

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 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 and 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 (*excluding specimen collected after office hour on Fridays, Saturdays, Sundays and public holidays*).
- e) For sample requesting karyotype: If cultured cells failed to settle in culture, PDL will notify referring doctor and proceed to PCR.

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
- 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: 8:45 am - 4:30 pm
(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.)
- 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 (*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)	Amniotic fluid	Falcon 2096 tube	30 mL in 3 tubes	7 working days
	Chorionic villi	Falcon 2096 tube with transport medium	8-10 mg	7 working days
	Placental tissue	Sterilin universal bottle with transport medium	At least $0.5 \times 0.5 \times 0.5\text{cm}^3$	14 working days
	Skin biopsy		$\sim 0.5 \times 0.5$ cm	
	Products of gestation		At least $0.5 \times 0.5 \times 0.5\text{cm}^3$	
	Cord blood	Heparin (if request for karyotype; otherwise EDTA)	0.2 mL	7 working days
	Foetal blood		0.2 mL	7 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)	Amniotic fluid	Falcon 2096 tube	20 mL in 3 tubes	15 calendar days
	Chorionic villi	Falcon 2096 tube with transport medium	5-10 mg	15 calendar days
	Placental tissue	Sterilin universal bottle with transport medium	At least $0.5 \times 0.5 \times 0.5\text{cm}^3$	28 calendar days
	Skin biopsy		$\sim 0.5 \times 0.5$ cm	
	Products of gestation		At least $0.5 \times 0.5 \times 0.5\text{cm}^3$	
	Cord blood	Heparin	2 mL	18 calendar days
	Foetal blood			7 calendar days
	Parental peripheral blood		2 mL from each parent	18 calendar days
Rapid Aneuploidy Detection (QF-PCR) /22q11.2 microdeletion Other molecular genetic tests (e.g. MSPCR, Fragile X, FGFR3 mutation detection, DNA sequencing, etc.)	Amniotic fluid	Falcon 2096 tube	2-4 mL	QF-PCR / 22q11.2 microdeletion: 2 working days
	Chorionic villi	Falcon 2096 tube with transport medium	0.2-0.4 mg (dissected)	
	Placental tissue	Sterilin universal bottle with transport medium	At least $0.5 \times 0.5 \times 0.5\text{cm}^3$	
	Skin biopsy		$\sim 0.5 \times 0.5$ cm	
	Products of gestation		At least $0.5 \times 0.5 \times 0.5\text{cm}^3$	
	Cord blood	EDTA	0.2 mL	Others: 7-28 working days (please contact lab.)
	Foetal blood		0.2 mL	
	Parental peripheral blood		3 mL from each parent	

*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).

(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 request form to PDL, TYH via hospital portering 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: <https://obsgyn.med.hku.hk/en/Services/Obstetrics/Maternal-Fetal-Medicine/Prenatal-Diagnosis>

Laboratory opening hours:
Monday to Friday: 8:45 am - 5:30 pm
(closed on Saturdays, Sundays and public holidays)

<i>Contacts:</i> 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 / Laboratory Director	Tel: 2589-2288 / 2589-2327	Other enquiry or complaint

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