Reg No 198904227G

KK Women's and Children's Hospital
SingHealth

CLINICAL GENETICS REQUEST (DNA TESTS)

Patient's name label

(For downtime use)
Name:
MRN:
Account number:
Date of birth:
Sex: M / F

Patient Type ☐ Gynae ☐ Obst ☐ Neonate ☐ Paed Med ☐ Paed Surg
Ward/Bed: _____ Clinic:_____ Class:_____

Clinical Diagnosis:	Relevant History / Findings / Family history:	Accompanying sample (s):
Please call lab for presymptomatic testing.		Please specify relationship (spouse / sibling / parent / child etc)

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

Name of consultant i/c

Date

Type of specimen:
☐ Peripheral blood in EDTA (3ml unless otherwise specified)
☐ Amniotic fluid (Gestation:_____ Amount:_____ml)
☐ Chorionic villi (Gestation:_____ Amount:_____mg)
☐ Fetal blood (1ml)
☐ Others: _____

Specimen taken Date:_____ Time:_____

Prenatal specimen requirement for specific diseases:
10mg CVS / 20ml AF for β-thalassaemia
15mg CVS / 30ml AF for α-thalassaemia, DM, FX

Please tick appropriate boxes below and delete where not applicable.

CONSENT REQUIRED FOR ALL TESTS (SEE NEXT PAGE)

DNA diagnostic tests for thalassaemia

Hb: _____ MCV: _____ MCH: _____
Hb electrophoresis: HbA2 _____ HbF:_____

DNA 108 ☐ Thalassaemia DNA screen
Hb electrophoresis, HbH inclusion bodies & DNA analysis for 5 α-thalassaemia deletional 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 α-thalassaemia mutations
DNA 101B ☐ DNA analysis for β-thalassaemia mutations
DNA 113A ☐ DNA sequencing α–globin genes
DNA 102A ☐ Prenatal test for α-thalassaemia**
DNA 102B ☐ Prenatal test for β-thalassaemia**

DNA diagnostic tests for following diseases

DNA 103† ☐ Huntington disease (HD)
DNA 104 ☐ Fragile X syndrome (FX)
DNA 105† ☐ Myotonic dystrophy (DM)
DNA 106† ☐ Spinocerebellar ataxia (SCA) screen
DNA 106A† ☐ Spinocerebellar ataxia (SCA) type _____
DNA 107 ☐ Spinal muscular atrophy (SMA)
DNA 114 ☐ Spinal muscular atrophy (SMA) carrier test
DNA 109 ☐ Y chromosome deletion
DNA 110 ☐ DNA methylation test for (Please circle one)
Prader-Willi / Angelman syndrome
DNA 111 ☐ Achondroplasia
DNA 112† ☐ Kennedy's Disease (KD or SBMA)
DNA 115 ☐ Craniosynostosis (hotspots in FGFR1, 2, 3 & TWIST)
DNA 116 ☐ Specific craniosynostosis syndrome (circle one):
Apert / Pfeiffer / Crouzon syndrome
(Analyse hotspots in FGFR2)
DNA 119 ☐ Congenital adrenal hyperplasia (21OH deficiency)
DNA 122 ☐ Duchenne muscular dystrophy (DMD/BMD)

DNA extraction

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

DNA diagnostic test for other diseases

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

DNA 001 ☐ Name of disease: _____
DNA 113 ☐ Targeted sequencing for specific variant
(please enclose report)
Gene: _____
Variant: _____
Chromosome coordinate (hg19, eg chr1:12345678):

_____

For validation of research findings in the proband only and NOT for detecting familial mutation.

Other prenatal tests**

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

DNA 002 ☐ Prenatal test for (Please circle one)
- Fragile X syndrome
- Spinal muscular atrophy
- Myotonic dystrophy (Type 1)
- Others:

_____

DNA 117 ☐ QF-PCR
(rapid detection of chromosome aneuploidies of chromosomes 13, 18, 21, X & Y)

HLA Genotyping

DNA 118 ☐ HLA-B*1502
DNA 121 ☐ HLA-B*5801

Send specimen to:
DNA Diagnostic and Research Lab
Basement 1, Children's Tower
KK Women's & Children's Hospital
Tel: (65) 6394 1395/6
Fax: (65) 6394 1397

† **Not for presymptomatic testing unless specially arranged by a clinical geneticist.**
** **All prenatal requests MUST be pre-arranged. This is to ensure full information and appropriate type and amount of specimen(s) will be available to carry out the test.**

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 or Dr Y M Tan through operator.

KK Women's and Children's Hospital
SingHealth

DNA Diagnostic & Research Laboratory
Genetics Service

CLINICAL GENETICS

Patient's name label

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Name:
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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 (please select one option):

☐ I consent to a portion of the remaining DNA to be made anonymous and used for quality control, medical education and research. Since the sample has been made anonymous, any results obtained cannot be traced to the original source and no results will be reported.

☐ I do not wish any remaining DNA to be used for quality control, medical education and research. My blood sample and DNA will be discarded after the test.

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 thalassaemia mutation screening (for Singapore citizen and permanent resident only):

If my / my child's test is positive,

☐ I agree to be referred to the National Thalassaemia Registry (NTR) for registration.
☐ I do not agree to be referred to the National Thalassaemia Registry (NTR) for registration.

Signature _____
Date _____

If person signing consent is the parent/guardian:
Name _____
NRIC No _____
Relationship to patient _____

Doctor / nurse taking consent

Signature _____
Name _____
Designation _____