

An Orange Socks Story: Nicole- Single Gene Mutation

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, we learn from Nicole about her son Jacob, who has a single gene mutation. He is the only documented case with this mutation; that's right, he's the only one in the world with this issue. In addition to this gene mutation, he has tracheomalacia as well as developmental delay.

Gerald: Nicole, thank you so much for taking the time for an Orange Socks interview by phone about your son Jacob. How old is Jacob?

Nicole: Jacob is 15 months now.

Gerald: 15 months old, and he has a rare genetic condition. Would you call it a mutation or a deletion or how would you describe that?

Nicole: It is a single gene mutation. It's a part of a single gene that has that mutation. The problem with the mutation is that it's never been documented before, so we don't know if there is anyone else out there who has a similar genetic mutation as Jacob, so right now we are learning about Jacob as we go.

Gerald: So as far as we know, he is the only one in the world?

Nicole: Yes.

Gerald: Okay, and what else does he have? He has tracheomalacia and developmental delay, I understand?

Nicole: Tracheomalacia. When I was pregnant with Jacob, we found during one of the ultrasounds that there was a large cyst forming in the back of the brain that caused his brain to form differently. Because of that, we don't know the impact it will definitely have or not have on his development. We do know that he is quite delayed, but is it because he spent the majority of his life in the hospital, most of that in ICU, or is it due to the differences in his brain? We do know there is a difference in the brain stem that no one can quite put into words. It just looks a little different, so because of that, there are a lot of questions about how that will affect him. So yes, he has what we would call a developmental delay. He was having some apnea spells which caused him to be diagnosed eventually with tracheomalacia, where his trachea is very weak and was actually closing up on itself.

Gerald: Interesting. So you found out in an ultrasound that he had this issue. Were they able to treat this in utero?

Nicole: No, not at all. When we first found out, we were given some pretty grim predictions as to what it would be like. At first they didn't even think I would be able to carry to term. Then we found a second opinion and a third opinion, and with each opinion, things got a little bit better. Things that they didn't see in the first ultrasound, they were able to see in the second ultrasound, and as Jacob developed in utero, it started to get better. They weren't sure what the cyst itself would do or not do, and nobody could do anything while I was pregnant; that was something that we would have to deal with afterwards. We are just now seeing neurologists, learning as we are going, because this is definitely something new. When they look at the brain scan, they can't find one similar to Jacob's.

Gerald: That's very interesting. What have been so far some of the difficult things or the hard things that you have had to face with Jacob?

Nicole: I think the most difficult thing was learning about Jacob when I was pregnant. When you're first pregnant, you're very excited and celebrating with everybody. Then you're given this news, and you don't know what to do with it. I work in special education, so I have always said to myself that whatever happens, happens. I have a large knowledge base that I can start on as a parent and then build from that as I get to know my child. That's the approach that I took with Jacob. My husband and I both thought that we would be dealing with just developmental delays, but when I gave birth to him, they swept him off because he wasn't breathing. That started the crazy journey that we've been on with Jacob, where we've spent the good part of almost a year at the hospital. We've had multiple blue calls with Jacob, so we've almost lost him quite a few times. I think that part has been the hardest. No one can actually tell us why or what's causing it. Eventually we got the tracheomalacia diagnosis, but that still hasn't solved some of the apnea spells because he still continues to have them even after having major surgery to correct it. I think knowing his diagnosis is something that we can't put a name to has been the hardest part, because we don't know what will happen to Jacob. We don't know how long he will live or how long he will be here with us. Will he develop, will he not develop, will this eventually go away? We just don't know. Having to travel with oxygen equipment and suction equipment and having him on a monitor to track his oxygen levels has been quite difficult for us and has made our whole life change. Part of that, too, is you have a really good support base going in, and then when something like this happens, there are people who can definitely handle it and stand by you, and then you notice that there are people who can't. I think that's been really hard, losing some of those friendships. Family is great, family is always there, but I think you really start to see who your true friendships are. It has been really hard because you can feel quite a load at times.

Gerald: Tell me, what has been his impact on your family, your immediate family as well as your extended family?

Nicole: Regarding my immediate family, I am so lucky and we're so lucky. They just jumped right in. I remember calling my mom the day that we found out that Jacob was going to have some difficulties, and her just sitting on the phone with me while I was crying and telling me it doesn't matter, we love Jacob no matter what. I think from that day forward, that's all I've seen from my immediate family. My sisters jumped right in; they have come to my side at the hospital; even the days that I don't want them there, they are there any way, because they are going to be there to support us. One of my sisters has taken the liberty of getting trained to deal with all the equipment so that she can come in and give us a little bit of a break to take care of Jacob. Immediate family has been great, and extended family, too, the more that they get to know. The unfortunate part with Jacob is that we don't take him around too many people, because if he gets sick, then that's something that could cause him to have some breathing difficulties, so people are getting to know him more through stories and pictures rather than actually getting to see him. We hope that once he gets a little bit stronger and we feel a bit more safe, that that can change.

Gerald: Super, so tell me a little bit about the joys that you've experienced with Jacob.

Nicole: I don't know where to even start with those because honestly, for as scary and difficult this journey has been, it has probably been the most amazing journey for both my husband and me. We always say that if we were told this again, we would never change our minds because the joy that Jacob brings us is just one of a kind. We see the joy in Jacob watching him develop in ways that people never thought would happen. He's kind of defying the odds, and I think the other thing is that we can sit here and say we remember the first day he smiled, or we remember the first day he made babbling sounds, because those things are huge. I would say the other joy that Jacob brings is that he's teaching us to live in the moment, appreciate the small things, stop worrying about the big things, stop having expectations and just start to appreciate the little things that happen every day.

Gerald: So Nicole, if I came to you with a child that had a similar issue, what advice would you give me?

Nicole: I think my greatest piece of advice would be not to have expectations. I think you need to let your child be an individual and allow them to develop in their own time, in their own ways, whether it be developing physically or not. I know that has been a real challenge for us. We've connected with other parents who have had little bits and pieces of what Jacob has. I think it's important to live in the moment honestly. Appreciating the little things in the daily victories are what get me through every single day. Once you can learn to let go of having those expectations and just being in the present, you'll see how amazing your life is with this little one.

Gerald: You've spent how much time in the hospital with him?

Nicole: When he was first born, we spent a good three weeks there in the NICU, then in the pediatric ward while we learned how to feed him. Jacob was NG-fed in the beginning of his life, then we had to learn how to feed him orally and he had to learn to eat orally, so that took some time. Then we came home for a little bit, and then he had his first apnea spell, which landed us back in the hospital. We were there at that point for a month, then home for Christmas, and then back in January. We didn't get back until July of this year, so on and off, we spent a year at the hospital.

Gerald: Wow, so a good chunk of his life he's been in and out of the hospital.

Nicole: A huge chunk of his life, and we always say that the people there are almost his family. We've developed amazing relationships with the doctors and the nurses and all the staff who work in the hospital. He's well-known there. Jacob has a great head of hair, and when people walk, they say, "There's Jacob and the hair!" They know Jacob, they all pray for Jacob and they are all rooting for Jacob, and I think they've become our family because we've had to be there for so long.

Gerald: With all the hassles and difficulties and the hospital trips and all that stuff you've had to go through for the last 15 months, is it worth it?

Nicole: Absolutely. I wouldn't change it for the world. I have the most amazing little boy, one of a kind. I can honestly say that. He literally makes me the happiest mom in the world. I am the strongest mom, and I've become a way better person because of him.

Gerald: That's excellent. Anything else that you'd like to add or say for this interview?

Nicole: I just think that coming from someone who had to deal with a new diagnosis and didn't know anybody who was in a similar circumstance, that getting out there and reading some blogs was important. Don't feel afraid or intimidated to reach out to somebody if they've touched you or you have a question. I found that to be so helpful to me. I would read blog after blog, and when I would come across somebody who was in a similar circumstance and read their story, I would send them an email and ask them for their advice or ask them how they dealt with something. Talking with a parent who's going through it is the best piece of advice I can give. The other piece is that my girlfriends who I've met through hospital stays have developed a really great relationship with me, and we call ourselves "our tribe." We can literally message, email or call each other, and we all live far away from each other. We can say, "Hey, Jacob has a G-tube." I would call them and say that Jacob has pulled out his G-tube again. That could be the end of the world for me that day, and they totally understand it. Surrounding yourself with people in similar circumstances is super helpful.

Gerald: That's super. I really appreciate your taking the time to talk with me about Jacob. I'm looking at his photograph; he's a beautiful little boy. He does have quite a head of hair.

Nicole: He does.

Gerald: That's great, and I'm very envious because I have no hair. I'm a bald guy, so good for him. Anyway Nicole, thank you for taking the time to speak with me this evening. I really appreciate it, and I've really enjoyed learning about Jacob and his extremely one-of-a-kind rare condition that doesn't even have a name. That's very interesting, and I thank you very much.

Nicole: Thank you.

Gerald: As we chatted after the interview, Nicole mentioned that at her 18-week check-up, once some of Jacob's issues were discovered, her attending physician gave her the option of terminating the pregnancy. She and everyone else who has come to know Jacob is very glad she did not listen to that advice. Thanks for listening to this episode. Orange Socks is an initiative of Rise Incorporated, 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.