You can request the prenatal screening test. Unlike the diagnostic test, the screening test cannot determine with certainty whether or not your baby has trisomy 21 but it indicates whether the probability or the risk of your child having this abnormality is low or high. The screening test does allow calculation of your individual probability with greater precision than simply basing it on your age. All pregnant women can undergo prenatal screening, no matter their age.

What does the prenatal screening test involve?

The prenatal screening test is done on blood drawn from the mother. If you decide to participate in the Trisomy 21 Prenatal Screening Program of Québec, full program details will be provided to you by the doctor who is caring for you during your pregnancy or another health professional. Among other things, you will have to provide two blood samples during your pregnancy:
- The first between the 10th and 13th week
- The second between the 14th and 16th week

An ultrasound may also be suggested, to verify the status of your pregnancy. During the ultrasound, the nuchal translucency of the baby may also be measured. Nuchal translucency is the quantity of fluid that collects within the nape of the fetal neck between the skin and the spinal column. Nuchal translucency thickness is often greater in babies with trisomy 21.

Results of the prenatal screening test

By combining the results of the two blood samples and your age, the test makes it possible to determine whether you have a low or high probability level.
- **Low probability**: The probability that you are bearing a baby with trisomy 21 is low and your doctor will not suggest that you undergo a diagnostic test.
- **High probability**: The probability that you are bearing a baby with trisomy 21 is high and your doctor will suggest that you undergo a diagnostic test.

It is important to emphasize that a low probability result does not guarantee that the baby is not affected. The prenatal screening test is a first step and thus only identifies your level of probability of bearing a baby with trisomy 21.

The screening test for trisomy 21 may detect a higher probability of other congenital anomalies such as the trisomy 18 chromosome or neural tube anomalies such as spina bifida. If screening detects a high probability of any of these anomalies, the health professional that monitored your pregnancy will inform you of the steps you will need to take.

ATTENTION: A prenatal screening test based on a single blood sample is less reliable than a test based on two samples. Both samples are therefore important.

THE PRENATAL SCREENING TEST

Two blood samples for more reliable results

If you only provide one blood sample for the prenatal screening test, the likelihood of having a baby with trisomy 21 will be assessed on the basis of the result of that sample alone.

THE DIAGNOSTIC TEST

If your prenatal screening test indicates that you are at high probability of bearing a baby with trisomy 21, you can find out for sure by undergoing a diagnostic test. The most widely used prenatal diagnostic technique is amniocentesis with a study of the chromosomes. This test makes it possible to determine whether or not the foetus has trisomy 21.

What does the diagnostic test involve?

Amniocentesis can be performed beginning the 15th week of pregnancy. A sample of the amniotic fluid surrounding your baby is collected using a small-gauge needle that is inserted in your abdomen. The technique does, however, involve a certain risk of complications, including the loss of your baby. It is mainly offered to women at high probability because of their family history or based on the result of the prenatal screening test.

Results of the diagnostic test

Amniocentesis performed during pregnancy can reveal that:
- The baby does not have trisomy 21 (This will be the case for most babies)
- The baby has trisomy 21
- The baby has other chromosome abnormalities (Your doctor will refer you to a genetic specialist)

The decision is entirely up to you. Certain women wish to know their probability of bearing a baby with trisomy 21 while others prefer not to know.

You may feel stress and anxiety during the process, either before the test, while waiting for the result, or when you receive the result. You can discuss the decision to participate in the prenatal screening program with your spouse, family and friends, your doctor, your midwife, another health professional, or groups of parents of children with an intellectual disability.
As shown in the graph, the probability of giving birth to a baby with trisomy 21 increases with age.

The purpose of the Trisomy 21 Prenatal Screening Program of Québec is to provide pregnant women and couples across Québec with access to voluntary prenatal screening for trisomy 21.

TRISOMY 21, AT A GLANCE
- Trisomy 21, also known as Down syndrome, is the most common viable chromosome abnormality.
- Individuals with trisomy 21 have a 3rd chromosome attached to the 21st pair, and thus have 47 chromosomes instead of 46.
- This abnormality affects about 1 in every 770 babies.
- Trisomy 21 affects the intellectual development of the child.
- Children with trisomy 21 may also suffer from other health problems (e.g., heart malformations).
- Trisomy 21 is generally not hereditary.
- All women may bear a foetus affected by this chromosome abnormality.
- The probability of having a baby with trisomy 21 increases with the age of the pregnant woman; the younger the woman, the lower the probability.

It is difficult to determine the intellectual limitations of children with trisomy 21. There are differences from person to person and from one environment to another. The amount of stimulation and support provided to children also has an impact. In most cases, however, a child with trisomy 21 will require varying degrees of support throughout his or her life. However, given a fair chance, individuals with trisomy 21 have the resources and potential to develop deep emotional relationships and to lead rewarding lives for themselves and their families.

For more information on prenatal screening for trisomy 21, you can talk with the doctor who is caring for you during your pregnancy or you can visit our website: www.msss.gouv.qc.ca/depistage-prenatal

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