GeneDx Announces Progress on GUARDIAN Study and Promise of Early Genomic Testing to End Rare Disease “Diagnostic Odyssey”

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- *Genome Sequencing and Interpretation Services Progressing Quickly for 1,000 Newborns Enrolled in Study to Date; On Track to Support up to 100,000 Study Participants in Next Four Years*

- *Initial Data and Analysis Presented at the ACMG (American College of Medical Genetics and Genomics) Annual Meeting Show Whole Genome Sequencing Identified Rare Genetic Conditions Not Otherwise Part of Standard Newborn Screening*

STAMFORD, Conn., March 20, 2023 (GLOBE NEWSWIRE) -- GeneDx Holdings Corp (GeneDx) (Nasdaq: WGS), a leader in delivering improved health outcomes through genomic and clinical insights, today announced it is continuing to support whole genome sequencing (WGS) and interpretation services for the GUARDIAN (Genomic Uniform-screening Against Rare Diseases In All Newborns) study, a research study recently launched by Columbia University using WGS to screen 100,000 newborns for more than 250 genetic conditions not currently included in standard newborn screening. Initial data from this study were presented at the ACMG Annual Meeting during the R. Rodney Howell Symposium, *Setting the Stage for Genomic Sequencing of All Newborns,* on Saturday, March 18, 2023.

As presented by Wendy Chung, M.D., Ph.D., a clinical and molecular geneticist, the Kennedy Family Professor of Pediatrics in Medicine and Chief of Clinical Genetics at Columbia University, and principal investigator for the study, of the 1,000 newborns enrolled to date, true positive screening outcomes were present in 2.6 percent of newborns. This included 15 confirmed cases with G6PD deficiency, a genetic disorder not integrated with standard newborn screening. G6PD deficiency is a result of decreased function in an enzyme called G6PD (glucose-6-phosphate dehydrogenase) and causes a breakdown of red blood cells in response to infections, certain drugs, foods, or stress, and is a risk for severe neonatal hyperbilirubinemia.

Additionally, reportable sequence variants from 238 genes screened in the GUARDIAN study were included in a retrospective analysis performed by GeneDx. For almost 25,000 individuals with positive exome or genome sequencing, more than 20% of individuals could have identified their genetic disease, on average, 7 to 11 years sooner had they received genome sequencing at birth.

“GUARDIAN is quickly teaching us a lot about how to implement genome sequencing in newborns,” said Paul Kruszka, M.D., Chief Medical Officer of GeneDx. “GeneDx is committed to delivering genetic testing services that end the extensive periods of time it can take for a patient to receive a diagnosis for their condition, commonly referred to as the diagnostic odyssey. We hope studies like the GUARDIAN study are launched across the U.S. to give more families access to this type of informative testing.”

For patients, early diagnosis means doctors and health systems can consider available interventions and therapies more quickly, often leading to improved quality-of-life. Additionally, early diagnosis has financial implications, including saving, on average, $30,000 per case due to reduced hospital stays and unnecessary or ineffective care for patients in the newborn intensive care unit (NICU)[1]. Further, as more patients are diagnosed with a rare disease, clinical trials can be established that help to more quickly develop therapies that can treat, or perhaps cure, rare diseases.

“The GUARDIAN study has the potential to transform children’s health by equitably diagnosing babies early in life at a time when treatment is most effective,” said Dr. Wendy Chung “Through this study, we are learning how to move genomic medicine forward at scale for the next generation.”

“Rarely in medicine is there such a monumental shift in how we deliver patient care. Genomic sequencing can and should be ubiquitous in NICU settings and is poised to become a standard for newborn screening,” said Katherine Stueland, President and CEO of GeneDx. “These data reinforce the power of genomic sequencing and the need to expand this more broadly. Had WGS been available at birth, we expect patients, and their families would avoid a diagnostic odyssey including years of inconclusive tests, thousands of dollars in medical expenses and the emotional toll that comes with seeking a diagnosis.”

About GUARDIAN
GUARDIAN (Genomic Uniform-screening Against Rare Diseases In All Newborns) is a research study sponsored by Columbia University in partnership with New York-Presbyterian, and the New York State Department of Health using whole genome sequencing (WGS) provided by GeneDx to screen 100,000 newborns for more than 250 genetic conditions not currently included in standard newborn screening. The goals of the study are to drive earlier diagnosis and treatment to improve the health of the babies who participate, generate evidence to support the expansion of newborn screening through genomic sequencing, and characterize the prevalence and natural history of rare genetic conditions. More information about GUARDIAN can be found at https://guardian-study.org/.

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 one of 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.

[1]ScienceDaily. (2017, October 19). Rapid whole-genome sequencing of neonatal ICU patients is useful and cost-effective.

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