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Screening for foetal chromosomal abnormalities

The purpose of this leaflet is to provide the patient with information about the purpose, nature, and interpretation of the results of screening for foetal chromosomal abnormalities.

The screening for foetal chromosomal abnormalities helps to identify pregnant women whose unborn children are at an increased risk of having a chromosomal abnormality (risk estimation is given for Down, Edwards and Patau syndromes) or a developmental disorder. Screening consists of a blood test and an ultrasound scan. In addition, the first trimester screening also assesses the risk for preeclampsia.

When and how does the screening take place?

Between 11 weeks to 13 weeks + 6 days of pregnancy, blood will be drawn, and an ultrasound scan will be performed. Based on the results of your blood test, ultrasound scan, and other data (age, weight, etc.), a computer program will calculate the probability of your future child having any of the most common chromosomal abnormalities.

Screening results

- **Low risk** – the risk is less than 1:1000. No further tests are needed.
- **Intermediate risk** – between 1:11-1:1000 for Down syndrome or between 1:11-1:100 for Edwards and Patau syndrome. In this case, we will offer you the option to perform an additional NIPT test (cell-free foetal DNA test using the mother's blood). A blood sample is collected and sent to the laboratory for testing. The results will be available in 10 working days, and will show whether the risk for chromosomal abnormalities is high or low. If NIPT indicates a high risk, we will offer you the possibility of a diagnostic test – amniocentesis (from 15 weeks of pregnancy).
- **High risk** – if the risk is 1:10 or higher, or the nuchal translucency measurement is 3.5 mm or more, we will offer you a diagnostic test (an invasive procedure - chorionic villus sampling or amniocentesis). In these cases, NIPT is not indicated.

Intermediate and high-risk results do not mean that your future child has a chromosomal abnormality, but that additional testing is indicated. False-positive results occur in 3.5-5% of cases. False-negative results are rare.

If the results of the first trimester screening show that you have an increased risk of developing preeclampsia, and there are no contraindications, aspirin prophylaxis will be recommended. Also, an additional ultrasound scan will be performed during the third trimester.

Additional diagnostic tests

Chorionic villus sampling – usually performed between the 12th and 13th week of pregnancy. Under ultrasound guidance, a small piece of tissue is collected from the placenta through the abdominal wall with a fine needle and sent to the laboratory for examination. The results will be available in about three weeks.

Amniocentesis can be performed from the 15th or 16th week of pregnancy. Under ultrasound guidance, a small amount of amniotic fluid is collected through the abdominal wall with a fine needle, and the foetal skin cells floating in the fluid are examined. The results will be available in approximately three weeks.

Additional diagnostic tests are voluntary. Before the procedure, the nature, purpose, risks, and possible consequences will be explained to you, and written consent will be obtained.

If it turns out that the foetus has a chromosomal abnormality, the geneticist will talk to you about the results of the test and the prognosis of the abnormality. According to Estonian law, a pregnancy can be terminated until the end of the 21st week of pregnancy if the unborn child is likely to have a severe mental or physical health problem. If you decide to terminate the pregnancy, contact your doctor or midwife for more information.

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Approved by the decision of the Medical and Nursing Care Quality Commission of *Aktsiaselts Ida-Tallinna Keskhaigla* on 26. August 2026 (Protocol No. 2.2-8/11-26)