Study by New York Genome Center and IBM Demonstrates Potential for AI and Whole Genome Sequencing to Scale Access to Precision Medicine

NEW YORK, NY - 11 Jul 2017: In a study published today in the July 11, 2017 issue of Neurology® Genetics, an official journal of the American Academy of Neurology, researchers at the New York Genome Center (NYGC), The Rockefeller University and other NYGC member institutions, and IBM (NYSE: IBM) bhave illustrated the potential of IBM Watson for Genomics to analyze complex genomic data from state-of-the-art DNA sequencing of whole genomes. The study compared multiple techniques – or assays – used to analyze genomic data from a glioblastoma patient's tumor cells and normal healthy cells.

The proof of concept study used a beta version of Watson for Genomics technology to help interpret whole genome sequencing (WGS) data for one patient. In the study, Watson was able to provide a report of potential clinically actionable insights within 10 minutes, compared to 160 hours of human analysis and curation required to arrive at similar conclusions for this patient.

The study also showed that WGS identified more clinically actionable mutations than the current standard of examining a limited subset of genes, known as a targeted panel. WGS currently requires significantly more manual analysis, so combining this method with artificial intelligence could help doctors identify potential therapies from WGS for more patients in less time.

Interpretation of genome sequencing data is a significant challenge because of the volume of genomic data to sift through, as well as the large, growing body of research on molecular drivers of cancer and potential targeted therapies. This informatics challenge is often a critical bottleneck when dealing with deadly cancers such as glioblastoma, with a median survival of less than 15 months following diagnosis.

"Our partnership has explored cutting-edge challenges and opportunities in harnessing genomics to help cancer patients. We provide initial insights into two critical issues: what clinical value can be extracted from different commercial and academic cancer genomic platforms, and how to think about scaling access to that value," noted the study's Principal Investigator, Robert Darnell, MD, PhD, Robert and Harriet Heilbrunn Professor and Senior Attending Physician at The Rockefeller University and Founding Director of the New York Genome Center.

In the study, NYGC researchers and bioinformatics experts analyzed DNA and RNA from a glioblastoma tumor specimen and DNA from the patient's normal blood, and compared potentially actionable insights to those derived from a commercial targeted panel that had previously been performed. The whole genome and RNA sequencing data were analyzed by a team of bioinformaticians and oncologists at the NYGC as well as a beta version of IBM Watson for Genomics, an automated system for prioritizing somatic variants and identifying potential therapies.

The beta version of Watson for Genomics processed abstracts and in some cases, full text articles from PubMed, a comprehensive source of more than 27 million citations for biomedical literature. With this information, the NYGC and Watson collaborated to identify gene alterations that can be therapeutically targeted.

"This study documents the strong potential of Watson for Genomics to help clinicians scale precision oncology more broadly," said Vanessa Michelini, Watson for Genomics Innovation Leader, IBM Watson Health. "Clinical and research leaders in cancer genomics are making tremendous progress towards bringing precision medicine to cancer patients, but genomic data interpretation is a significant obstacle, and that's where Watson can help."

The study was part of the NYGC's and its Institutional Founding Members' ongoing efforts to advance the use of next-generation sequencing, particularly WGS, in precision medicine. The NYGC and its founding member institutions are conducting additional studies involving Watson to help accelerate the discovery of potentially actionable sequence variants in various types of cancer, including an ongoing study that involves DNA and RNA from a larger cohort of glioblastoma patients, and a study of 200 patients with different types of cancer.

This study, conducted from 2015-2016, utilized a beta version of Watson for Genomics, which is now commercially available for genomic data interpretation through partnerships with Quest Diagnostics, Illumina, or as a cloud-based software for clinicians and researchers. Watson for Genomics is also used in clinical practice at the VA Health System.

About the New York Genome Center

The New York Genome Center is an independent, nonprofit academic research institution at the forefront of transforming biomedical research with the mission of advancing clinical care. A collaboration of premier academic, medical and industry leaders across the globe, the New York Genome Center has as its goal to translate genomic research into the development of new treatments, therapies and therapeutics against human disease. NYGC member organizations and partners are united in this unprecedented collaboration of technology, science and medicine, designed to harness the power of innovation and discoveries to advance genomic services. Their shared objective is the acceleration of medical genomics and precision medicine to benefit patients around the world. For more information, visit our website at http://www.nygenome.org.

Member institutions include: Albert Einstein College of Medicine, American Museum of Natural History, Cold Spring Harbor Laboratory, Columbia University, Hospital for Special Surgery, The Jackson Laboratory, Memorial Sloan Kettering Cancer Center, Icahn School of Medicine at Mount Sinai, NewYork-Presbyterian Hospital, The New York Stem Cell Foundation, New York University, Northwell Health, Princeton University, The Rockefeller University, Roswell Park Cancer Institute, Stony Brook University, Weill Cornell Medicine and IBM.

About IBM Watson Health

Watson is the first commercially available cognitive computing capability representing a new era in computing. The system, delivered through the cloud, analyzes high volumes of data, understands complex questions posed in natural language, and proposes evidence-based answers. Watson continuously learns, gaining in value and knowledge over time, from previous interactions. In April 2015, the company launched IBM Watson Health and the Watson Health Core cloud platform (now Watson Platform for Health). The new unit will help improve the ability of doctors, researchers and insurers to innovate by surfacing insights from the massive amount of personal health data being created and shared daily. The Watson Platform for Health can mask patient identities and allow for information to be shared and combined with a dynamic and constantly growing aggregated view of clinical, research and social health data. For more information on IBM Watson,

visit: ibm.com/watson. For more information on IBM Watson Health, visit: ibm.com/watsonhealth.

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