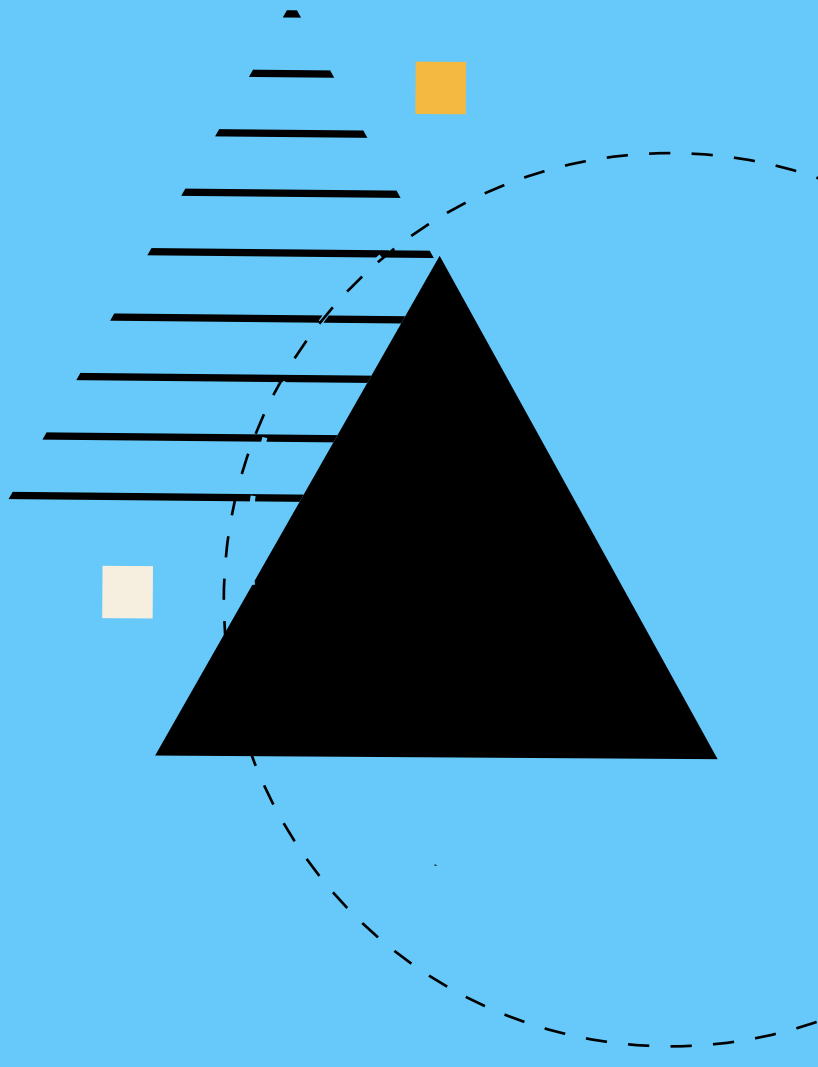


DNAnexus®

Overcome the four common data challenges in clinical diagnostics

How clinical diagnostic labs can flexibly scale to meet the demand for increased NGS test volume

Introduction

Today clinical diagnostic labs are at a crossroad. You face unprecedented demand for availability, flexibility, and scalability. At the same time, you need to maintain the quality of your services and keep your operations affordable.

In the era of next-generation sequencing (NGS), almost all components of the diagnostic workflow have gone digital—but that doesn't mean things have gotten easier. Workflows can suffer from a lack of standardization, interoperability, and connectivity at all levels. Applications cannot exchange data, systems are not connected, and data is locked in silos.

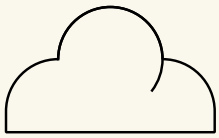
But lack of technology is not the problem. More technological solutions are available to you than ever before. Technologies like cloud, big data, and artificial intelligence can help improve health outcomes. But you also want to lower costs, improve diagnostic time and quality, and revolutionize the healthcare experience you provide for both patients and doctors.

Interconnectivity and interoperability are becoming essential, if you want to access the data needed to speed up diagnoses and make real-time decisions. Therefore, you need a comprehensive plan. Here, we address the four major data challenges in clinical diagnostics and how you can overcome these barriers.

Challenge #1

MANAGING TODAY'S HIGHER VOLUME OF SEQUENCING DATA

Since 2017, the demand for clinical diagnostics within the United States has increased by an average of 3.1% each year[1]. But with factors such as technological advancements, a rising elderly population, and consumers becoming increasingly aware of available genetic testing services, diagnostic labs are expected to see an exponential growth in NGS testing volume over the next 5 years[2].



Solution:

Move NGS analysis to the cloud

Moving your NGS analysis to the cloud will give you an environment that can flexibly scale to meet the demand for increased test volume, all while saving time and money. Cloud-based systems enable you to optimize analysis pipelines for quality, speed, runtime, and cost. The cloud eliminates bottlenecks in processing queues and server capacity.

Yet not all clouds are created equal, and clinical genomics has unique requirements. Look for vendors with solutions that are purpose-built for clinical genomics and who have experience supporting leading diagnostic testing organizations across a wide range of applications.

Challenge #2

THE EVER-CHANGING REGULATORY LANDSCAPE

Every clinical diagnostics company contends with growing—and increasingly complex—regulatory, certification, and security requirements. When a company expands to new countries, the task becomes tougher still, especially if this brings new requirements around data sovereignty and intellectual property (IP) protection. With Food and Drug Administration changes coming to regulate in-vitro diagnostic tests as medical devices, maintaining GxP compliance, including adherence to 21 CFR Part 11 requirements, it an important consideration as organiations navigate the changing regulatory landscape. In these conditions, do-it-yourself compliance solutions quickly become too expensive and complex to maintain compared to finding a partner with out-of-the box compliance.

Solution:

Mitigate risk by finding a trusted partner for security and compliance processes

Instead of hiring staff to continually monitor the everchanging regulatory landscape, work with a vendor with a proven track record helping clinical diagnostic labs succeed by building solutions that meet the industry's most strict compliance regulations, including:

- HIPAA
- CAP/CLIA
- 21 CFR Part 11
- GDPR
- IVDR
- Audit trail
- FedRAMP
- ISO 270001
- PCI DSS
- End-to-end encryption
- Strict production system access control
- Cyber Essentials
- Cyber Essentials Plus
- Data Privacy Framework

Challenge #3

THE NEED TO REDUCE OPERATIONAL COSTS

According to Laboratory News, in a 2021 survey of around 200 medical lab professionals in Europe, 76% of medical and research laboratories report rising lab operation costs[3]. Rising costs were reported across the entire range of laboratory work—from sample preparation to instrumentation and control. However, the cost of the informatics portion of the workflow is at higher risk for significant increase as more data is generated, analyzed, and stored per sample than ever before.

This is especially true for clinical diagnostic labs using proprietary, in-house informatics solutions, as these systems often become unexpectedly expensive in an effort to keep them reliable and efficient as you scale.

Solution: Adopt a purpose-built NGS informatics platform

Scalability and cost go hand-in-hand. To help keep cost per sample down, diagnostic laboratories should adopt a purpose-built NGS informatics platform capable of handling the enormous scale of NGS data in terms of data management and analysis.

Data management: Software maintenance and technical support is a continuous commitment. You have to keep your informatics platform up to date with ever-changing security and compliance regulations, software patches, and technology updates, potentially increasing staff workload and draining resources. These challenges can increase business costs such as:

- Increased headcount to manage operations
- Unplanned system downtime
- Extended turnaround times
- Delays getting new offerings to market

Selecting a trusted partner with a purpose-built NGS bioinformatics platform keeps your costs under control by:

- Managing maintenance activities, updates, and security patches
- Maintaining system regulatory compliance
- Increasing memory and storage when you are ready to scale
- Eliminating excessive compute times

Data processing: Two of the most resource-intensive components of a NGS workflow are data analysis and interpretation. Diagnostic laboratories should use scalable secondary analysis and tertiary analysis software solutions that:

- Reduce personnel hands-on time and effort
- Offer preconfigured workflows for industry-leading panels, such as the Illumina® TruSight™ Oncology 500 assay
- Provide access to subscription-based databases, such as the Human Somatic Mutation Database (HSMD) and Human Gene Mutation Database (HGMD®) Professional, with no additional fees
- Minimize risk of misclassifying variants

Challenge #4

PRESSURE TO DEVELOP AND DELIVER DIAGNOSTIC TESTS FASTER

The clinical diagnostic lab is at the heart of fast and accurate healthcare delivery. This means constant pressure for quick turnaround times, high-quality samples, and clear results. The demand for rapid genetic testing is at an all-time high. Even tests once considered routine are now under time pressure.

The more efficiently your lab can run tests and generate results, the more you can accomplish. Faster turnaround times can free up staff and resources for other activities, like growing your overall test catalog. And don't forget the reputation factor. You want to be the reliable go-to lab for the clinicians you serve — to be trusted for accuracy, professionalism, and speed.

Solution: Integrate, connect, and automate key processes

While the factors that can lead to poor turnaround times are numerous and varied, they can generally be grouped into two main categories: inefficient workflows and complexity of managing multiple tools. To accelerate turnaround times, build an integrated workflow that seamlessly goes from FASTQ to final report and automates key steps of the NGS workflow.

Points of workflow integration to reduce turnaround time

- Ability to directly upload raw FASTQ files from the sequencer to a secondary analysis software solution
- Ability to directly upload VCF files from a secondary analysis solution to a tertiary analysis solution
- Ability to store all data in the cloud without requiring local hardware

Key steps to automate in the NGS workflow:

- Security and regulatory compliance
- Software patches and technology updates
- Variant filtering and prioritization
- Secondary analysis of multiple sequencing runs simultaneously
- Variant classification according to professional guidelines (AMP/ASCO/CAP, ACMG/AMP)
- Variant curation
- Report generation

The Solution:

SCALE, STREAMLINE AND SIMPLIFY YOUR CLINICAL NGS WORKFLOW

For clinical diagnostic labs, a platform as a service combined with scalable informatics solutions is the self-running engine for growth within the field.

DNAnexus®, developer of the world's most secure, trusted platform for biomedical data analysis, offers an agnostic, end-to-end workflow for oncology and hereditary disease applications that goes from FASTQ to final report in a fraction of the time and cost (Figure 1).

THE DNANEXUS CONVENIENCE

Through the partnership, DNAnexus provides a secure, cloud-based platform for NGS secondary analysis that brings all your data and pipelines together in one place to deliver unparalleled efficiencies. The DNAnexus platform removes bottlenecks in secondary analysis—a computational- and storage-intensive process—through a cloud-based approach that minimizes the need for expensive hardware, additional IT resources, regulatory compliance management, and the development of advanced bioinformatics skills. Instead users rely on DNAnexus to address the administrative burden of maintaining IT infrastructure, compliance, and quality testing.

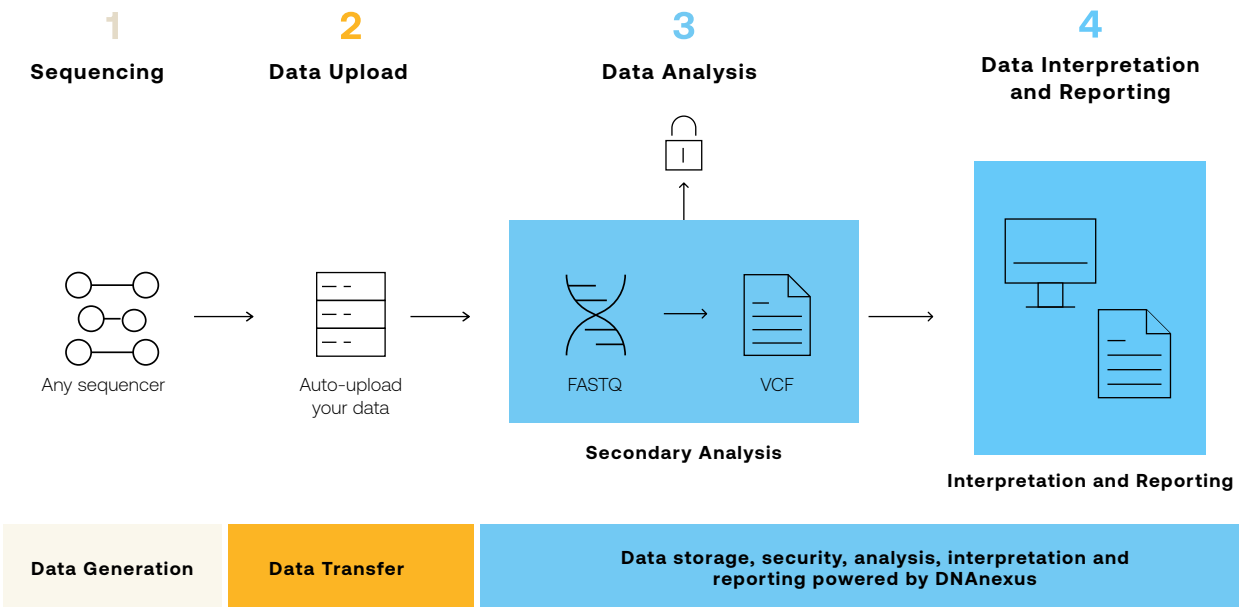


Figure 1. An integrated workflow from sequencing to final report

Empower your lab to do more, *with less*

To overcome today's data challenges, clinical diagnostic labs need scalable, cost-effective workflow solutions that deliver security, quality, and consistency, DNAnexus is ready to help you:

- Reduce time and effort with a unified, workflow-agnostic platform that lets you create, refine, validate, and execute pipelines with unprecedented speed.
- Scale your environment to meet demand for increased volume with exceptional uptime, powerful compute capacity, minimal infrastructure investment, and lower costs.
- Simplify compliance management and protect data with industry-leading, region-specific security and compliance that evolves overtime to meet requirements.
- Deliver patient test results faster with seamless variant analysis, interpretation, and reporting.

Take the first step to optimize your operations. See how DNAnexus can help your lab do more, with less.

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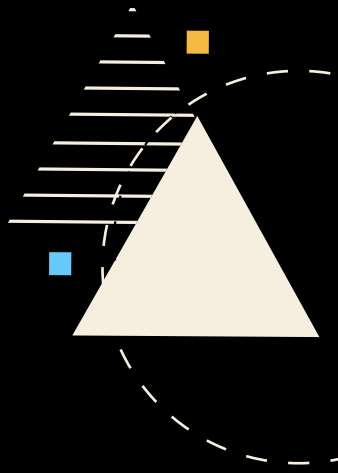

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PRODUCT DISCLAIMER

QCI Interpret is an evidence-based decision support software intended as an aid in the interpretation of variants observed in genomic nextgeneration sequencing data. The software evaluates genomic variants in the context of published biomedical literature, professional association guidelines, publicly available databases, annotations, drug labels, and clinical trials. Based on this evaluation, the software proposes a classification and bibliographic references to aid in the interpretation of observed variants. The software is NOT intended as a primary diagnostic tool by physicians or to be used as a substitute for professional healthcare advice. Each laboratory is responsible for ensuring compliance with applicable international, national, and local clinical laboratory regulations and other specific accreditations requirements.