Genetic Epilepsy Clinic

By Medical Director, M. Scott Perry

New technology and better understanding of the genetic origin of epilepsy is providing unprecedented insight into the disorder. Testing may now reveal causes of epilepsy in previously undiagnosed patients, resulting in improved treatment and a more precise prognosis. New genetic syndromes are increasingly discovered and providers will more frequently encounter genetic mutations of "unknown significance." These mutations require an expertise in both genetics and neurology to understand the implication of the test results on the care of the patient.

In response to this need, Cook Children's created the Genetic Epilepsy Clinic, staffed by pediatric epileptologists, M. Scott Perry M.D. and Dave Shahani M.D., along with metabolic geneticist, Alice Basinger, M.D. Our team can review the child's history, review existing testing, recommend additional evaluation, establish diagnosis, provide genetic counseling and help establish a treatment plan specifically designed to address the syndrome unique to the patient.

Diagnosing genetic epilepsy early in a child's life may limit unnecessary invasive testing, help avoid treatments which may worsen or aggravate the child's epilepsy and can ultimately lead to an improved outcome.

The Genetic Epilepsy Clinic helps children:

- With epilepsies caused by genetic mutations.
- Who had confusing/unclear test results for genetic epilepsy syndromes.
- Who fit the phenotypes of well-known genetic epilepsy, but whose testing was negative.

Common genetic epilepsy syndromes include those associated with:

- **Sodium channel disorders (Dravet syndrome, GEFS+)** ~ Sodium channel disorders represent a spectrum of epilepsy syndromes ranging from intermittent febrile seizures to more severe phenotypes such as Dravet syndrome. For some of these disorders (i.e. Dravet syndrome), antiepileptic drugs with sodium channel mechanisms may aggravate seizures, while other syndromes (i.e. SCN8a) may benefit from these drugs. Early diagnosis may prevent unnecessary testing and guide treatment to avoid medications which exacerbate these conditions.
- **Potassium channel disorders (KCNQ2/3)** ~ Potassium channel disorders often present with seizures in the infant's first days of life. In some phenotypes, the seizures can stop in infancy and are relatively benign; others can have seizures which persist into childhood and result in significant cognitive impairment. Early diagnosis may prevent unnecessary testing and guide treatment with medications specific to potassium channel dysfunction.
- **PCDH19** ~ Epilepsy associated with this gene resembles Dravet syndrome, but presents primarily in females.
- **Glut-1** ~ Glucose-transporter mutations result in a spectrum of epilepsy syndromes ranging from early onset of intractable seizures of multiple types, cognitive delay and microcephaly to milder phenotypes of early onset absence seizures (onset age < 4 years). Early diagnosis can improve treatment, as the ketogenic diet is a recognized therapy of choice for these patients.

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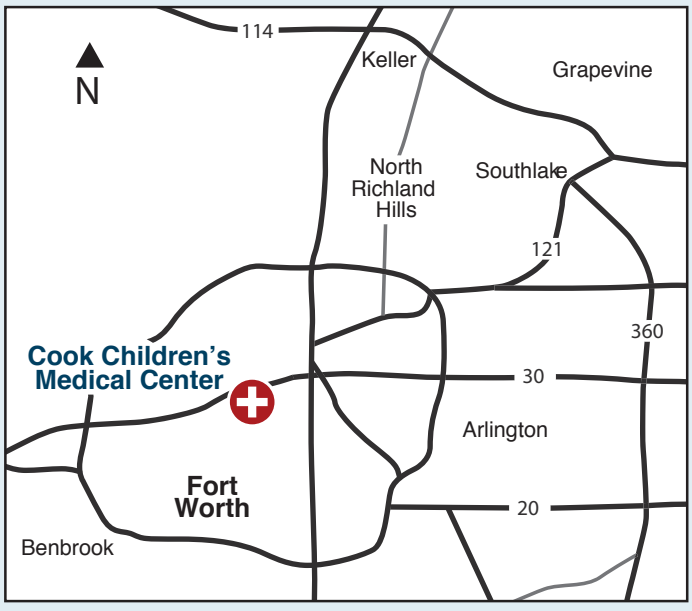
For referrals to the either the Genetic Epilepsy Clnic or Adult Genetic Epilepsy clinic call:
682-885-2500

Why choose Cook Children's Genetic Epilepsy Clinic for your patients?

We provide some of the nation's most advanced, child-friendly care by having:

A family-centered approach to diagnosis and treatment of epilepsy, reviewing all risks and benefits of care prior to making a final treatment decision.

- Specialists who work together in the exam and operating rooms.
- Experienced epileptologists and geneticists who identify the seizure type and create a unique treatment plan for each patient.
- Access to the latest therapies and research for genetic epilepsy syndromes.
- Comprehensive plan of care throughout life with transition to our Adult Genetic Epilepsy Clinic led by Hamid Kadiwala M.D.



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Case study:

A 2-year-old presented with status epilepticus characterized by hemiclonic seizure semiology. The family reported a history of multiple bouts of complex febrile seizures or febrile status and the patient had a delay in speech and motor development. An electroencephalogram (EEG) demonstrated generalized spike wave discharges and the patient's prior MRI was normal. Prior to presentation, she had been treated with oxcarbazepine for her unprovoked focal seizures.

SCN1A testing was ordered based on her clinical history, revealing a disease-causing mutation consistent with a diagnosis of Dravet syndrome. As a result, oxcarbazepine was discontinued, as it is known to exacerbate seizures in this syndrome. Valproate was initiated and the family was provided a rescue plan in hopes of preventing future bouts of status epilepticus.

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