WHAT TO DO IF THE RISK IS ELEVATED (SUPERIOR TO 1 OUT OF 250)?

This does not mean that the foetus is necessarily T21. To be sure, the caryotype of the foetus can be performed. The expectant mother is informed of this possibility. This exam counts the exact number of chromosomes 21 and establishes with certainty whether or not the child is T21.

Depending on the situation, a sampling of trophoblast (sampling cells of the placenta as soon as 11 weeks of amenorrhea) or an amniocentesis (sampling the cells of the amniotic fluid after the 15 th week of amenorrhea) might be suggested. These exams are done with a needle under ultrasound guidance, through the skin of the abdomen.

Miscarriage or premature labor risk due to these techniques is around 1%.

Results are available between a few days to 3 or 4 weeks depending on the technique used.

The expectant mother may request or refuse this sampling, which is covered by the health insurance.

WHAT TO DO IF THE COUNT OF CHROMOSOMES REVEALS A FOETUS WITH T21?

This situation requires assistance, taking the time to think things through, and getting information on the possible care of people with T21.

There are different possibilities:
- continue the pregnancy and welcome the child;
- continue the pregnancy and give the child to social services administred by the conseil general;
- request the termination of the pregnancy, called medical termination of the pregnancy, after decision by the prenatal diagnosis center.

To help the expectant mother, and her couple, to make their own choice, they may meet:
- professional care givers around them (gynecologist, obstetrician, family doctor, radiologist, midwife, pediatrician, genetician, psychologist...);
- members of the prenatal diagnosis center, in each region, where all the above professionals work in teams;
- specialized associations approved for accompanying patients with T21 and their families. The list can be provided by their doctor.

LIST OF ASSOCIATIONS

UNAPEI (Union nationale des parents d'enfants inadaptés)
15 rue Coysevox - 75876 Paris cedex 18
☎ : 01 44 85 50 50 - www.unapei.org

TRISOMIE 21 FRANCE
4 square François Margand BP 90249 - 42006 Saint-Etienne cedex 1
☎ : 04 77 37 87 29 - www.trisomie21-france.org

FONDATION JÉRÔME LEJEUNE
37 rue des Volontaires - 75015 Paris
☎ : 01 44 49 73 30 - www.fondationlejeune.org

REGARDS 21
11 bis rue de la République - 78100 Saint-Germain-en-Laye
www.regardsurlatrisomie21.org

LIST OF MULTIDISCIPLINARY PRENATAL DIAGNOSIS CENTRES

List of multidisciplinary prenatal diagnosis centres
http://www.agence-biomedecine.fr/uploads/document/liste_CPDPN_151110.pdf

List of regional perinatal centres
http://www.perinat-france.org/portail-grand-public/reseaux/reseaux-perinatals/les-reseaux-de-sante-regionaux-perinatals-432.html

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Accent Aigu - 02/2013

CNGOF
Collège National des Gynécologues et Obstétriciens Français

CNSF
Collège National des Sages-Femmes de France

FNMR Fédération Nationale des Médecins Radiologues

SFR sfr.radiologie.fr

Information for expecting mothers on the possibility of taking a voluntary screening test for trisomy 21

This document, for all expecting mothers, explains the currently available individual screening test for T21.

The individual screening test for T21 **measures the risk (high or low)** that the current foetus would be T21.

Two exams can be performed:
- a blood test on the expecting mother;
- an ultrasound examination of the foetus.

These two exams pose no risk for the pregnancy.

The screening test requires a written consent from the expectant mother and is not obligatory.

As the expecting mother may **choose**:
- to request or refuse the screening test for T21;
- to request or refuse an amniocentesis or sampling of the placenta if a screening test detects a high risk of T21;
- to change her mind at any moment.

WHAT IS T21?

T21, or Down's syndrome, erroneously called mongolism, is caused by an extra chromosome in the patient's cells.

Normally, in every cell of the human body there are 46 chromosomes, in 23 pairs. In T21, there is an extra chromosome in the 21st pair. The patient has therefor 47 chromosomes.

In the vast majority of cases, no one else in the family has a similar condition.

THIS EXTRA CHROMOSOME CAN HAVE MANY CONSEQUENCES, THE MOST FREQUENT ARE:

- Varying degrees of intellectual deficiency, with different chances for social integration and relative autonomy varying from one child with T21 to another, with possible autonomy.
- Education and the companionship being important factors in the development and well-being of these children.
- A characteristic aspect of the face (which does not exclude ressemblance to the parents) .
- A height rarely attaining more than 1m60 at adulthood.
- Diminished muscular tone called hypotonia.
- Malformations of variable severity, often in the heart and digestive system, for which some treatments exist.
- In some other cases, other problems, such as predisposition to certain types of blood disease.

HOW DO WE PERFORM THE DIAGNOSIS?

Before birth, a sample of the cells belonging to the foetus can provide its caryotype (the study of its chromosomes).
This exam reveals the presence or absence of the extra chromosome.
These fetal cells are in the amniotic fluid in which the foetus floats, or in the placenta.

The sampling via amniocentesis or trophoblast biopsy carries some risk of provoking a miscarriage.
This is why it is only recommended when there is an elevated risk that foetus is T21.

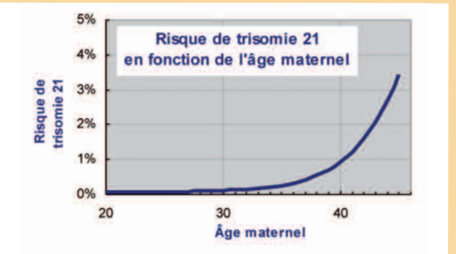
After birth, the presence of an extra chromosome 21 might be suspected from facial characteristics, or from some other malformation.
Certain detection of the disease requires a caryotype exam studying the child's chromosomes, which can reveal the extra chromosome.

HOW DO WE KNOW HOW ELEVATED THE RISK OF T21 IS?

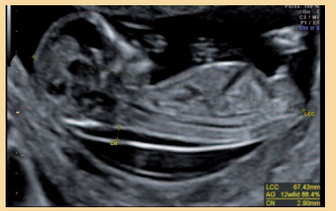
Low or high risk can be evaluated by a risk calculation, performed at the expectant mother's request.

A) **In the first trimester of the pregnancy**, the calculation is done on 3 items:

1. **The mother's age**: the older she is, the higher the risk.



2. **The measurement of the nuccal pad** (called **nuccal translucency**). During the first trimester ultrasound examination, the thicker the nuccal pad the higher the risk of T21. The measure of the nuccal thckness is made by a trained ultrasonographer (certified in a prenatal network of his area).



3. **A blood test**, usually performed after the ultrasound exam, to dose the mother's blood level of serum markers (blood proteins) in a specialized laboratory under control of the regional health agency.

The combination of these three values give an estimation of the risk of the foetus to be T21.

B) **During the second trimester**

If the screening test has not been performed during the first trimester, it is still possible until the 18th week of amenorrhea (weeks without menses).

The calculation then includes:

1. the age of the expectant mother;
2. the measure of the nuccal pad during the first trimester, if done by a certified ultrasonographer,
3. the measurement of different serum markers (blood test) specific to the second trimester.

Also note: a malformation may be detected during any of the three trimesters of pregnancy and represents a higher risk of T21.

HOW DO WE INTERPRET THE RISK?

The results are delivered to and interpreted by the prescribing doctor. This produces an estimated risk for the foetus to be T21, not a certainty.

If the risk is higher than 1/250 (1 out of 250 cases), it is considered to be an elevated risk.

Example of an elevated risk: 1/50 (1 out of 50 cases) This signifies that the foetus has 1 chance out of 50 (2% risk) to be T21. In other words, in 49 out of 50 cases (98% of the cases), the foetus is not T21.

If the risk is under 1/250, it is considered as low.

Example of a low risk: 1/1000. This would mean that the foetus has 1 chance out of 1000 (0.1%) to be T21, and so in 999 out of 1000 cases (99.9%) the child would not be T21.

In conclusion, remark that this calculation is not perfect. It will alarm 5% of pregnant women for whom the risk is called elevated, even though the grand majority of them will not have a T21 child.

On the other hand, at the opposite, in some rare cases, a risk that will have been estimated as low (less than 1 out of 250) will nonetheless be followed by a child born with T21.