

Disorders of sexual development/infertility

Contact details

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Samples required

- 1-2ml venous blood in plastic lithium heparin tube
- A completed request card should accompany all samples. Please provide clinical details

Patient details

To facilitate accurate testing and reporting please provide patient demographic details (full name, date of birth, address), details of any relevant family history (including proband's details) and full contact details for the referring clinician

Introduction

Cytogenetic testing can be requested if the patient has abnormal sexual development (e.g. delayed puberty), clinical features suggestive of a sex chromosome abnormality (e.g. Turner syndrome, Klinefelter syndrome), or is experiencing infertility.

Referrals

- Ambiguous genitalia/indeterminate gender (for neonates, can be in conjunction with 'rapid' testing for karyotypic sex)
- Delayed puberty or inappropriate secondary sexual development
- Short stature or amenorrhoea in females
- Oligozoospermia or azoospermia in males
- Premature ovarian failure
- Sperm and egg donors for NHS funded patients
- Couples undergoing assisted conception funded by the NHS

Service offered

The format of the service offered will vary depending on the patient's clinical features. The testing is performed by karyotyping (full or targeted), and additional techniques (e.g. fluorescent in situ hybridisation) may be used if an abnormality is suspected or detected during karyotyping.

Technical

For karyotyping, blood cultures are grown and harvested to yield metaphase cells which are analysed using light microscopy.

If the reason for referral is consistent with the possibility of a sex chromosome abnormality but not an autosome abnormality (e.g. short stature in a female; query Klinefelter syndrome) a targeted 30 cell score for the sex chromosomes is performed (not a full karyotype). A full karyotype is performed if the sex chromosomes and autosomes need to be examined.

Target reporting time

Routine analysis – 28 days

If clinical need indicates that an urgent result is required (e.g. current ongoing pregnancy) – 10 days. Please contact the laboratory for urgent cases.