**Transcript**

**Susannah’s Story with Dr. Wendy Chung and Luke Rosen**

Stan

Hello, I'm Stan Crooke, founder, Chairman, and CEO of n-Lorem. Welcome to the n-Lorem Podcast series. This is a podcast series that focuses solely, exclusively on the needs of nano-rare patients. that is, Patients that have a unique mutation that affects only them or are a member of a of a tiny group of patients of less than 30 worldwide. Today I'm privileged to have two guests. Luke Rosen is a parent of one of our patients. His seven-year-old daughter Susannah. Luke has had three successful careers. He is an actor whom you have probably seen in shows like Law and Order, and Orange is the New Black, and, you know, a number of other movies and media. And then, when he learned that Susannah was ill due to a rare genetic disease, he founded a patient efficacy group, which he continues to lead. And in the process of learning about Susannah's disease, he became such an expert that he was asked to be Vice president of patient engagement at Ovid Therapeutics. Recently, however, he's taken a leave of absence from Ovid to focus exclusively on Susannah, his family, and his foundation. Our second guest is Dr. Wendy Chung. Dr. Chung is the Kennedy family professor of pediatrics at Columbia University Medical Center, where she wears a large number of other hats such as the precision medicine resource leader in the Irving Institute, chief of clinical genetics, and many other roles. Wendy received her M.D. degree from Cornell University Medical School and her Ph.D. in genetics from Rockefeller University. She is a renowned genetics scientist and a medical geneticist and is unusual in the breadth of diseases and patients that she's dealt with but has a significant focus on neurological diseases and in particular autism spectrum. She has contributed groundbreaking genetics research and continues to care for many patients. She's won numerous awards and she is consistently recognized as an outstanding educator, and for her commitment to medical and graduate education. Welcome to both of you. It's great to have you. So, Luke, I'm going to begin with you. Why don't you tell us about life before you had Susannah?

Luke

Thanks Stan, and then thanks for having me here. It's a real pleasure. Before Susannah was Nat, our ten-year-old son now, who was three when Susannah was born. So, it was a very, you know, that feeling of being a new parent, and running all over the place, and not being certain if you're doing things right or wrong. I traveled a lot more than I do now and so it was like you said, I was an actor for about 20 years, and we had a very nice, you know, sort of life on the west side of Manhattan. And we had play groups. We were very fortunate to have Nat and of course very fortunate to have Susannah and yeah, that's before. It was kind of kind of a normal, it was a the old normal, we like to call it, because we have a new normal now, but it's the old normal.

Stan

How did you and your wife Sally meet?

Luke

So, we met in college. We both went to school in Connecticut. We met at Connecticut College, and she was dating a good friend of mine, he's not a friend of mine anymore.

Stan

One of those, eh, yeah, yeah. And is she a performer, too? Or doing those other things or?

Luke

Sally is an incredible, incredible actress. She's wonderful, but she doesn't care to admit it herself. Sally writes cookbooks and is incredible in the kitchen. And that's yeah, Sally is a is a wonderful writer and she's since slowed work down a lot. She cares, you know, both of us care for Susannah pretty much full time. But Sally is the is the person who keeps the engine running in our house for sure.

Stan

So now we're going to be talking about topics that are deeply personal and deeply painful, I'm sure. And so, Luke, I very much appreciate your willingness to share all that with us and. And I fully expect to cry somewhere during this interview. And that is exactly as it should be. Our heart should go out to people who are dealing with issues, particularly issues that affect their young children. So, Luke, what happened? You know, how did your life change after Susannah? You've already begun to tell us that, but I'd really be interested in how you learned when you first began to worry that there might be something wrong, what you had to go through to get a diagnosis, what you learned about the diagnosis, and how you how you proceeded from there.

Luke

Yeah, it was a very, I've said this before, and parents know that if parents have more than one child for the first kid, with Nat, I feel like I always say this to you Wendy, but with Nat, if he had a, you know, a little rash, we would Panic and run to the emergency room or something. Not quite, but with our second, with Susannah it was kind of like, oh, she's on her own timeline, you know. She'll get there. But we started noticing, there were a couple of different moments, where we noticed that things were really, really not going in the right direction. And it was Sally actually, in the, had Susannah in the bathtub, and we used to do this thing where we would say to the kids when they were in the bath when they were babies, you know, kick, kick, kick, kick, kick and we made a little song of it. Kick, kick, kick and Susannah just couldn't kick. She couldn't move her legs up like that. Then Sally said, "I think something's really wrong." And so that's when we went to, first, our pediatrician, and then our neurologist, and it became really clear that there was something significantly wrong with her health. The neurologist at the time told us that they were just very stumped. We did a lot of tests. MRIs, EEGs, everything, and the neurologist said that we should get genetic testing. Sally doesn't like me saying this, but if you're over a certain age, and we lived in Harlem at the time, so the kids were born at Columbia and, you know, Colombia is where we went when someone fell and got stitches, but if you're over a certain age when you're having a baby, what do they call it, geriatric pregnancy or something? Part of it is that you have to get a certain amount of genetic testing done. Very minimal, I now know, but so I thought, when the neurologist, I said, “oh, we've already done that,” that I didn't have a science background or don't have science background. I said, "we've already done that, we've had the genetic testing," but they said "no, there's a more extensive way to do it, you have to enroll in this research study." And so, one night we were in the hospital for a couple of nights for something that was going on with Susannah, and someone came in, and we enrolled in the research study. Quite some time went by until we finally were able to hear that she had a mutation in this gene, KIF1A. We didn't know what that meant at all. There was a moment when I was walking with her up Amsterdam Avenue, and she had very low muscle tone as a baby, but her legs shot up like planks and were stiff and she was scared and crying. That was the real moment where I thought something horribly wrong. Sally and I, after we heard the diagnosis KIF1A, we didn't know it. It was accompanied by a few research papers that did exist, and they were very scary papers to have. They said things like early death and brain atrophy and just these Words that were really hard to digest. I got this information before Sally did so I came home and I had this window of time where I knew, but Sally didn't know, and I wanted that window of time to keep going because I knew that what I was just going to have to say to Sally, and tell her what I just found out, and show her these papers, was going to be, and it was, quite literally heartbreaking. Then we started researching. We didn't know anything about this, so we started doing our own research and somebody said, “you need to find Wendy Chung.”

Stan

So, Luke, how old was Susanna when you first began to suspect there was a problem?

Luke

Susannah was three to four months old when we saw something was a little off, but not nothing that was incredibly concerning, until she was about one, one and a half.

Stan

How how long was it before you found your way to understanding that she had this unique mutation in this in this gene you knew nothing about?

Luke

How long was it from when we?

Stan

When you first went to your pediatrician.

Luke

So unfortunately, it took a long time. It took about three months for us to get the results back. If we could do it all over again, I would have asked our doctor at the time to just prescribe or order us the test instead of being part of the research study that we were in. That took us significantly longer than it would have otherwise. Those three months were really difficult because we were just waiting. We didn't know if we were ever going to find out what was wrong with her. Every doctor we went to told us they had no idea really what was going on. They could talk about the specific parts of her body. An ophthalmologist would tell us that her eye was moving one way because of one reason, and then a neurologist would say she's having this activity in her brain for one reason, but then no one could ever give us an exact, “this is what is wrong." That was about three months of not knowing anything and being really scared.

Stan

Three months of terror and then watching your little child go to terrifying things like MRI's and all that.

Luke

Yeah, you know what, someone forgot to tell us when Susannah was getting her first MRI that after she was getting sedated, the babies shake a little bit, and the person who was doing that forgot to tell us that and as Susannah's eyes were closing, I remember thinking, gosh, I don't want the last thing she sees before she goes under to see me crying because I was. Yeah, it was scary, you know, to see that. But it's normal.

Stan

And I'm sure you know now that you actually had a remarkably short period. The average time according to UDN data (the Undiagnosed Disease Network data) for one of these kinds of diseases to be identified and genetically characterized is something like 8 years. And of course, the vast majority of patients are never diagnosed. So, you learned that she had this mutation, and in a gene you didn't know about, and you found some papers that told you that things were bad. So how did your son handle all that?

Luke

He was Four. We're very honest in our family and in our house, but there are certain things that we wanted to protect him from. He's an incredible person, so he handled it in a way that was "this is just my sister, this is how she gets around, this is how she looks, this is how she acts,” but then there were some really scary times where he had to see some things, there still are every month. He sees things that are very difficult that kids shouldn't see but he handles things. He's 10 going on 60, you know.

Stan

How's Susannah doing today?

Luke

Today is a really good day. She doesn't usually make it through an entire day of school. We'll get a call from her aide, her para, who spends a day with her. We'll get a call that she's either had a seizure, or a bad fall or something. Or she's just, you know, not engaging with anybody, and she's staring off, and the teachers can't get her back.

Stan

Yes, she has seizures, and she has muscle control and muscle weakness, problems and attention challenges. Is she having some developmental delays as well as a result of all these problems?

Luke

Oh yeah, absolutely. Susannah, she's now started, she could walk several steps. Just a matter of months ago, but now she started crawling instead of walking, and she spends most of her time in her wheelchair. Yeah, her cognition is certainly not that of a seven-year-old but she smiles a lot, and she's very happy even when we know she's in pain. She's incredible.

Stan

She's beautiful, and you know, that's her life. It shouldn't be, but it is. So, you got to work like parents often do, in trying to understand what was going on with Susannah. And I would have to guess that you felt lost. The vocabulary was basically impossible to understand. As you learned more, you got more and more terrified, huh?

Luke

Absolutely. Yeah, the not knowing, and still right now, the just not knowing what tomorrow is going to be like for Susannah is really hard because she, every day, is kind of an improv, right? If she has one night where she has a nocturnal epilepsy, more than an epilepsy during the day, I mean she has both, but if she has a night that's filled with seizure activity then we know her day is going to be really hard for her, you know? She's very spastic now in her lower limbs. She also has neuropathies in hands and her feet. So, she is in a lot of pain in her hands, and she'll wake up in in her own way. Susannah has words. She can express herself, and in her own way, she'll tell us that it feels like her hands are burning on the inside, and that's hard to have a kid wake up and, yeah.

Stan

So, you meet Wendy, and you have a diagnosis. My guess is that your first question after that was, is there any treatment?

Luke

Yeah well, we met Wendy because somebody had told us that Wendy was the person who could articulate this and tell us what was happening, and maybe care for Susannah. So, I emailed Wendy and she emailed me back within minutes and asked if Sally and I wanted to come into her office and she could explain the implications of what KIF1A was and meant. So, we did, early in the morning, we went to Wendy's office at Columbia. I talked about this a lot; we were mentioning the elevator ride up there. Sally and I were really, really anxious, of course. Suzanna wasn't with us. It was one of the first times that we were in the hospital without Susanna. We had this moment where we were alone and got to spend time together. But that doesn't happen. Somebody has to be with Susannah 24 hours a day. You know, all the time. And when the elevator did open, and I remember this just so clearly, the people who were there, you know, there was a genetic counselor, and a social worker, and the nurse practitioner, and Sally and I knew that we were about to hear something that needed an army of people to help us with. And we went into this smaller room near Wendy's office and Wendy came in and told us. Just very clearly, and with this empathy that is really rare, what to expect and that there's not much known, we could find maybe 15 people in the world that had a mutation in KIF1A, and nobody with Susannah's at the time, and that it was a neurodegenerative disease likely with a progressive course, that she would probably have trouble seeing, which she does, she has vision problems, she would probably be in a in a chair as well. That was a hard conversation. I can't imagine ever having that again. There was a lot of crying, and you mentioned my son, Nat. And this is the moment, of all of this, about Susannah's health, and about this this diagnosis, and when Wendy was telling us what it was, the moment I remember most, and the hardest thing was I said in the middle of the conversation "How are we going to tell Nat? How are we going to tell Susannah's brother about this? How do you tell a kid this?" And Wendy looked at me and said he's going to be a remarkable young man one day. So, for some reason, that's what I remember most about this moment of finding out, this devastating disease that Susanna has was trying to figure out how I was going to explain it to my son. Then Wendy walked us out of the hospital, you know, it wasn't just a "here's a diagnosis, good luck," she walked us downstairs through the building, and even right when we got into the taxi, and there was a hug and an "I'll see you soon." So, we felt like we had this support that I now know not many people have.

Stan

And that's sadly very true. Luke, I wanna move to Wendy, but I do want to ask one last question. n-Lorem, what does it mean to you and your family that Susannah's application was accepted by n-Lorem as a potential for treatment?

Luke

Yeah, that's another moment that I'll never forget is when Wendy was able to tell us that. It means everything, Stan, that people are fighting for a treatment for her, and that it is actually going to hopefully become a reality, and that we might be able to change the course of Susanna's life. It's really hard to watch Susannah's life right now. To think that there could be a treatment for her that would not make it as severe and not make life so hard for her is just beyond measure. Words can't describe it, it's hard to. It means everything to us. Everything.

Stan

And you're sophisticated enough to know that there are no promises of any sort that anybody could make at this stage. So, it's hope, and hope for help, I suppose, is the way to think of it.

Luke

Absolutely. The desperation is teeming through our lives and our house. It's a hope for something that we didn't think would ever be there, and it is, so yeah.

Stan

Yeah, you know, just the hopelessness and isolation of patients that we are trying to help is detrimental to health. Hopelessness is bad for health and helplessness is terrible.

Luke

That's really well said, it is. It is grinding that hopelessness is. We're so thankful.

Stan

Well, we at n-Lorem feel privileged to have Susannah as a patient and to work with Wendy. And, you know, we'll keep our fingers crossed and do our level best and we'll all work together for Susannah. And I would say that I think every patient should experience the care that Wendy displayed, and sadly, I think you're right, Luke, that in far too few times is that the case. So, you are very fortunate in many, many ways. In the midst of, you know, a tragic situation. So, Wendy, I'm gonna turn to you now and we'll try to brighten up the conversation here maybe. So, I know your background, and of course I've admired your work, but I'm intrigued by how you do it. How do you manage every day? Well, talk about a clinic day, I guess. How do you manage seeing so many patients who are in positions similar to Susannah?

Wendy

Number one is I'm the one who's on the screen, but there's an army of people behind me. That's the real magic to all of this. So, we have a caring team. Luke mentioned, you know, our genetic counselors, our nurse practitioners, our social workers. But even beyond that, we've got an army of scientists, computational biologists, bench scientists, neuroscientists, artists, both here and around the world, who are all fighting for Susannah and all of Susannah's friends. So, as we do that, you know, I think we've learned, it's taken me, in some cases, 25 years to figure out some of the things that I do. But as we've done it, we've tried to scale ways to do it and put everyone to work. So even though the team that I mentioned are paid people on my team, the rest of our team is actually our families. Families like Luke, and I want to give Luke a lot of credit. He has mobilized a very powerful community of families behind him, and it's together that we get these things done. As we do this, we all have a role to play, and we all take feedback, and we iterate, and we get better and better as time goes on. We have partners like n-Lorem, which I can't, I mean I'm really good at some of the things that I do, but I can't do what you guys do. Together it's the dynamic, not just duo, but dynamic. I don't know what, a million people, but it's a lot of us all working together. The hope is that, although you know what we're doing for Susannah is important exactly for Susannah, the lessons that we're learning for Susannah, I'm convinced scale beyond Susannah. And so, it's how can we do this? Not just for Susannah, but Susannah's friends in KIF1A, other 500 or so neurogenetic conditions that I think about, the 7000 other rare genetic diseases that we know about, plus the others we're just discovering. I think those are really the incredible opportunities. But it's tough because right now, you know, Susannah is one of very few individuals with exactly her genetic change and we need to target her genetic change in this case, so she really is more than one in a million, you know, probably one in 50 million. But that doesn't mean we leave her behind.

Stan

It doesn't mean that at all. And you know, I think you handled these things as I handle them, and as Luke and parents and patients do. And that is you find ways to take control of what you can control and find ways to accept those things that you can't control. Otherwise, the process is too destructive. I imagine that rings pretty true to you, Luke.

Luke

Yeah, it does. It's pretty hard not to be able to control the outcome of the day, or just what is happening next, and that loss of control and lack of direction, or the spontaneous things that happen in life, are really hard. But like Wendy said, we have this community of people who are just so supportive. And yeah, it's a lot. Wendy has been kind enough to invite us to speak once in a while, every year, to her medical students. This year was pretty awesome because it fell on a voting day, so the kids didn't have school. So, Nat and Susannah came too, and I never want Nat to know what the word degenerative means. Although he's getting old enough to see that things are changing with his sister, but I had told Susannah I wanted to prep her for what was happening and I said that there'll be a microphone that one of us will have. Wendy pushed Susanna's wheelchair in, and then Susanna came and took the microphone. She sang a part of the song from Frozen, and it was in front of, you know, I don't know how many students were in there, but it seemed like a lot.

Wendy

An auditorium full of 200 students all watching Susannah perform.

Stan

Well, she's from a family of performers, so naturally she's going to be a star.

Luke

Yeah, yes she is. But seeing her do that is not something, and I never sell her short, I know she can do anything she wants to do, but there are things where, you know, you think about "am I ever going to get to see my daughter do..." And as I was sitting there watching her, even though we were talking to these medical school students, somebody said "Did you take a picture, did you record it?" I said "no, I just lived it." I got to see my daughter sing a song to a big audience. That was another great gift, so thank you Wendy.

Stan

Wendy, why don't you tell us about the gene KIF1A, and what it does, what the protein does, and why Susannah is sick.

Wendy

So, Susannah has a genetic variant. It started brand new with her. It wasn't inherited from either Luke or his wife, Sally. And the gene itself is a kinesin. It's a molecular motor, so it's responsible for taking things down these very long axons, or these very long projections out of neurons that go all the way from her head and the neurons in her head, but also the ones that go through her spinal cord and go down her legs to her toes and out to her fingertips. Those very long, if you think about it, the length you have to go from your head all the way down to your big toe, it's a long way to go. And so those motors are responsible for going a long, long way. And with this, Susannah's motor doesn't work very well. It has trouble in terms of, if you think about a train, sometimes it stalls on the tracks. It gets stuck and there's a traffic jam. Sometimes it falls off the tracks and, you know, it sort of loses its way in that way. And without being able to deliver the cargo of those trains, those cells die. They die over time, and once they're gone, I can't say for 100% sure, but I'm 99% sure they're not coming back. And so, as Luke described, this is unfortunately a one-way street. Luke and the families have a very obvious hashtag when you think about it. Hashtag stop the clock because that's what we think about in terms of time is ticking and once we start, you know, losing some of those cells, they don't come back. Susanna has this neuropathy, or this painful feeling that she has this burning. Susanna describes it, and some of the older patients have described it for me as well. It's a burning sensation that really hurts. I mean, like, deeply hurts in terms of your hands and your feet and just feeling tight, feeling like you can't really relax or let go. And with that, it's just a painful, uncomfortable feeling and we try medications, we try some of the same medications we use for other patients who have what we call neuropathies or these problems with their nerve cells. But it never really completely goes away, and at times it gets worse than other times. So that's part of it. But we literally, and I know this is hard for you to hear, Luke, so I'm sorry about this, but we literally see Susannah losing cells over time. We can image her brain, that MRI that Luke described, we can look at her brain. We can see parts of her brain that are shrinking, where we're losing those cells. They're dying off. We can look in her eyes. We can see her optic nerve going to her eye. We can see those cells dying off.

Stan

Is she losing sight then?

Wendy

Yes, she's losing sight, and it's so hard for Susannah. I mean, she really is my heroine. But it's so hard for her because she's got so many things battling against her. She has trouble with this plasticity, being able to move, but she wants to be a kid, right? She wants to just play and have fun. And you can imagine sometimes because she's not seeing things out of this, you know, corners of her eyes are seeing them clearly enough. Plus, she has trouble in terms of moving, and she has trouble with her balance. She falls a lot more than other kids do, and sometimes when she falls, she doesn't know what she's going to fall into. And so, it's just hard. Susannah's broken bones, more bones than her collarbone, her arm. And you know, when she breaks a bone, it's so hard for her, because then she's immobilized and it makes it that much harder for her to get back to where she was. And so, she has these, you know, it's sort of declining, but then, you know, it's almost like a big step down. And she's got to work, work, work to just try and get anywhere close to where she was, but it's hard. Within that, there's also been this other major major problem, which is that in terms of in her head, she's just got an electrical storm. The seizures, in other words, that are going on. Again I hate doing this to you, Luke, but this is no joke. We have just an incredible community, but we've lost so many of our children to this disease. And we've lost them due to some of these seizures in some cases unexpectedly. And I know that's what scares us to death. And I know Luke and Sally are just, if you can imagine, I don't think they've gotten a good night's sleep, you know, since Susanna was probably, I don't know, two and a half, three years old, because we've had kids that we've lost to seizures in the middle of the night. So literally Luke and Sally, one of them is sleeping with Susanna. Some cases Luke mentioned Nat, and Nat’s had, on occasion, to be the big brother, in terms of sort of being the angel watching over Susannah as she's sleeping and in some cases having a seizure, or throwing up, or having something caught and having to literally be able to make sure that she doesn't choke on her own vomit, or that she doesn't have a seizure, that she doesn't wake up from. It scares us because we've had kids that we've lost that way. As Susannah goes through this, as Luke said, we've tried just any number of medications for Susannah's seizures, and she's on a cocktail of medications. It's not just one, but it's a cocktail and it changes. It's not like you get it and you find the right mixture and you're good to go. It's like you might be good to go for a few months and then it's all over again trying to figure out what this is and making it so that she's not a zombie because, you know, there are some of these medications that are sedating and then she's just out of it. And that's not a life, you know. You want her to be a kid, but you also don't want to lose her or have her so that she's having so many, we call them postictal periods, times after a big seizure that she's just out of it for that reason. So, it's just, I can't tell you, it's a hard life. Susannah has many, I think of them as, angels watching over her. Besides her family, she's got her service dog, Pippin, who's just amazing. I mean, Pippin is a great for, I think, a great therapy dog for Susannah in general, but she's got her aid, she's got teachers, she's got therapists. I mean, it literally takes an army. But even still, there is a team hovering over Susannah 24/7. I kid you not. It really is 24/7 to be able to keep her safe. I have to say though, on the other hand, Susannah is just a joy. I mean, it is a joy to see these things, when she gets to eat an ice cream Sunday, or celebrate her birthday, or be able to go ice skating, but it's Susannah's form of ice skating at this time in terms of being able to go to the beach and just enjoy looking for shells or playing hockey or doing a car wash. I feel like I'm, even though I don't get to spend as much time with Susannah under COVID, I get so many pictures and videos that I get to be a part of the extended family, that it's just, it's that joy that motivates me and my team to do what we do every day because it's allowing kids like Susannah to live their richest, fullest lives, and we want to make them actually have those lives. That's the painful part. Every time we see her take a step back.

Stan

Why don't you spend just a minute on how we hope to treat Susannah, and why you think it might work?

Wendy

So, as I was talking about Susannah and these trains going down the train tracks, Susannah has one version of the gene that's perfectly fine. Those trains, I think, run on time they run on schedule, but she's got another copy of the train that is clogging things up. It's making a traffic jam. It's causing problems, and so essentially what we need to do, ironically, is get rid of one of her genes and be able to get that train out of the way, let the traffic flow and be able to maintain the health of those nerve cells. That's a tough order. We don't have, you know, a medication. I can't just say, you know, take two aspirin and call me in the morning. I don't even have something like the seizure medications. There's nothing we know of currently, in terms of just a regular pill that you can take, for Susannah to either treat the symptoms effectively, or to get at the root cause of the disease, and that we know that if we don't do something to get at the root cause she will continue to go downhill. With this, the sort of, just to put it in simplistic terms, the ASO that n-Lorem is developing is going to be able to suppress or to be able to prevent that bad KIF1A from being made, or at least decrease it to such a low level that it's not going to cause the mischief that it's causing. It's a tall order. And I want to emphasize this is not a trivial thing to do because we have to get rid of the bad KIF1A while maintaining the good KIF1A because we need those trains to still keep going down the train track. We've got a team of people at n-Lorem, and I do have to say a lot of my confidence is built in my history with dealing with another rare disease community called spinal muscular atrophy. And so, a lot of the playbook that we've taken for KIF1A is built on the antecedent about 12 years that we had experience with SMA. And that same strategy worked in terms of, Stan you take a lot of credit for this one, because for this, with SMA, this was our first FDA approved treatment for SMA, and I can't tell you how many children I buried with SMA before we had the ASO treatment that we had with that. And so it was that inspiration that really convinced me that we had a chance with KIF1A and I don't say it's a guarantee, but we've got a chance in terms of going forward and it's with that that I really do think the strategy should work, scientifically it makes sense. Stan and all of the other scientists at n-Lorem have just an incredible insight to the chemistry of how this works and to be able to design what are very, very specific molecules to be able to do this without being toxic, without doing any damage, and to fine tune this in just the exact way that we need. And they've been so incredibly generous. To literally donate all of that incredible experience and knowledge in the direction of children like Susannah. And there's, I can tell you, I know a lot of things. I don't know this. I don't know this chemistry. This isn't something that any scientist, like any one of us, could do just by thinking hard enough or, you know, sort of working enough at it. It really takes the decades of experience that Stan and his team have, and it's us working together with kif1a.org with the other families in terms of all making this work. And I'm convinced we're on the right road.

Stan

I am too and being involved with Spinraza, our drug for spinal muscular atrophy, was one of the many high points in my long career. But what we're doing at n-Lorem is even more gratifying to those of us who are doing it. And so, you know, the one thing that we have going for us with these nano rare diseases is they're all genetic. And so, once we understand the mutation, we have a potential target and now with ASOs we have genetic medicines. So, we can take that information directly and translate it to a medicine that we can use to treat a patient like Susannah. And, you know, to try to do that in 12 to 15 months, start to finish, is a real challenge. Especially in these more complex mutations where we have to be very specific for one part of the two-part gene system. So, it's challenging, but I think the technology is up to it. I also believe that it has to be scalable and if we make a commitment to a patient, we have to be ready to treat that patient for life, and we are. And we can do that. We can do it for free, and we are. So again, Luke and Wendy, it’s just a great privilege to know both of you. And I want to thank you for sharing all this with us. Before we close Luke, any final comment that you want to make?

Luke

Yeah, I you know, I just thought of, about three years ago, I took this video of our son Nat. Susannah was in her chair and Nat was teaching Susannah how to throw a coin into water to make a wish, and he explained it. You know, you have to throw it this way and then you make a wish, but don't tell anybody what that wish is. She threw it and it got in there and I emailed that video to Wendy. I said look at this, Nat's, you know, teaching Susannah, how to make a wish. And Wendy just wrote back one sentence. And that sentence was, I know what my wish is. And it's my wish too. And Stan, and your whole team, I think you know you're working so hard to make our wish come true. So, thank you.

Stan

Wendy, any final comment?

Wendy

Now, just to say that it's not easy. I know it's not easy for Luke and Susannah and Sally, and that every single day, we're there behind you. There are different things that we do to support Susannah. And together, we're a really powerful team and I know we're going to be able to get to something better.

Stan

Well, let me, I can't think of a better way to close this, we are all in pursuit of something better. Children should not suffer and so we'll do our best. So, I want to thank both of you very much. It's been a wonderful privilege to have you share all this with us and I'm sure we'll have more opportunity to talk in the future and I look forward to them being much more positive.

Wendy

So do I.

Kira

Does genetics fascinate you? Of course It does, your listening to this podcast. Discover new advances in the world of genetics with DNA Today podcast. I'm Kira Dineen. I'm one of the producers of the n-Lorem Patient Engagement Program podcast, and I'm also the host and producer of DNA Today podcast. I use my expertise in genetics to help you understand the complexities through interviews with leaders in the field, including the host of this podcast, Dr. Stan Crooke. He was on episode 141. DNA today explores genetic technologies like home DNA kits, CRISPR, groundbreaking research, and rare diseases. For a decade, DNA Today has brought you the voices of genetic pioneers. There's over 185 episodes, so plenty to keep you entertained and updated about genetic news. The show is a fan favorite, winning the People's Choice, Best Science and Medicine Podcasts award for the past two years. Learn more at dnapodcast.com and subscribe, rate, and review on Spotify, Apple Podcast, Google Podcasts, or wherever you're listening to this podcast. Discover new advances in the world of genetics with DNA Today podcast.

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 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 nlorem.org. Any questions can be sent into podcast@nlorem.org. Search n-Lorem on Twitter, Instagram, YouTube, LinkedIn, and Facebook to connect with us. Please rate and review the podcast on Apple, Spotify, or wherever you listen. This truly helps us climb the charts and allows others to find the show. This podcast is hosted by Dr. Stan Crooke, our videographer is Jon Magnuson of Mighty One Productions. Our producers are John Magnusson and Kira Deneen of DNA today. Thank you for listening.