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Information about antenatal investigations

Antenatal investigations:
We will discuss various screening methods with you as part of your antenatal care.
These tests (nuchal translucency measurement, maternal blood test, amniocentesis, placental biopsy) are performed to detect possible malformations or chromosomal disorders.
It is advisable to be well informed about the options for detecting malformations, and to have an idea of which examinations you would like to have.

Non-invasive examinations for risk evaluation with first trimester screening:
First trimester screening (FTS) is based on a combination of ultrasound examination and maternal blood test.
Timeframe: 12th to 14th week of pregnancy.

Ultrasound is used to assess fetal vitality, determine gestational age by measuring the crown-rump length, and measure nuchal translucency (NT).
Nuchal translucency is due to a collection of fluid under the skin at the back of the baby's neck. A larger-than-average clear space at the back of the neck may indicate a chromosomal disorder (e.g. Down's syndrome) or congenital organ deformation (e.g. a heart defect).

The maternal blood sample is used to test for two substances:
PAPP-A (pregnancy associated plasma protein-A) and free beta-hCG.
Individual risk of a chromosomal disorder or malformation can then be calculated using a combination of maternal age, nuchal translucency and the results of the blood test.

What does first trimester screening tell us?
In the vast majority of cases, FTS provides reassurance that your baby has a low risk of trisomy or serious malformation. In a few cases, however, FTS may indicate an increased risk of such an abnormality.
Even so, this does not mean your child has a problem: the examination is a screening test, not a definitive diagnostic test.

If screening indicates an increased risk, it is advisable to obtain detailed genetic counselling to discuss what other diagnostic examinations should be considered (genetic tests on maternal blood, invasive prenatal diagnosis).

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How precise is first trimester screening?
Over 85% of children with trisomy 21 have an abnormal FTS result. Babies with other chromosome abnormalities or malformations will also frequently return an abnormal FTS result. But it is important to understand that a normal FTS result is not a guarantee that your child does not have a problem: this is a screening test (for evaluation of risk), not a diagnostic test.

Genetic tests on maternal blood:
Analysis of the maternal blood gives a very strong indication (~ 99.9%) of whether or not your unborn child may have a chromosome disorder (essentially trisomy 13, 18 and 21). This method uses small quantities of fetal genetic material (DNA) present in the maternal blood.
With the change in health insurance policy that came into effect in July 2015, the cost of this test (chromosomes 13, 18 and 21 only) is now covered if the result of first trimester screening is abnormal (increased risk of >1:1000). You can of course also elect to have this test (e.g. due to age or the need to be sure). In this case, the test is considered a 'self-payer service' and we will invoice you directly.

Invasive antenatal diagnosis with chorionic villus sampling (CVS) and amniocentesis:
Chorionic villus sampling is an antenatal test in which a small sample is removed from the placenta for testing. The sample is obtained using a thin needle inserted through the mother's abdominal wall.
Amniocentesis is a process in which amniotic fluid is sampled using a needle inserted through the mother's abdominal wall. Chorionic villus sampling can be performed from the 11th week of pregnancy, amniocentesis from the 16th week.

Both methods provide a means of examining the number and structure of the chromosomes. A provisional result will be available within 24–48 hours; the definitive final report can take up to 2–3 weeks. Both CVS and amniocentesis carry a risk of miscarriage of approximately 0.5-1%. These invasive methods should therefore only be considered if an increased risk of trisomy has been identified.