

**SOUTH AUSTRALIAN & TASMANIAN MATERNAL SERUM  
ANTENATAL SCREENING  
(SAMSAS) PROGRAM**

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**Update 12  
Prenatal Screening for Down syndrome  
Age Specific Performance  
1<sup>st</sup> & 2<sup>nd</sup> Trimester Screening**

**The SAMSAS program** provides screening services to hospitals and private health practitioners in South Australia, Tasmania and Northern Territory. It also provides screening service support to the Princess Margaret Hospital in Western Australia.

**The SAMSAS first trimester screen** involves a risk calculation derived from the combination of two biochemical placental markers - free beta hCG and Papp-A, with the ultrasound marker nuchal translucency.

**The second trimester screen** uses biochemical markers only - alpha-fetoprotein, free beta hCG and unconjugated estriol.

**The likelihood ratio** derived from the marker combinations in 1<sup>st</sup> or 2<sup>nd</sup> trimester screens is used to adjust the risk at delivery of having a baby with Down syndrome. For both screens a risk cut off of 1:300 is used to categorise the pregnancy as either "at increased risk" or "not at increased risk". The cut off of 1:300 determines the percentage of pregnancies screened at increased risk (recall rate, RR) and the percentage of affected pregnancies which can be detected (detection rate, DR). For a 5% recall rate, we would expect to detect 75 – 90% of all affected pregnancies with Down syndrome in the first trimester, and 60-75% in the second trimester.

**In this Update** we use 65 first trimester and 80 second trimester marker profiles from Down syndrome pregnancies and over 1000 marker profiles from unaffected pregnancies to produce age-specific performance data for each trimester. These data are presented in table 1 and figures 1-3. The prevalence of Down syndrome at the time of screening was calculated for each maternal age in order to calculate the odds of having an affected pregnancy, following either an "at increased risk" or "not at increased risk" report. The prevalence used was calculated from the maternal age risk at delivery, adjusted for expected fetal loss after the time of screening from both 1<sup>st</sup> and 2<sup>nd</sup> trimesters.

**These data highlight the improved specificity achieved with the 1<sup>st</sup> trimester combined screen and the differential performance across age groups. We believe the data will assist in counselling and decision making by providing both health professional and patient with odds that better reflect outcomes.**

Details on how to request a first trimester screen follow on page 4.

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



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**1<sup>st</sup> & 2<sup>nd</sup> Trimester Screening**  
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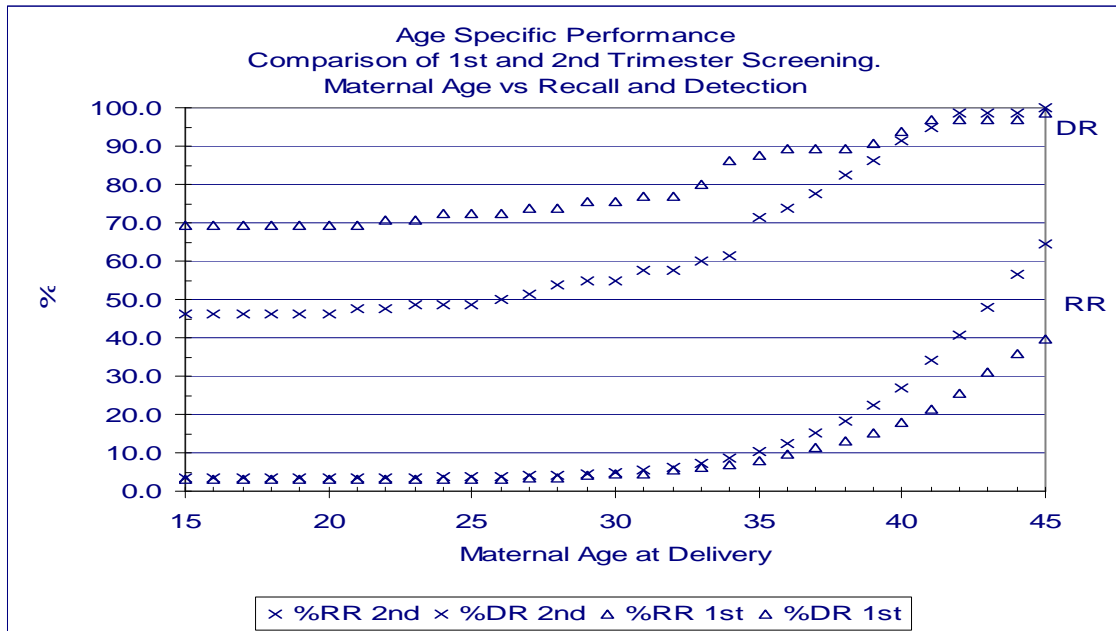
**HOW TO READ THIS TABLE**

For a 30 year old screened using the **1<sup>st</sup> Trimester Combined Screen**, there would be a **4.4%** chance of being screened “at increased risk” and a **75.4%** chance of detecting an affected pregnancy. For a pregnancy screened “at increased risk” there would be a **1 in 44** chance that the pregnancy is affected by Down syndrome and for a pregnancy screened “not at increased risk”, the chance would be **1 in 2,962**. Comparative performance figures for a second trimester screen are provided in the last 4 columns.

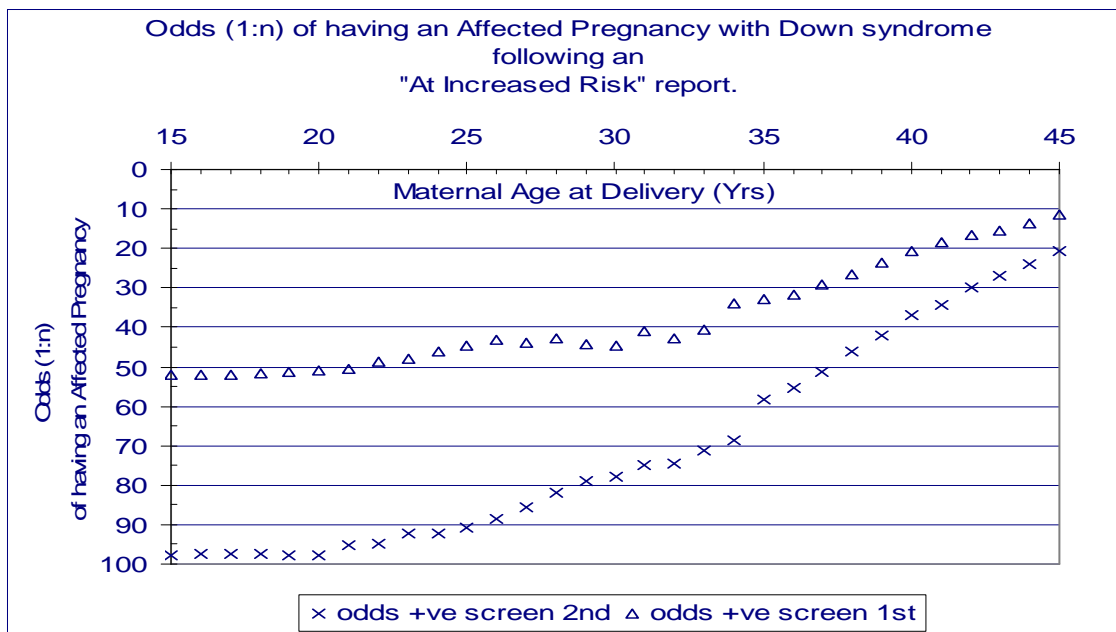
**Table 1**

<b>Maternal Age at Delivery Years</b>	<b>Maternal Age Risk at Delivery 1:n</b>	<b>1<sup>st</sup> Trimester Combined Screen % of Pregnancies Screened At Increased Risk</b>	<b>1<sup>st</sup> Trimester Combined Screen % of Affected Pregnancies Detected</b>	<b>1<sup>st</sup> Trimester Combined Screen Odds of having an Affected Pregnancy following an At Increased Risk Report 1:n</b>	<b>1<sup>st</sup> Trimester Combined Screen Odds of having an Affected Pregnancy following a Not at Increased Risk Report 1:n</b>	<b>2<sup>nd</sup> Trimester Screen % of Pregnancies Screened at Increased Risk</b>	<b>2<sup>nd</sup> Trimester Screen % of Affected Pregnancies Detected</b>	<b>2<sup>nd</sup> Trimester Screen Odds of having an Affected Pregnancy following an At Increased Risk Report 1:n</b>	<b>2<sup>nd</sup> Trimester Screen Odds of having an Affected Pregnancy following a Not at Increased Risk Report 1:n</b>
15	1663	3.1	69.2	52	3662	3.4	46.3	98	2391
16	1659	3.1	69.2	52	3654	3.4	46.3	98	2385
17	1654	3.1	69.2	52	3643	3.4	46.3	98	2378
18	1647	3.1	69.2	52	3627	3.4	46.3	98	2367
19	1638	3.1	69.2	51	3607	3.5	46.3	98	2354
20	1627	3.1	69.2	51	3583	3.5	46.3	98	2337
21	1611	3.1	69.2	51	3548	3.5	47.5	95	2369
22	1591	3.1	70.8	49	3696	3.5	47.5	95	2338
23	1564	3.1	70.8	48	3633	3.6	48.8	92	2354
24	1531	3.1	72.3	46	3749	3.7	48.8	92	2302
25	1487	3.1	72.3	45	3641	3.7	48.8	91	2235
26	1433	3.1	72.3	43	3509	3.9	50.0	88	2204
27	1367	3.4	73.8	44	3528	4.0	51.3	86	2153
28	1288	3.5	73.8	43	3321	4.3	53.8	82	2133
29	1195	4	75.4	44	3264	4.5	55.0	79	2028
30	1089	4.4	75.4	44	2962	4.9	55.0	78	1841
31	975	4.6	76.9	41	2819	5.5	57.5	75	1734
32	855	5.5	76.9	43	2448	6.3	57.5	75	1508
33	733	6.3	80	40	2404	7.3	60.0	71	1359
34	617	6.8	86.2	34	2917	8.5	61.3	68	1166
35	509	8.1	87.7	33	2662	10.2	71.3	58	1272
36	412	9.8	89.2	32	2409	12.4	73.8	55	1100
37	329	11.3	89.2	29	1891	15.1	77.5	51	993
38	259	13	89.2	26	1460	18.4	82.5	46	966
39	202	15.2	90.8	24	1303	22.4	86.3	42	912
40	156	17.9	93.8	21	1446	27.0	91.3	37	1041
41	119	21.5	96.9	18	1500	34.2	95.0	34	812
42	91	25.5	96.9	17	1531	40.6	98.8	30	775
43	69	30.9	96.9	15	1077	48.0	98.8	27	753
44	52	36	96.9	14	751	56.7	98.8	24	746
45	40	39.6	98.5	11	1127	64.7	100.0	21	n/a

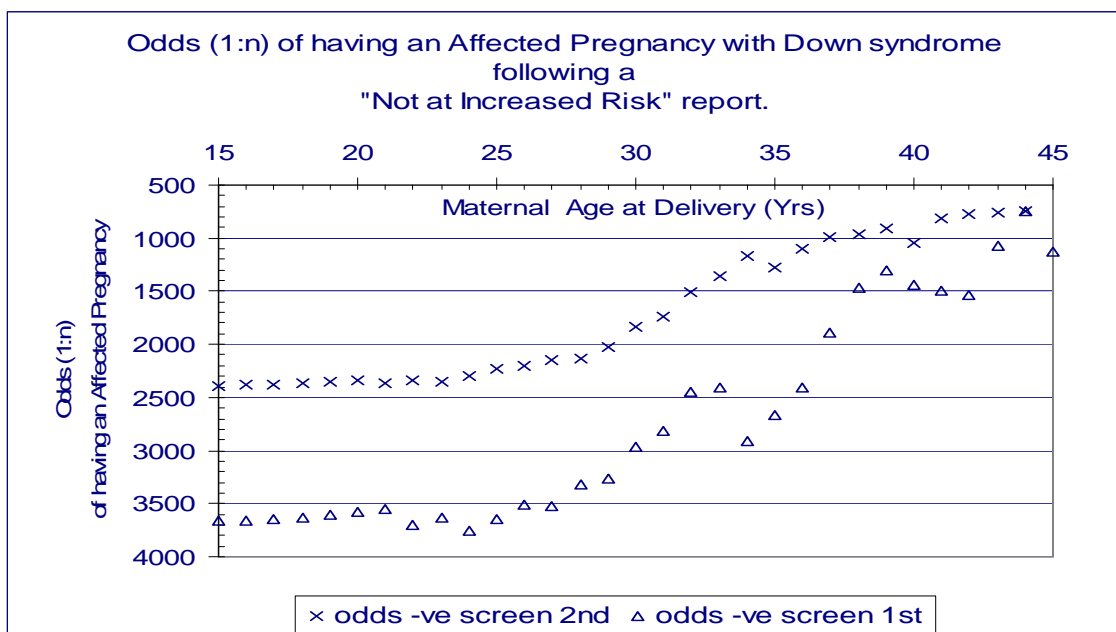
**Fig. 1**



**Fig. 2**



**Fig. 3**



## Requesting first trimester screening

Two request forms are required, one for the blood analysis and one for the nuchal translucency 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 10w0d – 13w6d,
  - (c) specify the ultrasound practice performing the nuchal translucency scan,
  - (d) refer **patient to the Privacy 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 **11w0d – 13w6d** 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 the National Privacy Legislation and Fair Information Code, refer **patient to the Privacy 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.

**Second trimester screening** of pregnancies for fetal Down syndrome and neural tube defects remains in place with the accepted timing of the blood sample being 14w0d to 20w6d. We recommend, 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.

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#### Gestational Age Windows for Antenatal Screening for Birth Defects

<b>1st Trimester</b>	<b>Blood sample 10w0d – 13w6d</b>	<b>Optimal gestation 10 - 12 weeks</b>
	<b>Ultrasound 11w0d – 13w6d</b>	<b>Optimal gestation 11 - 12 weeks</b>
<b>2nd Trimester</b>	<b>Blood sample 14w0d – 20w6d</b>	<b>Optimal gestation 16 weeks</b>