[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 about homocystinuria, or HCU. We'll hear from a family that has two children with HCU, including from the siblings themselves. 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.

In this episode, we’re talking with the Sullivan family, who have two children with HCU.

COLE: My name is Cole Sullivan.

SARAH: And I'm Sarah Sullivan.

COLBIE SULLIVAN: My name is Colbie, I’m 11 years old.

CAYLE SULLIVAN: My name is Cayle, I’m 9 years old.

SARAH: And we are going to share our HCU journey with you. So HCU is homocystinuria, and it is a rare metabolic disorder in which the body cannot break down protein. So the protein builds up in your body and can cause adverse reactions, such as blood clots, stroke, and various other things. So they can only eat foods that we supply for them and we have to count how much protein they have each day.

KATE: What Sarah was referring to – the build-up of protein in HCU – is specifically the build-up of two amino acids, which are components of protein called methionine and homocysteine. To date, there's no cure for HCU – but by taking certain medications plus following a diet low in whole protein, and taking special metabolic formulas, HCU can be successfully managed.

Colbie and Cayle will be back in a little bit to talk about what life is like with HCU, but now we turn to their parents to better understand how the disorder has changed their lives. Cole is an account manager for a software company, and Sarah is a registered nurse. They live outside of Bozeman, Montana and they were both so excited to welcome their first child, Colbie, into their family.

> SARAH: It's just instant love at first sight. I think I've never loved someone so much so quickly. Like you just love them right away. Colbie came really fast. She was normal and perfect and everything was going great.

KATE: All of that changed shortly after Colbie's 3rd birthday. At first, they thought she had a stomach bug, but it never went away. An MRI showed she had bleeding in the brain and a large blood clot. She was life-flighted to the nearest children's hospital where she spent a week in the ICU. A month later, they got the diagnosis: Colbie had HCU.

> COLE: The doctor just kind of pulled us in and it was just very unemotionally delivered. This is what your kid has. This is how your life is going to change. And it was a crash course in an hour.
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> SARAH: It was really hard. Like I felt like somebody stripped away my healthy, normal kid that I thought that I had.

KATE: Many inborn errors of metabolism are first identified through newborn screening, and then diagnosed with follow-up testing. HCU though is sometimes missed on newborn screening because some babies' blood methionine levels aren't high enough to be identified for further testing. That's why in some instances HCU is actually sometimes diagnosed later in life when a child or an adult begins to show symptoms. There's no indication before birth and there is nothing the parents could have done to prevent it. After Colbie's diagnoses Sarah and Cole had their one-year-old son, Cayle, tested as well. They found out that he too has HCU.

> COLE: How did you feel when we had Cayle tested?
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> SARAH: I just remember breaking down in tears and of course, right when I got off the phone with the doctor, then I called you. And I, I had to leave work for the day, I just couldn't -- we actually left our kids at daycare for a little while so that we could just kind of process the information before going and getting them.
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> COLE: We were definitely grieving. We took it really hard.
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> SARAH: There's so much unknown about homocystinuria and for a parent especially to not know what your child's future could really hold was so hard for us. You just want your kids to have the best life that they can have. And if you don't know that they are going to be able to have kids someday or if they're going to have to be on blood thinners for the rest of their life and not do activities; limitations is such a hard thing to have for your children. So I think it was all of those things. Grieving that you're not going to have a

normal Thanksgiving dinner or Christmas dinner or Easter egg hunt and, and have hard boiled eggs, like all our traditions and holidays are all focused around food.

COLE: You go to someone's birthday party, it's a pizza party and ice cream. And we, we concentrated, we really honed in on everything that we wouldn't be able to do. That was part of the grieving is this is really messing up our lives because we can't do this and we can't do that. I remember times when you would have mac and cheese, which was Colbie's favorite food. I remember you leaving the room.

SARAH: Yeah, I would eat in the closet. I had a lot of guilt. I just couldn't eat in front of Colbie or Cayle, like something that I knew that I would normally share with them or feed 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. And it was like as we moved through that grieving process, we finally realized, okay, we've got to stop talking about what we can't do and start talking about what we can do. And it was when we finally got that mentality of what the kids can do and started to plan for it, then when we were like, okay, we can make this work. We started to do more things.

SARAH: Once we kind of turned that corner I feel like we've traveled and we've done a lot of things and we don't let homocystinuria hold us back or our kids back in what they can do.

[MUSIC - "Endless Story" by Nick Petrov]

COLBIE: My name is Colbie. I'm 11 years old. I love taking care of my pets. Like, I can talk to them and they won't give me feedback and I want to be a vet when I'm older. And I have HCU.

CAYLE: My name is Cayle. I'm nine years old, and I play football, basketball, I ski, I also play baseball. And I have HCU.

COLE: Cayle, what is it like to live as a nine year old boy with HCU?

CAYLE: Hard.

COLE: What's hard about it?

CAYLE: Um, you have to keep track of your protein and the blood draws every other month. That's about it.

COLE: Can you think of anything good?

CAYLE: It's kind of nice just eating super healthy most of the time, instead of just eating junk food.

COLE: And Colbie, what is it like to live with HCU?

COLBIE: I think it's fun. We get to travel places because of HCU, because we're doing a study. It's also tough with the food when everybody else is having different food that you can't have. But for school, my mom packs us little treat bags and we keep them in a classroom and I can go get them when I want for a birthday. Yeah, it's kind of different from what they're having, but I'm, it's still having a treat, so it's not that different.

CAYLE: That's about the only thing that's different, because we still do a lot of the same stuff.

SARAH: Do you guys feel like you're normal kids?

COLBIE: Yes and no, because nobody's normal, first of all. And yes, because it's just like nobody can tell that you're different until you like, you actually tell them, they would just think I'm an ordinary kid.

COLE: And what about you, Cayle?

CAYLE: Like Colbie said, nobody's normal. Everybody has their own normal.

COLBIE: How was it like being parents for HCU kids?

COLE: I think the hardest part for us was not so much with you, Cayle, because you hadn't started on normal foods yet. But Colbie, you loved yogurt, you loved mac and cheese, you loved hot dogs. That's what you ate all the time. And almost overnight, we had to change your diet and trying to get you as a three year old to say, no, you can't have that. You can't have yogurt. You need to eat this or you need to drink that. So it was hard.

COLBIE: Was there a turning point for you guys?

SARAH: I would say like six months to a year after you guys were diagnosed, we started just slowly feeling more and more comfortable in, in our role as parents of children with HCU. And were getting more comfortable with making your foods and, um, medications. 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 we approach our first birthday party and different things like that. So I think also, just like with anything, after you get your first, your first holiday under your belt, your first birthday party, and 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 going to do it.

CAYLE: What advice would you give to parents that just got diagnosed?

COLE: It's okay to grieve. It's the stages that you're going to get through, it's a lifestyle change and it's going to be a grind for a while, and then before you know it, it is your new norm.

SARAH: I would also say that, you know, one of the hardest parts is you never stop advocating for your kids and you do have to really stay on top of all of those things to make sure that you get good care and that the school is doing the right thing for you and that the journey's not an easy one, but it's definitely doable. And you can lead a normal life.

CAYLE: I'm glad to have you all as family instead of someone else's family.

[MUSIC - “Everything's OK” by Porter Productions]

COLE: I love you both very, very much.

SARAH: I love you guys.

COLBIE: Me too.

CAYLE: Me three.

KATE: Thank you so much to Cole, Sarah, Colbie, and Cayle for sharing their HCU story with us. Nutricia is here to support families like yours and the Sullivans. We’re proud to provide a wide array of metabolic formulas for homocystinuria 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. Come 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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