+44 20 8123 2220
info@marketpublishers.com

Rare Disease Genetic Testing Market - A Global and Regional Analysis: Focus on Disease Type, Offering, Specialty Type, Sample Type, Trait Type, Technology, Age Group, End User, and Country Analysis - Analysis and Forecast, 2023-2033

https://marketpublishers.com/r/RF2571F00BABEN.html
Date: August 2023
Pages: 309
Price: US$ 5,500.00 (Single User License)
ID: RF2571F00BABEN

Abstracts

Introduction of Rare Disease Genetic Testing

The global rare disease genetic testing market was valued at $4.11 billion in 2022 and is anticipated to reach $16.69 billion by 2033, witnessing a CAGR of 13.69% during the forecast period 2023-2033. Cutting-edge technologies, such as whole-genome sequencing (WGS) and whole-exome sequencing (WES), involve the analysis of an individual's complete genetic code (genome) or specific protein-coding regions (exome), respectively. These methods help identify genetic variations, mutations, or abnormalities responsible for rare diseases.

Market Introduction

Before the advent of NGS (which includes WGS and WES), physicians relied on the Sanger sequencing-based tests, which could detect rare diseases but were extremely time-consuming. Additionally, till all the tests were completed, physicians had to rely on a general diagnostic odyssey or general diagnosis until definitive results were available. This delayed the treatment process. As per BIS research, the global rare disease genetic testing market includes panels and tests that can be used for more rare disease genetic testing. These are predominantly polymerase chain reaction (PCR) and next-generation sequencing (NGS) based tests, panels, and assays. The most common rare disease genetic testing panels include gastroenterology, endocrine and metabolism, and neurology disease, among others.

+44 20 8123 2220
info@marketpublishers.com

Industrial Impact

The global rare disease genetic testing market has witnessed significant growth, attributed to the increasing demand for early detection of rare diseases. The increasing prevalence of rare diseases has played a critical role in market growth. Furthermore, growing recognition and emphasis on early detection and prevention of rare diseases in pediatric populations around the world are also expected to influence market growth.

Rare disease genetic testing has immense potential to deliver next-level healthcare solutions. For instance, in January 2023, BGI Genomics, in collaboration with the National Centre for Cardiovascular Diseases, Fuwai Hospital, and the State Key Laboratory of Cardiovascular Diseases, developed the 'CardioGen automated interpretation system,' which serves as the genotype and phenotype database of monogenic cardiovascular disease. Other factors also impact the market growth, including increased patient demand and low turnaround time of these tests.

Market Segmentation:

Segmentation 1: by Disease Type

- Gastroenterology Disease
- Endocrine and Metabolism Disease
- Cardiovascular Disease
- Neurology Disease
- Hematology and Oncology Disease
- Dermatology Disease
- Other Diseases

Endocrine and Metabolism Disease Segment to Dominate the Global Rare Disease Genetic Testing Market (by Disease Type)

+44 20 8123 2220
info@marketpublishers.com

Based on disease type, the rare disease genetic testing market is led by endocrine and metabolism, which held a 21.88% share in 2022. The burden of endocrine and metabolism diseases often has a high prevalence in the general population, making their diagnosis and management particularly challenging. Their diagnosis addresses genetic testing such as NGS. Further, it also includes biomarker analysis, newborn screening, and molecular diagnostic techniques.

Segmentation 2: Offering

- Products
- Services

Services Segment to Dominate the Global Rare Disease Genetic Testing Market (by Offering)

Based on offerings, the rare disease genetic testing market is led by services, which held an 81.58% share in 2022. Services are specifically tailored to meet the distinct requirements of individuals affected by rare diseases, healthcare professionals, and researchers engaged in this field. The availability of these services complements the use of diagnostic products and facilitates comprehensive and personalized rare disease genetic testing.

Segmentation 3: by Specialty Type

- Molecular Genetic Tests
- Chromosomal Genetic Tests
- Biochemical Genetic Tests

Molecular Genetic Tests Occupying the Largest Share in the Market (by Specialty Type)

Based on specialty type, the rare disease genetic testing market is led by molecular genetic tests, which held a 58.89% share in 2022. The key reason for the increasing popularity of molecular genetic testing is that it provides crucial information for genetic counseling and family planning for individuals and families affected by rare diseases. By

+44 20 8123 2220
info@marketpublishers.com

identifying specific genetic mutations, families can gain insights into the likelihood of disease inheritance and assess the risk of passing on the condition to future generations. This type of testing is utilized to identify genetic disorders in newborns, allowing for prompt intervention and treatment initiation.

Segmentation 4: by Sample Type

- DNA Sample
- Buccal Swab Sample
- Blood Sample
- Saliva Sample
- Others

Blood Sample to Witness the Highest Growth Between 2023 and 2033

Based on sample type, the rare disease genetic testing market is led by blood samples, and it held a 33.99% share in 2022. Blood samples undergo processing to isolate different components, including white blood cells, serum, or plasma. These isolated components are then subjected to further analysis to detect genetic abnormalities, identify biochemical markers, or explore other diagnostic indicators. Genetic testing methods such as PCR, DNA sequencing, and microarrays can be employed on blood samples to identify specific genetic mutations that are associated with rare diseases.

Segmentation 5: by Trait Type

- Inherited Trait
- Acquired Trait

Inherited Trait to Witness the Highest Growth between 2023 and 2033

Based on trait type, the rare disease genetic testing market is led by inherited traits, which held an 82.59% share in 2022. Inherited traits are characteristics or conditions

+44 20 8123 2220
info@marketpublishers.com

that are passed down from parents to their offspring through genetic material, i.e., DNA. These traits result from variations or mutations in the genes and can increase the risk of developing certain rare diseases.

Segmentation 6: by Technology

- Sanger Sequencing
- PCR
- NGS
- Microarrays
- Others (Targeted Cytogenetics, Karyotyping, and FISH)

NGS to Dominate the Global Rare Disease Genetic Testing Market (by Technology)

Based on technology, the rare disease genetic testing market is led by next-generation sequencing (NGS), and it held a 43.80% share in 2022. NGS technology provides higher sensitivity and specificity in detecting genetic variants, including single-nucleotide variants (SNVs), insertions, deletions, and structural rearrangements. This technology also extends its applications to non-invasive prenatal testing, including the detection of fetal chromosomal abnormalities associated with certain rare genetic conditions.

Segmentation 7: by Age Group

- Prenatal
- Neonate and Infant
- Children and Adolescent
- Adult

Prenatal to Witness the Highest Growth between 2023 and 2033

+44 20 8123 2220
info@marketpublishers.com

Based on age group, the rare disease genetic testing market is led by the prenatal age group, which held a 39.68% share in 2022. Prenatal testing and diagnostics play a crucial role in identifying potential rare diseases and congenital abnormalities in the fetus. Prenatal testing methods such as chorionic villus sampling (CVS) and amniocentesis are employed to detect genetic disorders or developmental issues.

Segmentation 8: by End User

- Hospital
- Diagnostics Laboratories
- Other End Users

Hospitals to Dominate the Global Rare Disease Genetic Testing Market (by End User)

Based on end users, the rare disease genetic testing market is led by hospitals, which recorded a 53.12% share in 2022. Hospitals are at the forefront of adopting syndromic testing panels into routine healthcare procedures. Hospitals, particularly in leading regions such as North America and Europe, and also to a certain extent in Asia-Pacific, have incorporated syndromic testing panels and assays to provide superior care to patients.

Segmentation 9: by Region

- North America - U.S., Canada
- Europe - Germany, U.K., France, Italy, Russia, Spain, and Rest-of-Europe
- Asia-Pacific - Japan, India, China, South Korea, Australia, Singapore, and Rest-of-Asia-Pacific
- Latin America - Brazil, Mexico, and Rest-of-Latin America
- Rest-of-the-World

Regions such as North America and Europe have technologically advanced industries

and allocate considerable budgets to research and development (R&D) activities. Moreover, a maximum number of rare disease testing is done there, due to which these regions hold the maximum share in the global rare disease genetic testing market.

Recent Developments in the Rare Disease Genetic Testing Market

In April 2023, Agilent Technologies, Inc. and Theragen Bio collaborated in South Korea to advance bioinformatic solutions for cancer genomic profiling.

In April 2023, 3 billion receives accreditation from the California Department of Public Health (CDPH) as a clinical laboratory. With the CDPH accreditation, hospitals throughout California will now be able to utilize 3 billion's genetic tests for diagnostic purposes and can claim insurance for the tests.

In February 2023, Akili Labs and BGI Genomics established the first commercial clinical sequencing facility in Africa.

In January 2023, Premier Research and CENTOGENE formed a strategic partnership to hasten and reduce the risk of clinical development for rare diseases.

In February 2023, Illumina delivers the first NovaSeq X Plus sequencer and workflow insights on Illumina Complete Long Reads unveiled at Advances in Genome Biology and Technology (AGBT).

In March 2023, Invitae Announces Partnership with Epic to Streamline Genetic Testing. Through Aura, Epic's specialty diagnostics suite, Invitae will streamline interactions with provider organizations in the Epic community, making test result information available in providers' usual workflows so that it's easier to use genetic insights to inform treatment decisions.

Demand – Drivers, Challenges, and Opportunities

Market Demand Drivers:

Rapid Technological Advancements in Rare Disease Diagnosis: Technological advancements, such as next-generation sequencing (NGS), targeted gene panels, and artificial intelligence (AI)-based algorithms, have significantly improved diagnostic

+44 20 8123 2220
info@marketpublishers.com

accuracy for rare diseases. NGS enables comprehensive genomic analysis, allowing for the identification of disease-causing genetic mutations with higher precision. By identifying the underlying genetic mechanisms and individualizing treatment plans, technological advancements are improving patient outcomes, patient satisfaction, and overall market growth. These advancements have also led to the emergence of new areas of rare disease genetic testing, which have opened up new avenues for research and drug discovery.

Rising Prevalence of Genetic Disorders and Congenital Disorders: The rising prevalence of genetic disorders and congenital disorders has a significant impact on the growth of the global rare disease genetic testing market. Increased demand for diagnostic services, expansion of screening programs, focus on personalized medicine, research and development initiatives, supportive regulatory environment, and patient advocacy all contribute to the expanding market. Furthermore, the rising prevalence led to an expansion of screening programs. Governments and healthcare organizations are implementing newborn screening programs and prenatal genetic testing initiatives to identify these disorders early.

Growing Recognition and Emphasis on Early Detection and Prevention of Rare Diseases in Paediatric Populations: Increased consumer demand for early detection and prevention, coupled with a focus on personalized medicine and precision healthcare, is expected to fuel the growth of the global rare disease market. Awareness and education, patient empowerment, expanded screening programs, preventive healthcare strategies, supportive policies, and insurance coverage are all contributing to the expansion of diagnostic services and driving market demand for rare disease genetic testing.

Market Challenges:

Lack of Consolidation of Information Leading to Difficulty in Diagnosis: The lack of consolidation of information and the resulting difficulty in diagnosis poses significant challenges for the global rare disease genetic testing market. Rare diseases often have limited available information, fragmented research findings, and scattered expertise. The vast number of rare diseases, each with its unique set of symptoms and genetic variations, makes it challenging to consolidate comprehensive knowledge and expertise in a centralized manner.

Significant Capital Requirement for Research and Development Hindering Global Expansion Efforts: The significant capital requirement for research and development

+44 20 8123 2220
info@marketpublishers.com

(R&D) poses a notable challenge for the global rare disease genetic testing market. Research and development in the field of rare diseases often involve extensive genetic studies, clinical trials, and the development of innovative diagnostic tools and therapies. These activities require substantial financial investment, as they involve specialized expertise, laboratory infrastructure, regulatory compliance, and patient recruitment. The high costs associated with R&D act as a barrier, particularly for smaller companies and research institutions with limited resources, hindering their ability to undertake comprehensive research and development efforts. Rare diseases, by definition, affect a small patient population. Hence, the limited commercial market potential for treatments and diagnostics creates challenges in attracting sufficient private investment.

Market Opportunities:

Whole-Genome Sequencing Putting an End to the Rare Disease Diagnostic Odysseys: Whole-genome sequencing (WGS) has the potential to put an end to rare disease genetic testing odysseys and presents significant opportunities for the global rare disease genetic testing market. This technology enables a comprehensive analysis of an individual's entire genome, providing a comprehensive view of the genetic variations and mutations. This approach allows for the detection of both common and rare genetic variants, including those associated with rare diseases.

AI-Based Diagnostic Tools Diagnosing Rare Diseases by Identifying Potential Diagnoses: AI-based diagnostic tools that utilize machine learning and artificial intelligence algorithms present significant opportunities for the global rare disease market by improving the diagnosis of rare diseases. Artificial intelligence (AI) and machine learning algorithms are revolutionizing rare disease genetic testing by analyzing large datasets, identifying patterns, and making accurate predictions. For instance, in July 2021, Sema4, the parent organization of GeneDx, successfully concluded a transaction with CM Life Sciences, marking its debut as a publicly traded company. The company now operates as an AI-driven genomics and clinical data platform company.

Transitioning toward Increased Genetics Coverage in Reimbursement Policies: Transitioning toward increased genetics coverage in reimbursement policies presents a valuable opportunity for the global rare disease genetic testing market. It improves access to genetic testing, encourages early diagnosis and intervention, supports research and development efforts, stimulates innovation, reduces financial burdens for patients, and aligns with the principles of personalized medicine. These factors collectively contribute to improved diagnostic rates and advancements in rare disease

+44 20 8123 2220
info@marketpublishers.com

management.

How Can This Report Add Value to an Organization?

Product/Innovation Strategy: The global rare disease genetic testing market has been extensively segmented on the basis of various categories, such as disease type, specialty type, sample type, end-user, and region. This can help readers understand which segments account for the largest share and which are well-positioned to grow in the coming years.

Growth/Marketing Strategy: Partnerships, collaborations, product launches, and upgrades accounted for the maximum number of key developments, i.e., nearly 86.2% of the total developments in the global rare disease genetic testing market were between January 2021 and June 2023.

Competitive Strategy: The global rare disease genetic testing market has numerous established players paving their way into providing tests, panels, and assays in the market. Key players in the global rare disease genetic testing market analyzed and profiled in the study involve established players offering various disease-specific panels and multiplex instruments.

Key Market Players and Competition Synopsis

The companies that are profiled have been selected based on inputs gathered from primary experts and analyzing company coverage, product portfolio, and market penetration.

Key Companies Profiled:

- 23andMe Holding Co.
- 3billion, Inc.
- Agilent Technologies, Inc.
- Konica Minolta (Ambry Genetics, Inc.)
- BGI Group

+44 20 8123 2220
info@marketpublishers.com

CENTOGENE N.V.

Eurofins Scientific SE

GeneDx, Holdings Corp.

Azenta U.S., Inc. (GENEWIZ, Inc.)

Illumina, Inc.

Invitae Corporation

Laboratory Corporation of America Holdings

Medgenome

Myriad Genetics, Inc.

OPKO Health, Inc.

PerkinElmer, Inc.

Exact Sciences Corporation (Prevention Genetics)

QIAGEN N.V.

Quest Diagnostics Incorporated

Strand Life Science Pvt Ltd.

+44 20 8123 2220
info@marketpublishers.com

Contents

+44 20 8123 2220
info@marketpublishers.com

+44 20 8123 2220
info@marketpublishers.com

4 REGION

+44 20 8123 2220
info@marketpublishers.com

+44 20 8123 2220
info@marketpublishers.com

+44 20 8123 2220
info@marketpublishers.com

+44 20 8123 2220
info@marketpublishers.com

+44 20 8123 2220
info@marketpublishers.com

+44 20 8123 2220
info@marketpublishers.com

+44 20 8123 2220
info@marketpublishers.com

+44 20 8123 2220
info@marketpublishers.com

+44 20 8123 2220
info@marketpublishers.com

+44 20 8123 2220
info@marketpublishers.com

5 MARKETS - COMPETITIVE BENCHMARKING & COMPANY PROFILES

+44 20 8123 2220
info@marketpublishers.com

+44 20 8123 2220
info@marketpublishers.com

+44 20 8123 2220
info@marketpublishers.com

+44 20 8123 2220
info@marketpublishers.com

+44 20 8123 2220
info@marketpublishers.com

+44 20 8123 2220
info@marketpublishers.com

6 RESEARCH METHODOLOGY

+44 20 8123 2220
info@marketpublishers.com

List Of Figures

LIST OF FIGURES

+44 20 8123 2220
info@marketpublishers.com

+44 20 8123 2220
info@marketpublishers.com

+44 20 8123 2220
info@marketpublishers.com

List Of Tables

LIST OF TABLES

+44 20 8123 2220
info@marketpublishers.com

+44 20 8123 2220
info@marketpublishers.com

+44 20 8123 2220
info@marketpublishers.com

+44 20 8123 2220
info@marketpublishers.com

+44 20 8123 2220
info@marketpublishers.com

+44 20 8123 2220
info@marketpublishers.com

I would like to order

Product name: Rare Disease Genetic Testing Market - A Global and Regional Analysis: Focus on Disease Type, Offering, Specialty Type, Sample Type, Trait Type, Technology, Age Group, End User, and Country Analysis - Analysis and Forecast, 2023-2033

Product link: https://marketpublishers.com/r/RF2571F00BABEN.html

Price: US$ 5,500.00 (Single User License / Electronic Delivery)

If you want to order Corporate License or Hard Copy, please, contact our Customer Service:
info@marketpublishers.com

Payment

To pay by Credit Card (Visa, MasterCard, American Express, PayPal), please, click button on product page https://marketpublishers.com/r/RF2571F00BABEN.html

To pay by Wire Transfer, please, fill in your contact details in the form below:

First name:
Last name:
Email:
Company:
Address:
City:
Zip code:
Country:
Tel:
Fax:
Your message:

**All fields are required

Custumer signature _____

Please, note that by ordering from marketpublishers.com you are agreeing to our Terms & Conditions at https://marketpublishers.com/docs/terms.html

To place an order via fax simply print this form, fill in the information below

+44 20 8123 2220
info@marketpublishers.com

and fax the completed form to +44 20 7900 3970