

## Down Syndrome Screening

### Reporting Details:

	<b>First Trimester Combined Down Syndrome Screening</b>	<b>Second Trimester Biochemical Down Syndrome Screening</b>																						
<b>Turnaround Time**</b>	7 days (HA case) Please contact laboratory (private case) **Assuming both sample and <b>Laboratory Request Form</b> satisfy all requirements on arrival.	7 days																						
<b>Detection Rate</b>	90%	80%																						
<b>False Positive Rate</b>	5%	5%																						
<b>Risk Outcome by</b>	Gestational Adjusted Risk (Private cases) Term Adjusted Risk (HA case)	Term Adjusted Risk																						
<b>Risk Outcome Entity</b>	T21, T18, T13	T21, T18																						
<b>Screen Negative Threshold (SNT)</b>	Gestational Adjusted Risk (Private cases) <table border="1" data-bbox="537 1087 933 1178"> <thead> <tr> <th></th> <th>T21</th> <th>T18</th> <th>T13</th> </tr> </thead> <tbody> <tr> <td>SNT</td> <td>1:180</td> <td>1:150</td> <td>1:150</td> </tr> </tbody> </table> Term Adjusted Risk (HA case) <table border="1" data-bbox="537 1304 933 1394"> <thead> <tr> <th></th> <th>T21</th> <th>T18</th> <th>T13</th> </tr> </thead> <tbody> <tr> <td>SNT</td> <td>1:250</td> <td>1:250</td> <td>1:250</td> </tr> </tbody> </table>		T21	T18	T13	SNT	1:180	1:150	1:150		T21	T18	T13	SNT	1:250	1:250	1:250	Term Adjusted Risk <table border="1" data-bbox="959 1087 1268 1178"> <thead> <tr> <th></th> <th>T21</th> <th>T18</th> </tr> </thead> <tbody> <tr> <td>SNT</td> <td>1:250</td> <td>1:250</td> </tr> </tbody> </table>		T21	T18	SNT	1:250	1:250
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<b>Reporting media</b>	1. Automatic email or automatic fax to defaulted recipient (private cases) 2. Automatic uploading to HA ePR system (HA cases)																							
<b>Report Sample</b>	Appendix 3,4	Appendix 5,6																						

**Patient Details**

Name : TESTING\_HA ID : PESTING\_HA DOB : 10 Jul 1986  
Hospital: Prince of Wales Hospital (Requestor Dr TY Leung) Case No: TESTING\_HA Sex : F

**Maternal Details**

Ethnicity : East Asian (Chinese)  
Actual DOB: 10 Jul 1986  
Gravida: Parity :  
Smoker: No  
Diabetic: No

**Previous History of Aneuploidy**

Trisomy 21: No Trisomy 18: No  
Trisomy 13: No Other: No

**Pregnancy Details**

Conception: Spontaneous  
EDC: 18 Feb 2019  
Dating Method: By CRL (1st Trm)  
Age at EDC: 32.61 years

**Maternal Serum Details**

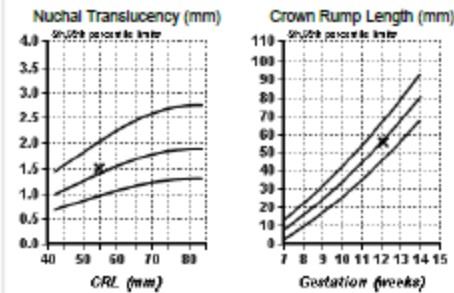
Lab Ref: TESTING\_HA Taken: 07 Aug 2018  
Sample: Clotted Blood Analysed: 08 Aug 2018  
Analyser: Kryptor K1009 Weight: 55.2 Kg

	Measurement	MoM
Free $\beta$ -hCG	93 IU/L	1.7284 MoM
PAPPA	4 IU/L	0.9942 MoM

**Ultrasound Examination Details**

Scan Date: 07 Aug 2018 Gest: 12 wks 1 days by EDD  
Sonographer: Leung Tak Yeung


**Fetus**  
Crown Rump Length 55.4 mm  
Nuchal Translucency 1.5 mm



**Term Specific Estimated Risk for Aneuploidy**

	Trisomy 21	Trisomy 18	Trisomy 13
Term Background Risk	1:670	1:7924	1:18654
Term Adjusted Risk :	1:6629 (Negative)	Negative	Negative

Screen Negative Threshold: Trisomy 21 < 1:250 ; Trisomy 18 < 1:250 ; Trisomy 13 < 1:250 ;

Reported by   
Tsang Ho Kau Fanny  
MT100984  
Report Date 09 Aug 2018 09:34

This is a screening and NOT a diagnostic test. A screen negative result does not exclude the possibility of a chromosomal abnormality. The adjusted risk is determined from the maternal age background risk, ultrasound factors (nuchal translucency) and maternal serum biochemistry (free  $\beta$ -hCG and PAPPA) which are adjusted for maternal and pregnancy characteristics. The adjusted risk is only valid if the assessment of Nuchal Translucency was performed by an accredited Sonographer who has undergone training and annual recertification. The Laboratory participates in the United Kingdom National External Quality Assurance Scheme (UKNEQAS). The model used to derive the patient specific risk is derived from information reported in the literature and research conducted by the Department of Obstetrics and Gynaecology of the Chinese University of Hong Kong.

Hardcopy for reference only. The latest version of report can be accessed via the ePR.

Appendix 3. First Trimester Combined Down Syndrome Screening Report  
(Screened Negative Report Sample)

**Patient Details**



Name : TESTING\_POSITIVE ID : TESTING\_POSITIVE DOB : 01 Jul 1986  
Hospital: Prince of Wales Hospital (Requester Dr. TY Leung) Case No: TESTING\_POSITIVE Sex : F

**Maternal Details**

Ethnicity : East Asian (Chinese)  
Actual DOB: 01 Jul 1986  
Gravida: 1 Parity : 0  
Smoker: No  
Diabetic: No

**Previous History of Aneuploidy**

Trisomy 21: No Trisomy 18: No  
Trisomy 13: No Other: No

**Pregnancy Details**

Conception: Spontaneous  
EDC: 18 Feb 2019  
Dating Method: By CRL (1st Trim)  
Age at EDC: 32.64 years

**Maternal Serum Details**

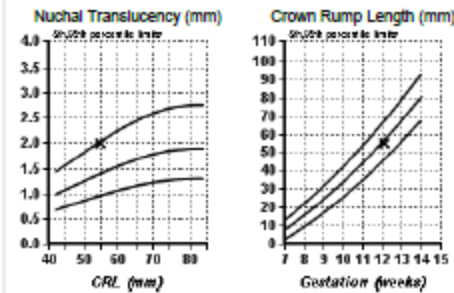
Lab Ref: Testing\_positive Taken: 07 Aug 2018  
Sample: Clotted Blood Analysed: 08 Aug 2018  
Analyser: Kryptor K1009 Weight: 55 Kg

	Measurement	MoM
Free β-hCG	93 IU/L	1.7231 MoM
PAPPA	1 IU/L	0.2474 MoM

**Ultrasound Examination Details**

Scan Date: 07 Aug 2018 Gest: 12 wks 1 days by EDD  
Sonographer: Ting Yuen Ha

**Fetus**  
Crown Rump Length 55 mm  
Nuchal Translucency 2 mm

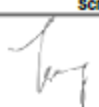


**Term Specific Estimated Risk for Aneuploidy**

	Trisomy 21	Trisomy 18	Trisomy 13
Term Background Risk	1:668	1:7891	1:18578
Term Adjusted Risk :	1.8 (Positive)	Negative	Negative

Screen Negative Threshold: Trisomy 21 < 1:250 ; Trisomy 18 < 1:250 ; Trisomy 13 < 1:250 ;

This is a screening and NOT a diagnostic test. A screen negative result does not exclude the possibility of a chromosomal abnormality. The adjusted risk is determined from the maternal age background risk, ultrasound factors (nuchal translucency) and maternal serum biochemistry (free β-hCG and PAPPA) which are adjusted for maternal and pregnancy characteristics. The adjusted risk is only valid if the assessment of Nuchal Translucency was performed by an accredited Sonographer who has undergone training and annual recertification. The Laboratory participates in the United Kingdom National External Quality Assurance Scheme (UKNEQAS). The model used to derive the patient specific risk is derived from information reported in the literature and research conducted by the Department of Obstetrics and Gynaecology of the Chinese University of Hong Kong.

Reported by:   
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Report Date 09 Aug 2018 09:51

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Appendix 4. First Trimester Combined Down Syndrome Screening Report  
(Screened **Positive** Report Sample)



**Patient Details**



Name : TESTING\_HA\_2ND TRIM      ID : TESTING\_HA\_2ND TRIM      DOB : 15 Aug 1990  
Hospital: Prince of Wales Hospital (Requester Dr. TY Leung)      Case No: TESTING\_HA\_2ND TRIM      Sex : F

**Maternal Details**

Ethnicity : East Asian (Chinese)  
Actual DOB: 15 Aug 1990  
Gravida: 1 Parity : 0  
Smoker: No  
Diabetic: No

**Previous History of Aneuploidy**

Trisomy 21: No      Trisomy 18: No  
Trisomy 13: No      Other: No

**Maternal Serum Details**

Lab Ref: Testing\_HA\_2nd trim      Taken: 02 Aug 2018  
Sample: Clotted Blood      Analysed: 07 Aug 2018  
Analyser: AutoDelfia      Weight: 50 Kg

	Measurement	MoM
AFP	100 ng/mL	1.9488 MoM
Free β-hCG	10 IU/L	0.8247 MoM
uE3	8 nmol/L	1.3192 MoM
Inhibin A	500 pg/mL	2.3243 MoM

**Pregnancy Details**

Conception: Spontaneous  
EDC: 06 Jan 2019  
Dating Method: From Screening Centre  
Age at EDC: 28.39 years

**Neural Tube Defect Screening**

Negative (AFP < 2.5 MoM)

**Term Specific Estimated Risk for Aneuploidy**

	<u>Trisomy 21</u>	<u>Trisomy 18</u>
Term Background Risk	1:1129	1:13332
Term Adjusted Risk :	1:11295 (Negative)	Negative

Screen Negative Threshold: Trisomy 21 < 1:250 ; Trisomy 18 < 1:250 ;

Reported by **Tsang Ho Kau Fanny**  
MT100984  
Report Date 09 Aug 2018 10:42

This is a screening and NOT a diagnostic test. A screen negative result does not exclude the possibility of a chromosomal abnormality. The adjusted risk is determined from the maternal age background risk and maternal serum biochemistry (free β-hCG, AFP, uE3, Inhibin) which are adjusted for maternal and pregnancy characteristics. The Laboratory participates in the United Kingdom National External Quality Assurance Scheme (UKNEQAS). The model used to derive the patient specific risk is derived from information reported in the literature and research conducted by the Department of Obstetrics and Gynaecology of the Chinese University of Hong Kong.

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**Appendix 5. Second Trimester Biochemical Down Syndrome Screening Report  
(Screened Negative Report Sample)**

**Patient Details**

Name : TESTING\_HA\_2ND TRIM\_POSITIVE ID : TESTING\_HA\_2ND TRIM\_POSITIVE DOB : 01 Dec 1990  
Hospital: Prince of Wales Hospital (Requester Dr. TY Leung) Case No: TESTING\_HA\_2ND TRIM\_ Sex : F



**Maternal Details**

Ethnicity : East Asian (Chinese)  
Actual DOB: 01 Dec 1990  
Gravida: 1 Parity : 0  
Smoker: No  
Diabetic: No

**Previous History of Aneuploidy**

Trisomy 21: No Trisomy 18: No  
Trisomy 13: No Other: No

**Maternal Serum Details**

Lab Ref: Testing\_HA\_2nd trim\_ Taken: 02 Aug 2018  
Sample: Clotted Blood Analysed: 07 Aug 2018  
Analyser: AutoDelfia Weight: 52 Kg

	Measurement	MoM
AFP	10 ng/mL	0.1993 MoM
Free $\beta$ -hCG	50 IU/L	4.2344 MoM
uE3	1 nmol/L	0.1666 MoM
Inhibin A	100 pg/mL	0.4739 MoM

**Pregnancy Details**

Conception: Spontaneous  
EDC: 06 Jan 2019  
Dating Method: From Screening Centre  
Age at EDC: 28.1 years

**Neural Tube Defect Screening**

Negative (AFP < 2.5 MoM)

**Term Specific Estimated Risk for Aneuploidy**

	Trisomy 21	Trisomy 18
Term Background Risk	1:1156	1:13655
Term Adjusted Risk :	1:3 (Positive)	Negative

Screen Negative Threshold: Trisomy 21 < 1:250 ; Trisomy 18 < 1:250 ;



Reported by Tsang Ho Kau Fanny  
MT100984  
Report Date 09 Aug 2018 10:42

This is a screening and NOT a diagnostic test. A screen negative result does not exclude the possibility of a chromosomal abnormality. The adjusted risk is determined from the maternal age background risk and maternal serum biochemistry (free  $\beta$ -hCG, AFP, uE3, Inhibin) which are adjusted for maternal and pregnancy characteristics. The Laboratory participates in the United Kingdom National External Quality Assurance Scheme (UKNEQAS). The model used to derive the patient specific risk is derived from information reported in the literature and research conducted by the Department of Obstetrics and Gynaecology of the Chinese University of Hong Kong.

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**Appendix 6. Second Trimester Biochemical Down Syndrome Screening Report  
(Screened Positive Report Sample)**