17th World Congress in Fetal Medicine

# First trimester combined screening for fetal aneuploidies enhanced with additional ultrasound markers: an 8-year prospective study

Nemescu D, Bruma A, Bratie A, Mihaila A, Luca Al
University of Medicine and Pharmacy "Gr. T. Popa, IASI, Romania

## Objective

To describe our screening population and to audit the performance of first-trimester screening for Down syndrome, based on combined test enhanced with additional ultrasound markers, since its implementation.

## Methods

Prospective study performed from 2009 to 2016, which included 1358 singleton fetuses, with a crown-rump length of 45-84 mm. The risk of aneuploidy was calculated using nuchal translucency, fetal heart rate (FHR), additional markers like nasal bone (NB), tricuspid flow (TF) and ductus venosus (DV), combined with maternal serum free β-human chorionic gonadotropin (fβ-hCG) and pregnancy-associated plasma protein-A (PAPP-A).

## Results

87% of patients had an evaluation of all additional ultrasound markers and in 97% at least two markers were assessed, in any combination. 70. 5% of patients were evaluated also through maternal serum biochemistry. The most common risk calculation used nuchal translucency, FHR, all additional ultrasound markers, fβ-hCG and PAPP-A in 851 (62. 7%) of cases. Adjusted risk for trisomy 21 was greater than 1: 100 in 65 (4. 8%) women. From them, 58 (87. 7%) have chosen an invasive test. There were 24 aneuploid fetuses (1. 7%). We identified 12 (50%) trisomy 21, 6 (25%) sex chromosome anomalies, the rest being triploidies and trisomy 18/13. The combined test detected 11 of the 12 cases of trisomy 21, having a first trimester detection rate of 91. 7%. 39 fetuses (2. 8%) had various types of structural anomalies.

## Conclusion

Combined test enhanced with all additional ultrasound markers did not showed a substantially improvement in T21 detection rate, compared to using only one of the additional markers.