Welcome to the Orange Socks Podcast, where we are inspiring life despite a diagnosis. I'm your host Dr. Gerald Nebeker. I was honored to interview Kristy and Andrew, their two sons grandma and grandpa about Hadley who has a diagnosis of Pfeifer syndrome. I enjoyed learning their story and I know you will too.

Kristy: It was kind of a long road. It started as just a normal pregnancy. We were thrilled when we found out it was going to be a girl. Then we went for our anatomy scan at 20 or 21 weeks, and they kept saying "oh her head is showing up really big." We just laughed about it because we thought it was Micah, our middle child who is just a big kid and we didn't really think anything of it. I should have because another ultrasound tech came in and did the measurements also, but I didn't. I've seen a lot of other ultrasounds so I should have but I didn't. Cut to that report gets sent to your Obstetrician who is my friend called me and says "it's not a big deal but I need to refer you to the maternal specialist because of some things that showed up on that ultrasound. And we thought it was no big deal. We were really busy at work opening our brand-new women's tower, I work in Labor and Delivery, so I know a lot about what was happening. I thought okay no problem I'll just walk across the building to the doctor's office while I was working. I didn't think to bring Andrew because I thought it was a quick little check-up. They started the ultrasound and its very obvious something is wrong. The tech got the doctor in the room and its someone who I've known for about 10 years at that point. Things just started unfolding. They didn't know what it was, they just knew something was very wrong. So that was terrifying. I was sent back to work and I was obviously a mess. The doctor couldn't see a kidney, something was wrong with her head but he didn't know what it was. So, the question was "what do we do now?" So, the next day we went to a fetal MRI which is really crazy to do when you're pregnant and you have to hold still but your baby is moving. The next day was an amnio. Then cut to the results of the amnio. Everything on the amniocentesis came back okay, like it's not this and its not that. It turns out that unless you are really searching for a certain type of what skeletal dysplasia

Andrew: Gene panel.

Kristy: Yeah, you have to know what you are ordering to narrow it down. So, it took the help of a second specialist to figure it out. She says "I'm 95% sure it is Crouzon's and don't Google it.

Gerald: Kristy of course Googled it. What you see when you Google it is old findings and the most severe cases. It makes it easy to jump to the worst-case scenario.

Kristy: So, you just jump to the worst thinking "is she going to be okay? Is she going to look like a little baby?" Your mind just goes in 10,000 different directions.

Gerald: During this time Andrew was also meeting with doctors and the doctors were still looking for definitive signs and symptoms but they weren't finding what they were looking for. It was only when they were referred out to another specialist that they ended up with a more concrete idea of what was going on. Andrew felt afraid when they specialist started talking about a Pfeifer diagnosis.

Andrew: Dr. A, we met a couple of times, and we weren't quite getting the direction that we were looking for, so it was only when we were referred out that we ended up with a more concrete idea of what was going on. That was basically the 1 major Dr.'s appointment I ever missed because of a work obligation, was the 1 where we kind of really nailed it down that Hadley had an FGFR2 gene mutation. I haven't made that mistake since. Initially there is fear, you know we talk about the Pfeiffer syndrome diagnosis, and I lot of that is centered around how we can accommodate her but also more on how she looks. As time has gone on it has become less about how she looks then about her overall total care needs. I think at some point in time the initial reaction was "what is it going to be like to have a child that looks different and has some needs?"

Kristy: But will act like a normal child, she will be walking and talking, and her biggest problem will be "what color of bow should I wear that day"

Andrew: Or social pressures, and ultimately, we are not dealing with social pressures. That is kind of a relief in some senses but kind of not. Initially just shock and then you work through it, and we had a dear friend come that next day and sit with us for a couple of hours and be able to just support us. And so, friendship communities that existed and exist as a result of the diagnosis and connecting with people online which is in the same community which is very strong and very positive that has been kind of the most amazing things about it for me personally, just the people that we have connected with all across the U.S. In some cases, beyond, that share a common bond and that care about each other it is pretty remarkable.

Gerald: They had 10 weeks of worry and unknowns while they were waiting for a diagnosis. While they got the official diagnosis Kristy was 34 weeks. After they received it, they tried to mentally prepare for her arrival. One doctor mentioned a possibility that if it wasn't a viable pregnancy Kristy could go a few states away because Kristy was so far along and have an abortion. But that wasn't an option for Kristy and Andrew. Besides there was not an absolute diagnosis at that point to know if the pregnancy was viable or not.

Kristy: I ended up being diabetic during this pregnancy, so I was already a wreck, and I couldn't eat anything that I wanted to. The plan was because of her craniosynostosis her brain and her skull may not be great for delivering naturally so we had a scheduled C-section, and she was breech for about eight weeks. However, the day of the C-section I kept having contractions, but I kept saying "oh because she is breech, she is not pushing it was a labor nurse thing turns out no, she had flipped, and I was five centimeters on the OR table, but she was going to come that day. She was C-section. Grammy got to be in the room too, two of my best friends were there too. one was taking care of me and the other was taking all of the photos. Then there was a big team of people, for her and for me. the room was filled the NICU team didn't know what to expect. The first thing to worry about is airway. So luckily, she was stubborn and couldn't get her intubated and then she didn't need to be intubated. It worked out because that would have been a longer issue to deal with.

Gerald: While in the waiting room for a doctor's appointment, someone looked at Hadley and asked Kristy "did you know she was going to be born like that?"

Kristy: Grammy and I were at therapy, and we were in the waiting room, and this is before COVID obviously and someone looked at her and said something they said, "and you knew she was going to be born like that?"

Andrew: I've been asked point blank in a therapy office by another parent about if we were given the option to abort. On a medical level it was never a focus.

Kristy: But so many other random people sitting next to you.

Andrew: Some John Smith at the doctor's office will bring it up. It is pretty invasive, and it is a snap judgement, and you look at the value of a life. They don't know my story at that point. That is a very brazen place to assert. It is pretty easy to dismiss also. The first time it might happen there is a little shock value, but definitely now or on some of the things that we share on social formats it is so easy to hit delete and move on and not waste time on that.

Kristy: I would say I have never had anyone say anything unkind to me however my husband told me, he does the Instagram I guess but he told me about some, and it is hard to believe.

Andrew: I like gallows humor; I like dark stuff but not about my child. There are boundaries there. As we have posted some things in recency that have had higher volume. It is a matter of numbers that you are going to get detractors. You are going to get people that have no connection or baring or interaction with your life and they can say off color things and that's okay, you just delete and move on.

Gerald: Andrew said he thought they would reach a level of autonomy and independence and as medical setbacks have happened; they have reached a place where Hadley requires total care. There have been two cycles of accepting: the initial diagnosis and subsequently realizing there is more going on than they had prepared for.

Andrew: We have reached a place where she is a total care child and maybe she didn't start out that way but that is who she is now and so the goal post moved a little, but that is okay. I think there have been kind of two cycles of accepting an initial diagnosis but then subsequently realizing that there is a little bit more going on than we had been set up for or prepared for and that is okay.

Gerald: Andrew has a brother and sister. Their family unit was strong, and they reacted well. His parents were out of town, and they came back when they heard Hadley's diagnosis. Andrew has some commonality with his sister. She had a child with an extreme diagnosis who passed away. Relative to Hadley she got a tracheostomy when she was 13 months old. They chose to do it. She was needing oxygen and has little airways and little ear canals. Everything in her mid-face is smaller when she got her trache it really helped her airway.

Kristy: Once she got her trache it was a big deal, that is her airway, and you can't lose your airway. We had issues with feeding in the beginning too which was really distressing because she was actually smaller than we thought she would be, and it was hard for her to gain weight. Feeding was a big issue in the beginning and oxygen became an issue as she was growing but needing a bit of oxygen. So how do you get all of this stuff to all of her appointments you're going to. With her trache you have to do a lot of training in the hospital. I'm a nurse and there is still a lot of learning for things you never thought you would do. I would say the total care stuff is mostly related to feeding which is all through her tube's and suctioning you always have to have her equipment with you. At night she is hooked up to the ventilator which it is not helping her to breathe it is just making her breathe at a certain rate.

Andrew: The first few months were kind of bliss. We were held in the NICU for 4 days and that was just to do some front-end screenings. She was breathing, breastfeeding, there was nothing to prevent her from coming home. We had about a month and a half where we had a child that looked different but was absolutely adorable. Apart from that it was very similar to our boys. Then there were syndrome specific things that started happening that kind of escalated and changed things. Her right eye started to hemorrhage out of its globe which happens to FGFR2 kids. So, we met with our craniofacial team with that and had set our first round of surgery but then eventually there was a day where her eye herniated and then it fixed in place, and we couldn't get her lid back over. That is where things escalated, and we got heavy in the medical. Just the progression from there on for about a year and a half of one discovery and need to be addressed after the other. To me the hardest thing for me is that we have a medically complex child who interacts with about 9 subspecialties or therapies and we're the hub to that and they are all spokes. We are not all in the same wheel, everything points out and comes back to us. And the lack of interdisciplinary collaboration or the fact we are non-medical, and we are driving that and what one doctor says has relevance to the next, but we are the ones left connecting the dots. That is probably the hardest thing to me in the grand scheme the care we get down. We have teams, we have wonderful support, but as far as really vetting how can we really maximize her quality of life and get her everything she needs, that is the biggest challenge.

Gerald: A typical day is Hadley is unhooked from the vent she uses to sleep at night and get ready for the day. She is immobile so moving her and all of the equipment and getting her fed, trache care, another feeding, exercise, and therapies they do. She is not going to school right now due to COVID which has given her opportunities to be social and to meet other people and allow her to grow. Most days are pleasant, nondramatic, and full of smiles. She is a happy little girl.

Kristy: I love having a girl. She is just so sweet, always so sweet, and smiley and I feel like she knows who we are and gets excited. She knows her brothers she loves her dog, it's not hard to find simple things that are just adorable about her.

Andrew: She is a happy little girl. But beyond that, she's given us so much perspective that we didn't have. You talked to our boys earlier today and they shared a little but their resilience in the whole matter and what they do is leaps and bounds more amazing because their parents are navigating and there are peaks and valleys. Peace isn't a process that you work through it is cyclical in nature. So, their resilience is amazing, I love that, but my life is markedly different than it was 5 years ago. It is better, but it is a heck of a lot harder and in ways that you would never think about unless you are given the opportunity and circumstance to navigate that.

Kristy: My life saver was connecting to another mom whose child had a similar syndrome and she just carried me through this thing. She lived in another state, and we would talk hundreds of times a day and it was like I could say whatever I wanted to say without any judgement, she didn't even know me. She just took me in and so what I would say to someone else is there is going to be someone else that will do that for you. It could be me. If I find out there is a new Pfeiffer baby coming into the world, I am going to connect with you, I'm going to pay it forward and do whatever I can do. Find your support.

Andrew: Kristy's point of connection is super important. We all need to lean on people. And beyond that you can only control things you can control. One of the things you can control is your attitude and approach through all of that. Even in markedly hard circumstances there is good. You just have to find it and you have to be looking and open to it. Our medical team would consist of an outstanding pediatrician but beyond that we work with a local neurosurgeon, neurologist, pulmonologist, GI, two eye doctors an ophthalmologist and ocular plastics

Kristy: We have a prosthetic guy too, an Ortho for her elbows that don't bend.

Andrew: Rehab

Kristy: Her heart is great. There is no cardiologist.

Andrew: So, there is about nine player's total.

Kristy: Then there is our team that is not local based. When she needs a big procedure done or we need some bigger answers we take it to the top hospital system in the U.S.

Gerald: Kristy and Andrew were asked how they make their situation more normal for their other two children. Andrew said Hadley is a family member and an essential part of the family unit all of our children have needs and deserve time. We have to peel off and have 1 on 1 time and talk. Having uncomfortable conversations is important it is an opportunity for them to express and so there has to be a regular check in if we observe a social que or an emotion building up, we say "hey bud I saw you doing this, do you want to talk about it? Could you share?" and it may not be in that moment that they will open up but at least they know that we are open to them and ultimately they will seek us out and make a physical connection if they want to share. The book "Wonder" was close to home. Andrew was very struck by the sister

and the parents in that book and the people around Augie the boys connected with the sister, watching how resilient their boys are through the grief is amazing. Grief is not something you work through it is cyclic in nature. I had the opportunity of speaking with Hadley's older brothers. Oliver and Micah.

Micah: Its different because she has a syndrome where she can't bend her elbows.
I am 4 years older than her.

Oliver: She is 5 and she likes to watch TV and she likes sleeping, she is pretty ordinary. I like to read books with her, listen to music with her and sometimes she just likes to listen to me. She laughs and smiles and sometimes she talks and sings.

Gerald: Having a sister with a disability is different he says. When asked what they think about when they are out in public, and someone asks a question or stares at her Oliver says, "we can answer questions." Oliver will tell friends about his sister if he trusts them. Micah likes to play peek-a-boo with Hadley.

Oliver: So at first without surgery she looks very different, but I realize that doesn't really matter. Everybody is different, everybody has their problems. And we all have solutions to those problems, and she does her very best. So sometimes I make food for her, I'll do errands if she needs some medical stuff I can just go into her room and grab it if my parents are occupied. I can throw things away, find bows. She likes bows to wear. She has a whole wall of them.

Micah: I sometimes pick out her outfits.

Oliver: I like how she smiles and laughs.

Gerald: I had the opportunity of speaking with Hadley's grandparents. The grandpa's reaction to finding out the diagnosis he said that if it had been their first granddaughter that had a rare syndrome or disability would have been very different. The first granddaughter only lived ten days.

Grandpa: As it were, I was there when Hadley was born, and we knew that she had this syndrome. We knew what to expect so that made it much better. We knew when she was born that we would have to have ongoing care and surgeries and such.

Gerald: Grandma's reaction was it was like finding out lightning had struck twice. She was there when Hadley was born so she knew what to expect so that made it better.

Grandma: Well, it's hard to see your children go through this. You have the grief of the child you thought you were going to have, but you also see what your children are dealing with. I think that is the hardest for me. So many major decisions and it is never just up to the doctor, it ends with them on what to do next. They haven't had a lot of guidance as far as I'm concerned.

Grandpa: You know it would be great if choices like this if there was someone in the medical world some specialty of doctors that would take them on beginning to end as kind of like the go-to person in charge because there is so much involved with directing and making decisions with someone like Hadley with all the specialists and the parents are just sitting there, they are doing all the research they can to figure out what to do next. It is a very difficult situation.

Gerald: Grandma says that she is involved in the daily care. She is honored to be involved, Hadley is a joy, and she loves taking care of her.

Grandma: All we can do is support. The hardest part is just supporting and being a cheerleader and supporting their decisions as they struggle with major life choices.

Grandpa: One of the hard things is just trying to figure out what is best for Hadley moving forward.

Grandma: And balancing the whole family, the boys. You know they say special needs well we all have special needs. We have different special needs, and the boys need to have a somewhat typical life too. Like what Andrew said we are all in a better place, we have all learned so much and those boys are going to be very enhanced in their lifetimes of what they are learning and how they react to people. We try to emphasize kindness in different things than we would have otherwise, and we look at people differently, I think. I've probably said this out loud before I was guilty of the if I saw somebody with a disabled child the thoughts go through your head "I don't know how you do it." And I would smile and then look away, I probably never talked to the child I probably didn't know the child's name I never asked the parents how they were doing. I was being polite, but I wasn't interacting. They are not an invisible community. The reason I post pictures all the time is just for awareness. I want people to see her as a person.

Gerald: Grandpa says that the last 6 months to a year Hadley has been very stable.

Grandpa: The last 6 months to a year she seems to be very stable. She really hasn't had any issues with her health. She seems to be very happy, content she is always smiling. I just sitting down next to her and having her hold my finger and to just chill out with her.

Grandma: Yeah, just to be around her and take care of her. She is easy to be around, she is a happy little girl and then watching the milestones. I think in another lifetime I should have been a physical therapist. Just trying to help her maintain. It has been hard with the pandemic of course; she is missing out and the whole disability community has missed out on their therapies and the things that they need to keep moving forward and we try to step in for some of those things for her. Because she is not getting occupation therapy, speech therapy, she's not getting physical therapy and she desperately needs all those things. So, when we can help do that in any way it's fun.

Gerald: Hadley got her name from Grandpa. Grandpa came up with the name Hadley Rose and said if you name her Hadley Rose, I'll give you $500 for each name and you can have $1,000

dollars if you gave her both names. They did and he gave them $1,000 dollars. Grandpa said that “basically we are chemistry sets.”

Grandpa: Basically what we are is chemistry sets. We are very complicated chemistry sets, we have all these different parts. We have proteins and enzymes, and all this sort of things in us and sometimes all of that doesn’t quite balance. With Hadley the Pfeiffer syndrome there is a condition where your bone structure in your head and other places doesn’t grow normally and if you have that in your head, it affects your spinal column it affects your breathing your throat your teeth your mouth your jaw, everything and your brain is up there too. So that is my understanding of it and it kind of affects a lot of things. It kind of focuses with the pressure in your brain and the bone structures of the openings and cavities.

Gerald: Grandma says that the sutures in Hadley brain made her skull fuse early. Her body was producing more bone fast so her skull was fused and so it created all of the other issues from there.

Grandma: So her skull was fused and it created all the other issues from there. So that needed to be opened. She had an emergent surgery and 4 months. It was supposed to be planned but it wasn’t because of the eye issue. In general the whole cranial community just being able to connect, I don’t’ know what people did years ago. This is so rare and like they said you get people from all over the world, it is just that rare that this community all comes together.

Gerald: Grandpa says he is lucky to have Hadley as his granddaughter. Grandma says that somedays you just have to simplify and enjoy the day.

Grandma: Somedays you just have to whittle it down and just enjoy each day. Nobody knows what your future is going to hold. You can grieve and experience joy at the same time. Just grab those moments. No-one knows what tomorrow is going to bring.

Gerald: What an honor it was to talk with Kristy and Andrew, their two sons Oliver and Micah and Grandma and Grandpa too to talk about Hadley and their journey with her.

Thanks for listening to this episode. Orange Socks is an initiative of Rise Incorporated. A nonprofit organization dedicated to supporting and advocating for people with disabilities. Follow Orange Socks on Facebook and Instagram and visit our website orangesocks.org for more stories and to find national and local resources to help parents of children with disabilities.