SSI03491

Aneuploidy Screening and NIPT (Non Invasive Prenatal Testing) in Obstetrics

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NIPT using cfDNA:
The presence of cell free foetal DNA in maternal serum was first described in 1997. This "foetal" DNA originates from the placenta and is rapidly cleared after delivery.

NIPT using cfDNA screening of maternal plasma can be performed from 10 weeks gestation and has greater sensitivity and sensitivity than combined serum/US screening described above, however there is no Medicare or private health insurance funding and therefore is completely funded by patients. Currently this is around $400.

Some women will opt to fund cfDNA as a primary screening tool, it is then advised that they do not proceed with SAMSAS screening as this may increase the false positive rate without increasing sensitivity. Women who opt for cfDNA as a primary screening tool should still be offered first trimester NT US to screen for structural anomalies, and if their US demonstrates anomalies may be offered invasive testing even with a low risk cfDNA result.

Three cfDNA tests are currently commercially available, are processed in Australia and have a turnaround time of approximately 1 week. SA Pathology is in the process of developing a NIPT, this is not currently available.

Harmony (Ariosa) through Clinpath utilises single nucleotide polymorphism technology. https://www.sonicgenetics.com.au/nipt/

Obese patients and those conceiving with IVF are more likely to have a test failure; however women with an aneuploid fetus are also more likely to have a test failure. All women with a test failure should be advised this places them at high risk for aneuploidy and they should be advised to have an invasive test.

All women who have either an amniocentesis or NIPT should have a registrar or consultant appointment two weeks later to discuss the result. This appointment could be booked at Modbury hospital unless the patient has transport/geographical issues with attending Modbury.

Based on the above flowchart:
SAMSAS Risk <1:500 reassure, routine ANC.

Anyone with a SAMSAS risk >1:500, structural anomaly on US or NT >3mm should be referred for registrar/consultant counselling in ANC.

SAMSAS Risk 1:250 1:500 advise of availability of cfDNA and cost associated. Patients that decline cfDNA but are concerned <</MCIa6( )-hat

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Consent

All Procedures involving the provision of medical treatment to a patient must be undertaken in accordance with the Consent to Medical Treatment and Palliative Care Act 1995.

Work Health Safety

The responsible manager must ensure all workers who undertake this Procedure receive adequate information, training, supervision and support. Staff following this Procedure have a duty of care for taking reasonable steps to protect their own health and safety and not adversely affecting another person while at work. Further information is available from the NALHN Work Health and Safety Services intranet.

Standards

National Safety and Quality Health Service Standards

Standard 1 Governance for Safety and Quality in Health Service Organisations	Standard 2 Partnering with Consumers	Standard 3 Preventing & Controlling

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