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

North Bristol NHS Trust

Analysis of Solid Tissues

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Sample Requirements:
Tissue culture transport medium is available from the Laboratory by arrangement Samples must not be sent in formal saline or other preservative. If transport media is unavailable sterile normal saline may be used.

See sample requirements page at www.nbt.nhs.uk/genetics

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 tests, family history, address and POSTCODE, NHS number,

Consent and Storage: (DNA and Cultured Cells)

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

Fibroblast cultures can be used to supplement blood cultures in patients who may have mixed cell types (mosaics), or for elucidating the karyotype in situations where blood cannot be obtained, e.g. pregnancy loss, aborted tissue, stillbirths or any type of post mortem study. Cultured cells can also be sent to other laboratories for the diagnosis of some inherited metabolic disorders.

Referrals may include the following;

- Stillbirths or miscarriages with any dysmorphic features
- Stillbirths or miscarriages where parents are known to carry chromosome rearrangements or abnormalities
- Early spontaneous abortions with a history of 3 recurrent miscarriages
- Confirmation of karyotype detected at prenatal diagnosis
- Termination of pregnancy for fetal abnormalities.
- Suspected biochemical defect (tissue will be cultured for metabolic studies and stored by prior arrangement)

Service Offered

Analysis will usually be by molecular genetic methods and may include:

- QF-PCR to detect common trisomies (13, 18 and 21), triploidy and sex chromosome aneuploidy; this test will also identify complete molar pregnancies
- Microarray CGH.. This will identify all trisomies and unbalanced rearrangements where there is gain or loss of a region .This technique does not detect balanced translocations or rearrangements that do not alter the copy number of the probe target sequences. It may detect mosaicism.
- In situ hybridisation using specific probes may be carried out where clinically indicated
- Cells will be cultured and frozen in long term storage for future testing only where specifically requested and according to the laboratory cell storage policy
- For live patients, where mosaicism is suspected, cells will be cultured and chromosome analysis or arrayCGH carried out.
- Samples where biochemical analysis is indicated will be cultured and frozen in long term storage; these may at any time be used for DNA extraction or referred on to specialist laboratories for biochemical testing.

Clinical Advice If clinical discussion is required we recommend you contact the Clinical Genetics service.

Target reporting Times

QFPCR and ArrayCGH report 42 days
Chromosome analysis (karyotyping) 42 days

Quality

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

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Information document No.60 Version 6
Active date of this version 30/07/2019
DETAILS CORRECT AT DATE OF PRINTING ONLY.
Approved by: Catherine Delmege