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REQUEST FORM FOR DNA ANALYSIS OF THALASSAEMIA AND HAEMOGLOBINOPATHIES

Please (√) below:

- ☐ DNA analysis of the alpha globin gene
- ☐ DNA analysis of the beta globin gene
- ☐ Confirmation for haemoglobinopathy

Patient Name	**Date of Birth:** **Age:**	**Ethnicity** □ **Malay** □ **Chinese** □ **Indian** □ **Others;**(specify)
Patient IC Number	**Gender** □ **Male** □ **Female**	**Type of Specimen:**
Patient MRN		**Date of Sampling:**
Ward/ Clinic:	**If female; Pregnant?** □ **YES, Weeks: ……** □ **No**	**Date Sent:**

CLINICAL SUMMARY/ FAMILY HISTORY/ FAMILY TREE.

Parental consanguinity: □ **YES** □ **NO**

THIS PART EXPLAINS SPECIMEN & TEST REQUIREMENTS/ CHECKLIST:

SPECIMEN REQUIREMENTS:
Fresh peripheral blood in EDTA tube (minimum 2 mL)

TEST REQUIREMENTS:
- ☐ A copy of recent iron study result (within 3 months).
- ☐ A copy of recent FBP result (within 3 months).
- ☐ A copy of Hb Analysis result of this patient that suggest for DNA analysis.

ADDITIONAL REQUIREMENTS FOR CASCADE SCREENING:
□ Index case
i) Name: _____
ii) IC/MRN No _____
iii) Diagnosis: _____
iv) Relationship to index case _____

□ A copy of DNA analysis for thalassemia syndrome/ haemoglobinopathy result of the INDEX case

This part is crucial for baseline correlation of the molecular results. Please ☐ all that applies

INDICATION OF TEST:
□ Diagnostic □ Antenatal
□ Form Four screening □ Cascade screening
□ Others (specify): _____

CLINICAL DIAGNOSIS:

CLINICAL STATUS:
□ **New Case**
- ☐ Trait
- ☐ Intermedia
- ☐ Major

□ **Follow Up**
- ☐ TDT (≥6 transfusions/ year)
- ☐ NTDT

Hb level at diagnosis : _____ g/dL

Hepatomegaly : □ **NO** □ **YES** _____ cm.

Splenomegaly : □ **NO** □ **YES** _____ cm.

Transfusion History ☐ **NIL** yet
☐ **YES** _____No./Year

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

REQUEST FORM FOR DNA ANALYSIS OF THALASSAEMIA AND HAEMOGLOBINOPATHIES

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 * ***THALASSAEMIA 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 thediagnosis.
- 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 thetest.
- There may be a possibility that the laboratory findings will be **UNINTERPRETABLE** or of unknown significance. In rarecircumstances, 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 membersmay 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 beadditional 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 fetalsample.

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 HSAAS 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. Iagree 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) tothe patient/ parent/ legal guardian SIGNATURE & OFFICIAL STAMP: NAME: DATE: