

Seizing Life, episode 80
Infantile Spasms – Part 2: The Hope of Research
Dr. Renée Shellhaas, Kari Rosbeck, Beth Dean
(Transcript)

- Kelly Cervantes: 00:00 Hi, I'm Kelly Cervantes, and this is Seizing Life, a biweekly podcast produced by CURE Epilepsy.
- Kelly Cervantes: 00:18 This week, we continue our panel conversation about infantile spasms by focusing on research currently being conducted on IS. Our panel includes Dr. Renee Shellhaas, a pediatric neurologist and director of research at C.S. Mott Children's Hospital at the University of Michigan, Kari Rosbeck, the president and CEO of the TSC Alliance and one of the founders of the Infantile Spasms Awareness Network, and Beth Dean, the CEO of CURE Epilepsy.
- Kelly Cervantes: 00:47 So Kari, obviously here at CURE Epilepsy, we love our research, and we do understand that there are some exciting new trials going on. Can you tell us about them?
- Kari Rosbeck: 00:56 I sure can, but let's go back to 2012 because that's when a core group of five TSC centers of excellence joined together to actually do the first biomarker study for epilepsy and TSC. That work was led by Dr. Martina Bebin at the University of Alabama, Birmingham, with support from the National Institutes of Health. And what they actually found was in newborns with TSC, there was a change in the EEG before the onset of seizures. So that biomarker allowed us to plan the first preventative clinical trial for epilepsy in the United States called the PREVeNT trial, again with funding from the National Institutes of Health, NINDS, National Institute of Neurological Disorders and Stroke, and with support from Lundbeck who provided drug, provided vigabatrin or the study. That recruited 86 babies, and we are waiting for the data to come out early next year. We're super excited and super hopeful.
- Kari Rosbeck: 02:02 We also have started a trial recently utilizing sirolimus, or an mTOR inhibitor, the same way, preventing epilepsy from ever developing in infants with TSC. And because sirolimus treats the underlying genetic pathway of TSC, we're very interested to see if there will be difference in outcomes between PREVeNT and what we're calling the STEPS trial. And the STEPS trial is being led by Cincinnati Children's Hospital and the center of excellence there.
- Kelly Cervantes: 02:34 Clinical trial is, I think, words to so many infantile spasm parent and caregivers' ears. Now, I know that CURE Epilepsy has been sort of on the forefront of some of this infantile spasms research, sort of this basic science. Beth, as the CEO of CURE

Epilepsy, can you talk to us about CURE's infantile spasms initiative and what the aims of that research were and what discoveries were made?

- Beth Dean: 03:03 Sure. So let me back up and start with our overall funding. So CURE Epilepsy has funded over \$4 million since 2011 towards infantile spasms. So this is a key area of research for us as an organization. But back in 2013, we actually initiated an infantile spasms team science program. So it was a specific initiative using eight different researchers to work on infantile spasms, but collaborate and share information as they did this. So this team science initiative for infantile spasms ran for several years, and it was really designed to accomplish a couple things. First, it was designed to look at and try and better understand the basic mechanisms of in infantile spasms, so what was going on or what is going on in the brains of these infants that's causing the spasms. And then from there, we wanted to identify biomarkers, identify novel drug targets-
- Kelly Cervantes: 04:07 Beth, I'm going to stop you for just a second there. Can you explain to us what a biomarker is?
- Beth Dean: 04:12 Sure. So biomarkers are ways using information from the body, so whether it's from blood or an EEG signature or genetics, but using some information from a person to try and determine if they're likely to, or maybe predisposed to, to get infantile spasms or get another disease. So we look for biomarkers as kind of signals or flags to tell us that somebody is more likely to get a disease than someone else without. So for infantile spasms, right, we wanted to try and find something that would let us know if a baby is maybe predisposed or likely to get infantile spasms.
- Kelly Cervantes: 05:01 All right, Beth. I know that you were mid answer, so I'm going to let you continue here.
- Beth Dean: 05:06 Sure. So, like I said, there were eight teams that were working as part of this infantile spasms initiative, and as a result of the work and the science that they conducted, the research, they were able to publish 19 journal articles. So these are kind of summaries of their research that appear in scientific and medical journals, kind of detailing the findings and the learnings they had. There are also seven additional manuscripts that are actually being drafted now, so those will become publications. Three of the researchers went on to get additional grants from the National Institutes of Health to continue their work in infantile spasms. And one investigator even got an additional... or got a patent for the work that they did.

Beth Dean: 05:48 So there were a lot of learnings that came out of this, and what's important is that these learnings, while none of them were a cure for infantile spasms, the results were published, the learnings are shared with the community so that other researchers can continue to build upon that research and move us towards a cure. So this is one of those, it takes a village, but lots of people are doing work and trying to understand the brain and the mechanisms. And by sharing this information through publications, we hope that others take these learnings and pick up the ball and take it a couple more yards down the field.

Kelly Cervantes: 06:27 So it sounds like there is a lot of exciting research that has recently been done and is currently being done. What other new trials or treatments should parents and caregivers be on the lookout for?

Beth Dean: 06:41 So obviously, we continue to seek more therapies and treatments for infantile spasms and improved treatments and therapies. There are clinical trials going on as we speak, some of them looking at drugs that are already approved in the marketplace and seeing whether or not they will work in infantile spasms, and then new compounds in development, which are in early clinical trials and being looked at. So there is definitely hope on the horizon. And this is why research is so important to continue to discover... to understand mechanisms, to discover a targets and treatments that will work on those targets, and then get those into clinical trials.

Brandon: 07:25 Hi, this is Brandon from CURE Epilepsy. Did you know that one in 26 Americans will develop epilepsy in their lifetime? For more than 20 years, CURE Epilepsy has funded cutting-edge, patient-focused research. Learn more about our mission to end epilepsy cureepilepsy.org. Now back to Seizing Life.

Kelly Cervantes: 07:45 All right. So we have thrown a lot of information out there. Imagine a new parent, they are just hearing about IS for the very first time. What is the most important pieces of information that you hope they take from this podcast?

Dr. Renée Shellhaas: 08:01 The very first thing is for families and parents, especially of babies who are at high risk, to know that their baby's at high risk, to know that infantile spasms are an emergency, to look for them, but not live every day in fear of the infantile spasms. If you think or you're worried that your baby might have in infantile spasms, call your doctor right away. If it's possible to get a video on your phone, that can be very, very helpful, just for your doctor to get a picture of what's going on with your baby. And then just know that although it's scary and although

we don't have all the answers, there is absolutely hope. And we do have some treatments that work well. And we're here and ready to work with any family that comes forward, whose baby has a new diagnosis of infantile spasms.

- Kari Rosbeck: 08:58 I just say, as a patient advocacy organization, we are here to walk this journey with you. You absolutely are not alone, and there are parents to support you, the TSC Alliance is here to support you, and many other advocacy organizations including CURE that are working so hard to find the answers to infantile spasms, and to one day, prevent them from ever happening.
- Beth Dean: 09:26 Let me add one more thing. You, as a parent, are the child's biggest advocate, so don't take no for an answer. If you need to get into your doctor and they can't see you right away, go to the emergency room. But as Ben said earlier in this program, it's an emergency, so push as hard as you need to, but get your baby taken care of.
- Kelly Cervantes: 09:49 Kari, Dr. Shellhaas, Beth, thank you all so, so much for joining me on this panel discussion episode of seizing life. Your experience in knowledge is going to help so many families who are walking this unfortunate road of infantile spasms. I just appreciate all of you and your time. Thank you.
- Dr. Renée Shellhaas: 10:11 Thank you.
- Kari Rosbeck: 10:12 Thank you.
- Kelly Cervantes: 10:16 Thank you, Dr. Shellhaas, Kari, and Beth for sharing your knowledge and insights on infantile spasms. As you heard from our guests, infantile spasms are considered a medical emergency and can deeply impact a child's physical and mental development. Early intervention can be a key to better outcomes. If you suspect an infant is experiencing IS, please consult a physician immediately. And if you feel the need for a second opinion, don't hesitate to get one. As we learned today, CURE Epilepsy has funded cutting edge infantile spasms research whose discovering are making a difference with this challenging condition. Please help us continue to push research forward by visiting cureepilepsy.org/donate. Your support and generosity are greatly appreciated. Thank you.
- Legal Disclaimer: 11:12 The opinions expressed in this podcast do not necessarily reflect the views of CURE Epilepsy. 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 Epilepsy strongly recommends that care and treatment decisions related to epilepsy and any other medical conditions be made in consultation with a patient's physician or other qualified healthcare professionals who are familiar with the individual's specific health situation.