STAGE 2:
The non-invasive prenatal genomic test OR the 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 screens for trisomy 21, trisomy 18 and trisomy 13.

The genomic test is proposed because it is **reliable** and **safe** (there is no risk of miscarriage compared 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;
- you are pregnant with twins (two fetuses);
- 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 very 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 very 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 a diagnostic test (amniocentesis) can determine with great certainty whether the baby has one of the trisomies.

AMNIOCENTESIS: DIAGNOSTIC TEST

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

Amniocentesis is a diagnostic test that allows 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 trisomies (trisomy 21, trisomy 18 or trisomy 13).
- The baby has one of the tested trisomies. This result is very 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 (approximately 1 in 500)
Waiting time for results	About 5 to 10 days	About 3 days with a rapid diagnostic test

Faced with the difficult decision of continuing or terminating your pregnancy, you may need help. You and your partner 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.

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.

PARTICIPATION IN PRENATAL SCREENING

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

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

For more information on prenatal screening and the limitations of biochemical and genomic tests, talk to the health care professional monitoring your pregnancy or visit **Québec.ca/depistage**.

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 partner, your loved ones, your doctor, your midwife, another health professional or other groups for parents who have a child with a trisomy.

QUÉBEC PRENATAL SCREENING PROGRAM

23-931-01A

Santé et Services sociaux Québec

Votre gouvernement

Québec

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.

SUMMARY

TRISOMY 21

- Trisomy 21, also known as Down Syndrome, is one of the most common trisomies (affecting 1 in 800 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, and that may vary from one child to the next.
- A child with trisomy 21 may suffer from other health problems (e.g., heart defects).

The level of stimulation and support offered to these children impacts their development. In most cases, individuals with trisomy 21 will need varying levels of support throughout their lives. However, given a fair chance, individuals with trisomy 21 have the resources and potential to develop deep emotional relationships and lead rewarding lives for themselves and their loved ones.

TRISOMY 18

- Trisomy 18 is a rare trisomy (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.
- Most babies born with trisomy 18 die soon after birth as a result of severe heart and brain defects or respiratory problems.

TRISOMY 13

- 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.

Trisomy 21, like trisomy 18 and trisomy 13, 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, trisomy 18 or trisomy 13.
- The probability of having a child with trisomy 21, trisomy 18 or trisomy 13 increases with the pregnant woman's age. The younger the woman, the lower the probability.

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.

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 reveals whether the probability of trisomy 21 is low or high. The test could also reveal a high probability of trisomy 18. It could be combined or not with the nuchal translucency measurement.

BLOOD TESTS

The biochemical test provides an analysis of the baby's proteins or hormones present in the mother's blood. The test involves having 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 abnormalities 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. This ultrasound will also determine if there is a single fetus or more than one.

The nuchal translucency measurement by ultrasound might also be proposed. It is performed between weeks 11 and 13. It measures the thickness of the fluid build-up at the back of the baby's neck. In babies with trisomy 21, the fluid build-up is often thicker than normal. When available, the nuchal translucency measurement is combined with the biochemical test to calculate the probability of a trisomy. Nuchal translucency measurement could involve costs when performed in a private clinic.

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 have 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 have the biochemical test). The non-invasive prenatal genomic test or an amniocentesis will be proposed.

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

At this stage, the test may reveal a high probability of trisomy 18.