Large study supports use of whole genome sequencing in standard cancer care

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In the largest study of its kind, scientists report how combining health data with whole genome sequence (WGS) data in patients with cancer can help doctors provide more tailored care for their patients.

The research, published in *Nature Medicine*, shows that linking WGS data to real-world clinical data can identify changes in cancer DNA that may be relevant for an individual patient's care, for example by helping identify what treatment might work best for them based on their cancer.

The study, led by Genomics England, NHS England, Queen Mary University of London, Guy's and St Thomas' NHS Foundation Trust and the University of Westminster, analyzed data covering over 30 types of solid tumors collected from more than 13,000 participants with cancer in the 100,000 Genomes Project. By looking at the genomic data alongside routine clinical data collected from participants over a 5-year period, such as hospital visits and the type of treatment they received, scientists were able to find specific genetic changes in the cancer associated with better or worse survival rates and improved patient outcomes.

The study showed that WGS could provide a more comprehensive view of a tumor's genetic landscape by detecting various genetic changes using a single test. This research uncovered significant findings across different cancer types, such as:

- Over 90% of brain tumors and over 50% of colon and lung cancers showed genetic changes that could affect how patients are treated, guiding decisions about surgery or specific treatments they might need.
- In more than 10% of sarcomas, larger DNA changes, known as

structural variants, were identified that can impact clinical care and treatment.
- In over 10% of ovarian cancers, the study pinpointed inherited risks offering crucial insights for clinical care.

The analysis also revealed patterns across several cancers and uncovered different types of genetic changes that might explain response to treatment or predict possible patient outcomes. Together, the findings show the value of combining genomic and clinical data at scale to help health care professionals make the best treatment decisions with their patients.

WGS allows physicians and researchers to read someone's entire genome—the 3.2 billion letters that make up our DNA—with just one single test. For patients with cancer, this technique can be used to compare DNA from their tumor to the DNA in their healthy tissues.

The 100,000 Genomes Project laid the foundations for the NHS to become the first national health system to offer WGS as part of routine care via the NHS Genomic Medicine ServiceThis study shows the value of investment in national infrastructure to generate clinical and genomic data at scale on patients and participants who consent to research in an NHS setting—allowing researchers to uncover insights to drive improved diagnosis, care and treatment for patients with cancer.

Dr. Nirupa Murugaesu, Principal Clinician, Cancer Genomics and Clinical Studies at Genomics England, Oncology Consultant and Cancer Genomics Lead at Guy's and St Thomas's NHS Foundation Trust, said, "This study is an important milestone in genomic medicine. We are starting to realize the promise of precision oncology that was envisioned ten years ago when the 100,000 Genomes Project was launched.

"We are showing how cancer genomics can be incorporated into

mainstream cancer care across a national health system and the benefits that can bring patients. By collecting long-term clinical data alongside genomic data, the study has created a first-of-its-kind resource for clinicians to better predict outcomes and tailor treatments, which will allow them to inform, prepare, and manage the expectations of patients more effectively."

Professor Sir Mark Caulfield, VP Health at Queen Mary University of London, added, "This study shows how we can transform cancer care from a 'one size fits all' approach into precision health care and defines specific genomic signatures that predict treatment response and outcomes, which may usher in expanded use of whole genomes for cancer care."

Dr. Alona Sosinsky, Scientific Director for Cancer at Genomics England, noted, "The 100,000 Genomes Project paved the way for delivering whole genome sequencing in cancer. This technology opens tremendous opportunities for precision oncology. Together with centrally collected clinical data our genomic cohort provides a rich dataset for basic and translational research. In this study, we demonstrate how Genomics England's unique national genomic research dataset can provide vital information to inform genomic testing in health care."

Helen White, Participant Panel Vice Chair for Cancer at Genomics England, observed, "This study has opened doors to better ways of diagnosing and treating cancer, but was only made possible by all those who, despite the challenges of a cancer diagnosis, gave their consent for their or their loved one's genomic and health data to be used in the 100,000 Genomes Project. Their invaluable contribution has made these advances possible, bringing hope to people affected by cancer of living well for longer."

Professor Dame Sue Hill, Chief Scientific Officer for NHS England and

Senior Responsible Officer for Genomics, stated, "With this new study, data from the 100,000 Genomes Project continues to build the evidence for the use of genomic testing to deliver precise molecular diagnoses to inform personalized treatments and interventions for patients.

"The insights gained in this study, in which genomic patterns or profiles have been mapped out in thousands of patients with different types of cancer, support and inform the NHS Genomic Medicine Service in providing a comprehensive genomic testing service for patients with cancer and signals a promising future for health care as we continue to hone and enhance the NHS use of genomics and tailor interventions for improved outcomes."

Andrew Stephenson, Health Minister, concluded, "This ground-breaking research demonstrates the power of genomics and is already helping to transform care, allowing patients to receive more tailored treatment and driving improved diagnosis. But we won't stop there. We want to harness this innovation to improve care for patients and affirm our position as a life sciences superpower—leading to quicker and more targeted interventions."

More information: Insights for precision oncology from the integration of genomic and clinical data of 13,880 tumors from the 100,000 Genomes Cancer Programme, *Nature Medicine* (2024). DOI: 10.1038/s41591-023-02682-0

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