An Orange Socks Story - Tiffany and Nathan: Aicardi and Turner’s syndrome

Interview by: Gerald Nebeker, President of Orange Socks

It was an honor for me to meet with Tiffany and Nathan to discuss their daughter, Ellie, who has been diagnosed with Aicardi syndrome and Turner syndrome. They were given the option to abort Ellie when the issues were detected at their 20-week check-up which, thankfully, they did not do. I was happy to meet now three-year-old Ellie, who you will hear periodically during our conversation. She has surpassed doctor-predicted limitations and is delightful.

Gerald: When did you find that Ellie had some issues?

Tiffany: For her 20-week ultrasound, we went in typically excited to find out the gender. I didn’t think anything crazy. They said she had some abnormalities with her stomach, I think, and that those were initial markers for Down syndrome. They sent me to Perinatology of Maternal Fetal Medicine at our local hospital, and we began our biweekly visits. It seems like at every visit, they noticed something more.

Nathan: Something different.

Tiffany: They started measuring the things they noticed the most. Initially her femurs were really short, and they kept that as a marker for Down syndrome. Every time we went back, we kind of just got a little more.

Nathan: You could tell that when they just got a little more, but they didn’t want to say it was Down syndrome. They would say things like, “We see this, but we don’t see this, and this is kind of different than what we normally see,” and you could tell they were just trying to find anything that they could. It wasn’t until about the third or fourth or maybe the fifth ultrasound that they finally said, “We are noticing some different things in her in the formation of her brain,” and that was when we were more like, “Okay, now what is it?” They said, “She is missing her corpus callosum,” which is the separator between the right and left hemispheres, and that is not typical with Downs, and they were not quite sure what this was. They just kept monitoring stuff, and when we really found out, it was really more like, “What do we have?”

Tiffany: What are we facing?

Gerald: When did you get a definitive diagnosis?

Nathan: An hour before we delivered.

Gerald: Really.

Nathan: Yes, we were sitting in the delivery room where she had been in labor, and we had been sitting there for maybe a couple of hours.

Tiffany: It was later in the afternoon when one of the NICU perinatologist came in.

Nathan: Running in.

Tiffany: That kind of scared us.

Nathan: He said, “I need a marker or pen or something,” and then he started drawing on this white board and explaining what he saw, and then finally he wrote the word Aicardi on the board, and he said, “This is, I think, what she’s got.”

Gerald: So, you were in labor.

Tiffany: Yeah, and it was a traumatic labor.

Gerald: He was running in all excited to tell us that he knows what…

Nathan: …That he has a diagnosis.

Gerald: That's funny.

Nathan: It was kind of crazy, and immediately we asked, "What's that?"

Tiffany: Family members were sitting in there with us.

Nathan: We've got medical people in our family, and we were asking, and nobody knew what it was. Everybody was just kind of explaining really quickly the different symptoms and the different prognosis. He was trying to give us as much information as probably what he had read an hour before in some medical journal, he was trying to regurgitate what he had just found. Then obviously the labor got a little more intense, and that kind of got pushed to the side. They rushed her in to do a c-section, and that changed everything for a minute because we were worried first about making sure we got her out of there healthy, and then we could deal with everything else.

Gerald: What kind of advice did they give you at your 20 weeks? They were thinking maybe you have a Down syndrome child, did any of them offer any advice at that point?

Tiffany: Well, I remember their offering us an amnio, and they wanted to do further testing and MRIs while I was pregnant.

Nathan: We knew we would have some challenges, so spending a bunch of time and money and effort …

Tiffany: …for what you know would stress ourselves out more wasn't worth it.

Nathan: We thought that we were going to have some challenges, and we were just going to deal with it. I know they tried to get us into counseling. They said, "Why don't you go talk to this lady." We talked with her for 10-15 minutes, and she asked if wanted to come back and talk some more, and we said, "No, that we were fine." She said, "Really?" We said, "Yes, we will figure this out. We'll figure out a way to make this work." We got the bill from her, and it was $600, and we decided we didn't need to see her any more for sure.

Gerald: That's funny.

Nathan: There weren't really a lot of specific instructions because I don't think they really knew how to approach it.

Gerald: She also has another diagnosis, right?

Nathan: Yes.

Tiffany: Yes.

Nathan: That wasn't realized until…

Tiffany: …three days…

Nathan: …after she was born.

Tiffany: Yeah.

Tiffany: After I delivered, must have been on all the drugs and stuff, but I kept saying that they needed to sample her umbilical cord for testing. I didn't know why, but they needed to do genetic testing. They did the testing and sent it in, and two or three days later we went in to visit her in the NICU, and one member of the neonatal team came in and said that they had found another diagnosis. They said that she had Turner syndrome, mosaic Turner.

Gerald: Really.

Nathan: Turner syndrome deals with the sex chromosomes, so you have the XX or XY, and they said she's X. They were not seeing anything on that other chromosome, and they wanted to do a further analysis which would take a couple of weeks. They did even further testing, and it came back that she was very mosaic, like 1 in 200 of chromosomes have the XX whereas everything else is X, so that was kind of an after-the-fact. Turner comes with a lot less grim of a diagnosis than Aicardi. Aicardi tends to have a lot more troubling things like heart issues, back issues and lots of developmental issues, so we thought that Turner was okay. She is going to be a little short and a little bit small, and will develop a little bit slower, but that's fine as we'll deal with that later. Let's make sure she can get through the other issues. We were rushing to get CT scans and blood work, and they were doing all these tests trying to figure out if her heart was okay.

Tiffany: Her digestive system…

Nathan: They did a bunch of tests on her digestive system, and that was the main focus of the Aicardi. The Turner syndrome didn't present an immediate concern, and so we started going down a list of all of the scary diagnoses that come with Aicardi. We started checking them off, "No, she doesn't have that, she doesn't have that, and she doesn't have that." That's when we said, "Okay, let's talk about the Turner and see what we are going to do deal with that." Our focus was turned more to the Turner because a lot of the Aicardi stuff is just it is what it is. We now just have to deal with the therapy to try and help with growth and different things like that.

Gerald: What have been some of the challenges with having a child with these two conditions and these two diagnoses?

Tiffany: Well, delayed milestones.

Gerald: Sure.

Tiffany: Our biggest challenge was that we didn't realize her feeding was really hard when we first brought her home. That was actually the reason it took a while to get her out of the NICU. We interviewed a surgical team to have a G-tube put in, and we just kept feeling like we needed to give her a chance to try and eat on her own. They didn't think she could, they didn't think she had the mental capacity, I guess. She proved them wrong, and she ate like a little champ, but when we brought her home, it took us a while to figure out what bottles worked well and what position we had to hold her in. It was just a little trickier, and just like everything she does, it took her a little bit longer to figure out how to do it.

Nathan: Just like with any other child, it's hard to determine. Maybe because we knew she had a diagnosis or special needs, we felt like things were a bigger deal, but a lot of times I look back and think, "We did that with all of our other kids too." We treated them the same way, and we didn't treat her any differently. It's just like we were a lot more aware of what was going on. I remember struggling with our boys eating and finding out what bottles and getting them to sleep, and all these different things. They had the same struggles, but you are just so much more aware and clued in….

Tiffany: …and nervous.

Nathan: …yes, and nervous, and not knowing what is going to happen. The thing that I guess is the challenge for me that I don't necessarily see as a challenge is that she requires a lot more attention with the feeding or with just everything, but to me, that's a blessing. It can be a challenge because it's a lot more attention, but it's also a lot more rewarding because you have to spend a lot more time and a lot more energy. Whereas it seems with the boys, sometimes we let the phone or the tv take care of them. It's funny because she likes that for a second, but then she wants your attention.

Gerald: Sure.

Gerald: How old is she now?

Tiffany: She is three.

Gerald: She is three years old. Let me ask a little easier question, what are the joys?

Nathan: When you walk into the room, it seems like she lights up, like she would rather be with you than with a toy. It's funny, because at first we started buying all these toys when we got home. We wanted to have these toys to help her with this and help her with that, but she could care less about the toys. If you're in the room, she wants to be in your lap or she wants to have your attention. If you have your phone and you're trying to send a text or look at it, she will push your phone away and grab your face like, "No I want you to talk to me, and I want you to focus on me," and I think that is what is so much fun. She really lights up, like when I came home just a few minutes ago, and I walked in the door, she was sitting on the floor over here. She has really bad eyesight, which is another thing with the Aicardi, but as soon as I walk into the kitchen, I don't have to say anything, she just starts screaming, and that's just so fun. You walk over there, and she's jumping like, "Give me a hug, give me a hug!" You can tell that she is a lot more sensitive to feelings and emotions because that's the way she communicates, through feelings and emotions, versus verbal or anything like that.

Gerald: Tiffany, do you have some things to add?

Tiffany: She knows when people need love. She's got a lot of love to give. About a year ago, it would take us almost an entire hour to walk down the hallway at our church because she wanted to hug everybody, even people she'd never met. It's okay, I'm not crying because I'm sad, I'm crying because I think some of my friends are my friends because of Ellie. They love Ellie. They want Ellie's love and attention. There are two sides to working so hard for milestones, you work and you work for something like sitting up, we worked so hard for sitting up for seven or eight months, and when she did, even her brothers were her biggest cheerleaders. Her brothers will say, "Mom, she just said 'da.' She just brings so much love and happiness.

Gerald: You alluded to this a minute ago, but what has been her impact on her siblings as well as your extended family?

Tiffany: It has brought us a little closer, and everyone wants to be right by her. Honestly, it wasn't even until this last week that her older brother kind of realized there was something different. They loved that even with therapists coming in and transitioning out of early intervention. She's got a fan base for sure. People just want to be around her.

Nathan: Right now, we are really trying to work on getting her to walk, that's our big focus. We are trying to get her walking, and the problem is that I'm always trying to get people to put her down, saying, "No, no, no, no, she needs to walk, don't carry her because she needs to learn to walk." Which won't happen if we are always carrying her around everywhere. When she goes to my parents for a visit, we walk in the door and she's got to sit on everybody's lap, and everybody wants to talk to her and see what she is doing.

Tiffany: She loves that.

Nathan: They love that too, so it's really hard. She is the center of attention wherever we go.

Gerald: So, Nate, if I were to come to you and say that I just received a diagnosis, that I have a child in utero with similar conditions to your daughter, what advice would you give me?

Nathan: Don't Google it! Sometimes when you initially start searching for answers, it seems like all you hear are the scary things and the worrisome things, and it's important because a lot of these diagnoses come with challenges. It is good to know what those challenges are, but just don't lose hope, because as soon as you can get past that and realize that you know that there are some challenges, there are so many more blessings that come with that. Realize that there is happiness that can come from that, and it's not all gloom and doom. The doctors will always give you the gloom and doom, and in fact, we changed our initial doctor because we just kept saying, "Well, what if she can do this, and what if she can prove us wrong," and he said, "She's not going to, she's not going to." I said that I was going to talk to someone else because we are going to give her a chance, and we are going to give her all the love that she might need. Whether it's good or bad, we are still going to love her, and so I would say just hold on because you just got to get through the hard part. Once you get them here, once you can hold them and once you can see their face, all of that fear goes away.

Gerald: Tiffany, what advice would you give?

Tiffany: You can still be normal, and like Nate said, you just can't lose hope. I think that is what got us through. That initial part is so hard, and it's okay to be sad for a minute, but once you get this little one, they have so much love and so much more than just a dang diagnosis. They bring out the best in everyone they are around, so I'd say to be hopeful.

Gerald: What do you think the future holds?

Tiffany: Gosh.

Nathan: I think about that sometimes, then I actually think I don't really care. Sometimes I wonder what she'll be like as a teenager and I think, "Whatever it is, whatever it brings, I don't care," because what I'm mostly concerned about is today and tomorrow and the next day, getting the most time and joy out of it as I can. It's not really hard with her because she is always so happy and so cheerful, but it's like we think about it, but at the same time, we don't really worry too much about it. I don't really worry about what she will be like in the future, I'm happy with who she is now. We'll give her all the love and care she needs now, and whatever the future brings, bring it on and we will deal with the challenges or with the happiness that comes with it. We'll just take it all in stride.

Gerald: Last question, would you do it again?

Tiffany: Yeah.

Gerald: Would you do it again, Nate?

Nathan: If I could take out the initial fear and what am I going to do and how am I going to deal with that and be at this point, absolutely. It was after six months when we realized that this really isn't so bad, that there are a lot of blessings that come with hindsight.

Tiffany: I also feel like we've already done that, and even though we already know the outcome will be so good, I'm not going to lie because it doesn't come easy, but the joy that they bring outweighs the bad any day.

Gerald: Thank you.

Tiffany: Thank you.