Northern Health and Social Care Trust

Freedom of Information Request Reference

Antenatal Screening for Chromosomal Conditions

1. **Does the Trust offer tests to detect chromosomal disorders, including Trisomy 21 and if so what are the type of tests?**
2. **If so, are these tests offered to all woman or only offered to woman over the age of 35?**
3. **If applicable when are woman offered these tests?**
4. **If a woman has screening tests and they return a high chance of a chromosomal disorder what information is provided to the woman?**

The Northern Health and Social Care Trust does offer antenatal screening tests for chromosomal conditions to all women who request this and also to women thought to be at higher risk of having a baby with a chromosomal condition for example women who are over 35years of age or have a family history of chromosomal conditions.

There are two types of blood tests available and are funded privately by the woman requesting the test-

- **The Quadruple Test** (£27) is a blood test available between 15weeks and 20 weeks (ideally between 15 - 17 weeks) of pregnancy and screens for Down syndrome (T21), measuring four substances in the mother's blood and together with taking into account the mother's age, weight, gestational age – measured from a dating scan calculates the risk of the woman having a baby with Down syndrome. This test detects about 80% of babies affected by Down syndrome, meaning that 1 in 5 cases are not detected. The test also can generate a false positive result in about 3% of cases, meaning that the result indicates that the baby has Down syndrome when the actually baby doesn't.

Reporting of the Results

The results take up to 2 weeks to be reported and are reported as "lower risk" or "increased risk". Lower risk is defined as having a less than 1 in 150 chance of having a baby with Down syndrome this incorporates approx. 95% of women. "Lower risk" results are reported back directly to woman.

"Increased risk" results are reported back to the antenatal clinic and the Named Consultant who will meet with the woman at the next available appointment to counsel, refer to genetics and Fetal Medicine where an

amniocentesis would be offered to confirm the diagnosis. If the woman does not wish to have an amniocentesis the option of having non-invasive prenatal testing (NIPT) would be discussed as this test would report up to 99% accuracy rate when screening for Down syndrome (this would have to be funded by the woman).

- **NIPT** (Harmony Test £295-£385) is a blood test that can be taken after the 10th week of pregnancy. The test can be used in singleton, twin and egg donor pregnancies and has been approved for use on women aged between 18-48 years of age. This test analyses cell free fetal DNA within the mother's blood and gives a strong indication of whether the fetus has a "high probability" or" low probability" of having the following conditions-

    - T21 (Down syndrome 99%)
    - T18 (Edwards syndrome 97%)
    - T13 (Patau syndrome 94%)
    - Sex chromosome aneuploidy (Klinefelter syndrome 99%)
    - Monosomy X (Turner syndrome 99%)
    - Fetal sex (99%)
    - 22q 11.2 deletion also known as DiGeorge syndrome (75%)

Anomaly Scan

All women are offered a routine anomaly scan between 18weeks - 20 weeks plus 6 days, this is normally carried out around 20weeks of pregnancy. Any anomalies detected are explained at the time to the woman and referral made to Fetal medicine were review or date of appointment will be arranged within 5-7 working days. Upon suspected diagnosis of a fetal anomaly the ultra-sonographer will email a copy of the referral form to the ANSC and the named consultant. As this can be a very anxious time for women they are given written information upon referral with the Maternity OPD and the ANSC contact details. The ANSC would telephone each woman referred to Fetal medicine to provide a point of contact for the woman and offer support.

Amniocentesis

Before having the test, women are given a clear explanation of the testing process and had discussed the possibility of receiving uncertain results.

Women who have under gone an amniocentesis with Fetal medicine will have results reported back to the ANSC who will inform the woman and the named consultant of the results. If the result identifies a genetic condition the woman

will be counselled face to face about the baby's condition, she will be provided with written information about the condition and how she feels about continuing the pregnancy will be sensitively explored. All women will be given time to process the test results as most women are overwhelmed by a diagnosis and uncertain of what this means long term.

For women who decided not to have an amniocentesis due to the risk of miscarriage and have had a "high probability" result from NIPT would be offered genetic testing with the baby's cord blood at delivery. This confirms or excludes the diagnosis for the parents early on and ensures paediatric follow up is implemented if required.

In addition, I attach Circular dated 21 October 2009 referencing NICE Clinical Guideline No 62 - Antenatal Care. The advice in the NICE guideline covers the routine care that all healthy woman can expect to receive during their pregnancy. Was this guidance implemented within the Trust and if not what were the reasons that this was not implemented?

The NHSCT adheres to the regional standard for antenatal screening and in keeping with this has not implemented the practice of offering routine antenatal screening for Down syndrome to all pregnant women.