

CASE STUDY

Ultragenyx: Overcoming Technological Challenges to Fast-Track R&D

Advancing Rare Disease Therapeutics with DNAnexus[®] Titan



Company Overview: A Rare Disease Pioneer

Ultragenyx, a biopharmaceutical company based in Novato, CA, is committed to bringing novel products to patients for the treatment of rare and ultra-rare diseases, with a focus on serious and debilitating genetic diseases. The company has rapidly built a diverse portfolio of approved therapies and product candidates aimed at addressing diseases with high unmet medical need, yet clear underlying biology. Ultragenyx's strategy is predicated upon time and cost-efficient drug development, with the goal of delivering safe and effective therapies to patients with the utmost urgency.

The Challenge: Decreasing Analysis Time of Multi-Omics Data

As Ultragenyx scaled its R&D efforts, leadership recognized a need for a more streamlined way to unlock the power of multi-omics data to accelerate their discovery process.

The small bioinformatics department is responsible for supporting all research and clinical initiatives for the entire company, running analytical pipelines and sharing analysis data back to those researchers for computationally-demanding data analyses including: variant calling from whole-exome and whole genome sequencing, RNA-seq, ATAC-seq, CHIP-seq, nanopore sequencing, and assembling genomes. Competing priorities and concurrent analyses resulted in long compute queues and backlogs, creating bottlenecks in the R&D process. The head of the bioinformatics department knew he needed a way to simplify and automate these highly technical and computationally-heavy analyses to reduce turnaround time, and make it easier for researchers to run specific analyses.

The Solution: A Purpose-Built Platform for NGS Analysis Pipelines

Ultragenyx chose to implement **DNAexus Titan™**, a unified NGS analysis platform, to bring data and pipelines together in one secure environment for streamlined multi-omics data analysis.

DNAexus Titan™ provides a cloud environment, enabling sharing of projects, data, and tools to team members who have approved access. This enables the bioinformaticians to build and maintain analysis applications and share them with research and clinical groups, thus empowering non-bioinformatician researchers to self-serve and conduct analyses on their data with well-maintained and version-controlled pipelines. Researchers can upload their data to DNAexus, conduct analysis with an approved pipeline, visualize, and share results with collaborators - all with an intuitive user interface. Moreover, DNAexus

facilitates the efficient delivery of analysis results from complex pipelines run by a bioinformatician. Data analysis, visualization, and sharing of results to other parts of the company can occur on one unified and secure platform.

In addition to solving collaboration challenges, DNAexus brought about new computing efficiencies to reduce turnaround time for results. The platform minimizes compute time through parallel processing, getting results in the hands of researchers faster.

DNAexus eliminates bottlenecks with a platform built for streamlined multi-omics analysis. With these new efficiencies in place, Ultragenyx has been able to rapidly scale to analyze more multi-omics data, without increasing the workload on the bioinformatics team.

Results: Fast-Track R&D

By implementing DNAexus, Ultragenyx was able to take advantage of cloud infrastructure to streamline data analysis and experience dramatic improvements in turnaround time and computing costs.

Efficiencies brought about by running pipelines on DNAexus decreased turnaround time 10 fold. The bioinformatics department now is able to support research and clinical groups more rapidly.

With transparent pricing per analysis run, team members can calculate costs before running jobs, resulting in predictable costs and the ability to tailor cloud instance type to minimize costs and maximize efficiency. In addition to experiencing tangible cost savings with DNAexus, according to the head of bioinformatics, the primary benefit is the ability to save time and bring drugs to market faster.

Delivery of rare disease therapeutics requires rapid discovery, and Ultragenyx is in the business of time. With the right infrastructure in place, Ultragenyx simplified the complexities of secondary analysis infrastructure, allowing researchers to focus on what's important - rapid rare disease discovery.

“You save the bioinformatician’s time as well as compute time, and therefore decrease turnaround time. This enables R&D researchers to answer questions and get rare disease treatments to market faster. You can’t put a price tag on that.”

SEAN DAUGHERTY

Associate Director of Bioinformatics, Ultragenyx

BENEFITS:

- › Solve infrastructure challenges to scale future growth
- › Build version-controlled and traceable pipelines
- › Empower non-bioinformatics researchers to self-serve with approved workflows
- › Enable bioinformaticians to seamlessly deliver results from complex pipelines
- › Experience decreased time to results