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Courtagen Offers Wide Array of Genetic Testing Options for Epilepsy

November is Epilepsy Awareness Month

WOBURN, MA – November 8, 2016 – Courtagen Life Sciences, Inc., an innovative molecular information company, focused on providing genetic diagnoses for neurological conditions through next generation sequencing, offers a broad range of genetic testing panels for epilepsy, including recently-added epiSEEK® Spotlight Panels, designed to target genes that have been reported in association with specific phenotypes for epilepsy and seizure disorders.

Epilepsy is Common

Epilepsy, or seizure disorders, are some of the most common neurological problems. It is estimated that 1 in 62 people in the United States will develop epilepsy at some point in their lifetime, with the highest incidence in children. Despite the significant development of anti-seizure medications, with over 30 medications available currently, as well as non-medication options, approximately one-third of patients with newly diagnosed epilepsy cases will become refractory to treatment.

The Burden and Costs

Children with a current diagnosis of epilepsy are significantly more likely than those never diagnosed to experience comorbidities such as developmental delay, autism, ADHD, behavior problems, headaches, anxiety, and depression. The main cost drivers for the care and management of children with epilepsy are hospital admissions, emergency room visits, anti-epileptic drugs (AEDs), and therapies to address comorbidities.

The Role and Utility of Genetics

Research over the past two decades has highlighted the importance of genetics in the etiology of many epilepsies. Many forms of epilepsy, and especially those with onset in infancy and childhood, are strongly influenced by genetics. Therefore, neurologists routinely consider genetic testing in the diagnostic workup of epilepsy syndromes and seizure disorders.

Knowing the genetic basis of a patient's epilepsy is valuable for obtaining a definitive diagnosis, estimating prognosis, determining recurrence risks, and guiding treatment choices. It may also give information about other associated neurologic or medical conditions that may arise over time. In many cases, the precise genetic diagnosis can be important in therapy selection, particularly when there are known contraindications or recommended treatment options based on genetic results. For example, in the case of glucose transporter type 1 (Glut-1) deficiency syndrome due to *SLC2A1* mutations, treatment with the ketogenic diet (and not anti-epileptic drugs) can prevent seizure activity and, if

implemented early, can protect the brain and prevent cognitive impairment, ultimately reducing the total cost of medical care for that child.

"With the rapid advancement in genetic knowledge, a patient's DNA can be the next important piece in unlocking access to better seizure control," says Dr. Minh Le, Pediatric Neurologist and Courtagen Medical Consultant. "Genetic testing can help a physician to make an accurate diagnosis early in the disease course, anticipate and hence prevent comorbidities, and select the most appropriate treatment based on the underlying etiology where proper early treatment could save a child's brain development and life."

Courtagen Genetic Testing Panels for Epilepsy
Identifying the best gene or genes to test is complex as there is considerable overlap in symptoms associated with the genes, making a tiered approach impractical. Next generation sequencing allows for concurrent analysis of multiple genes that have been associated with epilepsy and seizures in the most cost and time efficient manner. Courtagen offers a wide range of testing options, designed and interpreted by an expert clinical genetics team. Results are delivered in a concise clinical report to help determine diagnosis and assist with decisions about treatment and disease management.

- **epiSEEK® Spotlight Panels**: epiSEEK® Spotlight Panels: Fourteen tests that cover a range of seizure disorders, including Absence Seizures, Brain Malformation Seizures, Early Infantile Epileptic Encephalopathy (EIEE), Fever Sensitive Seizures, Focal Seizures, Infantile Spasms, Myoclonic Seizures, and Neonatal Seizures.
- **epiSEEK® Focus**: Designed to target the most common disease associated genes related to epileptic and seizure disorders.
- **epiSEEK® Comprehensive**: Designed to provide a thorough view, the epiSEEK Comprehensive panel enables genetic analysis and clinical interpretation of a wide range of genes associated with neurological function, and epileptic and seizure disorder phenotypes.
- **epiSEEK® Triome™**: Designed to provide the most complete picture, the epiSEEK Triome panel uses powerful trio analysis, by fully sequencing the parents' samples along with the patient, to enable extensive genetic analysis and clinical interpretation of an expanded selection of genes associated with neurological function, and epileptic and seizure disorder phenotypes.

With this a broad array of test offerings, Courtagen is able to provide clinicians, patients, and payers with even higher-quality information on genetic changes that have been shown to influence epilepsy.

To learn more about genetics and epilepsy visit http://info.courtagen.com/epilepsy-genetics/, or follow Courtagen on Facebook, Twitter, and Instagram.

About Courtagen Life Sciences, Inc.
Courtagen Life Sciences, Inc., located in Woburn, MA, is a CLIA/CAP certified molecular information company focused on the diagnosis of a range of neurological disorders. Courtagen operates a highly sophisticated Next Generation DNA Sequencing, bioinformatics, and clinical interpretation business. Courtagen's work helps physicians elucidate the linkages between the genotypes and phenotypes of various neurological diseases. For more information, please visit http://www.courtagen.com/

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