An Orange Socks Story – Gwen: Microcephaly, Asparagine Synthetase Deficiency or ASNS, Dwarfism, Cerebral Palsy, Epilepsy

Interviewed by: Gerald Nebeker, President of Orange Socks

Gerald: I really appreciated the time Gwen took to talk to me by phone. Her first child, a boy, was typical. So, when their second child was born with microcephaly, they were shocked and surprised because they were told there was a 25% chance of having another child with microcephaly. They were watching for any issues with the third pregnancy, and then that baby was diagnosed at 24 weeks with microcephaly. They were encouraged to abort the pregnancy because the medical professionals said they already had enough on their plate. They chose not to. I'm sure you will find Gwen's story interesting and inspiring. It turns out that both babies had asparagine synthetase deficiency, or ASNS for short, and it was ASNS that caused the microcephaly.

Gerald: Thank you so much for taking the time to meet with me over the phone to talk about your two daughters. Why don't you tell me about finding out that your children had some issues?

Gwen: Well, actually we had our first child when my husband and I were 22 and 23. We were pretty young, and our son was born typical, so we had no reason to be concerned that we had any type of a genetic condition to deal with. When we got pregnant with our second child, we found out it was a girl when she was around 19 weeks. We had a normal sonogram. Everything came back completely within normal limits. She was born at 39 1/2 weeks, she actually made it to term. The doctors noticed immediately that her head was pretty small, and she had some clonus with her arms, and they were pretty concerned and called in all the specialists to give her a thorough exam. Because we had her, when we had our third child, we knew that there likely could be a genetic risk. We had been searching for about four years at that point to locate the genes, and there had been none found. Some doctors said it could be an isolated incident, and others said that it was probably genetic. We didn't know what to believe, but they told us to plan on a 25% chance of recurrence. We felt like we could handle whatever came our way, and honestly, we thought it wasn't going to happen again. We did serial sonograms with our third pregnancy with Lola from about five weeks on, and we had a normal sonogram at all of our visits until we got to 22 weeks when they said that "We've got a little bit of an issue." Her head measurements were about two weeks behind, but that could be still within normal limits, they were not sure. Two weeks ahead or two weeks behind is still considered normal, but obviously we were still watching those measurements pretty closely. There was a chance that this could be something starting to happen, and we needed to watch it and track it. They told me to come back in a month, so we went back when I was 26 weeks pregnant, and they said we were either going to see that her head had caught back up and she wasn't going to be microcephalic, or we were going to see that her head was further behind. We would just have to see what the sonogram showed. The second that they put the wand on my tummy, I instantly knew. I didn't have a degree, but don't think that I didn't research it a million times. I knew exactly what numbers to listen for and look for, and I knew instantly that it

happened again. They confirmed microcephaly for her at 26 weeks, and her head was then five weeks behind.

Gerald: What type of advice did your doctors give you with these two pregnancies?

Gwen: At the first pregnancy, we had no reason to believe anything could happen because there was no history on either side of my husband's family or my family, or any type of issues whatsoever, so we didn't have risk factors. I had Claire at a birth-care center, and it was a very normal pregnancy. I didn't worry, I mean I was super cautious and ate organic, didn't paint my toenails and didn't dye my hair. I just I did what I did with Cal. I took my prenatal vitamins, and I went to all my appointments. I was super careful and cautious the whole way, and there was no reason to believe anything was going to happen. I had a pretty uneventful labor, and she was born, and instantly everyone freaked out, so I didn't have any advice with my first daughter.

Gerald: So then, with the first one, it was a total surprise when she was born that she had some issues.

Gwen: Yes, complete surprise.

Gerald: Ok.

Gerald: With the second one, they knew after some testing and found out then that she did have similar issues in utero.

Gwen: Yes, that came at about 22 to 26 weeks into the pregnancy, and perfectly explained the reason our first daughter, Claire, had had a normal sonogram at 19 weeks. We couldn't figure that out after she was born, we looked back at the 19-week sonogram and tried to see. We were working with a perinatologist, and we tried to see if there was something that got missed. Honestly, everything was completely within normal limits at 19 weeks, and apparently all was completely within normal limits for Lola as well, because I had a normal sonogram at 18 weeks with her. So, for us, it happens later in the pregnancy. The second half of the pregnancy is where it became increasingly obvious that we had microcephaly to deal with a second time.

Gerald: Did they counsel you any particular direction with this second pregnancy when they found out you had another child with this same issue?

Gwen: I talked with so many different people that what ended up happening was that we worked with a perinatologist for my second daughter's birth, so we already knew we had this 25% chance, or up to a 25% chance of it happening again, which is why they had us start working with the perinatologist. He was really helpful through the whole pregnancy, very positive and very encouraged by what he saw up until 18 weeks. The kicker is that the 18 weeks happened right before Christmas. He told us at that visit that that would be his last visit with us because he was moving and taking another job across the country. We tried to see if another perinatologist would accept us. I think there was only one other one

in town at the time, and she refused to accept new patients, so we were kind of up in the air. However, the way he left it with us at 18 weeks when everything was still looking good was the best Christmas present you could ever receive. Everything looked fantastic, so we were just happy-go-lucky, thinking everything was great, it didn't happen again, and everything was going to be fine. When we went to our next visit, our OB said that since the other perinatologist won't take you, he'd just have us go to a sonogram lab, and then the radiologist would read the report, and then he would advise us after he talked to him. He said they had done this before lots of times, it really was not that big of a deal, we were still going to be getting expert opinions about what going on. That was a little weird for us. Here we were at a really crucial point of our pregnancy thinking everything's ok, and we go into our 22 weeks thinking nothing's up, it's going to be fine. When we go and they say, "Oh, well, the head's two weeks behind." The first thought I had was that it's different sonogram equipment at a different place, and the sonogram honestly didn't look clear. Scott and I both thought we were looking at Atari graphics or something because it was so primitive and just very vague. I felt like the one we had with the perinatologist was super clear, you could see everything. So, I felt like there was little bit of room for error there. I remember saying to the OB that these sonogram pictures are not that great, and I don't know what's going on. He said, "Let's go back for second opinion." We went to a different place at 22 weeks, and they said the same thing, that they were getting the same readings. Then we thought, "Oh, my gosh, this means that something bad is happening." We tried to see if they'd let us come back in two weeks, because there's no way I can make it a month as that would be the longest month of my life, waiting to see what the next results are going to show. They said that it's going to go one of two ways, it's either going to be better, or a lot worse. That was the worst month ever, waiting and waiting, just trying to get any answers whatsoever regarding if it was happening again. We just tried to stay positive, and when it was confirmed when we went back, it was pretty devastating for both Scott and me. I don't know if I've cried as much in my whole life as I did those 24 hours after Lola was diagnosed. It just felt so unfair to have to go through it again, when we didn't have any reason to believe that it would happen again. They still hadn't been able to confirm that it was genetic. We really, truly thought we had a 75% chance or greater that everything would be fine, so when we were diagnosed, I remember going back to my OB who said, "You guys have been through an awful lot. Look Gwen, you have options, and I'm not going to judge you whichever option you take, but I would encourage you to check out every option." He wasn't pushing anything on me one way or the other, he just said the best advice he could give me is to put myself into both shoes, do my research, talk to people, make phone calls and consider every single option that's out there. He said, "This is your life. You've already got a lot on your plate. You've already got a four-year-old daughter who has a pretty profound condition and adding another daughter with a very profound condition would be life-changing for you. I just want you to feel like you've explored every option thoroughly."

Gerald: You obviously chose to have that daughter.

Gwen: Yes, and I did exactly what he said. I actually live in a town where there was a pretty famous doctor who did perform abortions, who I knew personally because I went to school with his daughters. I knew what kind of a person he was, and I knew that option was one that was certainly out there for me because he was well-known. It just so happened that

when I found out that we were going to have another child, people came out of the woodwork a little bit to say they had been through something similar, and here's the decision we made. I talked to those people and asked questions, and I went to their websites and read the testimonials. I actually called the office, and I remember being just completely horrified. I remember someone telling me there that making the decision to terminate the pregnancy was one of the most "loving things" I could do for that child. All I could feel was that I would be playing God, making the decision to terminate the life of a child who could bring me the kind of joy that Claire has brought me. How was that loving? I could be missing out on a child who ends up doing more than Claire is able to do, or who brings even more joy to our family. Who am I to play God? I was given this child for a reason, why am I trying to change this path? I'll be completely honest with you, I consider myself to be pro-choice, and I always have been my whole life, yet when I was in the situation personally to make that decision, it felt like the worst decision I could possibly make. It's weird how it's your baby who you've loved for 26 weeks, who you feel move every single day, all day every day, and who all of your hopes and dreams are pinned on. It actually wasn't that hard of a decision to make. Both Scott and I felt like it was 100% clear what we should do, and I told the lady off and hung up. We were having this baby, and Scott and I both agreed, and it was like the tides turned. It was like I went from bawling 24/7 to feeling hope. I was supposed to be these girls' mom, and I was finally embracing that fully and being the mom that they wanted me to be.

Gerald: That's a great story. Now tell me what is the diagnosis and how does it manifest itself in your daughters? Is there another type? There are lots of different things that this particular diagnosis causes, tell me about that.

Gwen: Just this past September, we were actually completely shocked when we got results back from Whole Exome Sequencing. We had given blood to two different research hospitals, one was a genetics clinic, and one was doing research. They said that it could be a month, or six months, or five years, or we may never get answers. "We don't know what to tell you. We are hoping that we are going to find something." They called me about three months after we had given blood at Children's Mercy Hospital, and they had said they had found the gene responsible for our girls' condition. We had gone through their whole lives, 15 years, saying, "Okay, our daughters have microcephaly, that is their diagnosis." I still feel like it's their primary diagnosis, but apparently the whole genome sequencing said that the girls have a gene that is affected called the ASNS gene, and it causes asparagine synthetase deficiency. That's a lot of big fancy words, but basically all of the symptoms that my girls have, and then some, are symptoms of asparagine synthetase deficiency. From what I'm learning, it is a spectrum disorder that can either affect the children more mildly, or more severely. Our daughters happen to be more on the severe to profound end. We asked questions at our genetics appointment about other kids who would be similar to our girls, and we were told that the children who were similar to our girls had already passed away, and that the girls were basically paving the way for kids with this diagnosis. They said they would probably be wanting more information down the road about how we had cared for the girls, like medicines, vaccines, diet and treatment methods because they felt like the girls were kind of pioneers for being a little bit more severely affected. Since then, I started a Facebook page for asparagine synthetase deficiency, and we have new members, so

finally I've been able to connect with some other families who have kids who are similar. Pretty much the common diagnosis is microcephaly, which just means small head and small brain. How the brain is affected varies from kid to kid. Some of them have just a small brain that is normal and functions pretty well. Several of these kids that I've met walk and talk, but it's limited, and they didn't do it until they were older. They've had a lot of physical therapy and have struggled along the way. Other children have brains that are more severely affected. They don't just have a normal brain that happens to be small, but also brain malformations, and that's where my girls fit in. They don't walk and don't talk, but communicate in their own way and are working on physical therapy skills so that they can learn to use a walker possibly. Claire, our daughter, has been working on that, and it has been really exciting. They're paving their own way as these kids are doing the same. It's something that is kind of underdiagnosed, like autism. It's going to run the gamut. On outward appearance, they don't appear to have a disability at all like kids who are very profoundly affected and who don't survive the first year of life. Our girls fit in there somewhere towards the more severe end, and some of these kiddos I've met are mildly to moderately affected. They have daily struggles that I can't even wrap my mind around, and we have daily struggles that they can't wrap their minds around, so it has been really neat to connect with these other families and see the similarities. Something else that was really cool is that there was something mentioned to me when we went to the genetics appointment about whether or not my daughters startled easily to loud noises or even to quiet noises, and I said, "Oh, my gosh, this has been something we've dealt with since the beginning. It's out of control, they can hear a pin drop three rooms away and are very, very hypersensitive with their hearing. So, one of the first questions I had to ask the other moms was, "Do your kids do this, too?" They said, "Oh, my gosh, it has been such an issue since she was born." It has been little things like that that we are finding. How nice it is to know another family who understands what we are going through.

Gerald: How old are your daughters now?

Gwen: Claire is 15 and Lola will be turning 11 in about a month.

Gerald: So, you've had many years of caring for your daughters.

Gwen: Yes.

Gerald: You've mentioned several things already, but I'm wondering if you could add some things that have been particularly difficult or hard in caring for your daughters.

Gwen: There are a few things that outsiders might look at and say, "Oh, my gosh, that's huge!" One is epileptic seizures. Most people are absolutely horrified and scared beyond belief to see seizures because to them, it's a sign of the brain malfunctioning. I know that they are dangerous and that kids have died from seizures, and I've watched friends' children die from seizures. It is absolutely horrifying and absolutely terrifying as a parent to see your kid have one. I remember when Claire had her first seizure, she was about three months old, and that scared the crap out of me. I couldn't even function. I was obsessive about writing down every single little thing that happened, like when it started and when it

stopped. I had to get to a clock as quickly as I could so I could time it. I wanted to be able to chronicle every single little thing that occurred with it. Over time, I began to realize how much I was focusing on all of the things that were wrong with my child, and all of the things that the medical community would look at and say that "This is a flaw with your child, this is something that is not right and not normal." Well, every single thing about my kids isn't normal. Let's be honest, we have very few things that are still normal left. I finally realized something with my daughters and their particular seizures that I see currently. I'm not saying that those won't change and that I don't have fears and risks or feel a little shudder, wishing I didn't have to witness those, but I started not making it such a big deal in our lives. For us, our girls were not going backwards, they were not losing skills, they were not regressing, they were not turning blue and they were not stopping breathing. We weren't having to give oxygen or rush them to the hospital. In fact, what we saw after the seizures was a little strange, because what we saw was more awareness. It was almost like we could equate it to their brains trying to work and trying to connect. The messages or neurons were firing, and it was like we saw more awareness and better eye contact. We saw the girls being more interactive after the seizures, so for us, it wasn't such a negative thing. I quit writing them down, and I quit noting all of them. We'd be out in public with somebody, and one of the girls would start to have a seizure or make a sound or smile, that's what our girls do a lot when they have seizures, and they'd say that "She's so cute, what's she doing?" I'd say that she is having a seizure, and they'd say, "Do you want to me to call 911?" I'd say, "No, we just we just ignore them." I mean, we are not ignoring them. We are right there, holding them and making sure that the seizures don't cause them to spit up. The seizures are such a tiny portion of who the girls are, such a tiny part of the whole of who Claire is and the whole of who Lola is, that for us, it was just something that we didn't give as much attention to. That being said, Claire was three months old when she started having them, and Lola started having them around 18 months, and I've been working to try some natural remedies that would help lessen the seizures. We've done better at trying to lessen the seizures using natural things, and that's been really exciting to see some progress, because I feel like sometimes the seizures do interfere with sleep for Lola, and with development for both girls, so that is definitely my mission. I'm continually trying to calm her, because I cannot use prescription medication with the girls, so I have had to go a more natural route, which was more familiar for me anyway, and try to find some answers. So, seizures are one thing I do want to conquer, but I don't fear them now, I just want to optimize my girls' lives.

Gerald: Tell me, what is a typical day for you?

Gwen: There is nothing typical about any of my days, nor are any two days the same. We are typically atypical. I can't say that nighttime is normal because they don't sleep steadily. Claire sleeps a little bit better than Lola, but she still wakes a bunch of times, and she just has the gift of being able to settle herself a little bit easier on her own whereas Lola requires to be held pretty much all night. We do monitor for seizures at night because we see more seizures at night. With Lola, her sleep is definitely an issue. She sleeps anywhere between 3-5 hours average a night, some nights less, some nights a hair more. Six hours is the most I've ever seen her sleep in the last couple of years, and that's not all in one stretch. We are talking an hour or two at a time, and that is a good night. Usually it's 20-minute stretches the whole night, so if we do not have a nurse here to watch the girls, we are

getting no sleep. My husband and I tag-team, and occasionally I'll have a babysitter who can step up and help, but that is not common, so night times are rough if we do not have a nurse here to stay up with Lola. We wake up in the mornings about 8-ish, and Lola's best time for sleep is between 8 and 11 in the morning. We have no idea why she sleeps between 8 and 11 in the morning, and that is not consistent, but on the whole, she sleeps better between 8 and 11 in the morning. The tricky part is we have therapy in the mornings. We have PT, OT, speak technology and we have a vision therapist who comes and two teachers who come, so that's like seven people who are trying to come Monday-Friday, every single day. They all come between 10 and 11-ish, so we usually end up waking her, which usually results in her having a seizure because she's finally gotten into a deep sleep which she couldn't get into the whole night. A lot of times, therapy is a struggle because she is agitated, and we've been known to let her sleep through therapy. If she has been sleep-deprived for a while, we miss it, which I don't like doing very often, maybe once every few months. We usually try to get them involved in therapy between 10 and noon-ish, and then they're exhausted, and they sleep sometimes, or they don't for a little bit afterwards. We usually are holding Lola for the rest of the day and evening. She still eats on a newborn schedule, so we are feeding her every 3-4 hours, and Claire is fed every 2-4 hours. We alternate water and food for her, so in between, we're trying to make fresh food for both of them if we can. I'm also trying to help with our business. My husband owns his own business, and I help run the business three days a week for a few hours, maybe a total of 13 or 14 hours a week. I have to cover myself with a babysitter so I can go help with the business, and then get back home. We try to squeeze in stuff for our son. A lot of times in the evenings, he'll have basketball games or practices, or we go with him to the gym. We hang out as a family and watch TV or go somewhere to eat dinner. We try to have as much normalcy for him as we can. We've always tried to do that. We are a pretty close-knit family, we just like spending time together.

Gerald: Maybe a little easier question would be what are the joys that you've experienced with these girls?

Gwen: There are a lot more joys than I thought were possible. The joys have come in little packages, like things you wouldn't expect, in stones not milestones. Like I've said before, there is joy in just seeing increased awareness, seeing increased eye contact, seeing the girls respond to their environment and to us, seeing them smile at things that we say that are funny and they have an appropriate response, and seeing them progress super slowly with their therapies. I know most people would look at it and say they don't see any progression as they are not walking or crawling, and they are not sitting up independently. I get it, but when you have a child who for 15 years couldn't sit and then to have trunk control on your lap with their body and their legs at 90 degrees on your lap, and you're able to hold their hips, and they are able to hold their heads up, maintain good head control, look at their environment and interact with their environment, and make choices with their heads or hands, that's huge for us. Most of the world might look in and say that they aren't doing what matters, which is walking. Claire has been in her walker, and she is figuring out that she can move herself now. She is not taking steps or running, and she is not trying to get from point A to point B at this point, but she is learning and figuring out that "When I push, I move, and when I move, they cheer for me, and when they cheer for me, I smile,"

and this is fun to see that she is progressing in that way. Lola is a little slower than Claire right now. The gains we are seeing with Lola are more cognitive than they are physical, and she's paying more attention to her surroundings, like passing her from me to our babysitter Tracy, and seeing her looking at her face. Then Tracy says, "Hi, how are you?" And she smiles at her. For us, that is such a joy to see, because for a decade, we didn't see anything like that. We are starting to see the girls making connections. They are both working on a program called the Eagle Eyes, which is a communication device. It is able to tracks eye movement, and the girls are able to make choices on the screen by making eye movement. We are hoping that will open some doors for them as well, which we are starting to already see with their being able to activate a target by looking at a target which causes a song to play, or something funny to happen, or a face to appear. They are getting cause and effect, and that's been really exciting to see, too.

Gerald: So, tell me what impact have these two girls had on your family, or immediate family, friends and neighbors?

Gwen: It has been pretty exciting to see our whole community band together to accept our family, and I don't mean our immediate little town. We live in a suburb of Wichita, the biggest city in Kansas, that has about 450,000 people, and people in our community know the girls. We sold some Claire and Lola bracelets to try and raise money to buy the Eagle Eye program, and I see people wearing those. I see people posting pictures all over the world from a lot of different countries, and that is really exciting. Our community itself, like the school that our son attends, has completely embraced our family and our girls. They have basically opened their arms to us. People see us all the time and ask us questions. They are happy to reach out to our girls. They know who the girls are, and they're proud of them. We try to get them up on the jumbotron when we go to local basketball games. The girls are well-known around here, and I love the fact that people aren't afraid to come up to us and ask to talk to the girls. They aren't afraid to reach out and talk to the girls and touch the girls and talk about things that they saw because the girls have been in the media a little bit. People have seen those interviews and are really touched by the girls' lives and inspired by their desire to be here and to give hope to other families who are in similar situations. It has just been really, really neat to see everyone come together. The bond has definitely been strengthened in places and weakened in others, and that's not all a bad thing as its kind of weeds out people who maybe can't handle it or aren't okay with it. While that's hard to see, it's also one less person for me to have to deal with who can't handle what we're doing, and if they can't, then I don't need to be around that negativity anyway. It has been really enlightening for all of us, but I would say overall, the resounding emotion has been support and love for our girls. My whole takeaway message of sharing the girls' lives is for other people to not be afraid to reach out to special-needs kids in their communities and their families. It feels good when people embrace our kids who are different, because many people are really good at elbowing their friend, pointing and whispering, and making awful comments. We don't pay attention to any of the negative, we focus on the love, because there's so much of that, and the support, and the stuff that we've gotten from not only the community, but our friends and family. It has united us more, if that makes sense.

Gerald: It does. Now let me ask you another question. If I came to you with the results a of a prenatal checkup where the diagnosis was similar to that of your daughters', what advice would you give me?

Gwen: I wrote a blog post on this very thing, maybe a year or so ago, and I am actually confronted by parents like this all the time. I'm probably talking to two or three right now about this very thing. I hear, "Oh, my gosh, my kid just got diagnosed with this, their head is measuring small, I'm scared to death, I want you to help me," and it feels really good to me to be able to reach out and give these parents what I wish that I had at the same point in my life, just tell them that they can do this and that they've got this. It's scary as hell, I'm not going to lie. It feels like so much unknown that you almost don't know how to move forward. You feel like you can't trust your "mommy instinct." You feel like your choices have been taken away, almost like you just have to do whatever the doctors tell you to do because it's such a scary thing and nobody knows what's going to happen. You've got to really lean on somebody else instead of trusting your own gut instinct. I just want people to know that they can trust their gut, and they can trust their heart. We still are the people who know them the best, they were in our tummies for months, and we loved them before anyone else even knew them. We are so closely tied to who they are, and it matters so much to us, and I think we need to absolutely trust our heart in this whole process. That's what we tried to do back in the day, but it was scary, and I remember feeling like I couldn't do it. I just wish that somebody had told me that there were a lot of kids who do defy the odds, that there are people who have microcephaly who are into their seventies and eighties. I wish someone had told me that you can have a child who is very profoundly affected who could live a quality life, being an important member of society, who is valued and respected and loved and cherished without ever having walked a step or spoken a word. I wish someone would have told me that back then, that I could still be happy, and that I could still find joy in the life that I had, that maybe there was a chance that this life wasn't an accident and that I was supposed to be this mom. I just wish somebody had told me that in the beginning, and I try to really make that point to these new moms, that this life is special and perfect and wonderful exactly the way that it is. Having these kids and thinking outside the box and not doing things exactly the way that you would with a typical child is basically upping your mommy game. It's making you a better person, making you stronger, uniting your family, and helping your kids to know how much they're loved and how important they are to you. I just really want people to know that they can do it and that they just need to believe and love and find joy in every single day.

Gerald: That's terrific advice. One last question…

Gwen: Sure.

Gerald: Any regrets?

Gwen: Zero regrets. I would do it exactly the same way a million times over again.

Gerald: Thank you very much.