

Global Rare Disease Diagnostics Market: Focus on Genetic Diseases, Service Providers, Competitive Landscape, and Country - Analysis and Forecast, 2018-2025

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Abstracts

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Rare diseases, also known as orphan diseases, have become a major health burden in the recent times. Comprising 6000-7000 life threatening diseases, rare diseases affect small percentage of the population. Every country has their own definition of rare disease based on the country's population. In the initial years, pharmaceutical companies were less interested in adopting them to develop treatments for such a small patient population, therefore the US FDA started giving orphan drug designations to the drug candidates intended to treat rare diseases. Orphan drug designations benefit companies from incentives for the development of these products until the marketing approval. The major challenge in rare diseases treatment is the lack of diagnosis, government policies, awareness and funding for R&D, along with long diagnostic delays.

The increasing patient pool for rare disease cancer, government funding to accelerate research in rare diseases, and interest of big pharmaceutical companies in manufacturing orphan drugs and orphan disease diagnostic devices are expected to be the major factors for the growth of the market. Pharmaceutical giants such as Shire and Qiagen are investing in the R&D for the development of innovative and improved diagnostic devices and assays for rare disease. Rise in the awareness level among people on rare diseases is also expected to fuel the growth of the market. The rare disease has different definitions across different countries. Rare diseases are genetically inherited and is difficult and expensive to diagnose and with are few

treatment options. A disease is known as rare in the United States if it affects fewer to 200,000 people, and in Europe, a disease is referred to as rare if it affects one in 2000 people. Rare diseases are also referred to those diseases that are underappreciated or ignored by the medical community and drug companies

The global rare disease diagnostic market is segmented based on disease type, test type, age, trait, end-user, and region. The disease type segment is further sub-segmented into gastroenterology, endocrine and metabolism disorders, cardiovascular disorders, neurology, hematology and oncology, dermatology, and other therapeutic areas. The test type segment is further segmented as genetic tests, general lab tests, imaging, and other physical tests for rare diseases. On the basis of trait type, the rare disease diagnostic market is segmented as inherited and acquired. Based on the age group, the rare disease diagnostic market is segmented into children and adults. On the basis of end user, the rare disease diagnostic marker is segmented as hospital laboratories, diagnostic laboratory, genetic testing laboratories, cancer research laboratories, and others genetic services area. The market analysis includes an in-depth examination of the key ecosystem players, key strategies, and developments taking place in this market. Additionally, it includes market dynamics (market drivers, opportunities, and challenges) and industry analysis. Geographically, the market can be segmented into five distinct regions including, North America, Europe, Asia-Pacific, Latin America, and Middle East Africa (MEA).

The purpose of the study is to gain a holistic view of the global rare disease diagnostic market in terms of various factors influencing it such as key market trends, competitive and regulatory aspects of the market, and scientific innovations with respect to gene sequencing. The scope of the report is centered upon conducting a detailed study of the diagnostic devices/services and assays used to diagnose rare disease. It involves wide range of diagnostic tests such as genetic tests (genome sequencing and exome sequencing), lab tests, imaging, and physical examinations that are performed in diagnostic laboratories, hospitals, cancer research institutes and other gene sequencing institutes/companies.

The rare disease diagnostic market is divided into six different segments: diseases, test type, 'trait, age, end user, and region. The report offers the reader with an opportunity to unlock comprehensive insights with respect to the market and helps in forming well informed strategic decisions. The research uncovers some of the substantial parameters that must be taken into consideration before entering the market.

This research report aims at answering questions related to various aspects of the

global market with the help of the key factors driving the market, threats that can possibly inhibit the overall market growth, and the current growth opportunities that are going to shape the future trajectory of the market expansion. The study considers the growth-share matrix model for a comprehensive study of the global enzyme market, and assesses the factors governing the same. Opportunity matrix and detailed product mapping have been included in the report. The market by region has been further sub-segmented in various countries, and in each sub-segment the key market trends, list of the key players, and recent developments, have been discussed.

Key questions answered in the report:

What are the major market drivers, restraints, and opportunities in the global rare disease diagnostic market?

What were the market shares of the leading segments and sub-segments of the global rare disease diagnostic market in 2017, and what will be shares in 2025?

How will each segment of the global rare disease diagnostic market grow during the forecast period, and what will be the revenue generated by each of the segments by the end of 2025?

What are the influencing factors that may affect the market share of the key players?

What are the key developmental strategies implemented by the key players to stand out in this market?

What are the major regulatory authorities/associations/consortiums affecting the global rare disease diagnostic market, and who are the key authorities facilitating development and approval of diagnostic products/services?

Which companies are holding gene sequencing services, and what is the epidemiology of the rare diseases?

What are the types of genetic tests involved in the diagnosis of rare diseases, and what is the market share for various test types performed in the diagnosis of rare diseases?

Which test type will be dominant among physicians for diagnosing rare diseases

in terms of revenue?

Which disease type is expected to have the highest CAGR during the forecast period?

Which test type is expected to register the highest CAGR for the global rare disease diagnostic market?

Which end user is expected to register the highest CAGR for the global rare disease diagnostic market?

Which geographical region will contribute to the highest revenue for rare disease diagnosis during the forecast period?

The report also profiles 15 companies including several key players that have been contributing significantly to the market. The key players of the market include Retrophin, Inc., 3billion, Inc., 23andMe, Inc., QIAGEN N.V., Illumina Inc., PerkinElmer, Inc., BGI, Partek, Inc., GENEWIZ, Centogene A.G., Strand Life Sciences Pvt Ltd., Eurofins Scientific, Laboratory Corporation of America, and Laboratory Corporation of America, among others.

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