GeneDx to Present 18 Abstracts at ACMG 2026, Showcasing the Scale of GeneDx Infinity™ and Real-World Genomic Leadership

March 9, 2026

GeneDx showcases leadership in exome and genome quality, expanded access through non-genetics ordering, and AI-driven innovations that enhance speed, scale, and diagnostic impact

GAITHERSBURG, Md.--(BUSINESS WIRE)--Mar. 9, 2026-- GeneDx (Nasdaq: WGS), the leader in rare disease diagnosis and improving health through the power of genomic data, today announced scientific contributions to be presented at the 2026 American College of Medical Genetics and Genomics (ACMG) Annual Meeting. GeneDx will present 18 accepted abstracts, including three platform presentations and two rapid fire poster talks, with research spanning sequencing innovations, breakthroughs in AI-supported interpretation, novel gene-disease discoveries, the real-world impact of earlier diagnoses, and more.

This research highlights the scale and clinical impact of GeneDx Infinity™ – the largest and most comprehensive rare disease genomic dataset – and GeneDx's leadership in bringing exome and genome testing to the standard of care. The research showcases how GeneDx leverages AI, combined with world-class data and clinical expertise, to enhance variant interpretation, accelerate analysis, and scale insights from Infinity.

"Only GeneDx can study exome and genome performance at the scale reflected in GeneDx Infinity – more than a million exomes and genomes linked to deep clinical context," said Dr. Linda Genen, MD, MPH, Chief Medical Officer at GeneDx. "The data we're presenting at ACMG 2026 show that when genomic testing is used earlier and interpreted with the power of real-world genomic intelligence, it doesn't just provide answers – it fundamentally changes care. Across diverse clinical settings and provider types, exome and genome testing shorten the diagnostic journey, guide medical management, and reduce healthcare costs for patients and health systems alike."

GeneDx Platform Presentations:

Utilization and Impact of Exome and Genome Sequencing by Non-Genetics Providers
Thursday, March 12 at 12 p.m. BCC Ballroom 1 – Melanie P. Napier, MSc, MSc, CGC, CCGC, GeneDx
Data from 135,000 WES/WGS tests (2022–2024) show that diagnostic yield and VUS rates were comparable between genetics and non-genetics providers, underscoring appropriate utilization and the expanding access to genomic testing.

A Rapid, Novel Approach to Rare Disease and Clinical Genetic Variant Discovery Using On-Flowcell Proximity Sequencing and Haplotype-Resolved Variant Calling
Thursday, March 12 at 12:30 p.m. BCC Ballroom III – GeneDx in partnership with Illumina
This study demonstrated 31/31 known difficult-to-sequence variants were detected using Constellation technology, reinforcing the potential of innovative sequencing methods to solve previously challenging cases.

Benchmarking Comprehensive Variant Detection in Challenging Genomic Regions Using TruPath™ Genome
Thursday, March 12 at 12:45 p.m. BCC Ballroom III – Joseph M. Devaney, PhD, GeneDx
A benchmarking study of 152 diverse DNA samples demonstrated 98.9% detection of expected variants, highlighting improved detection of structural variants and resolution of complex genomic regions using advanced sequencing approaches.

GeneDx to reveal latest AI advancements and context on diagnostic yield in two rapid fire poster talks

GeneDx leverages AI across its platform to enhance variant interpretation, accelerate analysis, and scale insights from the world's largest rare disease dataset. Results from GeneDx's latest AI-powered variant ranker, which prioritizes pathogenic variants with high recall to reduce manual review time and enable faster, more confident diagnoses, as well as new analyses from the largest clinical cohort to date demonstrating that diagnostic yield alone does not define test performance will be featured in the following rapid fire poster talks:

MIMI Ranker: An AI-Powered Tool for Pathogenic Variant Prioritization Tested on a Diverse Dataset of 16K Clinical Exomes and Genomes
Thursday, March 12 at 12:00 p.m. BCC Ballroom I – Amber Begtrup, PhD, FACMG, GeneDx
Evaluation of GeneDx's AI-based MIMI Ranker – the company's latest proprietary AI technology – demonstrated high recall of pathogenic and likely pathogenic variants while significantly reducing manual interpretation time, underscoring the importance of combining machine learning with large, representative datasets and genetics expertise.

Diagnostic Yield of Exome and Genome Sequencing in a Large and Diverse Cohort of Over 300,000 Patients: Context Matters

Thursday, March 12 at 12:00 p.m. BCC Ballroom I – Kirsten Kelly, MS, LSGC, GeneDx
Analysis of more than 300,000 cases collected over 12 years demonstrated that GeneDx's molecular diagnostic yield for exome and genome testing is 51.4% across age groups, clinical indications, and genetic ancestries. Findings provide one of the largest real-world benchmarks for clinical WES/WGS performance and emphasize the importance of the context around diagnostic yield data.

GeneDx data will also be featured in the following poster presentations:

- Benchmarking Variant Classification Quality: Concordance of an ACMG-Based Framework with >400,000 ClinVar Submissions from Over a Decade of Clinical Testing will be presented by Sarah R. Poll, PhD, GeneDx – Thursday, March 12 at 10:30 a.m. BCC Ballroom I
- Early Experiences of a Commercial Laboratory with Ultra-Rapid Genome Sequencing - Insights from 100 cases will be presented by Melanie P. Napier, MSc, MSc, CGC, CCGC, GeneDx – Thursday, March 12 at 10:30 a.m. BCC Ballroom I
- GeneMatcher-Facilitated Collaborative Research Ends Diagnostic Odysseys: From Candidate Gene Cohorts to Diagnostic Reports will be presented by Erin Torti, MS, CGC, GeneDx – Thursday, March 12 at 10:30 a.m. BCC Ballroom I
- High-Throughput Detection of Pathogenic Repeat Expansions Using PureTarget: Concordance with Clinical Assays and Mosaic Insights will be presented by Jessica Noya, MS, GeneDx – Thursday, March 12 at 10:30 a.m. BCC Ballroom I
- Reduction in Healthcare Costs for Children with Epilepsy Following Exome and Genome Sequencing: a SAVES-Kids Study will be presented by Colton Frazer, PharmD, MBA, RPh, GeneDx – Friday, March 13 at 10:30 a.m. BCC Ballroom I
- Reduction in Healthcare Resource Utilization for Children with Neurodevelopmental Disorders After Exome and Genome Sequencing: A SAVES-Kids Study will be presented by Colton Frazer, PharmD, MBA, RPh, GeneDx – Friday, March 13 at 10:30 a.m. BCC Ballroom I
- Solving the Unsolved: Epigenomic Insights from Long-Read Sequencing Improves Diagnosis of Mendelian Disorders will be presented by Eunjee Lee, PhD, GeneDx – Thursday, March 12 at 10:30 a.m. BCC Ballroom I

Through strategic collaborations with Illumina, CLS Foundation, BEACONS-NBS, GUARDIAN, and SeqFirst, GeneDx is helping drive exome and genome sequencing toward becoming the standard of care across diverse clinical settings. This work contributed to the following:

- SeqFirst Developmental Differences – Early whole genome sequencing offered using broad inclusion criteria improves access to early precise genetic diagnosis – Katrina Dipple, MD, PhD, FACMG – Thursday March 12 at 10:30 a.m. BCC Ballroom I
- Large-Scale Implementation of Rapid Genome Sequencing as a First-Tier Diagnostic in 662 Hospitalized Patients at a Tertiary Center, Ecenur Tuc Bengur MD – Thursday, March 12 at 10:30 a.m. BCC Ballroom I
- Variant Interpretation and Reporting Approaches in the GUARDIAN Study of Genomic Newborn Screening – Mehmet B. Duz, MD – Thursday March 12 at 10:30 a.m. BCC Ballroom I
- Hospital-wide implementation of first-tier rapid genome sequencing – Tara Wenger, MD, PhD, FACMG – Friday, March 13 at 10:30 a.m. BCC Ballroom I
- Gene List Development for the BEACONS-NBS study: The First U.S. Multi-State Genomic Newborn Screening Initiative – Britt Johnson, PhD, FACMG, GeneDx – Friday, March 13 at 10:30 a.m. BCC Ballroom I
- Building a Natural History and Patient Registry Platform for Coffin-Lowry Syndrome (CLS): A Roadmap for Therapeutic Readiness – Mary Hames – Friday, March 13 at 10:30 a.m. BCC Ballroom I

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 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, 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.

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