GeneDx Introduces New Telehealth Pathway to Expand Access to Exome Testing for Parents Seeking Answers

January 9, 2025

STAMFORD, Conn.--(BUSINESS WIRE)--Jan. 9, 2025-- GeneDx (Nasdaq: WGS), a leader in delivering improved health outcomes through genomic insights, today announced a new way to provide genetic counseling and test-ordering services for patients and families seeking access to genetic testing. The new telehealth testing pathway aims to shorten the diagnostic odyssey by connecting parents directly with genetic experts, increasing access to exome and genome testing.

Despite mounting clinical evidence for the benefits of exome testing in delivering a definitive diagnosis for rare disease patients, many individuals face barriers to access comprehensive genetic testing. The average time to an accurate diagnosis remains approximately 4-5 years, and in some populations, it can extend up to a decade. [1] At the same time, a child with neurodevelopmental disorders is likely to accrue over $10,000 in additional health costs and undergo more than 5 uninformative tests. [2] Every day that a child with a rare disease waits for a genetic diagnosis, their families lose the opportunity for disease-specific care plans, better resources, and the positive psychological impacts of improved understanding and support. Currently, the system presents significant challenges for parents seeking answers for their children's unexplained symptoms, with wait times for medical genetics and developmental/behavioral pediatricians increasing significantly over the past three years.[3]

"GeneDx understands that a genetic diagnosis can be life-changing for families, offering crucial insights that inform personalized care and treatment, which can lead to significantly improved health outcomes, including gene therapies and proactive condition management," said Dr. Paul Kruszka, MD, FACMG, Chief Medical Officer at GeneDx. "GeneDx is steadfast in our commitment to increasing access to exome and genome testing by responsibly removing the systematic barriers that have previously prevented patients and families from accessing this critical testing."

Too many children experience symptoms without a clear answer as to why. Meanwhile, genetic tests exist that may provide the answers these families desperately need. To bridge that gap, GeneDx is working with a leading telehealth genetics provider, to enable an easier way for parents to access actionable, accurate genetic testing for their child by streamlining the referral process to better support patients and families in need of exome testing. Parents can schedule testing, appointments, and receive a diagnosis in as soon as five weeks by visiting GeneDx.com/get-exome.

GeneDx's commitment to expanding access to testing extends beyond enabling new channels and focuses on ensuring all patients have equitable access to care. Recent findings highlight that racial disparities in delivering precise genetic diagnoses are due to limited access to testing, not differences in diagnostic yield across diverse populations. By expanding access to GeneDx's exome and genome testing, more patients can benefit from the genomic insights of over 700,000 exomes and genomes, increasing the likelihood of obtaining definitive diagnoses.

About GeneDx

At GeneDx (Nasdaq: WGS), we believe that everyone deserves personalized, targeted medical care—and that it all begins with a genetic diagnosis. Fueled by one of the world's largest rare disease data sets, our industry-leading exome and genome tests translate complex genomic data into clinical answers that unlock personalized health plans, accelerate drug discovery, and improve health system efficiencies. For more information, please visit genedx.com and connect with us on LinkedIn, X, Facebook, and Instagram.

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2. Soden SE, Saunders CJ, Willig LK, et al. Effectiveness of exome and genome sequencing guided by acuity of illness for diagnosis of neurodevelopmental disorders. Sci Transl Med. 2014 Dec 3;6(265):265ra168. doi: 10.1126/scitranslmed.3010076.
3. Klima, T. (2023). Access to Pediatric Specialty Care in California: Results of The Children's Specialty Care Coalition 2022 Member Survey (pp. 1–11). California: Practical Research Solutions.

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