An Orange Socks Story - Maria Update: Trisomy 18 or Edwards Syndrome

Interview by: Gerald Nebeker, President of Orange Socks

Welcome to the Orange Socks podcast, where we are inspiring life, despite a diagnosis. I'm your host, Dr. Gerald Nebeker. It was a pleasure to meet with Maria again. The first time was over two years ago when she was pregnant with her daughter, Serenity. That was such an anxious time. Serenity was diagnosed in utero with Trisomy 18 or Edwards syndrome. When Maria was a little over ten weeks along, the doctors gave her a very grim prognosis. The reality is most babies with Edward syndrome die before or shortly after birth. Because of that, Maria was advised to terminate her pregnancy on many different occasions. However, she did not want to do that. Maria wanted to give Serenity a chance.

Maria: I found out about her diagnosis at 10.3 weeks of conception. I was given all of the negative aspects of her diagnosis. I was told that there was no hope, that it was a life-threatening diagnosis. That I was only prolonging the inevitable, and that I just needed to let everything take its course. I was prompted on several occasions to terminate my pregnancy. My baby had a beating heart and I knew she was a female. And the same day I got the diagnosis, within the hour, I knew her name was to be Serenity Grace.

Gerald: When she was almost nine months along, Maria had to move to another state in order to find a doctor who would support her in giving birth to Serenity, and who would have the equipment and specialists there to support Serenity after she was born. While the doctor was supportive of Maria, she was told that she would only get about an hour with Serenity before she passed. Around that time, was when I first met Maria and she did her first Orange Socks interview with me. Her story was tender and compelling. She had to fight so hard just to give birth to Serenity. I never got to hear the rest of the story until Maria emailed me a little over two years later and said she was ready to share the outcome to the world. I was so excited to learn what happened, and this is what I learned:

Maria: So, prenatal care was very limited in the small town I lived in, and in the state I was residing in at that moment. When I asked certain questions about things we would do if my baby was in distress, the doctors told me they would be the ones to make the decision on whether or not we would perform an emergency c-section, I knew at that point that it was not their choice, this needed to be my choice. I was choosing life no matter what the diagnosis was. As long as she has a beating heart, I was to fight for her. The journey began and I ended up finding a doctor out of state and advocated for myself to get into the Ronald McDonald House in Orange County, and I found an amazing doctor that believed in life. This is where my prenatal care began. The journey of traveling hundreds of miles just to get the care I needed. It was decided that we would have a controlled labor. I was to be induced and seven weeks before my induction date. I knew that I needed to relocate and go back to the state of California where I would be delivering her. Prenatal care was always the same, it was still prompted that I was carrying a baby with a very life-threatening diagnosis. The neonatologist even said that I was prolonging the inevitable. Again, I was prompted, even in that state, that I shouldn't continue with the pregnancy and that I really needed to consider once she was born things like trachs, and G-tubes, feeding tubes. There was opposition to that for Trisomy 18 babies. I had a care plan, a birthing plan which could change at any moment, but I had asked for all life-saving efforts for her.

Gerald: The plan was to induce Maria on May 12, but Serenity had different plans, and the day before Maria went into labor. After eleven hours of labor, and after almost needing a c-section, Maria had Serenity.

Maria: I was scheduled for May 12, my baby was also diagnosed with a congenital heart disease in utero, so she would also have problems during labor, and she did. My God decided that May 11 would be the day she would come. My water bag broke at the Ronald McDonald House at 1 PM, and I was scheduled the following morning to be at the hospital at 5AM. The way it turned out, her heart would not have withstood the induction. She struggled during labor. It was eleven long hours of labor. I did have an epidural, and again, her heart was not doing well. So, I was told that if I continued with the epidural that I would be compromising Serenity. At that point I decided, “No more, let’s stop the medication now, and I’m going to do this as best as I can. No one can tell me how to do it or what to do, just let me do it.” I continued to labor without medication. Serenity’s heart continued to drop until the very end. Right before I delivered her, she had what they called D-SAT and her heart had dropped so low in the 20’s that there was hardly a heartbeat. They were preparing for an emergency c-section when my doctor said, “Okay momma, you’re on. Baby Serenity needs you now. Push.” And I did everything I could with everything I had in me to deliver her. I gave three of the largest pushes that I could ever give in my life, and there she was.

Gerald: She didn’t have any facial deformities like the doctors said she would. When she was in utero, her hands were clenched so when she was born, they thought she would have clubbed hands. She does not. She also came out crying, another thing the doctors said she would not do. They had about seven different professionals waiting to work with Serenity after she was born.

Maria: I had an entire team in the hospital that were waiting for her. There were seven professionals in the delivery room awaiting Serenity with the NICU incubator waiting for her arrival. They expected her to pass within minutes of delivery and they expected her to go on oxygen immediately, and none of that came true. She was able to breathe on her own, she was this beautiful baby girl that cried immediately. She weighed five pounds, [was] seventeen- and three-quarter inches [long] and she was just perfect. Just absolutely perfect. They expected her to be born with deformities, and the way it turned out, the geneticist was shocked. The team was shocked that she had a little toe that didn’t fully develop. She had slight rocker bottom feet. She was able to open and close her hands, in utero she kept them clamped shut so they assumed she would have clubbed hands and clubbed feet. She just had slight features that appeared that she may have Down syndrome, but again, they were shocked.

Gerald: Maria's birth and care plan said she wanted every life-saving measure for Serenity. Although, in the end, none were needed. Maria remembers praying that she would have just fifteen minutes with the baby to love her. When fifteen minutes were up, she prayed for fifteen minutes more.

Maria: I just prayed, and I asked God if He could just give me fifteen minutes to love her. To tell her how much I loved her and to tell her how she was just the most perfect beautiful baby. And when God gave me fifteen minutes, I asked Him if He could just give me fifteen minutes more. And as the hour went by, and they took her away and they checked her out I remembered just asking if I could maybe have another day with her to love on her. May 11th of this year Serenity turned two years old.

Gerald: An hour after Serenity's birth the doctors took her away to work with her and to do some tests. When the tests came back it showed that sixty percent of her chromosomes were trisomy 18 and the other forty percent were normal. This means she has Mosaic Trisomy 18. This is extremely rare. Serenity defied all the odds, she does not have a trach, nor does she use any extra measures to breathe, which is fairly common.

Maria: When Serenity was born, they did the testing. They did a rush STAT test on her, and the geneticist said your baby is what we call Mosaic Trisomy 18, very rare babies. Sixty percent of her cells have the third copy of the 18th chromosome, forty percent of her cells are completely normal. How each of her organs is affected, she is still telling us what she needs. She is on a feeding tube, she has had feeding issues since she was in utero. When she was born, she was on a feeding tube, first it was through the nose and now it's actually a button in her belly. But again, these are all things that the doctors did not know, and they wouldn't have ever known.

Gerald: Serenity is now two years old and she walks, runs, plays and talks. Although there have been some challenges along the way.

Maria: So neurologically, she's delayed. Her speech is delayed, she is learned how to sign, and she has very low muscle tone, so she falls a lot. She wears braces, but today for the interview, I decided we were going to leave the shoes and braces at home, I have her in sandals today. But she is delayed developmentally. She's had two swallow studies and it is neurological for her. Prognosis wise, I finally asked the question right before she turned 1. I was told that the chances of her making it to year 1 were great, the chances of her making it to four are good and the whether or not she will graduate from college, we don't know. But I've already made plans, she's going to outlive me. I've already spoken with my daughter who is 23, she is going to take care of her sister because she is going to outlive me. Just getting services for her, and again, because of the Trisomy 18 stigma, a lot of doctors don't believe in providing more services, and then they meet her, and they've been floored. I've been told things like she's a miracle from the medical profession, they can't believe she's walking and talking and running around, that she's not on any kind of breathing assistance. But it's been advocating, just having to advocate for her since birth. Having to make it back from one state to the other.

Gerald: After ten days of being home, Serenity was struggling to eat after being off of her feeding tube, Maria knew something was seriously wrong and she packed Serenity up at midnight and drove Serenity four hours to the hospital. Maria had to wait 10 hours before someone would look at her little daughter. The whole time Maria pleaded, “Something is seriously wrong with my baby, please help me.” When they finally looked at Serenity, they indeed realized that something seriously wrong was going on with her. After pushing for additional tests by Maria, it was discovered that Serenity’s brain was not working like everybody else’s, her brain does not send a signal to swallow and breathe and she chokes, so she is at risk for aspiration. Having a feeding button installed has fixed the problem and has helped tremendously. Serenity also has low muscle tone and she falls over frequently, so she wears braces to walk and she receives occupational therapy, speech therapy, physical therapy and feeding therapies.

Maria: We ended up in the hospital ten days after we got here. She was struggling to feed, and they had already taken her off of her feeding tube. I just noticed something was terribly wrong with my baby and she was crying a lot. I decided to take the four-hour trip to the nearest major medical that had a children’s hospital and all within the state I was residing in. And even getting there leaving here at midnight, which should have been a four-hour trip, turned into a five-and-a-half-hour trip. I just said, “Something is seriously wrong with my baby, please help me.” I spent about ten hours there before somebody would look at her and finally realize that something was seriously wrong with her. Through more advocating and testing, three weeks in the hospital, we found out that, yeah, she has serious breathing and feeding issues. Her brain is not working like everyone else’s. Her brain does not send the signals to swallow and breathe, so she is at risk for aspiration. We are still working on that. And for most of her life she was on a bolus feed once every three hours and then a continuous feed for ten hours throughout the night. As of a few months ago, we have upped her feeds to where she is having five a day, once every three hours on the hour. She also has lots of therapies, physical, feeding, speech.

Gerald: I’m impressed with Maria’s advocacy for Serenity through the pregnancy. Maria also had to advocate for herself and her birth plan during the last few months of pregnancy. Her doctor told her to prepare for life, but to also prepare for death. Maria had to plan Serenity’s funeral, but she was blessed with a community that donated for a funeral that didn’t happen. Maria continues as Serenity’s strongest advocate.

Maria: I’m out there advocating for her. Since she was two months old, I signed up with services to get early intervention for her.

Gerald: Despite the challenges they have had, Maria only expresses joy. Just Serenity smiling, breathing and waking up brings joy to Maria. She also finds comfort in knowing that Serenity’s beautiful face is going to light up as soon as she sees her. Maria says that Serenity is her whole world. Every milestone that she reaches is a new joy. She is so much more than the doctors predicted. Maria is so grateful as to be Serenity’s mom.

Maria: Breathing, waking up, just her smiling and knowing that that beautiful face is going to light up as soon as she sees me. Now, I do work full time, I have an attendant caregiver. She did have a nurse, skilled nursing, but we outgrew that about thirteen months in. We got off of the skilled nursing to transition into attendant care, which is amazing. Just to know that when I get home from work, that I make her day. She light's up, I'm her world, but she doesn't realize that she's my world. She's everything.

Gerald: Maria advises parents with children who have similar diagnoses to never give up, never stop advocating for your child. As long as your baby has a heartbeat, they are fighting, and we need to fight too. She urges parents to question everything. She says, "When you're at an appointment, ask a lot of questions. If there isn't a service provider that you think there should be, search for it. Seek out and advocate for services for your child. Just don't ever give up."

Maria: The best advice I can give is don't give up, don't give up no matter what. Doctors are practitioners. They are practicing medicine. I believe in a Higher Power, I believe in God, and He has the final say. My advice is to ask a lot of questions, question everything. Find out about your baby's diagnosis. No matter what, keep fighting. If your baby has a beating heart, then your baby has a shot. As long as you continue to fight, but your baby depends on you to depend on yourself, to ask all the right questions. I can't say it enough and emphasize it enough. Question everything. Everything. If there is a service out there that hasn't been provided, seek it. Seek it and fight for it. I was not offered services during my pregnancy. Genetic services, genetic counselling, which would give me better ultrasounds, 4D, that is something I had to fight for and ask for. And It was just to see what challenges and what medical professionals needed to be present during her delivery, that we would have a cardiologist, a neonatologist just to fight for her. So, the best advice I could give is just don't give up no matter what. Your baby hasn't stopped fighting, and your baby's heart has not stopped beating, so you don't give up no matter what.

Gerald: I was honored to meet with Maria again and catch up, and most importantly to meet Serenity. It was such a thrill for me. I can see how the world is a better place because Serenity and her dedicated mom are in it.

Thanks for listening to this episode, Orange Socks is an initiative of Rise Services Incorporated. A nonprofit organization 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.