GLOBAL PRENATAL & MATERNAL DIAGNOSTIC MARKET ANALYSIS TO 2020

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Global Prenatal, Maternal & Carrier Diagnostic Market Analysis to 2021
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1.0 Introduction

Prenatal tests (fetus, embryos and newborns) currently on the market analyze chromosomes, DNA, RNA, genes, and/or gene products to determine whether an abnormality is present that is causative of a specific disease. Since 2011, the launch of noninvasive prenatal tests (NIPTs) is revolutionizing the prenatal screening industry providing detection rates of >99% and false positive rates of <0.1%. These screening results are only indicative of the risk and not confirmative. To confirm positive results of NIPT screening tests, prenatal diagnostic tests such as amniocentesis and chorionic villus sampling (CVS) are performed. Prior to NIPTs, ultrasound and maternal serum tests were routinely used to screen fetusus for genetic abnormalities.

Globally, 50 countries have effective newborn genetic test programs, the implementation of which is in various stages. In the U.S., four million newborns are tested annually, with 3,000 found to have metabolic and genetic diseases. The purpose of newborn screening is to identify affected newborns quickly and provide them with treatment to prevent mental retardation, prolonged illness and death. The newborn genetic screening market is crowded with different technologies, the common being tandem mass spectrometry, pulse oximetry, enzyme-based assays, DNA assays and electrophoresis.

Preimplantation genetic screening and preimplantation genetic diagnosis (PGS/PGD) are genetic tests performed in eight-celled embryos before implantation during in vitro fertilization (IVF) for the detection of genetic abnormalities and sex. These tests can detect and diagnose chromosomal rearrangements, X-linked diseases and help in reducing the incidence of spontaneous abortions, increase implantation rates, prevent trisomic offsprings and avoid the risk of transmitting single gene disorders. However, PGS and PGD results are not 100% accurate and after pregnancy, if required the diagnostic tests such as amniocentesis and CVS are to be performed to confirm the positive results of PGS/PGD. In the U.S. alone, more than 63,000 babies were born through IVF in 2013 and compared to 2012 data; it was an increase of 2,000 births.

The most important development in the field of prenatal screening, is the introduction of NIPTs using cell-free fetal DNA (cffDNA) in 2011. Obstetricians and patients who are deterred by the 10% to 15% false positive rates of ultrasound screening and 5% false positive rates of maternal serum tests are eagerly embracing NIPTs which claim to have only <0.1% of false positive results.

Within just four years, molecular genetics firms have successfully produced eight different types of kits for the detection of genetic abnormalities in the fetus. These products have been gradually marginalizing the maternal serum tests and in another decade, maternal serum tests are expected to become obsolete. If, companies can develop NIPTs with a 100% accuracy rate, maternal serum tests, amniocentesis and CVS will all be completely overshadowed by NIPTs. Despite the growing popularity of NIPTs, maternal serum tests still continue to have some market due to their low price. While the maternal serum tests are available for just $150 to $200, NIPTs are priced between $795 and $2,762. However, Sequenom reportedly sold over 150,000 tests in 2013 and so pricing does not seem to be a deterrent factor. The introduction of technologies such as digital PCR and next-generation sequencing (NGS) or massively parallel sequencing (MPS) have also enabled accurate estimation of very small differences in chromosome-specific sequences in maternal blood.

1.1 Executive Summary

According to WHO, nearly 140 million babies are born every year, out of which 5 million die in the first month of life, mostly in developing countries. For example, in India, about 5% to 15% of sick newborns have a metabolic disorder. This emphasizes the need for newborn screening for preventing disability and death by early intervention, follow-up and counselling.

Globally, the use of maternal serum marker screening and ultrasound imaging for the detection of chromosome aneuploidies and other birth defects constitute a routine part of prenatal care in the first and/or second trimesters. Yet, both of these techniques have the disadvantages of high false positive rates, varying from 2% to 7%. If the results of these tests show that a fetus is at increased risk of aneuploidy, invasive approaches such as chorionic villus sampling (CVS) or amniocentesis are recommended for diagnosis.

Fetal Ultrasound Screening Market

Today, ultrasound is used on average five times per pregnancy before delivery. The fetal ultrasound screening market generated revenues of about $x million in 2015 with the potential to earn $x million in 2021, growing at a CAGR of x%. Fetal MRI prenatal screening is performed rarely, only when the ultrasound screening provides an ambiguous result and to detect suspected central nervous system (CNS) disorders in the fetus during the second trimester. The fetal MRI segment had generated estimated revenue of $x million in 2015 with a potential to earn $x million in 2021.

Maternal Serum Testing Market

The global maternal serum test market was worth $x million in 2015 and is forecast to grow and reach $x million by 2021. The market consists of test methods for:

- Pregnancy-associated plasma protein (PAPP-A)
- Alpha-fetoprotein (AFP)
- Human chorionic gonadotropin (hCG)
- Estriol
- Inhibin-A

Had it not been for the launch of noninvasive prenatal tests (NIPTs) in 2011, this market would have fared much better.

Noninvasive Prenatal Test Market

Noninvasive prenatal tests (NIPTs) are the future of prenatal screening and have shown signs of surpassing the traditional maternal serum test revenues by earning about $x million in 2015. With a CAGR of x%, this market is forecast to reach $x million in 2021.

Newborn Screening Market

Newborn screening is an accepted national health policy in about 52 countries. It is a well-entrenched routine of newborn care in the U.S., U.K., Japan, Australia and most western European countries for the last three decades. In the Asian continent, China, Philippines and Thailand have developed admirable newborn screening programs in a relatively short period of time. Mass spectrometry has become the standard technique for newborn screening. Using this technique, medical technologists are capable of screening as many as 30 metabolic disorders from a single sample of blood. The global market for newborn screening has been valued at $x million in 2015 and it will be worth about $x million by 2021.

Preimplantation Genetic Diagnosis Market

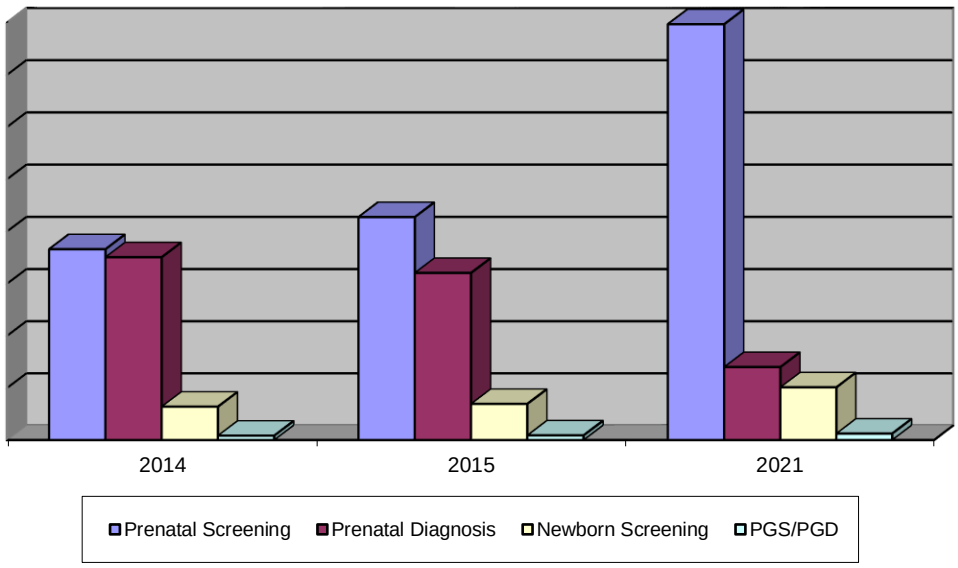
Preimplantation genetic diagnosis (PGD) has been in practice for more than twenty years in about 60 countries globally. PGD testing is performed to identify genetic defects in embryos. The tests are usually performed in cases of a known genetic defect, in late age pregnancy, or in patients with a history of repeated miscarriages. PCR, FISH, CGH, and SNP analysis are some of the most common techniques used, and PCR is the most widely used technology. The global market for PGD was worth about $x million in 2015 and this has been predicted to enlarge and reach $x million in 2021.

Table 1.1: Summary of Prenatal Screening, Prenatal Diagnosis, Newborn Screening and PGS/PGD Markets, Through 2021

Type of Test	2014 ($ Millions)	2015 ($ Millions)	2021 ($ Millions)	% CAGR 2015-2021
Prenatal screening:				
Ultrasound screening				
Fetal MRI screening				
Maternal serum screening				
Noninvasive prenatal screening (NIPT)				
Total for prenatal screening				
Prenatal diagnosis (amniocentesis and CVS				
Newborn screening				
PGS/PGD				
Total				

Source: www.kellyscipub.com

Figure 1.1: Summary of Prenatal Screening, Prenatal Diagnosis, Newborn Screening and PGS/PGD Markets, Through 2021



Source: www.kellyscipub.com

1.2 Objectives of this Report

Since the 1960s, technologies for testing fetuses for conditions including trisomies have been continually advancing. The initial nongenetic testing such as ultrasound and serum screening for protein level markers, have developed remarkably throughout the years. Yet, these conventional tests are limited in sensitivity and specificity. Parallel to the development of these conventional technologies, two invasive techniques called amniocentesis and chorionic villus sampling (CVS) were introduced in 1980s to offer near 100% detection rates for trisomies. Yet, the chances of injury to the fetus and the consequence of miscarriages deterred both patients and obstetricians from using these techniques and to this date the techniques are used in high-risk pregnancies for the confirmation of positive results from screening tests.

When human genome sequencing was successfully accomplished, experts in the healthcare industry expected that DNA sequencing would mainly focus on genetic diseases in adults. Instead, the impact of sequencing technology has been primarily on prenatal, newborn and PGD tests. In a short span of four years, the different types of maternal serum screening tests and fetal ultrasound tests are getting gradually replaced by the new technology of non-invasive prenatal tests (NIPTs). In the past four years, hundreds and thousands of these new tests have been performed in screening cffDNA from maternal blood.

The objective of this report is to provide an overview of the various technologies being employed for detecting genetic diseases in embryos, fetuses and newborns. The various chapters describe common pregnancy disorders, numerical chromosome disorders and structural chromosome disorders, single gene disorders inherited by fetuses, newborns and embryos. The global prenatal and maternal diagnostic test market is assessed with respect to:

- Fetal Ultrasound
- Prenatal MRI Screening
- Maternal Serum Screening Tests
- Noninvasive Prenatal Tests (NIPTs) using cffDNA
- NIPTs by Product
- Prenatal Diagnostic Invasive Tests
- Newborn Screening for Genetic Diseases
- Newborn Screening Market by Technology
- Preimplantation Screening/Diagnosis (PGS/PGD)

SWOT and merger/acquisition analysis is also performed as is a comprehensive documentation of the legislation pertaining to newborn screening by geography and how clinical programs are implemented in developed and developing markets.

Emerging trends in associated markets are also analysed in order to give the reader a comprehensive overview of how prenatal and maternal diagnostic testing is affected by the following industries:

- Molecular Diagnostics Market
- Liquid Biopsy Market
- Personalized Medicine Diagnostics Market
- In vitro Diagnostics (IVD) Market

1.3 Key Questions Answered in this Report

- What are the major prenatal pregnancy complications?
- What are the major genetic diseases detected in fetuses?
- What are the appropriate technologies for the detection of aneuploidies, microdeletions, duplications, copy number variations and translocations?
- How far is genetic counseling important in educating pregnant women and healthcare professionals?
- What is the impact of recent advances in clinical genomics on genetic counseling?
- What different noninvasive and invasive prenatal screening tests are performed during a pregnancy?
- What are the detection rate, true positive rate and true negative rate for NIPTs?
- Currently, in which countries are the NIPTs available?
- What is the cost of NIPTs region-wise?
- What is the uptake of conventional maternal serum tests, NIPTs and invasive diagnostic tests in the U.S.?
- What is the average cost of maternal serum screening, NIPTs, fetal ultrasound screening and invasive diagnostic tests in the U.S.?
- What are the strategies to be adopted for clinical implementation of NIPTs for all pregnancies?
- What is the "patient directed model" for the integration of NIPTs into healthcare systems?
- How does the detection rate of NIPTs compare with the rates of conventional maternal serum screening tests?
- What are the genetic disorders detectable by different prenatal screening and diagnostic tests?
- What is the reliability of amniocentesis and CVS results?
- Is the future of invasive diagnostic tests uncertain?
- Do the intact fetal cells in maternal blood have use in noninvasive prenatal diagnosis (NIPD)?
- What about the use of trophoblast cells obtained from cervix in NIPD?
- How many genetic conditions are detected during newborn genetic screening in the U.S.?
- What is the status of newborn genetic screening in developed, developing and other countries?
- What are the different preimplantation genetic screening (PGS) and preimplantation genetic diagnosis (PGD)?
- What are the currently available PGS/PGD testing products available in the market?
- What are the currently used advanced technologies in prenatal, newborn and PGD testing?
- What is the global market for fetal ultrasound screening, through 2021?
- Who are the market leaders in fetal ultrasound systems?
- What is the global and regional market for prenatal screening by fetal MRI, through 2021?
- What is the global and regional market for maternal screening tests, through 2021?
- What is the global and regional market for NIPTs using cffDNA, through 2021?
- What is the global and regional market for invasive prenatal diagnostic tests, through 2021?
- What is the global and regional market for newborn screening tests, through 2021?
- What is the global market for newborn screening tests by technology, through 2021?
- What is the global and regional market for pre-implantation genetic diagnosis (PGD), through 2021?
- What is the global market for molecular diagnostics, through 2021?
- What is the global market for liquid biopsy tests, through 2021?

- What is the overall global market for personalized medicine diagnostics, through 2021?
- What is the overall global market for *in vitro* diagnostics (IVD), through 2021?
- What are the recent acquisition activities in prenatal screening industry?
- What are the important factors that drive the growth of prenatal and newborn genetic testing industry?
- What are the challenges faced by the prenatal screening industry?
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