



Genetic Screening

Maternal Serum Screening

What is Maternal Serum Screening?

Maternal Serum Screening is the catch-all term used for a number of screening tests available in early pregnancy that each estimate a woman's chance of having a baby with Down syndrome, Trisomy 18 and open spina bifida.

The maternal serum screening options currently available in British Columbia include:

- Quadruple Marker Screen (QMS)
- Serum Integrated Prenatal Screen (SIPS)
- Integrated Prenatal Screen (IPS).

All three screening options are limited to specific times in pregnancy. Also, the eligibility for IPS is limited to groups of women with elevated risks. Please refer to the handout on Genetic Testing Options in BC for a complete list of the timelines and eligibility criteria.

What is involved with Maternal Serum Screening?

Quadruple Marker Screen

The quadruple marker screen is also known as the quad screen, or simply the QMS. It is a blood test that is offered to all women between 15 and 20 weeks of pregnancy.

Your blood is analyzed for levels of four different hormones associated with pregnancy: alpha-fetoprotein (AFP), unconjugated estriol (uE3), human chorionic gonadotropin (hCG) and dimeric inhibin-A (DIA). The fetus and/or the placenta play a role in the production of each of these, so they reflect the fetal condition to a certain extent.

Serum Integrated Pregnancy Screen

The Serum Integrated Pregnancy Screen has two steps:

- Step One is a blood test between 10 and 14 weeks for PAPPA. As with the QMS hormones, Pregnancy-Associated Plasma Protein (PAPPA), is associated with fetal condition.
- Step Two is a Quadruple Marker Screen between 15 and 20 weeks

Combining the risk estimates from both tests increases the accuracy of the final risk estimate. Currently in BC it is offered to ALL women.

Integrated Pregnancy Screen

The Integrated Pregnancy Screen has three parts:

- Step One is a blood test between 10 and 14 weeks for PAPPA
- Step Two is an ultrasound between 11 and 14 weeks for nuchal translucency. Nuchal translucency is the term used for the measurement of the amount of fluid behind the baby's neck. Babies with Down Syndrome tend to have a larger measurement.
- Step Three is a Quadruple Marker Screen (between 15 and 20 weeks)



Again, combining the risk estimates from all three tests increases the accuracy of the final risk estimate. Currently in BC it is only offered to women over 40, or who carry other risks:

- Women over 35 with 3 previous miscarriages
- Women carrying more than one baby
- Women with a previous child or pregnancy with Down syndrome or Trisomy 18 or Trisomy 13
- HIV positive women

Anyone who is eligible for the IPS but cannot have a nuchal translucency ultrasound is then eligible for the SIPS.

How are the results reported?

The results of each test/set of tests – also taking into account the woman’s age, race and weight, among other factors – are used to *estimate* the risk of giving birth to a baby with either Trisomy 21 (Down Syndrome), Trisomy 18 (Edward’s Syndrome) or Open Spina Bifida.

The results are sent to your midwife or doctor, usually within one to two weeks of the final step being completed. They will include a risk estimate (e.g. 1 in 300) for each of the three conditions. If your risk estimate for any one of the three is above a predefined statistical cutoff, then the test will be labeled “screen positive” and you will be eligible for follow-up testing with amniocentesis.

What can maternal serum screening tell me?

These tests are only relevant for the three conditions mentioned above: Down Syndrome, Trisomy 18 and Open Spina Bifida. Importantly, the test cannot tell you if your baby actually *has* any of the three conditions; it can only estimate the amount of risk of any one of these problems occurring. It also cannot give any information about how severe any one of these conditions might be. For example, with Down’s syndrome there is a variance from highly functional both mentally and physically, to total incompatibility with life.

There is a 1% chance of a false negative (meaning baby has one these conditions but the screen did not pick it up).

Are there any alternatives?

Depending on your age and what stage of pregnancy you are at, you may have the option of having an amniocentesis or chorionic villus sampling. These tests can provide a definitive diagnosis of the three syndromes/conditions in question, but are more invasive and include a risk of pregnancy loss or fetal limb damage.

All women are offered a routine ultrasound at 19 weeks of pregnancy. Ultrasound is used to diagnose major congenital abnormalities, environmental or genetic, including serious neural tube defects. It can also pick up indicators or “markers” of genetically inherited conditions such as Trisomy 18 and 21, but it cannot provide absolute diagnosis of these conditions.

■ Please refer to the handout on Genetic Testing Options in BC for timelines and eligibility.

Key points in making your decision

Of course everyone is looking for the reassurance of a normal result, but since these are all screening tests, not diagnostic, there is a deliberately high false-positive result (meaning baby is said to have a problem but is in fact healthy) in order to miss very few babies that do have one of these conditions.



Your decision about serum screening will be best informed by looking at a number of factors.

- If the result indicated an elevated risk, would you proceed to amniocentesis even though it carries a risk of pregnancy loss? If not, this test may not be for you.
- If you did further testing that confirmed a congenital abnormality, would you consider termination?
- If further testing confirmed abnormality, would you want to join support groups or get counseling to prepare for your baby? Would anything about your birth plans change if you knew your baby may require special care at birth?
- Would you be satisfied by just a detailed ultrasound, one which would pick up any major congenital abnormalities? If so, you may want to proceed directly to ultrasound.
- Importantly, if the initial result indicated elevated risk but further testing indicated no abnormalities, would you still continue to worry through your whole pregnancy?

Remember, no amount of genetic screening or testing can guarantee a normal baby.

Please refer to the website of the BC Prenatal Genetic Screening Program for more details, including age-related risks, detection rates and false positive incidence:

www.bcprenatalscreening.ca