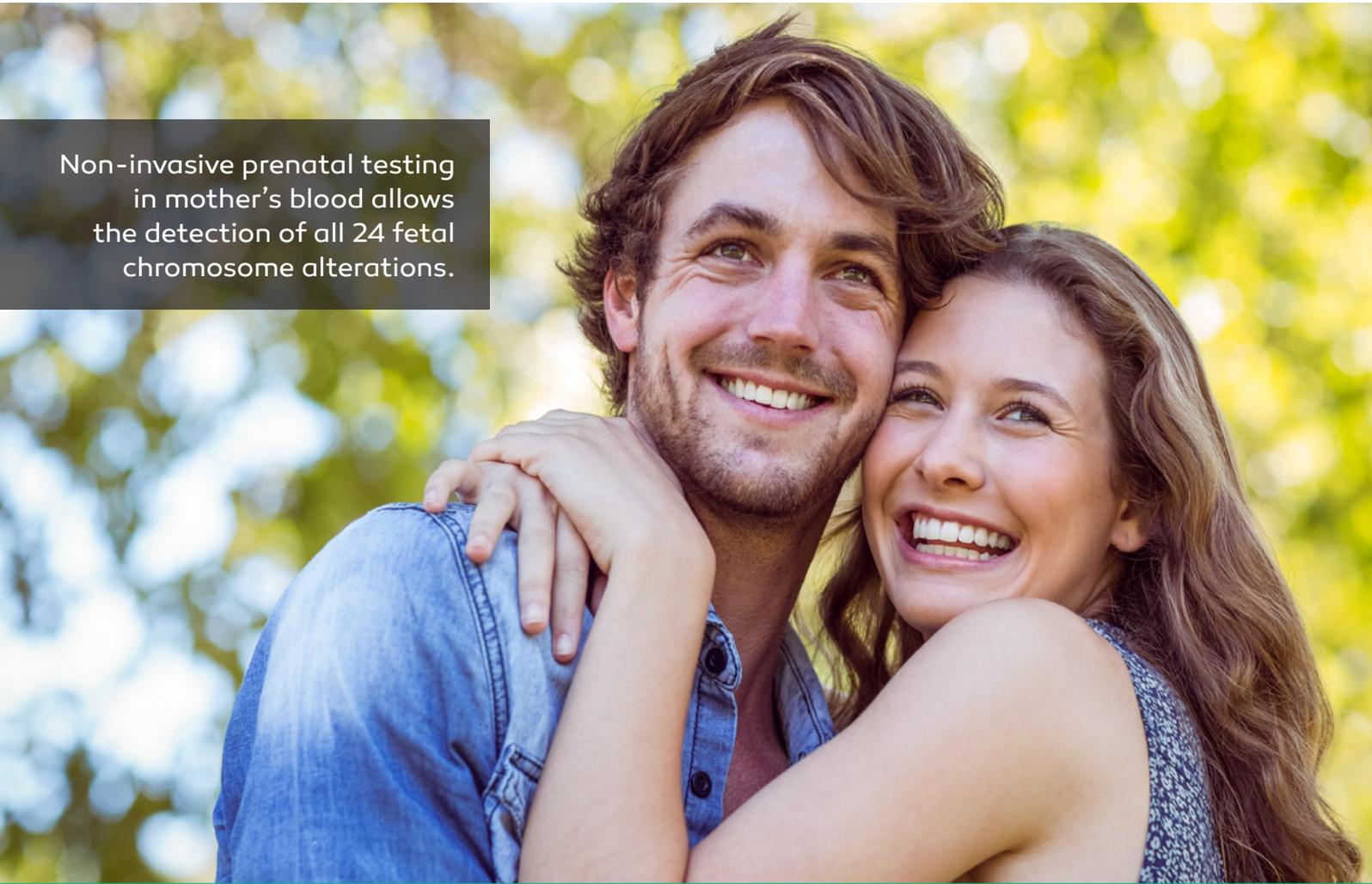


# NIPT

## NON INVASIVE PRENATAL TEST



Non-invasive prenatal testing in mother's blood allows the detection of all 24 fetal chromosome alterations.

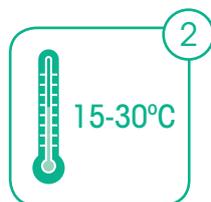


During pregnancy, small amounts of fetal cell-free DNA that pass through the placenta can be detected in the mother's blood. The non-invasive prenatal test (NIPT) screens this fetal DNA to identify the presence of alterations in the chromosomes of the baby. NIPT is the most reliable and safe prenatal screening option currently available, with no associated risks for either the mother or the fetus. From a single tube of mother's blood, drawn as early as 10 weeks in the pregnancy, this test allows to determine if the future baby presents any chromosome alteration.

## METHODOLOGY



1  
Mother's blood draw (10ml) at 10 weeks +



2  
Keep the blood sample at room temperature until shipping



3  
Ship at room temperature



4  
Cell-free fetal DNA analysis



5  
Results in 7 working days

## What is Non-Invasive Prenatal Testing?

The non-invasive prenatal test (NIPT) in mother's blood is a new prenatal screen test that allows to identify the presence of fetal chromosome anomalies. This test represents a great advance in prenatal screening.

By carrying out a simple blood test from week 10 of pregnancy, remnants of fetal genetic material that pass through the placenta can be detected in the mother's blood. From the study of this fetal material, it is possible to foresee the possibility that the future baby suffers from any chromosome alteration.

## How is NIPT performed?

The non-invasive prenatal test requires 10 ml blood sample. Once drawn, it must be sent at room temperature to our laboratory where an extraction of both maternal and fetal genetic material will be performed. This material will be then analyzed using state-of-the-art Next Generation Sequencing (NGS) technology.

From the analysis of the sequencing results, the amount of fetal and maternal DNA present for each of the chromosomes analyzed will be determined. And thus the probability that the fetus suffers from aneuploidies or microdeletions can be elucidated.

Our results reports are available in 7 days after sample reception.



## Who can benefit from NIPT?

Medical societies have recommended NIPT as an option for all pregnant women regardless of age or risk<sup>1,2</sup>. This screening test is aimed at patients with gestation of 10 weeks or more with single or twin pregnancies. It is particularly beneficial for woman of advanced maternal age ( $\geq 35$  years), who have had an abnormal result in the combined screening of the first trimester, an abnormal ultrasound or a medical history that suggests an increased risk of a pregnancy with chromosome aneuploidies.

## How reliable are NIPT results?

Non-Invasive Prenatal Testing results are the most precise prenatal screening tests results currently available, more precise than the traditional combined screening of the first trimester<sup>3</sup>. In general, the probability of obtaining a false positive or false negative result is lower than in other tests<sup>1-3</sup>.

NIPT detects the main trisomies: Down syndrome, Patau syndrome and Edwards syndrome, with a sensitivity and specificity higher than 98% and 99% respectively. This test is also useful to detect aneuploidies in the sex chromosomes with sensitivity and specificity greater than 95% and 99% respectively<sup>1-3</sup>.

This test also detects, with high specificity and sensibility the presence of microdeletions associated to the following syndromes: Angelman, Prader-Willi, 1p36 deletion, Cri du Chat, Wolf-Hirschhorn, Jacobsen, Langer-Giedion, DiGeorge II, Phelan-McDermid and 16p11.2-p12.2 deletion.

## What are the advantages iGLS NIPT service?

iGLS uses non-invasive prenatal test to determinate, by means of massive sequencing of the entire genome, the presence of fetal aneuploidies in all 24 chromosomes and the most common microdeletions. This test has significantly higher detection rates than traditional methods<sup>1-4</sup> and has shown excellent detection rates and very low false positive rates compared to other non-invasive prenatal diagnosis methods<sup>5</sup>.

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2. Gregg AR et al. Genet Med. 2016;18(10):1056-1065.
3. Bianchi DW et al. 2014;370(9):799-808.
4. Norton ME et al. N Engl J Med. 2015;372(17):1589-1597.
5. Gil MM et al. Ultrasound Obstet Gynecol. 2015;45(3):249-266.

**iGLS**

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