GeneDx to Highlight Key Research Findings at American Society of Human Genetics (ASHG) Annual Meeting

November 4, 2024

Data to be presented showcasing GeneDx's ongoing commitment to spearheading industry-leading research to drive transformational clinical utility

STAMFORD, Conn.--(BUSINESS WIRE)--Nov. 4, 2024-- GeneDx (Nasdaq: WGS), a leader in delivering improved health outcomes through genomic insights, today announced its scientific contributions at the 2024 American Society of Human Genetics (ASHG) annual meeting. GeneDx data will be presented across six platform presentations and five posters and will unveil findings from key research initiatives constructed on the backbones of its industry leading diverse dataset of more than 700,000 clinical exome and genomes.

GeneDx leverages its database as a critical tool in a number of studies to drive transformational clinical care for pediatric patients as the industry continues to look for strong evidence to expand the utilization and clinical utility of genome sequencing. Through these strategic collaborations with SeqFirst, The University of Washington, PacBio the Autism Sequencing Consortium and GUARDIAN, GeneDx showcases the accessibility, affordability and actionability of exome and whole genome sequencing (WGS) in pediatric patients.

Research to be presented this week at ASHG will include:

Rapid whole genome sequencing (rWGS) in the NICU leads to changes in clinical care:

- In collaboration with SeqFirst, patient cases were analyzed when a diagnosis was found with rWGS to understand how decisions were made with genomic sequencing and what is missed in its absence when only using conventional care protocols.

Racial disparities in an accurate genetic diagnosis:

- In one of the largest studies to look at ancestral backgrounds and genetic diagnosis, GeneDx, the University of Washington, and Geisinger explore the value of a diverse dataset to understand diagnostic yield and if it varies significantly based on ancestral background or if other factors are limiting access to a genetic diagnosis.

Data validation for long read sequencing:

- With growing interest in the field to explore the clinical utility of long read sequencing, validation data will be presented assessing the sensitivity of PacBio's HiFi long read sequencing to detect cases with a confirmed answer on short read whole genome sequencing (WGS) Additional cases where long read sequencing uncovered pathogenic variants that were difficult to detect on short read WGS will be presented.

Genetic variants linked to Autism Spectrum Disorder (ASD):

- Working alongside the Autism Sequencing Consortium, research identifies 230 new genes associated with ASD. This molecular evidence underscores the effectiveness and accuracy of genetic diagnostics compared to current methods, which rely on parents' or caregivers' accounts of their child's development or professional observations of behavior.

"The opportunity to present these findings at ASHG underscores GeneDx's commitment to advancing genomic research to move forward its application in clinical settings," said Dr. Paul Kruszka, MD, FACMG, Chief Medical Officer at GeneDx. "Our collaborations with leading research initiatives allow us to leverage our industry-leading dataset to drive innovation and improve patient outcomes. Across the board this work not only highlights the proven clinical utility of genomic testing but emphasizes the importance of equitable access."

GeneDx collaborated on the following:

Presentations:

- Wednesday, November 6, 9:15 am MT: Unveiling the crucial neuronal role of the proteasomal ATPase subunit gene PSMC5 in neurodevelopmental proteasomopathies. Janelle Stanton, PhD (University of Limerick, Ireland) - Room 505 Session 12
- Thursday, November 7 at 1:30 pm MT: Use of exclusion criteria to select critically ill newborns for rapid genome sequencing captures precise genetic diagnoses missed by use of conventional inclusion criteria. Tara Wenger, MD, PhD (University of Washington) – Room 505 Session 54

- Friday, November 8 at 10:45 am MT: Genome-wide profiling of highly similar paralogous genes using HiFi sequencing. Xiao Chen, PhD (PacBio) – Four Seasons Ballroom 4 Session 71
- Friday, November 8 at 11:15 am MT: Benchmarking detection of technically challenging pathogenic variants with long-read sequencing and a head-to-head comparison with short-read sequencing in a clinical diagnostic laboratory. Joseph M. Devaney, PhD (GeneDx) – Four Seasons Ballroom 4 Session 71
- Friday, November 8 at 1:45 pm MT: The largest to-date exome study of autism spectrum disorder triples the number of autism-associated genes. Frederick Satterstrom, PhD (Broad Institute) – Room 401 Session 78
- Friday, November 8 at 6:00 pm MT: Expanded newborn screening using genome sequencing for early actionable conditions: results of the first 10,000 participants enrolled in the GUARDIAN study. Wendy Chung, MD, PhD (Boston Children's Hospital) – Mile High Ballroom Session 86

Posters:

- Thursday, November 7 at 2:30 pm MT: Racial disparities in access to a precise genetic diagnosis are not due to differences in diagnostic yields. Jessica X. Chong, PhD (University of Washington)
- Thursday, November 7 at 2:30 pm MT: Partial methylation of a pathogenic *XYLT1* repeat expansion associated with intrafamilial variation in severity of Desbuquois dysplasia 2. Michael J. Bamshad, MD (University of Washington)
- Friday, November 8 at 2:30 pm MT: Genetic etiologies and diagnostic yield of exome sequencing in pediatric motor speech disorders. Marissa Mitchel, MS (Geisinger Autism & Developmental Medicine Institute)
- Friday, November 8 at 2:30 pm MT: Evaluating dosage sensitivity predictions for multigenic copy number variants to facilitate clinical interpretation. Erin Riggs, MS (Geisinger)
- Friday, November 8 at 2:30 pm MT: De novo variants in *GTF2H1* underlie variable syndromic developmental delay. Karynne Patterson, BS/BA (University of Washington)

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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