**Interview**

**Mostyn’s Story with Zoe Hummel Script**

**Narrator:** Are you searching for a meaningful Mother's Day gift? A gift that goes beyond the ordinary when that truly touches the heart? Well, this year you can let hope grow by donating to n-Lorem to support nano-rare patients in honor of a mother in your life. With your donation, n-Lorem will send a beautiful Mother's Day card with a personalized message directly to your loved one. But that's not all. As a special bonus this year, n-Lorem is including a decorative wildflower seed packet so that your loved one can plant the seeds of hope directly at home, watching as they bloom into a beautiful reminder of your love and support. Mother's Day is just around the corner, so don't wait. Visit nlorem.org today to make your donation and order your let hope Grow Mother's Day card and seed packet. n-Lorem, bringing hope and potential help to nanowire patients and honoring the mothers who inspire us every day.

**Stan:** Hello everyone, I'm Stan Crooke. I'm chairman and CEO of n-Lorem and I'm your host for the n-Lorem podcast series, the podcast series that focused exclusively on addressing the needs of patients a families with nano-rare mutations. Today we have a very special guest, Zoe Hummel, who is a mother of one of our patients. Zoe, welcome.

**Zoe:** Hi, thank you so much for having me here.

**Stan:** Oh, I'm delighted to have you and I should begin by thanking you and Clayton for organizing the opportunity for me to make the show. I actually got to throw out the first pitch for the Yankees spring training game in Tampa. Thanks to Zoe and Clayton. And I would say that I was quite a star. I threw a. Real stripe say that.

**Zoe:** I think you totally did a real strike for sure. You did great.

**Stan:** Yeah, well, you know, it's been some decades since I threw a ball. So, and I lost my baseball glove long ago, so Roseanne ordered a glove, which turned out to be weight like a like a 5 year old's glove, but it didn't matter anyway. She runs screaming when I throw the ball, so I never got to practice until the morning of the game, so I got to throw a few. And I fortunately didn't fall off the mound or break my leg or do anything else, and I didn't hit the mascot, which is a big plus.

**Zoe:** He did really good. We've been to a lot of baseball games and we've seen a lot of first pitches and you did great. You did excellent.

**Stan:** It was a real thrill. I mean, I mean, it is the Yankees and to stand next to those uniforms and everything was pretty cool. It was nice to share it with you guys in Mostyn and so on.

**Zoe:** We were happy you were able to come down and do that. Yeah, for sure. It was our honor.

**Stan:** It was a lot of fun and so Zoe and Clayton have a child, Mostyn, and I'm told to think of Boston and say Mostyn which Zoe and I have discussed is a Welsh name and when I looked it up it said moss covered rock, but she tells me no, it's a fortress. So I Accepted that.

**Zoe:** Fortress in a field in a field.

**Stan:** So, I think let's begin where I guess these things always begin. Really, how did, how did you and Clayton meet and we don't get in need to get in details but just have to meet and get together.

**Zoe:** Right. Clayton and I actually went to the same high school together and he asked me out in when I was in my 11th grade year and he was in his 12th grade year and I of course, said no, absolutely not. And he continued to ask me out a couple of times. And I just kept on telling him no. And then eventually I kind of broke down and he took me out and I brought my brother with me, actually, which probably didn't go as he planned, but we did have a good time. But that was it. And then a couple of years later, he was in college, and he was coming back for summer school. And I was getting ready to go into college. And we ran into each other and we kind of hung out during the summer, but then again, never really went into anything more than that. And then five years later, we met out at a restaurant and then I ran into him. He literally walked in the door and there he was with some friends. And he, like, looked at me and he was like, what are you doing here? And I was like what are you doing here? And then that was kind of it.

**Stan:** Well, he had gained a great deal of sophistication and poised by then, I'm sure.

**Zoe:** I actually he invited me on a date and I thought, you know, I don't know if I really wanna date this guy. I'm not really sure if I'm really into him. And I said I'm going to invite him to a jazz concert for the school that I was going to at University of South Florida, and I thought he's going to go and he's going to hate it. He's not going to like it at all. And I took him and we had the best time, and that was it. I literally called my best friend and I was like, I love this guy like, so that was it.

**Stan:** Well, it seems to have worked out pretty well. And Speaking of jazz concerts, I think it would be interesting just to spend a minute or so on your on your musical history and experience. Nice cat.

**Zoe:** You know he's. Very curious right now.

**Stan:** You know, and if my golden retriever were to come in, she would take over the conversation. So count your blessings. You don't have a loudmouth golden retriever, but you are a very accomplished violinist and have been in a number of productions and travel with a lot of different groups, including some rock bands that I know a bit about and other musical organizations. And just tell us a bit about how that felt, and it must have been pretty exciting to be in front of large crowds.

**Zoe:** Yeah. Oh my gosh. Yeah, it was. It was pretty incredible. But the first I think the first time that I kind of gotten taken by it was when I was touring Rod Stewart, and we actually played a show here in Tampa. And I remember being on stage and it was like as soon as you could hear the crowds screaming and screaming, and as soon as they lifted up the curtains or the show started. I mean, I felt like I wanted to throw up coughs like, cry, laugh, all at the same time. It was just the most incredible feeling I've ever had. It was amazing.

**Stan:** And you've done a wide range of music from hard rock to classical music and everything in between.

**Zoe:** Yeah. Yeah, I was classically trained. I started when I was two years old in Canada, and they have a big following called Suzuki method. It's a year training and my Sunday school teacher told my mom, hey, you know, you should have your daughter play violin. She's got perfect pitch. So, they started me at two, and I actually started playing when I was three because when you're two, you can't really play the violin. They just want to make sure you don't drop it. So, what they gave me was a wooden a little cardboard box with the ruler and a chopstick. And I learned how to hold the violin through the songs without dropping it. And once I could do that, then I was ready for my actual real violin. So, I went through all of those books and then I continued on through high school, did all of my you know encore, you know, first chair kind of stuff and played in the orchestra. And then I tried to audition to get into school for a violin and I did. And I ended up going to USF because I had a full scholarship there, and that just kind of led me to the right people. I just kind of kept them going through and I got did operas and I started doing some Broadway stuff and that led to more and more things, and then I was able to start getting into shows and touring with different artists and you know, being on stage and going on radio shows and it was just really fun. I had a really, really good time.

**Stan:** You know, people do it and they complain about it, but the truth is they love it most of the time, right?

**Zoe:** Yeah, I feel very blessed. Very blessed.

**Stan:** I would. I would bet that you do. And of course, you continue doing that even after you're married.

**Zoe:** Yes, yeah, I was doing a lot of chores and then right before we got married, I was supposed to go on tour with Brooks and Dunn and Alan Jackson. And it was a six-month long tour and Clayton was like, listen, you've been doing a lot of tours now. Maybe we should just get married and like maybe not do the tours anymore. So, I said okay, we'll get married. So, we got married. And I just, I still played the violin, but I just did more like recordings and session work exactly, and you know corporate events and things like that, and I loved it. I was hoping I was going to continue to do that you know, all throughout. But I feel like I said, very blessed that I had all the opportunities that I did to do the things that I was able to do.

**Stan:** And then Mostyn came along. So, talk to us about Mostyn. There was, you know, birth and when it became apparent something wasn't quite right.

**Zoe:** Yeah. So, I had a great pregnancy. I, you know, was really, really dedicated with my diet and all the food and everything. I wanted to make sure I was as healthy as I possibly could be, and I felt great. I remember when I got pregnant and I went to my doctor's appointment, he did the lab work, and I was 32 at the time. He comes back and he goes man, and he goes your lab work looks great. You like a 25-year-old and. I like looked at Clayton and I was like, yeah, see, I told you I was. So, I was very proud of myself. And so, I had a great pregnancy. I didn't have any issues with anything, had a natural birth with no, and nothing I didn't have after or anything, no medication. He was like only six hours. I mean, it was a really easy birth, and he came out. Everything was great. We had no problems. We went home the next day, and he seemed like at the time that everything was going well and as time progressed I started to kind of notice some little things about him. Just tiny things that were just a little bit off they seemed like to me, like he never crawled. He always like army walked first and then he went from army walking to standing and walking. And it wasn't delayed. It just wasn't, it was like 14 months which is not delayed, but it's just still not where you want to be, but I kept on to the pediatrician, and I say, you know, like he's, you know, he's not what he's walking kind of looks a little bit odd. It's like, oh, you know, it's okay. He just started it. And then I say he's not really saying a lot of words and he's kind of saying some just sounds and stuff he said he's a boy. Boys are always really, really slow and I was like, okay. I mean, he had good eye contact. He smiled. He did all those little cute baby stuff, but I wasn't really sure if he was progressing the way he should have. And then about when he was two years old is when I had him tested by our local my organization here. It’s called Early Steps. I don't know if they have that in other places, but it's called Early Steps and basically, they come out to your house, they evaluate your child and they think do they need speech therapy, physical therapy, occupational therapy. So, there was a woman who came out, she assessed him and she said, yeah, I think he could have, I think he could have all three of them, and I was like, oh, okay, so I told the pediatrician and he was like, no, I wouldn't worry about that. I think you're just getting yourself all stressed out. So, I was like, okay, fine. So, we just went along. Not really trying to push that thing, thinking that he was a boy, he was just going to eventually, you know, he wasn't sick. He seemed like he was happy; he was eating well and all that kind of stuff. So, I just, we didn't really push it. And then at three I remember my mom actually watched Mostyn and when we came home because we had gone out, we went to the Bahamas just for the weekend. And when we came home, I remember she said to me, you need to get him, we need to take him to like a doctor. Please don't be mad at me for saying this to you. But you do need to check. Have him tested by like a neurologist or something. There's something off. And I was like, okay fine. So, I actually took him to another speech therapist because I was like, well, he's not talking. So maybe you know. So, she evaluated him, and she said he absolutely needs speech, but he needs everything. And I was like, well, what do you mean he needs everything. She goes and started asking me all these questions about my birth. Did he have the correct oxygen. Like the question she was asking me seemed like she was trying to say that he had cerebral palsy. So, then I said, do you think he has cerebral palsy? She said I'm not a doctor, I can't tell you that. So, I was like, she's like, I just think you need to another pediatrician and get him really evaluated. I called Clayton and said, listen, you get an appointment with somebody. So, we got an appointment with the neurologist, and when we went to the neurology appointment, it was a of somebody that Clayton had worked with because he's in the medical industry. We went to the neurologist, and he evaluated him, and he said that he has cerebral palsy. So, we couldn't understand why he had cerebral palsy, because like, like I said, his birth was great. We didn't think anything seemed to be off. So, we scheduled him for an MRI and when the MRI came back it came back normal. So, we were still kind of like okay, so this our kid has cerebral palsy. And you know what else? What are we going to do now? So, we eventually got him into speech and physical therapy and occupational therapy, and it was all under that diagnosis of cerebral palsy. And everything seemed to be going okay. He kind of seemed like he was maybe progressing a little bit. He was a developmental delay, so you know, his words were always a little bit off and he was having a hard time, you know, fine motor skills. But he seemed like he was getting better, and at that point we were just like, okay, he seems like it's getting better, and I thought, well, you know he's let's put him into school. So, we put him in preschool, and I went to the preschool, and I stayed there, and I would go there during the day just because I felt kind of strange leaving him. I wasn't sure if they knew what was going on with him or anything like that, so they let me come to the school, which was great. And then eventually, like I said, he started to seem like it's getting better. And so now I would drop him off. And so, everything was seeming to go a little bit better and then we were going to put him into a Montessori school. That was right before he turned five, and oh, he actually was five. Sorry, right around five. We were going to put him in Montessori. And we were really excited about it. And I had met with the school and everything else. And then on December 31st, 2016, he had his first grand Mal seizure in the morning, and it came out of nowhere and we had no idea what to do. I mean, I remember calling out to Clayton and saying he's having he's having a seizure and he's. I'm like, what do I do? He's like, put him on the side. We put him on his side, called 911. And that's kind of what happened before that. Yeah.

**Stan:** So, let’s stop there for just a minute. And a story that we hear frequently, and it's a really tough balance. That's right, because Mom and Dad and Grandma and Grandpa are always the best observers. But we're also the most concerned, and good pediatricians will say all the things your good pediatrician said he's boy, he's in the normal range. If you could distill down to a single sentence or two, the lessons that you think are important out of that first part of your journey.

**Zoe:** I think if I knew what I knew now I know that I would have, I would tell somebody - listen in your gut if something feels wrong. You need to get it addressed and you need to find out why it is what it is you know. I should have. You know, everybody's taking them to get therapy and stuff like that. It's scary. As a mom, you know, you think something's wrong with my child. But if I was blessed to have another child now the minute I saw them not doing something they're supposed to be doing, I would take them to a specialist immediately and try to figure out why they weren't doing what they were doing.

**Stan:** The bottom line is trust your instincts.

**Zoe:** Trust your instincts. If you feel like something is wrong and something does not seem right, get the answers that you need until you feel like it's right.

**Stan:** Yeah. And you know from my perspective, a lot of this would be so much simpler for parents, pediatricians, and it's such a dramatic improvement in the world if we sequenced all of our newborns.

**Zoe:** Yes.

**Stan:** I mean, you'd have the information and then you would have the option of doing what needed to be done. But if you had that information, you knew there was a mutation that might be a problem you and the physician and everyone else involved would have been at a different level.

**Zoe:** Yeah. Totally you would be way more prepared mentally, emotionally and everything absolutely. Absolutely.

**Stan:** As an aside, one of our major goals at n-Lorem is to assure that genomic sequencing is incorporated into newborn evaluation protocols with appropriate safeguards. And there's now a significant lobbying effort going to drive that and the fact that we're able to treat some of these patients is, it's an important step. So, now you've got seizures, and there's very little that's more frightening for a parent than to have that first seizure happen, right?

**Zoe:** Yeah. Yeah, it was terrifying.

**Stan:** Yeah. And the truth is, there's nothing you can do. I mean, wait it out, right? Yeah. So now you're on high alert and you went back to the pediatric neurologist would be my guess.

**Zoe:** Yeah, we called him that day. You know, we took him to the hospital and they called him, and he put him on Trileptal. And we started on Trileptal. And within a day, he started developing a rash. So, we called the neurologist. And we said, hey, he's got a rash. He said, yeah, you got to stop. He can't be on that. So just immediately stop. Well, he didn't get his nighttime dose of that, and he immediately had another Grand Mal. So now we're really getting scared now because now, what are we going to do? So, we called him again and he put him on Keppra, and he said, hey, come to the office tomorrow. We need to an EEG on him. Yeah, EEG, EKG, I always get those two confused but EEG. So, we took him to his office, and they did an EEG on him. And for just a 15 minute one and he came out and he had a piece of paper, and he had a circle on one and a star on the other. And he says okay your son has got juvenile onset epilepsy can maybe go away by the time he's 8. It's not nothing like you need to be too worried about. It's going to go, probably go away. And I looked and I said, well, okay, you circled that but what's the star on that like, what's that from? He goes. Oh, don't worry about that. That's really, really bad. Well, come to find out, he had the really bad one, which is Lennox-Gastaut syndrome, and he didn't mention it that day. But I remember reading it and I remember kind of getting scared about that after I researched it and yeah, it was just so we didn't get diagnosed with that until a little bit later. It gets up, gets worse because the Keppra does not work, and he continues to have seizure after seizure after seizure. So now we're kind of flipping out. We're like, not sure what to do. We've never been around like seizures before. We turned off all the lights in the house. We put caution tape on our front door. Like I'm not kidding. We actually really did this. Turn off all the TV's we like, literally went like silent. We didn't want anybody to bother nobody. Knock on the door because he was having so many seizures and during that time, I had already tried to get into Boston Children's Hospital and that wasn't related to seizures. I was just trying to get into it because he got diagnosed with cerebral palsy and I heard that they were really good with neural, like neurological stuff. And I thought, let's try to get there was like a six-month waiting list and I was on a waiting list. So, I didn't think we could actually get into Boston Childrens. And I told Clay, and I was like, I don't know if we can try to see if we can get him there because we know it's really good claims. Well, Clayton's like okay, well, let me call around and see if I can find anybody that might know how somebody up there. So, he called one of his doctor friends. And he is a good he went to Boston, he went to Harvard, and he knew a lot of people around there. And Clayton was talking him on the phone. And he's like, yeah, I've got this friend. He says, I've got this friend, he's big philanthropist up there. His son has got a neurological problem. And he, like, made the wing over there at Boston Children's. He's like, I can tell you his name he's like but I don't know his phone number. He's like, I haven't talked to him in, like, 10 years. And Claytons like, yeah, just give me his number and I'll find him. I'll try to figure and I'll connect you with them so you can maybe help us. And he's like, he's, like, fine let's do that. He's like hey. Wait, wait. Hold on. Wait, wait. Wait, wait, wait, wait. What? And Claytons like what? What? What? He's like, you're not going to believe this right here on the desk, on my desk in the middle of my desk is this letter from this guy, his numbers right here. I've never I haven't spoken to him ten years. So, I'm going to call him right now. So, he called him, and within, I'm not kidding ten minutes clayton had the head of neurology at Boston Children's emailing him, saying, hey, can you be here in a week? We can get you in. So that was the mission. We're going to pack up everything. We're going to drive because we don't want to fly with him. We don't know how to deal with them with seizures. And we drive to Boston, and it's in January. And it's absolutely snowing like crazy, and it's black ice. And so, on our mission there they were taking my car and it was an SUV and the tire pressure kept on you know coming out so Clayton had to put air in the tires, and it keeps getting worse and worse and we get to this one stretch of area and it's we're I don't even know. I think we're about a day away from Boston. And we keep on trying to fill up the tires and somebody had cut the hose of the air pressure thing for like 15 miles. It was crazy. And it was in the freezing snow. And he's trying to get this pressure. So, we finally get to the dealership, and barely like almost like with the rim like not hopefully not damaged. We're thinking hopefully the rims not damaged, we get to the thing they take off the tire. There, he says, just put a new tire on there. Let's get going. The guys like, listen, we're not going to be able to put a new tire on this thing. The tire is not your problem. It's the rim. The rim is cracked. And we're like what he's like, yeah, he's like, and I can't. He's like, I can give you, like, a smaller car, but I can't. I don't have anything's going to be able to fit all the stuff that you need. So, Clayton was like alright, let's just call the dealership in Boston, tell them what we need, they'll have it there. And Clayton went to Home Depot and he buys gorilla glue and gorilla duct tape. And he glued and duct taped the rim. And we drove. The next day, through there to Boston to the dealership, got the new rim, got the thing literally. But like just like 5 minutes before our appointment parked, gotten in there and there we were at Boston Children's Hospital. So, we made it.

**Stan:** Literally a long trip and.

**Zoe:** Oh my gosh.

**Stan:** Yeah. And you know, as I listen to it, I think about all the patients we deal with, and I think as terrible as things were for you guys, you were unbelievably fortunate in some ways. Clayton was in the medical field. He had a friend who knew a friend and suddenly had an appointment that you couldn't get, and that to me, is a part of what the problem that they shouldn't have to have a friend of a friend to get care for your child, right?

**Zoe:** No. Now it's yeah, if we were stuck to try to find help here in Tampa and Florida, well first of all, at the time the genetic mutation that Mostyn has was only on the Boston Children's Epilepsy Panel. It was not anywhere here in Florida, so we would have done the testing here and they would have never known what was wrong with them and they weren't even getting it. So, I mean. Yeah, it's really, really sad when you think about it because we were very blessed. Like you said, we were very fortunate that we had that contact. Otherwise, I don't know where we would be or what we would have done.

**Stan:** So, you end up at the BCH and they immediately ran, I assume, that panel of genetic tests that is still a tiny fraction of all the genes that we know cause it, and out of that came a real diagnosis of what the problem was, I guess.

**Zoe:** Right. Yeah, they told us that they wanted to do the genetic testing because epilepsy in general is either unknown, structural, or genetic, and we knew it wasn't structural because he had the MRI's done and everything seemed normal. So yeah, we did the genetic testing and when it came back, he had three misspellings on two of them were on dominant genes but they were not something that they felt like was necessarily like an issue because Clayton had one of them and I had the other one and it was fine. I was fine. He was fine with no seizures. And then that KCNB1, one is the one that they found, and it was not from me or from Clayton. It was like as the genetic specialist at Boston children said, she said Mostyn is just very unlucky. That's exactly what she said.

**Stan:** That's exactly the right word. You know, one of what people don't understand is how complex that you have 6 billion genetic letters and it isn't boxing. It's, you know, a illiterate medieval monk. You know, in a candlelit cloister copying each letter and there are mistakes and you have all kinds of systems to correct it, but we're all walking around with hundreds of millions of these mistakes. And yet and Mostyn has the same number and yet he has a bad one, and that's bad luck.

**Zoe:** Yeah, yeah. When she said that, I was really taken by that because I was thinking like here is like a genetic specialist and she's actually saying that. So, it really, really struck me when I thought about it, you know, it's like here you have Mostyn and he has one gene that's causing everything that he has.

**Stan:** One actually one nucleotide in one gene, one extra nucleotide mistake in 6 billion. So now you know all that, and you have a diagnosis, and I suppose the next step is to talk about what the gene does. You want to just you just tell.

**Zoe:** Oh, what the gene does. Yeah, it's a potassium channel genetic gene KCNB1. And it has to do with excitability and basically, he's yeah, I mean, you're like, why am I explaining this? Not my forte.

**Stan:** Well, you know the fact is we're electrical machines and what our cells do is establish electrical fields by keeping charged particles out and some charged particles in, and we call those ions. And so those things are called ion channels and this is potassium ion channel and you can't have too much and you can't have too little. You have to have just the right amount. It's a Goldilocks kind of an affair for all of these ion channels. And so, whether it's a mutation that causes it to be overly active or less active, you end up with electrical currents that are wrong, and seizures are a part of the syndrome that had happened there. So, you have a diagnosis, now you understand that there is a problem and you can name it.

**Zoe:** Yeah.

**Stan:** And that's good. But it sort of the next sentence out of the physician's mouth probably was and there's no specific treatment for this that we're just going to have to use medications the best we can.

**Zoe:** Yeah. And you know, when we were going through this? With, you know, he's gone through almost 17 anticonvulsants. So that means, you know, in the in the seven years he's had this, we've started a drug. Realized it didn't work. Now mind you, we might be adding another drug. So, say he's on two now. Nothing is working. So, you have to get off the drug, start a new drug. And there's all these different levels and you're just going back and forth and back and forth, and it really feels like the doctors, you know, they don't know what's going to work and what's not going to work. And they're like, almost just throwing darts and like, okay, let's try this drug. And we're like, okay, why do we think this would be a good one, you know? And for Mostyn situation, the potassium channel thing, unfortunately, there is no anticonvulsant that's specific for potassium channel. There was one and they had to take it off the market because it turned the people blue. There hasn't been another one. So, we're really working with a lot of drugs that deal with sodium channels or other types of epilepsy, not potassium. So, it's really been hard. He's on three anticonvulsants and he still has seizures almost every day. I mean, it's just. It's crazy and it's sad. It's really sad watching, you know, somebody that you care about and go up and down on these medications because it doesn't make them feel good.

**Stan:** You know, and you know that the seizures are going to make the developmental delays worse. And I'm sure everyone told me that.

**Zoe:** He's yeah, he's declined so much, so much. I always tell, I tell people like if I showed you videos of him when he first started having seizures and I showed you a video of him when he was three years into seizures and then in five years and then now seven years like you would see the decline. It's just awful, you know, his verbal ability, his walking ability, everything about him. He's happy. You know, we try our best to keep him super happy and you know he's got the most beautiful like sweet heart and he's just he loves people. But yeah, he's absolutely declining. He's declining. And I don't know when the next seizure. Honestly, I honestly, I don't want to say that I don't know what next season is gonna kill him to be honest with you. That's the really the truth.

**Stan:** This is a good reason to cry bad, yeah. So just a couple of more thoughts about emotions for a minute. The response to illness is pretty universal. It's first fear and then anger at the unfairness of it all.

**Zoe:** Yeah.

**Stan:** And you actually understand how unbelievably unfair it is so how have you and Clayton managed? Through that emotional thicket of hope feeling, terrified, angry. No place to put your anger. Who can we blame? Nobody. It's just bad luck. How do you work your way through all that to a place where you can function and have a constructive life.

**Zoe:** I think like most importantly, is our faith. You know, we have the same belief. So, I think that's gotten us through a lot of those trials and tribulations and stuff. But I also think, you know, Clayton is so incredible about not just settling and he's about taking action and you know, we both try to do our best to research and really figure out what can we do like we have to be able to do something. There's got to be something you can do, right. We gotta figure out what can we do? And I think those are the things that have really helped us faith and just trying to get take action and try to figure out what else we can do. Find something. Whatever.

**Stan:** So, work your way through it. Find a way to convert it to steps that you can take, whether any of them work. At least you're doing your best and you know that.

**Zoe:** And it's definitely not, yeah, and it's definitely not easy. Like, it's not easy at all. It's really been challenging. It's very, very hard. You know, there's a lot of times where we feel extremely defeated, you know, but.

**Stan:** So now you've got a diagnosis. You know there's no specific treatment. You know that there's research going on in genetic sequencing that's happening and lots of things are going on and you, you spend your time now instead of making music instead of enjoying life as you once did, becoming a scientist. And a geneticist right?

**Zoe:** Yeah, Clayton really is the researcher on all that kind of stuff. Basically, the minute once Mostyn started having seizures, I just said I've got to take care of him full time.

**Stan:** So, life changed.

**Zoe:** Life changed a lot, yeah. Everything changed. I really felt like I lost myself, to be honest with you. Like as a person. Because the violin was really important to me, you know? But yeah, it was. It was a really challenging time in the very beginning when all of this went down. And I think Clayton felt the same way too. I mean, we were just, we had to do things differently. It is a lot differently not, you know, not just a little differently, a lot differently. And yeah, it is extremely overwhelming, overwhelming.

**Stan:** So now the next step is to find somebody who might be able to help, right?

**Zoe:** Yeah. So, Clayton had, like I said, he was all into the genetic stuff. And he asked a couple of doctors there about CRISPR and things like that, and he's trying to figure out maybe if since we know it was a gene thing, maybe there was something and we did find a company and we the first time they met us there were like we're going to be able to cure your son. We can cure him. And I was like, okay, and it was like, you know, you're telling me that. I mean, I actually when he when they told me that I remember I burst into tears because I couldn't believe someone was actually going to say that to me and tell me that they could do that. So, we had a lot of hope at that point and we started the process, but that was really expensive. We had to pay for it. I mean, do you want to know how much they wanted?

**Stan:** I'm appalled that anyone would ever do something like that, but I know it happens all the time. Nothing really important to hear. I would assume more than $1,000,000 would be my guess.

**Zoe:** Yeah. Yeah. So, they gave us like a plan and they told us that steps that we needed to take and that basically for them to be able to do what they needed to do to help Mostyn that it would be $4 million.

**Stan:** You know how that disgusts me? Do you know how infuriating I find that? One that company that would do it, two that they would give you the false hope of telling they could cure something they know nothing about, and then want $4 million from you.

**Zoe:** Yeah. So, Clayton is like what we need to like start selling things, and you know I mean we had to raise, we were thinking of how we raise money and we were trying to do whatever we could to start this thing because we wanted this just to get going and I mean, it was just really, really stressful. And then that company they were working on it. They weren't doing anything. They ran his genes and then we that we kept on checking in with them to see if anything was going on. We started like an initial payment with them and then a company came in and bought that company out. And when that happened they didn't want to be a part of helping Mostyn, so they yeah. Basically.

**Stan:** They didn't want to be a part of helping because they knew they couldn't.

**Zoe:** They couldn't probably, and they called us and they said we can't do anything for your son. They're like, we're sorry we can't do anything for your son so then I was like, really, really, devastated.

**Stan:** How long? What was the time?

**Zoe:** They told me it was. It had been like almost a year had gone by.

**Stan:** Then and then somebody sane comes along and says, hey, they have been telling you the truth basically.

**Zoe:** Yeah, we're not going to help you at all.

**Stan:** So you know, and this is story we hear. And so, I suppose, what sort of lesson we take from that that you think would be worth sharing with folks?

**Zoe:** Oh man, hopefully everything that you hear I don't know. I mean, I don't know. I mean, we have so much hope and it was really, really defeated. I don't even I mean I guess if I was going to tell somebody a lesson with regards to that situation is I'm going to say this because I think this is a good thing. It's okay, you know, it's going to be okay and let's try again. That's okay. It's going to be okay. Let's try again, and see if we can find something else.

**Stan:** So now n-Lorem comes along.

**Zoe:** Well, that and one thing, honestly, if we hadn't have gone with this other company, we wouldn't have met a nice man that worked for that company that told us about n-Lorem. And so, it all kind of worked together, to be honest.

**Stan:** How much? How much financial information did n-Lorem ask about you?

**Zoe:** They didn't they now they. So, we talked in the beginning. We had an interview. It wasn't with you, it was with Frank Bennett. And he met us, and I remember he was asking us some questions about Mostyn. He met Mostyn and then the very end, you know, Clayton says, well, you know, let us know what we need to start to like get, you know, to give you money and stuff and you said oh no, no, no, no, no, no, no, no, no, were not gonna make you pay for anything. This is for free. And I just burst into tears. I couldn't believe it because here we're, like, trying to figure out how to, like, refinance our house and, like, do all these things, try to help our son. And now this company is going to try to help him and they're not going to charge us anything for it. It was like it was like a miracle, to be honest.

**Stan:** I think if I were and I think your lesson is a good one, which is get to pick yourself up and dust yourself off and keep trying. But I think if someone promises security, that's the person to avoid. I guarantee you you've gotten no promises from n-Lorem, other than we'll do our best.

**Zoe:** Yeah. Yeah. No, I know.

**Stan:** And I think if someone asks you to pay, you really have to ask, why? And it may be legitimate but you know, I don't really think parents should have to pay for the care for their children. That should be something that we find a way to do as a society and so of course, we at n-Lorem are unwilling to accept financial information about families who apply. We don't have a place on the phone for it, and you and Clayton have been generous in trying to help us raise money, and we find that. But that's a very different proposition from charging somebody and making promises.

**Zoe:** We like, I mean we do that because we are so honored that you guys are wanting to do this to not just for Mostyn, but for all these other children. And because these families, they get to a point where they don't have hope. They have a child that has got something that nobody else has and most doctors just say okay well, I don't know what I can do. I can medicate them, I can. You know you can do therapy. One doctor wanted to cut Mostyn's brain in half. He thought that was going to help. And I mean, there's so much stuff and you just look at your life and you're thinking, is this going to be it? Like, is this going to be it? And so, we believe in everything that n-Lorem does. And like I said, we're honored to be a part of this because we believe in what you guys are doing for other families and other children, and it's going to help change the world. It really will. Because these kids are beautiful people and they deserve to be able to have a chance, you know.

**Stan:** One last question, you and Mostyn and Clayton came to our first colloquium. I can now pronounce that word. And we'll call it a meeting. That must have been a tough trip. I mean, Mostyn’s a happy boy and he loves baseball of course and he likes, you know, knocking things out of your hand, which is fun.

**Zoe:** Oh my gosh, little stinker.

**Stan:** How'd you manage that?

**Zoe:** Yeah, that was a little bit stressful because, you know, we wanted to make sure, because we you know, it was a professional meeting, so we wanted to make sure that Mostyn was at his best behavior, but he's Mostyn. So, you know, just we've gotten really good at kind of trying to maintain him the best that we can. You know, we know the things that he likes. And try to keep him entertained. Plus, he loves people and so he did great. I mean it's I always kind of we kind of go into a room and we kind of scout it out and we go, okay, that corner over there is where we need to go. We're going to put him over there because, you know, nobody will be behind him, nobody will be beside him. And then we can keep them kind of contained. So, once we do that, it does help. But he does get a few drinks or he doesn't get some plates of food every once in a while.

**Stan:** Oh, he was one of the stars of the meeting. His mom, you know, did okay too by playing violin or at our reception so you know, I know that this has been not altogether fun for you, but I think our listeners will appreciate you're sharing the story and some of the basic lessons that you've learned and not you know I think to sum up, the most important basic lesson is you just have to keep trying.

**Zoe:** You have to keep trying. Yeah, you have to just keep trying go forward because somebody, somewhere out there there's a Stan.

**Stan:** Absolutely. There's an n-Lorem. Anything else that I haven't asked, or any other comment that you'd like to make before we call it a day.

**Zoe:** There's an n-Lorem for sure. Now I mean nothing. I mean, I just like I had them like, we're honored to be a part of this group and we're really looking forward to the future, you know.

**Stan:** We're doing our best and I wish I could promise a positive outcome, I can't.

**Zoe:** You know, but we're just, we're happy that, you know, he's even accepted. So, we're really excited.

**Stan:** Wonderful to chat with you again and thanks again for bringing my baseball dreams to a fruition where I got to play with the Yankees for a pitch. And you give Mostyn a kiss for me.

**Zoe:** And Mostyn will give you a big fist bump. And probably knock a plate of food over, but other than that.

**Stan:** He's done before, okay, okay, take care.

**Zoe:** Thank you so much. Alright. Alright, bye bye.

**Narrator:** n-Lorem is a nonprofit committed to discovering and providing personalized, experimental treatments for free, for life to patients with genetic diseases that affect 1 to 30 patients worldwide referred to by n-Lorem as nano-rare, many of these patients progress and die without ever achieving a diagnosis. This is where in n-Lorem comes in. They do the impossible by providing hope, and for those that they can help, free lifetime treatment. For more information about n-Lorem or today's episode visit nlrem.org. Any questions can be sent into podcast@nlorem.org. Search n-Lorem on Twitter, Instagram, YouTube, LinkedIn and Facebook to connect with us. This video is hosted by Dr. Stan Crooke and produced with the help of the following professionals. Thank you for watching.