GeneDx Makes Significant Industry-Wide Contributions to New Disease Gene Discovery

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Company submitted nearly one quarter of all candidate gene submissions to GeneMatcher in 2022 and collaborated on 63 publications involving new disease-gene associations or expansion of phenotype discoveries

STAMFORD, Conn., Jan. 30, 2023 (GLOBE NEWSWIRE) -- Throughout 2022, GeneDx (Nasdaq: WGS), a leader in delivering improved health outcomes through genomic and clinical insights, contributed a significant amount – 22% – of total candidate gene submissions to GeneMatcher, a platform that facilitates new disease gene discovery and enables connections between patients, clinicians and researchers by allowing individuals to submit and search genes of interest.

GeneDx is dedicated to eradicating the multi-year diagnostic odyssey faced by many families seeking clinical diagnoses for critically ill patients. The company has the world's largest rare-disease dataset including more than 400,000 clinical exomes, 2.7 million phenotypes and over 1 million patient samples. Its data contributions to GeneMatcher enable research with a large number of global external collaborators to increase knowledge of candidate genes; GeneMatcher efforts have contributed to the diagnoses for more than 13,800 individuals tested by GeneDx, as well as countless others due to publication of findings.

"Our contributions to GeneMatcher are just one way we demonstrate our commitment to ending the diagnostic odyssey for patients," said Katherine Stueland, president and CEO of GeneDx, "By focusing on the bigger picture and utilizing our immense dataset and ability to interpret genetic findings at scale, we can continue to develop a more complete understanding of complex disease, improve diagnosis rates and help providers build effective treatment plans."

By The Numbers

GeneDx is a leader in scientific inquiry, striving to find answers for patients and contribute to medical literature. In 2022, an average of 13 patients tested by GeneDx were included in GeneMatcher publications each month, and an average of 18 GeneDx-ordering clinicians were included as authors on GeneMatcher publications each month.

Additionally, in 2022, the company:

- Contributed to 151 publications in peer-reviewed journals, significantly more than its closest competitors
- Collaborated on 33 publications that established new disease gene associations
- Collaborated on 30 publications that expanded knowledge of known disease gene associations, including broadened phenotypes, new disease mechanisms, or modes of inheritance

These endeavors strengthen the knowledge base for rare disease diagnostics, which is critical for treatment improvements and innovation. They also underscore the company's dedication to patient needs and commitment to collaboration and clinician input, as many providers are directly involved in GeneDx-related research and publications.

About GeneDx (formerly Sema4)

GeneDx 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, fueled by one of the world's largest rare disease data sets. For more information, please visit genedx.com and connect with us on LinkedIn, Facebook, and Instagram.

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