Seizing Life, episode 31
“The Best of Seizing Life”
Transcript

Voiceover:	00:10	This week on Seizing Life, we revisit some of our favorite episodes from the past year. We begin with an excerpt from our conversation with Cure's founder, Susan Axelrod.
Kelly Cervantes:	00:20	It's no wonder that you started Cure, but you are the mother of three children. You have a special needs child, your husband is traveling a ton. This is no small feat in the midst of all of this. Where was it born from?
Susan Axelrod:	00:38	It was born from the desperation that we all feel, those of us whose kids have not responded to medications and whose lives are being just destroyed, if not lost. And it came from slowly, again, talking about my age and how long ago this was, but slowly meeting other moms. The power of moms. One of the physicians that was on our board when we first started, used to say, "There's nothing like an angry mom."
Susan Axelrod:	01:09	And I used to say, "We're not angry. We are desperate. To me, there's a difference.
Kelly Cervantes:	01:16	There's a big difference.
Susan Axelrod:	01:18	I started to meet these other moms and we started to do some fundraising for support groups and things like that. And then really started talking and learning. I thought Lauren was the only one out there who didn't respond to medications. I honestly did. I thought epilepsy's certainly been cured and solved by now and I just have this fluky child.
Susan Axelrod:	01:44	Fast forward to meeting some moms and I'm like, no, there's a lot of us. There's a lot of us. It's a spectrum. There's some who lose their lives. There's some who can function fairly well. There's some, Lauren's sort of maybe in the middle there. So the impact is very variable, but we're all struggling and we all want more for our kids.
Voiceover:	02:15	Dr. Charles Marcucilli, division chief of pediatric neurology and director of pediatric epilepsy at Rush University Medical Center spoke with us about understanding the complicated nature of epilepsy.
Dr. Marcucilli:	02:27	We've struggled with defining epilepsy almost from the earliest observations. I think the reason why it's hard for people to understand relates to the wide range of severity, the widely different types of seizures that patients can experience, in

		terms of the causes as well. The fact that epilepsy is associated with so many comorbid conditions.
Kelly Cervantes:	03:03	Define comorbid.
Dr. Marcucilli:	03:04	So these are other disorders or conditions besides the epilepsy such as attention deficit disorder or anxiety or depression. And finally I think there is variability in terms of the impact of epilepsy on the individual in the family.
Kelly Cervantes:	03:25	Let's start with the severity that you mentioned first.
Dr. Marcucilli:	03:29	I like to use as an example of [absence 00:03:32] epilepsy or staring spells. We like to say that epilepsy is a common phenotype. That means what the condition or disorder looks like with multiple genotypes, meaning different causes. So I could have within my clinic three different patients with staring spells.
Dr. Marcucilli:	03:51	I could have a child who has 20 to 30 staring spells a day lasting 20 seconds each, and this child, hypothetically speaking, developed that epilepsy at about three to four years of age. And so this child probably has childhood absence epilepsy. Contrast this with another child who also has 20 to 30 staring spells per day brief, but in contrast to the first kid, this child has a generalized tonic clonic seizures and developed the seizure much later, nine, 10 years of age.
Dr. Marcucilli:	04:32	In the first case, that child will probably outgrow that epilepsy by the time she's a teenager and has gone through puberty. In the second case, that child has a lifelong condition that she'll have to deal with. A third case is a child who comes with a staring spell, perhaps developed the epilepsy at nine years of age, and this child will have longer staring spells and perhaps they can progress to generalized tonic clonic seizures.
Dr. Marcucilli:	05:08	The first two kids probably have a genetic cause and the third kid may have a genetic cause, but could also have other causes such as malformations of cortical development. What the public sees is the staring spells, but each have a different trajectory. The first one will likely outgrow it. The second one will have to deal with this through adulthood and the third, it may be uncertain and I think that leads me to the severity question.
Dr. Marcucilli:	05:39	I've taken care in the past of a patient, a young man, I won't give too many details here who essentially had one seizure per year. He'd have a seizure, come into clinic, I'd make a

		medication adjustment. I would then see him in six months. He's doing great. I'm patting myself on the back and then six months later he has another seizure, make another medication adjustment. Six months later he's doing well and I think I'm doing well. Six months later he has another seizure. After about three years, I was beginning to realize I wasn't really helping him much and that his pattern of seizures was once a year. And so I frequently ask the residents or my EEG technicians, could you live with or tolerate one one-minute generalized tonic seizure per year?
Dr. Marcucilli:	06:36	That's not really a fair question, right? Because they help me with my patients who have epilepsy and they put on EEG leads and the vast majority of them would say, "Dr. Marcucilli, I think I could do one seizure a year. Doesn't seem so severe." But what if I said this young man, and let's just say he's 22, for six months out of the year in most states, he can't drive. Now he can't go to school. He's going to have a difficult time getting or holding a job and this may affect his relationships. And suddenly one seizure a year isn't so benign. So we have this whole range of comorbidities that we need to consider. It's not just the seizures, but it's also what impact the seizure has on one's life.
Voiceover:	07:40	We spoke with DePaul University student Eva Wadzinski and her mother, Michelle, about the challenges of being a college student with epilepsy.
Eva Wadzinski:	07:47	I started in a community college because I didn't think I was at the health to go to a four year university and I also went through two brain surgeries in the process. And then I started DePaul, I started with four classes. It was not working out well, so I went down to three and it's been better.
Eva Wadzinski:	08:07	You kind of just have to push forward every day and you end up having to educate your professors, your classmates, your student of accommodations because no one seems to know what epilepsy is.
Kelly Cervantes:	08:20	That like leads perfectly into my next question is, what do you tell your professors and how do they respond?
Eva Wadzinski:	08:29	It differs.
Michelle Wadzinski:	08:30	We have a letter that we've been giving to every teacher since, I don't know, freshman year and we've modified it through, the same letter go in and make the modifications, print it out, give it to them, trust that they'll actually read it.

Eva Wadzinski:	08:45	And then I'll have one professor that's like, "Well you were gone because ...", but I was gone because of seizures, but they think I just skipped class. And so then I have to go my doctor, to my psychologist, to my student of accommodations to get everyone in to say like, no, this is not because I skipped classes because it's unsafe for me to be in class.
Eva Wadzinski:	09:05	And so that's tough when that happened.
Kelly Cervantes:	09:08	And it's creating more stress for you in an already stressful environment. I mean college is hard. and so that additional work has to be tough.
Kelly Cervantes:	09:23	What do you tell your peers, your fellow students?
Eva Wadzinski:	09:27	I don't usually tell them because it's more of a personal thing and especially if I'm not close to them or they're friends of mine, then it's like I'm just going to [inaudible 00:09:37]. But I've had seizures during classes and then it makes the professor more aware because I don't have tonic clonics unless I'm off of my medications. I have all these other ones that may not be apparent, but I had a seizure in my math class where my arm convulsed and it actually hit one of them girls and she was like, "Are you okay?" Like they thought I was probably on like some drug.
Michelle Wadzinski:	10:05	Which is just so frustrating, but that's where the thought process, because people are so uneducated about seizures.
Kelly Cervantes:	10:12	People fear what they don't understand. You know, if you take the time to learn and understand, I think then they're more supportive. If you think of it before epilepsy touched your life, how much did you know about epilepsy?
Michelle Wadzinski:	10:26	I knew the tonic [crosstalk 00:10:29] Gray's Anatomy.
Kelly Cervantes:	10:30	Exactly, exactly.
Kelly Cervantes:	10:32	And me too, I didn't know anything about it. And so I try to remind myself, unless there's a family member or a friend that has it, it's they don't understand. Yeah. And so first we try to go gently with education and hopefully that resolves everything.
Voiceover:	10:55	Dr. John Millichap, attending physician for Neurology and Epilepsy at Ann and Robert H. Lurie Children's Hospital discuss the benefits of genetic testing for epilepsy patients and their families.

Kelly Cervantes:	11:05	What is the connection between genetics and epilepsy?
Dr. Millichap:	11:08	Sure. So we know a lot about the causes of epilepsy and in the same breath I'll say we know very little. So we know of hundreds of genes that are related to epilepsy and having a variation in the genetic code for those specific genes alters the function of the machinery in the body that that gene codes for. So if there's a variation in that piece of machinery, in that channel, then the balance can be off. So where you have more electricity in that brain cell and more electricity then leads to seizures. So that's just one example. There can be other genes that are related to the actual structure of the brain, so they can lead to actual malformations of the brain that we can see on a picture like in an MRI.
Kelly Cervantes:	12:00	So talking about variants in the genetic code, not all epilepsies are necessarily inherited. You get your test results back and they're talking about this variant or that variant. What causes those variants if it's not hereditary?
Dr. Millichap:	12:18	Sure. This is one of the first things that I talked to parents about when I'm leading into talking about doing genetic testing or recommending genetic testing, is that most of the early onset childhood epilepsies are actually new in the child and not from the parents. So sort of back to basic biology after the egg and the sperm come together, then it forms the child and the DNA has to be copied every time the cell splits. And in a normal, in everybody, you, me, every time that that's copied, there's going to be a little mistakes, little changes to the DNA and sometimes those changes don't make any difference whatsoever and we just go on being ourselves.
Dr. Millichap:	13:03	And then sometimes early on one little change will be then important. It will change the function of that one gene, of that one piece of machinery that it can cause epilepsy and be in all subsequent cells in the body.
Kelly Cervantes:	13:18	What are the tests that are Evailable? What are the tests that you order as a clinician and what are you looking for in those tests?
Dr. Millichap:	13:26	Sure. I got into neurology about 11 years ago and at that point genetic testing was not used as a first line test whatsoever. Things have changed to the point where almost every patient that I see today, once I've established the diagnosis of epilepsy, taken a history and looked at the EEG, the MRI, my examination and I haven't determined a cause of the epilepsy, I'll talk to parents about the Evailability of genetic tests. And in many

		cases, the first test would be a focus test that would look at genes we know to be associated with epilepsy.
Dr. Millichap:	14:08	And this test is called an epilepsy gene panel and it has a high sensitivity for those specific genes that we know to be related to epilepsy. So maybe a hundred or 200 genes that have a strong association with epilepsy are included on the panel. And once we've done this test, we know that there's not a change in those genes because it looks at it with such depth and accuracy. And if that's unrevealing, we can go to the next step, which often involves the parents even more where we'll take the child's blood and then also the blood of the parents and look at all of the portions of the DNA that make machinery making make the body. That's called whole exome sequencing. And the reason we take the parents' blood is because as I said, we all have little variations that don't change anything in our bodies or have any significance. And so we kind of subtract any of those inherited variations from the child.
Kelly Cervantes:	15:18	Right. So if you see the variant in the parent and then you're also seeing the variant in the child, but the parent is symptom-free, then that's probably not your culprit.
Dr. Millichap:	15:27	Yeah. So the genetic testing company has a huge computer that does a lot of this initial sifting. And another thing that I'll constantly be reminding other doctors and trainees and parents is that the genetic testing that we do is not a static test. So if you had an MRI last week and the radiologist read it and the neurologist looked at it, that happened, that is what the MRI shows. But if you had a genetic test last month or six months ago or a year ago, that test is not static, we're always learning new things. The variations that were meaningless or the genes that were meaningless to epilepsy a year ago may today be the cause for your child's epilepsy.
Eva Wadzinski:	16:20	Dr. Takijah Heard, division head of Pediatric Neurology and Epilepsy at the North Shore Neurological Institute explained how the vagal nerve stimulator commonly known as VNS can provide seizure relief for some epilepsy patients.
Kelly Cervantes:	16:33	So you're here to talk to us today about vagal nerve stimulators, the VNS therapy, as a treatment for seizures. Can you explain the device to us and how it works?
Dr. Heard:	16:47	The vagal nerve stimulator happens to be a noninvasive sort of procedure that can be done as an outpatient. It's a non-open brain surgery. It happens to have a coil that sort of wraps around the vagus nerve and then they have a generator they

		sort of place on the left side in the, maybe a couple of inches below the clavicle.
Kelly Cervantes:	17:05	Okay.
Dr. Heard:	17:06	And so the device goes just underneath the skin. And then there are wires that come up and connect to the vagus nerve, which happens to be in the neck.
Kelly Cervantes:	17:16	Okay. And why does it work?
Dr. Heard:	17:19	So it ends up targeting the vagus nerve. The vagus nerves then sort of connects to the brain and sort of sends amounts of energy that happens to be treatment amounts of energy to sort of combat the seizure, which the addition of a [inaudible 00:17:31] amount of energy, to sort of stop the seizures.
Kelly Cervantes:	17:34	And how does it know when to work?
Dr. Heard:	17:37	And so it has great configurations in which it sort of has a timeframe at which it comes on. And then it has a timeframe in which it gives the amount of energy. So it can be 30 seconds, it could be up to a minute. And then we go up on the amount of energy and milliamps until we get to a therapeutic amount. Therapeutic is usually about 1.5 milliamps. And then that's whenever we start to notice that as it cycles, every five minutes, that you'll start to notice that the seizure frequency you will start to go down.
Susan Axelrod:	18:05	Ah, I see. So it is triggering the vagus nerve at whatever every five minutes or I'm sure whatever you set that to be at.
Dr. Heard:	18:15	Yep.
Susan Axelrod:	18:15	Now is it the kind of thing where if someone feels a seizure coming on that they can trigger it to try and stop the seizure?
Dr. Heard:	18:24	Great question. So they have a device, what's called a magnet. It looks very similar to an Apple watch. So it's a large device that you can put on like an Apple watch or you can wear it around your neck and you can trigger it by swiping it. So I usually will say put a W across the actual stimulator, that activates it, sending an extra jolt of energy that's usually about 0.5 milliamps larger than the normal routine amount.
Kelly Cervantes:	18:48	Okay. And then how in the world did they figure out that this worked? Do you have any idea?

Dr. Heard:	18:53	No, not really, but we definitely noticed that I'm the vagus nerve is one of the cranial nerves. And so this is one of the ones that happens to come out of the brain and we have easy access to. So is there a way in which we can sort of get to the brain without it actually being a open brain surgery?
Kelly Cervantes:	19:08	How is the VNS powered?
Dr. Heard:	19:12	So it has a battery. The battery life can last anywhere from five to 10 years. Usually about seven to eight years is usually the battery life for most. Obviously it could be shorter if we're sort of are doing the times in which the cycling happens much more frequently than five minutes and can last up to 10 years.
Kelly Cervantes:	19:27	And so what happens when the battery runs out? Because this is under your skin. It's not like you know the remote control where you can just pop new batteries in. There's a little more invasive.
Dr. Heard:	19:36	Well, not so much. So there is going to be the vagal nerve stimulator generator's going to be under the skin and so they make a small incision where it's at. It's sort of placed in a pocket. They remove the generator from out of that pocket and slide in a new generator. So again, it's a daytime procedure so it's not anything you have to be in the hospital for. You get it done, you put in a new battery, then you get to go home.
Voiceover:	19:59	Susan Axelrod concluded her conversation with us by discussing her hopes for the future of Cure and the need for investment and more research.
Kelly Cervantes:	20:07	Where do you want to see Cure go? Where do you want to see the epilepsy community push forward? Where do you want us to be 20 years from now?
Susan Axelrod:	20:18	I think that we are on the cusp, maybe another five to 10 years of being able, as we know, epilepsy is many, many diseases. So we can't say we're going to cure it all, but let's start tackling them. And that's what I'd like to see happening. And that's one of the reasons we've sort of approached infantile spasms or [inaudible 00:20:39] is let's zero in on this or prevention of epilepsy after head injury. This is a known risk factor, these are known things. Let's get smart here and sort of figure out how we can tackle this, break it into pieces for it happens.
Kelly Cervantes:	20:55	Stop it before it happens.

Susan Axelrod:	20:55	Right.
Kelly Cervantes:	20:56	Or know exactly what treatment is going to work for that specific epilepsy.
Susan Axelrod:	21:00	Exactly. Exactly. More funds are desperately needed and I don't want anybody to think that that's not the case. However, I do want to make sure that it's focused. I mean I think our impact has been really significant and there are other people and hopefully pharmaceutical companies, et cetera that will be interested in development of therapies. But if we can lay the ground work for them and be really smart about what we're doing. And I just want to get to the point, on my way over here this morning or actually before I left this morning to come here, I opened up my email and there was another email from somebody asking, baby just newly diagnosed and as it was hard for me to walk over and see you in [inaudible 00:21:55] that very first time I just, my heart breaks. It's an infant, it just shouldn't be happening. So I want to see that number greatly diminished.
Voiceover:	22:12	Cure is the leading non-governmental funder of epilepsy research. Since its creation in 1998, Cure has raised more than $60 million to fund over 230 projects in 15 countries. Please consider supporting Cure's mission to end epilepsy by making a donation today at cureepilepsy.org/donate. Thank you.
Voiceover:	22:37	The opinions expressed in this podcast do not necessarily reflect the views of Cure. The information contained herein is provided for general information only and does not offer medical advice or recommendations. Individuals should not rely on this information as a substitute for consultations with qualified healthcare professionals who are familiar with individual medical conditions and needs. Cure strongly recommends that care and treatment decisions related to epilepsy and any other medical condition be made in consultation with a patient's physician or other qualified healthcare professionals who are familiar with the individual's specific health situation.