Enabling Genomics-Guided Precision Medicine with Real-World Evidence

by Sean P. Scott

Molecular diagnostics testing plays a key role in precision medicine. It is embedded at every stage of the treatment pathway – from providing reliable information that informs the optimal treatment for the individual patient to precise monitoring of its effectiveness. A growing number of healthcare organization services (HCOs) and physicians are adopting molecular diagnostics to diagnose or predict susceptibility for inherited conditions, determine carrier status, screen for common disorders, predict drug response, select appropriate treatments, and monitor disease recurrence.

The introduction and expansion of molecular diagnostic and next-generation sequencing (NGS) technologies has enabled more patients to undergo testing for rare genetic diseases, such as cancer. Yet, approximately 10 percent of the global population is affected by rare genetic diseases, underscoring the need for improved pre-conception carrier screening, non-invasive prenatal testing (NIPT), pediatric rare disease testing, hereditary cancer risk assessment, and somatic cancer testing. For this to happen, genetic testing must become more accessible, affordable and consistent.

In the past decade, substantial progress has been made in clinical testing technology, predicating the establishment of regulatory and professional practice standards for the clinical wet lab. Today, we are witnessing a shift away from instrumentation and towards insight, with more attention focused on standardizing data analysis, interpretation and reporting to clinical care. The ability to interpret genetic test results remains the rate limiting factor for the broad adoption of NGS testing. Interpretation is dependent upon the standardization of the entire Sample to Insight workflow, from sample collection to clinical report.

Commercial- and provider-based clinical testing laboratories are transitioning to NGS-based testing and soon-to-be exome-based testing. These advanced, decision-support technologies are helping clinical labs meet the growing demands and needs of ordering physicians, while remaining competitive within the market. This critical path has shifted away from sample collection, sequencing, and generating data, to a more insight-intrinsic approach that enables physicians to derive actionable information from clinical NGS test reports. For this to happen, physicians must have access to clinical and real-world evidence that is reported and assembled in accordance with professional and regulatory guidelines. While numerous public and private databases have emerged to store and share this clinical data, real-world evidence is less accessible. Real-world data is complex, diverse, and locked in provider systems, making it difficult to glean actionable information outside of what is available in public databases.

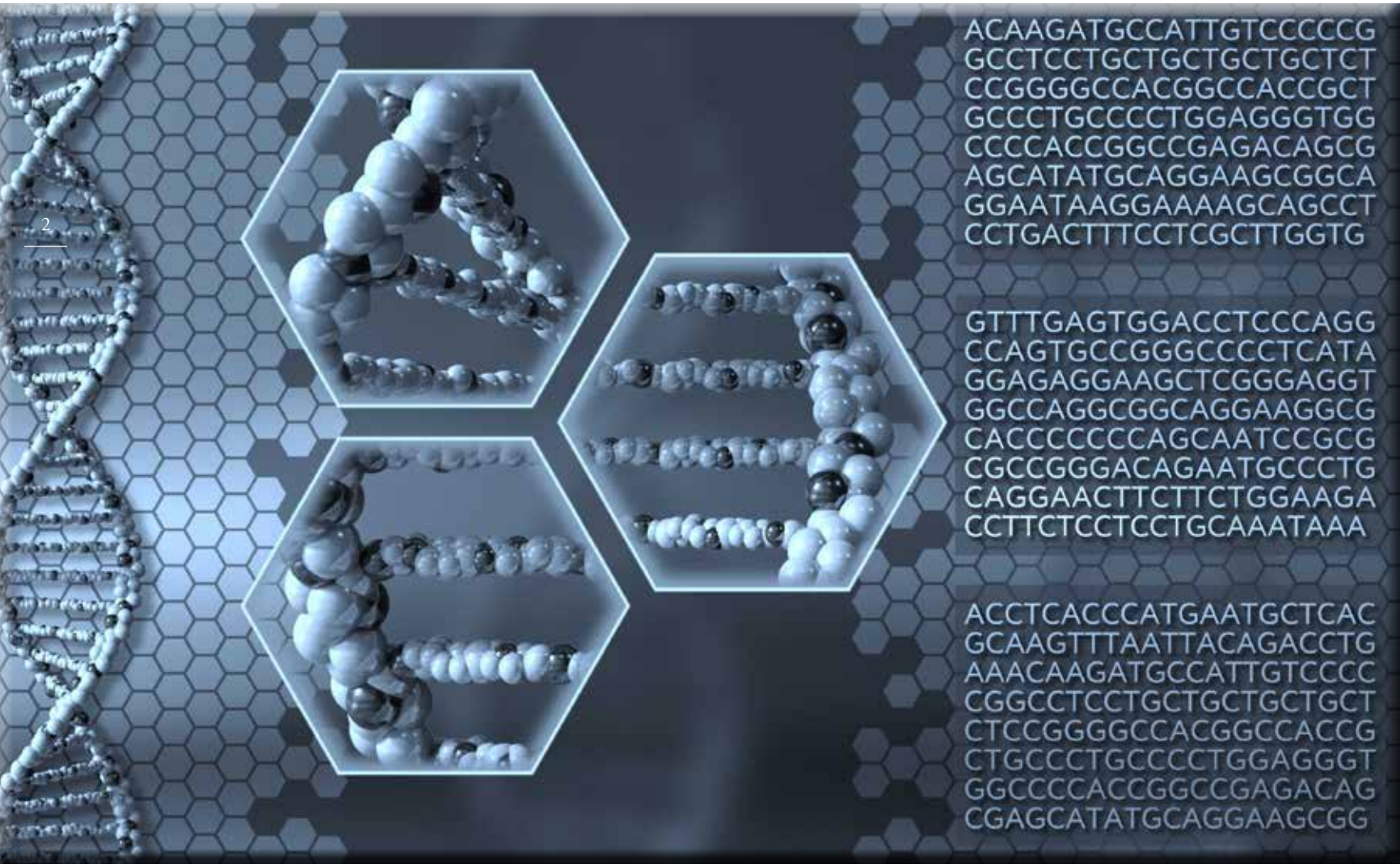
Real-world evidence is derived from the data of patients treated in real-world settings. The surge of electronic health records (EHRs), as well as the rise of wearables, is enabling researchers and providers to better understand the real-world patient experience. Genetic and biomarker data is increasingly being used to enhance traditional real-world evidence platforms to characterize disease pathways and establish associations with phenotypic expression. Aggregating and analyzing large, real-world evidence datasets, such as identifying targeted therapies and clinical trial options for a cancer patient with a unique genomic profile, has been instrumental in driving precision medicine forward. Now, HCOs and physicians have the opportunity to use real-world evidence datasets to identify even larger patient cohorts

with similar treatment decisions, treatment responses, patient outcomes and survival rates.

Across the healthcare sector, every role is being affected by this trend. From researchers involved in early stage drug discovery and development who can now study more specific subgroups of patients, through the FDA assessing the efficacy of a new drug within the approval process, to payers determining how to reimburse for off-label medications based upon available evidence and the expected patient benefits, to providers on the front lines of care, even to the patients themselves who want to understand more about their disease and potential treatments, the potential of real-world evidence in precision medicine is transformative.

The pharmaceutical industry has adopted genome-enabled drug discovery, and the market for molecular diagnostics has grown rapidly. There are now groundbreaking examples of the application of genomics across the stages of disease, from risk stratification and screening to diagnosis and prognosis of treatment. For some diseases, such as breast cancer, genome-based tools have been woven into clinical practice and disease management, but for most areas of medicine, the uptake of genomics has been slow.

The lynchpin is data-actionability. While a clinician's skill lies in having the expertise and judgment to evaluate every factor involved and making the correct call, the true barrier he or she faces – where evidence-based clinical decision support (CDS) tools become a critical ally – is the challenge of collecting, analyzing and using this data to guide treatment decisions especially in cases where there may be multiple variants involved and/or multiple treatment and clinical trial options. First-movers in genomics-guided precision medicine will be able to integrate data from all from multiple testing platforms, including real-world evidence, and combine it with patient clinical data from multiple sources to enable a 360-degree view of the patient and capture and enrich this data longitudinally.



Tapping into new real-world data sources of data – including biometric readings from wireless devices, patterns of health service use and costs from EHRs and medical claims, and insights on what people think and do drawn from their social media and internet use – can help bridge the divide between what's been shown to work in randomized controlled trials and what actually produces value as measured by patients, providers, and payers. Making sense of all this information is an enormous challenge: the amount of health care data grows by nearly 50 percent a year.[2] It requires a radically different set of analytic methods based on predictive analytics, machine learning, and artificial intelligence.

This type of integration requires a high level of experience and expertise – enter QIAGEN. A trusted partner for clinical laboratories worldwide, QIAGEN enables genomics-guided precision medicine. From sample collection tubes, to sample preparation, instrumentation, and standardized software that analyzes, interprets and reports NGS test results, QIAGEN supports each step of the workflow, helping labs embrace the potential of genomics medicine.

Sean P. Scott, Chief Business Officer and Vice President of Clinical Market Development at QIAGEN, explains the necessity and value of incorporating real-world evidence into genomics-guided precision medicine.

"There's no question that there is great promise for genomics-guided precision medicine in genetic diseases, and there is growing evidence of improved patient outcomes resulting from better testing technologies and evidence-based clinical decision support capabilities. But while the technology continues to advance, genomics-guided precision medicine will not become mainstream clinical practice until the data analysis, interpretation, reporting and decision-support challenges are addressed at scale and integrated with imaging data and clinical outcomes data," Scott says.

In his role with QIAGEN, Scott has been focused on solving the data interpretation challenge and enabling genomics-guided precision medicine capabilities in the clinical laboratory setting for the better part of the last decade. Whether the labs are large reference labs, commercial specialty labs, or small central labs within regional hospital systems, Scott and QIAGEN take a holistic "Sample to Insight" based approach to helping their customers develop best-in-class test menus and services that result in insightful and actionable results for the physicians and patients served by these laboratories. Mr. Scott's focus has recently shifted to helping HCOs develop genomics-guided precision medicine programs across their laboratories and physician groups. He is also working with partners in the pharmaceutical industry to identify and access real-world evidence datasets that better inform drug development, clinical trial protocol design, and patient cohort selections.

"Whether it's a small hospital network or a large commercial specialty lab, we're focused on helping them understand how to develop a more insightful and actionable report for the ordering physicians – detect and prevent disease or improve treatment roadmaps for patients that have failed first line care. This enables the physicians and the laboratories they serve to make better decisions for improved patient care and outcomes," Scott says.

Although Scott has been working on the cutting edge of genomics-guided precision medicine for more than a decade, the field is still a relatively new concept within the healthcare industry, he says. Therefore, leaders like Scott are critical to the successful and responsible implementation of NGS-based tests within today's clinical setting.
The healthcare industry is undergoing a dynamic shift away from DNA-based diagnostic testing towards theranostics and prognostics, and physicians are seeking supplemental information on treatment and trial options for their patients, Scott says. Thus, it is imperative that healthcare providers embrace the full potential of genomic data and standardize NGS testing according to professional association guidelines and institutional best practices.

"When you talk to the ordering physicians who are trying to draw insights or make treatment decisions based on genetic test results, there's still a large chasm between complete and accurate reports and reports that are insightful, actionable, or influential over the clinical decision-making process," Scott says.

Genomics-guided precision medicine promises to transform healthcare, but in order for this potential to be fully realized, providers, payers, regulators, and industry insiders must work collaboratively to ensure the seamless integration of this new technology across the clinical setting, Scott says. The building blocks for such a system are already forming, and healthcare providers need to develop capabilities for genomic integration, molecular profiling, and applying clinical outcomes data and real-world evidence at the point-of-care.

"You will not see true genomics-guided precision medicine capabilities within provider networks until they can figure out how to best integrate genomics data with clinical outcomes data and real-world evidence. I think that's the real challenge right now," Scott says. ■

References

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