

Brief information about

Early prenatal diagnosis

Engelska



Early prenatal diagnosis

It is important to note that all prenatal diagnosis tests are voluntary. It is always the pregnant woman who decides whether to have a test or not.

Early prenatal diagnosis is defined as tests performed before 22 completed weeks of pregnancy. However, most tests are performed much earlier.

If you undergo prenatal diagnosis tests, the results may indicate that the fetus has a chromosomal disorder and/or another malformation. If this is the case, you will be asked if you want further tests. You will be given information about the medical and social consequences of the disorder and about the care and treatment that can be offered. It will then be up to you to decide whether you want to continue or terminate the pregnancy. If you feel doubtful about making this decision, you should consider whether you really want to undergo prenatal diagnosis tests. Please note that the early and routine ultrasound scans (see below) are also prenatal diagnosis tests.

It is also important to remember that some conditions cannot be detected by prenatal diagnosis. Even if the examination or test results seem completely normal, there is still a slight risk that there may nonetheless be disorders that can affect the fetus's health.

If you have a family history of chromosomal disorders or hereditary illness, you should inform your midwife or doctor.

The public health system offers prenatal diagnosis tests free of charge, based on a medical assessment. If you undergo prenatal diagnosis at a private clinic, you may not be offered the same test at a public health facility. Ask your midwife whether this applies to you.

Early ultrasound scan (tidig ultraljud, TUL)

An early abdominal ultrasound scan is offered free of charge to all pregnant women in Västra Götaland. The purposes are to determine the length of the pregnancy, to identify pregnancies with more than one fetus and to detect serious malformations. The early scan should ideally be done between pregnancy weeks 12+4 and 13 + 6, but it can be scheduled up to week 15 + 0. Chromosomal disorders cannot be detected at the early scan.

CUB (KUB)

The CUB (combined ultrasound and biochemical test) estimates the **likelihood** of disorders involving an extra chromosome (trisomies). The CUB can estimate the likelihood of trisomy 21 (Down syndrome) and two other, more uncommon but more serious, chromosomal disorders (trisomy 13 and 18).

It is important to note that the CUB does not give a firm diagnosis of trisomy. Instead, it is a tool to **calculate the likelihood** of trisomy.

In Västra Götaland, the CUB is offered free of charge to all pregnant women aged 35 and up on the date of the last menstrual period.

A blood test is taken from the pregnant woman at pregnancy week 9+0 - 13+6 and levels of two proteins are measured. An abdominal ultrasound scan is done at pregnancy week 11+2 - 13+6, in order to measure the width of the fluid-filled space at the back of the fetus's neck (nuchal translucency). The results of these tests are combined with the woman's age to calculate the likelihood that the fetus has a trisomy. Optimally, the blood test should be taken at week 10+0. The scan should ideally be done at week 12+4 -13+6, because it is possible to do a high-quality early scan and determine the due date at the same time.

It is possible to do the CUB in a twin or other multiple pregnancy, but there may be difficulties making decisions based on the results. In these cases, specific counselling may be helpful.

If the CUB indicates a higher likelihood of a trisomy, further tests will be offered.

NIPT (non-invasive prenatal test)

The NIPT entails taking a blood test from a pregnant woman in order to examine the fetus's DNA. Like the CUB, the NIPT calculates the likelihood of a chromosomal disorder, but with a higher degree of accuracy. Like the CUB, the NIPT estimates the likelihood of trisomies 13, 18 and 21. It can be performed at pregnancy week 10 and onward.

The NIPT is offered in cases:

- in which a CUB has indicated a moderately elevated (1:51-1:200) likelihood of a trisomy

- in which amniocentesis or chorionic villous sampling (see below) are inappropriate (e.g. hepatitis, HIV, bleeding disorders)

The NIPT cannot be performed if there is more than one fetus, after oocyte donation, if the woman has a chromosomal disorder herself or if she has had any of the following during the last three months:

- transplantation (including stem cell transplantation)
- blood transfusion
- radiation therapy
- immunotherapy

Amniocentesis/chorionic villous sampling (AC/CVS)

A thin needle is inserted into the pregnant woman's abdomen to sample either the placenta or the cells in the amniotic fluid. The sample is most often analyzed with a method (QF-PCR) that yields results within a week for the trisomies 13, 18 and 21.

- Amniocentesis can be done at pregnancy week 15+0 at the earliest.
- CVS can be done at pregnancy week 11+0 at the earliest.
- These tests entail an increased risk of miscarriage of less than 0.5%.

Amniocentesis or CVS is offered in these cases:

- a CUB indicates a likelihood above 1:50, if the nuchal translucency is ≥ 3.5 mm or if the NIPT indicates a high likelihood of trisomy 13, 18 or 21
- there is a suspicion of malformation at an ultrasound scan
- previous baby or pregnancy with a chromosomal disorder
- history or family history of hereditary disease
- in exceptional cases, at age 35 and up, if the woman declines a CUB but wants a prenatal diagnosis test for chromosomal disorders

Routine ultrasound scan (rutinultraljud, RUL)

The routine ultrasound scan is offered free of charge to all pregnant women. It is performed at pregnancy week 18-20 to:

- determine pregnancy length and the date of delivery (unless this has already been determined at the early scan; see above).
- diagnose multiple pregnancies (if no early scan has been done)
- to detect serious malformations

- determine the location of the placenta

Some malformations (such as large spina bifida) are easy to see with ultrasound, and they are thus almost always detected. Other malformations (such as some heart defects) are more difficult to detect. Furthermore, not all malformations are detectable with ultrasound. In rare cases, a healthy fetus is mistakenly assessed as having a disorder. Sometimes it can be difficult to determine the significance of an unusual finding. This can result in uncertainty that can be difficult for the parents.

If you want to know the sex of the fetus, please tell the midwife before the scan. If the sex can be determined with a reasonable degree of certainty, s/he will tell you what it is.

What happens after the prenatal diagnosis tests?

In almost all cases, the test results are normal and the baby is completely healthy at birth. If, on the other hand, the tests indicate something abnormal, a doctor will make an assessment. Further tests may be offered and you may be asked whether you want an amniocentesis or a CVS. If your fetus does have a chromosomal disorder and/or some other malformation, you will be given extensive information and the opportunity to discuss what this means. No other person has the right to interfere with or try to influence the pregnant woman's decisions. However, sometimes it can be helpful to speak with a knowledgeable professional, such as a genetic counselor, pediatrician, psychologist or social worker, in order to arrive at the best decision for you. The links below can also be helpful, in order to learn more about what it means to be the parent of a child with a disability.

If you want to know more about prenatal diagnosis:

- Read the information on your local obstetrics department's home page.

Web links:

<http://www.1177.se/Fakta-och-rad/Undersokningar/Fosterdiagnostik/>

<http://www.fub.se>

<http://www.svenskadowndforeningen.se>

<http://www.agrenska.se>

<http://www.gensvar.se>

<http://www.socialstyrelsen.se/funktionshinder>

<http://www.nkkf.nu>