



KK Women's and  
Children's Hospital

SingHealth

Patient Type  Gynae  Obst  Neonate  
 Paed Med  Paed Surg  
Ward/Bed: \_\_\_\_\_ Clinic: \_\_\_\_\_ Class: \_\_\_\_\_

## CLINICAL GENETICS REQUEST (DNA TESTS)

Patient's name label

(For downtime use)

Name:

MRN:

Account number:

Date of birth:

Sex: M / F

<b>Clinical Diagnosis:</b>	Relevant History / Findings / Family history:	Accompanying sample (s):
<b>Please call lab for presymptomatic testing.</b>		Please specify relationship (spouse / sibling / parent / child etc)
Name & signature of requesting doctor Contact no. (if urgent)	Type of specimen: <input type="checkbox"/> Peripheral blood in EDTA (3ml unless otherwise specified) <input type="checkbox"/> Amniotic fluid (Gestation: _____ Amount: _____ ml) <input type="checkbox"/> Chorionic villi (Gestation: _____ Amount: _____ mg) <input type="checkbox"/> Fetal blood (1ml) <input type="checkbox"/> Others: _____	
Name of consultant i/c	Specimen taken Date: _____ Time: _____	
Date	<b>Prenatal specimen requirement for specific diseases:</b> 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

<p><b>DNA diagnostic tests for thalassaemia</b></p> <p>Hb: _____ MCV: _____ MCH: _____ Hb electrophoresis: HbA2 _____ HbF: _____</p> <p>DNA 108 <input type="checkbox"/> Thalassaemia DNA screen Hb electrophoresis, HbH inclusion bodies &amp; DNA analysis for 5 <math>\alpha</math> thalassaemia deletional mutations. Fresh EDTA blood (2xAdult 3mls; 2xPaeds 0.5ml) (Mon-Fri, 8am-6pm)</p> <p>DNA 101A <input type="checkbox"/> DNA analysis for <math>\alpha</math>-thalassaemia mutations DNA 101B <input type="checkbox"/> DNA analysis for <math>\beta</math>-thalassaemia mutations DNA 113A <input type="checkbox"/> DNA sequencing <math>\alpha</math>-globin genes DNA 113B <input type="checkbox"/> DNA sequencing <math>\beta</math>-globin genes DNA 102A <input type="checkbox"/> Prenatal test for <math>\alpha</math>-thalassaemia** DNA 102B <input type="checkbox"/> Prenatal test for <math>\beta</math>-thalassaemia**</p> <p><b>DNA diagnostic tests for following diseases</b></p> <p>DNA 103<sup>†</sup> <input type="checkbox"/> Huntington disease (HD) DNA 104 <input type="checkbox"/> Fragile X syndrome (FX) DNA 105<sup>†</sup> <input type="checkbox"/> Myotonic dystrophy (DM) DNA 106<sup>†</sup> <input type="checkbox"/> Spinocerebellar ataxia (SCA) screen DNA 106A<sup>†</sup> <input type="checkbox"/> Spinocerebellar ataxia (SCA) type _____ DNA 114 <input type="checkbox"/> Spinal muscular atrophy (SMA) DNA 109 <input type="checkbox"/> Y chromosome deletion DNA 110 <input type="checkbox"/> DNA methylation test for (Please circle one) Prader-Willi / Angelman syndrome DNA 111 <input type="checkbox"/> Achondroplasia (ACH) DNA 112<sup>†</sup> <input type="checkbox"/> Kennedy's Disease (KD or SBMA) DNA 115 <input type="checkbox"/> Craniosynostosis (hotspots in <i>FGFR1</i>, 2, 3 &amp; <i>TWIST</i>) DNA 116 <input type="checkbox"/> Specific craniosynostosis syndrome (circle one): Apert / Pfeiffer / Crouzon syndrome (Analyse hotspots in <i>FGFR2</i>) DNA 119 <input type="checkbox"/> Congenital adrenal hyperplasia (21OH deficiency) DNA 122 <input type="checkbox"/> Duchenne/Becker muscular dystrophy (DMD/BMD) DNA 126 <input type="checkbox"/> CCHS: <i>PHOX2B</i> testing</p> <p><b>DNA extraction</b></p> <p>DNA 004 <input type="checkbox"/> 3-5mls blood DNA 005 <input type="checkbox"/> Tissue/Cell culture/Amniotic fluid/CVS Please specify: _____</p>	<p><b>DNA diagnostic test for other diseases</b></p> <p>(Tests in this category are only carried out with prior arrangement)</p> <p>DNA 001 <input type="checkbox"/> Name of disease: _____ DNA 113 <input type="checkbox"/> Targeted sequencing for specific variant (please enclose report) Gene: _____ Variant: _____ RefSeq: _____ Chromosome coordinate (GRCh37 or GRCh38 please circle one): _____</p> <p><b>Other prenatal tests**</b></p> <p>(Tests in this category are only carried out with prior arrangement)</p> <p>DNA 002 <input type="checkbox"/> Prenatal test for (Please circle one) <input type="radio"/> Fragile X syndrome <input type="radio"/> Spinal muscular atrophy <input type="radio"/> Myotonic dystrophy (Type 1) <input type="radio"/> Others: _____</p> <p>DNA 117 <input type="checkbox"/> QF-PCR (rapid detection of chromosome aneuploidies of chromosomes 13, 18, 21, X &amp; Y)</p> <p><b>HLA Genotyping</b></p> <p>DNA 118 <input type="checkbox"/> HLA-B*1502 DNA 121 <input type="checkbox"/> HLA-B*5801</p>
	For lab use only
	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

<sup>†</sup> 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.

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

## CONSENT FORM

### CONSTITUTIONAL GENETIC TESTING (GENERAL 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 confirm a diagnosis, carrier status, or estimate your chance of developing certain health problems. The result of the genetic testing may be helpful for the doctors to better care for you.

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

**Name of the genetic test is** \_\_\_\_\_

#### What does genetic testing involve?

A blood and/or saliva sample and/or \_\_\_\_\_ (tissue sample) will be collected. The sample(s) will only be tested for the gene(s) / conditions listed above.

#### What are the possible results I could receive?

The results you could receive from genetic testing can 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 your health.

- **Genetic variant(s) identified**
  - A genetic change associated with your condition was identified
  - This confirms a genetic diagnosis, carrier status, or predisposition to disease, and may be helpful for medical management.
  - This result may have implications for family members.
- **Genetic variant(s) NOT identified**
  - No genetic change associated with your condition was identified
  - It does not exclude a genetic cause for your condition.
  - This result may reduce likelihood of a genetic / hereditary condition, but does not completely eliminate the possibility. 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 your condition, 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. You may currently have symptoms associated with the incidental findings, or you may or may not develop them in the future.
  - You have a choice whether you wish to receive such findings, if any.

### **What precautions must I take for genetic testing?**

It is important to inform your doctor / genetic counsellor if you have received a bone marrow transplant or recent blood transfusion as this may affect your result.

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

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

- Your test result can confirm a genetic diagnosis but cannot determine if, or when, the symptoms will manifest, nor can it provide information on the disease severity or recurrence.
- Your test result may have implications for your family members and can change the estimation 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, 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 your result.
- 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).
- Your 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 (General 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)