

An Orange Socks Story – Sarah: Hydrocephalus and Other Physical Differences

Interviewed by: Gerald Nebeker, President of Orange Socks

Gerald: I was happy that Sarah took the time to talk with me by phone about her daughter, Isabella, who has hydrocephaly and other physical issues. Sarah was pregnant with twin girls, and during a check-up, it was discovered that one of the girls, Isabella, had issues. Sarah, when did you find out that your daughter had hydrocephaly?

Sarah: We found out that she was starting to develop the hydrocephalus around 16 weeks, and from there, we would get a scan about every two weeks, which went to every week at the very end. They do measurements of ventricles, and there are two lateral ventricles inside the brain, so they would just measure those at every scan, and they knew she definitely had it.

Gerald: You got the diagnosis in utero that your baby, who you named Isabella, had hydrocephaly. What advice did your attending physician give you?

Sarah: Because we were having identical twins, she did mention possible termination if it were just a singleton pregnancy instead of identical twins, she would be terminated. Ariella, her twin sister was completely normal, and everything was growing correctly with her. So, we continued on with the pregnancy. From then on, no one really pushed termination as much because we were having identical twins. It was like, “Well, one's doing good, and we already know that the other one has this. So, let's just continue on and see at birth what happens.”

Gerald: Tell me, what have been some of the challenges? You had twins, so that's a challenge in and of itself. You mentioned that because she had a sister in utero that you weren't pushed to abort the baby, so that wasn't really something that you necessarily had to contend with. What have been the challenges since her birth?

Sarah: I would say one of the first challenges was due to her enlarged head. We already knew that we were going to have a scheduled C-section. If she were a singleton pregnancy, I'm not sure how far along they would have pushed the pregnancy. With identical twin pregnancies, they usually don't go past 37 weeks gestation because of the placenta breaking down. We knew that was scheduled, and that was fine. It was the safest way for them to be born. I guess that was one of the things that we didn't really have much of a choice for. Some of the other challenges we've had are that we found out when she was born, she also has other birth defects. She was born with a perforated anus, so she doesn't have an anus, and she also has a persistent cloaca where her vagina and urethra are connected inside. So, she only has one exit, and that is where she urinates. The first initial challenge that we had was that she couldn't poop, so we had to do surgery so she can go to the bathroom. She has an ostomy bag on the left side of her abdomen where she goes to the bathroom. That was totally new for us. It's honestly like changing a diaper, but at the beginning, we had to learn how to correctly place the bag and how to maintain her skin around the stomas so they don't get irritated. That was a little bit challenging. Then on day four of life, she had brain surgery. They used a couple of caps on her brain in order to get the fluid out temporarily, to relieve that pressure inside of her head. Her type of hydrocephaly is the congenital type, so it's a blockage between the third and fourth ventricle called aqueductal stenosis. There's a tiny little tube that drains from the third to the fourth ventricle which didn't form properly because of that blockage, so the fluid is building up over time. The neurosurgeon made a new path, a new hole for it to flow and regulate. He also cauterized the part of her brain that makes more spinal fluid, and that will help bring down the fluid as well. Because she had two procedures done so far it's working, but there's a chance of it failing, and if that hole that he made closes up, she will have to go in and get a shunt, and that involves the tubing draining into her abdomen. That's why they didn't go with the shunt to begin with because she already had the abdominal surgery. Then eventually, maybe around six months of age, she's going to be having reconstructive surgery on her anus,

her vagina and urethra. We're not sure if it will be done at the same time or separately, and then she'll have her ostomy closed up on her belly once everything is done. So, it's another surgery after the fact.

Gerald: So, she's had to have several surgeries and has several to go. Does she also have an intellectual disability or not? Do you know that?

Sarah: Yes. They first mentioned VACTRL to us, and I'm not sure if that's another genetic term as we didn't know any of this stuff, but VACTRL affects the midline of the body as far as genetic defects. She does have a tilted vertebra on her spine, but she does not have VACTRL. I didn't want to bring it up like she had it, but it was mentioned that she could have it. She does not have VACTRL. We did have her genetic testing done. She has a microduplication on chromosome 16 short arm, and a microduplication on chromosome 17 short arm. The diagnosis for 17 is called Charcot-Marie-Tooth syndrome, which is just the name of the doctors who discovered this syndrome and has nothing to do with her teeth, which is funny because a lot of people ask, "Tooth? What does that mean?" Charcot-Marie-Tooth syndrome is a neurological muscular disorder where her lower limb muscles, basically from her knee down, will deteriorate over time, and she will become weaker in her hands as well. This type of syndrome can either mildly affect her or she can have it greatly affect her. They don't predict her being in a wheelchair or anything, but she may need braces as she gets older. There's no syndrome with that 16 duplication, but it's linked to learning disorders, speech delay, ADHD, autism and schizophrenia. However, that can also either be super mild, as if you wouldn't even notice any symptoms from that, or on the other hand a little more severe, where we know she's autistic or that she has ADHD. It's kind of whatever she was born with.

Gerald: So that's something you will have to wait and find out later?

Sarah: Yes.

Gerald: Let me ask you a little about the joys, because a lot of times we focus in on the things a child can't do, or the surgeries or whatever. So, tell me the joys of having Isabella.

Sarah: I would have to say first that she has this amazing peace about her. She hardly cries, and she's the sweetest baby ever. With her being in the NICU the first month, I know the NICU is hard and she was different than most NICU kids. She was premature, but she had a lot of other things going on with her, and when you look at her, the hydrocephalus is obvious as she has a large head. But when you look at her, you can't really tell that she has anything wrong. That's what really affected us the most. We didn't know all the other stuff. Despite all of that, all these surgeries and how many times she's been put to sleep for procedures, she's just so sweet. There's something special about her. We can't put our finger on it. My mother-in-law, my mom, my husband and I all just see this peaceful, sweetest baby we've ever seen. Comparing her to her sister is kind of funny because her sister's all spunky and more on the fussier side, but Isabella just has this aura about her where she's just so at peace with life. We've really enjoyed having her. Also, the fact that she's a twin and grew inside next to her sister makes her really sensitive to touch. When she's touched or held, she reacts so much stronger than what we've noticed with our other children. She's just so connected to you, it's just unreal. That's one of the things we've really noticed that's been a real joy to have her at home and her reaction. Our five-year-old son says, "Mom you drank too much water, that's why she has too much water in her head."

Gerald: That's funny! So funny that he would say that.

Sarah: I know right? We've had a real good support system with our family.

Gerald: I was going to ask you a question about that. How has Isabella's birth and her life so far affected your extended family? You mentioned briefly your immediate family, but how about your extended family? How have they reacted to her? What have been their thoughts?

Sarah: Everyone has been really supportive. We didn't really talk about her having hydrocephalus until about a month before we gave birth. We didn't want to advertise it. I don't know why we didn't. I don't know if we thought, "Oh, she's going to be fine at first" or, I don't know. We just kind of had this idea that

we didn't need to tell anyone, and then we realized that we did need to tell people that she has what she has and that she's special because of that. Our families reacted very well, and were very supportive and loving. They've asked if there's anything they can do for us. Unfortunately, we don't live next to our family. My husband is in the military, and we are not able to live next to our close family, so they have to fly to come see us. But we've had that support. When they do come, it's so awesome.

Gerald: That's terrific.

Sarah: Yeah.

Gerald: So let me ask you this, if I came to you with a diagnosis that I had a child in utero who had some of the conditions that your daughter, Isabella, has, diagnosed with hydrocephaly and maybe some of the other conditions that you mentioned, what advice would you give me?

Sarah: First, I would not say termination at all. With technology nowadays, they have the ability to help with surgery depending on what type of hydrocephalus has developed. I would say some things to expect are NICU time, or your child is going to have surgeries right off the bat, which is hard to deal with because they are so tiny. But they are actually stronger than we give them credit for. I would say to keep on pressing on, pray, get early intervention. Go in as soon as possible for physical therapy so you can have that going for them to make up for their head being so large and heavy. That's pretty much everything I would say.

Gerald: Well, wonderful. Any other thoughts that you want to add?

Sarah: No, I'm just really glad that we got to talk. This is a great outlet, your website. I really enjoy it. I like how it is a place for people to reach out when they hear all this doom and gloom, there are positive stories to see. The medical field can be kind of cutthroat sometimes. It makes you feel like there's no hope sometimes. But when you see others and their testimonies and what they've gone through, it's great. We're all here to support each other, and it feels really good.

Gerald: I appreciate Sarah and her candor. I am certain her story will be an inspiration to other parents.