

Global Artificial Intelligence (AI)-Powered Rare Disease Diagnosis Market to Reach \$5.49 Bn at 28.9% CAGR by 2029

*The Business Research Company's
Artificial Intelligence (AI)-Powered Rare
Disease Diagnosis Global Market Report
2025 – Market Size And Forecast 2025-
2034*

LONDON, GREATER LONDON, UNITED
KINGDOM, October 9, 2025

/EINPresswire.com/ -- How Large Will

The [Artificial Intelligence \(AI\)-Powered Rare Disease Diagnosis Market](#) Be By 2025?

The market size for the AI-enhanced rare disease diagnosis industry has seen considerable growth in the past years. The projection is from \$1.54 billion in 2024 to jump to \$1.99 billion in 2025, with a CAGR of 29.1%. The notable increase during the historic period can be traced back



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to the growing use of AI in the healthcare sector, increased investments in research for rare diseases, a surge in awareness regarding early diagnosis, the development of healthcare infrastructure, and rising government backing for digital health.

Over the next few years, the market size of AI-based rare disease diagnosis is likely to experience significant expansion, growing to \$5.50 billion in 2029 with a CAGR of 28.9%. Various factors are set to contribute to this growth including the elevated utilization of machine learning

algorithms, heightened demand for tailor-made medicine, enhanced partnerships between AI start-ups and hospitals, the increase in accessible genomic data, and broader digitalization within the healthcare sector. Future trends to watch out for include progression in deep learning models, novel approaches in predictive diagnostics, advancements in the utilization of natural language processing for medical data, study and growth in the field of multi-omics integration, along with continuous refinements in AI-oriented imaging technologies.

Download a free sample of the artificial intelligence (ai)-powered rare disease diagnosis market

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What Are The Major Driving Forces Influencing The Artificial Intelligence (AI)-Powered Rare Disease Diagnosis Market Landscape?

The ongoing rise in autoimmune disorders is anticipated to stimulate the expansion of the artificial intelligence (AI) enhanced rare disease diagnosis market. Autoimmune disorders are characterized by the immune system erroneously targeting the body's own cells and tissues, causing swelling and harm. This escalation in autoimmune disorders is attributed to an increasing number of people suffering from immune system irregularities that fuel a greater requirement for early detection, superior treatments, and ongoing tracking to effectively manage symptoms. AI-enhanced rare disease diagnosis aids in managing autoimmune disorders by swiftly pinpointing intricate conditions, facilitating customized treatment choices, and refining the overall precision of diagnosis. For example, based on data from the Australian Institute of Health and Welfare, a governmental entity in Australia, in 2022, approximately 514,000 individuals (2.0%) in Australia were coping with rheumatoid arthritis, a systemic autoimmune disorder where the immune system targets its own tissues. Hence, the rising occurrence of autoimmune disorders is pushing the growth of the AI-powered rare disease diagnosis market.

Who Are The Top Players In The Artificial Intelligence (AI)-Powered Rare Disease Diagnosis Market?

Major players in the Artificial Intelligence (AI)-Powered Rare Disease Diagnosis Global Market Report 2025 include:

- Ambry Genetics Corporation
- MedGenome Inc.
- Sophia Genetics SA
- Variantyx Inc.
- Facial Dysmorphology Novel Analysis
- Atropos Health Inc.
- Congenica Ltd.
- Saventic Care Sp. z o.o.
- Rapid Innovation LLC
- Centogene N.V.

What Are The Key Trends Shaping The [Artificial Intelligence \(AI\)-Powered Rare Disease Diagnosis Industry](#)?

Prominent players in the sphere of artificial intelligence (AI) applied to the diagnosis of rare diseases are turning their attention towards developing state-of-the-art technology such as consolidated genomic diagnostic tools. These innovative solutions are designed to augment diagnostic precision, trim down processing times, and offer a more thorough understanding of intricate genetic conditions. The said solutions are composed of holistic genomic sequencing and AI-guided analysis, providing complete insights on genetic data. One such instance occurred in October 2023, with Oxford Nanopore Technologies plc, a biotechnology company from the UK,

and Fabric Genomics Inc., an American company specialising in AI, unveiling an integrated whole-genome sequencing solution aimed at hastening the diagnosis of rare diseases. This solution merges rapid sequencing with AI-guided variant analysis to yield more precise and useful clinical insights. Integrating real-time nanopore sequencing with AI driven genomic analysis swiftly identifies rare genetic variants. This results in robust, actionable insights for clinicians, enhancing the speed and precision of diagnostics especially in pediatric care.

Market Share And Forecast By Segment In The Global Artificial Intelligence (AI)-Powered Rare Disease Diagnosis Market

The artificial intelligence (AI)-powered rare disease diagnosis market covered in this report is segmented as

- 1) By Component: Software, Hardware, Services
- 2) By Technology: Machine Learning, Deep Learning, Natural Language Processing, Computer Vision, Other Technologies
- 3) By Deployment Mode: Cloud-Based, On-Premises
- 4) By Diagnosis Type: Genetic Disorders, Autoimmune Disorders, Metabolic Disorders, Infectious Diseases, Other Diagnosis Types
- 5) By End-User: Hospitals, Diagnostic Centers, Research Institutes, Specialty Clinics, Other End-Users

Subsegments:

- 1) By Software: Clinical Decision Support, Data Analytics Platforms, Genomic Interpretation Tools, Machine Learning Algorithms, Diagnostic Reporting Software
- 2) By Hardware: Sequencing Instruments, Imaging Devices, Laboratory Equipment, Computational Servers, Diagnostic Sensors
- 3) By Services: Consulting Services, Implementation And Integration, Training And Support, Data Management Services, Maintenance And Upgrades

View the full artificial intelligence (ai)-powered rare disease diagnosis market report:

<https://www.thebusinessresearchcompany.com/report/artificial-intelligence-ai-powered-rare-disease-diagnosis-global-market-report>

Artificial Intelligence (AI)-Powered Rare Disease Diagnosis Market Regional Insights

In the 2025 Global Market Report for AI-Based Rare Disease Diagnosis, North America was named as the leading region. It's also projected that the Asia-Pacific region will experience the quickest expansion within the forecasted timeframe. The report encompasses a range of regions, specifically Asia-Pacific, Western Europe, Eastern Europe, North America, South America, Middle East, and Africa.

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