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THE CURE

Easy test. Tough answers.

What if parents could access the genetic makeup of a fetus in the first trimester of a pregnancy? And what if they could gather this information through a procedure that was largely risk free and almost completely noninvasive?

Well, those what-ifs have arrived. The era of noninvasive prenatal testing (NIPT) is with us, bringing a host of profound clinical, ethical and legal challenges that we've barely begun to explore.

The technology that got us here is, on the surface, straightforward. Draw blood from the mother. Find and isolate the relevant free-floating fetal DNA. Analyze.

Several companies already offer NIPT for common aneuploidy conditions, like Down syndrome, though the test is not yet considered reliable enough to be truly diagnostic (positive results must be confirmed by another recognized method). In Canada, the test is not currently offered in the public system (clinical studies are ongoing), but its use may be discussed with some women as a possible private option.

These are early days for NIPT. There is no doubt that in the relatively near future, parents will be able to use NIPT technology to reveal information about a host of genetic predispositions, such as whether a fetus carries a mutation that increases the risk of heart disease or various kinds of adult onset cancers. Indeed, the sophistication of genetic sequencing technology has advanced at such an incredible pace that we face the possibility of using NIPT to sequence a fetus' whole genome — an achievement already accomplished in the research setting.

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In other words, parents may soon be able access the entire genetic profile of a fetus. And the technology is evolving so quickly that it has been suggested that the global market for NIPT is already worth over $1 billion.

While previous forms of prenatal diagnostic technologies have often elicited intense policy debates — particularly in the context of nonmedical uses such as sex selection — the availability of whole-genome NIPT seems likely to take these questions to an even higher level.

To be clear, from a clinical perspective, the availability of reliable NIPT will be a tremendous advance. It promises more definitive diagnostic capabilities than existing techniques and is associated with fewer risks, particularly to the fetus, compared with amniocentesis. The test is also far easier to conduct.

But it is precisely this simplicity and diminished risk that causes concern. Deciding to have a blood test seems a less dramatic decision for an expectant mother than opting for amniocentesis, which requires a careful weighing of risk and benefit. Some observers worry that the informed consent procedures will not be as comprehensive, or that parents and practitioners will view the test as a part of the prenatal routine and will therefore fail to carefully consider the ramifications of NIPT.

There is also a fear — unsupported by evidence at this point — that the ease of NIPT will increase pressure to act upon the results of the test if it sets off genetic warning signals.

This anxiety extends to the possible use of NIPT to identify susceptibility genes for late-onset diseases and conditions like diabetes or obesity, or for nonmedical purposes such as selecting particular genetically determined traits, gender being the most obvious.

Agreeing on the boundaries for application of this technology — and how best to deploy it in our public health care system — will be challenging. Let's say that, as a society, we come to some degree of agreement about the kind of genetic information that should be available to parents through NIPT. Let's say, hypothetically, that we think parents should have access only to a particular set of highly predictive disease-causing genetic mutations.

As a strong supporter of individual and reproductive autonomy, I am not necessarily agreeing with this policy approach. But how would we, as a society, design and implement a policy that accommodates the ethical and legal strains raised by this new technology? What kind of laws would be needed? How would they be policed?

Canadians currently have a right to access their health information. This right is codified in provincial health information laws and has been affirmed by the Supreme Court of Canada. If a woman gets an ultrasound during her pregnancy, she can ask for, and has a right to receive, all the information disclosed by that procedure including available information suggesting the sex of the fetus. A clinician cannot, out of concern for how the woman will use the information, decline the request.

Given this legal framework, a woman who gets NIPT would have the right to access to all the information disclosed by the procedure. Health care providers do not have the option of picking and choosing what to reveal to the mother. Moreover, physicians can't interrogate women about how they intend to use NIPT information prior to offering the test.

We could, of course, choose to enact a new law or amend all the existing provincial health information legislation to create specific rules for NIPT. (Since health is a provincial jurisdiction, this would require action by every province, further complicating the policy-making process and likely leading to a checkerboard of laws across the country.) Other countries, particularly in Asia, have passed laws restricting access to prenatal information, largely as a result of the high level of concern about sex selection.

But for Canada, this would be a drastic departure from the existing norms on consent and personal autonomy. Nor is it easy to imagine provincial governments welcoming the inevitable public furor that would accompany a legislative debate associated with reproductive autonomy, the moral status of the fetus and the termination of pregnancies.

I have always been cautious about predicting the near future implementation of "revolutionizing" health care technologies. They never seem to happen in the predicted "near future" or "revolutionizing" manner.

But this revolution seems inevitable. Many technical hurdles remain, but NIPT seems to be evolving at a truly remarkable pace. And there is virtually no chance that Canada's legal norms will change before a range of sophisticated NIPT technologies are available.

Given this reality, our first and most logical policy response should be education and a comprehensive pretest consent and counselling process for mothers and fathers. While there are single-gene disorders and conditions for which NIPT will provide relatively definitive results, the vast majority of information that would be provided by a whole-genome scan is far from being highly predictive of an unborn child's future health.

Our health is determined by an inconceivably complex mix of countless biological and social factors, from lifestyle choices to geography, our educational opportunities and peers, ever-improving health care and public health developments and, to a degree far less than often portrayed in the popular press, genetics. That truth needs to be told, and reinforced.

The emergence of NIPT will require some difficult policy choices, but will also be an opportunity to improve public awareness. Its emergence also offers us a moment to examine again what it means to be human in a time when technology asks that question in new and difficult ways. This is a chance to remind ourselves that genes do not, necessarily, hold our destiny, no matter how thoroughly or early we map them. ■