GeneDx Presents New Data at the American Epilepsy Society Annual Meeting Demonstrating That Exome Sequencing Outperforms Multi-Gene Panels for Patients with Epilepsy

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New research from one of the largest exome sequencing studies of patients with epilepsy adds to the mounting evidence that exome sequencing offers superior clinical results compared to multi-gene panels

STAMFORD, Conn., Dec. 01, 2023 (GLOBE NEWSWIRE) -- GeneDx (Nasdaq: WGS), a leader in delivering improved health outcomes through genomic and clinical insights, today announced new data to be presented at the American Epilepsy Society (AES) annual meeting in Orlando, Florida, December 1-5, 2023, that supports the use of exome sequencing as a first-tier test for patients with epilepsy.

In a new study of 22,500 individuals with a clinical history of seizures or suspected seizures, researchers at GeneDx found that:

- Nearly 25% of this patient cohort received a genetic diagnosis through exome sequencing. Previously published literature cites the diagnostic rate of multi-gene panels for patients with epilepsy at 19%.[1]
- Notably, 78% of patients in this exome positive cohort had prior genetic testing, suggesting that the previous tests did not resolve all clinical questions.
- The gap in diagnosis between exome sequencing and the multi-gene epilepsy panels can be largely attributed to the fact that more than half (57%) of seizure-related genes are not included on many commercially available epilepsy panels.
- Of those patients with a positive genetic diagnosis from exome sequencing, 11% had a variant in a treatment related gene.

These data were collected between 2019 and 2022. In 2022, guidelines were published by the National Society of Genetic Counseling (NSGC) and endorsed by AES, recommending exome as a first-tier test for individuals with unexplained epilepsy.

GeneDx will also present data at the conference that shows insurance may offer better coverage for exome sequencing than multi-gene panels for patients with epilepsy. The review of publicly available coverage policies from 12 national payers, regional payers, state Medicaid programs, and laboratory benefit managers, revealed that for outpatient individuals with epilepsy, broader payer coverage exists for exome sequencing than for multi-gene panels.

“The combination of these studies paints a striking picture for the use of exome sequencing for patients with epilepsy,” said Paul Kruszka, Chief Medical Officer at GeneDx. “Between expanding payer coverage, the diagnostic power of exome sequencing, and the clinical guidelines, providers should feel confident in offering exome sequencing as a first-tier test to their patients. The time is now for exome sequencing to be embraced as the standard of care for this patient population.”

Epilepsy impacts approximately 1.2% of the US population, with just under a half a million cases in pediatric patients.[2] Nearly 50% of unexplained epilepsy cases likely have a genetic cause.[1] Identifying the specific disease-causing variant in epilepsy patients enables a more precise treatment plan, including avoiding, stopping, or initiating specific medication or diet recommendations, and can qualify patients for clinical trials.

At the conference, GeneDx will also be hosting an industry sponsored symposium discussing the ways in which exome sequencing can help epilepsy patients find answers sooner. In that panel discussion:

- Dr. Heather Mefford, MD, PhD, FACMG, of the Center for Pediatric Neurological Disease Research at St. Jude’s Children’s Research Hospital, will review the current guidelines, which support exome sequencing as a first-tier test for patients with epilepsy, and describe the clinical utility for patient care and management.
- Parents of three individuals with epilepsy will share their lived experiences with genetic testing, including the psychological benefits of having a precise diagnosis, having a clearer understanding of how their child’s symptoms may change as they age, and the sense of belonging and community a genetic diagnosis can provide.

Full 2023 AES Conference participation:

- **Platform Session Clinical Research/Poster 1.204**: Exome-Based Testing for Patients with Seizures: Advantages over Panel-Based Testing (Michelle M Morrow, Kirsty McWalter, Elizabeth Butler, Lindsay Havens, Melanie P Napier, Karen E Wain, Paul Kruszka)
- **Poster 2.482**: Payer Coverage for Epilepsy: Broader Coverage for Exome Sequencing Compared to Multigene Panels and Genome Sequencing (Sarah Soto, Stephanie Gandomi, Jane Juusola)
- **Corporate Satellite Symposium:** The Clinical and Personal Value of a Genetic Diagnosis for Epilepsy: Patient and Caregiver Perspectives

About GeneDx

GeneDx (Nasdaq: WGS) delivers personalized and actionable health insights to inform diagnosis, direct treatment, and improve drug discovery. The company is uniquely positioned to accelerate the use of genomic and large-scale clinical information to enable precision medicine as the standard of care. GeneDx is at the forefront of transforming healthcare through its industry-leading exome and genome testing and interpretation, fueled by one of the world's largest rare disease data sets. For more information, please visit GeneDx.com and connect with us on LinkedIn, Facebook, Twitter, and Instagram.

Investor Relations Contact:
Investors@GeneDx.com

Media Contact:
Press@GeneDx.com

References

1. Sheidley BR, Malinowski J, Bergner AL, *et al.* Genetic Testing for the Epilepsies: A Systematic Review. *Epilepsia*. 2022 Feb;63(2):375-387. doi: 10.1111/epi.17141. Epub 2021 Dec 10.
2. Zack MM, Kobau R. National and State Estimates of the Numbers of Adults and Children with Active Epilepsy — United States, 2015*MMWR*. 2017;66:821–825. DOI: 10.15585/mmwr.mm6631a1