



Patient Information

First-trimester ultrasound (nuchal translucency measurement)

Dear expectant mother,

You have attended our practice today to undergo measurement of the nuchal translucency of your unborn child. Before the examination, it is important for you to have some background information on the purpose, possibilities or limitations, and potential problems associated with this examination, which the following pages should provide. This information serves as the basis for the pre-procedural medical discussion prior to examination.

Background:

It is known that the risk of a child developing chromosomal disorders (misdistribution of the carriers of genetic material) increases with the age of the expectant mother. The most commonly occurring chromosomal disorder is Down's syndrome, in which chromosome 21 is created 3 times instead of twice, for which reason it is also known as trisomy 21. This is followed by trisomy 18 (chromosome 18 created three times, Edward's syndrome) and trisomy 13 (chromosome 13 created three times, Patau's syndrome). Due to this rise in chromosomal disorders with increasing age, in Germany all expectant mothers who are 35 or older at the time of birth are offered an amniotic fluid test to determine chromosomes (age indication), in accordance with maternity guidelines. However as most expectant mothers over the age of 35 give birth to healthy children, and as around half of all children with Down's syndrome are born to women under the age of 35, it must be stated that age alone is only one limitedly usable parameter for or against a decision to undergo an amniotic fluid test.

In recent years, it has emerged that many foetuses with chromosomal disorders exhibit special features (called markers) or organic malformations in ultrasound examinations which are sometimes also detectable in early stages of pregnancy. The detection of such features is not conclusive proof, but indicates an increased risk of the existence of a chromosomal disorder, while the absence of such features reduces the risk. The most well-known feature in early pregnancy that can indicate a chromosomal disorder is increased nuchal translucency, which is why this examination is commonly also referred to as "nuchal translucency measurement".

However, this name is no longer accurate for the examination as the rapid advances in technology and expanding experience of examiners now also makes it possible to identify most severe organic malformations at this early stage of pregnancy. For this reasons, we now prefer to refer to the "early exclusion of malformations" or first-trimester ultrasound.

When is the examination performed?

This type of examination is restricted to a relatively narrow time window, within which it can form the basis for very reliable statements. This time window ranges from the 11th week of pregnancy to the 13th week of pregnancy. Outside this timeframe, risk analyses based on the measurement of nuchal translucency are not permitted.

How is the examination performed?

The examination is an ultrasound examination which presents no risk to the pregnancy and is usually performed via the abdominal wall. If visibility conditions are very unfavourable (thick abdominal wall, awkward position of the embryo or uterus), it may be necessary to perform the examination transvaginally (via the vagina).

What is examined ?

The examination entails an inspection of the developmental state of the embryo and an initial organ examination, if possible at this early gestational age. This makes it possible to identify or exclude most serious malformations.

With regard to the risk situation for chromosomal disorders, different features of the child are determined that can be included in the risk determination:

- *Nuchal translucency (thickness of nuchal fold):* The nuchal translucency is a structure that can be detected in every foetus at this gestational age, but its thickness may differ. In general, the risk of the existence of a chromosomal disorder increases with the thickness of the nuchal translucency. It is important to know that a thicker nuchal translucency does not indicate the existence of any disease per se. An embryo with a thicker nuchal translucency is not ill as a result, and healthy children may later have a thickened nuchal translucency also.
- *Nasal bone:* Since it is known, particularly in the case of foetuses with Down's syndrome, that the nasal bone is often undeveloped or very weakly developed at this gestational age, the visibility of the nasal bone is examined. If the nasal bone is missing, the risk of the presence of Down's syndrome is increased.
- *More recent additional markers:* Recently, additional parameters have been included in the risk determination that further increase the validity and reliability of the calculation. These include the blood flow in the ductus venosus (vessel connecting the umbilical vein and the inferior vena cava of the child) and examination of the tricuspid valve (heart valve between the right atrium and the right ventricle) for venous return of the blood. These parameters are often abnormal in children with Down's syndrome. Here again, it is important to know that abnormal blood flows can also be observed in completely healthy children. These additional parameters cannot be fully depicted in every examination situation and it will not be possible to include these in all examinations for risk determination.

Blood test (biochemistry): In order to further increase the validity of risk determination, the determination of the concentration of two substances in the blood of expectant mothers can be used. These are the pregnancy hormones (β -HCG) and the PAPP-A (protein formed only during pregnancy). The level of concentration of these substances in the blood and the ratio of the two substances to each other are analysed and so are included in the risk determination.

Summary:

Using the **age of the mother**, the **thickness of the nuchal translucency**, possibly also the visibility of the nasal bone, the partial inclusion of the new markers and the **biochemistry**, a specific risk of the presence of a chromosomal disorder can be determined for your personal situation. The probability of identifying an embryo with Down's syndrome in this way is over 90%.

This early ultrasound can also be used to exclude more serious malformations at an early stage. However, this ultrasound examination does not replace the more detailed ultrasound (detailed diagnostic) normally performed after the 20th week of pregnancy.

Please consider the following points:

This examination can never safely exclude the presence of a malformation, disease or chromosomal disorder. The first-trimester ultrasound cannot therefore give you any guarantee of a fully healthy child. Only a risk assessment is possible for a chromosomal disorder, not a definitive diagnosis. A chromosomal disorder can only be safely excluded with an invasive examination such as an amniotic fluid test.

Most examinations reveal no abnormalities, which can help to relieve anxieties and contribute to an uneventful pregnancy. This can also be helpful when making decisions about further examinations (e.g. amniotic fluid test).

However, if abnormalities are detected in the examination, this often leads to upset and conflict, which in some cases can be significant. In such an eventuality, we will inform you promptly and support you, in consultation with other physicians also (e.g. human geneticists, paediatricians or paediatric surgeons). We also advise you to seek psychosocial counselling in these situations particularly and can arrange contact at your request.

Genetic counselling:

In addition to this patient information, the Gene Diagnostics Act (GenDG) stipulates that expectant mothers must be offered genetic counselling before a first-trimester ultrasound and after the examination results are obtained.

Genetic counselling before a genetic test pursuant to GenDG includes:

Answering your personal questions,

- Evaluation of existing medical findings or reports on findings,
- Examination-based compilation of abnormalities in your personal and family health prehistory (anamnesis),
- Provision of information on the need for a genetic test arising from your questions or prehistory, and information on the possibilities, limitations and material extraction-associated risks of the examination procedure being explained,
- Estimation of the genetic risks including discussion of the significance of all information for your life and family planning, and possibly for your health,
- Support options available for dealing with the physical and psychological stresses caused by the examination and its results,
- Assessment of the need for detailed genetic counselling by a specialist in human genetics.

We usually provide this counselling directly in conjunction with the pre-procedural discussion and the communication of results, but if you feel that you have already received sufficient information and counselling, you can waive further genetic counselling in writing.

Please tick here to indicate whether you wish further counselling, want to waive this, or have already received genetic counselling.

Consent:

I have already received genetic counselling in preparation for the first-trimester ultrasound:

Yes: No:

I wish to receive further genetic counselling before the examination:

Yes: No:

I wish to receive genetic counselling when the examination findings are available

Yes: Only if findings are abnormal: No:

Communication of examination results:

The physician who performed the first-trimester ultrasound examination will inform you of the examination results.

Other people (partner, attending gynaecologist) can only be informed with your express written consent.

You have the right not to acknowledge the results of the examination or parts thereof and to have these destroyed.

I consent to disclosure of the results of the ultrasound examination to my

Gynaecologist _____ at:

Other physicians: _____

Other people: _____

Personal questions: _____

Physician's notes: _____

I received detailed information about the planned examination in the pre-procedural discussion with
Dr. _____

All questions I feel were important regarding the type of examination and its significance were discussed and answered for me comprehensibly. I feel well informed, have no further questions and consent to the examination. I do not need any further time for consideration.

You can revoke all of your consents at any time with effect for the future.

Place/date Signature of expectant mother

Place/date Signature of physician