

Prenatal Diagnostic Laboratory, Tsan Yuk Hospital

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

Thalassaemia Genetic Testing

Instructions (HA)

1. Instructions to doctors (HA) 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), 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 β 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
 - We prefer to receive samples on Mondays or Tuesdays. If needed, we shall receive samples on other days of the week. Please call us to confirm.
 - Samples are sent to PDL via hospital portering service. Please arrange with portering service team of the referring hospital.

*Specimen Reception hours: Monday to Friday: 9:00am - 4:30pm
(excluding Saturdays, Sundays and public holidays)*

