PRENATAL EXAMINATIONS AT A GLANCE

ULTRASOUND

With an ultrasound, the unborn child can be imaged on a monitor. For this, the doctor guides a transducer over the belly of the pregnant woman, and a computer produces a picture from the reverberated sound waves. In the early pregnancy, the ultrasound can also be performed through the vagina.

Pregnancy care provides for three basic ultrasound examinations to examine the 10th, 20th and 30th week of pregnancy after the last menstruation (p.m.).* In the case of abnormalities, further special examinations such as Doppler ultrasound or a detailed diagnosis are recommended. A "neck fold ultrasound" is part of the first trimester test (see first trimester test).

WHAT CAN I LEARN?

Basic ultrasound

- 9th-12th Week: The gestational age is determined and whether more than one child is to be expected. The external body shape and the heartbeat are also assessed.
- 19th-22nd Week: The age-appropriate development of the unborn child is tested. The head, abdomen and thigh bones are measured as well as the placenta and the quantity of amniotic fluid.
- 29th-32nd Week: The placenta as well as the growth and position of the child are examined. This is important information for birth planning.

Extended basic ultrasound

In the 19th-22nd Week, you can also opt for an extended basic ultrasound, in which the head, brain, spine, abdomen and back as well as some organs are checked.

Diagnostic Doppler ultrasonography

This is recommended from the 20th Week, if it is suspected that the child is not being adequately nourished. The blood flow to the uterus and in certain blood vessels of the unborn is measured.

* p.m.: Pregnancy duration, calculated from the first day of the last menstrual period (post menstruation). This is the number gynaecologists and midwives count and enter into the mother's medical papers.

Precise diagnosis or organ ultrasound

This examination is also referred to as malformation ultrasound. It is performed with a high-resolution ultrasound device in special centres or practices. As of the 13th Week, many organs can be examined more closely, even the heart (echo cardiography). About half of all serious malformations can be seen at this time. Another organ ultrasound is almost always recommended to be performed in the 20th-22nd Week.

A 3D/4D ultrasound produces a spatial image of the unborn, but rarely provides medically important information.

HOW RELIABLE IS THE RESULT?

The quality of ultrasound findings depends primarily on the experience of the examiner and on the quality of the ultrasound device. A low level of amniotic fluid or an unfavourable position of the child complicates the assessment, as do scars or a thick belly of the pregnant woman.

Even under favourable conditions, every defect cannot be seen in the ultrasound, and false assessments can occur.

WHAT ARE THE EXAMINATION RISKS?

Normal ultrasound is considered harmless to the unborn child. With the Doppler ultrasound, heat is generated in the examined tissue, so it should only be applied in a justified way in the early pregnancy and be kept as short as possible. Modern ultrasonic devices have built-in control systems for this purpose.

COSTS

The basic ultrasound, also the extended version, is paid by the health insurance company, as well as further ultrasound examinations needed to clarify abnormalities. Patients that request ultrasounds must pay for them themselves.

PROS

- The results can already be discussed during the examination.
- An inconspicuous finding can be calming and reassuring.
- Seeing an image of the unborn can create a feeling of closeness to the child.

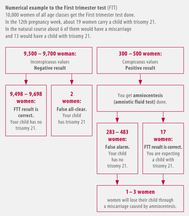
CONS

- Identified abnormalities, even insignificant ones, can be troublesome and often lead to further examinations.

FIRST TRIMESTER TEST OR FIRST TRIMESTER SCREENING (ETT OR ETS)

The probability for certain chromosomal abnormalities in the unborn is calculated from different blood values and ultrasound measurements. It also takes into account the age of the pregnant woman and the gestation period. The first trimester test only provides a risk classification, not a safe diagnosis.

This test includes a blood test in the 10th-14th week on two hormonal and protein values and an ultrasound in the 12th-14th week, in which the neck folds (= neck transparency, NT) and possibly further characteristics of the unborn child are measured. In the 19th-20th Week, the test can be supplemented by further blood tests. If the period for the first trimester test has passed, then the blood can only be examined for two, three or four proteins up to the 20th week. At that point, one speaks of the second trimester test.



WHAT CAN I LEARN?

From the results, the probability of a trisomy 21, 18 or 13 is calculated. An enlarged neck fold may also indicate a heart defect or other abnormalities. Additional chromosome aberration. In the 15th-20th week an increased AFP value* can be an indication of an open back or an unclosed abdominal wall.

The results are available within a few days. If it is conspicuous, a detailed ultrasound, a genetic blood test for chromosome aberrations and/or an amniotic fluid examination can provide more clarity.

HOW RELIABLE IS THE RESULT?

In the first trimester test, there are often false alarms: in approximately 96 out of 100 conspicuous findings, further studies show that the child has no chromosome aberration. The test does not recognise about 10 out of 100 chromosome deviations.

An example: 10,000 women of all ages undergo the first trimester test – in about 19 women, one can expect a trisomy 21 in the 12th week in her unborn child. In the natural course of things, six women will experience a miscarriage before birth.

- 9,500 to 9,700 women receive an inconspicuous finding. Among them are two women in whom the test does not recognise the child's trisomy 21– the test leads to a false reassurance.
- 300 to 500 women get a conspicuous result. Among them are 17 women whose child actually has a trisomy 21. For all others, it amounts to a false alarm.

The neck fold ultrasound alone as well as the second trimester test is less accurate than the first trimester test.

* AFP (Alphafetoprotein): Protein that is released into the blood and into the amniotic fluid by the child.

COSTS

The First Trimester Test has to be paid for and costs between 120 and 200 EUR.

PROS

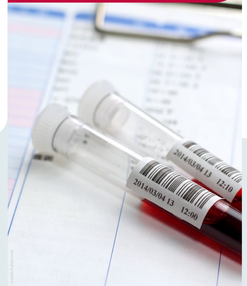
- Inconspicuous values calm down.
- Depending on the personal attitude of the woman, an invasive examination of the amniotic fluid is superfluous.

CONS

- The predicted probability data are difficult to understand and evaluate.
- The test often leads to further examinations in case of unclear or positive findings.
- 3 to 5 out of 100 women who undergo the test are unsettled by a false alarm.

GENETIC BLOOD TESTS FOR CHROMOSOME ABERRATIONS (NIPT)

From the blood of the pregnant woman traces of the child's genetic material are filtered out and sorted. The probability of certain chromosomal abnormalities of the unborn is estimated on their distribution. These "non-invasive prenatal tests" (NIPT) can be performed from the 10th pregnancy week. In particular, they are offered to women from the so-called risk group, whose first trimester test result was conspicuous, who experienced a chromosomal deviation in a previous pregnancy or in the family or who are over 35 years old.



WHAT CAN I LEARN?

The tests are designed to determine whether the chromosomes 13, 18 or 21 are triple (trisomy), and, depending on the manufacturer, if abnormalities of the sex chromosomes X and Y occur.

HOW RELIABLE IS THE RESULT?

In each case, three findings are possible and are specified separately for each chromosomal deviation: "Low risk", "high risk" and "unclear finding".

"Low risk" means: The chromosomal deviation can be excluded at approximately 100 percent.

"High risk" for trisomy 21 means for women from the risk group: 9 out of 10 women with this finding actually have a child with trisomy 21. For women who do not belong to the high-risk group and for other chromosome abnormalities, the test is less reliable. In other words, in such cases it often leads to false alarms.

A conspicuous result must be confirmed by a chorion villus biopsy or amniotic fluid test in order to rule out a false alarm.

In up to 5 out of 100 women, the test does not provide any evaluable results ("unclear findings"). In such cases the test can be repeated using a new blood sample.

COSTS

You usually have to pay for blood tests for chromosomal abnormalities yourself and they cost between 200 and 650 euros, depending on the scope and provider, plus about 50 to 60 euros for consultation and blood sampling.

PROS

- The result is much more accurate than the First Trimester Test.
- An inconspicuous test result makes it easier to forego an invasive examination.

CONS

- There are frequent false alarms among women who do not belong to the high-risk group.
- Approximately one to two weeks waiting time for the findings.

CHORIONIC VILLUS SAMPLING AND AMNIOTIC FLUID TEST (AMNIOCENTESIS)

These tests are used to determine the chromosome set of the child, in this case cells are taken from the body of the woman, which usually have the same set of chromosomes as the child. Under ultrasound surveillance, a fine hollow needle is advanced through the woman's abdomen (invasive examinations).

- Chorionic villus sampling (from week 11): A sample is taken from the placenta.
- Amniotic fluid test (from 15th week): The doctor advances a hollow needle into the amniotic sac and aspirates a sample of the amniotic fluid.

The child's cells are isolated from the sample in the laboratory and examined for their chromosome set.

As a result of both tests women may feel a slight pulling in the abdomen for a few hours to days. During the first days after the procedure, you should refrain from physical exertion and take a sick leave for two days.

WHAT CAN I LEARN?

The number and basic structure of the chromosomes are investigated. The sex can be revealed on request from the 12th week p.c. As a rule, AFP and AChE* are determined in addition to the amniotic fluid. Elevated values may indicate an open spine or an open abdominal wall.

Genetic analysis for certain hereditary diseases, e.g. muscle or metabolic diseases, is only performed if the disease occurs in the family or there are indications from other analyses.

With the chorion villus biopsy a provisional result is obtained after one to seven days, the final results after about two weeks.

For the amniotic fluid test, a partial result (FISH test) regarding the chromosomes 13, 18, 21 as well as the sex chromosomes X and Y can be prepared upon request after one to two days The waiting time until the final results is two to three weeks.

HOW RELIABLE IS THE RESULT?

More than 99 out of 100 women receive an accurate result. Sometimes not all the cells examined have the same chromosome pattern (mosaic), in such cases the results are unclear. This is more common in the chorionic villus sampling than in the amniotic fluid test.

Sometimes no evaluation is possible and a decision has to be taken about a further puncture.

WHAT ARE THE EXAMINATION RISKS?

The puncture can lead to contractions or amniotic fluid leakage and in the worst case trigger a miscarriage. This affects 1 to 4 of 200 women after chorionic villus sampling. After an amniotic fluid test, 1 out of 200 women lose their child.

With the amniotic fluid test, an injury to the unborn is possible, but it is extremely rare..

* AChE (acetylcholinesterase): Protein, which is released by the baby into the blood and the amniotic fluid.

COSTS

The costs are borne by the health insurance company, if the examination is medically justified.

PROS

- Reliable results
- Chorionic villus sampling: possible early in pregnancy (11th week) and fast preliminary results (1 to 7 days)
- Amniocentesis: additional information on open spine/ open abdominal wall

CONS

- Miscarriage risk is often higher than the probability of chromosome aberrations
- Two days physical rest and possible work loss
- 14 to 21 days waiting time until the final results.
- Building a relationship with the child can be delayed.

PERCUTANEOUS UMBILICAL BLOOD SAMPLING (CORDOCENTESIS, CHORDOCENTESIS)

During umbilical blood sampling (from the 18th week), a fine hollow needle is advanced through the abdomen and the amniotic sac into the umbilical cord vein under ultrasound guidance. Then some blood of the unborn is drawn. The test is carried out only in special centres.

WHAT CAN I LEARN?

The blood of the child is examined for anaemia, Rh incompatibility or treatable infections. A chromosome analysis and gene analysis are also possible.

HOW RELIABLE IS THE RESULT?

The findings are very reliable as the blood of the child is examined directly.

WHAT ARE THE EXAMINATION RISKS?

The puncture can cause contractions or a bleeding; in 1 to 3 out of 100 women it leads to a miscarriage.

COSTS

The costs are borne by the health insurance company, if the examination is medically justified.

PROS

- Reliable results
- An immediate blood transfusion is possible in cases of anaemia.

CONS

- Increased miscarriage risk
- Only possible in special centres

pregnancy

Prenatal diagnosis

Advice, methods and assistance. An overview

BZgA
Bundeszentrale für gesundheitliche Aufklärung

Dear Reader,

Being pregnant, experiencing a birth, becoming mother and father: family planning and pregnancy almost always means an intense phase of feelings, desires, hopes and also uncertainties. If you want to become pregnant or are already pregnant, ask yourself: How will the child change my/our life and everyday living? What can I do for my child? And will it be healthy? Even though a lot of health risks are being discussed nowadays, most children are born healthy.

> A very tiny number diseases and disabilities are congenital and only a small number of those are recognisable before birth.

The doctor or midwife monitors the pregnancy and the development of the child during the examinations of the pregnant woman. In addition, gynaecological practices also offer various prenatal tests, which look for impairments in the unborn child. You must pay for some of these examinations yourself, as the costs are not covered by the statutory health insurance. You can decide for yourself whether you want to use such examinations, and if so, which ones.

WHAT IS **PRENATAL DIAGNOSIS?**

Tests that search specifically for indications of malformations or disorders in the unborn are summarised under the term “prenatal diagnosis”. These include certain ultrasound examinations, blood tests as well as examinations of early placental tissue (chorionic villus sampling) or amniotic fluid.

In so doing, there is a differentiation

- between non-invasive tests which do not interfere with the woman’s body and provide a first individual risk assessment (such as the first trimester test, see below)
- and invasive procedures, which encroach on the woman’s body. These tests provide more accurate results on malformations or abnormalities in the unborn child. This includes, for example, an amniocentesis, in which a needle penetrates into the woman‘s body. It is associated with an increased risk of losing the child through a miscarriage.

> The test results usually do not say much about how serious a disease or disability is and what limitations it will bring with it.

Ultrasound plays a dual role in antenatal care. It helps to check the normal course of pregnancy. However, it is also possible to detect abnormalities of the unborn child, which can force parents to face difficult decisions. With high-resolution ultrasound, a specialist practice can also search for malformations of the unborn child (detailed diagnosis).

More information on the most common methods and their meaning can be found on the back of this leaflet.

“WHAT EXAMINATIONS SHOULD I HAVE DONE?
WHAT DO I WANT TO KNOW
ABOUT MY CHILD DURING PREGNANCY?
CAN I REFUSE PRENATAL EXAMINATIONS?”

MEDICAL **EDUCATION** AND **COUNSELLING**

All examinations are voluntary. That is, you have a right to know, but also a right not to know. In any case, it is advisable to know in advance which offers you want to use and which you do not.

Your doctor is legally obliged to inform you before prenatal diagnosis about the nature of the test and its risks, and to discuss possible results and the subsequent consequences with you. This applies to the chorionic villus biopsy and the amniotic fluid examination, but also to specialised ultrasound examinations, the first trimester test and the genetic blood test for chromosome aberrations.* The doctor must also point out to you that you are entitled to psychosocial counselling in a pregnancy-counselling centre.

REASSURANCE OR **UNCERTAINTY?**

Many women and couples hope to receive the soothing certainty that their child is healthy from prenatal diagnostic tests, although the tests can provide differing indications that the unborn child does not have certain impairments. However, prenatal diagnostics cannot provide a guarantee of a healthy child. Only a certain number of impairments can be determined at all during pregnancy. Some of the tests can only estimate how likely a deviation from the norm is, and can cause parents to become worried by false alarms. In addition, the tests can also overlook diseases or disabilities.

Nevertheless, a normal result can calm pregnant women if, for example, they have already lost a child or have an inherited disease in the family. Other women experience the constant stress of medical risks which can become a burden that

* Chromosomes contain the entire genetic information (genes). Each human cell contains a chromosome set. This consists of 22 chromosome pairs and two sex chromosomes (XX or XY) in most people. If the chromosomal set is altered, medicine calls this a chromosome disorder or variation.

interferes with the development of a relationship with the child – especially in the first trimester of pregnancy. Unclear findings can overshadow the pregnancy. If they lead to further prenatal examinations, the period of anxiety and uncertainty is prolonged.

> No therapy is available for most prenatal disabilities or diseases.

HOW DO I MAKE **A DECISION**?

Prenatal diagnostic tests do not protect against disabilities, but instead look specifically for abnormal deviations in the unborn child. In some cases, the early knowledge of a disease can improve the child’s start in life. For example, in the case of a heart defect of the child, the childbirth can be planned specifically in a specialised hospital with a neonatal intensive care unit. However, in most cases no treatment is available. Then the parents-to-be will face a difficult decision: Do we take our child as it is, or do we want to abort the pregnancy?

“WHAT DO I DO AFTER AN UNUSUAL FINDING?
HOW DO I IMAGINE LIFE WITH A DISABLED CHILD?
WHAT SUPPORT IS AVAILABLE? WHAT IMPACT DOES THIS
HAVE ON OUR PARTNERSHIP AND THE OTHER CHILDREN?”

THE **ABORTION**

After the twelfth pregnancy week after conception (pc)* a pregnancy may only be interrupted if there is a medical indication. This means that the continuation of the pregnancy seriously jeopardises the physical or mental health of the pregnant woman, according to medical opinion, and this danger cannot be reasonably averted in any other way. Only then is a pregnancy termination legally permissible.

Furthermore, the Pregnancy Conflict Act prescribes that a three-day consideration period be provided after the notification of the diagnosis. Only then can the written indication be issued that is necessary for the abortion. The pregnant woman and her family can use this time for an independent consultation. They have a legal right to talk to a pregnancy counselling centre to clarify their feelings and thoughts and make a decision.

The parents will therefore not be alone on the termination of the pregnancy. And in the case of an abnormal test result, the medical consent to terminate is not automatically granted. Particular care is taken with regard to the medical indication after the 20th week of pregnancy p.c., because at that point the child is already viable outside the mother‘s body.

“HOW DO I MANAGE AN ABORTION?
WHO CAN SUPPORT ME? HOW CAN I SAY FAREWELL?”

In the event of a pregnancy termination after 12 weeks p.c., a miscarriage is initiated with labour pain inducing drugs. The process can last from a few hours to a few days and takes place in a delivery room or a labour room. The woman is supervised by a midwife and a doctor.

For more information, please visit
www.familienplanung.de/pnd.

* p.c.: Pregnancy period, calculated from conception (post conceptionem). This is how the law regulating the termination of pregnancy (§ 218 StGB) counts days and weeks.

CONSULTING – ORIENTATION AND **ASSISTANCE**

The numerous medical tests, technical terms and findings can affect your own feelings about pregnancy and the child. Some women and couples are looking for a place where they can talk about their questions, fears and thoughts. Experience has shown that in addition to medical facts, feelings, life experience and intuition play an important role in decision-making.

A pregnancy counselling centre can be a place where you can get answers to your questions. Together with a consultant, you can calmly consider and find your own way.

You can take the advice before the prenatal diagnosis, in the time waiting for the results and/or after the findings. You will also find support there before and/or after a pregnancy termination based on medical indication. The consultation is confidential and free of charge.

Likewise, counsellors can accompany you if you are expecting a child with disabilities and, among other things, provide you with contacts to a self-help group or to support groups for disabled people. A counselling centre can provide you with expert information on financial assistance and other support.

On **www.familienplanung.de/beratungsstellensuche** you will find a counselling office near you.

WHICH **DISABILITIES** CAN BE **RECOGNISED PRENATALLY**

Diseases of the newborn can have many causes, such as infections for the mother, a premature birth or birth complications. A few of the abnormalities are inherited or happen randomly during the fusion of egg and sperm cells. These include chromosomal aberrations that can be detected by the first trimester test, the genetic blood test, the chorionic villus biopsy or the amniotic fluid examination. Heart defects and other organ deficiencies are usually identified in the ultrasound.

If the unborn child has a serious disability, a miscarriage often occurs, often in the first trimester of pregnancy. Therefore, early tests also detect abnormalities in unborn babies where the pregnancy would have ended by miscarriage itself.

Malformations of the heart, kidneys, central nervous system and skeletal system are most frequently detected. About ten percent of the conspicuous findings affect the chromosomes.* The following is an overview of some deviations in the developing child. The data on frequency of occurrence are based on estimates and extrapolations. They indicate approximately how many women learn that their child has or had a disability in pregnancy, after childbirth or after a miscarriage.

* Source: EUROCAT, a European network for the detection of childhood anomalies that occur during pregnancy or after birth. Limited regions from 23 countries deliver data to EUROCAT (www.eurocat-network.eu). From Germany, Sachsen-Anhalt and the Mainz region report their findings.

Heart defects/abnormalities

Heart defects usually only become evident after birth, when the child’s circulatory system changes and the newborn has to breathe independently. Many early heart defects are easily treatable and the children can often lead a largely normal life. Childhood cardiac arrhythmias can sometimes be treated during pregnancy by women taking medication.

In 80 out of 10,000 pregnant women, the unborn child has a heart defect, though the extent of those defects can vary greatly.

Malformations on the spine, spinal cord and brain

These malformations are also referred to as neural tube defects and can vary greatly. For example, spina bifida can cause slight paralysis leading to difficulty walking, but paraplegia (total paralysis from the waist down) is also possible. In addition, the head can be greatly enlarged if the drainage of cerebrospinal fluid (CSF) is restricted (this is known as hydrocephalus). If such malformations are detected early, usually with ultrasound, this allows careful birth planning and competent medical care for the newborn in a special centre.

The most serious of these malformations is anencephaly. This is where parts of the skull and brain have not developed and the children die shortly after birth.

Approximately 8 out of 10,000 pregnant women are carrying a child with neural tube defects, of which about every other child has spina bifida.

Chromosome deviations

Trisomy 21 (Down Syndrome)
These children usually develop more slowly, initially recognisable in their movements, later also mentally. Some need a lot of support; others can gain a school-leaving certificate and pursue a profession. How much the child as a whole will be impaired cannot be predicted during pregnancy. Almost half of the children with trisomy 21 have a heart defect, and about seven percent a bowel constriction or an intestinal obstruction, which can be remedied by surgery.

Among 10,000 pregnant women are about 20 whose child has Down Syndrome. This corresponds to a risk of 1:500. The age of the woman increases the probability of Down Syndrome: in a 25-year-old it is 1:1,400 at the end of pregnancy, 1:360 for a 35-year-old and 1:100 for a 40-year-old.

Trisomy 18 (Edwards Syndrome)
and **Trisomy 13** (Pätau Syndrome)
Children with these chromosomal abnormalities often have malformations to the head and brain, heart, limbs and/or other organs. Most die in pregnancy or in the first weeks after birth. Of 10,000 pregnant women, about 5 have a child with trisomy 18 and 2 a child with trisomy 13.

Monosomy X (Ullrich-Turner Syndrome)
This is a deviation of the sex chromosomes and only girls are affected. They are smaller than average and almost always sterile. Their mental development is normal. Children have an Ullrich-Turner syndrome in approximately 2 out of 10,000 pregnant women.

Klinefelter’s syndrome
This deviation of the sex chromosomes affects only boys and often remains unnoticed. The boys are usually above average in size, sometimes slowed in their motor and mental development, and almost always infertile. Among 10,000 pregnant women are about 10 whose unborn child has Klinefelter‘s syndrome.

On **www.familienplanung.de/pnd** you will find further information about the individual testing methods.

pregnancy

You can find out where you can get counselling in your region on
www.familienplanung.de/beratungsstellensuche
or at the following institutions:

AWO Arbeiterwohlfahrt Bundesverband e.V.
Tel.: +49(0)30/26 30 9-0 | www.awo-schwanger.de | www.awo.org

Deutscher Caritasverband e.V.
Tel.: +49(0)761/2 00-0 | E-Mail: info@caritas.de or Contact form
www.caritas.de → Help and counselling

Der Paritätische Gesamtverband
Tel.: +49(0)30/24636-0 | E-Mail: info@paritaet.org | www.paritaet.org

Deutsches Rotes Kreuz e.V.
Tel.: +49(0)30/8 54 04-0 | www.drk.de

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pro familia Bundesverband e.V.
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On **www.familienplanung.de/pnd** you will find further information about the individual testing methods.

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