

Postnatal Cytogenetic Analysis

Contact details:

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Pathology Sciences
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Head of department:

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Molecular Genetics:

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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 10 calendar days
Routine chromosome analysis: TAT 28 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