An Orange Socks Story: Kaitlin- Genetic Disorder
Interview by: Gerald Nebeker, President of Orange Socks

Gerald: Welcome to the Orange Socks Podcast where we are inspiring life despite a diagnosis. I'm your host, Dr. Gerald Nebeker. In this episode, I speak with Kaitlin about her son Mason who is 20 months old. Mason is medically complex because of a deletion of two separate chromosomes, making him unique in the world. He has a congenital heart defect, congestive heart failure, microcephaly, optic nerve hyperplasia, bilateral auditory neuropathy, developmental delay, epilepsy and many other conditions, yet he is the light of his parents' life. I know you're going to enjoy hearing this story.

Gerald: Kaitlin, thank you so very much for taking the time to talk with me today about your son Mason. I'm honored that you would take that time. Tell me a little bit about Mason who is now 20 months old.

Kaitlin: Mason is the light of our life. We were told we would have a great pregnancy followed by a normal delivery of a normal child, but it didn't happen. He has gone through many difficulties and many different diagnoses, but nothing has held him back from being the happiest child. The love that he is surrounded by and the positivity he puts into everyone's life is amazing to watch, especially with his older brother. My goodness, just watching them together is unreal, and I'm so blessed to be a witness to them and to be their mom.

Gerald: That's super. What kind of diagnoses does he have? He has a whole bunch of them. I'm looking at a little bit of your information, and gee whiz, he's got a lot of stuff going on.

Kaitlin: Yes, he does. His main diagnosis, or somewhat of a diagnosis, is his genetic disorder. He has a deletion of two different chromosomes, the long arm of his sixth chromosome and the long arm of his 11th chromosome. They don't really know much about it. We were told that about 85 cases are known in the world with just the sixth deletion, and there is nothing on the 11th deletion, so they basically just told us the different anomalies that are included, which are heart defects. He has congestive heart failure and congenital heart disease which was repaired. He had a large VSD and ASD, which are holes between the chambers of his heart which was able to be repaired, and he has been thriving since. They also said that a lot of facial anomalies come with it, too, which they call dysmorphic features. That was very hard to hear from the doctors; that was basically the first thing we heard out of their mouths after he was born. I didn't get to see him, but they said he's missing his right finger and has dysmorphic features. Dysmorphic sounds like such an ugly word, so it was heartbreaking to hear. He also has optic nerve hypoplasia where both of the nerves in his eyes were not fully formed. He has hearing problems which we aren't too sure about yet; we have still have to have a hearing test. We know he can hear some, but to what extent, we still don't know. He has global developmental delay,s

so even though he is 20 months old, he is about at a three-month-old level. We are still working on head control, and once we can get that, all the equipment that will be available to him will really change his world. I cannot wait for that. He has globalization-related epilepsy, so he doesn't quite have full-blown seizures right now, but his EEGs did show focal slowing. He has had brain hemorrhages and brain cysts, so there's a lot going on with him. He is completely tube-fed, but we are looking forward to tomorrow when he has a swallow study, so hopefully he can start drinking from a bottle again or start taking purees. Even if he can't, we are excited because we are starting a blended diet for him so he'll be getting actual human food through his tube which I know will let him thrive even more. We just learn to look forward to the small things and get excited over everything. There are definitely more blessings than negatives here.

Gerald: Well, that's awesome. Over the last 20 months, what have been the hardest things for you and your husband to deal with?

Kaitlin: My husband and I have not been able to sleep in the same bed since he's been born. When we got out of the NICU, they said that since he's in heart failure, these are the signs that you need to watch for so that he's stable at home, and if any of these arise, you take him immediately to the ER. I couldn't even fathom sleeping in bed, thinking I'd sleep too good to miss something, so I slept on the couch with him. He was in his bassinet so his head was within a foot from me, so he was close. Even with his being so close, I've woken up to his not breathing so many times even though he has had his heart repaired. Waking up to your child not breathing is so scary, so I still sleep on the couch with his head near me so if anything arises, I'm right there. I haven't slept in the bed in two years. I feel like if I sleep in a bed, I'm going to sleep so good that I won't hear him, and that's not fair to him. That's our biggest struggle, just finding our new normal and understanding why I'm doing this or what's best for Mason. It's also a struggle for different people to understand that we can't go out and do certain thing,s because if he has a surgery coming up, we need to keep him healthy. This is why he hasn't had a hearing test in two years; they keep having to get rescheduled because he's to the point now where he has to be under anesthesia for it. He has been under general anesthesia so many times that they don't want to intubate him unless it's absolutely necessary. We can't even have a normal babysitter for him. Things can change with Mason in the blink of an eye. He can go from happy to aspirating in a matter of minutes. We've had to call 911, and it's scary, because we've dealt with it so we know what to expect, but I couldn't even imagine putting someone else in that situation. If that happens, it wouldn't be fair. I don't expect them to understand because before I had Mason, I didn't understand anything of the special needs world. Being in it now has opened my eyes to so much. Those are most of our main struggles as well as equipment and not having a diagnosis for Mason. Even though they call iat a "genetic disorder," there's no specific disorder for him, so all the insurance companies request the equipment, but they don't know if he will die from it because they don't know what to expect in his future. It's a full-time job between calling insurance companies and making sure my

son is getting what he needs, and that's another struggle. However, all the positives really outweigh all the negatives.

Gerald: Tell me all the joys that he's brought into your life and to the life of your husband and your other child. You have a five-year-old; tell me a little bit about the joys Mason has brought into your immediate family?

Kaitlin: I've never seen his older brother so loving and so happy until Mason came into his life. We always joke and say that he's the babysitter of the group. He can be in a room full of kids, and before he has fun, he needs to make sure everybody is okay. If anybody asks about his brother, he says,"My brother's fine," or "These are glasses to help my brother." He's like his own advocate; it's just so beautiful to watch. All of the kids at my son's school surround him and ask questions, and Matthew is just so loving about his brother. He's so proud of him, and he notices no differences. He always holds Mason's hand, the one with his finger missing, and he has not asked one question. He just kisses it and holds it, and he tells everybody that this is my brother. Greg and I have learned our new normal and learned to embrace every milestone. Mason is very delayed, so we celebrate anything that comes up, and it's exciting. Every day of his life is celebrated, and the love that Mason has brought to our family and to our extended family and people we don't know, plus the kindness that we've seen from this, is just amazing. We have my son's schools wanting to do fundraisers to help us give back to the hospital that has helped Mason. We get to throw these huge Christmas parties at hospitals, and everybody is just so kind with their donations and everybody wanting to help. It's so great to watch, and it's emotionally overwhelming with the kindness. With all the negatives that we were told, there's so much more positive. He's just the light of our lives. He's so happy all the time, and with everything he's been through, the fact that he's still able to smile and laugh every day really shows us that we can be just as strong as he is in this journey as well.

Gerald: That's super. What has his impact been on your extended family?

Kaitlin: My family is so supportive. When we had Mason and I wasn't able to see him and we didn't know what was going on other than he was just rushed away, I had to call my mom and tell her that we had him, but we don't know what's wrong, can you come here right now because I need some emotional support. They all came; they were all so supportive. The same is with his family. There are some family members who are still not understanding, or they think that once he hits a milestone, he'll be completely normal. We embrace that he's different; we love everything about it, and they are starting to as well. We don't expect them to understand everything as fully as we do, because sometimes I look back on how I got through all of this with such little knowledge compared to what we know now about his diagnosis, so I don't expect them to understand. If somebody sort of lets us down or isn't very understanding, it surprises us. We have to remind ourselves that they don't understand, and it's not their fault. For the most part, our families have been an

absolute blessing. My mom is pretty much the only one who can really help me watch Mason other than his dad, so she's been a Godsend as well.

Gerald: If I were to come to you having just found out that a child I had just borne has complex issues like Mason, having lots of complex issues going on and lots of different diagnoses, what advice would you give me?

Kaitlin: You are your child's voice, so do not be afraid to advocate or question things. Don't be afraid to ask questions. If you are able to find a family or support group, I would say get on that right away, because there's no worse feeling than feeling alone. There are more people going through this, and I've never once had a family turn me away from asking questions. I love questions, and I love being able to educate people on my son. So do not be afraid to ask questions and to reach out and really enjoy every single moment with him. There are so many moments where I look back and see that I was such a stress case during his first couple of months of life, when I wish I could have taken a deep breath, embraced what was happening and really enjoy that time. The days are going to pass, so enjoy every moment that you have with them, celebrate their life and give them the most love that they could ask for.

Gerald: What's your greatest fear with Mason?

Kaitlin: On his 11th chromosome, one of the deletions is a whole gene deletion called the SDHD Gene, and it gives him a much increased chance of hereditary cancer. It runs in my family, and when cancer is so relevant now, it scares me that he's going to have to go through that as well. We still have so many specialists that we have yet to see, and I just don't want him to have to go through anything else besides what he has already been through. I know we can get through it, especially him because he's such a fighter. I just need him here with me. That's my biggest fear.

Gerald: You've gone through a lot. You have a child with complex medical needs, and it sounds like he's one of a kind. There isn't anyone else out there quite like him. Besides lots of medical stuff, you haven't slept in the same bed with your husband for almost two years. You have him right by your side. Are all the sacrifices and all the work worth it?

Kaitlin: I don't even view them as sacrifices. A lot of parents come up to us and say, "Oh, man, if we were in your situation, there's no way we could do it." We look at them and say, "Yeah, you can. That's your child, and you will do everything for them." It's not really even a sacrifice. My mom always told me that you live your life, and when you decide to have kids, it's their turn. I would sacrifice my whole life to make sure they have their best life, so it's so worth it. I can't imagine my life without him or where we would be in this life without him. He's our light; he's everything to us, and he's everything to everybody around him, too. It's incredible to watch the people who come into contact with him.

Gerald: That's super. Any last thoughts that you'd like to share with the world?

Kaitlin: Be kind to everybody. The first time I heard the doctors say that he had dysmorphic features, the first thought that came to mind was that the world is so mean, so I need to think of ways to protect him. I feel like a mom shouldn't have to worry about that, so just be kind to everybody, be accepting, teach your kids about the differences and just spread love, just spread love.

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 and children with disabilities.