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 15 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. Another option is having an amniocentesis test which 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 or ultrasound findings. Women who choose NIPT first without the SIPS/IPS test must cover the cost, which is in the order of $500–$650 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, 275 have a test result that says they are at higher risk for carrying a fetus with T21. If these 275 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, 4,725 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 275 women whose test results show they are at increased risk of carrying a fetus with T21, 262 are actually NOT carrying a fetus with T21. Many of these women will experience anxiety. ❑ **May have to face difficult decisions** 275 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 4,725 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?	❑	❑

References

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No conflict of interest: Neither the granting agency, the authors, nor their affiliated organizations have any interests at stake in the decisions made by patients after using this decision aid.