UZ LEUVEN | CENTRUM MENSELIJKE ERFELIJKHEID

NIPT-PLUZ: Non-Invasive Prenatal Test

Information for patients

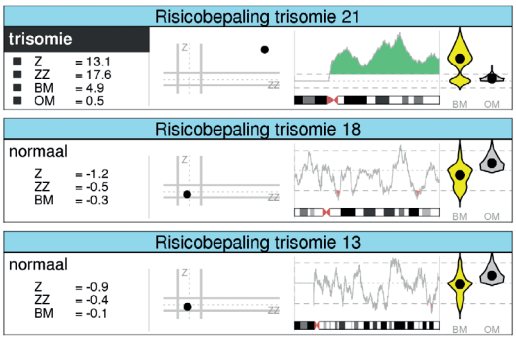
WHAT IS TRISOMY 21 OR DOWN SYNDROME?

A baby with trisomy 21, commonly known as Down syndrome, has three copies of chromosome 21 instead of the normal two copies. The main feature of trisomy 21 is intellectual disability, although in a minority of cases a congenital heart defect or other physical abnormality may be detected via ultrasound examination. The risk for trisomy 21 increases with the age of the mother, rising sharply from the age of 36.

Next to trisomy 21, trisomy 18 (Edwards syndrome) and trisomy 13 (Patau syndrome) are the viable chromosomal abnormalities. Therefore, for those who did not yet have a combined test, the NIPT (non-invasive prenatal test) is available to estimate the risk of a baby with trisomy 21, 18 or 13.

WHAT IS THE NON-INVASIVE PRENATAL TEST (NIPT)?

During pregnancy, DNA fragments of the baby can be found circulating in the blood of the mother. By measuring these DNA fragments of the baby in the maternal circulation, the sex of the baby is determined as well as the number of copies of chromosome 21, 18 and 13, allowing for detection of trisomy 21, 18 and 13. The blood collection for this test can be carried out as from 10 weeks of gestation, because from then on there is sufficient DNA of the baby present in the maternal blood.



Using the NIPT-PLUZ, performed in all genetics centers in Belgium including UZ Leuven, all chromosomes are analyzed. Therefore, in rare cases (~0,5% of all samples), NIPT can also detect other chromosome abnormalities, for example a trisomy of a chromosome other than 13, 18 or 21 or another chromosome abnormality that is clinically relevant for the mother's and/or the baby's health. The Belgian Advisory Committee on Bioethics recommends to also disclose these abnormalities to the pregnant woman (Opinion no. 66).

The NIPT performed within the Center of Human Genetics in Leuven is an in-house optimized and validated test. This unique analysis method was awarded the ISO 15189 quality label of the government (BELAC accreditation) and was published in various scientific journals.

As a genetic center, we also provide multidisciplinary expertise that guarantees the correct interpretation and follow-up of the NIPT in accordance with the national guidelines of the Belgian Society of Human Genetics (BeSHG) and the advice of the Belgian Advisory Committee on Bioethics.

NIPT is NOT able to detect:

- Mosaic trisomy 21, 18 or 13 (not all the cells have a trisomy)
- Small chromosome abnormalities (microdeletions or microduplications)
- Monogenic (single gene) disorders (such as cystic fibrosis or fragile X syndrome)
- Numerical abnormalities of the sex chromosomes (such as Turner syndrome or Klinefelter syndrome)

NIPT is NOT appropriate in any of the following situations:

- Ultrasound abnormalities in your baby (including a nuchal translucency of >3.5mm).
- The mother has had stem cell therapy or an organ transplant prior to the NIPT.
- The mother has cancer.

WHAT ARE THE POSSIBLE RESULTS OF THE NIPT?

1. A normal NIPT result means that no indication was found for the presence of trisomy 21, 18 or 13 in the baby. The test has an unprecedented sensitivity of almost 100% for the detection of trisomy 21, 18 and 13. Therefore, it is much more reliable than the combined test. As NIPT is a screening test and not a diagnostic test, a normal result cannot exclude trisomy 21, 18 or 13 for 100%.

2. An abnormal NIPT result indicates the presence of an abnormal number of chromosomes 21, 18 or 13. This result should always be confirmed by an invasive test (preferably by amniocentesis). By carrying out this diagnostic examination directly on baby's DNA, you will have complete certainty as to whether or not the baby has trisomy 21, 18 or 13.

   When the NIPT shows another clinically relevant chromosomal abnormality, the Centre for Human Genetics or your doctor will inform you accordingly and offer you the appropriate multidisciplinary follow-up.

3. The NIPT is inconclusive and can therefore neither exclude nor confirm the presence of trisomy 21, 18 or 13 in the baby. An inconclusive result occurs in less than 1% of the analyzed samples. In this case, further follow-up is recommended, either by a new NIPT analysis on a second blood sample (at no additional cost), or by an additional ultrasound followed by an invasive test (amniocentesis).

4. In less than 0,5% of the cases, the NIPT fails. If this is due to technical issues, the NIPT can be repeated once on a second blood sample (at no additional cost), or you can opt for another test.

The results can be consulted in your online medical file through "mynexuzhealth" (www.mynexuzhealth.be). In case of an abnormal result, your doctor or the Centre of Human Genetics will inform you.

If you have any further questions after reading this information, you may contact your gynaecologist or a genetics centre of your choice.

More information on the NIPT-PLUZ analysis as well as on the cost of the test, can be found on our website www.uzleuven.be/nipt.

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