

## Non-Invasive Prenatal Testing (NIPT)

NIPT is a safe non-invasive prenatal test which evaluates whether a pregnancy has a high chance of trisomy 13 (Patau syndrome), trisomy 18 (Edwards syndrome) or trisomy 21 (Down syndrome). Although NIPT is not diagnostic, NIPT identifies more than 99% of babies with Down's syndrome, 97% with Edward's syndrome and 87% with Patau syndrome. Testing is performed on a blood sample from the mother. The test can be performed as early as 10 weeks gestation.

### NIPT testing criteria

- NIPT is offered routinely by the NHS to 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 is now available for twin pregnancies

NIPT 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
- Either of the parents are a carrier of a Robertsonian translocation involving chromosomes 13 and 21

These patients should be referred to the Clinical Genetics Service when thinking about another pregnancy, or as early as possible during pregnancy, to discuss their testing options. The referral should be made using the standard All Wales Medical Genomics Service referral form.

### NIPT testing requirements

The test is NOT suitable for the following pregnancies;

- If the mother has a known malignancy
- If the mother has a chromosome anomaly
- If the mother has undergone a blood transfusion in the last four months
- If the mother has undergone transplant surgery, immunotherapy or stem cell therapy

### Sample requirements

10mls of maternal blood in a STRECK tube (tube available on request from the laboratory). The samples must be labelled with three identifiers and sample collection date and time. Samples must be accompanied by a NIPT referral form (available on website).

Consent for genetic testing is assumed when the request for testing is received. It is the responsibility of the referring clinician to ensure that appropriate consent has been obtained.

### Turnaround time (TAT) – 14 calendar days

Please contact the laboratory directly on 029 2184 4023 should you want further information on how to access this test.

**Contact details**

All Wales Genomics Laboratory  
Institute of Medical Genetics,  
University Hospital of Wales  
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<http://www.wales.nhs.uk/AWMGS/>