NIPT *vs* combinated test for predicting aneuploidies: a case report

Fiorella **Bucci**, Martina **Marmorato** *, Giulia **Calonaci**, Stefano **Masoni**

Department of Gynecology and Obstetrics, Valli Etrusche Hospital, Livorno, Italy.

DOI: 10.36129/jog.2022.S02

Background. Prenatal screening for trisomy 21-18-13 and selected sex chromosome aneuploidies can be performed using next-generation sequencing of cell-free DNA (cfDNA) in the maternal circulation or a combined test with free ß-hCG, PAPP-A dosage and nuchal translucency misuration.

Case presentation. A 32-year-old healthy caucasian woman, first spontaneous pregnancy, performed a non invasive prenatal test (NIPT) at 10 weeks of pregnancy which resulted in low risk for Trisomy 13, 18 and 21 and major aneuploidies. Fetal fraction was 4%.

At 13 weeks, the same patient performed a combined test with high-risk result for trisomy 18 and 21 (1:58 and 1: 204 respectively), low risk for trisomy 13 (1:1049). Despite medical advice, she did not undergo genetic counseling.

At 15 weeks of pregnancy the patient was diagnosed with internal abortion. The response of the molecular investigation on the skin biopsy was: condition compatible with triploid chromosomal structure. Histological examination of the placenta showed no abnormalities.

Conclusions. Triploidy is a rare chromosomal abnormality unrelated to maternal age. The prevalence at 12 weeks is about 1 in 2000 but it is highly lethal and very rarely observed in live births. NIPT has a high predictive value, but it still remains a screening test for the rare false negatives. The reasons for false negatives are: confined placental mosaicism, borderline low fetal fraction, maternal copy number variants, technical issues. In case of discrepancy between the two screening tests, it is always advisable to carry out genetic counseling.