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OPEN PEER COMMENTARIES

Delineating the Scope of NIPT: Ethics Meets Practice

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As noninvasive prenatal testing (NIPT) is being implemented as a first-trimester prenatal screening modality in healthcare systems around the world, it raises ethical concerns. In theory, NIPT allows for interrogation of the entire fetal genome. While in previous years, first-trimester prenatal screening was aimed at the detection of a limited set of chromosomal abnormalities, it is now possible to use NIPT to detect copy number variants, single-gene disorders, and microdeletion syndromes. Thus, in the future, NIPT may enable pregnant women and their partners to learn all kinds of information about their future child. This information may be useful for reproductive decision-making, as it may provide reasons for women or couples to consider termination of pregnancy. However, if pregnant women or couples decide, after screening, to continue their pregnancies, children may grow up knowing about future disease risks, which may be burdensome and harmful. Also, this may violate children's right to an open future, as it takes away from children the opportunity to decide for themselves whether or not they want to learn this information. Thus, parental access to fetal genomic information may need to be restricted—mainly, as argued by Bayefsky and Berkman, for these two reasons: the best interests of future children and their anticipatory autonomy interests (Bayefsky and Berkman 2022). They present a framework for delineating an ethically appropriate scope of NIPT, distinguishing information that should be recommended from information that should not, which should be upheld by physicians' associations, not by the state. I draw on recent experiences with NIPT in my country, the Netherlands, to argue that that there *is* an ethical (and legal) basis for state intervention in limiting parental access to the fetal genome, that healthcare professionals should not recommend prenatal screening at all, and that the ethically appropriate scope of NIPT should be delineated more strictly than do Bayefsky and Berkman.

An alternative rationale for state intervention is rooted in the medical-ethical principle of nonmaleficence (Beauchamp and Childress 1979), and in a constitutional obligation for the state to protect and promote the health of the population (Ministry of the Interior and Kingdom Relations 2018). The Netherlands has a law, the Population Screening Act, which was introduced in 1992 to protect the population against harmful or unnecessary screening programs. In line with the existing international ethical framework for screening (Kater-Kuipers et al. 2018), the Dutch law recognizes that screening is inevitably associated with harms, costs and burdens, which must be outweighed by the benefits for screening participants. The risks of screening include those associated with false positive and false negative test results, ranging from the physical risks and financial costs of unnecessary follow-up diagnostics and medical intervention to psychological effects such as undue anxiety and vigilance. More fundamentally, screening—or the offering of medical research to populations to detect (risk factors for) diseases—tends to render healthy people sick. This means that to justify screening, adverse consequences must be offset by significant health benefits. Screening must be proportionate.

In most population screening programs, potential benefits are medical in nature. The results of screening must be "actionable": based on the results, it

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should be possible to start preventive or therapeutic interventions which lead to improved health outcomes. In first-trimester prenatal screening programs, however, the benefits should be understood in terms of strengthening reproductive autonomy (Kater-Kuipers et al. 2018). The information offered through prenatal screening should be associated with "action options" leading to better outcomes in terms of reproductive autonomy. From this requirement it follows that the information offered should at minimum be sufficiently clear to allow pregnant women to make reproductive decisions.

The Netherlands is the only country besides Belgium in which NIPT is offered as a first-tier screening test to all pregnant women, and is funded—in part—through the national healthcare system (Dutch women must make an out-of-pocket co-payment of 175 euro (approximately 195 US dollars)). There is no private market for NIPT, as based on the Population Screening Act, the offering of NIPT is prohibited without a license. The government has licensed a group of laboratories to screen for trisomies 13, 18 and 21, and to conduct research on "additional findings," including chromosomal aberrations other than trisomies 13, 18 and 21. While the clinical validity of NIPT results indicating increased risk of trisomies 13, 18 and 21 is high, the validity of results indicating other chromosomal aberrations was found to be lower (Van der Meij et al. 2019). In the Netherlands, therefore, it is not currently recommended to disclose findings beyond trisomies 13, 18 and 21, based on unclear or limited clinical validity (Health Council of the Netherlands 2020), unless women explicitly ask for it. This limit serves to protect pregnant women and their partners against insufficiently informative screening test results.

In their paper, Bayefsky and Berkman are not fully consistent when delineating their first category of information—information which should be recommended by healthcare professionals and reimbursed by health insurers. They write that NIPT should include "information that is useful for parental actions in the short-term" (Bayefsky and Berkman 2021, 24) or "any information necessary for decision-making or preparation in pregnancy" (30) or "information with direct bearing on the decision to terminate" (18). I believe that the latter delineation is most appropriate and most in line with the alternative rationale described above. Bayefsky and Berkman are at pains to acknowledge that the information offered through screening need not be used to terminate a pregnancy, but can also justifiably be used to prepare for the birth of an affected child. They suggest that "many, if not most, parents want to use the information to prepare" (Bayefsky and Berkman 2021, 21). I fully agree that pregnant women or couples should be free to choose to continue pregnancies after taking part in screening and after receiving positive results, and that they should receive all the care and support they need when doing so. This is part and parcel of respecting reproductive autonomy. But it does not mean that the primary purpose (or one of the primary purposes) of first-trimester prenatal screening should be for pregnant women or couples to receive (all kinds of) information that they may use to "prepare" for pregnancy and childbirth. Rather, the primary purpose of screening is to allow them to avoid having a child with a serious disability, if this is in line with their personal values, family interests, life plans, etc. Therefore, I would support delineating the ethically appropriate scope of NIPT such that it includes only information that is clinically valid and has direct bearing on decision-making about termination of pregnancy.

Bayefsky and Berkman argue that regulating the scope of NIPT should not be done by the state, but by associations of healthcare professionals. In the Netherlands, such a model for professional self-regulation has been implemented when NIPT was introduced in the Netherlands (Van der Meij et al. 2019). In this model, Dutch obstetricians or midwives will always offer prenatal screening in a neutral manner. It is felt that women should be free to choose to participate in screening or not to participate in screening, and that the exertion of pressure (however subtle) to take part in screening, should be avoided (Kater-Kuipers et al. 2020). National professional guidelines for pretest counseling thus require healthcare professionals to provide information about prenatal screening in a step-wise manner—to make an "information offer" first—to avoid directiveness: pregnant women and their partners are asked first whether they wish to be informed about opportunities for prenatal screening, if they do not, this preference is respected (National Institute for Public Health and the Environment (RIVM) 2018). Thus, screening is not *recommended* to pregnant women at all.

This step-wise approach to informed consent for prenatal screening results in a relatively low uptake of NIPT of approximately 46% (Van der Meij, Groot-van Mooren, et al. 2021). Dutch women who choose not to participate in first-trimester prenatal screening do so because they consider Down syndrome not severe enough to justify termination of pregnancy (Crombag et al. 2016), or because they feel that "every child is

welcome" (Van der Meij, Njio, et al. 2021). There are indications that in the Netherlands, there is little societal pressure for pregnant women to take part in screening (Kater-Kuipers et al. 2021; Van der Meij, Njio, et al. 2021).

The experiences with the implementation of NIPT in the Netherlands in recent years suggest that the aim of screening—reproductive autonomy—can be met in a healthcare system in which NIPT is regulated by the state, the scope of NIPT is strictly delineated, and healthcare professionals explicitly underline the freedom of pregnant women and their partners to choose or forego first-trimester prenatal screening.

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