Genomenon

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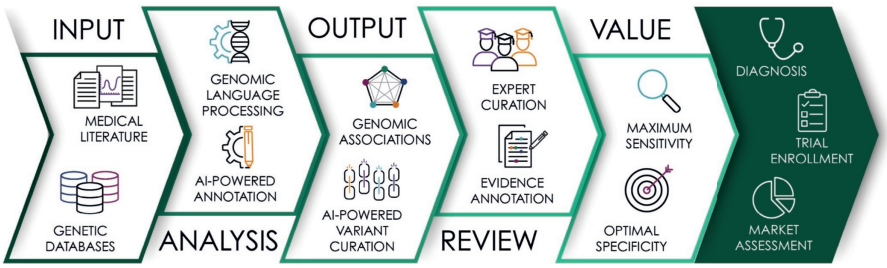
GENOMENON®
AI-DRIVEN GENOMICS

Improving diagnostic rates for rare diseases by streamlining genomic interpretation

Genomics company Genomenon is combining the power of artificial intelligence with the experience of expert curators to produce an accurate assessment of genetic variants that cause disease to expedite development of diagnostics and target identification.

Genetics defines nearly 80% of all rare diseases. Despite readily available genome sequencing, many people with a rare disease experience diagnostic delays, or are never properly diagnosed. This is due in large part to challenges that clinicians face in identifying, assessing, annotating, and documenting genomic evidence to guide patient care. Even when a diagnosis is available, awareness of new trials and drug therapies in the clinical community is limited.

Addressing these challenges is Genomenon, an artificial intelligence (AI)-driven genomics company at the forefront of a new era of precision medicine. The same company that built the Mastermind genomic search engine—the most comprehensive search engine of empirical evidence for diagnosing genetic disease—is curating the entire human genome with the goal of increasing the accurate diagnosis and effective treatment of patients with a rare disease.



Process, value, and applications for Genomenon's genomic data solutions.

A unique combinatorial approach

Genomenon combines the computational power of AI and the critical eye of human curation to produce the most complete and accurate assessment of genetic variants that cause disease.

"Genomenon uniquely merges the scalability and sensitivity of AI with the specificity and accuracy that can only come from experienced human curators," explained Mark Kiel, founder and chief scientific officer at Genomenon. "Resolving genetic ambiguities and critically assessing the meaningfulness of clinical information is paramount. In our approach, every scientific assertion for every variant is reviewed by experts and supported by empirical evidence from the medical literature."

Curating the genome

Using this novel approach, Genomenon is curating every gene in the entire human genome, and is making the insights broadly available. By rapidly producing comprehensive and expertly curated variant landscapes comprising all disease-causing variants for any gene, Genomenon enables pharmaceutical companies to select optimal inclusion and exclusion criteria for clinical trial design, develop companion diagnostics, and accelerate target discovery for precision medicine drug programs. This disease-specific curated content is then made available across thousands of diagnostics labs in more than 140 countries through its Mastermind genomic search engine. This approach not only gives clinicians access to comprehensive and clinically meaningful evidence, but also raises awareness of available clinical trials and therapies.

> **"Genomenon enables pharma companies to select optimal inclusion and exclusion criteria for clinical trial design, develop companion diagnostics, and accelerate target discovery for precision medicine drug programs**

"To fully realize the promise of precision medicine in routine care, every variant in the human genome must be pre-curated," said Kiel. "Only with the speed and scale afforded by AI technology will it be possible to achieve this within the next several years."

Rare disease diagnosis

Genomenon's initial focus is on genes that cause early-onset diseases that are deadly but potentially treatable. One of these diseases is ENPP1 deficiency, a rare and devastating disorder that causes the calcification and narrowing of arteries and is lethal in approximately 50% of babies. Inozyme Pharma has developed a novel therapeutic for which there is a promising clinical trial—but for clinicians to intervene with these therapies they first need to be made aware of the trial.

In an ongoing partnership with Inozyme, Genomenon has produced the world's most comprehensive variant landscape for ENPP1 deficiency that identifies whether certain genetic variations are known to be pathogenic. The research data have been published in Mastermind, along with information about available clinical trials and potential treatment options. "The dream for any rare disease is an earlier diagnosis and effective treatment, but the best drug in the world is useless if patients or physicians don't know about it," explained Catherine Nester, vice president of physician and patient strategies at Inozyme Pharma. "In the rare disease space, knowledge is power, and Genomenon's curated content has real potential for powerful impact."

Value to pharma

Inozyme Pharma is just one company that Genomenon is helping to overcome the challenges of rare disease diagnosis and treatment—and this is only the beginning. Genomenon is collaborating with Stephen Kingsmore and the Rady Children's Institute for Genomic Medicine in San Diego, California, United States, as the supplier of curated variant data on ~400 genetic diseases for the BeginNGS program. The end result of this pioneering effort will be more patients connected to life-saving therapies—faster and with more accuracy.

Genomenon is seeking partnerships with organizations looking to improve genetic diagnosis of rare disease, design effective trials, better define disease prevalence and/or connect patients to trials or approved therapies. "As use of sequencing drives exponential growth in precision medicine, Genomenon's platform is the ideal solution to bring life-changing treatment to patients while enabling pharmaceutical companies to more efficiently discover, develop, and distribute new targeted therapies," said Mike Klein, chief executive officer at Genomenon. "By making the best data more readily available, we are on a mission to ensure that no rare disease patient goes undiagnosed or untreated."

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