

An Orange Socks Story - Katherine and Jeff: EVC Follow Up

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. I interviewed Katherine and Jeff previously about their second daughter, Bella, who was given a prenatal terminal diagnosis. Katherine and Jeff are in some ways poster children for Orange Socks. Their story is about how deciding to carry your baby to term can turn out very differently than what the doctors predicted.

Katherine: The first appointment where they diagnosed her with a lethal form of skeletal dysplasia, we were strongly encouraged to terminate the pregnancy. We were told that no other parents who were given this diagnosis had carried to term. We were also told to consider the emotional effects that carrying this child might have on our older daughter since knew Bella was going to die. We left that appointment telling them, "No, we are going to give this baby a chance to fight." They told us, "We'll see you back in a couple of weeks you're going to change your mind." They realized they were wrong in her diagnosis when her ribcage actually did grow and they didn't expect that. They then gave us another diagnosis, but then that second diagnosis was also lethal.

Jeff: Bella's heart defect was almost placed to the side of importance. The doctors kind of approached it like, "Just so you know, she also has a heart defect. Not that that matters because her diagnosis is lethal."

Gerald: But the doctors were wrong twice. After Bella's birth, she was diagnosed with Ellis Vankerfeldt or EVC, which is extremely rare.

Katherine: She is now almost 3 years old and she has had her heart repair surgery. Like most things that Bella has gone through, she excelled through that surgery and excelled through the recovery. She was winning hearts marching the halls of the hospital within a couple days of that surgery. More than the physical effect of it, it was harder for her to overcome the emotional effect of the scariness of surgery. But her heart's repaired, her lungs look great and she's hitting all of her milestones either on time or early. She really is just a picture of best-case scenario for EVC.

Gerald: Bella has given Katherine and Jeff a run for their money.

Katherine: Oh, she brings so much laughter into our lives, she's a little spit fire. She's 100 percent whatever she feels, whatever she does, she's always at 100 percent.

Gerald: Many people had opinions about Katherine and Jeff having another child.

Katherine: We were even given the option from genetic counselors. They told us, "You can do IVF and test the embryos to see if they have EVC and we can only implant those that don't, so if you want more children there are ways to avoid it."

Jeff: Given the blessing that Bella has been in our life, we weren't afraid, we knew the risk existed. But what is the risk? If it's the risk of having another Bella, we will take that! We love Bella. It is our opinion that Bella's perfect. We wouldn't trade Bella for a child that doesn't have EVC simply because of the diagnosis, so we said, "No, we are not going to do that."

Katherine: There is definitely fear going into it. We knew that there was a chance of having another child with EVC and that child might struggle more, and that child might not survive. We went into delivery day not knowing if this is a child that we are only going to have with us for a short time, or is she going to be able to survive in this world. The only thing that really got us through that and gave us the courage for that is our firm belief that life is precious, life isn't just on this earth, it continues afterward. So, bringing in another little soul even if her life was short, it would not be void of meaning. We had seen Bella's story and her impact on so many people. We knew that Shiloh's story would impact other's as well. Shiloh in the bible is known as a place of peace, not because of its geographical place or anything like that, but it's where God's presence rested. We knew that was the source of our peace, and that was source of our strength. Regardless of that outcome, we would have that. So, going in to deciding to get pregnant, and throughout the pregnancy, we had a great deal of peace.

Gerald: After getting pregnant with Shiloh they waited patiently for the 19-week ultrasound.

Katherine: As soon as the measurement popped up on the screen for the arm and the leg measurements, we knew that child had EVC. It was a really emotional appointment with the ultrasound tech.

Jeff: I remember these were measurements she hadn't seen before as well as a heart defect. For the tech these were not good things, but for us we knew that they were compatible with life. If it were not for Bella, we would have been given a terminal diagnosis.

Katherine: The tech was crying at the end of the appointment and we were crying too. We told her, "It's okay, we have hope and we've seen great things happen, so it's not without hope."

Gerald: Their doctors didn't push them to terminate the pregnancy this time.

Katherine: They knew already that this was a baby that we were going to want and we were going to give every chance to fight. It was interesting to see though how that experience with Bella had even changed their attitudes and the way they talked about Shiloh. When we were pregnant with Bella, they were always telling us worst-case scenario. They kept talking to us about the day Bella was going to be born. They told us of the steps after birth. She would be taken away and assessed. That assessment will determine if she's compatible or incompatible with life, and if she will be brought back to us to die in our arms. She could also be put on

oxygen support and then they will determine if they could help her. They talked through all the possibilities with very grim outcomes. With Shiloh there was none of that discussion. Sometimes we felt like, "You do understand that Bella is the best-case scenario and we are not guaranteed the same outcome with Shiloh." EVC is somewhere between 30-50% lethal, so we knew that we were not guaranteed a child who thrived as much as Bella has with EVC. The day we delivered Shiloh, I was feeling the need to push and then all of a sudden someone in the room was like "Wait a second, we need to call the NICU, like we need Peds up here." I had to hold off pushing for a minute for them to get some doctors in the room.

Jeff: Stark contrast with Bella, we had like a dozen doctors in the room.

Katherine: Yeah it was a much smaller team, which was interesting because Shiloh actually ended up needing more assistance after birth than Bella did. Bella came out screaming and scoring nines on her APGAR's. She just didn't need any assistance, whereas Shiloh was not screaming, clearly in distress, and they immediately had to give her oxygen support. She had to go down to the NICU fast after delivery.

Gerald: I asked Katherine to describe EVC.

Katherine: Ellis Van Creveld primarily affects bone growth. EVC is a form of dwarfism, people with EVC are Little People and they will always be small. A couple of the unique things that present to this form of dwarfism are they have an extra digit in their hands, so they have six fingers, as well as heart defects specifically associated with EVC.

Jeff: Usually repairable.

Katherine: The most common is actually what Shiloh has, which is the combined atrium, so she is missing that atrial septum in the heart. So, there's often that heart defect, in addition to the short limb bones in their arms and in their legs. People with EVC tend to have dental abnormalities as well. Bella is almost three and she has four teeth that are very well erupted and three that are somewhat erupted, and we are not sure how many teeth she will actually end up having. It's a mystery that we'll find out as she grows.

Jeff: We have so many ways to correct things like that relatively easy.

Katherine: The reason there is a high lethality to it is due the bone growth of the rib cage. If the rib cage doesn't grow large enough to have sufficient lung tissue to support life, especially when you are talking about someone with restricted lung growth, add in a heart defect and that makes things a little bit more complicated.

Gerald: Although Shiloh has EVC like her sister Bella, her symptoms have been different.

Jeff: Her ribs are shaped differently than Bella's. Shiloh was given a 50% chance by the pulmonologist that she is going to have to do a rib expansion surgery. Another thing that's

different than Bella is that she's also got a more complicated heart defect, she has no wall between her two atriums. She also has oxygen-depleted blood mixed in with oxygen-rich blood, so that needs to be fixed. Along with the valve issue, she's got a couple challenges or obstacles ahead.

Katherine: She works quite a bit harder to breathe, it's a delicate balance right now between helping out her heart and helping out her lungs. Sometimes what you would do for one would hurt the other, pulmonology and cardiology work closely together with her to keep her on the optimum path here until she's big enough and at a good age for heart surgery.

Gerald: Bella and Shiloh's older sister is five and loves her baby sisters with EVC.

Katherine: She sees her sisters as entirely normal. We started to have some conversations and she may have a real limited grasp at this age, which will certainly grow as she and her sisters grow, but she interacts with her sisters just like you would see any other siblings interact. We couldn't have a better older daughter in our family. She's so good with her sisters and the perfect leader of the two of them.

Gerald: Katherine and Jeff eventually surrendered to God and moved forward with faith. For people on the outside looking in, they are often panicked at the thought of multiple children with disabilities. After interviewing parents with children who have multiple children with disabilities, it's clear that they feel so grateful, and commonly feel that it's the best thing that's happened to their family. Katherine and Jeff are no different.

Katherine: When you are told that you're not going to have time with a child and then you get day after day after day with them, even when things look bad, you still remain grateful that they are here. I think we more appropriately realize the big things and the little things with Shiloh. Whereas Bella being so on track in milestones and wanting her to excel and do well in everything. Shiloh is on her own little pace of development that's a slower pace and we honestly don't really care. She's alive, she's doing well, she's not in the hospital, she's not on oxygen support. Those are our victories, so those milestones and markers are important to see that your child is growing and developing, but they lose some of their importance. The little annoyances of life are properly put in perspective as little.

Gerald: Katherine and Jeff have advice for parents who are given a prenatal diagnosis similar to their daughter's.

Jeff: When we were first given our lethal diagnosis, it was because the chest wasn't going to support lung tissue, and in their opinion, given all the cases they have seen up until that point was that the chest wasn't going to grow or grow very marginally past that point. In our experience in both Bella and Shiloh, the ribcage continued to grow. When they were born it was still smaller, but it continued to grow, that's when we are in support of life. If somebody else has a diagnosis out there whereby the rib cage doesn't look large enough to support life at a younger age, just recognize that it can grow, and it does grow. At least in our case it did.

Katherine: And that lung function can improve, which with a lot of conditions isn't the case. We've talked with our cardiologist and she said, "We're used to seeing kids who are, if anything, going to get worse, but EVC kids, actually, with age usually improve."

Jeff: People out there won't have been diagnosed with EVC in the lungs, but it can present itself in different ways, probably a shorter polydactyly or thane atrophic dysplasia or something that's fatal. My advice would be it let it play out, let God do his work. Often times their experience leads to a certain outcome, but not all cases.

Gerald: I asked Katherine and Jeff if they wondered about the future much, or if they live one day at a time.

Jeff: We try to live one day at a time. At times there are things that make us think about the future, "Oh, what's that going to look like? Well, gee, that's going to be a challenge in this way or that way."

Katherine: I try not to live too much in the future because if I think about how I'm going to handle all of the situations that are going to present themselves, I get overwhelmed and I think, "How am I going to be able to handle that, or how am I going to be able to do this?" I had that experience even when I was pregnant with Shiloh and with Bella's heart surgery coming up with a newborn, who I didn't know what her health condition was going to be like and how on earth am I going to be able to take care of this one daughter getting heart surgery with a newborn. It's so easy to get worked up and think, "This is impossible, I can't do it," but what we've seen time and time again is you just do it, it works out and you figure it out. It's usually not as hard as you imagine it is going to be. We try not to consume ourselves too much with worry for the future, but at the same time informing ourselves of issues that we need to be aware of. This time around with Shiloh, we've realized how much we need to be an advocate for our child. The medical community does their absolute best, but we can definitely be of assistance in her care and in her treatment. It's been hugely beneficial for us to get connected to other EVC parents. You know, some of those connections actually led to us discovering a doctor out in Pennsylvania. She works with the Pennsylvania Dutch Amish, which is the most common place where you see EVC. So, she's seen more EVC cases than any other doctor and actually published an article about the benefit of postponing cardiac surgery so that their lungs could actually improve and so that you don't end up needing a trach or other interventions for lungs after surgery. The original plan with Shiloh was to have surgery this spring and then we made contact with this doctor, and our cardiologist is amazing, together they decided okay looks like best case for Shiloh is actually to postpone surgery. That's the benefit of the age we live in, and because of Facebook we can be connected to parents with the same diagnosis, share information with each other and share experiences on something that's so rare that most of our doctors haven't really encountered it really, if not at all.

Gerald: Having two children with EVC has blessed Katherine and Jeff.

Jeff: Other than the fact they are smaller and we have to make adjustments in the house because they can't reach things or they can't pull up things that somebody of average stature often could, I sometimes forget that she was in some way or that the world might call her disabled because she really is like any other child. We have a lot of stools around the house which we tend to stub our toes on.

Katherine: And there's going to be certain challenges and changes to life and life isn't going to look the same, but you're in for an awesome ride getting to watch these kids fight and surpass challenges and difficulties. Seeing them just so full of life is such an awesome privilege. You're not going to regret giving them the chance to fight.

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