

EP10.49 – Combined screening test versus non-invasive prenatal test for first-trimester aneuploidy screening: multicenter cohort study from Emilia-Romagna, Italy

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Background:

In recent years, non-invasive prenatal testing (NIPT) analyzing cell-free fetal DNA has emerged as an alternative to first-trimester combined screening (CST) for fetal aneuploidy. While CST relies on maternal serum markers and ultrasound parameters, NIPT offers significantly higher sensitivity and lower false-positive rates for primary trisomies. In 2024, the Emilia-Romagna Region, Italy, introduced universal NIPT without prior CST as the primary prenatal screening strategy for fetal trisomies.

Objective:

To compare the number and indications for invasive procedures in two historical cohorts from consecutive years in a Northern Italian region.

Methods:

Multicenter retrospective observational cohort study, involving 6 referral fetal medicine units in Emilia-Romagna, Italy.

Cohort 1: screening based on the CST;

Cohort 2: screening based on NIPT associated with conventional first-trimester screening ultrasound.

Results:

Figure shows comparison between two cohorts. Moreover, no differences were recorded in terms of rate of first trimester diagnosis or suspicion of fetal anomaly, nuchal translucency > 3.5 mm and fetal anomaly diagnosed in the second trimester.

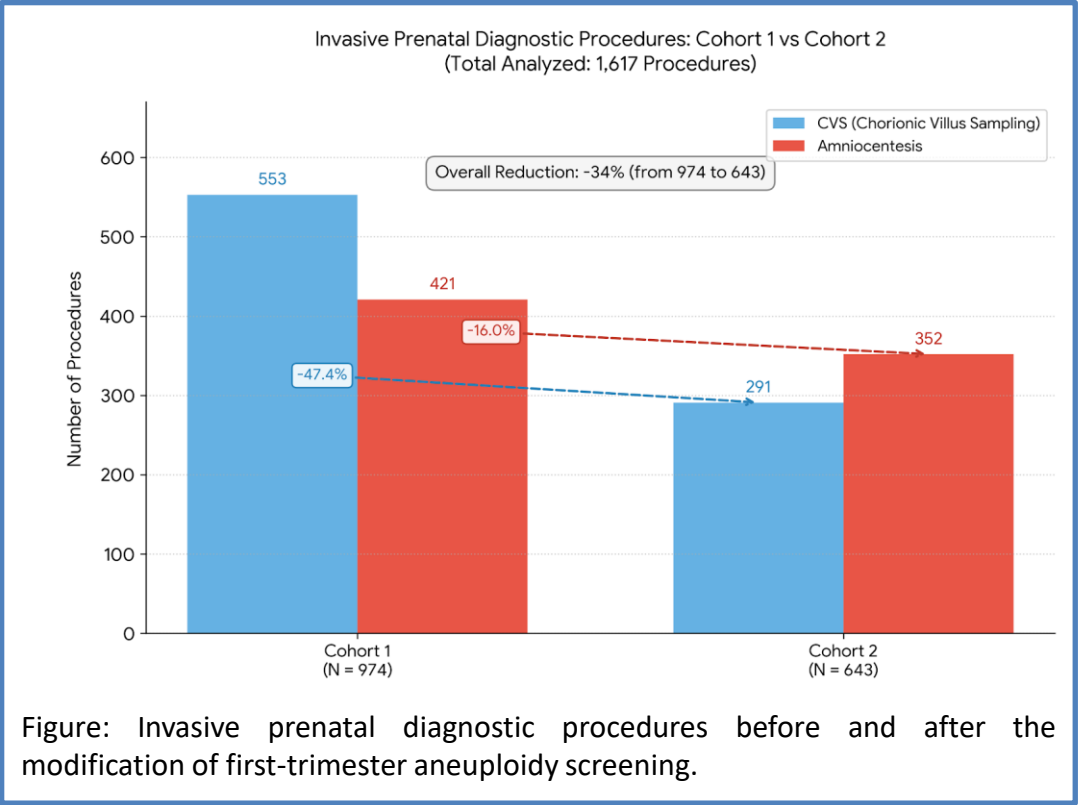


Figure: Invasive prenatal diagnostic procedures before and after the modification of first-trimester aneuploidy screening.

Conclusions

First trimester aneuploidy screening by means of NIPT on a regional level is associated with an over 30% reduction of invasive procedures for prenatal diagnosis and seems not to increase the number of cases of late diagnosis of fetal anomaly and NT>3.5 mm