GeneDx to Showcase Data from Largest Genomic Newborn Screening Cohort of 14,000 Newborns

October 8, 2024

Data to be presented at the International Conference on Newborn Sequencing (ICoNS) highlighting lessons learned from the GUARDIAN and Early Check newborn sequencing studies showcasing GeneDx's deep expertise as the leader for gNBS

STAMFORD, Conn.--(BUSINESS WIRE)--Oct. 8, 2024-- GeneDx (Nasdaq: WGS), a leader in delivering improved health outcomes through genomic insights, today announced it has provided genomic newborn screenings (gNBS) for more than 14,000 infants through its participation in groundbreaking research studies which aim to explore the clinical utility and implementation of utilizing genome sequencing to expand standard newborn screening (NBS). Through these studies, GeneDx has conducted more gNBS than any other lab and gained experience understanding diverse multi-site implementation strategies, positioning GeneDx as a laboratory leader set to revolutionize the standard approach to NBS.

Early diagnosis is critical to ensuring patients have the best chance at fighting the progression of disease. The broad availability of gNBS can significantly decrease time-to-diagnosis, which is particularly critical for diagnoses with actionable outcomes, including associated treatments and/or interventions. Through a retrospective analysis of its large database - currently more than 700,000 exomes and genomes - GeneDx looked to identify positive findings that would have been reported at birth had gNBS been available to patients as newborns. This analysis showed that greater than 21% of patients would have received a diagnosis earlier, on average by more than 8 years.

As the lab behind industry moving research studies, including GUARDIAN (Genomic Uniform-screening Against Rare Diseases In All Newborns), which has served more than 10,000 infants, and its early involvement in Early Check, supporting 2,000 infants, GeneDx has a unique understanding of the complexities of offering gNBS at scale. Interpretation of gNBS is heavily dependent on having a robust and diverse database of clinically significant variants to reduce the burden of analysis. Additionally, expertise in technical evaluation and understanding sequencing data outputs are necessary to inform variant interpretation and deliver a more definitive diagnosis.

"GeneDx's participation in GUARDIAN and Early Check allowed us to not only help end the diagnostic odyssey by delivering actionable diagnoses for hundreds of newborns before symptoms ever started, but also gave us the opportunity to showcase the power and impact of genomic newborn screening in this setting," said Dr. Paul Kruszka, MD, FACMG, Chief Medical Officer at GeneDx. "Over the past few years science and technology have allowed us to make remarkable strides that both lower costs and improve turnaround time that allows GeneDx to bring genomic newborn screening to families at scale today."

GeneDx is proud to partner with Columbia University Irving Medical Center, New York-Presbyterian and the New York State Department of Health on the GUARDIAN study, and with the North Carolina State Laboratory of Public Health and the University of North Carolina at Chapel Hill for the Early Check study.

Dr. Kruszka is set to present "Lessons Learned: 14,000+ Infants Screened with Newborn Sequencing [1]" at the International Conference on Newborn Sequencing (ICoNS) this week in New York City.

GeneDx's ongoing work to support research within gNBS underscores the company's vision for a world where any genetic disorder is diagnosed quickly to prevent disease progression and ensure long and healthy lives for all. As gNBS continues to advance, GeneDx remains dedicated to being at the forefront by harnessing genomic insights for early disease detection and one day ending the diagnostic odyssey.

For more information about GeneDx and our pioneering work in gNBS, visit genedx.com/newborn-screening.

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.

[1] The findings and conclusions in this presentation are those of the authors and do not necessarily represent the views of the North Carolina Department of Health and Human Services, Division of Public Health

View source version on businesswire.com: https://www.businesswire.com/news/home/20241008301393/en/

Press@genedx.com
Investors@genedx.com

Source: GeneDx