Prenatal Diagnostic Laboratory, Tsan Yuk Hospital

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

Chromosomes and molecular genetic analysis for postnatal study

Instructions (HA)

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

1. Instructions to doctors (HA) on sending samples

- 1.1 Complete one-page request form "**Request form for Postnatal Studies**" (TYH-REQ-POST), which <u>can</u> 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 (*excluding specimen collected after office hour on Fridays, Saturdays, Sundays and public holidays*).
- 1.2 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
 - 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: 8:45am - 4:30pm

(excluding Saturdays, Sundays and public holidays)

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 (excluding specimen collected after office hour on Fridays, Saturdays, Sundays and public holidays)

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

Table 1. Tests and specimen collection specifications

Laboratory test	Nature of specimen	Container (provided upon request)	Specimen amount/volume	Turn-around time*
Chromosomal microarray analysis, CMA (+/- Karyotype)	Peripheral blood (Proband)	EDTA (if request for karyotype and FISH analysis, use Heparin)	2 mL	14 working days
	Parental peripheral blood (for inheritance study in CMA)	EDTA (not for karyotype)	3 mL from each parent	7-14 working days (if inheritance study is performed in CMA)
Chromosomes study (Karyotype only)	Peripheral blood (Proband)	Heparin	2 mL	18 calendar days
	Parental peripheral blood (for inheritance study)		2 mL from each parent	18 calendar days
Rapid Aneuploidy Detection (QF-PCR) / 22q11.2 microdeletion	Peripheral blood (Proband)	EDTA (if request for karyotype and FISH analysis, use Heparin)	2 mL	QF-PCR / 22q11.2 microdeletion: 2 working days
Other molecular genetic tests (e.g. MSPCR, Fragile X, FGFR3 mutation detection, DNA sequencing, etc.)	Parental peripheral blood (for inheritance study)		3 mL from each parent	Others: 7-28 working days (please contact lab.)

^{*}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. Chromosomal microarray, Cytogenetics, QF-PCR, 22q11.2 microdeletion reports can be accessed via Electronic Patient Record (ePR).

4. Payment method

Self-financed tests

➤ <u>QMH</u>

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 Laboratory opening hours:

Room 2-10, Tsan Yuk Hospital, 30 Hospital Road Monday to Friday: 8:45am - 5:30pm

Sai Ying Pun, Hong Kong (closed on Saturdays, Sundays and public holidays)

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

Contacts: Senior Med. Tech. Tel: 2589-2288 Chromosomes study (Karyotype)

Scientific Officer Tel: 2589-2327 / 2589-2328 Molecular genetic testing

(e.g. CMA, QF-PCR, MSPCR, Fragile X, FGFR3,

DNA sequencing, UPD, FISH, etc.)

Duty Officer / Tel: 2589-2288 / 2589-2327 Other enquiry or complaint

Laboratory Director