

BC PRENATAL GENETIC SCREENING PROGRAM UPDATE

May 2019

Effective immediately, the BC Prenatal Genetic Screening Program has changed the Down syndrome positive screen cut-off, which will result in a higher detection rate.

With the increased availability of 2nd tier funded Non-Invasive Prenatal Testing (NIPT), BC data supports changing the risk cut-offs of SIPS and QUAD to 1/900. This will improve the Down syndrome detection rate from 85% to 89%. By changing the risk cut-off to 1/900, 10% of women will receive a screen positive result and have the option of NIPT. Amniocentesis will be an option for women with a positive SIPS/QUAD result with a risk greater than 1/300. The 1/200 screen cut-off for IPS remains unchanged, with a Down syndrome detection rate already greater than 95%.

HOW WILL THIS AFFECT MY PRACTICE?

Ordering of SIPS, IPS or QUAD remains the same. Only the management of patients with a positive SIPS/QUAD screen has changed.

| OLD WAY | NEW WAY |
|---|---|
| SIPS risk 1/300 or greater: Screen positive. Offer patient either funded NIPT or Amnio. | SIPS or QUAD risk 1/300 or greater: Screen positive. Offer patient either funded NIPT or Amnio. |
| QUAD risk 1/385 or greater: Screen positive. Offer patient either funded NIPT or Amnio. | SIPS or QUAD risk 1/301 – 1/900: Screen positive. Offer patient funded NIPT ONLY. Not eligible for Amnio. |

If NIPT result is positive (high risk for trisomy 13, 18, 21), patient should be referred to medical genetics for counselling and invasive diagnostic testing.

IPS ELIGIBILITY

A reminder that the following women are eligible for an NT ultrasound:

- Women 35 years or older at EDD;
- Twin pregnancies regardless of maternal age;
- Pregnant following IVF with ICSI.

For pregnant patients screened with IPS, there is no change to the IPS risk calculation or management of positive screen results.

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

- PSBC Obstetric Guideline on Prenatal Screening
- Prenatal Genetic Screening Decision Aid
- Patient pamphlets “*Prenatal Genetic Screening – It’s your choice*” (available in multiple languages)
- Screen positive info sheets “*What does it mean and what do I do now?*” (available in multiple languages)

BC Prenatal Genetic Screening Program



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