First-trimester nuchal translucency measurements: using a transverse or sagittal plane?

TO THE EDITORS: A nuchal translucency (NT) examination is currently a routine first-trimester ultrasound. The presence of a thickened NT (≥3.5 or ≥3.0 mm) is associated with common and uncommon aneuploidies and with a wide variety of genetic syndromes and structural anomalies, even when the karyotype is normal. Measurement of the NT requires specific and standardized assessment and careful attention to technique, which might be time-consuming, especially when the fetus is not well-positioned.

To overcome this limitation, Montaguti et al investigated the ability of a transverse view of the fetal head to detect increased fetal NT at 11 to 13 weeks’ gestation. In a total of 1023 women referred for the first-trimester combined screening, an excellent correlation was found between sagittal and transverse NT measurements. Significantly less time was required to obtain transverse NT measurements than sagittal measurements, particularly when the fetus had an unfavorable position. The authors concluded that increased NT can be reliably identified by using transverse views, and in some cases this may technically be advantageous. However, their study lacked some basic information that is required to sustain the conclusion.

Although measurement of NT in the transverse plane accurately identified cases with a sagittal measurement of ≥3.0, it is unclear how many cases had NT >3.0 mm and >3.5 mm, respectively, in this study. In 45 of 1023 (4.4%) patients, a high risk of trisomies 21, 13 or 18, or both was calculated from either the combined test or noninvasive prenatal testing (NIPT). Were cases with increased NT included among these 45 patients, and how many such cases were there? Was an increased fetal NT an indication of invasive testing in their algorithm? In the 33 patients who decided to undergo chorionic villus sampling, aneuploidies were detected in 6 (18%). Did the 3 cases (of trisomy 18, 9, and 16, respectively) have increased fetal NT measurements or other structural anomalies on first-trimester ultrasound?

NIPT is transforming the landscape of prenatal care. It can be expected that NIPT will ultimately become the primary screening tool for common trisomies. Therefore, we agree with Montaguti et al that routine NT measurement in the era of NIPT screening for aneuploidy might not be necessary. In a screening algorithm with NIPT (Figure), ultrasonographic screening for large NT or other structural anomalies is enough.

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We are grateful to the authors of this letter to the Editor for the interest shown in our study. In our study, we have shown that the identification of increased nuchal translucency (NT) ≥3.0 mm and ≥3.5 mm can also be reliably performed using axial planes in women in the first trimester of pregnancy. Patients were recruited from the population undergoing a larger trial promoted by the health authorities of the Emilia Romagna region to compare the performance of the combined test with that of noninvasive prenatal testing (NIPT) for trisomies of chromosomes 21, 18, and 13. According to the protocol of the study, an invasive procedure was offered to patients that had a positive combined test (risk of trisomy 21 >1:300, risk of trisomy 13 or 18 >1:150), a positive NIPT, or a NT of ≥3.5 mm. Among the 1023 women of our study, 14 (1.4%) had an NT of 3.5 mm, and 2 (0.2%) had NTs of 3.0 mm and 3.4 mm, respectively. Forty-five (4.4%) had a high risk of aneuploidies at the combined test or at the NIPT. All the 16 fetuses with an NT of >3.0 mm were in the latter group of high risk, whereas the other 29 presented an NT within normal ranges. We did not find any fetus with an increased NT and a low risk of aneuploidies by NIPT or combined test. All the 16 fetuses with an NT of ≥3.5 mm had a high risk of aneuploidy and may be addressed directly to the determination of the fetal karyotype. We stress again that our study did not aim to suggest that sagittal planes are replaced by transverse views in the precise quantification of NT required for combined test risk assessment. In pregnancies undergoing NIPT instead, NT assessment may be initiated in the transverse plane, particularly in cases with the fetus in unfavorable position, limiting a sagittal view for cases with an excessive measurement.

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