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1TT and NIPT

Firsttrimester-Screening and Non-Invasive-Prenatal-Testing

Information for the patient

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The classical first trimester screening

In addition to ultrasound examinations by your gynecologist, blood will often be drawn during your pregnancy to perform certain analyses in the laboratory. For example, blood type is determined, hormones and immunity status is checked and much more. An additional test, which is offered to every pregnant woman, is the so-called first-trimester test (1TT).

This is a screening test, which serves to discover pregnancies with an increased risk for the presence of a trisomy so that further tests can then be offered to you.

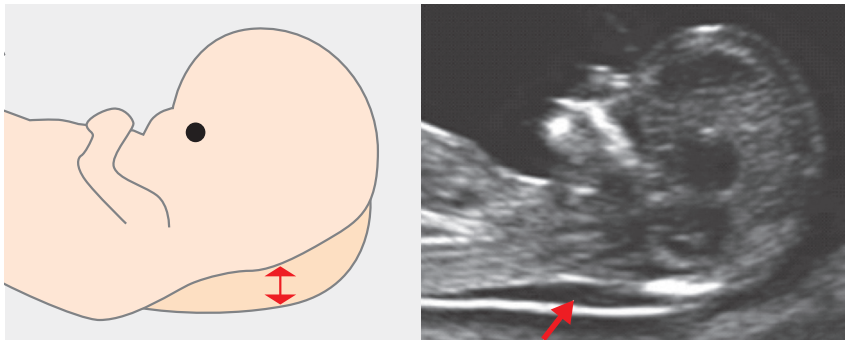
What is a trisomy?

Humans have 46 chromosomes in each cell, including 23 chromosomes inherited from the father and 23 from the mother. They carry genetic information of a person. Trisomy is a chromosomal disorder that occurs when a certain chromosome is present three times instead of two times. This excess of genetic information has serious consequences.
Trisomy 21 (Down syndrome) is the most common trisomy. It is accompanied by mental retardation and often other organs such as the heart are also affected. With increasing age of the mother, the risk of pregnancy with trisomy 21 is greater. In a 40-year old woman, the age risk is 1:100 (1%), while a 20-year-old woman has a smaller risk of 1:1500 (0.07%). The trisomies 13 and 18 are much rarer than the trisomy 21. These chromosomal disorders are associated with severe malformations, which makes the survival of the fetus impossible in most cases.

Basics of the first trimester screening test

Ultrasound and biochemistry

The first trimester screening test consists of a combination of an ultrasound examination and a blood withdrawal, which are both carried out in the first third of pregnancy. Besides many other aspects, the gestational age is also determined by ultrasound. This is done by measuring the fetus from the crown to the tailbone (crown-rump length). Based on this measurement, the exact week and day of the pregnancy can be deducted. Determining the week of pregnancy is much more accurate than a calculation after the last menstrual period. Thereafter, the nuchal translucency (NT) is measured. Nuchal translucency refers to a collection of fluid under the neck skin of the fetus. In fetuses with trisomy 21 the NT is increased.



Nuchal translucency in a diagram and an ultrasound image

Risk calculation

In maternal blood, two markers are studied in the laboratory, which are formed by the placenta: PAPP-A (pregnancy-associated plasma protein A) and the free β-HCG (Free Beta Human Chorionic Gonadotropin). With a special computer program, the individual risk of the presence of trisomy in the ongoing pregnancy is calculated using the combination of maternal age, history (smoking, ethnicity, diabetes, any trisomy in the previous pregnancies, weight), nuchal translucency, concentration of the markers and week of pregnancy. This risk is termed the overall risk or combined risk. The background risk is termed as basis risk or age risk based on the maternal age.

Significance

Most first trimester tests are unremarkable, they do not indicate an increased risk for your pregnancy. However, the test can also show that the risk is increased. Suspicious findings do not mean that your unborn child has a trisomy as the first trimester test is merely a screening method and not a definitive, diagnostic test. In case of a suspicious first trimester test, one must discuss with the physician the necessity of further testing (NIPT = Non-invasive prenatal test, chorionic villus sampling or amniocentesis).

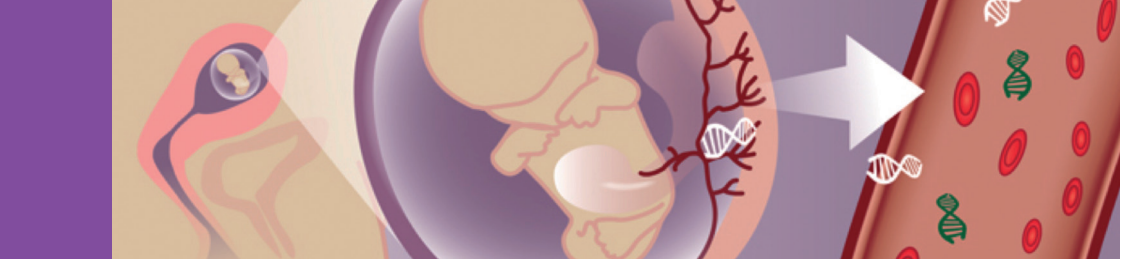
The first trimester test can help to discover about 85–90% of all trisomy 21 pregnancies, i.e. 10% of all pregnancies with trisomy 21 have normal first trimester test results. Anomalies and malformations other than trisomies 13, 18 and 21 cannot be detected by using the first trimester screening test.

Special conditions (twins, IVF, egg donation)

Even in twin pregnancies and IVF pregnancies, the first trimester test can be used in the case of egg donation, the age of the donor on the day of egg retrieval has to be considered.

Cost assumption

The first trimester screening test is covered by health insurance. A first trimester screening test with a risk greater than 1:1000 (e.g. 1:600) is a prerequisite for the cost assumption of NIPT (see below) by the health insurance company.



The non-invasive prenatal test (NIPT) harmony™

Principle

The placenta releases tiny bits of genetic material (DNA) of the unborn child in your blood. They make up for about 10% of cell-free DNA in maternal blood. With NIPT (non-invasive prenatal test), this cell-free child DNA is examined and the trisomies 21, 13 and 18 can be ruled out. The determination of sex is also possible. In addition, rarely occurring maldistribution of sex chromosomes (e.g. Turner syndrome) can be determined.
To make this possible, these bits of DNA are propagated, analyzed and assigned to the corresponding chromosomes. With bioinformatic methods, it is determined whether your pregnancy is characterized by trisomy 21, 13 or 18 and how the sex chromosomes X and Y are distributed.

Time

The NIPT (Harmony test) can be performed from the 10th pregnancy weeks onwards (10+0). For a cost-takeover by the insurance companies, a maternal serum screening (first trimester test) must have been carried out in advance and the overall risk must be greater than 1:1000 (e.g. 1:800).

Gender determination

The determination of gender is possible using the Harmony test. For legal reasons the gender may only be disclosed after the 12th pregnancy week (12+6). For the performance of the NIPT the determination of the gender is not necessary.

Special cases (e.g. twins, IVF, egg donation)

It is possible to perform NIPT even in case of IVF pregnancies or twin pregnancies. For the purposes, it is important to know whether egg donation has taken place.

Short duration till availability of results

It takes about 3 to 4 working days to get the result.

Possible problems

In a few cases, sufficient DNA of a child cannot be obtained from the first blood sample and therefore a second one may be needed.

Test accuracy

As much as 99.9% of all cases of trisomy 21 are correctly identified by the test. However, there are rare cases where false positive or false negative results may be obtained due to atypical placental development. Therefore abnormal findings must be confirmed by chorionic villus sampling or amniocentesis. If your child has disorder different than the trisomies mentioned above, the NIPT will not detect it.

Contact persons

Dr. Ute Wiedemann
T 044 269 99 99

Dr. Dieter von Au
T 044 269 99 99

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MEDIZINISCHE LABORATORIEN Dr. F. KAEPPELI AG
Wolfbachstrasse 17, Postfach, 8024 Zürich
Telefon 044 269 99 99, Fax 044 269 99 09
info@medica.ch www.medica.ch

www.medica.ch