Hospital funded Non-Invasive Prenatal Testing for women at increased risk of aneuploidy based on first or second trimester screening: a retrospective cohort study.

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Transformation: Making Waves

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Introduction

Screening for aneuploidy in pregnancy with Non-Invasive Prenatal Testing (NIPT) incurs an out-of-pocket cost of $449[1], and is not covered by Medicare, nor has it been routinely funded by hospitals as a second-tier test. This is cost-prohibitive for many women, including those who could benefit from NIPT as a second-tier screening test, where the alternative is invasive diagnostic testing with chorionic villus sampling (CVS) or amniocentesis.

Contingent screening for those at intermediate risk has a high overall detection rate (97.8%) for major trisomies and may be a cost effective and feasible first trimester screening test for aneuploidies in the public health system[2].

The Royal Hobart Hospital (RHH) introduced hospital funded NIPT (HF-NIPT) as a possible alternative to diagnostic testing for women at increased risk of aneuploidy who met strict clinical criteria.

Objective

To assess whether HF-NIPT for women with an increased risk of fetal aneuploidy reduces invasive procedure rates at our tertiary centre.

Methodology

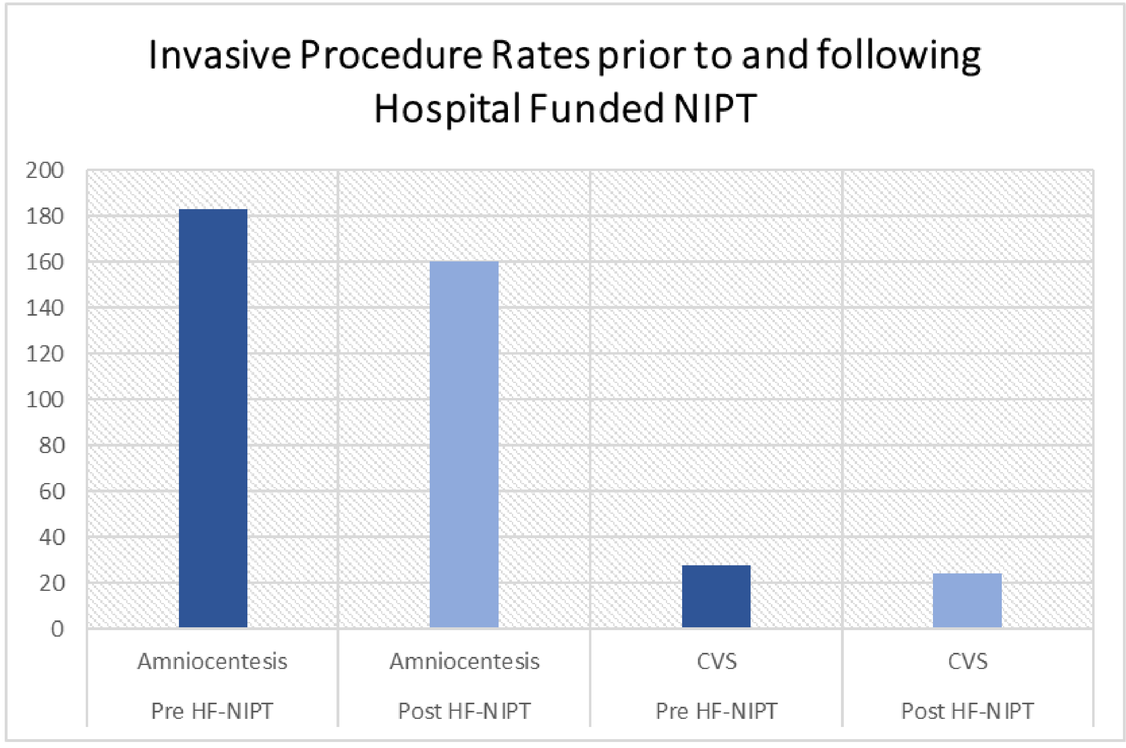
From the 1st of June 2017, women referred with a high risk first or second trimester screen, who met strict clinical criteria (Table 1), were offered HF-NIPT (*percept*®) or diagnostic testing (CVS or amniocentesis).

Table 1: Criteria for HF-NIPT at RHH
Risk for trisomy on combined first trimester screen between 1:50 to 1:300.
In the following indications, after detailed counselling: • Risk for trisomy greater than 1:50, • Fetal anatomical abnormality or severe early growth restriction, • NT > 3.4mm, or • PAPP-A or BHCG < 0.2MoM. *invasive testing should be offered as first line as the risk of atypical chromosomal abnormalities is greater.
In those with a risk of certain balanced translocations *following detailed genetic counselling and discussion with the cytogenetics laboratory.

Data was extracted from the cytogenetics laboratory software to capture all NIPT, CVS and amniocentesis performed from 1st November 2012 to 31st January 2022, in order to compare the rates of invasive procedures prior to and after the implementation of HF-NIPT.

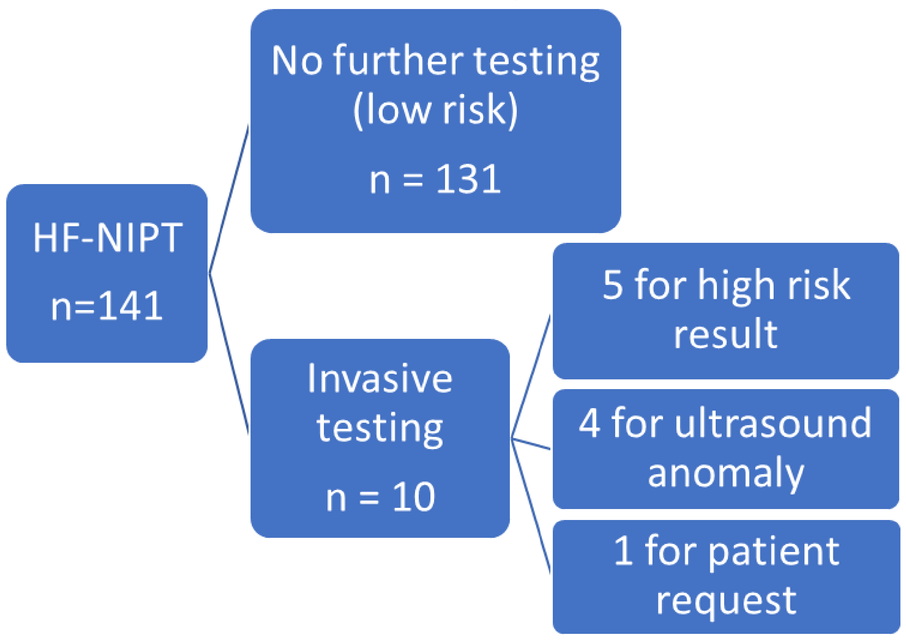
Results

183 amniocentesis and 28 CVS were performed prior to the introduction of HF-NIPT, compared with 160 amniocentesis and 24 CVS after. This equates to a 12.5% reduction in amniocentesis and a 14% reduction in CVS over 4.6 years.



141 HF-NIPTs were performed, with the main indication being high risk first and second trimester screen.

10 went on to have diagnostic testing (five due to high risk NIPT, four due to ultrasound anomalies, and one due to patient request). One HF-NIPT was a false negative, where microarray from amniotic fluid revealed triploidy (69, XXX). There were no false positive HF-NIPTs.



Conclusion

HF-NIPT as a second-tier screening test led to a significant reduction in the rate of invasive diagnostic testing at our tertiary centre.

Widespread uptake of this model of contingent screening should be evaluated, assessing the false negative rate, cost effectiveness and patient satisfaction.

References

1. Victorian Clinical Genetics Service, Percept™ NIPT, viewed 28th August 2022, <https://www.vcgs.org.au/tests/perceptnipt>
2. Colosi E, D'Ambrosio V, Periti E, *First trimester contingent screening for trisomies 21,18,13: is this model cost efficient and feasible in public health system?,* J Matern Fetal Neonatal Med. 2017 Jan 4:1-6.