

# **Global Rare Disease Diagnostics Market: Focus on Disease, Trait Type, Products, Age Group, Test Type, Technology, End User, Country Data (15 Countries), and Competitive Landscape - Analysis and Forecast, 2020-2030**

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## **Abstracts**

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Market Report Coverage - Rare Disease Diagnostics

Market Segmentation

Disease Type: Gastroenterology Disease, Endocrine and Metabolism Disorders Disease, Cardiovascular Disorders, Neurology Disease, Hematology and Oncology Disease, Dermatology Disease

Product Type: Kits and Assays, Panels

Sample Type: DNA Sample, Buccal Swab Sample, Blood, Saliva

Technology: Next-Generation Sequencing, Whole Genome Sequencing, Whole Exome Sequencing, Microarrays

Trait Type: Inherited, Acquired

Test Type: Genetic Test, General Lab Test, Imaging Test

Age Group: Children Age, Adult Age

End User: Hospital Laboratories, Diagnostics Laboratories, Genetic Testing Laboratories, Cancer Research Laboratories, Cancer Research Institutes, Universities Research Laboratories

## Regional Segmentation

North America – U.S., Canada

Europe – Germany, France, Italy, U.K., Spain, Russia, Rest-of-Europe

Asia-Pacific – Japan, China, India, Australia, Singapore, Rest-of-APAC

Latin America – Brazil, Mexico, Rest-of-Latin America

Rest-of-the-World

## Growth Drivers

Rising Prevalence of Genetic Disorders

Increasing Research and Development in the Field of Rare Disease Diagnostics

Rare Genome Projects Leading to Rise in Awareness Regarding Treatment of Disease

Commitment of Big Pharma and Biotech Companies to Rare Diseases

## Market Challenges

Expensive Sequencing Procedures and Their Applications in Medical Treatments

High Capital Requirement Hampering the Expansion of Global Reach

## Stringent Regulatory Standards

### Market Opportunities

Technological Advancements in Sample Preparation for Whole Genome and Exome Sequencing in Rare Disease Sequencing

Huge Opportunity for Pharma and Medical Devices and Biotechnology Companies

### Key Companies Profiled

23andMe Inc., 3billion, Inc., Agilent Technologies, Inc., Beijing Genomics Institute (BGI), CENTOGENE N.V., Eurofins Scientific SE, GENEWIZ, Inc., Illumina, Inc., In-Depth Genomics, Invitae Corporation, Laboratory Corporation of America Holdings, OPKO Health, PerkinElmer, Inc., Quest Diagnostics Incorporated, Traverre Therapeutics, Inc.

### Key Questions Answered in this Report:

What are the possible long-term and short-term impacts of rare disease testing on the human health continuum?

What are the major market drivers, challenges, and opportunities in the rare disease testing?

What are the key development strategies being implemented by the major players in order to sustain in the competitive market?

What are the key regulatory implications in developed and developing regions for the global rare disease diagnostics market?

How are service-based companies impacting the growth of the global rare disease testing industry and further shaping up future trends?

How is each segment of the market expected to grow during the forecast period from 2020 to 2030?

Who are the leading players with significant offerings to the global rare disease diagnostics market and what is the expected market dominance for each of these leading players?

Which companies are anticipated to be highly disruptive in the future, and why?

What are the unmet needs that are yet to be met by the global rare disease diagnostics market with respect to the application areas?

What are the dynamics of various application areas and countries, which are impacting the global rare disease diagnostics market?

What are the new market opportunities of various technologies that are influencing the growth of the global rare disease diagnostics market?

How is each segment of the market is expected to grow during the forecast period from 2020 to 2030? Following are the segment types:

Product

Sample

Technology

Age group

Trait

Test

End user

Region

## Market Overview

Rare disease diagnostics has grown significantly since the technology was first commercialized with whole genome and exome sequencing, but it is important to

quantify that growth and describe future trends. The genome testing industry is proliferating, and its growth is expected to continue at a torrid pace. However, there are significant challenges that may dampen future growth, if not addressed.

Our healthcare experts have found rare disease diagnostics to be one of the most rapidly evolving technologies, and the global market is predicted to grow at a CAGR of 8.57% over the forecast period of 2020-2030.

The unmet clinical needs for better tools to predict, diagnose, treat, and monitor diseases are acting as significant factors driving the growth of the sequencing industry. Other factors fueling the growth include increased understanding of the molecular basis of diseases, patient demand, industry investment, and regulations that allow marketing of tests without the FDA approval.

Despite rapid advanced sequencing industry growth, there are several key issues that need to be addressed to facilitate future growth. The relatively high total costs of delivering sequencing test results compared with other technology platforms, and limited coverage by payers, are the key challenges to the growth of this industry. Whole genome and exome sequencing remain relatively costly, requiring initial equipment investment, specialized workforce requirements, and time-intensive variant interpretation.

Within the research report, the market has been segmented on the basis of disease type, product, sample, age group, testing type, technology, end user, and region. Each of these segments cover the snapshot of the market over the projected years, the inclination of the market revenue, underlying patterns, and trends by using analytics on the primary and secondary data obtained.

### Competitive Landscape

The exponential rise in the application of next-generation sequencing on the global level has created a buzz among companies to invest in the products and services of next-generation sequencing used for rare disease diagnostics. Due to the diverse product portfolio and intense market penetration, whole genome and exome have been pioneers in this field and significant competitors in this market.

Based on region, North America holds the largest share, owing to improved healthcare infrastructure, rise in per capita income, and improvised reimbursement policies in the region. Apart from this, Latin America and the Asia-Pacific are anticipated to grow at the

fastest CAGR during the forecast period.

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