An Orange Socks Story – Nikkie and Sean: Osteogenesis Imperfecta

Interview by: Gerald Nebeker, President of Orange Socks.

Welcome to the Orange Socks Podcast, where we are inspiring life despite a diagnosis. I'm your host, Dr. Gerald Nebeker. Nikki and Sean describe the moments leading up to when they discovered their son's diagnosis of osteogenesis imperfecta, which is a brittle bone condition.

Nikki: We went in for a routine 20-week ultrasound and 45 minutes went by. At the very end she said, "The baby's legs are short. I can't find the babies legs." She said, "Well, I'm going to refer you to the bigger hospital." So, we went down there and he said, "Yes, we think it's a form of dwarfism based on his femur length. His femurs are shortened and curved, and so that's a marker for other conditions." We had to keep going back for follow up ultrasounds. At around the 28th week it became progressively clear that it was more than just dwarfism, he referred to it as brittle bone. He didn't think it was that until the 28th week ultrasound. He said, "I think, based on what I'm seeing here, I think it's osteogenesis imperfecta." So, we ended up getting an amniocentesis done. We really thought about it, whether that was something we wanted to do or not. I thought, "Well, if nothing else, it will prove the doctors wrong that it's not this condition and we can at least know a little bit more." At 32 weeks, we got the results back that it was Osteogenesis Imperfecta.

Gerald: Sean had feelings of uncertainty after receiving the news. But ultimately, he came to realize that caring for a child in this way could be a beautiful blessing.

Sean: During the ultrasound he showed us his head was soft. You get a knot in your stomach; you don't want anything to be wrong with your kids, but it didn't get worse than that. We started to understand what this was and we talked about it as pragmatically as we could. The worst-case, and best-case scenarios. We started to just mentally move into the thought process.

Gerald: The couple did a lot of research and addressed many misconceptions when they embarked on learning about their son's condition, and they faced a tough decision whether or not to continue the pregnancy.

Nikki: At 20 weeks we went straight to genetic counseling, and they said, "Well you have 24 weeks to terminate if that's what you want to do." Whenever we found out the diagnosis officially at 28 weeks, we wanted to explore our options, "Is this something we really should continue with? This is really scary. Do we want to bring a child into this world who will suffer physically and break bones all the time?" We thought about what his future would bring. Having bones that break for no reason, responsibly, can we really bring a child into the world that is going to suffer like that? We kind of had to wrap our minds around it, but I think the unknowing part was really hard for me because they couldn't tell us if it was lethal. They couldn't tell us if the baby would survive birth. They gave us no information at birth about what we could expect. Those last five months of my pregnancy was about a lot of faith and you know staying present and moving through it. It was so unknown. Sean and I decided to not share this information with our family.

We told a couple of people, because we really didn't know. We just wanted to allow whatever was meant to be, be and we didn't want other people's negativity to affect us. Because we didn't know, nobody knew if he was going to survive at all.

Gerald: Nikki discusses the fractures her son suffered in utero and during the birth process. She credited this research and learning with minimizing these issues as much as possible.

Nikki: The radiologist counted 26 plus fractures that were both in utero and from birth.

Sean: My wife did as much research as she could immediately and really took ownership over how this was going to work through the pregnancy. She did the research and she said, "You know, based on what I've learned, that a C-section isn't necessarily safer for him. They need to go in and grab the torso." His torso is his ribs and his heart, that was a big concern, and she said, "I want to do this natural," and so we put his legs and his arms at risk more than his torso. There was no science behind it. She did it without medicine, without drugs. They must have piled 35 people into the room to watch because knowing he had OI, knowing that there was no drugs, a natural birth with a baby with OI, I don't know if it's ever been done before. I give my wife so much credit.

Nikki: I was really confused as to why they did not refer us out to specialists. I had to go do my own research whenever I was pregnant to find OI specialists. They did not refer me to anybody, and I thought that was strange for our medical providers. I had a conversation with a genetic doctor in Delaware, they specialize in OI there. He said, "I have a research paper that I was a part of, that studied vaginal birth or cesarean birth the fracture risk is the same. It's not substantial. There is no reason to do a cesarean for fracture risk alone." He did have multiple fractures in various stages of healing, but they didn't splint them or anything. His APGAR scores were really good. He didn't need any oxygen and he just did really well. He was only in the NICU for five days.

Gerald: The couple describes the biggest challenges they have faced up to this point.

Nikki: The pregnancy was really hard for me. It was really hard for me because I previously had a home birth and I had this idea of what my fourth pregnancy and child was going to be like. There is this beautiful poem called "Welcome to Holland" in the special needs community. That is why I'm so passionate about this whole thing. You think you find yourself in this one place, but you find yourself in this whole other place. Nobody told you that this other place is way more meaningful and profound than you had ever initially planned. It deepened my faith, but the hardest thing for me was holding this pregnancy and realizing that my worst fears may be happening. The worst things that I could ever imagine going through, might actually be happening to me and to my family, and how am I going to tell my children that they may lose their little baby brother. That is where I strengthened my faith, because I kept hearing God say, "Give him a shot. His life and his soul and his faith and purpose is far greater than any fear that you have. This isn't about you." And although I felt like, "Okay, we could lose this child," I also simultaneously felt this strong sense of, "It's going to be okay, you're going to have this baby, it's going to be great, he's going to do great things and you're just going to have to get out of the way." Those experiences were really hard for me. Now he is doing so good and life is just coming together, everything is really good.

Gerald: The siblings are fantastic with their brother. His special spirit has given them empathy and made their love grow.

Sean: They have all matured a lot because of it, all differently. We have an 11-year-old, four-year-old and an eight-year-old, but they are all very careful. There hasn't been one accident with the children not being careful enough around him. I never had a doubt, as soon as I knew that there was an option that this could be okay, I just kind of focused on that being the only possibility. That's easier for me mentally, and with anything in life. I had to keep telling Nikki the whole time through the hard part, "Honey we are going to have a beautiful baby boy, a little Italian looking baby." And we got it, I get to say I was right on that.

Gerald: Nikki shares what she hopes other parents would know if they are walking a similar path.

Nikki: I wish I had known then more of the better case scenarios. More of just information, more of support, more of this community, more of the sense of what possibilities are specific to my son's condition. We would like to know all of the answers about everything at all times, that would be nice, but it would ruin the fun. I would say that I have a relationship with God, I believe in God, and that really, it's in God's hands, and that God is already there. God already knows the outcome, and it's not your job to understand or to know. It's your job to just be present and to move through this experience because you aren't supposed to know what is going on necessarily. You aren't supposed to know everything. Everything that you are feeling right now is totally okay. It's the way it's supposed to be and you're going to move through this. This child is on a path all on his or her own that has nothing to do with you. You've been given this child to care for. You go through that period of grief. You can be sad obviously, process your emotions, that's super important, but at one point you have to decide, every moment moving here is a moment I'm never going to get back. Whatever the outcome is about this, I'm never going to get this moment back with myself, my future my children that are here, my husband, my purpose here on earth. Feel the emotion as it comes, get wrapped up in it and recognize that it's important to find those moments of joy. Make those moments now.

Sean: I got this mindset early on and I would want to give this to somebody else, "It's going to be what it is. It's going to be exactly what it's supposed to be." That's advice for everything in life.

Gerald: Caring for a child with brittle bones is a day-to-day journey.

Nikki: When he was born, he was extremely fragile. They used to be called "pillow babies," where you'd put them on the pillow and you would never move them. Now we know that they are supposed to be moved and they are supposed to get exercise and to put them in the water a lot to help him move. His bones have gotten stronger, he hasn't had any additional breaks. He had one when he was five months old in his arm, just from changing him. I'm careful when I pick him up. I don't pull on his legs, I don't pull on his limbs, because the mutation is in his collagen gene, every time his bones break, they do this remodeling process. That's what bones do, and that's why kids bones grow so quickly, when they are little they are constantly remodeling. Like my son broke his collarbone. Our only break in 2018 was my son's collarbone. When he gets treatment, it treats the bone that already grew and it draws calcium and blood and adds it to the bone. So, this treatment actually does strengthen the bone, and it's why he has to get treatments every two months. So, his bones will always be fragile, they will never be non-OI bones, but they are getting stronger and they will be stronger than they have ever been.

Sean: We didn't know if he was ever going to crawl. When he was born, we asked about his legs getting weird because we wanted to give him the opportunity to be mobile. And even the surgeon says, "Well if he shows signs of mobility." Sure enough, he gets up and pulls his whole body along. He is just so much better. He's going to do a lot of things, live a very fulfilled, very happy life.

Gerald: Finally, the couple wishes that others can understand that this journey is bigger than just them.

Nikki: I feel like it does take a lot of faith, and if you can find people that are supportive of you and your conviction, your story is going to pan out however it's meant to be. There are certain things that define you in life, and I think that it's all a good journey. Be as positive as you can be, and have a lot of faith.

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