Section: Miscellaneous

JAMP

Original Research Article

# NUCHAL TRANSLUCENCY SCREENING: ENHANCING EARLY DETECTION OF FETAL DISORDERS

*Received : 01/05/2026*
*Received in revised form : 12/06/2026*
*Accepted : 27/06/2026*

*Keywords:*
*Nuchal translucency, crown-rump length, prenatal screening, fetal anomalies, chromosomal abnormalities, first trimester.*

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DOI: 10.47009/jamp.2026.8.4.19

Source of Support: Nil,
Conflict of Interest: None declared

*Int J Acad Med Pharm*
2026; 8 (4); 106-110

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## ABSTRACT

**Background:** Nuchal translucency (NT) screening is an important first-trimester prenatal tool for the early detection of fetal chromosomal and structural abnormalities. It serves as a non-invasive marker for identifying aneuploidies such as Down syndrome, Trisomy 18, and Trisomy 13, as well as congenital heart defects. **Materials and Methods:** This prospective observational study was carried out in the Department of Anatomy in collaboration with the Departments of Obstetrics and Gynecology and Radiodiagnosis at Sarojini Naidu Medical College from January 2026 to May 2026. A total of 218 pregnant women with singleton pregnancies between 11 weeks and 13 weeks + 6 days of gestation were included. NT measurements were performed according to Fetal Medicine Foundation guidelines, and CRL was measured between 45 mm and 84 mm. **Results:** The majority of participants (58.7%) belonged to the age group of 25–30 years, with a mean gestational age of 12.4 ± 0.8 weeks. The mean NT thickness was 1.72 ± 0.54 mm, with a median of 1.70 mm and a 95th percentile of 2.8 mm. A significant positive correlation was observed between CRL and NT thickness ($r = 0.31$, $p < 0.001$), indicating that NT thickness increases with fetal growth. Increased NT above the CRL-specific 95th percentile was observed in 5.0% of cases, while 3.7% had NT >3.0 mm. **Conclusion:** The study confirms that NT measurement is a reliable and effective first-trimester screening tool. CRL-adjusted NT percentile charts improve the accuracy of fetal risk assessment and aid in the early identification of high-risk pregnancies, enabling timely counseling and intervention.

## INTRODUCTION

Nuchal Translucency screening is the first-trimester prenatal work-up. It is one of the first non-invasive ways to get a understanding on the fetus and observed any chromosomal or structural issues that may be present. It is done once the pregnancy has reached 11 weeks but before 13 weeks and 6 days, it provided the fetal crown-rump length is in the 45 to 84 mm range. In essence, Nuchal Translucency is the sonographic measurement of the fluid-filled subcutaneous space at the back of the neck. A thicker than normal reading is a strong indicator of aneuploidies like Down syndrome, Trisomy 18 or 13, not to mention congenital heart problems and other genetic syndromes.[1]

This screening was introduced in the 1990s revolutionized prenatal screening for shifting the focus toward early risk , enabling timely counseling, diagnostic testing, and decision-making for expectant parents. When it combined with maternal serum biomarkers—such as pregnancy-associated plasma protein-A (PAPP-A) and free β-human chorionic gonadotropin (β-hCG)—along with maternal age, Nuchal Translucency screening forms the basis of the combined first-trimester screening test, achieving detection rates of approximately 85–90% for major aneuploidies with relatively low false-positive rates.[2]

Its also identifying chromosomal abnormalities, Nuchal Translucency measurement provides valuable insight into early fetal anatomy and physiology. It also can reflect underlying cardiac dysfunction, lymphatic drainage abnormalities, or any altered extracellular matrix composition, which making it very important marker of broader fetal health.[3] As prenatal medicine increasingly integrates advanced technologies such as Cell-free fetal DNA analysis, its screening remains highly relevant, particularly in settings where

comprehensive genetic testing may not be readily accessible or as part of a multimodal screening strategy.[4]
Thus, NT screening serves as a critical “first clue” in fetal assessment—bridging in traditional ultrasound evaluation with modern genomic medicine and significantly influencing the trajectory of prenatal care.

## MATERIALS AND METHODS

This prospective observational study was conducted in the Department of Anatomy, with collaboration of the Departments of Obstetrics and Gynecology and Radiodiagnosis, at Sarojini Naidu Medical College over a period of January 2026 to may 2026 . The study was undertaken to evaluate the significance of Nuchal Translucency measurement as an early marker of fetal chromosomal and structural abnormalities, with emphasis on its anatomical and developmental correlations.
A total of 218 pregnant women with singleton gestation between 11 weeks and 13 weeks + 6 days were recruited from the antenatal outpatient clinic of the Department of Obstetrics and Gynecology. Gestational age was confirmed based on the last menstrual period and ultrasonographic crown-rump length (CRL) measurements between 45 mm and 84 mm, in accordance with the recommendations of the Fetal Medicine Foundation. Cases of multiple pregnancies, uncertain gestational age, fetal demise, and pregnancies complicated by major maternal systemic illnesses were excluded from the study.

## RESULTS

**Table 1: Baseline Demographic Characteristics of Study Participants**

| Variables | Number (n=218) | Percentage (%) |
|---|---|---|
| Maternal Age (years) | | |
| <25 years | 24 | 11.0 |
| 25–30 years | 128 | 58.7 |
| >30 years | 66 | 30.3 |
| Parity | | |
| Primigravida | 114 | 52.3 |
| Multigravida | 104 | 47.7 |
| Mean gestational age (weeks) | 12.4 ± 0.8 | — |

The majority of study participants belonged to the 25–30 years age group (58.7%), followed by those aged above 30 years (30.3%), while only 11.0% were below 25 years. Primigravida constituted a slightly higher proportion than multigravida (52.3% vs. 47.7%). The mean gestational age at assessment was 12.4 ± 0.8 weeks, confirming that nuchal translucency (NT) measurements were performed within the standard first-trimester screening period (11–13+6 weeks).

**Table 2: Mean ± SD of Crown-Rump Length (CRL) and Gestational Age (GA) in Pregnant Women with Normal and Increased Nuchal Translucency Thickness**

| Parameters | Normal NT (n=196) Mean ± SD | Increased NT (n=22) Mean ± SD | p-value |
|---|---|---|---|
| Crown-Rump Length (CRL) (mm) | 61.8 ± 9.4 | 64.2 ± 8.7 | 0.218 |
| Gestational Age (weeks) | 12.2 ± 0.7 | 12.6 ± 0.6 | 0.143 |

The mean crown-rump length (CRL) was slightly higher in the increased NT group (64.2 ± 8.7 mm) compared to the normal NT group (61.8 ± 9.4 mm), although this difference was not statistically significant (p = 0.218). Similarly, the mean gestational age was marginally higher in cases with increased NT (12.6 ± 0.6 weeks) compared to those with normal NT (12.2 ± 0.7 weeks), but this difference was also not statistically significant (p = 0.143). However, subsequent correlation analysis demonstrated a significant positive relationship between CRL and NT thickness, indicating that NT increases progressively with fetal growth.

**Table 3: Percentile Values of Nuchal Translucency Thickness for 10 mm Crown-Rump Length (CRL) Intervals**

| CRL Interval (mm) | Number of Cases (n) | 5th Percentile (mm) | 50th Percentile (Median) (mm) | 95th Percentile (mm) |
|---|---|---|---|---|
| 45–54 | 38 | 0.9 | 1.4 | 2.3 |
| 55–64 | 62 | 1.0 | 1.6 | 2.5 |
| 65–74 | 71 | 1.1 | 1.8 | 2.8 |
| 75–84 | 47 | 1.2 | 2.0 | 3.1 |

The percentile analysis demonstrated a gradual increase in NT thickness with increasing crown-rump length. The median NT value increased from 1.4 mm in fetuses with CRL between 45–54 mm to 2.0 mm in those with CRL between 75–84 mm. Similarly, the 95th percentile values progressively increased from 2.3 mm to 3.1 mm across the CRL intervals, reflecting the physiological relationship between fetal growth and NT thickness.

Table 4: Distribution of Nuchal Translucency Thickness Across Measurement Ranges

| NT Thickness Range (mm) | Number of Cases (n=218) | Percentage (%) |
|---|---|---|
| ≤ 1.0 mm | 32 | 14.7 |
| 1.1 – 1.5 mm | 68 | 31.2 |
| 1.6 – 2.0 mm | 74 | 33.9 |
| 2.1 – 2.5 mm | 26 | 11.9 |
| > 2.5 mm | 18 | 8.3 |

The distribution of NT measurements showed that the majority of fetuses (65.1%) had NT thickness between 1.1 mm and 2.0 mm, which lies within the expected physiological range during the first trimester. A smaller proportion (20.2%) had NT values greater than 2.0 mm, while 8.3% had NT thickness above 2.5 mm, representing a clinically important subgroup for further evaluation.

Table 5: Overall Descriptive Statistics of Nuchal Translucency Thickness

| Parameters | Value |
|---|---|
| Mean NT thickness (mm) | 1.72 ± 0.54 |
| Median NT thickness (mm) | 1.70 |
| Minimum NT thickness (mm) | 0.8 |
| Maximum NT thickness (mm) | 3.6 |
| 5th Percentile (mm) | 0.9 |
| 95th Percentile (mm) | 2.8 |

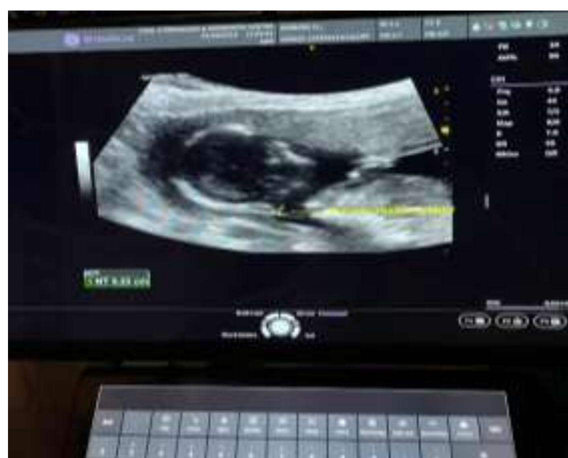

Figure 1: Ultrasonographic measurement of nuchal translucency (NT) in a first-trimester fetus (11–13+6 weeks gestation).

The overall mean NT thickness among the study population was 1.72 ± 0.54 mm, with values ranging from 0.8 mm to 3.6 mm. The median NT thickness was 1.70 mm, indicating that most measurements clustered within the normal physiological range. The overall 95th percentile was 2.8 mm, consistent with the CRL-stratified percentile analysis.

Table 6: Correlation Between Crown-Rump Length (CRL) and Nuchal Translucency Thickness

| Variables | Correlation Coefficient ® | p-value |
|---|---|---|
| CRL vs NT thickness | 0.31 | <0.001 |

A statistically significant mild-to-moderate positive correlation was observed between crown-rump length and NT thickness ($r = 0.31$, $p < 0.001$), indicating that NT thickness increases proportionally with fetal growth. This finding supports the physiological relationship between fetal size and NT measurements and emphasizes the importance of interpreting NT values according to gestational age and CRL.

Table 7: Prevalence of Increased Nuchal Translucency Thickness Based on CRL-Specific Standard Cut-off Values

| NT Threshold | Number of Cases (n=218) | Percentage (%) |
|---|---|---|
| ≥95th percentile (CRL-specific) | 11 | 5.0 |
| >3.0 mm | 8 | 3.7 |
| >3.5 mm | 2 | 0.9 |

Out of 218 fetuses, 11 cases (5.0%) demonstrated NT measurements above the CRL-specific 95th percentile, consistent with expected percentile distribution. Additionally, 8 cases (3.7%) had NT thickness greater than 3.0 mm, while only 2 cases (0.9%) exceeded 3.5 mm. These findings indicate that a small but clinically important proportion of the study population fell into higher-risk categories warranting further diagnostic evaluation.

## DISCUSSION

Our present study was undertaken to evaluate the pattern of nuchal translucency thickness in relation to


ISSN (O): 2687-5365; ISSN (P): 2753-6556

crown-rump length during the first trimester of pregnancy and to establish normative percentile values in the local population. nuchal translucency measurement performed between 11 and 13+6 weeks of gestation is considered the standard screening window, as recommended by the Fetal Medicine Foundation, because this period provides optimal fetal visualization and allows accurate measurement of NT thickness.[1] our finding shows the mean gestational age at examination was 12.4 ± 0.8 weeks, which is well within the recommended range and comparable to the gestational age range used in previous landmark studies.[2,3]

The baseline demographic characteristics revealed that the majority of participants were between 25–30 years of age (58.7%), followed by women older than 30 years (30.3%). Primigravida women constituted 52.3% of the study population. Maternal age is a well-established factor in fetal chromosomal risk assessment, and its integration into first-trimester screening significantly improves detection rates for trisomy 21 and other aneuploidies 4,5. Similar maternal age distributions have been reported in multicentric screening studies, emphasizing the importance of maternal demographic characteristics in prenatal risk stratification.[6]

In the present study, the mean CRL was slightly higher in fetuses with increased NT (64.2 ± 8.7 mm) compared to those with normal NT (61.8 ± 9.4 mm), though this difference was not statistically significant ($p = 0.218$). Likewise, the gestational age was marginally higher in the increased NT group but without statistical significance. These findings suggest that increased NT thickness is not merely dependent on gestational age but reflects individual fetal variation. Similar observations were reported by Spencer et al,[7] who demonstrated that NT thickness must always be interpreted in relation to CRL. Nicolaides also highlighted that CRL-adjusted NT assessment provides greater diagnostic accuracy than using fixed cut-off values.[8]

A major finding of this study was the progressive rise in NT thickness with increasing CRL. The median NT thickness increased from 1.4 mm in the CRL group of 45–54 mm to 2.0 mm in the 75–84 mm CRL group, while the 95th percentile increased from 2.3 mm to 3.1 mm. This trend is consistent with the original FMF normative charts and confirms the physiological relationship between fetal growth and NT thickness. Similar findings were reported by Pandya et al,[9] and Wright et al,[10] who established that NT increases linearly with CRL. This progressive increase has been attributed to the maturation of the fetal lymphatic system and transient accumulation of subcutaneous fluid.[11]

The NT distribution pattern in the present study showed that 65.1% of fetuses had NT values between 1.1 and 2.0 mm, which is considered within the normal physiological range. Only 8.3% had NT values above 2.5 mm. These findings are comparable with those of Sebire et al,[12] who reported that most fetuses in routine screening populations demonstrate NT values below 2.5 mm, with only a minority showing abnormal thickening. Similarly, Snijders et al,[13] found that NT values above the 95th percentile are relatively uncommon but carry significant diagnostic implications.

The overall mean NT thickness in the present study was 1.72 ± 0.54 mm, with a median of 1.70 mm and a 95th percentile value of 2.8 mm. These findings are comparable to those reported by Salomon et al,[14] who established reference ranges for NT thickness across the first trimester. Their study showed a similar average NT value and emphasized the need for population-specific reference ranges. This similarity suggests that the present study population follows the internationally accepted NT growth pattern.

A statistically significant positive correlation was observed between CRL and NT thickness ($r = 0.31$, $p < 0.001$), confirming that NT increases proportionally with fetal growth. This finding is in agreement with previous studies by Kagan et al,[15] and Wapner et al,[16] both of whom demonstrated that CRL-specific NT interpretation improves screening sensitivity. This reinforces the principle that NT values should always be interpreted relative to fetal size rather than as isolated measurements.

In the current study, 5.0% of fetuses had NT values above the CRL-specific 95$^{th}$ percentile, which is statistically expected and confirms the reliability of the constructed percentile chart. Additionally, 3.7% of fetuses had NT >3.0 mm, while 0.9% exceeded 3.5 mm. Similar prevalence rates were reported in the SURUSS trial by Wald et al,[17] where elevated NT values were observed in approximately 4–5% of screened pregnancies. Malone et al,[18] also reported comparable findings and emphasized that increased NT is strongly associated with chromosomal abnormalities.

The significance of increased NT extends beyond chromosomal abnormalities. Hyett et al,[11] demonstrated a strong association between increased NT and congenital heart defects, even in chromosomally normal fetuses. Bilardo et al,[19] further reported that euploid fetuses with increased NT remain at higher risk of structural anomalies and adverse pregnancy outcomes. Therefore, increased NT should not be dismissed even after normal karyotype results.

The American College of Obstetricians and Gynecologists (ACOG) recommends that fetuses with NT measurements above the 95$^{th}$ percentile or greater than 3.0 mm should undergo detailed counseling, biochemical screening, and, when indicated, invasive diagnostic testing.[20] The present study identified 11 such cases, highlighting the practical importance of NT measurement in routine antenatal care. Early identification of these high-risk fetuses provides an opportunity for timely intervention and improved pregnancy outcomes.

Overall, the findings of the present study are in close agreement with previous international studies. The progressive increase of NT with CRL, the normal

distribution of NT values, and the prevalence of increased NT all support the validity of CRL-specific percentile charts in first-trimester screening. These findings reinforce the role of NT as an essential component of prenatal screening and support its routine use for early identification of high-risk pregnancies. Most importantly, the present study contributes local normative data that may help improve the accuracy of fetal screening in the regional population.

## CONCLUSION

The present study reinforces the importance of first-trimester NT measurement as a reliable and effective screening tool. The findings align with established evidence and support the incorporation of CRL-adjusted NT percentile charts into routine prenatal screening protocols for early identification of high-risk pregnancies.

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