

DRAGEN™ Pipelines on BaseSpace™ Sequence Hub

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

- Analyze a whole human genome in ~35 minutes or a whole human exome in ~ 8 minutes with high analytical sensitivity and specificity
- Stream data directly from the sequencing system to BaseSpace Sequence Hub to launch DRAGEN analysis pipelines simply
- Operate in the cloud and use resources on-demand to minimize costs and meet laboratory needs
- Assure data privacy with a security-first platform that is independently audited and certified with various regulations



Introduction

Illumina DRAGEN software provides accurate and rapid secondary analysis of next-generation sequencing (NGS) data. DRAGEN secondary analysis 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 DRAGEN software with the user-friendly interface and low-cost pricing model of BaseSpace Sequence Hub enables users of all levels of bioinformatics expertise to use leading analysis tools to extract meaningful insights from sequencing experiments.

Accurate, fast analysis

DRAGEN secondary analysis generates exceptionally accurate results. In the 2020 Precision FDA Truth Challenge V2 (PrecisionFDA V2), DRAGEN v3.7 won most accurate in All Benchmark Regions and Difficult to Map regions for Illumina sequencing data.^{1,2} In addition to accuracy, DRAGEN software enables rapid secondary analysis, as demonstrated by an independent institution that used the platform to set a speed record in genomic analysis.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 DRAGEN software address common challenges in genomic analysis, such as lengthy compute times, consistent accuracy, and massive volumes of data. Various DRAGEN pipelines are currently available on BaseSpace Sequence Hub to support multiple sequencing applications (Table 1). Additional pipelines and new versions are released on a regular cadence.

 For a comprehensive list of pipelines, visit illumina.com/DRAGEN.

Simple workflow

DRAGEN software on BaseSpace Sequence Hub integrates leading secondary analysis pipelines into a simple workflow. Users can monitor runs in real time and stream data securely and directly from instruments into the cloud

Table 1: DRAGEN pipelines 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, scales to thousands of samples at expedited speeds with uncompromising accuracy
DRAGEN Methylation Pipeline	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	Proprietary reference used by the DRAGEN apps built with FASTA files

ecosystem for push-button analysis using several DRAGEN pipelines (Figure 1). After secondary analysis is complete, users can easily store, share, and conduct other forms of data management directly in BaseSpace Sequence Hub (Figure 2).

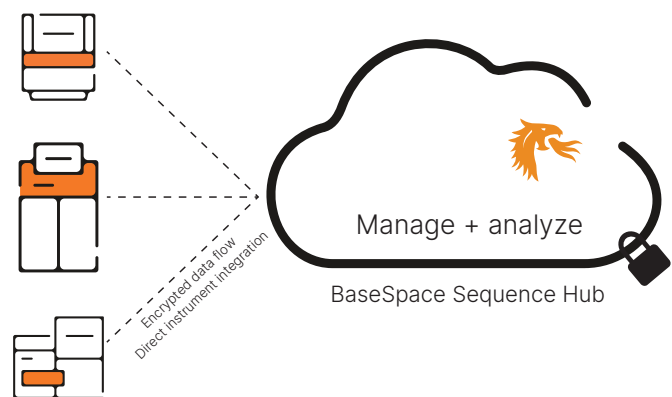


Figure 1: Data management and analysis—Connect one or more Illumina instruments to BaseSpace Sequence Hub for automatic data transfer, analysis using DRAGEN apps, management, storage, and sharing.

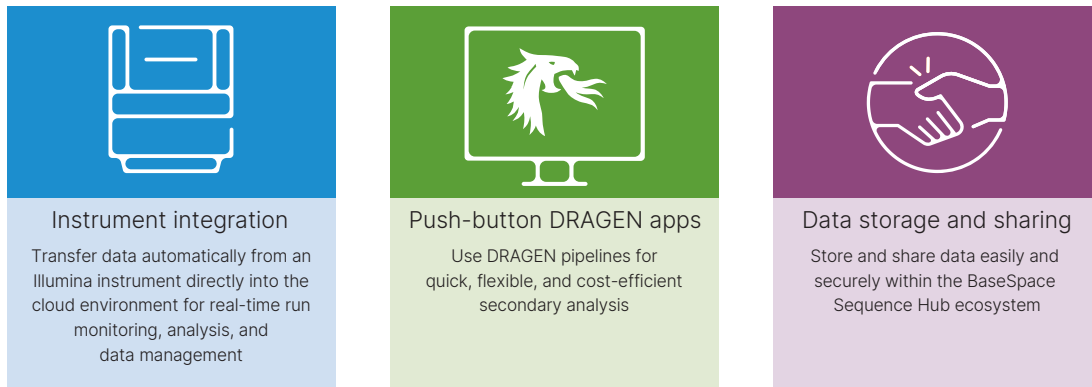


Figure 2: Simplified data analysis—DRAGEN secondary analysis on BaseSpace Sequence Hub couples accuracy and efficiency with simplicity and security.

Low-cost, scalable platform

DRAGEN pipelines on BaseSpace Sequence Hub remove the need to purchase on-premises computing 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 when 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 Health Insurance Portability and Accountability Act (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).

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.

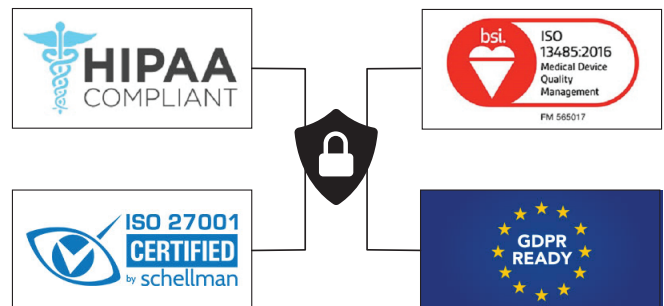


Figure 3: Security-first BaseSpace Sequence Hub— Independently audited and certified for HIPAA compliance, ISO 207001, ISO 13485, and GDPR readiness.

Learn more

[DRAGEN secondary analysis](#)

[BaseSpace Sequence Hub](#)

References

1. Food and Drug Administration. Truth Challenge V2: Calling Variants from Short and Long Reads in Difficult-to-Map Regions. precision.fda.gov/challenges/10. Accessed March 14, 2024.
2. Illumina. DRAGEN Sets New Standard for Data Accuracy in PrecisionFDA Benchmark Data. Optimizing Variant Calling Performance with Illumina Machine Learning and DRAGEN Graph. illumina.com/science/genomics-research/articles/dragen-shines-again-precisionfda-truth-challenge-v2.html. Accessed March 14, 2024.
3. The San Diego Union Tribune. Rady Children's Institute sets Guinness world record. February 12, 2018. sandiegouniontribune.com/news/health/sd-no-rady-record20180209-story.html. Accessed March 14, 2024.



1.800.809.4566 toll-free (US) | +1.858.202.4566 tel
techsupport@illumina.com | www.illumina.com

© 2024 Illumina, Inc. All rights reserved. All trademarks are the property of Illumina, Inc. or their respective owners.
For specific trademark information, see www.illumina.com/company/legal.html.
970-2019-015-B