

Pränatale Diagnostik Berlin - Lichtenberg

Dres. J. Paulick, A. Sarut Lopez, K. Ramos

Surname:		Name:		Date of Birth:	
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Information on diagnostic punctures

A medical issue has arisen during your pregnancy that requires invasive diagnostics. This involves puncturing the amniotic cavity or placenta with a thin hollow needle through the abdominal wall in order to obtain cells from the fetus or placenta for genetic testing. The most common indications (medical reasons) for diagnostic punctures during pregnancy are:

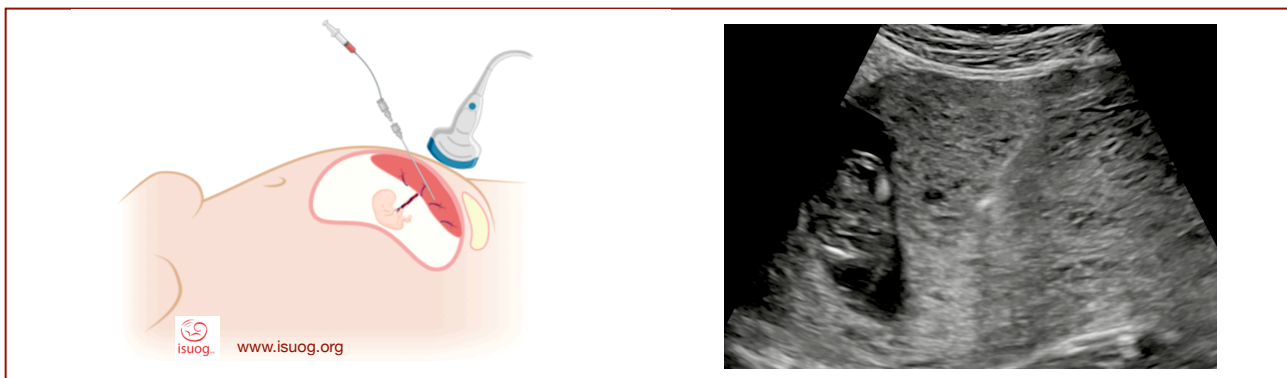
- Suspicion or exclusion of a genetic disorder in the expected child, for example if the pregnant woman or the father of the child is older, if there is a suspicion of malformation or in the case of abnormal risk tests, such as nuchal translucency measurement or NIPT (non-invasive prenatal test)
- Suspected infection of the fetus (toxoplasmosis, cytomegalovirus)

Various forms of material collection are available for invasive diagnostics. Which method is used in your case depends on the medical issue, the location of the placenta and the gestational age.

The procedure is similar for all forms of invasive diagnostics:

A detailed ultrasound examination takes place before each of these measures. The skin is then disinfected to prevent the introduction of bacteria and viruses. A thin needle is then inserted into the desired region under careful constant ultrasound guidance. This prevents unintentional injury of the fetus or neighboring organs. The desired material (amniotic fluid, chorionic villi) is obtained using a needle with a syringe attached. The women concerned often feel an unpleasant pressure in the lower abdomen, comparable to a blood sample or vaccination. Maternal complications following diagnostic punctures are extremely rare and are usually limited to some abdominal discomfort, minor hematomas in the abdominal wall or cardiovascular dysregulation.

Chorionic villus sampling (CVS: placenta puncturing)



Period: 11 - 14 weeks gestation

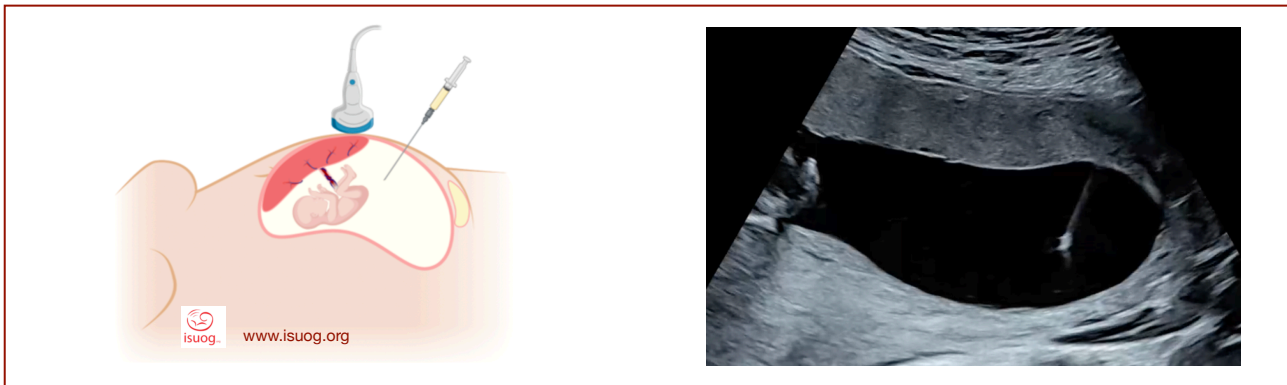
Advantage: Early result, early certainty and early decision in the event of illness.

The test is suitable for clarifying most genetic issues that are known in early pregnancy or even before pregnancy. A further advantage is that the inner pregnancy envelope, the so-called egg membranes (amnion), are not damaged, as the chorion lies outside this inner envelope. The short-term culture (preliminary findings) is usually available after 24-48 hours, the final findings after around 14 days.

Disadvantage: In approx. 2% of cases there are unclear findings (mosaics), which must be clarified with an amniocentesis.

Risk: 0.3-0.5% risk of miscarriage in our patients. As spontaneous miscarriages due to fetal developmental disorders can also occur at this time, it is difficult in individual cases to establish a causal connection with the chorionic villus sampling or to rule it out with certainty if a miscarriage has occurred.

Amniocentesis (AC: amniocentesis)



Period: from 15+0 weeks' gestation

Advantage:

The procedure is slightly less unpleasant for the pregnant woman.

The final result of the examination (karyogram) is available after approx. 14 days, results of the trio exome examination after approx. 3 weeks. A rapid test for fetal trisomies 21, 18 and 13 (FISH) can provide a result after 24-48 hours.

Risk: 0.1 - 0.2% risk of miscarriage in our patients. Clinically relevant unintentional injuries to the fetus during the puncture are extremely unlikely and have not occurred in our practice to date. In very rare cases, small amounts of amniotic fluid may be discharged temporarily (leakage), which stops on its own after a short time.

What are the limits?

A general examination for all diseases is not possible. Only those diseases whose genetic origin is known can be detected.

What can you do to reduce the risk of miscarriage?

We recommend to rest for the first 2 days after the procedure and refrain from exercising and lifting heavy loads (over 5 kilograms).

A check-up with your gynecologist within a week of the puncture is recommended. If you require a certificate of incapacity for work for 2 days, please contact us.

If you have the following complaints, please consult your gynecologist or a gynecological clinic: Bleeding, discharge of amniotic fluid, persistent or increasing abdominal pain, fever.

Please bear in mind:

Neither invasive diagnostics nor ultrasound examinations carried out professionally with high-end equipment are able to detect all diseases. For example, blindness, deafness and various forms of mental developmental disorders cannot be diagnosed prenatally.

Ethical and psychosocial conflicts can arise for expecting parents during the course and as a result of today's diagnostic puncture. These conflicts are particularly likely in cases of proven serious fetal diseases or indications of genetic diseases. According to §2 of the Pregnancy Conflict Act, women and men are entitled to free psychosocial counseling in these cases. Please contact us for addresses, which we will be happy to provide you with. In connection with a diagnostic puncture, we recommend that you take advantage of genetic counseling. This involves a detailed survey of your own and your family's medical history. Based on the latest scientific findings, the pedigree survey will determine whether there are familial or other risks of diseases for you, your offspring or the embryo and, if necessary, discuss treatment options.

If, in the course of today's consultation or in the course of the examination, it becomes clear that you do not wish to undergo this examination, you have the opportunity to indicate this at any time and exercise your right not to know. You can withdraw your consent to the examination at any time.

Patienten - ID:

Please answer the following questions:

I have already received adequate advice in preparation for today's procedure at a specialist practice for medical genetics:

Yes No

I would like to have another genetic consultation in a specialist practice for medical genetics after the test results are available:

Yes No Only in case of abnormal findings

All data is recorded and stored electronically in our practice. The statutory retention period for the findings collected in this context is 10 years. According to §12 of the German Genetic Diagnostics Act, the documents must then be destroyed. However, in the context of quality assurance and scientific work, we also require some data beyond this period. These are used in pseudonymized form and in compliance with the statutory data protection regulations. It may also be useful for you and your descendants to have the data collected available even after 10 years and beyond.

I agree that the data collected may be stored for longer than 10 years:

Yes No

I consent to the transmission of the results of the diagnostic puncture as follows:

Yes No

Gynecologist:

Other doctors /persons:

I agree that the results of the genetic analysis may also be transmitted directly to me by the staff of the Medicovert Practice for Human Genetics Berlin Lichtenberg - Drs. Kinzel, Zschieschang, Hampel, Cronauer, Engfer, Hebebrand, Khuller:

Yes No

I would like a copy of the genetic findings:

Yes No

I will make any remaining test material available to the analysis laboratory in anonymized form for comparative tests.

Yes No

Questions / comments on the personal interview:

I have read this information, understood the explanation and was able to ask my remaining questions during the preliminary discussion. All the information was presented in a form that I could understand and my questions about the purpose, type, scope, significance and the results that could be achieved with this requested examination were answered in detail and clearly.

I agree to the examination and any further examinations that are necessary for the preparation of the requested findings and do not wish to have any further time for reflection.

Yes No

I have been informed about the right not to know, the right to seek conflict advice and my right to withdraw my consent at any time.

Yes No

Date: Signature of patient

Date: Signature of doctor