# An Orange Socks Story: Gina-Down Syndrome and SMA

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 Gina about her 12-year-old daughter, Larkin, who has Down syndrome and spinal muscular atrophy or SMA. Gina says Larkin is the only person known to have both Down syndrome and SMA. I know you're going to enjoy learning of the positive impact Larkin has made on Gina and her family.

Gerald: Gina, thank you so much for taking the time to do an Orange Socks interview with me today about your daughter, Larkin who has Down syndrome as well as spinal muscular atrophy or SMA. When did you find out that your daughter had these issues?

Gina: We did not find out that Larkin had Down syndrome until she was delivered. I had her via C-section, and my doctor showed her to me over the little drape; I knew immediately she had Down syndrome. He looked at me, and I looked at him, and because he had been my OB for 18 years, it was just a silent conversation between us. We didn't have to say a word, we both knew what it was. Larkin's dad had no idea, so he was just happy. She was in respiratory distress, so I got to give her a kiss and hold her briefly, then they took her to the NICU. That was the first diagnosis we found out obviously right away. She also has SMA Type 1, which is the most severe type of SMA, and we didn't find that out until she was two-and-a-half months old, so that was a stressful ordeal. We were in the NICU for a long time, actually twice. She was not able to breastfeed or suck on a bottle, so she had an NG tube which went down her nose. Of course, when you're in a place for a long time, you become fond of people, and I became fond of her nurses. They said, "Gina, why don't you do a feeding tube, a gastrostomy, and that way we can get her home. She just has Down syndrome and is just low-toned, but she'll get stronger." That is what our focus was on, that she was just weak and was going to get stronger. I believed what I was hearing because I had four other kids who were perfectly healthy, and we were naïve, I guess. We didn't know anything in the medical field, so we went ahead and had the feeding tube surgery and took her home. She came home with a nasal cannula and an O2 monitor, and that was it. She was home for 11 days, and I tell everyone that it was the most stressful 11 days of my life because I knew she was aspirating. They wanted me to try and bottle-feed her, to try and work on that, but I could tell that she was aspirating and it was crazy. I was also feeding her via her feeding tube through her stomach, and as the days progressed, I was taking pictures as I am an avid photographer. Looking back, I can see the slow progression of her just getting more sick, and it makes me sad. and Before we had left the hospital, they said that this was the temporary GT feeding tube in her stomach, and if for some reason it comes out and the hole closes up, we would have 20 minutes to put in this thing they gave us, but we needed to take her to the hospital if that ever occurred. I called Larkin's dad at work and said, "I think her feeding tube is loose. We should probably take her to the hospital." When he got home, we took her to the hospital. After the 11 days, her O2 monitor had alarmed a couple of times, but then went back up, and again, I'm a novice so I just thought that was normal. Anyway, we took her to the hospital, and she was dusty in color. They asked how long she had been like that, and she had beeped on our walk in from the car but then had come up again. They took her right back to the NICU, and we were very fortunate because usually when babies leave the NICU, they are not allowed back in, they usually go to the PICU, which is the Pediatric Intensive Care Unit. They accepted her back into the NICU into an isolation room, and within a few days, she coded and had to be intubated, and at that point, they consulted a neurologist. When the neurologist came in, it was literally heart-breaking because he said he suspected spinal muscular atrophy. I had heard of muscular dystrophy but I had never heard of spinal muscular atrophy, so I asked what that was. He point-blank said not to worry about it until the DNA test comes back, and he wouldn't tell me what it was. I immediately went home and Googled SMA, and at that point, I just broke down. What I read was that it was the number-one genetic killer of age two and under, and they lose the muscles to breathe and feel well. That's all I remember. I had to wait 14 days for the test to come back, and it was positive. In those two weeks, I knew what I was going to do if it was going to come back. I knew my options were to have her trached and put on a ventilator or basically extubate her and let nature take its course. That's exactly how the doctor explained it to me, and I couldn't even imagine doing that I just couldn't do it. At two-and-a-half months old, she had the trach surgery, and she's been on a vent ever since. I'm not going to tell you it has been easy, it has been a roller coaster, but well worth it. She has been such a blessing.

Gerald: So, she's 12 years old now.

Gina: Almost 12.

Gerald: Almost 12.

Gina: Yes, January 11

Gerald: Over these last 12 years, what's been the hardest part for you?

Gina: I get emotional over it because lots of things have happened in the almost 12 years. It's very hard on your marriage, and it's very hard on your family. I have four other children. When Larkin was born, I had a four-year-old, my older three kids were in high school, so they were pretty self-sufficient, but our son was four, and he has had to learn how to do a lot of things on his own. He has been put on the back burner. He's amazing. God knew that I should have him because He knew that Larkin would be ours one day. He is the happiest kid I've ever met and so loving, and so I'm very thankful for him. I'm very thankful for all my kids. It's very hard taking care of a special-needs child, but it's even harder when they're hooked up to so many machines that you can't really leave. I stay in her room all the time if I don't have nurses, so I'd say that's been difficult because of her care. I'm used to her, she's a piece of cake. It's so easy to take care of her. She's happy, she loves to read, she loves music, and she laughs. She can't communicate verbally, but she has some facial movements. At about four years old, she lost her muscles to smile, but you can tell when she's happy. You can just see her eyes light up. She's just like my best friend. I hold her hand all night long. When I come home from work, I do everything I need to do before the nurse leaves, and then it's my turn to be with her. I literally just hold her hand all night long, and we watch TV or we paint fingernails, and we just make the best of it.

Gerald: So, tell me about the joys. You've mentioned a few of them, but what have been the joys in having her in your life?

Gina: Oh, my gosh, like I said, she's such a happy girl, and I'd say she never complains, not that she could verbally complain, but you know when a kid is happy and you knew when they are sad and hurting. She rarely ever cries, the only time is when we call it "getting too junky," if there's mucous in her lungs that needs to come out, or we have to hold her trach so it holds the mucous in her lungs, and we hold her nose and her mouth. There are three things that need to be suctioned, and she can basically tell us just by crying or a grimace on her face before her machine even alarms that something is wrong. The joy with her is just being able to be with her. I love introducing her to new things, seeing things through her eyes and how she responds. Today my work Santa came in, and I videotaped a message from Santa to Larkin. I was so excited to show her, and she was happy. It's just so magical with her, so happy being able to be with her. Those are my joys. She loves everyone, and we have so many more friends just because of Larkin.

Gerald: You mentioned her brother who's just immediately older than her, but what impact has her life had on your immediate family with maybe your other children?

Gina: Honestly, I think my kids are very compassionate towards others, especially disabled people and children. Their hearts are just so full of love for them, and they're the first ones to stick up for someone who is not typical. So, she has been a blessing to us. She's normal, I mean, we've lived with this forever, so we don't look at her and see what she is, we look at her and see her.

Gerald: What has been her impact on your extended family?

Gina: We don't really have a huge extended family. She does share a birthday with my dad, and they really have a tight bond that is amazing to see. There can be times where she does make her noises that's like her talking. She won't say a peep to me, but he'll walk in the door, and she's "aaa aaa." She has words to tell him, and he'll kiss her forehead, and she'll just look for more kisses. They have a really unique bond that's neat to watch. I think that our extended family is more compassionate towards others and knowing what's important, not the material things, but just being together. I think that's how she has impacted our lives.

Gerald: If I were to come to you just having received a diagnosis or confirmation that I had a young child with Down syndrome and SMA at the same time, what advice would you give me?

Gina: First, I would say you're unicorn number two, because as far as we know, Larkin is the only person in the world with Down syndrome and SMA type 1, so we call her a unicorn. I would try to be as much help as I could. I would answer questions. I would not give false hope, but I would give hope. I think it's what you put into the care of your child because it is a lot of care. We don't let anyone come into our home who is sick or who has been sick recently. We say that we're germophobes. We wash our hands all the time. We make sure her environment is 100 percent clean, 100 percent of the time. I would just give them pointers and say that they can do it, because you can do it, you can make it work and it's worth it.

Gerald: Any closing remarks? Anything that you'd like to say that we haven't covered already?

Gina: This always makes me emotional, but after Larkin was born and I was still in the hospital, my doctor called me from his home and said, "At the time, we knew that she had Down syndrome. God knew that she needed you, and you needed her." I truly do believe that, because she's made me so much stronger, more independent, more outgoing and more bold. I see that for myself, and I see that for her. I have to advocate for her. There are so many times that I've saved her life, literally saved her life. Even though I have nurses here, I'm the one who takes over. I'm the one who's on-call 24/7. I run in in the middle of the night and save her life, and I kind of feel like she saves mine because she is amazing.

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.