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Praxis für pränatale Diagnostik München

Nuchal translucency scan / first trimester screening in 12th – 14th week of pregnancy

Reasons for this examination
These day many women and couples want an individual risk assessment for chromosomal abnormalities, heart defects or other malformations.
One of the most common developmental defects is Down's Syndrome, where there is an additional copy of chromosome 21 (trisomy 21). The risk of Down's Syndrome does indeed increase with the age of the pregnant woman, but as around 70 % of children suffering from Down's Syndrome these days are born to women younger than 35, young patients especially may also want to assess the individual risk for their child. On the other hand, for women older than 35 with a low individual risk, a genetic examination e.g. by means of amniotic fluid puncture with the corresponding complication rate, is often avoidable.

In addition, many malformations – particularly of the head and brain, arms and legs, the bladder, gap formations in the abdominal wall and spine and defects of the diaphragm – can already be detected during the first trimester (early organ ultrasound). Furthermore, some of the serious heart defects can already be detected.

The examination that we offer thus goes far beyond "trisomy 21 screening". For this reason, a test on the mother's blood for trisomy 21 (so-called non-invasive prenatal test - NIPT) can by no means replace first trimester screening either.

What is first trimester screening?
The foetal nuchal fold is a small accumulation of liquid under the neck's skin, which can normally be measured by ultrasound in the period between the 11+0 and 13+6 week of pregnancy. A widening of this nuchal fold can occur in children with chromosomal abnormalities, but also in case of other malformations such as heart defects. A combination of various measurements goes into the result of the examination:

- measurements from the ultrasound scan
- measurements from a blood test (biochemical)

The age of the pregnant woman, the size of the child and the thickness of the nuchal fold can be used to calculate an individual risk of chromosomal abnormalities, e.g. trisomy 21 (Down's Syndrome), but also trisomy 13 (Patau's Syndrome) and trisomy 18 (Edward's Syndrome). Other sonographic attributes, such as the nasal bone, blood flow properties or the heart rate, can also be integrated in the analysis. The decision for further or invasive diagnostics (puncture) can thus be made on a more individual basis.

The risk of other, non-chromosomal malformations (e.g. heart defects) also correlates to the width of the nuchal fold, meaning that targeted diagnostics can be arranged. The discovery rate of Down's Syndrome is around 75 %. This discovery rate can

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be increased by a further 10-15 % to 85-90 % when combined with a test on the mother's blood. Two substances formed in the placenta, PAPP-A and free beta HCG, are measured in the pregnant woman's blood in this respect.

Trisomy 21: Detection rate of the different methods:

Age alone (> 35 years old)	30 %
Age + nuchal translucency scan	75 %
Age + nuchal translucency scan + maternal blood test	85-90 %

Studies have shown that in case of a later examination with blood analysis in approx. the 11th week of pregnancy and an ultrasound scan in approx. the 13th week of pregnancy, compared to simultaneously analysing these components, the highest rate of detection can be achieved with ultrasound screening for chromosome anomalies. If at all possible, this form of ultrasound procedure should therefore be pursued.

Furthermore, it is thereby possible to incorporate the so-called second step parameters (nasal bone, ductus venosus, tricuspid valve) from advanced screening in the risk analysis according to the FMF London guidelines. If no clear allocation to the high or low risk group is possible by age, biochemical testing and nuchal translucency measurement, these additional markers may enable a further diagnostic risk differentiation. Besides an early organ and heart ultrasound, these parameters are studied by us in addition as part of first trimester screening. The overall result can be discussed immediately after the ultrasound scan.

Benefits and limits of the examination

First trimester screening involves no examination risk for the child. The examination enables a risk assessment and can thus be used to help decide for or against so-called invasive diagnostics, such as taking amniotic fluid (amniocentesis) or placenta tissue (chorionic villus sampling) or also the non-invasive prenatal tests (NIPT). Inconspicuous results are no guarantee for a child with no genetic disorders or other malformations. It is only possible to rule out a chromosomal abnormality with certainty by performing an amniocentesis or chorionic villus sampling.

The risk assessment from the nuchal translucency screening and the decision linked to this for or against a further examination during pregnancy must be incorporated in an individual consultation, which is carried out by us before and after the examination.

The test available since 2012 for trisomy 21, 13 and 18 (NIPT) from the mother's blood cannot replace the first trimester screening, as it is unable to make a statement about other chromosomal defects, genetic disorders or foetal malformations. In certain situations it can be useful as a complement to the first trimester screening.
We can advise you about this as part of the ultrasound scan. Before the test can be carried out, a consultation from a specialist for human genetics is also necessary.

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Pre-eclampsia screening
Measuring the blood flow of the arteries in the womb represents an extension to first trimester screening. From this, combined with the mother's blood pressure and a blood count (PAPP-A), the likelihood for the occurrence of pre-eclampsia (high blood pressure, proteinuria and water retention during pregnancy, so-called toxaemia of pregnancy) can be determined. This may be useful, for example if pre-eclampsia occurred in a previous pregnancy or there are other risk factors. The risks of the occurrence of a placenta under-function can also be assessed. If detected early, prevention measures and appropriate checkups can be arranged.

Costs of the examination
First trimester screening is an individual medical service and is currently not reimbursed by the statutory health insurance schemes. You will be given a detailed private liquidation according to the scale of charges for doctors (GOÄ). You can find further details on our website pages at www.praenatalschall.de or by calling us on the phone.