Chorionic villus sampling - Punction of the placenta

Frauenheilkunde und
Pränatale Medizin

Dear expectant mother,

You have attended our practice today to undergo chorionic villus sampling. 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 chorionic villus sampling.

In Germany, all expectant mothers who are 35 or older at the time of birth are offered a prenatal diagnostic testing (known as “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 rises 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).

The chorion is a layer of cells on the exterior of the amniotic sac. The chorion cells give rise to chorion villi, which eventually form the foetal part of the placenta. Although these cells are not part of the foetus, they are usually genetically identical.

As chorionic villus sampling can be performed from the end of the 11th week of pregnancy, it is suitable for couples seeking the earliest possible test results.

Other indications for chorionic villus sampling can include abnormal findings in the ultrasound examination (e.g. increased nuchal translucency), abnormal blood tests (risk tests) or chromosomal mutations in the family or the parents. Chorionic villus sampling is also often offered in the case of metabolic disorders or known congenital disorders in the family.

Limitations of the test:

In chorionic villus sampling, a direct preparation is created from part of the tissue sample extracted. Using special techniques, the rough structure and the number of chromosomes can be determined within 24-48 hours. A normal result largely excludes the most common chromosomal disorders (Trisomies 21, 18 and 13, Turner’s syndrome).

A long-term culture is then created from the remaining tissue, the results of which are usually available after around 14 days. The examination of the cultured cells then forms the final result, in which even smaller anomalies (where microscopically identifiable) can be registered or excluded.

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.

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 chorionic sampling. Particularly where serious foetal diseases are detected or in the event of a miscarriage resulting from chorionic sampling, 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 test 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 or our human geneticists 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,

Consent:

Communication of examination results:

The physician who performed the chorionic sampling 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 chorionic sampling 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

The sample does not always contain enough tissue, so in rare cases direct preparation may be waived, and the results of the long-term culture must be awaited.

In rare cases, insufficient growth may be achieved in the long-term culture (referred to as culture failure). It may then be necessary to repeat the chorionic villi sampling, or (depending on the gestational age) perform an amniotic fluid test (amniocentesis).

In rare cases, chromosomal mosaics arise. This is where different chromosomal patterns are found in the cells of the direct preparation and/or the long-term culture. Additional procedures may be necessary for further clarification (amniotic fluid test, umbilical cord punction).

As no amniotic fluid is extracted in chorionic villus sampling to determine the alpha-fetoprotein (AFP) for detecting open fissure formations of the back or abdominal wall, an AFP determination should be performed in the 16^{th} – 18^{th} week of pregnancy using the blood of the mother and/or an ultrasound examination performed in 20^{th} – 22^{nd} week of pregnancy to exclude such defects.

Note that a normal set of chromosomes does not always mean that your child is healthy. Many foetal malformations and diseases (e.g. heart defects, limb malformations, metabolic disorders) cannot be excluded using this test. However, some of these diseases can be detected using a high-resolution ultrasound examination in the 20^{th} – 22^{nd} week of pregnancy.

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 chorionic villus sampling. 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 suck out some villi.
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.

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 0.5% of punctions.
In very rare cases, intermittent 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.