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MOLECULAR DIAGNOSTICS IN GENETIC TESTING *(SAMPLE COPY, NOT FOR RESALE)*

Trends, Industry Participants, Product Overviews and Market Drivers

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SAMPLE

1. Overview

1.1 Statement of Report

Molecular diagnostics for genetic testing brings advanced analytical techniques to the diagnosis and treatment of genetic disorders. The confluence of breakthroughs in genomics and proteomics and the development of microarray devices to measure analytes in the blood and various body tissues are driving significant growth in the segment. Major developments include the integration of specialty labs and gene expression profiling into clinical practice, the introduction and rapid growth of cell-free fetal DNA prenatal testing, the development of companion diagnostics for drug development, the widespread installed base of automated instruments for molecular testing and the development of personalized medicine. The genetic testing space is one of the most profitable sectors of molecular diagnostics and is expected to be an area of high growth and corporate change throughout the forecast period. This TriMark Publications report describes the emerging field of molecular diagnostics for genetic testing. This review analyzes the size and growth of the molecular diagnostics for genetic testing market, including the factors that influence the various market segments within it and the dollar volume of sales, both in the U.S. and worldwide. Moreover, this analysis profiles the leading companies focused on the molecular diagnostics for genetic testing sector. Also examined are:

- Polymerase chain reaction (PCR) technology platforms.
- Clinical applications of nucleic acid based genetic testing.
- The market for molecular diagnostic tests associated with diagnosing genetic disorders.
- Companies participating in this sector.
- New technology platforms and diagnostic test kits.
- Trends in the molecular diagnostics industry sector.
- The internal structure of the molecular diagnostics clinical testing sector.

The genetic testing market has been divided into the following segments for ease of examination:

- Diagnostic testing of genetic disorders.
- Carrier testing for genetic disorders.
- Pre-implantation genetic diagnosis.
- Prenatal/*In utero* genetic screening.
- Newborn genetic screening tests.
- Predictive and pre-symptomatic testing for adult onset disorders.
- Pharmacogenomics and pharmacogenetics.
- Tissue and blood typing.
- Paternity testing.
- Criminal and forensics testing.
- Genealogical DNA Testing.

1.2 About this Report

This report includes the following features:

- It examines the generally accepted clinical analytical activities in use today in the molecular diagnostics (MDx) sector for diagnosis and management of genetic disorders. It includes the prevalent devices and the accompanying reagents and supplies as utilized in hospitals and large reference and specialty Clinical Laboratory Improvement Act (CLIA) licensed laboratories.
- It discusses the potential benefits of the molecular diagnostics technique for various sectors of the medical and scientific communities. It also assesses the market drivers and bottlenecks for MDx tests from the perspective of these communities.

- It establishes the current total MDx market size and future growth of the molecular diagnostics market for genetic testing and analyzes the current size and growth of key segments.
- It assesses various business models in molecular diagnostics for genetic testing, including CLIA-licensed specialty labs, general reference labs and reagent kit marketing, as well as provides strategic recommendations for near-term business opportunities.
- It examines the products offered and roles played by companies that have invested significantly in this market, and it provides current and forecasted market shares by these companies.
- It discusses new collaborative business models that bring together diagnostics and therapeutics.
- It evaluates the role that genetic testing prognostic assays can play in personalized medicine.

1.3 Scope of the Report

The goal of this study is to review the market for molecular diagnostics for genetic testing equipment and supplies using reagents and instruments for analysis of individual components in body tissues and fluids. Toward this goal, this review addresses the following key questions and topics:

- Which companies are utilizing new, cutting-edge technologies to develop, validate and market MDx tests for clinical use in genetic testing management?
- What are the current impediments to incorporating promising molecular tests into practice?
- Which new MDx genetic tests show the most promise for regulatory approval?
- What are the economic challenges to gaining approval?
- What kinds of MDx genetic tests are best?
- How does regulatory oversight drive approval and adoption of new technologies?
- Which strategic alliances show the greatest synergy in bringing molecular diagnostics tests to market?
- Which shared technologies are driving the most encouraging developments of new genetic testing methods?
- Estimate the current and future U.S. and global markets for molecular diagnostics for genetic testing.
- Examine market drivers that have resulted in the global race for new genetic testing diagnostics.
- Assess market opportunities.
- Discuss product development challenges in relation to regulatory constraints, legislative constraints and technical challenges.
- Analyze the need for MDx for the different genetic testing indications.
- Understand the impact of current products and the future of molecular diagnostics industry.
- Provide insight into the MDx genetic testing products in the pipeline and the companies that strive to bring these products to the market in the immediate future.
- Gain insight into the current applications of MDx in a wide variety of genetic testing segments.
- Explore drug discovery efforts in relation to MDx methods.
- Analyze the usual hurdles new MDx methods are encountering to reach the market and the right path to the market for these products.
- Review the current licensing, investing and partnering activities in molecular diagnostics sector.
- Assess business models and requirements for a successful MDx industry.
- Examine funding scenario for the MDx sector.
- Identify the key players in the MDx for genetic testing industry and their contribution to this market.

Key questions answered in this study are:

- What are the products that utilize MDx for genetic testing?
- What applications offer the greatest potential for genetic tests using MDx technology platforms?
- What is the current global market for MDx for genetic testing?

- What development activities are taking place globally in the MDx for genetic testing sector?
- How many companies are involved in the development of MDx products for genetic testing?
- What patents have been issued for MDx products in genetic testing?
- What is the effect of patent expiration on genetic tests utilizing molecular diagnostics?
- What market drivers are responsible for the global growth of genetic testing?
- What regulatory and technical challenges are being confronted by the MDx industry?
- What are the current promising developments in the MDx for genetic testing sector?
- What is the latest position of the clinical studies and product pipeline in MDx for genetic testing sector?
- What is the potential population in the U.S. for genetic testing?
- Do the developing countries offer scope for the growth of MDx products related to genetic testing?
- What are the MDx for genetic testing products available in the market place?
- How many companies are involved in the development of MDx for genetic testing?
- Which firms are involved in the development of government-oriented products?
- Which companies are focused on the development of paternity products?
- Which companies are focusing on clinical MDx for genetic testing products?
- What are the different business models suitable for the different types of MDx for genetic testing products?
- What are the requirements for the commercial manufacturing of MDx products?
- What are the different funding sources in the U.S. for the development of MDx products?

This report contains:

- Current market opportunities for MDx products for genetic testing.
- Product development challenges confronted by the MDx industry.
- A brief discussion on the need for molecular diagnostics.
- The future direction of the emerging MDx products for genetic testing.
- The overall picture of pipeline products in the MDx for genetic testing sector and the companies involved.
- A market projection for global MDx for genetic testing products.
- Listing and explaining the most widely used MDx products for genetic testing in the market.
- Presentation of the global picture of the MDx for genetic testing market with particular reference to the leading countries.

This examination surveys most of the biotech companies known to be currently marketing, manufacturing or developing instruments and reagents for the genetic testing market in both the U.S. and the world. Each company is discussed in depth, with sections on its history, product line, business and marketing analysis, along with a subjective commentary of the company's market position. The U.S. is the focus of this report. Primary attention is paid to the specialty and reference lab market segment and, separately, to the instruments, reagents and supplies marketed by the leading companies in this segment. Market size, growth rates and market components for instruments, reagents, controls and consumables used in this area are also analyzed.

1.4 Objectives

The main objectives of this analysis are:

- Identifying viable technology drivers through a comprehensive look at platform technologies for molecular diagnostics in genetic testing, including probe-based nucleic acid assays, microarrays and sequencing.
- Obtaining a complete understanding of the chief characteristics of molecular diagnostics tests as they are used in genetic testing (predictive, screening, prognostic, monitoring, pharmacogenomic and theranostic tests) from their basic principles to their applications.
- Discovering feasible market opportunities by identifying high-growth applications in different genetic testing areas.
- Focusing on global industry development through an in-depth analysis of the major world markets for molecular diagnostics for genetic testing, including growth forecasts.

The emphasis in this report is on the clinical use of molecular diagnostics tests for genetic testing. The reader should consult other TriMark Publications reports on the TriMark website for detailed discussions of important individual market segments related to the molecular diagnostics market or routine testing. In addition to this report, TriMark Publications offers a complete suite of market reports aimed at the molecular diagnostic space including: *Molecular Diagnostics Markets*, *Molecular Diagnostics in Cancer Testing* and *Molecular Diagnostics in Infectious Disease Testing*.

1.5 Methodology

The author of this report holds a Master's in immunology and has substantial experience in science writing and as a medical industry analyst. She also has many years of laboratory experience and has conducted laboratory testing and instrument and reagent development for biotech companies. The editor of this report holds a Ph.D. in biochemistry from the University of Minnesota and has had post-doctoral experience at the University of Connecticut School of Medicine. He has taught at Quinnipiac University, the Tufts School of Medicine and the New York Institute of Technology. He has been a senior scientist at DuPont and Pfizer Pharmaceutical Laboratories in drug development and diagnostic testing. He also has many decades of experience in science writing and as a medical industry analyst. He has over 40 years of experience in laboratory testing and instrument and reagent development technology as a licensed clinical laboratory director, as well as extensive experience in senior level management positions in biotech and medical service companies.

Company-specific information is obtained mainly from industry trade publications, academic journals, news and research articles, press releases and corporate websites, as well as annual reports for publicly-held firms. Additional sources of information include non-governmental organizations (NGOs) such as the World Health Organization (WHO) and governmental entities such as the U.S. Department of Health and Human Services (HHS), the National Institutes of Health (NIH), the Food and Drug Administration (FDA) and the Centers for Disease Control and Prevention (CDC). Where possible and practicable, the most recent data available have been used.

Some of the statistical information was taken from Biotechnology Associates' databases and from TriMark's private data stores. The information in this study was obtained from sources that we believe to be reliable, but TriMark does not guarantee the accuracy, adequacy or completeness of any information or omission or for the results obtained by the use of such information. Key information from the business literature was used as a basis to conduct dialogue with and obtain expert opinion from market professionals regarding commercial potential and market sizes. Senior managers from major company players were interviewed for part of the information in this report.

Primary Sources

TriMark collects information from hundreds of Database Tables and many comprehensive multi-client research projects, as well as Sector Snapshots that it publishes annually. TriMark extracts relevant data and analytics from its research as part of this data collection.

Secondary Sources

TriMark uses research publications, journals, magazines, newspapers, newsletters, industry reports, investment research reports, trade and industry association reports, government-affiliated trade releases and other published information as part of its secondary research materials. The information is then analyzed and translated by the Industry Research Group into a TriMark study. The Editorial Group reviews the complete package with product and market forecasts, critical industry trends, threats and opportunities, competitive strategies and market share determinations.

TriMark Publications Report, Research and Data Acquisition Structure

The general sequence of research and analysis activity prior to the publication of every report in TriMark Publications includes the following items:

- Completing an extensive secondary research effort on an important market sector, including gathering all relevant information from corporate reporting, publicly-available data and proprietary databases.
- Formulating a study outline with the assigned writer, including important items, as follows:
 - Market and product segment grouping, and evaluating their relative significance.
 - Key competitors' evaluations, including their relative positions in the business and other relevant facts to prioritize diligence levels and assist in designing a primary research strategy.
 - End-user research to evaluate analytical significance in market estimation.
 - Supply chain research and analysis to identify any factors affecting the market.
 - New technology platforms and cutting-edge applications.
- Identifying the key technology and market trends that drive or affect these markets.
- Assessing the regional significance for each product and market segment for proper emphasis of further regional/national primary and secondary research.
- Completing a confirmatory primary research assessment of the report's findings with the assistance of expert panel partners from the industry being analyzed.

For projection for the future values and growth rates of specific markets our analysts use a proprietary forecast spreadsheet, which takes into account a wide variety of market indices such as inflation rates and anticipated increases/decreases in funding of specific markets. Then, other factors such as new market introductions and acquisitions are manually incorporated into the spreadsheet.

1.6 Executive Summary

Molecular diagnostics is a rapidly advancing area of research and medicine, with new technologies and applications being continually added. A substantial part of MDx technology has been devoted to human DNA detection for:

- Detecting and diagnosing a specific disease.
- Assessing an individual's probability of developing a specific disease.
- Predicting the effectiveness of targeted drug therapies in individuals with a specific disease (pharmacogenetics/pharmacogenomics).
- Ascertaining hereditary and/or relationship information (formerly called paternity testing).
- Identity testing for forensics or criminal investigations (also referred to as DNA testing).
- Tissue typing for transplantation.

Over the past several years, this rapidly evolving field has seen several fascinating developments, including:

- Integration of specialty labs and gene expression profiling into clinical practice.
- Introduction and rapid growth of cell-free fetal DNA prenatal testing.
- Development of companion diagnostics for drug development.
- Widespread installed base of automated instruments for molecular testing.
- Development of personalized medicine.
- Rapid expansion of MDx test menus.
- Impact on pharmacogenomics and molecular epidemiology.
- Integration of specialty labs and gene expression profiling into clinical practice.
- Integration into therapeutic choices for cancer and the use of diagnostics for predicting disease recurrence.
- Development of lab-on-a-chip devices.
- Use of gene expression profiling for determining the efficacy of therapeutic drugs for cancer.

The technologies that are described as molecular diagnostics include:

- First-generation amplification, DNA probes, fluorescent *in situ* hybridization (FISH).
- Second-generation biochips and microfluidics.
- Next generation signal detection, biosensors and molecular labels, and gene expression profiling using microarrays.

Small- and medium-sized companies with innovative products and technology platforms have great opportunities for success in the field of molecular diagnostics. The exciting thing here is that this market segment is characterized by unprecedented growth rates, which stand in contrast with the low or even negative growth rates of mature laboratory-testing segments in fields such as hematology, chemistry and microbiology. Research in genomics has led to a new healthcare paradigm, where a disease is understood at the molecular level, allowing patients to be diagnosed based on genetic their own unique information and then treated with drugs designed to work on specific molecular targets.

Worldwide, the molecular genetic testing market was \$ ██████ in ██████, and is expected to increase at a compound annual growth rate (CAGR) of ██████% to \$ ██████ by ██████. The U.S. represents the largest market for genetic testing worldwide, with approximately ██████% of the genetic testing market. Leading players in the genetic testing market include: Roche, Hologic, Abbott Diagnostics, Qiagen, Sequenom, Natera, PerkinElmer Genetics, Immunocor, Life Technologies, Progenika and Counsyl.

Genetic disease management, which includes screening, diagnosis and monitoring, continues to be a market with strong clinical need. One of the most frequently performed screening assays for genetic disorders in the U.S. is the detection of mutations in the CFTR (Cystic Fibrosis Trans-Membrane Conductance Regulator) gene. The molecular market is expected to expand for newborn screening as more advanced technology becomes integrated into recommended protocols for screening. In addition, advances in genetic testing procedures will promote testing for the early detection of Alzheimer disease, hemochromatosis, breast cancer, colorectal cancer, diabetes in young people and rare forms of amyotrophic lateral sclerosis. Moreover, the market will thrive with the personalization of diagnosis and therapy by identifying genes associated with complex diseases, optimizing the drug response and reducing side effects and failure rates.

Pharmaceutical companies are investing billions of dollars in the development of high-potential targeted therapies, one of the fastest growing segments of oncology drug development. Many of these therapies will require a specific test (referred to as a “theranostic” or “companion diagnostic”) to assist physicians in selecting the right drug for the right patient. The theranostic is likely to accelerate the process for drug approval and market introduction by guiding selection of the most appropriate patients for the clinical trials. The FDA’s “Critical Path Initiative” is facilitating a national effort to modernize the scientific process through which a potential human drug, biological product or medical device is transformed from a discovery or “proof-of-concept” into a medical product.

Segments with significant revenue potential will benefit from the principal advantages of molecular diagnostics: sensitivity, specificity and speed. In addition, non-invasive or minimally-invasive procedures for obtaining the material for those tests will facilitate implementation of these technologies. Cross benefits, such as near-instant diagnostic results, better targeted therapies and shorter hospitalization times will compensate for higher costs for molecular tests compared with traditional analyses, such as microbiological assays. However, molecular diagnostics will help abate the cost for diagnosis, therapy and healthcare altogether in the long term.

Key drivers of growth in the molecular genetic testing market include:

- Personalization of diagnosis and therapy by identifying genes associated with complex diseases, optimizing the drug response and reducing side effects and failure rates (pharmacogenetics).
- Increased interaction between pharmaceutical companies and molecular diagnostics manufacturers to continue to development of companion diagnostic assays, which will aid in determining whether a given patient qualifies for a particular drug (pharmacogenomics).
- Need for faster methods of diagnosing disease states and medical disorders earlier, and for a powerful, reliable tool for fast therapy decisions (automatization).
- Need for an automated, easy-to-handle tool that optimizes sample preparation, analysis and data evaluation (lab-on-a-chip). These developmental improvements will lead to the availability of molecular diagnostics devices that can be used for near-patient or point-of-care testing.
- Need to contain or decrease healthcare costs without compromising accuracy or reliability.