Test results and actions

- Test results are available **within 7 calendar days** from reception of the blood sample in our laboratory.

- **Normal:**
  - ✓ No indication of fetal trisomy 21, 18 or 13
  - ✓ Standard pregnancy follow-up

- **Abnormal:**
  - ✓ Strong indication of fetal trisomy 21, 18 or 13
  - ✓ Confirmation by amniocentesis

- **Inconclusive result for chromosome 21, 18 or 13:**
  - ✓ Trisomy 21, 18 or 13 cannot be confirmed nor excluded
  - ✓ Repeat NIPT on new blood sample (no extra cost) OR ultrasound follow-up + amniocentesis

- **Failed test:**
  - ✓ No reliable analysis possible
  - ✓ Repeat NIPT on new blood sample (no extra cost)

Quality control

We appreciate your feedback!

Please update us with the outcome of the pregnancy after NIPT via *cme.nipt@uzleuven.be*

- Ultrasound abnormalities
- Spontaneous miscarriages
- Discrepant results (false positives/negatives)
- Invasive testing:
  - ✓ Tissue type: CVS / amniotic fluid
  - ✓ Type of analysis: array / FISH / qPCR
  - ✓ Result (as compared to NIPT)

Scientific publications

(1) Bayindir, B. *et al.* Noninvasive prenatal testing using a novel analysis pipeline to screen for all autosomal fetal aneuploidies improves pregnancy management. Eur J Hum Genet 23(10): 1286-93 (2015)

(2) Vandenberghe, P. *et al.* Non-invasive detection of genomic imbalances in Hodgkin/Reed-Sternberg cells in early and advanced stage Hodgkin's lymphoma by sequencing of circulating cell-free DNA: a technical proof-of-principle study. Lancet Haematol 2(2):e55-65 (2015)

(3) Amant, F. *et al.* Presymptomatic identification of cancers in pregnant women during noninvasive prenatal testing. JAMA Oncology 1(6): 814-9 (2015)

(4) Brady, P. *et al.* Clinical implementation of NIPT – technical and biological challenges. Clin Genet 89(5): 523-30 (2016)

(5) Brison, N. *et al.* Accuracy and clinical value of maternal incidental findings during noninvasive prenatal testing for fetal aneuploidies. Genet Med 113 (2016)

(6) Brison, N. *et al.* Predicting fetoplacental chromosomal mosaicism during non-invasive prenatal testing. *Manuscript submitted.* Prenat Diagn (2017)

(7) Neofytou, M. *et al.* Conflicting noninvasive prenatal and obstetric sex determination due to liver transplant. *Manuscript submitted.* Prenat Diagn (2017)

Disclaimer:

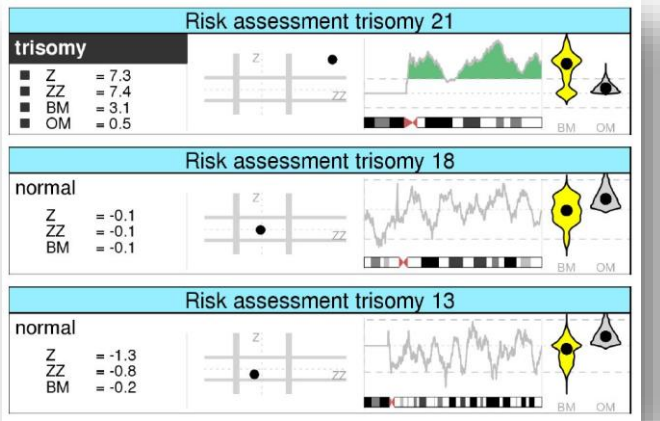
NIPT is a non-invasive prenatal screening test for detection of trisomy 21, 18 and 13 from 10 weeks of gestation onwards. An abnormal result should always be confirmed by invasive prenatal testing (preferably by amniocentesis). NIPT also detects the sex of the fetus. However, sex chromosomal aneuploidies cannot be detected. In rare cases, NIPT may detect other chromosomal abnormalities such as other fetal autosomal trisomies or a clinically relevant chromosomal abnormality in the pregnant mother. NIPT is not able to detect mosaicism, microdeletions, microduplications or monogenic disorders. When ultrasound abnormalities are present in the fetus, an invasive test is indicated. NIPT cannot be performed when the mother has had a stem cell transplant or an organ/tissue transplant.

Contact us at:
cme.nipt@uzleuven.be
+32 (0)16 34 59 03

More information:
www.uzleuven.be/nipt

NON-INVASIVE PRENATAL TESTING

NIPT



First centre in Belgium and in Europe to perform NIPT

In-house developed and optimized genome-wide analysis

Validated and accredited for the detection of trisomy 21, 18 and 13 as well as fetal sex

ISO 15189

Diagnostic Experience at UZ Leuven

November 2013 – July 1, 2017

~25,000 samples have been analyzed

>99% of cases received a result on first sampling

Test Performance

Unprecedented sensitivity of almost 100% for detection of fetal trisomy 21, 18 and 13 in singleton pregnancies

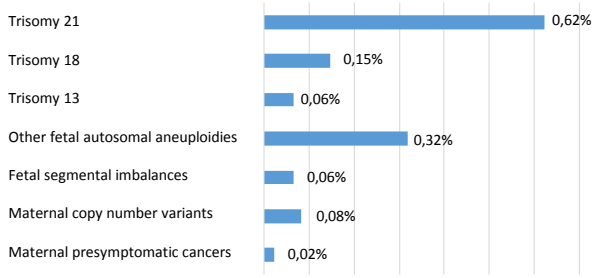
Much more reliable than the combined test

	Observed sensitivity	Observed specificity	PPV	NPV
Trisomy 21	99,44%°	99,98%	97,79%	100%
Trisomy 18	95%*	99,99%	92,68%	99,99%
Trisomy 13	100%	99,98%	78,95%	100%

° 1 false negative mosaic trisomy 21 (~75% of cells)
* 1 false negative trisomy 18 in 1 singleton and 1 early DCDA twin pregnancy

Incidental Findings

Genome-wide analysis allows detection of other clinically relevant maternal / fetal chromosomal abnormalities



IMPORTANT NOTICE:

NIPT at CME-UZ Leuven is offered in full compliance with the national guidelines for NIPT testing and management of incidental findings of the Belgian Society of Human Genetics (www.BeSHG.be) and the Belgian Advisory Committee on Bioethics (Opinion no. 66). CME-UZ Leuven is part of the national consortium which drafted and issued Good Practice Guidelines. The consortium is comprised of 8 genetic centres, all of which are nationally accredited by the Ministry of Health.

FAQs

When? ≥10 weeks of gestation

Blood collection?

- cfDNA tubes [Roche] (white cap) = **preferred**
- Streck tubes (camouflage pattern)

Contra-indications?

- Ultrasound abnormalities (incl. NT>3,5mm)
- Mothers who had a stem cell transplant or an organ/tissue transplant

Please indicate the following information on the request form for correct interpretation of the NIPT results:

- Mono- or dizygotic twin pregnancy
- Vanishing twin pregnancy
- Mothers with high pre-pregnancy weight (>100kg)
- Mothers on heparin therapy
- Mothers with lupus
- Mothers who have (had) cancer

Cost for the pregnant women?

- Pregnant women with a Belgian medical insurance: 8,68€ (as from July 1, 2017)
- Others: 260€

To change how to receive your patients NIPT results (by eHealth/KWS inbox/E-mail/fax/post):

Please send an E-mail to aflevervoorkeur@uzleuven.be