QUÉBEC PRENATAL SCREENING PROGRAM

Information document for Pregnant Women and Couples

June 1, 2020

Notice

This information document is for pregnant women and couples. The health care professional monitoring the pregnancy is invited to give them a copy and inform and support them in their decision on whether to participate in the Québec Prenatal Screening Program.

This document is **temporary** and will be replaced by the **pamphlet** once the COVID-19 health crisis has been mitigated or has passed.

Votre gouvernement

Québec

QUÉBEC PRENATAL SCREENING PROGRAM

The Québec Prenatal Screening Program aims to make prenatal screening for trisomy 21, trisomy 18 and trisomy 13 accessible to pregnant women and couples in Québec, on a voluntary basis.

TRISOMY 21 - SUMMARY

- Trisomy 21, also known as Down Syndrome, is one of the most common chromosomal abnormalities (affecting 1 in 770 babies).
- A person with trisomy 21 has a 3rd copy of chromosome 21. Therefore, the individual has 47 chromosomes instead of 46.
- Trisomy 21 affects the child's intellectual development.
- A child with trisomy 21 may suffer from other health problems (e.g., heart defects).
- Trisomy 21 is not hereditary in more than 95% of cases and therefore occurs randomly. Less than 5% of cases are inherited.
- Any woman may carry a baby with trisomy 21.
- The probability of having a child with trisomy 21 increases with the pregnant woman's age. The younger the woman, the lower the probability.

The intellectual limitations presented by a child with trisomy 21 are hard to predict. Differences have been observed from one individual to the next and one environment to the next. The degree of stimulation and support offered to the children also has an impact. In most cases, individuals with trisomy 21 will need varying levels of support throughout their lives. With the right amount of support, individuals with trisomy 21 have the resources and potential to develop deep emotional relationships and lead rewarding lives for themselves and others.

TRISOMY 18 - SUMMARY

- Trisomy 18 is a rare chromosomal abnormality (affecting approximately 1 in 4,500 babies at birth).
- Trisomy 18 is caused by the presence of a 3rd copy of chromosome 18.
- Trisomy 18 is often associated with severe birth defects.
- Most pregnancies with a baby having trisomy 18 end in miscarriage.
- Babies born with trisomy 18 die within days after birth as a result of severe heart and brain defects or respiratory problems.
- Like trisomy 21, trisomy 18 occurs randomly in most cases.

TRISOMY 13 - SUMMARY

- Trisomy 13 is a rare chromosomal abnormality (affecting approximately 1 in 7,000 babies at birth).
- Trisomy 13 is caused by the presence of a 3rd copy of chromosome 13.
- Trisomy 13 is very severe and often associated with miscarriage or multiple defects.
- Most babies born with trisomy 13 die soon after birth.
- Like trisomy 21 and trisomy 18, trisomy 13 occurs randomly in most cases.

DESCRIPTION OF THE QUÉBEC PRENATAL SCREENING PROGRAM

The program screens for trisomy 21, but it could also reveal trisomy 18 or trisomy 13 during the pregnancy. Conducting prenatal screening allows us to calculate the probability that your child has one of these trisomies. There are other chromosomal abnormalities but the public program, which is based on Canadian recommendations, does not screen for them. The screening results might lead you to make a decision on whether to continue the pregnancy.

The Québec Prenatal Screening Program includes:

1. Performing the biochemical test, including a nuchal translucency measurement, if possible;
2. Performing the non-invasive prenatal genomic test or performing amniocentesis for further investigation if the biochemical test indicates a high probability of fetal trisomy.

It is up to you to decide whether to proceed with prenatal screening and use the results obtained to make an informed decision. The choice is yours at every stage.

STAGE 1: The biochemical test (with or without nuchal translucency)

The biochemical screening test, with or without the nuchal translucency measurement, reveals whether the probability of trisomy 21 is low or high. The test could also reveal a high probability of trisomy 18.

Blood tests

The first stage in screening, the biochemical test, provides an analysis of the baby's proteins or hormones present in the mother's blood. The test involves the administration of **two blood tests** during your pregnancy:

- the first between weeks 10 and 13;
- the second between weeks 14 and 16;

NOTE: It is important for both blood tests to be performed during the right period. The screening test is more reliable using two blood tests than using just one.

First trimester ultrasound

A **first trimester ultrasound** will be proposed between weeks 11 and 14 to see how your pregnancy is progressing and identify possible defects in your baby. This ultrasound is used to precisely estimate how long you've been pregnant to allow a more reliable calculation of the probability of having a baby with a trisomy.

The **nuchal translucency measurement** by ultrasound might also be proposed. It is performed between weeks 11 and 13. It allows a measurement to be taken of a thickening due to an accumulation of liquid in the back of the baby's neck. For babies with trisomy 21, the nuchal translucency is often thicker than normal. When available, the nuchal translucency measurement is combined with the biochemical test to improve even further the calculation of the probability of a trisomy.

Results of the biochemical test (with or without nuchal translucency)

- **Low probability**: the probability that you are carrying a child with trisomy 21 is low and no additional tests are necessary (more than 95% of women get this result when they get the biochemical test).

  A low probability **does not guarantee** that the baby does not have trisomy 21. Considering the natural differences between individuals and the limitations of the test, it does not allow all babies with trisomy 21 to be identified.

- **High probability**: It is possible that you are carrying a child with trisomy 21 (3% to 4% of women get this result when they get the biochemical test). A follow-up will be proposed to confirm or refute the presence of a chromosomal abnormality

  A high probability does not necessarily mean that the baby has trisomy 21.

As part of this test, a similar calculation of probability is made for trisomy 18. The results for trisomy 13 are comparable to those for trisomy 18. For that reason, we cannot distinguish between the two at this stage of screening.

If the biochemical screening test indicates a high probability, the non-invasive prenatal genomic test or an amniocentesis will be proposed. It is up to you to decide whether to take one test or the other.

STAGE 2: The non-invasive prenatal genomic test or amniocentesis

Non-invasive prenatal genomic test (NIPT)

The genomic test is a screening test that will be proposed if you have a high probability of having a baby with trisomy 21 (or trisomy 18) following the program's biochemical test. The genomic test is performed by taking a sample of the mother's blood and analyzing DNA fragments from the placenta.

The genomic test detects or refutes whether the baby has trisomy 21, trisomy 18 or trisomy 13.

The genomic test is proposed because it is **reliable** and **safe** (there is no risk of miscarriage as with amniocentesis).

The genomic test might be proposed to you right away (instead of the biochemical test) if you are in one of the following situations:

- you have had a previous pregnancy in which the baby had trisomy 21, trisomy 18 or trisomy 13;
- you will be 40 years old or older at the time of delivery;
- the test is prescribed following a consultation in genetic medicine.

Results of the genomic test

- **Low probability**: this result indicates that the baby has a low probability of having one of the tested trisomies, even if the result of the biochemical test showed a high probability initially. A "low probability" result of the genomic test is reliable (over 99%) and there is no need to conduct further investigations.

- **High probability**: this result indicates that the baby is likely to have one of the three tested trisomies. However, a "high probability" result is not 100% certain and only the amniocentesis can determine with great certainty whether the baby has one of the trisomies.

Since it has some limitations, the prenatal genomic test:

- Does not identify all babies with one of the tested chromosomal abnormalities.
- Does not eliminate the possibility of a false-positive result. In fact, there is a possibility that the baby does not have one of the trisomies even if the result is positive. For that reason, amniocentesis is proposed in the event of a positive result.
- Does not work for a small percentage of women. In this situation, the health professional monitoring your pregnancy can discuss your options, notably amniocentesis, with you.
- Does not screen for all genetic diseases causing abnormalities, intellectual delays or autism.

Diagnostic test: amniocentesis

Amniocentesis remains an option if the biochemical test indicates a high probability.

Amniocentesis is a diagnostic test that allows us to identify the number of each chromosome. This method is reliable in determining whether the baby has a trisomy, but it presents risks to the pregnancy, such as miscarriage. Amniocentesis, which is performed by inserting a fine needle into your abdomen, allows a small amount of amniotic fluid containing the baby's cells to be collected.

Results of the amniocentesis

- The baby does not have one of the tested chromosomal abnormalities.
- The baby has one of the tested chromosomal abnormalities. This result is reliable and allows you to make a decision about your pregnancy.

You can consult the table, which compares the genomic test and amniocentesis, and discuss the pros and cons of each method with a health care professional.

Genomic test and amniocentesis: characteristics

	Non-invasive prenatal genomic test	Amniocentesis (diagnostic test)
If the test result is negative	The result is over 99% reliable	This result is the most reliable
If the test result is positive	The result is probable but must be confirmed	The result is nearly 100% reliable
Risk for the pregnancy	None	Miscarriage (1 in 500 to 1 in 200)
Waiting time for results	About 5 to 10 days	About 3 days with a rapid diagnostic test

If your baby has trisomy 21, you will have a decision to make:

- Continue your pregnancy and prepare to have a child with trisomy 21;
- Terminate your pregnancy and live with the accompanying grief.

If the baby has trisomy 18 or 13, you will also have to make a decision on whether to continue the pregnancy.

Faced with the difficult decision of continuing or terminating your pregnancy, you may need help. You and your spouse should not hesitate to discuss the decision with your loved ones or a health care professional. You can also contact support groups for parents of children with trisomy 21. This could help you make the best decision for you.

Can you choose whether to proceed with the prenatal screening test?

Yes. The decision is entirely up to you. Some women want to have this information during the pregnancy and others don't.

Also, keep in mind that at any time and at any stage, you can choose to end your participation in the program.

Participating in the program might cause you stress and anxiety before getting the tests, while awaiting your results or when receiving your results. You can discuss the decision on whether to participate in the Québec Prenatal Screening Program with your spouse, your loved ones, your doctor, your midwife, another health professional or other groups for parents who have a child with a trisomy.

For more information on prenatal screening, talk to the health care professional monitoring your pregnancy.