

New treatment to reduce seizures among infants with Sturge-Weber syndrome

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Researchers at Kennedy Krieger Institute and Boston Children's Hospital have discovered a new way to decrease early seizure likelihood and improve neurologic outcomes among infants with Sturge-Weber

Syndrome (SWS). This collaboration is the first multi-center study of its kind focused on this rare condition.

Almost 90% of infants with SWS experience a seizure before the age of two, and these early seizures can contribute to brain injury and worse long-term outcomes. Researchers have found that [young patients](#) who were treated presymptomatically for SWS were significantly less likely to have early seizures.

Dr. Anne Comi, director of the Hunter Nelson Sturge-Weber Center at Kennedy Krieger and co-principal investigator on this project, says that this groundbreaking discovery will impact the quality of life for young SWS patients.

"By treating these infants ahead of time, we can lower those chances of experiencing early seizures by almost half," Dr. Comi said. "In our [study group](#), we reduced the percentage of patients diagnosed with early seizures from 88% to 53% and improved outcome at two years. This breakthrough shows that treating infants before they start showing major symptoms improves their quality of life."

SWS is a neurovascular disorder often characterized by a facial birthmark, called a port-wine birthmark, located on the upper face. The birthmark is typically an indicator that a patient is considered at high risk for abnormal vessels in the brain and possible seizures.

In addition to presymptomatic treatment, researchers also found that conducting a clinical electroencephalogram (EEG), a test that measures [electrical activity](#) in the brain, can predict seizure onset. Dr. Comi believes these results will change treatment approaches across the globe.

"Stopping the seizures ahead of time means we lower the risk of brain injuries." Dr. Comi said. "Infants with a facial port-wine birthmark

should be seen urgently by a neurologic specialist so that we can conduct a clinical EEG, evaluate brain activity, do proper MRI imaging, and develop a presymptomatic treatment plan."

The presymptomatic treatment involves administering a combination of anti-seizure medication and [low-dose aspirin](#). Previously, intervention did not begin until after patients were already experiencing seizures. However, by that point, infants often developed brain injuries due to venous strokes, leading to regression in cognitive development.

"SWS is a developmental disorder with many different causes, and there was an urgency to find an intervention to protect high risk babies in order to improve their outcomes," said Dr. Anna Pinto, Director of the Sturge Weber Clinic at Boston Children's Hospital and co-principal investigator on the project.

"Without [early intervention](#), there are worse outcomes," Dr. Comi said. "Seizures disrupt blood flow which causes cortical damage, leading to cognitive impairments in patients by school age. Using anti-seizure medication and low-dose aspirin addresses the [blood flow](#) issues and high risk of early seizures."

The next steps will be using the data collected to understand how to predict early seizure onset better. Dr. Yangming Ou, a faculty member in the Fetal-Neonatal NeuroImaging Data Science Center at Boston Children's and a co-investigator in this work, says that [artificial intelligence](#) (AI) will be a useful tool to treat SWS patients earlier.

"We have begun training AI tools using this data, collected from past clinical care of kids, to determine better predictors of first [seizure](#) onset and the impact of treating patients before symptoms appear," Dr. Ou said. "Our AI methods can help clinicians determine the best course of treatment for each individual patient with this rare disease."

Multiple centers around the country are now planning to use presymptomatic treatment among their young patients. Dr. Comi says the results are compelling and will help families who are impacted by SWS. Kennedy Krieger and Boston Children's Hospital are seeking patients who can benefit from this new learning to participate in next-level research.

"We are excited about these findings," Dr. Comi said. "Future research will need to focus on longer-term neuro-cognitive outcome after presymptomatic treatment and prediction of which children are most likely to respond well to this new clinical care approach."

Provided by Kennedy Krieger Institute

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