AWMGS
All Wales Medical Genomics Service

Pilot study – Non-Invasive-Prenatal Testing (NIPT) in pregnancies with fetal anomalies

BACKGROUND

The All Wales Genomics Laboratory is conducting a pilot study to evaluate the use of NIPT for pregnancies in the presence of certain fetal anomalies on ultrasound scan.

The laboratory already offers a NIPT service for pregnant women in Wales who have an increased chance (>1 in 150) of trisomy 13, 18 and/or 21 following combined or quad screening. NIPT testing is also offered by the NHS in Wales at the request of the Prenatal Clinical Genetics Service if the patient has a child or a previous pregnancy with trisomy 13, 18 or 21, or either of the parents are a carrier of a Robertsonian translocation involving chromosomes 13 and 21.

The purpose of the pilot study is to evaluate the clinical utility of extending our service to include pregnancies with certain fetal anomalies in instances where **the pregnant woman does not wish to have an invasive test.** The test may be requested by the patient's obstetric team (Clinical Genetics approval is not required). The duration of the study will be one year, commencing with immediate effect.

In order to participate in the pilot study, the patient will need to meet the following criteria;

STUDY ACCEPTANCE CRITERIA

Pregnant patient does not wish to have an invasive test **AND** the ultrasound scan shows one of the following;

- Multiple fetal anomalies strongly suggestive of trisomy 13, 18 or 21
- Atrioventricular Septal Defect (AVSD) as a sole anomaly
- Exomphalos as a sole anomaly
- Duodenal atresia associated with other anomalies
- Isolated pleural effusion
- Isolated cystic hygroma*

* The NIPT test does not check for Turner syndrome which is an important cause of cystic hygroma.

The following have been EXCLUDED;

- Blood transfusion within 4 months
- Transplant surgery
- Immunotherapy/ stem cell therapy
- Maternal malignancy
- Multiple pregnancy >2 fetuses
- Pregnancy with vanishing twin

HOW TO REFER A PATIENT FOR THE STUDY

1. Complete NIPT referral form for fetal anomalies (pilot study) - *available on laboratory website – see below*
2. Collect 7-10mls of maternal blood in a Streck tube (tubes available on request)
3. Send the sample and accompanying request form to the All Wales Genomics Laboratory (full address below). The sample must be received by the laboratory within 5 days of sampling.
4. Please inform the laboratory that the sample is on its way; by telephone: 029 218 44072 or email Lab.genetics@wales.nhs.uk

The report will be returned to the referring clinician within a turnaround time of up to 14 calendar days. Please ensure that you have provided an email address for the return of the report in the appropriate box on the referral form.

If you have any questions or require any additional information please contact the laboratory.

Contact details

All Wales Genomics Laboratory

Canolfan Iechyd Genomig Cymru/Wales Genomic Health Centre

Cardiff Edge Business Park

Longwood Drive

Whitchurch

CARDIFF

CF14 7YU

Tel: 029 2184 2641

Fax: 029 2184 4043

Lab.genetics@wales.nhs.uk