Test results and actions

- 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 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


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. When ultrasound abnormalities are present in the fetus, an invasive test is indicated. NIPT is not able to detect mosaicism, microdeletions, microduplications or monogenic disorders. NIPT should be performed with caution in cases of (vanishing) twin or multiple pregnancies, when the mother has (had) cancer, when the mother recently had heparin therapy or a blood transfusion, and when the mother has had immunotherapy, a stem cell transplant or an organ/tissue transplant within 3 months prior to the pregnancy.

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

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

More information:
www.uzleuven.be/nipt
Diagnostic Experience at UZ Leuven
November 2013 – Mid April 2017

Over 21,000 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

No false negatives for trisomy 21 and 13

False positive in <0.05% of cases

Incidental Findings

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

<table>
<thead>
<tr>
<th>Condition</th>
<th>Observed sensitivity</th>
<th>Observed specificity</th>
<th>PPV</th>
<th>NPV</th>
</tr>
</thead>
<tbody>
<tr>
<td>Trisomy 21</td>
<td>100%</td>
<td>99.99%</td>
<td>98.12%</td>
<td>100%</td>
</tr>
<tr>
<td>Trisomy 18</td>
<td>97.14%*</td>
<td>100%</td>
<td>97.14%</td>
<td>99.99%</td>
</tr>
<tr>
<td>Trisomy 13</td>
<td>100%</td>
<td>99.98%</td>
<td>64.29%</td>
<td>100%</td>
</tr>
</tbody>
</table>

* 1 false negative trisomy 18

IMPORTANT NOTICE:

NIPT at CME-UZ Leuven is offered in full compliance with the BeSHG National Guidelines for NIPT testing and management of incidental findings (www.BeSHG.be). 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 on heparin therapy or who had a blood transfusion
- Mothers who had immunotherapy, a stem cell transplant or an organ/tissue transplant within 3 months prior to the pregnancy
- Mothers with a chromosome abnormality
- Mothers with cancer

Twin pregnancies and high pre-pregnancy BMI?

NIPT can be performed in case of:

- Monozygotic / Dizygotic twin pregnancies
- Vanishing twin pregnancies
- High pre-pregnancy weight (>100kg)

Please provide this information on the request form for correct interpretation of the NIPT results.

Cost for the pregnant women?

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