OBSTETRICS AND GYNAECOLOGY
CLINICAL PRACTICE GUIDELINE

NIPT Prenatal Testing for Fetal Aneuploidy

Scope (Staff):	Obstetrics and Midwifery
Scope (Area):	Obstetrics and Gynaecology

This document should be read in conjunction with the Disclaimer.

Aim

To describe NIPT and its place in antenatal aneuploidy screening to assist medical staff and their patients make an informed decision about its use.

Background

Non-invasive Prenatal Testing (NIPT) uses cell-free DNA of placental origin in the maternal serum to screen for fetal aneuploidy. Multiple independent studies have demonstrated the clinical validity of maternal plasma cell-free DNA sequencing for the detection of fetal trisomy 21 in high risk women. The NEXT trial, published in 2015, has subsequently demonstrated the same high performance characteristics in low-risk women. Cell-free DNA based NIPT has high sensitivity (>99.5%) and specificity(99.8%) as a screening test for trisomy 21 with an overall positive predictive value of 80% (compared with 3.4% for combined First Trimester Screening).

Key Points

1. NIPT is not a diagnostic test and high risk results still require confirmation with an invasive test e.g. amniocentesis or CVS.
2. A routine 12 - 14 week gestation ultrasound scan is still required if NIPT is undertaken to assess for multiple pregnancy and major fetal abnormalities (e.g. anencephaly).
3. Currently there is no Medicare funding or private health insurance rebate for NIPT.aneuploidy screening and NIPT has significant direct costs to the patient ($395-695, depending on the specific provider used) and has a turn-around time of approximately 7-14 days.
4. The results report a risk assessment for trisomy 21, 18 and 13.
5. It can also report on the sex of the fetus and some sex chromosome abnormalities although the performance characteristics are lower than for autosomes
6. Maternal serum can be collected from 10 weeks gestation.

7. There is a test failure rate of up to 5% (this is higher as body mass index increases: the test failure rate is likely to be 50% at a maternal weight of 160kg). In women with test failure there is an increased risk of aneuploidy (6-fold increase in a low risk population).
8. NIPT can be used in twin pregnancies but will only test for trisomies 21,18 and 13. The performance characteristics are lower in twin pregnancies than in singleton pregnancies.
9. NIPT has not been validated for triplet or higher order multiple pregnancies.
10. The laboratory must be advised of the presence of a non-viable fetus together with a viable twin.
11. NIPT is not the appropriate test for aneuploidy if there are fetal abnormalities present on ultrasound. In this circumstance a CVS or amniocentesis should be performed

Pre Screening Counselling

Counselling prior to testing should include

- The sensitivity rate.
- The positive predictive value- this is influenced by test performance and population prevalence and may be as low as 50% for some aneuploid conditions.
- The reported “99%” probability for high risk results is a description of the test and is not equivalent to the positive predictive value which is dependent upon both test and patient factors
- The screening only covers trisomies 21,18 and 13 and X and Y chromosomes (although some laboratories are now offering testing for other aneuplodies such as trisomy 9,16 and 22 and microdeletion / duplication syndromes such as DiGeorge Syndrome ,Prader-Willi / Angelman Sydrome).
- The accuracy of sex determination is 99%
- Any abnormal results will require invasive testing i.e. amniocentesis or CVS
- The approximate cost to the patient.

Keywords:	NIPT, fetal, Aneuploidy, trisomies, ultrasound, maternal, trisomy, abnormality		
Document owner:	Co-Directors, Obstetrics and Gynaecology		
Author / Reviewer:	Clinical Guidelines Co-ordinator		
Date first issued:	June 2016		
Last reviewed:	August 2024	Next review date:	February 2025
Endorsed by:	Midwifery and Obstetrics Clinical Practice and Outcomes Committee	Date:	August 2024

NSQHS Standards Applicable:	Std 1: Clinical Governance

Printed or personally saved electronic copies of this document are considered uncontrolled. Access the current version from the WNHS website.

Version History

Version Number	Date	Summary
1.0	June 2016	First version
2.0	August 2024	Clinical decision by Executive to extend review date by 12 months to August 2025

The health impact upon Aboriginal people has been considered, and where relevant incorporated and appropriately addressed in the development of this policy.

This document can be made available in alternative formats on request for a person with a disability.