GeneDx to Showcase Pioneering Research Findings at American Society of Human Genetics (ASHG) Annual Meeting

October 8, 2025

With unmatched scale and diversity, GeneDx Infinity™ powers scientific discovery while enabling the most precise clinical rare disease diagnoses for patients and families today

GAITHERSBURG, Md.--(BUSINESS WIRE)--Oct. 8, 2025-- GeneDx (Nasdaq: WGS), a leader in delivering improved health outcomes through genomic insights, today announced pioneering scientific contributions that will be presented at the American Society of Human Genetics (ASHG) Annual Meeting.

At ASHG, GeneDx will unveil research findings drawn from GeneDx Infinity™, a dataset of nearly one million exomes and genomes and over seven million phenotypic datapoints. GeneDx will showcase 14 pioneering research studies highlighting advancements in genomic newborn screening, neurodevelopmental disorders, diagnostic technologies, and machine learning applications in genomics. This work has been fueled by GeneDx Infinity - the largest rare disease dataset and the only resource with the depth and diversity needed to fuel equitable care and research.

Throughout the week at ASHG, GeneDx will be presenting data that showcases advances in the following key areas:

Unlocking new discoveries and shortening the diagnostic odyssey with GeneDx Infinity:

- GeneDx Infinity is driving genomic discovery and diagnostic utility, leveraging large-scale exome and genome sequencing data to identify novel disease-associated genes, enhance gene-disease curation, and improve variant detection for complex conditions like intellectual disability, congenital heart disease, and hearing loss, while also demonstrating high concordance with traditional methods for CNV detection.

AI and machine learning approaches uncovering the genetic basis of rare diseases:

- GeneDx's advanced machine learning techniques are accelerating novel gene discovery and streamlining variant classification at scale, enabling accurate and rapid diagnoses and offering new insight into gene–disease associations.

The genetic causes of autism:

- In a study of over 62,000 individuals affected with autism, researchers highlighted a core set of genes associated with autism and found moderate genetic correlations between autism and schizophrenia, epilepsy, and bipolar disorder.

Delivering earlier diagnoses with genomic newborn screening (gNBS):

- Updated results from the GUARDIAN study reports data from 15,000 newborns and demonstrates high enrollment rates, positive parental experiences, and meaningful follow-up outcomes – reinforcing the value of gNBS in accelerating time to diagnosis.

Clinical validation for long read sequencing:

- Long-read sequencing approaches demonstrate strong potential to improve clinical diagnostics, accurately detecting repeat expansions, resolving difficult-to-sequence regions, and benchmarking against short-read pipelines in critically ill newborns, offering foundational data for the potential implementation of long-read platforms in high-throughput clinical laboratories in the future.

GeneDx collaborated on the following:

Platform Presentations:

- **Thursday, October 16, 8:30-10:00 am ET:** GUARDIAN Expanded NBS Study: Short- and Medium-Term Follow-Up with the First 15,000 Participants Enrolled. *Brenna Boyd, MS, CGC* (Columbia University Irving Medical Center)
- **Thursday, October 16, 1:30-2:30 pm ET:** Shared and distinct genetic architectures of autism and neuropsychiatric disorders. *Jack Fu, PhD* (Massachusetts General Hospital)

Posters:

- Beyond panels: the superior diagnostic utility of exome/genome sequencing in hearing loss *Bobbi McGivern, MS, LCGC* (GeneDx) – Poster 7012W
- Benchmarking long-read variant sensitivity across ONT and PacBio platforms using known clinically reported variants in a

cohort of critically ill newborns *Colby T. Marvin* (University of Washington) - Poster 4071F
- The clinical impact of a framework for validating gene-disease associations in a high-throughput clinical laboratory *Maria Guillen Sacoto, MD, FACMG* (GeneDx) - Poster 2057F
- Copy Number Variant Detection by Exome or Genome Sequencing Is Highly Concordant with Chromosomal Microarray *Sarah Poll, PhD (*GeneDx*)* - Poster 8009F
- Concordance of results from Cas9-Targeted Sequencing and LR-WGS with Results from Validated Clinical Methods for the diagnosis of Repeat Expansion Disorders *Jessica Noya, MS (*GeneDx and PacBio) - Poster 8026T
- Resolution of the D4Z4 repeat responsible for facioscapulohumeral muscular dystrophy with HiFi sequencing *Xiao Chen, PhD* (PacBio) - Poster 4104F
- Gene-based burden testing of rare variants identifies established and novel genetic contributors to Intellectual Disability *Thorhildur Juliusdottir, PhD* (GeneDx) - Poster 9147W
- SeqFirst: The value of a "sequencing first" approach: qualitative interviews with SeqFirst families *Mike Bamshad, MD and Olivia Sommerland, MPH* (University of Washington) - Poster 3026F
- A machine learning approach for identifying de novo variants with high accuracy in a high-throughput genomic testing laboratory *Robert Kueffner, PhD* (GeneDx) - Poster 5049F
- Large-scale WES integrating research and clinical genetic testing cohorts identifies novel risk genes for congenital heart disease *Wendy Chung, MD, PhD and Wenxing Li, MS, PhD* (Columbia University Irving Medical Center) - Poster 9116W
- Machine Learning Approach Identifies Genes with Rare Variants Predisposing to Thoracic Aortic Aneurysms and Acute Aortic Dissections *David R. Murdock, MD and Dong-chuan Guo, MS, PhD* (University of Texas Health Science Center at Houston) - Poster 5010W
- Using GORdb to leverage large scale genotype and phenotype data to confirm and explore new gene disease associations *Hildur Olafsdottir, PhD* (GeneDx) - Poster 2056F

Additional CoLab Industry Sessions:

- **Wednesday, October 15, 12:00-1:00pm ET:** Redefining the limits of what's possible: Sequencing reimagined. *Speakers: Steve Barnard, PhD* (Illumina), *Joe Devaney, PhD* (GeneDx), *Bekim Sadikovic, PhD* (London Health Sciences Centre Research Institute)
- **Wednesday, October 15, 3:15-3:45pm ET:** HiFi sequencing at scale: Targeted native DNA sequencing with PureTarget. *Sarah Kingan, PhD* (PacBio)*, Keith Nykamp, PhD* (GeneDx)*, Zach Freeman, PhD* (University of Michigan)
- **Thursday, October 16, 3:15-3:45pm ET:** Accurate and Scalable WGS variant prioritization for phenotype-free newborn screening (NBS) with GEM AI. *Mark Yandell, PhD* (University of Utah) and *Samuel Strom, PhD, FACMG* (Fabric Genomics)

About GeneDx

GeneDx (Nasdaq: WGS) is the global leader in rare disease diagnosis, transforming the way medicine is practiced by making genomics the starting point for health, not the last resort. We bring together unmatched clinical expertise, advanced technology, and the power of GeneDx Infinity™ – the largest rare disease dataset – built over 25 years from millions of genomic tests and deep clinical insights. This unparalleled foundation powers our ExomeDx and GenomeDx tests, giving clinicians the highest likelihood of delivering a timely, accurate diagnosis. GeneDx is shaping the future of healthcare by moving the standard of care from sick care to proactive healthcare. While our roots are in rare disease diagnosis, our commitment extends beyond – growing with the families we serve – as a trusted partner at every stage of life. 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 implement plans to accelerate scientific discoveries and unlock other value in the rare disease space, (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) our ability to pursue our new strategic direction. 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, 2024 and our Quarterly Reports on Form 10-Q for the fiscal quarters ended March 31, 2025 and June 30, 2025, 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/20251008971231/en/

Investor Relations Contact:
Investors@GeneDx.com

Media Contact:
Press@GeneDx.com

Source: GeneDx