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Emedgene

AI-powered software that streamlines germline variant interpretation for rare disease genomics and other research applications.

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

Overview

Emedgene software, offered by Illumina, is a comprehensive research tool designed by geneticists to streamline tertiary analysis workflows for rare disease genomics and other germline research applications. It supports data from Whole-Genome Sequencing (WGS), Whole-Exome Sequencing (WES), targeted panels, and microarrays. The software is powered by Explainable AI (XAI) and automation, which enables a 2–5x increase in efficiency and a 50–75% reduction in total workflow time per subject compared to manual interpretation.

The XAI prioritizes high-confidence candidate variants and provides a transparent evidence graph, linking to relevant literature and databases, which helps variant interpretation research scientists quickly review and confidently scale their operations. The AI model has been validated with high accuracy (e.g., 97% overall for subjects analyzed in one study).

Key Features and Capabilities:

Explainable AI (XAI): Prioritizes variants with transparent, evidence-backed insights.
Automation: Optimizes workflows for user-defined Standard Operating Procedures (SOPs) across various test types.
Automated Classification: Provides time-saving automated ACMG classifications for comprehensive variant types, including SNVs, indels, CNVs, SVs, and mtDNA variants.
API Interoperability: Powerful API for integrating tertiary analysis with LIMS, storage, and pipelines.
Customizable Reporting: Allows users to customize, edit, and automatically populate research reports, which can be downloaded in PDF or JSON format.
Data Input Flexibility: Compatible with FASTQ and VCF file formats from virtually any secondary variant caller.

Emedgene integrates with Illumina's secondary analysis and data storage platforms, including DRAGEN Secondary Analysis, BaseSpace Sequence Hub, and Illumina Connected Analytics, offering a single-vendor solution from sample processing through research report generation. The software is intended for Research Use Only.

Key Features

- Explainable AI (XAI) for variant prioritization
- Automated ACMG classifications (SNVs, indels, CNVs, SVs, mtDNA)
- High-throughput workflow automation
- Customizable and automated research report generation
- API interoperability for LIMS/storage integration
- Support for WGS, WES, targeted panels, and microarrays
- Configurable Standard Operating Procedures (SOPs)
- Evidence graph compilation with literature/database links

Pricing

Model: enterprise

Pricing is not publicly disclosed. Contact Illumina sales for a custom quotation, subscription information, and a supported evaluation experience.

Target Company Size: medium, enterprise

Integrations

Illumina DRAGEN Secondary Analysis, BaseSpace Sequence Hub, Illumina Connected Analytics, Laboratory Information Management Systems (LIMS) (via API)

Compliance & Certifications

HIPAA, GDPR, ISO 27001, ISO 27701, SOC 1 Type II, SOC 2 Type II, APEC PRP

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