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

Prenatal Genetic Testing

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

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

Foetal Blood: at least 1ml in lithium heparin

Parental blood samples (5-7mls EDTA):
- If the fetal sample is blood stained, a maternal blood is required.
- For referrals with structural abnormality or NT ≥ 3.5mm parental bloods are required.

Samples should be accompanied by a FULLY completed request form (available as download at www.nbt.nhs.uk/genetics or from the laboratory).

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
The main indication for Prenatal Genetic Testing is a pregnancy that has been identified as being at risk of a genetic disorder. This risk may have been identified by:
- Antenatal Down syndrome screening
- Abnormalities detected on ultrasound scan
- A family history of a chromosome abnormality

Service offered: Please refer to Prenatal Genetic Testing for full details.
- Rapid Aneuploidy Testing for common trisomies by QF PCR on all prenatal samples.
- **Referrals without structural abnormality**: QF-PCR only, with no further testing.
- **Referrals with structural abnormality or NT ≥ 3.5mm:** Microarray CGH (aCGH) in the event of a normal QF-PCR result (or sex chromosome aneuploidy that does not explain the scan findings).
- Chromosome analysis by karyotyping is only performed for confirmation of abnormal QF-PCR results or at the request of clinical genetics.
- Cell culture may be performed for referral to other laboratories for additional testing using DNA or biochemical analysis. Please contact Laboratory to discuss your specific requirements.
- Long term storage of cultured cells suitable for DNA analysis or biochemical analysis is carried out where specifically indicated

Referrals
Referrals are accepted from Obstetrics and Clinical Genetics

Target reporting Times

Rapid aneuploidy testing by QF PCR: 3 calendar days

Microarray CGH or karyotype: 14 calendar days

**Low gestation (15 wks) AF samples and small CVS samples may take longer to achieve a result by aCGH.*

Quality
- BGL participates in all appropriate UK NEQAS EQA schemes for this service

Laboratory Contact: Catherine Delmege (0117 414 6149)
Clinical Advice: If clinical discussion is required we recommend you contact the Clinical Genetics service.

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DETAILS CORRECT AT DATE OF PRINTING ONLY.
Approved by: Catherine Delmege