

CYTOGENETICS



Patient's Name Label

For Downtime Use:

Name:

MRN:

Account Number:

Date of Birth:

Sex: M / F (Circle One)

Ward/Bed: _____ Clinic: _____ Class: _____

 Patient Type Gynae Obst Neo
 Paed Medicine Paed Surgery

Laboratory Barcode

Clinical Indications for test

For Laboratory Use Only

Relevant History/Findings/Treatment

Name & Signature of Requesting Doctor

Pager / Contact No (indicate if urgent)

Name of Consultant I/C

Date

Type of Specimen

Please specify:

Date & Time Specimen Taken

Date _____ Time _____ am / pm

Please (tick) appropriate boxes below

CHROMOSOME ANALYSIS

CY0170	<input type="checkbox"/>	Peripheral blood*
CY0010	<input type="checkbox"/>	Amniotic fluid*
CY0050	<input type="checkbox"/>	Chorionic villi*
CY0103	<input type="checkbox"/>	Fetal blood (cordocentesis)*
CY0171	<input type="checkbox"/>	Product of conception (POC)*
CY0171	<input type="checkbox"/>	Skin Biopsy*
CY0020	<input type="checkbox"/>	Bone marrow
CY0020	<input type="checkbox"/>	Leukemic Blood
CY0024	<input type="checkbox"/>	Lymph node
CY0023	<input type="checkbox"/>	Solid cancer tissue / tumour
CY0026	<input type="checkbox"/>	Cell line / stem cell

CHROMOSOME MICROARRAY ANALYSIS

CY0300	<input type="checkbox"/>	Prenatal SNP Array with Limited Karyotype*#
CY0301	<input type="checkbox"/>	Prenatal SNP Array#
CY0302	<input type="checkbox"/>	CytoScan 750K Array (Further study for Prenatal Array)^
CY0302	<input type="checkbox"/>	CytoScan 750K Array (Product of Conception)^
CY0303	<input type="checkbox"/>	Molecular Inversion Probe Array
CY0303X	<input type="checkbox"/>	Molecular Inversion Probe Array with DNA Extraction
DNA120	<input type="checkbox"/>	Chromosome Microarray Analysis^ (Refer to Page 2)

SPECIAL STUDIES

CY0200	<input type="checkbox"/>	Tissue culture
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FISH - NEUROBLASTOMA

CY0214	<input type="checkbox"/>	NMYC (2p24.1) dual colour
CY0213	<input type="checkbox"/>	Neuroblastoma panel (3 probes) MYCN/1p/11q

FISH - NEUROBLASTOMA on paraffin sections

CY0215	<input type="checkbox"/>	NMYC (2p24.1) dual colour
CY0218	<input type="checkbox"/>	Neuroblastoma panel (3 probes) MYCN/1p/11q

FISH for TUMOUR on paraffin sections

CY0215	<input type="checkbox"/>	EWSR1 (22q12) Break Apart
CY0215	<input type="checkbox"/>	RELA (11q13.1) Break Apart
CY0215	<input type="checkbox"/>	c11orf95 (11q13.1) Break Apart
CY0219	<input type="checkbox"/>	HER2 (ERBB2) DNA probe †
CY0215	<input type="checkbox"/>	Others, please contact lab

FISH - WITH CHROMOSOME ANALYSIS *

CY0101	<input type="checkbox"/>	DiGeorge Region Probe (TUPLE1/HIRA)
CY0101	<input type="checkbox"/>	Williams Region Probe (ELN)
CY0101	<input type="checkbox"/>	PWS/AS Region Probe (SNRPN)
CY0101	<input type="checkbox"/>	Wolf Hirschhorn Region Probe (WHS)
CY0101	<input type="checkbox"/>	Cri du Chat Region Probe (EGR1)
CY0101	<input type="checkbox"/>	22q13 Deletion Probe (ARSA)
CY0101	<input type="checkbox"/>	13q14 Patau Region Probe (RB1)
CY0101	<input type="checkbox"/>	21q22.13-q22.2 Down Region Probe (D21S342)
CY0101	<input type="checkbox"/>	Centromeric 18 Probe
CY0101	<input type="checkbox"/>	Centromeric X and Y Probe (CEP X, Y)
CY0101	<input type="checkbox"/>	SRY probe

FISH - (METAPHASE) ADD-ON *

CY0211	<input type="checkbox"/>	DiGeorge Region Probe (TUPLE1/HIRA)
CY0211	<input type="checkbox"/>	Williams Region Probe (ELN)
CY0211	<input type="checkbox"/>	PWS/AS Region Probe (SNRPN)
CY0211	<input type="checkbox"/>	Wolf Hirschhorn Region Probe (WHS)
CY0211	<input type="checkbox"/>	Cri du Chat Region Probe (EGR1)
CY0211	<input type="checkbox"/>	22q13 Deletion Probe (ARSA)
CY0211	<input type="checkbox"/>	13q14 Patau Region Probe (RB1)
CY0211	<input type="checkbox"/>	21q22.13-q22.2 Down Region Probe (D21S342)
CY0211	<input type="checkbox"/>	Centromeric 18 Probe
CY0211	<input type="checkbox"/>	Centromeric X and Y Probe (CEP X, Y)
CY0211	<input type="checkbox"/>	SRY probe
CY0211	<input type="checkbox"/>	Others / customized probe, please contact lab

FISH - (INTERPHASE DIRECT)

CY0210	<input type="checkbox"/>	Trisomy 13 (RB1)*
CY0210	<input type="checkbox"/>	Trisomy 21 (D21S342)*
CY0210	<input type="checkbox"/>	Trisomy 18 (CEP 18)*
CY0210	<input type="checkbox"/>	Centromeric X and Y Probe (CEP X, Y)*
CY0216	<input type="checkbox"/>	Aneuploidy panel (Trisomy 13, 18, 21, X and Y)*
CY0217	<input type="checkbox"/>	Aneuploidy panel (Trisomy 13, 18, 21, X and Y) on paraffin*
CY0210	<input type="checkbox"/>	Others, please contact lab

† If ordering HER2 testing, please indicate the following:

IHC result: [] 0 [] 1+ [] 2+ [] 3+

Cold ischaemia timing: _____

Fixation timing: _____

* Consent form: "Karyotyping/ Fluorescence In Situ Hybridisation (FISH) For Constitutional Genetic Testing" (85060-FM-MB-103)

Consent form: "Prenatal Chromosome Microarray Analysis" (85060-FM-MB-104)

^ Consent form: "Constitutional Genetic Testing (General Genetics)" (85060-FM-MB-101)

Please send specimens to Cytogenetics Laboratory, Basement 1, Children's Tower. For enquiries, please call 6394-1392.

DNA120: Chromosome Microarray Analysis (180K High Resolution Copy Number)

Clinical indications: Please list all the clinical features the patient is known to have

Family History

- Parents consanguineous
 Parents with ≥ 2 miscarriages
 Other relatives with similar clinical history
 Explain: _____

Perinatal History

- IUGR
 Non-immune hydrops fetalis
 Oligohydramnios
 Polyhydramnios
 Prematurity
 Other: _____

Developmental

- Fine motor delay
 Gross motor delay
 Speech delay
 Other: _____

Cognitive

- Intellectual disability/MR
 List IQ, if known: _____
 Learning disability
 Other: _____

Behavioural/Psychiatric

- Attention deficit hyperactivity disorder
 Autism spectrum disorder
 Other behavioural/ psychiatric abnormality
 Specify: _____

Karyotype:

- Not done
 Normal: 46,XX / 46,XY
 Others: _____

Hearing/Vision

- Abnormality of Vision
 Specify: _____
 Abnormality of Eye Movement
 Specify: _____
 Hearing loss
 Specify: _____
 Other: _____

Growth

- Failure to thrive
 Overgrowth
 Short stature
 Other: _____

Cardiac

- Atrial septal defect
 Coarctation of the aorta
 Tetralogy of Fallot
 Ventricular septal defect
 Other: _____

Craniofacial

- Cleft lip
 Cleft palate
 Coloboma of eye
 Craniosynotosis
 Dysmorphic facial features
 Specify: _____
 Ear malformation
 Specify: _____
 Macrocephaly
 Microcephaly
 Other: _____

Cutaneous

- Hyperpigmentation
 Hypopigmentation
 Other: _____

Gastrointestinal

- Anal atresia/imperforate anus
 Gastroschisis
 Omphalocele
 Pyloric stenosis
 Tracheoesophageal fistula
 Other: _____

Genitourinary

- Ambiguous genitalia
 Cryptorchidism
 Hydronephrosis
 Hypospadias
 Kidney malformation
 Specify: _____
 Other: _____

Musculoskeletal

- Club foot
 Contractures
 Diaphragmatic hernia
 Limb anomaly
 Specify: _____
 Polydactyly
 Specify: _____
 Syndactyly
 Specify: _____
 Vertebral anomaly
 Specify: _____
 Other: _____

Neurological

- Cerebral palsy
 Encephalopathy
 Hypertonia/ spasticity
 Hypotonia
 Seizures
 Structural brain anomaly
 Specify: _____
 Other: _____