104 FOOD

KATE MILEY: 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 talking about adjusting to a special medical diet with four families living with inborn errors of metabolism. 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.

KATE: In this episode we're exploring how four families learned to adjust to their children's medical diets. 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

KATE: First we hear from Lacy Shaffer and Julie Buldoc Defilippo who both have young children with Phenylketonuria or PKU.

> LACY: My name is Lacey Schaefer, my daughter, Aspen, two and a half was diagnosed with phenylketonuria.
> JULIE: And hi, my name is Julie Buldoc Defilippo I have a son who's five years old, his name is Johnny and he has PKU.

KATE: PKU is a rare genetic disorder that makes it difficult for the body to metabolize protein. In a previous episode, Lacy and Julie talk about understanding the diagnosis. 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. But adjusting to that diet can be a shock for a new parent.

JULIE: 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. So, yeah, it was so scary.

LACY: I was nowhere near educated enough to know how...I thought it was meat, eggs, dairy. Whoa, no, no, no. It's flour. It's chips. It's just every single thing that's in the world and everything is surrounded around food, holidays, events, birthdays, everything.

JULIE: So for the first year, Johnny didn't go to daycare. He stayed with my mom, so we had to trust her. And at first I would like make the formula and send it and just tell her the amount that he had to drink. And finally my mom was like, I could make it. I just had to tear the bandaid off. So we bought her a scale and she learned to make it. And that that became a lot, a whole lot easier.

LACY: I would go down to the grain on the scale, I wouldn't let anyone else touch it. My mom, if she did mix the formula, she would have to take a picture of the scale and send it to me before I allowed her to give it to her. And I just got very, you know, over zealous about every single aspect of it. And now if I was to have another one, I would say you're getting six scoops, let's go drink your milk. I cannot tell you the countless hours I spent weighing things over and over again because I didn't trust myself. I mean, some things really are small potatoes, no pun intended, but they really are. Some things are just small potatoes. Things have become a second nature.

JULIE: It just becomes second nature.

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 David, 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.

JULIE: The time consumption is tough because my husband and I eat one meal, Johnny eats a different meal and David sometimes will have ours. But typically I'm cooking something different for David. And I think the hard part comes with like the medical food and what he likes, like, he tends to eat the same things a lot. And so I feel bad about that, like that he doesn't have much variety in his diet. And so it's just it will take him hours to drink like four ounces. And so that's our struggle. So we have to kind of sit him down and like give him around, like take a sip and coach him for hours on drinking his

formula. It's exhausting and it does make me feel guilty because it's taking him away from playing and being a five year old and being a child.

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. So letting my husband help, because at first I wasn't even letting that happen. I think sharing the burden has definitely been helpful with the management of PKU. I basically handle all the food and writing everything down and he handles the formula.

LACY: So what advice would you have for the new parents?

JULIE: I would say give it some time, it gets better and to ask for help, it's OK 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. And so I would say just give it some time.

[MUSIC]

KATE: The Sullivan family had a similar experience adjusting to their new reality. They have two children with a different metabolic disorder, homocystinuria or HCU.

COLE: My name is Cole Sullivan

SARAH: And I'm Sarah Sullivan, and we have two children with HCU Colby, who is 11 years old, and Cayle, who is nine years old.

KATE: HCU is another rare metabolic disorder in which the body cannot break down protein. The diet management is similar to PKU, in that individuals have to limit foods high in protein and in many cases consume a special metabolic formula. Unlike PKU, HCU is sometimes diagnosed later in life. Colby was diagnosed at age 3 and Cayle when he was 1.

SARAH: It's just added a layer of difficulty for a lot of situations and made us have to think outside the box when we go to do things or have holidays and meals. And it's just made it a little bit more challenging, I think. But I think there's a lot about parenthood that's challenging that when you first find out you're going to have a baby, you don't realize.

COLE: when we're having the baby, everything you think is just like, oh, everything's going to be great, they're going to be snuggled up and everything's cute, you know? And in reality, it's it's a lot of work. It's a lot of physical work as far as the sleeping, getting them to bed, the feeding, and then with the added complexity of homocystinuria, you know, it's a mental grind to being a parent because you you are not only making sure

that you're being a parent for the kid, but you're also got their health at the forefront of your mind, their diet at the forefront of your mind.

COLE: After they were diagnosed, we didn't we didn't go out to dinner. We didn't go out to eat. We didn't go to functions. We stayed home. We stuck around the house, our comfort zones where we had meals and we had the food that the kids could eat. Because we were trying to emulate everything that we ate.

COLE: I remember times when you would have Mac and cheese, which was Colby's favorite food. I remember you leaving the room.

SARAH: Yeah, I would go in another room or in the closet to eat because I just couldn't I just couldn't bear to eat around them. It just broke my heart.

COLE: And we kind of had this, like, epiphany of, you know, as soon as these kids see the people that they know and love eating different foods, when they go to school, when they go to birthday parties, when they go elsewhere to people that they don't know as well, they wouldn't be so honed in on what they couldn't have. It was just the norm. Mom and dad eat this food and I eat this food and it has actually paid off for us. They know that they are going to have different food than what mom and dad have.

COLE: I think as we went through the grieving process and learned more and more about it and became more comfortable with some of the foods and know that you, we can still make foods for them that they enjoy and that are sometimes like ours and sometimes not. And they're OK with that, that we became in our new normal. And that's what we always like to call it, that we're comfortable in our new normal.

SARAH: And we had a great dietitian that I could call and email any time of the day. And she really helped us through a lot of things like how do I how do we approach our first birthday party and different things like that. So I think also, just like with anything, after you get your first holiday under your belt, your first birthday party, your first birthdays, once we got all of those things done, it just felt like we could do this. Like this is something that we can do and it's not the end of the world. And we're we're going to do it.

KATE: Now we hear from Jordan and Andre Coleman who have an 8-year-old son with Maple Syrup Urine Disease, or MSUD. You can listen to a previous episode with the Coleman's to learn a bit more about MSUD. Management of MSUD also includes a diet low in whole protein plus metabolic formula.

JORDANN: My name is Jordan Coleman.
ANDRE: And I'm Andre Coleman.
JORDANN: And we are parents to Carter, who has maple syrup urine disease.

JORDANN: Fortunately for us, our metabolics team had given us, I guess, or to try to just talk to us about what life would be like in the future and that being on diet, he could lead a kind of normal, quote unquote, life. And so once we kind of got once he gotten to a point where I wasn't worried about whether or not he was going to live and I knew he was going to live, then it was like, OK, what are we going to do to make sure that he is as happy and healthy as a kid is as he possibly can be.

ANDRE: You know, it's a modified diet. Again, there will be some bumps along the way, but it's a modified diet and there are certain foods that I don't like.

JORDANN: So meal times are...I'm basically a short order cook for Carter while trying to make something for the rest of the family. So a lot of times I try to mirror some of the similar foods. So if we're having pasta, I'll try to make pasta for him or we're having burgers and fries, I'll do a veggie burger or a low protein burger for him with French fries. So I try to keep it somewhat similar, but I am having to make two meals depending on a side dish that we're having. If it's the vegetable, he can he can have that. But there are there are times where I get started on the dish for the four of us. And then we got 15 minutes left and I'm like, oh, crap, I haven't made anything for him yet. And then I'm trying to scramble to get something done. So a few more dishes. There are times where I'll spend a weekend preparing stuff in bulk to make it easier to make. But I mean, he kind of has like a lane that he likes to stick in for food. So he's not super picky, but he has the stuff that he likes. So we know that there is always something that we can pull into that we just want to step back a little bit to be able to enjoy it.

ANDRE: We have three kids, they vary in age from 10 to eight to two. We have Carter, who has maple syrup urine disease. And that requires, you know, certain attention from your daily routine of preparing his formula to monitoring his diet throughout the day. But at the same time, we have to be mindful of our other two kids as well who, you know, don't have maple syrup urine disease, but require the same focus and attention as any kids would require of their parents.

JORDANN: Carter's sister has been amazing. She's aware of his diet restrictions, I think at times she encourages him when he has more formula to drink or his daily formula to supplement some of the proteins that he does not get through his regular diet, and then there are times when she encourages him at the dinner table to eat certain foods or to finish food that is on his plate. So she's aware, I personally believe that she looks out for him to make sure that he's sticking to his diet.

JORDANN: I would tell a newly diagnosed family that eventually, things will be OK, there is a really steep learning curve in the beginning because pretty much one hundred percent of the time nobody has ever heard of MSUD before you get a call about the diagnosis. So it's scary, especially when you know what some of the severe

complications are. But with proper diet and drinking formula, kids really can lead normal lives.

KATE: Thank you so much to Julie Buldoc Defilippo, Lacy Shafer, Sarah and Cole Sullivan, and Jordann and Andre Coleman for sharing their 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 inherited disorders. 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.

KATE: 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 or your favorite podcast platform.

KATE: 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.