Chromosomes and molecular genetic analysis for prenatal diagnosis

Instructions (HA)

(Chromosomal microarray, karyotype, QF-PCR, molecular genetics analysis)

1. Instructions to doctors (HA) on sending samples

1.1 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) (Please indicate on the request form or consent form to show the relationship of the couple e.g. Husband of or partner of pregnant woman’s name)

IMPORTANT NOTES:

a) Use a separate request form for each prenatal and parental samples
b) For requesting molecular genetic tests on fetal specimen or suspecting maternal cell contamination in the fetal specimen, maternal peripheral blood sample shall be provided for maternal cell contamination test. (For requesting thalassaemia genetic testing, please refer to “Instructions to doctors on Thal” (TYH-INF-INST-THAL) for Thalassaemia Genetic Testing.)
c) Send specimen in appropriate containers labelled with at least two patient’s identifiers
d) 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.

1.2 Obtain patient consent using a generic “Consent Form for Genetic Testing” (TYH-REC-CONSENT) for each patient/subject being referred for cytogenetic/genetic testing in PDL, TYH. This includes choice of patient sample storage for further investigation or research. It explains the limitation of genetic result interpretation and the possibility of unveiling incidental findings.

- 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, pregnant woman and her partner shall independently sign one form.
- Send the completed patient consent form with request form for each patient sample.

1.3 Sample delivery

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)

1.4 Tests and specimen collection specification (see next page, Table 1)

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, hemolysed blood, 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.
Table 1. Tests and specimen collection specifications

<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>Chromosomal microarray analysis, CMA (+/- Karyotype)</td>
<td>Amniotic fluid</td>
<td>Falcon 2096 tube</td>
<td>30 mL in 3 tubes</td>
<td>7 working days</td>
</tr>
<tr>
<td></td>
<td>Chorionic villi</td>
<td>Falcon 2096 tube with transport medium</td>
<td>8-10 mg</td>
<td>7 working days</td>
</tr>
<tr>
<td></td>
<td>Placental tissue</td>
<td>Sterilin universal bottle with transport medium</td>
<td>At least $0.5 \times 0.5 \times 0.5$ cm$^3$</td>
<td>14 working days</td>
</tr>
<tr>
<td></td>
<td>Skin biopsy</td>
<td>Sterilin universal bottle with transport medium</td>
<td>~$0.5 \times 0.5$ cm</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Products of gestation</td>
<td>Sterilin universal bottle with transport medium</td>
<td>At least $0.5 \times 0.5 \times 0.5$ cm$^3$</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Cord blood</td>
<td>Heparin (if request for karyotype; otherwise EDTA)</td>
<td>0.2 mL</td>
<td>7 working days</td>
</tr>
<tr>
<td></td>
<td>Foetal blood</td>
<td></td>
<td>0.2 mL</td>
<td>7 working days</td>
</tr>
<tr>
<td></td>
<td>Parental peripheral blood</td>
<td>EDTA</td>
<td>3 mL from each parent</td>
<td>7-14 working days (if inheritance study is performed)</td>
</tr>
<tr>
<td></td>
<td>Amniotic fluid</td>
<td>Falcon 2096 tube</td>
<td>20 mL in 3 tubes</td>
<td>15 calendar days</td>
</tr>
<tr>
<td></td>
<td>Chorionic villi</td>
<td>Falcon 2096 tube with transport medium</td>
<td>5-10 mg</td>
<td>15 calendar days</td>
</tr>
<tr>
<td></td>
<td>Placental tissue</td>
<td>Sterilin universal bottle with transport medium</td>
<td>At least $0.5 \times 0.5 \times 0.5$ cm$^3$</td>
<td>28 calendar days</td>
</tr>
<tr>
<td></td>
<td>Skin biopsy</td>
<td>Sterilin universal bottle with transport medium</td>
<td>~$0.5 \times 0.5$ cm</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Products of gestation</td>
<td>Sterilin universal bottle with transport medium</td>
<td>At least $0.5 \times 0.5 \times 0.5$ cm$^3$</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Cord blood</td>
<td>Heparin</td>
<td>2 mL</td>
<td>18 calendar days</td>
</tr>
<tr>
<td></td>
<td>Foetal blood</td>
<td></td>
<td>7 calendar days</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Parental peripheral blood</td>
<td>Heparin</td>
<td>2 mL from each parent</td>
<td>18 calendar days</td>
</tr>
<tr>
<td>Rapid Aneuploidy Detection (QF-PCR) /22q11.2 microdeletion</td>
<td>Amniotic fluid</td>
<td>Falcon 2096 tube</td>
<td>2-4 mL</td>
<td>QF-PCR / 22q11.2 microdeletion: 2 working days</td>
</tr>
<tr>
<td>Other molecular genetic tests (e.g. MSPCR, Fragile X, FGFR3 mutation detection, DNA sequencing, etc.)</td>
<td>Chorionic villi</td>
<td>Falcon 2096 tube with transport medium</td>
<td>0.2-0.4 mg (dissected)</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Placental tissue</td>
<td>Sterilin universal bottle with transport medium</td>
<td>At least $0.5 \times 0.5 \times 0.5$ cm$^3$</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Skin biopsy</td>
<td>Sterilin universal bottle with transport medium</td>
<td>~$0.5 \times 0.5$ cm</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Products of gestation</td>
<td>Sterilin universal bottle with transport medium</td>
<td>At least $0.5 \times 0.5 \times 0.5$ cm$^3$</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Cord blood</td>
<td>EDTA</td>
<td>0.2 mL</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Foetal blood</td>
<td></td>
<td>0.2 mL</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Parental peripheral blood</td>
<td>EDTA</td>
<td>3 mL from each parent</td>
<td></td>
</tr>
</tbody>
</table>

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

All reports will be faxed and sent to your office by messengers. Cytogenetics, QF-PCR, 22q11.2 microdeletion reports can be accessed via Electronic Patient Record (ePR).

(Note: After delivery of the baby or miscarriage, please complete the section of “Reply form - Pregnancy outcome (to PDL, TYH)” and return a copy of the form (by fax or by post) for auditing purpose.)

4. Payment method

Self-financed tests

- **QMH**
  
  Instruct patient (with the completed HKU Charging Form) to pay at QMH/TYH Shroff. Send copy of payment receipt, specimen and request form to PDL, TYH via hospital portering service.

- **Other HA hospitals**
  
  Instruct patient to write a crossed cheque in Hong Kong Dollars payable to "Hospital Authority - Queen Mary Hospital”, or “醫院管理局 - 玛麗醫院”.

  Send the crossed cheque along with the completed HKU charging form, the samples and request form to PDL via hospital portering service, where the charging form can be obtained upon request.

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:** 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. W.K. Tam**
  (Senior Med. Tech.)
  Tel: 2589-2288
- **Dr Kelvin Chan**
  (Scientific Officer i/c)
  Tel: 2589-2327 / 2589-2316
- **Mr Patrick Au**
  (Scientific Officer)
  Tel: 2589-2328 / 2589-2316
- **Duty Officer / Laboratory Director**
  Tel: 2589-2288 / 2589-2327

**Chromosomes study (Karyotype)**

**Molecular genetic testing**
(e.g. CMA, QF-PCR, MSPCR, Fragile X, FGFR3, DNA sequencing, UPD, FISH, etc.)

**Other enquiry or complaint**

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