

## UCLA Infantile Spasms Program addresses rare, potentially devastating form of epilepsy



**The UCLA Division of Pediatric Neurology** offers one of the nation's largest, most active programs of clinical care and research for a potentially devastating form of epilepsy called infantile spasms. If untreated, infantile spasms can leave children with lifelong developmental impairment, including severe intellectual disability.

Epilepsy is one of the most common neurological diseases, affecting about 1 percent of the U.S. population. By contrast, infantile spasms is relatively rare, with about 2,000 new cases reported annually. The condition tends to affect children under the age of 2 years. Unlike adult epilepsy, which is often marked by dramatic seizures, infantile spasms is characterized by clusters of often subtle seizures that each lasts just a few seconds. These seizures typically include sudden lifting of the arms or bending at the waist, and can be difficult to differentiate from normal infant behavior, complicating early detection.

This high-stakes disorder can cause irreversible devastation in as little as a month. If infantile spasms is suspected, UCLA pediatric neurologists recommend rapid confirmation of the diagnosis and targeted intervention.

### Research aims to better predict treatment outcomes

UCLA's Infantile Spasms Program has an ambitious, multi-faceted research agenda, encompassing four primary areas: comparisons of standardized drug protocols, improvements to EEG to better characterize infantile spasms and predict outcomes, characterization of the potential role of surgical options, and identification of genetic predictors for this neurological condition.

"One of our goals is to be able to predict who will respond to various therapies, so we can begin the most effective treatment promptly," says pediatric neurologist Shaun Hussain, MD, MS, director of the UCLA Infantile Spasms Program and assistant clinical professor of pediatrics. "Another goal is determining if some patients should avoid risky and expensive medical therapies in favor of potentially curative surgery."

UCLA specialists are committed to educating families about infantile spasms and to reaching out to pediatricians and family physicians, many of whom infrequently encounter the condition. "We want physicians to be keenly aware of infantile spasms," says Dr. Hussain, "and to refer children to us as early as possible to increase the chances of effective intervention."

## No single cause

There is no one cause for infantile spasms. Almost any injury to the brain or abnormality can be a causal factor. In one of the paradoxes of this condition, cases with no known cause — about 10 percent of those affected — seem to respond better to intervention. Depending on the underlying cause, normal development is possible if treatment is prompt and successful. In its most positive outcomes, the condition represents one of the few opportunities to cure epilepsy.

The UCLA Infantile Spasms Program employs the gold-standard diagnostic tool: video-electroencephalography or video-EEG. Infantile spasms has a particular EEG signature, a condition called hypsarrhythmia — chaotic, disorganized electrical brain activity with no recognizable pattern — which is present in most cases. Successful treatment must address both the spasms and hypsarrhythmia.

## Current treatments

First-line treatment for infantile spasms is usually hormonal therapy, including prednisolone and adrenocorticotropic hormone (ACTH), as well as the anti-convulsant vigabatrin. These treatments can produce serious adverse effects such as immune system suppression and high blood pressure with hormonal therapy, or visual (retinal) injury from vigabatrin. There is about a 75 percent response rate, with about two-thirds of responders staying spasm-free. As yet, doctors have no way of predicting which patients will respond to which medications or to surgical interventions to remove seizure-generating parts of the brain.

## UCLA program offers multiple strengths

As a top-tier center for infantile spasms, UCLA takes on the most difficult and complex cases. The program is distinguished by its size, active slate of clinical trials and comprehensive, multidisciplinary approach, drawing families from all over the world. Faculty members include specialists in developmental and metabolic storage diseases, epilepsy and EEG technology, along with experienced neurosurgeons in the Pediatric Epilepsy Surgery program. Indeed, doctors at UCLA pioneered surgical therapy for carefully selected patients with infantile spasms in the eighties, and since have introduced many technical advances. Doctors follow patients in the Infantile Spasms Clinic, which offers a full spectrum of proven medications, as well as the ketogenic diet, a high-fat, low-carbohydrate diet that may be helpful in the management of many different types of epilepsy.

Adding to these strengths is the work of UCLA's Neuroscience Genomics Core, which is at the forefront of next-generation genomics sequencing. A better understanding of the genetics of infantile spasms could result in more personalized therapies.

## Participating Physicians

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