Congenica®

Enabling genomic medicine

Congenica AI + Exomiser

Focus on the variants that matter

Data interpretation is by far the most time-consuming and resource-intensive step of the NGS workflow. Any time savings that can be found have a significant impact on overall costs and sample throughput. The use of artificial intelligence (AI) combines with other features of the Congenica clinical decision support platform to enable rapid analysis, interpretation and reporting of complex genomic data to aid the diagnosis of genetic diseases.

Efficient prioritization of causal variants

Congenica AI uses machine learning to classify and rank all variants so you can focus on those that matter the most. Providing full transparency via specific lines of evidence supporting each assigned classification and ranking, Congenica AI enables you to work more efficiently, complete cases faster and achieve a high diagnostic yield – all with complete confidence in the quality of your results.

Key benefits of Congenica AI

- ✓ Reduced analysis time (up to 4x faster)
- ✓ Increased diagnostic yield
- ✓ Complete transparency of predictions
- ✓ Greater confidence in your results

Reduce your analysis time while increasing diagnostic yield

Congenica AI uses key attributes including allele frequency, predicted variant effect, predicted deleteriousness and more to classify all variants as Pathogenic (P), Likely Pathogenic (LP), Likely Benign (LB) or Benign (B). Each prediction is rated on a 1-star to 5-star confidence scale.

When Human Phenotype Ontology (HPO) terms are provided, Congenica AI additionally ranks all P/LP variants by their predicted contribution to the patient's phenotypes – both the primary reported phenotype as well as extended phenotypes identified by traversing up the HPO hierarchy. Predictions are based on the gene's OMIM disease association and mode of inheritance (MOI).

Congenica AI's ranking places the causal variant within the top 10 for 85% of diagnosed cases, helping you solve more cases while simultaneously reducing your analysis time.

Solving the unsolvable

When the case of a 1-½ year old boy suffering from epilepsy was reanalyzed, Congenica AI brought the previously overlooked causal variant to the top of the list.

Read the case study

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Complete transparency

Congenica AI predictions are always supported by the individual lines of evidence considered by the model so that you can readily assess the reasons behind each variant's ranking. Congenica AI's built-in explainability enables you to:

- Quickly identify and review phenotypically relevant P/LP variants
- Invest time reviewing ACMG classification criteria, assisted by ACMG auto classification, for only the most likely causal variants
- Confidently ignore B/LB variants

Analyze, interpret and diagnose cases with the assurance of knowing how each variant's ranking was determined.

AI enables confident interpretation of any case

Congenica AI was trained on a data set comprised of more than 35,000 variants derived from over 10,000 patient cases. A broad range of rare disease phenotypes were represented, spanning more than 50 distinct clinical indications. With such a large and varied training set, Congenica AI can be applied to solve any inherited disease case without concerns of bias.

Congenica is a CE Marked In Vitro Diagnostic platform*. As an integral component of the platform, Congenica AI is subject to rigorous quality standards. Between software releases, the model continues to learn in the background from new patient data, further refining its ability to accurately classify and rank variants. Following rigorous testing as part of the software release process, the updated, refined model is introduced, providing access to continued performance improvement within a carefully regulated environment.

Validation study

An accurate model for predicting causal variants

Congenica AI has been demonstrated to:

- have 95.5% accuracy in predicting the pathogenicity of Clinvar 3- and 4-star variants
- rank the causal variant in the top 10 in 85% of cases of solved cases
- correctly reclassify 92.8% of VUS as P/LP or B/LB

Download the study

Combine with Exomiser for even greater variant ranking power

Exomiser – a class-leading variant prioritization tool in its own right – is seamlessly integrated into the platform and made available alongside Congenica AI.

For samples with provided HPO terms, Exomiser prioritizes variants by leveraging information on variant frequency, predicted pathogenicity, and gene-phenotype associations derived from human diseases, model organisms, and protein–protein interactions.

When Congenica AI and Exomiser are used side-by-side, ranking of the causal variant within the top 10 rises to 88% of solved cases.

Discover Congenica today

Discover how Congenica AI helps you focus on the variants that matter.

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Request a demo
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