DNA ANALYSIS FOR THALASSEMIA SYNDROMES

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Specimen Requirements:
- *Adults: ~2.5 mL, Peads: ~0.5mL peripheral blood in EDTA tube (lavender/purple/ green cap)*
- *All paediatrics samples must be accompanied with parents' samples.*

FOR LAB USE ONLY (MGM/ MT)

PATIENT INFORMATION *Maklumat pesakit*:

1. Patient Name *Nama Pesakit*	:	2. Date of Birth: *Tarikh Lahir*	3. Age : *Umur*
4. Patient ID/ IC No. *No Kad Pengenalan*	:	5. Ethnicity *Etnik* : ☐ Malay ☐ Chinese ☐ Indian ☐ Others; Please specify: _____	6. Gender *Jantina* ☐ Male ☐ Female 7. If female; Pregnant? ☐ YES, Weeks:…… ☐ No
8. Address to send report: *Alamat untuk penghantaran laporan*		9. Hosp/ Ward/ Clinic: *Hospital/ Wad/ Klinik* 10. Hosp. Lab No.: *No Makmal* 11. Type of Specimen: *Jenis spesimen*	
Tel/ Fax No.		12. Date of Sampling: *Tarikh pengambilan spesimen*	Date Sent: *Tarikh Hantar*

FAMILY HISTORY **(INCLUDE FAMILY PEDIGREE/ TREE)** *Sejarah keluarga (Sertakan gambarajah pokok keluarga)*

Parental consanguinity: ☐ **YES** ☐ **NO**

Clinical Diagnosis: _____

Clinical Status:
☐ **NEW CASE** ☐ **FOLLOW UP**
☐ Trait ☐ TDT (≥6 transfusions/ year)
☐ Intermedia ☐ NTDT
☐ Major

Has anyone in this patient's family or spouse ever had DNA testing for thalassemia/ hemoglobinopathy?
☐ **YES** ☐ **NO**

If **YES**, what was the result(s): _____

If test was done in IMR/ HKL previously, please state IMR/ HKL Hematology Lab No.: _____

CLINICAL FEATURES:

Age at first diagnosis	: _____	Months/ Years		
Hb Level at diagnosis	: _____	g/dL		
Hepatomegaly	: ☐ **NO**	☐ **YES**	_____	cm.
Splenomegaly	: ☐ **NO**	☐ **YES**	_____	cm.
Transfusion History	: ☐ **NIL** yet	☐ **YES**	_____	No./ Year

TYPE OF TEST REQUEST:
☐ DNA analysis of the beta globin gene
☐ DNA analysis of the alpha globin gene
☐ Confirmatory test for thalassemia/ hemoglobinopathy
☐ DNA analysis alpha for further testing

Is this patient the **INDEX** case? ☐ **YES** ☐ **NO**
Adakah pesakit ini merupakan kes INDEKS

If **NO**, Please state
NAME & **IC No.** of the **INDEX** case:
Jika TIDAK, Nyatakan NAMA, No Kad Pengenalan kes INDEKS

Relationship to **INDEX** case:
Hubungan dengan kes INDEKS

IMPORTANT CHECKLIST; Please include with this form:
☐ A copy of recent FBC result of this patient
☐ A copy of Hb Analysis result of this patient
☐ A copy of affected relative's genetic result, if not done in IMR.
☐ For requests to exclude 'normal/ boderline HbA2 beta thal trait', please include results of iron studies and DNA analysis of alpha globin gene.
☐ Hb analysis report Is pending; Hosp: _____;
Date sample sent: _____

Official stamp of Requesting Doctor:

Name, Signature & Date

INFORMED CONSENT FOR DNA TESTS

TEST INFORMATIONS:

Name of Disease/ Test : _____

I understand the following:

1. This test is specific for _____.
   - 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.
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, rare technical error, misincorporation of DNA bases by the enzyme used to perform the test, sample misidentification, sample contamination, primer site mutations and general laboratory 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. 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.
7. In order to help me understand the test results, the results will be reported to me only through a doctor or genetic counsellor.
8. This report shall be used **ONLY** for clinical interpretation.
9. This report shall **NOT** be used for any forensic purposes or is **NOT VALID** for forensic interpretation.
10. 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.
2. Besides rare DNA variation and the technical error, erroneous results may also arise from maternal contamination of the fetal sample.

INFORMED 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 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. SIGNATURE : NAME & IC NO.: DATE:	I have fully explained the nature of the requested test(s) to the patient/ parent/ legal guardian SIGNATURE & OFFICIAL STAMP: NAME : DATE :