Women's & Children's Hospital

Prenatal Screening for Congenital Anomalies in South Australia 2013

South Australian Birth Defects Register
Women's and Children's Hospital
Adelaide, South Australia
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Government of South Australia
SA Health

Women's & Children's Hospital

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South Australian Birth Defects Register Staff

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Executive Summary

In 2013:

- 280 chorionic villus samplings and 823 amniocenteses were performed on South Australian women, representing 1.4% and 4.1% of all women who gave birth, respectively.
- Maternal age was a factor in 50% of chorionic villus samplings and 47% of amniocenteses.
- 2 fetuses underwent fetal blood sampling, with one subsequent fetal transfusion.
- The South Australian Maternal Serum Antenatal Screening Programme (SAMSAS) screened 2,033 pregnancies (10.2% of all pregnancies) at 15-20 weeks gestation for a fetal neural tube defect by estimation of maternal serum alpha-fetoprotein. This can be compared to a peak of around 83% in the early-mid 1990s, reflecting increasing reliance on ultrasound as the main screening method for neural tube defects.
- 23 of 24 cases of neural tube defect that had screening by maternal serum screening, ultrasound screening or both, were detected prenatally.
- SAMSAS used first trimester combined or second trimester maternal serum screening to detect Down syndrome in 17,153 pregnancies (86.1% of all pregnancies in SA).
- Of the 58 Down syndrome cases prenatally screened or tested by one or more screening or testing method, 55 (94.8%) were detected. The screening or diagnostic methods used were:
    - First trimester combined screen (nuchal translucency and maternal serum screen),
    - Second trimester maternal serum screen,
    - Chorionic villus sampling,
    - Amniocentesis,
    - Ultrasound alone as the first indication of Down syndrome.
- There have been significant changes over time in the proportion of pregnancies in which prenatal diagnosis by chorionic villus sampling or amniocentesis is performed. In 1986 this proportion was 5% and peaked in 1996 with 12%. In 2013 this proportion was 5.5%. A contributing factor to the fall in recent years has been a decrease in the proportion of women 35 years and older using chorionic villus sampling and amniocentesis, following the introduction of first trimester Down syndrome screening, and more recently, the introduction of non-invasive prenatal testing.

Introduction

The Annual Report of Prenatal Screening for Congenital Anomalies in South Australia records the 2013 experience based on the techniques of chorionic villus sampling (CVS), amniocentesis, fetal blood sampling, first trimester combined screening by nuchal translucency and maternal serum screening, and second trimester maternal serum screening for neural tube defects and Down syndrome.

In South Australia, screening for open neural tube defects began in 1978. In 1990 and 1991, second trimester screening for Down syndrome and other pregnancy pathologies was introduced, followed by first trimester screening in 2000. Integrated screening was introduced in 2009, using combinations of both first and second trimester markers. More recently, the introduction of non-invasive prenatal testing has enabled pregnant women to have a blood test from 10 weeks gestation. This blood sample is then analysed for fetal trisomies 21, 18 and 13, (and sometimes for X-chromosome abnormalities) with varying levels of sensitivity and specificity. As with the first and second trimester screening programs, non-invasive prenatal testing is considered to be a screening test, and pregnant women should be counselled and always offered confirmation of abnormal results with diagnostic testing.

Diagnostic tests are invasive and carry a small risk to the pregnancy, and are generally performed after a high risk screen. CVS is performed in the 1st trimester, and amniocentesis is performed in the 2nd trimester. Patients will choose the most appropriate test for them after counselling. Some choose to proceed to diagnostic testing without screening tests.

No attempt has been made to compile information on pregnancies where ultrasound was the sole diagnostic technique used to detect birth defects. Its role in the detection of neural tube defects is recorded in the review of the maternal serum alpha-fetoprotein screening programme.

It should be noted that this report included cases *screened* in each calendar year. This contrasts with the SA Birth Defects Register report which includes cases *born or terminated* in each calendar year.

We are grateful to the following groups for providing data for this report:

- South Australian Maternal Serum Antenatal Screening Programme (SAMSAS)
- Genetics and Molecular Pathology Directorate, SA Pathology
- Pregnancy Outcome Statistics Unit, SA Health
- Clinical Information Services, Women's and Babies Division, WCH

Table 1: Screening and diagnostic tools used to detect congenital anomalies

Screen/Test	First Trimester	Second Trimester
Screening for Down syndrome	Non-invasive prenatal testing ≥ 10^{+0} weeks	Maternal Serum Screening (βHCG/uE_3/AFP) 14^{+0} to 20^{+6} weeks
	Nuchal Translucency (NT) plus Maternal Serum Screening (βHCG/PAPP-A) 9^{+0} to 13^{+6} weeks	
Screening for neural tube defects	Ultrasound from 12 weeks	Maternal Serum Screening (AFP) 14^{+0} to 20^{+6} weeks Ultrasound*
Screening for congenital malformations	Ultrasound** from 12 weeks	Ultrasound 19-20 weeks
Diagnostic testing tools for chromosome abnormalities and genetic disorders	Chorionic villus sampling (CVS) 10-13 weeks	Amniocentesis After 15 weeks

**Ultrasound can detect neural tube defects at any stage of the 2nd trimester, but the planned time for screening is typically at the routine morphology scan at 19-20 weeks.*

*** Whilst not part of a routine screening programme, the 12 week ultrasound is increasingly capable of detecting fetal anomalies*

βHCG – beta human chorionic gonadotropin; PAPP-A – Pregnancy-associated plasma protein A; uE_3 – Unconjugated estriol; AFP – Alpha-fetoprotein

Trends in chorionic villus sampling and amniocentesis

Since 1986, the percentage of all women undertaking prenatal diagnostic CVS or amniocentesis peaked at 12% in 1996 (Figure 1). In 2013 the proportion was 5.5% of all women who gave birth, lower than 2012 (Table 2). This is only a slightly higher percentage than those undertaken in 1986 (5.0%). Over time, there has been an increase in prenatal diagnoses by CVS as a percentage of the total prenatal diagnoses and therefore a decrease in diagnoses by amniocentesis. Reasons for this increase may include first trimester screening results being available in time for CVS to be offered, or increased utilisation of first trimester screening.

Figure 1: Percentage of chorionic villus sampling and amniocentesis by year for all women: all indications

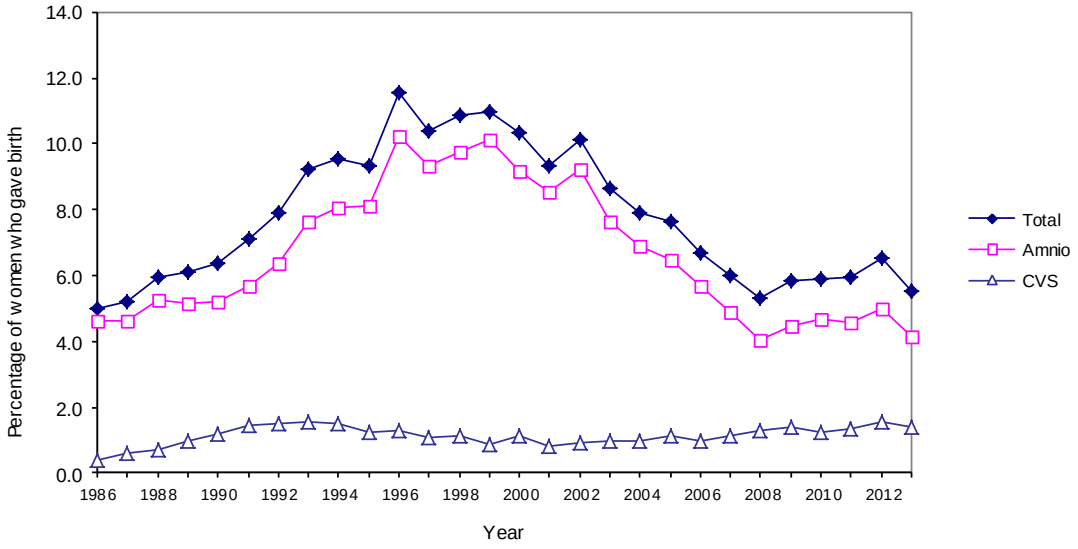


Table 2: Prenatal diagnosis by technique						
Year	**CVS (% total)**		**Amniocentesis (% total)**		**Total**	**Percentage of women who gave birth**
1986-1990	747	(13.4%)	4810	(86.6%)	5557	5.7%
1991-1995	1412	(16.8%)	7012	(83.2%)	8424	8.6%
1996-2000	995	(10.0%)	8908	(90.0%)	9903	10.9%
2001-2005	842	(11.0%)	6790	(89.0%)	7632	8.7%
2006-2010	1160	(20.1%)	4604	(79.9%)	5764	5.9%
2011	269	(22.6%)	921	(77.4%)	1190	5.9%
2012	316	(23.8%)	1013	(76.2%)	1329	6.5%
2013	280	(25.4%)	823	(74.6%)	1103	5.5%

Trends in chorionic villus sampling and amniocentesis

The number of CVS performed in 2013 was 280 (1.4% of all women who gave birth), a decrease compared with 2012 when 316 (1.6%) samplings were performed. Maternal age was a factor in 141 (50%) of all CVS performed in 2013.

The number of amniocenteses performed in 2013 on SA women was 823 (4.1% of all women who gave birth), a decrease compared with 2012 when 1,013 (5.0%) amniocenteses were performed. Maternal age was a factor in 387 (47%) of all amniocenteses performed in 2013.

Table 3: Chorionic villus sampling and amniocentesis, SA 2013: Indications

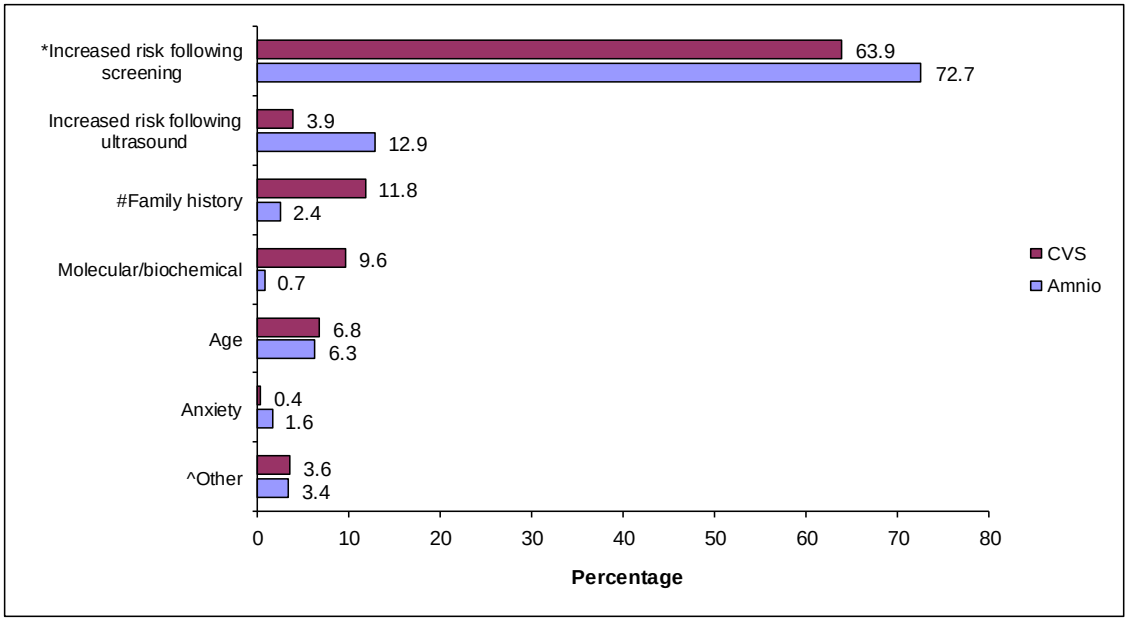
Indication	CVS		Amniocentesis		Total
	<35 yrs	≥35 yrs	<35 yrs	≥35 yrs	
Screening At increased risk of chromosomal abnormality following screening by MSS* or NT[#]					
1. 1st trimester combined screen	89	87	287	257	720
2. 1st trimester NT alone	0	0	0	0	0
3. 1st trimester MSS alone	1	0	0	1	2
4. 2nd trimester MSS alone	-	-	18	23	41
5. Integrated screen	-	-	6	4	10
At increased risk of NTD[^] following MSS*	-	-	0	0	0
At increased risk of chromosomal abnormality following non-invasive prenatal testing[$]	0	2	1	1	4
Abnormality found on ultrasound	5	6	81	25	117
Family History Previous child with					
1. Down syndrome	1	4	2	5	12
2. Other chromosome abnormality	7	4	0	3	14
3. Neural tube defect	0	0	0	0	0
Family history of					
1. Down syndrome	0	0	0	0	0
2. Other chromosome abnormality[%]	13	4	7	3	27
3. Neural tube defect	0	0	0	0	0
At increased risk of a disorder diagnosed by molecular or biochemical techniques	18	9	5	1	33
Other Indications Maternal age ≥ 35 years	-	19	-	52	71
Maternal anxiety/maternal age < 35 years	1	-	13	-	14
Following abnormal CVS result	-	-	1	2	3
Other (eg failed cordocentesis)	0	0	8	4	12
Blood group antibodies	0	0	1	1	2
Paternity	2	1	2	0	5
Multiple reasons	2	5	4	5	16
Total	**139**	**141**	**436**	**387**	**1103**

** Maternal serum screening; [#] Nuchal translucency; [^] Neural tube defect; [$] two of these women also had high risk 1st trimester combined screening; [%]includes translocation carrier parent*

Trends in chorionic villus sampling and amniocentesis

Figure 2 illustrates the main reasons for having chorionic villus sampling or amniocentesis in 2013. The most common reason for having chorionic villus sampling or amniocentesis was increased risk following screening during pregnancy (63.9% and 72.7% respectively). CVS was more commonly performed for pre-existing conditions, such as family history of chromosomal defects, or where the fetus was at increased risk of a disorder diagnosed by molecular or biochemical techniques. In contrast, amniocentesis was more often utilised when increased risk of abnormalities were identified during the pregnancy, such as an increased risk following ultrasound, or following first or second trimester screening. Maternal age also remained a significant factor for both CVS and amniocentesis.

Figure 2: Chorionic villus sampling and amniocentesis, SA 2013: Utilisation by reason



**any increased risk of chromosomal abnormality or neural tube defect following first or second trimester screening, including non-invasive fetal DNA testing.*
[#]any family history or previous child with Down syndrome, other chromosome abnormality or neural tube defect.
^includes: following abnormal CVS result, other specified reasons, multiple reasons, blood group/antibody testing, paternity or unknown indication.

Trends in chorionic villus sampling and amniocentesis

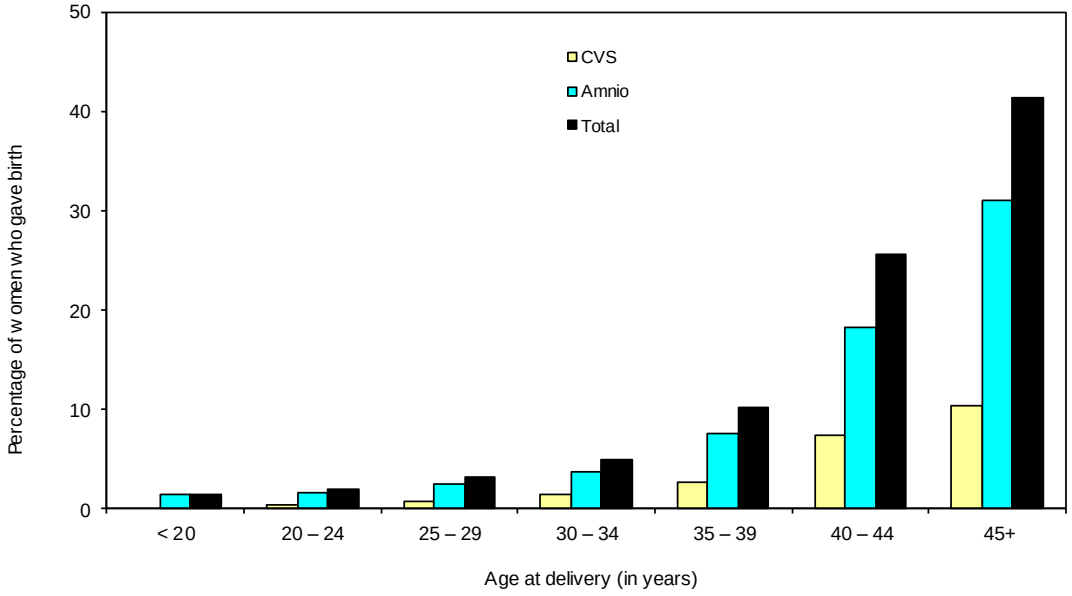
Table 4 and Figure 3 illustrate the utilisation of chorionic villus sampling and amniocentesis by maternal age in 2013. Women in the 45+ age group had the highest utilisation, with 41.4% of all women giving birth in that age range undergoing one of these diagnostic tests.

Table 4: Chorionic villus sampling and amniocentesis: Utilisation by maternal age

Age*	CVS	Amniocentesis	Total	No. of women who gave birth	Percentage^
<20	0	9	9	666	1.4
20-24	9	45	54	2881	1.9
25-29	41	144	185	5858	3.2
30-34	89	238	327	6576	5.0
35-39	82	240	322	3158	10.2
40-44	56	138	194	757	25.6
45+	3	9	12	29	41.4
Total	**280**	**823**	**1103**	**19925**	**5.5**

**Age in years at expected delivery date; ^Percentage of women who gave birth in that age range*

Figure 3: Chorionic villus sampling and amniocentesis: Utilisation by maternal age



Trends in chorionic villus sampling and amniocentesis

Figure 4 shows the proportion of chorionic villus sampling or amniocenteses undertaken by women ≥ 35 years of age. Although the number of women who gave birth at age 35 years and over has risen from 1,285 in 1986 to 3,499 in 2013, the proportion of women in this age group having chorionic villus sampling or amniocentesis has been steadily decreasing over time, and plateauing in the last few years (Figure 4). This is most evident from 1996 onwards. This decrease may be explained by an increasing proportion of women having first trimester screening and therefore less women directly requesting chorionic villus sampling or amniocentesis (see Figures 5 and 6).

Figure 4: Percentage of chorionic villus sampling and amniocentesis by year for maternal age ≥35 years, SA 1986-2013: all indications

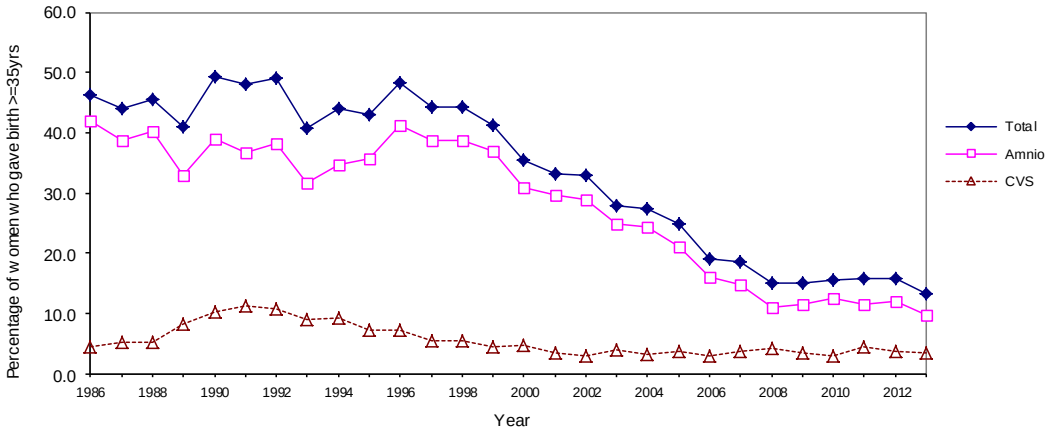
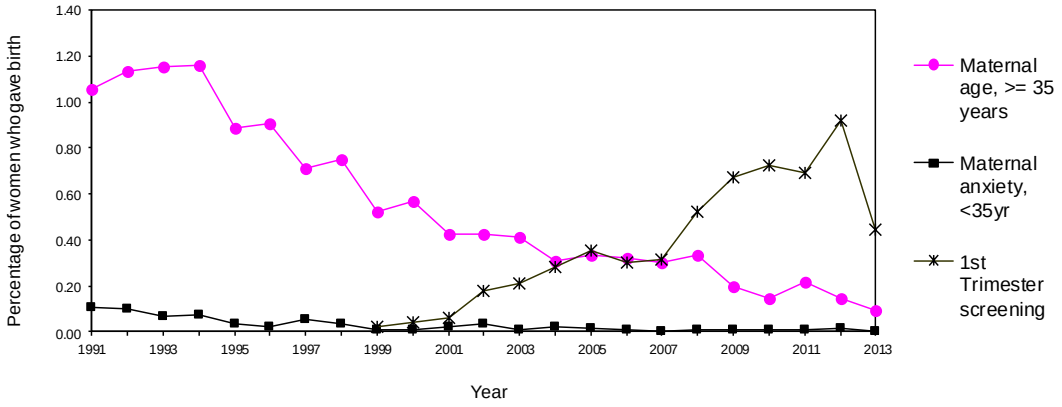


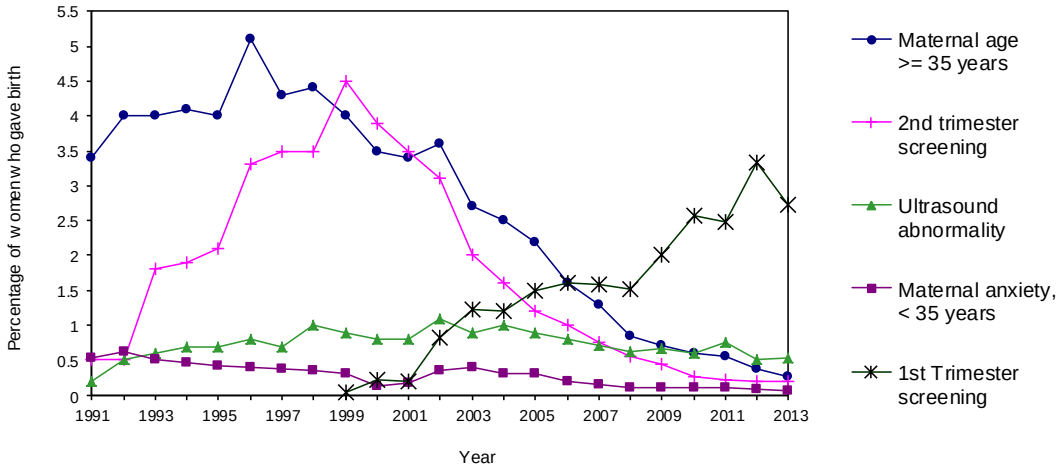
Figure 5: Indications for chorionic villus sampling by year, SA 1991*-2013: Percentage of all women who gave birth



**No routine screening prior to 1991*

Trends in chorionic villus sampling and amniocentesis

Figure 6: Indications for amniocentesis by year, SA 1991*-2013: Percentage of all women who gave birth



**No routine screening prior to 1991*

Fetal blood sampling

Fetal blood is obtained from the umbilical cord or a blood vessel in the fetal liver by an ultrasound guided needle technique. In 2013, 2 fetal blood samplings were performed. A fetal transfusion was performed in one of the fetuses during the pregnancy.

Table 5: Fetal blood sampling, SA 2013: Indications

Indication	No. of procedures	No. of fetuses	Outcome
Rhesus or other isoimmunisation	2	2	1 fetal transfusion
Total	**2**	**2**	

Disorders diagnosed by molecular or biochemical techniques

Table 6 provides information on the specific disorders that were diagnosed by molecular or biochemical techniques in 2013. The indications for chorionic villus sampling or amniocentesis in this table are most often associated with a family history of the specific disorder being investigated.

Table 6: Disorders diagnosed by molecular or biochemical techniques, SA 2013

Indication	Chorionic villus sampling		Amniocentesis	
	Tested	Affected	Tested	Affected
Achondroplasia	-	-	1	1
Acrocallosal syndrome	1	0	-	-
Adrenoleukodystrophy	1	0	-	-
Alport syndrome	1	0	-	-
Cystic fibrosis	4	1	-	-
Familial adenomatous polyposis	1	1	-	-
Fragile X syndrome	1	1	1	0
Glutaric aciduria type II	1	1	1	0
Glycogen storage disease type II (Pompe disease)	1	0	-	-
Haemophilia	2	0	-	-
Hereditary angiodema	1	1	-	-
Huntington disease	3	2	-	-
Incontinentia pigmenti	1	0	-	-
Myotonic dystrophy	3	2	2	2
Panhypopituitarism	1	0	-	-
Pontocerebellar hypoplasia	1	0	1	0
Pyruvate carboxylase deficiency	1	0	-	-
Smith-Lemli-Opitz syndrome	1	0	-	-
Spinal muscular atrophy	2	1	1	0
Thalassaemia	2	0	2	1
Williams syndrome	-	-	1	0
X-linked hydrocephalus	1	0	-	-
X-linked intellectual disability	1	0	-	-
X-linked retinitis pigmentosa	1	0	1	1
Zellweger syndrome	1	1	-	-
Blood group testing (including Rhesus)	-	-	1	n/a

Maternal serum screening for neural tube defects in the 2nd trimester

In 2013, 2,033 pregnancies were screened by estimation of maternal serum alpha-fetoprotein by the South Australian Maternal Serum Antenatal Screening (SAMSAS) programme at 15-20 weeks gestation for a fetal neural tube defect. This represents 10.2% of all pregnancies.

There were 25 cases of neural tube defect in SA births whose mothers reached 14 weeks of pregnancy on or after 1st January 2013 or were no more than 20^{+6} weeks by 31st December 2013 and hence would have been eligible for maternal serum alpha-fetoprotein screening during the 2013 screening year. This figure includes all neural tube defects confirmed in terminations of pregnancy or in births (≥ 400g or ≥ 20 weeks gestation).

95.8% of fetuses with a neural tube defect that had screening by either maternal serum alpha-fetoprotein or ultrasound, or both, were detected prenatally.

Table 7: Maternal serum screening for neural tube defects, SA 2013: SAMSAS

Number of pregnancies screened 2013	2,033
Total women who gave birth in SA in 2013	19,925
Percentage of pregnancies screened	10.2%

Table 8: Neural tube defects screening, SA 2013: Cases detected / Not detected

Method of detection	Anencephaly	Spina bifida	Encephalocele	Total
1. AFP screen abnormal, as first indication of neural tube defect	1	0	0	1
2. Ultrasound abnormal, as first indication of neural tube defect				
(a) No serum AFP	1	7	2	10
(b) Serum AFP normal	1	1	0	2
(c) Too early for AFP	10	0	0	10
3. Not detected on AFP and/or ultrasound screening	0	0	1	1
4. No screening	0	1	0	1
Total[#]	**13**	**9**	**3**	**25**

AFP = alpha-fetoprotein; [#]Neural tube defects detected as a result of maternal serum AFP and/or ultrasound screening or elective testing because of previous affected child, or from examination of child at delivery.

Table 9: Neural tube defects screening, SA 2013: Outcome of cases detected

Outcome	Anencephaly	Spina bifida	Encephalocele	Total
Livebirth	0	2	1	3
Stillbirth	1	0	0	1
Termination of pregnancy	12	7	2	21
Total[#*]	**13**	**9**	**3**	**25**

[#]Neural tube defects detected as a result of maternal serum AFP and/or ultrasound screening or elective testing

**Does not include one case detected interstate and born in SA*

Screening for Down syndrome

There are both screening and diagnostic tests for Down syndrome during pregnancy. The screening tests include nuchal translucency (NT) screening, 1st trimester maternal serum screening (MSS), and the most commonly used 1st trimester test, combined NT and 1st trimester MSS. Second trimester MSS is also available if 1st trimester screening has not been performed. Integrated screening is now offered for patients who have had both 1st and 2nd trimester screening. More recently, non-invasive prenatal testing has become available in South Australia, via pathology providers who then send the maternal blood sample to the USA or China for testing and reporting.

Diagnostic tests are invasive and carry a small risk to the pregnancy; they are chorionic villus sampling (CVS) performed in the 1st trimester, and amniocentesis performed in the 2nd trimester. They are generally performed after a high risk screen. Patients will choose the most appropriate test for them after counselling. Some choose to proceed to diagnostic testing without screening tests.

Pregnancies are screened in the 1st trimester by the combination of nuchal translucency screening and maternal serum screening (free βHCG and PAPP-A). Software developed by SAMSAS or the Fetal Medicine Foundation (FMF) is used to estimate the risk for each pregnancy, based on blood analyte and nuchal translucency results and maternal age. Pregnancies are screened in the 2nd trimester by maternal serum screening. Table 10 details the cut-off points for recommending consideration of CVS or amniocentesis for the different risk providers.

NT thickness for each fetus in multiple pregnancies parallels that of singleton pregnancies. For dichorionic twins, a Down syndrome risk for each fetus is issued using the combination of NT thickness and maternal age. For monochorionic twins, who should be genetically identical, a single risk assessment is reported. These twins will have an identical maternal age-related Down syndrome risk, but may show different NT thickness; the greater risk is reported for counselling purposes. A 75% detection rate of affected multiple pregnancies is achievable according to published data using 1:250 as the cut-off. The use of serum markers for multiple pregnancies greater than twins is currently not offered in SA but marker levels are measured for future studies to assess their possible utility.

Women were allocated to the 2013 screening year if their screening test was performed in 2013. Where no screen was performed, women were allocated to the 2013 screening year if they were eligible to have completed their antenatal screening in 2013. Because there was no formal screening program which incorporates non-invasive prenatal testing in 2013, we are only reporting this when specifically informed that it was used in a particular pregnancy.

There were 64 cases of Down syndrome included in the 2013 screening year. This figure includes all Down syndrome cases confirmed in terminations of pregnancy or in births (≥ 400g or ≥ 20 weeks gestation). Of the 58 Down syndrome cases prenatally screened or tested by one or more screening or testing method, 55 (94.8%) were detected.

Screening for Down syndrome

Table 10: Risk providers for Down syndrome screening tests and risk cut-off points[#]						
Risk provider	**1st trimester screen**		**2nd trimester screen**		**Integrated screen**	
	Test	**Risk cut-off**	**Test**	**Risk cut-off**	**Test**	**Risk cut-off**
SAMSAS*	✓	1:250	✓	1:250	✓	1:250
FMF	✓	1:300	✗	-	✗	-

**SAMSAS changed from risk cut-off 1:300 to 1:250 in April 2009*
[#]Risk cut-off points are used for recommending consideration of further diagnostic testing, including chorionic villus sampling or amniocentesis

Table 11: Down syndrome screening, SA 2013: All testing laboratories		
	Pregnancies Screened*	
First trimester screening	15,592	(78.3%)
Second trimester screening only[^]	1,561	(7.8%)
Total number of pregnancies screened[#]	17,153	(86.1%)

**numbers are expressed as a percentage of the total number of women who gave birth in SA in 2013 (19,925)*
[^]does not include 472 women who had 2nd trimester screening for neural tube defects only
[#]A further 0.6% of women who gave birth (129 cases) did not have screening, and instead had a diagnostic CVS or amniocentesis either specifically for Down syndrome (n=40), or for other reasons (n=89) resulting in a total of 86.7% of pregnancies having some form of investigation to screen for or identify Down syndrome.

Table 12: Down syndrome screening, SA 2013: Cases detected / Not detected	
1. Cases detected: Pregnancies screened (by screening method n=55)*	
First trimester screening (nuchal translucency plus MSS[+])	47
Second trimester screening (MSS[+])	3
CVS (without prior screening)	3
Amniocentesis (without prior screening)	0
Ultrasound	2
2. Not detected prenatally (n=9)	
First trimester screening (nuchal translucency plus MSS[+])	3
Second trimester screening (MSS[+])	0
Integrated screening (first and second trimester screening markers)	0
Not screened (diagnosed after birth)	6
Total	**64**

[+]MSS = maternal serum screening;
** Three cases also had non-invasive prenatal testing performed, in association with other screening or detection methods*

Screening for Down syndrome

Table 13: Down syndrome screening, SA 2013: Outcome of cases	
Livebirth	16
Stillbirth	0
Termination of pregnancy	48
Total	**64**

Due to changes in reporting processes, Tables 14 and 15 only provide information for pregnancies where SAMSAS calculated and reported the Down syndrome risk following 1st trimester combined biochemical and nuchal translucency. Pregnancies screened using Fetal Maternal Foundation (FMF) software have not been reported to the SA Birth Defects Register from 2010 onwards.

Table 14: First trimester combined biochemical and nuchal translucency screening by maternal age, SA 2013: SAMSAS only

Maternal age	<35 years	≥35 years	Total
Number of pregnancies screened with valid risks reported* (% of pregnancies screened)	10,281 (81.3%)	2,370 (18.7%)	**12,651 (100%)**
Identified as "increased risk" after correction of gestational age (% of pregnancies screened)	388 (3.7%)	404 (17.0%)	**792 (6.3%)**
Total CVS/Amniocentesis performed on pregnancies identified as "increased risk" (%)	283 (72.9%)	253 (62.6%)	**536 (67.7%)**
Affected pregnancies in screened population	16	23	**39**
Affected pregnancies among those screened at "increased risk"	15	21	**36**
Sensitivity (%)	93.8	91.3	**92.3**
Risk of an affected pregnancy in those at "increased risk" (risk ≥1:250) on screening (positive predictive value, PPV)	1:26	1:19	**1:22**

**"pregnancies screened with valid risks reported" exclude pregnancies which are <9 weeks and >14 weeks gestation and duplicate samples*

Screening for Down syndrome

Table 15: First trimester combined biochemical and nuchal translucency screening by maternal age, SA 2001-2013: SAMSAS only			
Maternal age	**<35 years**	**≥35 years**	**Total**
Number of pregnancies screened with valid risks reported* (% of pregnancies screened)	86,679 (79.7%)	22,058 (20.3%)	**108,737 (100%)**
Identified as "increased risk" after correction of gestational age (% of pregnancies screened)	2,771 (3.2%)	2,739 (12.4%)	**5,510 (5.1%)**
Total CVS/Amniocentesis performed on pregnancies identified as "increased risk" (%)	2,127 (76.8%)	1,972 (72.0%)	**4,099 (74.4%)**
Affected pregnancies in screened population	124	171	**295**
Affected pregnancies among those screened at "increased risk"	105	155	**260**
Sensitivity (%)	84.7	90.6	**88.1**
Risk of an affected pregnancy in those at "increased risk" (risk ≥1:250) on screening (positive predictive value, PPV)	1:26	1:18	**1:21**
**"pregnancies screened with valid risks reported" exclude pregnancies which are <9 weeks and >14 weeks gestation and duplicate samples*			

Table 16 provides information for pregnancies where SAMSAS collected and reported the Down syndrome risk following 2nd trimester maternal serum screening.

Table 16: Second trimester maternal serum screening by maternal age, SA 2013: SAMSAS only			
Maternal age	**<35 years**	**≥35 years**	**Total**
Number of pregnancies screened with valid risks reported* (% of pregnancies screened)	835 (83.5%)	165 (16.5%)	**1,000 (100%)**
Identified as "increased risk" after correction of gestational age (% of pregnancies screened)	26 (3.1%)	33 (20.0%)	**59 (5.9%)**
Amniocentesis performed on pregnancies identified as "increased risk" (%)	16 (61.5%)	21 (63.6%)	**37 (62.7%)**
Affected pregnancies in screened population	1	2	**3**
Affected pregnancies among those screened at "increased risk"	1	2	**3**
Sensitivity (%)	100	100	**100**
Risk of an affected pregnancy in those at "increased risk" (risk ≥1:250) on screening (positive predictive value, PPV)	1:26	1:16	**1:19**
**"pregnancies screened with valid risks reported" exclude pregnancies which are <14 weeks and >20 weeks gestation, duplicate samples, and those requested for neural tube defect risk only.*			

Screening for Down syndrome

Table 17 presents South Australian data (1986-2013) for the risk that a woman of a given age will give birth to a baby with Down syndrome. As illustrated in the table below, this data is comparable with published international data. Variations seen within and between the two datasets may reflect a number of factors: the relatively small number of women in different age groups; the numbers of births recorded (approximately 540,000 for SA vs approximately 6,000,000 for International); and the data collection periods (1986-2013 for SA vs 1990-1998 for International).

Table 17: Risk of Down syndrome by maternal age, SA 1986-2013

Mother's age at delivery	South Australia	International*
16	1:2378	1:2013
17	1:2542	1:1599
18	1:1339	1:1789
19	1:1611	1:1440
20	1:2725	1:1441
21	1:1568	1:1409
22	1:1096	1:1465
23	1:1213	1:1346
24	1:1409	1:1396
25	1:1297	1:1383
26	1:1181	1:1187
27	1:1208	1:1235
28	1:1119	1:1147
29	1:1215	1:1002
30	1:800	1:959
31	1:676	1:837
32	1:558	1:702
33	1:428	1:589
34	1:314	1:430
35	1:254	1:338
36	1:203	1:259
37	1:212	1:201
38	1:129	1:162
39	1:125	1:113
40	1:67	1:84
41	1:58	1:69
42	1:47	1:52
43	1:37	1:37
44	1:26	1:38
45+	1:34	1:30

**Data obtained from:*

- *Morris JK, Mutton DE and Alberman E. Revised estimates of the maternal age specific live birth prevalence of Down's syndrome. Journal of Medical Screening 2002; 9: 2 (for maternal ages 16-44)*
- *Morris JK, De Vigan C, Mutton DE and Alberman E. Risk of a Down syndrome live birth in women 45 years of age and older. Prenatal Diagnosis 2005; 25: 275-278 (for maternal ages 45 and greater)*