Industry Brief

How Diagnostic Testing Is Powering Precision Medicine

AI and accelerated genomic analysis provide fast and accurate biological insights.

Image provided by Natera.

Overview

An important step in providing effective care is determining what disease or illness a person may have. Because of this, diagnostic testing is used across medical fields to accurately diagnose and inform treatments for patients.

Advanced genomic technologies such as cell-free DNA (cfDNA) liquid biopsy, targeted gene panels, whole-exome sequencing (WES), whole-genome sequencing (WGS), methylation profiling, and transcriptome analysis have greatly expanded diagnostic capabilities. These technologies support testing of a multitude of conditions and diseases across oncology, prenatal and newborn screening, infectious diseases, and rare genetic diseases.

New applications for genomics-based testing include but aren't limited to:

Oncology

> **Early Detection:** Liquid biopsy measures cfDNA in fluids. This can provide a noninvasive approach to early detection before tumors are visible on imaging scans.

> **Treatment Monitoring:** cfDNA is useful in noninvasive monitoring like minimal residual disease (MRD). MRD is used to detect residual cancer cells, like blood or tissue, that may be present post-treatment.

> **Tumor Profiling:** WGS of a tumor as well as matched normal tissue can be used to understand the composition of a tumor and regions of the genome relevant to cancer progression and treatment.

Prenatal and Newborn Screening

> **Noninvasive Prenatal Testing (NIPT):** cfDNA from maternal blood can detect fetal chromosomal abnormalities with high sensitivity and specificity, reducing the need for invasive procedures like amniocentesis.

> **Early Detection of Hereditary Conditions:** WGS and WES allow rapid identification of rare or actionable genetic disorders in newborns, supporting timely interventions and informed treatment decisions.

Genetic and Rare Diseases

> **Rare Diseases:** WGS has been particularly useful in genetic testing for rare diseases, where patients can undergo a diagnostic odyssey when trying to understand their underlying condition.

> **Critically Ill Patients:** Genomics is being used when turnaround time for results is critical, such as when screening critically ill newborns, and for infectious disease profiling and diagnosis.

Key Challenges

> **Accuracy Challenges:** Detecting low-frequency DNA in liquid biopsies makes sensitivity critical.

> **False Positives:** Inaccurate test results and outcomes increase costs and delay correct treatment.

> **Cost and Speed Barriers:** High sequencing, overhead, quality control, and reimbursement delays slow turnaround.

> **Impact:** Addressing these challenges enables faster, more accurate diagnoses and better patient outcomes.

Benefits for Healthcare Organizations

> Reduce turnaround time with faster and accurate testing

> Early detection and precise DNA methylation analysis with BWA-Meth

> Ability to identify cancer mutations with Mutect2

Challenges

Traditional Challenges: Accuracy and Turnaround Time

Improving accuracy in liquid biopsy is particularly challenging due to the low volume of relevant DNA in blood samples. False positives are particularly detrimental in liquid biopsy approaches, because they can lead to incorrect diagnosis, resulting in retesting and incorrect treatment. Even more importantly, higher accuracy improves patient experiences and enables better outcomes through more reliable diagnoses.

Additionally, turnaround time remains a significant bottleneck. Specific to data generation and processing, both whole-genome sequencing and liquid biopsies require a larger amount of sequencing data, which extends data processing time and delivery of results. In scenarios where turnaround time is critical, moving from hours to minutes can have a significant impact on downstream decision making.

Emerging Challenges and Opportunities: Better Detection and Insights

With precision medicine, researchers are looking to harness multimodal and multiomics datasets—spanning genomics, methylation, transcriptomics, proteomics, and patient metadata—to unlock earlier disease detection, diagnose novel conditions, and identify precision therapeutic opportunities. Integrating molecular and patient metadata will only help to unlock additional insights.

Scientific and technical challenges associated with precision medicine continue to evolve. With the prominence of AI and machine learning, models are only as valuable as the data used to train them. To develop fair and generalizable methods, diverse datasets that are representative enough to avoid bias are critical. As new diseases or phenotypes emerge, molecular profiling and data analysis need to be intelligent enough to adapt, learn, and accommodate multimodal and temporal datasets for evolving scientific understanding.

NVIDIA Solutions

Improving Accuracy With NVIDIA Parabricks

For faster, consistent, and more accurate analysis, companies can use NVIDIA Parabricks, a GPU-accelerated software suite for secondary genomic analysis. Parabricks significantly reduces data processing time, reduces cost, and optimizes performance for several diagnostic processes.

For whole-genome, whole-exome sequencing and targeted gene panels, established tools like BWA-MEM, HaplotypeCaller, and DeepVariant are helpful for alignment and germline variant calling. With them, researchers can identify variants relevant to rare diseases, critically ill patients, and patient risk profiles. Additionally, next-generation graph and deep learning-based tools, such as Giraffe from UCSC and DeepVariant from Google, offer improved performance in both alignment and variant detection. By leveraging population-aware and pangenome references, these methods enhance accuracy.

Specifically for liquid biopsy and oncology, BWA-Meth is used for fast and accurate alignment for bisulfite sequencing (BS-seq), which is essential for sensitive and specific detection of DNA methylation patterns. This capability underpins early cancer detection and accurate methylation-based diagnostics. For somatic variant analysis, Mutect2, developed by the Broad Institute's Genome Analysis Toolkit (GATK), is a leading tool for identifying somatic mutations. DeepSomatic, from Google, provides a deep learning-based alternative for somatic variant calling. Both tools are designed to distinguish true somatic mutations from alterations present in normal tissue. This is particularly valuable for clinical oncology and the development of molecular diagnostics. Additionally, STAR and STAR-Fusion are widely utilized in oncology for the detection of transcriptome fusions and comprehensive expression profiling. By enhancing sensitivity and analytical depth, these tools deliver robust expression profiling and fusion detection across a range of transcriptome panels.

Parabricks provides GPU-accelerated implementations of these tools, enabling clinical geneticists to assess identified variants and inform diagnoses and treatments faster, while producing results that are identical or highly similar to baseline CPU tools.

	Alignment	**Variant Calling**
Germline: WGS, WES, and Targeted Gene Panels	BWA-MEM and Giraffe	HaplotypeCaller and DeepVariant
Somatic: WGS, WES, and Targeted Gene Panels	BWA-MEM and Giraffe	DeepSomatic and Mutect2
Oncology: Liquid Biopsy	BWA-Meth	DeepSomatic and Mutect2
Oncology: Transcriptome Fusion Panel and Expression Profiling	STAR	STAR-Fusion

List of tools that are available as GPU-accelerated implementations within NVIDIA Parabricks. These tools address alignment and variant calling steps that are relevant for germline, somatic, and oncology use. cases.

Leaders in Diagnostics

Freenome

Freenome has developed a multiomics platform that uses a blood test to detect cancer at its earliest, most treatable stages. Freenome's platform analyzes genomic, epigenomic, and proteomic biomarkers and uses AI, machine learning, and deep learning models to detect cancer-specific signals in the bloodstream. The company's first screening test for colorectal cancer was recently clinically validated in a large, multi-center prospective study. It's currently under review by the U.S. Food and Drug Administration and is anticipated to be available in 2026 for the 120 million people in the U.S. who are eligible for this screening.

Image provided by Freenome.

> "Our research team has presented data on an attention-based deep learning model that operates on billions of cell-free tumor DNA fragments per subject, generalizes to an independent hold-out cohort, and exhibits performance improvement with increasing training data volume. Even at low sample numbers (in thousands), this deep learning model outperformed a state-of-the-art machine learning method and can be applied to other indications, thus paving the way for effective deep learning in blood-based early cancer detection.
>
> The 2026 launch of our colorectal and lung cancer screening tests using the same assay workflow will spark the creation of one of the largest population-level health repositories, with millions of genomic datasets integrated with electronic health records and multimodal patient information. This underscores the indispensable role of collaborating with NVIDIA to achieve the scale and capabilities necessary to translate groundbreaking research into further clinical applications."

Jimmy Lin, M.D., Ph.D., MHS,
Chief Scientific Officer, Freenome

Natera

As a global leader in cfDNA testing, Natera provides personalized genetic testing and diagnostics. Specifically dedicated to oncology, women's health, and organ health, Natera works to inform patients earlier so they can lead longer, healthier lives.

Image provided by Natera.

> "At Natera, we're building proprietary genomic foundation models trained on hundreds of thousands of tumors with multimodal data—including DNA, longitudinal circulating tumor DNA (ctDNA) liquid biopsy, RNA, and imaging. Running on NVIDIA H100 Tensor Core GPUs, these models accelerate therapeutic biomarker discovery and validation, power patient-specific therapy design, and help optimize clinical trial stratification and next-line treatment decisions. In parallel, we're developing cancer genome analysis workflows accelerated by NVIDIA Parabricks to scale computationally intensive pipelines. This is AI, not just for diagnostics, but for transforming how personalized therapies are developed, optimized, and delivered to patients."

Helio Costa, Ph.D.
Head of Therapeutics and Innovations, Natera

Guardant Health

Guardant Health is a leading precision oncology company that works to transform patient care and accelerate new cancer therapies. In 2024, they announced FDA approval for Shield, their blood test for colorectal cancer screening.

Image provided by Guardant Health.

> "Ensuring accurate and timely results are the most important aspects of any diagnostic testing. NVIDIA GPUs enable us to optimize our bioinformatics pipeline and reduce model training time to further improve diagnostic accuracy and enable us to iterate more quickly on new innovations. In addition, NVIDIA GPUs allow us to fine-tune and run popular AI models within our existing infrastructure to enhance our operational capabilities and improve efficiencies across many workflows."

Kenny Speer
Vice President, Bioinformatics Software Engineering, Guardant Health

SOPHiA GENETICS

The SOPHiA DDM Platform, a cloud-native, software-as-a-service platform, empowers healthcare institutions with robust, data-driven insights. Using advanced AI algorithms, the platform computes, standardizes, and analyzes complex health data related to cancers and rare and inherited disorders, driving more informed decisions. With a diverse portfolio of applications, including advanced liquid biopsy testing for noninvasive cancer genomic profiling and monitoring, SOPHiA GENETICS is driving advancements in oncology research. By providing access to cutting-edge technology worldwide, the company is committed to transforming precision medicine on a global scale.

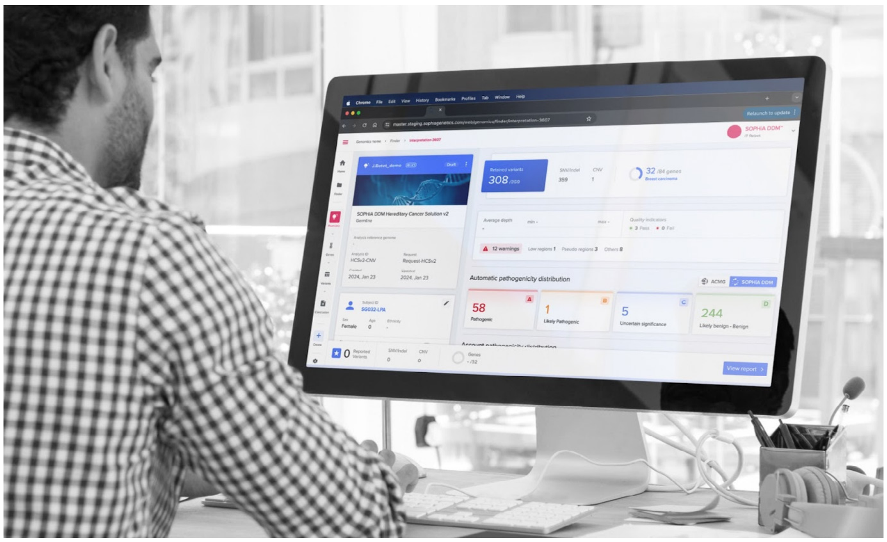

Image provided by SOPHiA GENETICS.

> "It is our continued goal to improve health outcomes for patients globally by expanding access to precision oncology and equipping local health institutions with the tools and technology needed to practice data-driven medicine."

Jurgi Camblong
CEO and Co-Founder, SOPHiA GENETICS

Powering Scalable and Accurate Diagnostic Testing

The diagnostics landscape is evolving rapidly. As a result, new challenges emerge when trying to balance increased accuracy with reduced cost and turnaround time. From requiring high-accuracy models that can be trained specifically to their own gene models to running batched samples for timely diagnosis at scale, the primary goal is to make fast and reliable testing available to the masses.

Diagnostic companies continue to pave the way for preventative healthcare and empower patients to be their own advocates. Ensuring diagnostic testing remains fast, accessible, and accurate to everyone is pivotal to this and to progressing precision medicine.

Ready to Get Started?

To learn more about diagnostics and NVIDIA Parabricks, visit nvidia.com/parabricks

Contact Sales at: www.nvidia.com/en-us/industries/healthcare/contact-sales

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