Genomenon and Diploid Sign Partnership Agreement to Accelerate Rare Disease Diagnostics

Mastermind Genomic Search Engine Now Integrated Into Diploid's Moon Diagnostic Software

ANN ARBOR, MI and LEUVEN, BELGIUM; February 19, 2019 -- Diploid, leading AI-based software provider for rare disease diagnostics, and Genomenon®, the genomic search company, announced today that they have entered into a partnership to improve genome interpretation for rare disease diagnostics.

With its Moon software, Diploid provides the most efficient workflow for rare disease diagnostics. Moon allows its users to go from VCF to causal variant in just minutes thanks to advanced machine learning and deep phenotype integration. As a result, the software was selected by Dr. Stephen Kingsmore at Rady Children's Hospital to establish a new world record for fastest genome interpretation in the NICU (Read the Rady story).

"Moon returns a very short list of variants that might be relevant for the patient's phenotype: typically about 4-8 variants for exome or whole genome data" says Peter Schols, Diploid's founder and CEO. "This breakthrough saves clinical laboratory scientists a lot of time compared to manually filtering and curating hundreds or even thousands of variants."

Cyrielle Kint, Diploid's CSO, adds: "We wanted to make the process of evaluating the handful of remaining variants reported by Moon even more efficient, and that's why we are integrating with Genomenon's Mastermind®, the clear leader in genomic search engines."

Genomenon's Mastermind Genomic Search Engine filters and prioritizes millions of genomic articles to find the most useful and meaningful citations, saving scientists hours of literature curation per case and improving the chances of finding key genetic information that could be critical for a patient's diagnosis.

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The new integration allows users to see which candidate variants are mentioned in relevant publications and gives users direct access to Mastermind from Moon's user interface.

"We're thrilled to partner with Diploid to reduce the time needed to identify the cause of genetic illness that can be life-threatening," said Mike Klein, CEO of Genomenon. "With integrated Mastermind search results, Moon users get an immediate view of the literature supporting each variant, and can speed their genomic interpretation for rare diseases."

Both companies will be demonstrating the integration at the ACMG Annual Clinical Genomics Meeting conference taking place in Seattle, Washington April 2-6, 2019.

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.

About Diploid
Diploid is on a mission to provide clinical labs, hospitals and nation-wide sequencing programs with the tools to diagnose every rare disease patient. Its Moon platform is the first software worldwide to use artificial intelligence for rare disease diagnostics.

Thanks to its use of advanced machine learning and deep phenotype integration, Moon is now widely recognised as the fastest genome interpretation solution, going from whole genome VCF to diagnosis in minutes.

In addition, Moon's autopilot feature enables unsupervised reanalysis of negative cases, making continuous genome interpretation a reality.
Learn more at www.diploid.com/moon.

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