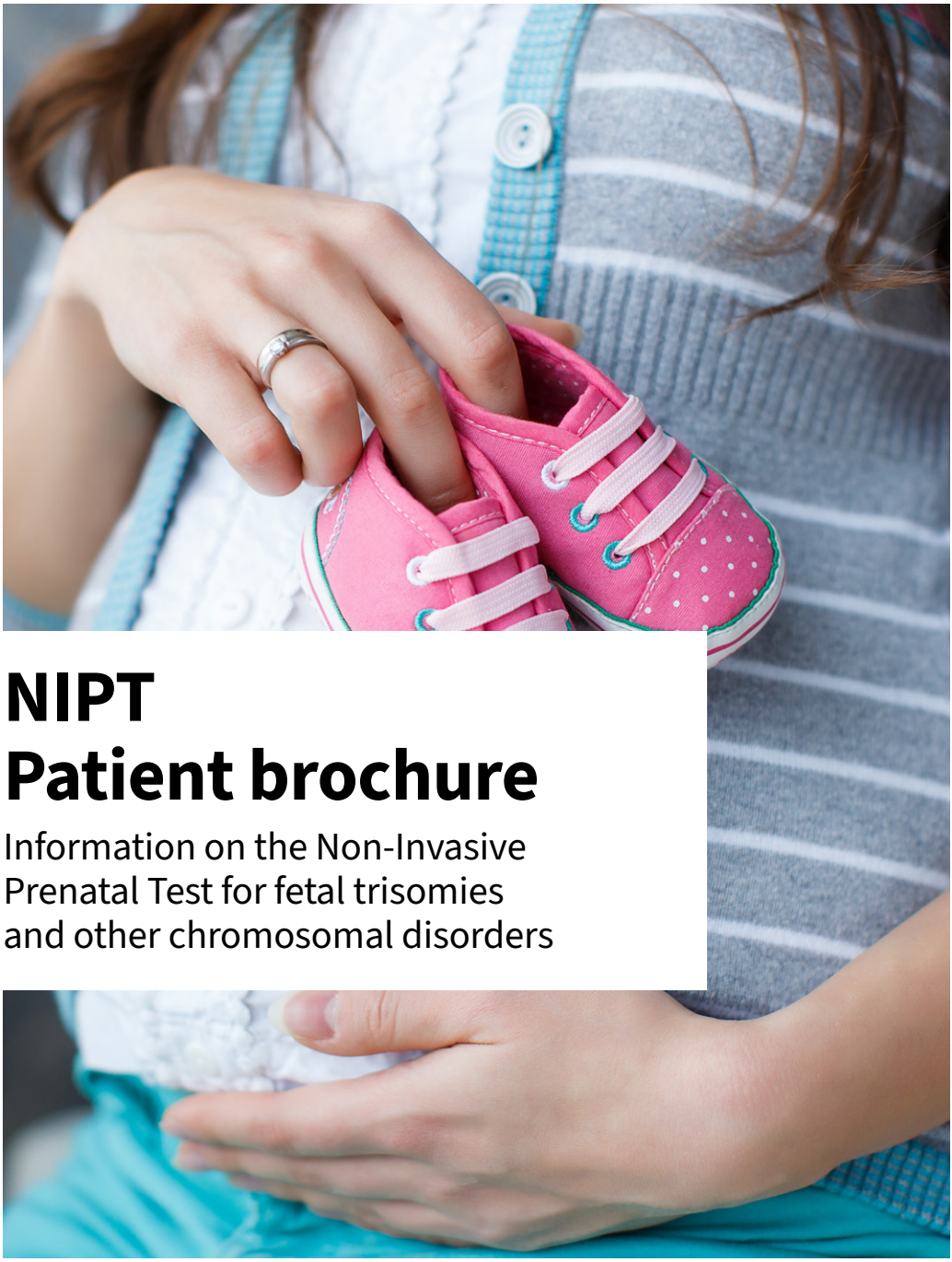
Universität Zürich[UZH]

Institute of Medical Genetics
Genetic Diagnostics Laboratory



NIPT
Patient brochure

Information on the Non-Invasive Prenatal Test for fetal trisomies and other chromosomal disorders

What can the NIPT analyses we offer detect?
The Institute of Medical Genetics at the University of Zurich offers a non-invasive prenatal test (NIPT), which is based on Illumina high-throughput sequencing technology. All tests are performed and analysed on-site in our Genetic Diagnostics Laboratory.
The tests we offer are the following :

- **"Basic Trisomy NIPT"** (Illumina VeriSeq® basic NIPT)
  NIPT for the exclusion of the common trisomies (13, 18, 21) and of the numerical abnormalities of the sex chromosomes X and Y (e.g. Turner or Klinefelter syndrome), as well as determination of the fetal sex, if desired.
- **"Simple Extension" to all chromosomes** (Illumina VeriSeq®-NIPT)
  All chromosomes (1-22, XY) are examined for trisomies and monosomies, as well as for large deletions and duplications (≥7Mb).
- **"Combination Package" for all chromosomal incl. microdeletions / microduplications** (Illumina VeriSeq®-NIPT and in house IMG-NIPT)
  All chromosomes (1-22, XY) are examined for trisomies and monosomies, for large deletions and duplications (≥7Mb), as well as for microdeletions / microduplications (≥3Mb). However, this test is only possible in presence of a fetal fraction of at least 6% and it is only carried out and charged for in this case.

When and for whom is the non-invasive prenatal test (NIPT) possible?
The test is possible from the 10th week of gestation (i.e. from 9+1 weeks). However, the likelihood of obtaining a reliable result increases at later gestational age, since more fetal DNA is usually detectable. Our NIPT tests can be used for both single and multiple gestation pregnancies and in case of egg donation.

How long do I have to wait for a result?
A result is usually available within 1-2 weeks. The findings are immediately sent to the requesting doctor by e-mail. Unless there is a medical indication for sex determination, the fetal sex may only be communicated after the 12th week of gestation.

How can fetal chromosomal disorders be detected in maternal blood?
DNA fragments originating from the placenta circulate in the mother's blood. In the vast majority of the cases, such fragments match the fetal genetic material and can thus provide indications of possible chromosomal disorders in the fetus.

What is the significance of the NIPT result?
A normal result in the **"Basic Trisomy NIPT"** means that there is no evidence of trisomy in the chromosomes tested. In singleton pregnancies the detection for trisomies 13, 18 and 21 is > 99.9%. In twin pregnancies, the detection rate may be slightly lower. The number of sex chromosomes is correctly detected by NIPT in over 90% in singleton pregnancies. In multiple gestation pregnancies the number of sex chromosomes is more difficult to be determined, in rare cases it is not possible.
The **"Simple Extension"** and the **"Combination Package"** have a detection rate for trisomies affecting other chromosomes of 96.4%. For other abnormalities (deletions / duplications), the detection rate and test accuracy depend on the size of the abnormality and on the amount of the circulating fetal DNA. This means that not all chromosomal abnormalities can be detected with the **"Simple Extension"**, nor with the **"Combination Package"**. Furthermore, the trisomy of all chromosomes simultaneously (triploidy) cannot be detected with any of the tests we offer, due to technical limitations.

Before drawing any conclusions from an abnormal NIPT result, it's essential to confirm it through a diagnostic test, by means of an invasive procedure (e.g. amniocentesis). This precaution is necessary because anomalies identified in NIPT could be confined to the placenta, a (vanishing) twin, or potentially be caused the presence of variants in the pregnant woman herself.

What other findings could be identified with NIPT?
In 1-2% of the cases, the tests can either provide an unclear result or a no result. In such cases, the test can be repeated once without additional costs.
Medically relevant chromosomal abnormalities affecting the pregnant woman can be detected with the **"Simple Extension"** and the **"Combination Package"**. In rare cases, such abnormalities can indicate for example the presence of presymptomatic cancer in the pregnant woman. If you do not wish to receive such information, you can object to this on the request form.

Is NIPT the right test for me?
When facing a significantly higher risk for fetal trisomy or when abnormal fetal ultrasound findings are present, choosing an invasive procedure (like chorionic villus sampling or amniocentesis) would yield faster results and detect more chromosomal abnormalities compared to NIPT. Additionally, depending on the indication, gene analyses can also be carried out from the chorionic villa or from the amniocentesis. The risk of miscarriage following invasive procedures is less than 0.5% (< 1:200) for experienced physicians.
If there is a hereditary disease or a disability affecting a member of your family, the genetic cause should first be clarified. This permits a targeted search in the foetus. For diagnostic clarification of yet unclear familial diseases or for general genetic counselling, our team of specialists will be happy to assist you in our outpatient clinic (invoices accordingly to TARMED). For appointments: sprechstunde@medgen.uzh.ch

What are the costs for the various tests and what is covered by the basic insurance?
The **"Basic Trisomy NIPT"** for chromosomes 13, 18, 21, X and Y, including determination of the fetal sex, costs CHF 459 plus CHF 21.60 order tax. If a first-trimester test showed a risk for trisomy of at least 1:1000 and if the NIPT is performed from the 12th week of gestation, the costs are usually reimbursed by your basic health insurance.
For an additional charge of CHF 250, a **"Simple Extension"*1** can be carried out (not covered by basic health insurance).
A **"Combination Package"*1** costs CHF 350 in addition to the "Basic Trisomy NIPT" (not covered by basic health insurance).

*1 The „Simple Extension" and the „Combination Package" can only be ordered in conjunction with the „Basic Trisomy NIPT". If a „Basic Trisomy NIPT" was performed externally and you would like the „Simple Extension" or the „Combination Package", a new basic trisomy NIPT is required in our laboratory.

Who can arrange a NIPT?
Specialists in gynaecology and obstetrics with a focus on feto-maternal medicine, specialists in medical genetics and physicians with a certificate of competence in pregnancy ultrasound from the SGUM. They can request special blood collection kits from us. If the request form has been completed in advance and signed by an authorized physician, blood can also be taken at our institute by prior appointment.

Where can I find more information?
You can find more information on genetic counselling and prenatal diagnostics at www.medgen.uzh.ch/sprechstunde.

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Editor
Institute of Medical Genetics

Director
Prof. Dr. med. Anita Rauch
FMH Medical Genetics
FAMH Medical Genetics

Head of department
PD Dr. phil. biochem.
Beatrice Oneda
FAMH Medical Genetics

Print
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Address
University of Zurich
Institute of Medical Genetics
Wagistrasse 12
8952 Schlieren
Switzerland
Tel. + 41 44 556 33 00
sprechstunde@medgen.uzh.ch

Website
www.medgen.uzh.ch