

# **Rare Disease Genetic Testing Market Size, Trends, Analysis, and Outlook By Disease (Neurological Disorders, Immunological Disorders, Hematology Diseases, Endocrine & Metabolism Diseases, Cancer, Musculoskeletal Disorders, Cardiovascular Disorders, Dermatology Disease, Others), By Technology (Next Generation Sequencing, Array Technology, PCR-based Testing, FISH, Sanger Sequencing, Karyotyping), By Specialty (Molecular Genetic Tests, Chromosomal Genetic Tests, Biochemical Genetic Tests), By End-user (Research Laboratories & CROs, Hospitals & Clinics, Diagnostic Laboratories), by Region, Country, Segment, and Companies, 2024-2030**

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## **Abstracts**

The global Rare Disease Genetic Testing market size is poised to register 14.52% growth from 2024 to 2030, presenting significant growth prospects for companies operating in the industry. The industry study analyzes the global Rare Disease Genetic Testing market across By Disease (Neurological Disorders, Immunological Disorders, Hematology Diseases, Endocrine & Metabolism Diseases, Cancer, Musculoskeletal Disorders, Cardiovascular Disorders, Dermatology Disease, Others), By Technology (Next Generation Sequencing, Array Technology, PCR-based Testing, FISH, Sanger Sequencing, Karyotyping), By Specialty (Molecular Genetic Tests, Chromosomal Genetic Tests, Biochemical Genetic Tests), By End-user (Research Laboratories & CROs, Hospitals & Clinics, Diagnostic Laboratories).

The Rare Disease Genetic Testing Market is experiencing growth propelled by increasing recognition of rare genetic disorders, rising demand for early and accurate diagnosis, and advancements in genetic testing technologies and bioinformatics analysis. Rare disease genetic testing encompasses a variety of molecular and genomic assays aimed at identifying pathogenic variants and mutations associated with rare inherited diseases. Key trends shaping its future include the development of next-generation sequencing (NGS) panels and exome sequencing assays for comprehensive and high-throughput genetic analysis, integration of bioinformatics algorithms and databases for variant interpretation and classification, and customization of genetic testing strategies for specific disease phenotypes and patient populations. Moreover, increasing adoption of newborn screening and population-based genetic screening programs, expansion of genetic counseling services and rare disease registries, and regulatory approvals for new genetic testing platforms contribute to market expansion.

## Rare Disease Genetic Testing Market Drivers, Trends, Opportunities, and Growth Opportunities

This comprehensive study discusses the latest trends and the most pressing challenges for industry players and investors. The Rare Disease Genetic Testing market research analyses the global market trends, key drivers, challenges, and opportunities in the industry. In addition, the latest Future of Rare Disease Genetic Testing survey report provides the market size outlook across types, applications, and other segments across the world and regions. It provides data-driven insights and actionable recommendations for companies in the Rare Disease Genetic Testing industry.

## Key market trends defining the global Rare Disease Genetic Testing demand in 2024 and Beyond

The industry continues to remain an attractive hub for opportunities for both domestic and global vendors. As the market evolves, factors such as emerging market dynamics, demand from end-user sectors, a growing patient base, changes in consumption patterns, and widening distribution channels continue to play a major role.

## Rare Disease Genetic Testing Market Segmentation- Industry Share, Market Size, and Outlook to 2030

The Rare Disease Genetic Testing industry comprises a wide range of segments and sub-segments. The rising demand for these product types and applications is

supporting companies to increase their investment levels across niche segments. Accordingly, leading companies plan to generate a large share of their future revenue growth from expansion into these niche segments. The report presents the market size outlook across segments to support Rare Disease Genetic Testing companies scaling up production in these sub-segments with a focus on expanding into emerging countries.

Key strategies adopted by companies within the Rare Disease Genetic Testing industry

Leading Rare Disease Genetic Testing companies are boosting investments to capitalize on untapped potential and future possibilities across niche market segments and surging demand conditions in key regions. Further, companies are leveraging advanced technologies to unlock opportunities and achieve operational excellence. The report provides key strategies opted for by the top 10 Rare Disease Genetic Testing companies.

Rare Disease Genetic Testing Market Study- Strategic Analysis Review

The Rare Disease Genetic Testing market research report dives deep into the qualitative factors shaping the market, empowering you to make informed decisions-

**Industry Dynamics:** Porter's Five Forces analysis to understand bargaining power, competitive rivalry, and threats that impact long-term strategy formulation.

**Strategic Insights:** Provides valuable perspectives on key players and their approaches based on comprehensive strategy analysis.

**Internal Strengths and Weaknesses:** Develop targeted strategies to leverage strengths, address weaknesses, and capitalize on market opportunities.

**Future Possibilities:** Prepare for diverse outcomes with in-depth scenario analysis. Explore potential market disruptions, technology advancements, and economic changes.

Rare Disease Genetic Testing Market Size Outlook- Historic and Forecast Revenue in Three Cases

The Rare Disease Genetic Testing industry report provides a detailed analysis and outlook of revenue generated by companies from 2018 to 2023. Further, with actual data for 2023, the report forecasts the market size outlook from 2024 to 2030 in three case scenarios- low case, reference case, and high case scenarios.

### Rare Disease Genetic Testing Country Analysis and Revenue Outlook to 2030

The report analyses 22 countries worldwide including the key driving forces and market size outlook from 2021 to 2030. In addition, region analysis across Asia Pacific, Europe, the Middle East, Africa, North America, and South America is included in the study. For each of the six regions, the market size outlook by segments is forecast for 2030.

### North America Rare Disease Genetic Testing Market Size Outlook- Companies plan for focused investments in a changing environment

The US continues to remain the market leader in North America, driven by a large consumer base, the presence of well-established providers, and a strong end-user industry demand. Leading companies focus on new product launches in the changing environment. The US economy is expected to grow in 2024 (around 2.2% growth in 2024), potentially driving demand for various Rare Disease Genetic Testing market segments. Similarly, Strong end-user demand is encouraging Canadian Rare Disease Genetic Testing companies to invest in niche segments. Further, as Mexico continues to strengthen its trade relations and invest in technological advancements, the Mexico Rare Disease Genetic Testing market is expected to experience significant expansion, offering lucrative opportunities for both domestic and international stakeholders.

### Europe Rare Disease Genetic Testing Market Size Outlook-Companies investing in assessing consumers, categories, competitors, and capabilities

The German industry remains the major market for companies in the European Rare Disease Genetic Testing industry with consumers in Germany, France, the UK, Spain, Italy, and others anticipated to register a steady demand throughout the forecast period, driving the overall market prospects. In addition, the proactive approach of businesses in identifying and leveraging new growth prospects positions the European Rare Disease Genetic Testing market for an upward trajectory, fostering both domestic and international interest. Leading brands operating in the industry are emphasizing effective marketing strategies, innovative product offerings, and a keen understanding of consumer preferences.

Asia Pacific Rare Disease Genetic Testing Market Size Outlook- an attractive hub for opportunities for both local and global companies

The increasing prevalence of indications, robust healthcare expenditure, and increasing investments in healthcare infrastructure drive the demand for Rare Disease Genetic Testing in Asia Pacific. In particular, China, India, and South East Asian Rare Disease Genetic Testing markets present a compelling outlook for 2030, acting as a magnet for both domestic and multinational manufacturers seeking growth opportunities. Similarly, with a burgeoning population and a rising middle class, India offers a vast consumer market. Japanese and Korean companies are quickly aligning their strategies to navigate changes, explore new markets, and enhance their competitive edge. Our report utilizes in-depth interviews with industry experts and comprehensive data analysis to provide a comprehensive outlook of 6 major markets in the region.

Latin America Rare Disease Genetic Testing Market Size Outlook- Continued urbanization and rising income levels

Rising income levels contribute to greater purchasing power among consumers, spurring consumption and creating opportunities for market expansion. Continued urbanization and rising income levels are expected to sustainably drive consumption growth in the medium to long term.

Middle East and Africa Rare Disease Genetic Testing Market Size Outlook- continues its upward trajectory across segments

Robust demand from Middle Eastern countries including Saudi Arabia, the UAE, Qatar, Kuwait, and other GCC countries supports the overall Middle East Rare Disease Genetic Testing market potential. Fueled by increasing healthcare expenditure of individuals, growing population, and high prevalence across a few markets drives the demand for Rare Disease Genetic Testing.

Rare Disease Genetic Testing Market Company Profiles

The global Rare Disease Genetic Testing market is characterized by intense competitive conditions with leading companies opting for aggressive marketing to gain market shares. The report presents business descriptions, SWOT analysis, growth strategies, and financial profiles. Leading companies included in the study are 3billion Inc, Ambry Genetics, Artemis DNA, Arup Laboratories, Baylor Genetics, Centogene N.V., Color Genomics Inc, Coopersurgical Inc, Eurofins Scientific, Fulgent Genetics Inc,

Health Network Laboratories, Invitae Corp, Laboratory Corp of America Holdings, Macrogen Inc, Myriad Genetics Inc, Opko Health Inc, Perkin Elmer Inc, PreventionGenetics, Progenity Inc, Quest Diagnostics Inc, Realm IDX Inc, Strand Life Sciences

## Recent Rare Disease Genetic Testing Market Developments

The global Rare Disease Genetic Testing market study presents recent market news and developments including new product launches, mergers, acquisitions, expansions, product approvals, and other updates in the industry.

## Rare Disease Genetic Testing Market Report Scope

Parameters: Revenue, Volume Price

Study Period: 2023 (Base Year); 2018- 2023 (Historic Period); 2024- 2030 (Forecast Period)

Currency: USD; (Upon request, can be provided in Euro, JPY, GBP, and other Local Currency)

## Qualitative Analysis

Pricing Analysis

Value Chain Analysis

SWOT Profile

Market Dynamics- Trends, Drivers, Challenges

Porter's Five Forces Analysis

Macroeconomic Impact Analysis

Case Scenarios- Low, Base, High

## Market Segmentation:

*Rare Disease Genetic Testing Market Size, Trends, Analysis, and Outlook By Disease (Neurological Disorders, Im...*

## By Disease

Neurological Disorders

Immunological Disorders

Hematology Diseases

Endocrine & Metabolism Diseases

Cancer

Musculoskeletal Disorders

Cardiovascular Disorders

Dermatology Disease

Others

## By Technology

Next Generation Sequencing

-Whole Exome Sequencing

-Whole Genome Sequencing

Array Technology

PCR-based Testing

FISH

Sanger Sequencing

Karyotyping

## By Specialty

Molecular Genetic Tests

Chromosomal Genetic Tests

Biochemical Genetic Tests

## By End-User

Research Laboratories & CROs

Hospitals & Clinics

Diagnostic Laboratories

## Geographical Segmentation:

North America (3 markets)

Europe (6 markets)

Asia Pacific (6 markets)

Latin America (3 markets)

Middle East Africa (5 markets)

## Companies

3billion Inc

Ambry Genetics

Artemis DNA

Arup Laboratories



Baylor Genetics

Centogene N.V.

Color Genomics Inc

Coopersurgical Inc

Eurofins Scientific

Fulgent Genetics Inc

Health Network Laboratories

Invitae Corp

Laboratory Corp of America Holdings

Macrogen Inc

Myriad Genetics Inc

Opko Health Inc

Perkin Elmer Inc

PreventionGenetics

Progenity Inc

Quest Diagnostics Inc

Realm IDX Inc

Strand Life Sciences

Formats Available: Excel, PDF, and PPT

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Endocrine & Metabolism Diseases  
Cancer  
Musculoskeletal Disorders  
Cardiovascular Disorders  
Dermatology Disease  
Others  
By Technology  
Next Generation Sequencing  
-Whole Exome Sequencing  
-Whole Genome Sequencing  
Array Technology  
PCR-based Testing  
FISH  
Sanger Sequencing  
Karyotyping  
By Specialty  
Molecular Genetic Tests  
Chromosomal Genetic Tests  
Biochemical Genetic Tests  
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Research Laboratories & CROs  
Hospitals & Clinics  
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### **3BILLION INC**

Ambry Genetics  
Artemis DNA  
Arup Laboratories  
Baylor Genetics  
Centogene N.V.  
Color Genomics Inc  
Coopersurgical Inc  
Eurofins Scientific  
Fulgent Genetics Inc  
Health Network Laboratories  
Invitae Corp  
Laboratory Corp of America Holdings  
Macrogen Inc  
Myriad Genetics Inc  
Opko Health Inc  
Perkin Elmer Inc  
PreventionGenetics  
Progenity Inc  
Quest Diagnostics Inc  
Realm IDX Inc  
Strand Life Sciences

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