

RESEARCH ARTICLE

Analysis of Nuchal Translucency Measurements and Their Association with Pregnancy Outcomes

Ense Kalınlığı Ölçümlerinin Analizi ve Gebelik Sonuçlarıyla İlişkisi

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ABSTRACT

Objective: This study aimed to assess the prenatal and perinatal outcomes of fetuses with increased nuchal translucency (NT) and examine the relationship between NT thickness, chromosomal findings, and adverse pregnancy outcomes, such as fetal loss and neonatal morbidity.

Materials and Methods: This retrospective study included pregnant women referred to the perinatology clinic between February 2023 and July 2025 due to increased NT (≥ 3.0 mm) detected during first-trimester ultrasonography. Detailed ultrasonographic evaluation and invasive prenatal diagnostic testing were performed. Karyotyping and chromosomal microarray analysis (CMA) were conducted in all cases, and whole-exome sequencing (WES) was selectively applied.

Results: A total of 39 fetuses with increased NT were evaluated. The median NT value was 4.0 mm, and 35.9% of cases had NT values ≥ 4.5 mm, representing a high-risk subgroup. Chromosomal abnormalities were detected in 56.4% of fetuses, with trisomy 21 being the most common abnormality (35.9%). Additional ultrasonographic findings were observed in 64.1% of fetuses with a normal karyotype, among whom intrauterine fetal loss and neonatal morbidity rates were 15.4%. Chromosomal microarray analysis provided additional diagnostic yield beyond karyotyping. Whole-exome sequencing did not identify pathogenic variants; however, one case resulted in intrauterine fetal loss. These findings indicate that increased NT may be associated with adverse pregnancy outcomes even in the absence of detectable genetic abnormalities.

Conclusion: Increased NT is a strong predictive marker of chromosomal abnormalities, structural anomalies, and adverse neonatal outcomes, regardless of genetic test results. Chromosomal microarray analysis should be considered the minimum diagnostic standard, while advanced molecular testing, serial ultrasound follow-up, and comprehensive counselling may be beneficial in selected cases.

Keywords: Nuchal Translucency, Chromosomal Microarray Analysis, Prenatal Diagnosis.

ÖZET

Amaç: Bu çalışmanın amacı, artmış nuchal saydamlık (NT) saptanan fetüslerin prenatal ve perinatal sonuçlarını değerlendirmek ve NT kalınlığı ile kromozomal bulgular, fetal kayıp ve neonatal morbidite gibi olumsuz gebelik sonuçları arasındaki ilişkiyi araştırmaktır.

Gereç ve Yöntem: Bu retrospektif çalışmaya, 2023 yılı Şubat ayı ile 2025 yılı Temmuz ayı arasında, birinci trimester ultrason taramasında artmış NT ($\geq 3,0$ mm) saptanması nedeniyle perinatoloji kliniğine yönlendirilen gebeler dahil edildi. Tüm olgular ayrıntılı ultrasonografik değerlendirmeden geçirildi ve invaziv prenatal tanı testleri önerildi. Tüm vakalarda karyotipleme ve kromozomal mikrodizi analizi (CMA) yapıldı, gerektiğinde seçilmiş fetüslerde tüm ekzom dizileme (WES) uygulandı. Demografik özellikler, ultrason bulguları, genetik sonuçlar ve gebelik sonuçları analiz edilmiştir.

Bulgular: Artmış NT saptanan toplam 39 fetüs çalışmaya dahil edildi. Median NT değeri 4,0 mm idi ve olguların %35,9'unda NT $\geq 4,5$ mm olarak belirlendi; bu grup yüksek riskli olguların önemli bir kısmını oluşturdu. Fetüslerin %56,4'ünde kromozomal anomali tespit edildi ve en sık görülen anomali Trizomi 21 (%35,9). Normal fetal karyotipe sahip fetüslerin %64,1'inde ek ultrasonografik bulgular saptandı; bu grupta intrauterin fetal kayıp ve neonatal morbidite oranları %15,4 ile daha yüksek bulundu. Karyotiple karşılaştırıldığında CMA ek

tanısal katkı sağladı. Gerçekleştirilen WES analizinde patojenik varyant saptanmadı, ancak bu olgulardan birinde intrauterin fetal kayıp gelişti. Genetik anomali saptanmasa dahi artmış NT varlığının olumsuz gebelik sonuçlarıyla ilişkili olabileceği gösterildi.

Sonuç: Artmış NT, kromozomal anomaliler, yapısal malformasyonlar ve olumsuz perinatal sonuçlar açısından yüksek riski gösteren önemli bir prognostik belirteçtir. Kromozomal mikrodizi analizinin asgari tanısal standart olarak değerlendirilmesi, ileri moleküler testlerin seçici kullanımı, ultrason takipleri ve genetik danışmanlık, optimal klinik yönetim için gereklidir.

Anahtar Sözcükler: Ense Kalınlığı, Kromozomal Mikrodizi Analizi, Prenatal Tanı.

The measurement of NT during the first-trimester ultrasound examination has become an integral component of prenatal screening for chromosomal abnormalities, particularly trisomies 21, 18, and 13 (1). Increased NT, defined as a subcutaneous fluid collection at the fetal neck greater than the 95th or 99th percentile for crown-rump length, has been identified as an important sonographic marker not only for aneuploidies but also for a broad spectrum of structural malformations, genetic syndromes, and adverse perinatal outcomes (1, 2). While the association between increased NT and chromosomal abnormalities is well established, a substantial proportion of fetuses with increased NT measurements exhibit normal karyotypes yet remain at increased risk for adverse outcomes. These include miscarriage, intrauterine fetal death, structural anomalies-particularly congenital heart defects-and neonatal morbidity (2). Lithner et al. (2) demonstrated that, even in euploid fetuses, the risk of unfavorable pregnancy outcomes rises in proportion to the degree of NT elevation, with structural defects observed in nearly one-fifth of such pregnancies. Similarly, Bet et al. (1) reported that fetuses with early increased NT, even when the measurement normalized later in gestation, continued to carry a higher risk of genetic or structural abnormalities and pregnancy loss compared with those with persistently normal NT. Recent prospective and retrospective analyses emphasize that increased NT represents a non-specific but significant marker of early embryonic developmental disturbance rather than an isolated finding (1, 2). Studies combining advanced genetic testing methods-such as chromosomal microarray and whole-exome sequencing-have revealed that some cases of increased NT in chromosomally normal fetuses may be associated with submicroscopic or monogenic disorders (3, 4). Therefore, comprehensive follow-up involving detailed anomaly scans, fetal echocardiography, and, when indicated, molecular testing is recommended to clarify prognosis and guide parental counseling (5). Given the heterogeneity in outcomes and the limited data regarding pregnancy prognosis among fetuses with increased NT but normal karyotype, further research focusing on detailed obstetric and neonatal outcomes is warranted. The present study aims to describe the demographic, obstetric, and perinatal characteristics of pregnancies with increased NT and to evaluate the relationship between NT measurement and subsequent pregnancy outcomes.

MATERIALS AND METHODS

Study design and setting

This retrospective study was conducted at the Department of Perinatology, Firat University Faculty of Medicine, Firat University, Turkey. The study included pregnant women who were referred to the perinatology clinic between February 2023 and July 2025 due to increased NT detected during first-trimester screening ultrasound. Ethical approval was obtained from the Firat University Non-Interventional Research Ethics Committee (approval no: 15/31, dated 23.10.2025). The study was conducted in accordance with the Declaration of Helsinki.

Study population

The study included pregnant women who were referred for the first-trimester combined screening test and were found to have increased NT measurements. An NT value of ≥ 3.0 mm, corresponding approximately to the 99th percentile for crown-rump length, was accepted as the cut-off for increased NT in accordance with previous studies (6). Cases with incomplete data or multiple pregnancies were excluded from the study.

Data collection

Patient data were obtained from the electronic hospital database and perinatology registry books. Demographic characteristics including maternal age, gestational week at procedure, gestational week at delivery, and birth weight were recorded. In addition, ultrasonographic findings, invasive diagnostic procedures such as chorionic villus sampling (CVS) and amniocentesis, and their respective results were reviewed. Chromosomal analysis results including karyotype findings (normal, trisomy 21, trisomy 18, Turner syndrome, triploidy, duplication, and deletion) and perinatal outcomes (termination, intrauterine fetal loss, live birth, and neonatal intensive care unit admission) were retrieved. The presence and distribution of associated ultrasound findings such as cystic hygroma, cardiac, central nervous system, and urinary system anomalies, minor markers, and other malformations were also documented.

Ultrasound evaluation

All ultrasonographic examinations were performed using a Voluson E 6 ultrasound device (GE Healthcare, USA) equipped with a 4-8 MHz transabdomi-

nal probe. Nuchal translucency was measured according to the ISUOG (2023) practice guidelines, in the midsagittal plane where the fetal head and upper thorax were visualized. The NT measurements were obtained in the sagittal plane of the fetus, according to the standardized guidelines, by a single experienced operator (Ç.A.) to minimize interobserver variability.

Invasive diagnostic procedures

All patients with increased NT were counseled regarding the potential association with chromosomal abnormalities and were offered invasive prenatal diagnostic testing. The type of invasive procedure was determined based on the placental location. CVS was performed in cases where the placenta was located anteriorly and accessible. Amniocentesis was carried out for patients with posteriorly positioned placentas or when CVS was technically not feasible. Procedures were performed by experienced perinatologists under sterile conditions, and informed consent was obtained from all participants.

Following invasive sampling, chromosomal analysis was performed using both conventional karyotyping and CMA, which was considered the minimum diagnostic standard for all cases. WES was selectively considered in cases with significantly increased NT measurements, normal karyotype and CMA results, and a clinical suspicion of monogenic disorders based on ultrasound findings or pregnancy course.

Some patients declined invasive testing after counseling and were followed up with non-invasive prenatal testing or serial ultrasonographic evaluations according to clinical guidelines.

Outcomes

The primary outcomes of the study included the types and frequencies of chromosomal abnormalities identified in fetuses with increased NT, the rate and distribution of invasive procedures, and the overall pregnancy outcomes. Pregnancy outcomes were categorized as live birth, termination, or intrauterine fetal loss, and the need for neonatal intensive care unit (NICU) admission was also evaluated. Secondary outcomes involved the relationship between NT thickness and the presence of additional structural or soft ultrasound markers. These outcomes were used to assess the diagnostic contribution of NT measurement to prenatal evaluation and the prognostic value of increased NT in determining fetal and neonatal prognosis.

Statistical analysis

All statistical analyses were performed using SPSS version 22.0 (IBM Corp., Armonk, NY, USA). Continuous variables were expressed as median (interquartile range), and categorical variables as number (percentage). Descriptive analyses were

used to summarize NT values, chromosomal results, ultrasound findings, and pregnancy outcomes.

RESULTS

A total of 39 patients referred to the perinatology clinic due to increased nuchal translucency were evaluated. The median nuchal translucency (NT) value was 4.0 mm (interquartile range, 3.3-5.0 mm). The median maternal age was 32.7 years (27.0-37.0), and the median gestational week at the time of the procedure was 12.2 weeks (12.0-12.5). The median gestational week at delivery was 38.5 weeks (38.0-39.3), and the median birth weight was 3050 g (2800-3300). CVS was performed in 22 cases (56.4%), whereas amniocentesis was performed in 5 cases (12.8%). Additional ultrasound findings were detected in 25 fetuses (64.1%). Among the fetuses, 20 (51.3%) were female and 19 (48.7%) were male. The rate of cesarean delivery was 62.5% (n=20), whereas 37.5% (n=12) delivered vaginally. Regarding pregnancy outcomes, 17 cases (43.6%) resulted in term delivery, 16 (41.0%) were terminated, and 6 (15.4%) resulted in intrauterine fetal loss. Neonatal intensive care unit (NICU) admission was required in 3 newborns (17.7%) (Table 1).

Table 1. Descriptive characteristics of the study population.

Variable	Median (Q1-Q3) / n (%)
Nuchal translucency (mm)	4.0 (3.3-5.0)
Maternal age (years)	32.7 (27.0-37.0)
Gestational week at procedure	12.2 (12.0-12.5)
Gestational week at delivery	38.5 (38.0-39.3)
Birth weight (g)	3050.0 (2800.0-3300.0)
Chorionic villus sampling (CVS)	22 (56.4%)
Amniocentesis	5 (12.8%)
Presence of additional ultrasound findings	25 (64.1%)
Fetal sex	
Female	20 (51.3%)
Male	19 (48.7%)
Mode of delivery	
Vaginal delivery	12 (37.5%)
Cesarean section	20 (62.5%)
Pregnancy outcome	
Term delivery	17 (43.6%)
Termination	16 (41.0%)
Intrauterine fetal loss	6 (15.4%)
NICU admission	3 (17.7%)

Values are presented as median (interquartile range) or number (percentage). NICU: Neonatal intensive care unit.

Among the fetuses with increased NT, additional ultrasound abnormalities were observed in 25 cases (64.1%). The most common finding was cystic hygroma, detected in 14 fetuses (35.9%). Minor markers, including soft sonographic findings such as echogenic intracardiac focus or mild pyelectasis, were present in 7 cases (17.9%). Structural anomalies were also identified, including urinary system anomalies in 3 fetuses (7.7%), central nervous system anomalies in 2 (5.1%), and cardiac anomalies in 1 (2.6%). Hydrops fetalis was observed in 1 fetus (2.6%), while other less frequent abnormalities were documented in 4 cases (10.3%) (Table 2).

Table 2. Distribution of additional ultrasound findings

Ultrasound finding	n (%)
Cystic hygroma	14 (35.9%)
Cardiac anomaly	1 (2.6%)
Hydrops fetalis	1 (2.6%)
Urinary system anomaly	3 (7.7%)
Central nervous system anomaly	2 (5.1%)
Presence of minor marker(s)	7 (17.9%)
Other findings	4 (10.3%)

Values are presented as number (percentage). Minor markers include soft ultrasound findings such as echogenic intracardiac focus or mild pyelectasis.

In our study, when NT thickness was evaluated categorically (n =39), approximately two-thirds of the cases were found to be ≤ 4.4 mm (3.0-3.5 mm: 30.8%; 3.5-4.4 mm: 33.3%; cumulative rate: 64.1%). Cases with NT values ≥ 4.5 mm constituted 35.9% of the total cohort (4.5-5.4 mm: 12.8%; 5.5-6.4 mm: 15.4%; >6.5 mm: 7.7%), indicating that a significant portion were in the markedly increased NT group (Table 3).

Table 3. Distribution of Nuchal Translucency Thickness Categories

NT range (mm)	n (%)
3.0–3.5	12 (30.8%)
3.5–4.4	13 (33.3%)
4.5–5.4	5 (12.8%)
5.5–6.4	6 (15.4%)
>6.5	3 (7.7%)

This distribution indicates that a considerable proportion of the cohort belonged to the high-risk NT category.

Karyotype analysis was successfully performed in 39 fetuses who underwent invasive prenatal testing. Normal chromosomal results were observed in 17 cases (43.6%), whereas chromosomal abnormalities were identified in 22 fetuses (56.4%). The most frequent abnormality was trisomy 21, detected in 14

fetuses (35.9%), followed by trisomy 18 in 3 fetuses (7.7%). Less common chromosomal abnormalities included triploidy in 1 case (2.6%), Turner syndrome in 1 (2.6%), duplication in 1 (2.6%), and deletion in 2 (5.1%) cases (Table 4).

Table 4. Distribution of karyotype results

Karyotype result	n (%)
Normal	17 (43.6%)
Trisomy 21	14 (35.9%)
Trisomy 18	3 (7.7%)
Triploidy	1 (2.6%)
Turner syndrome	1 (2.6%)
Duplication	1 (2.6%)
Deletion	2 (5.1%)
Total	39 (100.0%)

Values are presented as number (percentage).

DISCUSSION

In the present study, we evaluated the prenatal and perinatal outcomes of pregnancies referred to the perinatology clinic due to increased NT thickness. The median NT value in our cohort was 4.0 mm, and more than half of the cases underwent invasive diagnostic testing. Chromosomal abnormalities were identified in approximately half of the fetuses, with trisomy 21 being the most common, followed by trisomy 18 and other structural chromosomal anomalies. Additional ultrasound abnormalities were present in nearly two-thirds of the cases, most frequently cystic hygroma and minor sonographic markers, while a smaller proportion exhibited urinary, central nervous system, or cardiac anomalies. Although a considerable number of pregnancies resulted in live births, the rates of pregnancy termination and intrauterine fetal loss remained high, reflecting the heterogeneous prognosis associated with increased NT. These findings highlight that increased NT values, even when measured during early gestation, continue to represent a significant predictor of chromosomal abnormalities and adverse perinatal outcomes.

In the present study, NT values of ≥ 4.5 mm constituted 35.9% of the cohort, indicating that more than one-third of fetuses fell within the high-risk category for genetic or structural pathology. Recent evidence supports a stepwise association between increasing NT thickness and adverse outcomes, independent of chromosomal status, suggesting that NT is a marker of generalized embryological dysregulation rather than a condition-specific finding (6). Increased NT, typically defined as a thickness above the 95th or 99th percentile for crown-rump length, has been shown to correlate strongly with chromosomal abnormalities, particularly trisomy 21, 18, and 13 (7). Beyond aneuploidy, increased NT is also associated with structural malformations-most notably congenital heart defects-and a variety of gene-

tic syndromes, including monogenic and submicroscopic chromosomal disorders (6). Even in fetuses with normal karyotypes, an increased NT measurement carries a higher risk of miscarriage, intrauterine fetal demise, and adverse perinatal outcomes (6, 7). Kelly et al. (8) highlighted that fetuses with NT measurements exceeding the 99th percentile are at increased risk of RASopathies, congenital cardiac defects, and single-gene disorders, even in the absence of detectable chromosomal abnormalities, underscoring the importance of molecular testing and advanced imaging in this population. Therefore, NT assessment serves as an early, non-invasive, and sensitive indicator that guides further diagnostic evaluation, including invasive testing, detailed anatomical scanning, and, when indicated, molecular genetic analysis (2, 5). Sharifzadeh et al. (9) reported that reliance on a single NT cutoff may underestimate risk in cases with progressive NT elevation, advocating instead for percentile-based stratification to optimize prognostic accuracy.

Although the proportion of fetuses with a normal karyotype in our study was 43.6%, the high rate of additional ultrasound findings in these cases, at 64.1%, and the significantly higher rate of intrauterine loss and neonatal morbidity, demonstrate that increased NT is not merely an isolated marker associated with chromosomal abnormalities but also a nonspecific but strong indicator of structural or functional disorders occurring during early embryonic development. Similarly, the literature has reported that increased NT in fetuses with a normal karyotype is significantly associated with cardiac anomalies, genetic syndromes, and intrauterine fetal loss (10). Bilardo et al. (10) emphasized that a structural or genetic anomaly develops in 32% of cases with a normal karyotype and $NT \geq 3$ mm, and that this condition is an early reflection of embryonic developmental disorder. Van Vugt et al. (11) reported that the risk of intrauterine death and neonatal loss was significantly increased in fetuses with increased NT and normal karyotype, even if no structural defects were detected in follow-up ultrasounds. Additionally, studies in large cohorts have shown that the risk of pregnancy loss, fetal anomalies, cardiac malformations, and preterm birth increases linearly with increasing NT thickness (2, 12). One of the key findings of our study is the high incidence of adverse pregnancy outcomes even in fetuses with normal karyotypes. Additional ultrasonographic findings were detected in 64.1% of cases with normal karyotypes, and the rates of intrauterine fetal loss and neonatal morbidity were significantly increased in this group. This finding suggests that increased NT may be an indicator not only of chromosomal abnormalities but also of structural or functional disorders in early embryonic development. Normal genetic test results should not be considered an absolute indicator of good prognosis in the presence of increased NT, and these cases

should be monitored with close ultrasonographic follow-up. Based on these findings, it can be concluded that increased NT reflects a multifactorial pathophysiological process due to hemodynamic disorders, lymphatic drainage insufficiency, or connective tissue defects that occur during embryonic development, and therefore should be carefully evaluated in clinical follow-up even if the karyotype is normal (9).

In the current study, all fetuses with increased NT underwent karyotyping and chromosomal microarray analysis. In our cohort, chromosomal microarray analysis provided an additional diagnostic yield of approximately 7.7% beyond conventional karyotyping, identifying submicroscopic copy number variations that would not have been detected by standard cytogenetic analysis alone. This allowed for comprehensive evaluation of chromosomal and submicroscopic copy number variations in the cases. WES was performed in two cases, one with a normal sequencing result and a healthy birth at term, and the other with an intrauterine fetal demise at 27 weeks' gestation despite a normal WES result. These findings are consistent with the idea that increased NT is the result of a wide range of genetic etiologies rather than a single disease entity. Recent studies have shown that CMA provides an incremental diagnostic yield of 4-6% in fetuses with increased NT and a normal karyotype, and that detection rates increase in parallel with NT thickness (13). In a 2023 meta-analysis by Girolamo et al. (14) it was stated that WES provided a 3-8% additional diagnostic contribution in cases of isolated increased NT with normal CMA. The same meta-analysis also emphasized that in cases with $NT > 5.5$ mm, the additional diagnostic contribution with WES could reach up to 30-34% (14). In this meta-analysis, it was emphasized that CMA should be considered the minimum diagnostic standard in fetuses with increased NT, but WES or genome sequencing should be selectively recommended in cases with significantly increased NT. In a study published by Choy et al. in 2019 (15), it was reported that when genome sequencing was applied after the standard CMA test in cases of increased NT, the diagnosis rate increased from 16% to 32%.

This study has some limitations. First, the retrospective design of the study limits the possibility of making causal inferences. Second, reflecting a single-center experience and the relatively small sample size may limit the generalizability of the findings. Furthermore, the fact that WES was only applied to a limited number of cases may have prevented the full determination of the true prevalence of monogenic diseases. Nevertheless, despite these limitations, the homogeneous patient group, detailed ultrasonographic evaluations, and systematic genetic analysis approach support the clinical value of the results obtained.

Overall, our findings suggest that increased NT remains clinically relevant even in the absence of chromosomal abnormalities.

Although WES was applied to a limited number of patients in our study, the fact that intrauterine pregnancy loss occurred despite normal sequencing results highlights that increased NT may reflect not only genomic alterations but also early developmental disorders that cannot be fully explained by current genetic testing technologies. Therefore, clinical management should be individualized based on NT thickness, the presence of additional sonographic findings, and longitudinal assessment, rather than

relying solely on karyotyping or molecular testing results.

Conclusion

Increased NT remains a clinically significant finding regardless of karyotype results and is associated with an increased risk of structural anomalies and adverse perinatal outcomes. Chromosomal microarray analysis should be performed in all cases, and whole-exome or genome sequencing may be considered in selected fetuses with significantly increased NT or developing ultrasound abnormalities.

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