



# DNA ANALYSIS FOR THALASSAEMIA SYNDROMES & HAEMOGLOBINOPATHIES

For IMR/ HKL/ HSB used only  
Type of specimen  blood  DNA  others

Please  below, WHERE you wish to send the sample and TEST request:

<input type="checkbox"/> DNA analysis of the beta globin gene <input type="checkbox"/> Further testing for alpha/beta globin genes* <input type="checkbox"/> Confirmation for haemoglobinopathy <small>*Note: Only after common alpha/ beta globin gene mutations have been excluded by HKL/ HSB</small> <b>Molecular Genetics Laboratory</b> <b>Haematology Unit, Cancer Research Centre</b> <b>Institute for Medical Research</b> <b>National Institutes of Health</b> Jalan Setia Murni U13/52, 40170, Shah Alam, Selangor, Malaysia. Phone: 03-3362 8644 Email: mgm.imr@gmail.com Website: www.imr.gov.my	<input type="checkbox"/> DNA analysis of the alpha globin gene <input type="checkbox"/> DNA analysis of the beta globin gene <input type="checkbox"/> Confirmation for haemoglobinopathy (HbE, HbS and HbC only) <b>Molecular Hematology Laboratory</b> <b>Haematology Unit, Pathology Department</b> <b>Hospital Kuala Lumpur</b> 50586 Kuala Lumpur, Malaysia. Phone: 03-26155748/ 5746 Email: hematologihkl@moh.gov.my Website: www.hkl.gov.my	<input type="checkbox"/> DNA analysis of the alpha globin gene <b>Haematology Unit</b> <b>Pathology Department</b> <b>Hospital Sultanah Bahiyah</b> Km6, Jln Langgar, Bandar Alor Setar, 05460 Alor Setar, Kedah, Malaysia. Phone: 04-740 6250/ 6251 Fax: 04-740 6275 Email: patologi.hsb@moh.gov.my Website: www.hsbas.moh.gov.my
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<b>Patient Name:</b>	<b>Date of Birth:</b>	<b>Ethnicity:</b> <input type="checkbox"/> Malay <input type="checkbox"/> Chinese <input type="checkbox"/> Indian <input type="checkbox"/> Others;(specify) _____
	<b>Age:</b>	
<b>Patient ID/IC Number:</b>	<b>Gender:</b> <input type="checkbox"/> Male <input type="checkbox"/> Female <b>If female; Pregnant?</b> <input type="checkbox"/> YES, Weeks: ..... <input type="checkbox"/> No	<b>Hosp/ Ward/ Clinic:</b>
<b>Address of KK or hospital to send report:</b>	<b>Type of Specimen:</b>	<b>Date of Sampling:</b>
<b>Tel Number:</b>		<b>Date Sent:</b>

**CLINICAL SUMMARY/ FAMILY HISTORY/ FAMILY TREE.**

Parental consanguinity:  YES  NO

This information is crucial for baseline correlation of molecular results. Please  all that applies

**INDICATION OF TEST:**

- Diagnostic:  Antenatal  Others \_\_\_\_\_  
 Screening:  Form Four  Cascade screening  
 Others (specify): \_\_\_\_\_

**CLINICAL DIAGNOSIS:**

**CLINICAL STATUS:**

- |                                     |  |
|-------------------------------------|--|
| <input type="checkbox"/> New Case   | <input type="checkbox"/> Follow Up                   |
| <input type="checkbox"/> Trait      | <input type="checkbox"/> TDT (≥6 transfusions/ year) |
| <input type="checkbox"/> Intermedia | <input type="checkbox"/> NTDT                        |
| <input type="checkbox"/> Major      |  |
- Hb level at diagnosis : \_\_\_\_\_ g/dL  
 Age at diagnosis : \_\_\_\_\_  
 Hepatomegaly :  NO  YES \_\_\_\_\_ cm.  
 Splenomegaly :  NO  YES \_\_\_\_\_ cm.  
 Transfusion History  NIL yet  
 YES \_\_\_\_\_ No./Year  
 Last transfusion date \_\_\_\_\_

**THIS PART EXPLAINS SPECIMEN & TEST REQUIREMENTS/ CHECKLIST:**

**SPECIMEN REQUIREMENTS:**

Peripheral blood in EDTA tube (must arrive at lab within 2 week)  
 Adults: ~2.5 mL  Paeds: ~0.5mL

**TEST REQUIREMENTS:**

- A copy of recent FBC (<3 months) result of this patient  
 A copy of Hb Analysis result of this patient  
 All paediatrics (≤ 12 y.o.) sample to be referred to IMR, must be accompanied by both parents' FBC and Hb analysis results

**ADDITIONAL REQUIREMENTS FOR CASCADE**

**SCREENING:**  Index case

- i) Name: \_\_\_\_\_  
 ii) IC/Lab No: \_\_\_\_\_  
 iii) Diagnosis: \_\_\_\_\_  
 iv) Relationship to index case: \_\_\_\_\_

A copy of DNA analysis for thalassaemia syndromes/ haemoglobinopathy result of INDEX case

If Hb analysis report of this patient is pending:

- i) Hospital performing the test: \_\_\_\_\_  
 ii) Date of sample sent: \_\_\_\_\_

Official stamp of Requesting Doctor  
(Name, Signature & Date)

## INFORMED CONSENT FOR DNA TESTS

Test information: **DNA ANALYSIS OF THALASSAEMIA SYNDROMES & HAEMOGLOBINOPATHIES**

Patient Name:  
.....

Patient ID:  
.....

I understand the following:

This test is specific for \* **THALASSEMIA SYNDROMES & HAEMOGLOBINOPATHIES**

- A **POSITIVE** result is an indication that I may be predisposed to or have the specific disease, or condition. Further testing may be needed to confirm the diagnosis.
  - There is a chance that I will have this genetic condition but that the genetic test results will be **NEGATIVE**. Due to limitations in the technology and incomplete knowledge of genes, some changes in DNA or protein products that cause disease, may not be detected by the test.
  - There may be a possibility that the laboratory findings will be **UNINTERPRETABLE** or of unknown significance. In rare circumstances, findings may be suggestive of a condition different than the diagnosis that was originally considered.
1. 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.
  2. Erroneous results and incorrect interpretation may occur because of **rare variation in the DNA of the individual, rare technical error, disincorporation of DNA bases by the enzyme used to perform the test, sample misidentification, sample contamination, primer site mutations and general laboratory error.**
  3. Accurate interpretation of the DNA test result depends on **correct information** about the **clinical diagnosis** and about the **biological relationships** within the family.
  4. DNA testing may reveal **non-paternity**, meaning that the stated father is not the biological father.
  5. The tests offered are considered to be **the best available at this time**. If technology improves and more mutations (gene defects) are detectable in future, I authorize the Laboratory to re-analyse, at the Laboratory's option, any remaining DNA for the same disease without being informed. If the sample is insufficient, my doctor may ask me for a fresh sample. There may be additional fees for such tests.
  6. In order to help me understand the test results, the results will be reported to me only through a doctor or genetic counsellor.
  7. This report shall be used **ONLY** for clinical interpretation.
  8. This report shall **NOT** be used for any forensic purposes or is **NOT VALID** for forensic interpretation.
  9. This report shall **NOT** be used in any courts of law or in legal matters and is **NOT VALID** for legal interpretation.

For prenatal testing, the following also apply:

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

**WRITTEN CONSENT:**

1. A biologic specimen (blood, tissue, amniotic fluid or chorionic villi) will be collected for DNA tests for the above condition.
2. 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. I understand that any biologic specimens obtained for the purpose of this genetic testing become the exclusive property of HKL/ IMR's laboratories. After the specific test(s) requested have been completed, the laboratory may dispose, retain, or use the de-identified specimen(s) for test validation or education; i.e publication into journals. I understand that my identity will be protected.
3. DNA results are strictly **CONFIDENTIAL** and will not be released to anyone including my relatives/ other than my doctors without my consent.
4. 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).

To be completed by the:	
PATIENT/ PARENTS/ LEGAL GUARDIAN	DOCTOR/ COUNSELLOR
I have read and received a copy of this consent form. I understand the information provided in this document and I have had the opportunity to ask questions about testing, the procedure and the associated risks, benefits and limitations. I agree to have genetic testing and accept the risks and limitations.	I have fully explained the nature of the requested test(s) to the patient/ parent/ legal guardian
SIGNATURE : NAME & IC NO.: DATE:	SIGNATURE & OFFICIAL STAMP: NAME : DATE :