Consider the tumor. Once viewed as a uniform wad of deadly malignancy, it’s now known as a collection of diverse cell types, driven by a wide range of genetic mutations and cooperating in a complicated ecosystem.

Scientists continue to identify more and more of these mutations, a crucial task in the search for more targeted, and potentially much more effective, therapies.

“There can be tremendous genetic diversity within a single tumor,” said Bryce Olson, Intel’s global marketing director for health and life sciences. That’s why they’ve been so hard to eradicate. The therapies in general use today—chemotherapy, radiation, and surgery—are broadbased. But with cancer, one size doesn’t fit all.

“We’ve needed a more targeted approach for a long time, one that addresses the genetic mutations that are driving cancer cells,” said Olson. “And we’re finally starting to get therapies that go after those mutations at the molecular level.”

The new treatments are based on genomic sequencing, which has only recently become cost-effective enough for clinical use. In 2003, the human genome was first sequenced for $2.7 billion. Today, the price of sequencing an entire genome is closer to $1,000, and costs continue to fall.

Sequencing identifies the key genetic mutations for which targeted therapies and immunotherapies can be used. These therapies attack the genetic vulnerabilities of individual tumor cells or supercharge the human immune system, yielding antibodies that can attack cancer at the molecular level. Results to date have ranged from promising to stellar, and Olson—an advanced-stage cancer survivor—can personally attest to that.

“The median survival time for advanced prostate cancer patients like me, who have bone metastases and are progressing after chemo, is 21 months,” he said. “I passed that milestone eight months ago.” Olson used genomic sequencing to identify and disrupt a mutated cell signaling pathway, after entering a clinical trial targeting that specific pathway.

Next-generation sequencing (NGS), bolstered by powerful data analytics and the clinical interpretation of genetic variants in a patient’s cancer, is on the cusp
of transforming healthcare. We are at a watershed moment in medical history, comparable to the discovery of microorganisms, the development of vaccines, and the creation of antibiotics.

Data is at the heart of this revolution. The value of NGS grows as more data is collected, and that will accrue only as more people are sequenced and their conditions are profiled. The progress of precision medicine now depends largely on getting that data from discoveries into the hands of the doctors on the front lines of patient care.

**Mastering data**

Oncologists are hard pressed to incorporate new genomic discoveries into everyday practice. They have precious little time to begin with: They’re grappling with an aging population plagued by chronic diseases, with no increase in time to spend educating patients on the new taxonomy of cancer that focuses on its molecular drivers.

Many oncologists don’t use genomic data simply because they lack the software tools, both visual and analytic, to quickly turn data into insights that can help identify the most appropriate therapies.

Compounding the problem, genomic sequencing results usually aren’t well integrated into electronic health records (EHRs) and clinical decision support systems. Different payers also have varying standards for assessing clinical utility, which leads to inconsistent coverage and reimbursement for NGS-based testing.

Finally, even with today’s potent technologies, molecular profiling and clinical interpretation take weeks—time that some patients may not have.

These issues pose a significant roadblock to the widespread adoption of genomics-based precision medicine. Providers can, however, advance the clinical application of precision medicine with a few basic strategies.

**Stopping the “cycle of inaction”**

First, providers and healthcare organizations can focus their efforts on areas with the biggest impact on care and where reimbursement is most likely. For now, that means primarily oncology, pediatrics, reproductive health, and pharmacogenomics, all of which have a larger known genetic component. Some EHRs have tools that can tell physicians which genetic tests are reimbursed and which require out-of-pocket payments from patients. As researchers continue to learn more about the genetics of other conditions, the list of high-impact areas will grow.

Organizations will also need to incorporate precision medicine into their governance bodies if they haven’t already done so. Making precision medicine a reality requires investment and likely major changes across the organization,

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—Jennifer Esposito
Worldwide General Manager, Health and Life Sciences, Intel
including stronger collaboration among clinicians, pathologists, and medical tumor boards. Cross-functional executive support at all stages is critical to successfully implementing these changes.

With that executive-sponsored cross-functional team, the next step is determining the right approach and engaging the necessary partners, from sequencing to bioinformatics to clinical decision support, and deciding how much to do internally.

For example, smaller hospitals might work with a commercial diagnostic test company that handles all the sequencing, analytics, and clinical decision support tools to bring actionable data to doctors. A larger hospital may find it more cost-effective to have its own lab for genetic testing, with annotated test results delivered to physicians at the point of care. Given the size of that investment—including personnel trained in DNA extraction, sequencer operation, bioinformatics, genomic data analysis, and related aspects—that course may only make sense for larger organizations.

Ultimately, providers will expect infrastructure and tools that enable their EHRs to include a real-time interface with genetic databanks, as well as designated fields for genetic variant information to ensure genomic data isn't relegated to easily overlooked text or comment fields. May organizations will also need more sophisticated algorithms and analytics to manage patients at the individual level.

"When more points of care have both genomic sequencing and data analytics at their fingertips, and a team skilled in the application of genomic methods in a clinical setting, more people will receive these new treatments," said Jennifer Esposito, Intel's worldwide general manager, health and life sciences.

The most significant stakeholders in NGS, of course, are the patients. Their interests must remain paramount as the genomic revolution evolves. Space must be created in the clinical framework for self-advocacy; when patients speak, physicians, researchers, and regulators alike must listen closely and accommodate demands.

Patients will need user-friendly educational materials to understand their options and possible outcomes. And as they learn more about precision medicine, more people will expect personalized treatment as a matter of course. Providers will risk losing patients if they don't have a solution that can handle the complexities of precision medicine, and that includes the right staff, instruments, hardware, and software.

That's why Intel has collaborated with QIAGEN, a company with more than three decades of experience in bioinformatics. The QIAGEN "sample-to-insight" solution supports the entire diagnostic workflow, from sample preparation and sequencing to data analysis, interpretation, and reporting, as well as expertise in clinical decision support with testing laboratories and the physicians they serve.

"THE SIZE OF GENOMIC DATASETS DOUBLES ABOUT EVERY EIGHT MONTHS."

—Erik Banks
Director of the Data Sciences and Data Engineering group at the Broad Institute

"JAMA Oncology, November 2016"
“Central to the Intel and QIAGEN collaboration is enabling molecular diagnostics labs and healthcare providers to develop and operationalize scalable, compliant, and secure molecular and genomics data analysis, interpretation, and reporting solutions to implement genomics-guided clinical decision support at the point of care,” said Sean Scott, QIAGEN’s VP, business development for genomics and bioinformatics.

The collaboration is yielding results.

A Danish hospital using the QIAGEN Clinical Insight software platform found clinically actionable mutations in 40 percent of advanced cancer patients who were running out of options. Half of that group received treatment plans that were less toxic and more likely to work.

Intel is also collaborating with the Broad Institute of MIT and Harvard to scale the ability to analyze massive amounts of genomic data from diverse sources worldwide.

“The size of genomic datasets doubles about every eight months and, as it does, the challenge of acquiring, processing, storing, and analyzing this information increases as well,” said Eric Banks, director of the Data Sciences and Data Engineering group at the Broad Institute.

“Working with Intel, we plan to build out solutions that can work across different infrastructures to facilitate efficient processing of these growing datasets, and then make these tools openly available for researchers worldwide,” said Banks. “Our work is a step toward building something analogous to a superhighway to connect disparate databases of genomic information for the advancement of research and precision medicine.”

This convergence of massive genomic datasets and bleeding-edge analytics will help realize a long-cherished dream: the prevention of a vast array of maladies, and unequivocal cures for those diseases that do manifest.

After all, precision medicine isn’t about geneticists making stunning breakthroughs, or physicians applying the therapies that result from those discoveries. It’s about the patients who are cured or in extended remission, and the many who would have died without targeted treatments.