DATA SHEET

DRAGEN™ apps on BaseSpace™ Sequence Hub

Accurate, comprehensive, and efficienct analysis in an easy-to-use, cloud-based environment

- Stream data directly from the sequencing system to BaseSpace Sequence Hub for seamless launch of DRAGEN secondary analysis applications
- Operate in the cloud and use resources on demand to minimize costs and meet laboratory needs
- Help ensure data privacy with a security-first platform that is independently audited and certified to global standards

illumina®

Introduction

Illumina DRAGEN software provides accurate, comprehensive, and efficient secondary analysis of next-generation sequencing (NGS) data. DRAGEN secondary analysis applications and pipelines are available on BaseSpace Sequence Hub, an easy-to-use, secure cloud-based software for simplified run management, monitoring, and bioinformatics. Combining the accuracy and speed of DRAGEN software with the user-friendly interface and low-cost pricing model of BaseSpace Sequence Hub enables meaningful insights from sequencing experiments for users of all informatics experience levels.

Accurate and comprehensive analysis

DRAGEN secondary analysis generates exceptionally accurate and comprehensive 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, DRAGEN software enables rapid secondary analysis, as demonstrated by an independent institution that set a speed record using the platform in genomic analysis.[3] This optimized performance is available for a wide variety of genomic analysis solutions, including binary base call (BCL) file conversion, mapping, alignment, sorting, duplicate marking, and haplotype variant calling. As described in the study published in *Nature Biotechnology*, DRAGEN software addresses common challenges in genomic analysis, such as lengthy compute times, call consistency, and handling large volumes of data.[4] Several DRAGEN apps are available on BaseSpace Sequence Hub to support multiple sequencing applications (Table 1). New apps and updated versions are released on a regular cadence.

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

Simple workflow

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

Table 1: Example DRAGEN apps available on BaseSpace Sequence Hub

App	Description
DRAGEN Germline	Mapping and aligning to reference for variant calling; includes advanced error model calibration for increased accuracy and repeat expansion detection and genotyping through Illumina Expansion Hunter
DRAGEN Somatic	Somatic variant detection in tumor samples; includes tumor-only and tumor–normal modes
DRAGEN Enrichment	Combines DRAGEN Germline and Somatic callers into a pipeline designed to analyze enrichment samples; includes a full suite of enrichment metrics and reporting
DRAGEN RNA	Rapid alignment and splice junction mapping, quantification, and fusion detection
DRAGEN Joint Genotyping/ Population	Joint variant calling across multiple genomes; scales to thousands of samples at expedited speeds with uncompromising accuracy
DRAGEN Methylation	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	Accepts FASTA file to build a proprietary, non-standard DRAGEN reference

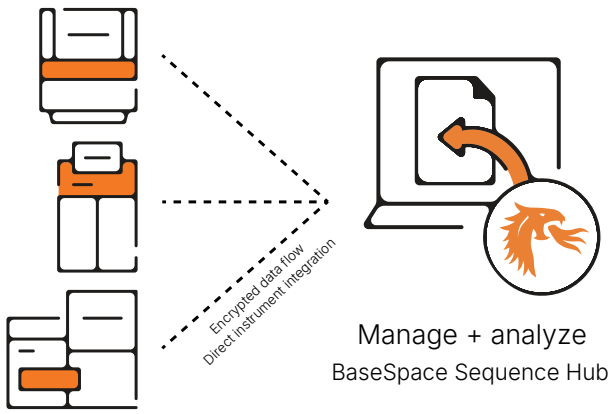

Figure 1: Data management and analysis—Connect one or more Illumina instruments to BaseSpace Sequence Hub for automatic data transfer, analysis using DRAGEN apps, management, storage, and sharing.

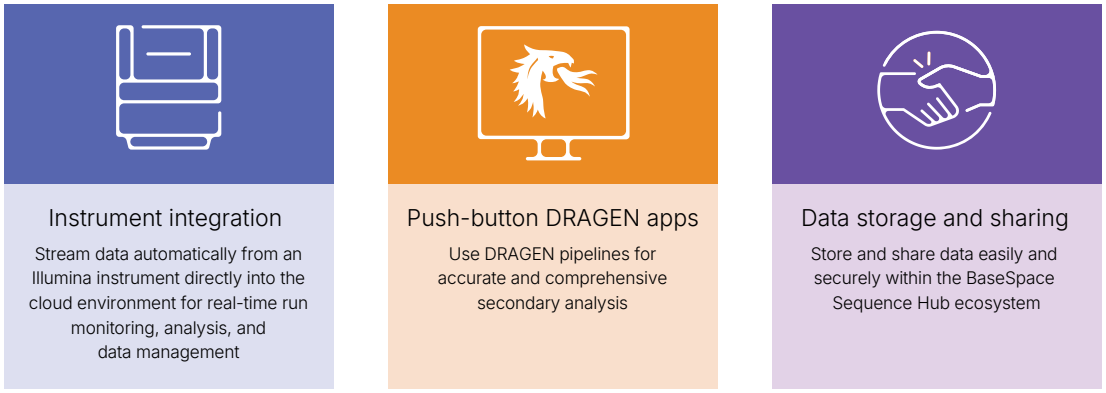

Figure 2: Simplified data analysis—DRAGEN secondary analysis on BaseSpace Sequence Hub couples accuracy and efficiency with simplicity and security.

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 on BaseSpace Sequence Hub (Figure 2).

Low-cost, scalable platform

DRAGEN apps on BaseSpace Sequence Hub eliminate the need to purchase on-premises computing and storage, reducing upfront costs, power consumption, and maintenance. DRAGEN apps cost ~8 iCredits/genome and 2 iCredits/exome for small variant calling.*

DRAGEN apps 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

Security is of paramount importance when making the decision to move genomic data to cloud-based analysis and storage. In BaseSpace Sequence Hub, data are protected through several physical, electronic, and administrative measures. Data for upload are encrypted using the AES256 standard and protected by transfer layer security (TLS). Data within BaseSpace Sequence Hub are hosted on Amazon Web Services (AWS), which is compliant with industry-accepted security standards.[5] Enterprise subscriptions offer an additional level of security. Enterprise customers are provided their own domain and the ability to use their own SAML 2.0-supported authentication service to manage users and passwords. BaseSpace Sequence Hub also supports Enterprise customers in a Health Insurance Portability and Accountability Act (HIPAA)-regulated environment with a Business Associate Agreement (BAA). More information about security features can be found in the BaseSpace Sequence Hub security and privacy brief.

Free trial

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

- 1 TB included storage
- 250 iCredits—used to purchase additional data storage and analysis options
- All BaseSpace Sequence Hub apps
- Access to instrument run monitoring capabilities
- Included demultiplexing for runs streamed into a customer's account

For a free trial or to upgrade to a Professional or Enterprise subscription account, visit BaseSpace Sequence Hub Ordering, or contact your local sales representative.

* Actual analysis costs vary based on input sample characteristics, analysis parameters selected, instance type, and location.

Learn more

DRAGEN secondary analysis

BaseSpace Sequence Hub

References

1. US Food and Drug Administration. PrecisionFDA 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 sccuracy 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. Published 2022. Accessed March 14, 2024.
3. The San Diego Union-Tribune. Rady Children's Institute sets Guinness world record. sandiegouniontribune.com/news/health/sd-no-rady-record20180209-story.html. Published 2018. Accessed March 14, 2024.
4. Behera S, Catreux S, Rossi M, et al. Comprehensive genome analysis and variant detection at scale using DRAGEN. *Nat Biotechnol*. Published online October 25, 2024:1-15. doi:10.1038/s41587-024-02382-1
5. AWS Cloud Security. aws.amazon.com/security/. Accessed March 13, 2020.

illumina®

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

M-GL-03292 v1.0