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Rare Disease Diagnostics Market - Global Industry Size, Share, Trends, Opportunity, and Forecast, 2017-2027 Segmented By Indication (Gastroenterology Disease, Endocrine & Metabolism Disorders Disease, Cardiovascular Disorders, Hematology & Oncology Disease, Others), By Trait Type (Inherited v/s Acquired), By Test Type (Genetic Tests, General Lab Tests, Diagnostic Imaging, Others), By Technology (Next Generation Sequencing, Whole Genome Sequencing, Whole Exome Sequencing, Microarrays, Others), By Sample Type (DNA Sample, Blood Sample, Saliva Sample, Others), By End User (Hospitals & Clinics, Academic & Research Institutes, Others), By company and By Region

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Abstracts

The global rare disease diagnostics market is anticipated to observe impressive growth during the forecast period 2023-2027. The major factors include the growing incidence of rare disorders, adoption of advanced technology, and increase in investments in R&D are impelling the growth of the market. Rare diseases or orphan diseases is any disease that affects a small percentage of the population. The other factors supporting the market's growth are, increasing government initiatives for spreading awareness, expanding patient registry for rare diseases, declining sequencing cost, rising geriatric population, the growing launch of programs & new product development in rare disease

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diagnosis, and rise in research and development activities.

The rising incidence of rare disorders will fuel the market growth.

Rare diseases are not rarer as there are many people who are suffering from the disease. They affect more people than the combination of AIDS & Cancer. For instance, as per the European Journal of Human Genetics, the number of people living with a rare disease is estimated at 300 million across the globe. Owing to the rise in the number of people suffering from rare diseases such as Agammaglobulinemia, Goodpasture Syndrome, Granulomatosis with Polyangiitis (GPA, formerly Wegener Granulomatosis), Leukocyte Adhesion Deficiency, Pediatric Bruton Agammaglobulinemia, Pediatric Severe Combined Immunodeficiency, Schnitzler Syndrome and others, the surge in demand for the diagnosis is bolstering the growth of the market.

Technological Advancements in Disease Diagnosis

The rise in the adoption of advanced technology in the field of rare diseases diagnostic is augmenting the growth of the market. New innovative technologies and advancements such as artificial intelligence (AI), NGS-based testing, and automation technology are assisting in reducing time and cost and enabling more accurate diagnosis when compared to standard manual analysis. For instance, increase in the adoption of the Whole Exome Sequencing (WES) genetic testing tool. The technique aims only at exons, which make 1-2% of the whole genome and include 80% of all disease-causing mutations.

Rising Awareness of diagnosis of rare diseases

Early diagnosis of rare diseases is essential as it can help people to treat these conditions timely without suffering. Thus, the rise in awareness about rare diseases among the general public and patients is facilitating the growth of the market. Various initiatives by government and non-government organizations are taken to make people aware of the early diagnosis and treatment of the disease. For instance, every year, rare diseases day at NIH takes place, which aims to raise awareness about rare diseases and the impact they have on patient’s lives.

Market Segmentation

The global rare disease diagnostics market is segmented into indication, trait type, test type, technology, sample type, end user, and company. Based on indication, the market

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is divided into gastroenterology disease, endocrine & metabolism disorders disease, cardiovascular disorders, hematology & oncology disease, and others. Based on trait type, the market is divided into inherited and acquired. Based on test type, the market is segmented into genetic tests, general lab tests, diagnostic imaging, and others. Based on technology, the market is divided into next-generation sequencing, whole genome sequencing, whole exome sequencing, microarrays, and others. Based on sample type, the market is divided into DNA samples, blood sample, saliva samples, and others. Based on end-user, the market is divided into hospitals & clinics, academic & research institutes, and others. In terms of country, the United States is expected to be a lucrative market in the forecast period due to rising occurrences of rare diseases in the country.

Market Players

Agilent Technologies, Inc., Illumina, Inc., QIAGEN NV, PerkinElmer, Inc, Quest Diagnostics, Inc., CENTOGENE N.V., 23andMe, Inc., 3billion, Inc., Eurofins Scientific SE, and Invitae Corporation are some of the leading companies operating in the market.

Report Scope:

In this report, global rare disease diagnostics market has been segmented into the following categories, in addition to the industry trends which have also been detailed below:

Rare Disease Diagnostics Market, By Indication:

Gastroenterology Disease

Endocrine & Metabolism Disorders Disease

Cardiovascular Disorders

Hematology & Oncology Disease

Others

Rare Disease Diagnostics Market, By Trait Type:

Inherited

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Acquired

Rare Disease Diagnostics Market, By Test Type:

Genetic Tests

General Lab Tests

Diagnostic Imaging

Others

Rare Disease Diagnostics Market, By Technology:

Next Generation Sequencing

Whole Genome Sequencing

Whole Exome Sequencing

Microarrays

Others

Rare Disease Diagnostics Market, By Sample Type:

DNA Sample

Blood Sample

Saliva Sample

Others

Rare Disease Diagnostics Market, By End User:

Hospitals & Clinics

Academic & Research Institutes

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- Others

Rare Disease Diagnostics Market, By Region:

- North America
  - United States
  - Canada
  - Mexico
- Asia-Pacific
  - China
  - India
  - Japan
  - Australia
  - South Korea
- Europe & CIS
  - Germany
  - France
  - United Kingdom
  - Spain
  - Italy
- South America

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Brazil

Argentina

Colombia

Middle East & Africa

South Africa

Saudi Arabia

UAE

Competitive Landscape

Company Profiles: Detailed analysis of the major companies present in Global Rare Disease Diagnostics Market

Available Customizations:

With the given market data, TechSci 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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