GeneDx Presents New Data at ACMG Demonstrating the Benefits of Exome Sequencing Over Chromosomal Microarray

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New research released at ACMG Annual Clinical Genetics Meeting reiterates the superiority of exome sequencing's role in shortening the diagnostic odyssey

STAMFORD, Conn., March 16, 2023 (GLOBE NEWSWIRE) -- GeneDx Holdings Corp. (GeneDx) (Nasdaq: WGS), a leader in delivering improved health outcomes through genomic and clinical insights, announced new research today at the American College of Medical Genetics and Genomics (ACMG) Annual Clinical Genetics Meeting which demonstrates exome sequencing's diagnostic advantages in shortening patients' and families' diagnostic odysseys when compared with chromosomal microarray (CMA).

To determine whether exome sequencing is comparable to CMA with respect to copy number variant (CNV) yield, GeneDx compared the reported CNV results on over 8,000 probands who had CMA and exome sequencing between May 2019 and May 2022. Overall, exome had a CNV detection rate of 93.4%, factoring in that some CMA-reported CNVs were detected by exome sequencing but not reported due to lack of phenotypic fit. Additionally, for CMA negative cases, exome sequencing found a CNV in 1.3% of these cases, mostly due to limitations in CMA probe coverage.

CNVs represent regions of the genome that can vary in the number of copies present between individuals, often leading to neurodevelopmental disorders and for which CMA had historically been considered the gold-standard for detection. This research shows exome sequencing's value in detecting both large and small CNVs, including insertions, deletions and duplications of DNA segments, compared to CMA. These data, coupled with industry guidelines stating that exome should be considered a first-tier test for individuals with congenital anomalies, developmental delay, or intellectual disability, and the increasing and expanding payor coverage, can assist providers in more confidently ordering exome testing, reducing the number of tests ordered per patient and expediting the time to diagnosis.

"Families of rare disease patients need answers, and providers should be empowered to order the most informative tests first in order to shorten the diagnostic odyssey," said Dr. Paul Kruszka, Chief Medical Officer of GeneDx. "Exome sequencing has a much higher diagnostic yield as it covers both sequence variants and CNVs, whereas CMA is limited to detecting only CNVs. Innovating testing procedures and better informing providers about their options will help improve diagnosis rates and accelerate patients' journeys from symptom onset to treatment plans."

GeneDx is dedicated to eradicating the multi-year diagnostic odyssey faced by many families seeking clinical diagnoses. The company has the world's largest rare-disease dataset, including more than 400,000 clinical exomes, 2.7 million phenotypes and over 1 million patient samples, which allows it to interpret genetic findings at scale.

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, 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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