

Rare Disease Genetic Testing Market - Global Industry Size, Share, Trends, Opportunity, and Forecast, 2018-2028 Segmented By Disease Type (Neurological Disease, Immunological Disorders, Hematology Diseases, Endocrine & Metabolism Diseases, Cancer, Musculoskeletal Disorders, Cardiovascular Disorders (CVDs), Dermatology Disease, Others), By Speciality (Molecular Genetic Tests, Chromosomal Genetic Tests, Biochemical Genetic Tests) By Technology (Next-Generation Sequencing (NGS), Whole Exome Sequencing, Whole Genome Sequencing, Array Technology, PCR-based Testing, FISH, Sanger Sequencing, Karyotyping), By End User (Research Laboratories & CROs, Hospitals & Clinics, Diagnostic Laboratories), By Region, Competition

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Abstracts

The Global Rare Disease Genetic Testing Market was valued at USD 789.83 Million in 2022 and is expected to experience robust growth during the forecast period, with a projected Compound Annual Growth Rate (CAGR) of 8.04% and expected to reach USD 1249.63 Million through 2028. Rare Disease Genetic Testing involves the diagnosis of genetic mutations or alterations responsible for rare diseases. These diseases have a low prevalence, affecting only a small percentage of the population. They are often caused by genetic mutations or alterations in specific genes. Genetic

testing for rare diseases analyzes an individual's DNA to identify abnormalities or mutations that may be causing their symptoms or condition. This aids in accurate diagnosis, understanding the underlying cause of the disease, and guiding personalized treatment strategies. Various techniques are used for rare disease genetic testing, including next-generation sequencing (NGS), which allows the analysis of multiple genes or the entire genome simultaneously. This technology enhances the efficiency and accuracy of genetic testing, enabling the detection of rare genetic mutations.

Key Market Drivers

Increasing Prevalence of Rare Diseases Drives Market Growth: The rising prevalence of rare diseases is a significant driver for the global rare disease genetic testing market. The prevalence rate of rare diseases in the population was estimated at 3.5-5.9%, translating to approximately 260-440 million people globally. As understanding of genetics and rare diseases improves, genetic testing becomes crucial for accurate and early diagnosis. It enables the identification of specific genetic mutations associated with rare diseases, confirming diagnoses, and enabling targeted treatment plans. Increased awareness of rare diseases among healthcare providers and patients has led to higher demand for accurate and timely diagnosis. Patients and their families are more likely to seek genetic testing for suspected rare diseases, contributing to market growth. The concept of precision medicine, which tailors medical care to an individual's genetic makeup, further drives the demand for genetic testing.

Increasing Demand for Personalized Medication Drives Market Growth: The growing demand for personalized medicine significantly influences the global rare disease genetic testing market. Personalized medicine involves tailoring medical treatment to individual patient characteristics, including genetic makeup. It anticipates which treatments will be most effective based on genetic information. Genetic testing is crucial for identifying genetic mutations underlying rare diseases, confirming diagnoses, and understanding their causes. Personalized medicine aims to target disease root causes, making genetic testing essential for treatment tailoring. Genetic testing enables precise and early rare disease diagnosis, especially important for conditions with vague or overlapping symptoms. Accurate diagnosis ensures appropriate and effective treatment options. Genetic testing also identifies potential drug targets, predicts treatment responses, and guides therapy selection, particularly important for rare diseases with limited standard treatments.

Increasing Awareness and Advocacy Drive Market Growth: Rising awareness and advocacy for rare diseases contribute to the growth of the global rare disease genetic

testing market. Approximately 7,000 rare diseases affect 25-30 million Americans, leading to increased awareness. Advocacy efforts raise the profile of genetic testing in managing rare diseases, driving demand for testing services. Increased awareness and advocacy lead to greater genetic testing consideration among patients, healthcare providers, and policymakers. As awareness spreads about rare diseases and the importance of genetic testing, patients seek genetic testing for accurate diagnosis and personalized treatment. Advocacy initiatives emphasize early diagnosis significance for improved patient outcomes, leading to higher demand for testing services. Collaborations between advocacy groups, healthcare providers, and researchers improve genetic testing access and funding, contributing to market growth.

Key Market Challenges

Diagnostic Complexity Poses Testing Challenges: Rare diseases' complexity can lead to delayed or misdiagnosis. Healthcare providers may struggle to identify underlying causes of symptoms, leading to incorrect treatments or interventions. Rare diseases have heterogeneous symptoms and often lack standardized diagnostic criteria, making genetic testing challenging. Rare diseases' genetic basis can involve multiple genes and variants, complicating precise mutation identification. Advanced testing technologies and bioinformatics analysis are required to identify rare disease genetic mutations accurately.

Limited Knowledge and Awareness Hinders Testing Adoption: Limited awareness among healthcare providers hinders genetic testing consideration for rare diseases. Lack of familiarity with rare disease clinical presentations and genetic profiles leads to underutilization of genetic testing. Misdiagnosis due to limited knowledge results in inappropriate treatments. Limited awareness affects genetic testing selection, as providers may not recognize genetic markers associated with specific rare diseases. Limited awareness also slows rare disease research progress and test development.

Ethical and Privacy Concerns Impact Testing Uptake: Ethical and privacy concerns affect rare disease genetic testing adoption. Obtaining informed consent for genetic testing is complex, especially for rare diseases. Privacy and security concerns around genetic data discourage testing participation. Genetic testing may reveal unexpected findings, creating ethical dilemmas. Genetic data may also affect family members who have not consented to testing. Ethical considerations are influenced by cultural and religious beliefs, requiring careful testing management.

Key Market Trends

Growth of Direct-to-Consumer (DTC) Testing: While DTC testing's growth in the global rare disease genetic testing market is relatively limited compared to other genetic testing areas, such as ancestry or health traits, it plays a role. DTC tests empower consumers with genetic information, potentially identifying markers associated with rare diseases. Even if not exclusively for rare diseases, DTC testing increases general genetics and health awareness. DTC tests can identify rare genetic variants associated with conditions. Some companies offer options to contribute genetic data to research databases, aiding rare disease research and discoveries. Ethical concerns and regulatory oversight impact DTC testing's accuracy and implications.

Segmental Insights

Disease Type Insights: The Endocrine & Metabolism segment dominated the 2022 Rare Disease Genetic Testing market and is projected to continue growing. Many rare diseases in this category have clear genetic bases, with mutations affecting hormone production and metabolism. Genetic testing aids early diagnosis and intervention. Genetic information guides personalized treatment plans, targeting specific genetic mutations and improving outcomes.

Technology Insights: Next-Generation Sequencing (NGS) dominated the 2022 Rare Disease Genetic Testing market and will likely continue growing. NGS revolutionizes genetic testing, offering speed, affordability, accuracy, and the analysis of large genetic datasets. It aids the identification of rare genetic mutations and enhances diagnosis and treatment precision.

Specialty Insight: Molecular genetic tests dominated the 2022 Rare Disease Genetic Testing market and are predicted to continue growing. Molecular tests analyze DNA, RNA, or proteins, diagnosing rare diseases, monitoring progression, and identifying risk.

End-Use Industry Insights: Research & Contract Research Organizations (CROs) dominated the 2022 Rare Disease Genetic Testing market and will likely continue growing. They develop genetic tests, conduct trials, and offer testing services. Research & CROs study genetic bases of rare diseases, develop treatments, and improve diagnosis and management.

Regional Insights

North America leads the Global Rare Disease Genetic Testing Market, primarily the

United States and Canada. These countries possess advanced healthcare infrastructure supporting genetic testing technology development, adoption, and integration. The region's high rare disease prevalence results from factors like an aging population and increased awareness. This region's prominent healthcare landscape supports genetic testing growth and adoption.

Key Market Players

Centogene N.V.

Invitae Corp.

3billion, Inc.

Arup Laboratories

Eurofins Scientific

Strand Life Sciences

Ambry Genetics

Perkin Elmer, Inc.

Realm IDX, Inc.

Macrogen, Inc.

Baylor Genetics

Report Scope:

In this report, the Global Rare Disease Genetic Testing Market has been segmented into the following categories, in addition to the industry trends which have also been detailed below:

Global Rare Disease Genetic Testing Market, By Disease Type:

Neurological Disease

Immunological Disorders

Hematology Diseases

Endocrine & Metabolism Diseases

Cancer

Musculoskeletal Disorders

Cardiovascular Disorders (CVDs)

Dermatology Disease

Others

Global Rare Disease Genetic Testing Market, By Speciality:

Molecular Genetic Tests

Chromosomal Genetic Tests

Biochemical Genetic Tests

Global Rare Disease Genetic Testing Market, By Technology:

Next-Generation Sequencing (NGS)

Whole Exome Sequencing

Whole Genome Sequencing

Array Technology

PCR-based Testing

FISH

Sanger Sequencing

Karyotyping

Global Rare Disease Genetic Testing Market, By End User:

Research Laboratories & CROs

Hospitals & Clinics

Diagnostic Laboratories

Global Rare Disease Genetic Testing Market, By region:

North America

United States

Canada

Mexico

Asia-Pacific

China

India

South Korea

Australia

Japan

Europe

Germany

France

United Kingdom

Spain

Italy

South America

Brazil

Argentina

Colombia

Middle East & Africa

South Africa

Saudi Arabia

UAE

Competitive Landscape

Company Profiles: Detailed analysis of the major companies present in the Global Rare Disease Genetic Testing Market.

Available Customizations:

Global Rare Disease Genetic Testings Market report with the given market data, Tech Sci Research offers customizations according to a company's specific needs. The following customization options are available for the report:

Company Information

Detailed analysis and profiling of additional market players (up to five).

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