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Prenatal screening for aneuploidies after preimplantation genetic testing

Albaigés G, Prats P, Rodríguez-Melcón A, Palacios G, Rodríguez I, Racca, A, Parriego M, Serra B.
Hospital Universitario Dexeus., Barcelona, Spain

Objective

The main aim of this study is to report the performance of the first trimester aneuploidy screening in pregnancies conceived by in vitro fertilisation (IVF) and preimplantation genetic testing (PGT-A) of transferred embryos in our medical setting. Secondarily, we aim to discuss the need for a different prenatal screening strategy in this specific group of patients.

Methods

Retrospective cohort study in a single tertiary care centre between January 2013 and June 2022. 20,237 women have had prenatal follow-up in our clinical centre and were included in our study. We divided them in 3 groups: singleton pregnancies conceived after the transfer of a PGT-A screened euploid-embryo (n=511), singleton pregnancies conceived after IVF without PGT-A (n= 3,291) and singleton naturally conceived pregnancies (n= 16,436). Maternal demographic characteristics and type of conception were collected from our electronic medical records. PGT-A procedure was performed according to established standard protocols. Prenatal screening of aneuploidies was performed combining maternal serum biomarkers (free-ßHCG and PAPP-A) with ultrasonographic markers (nuchal translucency [NT] and ductus venosus pulsatility index [DV-PI]) and maternal age. The risk assessment was calculated using the conventional combined test in two steps: blood sampling for the study of maternal serum free-ßHCG and PAPP-A was performed between 8^{+0} and 13^{+6}-weeks' gestation and the NT scan was performed between 11^{+0} and 13^{+6} weeks' (crown-rump length (CRL) 45-84 mm). Fetal chromosomal status was determined either by amniocentesis or chorionic villus sampling (CVS) when requested or by phenotypic evaluation after delivery by the attending paediatricians.

Results

The conventional combined test for PGT-A pregnancies had a specificity of 91% [CI95%: 88%-93%] and the sensitivity could not be calculated since there were no cases of fetal aneuploidy in this group. Further analysis was performed to determine the performance of the test in relation to maternal age. The specificity of the test in the PGT-A group decreased from 95.4% [CI95%: 87.1%-99.0%] in women younger than 35-years to 90.3% [CI95%: 87.2%-92.9%] in women of 35 years or older In 89.1% of pregnancies conceived after IVF with PGT-A with a high-risk for T21, 18 or 13, the result was related to advanced maternal age. Regarding the variables studied in the conventional combined first trimester screening test, only significant differences were found in the MoM corrected-Log ß-HCG between pregnant women after PGT-A and women with naturally conceived pregnancies.

Conclusion

The current screening strategy for the viable chromosomal abnormalities (trisomy 21, 18 and 13) combining maternal age with biochemical and ultrasonographic markers has been shown to penalize pregnancies undergoing PGT-A, due to the great weight of age in the a priori risk. These strategies generate unnecessary tests with the consequent economic and emotional cost for patients. We propose abandoning the current strategy and establishing a new protocol for these patients, combining NIPT with first trimester ultrasound markers, which allows for the identification of other genetic or structural conditions that have not been screened by PGT-A and may be present in the fetus.