Invasive diagnosis after first trimester aneuploidy screening

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In our paper, we have shown that the identification of increased nuchal translucency (NT), greater than or equal to 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 from the Health Authorities of the Emilia Romagna Region to compare the performance of the combined test with non-invasive 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 greater than 1:300, risk of trisomy 13/18 greater than 1:150), a positive NIPT or a NT of 3.5 mm or more. Among the 1023 women of our study, 14 (1.4%) and 2 (0.2%) an NT of 3.5 mm and among 3.0 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 more than 3.0 mm were in the latter group of high risk, while 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 a high-risk aneuploidies screening, 33 decided to perform chorionic villous sampling with 6 (18%) anomalies detected. There were 3 cases of trisomies 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 septation and severe hydrops; the pregnancies with trisomy 16 had no anatomical malformation detectable on first trimester ultrasound.

Our results 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 aneuploidies and may be addressed directly to the determination of the fetal karyotype. We stress again that the aim of our study was not to suggest that sagittal planes are replaced by transverse views in the precise quantification of NT required for combined test risk assessment. In those pregnancies undergoing NIPT, instead, NT assessment may be initiated in the transverse plane, in
particular in those cases with the fetus in unfavorable position, limiting a sagittal view for cases with an excessive measurement.

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