[MUSIC “Look Inside” by Sirus Music]

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 with an 8-year old and his parents about living with maple syrup urine disease, or MSUD. 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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KATE: 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 the story.

KATE: Today, we’re talking with the Coleman family about their journey with maple syrup urine disease, or MSUD.

JORDANN: My name is Jordann Coleman.

ANDRE COLEMAN: And I'm Andre Coleman.

CARTER COLEMAN: And uh my name's Carter. I have MSUD.

KATE: Carter will be back later to talk about what it’s like as a kid with MSUD. But for now we turn to his parents to better understand how MSUD has changed their lives. Jordann is an insurance broker, and Andre is a maritime executive. They live in the San Francisco Bay area with their three children.

JORDANN: MSUD is what's called an inborn error metabolism, and what that means is that the body cannot properly process or metabolize protein and it's not the full protein, but it's an amino acid that is found in protein. And so because the body cannot process

these amino acids, they build up in the blood and can become toxic to the brain.What can happen is if there is too much of a buildup, it can cause brain damage, which can lead to mental impairment, or it can in severe cases lead to coma and also death.

KATE: Fortunately, nutrition management, including special metabolic formulas, are available to help manage the disorder and prevent many of these problems.

Maple syrup urine disease is an inherited, genetic disorder. Both parents need to be carriers for their child to have MSUD. And neither Andre nor Jordann knew they were carriers. There's no indication before birth, and there is nothing the parents could have done to prevent it.

JORDANN: In the womb, Carter was so active. He kicked and moved all of the time. To see him and to hold him for the first time, I was just happy.

KATE: It was the day before Thanksgiving, Carter was just 4-days-old, and Jordann and Andre were excitedly introducing him to their families. Then their pediatrician called. Something came up on Carter's newborn screen.

JORDANN: We went from being just blissfully happy to it felt like everything was crashing down on us. This can't happen to us. We're two healthy people and there is no history of anything in either of our families. So this can't, this can't be right.

KATE: Thanksgiving dinner was cancelled. They immediately piled into their car with Andre's mom, their 2-year-old daughter, and Carter to make the hour-long journey to the nearest children's hospital.

JORDANN: I remember sitting in the back in the car seat next to Carter and he's sleeping. And I'm just looking at him and thinking, what is going on in there, buddy? And just hoping and praying that he didn't have it and that we were going to get there and this was going to be a big misunderstanding.

ANDRE: And I think when we arrived at the hospital and were basically pushed up front in the emergency room, it was a feeling of, okay, this is serious and are we on borrowed time with Carter?

JORDANN: We had to completely just immediately shift from hoping that nothing was wrong to being like, okay, now we have to put together a plan of attack. Is there anything that brought you comfort in the first few weeks after his diagnosis?

ANDRE: While Carter's disease is specific to him and to us – for me, it was an understanding that almost every individual has some type of challenge in their life that they face in some different shape or form, whether it's external, internal influence or environment, whatever it may be. For me, it was like, okay, what is it? Let's learn about it and then let's move forward with living with it as we understand how to treat the disease. So what brought you comfort?

JORDANN: You know, everybody that we spoke with, friends, family, had never even heard of MSUD. Although they were sympathetic we had no common ground. So we actually felt isolated in the fact that I could not talk about this experience with anybody other than you. And we found a Facebook group for MSUD families. And, um, seeing how other families were living their lives made us realize that, okay, it's not going to completely upend the way that we live. It's going to be different. We're going to have to make adjustments, but it's not like we are going to, our whole world is going to be

completely turned upside down. I find it therapeutic to have other people that understand our life, the good and the bad.

ANDRE: I do agree with you that joining that Facebook group and just you see those who have lived with it longer and seeing the normal life, it's one of those things where it is what it is. There will be some bumps along the way, but it's a modified diet and there are certain foods that I don't like, so it's not the worst thing in the world.

JORDANN: One of the impairments that can happen with having MSUD is cognitive delays. Is there anything specifically that happened in Carter's life that made you realize that he was okay?

ANDRE: I will say, early on I thought he was a lot less vocal than his older sister when she was an infant slash toddler. I can't pinpoint the exact age or moment in time, but at some point in time I'd say around two to three years old, his level of communication increased immensely. And to this day, he loves to talk, uh, he is very loud.

JORDANN: It's funny, there's times that we might be at dinner and he just, Carter just will not stop talking. And we just look at each other and we go, remember when we were so concerned that there were cognitive delays and he wouldn't talk and now he will not shut up? [LAUGHTER] If we would have known this back when we first got the diagnosis, we could have saved ourselves a lot of stress. [LAUGHTER]

[MUSIC - “Imaginary Place” by L-Ray Music]

CARTER: My name's Carter uh I’m in second grade. I am eight years old. My favorite color’s gold and black. I like playing sports and I have MSUD. Being a kid with MSUD is normal, uh, you can’t eat certain foods, but like, you can do lots of other stuff. What’s your favorite thing about me?

ANDRE: My favorite thing is your level of energy. [LAUGHTER] I've never seen someone who, uh, when they start their day, is at 110% throughout the day until they finally put their head down on their, on their pillow and go to bed.

JORDANN: Carter, my favorite thing about you is your charm. Probably since the day you were born you have had the ability to instantly have people like you. There is something about you that is just magnetic and it's something that I have loved watching develop in you.

ANDRE: Carter, what are your dreams for yourself?

CARTER: To play football, do fun stuff, and yeah, to have a house.

[LAUGHTER]

CARTER: Is there anything you want to say to me that you never said to me before?

JORDANN: Yes. Um, Carter, I think you are a miracle. You make me proud every single day, and you truly are a light in my life.

CARTER: Hmm. You?

[LAUGHTER]

ANDRE: Um, let's see. I just admire you and appreciate your day to day approach through life and through living with Maple Syrup Urine Disease. Even when there are downtimes, if you're not feeling well, your ability to rebound and your drive to get back to just your normal routine, it motivates me. So, I appreciate that about you.

JORDANN: Carter, if you met some parents who just had a baby and they found out that the baby had MSUD, what would you tell those parents?

CARTER: That it will be okay. It's normal and you just can't have certain foods but you can do normal stuff.

JORDANN: So I think my advice for new parents is that you will walk on a tightrope in the journey. Eventually, things will be ok. And it's completely overwhelming in the beginning but it becomes part of how you live your life, but it does not dominate our life. And so you become an expert tightrope walker.

[MUSIC - "Imaginary Place" by L-Ray Music]

KATE: Thank you so much to Jordann, Andre, and Carter for sharing their MSUD story with us. Nutricia is here to support families like yours and the Colemans. We're proud to provide a wide array of metabolic formulas for MSUD 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.

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.

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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