BC Prenatal Genetic Screening Program

An aid to decision-making

Should I take the SIPS / IPS test to screen for Trisomy 21 (Down syndrome)?

❖ Who might think about being tested?

All pregnant women can have this test. The risk of Trisomy 21 increases with a woman's age.

AGE RELATED RISK OF ANOMALIES IN FETUS AT BIRTH			
Mother's Age	**Chance of Trisomy 21 (Down Syndrome)**	**Chance of Trisomy 18**	**Chance of Neural Tube Defect**
25	1 in 2,500	1 in 25,000	1 in 1,000 for all ages
30	1 in 840	1 in 8,400	
35	1 in 356	1 in 3,560	
38	1 in 166	1 in 1,066	
40	1 in 94	1 in 940	

❖ A decision to make

- **Doing and not doing the test are both good choices. Making the decision might be easier if you:**
  - ✔ Base your decision on the best scientific information available
  - ✔ Base your decision on your values and preferences
  - ✔ Share your thoughts with your doctor / midwife and your family

❖ Information to help you make the decision

- **What is Trisomy 21 (T21) or Down Syndrome?**
  - ✔ It is caused by an extra copy of chromosome 21 which affects how the baby develops and grows.
  - ✔ People with T21 have almond-shaped eyes, a round face, poor muscle tone, greater risk of vision and hearing problems, heart, stomach and bowel defects, and intellectual disabilities that can be mild or moderate.
  - ✔ 60% of children with T21 need specialized home care.
  - ✔ Some adults with T21 have jobs and are almost completely independent.
  - ✔ People with T21 can have meaningful emotional relationships and lead lives that are fulfilling for themselves and their family and friends. They usually live into their 50's.
- **What is Trisomy 18 (T18)?**
  - ✔ It is caused by having an extra copy of chromosome 18.
  - ✔ Many pregnancies with T18 will miscarry.
  - ✔ Babies that are born with T18 rarely live more than a few days or months because of serious heart and brain defects and poor growth before and after birth.

- **What is a Neural Tube Defect?**
  - ✔ An open neural tube defect (NTD) occurs when the brain or spinal cord does not form properly.
  - ✔ Spina bifida is a NTD in which the spine does not completely close. People with spina bifida may have both physical and mental disabilities.
  - ✔ Anencephaly is an open NTD involving the brain. A baby with anencephaly will be stillborn or die shortly after birth.
- **What is the Serum Integrated Prenatal Screening test (SIPS) and Integrated Prenatal Screening test (IPS)?**
  - ✔ **SIPS** is two blood samples taken:
    1st between 9 weeks and the end of the 13th week
    2nd between 14 weeks and the end of the 20th week (preferably between 15 and 16 weeks)
  - ✔ **IPS** is the SIPS test along with a special ultrasound of the neck folds of the fetus. This test is offered to women age 35 years or older at the time of delivery, and women carrying twins.
  - ✔ The result of SIPS/IPS is available about 10 days after the second blood test.
- **What is the SIPS/IPS test for?**
  - ✔ This test tells you if you have a higher chance of carrying a fetus with T21, T18, or a NTD.
  - ✔ If the chance is high for either T21 or T18, your doctor/midwife will offer you NIPT (covered by MSP). NIPT is another (blood sample) screening test for T21 and T18 that has a higher accuracy than SIPS / IPS. Depending on the level of risk indicated on your SIPS/IPS screen result, you may also have the option of amniocentesis. Amniocentesis is an invasive diagnostic test that will tell you for sure if you are carrying a fetus with T21 or T18.
  - ✔ This information can help you decide whether to prepare for a child with special needs or consider ending the pregnancy.
- **What other options are available for me on my BC medical plan?**
  - ✔ If you are 40 years or older on the due date, you can choose to have an amniocentesis first without having the SIPS/IPS test.

    An amniocentesis is a diagnostic test that checks the chromosomes of fetuses that are at higher risk of an abnormality. A small sample of the liquid around the fetus is taken using a needle inserted through the mother's abdomen while watching with an ultrasound. This procedure is associated with a risk of 1 in 200 of losing the pregnancy.
- **What private pay screening options might be available?**
  - ✔ A First Trimester Screening Test (FTS) is an option that consists of one blood test and a special ultrasound, both taken around 11 weeks. The results are available the same day or within a few days. This test costs about $500.
  - ✔ A Non-Invasive Prenatal Test (NIPT) is a single blood test taken anytime after 10 weeks. The result is available in 10 days and is highly accurate for T21 and T18. NIPT is covered by MSP only for: women at higher risk for T21/T18 based on SIPS/IPS results, a history of a previous pregnancy with trisomy 21, trisomy 18 or trisomy 13, or ultrasound findings. Other women who choose NIPT as their screening test without doing the SIPS/IPS test must cover the cost, which varies depending on the commercial test used.
  - ✔ Neither FTS nor NIPT screen for a neural tube defect. If you chose one of these tests, screening for neural tube defect will be done by your detailed ultrasound at 19–20 weeks gestation.
- **SIPS, IPS, and the private pay tests (FTS and NIPT) are all screening tests that will tell you your chance of carrying a fetus with T21 or T18. *Only an amniocentesis test can tell you for sure.***

Doing or not doing the SIPS / IPS Test *(follow along with the visual aid diagram)*

Although the SIPS/IPS test can detect a pregnancy at increased risk of T18, most cases will also be detected by ultrasound. For these reasons, the benefits and harms of doing or not doing SIPS/IPS test will focus on screening for T21.

DOING the test

Benefits	Harms
❑ **Know your chances of carrying a fetus with T21** Out of 5,000 women screened, 500 have a test result that says they are at higher risk for carrying a fetus with T21. If these 500 women have NIPT or an amniocentesis to know for sure, only 13 would actually be carrying a fetus with T21. ❑ **Prepare to end the pregnancy** Some women who know they are carrying a fetus with T21 will choose to end the pregnancy. ❑ **Prepare for a child with T21** Some women who know they are carrying a fetus with T21 will choose to continue the pregnancy and can prepare for a child with T21 or may consider an adoption plan. ❑ **Reassurance** Out of 5,000 women who take the test, 4,500 have a result that means they are at low risk for carrying a fetus with T21. These women are reassured.	❑ **Anxiety while waiting for results** Women waiting for test results have anxiety levels 10 times higher than normal. ❑ **False Alarm** Out of the 500 women whose test results show they are at increased risk of carrying a fetus with T21, 487 are actually NOT carrying a fetus with T21. Many of these women will experience anxiety. ❑ **May have to face difficult decisions** 500 women whose test results show they are at increased risk of carrying a fetus with T21 will need to decide about having further testing (NIPT or amniocentesis). Those who have testing and are shown to actually have a fetus with T21 will need to make a decision about whether to continue or end the pregnancy. ❑ **False Reassurance** Of the 4,500 women whose test results show they are at low risk for carrying a fetus with T21, 2 will actually be carrying a fetus with T21. These 2 women are falsely reassured.

NOT DOING the test

Benefits	Harms
❑ **Avoid anxiety and unnecessary extra testing** ❑ **Stay true to your personal convictions and values** For some women, not doing the test is the right choice for their personal or family's convictions. ❑ **Avoid difficult decisions** Not doing the test can avoid the anxiety and stress of making a decision about continuing or ending the pregnancy if the fetus has T21.	❑ **Not knowing your risk of carrying a fetus with T21** Out of 5,000 women who do not take the test, 15 women are carrying a fetus with T21. These women cannot prepare for giving birth to a baby with T21. ❑ **Anxiety from not knowing** Women who don't take the test may be anxious because they don't know if their child will have T21 or not. ❑ **Possible social pressure to do the test**

❖ *Discussion with Your Care Provider*

- ❍ **What is your chance of having a baby born with T21, T18 and Neural Tube Defect based on your age? Check the table on page 1 to know your risks.**
- ❍ **Check your understanding of:**
  - ✔ What are the tests for
  - ✔ How and when you get results
  - ✔ Options for further testing if your screen result shows a high risk
  - ✔ Private pay options
  - ✔ Benefits and harms of the tests

❖ *What are the benefits and harms that matter most to you?*

DOING the test	NOT DOING the test
Benefits	***Benefits***
Harms	***Harms***

❖ *What is your decision?*

❑ **Do the test** ❑ **Don't do the test** ❑ **I don't know**

❖ *Are you comfortable with this decision?*

		Yes	No
Sure of myself	1) Do you feel sure about the best choice for you?	❑	❑
Understand information	2) Do you know the benefits and harms of doing or not doing the test?	❑	❑
Risks and Benefits	3) Are you clear about which benefits and harms matter most to you?	❑	❑
Encouragement	4) Do you have enough support and advice to make a choice?	❑	❑

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References

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