WHITEPAPER

Precision Medicine Requires **Scalable Data Sharing, Data Protection**

Myriad Genetics' Precise Treatment Registry offers the data access and safeguards needed to spur clinical innovation

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As healthcare organizations aim to maximize the potential of precision medicine, they are apt to realize that data-driven clinical and genetic insights are vital to advancing scientific innovation, supporting research, and improving health outcomes. Today's precision health data is multimodal in nature and massive in volume. To maximize the potential of this data, organizations must be able to ingest, harmonize, and integrate data at scale while also ensuring that data privacy and security standards are maintained. Overcoming these challenges opens the door to new possibilities for organizations invested in precision medicine, enabling advanced scientific data analysis and faster time-to-insight for researchers.

Myriad Genetics, a leading genetic testing and precision medicine company committed to improving patient care across medical specialities through genetic insights, and DNAnexus, a leading platform for secure, scalable, and cloud-based bioinformatics data analysis, are working together to move in this direction. Grounded in the belief that access to quality data increases the clinical value of genetic tests, Myriad Genetics pioneered the Myriad Precise™ Treatment Registry in collaboration with DNAnexus. This innovative registry addresses key challenges associated with sharing large scale clinico-genomic data, supporting the next generation of precision medicine advancements by combining structured, de-identified clinical data with somatic, germline, and/or genomic instability testing data. For example, clinicians can compare outcomes of patients with the same test result across varying clinical or genetic factors to understand how to better manage progression of disease.

"Our mission is to advance the understanding of cancer and allow people to live healthier, longer lives. Data is the key to gaining more understanding," said Thomas P. Slavin, M.D., Chief Medical Officer, Myriad Genetics. "We realized that our data could provide significant benefits to our customers, partners, and to the field in general, if delivered in a safe, usable manner."

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THOMAS P. SLAVIN, M.D.,
Chief Medical Officer
Myriad Genetics.

Dealing with data difficulties

By leveraging the DNAnexus platform to support the Precise Treatment Registry, Myriad is moving beyond the difficulties associated with the data sharing status quo in the healthcare industry.

"The Precise Treatment Registry is getting valuable data into the hands of scientists driving research to advance cancer care," Slavin said. "A real cohort browser where people can look at real-world data in real-time. It's very concrete. It's not smoke and mirrors. With the Precise Treatment Registry, it is possible for researchers, clinicians, and other users to log on, look at the data, manipulate the data, do real-world queries, and test real-world hypotheses."

By leveraging the DNAnexus infrastructure, the Precise Treatment Registry is successfully addressing a variety of stymying challenges:

Scale of genomic data. The sheer size of genomic data makes it onerous to move from system to system. Genomic data can stretch from many terabytes to well into the petabytes. As such, genomic data sets are challenging the notion that downloading data is a viable data sharing paradigm.

The DNAnexus platform supports a "bring compute to data" model where sharing data sets and cohort data does not require users to physically move data files. The platform empowers users to invite other researchers and clinicians to participate, who then can get value from the data immediately without needing to wait days or weeks to simply transfer the data. Instead, researchers and clinicians collaborate in real time via the Precise Treatment Registry.

"We are using a model where users no longer download data. Instead, the users come to environments preloaded and tailored to the data sets where the data owners can ensure regulatory control of the data. The users benefit with a turnkey environment that is ready to go for them to conduct research." said John Ellithorpe, President, DNAnexus.

Integration of common ontologies. Creating mappings between concepts in different ontologies is a critical component of successful data sharing. Ontologies provide a clear definition of concepts in a given domain. For example, SNOMED defines weight as "body weight" and code "27113001." Another ontology, however, might use a different term and code for the same concept. With the DNAnexus platform, it is possible to accurately integrate various ontologies within the data set. Researchers can confidently trust that the concept will carry the same meaning across the data set. In addition, researchers can rely on their existing knowledge of various ontologies and are not required to learn new terms and definitions to work with the data set.

Fragmentation of data sources. Precision medicine often requires pulling "nuggets of information from multiple different systems and then synthesizing all that information,"

according to Kevin R. Haas, Chief Technology Officer, Myriad Genetics. A common challenge faced by many healthcare organizations is they "do not employ an army of software engineers or data scientists" that can do this work, Haas said.

The registry is preloaded with de-identified data on thousands of patients results, along with clinical components from the test requisition form (TRF). Preloading data allows for immediate exploration for clinicians and researchers who have been fully onboarded. In addition, de-identified clinical data and results from tests ordered can be uploaded, including Precise™ Tumor, MyChoice® CDx, MyRisk or BRACAnalysis CDx®, and Prolaris®.

Once access is granted to the research portal, users will be able to carry out queries and analyses that will help inform and foster research opportunities. Some examples include:

- Expanding somatic-germline interactions and associations
- Comparing individual data (in process) to aggregate group data sets

Assurances of data privacy. When working with healthcare data – especially genomic data – it's important to address privacy, security, and consent concerns. Genomics is particularly sensitive as it's like a person's fingerprint, offering unique, readily identifiable information about individual patients.

"Organizations have to be respectful of data and make sure they are on the right side of history when it comes to such concerns," Haas said. Indeed, when organizations meticulously address these data security issues, researchers and clinicians are apt to be "most eager to partner with them," Haas said. "When allowing access to needed data while simultaneously protecting it, an 'ad-hoc' approach simply does not work."

The Treatment Registry was developed from the ground up with security and privacy built in. With the DNAnexus infrastructure in place, the registry specifically manages "which data is accessible, which data is anonymized for which purposes, which consent is required, and who has access to which cohorts. It's imperative to have a framework or infrastructure that proactively manages privacy and compliance," Haas pointed out.

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The platform also supports all the necessary data access controls, privacy and compliance functions, and certifications needed for Institutional Review Board (IRB)-approved studies.

Plenty of potential

With this platform in place, researchers and clinicians who use the registry are able to:

Quickly conduct research studies. "Researchers really like having the ability to test hypotheses and start thinking through things in real time. It's a great platform for researchers, residents, and fellows who are working in hospital systems who need quick answers," Slavin said. "With the data in this platform, users can literally produce an abstract in as little as 45 minutes, which is incredible. We're able to advance the field by providing researchers with access to data at their fingertips."

Make meaningful comparisons. Clinicians and researchers often need to compare what they are doing with the patients in front of them to other cohorts of similar patients. With the data available in the platform, it will be easy to see the interventions and results associated with patients with similar conditions. "What have others like them done? How were the outcomes? What were the genomic signatures? By answering these questions, users will be able to optimize the protocol, the treatment opportunities, and treatment options for the person sitting in front of them," Slavin noted. "It's nice to have the knowledge of not just yourself and the 10 other patients you may be seeing at that point, but of the 10,000 patients that are part of the larger hospital or health system cohort."

WORKING WITH THE RIGHT PARTNER

When launching the Precise Treatment Registry, Myriad wanted to quickly find a way to effectively and securely share data with researchers and clinicians.

To internally build the infrastructure required for such data sharing would simply be too labor intensive and take too much time. In essence, the opportunity cost associated with internal development was prohibitive.

Myriad's leaders acknowledged that working with a partner that could provide a proven, ready-to-use platform was critical to provide the speed-to-market advantages needed.

In addition, they realized that researchers and clinicians are apt to shy away from using an internally developed platform that is not commonly used across the industry.

"They just don't want to learn and trust another person's app or infrastructure. We wanted to embrace a trusted and respected platform that already had the buy-in and engagement of end-users," Haas said. "It's the right thing to do. It's just better to embrace a common third-party standard that everyone can buy into and share more than just our data on."

Myriad leaders decided to work with DNAnexus to provide the platform for the Precise Treatment Registry. DNAnexus was not only able to provide a ready-to-go, secure, compliant and highly scalable environment for Myriad's clinico-genomic data, but they also acted as a key data design partner in understanding what the provider and researcher users need to do with the information.

"There certainly is sophisticated infrastructure. It's behind the scenes. It has the horsepower of AWS and Apache Spark," Haas said. "It also provides great data visualizations across both clinical and genomic information." ■

Support advanced scientific data analysis. The platform also empowers users to conduct sophisticated studies. “For example, if researchers want to do something very sophisticated, they can move forward without worrying about having the horsepower of discovering new informative single nucleotide polymorphisms for polygenic risk scores or making associations between gene mutations and ancestry. You have the ability to run these large-scale, large cohort data set analyses that can scale out to thousands of cores and machines in the cloud and access terabytes or petabytes of data,” Haas said.

Push precision medicine forward. The DNAnexus platform empowers Myriad to become a better partner to researchers and clinicians — and therefore pushes precision medicine forward. “With this platform in place, we’re able to become more collaborative and provide improved access to data. We’re [Myriad] enrolling in partnerships with different research studies and pharmaceutical partners that will lead into something that needs statutory oversight that you’d expect from an FDA-approved product,” Haas said.

Slavin added, “The DNAnexus backed portal empowers researchers with tools that make it possible to innovate. As a result, we can work easily with the medical community to help advance cancer care.”

DNAnexus is proud of the partnership with Myriad to create their Precise Treatment Registry. “We have only scratched the surface of the potential for precision medicine,” Ellithorpe concluded. DNAnexus is looking for other healthcare organizations to partner with as they seek to advance their precision medicine initiatives. ■

- Come visit Myriad’s website and sign up for access to Precise Treatment Registry: https://myriad.com/oncology/precise-registry/
- To learn more about DNAnexus, go to: https://www.dnanexus.com/use-cases/clinical-diagnostics