What is Maternal Serum Screening?

The Maternal Serum Screen (MSS) is an optional blood test that is available to all pregnant women in British Columbia. This test, which poses no risk to the pregnancy, estimates a woman’s chance of having a baby with Down syndrome (Trisomy 21), Open Neural Tube Defects (e.g., Spina Bifida) and Edwards syndrome (Trisomy 18). It is very important to understand that this screening test is NOT a diagnostic test; it can only indicate if a pregnancy is at a greater or lesser risk of the above conditions. Women who are found to be at increased risk are offered further diagnostic testing such as detailed ultrasound examination and/or amniocentesis.

Not all affected pregnancies are detected by the MSS. The screen can detect approximately 79% of cases of Down syndrome, 85% of Open spina bifida and 60% of Trisomy 18.

How and when is the test done?
The MSS is two blood tests that can ONLY be done at specific times in pregnancy: the first between 10 and 14 weeks of pregnancy, and the second between 15 and 21 weeks of pregnancy. If you have missed the first blood test, you can still do the second, but the results will not be as accurate. The test measures the levels of several proteins in your blood, such as PAPP-A, alpha-fetoprotein (AFP), unconjugated estriol (uE3), and human chorionic gonadotropin (hCG). These are made by the baby or the placenta, and are measured in the expectant mother’s blood. The levels of these proteins are different in some pregnancies affected with Down syndrome, neural tube defects, or Trisomy 18. These protein levels in combination with maternal age, weight, and other factors are used to estimate the risk in each pregnancy.

What is a Nuchal Translucency Ultrasound?
A nuchal translucency ultrasound provides a measurement of the skin on the back of the fetus’s neck. This measurement, when combined with the blood test results, lowers the false positive rate of the test. This measurement must be taken between 10 and 14 weeks of pregnancy. Nuchal translucency ultrasounds are covered by MSP for the following women:

a) Women ≥ 35 year old at expected date of delivery (EDD);

b) Women with twin pregnancies;

c) Women who have a history of a previous child or fetus with Down syndrome, trisomy 18 or trisomy 13;

d) Women who are HIV positive; and

e) Women pregnant following in vitro fertilization with intracytoplasmic sperm injection (IVF with ICSI)

Women who are not covered by MSP for a nuchal translucency ultrasound may choose to pay to have one done. Visit the Early Pregnancy Assessment Unit’s website for more information about this option: www.epau.ca, or ask your midwife for more information.

What does a ‘negative’ result mean?
Negative results (‘below the screen cut off’) are reassuring. These results indicate that the baby’s chance of having Down syndrome, Open Neural Tube Defects or Trisomy 18 is low. A negative result is correct 99.9% of the time, however, the chance that the baby is affected is not zero. In this situation, further diagnostic testing by amniocentesis would not be offered unless a woman is already eligible for amniocentesis based on age alone (age 40 or over). This is because the chance that the baby is affected is lower than the risk of pregnancy loss associated with the amniocentesis procedure itself.

What comes after a ‘positive’ result?
About 8-10% of all women who have MSS will have a ‘positive’ result, or, in other words, a result which is above the screen cut off. Even with a ‘positive’ screening result, most fetuses do not have a problem. It simply means that the chance for Down syndrome, neural tube defects, or Trisomy 18 is increased to a level which is sufficient to offer further testing (detailed ultrasound and/or amniocentesis). The vast majority of women who have a positive MSS result have healthy, unaffected babies.

How and when are results available?
Results are usually available 7-10 days after the second blood test (16-20w of pregnancy). All results above the screen cut off (positive) are telephoned to the midwife’s office for discussion. Accurate pregnancy dating is essential for this test, so a dating ultrasound is sometimes recommended before further follow-up. Options for follow up include genetic counselling to review the results and discuss further testing options and diagnostic testing (such as detailed ultrasound and/or amniocentesis). Results below the screen cut off are sent by mail to the midwifery office and the result given to you as soon as possible.
Maternal Serum Screening (MSS) – Key points!

1. The test is offered to all women
2. The blood tests are performed at 10-14 and 15-21 weeks of pregnancy
3. A Nuchal Translucency ultrasound is available at 10-14 weeks of pregnancy for women 35 and older.
4. Maternal blood samples are used for the test
5. If the screen result is ‘positive,’ follow up options include genetic counselling and further testing by detailed U/S and amniocentesis
6. If dates are uncertain, a dating U/S performed early in the pregnancy can reduce false positives
7. Women who will be 40 years of age at delivery can forego maternal serum screening as they can choose to go directly for amniocentesis if they wish

Advantages of MSS

1. A negative result may offer some reassurance
2. It may help in decision making. For example, if an Open Neural Tube Defect is detected before birth, heath care professionals can provide parents with information and support. They can plan for delivery in a specially equipped medical centre so that the baby can have any necessary surgery or treatment soon after birth.

Limitations

1. MSS only screens for 3 things (Down syndrome, Trisomy 18, and neural tube defects only) and does not pick up every baby with those conditions
2. Results take about 7-10 days to come back. This is an anxious time for many parents.
3. About 8-10% of all women get ‘positive’ results, however only 2-4% of all positives are true positives. Most women with a ‘screen positive’ result have normal babies.
4. A negative result does not guarantee that everything is all right with the baby.
5. Even if disability is confirmed by a diagnostic test (amniocentesis), it does not measure the degree to which the baby is affected.
6. Advanced maternal age also increases the risk for chromosomal abnormalities other than Down’s syndrome. There is no way of screening for all of these conditions.

Important questions to consider before choosing to have this screening test

(See Appendices 1 & 2)

1. How do you feel about having a baby with a disability such as Down Syndrome, Spina Bifida or Trisomy 18?
2. Would you want the type of information that this screening test can provide?
3. If the result is ‘above the screen cut-off’ (otherwise ‘positive’), amniocentesis is an option. This test has a miscarriage risk of about 1 in 200 (0.5 %). Would this be an acceptable option for you?
4. Consider whether knowing about a birth defect would change your medical, birthing or parenting plans. Would you consider termination if your baby had one of these problems?

What is Down Syndrome? (Trisomy 21)

Down syndrome is a condition caused by the presence of an extra chromosome. It is one of the most common genetic birth defects, affecting approximately 1 in 800 to 1000 babies. It can be hereditary or accidental. Down syndrome is a common cause of learning difficulty and is often associated characteristic facial features and certain medical problems. About 40-50% of affected individuals have congenital heart problems, and there is risk of ophthamal (eye) and hearing impairment as well as thyroid and gastrointestinal problems. Life expectancy is sometimes reduced, however, many adults with this condition are now living into their 50’s and 60’s. The degree of learning difficulties is variable. There is no way to predict how serious any of the disabilities will be.

Children with Down syndrome usually can do most things that any young child can do, such as walking, talking, dressing and being toilet-trained. However, they generally start learning these things later than other children. The outlook for these children is far brighter than it once was. Today, an increasing number of adults with Down syndrome live semi-independently in community group homes where they take care of themselves, participate in household chores, develop friendships, partake in leisure activities and work in their communities.
Any woman at any age can have a baby with Down syndrome. However, the chance is higher the older the woman is. A woman who is 40 has a much higher chance of having a baby with Down syndrome than a woman who is 20. The father’s age may also affect the chance that the baby has Down’s syndrome, though research in this area is still ongoing. Because most women have their babies between 20 and 35, most babies with Down syndrome are born to younger women.

**What is Trisomy 18? (Edward’s Syndrome)**

This is a rare disorder caused by an extra chromosome at the 18th pair. Like trisomy 21 (Down syndrome), trisomy 18 affects all systems of the body and causes distinct facial features. It is estimated to occur in 1 in 6,000 – 8,000 live births. Most babies (95%) die before or within one hour of birth. Babies with this condition have severe mental disability and major physical problems and require skilled medical care. Infants have a 5% chance of surviving to age 1 year.

**What is Spina Bifida?**

This occurs when there is an opening in the spine which can cause nerve damage. Spina Bifida affects 1 in every 2,000 live births each year in the US. It occurs more frequently among Hispanics and people of European descent and less commonly among Ashkenazi Jews, Asians and African-Americans.

Spina bifida varies in severity and in the level of disability. There is no way to predict exactly how serious these problems will be. This can range from individuals that lead full and active lives to individuals with physical disability which can include weakness or paralysis of the legs, incontinence, and hydrocephaly (fluid on the brain) which can cause mental developmental delays.

With treatment, children with spina bifida can usually become active individuals. At least 70% of children with spina bifida have normal intelligence, although some do have learning difficulties. A baby with the most severe form of spina bifida usually requires surgery within 24 to 48 hours after birth to tuck exposed nerves and spinal cord back inside the spinal canal and cover them with muscle and skin. Prompt surgery helps prevent additional nerve damage from infection or trauma.

There appears to be a hereditary factor and an increased risk for women who have had a previously affected pregnancy. However, 95% of babies born with spina bifida and other neural tube defects are born to parents with no family history of these disorders. There is also a possible increased risk for women who take anti-epileptic medications. The chance does not depend on the age of the mother. Ensuring adequate folic acid intake before and during early pregnancy is hugely protective.
Understanding Prenatal Screening: a Visual Aid for Patients

This visual aid illustrates 5000 pregnant women who chose Down Syndrome screening.

- The majority of women will **Screen Negative** and have a normal, healthy baby. (true negative)
- Very rarely a woman will **Screen Negative** but will have a baby with Down Syndrome. (false negative)
- A small percentage of women will **Screen Positive** but the majority will have normal, healthy babies. (false positive)
- A small percentage of women will **Screen Positive** and have a baby with Down Syndrome. (true positive)

Statistics based on singleton pregnancies choosing SBPS.

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