

## TRIPLE TEST

### WHAT IS THE TRIPLE TEST?

The triple test is an analysis of substances in the serum of a pregnant woman. It is performed between the completed 14<sup>th</sup> and 20<sup>th</sup> week of pregnancy and consists of the determination of three substances:

- AFP (alpha-fetoprotein)
- $\beta$ -hCG (human chorionic gonadotropin)
- E3 (free estriol)

On the basis of these three measured substances - in addition to the maternal age and other factors – the probability of whether the unborn child has a Down syndrome (trisomy 21) or not, can be estimated. Additionally, elevated AFP values can indicate malformations with a structural defect that affects skin permeability such as a neural tube defect and an omphalocele (spina bifida, “open back” and “open venter”).

### WHAT DOES THE TRIPLE TEST RESULT INDICATE?

The risk of having a child with Down syndrome depends on the maternal age. The risk of a 20 year old pregnant woman is about 1:1,667, this means that one individual out of 1,667 women of this age will bear a child with Down syndrome. The risk increases with maternal age and for a 35-year old woman it is 1:380.

The triple test can estimate the risk for the presence of a trisomy 21 in an unborn child. This risk estimation is more exact than a risk estimation based on maternal age alone.

The only method which can prenatally clearly exclude the presence of a trisomy 21 is the analysis of the chromosomes from the cells of amniotic fluid. However, to perform such a chromosome analysis it is necessary to take a sample of amniotic fluid (amniocentesis). Amniocentesis is associated with the risk of abortion caused by this intervention. This risk is given to be between 1:100- 1:200 (0,5-1%).

If the result of the triple test reveals a risk, which is higher than the risk of losing the unborn child through an amniocentesis, this method as an additional examination is discussed and offered. Usually amniocentesis is offered if the risk for trisomy 21 exceeds 1:270 -1:380.

After genetic counselling and a detailed ultrasonography, a sample from the amniotic fluid can be taken in order to examine the child's chromosomes.

The triple test only gives information about the probability of the unborn child having a trisomy 21.

The triple test is a method of risk estimation and is therefore not a diagnostic, but a screening method.

An exact prenatal diagnosis of trisomy 21 can only be made with the analysis of the fetal chromosomes.

### WHEN IS IT REASONABLE TO PERFORM A TRIPLE TEST?

According to the maternity guidelines in some countries, pregnant women of the age of 35 years and over must be informed about the possibilities of prenatal diagnostics. Pregnant women of this age, who do not favour the performance of an amniocentesis, which is accompanied by a rate of abortion of 0,5 - 1 %, can first have a triple test performed to obtain a risk estimation.

About 70 % of all children with trisomy 21 are borne by women who have not yet reached the age of 35. Since the probability for the occurrence of a trisomy 21 in this age group is smaller than the risk of losing a healthy child by an amniocentesis, this invasive interference and with it the diagnosis are rarely performed in women under 35 years of age. About 70 % of the cases of trisomy 21 can be recognized by the triple test.

Neural tube defects can be recognized with relative certainty by the determination of the AFP values.

## FIRST TRIMESTER SCREENING

With the first trimester screening, a combination of ultrasound examination and measurement of the values of two substances in the blood, the risk for certain chromosome alterations can be measured between the 11<sup>th</sup> and the end of 13<sup>th</sup> week of pregnancy. At this developmental stage, chromosomal alterations in an unborn child can often be recognized by an accumulation of liquid in the area of the neck (Nuchal Translucency: NT).

The measurement of nuchal translucency presupposes careful examination and appropriate experience of the examiner.

With the data of the ultrasound examination in combination with the age of the pregnant woman and two blood values, the PAPP- A and the free  $\beta$ -hCG, an individual risk for the most frequent chromosome abnormalities of the unborn child can be estimated.

As with the triple test this is a method for risk estimation. It can not definitely be determined whether the child has a trisomy 21 or not. The detection rate for trisomy 21 is described to be 85 %.

The exclusion of a chromosomal alteration or its diagnosis can only be made with the analysis of the fetal chromosomes. For this chorionic villi biopsy or amniocentesis is necessary.

An advantage of the first trimester screening in comparison to the triple test can be the fact that it is performed at an earlier stage of pregnancy. A statement on neural tube defects ("open back") or other malformations (such as "open venter") cannot be made. Such malformations can be detected with an analysis such as triple test or by the measurement of the AFP value only.

The highest detection rate regarding malformations of the child are reached using a combination of the first trimester screening and the triple test. According to latest information this combination has a detection rate of approx. 94% (1, 2, 3).

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## REFERENCES

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## ADDITIONAL DIAGNOSTICS DURING PREGNANCY

TRIPLE TEST

FIRST TRIMESTER SCREENING



<http://www.users.bigpond.com/aussieshutterbug/00-Mirrors.htm>