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Non-invasive Prenatal Testing Runs Global

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2023 is the year of the growing preference for early disease detection and prevention, minimally invasive testing methods, and consumerization in diagnostic testing. Non-invasive prenatal testing (NIPT) is a powerful tool empowering clinicians and prospective parents with valuable clinical insights to make informed decisions regarding pregnancy management. With guidelines for NIPT expanding and payers reimbursing for screening procedures, clinically validated virtual care platforms will open the aperture to a larger addressable NIPT market.

Non-invasive prenatal testing (NIPT) requires the evaluation of fetal cells or cell-free DNA obtained from a maternal blood sample during pregnancy. Since its debut in the United States and China/Hong Kong in 2011, the molecular screening test used to detect fetal congenital anomalies is the fastest-spreading prenatal genetic technology globally, accounting for 38 per cent of the prenatal genetic testing market.

Technological innovations shaped by commercial interests

Although Next Generation Sequencing (NGS)-based methods have been majorly used, technology evolutions encompassing qPCR, dPCR, and amplification technologies aim to further drive the quality of tests and reduce costs. The commercial sector is broadening its scope to include sex chromosome aneuploidies, rare autosomal trisomies, and sub-microscopic copy-number variants. Automated in-lab workflows and remote patient monitoring in pregnancy are enabling providers to better manage pregnancies and risks.

A new generation of screening tests is promising to enhance pregnancy management by providing information about placental development acquired in real-time with trophoblast cells, implying a focus on targeted management of at-risk pregnancies. As the technology evolves, debates surrounding the expansion of NIPT beyond chromosomal abnormalities will increase. Hence for test developers, the role of private clinics and biomedical professionals is key to spearheading access, especially in underpenetrated regions. Another strategy central to attracting customers includes validation studies.

While clear guidelines and services to address potential medical and ethical issues are critical for the responsible future use of NIPT, leveraging complementary informatics technologies that enhance data analysis and reporting capabilities will be vital in accelerating new product development, in addition to expanding core intellectual property portfolio and sample preparation technology.

Commercial potential across geographies

With 140 million babies born globally, NIPT represents an exciting opportunity, with many geographies still in the nascent stages of adoption. A large US market offers significant potential. There are about 4 million babies born in the United States each year, and 97 per cent of high-risk pregnancies and 56 per cent of average-risk pregnancies are covered. NIPT is widely adopted throughout Europe, though only a few countries have a national policy that regulates its use. NIPT for all pregnancies is covered in the Netherlands and Belgium as a first-tier screening test. Within Asia-Pacific, the rising number of women postponing pregnancy until later in life across China, Australia, India, and Japan is favouring adoption. Thailand and Vietnam are other markets with sizable populations offering significant opportunities. Expanded clinical applications, such as autosomes and microdeletions, spur innovation in the sector, making it a viable industry in the long term.

Growing competitiveness with substantial new entry

The market is in the growth phase. The dominance of large-scale companies in the industry, including Natera, Illumina, LabCorp, Roche Molecular Systems/BioReference Laboratories, and BGI Genomics, is challenged by new entrants in the field embracing non-PCR, non-NGS-based methods to enable broader adoption, like the Vanadis NIPT system from PerkinElmer. To further reduce implementation costs, companies like Atila Biosystems are harnessing the power of digital PCR to detect targets that indicate possible chromosomal anomalies—trisomy 21 associated with Down Syndrome, trisomy 18 associated with Edwards Syndrome, and trisomy 13 associated with Patau Syndrome and foetal fractions.

As NIPT marches toward becoming the standard screening tool for pregnancies worldwide, companies like Bionano Genomics emerge as game changers, with optical genome mapping (OGM) analysis providing high-throughput, comprehensive follow-up genome analysis in case of a positive NIPT screen or for high-risk pregnancies following an abnormal ultrasound. OGM can be developed as the basis of a new standard of care in prenatal genetic analysis because of its ability to identify all types of structural variants. Other notable new participants encompass Dutch biotech VyCAP, Danish ARCEDI Biotech, and US-based RareCyte, which developed methods for separating circulating trophoblast cells from maternal blood samples. Rapid advancements in novel techniques imply an enriching product matrix for decentralised NIPT.

Growth strategy to address the surge in test volume

2023 is the year of the growing preference for early disease detection and prevention, minimally invasive testing methods, and consumerization in diagnostic testing. The three macro trends imply volume growth and improved test economics for NIPT, driven by growing awareness of preventing chromosomal anomalies such as Downs Syndrome. With guidelines for NIPT expanding and payers reimbursing for screening procedures, clinically validated virtual care platforms that enable obstetrician-gynaecologists to deliver prenatal care offer risk-specific experiences, detect risk more quickly, and automate critical elements of care, driving the adoption of guideline-recommended NIPT. As adoptions surge, workflow improvements via artificial intelligence-based technologies (deep neural networks and probabilistic modelling) learning from millions of processed samples are poised to improve positive predictive value, lower the cost of goods sold, and reduce the no-call rate.

Cell-based NIPTs represent another promising field in research, addressing the challenges associated with whole-fetal-cell isolation from the maternal blood draw. However, evidence will decide the success of this next-generation approach in bringing tests from screening to diagnostics. The path of fetal cells might be long, but the rewards are promising. New technologies must undergo validation for responsible clinical implementation. Increasing public awareness of testing is critical to prepare prospective parents for crucial decision-making. Remote pregnancy monitoring platforms delivering medical-grade fetal wellness checks will empower providers and patients to expand care delivery. Due to the advanced nature of technology, achieving economies of scale is key to incremental growth. Hence, techniques should enable providers to support expectant parents with more accurate, timely, and personalised pregnancy management options.

NIPT is a powerful tool empowering clinicians and prospective parents with valuable clinical insights to make informed decisions regarding pregnancy management. Technological advances and responsible innovation make it a promising screening avenue for the detection of additional genetic conditions. Industry participants carving distribution and marketing agreements to expand access and coverage, in addition to product development and commercialization agreements, will effectively navigate the coming age of distributable kits, offering new solutions to expand testing to all autosomes and microdeletions. Empowering laboratories with computational frameworks for sequencing-based NIPT analysis to enable the characterisation of disease transitions and their reflection in the blood will open the aperture to a larger addressable NIPT market.

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