QUÉBEC PRENATAL SCREENING PROGRAM

Additional Information for Pregnant Women and Couples

June 11, 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 website 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. There are other chromosomal abnormalities but the public program, which is based on Canadian recommendations, does not screen for them.

The Quebec Prenatal Screening Program is offered free of charge in the public health network.

The screening is offered during pregnancy follow-up but it is not a routine exam. Your doctor or midwife must ensure that you have received all the information you need to decide whether you wish to participate. This decision is entirely up to you. The professional monitoring your pregnancy must also ask you to sign the consent form if you wish to participate in the program.

Before signing the consent form, you need a clear understanding of what trisomies 21, 18 and 13 are, what information you will gain from this screening and the decisions you might have to make.

By participating in the screening program, you will be able to find out as soon as possible if the child you are carrying presents a high probability of having one of the three tested trisomies. When the probability is high, additional analyses are offered to confirm that your baby has one of the trisomies. If that is the case, you may have to decide if you want to continue your pregnancy, and give birth to the child, or if you want to terminate your pregnancy.

The purpose of this document is to present you with the relevant information on the Quebec Prenatal Screening Program to help you decide whether to participate in the program.

Trisomy 21

Trisomy 21, also known as Down Syndrome, is caused by the presence of an extra copy of chromosome 21 (each human cell has normally 23 pairs of chromosomes). This chromosomal abnormality is one of the most common. Trisomy 21 is not generally hereditary and therefore occurs at random.

All people with trisomy 21 have some degree of intellectual disability, varying from mild to severe. Some people may need a lot of support and guidance throughout their life, while others may have a job and lead an almost independent life. It is impossible to determine the level of autonomy and intellectual functioning that a child with trisomy 21 may develop.

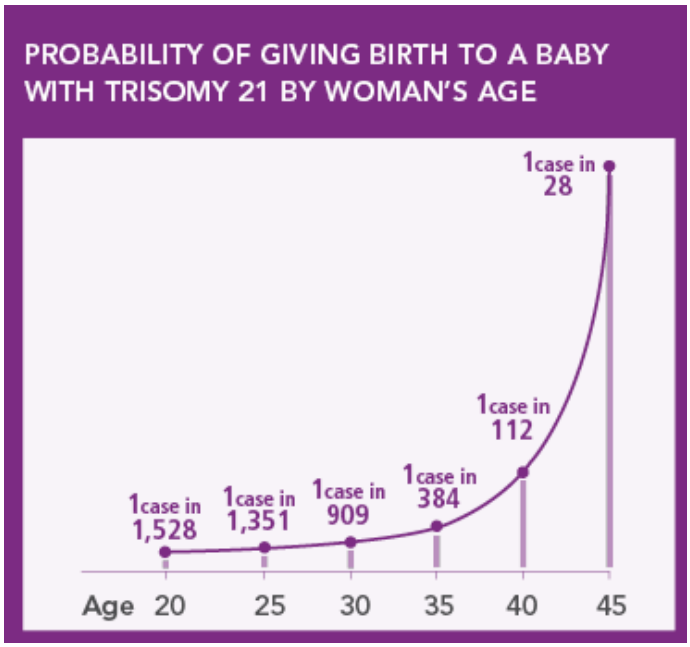
Most children with trisomy 21 can walk and talk, but it takes longer for them to learn language and motor skills. The interventions and support these children receive from an early age influence their development. In addition to intellectual disability, these children may have other types of health problems, such as heart and intestinal defects, or epilepsy.

People with trisomy 21 have the potential to establish deep emotional relationships and lead rewarding lives for themselves and their loved ones.

Frequency of trisomy 21

About 1 baby in 770 is born with trisomy 21. Any pregnant woman, regardless of her age, may be carrying a child with trisomy 21. However, the probability increases with age.

Figure 1 shows that the probability of carrying a child with trisomy 21 increases with the woman's age. At 20 years old, the probability is 1 in 1,528 pregnant women, while at age 45, the probability increases to 1 in 28 pregnant women.



Source: CUCKLE, H.S., et al., 1987

Figure 1. Probability of giving birth to a baby with trisomy 21 by woman's age

Trisomy 18

Trisomy 18, also known as Edwards Syndrome, is caused by the presence of an extra copy of chromosome 18 (each human cell has normally 23 pairs of chromosomes). This chromosomal abnormality is the most common after trisomy 21.

Trisomy 18 is rare and affects approximately 1 in 4,500 babies at birth. However, nearly 95% of pregnancies in which the baby has trisomy 18 end in miscarriage or the baby's death before birth. Any pregnant woman can carry a baby with this chromosomal abnormality but the probability increases with age. Like trisomy 21, trisomy 18 occurs randomly in most cases.

People with trisomy 18 have stunted growth before and after birth. They have an intellectual disability and severe global developmental delay. Most of these people have abnormalities of the hands and feet. Major malformations of internal organs, particularly the heart and kidneys, are common. Most babies born with trisomy 18 will die within the first weeks of life. Only 5 to 10% survive beyond a year. However, in exceptional cases, some babies reach adulthood. In all cases, babies living with trisomy 18 will receive support and comfort care or may receive some treatment, depending on the symptoms.

Trisomy 13

Trisomy 13, also known as Patau Syndrome, is caused by the presence of an extra copy of chromosome 13 (each human cell has normally 23 pairs of chromosomes).

Trisomy 13 affects approximately 1 in 7,000 babies at birth. Trisomy 13 is very severe and often associated with a miscarriage or multiple defects. Over 95% of affected babies die before birth. Of the pregnancies carried to term, half the babies will die during the first month of life and 90% will die before one year of cardiac, kidney and neurological complications. In exceptional cases, the baby may live longer if he or she does not have major brain malformations. Any pregnant woman can carry a baby with this chromosomal abnormality but the probability increases with age.

Trisomy 13 is characterized by facial abnormalities and limb deformities, very severe neurological problems and brain, heart and urogenital malformations. Intellectual and developmental delays are severe.

Generally, medical care is limited to providing support and comfort.

Stages of screening

The Québec Prenatal Screening Program includes:

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

Figure 2 shows the different stages of prenatal screening, in particular, the moments when you will have to make a decision.

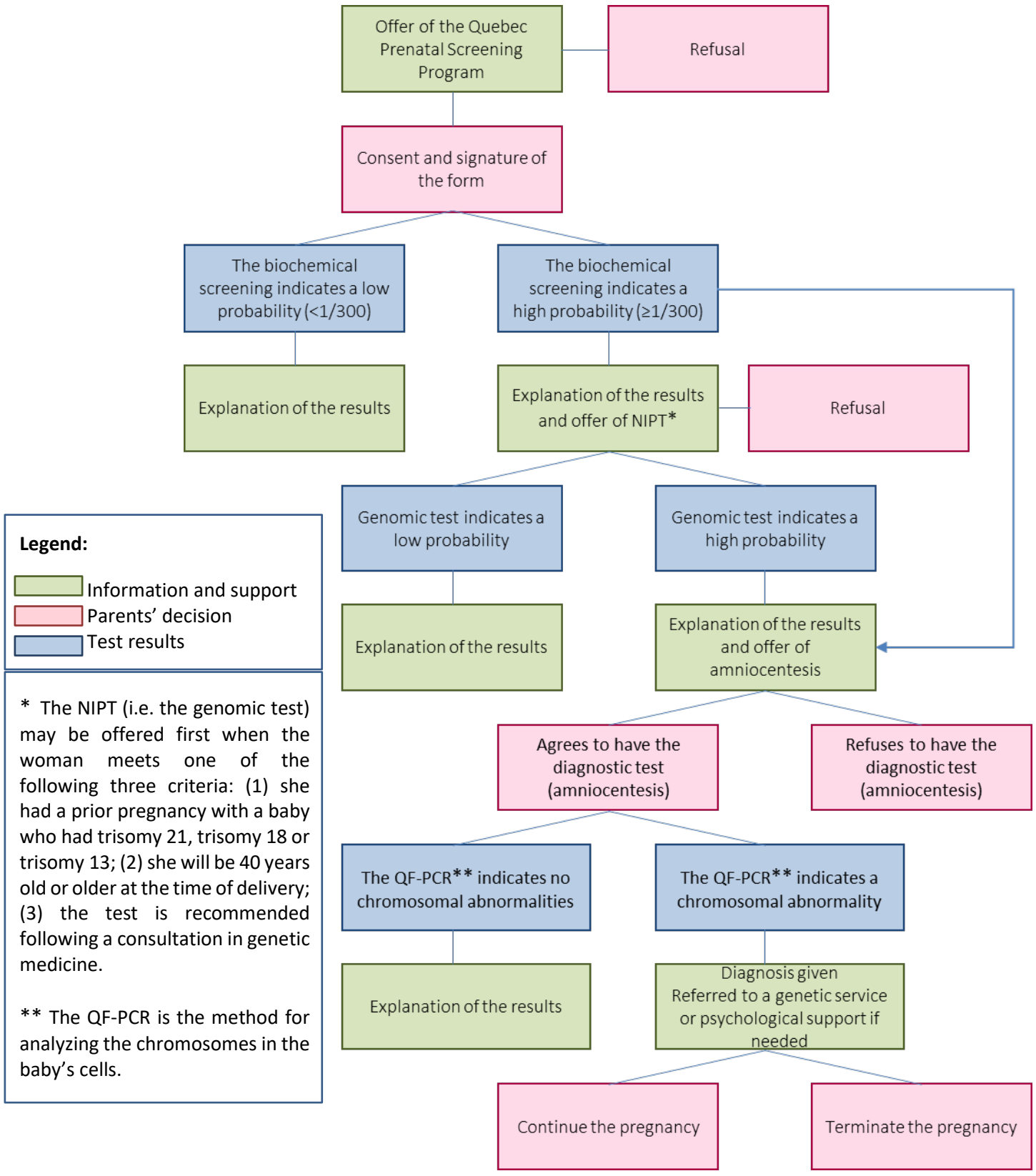


Figure 2. Stages of prenatal screening

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;

Both blood tests are important and ensure more reliable results than a single blood test. It is therefore preferable, if you decide to participate in the Quebec Prenatal Screening Program, to have two blood tests done at the right time. The blood tests present no danger to the pregnancy.

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)

The results of the two blood samples and nuchal translucency measurement (if available) will indicate whether the probability of trisomy 21 is low or high:

Low probability (probability less than 1 in 300): 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 (probability equal to or higher than 1 in 300): 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.

At this stage, your doctor or midwife will recommend that you move on to stage 2 of the program. The decision is yours.

STAGE 2: The non-invasive prenatal genomic test OR the amniocentesis

Depending on the results you receive in stage 1 of the program, your doctor or midwife might offer you the non-invasive prenatal genomic test or recommend that you go straight to amniocentesis. You will be given the explanations and information you need to decide which option is preferable to you. Table 1 summarizes the characteristics of both tests.

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.

Amniocentesis: diagnostic test

Amniocentesis is a diagnostic test offered to you if the result of the non-invasive prenatal genomic test shows a high probability for trisomy 21, trisomy 18 or trisomy 13. Some women or some couples may want to go straight to amniocentesis, without doing the genomic test, despite the risks it might cause (see table 1). This possibility can be discussed with the professional responsible for monitoring your pregnancy.

The test consists of inserting a fine needle into your abdomen to collect a small quantity of the fluid surrounding the baby in the uterus (amniotic fluid). This fluid contains the baby's cells and amniocentesis allows us to analyze the chromosomes in these cells. The test can be performed starting in the 15th week of pregnancy.

Amniocentesis is the only test that can confirm with great certainty whether your baby has one of the three tested trisomies, just as it can confirm with the same certainty that your baby does not have one of these trisomies.

Possible complications associated with amniocentesis

Amniocentesis carries risks to the pregnancy, the main one being miscarriage. This risk is about 1 miscarriage in 300 women who have amniocentesis. Therefore, this test is offered only to pregnant women who present a high probability on the screening test. Certain minor complications are also observed, the most common (2 to 5% of cases) being:

- a slight leakage of amniotic fluid;
- uterine contractions;
- abdominal pain.

Results of the amniocentesis

Absence of trisomy: your baby does not have trisomy 21, trisomy 18 or la trisomy 13. This result is very reliable.

Presence of trisomy 21: your baby has trisomy 21. This result is very reliable.

You must then decide if you wish to:

- Continue your pregnancy and prepare to parent a child with trisomy 21;
- Continue your pregnancy and place the child for adoption;
- Terminate your pregnancy and deal with the grief that might cause.

The decision must be made when there is no way to predict the severity of the intellectual disability or the level of autonomy your child might have and without knowing if your child has physical deformities or other health problems.

Presence of trisomy 18: your baby has trisomy 18 or trisomy 13. This result is very reliable.

You must then decide if you wish to:

- Continue the pregnancy;

- Terminate the pregnancy.

The decision must take into consideration the severity of the deformities associated with these trisomies and the fact that 90% of affected newborns will not live longer than a year. The decision must also be made when it is impossible to predict with certainty the condition of your baby at birth.

Table 1. 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

Keep in mind that at any time and at any stage, you can end your participation in the program.

Special conditions

Women who have a multiple pregnancy (carrying more than one baby) cannot use the Quebec Prenatal Screening Program due to its current limitations. Women pregnant with twins or triplets, for example, must talk to their doctor to discuss the options available for them.

Costs

All stages of the Quebec Prenatal Screening Program are offered free of charge to all pregnant women with a Régie de l'assurance maladie du Québec (RAMQ) health card. Some women with a special status may also have access to it. However, there may be costs associated with the nuchal translucency measurement if it is performed at a private clinic.

Voluntary participation

Participation in the Quebec Prenatal Screening Program is voluntary. The decision is personal and entirely up to you. To help you with your decision making, you can discuss the testing with your partner, your loved ones, the health professional monitoring your pregnancy, a genetic professional or support groups for parents of children with trisomy 21, trisomy 18 or trisomy 13.

For clarity, you could also ask yourself the following questions:

- Do I want to know my probability of having a baby with trisomy 21, trisomy 18 or trisomy 13?

- How will I react if the results of the screening tests are abnormal?
- Do I want to proceed with amniocentesis if the screening is abnormal, despite the risk of miscarriage?
- Will I want to end my pregnancy if I find out my baby has trisomy 21, trisomy 18 or trisomy 13?
- Will I want to continue my pregnancy if I find out my baby has trisomy 21, trisomy 18 or trisomy 13, and raise my child or place him or her for adoption?

Remember that the program is voluntary and that you can choose whether or not to undergo the screening tests, depending on your situation, wishes and values. This decision is personal and the reasons behind your choices are your own. You can also refuse to proceed with amniocentesis if it is offered to you. Moreover, if you wish, you can decide to withdraw from the program at any time.

If you decide to participate in the Quebec Prenatal Screening Program, your doctor or midwife will have you sign a consent form and will help you with the process.

If you decide not to participate in the Quebec Prenatal Screening Program, you do not have to sign anything but your health professional must note your refusal. Your doctor or midwife will answer your questions and inform you of the steps of the pregnancy follow-up.

Help and support

Deciding whether to participate in the Quebec Prenatal Screening Program is not necessarily easy. Deciding to terminate or continue a pregnancy knowing that the baby has a chromosomal abnormality is even harder.

If the results of your participation in the program lead you to make a choice between continuing or terminating your pregnancy, you might need help.

Your doctor or midwife can direct you to psychological services to support you in your reflection. Don’t hesitate to discuss this choice with your loved ones or a health professional. You can also contact support groups for parents who have children with trisomy 21, which might help you in making the best decision for you.

For the contact information of groups in your region, ask the professional who is monitoring your pregnancy.