Invasive diagnosis after first-trimester aneuploidy screening

We are grateful to the authors of this letter to the Editor for the interest shown in our study.1 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. Among the women with high-risk aneuploidy screening, 33 decided to have chorionic villus sampling, with 6 (18%) anomalies detected. There were 3 cases of trisomy 21, all with an increased NT (3.7 and 4.4 mm, respectively, and one with generalized hydrops). The fetus with trisomy 9 had a very enlarged NT with septations and severe hydrops; the pregnancies with trisomy 16 had no anatomic malformation detectable on first-trimester ultrasound.

The results of our study are well-correlated with previous experience that the identification of increased NT is a crucial part of the first-trimester ultrasound,2,3 most of all because these fetuses have 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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REFERENCES