Bristol Genetics Laboratory is a UKAS accredited medical laboratory No.9307.

North Bristol NHS Trust

Rapid Aneuploidy Testing

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Sample Required

See Sample requirements page at www.nbt.nhs.uk/genetics **for full details**

Amniotic fluid: 15-20 ml in sterile, leak proof, plain (no anticoagulant) plastic universal container (Sterilin or Nunc are recommended)

Chorionic Villus Sample (CVS): 10-25 mg should be sent in transport media supplied by the laboratory

Blood: at least 1ml in lithium heparin

Samples should be accompanied by a FULLY completed request form (available as download at www.nbt.nhs.uk/genetics or from the laboratory).
Please include details of the test required, family history, address and POSTCODE, NHS number, referring clinician and centre.

Consent and Storage:

All genetic testing requires consent. **It is the responsibility of the referring clinician to ensure that appropriate consent has been obtained.**

Stored material from all referrals may be retained for quality assurance purposes and may be used anonymously for the development of new tests for the disorder in question.

Clinical Background and Genetics

Quantitative Fluorescent Polymerase chain Reaction (QF-PCR) analysis examines highly polymorphic repetitive DNA sequences 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) and sex chromosome complements.

Service Offered

This test is carried out on all chorionic villus samples, amniotic fluids and foetal blood samples. It may also be undertaken on neonatal bloods referred for cytogenetic investigation.
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

- Testing is carried out using the Elucigene® QST*R® Plus v2 assay. This test identifies trisomy 13, 18, 21 and the sex chromosome complement. Additional testing may be undertaken using additional Elucigene® QST*R® kits for some cases.
- The test does not exclude mosaicism or structural abnormalities involving these or other chromosomes.
- This test might be carried out in conjunction with array CGH analysis or full chromosome analysis of cultured cells where appropriate and when the initial sample size is sufficient for all tests.

Prenatal samples referred from pregnancies with no indication of a structural abnormality will not have any further testing after a normal QF-PCR result. Abnormal QF-PCR results will be followed up by full chromosome analysis.

Patients should be advised that:

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

Target reporting Times

3 calendar days from receipt of the sample.

Samples must arrive at the laboratory by 2.30pm to be processed on the day of receipt.

Quality

BGL participates in all appropriate UK EQA schemes for this service.

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