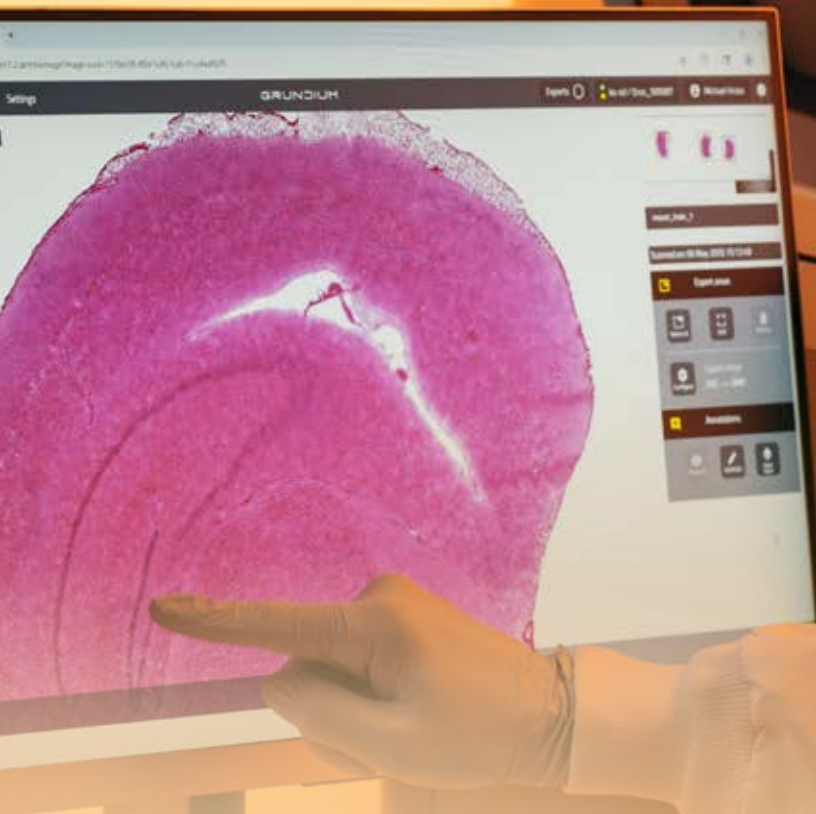




Illumina BioInsight Platform

Deeply Integrated Data
Analysis and Interpretation

The Illumina BioInsight Platform is a secure, unified ecosystem supporting high-quality genomics and multiomics data analysis, interpretation, and integration with knowledge and reference datasets. Analyze across scales, from individual samples populations and cohorts, all in one place.

Its flexible architecture is designed to meet the demands of diverse environments, from pharmaceutical research to academic institutions and population-scale programs, 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. The Illumina BioInsight Platform removes barriers between assays and informatics, seamlessly linking samples to knowledge and interpretation across use cases, from multi-omic exploration to population-scale, translational research.

Built for current and future omics initiatives

Designed for the Future of Omics
Analyze omic data in one environment that seamlessly integrates a broad menu of applications to help researchers maximally extract insights from one to many omes at virtually any scale.

Your Program, Accelerated

When the tools keep pace, your science leads. The BioInsight Platform is designed to handle a range of project sizes so that science can scale without costly infrastructure and maintenance.

Workflow Coverage

BioInsight Platform delivers an integrated data journey. Manage runs, process data, interpret results, explore databases, and product publication-ready figures in one place.

AI-Powered Insight & Annotations

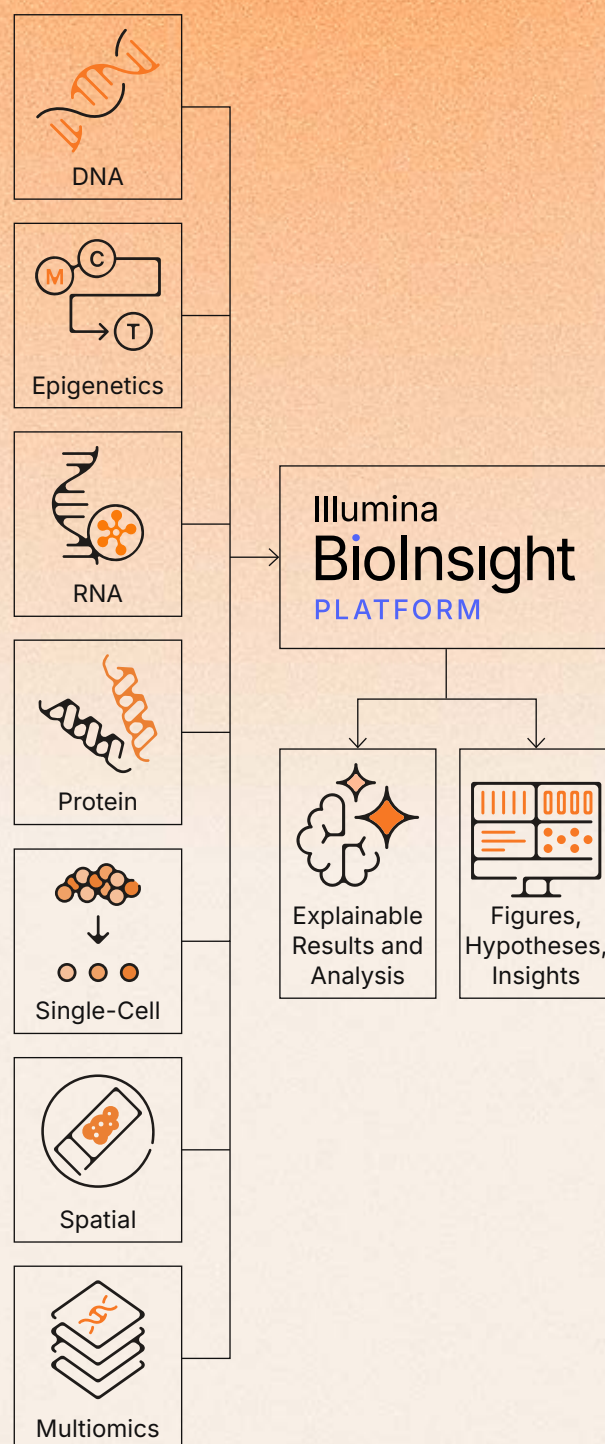
Accelerate interpretation with integrated outputs from groundbreaking AI tools like SpliceAI, paired with continuously updated, context-aware annotations that span cohorts, diseases, and data types.

Security, Compliance & Collaboration

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

Fully Integrated & 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.



Run



BaseSpace™

Sequencer-to-cloud connectivity

Getting access to your data should be simple: Manage your samples and complete workflow, improve efficiency, ensure traceability, and manage complex workflows at scale.

BaseSpace and run management rapidly and seamlessly prepare your data for downstream analysis.

Analyze



DRAGEN™

Secondary analysis

Rapidly call every variant type, quantify every molecular signal, and trust every result from a perpetually-improving engine with over a decade in proven performance. DRAGEN is a 4-time winner of the PrecisionFDA Truth challenge and scales to population-level without re-processing cohorts (DRAGEN IGG).

Interpret



Emedgene™

Variant interpretation
germline



**DRAGEN
Annotation**

AI and machine
learning-enabled
variant prioritization

Uncover new gene-disease connections, resolve and curate variants across basic, cancer, genetic and rare disease research.

BioInsight Platform seamlessly integrates field-leading machine learning models from the BioInsight AI Lab, enabling researchers to evaluate variant pathogenicity, splicing, gene regulation, and disease relevance within a single analysis environment.

Explore



**Connected
Multiomics**

Multiomic discovery

Discover what no single ome can reveal: ICM is an interactive platform for visualizing and exploring multiomic and multimodal datasets. Produce publication-ready figures and findings for single-cell, spatial, methylation, transcriptomics, and more.

Enrich



BioInsight Data

Curated genomic and multiomic data

Curate institutional knowledge and enrich your results with genomic cohorts, multiomic datasets, publications, and public data linked to real-world phenotypes.

Explore discovery cohorts, cellular atlases, proof-of-concept data, and more.



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

Integrated Solutions Spanning the Research Continuum

1. Discover

2. Interpret

3. Translate

Illumina Connected Multiomics: Intuitive, Scalable Multiomic Data Analysis

Illumina Connected Multiomics (ICM) is a powerful solution that integrates multiomic and multimodal analysis, all within an intuitive user interface.

With ICM, researchers can match and integrate omic layers, analyze jointly with collaborators, validate results against over 27,000 curated studies, and build cumulative institutional knowledge.



Learn more about ICM,
schedule a demo, or start a free trial

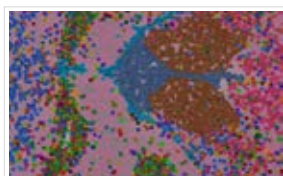
Visit: <https://www.illumina.com/products/by-type/informatics-products/connected-multiomics.html>



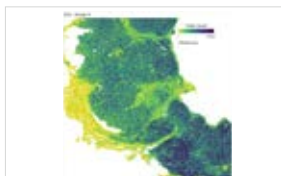
Explore multiomic data to uncover biological insights in context, build cumulative knowledge, and maximize the impact of every dataset.

Discover subtypes invisible to single-omic analysis.

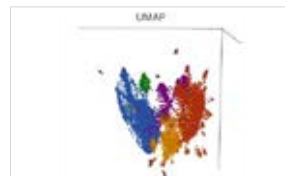
Jointly analyze multiple matched omic layers via integrated multiomics methods.



Spatial Context



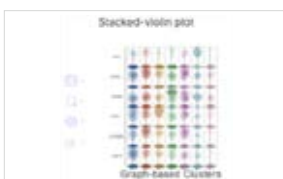
Tissue Organization



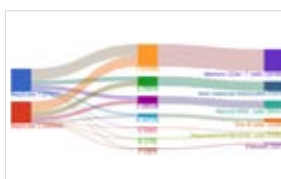
Cell-Type Clustering

Validate findings in a click.

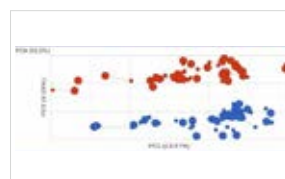
Test molecular signatures against 27K+ curated studies and 20K+ biosets.



Expression Patterns



Cross-Modality Relationships



Molecular Signatures

Ask question in plain language.

Query biological signatures against curated knowledge base atlases using natural language.

Build on every study.

Cumulative insights compound across studies, modalities, and collaborators.



Omic layers jointly analyzed in a single study



Curated biosets for cross-study validation



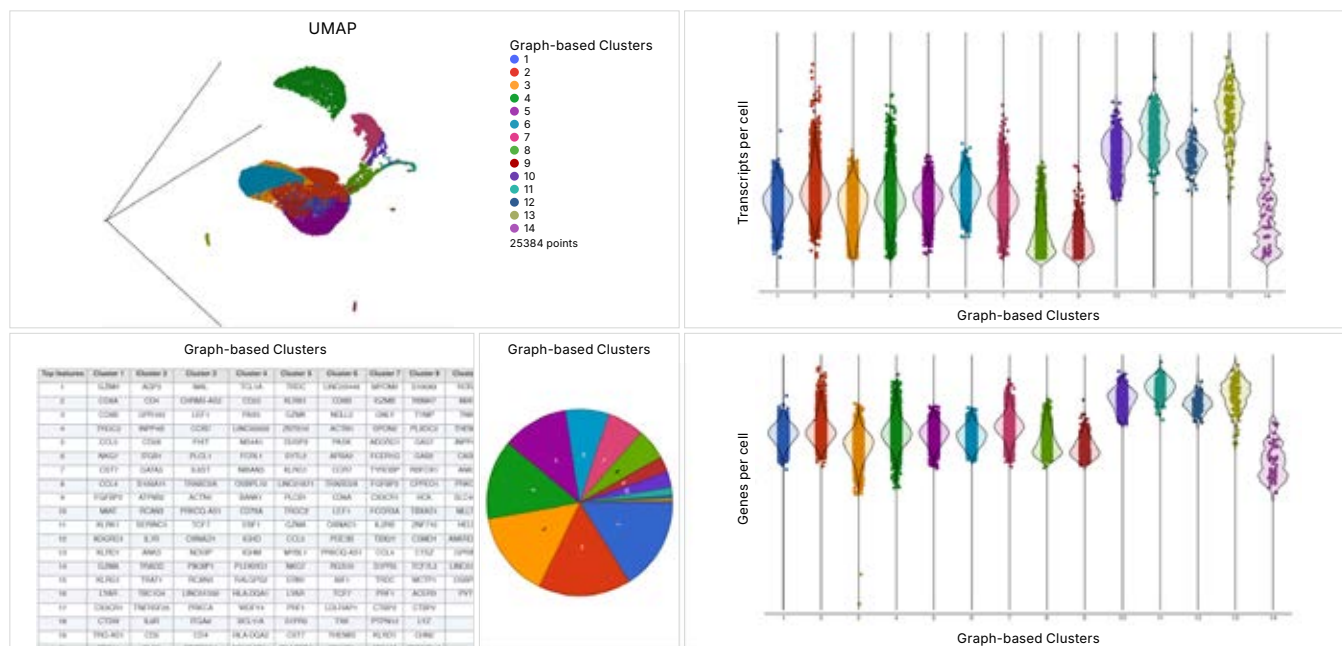
Accessible independent studies

Multiomic data analysis for any lab

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.

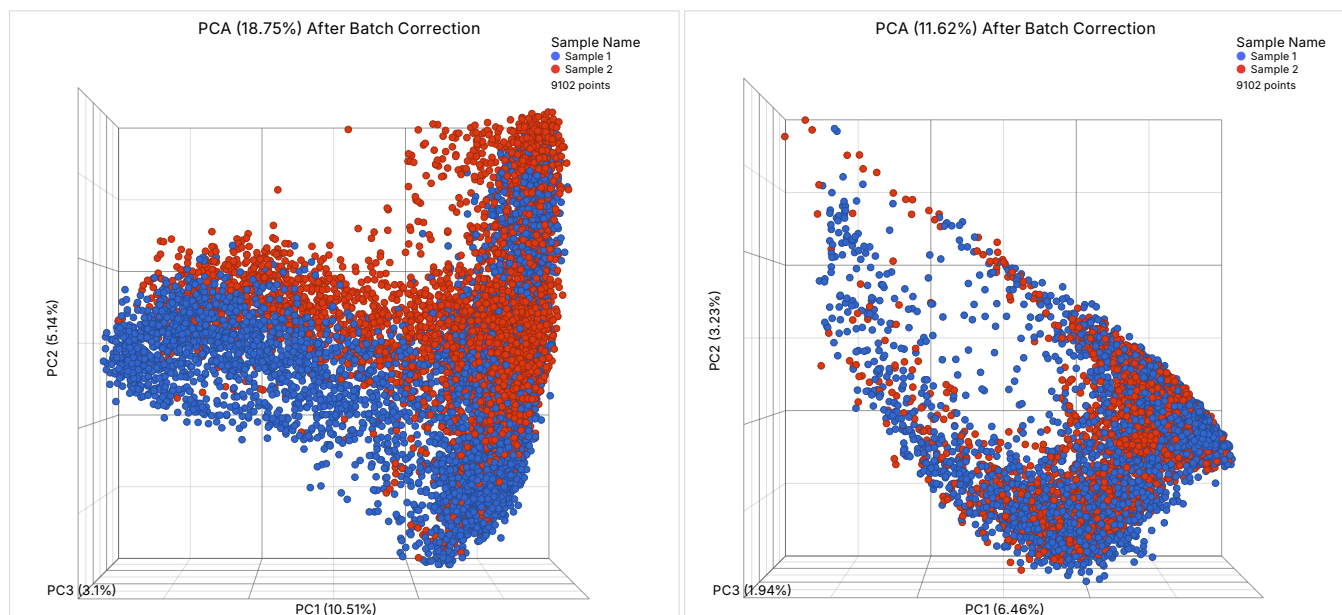
The robust infrastructure is designed to help bioinformaticians focus on meaningful collaborations instead of routine tasks, while also decreasing upkeep costs and maintenance.

Interactive visualization examples:



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.



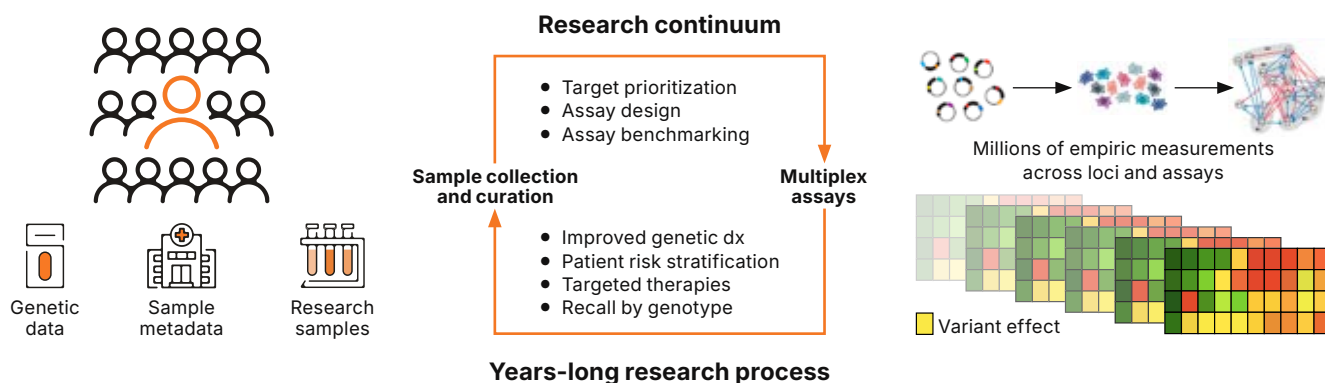
Batch Effect

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

Integrated Solutions Spanning the Research Continuum



Finding a variant is fast.
Understanding its function is often **measured in years**.*



Illumina AI models can reduce the time between research and impact

Rapid variant function prediction can **reduce the time required** to characterize and understand novel variants.

* Gregory M Findlay, Linking genome variants to disease: scalable approaches to test the functional impact of human mutations, Human Molecular Genetics, Volume 30, Issue R2, 15 October 2021, Pages R187–R197, <https://doi.org/10.1093/hmg/ddab219>, <https://globalgenes.org/rare-disease-facts/> (Accessed on 8/17/26)

Illumina's most advanced genome interrogation tools are now easily accessible on the **BioInsight Platform**



DRAGEN Annotation

Introducing: **DRAGEN Annotation**

The BioInsight AI Lab developed DRAGEN Annotation to solve a critical bottleneck in genetic disease research and drug discovery: variant interpretation.

More than 90% of protein-coding variants lack functional characterization, limiting the discovery of disease mechanisms and therapeutic targets.** By integrating advanced genome interrogation capabilities within the BioInsight platform, DRAGEN Annotation helps researchers **interpret the biological impact of genetic variation, uncover gene-disease relationships, and accelerate the path from genomic data to field impact.**

** Tejura M, Chen Y, McEwen AE, Stewart R, Sverchkov Y, et al. A scalable approach to resolving variants of uncertain significance. bioRxiv [Preprint]. 2026 Feb 23:2026.02.14.705848. doi: 10.64898/2026.02.14.705848. PMID: 41727046; PMCID: PMC12918978.C

Understanding the consequences of genetic variation

DRAGEN Annotation reveals how genetic variants influence genes, transcripts, splicing, proteins, and regulatory elements, helping researchers understand the biological mechanisms that drive disease.

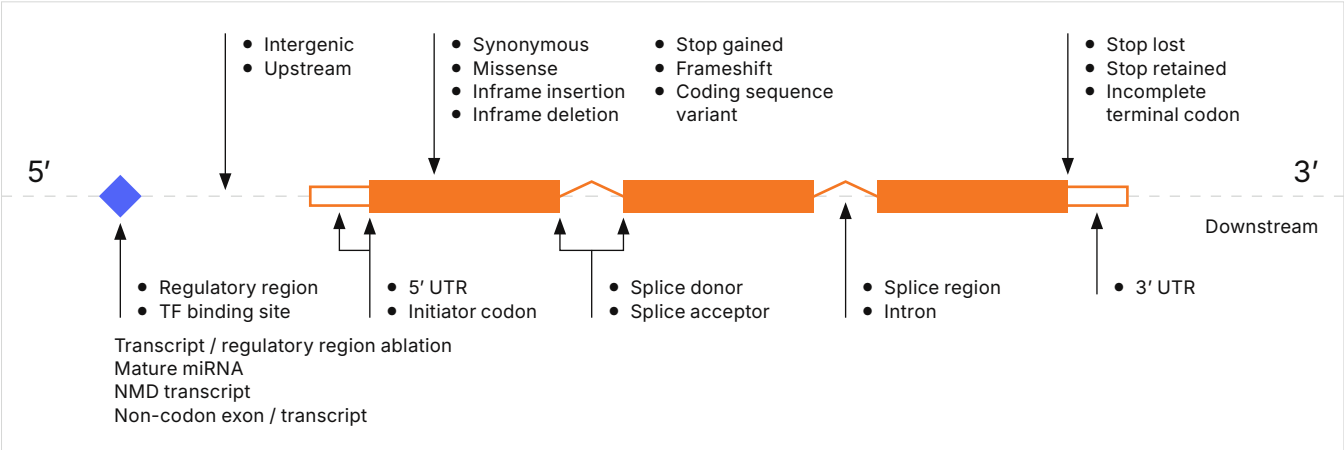
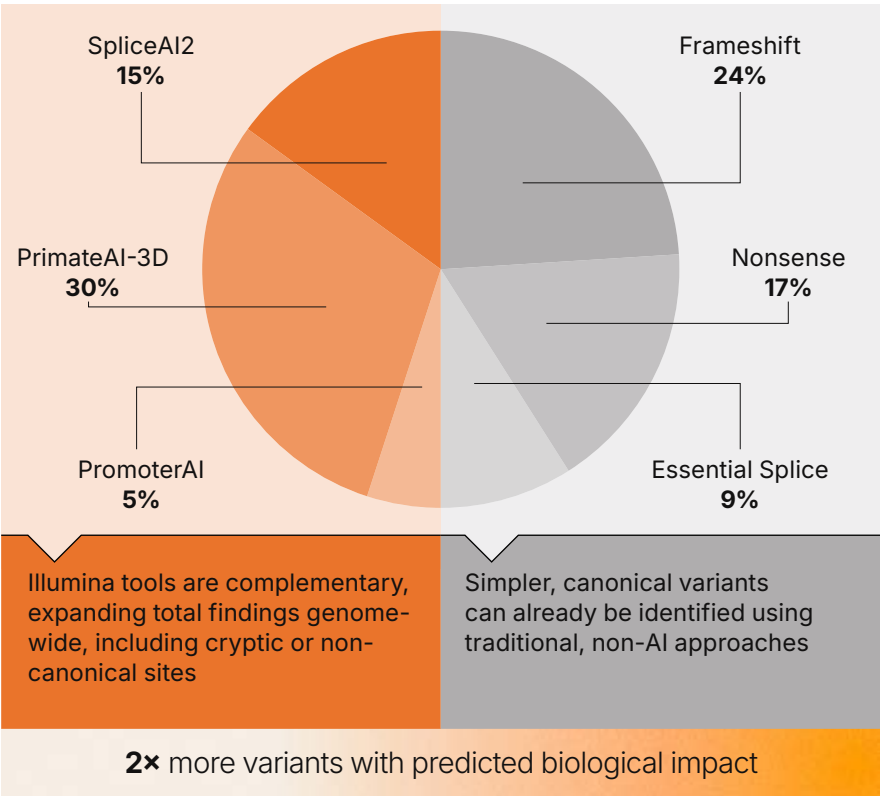


Figure 1 – Examples of DRAGEN Annotation functions. Standardized Sequence Ontology consequence terms help researchers understand how a variant may affect gene structure, splicing, protein function, and other biological processes.



In a rare disease research cohort, twice as many variants with predicted impacts on biology were identified by adding Illumina AI models***

Combined analysis with Illumina AI models finds variants missed by traditional methods



***Source: Illumina R&D

Variant context provided through:

- Primate AI-3D
- Promoter AI
- SpliceAI2
- PhyloPPrimate
- 1000 Genomes Project
- Alpha Missense
- Cancer Hotspots
- ClinGen
- ClinVar
- DANN
- dbSNP
- DECIPHER
- FusionCatcher
- GERP
- GME Variome
- gnomAD (GRCh37)
- gnomAD (GRCh38)
- MITOMAP
- MultiZ 100 way
- REVEL
- TOPMed
- AlphaMissense
- ABRaOM

Integrated Solutions Spanning the Research Continuum



The BioInsight Platform hosts market-leading tertiary analysis solutions for clinical research

The BioInsight platform provides access to multiple tertiary analysis solutions for clinical research, including Emedgene. Emedgene unifies genomic interpretation across diverse germline workflows, enabling researchers to efficiently assess and prioritize findings across multiple assay formats and variant types.



Explainable AI-Powered Prioritization

Leverage explainable AI (XAI) to prioritize disease-relevant variants with integrated genetic, phenotypic, and literature evidence



Configurable Workflows & Automation

Standardize interpretation workflows with configurable SOPs, automation, and ACMG-guided classification support



Curated Evidence Integration

Combine continuously updated public knowledge sources with internal and network-curated data to support faster, more confident analyses



Custom Reporting & Health IT Connectivity

Generate tailored reports and integrate findings seamlessly with laboratory and health IT systems



Learn more about Emedgene, schedule a demo, or try it for free on the BioInsight Platform

Visit: <https://www.illumina.com/products/by-type/informatics-products/emedgene.html>

Prioritized Variant Accuracy

2025 Illumina Study – Evaluated on a proprietary Illumina dataset of 1,395 cases*



98.7% previously reported variants identified by the AI-prioritized list



97% previously reported variants ranked within the top three positions



4 median number of variants prioritized for review

*Illumina data on file, 2025

Baylor Genetics (2023) showed Emedgene's explainable AI flagged 100% of reported variants as candidates, with **96.7% ranked in the top 10**



Read the publication

Visit: <https://ilmn.ly/automated-variant-analysis>

Explainability matters in genomics

Emedgene helps researchers rapidly identify high-confidence, disease-relevant variants while maintaining complete transparency into the underlying evidence. By linking prioritized findings to supporting literature, phenotype

associations, inheritance patterns, and curated databases, Emedgene streamlines variant review and interpretation while avoiding the “black box” approach common to many analysis tools.

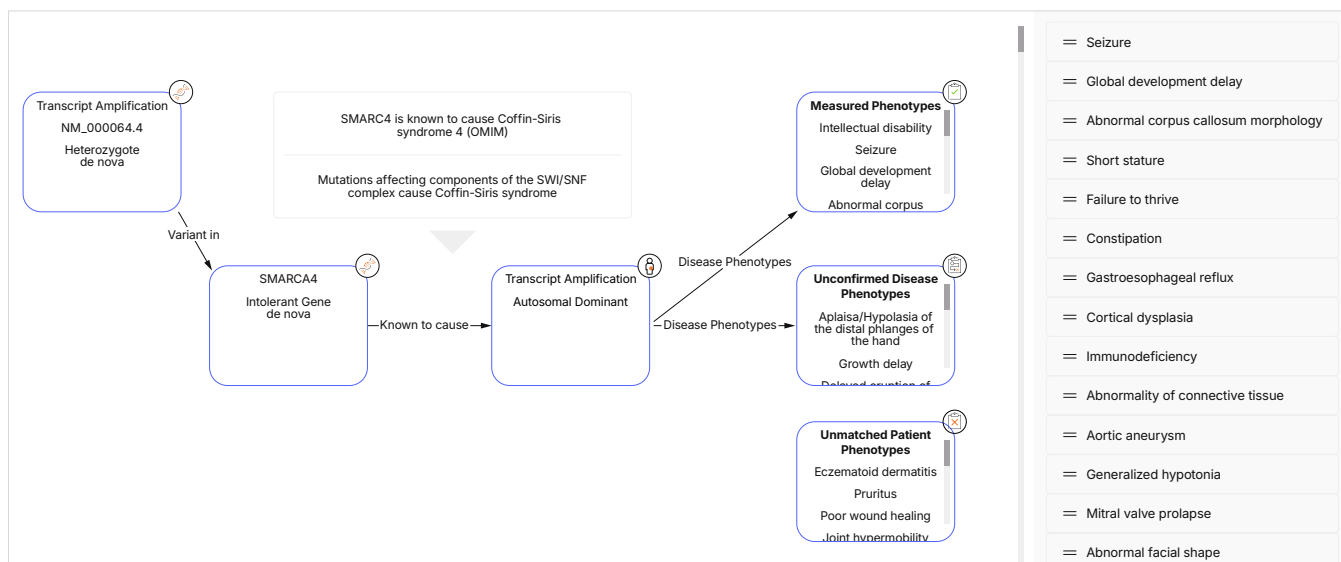


Figure 2 – Transparent logic: Every hypothesis is backed by literature and knowledge base sources

Integrated and Streamlined

Emedgene is fully-integrated with DRAGEN secondary analysis as part of the BioInsight Platform, streamlining variant interpretation up to **75% per subject**. Automate workflows with API integrations to leading LIMS, storage, pipelines, and more.

Scalable by Design

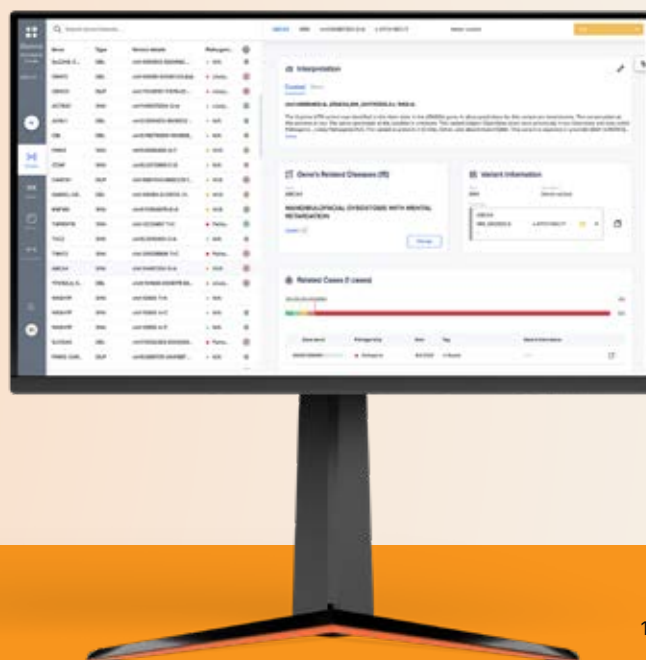
Confidently keep pace with evolving science, technology, and test volumes with up-to-date knowledge sources, automated curation capabilities, and a team of experts to support your journey. Emedgene scales from single sample workflows to population scale studies.

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.



Explore the BioInsight
Platform or request a
personalized demonstration
using the QR code or link



Visit: <https://platform.login.illumina.com/platform-home/choose-subscription-plan?plan=free-trial>