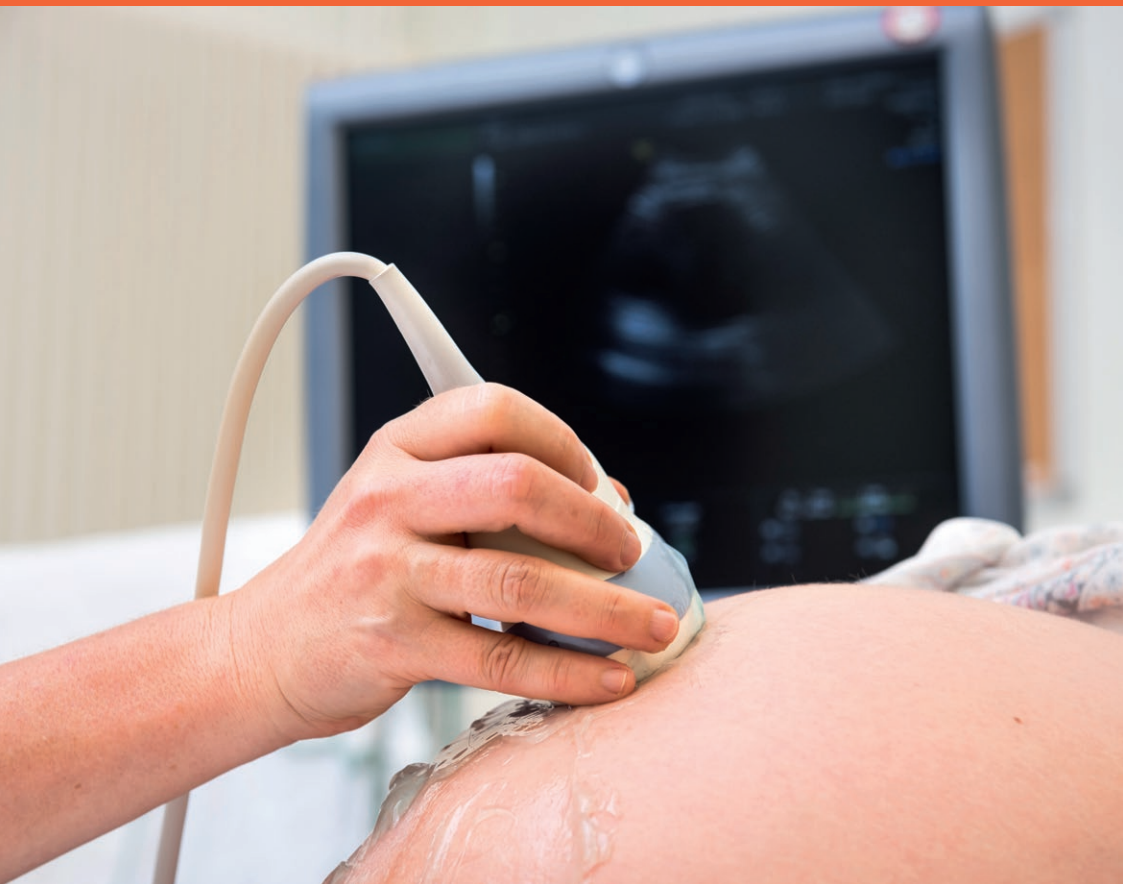
Anglais

PRENATAL SCREENING
FOR TRISOMIES 21, 18 AND 13

Screening tests and diagnostic examinations



Hôpitaux Universitaires Genève

Introduction

The aim of this leaflet is to explain to you what the double early screening test for trisomies 21, 18 and 13 consists of. It is offered to every pregnant woman in the first trimester of the pregnancy, between the 11th and 14th week. It is used to determine the risk of chromosome abnormalities in the unborn child. This test is not compulsory and is only carried out with your consent. According to the risk assessed, more extensive tests may be discussed.

What does the test consist of?

It consists of an ultrasound, performed at about 12 weeks, and a maternal blood analysis which may be performed as early as 9 weeks.

Nuchal translucency

The most important element for calculating the risk is the ultrasound measurement of the thickness of the nape of the neck of the embryo which forms a zone called nuchal translucency. The risk increases with the thickness of this.

Furthermore, a thick nuchal translucency may be an important index for other fetal abnormalities, but may also be observed in children where the development is totally normal.

Maternal serum markers

The maternal blood sample measures a hormone, free β-hCG, and a protein, PAPP-A. These substances are found in the blood of all pregnant women, but with a different level for a fetus which is a trisomy 21, 18 or 13 carrier.

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INFO

If this test is not performed during the first trimester, a test can be performed during the second trimester. It is based on the age of the future mother and two parameters of maternal blood – AFP (alpha-fetoprotein) and β-hCG. In this case only the risk of trisomy 21 is assessed.

How to interpret **the results?**

INFO

The risk of trisomy 21 increases with age. It is 1 case per 1600 births for mothers aged 20, 1 per 900 up to age 30, 1 per 380 up to age 35 and 1 per 20 births at age 45.

The age of the future mother, combined with the nuchal translucency and blood tests, enables a calculation of the statistical probability of having a fetus which is a carrier one of these three trisomies.

A low risk

The result is considered as low when the risk of trisomy falls within 1/1001 and 1/20,000 births. This means there would have to be more than 1000 births to find one case of trisomy. For these future mothers, the risk is really low. No further tests are required apart from the normal ultrasound monitoring.

An increased or intermediate risk

A result is considered as "increased" or "intermediate" when the risk of trisomy is higher than 1 in 1000 (1/500, 1/150, 1/50, etc.). This result does not prove the presence of a trisomy.

According to the risk level, other tests can be offered on a case by case basis:

- A non-invasive prenatal test (Genatest) in the case of an intermediate risk higher than 1 in 1000 and if the ultrasound is normal.
- An invasive prenatal diagnostic (chorionic villus sampling, amniocentesis) in the case of high risk (combination of a risk higher than 1 in 1000 with other criteria such as ultrasound abnormalities, multiple pregnancies, very high nuchal translucency, etc.).

What to do if you are at **increased risk?**

Depending on the risk of trisomy 21, 18 or 13, two types of additional tests are possible.

A non-invasive prenatal test (Genatest)

In the case of intermediate risk, the Genatest is offered for screening for trisomies 21, 18 and 13 in the fetus, except if the ultrasound reveals a deformity or increased nuchal translucency. In these specific situations, a diagnosis by means of chorionic villus sampling or amniocentesis is offered immediately.

Performed by taking a maternal blood sample, the Genatest analyses the genetic material of the fetus (its DNA) released by the placenta into the blood of the pregnant woman. This test does not present any immediate risk for the pregnancy. The results are available within 5-10 working days after the blood sample was taken.

If the test indicates that the fetus is very probably a carrier of the trisomy concerned, the result must be confirmed by an invasive diagnostic test (chorionic villus sampling or amniocentesis).

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FIND OUT MORE

For more information, please read *Genatest, a non-invasive prenatal test.*

A prenatal diagnostic

To find out specifically if the fetus is affected or not by trisomy 21, 18 or 13, the doctor performs a choriocentesis or an amniocentesis:

- Choriocentesis, or chorionic villus sampling (CVS), consists of removing a minuscule sample of the future placenta by means of a needle guided by ultrasound control.
- Amniocentesis consists of removing a small quantity of amniotic fluid with a needle, under ultrasound control.

These two examinations enable all fetal chromosomes to be analysed.

These are invasive procedures which may, in rare cases (0.2 to 0.5%), trigger a miscarriage.

The initial results are available within 48 hours and the definitive results after two weeks.

INFO

You will be offered a specific consultation if the tests of the first trimester of your pregnancy show an intermediate or high risk in order to provide some explanations on the diagnostic techniques and to explain the choice of test to you.

Practical **information**

DGO • Dépistage prénatal des trisomies 21, 18 et 13 • Août 2017

Contact us

022 372 42 29
www.hug-ge.ch/gynecologie-obstetrique

Invoicing

The screening test in the first trimester of the pregnancy (ultrasound and maternal blood analysis) and the costs of the chorionic villus sampling or amniocentesis are assumed by the compulsory health insurance.

The non-invasive prenatal test is reimbursed within the basic benefits of LAMal if the risk of trisomy is higher than 1 in 1000.

Division of Obstetrics

Fetal Medicine and
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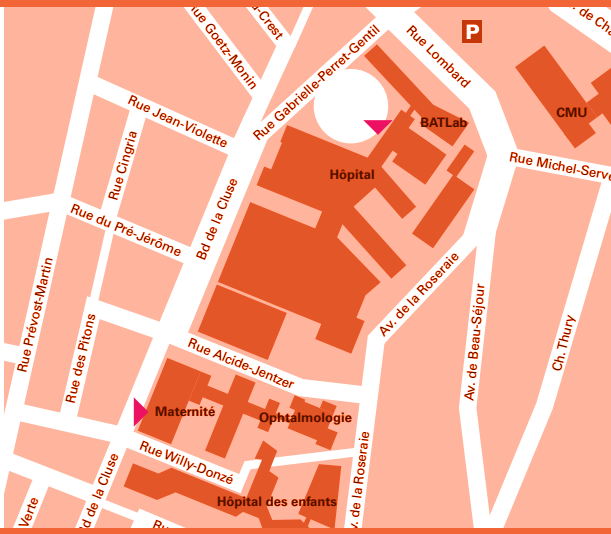
How to find us

Tram 12 and 18,
"Augustins" stop
Bus 35,
"Maternité-Pédiatrie" stop



This brochure, tested with patients, has been compiled by the division of obstetrics in collaboration with the information group for patients and relatives (GIPP) of the HUG.