An Orange Socks Story - Katherine and Jeff: Ellis-Van Creveld Syndrome
Interview by: Gerald Nebeker, President of Orange Socks

Gerald: I was honored that Katherine and Jeff would take the time for an Orange Socks interview to talk about their experience with their daughter, Bella, who has Ellis-Van Creveld Syndrome, a very rare condition. Theirs is an interesting story because Bella received two diagnoses in utero, conditions where doctors advised them to abort Bella because the conditions were fatal. Had they followed that guidance, we wouldn't know Bella today, who is a delightful two-year-old. Both diagnoses were wrong. You can follow Katherine on Instagram and her blog at Kathinchina.com.

Gerald: Katherine and Jeff, thank you so very much for taking the time to meet with me. When did you find out that Bella had Ellis-Van Creveld Syndrome?

Katherine: We didn't find out, actually, her specific diagnosis until a couple of months after she was born, but it was at our anatomy scan. Going into that, we had one healthy child already, and the anatomy scan is when you find out if it's a boy or girl, and you don't really think about the other things. We had that scan at about 18-and-a-half weeks, and as we did the scan, I noticed that the person scanning kept going back to Bella's long bones and her arms and her legs multiple times. I could see the number on the screen was about two weeks behind, so we could tell something was up. The tech had been really chatty before those measurements and really friendly, and all of the sudden she got really quiet. Like any parent would, we asked how significant is that two weeks, and she said, "You know, your OB will call you and discuss the results with you." She was very quiet the rest of the time. We finished the scan and went right outside the office to the little patio table and immediately pulled out our phones and googled because that's what we do these days. We were looking up limb difference, and there were a lot of different, scary possibilities that popped up. My husband had to head back to work, and I had to go pick up our older daughter, and we knew we'd talk to the doctor later, but we were fortunate in that one of our good friends is actually a radiologist. I texted him and asked him to look for my scan and read it. He gave me a call after he read it and said, "You know, that is a significant lag in those bones, probably not just that she's small. There is probably something going on." He mentioned a few possibilities and the fact that there'd probably need to be further testing. My husband and I were actually heading out the next day to go on a little "baby-moon" trip, the last trip before having a second child in our home. We were about five minutes from our destination when we got the call from the OB, and she said, "I want you to come in and have your blood drawn, and we want to do some testing to see if we can see anything just from the blood work." They were testing for the most common forms of trisomy at first, so when we got back from the trip, we did that, and all of those results came back negative. They sent us to a bigger city about three hours away to go to a university hospital to have a more detailed ultrasound and to get further information. When we went for that scan, both of us were really at peace going into

it. We knew a range of possibilities, and we had talked a lot about it and about the fact that we could be having a child who would be with us for the rest of our lives, and we were just eager to find out what was going on with her.

Jeff: Although we were open to a wide range of possibilities, I don't think either of us really expected an outcome where she wouldn't be with us.

Katherine: No.

Jeff: So that wasn't our expectation. There might be some challenges, but nothing we couldn't overcome and certainly nothing that would be fatal.

Katherine: So, we went to that scan, and the tech was amazed by how active Bella was because she was doing acrobatics throughout that whole scan. At the end of it, she said, "I'm going to go talk to the neonatologist and see if we need any more pictures." She came back to the room after that discussion, and once again, her cheerfulness had disappeared. She told us, "I think we have a good idea of what's going on. You'll get to talk to one of the doctors a little bit later." We had an hour or two break, and Jeff was convinced that it was dwarfism, saying he thought she was going to be a little person. During that break, we talked about that, and we talked about how we were okay with that. This was something that we could face as a family. The first person we met with after the scan was actually the genetic counselor. I've learned that with the doctors, usually their first question is "What do you know about what's going on?" They make you go into it first, which always drives me nuts, because I just want to get to the point. Jeff dutifully told his guesses about what he thought was going on, and then the doctor's first sentence, which I'll never forget, was "Your baby is not doing very well." To us, that was such a shock, because we had just watched her dance across the screen, and she was so full of life that to hear those words was just very jarring. The doctor went on to explain that not only were her arms and her legs measuring shorter than average, but her thoracic cavity and rib cage was much smaller as well. This had led them to a diagnosis of what's called thanatphoric dysplasia, which is a form of dwarfism, but it's a lethal form of dwarfism. She told us that there was a 99.9% chance that Bella's rib cage would not grow much, if at all, beyond this point, and because of that, when she was born, there wouldn't be sufficient lung tissue, and she wouldn't be able to breathe, so we would not have much time with our daughter. Then kind of as an afterthought, she mentioned "I don't want you to see this in your charts later and wonder why we didn't tell you, but she also has a heart defect, but that's not going to be what ends up being lethal for her." At that point, I think we were both a little stunned, and we asked her, "What does this mean going forward, what does this look like?" She responded "Well, since this is terminal, most parents will choose to terminate the pregnancy." We told her right then that was not going to be our choice. We were not going to terminate. It really never was an option we considered. She said "Okay," and she got some family history, and then we met with the doctors and went through all the details again where they talked about her basically suffocating after she was born because of not having enough lung tissue. I asked one

of the doctors, “With the kids that you’ve seen born with this diagnosis, how much time did they have?” I was trying to picture that day when she was born and know if we were going to have minutes with her or hours. She said, “I can’t personally tell you that because I’ve never seen a live birth with this diagnosis.” That led me to the question, “Do they usually pass away before they’re born?” She said, “No, it’s just that every single parent given this diagnosis has chosen to terminate.” That was heartbreaking and a little shocking to hear, but one of the head doctors came in soon after that, and he talked to us as well. It became very clear, very quickly that he didn’t agree with our decision to continue. I don’t think that was out of any ill will or anything like that, but he saw this baby, and he knew there was nothing he could do to save this baby. He was trying to give us what he thought was the least-painful path going forward, which he thought would just be to stop it at this point. He even mentioned to me, “Think of your other daughter at home,” who was about two at that time, “and think of how emotional you are and how emotional you’re going to be the next couple of months, and how hard that’s going to be on your daughter.” He said that we needed to think about her, too, in making this decision. He looked at me and said, “I know today you think you’re not going to change your mind, but you’re going to change your mind. We’ll see you back here, and you’re going to change your mind. Just so you know, you don’t have a deadline because this is a lethal diagnosis. We can terminate at any point, so you don’t have to decide by a certain week in your pregnancy make the decision.” He said, whenever you do make that decision, we’ll be here, and we’ll be able to do it.” It was a very hopeless situation that was presented to us, but we went home, and both of us agreed that there was no chance that we wanted to terminate this. We knew that we might not get much time with her, but we wanted to give her any chance she had. We didn’t choose the day she was conceived, and we refused to choose the day that she would die. We decided to continue, and that meant that we had to head back to the university hospital every month for check-ups and ultrasounds. Four weeks later, we went back, and they took the measurement of the thoracic cavity, and it had grown four weeks’ worth of time. They had told us that it was not going to grow at all, and if it grows, it’s going to be very insignificant. We were meeting with the doctors after that, and I remember my husband looked at the doctor and said, “Does this mean there is hope?” She said, “I’ll give you a sliver of hope.” We knew that good things were happening. We went back another month later, and sure enough, her rib cage had grown once again in a month’s time. This rib cage that was lagging with her arms and legs all of a sudden flipped around and shot off, and that was the one thing that we were asking people to pray for with her. She had short arms and legs that weren’t going to determine the course of her life, but it was the rib cage that we needed to grow. The name we chose for her was Arabella, which means striving in prayer and beautiful altar. We felt like we were giving this daughter up to God and saying, “Do with her as you will,” and asking people to strive with us in prayer. We were watching this rib cage grow unexplainably, and the second time that it grew that much, they told us they didn’t think their initial diagnosis was correct. They didn’t think it was TD anymore, but that meant searching for a new diagnosis. They had a conference with the doctor, and they weren’t really sure what they were looking at because it wasn’t fitting into any of the more common forms of dwarfism.

There are a lot of different forms of dwarfism, and she wasn't really checking the boxes for the ones they were looking at yet. We kept going back, and it kept growing, and then we headed back about a month before she was born. We were having an echo on her heart on the grown scan, and it was at that appointment when we sat down in the conference room and the doctors said, "I think we figured it out. I think we know what she has. We're thinking that she has Short rib polydactyly syndrome." They thought they saw an extra finger and an extra toe on the ultrasound, and that the shape of her rib cage seemed to point to that. It was at this point they told us that Short rib polydactyly syndrome once again is 100% lethal. We had moved away from the first lethal diagnosis where we had hope growing and thinking our daughter had a chance, and then a month before she was born, we were given a second lethal diagnosis. How did you feel with that one, honey?

Jeff: The very fact that they had misdiagnosed the first go-round gave us some confidence that they may have misdiagnosed the second, so while it was hard, and we had no scientific basis for hope at that point, we had hope. We went into delivery day with some hope, recognizing that she would probably have challenges even if she did survive, which was something they told us she probably wouldn't. Even if she did, she would almost definitely have to go to the NICU and probably stay at the NICU for quite some time.

Katherine: Going into delivery day, they told us three ways that this was possibly going to look. The first is after she's born, we take her away to assess her. If it's clear that she is really struggling to breathe and intervention isn't going to help her, we'll bring her right back to you, and you will get to spend as much time as possible. That was what they presented to us as the most likely of outcomes. The second was that we take her away and have her assessed, and we are not sure if assistance will help her, so we'll take her to the NICU and try to give her some breathing assistance and assess whether or not that's going to be successful in helping her. If at any point it looks like this really isn't going to work, once again we bring her to you, and you get to spend time with her. The third and least likely outcome was after she's born, she needs some assistance, but it looks like she can make a fight for her life, and so she stays in the NICU. That was the possible picture that they gave us. My husband had researched burial plots, and I had researched and found a foundation that sends burial gowns made out of old wedding dresses, and though we had hope, we were also prepared for the possibility that we would not bring our daughter home. She was born on a Sunday around 12:30, screaming her head off, which I knew immediately was a really good sign, because you have to have some lung tissue to scream, but I had also read enough stories of moms who had lost their babies that also screamed when they were born, so I knew it wasn't this guarantee that she was going to be okay. They took her right away for assessment, and my husband and good friend who is a photographer left the room with her to go to the assessment room. I was left there by myself with all of the fun post-birth things happening to me. I waited a couple of minutes and I asked if someone could please go check on my baby and tell me how she's doing. They sent someone over to the assessment room, and she came back a few minutes later and said, "Your daughter scored nines on her

APGAR. She's doing great, and they are going to bring her back to you in a couple of minutes." They weren't bringing her back to die, they were bringing her back a very pink, very much breathing, beautiful baby girl. The next thing I asked was, "How many fingers and toes does she have?" which is the perpetual joke when you have a child, but since that was the thing that pointed to this one diagnosis, I was actually quite curious as to how many fingers and toes she had. The nurse said, "I'm not sure, you're going to have to look when she gets here." Sure enough, she had six fingers, but only five toes, so she was brought back to us within 5-10 minutes, and she didn't leave us the rest of the time she was there. She never went to the NICU, she was on the regular maternity ward, and three days later, we headed home with her, with no oxygen and no monitoring technology, just a miraculously breathing baby girl. We had to constantly pinch ourselves that was actually happening, but then about a month or two after she was born, we met with a geneticist who said that they were never quite confident in their diagnosis of Ellis-Van Creveld, which turns out to be a subset of Short rib polydactyly syndrome, how many, four?

Jeff: There are five different kinds, and only two are compatible with life. Ellis-Van Creveld has the highest probability of living, I think it's somewhere between 30% and 50% fatal. Ellis-Van Creveld has much better odds than the other alternatives.

Katherine: I think the wide range of lethality is because there are so few cases in the world that they don't have a precise number.

Jeff: As I understand, there are a few hundred cases of Ellis-Van Creveld.

Katherine: Worldwide.

Jeff: It's quite rare.

Katherine: Yeah.

Jeff: So, Katherine has to carry the gene, and I have to carry the gene, and it's very unlikely for us to carry the gene and then for it to manifest itself, we both have to contribute.

Gerald: Any family history or anything?

Katherine: No, it has never shown itself in our family, which shows you the rarity of actually matching up the two, but it's auto-recessive, so if you do both carry it, you have a one in four chance.

Jeff: Katherine is pregnant again with our third child, and we found out that she likewise has Ellis-Van Creveld.

Gerald: And they are sure about this diagnosis?

Katherine: They are sure about this diagnosis, because of it being auto-recessive, she has the six fingers, she has the heart defect that is very similar to her sister's, and when we went into our anatomy scan with Shiloh, our third daughter, as soon as we saw the long-bone measurement in the arm pop up on the screen, we knew we had another baby with Ellis-Van Creveld.

Jeff: Okay, we are going to do this again this is yet another journey, but in this case, we already kind of knew what that journey would look like.

Katherine: But we knew when we chose to get pregnant again that there was the possibility.

Jeff: Probability.

Katherine: We were okay with that. When we talked to doctors after Bella was born, they wanted to make it really clear to us that this is auto-recessive, and we had a good chance of having another child with this. They said we could do artificial insemination, and they could test the egg to see if the baby had Ellis-Van Creveld and not implant those who had it, and neither of us wanted to go that route if you look at Bella.

Jeff: Yeah, we would never trade Bella, she's great. She screams at night, which is kind of annoying…

Katherine: She's quiet now.

Jeff: She does annoying things that most two-year-old's do, but she is wonderful. We wouldn't trade her for anything. She is a joy and a delight and it's fun to watch her and her older sister interact.

Katherine: To try and avoid what she has is just isn't something that we would do even knowing the risks. And so, with this baby, we don't know what delivery day is going to look like, and we don't know what the months following are going to look like.

Jeff: Bella is going to have heart surgery shortly after.

Katherine: Shortly after Shiloh comes.

Jeff: Here in the fall, so there are a number of different variables. The reality is that when we were going in to Bella before we even had that first ultrasound, if they had told us that these are the challenges that you're going to have with her diagnosis, we probably would have been very frightened about what that might look like for our family, not that we wouldn't choose that, but it obviously presents hurdles. We've had so much fun that we don't look at them as hurdles necessarily, we look at them

just as life, and those are a journey. They are just the things that you have to deal with.

Katherine: I think that once you're told that your child isn't going to live, then everything else pales in comparison. Whereas before we were pregnant with Arabella, to find out our child had a significant heart defect that required open-heart surgery maybe months after birth or within a couple of years after birth, that probably would have been something that terrified me. Whereas when they told us Shiloh had a heart defect, the only thing I really cared about was is it one that can be repaired. It wasn't even that scary to face, because the child has life, and that's really all that matters, the other things you figure out as you go.

Jeff: You don't need to figure them out on day one, just figure them out over time.

Gerald: So, Bella had essentially two fatal diagnoses before she was born, and you were strongly encouraged to end the pregnancy because of various reasons the doctor gave you. Have you ever thought about what if you made a different decision?

Katherine: I have thought about if we had made a different decision because TD, the first diagnosis, is not auto-recessive, it's a random genetic mutation, and so we likely would have tried to get pregnant again. Then if we got pregnant again, and if the same sorts of things showed up, we'd probably start to question if this really was random genetic or something else going on. Then if we had continued with that pregnancy and that child had lived, I can't even imagine what we would have felt like, knowing if we had given that first child a chance, she could have made it.

Jeff: We are blessed at a time when we have ultrasounds and access to information, but ultimately, they are just guesses, and we don't want those guesses to get in the way of life.

Gerald: Tell me about the impact that Bella has had on your immediate family and friends. I mean, she's obviously delightful, as is your other daughter. I'm sure your family grieved with you during the dark diagnoses, and then she was born so robust. Tell me a little bit about what impact that has had on your family.

Katherine: She has had a huge impact on actually a huge number of people. After we received that initial diagnosis, I started blogging about her. I had a blog from when I lived in China, so I picked it back up and started telling her story, mainly because it was so hard to tell it again and again to every person. I wanted one place where people could go and read updates about her so I wouldn't have to go through the motions of saying things over and over again. That blog ended up getting shared far and wide, and so Bella's story went far and wide to people just being amazed at what happened that really defied expectations. I think the day she was born there were something like 100,000 hits.

Gerald: Oh, my.

Katherine: On the blog that day, we could feel this great multitude of people who were making her name true of striving of prayer and then rejoicing at the fact that she lived. In fact, our local newspaper even did a story on her. It was a front-page story of this baby who defied the odds which we just saw as an awesome opportunity to do what Orange Socks is doing, simply spreading awareness that things can end differently than what's expected. We don't know with certainty, so give life a chance. Our families, of course, were super encouraged by it. There is something special about Bella that wherever we go, people are drawn to her. It's something that you don't see that often, but we are constantly getting stopped because she has this way of locking eye contact with people and waving at them and engaging with them, and you watch her just spread joy. One day I was at the grocery store, and one of the people who worked in the floral department pulled me aside and said, "I watch you guys come in every single week, and I watch how the faces of people around you change every single week when they see you." She just has that magnetic quality to her, and we are getting to the point where people are starting to realize she's really small for how she's acting. At first, they would think that she is just an average baby, but now people are saying that she's really small, so we say she has a form of dwarfism, and they actually didn't think she was going to make it, but she's thriving. Just the opportunity to spread her story wherever we go is really, really cool.

Gerald: That's great. Jeff, if I were to come to you just having received a diagnosis that I had a child a with Ellis-Van Creveld, what advice would you give me?

Jeff: Enjoy it, soak it up and recognize that it is going to come with some challenges for sure, so get that support network around you. Bella is a joy, and at this point, we don't even see Bella as any different than her older sister. At times, I tend to forget because she acts as any child her age would act. It's only when she gets a really bad cold that we recognize then she gets into a state where she has pneumonia-like symptoms. Her lungs and her heart are not like everybody else's, but it's easy to forget because she is a joy and really is like every other child.

Gerald: And she has a heart issue that's going to be worked on. What specifically is it about her heart that they need to repair?

Katherine: When she was first born, she had both an ASD and a VSD, so two holes in her heart. VSDs are more significant, and hers was very small and actually has closed itself on its own, but her ASD, the hole between the wall and the two chambers of her atrium, is quite large.

Jeff: She almost has no wall, it's what they call a common atrium if you don't have a wall.

Katherine: Hers is not.

Jeff: Hers is not quite a common, but it's very close.

Katherine: Shiloh, our second daughter, has a complete common atrium, she has no atrial wall, so it's just one.

Jeff: Three chambers basically.

Katherine: Three chambers, and then in addition to that, the valves that are going from the atriums to the ventricles are different than the normal heart anatomy. It's almost like they have a singular valve was how the cardiologist explained it to us. There is some banding of tissue in the middle of it that allows those valves to operate separately, which is probably why Bella has been able to do so well up till the age of two, but the valves will also get repaired.

Gerald: And they think they can do pretty good with that, that's awesome.

Katherine: Yeah, they think she'll need open-heart surgery, and it's a significant open-heart surgery, but it's one that they think has a very high likelihood of success, and she probably won't need another surgery after that.

Gerald: That's great.

Katherine: She's slated to have heart surgery this October, depending somewhat on how Shiloh is doing. If Shiloh is really struggling, then we'll probably postpone it, but they'd like to get her in before another cold and flu season, just because those tend to hit her hard.

Gerald: Katherine, you get the final word. What final word would you want to give?

Katherine: My final word is give your baby a chance. We wouldn't trade what we have been through for 10 healthy babies. We think we are some of the most blessed people to have been on the road that we have been on, and it can be really scary and it can be really stressful. I know that right now as I'm about three weeks away from giving birth, and really not knowing what the outcome is going to be, you surround yourself with good community and sit back and watch what the Lord might do. It can be pretty incredible to see. It'll give you a perspective on life that you wouldn't want to trade for anything else.

Gerald: Wonderful, you guys are awesome. Thank you for taking the time to meet with me and to tell your story, it's very inspirational.

Jeff: Thank you.

Katherine: Thank you.