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:

Please call lab for presymptomatic testing.

Relevant History / Findings / Family history:

Accompanying sample (s):

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 thalassaemia, SMA, ACH
15mg CVS / 30ml AF for DM, FX

Please tick appropriate boxes below and delete where not applicable.

CONSENT REQUIRED FOR ALL TESTS

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 (2xAdult 3mls; 2xPaeds 0.5ml) (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 113B ☐ DNA sequencing β–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 114 ☐ Spinal muscular atrophy (SMA)
DNA 109 ☐ Y chromosome deletion
DNA 110 ☐ DNA methylation test for (Please circle one)
Prader-Willi / Angelman syndrome
DNA 111 ☐ Achondroplasia (ACH)
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/Becker muscular dystrophy (DMD/BMD)
DNA 126 ☐ CCHS: *PHOX2B* testing

DNA extraction

DNA 004 ☐ 3-5mls blood
DNA 005 ☐ Tissue/Cell culture/Amniotic fluid/CVS
Please 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: _____
RefSeq: _____
Chromosome coordinate (GRCh37 or GRCh38 please circle one):
_____

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

For lab use only

† **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) wil will be available to carry out the test.**

For after office hour queries on DNA tests, please contact Dr H Y Law / Dr Alexis Wang / Dr Yeo Tai Wai through operator.