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Praxis für pränatale Diagnostik München

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Amniocentesis / amniotic fluid puncture

Reasons for an amniocentesis – indications:
The term "invasive diagnostics" refers to procedures used to take cell's from the child for further examination. Whilst developmental defects and organ anomalies are detected with the pure ultrasound scan, amniocentesis is used to detect chromosomal abnormalities, meaning changes in the number and structure of the chromosomes. Normally every person has 46 chromosomes in each cell of their body, which carry genetic information.
Around the conception stage, random defects can occur during division. The most familiar of these random errors is trisomy 21, Down's Syndrome. In every pregnancy there is a certain degree of risk that the child could have a random defect of this kind. This risk increases the older the pregnant woman is (age indication).

Reasons for an amniocentesis may be conspicuous findings from the screening in the first trimester (measurement of nuchal translucency and the serum parameters beta hCG and PAPP-A), from the ultrasound scan of the foetus or in case of inherited and frequently occurring disorders (molecular genetic diagnosis). The expectant parents' wish for the utmost certainty can also be an indication for amniocentesis.

Although heart defects and other structural malformations are often found alongside many chromosomal abnormalities (such as Down's Syndrome), in some cases the organ diagnostics reveal no noticeable abnormalities. The risk of a chromosomal abnormality is indeed reduced if the ultrasound scan is normal, but this cannot be definitely ruled out with ultrasound. It is only possible to rule out a chromosomal abnormality with certainty by means of invasive diagnostics. The blood test for trisomy 21, 13 and 18 from the mother's blood, which has been available for a few years, is unable to fully answer many questions resulting from such situations and therefore does not replace invasive diagnostics in all cases. This blood test may be helpful in certain situations, however, and we will be happy to advise you about this face to face.

The actual hereditary conditions are based on genetic changes, which have either just emerged or were already present in one or both parents. Examples of these kinds of conditions are cystic fibrosis or other metabolic disorders. With the majority of possible problems, the ways of detecting a condition and also any consequences of it must be clarified during a human genetic consultation before carrying out invasive diagnostics. If no cases of disorders have previously occurred in the families, prenatal screening tests for the presence of such conditions are only seldom possible.

Examination process:
Before each amniocentesis, a detailed ultrasound scan is performed. Under constant ultrasound guidance, a very thin needle is inserted into the amniotic cavity in such a way that any injury to the child is extremely unlikely. The puncture process takes place under sterile conditions and without a local anaesthetic.

The intervention only lasts a few minutes. Approx. 15 ml of amniotic fluid is extracted and cultures set up of the baby's cells contained in the fluid. As these cell cultures need time to grow and multiply, the result of the amniotic fluid analysis is only available approx. 2 weeks after the procedure. In particular cases or on request, an additional quick test (FISH analysis) can be carried out, which can rule out or verify the three most common chromosomal abnormalities - trisomy 21 (Down's syndrome), trisomy 18 (Edward's Syndrome) and trisomy 13 (Patau's Syndrome) - as well as defects in the chromosomes that determine the child's sex,

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with a high degree of certainty just one day after the examination. Thus nearly 85 % of pathological diagnoses can be quickly diagnosed.

Determining a protein (alpha fetoprotein) and an enzyme in the amniotic fluid in addition enables gap formations in the spine (Spina bifida) to be detected with a high degree of certainty. This is not possible with chorionic villus sampling. Gap formations can also be ruled out with a high degree of certainty by detailed malformation diagnostics, however, meaning that the significance of the AFP diagnosis is relatively low these days and does not represent an independent indication to carry out invasive diagnostics.

After the examination:

After the puncture process you will normally stay at the practice for another half an hour. We advise you to recuperate for two to three days after the procedure, whereby light activities can be pursued. During the next few days after the puncture, you should see your gynaecologist for a checkup.
If any symptoms occur (pain, bleeding, amniotic fluid discharge), get in touch directly with your gynaecologist or us.

Possible complications from the procedure:

Complications seldom occur but cannot be ruled out despite the examination being performed with the utmost care. Occasionally there may be a temporary discharge of amniotic fluid or bleeding. In most cases the pregnancy can be maintained by taking suitable action (recuperation, poss. in-patient monitoring).
A miscarriage or premature birth can be expected in approx. 1 out of 200 punctures. Risks for the mother, such as infections and bleeding, are extremely rare on the other hand.
As invasive measures are performed by our practice on a routine basis and in compliance with all the necessary quality assurance measures, complications are extremely unlikely. Thanks to constant sonographic guidance during the puncture process, direct damage to the foetus can be prevented.

Value of the diagnostics and possible problems:

By means of amniocentesis, many but not all chromosomal abnormalities can be ruled out with a very high degree of certainty.
In seldom cases, even though the procedure is performed carefully, there may be no result or an unclear one (e.g. because the cells do not reproduce normally or different chromosomal divisions are found). It may then be necessary to repeat the amniocentesis.
Occasionally one sees chromosomal material that cannot be clearly identified and whose significance for the development of the child remains unclear (so-called marker chromosomes). For further clarification, it may be necessary to examine other cells from the child (e.g. by puncturing the umbilical cord) or the chromosomes from the parents' blood.
Changes to very small chromosome segments or to single genes cannot be detected under the microscope. The array CGH diagnosis provides an approx. 100 times higher resolution compared to conventional methods and is thus able to detect minor abnormalities (microdeletion syndromes) at submicroscopic level. This technology is not paid for by the statutory health insurance schemes in routine cases, however. Additionally, there are specific gene panels (clinical exome sequencing, CES) which can also be a valuable tool in the setting of sonographic abnormalities.

In the event of abnormalities or unclear findings, we recommend a consultation from a specialist in human genetics, who will discuss these with you face to face. On request or in case of specific issues, a specialist genetic consultation may also be useful before an amniocentesis.

Limits of the examination:

The possibility that the expected child will reveal other physical or mental abnormalities, despite the diagnosis of a normal set of chromosomes, cannot be ruled out. Although developmental defects or organ malformations can be shown with high probability by differential organ diagnostics (organ ultrasound) in the 20th - 22nd week of pregnancy, an unremarkable examination can never fully guarantee a healthy child.