



Next-Generation Gene Sequencing from Athena Diagnostics Streamlines Epilepsy Diagnosis

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Quest Diagnostics' specialty neurology business unveils 10 new epilepsy diagnostic services, including industry's first clinical multi-analyte test for autoimmune epilepsy
Test services to be unveiled at the AAN Annual Meeting, April 26 - May 4 in Philadelphia

MADISON, N.J., April 23, 2014 /PRNewswire/ -- Quest Diagnostics (NYSE: DGX), the world's leading provider of diagnostic information services, today announced a suite of new testing services from its Athena Diagnostics specialty neurology business designed to help physicians more reliably and quickly diagnose the cause of several forms of epilepsy in adults and children. The new lab-developed tests, based on next-generation gene sequencing and neuroimmunological testing, include the first clinically available multi-analyte test panel to analyze five antibodies associated with autoimmune epilepsy, which accounts for up to 10 percent of epilepsy cases.



Epilepsy is a group of neurological disorders often characterized by seizures and cognitive impairment due to electrical disturbances in the brain. While brain tumors, trauma or infection can cause epilepsy, the condition can be inherited genetically within families. Some forms of epilepsy may be treated with antiepileptic pharmacotherapy or surgical procedures, but the type and efficacy of treatment depends on the specific epileptic disorder, among other factors. Up to 2.3 million adults in the U.S. have epilepsy and nearly 150,000 develop the condition each year, according to estimates from the U.S. Centers for Disease Control and Prevention.

Athena Diagnostics will unveil the new services during the American Academy of Neurology Annual Meeting, April 26 - May 4, in Philadelphia (Exhibit Number 813).

Next-Generation Gene Sequencing Aids Clinical Practice

"One of the great challenges in medicine today is how to transform mass quantities of genetic sequencing data into clinically relevant information that enlightens, rather than confuses, diagnosis," said Jay Wohlgenuth, M.D., senior vice president, science and innovation, Quest Diagnostics. "Athena's new epilepsy services exemplify how we are taking that challenge head on, by creating services that support the real-world clinical approach physicians take to evaluate, diagnose and treat patients across a continuum of care."

Physicians typically diagnose the type of epilepsy based on a comprehensive medical exam that includes assessing signs and symptoms and results of neuroimaging and electroencephalography (EEG) tests. In recent years, next-generation gene sequencing has enabled researchers to sequence the entire human genome to identify pathogenic genetic variants that cause genetic forms of the disorder. Most clinically available diagnostic tests perform sequencing on hundreds of genes and provide test results reaching dozens of possible disease-causing variants. The physician must then review and interpret this data for clinical significance for the individual patient.

To address this limitation, Athena has developed nine sequencing test panels according to epilepsy type to aid the diagnosis of discrete forms of epilepsy ranging from generalized to syndromic. The panels, called Epilepsy Advanced Sequencing Evaluations, are arranged by clinical phenotype and EEG test findings, with testing performed only on genes associated with a specific epilepsy type. Physicians may therefore select a panel and receive a test report with clinically useful sequencing information to diagnose epilepsy at the molecular level. This approach may avoid the excessive use of other neurological testing, including imaging studies of the brain.

"The cause of epilepsy is unknown in more than a third of individuals, and current diagnostic approaches can provide unclear results. By focusing on the neurological presentation and EEG findings, our test services differ from offerings that provide a plethora of sequencing data without a larger context for evaluating the patient," said Joseph J. Higgins, MD, medical director, neurology, Athena Diagnostics. "A neurogenetic diagnosis by molecular blood testing may help reduce the likelihood of laborious, stressful and expensive diagnostic procedures, and, in some cases, help the physician to select effective antiepileptic pharmacotherapy or other treatments."

Athena Diagnostics also analyzes variants of unknown significance based on its experience in neurogenetic testing to further enhance genetic test interpretation for the physician. "We use novel prediction tools to reliably assign pathogenicity scores to variants to help physicians properly interpret genetic testing results," added Dr. Higgins.

Autoimmune Epilepsy – A Disorder That Can Elude Reliable Diagnosis

A study published in the March 2014 edition of JAMA Neurology found that autoimmune disorders were associated with elevated risk of epilepsy, and that "patients with either condition should undergo surveillance for the other." Autoimmune disorders include type 1 diabetes mellitus, rheumatoid

arthritis and Crohn's disease.

Epilepsy caused by autoimmune factors may respond to a combination of immunotherapy and antiepileptic drugs, but the condition is often mistaken for psychiatric and other cognitive disorders. Current diagnostic techniques, including MRI, cerebrospinal fluid and brain biopsy, often provide unclear diagnostic information.

The Athena Diagnostics Autoimmune Epilepsy Evaluation lab-developed test identifies antibodies directed against neuronal cell-surface antigens, such as NMDAR, VGKC-complex, LGI1 and CASPR2 as well as neuronal GAD65. It is the first clinically available multi-analyte test in the United States to aid the diagnosis of autoimmune epilepsy.

"A growing body of literature demonstrates an autoimmune basis in the etiology of some forms of epilepsy, and that standard therapies alone may not be sufficient for treating this disorder," said Dr. Higgins. "Our multi-analyte panel will enable the physician to streamline the evaluation of a patient based on the presence of antibodies well correlated in the literature with autoimmune epilepsy. With this information, a physician may arrive at a reliable diagnosis more quickly, and initiate immunotherapy as well as traditional anticonvulsant therapy."

"A reliable diagnosis and early treatment plan is of obvious benefit to the patient," said Dr. Higgins. "But it also affords significant potential cost savings for the health care system."

All of the new epilepsy tests are available nationally, with the exception of New York, where state approval is expected within the next six weeks.

About Athena Diagnostics

Athena Diagnostics, a business of Quest Diagnostics, is a leader in diagnostic testing for neurological diseases and offers innovative tests for Alzheimer's disease, muscular dystrophy and other neuromuscular and developmental disorders. Athena is dedicated to providing neurologists and other physicians and specialists with insights that can improve patient health. For more information, visit AthenaDiagnostics.com.

About Quest Diagnostics

Quest Diagnostics is the world's leading provider of diagnostic information services that patients and doctors need to make better healthcare decisions. The company offers the broadest access to diagnostic information services through its network of laboratories and patient service centers, and provides interpretive consultation through its extensive medical and scientific staff. Quest Diagnostics is a pioneer in developing innovative diagnostic tests and advanced healthcare information technology solutions that help improve patient care. For more information, visit QuestDiagnostics.com. Follow us at [Facebook.com/QuestDiagnostics](https://www.facebook.com/QuestDiagnostics) and [Twitter.com/QuestDX](https://twitter.com/QuestDX).

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