Comment on: Patients' perception of risk: informed choice in prenatal testing for foetal aneuploidy

We read with great interest the article entitled "Patients' perception of risk: informed choice in prenatal testing for foetal aneuploidy".[1] As the authors reminded us that it "*behoves the clinician to be aware of newer tests for increasing accuracy with no risk of miscarriage*" and attempted to summarise various prenatal screening and diagnostic tests, it is imperative that readers of the article be made aware of the fact that contemporary practice at hospitals in Singapore has long surpassed what is mentioned in the article.

In the last decade, prenatal screening for Down syndrome has transitioned from the second trimester triple test to the first trimester screen (FTS). In Singapore, to our knowledge, obstetricians follow the standards of the Fetal Medicine Foundation (FMF). In contrast to the gestational age of 9–14 weeks quoted by Choolani and Biswas, the gestational age for measuring the nuchal translucency (NT), as recommended by the FMF, is 11–13 weeks and six days.[2] When combined with maternal age, NT on its own allows a 75%–80% detection rate at a 5% false positive rate for Down syndrome.[3,4] In addition, the authors failed to discuss the value of NT measurement beyond simply screening for Down syndrome. A thickened NT also sheds light on the possibility of an increased risk for foetal death, perinatal loss, chromosome disorders, including trisomy 13, trisomy 18, Turner syndrome and genetic diseases, including some cases of alpha thalassaemia. Many genetic syndromes have subtle features that can be difficult to detect by cursory routine ultrasonography. An increased NT is also associated with many structural malformations such as cardiac defects in the foetus. If the anomaly scan at 20 weeks is normal, it is believed that the chance of the foetus having an adverse outcome is most likely low.[5-7] The usefulness of the NT measurement as an early screen for pregnancy complications and foetal abnormalities should not be overlooked. We agree with the authors that the common practice of combining the NT measurement with the analysis of maternal serum free β-hCG and PAPP-A further improves the detection rate of Down syndrome to 85%–90% with a 5% false positive rate.[3,4,8,9] However, multiple studies have also demonstrated that the detection rate of the FTS may even be further improved to 93% by analysing serum free β-hCG and PAPP-A at 9–10 weeks gestational age.[10-12] It is therefore important to clarify that the combined screen is not only capable of yielding a more than 90% detection rate for Down syndrome, but is also more than just a simple screening tool for Down syndrome alone.

The FTS has been routinely offered to patients at KK Women's and Children's Hospital for the past eight years. In fact, in the last eight years, our hospital has also incorporated other specific markers of Down syndrome, including the absence of the nasal bone, and more recently, reversed a-wave in the ductus venosus and tricuspid regurgitation. These markers can further improve the detection rate of Down syndrome to 93%–96% and also lower the false positive rate to 2.5%.[13-15] The FTS has moved beyond the simple combined screen and beyond simply a screening test for Down syndrome. In fact, the specific details of the timing of the FTS and the useful information gleaned from the FTS to help guide antenatal care beyond Down syndrome screening has been recognised by the RCOG and NHS for about a decade.[16-18] It therefore behooves authors of academic papers to be aware of not only "*newer tests for increasing accuracy with no risk for miscarriage*", but also what constitutes standard clinical practice.

Yours sincerely,

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