

# Quest

## PODCAST



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### **Episode 1: Receiving a Diagnosis** **Friday, August 6, 2021**

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Mindy Henderson: Welcome to the Quest podcast, proudly presented by the Muscular Dystrophy Association, as part of the Quest family of content. I'm your host, Mindy Henderson. Together, we are here to bring thoughtful conversation to the neuromuscular disease community and beyond about issues effecting those with neuromuscular disease and other disabilities, and those who love them. We are here for you to educate and inform, to demystify, to inspire and to entertain.

We're particularly excited to be launching this podcast during the summer months, which celebrate both Disability Pride month in July, and SMA Awareness month in August. We are here shining a light on all that makes you, you, whether you are one of us, love someone who is, or are on another journey all together, thanks for joining. Now, let's get started.

For anyone with a neuromuscular disease. The journey begins with diagnosis, a name for the symptoms that are manifesting. Just getting to the diagnosis can be a convoluted odyssey and take months, if not years. And even with a confirmed diagnosis, the odyssey is just beginning. Today, we'll dive into that conversation from multiple perspectives. An individual diagnosed in infancy, an individual diagnosed in early adulthood, and a parent who went on the journey as an advocate for their child. We'll talk about each of their stories and the path they took to get a diagnosis, how it impacted their lives, how they've learned to live with it, what's been hard, what's been easier, and how they've coped and made a life that works for them.

So with me today, I have Paloma Juarez, she and her husband Brian Way are parents to Vaun, who's five years old, and twins, Koen and Zavier who are

almost 10 years old. Vaun and Koen both have infantile-onset Pompe's disease, and the family lives in Prairie Village, Kansas.

I also have Chris Anselmo, who works at the Muscular Dystrophy Association, in a partnership management role. Chris lives with limb-girdle muscular dystrophy type 2B. He was diagnosed with the disease when he was 18 years old, the result of a routine blood test after a car accident, and before he had any physical symptoms. Today, Chris is 34 and has lived with increasing muscle weakness for the last 13 years. And then there's me, I was diagnosed with spinal muscular atrophy type 2 at 15 months, and I plan to speak about growing up with a disability. So Chris and Paloma. Welcome. Thank you so much for being here.

Paloma Juarez: Hi.

Chris Anselmo: Yeah. Thanks for having us.

Mindy Henderson: Absolutely. So I'm just going to jump right in you guys, Paloma, let's start with you. Would you tell me about the journey to get a diagnosis for your kids? And just how you knew something was wrong, and where things went from there?

Paloma Juarez: Sure. So just to correct you, Koen and Zavier are actually only 10 months old, not 10 years old.

Mindy Henderson: Oh, I'm so sorry.

Paloma Juarez: That's okay. So actually our experience is kind of unique, especially in the Pompe's world. So Vaun was born full-term at a hospital here in Kansas City, Missouri. We had done a birthing class and heard something about newborn screening, and they kind of mentioned, "Hey, you'll probably never hear about it, just know that they'll prick your newborns heel, run a test, generally nothing shows up." Little did we know that a week after he was born, somebody did call us about it. And his newborn screening showed a low enzyme level, which indicated possible Pompe's disease.

We found that out on a Friday. On that following Monday after researching and stewing and crying all weekend, we met with a genetic therapist and counselor. An echo was done that day, some blood work was done that day, but once they saw hypertrophic cardiomyopathy, they felt confident with the low enzyme level, from the newborn screening, coupled with the heart problems to begin the diagnosis for infantile-onset Pompe's disease.

Mindy Henderson: Wow. And what did they tell you about that? What did they say that the prog was and what you should expect?

Paloma Juarez: So, again, five years ago, newborn screening was just very brand new for Pompe's disease, only five states screened for it. So most children Vaun's age are diagnosed clinically. And so the outcomes for a clinical patient and what we were told were, our son wouldn't be able to walk, he would likely be on a ventilator, wouldn't be able to eat or swallow on his own, best case scenario that

he would live, perhaps, till two or three years old, but that we needed to prepare ourselves for a short life full of complications.

Mindy Henderson: Wow.

Paloma Juarez: Because Vaun was diagnosed via newborn screening, we began what's called enzyme replacement therapy for him, by the time he was 24 days old. And his story is a very unique and different one, and one that we're hoping as more states adopt newborn screening policies for Pompe's disease, that can become a popular story for others. Vaun walks today. Vaun talks. He's met every single milestone. He eats independently. He starts kindergarten in the fall. His fifth birthday was one that was super emotional for us, because we really just never thought that we'd even get to that point. So to be here and be having a child that's seizing and having every opportunity as his peers, has been really quite spectacular and miraculous to us.

Mindy Henderson: Oh my gosh, was the story similar for your other sort of walkthrough this journey.

Paloma Juarez: So, because we knew ahead of time that Vaun had this diagnosis and that Brian and I are both carriers for it, when we found out that we were expecting, we set an amnio for 20 weeks, and had the genetic testing done through there. So Koen and Zavier, we knew their diagnosis' before they were born. So Koen has the exact same mutation that Vaun has, and Zavier hit a different way. He is actually not even a carrier. He's unaffected by the gene completely.

Mindy Henderson: Wow.

Paloma Juarez: Yeah. Two very different perspectives.

Mindy Henderson: Interesting. So, Chris, we know that you were diagnosed when you were 18, you had a car accident that sort of led to your diagnosis. What did the doctors tell you at the time about your illness and what you should expect?

Chris Anselmo: I mean, the whole thing really just seemed like a big fluke, because I didn't have any symptoms at the time, yet, I was told that I had some sort of muscle breakdown going on inside of me. And it seemed very surprising, because it just didn't match up with the experience that I was living. I was growing up doing a lot of sports, I played basketball, was very active. So to hear that I had a muscle disease was really kind of a shock. I mean, if you think about the previous year of my diagnostic journey, so started with a car accident, when I was 17 years old, I didn't formally get diagnosed, however, until I was 18.

I saw all sorts of different specialists. They were trying to figure out why my creatine kinase levels were so high. So for a typical person, it should only be a few hundred, mine was like 35,000 or something.

Mindy Henderson: Oh wow.

Chris Anselmo: So initially, they thought that I had suffered an internal injury as a result of the car accident. When it was clear, it was not the case, that kind of led me on a whole journey where they're just trying to figure out what it was. So I kind of had this uncertain cloud over my head for about a year. And after seeing a few different specialists, they decided to do a muscle biopsy, because they figured it might be muscle related. And they determined that I was missing this protein called dysferlin, and the neurologist at the time knew a little bit about it, but she really wasn't too sure what the overall prognosis was. Because this was back in 2004, there just wasn't that much information about it. It's a very rare thing to be missing this dysferlin protein.

And just based off of her kind of analysis and literature, she thought that, perhaps, this might be something that would manifest later in the life, my '30s and '40s. Now, I was just about ready to go to college, so she said, "This shouldn't affect you going to college. You should be fine. You don't even really have to think about it, just don't become like a body builder or a marathon runner." Which is fine, because I hated exercising anyways, but it really wasn't anything that I needed to worry too much about. And I went to college, I was fine.

And then shortly after graduating from college in 2008, I just started noticing just random symptoms of just muscle weakness or just not being able to do something that I was once able to do quite as well, like going for a run, carrying heavy objects up the stairs. I was winded after climbing several flights of stairs, that sort of thing. And it took a while to figure out this might be the disease that I was told I had five years ago.

So I went to an adult neurologist in Boston, where I was living at the time, who confirmed that it was what he called dysferlinopathy. It's got many different names. It's kind of known as limb-girdle muscular dystrophy today. But that I had basically a muscle disease and that it actually was taking place now. So it wasn't something that was going to happen later in life, it's actually something that was happening to me now. That the muscle weakness I was experiencing was part of the disease and that it was just likely continue.

Mindy Henderson: Okay. So let me back up just a little bit, when you were in college, what was that like, knowing that there was something there, it hadn't manifested yet? Were you able to put it out of your mind and live your life throughout those college years?

Chris Anselmo: Yeah. I barely even thought about it. I knew I had something, but it wasn't front and center in my mind. It was just something that was kind of below the surface. I even forgot what it was called. I didn't even think about it for like four years. I just was consumed by other things. But I didn't really notice any limitations. I was able to kind of do everything that anybody else my age could do. I went to the gym. I continued to play basketball. Looking back on it now, there probably were some warning signs in terms of just recovery from exercise, that sort of thing. But for the most part, it was not part of my consciousness at that point.

Mindy Henderson: Gotcha.

Chris Anselmo: I was just consumed with just being a normal college kid.

Mindy Henderson: Well, yeah, and so I guess I'll just kind of interject my story here. I was diagnosed with spinal muscular atrophy type 2, there were three different types, when I was only about 15 months old. And I guess my story is different from both of yours, because I don't really remember the initial diagnosis. Paloma much like you and your journey, that was up to my parents to navigate. And basically what happened with me was, I started to walk and stand when babies normally do that sort of thing, and very quickly thereafter kind of stopped. It was almost like I lost interest, to hear my parents talk about it. Just kind of couldn't be bothered to get up and walk around anymore.

And my parents took me to the doctor, the pediatrician, who actually said that I was fine. He said that it was probably a phase that I was going through and just to kind of let it ride out. And so, my parents, though, they knew that there was something else, and I like to call it my medical scavenger hunt that we kind of embarked on and ended up at the Mayo Clinic, diagnosed by the head of neurology with this condition. And this was 40 something years ago. And so there was a lot less known about this condition than there is now.

My parents were actually told that I would, in fact, continue to lose my ability to stand and walk, which I have, but they were also told that I would lose all of my cognitive function, and that I probably wouldn't live to be three. And so they talked to all the experts, there was no treatment, no cures, nothing really at the time that could be done about it, but they did start physical therapy with me.

And it was subtle, but bit by bit, I did start to make some small improvements and then I had a third birthday, and I've had a lot of birthdays since then, so that's really kind of how my story came to be. I ended up in a wheelchair pretty quickly, I think I rode around in strollers and things as a toddler, but then moved into a wheelchair when the time was right for that. And didn't really have much time where I stood or walked. So that's kind of how my story went.

So Paloma, let me come back to you. How did the news of this diagnosis land with you? What were your initial concern and fears? And how did you work through the shock and the emotion of it?

Paloma Juarez: So I feel like at, first, your fight kicks in. There wasn't, in our mind, for at least those first like six months, where you had this moment of maybe grieving over the diagnosis. I feel like those feelings came much later. For us, it was, what is our plan? What are the next steps? How do we create and give an incredible life to this precious little person that we have in our arms? There wasn't this moment of kind of where you wrapped yourself in the emotion of all of it. We just wanted to do and research and figure out what the next steps were.

So Brian likes to call it our emergency room time, where everything was just really fast and working towards a solution. I'd say probably like as his first birthday approached, we kind of allowed ourselves to really talk about those feelings, and what our concerns for the future were/ with the big question being, does this mean that he is our only child?

And so for a long time, we really just worked with the idea that he was our special gift, trying to be the best partner for one another that we could, but really our focus has always been helping Vaun, and then helping within our community. I get a lot of therapy out of that.

I think the other hard thing was though, my friends were building families at the same time, and my husband and I run a girl's volleyball club, and so we were working with young athletes all the time. And there were moments where you felt almost this feeling of jealousy at times, where you're like, "Well, why do you get to have these just regular experiences without the stress or the unknown?"

And I think that between us really working harder with our athletes, it gave us the opportunity to get to know them more, kind of embrace this idea of let's take that idea of jealousy of being "normal," and let's really work to just know people more. The more that engaged with our athletes, the more you realize every child, every kid, every family had something that they were working through or some struggle that they were overcoming. And I think instead of manifesting those feelings of jealousy, it really made us feel like, "Okay, this is just part of parenthood. Every parent's going to go through some kind of journey, maybe to some more significant than others," but that feeling of community with other parents really helped us through those feelings.

Mindy Henderson: That is such great perspective. I've never quite heard it that way. And so often we get so kind of bogged down in our own adversity, and what's going on in our lives, that, I personally agree with you, I think it can be so helpful to look around at what's going on with other families. And just to know that their struggle may not be your struggle, but guaranteed, they have a struggle. I love that.

Paloma Juarez: Absolutely. I really feel like the more that we could take this idea of, yes, we understand that his journey is different. Yes, we know that he's got challenges that others will never face. I think, for us, it was about finding that balance of having real positivity. How do we look at the world in a way that's going to help him continue to grow and move forward, and not have these feelings of despair? Like why me? And so I really felt like, for him to be able to do that, we had to really work on that perspective for ourselves.

And, again, the more that we can find and talk to, and like you said, embrace this idea that everybody has some sort of adversity that their work me through. What can I learn from the way that they handle that, it'll make us better parents and leaders for Vaun, and then Vaun to be able to navigate those things in the future on his own?

Mindy Henderson: Yeah. So did that intentionality to your mindset, did that come naturally to you? Or was that a decision that you actively made to look at it from a more kind of thoughtful, positive place?

Paloma Juarez: I think it comes from twofold. So my family is this crazy, large, loud Mexican, Catholic family, and so we were always raised that you just power through and you're strong and let your faith carry you through. And my husband, Brian, is very much, he's very logical and he loves to learn. And so we kind of combined

those things, where I was the emotional one, just ready to carry us up and fight. And he was the one that was very tactical. And we did a lot of research on different sociology ideas, psychology ideas, what it means to just be a learner and embrace some of this. And so I think our ying and yang attitudes actually just helped us naturally create a plan that was going to be one that could sustain us and carry us through as a couple, as individuals, and then as parents for Vaun, and now Koen and Zavier.

Mindy Henderson: That's so great. So Chris, once your symptoms did start to manifest after graduating college, what was that like emotionally for you? And how did you work through it?

Chris Anselmo: It's actually interesting hearing Paloma talk about all the different things that helped them with their son, because that's a lot of learnings that I could have benefited from during that early period. I eventually got to that point, but in the beginning, I did basically the opposite of that. I had a really hard time reconciling what the neurologist that I saw after college had told me that the disease is happening now, that I was going to be getting weaker. I'd eventually end up in a wheelchair in 10 years.

What he said versus what I was holding onto, what the neurologist from several years before had said, that this isn't something that I had to deal with right now, it'd up later in life, by then there certainly would probably be a treatment. Don't worry about it. So it took a really long time. And I really struggled with accepting that the disease was happening to me now at age 22, 23. And this was something that I was going to have to deal with, and my life was about to get infinitely more difficult.

I didn't handle that well at all, actually, in the first two or three years. All the things they tell you that would help you during that time, I basically didn't do. So rather than reach out to people for help and support, I became kind of a hermit. I didn't want to open up to anybody. I wasn't a very pleasant person at that time. So it was just so consumed by just frustration of what was happening to me. I stopped being able to run. I started fall about a year or so after that. I was just very jealous that my roommates, my friends, they're all kind of advancing in their lives, they're buying houses, they're getting married, they're getting a dog-

Mindy Henderson: Sure.

Chris Anselmo: ... and I was just worried about can I get to work without falling? So it was very tough. I kind of just did not want to deal with what is happening. I didn't seek out any really other patients that had the conditions. I didn't want to know what my future was going to hold. I kind of held to this false notion that there was something that I could do to keep the weakness from happening, to kind of stop it in its place. So I altered my diet. I tried to exercise very, very carefully, so I didn't overexert myself, but it didn't work. I tried physical therapy, tried to be as limber as possible, tried to stretch, didn't work.

And all of that together, it just became very difficult to deal with. Because, again, I was a young adult. I had my whole life ahead of me, and then to have all this

happen, was really just hard to accept. And it took several years to get to that point, but in the beginning, I did not handle it well at all.

Mindy Henderson: Yeah. And like you said, understandably so, you were, as you said, a young adult and none of your other friends were having to deal with this kind of thing. And you didn't, maybe have the life experience yet that, say, Paloma had, and her husband, to look at the situation a little bit differently.

Chris Anselmo: Yeah. I didn't have the right perspective. I mean, everybody deals with a life changing diagnosis in their own way. There's no necessarily wrong way or right way to handle it. It's just a matter of how you get the news, the support system around you, your mindset. And I kind of think of it, now, in terms of a continuum. There's people that get a diagnosis like that, that become completely jaded. And then there's people on the other end of the spectrum that just see it as one minor speed bump in the road of life. That have the right mindset to be able to tackle it, and that are just abundantly optimistic. And if you think of it in terms of that spectrum, I was much closer to just the angry, jaded side early on. And it took me several years to move the needle a little bit towards the more optimistic side.

Mindy Henderson: So what was the turning point on that? Or was there a single moment or time period that comes to mind where things changed for you? Because, clearly, that's not who you are today.

Chris Anselmo: There wasn't necessarily an epiphany moment where it just made perfect sense that I needed to change how I was living my life. It was more just a sense where I was just kind of sick and tired of just not achieving my goals, of just being kind of angry all the time, being frustrated, of being very reactive to what was happening to me. And I just thought about my goals that I had had for myself. I wanted to be a writer. I wanted to go back to school to get a master's degree. So I was maybe 26 or so at the time, and I just took out a piece of paper and I just kind of wrote down my goals. And I just got backtrack, where like, given the circumstances, given that I can't change these things, and that was the hardest part, I just for so long was struggling to accept that I couldn't change certain circumstances, given those circumstances, how can I still achieve my goals?

And I was able to, thinking about what Paloma had said about her husband being very logical and process-oriented, I just kind of mapped out, if I wanted to go to grad school, for example, how would I make that work? What resources would I need? What help would I need? How would I make it work financially? How would I juggle that and having a job, et cetera? And I was able to see like, hey, okay, maybe I can't do it the way that I expected to do it, or the way that I thought maybe 10 years ago that I would do it, but there's a way to do it. And once I kind of saw that you could still achieve your goals in life, you just might have to do it a different way, that really helped to open doors that had previous closed in my mind.

Because for so long, I just didn't think certain things were possible or they'd just be too difficult. And a lot of that was kind of eating away at me, and all of a sudden I was able to see, gradually, that that's not the case. There are still all



these opportunities. These goals would have to be achieved differently from maybe what I had expected, but they were still possible. And that really just helped to kind of reorient me into the right direction towards being able to live a productive life, to kind of abandon some of the anger and jealousy that I had experienced previously, and to just move forward towards a more productive and optimistic future.

Mindy Henderson: Oh, that's so good. And I love that that was the approach that you took, just looking at what your goals were, what you wanted your life to be. I think that that can be a really powerful exercise for anybody, regardless of limitations or where they are in their life. Because like we were saying earlier, everybody's got challenges and you may not be able to get to a destination the way that you thought you were going to. But I think just knowing what you want, who you are, can go a long way toward the first step to figuring it out.

And for me, I grew up in a family, clearly, my parents were fighters, and they had a lot of influence over me and they really raised me to live without excuses. I had an able... or still do have an able bodied sister, and they raised me with the same expectations that they had for her. I was expected to do well in school, to have friends, to have interests, to have dreams for myself and a vision for my life.

And so I think that, Chris, a lot of the work that you maybe had to do for yourself, for me, it came down to you having parents with the foresight to be able to kind of instill that in me, and enable me with those sort of life skills to architect a life that worked for me as I went.

So Paloma, I'm going to come back to you for just a second. We're talking about two very different kinds of scenario, obviously, Chris and I are the actual people that the diagnosis was given to, and then you are here representative of a parent whose child was diagnosed. And I'm wonder, if, I suspect that there might be an additional layer of helplessness or maybe guilt that comes along with a diagnosis like this for your child, was that true for you?

Paloma Juarez: For sure. I mean, I think I have all those feelings that Chris was talking about, but I think the difference was that, as the person that has to lead this child and teach them how to be the best version of themselves, it's just not feelings that you can linger on too long. So, I mean, I have had feelings of this idea of guilt, but to be quite honest, I have feelings of guilt like that with Zavier, who has no diagnosis. I think sometimes that's just the experience of being a parent, being someone that cares so deeply for someone, and wanting to be able to provide and give the best life possible.

As far as helplessness, yeah there are certainly moments where we felt helpless. So we, right or wrong, work to control the things that we're allowed to control, getting him into therapy as often as possible, making sure that doctors communicate with us, not because we're his parents, but because they should, making ensure that we put him in any and every time experience it's going to make him feel like a confident person and not somebody that feels disabled. And so I think that maybe the helpless side of it is, maybe, where we can learn to do

better, but for our us, it's just been about seizing control where we can have some control, because there are so many things that are outside of it for us.

Mindy Henderson: Yeah. So, so wise. And Paloma, can you tell me what life has been like now that you've got a few years of this type of journey behind you, and what their life is like today?

Paloma Juarez: Yeah. So again, like Vaun, is incredibly, and Koen's on his way to being, I mean, don't get me wrong Koen's very, very lucky, but Vaun kind of paved that way for him, you know?

Mindy Henderson: Yeah.

Paloma Juarez: So for us, it just always been staying one step ahead of things. So his center of excellence, Dr. Kishnani out of Duke, they're very much, "We don't wait for symptoms or things to start showing a trend of not the direction we want to go. We always stay super proactive on things." So Vaun started physical therapy and occupational therapy when he was three months old. We slowly ended up adding speech therapy and feeding therapy, as we noticed some weakness in his oral muscles. But for the most part, really, it's just kind of managing the schedule. So both boys have weekly infusions now, and that takes a whole day.

Mindy Henderson: Oh, wow.

Paloma Juarez: We're fortunate enough to be able to do that inside the home. So we have a wonderful care team that comes to the house weekly, has these wonderful relationships with both boys, not just as their nurses, but really because they care about them. They're people in their lives. And I think that's been the most awesome experiences, while we have these challenges, our sons are able to meet some really spectacular, caring, kind people that are not just advocates, but really teach our boys how to be a good member of this wonderful world.

Mindy Henderson: Wow.

Paloma Juarez: And I think those are all relationships that you want for them. So that's been incredibly fortunate to us, but really quite awesome, trying to find the things that we can take away that may be experience really, not so dreary, but hey, this is life, and yet we're getting good experiences out of it.

It's weird to be home sometimes for medical deliveries once a week. And some of our neighbors see us taking out bigger things of trash than them, and that's kind of how conversations can be started up at times. But I think, for the most part, it's just feeling like you can get into a routine with things helps. So on these days, you're in therapy, on these days, we have infusion days, and kind of making infusion days feel more special for them, right? So Vaun gets-

Mindy Henderson: Yeah.

Paloma Juarez: ... unlimited access to his technology on infusion days. And try and make like a fun breakfast or pawpaw, my father, will bring the boys over lunch, so trying to

make it a very special day for them, so that it doesn't feel so high anxiety or cumbersome for them. Especially as Vaun gets older and isn't just going with the flow, but has questions and wanting to understand these ideas a little more, like why do we have to do this today?

So that, hopefully, as he grows older, the transition still is one that doesn't feel like, oh my God, I don't want to do this anymore. Because this will be a lifelong treatment, infusions weekly for the foreseeable future, until something cooler and more medically advanced comes along. So I think a routine is the best way for us to cope through all of it.

Mindy Henderson: That makes a lot of sense, gives kind of a sense of predictability and control to your situation. And I love, also, that you're making those infusion days special, because as inconvenient and maybe unpleasant as the infusions may be, as the treatments may be, they're life saving, and they're such miracles. Myself as well, with spinal muscular atrophy, I've been getting the Spinraza treatments, which is a spinal injection into the spinal fluid, once every three months, which is inconvenient and painful.

And I've got a tricky spine that requires some really sophisticated equipment and doctors to navigate. But I also recognize what a gift it was. I lived 44 years of my life with there being absolutely nothing that anyone could do about this. And so seeing what a special, celebratory thing this is in our lives, I love that you do that for your kids.

Paloma Juarez: Yeah. I think it was one of those things where we, at first, thought, because Vaun's been doing this since he was 24 days old, we just thought that maybe he wouldn't know any different, because this is just what he's done all his life. But somewhere around the age of three, we started noticing some anxiety on those days, getting really annoyed with kind of being confined for the day or whatever. So we really had to make sure that we were consistent with, "You have to do this, because you have to have it," right?

Mindy Henderson: Right.

Paloma Juarez: In a way that he can kind of understand that without it being demanded, wanting him to feel a part of it. And we take twos for him on that too. If there's ever a day where maybe he flinches or wants to go for a walk outside, we make sure that he can have some flexibility in communicating those things to us and his nurse. And then being accommodating where we can. We, obviously, can't go to like the park or anything, but if we want to go for a stroll around the block or whatever, as long as his nurse comes with us, we can do those things.

And so we're learning a lot from Vaun in hopes that, because, wow, Koen is just so different. He's our unruly one already. Keeping an infusion pump away from two 10 months old twins is difficult.

Mindy Henderson: Oh boy.

Paloma Juarez: But we're hoping, again, to learn from kinds of behaviors that we've gone through with Vaun, and things that worked and things that didn't, that hopefully our a little wild child can kind of stay compliant, and be helpful here at home, in the future too.

Mindy Henderson: Definitely. And I want to go back, too, to something that you said a few minutes ago, just talking about your nursing staff, and the caretakers and other people. And for me per personally, I always try to see the good in the bad and to find the lessons that can be learned and to find the purpose, and all of those things. And I think that one of the best things that comes from a disability like this, or neuromuscular conditions, is you get to see the compassion in people and you get to see the side of people that's so kind and so caring. Not a 100% of the time, but I think that we maybe get to see a lot more than other people, who are not going through this particular challenge, which I consider to be a blessing in my life.

Paloma Juarez: Absolutely. Don't get me wrong. Everybody's going to have different experiences, and finding the right care team for you, it can take a lot of time. But, again, I try and think about the therapist or the nurses that have made such an impact already on our five-year-old. Again, it warms your heart, because those are the relationships and things that grow a person. And so we're incredibly fortunate for these people.

I think the other awesome thing is we've had players that have either taken a real liking to our family and our kids and stuff. And some of them are going on thinking about doing and majoring in these things in the future and stuff. So I think that there's definitely such a need for that, that we love that our awesome stories and experiences can be inspiring for people looking to have those opportunities, not necessarily as a patient, but as a caregiver.

Mindy Henderson: Yeah. It's so true. And I will also say, I've created a life for myself with quite a bit of independence. I drive a car. I am married. I have a daughter. And I always say that, whoever created me, whatever you believe, had a really good sense of humor. Because I was created with these physical limitations that make me dependent on people so much of the time, and yet I have a fiercely independent spirit. And so it's been kind of an interesting contradiction to grow up with.

And it's forced me to really work outside the box, and think outside the box to learn how to live independently, and how to employ personal care assistants, who just like you're saying, at times have become some of my best friend. And years after they stop working for me, they continue to be in my life in some way or another. So, yeah, there's a lot about this kind of lifestyle, I think, that just lets us see the goodness in people. So Chris, what have you learned up about yourself as you've gone on this, what? 13-year journey that you've had so far?

Chris Anselmo: Yeah. I mean, it's been interesting hearing you talk about the impact of having other people in your life that are kind of going along this journey with you. And I can really relate to that. I think one of my greatest learnings, if not my greatest learning, of all of this was learning how to ask for help. How to let people in on what was going on.

I haven't even talked about my family. I haven't talked about my parents or my friends, but they've been instrumental on this journey, and just helping me anyway that they can. And I think part of what I struggle both in the beginning was just learning how to ask for help. Not realizing everybody around me wanted to help, they just weren't really sure how to do that. They didn't want to be intrusive. They didn't want to just assume that I needed help with certain things.

Once I realized that, once I started to accept that, even asking for help as a form of independence, because you know what your limitations are, you know what you need help with. The sooner you ask for help to do something, the sooner you can go back to just continuing with your life and achieving your goals, moving forward.

Mindy Henderson: Yeah. And it's kind of a way of advocating for yourself, too, right?

Chris Anselmo: Exactly. It's something that everybody with everybody struggles to ask for help. But I just got to a point where just I was having so many different issues, physically, that I just needed to start asking people to carry things and whatnot, pick me up if I fall and that kind of thing. And once I connected with other people that had the conditions, different forms of muscular dystrophy, and even other people that had rare diseases, once I shared with my parents just how much I was struggling, once I shared with my friends that I could no longer hide that I was having physical weakness, they've all been super helpful, super compassionate about making sure that I had everything I needed to kind of deal with this.

And I didn't really realize how much they were affected by it as well, in the fact that, you have a disease, you think that you're just living this by yourself, but it affects your entire circle. It affects your parents. It affects your family. It affects your friends. They don't want to see you get weaker. They don't want to see you struggling to walk or to fall on the ground. But once I realized that and shifted the focus off of myself, stopped worrying about how I felt all the time, and started opening up to people, it really became a reciprocal process where I started to connect with people in a new way.

People shared with me things, I had coworkers that would share with me things, that I don't think that they've shared with many other people, in terms of things have struggled with in their life. I've had friends that have disclosed things to me that I never expected, in terms of what they might have been dealing with. And you kind of form that connection, because they kind of have seen your story, they've seen you live it out. And once you start to take hold of what's going on with you, and you start to figure out a way to live a productive and happy, or not always happy, but productive and more optimistic life, that really affects other people. And they kind of look to you as sort of a role model.

I mean, I still don't think of myself as a role model, but I never really thought that just me living with this disease would affect people in that way. And then all of that helps me to accept what was going on and to accept that this is part of my life, this is who I am, but there's so much more to me than my disease. That

was the greatest learning, and that's just one of those things that I wished that I had learned several years prior. But again, everybody deals with a disease like this in their own way. And there's no set and fast roadmap that by year two, you have to fully accept it, by year three, you have to start achieving goals that you had set for yourself. I just found that opening up to people, learning how to ask for help, you really just form really special bonds with people.

And there will always be people that, they might not be comfortable with it or they might not know what to say or anything like that, but they're few and far between I find. When you're authentic and you're able to kind of show, like, this is my disease, this is what I'm dealing with, this does not define who I am. And I can empathize with you if you're struggling with something. I think once they realize that, that you can be somebody that they can talk to for advice, or even to use you as kind of a roadmap for how to deal with your own situation, that's really kind of an empowering feeling.

Mindy Henderson: Yeah. Oh, you make such good points. I don't even know where to start. I think the first thing that's coming to mind listening to you is that the power, I think, of sharing our stories and our vulnerabilities is huge. I think that is a sign of strength to be able to talk about the hard things in your life, and to be able to ask for help, like you're saying, it's empowering. And with something like a physical disability, it's so visible, and so people can't help but notice our particular adversity, whereas somebody else walking down the street, you may not know what they're going through in their life. I think that, like you were saying, it makes you a role model, and people can't help, but see it and notice the way that you're living and want to follow suit. Does that make sense?

Chris Anselmo: Yeah. I think you hit the nail on the head. I think one thing that, as I've kind of grown in how I handle this condition, one of the things that I've been thinking about a lot is, obviously, if I do any writing or public speaking, I'm kind of conveying my story. Obviously, you don't want to come off as an inspiration to the point where people would take pity on you, that just drives me up a wall.

Mindy Henderson: No, of course not.

Chris Anselmo: But I do feel like, rather than being like an inspiration where that happens, where pity happens, you can be an inspiration where you actually change behaviors in other people's lives, in terms of how you dealt with the challenge, how you live with it, your attitude, how you treat other people. And that's what I've really tried to focus on is I'm no more "inspirational," I say that in air quotes, than anybody else, but I have a certain set of circumstances that I'm living with, that I've struggled with, I try not to hide that, that it has been a challenge. But I think something you had said earlier, that everybody's sort of dealing with something, whether a physical disability, whether it's something going on in somebody's lives, and I just try to always think that.

Everybody's got something holding them back, something that they're fighting against, mine just happens to be physical, but that doesn't make me... I mean, I could be inspiring, but I'm not any less of a person, because I have this. I'm not diminished in anyway, just because I have to do something in a different way. My

muscle weakness is just, it's a part of who I am, but it is not all of who I am. And I've been able to connect with people a lot in that way by just not being afraid to share what I shared earlier on, when I was saying that I really struggled with this.

It's important to be optimistic and to also just acknowledge that, yes, certain things are difficult, but life just as a whole is not an easy thing to get through.

Mindy Henderson: Absolutely.

Chris Anselmo: And just anything that we can do to just be authentic, to be a good role models, to connect with people, I think is probably one of the most empowering things that you can do. And you could really positively change people's lives in a way that isn't strictly, just as an example of just somebody that like, "Oh, I'm having a bad day, but at least I'm not him," that kind of thing. So avoiding that, I don't really know what to call it, but there's definitely a way that we can convey our story that is uplifting, that is meaningful. And that's been one of the greatest blessings of having this disease. It hasn't all been bad. I have been able to connect with other people in ways that I don't think of what have, if I did not have this, and I don't take that lightly.

Mindy Henderson: Absolutely. Absolutely. And like you say, I think just storytelling in general is a powerful thing. And whatever your story is, I think it empowers other people to know that they can tell their story too, and they can talk about their challenges and their hard times. And I think it helps us all feel a little bit less lonely when someone one shares their story and can give that to somebody else. And to let them know that they're not alone in their struggles.

Chris Anselmo: I think it helps to just create a sense of connection that I think this world desperately needs.

Mindy Henderson: Absolutely. So Paloma, what's the biggest lesson that you've learned as a result of going through all of this? And what advice can you give to someone who may be listening and going through something similar?

Paloma Juarez: I think when talking to other parents that go through things like this, whether it be somebody whose child maybe is having a learning issue at school, or another Pompe's family that just got handed this diagnosis and feels like their whole world has been devastated. I think the important thing, and I think both of you have done a really awesome job of touching on this, and it really does help me in our journey and the kind of adult that we want Vaun to be, can he one day have the perspective that you guys have, because we've given him tools to do that? I think the most important thing is, you've got to, A, allow yourself to feel what you feel, and embrace those feelings, and understand that what you felt yesterday may not be the thing that you feel tomorrow. But, B, really, again, understanding that you are never alone in this, there's community out there that certainly does really help.

I never really understood what that idea of it takes a village to raise the child, but it certainly does. Whether it's, again, a disability or not, I think the more relationships and community that you can have, the better human being you're

going to be, and the better experience. I mean, I think everybody, like you guys have touched on, you just want to be able to connect and that's a huge thing. And sometimes I think when you get these diagnosis' you worry, "Am I going to be able to connect? Will my child be able to connect, because they're labeled different?" And so I think that's it, it's really just embracing your feelings and then really just grabbing hold and reaching out to your community, whatever that is. I think all of those relationships really matter and really help you along the way.

Mindy Henderson:

That's really good. And I think that, for me, the biggest lessons that I've learned along the way, are number one, I truly believe that there is always hope. As dark as something may feel, hope is just the belief that something can be different or get better. And I believe that there's always hope to be had, regardless of what the situation is. And the old adage of, this too shall pass, it will. This hard moment that I'm having because of X, Y, or Z limitation, that moment's going to pass, and I'll laugh again and I'll have more experiences and things will be okay again.

And I think the other thing that I've learned as I've walked through my life and figuring out how to make things work, if you're at the very beginning of this, you may feel overwhelmed by the things that you believe are not going to be possible. But like Chris was saying, it's just a question of looking at things a different way and asking one question at a time,, and finding the answer to that question and the way to make that particular piece of the puzzle work. And then you move on to the next question. That's been a process that I've used again and again and again in my life, that's gotten me to college. It's gotten me to living in an apartment, independently. It's gotten me to driving a car. So I think that's the advice that I would leave the audience with as well.

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