White Paper

# 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. There is often a lack in 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. There are more technological solutions 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 that 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 entails 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. In these conditions, do-it-yourself compliance solutions quickly become too expensive and complex to maintain.

### Solution: Mitigate risk by offloading security and compliance processes

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

- HIPAA
- CAP/CLIA
- 21 CFR § 11
- GDPR
- IVDR
- End-to-end encryption
- Strict production system access control
- Audit trail

## 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 dealing with rising prices in lab operations (3). While rising operational costs were reported across the entire range of laboratory work—from sample preparation to instrumentation and control—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 performant 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, all of which becomes added work for your existing staff and often an endless resource drain.
  These challenges can have a real cost to your business in terms of needing to hire additional headcount to manage operations, unplanned system downtime, extended turnaround times, and delays in getting new offerings to market. Selecting a purpose-built NGS informatics platform from an outside vendor lets you offload maintenance activities, easily add memory and storage, and eliminate excessive compute times, which will help keep your costs under control.

- **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
    - And 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, and even tests once considered routine are now under time pressure.

Furthermore, the more efficiently labs can run tests and generate results, the more they can accomplish. Faster turnaround times can free up staff and resources for other activities, like growing the overall test menu. And let's not forget the reputation factor. Labs want to be the reliable go-to for the clinicians they serve. They want 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 the complexity of managing multiple tools. To accelerate turnaround times, diagnostic labs should 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 patch and technology updates
- Secondary analysis of multiple sequencing runs simultaneously
- Variant filtering and prioritization
- Variant classification according to professional guidelines (AMP/ASCO/CAP, ACMG/AMP)
- Variant curation
- Report generation

## The How: 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, and QIAGEN® Digital Insights, owner of the world's largest, fastest growing genomic knowledge base, are partnering to provide clinical diagnostic labs 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.

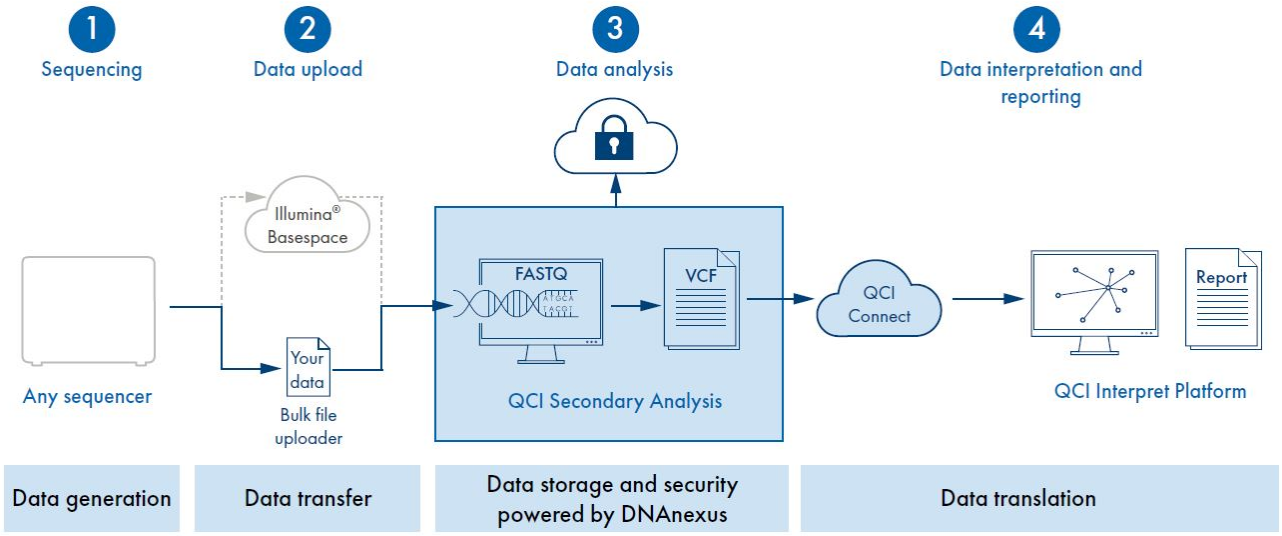


**Figure 1.** An integrated workflow from FASTQ to final report

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. Users instead rely on DNAnexus to address the administrative burden of maintaining IT infrastructure, compliance, and quality testing.

## The QCI advantage

The QIAGEN Clinical Insights (QCI®) portfolio is an integrated, universal solution for secondary and tertiary analysis in molecular pathology, clinical genetics, and translational research. Composed of three offerings—QCI Secondary Analysis and QCI Interpret—the QCI portfolio streamlines NGS data analysis and variant interpretation, enabling users to go from raw NGS instrument data to clinical report with an 85-90% reduction in turnaround time regardless of panel size.

QCI Secondary Analysis is a cloud-based service powered by DNAnexus. Scalable and workflow agnostic, QCI Secondary Analysis can be used with any NGS instrument and panel combination and seamlessly connects to QCI Interpret, QIAGEN's platform for clinical NGS interpretation.

Highly flexible and efficient, QCI Secondary Analysis requires minimal resources and expertise to automate and rapidly analyze an entire sequencing run in just five simple steps.

VCF files can then be directly uploaded to QCI Interpret for downstream interpretation and reporting.
QCI Interpret dynamically and transparently computes variant pathogenicity and actionability based on the AMP/ASCO/CAP or ACMG/AMP guidelines. Users can then auto-generate custom, patient-specific reports with the latest diagnostic and prognostic information, as well as relevant biomarker directed therapies and clinical trials. To date, QCI Interpret been used to analyze and interpret more than 3 million patient molecular profiles for hereditary and oncological diseases worldwide.

## The QKB difference

The content core of the QCI portfolio, the QIAGEN Knowledge Base (QKB), is powered by Augmented Molecular Intelligence, the combination of artificial intelligence (AI) and human expertise to advance and accelerate confident clinical decision making.

A key differentiator of the QCI portfolio, the application of Augmented Molecular Intelligence leverages AI and machine learning to efficiently identify, extract and align evidence from scientific literature and over 40 public and proprietary databases in the QKB. Then, over 200 MD- and PhD-level expert curators interpret, contextualize, augment, and verify the quality of the information before inclusion in the knowledge base. The approach ensures users have access to high quality molecular intelligence to augment their own decisions.


## 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. The partnership between QIAGEN Digital Insights and DNAnexus provides clinical diagnostic labs with an ultra-fast workflow from FASTQ to final report that enables you to:

- 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 automated variant analysis, interpretation, and reporting.

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

To learn more, please visit: **www.digitalinsights.qiagen.com/qci-secondary-analysis/**

## References


1. IBISWorld. (2021b, October 23). IBISWorld - Industry Market Research, Reports, and Statistics. Diagnostic & Medical Laboratories in the US - Market Size 2005–2027. https://www.ibisworld.com/industry-statistics/market-size/diagnostic-medical-laboratories-united-states/
2. Business Wire. (2021, June 11). Global Next-Generation Sequencing Market to 2025 - Impact of COVID-19. Research and Markets. https://www.businesswire.com/news/home/20210611005333/en/Global-Next-Generation-Sequencing-Market-to-2025---Impact-of-COVID-19---ResearchAndMarkets.com
3. Castanho, G. (2022, February 23). MLO's 2022 Annual Salary Survey of laboratory professionals. Medical Laboratory Observer (MLO). https://www.mlo-online.com/management/careers/article/21257623/mlos-2022-annual-salary-survey-of-laboratory-professionals


## Product Disclaimer

QCI Interpret is an evidence-based decision support software intended as an aid in the interpretation of variants observed in genomic next-generation 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.