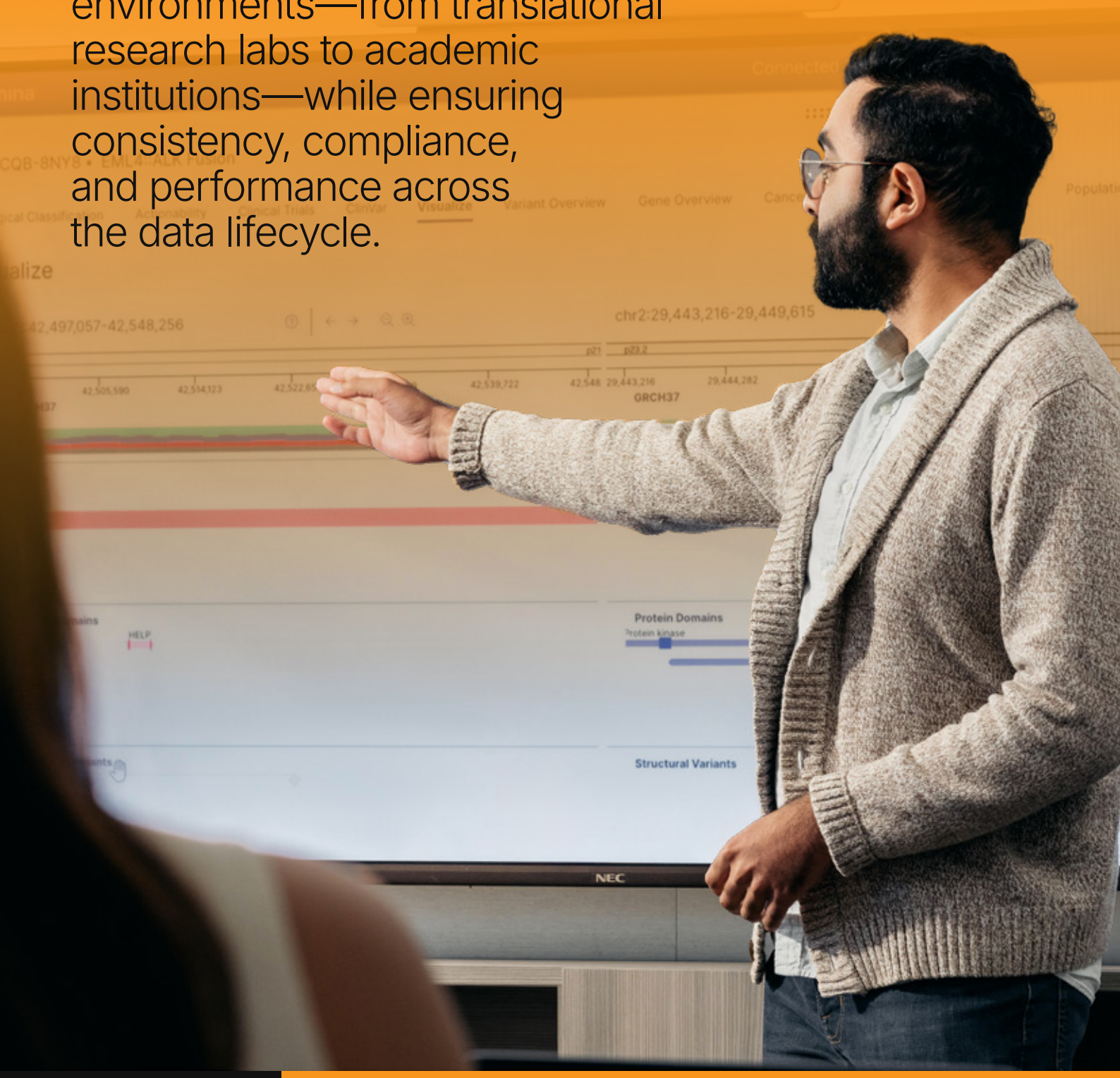
illumina®

Illumina Connected Software

Deeply Integrated Data Analysis and Interpretation

Illumina Connected Software is a secure, enterprise-ready suite built on a modular infrastructure to support high-quality data interpretation at scale.

Its flexible architecture is designed to meet the demands of diverse environments—from translational research labs to academic institutions—while ensuring consistency, compliance, and performance across the data lifecycle.



Omics Without Silos: A Unified Software Experience

Secondary and tertiary analysis solutions across sequencing applications are traditionally disjointed, often leading to issues in dataset interoperability, scalability, and long-term utility. In the absence of fully workflow-integrated analysis pipelines, organizations have adopted a mix of proprietary and open-source tools, many of which are not designed to work together, function at scale, or accommodate diverse data inputs. Illumina Connected Software removes barriers between assays and informatics, seamlessly linking samples to insights across use cases, from multi-omic exploration to population-scale, clinically-relevant research.

Built for current and future omics initiatives

Designed for the Future of Omics
One environment that seamlessly integrates a broad menu of applications to help researchers unlock deeper insights from one to many omes at virtually any scale.

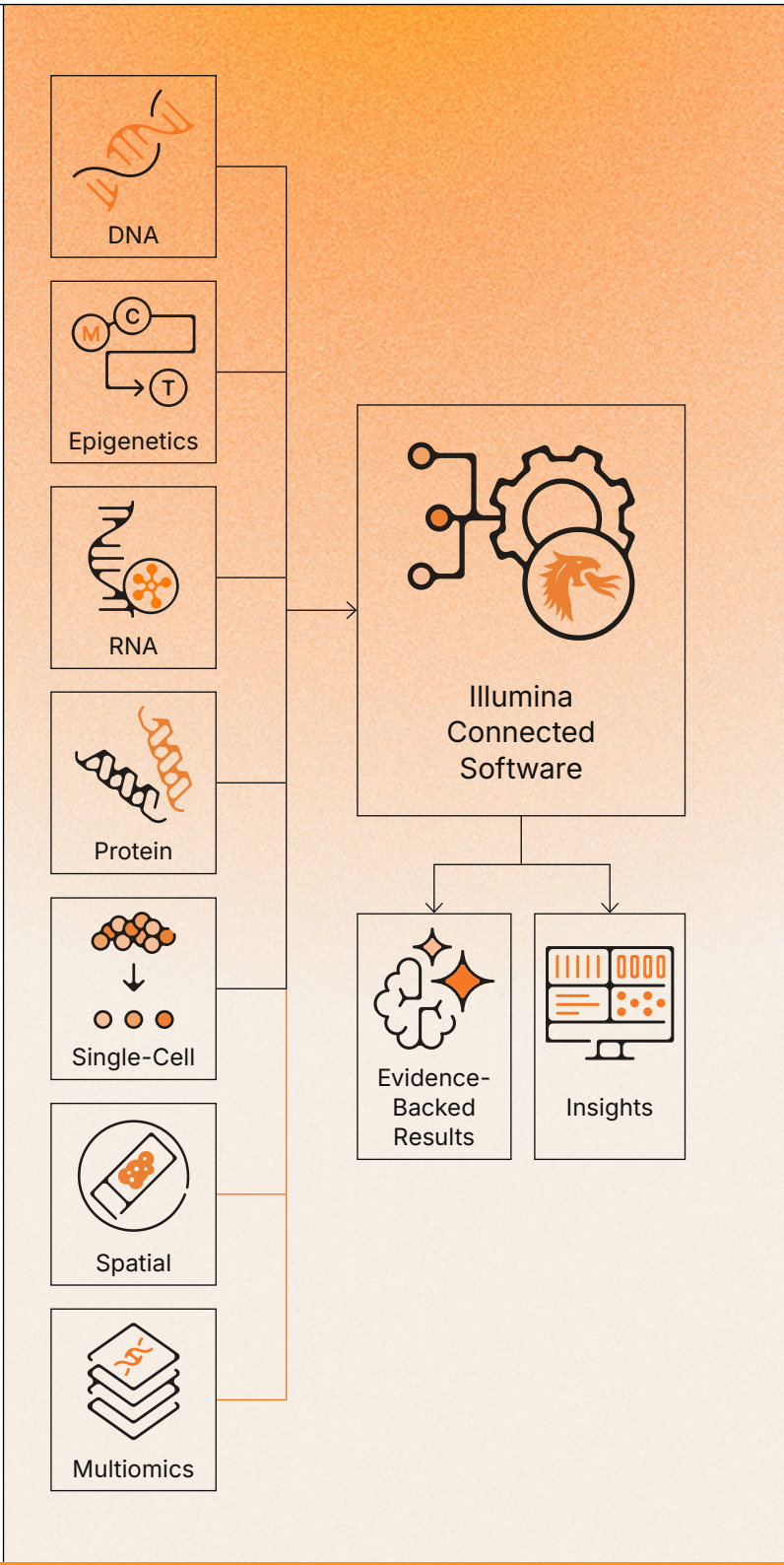
Enterprise-Ready Infrastructure
Designed to reliably handle datasets ranging from individual projects through national genomics programs and multi-institutional collaborations.

End-to-End Workflow Coverage
From sample accessioning to secondary analysis, to discovery and insight—Connected software delivers an integrated data journey.

AI-Powered Insight & Connected Annotations
Accelerate interpretation with integrated outputs from groundbreaking AI tools like PrimateAI-3D, paired with continuously updated, context-aware annotations that span cohorts, diseases, and data types.

Security, Compliance and Collaboration
Built with global standards, robust security, compliance and privacy measures in mind to enable secure collaboration across institutions and borders.

Fully Integrated and Streamlined
Connect powerful informatics with the industry's most-comprehensive set of instruments and omics approaches to reduce touchpoints and streamline data flow saving transfer time and energy.



Sample and run management

Clarity LIMS™: Sample and workflow management software optimized for genomics labs. Improve efficiency, ensure traceability, and manage complex workflows at scale.

Secondary Analysis

DRAGEN™ Secondary Analysis: Highly accurate, comprehensive and efficient secondary analysis. 4-time winner of the PrecisionFDA Truth Challenge. Includes iterative genotyping and aggregation and batchwise processing to enable scalable population-level variant calling without re-processing-the full cohort (DRAGEN IGG).

Data Curation and Management

Illumina Connected Analytics (ICA): A secure cloud-based data platform for data curation, storage, and analysis—built for collaboration, reproducibility, and cohort-scale exploration.

- Create and Maintain Cohorts—ICA Cohorts
- Accurately Annotate Variants—Connected Annotations
- Data Aggregation and Querying—ICA Base

Discovery, Insight, Interpretation

Emedgene™ (for Genetic Disease): Illumina's premier variant interpretation software for rare disease and other research applications that streamlines tertiary analysis and reporting workflows, powered by explainable AI.

Illumina Connected Insights (for Oncology): ICI streamlines clinical research reporting in oncology with configurable evidence-driven decision support integrated into your workflow.

Illumina Connected Multiomics: is an interactive platform for visualizing and exploring multiomic and multimodal datasets. Produce publication-ready figures and findings for single-cell, spatial, 5-base WGS (methyl-seq), transcriptomics, and more.

Illumina Connected Annotations: A powerful AI-driven tool used to analyze, annotate and interpret datasets. Connect your data with PrimateAI-3D, SpliceAI, COSMIC, and OMIM—adding a full suite of AI annotations to an Illumina genome in <1min and saving 50-70% time per case with prioritization and evidence.

> “Illumina Connected Multiomics is a powerful solution that integrates multiomic and multimodal analysis.”

Connected Solutions Spanning the Research Continuum

1. Discovery | 2. Clinical Oncology Research | 3. Clinical Genetic Disease Research

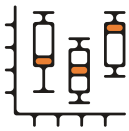
Illumina Connected Multiomics: Intuitive, Scalable Multiomic Data Analysis

Illumina Connected Multiomics (ICM) is a powerful solution that integrates multiomic and multimodal analysis, creating seamless workflows from sample to insights.

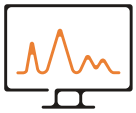
It empowers researchers to delve into complex data with ease, uncover profound biological insights, and accelerate groundbreaking discoveries.

Learn more on the Illumina Website

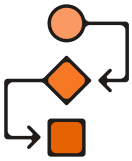
Confidently analyze multiomic data



Interactive Visualizations
Information-rich and publication-ready visualizations.

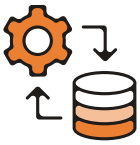


Powerful Statistical Algorithms
Robust statistical methods for identifying key features.



Data Integration and Streamlined Workflows
Built to provide seamless multiomic workflows from assays to insights.

Security-First Infrastructure
Built with industry standard security, privacy, and compliance standards for a trusted data environment.



Integrated Data Management and Efficient Processes
Import, compile, and annotate data with phenotypic metadata to produce detailed studies.

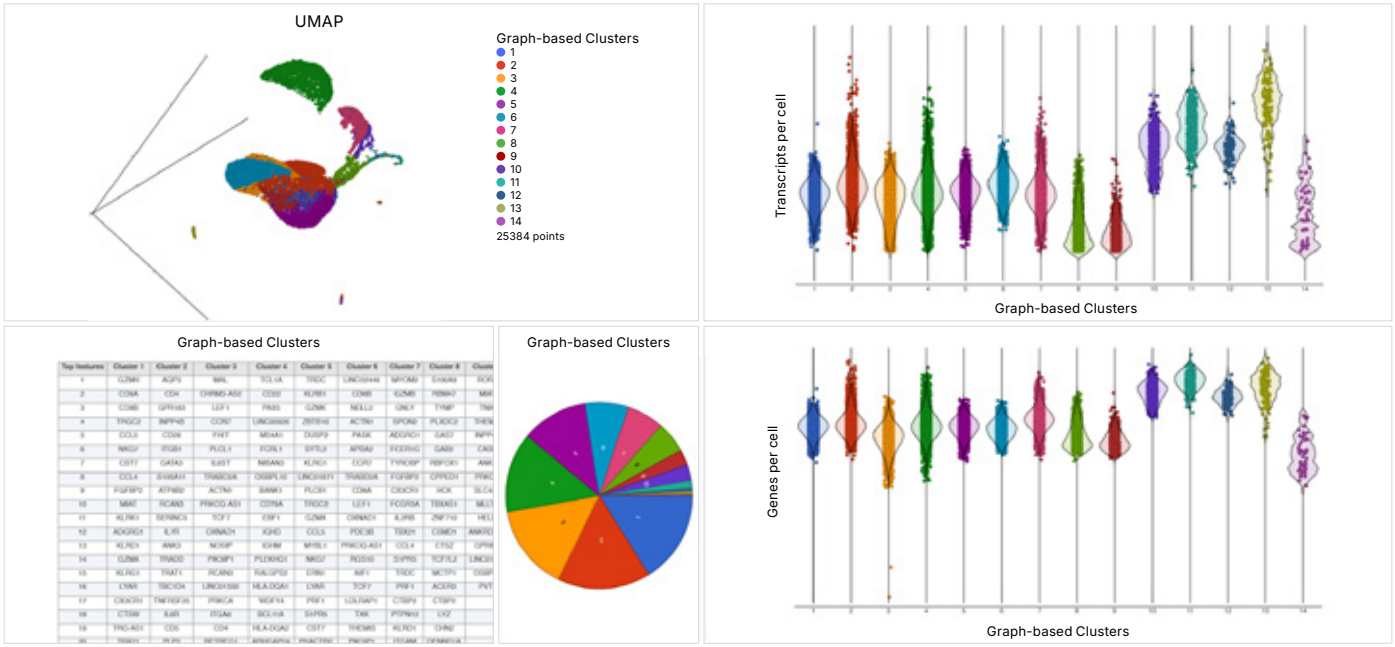
Adaptable and Scalable Solutions
Designed to scale for multiomics studies of any size and support diverse sample types including bulk, single-cell, spatial, protein, methylation, and more.

Streamlined, End-to-End Workflows

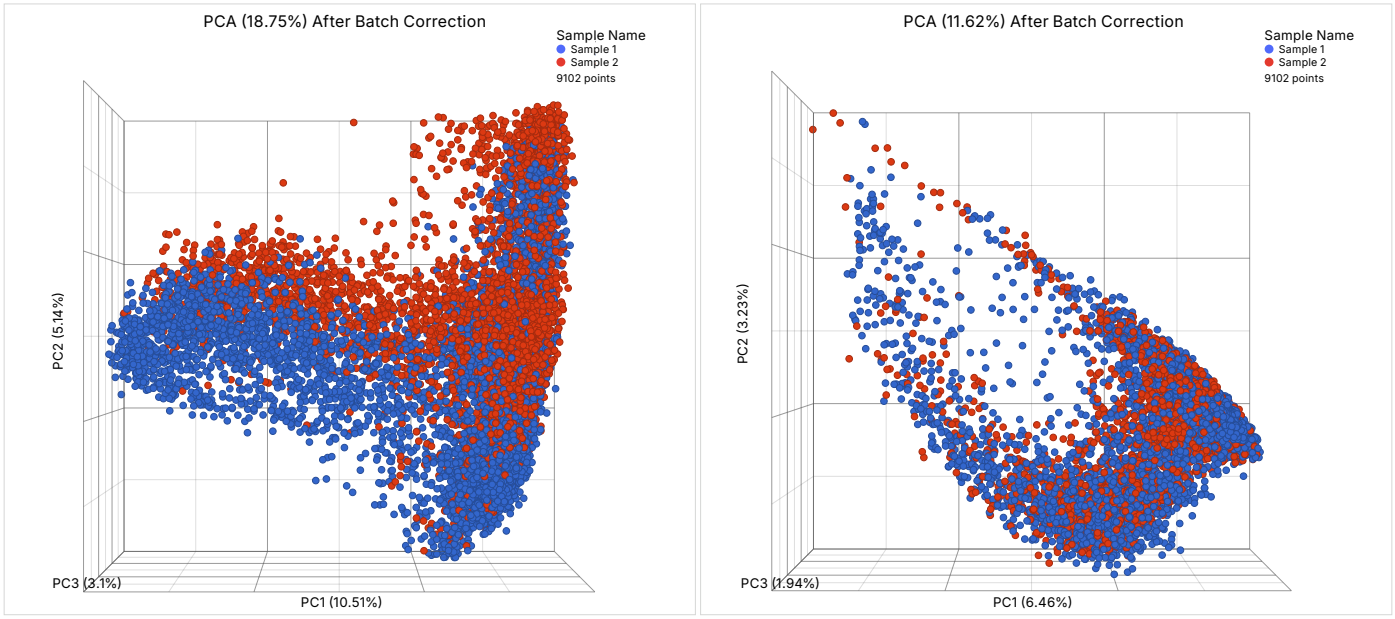
Out-of-the-box workflows support Illumina assays and leverage the accuracy of DRAGEN secondary analysis. Together, DRAGEN and ICM make complex data analysis accessible for researchers of all skill levels.

The robust infrastructure is designed for both multiomic and multimodal research including bulk, single-cell, and spatial studies.

Interactive visualization



Single-Cell
Synchronized views show readouts such as cell populations (UMAP), single-cell sequencing metrics (ex: genes/cell, transcripts/cell), differentially expressed genes, and more.



Batch Effect
PCA showing before and after batch effect correction and how correction can help mitigate batch effects.

Connected Solutions Spanning the Research Continuum

1. Discovery	2. Clinical Oncology Research	3. Clinical Genetic Disease Research

Illumina Connected Insights: Overcome Interpretation Bottlenecks in Translational and Clinical Oncology Research

Illumina Connected Insights streamlines interpretation of variants across multiple "omes" by integrating 55 curated knowledge sources with automated sample processing – from data upload through to report generation. This scalable platform supports diverse translational and clinical research applications, enabling labs to accelerate insights while maintaining security, flexibility, and compliance.

Learn more on the Illumina Website

Confidently connect your analysis workflows with published insights

Interpretation Engine
integrate your data with relevant clinical trials, drug labels, guidelines, and more for clinical research.

Multi-Ome, Versatile Results
Identify variants across DNA and RNA signatures, and compare signatures with published disease biomarker patterns.

Users can also add variants not present in the VCF or not called from FASTQ to their case to complement NGS data with data ingested from other genetic tests (long-read sequencing, optical mapping, CGH, SNP array, karyotyping/ FISH, repeat-primed PCR, MLPA, Southern blot, etc).

Seamless Connectivity
Ready to support TSO500 analysis and other oncology workflows, remove unnecessary touchpoints and manual data movement by directly integrating with tools like DRAGEN secondary analysis.

Privacy and Security
Generate your lab's private curated knowledge, backed by security-first infrastructure that conforms to privacy and compliance software requirements and regulations.

Globally-Deployed, Regionally-Relevant
Configure regional content, preferences and language to meet regional needs.

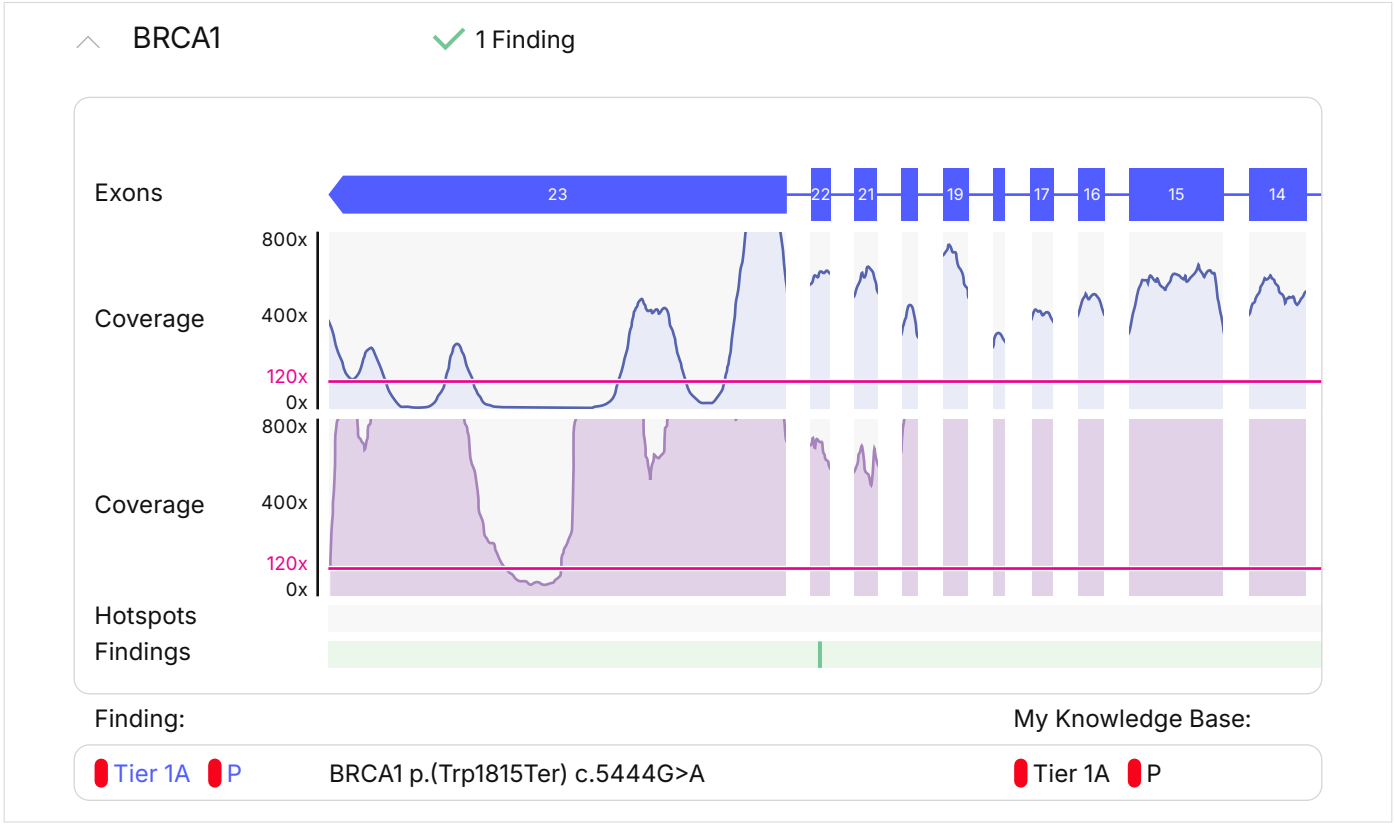


Figure 1 – Key Findings from Default Filters

Connected Solutions Spanning the Research Continuum

1. Discovery | 2. Clinical Oncology Research | **3. Clinical Genetic Disease Research**

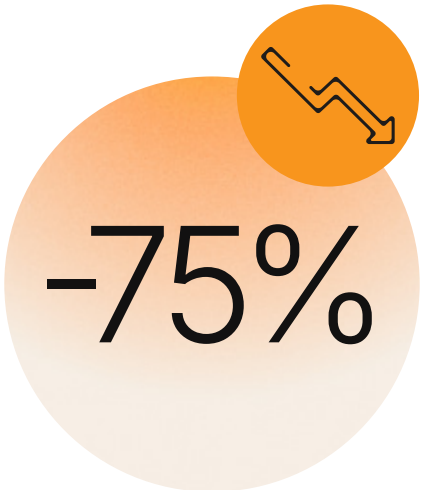
Market-leading AI Germline Variant interpretation for Genetic Disease with Emedgene

Emedgene is a software platform that leverages Explainable AI (XAI) to assist clinical genetic disease researchers in interpreting germline genetic variants, offering a prioritized shortlist of top candidates along with variant-specific explanations. The XAI model was featured in a 2023 validation study published with Baylor Genetics and published in Genetics in Medicine, in which the prioritized variants generated using XAI were successfully validated against manual curation in 98 percent of subject trios, 93 percent of single subjects, and 97 percent overall for subjects analyzed.

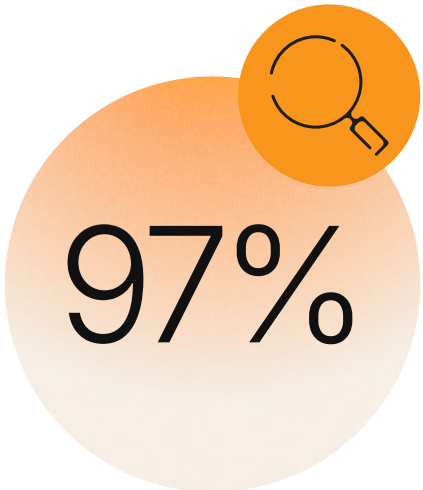
Learn more about Emedgene on the Illumina Website

The role of AI in genomics is to reduce noise and highlight data with the most evidence

Efficiency
Support scale and efficiency gains



Time Per Subject
Reduce turnaround time and costs



Accuracy
Prioritize variants across applications confidently

Emedgene presents the user with a small number of high-confidence candidate variants that are most likely to solve the case

Evidence and Transparency
Confidently interpret variants with evidence-backed, transparent sourcing. Every variant shortlisted by the AI is presented with a full chain of evidence (Fig. 2), including links to databases and publications, opening the AI blackbox and streamlining variant curation.

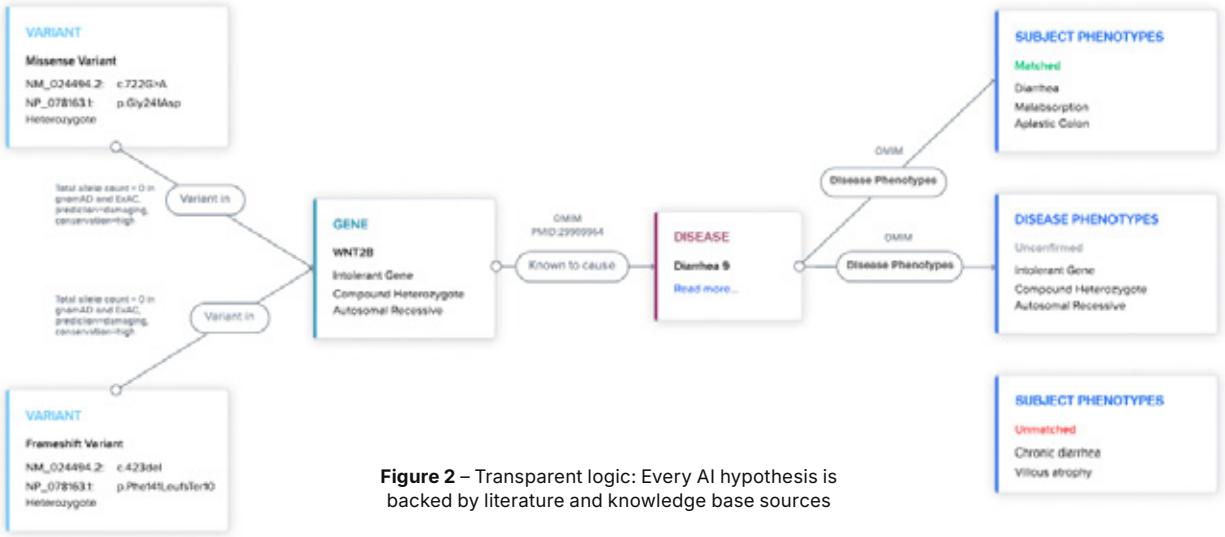


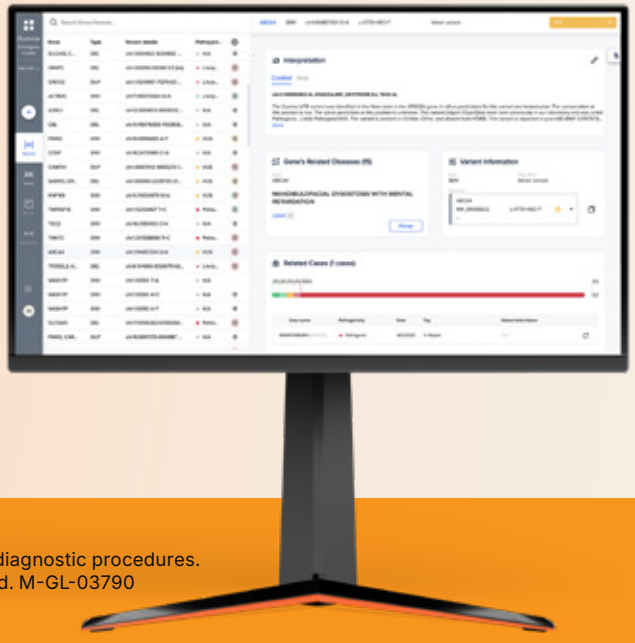
Figure 2 – Transparent logic: Every AI hypothesis is backed by literature and knowledge base sources

Integrated and Streamlined
Emedgene is fully-integrated with DRAGEN secondary analysis, streamlining variant interpretation up to 75% per subject. Automate workflows with API integrations to leading LIMS, storage, pipelines, and more.

Scalable by Design
Confidently keep pace with evolving science, technology, and test volumes with up-to-date knowledge sources, automated curation capabilities, and a team of experts to support your journey. Emedgene scales from single sample workflows to population scale studies.

Flexible Input for Highly-Customized Needs
Adopt a customized workflow for your lab and Interpret results from whole genomes, whole exomes, virtual panels, or targeted sequencing data, all in one platform.

Custom Reporting and Collaboration
Maintain a knowledge base of your organization's curated and tagged genomic data, all of which can be customized and automatically populated into reports. Share curated variants securely across a private network of labs and facilitate team communications.



Try DRAGEN 4.4 Today

Request a FREE demo using the QR code or link*

*https://www.illumina.com/destination/dragen-demo.html

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