Natalie Ladly – S2E8 Extra Transcript

My name is Natalie Ladly, and I'm the president and fundraising chair for CDKL5 Canada. We live in Heathcoate, which is just outside the Collingwood area in Ontario, and we have three children. Our youngest daughter, Brynn, has CDLK deficiency

Disorder, also known as CDD. She's seven. And then we have our oldest daughter is Reece. She's 12 and Cullen is ten. CDKL5 five is a mutation in the CDKL5 gene, which essentially stops the production of the protein that's essential for normal brain development and the development of neurons.

And so the impact of that is: children who have intractable epilepsy, and neurodevelopmental delays so it can impact cognition, motor skills, vision and speech. Brynn was born full term. She was a healthy baby. I had a healthy pregnancy and around six weeks of age, she started having seizures. She was put on some anti-epileptic meds and by the time she was four months old she was on three different anti-epileptics with no relief from seizures. So, we went through with pet CT scans, and MRIs and there was no abnormalities, at which point the neurologist suggested that we do genetic testing. It was right around nine months old when we got the diagnosis, which was unfortunate timing for us because she happened to be having a little honeymoon period. And so she'd been seizure free for three weeks. And we had convinced ourselves that she just had this random infant epilepsy and she'd outgrown it, and then, of course, we got the devastating news that she had CDD.

Getting the diagnosis was really tough. I happened to be alone at the hospital when we got the news. So all of a sudden you're receiving this information and you're looking at your baby, who at the time I thought, “Oh, you know, she's doing great.” You know, she wasn't. hitting her milestones, but, you know, it becomes your new normal and you just sort of think, well, she's having a really good day and, you know, she's doing good based on how she's been doing in general. And all of a sudden. You find out that, you know, your child is likely never going to walk and never going to talk and will never live independently. And, you know, it was crushing.

Like it was so hard for me to wrap my head around how this doctor could know with certainty all of these things about my child and you know, and then all of a sudden you hit like this wall of grief because all of these hopes and expectations you have that you don't even realize you have, you know, you realize that none of those are going to happen.

And so it was it was pretty rough the first couple of days but then I came home and my husband like, you know talked about it and you know, after going through like the vast amount of symptoms that are associated with CDKL5, we realized that the only say to really deal with that was just to take one day at a time and just, you know, with the same attitude that I, you know, I walked into the hospital thinking she's doing great. Meanwhile, you know, she was a nine-month-old who couldn't sit up and was having. seizures previously. And, you know, like we knew she was behind, but just take it one day at a time. And that's how we can move forward. And so that's kind of what we've been doing. We just address each symptom as it comes up and try not to get overwhelmed with the caregiving aspect of it.

Brynn is a spunky seven year old. If you were to see her in an arena or at a soccer game, which is where we spend most of our time you would probably think that she just, you know, is oblivious to the world around her. She's seven years old. She's in a wheelchair.

She's nonverbal. She's has a feeding tube. On initial glance she doesn't look like she' very engaged. But everyone who knows her well, I always know when a teacher knows her well, because they say, oh, she's such a drama queen. And I'm like, okay, that's Brynn. So somehow, without speaking a word and she doesn't gesture. She really has very limited ways of communicating but she manages to capture everybody's heart. And Eric and I always say that's her superpower, is that she charms. Everybody that she meets.

She loves her friends, she's in school, she's included in her Grade 2 classroom and participates in as many different activities as she can, and she goes horseback riding as part of her hippotherapy. Hippotherapy as a type of physiotherapy. So Brynn sits on the horse, she rides frontward, she rides backwards, she lies down on her stomach and on her back. And it not only provides her impact that she doesn't get because she doesn't walk, but it also allows her hips to move in the motion with the stride of the horse when it walks, moves her hips in the way that mimics walking. And so it's really good for her joints. And it also gives her the feeling of the stepping over motion that she doesn't get from walking.

She loves being in her swing and in the swimming pool. And, you know, there's an abundance of activities that she thoroughly enjoys participating in. She just misses a lot of the sort of typical activities that a seven-year-old would be doing.

Brynn communicates a lot with her eyes she gives a stink eye If she's not happy. With you far more often than she gets a smile, if I'm being honest. And she often if she's excited, she'll kick her legs out or she'll sometimes squeal. She will give us smiles occasionally, but more often than not, she's just quite content. And she's a very mild mannered little girl.

She doesn't complain. She never cries. And we joke that she's the easiest to get along with because she doesn't talk back. And I say to the other kids, yeah Brynn is our are favourite because she never gives us any grief about anything.

So Reese and Cullen both have really close relationships with brand, but they interact with their really differently. So, you know, they both are. Affectionate. And care about her and care for her. But Reese, who's 12, is much more practical in her approach. She likes to help. She sets up. Feeds for brand. She'll change her. She makes. Her medicine. She's very hands on in that respect. And then Cullen is like a sensitive soul who just will like, climb on the couch with her and smuggler and reader a million Peppa Pig Books. And so it's interesting, they have totally different relationships, but I think she values both. You know, she gets. To engage with both of them and just in different ways.

Brynn is currently on three anti-epileptic medications and she also takes CBD oil, cannabinoid oil, which helps control her spasms. She's having approximately two seizures a day that vary in length. Right now between four. And 5 minutes, which is good compared to this time last year when she was having her average seizure length. Last year was between ten. And 12 minutes, and that for us, we find it's the most difficult part of CDKL5 five. There's a huge array of symptoms and they all require different levels of care. But the seizures are the part that impacts our life the most. It sort of stops whatever activities you're doing. If we're having dinner or when, I have to get up and leave the table. If we're in public, it doesn't bother us, but it bothers everybody who's around us. It makes people uncomfortable, which, you know is never a great feeling for for us or for Brynn. It steals all of Brenda's joy. When she's having a good day with no seizures, she's bright eyed and she's engaging. And, you know, you can really feel like you're communicating with her and she's reciprocating that. But the seizures just steal the show like she sleeps and she's

exhausted and she has a hard time clearing her airways. And, you know, it just it's just really burdensome.

After Brynn was diagnosed, we made it till she was around two and then we quickly realized that working a normal 9 to 5 job wasn't going to make sense for us anymore. Between her doctor's appointments in Toronto, the physiotherapist, the occupational therapist, and you know, just even seeing the regular Pediatrician it wasn't sustainable for me to continue working full time. So a business opportunity came up that was totally out of left field and my husband pushed me and encouraged me to take it on. And so now I run my own business and it's taken a huge weight off our shoulders because it allows me to have some flexibility and I can make time for Brynn without feeling like I'm doing it at the expense of an employer.

I ended up joining the board of CDKL5 Canada when Brynn was one and a half. And it was for me at the time in a situation where I felt like I had no control and I had really no ability to do anything for Brynn to help her other than caring for her. Being on the board made me feel like, okay, this is something that I can accomplish. I can work towards, schools, we can help create awareness, we can fundraise and invest That money into research and maybe one day they'll be treatments or a cure. And even if it's not for Brynn, then if other families don't have to suffer through the experiences that we are suffering through, then you know then it's all worth it.

So having Dr. Eubanks is a gift for patients with CDKL5 because it's so hard to find researchers who are interested in studying rare diseases, first of all because they're not well known. So a lot of people aren't even aware that they exist. But also because there's not necessarily the funding and there's not people willing to invest. Dr. Eubanks found us, I think, through connection, through Rett Syndrome. And he's just been incredible. The Lilly Foundation is a non-profit organization that runs out of London and they host a scientific forum. They've been hosting these forums for about ten years now and Dr. Eubanks is one of 12. They call him one of the pioneers who has been attending the city CDLK5 Forum right from the beginning and has never missed a year. His research is so progressive. And he's making such a big impact. And, you know, just when you speak to him that he cares so deeply about the work he's doing. It's not just some random rare disease to him, he cares and he asks about Brynn and he, you know, gives feedback about symptoms or strange things that happen that I'm just kind of shaking my head and trying to wrap my head around. And. You know, he wants to be a part of it and his passion and his dedication, It makes a huge difference. On days where it feels hopeless for us, you know, we're up all through the night, and sleep deprived and oftentimes just kind of, you know, you have days where you're like, how am I going to keep doing this? Like, I'm exhausted and you know, it feels like everything just piles up. And then I'm like, okay. But Dr. Eubanks is in the lab and he is plugging away and he's making a difference for us. And and that's all you need. Sometimes you just need to know that you've got somebody in your corner who is, you know, rooting for you and trying to make the world a better place, really.

Caring for a child with special needs-it's hard. It's a heavy load. And I would be lying if I said, you know, it's all sunshine and roses. We were a silver lining family. And we do try to, you know, find the humour in. Situations and try to make the best of it, because. At the end of the day, we don't really have a choice. You know if we can have a healthy child that would always be the option that you would choose. So, you know, it's, it's a struggle, but the silver linings are that, you know, it does make you incredibly grateful for when your child is healthy and thriving. She genuinely, somehow, without speaking a single word,

brings out the best in everybody around her, including us, our friends and family, our community. Everybody reaches out and digs deep in their pockets and in their hearts and you know, she's included at school in ways that I never could have ever expected or imagined. And, you know, it just kind of makes your heart swell. And I'm like, oh, my gosh, I didn't, you know, I would never have known that this world existed if it wasn't for Brynn. So, yeah, I mean, it's certainly not. What I expected when. You know, when we tried for our third child, if I, if I had had known I'd had a window. I mean, no, my gosh, I can't imagine life without Brynn. But it's, you know, it's amazing you have healthy kids and you just you never dawn, just think that this could be somebody's reality.

For rare diseases, it needs to be a collaboration between the scientific community and the patient advocacy groups, because really, at the end of the day, the knowledge is they're out there, but they're dealing with so many different disorders and they're sort of inundated with everything. And there's not the volume in the rare disease community for practitioners to really be able to treat patients like specific to their disease. And I think if you can find a connection like what we have with Dr. Eubanks, where we can support him monetarily in a small way and he can be engaged with us and understand, you know, get a real-life picture of what he's looking at because, you know, he's joked with me and said, you know. We look at it on such a microscope level. And then you meet children and you're like. This is. What we're here for. You know, like, this is this is what we're working towards. And I think having that connection and having the support and working together in the rare disease community, more so than in, you know, regular areas of health. I think that's the most impactful thing.