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## **Implementing Non-Invasive Prenatal Testing (NIPT): Perspectives of patients and professionals**

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The introduction of Non-Invasive Prenatal Testing (NIPT), also referred to as cell-free DNA (cfDNA) testing, has created a significant change in prenatal care, affecting several stakeholder groups. The main objective of this thesis was to gain insight into the perspectives of stakeholders involved in the implementation process of NIPT. The studies in this thesis focused on the attitudes of pregnant women and their experiences with being offered NIPT and also explored the opinions of health professionals and parents of children with Down syndrome.

## **PART I. TOWARDS IMPLEMENTATION OF NON-INVASIVE PRENATAL SCREENING**

**Chapter 2** describes the attitudes of pregnant women and male partners towards NIPT for fetal aneuploidy and for a wider range of disorders in the future. Their attitudes were explored through (online) focus groups (n=47 participants) and individual interviews (n=13). It was evident that participants preferred NIPT over the first-trimester combined test (FCT) because it allows for more accurate, safe and earlier testing. Participants thought that NIPT would therefore lower the barrier for screening. Concerns were expressed about potential uncritical use of NIPT and pressure to accept this nearly perfect test. Pregnant women thought testing for a wider range of disorders with NIPT could be beneficial for severe disorders, but believed there should be a limit to what it tests for and that testing for minor abnormalities should be avoided.

**Chapter 3** presents the results of an online questionnaire on the Dutch pregnancy fair website, completed by 381 pregnant women, which explored their opinions on the impact of NIPT and the use of NIPT for a wider range of disorders. About half (51%) of the pregnant women were interested in having NIPT, including 33% of women who had declined FCT, and most believed that the uptake of prenatal screening would increase with the introduction of NIPT. If NIPT were to be applied for broader testing, they thought it should be allowed for life-threatening and/or severe mental- or physical disorders. Women feared that offering testing for more disorders could result in information overload, but nonetheless preferred to have an individual choice from a list of disorders when offered this broader test.

**Chapter 4** combines data from the online questionnaire on the Dutch pregnancy fair website and focus groups among pregnant women. The aim of this study was to explore women's reasons for declining FCT and how these relate to accepting or declining future (hypothetical) use of NIPT. This study demonstrated that women declining FCT for reasons relating to attitudes towards Down syndrome and termination of pregnancy are more likely to decline NIPT as well or see its value in preparing for the birth of a child with a disability, while women declining for test-related reasons are more likely to accept the 'safer' NIPT test. Based on these results, uncritical use of NIPT

is not likely, provided that women have the opportunity to reflect on their personal values.

**Chapter 5** describes a questionnaire study among 240 obstetric health professionals which aimed to investigate their opinions on offering NIPT as a first-tier screening test for fetal aneuploidy and as a test for a wider range of disorders in the future. Most health professionals (72%) believed that NIPT should replace the FCT, although 43% preferred to maintain the nuchal translucency measurement. It was expected that the use of NIPT would lead to an increased uptake of prenatal screening. Participants also believed that NIPT is an easier test to explain, thereby simplifying counseling. If NIPT were to be offered as a broader test, health professionals – in contrast to pregnant women- thought this should be done as a fixed list of disorders, and the great majority agreed to screening for disorders characterized by neonatal death or disorders for which children would need medical care throughout their lives.

In **Chapter 6** a qualitative study using focus groups (n=16 participants) and interviews (n=11) to explore the attitude of parents of children with Down syndrome towards NIPT is described. Although some parents were critical about the practice of screening for Down syndrome, they did think NIPT to be beneficial as it enables people to know whether the fetus is affected and to prepare themselves without risking miscarriage. Parents seemed concerned about the uncritical use of NIPT and feared that a higher abortion rate caused by NIPT would lead to less acceptance and facilities for children with Down syndrome, resulting in more people deciding to screen. The importance of good counseling and balanced, accurate information about Down syndrome was stressed. Parents were positive towards testing for more disorders since they believed that this could divert the focus away from Down syndrome. In contrast, it was also perceived as worrisome due to the potential loss of diversity in society and the uncertainty of where it would all end.

## **PART II. EVALUATION OF THE IMPLEMENTATION AND OFFER OF NON-INVASIVE PRENATAL TESTING**

**Chapter 7** discusses the implementation of NIPT in Dutch prenatal care by describing the constraining and enabling factors and the conditions for successful implementation. A mixed methods approach involving stakeholder interviews (n=13), document analysis and (participatory) observations of the Dutch NIPT Consortium meetings was used to generate findings, which were then interpreted with the use of the Diffusion of Innovation Theory and a Network of Actors model. Results showed that implementing advanced technologies such as NIPT benefit from a learning phase in which the technical performance and women's experiences can be evaluated. Such a

coordinated learning phase, involving all stakeholders, stimulates the process of responsible implementation.

**Chapter 8** reports preferences and decision-making among women at increased risk for fetal aneuploidy based on first-trimester combined testing ( $\geq 1:200$ ) or medical history offered NIPT as a contingent screening test in the context of a nationwide implementation study (TRIDENT study). Results of a questionnaire, filled out by 1091 women after pre-test counseling, showed that the great majority prefer NIPT over invasive testing (amniocentesis or chorionic villus sampling) because it is safer. Most women (90%) had sufficient knowledge on NIPT and 78% made an informed choice. Informed choice was associated with less decisional conflict and less anxiety. Women with a low level of education and women with inadequate health literacy were less likely to make an informed choice. It was shown that women who chose NIPT had less intention to terminate their pregnancy in the case of Down syndrome compared to women choosing invasive testing, possibly indicating that they more often undergo NIPT just to prepare themselves. Continuous attention for counseling is required, also outside the study context, especially for low-educated and less health-literate women.

**Chapter 9** describes high-risk pregnant women's feelings, emotional well-being and satisfaction after receiving NIPT results. A questionnaire filled out after receiving test results ( $n=682$ ) showed that the great majority of women (96%) were glad to have been offered NIPT and did not regret having had NIPT. The waiting time for results was perceived as too long by most women (68%). It was shown that women were reassured by a normal NIPT result and that their level of anxiety returned to normal. Women who had inadequate health literacy and those who had NIPT based on a medical history experienced significantly higher anxiety after a normal NIPT result compared to those with adequate health literacy and no medical history. These women might benefit from extra explanation or guidance after receiving NIPT test results.

## **CONCLUDING REMARKS**

Stakeholders are interested in NIPT and testing for a wider range of disorders. Introducing NIPT as an alternative to invasive testing for pregnant women at high-risk for fetal aneuploidy within a study context assisted in a responsible implementation and led to an offer that satisfied women and allowed them to make an informed choice. Concerns about routinization or uncritical use and pressure to test when implementing NIPT as a first-tier screening test should be avoided by safeguarding informed decision-making through the training of healthcare professionals and realizing effective pre-test counseling. Innovative strategies and counseling aids could be helpful, especially, but not exclusively, for women with low educational levels and/or inadequate health

literacy. When testing for a wider range of disorders, it is important to find a way that facilitates rather than undermines well-informed decision-making.