



**KK Women's and  
Children's Hospital**  
SingHealth

## CLINICAL GENETICS

Patient's name label

(For downtime use)

Name:

MRN:

Account number:

Date of birth:

Sex: M / F

Patient Type  Gynae  Obst  Neo

Paed Med  Paed Surg

Ward/Bed: \_\_\_\_\_ Clinic: \_\_\_\_\_ Class: \_\_\_\_\_

### Clinical Diagnosis:

Relevant History / Findings/Family history:

For laboratory use

For thalassaemia tests:

MCV: \_\_\_\_\_ Hb: \_\_\_\_\_

Hb electrophoresis: HbA2 \_\_\_\_\_ HbF: \_\_\_\_\_

Name & signature of requesting doctor  
Contact no. (if urgent)

Type of specimen:  Peripheral blood  Amniotic fluid  
 Bone marrow  Chorionic villus sample  
 Leukemic blood  Fetal blood  
 Lymph node  Product of conception  
 Skin biopsy  Fetal tissue: \_\_\_\_\_  
 Others: \_\_\_\_\_  Embryonic stem cell lines

Name of consultant i/c

Date & time specimen taken

Date

Date: \_\_\_\_\_ Time: \_\_\_\_\_

**Please tick appropriate box/es below and delete where not applicable.**

### CYTOGENETIC TESTS (Lab no: 63941392)

#### Karyotype (Chromosome analysis)

- CY0170\*  Peripheral blood  
 CY0010  Amniotic fluid  
 CY0050  Chorionic villus sample (CVS)  
 CY0103\*  Fetal blood  
 CY0171  Product of conception /  
 fetal tissue: \_\_\_\_\_  
 CY0020  Bone marrow / Leukemic blood\* / Lymph node /  
 Tumour / Embryonic stem cell line

#### Fluorescence in-situ hybridization (FISH)

- CY0210\*  Direct FISH without karyotype  
 CY0211\*  Microdeletion FISH add-on  
 CY0212\*  Add-on FISH  
 CY0101\*  FISH (includes karyotype)  
 DiGeorge / VCF / 22q11 deletion syndrome  
 Williams syndrome  
 Miller-Dieker syndrome  
 Smith-Magenis syndrome  
 Wolf Hirschhorn syndrome  
 Cri du Chat syndrome  
 Phelan syndrome (22q13 deletion)  
 Trisomy 13  
 Trisomy 18  
 Trisomy 21  
 X and Y  
 N-myc  
 TEL/AML1  
 Others: \_\_\_\_\_  
 CY0213  Neuroblastoma panel

#### Others

- CY0200  Tissue culture  
 Other cytogenetic test: \_\_\_\_\_

### DNA TESTS (Lab no: 63941395/6) CONSENT REQUIRED

#### DNA diagnostic tests for thalassaemia

- DNA 108\*\*  Thalassaemia DNA screen (12 mths and above)  
 include Hb electrophoresis, HbH inclusion bodies & DNA analysis for  
 5  $\alpha$ -thalassaemia deletion mutations.  
 Fresh EDTA blood (Adult 3mls X 2; Paeds 0.5ml X 2)  
 Send within 4 hrs of collection (Mon - Fri, 8am - 6pm)  
 DNA 101A\*\*  DNA analysis for  $\alpha$ -thalassaemia mutations  
 DNA 101B\*\*  DNA analysis for  $\beta$ -thalassaemia mutations  
 DNA 113A\*\*  DNA sequencing  $\alpha$ -globin genes  
 DNA 113B\*\*  DNA sequencing  $\beta$ -globin gene  
 DNA 102A\*\*  Prenatal test for  $\alpha$ -thalassaemia  
 DNA 102B\*\*  Prenatal test for  $\beta$ -thalassaemia

#### DNA diagnostic tests

- DNA 103\*\*  Huntington disease (HD)  
 DNA 104\*\*  Fragile X syndrome  
 DNA 105\*\*  Myotonic dystrophy (DM)  
 DNA 106\*\*  Spinocerebellar ataxia (SCA)  
 DNA 107\*\*  Spinal muscular atrophy (SMA)  
 DNA 114\*\*  Spinal muscular atrophy (SMA) carrier test  
 DNA 109\*\*  Y chromosome deletion  
 DNA 111\*\*  Achondroplasia  
 DNA 112\*\*  Kennedy's Disease (KD or SBMA)

#### DNA diagnostic test for other disease

(Tests in this category are only carried out with prior arrangement)

- DNA 001\*\*  Name of diseases: \_\_\_\_\_  
 DNA 002\*\*  Prenatal test for: \_\_\_\_\_  
 DNA 113\*\*  DNA sequencing for specific mutation in:  
 Gene: \_\_\_\_\_  
 (Report for identified family mutation necessary for this request)

#### DNA extraction

- DNA 003\*\*  0.5ml blood  
 DNA 004\*\*  3-5mls blood / chorionic villus sample (CVS)  
 DNA 005\*\*  Tissue: specify \_\_\_\_\_

### Prader-Willi syndrome / Angelman syndrome (Please circle one)

DNA110\*\*  Screening by DNA methylation test

CY0101\*  FISH analysis (includes karyotype)

**Tests marked \*** Sodium heparin tube (green-topped vacuette); send to Cytogenetics Laboratory, B1, Children's Tower. (6394-1392) Please call the lab to get an appointment before sending the sample.

**Tests marked \*\*** FILL UP CONSENT FORM ON NEXT PAGE

3ml EDTA tube (purple-topped vacuette) for blood samples; send to DNA Diagnostic and Research Laboratory, B1, Children's Tower. (63941395/6)

All DNA tests are developed "in-house", pending registration with HSA.

For after office hour queries on DNA tests, please contact Dr H Y Law through operator.



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**DNA Diagnostic & Research Laboratory  
Genetics Service**

### CONSENT FOR DNA TESTS

Name of Disease / Test: \_\_\_\_\_

I give consent for myself / my child to be tested for this condition.

I understand the following:

- 1 A blood/tissue sample will be collected for DNA tests for the above condition.
- 2 The potential benefit of this test is to confirm the diagnosis of the condition and to determine which other family members may be carriers or have increased risk of having the defective gene.
- 3 Erroneous results and incorrect interpretation may occur because of rare variation in the DNA of the individual and rare technical error.
- 4 Accurate interpretation of the DNA test result depends on correct information about the clinical diagnosis and about the biological relationships within the family.
- 5 DNA testing may reveal non-paternity, meaning that the stated father is not the biological father.
- 6 If technology improves and more mutations (gene defects) are detectable in future, I authorise the Laboratory to re-analyse, at the Laboratory's option, any remaining DNA for the same disease. If the sample is insufficient, my doctor may ask me for a fresh sample. There may be additional fees for such tests.
- 7 After DNA testing is completed, a small amount of my DNA may be made anonymous and used for medical education, quality control or research. Since the samples have been anonymised, any research results obtained cannot be reported to me.
- 8 In order to help me understand the test results, the results will be reported to me only through a doctor or genetic counsellor.
- 9 DNA results are strictly confidential and will not be released to anyone other than my doctors without my consent.
- 10 Some individuals who have chosen to have predictive DNA testing and been found to carry the gene leading to the disease have experienced discrimination (insurance, employment and social).

For prenatal testing, the following also apply:

- 1 This DNA test will determine the status of the fetus for this disease.
- 2 Besides rare DNA variation and the technical error, erroneous results may also arise from maternal contamination of the fetal sample.

For linkage analysis, the following also apply:

- 1 Linkage analysis is used in some cases where direct detection of the mutation (gene defect) cannot be performed.
- 2 Because of naturally occurring DNA recombination, the accuracy of linkage analysis in predicting carrier or affected status is not 100% and is usually reported as a probability.
- 3 Genetic markers used for linkage analysis may not be informative in every family. In this case, DNA testing cannot provide results for the family or some members of the family.

Signature \_\_\_\_\_

Doctor / nurse taking consent

Date \_\_\_\_\_

Signature \_\_\_\_\_

If person signing consent is the parent/guardian:

Name \_\_\_\_\_

Name \_\_\_\_\_

Designation \_\_\_\_\_

NRIC No \_\_\_\_\_

Relationship to patient \_\_\_\_\_