REVIEW OF NON-INVASIVE PRENATAL TESTING IN CLINICAL PRACTICE

Stratoudakis G, Ebrahim H, Papastamatiou M, Dalakoura D, Archontakis G, Patramani S, Daskalakis G.

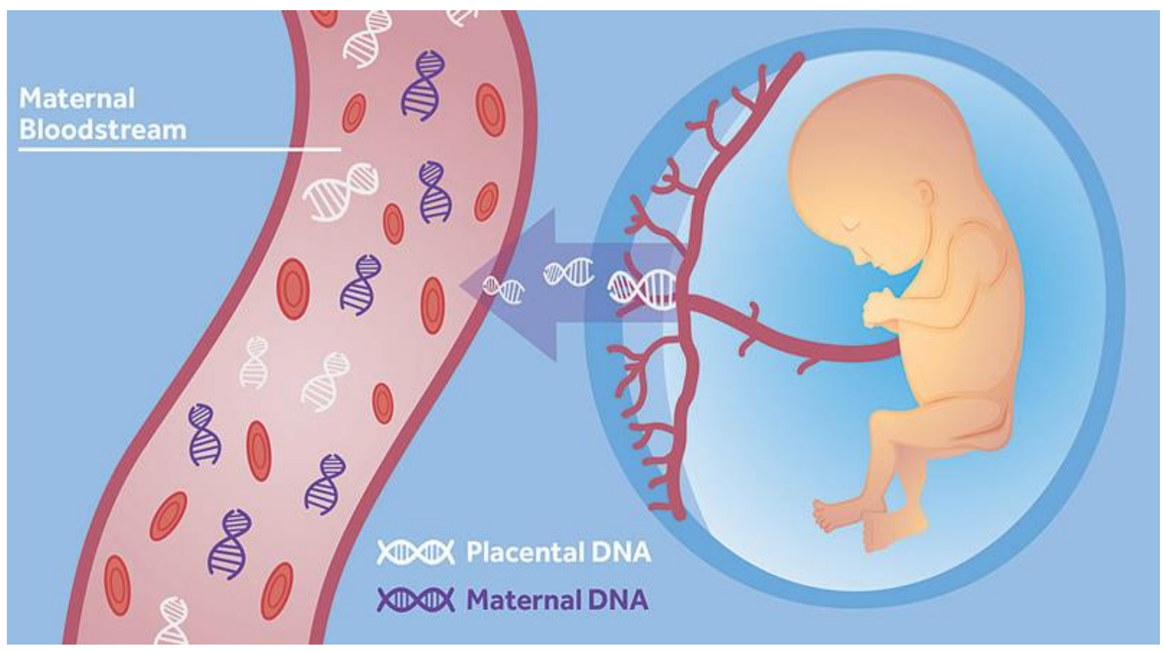
Department of Obstetrics & Gynecology, General Hospital of Chania, Crete, Greece

Introduction

Non-invasive prenatal testing (NIPT) as a screening tool for fetal aneuploidies and other chromosomal anomalies has been gaining ground due to its non-invasiveness, high detection rate and low false-positive rate. NIPT can be offered as a first- line screening test during the first trimester or following a high-risk result in combined first trimester screening. During pregnancy, DNA fragments are released from placental cytotrophoblastic cells into the maternal bloodstream. Advances in genomic sequencing have allowed analysis of the fetal cell-free DNA detected in maternal plasma in order to screen for chromosomal trisomies, deletions, duplications and sex chromosome anomalies. Our objective is to asses the value of NIPT in detecting the aforementioned abnormalities, compared with data from karyotyping and pregnancy outcomes.

Materials and Methods

We studied the files of 1000 patients with singleton pregnancies followed up in our department of Obstetrics and Gynecology in Chania General Hospital, Crete, Greece, from 2016-2019. In the study we included patients in the first and second trimester of pregnancy, who either opted for NIPT before combined first trimester screening, or received it after cFTS results indicated a high risk for chromosomal anomalies. In patients with NIPT results demonstrating a high risk of aneuploidies, deletions, duplications or sex chromosome abnormalities, amniocentesis was offered for karyotype analysis. Pregnancy outcomes were subsequently followed up.



Results

We studied 1003 patients aged 19-46, with a median age of 32.1. In 32 (3.19%) cases, results indicated high risk for trisomies 21, 18 and 13 and another 11 (1.09%) came back as high-risk for sex chromosome abnormalities. Genetic consultation was offered and 40 out of 43 opted for amniocentesis. In 38 cases (95%), chromosomal anomalies were identified, with 16 (42%) accounting for 21 Trisomy, 5 (13.1%) for 18 Trisomy and 2 (5.2%) for 13 Trisomy. Deletions and duplications were found in 6 (15.7%) and 5 (13.1%) cases respectively. Sex chromosome abnomalities were confirmed in 4 cases (10.5%). During follow-up, chromosomal anomalies were not detected in the remaining pregnancies.

Conclusion

As a prenatal screening tool, the positive predictive value of NIPT is very high for trisomies, deletions and duplications and lower for sex chromosome abnormalities. False-positive and false-negative results regarding trisomies are extremely low, which can significantly reduce the number of patients undergoing invasive prenatal testing such as chorionic villus sampling and amniocentesis.