Dear expectant mother,

You have attended our practice today to undergo an amniotic fluid test (amniocentesis). Prior to the procedure, it is important that you have understood the possibilities, limitations and risks of this test. This patient information sheet should give you some basic information before the discussion with your physician.

Reasons (indications) for an amniocentesis:

The amniotic fluid test serves as an additional diagnostic tool during pregnancy, primarily to determine chromosomal disorders (disorders of the genetic material of the unborn child). This test is performed on pregnant women who exhibit an increased risk regarding these genetic diseases on the basis of their prehistory or findings during pregnancy, or if they wish to obtain further information for personal reasons.

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. This is because the risk of misdistribution of chromosomes (the carriers of the genetic material in the cells) during development of the ovum increases with the age of the mother. This incorrect cell division can cause the number of chromosomes to change, for example. The most commonly occurring chromosomal disorder is Down’s syndrome, in which chromosome 21 is present 3 times instead of twice (trisomy 21).

Other reasons for an amniocentesis can include abnormal findings in the ultrasound examination, abnormal blood tests (risk tests) or chromosomal mutations in the family or the parents. An amniotic fluid test can also be useful where infection of the unborn child (e.g. by the cytomegalovirus or in the event of toxoplasmosis) is suspected.

In this way, the amniotic fluid test can be used to safely exclude certain foetal diseases and alleviate the fear of a possible disease or malformation of the child.

Limitations of the test:

Analysis of the amniotic fluid includes testing of the chromosomes and determination of the alpha-fetoprotein (a foetal protein which passes into the amniotic fluid in greater concentration in the presence of fissure formations of the back or abdominal wall) as standard. If there are known congenital disorders in the family (in which usually only small sections of a chromosome – the genes - are mutated rather than an entire chromosome), in some cases it is also possible to screen for these (this is then called a molecular genetic test).

However it is never possible to exclude all conceivable diseases.
In rare cases, an unclear result or no result may be obtained despite careful performance of the test (e.g. because the cells to not multiply properly, or different chromosomal distributions are found). It may then be necessary to repeat the amniocentesis.

Alternatives:

Depending on the gestational age and issue at hand, other test methods may come into consideration for you (detailed ultrasound examination, risk assessment using specific blood tests). If you require more detailed information about this, please mention this in the pre-procedural discussion.

Outline of the test:

A detailed ultrasound examination is performed before every amniocentesis. This is followed by skin disinfection to prevent the introduction of bacteria or viruses.

Under ultrasound monitoring, a thin needle is then guided to the required region. This ensures that the target region is reached directly and rapidly. The visual monitoring also minimises the risk of accidental injury to the foetus or adjacent organs. An attached syringe is then used to extract the amniotic fluid sample.

The pain involved is perceived by women undergoing the procedure as somewhat unpleasant pressure in the lower abdomen and is said to be similar to a blood sample extraction or vaccination. For this reason, the administration of a painkiller or local anaesthetic is not necessary.

The foetal cells present in the amniotic fluid sample are then multiplied in a suitable laboratory. When enough cells have grown, the analysis can be performed. This takes an average of 14 days.

It is also possible to obtain the result for individual chromosomal disorders such as trisomy 21 (Down’s syndrome) within 24 hours using a rapid testing procedure (known as FISH diagnostic testing). These rapid tests are not usually included in the benefits catalogues of statutory health insurers and must be paid for by you if you want these. If you have specific questions about this please contact us.

Possible complications:

Complications rarely occur, but of course cannot be excluded in individual cases despite careful performance of the test.

A miscarriage occurs after around 0.3 – 0.5% of punctions.

In very rare cases, temporary amniotic fluid discharge or bleeding results; in most cases the pregnancy can be sustained using suitable measures (rest, possibly inpatient monitoring).

Injuries to adjacent organs (e.g. bladder, intestine or blood vessels) or infections are even rarer.

Injuries to the child are extremely rare.

Behaviour after the procedure:

On the day of the procedure and the following day, you should avoid significant exertion (please avoid sexual intercourse also).

A follow-up examination by the attending gynaecologist is advisable within one week after punction.

Please visit your attending gynaecologist, us or a gynaecological clinic for an examination if you notice:
- Bleeding
- Amniotic fluid discharge
- Persistent or increasing stomach pain.
Please consider the following:

The vast majority of tests reveal no abnormalities, which can help to relieve anxieties and contribute to an uneventful pregnancy.

However, parents-to-be can experience ethical and psychosocial conflicts arising from the performance and results of the amniocentesis. Particularly where serious foetal diseases are detected or in the event of a miscarriage resulting from an amniocentesis, these conflicts are to be expected. However we will inform you promptly and support you, in consultation with other physicians also (e.g. human geneticists and paediatricians, self-help groups etc.). 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 an amniotic fluid test 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 outlined here,
- 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 amniotic fluid test:
Yes: □  No: □

I wish to receive further genetic counselling before the examination:
Yes: □  No: □

I wish to receive further genetic counselling before the test results are available:
Yes: □  Only if findings are abnormal: □  No: □
**Consent:**

**Communication of examination results:**

The physician who performed the amniotic fluid test will inform you of the test 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 amniotic fluid test 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 and significance of the test, its risks and complications were discussed and answered for me comprehensibly. I feel well informed, have no further questions, and consent to the test and the extraction of the genetic sample. 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

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