GeneDx honors Rare Disease Day and announces presentations and research at the 2023 American College of Medical Genetics and Genomics Annual Clinical Genetics Meeting

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Research illustrates company's progress in working to end the diagnostic odyssey and includes data with implications for how care is delivered to patients beginning at birth

STAMFORD, Conn., Feb. 28, 2023 (GLOBE NEWSWIRE) -- GeneDx (Nasdaq: WGS), a leader in delivering improved health outcomes through genomic and clinical insights, today announced its contributions to eight posters, one platform presentation and one symposium session to be released at the upcoming 2023 American College of Medical Genetics and Genomics (ACMG) Annual Clinical Genetics Meeting. The announcement coincides with Rare Disease Day and is just the latest example of the company's dedication to eradicating the multi-year diagnostic odyssey faced by many families seeking clinical diagnoses for critically ill patients.

GeneDx is a leader in rare disease diagnosis and has spent the last 20 years building one of the largest rare disease datasets in the genomics industry, including data from more than 400,000 clinical exomes. These data play a critical role in the company's research -- and in the insights it delivers to providers -- to enable the delivery of informed, patient-centered care. It also supports the identification, discovery and development of creating therapeutics to manage and treat rare genetic diseases.

"While today is officially recognized as Rare Disease Day, every day is rare disease day for us as we work tirelessly to shorten and ultimately end the diagnostic odyssey," said Katherine Stueland, GeneDx President and CEO. "With the increasing focus on patients with rare disease, we're proud to bolster the medical community's ability to diagnose disease more accurately and rapidly. Our research contributions are evidence of our investment into unlocking and advancing the power of genomic information to become the standard of care in diagnosing and treating rare disease."

The research and collaborations being shown at ACMG are just the latest demonstration of the company's continued success in leveraging genetic data and technology to generate clinical insights that lead to improved and faster diagnoses, new and better patient treatments and improved outcomes.

GeneDx's presence at ACMG on March 15 at the Salt Palace Convention Center in Salt Lake City, Utah, is as follows:

ORAL PRESENTATION RESEARCH COLLABORATIONS

- *Genomic Uniform-screening Against Rare Diseases in All Newborns (GUARDIAN): Presented by Dr. Wendy Chung, MD, PhD, Overall Study Principal Investigator, Columbia University: [Part of the R. Rodney Howell Symposium, March 15, 2023, 10:45 AM MT]*
- Developing a Framework for Sequence Variant Interpretation for Multiple X-linked Inborn Errors of Metabolism: The ClinGen IEM Working Group Experience *In partnership with ClinGen [March 17, 2023 3:30 PM MT]*

GENEDX POSTER PRESENTATION RESEARCH COLLABORATIONS

- *In collaboration with Tara Wenger and Alexandra Keefe of University of Washington:*
  - *Parental Perspectives on SeqFirst Neonatal Rapid Genome Sequencing*
  - *SeqFirst: Impact of a Precise Genetic Diagnosis on End-of-Life Decision Making in the NICU*

POSTER PRESENTATIONS SHOWCASING STUDIES LED BY GENEDX

- *Diagnostic Yield of Copy Number Variants by Exome Sequencing versus Chromosomal Microarray*
- *Four Cases of Mosiac Triploidy Identified by Trio Exome Sequencing*

POSTER PRESENTATION RESEARCH COLLABORATIONS: RARE DISEASE DISCOVERY AND CONTRIBUTIONS TO PROFESSIONAL DECISION-MAKING BODIES

Rare Disease Discovery:

- *De novo* missense variants in *ZBTB47* cause a neurodevelopmental phenotype of developmental delays, seizures, and possible movement abnormalities

- Mutation of Cohesin Release Factors *WAPL, PDS5A,* and *PDS5B* Defines a New Class of Cohesinopathies

Professional Decision-Making Bodies:

- *ATM* and *PALB2* Variant Curation Guidelines Progress Update: ClinGen Hereditary Breast, Ovarian, and Pancreatic Cancer Variant Curation Expert Panel *In partnership with ClinGen*
- The ClinGen ENIGMA BRCA1/2 Expert Panel: a dynamic framework for evidence-based recommendations to improve classification criteria for variants in BRCA1/2 *In partnership with ClinGen*

About GeneDx
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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