Masterclasses

M12.2 Diagnosis Before Birth: For Mothers and for Babies 10:45 Theatre 2

Non-invasive Prenatal Testing as Primary Screening for Down Syndrome

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The performance of non-invasive prenatal testing (NIPT) is superior to the Down screening methods currently in use for both high- and low-risk pregnancies. In terms of benefits and harms, NIPT as first-tier screening test is preferred. The concern over loss of benefits from current Down screening strategy after its replacement by NIPT is not substantiated. The ethical principles of equity and reproductive autonomy also favour NIPT for universal screening. Evidence from the US demonstrated that, from a social perspective, it's cost effective to replace current Down screening strategies with NIPT if the cost of NIPT is no higher than USD453.[1] A preliminary analysis showed that when the cost of NIPT is below USD300, current Down screening strategies in the Hospital Authority could potentially be replaced by NIPT without increasing the expense per case of trisomy 21 diagnosed from a social perspective. As the price of NIPT is now down to USD300 and below, universal application of NIPT can be economically justified. In fact, it was recognised that NIPT could be offered below USD250 and yet the provider is already making a good profit from it.[2] The use of NIPT as a primary screening test for all pregnant women is also endorsed by the International Society of Prenatal Diagnosis (ISPD).[3]

In Hong Kong, the universal first trimester combined screening (FTS) using fetal ultrasonographic measurement of nuchal translucency and serum biochemical markers to detect common aneuploidies has implemented since 2010. However, since 2011 when non-invasive prenatal testing (NIPT) for aneuploidy using cell-free DNA (cfDNA) in maternal plasma came into clinical use, this has resulted in tremendous changes in our prenatal counselling and testing. Although NIPT has a higher detection rate and lower false positive rate in detecting Down's syndrome, the implementation as primary screening to replace the current system will lead to missing more other fetal chromosomal abnormalities and it is not cost-effective. The apparent benefit of reduction of miscarriage from avoiding invasive prenatal diagnostic procedures may have been overestimated as well. Instead of implementing as primary screening, incorporating NIPT into current universal screening strategy as the contingent screening will gain the benefit of improving the detection rate without missing other fetal chromosomal abnormalities, and is relatively cost-effective.

References:

1. *Fairbrother G, Burigo J, Sharon T, Song K. Prenatal screening for fetal aneuploidies with cell-free DNA in the general pregnancy population: a cost-effectiveness analysis. J Matern Fetal Nenatal Med 2015; 29(7): 1160-4.*
2. *Minear MA, Lewis C, Pradhan S, Chandrasekharan S. Global perspectives on clinical adoption of NIPT. Prenat Diagn 2015; 35: 959-67.*
3. *Benn P, Borrell A, Chiu RWK, Cuckle H, Dugoff L, Faas B, et al. Position statement from the chromosome abnormality screening committee on behalf of the board of the International Society for Prenatal Diagnosis. Prenat Diagn 2015; 35: 725-34.*