

June 12, 2018



GeneDx Announces Completion of 100,000 Exome Sequences

Milestone achievement offers significant contribution to clinically relevant genetic variants used to diagnose rare and genetic diseases

GAITHERSBURG, Md., June 12, 2018 (GLOBE NEWSWIRE) -- **GeneDx, Inc.**, a subsidiary of OPKO's BioReference Laboratories and a recognized leader in genetic testing, today announced that the Company has performed clinical Exome Sequencing on more than 100,000 individuals. This Company milestone represents one of the largest cohorts of sequenced exomes by an independent clinical laboratory in the world. It also greatly expands the database of known clinically relevant genetic variants used as tools to help diagnose rare diseases.

"This milestone is a huge step forward in patient care," said Dr. Jane Juusola, Director of Clinical Genomics at GeneDx. "GeneDx was founded with the mission to help patients and families by making genetic testing available to as many people as possible. This large and unique data set will further help in providing answers for those who might otherwise go undiagnosed."

Exome sequencing is a process by which the protein-coding genes in the human genome are sequenced. It is used to help diagnose disease by identifying variations in these genes that may lead to a variety of neurological, metabolic or other disorders. In total there are about 20,000 genes.

Over the past three years, GeneDx has helped discover and publish more than 62 disease-causing genes, thereby contributing to the phenotypic, or observable characteristics, involved in identifying and further understanding developmental delay and intellectual disability. GeneDx has grown in exome volume by a compound annual growth rate of 55% since its inception. With over 100 professionals dedicated to exomes and genomes, including MDs, PhDs and genetic counselors, GeneDx has developed a suite of patient-focused exome and genome tests for a variety of clinical indications and scenarios, including family-based testing, rapid testing for critically ill patients, prenatal testing and exome testing in parallel with mitochondrial genetic testing.

"We are very excited about what we have accomplished and what we continue to do each day," said Dr. Ben Solomon, Managing Director of GeneDx. "This expanded data set, along with our proprietary state-of-the-art variant analysis software, allows us to continually provide patients and clinicians with highly accurate variant interpretation results that can help make a definitive diagnosis possible in very complex cases."

For GeneDx's complete list of testing options, please visit its website www.genedx.com or email zebras@genedx.com.

About GeneDx

GeneDx is a global leader in genomics, providing testing to patients and their families in more than 55 countries. Led by its world-renowned whole exome sequencing program, GeneDx has an acknowledged expertise in rare and ultra-rare genetic disorders, as well as one of the broadest menus of sequencing services available among commercial laboratories. 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 www.genedx.com.

Contact

Ben Solomon, MD, FACMG, 301-519-2100
bsolomon@genedx.com
Managing Director, GeneDx



Source: OPKO Health, Inc.