An Orange Socks Story – Nancy and Daniel: PPP2R5D

Interviewed by: Gerald Nebeker, President of Orange Socks

Gerald: I was thrilled to meet with Daniel and Nancy about their daughter Avianna, who at three years old was diagnosed with a condition so rare there are only 28 in the world with it. In fact, it doesn't really even have a name yet and is referred to only as PPP2R5D.

Gerald: Nancy, when did you find out that your daughter had issues?

Nancy: When she was probably a couple of months old and she wasn't able to do the typical milestones for her age.

Gerald: And you had gone to doctors…

Nancy: Yes, we went to her primary who suggested starting physical therapy, and then we just noticed more and more milestones that weren't able to be reached. We started getting referrals for a geneticist or a neurologist, I think, too, and that is when the guessing game began.

Gerald: When did they actually come up with a diagnosis?

Nancy: It would be three years ago in the summer.

Gerald: So it took them three years.

Nancy: Yeah.

Gerald; What is that diagnosis?

Nancy: it's a variation of PPP2R5D.

Gerald: How many people in the world have this same diagnosis?

Nancy: Around the world, there are 28 or so families that have a loved one with this diagnosis that we know of.

Gerald: A very rare condition.

Nancy: Yes.

Gerald: Daniel, what were your thoughts when you finally got a diagnosis three years in, and you had a child with PPP2R5D?

Daniel: At first, it took a while. I probably spent a couple of weeks thinking about it, because being a a first-time parent, you hear about families or parents who have child with disabilities, but you never think it could be you. It's just something that you had to accept, and there was nothing that we could do other than love her and try to get her the help that that she needs. Mom does a great job of that, and it was tough for sure.

Gerald: So what types of things do you need to do with her?

Nancy: Well, right now, it's a full-time schedule of therapies. Her biggest thing is her speech, so she does two hours of speech a week in school and out of school, and then she has physical therapy because of her low muscle tone. We go to Utah every six months when before it was every three months. We would fly to Utah for her severe scoliosis so she can get treated at the Hospital there for a bunch of therapies. We are also looking on the computer for other ideas and to see if another report comes up about her mutation because we don't know anything. She is what's giving all the doctors the information for it.

Gerald: Right, so Daniel, what are some of the hard things and some of the challenges you've experienced in having a daughter with a disability?

Daniel: She is six now, and I I just had a conversation with another parent a couple of days ago about this. In the beginning, it was tough, especially more with her brother because he's able to do things that she isn't. He is so much further advanced that you could see her struggles as time passed. I almost got used to it, and I almost felt as if nothing was wrong, that it was just her. I think really things set in when she is around other kids, not so much with her brother anymore, but when she is with a group of kids playing and they are all able to run around, jump up and down, and go up and down the slide. We have conversations with their parents, and it definitely is tough to think about the tough decisions.

Gerald: Let me ask you the same question: what have been some of the hard parts for you? Obviously there are all the therapies and all sorts of things, but what else has been tough for you?

Nancy: Not being able to have a conversation with her, like how was your day at school, and her not being able to respond like, "Oh, I did this or this." She is not able to do that kind of thing and like he said, she can't do a lot of the stuff that other kids can do, like go to the slide or race with somebody. There are a lot of times where we are in a group of people and the kids are playing with each other, and she is just doing her own thing, standing back and playing by herself, playing with other stuff not around all the other kids. I think that's the hardest thing and thinking about how is her future going to be.

Gerald: Sure.

Daniel: to give you a good picture of maybe what she is describing is when she started school, t was almost like a game of charades, where she would do signs and grunts and groans, and you'd have to figure out what she wanted. That has slowly turned into one word or two words, and now she is able to put a few more words together. Because we are around her every day, we can probably understand her where friends or others wouldn't be able to pick up what she is trying to say. It's a lot better now, but in the beginning, it was really tough because she was severely delayed in several categories. The biggest thing was speech; that was tough, and that was always something in the back of my mind as friends with kids even a year or years younger could speak full sentences. If they needed something, they could talk to their kid, and it's tough not being able to do the same.

Gerald: A little easier question, perhaps: Nancy, what are the joys?

Nancy: She sees everything in a completely other light. She is so happy. In a bad situation or a sad situation, she just sees the best outcome. She fights, and she never gives up. She is always trying her best, and she's just so caring and has such a beautiful soul.

Gerald: What are your joys?

Daniel: She's a fighter. A friend probably said it best: there are problems around the world, and we complain about the smallest things, and yet even with everything that she has to go through, she still wakes up every day with a smile on her face as if everything is okay, and I love her for just that alone.

Gerald: What has been her impact on your family? She is the oldest of the children that you have, so what has been that impact as well as the impact on your extended families or even neighbors who she interacts with?

Daniel: I I got this feeling the day that she was born. When I talked to family members or to friends who were parents, they would always say, "Everything is going to turn out okay." This went on for years, and I saw that things weren't okay, and to this day, I don't think everybody understands. They spend maybe a couple of hours with her, where when she is with us by herself, she acts totally differently. She keeps more to herself when she is around families or friends, so I think they are not able to see entirely what what we go through or what we deal with on a daily basis.

Gerald: What are your thoughts about the impact of of her life on others who she associates with?

Nancy: The same thing of never giving up on things. They see that all the things she's been through, and she still does everything with a smile on her face and continues to fight to learn. Everything takes her so much more work, even to walk across the street, but she is still going, and I think for our family, we do not give up and keep going and appreciate the small simple things in life.

Gerald: Knowing what you know now, if I came to you with either a young child or in utero with this same diagnosis, what advice would you give me?

Nancy: Don't give up fighting for your child. Be the biggest advocate, because if that wasn't the case for us, I don't know where she would be right now and if she would even be able to walk and talk. Just fight for them and be their biggest advocate. When we were diagnosed, the doctor basically told us, "This is what she has. I can't give you any information. We have nothing to give you." It was just a piece of paper with different words and letters, and I had no idea what they meant. When they told us there were only about seven families in the world that they knew of with this diagnosis, I remember coming home and searching online and not finding anything. I reached out to different doctors and other research centers, asking for anything, and nothing was there. Then we posted this big thing on social media, and we found a couple of families. Then we started a Facebook page, and now we have an organization that is going to help fund research for this mutation. If it weren't for the parents fighting for their kids, I don't think we would have any of the things that we do.

Gerald: Oh, that is great. Was it worth it?

Nancy: 100%. She might not see any of the outcome of all the research, but she is going to help to change the world and help others out there.

Gerald: Wonderful. Thank you, guys. Thank you for taking the time to meet with me.

Nancy: Thank you.