Rapid Aneuploidy Testing

- Quantitative Fluorescent Polymerase chain Reaction (QFPCR) analysis examines highly polymorphic repetitive sequences found within extracted DNA and is used to identify the three most common viable autosomal trisomies: trisomy 21 (Down syndrome), trisomy 18 (Edwards syndrome) and trisomy 13 (Patau syndrome).
- This test is carried out on all chorionic villus samples, foetal and neonatal bloods and on amniotic fluids by request.
- The main indication for this test is the suspicion of a chromosome aneuploidy syndrome (trisomy 13, 18 or 21), this may be identified prenatally by:
  - Antenatal Down syndrome screening
  - Abnormalities detected on ultrasound scan
  - A previous aneuploid pregnancy
  - Parent known to carry a chromosome abnormality which gives a risk of a specific aneuploidy syndrome.

- Postnatally it will be carried out on all neonates with a suspected chromosome abnormality.
- The standard test identifies trisomy 13, 18, 21. Additional testing may be undertaken to identify the sex chromosome complement.
- The test does not detect mosaicism or structural abnormalities involving these or other chromosomes.
- This test is usually carried out in conjunction with full chromosome analysis of cultured cells where initial sample size is sufficient for both tests.
- The additional Sex chromosome testing will be undertaken:
  - Prenatally where testing is for an X linked disorder
  - Where ultrasound scan or postnatal examination is suggestive of Turner syndrome
  - Postnatally on babies with indeterminate sex at birth.

Patients should be advised that:
- Maternal contamination (prenatal samples), mosaicism or other factors may complicate analysis of QFPCR.
- Samples may fail to give a results (usually due to maternal cell contamination).
- Abnormalities may be detected on the full chromosome analysis that could not be detected by QFPCR analysis.

Target reporting Times

Anticipated reporting time: 1-3 working days from receipt of the sample. Samples must arrive at the laboratory by 2.30pm to be processed on the day of receipt.

Please contact the laboratory for up to date prices.

Laboratory contact: For enquiries contact: Lisa.burvill-holmes@nbt.nhs.uk