An Orange Socks Story: Josh and Kayla: ADNP

Interview by: Gerald Nebeker, President of Orange Socks

Welcome to the Orange Socks Podcast, where we are inspiring life, despite a diagnosis. I'm your host, Dr. Gerald Nebeker. In today's episode, I speak with Kayla and Josh about their son, Parker, who has ADNP syndrome. A rare genetic condition that causes, among other things, intellectual disability, autism spectrum disorder and distinctive facial features. I had the privilege of meeting Parker, what a beautiful and sweet little boy. I know you're going to enjoy hearing Kayla's and Josh's story.

Gerald: Alright, so Kayla and Josh, thank you so much for taking the time for an Orange Socks interview for your son, Parker, who has ADNP syndrome. Kayla, tell me about ADNP syndrome.

Kayla: ADNP syndrome is a rare, complicated, neuro-developmental condition.

Josh: You said it better than I could.

Kayla: Parker was about the 85th person in the world diagnosed with it. Now we estimate around 140. You know, you start out with global developmental delays, speech is severely affected, many of them have autism and intellectual disability. It affects, you know, various systems, a lot of the kids have heart defects, sleep apnea.

Josh: Abnormal MRIs.

Kayla: Abnormal MRIs. Some children have had brain atrophy, so it affects a lot of different things, and the children in different ways.

Josh: Basically, a slight mutation in the ADNP gene. I guess a lot of kids have different mutations of that gene, some with same and some alone, but they all kind of do some of the same things to the kids in a varying degree.

Kayla: Something that's very interesting about it, is that the autistic traits that these kids have are the same, versus there being a big spectrum. They seem to have the same type of traits, like they like adults, they warm well with adults, but not so well with other children.

Josh: All love music, all love water.

Gerald: That's interesting so.

Kayla: A happy demeanor.

Josh: They are happy kids.

Gerald: So, Josh, when did you find out that your son had this diagnosis?

Josh: Well, we went through, you know, he's our first kid. Part of it, you don't know what to expect with your first kid, you start realizing at a certain point that he's not doing things like the other kids are doing, and you know, you go to your pediatrician and he talks about the huge curve difference there are between just kids in general. So, for the longest time, you know, you want to think that there's nothing wrong. So, I want to say around 12 months, well he probably just started crawling.

Kayla: Yeah, he was army crawling, but not crawling on his hands and knees.

Josh: Yeah, so there's just global delay, really. And then, there was a year long deal of just trying to rule things out. We had a really good genetics doctor, and he could tell by looking at him, and just everything, that he probably had a genetic condition. So, he put us through to get the testing, the whole exome sequencing, which is still kind of a new deal. And from what we understand, a lot of people have a hard time getting that done as quickly as we did. So, we are actually pretty lucky to get that knocked out after our first or second visit, and our insurance covered it, and stuff like that. We were lucky to go through that process to get that diagnosis, so around a year ago, two and a half.

Gerald: Kayla, you noticed that he was missing some developmental milestones you had testing done, and finally they came up with a diagnosis. What were your thoughts when you finally found out that Parker has ADNP?

Kayla: It was really overwhelming. I didn't expect to get a diagnosis. I had read that there's maybe a 25 % chance that we would get one from the whole exome sequence thing. Just, generally, I was really surprised then for it to be something so rare was-I just remember not being able to sleep, and all I could hear all night was like ADNP syndrome, you know, just in my head over and over again. But, luckily, there's a parent who started the ADNP Kids Foundation, and a parents support group on Facebook. So, we were able to connect with other families, which has been wonderful. The parents of older children have just guided us so much, it's been a huge blessing.

Gerald: So, what's been some of the hardest things that you've had to deal with this diagnosis, whether it be care, or I don't know, what it would be. I've never met anybody with this syndrome, so I'm glad to have met him. And he's a doll, by the way, he's a cute kid. So, he's fun to meet, but what was the hardest thing about either the care, or learning about this diagnosis?

Kayla: Probably realizing that the chances of him living independently as an adult are very slim. I think, kind of coming to terms with that. Just the whole change in what I expected his life to be, and now I feel like Josh and I are coming to some peace over that. And honestly, Parker is such a joy, and we just love him so much that sometimes I don't want him to ever leave me. Then, also, the communication barrier is very difficult, but he just started saying his first word that he has said enough to where we can say he has a word. So hearing his voice has been-

Gerald: And how old is he?

Kayla: Three and a half.

Gerald: Three and a half, okay. So, Josh, what was the hardest thing for you when you finally realized you had a child with a disability?

Josh: You know, when I first found out, I kind of had in the back of my mind, I think I mentally prepared myself for the possibility a little bit better than Kayla. Not saying I thought he had something, I'm trying to be a positive person regardless of what the outcome was. So, I had already made up my mind, "Alright, if Parker's got something, we are just going to ride this out. He's awesome, that's all that matters." You know, he wasn't in my womb, so I wasn't as connected as Kayla is. And she works so hard for him every day in many ways. As time went on, I got the diagnosis, and alright, great. You know, we got a diagnosis, we move from here, we got info, you know, moving on. The hardest thing is seeing him with other kids. So, I love sports, you know, that's kind of my hobby. You, just when you have a boy, you have an idea of what you're expecting your life with your son to be. Your typical take him to the baseball game or the football game. So, you know, and that kind of just goes out the window. I mean, there's other things that can be done, but your typical picture of how your life is going to be with your son kind of got blown up pretty quickly. And it's sad, you know, when you see other children playing, and he doesn't play the same way. Little things like that, you know, the day-to-day stuff. Obviously, a little bit harder when you have a kid with disabilities, but he does things that other kids don't do that make him such a blessing and joy. Taking it one step at a time, where those bad things don't seem quite as bad, you know.

Gerald: While you're at that, why don't you tell me some of the joys that you've realized with Parker?

Josh: He's a happy kid, like you saw. Not to say he doesn't have his little tantrums, but he's got this inner peace and joy that just contagious. I mean, sometimes you'll see a stranger, even a stranger you probably don't want him to go up and talk to, and he'll just go up there and he'll give him the biggest hug, or want them to hold him or something like that. I mean, he puts a smile on everyone's face, and that's just who he is. He doesn't have to try to do it, you know, or feel like he needs to. He's just a happy, mischievous, loving and even caring, I would say. I mean, he's a snow monster, you know.

Gerald: So what are some of your joys?

Kayla: I mean, Parker, just kind of what Josh, said he brings joy to everyone he meets. I feel like he is changing all of us for the better. I feel like anyone that gets to know him, their lives are impacted by him. I even see, kind of, our group of friends, all their children are typically developing, and as they kind of learn about Parker, I can see him touch them as well.

Josh: Yeah.

Kayla: Don't you think?

Josh: Absolutely, you know, I was surprised on how many parents have children that have some sort of disability. I mean, you see them and stuff, but we were never touched in a way prior to this completely personally, with a close friend or family member. So, it was kind of all new to us, but once this has started, it's actually more prevalent than you would think. And so many people are going through similar things that we are going through, and they all say the same thing. You know, it's hard, but it is awesome. It's so worth it, and everything like that. And it's nice to show that to so many people that are close to us, realize that your child is a little different, or a lot different. That everything is good, and it teaches other kids just to be good with it too. It's a really good learning experience for everyone around us, you know, and everyone seems to be pretty receptive. I mean, they love him, you know.

Kayla: And also, you know, just like you said about the joys, I would say that whenever its been a really hard day with all the therapy, or he's had a meltdown or whatever and I'm upset about it, and Josh will always remind me, "Just forget about all that, and just go play with him." And sure enough, he gives me one of those huge hugs and-

Josh: Grin.

Kayla: And yeah.

Gerald: That's great. What has been his impact on your family, your extended family Kayla?

Kayla: I feel like he has made us, all of our extended family, more selfless.

Josh: Softer.

Kayla: Softer, yes, he has softened everyone up quite a bit, I would say. I see him really touching my dad in particular.

Gerald: Josh, what impact has he had on your extended family?

Josh: They live in a different city, so when you're kind of going through this they want to be positive through everything. Before his diagnosis, it was always just like, you know, "He's going to grow out of this." You like to be positive, but you also like to face facts when they present themselves. But, whenever it came to the point of actually getting a diagnosis, they've been great. I mean, for the most part, positive people in general, you know. But, just an outpouring of love and support, and they just want to help. And anything that they can do, they want to help, but they all love him. He loves all of them and, again, they just kind of bring everyone together. He brings the love out in this case. The love is already there, but it just exponentially grows when you have a common denominator of Parker.

Gerald: So, Kayla, what impact has Parker had on your extended family?

Kayla: I would say that Parker just brings out the best in everybody. I mean, I'm fortunate to have a wonderful family, and they're just incredibly supportive. Especially through the whole diagnostic process, and all the therapy appointments. I mean, they are just wonderful, and I feel like we're already a really close family and Parker has brought us even closer.

Gerald: So, Josh, if I were to come to you and just having received the same diagnosis looking for advice, what advice would you give me?

Josh: Enjoy it, it's just a blessing. It doesn't sound like it when you first get the news, it probably doesn't look like it, it's scary. But, man, it's a joy. It's a roller coaster. It's hard, I think I would never change anything about him, I really wouldn't. And it almost sounds cliché because you go back and you look up other things, and you read about special needs kids. And all the parents, a lot of them, say they're special, they are great. And, before I would kind of second guess that to be honest with you. "They are just saying that," in my head. But, man, it is true. I would not do anything differently. I love that kid, I really do. If there was a cure, if you gave me a cure pill and gave it to him, yeah, I would give it to him. I want my kid to do things that other kids can do, but right now, me talking to you, he's an absolute joy, and I'm completely fine with it. Hard, but a joy.

Gerald: Kayla, what advice would you give me?

Kayla: Everything that Josh said, and also get connected with parents of children with the same diagnosis. The parents of the older children, they want to help you, and they want to guide you, and they're going to give you information that's going to help your child.

Josh: Kayla is in the trenches, I get to go to work everyday, she's the one that works her butt off going to therapy visits. Two and a half hour drive just for speech, you know, and there's only so much I can do, positively. And there's only so much I can do. I mean, you can't talk to a guy the same way you talk to your girlfriends. There's only so much. It's going to be okay, and her believe me, I think. So, I think having some understanding friends which, I think, sometimes is harder said than done. I think we've been lucky to have a few. A good support group, I think, is important, don't you think? To a certain point.

Kayla: Definitely, and knowing it's hard, and that its okay to talk about it honestly.

Gerald: Well, you guys are great. Kayla, I'm going to give you the last words. Any final comments you want to make about your life with Parker, or anything that you want the world to know about your life?

Kayla: Our lives are much better because of Parker. It's difficult, but it's so worth it. I just love that boy so much, and he really does just bring joy to everyone he meets. And he is just the world to us, and we feel so fortunate that God chose us to be his parents.

Gerald: Parker is the first person I've met with ADNP syndrome. I learned a lot about it from Kayla and Josh. An early marker of it is that children with it, get all of their teeth by the time they are 1 years old. I also learned how delightful they can be, and that there is an ADNP foundation where parents can go for specific information, and to connect with other parents.

Thanks for listening to this episode. Orange Socks is an initiative of Rise Incorporated, a nonprofit organization that is 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.