Looking to the experience obtained from implementing Non-Invasive prenatal testing of common chromosomal aneuploidies using cell-free fetal DNA in Iran

Mohammad Keramatipour1*, Daniz Kooshavar[1], Zahra Golchehre[1], Masoumeh Razipour[1]
[1] Department of Medical Genetics, Tehran University of Medical Sciences, Tehran, Iran
* Corresponding author: keramatipour@sina.tums.ac.ir

Introduction:
Conventional prenatal screening of chromosomal abnormalities, relies exclusively on biochemical and sonographic assessment in the first and second trimesters. With a 5% false-positive rate (FPR), conventional screening has a detection rate of about 60–95% for trisomy 21. From 2008 a new method called non-invasive prenatal testing (NIPT) was established which used sequencing of cell-free fetal DNA (cffDNA) in maternal plasma. The main aim was to reduce the incidence of unnecessary invasive procedures and iatrogenic fetal loss. NIPT showed a significantly low FPR.
In recent years, NIPT was introduced in Iran fetal screening market,. The present study was aimed to report the experience with NIPT of common chromosomal aneuploidies based on the analysis of cffDNA in maternal plasma through Massively Parallel Signature Sequencing (MPSS).

Material and methods:
The study was performed between October 2014 and December 2015 (a period of 15 months). A total number of 514 NIPT screenings were performed in 14 centers, using NIFTY test for trisomies 21, 18, 13 and sex chromosome aneuploidies (SCA).
This test is based on low-coverage whole genome MPSS of the cell-free DNA in maternal plasma. All the results were then validated by either Karyotyping/QF-PCR or pregnancy outcome follow-ups.

Results:
Overall 23 out of 514 tests (4.47%) was failed in the first attempt and required repeated blood sampling. One test was repeated twice. Test repeats were successful and there were no test failures.
Outcome data were available in 273 out of 514 NIPTs (53.11%) to the date of writing this report. Trisomy 21 was confirmed in 7 out of 8 cases with positive T21 results among 273 NIPTs. Sensitivity and specificity for T21 screening were 100% and 99.62% respectively. One XYY result was also reported and confirmed.

Conclusion:
Due to high sensitivity and specificity obtained in this study we conclude that NIFTY is a reliable NIPT for screening common chromosomal aneuploidies. More samples and outcome data during the time will indeed provide more accurate evaluation of the test accuracy.