For patients living in **Fraser**, **Interior**, **Island**, and **Vancouver Coastal Health**

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

What is prenatal genetic screening?

It is a blood test available to all pregnant women/individuals 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/individuals of all ages. In fact, the chance of having a baby with Down syndrome or trisomy 18 is lower in younger women/individuals and higher in older women/individuals.

Although most babies are born healthy, all women/individuals 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 between 11 and just under 14 weeks of pregnancy. Although adding the NT gives more information for the screen result, the blood tests described above 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 or having diagnostic testing by chorionic villi sampling or amniocentesis. These options are further described on page 6.

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.

Talk to your health care provider about your screening options.

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.

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

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/individuals 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/individuals have prenatal genetic screening results showing chances are low for these conditions.

Although 1 in 10 women/individuals will have a screen positive result, most of these women/individuals 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/individual ages.

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

Your health care provider will first arrange a detailed ultrasound and then the necessary follow-up depending on whether or not an abnormality is detected 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 which is more accurate and is called prenatal cell-free DNA screening commonly known as NIPT. 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/individuals 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 prenatal cell-free DNA screening, commonly known as NIPT?

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 but not 100%. An amniocentesis would then be offered to confirm the positive NIPT result. The NIPT test result is available in 10 days. The NIPT test is covered by the provincial medical plan for women/individuals with a positive (SIPS/IPS/Quad) screen result for Down syndrome or trisomy 18, as well as for women/individuals with a previous pregnancy or baby with trisomy 21, 18, or 13.

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/individuals 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/individuals 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/individuals 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, 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/individuals 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 Perinatal Services BC (PSBC) at 604-877-2121.

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