus just to get a general idea of what we're looking at and the initial response. When we presented it, we immediately got a pretty abnormal response. So at 80 dB we can see a beautiful wave I and II, maybe an emergence of wave III, but other than that, the remainder of the response is pretty abnormal. So not able to follow a wave V down to threshold with this type of waveform, but I was able to follow that wave I.

So decrease the intensity down to a level that was essentially within normal range. And since he was under anesthesia, I wanna do as much as I could. So I presented the 80 dB click to a rarefaction and condensation, and you can see here that they actually aligned pretty well. So there's no reversal of a cochlear microphonic in that early stage. We still have a beautiful wave I and wave III. I completed bone conduction, and once again, no wave V, but was able to track that wave I down into the normal range. Tone burst testing, I did at 500 and 4000 hertz, and once again 4,000 hertz was tracked down essentially into normal range. While I was doing the testing, I contacted the radiologist just to kind of get an idea of what she was seeing on the MRI results.

And she indicated for the most part that the myelination patterns were very normal for his age. She didn't see any abnormal fluid collection and she saw a really well-formed corpus callosum. They also did a temporal bone study given that he failed his newborn hearing screening and they thought that he might have some hearing loss. This too was also very normal, and so the inner ear structures, including the semicircular canals and the cochlea and the vestibule were all unremarkable. He did not have any vestibular aqueduct enlargement. And so her outcomes were a normal brain and temporal bone. All right, so in reporting the results to the pediatrician and anyone kind of going forward, the most important thing that I wanted to point out was that this was an abnormal ABR, and I was very specific about describing the components of the waveform.

So those wave I and II were present and robust, but the later components were absent. Overall, these findings are very consistent with the disorder at the level of the auditory brainstem pathway. What we would consider of neural hearing loss versus an auditory neuropathy. When talking to the mother, it's always very difficult to, you know, go over or discuss test results with a parent and have to tell them that the ABR was abnormal and more so when we don't have any really good information about how we expect

Ethan to hear in the future. So I talked a little bit about the difference between detection and discrimination, similar to what I would do with a child who's been diagnosed with auditory neuropathy.

And mom was very confident in stating that she felt that Ethan was able to hear sounds and that he even startled to noises. I was also cautiously optimistic given that he had very present otoacoustic emissions and his wave one was measured down into that normal range. So my recommendations to his mother was to repeat the ABR in three months. And in the meantime, I would not wanted her to see an ear, nose and throat doctor. She had already been referred for ophthalmology and was waiting an appointment. We got him a referral over to our Arizona Early Intervention program. I really encouraged mom to get involved in our Arizona Hands and Voices and guide by her side.

So we have a very active program here in Arizona that can be helpful, and I knew that this was gonna be a long path and journey for her given that we didn't have any definitive results as to how Ethan was gonna hear at this point in time. And then most importantly, we wanted to get him into neurology. Ethan returned for a follow-up ABR in at three months. He was still under six months of age at that time, so I was able to do a non-sedated ABR. Mom reported in her interim history that she had seen the ENT. They didn't have any specific recommendations other than to follow up with the BAER study as recommended.

He had also seen an ophthalmologist and they did not feel that he had any need for treatment at that time. And then the neurologist told mom that the ABR, I'm sorry, the MRI was normal and that they wanted to see him back in one year. So before we started the ABR, we did a tympanometry with 1000 hertz probe tone, which was peaked and normal. He had very present and robust otoacoustic emissions. And then he was an amazing baby and sat beautifully for his acoustic reflexes, which were also

present. So having these present emissions once again kind of ruled out that auditory neuropathy component, 'cause typically they would be absent. So because he was without sedation, I didn't want to jump in with a very high intensity click stimulus.

So I started out at 60 dB NHL hoping that his ABR had resolved. And this is the waveform that emerged. So results were still pretty grossly abnormal. And since I had completed all of the other testing while he was under anesthesia, I decided to do a rate study. So this is his rate study here. So at higher intensity levels, we see that wave I, II and a little bit of a III emerging. But as we decrease in stimulus rate, we start to see that III really perk up and then a V emerge. Overall, the waveform morphology is still grossly abnormal and the latency is very delayed. I did wake up before I got an 11.7 response on the left side, but once again, you can see a little bit of a five kind of peaking out there.

All right, so my recommendation to mom at that point in time was to return for behavioral testing. Mom would continued to be very consistent in her feeling that he was hearing what was going on in his environment and responding to sounds. So at eight months of age, he returned and he was an extremely alert and attentive eight-month old. He was sitting with support, but also had really good head and neck control. Mom was insistent that he heard all of his family members' voices, particularly his mother. And even a couple days before the appointment, his sister had turned the television on behind him and he actually turned around to try to find it. So we put him in the booth and presented some sounds, and he had excellent localization both to the right and to the left speaker, responses that made it clear that he truly was able to hear his environment.

His otoacoustic emissions were still present and robust and he had normal tympanograms at the time. My recommendations to mom at that point in time, because the neurologist really didn't wanna see him for a whole another year, was to consider

getting a second opinion for neurology. And I also talked to her a little bit about genetics testing and then a speech language evaluation. So at this point he was eight months old. And we have an AVT therapist here that specializes in kind of that early communication component, and we wanted to get a baseline and make sure that he was on track. We're gonna continue monitoring with ENT and audiology and then, of course, his AzEIP services. The second opinion, a neurology consult confirmed that he needed genetic testing, so they referred for genetic testing, and they also recommended serial MRIs.

He followed up consistently with audiology and ENT and ophthalmology, and all of the time undergoing his AzEIP services. So for Ethan, the key component that kind of helped us figure out the reason for his hearing loss was his genetic results. So his genetic panel found that he had one gene for the USH2, which geneticists considered non-significant or undetermined significance. But the key gene panel was the GJC2 and not to be confused with the GJB2, which is a hearing loss gene. This one is associated with hypomyelinating leukodystrophy. So both of his parents were tested for this gene and his mother was found to have a pathogenic variant. So he is currently waiting to be seen at a clinic across the country in the leukodystrophy clinic.

This particular leukodystrophy is typically presents in neonatal, an infant period, and it is slowly progressing. It starts out with nystagmus, delayed motor development, ataxia, dysarthria, and then progressive spasticity. There have also had some mild peripheral neuropathy documented, and then, in rare occasions, hearing loss and optic atrophy. Ethan has gone on to have serial MRIs. Overtime we're finding that his myelination is abnormal now. He has some hypomyelinisation in the occipital, parietal and temporal lobes. He also has some hypomyelinisation in his corpus callosum now and some general volume loss. He continues to develop slowly. He's not walking at this time, or, at least, wasn't at the last time that I saw him. So we'll see in a couple of weeks where he is today.

These are his behavioral tests over time. Once again, feel very fortunate, because he's an excellent tester. His behavioral responses have all been in the normal range. When he comes in next time, we're gonna work with some insert earphones and work on trying to get some ear specific information. So overall, I wanted to present this case to kind of show the difference between auditory neuropathy and more of a neural type hearing loss. So with a neural type hearing loss, wave I does not reverse impolarity with their rare and con stimuli. And oftentimes you can track wave I down to softer levels. With A NSD, we see that early cochlea microphonic reversing out to several milliseconds. Some of our key features with Ethan was early diagnosis of the nystagmus and the abnormal ABR.

He is clearly not an ANSD case. And for him the key component was his genetic panel. So finding out, you know, his diagnosis that kind of plays into what we're seeing on the ABR. He has had MRI changes over time, but his behavioral responses and his auditory development at this point in time seem normal and appropriate. He continues to have present acoustic reflexes and present otoacoustic emissions. I thank you for listening to my presentation and at this point in time I'm gonna turn you over to Wendy Steuerwald who is going to open up to questions.

- Okay, hello, everyone. Oh, my goodness, you guys, I'm so impressed. Those are such great case studies. I learned a lot. Okay, we have some questions. And for the audience, if you have questions, please enter them into the Q&A. First question we have is for Lynn, someone is wondering what is an incomplete test?

- An incomplete test with a newborn hearing screening, we would consider like a stop and a start as an incomplete test. So if they started the screening and something went wrong, let's say the electrodes came off or the earphone fell out, they may stop the test. Or there are times where when the babies aren't passing, they stop it before the

test completes all the way and then restart the test. So we would consider it incomplete. If you basically didn't get that pass or fail result, they've stopped it before you've gotten that result.

- Is there any sort of number of times, Lynn, that you can do that? I mean, like, if you start the test and the baby starts to cry, is it okay to stop or should you keep going, or how does that all work?

- It's hard to, I guess, have a number on that, but at least from our standpoint when we look at that, we really don't wanna ever push start on that test until the baby is swaddled calm, fed. So we can know that our test is gonna be complete and accurate, but that is something that we do track in our own sort of screening world here. We look at the stops and starts and the incompletes. And we don't want a baby to basically be screened more than five times. So meaning that we consider that to be an incomplete, including, I'll see if I can explain that. We want that each ear was tested one time and let's say, that baby didn't pass, so then you come back and test that baby again and you stopped and started and then you wanted to start again.

So that would be the third, and then the next pass or fail result that you get from each ear would be four and five. I'm not sure that explained that clearly without showing it to you, but ultimately we don't want the baby to be screened five times. So how that looks, that really gives you one time to stop and start the test without, we kind of think, over-screening, 'cause situations do come up when the baby does just get too mad and you're in it and you have to stop it. But ideally, you should never have to do that.

- Sure, there's actually a comment saying there were so many of those with the tests and it's like, yes, yes.

- Yes.

- On your presentation there was like how many, like 11?

- 11 at least on one side. And yeah, that is a situation where when we do see that in the state database, a screenshot is usually sent to me, and then if that's our screeners then we have a discussion. But again, we're tracking that, so half the time I already know about those babies. But if it is from an outside facility, we go ahead and pass that on to the state letting them know like, hey, this might be an education opportunity, you know, for the site. And we do have hospitals that we do see that from pretty routinely. I think it's just a lack of education and probably support in that scenario.

- Sure.

- Or lack of maybe audiologist oversight, I'm not sure.

- That makes sense. The next question is for Maddie. Maddie, what is an acoustic seizure?

- So an acoustic seizure is also known as an audiogenic seizure and it is a seizure activity that is prompted by, like, a loud input level or in response to sound specifically. In Walter's case, he has not had any of those presentations, but that is something that's associated with the GIPC3 mutation, and so it's that seizure activity in response to loud stimuli.

- Wonderful. Maddie, another one for you. Can you review the GIPC3 mutation again?

- Yes, so again, that GIPC3 mutation was something that was new to me as well. And what the literature review found is it's associated with that moderate sensory neural hearing loss, those acoustic or audiogenic seizures that I just described. But most

specifically there is kind of a disarray of the inner and outer hair cells, more specifically those inner hair cells looking at kind of that they're not quite clustered as well, they're not quite as strong and you're seeing the impact at a hearing level because of that. And I've also included my references for the GIPC3 if anybody wants to go and review those articles. One was specifically looking at the mutation in some mice models, which I found really interesting.

And then the other one is a complete literature review of all of the available articles regarding GIPC3, which provides an excellent overview.

- Great. That's very interesting. Just another quick question, 'cause we're just about out of time. Allie, where can people get recorded Spanish material?

- Yeah, so we got all of ours from Auditec. It's just auditec.com, A-U-D-I-T-E-C.com. Super quick and easy process for us to order those and get them in quickly.

- Awesome, and a follow-up question, how would you recommend people score those if they don't speak Spanish?

- A really great area, I wish there was a really clear answer to that. I really think the only solution is to do your best. I know at a cochlear implant conference I was at a couple years ago, there was some discussions about this and there really is no standardized way unfortunately. You know, I'm lucky that I took enough years where I understand a fair amount and feel comfortable scoring it myself with repeated words, but I know a lot of our other staff does not. So I think really it's just evaluate your resources. If there's an interpreter you can use that might be great. If you can use the family, it definitely takes a lot of coaching to get them to present at the right level for monitored live voice, things like that.

You know, they can present 'em, they can also help score, but you really wanna make sure they're not biased. You know, this mom and I are pretty well in agreement on everything, so I was very fortunate that it's easy, but it's definitely not always the case. You know, there's also been some stuff when I was researching there's really not a clear answer. And one of the suggestions was to consider using like a non-speech test to assess discrimination. One of 'em talked about like dichotic digits. I just didn't really dive too much into that quite yet.

- Awesome, okay, this is the final question, 'cause we're out of time, but I just need to ask it. All of your cases dealt with a lot of cases that were not clear, they didn't go in a straight line, and you weren't really sure what was gonna happen next. How did you counsel the parents on everything that was going on when you weren't really sure what was gonna happen?

- I can start with this one. I think that especially in pediatric cases, you're working with the family as the whole. I think transparency is like a key piece of working with those families, and kind of being upfront and honest with them, with the information that you do have, talking about the information that you want to get and why you want to get that information, and kind of building that trust with families. And the more honest you can be with them, even if it's, you know what, I don't know why this is happening right now, but we need to do some more research. I think that that helps kind of build and strengthen the relationship you have with a family even when there's twists and turns.

So if something more comes up then they're prepared for that.

- That makes sense. Okay, well, I want to thank our presenters today. You guys are awesome. I learned so much. Thank you for your time and sharing your expertise. And audience, thank you for joining us today. And as you know, this presentation is being

recorded, so you can go back and watch it again anytime. Okay, thank you all. Have a good day. Bye.

- Thank you.