UK Genetic Testing Network

Clinical Cytogenetics – Core Services (July 2015)

The UKGTN has worked with the Association for Clinical Genetic Science, through the UKGTN Laboratory Membership and Audit Working Group, to draw up a list of tests (detailed below) that are considered to be commonly offered by cytogenetics laboratories. They are normally available in every laboratory as a fully funded service.

Laboratories can apply to provide tests not listed below by completing a genetic test evaluation form (gene dossier) for the specific test. The gene dossier will be evaluated by the Genetic Test Evaluation Working Group. Currently this refers to constitutional testing only.

The core services listed are undertaken by karyotype analysis, microarray analysis or by targeted tests (QF-PCR, FISH & MLPA), as appropriate.

Prenatal testing

Abnormal prenatal ultrasound scan analyses
At risk patients identified from Down screening
Culture for investigations by other disciplines
Family history of chromosome rearrangement

Postnatal testing
Neonatal/paediatric/adolescent investigations

Ambiguous genitalia/indeterminate gender.
Chimerism testing
Delayed puberty, or disorders of secondary sexual development.
Dysmorphism
Growth delay
Intra uterine death
Muscular/neurological presentation with possible cytogenetic cause
Phenotypic features suggestive of chromosomal mosaicism
Prenatal follow-up investigations
Recognised microdeletion/microduplication syndromes
Recognised syndromes with a cytogenetic aetiology
Short stature and / or amenorrhoea in females.
Unexplained learning difficulties/developmental delay

Postnatal testing
Adult investigations

Chimerism testing
Couples undergoing assisted conception for NHS funded patients only
Dysmorphism
Family follow up investigations

Family history of a known chromosome abnormality other than simple aneuploidy due to non-disjunction
Growth delay
Intra uterine death
Muscular/neurological presentation with possible cytogenetic cause
Oligozoospermia or azoospermia in males.
Parental analysis after pregnancy loss of a fetus with multiple congenital abnormalities or severe IUGR or unexplained stillbirth/neonatal death
Parental analysis after three or more unexplained miscarriages and clinical indication to investigate (eg family history).

Premature ovarian failure.
Prenatal follow-up investigations
Recognised microdeletion/microduplication syndromes
Recognised syndromes with a cytogenetic aetiology
Sperm and egg donors for NHS funded patients only.
Suspected family history of chromosome abnormality where the chromosome abnormality of the affected individual is not known.
Unexplained learning difficulties/developmental delay with dysmorphism or congenital anomalies

Post mortem perinatal, fetal or placental testing for chromosome imbalance

Cases of perinatal death under investigation by the Coroner when requested.
Confirmation of complete or partial molar pregnancies
Fetuses/stillbirths/neonatal deaths with congenital abnormality suggestive of a chromosomal anomaly or with IUGR.
Follow up confirmation of prenatal cytogenetic findings on post termination tissue.
Follow up of prenatal ultrasound findings suggestive of a chromosomal anomaly.
Spontaneous abortion less than 16 weeks where the couple is receiving assisted conception and/or treatment for recurrent pregnancy loss.
Spontaneous abortion where the couple has a known chromosome rearrangement.
Third (or more) unexplained miscarriage (<16 weeks)
Unexplained IUD/spontaneous abortion (³16 weeks).
Unexplained stillbirth or neonatal death (³24 weeks).

Other tests provided when clinically indicated.

Cell culture as a prelude to investigations by other laboratories/services.
Chromosome breakage syndromes.
Confirmation or further characterisation of a chromosome anomaly
Cryptic chromosomal imbalance investigations