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EHDI Annual Conference

Family Perspectives on Navigating a CHARGE Syndrome Diagnosis

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>> Good morning everybody, my name is Jamie, I'm the room moderator. Just to let you know there is a few exit signs. If there was a fire, exits over there and behind here.

We are limited on time, and I'm sorry to rush everybody, but we have another fabulous presentation going on. So without further ado --

>> PENNI ECHOLS: Thank you so much, and thank you ladies for sharing your story. It is wonderful to see parts of our stories represented in yours, and we are happy to be part of the CHARGE family with you.

My name is Penni Echols. I wear many hats but the hat I am wearing today is that I am a member of the Board of Directors of the CHARGE syndrome foundation where we work to improve the lives of all people with CHARGE syndrome.

I'm joined today by Iris and Staci who are going to tell you part of their story, mostly relating to their child's first year or so and getting that diagnosis with their hearing.

Babies with CHARGE syndrome are often born with life-threatening birth defects. They spend many months in the hospital and undergo many surgeries and treatments. Swallowing and breathing problems make life difficult even when they come home. Most have hearing loss, vision loss, and balance problems that delay the development and communication. And despite the seemingly insurmountable obstacles, children with CHARGE syndrome often far surpass their medical, physical, educational, and social expectations.

The name CHARGE was a clever way in 1981 to refer to a newly recognized cluster of features seen in a number of children. Over the years it has become clear that CHARGE is indeed a syndrome, and at least one gene causing the CHARGE syndrome has been discovered. The letters CHARGE stand for coloboma of the eye, heart defects, atresia of the choanae, restriction in growth and development, and ear abnormalities including Deafness. These features are no longer the only factors in the diagnosis, but we are keeping the name. We love it. We love to CHARGE on.

Today my friends will share part of their story, and then I will wrap up to help you identify some of the barriers that our family stays in meeting their one, three, and six benchmarks. These are our kids. They thrive.

>> IRIS FILKINS: Hi everyone, I'm Iris Filkins, one of the Colorado state liaison for the CHARGE syndrome foundation. And this is my family. My husband Kyle and I are local to the Denver area, we are about 20 minutes west of here in [can't understand]

This is my daughter Remy, this is her sign name, and we also like to call her Remy Roo and Remy will be three years old next month and she has bilateral cochlear implants and loves Sesame Street.

To get us started, because our family is learning ASL I thought it would be fun to teach you a couple of science from Sesame Street. If you do the letter E and your nose, that is Elmo for his big orange nose. You can do Big Bird, Cookie Monster, and those are some of Remy's favorite signs right now. She uses 50 or 60 expressive signs. Obviously some days are better than others, but she is picking up ASL. With her cochlear implants we are still not quite sure what she can hear, but she was an everyday with her glasses. We are hopeful that since she realizes that sound is infected meaning.

When he also attends a toddler program here in Denver at the anchor center for blind children. We have a very inclusive program that we were able to incorporate ASL into using Remy's community intervenor for Deaf/Blind children.

And we are planning for her to attend preschool in the fall. We are just now starting the IEP process. And trying to figure out what accommodations and resources Remy will need for schools and she is Deaf/Blind.

So that leads me into Remy's medical journey. She has been a warrior since day one. She was born at 34 weeks. My pregnancy was very normal but had extra amniotic fluid that triggered an early labor and an emergency C-section. Remy was in the NICU for seven weeks and it echoes a lot of take's story. She had three surgeries in those first weeks and what felt like diagnosis after diagnosis. It was the shock of a lifetime and a diagnosis I don't think I can begin to explain right now.

Kate did a great job. She has come very far and I thought she did a great job because it was a shot of a lifetime and we were not only learning a lot about Remy at that time but also ourselves in the medical field and everything that a CPA knows nothing about. I prefer to be in spreadsheets, so this was definitely an adjustment for us.

So that leads me into her diagnosis. Remy was diagnosed with CHARGE syndrome at one we call. She has a lot of the classic characteristics of CHARGE syndrome including bilateral colobomas of her retina and optic nerve. She has microphthalmia of her right eye. She had choanal atresia which was a complete blockage in her nasal passage. That was her first surgery at five days old too she also has internal and external ear abnormalities, cranial differences, and malrotation in her intestines. That was her second surgery at six days old, right after the choanal atresia she had an emergency surgery the next day. And then silent desperation was detected in the NICU resulting in her getting a G-tube.

Those were the first three surgeries and less than six weeks of life.

Leaning more into her hearing loss diagnosis, a lot of the life-threatening areas of the choanal atresia and the malrotation, those were priority number one when we were transferred. But to get there we had to be transferred to the level for NICU because Remy was born in a small hospital in northern Denver and they just were not equipped to handle what they were seeing with Remy. A similar story where she was taken to the NICU, we get general information but they just had no answers for us. So when Remy was two days old she was transferred to Rocky Mountain Hospital for children and they did two rounds of CT and MRI imaging to learn more about her anatomy. Originally they were trained to get images of the nasal passage because they could not get the NG tube down, but in that process they were also trying to see her middle and in her ear, her auditory nerve, and really anything we could get a sense of. Unfortunately they did not see an auditory nerve in the imaging, but they were able to confirm that Remy had malformed cochleae and missing semicircular canals which are very common with CHARGE syndrome.

After that imaging, Remy was referred for a sleeping AVR which is the auditory brainstem response test both in the NICU and also outpatient. The AVR results were mixed though. She had three sleeping AVRs done and we originally thought that Remy had moderate to severe hearing loss, so she started using hearing aids and three months old. But we never saw a response, like ever. And it is easy to say that a child is young and close to a newborn and isn't responding and they don't know sound as meaning, but when she was six months old we decided to get ear tubes to drain fluid and see if that would improve her hearing. And that time is sedated AVR was done, and we found out that Remy's level of hearing loss was more significant than we originally thought. So the conversation switched from using hearing aids to cochlear implants.

But Remy is obviously a very complex little girl, and that applies to her ear anatomy as well. So we had to consult with a few searches to find the right team for Remy. That included both Dr. HUPO at Rocky Mountain your.

Center to be her cochlear implant certain, and also Dr. Mitchell at Rocky Mountain pediatric ENT to be there to evaluate her narrow airway, to intubate her, to be there for added support because with CHARGE syndrome it is never just one thing. There are 10 to 20 things we are considering and sedation was never easy.

Both surgeons were present for both for cochlear implant surgeries which was at 13-month-old for her right side and then at 21-month-old for her left side.

So with all of that, Remy has said both cochlear implants for over a year now. And our communication approach is still primarily sign language. We get ASL lessons through early intervention with our county, and Remy also has a community intervenor for Deaf/Blind children and helps us when we are out and about. We are also using spoken language with Remy, and we narrate everything we can for her. But we are still not sure what speed she can hear. There are definitely sounds in the higher frequencies like battles or musical instruments or maybe her feeding pump beeping when it is done that Remy can year, and it is repeatable.

But still she is not using spoken language, and so we are still focused on ASL. And we also realized the limitations of technology. And that sometimes the CIs fail and that she will need language even without those devices.

In the very beginning we were also very focused on touch cues. In the NICU we were told that Remy was Deaf/Blind. We knew that she had impairments in both that were going to make it to where she had a dual sensory loss, and then the NICU as a parent that is terrifying. I don't know any other Deaf/Blind people in my regular life. There is obviously a few stories that you hear.

So we wanted Remy to have positive touch. I an accountant, hospitals are scary, and we wanted her to know our touch. So we did touch cues if they were going to come into do anything with her nose, we would tap not only one nostril but two if they were coming in for a second swab or a second sailing. Or we would touch her eyes, if they were going to take a leadoff and I would sign I'm sorry and they would take it off, just so they have that -- and I guess that puts us in the total communication realm for Remy people are we wanted to have access to language no matter what the language looks like. So we are also trialing AAC devices to see what works for Remy.

But you want to communicate with each other nothing about five ways of doing it. The AAC devices new but like anything else we're trying to figure it out for her.

If I could go back, what is one thing I would tell myself. First I would tell myself to stay strong and that you will find the strength, even if you don't think you well.

Secondly is to connect with other medical parents. Those connections are both on a personal level but also a knowledge sharing and resource sharing level. Making those connections will be crucial for navigating this unexpected journey, because parents know this life better than any professional can. I am so happy that people are attending the session to get a different perspective for to make a connection with a parent, because parents are the ones that know this world inside and out. They are the ones who know all of the tips and tricks like what headbands to get on NCR want pilot caps work on their babies or tips for keeping the magnet of the CIs on.

When Remy got activated, her audiologist, Allison, told us these will fall off 100 times a day. But I want you to put them on 101 times.

And I thought to myself there is no way. This has to be an exaggeration, 100 times, how are we going to do all of this? And sure enough Remy's CIM tracks the number of coil office and she was averaging close to 200 times a day in the very beginning.

But another mom recommended a headband on and see which led me to these really cool plastic pieces that I attached to Remy's classes, and now that is one less accessory we have to manage. Enter processing wearing time increased significantly.

Those other cool things that parents are not only creating for their kiddos, but sharing with other medical parents. So that would be my one piece of advice for myself but also ratio) Sammy today. I want to thank you for letting me share Remy's story, and I will let Staci tell you theirs.

>> STACI PETRALBA: I'm Staci Petralba, an audiologist and a mother of four-year-old Allison and two-year-old Benjamin was born with CHARGE syndrome. My family and I live in Las Vegas.

This is my son Benjamin. He will be three years old in May. He currently gets around by crawling or in his gait trainer. He is 100 percent tube fed and he has never done oral feeds. He wears a hearing aid in the right ear and a cochlear implant in his left ear. He communicates by vocalizations and signs. He has about 20 to 30 words that he says or signs. He really enjoys reading books, brown bear and [can't understand] are his current favorites. And he would be the happiest person in the world if he could just sit in your lap and rock himself back and forth all day.

Benjamin was full-term at 38 weeks. We did not know anything was wrong before he was born. It was a typical uncomplicated pregnancy. All I did have was extra fluid, but my doctor was not really concerned about it. I went into labor spontaneously and he quickly made his debut.

After Benjamin was born the nurses became concerned pretty quickly. He would pink up when he was crying, but if he was not crying he was turning blue. So they immediately were suspecting choanal atresia and his was in both nasal passages so he wasn't getting any air at all unless he was crying.

He was taken to the NICU right away, and two hours later they came back into the room and informed us they had to intubate him to stabilize his airway and there was a holistic things I have found including severe and life-threatening heart deficits, choanal atresia, abnormal ear shape, and some other things I don't really remember.

He was then transferred to a different hospital where the children's cardiac program is, and the next week was spent getting to know Benjamin which really just means doing a bunch of tests. All of which found more things wrong with him.

After a few days of this I just wanted to tell them to stop doing tests, we have enough diagnoses, we don't need anymore. We found out about kidney of normality is, he had a brain bleed, he had colobomas of both eyes on the optic disc, etc., and we have added more along the way.

After an open heart surgery, he was transferred out of state to primary Children's Hospital in Salt Lake City for his choanal atresia repair because there was not anyone in Las Vegas who was able to do that.

We heard the term CHARGE the day he was born and they were pretty much immediately suspecting CHARGE because he has all of the major diagnostic criteria. And I think what really points physicians to CHARGE is the combination of choanal atresia and colobomas. Because those two occurring together is pretty rare, so usually it means CHARGE.

It was helpful to have that diagnosis so we knew what we were dealing with, and to help us find resources. And later on that was confirmed with CHD 7 mutation with genetics.

The hearing loss diagnosis. Benjamin had his hearing test per my request at primary Children's Hospital while we were waiting for his choanal atresia surgery. He never actually failed a screening because they were suspecting CHARGE, they just want to do a diagnostic ADR right away. So he was about three weeks old when he had his hearing test, and by that time I do my research on CHARGE and I pretty much knew they were going to find some hearing loss. And really I was just hoping that he had an auditory nerves, because that would mean that we could at least two cochlear implants.

When I found out the hearing loss in his right ear was in the mild to moderate range, I was actually pretty excited about that.

When they got back to Las Vegas and he was discharged from hospital, he went on to have ADRs with early intervention in a private practice audiologist confirming those results.

Choosing a communication approach for Benjamin was not as straightforward as it may have been if he didn't have the additional medical issues. As a principal, I believe in total communication. But in reality in the first year of life, we didn't really know what was going to work. He was not really responsive to interaction and it was not until he had his first major heart repair at nine years old and another and we surgery 15 months old that he began to make improvements developmentally and became more interactive and responsive.

Early on I had a really hard time with his vision diagnosis. Initially we did not get much information about what is functional vision was like. We were just told that he has colobomas, they are big, and they are covering his optic disc. It wasn't until Benjamin was about a year and a half old that we were confident that he could see well enough to sign.

To this date we have not received much support from early intervention with introducing sign language. Our speech pathologist with early intervention works on general language stuff, but she doesn't really sign or have much experience with hearing loss. We recently met a semi retired teacher of the Deaf who we are paying privately for tutoring sessions as she has had the most awful tips for helping Benjamin. So Las Vegas does not have the wonderful resources that these other ladies have experienced.

Voice quality was another concern. After Benjamin's major heart surgery at nine months old, is left vocal cord got paralyzed from the surgery. So for about two months we didn't hear any sound. No crying, nothing. It was terrifying. Since then his voice quality has improved significantly, but at the time we didn't know if he was going to be able to use his voice.

Benjamin got his hearing aids at four months old, and his cochlear implant in the left hear that two and a half years old. There was some delay in getting the cochlear implant due to other surgeries that were more important and due to having to coordinate care out of state because he gets ENT and audiology in Salt Lake City, so we travel for that.

I made the decision to pursue a cochlear implant for his left ear because it was impossible to get his left hearing aid to meet prescriptive targets without feedback. And I just did not feel like he was getting much access to high-frequency information without an implant. So I also felt like a cochlear implant would provide them with more stable access to sound because we would not have to worry about fluctuations in hearing due to middle ear people issues are often common with CHARGE syndrome.

We have struggled a lot with keeping Benjamin's hearing devices on. You believe them on if you are holding him or doing something with it, but as soon as you walk away or set him down they are off and he is chewing on them. That is a challenge we are still working on, and there is a session on that later today so I might be going to that, how to keep the device on, I need all of the tricks.

Looking back, what is one thing I would tell myself. I would say just don't second-guess yourself. Trust your mommy instinct. It's easy to look back and think what if we did this or what if we did that, and I will just say be confident and know what you are doing is best for you and your family.

>> PENNI ECHOLS: Thank you also much for agreeing to do this. It is so hard for me to tell my story, and I take so much joy in watching you develop your own and sharing that with everyone. Thank you so much.

This is a short list of some of the reasons why children with CHARGE syndrome and other medical complications don't meet their one, three, six benchmarks.

I don't want to read them all, it would take too long. You can look at our slides later.

A CHARGE syndrome foundation help. We are so excited, we have a full-time outreach director named Lourdes [can't understand] and she speaks Spanish. She is the nicest and most wonderful person you will ever meet. If you come across a family that is struggling, she can get you connected to liaisons.

We have medical travel assistance for families that need to travel and additional costs that are not covered by insurance. Recreational grants for families. Tons of information for professionals, including these grant opportunities that are not always claimed all the way. Not just for scientific prefer clinical as well.

We invite everyone to come to our conference. It's the best time ever. Family to family support matters. Find your state's liaison. We are very close to having a liaison in every state. States that do not have volunteer parents have a regional facilitator that covers the area.

State [can't understand] projects. Would you like to come to conference in 2025? Is going to be a great party.

Finally we invite everyone to come to our walk and roll in May, it's a good time for families to get together, it is free to participate, and we would love to party with you.

Thank you so much.

>> Thank you guys for coming, and also make sure that you send some reviews in the registration. There is a little scanner so you can give some feedback, and also get in contact with the presenters. If you guys have further questions, are you guys available in the hallway? There is such limited time in such great resources right now. So thank you again for coming. Thank you guys.