Non-Invasive Prenatal Testing: an audit of its indications and uptake in a tertiary hospital



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Introduction

- Non-invasive prenatal testing (NIPT) is a revolutionary screening method for pregnancies considered at high risk of aneuploidy.
- Given its superior rate of detection over combined first trimester screening (cFTS), studies on its uptake in large healthcare facilities are warranted to understand the impact of its introduction on existing screening methods.

Objective

To examine the uptake of NIPT in a tertiary hospital including: the number of NIPT performed, indications for its referral, the results and the number of invasive procedures.

Methods

- An audit was conducted of all cases of NIPT referred through the Maternal Fetal Medicine Unit of RBWH between 7th October 2015 and 7th April 2018. Patients' details and test results were obtained through private pathology centres. Further information were obtained from the Queensland Integrated Electronic Medical Record system.
- Data extraction and analysis was completed with respect to patient characteristics, obstetric and antenatal history, indications for NIPT, results of testing, and invasive procedures if performed.

Table 1. Reasons for NIPT referral



Results

- NIPT screening was performed in 90 women (91 tests) with a mean age of 33.4 years (19-45) and mean BMI of 24.3 (18.3-50.5).
- The commonest reason for referral was a high risk cFTS, followed by abnormal findings on fetal morphology scan, and previously confirmed trisomies.
- NIPT returned high risk in only one case for trisomy 21 with no false positive or false negative results.
- Amniocentesis was performed in six cases (6.59%). Indications include previously confirmed aneuploidy, and to exclude genetic conditions in carrier mothers.

Conclusion

The uptake of NIPT is popular amongst high risk pregnancies and has increased secondary to reducing costs of testing. Our audit on uptake of NIPT provides insight into the various indications for the test and reflects patients' screening preferences.