

Rare Disease Genetic Testing Market CAGR to be at 13.3% from 2025 to 2029 | \$1.93 Billion Industry Revenue by 2029

The Business Research Company's Rare Disease Genetic Testing Global Market Report 2025 – Market Size, Trends, And Global Forecast 2025-2034

LONDON, GREATER LONDON, UNITED KINGDOM, September 15, 2025

[/EINPresswire.com/ -- What Is The Projected Market Size & Growth Rate Of The Rare Disease Genetic Testing Market?](#)

The market size for genetic testing for rare diseases has experienced substantial growth in the past few years. The market is predicted to expand from \$1.03 billion in 2024 to \$1.17 billion in 2025, reflecting a compound annual growth rate (CAGR) of 13.6%. The previous growth can be



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credited to the increasing awareness of rare genetic conditions, a rise in the demand for quick and precise diagnoses, an increase in the government's funding for research into rare diseases, the ever-growing availability of genetic testing services, an upward trend in investments in precision medicine, and the increasing occurrence of rare genetic disorders.

The market size for testing for rare genetic diseases is projected to dramatically increase in the coming years, reaching \$1.93 billion in 2029, growing at a compound annual growth rate (CAGR) of 13.3%. This growth in the

predicted time frame can be credited to factors such as the rising utilization of next-generation sequencing techniques, a heightened emphasis on personalized medicine, increased investment in genetic research, the expansion of healthcare systems in developing markets, an upswing in diagnostic testing, a surge in hereditary cancer testing, and an escalating demand for non-invasive diagnostic solutions. Key trends expected during the forecast period are the progression in genome editing technologies, sophisticated bioinformatics tools for analyzing genetic data, innovations in multi-gene panel testing, technological advancements in portable genetic testing



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devices, and new methods in sample collection techniques.

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What Is The Crucial Factor Driving The Global Rare Disease Genetic Testing Market?

The surge in genetic disorders is anticipated to stimulate the expansion of the rare disease genetic testing market. Genetic disorders are medical conditions prompted by abnormalities in a person's deoxyribonucleic acid, bequeathed by either or both parents, or from new anomalies. The rise in genetic disorders can be attributed to advancing parental age since older parents have an elevated risk of transmitting genetic mutations to their offspring. Rare disease genetic testing aids in accurately diagnosing these disorders at an early stage by detecting specific mutations. It facilitates customized treatment plans, thereby enhancing patient care and promoting better long-term health results. For instance, the National Health Service, a UK-based government department, reported in May 2024 that 17,000 individuals are currently living with sickle cell disease, a hereditary genetic blood disorder, with approximately 250 new cases being identified annually. Consequently, the rise in genetic disorders is propelling the expansion of the rare disease genetic testing market.

Who Are The Emerging Players In The Rare Disease Genetic Testing Market?

Major players in the Rare Disease Genetic Testing Global Market Report 2025 include:

- Thermo Fisher Scientific Inc.
- Quest Diagnostics Inc.
- Eurofins Scientific SE
- Agilent Technologies Inc.
- Illumina Inc.
- QIAGEN N.V.
- Natera Inc.
- OPKO Health Inc.
- Myriad Genetics Inc.
- Fulgent Genetics Inc.

What Are The Key Trends Shaping The Rare Disease Genetic Testing Industry?

Prominent businesses in the rare disease genetic testing market are keen on innovating technologically superior solutions like digital genomics platforms. These platforms aim to enrich the efficacy, precision, and adaptability of diagnosing rare diseases. Such platforms are superior software systems that amalgamate genomic data interpretation, analysis, and reporting to expedite and streamline genetic testing and diagnosis. As an illustration, Strand Life Sciences Private Limited, an Indian computer software enterprise, initiated the StrandOmics Portal in March 2025. This digital platform is targeted to increase the precision and efficiency of rare disease diagnosis. The portal grants detailed insights into genetic variations to clinicians, which aids in faster and more accurate diagnoses. It also supports advanced variant interpretation,

encompassing prenatal diagnostics, and allows independent genetic data reviews. This introduction showcases a firm commitment to promoting precision medicine and equipping healthcare professionals with efficient tools to improve patient outcomes.

What Segments Are Covered In The Rare Disease Genetic Testing Market Report?

The rare disease genetic testing market covered in this report is segmented

- 1) By Disease Type: Immunological Disorders, Cardiovascular Disorders, Neurological Disease, Endocrine And Metabolism Disorders Diseases, Hematological Diseases, Dermatological Disease, Other Disease Types
- 2) By Technology: Next-Generation Sequencing, Sanger Sequencing, Polymerase Chain Reaction Based Testing, Array Based Testing, Fluorescence In Situ Hybridization, Other Technologies
- 3) By Specialty: Molecular Genetic Tests, Chromosomal Genetic Tests, Biochemical Genetic Tests
- 4) By End-User: Hospitals And Clinics, Research Laboratories And Contract Research Organizations, Diagnostic Laboratories, Other End Users

Subsegments:

- 1) By Immunological Disorders: Autoimmune Disorders, Allergic Conditions, Primary Immunodeficiency Disorders, Inflammatory Disorders
- 2) By Cardiovascular Disorders: Congenital Heart Defects, Cardiomyopathies, Aortic Aneurysms, Pulmonary Hypertension
- 3) By Neurological Disease: Neurodegenerative Disorders, Neuromuscular Disorders, Epileptic Syndromes, Movement Disorders
- 4) By Endocrine And Metabolism Disorders Diseases: Inherited Metabolic Disorders, Thyroid Disorders, Adrenal Disorders, Diabetes Related Genetic Conditions
- 5) By Hematological Diseases: Hemophilia, Thalassemia, Sickle Cell Disease, Clotting Disorders
- 6) By Dermatological Disease: Genetic Skin Disorders, Connective Tissue Disorders, Pigmentary Disorders, Keratinization Disorders
- 7) By Other Disease Types: Skeletal Dysplasia, Renal Genetic Disorders, Ophthalmic Genetic Conditions, Gastrointestinal Genetic Conditions

View the full rare disease genetic testing market report:

<https://www.thebusinessresearchcompany.com/report/rare-disease-genetic-testing-global-market-report>

Which Region Is Projected To Hold The Largest Market Share In The Global Rare Disease Genetic Testing Market?

In the Rare Disease Genetic Testing Global Market Report 2025, North America emerged as the leading region in 2024, with the Asia-Pacific region predicted to experience the most rapid growth in the forecast period. The report provides coverage for several regions including Asia-Pacific, Western Europe, Eastern Europe, North America, South America, the Middle East, and Africa.

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