

Information for parents about prenatal screening tests

Screening for fetal chromosomal and congenital abnormalities

Screening tests during pregnancy are voluntary and free-of-charge. Prenatal screening tests are applied for through the Child Welfare Clinic (neuvola). We hope that the person expecting will come to the screenings with their spouse or support person without children.

The person expecting has the opportunity to participate in two screening ultrasound scans, one of which is done in early pregnancy and the other in mid-pregnancy. Screening for fetal chromosomal abnormalities takes place in early pregnancy with the so-called NIPT (non-invasive prenatal testing) examination. Chromosome screening assesses the risk of trisomy 21 (Down syndrome), trisomy 18 (Edwards syndrome) and trisomy 13. The risk of significant congenital abnormalities is screened in the mid-pregnancy structural ultrasound scan.

Before you decide to participate in prenatal screenings, it is good to consider the possible consequences. The results of screening tests are usually normal, but the results can also indicate an increased risk of an abnormality or illness for the fetus, in which case further follow-up tests are possible. After the results are out, you may even be faced with the choice of continuing or terminating the pregnancy. Although screening tests are reliable, they do not detect all chromosomal and congenital abnormalities. It should also be noted that not all newborn diseases are caused by chromosomal or congenital abnormalities. Rarely, despite normal screening findings, a newborn child may be diagnosed with a chromosomal or congenital abnormality after birth (a false negative screening finding) and correspondingly a suspicion of an abnormality may arise during the screening, even though the child is healthy (a false positive screening finding).

Do you participate in prenatal screening?

Diagram of different screening options



A. Yes, I participate in chromosomal and congenital abnormalities screenings

In this case, you will participate in two ultrasound scans, one of which is done in early pregnancy and the other in mid-pregnancy.

An early pregnancy ultrasound scan is scheduled for weeks 12–14 of pregnancy, when it is confirmed that the fetus is alive, the duration of the pregnancy is estimated, the number of fetuses and placentas is taken into account, and the person expecting is referred for a NIPT sample.

A structural ultrasound examination is performed around 21 weeks of pregnancy.

B. I do not want to participate in chromosome screening, but I choose ultrasound scans

In this case, you will participate in an early pregnancy ultrasound scan during the 12-14th week of pregnancy and a mid-pregnancy structural ultrasound scan either around 21 weeks or after 24 weeks of pregnancy.

C. I do not want to participate in screening for fetal chromosomal abnormalities or in ultrasound scan

If you do not want to participate in chromosome screening or ultrasound scans, your pregnancy will be monitored in your Child Welfare Clinic (neuvola).

More detailed information about screenings

Chromosomal abnormality screening by NIPT

After an ultrasound examination, all mothers in early pregnancy can participate in a voluntary NIPT (Non-invasive prenatal test) examination, which investigates the most common chromosomal abnormalities of the fetus. The test is based on examining the fetal genetic material (DNA) in the mother's blood. We screen trisomies 21, 18 and 13 by using the NIPT test. The sensitivity of NIPT screening for trisomy 21 is about 99% and for other abnormalities about 93%.

After the mother's blood sample is taken, the result will be ready in about a week, and you can find the NIPT answer in MyKanta. If the result of the NIPT test is abnormal, you will be informed of the result by phone and get information about follow-up tests. The fetal chromosome finding is confirmed by an amniocentesis.

Screening for congenital abnormalities

Structural ultrasound scan in mid-pregnancy

Mid-pregnancy ultrasound scan is done at around 21 weeks of pregnancy. Its purpose is to identify severe congenital abnormalities of the fetus. A midwife performs the screening scan over the abdomen. If necessary, a midwife consults a doctor or allocates you for further follow-up tests. Fetal growth and structures, the amount of amniotic fluid and the location of the placenta will be examined in the screening scan. If the findings of the ultrasound scan are abnormal or screening cannot be performed reliably, you will be referred for follow-up tests at Majakka hospital's Maternity Outpatient Clinic.

The purpose of the follow-up tests is to give parents information about a possible congenital abnormality and its effect on the life of the fetus and newborn. If necessary, in addition to the obstetrician, a geneticist, a pediatrician, a pediatric cardiologist and/or a pediatric surgeon are involved providing guidance.

According to Finnish legislation, if the parents wish, they can apply for permission to terminate the pregnancy if the fetus is found to have a severe chromosomal or structural abnormality. A pregnancy termination permit is applied from Valvira, which can grant a pregnancy termination permit up to 24+0 weeks of pregnancy.

A structural ultrasound scan performed after the 24th week of pregnancy

Instead of a mid-pregnancy ultrasound, you can participate in a structural ultrasound scan after 24 weeks of pregnancy. Determining a congenital abnormality during pregnancy can improve the prognosis of the unborn child by directing the pregnancy monitoring and delivery to a hospital, where the pregnant woman and the unborn child will receive the best possible help. A pregnancy termination permit cannot be applied due to a fetal congenital abnormality after 24+0 weeks of pregnancy.

Noteworthy

- Participation in all screening tests and follow-up tests is voluntary.
- Screening tests do not find all chromosomal and congenital abnormalities.
- If you are referred to the maternity outpatient clinic because of your own illness or an increased risk of chromosomal and congenital abnormalities of the fetus, an early pregnancy ultrasound scan or a structural ultrasound scan by a doctor can be combined with your outpatient clinic visit, and the outpatient clinic visit will then be charged an outpatient clinic fee.

You can get more information about prenatal screening tests, e.g.:

- From the health nurse and doctor of your own Child Welfare Clinic (neuvola)
- From the website of Tyks Maternity Outpatient Clinic
- <u>Terveyskylän Naistalo</u> (in Finnish)