An Orange Socks Story – Valorie: Cockayne Syndrome

Interview by: Gerald Nebeker, President of Orange Socks

Gerald: I was grateful for Valorie, who took the time to meet with me in her office for an Orange Socks interview about her son, Jace, who was diagnosed with Cockayne syndrome at age seven. Although they knew Jace had challenges much earlier, Cockayne syndrome is a very rare genetic condition that is fatal. Jace lived 20 years and passed away just 12 weeks ago. Valorie had to become Jace's champion, having to fight for insurance coverage for essential procedures to help him. Her advocacy is now benefitting other parents who have sought Valorie's advice on various issues, and she is even starting a nonprofit organization called Team Jace to benefit others.

Gerald: Valorie, thank you so much for taking the time to meet with me to talk about your son, Jace. When did you find out that he had Cockayne syndrome?

Valorie: He was seven and a half. We had been searching for many years. Cockayne syndrome is progressive, so as time goes on, these children develop more and more of the symptoms. In the beginning, he didn't have any hearing loss, and that's one of the big things normally. He did have microcephaly, small head size, and he was very sensitive to the sun, which I assumed was because he was blonde-haired and blue-eyed. I'm fair and burn easily, so I assumed the sensitivity was because he was my child. Looking back, if you type in microcephaly and sun sensitivity online, the first thing that comes up is Cockayne syndrome, but it was not one of the symptoms that I ever threw in that basket. I thought that was just one of those neutral things that really didn't matter. We started taking him to dermatologists at about two and a half years old, because even with tons of sunscreen on, he would still burn and blister, and his eyes would burn. We had been to several dermatologists, and actually the first person who said Cockayne syndrome to us was a dermatologist at Primary Children's.

Gerald: Interesting, and this is a rare condition.

Valorie: Very rare.

Gerald: Tell me a little bit about how you were actually able to confirm the diagnosis.

Valorie: At that time, there was no testing available in the United States. I found a doctor in England who could do the testing, so they did skin biopsies at Primary Children's, put them on a plane and got them to him as quickly as possible. He cultured those cells, actually exposing those skin cells to UV. All of us are exposed to UV every day, and even though Jace was very sensitive to the sun, he didn't sunburn like a typical person. Jace didn't have any reaction at all for about 24 hours, then he would get blistered and have tons of swelling.

Gerald: Tell me about that day when you actually found out that he for sure had that diagnosis.

Valorie: That was a tough day. It was the 20th of May, and I was home alone with my two kids, my husband was at work. I got an email confirmation that they had found Cockayne syndrome in Jace's cells. I had to contact his geneticist, which started this ball rolling. The geneticist wanted him to have another MRI as one of the symptoms a lot of times is calcification either in the basal ganglia or in the cerebellum. Jace had had MRIs before, but he did not have the calcifications then, but because Cockayne syndrome is progressive, what he didn't have in the past doesn't mean much, so they scheduled an MRI. He had to be sedated because even though he was seven, he was functioning more at a three- or four-year level. He had calcifications in both places, which is another rare thing, as most kids have them in one or the other. That was a punch in the gut you get as a parent, knowing we couldn't fix the syndrome, but asking what we could do to help him. He was very thin, at seven and a half, he weighed 35 pounds. I started pushing for a feeding tube, and that was not an easy road. It was a rare syndrome so doctors had not seen a child with that syndrome, so didn't have a set treatment plan. I finally got the doctors to understand that this kid won't eat ice cream, won't eat candy bars and won't eat cookies. It's not like I'm trying to feed him brown rice when I can't get him to eat, he won't eat anything and he won't drink. I would say to him, "If you want to ride your bike, you have to drink two sips of this." That went on our entire day. He was having a lot of issues because he was so dehydrated and malnourished. When I finally talked, really forced, the doctor to do the surgery, my insurance didn't want to pay for it, so then we had to go through all of the jumping through hoops to convince the insurance company that this was necessary. Even though he was on probably the fifth percentile for weight, we still had to go through a month of meetings with a dietician and following what they suggested, which of course didn't help and he actually lost weight. They suggested that we offer food and if he doesn't eat it, then he doesn't get to eat until the next meal. Well, Jace was fine with that, Jace thought it was awesome that he never had to eat, so he lost weight. He had extreme reflux, so the feeding tube surgery was not just a simple out-patient, he had to have stomach surgery as well, the fundal application of the reflux. Even after that surgery, the doctors wanted to treat him like a typical seven-year-old, and they sent us home with a feeding plan where he was supposed to have eight cans per day. He had a feeding tube for 13 years, and the highest we ever got was five cans. Probably one of the hardest parts of the syndrome is that even the doctors who are trying to help are not sure how to help, and in a lot of ways, their suggestions hurt us. I had to decide that maybe they didn't know best, which was not my natural perspective with a doctor when this journey of ours started out. I believed that you do what the doctors say because they know best. I'm an accountant, not a doctor, and I don't know what's best, but I did find throughout that journey that I know my son best, and I know what's best for him. It took a lot of time, research and spending time with him to know what worked and what didn't. There were several heated discussions with doctors because they're doctors and they know.

Gerald: You probably did actually have more knowledge about the syndrome than the doctors because they really are only going to see one.

Valorie: Right.

Gerald: Or none in their entire practice.

Valorie: Jace's geneticist was towards the end of her career, and she had seen one other child with Cockayne syndrome when she was in her 20s, and it was even a different type. Jace had type one. Type one children are normally born pretty healthy and develop normally until they are about a year old, and then they start to struggle. Jace was born with a small head size, but normal head size by the time he was a year old. It was not even on the chart. He walked at one year old. Between one and two, when they start to combine words, he wasn't doing that. We're very lucky that in our area, we have a wonderful special needs school. He stared there at three and went for 17 years until we put him on hospice and he really couldn't go much anymore.

Gerald: What has been the hardest part with Jace, because he's passed away?

Valorie: Right.

Gerald: He lived until he was 20 years old and passed away just a handful of weeks ago.

Valorie: Right, he passed away the 25th of May, so it has been 12 weeks.

Gerald: So not very long, but in terms of that 20 years, what would you say would be the hardest thing?

Valorie: Watching your child suffer and knowing that you have to allow a doctor to perform things that are painful, but they have to have it. There were so many times that I would say that I wish I could do it; I wish I could let them cut my Achilles' tendon and tear it and cast me, and things like that. That was difficult, but Jace had a very positive way of looking at things. He wasn't a complainer, he didn't cry and whine about the pain. He was a tough guy, so it made that part a little bit easier; however, the progression of the syndrome and constantly watching him lose abilities was difficult.

Gerald: Because it is a fatal syndrome.

Valorie: Right, always fatal, absolutely. At one point, he would ride his pedal bike without training wheels and pop wheelies, and slowly he lost that ability. We got him a trike that he could ride, but he couldn't pop wheelies anymore. But like I said, he had this great outlook where he didn't say, "I don't want to ride the trike because it makes me sad. I want to ride the two-wheeler." He said, "Oh, an awesome blue

trike!" If it was blue, you could get Jace to do just about anything. Jace made a lot of those things easier, but it's tough to watch a sweet person just continually lose.

Gerald: What were some of the joys?

Valorie: Jace had no concept that there was anything really wrong with him. He was only 4'2, but he thought he was a 6' tall man. He thought he was the strongest person ever. Watching him with people and watching callous, closed-off people, they had no weapons towards him whatsoever. He had a way with people that I'd never seen any other person have, this skill where he could instantly melt people. He really had a loving way about him. Most of us think that by the time we get to be about 9 or 10 years old, we start to worry about what other people think and how we look and more of the world view, and he was never affected by that at all.

Gerald: Interesting, and you have two other children.

Valorie: I actually have three other sons.

Gerald: Are they older?

Valorie: The oldest one is 23. He is technically not my child as he is my stepson, but I love him to death. Then Jace, and then I have an 18-year-old, and we adopted a baby when he was born who is seven now.

Gerald: What was the impact of Jace's life on your family, your immediate family as well as your extended family?

Valorie: It's tough, very, very tough. Jace required 24-hour care, and the type of care changed over the years, but he's always required eyes-on. He couldn't be left alone for five minutes, he was a 24/7 kind of kid. Your life adjusts for that, and one of the biggest problems with the other kids was when Jace was in pain or sick, he was the top priority. That meant that the other kids had to be second. It was a struggle, because I am one of those kinds of moms who think that my kids come first. My husband and I didn't go on dates, we've never been on a cruise, we've never been away overnight from our kids. It just wasn't an option with him. He was very much a momma's boy, and if I was within two feet, he was perfectly fine, but if I wasn't, he was very unhappy. One of the hardest things was that the other kids had to learn that they were not quite as important, and that's their perspective, definitely not my perspective. I always read my kids stories and cuddled them to sleep every night, but my kids have grown up thinking that I loved Jace more, and that's hard for me to take.

Gerald: If I came to you just having received a diagnosis that I had a child with the same syndrome and I was seeking your advice, what advice would you give me?

Valorie: There are two organizations that support families here in America, Share and Care Cockayne Syndrome Network, which is wonderful. They have a retreat every year. We've gone to several, but the last five or six years we haven't gone, because it was too hard for Jace to travel. There's one in England called Amy and Friends, which is run by a dear friend of mine, Jane Hughes. I would say the number one thing is to get in contact with these people. They will get you linked up with all of the parents. Number two, keep your kids out of the sun. Number three, don't give them medication unless it's absolutely necessary. They wanted Jace to be on Baclofen at a very young age, but I refused. We did water therapy, physical therapy and essential oil massage, everything we could think of, but a lot of the Cockayne's kids struggle with liver issues and kidney issues, and if you pump your child full of medications, it takes a toll. I would also say to take a breath and try to enjoy today, because when you know the cliff's coming, you brace just naturally. It took me probably about a year to realize that I wasn't making memories because I was so fixated on this disaster that's coming that I was missing out on what we're doing today. I say to all my friends, even the people who have typical kids, take five minutes and sit down in the dirt with your kids. I think it's even more important when you have a special needs child. It's as important to do that with your special needs child as much so as your typical children. Our world is out of control, and we are all running as fast as we can possibly run, but looking back, now that Jace is gone, the things that I remember are those little five-minute things where we stopped and picked sunflowers and silly things. Jace loved pocket knives, and he would say, "I was so good, mom, I was so good, can we go to Cabela's? I want to buy a knife." I didn't want to go, but we'd go anyway, and looking back now, I'm so glad. Those decisions you make when you're tired and really don't want to do it and don't necessarily need to be buying something, just make a memory today, something that's going to stick in your head. The things that I did that I thought made me a good mother are not the things that my other kids remember, you know, making sure dinner is on the table, making sure they're clean and have clean clothes, that they got to school on time and did their homework, and their homework's in their backpack, and their backpack is by the door. Kids don't care about that. I think it's important, but it's not something that's going to help your child to become a balanced, loving adult. That's just kind of the noise in the background. Make sure that you are taking the five minutes that really matters.

Gerald: You got a definitive diagnosis when he was seven. You knew he had issues before, but you didn't have a label for it. He lived 20 years, you cared for him, you saw him progress, you saw him regress, had to battle with insurance companies and who knows what else in those 20 years that you had to deal with him. Was it worth it?

Valorie: Oh, absolutely. The fact that my life is different doesn't describe it, because in those last few years, Jace couldn't swallow, so I developed this almost ninja-like hearing. I could hear him even if he was in the other room if he happened to swallow a little bit odd. He was a silent choker, never made a noise, never moved his hands, he would just be choking. It's difficult to stop that sense. My struggle now is to try to

revert back to a mom of three healthy boys. I don't need to have this super hearing. It's not going back as quickly as I thought it would. As for fighting with insurance companies, I'm an expert at that. If you have a handicapped child and you're told that something's not covered, fight it. I am a force to be reckoned with when it comes to those kind of things. Jace went to physical therapy three times a week for 11 years because I wouldn't shut up. I actually sent a spreadsheet via registered mail comparing a year's worth of physical therapy to a surgery with a picture of Jace to the benefits coordinator of my husband's employer. I didn't just do it once, I did it many, many times until they finally issued an override. I said, "This isn't saving you money, it's not." I would always appeal, and even when the appeal was denied, I didn't stop at that. My son received excellent care in so many ways by not leaving it up to the insurance companies or even the doctors, because a lot of times even the doctors realized that it wouldn't be paid for, so they didn't want to offer it. I'm starting a nonprofit called Team Jace, and the number one thing I would like to focus on is helping people get the equipment that their child needs. A big part of it is just going to be an exchange program, because after Jace passed away, I had an awesome stroller that cost me $7,000 sitting in my garage. I first tried to give it to a friend, but she couldn't use it, so we did this kind of three-way trade where I gave mine to someone else and then she received one, and it worked out great. Get on Team Jace's page on Facebook and contact me, I will help you find what you need, and if that doesn't work, we'll find another way. I think so many parents just feel helpless because you're at so many people's mercy, you're at your child's syndrome's mercy, and the doctors'. What will the doctors do, what will the insurance pay for? It's not up to them, it's up to you and what you're willing to do.

Gerald: Valorie, thank you for taking the time.

Valorie: You are very welcome.