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Supporting women's autonomy in prenatal testing

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Noninvasive fetal genetic sequencing done early in pregnancy is poised to become a routine part of prenatal care. While it could offer patients substantial benefits, there is a risk that it will be integrated into care "without the robust, evidence-based informed consent process necessary for respecting women's autonomy," states an article in the August 10 issue of the *New England Journal of Medicine*.

"If that happens, patients will be asked to decide whether to undergo invasive diagnostic testing and then to consider whether to terminate or continue their pregnancy without a full understanding of the results."

The lead author is Josephine Johnston, LLBB, MBHL, director of research at The Hastings Center; co-authors are Ruth M. Farrell, MD, an obstetrician-gynecologist at the Cleveland Clinic, and Erik Parens, PhD, senior research scholar at The Hastings Center. Johnston is the principal investigator and Parens is an investigator on a project on goals and practices for next-generation prenatal testing, supported by a grant from the National Human Genome Research Institute of the National Institutes of Health.

The authors anticipate the advent of noninvasive diagnostic tests that can sequence the entire genome of a fetus from whole fetal cells circulating in the mother's blood. They would yield vastly more information than the screening tests now in use—themselves fairly new—which analyze fragments of fetal DNA in the mother's blood.

The article calls for a broad range of changes in policy and practice to enable clinicians to give women the information they need to provide fully informed consent in prenatal testing.

While the meaning of prenatal genetic tests results has always been complex and often difficult to interpret, the complexity "grows exponentially when we move from tests that can generate a handful of results to information about hundreds or thousands of genes," the authors write. Many of these results will reveal genetic variations whose significance is unclear or unknown, leaving women unsure what to do with information they may wish they did not have.

The authors cite barriers to women giving fully informed consent to prenatal genetic testing. "Since the 1980s, prenatal screening tests for a small number of traits including Down's syndrome have become routinized in ways that can undermine informed consent," they write. "Specifically, studies show that women have undergone prenatal genetic screening and diagnostic tests with only a limited understanding of the indications and ramifications of the information that the tests return." Patients and providers have also engaged in the "collective fiction" that screening can improve a fetus's health and a "collective silence" that a positive result could lead a woman to decide to have an abortion.

"The need for fully informed consent in prenatal screening and testing has never been more urgent," the authors conclude. "Meeting this need will require adoption of reimbursement policies and professional practice guidelines that support clinicians in breaking with current routine practices, which too often involve dispensing with or failing to adequately carry out an informed consent process."

Other recommendations include: funding to develop education and counseling approaches to help patients decide whether to be tested and what to do with the results; social welfare and other policies that support

people with disabilities and their families "so that women's choices are less likely to be constrained by financial concerns or fear for the future welfare of a disabled child;" and access to abortion services. Johnston discusses their recommendations in the *New England Journal of Medicine*'s podcast .

More information: Josephine Johnston et al. Supporting Women's Autonomy in Prenatal Testing, *New England Journal of Medicine* (2017). DOI: 10.1056/NEJMp1703425

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