An Orange Socks Story- Sarah: Potocki-Shaffer Syndrome

Interview by: Gerald Nebeker, President of Orange Socks

Gerald: I am so grateful to Sarah for taking the time for an Orange Socks phone Interview. Her daughter, Betty, has been diagnosed with Potocki-Shaffer syndrome, a very rare genetic disorder. Betty's life inspired Sarah to create the podcast, bringingupbetty.com, that features stories of parents of children with special needs.

Gerald: First question is when did you find out that that your daughter had Potocki-Shaffer syndrome?

Sarah: She wasn't officially diagnosed until she was about eight months old, but we kind of had a long journey to get there. When we did the standard 20-week ultrasound, there were a few red flags that came up on that normal ultrasound you get at the doctor's office. They said that there were three soft markers to indicate that there could be a problem chromosomally with our baby. The doctor requested some additional blood work, and then a level two ultrasound. We went to maternal fetal medicine and had a follow-up with that, then we had that level two ultrasound. By then, they had gotten the results of the additional blood work, and with all of that additional information, they said, "Oh no, your baby looks great, like no worries. Just enjoy the rest of your pregnancy. You should be expecting a totally healthy baby." We had kind of this rollercoaster during the pregnancy, but then we thought we were good to go. When she was born, we noticed she was different from our first daughter. Betty is our second, and she was super sleepy, like not super engaged, but everyone had told us before she was born that this little baby is going to be her own person and don't compare her to her sister, so we were trying to do that. We thought she was just different from her sister, so we chalked it up to that. Then as time went on, we started to have some more serious concerns. When she was two months old, we went in for her check. One thing that she had was an umbilical hernia. It seemed huge to us. It was in her belly button area, stuck out about two inches from her stomach and was just protruding. It seemed like a huge concern, and she also had a weird choking, coughing, gagging thing she would do. She wasn't smiling, and she wasn't looking at us. The doctor said, "That this little hernia looks like a big deal, but it is the least of your concerns," and that was totally a sobering moment. He wrote that she had global development delays on an early intervention pamphlet and told me to call them. I thought, "Man, she's only two months old. How could she be globally delayed at this point already?" There were some milestones she was already missing, but we were really encouraged by our doctors who said that with just some additional help, she would catch up to her peers and continue to progress. He said, "Man, if she's just two months behind, then in a year or two years, no one would be able to perceive the difference." We hung onto that for a while. As time went on, it wasn't as if she wasn't progressing, but it was so slow that we wondered if there was more to it. The doctor had definitely referred us to several additional specialists, and all of these tests were coming back normal. We did like a CAT scan, an MRI, a chest x-ray, a study and just test after test with every concern that we had

coming back normal, so we didn't really know what to think. We felt that we were dealing with a potentially really scary thing, but we didn't know what it was. Part of that just made everyday life harder because any time she would get a little rash or have a new cough or something, we were like "Oh no, is this the beginning of a manifestation of some huge thing that we just haven't discovered yet?" Finally, we got in to see a geneticist. Our neurologist requested genetic testing, and our insurance agreed to cover it finally. It's quite expensive, so it wasn't something that we felt confident that we needed to do, to pay the seven thousand dollars for this one test. We finally saw a geneticist and for whatever reason, when he requested it, our insurance said they would cover it. I think there were three things he was looking for just based on her development at that age and some of her physical characteristics. One of the things he had on his list was a significantly decreased life expectancy of 18 months or so, and that was very scary because that would mean we were halfway done with our baby's life, potentially. Thankfully, those tests came back negative. The genetic testing did reveal that she was missing a part of her 11th chromosome which was associated with Potocki-Shaffer syndrome. There's not a lot of information about it, because at the time they told us, there were four known cases in the world. There has only been one clinical study done about the syndrome, and there was only a six-page report on that. By the time we actually met with the geneticist to discuss her diagnosis, I had memorized all the information that was out there. Before she was diagnosed, I was desperate to have a diagnosis. I thought if we find out what it is, then we'll have an action plan, a roadmap. We'll know what to expect; we'll know what to do to help her. That just wasn't the case because it's so rare with such a huge spectrum of outcomes that, really, we had a name for it, but we had no other information.

Gerald: So, in your case, the diagnosis offered no comfort or help at all?

Sarah: Not really, although with that diagnosis, we were able to find some other families relatively quickly through Facebook. Surprisingly there was a Facebook group, and then there were some good organizations that can put you in touch with other families that have this syndrome, so we got in touch with a few other families. That was the biggest thing for us, just knowing people who understood and who could help us know what to expect, what they had dealt with and what life was like with their children. That was probably the biggest thing even though we didn't get clinically a lot of information or a name that could help put us in touch with other people further down the road with it who could tell us what we were dealing with.

Gerald: Okay, what have been the hardest things for you in the last four years with you with your daughter?

Sarah: There are day-to-day hard things which some of the other families who have children with Potocki-Shaffer syndrome haven't really dealt with very much. So far, that's been hard, because we feel a little bit alone with it. I'd say one of the biggest things that's come up is some of her sensory issues. She has a sensory processing disorder, and it makes everyday happenings around the house feel impossible to her, so she will scream and cry when we, for example, open up the mayo, open an envelope or a box or scissors, rifling through a box of Lego's or if we are preparing food in front of her. There are the funniest things that you would never put

together, especially related to sound. As we are expecting our third baby, we got a new van with automatic doors on the side that she just cannot handle, so the first several months, every time we got in and out of the car, it was a total meltdown. That's been a challenge. Also, the fact that she's growing is difficult because she doesn't walk yet. She weighs about 40 pounds right now, and I know 40 pounds is 40 pounds, but it's a heavy 40 pounds, which is weird because she has low muscle tone. At the same time, she can be very strong and has this way of throwing her weight around that is really taxing on anyone who is trying to carry her or help her. That's been a challenge. Then I'd say the thing that kind of creeps in that I have to push aside are thoughts about the future. Because we were parents of a typically developing child first, we have come to understand eventually that those things are just a phase, that most of the hard stuff is just a phase whether it's battles at bedtime or mealtime or tantrums, your kids will normally go through those and then come out on the other side, and you both learn something along the way. With Betty, those phases can be really, really long because her development is slower, so it's hard to remember that this could possibly just be a phase. It's easy to think, "Oh my gosh, the rest of our life is going to be like this." I just have to remember that we definitely don't have a crystal ball, and we don't know what's going to happen and how far she will develop and all the skills that she can possibly gain, and we hold on to that hope that there are good things to come, too.

Gerald: Tell me about the joys.

Sarah: She has a sweet personality and spirit, and we just love her. She brings so much joy. I love watching the interaction she has with her older sister. A lot of my friends who have more than one child are dealing with a lot of sibling rivalries and battles, and we really don't have to deal with that at all at our house. They just have a really sweet relationship that I don't know they would have had if Betty was typically developing. There is an extra bit of sensitivity and tenderness and a love that that is a part of our family culture and hopefully running more compassion toward everyone around us. There have been several times where right when we're kind of at a breaking point, some big thing happens that is either developmental or medical progress for Betty, most recently with the birth of our third child who's just six weeks old now. Things are just kind of chaotic, we're living in survival mode. We have been dealing with a lot of challenges with her. And then in a week's time or a month's time, it seems like her vocabulary doubled almost overnight, which is huge for her, because her communication has been a big challenge. So, it seems like every time we hit a road block in one area, there might be a developmental leap in another area, and that's been really encouraging.

Gerald: You mentioned that she's had impact on siblings in the interaction that she has with her older sibling. What impact has she had on your extended family or your neighbors or friends?

Sarah: It has been really sweet to see her born at a time when in our little neighborhood, there were six babies due right around the time she was due, so I was excited to have her grow up with this little pack of friends in her neighborhood. Then as she was diagnosed and obviously not keeping up, like it wasn't just a little extra help in catching up kind of thing, I realized that she wouldn't be in that little pack with her peers. That's what I thought. However, even those

little friends have been huge defenders of her, so cute and sweet, and they just love her. That has been so neat to watch. As far as extended family, we don't have cousins that live in the same state or anything, but when those kids come to town, they all just want to see and hold and play with Betty. I don't know if it's because of Betty, but my oldest niece has just finished her freshman year at BYU studying special education. I think that probably because of a little more exposure to her cousin, a child with special needs, she has seen just how special they are and the extra help that they need, and she wants to pursue that for her career, so I think that is really cool.

Gerald: Just another question. If I came to you with either an in utero or shortly thereafter similar diagnosis, or in your case, with a similar syndrome, what advice would you give me?

Sarah: I would say to learn everything you can to help your child with the best life that you can. Give them every access to learning and development, and at the same time, the probably best advice that we've gotten is to not let a stack of symptoms get in the way or cloud your view of your child. When Betty was just two or three months old, we were just starting to unravel this puzzle of what was going on with her. I remember her pediatrician telling us not to let her become a stack of symptoms, that we had a beautiful daughter, so we should go home and just love her. That is just the truth, no matter what your child is dealing with. I think it's really unfortunate that we live in a world where a lot of people form their expectations based on a blood test. I don't think we should do that, it doesn't benefit anyone. We were well-informed about the clinical side of things, but at the same time, we didn't let that put any road blocks ahead of our daughter. We have realistic expectations, but we also aren't going to let that be formed by the medical community. I guess I would just encourage other parents to do the same.

Gerald: I appreciate that you have a daughter with a disability, Potocki-Shaffer syndrome, which is pretty rare and not a lot is known about it. You have a lot of responsibilities with your other children, and you have some physical demands also in caring for Betty. Was it worth it?

Sarah: Oh, for sure, for sure. She is a part of our family. This might sound weird, but I really can't imagine her any other way. This is just a part of who she is, and it's a part of who our family is, and thank goodness for that because she has taught us so many lessons that are beyond what we could ever know or experience if she were not a part of our family. It was definitely worth it.

Gerald: Great. Thank you so much for taking the time to talk to me.

Sarah: Sure, my pleasure.

Gerald: I appreciate it.