

Event

Non-invasive prenatal genetic testing (NIPT) is a blood test for pregnant women that screens for certain fetal genetic disorders, such as Down syndrome. The test works by analyzing fetal DNA fragments circulating in maternal blood. There has been a lot of excitement surrounding this technology due to its non-invasive nature, high level of accuracy, ability to be employed at a relatively early point during pregnancy, and potential to reduce the number of fetal losses associated with more invasive procedures. It raises, however, important questions of disclosure and consent.

Significance

There is some uncertainty about how NIPT should fit within the current, provincially-funded, framework of prenatal screening and diagnosis. NIPT is currently less accurate and able to detect a more limited range of disorders than invasive diagnostic tests, such as amniocentesis or chorionic villus sampling (CVS). As a result, invasive testing continues to be recommended to confirm positive NIPT results¹. Additionally, in most Canadian provinces and territories, NIPT is not provincially funded and only available to high risk pregnant patients who are able to pay for the test themselves. These issues raise questions about the level of disclosure and standard of care required of physicians treating pregnant patients².

Analysis

Although amniocentesis is still considered the “gold standard” in prenatal genetic diagnosis³, physicians have an obligation to disclose the risks and benefits of a procedure, as well as potential less-invasive alternatives⁴. If a fetus is lost as a result of an invasive procedure, whether NIPT was discussed as a potential screening tool may be relevant in assessing potential liability. The legal test for disclosure is whether a reasonable person in the patient’s circumstances would have consented to the procedure had all of the relevant information been disclosed⁵. Courts may find that a reasonable person in a patient’s position would want to know of all possible screening and diagnostic options, particularly if she has expressed concerns over the specific disorders detectable by NIPT or the possibility of fetal loss associated with invasive testing. This will be increasingly important if NIPT reaches diagnostic capabilities. As the standard of care can change over time, physicians must “keep pace with advances in medical science”⁶. When discussing NIPT with patients, it is important that physicians disclose the limitations of NIPT in terms of accuracy and range of disorders that can be detected to prevent patients from having false assurances as to the health of their fetuses. A failure to adequately disclose this information could result in liability if a patient declines invasive diagnostic testing believing NIPT to be comprehensive and diagnostic, and a baby is born with a genetic disorder that would have been detected by amniocentesis or CVS. Discussions about NIPT with patients will also become increasingly important as this technology gains acceptance for use in low-risk patients who may be carrying affected fetuses with disorders that would go undetected under the conventional, less accurate screening process.

Conclusion

It is important that the implications and limitations of NIPT be thoroughly discussed with patients as well as the availability of genetic counselling services. As the technology behind NIPT is changing rapidly, it is important that physicians keep up to date on technological developments and professional recommendations. The role of professional societies in this regard will be important.

1. Langlois S, Brock J-A, Genetics Committee, et al. Current status in non-invasive prenatal detection of down syndrome, trisomy 18, and trisomy 13 using cell-free DNA in maternal plasma. *J Obstet Gynaecol Can.* 2013;35(2):177-181.
2. **Toews M, Caulfield T. Physician Liability and Non-Invasive Pre-Natal Testing. *J Obstet Gynaecol Can* 2014;36(10):907–914.**
3. Benn P, Cuckle H, Pergament E. Non-invasive prenatal testing for aneuploidy: current status and future prospects: NIPT for aneuploidy. *Ultrasound in Obstetrics & Gynecology.* 2013;42(1):15-33. doi:10.1002/uog.12513.
4. *Cory v. Bass*, 2012 ABCA 136, 522 A.R. 220.
5. *Reibl v. Hughes*, [1980] 2 SCR 880 (SCC).
6. E. I. Picard & G. B. Robertson, *Legal Liability of Doctors and Hospitals in Canada*, 3rd ed., (1996), at pp. 276-77.