



Update 14
Changes to Maternal Serum Screening
for
Down Syndrome, Trisomy 18 and Open Neural Tube Defects

March 2009

Dear Colleague,

The South Australian Maternal Screening Antenatal Screening (SAMSAS) Program Advisory Group has recommended the following changes to the Program as part of ongoing review and development of maternal screening services.

A. Screening window ¹

Maternal blood samples for 1st trimester screening will be accepted from 9 weeks 0 days. The first trimester screening window changes to *9w0d – 13w6d*. The window for nuchal translucency remains at 11w0d – 13w6d. The second trimester screening window also remains unchanged at 14w0d – 20w6d.

B. Reporting of risk odds ¹

Risk odds of the foetus having either Down Syndrome or Trisomy 18 will be expressed as *odds at the time of screening* instead of odds at term. Maternal age related risks will also be expressed at time of screening. This will apply for both first and second trimester screens.

C. Decision point ¹

The decision point for offering a patient an invasive diagnostic test will become *1 in 250* for both first and second trimester screens. This is in response to (B) and will maintain the balance between detection rate and false positive rate.

D. Too late and too early reports

Following an ultrasound, gestation may be different to expected and the wrong screen type will have been ordered. To avoid reports such as “too late for a first trimester screen” or “too early for a second trimester screen”, *tick both boxes on the SAMSAS request form*. If both “First Trimester Screen” and “Second Trimester Screen” boxes are ticked, SAMSAS will perform the appropriate screen according to the best estimate of gestation, based either on the CRL at the time of the nuchal translucency scan or by the average biometry for more advanced pregnancies when the CRL is > 84 mm.

E. Integrated screening ²

Integrated screening uses combinations of both first and second trimester markers and results in lower false positive rates and higher detection rates than either first or second trimester screens alone. However, the first trimester combined nuchal translucency and biochemical screen remains the primary population screen of choice. Integrated screening *is not recommended as the primary population screening strategy* for a number of practical reasons.

Integrated tests ²

Integrated testing (as opposed to integrated population screening) will be offered by SAMSAS:

- 1) If the patient is undecided after her first trimester combined screen and requests a repeat screen, then second trimester markers can be requested for a *full combined integrated test*.
- 2) If a first trimester blood is received, first trimester nuchal translucency is not measured and a second trimester blood sample is received subsequently – a *biochemistry only integrated test* will be performed.
- 3) If a first trimester nuchal translucency is measured but the blood sample is too late (14+ weeks) for a first trimester combined screen – the nuchal translucency will be used *in combination with second trimester markers*.
- 4) If specifically asked for (with the requester taking primary responsibility for appropriate requesting), co-ordination of first and second trimester blood samples and first trimester nuchal translucency provides clear instruction on when the risk odds are required i.e. after completion of the second trimester biochemical testing.

F. Thickened Nuchal Translucency ³

Published studies have shown that chromosomally normal foetuses with increased nuchal translucencies have an increased risk of cardiac and other foetal morphological abnormalities.

- 1) A nuchal translucency thickness of ≥ 3 mm is considered to warrant further investigation.
- 2) In cases where the nuchal translucency is ≥ 3 mm, the following sub-note will be included in the SAMSAS report; *“Please note that the nuchal translucency is ≥ 3 mm. This is associated with an increased risk of cardiac and other foetal morphological abnormalities. Review at morphology scan is advised.”*

These changes will be phased in from April 2009.

Please direct any queries to Mr Robert Cocciolone
phone: 08 8161 7285
email: robert.cocciolone@health.sa.gov.au

Yours sincerely,



Robert Cocciolone
BAppSc, Med Lab Sc, Head, Antenatal Screening (SAMSAS) Program

References

1. HGSA/RANZCOG College Statement C-Obs 4, July 2007
2. Cocciolone et al Combining first and second trimester markers for Down syndrome screening: Think twice Aust N Z J Obstet Gynaecol 2008;48: 492-500
3. Nicolaides et al Increased fetal nuchal translucency at 11-14 weeks Prenat Diag 2002;22:308-315