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

Postnatal Cytogenetic Analysis

North Bristol NHS Trust

Contact details:
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Head of Department:
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Consultant Lead for Rare Disease:
Maggie Williams, FRCPath

Service Lead: Catherine Delmege
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Sample Required:
Adult: 3-5ml blood in lithium heparin
Paediatric: at least 1ml blood in lithium heparin (preferably>2ml)

Samples should be accompanied by a FULLY completed request form, available as download at www.nbt.nhs.uk/genetics

Please include details of referral reason, tests, family history, address and POSTCODE, NHS number, referring clinician and hospital.

Consent and Storage:
(DNA and Cell suspension)

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
Chromosome analysis by karyotyping may be indicated in the following groups of referrals:

Paediatric referrals: Disorder of sex development (DSD) in neonates, delayed puberty or other atypical sexual development.

Adult referrals:
Infertility and family history of a chromosome abnormality.

Testing is carried out by **arrayCGH** for patients with intellectual disability/learning difficulties/developmental delay and for patients with dysmorphism and/or congenital abnormalities

Service offered

- Chromosome analysis by karyotyping
- Rapid aneuploidy testing by QF PCR (available for urgent referrals where appropriate, see below)
- Specific testing by fluorescence in situ hybridisation (FISH):contact the laboratory for further information
- Array Comparative Genomic Hybridisation (aCGH) testing – see separate proforma,

Urgent Referrals

The following referrals will be classed as urgent

- Patient presenting in pregnancy with family history of chromosome abnormality at birth
- Indeterminate gender at birth
- Neonates with a suspected chromosome abnormality
- Parents of a structural chromosome rearrangement detected at prenatal diagnosis
- Specific clinical need (please indicate this on the referral form or contact laboratory)

Target reporting Times

Rapid aneuploidy testing by QF PCR: TAT 3 working days
Urgent chromosome analysis: TAT 14 calendar days
Routine chromosome analysis: TAT 42 calendar days

For an up to date prices please contact the laboratory

Quality

- BGL participates in the appropriate UK NEQAS schemes for postnatal cytogenetic referrals.

Clinical Advice: If clinical discussion is required we recommend you contact the Clinical Genetics service.
Laboratory contact: For enquiries contact Catherine.Delmege@nbt.nhs.uk

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