South Community
BIRTH PROGRAM

GENETIC SCREENING EDUCATION
(please read before your first appointment)

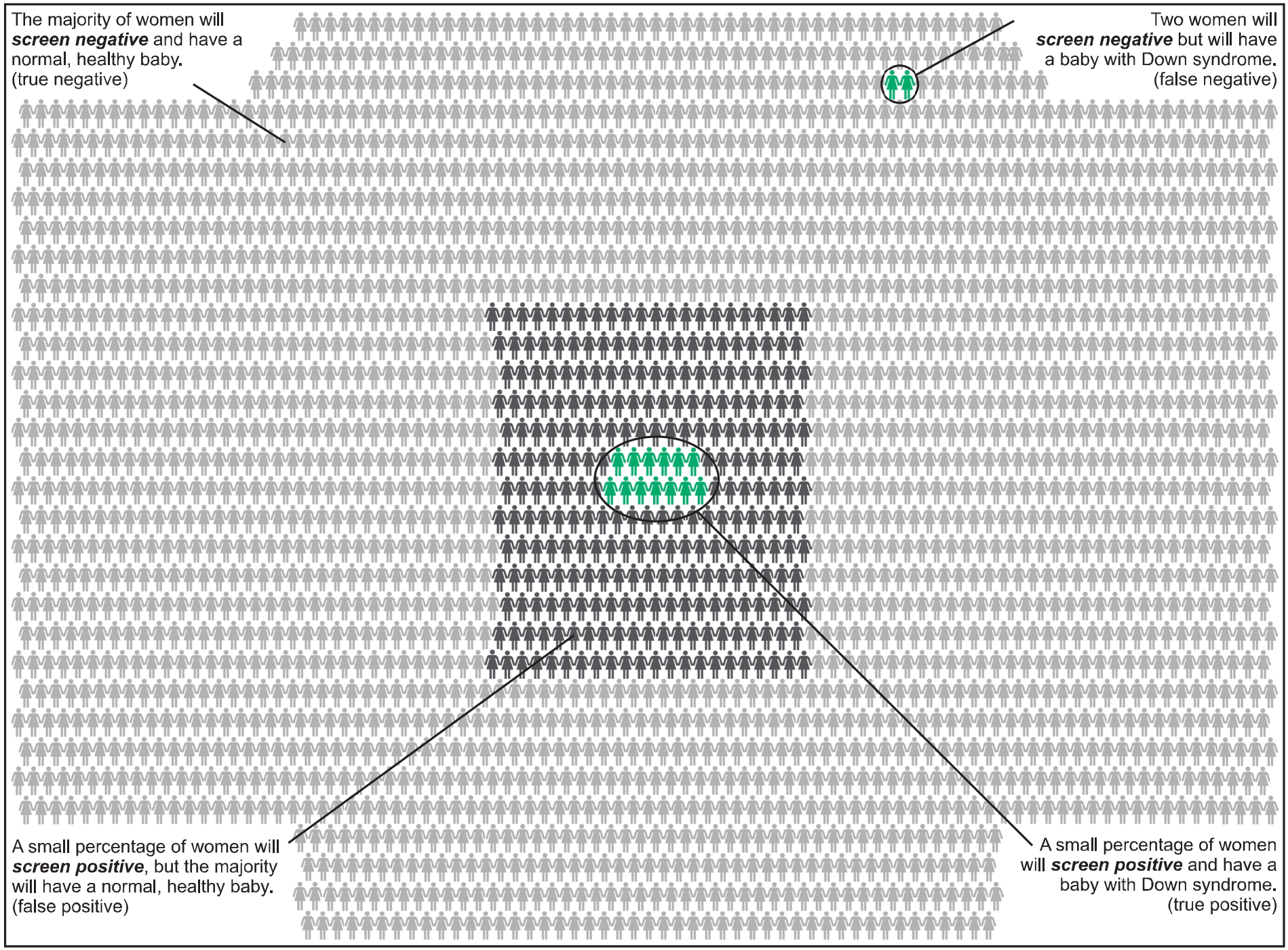
To learn more about genetic screening please click the following link to watch a video from the Perinatal Services of British Columbia.

CLICK FOR VIDEO HERE: https://www.youtube.com/watch?v=z39_COwX-H4&feature=youtu.be

The additional pages of this document will go through your options for genetic screening and your medical provider will discuss this with you at your first appointment.

Understanding Prenatal Screening: A Visual Aid for Patients

This visual aid illustrates 5,000 pregnant women who chose Down Syndrome screening.



Statistics based on singleton pregnancies choosing screening in BC.

Prenatal Genetic Screening

It's *your* choice

Learn more about the options available to you

BC Prenatal Genetic Screening Program

Perinatal Services BC
Provincial Health Services Authority

Provincial Health Services Authority
Province-wide solutions. Better health.

www.bcprenatalscreening.ca

What is prenatal genetic screening?

It is a blood test available to all pregnant women in British Columbia. This screening tells you the chance of your baby having Down syndrome, trisomy 18 or an open neural tube defect.

What are Down syndrome, trisomy 18 and open neural tube defects?

Down syndrome happens when a baby has an extra chromosome. Chromosomes tell our bodies how to grow and develop. When there is an extra chromosome there is too much information. This changes the way the body grows and develops. People with Down syndrome have mild to moderate intellectual delays. They also have a higher chance of some health problems. There is no way to know how serious the problems will be. People with Down syndrome usually live into their 50s.

Trisomy 18 also happens when a baby has an extra chromosome. Many pregnancies with trisomy 18 miscarry. If the baby is born, he or she rarely lives past the first few days or weeks. These babies have serious heart and brain defects.

Open neural tube defects happen when the brain or spinal cord does not form properly. When an open neural tube defect involves the spinal cord, it is called spina bifida. It can result in both physical and mental disabilities. Life expectancy depends on how serious the condition is. An open neural tube defect involving the brain is called anencephaly. Babies with anencephaly will be stillborn or die shortly after birth.

What are the chances I will have a baby with one of these conditions?

The chance of having a baby with Down syndrome is about 1 in 700 and the chance of having a baby with trisomy 18 is about 1 in 7,000. These numbers are averages for women of all ages. In fact, the chance of having a baby with Down syndrome or trisomy 18 is lower in younger women and higher in older women.

Although most babies are born healthy, all women have a chance of having a baby with Down syndrome, trisomy 18 or an open neural tube defect – even if they and their families are healthy.

Mother's age (years)	Chance of Down syndrome	Chance of trisomy 18
25	1 in 1,250	1 in 12,500
30	1 in 840	1 in 8,400
35	1 in 356	1 in 3,560
40	1 in 94	1 in 940
45	1 in 24	1 in 240

If you or your partner has had a baby with Down syndrome or another chromosome condition, your chance in another pregnancy is increased.

The chance of having a baby with an open neural tube defect is the same no matter what your age – about 1 in 1,000.

Prenatal screening will tell you your chance of having a baby with Down syndrome, trisomy 18 or an open neural tube defect in this pregnancy.

It is your choice whether to have prenatal genetic screening.

The earlier you see your health care provider, the more options you have.

How, when and where is prenatal genetic screening done?

Two blood tests are taken at your local lab:

- **Blood test #1:** between 9 and just under 14 weeks of pregnancy
- **Blood test #2:** between 14 and just under 21 weeks of pregnancy (best to have test #2 done as early as possible – ideally before 16 weeks)

If one misses the first blood test, one may still have the second blood test. However, it is best to have both blood tests when possible. Having both improves the accuracy of the screen result.

Results of screening are available within ten days after the second blood test.

If you have an increased chance of having a baby with Down syndrome or trisomy 18 due to your age, you will be offered a special ultrasound. The ultrasound would be in addition to the blood tests. The ultrasound measures the fluid space at the back of your baby's neck. It is called a **nuchal translucency or NT** ultrasound. The NT is done

Talk to your health care provider about your screening options.

Whatever you choose, it will not affect your care. If screening is not right for you, please tell your health care provider.

between 11 and just under 14 weeks of pregnancy. Although adding the NT gives more information for the screen result, the blood tests described on page 3 are very good screens on their own.

What if I have had a previous pregnancy with Down syndrome or trisomy 18 or 13?

You will have the option of screening with NIPT.

Skip to page 6 for more information.

What if I will be 40 or over when my baby is born?

You will have the option of screening with the two blood tests and NT ultrasound, as just described. You will also be offered the choice to have diagnostic testing that tells you for sure if your baby has Down syndrome or trisomy 18. Types of diagnostic testing are chorionic villi sampling or amniocentesis. You may also decide not to have prenatal genetic screening or diagnostic testing.

What if I am pregnant with twins?

If you are less than 14 weeks pregnant, you will be offered both an NT ultrasound and the prenatal screening blood tests described on page 3. If an NT ultrasound is not available, or you are more than 14 weeks pregnant, you will still be offered the blood test(s) described on page 3. If you will be 35 or older when your babies are born, you will have the option of amniocentesis.

What happens after I have had the blood tests?

The result of the prenatal screening will most likely show your chance of having a baby with one of these conditions is low. This is called a **"screen negative"** result. This result is correct over 99.9% of the time but it does not mean your chance of having a baby with one of these conditions is zero.

If the result shows your chance of having a baby with one of these conditions is high enough, you will receive a **"screen positive"** result. This prenatal screen result does not mean your baby has the

condition for sure. In fact most women with this result do not have a baby with one of these conditions. More testing will be offered to you to give you a definite answer.

Most women have prenatal genetic screening results showing chances are low for these conditions.

Although 1 in 10 women will have a screen positive result, most of these women will not have a baby with Down syndrome, trisomy 18 or an open neural tube defect.

The chance of having a screen positive result increases as a woman ages.

What test will I be offered if I have a screen positive result for an open neural tube defect?

You will be offered a detailed ultrasound. You will also be offered an appointment with a maternal fetal medicine doctor or a genetic counsellor at one of BC's medical genetics clinics (Vancouver or Victoria). If your baby has an open neural tube defect, this is usually seen on the ultrasound scan.

What test will I be offered if I have a screen positive result for Down syndrome or trisomy 18?

You will be offered another screening blood test called NIPT (Non-Invasive Prenatal Testing). You may also have the option of a diagnostic test called amniocentesis, depending on the level of risk indicated by your positive screen result. Some women will choose to have additional testing, some will not. It is your choice.

While prenatal genetic screening tells you the chance of your baby having Down syndrome or trisomy 18, you would need a diagnostic test, such as an amniocentesis, to find out for sure.

What is NIPT (Non-Invasive Prenatal Testing)?

It is a safe and highly accurate screening test for Down syndrome and trisomy 18 that is done through a blood test. It detects almost all babies with Down syndrome and trisomy 18. This means that if the test is negative, the chance of Down syndrome or trisomy 18 is extremely small. If the test is positive, the chance is high. An amniocentesis would then be offered to confirm the positive NIPT. The NIPT test result is available in 10 days. For women who receive a positive (IPS/SIPS/Quad) screen result for Down syndrome or trisomy 18, this NIPT test is covered by the provincial medical plan.

What is an amniocentesis?

It is a diagnostic test which tells you if your baby truly has one of these conditions. A small amount of fluid is taken from around your baby by putting a very fine needle into your belly. About three teaspoons are taken. The needle is guided by ultrasound so it does not touch the baby. This fluid sample is looked at to find out whether or not the baby has Down syndrome, trisomy 18 or another chromosome condition. Amniocentesis has a 1 in 200 (0.5%) chance of pregnancy loss.

What if the further testing confirms that my baby has one of these conditions?

If the testing confirms your baby has Down syndrome, trisomy 18 or an open neural tube defect, there are people you can talk to who will help you. Your health care provider, as well as medical geneticists or genetic counsellors, are there to discuss your choices with you and help you make a decision that is right for you. Your choices include continuing the pregnancy, ending the pregnancy, or making an adoption plan.

Making a decision

Is prenatal genetic screening right for me?

Many women find it difficult to decide whether or not to have prenatal genetic screening. Here are some questions to think about that may help you decide.

- Do I want to know if my baby has Down syndrome, trisomy 18 or an open neural tube defect before the baby is born?
- What would I do if my diagnostic test result showed my baby had one of these conditions? Would I end the pregnancy? Would I want to know so that I could prepare for a child with special needs? Would I consider an adoption plan for the baby?
- How will this information affect my feelings throughout the pregnancy? Would having a screen positive result cause me too much worry?

Points to keep in mind

- Most women have a prenatal genetic screening result showing chances are low for these conditions.
- Although some will have a screen positive result, most of these women will not have a baby with Down syndrome, trisomy 18 or open neural tube defect.
- Prenatal screening detects most babies with Down syndrome, trisomy 18 or an open neural tube defect, but not all.
- Sometimes prenatal screening may detect other medical conditions in your baby.
- It is important to remember that no test detects every type of physical or mental condition.
- Talk to your health care provider if you need more information to help make your decision.

More information about prenatal genetic screening is available on our website *www.bcprenatalscreening.ca*

If you have questions or need more information, please talk to your health care provider.

What else might you like to know?

The BC Prenatal Genetic Screening Program is part of Perinatal Services BC, an agency within the Provincial Health Services Authority (PHSA). The BC Prenatal Genetic Screening Program operates across several facilities in the province. While analysis of the initial blood tests takes place at the laboratory at the Children's and Women's Health Centre of BC, further diagnostic testing, if required, takes place at other facilities in BC. Regardless of the point of collection, prenatal genetic screening information is provided to the BC Prenatal Genetic Screening Program and, in combination with other information received, is used to provide safer, more accurate tests, measure outcomes and evaluate and disseminate new evidence/knowledge.

We are committed to protecting the privacy of personal information

For women choosing to have prenatal genetic screening, it is important to know that the BC Prenatal Genetic Screening Program collects, uses and discloses personal information only as authorized under section 26 (c), 33 and 35 of the BC Freedom of Information and Protection of Privacy Act, other legislation and PHSA's Privacy and Confidentiality Policy. We respect your right to personal privacy and take all reasonable steps to make sure that personal information is treated confidentially, is only used and further disclosed for the purposes described above and is securely stored. Reports generated from the information collected are always in summarized form and do not include names or other identifying information. Should you have any questions regarding the collection, use or disclosure of your personal information, please contact the Privacy Advisor for Perinatal Services BC (PSBC) at 604-877-2121.

BC Prenatal Genetic Screening Program

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April 2019

Prenatal Genetic Screening

It is your choice whether or not to have prenatal screening and/or a diagnostic test.

Genetic screening is offered to all women in pregnancy. Depending on your age and other risk factors, different tests may be offered to you by your midwife or doctor.

The Conditions

The following conditions are screened for:

Down Syndrome or Trisomy 21

People with Down Syndrome usually have mild to moderate intellectual delay. Individuals with Down syndrome may have a greater incidence of health conditions than the average person such as heart, stomach, bowel, thyroid, vision and hearing problems. Treatment is available for many of these conditions. Each person with Down syndrome is different. There is no way to test how serious the disabilities will be. Today, many people with Down syndrome generally live to 50-60. In general, about 1 in 1000 babies born will have Down Syndrome. The chance of having a child with Down Syndrome increases with the mother's age.

Trisomy 18

Babies with trisomy 18 have both mental and physical disabilities. Many pregnancies with Trisomy 18 will end in a miscarriage. Many babies born with Trisomy 18 do not survive past the first few months of life. In general about 1 in 6,000 babies born will have Trisomy 18. The chance of having a child with Trisomy 18 increases with the mother's age.

Open Neural Tube Defects

These conditions occur when the brain or spinal cord does not form properly. An open neural tube defect (OTD) involving the spinal cord is called Spina Bifida. Spina Bifida causes physical disabilities such as difficulty walking, and controlling the bladder and/or bowel. People with Spina Bifida may also have mental disabilities. Treatment can help with many of the physical disabilities. An OTD defect involving the brain is called anencephaly. A baby born with anencephaly will die shortly after the birth. In Canada, the chance of having a baby with an open neural tube defect is about 1 in 2,000 births. The chance of a child being born with this condition does not increase with the mother's age.

Reasons you may choose to have a screening test

• Some people have screening because they would like the information before their baby is born and would like to prepare for a child with special needs. You can go to support groups where you can talk to other parents to learn how having a baby with one of these conditions can affect your family's life and find out what help is available to you.

• Some people have screening because they would consider giving the baby up for adoption if their baby was found to have one of these conditions.

• Some people have screening because they would end the pregnancy if their baby was found to have one of these conditions.

The fine print

• **There is no cure** for Down syndrome, Trisomy 18 or Open Neural Tube defect.

Every pregnancy has 2 or 3 chances in 100 (2-3%) of having a condition that is not found by prenatal screening tests.[1] Genetic testing is not available for every condition and not all health conditions are genetic. So a "normal" screening result does not guarantee that a child will be healthy.

Reasons you may choose NOT to have a screening test

Some people prefer not worry about a problem until after the baby is born.

Some people prefer not to risk having a false positive test that might cause more worry.

Remember most babies are born healthy

[1] A guide to understanding prenatal screening tests - for women and their families (2007)

Screening Tests Available

Depending on how far along you are in your pregnancy, as well as your risk factors, your midwife or doctor will offer the best test for you.

The following are the test that are covered by MSP (no cost to you) now:

Serum Integrated Prenatal Screen (SIPS): Two blood tests, one at 9-13 weeks and the second test at 15-20 weeks. *Note: results are not given until after the second blood test is done*

Integrated Prenatal Screen (IPS): The same as SIPS with the addition of a Nuchal Translucency (NT) ultrasound of the baby, which looks at the thickness of the neck folds. This additional test is only available if you are over the age of 35 in Vancouver.

Quad Screen (Quad): One blood test only taken between 15-20 weeks, if the first blood test for SIPS is missed.

Test results are given as a percent chance that your baby has one of the conditions described above. The results will be sent to your midwife or doctor, who will then contact you

Private Genetic Screening Tests

First Trimester Screen (FTS): Is a blood test and ultrasound done around 11 weeks. The ultrasound looks at nuchal transluscency as in IPS plus other measurements. A risk is given of having a baby affected. Results of FTS are available the same day or within a few days of the test. The cost of private FTS is approximately $500.

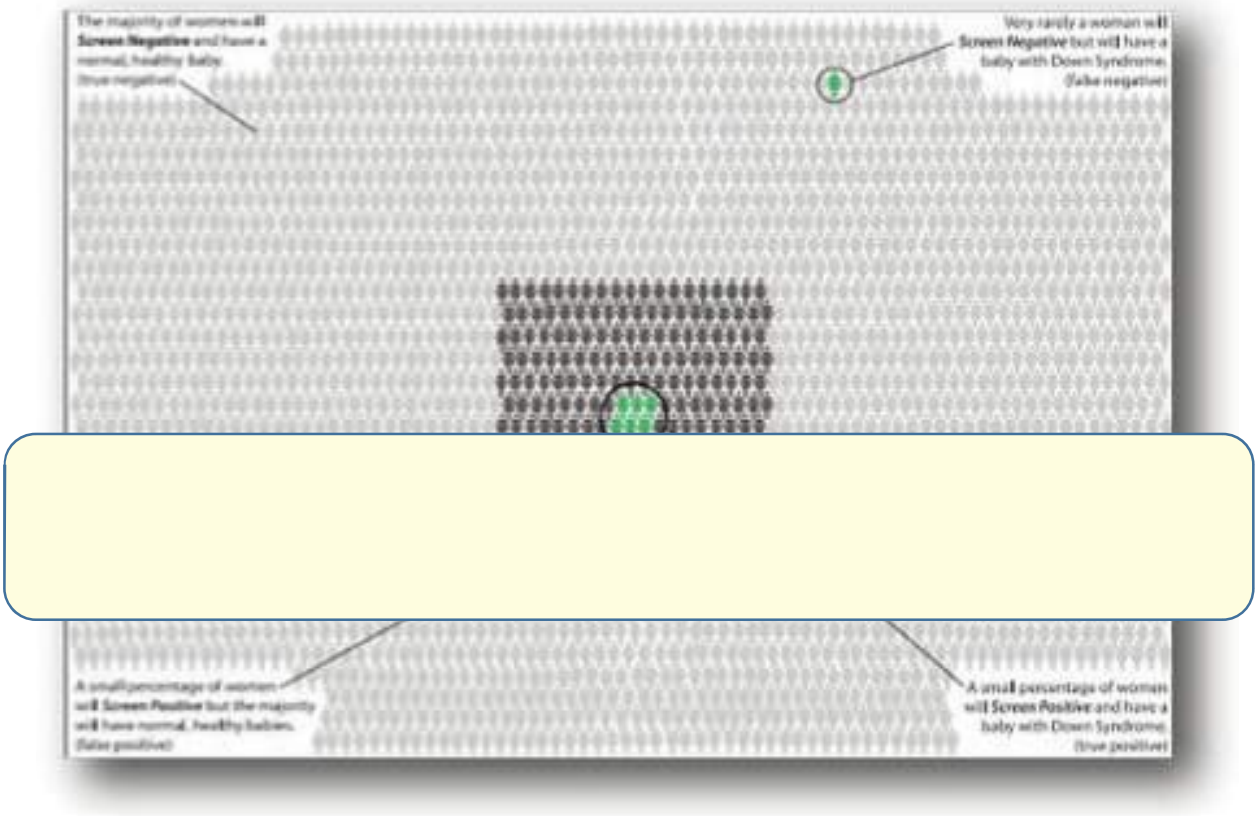
Non-Invasive Prenatal Screen (NIPT): Is a blood test that can be performed as early as 10 weeks of pregnancy. NIPT has a detection rate of Down syndrome greater than 99.9% but also has a false positive rate. The cost is approximately $800*. Although it cannot give you an answer that is 100% certain, it is the most accurate screening available at this time. This test can also tell you if you are having a boy or a girl. Occasionally (about 20% of the time) the test cannot give a result at all due to technical reasons.

*** Prices for Private Pay Screening are going down - Please check with providers for updating pricing! ***

Both private screening options will have results available sooner than the public option, giving families more time to decide whether to have more testing (see pg 5)

Understanding Your Screening Results

If your screening test result is "negative" you can feel fairly certain that your baby will NOT be born with one of the conditions tested for. The chance of the test being wrong is very small : about 1 in 1000 when the result is negative. This is called a *false negative.*



If your screening test result is "positive" it means that the chance is higher than expected for your age that you may have a baby with one of the conditions. You will need to decide if you want to do more testing to find out for sure. The chance that the test result is wrong is high when the screen is positive. Only a small number of women actually have a baby with a condition when the screening test is positive. There is about a 90% chance that your baby is healthy, depending on your age and the condition. The green women in the center represent the true positives while the black women represent those women who have tested positive and are actually negative. When you test positive you may should have more discussion about what this means with your doctor or midwife or with a genetic counsellor, and will be offered more tests to determine if the result is a true positive or a false positive.

Some women who are at higher risk of Down Syndrome and the other conditions mentioned above (such as over age 40) may choose to have the most accurate diagnostic test without having the screening blood tests first.

Diagnostic Testing

Amniocentesis

A very thin needle is put into the woman's belly into the womb. A few teaspoons of the fluid surrounding the baby (amniotic fluid) is taken out through the needle. The needle is guided by ultrasound, so that it does not touch the baby.

The amniotic fluid contains skin cells from the baby. Each cell has a complete set of the baby's chromosomes. A picture is taken of the chromosomes so they can be counted and looked at closely. Amniocentesis is performed after 15 weeks and takes 2 weeks to get a result.

Amniocentesis can tell you for sure if the baby has Down Syndrome or Trisomy 18.

Amniocentesis is associated with a 1 in 200 risk of miscarriage. In other words, if 200 women have an amniocentesis, 1 would lose her pregnancy as a result of the procedure.

Chorionic Villus Sampling (CVS)

Chorionic villus sampling is a diagnostic test in which a needle is put either into the woman's belly or the vagina. The needle is used to collect a small piece of the placenta, known as the chorionic villus. The needle is guided by ultrasound so that it does not touch the baby. CVS can also tell you for sure whether or not the baby has Down Syndrome or Trisomy 18.

CVS is done between 11 – 13 weeks of pregnancy, it may be offered to women who are eligible for diagnostic testing without prior screening. The result of CVS is ready in 2 – 3 weeks.

There is a 1 in 100 chance risk of miscarriage, that the pregnancy will be lost, with a CVS test.

Detailed Ultrasound

This ultrasound is usually done between 18 and 20 weeks of pregnancy. You may decide to only have a detailed ultrasound (without the blood tests), which often can show any of the three conditions tested for and possibly show other health issues such as differences in organs or body.

In a detailed ultrasound examination, a specialist looks carefully at every part of the baby to ensure the baby is growing and developing as expected. The ultrasound can tell whether the baby has an open neural tube defect. It can also confirm how far along you are in the pregnancy, and whether twins are present. Many conditions cannot be seen on ultrasound, so a normal ultrasound does not guarantee that the baby will be normal.

FOR MORE INFORMATION TALK WITH YOUR DOCTOR OR MIDWIFE OR VISIT THE SITE:

http://www.perinatalservicesbc.ca

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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A Screen Positive Result

What does it mean and what do I do now?

BC Prenatal Genetic Screening Program

Perinatal Services BC
Provincial Health Services Authority

You have learned that the result of your prenatal genetic screening is "screen positive." The screening was to find out the chance of your baby having Down syndrome, trisomy 18, or an open neural tube defect.

What does a "screen positive" result mean?

This result does not mean your baby has Down syndrome, trisomy 18, or an open neural tube defect. In fact, most women with this result do not have a baby with one of these conditions. The result means that the chance of your baby having Down syndrome, trisomy 18, or an open neural tube defect is high enough that further testing is offered to determine if your baby has any of these three conditions.

As part of your screen result, you have been given a number that estimates the chance your baby has one of the conditions screened.

Here is an example of a screen positive result:

The *chance* of Down syndrome is 1:100. This is the same as saying:

- There is a 1% chance your baby will have Down syndrome **or**
- There is a 99% chance your baby will not have Down syndrome **or**
- If 100 women had the same result, one of them would have a baby with Down syndrome and the other 99 would not.

According to your screen result, you screened positive for:

_____

The chance your baby has this condition is:

1 in _____ which is _____%

What happens now?

Your health care provider may need to confirm how far along you are in your pregnancy. It is important to make sure the pregnancy dating used for the prenatal screen result is correct.

If you have a screen positive result for an open neural tube defect, you will be offered a detailed ultrasound and an appointment with a maternal fetal medicine doctor or a genetic counsellor at one of BC's medical genetics clinics in Vancouver or Victoria. If your baby has an open neural tube defect, this is usually seen on the ultrasound scan.

If you have a screen positive result for Down syndrome or trisomy 18, you will be offered another blood test called NIPT (for Non-Invasive Prenatal Testing). You may also have the option of an amniocentesis, depending on the level of risk indicated by your screen positive result.

What is Non-Invasive Prenatal Testing (NIPT)?

It is a more accurate screening test for Down syndrome and trisomy 18 than SIPS / IPS / Quad and it is done through a blood test. It detects almost all babies with Down syndrome and trisomy 18 with very few false positive results. NIPT will give you a new risk (very low or very high) of having a baby with Down syndrome, trisomy 18, or trisomy 13. If the risk is very low, no further testing would be recommended. If the risk is very high, an amniocentesis would be needed to confirm the result. The NIPT test result is available in 10 days. For women with a positive SIPS / IPS / Quad screen, NIPT is funded by MSP.

What is an amniocentesis?

It is a diagnostic test. It tells you for sure if your baby has Down syndrome or trisomy 18. It is done by putting a very fine needle into your belly to remove a few teaspoons of amniotic fluid from around the baby. The needle is guided by ultrasound so it does not touch the baby. The baby's cells that are in the fluid sample are looked at to find out whether your baby does or does not have Down syndrome or trisomy 18. When an amniocentesis is done for a positive screen, results are available in 3 days.

Amniocentesis has a 1 in 200 (0.5%) chance of pregnancy loss. In other words, if 200 women have an amniocentesis, one would lose the pregnancy as a result of the procedure.

Should I have extra testing?

It is your choice. Not every woman who has a screen positive result wants extra testing. Think about these questions to help make up your mind.

- Do I think the chance that is given on my screen result is high or low?
- Do I want to know if my baby has Down syndrome or trisomy 18 before the baby is born?
- If I find out that my baby has Down syndrome or trisomy 18, what will I do with this information? Will I end the pregnancy? Will I continue the pregnancy? Will I use the information to prepare for a child with special needs? Will I make an adoption plan for the baby?
- Will I feel too worried for the rest of my pregnancy if I do not have additional testing?

If I want extra testing and am given the option of NIPT or amniocentesis, which one should I have?

It is your choice. Think about these two questions to help you make up your mind.

- Will I be reassured if my NIPT result shows a very low risk of Down syndrome, trisomy 18, and trisomy 13?
- Do I need a result that is 100% accurate (amniocentesis and diagnostic test) even if it means taking the 1 in 200 chance of pregnancy loss?

What if the result of the amniocentesis or detailed ultrasound shows that the baby has one of these conditions?

Your health care provider, as well as medical geneticists or genetic counsellors, are there to discuss your choices with you and to help you make a decision that is right for you.

Please take this home to read, think about and talk over with your partner and family. If you have more questions, or feel a genetic counselling appointment to talk about your options would be helpful, talk to your health care provider.