E. Gulbja Laboratorija

NIPT

INFORMATION FOR A PATIENT

WHAT IS NIPT ND WHY IT IS BEING DONE?
NIPT (Non Invasive Prenatal Test) or non-invasive prenatal examination of the chromosomes of the fetus in maternal bloodstream is a test which is being performed during your pregnancy to make sure that the set of fetal chromosomes is normal.

NIPT can provide valuable information in the following cases:

- pregnancy screening results indicate an elevated risk
- abnormal ultrasound results
- maternal age above 35 years
- medical history of aneuploidies

The test can also be performed in the case when the risk is not elevated.
In most cases the test allows to avoid invasive manipulations – CVS or amniocentesis which carry the risk of spontaneus abortion (~1%).
NIPT can be performed starting from the 10th week of pregnancy although the recommended time for the test is right after the pregnancy screening test results are available. The test can also be done in twin pregnancies.

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WHAT IS DETECTED BY NIPT AND NIPT PLUS TESTS?

NIPT

Intended for the detection of the most common chromosomal aneuploidies:

- Trisomy 13 – Patau syndrome
- Trisomy 18 – Edwards syndrome
- Trisomy 21 – Down syndrome
- Sex chromosome abnormalities (X and Y)

NIPT PLUS

Intended for screening of aneuploidies of all chromosomes as well as the detection of the most common microdeletions: Cri du chat, 1p36 deletion, DiGeorge II, Wolf Hirschorn, Prader-Willi/Angelman, Jacobsen, Langer Giedion, Phelan McDermid, 16p11 deletion.

WHAT IS THE TESTING MATERIAL FOR THE NIPT?

NIPT is a genetic test and therefore a genetic material - DNA - is needed.
The material is a free circulating (cell-free) fetal (placental) DNA in maternal bloodstream.
Cell-free DNA is available in every individual by being released from the cells when they die at the end of a normal cell life cycle. Placental DNA enter the maternal bloodstream during pregnancy.

PREREQUISITIES FOR NIPT

A blood sample for NIPT can be collected at any of E. Gulbis Laboratory service centers. Sampling should be avoided on Fridays, Saturdays and Sundays.
An important factor for a valid result is fetal DNA fraction. Minimal fetal DNA fraction for precise interpretation of the results is 3.5%. In rare cases the fetal fraction is too low. Usually it rises as the pregnancy progresses but in some cases does not reach the needed percentage which might be due to the weight of the patient or other individual physiological characteristics. The fetal fraction itself is not an indicator of any chromosomal anomalies.

In the cases when the fetal fraction lower than 3.5% the laboratory is asking for an additional sample at least 15 days after the first sampling was done. In such cases the testing is repeated free of charge.
In very rare cases results can be inconclusive – in such cases an additional sample is also requested.

WHAT SHOULD I DO IF MY NIPT RESULT IS POSITIVE (HIGH RISK)?

In such cases your doctor should evaluate the results by also taking into consideration other test results (eg. US, 1st trimester screening, personal and family history) in order to determine the next steps.
NIPT results can only be confirmed by an invasive procedure – CVS or amniocentesis.