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Supporting Women's Autonomy in Prenatal Testing

Josephine Johnston, L.L.B., M.B.H.L.,

The Hastings Center

Ruth Farrell, M.D., M.A., and

Women's Health Institute, Departments of OB/GYN and Bioethics: Cleveland Clinic

Erik Parens, Ph.D.

The Hastings Center

Early and noninvasive fetal genetic sequencing is on the horizon.¹ Such expanded prenatal testing could offer patients substantial benefits. But current practices in prenatal screening and the complex nature of genomic science and technology create the risk that these tests will be integrated into care without the robust, evidence-based informed consent processes necessary for respecting women's autonomy. 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.

As genetic testing technology is advancing, access to abortion is eroding and the constitutional status of abortion rights is under threat. Given the political environment and the fact that prenatal testing can lead to pregnancy termination, some supporters of abortion rights may argue that now is an inopportune moment to raise concerns about prenatal testing. We disagree. Support for access to prenatal genetic tests and abortion services and advocacy for robust informed consent processes grow out of the same ethical commitment to respect for autonomy.

Since the inception of their field in the 1970s, genetic counselors have championed the ethical ideal of engaging with patients in a process of informed consent, which has included nondirective counseling on whether to undergo testing, the meaning of test results, and next steps. That ideal, especially in the prenatal context, can be traced in part to genetic counselors' awareness of the misdeeds of eugenicists, who sought to prevent pregnant women from having "unfit" children. Obstetricians have formally adopted the same commitment to informed consent and nondirective counseling.

Important barriers stand in the way of achieving this goal. 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. 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 can

Corresponding Author: johnstonj@thehastingscenter.org.

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return.² Medical anthropologists studying various prenatal screening methods have found that routinization may also include the “collective fiction” that screening can improve fetuses’ health and a “collective silence” regarding the fact that a positive screening result could eventually lead to a decision to abort.

Many factors have contributed to this routinization. Because there aren’t nearly enough health care providers with expertise in prenatal genetics (including genetic counselors and maternal–fetal medicine specialists) to meet with all pregnant women, pretest and posttest counseling often falls to primary prenatal care providers. The evolving constraints on clinical practice allow very limited time during visits for conversations with patients about prenatal testing, disability, and pregnancy termination. Furthermore, research has shown that these clinicians can be quite directive when counseling patients about prenatal testing, perhaps owing to their perceptions of the value of such information and the emphasis of their training. Furthermore, the evolving constraints on clinical practice allow very limited time during visits for conversations with patients about prenatal testing, disability, and pregnancy termination. Finally, the threat of litigation has fueled the kinds of routinized practices that propel patients toward testing.

Offering patients a robust informed consent process will become more demanding if, as seems imminent, prenatal sequencing becomes widely available. Although some commentators are optimistic that these hurdles can be overcome, use of sequencing in a variety of research and clinical contexts is showing that the meaning of results is complex and rapidly evolving.

Of course, the meaning of prenatal genetic test results has always been complex. Very little phenotypic information is available about a fetus, and the variable expressivity of disorders and the incomplete penetrance of most genetic variants have long posed interpretive challenges. But the complexity grows exponentially when we move from tests that can generate a handful of results to those that can reveal information about hundreds or thousands of genes.

In addition, policies and practices for sequencing the genome of the unborn fetus will need to address management of results revealing what are called “variants of uncertain significance” and what we might call “variants of misattributed significance.” Many gene variants whose significance is unclear or unknown are later reclassified as benign, and there are also genomic variants whose meaning is thought to be understood, until that understanding turns out to be wrong. Clinicians at the Mayo Clinic, for instance, recently reported that a cardiac defibrillator was implanted in an adolescent boy who carried a variant that a genetic testing company had reported as a “probable deleterious mutation” but that the Mayo team subsequently discovered had not caused heart problems in the child.³ Such a scenario highlights the importance of addressing the possibility of variants of uncertain or misattributed significance. It is not difficult to imagine a woman terminating a pregnancy because she is told that the fetus carries a pathogenic variant and then have that variant turn out to be benign or of minor impact or to lead to a condition that is readily treatable.

One might argue that detailed informed consent processes preceding receipt of genetic information are unnecessary because people want as much genetic information as they can get. Yet two recent studies suggest that people's desire for genomic information in the prenatal context varies.

In a study of pregnant women receiving abnormal prenatal chromosome microarray results, Barbara Bernhardt and colleagues found that although many women initially saw the offer of more information about their fetus as "too good to pass up," they were blindsided by the results.⁴ Many of the women who received uninterpretable or uncertain results viewed them as "toxic knowledge" that they wished they did not have. The researchers therefore called for "a thorough discussion of the various uncertainties associated with abnormal [testing] results" as part of pre-test counseling.

When Miriam Kuppermann and colleagues tested a computerized, interactive decision-support guide with pregnant women, they found that those who used it were both better informed about the benefits and risks of various prenatal tests and less likely to opt for diagnostic testing than patients who were randomly assigned to receive standard care.⁵ The study authors concluded both that women can be helped to better understand their options and that clinicians need to underscore that forgoing testing is "a reasonable choice."

It is daunting to envision informed consent processes robust enough to prepare patients to receive information as voluminous and complex as sequencing results. Yet these studies clearly indicate that women have a strong preference for receiving help in parsing the distinction between empowering and disempowering information. As part of that process, clinicians have an ethical responsibility to discuss the small but real possibility that information from testing will lead women to face pivotal decisions about ending or continuing their pregnancy.

The need for fully informed consent in prenatal screening and testing has never been more urgent. 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. It will also require funding for development of approaches to pretest and post-test education and counseling that empower patients to decide whether to be tested and what to do after receiving their results. Policies in a range of areas, from education to social welfare to health care, that support people with disabilities and their families are also needed 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 respect for women's autonomy also crucially requires access to abortion services. Only with these practices and policies in place can women's decisions about prenatal screening, diagnostic testing, and termination or continuation of pregnancy be truly free and informed.

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