

# GENOMENON, INC. RECEIVES THE 2023 TECHNOLOGY INNOVATION LEADERSHIP AWARD

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*Identified as best in class in the global pharma &  
clinical diagnostics genomics industries*

Best Practices Criteria for World-Class Performance

Frost & Sullivan applies a rigorous analytical process to evaluate multiple nominees for each award category before determining the final award recipient. The process involves a detailed evaluation of best practices criteria across two dimensions for each nominated company. Genomenon excels in many of the criteria in the global pharma and clinical diagnostics genomics markets.

AWARD CRITERIA	
Technology Leverage	Business Impact
Commitment to Innovation	Financial Performance
Commitment to Creativity	Customer Acquisition
Stage Gate Efficiency	Operational Efficiency
Commercialization Success	Growth Potential
Application Diversity	Human Capital

Genomenon: Innovative, Outcomes-driven, Committed

Genomenon is a genomic intelligence software company that combines AI-powered Genomic Language Processing (GLP) with expert review by genetic scientists to deliver the most comprehensive knowledgebase of genomic evidence available. Their fit-for-purpose AI and systematic approach to identifying and indexing published genomic evidence gives them a unique advantage over other open access and commercial genomic databases in this space. Clinical diagnostic labs rely on Genomenon’s Mastermind Genomic Search Engine to find critical information needed to diagnose rare genetic diseases and identify the most appropriate patient treatment options. Pharmaceutical companies engaged in the development of rare disease therapeutics partner with Genomenon to gain deep insights into the genetic drivers of disease. The Genomenon scientific team leverages information in the Mastermind knowledgebase to provide the genomic intelligence pharmaceutical companies need to better understand disease mechanisms, more accurately estimate disease prevalence, design clinical trials, and prepare documentation for regulatory approvals.

Over the past 6 years, Genomenon has grown from a University of Michigan startup to a well-established genomics software provider, recognized for its fast turnaround, extensible solutions, and patented Genomic Language Processing (GLP) engineered to address the myriad of issues that complicate genomic evidence searches for gene variants, published patient information, and details about clinical trials, and available treatments.

In 2020, Frost & Sullivan recognized Genomenon for its proven genomics methodology, interpretation

and technology that supports development of therapeutics for personalized medicine and remains impressed with the company's continuing innovation and sustained leadership.

### ***Commitment to Providing Comprehensive Genomic Evidence***

Backed by world-class subject matter experts, Genomenon introduced its ground-breaking Mastermind Genomic Search Engine in 2017. The company has since refined its holistic approach to compiling genomic evidence, continuously building its technology to bridge industry gaps and updating its knowledgebase to stay current with the tsunami of peer-reviewed papers and supplemental materials published each year.

The Mastermind Genomic Search Engine is a comprehensive search and association engine that identifies gene, variant, disease, phenotype, and therapy evidence from published scientific articles.<sup>1</sup> It expedites diagnosis and treatment decisions for genetic testing labs. The solution leverages AI-driven genomics to accelerate variant interpretation, providing immediate insight into the full text of millions of scientific articles.

While trailblazing innovation, Genomenon focuses on continuous development, bringing further breakthroughs to the market. Notably, it has made a decisive step forward since Frost & Sullivan's recognition in 2020. Today, it covers over nine million full-text genomic articles (compared to more than

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**- Pavel Zhebrouski,  
Best Practices Research Analyst**

five million in 2020) and over three million supplemental data sets, with 22.4 million identified variants (versus five million in 2020).<sup>2</sup>

As a result, Genomenon significantly differentiates in the space, as Mastermind Genomic Search Engine has 100 times more genomic evidence and 50 times more variants than other available sources.<sup>3</sup>

The company brings a unique approach to the market, diving into the data in a way nobody else

can do. Traditional search tool design does not account for the many ways a disease, gene, or variant is referenced, making identification difficult and labor-intensive. In contrast, in responding to these challenges, Genomenon built a unique set of heuristic algorithms to pull the needed information out of the literature. The company distinctly differentiates in the space by combining AI engines and expert human curation to provide the most accurate interpretation of disease-causing variants. As a result, it reduces the turnaround time and, more importantly, the risk of missing a patient diagnosis.

Today, tens of thousands of doctors in over 140 countries leverage the Mastermind Genomic Search Engine, connecting their patient's variants with the published genomic research to diagnose and treat patients promptly, and diagnose babies with rare diseases more effectively.

Recently, Genomenon integrated patient variant data from the National Institutes of Health's (NIH)

<sup>1</sup> <https://www.genomenon.com/mastermind-faq/>, accessed November 2022

<sup>2</sup> Frost & Sullivan Interview with Genomenon, (October 2022)

<sup>3</sup> <https://www.genomenon.com/mastermind/>, accessed November 2022



ClinVar database into its Mastermind Genomic Search Engine, ensuring it brings the most comprehensive view to its customers.

### ***AI-Driven Genomic Solutions for Rare Disease Drug Development***

In addition to supporting clinical diagnostics applications, Genomenon's expert genomic science team partners with pharma and biotech companies developing orphan drugs to deliver comprehensive and customized reports on genomic landscapes for rare genetic disease. Genomenon's Pharma solutions harness the power of the Mastermind knowledgebase to identify genomic biomarkers, pathogenic variants, clinical trial target selection, and other data needed to support go-no-go decision-making in translational and clinical stage drug development and delivery. These solutions provide a comprehensive understanding of the genomic drivers of any disease and their mechanism of action to inform the genomic strategy of the top global pharmaceutical companies.

Fueled by Mastermind's comprehensive knowledgebase, the company's solutions are customized to meet its pharma customers' demands for actionable data. It specializes in providing the genomic intelligence required to better understand disease mechanisms, reveal the patient universe, validate disease prevalence, and increase diagnostic yield. New curated information uncovered during these highly collaborative projects are added to the Mastermind knowledgebase to support the greater research community and genetic testing labs.

The company can prepare a comprehensive "variant landscape" for any genetic disease of interest that identifies the genetic profile of the patients suitable for clinical trials or therapeutics that target specific gene mutations. Genomenon's genome scientists review and interpret relevant genes following clinical guidelines to deliver a pathogenicity score for each gene variant. In addition, Genomenon documents the underlying mechanism of action to ensure its pharma partners can identify the most appropriate patient profiles when designing clinical cohorts to increase the probability of clinical trial success.

This comprehensive picture of the multi-faceted genomic landscape of rare disease genetics arms pharma companies with the data and insights needed to determine market potential and validate evidence-based estimates of disease prevalence. It gives the company's pharma partners the variant and population data required to calculate disease prevalence more accurately in a population. Genomenon further drives disease, clinical trial, and treatment awareness through its global network of clinical diagnostic labs. Through this clinical connection, it makes genomic data uncovered through pharma partnership available, increasing diagnostic yield and clinical awareness.

Frost & Sullivan identifies Genomenon's solutions as ground-breaking, innovative technology. Its strong intellectual property portfolio safeguards its pioneering technology, adding value relative to its growth potential, thus securing a competitive advantage.

### ***A Proven Track Record***

Genomenon has a proven track record. The company efficiently supports over 20,000 users (pathologists, geneticists, and scientists) in 140 countries with over 2,000 laboratories using Mastermind for genetic

diagnosis.<sup>4</sup> Recently, the company completed a \$20 million Series B financing round led by Beringea and Spring Mountain Capital to drive further growth in the pharmaceutical and clinical diagnostic markets.<sup>5</sup>

Genomenon continues its solid growth stemming from its branding strategy and product delivery, which makes it a partner of choice for industry leaders. Notably, the company partners with Rady Children's Institute for Genomic Medicine (RCIGM) on rapid whole-genome sequencing to diagnose and treat critically ill babies with genetic diseases. Using AI-driven genomic data, Genomenon curates all genes included in RCIGM's recently announced BeginNGS™ newborn screening panel.<sup>6</sup>

The company has partnered with innovative pharmaceutical and biotechnology companies, such as Inozyme Pharma, to make critical information on ENPP1 deficiency available to genetic testing labs and clinicians across the globe.

***"The ENPP1 genomic landscape provided by Genomenon has enriched our understanding of this genetic disorder and continues to inform our research and clinical trial design."***<sup>7</sup>

- Catherine Nester, VP of Physician and Patient Strategies, Inozyme Pharma

Leveraging its Mastermind Genomic Search Engine, Genomenon produced a comprehensive variant landscape for ENPP1 deficiency, enabling opportunities for better diagnosis and treatment decisions against this rare disease.<sup>8,9</sup> Genomenon and Inozyme then extended this study to recalculate ENPP1 deficiency disease prevalence to reveal it was more than triple the previous estimate.

Frost & Sullivan anticipates rapid, widespread technology adoption, with the company's first-mover status strengthening its position on emerging opportunities.

### ***Innovative Technology Provides Sustainable Leadership***

With its customer-focused strategy, Genomenon consistently brings to market innovative best-in-class solutions and hastens new applications. Its mission is to make genomic information actionable. In fulfilling this goal, the company aims to curate the human genome and document all disease-gene-variant-phenotype-and therapy relationships across the medical literature. This extraordinary genomic intelligence resource will be invaluable for supporting the rapidly evolving field of precision medicine, increasing diagnosis speed, and enabling clinicians to make quick therapeutic decisions. Genomenon's innovative approach supports the next-generation sequencing program for newborns and expands the company's Mastermind Genomic Search Engine. With disease-specific curated content, it guides

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<sup>4</sup> Frost & Sullivan Interview with Genomenon, (October 2022)

<sup>5</sup> Genomenon Raises \$20M Series B Financing, Launching a New Era in AI-Powered Genomic Data, <https://www.genomenon.com/genomenon-raises-20-million-series-b-financing-launching-new-era-ai-powered-data/>, accessed November 2022

<sup>6</sup> Genomenon Partners with Rady Children's Institute for Genomic Medicine on Newborn Screening by Next Generation Sequencing, <https://www.genomenon.com/genomenon-partners-with-rady-childrens-institute-genomic-medicine-newborn-screening-next-generation-sequencing/>, accessed November 2022

<sup>7</sup> New Study Shows Comprehensive Genomic Evidence is Critical for Calculating Rare Disease Prevalence <https://www.genomenon.com/new-study-shows-comprehensive-genomic-evidence-critical-for-calculating-rare-disease-prevalence/> accessed February 2023

<sup>8</sup> Genomenon and Inozyme Pharma Announce Partnership to Advance Genetic Diagnosis for Rare Disease Patients, <https://www.genomenon.com/genomenon-inozyme-pharma-announce-partnership-advance-genetic-diagnosis-rare-disease-patients/>, accessed November 2022

<sup>9</sup> Chunn et al (2022), Orphanet Journal of Rare Diseases.

customers to the literature, enabling them to find answers on every variant classified by clinical guidelines while backed by evidence for diagnosis.

At the same time, Genomenon incorporates customer feedback into its product roadmap to maximize short-term growth opportunities while providing a path to future revenues. The company remains informed and close to customer experience and needs, collecting feedback and sharing best practices,

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which drives industry-wide innovations for broader customer success. It provides significant customer value as customers who have benefited from its technology are loyal and consistently return to initiate new programs, licensing more datasets from Genomenon to further develop their work in clinical testing.

While evolving from a technology standpoint, the company never loses sight of its customers' perspectives. Its brand maintains a global presence

while meeting customer-specific needs. Given today's landscape, Frost & Sullivan believes Genomenon is in a prime position to increase its market share in this highly competitive advanced genomics industry.

## Conclusion

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Technology innovation is a critical success factor for the genomics software market. Yet, with many options available, market stakeholders need to leverage the most appropriate and best technology-based solutions to optimize their market impact. With its unique Genomic Language Processing, extensive knowledgebase of genomic evidence, and expert genomic science team, Genomenon powers rare disease pharmaceutical and biotechnology companies to make informed decisions about promising candidates in their pipeline and help propel novel targeted therapeutics to market to improve the quality of life of patients. The company stands out from competitors based on its commitment to innovation and creativity, scientific rigor and accuracy, while achieving commercial success. Leveraging its deep expertise, Genomenon is committed to curating the human genome to make it actionable to support development and delivery of precision medicines, increase diagnosis speed, and enable clinicians to make timely therapeutic decisions.

For its strong overall performance, Genomenon is recognized with Frost & Sullivan's 2023 Global Technology Innovation Leadership Award in the global pharma and clinical diagnostics genomics markets.

## What You Need to Know about the Technology Innovation Leadership Recognition

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Frost & Sullivan's Technology Innovation Leadership Award recognizes the company that has introduced the best underlying technology for achieving remarkable product and customer success while driving future business value.

### Best Practices Award Analysis

For the Technology Innovation Leadership Award, Frost & Sullivan analysts independently evaluated the criteria listed below.

#### *Technology Leverage*

**Commitment to Innovation:** Continuous emerging technology adoption and creation enables new product development and enhances product performance

**Commitment to Creativity:** Company leverages technology advancements to push the limits of form and function in the pursuit of white space innovation

**Stage Gate Efficiency:** Technology adoption enhances the stage gate process for launching new products and solutions

**Commercialization Success:** Company displays a proven track record of taking new technologies to market with a high success rate

**Application Diversity:** Company develops and/or integrates technology that serves multiple applications and multiple environments

#### *Business Impact*

**Financial Performance:** Strong overall financial performance is achieved in terms of revenues, revenue growth, operating margin, and other key financial metrics

**Customer Acquisition:** Customer-facing processes support efficient and consistent new customer acquisition while enhancing customer retention

**Operational Efficiency:** Company staff performs assigned tasks productively, quickly, and to a high-quality standard

**Growth Potential:** Growth is fostered by a strong customer focus that strengthens the brand and reinforces customer loyalty

**Human Capital:** Commitment to quality and to customers characterize the company culture, which in turn enhances employee morale and retention

## About Frost & Sullivan

Frost & Sullivan is the Growth Pipeline Company™. We power our clients to a future shaped by growth. Our Growth Pipeline as a Service™ provides the CEO and the CEO's growth team with a continuous and rigorous platform of growth opportunities, ensuring long-term success. To achieve positive outcomes, our team leverages over 60 years of experience, coaching organizations of all types and sizes across 6 continents with our proven best practices. To power your Growth Pipeline future, visit Frost & Sullivan at <http://www.frost.com>.

## The Growth Pipeline Engine™

Frost & Sullivan's proprietary model to systematically create ongoing growth opportunities and strategies for our clients is fuelled by the Innovation Generator™.

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### Key Impacts:

- **Growth Pipeline:** Continuous Flow of Growth Opportunities
- **Growth Strategies:** Proven Best Practices
- **Innovation Culture:** Optimized Customer Experience
- **ROI & Margin:** Implementation Excellence
- **Transformational Growth:** Industry Leadership



## The Innovation Generator™

Our 6 analytical perspectives are crucial in capturing the broadest range of innovative growth opportunities, most of which occur at the points of these perspectives.

### Analytical Perspectives:

- **Mega Trend (MT)**
- **Business Model (BM)**
- **Technology (TE)**
- **Industries (IN)**
- **Customer (CU)**
- **Geographies (GE)**

