

emedgene

## Improve dry lab efficiency and scale your genomic interpretation operation using an end-to-end AI-based clinical genomics platform

### WHY

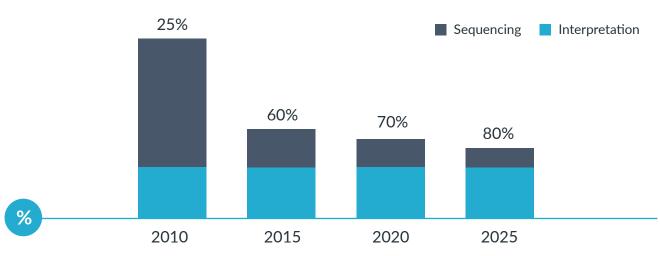
While the cost of sequencing has fallen dramatically, the cost of interpretation remains unchanged. Interpreting today is a labor-intensive effort combining data mining with hours of sifting through publications for relevant information.

To add to the complexity, most pathogenic mutations are unknown. Without incorporating new artificial intelligence tools, the cost of interpretation will continue to rise, and clinical labs will be hard pressed to scale their operations.

### WHAT

The Emedgene automated genomic interpretation platform brings clinical labs the end-to-end AI needed to scale their interpretation operation without increasing headcount, while maintaining high accuracy.





### Interpretation is both a bottleneck and a major cost driver

# Re-Emedgening Genomic Interpretation

# Ada

#### **Al Interpretation Engine**

Pinpoints pinpoints causative mutations and conveys evidence, for an easier path to clinical decisions. Includes:

- The AI knowledge graph, a complete map of variant, gene, mechanisms and phenotypes and that is always up to date.
- Uses NLP to incorporate new scientifically validated - unstructured and structured information from publications.
- A proprietary set of machine learning algorithms developed with leading geneticists that regularly generate novel knowledge and can pinpoint causative mutations.

# Wells

The Clinical Workbench

A full-featured NGS lab solution for the entire interpretation process.

- Improve productivity with a managed clinical workflow that facilitates team collaboration and optimizes case results.
- Designed for labs performing a broad portfolio of tests that require NGS quality control.
- Supports clinical reporting and archive, and is built on robust, secure and compliant IT infrastructure.

## BENEFITS

- Increase the accuracy of your analyses
- Pinpoint pathogenic mutations
- Save time with the automatic evidence builder & reporting
- Continuous reanalysis of unresolved cases scales backward testing
- Rigorous scientific oversight
- A full-featured lab solution

# Ada Features



### AI KNOWLEDGE GRAPH

Incorporates published literature and public databases into a single knowledge base with a unified ontology, mapping the complex connections between variants, genes, mechanisms and phenotypes.



### NLP OF PUBLICATIONS

Emedgene uses Natural Language Processing to extract both structured and unstructured information from new scientific publications and textual resources.



Presents all the available data that led to a specific variant identification, in order to provide the most time efficient decision support for geneticists. Evidence can be automatically incorporated in reporting.



Use Ada on Wells, or easily integrate with your in-house tools and workflow or with any other platform.



#### AUTOMATED GENETIC INTERPRETATION

Custom machine learning algorithms that regularly uncover clinically meaningful association models within the AI knowledge graph. Using these algorithms, Emedgene successfully pinpoints the causative mutations for both known and unknown genes.



Unique phenotype matching algorithms that use NLP to extract and map phenotypes and contextualize them from case data.



The emedgene workbench can continuously re-analyze unresolved cases using new information collected by the Al knowledge graph or created via machine learning algorithms.

# Wells Features



MANAGED CLINICAL WORKFLOW



CLINICAL GRADE NGS QUALITY CONTROL

A powerful analysis platform with an intuitive UI built to facilitate team collaboration and optimize case results.



### ROBUST NGS LAB SOLUTION

Broad portfolio of tests including customized gene panels, whole-exome and whole-genome.



#### AUTOMATED REPORTING

Build automatic reporting using the Emedgene API & meet your organizations' reporting standards.



### ROBUST IT INFRASTRUCTURE



COMPLIANT WITH ACMG GUIDELINES

Emedgene is clinically validated in large scale comparative studies. We're collaborating with leading UDN sites on interpreting their most difficult cases and are a provider of the Israeli 100k Genome Project.



Increased yield through automatic identification of mutations



**Time saved** on interpretation & reporting



### LEADING A PARADIGM SHIFT IN GENETIC MEDICINE

emedgene was founded on the vision that genetic medicine will inevitably become a part of the standard of care, and on the strong belief that what has already become a known fact in science, should

be used to serve the community of patients.

We share the commitment of our users, geneticists and clinicians alike, to provide better care to patients in need every day.



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