GeneDx to Showcase Key Research and Innovation at the 2025 American College of Medical Genetics (ACMG) Annual Meeting

March 13, 2025

Presentations to highlight how GeneDx is leveraging AI to drive both better patient care and scale

GAITHERSBURG, Md.--(BUSINESS WIRE)--Mar. 13, 2025-- GeneDx (Nasdaq: WGS), a leader in delivering improved health outcomes through genomic insights, announced today its scientific contributions will be presented at the 2025 American College of Medical Genetics and Genomics (ACMG) Annual Clinical Genetics Meeting. GeneDx will showcase its leadership in genomic research and innovation, driving the industry forward with three platform presentations and two poster presentations.

GeneDx earned one of the limited spots available for an industry-specific platform presentation on its AI-powered gene ranker embedded in its interpretation platform to enhance the speed and efficiency of clinical analysis. Through the combination of GeneDx's industry leading proprietary dataset, publicly available data, and the power of AI, Multiscore prioritizes genes with positive findings in exome and genome sequences by ranking them based on alignment with a patient's clinical presentation. GeneDx's AI-powered gene ranker accelerates analysis, reducing turnaround times and costs for patients and ultimately increasing access to care.

- **Multiscore, a Gene Ranker Powered by Artificial Intelligence and Real-World Clinical Data, Shows High Sensitivity in 10,000 Exomes and Genomes** will be presented by Vinnie Ustach, PhD (GeneDx) on Thursday, March 20, 2025 at 12:00 pm PT in Meeting Room: Petree Hall D

Featured as a Top 20 Poster, GeneDx will also present findings from an RNA sequencing program used to aid in variant of uncertain significance (VUS) resolution for patients that received exome-based testing for rare disease.

- **Clinical Impact of RNA Sequencing on VUS Resolution in a Diverse Rare Disease Cohort of Over 100,000 Patients** will be presented by Melanie Napier, MSc, MSc, CGC, CCGC (GeneDx) on Thursday, March 20 at 12:00 pm PT in the Exhibit Hall Learning Lounge and Friday, March 21 at 10:30 am PT at West Exhibit Hall A

"GeneDx's research, collaborations and AI-powered innovations being presented at ACMG reflect our commitment to advancing the utilization of exome and genome testing to improve patient care," said Dr. Paul Kruszka, MD, FACMG, Chief Medical Officer at GeneDx. "By investing in industry-changing research and harnessing cutting-edge technology, we are enhancing the speed, accuracy, and impact of genetic insights, ultimately enabling a precise diagnosis, faster and better outcomes for patients, and also driving cost efficiency."

GeneDx leverages its industry-leading dataset, exome and genome across numerous studies to advance clinical care for pediatric patients and showcase strong evidence to expand utilization. Through strategic collaborations with PacBio, GUARDIAN, and Seqfirst, GeneDx champions research to drive forward exome and genome sequencing as the standard of care in diverse clinical settings.

GeneDx collaborated on the following posters and presentations:

- **Resolution of large complex VNTRs with HiFi sequencing: applications in LPA Kringle IV-type 2 repeat and D4Z4 repeat** will be presented by Xiao Chen, PhD on Thursday, March 20 at 12:00 pm PT in Meeting Room: Petree Hall D
- **GUARDIAN Expanded Newborn Screening Study: Early Experiences** will be presented by Brenna Boyd, MS, CGC on Friday, March 21 at 1:30 pm PT in Meeting Room: 501 ABC
- **Parental needs during pediatric whole genome sequencing for developmental disorders: an interview study** will be presented by Priyanka Murali, MS, CGC on Thursday, March 20 at 10:30 am PT in West Exhibit Hall A

About GeneDx:
At GeneDx (Nasdaq: WGS), we believe that everyone deserves personalized, targeted medical care—and that it all begins with a genetic diagnosis. Fueled by one of the world's largest, rare disease data sets, our industry-leading exome and genome tests translate complex genomic data into clinical answers that unlock personalized health plans, accelerate drug discovery, and improve health system efficiencies. For more information, please visit genedx.com and connect with us on LinkedIn, Facebook, and Instagram.

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