



GeneDx Advances Clinical Adoption of Genomic Medicine with 27 Abstracts at ASHG 2026

October 19, 2026

Research strengthens the evidence for exome and genome sequencing as standard of care across rare disease, autism, newborn screening and beyond

GAITHERSBURG, Md.--(BUSINESS WIRE)--Oct. 19, 2026-- GeneDx, the leader in rare disease diagnosis and improving health through the power of genomic data, today announced 27 accepted scientific abstracts at the American Society of Human Genetics (ASHG) 2026 Annual Meeting, highlighting how exome and genome sequencing are advancing scientific discovery, earlier diagnosis and more precise care.

GeneDx research at ASHG 2026 highlights five areas:

- **Scaling genomic interpretation with AI and machine learning:** A GeneDx-led platform presentation explores machine learning to prioritize relevant copy number variants (CNVs), reflecting the company's human-first, human-last approach to leveraging AI decision support tools in exome and genome sequencing analysis.
- **Advancing exome and genome sequencing as standard of care:** Research from more than 100,000 individuals with autism examines factors influencing molecular diagnostic yield, adding evidence to support broad genomic testing for complex neurodevelopmental conditions.
- **Translating clinical data into actionable insights:** Studies explore genotype-first reanalysis, emerging genotype-phenotype associations, rare variant detection and the health-economic value of first-line exome and genome sequencing.
- **Accelerating gene discovery:** Nine abstracts using GeneDx Infinity™, GeneMatcher and global research collaborations to identify novel disease-associated genes and expand understanding of developmental and neurodevelopmental disorders.
- **Expanding equitable genomic newborn screening:** Findings from the GUARDIAN study examine positive rates and variant findings in more than 19,000 newborns across diverse genetic ancestries, informing the implementation of genomic newborn screening at scale.

"Advancing genomic medicine means turning scientific discovery into meaningful answers for patients," said Linda Genen, MD, MPH, Chief Medical Officer at GeneDx. "Through our clinical data, technology and research collaborations, we're building the evidence to make exome and genome sequencing a more accessible and actionable part of care across specialties and throughout life."

The research includes three platform presentations, three rapid-fire poster talks and a Reviewer's Choice poster, an honor awarded to the top 10% of posters at ASHG.

Featured GeneDx-Led and Collaborative Presentations

Platform Presentations:

- Genomic Newborn Screening Across Diverse Genetic Ancestries: GUARDIAN Positive Rates and Variant Findings Among >19,000 Newborns
 - Britt Johnson, GeneDx – October 21, 11:15–11:30 a.m. | Room 517A
- Continuous clinical prioritization of copy number variants (CNVs) through dynamic feature reconstruction
 - Robert Kueffner, GeneDx – October 22, 9:15–9:30 a.m. | Room 510
- Factors influencing molecular diagnostic yield in a clinical autism exome sequencing cohort of over 100,000 individuals
 - Kendall King, GeneDx – October 22, 1:30–1:45 p.m. | Room 510

Rapid-Fire Poster Talks:

- Variants in SFPQ cause a dominant neurodevelopmental syndrome
 - October 21, 10:30–10:33 a.m. | Room 710A
- Analysis of 184,786 developmental disorder trios identifies over a hundred novel DD-associated genes and candidate haplolethal genes
 - October 21, 10:35–10:38 a.m. | Room 710A
- Real-world prevalence of clinically significant RNU4-2 variants and the value of genotype-first reanalysis
 - Shelly Galasinski, GeneDx – October 21, 10:39–10:42 a.m. | Room 710A

Reviewer's Choice Poster:

- Large-Scale Gene-Based Burden Analysis Reveals the Genetic Architecture of Microcephaly in 25,000 Individuals
 - GeneDx – October 23, 2:30–4:30 p.m.

Additional accepted research spans autism gene discovery, sequencing technologies, structural variant detection, AI-assisted phenotype modeling, rapid neonatal genome sequencing, cardiovascular genetics and the economic impact of genomic testing. All study results remain under embargo until their respective ASHG presentations or poster sessions.

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 1,000 research publications, GeneDx is building the network that will drive the future of genomic precision medicine. For more information, visit [genedx.com](https://www.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, and (iv) future expansion of insurance coverage for exome and genome testing. 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, our Quarterly Report on Form 10-Q for the fiscal quarter ended June 30, 2026, filed with the SEC on August 3, 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.

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