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 “Request Form for Laboratory Studies” (TYH-REQ-LAB-HA) with the following information and use the form for sending samples, one form for one sample: blood, amniotic fluid or chorionic villi:
   - Patient details (patient’s demographics, HKID or document ID, PDC/clinic/hospital no.)
   - Name and ID no. of wife/mother on the request form for sample from husband/father or affected child
   - 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).

   Important note:
   a) Obtain patient consent for sample collection and/or storage, with a separate consent for genetic testing if applicable.
   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.

1.4 We shall note the haematological findings and reply with an appointment for sending
   (i) blood sample from wife or husband 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

1.5 Attach laboratory reports on haematological study (CBP and Hb pattern) of the couple when sending prenatal samples for thalassaemia genetic testing.

1.6 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.

1.7 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
   Saturday: 9:00am - 11:30am
   (excluding public holidays and Sundays)
1.8 Tests and specimen collection

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

*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 foetal specimen if QF-PCR or molecular genetics study is requested on the foetal specimen.
- specimen cannot reach the laboratory within 48 hours.

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

3. Reporting

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

4. 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: http://www.obsgyn.hku.hk/prenatal_diagnosis

Laboratory opening hours:
Monday to Friday: 9:00am - 5:30pm
Saturday: 9:00am - 12:30pm
(closed on public holidays & Sundays)

Contacts:
Mr. Patrick Au
(Scientific Officer)
Tel: 2589-2315
Thalassaemia genetic testing

Duty Officer / Laboratory Director
Tel: 2589-2288 /
2589-2327
Fax: 25172373
Other enquiry or complaint

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