Comment on authors' reply: patients' perception of risk: informed choice in prenatal testing for foetal aneuploidy

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We are heartened to read that the authors found our commentary insightful.[1] There is currently a myriad of prenatal tests available to patients, and in order for clinicians to help patients make a truly informed decision about their test options, it is integral that they have thorough knowledge of the details of the tests and how they compare with each other.

We strongly feel that the authors' view that "*additional benefits such as identifying cardiac defects in the foetus may be a bonus*"[1] needs to be addressed. It is widely known that congenital heart defects are the most common type of congenital malformation, with an incidence of 1 in 100 to 1 in 150.[2] Compared to the 1 in 700 birth incidence of Down syndrome (DS) and the 1 in 7,000 to 1 in 10,000 birth incidence of Trisomy 18 (T18),[3-5] congenital heart defects are 7–100 times more prevalent and hence a greater cause of concern for the general population. Given such high frequencies, efforts taken to identify heart defects during pregnancy is not simply 'a bonus'. Early screening for possible cardiac defects will only be for the benefit of patients, as it will prompt clinicians to be more vigilant in their care – either by recommending a foetal echocardiogram, or at least by paying careful attention to the foetal heart during the routine 20 weeks ultrasound scan.

Although the authors explained that the purpose of their paper was "to offer informed choices in the prenatal testing for foetal aneuploidy",[1] we find that it is crucial that we address the definition of informed choice. We agree with the authors that decisions about testing should be made autonomously by the patient, but readers should be reminded that patients are only able to make an informed choice if they have sufficient information in hand about their test options, including details of the conditions covered by the test, and understand the risks, benefits and limitations of the tests they are considering. The role of genetic counselling in noninvasive prenatal testing (NIPT) has been addressed by many concerned parties.[6-8] The National Society of Genetic Counselors (NSGC), for example, recommends that "*pretest counseling for NIPT include information about the disorders that it may detect, its limitations in detecting these conditions, and its unproven role in detecting other conditions*".[9] It has been clearly understood that although the detection for DS and T18 using NIPT is higher than that of first trimester screening (FTS), NIPT will not detect any other abnormalities that would have been suggested in FTS. In fact, the American College of Obstetrics and Gynecology guidelines emphasise that "*counseling regarding the limitations of cell free fetal DNA testing should include a discussion that the screening test provides information regarding only trisomy 21 and trisomy 18 and, in some laboratories, trisomy 13*".[10] Like the FTS, NIPT is a screening tool for DS, T18 and perhaps even T13. Unfortunately, unlike FTS, NIPT is unable to reveal any insight into other concerns about the pregnancy, such as genetic diseases, structural defects like cardiac defects, and adverse pregnancy outcomes.

Simply focusing on the detection rates of NIPT on trisomies alone does not constitute 'informed choice'. We feel that it is imperative for readers to be reminded of the limitations of NIPT and the importance of providing appropriate counsel to patients regarding the full details of their test options. We also need to be careful about how we integrate NIPT into a patient's obstetric care. According to the old adage, one should not lose sight of the forest for the trees. Similarly, we are to remind ourselves not to get caught up in the niftiness of NIPT and lose sight of the valuable information that various other screening tests have to offer.

Yours sincerely,

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