GeneDx Continues its Leadership in Genomics Research, Refining Gene-Disease Relationships and Impacting Results of >22,000 Patients

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STAMFORD, Conn.--(BUSINESS WIRE)--Jan. 8, 2025-- GeneDx (Nasdaq: WGS), a leader in delivering improved health outcomes through genomic insights, continues its investment in advancing scientific discovery through understanding gene-disease relationships, contributing to more than 85 peer-reviewed publications in 2024. These contributions, added to GeneDx's years of dedicated research efforts, influenced the broadening of phenotypes, the discovery of new disease mechanisms and new modes of inheritance, and ultimately impacted results of 12% of all patients who have received exome or genome sequencing from GeneDx.

In 2024, GeneDx also contributed nearly one quarter of all submissions to GeneMatcher, a platform that facilitates disease-gene discovery by sharing genetic findings among patients, clinicians, and researchers. For each "candidate gene"—a gene that is highly suspected (but not yet proven) to be linked to a specific disease—identified byGeneDx, the lab submits the finding to GeneMatcher, which then connects researchers and clinicians around the world so they can work together to better understand genetic contributions to rare disease. To date, this research has impacted >22,000 patients tested at GeneDx and, in the future, will influence the results of many additional patients seeking diagnoses or answers.

"The combination of our high-quality genomic testing and our dedicated involvement in research keeps GeneDx at the forefront of genomics and uniquely positions us to influence the future of healthcare," said Paul Kruszka, MD, FACMG, Chief Medical Officer at GeneDx. "We're committed to improving the lives of patients and their families by delivering definitive diagnoses, and that ultimately begins with a deep understanding of gene-disease relationships. We're honored to contribute our knowledge to the broader scientific community to ultimately help more patients."

By the numbers

GeneDx is a leader in scientific inquiry, striving to find answers for more patients. Through ongoing research efforts in 2024, GeneDx:

- Contributed to 86 publications in peer reviewed journals, bringing the total to over 1,121 publications to date and demonstrating the company's leadership in the commercial laboratory space
- Collaborated on 26 publications that expanded current knowledge of known disease-gene associations, including broadening phenotypes, new disease mechanisms, and new modes of inheritance
- Collaborated on 35 publications that reported new disease-gene relationships
- Increased its cumulative GeneMatcher submissions to more than 21,315—accounting for 22.2% of all such submissions and making GeneDx the largest contributor of the 16,812 laboratories, researchers, and clinicians taking part in the initiative

Recognizing that GeneDx ordering clinicians have a vested interest in finding definitive answers for their patients, GeneDx proactively invites them to take part in research around their patients' candidate gene results. In 2024, almost 150 GeneDx ordering clinicians were included as co-authors on the company's GeneMatcher publications, bringing the all-time total to 638 unique ordering clinician co-authors.

These endeavors underscore the company's commitment to improving healthcare for rare disease patients through genomic answers, its dedication to involving ordering clinicians in research efforts that benefit their patients, and its impact on the fundamental understanding of genetics—powering future treatment advancements and innovation.

About GeneDx:
GeneDx (Nasdaq: WGS) delivers personalized and actionable health insights to inform diagnosis, direct treatment, and improve drug discovery. The company is uniquely positioned to accelerate the use of genomic and large-scale clinical information to enable precision medicine as the standard of care. GeneDx is at the forefront of transforming healthcare through its industry-leading exome and genome testing and interpretation services, fueled by the world's largest, rare disease data sets. For more information, please visit www.genedx.com and connect with us on LinkedIn, Facebook, and Instagram.

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