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Screening for foetal chromosomal abnormalities

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

The purpose of screening for foetal chromosomal abnormalities is to identify pregnant women whose unborn children are at increased risk of developing a chromosomal abnormality (primarily Down, Edwards and Patau syndromes) or another type of developmental disorder). Risk of chromosomal abnormalities is determined using combined screening – a blood test and an ultrasound – to assess the markers for chromosomal abnormalities. Screening only assesses the risk of foetal chromosomal abnormalities.

Blood tests and ultrasound examinations are performed from **11 weeks to 13 weeks and 6 days of the pregnancy.**

A computer program is used to calculate the likelihood of your chid having any of the most common chromosomal abnormalities based on your blood analysis, ultrasound results and other data (age, weight, etc.).

Screening results

- If screening indicates **low risk** (over 1:1000), there is no need to perform any additional tests.
- If screening indicates **medium risk** (between 1:11 and 1:1000 for Down syndrome or between 1:110 and 1:100 for Edwards and Patau syndrome), we offer the option to perform a NIPT test (cell-free foetal DNA test using the mother's blood) free of charge. To this end, we will use venous blood for lab testing. You will be informed of the results within 10 business days. If the NIPT test indicates low risk, no further testing is required. If the NIPT test reveals high risk, we offer either a chorionic villus biopsy or amniotic fluid testing to specify the diagnosis.
- If screening indicates **high risk** (1:10 or above or if nuchal translucency measures 3.5 mm or more), we will immediately offer the chance to undergo a chorionic villus biopsy or amniotic fluid testing.

Medium and high risk does not mean your future child will have chromosomal abnormalities. The rate of false positive results is 3.5-5%. False negatives are rare.

Additional testing available

Chorionic villus biopsy is usually performed during weeks 12-13 of the pregnancy. It involves taking a small tissue sample from the growing placenta with a thin needle through the abdominal wall under ultrasound guidance. The tissue sample is then sent to the laboratory for examination. The results come back within two weeks.

Amniocentesis is usually performed starting from week 16 of the pregnancy. It is used to examine the foetal cells present in amniotic fluid. A small amount of amniotic fluid is taken from the amniotic sac with a thin needle through the abdominal wall under ultrasound guidance. The results of amniocentesis come back within three weeks.

Additional testing is voluntary and you have the right to decide whether you would like to take them. You will be informed of the nature, purpose, risks and consequences of any tests beforehand and asked to sign a consent form.

If test results put the foetus at high risk of chromosomal abnormalities, the results of the test as well as the diagnosis outlook will be explained to you. According to the Termination of Pregnancy and Sterilisation Act of Estonia, pregnancy may be terminated before the end of the 21st week if it is likely that the unborn child has a severe mental or physical disability. If you decide to terminate your pregnancy, contact your doctor or midwife for further information.

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