Neonate testing (rapid) for karyotypic sex and common aneuploidies

Introduction

Neonatal testing for karyotypic sex and common aneuploidies is performed upon specific clinician request (using the referral form) in order to provide a rapid result.

Referrals

- Neonates (<3 months of age) with clinical features suggestive of trisomy 13 (Patau syndrome), trisomy 18 (Edwards syndrome), or trisomy 21 (Down syndrome)
- Neonates (<3 months of age) with ambiguous genitalia

NB testing is not offered for a phenotypically normal baby on the basis of an increased risk of Down syndrome suggested by prenatal screening

Service offered

Testing is offered to neonates with appropriate clinical features (as above).

Technical

The testing is usually performed on interphase cells using FISH probes specific to the chromosomes/regions of interest.

As an alternative to interphase FISH, aneuploidy testing can be performed by karyotyping metaphase cells.

The testing is targeted, and therefore does not exclude the possibility of additional chromosome abnormalities, or investigate the possibility of mosaicism.

If the aneuploidy result is abnormal, karyotyping of metaphase cells will be performed to confirm the result. If the aneuploidy result is normal, further testing will be performed to identify the presence of other chromosome abnormalities; this will be done by karyotyping or microarray depending on the sample received and the clinical phenotype of the patient.

Target reporting time

3 calendar days from date of sample receipt for rapid result