

## Prenatal Diagnostic Laboratory, Tsan Yuk Hospital

Address: 30, Hospital Road, Hong Kong Tel: (852) 2589-2218 Fax: (852) 2517-2373

### Thalassaemia Genetic Testing

#### Instructions (Private)

#### 1. Instructions to doctors (private) on sending samples

- 1.1 Complete the “**Thalassaemia Screen Request**” (TYH-REQ-THAL-SCREEN) form.
- 1.2 Fax the completed “**Thalassaemia Screen Request**” form together with copy of laboratory reports of haematological study (CBP and Hb pattern) of the couple to our laboratory (Fax no.:2517-2373).
  - Correlation with CBP and Hb pattern results is necessary for correct interpretation of genetic test result for thalassaemia.
  - If no prior haematological study is available, please contact our laboratory for further arrangement.
- 1.3 Complete a 2-page request form “**Request form for laboratory studies - Prenatal Diagnosis / Reproductive Medicine Investigation**” (TYH-REQ-PRE-RMI) for **Prenatal Diagnosis / Reproductive Medicine Investigation**, which can be provided from the laboratory upon request, with the following information:
  - Patient details (patient’s demographics, HKID or document ID, PDC/clinic/hospital no.)
  - Referring doctor details (name of referring doctor, contact details and address for report)
  - Specimen details (nature of specimen, date and time of sampling)
  - Test(s) requested
  - Clinical details (information needed for examination performance and results interpretation may include patient’s ancestry, family history, consanguinity)

#### IMPORTANT NOTES:

- a) Use a separate request form for each prenatal and parental samples
  - b) Send specimen in appropriate containers labelled with at least two patient’s identifiers
  - c) If the sample cannot be sent to the laboratory on the day of sampling, please keep it at the door of refrigerator (4°C) and arrange the sample to reach the laboratory within 48 hours (*excluding specimen collected after office hour on Fridays, Saturdays, Sundays and public holidays*).
- 1.4 Obtain patient consent using “**Consent form for Genetic and Genomic Investigations**” (TYH-REC-CONF-GG) for each patient/subject being referred for cytogenetic/genetic testing in PDL, TYH.
    - Complete one consent form for one subject
    - The pregnant woman shall sign the form on behalf of the fetus
    - In case couple testing is required, woman and her partner shall independently sign one form.
    - Send the completed patient consent form with request form for each patient sample.
  - 1.5 We shall note the haematological findings and reply with an appointment for sending
    - (i) blood sample from woman or her partner for exclusion of alpha thal
    - or (ii) blood samples from beta thal couple for mutation workup
    - or (iii) blood samples from alpha thal couple together with amniotic fluid or chorionic villi
    - or (iv) amniotic fluid or chorionic villi for those with previous prenatal diagnosis
  - 1.6 Attach laboratory reports on haematological study (CBP and Hb pattern) of the couple when sending prenatal samples for thalassaemia genetic testing.
  - 1.7 For  $\beta$  thal couples, please send blood samples for mutation workup before prenatal diagnosis. We shall contact you if prenatal testing is feasible, within a week.
  - 1.8 Sample delivery
    - Courier service: Call Thunder Express Service Co. Ltd. at 2397-8781 or 2380-9122, Ms. Monica Leung on or before 11:00 am to arrange specimen pick up at 12:00 noon. Courier service is only available from Monday to Friday (excluding public holidays).  
*(Note: Please contact the laboratory to arrange specimen delivery to PDL, TYH beyond the aforementioned specimen pick up time from Monday to Friday. A special courier service fee applies.)*
    - Self drop-in: Your own personnel can bring the collected specimen to PDL, TYH within:  
*Specimen Reception hours: Monday to Friday: 8:45am - 4:30pm*  
*(excluding Saturdays, Sundays and public holidays)*

### 1.9 Tests and specimen collection

Laboratory test	Nature of specimen	Container (provided upon request)	Specimen amount/volume	Turn-around Time*
Genetic testing for thal	Amniotic fluid	Falcon 2096 tube	10-15 mL	7 working days (excluding public holidays and Sundays)
	Chorionic villi	Falcon 2096 tube with transport medium	5 mg villi	
Genetic testing for thal and chromosomes study	Amniotic fluid	Falcon 2096 tube	30 mL	
	Chorionic villi	Falcon 2096 tube with transport medium	15 mg villi	
Genetic testing for thal on couple	Peripheral blood	EDTA	3 mL	

\*Turn-around time may vary and be subjected to the quality and the quantity of the received specimen.

### 2. Rejection of sample

A specimen may be rejected when the following condition is observed:

- unlabelled or incorrectly labelled
- specimen container leaks
- not suitable for analysis (e.g. using incorrect container, frozen specimen, peripheral blood from subject having recent blood transfusion or taking immunosuppressive drugs, etc.)
- contaminated with maternal blood in the fetal specimen
- specimen cannot reach the laboratory within 48 hours (*excluding specimen collected after office hour on Fridays, Saturdays, Sundays and public holidays*)

In such events, you will be contacted for further actions.

### 3. Reporting

All reports will be faxed and sent by post to your office.

### 4. Payment method

- Monthly invoice will be issued from Queen Mary Hospital Finance Department
- Alternative payment method requires prior arrangement with the laboratory.

### 5. Address and contact information

*Address:* Prenatal Diagnostic Laboratory  
Room 2-10, Tsan Yuk Hospital, 30 Hospital Road  
Sai Ying Pun, Hong Kong

*Tel:* 2589-2208, 2589-2288  
*Fax:* (lab) 2857-5407, (office) 2517-2373

*Website:* <https://obsgyn.med.hku.hk/en/Services/Obstetrics/Maternal-Fetal-Medicine/Prenatal-Diagnosis>

*Laboratory opening hours:*  
Monday to Friday: 8:45am - 5:30pm  
(closed on Saturdays, Sundays and public holidays)

*Contacts:* Scientific Officer

Tel: 2589-2327 / 2589-2328

Thalassaemia genetic testing

Duty Officer / Laboratory Director

Tel: 2589-2288 / 2589-2327

Other enquiry or complaint

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