An Orange Socks Story: Cora and Tray- Rett 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 this episode, I speak with Cora and Tray about their twin daughters Charlie and Olivia, who have Rett syndrome. According to rettsyndrome.org, Rett syndrome is a rare, non-inherited, genetic, post-natal neurological disorder that occurs almost exclusively in girls and leads to severe impairment affecting nearly every aspect of the child's life: the ability to speak, walk, eat and even breathe easily. Cora and Tray's situation is unique, with only a handful of people in North America in similar circumstances.

Gerald: Tray and Cora, thank you so much for taking the time this evening to speak with me about your identical twin daughters Charlie and Olivia, who have Rett syndrome. Cora, when did you find out that your daughters had Rett syndrome?

Cora: We found out that our daughters had Rett syndrome on September 15th of 2017. That day was rough. We had been waiting for genetic testing results from our developmental pediatrician for about six weeks prior to that. Earlier in 2017, the girls were diagnosed with a global developmental delay, and they started receiving therapies through early intervention. Not much progress was being made, and we actually asked to be referred to a specialist because we knew that something wasn't quite right. They weren't hitting their milestones when they should have, so we needed to take the next step. On August 7th of 2017, we went to Vanderbilt to see a developmental pediatrician. After about an hour of being asked all kinds of questions and the girls were starting to melt down in the office, she actually diagnosed them with Autism and then immediately said she also wanted to do genetic testing for a rare neurological disorder called Rett syndrome. We had obviously never heard of it. It was extremely rare, and she said, "Don't Google it; it's very rare and they probably don't have it, but we just need to make sure." We drove home and Googled it immediately. I think most parents do that. We waited six weeks to get the actual phone call that they both had the genetic mutation, and it was Rett syndrome.

Gerald: How old were they at that time?

Cora: They had just turned two in May, so they were about 27 months.

Gerald: You knew something was up, and you had some testing done. Tray, what were your thoughts when you got a definitive diagnosis?

Tray: It's kind of an interesting question, because we talked about this in the months leading up to the initial "Hey, let's check this out and see if it is even though it's really rare." The more we looked into it during that 4-6 week period, we came to the

realization that it was probably what it is. I think a lot of our grieving was done in that time-frame, but then when we actually got the diagnosis, it was almost a sense of relief, because if it hadn't been that diagnosis, then we would have had to start the journey over again to figure out what it was. That gave us a game plan, and we knew what we needed to do and what resources we needed to approach and all of that. It was a mixed bag of emotion. Obviously, it's never easy to hear that, but I think it was made a little bit easier because we had prepared ourselves.

Gerald: How many identical twins have Rett syndrome in the United States? Tray, I think you mentioned when we were talking before we started, how many did we figure there are in the United States?

Tray: In North America, not including Canada, they're the only case that we know of that are their age.

Gerald: Wow.

Cora: Last time we checked, there were eight confirmed cases.

Gerald: Cora, you were mentioning that this effect is more common in girls than boys?

Cora: It is, yes. It's a chromosomal mutation in the MECP2 Protein on the X gene, so it's predominantly in girls, but very rarely it's also seen in boys. It tends to be more severe in boys because they only have the one copy of the gene; it's mainly girls.

Gerald: Cora, what's been the hardest thing for you so far in their young lives now that they're two years old?

Cora: They will be three in a few months. Initially, the hardest thing was that time period of waiting and knowing something's not right but not knowing what it is; that was really difficult. Like Tray said, when we actually got the diagnosis, it did feel like a sense of relief. As far as the hardest thing now, the biggest thing that we're dealing with currently and probably will until there's a cure for Rett syndrome is the communication aspect. They are non-verbal. The MECP 2 gene affects lots of other things in the body; it controls motor function and has aspects to communication and even breathing, so the level of severity is wide. We were told our girls have a milder form of it, but there are girls and boys who are completely on the other end where they have daily seizures, cannot use their hands at all and grind their teeth constantly. There's just a list of things that could be very severe for us. The girls don't have seizures, but they do have some breathing abnormalities and hand stereotypy. The hardest thing is communication as they are non-verbal. That's the hardest aspect of every day with their not being able to communicate their needs to us, and it causes them anxiety. Just imagine being hungry or thirsty or wanting something yet there's no way for you to express that, and you get frustrated because all the cognitive abilities are there, but the wires aren't connecting to make that

happen. That's frustrating for them; we can see that. We decided to make that our main area of focus, so we do a lot of speech therapy, and we attend a lot of educational webinars and seminars about how to make this easier for everybody in our family. We actually just got approved for our second Tobii gaze device which helps them to communicate by tracking their eye gaze for different symbols. They are catching on fast, so it's becoming one of the more joyful things instead of a hard thing, so we are turning it into a positive.

Gerald: Well, that's a good segue into my next question. What have been the joys?

Cora: Oh, gosh, it's easier now that we are past the fog of a brand-new diagnosis. I feel like it's more joy throughout the day than hard stuff, which is great, because it didn't feel like that in the very beginning. We get to celebrate the little milestones. With Rett syndrome, usually you don't show any symptoms until around 12-18 months, and then you go through a period of regression where you rapidly start to lose these skills and lose any words you once said. Our girls used to say a few words, but stopped. and the hand movements and things like that started. Now that's leveling out a little bit. The girls are getting better at taking steps up the stairs, and they are actually improving in areas that we were told they wouldn't improve much at all, so that's exciting for us. They are really funny girls; our girls are hilarious. They understand humor, and they laugh when it's appropriate to laugh, so they are really witty, hilarious girls, and I think that's just a gift that keeps on giving. Honey, what do you think the joys are?

Tray: There are a number of specific things that we noticed or mentioned like walking up the stairs, improving their overall motor skills and the physical capabilities that they have. They are actually growing. To give you a specific example, Charlie and Olivia both have these balance bikes, and they ride them around the house all around our living room and island. We took Charlie from going just a couple of moves forward and a couple of moves back with no real direction where she would just bump in and bounce to a month or two later where she literally will navigate this thing and take it all the way through our bedroom into our bathroom doing three-point turns and keeping on cruising. Those are things that you don't really see when it comes to their diagnosis. Things like that are fun and just really exciting for us, where with a typical two-and-a-half to three-year-old, you'd think it was no big deal. For us, it's a huge step in the right direction, so we get to celebrate all of that stuff.

Gerald: What has been their impact on your family?

Tray: I would say overall it has broadened their horizon and more or less brought everybody closer together. I feel like everybody wants to help out; they want to learn what they can do, how they can do it and how they can better support the girls. It's not only about doing things for them. They can know what Cora and I are going to be doing ona day-to-day basis, and I feel that they've been a really strong support system for our family as a whole, so we really couldn't ask for more.

Gerald: Cora, do you have anything to add to that?

Cora: I think it has been a shock, and it still is for some family members, but they've put that feeling aside and focused on the positive. We are doing that as a family. Everybody's been extremely supportive, and the girls have a lot of grandparents so that helps. We are very lucky that they all live locally. Having that family support is important, and we may have been in a totally different place if we didn't have them here.

Gerald: That's wonderful. Cora, if I were to come to you just having found out that I had a child with Rett syndrome, what advice would you give me?

Cora: I would say to reach out to your community. There are lots of families out there and people in similar situations, and they have more experience in this game, especially with these rare diseases. There are a lot of neurological diseases and a lot of aspects and different areas that go into it. Another piece of advice would be to manage your time. You have the communication side of things and the medical side, which can be a very large aspect by itself; you also have education and social interaction. There are a lot of different areas, so don't spread yourself too thin; focus on one area at a time, find the proper resources and accept help when it's offered to you. Tray and I are very type A, go-get-em, we we-can-handle-everything-in-the-world type people, and we quickly found out that that's just a silly idea because you can't do everything. What you can do is take everything in small chunks. Love your children and focus on that first, and everything will fall into place if you just have patience with yourself. You're so much stronger than you think you are. You don't have to do it all on your own; that's why there are resources and communities out there like Orange Socks to help parents understand that this doesn't have to be so lonely, it can actually be a really positive experience for you.

Gerald: That's awesome. Any closing words that you want to say?

Tray: We've talked about this before, and Cora's done a fantastic job with everything. She has just taken the reins on most of what pertains to Rett syndrome, and what I would say as a closing remark would be become ad advocate. Don't be scared or shy. Awareness is key when it comes to a diagnosis, especially a rare one, because more resources become available when more people know about your condition.

Gerald: Okay, awesome.

Cora: Yeah, I agree.

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