Infantile Spasms Transcript

Kelly Cervantes: 00:00 I'm Kelly Cervantes, and this is *Seizing Life*, a weekly podcast produced by Citizens United for Research in Epilepsy, CURE.

Kelly Cervantes: 00:18 On today's episode, we are joined by Amy Brin Miller. For the past two decades, Amy has worked on behalf of children with special needs and their families. Since 2015, she has been the Executive Director of the Child Neurology Foundation, where she continues to guide the organization to ensure that all children affected by neurological disorders can achieve their full potential.

Kelly Cervantes: 00:39 Today, she discusses infantile spasms, a devastating form of epilepsy that my family is all too familiar with but is relatively unknown by most. Amy, thank you so much for joining us today.

Amy Brin Miller: 00:52 Absolutely. I am thrilled to be here. Thanks for having me.

Kelly Cervantes: 00:54 Of course. So, infantile spasms. Let's talk.

Amy Brin Miller: 00:59 Yeah, the word ... It sounds very innocuous, but it is not nearly as scary of a word as the condition actually is.

Kelly Cervantes: 01:09 What are infantile spasms?

Amy Brin Miller: 01:11 Spasms make you think that someone's just kind of flailing around, when, actually, infantile spasms are a catastrophic form of pediatric epilepsy. It's very, very rare, and it starts with very few signs.

Kelly Cervantes: 01:26 Subtle.

Amy Brin Miller: 01:27 Subtle signs. Just head bobs. Sometimes people think it's maybe an upset tummy and some GERD, or maybe it's just the usual baby reflex that's happening, but, actually, it's pretty significant. Changes are going on in the brain and there is major seizure activity.

Kelly Cervantes: 01:44 So how are infantile spasms diagnosed?

Amy Brin Miller: 01:49 Even though it's a rare disease, what's interesting about it is that it actually has a pretty clear diagnostic criterion. What needs to happen is the child needs to have an EEG. That's just a test that measures what's going on inside the brain. The brain activity. When the physician reads that test, he or she is looking for a very distinctive pattern which is called Hypsarrhythmia. It's

		a chaotic pattern of brain activity that shows the infantile spasms are happening. Unfortunately, just because a child is having infantile spasms and has an EEG, it doesn't always show up right away. We always talk to families about the importance of requesting a longer EEG to make sure that you're capturing that brain activity for as long as you can to see if hypsarrhythmia is present.
Kelly Cervantes:	02:41	My understanding is that the hypsarrhythmia is also far more prevalent sometimes in sleep, which really makes that overnight, 24-hour-plus EEG really, really imperative to catching that brainwave pattern.
Amy Brin Miller:	02:57	What you're talking about, Kelly, just shows what happens when a parent becomes more and more educated about a disease. When you first learned about infantile spasms, you probably weren't thinking about when Adelaide was having the seizures, and what would be the maximum opportunity to capture them. Let me first just say that you're exactly right with what you're saying, but there’s another reason why it's so important that families start to understand what they're seeing when they know something is wrong with their child. It is important that they act, and that they're in a relationship with a clinician so they can say, “You know, so we did an hour EEG, but we still didn't capture it. What would you think about doing an overnight EEG?” and feeling empowered to ask that question and have that conversation with their clinician.
Kelly Cervantes:	03:42	Our daughter was diagnosed at nine months with infantile spasms. She'd had her first seizure at seven months and, now looking back on it, even looking back on videos that we took in between that seven to nine-month range, I'm pretty sure that she was having spasm clusters for weeks, if not a month, prior to us actually getting that diagnosis. That is heartbreaking to think about, that you're missing something, but I consider myself a very attentive parent. We were already meeting with neurologists, and we knew that something was not quite right when we were looking for things. And we still didn't catch it immediately, which just goes to show how subtle these signs are.
Kelly Cervantes:	04:26	Like you mentioned, what can parents look for? What do infantile spasms look like, specifically?

Amy Brin Miller:	04:34	Again, I wish there could be just one form, but the biggest thing I would always suggest to the family is, if you feel like something's wrong, pay attention to that. Then connect the dots. If you start seeing head-bobs, or if you start seeing what are sometimes referred to as a jackknife kind of posturing. Those can all be signs of something bigger happening. It's not just a typical reflex or, like I said, GERD.
Amy Brin Miller:	05:03	When I came to the foundation, I was trying to get up to speed pretty quickly on infantile spasms. We had a presence in that space for a while, trying to understand really the family's journey to getting to the diagnosis. Like I said, even though it's a rare disorder, unlike other rare disorders, there's a clear way to diagnose.
Amy Brin Miller:	05:25	So we partnered with another organization, Tuberous Sclerosis Alliance, and we interviewed about 15 families to understand when they first knew something was wrong and how long did it take them to act. Who did they go to? Then, eventually, how long was the timeline to get the I.S. diagnosis? I share this just in response to you sharing about your family's story.
Amy Brin Miller:	05:47	Of these 15 families, over 90% of them knew something was wrong within a week of first seeing a head-bob or a jackknife reflex. Within another week, they accessed healthcare. They were doing everything they should. And who did they access? They went to the emergency room, or they went to the pediatrician. Basically, two weeks went by from the first sign that something was happening with that child. It took them an average of six months to get the diagnosis of I.S., and that was after seeing an average of four physicians.
Amy Brin Miller:	06:20	This is, unfortunately, the norm for the families in our community. That's why the foundation and CURE, along with 26 other national organizations, have been working together to increase awareness about this devastating disease. It is so vital. This is no good. Right?
Kelly Cervantes:	06:39	[crosstalk 00:06:39] completely unacceptable [crosstalk 00:06:41].
Amy Brin Miller:	06:41	We can do something about this.
Kelly Cervantes:	06:42	Yes.

Amy Brin Miller:	06:43	It's going to take all hands-on deck to do it.
Kelly Cervantes:	06:45	When should parents be most on alert? When do infantile spasms usually present?
Amy Brin Miller:	06:52	That's a great, great question. Really, from birth to six months is the greatest risk. My son just turned six months. You kind of hit that, and you think, “Oh, we're going to be sitting up.” I honestly just took a big sigh of relief because we were out of that risky period. That does not mean though that an older child cannot develop I.S.
Kelly Cervantes:	07:12	I was going to say, Adelaide was diagnosed at nine months, and then it came back several times. Most recently, when she was 2 1/2, which is ... That's not an infant.
Amy Brin Miller:	07:22	That's exactly right. It again speaks to your comment earlier that the name of this disease doesn't really play nicely with creating awareness and understanding. It’s an oxymoron. I think parents need to understand that it's from birth to six months. I mean, a goal of ISAN is that every mother and father in America should have this on their radar, just like the “Back to Sleep” campaigns, and our whole country knows now we put babies to sleep on their back.
Amy Brin Miller:	07:53	Also, you should trust that gut. Just because your child is seven months or a year old, and you start seeing these head-bobs, be active. Know what to do and have the conversation with your provider.
Kelly Cervantes:	08:05	Absolutely. So, you talk about it being very rare. I think the number is ... I think there are 1,200 children diagnosed [crosstalk 00:08:12] every year.
Amy Brin Miller:	08:15	In our country. In the United States.
Kelly Cervantes:	08:17	This sounds relatively rare, but I have to say that we know three or four families who live within several miles of us who all got the diagnosis within a year of Adelaide. Granted, we live in the big City of Chicago, so it's a little more compact, but these people are out there. The diagnoses are out there. If you're not talking about one in a million, it is happening, and a pediatrician, and certainly an epileptologist and neurologist, is going to see it more frequently. Maybe only one or two cases

over the course of a pediatrician's practice. Why is it so devastating?

Amy Brin Miller: 09:00 It's devastating for a couple of reasons. I mean, for the disease state itself, it's devastating because that chaotic brain pattern just wreaks havoc on a developing brain. Any damage that's happening to that brain is incredibly scary. Those neurons are forming, and we want that brain to be as healthy as possible to give that child his or her optimal potential. If there's an assault on that brain, we want to stop that trauma from happening. That's what people need to understand about seizures. It's an assault on their brain, and you have to stop it at all costs.

Amy Brin Miller: 09:44 The other reason that I think that I.S. is devastating, and I think it speaks a little bit to what you were saying about more cases being out there than we know about, is the fact that it goes underdiagnosed. The fact that these children are seizing for weeks or months at a time, and, by the time they do get a diagnosis, it might not be I.S. at that point. They might get diagnosed with another form of epilepsy just because of age or, at that point, the presentation of the disease. That, to me as a healthcare provider, is absolutely devastating, that the child had a need and, for whatever reason, we missed it.

Kelly Cervantes: 10:19 Because infantile spasms are treatable in some cases.

Amy Brin Miller: 10:24 Absolutely.

Kelly Cervantes: 10:24 Some children, some babies who are diagnosed and get the frontline treatments within a relatively quick period of time of the seizure presentation, can achieve seizure freedom and go on to lead normal, if not relatively, normal lives. That is an absolutely possible outcome as long as it’s diagnosed and treated immediately.

Amy Brin Miller: 10:48 Yeah. There's an urgency to treat because that's what everyone wants, for a child to reach his or her optimal potential. To do that, you have to stop the seizures. You're exactly correct that if we can stop the seizures, then there's a chance the child might have mild developmental delay and, with appropriate therapies, could go on to a typical, functional level. But, the longer the seizures go unmanaged, the more assault is happening on that developing brain.

Kelly Cervantes: 11:20 Right.

Amy Brin Miller:	11:20	So, the more we can do to create better awareness, the more help these children can get in being diagnosed and appropriately treated. It's a responsibility for all of us.
Speaker 4:	11:31	Enjoying this episode of *Seizing Life*? Learn more about epilepsy by visiting cureepilepsy.org. Since 1998, CURE has raised more than $60 million to help fund over 220 cutting-edge epilepsy research projects in 15 countries around the world.
Speaker 4:	11:45	Now, back to this episode of *Seizing Life*.
Kelly Cervantes:	11:48	I want to bring ISAN back in because I think that you don't see this a lot in the scientific and research community. Organizations coming together and creating an alliance like this is rare. I'm so proud that CURE is a part of this, and we're just so grateful for the Child Neurology Foundation to house ISAN. I got to attend a meeting last year, and I was just so impressed with how passionate all the organizations are.
Amy Brin Miller:	12:23	I think why ISAN is so successful is it was born out of the community's request and the community's need. I think, when you respond to a need, then you're set up for a really successful venture. ISAN, or the Infantile Spasms Action Network, is a collaborative advocacy model of 26 national and international organizations that have come together to raise awareness about infantile spasms. And you're exactly right that this is a model that's very unique, which is why, I think, we were able to bring together so many organizations, and every year we add more partners. So, it started in 2016 with 15, and, now in 2019, we're at 26.
Amy Brin Miller:	13:04	In that, you have the patient advocacy community that touches infantile spasms. Again, if it's maybe a form of epilepsy like Lennox-Gastaut that I.S. evolves into, then you've got them at the table. With CURE, Epilepsy Foundation of America, and Tuberous Sclerosis Alliance, their patient populations have very large incidents of I.S. Also, at the table, you have all the physician and nurse organizations that touch I.S. from the neurology standpoint, including pediatricians and emergency physicians. Then, you have corporate partners at the table who have an interest in serving this patient population.
Amy Brin Miller:	13:44	The goal is to speak with one voice because, if you are trying to make an impact and raise awareness, why in the world would you want splintered communication going out? One of the first

		things that we did with ISAN was establish the Infantile Spasms Awareness Week, which is every year from December 1 through December 7. The second thing we did was we started to look at other disease models, and how they created awareness. We looked at things like a stroke, where there was an opportunity to develop a mnemonic that would be branded and easily recognizable but would also include steps that the general public could take once they're activated to this message.
Amy Brin Miller:	14:24	That was really in 2017, when ISAN developed the S.T.O.P. mnemonic, which really brings awareness to the urgency around infantile spasms and gives families immediate steps to take once they have seen the sign.
Kelly Cervantes:	14:41	What does S.T.O.P. stand for?
Amy Brin Miller:	14:43	S means "see the signs." That's where we are wanting families to feel empowered when they start seeing those head-bobs, those jackknife movements, and knowing there's something in your internal gut saying that something's wrong. T is "take a video." We live in a digital age so there no longer needs to be a situation where you're just verbally having to pontificate and put words together about what you're seeing. Shoot a video, and email or text it over to your physician to show it to him or her. O is "obtain the diagnosis." That really speaks to having the conversation with the provider that you want an EEG because the EEG leads to the hard diagnosis of infantile spasms. Then P means "prioritize treatment." That's when we are wanting the families to engage in a healthy conversation with their clinicians about what the treatment options are and which ones work best for their child.
Kelly Cervantes:	15:37	That leads me perfectly into the discussion of treatments. We mentioned earlier that infantile spasms for some kids are treatable. Those treatments are terrifying.
Amy Brin Miller:	15:53	When the clinician comes to you, you have to, for some of them, sign-off on paperwork about vision loss or all of these other side effects. There can be incredibly strong steroids that you're injecting into your child's body. They're scary.
Kelly Cervantes:	16:10	Yes.
Amy Brin Miller:	16:12	What I think is scarier is not treating it. These treatments are not popping an aspirin. I mean, they're time-intensive

		resources, and they should be made with due diligence and good conversation. What families absolutely need to hold on to, I would say at the forefront of that decision making, is that by not having appropriate treatment, you are continuing to allow your child's brain to be damaged. The best way you can stop that damage is by taking one of these treatments.
Amy Brin Miller:	16:51	That again, definitely from ISAN, but also the Child Neurology Foundation, is a lot of the work that we do in empowering, educating, and supporting families to understand how to best partner with their medical professional. How do you have these difficult conversations? How do you express what your goals of care are and be able to hear how the provider's coming back in his or her assessment of what those treatment options are? These treatments do have risks, but that doesn't mean that those risks outweigh the risk of not stopping the spasms.
Kelly Cervantes:	17:27	We've touched on the fact that not every pediatrician is familiar with infantile spasms. Let's say a parent is concerned that gut-alarm is going off. Something is wrong. They see an infantile spasms video, they take a video of their own child, and they take it into their pediatrician. Their pediatrician, however, is like, "I don't know, it's probably just a really sensitive reflex or startle." What next steps can that parent take?
Amy Brin Miller:	17:56	First, I do want to also say that we're talking about one in 12,000 children, so I think it's important to give the pediatricians and emergency physicians a bit of a break in terms of their big role being to reassure the family. I think that's okay. I think that the issue here is, when you've got a parent that is saying there is something wrong, that provider needs to be responsible to that need. So, if the family is feeling that the provider they're talking to is not being responsible with their feedback, then I would encourage that family to go to another provider.
Amy Brin Miller:	18:38	If you've accessed your primary care provider, then I would go to an emergency room. Or, depending on your insurance, do you need a referral? Or can you go ahead and see a neurologist? I think it's also important for us as advocates to make sure that, when we're giving this anticipatory guidance to families, we aren't saying you have to go to see the nation's expert in I.S. who lives five states away. That's just not realistic for a lot of families. Just based on life.

Amy Brin Miller: 19:14 That, again, is the goal of ISAN. That any provider should be aware of I.S. and be able to respond to the family's need. This doesn't necessarily mean that a pediatrician is going to feel comfortable making the diagnosis of a rare pediatric epilepsy condition. That's asking a lot of a provider. But he or she should be able to know who to refer that patient and family to who is comfortable making that diagnosis.

Amy Brin Miller: 19:44 I think it goes back to a little bit of understanding of the various players who are involved in this. At the core, it’s most important making sure that patient and family are active and educated to maneuver the situation based on their goals of care.

Kelly Cervantes: 20:06 I just think that's incredible advice. If all else fails, you go to that emergency room.

Amy Brin Miller: 20:12 That's exactly right. And you keep asking. The other kind of resource that we should talk about is called our Family Support and Empowerment Program. We promote it through ISAN, but it's a program that the Child Neurology Foundation administers. This is a program that right now is working with families in all 50 states and 58 countries. What we do is, when a patient or family contacts the foundation, we pair them up with a parent who has walked a little bit further along in their path. They've received some training, some mentorship, and they understand how to help support the family and the patient where they're at. Whether that's with information or resource navigation, or just to help with the emotional experience that this family is going through.

Amy Brin Miller: 21:00 So this is another resource. It's 24/7 available, so families can call if they're hitting a brick wall, saying, "I'm doing everything you're telling me to do and no one is still listening." Don't feel like you're alone. We talked about this earlier, that a lot of families feel so isolated. This is a great inroad, and it gets you connected to a larger network of people who can also help you problem-solve. Unfortunately, healthcare from the outside looks like it should be black and white. But healthcare isn't. For those of us who have walked and lived in it, unfortunately, it is not black and white.

Amy Brin Miller: 21:37 When families have these sorts of emergencies, they immediately think, “Oh, I should be able to do x, y, and z and it should move on.” Hopefully, it does. But in case it doesn't, know that you're not alone. Know that you can go and get some

		additional resources that are free. Feel empowered that there are people out there who want to walk with you and they want to help you.
Kelly Cervantes:	21:58	Without ISAN, the Child Neurology Foundation, and these organizations, we wouldn’t have Infantile Spasms Awareness Week, and the S.T.O.P. mnemonic, as well as an incredible video, which ISAN launched at the end of last year. This video can be viewed at www.seizinglife.org/ISAN, as well as on the Infantile Spasms Action Network website. It clearly shows babies who are having infantile spasms, so parents can know specifically what to look for. This way pediatricians have a reference point.
Amy Brin Miller:	22:41	Totally. It's Monica Jones from the Brain Recovery Project who produced that video. ISAN gives a little bit of grant money to our partners to basically let them focus on their strengths, which then furthers the mission and message of ISAN. There are multiple levels of giving these families the tools to be able to navigate the conversations that are needed to get the diagnosis immediately.
Amy Brin Miller:	23:09	At the end of the day, I'm a big believer that this is our watch. We're the adults right now in this world, and what is happening is unacceptable. If we don't start demanding change and working towards it, then shame on us. I hope that in my lifetime, the state of I.S. and some of these other rare disorders is just completely different because we're able to activate families appropriately, as well as partner them with the clinicians who know how to take care of these diseases.
Kelly Cervantes:	23:40	Amy, thank you so much for coming.
Amy Brin Miller:	23:42	Absolutely.
Kelly Cervantes:	23:43	And for chatting and sharing your wealth of information on this topic and the resources available to parents.
Amy Brin Miller:	23:49	I wish everyone said I had a wealth of information. Most people are like, enough already. But, thanks for having me over, and it's always, always so fun to spend some time with you Kelly. Thank you for everything you're doing. Not just for your family, but for all the families who are out there. Our whole community is better because of you.

Kelly Cervantes:	24:04	Thank you so much. I wouldn't have it any other way.
Kelly Cervantes:	24:10	Thank you again, Amy, for all you're doing to bring awareness and education to the general public, as well as the medical community. Infantile spasms are an emergency and need to be treated as such. This has to start with increased caregiver knowledge, followed by clinician support. If you want to know more about the diagnostic and treatment challenges of infantile spasms, then be sure to check out a free webinar at CUREepilepsy.org/InfantileSpasms.
Kelly Cervantes:	24:37	Finally, you can sign up for information about upcoming podcasts, or listen to past episodes, at CUREepilepsy.org/SeizingLife. Thanks so much.
Disclaimer:	24:54	The opinions expressed in this podcast do not necessarily reflect the views of CURE. The information contained here 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.
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