Non-invasive prenatal test (NIPT) – PATIENT INFORMATION

NIPT TEST ALTERNATIVES

The most common chromosome abnormalities of the fetus are 21 trisomy (Down syndrome), 18 trisomy (Edwards syndrome) and 13 trisomy (Patau's syndrome) and sex chromosome copynumber abnormalities like monosomy X (Turner's syndrome). New **N**on-**I**nvasive **P**renatal **T**est (**NIPT**) enables to measure the risk for the most common chromosome abnormalities from maternal blood sample. The test utilizes cell free fetal DNA (cffDNA) in the maternal plasma sample, which can be detected from pregnancy week 10. The aim of the NIPT test is to measure whether fetus has aneuploidy of studied chromosomes/chromosome regions (either too much or missing chromosomes). Basic NIPT-test detects trisomies 13, 18, 21 and the sex chromosomes (B –NIPTtri, KL 6373) and when requested, also six common microdeletions [22q11.2,15q11.2 (AG/PWS), 1p36,4p-/WolfHirshhorn,5p-/Cri du Chat] (B –NIPTdel, KL 6374). In twin pregnancies trisomies 13,18 and 21 can be detected and the presence of Y chromosome. The sex of the fetus is given if the patient wish to know it (marked in the NIPT test request form). More information is available of the NIPT test on our web pages: www.yml.fi.

ADVANTAGES OF THE NIPT TEST

The main advantage of the NIPT test is that it is non-invasive, which means that the fetus is untouched with this procedure. Therefore by non-invasive test the miscarriage risk can be avoided. In addition the NIPT test is much more specific and reliable than other screening tests and the result is achieved quickly in the first trimester period of the pregnancy. NIPT test is possible to be performed from pregnancy week 10.

LIMITATIONS OF THE NIPT TEST

The test measures the aneuploidy risk for the studied chromosomes/chromosome regions. Therefore the test does not exclude any other chromosome abnormality not included in the test panel. Even the NIPT test is reliable, there is a low risk for false result (false positive results are detected 0,2% and false negative results 0,02%). A remarkable overweight of the mother (>~120-140 kg) may decrease the success rate of the NIPT test (the amount of cffDNA may be too low). The NIPT test is a screening test (not diagnostic) and the abnormal result need to be confirmed using an invasive test from amniotic fluid or chorionic villus sample.

NIPT test can be performed also in twin pregnancies and pregnancies which have been given IVF treatments. In twin pregnancies trisomies 13,18 and 21 can be detected and the presence of Y chromosome. The sex of the twins is reliably detected only if both fetuses are girls (Y chromosome not detected). If one of the twins is abnormal, that cannot be specified which one. Therefore an invasive test is needed to confirm the abnormal result.

NIPT TEST IN PRACTICE

A normal blood sample is needed for NIPT test. Precounseling concerning the NIPT test is given before the procedure and a special NIPT test request is filled. The tests are send to Genesis Genetics in London (Cooper Genomics UK). By signing the request form patient agrees to have been informed enough of NIPT testing, which includes the meaning, benefits, risks and limitations of this test (Patient information). All clinical information concerning the test and results are confidential. Yhtyneet Medix laboratoriot takes care of the sample logistics and is responsible of the handling and reporting the result to the healthcare provider. The patient gets the result and may ask more information from the healthcare provider if needed.