In a dozen years, sequencing of the human genome has been transformed from a Herculean task to one requiring as little as a day and a thousand dollars. This dramatic improvement in sequencing technology promises important benefits to human health, but realizing this potential in everyday practice requires overcoming new challenges. DNAnexus, the global leader in cloud-based genome informatics and data management, offers a universal solution to the informatics challenges posed by next-generation sequencing (NGS), providing the backbone technology needed to apply NGS to the diagnosis and treatment of disease (Box 1).

“Medical progress is driven by inquisitive and impassioned clinicians, researchers and data scientists determined to make a difference,” said DNAnexus chief medical officer David Shaywitz. “Our goal is to support and empower these champions.” The DNAnexus Platform speeds the delivery of clinical innovation to patients with a turnkey computational and data management solution that is efficient, customizable and compliant with clinical regulatory demands, yet flexible enough to integrate other NGS technologies into a customer-specific pipeline. DNAnexus serves a broad range of global partners, from prenatal testing companies to the US Food and Drug Administration (FDA).

NGS challenges

Delivering on the promise of NGS will require overcoming critical challenges, including the quantity of data involved, the need for comprehensive regulatory compliance, stringent controls on patient data privacy and security, the need for collaboration across institutional firewalls, and integration with upstream and downstream systems, including sequencing platforms. “Our clinical customers need data management to be HIPAA [Health Insurance Portability and Accountability Act] compliant and secure at all steps in the process. This becomes even more important at scale, when petabytes of data are involved,” explained Richard Daly, chief executive officer at DNAnexus.

Extending global reach

DNAnexus has partnered with Natera, a leader in noninvasive genetic testing, to support data analysis, storage and sharing. Using the DNAnexus Platform, Natera’s remote global partner laboratories can upload sequencing data to a single secure, CLIA-certified environment. This has allowed Natera to rapidly expand into new markets by minimizing IT friction points. According to Daly, “The DNAnexus Platform serves as a compliant command center supporting multiple testing sites and enabling providers such as Natera with the technical platform to market and monetize their tests globally.”

Advancing cancer care

When Intermountain Precision Genomics, an in-house laboratory for Intermountain Healthcare, was searching for a bioinformatics solution, the ability to share research data and rapidly scale the effort across their 22 hospitals and medical group was a key criterion. Intermountain and DNAnexus worked together to develop a cloud-based bioinformatics pipeline to translate raw sequence data into interpretable variants. “We selected DNAnexus because of the company’s leading bioinformatics and cloud computing expertise, combined with the best-in-class security and compliance standards of the Platform,” said Lincoln Nadauld, medical director of Intermountain Precision Genomics. With these data, Intermountain’s oncologists can identify tumor-specific mutations and determine the best treatment option for each patient. “The underlying infrastructure that DNAnexus provides allows us to focus on our core competencies of R&D and patient care while revolutionizing cancer treatment.”

Leading open-source NGS R&D

As a leader in the field, DNAnexus was awarded a contract to create precisionFDA, an open-source platform for sharing of genomic-testing information. As part of the White House’s Precision Medicine Initiative, the FDA is tasked with developing a streamlined approach to evaluating diagnostics that rely on NGS technology, and precisionFDA is a critical part of that effort. DNAnexus anticipates that precisionFDA will be used by test developers, standard-making bodies, biopharmaceutical companies, health care providers, academic medical centers and patient advocacy groups.

For diagnostic developers, the precisionFDA Platform offers a venue for comparing new tests to approved ones and sharing results with collaborators and the FDA. As participants grow and more NGS data become available, this grassroots effort is expected to generate reference data sets and ultimately help to define industry standards. “DNAnexus is proud to be creating a community around open-source genomic-analysis pipelines, reference data and analytical processing resources,” said Daly. “The Platform will enable the managing and sharing of genomic data at an unprecedented level.”

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