An Orange Socks Story – Katie and Nathan: Rhizomelic Chondrodysplasia Punctat, or RCDP

Interviewed by: Gerald Nebeker, President of Orange Socks

Gerald: What an honor it was for me to meet with Katie and Nathan about their daughter Claire, who has Rhizomelic Chondrodysplasia Punctata or RCDP, a very rare and fatal condition. Claire is the first person I've met with RCDP, and she is wonderful. Katie and Nathan's love and enthusiasm for their daughter is very evident, and their positive attitude is compelling.

Gerald: When did you find out that your daughter had a disability?

Katie: At my 20-week ultrasound, we found out Claire was a girl, and then they said that her arms were measuring a little short and suggested we come in for a follow-up four weeks later. We went back four weeks later and we didn't even think anything about it as first-time parents and especially if the doctors are saying "I'm sure it's nothing." At 24 weeks, they said, "I'm really sorry, but she definitely has a form of skeletal dysplasia which is dwarfism."

Gerald: That's when you knew. What type of counseling did they give you?

Nathan: Honestly, it was a little lacking. The ultrasound tech was acting a little funny, and so I thought something was up. She disappeared, and the doctor came back with a…

Katie: … a big book.

Nathan: It was a big textbook, and he said she has some form of skeletal dysplasia, most likely a chondrodysplasia. Here is a print out about it.

Katie: Yeah.

Nathan: They left us on our own for a little bit to kind of take it in, and they offered to set us up with counselors and things like that, but initially there wasn't much said.

Katie: It was more like, here you go, here are some pieces of paper, sorry, enjoy looking it all up online yourself.

Gerald: Interesting. So what were your thoughts?

Nathan: It was a pretty big blow. It was more just shock there for a little while, and then a lot of tears, then kind of being together just trying to understand the paradigm shift of a sudden knowing that everything was going to be different.

Gerald: What were your thoughts, Katie?

Katie: I totally agree with Nate. I feel like you get married, you have a family, you have kids, you become grandparents, and life done. Then I feel like you find this, and you're like, okay, everyone is on the same path and all of a sudden, I felt like I had to take a hard right, and you kind of want to be on that other path, like this is not what I planned, this is not what I expected. "What did I do?" goes through your mind, and you think. "Did I eat something funny? Was I mean to someone?" It's totally irrational, but you go through those thoughts. Am I getting punished? Is there something I could have done to make this different? It's just really scary because we've never gone through anything like that before.

Gerald: Sure.

Katie: I have some friends with special needs, but I think it's different from having a friend to being a parent, and when you don't know and something is that rare and that people don't have, I feel like if they had said something like your child has Down syndrome, then we would have known what that is. With achondroplasia, it's like, what is that, and then on top of that, they said we think it's that but we can't be 100% sure. Then, of course, you spend hours and hours looking up things online, and you're just kind of lost in this big haze; you don't know what's going on.

Gerald: Sure.

Katie: You're just trying to get back to the path, and you just know you can't.

Gerald: Right. So she was born, and she has equipment that I'm seeing, so why don't you tell me, Katie, what are some of the hard things or the challenges that you have in working with Claire or in taking care of Claire?

Katie: I have to think about this for a second. There are a lot of things. It's kind of hard emotionally, and I'm sorry, I don't even know why I am crying. It can be hard to take care of a child who you know doesn't have long on this earth. You just want to give them the best life they can have, and you want to love them, and you want to give them so much joy. When we found out that Claire had this terminal dwarfism, this Rhizomelic Chrondrodysplasia Punctata, my first thought wasn't, "Oh, she's disabled" or "She has a learning disability or a physical disability," it was "We just don't have that long with her," so it's definitely a really heavy emotional toll. I would never trade it for anything. I feel like if I were to put all my trials in a basket of my whole life, I would pick Claire every time, because she is not a trial; she is just a great joy. Yes, it's hard. For medical things, I've learned how to place NG tubes and suction machines and feeding, but in the end, it doesn't matter. In fact, I think Nate and I have talked how I forget that kids don't eat through feeding tubes, and I forget they don't need suction machines, because that's just our normal. Our normal is just taking care of Claire that way, and that is fine, and I would take care of her 100 more years. I don't care much about medical stuff, but it's definitely a more emotional toll than anything. You always hear that comparison is like the thief of joy, and I think that's true. It can be true that my friends had kids around the same time as Claire, and there is an obvious difference, but like I said, I just would never trade it. I don't think people whose kids are different would say, "Oh, yeah, I would give up my kid in a second." I wouldn't trade that for the world.

Gerald: Nathan, what's the hard part for you?

Nathan: Other than, as Katie already mentioned, the emotional weight, just knowing that there is something that is going to happen that's going to crush you and not knowing if you're going to be able to come back from it. It's obviously the big one, not knowing. I don't know how I'll cope with it, but I try not to think about it because otherwise I can't function. I start to go to a dark place, and I don't want to be in a dark place because I want to enjoy her now.

Gerald: So her condition is a fatal condition.

Nathan: It is.

Gerald: What's the typical life span?

Nathan: 40% of the children with her condition don't make it to their first year; very few make it to the age of 10.

Katie: A child who lives 5 years is considered a long-term survivor.

Gerald: Katie, describe for me your daily routine.

Katie: Is it a good day? Is she okay in the morning? Is that what you mean?

Gerald: Just describe a typical day.

Katie: Some days, if she's already kind of upset, that throws the whole day off . You wake up, Nate goes to work, and I have my alarm set about 10 minutes before her feeding, so I'm groggily getting out of bed to go to the bathroom to take care of all the "me" stuff I need to take care of in the morning. Then I go downstairs and mix her formula. I bring it upstairs and prepare her feeding bag and her pump and all that, and usually if I'm lucky, she's still asleep by the time I start, so I'll set that up, hook her up and then if she is still asleep, I'll go grab breakfast and come upstairs. She is usually really upset already and trying to wake up. With her condition, she produces a lot of secretions, just crazy amounts of secretions, so we have our big suction machines, and I'll spend 10 minutes just suctioning her out. You cannot believe how many boogers this kid produces; it's just crazy. I take her out of her crib and wash her down because she doesn't have much mobility, and she tends to get sweaty at night, so I make sure I get her armpits and under her legs, the places that she is not really moving because it's just build-up. I make sure she is clean and get her dressed for the day, and we come down the stairs, and she'll usually nap for a little bit, which is great because then I can get dressed. I think it's important that you need to take care of yourself for a minute, and then we'll start her next feeding, and during that. we'll start to do play time, or I usually read her stories and then I read her books, and then we'll do at-home physical therapy for the next hour and a half. Because I can do it at my home, it can be spaced out versus when we go to therapy. We can do all the really hard-core stuff in an hour, and she is exhausted, so I stretch her out to work on her fingers and her movement. We roll her around and make her neck move to side to side because she just does not have a lot of mobility at al,l so we'll do that and maybe she'll nap again, maybe not. That is where the tricky part comes in; I feel like any mom doesn't always know when their kids are going to nap, and that's just the joy of unpredictable children. As the day is going on, we're like suctioning and suctioning, , we're feeding and making sure everything is okay, and on occasion I'll call a doctor or supplies to be reordered for her stuff and make sure that is all clean and nice. I feel like I'm cleaning a lot because with a medically fragile child, you just have to make sure your house is clean because you just don't want to bring in any germs or anything. Oh, my gosh, what do I do all day? I know I'm busy all day, and now saying it all out loud makes me feel like I don't do anything all day.

Nathan: On a rare occasion, you'll get a walk.

Katie: On a rare occasion, we get a walk, and the weather is starting to get nice, so we can venture outside, but in the winter, we just keep her inside; we've had gorgeous weather recently.

Gerald: Sure and you are busy all day.

Katie: I know, and well, she really can't be left alone.

Gerald: Sure.

Katie: I go to the bathroom with the door open just in case I hear her coughing or gagging, and you gotta go take care of that. Then you gotta get dinner ready at some point and remember to eat yourself, and that only rarely happens. It'll be three o'clock, and I'll wonder why didn't eat lunch; I just I missed it. I don't even know how that happens. Then more physical therapy, laundry, cleaning again, taking care of bills; my day is so boring. I need to find an activity or something. I'm listening to it out loud, and oh, my gosh, I love being with her, but that just sounds so repetitive.

Gerald: It actually sounds pretty normal with a child who is medically fragile.

Katie: Yeah.

Gerald: Nathan, let me ask you maybe an easier question - what are the joys?

Nathan: That's way easy. It's just holding her and trying to get her to smile or laugh. In the last few months, she is much more quick to give a smile than she was before, so playing with her is definitely the best.

Gerald: Yeah, that's great. How about for you? What are your joys?

Katie: I agree with Nate. I think just watching her grow has been crazy. I knew that I wanted to see my kid grow, but for us, with a child who has a lot of disabilities, just seeing the small things she does is monumental and brings you so much happiness. She was born with cataracts in both eyes, and we got them removed, and just seeing her see clearly for the first time was really joyful, and even now, I love seeing her looking and playing. I'll put on a little sensory show for her, and I love seeing her eyes light up and her laughing at that, and I just love being with her and making memories with her.

Nathan: I'd like to chime in. One more is when Katie tells me her favorite sleeping place is when she is snuggled up on my chest.

Katie: It's true.

Nathan: I love when she takes a little nap on my chest, and I just stroke her head, and we don't do much, maybe watch TV or something like that, but I love having her just snuggled up on me.

Gerald: Katie, tell me what impact has her life had on your extended family?

Katie: After Claire was born, I remember my sister said something, and I thought, "Wow, that is exactly how I feel!" Now that Claire is born and she has all those problems, Steph told me I realized how much things don't matter, and she didn't mean that her life didn't matter, just that Instagram doesn't matter, Facebook doesn't matter, and it doesn't matter that my house isn't perfectly clean. She just realized what actually makes you happy in life, and I think you know that's been a big one. I think you know that certain members of the family were a little uncomfortable with Claire's condition; they were a little standoffish. "I don't want to be around a child who is terminally ill," not because they didn't love Claire, I just think they didn't know how to react, and they didn't know all these things. I remember we had gotten letters from some people, and it was shocking how many people said they were sorry, not congratulations, you just had this baby. I feel like you have people in both camps; you have people who were kind of uncomfortable and kind of standoffish who didn't know what to say or what to do, and then you have people who were totally cool with it and were happy for us; they didn't care.

Gerald: How about your side, Nathan?

Nathan: It's amazing not necessarily what they did, but how my mood would be impacted by how I would interpret something like a kind gesture saying they were sorry, that it is unfortunate that happened. Obviously, everyone was trying to do it with love, but some days you'd really not want their pity. This is how it is, and other days, it would be thank you very much, it's fine. A of the time it was how I was doing on that day some days it was just very easy to explain everything going on, and other days it was just emotionally difficult to explain it, especially in the beginning. Iit was much more of a roller coaster of how I would interact with the world and how it is now. It's a little more stable, and you know there are still ups and downs, For the most part, they took it well though some definitely were at odds with her; you could tell they were walking on ice, and they knew they were walking on ice. and then others were just wondering whether they were going to smash the ice or not. They didn't care; they were just going to do it. Ultimately, that's who they are, and ultimately it was actually almost a little easier with their staying true to their character rather than trying to walk on ice or on eggs shells all the time.

Gerald: I am wondering if I came to you at my 20-week pregnancy and had a similar diagnosis as your daughter, what advice would you give me?

Nathan: It's going to be okay.

Katie: Yeah, oh yeah.

Nathan: It's going to be hard, but it's going to be okay. You'll have a lot of hardships, but there are going to be so many joys so enjoy what you have now, and it will work out; just keep going.

Katie: I totally agree. When you first find out the diagnosis, you probably think that matters a lot right then, and in some respects it does and in some ways it doesn't. You're welcoming a child into this world who is loving and whose life matters, so yeah, things are going to be a little different and things are going to be scary, but you're not going to love your child any less. That was something that I struggled with, not that I thought I wasn't going to love Claire, but I honestly thought, "Oh, my gosh, is my child going to have this disability and is all I'm going to see is their disability and I'm going to see the disability first and the child second," and that doesn't happen. I wish I could go back and tell myself that my child has a disability and that's it. You'll forget some days because Claire is Claire; it doesn't matter. I wish I could go back and tell myself that or tell someone who has the same condition that you are going to have such a great time; they will just be your whole world.

Gerald: Claire was diagnosed with Rhizolmelic Chondrodysplasia Punctata at 24 weeks, and their doctor apologized to Katie and Nathan for not finding it out sooner so they could have decided to abort Claire, something Katie and Nathan would have not opted for anyway and is a horrifying vision now to them as they love and care for their precious little girl.