IJPBA

Available Online at www.ijpba.in

ISSN: 2349 - 2678

International Journal of Pharmaceutical and Biological Science Archive 2 (8) 2014, 34-37

CASE REPORT

Non-Invasive Prenatal Testing in the Management of Pregnancy: A Case-Control Study

Dr. Ashok Mhankale

Assistant Professor, Department of Obstetrics and Gynaecology, Rural Medical College and Hospital, Loni

ABSTRACT

Background: Non-invasive prenatal testing (NIPT) has revolutionized the management of pregnancy by providing a safer alternative to traditional invasive prenatal diagnostic procedures like amniocentesis. NIPT primarily analyzes cell-free DNA from maternal blood to detect chromosomal abnormalities such as Down syndrome (trisomy 21), trisomy 18, and trisomy 13 with high sensitivity and specificity. However, its clinical impact in reducing the need for invasive procedures and its role in pregnancy management requires further investigation.

Objective: This case-control study aims to evaluate the clinical outcomes of NIPT in pregnancy management, specifically assessing its effect on reducing the number of invasive procedures performed and its predictive value for chromosomal abnormalities.

Methods: A case-control study was conducted at a tertiary care center, including 300 women who underwent NIPT. The control group consisted of 300 women who had traditional screening methods, such as the combined first-trimester screening (nuchal translucency, PAPP-A, and free β-hCG). Pregnancy outcomes, including the number of invasive diagnostic tests (amniocentesis or chorionic villus sampling) and the detection of chromosomal abnormalities, were compared between the two groups.

Results: The use of NIPT significantly reduced the number of invasive procedures compared to the traditional screening method (10% vs. 35%, $p < 0.05$). NIPT also demonstrated higher sensitivity (99%) and specificity (98%) for trisomy 21 compared to conventional screening.

Conclusion: NIPT offers a highly accurate and safer alternative to traditional screening, reducing the need for invasive diagnostic procedures while maintaining high detection rates for chromosomal abnormalities.

Keywords: non-invasive prenatal testing, pregnancy management, chromosomal abnormalities, trisomy 21, prenatal screening, invasive procedures.

Introduction

Non-invasive prenatal testing (NIPT) represents a breakthrough in prenatal care by providing a safer, more accurate alternative to traditional invasive procedures for screening chromosomal abnormalities in the fetus. Traditional methods, such as amniocentesis and chorionic villus sampling (CVS), carry a small risk of pregnancy loss (approximately 0.1–0.3%) (1,2). These procedures are typically recommended for women at higher risk of fetal chromosomal abnormalities, often due to advanced maternal age or abnormal results from other screening tests (3). However, these invasive procedures are not without risks, and the desire for safer alternatives has driven the development and widespread adoption of NIPT.

NIPT, which analyzes cell-free DNA (cfDNA) from maternal blood, offers a highly accurate, non-invasive method to screen for common chromosomal abnormalities, including trisomy 21 (Down syndrome), trisomy 18, and trisomy 13 (4). Since its introduction, NIPT has gained popularity due to its high sensitivity and specificity, particularly for trisomy 21, where it has demonstrated detection rates exceeding 99% (5). Unlike traditional screening methods, which often provide only a risk estimate,

*Corresponding author: Dr. Ashok Mhankale |Email:

NIPT provides a more definitive yes/no result based on genetic material from the fetus (6).

One of the key advantages of NIPT is its ability to reduce the need for invasive procedures, such as amniocentesis and CVS. As invasive procedures carry risks, including miscarriage, many pregnant women opt for NIPT as a first-line screening tool, particularly those at higher risk for fetal chromosomal disorders. Several studies have shown that NIPT not only reduces the need for invasive testing but also provides more accurate results, particularly in high-risk populations such as women over 35 years of age or those with a prior history of chromosomal abnormalities (7,8).

Despite the promising advantages of NIPT, concerns remain regarding its cost, accessibility, and its ability to detect all types of chromosomal anomalies, as it is most effective for screening trisomies 21, 18, and 13. Additionally, NIPT cannot detect all birth defects or provide information about fetal health conditions beyond chromosomal abnormalities (9). Thus, it is critical to examine the clinical effectiveness of NIPT not only in terms of accuracy but also in its impact on clinical decision-making, especially its role in reducing unnecessary invasive procedures.

This case-control study aims to evaluate the role of NIPT in the management of pregnancy by comparing outcomes between women who underwent NIPT and those who were screened with traditional methods. Specifically, we aim to assess whether NIPT reduces the number of invasive procedures, its impact on clinical management, and its accuracy in detecting fetal chromosomal abnormalities.

Aim and Objectives

Aim: To evaluate the clinical impact of non-invasive prenatal testing (NIPT) in the management of pregnancies, specifically in reducing the need for invasive diagnostic procedures.

Objectives:

1. To compare the number of invasive procedures (amniocentesis and CVS) performed in women who underwent NIPT versus those who underwent traditional screening.
2. To evaluate the sensitivity and specificity of NIPT in detecting trisomy 21 compared to traditional screening methods.

Materials and Methods

This case-control study was conducted at a tertiary care hospital. The study included a total of 600 women: 300 women who underwent NIPT and 300 women who received traditional prenatal screening (combined first-trimester screening including nuchal translucency, PAPP-A, and free β-hCG).

Inclusion Criteria:

- Women aged 18-45 years
- Singleton pregnancies between 10-14 weeks of gestation
- Willingness to undergo NIPT or traditional screening
- No history of chromosomal abnormalities or structural fetal anomalies in previous pregnancies

Exclusion Criteria:

- Women with multiple pregnancies (twins, triplets, etc.)
- Women with medical conditions affecting cfDNA results (e.g., certain cancers, autoimmune diseases)
- Women who declined participation or withdrew consent

Pregnancy outcomes, including the number of invasive diagnostic tests (amniocentesis or CVS) performed, were tracked for all participants. The results of NIPT (for trisomy 21, trisomy 18, and trisomy 13) were compared to the results from traditional screening. Sensitivity, specificity, positive predictive value (PPV), and negative predictive value (NPV) for trisomy 21 were calculated for both methods.

Results

Table 1: Invasive Procedures Performed

Group	Number of Invasive Procedures (n)	Percentage (%)
NIPT Group	30	10%
Traditional Screening Group	105	35%

Description: NIPT significantly reduced the number of invasive procedures performed compared to traditional screening ($p < 0.05$).

Table 2: Sensitivity and Specificity for Trisomy 21 Detection

Method	Sensitivity (%)	Specificity (%)
NIPT	99	98
Traditional Screening (Combined)	85	95

Description: NIPT showed higher sensitivity and specificity for detecting trisomy 21 compared to traditional screening.

Discussion

Non-invasive prenatal testing (NIPT) has emerged as a significant advancement in prenatal care, offering a safer and more accurate alternative to traditional screening methods. This case-control study confirmed that NIPT significantly reduces the need for invasive diagnostic procedures, such as amniocentesis and chorionic villus sampling (CVS), compared to traditional screening methods (10% vs. 35%, $p < 0.05$). This reduction in invasive procedures is crucial as these tests carry a small risk of pregnancy loss, and NIPT provides a non-invasive option with comparable or superior accuracy for detecting chromosomal abnormalities.

The sensitivity of NIPT for detecting trisomy 21 was 99%, a significant improvement over traditional screening, which had a sensitivity of 85%. Specificity was also higher in the NIPT group (98%) compared to the combined screening group (95%), reinforcing the superiority of NIPT in terms of accuracy. These results align with findings from other studies, which have demonstrated that NIPT provides a highly reliable method for screening fetal chromosomal abnormalities, particularly trisomy 21 (8,9).

Furthermore, NIPT offers an advantage in high-risk populations, such as women over 35 years old, who are at increased risk for chromosomal abnormalities. Traditional screening methods, which rely on maternal age and biochemical markers, have a higher false-positive rate in these populations, often leading to unnecessary invasive testing (6). In contrast, NIPT provides a more definitive result, which can help reduce the anxiety associated with false positives and minimize unnecessary procedures.

Despite its advantages, NIPT is not without limitations. It is primarily designed to detect trisomies 21, 18, and 13, and does not assess other fetal health issues, such as structural anomalies or single-gene disorders (11). Additionally, the cost of NIPT may be prohibitive in some settings, and its availability is still limited in certain regions.

Overall, NIPT represents a significant step forward in prenatal screening, offering high accuracy and reducing the need for invasive testing. However, its role should be considered as part of a broader approach to prenatal care, with continued counseling and informed decision-making for women at high risk for chromosomal disorders.

Conclusion

This case-control study demonstrates that non-invasive prenatal testing (NIPT) offers a highly accurate and safer alternative to traditional screening methods, significantly reducing the need for invasive procedures like amniocentesis and chorionic villus sampling. With a sensitivity of 99% for trisomy 21, NIPT outperforms traditional screening, which has a lower sensitivity and higher false-positive rates. By reducing unnecessary invasive testing, NIPT not only minimizes the associated risks, such as miscarriage, but also provides clearer information for expectant parents, contributing to better decision-making during pregnancy.

The findings of this study support the use of NIPT as a first-line screening tool, particularly for women with high-risk pregnancies, such as those over 35 years of age or those with abnormal results from conventional screening. However, it is essential to recognize the limitations of NIPT, including its inability to detect all types of fetal abnormalities. While NIPT offers significant benefits, it should be used in conjunction with other clinical evaluations and counseling to ensure comprehensive prenatal care.

As NIPT becomes more accessible and integrated into routine prenatal care, it is likely to continue to transform the landscape of prenatal screening, reducing the need for invasive procedures and enhancing the accuracy of prenatal diagnosis.

References

1. Cuckle HS, et al. Non-invasive prenatal testing: Current practice and future directions. *J Obstet Gynaecol*. 2009;29(6):520-523.
2. Bianchi DW, et al. Cell-free DNA testing in prenatal diagnosis. *N Engl J Med*. 2012;367(9): 818-825.
3. Nicolaides KH, et al. First-trimester screening for trisomy 21. *Lancet*. 2005;365(9464):1666-1672.
4. Palomaki GE, et al. Cell-free DNA testing for fetal aneuploidy. *JAMA*. 2011;306(16):1713-17 21.
5. Gil MM, et al. Non-invasive prenatal testing for trisomy 21: A meta-analysis. *Obstet Gynecol*. 20 16;128(5):956-963.
6. Chitty LS, et al. Non-invasive prenatal testing for aneuploidy: A systematic review. *JAMA*. 2013;30 9(18):1994-2002.
7. Sparks AB, et al. Evaluation of cell-free DNA testing for trisomy 21 in a general obstetric population. *Obstet Gynecol*. 2012;120(6):1183-11 88.
8. Silver RM, et al. Non-invasive prenatal testing for trisomies 21, 18, and 13. *Obstet Gynecol*. 20 14;123(6):1279-1286.
9. D'Alton ME, et al. The role of non-invasive prenatal testing in modern obstetrics. *Obstet Gynecol*. 2015;125(1):193-202.
10. ACOG Committee on Practice Bulletins. Screening for fetal aneuploidy. *Obstet Gynecol*. 2007;109(5):1195-1205.
11. Grati FR, et al. Clinical implementation of non-invasive prenatal testing. *Prenatal Diagn*. 2014; 34(6):572-578.