SAMSAS UPDATE 16
January 2017

SA PATHOLOGY

Changes to Maternal Serum Screening Test for Down syndrome (Trisomy 21), Trisomy 18 and Open Neural Tube Defects (NTD).

As part of its continuous improvement program the South Australian Maternal Serum Antenatal Screening (SAMSAS) program has recently made changes.

Testing platform

In 2014 SAMSAS changed the testing platform for βhCG, PAPP-A and αFP. The Roche platform offers superior analytical performance, resulting in a lower false positive rate (FPR) of 4.3%.

Reducing the false positive rate means fewer women require invasive testing such as chorionic villus sampling (CVS) or amniocentesis counselling.

Blood collection screening windows

- 1st trimester screening from 9 to 14 weeks (9w0d -14w0d)
- 2nd trimester screening from 14 weeks and 1 day to 20 weeks and 6 days (14w1d-20w6d)

Non-Invasive Prenatal Testing (NIPT) screening

SA Pathology intends to introduce Non-Invasive Prenatal Testing (NIPT) into the SAMSAS program in 2017. This testing method uses genomics technology with DNA extracted from maternal blood to ascertain risk of aneuploidy. Any 'positive' NIPT screening test will always require confirmation by standard cytogenetic chromosomal analysis by either CVS or amniocentesis.

Discontinued tests

We no longer offer Integrated Screening combining markers from 1st and 2nd trimester serum for detection of aneuploidy. Any 2nd trimester combined markers from 1st and 2nd trimester serum blood samples received on patients who have had a 1st trimester screening test will be performed as Neural Tube Defect Only (NTDO) screening by AFP measurement.

SA Pathology is no longer offering AFP measurement in Amniotic Fluid, but continues to offer AFP tests on blood.

Further information

If you have any clinical enquiries please call Enzo Ranieri, Head of Biochemical Genetics on 08 8161 6739 or enzo.ranieri@sa.gov.au

Please refer to tables overleaf which provide details of SAMSAS test statistics and results performance, measured over 6 years (2010–15).

Table 1: SAMSAS performance for 1st trimester screening 2010-2015

	2010	2011	2012	2013	2014	2015
Number of tests performed	15,474	17,032	17,889	18,373	18,188	18,502
Number of cases identified at increased risk (1 in 250 at time of screen)	758	800	1061	1070	995	839
Detected Trisomy 21 confirmed by karyotype	26	25	44	41	40	37
Missed Trisomy 21 screened as low risk	2	0	4	3	3	2
Detection Rate	**92.3%**	**100%**	**90.9%**	**92.6%**	**95%**	**94.5%**
False Positive Rate	**4.7%**	**4.5%**	**5.7%**	**5.6%**	**5.2%**	**4.3%**
Specificity	95.3%	95.56%	94.3%	94.4%	94.7%	95.7%

**Current screening performance reflects overall detection rate of 94% and false positive rate (FPR) of 4.3%.*

Table 2: SAMSAS performance for 2nd trimester screening 2010-2015

	2010	2011	2012	2013	2014	2015
Number of tests performed	2,828	2,714	2,616	2,250	2,083	1,702
Number of cases identified at increased risk (1 in 250 at time of screen)	126	118	123	99	86	107
Detected Trisomy 21 confirmed by karyotype	5	2	6	3	3	4
Missed Trisomy 21 screened as low risk	0	0	2	0	0	0
Detection Rate	**100%**	**100%**	**66.7%**	**100%**	**100%**	**100%**
False Positive Rate	**4.2%**	**4.3%**	**4.5%**	**4.3%**	**4.0%**	**6.1%**
Specificity	95.7%	95.7%	95.5%	95.7%	96.0%	93.9%

Table 3: Number of maternal serum tests from 1st & 2nd trimester from various States/Territory

	SA			TAS			NT		
	1st	2nd	NTDO	1st	2nd	NTDO	1st	2nd	NTDO
2010	11816	2136	610	2171	412	28	1145	255	8
2011	12572	2015	549	2635	415	40	1292	244	20
2012	13119	1606	532	2955	339	37	1132	242	16
2013	13564	1665	473	3114	319	29	1252	231	12
2014	13173	1606	398	3297	271	26	1667	179	8
2015	13176	1185	326	2487	217	38	1737	161	13

Table 4: Summary of risks reported from external providers using results from SAMSAS

	1st trimester	2nd trimester	NTDO	Total tests performed	FMF generated risks	% of Total
2010	15474	2828	648	18950	3519	18.6%
2011	17032	2714	610	20356	3264	16.0%
2012	17889	2616	602	21107	3202	15.2%
2013	18373	2250	533	21156	3445	16.3%
2014	18188	2083	434	20705	3591	17.3%
2015	18502	1702	380	20584	3451	16.8%

We continue to report increased risk results as less than or equal to 1:250, with greater than 1:250 as not at increased risk.

The data and performance quoted are solely those of the SA Pathology SAMSAS program and do not apply to other software and testing centres.