

[MUSIC “Look Inside” by Sirius Music]

KATE: Welcome to Caring for Rare: Stories of Rare Metabolic Disorders- a podcast that explores what life is like with a rare metabolic disorder. On this episode, we're exploring phenylketonuria, or PKU, with two moms of young children. Though these disorders are rare, you can find community and support, you are not alone.

JORDANN COLEMAN: We went from blissfully happy to it felt like everything was crashing down.

LACY SHAFFER: I felt like a first-time mom again because now I have a child that has a disorder that the entire world isn't familiar with.

COLE SULLIVAN: We've got to stop talking about what we can't do and start talking about what we can do.

JULIE BOLDUC DEFILIPPO: Seeing Johnny hit his milestones was very reassuring.

CARTER COLEMAN: You just can't have certain foods, but you can do normal stuff.

JORDANN: I find it therapeutic to have other people that understand our life.

SARAH SULLIVAN: We could do this, like this is something that we can do.

KATE: Caring for Rare is brought to you by Nutricia North America, a manufacturer of formulas and low protein foods for inborn errors of metabolism. We're proud to have served the metabolic community for more than 30 years. I'm your host, Kate Miley, a member of the Nutricia team since 2019.

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In each episode, we'll hear a family's personal experience with a rare metabolic disorder. Nutricia has partnered with these families to bring you their stories. I just want to note that this podcast may not be representative of all rare metabolic disorders, and this in no way replaces the care, advice, and medical supervision of your metabolic healthcare team. The following content is for informational purposes only, but we're hoping you'll find comfort and connection in these stories.

Today, we're listening in on a conversation between Lacy Shaffer, and Julie Bolduc Defilippo. Lacy is a stay-at-home mom from Louisiana, and Julie is a professor of social work from Massachusetts. They'll be talking through their families' journeys with phenylketonuria, or PKU. This is the very first time they've talked.

LACY: I'm Lacy Shaffer. I have a daughter, Aspen Shaffer, who is two and a half years old that was diagnosed at five days old with phenylketonuria.

JULIE: My name is Julie Bolduc Defilippo. I have a son who's five years old. His name is Johnny and he has PKU.

KATE: For those unfamiliar, PKU is an inherited genetic disorder - both parents must be carriers of PKU or have PKU themselves. And neither Lacy and her husband, nor Julie and her husband knew they were carriers. There's typically no indication before birth and there is nothing the families could have done to prevent it. Here - I'll let Julie explain the rest.

JULIE: So PKU is a condition where people are lacking a certain gene that allows them to process protein or more specifically phenylalanine, that's found in protein, and if they're given too much protein over time, it will lead to severe cognitive impairment.

KATE: To date, there's no cure for PKU, but with a diet low in whole protein as well as special metabolic formulas it can be successfully managed. Both Lacy's and Julie's kids who have PKU are thriving.

JULIE: Can you tell me about Aspen?

LACY: Oh jeez. PKU aside, she's a beautiful disaster, can I tell you. She has the biggest personality ever. She was born six weeks early, actually. She had to come out swinging. You know, her first breath was to fight. So that's what she does, PKU or not. It's hard to even see the PKU because of all of her other things, you know, that makes her so great. So that's Aspen. Tell me about Johnny.

JULIE: He's very smart, sometimes a little too smart for his own good.

LACY: Right.

JULIE: He does karate, piano, and hockey. He wants to be a ninja and a social worker, like his mom, when he grows up. Johnny was born early as well. I've always thought of Johnny as a fighter as well. And PKU, it's just like one little tiny part of him now.

LACY: Right on. I agree. How did it feel when you got the diagnosis?

JULIE: The world just came crashing down. We were just so scared, I was crying. We felt like we were in good hands but we were really just unsure of what this meant for his future. And I'll always remember the doctor said to us, you know, every hope and dream you've ever had for your child is still going to come true. And that really stayed with me.

JULIE: So we went home that night and the nutritionist had given us her cell phone and we gave Johnny his first little bit of formula and he coughed it up. And as new parents, we just, we didn't know what to do and the fact that we now had this baby with a condition and we didn't really fully understand it. We called the nutritionist at like 10 o'clock at night and we were like, we think he's allergic to the formula. And she's like I've never had a baby allergic to the formula. He probably just, normal cough up. And we had a lot of questions and a lot of emotion. And she walked us through at like 11 o'clock at night on the phone with us for like an hour and a half.

LACY: Even though I had been one already, I felt like a first time mom again because now I have a child that has a disorder that not only I don't know much about, but the entire world isn't familiar with. And then the isolation. Ugh, I don't know about you, but any time I had to tell someone she had PKU, the long-winded explanation that you were having to get ready and prepare yourself for after. Oh, what's that? Oh, she'll outgrow that. No, she's missing an enzyme. Oh no sure oh, is it one of those vegan things? I'm like, yep, we're over here just making our two-year-old trend with the vegan diet. Come on, you know, it's a lot to try to do right.

JULIE: Right.

LACY: And there's no right way.

JULIE: Was there a turning point for you when things got better?

LACY: I think for me, it was when I started seeing her become an actual human and not a baby. When I saw her start interacting with us, with her brother, with other people, when I would tell someone and they're like, wait, that baby don't have anything wrong with her. Like, I needed to be slapped in the face with the only one making it about PKU at this moment is you.

JULIE: Yeah, I definitely think, like seeing Johnny hit his milestones was very reassuring, and I think to a PKU parent it becomes even more meaningful. I actually got on camera the first time he sat up and you would have thought he got into Harvard how excited I was. And he was just sitting up and laughing and I was like over the moon and every milestone—

LACY: This kid's going places.

JULIE: —has been like that. Yeah, exactly. Every milestone that he continues to hit it's just more reassurance that he is going to be just fine.

LACY: Does Johnny have any siblings?

JULIE: Yeah. So we have a three and a half year old who does not have PKU.

LACY: Okay, so we're super similar because, in reverse, I have a six-year-old son who is non-PKU. How do you manage it with having the three-year-old not having PKU?

JULIE: So my three and a half year old, he's very picky. He only eats meat. So it's like complete opposite kids. I have one that will only eat bacon, steak, and like roast beef and the other who can't have any of that. So I think the time consumption is tough because everybody's eating something different, which takes a lot of time to prepare. We also noticed that Johnny felt very left out. So when David started eating those foods, he would ask a lot of questions. He felt isolated. So we just talk with him honestly about that and that it's okay to have those feelings. And it's hard, as a parent, like feeding your kid is like the most basic thing that you can do as a mother. And like, I can't feed my kid the way I want to. And I didn't even fully understand, like, the way I was supposed to at first.

LACY: Oh, it's so scary.

JULIE: Yeah, it was so scary. And I felt like that was one of the basic things that we do. That's how we care for each other.

LACY: So what has been the most helpful for you to manage your family life?

JULIE: I, I'm somebody that tends to do everything like I take on the world and I try to do everything all my own and try to manage it. And I had this expectation of what parenting would be and what type of mother I would be. And so letting go of that and asking for help and letting people in really helped a lot, especially with the emotional toll that everything had taken on me. So letting my husband help even, because at first I wasn't even letting that happen. And now it's about 50/50 in the management. So it feels very doable because we both take on some part of it. That definitely helped a lot.

LACY: Do you have any friends in the PKU world that you've connected with in any type of way?

JULIE: Yeah, so I found a lot of support on social media through the PKU parenting groups

LACY: Umm hmm.

JULIE: And then through our local chapter called the New England Connection for People with PKU and Allied Disorders. They do like an apple picking every year and a few other events. And it was really helpful to go to those events because people would bring their college-aged kids with PKU. I could see somebody like in the future, super successful, totally well-adjusted, and they have PKU. I was like, okay, like I can get my head around that and it gave me a lot of hope.

LACY: So what advice would you have for the new parents just getting this dreaded phone call?

JULIE: Give it some time. It gets better. And to ask for help. It's okay to ask for help from family members, from your spouse, from your team as well. You can't possibly know everything about PKU the first day you find out your child has it, but you will over time, you become an expert.

LACY: Agreed. I spent my whole first year of new baby love that you get worrying over trying to perfect something that's imperfect because you're going to see it's going to be okay. It's going to be okay. It might not be great all the time, but parenthood isn't great all the time anyway. And we just take our thing as part of us and we, we live life and these children are going to just do that, they're just going to live life and love it.

[MUSIC - "Sunbang" by Elephant Music]

JULIE: I almost wish that our children could see themselves through our eyes. Johnny is my hero and I want him to feel like he can overcome any barriers that are put in his way, whether it's with PKU or with any obstacle in his life. PKU is just one little piece of it. So I have no doubt that he's going to do extraordinary things.

LACY: My future hope for Aspen is that she is proud of who she is because I'm proud of who she is.

KATE: Thank you so much to Lacy and Julie for sharing their PKU stories with us.

Nutricia is here to support families like yours and theirs. We're proud to provide a wide array of metabolic formulas for PKU and other conditions.

And our Nutricia Connect platform provides resources like this podcast, reimbursement support for our metabolic formulas, and additional support like low protein recipes. Join us at [NutriciaMetabolics.com](https://NutriciaMetabolics.com).

We hope you'll check out our other podcast episodes to hear more first person stories from people living with rare metabolic disorders. Find them on [NutriciaMetabolics.com](https://NutriciaMetabolics.com) or your favorite podcast platform.

Caring for Rare: Stories of Rare Metabolic Disorders is brought to you by Nutricia North America. It was produced by Alletta Cooper, Ulrike Reichert, and me, Kate Miley. Thank you so much for listening, and be well.

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