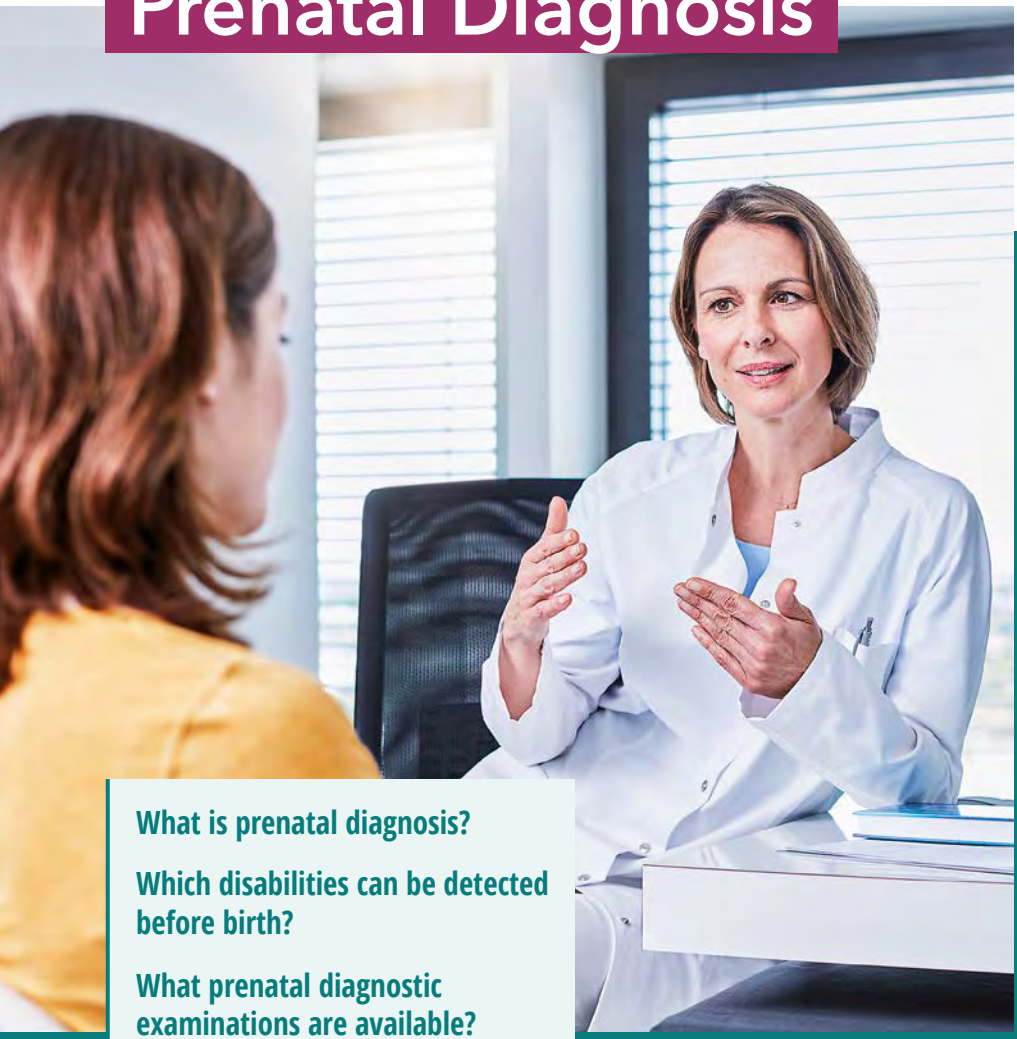
ADVICE, METHODS AND ASSISTANCE

Prenatal Diagnosis



What is prenatal diagnosis?

Which disabilities can be detected before birth?

What prenatal diagnostic examinations are available?

Federal Institute of Public Health

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Content

What is prenatal diagnosis? **2**

Non-invasive and invasive procedures 3
Important considerations 4
Medical information and advice 5
What is the next step after an abnormal result? 5

Which disabilities can be detected before birth? **8**

Overview of disabilities that can be detected before birth 9
Heart defects 9
Neural tube defects 10
Chromosomal abnormalities 10

What prenatal diagnostic examinations are available? **12**

Overview: Prenatal diagnostic examinations 13
Ultrasound 14
First trimester screening (FTS) 16
Blood tests for genetic abnormalities:
Non-invasive prenatal tests (NIPT) 18
Puncture of the placenta (chorionic villus sampling) 20
Amniotic fluid examination (amniocentesis) 22
Puncture of the umbilical cord (chordocentesis) 24

Advice centres **25**

Foreword

Dear Reader,

Even though there is a lot of talk about risks associated with pregnancies, the vast majority of children are born healthy.

> Few diseases or disabilities are congenital and only some of them can be detected before birth.

In order to detect health risks for the mother and child at an early stage, midwives and doctors monitor the course of the pregnancy and the development of the child very carefully. These examinations – prenatal care – are paid for by the health insurance provider. In the case of abnormalities, there are additional prenatal examinations and tests that search specifically for malformations and disorders in the unborn child. Some of these examinations are only carried out by specialised gynaecologists who specifically deal with prenatal diagnosis.

Some parents want these additional prenatal examinations although there are no particular risks or abnormalities. In such cases, they have to pay for these tests themselves. The statutory health insurance providers therefore only cover the costs if they are medically substantiated.

All examinations are voluntary. Irrespective of whether or not there are specific abnormalities and risks, you may decide for yourself whether you want to have prenatal diagnostic examinations and if so which ones.

The brochure provides an overview of prenatal diagnostic examinations as well as advisory and support services and can help you make your personal decision.

This should help expectant parents to consciously decide whether or not to have prenatal diagnostic examinations.

What is prenatal diagnosis?

“Prenatal diagnosis” means examinations that search specifically for indications of genetic abnormalities, malformations or developmental disorders in the unborn child. In any case, however, the following applies: Not every disease or disability is the same. The results of prenatal examinations therefore usually say little about how severe a disease or disability will be in an individual case and what limitations they will precisely entail.

Non-invasive and invasive procedures

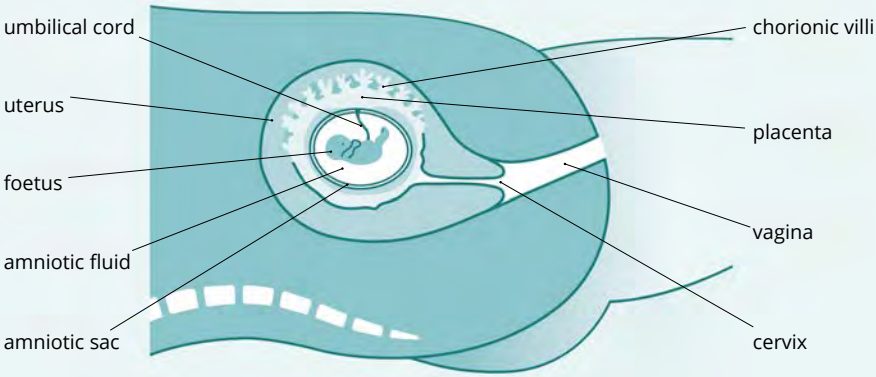
There are non-invasive and invasive procedures:

- **Non-invasive procedures** do not interfere with the woman's body or require only a blood sample from the mother. They therefore pose no risk to the woman or the unborn baby. These include ultrasound examinations, first trimester screening (FTS) and genetic blood tests (NIPT).
- **Invasive procedures** are associated with interference in the woman's body. A hollow needle is used to pierce the placenta, the foetal cavity or the umbilical cord to remove and examine cells. This carries a low risk of miscarriage. These include puncture of the placenta (chorionic villus sampling) and examination of the amniotic fluid (amniocentesis). Puncture of the umbilical cord (chordocentesis) is rarely used.

Some non-invasive procedures provide only probabilities so their significance is limited. In order to specifically confirm the suspicion of an abnormality, invasive procedures will be considered as a next step. Many women and couples fear that such an intervention will endanger their unborn child. But the tests are now much safer than before. For example, the risk of miscarriage from an amniocentesis test or chorionic villus sampling is now 0.5 to 1 %.

GRAPHIC 1

Organs and structures in the 12th to 16th week of pregnancy



Important considerations

Expectant parents hope that prenatal diagnostic examinations will give them the reassuring certainty that their child is healthy. However, some of the tests can only estimate how likely it is that an abnormality will occur. You may be concerned by false-positive results ("false alarms"). The examinations may also overlook diseases or disabilities (false-negative results).

In any case, the following applies: Prenatal diagnosis cannot guarantee a healthy child.

If doctors find an abnormality, further examinations can sometimes provide more precise insights. You decide for yourself whether you are ready for these examinations and the associated stresses and risks. Doctors therefore provide detailed advice on all aspects of these examinations. Pregnancy counselling centres also assist you with these considerations so that you can make an informed decision.

Important questions that expectant parents need to consider before opting for an examination include the following:

- What will I find out about my child during pregnancy?
- What reasons are there for carrying out the examination and what may be the reasons against it?
- How long does it take until the result is obtained?
- How reliable is the result of the examination?
- What do I do if the result is abnormal?
- Would I then have further examinations to be able to better assess the situation even if this means a risk for the mother and child?
- What happens if the abnormality is confirmed?
- How do we handle an abnormality as a couple or family?

Other considerations to be taken into account: Expectant parents can find the constant emphasis on medical risks stressful, which can damage the relationship that they are building with their child – particularly in the first trimester of the pregnancy. However, a normal result may reassure pregnant women, for example, if they have already lost a child, if there is a hereditary disease in the family or if they are afraid of a developmental disorder.

Medical information and advice

Prenatal diagnosis is always voluntary. You can decide for yourself which examinations and tests you want to have and which you do not want to have. You have a right to know, but also a right not to know. It is therefore advisable to get good information in advance about what services are available and the significance of the respective examination results.

Your doctor is legally obliged to give you information before each prenatal diagnostic examination. The conversation includes information on

- The type of the test and its risks
- The possible results
- The resulting consequences.

The doctor must also inform you that you are entitled to have psychosocial counselling in a recognised pregnancy counselling centre. You can get advice before making a decision, while waiting on a result and after you have obtained the result – this is free of charge and can also be anonymous if you wish.

What is the next step after an abnormal result?

If abnormalities are found in your unborn child, doctors discuss the result with you in detail. It may be that the doctors recommend further examinations to determine a reliable diagnosis. This can also involve invasive procedures.

Doctors discuss with you the option of treating your child, even during the pregnancy. But for many disabilities or diseases detected before birth, there are no therapeutic approaches of this kind. But in some cases, early knowledge of a disease may improve the child's start in life.

For example, if the child has a heart defect, it is possible to specifically plan for their delivery in a perinatal centre with a specialist children's hospital.

If there are no postnatal treatment options, you will receive individual advice and support – from doctors, but also from counsellors at a pregnancy counselling centre if you wish. They can help with the following considerations: How do I imagine life with a disabled child? Are we ready to get to know our child with all their characteristics and needs or do we want to terminate the pregnancy? Parent associations organised by parents with disabled children may also be a helpful point of contact. An insight into life with a disabled child can provide an additional perspective.

The counsellors at the pregnancy counselling centre will show you possibilities for assistance and support for life with a disabled child. They will help you collect information and give you other important contacts. If you decide not to continue with the pregnancy, they will discuss the next steps with you. Sometimes it is also evident that your child will only have a very short life expectancy. The counsellors will help you to organise or provide the best possible care during this time ("palliative support").

Advice – information - support

Prenatal diagnostic examination results may have far-reaching consequences. Expectant parents need a place where they can speak about their issues, fears and expectations. We know from experience that in addition to medical facts, feelings, life experiences and intuition play a major role when handling prenatal diagnostic results. Your gynaecologist or prenatal diagnostician will give you advice during this difficult time. The pregnancy counselling centre is also an important place where you will receive psychosocial assistance and support to find answers to your questions.

If you are considering an abortion, the pregnancy counselling centres will help you before and after this difficult decision. The advice is confidential and free of charge. The specialist in the counselling centre will continue to support you, regardless of the decisions you make. If you are expecting a child with disabilities, the counselling centre can, for example, arrange contact with parents in the same situation or refer you to the relevant associations and organisations.

A counselling centre can also provide competent information on financial assistance and support to which you are entitled. You will also find support there if you need to say goodbye to your child because they will live only for a short time.

Further information on how to deal with an abnormal result and contact addresses for associations can be found in the "Special Circumstances" brochure published by the Federal Institute of Public Health.

> You will find a counselling centre near you at www.familienplanung.de/beratungsstellensuche.

Abortion

Sometimes there are several reasons for considering an abortion. It is possible to have an abortion without penalty up to twelve weeks after conception. This corresponds to the 14th week of pregnancy if the calculation is not made from the day of conception, but the first day of the last period. After this time, an abortion may only be carried out if a medical indication is available. This means: According to a medical assessment, the continuation of the pregnancy will seriously endanger the physical or mental health of the pregnant woman and this risk cannot be averted in any other reasonable way. Only then is an abortion legally permissible. The expectant mother can therefore not decide on the late termination of the pregnancy alone. And in the event of an abnormal result, medical consent to an abortion is not automatically given. Before and after the difficult decision to terminate a pregnancy, you have a legal right to talk to a pregnancy counselling centre to clarify your feelings and thoughts and to make a decision.

The Pregnancy Conflict Act also provides for a three-day reflection period after notification of the diagnosis before the doctor issues the written medical indication.

If an abortion is carried out after the end of the 14th week of pregnancy, the woman usually receives labour-inducing medication to initiate the birth. A few hours to a few days may pass before the intended miscarriage occurs. The initiated birth takes place in a delivery room or labour ward. The woman is cared for by a midwife and a doctor.

The medical indication for an abortion is carefully weighed up by doctors after considering the child's ability to survive outside the womb. It is then necessary to induce cardiac arrest in the child before initiating the birth (foeticide).

Which disabilities can be detected before birth?

Diseases in newborn babies that affect their long-term health can have many causes, such as infections, environmental influences or underdevelopment in a premature birth. Only a small proportion of these are hereditary or occur randomly when the egg and sperm cells join.

Overview of disabilities that can be detected before birth

Chromosomal[1] abnormalities are hereditary or accidental disabilities that occur when the egg and sperm cells join. They may be detected by the first trimester test, the genetic blood test (NIPT), chorionic villus sampling or amniocentesis. Heart defects and other organ malformations are usually detected by ultrasound. If an unborn baby has a serious disability, it commonly leads to a miscarriage, often in the first trimester of the pregnancy.

The most common malformations in the heart, kidneys, central nervous system and skeletal system are found in unborn babies. In about 10 % of the abnormal results, the causes are related to the chromosomes. Below you will find an overview of some congenital disabilities (heart defects, malformations and chromosomal abnormalities, single gene disorders). The frequency data is based on extrapolations and estimates[2, 3]. It estimates approximately how many women in pregnancy, after childbirth or after a miscarriage learn that their child has or has had a disability.

Heart defects

Frequency: In about 0.8 % of pregnancies, the unborn baby has a heart defect.

Heart defects usually only appear after birth when the child's circulation changes and the newborn baby breathes independently. Many congenital heart defects are well treatable and the children can often lead a largely normal life. Childhood cardiac arrhythmias can sometimes be treated during pregnancy by the woman taking medication.

1 Chromosomes contain the entire genetic information (genes). Each human cell contains a set of chromosomes. In most people, this consists of 22 pairs of chromosomes and two sex chromosomes (46 XX or 46 XY). If the set of chromosomes is changed, medicine talks about a chromosomal disorder or deviation.

2 Source: EUROCAT, a European network for the detection of childhood anomalies that occur during pregnancy or after birth (https://eu-rd-platform.jrc.ec.europa.eu/eurocat_en).

3 There is no EUROCAT data available for Klinefelter syndrome. The frequency data is based on international studies.

Neural tube defects

Frequency: These malformations occur in about 0.1 % of pregnancies. Spina bifida represents about half of these cases.

Neural tube defects can be very different. For example, in the case of spina bifida: Some people have fewer problems and are still able to walk, but some are more affected and require a wheelchair for their entire life. If such malformations are detected early, this enables careful birth planning and competent medical care for the newborn baby in a special centre. In some cases, the child's head is greatly enlarged when the drainage pathways of the cerebrospinal fluid are affected (hydrocephalus or water on the brain). The most serious of these malformations is anencephaly. In this case, parts of the skull bone and brain have not developed and the children die shortly after birth. Neural tube defects are usually detected by ultrasound.

Chromosomal abnormalities

Trisomy 21 (Down syndrome)

Frequency: In about 0.2 % of pregnancies, the unborn baby has trisomy 21. The probability of the child having trisomy 21 increases with the woman's age: At the end of the pregnancy, this affects 8 in 10,000 in a 25-year-old, 29 in 10,000 in a 35-year-old and 100 in 10,000 in a 40-year-old.

Children with Down syndrome usually develop more slowly, initially physically and later mentally. A trisomy is associated with physical abnormalities; often the child has heart defects or malformations in the digestive tract. Physical limitations can usually be estimated fairly accurately. How the child will develop mentally, on the other hand, cannot be predicted during the pregnancy. Some people with Down syndrome need a lot of support later in their life while others can pursue a profession and lead an independent life.

Trisomy 18 (Edward's syndrome) and trisomy 13 (Patau syndrome)

Frequency: In about 0.1 % of pregnancies, the unborn baby has trisomy 18 or trisomy 13.

Children with these chromosomal abnormalities often have malformations of the head and brain, heart, limbs and/or other organs. Most children die during the pregnancy or in the first weeks after birth, but children who are born alive rarely reach five years of age or older.

Other chromosomal abnormalities

Frequency: Turner syndrome occurs in about 0.03 % of all pregnancies. The frequency of Klinefelter syndrome can be estimated at approximately 0.1 to 0.25 %.

Abnormalities in sex chromosomes such as Turner syndrome (monosomy X) in girls or Klinefelter syndrome in boys may lead to physical abnormalities such as dwarfism or infertility. In very many cases, only slight abnormalities exist, meaning that this genetic disorder is only discovered at a later stage or not at all. If therapy is necessary, those affected often benefit from the earliest possible treatment. Then it is advantageous to diagnose the syndrome at an early stage.

Single gene diseases

In addition, there are diseases that are abnormalities in individual chromosomes (single-gene diseases). Such abnormalities lead to different disease patterns such as

- Blood diseases, e.g. haemophilia
- Muscle diseases, e.g. Duchenne muscular dystrophy
- Metabolic diseases, e.g. cystic fibrosis

Such gene changes occur regardless of the age of the pregnant woman and occur in about 1 % of pregnancies, in some cases even less frequently.

If an unborn baby has a serious chromosomal disorder, a miscarriage often occurs automatically in the first trimester of the pregnancy.

What prenatal diagnostic examinations are available?

Along with the regular prenatal care, prenatal diagnostic examinations aim to clarify any abnormalities or problems in the child. Depending on the week of pregnancy and the problem, different methods may be used – which in turn provide results with varying degrees of reliability. A consultation helps in making the decision for or against a particular test. Contact points include gynaecology practices, centres and practices specialising in prenatal diagnosis and pregnancy counselling centres. Below you will find an overview of the individual examinations.

Overview
Prenatal diagnostic examinations

All examinations are voluntarily. You decide for yourself which examinations you want to have and which you do not want to have (irrespective of whether there are certain risks or abnormalities).

From the 10th week of pregnancy

Blood tests for genetic abnormalities: Prenatal tests (NIPT)

Important information on page 18

From the 12th week of pregnancy

Puncture of the placenta (Chorionic villus sampling)

Important information on page 20

11th to 14th week of pregnancy

First trimester screening (FTS)

Important information on page 16

From the 16th week of pregnancy

Amniotic fluid examination (Amniocentesis)

Important information on page 22

From the 18th week of pregnancy

Puncture of the umbilical cord (Chordocentesis)

Important information on page 24

20th week of pregnancy

Doppler ultrasound

Important information on page 14

20th to 22nd week of pregnancy

Precise diagnostics or an organ ultrasound

Important information on page 14

invasive

non-invasive

Ultrasound

Ultrasound examinations are part of the regular examinations in prenatal care and are carried out around the 10th, 20th and 30th week of pregnancy. At the ultrasound examination around the 20th week of pregnancy, the pregnant woman may also opt for the extended ultrasound examination, which examines the organs more closely. This examination is usually carried out by a gynaecologist. In the event of specific risks or a suspected developmental disorder, the precise diagnostic ultrasound examination ("organ ultrasound") is usually carried out by a prenatal diagnostician (see right column).

Doppler ultrasound

From the 20th week, a Doppler ultrasound examines the blood flow in the blood vessels of the uterus and in certain blood vessels of the unborn baby. Such an examination therefore checks whether the embryo is getting an adequate supply of blood. The reason for a Doppler ultrasound may be that the foetus is comparatively small.

Precise diagnostics or an organ ultrasound

This examination is also called a precise diagnostic ultrasound or second trimester screening. It is carried out with high-resolution ultrasound equipment in special centres or practices. This examination is offered in the 20th to 22nd week of pregnancy.

Most of the serious malformations may be detected at this time. However, about half of all serious malformations can be diagnosed as early as the 13th week of pregnancy as part of the first trimester screening, which is not part of the standard care (see p. 16).

3D / 4D ultrasound

This examination produces a spatial image of the unborn baby, but rarely provides medically significant information.

How reliable is the result?

The quality of any ultrasound result depends primarily on the experience of the doctor and the quality of the ultrasound equipment. In addition, if there is minimal amniotic fluid or the child is in an unfavourable position or the pregnant woman has scarring or a thick abdominal wall, this may complicate the assessment. Even under favourable conditions, the ultrasound does not detect every malformation and false assessments may occur.

What are the risks?

In principle, the ultrasound is considered harmless to the unborn baby. The Doppler ultrasound, on the other hand, generates heat in the tissue that is examined so it should only be used for justified reasons and for as short as possible in early pregnancy. Modern ultrasound equipment has built-in control systems for this purpose.

Costs

The basic ultrasound and the extended ultrasound are paid for by the health insurance provider because they are part of the standard examinations. If further ultrasound examinations are necessary to clarify any abnormalities, the health insurance provider will also cover this. You must pay for any additional ultrasound examinations that you request yourself.

Advantages

- The result may already be discussed during the examination.
- A normal result may provide reassurance.
- Seeing an image of the unborn baby can create a feeling of closeness to the child.

Disadvantages

- Any abnormalities detected, even minor ones, can cause concern and often lead to further examinations.

First trimester screening (FTS)

First trimester screening (FTS) is carried out between the 11th and 14th week of pregnancy. In addition, the organs are assessed and the risk of pregnancy-induced hypertension disease (pre-eclampsia) is assessed.

The age of the pregnant woman and the length of pregnancy also play a role in assessing the risk of a chromosomal disorder.

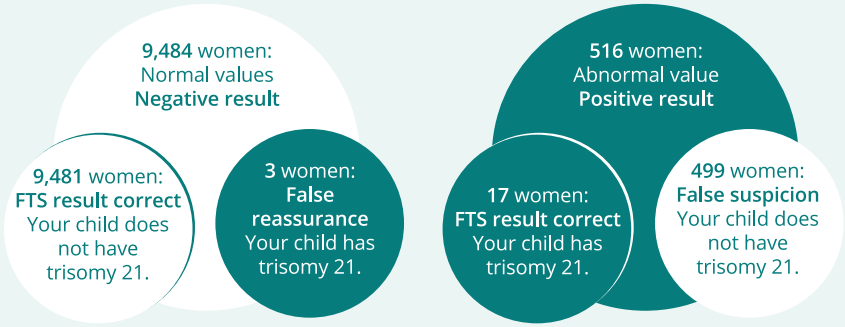
FTS includes the following:

- A blood test for two hormone and protein levels
- An ultrasound, in which the foetal nuchal fold (= nuchal translucency, NT) and possibly other characteristics of the unborn baby are measured.
- A measurement of the resistance in the uterine arteries
- A measurement of the mother's blood pressure

GRAPHIC 2

Diagram of the first trimester screening (FTS)

10,000 woman from all ages have the FTS.



The Diagram is based on the assumption that trisomy 21 occurs in 1 out of 500 of the unborn babies examined. First trimester screening detects trisomy 21 in 85 % of the unborn babies. It does not detect 15 % of the cases. In 5 % of the unborn babies examined, the test is positive, although the unborn babies are healthy (=false-positive)

Source: Guidelines of the Genetic Diagnostics Commission at the Robert Koch Institute [Richtlinie der Gendiagnostik-Kommission (GEKO) für die Anforderungen an die Durchführung der vorgeburtlichen Risikoabklärung sowie an die insoweit erforderlichen Maßnahmen zur Qualitätssicherung gemäß 23 Abs. 2 Nr. 5 GenDG. Bundesgesundheitsbl. 56, 1023–1027 (2013)].

What do I find out?

The results do not result in a diagnosis. The screening only calculates the probability of the child having trisomy 13, 18 or 21. The ultrasound may also reveal malformations in the child, for example, malformations of the arms and legs or of internal organs such as the heart or the brain.

If the screening detects an abnormality, further examinations bring more clarity, such as a further ultrasound examination or invasive diagnosis.

How reliable is the result?

First trimester screening often provides false abnormal results: Assuming 10,000 women attend the screening, then 516 receive an abnormal result. In 499 women, further examinations determine that the child does not have trisomy 21. In 3 out of 10,000 pregnancies, the test does not detect trisomy 21.

The adjacent numerical example in graphic 2 (see p. 16) explains the reliability of the results of first trimester screening.

Some parents choose only to have a nuchal fold ultrasound. In this case, the results are less accurate than with the complete screening.

Costs

Expectant parents have to pay for first trimester screening themselves. It costs between 150 and 250 euros.

Advantages

- Parents learn how high the risk is that their child has certain diseases, chromosomal abnormalities and severe malformations.
- By determining the risk of pre-eclampsia, changes may be made to the care of pregnant women.
- A normal result is reassuring.

Disadvantages

- The probability information is difficult to understand and evaluate.
- The test often leads to further examinations in the case of abnormalities or unclear results.
- 3 to 5 % of the women who take the test become anxious after a false abnormal result.

Blood tests for genetic abnormalities: Non-invasive prenatal tests (NIPT)

From a blood sample taken from the pregnant woman, components of the genetic material are filtered out that may be assigned to the pregnancy. These DNA fragments are then examined.

In this way, an estimate can be given as to the probability of certain chromosomal abnormalities in the unborn baby. These "non-invasive prenatal tests" (NIPT) are available from the 10th week of pregnancy. In a joint discussion with your gynaecologist, you will explore whether you have an increased risk so that the test may be implemented.

What do I find out?

The test determines the risk of trisomy 13, 18 and 21. The parents may also arrange additional examinations, such as to determine abnormalities in the sex chromosomes X and Y. The result is usually available after a few working days.

Further examinations, for example to check for single-gene diseases such as cystic fibrosis, are part of the research. Many medical societies do not recommend these types of genetic tests at the moment. The same applies to examinations for microdeletions. These are gene mutations in which parts of chromosomes are missing.

How reliable is the result?

The reliability of the test result is not always the same. In addition, it should be considered that this test is also a risk calculation. Whether or not a chromosome disorder is actually present depends on many factors.

This can be clearly demonstrated by the test for trisomy 21. In this case, there are three possible test results:

- **"The test is normal":** This result is usually very reliable. No further examinations are required.
- **"The test is abnormal":** The reliability of the result depends, among other things, on the age of the woman: In 40-year-old women, an abnormal test result for trisomy 21 is frequently confirmed (in 93 out of 100 cases). In 20-year-old women, on the other hand, the suspected abnormality is false in every second case (in 48 out of 100 cases).

- **"The test cannot be evaluated":** If there is too little pregnancy-specific genetic material in the blood sample, no result may be obtained. This is the case with up to 3 to 5 out of 100 NIPTs. In these cases, the NIPT may be repeated about 2 weeks later. Two thirds of the tests may then be evaluated. If an analysis is still not possible after the second evaluation, this indicates an increasing risk of chromosomal abnormality so a diagnostic puncture procedure may be discussed.

NIPTs for sex chromosomal abnormalities are much less reliable than NIPTs for trisomies. An example of this is the examination for Turner syndrome: Only 25 % of the positive results are really positive, so only a quarter of the abnormal results actually show that the child has Turner syndrome. There is not enough data available for the tests for single gene diseases to assess their reliability.

Costs

The trisomy blood test costs upwards of 170 euros, depending on the scope and the provider. Additional costs are incurred for additional analyses. The health insurance provider bears the costs of the test if it is medically substantiated.

Advantages

- The examination is not harmful for both the pregnant woman and the child.
- Normal test results may eliminate the need for invasive examinations.

Disadvantages

- Organ malformations that are not caused by chromosomal abnormalities are not detected.
- If the test determines an "abnormal" result, only an invasive examination (usually an amniocentesis test) can provide clarity.
- There are more frequent false abnormal results in younger women, as they are not in the risk group, as well as in very rare genetic syndromes or diseases.

Puncture of the placenta (chorionic villus sampling)

Tissue is taken from the placenta during this examination. The collected cells are suitable for this purpose because they are generally genetically similar to the cells of the unborn child. The doctor obtains the tissue sample by inserting a fine hollow needle through the woman's abdominal wall. Chorionic villus sampling is available from the 12th week of pregnancy. A laboratory then isolates the child's cells and examines the chromosome set.

After the examination, women may feel a slight pulling in the abdomen for a few hours to a few days. You should take care of your body for the first few days after the procedure and go on sick leave for a few days if required.

What do I find out?

The number and the rough structure of the chromosomes are initially examined. Conclusions on chromosomal abnormalities may be drawn from this, such as whether trisomy 13, 18 or 21 is present. In addition, sometimes a genetic analysis is carried out for certain hereditary diseases, for example, muscular or metabolic diseases. This is the case, for example, if the disease occurs in the family of the parents or if there are indications from previous examinations. In chorionic villus sampling, a preliminary result is available after one to two days and the final result after about two weeks.

How reliable is the result?

The result of the gene analysis is very reliable: The result is correct in 99 % of the examinations. It is rare that the examined cells do not all have the same chromosome pattern; in this case, the result is unclear. Sometimes no evaluation is possible and it is necessary to decide whether the examination should be repeated.

What are the risks?

The puncture procedure sometimes induces labour or a discharge of amniotic fluid and miscarriage occurs in about 0.5 to 1 % of all interventions.

Costs

The costs are covered by the health insurance provider if the examination is medically substantiated. This is the case, for example, if the child is suspected of having a disease due to the medical history or an abnormal result.

Advantages

- The result is very reliable and accurate in 99 % of cases. The examination is therefore suitable for confirming questionable results from previous examinations.
- Chorionic villus sampling is available from the 12th week of pregnancy and a preliminary result is available after one to two days.

Disadvantages

- It carries a (low) risk of miscarriage.
- The waiting time for the final result is 14 days.
- The waiting time may put a strain on building a relationship with the child.

Amniotic fluid examination (amniocentesis)

During this procedure, amniotic fluid is taken and the cells of the child contained therein are examined for genetic abnormalities. The doctor inserts a thin hollow needle through the abdominal wall into the amniotic sac of the woman. The child's cells are then multiplied and examined in a laboratory. Examination of the amniotic fluid is available from the 16th week of pregnancy. After the examination, women may feel a slight pulling in the abdomen for a few hours to a few days. You should take care of your body for the first few days after the procedure and go on sick leave for a few days if required.

What do I find out?

The number and the rough structure of the chromosomes are initially examined. Conclusions on chromosomal abnormalities may be drawn from this, such as whether trisomy 13, 18 or 21 is present. Sometimes a genetic analysis is carried out for certain hereditary diseases, for example, muscular or metabolic diseases. This is the case, for example, if the disease occurs in the family of the parents or if there are indications from previous examinations.

If desired, a partial result (FISH test or PCR test) for chromosomes 13, 18 and 21 as well as for the sex chromosomes X and Y is generated during the amniotic fluid examination after one to two days. However, this is not as certain as the final result. It takes around two weeks to get the complete result.

How reliable is the result?

The result of the gene analysis is very reliable as with chorionic villus sampling: The result is correct in 99 % of the examinations. It is rare that the examined cells do not all have the same chromosome pattern; in this case, the result is unclear. However, this is even rarer in amniocentesis than in chorionic villus sampling. Sometimes no evaluation is possible and it is necessary to decide whether the examination should be repeated.

What are the risks?

The puncture procedure may induce labour or a discharge of amniotic fluid and miscarriage occurs in about 0.5 to 1 % of all interventions. In the amniocentesis procedure, an injury to the unborn baby is possible, but it is extremely rare.

Costs

The costs are covered by the health insurance provider if the examination is medically substantiated, for example, if abnormalities have arisen during a preliminary examination. The costs for the FISH test are not covered by the health insurance provider.

Advantages

- The examination is therefore suitable for confirming questionable results from previous examinations.
- Examination of the amniotic fluid is available from the 16th week of pregnancy and a preliminary result is available after one to two days.

Disadvantages

- It carries a (low) risk of miscarriage and injury to the unborn baby.
- The waiting time for the final result is around 14 days.
- Building a relationship with the child can be stressful due to the waiting time.

Puncture of the umbilical cord (chordocentesis)

In the chordocentesis procedure, foetal blood is taken from the umbilical cord vein. The procedure is carried out in the case of certain problems, for example, if the child is suspected of being anaemic. To remove a sample, a fine hollow needle is pushed through the abdominal wall and amniotic sac into the umbilical cord vein using ultrasound. The doctor may also use access to the umbilical vein to treat the unborn baby. For example, the child may receive a blood transfusion or medication in this way. The chordocentesis procedure is carried out only in special centres. It is available from the 18th week of pregnancy.

What do I find out?

A chordocentesis procedure is mainly performed in the case of special medical problems such as the diagnosis of anaemia in the child. Otherwise, doctors usually recommend chorionic villus sampling or an amniocentesis test.

How reliable is the result?

The results are very reliable as the child's blood is examined directly.

What are the risks?

The puncture procedure may induce labour or bleeding and in the worst case a miscarriage may occur. Complications occur in 1 to 3 % of the interventions.

Costs

The costs are covered by the health insurance provider if the examination is medically substantiated.

Advantages

- The result is very reliable as the child's blood is examined directly.
- In some cases, direct access to the umbilical vein allows for the child to receive therapy. An example would be a blood transfusion in the case of anaemia in the child.

Disadvantages

- There is an increased risk of miscarriage.
- The examination is only available in special centres.

Advice centres

You can find information on advisory services in the region at familienplanung.de or from the following institutions:

AWO Arbeiterwohlfahrt Bundesverband e. V.
Phone +49 (0)30/2 63 09-0
E-mail: info@awo.org
www.awo-schwanger.de
www.awo.org

Deutscher Caritasverband e. V.
Phone +49 (0)7 61/2 00-0
E-mail: info@caritas.de
www.caritas.de

Der Paritätische Gesamtverband e. V.
Phone +49 (0)30/2 46 36-0
E-mail: info@paritaet.org
www.der-paritaetische.de

Deutsches Rotes Kreuz e. V.
Phone +49 (0)30/8 54 04-0
E-mail: drk@drk.de
www.drk.de

Diakonie Deutschland
Phone +49 (0)30/6 52 11-0
E-mail: diakonie@diakonie.de
www.diakonie.de

donum vitae e. V.
Phone +49 (0)2 28/36 94 88-0
E-mail: info@donumvitae.org
www.donumvitae.org

Evangelische Konferenz für Familien- und Lebensberatung e. V.
Phone +49 (0)30/51 57 53 -73
E-mail: info@ekful.de
www.ekful.de

pro familia Bundesverband e. V.
Phone +49 (0)69/26 95 77-90
E-mail: info@profamilia.de
www.profamilia.de

Sozialdienst katholischer Frauen – Gesamtverein e. V.
Phone +49 (0)2 31/55 70 26-0
E-mail: info@skf-zentrale.de
www.skf-zentrale.de

You will find a counselling centre near you at www.familienplanung.de/beratungsstellensuche.

ADVICE, METHODS AND ASSISTANCE

Prenatal Diagnosis

Imprint

Publisher
Federal Institute of Public Health
Maarweg 149–161, 50825 Cologne

www.bioeg.de
www.familienplanung.de
Instagram: @bioeg

Editorial
Annika Hesse, Anastasia Doré

Expert advice
Prof. Dr. Karl Oliver Kagan

Concept and Design
Kühn Medienkonzept & Design GmbH,
Ruppichteroth, Köln

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Edition
Last updated: April 2025

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Order address
by mail: Federal Institute of Public Health,
50819 Cologne
by fax: +49 (0)2 21/8992-257
by e-mail: bestellung@bioeg.de
Item number: 13625101

Federal Institute of Public Health

In cooperation with
Bundesarbeitsgemeinschaft der Freien Wohlfahrtspflege