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ORIGINAL RESEARCH

Non-Invasive Prenatal Testing in the Kingdom of Saudi Arabia: Current Status of Adoption and Roadmap for the Future

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Background: Non-invasive prenatal testing (NIPT) has emerged as a significant advancement in prenatal screening that offers safer and more accurate detection of chromosomal abnormalities compared to conventional methods. The study aimed to evaluate the status of NIPT adoption in the Kingdom of Saudi Arabia (KSA).

Methods: A comprehensive 3-phased study was conducted to examine the status of NIPT in KSA. In Phase I targeted literature review was conducted followed by Phase II and Phase III which involved qualitative interview-based exploration with key stakeholders and round table discussion with key opinion leaders, respectively.

Results: Key stakeholders in KSA underscore NIPT's clinical value and economic benefits while addressing coverage disparities and the push for national guidelines. In KSA, NIPT prescription is influenced by multiple factors such as logistics, personnel, cost, accessibility, policy, and validation. Addressing these factors is important for the widespread adoption of NIPT as a primary screening test. Key opinion leaders suggest that accurate infrastructure, multidisciplinary care, patient education, and expansion of NIPT's scope are crucial. To address current challenges, proactive collaboration of both public and private sectors is essential. NIPT usage has increased in KSA over time. It has now been recommended for all pregnant women, leading to an increase in demand for national guidelines to regulate the practice along with awareness campaigns about the value of testing.

Conclusion: A structured, phased roadmap for implementing NIPT in Saudi Arabia is crucial to ensure cost-effectiveness, cultural and ethical appropriateness, and nationwide access for all pregnant women.

Keywords: non-invasive prenatal testing, NIPT, Kingdom of Saudi Arabia, KSA, prenatal care, Invasive prenatal testing

Key Points

What is already known about this topic?

- The field of prenatal screening has transformed with the emergence of non-invasive prenatal testing (NIPT).
- Although NIPT proves to be beneficial for early screening of aneuploidy, its availability is still limited in the Kingdom of Saudi Arabia (KSA).

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What does this study add?

- This study examines the current state of NIPT adoption in KSA and future prospects.
- Patients' awareness, national guidelines, and accurate infrastructure are crucial for the widespread adoption of NIPT in the KSA.

Introduction

The field of prenatal screening has undergone a transformative evolution with the emergence of non-invasive prenatal testing (NIPT), as it offers a more accurate and safer approach for screening chromosomal abnormalities in the fetus. NIPT is a groundbreaking technique that involves a simple blood draw, which reduces the risk of miscarriage.[1] It is based on the analysis of cell-free fetal (cff) DNA.[2] The fragments circulating in maternal serum are analyzed using next-generation sequencing to detect various congenital anomalies.[3–5]

Globally, NIPT is increasingly integrated into routine prenatal screening programs across high- and middle-income countries, improving pregnancy management and informed decision-making. Its expanding applications now include screening for sex chromosome aneuploidies and microdeletions, reflecting its growing clinical value.[6]

Congenital anomalies are one of the leading causes of neonatal mortality worldwide, accounting for approximately 20% mortality rate among children.[7] The World Health Organization (WHO) reports that nearly 240,000 infants die due to congenital disorders within the first 28 days of their life annually (https://www.who.int/news-room/fact-sheets/detail/birth-defects). Within the Kingdom of Saudi Arabia (KSA), chromosomal abnormalities (aneuploidies) are known to account for approximately 6.7% of recurrent pregnancy losses.[8] The most common aneuploidies among live births are of sex chromosomes and trisomy 13, 18, and 21 which are also known as Patau syndrome, Edward syndrome, and Down syndrome, respectively.

Maternal serum markers and ultrasound imaging are the conventional screening methods used to identify congenital anomalies.[9–11] However, these methods have relatively high false positive rates of 2–7%. Invasive procedures like amniocentesis or chorionic villus sampling (CVS) carry potential risks, such as the risk of maternal infection, bleeding, fluid leakage, and miscarriage.[12]

NIPT demonstrates lower false positives in detecting chromosomal abnormalities as compared to traditional screening methods. NIPT is known to be 99% sensitive and 99.9% specific for trisomy 13, 18, and 21, and for sex chromosome abnormalities. It can be conducted as early as ten weeks of gestation without any risk of miscarriage or harm to the fetus.[13]

NIPT also offers several advantages, including cost-efficiency through reduced invasive procedures, better resource allocation for healthcare, early intervention, improved patient experience, and decreased legal liability, which ultimately leads to improved health outcomes and potential cost savings for the healthcare system.[14] Incorporation of NIPT in the present healthcare system can enhance the efficiency of the prenatal care processes, which could result in improved collaboration among healthcare practitioners, like gynecologists and geneticists, and deliver more holistic care for patients.

Despite its effectiveness in detecting common aneuploidies, NIPT faces several challenges, including inability to identify monogenic disorders, risks of false positives and negatives, lower reliability in twin or mosaic pregnancies, absence of standard protocols, uncertain real-world outcomes and ethical concerns.[15] Country-specific challenges in adopting NIPT include disparities in healthcare infrastructure, where low- and middle-income countries often lack access to advanced diagnostic facilities.[16] Cost and reimbursement issues remain major barriers, as NIPT is often expensive and not covered by public insurance in many regions.[17] Additionally, limited awareness among healthcare providers and patients and a shortage of trained genetic counselors hinder its widespread implementation, especially in rural and resource-limited settings.[18]

Although NIPT proves to be beneficial for early screening of aneuploidy, its availability is still limited in the KSA. Some of the major challenges in NIPT adoption are policy barriers, gaps in infrastructure, lack of screening programs, inconsistency in the identification of high-risk pregnancies, and lack of awareness and education among patients and health care providers.[19,20] Moreover, as people of the KSA follow Islamic laws for pregnancy and abortion, cultural and religious perceptions also play a crucial role in the adoption of prenatal screening programs like NIPT. As per the Islamic religion, abortion is prohibited except in critical conditions.[21]

Despite the clinical benefits of NIPT, there is a lack of comprehensive evidence regarding its adoption, accessibility, and integration into prenatal care pathways within the KSA. Existing studies have largely focused on technical performance or global utilization, leaving significant gaps in understanding the local implementation barriers, patient awareness, policy considerations, and cultural influences on NIPT uptake.[20]

In Saudi Arabia, prenatal screening is currently guided by the Ministry of Health (MoH) through national maternal health programs that include first- and second-trimester ultrasound scans and biochemical testing for high-risk pregnancies.[4] However, there are no standardized national guidelines for the routine use of NIPT, and its availability is largely limited to private tertiary care hospitals and select academic centers.

Thus, understanding these multifaceted challenges is essential to ensure the safe, culturally appropriate, and cost-effective implementation of NIPT in the Saudi context. Nevertheless, in the past few years, KSA has established itself as a frontrunner in furnishing an enhanced and more patient-focused, value-based healthcare system. NIPT has the potential to enhance prenatal care in the KSA[22] but addressing local guidelines, policy, regulatory, and access-related challenges is essential for its safe and effective expanded use.

Therefore, the main objective of this study was to provide a comprehensive analysis of the status of NIPT adoption within the KSA healthcare system, which included a detailed examination of the existing guidelines and the factors responsible for NIPT acceptance within the KSA. Furthermore, the study sought to scrutinize awareness among the patients and the implementation of NIPT, identify the gaps, and recommend strategies to address the current challenges and unmet needs for NIPT adoption in the KSA.

Methods

The study was conducted in three phases, with the primary aim of developing a comprehensive advocacy strategy for the implementation of NIPT in the KSA (Figure 1). The strategy was tailored to meet the specific needs and requirements of the country.

Phase I: Comprehensive Literature Review

To understand the status of NIPT over the last 10 years, a targeted literature review was conducted using the EMBASE and MEDLINE databases. The search included keywords such as prenatal care, high-risk pregnancies, prenatal guidelines, NIPT, prenatal screening, etc. Only studies published in English were selected and evaluated to gather information

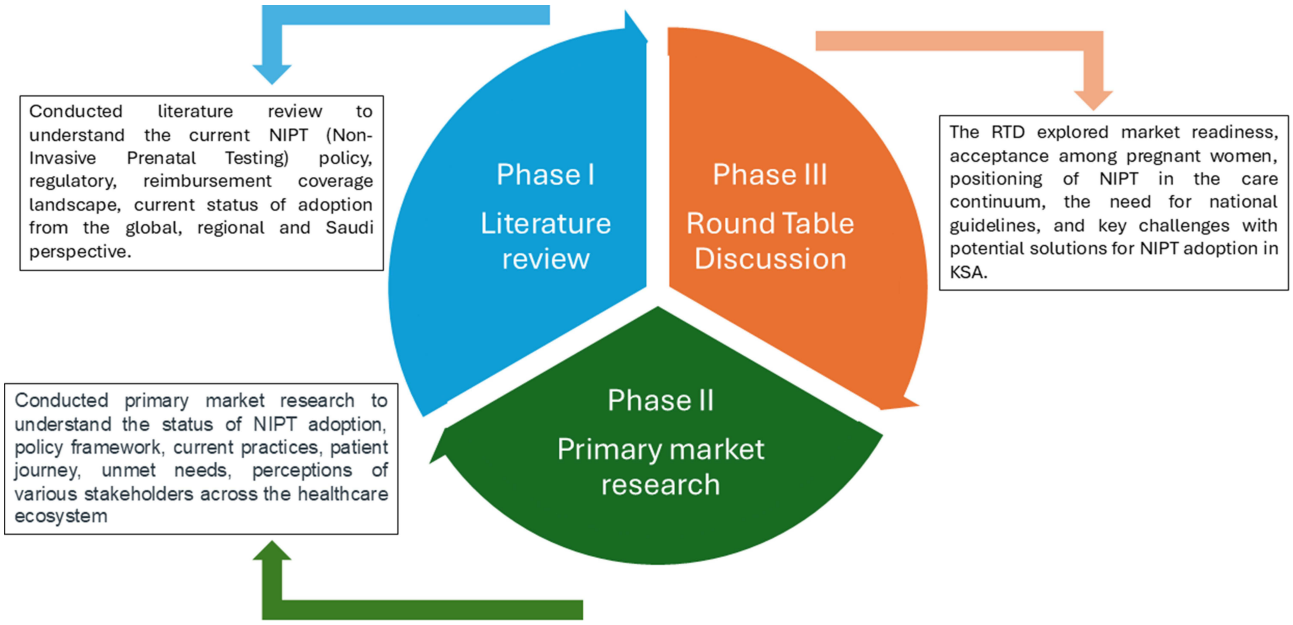

Figure 1 Three-phase qualitative research approach to assess the adoption of Non-Invasive Prenatal Testing (NIPT) in Saudi Arabia.

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about policies, regulations, reimbursement coverage, and the adoption status of NIPT globally and in the KSA. Based on the targeted literature review, specific topics to be discussed and elaborated from the KSA perspective were identified.

Phase II: Primary Market Research

After completing a targeted literature review, the second phase of the study involved qualitative semi-structured interviews which were conducted from July 2023 to September 2023. Various experts and key stakeholders were approached via. telecommunication and asked initial questions to gauge their interest and suitability for the interview. Subsequently, in-depth interviews were conducted face-to-face using a questionnaire to comprehend their perceptions, preferences, and to identify potential misconceptions or reservations about NIPT. The aim was also to understand key unmet needs in prenatal care and gather views regarding private-public partnerships and engagement plans (Supplementary file: NIPT questionnaire). The interviews were recorded and analyzed to draw consensus.

A total of twenty-nine key opinion leaders (KOLs) from public and private settings in KSA were actively engaged and interviewed. The majority were obstetricians/gynecologists or maternal and fetal specialists (59%), followed by geneticists (27%). Some of the KOLs were involved both in medical practice and regulatory activities like regulators, policy officials, or payers (14%) in the heterogenous healthcare ecosystem of the KSA (Table 1).

The overarching goal of this phase was to comprehend the policy framework, current practices, patient journey, unmet needs and challenges, and the perception of various stakeholders across the healthcare system. This was done to validate the existing literature and bridge gaps in the available data.

Phase III: Round Table Discussion

Phase III of this study was a Round Table Discussion (RTD) which was held in October 2023 with a group of 13 KOLs encompassing regulators, HCPs, and health economists. These KOLs were approached based on their expertise and experience in the healthcare industry and their role in the patients' journey during prenatal care. These KOLs were not involved during the primary market research. Among all, some of the KOLs held multiple stakeholder positions. For instance, 3 of the KOLs were both regulators and geneticists, and 2 KOLs were both payers and regulators. The majority of the KOLs were HCPs, including six geneticists, three obstetricians and gynecologists, and one maternal-fetal medicine consultant (Table 1).

Table 1 Profile of Key Opinion Leaders (KOLs) Participating in Phase II and Phase III of the Study

Phase II		
Category	**Number of KOLs**	**Percentage (%)**
Obstetricians / Gynecologists or Maternal & Fetal Specialists	17	59%
Geneticists	8	27%
Dual role (Medical practice + Regulatory / Policy / Payer)	4	14%
Phase III		
Category	**Number of KOLs**	**Percentage (%)**
Regulators	5	38.5%
Payers	2	15.4%
Health Economists	1	7.7%
Geneticists	6	46.2%
Obstetricians / Gynecologists	3	23.1%
Maternal-Fetal Medicine Consultant	1	7.7%

The theme and discussion points for the RTD were derived from insights obtained through targeted literature analysis and primary market research, which offered a comprehensive understanding of the current status of non-invasive prenatal testing (NIPT) practices and guidelines worldwide. The discussion encompassed topics such as unmet needs, challenges, and potential solutions, with the objective of formulating a national ethical and regulatory framework for NIPT. KOLs opinions and suggestions were recorded and summarized.

Results

Phase I: Targeted Literature Review

NIPT Evolution

NIPT is a blood test that was introduced in 2011 for detecting Down's syndrome (T21) with 100% sensitivity and 97.9% specificity.[23] For other chromosomal abnormalities, like Edward syndrome (T18), Patau syndrome (T13), sex chromosomal abnormalities, and autosomal abnormalities, the sensitivity of NIPT is approximately 99% and false positive rates below 1%.[24,25] Initially, NIPT was validated for high-risk pregnancies only, but later it was also validated for the low-risk ones. In 2020, the American College of Obstetricians and Gynecologists (ACOG) updated its guidelines to state that NIPT can be used in all pregnancies.[26] Currently, NIPT is used worldwide as a screening test for the common trisomy 13, 18, and 21, and sex chromosome abnormalities.[27] In the KSA, King Faisal Specialist Hospital and Research Center (KFSH) in Riyadh introduced NIPT in 2013,[20] and recently, King Abdulaziz Medical City, Ministry of National Guard Health Affairs (MNG-HA) in Riyadh, KSA, established a localized NIPT facility for the first time in Saudi Arabia and began operating it in their laboratories.[8]

Policies and Regulations Advocating NIPT

Various guidelines worldwide advocate for screening of fetal chromosomal anomalies. Following are some of the internationally recognized guidelines on NIPT:

In 2015, the United Kingdom (UK) National Screening Committee (NSC) recommended using NIPT as a second-tier test for 13, 18, and 21 trisomy. NIPT, as a second-tier test, is used to confirm the results of first-tier primary screening that has identified any risk of chromosomal abnormality. The recommendation states that women who opt for the initial screening test and receive a higher-chance combined test score of greater than or equal to 1 in 150 in the first or second-trimester quadruple test are eligible for the NIPT offer.[28] (Down's syndrome - UK National Screening Committee (UK NSC) - GOV.UK (view-health-screening-recommendations.service.gov.uk)).

In 2013, the Genetics Committee of the Society of Obstetricians and Gynecologists of Canada (SOGC) recommended offering NIPT to pregnant women identified as being at increased risk for fetal aneuploidies through provincial or territorial screening, positioning it as a second-tier screening test).[29]

For the United States (US) population, the ACOG and the Society for Maternal-Fetal Medicine (SMFM) issued an updated ACOG Practice guideline in 2020. It recommends that all pregnant women be offered prenatal genetic screening and diagnostic testing options, regardless of age or chromosomal abnormality risk. It also advocates the utilization of cell-free DNA screening as a follow-up for patients with a positive serum-analyte screening test results, providing an alternative for those who refrain from a diagnostic test. However, women need to discuss with their healthcare professionals (HCPs) and make an informed decision regarding screening and diagnostic testing. Around 25–50% of pregnant women in the US have been using NIPT as a second-tier test.[28]

Recently, in their practice guidelines, American College of Medical Genetics and Genomics (ACMG) has strongly recommended NIPT over traditional screening methods for all pregnant patients with singleton and twin gestations for fetal trisomy 21, 18, and 13 and strongly recommends NIPT be offered to patients to screen for fetal sex chromosome aneuploidy.[30]

The International Society for Prenatal Diagnosis (ISPD) published its first position statement on the use of NIPT to screen for aneuploidy in 2015. The guidelines suggest cfDNA screening for common autosomal trisomy in the case of twin pregnancies.[31] ISPD has also published a fresh position statement on the use of NIPT in singleton pregnancies in 2023 which states that NIPT is the most accurate screening test for the common autosomal aneuploidies in unselected

singleton populations, and those at known increased probability. It can be offered in primary or contingent screening models with context-specific considerations in local health policy influencing decisions and implementation models.[32]

Reimbursement Coverage Landscape

The implementation of NIPT at the global level also exhibited wide heterogeneity based on reimbursement criteria. In the US, ACOG states that nearly all high-risk pregnancy patients are covered for NIPT, and around 80% of insured patients are covered regardless of their age or risk level. However, private insurance and state Medicaid coverage for NIPT can vary significantly. Some state Medicaid programs do not cover NIPT at all, while others only cover it from specific companies.[17]

NIPT testing has been incorporated into the national policies in less than half of European countries, and its actual use by pregnant women is lower than 25%.[33] Some countries (eg, Belgium and the Netherlands) reimburse NIPT tests to all pregnant women.[34] However, certain countries reserve NIPT for high-risk pregnancies with a threshold ranging from 1:100 to 1:1000, and some countries restrict NIPT to very high-risk pregnancies.[35]

Canada allows provinces and territories to decide whether to include NIPT in their prenatal screening program. NIPT is publicly funded in the provinces of Ontario, British Columbia and Quebec, and the territory of Yukon, and is available across the rest of Canada for an out-of-pocket cost.[28,36]

In Lebanon, NIPT is offered mainly as a second-tier screening test, ordered by a physician based on their preferences over criteria defining high-risk pregnancies. However, the tests are available with no availability of public funding.[28]

Current Status of NIPT Adoption in the World

Globally, physicians showed a broad acceptance of NIPT, and the majority of them are recommending these tests in their practice. A study conducted in 2015 with 49 respondents from 28 different countries (including nine from Asia, three from Australia, 23 from Europe, four from the Middle East, six from North America, and four from South America) confirmed that NIPT is readily available through the private sector. Almost 90% of the respondents confirmed that they recommend NIPT in their practice. However, eligibility criteria for NIPT vary among physicians, with the majority recommending it only for high-risk pregnancies (41 out of 90), while others recommend it for all pregnant women (23 out of 90) and those who can pay for it (21 out of 90). Physicians also noted that patients' ability to pay for the test is a major factor that they consider when recommending NIPT.[37]

According to a study published in 2020, the global uptake of NIPT reached 22.4% in 2018, reflecting a significant rise in its adoption as a prenatal screening tool. Countries such as the US, UK, Canada, Australia, and various European nations have been at the forefront of NIPT implementation[38] (Table 2).

Chromosomal coverage for NIPT also varies across nations.[3,33,39] Among European countries, France, Norway, Austria, Wales, and Finland offer it as a test for chromosomes 13, 18, and 21, and six nations- Belgium, the Netherlands, Lithuania, Greece, Cyprus and Italy- also include microdeletions and or whole-genome coverage in the test.[3,17,33]

Table 2 Status of NIPT Adoption in Different Countries

Country	Birth Rate	NIPT Uptake
United States	12.001 per 1000 people	25–50% of all pregnant women receive NIPT
United Kingdom	11.377 per 1000 people	Around 50% of all pregnant women receive NIPT
Sweden	11.795 per 1000 people	<25% of women receiving NIPT
United Arab Emirates	10.110 per 1000 people	No data available
Kingdom of Saudi Arabia	16.631 per 1000 people	No data available

Phase II: Primary Market Research

Most of the respondents of the questionnaire were involved in high-risk pregnancy care and were devoting over 70% of their time to direct patient care. They also held decision-making roles and had an average of 5–7 years of experience in their respective fields. On average, 50–200 pregnant women, including high-risk pregnancies, visit these specialists each month. Private hospital settings had a higher proportion of pregnant women visits, followed by the MoH.

Most of the respondents showed a strong inclination towards prescribing NIPT for pregnant women with a higher risk of chromosomal abnormalities. There is a unanimous consensus among HCPs on the use of NIPT for high-risk pregnancies in the KSA. However, the criteria for NIPT prescription in high-risk pregnancies showed variations across sectors and experts, with high-risk screen results, maternal age of more than 35 years, and previous pregnancy complications being the most cited factors.

More than 80% of respondents prefer NIPT for pregnant women identified as high-risk for aneuploidies. Majority of the respondents considered maternal age as the primary criterion for prescribing NIPT, followed by other risk factors such as high-risk screening results from other tests like ultrasound, previous pregnancy with aneuploidy, family history. However, 10 of the respondents also suggested that NIPT must be offered to all pregnant women irrespective of the risk (Figure 2). In some cases, patient preference also triggers NIPT prescription by the HCPs in their routine clinical practice. Consanguineous marriages, though not directly related to the chromosomal aneuploidies, are considered by some HCPs as one of the criteria for NIPT prescription. From Figure 2, it can be observed that HCPs or questionnaire responders considered more than one factor while prescribing NIPT during their clinical practice.

From the semi-structured interview results, it was concluded that in the KSA, NIPT is generally viewed as a primary screening test, while invasive prenatal testing (IPT) is often reserved for confirmatory diagnostic testing across different sectors. Confirmation through an IPT is suggested only when other screening methods indicate potential abnormalities. Therefore, NIPT is preferred for high-risk pregnancies by HCPs in KSA due to its precision, performance metrics, early detection of an anomaly, and non-invasive nature. Moreover, experts also confirmed that the ACOG guidelines serve as the primary reference for NIPT recommendations among HCPs in KSA.

Phase III: Round Table Discussion with Key Opinion Leaders

In Phase III, RTD was held in October 2023 with the objective of validating insights from Phases I and II, exploring barriers and opportunities for NIPT adoption in Saudi Arabia, and co-developing recommendations for a national ethical and regulatory framework. A total of 13 KOLs, representing a mix of clinical experts, regulators, and policymakers from diverse institutions were involved in the discussion.

During the RTD, KOLs discussed the following topics and provided their opinions which were then summarized to draw consensus:

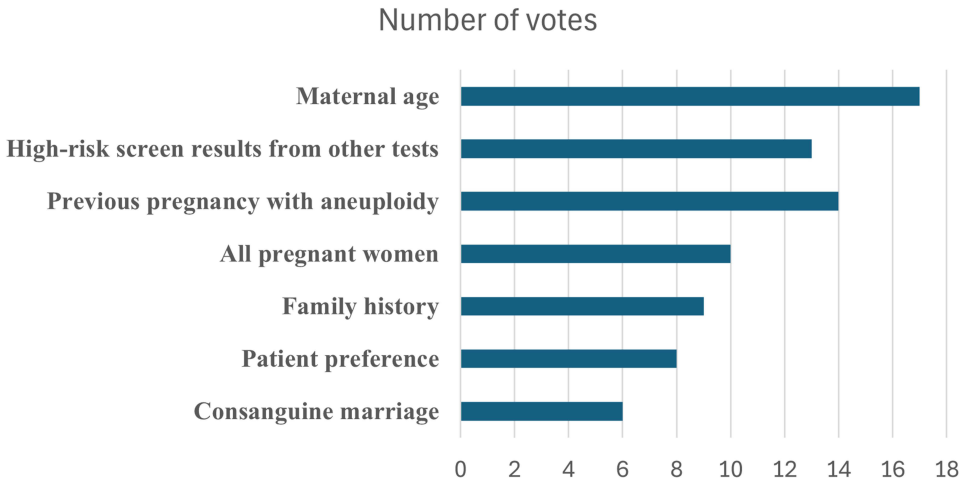

Figure 2 Criteria considered by experts while recommending NIPT.

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Market Readiness for NIPT Adoption in KSA

According to the majority of KOLs, KSA is ready to implement NIPT at a national level. These KOLs have experience in their respective fields and are familiar with the technical requirements necessary for NIPT testing. Most of them are already performing NIPT in their respective facilities.

Ministry of National Guard Health Affairs (MNGHA) covers NIPT for all those who are officially attached to the organization and aims to drive NIPT best practices to the forefront of prenatal care in the KSA through resource pooling and to establish a centre of excellence (COE) hub. The MNGHA also emphasized the cost-effectiveness of the test if the budget impact of having a child with chromosomal abnormality on families and the government is considered in the analysis.

Moreover, KFSH provides NIPT coverage for all its local patients. KFSH and MNGHA are also keen on establishing a COE Hub for the KSA, with the aim to maximize resource pooling and promote a multidisciplinary continuum of care. The members from the MOH stated that they occasionally direct eligible patients to secondary hospitals such as King Fahad Medical City (KFMC), for NIPT testing. The Ministry of Defense (MODA) aims to extend its coverage to include NIPT for high-risk patients in the near future, thereby upgrading their healthcare support as well as services to expectant parents.

However, KOLs have reported facing logistical issues and unavailability of resources such as testing kits, instruments, trained staff, and infrastructure, which hinders the continuation of testing facilities.

Acceptance Among Pregnant Women for NIPT

All the KOLs unanimously expressed that the acceptance and perception of NIPT among pregnant women can be influenced by some misconceptions that need to be clarified through pre-and post-test counseling. It is important to educate patients that NIPT is a screening test and not a confirmatory diagnostic test. One common misconception is that NIPT's primary use is to reveal the gender of the baby, which needs to be changed through appropriate education and counseling.

HCPs, physicians, and private laboratories should be educated about the importance of counseling and effective patient management to practically implement these measures. However, a major challenge observed in NIPT adoption is the lack of multidisciplinary collaboration and patient referral within the healthcare system, which needs to be addressed. Currently, patients are offered NIPT by their HCP, which solely depends on HCP's awareness and expertise. In the majority of cases, patients do not receive any pre- or post-counseling for the test. This leaves the patients in a dilemma, and they generally do not know what will happen following a positive NIPT result.

Positioning of NIPT in Patient's Journey Continuum of Care

According to the KOLs, it is essential to inform patients about available screening tests during their first visit to HCPs. The KOLs also emphasized the significance of obtaining informed consent from patients before conducting NIPT. Some KOLs also suggested that public awareness can be raised by providing information about the test through various communication channels such as brochures, videos, digital resources, and professional training.

KOLs also emphasized the need to implement KSA-specific national guidelines that focus on educating doctors on the importance of counseling and the need to address insurance coverage. Furthermore, they mentioned that private laboratories should be regulated by legislation that obliges them to not accept NIPT test requests without consent which confirms that the pregnant women underwent a pre-test counseling and have a provision for a post-test counseling as well.

Furthermore, if NIPT has indicated a positive result for aneuploidies, it should always be confirmed with an invasive diagnostic test through amniotic fluid, chorionic villi, or blood from umbilical cord.

NIPT Guidelines in KSA

All the KOLs unanimously agreed on the significance of having national guidelines specifically on NIPT. Currently, the majority of the HCPs are following international guidelines set by ACOG. However, having a national or local guideline specific to KSA could be more beneficial as it would help in standardizing the practice in the country and provide equitable benefits to both payers and patients.

Some KOLs also highlighted the need for locally produced health economic evidence to validate the cost-effectiveness of integrating NIPT into prenatal care. The cost-efficiency of NIPT also plays a key role in its adoption. If the test is perceived as expensive or inefficient, it may deter physicians from prescribing it, especially in settings where resources are limited. Therefore, there is a rising need for cost-effectiveness analysis (CEA) and budget impact analysis (BIA) localized to the sector context to position the economic value of NIPT.

KOLs have suggested that the national health insurance policy should cover NIPT, but only if appropriate regulations and guidelines accompany it. Currently, insurance covers the management of pregnant women, therefore, if NIPT meets the necessary criteria, it can also be covered. It is vital to establish strict national guidelines in the KSA to allow their translation into coverage policies enforced by private insurance companies nationally. Table 3 shows key areas that need to be addressed for successful utilization of NIPT in KSA.

KOLs have also expressed concerns about unregulated malpractice like sex determination, unnecessary testing, and abortions and have suggested accurate and specific guidelines for better compliance. For example, implementing informed consent before the test ensures that the patient is provided with all the necessary information beforehand and that both the HCP and patient are aware of each other's intentions and choices. Another recommendation was to have an abortion policy that is specific to trisomy 13, 18, and 21, and for sex chromosome abnormalities. Presently, abortion is prohibited in the KSA except in some cases which are considered critical by the counselors and doctors. However, in some cases, such as due to religious reasons, some of the patients might not want to consider abortion. NIPT is not a mandate, it is an individual's choice. If they do not want to consider abortion even after a positive result, they should not be coerced to undergo it.

The conditions which are considered critical for an abortion are generally defined by ethical committees of the hospitals which are the main approvers of abortions. However, every hospital has its own criteria to define critical conditions in abortion cases.[40] Therefore, it is crucial to address all these matters in the policies and communicate them effectively to the patients.

Other Factors and Challenges Influencing NIPT Adoption in KSA and Their Potential Solutions

During the discussion, KOLs highlighted various factors and challenges that should be addressed to increase the widespread adoption of NIPT in KSA. One of the most important factors discussed was the inefficient logistical process. It is crucial to streamline and ensure that NIPT is conducted within the vital gestation period, which starts from ten weeks of gestation. However, logistic delays or issues can result in missed opportunities or inaccurate results, which could impact both physicians' and patients' confidence.

To bring efficiency, the expertise of the staff involved in the administration and interpretation of NIPT is also absolutely crucial. The absence of trained experts can jeopardize the reliability and effectiveness of the test. This implies that for NIPT to be widely accepted and prescribed in the KSA, there should be a robust system of training and certification for personnel. Digital applications have the potential to revolutionize multidisciplinary care by providing

Table 3 Three Key Areas That Needs to Be Addressed for Successful Utilization of NIPT in KSA

Key Area	Description
Informed decision	A thorough understanding and informed consent are crucial for patients undergoing NIPT. Pre- and post-test counseling ensures patients are equipped to make informed decisions.
National guidelines	The regulatory system in Saudi Arabia highlights the necessity of a national policy to facilitate reimbursement by both the public and private sectors
Accuracy	The accuracy of NIPT testing largely depends on infrastructure and logistics that need to be upgraded regularly to meet demand.

Abbreviations: NIPT; Non invasive prenatal testing; MoH, Ministry of Health; KSA; Kingdom of Saudi Arabia; CVS, Chorionic Villus Sampling; ACOG, American College of Obstetricians and Gynecologists; KFSH, King Faisal Specialist Hospital and Research Center; NSC, National Screening Committee; SOGC, Society of Obstetricians and Gynecologists of Canada; SMFM, Society for Maternal-Fetal Medicine; HCPs, Healthcare Professionals; ACMG, American College of Medical Genetics and Genomics; ISPD, International Society for Prenatal Diagnosis; RTD, Round Table Discussion; KOL, Key Opinion Leader; MNGHA, Ministry of National Guard-Health Affairs; COE, Centre of Excellence; KFMC, King Fahad Medical City; MODA, Ministry of Defence; CEA, Cost-Effectiveness Analysis; BIA, Budget Impact Analysis.

Arabic information and counseling contacts to patients. Furthermore, offering training to general practitioners in rural areas ensures that best practices are implemented early during pregnancy.

Apart from the above consensus, certain differences in opinion emerged regarding the scope and pace of NIPT rollout with some experts advocating for immediate universal screening to maximize early detection, while others recommended a phased approach starting with high-risk pregnancies to ensure cost-effectiveness and system readiness before wider implementation.

Discussion

NIPT screening is used for the screening of aneuploidies, and due to its non-invasive nature and performance metrics, it is considered by HCPs and patients as an attractive alternative to traditional screening tests, which are less accurate. Thereby, NIPT can reduce the number of risky procedures like amniocentesis and CVS.[4,25,38]

NIPT has the potential to improve decision-making processes and management choices, thereby delivering clinical benefits to patients and economic advantages to the healthcare system. NIPT is globally acknowledged for its accuracy and, therefore, adopted by global healthcare systems.[41–43]

This study discusses the efforts initiated by KSA to develop the NIPT framework, investigate its current use, and discuss prospects for its adoption in the national healthcare system. For NIPT implementation, it is crucial to finalize the policy to prevent malpractice, foster infrastructure development, and assure reimbursement at a sub-national level to enhance healthcare standards and patient support.

It is essential for individuals considering undergoing NIPT to evaluate the advantages and limitations of the test by discussing it with their HCPs to make an informed decision. Although NIPT has a plethora of advantages, there are certain challenges associated with this test that must be considered while opting for the test. NIPT can also yield false positive and false negative results, and it is more expensive than traditional screening tests. Another factor that could lead to misuse of NIPT is the gender revelation that could potentially lead to selective abortions, thereby affecting gender neutrality. It is imperative to educate HCPs and the general public that NIPT is a screening test for various chromosomal abnormalities, including Down syndrome (Trisomy 21), Patau syndrome (Trisomy 13), Edward syndrome (Trisomy 18), and sex chromosomal abnormalities. Depending on the technology utilized, it may not detect rare genetic disorders like, point mutations, structural rearrangement and mosaic.[44]

A combination of factors like logistics, personnel, cost, accessibility, policy, and validation of the test influences NIPT prescription. Effectively addressing these attributes is crucial for the widespread adoption and trust in NIPT. Experts emphasize the need for a centralized infrastructure, a multidisciplinary continuum of care, patient education, and expansion of the scope of NIPT to meet the specific needs of the local population. Increasing awareness about NIPT using numerous tools such as educational brochures, media campaigns, and interactive apps conjugated with the empowerment of HCPs through appropriate training and collaborations ensures thorough patient comprehension and consistent counseling. Strategic public-private partnership is essential for providing significant opportunities for advancement in NIPT. To address current challenges and ensure the integration of both sectors, proactive collaboration between both sectors is the need of the hour.

This study has certain limitations. Phase I relied on secondary data obtained from published literature which may not fully reflect real-world practice variations in NIPT implementation within KSA. Phase II included a limited number of respondents, primarily from tertiary and private healthcare facilities, which may not represent the perspectives of all healthcare providers across the KSA. In phase III, findings relied on expert opinion, which may introduce subjective bias and may not represent the views of all stakeholders. Furthermore, lack of direct patient perspectives would have restricted the understanding of awareness, acceptability, and cultural factors surrounding NIPT in the real-world scenario.

Conclusion

In recent years, the KSA has seen an increase in the usage of NIPT. This is due to the advancements, the cost drop in NGS, and the growing awareness of NIPT, which has led to pregnant women opting for this method as it is considered an accurate way to screen for chromosomal abnormalities in the fetus. NIPT offers a highly accurate, non-invasive screening option for common chromosomal abnormalities such as trisomy 21, 18, and 13, as well as sex chromosome aneuploidies;

however, it does not reliably detect all genetic disorders, such as point mutations, structural rearrangements, or rare chromosomal abnormalities.

Healthcare providers in the KSA also acknowledge NIPT as a reliable option and recommend it to be available for all pregnant women irrespective of the risk, further contributing to the acceptance of the procedure among the local population.

A structured roadmap for implementing a Managed Entry Agreement (MEA) for NIPT in Saudi Arabia is essential and should begin with forming a national steering committee to align on objectives, address cultural, ethical, and regulatory needs, and draft national guidelines defining eligibility (initially high-risk pregnancies), counseling protocols, and reimbursement mechanisms. A pilot phase in tertiary hospitals should establish infrastructure, train HCPs, set up a national registry, and test financial or outcome-based MEA models, followed by phased nationwide rollout integrating NIPT into existing prenatal screening programs, expanding coverage (including rural areas), and ensuring pre- and post-test counseling. Continuous monitoring of uptake, cost-effectiveness, and patient outcomes will inform iterative policy updates, with public education campaigns and ethical governance ensuring equitable, culturally appropriate access.

Data Sharing Statement

Data sharing is not applicable to this article as no datasets were generated or analyzed during the current study.

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References

1. Horn R. NIPT and the concerns regarding routinisation. *Eur J Hum Genet*. 2022;30(6):637–638. doi:10.1038/s41431-022-01053-6
2. Alyafee Y, Al Tuwaijri A, Umair M, et al. Non-invasive prenatal testing for autosomal recessive disorders: a new promising approach. *Front Genet*. 2022;13:1047474. doi:10.3389/fgene.2022.1047474
3. Alberry MS, Aziz E, Ahmed SR, Abdel-Fattah S. Non invasive prenatal testing (NIPT) for common aneuploidies and beyond. *Eur J Obstet Gynecol Reprod Biol*. 2021;258:424–429. doi:10.1016/j.ejogrb.2021.01.008
4. Jayashankar SS, Nasaruddin ML, Hassan MF, et al. Non-Invasive Prenatal Testing (NIPT): reliability, challenges, and future directions. *Diagnostics*. 2023;13(15):2570. doi:10.3390/diagnostics13152570
5. Pös O, Budiš J, Szemes T. Recent trends in prenatal genetic screening and testing. *F1000Res*. 2019;8:F1000FacultyRev–764. doi:10.12688/f1000research.16837.1
6. Lu X, Wang C, Sun Y, Tang J, Tong K, Zhu J. Noninvasive prenatal testing for assessing foetal sex chromosome aneuploidy: a retrospective study of 45,773 cases. *Mol Cytogenet*. 2021;14(1):1. doi:10.1186/s13039-020-00521-2
7. Zolfizadeh F, Ghorbani M, Soltani M, et al. Factors associated with infant mortality due to congenital anomalies: a population-based case-control study. *Iran J Public Health*. 2022;51(5):1118–1124. doi:10.18502/ijph.v51i5.9411
8. Alyafee Y, Al Tuwaijri A, Alam Q, et al. Next generation sequencing based Non-invasive Prenatal Testing (NIPT): first report from Saudi Arabia. *Front Genet*. 2021;12:630787. doi:10.3389/fgene.2021.630787
9. Cuckle H. Prenatal screening using maternal markers. *J Clin Med*. 2014;3(2):504–520. doi:10.3390/jcm3020504
10. Muller F, Bussières L. Maternal serum markers for fetal trisomy 21 screening. *Eur J Obstet Gynecol Reprod Biol*. 1996;65(1):3–6. doi:10.1016/0028-2243(95)02293-2
11. Yoshizato T, Kozuma Y, Horinouchi T, Shinagawa T, Yokomine M, Ushijima K. Diagnosis of fetal abnormalities during the first trimester. *Kurume Med J*. 2021;66(2):85–92. doi:10.2739/kurumemedj.MS662002
12. Georgsson S, Sahlin E, Iwarsson M, Nordenskjöld M, Gustavsson P, Iwarsson E. Knowledge and attitudes regarding Non-Invasive Prenatal Testing (NIPT) and preferences for risk information among high school students in Sweden. *J Genet Couns*. 2017;26(3):447–454. doi:10.1007/s10897-016-9997-y
13. Health Quality Ontario. Noninvasive prenatal testing for trisomies 21, 18, and 13, sex chromosome aneuploidies, and microdeletions: a health technology assessment. *Ont Health Technol Assess Ser*. 2019;19(4):1–166.
14. Andari MVC, Bussamra SLC, Tedesco TGD, et al. Noninvasive prenatal testing: benefits and limitations of the available tests. *Ceska Gynekol*. 2020;85(1):41–48.

15. Lutgendorf MA, Stoll KA, Knutzen DM, Foglia LM. Noninvasive prenatal testing: limitations and unanswered questions. *Genet Med*. 2014;16 (4):281–285. doi:10.1038/gim.2013.126
16. Labuschagne R, Aldous C, Vorster E, Walters S. Scoping review: the current landscape of NIPT in South Africa. *J Community Genet*. 2025;16 (3):227–241. doi:10.1007/s12687-025-00802-6
17. Allyse M, Minear MA, Berson E, et al. Non-invasive prenatal testing: a review of international implementation and challenges. *Int J Womens Health*. 2015;7:113–126. doi:10.2147/IJWH.S67124
18. Emmet M, Stein Q, Thorpe E, Campion M. Experiences of genetic counselors practicing in rural areas. *Journal of Genetic Counseling*. 2018;27 (1):140–154. doi:10.1007/s10897-017-0131-6
19. Akiel MA, Mohamud MS, Masuadi EM, Alamri HS. Knowledge and attitude of pregnant women in the Kingdom of Saudi Arabia toward noninvasive prenatal testing: a single center study. *Mol Genet Genomic Med*. 2022;10(7):e1960. doi:10.1002/mgg3.1960
20. Bahkali NM, Eissa GA, Shaheen AMB, Sanedi AM, Bahkali DM. Knowledge and attitude regarding Non-Invasive Prenatal Testing (NIPT) among women: a cross-sectional study in Saudi Arabia. *Clin Exp Obstet Gynecol*. 2023;50(10):220. doi:10.31083/j.ceog5010220
21. AbdulAzeez S, Al Qahtani NH, Almandil NB, et al. Genetic disorder prenatal diagnosis and pregnancy termination practices among high consanguinity population, Saudi Arabia. *Sci Rep*. 2019;9(1):17248. doi:10.1038/s41598-019-53655-8
22. Malik SD, Al-Shafai M, Abdallah AM. The special features of prenatal and preimplantation genetic Counseling in Arab Countries. *Genes*. 2022;13 (2):167. doi:10.3390/genes13020167
23. Chiu RWK, Akolekar R, Zheng YWL, et al. Non-invasive prenatal assessment of trisomy 21 by multiplexed maternal plasma DNA sequencing: large scale validity study. *BMJ*. 2011:342:c7401. doi:10.1136/bmj.c7401
24. Dai R, Yu Y, Zhang H, et al. Analysis of 17,428 pregnant women undergoing non-invasive prenatal testing for fetal chromosome in Northeast China. *Medicine*. 2021;100(6):e24740. doi:10.1097/MD.0000000000024740
25. Zhang H, Gao Y, Jiang F, et al. Non-invasive prenatal testing for trisomies 21, 18 and 13: clinical experience from 146,958 pregnancies. *Ultrasound Obstet Gynecol*. 2015;45(5):530–538. doi:10.1002/uog.14792
26. American College of Obstetricians and Gynecologists'. Committee on practice bulletins—obstetrics, committee on genetics, society for maternal-fetal medicine. screening for fetal chromosomal abnormalities: ACOG practice bulletin, number 226. *Obstet Gynecol*. 2020;136(4):e48–e69. doi10.1097/AOG.0000000000004084
27. Steffensen EH, Skakkebæk A, Gadsbøll K, et al. Inclusion of sex chromosomes in noninvasive prenatal testing in Asia, Australia, Europe and the USA: a survey study. *Prenatal Diagnosis*. 2023;43(2):144–155. doi:10.1002/pd.6322
28. Ravitsky V, Roy MC, Haidar H, et al. The emergence and global spread of noninvasive prenatal testing. *Annu Rev Genom Hum Genet*. 2021;22 (1):309–338. doi:10.1146/annurev-genom-083118-015053
29. Birko S, Ravitsky V, Dupras C, et al. The value of non-invasive prenatal testing: preferences of Canadian pregnant women, their partners, and health professionals regarding NIPT use and access. *BMC Pregnancy Childbirth*. 2019;19(1):22. doi:10.1186/s12884-018-2153-y
30. Dungan JS, Klugman S, Darilek S, et al. Noninvasive prenatal screening (NIPS) for fetal chromosome abnormalities in a general-risk population: an evidence-based clinical guideline of the American College of Medical Genetics and Genomics (ACMG). *Genet Med*. 2023;25(2):100336. doi:10.1016/j.gim.2022.11.004
31. Palomaki GE, Chiu RWK, Pertile MD, et al. International society for prenatal diagnosis position statement: cell free (cf) DNA screening for down syndrome in multiple pregnancies. *Prenatal Diagnosis*. 2021;41(10):1222–1232. doi:10.1002/pd.5832
32. Hui L, Ellis K, Mayen D, et al. Position statement from the international society for prenatal diagnosis on the use of non-invasive prenatal testing for the detection of fetal chromosomal conditions in singleton pregnancies. *Prenatal Diagnosis*. 2023;43(7):814–828. doi:10.1002/pd.6357
33. Gadsbøll K, Petersen OB, Gatinois V, et al. Current use of noninvasive prenatal testing in Europe, Australia and the USA: a graphical presentation. *Acta Obstet Gynecol Scand*. 2020;99(6):722–730. doi:10.1111/aogs.13841
34. Lannoo L, Van Der Meij KRM, Bekker MN, et al. A cross-country comparison of pregnant women's decision-making and perspectives when opting for non-invasive prenatal testing in the Netherlands and Belgium. *Prenatal Diagnosis*. 2023;43(3):294–303. doi:10.1002/pd.6329
35. Sebire E, Rodrigo CH, Bhattacharya S, Black M, Wood R, Vieira R. The implementation and impact of non-invasive prenatal testing (NIPT) for Down's syndrome into antenatal screening programmes: a systematic review and meta-analysis. *PLoS One*. 2024;19(5):e0298643. doi:10.1371/journal.pone.0298643
36. Haidar H, Birko S, Laberge AM, Le Clerc-Blain J, Ravitsky V. Views of Canadian healthcare professionals on the future uses of non-invasive prenatal testing: a mixed method study. *Eur J Hum Genet*. 2022;30(11):1269–1275. doi:10.1038/s41431-022-01151-5
37. Minear MA, Lewis C, Pradhan S, Chandrasekharan S. Global perspectives on clinical adoption of NIPT. *Prenatal Diagnosis*. 2015;35(10):959–967. doi:10.1002/pd.4637
38. Neofytou M. Predicting fetoplacental mosaicism during cfDNA-based NIPT. *Curr Opin Obstet Gynecol*. 2020;32(2):152–158. doi:10.1097/GCO.0000000000000610
39. Dondorp W, de Wert G, Bombard Y, et al. Non-invasive prenatal testing for aneuploidy and beyond: challenges of responsible innovation in prenatal screening. *Eur J Hum Genet*. 2015;23(11):1592. doi:10.1038/ejhg.2015.109
40. Alharbi RH, Alajmani L, Alhajrasi RK, Hindi MO, Alsaywid BS, Lytras MD. Perception of the Saudi population on abortion decisions in congenital fetal anomalies. *Cureus*. 2022. doi:10.7759/cureus.32587
41. Ye X, Lin S, Song X, et al. Identification of copy number variants by NGS-based NIPT at low sequencing depth. *Eur J Obstet Gynecol Reprod Biol*. 2021;256:297–301. doi:10.1016/j.ejogrb.2020.11.026
42. Qi QG, Tuo Y, Liu LX, Yu CX, Wu AN. Amniocentesis and Next Generation Sequencing (NGS)-based noninvasive prenatal DNA Testing (NIPT) for prenatal diagnosis of fetal chromosomal disorders. *Int J Gen Med*. 2021;14:1811–1817. doi:10.2147/IJGM.S297585
43. Koumbaris G, Achilleos A, Nicolaou M, et al. Targeted capture enrichment followed by NGS: development and validation of a single comprehensive NIPT for chromosomal aneuploidies, microdeletion syndromes and monogenic diseases. *Mol Cytogenet*. 2019;12:48. doi:10.1186/s13039-019-0459-8
44. Chen YP, He ZQ, Shi Y, et al. Not all chromosome aberrations can be detected by NIPT in women at advanced maternal age: a multicenter retrospective study. *Clin Chim Acta*. 2018;486:232–236. doi:10.1016/j.cca.2018.08.018