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

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

**Laboratory contact:** For enquiries contact
Catherine.Delmege@nbt.nhs.uk

---

Information document No 54 Version 10
Active date of this version 19/02/2018

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

*Exceptional healthcare, personally delivered*