GeneDx Announces the Sequencing of More Than 300,000 Clinical Patient Exomes

Largest clinical dataset of its kind driving diagnosis and discovery worldwide

GAITHERSBURG, MD, Sept. 21, 2021 – GeneDx, Inc., a leader in genomic analysis, a wholly owned subsidiary of BioReference Laboratories, Inc., an OPKO Health company (NASDAQ:OPK), today announced it has completed clinical genetic exome sequencing for more than 300,000 patients, making the company's dataset the largest of its kind in the world. Supported by matching phenotypes, the company's market-leading genomic analysis and interpretation capabilities have created a diagnosis and discovery engine advancing genetic medicine worldwide.

"The transition to widespread use of exome and ultimately whole genome sequencing rather than multi-gene panels promises to radically simplify the use of genomic information in healthcare. The rapid acceleration in demand for our exome testing, including nearly 100,000 exomes completed in the last year alone, shows this transition is well underway," said Katherine Stueland, president and CEO of GeneDx. "Often our patients are infants in the neonatal intensive care unit whose parents are facing a daunting process to figure out what's wrong with their child. Going immediately to sequencing the genomes of baby and parents dramatically curtails that quest, enabling the care team to have the right answer on what's wrong so they can intervene quickly."

With the volume of clinical exome sequencing accelerating more than 40 percent annually, GeneDx has played a pivotal role in supporting rare disease diagnosis for hundreds of thousands of patients while also expanding understanding of gene-disease relationships that improve diagnosis and treatment for patients worldwide. The company's extensive experience with exome sequencing results in a definitive diagnosis in 20% more cases with 27% fewer variants of unknown significance compared to public datasets.[1] The variant interpretation framework that supports the company's exome service also underlies its rapid whole genome sequencing service, which provides a complete genetic picture of a patient in seven days or less.

In addition to best-in-class interpretation and classification, GeneDx's interpretation capabilities and dataset have been an important discovery engine. Nearly a quarter of cases diagnosed today include gene-disease relationships originally identified by GeneDx, and each year company researchers collaborate on dozens of publications of new gene-disease relationships, significantly expanding medical understanding of the genetic underpinnings of developmental delay, intellectual disability and other genetic diseases.

Exome and whole genome sequencing are genetic tests designed to provide a comprehensive look at an individual's DNA. The broadest test, whole genome sequencing, analyzes the full genetic sequence of an individual, allowing clinicians to assess any variants that may be associated with health concerns. Exome sequencing concentrates on a subset of the genome, analyzing the roughly 20,000 protein-coding genes that are the source of most genetically-linked health issues. Each type of testing provides more complete information compared to the targeted multi-gene panels clinicians typically use to look for genetic underpinnings of health conditions. The American College of Medical Genetics and Genomics recently published guidelines strongly recommending exome or genome sequencing as a first or second-line test in children with intellectual disability, developmental delay or multiple congenital abnormalities.[2]

About GeneDx

GeneDx, Inc. is a global leader in genomics, providing testing to patients and their families from more than 55 countries. Originally founded by scientists from the National Institutes of Health, GeneDx offers a world-renowned clinical genomics program with particular expertise in rare and ultra-rare genetic disorders. In addition to its market-leading exome sequencing service, GeneDx offers a suite of additional genetic testing services, including diagnostic testing for hereditary cancers, cardiac, mitochondrial, neurological disorders, prenatal diagnostics, and targeted variant testing. GeneDx is a subsidiary of BioReference Laboratories, Inc., a wholly owned subsidiary of OPKO Health, Inc. To learn more, please visit http://www.genedx.com.

About OPKO Health
OPKO is a multinational biopharmaceutical and diagnostics company that seeks to establish industry-leading positions in large, rapidly growing markets by leveraging its discovery, development, and commercialization expertise and novel and proprietary technologies. For more information, visit www.opko.com.

Cautionary Statement Regarding Forward-Looking Statements
This press release contains "forward-looking statements," as that term is defined under the Private Securities Litigation Reform Act of 1995 (PSLRA), which statements may be identified by words such as "expects," "plans," "projects," "will," "may," "anticipates," "believes," "should," "intends," "estimates," and other words of similar meaning, including statements regarding GeneDx's test offerings, the adoption and widespread use of exome and whole genome sequencing, the utility and accuracy of exome and whole genome sequencing, the ability to provide rapid whole genome interpretation within seven days or less, whether GeneDx's interpretation capabilities and dataset will be an important discovery engine, whether healthcare providers will be able to properly treat conditions based on the results of exome and whole genome sequencing, as well as other non-historical statements about our expectations, beliefs or intentions regarding our business, technologies and products, financial condition, strategies or prospects. Many factors could cause our actual activities or results to differ materially from the activities and results anticipated in forward-looking statements. These factors include those described in the OPKO Health, Inc. Annual Reports on Form 10-K filed and to be filed with the Securities and Exchange Commission and in its other filings with the Securities and Exchange Commission. In addition, forward-looking statements may also be adversely affected by equipment and reagent shortages, general market factors, competitive product development, product availability, federal and state regulations and legislation, the regulatory process for new products, manufacturing issues that may arise, patent positions and litigation, among other factors. The forward-looking statements contained in this press release speak only as of the date the statements were made, and we do not undertake any obligation to update forward-looking statements. We intend that all forward-looking statements be subject to the safe-harbor provisions of the PSLRA.

References:

1. Data on file
2. Manickam, K., McClain, M.R., Demmer, L.A. *et al.* Exome and genome sequencing for pediatric patients with congenital anomalies or intellectual disability: an evidence-based clinical guideline of the American College of Medical Genetics and Genomics (ACMG). *Genet Med* (2021). https://doi.org/10.1038/s41436-021-01242-6

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