Objective
NIPT was introduced in the first trimester screening program for Trisomy 21 in the Northern Region of Denmark in March 2013. In each high-risk pregnancy, defined as the risk of Trisomy 21 ≥ 1/300 (combined screening), the woman is offered additional prenatal diagnostic testing of either chorionic villous sampling (CVS) or NIPT. If the nuchal translucency is increased above 3.5 mm, the woman is advised to have a CVS for a full fetal karyotype, as this subgroup of woman are supposed to have an increased risk of chromosomal abnormality other than Trisomy 21. The aim of this study was to investigate, how the implementation of NIPT changed the prenatal diagnosis of Trisomy 21 among high-risk singleton pregnancies in the Northern Region of Denmark.

Methods
Singleton high-risk pregnancies were identified by combined screening in the following two groups: Group 1 (before the implementation of NIPT) 1/3 2011 – 1/2 2013 and Group 2 (after the implementation of NIPT) 1/3 2013 – 1/2 2015. In each group, the distribution of prenatal tests were described.

Results
In each group the total of singleton high-risk pregnancies were; Group 1, n=253 (screen positive rate: 2.79%) and Group 2, n=302 (screen positive rate: 3.42%). The distribution of diagnostic prenatal tests was investigated in each group. The implementation of NIPT was followed by a reduction in the invasive rate among high-risk singleton pregnancies from 70% to 48% (p<0.001). The standard neonatal examination did not reveal any clinical signs of undetected chromosomal abnormalities in the NIPT group. In the NIPT group, four cases of trisomy 21 were identified by NIPT and in each case the diagnosis was confirmed by amniocentesis or at birth. Furthermore, three cases of sex chromosomal abnormalities were identified by NIPT; two cases of Monosomy X and one case of Triploidy X. CVS confirmed only one case of Monosomy, and the last two cases were false positives.

Conclusion
The implementation of NIPT in the screening program for Trisomy 21 in the Northern Region of Denmark was followed by a markedly reduction in the invasive rate, without any clinical increase of undetected chromosomal abnormalities. NIPT successfully detected four cases of Trisomy 21, however two out of three sex chromosomal abnormalities were false positives.