



## **Non-invasive prenatal testing as a primary screening method for common foetal aneuploidies in a general Belgian population**

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### **Objective**

Non-invasive prenatal testing (NIPT) using circulating cell-free DNA is reimbursed by the Belgian healthcare system since July 2017 as a first-line screening method for trisomy 21, 18, and 13. The aim was to compare NIPT with combined first or second trimester serum screening in a general population of pregnant women.

### **Methods**

We report a single-centre retrospective analysis of the general pregnant population at CHR Citadelle Hospital in Liege. A first group of 3, 654 patients having had NIPT screening between 01/07/2017 and 31/12/17 was compared to second group of 2, 160 women having had combined first or second trimester serum screening between 01/01/16 and 31/12/2016. Analysis were performed with the Harmony Prenatal Test from Roche-Ariosa for the NIPT and Delfia-Express from Perkin-Elmer for the combined first or second trimester serum screening. The indications for prenatal invasive procedures were for NIPT an abnormal result and for combined testing a risk of  $> 1/250$ .

### **Results**

There was no significant difference in demographics of the two groups (age, patient weight, number of twin pregnancies and IVF-treatment). For combined testing, 92.08% of the patients were screening negative, 2.78% were screening positive and 5.14% had dubious results (low risk for combined test with NT, but high risk biochemical test without NT). In the NIPT-group, 99.23% were screened low risk, 0.27% at high risk (T21, T18 and T13) and 0.27% ended after at least two blood samples with inconclusive results. The number of needed invasive procedures to clarify the screening results passed from 2.78% in the combined screening group to 0.63% for the NIPT group (high risk and inconclusive result). This difference is significant ( $p < 0.0001$ ).

### **Conclusion**

NIPT for the screening of common foetal aneuploidies in a general pregnant population is feasible as a first line test. A significant reduction of invasive testing has been observed in the NIPT group compared to combined first or second trimester serum screening. This goal of the Belgium health care system in starting the reimbursement of NIPT for every pregnant woman seems to be achieved.