Instructions to doctors (HA) on sending samples

1.1 Complete one-page request form “Request form for Postnatal Studies” (TYH-REQ-POST), which can be provided from the laboratory upon request, with the following information:

- Patient details (patient’s demographics, HKID or document ID, clinic/hospital no., relationship to proband with proband’s name and ID)
- 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 NOTES:**

a) Use a separate request form for each subject
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.

1.2 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
- Either parent/guardian shall sign the form on behalf of his/her child
- Each parent shall independently sign one form if parental testing is required.
- Send the completed patient consent form with request form

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.)
- 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>Peripheral blood (Proband)</td>
<td>EDTA (if request for karyotype and FISH analysis, use Heparin)</td>
<td>2 mL</td>
<td>14 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>(for inheritance study in CMA)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Chromosomes study (Karyotype only)</td>
<td>Peripheral blood (Proband)</td>
<td>Heparin</td>
<td>2 mL</td>
<td>18 calendar days</td>
</tr>
<tr>
<td></td>
<td>Parental peripheral blood</td>
<td></td>
<td>2 mL from each parent</td>
<td>18 calendar days</td>
</tr>
<tr>
<td></td>
<td>(for inheritance study)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Rapid Aneuploidy Detection (QF-PCR) / 22q11.2 microdeletion</td>
<td>Peripheral blood (Proband)</td>
<td>EDTA (if request for karyotype and FISH analysis, use Heparin)</td>
<td>2 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>Parental peripheral blood</td>
<td></td>
<td>3 mL from each parent</td>
<td>Others: 7-28 working days (please contact lab.)</td>
</tr>
<tr>
<td></td>
<td>(for inheritance study)</td>
<td></td>
<td></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).

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 - 瑪麗醫院".
  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](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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