Genomics

In many next-generation sequencing laboratories around the world, the variant calling format (VCF) file is curated by expert personnel using manual or semi-automated processes. These processes require familiarity with ever-expanding literature pertaining to the significance of specific mutations as well as the availability of therapeutic options that may target those mutations—whether those therapeutic options are FDA approved or investigational.

While this process is carried out effectively in most laboratories through manual curation, studies below demonstrate that Watson for Genomics (WfG) can classify and categorize actionable variants with great accuracy and in less time than is required when performed with conventional methodology. For example, one study found that Watson for Genomics accurately interpreted whole genome sequencing data for a glioblastoma patient in 10 minutes.

Other evidence indicates that WfG can identify new potentially actionable variants that were not identified manually.[21] With the rise of precision medicine, technologies that can quickly associate genomic data with potential therapeutic options play an important role in supporting oncology care providers in evaluating treatment options.

21 Patel N et al. Enhancing next-generation sequencing-guided cancer care through cognitive computing [published online November 20, 2017]. Oncologist. 2018;23(2): 179-185. doi: 10.1634/theoncologist.2017-0170

Clinical insights for hematological malignancies from an artificial intelligence decision-support tool

Kim M et al. ASCO Annual Meeting 2019

Link to study →

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WfG variant interpretation correlated well with manually curated expert opinion and identified clinically actionable insights missed by manual interpretation... WfG has obviated the need for labor-intensive manual curation of clinical trials and therapy, enabling our center to exponentially scale our NGS operations.

Excerpt from abstract

54
South Korean patient cases with hematological malignancies were analyzed by Watson for Genomics (WfG)

10
cases were randomly selected for manual interpretation analysis

71%
of cases had at least one clinically actionable therapeutic alteration

33%
of cases had genes that were targeted by a US FDA approved therapy

20%
of cases without therapeutic alterations, WfG identified additional diagnostic or prognostic insights

90%
of the manually interpreted cases were concordant with WfG analysis

WfG identified 9 more (33%) clinically actionable variants not found in manual assessment.

Enhancing NGS-guided cancer center care through cognitive computing

Patel N et al. The Oncologist. 2018;23(2):179-185

Link to study →

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Molecular tumor boards empowered by cognitive computing can significantly improve patient care by providing a fast, cost-effective, and comprehensive approach for data analysis in the delivery of precision medicine.

Excerpt from abstract

1,018

Watson for Genomics analyzed 1,018 patient cases previously sequenced and analyzed

Providing current, accurate information on newly approved therapeutic options and open clinical trials requires considerable manual curation performed mainly by members of molecular tumor boards (MTBs).

Watson for Genomics’ automated analysis of genomic data took under 3 minutes per patient case.

under 3 min

In 32% of the patient cases, Watson for Genomics found additional potentially clinically actionable variants that a molecular tumor board had not identified.

32%

Comparative analysis of target gene exon sequencing by cognitive technology using next generation sequencing platform in Chinese patients with lung cancer*

Zhang et al. ASCO Annual Meeting 2018
*no contributing IBM author

Link to study →

"These findings suggest the unique role for cognitive computing in the detection of genetic alterations which may inform opportunities for investigational targeted cancer therapies.

Excerpt from abstract

115
tissue samples

Study compared annotation and reporting differences between an actionable mutation list generated by Watson for Genomics to the list generated by a bioinformatics molecular tumor team at Guangdong Lung Cancer Institute.

Watson identified an average of 1.54 additional mutations

Watson found the same mutations as manual annotation