

BC PRENATAL GENETIC SCREENING PROGRAM UPDATE

February 2014

An updated Guideline on prenatal genetic screening is now available on-line for downloading at www.bcprenatalscreening.ca. The updated tables and algorithms are enclosed.

THE REVISED GUIDELINE AND ALL RELATED SCREENING DOCUMENTS REFLECT THE FOLLOWING KEY CHANGES:

- 1) Timing of blood collection for blood test #1 (part 1 of SIPS/IPS) measuring serum PAPP-A can now occur as early as 9 weeks. This means that it is done between **9 – 13⁺⁶ weeks gestation**.
- 2) For women undergoing screening for Down syndrome, screening for open neural tube defect (ONTD) via maternal serum AFP (MSAFP) will continue as part of SIPS/IPS/Quad. However given the detection rate of ONTD is higher with a detailed ultrasound than with MSAFP, **ordering MSAFP alone for ONTD is no longer indicated** except for women with a BMI ≥ 40 , or those with limited access to a quality 18–20 week ultrasound.
- 3) **Serum analyte cut-offs for PAPP-A and hCG to predict adverse obstetrical outcome** (such as stillbirth, preterm birth < 34 weeks, HELLP syndrome, pre-eclampsia) **have been modified** to reduce the false positive rate and improve the predictive value. The cut-offs are now PAPP-A ≤ 0.15 MoM and hCG ≥ 4.0 MoM.
- 4) **Non-invasive Prenatal Testing (NIPT) is available (self-pay)** to high risk pregnant women in lieu of an amniocentesis via referral to Vancouver or Victoria medical genetics departments. Go to our website www.bcprenatalscreening.ca for more NIPT information including how to access testing.

TWO MORE KEY POINTS TO REMEMBER:

- 1) **Timing is everything.** Continue to offer prenatal genetic screening to all pregnant patients early (within the 1st trimester) so that SIPS/IPS is an option for them. Arrange for the 2nd blood collection (part 2 of SIPS/IPS) to be done early within the eligible timeframe. Aim for 15⁺²–16 weeks to ensure sufficient decision-making time for your patient if any further testing is indicated.
- 2) If patient has a 1st trimester ultrasound done, document the CRL measurement and scan date on the patient's SIPS/IPS/Quad lab requisition to support most accurate screen risk result. Alternatively, a copy of the ultrasound report can either be faxed to the C&W biochemistry lab or attached to the patient's lab requisition.

BC Prenatal Genetic Screening Program



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Go to our website www.bcprenatalscreening.ca to download or order updated versions of:

- Patient pamphlets “*Prenatal Genetic Screening – It’s your choice*”
- Screen positive info sheets “*What does it mean and what do I do now?*” (both available in multiple languages)
- C&W biochemistry lab requisition