Original Article

Evaluation of Non-Invasive Prenatal Testing and Early Ultrasound Findings in Fetuses with Raised Nuchal Translucency

Mugheera Hussain, Maryum Noor Malik, Shazia Nayyar, Iqra Tahir, Atika Shahzadi, Jawairiah Liaqat

ABSTRACT

Objective: To assess the association between non-invasive prenatal testing (NIPT) results and first-trimester ultrasound findings in pregnancies with increased nuchal translucency (NT) and their ability to predict adverse fetal outcomes.

Methodology: A retrospective analytical cohort study was carried out at the Combined Military Hospital (CMH), Kharian, Pakistan, between January 2022 and December 2025. The study included 180 singleton pregnancies showing an NT measurement of 3.0 mm or more on first-trimester ultrasound. Findings from NIPT were compared with ultrasound results and verified through invasive diagnostic procedures or postnatal outcomes. Diagnostic accuracy was assessed by calculating sensitivity, specificity, predictive values, and the area under the ROC curve (AUC).

Results: The mean maternal age was 31.2±4.6 years, and the mean gestational age at ultrasound was 12.4±0.6 weeks. Among 180 pregnant women, 54 (30%) had adverse pregnancy outcomes, including chromosomal abnormalities, major structural anomalies, or pregnancy loss. NIPT demonstrated a sensitivity of 95.4%, specificity of 98.6%, and area under the curve (AUC) of 0.97 for detecting aneuploidy. First-trimester ultrasound alone showed an AUC of 0.83, while the combined approach achieved an AUC of 0.99, yielding superior diagnostic accuracy.

Conclusion: Increased NT remains a critical early marker of adverse fetal outcomes. NIPT provides excellent detection of common aneuploidies; however, combining it with detailed first-trimester ultrasound significantly enhances diagnostic and prognostic accuracy.

KEYWORDS: Aneuploidy; Chromosome Disorders; Genetic Testing; Nuchal Translucency Measurement; Prenatal Diagnosis; Ultrasonography.

INTRODUCTION

Nuchal translucency (NT) measured by first-trimester ultrasound is a well-established marker for chromosomal aneuploidy and a wide spectrum of structural and genetic disorders; increasing NT thickness correlates with higher risk of adverse perinatal outcomes.[1,2] An increased nuchal translucency (NT) measurement in the first trimester often raises concern for underlying chromosomal or structural fetal abnormalities. Combining non-invasive prenatal testing (NIPT) with detailed early ultrasound may improve the detection and prediction of such adverse outcomes. Non-invasive prenatal testing (NIPT), using cell-free DNA, has transformed prenatal screening by providing high sensitivity and positive predictive value for common aneuploidies, thereby reducing the need for invasive diagnostic procedures.[3]

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Despite the strong performance of NIPT for trisomies 21, 18, and 13, contemporary cohorts have shown that a normal NIPT result does not fully exclude risk in fetuses with increased NT.[4] Fetuses with elevated NT may harbor cardiac malformations, rare copy-number variants, single-gene disorders, or structural anomalies that escape targeted NIPT, and these conditions can adversely affect pregnancy outcomes.[2,5,6]

Recent large retrospective and prospective analyses indicate that combining calibrated NT measurement with detailed first-trimester ultrasound and, where indicated, genome-wide or expanded NIPT increases diagnostic yield and improves prognostication for live birth, termination, and perinatal morbidity. Nonetheless, the magnitude of incremental benefit depends on the NT threshold, ultrasound findings, and the NIPT platform, producing variability in clinical pathways and counseling practices.[5,7]

In Pakistan and the South-East Asian region access to standardized first-trimester screening, accredited NT measurement, and cfDNA testing is heterogeneous; national guidance and institutional reports emphasize locally adapted algorithms, sonographer training, and clear counseling frameworks to manage resource and cultural constraints. Regional data and context-sensitive recommendations are therefore essential to optimize practice.[8,9]

This study aims to compare adverse pregnancy outcomes among fetuses with NT ≥3.0 mm according to NIPT and first-trimester ultrasound findings, and to estimate the diagnostic and prognostic yield of each modality alone and in combination. By quantifying rates of chromosomal abnormalities, structural defects, pregnancy loss, and liveborn outcomes across strata, the study will provide clinicians and policymakers with actionable data to refine referral thresholds, select appropriate NIPT strategies, and counsel families regarding prognosis, follow-up imaging, and invasive testing decisions in resource-limited settings, and improve perinatal outcomes and support national screening protocols.

METHODOLOGY

This retrospective analytical cohort study was carried out at the Combined Military Hospital (CMH), Kharian, Pakistan, from January 2022 to December 2025. The study included 180 pregnant women who underwent first-trimester ultrasound screening between 11 + 0 and 13 + 6 weeks of gestation, during which nuchal translucency (NT) was measured. All cases in which the fetal NT measurement was equal to or greater than 3.0 mm were identified from the hospital's fetal medicine database. These patients subsequently underwent non-invasive prenatal testing (NIPT) and were followed up for pregnancy outcomes.

All singleton pregnancies fulfilling the inclusion criteria were included. Women with multiple gestations, missing NIPT data, known maternal systemic diseases affecting fetal development, or incomplete follow-up records were excluded. The study sample was estimated using the WHO calculator, assuming a 15% prevalence of adverse pregnancy outcomes among fetuses with increased NT, based on previously published literature. With a 5% margin of error and a 95% confidence level, the calculated sample size was 196; however, a total of 180 cases were included based on available data during the study period.[10] Data were retrieved from hospital electronic records, ultrasound logs, and laboratory databases. Demographic and clinical parameters such as maternal age, parity, gravidity, body mass index (BMI), gestational age at ultrasound, and prior obstetric history were recorded. Ultrasound parameters included NT measurement, crown–rump length (CRL), and detection of any additional structural anomalies. NT measurements were performed by certified sonographers using standardized protocols and equipment, ensuring optimal mid-sagittal imaging, correct fetal position, and magnification. All measurements were documented in millimeters, and corresponding images were archived.

Following the identification of increased NT, NIPT

was performed through an accredited external laboratory using cell-free fetal DNA extracted from maternal plasma. Tests primarily screened for trisomy 21, 18, and 13, with extended panels including sex chromosome aneuploidies and selected microdeletions when clinically indicated. Cases with high-risk or inconclusive NIPT results were offered invasive diagnostic procedures such as chorionic villus sampling (CVS) or amniocentesis for cytogenetic confirmation through karyotyping or chromosomal microarray analysis, depending on availability.

Pregnancy outcomes were obtained from medical records, follow-up data, and neonatal records. The primary outcome of the study was the presence of chromosomal abnormalities. Secon-dary outcomes included a composite adverse pregnancy outcome, defined as the occurrence of structural anomalies, miscarriage, intrauterine fetal demise, termination of pregnancy, or abnormal neonatal outcome. Individual outcome categories were also recorded separately. Neonatal outcomes were assessed at birth or during the early postnatal period.

All collected data were anonymized before analysis, and each case was assigned a unique study identification code. Data entry and verification were performed by two independent investigators (postgraduate trainees) to ensure accuracy. All statistical analyses were carried out using SPSS version 25. Baseline maternal and fetal characteristics were summarized using descriptive measures, with continuous variables reported as mean ± SD or as median and interquartile range, while categorical variables expressed as counts and percentages. Differences between groups were assessed using the chi-square test for categorical variables and either the independent t-test for continuous variables, as appropriate. Logistic regression models were applied to evaluate the relationship between ultrasound findings, NIPT results, and adverse pregnancy outcomes, while accounting for potential confounding factors such as maternal age, BMI, and gestational age. Measures of diagnostic performance—including sensitivity, specificity, positive and negative predictive values, and the area under the receiver operating characteristic curve (AUC)—were calculated for NIPT, first-trimester ultrasound, and the combination of both tests. Additional subgroup analyses were performed according to NT categories (3.0–3.4 mm, 3.5–4.9 mm, and ≥5.0 mm) to determine how predictive accuracy varied across different NT ranges.

As the study was retrospective study based on review of existing medical records, no direct patient contact was involved and the requirement for informed consent was waived. The ethics approval was granted by Institutional Ethics Review Board of Combined Military Hospital (CMH), Kharian Cantt, Pakistan (Approval No.: CKMC/IERB/AC-00284, Dated: 15 December, 2025) prior to data analysis, in accordance with institutional guidelines.

RESULTS

A total of 180 pregnant women included in the study. The mean maternal age was 31.2±4.6 years (range: 21–43), and the mean gestational age at ultrasound was 12.4±0.6 weeks. The majority of participants were multigravida (58.3%), 98 (54.4%) were parous women with at least one prior viable delivery, and the mean nuchal translucency (NT) measurement was 3.8 ± 0.7 mm (Table I).

Table I: Maternal and Fetal Baseline Features (n = 180)

Variable	Mean ± SD / n (%)
Maternal age (years)	31.2±4.6
Gestational age at scan (weeks)	12.4±0.6
Gravidity (≥2)	105 (58.3%)
Parity (≥1)	98 (54.4%)
BMI (kg/m^2)	26.8±3.5
Crown–rump length (mm)	63.2±5.8
Mean NT measurement (mm)	3.8±0.7
NT 3.0–3.4 mm	72 (40.0%)
NT 3.5–4.9 mm	78 (43.3%)
NT ≥ 5.0 mm	30 (16.7%)

Among 180 cases, NIPT yielded positive results in 26 (14.4%), negative in 150 (83.3%), and inconclusive in 4 (2.2%) due to low fetal fraction. Among the positive results, trisomy 21 was the most frequent (46.1%), followed by trisomy 18 (26.9%), trisomy 13 (11.5%), and sex chromosome anomalies (15.3%).

Invasive diagnostic testing was performed in 48 (26.7%) cases. Chromosomal abnormalities (primary outcome) were confirmed in 22 (12.2%) fetuses. Overall, NIPT showed superior diagnostic performance compared with ultrasound alone, and the combined approach yielded the highest accuracy (Table II). A total of 54 (30%) pregnancies experienced the secondary composite adverse outcome, which included chromosomal abnormalities, major structural defects, miscarriages, intrauterine fetal demise, or termination of pregnancy.

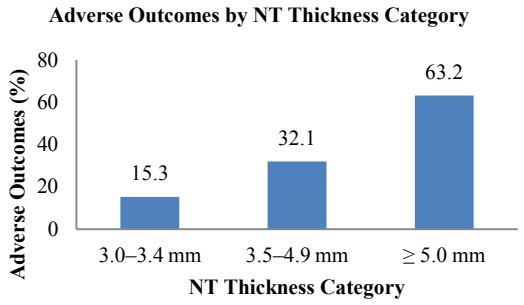
Table II: Diagnostic Performance of NIPT and Ultrasound in Detecting Chromosomal Abnormalities

Parameter	NIPT	Ultrasound alone	Combined NIPT + US
Sensitivity (%)	95.4	72.7	98.1
Specificity (%)	98.6	88.5	98.8
PPV (%)	88.5	56.3	90.4
NPV (%)	99.3	92.7	99.6

Overall, 54 (30%) pregnancies had adverse outcomes, which included chromosomal abnormalities, major structural defects, miscarriages, intrauterine fetal demise, or termination of pregnancy. The remaining 126 (70%) resulted in live births with normal postnatal outcomes. These data reflect that chromosomal abnormalities were considered the primary outcome, while the broader group of adverse outcomes constituted the secondary composite endpoint.

When stratified by NT category, adverse outcomes increased significantly with NT thickness: 15.3% for NT 3.0–3.4 mm, 32.1% for NT 3.5–4.9 mm, and 63.3% for NT ≥ 5.0 mm ($p < 0.001$) (Figure 1).

Figure I. Association between NT Thickness and Adverse Pregnancy Outcomes



Graph showing an upward trend in adverse outcome rates by NT category.

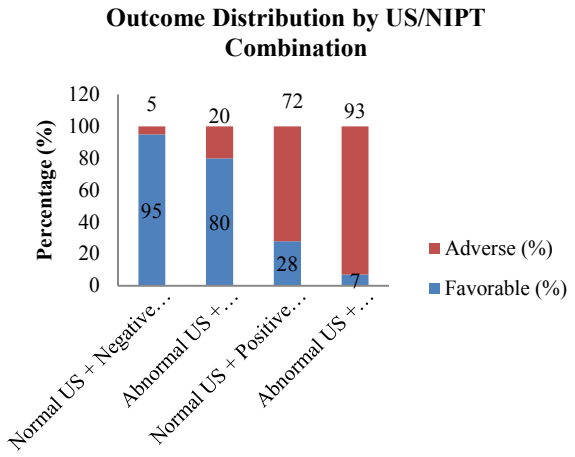
Among 150 pregnancies with a negative NIPT, 8 (5.3%) resulted in structural anomalies despite a normal karyotype, including cardiac and renal defects. In contrast, 84.6% of positive NIPT cases had either chromosomal or structural abnormalities confirmed postnatally or after invasive testing. The combination of abnormal ultrasound plus positive NIPT showed the highest association with adverse outcome ($p < 0.001$).

Logistic regression revealed that increased NT ≥ 5.0 mm (OR = 7.4; 95% CI 3.0–18.2; $p < 0.001$) and positive NIPT (OR = 6.9; 95% CI 2.7–17.6; $p < 0.001$) were independent predictors of adverse pregnancy outcome after adjusting for maternal age and BMI (Table III).

Table III. Predictors of Adverse Pregnancy Outcomes Identified by Logistic Regression

Variable	Adjusted OR	95% CI	P-value
Maternal age > 35 years	1.6	0.7 – 3.7	0.23
NT ≥ 5.0 mm	7.4	3.0 – 18.2	< 0.001
Positive NIPT	6.9	2.7 – 17.6	< 0.001
Structural anomaly on ultrasound	4.5	1.9 – 10.4	0.001
BMI > 30 kg/m^2	1.2	0.5 – 2.8	0.62

Figure II: Pregnancy Outcomes by NIPT and Ultrasound Findings

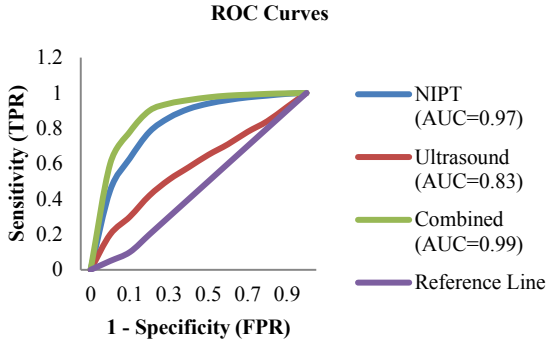


The figure II illustrates that the combination of ultrasound and NIPT offers the best predictive performance for identifying high-risk pregnancies (Normal ultrasound + negative NIPT: 5% adverse, Abnormal ultrasound + negative NIPT: 20% adverse, Normal ultrasound + positive NIPT: 72% adverse, Abnormal ultrasound + positive NIPT: 93% adverse).

ROC curve analysis was performed to evaluate the diagnostic accuracy of NIPT, first-trimester ultrasound, and their combination in predicting chromosomal or structural abnormalities. The AUC for NIPT was 0.97 (95% CI: 0.94–0.99), indicating excellent discriminative ability. In contrast, the AUC for ultrasound alone was 0.83 (95% CI: 0.76–0.90), while the combined model (NIPT + ultrasound) attained an AUC of 0.99 (95% CI: 0.97–1.00), reflecting near-perfect diagnostic accuracy (Figure 3).

ROC curve analysis was performed to evaluate the diagnostic accuracy of NIPT, first-trimester ultrasound, and their combination in predicting chromosomal abnormalities (primary outcome) and associated structural defects (secondary outcome). The AUC for NIPT was 0.97, indicating excellent discriminative ability for the primary outcome. In contrast, the AUC for ultrasound alone was 0.83, while the combined model (NIPT + ultrasound) attained an AUC of 0.99 (Figure 3).

Figure 3: ROC Curve Comparison of Diagnostic Accuracy



DISCUSSION

In this study involving 180 pregnancies with an increased NT measurement (≥3.0 mm), chromosomal abnormalities were defined as the primary outcome, and about one-third of pregnancies experienced secondary composite adverse outcomes. The likelihood of complications rose with greater NT thickness. NIPT performed very well in detecting common chromosomal abnormalities, showing a sensitivity of 95.4% and specificity of 98.6%. When findings from NIPT were combined with detailed first-trimester ultrasound results, the ability to predict both primary and secondary outcomes improved even further, with areas under the ROC curve of 0.97 for NIPT alone, 0.83 for ultrasound, and 0.99 for the combined assessment. These findings support an integrated diagnostic pathway in which molecular screening and careful sonographic assessment provide complementary information for counseling and decision-making.

The observed proportion of chromosomal abnormalities (primary outcome) and the overall secondary composite adverse outcomes in this study is broadly consistent with large contemporary series that report elevated risks among pregnancies with NT ≥ 3.0 mm, although reported rates vary by population, ascertainment, and the depth of genetic testing used. Several recent multicenter cohorts and registry analyses have documented adverse outcome rates ranging from ~25–32% for NT thresholds around 3.0 mm, particularly when

microarray and extended genetic testing were applied to cases with abnormal ultrasound or positive screening.[11,12] Differences between studies likely reflect variable inclusion of terminations for major anomalies, availability of chromosomal microarray, and whether isolated structural anomalies with normal karyotype were counted as adverse outcomes.

The high sensitivity and specificity of NIPT in this study align with numerous recent evaluations showing excellent performance of cell-free DNA screening for trisomy 21, 18 and 13 in high- and mixed-risk populations.[13-15] In this study, PPV (88.5%) and NPV (99.3%) approximate those reported in other tertiary-center cohorts where pretest probability is enriched by sonographic markers such as increased NT.[13,16] Notably, NIPT remains primarily targeted to common whole-chromosome aneuploidies; residual risk for pathogenic copy-number variants and single-gene disorders persists when NIPT is negative — a pattern mirrored in 8 cases (5.3% of negative NIPT) with structural anomalies and normal karyotype. Such limitations have been reported in previous studies and underline the importance of ongoing ultrasound monitoring and additional genetic evaluation when an increased NT is observed, even if NIPT results are negative.[17,18]

The ROC results highlight the added benefit of using both NIPT and first-trimester ultrasound together. Ultrasound can identify structural anomalies and subtle markers that NIPT may overlook, while NIPT remains highly effective in detecting common chromosomal abnormalities. Several recent studies comparing standard NIPT, expanded NIPT, and early ultrasound have reached similar conclusions: combining these approaches improves sensitivity and PPV for a wider spectrum of chromosomal and structural disorders. However, broader testing panels can slightly increase false-positive rates.[12,14] These findings support a practical strategy in which standard NIPT is paired with careful first-trimester ultrasound for most pregnancies, while genome-wide or chromosomal microarray testing is reserved for cases with persistent ultrasound abnormalities or markedly increased NT.

In this analysis, NT measurements of 5.0 mm or higher and a positive NIPT result emerged as the strongest independent predictors of chromosomal abnormalities (primary outcome) and were also associated with increased risk of secondary adverse outcomes. This aligns with previous studies showing that the risk of chromosomal or structural abnormalities increases progressively with higher NT values.[1,19] In clinical practice, these findings suggest that invasive testing should be offered to patients with NT $\geq$ 5.0 mm or a positive NIPT, while moderate NT elevations with negative NIPT require careful follow-up with detailed ultrasound. For those with moderate NT elevations and negative NIPT, careful follow-up with detailed ultrasound remains essential to monitor fetal development.

Local healthcare context plays an important role in prenatal care. In many low- and middle-income settings, availability of skilled sonographers, accredited NIPT laboratories, and invasive testing options can be limited, which affects both the detection of primary outcomes and the observation of secondary adverse outcomes.[20] This study emphasizes that even when NIPT is accessible, proper sonographer training and clear referral protocols are crucial for reliable results and effective counseling.

CONCLUSION

In pregnancies with increased NT, NIPT provides excellent detection of common aneuploidies but does not obviate the need for detailed first-trimester ultrasound — the combination yields superior diagnostic and prognostic accuracy and should be included in local algorithms for further genetic testing and counseling.

***Limitations**:* Being a single-center study retrospective analysis, this study may be subject to selection bias and limitations related to incomplete

data recording. In addition, only women with increased NT who underwent NIPT and had complete follow-up were included, while those who declined NIPT or were lost to follow-up were excluded, which may have introduced selection bias and affected the generalizability of the findings. Furthermore, not all participants underwent invasive diagnostic testing, raising the possibility of verification bias, as chromosomal status in some cases was inferred from postnatal outcomes rather than confirmed through a uniform reference standard. Chromosomal microarray testing was not performed, so some pathogenic copy-number variants could have gone undetected. Nonetheless, the concordance of key findings with other recent series suggests robustness.

Financial Disclosures: None
Conflict of Interest: None

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Date of Submission: 28-01-2026
Revised Date: 15-02-2026
Accepted Date: 10-04-2026