Communiqués de presse

Une étude réalisée par le New York Genome Center et IBM démontre le potentiel de l'IA pour le séquençage complet du génome afin d'améliorer l'accès à la médecine de précision

Dans une étude publiée dans le numéro du 11 juillet 2017 de Neurology® Genetics, journal officiel de l'American Academy of Neurology, les chercheurs du New York Genome Center (NYGC), The Rockefeller University, d'autres institutions membres du NYGC et IBM ont illustré le potentiel d'IBM Watson pour la génomique à analyser des données génomiques complexes à partir d'un séquençage d'ADN de pointe de génomes entiers. L'étude a comparé des techniques multiples - ou des tests - utilisés pour analyser les données génomiques provenant des cellules tumorales d'un glioblastome et des cellules saines normales.

Armonk - 11 juil. 2011: Dans une étude publiée dans le numéro du 11 juillet 2017 de Neurology® Genetics, journal officiel de l'American Academy of Neurology, les chercheurs du New York Genome Center (NYGC), The Rockefeller University, d'autres institutions membres du NYGC et IBM ont illustré le potentiel d'IBM Watson pour la génomique à analyser des données génomiques complexes à partir d'un séquençage d'ADN de pointe de génomes entiers. L'étude a comparé des techniques multiples - ou des tests - utilisés pour analyser les données génomiques provenant des cellules tumorales d'un glioblastome et des cellules saines normales.

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 (July 11, 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 have 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.