

Prenatal Diagnostic Laboratory, Tsan Yuk Hospital

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

Down syndrome screening tests – Instructions (HA)

1. Instructions to doctors (HA) prior to making test request

1.1 Pre-test explanation

Please explain to couples that the biochemical test is a **screening test**. It is not a diagnostic test. **A screen negative result does not exclude the possibility of Down syndrome because screening does not detect all affected pregnancies.**

Please refer to the following table for the detection rate (with screen positive rate of 5%).

Comparison of the detection rates (with screen positive rate of 5%) among different Down syndrome screening tests

Down syndrome screening tests	Gestational age (weeks)	Tests	Detection rate with screen positive rate of 5% (%)
Nuchal Screening	11 - 13 ⁺⁶	NT measurement	69 ^(a) – 79 ^(b)
1 st Trimester Combined Screening Test	11 - 13 ⁺⁶	NT measurement + PAPP-A + free β -hCG	Singleton pregnancies 88 ^(b)
			Twin pregnancies ^(b) Monochorionic 90 Dichorionic 79 All 81
2 nd Trimester Quadruple Screening Test	16 - 19 ⁺⁶	AFP + free β -hCG + uE3 + Inhibin A	83 ^(c)

(a) Lam YH et al. Comparison and integration of first trimester fetal nuchal translucency and second trimester maternal serum screening for fetal Down syndrome. *Prenat Diagn.* 2002 Aug;22(8):730-5.

(b) Wald NJ, Bestwick JP, Huttly WJ, et al. Prenatal screening for Down syndrome in twin pregnancies: Estimates of screening performance based on 61 affected and 7302 unaffected twin pregnancies. *Prenat Diagn.* 2018;38:1079–1085. <https://doi.org/10.1002/pd.5381>

(c) Wald NJ et al. First and second trimester antenatal screening for Down's syndrome: the results of the Serum, Urine and Ultrasound Screening Study (SURUSS). *J Med Screen.* 2003;10(2):56-104. Erratum in: *J Med Screen.* 2006;13(1):51-2.

1.2 A trained and qualified sonographer shall obtain an operator code from PDL, TYH in order to request for Nuchal Screening and 1st Trimester Combined Screening tests which involve nuchal measurements.

Please send a copy of the 'FMF Certificate of competence in the measurement of nuchal translucency' to Scientific Officer (Tel: 2589-2327) who will assign an operator code for each operator.

(Important note: Operators are responsible to renew their FMF Certificates yearly).

2. Instructions to doctors on sending samples

2.1 Complete an appropriate request form as indicated in the table in Section 2.2 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 of sampling)
- Test(s) requested
- Gestational weeks and EDC (by scan or by date)
- Clinical details (information needed for examination performance and results interpretation including patient’s ancestry, family history)
- Referring doctor’s / nurse’s signature (it is assumed that consent of patient has been obtained by signing the request form)
- Important note:
 - a) For assisted reproduction with embryo transfer, please indicate whether fresh or frozen embryos were transferred, and provide the date of egg collection, date of embryo transfer, and donor’s date of birth or age (if applicable).
 - b) Complete the nuchal translucency measurement section including the operator code (please refer to Section 1.2 on how to obtain an operator code).
 - c) For pregnancies with a vanishing twin, please refer to the following table for the appropriate type of Down syndrome screening test.

Gestational age (weeks)	Ultrasound scan finding of demised twin	Down syndrome screening tests
11 - 13 ⁺⁶	with fetal pole	Nuchal Screening
	with gestation sac, no fetal pole	Either Nuchal Screening or 2 nd Trimester Quadruple test preferably after 18 wk
16 - 19 ⁺⁶	with gestation sac, no fetal pole	2 nd Trimester Quadruple test preferably after 18 wk
	with fetal pole	No biochemical screening

(Spencer K et al. First trimester aneuploidy screening in the presence of a vanishing twin: implications for maternal serum markers. Prenat Diagn 2010 Mar;30(3):235–40.)

- d) The collected blood sample shall reach the laboratory within 48 hours, counting from the time of blood collection. If the sample cannot be sent to the laboratory on the day of blood collection, please keep it at the door of refrigerator (2-8°C). Samples should be delivered in a courier box/bag on ice.

2.2 Specimen specification

Collect 5 mL of peripheral blood into a serum tube (*prohibit using barrier gel tube*) labelled with at least two patient’s identifiers. Cap and invert the tube well.

Request forms and specimen specification for the corresponding test as specified in the following table:

Down syndrome screening tests	Gestational age (weeks)	CRL (mm)	Specimen (volume)	Container (provided upon request)	Request form	Turn-around time
Nuchal Screening	11 - 13 ⁺⁶	42 - 83	--	--	WHITE form Request form for Nuchal Screening (HA) (TYH-REQ-NUCHAL-HA)	3 working days
1 st Trimester Combined Screening Test	11 - 13 ⁺⁶	42 - 83	Peripheral blood (5 mL)	Serum tube*	BLUE form Request form for 1st trimester DS (HA) (TYH-REQ-1stDS-HA)	
2 nd Trimester Quadruple Screening Test	16 - 19 ⁺⁶	--	Peripheral blood (5 mL)	Serum tube*	YELLOW form Request form for 2nd trimester DS (HA) (TYH-REQ-2ndDS-HA)	

*Prohibit using barrier gel tube

2.3 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)

3 Rejection of sample

A specimen may be rejected when the following condition is observed:

- unlabelled or incorrectly labelled specimen container
- specimen container leaks
- not suitable for analysis (e.g. frozen or grossly haemolysed, lipaemic or icteric blood sample, using incorrect container, etc.)
- specimen cannot reach the laboratory within 48 hours after blood collection
- specimen unfit for the tests described in Section 2.1c and 2.2

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

4 Reporting

- All reports will be sent to your office by messengers. Screen positive reports will be faxed to your office. All Down syndrome screening reports can be accessed via Electronic Patient Record (ePR).
- Turn-around-time (TAT): 3 working days

(Note: After delivery of the baby or miscarriage, please complete the section of “Reply from Obstetric Units” at the bottom of the report and then return a copy of the report (by fax or by post) for auditing purpose.)

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: 8:45am - 5:30pm

(closed on Saturdays, Sundays and public holidays)

Contacts: Medical Technologist
Senior Medical Technologist
Scientific Officer

Tel: 2589-2212
Tel: 2589-2288
Tel: 2589-2327 / 2589-2328

} Down syndrome
screening service

Duty Officer / Laboratory Director

Tel: 2589-2288 / 2589-2327
Fax: 2517-2373

} Other enquiry or
complaint

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