

For more information on
infantile spasms, please visit:

www.infantilespasmsinfo.org

For more information on the
Child Neurology Foundation, please visit:

www.childneurologyfoundation.org



“What we hope to do on the Child Neurology Foundation’s website and on our IS designated website (www.infantilespasmsinfo.org) is to provide information you can trust. There are so many sources of information out there that aren’t vetted by anybody, not by a doctor, let alone a child neurologist. We want to make sure that this information is completely honorable, vetted, and trustworthy so that parents are as informed as possible before going to see their pediatrician or child neurologist.”

- John Stone, Executive Director, Child Neurology Foundation

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Understanding Infantile Spasms (IS)

INFANTILE SPASMS

Creating Hope for Better Developmental Outcomes



- Logan, age 4, with his doctor, Jong Rho

What is Infantile Spasms (IS)?

“Infantile spasms is classified as a catastrophic childhood seizure disorder, and it’s not catastrophic because of the way the spasms look. If a child is having infantile spasms, it doesn’t look like such a big thing... just little jerks. It does not look frightening, yet the impact could not be greater. That child with infantile spasms is now threatened with a very poor developmental outcome.

Unfortunately, the majority of children are going to eventually be found to be cognitively impaired, and most will continue to have other seizure disorders later on.”

Don Shields, M.D.
Professor Emeritus of
Neurology and Pediatrics
David Geffen School of
Medicine at UCLA



Don Shields, MD

Infantile Spasms typically begins in the first 4 to 8 months of life and is characterized by flexion (bending and jerking) of the trunk (torso) or neck and extremities (arms and legs). An episode can range from a subtle head jerk to a flexion that lasts for a few seconds. Most often, the spasms occur in clusters.

Infantile spasms are characterized by hypsarrhythmia (hips-A-'rith-mE-uh), a highly disorganized and chaotic pattern of electroencephalogram (EEG) abnormality. The presence of hypsarrhythmia, which does not typically occur with other forms of epilepsy, can help to confirm a diagnosis of IS. Furthermore, there is a strong correlation between hypsarrhythmia



and the cognitive impairment (damage) and developmental delays that are often associated with infantile spasms. That’s why an EEG is so important; it helps determine what the underlying disorder is that needs to be treated.

What Causes Infantile Spasms?

Unfortunately, not much is known about what triggers infantile spasms. Scientific research is needed to enhance the understanding of infantile spasms – and better understanding has the potential to lead to improved treatment and developmental outcomes for patients with IS.

What is Symptomatic Infantile Spasms?

In about 70% of cases, the cause of IS can be linked to causes such as a central nervous system (CNS) infection, brain developmental abnormalities, or genetic abnormalities. These cases of IS are considered to be “symptomatic.” Understanding the cause or origin of IS can impact the course of treatment selected, which can ultimately affect a child’s prognosis (outcome).

What is Cryptogenic Infantile Spasms?

In approximately 30% of cases, there is no known cause for the IS. When the cause or origin of IS cannot be identified, the condition is characterized as “cryptogenic.”

How is Infantile Spasms Diagnosed?

While infantile spasms is a rare form of epilepsy, it can be identified by its distinct (unique) characteristics if physicians and caregivers are aware of the telltale signs. Infantile spasms can be mistaken for colic, but colic does not typically occur in the clusters that are typical of IS. Infantile spasms is typically diagnosed by observing spasms in a child who is in infancy who also displays hypsarrhythmic EEGs.

“Colic is the most common initial diagnosis made; but the spasms occur at times that aren’t necessarily associated with feeding – which colic is – and they occur in a series, which makes them unique in contrast to colic.”

John Bodensteiner, M.D.
Senior Associate Consultant
Mayo Clinic



John Bodensteiner, MD

Patients with spasms will present in a variety of ways, but the most typical is a recognition of little flexion jerks that can be mistaken for a little startle reflex in children, and that happens fairly commonly. The thing that really triggers knowing that there’s something wrong is the child’s development either stops advancing or actually draws back some so that they lose developmental milestones. Then you know there’s a real problem. And even if the spasms are kind of subtle, that will bring them to the attention of a medical professional.

Why is it Important to Diagnose and Treat Infantile Spasms?

Infantile spasms can result in developmental delay and cognitive development issues. Babies often stop achieving developmental milestones and may even lose skills such as sitting, babbling or rolling over in conjunction with the onset of IS. For this very reason, early recognition, diagnosis and treatment of infantile spasms is critical and can improve the developmental outcomes of infants diagnosed with IS.

It is important to identify spasms early on and to treat as soon as possible because there’s growing evidence that delay in treatment reduces the probability that the patient will do well developmentally.



“My life has been pretty normal. I make friends with kids all the time. They accept me for what I have and who I am. I do activities outside of school, like swimming and other sports. I just feel like a regular kid. Next year, in fact, I’m planning to attend college.” - Ty Onda, 18



The Onda Family

More than most families, the Ondas know about the effects of infantile spasms. Ty Onda, now 18, had his seizures stopped as an infant, although he still suffers from the underlying cause of his disease, tuberous sclerosis. Ty, a competitive swimmer and good student, heads to college next year in the hopes of studying biology. His step-sister, Mami, was also diagnosed with infantile spasms, but suffered from refractory seizures which stunted her developmental progress. Mami’s step father, Rocky, said even though her cognitive development was impeded, she still lives a “happy and good life.” Ty’s infantile spasms were diagnosed quickly, in part, because of their experience with the disorder with Mami.



“I’m very proud of what Ty has accomplished to date and what he will be able to do in the future.”

- Rocky Onda, Ty’s Father

How is Infantile Spasms Treated?

In infantile spasms, the goal of treatment is to eliminate the spasms and hypsarrhythmia as quickly as possible. Awareness and proper identification can result in the selection of appropriate therapy that can improve a patient's developmental outcomes.

"Even though it has been close to seven years for Logan being seizure free, it's still very close to our hearts and something we can remember like it was yesterday. So when you look at him, when he does something for the first time like take a swimming lesson or write his name, it's just a little extra special knowing what he went through and where he was to where he is now." - Tiffany and Ryan Allen

The Allen Family from Arizona knew something was wrong with their infant son, Logan, when his tiny body was gripped by seizures. By quickly getting to a child neurologist, Dr. Jong Rho, Logan's seizures were controlled. Now 7 years old, Logan is developmentally on track with his peers.

"Happily for Logan, early, aggressive intervention with ACTH wound up being potentially very helpful for him long-term. He's not had a recurrence in terms of any type of seizure problem and, for that matter, I would say that most people who meet him these days would not ever guess that he had such a catastrophic epileptic condition as a young infant."

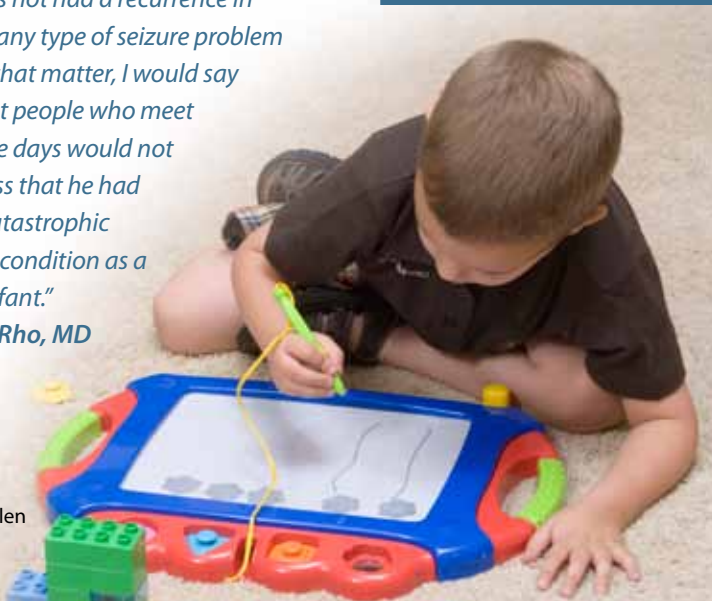
Jong M. Rho, MD



The Allen Family



Ryan and Logan Allen



Logan Allen
at play

Physician's Role

Patients often first present to their pediatricians. Once a proper diagnosis is made, treatment often includes the involvement of child neurologists, neurosurgeons, and/or dietitians and other health care professionals who specialize in epilepsy care.

"There are a number of different strategies that have been used over the years to try to treat these spasms. Treatment includes medications, surgical interventions, such as taking out abnormal pieces of brain that might have developed aberrantly during development, or other non-pharmacologic treatments, such as the ketogenic diet, which is a non-drug form that can be very effective for treating some patients with spasms as well. No single therapeutic intervention has proven to be universally effective, so really the clinicians have to look at all of the treatment options and try to tailor them as much as possible to the particular patient with spasms."

Jong M. Rho, MD
Head, Division of Pediatric Neurology
Alberta Children's Hospital

Medication

There is no consensus on the best initial approach for infantile spasms. Many experts recommend a short course of hormonal therapy (ACTHar) but others use oral antiepileptic drugs (vigabatrin or topiramate) or oral steroids (prednisolone). H.P. ACTHar[®] Gel and Sabril[®] vigabatrin are the only FDA approved treatments for infantile spasms. Other approaches such as the ketogenic diet or valproate can be effective if the primary treatment fails.

H.P. ACTHar[®] Gel has a direct effect on the brain and may work by helping the body produce natural hormones with the goal of providing rapid resolution of spasms with clinically significant improvement in the chaotic EEG pattern (hypsarrhythmia) often times within days to weeks at approved dose. Potential side effects include infection, hypertension, irritability, and changes in appetite.

Vigabatrin works by increasing the availability of the main inhibitory neurotransmitter in the brain. While relatively safe and effective,

there is a risk of permanent vision loss which must be balanced against the consequences of untreated spasms. Vigabatrin is especially effective in children with tuberous sclerosis.

Off-label use of oral steroids can be effective, but fewer children respond than to hormonal treatment. Side effects are similar to Acthar.

“There are two treatment options that are generally considered to be the most effective, and that’s ACTH and Vigabatrin, and which of those two treatment options one chooses at the beginning depends a lot on the patient and what kind of underlying problem we might identify.”

Don Shields, M.D.

Professor Emeritus of Neurology and Pediatrics

David Geffen School of Medicine at UCLA

Vigabatrin appears to be most effective in treating infantile spasms caused by tuberous sclerosis (a disorder which is associated with damage to the brain, skin, heart, and kidneys).

Diet and Non-Medication Treatment Options

Ketogenic diet: This form of therapy for IS has been around since the 1920s and is sometimes used in combination with medication therapy. A ketogenic diet is a high fat, adequate protein, low carbohydrate diet. Recent reports show that a ketogenic diet may help to control spasms in children who don’t respond to other therapies like medication.

High-dose IV immunoglobulin: This form of treatment has been used to treat a number of seizure disorders and there is some evidence that it can be effective in treating cryptogenic (unknown cause) infantile spasms. Because of the limited available data in treating IS, this treatment option is usually considered only after other medical therapies have failed.

Surgery: When patients have failed medication treatment options and when a localized area of brain abnormality can be identified, cortical resection (removal of the brain abnormality) has the potential to control the seizures and improve developmental outcomes.



What the Child Neurology Foundation is Doing to Make a Difference

Founded in October 2000, the Child Neurology Foundation was created as the outreach and philanthropic arm of the Child Neurology Society. Members of the Child Neurology Society include more than 1,300 child neurologists from the U.S. and Canada, as well as more than 30 other countries around the world.

Mission of the Child Neurology Foundation

- Advocate for children and adolescents with neurologic and developmental disorders
- Fund neurologic research of young investigators
- Promote awareness of career opportunities in child neurology
- Provide public, professional, and patient education programs
- Support the activities and mission of the Child Neurology Society

Awards

Since 2001, the Child Neurology Foundation has awarded more than \$1 million in scholarships, research grants, and other awards to more than 50 medical professionals and students.

Research

As part of its mission, the Child Neurology Foundation is dedicated to furthering scientific research to develop better treatments and ultimately cures for childhood neurologic diseases. In 2009, CNF received funding for Infantile Spasms research. The inaugural “CNF 2010 Logan Infantile Spasms Research Award” has been awarded to Catherine Chu-Shore, Ph.D. of Massachusetts General Hospital, who will focus her research on the “Functional Network Connectivity” in Infantile Spasms.





Shannon and Rod Geoghegan – daughter Hannah diagnosed with IS at age 6 months – seen above at 1 year old

“Before she was diagnosed with infantile spasms, Hannah was babbling and making sounds. She lost her vocalization for awhile. At around 8 months old, post-treatment, she was able to sit unassisted and started laughing and making noises again. My greatest sadness is that there have been so many things that have happened in this past year - and I feel like we’ve missed out on enjoying each moment. I think we’re just starting to be able to learn to enjoy those little moments - we celebrate each and every thing Hannah does.”

-- Shannon and Rod Geoghegans on importance of being aware and early/aggressive diagnosis/treatment

Always consult your physician before selecting a treatment plan. All information provided by the Child Neurology Foundation on its website or in printed material is for informational and educational purposes only. Readers are encouraged to confirm the information contained herein with other sources. Patients and consumers should review the information carefully with their professional health care provider. Side effects and drug interactions are always a possibility. This information is not intended to replace medical advice offered by physicians.

Infantile Spasms Awareness Week (ISAW)

In 2009, the Child Neurology Foundation proudly announced the creation of Infantile Spasms Awareness Week. The goal of IS Awareness Week is to provide pediatricians, child neurologists, parents and caregivers with objective educational tools which will increase awareness and understanding of infantile spasms. Early and aggressive treatment can impact the prognosis of infants diagnosed with IS. This important educational initiative will help to ensure that physicians have an increased awareness and understanding of IS. It will also make much needed educational materials available to the parents and caregivers of afflicted infants.



“Infantile Spasms Awareness Week is a great project for many reasons. Most obviously, it shows the importance of the recognition of such a potentially devastating disorder. It also shows the fact that industry and the child neurology community can cooperate in a responsible and ethical way and advance the causes in which we both believe.”

Lawrence Brown, M.D.
Associate Professor of Neurology and Pediatrics
Children’s Hospital of Philadelphia

