

**Seizing Life, episode 79**  
**Infantile Spasms – Part 1: A Medical Emergency**  
**Transcript**

- Kelly Cervantes: 00:00 Hi, I'm Kelly Cervantes and this is Seizing Life, a bi-weekly podcast produced by CURE Epilepsy. Today on Seizing Life, we address the topic of Infantile Spasms through a panel of experts who bring a range of knowledge, perspectives and experiences with IS. Our panel includes Dr. Renée Shellhaas, a pediatric neurologist and Director of Research at CS 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 Action Network. Beth Dean, the CEO of CURE Epilepsy. And in this episode, I will have a dual role serving as both host and contributor.
- Kelly Cervantes: 00:54 Dr. Shellhaas, Kari, Beth, thank you all so much for joining us today. Dr. Shellhaas, I'm going to start with you with a very sort of basic introduction, what is infantile spasms or West syndrome? And are they the same thing? And then how prevalent is the syndrome?
- Renée Shellhaas: 01:15 Hello. Thank you so much for inviting me. This is such an important topic, and I'm glad to speak about it. Infantile spasms is a very important kind of seizure that is rare in that it happens in around 1 in 2 or 3,000 babies most often, when they're between four and eight months of age, but can happen up to about age two. It's a specific kind of seizure, where a baby will have what we call a cluster or a repeated pattern of events where they may arch or come forward with their head and sometimes their arms, most often as the falling asleep or as they're waking up from sleep.
- Renée Shellhaas: 02:00 This particular kind of seizure is a hallmark or a feature of something called West syndrome but can happen without having all of the features of West syndrome. West syndrome is having infantile spasms, this particular kind of seizure, as well as a particular kind of pattern on a brainwave test called an EEG. So that brainwave pattern is called hypsarrhythmia and having abnormal development. So many children with infantile spasms will have those three things together, but there are also a lot of children who will simply have infantile spasms without having West syndrome.
- Kelly Cervantes: 02:37 Interesting. So you said they most commonly appear between ages four to eight months, but they can happen a little later. So you talked about what a parent should look for, these sort of different movements and arching. How easy is it for a parent or caregiver to spot those movements? Is that reflux? It could be a bunch of different things. And I think as parents, so often we're

trained that we might be overreacting or looking for things that aren't there. So how do you know when it is infantile spasms when it is a seizure and when it's something else?

- Renée Shellhaas: 03:23 It's a really great question. So I think the first thing is, as a parent your baby the very best, you're an expert on what their usual behavior is. And infantile spasms doesn't look like your baby's usual behavior. Infantile spasms and other kinds of seizures tend to happen as the same thing over and over again, as a pattern. It doesn't happen just one time, it happens in clusters, or it'll happen over and over again over a period of minutes or sometimes even up to an hour, but several times a day, so that a baby may have these 100 times a day at the beginning.
- Renée Shellhaas: 03:59 But the most important thing is it looks different from what your baby normally does, and that each time it looks about the same as the time before. I think bottom line though is, if you're worried that your baby's having infantile spasms, you have to ask for help. Because without having a neurologist take a look at your baby and without having an EEG, that brainwave test to help confirm the diagnosis, we really don't know for sure. And so the stakes are high enough that if you're worried, you really should be seeking out help.
- Kelly Cervantes: 04:31 Yeah. I think for us on our journey with my daughter, Adelaide, when she was first diagnosed, she had had a seizure first, but sort of a general seizure. No one had mentioned infantile spasms to us. We weren't looking for it. I had never heard of it before. But what we noticed ... She was she had hypotonia, so low muscle tone, and she had difficulty holding her head regardless. But we started noticing these repetitive head drops that even though ... It was more than her just not being able to hold her head up like usual or she would get tired. It was almost like syncope. And it started happening more and more frequently.
- Kelly Cervantes: 05:17 We would notice it a couple times, it will cluster. It would be like, is that something? Is it not something? And we let it go. Looking back, she was probably having some form of these clusters for a week or two before it got bad enough that it was happening so regularly and so rhythmically, that we were able to recognize that this was so much more than hypotonia. And we called the doctor and she suggested that we bring her into the emergency room so that we could get her admitted. Is that generally the experience that you see?

- Renée Shellhaas: 05:55 So it certainly can be and I think it's a ... You're telling a sort of a classic story of a new onset of infantile spasms. I think the other thing that oftentimes parents will be able to comment on in hindsight is that their baby doesn't feel as bright, they're not as engaged. Or maybe they're not ... Their tone might not be as good as it used to be. Or they used to be able to sit up or rollover, and they're not doing that anymore. And so that we call developmental regression or loss of abilities is also a red flag. So if somebody's baby is having these kind of repetitive events, they have head drops, or their arms coming up, and the baby seems to have regression, that would be a big concern.
- Kelly Cervantes: 06:43 So what are the challenges that parents might face when getting that accurate diagnosis? Because I know that in speaking with other parents who have had children diagnosed with infantile spasms, I've heard horror stories of trying to get a diagnosis. So what is it that can make it so difficult to get that diagnosis?
- Renée Shellhaas: 07:02 Well, I think like you said at the beginning, babies do all kinds of different behaviors. And it's more likely that a baby's going to have reflux than infantile spasms, because reflux is a very common thing for babies to have. And so, I think, oftentimes, we sort of look at each other as parents and we look at other kids and we say, gosh, should we be worried? Or are we confident enough to say there's something definitely wrong? And pediatricians most oftentimes see normal babies too. And so the first instinct is that common things are common, and so most likely baby's going to be fine.
- Renée Shellhaas: 07:42 I think the follow-up there is, again, using your gut as a parent. If you don't think your baby's okay, then you need to push harder. I think also that most of the time, babies who develop infantile spasms have had other challenges already in their life. Not always, but most of the time. And so if you know that your baby's already had a seizure, for example, and a young infant, you might have been counseled or you might ask about infantile spasms as a possibility. Or if you know your baby had neonatal seizures or tuberous sclerosis syndrome, for example, your pediatrician or your neurologist or any other caregiver might be able to have a clue to ask.
- Renée Shellhaas: 08:32 Where it gets really tough as if you have a baby who you didn't know and this was the first thing that came up. And there again, pushing hard to get a diagnosis if something's wrong. And having access to a phone that you can take a video and show to somebody can sometimes really seal the deal.

Kelly Cervantes: 08:52 Yeah. Taking that video is so, so important. But you have to advocate, you have to have the fight for your child.

Brandon: 09:03 Hi, this is Brandon from CURE Epilepsy. Did you know that 1 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 at [cureepilepsy.org](http://cureepilepsy.org). Now back to Seizing Life.

Kelly Cervantes: 09:23 So how is infantile spasms diagnosed? What is that diagnostic journey look like?

Renée Shellhaas: 09:31 Sure thing. So the gold standard, if you will, for infantile spasms is going to be having an EEG or brainwave test and have the baby do the event in question that have an infantile spasm. And that has a specific pattern that we typically will see on EEG, and usually it's pretty obvious and that that event is associated with sort of what we call a detriment. So all of the brainwave kind of slow down and get quiet for a second or two, it happens in a pattern and in a repeated pattern for a particular baby. And very often between the seizures, the EEG is also very abnormal. So that's the standard.

Renée Shellhaas: 10:13 We know though that sometimes and depending on where you live, it may not be easy to get to an EEG right away. And actually, as part of response to the COVID-19 pandemic, professional societies did come out with what we call crisis standards of care for diagnosis of infantile spasms, where we can use the video in the story and talking with a family to make a presumptive diagnosis. But the gold standard is really the EEG.

Kelly Cervantes: 10:40 And you talked about hypsarrhythmia earlier as one of the diagnostic pieces that often goes hand in hand with infantile spasms. What is that?

Renée Shellhaas: 10:50 Sure. So hypsarrhythmia is a very abnormal EEG pattern, where the brain waves are chaotic, so they're not organized like a normal baby's brain waves would be, and that have what we call spikes. So abnormal patterns that are associated with seizures in anybody but then combined with a story of infantile spasms helps us to make the diagnosis. I will say, though, that up to a third of babies who have infantile spasms do not have hypsarrhythmia on EEG. So if the concern is really high, and the baby doesn't have hypsarrhythmia, we still need to push hard to get a good diagnosis because the treatment is about the seizure, not about the EEG.

Kelly Cervantes: 11:35 That's fascinating. I wasn't aware of that. So what are the risks at play the longer that diagnostic journey takes?

Renée Shellhaas: 11:45 Oftentimes, parents will ask me, are those seizures hurting my baby? It's hard to watch a baby have a seizure, even if it's only a second or two at a time. Those individual seizures, those individual infantile spasms are not hurting the baby. But the longer that baby goes without diagnosis and treatment for infantile spasms, the higher the risk of having long term developmental problems and the higher the risk of having long term epilepsy that's hard to treat. So the flip side is the sooner we make the diagnosis and the sooner we start effective therapy, the better than baby's chances of having a good outcome.

Kelly Cervantes: 12:27 Now, Kari, we touched a little bit earlier on these comorbidities with tuberous sclerosis in particular. Can you sort of explain that connection between IS and TSC? And what is TSC?

Kari Rosbeck: 12:45 Sure. Tuberous sclerosis complex or TSC is a rare genetic disorder that causes tumors to grow throughout the body. So predominantly the brain, heart, kidney, liver, lungs, skin. It is the leading genetic cause of epilepsy. So about 85% of individuals with TSC will have epilepsy. The brain dysfunction in TSC really impacts quality of life, from seizures, to developmental delays and intellectual disabilities, to behavioral challenges and infantile spasms. So about a third of infants with TSC will develop infantile spasms.

Kelly Cervantes: 13:25 So is that community educated on infantile spasms? Is this something then that they are told to be on the lookout for?

Kari Rosbeck: 13:34 100%. And I think as we have furthered our research in TSC and really understood that, babies with TSC can actually be diagnosed in utero with the appearance of two or more heart tumors. So they'll receive a probable diagnosis in utero if it appears on just a routine ultrasound. And so those babies, hopefully, will be sent to a neurologist at birth for a confirmed diagnosis. And then we work really hard with the neurology, child neurology community, and through our own social media, our website to really engage in educating those families.

Kari Rosbeck: 14:14 Really important as you were talking earlier, Kelly, what can families do to advocate? Other than taking a video, it's going and educating yourself about what infantile spasms might look like. So as you all were talking about, it can be as simple as a head nod that if you're not watching, you can miss. So it's really important for us at the TSC Alliance to educate our community,

for child neurologists to educate the community. And then we have a cadre of volunteers on social media that as soon as somebody comes to our Facebook page, they jump in and educate parents as well. So it's kind of a three-part educational series there.

- Kelly Cervantes: 14:57 Well, I can see just how important research can be because if we can figure out which babies are higher at risk of developing infantile spasms in utero, then we can inform those parents and educate those parents so that they can be on the lookout so we can get that diagnosis sooner. So Doctor, can you explain what that treatment looks like?
- Renée Shellhaas: 15:20 Over the last 5 or 10 years, we've really made great advances in understanding what the standard treatments are for infantile spasms. And what we found as a research community is that there are three standard approaches for infantile spasms. There is a medicine, it's an injectable, high dose steroid called ACTH. There is an oral medicine that's prednisolone or another steroid that we can give by mouth. And then there is a specific medicine called Vigabatrin. Those three medicines all can work for infantile spasms treatment.
- Renée Shellhaas: 15:56 Everything else that we have tried has failed. So really important for the child neurology community to know there are only three choices. And there are reasons that you might pick one of those three. For example, there's some really good data to suggest that babies who have tuberous sclerosis complex respond fast to Vigabatrin. And so that's going to be my first choice if I know the baby has TS. For other babies, it's a little bit less clear, but starting with one of those three medicines is certainly the way to go. And the way that we do that is we hit them hard at the beginning with high doses of medicine. And within about a week, usually we have a sense of whether it's working or not.
- Renée Shellhaas: 16:46 By 14 days of treatment, we're reassessing, we're looking at that EEG again, and we're talking to parents to say, are you seeing any more seizures? And if things are starting to get better, then we can start to slowly actually taper down if they've been on either oral steroids or ACTH. So that they have a month of their first line treatment. In the best case scenario, the spasms are gone, baby does well, and we watch them really carefully. But they do well in the long run. If however, at that 14-day mark or before then things are getting worse or not getting better, we may switch to a different one of those three treatments to see if we can get a better response.

- Kelly Cervantes: 17:26 I understand that some of those medications can also be used together in conjunction. I've personally read some studies that say to use Vigabatrin and ACTH together. All of these drugs really stink, I think is a really nice way to put it. They all have pretty scary side effects. And I think that when the doctors are talking to parents and caregivers about these drugs, they can be pretty scary. What sort of reassurances can you provide a spasm can look pretty, not so bad compared to what the side effects of these drugs can be? Why is it so important for a parent to put their kid on one of these kind of scary drugs?
- Renée Shellhaas: 18:18 That's a really good question. All of the medicines that we use have side effects, for sure. And that's why sticking with one of the three standard therapies is so important. We don't want to give a baby one of the medicines that we now know doesn't work and expose them to potential side effects from those medicines that don't work. For the ones that we do have, yes, they have scary sounding side effects, and they are scary side effects. For the most part, we can mitigate those. And for families who are going to have ACTH or oral steroids as their first line treatment, the main thing that we see is really fussy, cranky babies. And they're hungry because steroids make us hungry.
- Renée Shellhaas: 19:01 Although it's easy from my end to say this, what I say to families is, go ahead and feed them. If they're hungry, we're just going to feed them and they're going to gain weight. That's okay. And remind ourselves that this is temporary. That we're going to be on high dose steroids only for a few weeks. And those weeks I know can seem like forever, but it is temporary. And if in the long run that's the right treatment that's going to take care of these seizures, we'll all be glad that we did it.
- Renée Shellhaas: 19:30 We also talk about self-care for parents and who can we rely on when we need a break especially if the baby's really cranky and fussy. When can mom or dad step away and have a nap or go for a walk to make sure that we're really supporting because this is tough. And oftentimes it's really throwing into a community that families weren't associated with before and just finding your people to help support is really important.
- Renée Shellhaas: 20:01 For Vigabatrin has a separate set of side effects, and we talk very carefully about those. The main one that families need to be aware of is the possibility of visual field loss. What does that mean? It means that for some people who take this medicine for a long period of time, they don't see well, generally, on the outside of their vision. And that doesn't get better if it happens. The good news is that the less time that you need the medicine,

the less likely it is that that visual field loss is going to happen. Overall, the feeling in the community is that the risk is worth the potential benefit to make sure that we're doing the right thing for baby.

- Renée Shellhaas: 20:48 Now, you said, what about giving combination therapy, there are some data to suggest that you might get a faster response with combination therapy. There are still more studies to be done to really understand the very best way to do that, but it's certainly something that some places will offer. And family should talk about what are the pros and cons of starting with two drugs all at once versus starting with one and then quickly switching to another one if it doesn't work.
- Kari Rosbeck: 21:15 If I could add, Kelly, to that with Vigabatrin that's used a lot in the TSC community, we often say, stopping infantile spasms is so important. They may not be able to drive, which is the concern with peripheral vision loss, if you don't stop the seizures, that's the paramount thing, that's so important.
- Kelly Cervantes: 21:39 Yeah. I think that it's such an important message to get across to parents is that stopping the seizures is so much more important than the ugly side effects. I can say, having experienced giving my child all three of those medications on multiple occasions, because unfortunately, my daughter was one where the drugs would work for a short period of time, and then unfortunately, the spasms would return, be it a couple months later or a year later. And so we just kept trying different things. And her brain, unfortunately, always found a way to work around the treatment. Her brain was just set on seizing, I suppose.
- Kelly Cervantes: 22:28 But there isn't one, I don't think that's ... It's really a conversation that you have to have with your doctor and figuring out what is the best. And if you do have an underlying cause, helping that try to determine the best medication choice, but the most important thing is to get on one of those three medications as quickly as possible. So what does successful treatment of infantile spasms look like with one of these three medications?
- Renée Shellhaas: 23:00 So successful treatment is what we're all aiming for, it's the best case scenario. And I think of it as rebooting the computer. Things were going wrong and we start with high doses of medicines, and we see those spasms melt away. And that might look like a cluster of spasms that used to last for 30 minutes becomes 10 minutes, 5 minutes, 2 spasms, 1 spasm and it's gone. Or they may become less and less obvious, even for



parents who know exactly what they're looking for. And at the same time, the baby becomes brighter and starts to reengage. If they had any kind of developmental regression, you might start to see skills develop.

- Renée Shellhaas: 23:46 So I had a baby years ago now, who responded beautifully to ACTH. And that child had ... She'd been sitting and like starting to crawl and her spasms began. And by a month of treatment, she was walking, and she was bright and ready to go. That's what we're looking for, is the seizures go away and the babies engage. Now we'll follow on EEG. And part of that is we want to see the EEG clean up and look as normal as possible. But the most important thing is how the baby's doing.
- Renée Shellhaas: 24:21 One of the reasons will repeat the EEG is sometimes those spasms can become so subtle that we really can't tell unless we have a video and an EEG running at the same time so we can compare. And if we're that close, and we might push for another week or two of higher dose medicine just to kind of finish things out strong so that we have the best possible outcome.
- Kelly Cervantes: 24:44 Now, my situation with my daughter, Adelaide, was extreme. And so I always I'm wary of any parent who is walking into this diagnosis to necessarily compare to my family's story because it is on this very extreme end of the spectrum. But for my daughter, the spasms did come back, along with other seizure types. What is that recurrence look like? And is that something that all parents need to be aware of?
- Renée Shellhaas: 25:15 Absolutely. So even if we get that best case scenario where the seizures seem to be going away, we're going to be in close touch, and be watching that baby carefully. Because it is possible that spasms could recur, especially after we taper them down off of those medicines, or that other seizures could arise. And we need to know what to look for. Oftentimes, by the time we're treating infantile spasms and we've gone through a diagnostic workup, so babies had a brain MRI, or they've had genetic testing, that'll help us understand what the risk is and other kinds of seizures or recurrent infantile spasms. And that will help us with guiding families about what to look for and what to expect.
- Renée Shellhaas: 25:57 But it is not uncommon, unfortunately, that children will have a recurrence of their infantile spasms even after they seem to have resolved. And then it's a case by case basis about maybe they responded so well the first time to a treatment, maybe we want to go back to that same treatment, because we just need a

second round. Maybe we need to try different ones. Maybe the baby was on Vigabatrin before and we're going to switch to prednisone or ACTH, or vice versa, depending on each individual situation.

- Renée Shellhaas: 26:30 But I think the main thing is, if you have a sense, I'm worried that something's going wrong in the seizures and back to pick up the phone right away and call your neurologist and get seen again quickly, so that we can again, restart the right therapy and make sure we're doing the right thing by baby.
- Kelly Cervantes: 26:49 So given how difficult it can be for a parent to understand IS, and it was mentioned earlier that a pediatrician may not see a lot of IS patients over the course of their career, are there guidelines in place to help physicians diagnose infantile spasms?
- Kari Rosbeck: 27:10 Right. So infantile spasms can be difficult to diagnose. But the good news is that we have good consensus around those three standard treatments, and we have guidelines from the American Academy of Neurology and the Child Neurology Society, about how to start those treatments. We also have guidance from, again, Child Neurology Society and the Pediatric Epilepsy Research Consortium about how to do efficient diagnosis and treatment initiation in the era of the COVID pandemic, where we had to change some of our medical practice, but still want to highlight that early diagnosis and treatment is an emergency.
- Kari Rosbeck: 27:54 And so we don't want to let the COVID pandemic in the way of getting that diagnosis and treatment in place. And it doesn't matter what the reason is for the infantile spasms. No matter what, the earlier we can make that diagnosis and start the right medicine, the better the infant will do.
- Brandon: 28:16 Join us for part two, where we focus on the research currently being done on infantile spasms and the hope that it promises. Part two will be available to download and stream on Wednesday, December 15th, at CURE Epilepsy's website, YouTube channel and most podcasting platforms.
- Brandon: 28:38 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 health care professionals who are familiar with individual medical conditions and needs.

Brandon:

29:00

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