An Orange Socks Story: Lauricia- Cornelia de Lange Syndrome

Interview by: Gerald Nebeker, President of Orange Socks

Gerald: I am thankful to Lauricia who took the time for an Orange Socks interview by phone to talk about her son Zacchaeus, who has Cornelia de Lange Syndrome, a very rare condition.

Gerald: Lauricia, thank you so very much for taking the time to speak with me. I'm honored that you would talk about your son. Tell me, when did you find out that Zacchaeus had Cornelia de Lange Syndrome?

Lauricia: Initially we found out about his diagnosis just an hour or so after he was born via C-section; however, we had multiple indications that he had the Syndrome prenatally. We were doing lots of different testing and frequent ultrasounds, and it started at our eight-week appointment. The doctor said, "You're measuring just a little bit small, but it doesn't seem to be a concern at this time." We kind of just ran with that, and then at our 14-week ultrasound, he measured two weeks behind, so he was then at 11 weeks. They said he was still a little small, but they weren't seeing anything too concerning. Fast-forward to our anatomy scan at 20 weeks. He was measuring anywhere between 16 and 17 weeks, and at that point, they were seeing many abnormalities. The biggest thing obviously was his slow growth. He was not measuring where he should be or where I should have been gestationally. He had micrognathia, which is an undersized jaw, so they were seeing some facial defects as well. At that point, I was seeing a midwife, and they referred us to maternal fetal medicine. During that time, they had done some more testing. We had an amniocentesis, a chromosome analysis and a microarray, and surprisingly, all those tests came back completely normal. They decided to refer us to a children's hospital, so we were collaborating with the children's hospital, and they were doing their own work-ups. I had a full body extremity MRI and a fetal echo, and on one of my last ultrasounds, my doctor actually said, "I think I'm going to do this ultrasound myself instead of the stenographer." As we were doing the ultrasound, I was watching his facial expressions and trying to read what's going on. He said, "Your son has abnormally long eyelashes for his gestational age. I suspect he may have this Syndrome called Cornelia de Lange Syndrome, and he wanted us to meet with a geneticist. We did not have the chance to meet with the geneticist before we was born prematurely, but that diagnosis was confirmed clinically at birth, and then six weeks later, testing confirmed it.

Gerald: So, what advice or counseling did your physician offer you when they found that they had some issues during the pregnancy with the baby?

Lauricia: Initially, I don't feel like we were really pressured, but our first offer was to terminate the pregnancy. We had just gotten the news about everything that was going on, and we felt we didn't really have enough time to even process what was

happening. They gave us a minute and left me and my husband. We cried, and we talked, and we decided we were not going to terminate the pregnancy. When they came back in, they gave us other options and said they would like to do an amniocentesis. That's when the testing began. It was hard emotionally, very hard. I was something new to us and new to our family. Even though it's a gene mutation, we have not had anyone in the family with any gene mutation of this sort. They decided to offer us counseling, but I declined that as I had lots of family support. My family was tremendously supportive, and I felt like we could get through it as a family together.

Gerald: Cornelia de Lange Syndrome is pretty rare. Can you tell me a little bit about it?

Lauricia: Absolutely. It occurs in 1 in 10,000 live births. There's not too much research into the Syndrome since it's such a rare disorder, but what it does show is restricted growth and potentially no limbs or partial limbs, congenital organ defects and developmental delays. Even though we really don't know where that falls on the Syndrome, it could be anywhere from severe to mild. We really don't know what the prognosis of that is, but we do know that the biggest thing is that all of the kids with the Syndrome have similar features in their faces. They have what I like to call his unibrow, and he has very long eyelashes. These kids are really small with small fingers and toes that are symmetrical to their body, but they are abnormally small. While my son does not have seizures, they could potentially have seizures coupled with their growth, motor or fine motor skills, walking, crawling, and definitely eating disorders. Many of them are on trachs or are on oxygen. My son does have a cleft palate, so that, along with his micrognathia, makes it that much more complicated to eat. We are just taking it one day at a time, and we hope that there will be more research and more knowledge about his Syndrome as we continue to grow with him.

Gerald: Some of the issues they can fix, like the cleft palate and heart issues. Does he have heart issues?

Lauricia: Zacchaeus was born with a VSD and a pulmonary stenosis, so when he was about five months old, he had open-heart surgery to repair them. There is some mild leakage, so at some point in his life, he will have to have another open-heart surgery. His first surgery was a G-tube. As I said before, he didn't eat, so his main nutrition comes from his G-tube. His second surgery was the open-heart surgery, and his third surgery was a mandibular distraction. That moved his jaw forward to open up his airway so his sleep apnea could potentially be resolved and eating could get better for him. We had the hardware removed a couple of weeks later. We have been waiting on the hospital trips a lot, but it's all in the best interest and benefits his condition. We are giving him a break from surgery for now, and in a year, we are hoping to fix his cleft palate. Unfortunately, we can't do that now as it would compromise his airway. but we hope to fix that and give him that nutritional value and hope that will help him and add to his quality of life.

Gerald: So you think that down the road, you might be able to remove the G-tube and he might be able to eat by mouth then?

Lauricia: That is ultimately the goal. Potentially, he might still have to have the G-tube for some nutritional value for more calories and such, but we are hoping that he will be able to eat enough to supplement the calories that he is getting from the G-tube and to help with his fine motor and sensory skills and other things that go along with that. We work really hard with his therapists who work on these sort of things. One of his therapists is for vision to help with astigmatism in his eye. One of his things with the Syndrome as well is his eyelids are set lower than ours would be, so his vision could be affected by that. We work really hard to strengthen his eyes, his vision and tracking to continue helping other parts of him to grow and develop properly.

Gerald: Interesting. Thank you for explaining a little bit about Cornelia de Lange Syndrome. You obviously didn't abort the pregnancy and you had your son. What have been some of the greatest challenges that you've faced in raising him? He is how old now?

Lauricia: He is three months over two years old.

Gerald: Okay, so over the last two years, what have been some of the challenges?

Lauricia: I'd say our biggest challenge is learning how to balance everything. It has been a struggle to learn how to balance our work life and our older son. My son has four different therapists, and he sees 10 different specialists, so it has been challenging balancing out all his therapies, appointments and our outside life, and trying to just optimize his life while trying to give attention to our older son. Trying to normalize everything and balance everything has been quite a challenge, but it's always rewarding as well.

Gerald: Tell me about the joys.

Lauricia: Oh, the joys are never ending. He's awesome. He has taught us so much about perseverance. I think one of the biggest joys we've gotten out of him, besides himself, is that he's brought my family so close together, and not just my immediate family of my husband and my son. Our siblings, grandparents, aunts, uncles and everybody have been such a tremendous support system. You bond together. He has taught us how to self-educate ourselves on how to better his life; he's been really amazing. We've been able to advocate for him really well, and it has been awesome to learn and to grow with him, because unfortunately, there's not much research on his Syndrome. We don't know entirely his prognosis, but it has been such a joy to learn and accept every single phase of development in his journey. It has been amazing to just enjoy that with him, include ourselves and be there with him to support him.

Gerald: That's wonderful. I guess I'm wondering if I came to you with a prenatal diagnosis of Cornelia de Lange Syndrome, or I found out after birth, what advice would you give me?

Lauricia: My main advice to any parent who has any type of diagnosis prenatally in utero would be to use your resources. A lot of people are not fully aware of the extent of resources that are out there. You don't quite understand that until you have to start using them. Utilize them; they are there to help you. It can be scary and very intimidating because you don't really know what to expect as a parent expecting a special child, but they are there to help you and guide you. Sometimes you just have to accept it and let them help you and guide you, and kind of walk with you through the journey and be a guidance to you. That has been very important for me, because had I not had the support system or proper resources, I think it could have been a lot more emotionally damaging than not using the resources available.

Gerald: You have to take him to lot of doctors and lots of therapists. My assumption is that he's a lot more work than your other child. You have another son?

Lauricia: I have a six-year-old who is a very healthy and rambunctious little boy.

Gerald: But with all the work that this little guy is taking, is it worth it?

Lauricia: Oh, absolutely. Another thing that we've enjoyed is that we only have one other child. I was a little worried to say the least at how he was going to take everything, because we spent a lot of time in the NICU and Zacchaeus has had multiple surgeries, so we've spent a lot of time doing the hospital life. I was just a little worried as to how my son would take it. I feel like he's kind of accepted it as well, and it has taught him to love in a different way. He is so gentle with his brother and loving with him. We all have come to grow together as a family, and my fears that I initially had are not even present any more, because I am so confident in our love for him that none of that matters any more.

Gerald: That's great. Any final remarks or anything else that you'd like to say?

Lauricia: I just wanted to say that looking back at it all now, it really makes me appreciate him. I want everyone to know that sometimes you're going to have diagnoses that are unknown up until delivery, and even maybe years after delivery, you may not have a definitive diagnosis. That's the scariest thing people can come across, because we weren't really sure if we were planning for a funeral or planning for a baby shower, and that was really hurtful for us. We didn't know what to expect. That is really scary for people, but you really can be strong about it because like I said, there is so much support out there. Even parent to parent, many doctors, many clinics and many resources that you can use that can help you to be strong enough to do it. It is definitely worth it, and I would never advise someone to terminate a pregnancy because these children are the biggest blessings, and they will give you

all the strength you need. You'll learn to advocate as a parent through their journey, and it's amazing. I would be there right with you and do the journey with you if I had to do it all over again.

Gerald: That's wonderful. You're awesome. Thank you so much for taking a few minutes to talk with me; I'm honored that you did.

Lauricia: Thank you so much for listening to his story. I hope that it helps other parents become stronger, and I really appreciate that you'd take the time to interview us for our story.