SOUTH AUSTRALIAN MATERNAL SERUM ANTENATAL SCREENING (SAMSAS) PROGRAMME

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Update 11
Down syndrome Audit
First Trimester Screening

SAMSAS is pleased to present performance figures from an audit of 6,255 valid risks based on the combined ultrasound and biochemical screening for fetal Down syndrome in first trimester.

The auditing of this data was performed as described in the paper *Ryall et al*., Karyotypes found in the population declared at increased risk of Down syndrome following maternal serum screening. *Prenat Diagn 2001:21: 553-557*. This process achieves ascertainment of 98.5% of the population screened.

Our expectations from first trimester screening programme was to have a performance that was at least as effective or better than second trimester screening programme. Two objective markers which can be used to measure effectiveness are recall and detection rates expressed as a % of the population screened. Recall rate is simply the number of pregnancies screened at increased risk of Down syndrome. The detection rate is the number of affected pregnancies screened at increased risk relative to the number of affected pregnancies in the screened population.

When comparing detection rates one must allow for the fetal loss which would occur between the first and second trimester period. One such study has done this and results are summarised in table 1. Reference, *Prenat Diagn* 2001; 21: 788-793 What is the true fetal loss rate in pregnancies affected by trisomy 21 and how does this influence whether first trimester detection rates are superior to those in the second trimester?

Table 1 First trimester detection rate needed to better that of the respective second trimester detection rate using various estimates of fetal loss and second trimester detection rates.

	60% Second trimester detection	65% Second trimester detection	70% Second trimester detection	75% Second trimester detection
1st Trimester detection needed	**67.72**	**72.18**	**76.52**	**80.7**

Table 2 shows the relative performances of the various screening modalities possible in first trimester compared to that in second trimester.

To make comparisons easier a fixed recall rate of 5% was chosen.

Table 2

Screening Modality	Number Screened	% Recalled	Number Of Pregnancies Affected With Down syndrome	**% of affected pregnancies Detected**
2nd Trimester	11,782	5	21	61.9
1st Trimester Combined Biochemistry & Nuchal Translucency	**6,255**	**5**	**18**	**77.8**
1st Trimester Biochemistry Only	6,255	5	18	65
1st Trimester Nuchal Translucency only	6,255	5	18	50

Table 2 shows that the performance of the combined first trimester screening strategy (allowing for fetal loss) far exceeds that of the second trimester programme, first trimester biochemistry only and nuchal translucency only screening. In all screening modalities maternal age risk at delivery was used in the risk calculation.

The actual detection for the above first trimester cases using the combined strategy was 88.9% (16/18) with a recall rate of 6.0%, using a risk cut off of 1:300. The median maternal age at delivery for this group was 31.7 years.

Figure 1 compares the separation of unaffected and affected populations using first and second trimester markers. From this it can be seen that the combined screening strategy in first trimester gives less overlap between the unaffected and affected populations, accounting for the greater sensitivity over the second trimester markers.

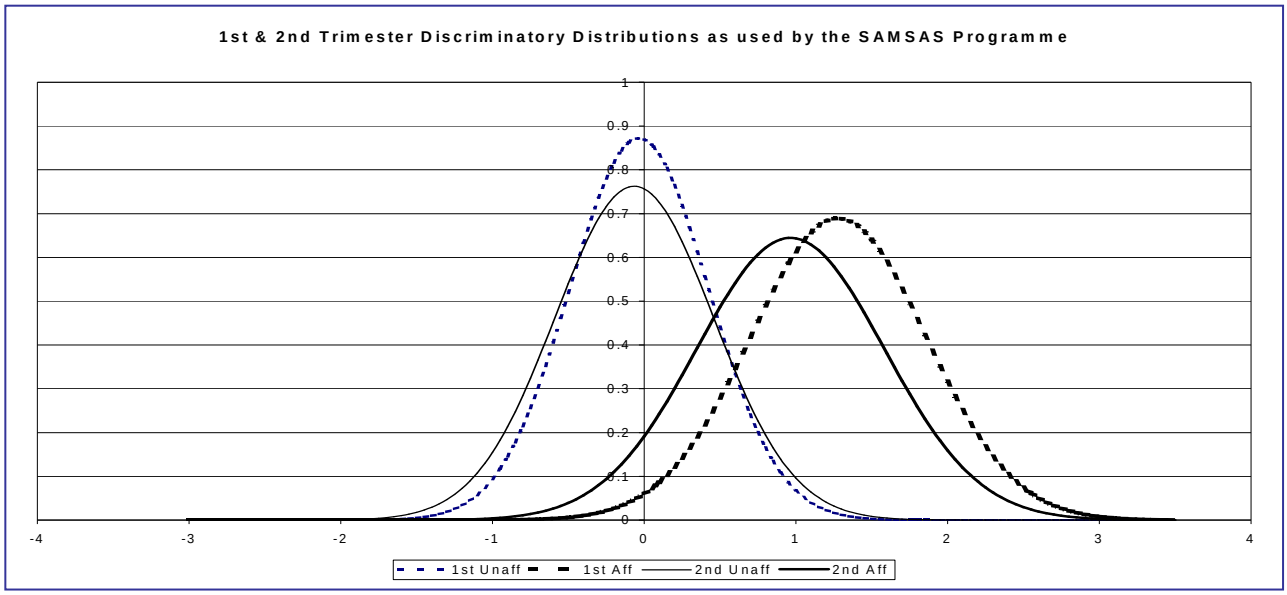

Figure 1

Figure 2 shows the distribution of each marker in the 18 cases affected with Down syndrome, in the screened population.

The median values were **NT_MoM = 1.33, BHCG_MoM = 1.93 and PAPP_A_MoM = 0.33.** It is worth noting this group of Downs have relatively low nuchal thicknesses, with only 8 of the 18 having measurements greater than 2mm. An explanation for this would be that some selection was occurring however this does not appear to be happening as the observed prevalence of Downs in this screened group of 1:347 (18/6255) is as expected.

This finding does however further support the use of the combined screening strategy.

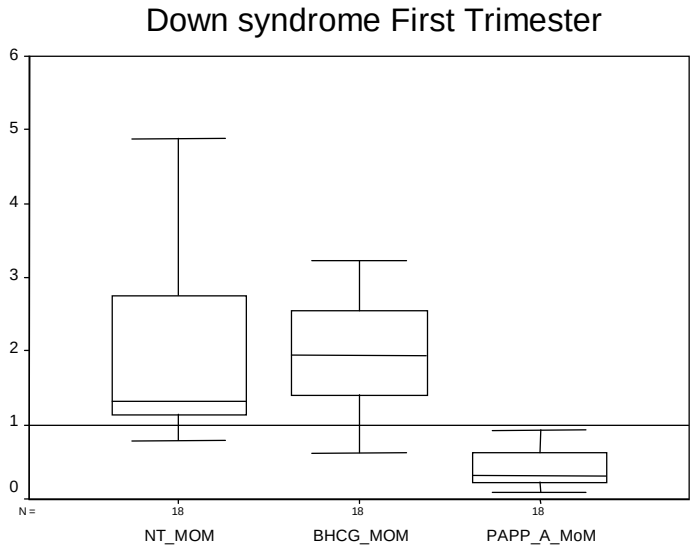

Figure 2

*** The data presented and the performances quoted in this report are those of the SAMSAS programme and do not apply to other software or testing centres.**

**** PTO for Requesting Procedure.**

Second trimester screening of pregnancies for fetal Down syndrome and neural tube defects **remains in place** and can still be requested as before. We recommend, however, that if a pregnancy is screened in first trimester then any request in second trimester be confined to **neural tube defect (NTD) screening only**. First trimester screening does not include detection of fetal NTDs.

Requesting first trimester screening

Two request forms are required, one for the blood analysis and one for the ultrasound scan.

BLOOD ANALYSIS

1. 5-10 mL clotted blood sample, taken between 10 and 13 weeks gestation is required. A list of collection centres is provided on the reverse of the SAMSAS request form.
2. Use a SAMSAS request form, telephone (08) 8161 7285 if you require some of these,
   (a) the test request is "**first trimester screen**",
   (b) complete the gestational age information, the gestation must be between 10+ and 13+ weeks,
   (c) specify the ultrasound practice performing the nuchal translucency scan,
   (d) refer **patient to the Privacy Act Disclosure on the SAMSAS request form**,
   (e) give the patient the SAMSAS pre-test information booklet,
   (f) send the blood specimen to Women's and Children's Hospital, **for interstate or remote areas check with SAMSAS on what services are available.**

ULTRASOUND

3. Book a **Nuchal Translucency** scan with the imaging group of choice. The fetus must be between **11+ and 13+ weeks** gestation at the time of the scan.
4. Complete an ultrasound request form, specifying "**risk of fetal abnormality**"; and **"Copy to SAMSAS"**. To comply with National Privacy Legislation, refer **patient to the Privacy Act Disclosure on the SAMSAS request form**.

SAMSAS will coordinate the results with the ultrasound practice and you will receive a single report giving the risks calculated for the pregnancy. Post-test information booklets are provided with all reports issued by SAMSAS on pregnancies found at increased risk of fetal abnormality.

Availability of first trimester screening

Combined ultrasound and biochemistry screening is not at present offered through all hospitals/clinics. Check with the hospital/clinic concerned.

Costs

For privately insured patients SAMSAS continues its policy of accepting 'Medicare only' for the serum biochemistry analyses. There may be a gap payment for the ultrasound measurement. Check with the practice providing this service.

Robert Cocciolone, BAppSc, Med Lab Sc
on behalf of the South Australian Maternal Serum Antenatal Screening (SAMSAS) Programme

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Gestational Age Windows for Antenatal Screening for Birth Defects

First Trimester	**Blood sample 10w0d – 13w6d**	**Ultrasound 11w0d – 13w6d**
Second Trimester	**Blood sample 14w0d – 20w6d**	