GeneDx to Expand Access to Exome Testing for Pediatric Epilepsy Patients with New Partners

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STAMFORD, Conn.--(BUSINESS WIRE)--Dec. 5, 2024-- GeneDx (Nasdaq: WGS), a leader in delivering improved health outcomes through genomic insights, today announced Biogen (Nasdaq: BIIB), Praxis Precision Medicines (Nasdaq: PRAX) and Stoke Therapeutics (Nasdaq: STOK) as the founding partners to its Patient Access Program for pediatric epilepsy, which provides access to whole exome sequencing.

Currently, access to a definitive genetic diagnosis in pediatric epilepsy remains limited, as evidenced by the 5–8-year diagnostic odyssey that many children face.[1] While patients may receive targeted multi-gene panel testing, there are more than 700 different genes related to seizures, and less than 50% of those genes are included on most commercially available gene panels.[2] This program increases access to exome testing, increasing the chance of receiving a definitive diagnosis. Nearly 25% of patients with seizures received a genetic diagnosis through exome testing, compared to a previously reported 19% diagnostic yield for epilepsy gene panels.[3]

The Patient Access Program helps to ensure more equitable care across pediatric epilepsy populations. Exome testing is recommended as a first-line test for patients with unexplained epilepsy by the National Society of Genetic Counselors and these guidelines are endorsed by the American Epilepsy Society.[4] In addition, research shows that for those with a genetic diagnosis, the knowledge has implications for treatment and management in up to 80% of people.[5] However, despite the overwhelming clinical support and guidelines for testing, access to testing is sparse, racial disparities exist due to lack of access, and the journey to obtain a genetic diagnosis for rare disorders, including epilepsy, can take years. The Patient Access Program aims to address these challenges by increasing access to whole exome sequencing for pediatric epilepsy patients.

In addition to helping patients receive a genetic diagnosis, the insights generated from testing will contribute to GeneDx's industry leading rare disease data set. GeneDx has robust de-identified data from more than 700,000 exome and genome results that can help researchers better understand gene-disease relationships for patients with seizures.

"While epilepsy is a fairly common condition, affecting nearly a half a million children under age 18 in the US, its genetic origins are still insufficiently understood. Through increased access to exome testing more patients may not only get potential answers for their symptoms, but it may also allow the possibility of personalized treatments and therapies in the future," said Melanie Duquette, Chief Growth Officer of GeneDx. "GeneDx has the unique ability to deliver answers to patients to improve their health, simultaneously unlocking insights for biopharma companies who are investing to develop potential therapies to treat similarly-situated patients, all while adding a deeper understanding of gene-disease relationships to our already robust database."

"At Praxis, we are proud to lead with the largest epilepsy-focused portfolio in the industry, which includes groundbreaking therapies like relutrigine," said Steven Petrou, Chief Scientific Officer and co-founder of Praxis Precision Medicines. "The success of the EMBOLD study and the ongoing work in our EMBRAVE study underscore the transformative potential of our Cerebrum and Solidus platforms to accelerate drug discovery and development for patients with severe epilepsy. By combining these advances with the GeneDx Patient Access Program, we are not only refining the understanding of epilepsy's genetic underpinnings but also enhancing trial recruitment and speeding the delivery of innovative treatments to patients who need them most. It's an exciting time as we push the boundaries of what's possible for these families."

"A proper genetic diagnosis is a critical first step in getting patients with epilepsy the care and treatment they need," said Barry Ticho, M.D., Ph.D., Chief Medical Officer of Stoke Therapeutics. "As our understanding of the genetic causes of epilepsy continues to increase, we are unlocking the potential for new genetically targeted treatments that address the underlying cause of the disease rather than only the symptoms. We are pleased to be partnering with GeneDx to provide greater equity in the availability of testing and to work together toward a common goal of improving outcomes for patients."

To be eligible for the Patient Access Program, epilepsy patients and their providers must meet certain criteria, including the following:

- Patient must be less than 18 years of age and reside in the United States
- Patient must have experienced their first unprovoked seizure under 8 years of age
- Patient must not have had prior genetic testing performed by a clinical laboratory that confirmed a diagnosis of a neurodevelopmental disorder (NDD)
- Ordering provider must be authorized under applicable law to order genetic testing in the United States

To learn more, visit genedx.com/epilepsy.

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 services, fueled by one of the world's largest, rare disease data sets. For more information, please visit www.genedx.com and connect with us on LinkedIn, Facebook, and Instagram.

References

1. Marwaha S, Knowles JW, and Ashley EA. A guide for the diagnosis of rare and undiagnosed disease: beyond the exome. Genome Med. 2022 Feb 28;14(1):23. doi: 10.1186/s13073-022-01026-w.

2. Butler L, et al. Exome-based testing for patients with seizures: Advantages over panel-based testing. Poster presented at American Epilepsy Society Annual Meeting; December 2, 2023; Orlando, FL.

3. Smith L, Malinowski J, Ceuleman S, *et al.* Genetic testing and counseling for the unexplained epilepsies: An evidence-based practice guideline of the National Society of Genetic Counselors. *J Genet Couns.* 2023 Apr;32(2):266-280. doi: 10.1002/jgc4.1646.

4. 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.

5. Marwaha S, Knowles JW, and Ashley EA. A guide for the diagnosis of rare and undiagnosed disease. *Genome Med.* 2022 Feb 28;14(1):23. doi: 10.1186/s13073-022-01026-w.

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