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 and has ADNP syndrome. When did you find out that Roland had ADNP syndrome?

Genie: We found out on August 16, 2016, that Roland tested positive for the ADNP pathogenic gene mutation. It was a shock to us. My husband and I had done some genetic testing with Roland in utero, not just the normal amnio and fragile X, but we had extensive genetic testing done through Mount Sinai for every autistic type of related disorder in 2012 when he was in utero. ADNP was not part of that blood work because it was not discovered until the year that Roland was born. The diagnosis was 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 was an older patient in his mid 20's. They reached out to the family prior to our son's diagnosis, but the family did not want to reach out to us. We only had a stack of papers that explained very little about ADNP, the mutations and his mutation in particular. Then we got a recommendation to reach out to Sandra Sermone, the head of the ADNP Foundation, who at that point had about 54 children diagnosed when Roland was diagnosed. Little was known about it, and we were advised to follow up with our neurologist, who we were very close with. They conducted major testing, EKG, EEG, and brain scan to make sure that Roland didn't have any other issues. That was not an easy time in our lives, having Roland as part of a blended family of five children. It was honestly very hard in the beginning, but within a few days of the diagnosis, in speaking with the founder of the foundation on Facebook, my husband and I were relieved because we had an answer. We were told for so many years that it was the "Norman syndrome," a global set of delays. We were told this by numerous neurologists, an endocrinologist, our eye doctor, ENTs, and our whole team. We were also told this in trying to get an evaluation done through the state. When we found out that there was actually something significantly wrong with him, it came as a shock, but it also answered many questions, and it was a relief to finally find out the truth.

Gerald: You obviously suspected something. You had all the in-utero testing, and everything came back negative, so you had a healthy baby boy. Then you started to notice some things were different or he was missing some developmental milestones; you had some suspicions that something was amiss with Roland or something was different from other children, is that correct?

Genie: That is correct. To be honest, after we brought him home from the hospital, we were having a bit of a hard time breast-feeding and Roland latching on, so I brought him back shortly after to see an ENT. The ENT said very off-hand that he had never seen a child who had such narrow nasal passages and narrow ear canals, and he had a very hard time doing an endoscopy. That was a bit of a red flag, but it didn't seem too alarming at that particular time. He said Roland had some reflux, and we could do (A) nothing about it, (B) give him medication, or (C) elevate his bed a little bit. That was the beginning of what's going on. 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 a lot of time turning his head directly to those and contorting his head so that he could look at the light. I always thought that was a little strange, but he did meet many of his milestones up until12-15 months of age. We had concerns about his eye contact since birth, and we kept getting that checked regularly. His eyes started crossing at a very young age, so we started taking him to different ophthalmologists for opinions. One ophthalmologist wanted to patch him and that didn't work, then they wanted to do eye-drop therapy, which we tried, but he had an allergic reaction to the Atropine which you drop into his healthier eye so his weaker eye can catch up. His eye never went back to normal dilation, and they had never seen that happen, so that was a red flag. 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. He was not able to repeat them or retain them, but he was rolling over appropriately, sitting up appropriately and standing up properly. Walking was very, very delayed. The other thing that we noticed was that one of his eyelids was very droopy, and I took him into the ophthalmologist for that. The doctor said that he hadn't really seen that and wanted another opinion. Even with all these weird things that were happening, Roland was meeting the standards for a state evaluation. When he was being evaluated, they weren't really seeing the red flags, but when people come into your home, they are only seeing a snippet for about an hour to an hour and half; they are not seeing the whole child and the whole picture. The fact that he had two eye surgeries is quite a big deal at such a young age, and the fact that he had to have his tonsils and adenoids removed at a very young age. Before the diagnosis, he wasn't sleeping at night. As he was growing, he wasn't able to sleep through the night; he was gasping, and I thought it was sleep apnea. As it turned out, children with ADNP have very low muscle tone, so when you go to sleep, the inside of your nasal passages tend to droop. If your adenoids are enlarged, you have a small space for breathing, so he was waking up a lot, gasping for air. A lot of these it was kind of like as my husband likes to say connecting the dots a lot of strange things were occurring, but we couldn't connect the dots. That led us to want to explore more, talk more with our team at Weill Cornell and really get to the bottom of it, and that's what brought us to further testing.

Gerald: Before you got the definitive diagnosis, you were essentially chasing the signs and symptoms of ADNP.

Genie: Absolutely, and I would like to mention that we noticed that his teeth were coming in much quicker than our other children, and they were coming in very small, almost like Chiclet teeth. I took him to a dentist because my husband and I don't ever leave a stone unturned, and the dentist did say that he hadn't seen a child at that age have a full set of teeth, let alone such small ones, but nobody was every really alarmed. At that particular time in his life, he looked developmentally appropriate in terms of his age, his weight and his height with the exception of the droopy eyelid and his crossed eyes. Prior to surgery, he wasn't displaying some of the things that he displays now as he's grown older.

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

Gerald: What are some of the joys?

Genie: Lots of joys, huge amounts of joys. He is a really, really happy child with a lot of giggles. He loves water play and has a hearty appetite although he eats completely gluten- and dairy-free and low-sugar, and he takes a ton of supplements. He enjoys eating, and he enjoys being part of a group. He's particularly good with adults. He has a really hard time with his sibling who is seven, but he's a lot better with the older siblings. He enjoys music, he enjoys swimming and he loves the ocean. All in all, he's a really happy kid. He loves cars; he has very limited words, but one of his favorite words is car, so he'll spend a lot of time outside looking at the cars, and getting in and out of cars. He's just a joy to be around. 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. 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 be part of our community.

Gerald: What has been his impact on his siblings or your extended family, family generally or friend's and neighbors?

Genie: It has been an interesting journey. I was talking to another ADNP's father at an ADNP's gathering at Mount Sinai's hospital at 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 empathize 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 that what we face on a day-to-day basis. A huge challenge that Roland has had and that is 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 that has gotten better because we have a special-needs bed for him, which he adores. In general, it's hard to take him to restaurants. We no longer take him to restaurants or to places that are very loud. We've had to change and alter our social life a bit just because we have to think ahead of how things will impact him and factor in his nap. He's quite good with his half-siblings, the twins who are 16 and his half-brother who is 14. Maybe it's because they look more like adults to him, and many ADNP 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. It has been 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 or pinch her. We have worked really hard with an ABCA specialist with both children 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 acting in an aggressive manner. 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: 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. There's the initial shock of hearing that your child has a rare syndrome—there are about 160 diagnosed children around the world with this syndrome. and that is very scary. There's also the fear that a lot of ADNP parents have that I do not have and that is about the life expectancy. I don't look at his syndrome in terms of his life expectancy; I look at it as we are living in the here-and–now, and I'm never going to give up hope. Although he's non-verbal, I know in my heart that he will speak. When we got this diagnosis and reached out to other ADNP parents, they had different opinions, because perhaps their children were

manifesting different forms of the mutation. It can be harder for some people than it is for others, because maybe their child can talk but in calipers and cant watch etcetera every child with the mutation takes different forms. Across the board, these kids all have teeth that came in early, they all love water play, they do not know the difference between hot and cold and they all have certain things that are similar, but they also really vary. When parents reach out to me, I tell them to live in the moment, have belief and advocate for your child. You are the only person who can advocate for your child if your child is non-verbal. When my husband and I started going through the state for an IEP, that's when we really looked at each other and said that we needed to do something more than what we are doing for Roland. We needed to start a foundation, and we threw a board member at the Mount Sinai hospital where Roland was born. We reached out to the Seaver Autism Institute and had Roland undergo a full psychiatric evaluation there. We met with them and started a foundation, and we brought in Sandra, the head of the ADNP Foundation. I'm now the President of the ADNP Foundation under Sandra who is supporting our endeavor, which is to raise 200,00 dollars for 10 patients to be fully evaluated at the Seaver Autism Institute in hopes of finding a cure or perhaps funding a clinical trial. Dr. Alana Doges, the founder of the ADNP Foundation, is looking into an intranasal spray drug that perhaps could help children with ADNP, and Mount Sinai is looking into stem-cell research for autistic-related disorders. Part of our mission is to not give up hope and to allow our son to be a part of the study. My husband and I were part of the study as well, and that involved huge amounts of blood sampling, MRIs, EKGs and EEGs. 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 want to go back to the sibling conversation, because before we got the diagnosis and Roland was non-verbal, we were having a family get-together. I'd been so upset that he had a couple of words and then lost the words. During the family get-together, one night we asked everyone to predict when Roland is going to speak. My father was saying Thanksgiving, my mom said Christmas, I said my birthday, his siblings were saying all different dates, my husband said a date, and our caregivers were saying dates. Here we had a list, and we were all guesstimating when Roland was going to speak. His seven-year-old sister, who obviously was a couple of years younger at that point, said, "On Star Fish Day." We all looked at her and asked, "What's Star Fish Day?" She said, "When he's ready." That shows that she knew innately as his sister and as growing up with him that something was wrong, and it was her way of telling all of us to calm down, he's going to speak when he is ready. I think about that little story probably every day.

Gerald: That's a great story.

Genie: The way that we live our life is Star Fish Day, any little milestone that he makes, any little word that he says. Recently he said Kale Chip when he was eating a kale chip, and we haven't heard it since. That is enough to celebrate Star Fish Day

because he said the word kale chip. If he can climb up a ladder to get to the top of the slide, that's another Star Fish Day moment. I would advise all parents with kids who have really rare syndromes who are really challenged every day to wake up and decide to celebrate their child's Star Fish Day.

Gerald: Thanks for listening to this episode. Orange Socks is an initiative of Rise Incorporated, a non-profit 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.