Our experience in applying non-invasive prenatal testing

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Objective

The growing application of prenatal testing has completely changed by the arrival of Invasive Prenatal Tests (NIPT). All of them are based on the analysis of fetal DNA (cell free fetal DNA- cf DNA). When compared with previous non-invasive screening tests, non-invasive prenatal tests have significantly higher accuracy in detecting most common chromosomal trisomies, fetal sex, sexual chromosomal aneuploidy and chromosomal deletions.

Methods

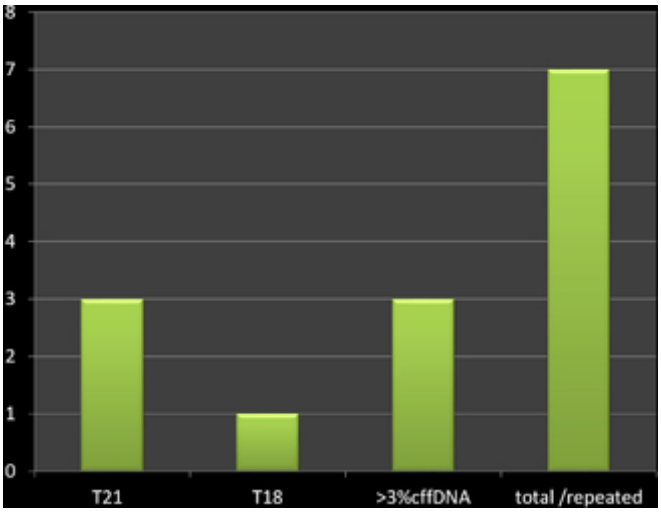
We tested 108 pregnant women between May 2014 and October 2016. 103 women had singleton pregnancies and five carried twins. Blood samples were taken during the 10th and 18th week of pregnancy. Previous conventional screening tests were not preformed on all participants. 57 had a first assessment of the risk in the first quarter, on the basis of age (35 and over) and nuchal translucency. A combined first-trimester screening test, based on age risk, NT and biochemistry (cut off 1/ 250), showed a high-risk result for 29 women. In addition, 22 women without high risk factors, decided for NIPT for personal reasons. Before giving a blood sample (10 ml of venous blood), all patients were informed about non-invasive prenatal testing and signed the consent. An online questionnaire on previous and current pregnancies was completed. Samples were analyzed in relevant diagnostic laboratories. Patients with high-risk results NIPT had amniocentesis (invasive diagnostic procedures). Patients with low risk results had the usually antenatal care.

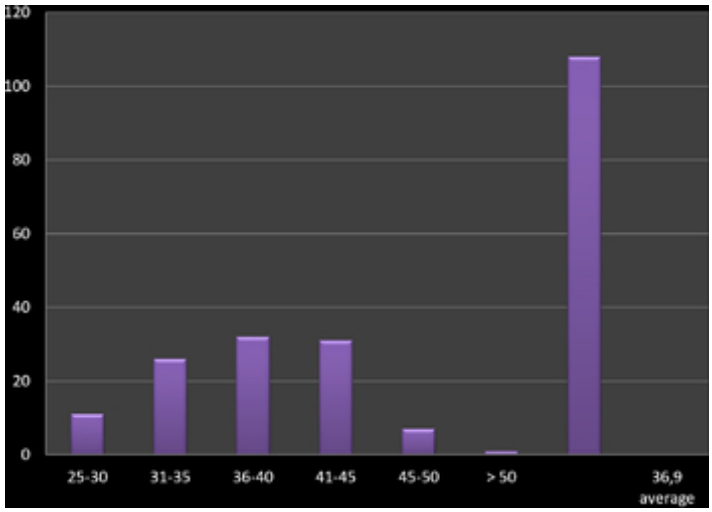
Results

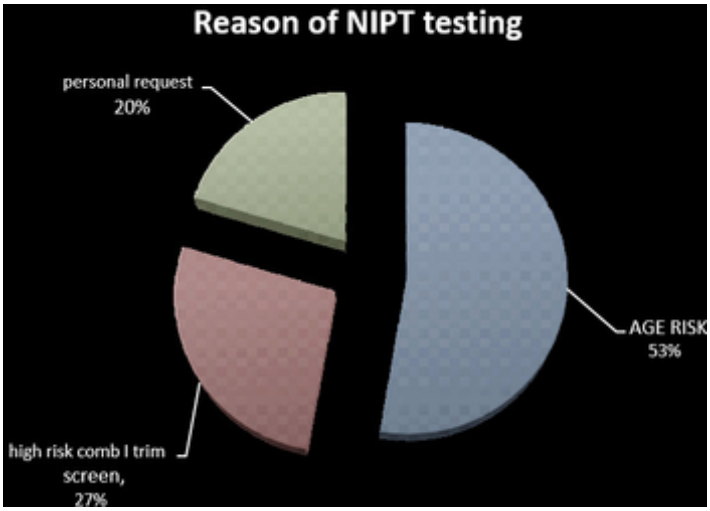
The youngest tested patient was 25 and the oldest was 52 years (egg donation). The average age tested was 36,9 years. Results were awaited for 8-10 days. The test was repeated in 7 cases; in 3 cases due to high risk for T21, in 1 case due to high risk for T18, which was confirmed by amniocentesis and in 3 cases due to cff DNA concentration being less than 3%. Repeated analysis showed low risk. There were no false positive or false negative results.

Conclusion

One of the biggest values in almost every parent's life is having a healthy child. Non-invasive prenatal testing in routine clinical practice could significantly reduce the number of unnecessary amniocentesis/diagnostic invasive procedures that could lead to fetal loss. NIPT is used both as a first and second line screening test for aneuploidy. Unfortunately, the application of this test is still dependent on financial capabilities rather than medical indications.






Reason of NIPT testing
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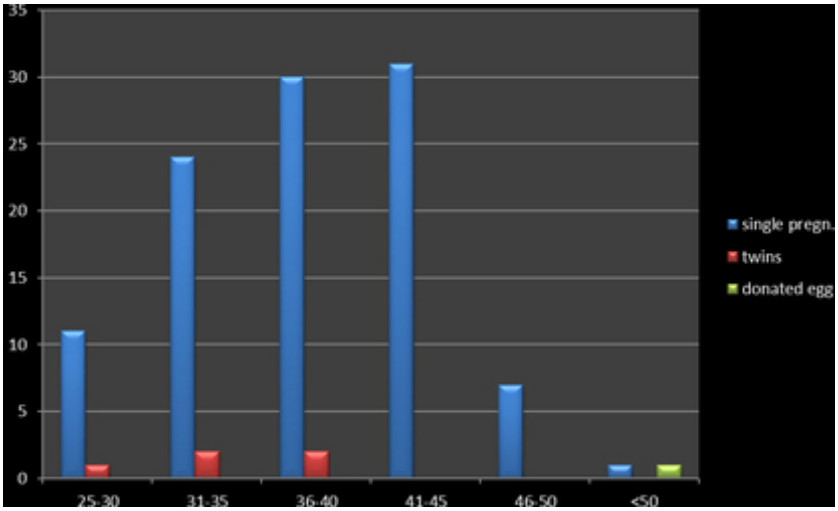
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