An Orange Socks Story: Genie- ADNP Syndrome

Interview by: Gerald Nebeker, President of Orange Socks

Gerald: Welcome to the Orange Socks Podcast, where we are inspiring life despite a diagnosis. I am your host Dr. Gerald Nebeker, in today's episode I speak with Genie about her son, Roland, who has ADNP syndrome. I know you're going to love hearing their story.

Gerald: Genie, thank you so much for taking the time to do an Orange Socks interview with me today about your son, Roland, who is five years old who has ADNP syndrome. So, tell me when did you find out that Roland had ADNP syndrome?

Genie: We found out on august 16, 2016 that Roland actually tested positive for the ADNP pathogenic gene mutation, and it was a shock to us. My husband and I had done some genetic testing with Roland in utero, not just your normal amnio and fragile X. But we had extensive genetic testing done through Mount Sinai for every autistic type of related disorder known to man, back in 2012 when he was in utero. And ADNP was actually not part of that blood work because it was not discovered until the year that Roland was born, so the results were quite shocking to the two of us. There was not a lot known about ADNP when we were told by our genetic doctor at Weill Cornell, they had only heard of one other patient who they had diagnosed with ADNP, who has an older patient in his mid 20's. And they reached out to the family prior to our son's diagnosis and the family did not want to reach out to us, and so we had a stack of papers that explained very little about ADNP and the mutations, and his mutation in particular. But a recommendation that we reach out to Sandra Sermone, who is the head of the ADNP foundation, who at that point had about 54 children diagnosed when Roland was diagnosed. And then little was known about it, and pretty much wanting you to follow up with your neurologist, who we were very close with and they followed up with some major testing, EKG, EEG, brain scan, etcetera to make sure that Roland didn't have any other issues that we were not aware of. So, it was not an easy time in our lives to have Roland part of a blended family of five children, and it was honestly very hard in the beginning. But at some point, my husband and I, within a few days of the diagnosis in speaking with the founder of the foundation on Facebook, we were actually relieved because we actually had an answer. And we were told for so many years that it was the quote-on-quote Norman syndrome, a global set of delays, not only were we told this by numerous neurologists, endocrinologist, our eye doctor, ENT's, our whole team, we were also told this in trying to get an evaluation done through the state. So, when we found out that there is actually something significantly wrong with him, it came as a shock, but at the same point it really answered many questions that we had lingering, and it was a relief to finally find out the truth.

Gerald: So, you obviously suspected something. You had all this in utero testing, everything came back negative. So you had a healthy baby boy, and then you started to notice some things were different, or he was missing some developmental mile stones, or anyway you had some suspicions that something was amiss with Roland? Or something different than other children is that correct?

Genie: That is correct. To be honest, straight off after we brought him home from the hospital, we were having a bit of a hard time breast feeding and Roland latching on, and so I brought him back shortly after to see an ENT. And the ENT said something, you know, very offhand, something like, "I've never seen a child who had such narrow nasal passages and narrow ear canals." And had a very hard time doing an endoscopy. That kind of was a bit of a red flag, but didn't seem so alarming at that particular time. And said he had some acid reflux and we could do, A. nothing about it, B. give him a medication, or C. elevate his bed a little bit. And that was kind of the beginning of the, "Hum, what's going on?" And then I noticed that there wasn't a lot of mommy-child eye contact and eye tracking at a very young age. My husband and I noticed that he was spending a lot of time looking out the window, or if he was in the crib and there was a TV in the distance, or a light on he would spend just a lot of time turning his head directly to those and contorting his head so that he could look at the light. And I always thought that that was a little strange, but he did meet many of his milestones up till 12-15 months of age. We did have concerns about his eye contact since birth, and we kept getting that checked regularly, but his eyes started crossing. At a very young age we started bringing him to different eye doctors, ophthalmologists for opinions, and one ophthalmologist wanted to patch him and that didn't work. And they wanted to do eye drop therapy, which we tried, then he had an allergic reaction to that Atropine where you drop his healthier eye so his weaker eye can catch up, and his eye never went back to normal dilation, which they had never seen. That was a bit of a red flag, but while he was having all these issues, he was meeting some of the milestones, in fact, he was saying simple words. I've never heard "mommy" still to this day, but he was saying simple words, but he was not able to repeat them or retain them. And he was rolling over appropriately, sitting up appropriately, brought himself to standing up properly, but his walking was very, very delayed. The other thing that we noticed that kind of stood out was one of his eye lids was very droopy, and I brought him into the ophthalmologist for that and he did note that, he said, "You know, I haven't really seen that, let's get this other opinion." But with all these kind of weird things that were happening, he was kind of meeting the standards, I would say, for a state evaluation. So, when he was being evaluated, they weren't really seeing the red flags, but of course when people come into your home they are only seeing a snippet for about an hour, an hour and half, they are not seeing the whole child and the whole picture. I would say the fact that he had two eye surgeries is quite a big deal at a very young age, and I would also say the fact that he had to have both his tonsils and adenoids removal at a very young age because of what was happening. Before the diagnosis, was he wasn't sleeping at night, he had pretty good sleep, but as he was growing he wasn't able to sleep through the night and he was gasping, and I thought, "Sleep apnea." And as it turned out, children with ADNP have very low muscle tone, and when you have very

low muscle tone and you go to sleep, your whole kind of insides of your whole nasal passage tends to droop. If your adenoids are enlarged, you really have a small space for breathing so he was waking up a lot gasping for air so a lot of these, it was kind of like, as my husband likes to say, "Connecting the dots." A lot of strange things that were occurring, but we couldn't connect the dots. So that led us to want to explore more and talk more with our team at Weill Cornell and really get the bottom of it, and that's what brought us to further testing.

Gerald: So, essentially, before you got the definitive diagnosis, you were essentially chasing signs and symptoms of ADNP?

Genie: Absolutely, and I would like to mention, we noticed that his teeth were coming in rapidly, much quicker than our other children. And they were coming in very small almost like chicklet teeth. And I actually brought him in to a dentist because my husband and I really feel like we don't ever leave a stone unturned, and the dentist did note, "I haven't seen a child at this age have a full set of teeth, let alone such small ones." But nobody was every really alarmed because at that particular time in his life, he looked developmentally appropriate in terms of his age, his weight, his height with the exception of the droopy eyelid, and his eyes crossing prior to surgery. He wasn't displaying some of the things that he displays now as he's grown older.

Gerald: So, tell me what has been some of the greatest challenges that you've had so far in his five years?

Genie: Yesterday was a really challenging moment for me, I brought his sister, Devin, to a birthday party. And I bought, I went to go buy a gift at the store next to the birthday party for the little boy whose birthday it was, and I decided to buy Roland a Valentine's present. And when the woman asked what type of wrapping paper I wanted for the little boys birthday, I said, "Birthday paper, whatever you would have that would be for a celebration." And then when she asked me about wrapping Roland's gift, she said, "Well, what's your son's favorite color?" And I said, "I honestly don't know," and she said, "Why don't you know? How old is he?" And I said that he's five, and she said, "Oh, well, he doesn't have a favorite color?" And she said, "It's okay if he doesn't have a favorite color," and I said, "No, he's severely autistic, and he has a very rare neurodevelopmental syndrome called ADNP." As what it's doing to me right now, is really upsetting. So there are moments in time that are challenging in public, when you're kind of faced with talking about your day-to-day reality with your child, but at the same time, I'm extremely poignant because it makes you realize, "I'm going to learn my sons favorite color." So, when I came back home, I decided to reach out to our ABA therapist, and I want to figure out his favorite color by giving him color discrimination and having him point to his favorite color. So, certain things like that are a bit hard to cope with, then I want to find out a way to have him express to us a favorite color although he can't speak, maybe he could point to it.

Gerald: So, tell me what are some of the joys?

Genie: Lots of joys, huge amounts of joys. He is a really, really happy child. A lot of giggles, loves water play, has a hearty appetite. Although he eats completely gluten, dairy free and low sugar, and takes a ton of supplements. And so, he enjoys eating and he enjoys being part of a group. He's particularly good with adults, he was a really hard time with his sibling, who's 7, and he's a lot easier with the older siblings. But he enjoys music, he enjoys swimming, he loves the ocean. And all in all, he's a really happy kid. He loves cars, and he has very limited words, one of his favorite words is "car." So, he'll spend a lot of time outside looking at the cars, getting in and out of cars, etcetera. But he's just a joy to be around because he's not a morose child, if anything, his personality is bubbly, giggly and happy, so he's really easy to be around and really enjoyable to take to activities and to do things with. You know, it's challenging to have him in public places, but we do not hold back, and as a family we try our hardest to integrate him into society and have him part of our community.

Gerald: So, tell me what's been his impact on his siblings or your extended family, family generally, or friend's, neighbors, what has been his impact on them?

Genie: It's been an interesting journey. I was talking to another ADNP's father at, of course, our ADNP's gathering at Mount Sinai's hospital, the Seaver Autism Institute a couple of weeks ago, and the father said, "My wife and I feel like we have lost a lot of friends due to our daughter's diagnosis." I could resonate with what the father was saying, I wouldn't say that we have lost friends, I just don't think a lot of our friends understand the enormity of the syndrome and what we face on a day to day basis. A huge challenge that Roland has had, and its surely getting better, has been sleep. And that has taken a huge toll on me and his siblings because we are all on the same floor. Recently, they've gotten better because we have a special needs bed for him which he adores, but it's hard in general to take him to restaurants. We no longer take him to restaurants. We don't take him to places that are very loud. Our social life we've kind of had to change and alter a bit just because we had to think ahead of how things will impact him and factor in his nap, etcetera. He's quite good with his half-siblings, the twins that are sixteen and his half-brother, who's 14, and maybe it's because they look more like adults to him. And many ADNP's kids respond better to adult figures than they do to children around their own age. The largest impact that he has had is on his seven-year-old sister, and it's been very, very hard for her to have a special needs brother. She is extremely compassionate, but she's also a magnet for hair-pulling, and Roland spends a lot of time trying to pull her hair, pinch her, etcetera. So, we have really worked really hard with a ABCA specialist with both children in particular to help our daughter to tolerate the hair pulling and not fight back in order to not give in to the attention that he is trying to get while he's pulling hair, or trying to act in an aggressive manner. So, it is a huge challenge for her, and I think deep down inside she is an extremely compassionate loving sister, but for a seven-year-old it's quite hard.

Gerald: Sure, so if I were to come to you just having received a diagnosis that my child had ADNP syndrome, what advice would you give me?

Genie: I love that question. Best advice that I could give anybody when they get a diagnosis, is that it will get better after you hear the initial shock that your child has a rare syndrome. That at this point in time, we have about 160 diagnosed children around the world with this syndrome, that is very scary. There's also the fear that a lot of ADNP's parents have that I do not have, and that is what's the life expectancy? I don't look at his syndrome in terms of what's his life expectancy, I look at his syndrome as we are living in the here and the now, and I'm never going to give up hope. Although he's nonverbal, I know in my heart that he will speak. When we got this diagnosis and we reached out to other ADNP's parents, they had different opinions because perhaps their children were manifesting different forms of the mutation. And so, it can be harder for some people than it is for others because maybe their child can talk, but in calipers and can't walk, etcetera. Every child with a mutation has a different form, so although many of these kids across the board, they all have teeth that came in early, they all love water play, they all do not know the difference between hot and cold, they all have certain things that are similar. They really, really vary, so I do tell parents when they reach out to me, "Live in the moment, have belief and advocate for your child. You are the only person that can advocate for your child, if your child is nonverbal." When my husband and I started going through the state for an IEP, that's when we really looked at each other and said we need to do something more than what we are doing for Roland we need to start a foundation. And we through a board member at the Mount Sinai hospital where Roland and Charlotte and Taylor were born, we reached out to the Seaver Autism Institute, who is doing a study on sealempidermic syndrome which is an autistic related disorder as well as Fauxstone1. And we reached out to them, and had Roland undergo a fully neuropsychiatric evaluation there. And met with them and started a foundation, and brought Sandra Sermone in who is the head of the ADNP's foundation I'm now the P of the ADNP's foundation under Sandra. And Sandra is supporting our endeavor, and our endeavor really is to raise 200,000 dollars to have 10 patients fully evaluated at the Seaver Autism Institute, in hopes of a cure. Perhaps a clinical trial, that they are looking into with the founder, of which this is public the founder of the ADNP, Dr. Alana Doges, is looking into a drug, an intranasal spray that perhaps could help children with ADNP. And Mount Sinai is certainly looking into stem cell research for autistic related disorders, and so that is part of our mission is to not give up hope and to allow our son to be a part of the study. And my husband and I were a part of the study as well that involved huge amounts of blood sampling, MRI's, EKG's, EEG's, etcetera. As we have a strong belief that down the road there will be some sort of cure for children with this syndrome.

Gerald: Good for you, that's great.

Genie: I have a wonderful story, and I wanted to go back to the sibling conversation because before we got the diagnosis and Roland was nonverbal, we were having a family get together and I'd been so upset that he had a couple of words and then he

had lost the word. And we were having a family get together, and we said on a night, "Well, everybody, let's predict when Roland is going to speak," and my father was throwing out, "Thanksgiving!" My mom said, "Christmas!" I said, "My birthday!" His siblings were saying all different dates, my husband said a date, our caregivers were saying dates, so here we had a list and we were all guesstimating when Roland was going to speak, and his seven-year-old sister, who obviously was a couple of years younger at that point, said, "On starfish day." And we all looked at her and said, "Huh? What's starfish day?" And she said, "When he's ready." And so, I think that to me shows that she knew innately as his sister, and as growing up with him, that something was wrong. And it was her way of explaining to all of us that, "Calm down, he's going to speak when he is ready." And I think about that little story probably every day.

Gerald: That's a great story.

Genie: So, the way that we live our life is starfish day, any little milestone that he makes, any little word. Recently, he said the word, "kale chip" when he was eating a kale chip, and then we haven't heard it since. That is enough to celebrate starfish day because he said the word "kale chip." Or if he can climb up a ladder to get to the top of the slide, that's another starfish day moment. So, I would advise all parents who have kids with really rare syndromes that are really challenging every day to wake up and decide to celebrate their child's starfish day.

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