

# EPILEPSY

# NEUROGENETICS

# INITIATIVE

Advancing Diagnosis  
and Care Through  
Genetic Testing



Epilepsy is a common brain disease defined by the presence of seizures. For many children, the cause of epilepsy is genetic. Until recently, little has been known about the genes that cause the condition, so treatment has been imprecise and not targeted toward the underlying cause. Many families spend years searching for answers to alleviate their child's suffering. At Children's Hospital of Philadelphia, we are dedicated to improving care for children with epilepsy. Thanks to recent rapid advances in our understanding of the role genetic variants play in epilepsy and the development of genetic testing technologies, we can now identify the genes causing a child's epilepsy, which can put an end to a family's search for answers.

## WHY WE'RE DIFFERENT

Our Epilepsy Neurogenetics Initiative (ENGIN) is a unique service that exists within one of the largest neurogenetics programs in the world. We pair cutting-edge clinical care and genetic testing to identify the genetic variants causing a child's epilepsy, with innovative research aimed at the development of precision approaches to treatment.

### We offer:

- Unparalleled access to genetic testing
- Extensive team of neurogeneticists, genetic counselors, pediatric epilepsy experts, a neurosurgeon and therapists
- Individualized treatment plan
- Access to the comprehensive care necessary to manage the full range of issues associated with epilepsy

## WHAT WE OFFER

### Establishing a genetic diagnosis can:

- Provide a family answers as to why their child developed epilepsy, the disease prognosis and risk of recurrence in other family members
- Inform treatment and management
- Allow the family to connect with other families of children with the same diagnosis
- Enable research to better understand disease mechanisms and develop precision treatments
- Reduce or eliminate the need for additional invasive diagnostic testing

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## WE LEAVE NO STONE UNTURNED

The Epilepsy Neurogenetics Initiative is unique in that we integrate genetics into the diagnosis and management of each child with epilepsy. Our comprehensive evaluation includes:

- Genetic testing, which can include:
  - Chromosomal microarray analysis
  - Epilepsy panel
  - Whole exome sequencing
- Physical and neurological examination
- Review of medical and family history
- Neurosurgical evaluation for resective brain surgery or implantation of neuromodulatory devices
- Evaluation for dietary therapy
- Genetic counseling
- Physical and occupational therapy evaluation

Based on the results of the evaluation, our clinical and research teams work with referring physicians and families to create a personalized care plan. We offer a full range of epilepsy therapies, from medication and dietary treatment to epilepsy surgery. All patients are offered the opportunity to enroll in our research studies, which help us learn more about the child's condition and refine their treatment, and will help us develop precision treatments that benefit children around the world.

## TREATING ALL IN SEARCH OF ANSWERS

Families come to us from all over the country and the world due to our unparalleled access to genetic tests and our efforts to translate discoveries into effective treatments. Any child with epilepsy may be eligible for referral to our program.

We partner with families, primary care providers and referrers to provide the best care available.

*Children's Hospital of Philadelphia recognizes the importance of this field of discovery and what it can do for children with epilepsy and their families. In an effort to continually elevate care, we have made a significant investment in our Neurogenetics and Epilepsy programs.*



### PARTNER WITH US

To refer a patient or request a second opinion:  
267-426-6298  
[CHOPUSA@email.chop.edu](mailto:CHOPUSA@email.chop.edu)

**LEARN MORE**  
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