An Orange Socks Story – Amanda: Pfeiffer Syndrome

Interview by: Gerald Nebeker, President of Orange Socks

Gerald: Welcome to the Orange Socks podcast, where we are inspiring life despite a diagnosis. I am your host, Dr. Gerald Nebeker. In this episode, I speak with Amanda by phone about her daughter, Emmy, who has a rare condition called Pfeiffer syndrome. Emmy's parents created a Facebook page for her, and I invite you to visit it; just search for Emerson the Unicorn. It's a delightful page by her family. I learned a lot from Amanda, and I know you will too.

Gerald: Amanda, thank you very much for taking the time to do an Orange Socks interview with me today. We are talking about your daughter, Emmy, who has Pfeiffer syndrome. I've never heard of Pfeiffer syndrome, could you tell me a little bit about it?

Amanda: Not many people have heard of Pfeiffer syndrome. It's about a 1-in-100,000-births syndrome that has multiple issues. The most obvious one is the craniosynostosis, which is early skull fusion. The sutures in her skull began fusing while she was in the womb, and that can cause multiple things. In her case, she had pretty significant proptosis, which is bulging of the eyes due to shallow orbital sockets, and her skull is oddly shaped because of the way it had already began to fuse. That can cause multiple neurological issues because of delayed brain growth. Some Pfeiffer syndrome specific markers are white thumb, white toes, the craniosynostosis, gosh, there are a lot of things that go along with it.

Gerald: When did you find out that Emmy had this syndrome?

Amanda: Oddly enough, we found out at our 20-week anatomy scan. I'm an older mother, so we had all the DNA testing for all the different things that they test for, and all that came back completely normal, no issues whatsoever with her. So, we went in there thinking it was just going to be a quick anatomy scan just to make sure everything is growing fine. We already knew she was a girl because of the DNA testing, and we found out at that 20-week scan that her skull had already began to fuse. It was a really tense moment, watching the technician go over and over the same areas, and we just knew immediately that something was not right.

Gerald: When did you, then, actually get the definitive diagnosis that it was Pfeiffer syndrome?

Amanda: We didn't get that until after she was born. Before that, we scheduled a visit with our craniofacial team at our local children's hospital, and the doctors there looked over the scans and the 3D imaging that they had done. The doctor said that based on what he could see, he believed it was Pfeiffer syndrome. However, there are multiple syndromes that fall into that same area like Crouzon and Apert's syndrome. There are lots of different syndromes with that same genetic mutation,

but he believed it was Pfeiffer syndrome. He offered us genetic testing, but because it would have taken an amniocentesis, we opted out. We figured we would find out once she got here when we could get the definitive diagnosis.

Gerald: Did your doctor give you any advice at that time, when you found out that there was an anomaly with the baby?

Amanda: Yeah, he did, and it was incredible. I will start by saying that the initial doctor we saw after the scan was not helpful. She didn't know exactly what it all meant. She knew that there was a pretty severe skull fusion at that appointment and presented us with our termination options, she didn't give us any other information. We are the kind of people who want to have a second opinion, we want to make sure we know what we are doing before we make big decisions of any sort, and that's just who we are. We saw a top craniofacial doctor who gave us lots of stories of kids that he sees with the various syndromes that it could be, including Pfeiffer, and he told us realistically that it was going to be a very long road. She's going to require lots of surgeries and lots of intervention, and it could be a long journey for her, but that the kids that he sees are happy and healthy. Many of them go off to college. There are a wide variety of outcomes for these kids, and he gave us very straightforward information about it, and gave us a lot of hope.

Gerald: That's good. So, your first physician essentially then only offered you termination options as opposed to raising this child, is that correct?

Amanda: Yeah, I never saw her again, and there's nothing political about it, it's just that's not the kind of thing that you want to hear at this kind of appointment. We understood where she was coming from, but then we saw another obstetrician besides the doctor who helped us get the diagnosis, and he was wonderful in agreeing with going forward with the pregnancy, taking care of her and me, and keeping a very close eye on her so that we could make those interventions with her delivery. He was incredible. We immediately fired the first one.

Gerald: That's awesome, and Emmy is how old now?

Amanda: She's getting ready to be five months old.

Gerald: During these five months, what have been some of the hardest things that you and your family or you and your husband have had to deal with?

Amanda: Wow, that is a loaded question. Emmy was born in July, and she spent the first four months of her life in the hospital. That was pretty difficult for us as a family. We have a six-year-old as well, and it was very difficult for him during that time because our family was really disjointed. One of us had to be home with him, and one of us had to be at the hospital with her. There were lots of on-the-fly medical decisions that were very difficult for her. Going into it, we knew some of the things that could happen. Of course, we had high hopes that it wouldn't be very

difficult, but it has turned out to be pretty difficult. We had to make some tough choices as far as how to treat her. She got a tracheostomy and had to get a rotation in her bowels fixed, and there have just been a lot of things that we were prepared for, but we actually weren't prepared for.

Gerald: Tell me about some of the joys.

Amanda: Oh, she's incredible. We had to take her two weeks early because they noticed in some of the scans that her hydrocephalus was becoming very, very bad, and they wanted to go ahead and get her out and do interventions with a shunt to hopefully help her brain development. Every day is so incredible with her, because we are seeing so many amazing signs that she's developing. All the doctors would tell us that they never thought she would be able to breathe on her own, and she can. We still had to eventually get a tracheostomy, but for a while, she was able to do these things that they kept telling us she would not be able to do, and she keeps doing that every day.

Gerald: That's neat. What has been her impact on your family as well as your extended family and friends?

Amanda: Well, unfortunately, we are in a situation where we do not have our family close by. We are in a transport area, so most of our family lives about 2,000 miles away. It's difficult because we don't have a lot of family support out here, and she requires a lot of care with her different little quirks. My son sort of gets pushed to the side a little bit right now. We try to explain to him that it won't be forever because eventually she will be bigger and won't require as much attention as she does now, but it's tough on him. It's also pretty rough on us because of her tracheostomy and things like that. Leaving the house is quite a production, it requires two adults in the car to go places with her, so we don't get out much these days.

Gerald: Interesting. If I were to come to you, just having received a diagnosis that my child had Pfeiffer syndrome in utero, what advice would you give me?

Amanda: Be flexible. If you want to do this, you can, but you have to be flexible. You have to roll with the punches. It is not going to be easy. It doesn't matter the severity, because Pfeiffer syndrome has a pretty wide range of symptoms, and even at the least severe, your child is going to have multiple skull surgeries over the course of their lifetime, and that's hard to see. It's so difficult, but these kids can be, and often are, incredible. Cognitively and physically, they can do everything, they are just typical kids who don't look typical.

Gerald: What would be your daily routine with her? What types of things do you have to do with her trach and those sorts of things?

Amanda: She's pretty much a typical baby in a lot of ways, so we don't do a whole lot of different things other than baby things. The trach is a little more complex. She is also on a ventilator when she sleeps. She has pretty severe central apnea because her skull is pressing on her brain stem, and that's one of those things that surgery will fix eventually. We do have to keep her on the ventilator overnight or when she is deep asleep. Every day we have to do trach care, where we clean around her neck and make sure everything is nice and clean and dry, and she's not getting rashes or anything around the trach. It was one of those things where we thought it was going to be so difficult and so complex once we got home, and we were so overwhelmed with how we were going to do everything, but it turned out to be kind of easy. Once you get into the routine of it, it's not bad, the trach care is actually pretty minimal. It requires suction and cleaning, but other than that, it's really not hard.

Gerald: That's great. Thinking about your experience and what you've gone through, are there some last words that you'd like to tell folks who will be listening to your story?

Amanda: From our perspective, our daughter is developing very well. A lot of these kids who look significantly different, especially in the craniofacial ways, are typical kids. Inside, they are just kids, and they want to be treated just like any other kid. It's okay for you to ask questions about them, because Emmy does look strikingly different from other babies right now. I love answering questions about Emmy, because she is so incredible. We just had an instance at the hospital where a five-year-old child looked at her and kind of laughed and said, "Her eyes are funny," and it was that moment where I made the decision to say something. Do I scold her, or do I educate her, or do I just walk away and cry? I found out that through talking about it with the little girl, it helped me feel better. I was very happy that her mom got involved in the conversation and involved my daughter in the conversation. That's what we want, we want people to be interested in Emmy, and we want people to be kind to her, but we want questions and talking about her, and hopefully that will lead to her being included more when she is older.

Gerald: Thanks for listening to this episode. Orange Socks is an initiative of Rise 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.