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 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 (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 (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 / 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".
For after office hour queries on DNA tests, please contact Dr H Y Law or Dr Y M Tan through operator.

CONSENT FORM

CONSTITUTIONAL GENETIC TESTING (PRENATAL GENETICS)	ACCOUNT NO. NRIC NO. NAME ADDRESS SEX/BIRTH DATE/RACE DATE AND TIME OF ADMISSION

What is genetic testing?

Genetic testing is the analysis of genetic information of an individual.

Why do I need genetic testing?

You have been offered genetic testing to provide a genetic diagnosis for this pregnancy. The result of the genetic testing may be helpful for the doctors to better care for you and your pregnancy.

In your case, you are offered genetic testing for the following **suspected/ clinical genetic condition(s)**:_____

Name of the genetic test: _____

_____

What does genetic testing involve?

A chorionic villus / amniotic fluid sample (please delete accordingly) or _____ (sample) will be collected. The sample(s) will only be tested for the gene(s) / condition(s) listed above.

What are the possible results I could receive?

The results you could receive from genetic testing depend on the type of genetic test that was done. Some gene changes can result in a faulty gene and cause certain health problems, whereas other gene changes have no effect on health.

- **Genetic variant(s) identified**
  - A genetic change was identified
  - This confirms a genetic diagnosis and may be helpful for medical management.
  - This result may have implications for family members.
- **Genetic variant(s) NOT identified**
  - No genetic change was identified
  - This result may reduce likelihood of the genetic condition(s) tested for, but does not completely eliminate the possibility of such condition(s), or other genetic condition(s) that were not analysed. Further testing may be required.
  - This result could also be due to limitations in current technology and/or knowledge.
- **Variant(s) of Uncertain Significance (VUS) identified**
  - A genetic change was identified, however, there is insufficient information about the genetic change to associate it with an abnormal outcome, at the time of testing.
  - This result will not be used to direct medical management, unless deemed significant by your medical doctor.
  - Testing other family members may be helpful to clarify if this result is truly associated with a genetic condition / predisposition to disease.

  - This variant may be reclassified over time, when more information is available. The laboratory may issue a revised report / addendum, and you will be informed.
- **Incidental findings**
  - These are genetic changes that may not be related to the reason for testing. This may or may not have significant implications on the outcome of the pregnancy.
  - You have a choice whether you wish to receive such findings, if any.

What precautions must I take for genetic testing?

If a separate blood draw is required from you and/or the biological father, please inform the doctor/genetic counsellor if either of you have had a bone marrow transplant or blood transfusion performed in the past.

What are the *potential* risks and limitations of genetic testing?

The genetic testing and results may come with some risks and/or limitations:

- The test result can confirm a genetic diagnosis for this pregnancy but cannot determine if, or when, the symptoms will manifest, nor can it provide information on the disease severity or recurrence.
- The test result may not only have implications for the pregnancy, but also other members of the family as it may change their understanding of their genetic risk.
- Genetic test results may result in some forms of discrimination (insurance, employment or other) as they form part of your medical records and may be accessed by and/or disclosed to a third party who has obtained your necessary consent or when such access is allowed or required by law.
- There is a small chance of error in the results due to, but not limited to, limitations in technology, sample contamination, including maternal cell contamination, inconsistencies or differences in classification of variant(s), and/or lack of clinical knowledge and inaccuracies in family history knowledge.
- Genetic testing, in rare cases, may reveal non-paternity/maternity of a presumed parent in your family.
- Occasionally, the laboratory may require additional sample(s) from you and/or family members to clarify the result. In case of insufficient sample, an additional sample would also be required.
- People react differently to receiving genetic test results. You can request for additional support before proceeding with this genetic test and/or after receiving the results.

What can I expect after the test?

- Due to the complexity of the test, your results will only be made available to you by a genetic counsellor or suitably qualified and appointed healthcare professional.
- This genetic test result will be stored in your medical records, which will be accessible by the medical team(s) responsible for your care.
- The results are confidential and will only be released to other medical professionals involved in your care and/or other parties with your written consent or as otherwise allowed by law.
- Any remaining unused portion of the sample may be stored for validation, process development, and/or quality control studies, according to the laboratory's sample retention policies.
- Further testing and/or future re-analysis requested may incur additional charges and/or require an additional sample to be taken and/or may delay the time taken to get a final result.
- To assist with result interpretation, your de-identified genetic results and clinical information may be added to scientific databases (local and/or international).

- The result(s) may be useful to your family members to receive genetic testing. Please inform your doctor/genetic counsellor if you consent to sharing your genetic testing results with your family members.

What are my options?

Genetic testing is voluntary; you may choose not to proceed. You may also withdraw from the genetic testing at any point, before the test is completed. If consent is withdrawn, the sample will be discarded and no report will be issued. However, charges would apply once the test request has been received and processed.

Others (to be filled by Healthcare Professional) [if applicable]

Part I – Patient's Declaration

1. I, _____ (NRIC/Passport No. _____), have read this information sheet and confirm that I understand the nature, purpose, risks, limitations, and options with regard to **Constitutional Genetic Testing (Prenatal Genetics)** ("Test").
2. I acknowledge that the risks and limitation(s) listed are not intended to be exhaustive. I have had an opportunity to ask for more information about (i) the above-mentioned risks and limitations; (ii) the risks in general; and (iii) specific concern(s) of relevance to me.
3. I hereby consent to undergo the Test.
4. I understand and agree that the Test will be performed by the appropriate SingHealth institution (with the involvement of external providers, if necessary) and I will be admitted and/or registered as a patient of that SingHealth institution.
5. I understand that I have the option to choose whether to receive incidental findings.

   ☐ I wish to receive incidental findings.

   ☐ I **do not** wish to receive incidental findings.

   ☐ Not applicable
6. I understand that my result(s) may be useful for my family members for genetic counseling and testing.

   ☐ I consent to sharing my result(s) with my family members. They will be required to provide my name and NRIC/FIN/Passport number.

   ☐ I **do not** consent to sharing my result(s) with my family members.
7. In the event I am uncontactable, the test results may be made known to:

   Name: _____ NRIC (last 4 digits): _____
   Contact details: _____ Relationship: _____

   Name: _____ NRIC (last 4 digits): _____
   Contact details: _____ Relationship: _____

_____ _____
(Signature/**[*Left/Right]** Thumbprint of Patient) (Date of Signing)

_____ _____
(Name of Witness) (Designation of Witness)

_____ _____
(Signature of Witness) (Date of Signing)

*** Please delete accordingly**

Part II – Parent's / Legal Guardian's / Donee's / Deputy's Declaration (herein referred to as the "Authorised Person") (if applicable)

1. I, _____ (NRIC/Passport No. _____), the ***Parent / Legal Guardian / Donee / Deputy** of _____ (NRIC/Passport No. _____) ("Patient"), have read this information sheet and confirm that I understand the nature, purpose, risks, limitations and options with regard to **Constitutional Genetic Testing (General Genetics)** ("Test").
2. I acknowledge that the risks and limitation(s) of the Test listed are not intended to be exhaustive. I have had an opportunity to ask for more information about (i) the above-mentioned risks and limitations; (ii) the risks in general; and (iii) specific concern(s) of relevance to the Patient.
3. I hereby consent for the Patient to undergo the Test.
4. I understand and agree that the Test will be performed by the appropriate SingHealth institution (with the involvement of external providers if necessary) and the Patient will be admitted and/or registered as a patient of that SingHealth institution.
5. I understand that I have the option to choose whether to receive incidental findings.

   ☐ I wish to receive incidental findings.

   ☐ I **do not** wish to receive incidental findings.

   ☐ Not applicable
6. I understand that the Patient's result(s) may be useful for the Patient's family members for genetic counseling and testing.

   ☐ I consent to sharing Patient's result(s) with Patient's family members. They will be required to provide Patient's name and NRIC/FIN/Passport number.

   ☐ I **do not** consent to sharing Patient's result(s) with Patient's family members.
7. In the event I am uncontactable, the test results may be made known to:

   Name: _____ NRIC (last 4 digits): _____

   Contact details: _____ Relationship: _____

   Name: _____ NRIC (last 4 digits): _____

   Contact details: _____ Relationship: _____

_____ _____

(Signature/**[*Left/Right]** Thumbprint of Authorised Person) (Date of Signing)

_____ _____

(Name of Witness) (Designation of Witness)

_____ _____

(Signature of Witness) (Date of Signing)

*** Please delete accordingly**

Part III – Healthcare Professional's Declaration

I confirm that I have explained to the Patient, or the Authorised Person (if applicable), the Patient's medical condition as well as the nature, purpose, risks, limitations, and alternatives with regard to the Test and have addressed queries of the Patient, or the Authorised Person (if applicable).

_____ _____

(Signature, Full Name, and Professional Registration / *Employee No. of Healthcare Professional)

(Date of Signing)

**Only for those without professional registration number*

Part IV – Interpreter's Declaration (if applicable)

I, _____, confirm that I have interpreted to the Patient, or the Authorised Person (if applicable), the Healthcare Professional's explanation of the Patient's medical condition, nature, purpose, risks, limitations, and alternatives with regard to the Test and the Healthcare Professional's response to the Patient's, or the Authorised Person's (if applicable), queries in _____ (language / dialect).

_____ _____

(Signature of Interpreter)

(Date of Signing)