An Orange Socks Story: Jori and Dan- Autosomal Recessive Centronuclear Myopathy type 5

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. I have Jori and Dan on the podcast today talking about their son Luxton. He received a diagnosis when Jori was 32 weeks pregnant with Luxton's little brother Atler.

Jori: Autosomal Recessive Centronuclear Myopathy type 5. There are three cases that they've found, but Luxton's is the only case with the two mutated genes. He had a mutated gene from me and a mutated gene from my husband.

Gerald: Jori and Dan described the first few days after Luxton was born.

Jori: He couldn't breathe on his own, they called a code blue.

Dan: They thought something was wrong right off the bat but nobody really knew what. The first assumption was it was malnutrition that he wasn't getting enough nourishment.

Jori: They had like a laundry list of things that they thought it could be. His MRI showed some brain bleeds. They thought he had Gyneschemic brain injuries because he did have a rough delivery so he was restricted in utero.

Gerald: After bringing Luxton home from the hospital Jori and Dan tried to handle the health issues Luxton was facing.

Jori: He was three weeks old and they decided to send us home. I'm a nurse and so he had a feeding tube in his nose when he was born. He wasn't able to swallow or suck. He had a ton of secretions and he didn't move. We called him a slinky, he's a lot better now.

Dan: We noticed he started to get more and more irritable to where we couldn't console him. We would try to burp him, change him, rock him, and feed him. The only time he would sleep was when he would get worn out and in the back of our minds we were thinking something is wrong.

Jori: He was three weeks old. We were getting ready to go to bed and all of a sudden, his oxygen rate dropped to like the 20s and his heart rate we couldn't find it. We were admitted for another three months on and off and they still couldn't find anything. He was getting a little bit stronger so we got a G-Tube placed and then they were like "its terminal whatever he has so don't expect him to look at you, don't expect him to laugh. He's never going to play with toys. He's never going to walk,

he's never going to crawl. He's never going to sit up, he's going to be a vegetable." and they didn't give us very much hope. One doctor wanted us to put him in a long-term care facility and that was really hard for us because we had no answers brand new baby.

Dan: Then he started throwing up, we were told he had Pilar Stenosis.

Jori: Its where the muscles in your stomach close off so everything, we were feeding him through his feeding tube had nowhere to go so it was coming back up.

Dan: When he came home, we lasted a day roughly because into the night he started throwing up gasping a lot. They had to do an emergency surgery on him because they found that he had that pilo. In the back of our minds the entire time was "something is wrong. What is it, it doesn't feel right, it doesn't seem right, what you are telling me?"

Gerald: There was finally one specific test that assisted in eventually determining a diagnosis.

Jori: They ran a test called a microarray. It showed small deletion on chromosome two. We didn't get an official diagnosis till he was almost 16 months.

Gerald: Dan talk's about how the diagnosis affected him.

Dan: I kind of had a hard time with that. They could tell me that he won't do certain things but I can actually see what he can do and I can work with him and see what I can have him do and what kind of life he can lead. He's starting to stand, I just look at it as he's got a condition that makes him have to work harder.

Gerald: Jori felt overwhelmed after receiving the news of Luxton's condition.

Jori: Well we got the diagnosis and I remember the lady walking in the room. she was like "so we found something this is a gene it affects but we have no other records of this being found. The other two records that are very similar is both kids are trache vented and doctors said that they don't have a very good prognosis. I was devastated and then I went to why us why our baby.

Gerald: But as time went on Jori and Dan were able to see the blessings they have come to enjoy and love Luxton's unique qualities.

Jori: He rolls and now he is lifting his head up off the ground. He does things they told us he would never even, do they are coming to us for answers and sending their cases occasionally. If I see another 18-month-old running around going down slides, sometimes I'll let myself go there but not for very long. He does things that other moms don't get to experience, we just have a very special bond. I feel we wouldn't have it if he didn't have this diagnosis, he is a magician with his feet so he didn't use

his hands for a long time he's always happy I think the only time he's ever really upset or cried is when he's in pain or something's bothering him. He has a favorite song it's by Michael Jackson. He loves to dance even though he doesn't stand, he is so funny.

Dan: He's definitely one of the happiest babies I've ever been around. After we got everything figured out, he's just had time to excel. He's never been one to just cry he loves to be around people he's a snuggler. For a lot of the time he's gotten more and more curious about the world and it's just funny to see that curiosity for him to take place. I love coming home and seeing him dance around me and get excited its really wonderful having him be a part of our family. He's just seems to have this determination to never really give up.

Gerald: Jori shares her advice for other parents who are going through similar situations.

Jori: I would probably tell you what my husband told me three months into us not knowing what was going on. I was having a really hard time, I was feeling guilty I was feeling sad, jealous, like I couldn't do it, I wasn't worthy, or why me. We were walking down the hall of the hospital and I was like "I can't do this." he stopped me and was like "Jori stop listening to the doctors, stop looking at everything they are telling you what's wrong and just be his mom." and it was something so simple. "Look at him and what he needs, then how can I give him the best life possible." and I stopped looking at what he's never going to do and what everyone's labeling my child as and once I stopped doing that, I was able to pay attention to what he can do; "oh he is making really big leaps." and then I started getting joy out of that, I mean I still get sad sometimes. But he can do so many things that they said he would never do. When I started focusing on the positive and asking how can I help him progress, it brought all the joy back into everything I thought went away when he was diagnosed.

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