

Nemours Children's Health Researcher Receives Prestigious \$2M Federal Research Grant to Study Rare Genetic Condition Tied to Epilepsy and Autism

Research team will study the mechanisms underlying seizures and cognitive impairments in tuberous sclerosis complex

WILMINGTON, Del. (June 11, 2024) — Amanda Hernan, PhD, Head of the Dynamics in Epilepsy and Cognitive Development (DECODE) Lab at Nemours Children's Health, Delaware Valley has been awarded a five-year, \$2 million R01 research grant from the National Institutes of Health (NIH) to study tuberous sclerosis complex (TSC), a leading genetic cause of epilepsy and autism spectrum disorder.

Hernan's study, titled "*Leveraging genetically encoded heterogeneity to understand TANDs and seizures in novel models of TSC*," seeks to increase understanding of this disorder, in which noncancerous tumors compromise brain development and function. Up to 90% of patients diagnosed with TSC have epilepsy, which involves recurring seizures caused by abnormal electrical impulses in brain tissue. In addition, 70% also have TSC-associated neuropsychiatric disorders (TANDs), including autism spectrum disorder, attention deficit hyperactivity disorder and learning difficulties.

Hernan and her team are focused on the association between epilepsy and cognitive impairment in TSC. They anticipate that understanding ways in which TSC causes seizures and cognitive impairment could help researchers develop and identify better treatment options.

"TSC is a heterogeneous condition, therefore, even within a single family, the same mutation in one of the genes that causes TSC can produce different outcomes from one family member to another," Hernan explained. "In this grant specifically, we're utilizing this heterogeneity to help us understand the neural network mechanism underlying the development of seizures and TANDs."

Hernan joined Nemours Children's Health in 2021. In 2017, as a postdoctoral fellow at the University of Vermont, Hernan received an NIH K22 Transition to Independence award. She has also received a Delaware IDeA Network of Biomedical Research Excellence award and a generous donation from the Holt Foundation to support her research efforts.

"Translational research is the fundamental backbone upon which clinical care is based. Research at Nemours Children's is crucial for driving innovation and advancements in medicine that ultimately lead to improved outcomes and quality of life for our patients," said Hernan. "This NIH funding and support for the amazing DECODE lab team is critical to advancing our science that is dedicated to enhancing the health of children living with TSC."

About Nemours Children's Health

Nemours Children's Health is one of the nation's largest multistate pediatric health systems, which includes two freestanding children's hospitals and a network of more than 70 primary and specialty care practices. Nemours Children's seeks to transform the health of children by adopting a holistic health model that utilizes innovative, safe, and high-quality care, while also addressing children's needs well beyond medicine. In producing the highly acclaimed, award-winning pediatric medicine podcast Well Beyond Medicine, Nemours underscores that commitment by featuring the people, programs and partnerships addressing whole child health. Nemours Children's also powers the world's most-visited website for information on the health of children and teens, Nemours KidsHealth.org.

The Nemours Foundation, established through the legacy and philanthropy of Alfred I. duPont, provides pediatric clinical care, research, education, advocacy, and prevention programs to the children, families and communities it serves. For more information, visit [Nemours.org](https://nemours.org).

<https://nemours.mediaroom.com/2024-06-11-Nemours-Childrens-Health-Researcher-Receives-Prestigious-2M-Federal-Research-Grant-to-Study-Rare-Genetic-Condition-Tied-to-Epilepsy-and-Autism>