Prenatal Genetic Screening

Prenatal genetic screening is optional and available to all pregnant clients in British Columbia who are covered by MSP. These tests evaluate your chance of having a child affected by one of the following conditions:

- **Down syndrome**--an extra copy of chromosome 21. Down syndrome may cause mild to moderate mental disability, atypical facial features, heart abnormalities, or other physical disabilities. People with Down syndrome can be minimally or profoundly affected. It is impossible to tell the degree of effect in advance from genetic testing.
- **Trisomy 18**--a chromosomal abnormality in which an individual has an extra copy of chromosome 18. Individuals with Trisomy 18 have severe mental and physical disabilities. The majority of babies with Trisomy 18 do not survive past a few days or weeks of life. There is no cure for this condition.
- **Open Neural Tube Defects (ONTD)**--which develop into the spinal cord and brain. One form is open spina bifida, which can cause paralysis or problems walking, learning disabilities and bowel and bladder control difficulties. Anencephaly is a more severe NTD, in which the brain and skull do not develop properly. Babies with anencephaly cannot survive outside the womb.
- Other rarer abnormalities such as **trisomy 13/15** and, in the case of NIPT, some genetic microdeletions.

All couples have a small chance of having a baby with Down syndrome, trisomy 18 or ONTD regardless of age, personal medical history, or family history.

The risk of having a child affected by Down syndrome, trisomy 18, or other chromosomal abnormalities increases as a client gets older, as shown in this table.

Maternal Age	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

Screening Options

Standard screening tests measure the levels of specific proteins (NIPT measures fractionated fetal DNA) in the client's blood. In addition to these protein levels, the risk estimates are calculated using the client's age at delivery, current weight, diabetic status, ethnicity, and the gestational age of the pregnancy.

It is important to understand that these tests can only tell you if you are at an increased or decreased risk of the few conditions listed above. There are many other conditions that we do not have the ability to screen for. Prenatal genetic screening tests cannot provide a diagnosis or a guarantee that your baby is completely "healthy" or "perfect".

There are 4 prenatal genetic screens currently available in BC. These are screening tests. If the result is abnormal, you will be offered further testing and genetic counseling to confirm the diagnosis.

- **Quad screen (Quad)**--one blood test at 15+0 - 20+6 weeks gestation. Offered to all pregnant women who are more than 14 weeks but less than 20+6 weeks gestation, regardless of age or risk factors. Detects 77% of affected pregnancies. Results are available in about 10 days.
- **Serum integrated prenatal screen (SIPS)**--two blood tests, at 10+0 - 13+6 weeks and 14+0 - 20+6 weeks. The 11th week and the 15 - 16th week are the best times for sample collection. Offered to all pregnant women regardless of age or risk factors. Detects 85% of affected pregnancies. Results are available about 10 days after the second sample is drawn.
- **Integrated prenatal screen (IPS)**--early ultrasound at 11-14 weeks, and two blood tests, at 10+0 - 13+6 weeks and 14+0 - 20+0 weeks. The 11th week and the 15 - 16th week are the best times for sample collection. Offered to pregnant women who will be ≥35 years old at EDD; have twin pregnancies; have a history of a previous child or fetus with Down syndrome, trisomy 18 or trisomy 13; are HIV-positive; or are pregnant following in vitro fertilization with ICSI. This test detects 91% of affected pregnancies. Results are available about 10 days after the second sample is drawn.
- **Non Invasive Prenatal Testing (NIPT)**--one blood test that can be done as early as 9-10 weeks. Provides a detection rate of 99% for Down syndrome, 97% for trisomy 18 and 80% for trisomy 13. The test has a 0.5% false positive rate. There is no risk to the pregnancy. It is covered by MSP if you have had an abnormal SIPS or IPS result. You have the option to choose this test instead of SIPS or IPS, but the cost is not covered by MSP. The cost is approximately $120 to $795, depending on which options you choose. Results are available in about 10 days.

Diagnostic Testing Options

The following diagnostic tests differ from previously described screening tests. They provide a definite diagnosis as they look specifically at the chromosomes of the baby to determine if it is affected by Down syndrome or trisomy 18. These tests are offered to women who meet the following criteria:

- all clients 40 years or older at time of due date, or 35 years or older if expecting twins, and
- any client who has had a positive genetic screen result or elevated nuchal translucency measurement.

There are two diagnostic testing options:

- **Amniocentesis** – done by inserting a very fine needle through the women's abdomen and into the uterus. From there, a few teaspoons of the amniotic fluid surrounding the baby is drawn. There are fetal cells floating in the amniotic fluid, and DNA from these cells is examined in a laboratory. During the entire procedure, the baby's position is monitored by ultrasound to ensure that the needle does not make contact with the baby. Amniocentesis is performed after 15 weeks of pregnancy. The increased risk of miscarriage as a result of the procedure is 1 of every 200 women. The results take 2-3 weeks to be completed, though a preliminary result is often available within 1 week.
- **Chorionic Villus Sampling (CVS)** – done by inserting a needle through the client's abdomen or cervix. The needle is used to collect a small piece of placental tissue. The procedure is done with ultrasound guidance to ensure that the needle does not make contact with the baby. CVS is done between 10-12 1/2 weeks of pregnancy. Risks of the procedure include an increased risk of miscarriage of 1-2 in 100, and an increased risk of limb malformation (arms, legs, hands, or feet) of 1 in 1,000-2,000. The results take 2-3 weeks.

The diagnostic test for ONTD is a detailed ultrasound done routinely between 18 - 20 weeks gestation. The ultrasound looks at the baby's anatomy, and can identify some forms of abnormalities, such as open spina bifida. However, ultrasound may not detect all cases of abnormalities.

What to Consider

Genetic screening tests are optional. The risk information provided by the screen is useful to some people, but not to everyone. You need to decide if the information provided is of value to you, and if you want to have the test. Screening can be anxiety-provoking because of the potential information it can provide, the time lag between testing and receiving results, and because of the possibility of false-positive results. Here are some questions to consider before deciding whether or not to have prenatal genetic screening:

- Do I want to know if my baby has Down syndrome, trisomy 18, or ONTD before the baby is born? How will I use this information?
- Would I want to have an amniocentesis or CVS if my screen result said my risk was high?
- Would I be willing to risk a miscarriage from one of the diagnostic tests?
- What would I do if I knew that my baby had Down syndrome or another problem? Would I manage the pregnancy differently? Would I continue with the pregnancy?

INFORMED CONSENT

I have read and understand the above information and have had my questions answered.

- ❑ I choose to have *one* of the following prenatal genetic screening: □ Quad screen □ SIPS □ IPS □ NIPT
  I understand that if the tests are positive, I will be offered amniocentesis or CVS for diagnosis.
- ❑ I am 40 years or older and choose to have diagnostic testing (amnio or CVS), without other screening first.
- ❑ I choose to not have prenatal genetic testing done. I understand that if abnormalities are found on a detailed ultrasound, I will be offered further testing. I understand that I am refusing standard care.

Client's Signature: _____Date: _____

Care Provider's Signature: _____ Date: _____

Reference: BC Prenatal Screening: http://bcprenatalscreening.ca