Genomenon Adds ACMG Classification Features to the Mastermind Genomic Search Engine

Mastermind Uses Machine Learning to Prioritize Medical Literature According to ACMG Variant Interpretation Guidelines

ANN ARBOR, MICHIGAN - April 9, 2018 - Genomenon, the leading genomic search engine company, announced support for ACMG variant classification within the Mastermind Genomic Search Engine. Mastermind users can accelerate their genomic interpretation by searching the scientific literature that fits each individual ACMG classification criteria.

The American College of Medical Genomics, in collaboration with Association for Molecular Pathology (AMP) and the College of American Pathologists (CAP), released updated standards & guidelines for the interpretation of genomic variants in May of 2015. The guidelines recommend the use of specific standard terminology for describing genetic variants as “pathogenic”, “likely pathogenic” or “uncertain significance”, “likely benign” and “benign”. The guidelines also recommend a process for classification of variants into these five categories based on specific scientific evidence.

While some of the evidence can be based on population data or computational data, genomic variants cannot be classified as pathogenic without citing evidence from peer-reviewed scientific literature. Mastermind is the only genomic search engine that provides an extensive search of all the scientific literature according to ACMG classification guidelines. Users can search by disease, gene, and ACMG criteria to find clinically prioritized scientific evidence that can be cited in patient reports.

Mastermind is the world’s first genomic search engine that connects patient’s genomic data with the evidence from the scientific literature, a key requirement in analyzing a patient’s DNA in precision medicine for rare genetic diseases. Genomenon has deployed its proprietary Genomic Language Processing (GLP) algorithms to recognize key genomic information in the medical literature, and utilized advanced machine learning to decipher the clinical relevance of each article according to the different ACMG classification guidelines.

Genomenon will be demonstrating the next generation of the Mastermind Genomic Search Engine, including the new ACMG variant interpretation support, at the American College of Medical Genomics Annual Meeting April 11 – 13 in Charlotte, North Carolina.

About Genomenon
Genomenon connects patient genomic data with evidence from the scientific literature to diagnose and deliver better patient outcomes. Our web-based Mastermind Genomic Search Engine has indexed the scientific evidence from millions of medical articles, cataloguing the genetic relationships between DNA and human diseases, including cancer. For more information, visit www.genomenon.com.

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