GeneDx Leads Rare Disease Genomics with Unmatched Research and Clinical Impact in 2025

January 5, 2026

Industry-leading research powered by GeneDx Infinity™ accelerates the transition of genomics from discovery to the standard of clinical care, driving better outcomes for patients across healthcare settings

GAITHERSBURG, Md.--(BUSINESS WIRE)--Jan. 5, 2026-- GeneDx (Nasdaq: WGS), a leader in delivering improved health outcomes through genomic insights, today announced a record-setting year for its Medical Affairs and research programs, reinforcing its position as the global leader in rare disease genomics. In 2025, GeneDx published 79 peer-reviewed studies bringing its total to over 1,100 peer-reviewed studies – all powered by GeneDx Infinity™ – delivering the most prolific and clinically impactful research portfolios in rare disease genomics.

GeneDx Infinity™, the world's largest rare disease genomic dataset, is built on more than 25 years of real-world clinical experience and includes nearly 1 million exomes and genomes enriched for rare disease and deep phenotypic data across diverse populations. This continuously growing dataset fuels GeneDx's industry-leading research, enabling precise diagnosis, faster discovery, stronger evidence generation, and precision genomic medicine across healthcare settings.

GeneDx's 2025 research output included four strategic, practice-shaping publications, 56 GeneMatcher-enabled discoveries, and 38 strategic conference abstracts presented at the world's leading medical and scientific forums. Together, these contributions demonstrate GeneDx's unmatched ability to move genomics beyond research and into routine clinical care – setting the standard for how genome-first medicine is delivered across newborn, pediatric, and rare disease populations.

Among the year's most influential was the GUARDIAN (Genomic Uniform-screening Against Rare Disease In All Newborns) study which was recognized as part of *JAMA*'s annual **Research of the Year Roundup**. This highly selective honor is reserved for the most impactful scientific contributions published across all *JAMA* journals. The recognition highlights the significance of GUARDIAN's peer-reviewed findings, which demonstrate the power of genomic newborn screening to identify serious, actionable childhood conditions more broadly than traditional newborn screening.

GeneDx also advanced genome-first clinical care across multiple care settings through a series of landmark publications, including:

- **SeqFirst Neo** – establishing rapid genome sequencing as a pathway to equitable, precise diagnosis for critically ill newborns (Q1)
- **SeqFirst Floor** – demonstrating the successful implementation of first-tier rapid genome sequencing in non-critical pediatric wards (Q2)
- **Advantages of exome sequencing over panel testing for seizure indications** – reinforcing the clinical superiority of comprehensive genomic testing over stepwise, panel-based approaches (Q4)

"GeneDx defines what leadership in rare disease genomics looks like," said Britt Johnson, PhD, FACMG and Senior Vice President of Medical Affairs at GeneDx. "Our research doesn't just advance scientific understanding – it changes clinical practice. In 2025, we showed at unprecedented scale how a genomic-first testing approach delivers precise diagnoses, greater equity, and more confident care decisions. This is how genomics becomes the standard of care and we can deliver better care for patients through true genomic precision medicine."

Further strengthening its leadership, GeneDx supported 56 GeneMatcher publications in 2025 – catalyzing global collaboration to accelerate gene–disease discovery and deliver long-overdue answers to patients who have historically gone undiagnosed. Across these GeneMatcher publications, GeneDx partnered with 175 clinicians from 74 institutions, helping provide diagnoses for 136 patients. These efforts also resulted in 31 newly clinically validated gene–disease associations in 2025, bringing the total to over 500 new gene-disease validations that GeneDx has contributed to, extending the impact to future patients who will benefit through faster, more precise diagnoses.

"With unmatched clinical depth, real-world evidence, and a proven ability to translate discovery into care, GeneDx is paving the way for a genome-first healthcare system," said Katherine Stueland, President and CEO of GeneDx. "Through our research and collaborations, we are demonstrating how healthcare can work better for patients, enabling earlier diagnosis, precision care, and better outcomes the standard, not the exception."

About GeneDx
GeneDx (Nasdaq: WGS) is the global leader in rare disease diagnosis, with a mission 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 advance gene-disease discovery and implement plans to accelerate and unlock the full potential of precision medicine, (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, June 30, 2025, and September 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.

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