Rare Genomics Institute Demonstrates the Power of Genomenon's AI-Based Technology in Diagnosis

Genomenon's Mastermind Genomic Search Engine Connects Patient with Rare Disease Diagnosis

LOS ANGELES, CA; February 28, 2019 -- Rare Genomics Institute, a nonprofit providing direct support to undiagnosed rare disease patients, announced on Rare Disease Day that they were able to diagnose a previously undiagnosed patient with the use of the Mastermind® Genomic Search Engine, a clinical decision support tool by Genomenon®.

The Patient Research Services of the Rare Genomics Institute has created an ecosystem of leading technology partners and genetic experts from top research institutions around the world to give patients pro-bono access to world-class genomic sequencing, data analysis and interpretation services. Often, Rare Genomics works with their partners and volunteer experts to re-analyze cases that have hit a dead end.

Rare Genomics partner Genomenon, a Big Data genomics company, uses Artificial Intelligence (AI) to connect the genetic mutations buried in 30 million medical research publications with patient data obtained from genetic sequencing. Using technology that wasn't available just a few years ago, Genomenon puts the research for over 4.1 million genomic variants at the scientist's fingertips to make sure that no stone is left unturned in providing a comprehensive diagnosis.

In a recent case, a patient had their whole exome sequenced and analyzed by a leading genetics laboratory as part of their long diagnostic odyssey. The lab was unable to find any clinically relevant genetic mutations that could provide a diagnosis. That was where Rare Genomics Institute stepped in; Dr. Lipika Ray, a computational geneticist on the Patient Research Services team, reanalyzed the patient's DNA, which included a search of the Mastermind Genomic Search Engine.

With Mastermind, Dr. Ray was able to find a single research report in the scientific literature that matched the patient's DNA data. The patient in the report shared similar symptoms with the patient being analyzed. With this finding, RG recommended that the patient be re-examined based on the diagnosis found in the scientific research.

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“I can say with certainty that without the findings obtained from Genomenon, I would not have been able to provide a diagnosis for this patient,” said Dr. Ray. “Sometimes there is only one research paper that can connect a patient’s DNA with a diagnosis. Searching through millions of research papers to find a patient’s genetic mutation can be like trying to find a needle in a haystack. With advanced AI techniques used by Genomenon, the needle can pop right into view.”

“This case is the perfect example of the innovative opportunities we try to bring to rare disease patients that have been fighting for so long with no answer,” said Romina Ortiz, COO Rare Genomics.

Find the complete diagnosis story on RareGenomics.org or Genomenon.com.

About Rare Genomics Institute
Rare Genomics Institute (RG) was founded in 2011 to fill the health care gap for undiagnosed rare disease patients and supporting research in rare diseases. RG helps rare disease patients find a diagnosis, treatment, and pathway to a cure by individualized access, coordination and execution of genetic sequencing and research services with RG and its affiliates.

RG also supports rare disease advocacy by fostering an online community of rare disease patients, and supporting rare disease research through a yearly grant competition. We hope that these efforts slowly push science and care forward to meet the needs of the patients affected by rare diseases. For more information, visit RareGenomics.org.

About Genomenon
Genomenon powers evidence-based genomics for faster; more comprehensive diagnosis and treatment decisions. Their flagship product, the Mastermind Genomic Search Engine provides immediate insight into the published genomic research for every disease, gene, and variant found in the literature.

Used by hundreds of diagnostic labs around the world, Mastermind accelerates genomic interpretation by providing unique insight into genomic relationships found in the full text of millions of scientific articles.

Pharmaceutical researchers license the Mastermind database for a comprehensive genomic landscape associated with any given disease – to identify and prioritize genomic biomarkers for drug discovery and clinical trial targets. For more information, visit www.genomenon.com.

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