GeneDx Research Demonstrates Medical Actionability of Exome and Genome Sequencing Results in Large Real-World Study of More Than 300,000 Patients

November 10, 2026

Findings presented at NSGC 2026 show 62% of patients with pathogenic or likely pathogenic findings had results associated with opportunities to inform medical management

GAITHERSBURG, Md.--(BUSINESS WIRE)--Nov. 10, 2026-- GeneDx, the leader in rare disease diagnosis and improving health through the power of genomic data, today announced new research to be presented at the National Society of Genetic Counselors (NSGC) 2026 Annual Conference, November 11–14. Across four scientific posters and three educational sessions, GeneDx will highlight how genomic data, advanced technology and clinical expertise are helping translate genetic diagnoses into more informed patient care.

New research analyzing exome and genome sequencing results from more than 300,000 patients found that 62% of patients with pathogenic or likely pathogenic findings (62,459 of 101,390) had results associated with medical actionability, including opportunities to inform treatment, disease surveillance, acute management and long-term care. These findings demonstrate the potential impact of genomic sequencing to guide clinical decisions and inform care beyond the initial diagnosis.

“A genetic diagnosis has the power to change the course of care,” said Linda Genen, MD, MPH, Chief Medical Officer of GeneDx. “Our research reinforces the clinical impact of genomic sequencing, not only in delivering answers, but in identifying potential opportunities to inform treatment and change medical management. By equipping genetic counselors with the data, technology and expertise to translate these insights into action, we can help more patients and families realize the full value of genomic medicine.”

Additional research highlights:

- **The Ripple Effect of Gene Discovery: Real-World Impact of GeneMatcher-Facilitated Collaborations** – As one of the largest contributors to GeneMatcher, GeneDx's collaborative research contributed to 792 definitive diagnoses over 4.5 years, with more than 90% of 103 candidate gene publications resulting in at least one validated gene-disease association or gene upgrade. The findings demonstrate how continued gene discovery can deliver answers to previously undiagnosed patients – November 11, 5:30 p.m. Poster number: GENT211
- **The Diagnostic Yield of Exome and Genome Sequencing in a Large and Diverse Cohort of Over 300,000 Patients: Context Matters** – Research involving more than 300,000 patients reinforces the importance of clinical context in understanding diagnostic yield – November 12, 4:30 p.m. Poster number: GENT200
- **Assessing Medical Actionability of Exome and Genome Sequencing Results in Over 300,000 Individuals from a Large Real-World Diagnostic Cohort** – November 12, 4:30 p.m. Poster number: IND264
- **Scalable Genomic Consent in the NICU and Beyond** – Highlighting the expansion of the My46 digital engagement platform to support genomic counseling and consent in neonatal intensive care settings and beyond. Poster format: Virtual

GeneDx will also participate in three educational sessions:

- **CSI: Clinical Sequencing Investigation: Applying Genomic Evidence to Solve the Case** | November 11, 12:15–1:30 p.m. A GeneDx-sponsored interactive symposium led by Britt Johnson, PhD, FACMG and Flavia Facio, MS, CGC, demonstrating how genomic evidence and AI-powered tools support clinical interpretation.
- **Beyond Algorithms: How AI and Machine Learning Leverage Large-Scale Data to Inform Gene Curation and Variant Classification at Scale** | November 11, 2–3 p.m. Exploring how AI and machine learning can support gene curation and variant classification while emphasizing clinical judgment and human oversight.
- **Hire Power: Unleashing Your Inner Business Case Superhero** | November 13, 1:45–3:45 p.m. Equipping genetic counselors with practical tools to advocate for expanded services and improve patient access to care.

About GeneDx

GeneDx’s (Nasdaq: WGS) mission is to empower everyone to live their healthiest life through genomics. GeneDx combines unmatched clinical expertise, advanced technology, and the power of GeneDx Infinity ™– the world’s largest rare disease genomic dataset. This unparalleled foundation powers GeneDx’s ExomeDx ™and GenomeDx® tests – ranked #1 by expert geneticists and granted FDA Breakthrough Device designation – enabling clinicians to deliver precise, fast, and actionable diagnoses. GeneDx Infinity also fuels discovery for biopharma, with the most powerful AI-driven genomic intelligence. A genomics pioneer over the last 25 years, diagnosing more than 4,800 genetic diseases and publishing more than 1,100 research publications, GeneDx is building the network that will drive the future of genomic precision medicine. For more information, visit **genedx.com** and connect with us on **LinkedIn**, **Facebook**, and **Instagram**.

Forward Looking Statements

This press release may contain “forward-looking statements” within the meaning of Section 21E of the Securities Exchange Act of 1934, as amended, and the U.S. Private Securities Litigation Reform Act of 1995. These forward-looking statements generally are identified by the words “believe,” “project,” “expect,” “anticipate,” “estimate,” “intend,” “strategy,” “future,” “opportunity,” “plan,” “may,” “should,” “will,” “would,” “will be,” “will continue,” “will likely result,” and similar expressions. Forward-looking statements are predictions, projections and other statements about future events that are based on current expectations and assumptions and, as a result, are subject to risks and uncertainties. Many factors could cause actual future events to differ materially from the forward-looking statements in this press release, including but not limited to: (i) our ability to advance gene-disease discovery, (ii) the risk of downturns and a changing regulatory landscape in the highly competitive healthcare industry, (iii) the size and growth of the market in which we operate, (iv) future expansion of insurance coverage for exome and genome testing, and (v) the anticipated impacts of regulatory reimbursement changes. The foregoing list of factors is not exhaustive. A further list and description of risks, uncertainties and other matters can be found in the “Risk Factors” section of our Annual Report on Form 10-K for the fiscal year ended December 31, 2025 and our Quarterly Report on Form 10-Q for the fiscal quarter ended March 31, 2026, filed with the SEC on May 4, 2026, Quarterly Report on Form 10-Q for the fiscal quarter ended June 30, 2026, filed with the SEC on August 3, 2026, and our Quarterly Report on Form 10-Q for the fiscal quarter ended September 30, 2026, filed with the SEC on November 9, 2026, and other documents filed by us from time to time with the SEC. These filings identify and address other important risks and uncertainties that could cause actual events and results to differ materially from those contained in the forward-looking statements. Forward-looking statements speak only as of the date they are made. Readers are cautioned not to put undue reliance on forward-looking statements, and we assume no obligation and do not intend to update or revise these forward-looking statements, whether as a result of new information, future events, or otherwise. We do not give any assurance that we will achieve our expectations.

View source version on businesswire.com: https://www.businesswire.com/news/home/20261110972518/en/

Investors@GeneDx.com
Press@GeneDx.com

Source: GeneDx