RESULTS OF FIRST YEAR NIPT FOR ALL PREGNANT WOMEN IN THE NETHERLANDS

Non-invasive prenatal testing (NIPT) is being offered as a first-tier test to all pregnant women in The Netherlands since April 2017. Now, Karuna van der Meij et al. have published the results of the first year of NIPT (TRIDENT-2 study, licensed by the Ministry of Health), based on 73,329 pregnancies (The American Journal of Human Genetics (2019). Of all pregnant women, 42% participated and of these, 78% opted for genome-wide NIPT. Because of intensive cytogenetic follow-up studies in case of abnormal findings, this study provides an accurate perspective on the findings that can be expected in a low-risk population in addition to the common trisomies (13, 18, 21). The percentage of additional findings (0.36%) was in the same order of magnitude as trisomy 21 (0.33%) and included cases with confined placental mosaicism, other trisomies in the fetus, chromosomal rearrangements in the mother and - in rare cases complex profiles suggestive of a maternal malignancy. This study provides an input for the debate on whether NIPT should be performed genomewide or should be restricted to the trisomies. There is a trade-off between the benefits of finding other chromosomal anomalies in the fetus on the one hand and, on the other, increased parental anxiety and unnecessary invasive procedures.