

DRAGEN™ Pipelines on BaseSpace™ Sequence Hub

Accurate, rapid secondary analysis in an easy-to-use, cloud-based environment.

Highlights

- **Accurate, fast analysis**

Analyze a whole human genome at 30× coverage in ~35 minutes or a whole exome at 100× coverage in ~8 minutes with high analytical sensitivity and specificity

- **Simple workflow**

Stream data directly from the sequencing system to BaseSpace Sequence Hub and launch DRAGEN analysis pipelines with the push of a button

- **Low-cost, scalable platform**

Operate in the cloud and use resources on-demand to minimize costs and meet laboratory needs

- **Secure, compliant environment**

Assure data privacy with a security-first platform independently audited and certified for HIPAA compliance,* ISO 27001, and ISO 13485

Introduction

The Illumina DRAGEN (Dynamic Read Analysis for GENomics) Bio-IT Platform provides accurate, ultra-rapid secondary analysis of next-generation sequencing (NGS) data. The DRAGEN platform is available in the cloud through BaseSpace Sequence Hub, the easy-to-use, security-first Illumina cloud-computing data management platform. Combining the accuracy and speed of the DRAGEN platform with the user-friendly interface and low-cost pricing model of BaseSpace Sequence Hub enables users of all levels of bioinformatics expertise to leverage leading analysis tools to extract meaningful insights from sequencing experiments.

Accurate, fast analysis

The DRAGEN platform is engineered to remove biases and other sources of error, ensuring highly accurate results. In the 2017 Precision FDA Hidden Treasures – Warm Up Challenge, the DRAGEN platform received the highest score in five out of six accuracy measures for whole-genome variant calling among platforms that recognized all 50 variants.¹ In addition to accuracy, the DRAGEN platform enables ultra-rapid secondary analysis, as demonstrated by two independent institutions that used the platform to set speed records in genomic analysis.^{2,3} This optimized performance is available for a wide variety of genomic analysis solutions, including BCL conversion, mapping, alignment, sorting, duplicate marking, and haplotype variant calling. Fundamental features of the DRAGEN Platform address common challenges in genomic analysis, such as lengthy compute times, consistent accuracy, and massive volumes of data.

*HIPAA compatibility applies in the US only with BaseSpace Enterprise

A variety of DRAGEN pipelines are currently available on BaseSpace Sequence Hub to support multiple sequencing applications, including exome, genome, RNA, and methylation (Table 1). Additional pipelines and new versions are released on a regular cadence. For a comprehensive list of pipelines, visit www.illumina.com/DRAGEN.

Table 1: DRAGEN Pipelines currently available on BaseSpace Sequence Hub

Pipeline	Application
DRAGEN Germline Pipeline	End-to-end (BCL→VCF) NGS analysis, including advanced error model calibration for increased accuracy, and repeat expansion detection and genotyping through Illumina Expansion Hunter
DRAGEN Somatic Pipeline	Somatic variant detection in tumor samples, includes tumor-only and tumor-normal modes
DRAGEN RNA-Seq Pipeline	Rapid alignment and splice junction mapping, quantification, and fusion detection
DRAGEN Joint Genotyping/Population Pipeline	Joint variant calling across multiple genomes and scales to thousands of samples at expedited speeds with uncompromising accuracy
DRAGEN Methylation Pipeline	Ultra-rapid analysis of whole-genome and targeted bisulfite DNA sequence data; compatible with Illumina TruSeq™ DNA Methylation and TruSeq Methyl Capture library prep kits
DRAGEN Reference Builder	Uses FASTA files to build the proprietary reference used by the DRAGEN apps

Simple workflow

The DRAGEN Platform on BaseSpace Sequence Hub integrates leading secondary analysis pipelines into a simple workflow. Users can monitor runs in real time and securely stream data directly from instruments into the cloud ecosystem for push-button analysis using a number of DRAGEN pipelines (Figure 1). Once secondary analysis is complete, users can easily store, share, and conduct other forms of data management directly in BaseSpace Sequence Hub (Figure 2).

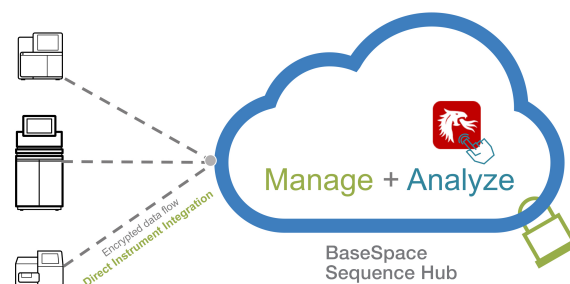


Figure 1: Data management with plug-and-play instrument integration and push-button analysis—Easily connect BaseSpace Sequence Hub to one or multiple Illumina instruments for automatic data transfer from the system into the cloud-based ecosystem for data analysis using DRAGEN apps, and data management, storage, and sharing.

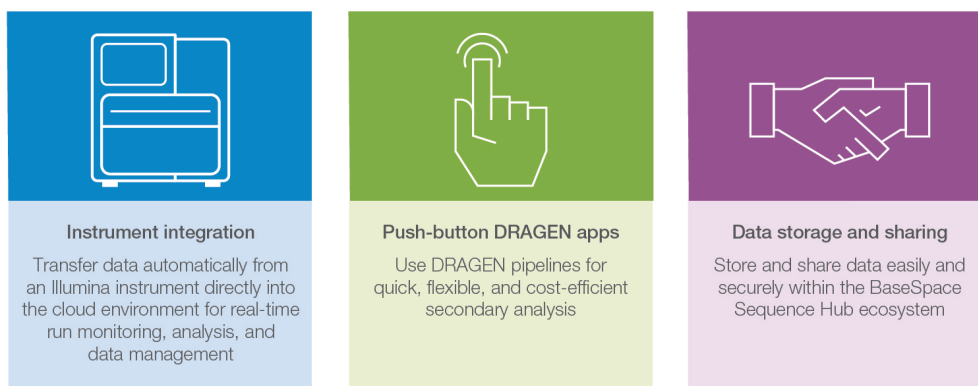


Figure 2: Simplified data analysis—The DRAGEN platform on BaseSpace Sequence Hub couples DRAGEN accuracy and speed with BaseSpace Sequence Hub simplicity and security.

Low-cost, scalable platform

DRAGEN pipelines on BaseSpace Sequence Hub remove the need to purchase on-premise compute and storage, reducing upfront costs, power consumption, and maintenance. DRAGEN apps cost ~ \$5 USD/genome and less than \$2 USD/exome.[†]

DRAGEN pipelines can be used on-demand for small studies or scaled up according to laboratory needs. With BaseSpace Sequence Hub, users can run multiple samples in parallel, and scale up operations without investing in additional hardware infrastructure.

Secure, compliant environment

BaseSpace Sequence Hub imports data directly from the sequencing instrument during the run, enabling customers to begin data analysis as soon as the run completes. Several security measures protect data in transit while communication occurs between the sequencing instruments and the data analysis and storage servers. BaseSpace Sequence Hub has been independently audited and certified for HIPAA compliance,* ISO 27001, and ISO 13485. It is built to enable data privacy and compliance with GDPR, including end-to-end encryption, auditing, and fine-grained access control (Figure 3).



Figure 3: Security-first BaseSpace Sequence Hub —Independently audited and certified for HIPAA compliance, ISO 27001, ISO 13485, and GDPR readiness.

[†]Approximate cost; varies based on sample

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Free trial

BaseSpace Sequence Hub offers a limited 30-day free trial for new accounts. New free trial accounts have access to:

- 1 TB free storage — Purchase additional storage with promotional iCredits
- 250 iCredits — Use for additional storage, compute, and third-party app fees
- All BaseSpace Sequence Hub apps

Contact your local sales representative to upgrade the free trial to a Professional or Enterprise subscription account.

Learn more

To learn more about the DRAGEN Bio-IT Platform, visit www.illumina.com/DRAGEN. To learn more about BaseSpace Sequence Hub, visit www.illumina.com/basespace.

References

1. Precision FDA Hidden Treasures Warm Up. precision.fda.gov/challenges/1/view/results. Accessed September 14, 2018.
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