Non-invasive prenatal diagnosis: an ethical imperative

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In their Ethics watch article (An offer you can't refuse? Ethical implications of non-invasive prenatal diagnosis. Nature Rev. Genet. 10, 515 (2009)), Schmitz et al. argue that the implementation of non-invasive prenatal diagnosis (NIPD) for fetal aneuploidies would pose a threat to the reproductive autonomy of women by impeding the provision of adequate pre-test counselling. I argue that the introduction of NIPD would in fact increase reproductive autonomy by allowing women to access information without subjecting their pregnancy to the risk posed by amniocentesis or chorionic villus sampling (CVS).

Women considering invasive testing are faced with a distressing dilemma: they must weigh the risk of bringing to term an affected fetus against the risk of losing a healthy one. To assist women and their partners in this decision and to inform them about the implications of possible abnormalities, pre-test counselling has become a routine component of prenatal diagnosis in most countries. Schmitz et al. argue that NIPD should replace current invasive testing only once “a new organizational setting” is in place to ensure that women are offered adequate pre-test counselling. However, delaying the introduction of NIPD into clinical practice would be ethically misguided, as explained below.

First, by eliminating the risk of pregnancy loss, NIPD would be of immense medical benefit and should therefore be available as soon as it is ready to be clinically implemented, even if the demand for it is initially so high as to prevent comprehensive pre-test counselling.

Second, because NIPD eliminates the risk of pregnancy loss, genetic counsellors would be able to focus on discussing the possible results of the test — and the alternatives open to women and their families — rather than spend a substantial amount of time and effort discussing the risk inherent in the test. Non-invasive prenatal diagnosis would therefore change the context of counselling in a way that would promote autonomous decision making.

Third, NIPD — like CVS — can be performed much earlier in the pregnancy than amniocentesis. However, CVS carries the same or an even higher risk of miscarriage compared with amniocentesis and is dependent on the availability of highly trained professionals. Non-invasive prenatal diagnosis would therefore enhance reproductive autonomy by allowing women more time to gather information. Furthermore, terminating a pregnancy during the first trimester rather than during the second trimester is medically safer, emotionally less traumatic (for some) and more easily available (some providers refuse to terminate beyond 12 weeks' gestation).

Fourth, diagnostic testing need not merely "enable the pregnant woman to decide whether to live with an impaired child or to terminate the pregnancy". Rather, the information may be sought to prepare for the birth of a child with special needs. Indeed, in the survey quoted by Schmitz et al. to demonstrate the inadequacy of current consent for prenatal testing, 23.2% of women indicated that prenatal diagnosis would enable "planning and provision of care for the baby in good time in the event of a disability". Access to NIPD would therefore enhance women's autonomy by allowing them to prepare for the outcome of their pregnancy, not just to make a decision about terminating it.

In conclusion, NIPD should be implemented as soon as it passes the appropriate threshold of accuracy and can safely replace current diagnostic testing. Although we should consider appropriate modifications to our current counselling and consent mechanisms in preparation for such implementation, these considerations do not justify withholding this valuable new technology.

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