

Nuchal Translucency (NT) Ultrasound

Since February 2009, numerous sonographers and sonologists have undergone training and certification for providing nuchal translucency ultrasound. Increasing the number of certified NT sites in BC has led to a significant improvement in access and eligibility for NT. Eligible women can have an NT ultrasound done at any of the following NT sites:

NT Sites as of January 2010		
City	Site	Booking Number
Coquitlam	MedRay Imaging (community imaging clinic)	Ph: 604-941-7611 x722 F: 604-942-4612
Kelowna	Kelowna Regional Fertility Centre	Ph: 250-861-6811 F: 250-861-6814
New Westminster	Royal Columbian Hospital in Medical Imaging area	Ph: 604-520-4132
North Vancouver	North Shore Medical Imaging	Ph: 604-987-9729 (XRAY) F: 604-984-8395
Prince Rupert	Prince Rupert Regional Hospital	F: 250-622-6178
Surrey	Surrey Memorial Hospital in Family Birthing area	Ph: 604-930-7884
Vancouver	BC Women's Hospital in Diagnostic Ambulatory Program area	Ph: 604-875-2900 (call first to book appt, then fax requisition) F: 604-875-3013
	St. Paul's Hospital in Medical Imaging area	Ph: 604-806-8161
	Greig Associates (community imaging clinic)	Ph: 604-321-6774
Victoria	Victoria General Hospital in Medical Imaging area	F: 250-381-8053
	Royal Jubilee Hospital in Medical Imaging area	
	Island Ultrasound (VIHA-operated ultrasound clinic)	

Work is in progress for the implementation of additional sites in the Interior Health Authority and Northern Health Authority.

Our website www.bcprenatalscreening.ca keeps an up-to-date list of NT sites. Visit it periodically to see what is new.

DID YOU KNOW THAT...

...You can help us ensure that we provide you and your patient the most accurate risk estimate by:

- Indicating on the lab requisition if your patient will be having an NT and if known, at what NT site. We will then make sure we have the NT data when calculating the risk.
- Using the new lab requisition form (included) and carefully answering **all** questions.

...You can order more copies of the patient pamphlets (that have now been translated in Traditional Chinese, Simplified Chinese, and Punjabi) on our website www.bcprenatalscreening.ca (click on the *For Health Care Providers* tab.)

...Our website has a "dating" tool, (found on the *Home* page) which was designed to help patients know what they are eligible for and when these tests are done. You may find this tool helpful too!

BC PRENATAL GENETIC SCREENING PROGRAM UPDATE

January 2010

In February 2009, the BC Prenatal Genetic Screening Program adopted a serum-based approach to prenatal genetic screening with nuchal translucency (NT) ultrasound added for women at higher risk of having a fetus with Down syndrome or trisomy 18 and women with multiple gestations. Since the launch of the program, we have made significant progress in the implementation of NT ultrasound sites increasing our capacity to offer Integrated Prenatal Screening (IPS) (NT plus first and second trimester serum markers) to a larger proportion of women at higher risk. In addition, our commitment to provide screening that is evidence-based means that we must adjust our approach as new evidence is published.

This newsletter will focus on:

- Changes to the *Guideline: Prenatal Screening for Down Syndrome, Trisomy 18 and Neural Tube Defects*
- Information on current utilization
- Answers to Frequently Asked Questions
- Practical points

The tables and algorithms enclosed with this newsletter **REPLACE** those distributed with the Guideline in February 2009. The updated Guideline (in PDF) reflecting these January 2010 changes is available for download on our website (www.bcprenatalscreening.ca).

CHANGES TO THE SCREENING GUIDELINE:

1. New eligibility criteria for NT ultrasound (as part of IPS):

As of January 2010, the following women qualify for NT ultrasound to be done in conjunction with first and second trimester serum markers (IPS):

- Women ≥ 36 years old at expected date of delivery (EDD);
- Women with twin pregnancies;
- Women who have a history of a previous child or fetus with Down syndrome, trisomy 18 or trisomy 13;
- Women ≥ 35 years old with 3 or more miscarriages;
- Women who are HIV positive; and
- Women pregnant following *In vitro* fertilization and intracytoplasmic sperm injection.

2. Screening in twin pregnancies:

Based on published studies, screening twin pregnancies with both NT ultrasound and serum markers reduces the false positive rate while maintaining a high detection rate as compared to NT ultrasound alone.

As of January 2010, women pregnant with twins should be screened by NT ultrasound and first and second trimester serum markers (IPS). If NT is not available, SIPS should be offered. If the patient presents after 13 weeks 6 days gestation, Quad should be offered. Women who are ≥ 35 years old have the option of amniocentesis without prior screening.

Screening in higher multiples will remain based on NT alone.

BC Prenatal Genetic Screening Program

PART OF THE BC PERINATAL HEALTH PROGRAM

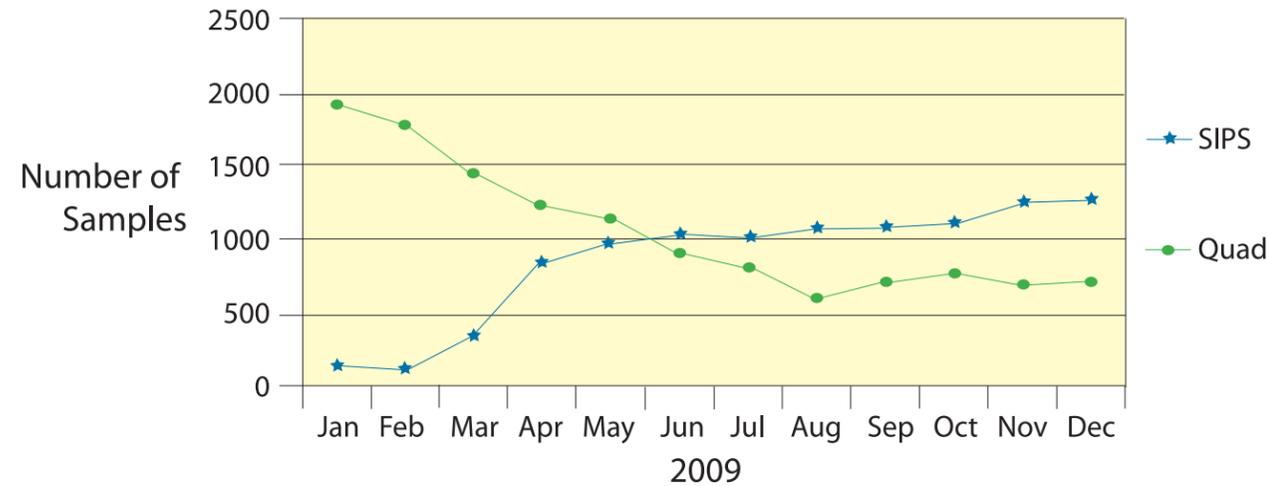
F5 - 4500 Oak Street
Vancouver, BC
Canada V6H 3N1
Tel: 604.875.3772
Fax: 604.875.3747
www.bcprenatalscreening.ca



UTILIZATION OF PRENATAL SCREENING TESTS

Since SIPS became available to all women in February 2009, the Prenatal Biochemistry Laboratory has seen a progressive increase in the utilization of SIPS with a decrease in the utilization of Quad.

This is a GOOD change in clinical practice since SIPS has a lower false positive rate than Quad.



ESTIMATED 2009 UPTAKE OF SCREENING IN SINGLETON PREGNANCIES AND TYPE OF SCREENING TESTS USED

Age	2009		Screening Test		
	number of screens performed	percent of pregnancies screened	Quad	SIPS	IPS
<35	15,557	44%	58%	41%	1%
35 – 39	5,640	70%	33%	59%	8%
40+	1,026	57%	22%	29%	49%

Amongst the 35 – 39 year old women, approximately 67% of them have SIPS or IPS and 33% have Quad. In contrast, amongst the <35 year old women, only 42% have SIPS or IPS and 58% have Quad.

We could improve screening by educating and encouraging pregnant patients to see their health care provider early in their pregnancy (before 10 weeks gestation). This will give them the option of having a screening test with a lower false positive rate (such as SIPS or IPS) and thus less chance of being offered an amniocentesis to have a definite diagnosis.

FREQUENTLY ASKED QUESTIONS

Is screening for Trisomy 18 still being done as no risk is displayed on the reports?

Yes. The program still screens for trisomy 18 but the software used does not display the risk for Trisomy 18 unless it is above the screen cut-off (1:300).

Why do the different screening tests have different risk cut-offs?

For any given screening test, a risk cut-off is set such that a desired detection rate is reached with an acceptable false positive rate. The different risk cut-offs chosen for Quad, SIPS and IPS were selected so that each test reached an equivalent detection rate. One can do that with a risk cut-off of 1:200 for IPS, 1:300 for SIPS and 1:385 for Quad. With these cut-offs, the false positive rate will be different for the different tests with the lowest false positive rate with IPS and the highest with Quad.

Can a detailed second trimester ultrasound (18 – 20 weeks) be used to modify the risk of Down syndrome assessed by the screen?

Yes. An 18 – 20 week ultrasound without any soft markers or anomalies is capable of reducing the estimated risk of trisomy 21 by approximately 50%.

How do I counsel a patient who is found on the 18 – 20 week ultrasound to carry a fetus with a choroid plexus cyst(s) (CPC)?

The finding of isolated choroid plexus cyst(s) (CPC) on ultrasound should be interpreted in conjunction with the prenatal screening (SIPS, IPS, or Quad) result or age alone if the woman is greater than 20+6 weeks gestation and Quad screen can not be offered.

The risk of trisomy 18 in the presence of isolated CPC remains low if the screening test was negative and the woman is less than 35 years old. In those situations, women should be reassured. An amniocentesis should be offered only if the CPC is seen in association with either (a) a screen positive result for Down syndrome or trisomy 18; or (b) another abnormality(ies) on ultrasound; or (c) a woman 35 years or older.

How can the test performance of IPS, SIPS, or Quad be used in counselling patients?

The performance of the tests may be useful in counselling patients *prior* to screening and help patients decide whether to have screening and which screening test they may want to have.

It helps patients understand that:

- For all screening tests, a negative test result is reassuring as the remaining risk of Down syndrome would be low.
- Screening tests have different false positive rates with the lowest rate being with IPS, followed by SIPS and Quad.

This means that if a woman has a screening test that combines first and second trimester screening (IPS or SIPS), her chance of being offered an amniocentesis for a definite diagnosis is lower than if she has a Quad test.

- All screening tests have false positive rates so having a positive screen does not mean that the baby has Down syndrome.

Why do (according to the tables provided) women with a negative test result have such a low chance (<0.1%) of having a baby with Down syndrome if the detection rates range from 76% to 97%?

The incidence of Down syndrome is 1 in 700 (range from 1 in 1,250 in 25-year old women to 1 in 94 in 40-year old women). Given this incidence, if the screening tests detect 76% to 97% of cases, as a group, the women with negative screen results are left with a chance of Down syndrome of less than 1 in 1000 (3 – 24% of their original chance depending on the age of the women and the test used). The screening tests have a very high negative predictive value –99.9%. As a group, women who screen negative have a 99.9% chance that their baby is unaffected.

In practical terms, what does the screen positive rate mean?

The screen positive rate is the chance that a test will be above the screen cut-off and the result will be reported as screen positive. This rate includes both false positives and true positives. A false positive means that the screen is positive but the baby is unaffected whereas a true positive means that the pregnancy screens positive and the baby is affected. The screen positive rate varies for the different tests and for women of different ages. However, for all tests and all age groups, the majority of women who screen positive have an unaffected pregnancy.

For example, women less than 35 years of age, who choose SIPS, have a 3.7% chance of having a screen positive result. Of those women who screen positive, 97% of them will have an unaffected pregnancy. These women have a false positive screen result.

In practical terms, once a woman has a positive screen result, should she be counselled as per the risk estimate on her report?

Yes. The risk estimate provided on her report is the best estimate of her risk for having an affected pregnancy based on the combination of her age, the results of her blood test(s) and, if available, the nuchal translucency measurement. The patient's calculated risk as indicated on her report should be used in the counselling.

For example, the 32 year old woman who had SIPS and screens positive with a risk of Down syndrome of 1 in 240 should be counselled that the best estimate of her risk is 1 in 240. This means that for 240 women with this same risk, one will have an affected baby and 239 will not.