Ninalia
by eurofins biomnis

Non-Invasive Prenatal Testing for trisomies 21, 18 and 13 (NIPT)

The prenatal trisomy screening test based on a simple maternal blood sample

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What is the Ninalia screening test?

Ninalia Non-invasive Prenatal Testing (NIPT) is an innovative genetic screening test for chromosomal abnormalities.
This test uses circulating cell-free DNA (cfDNA) from a maternal blood sample without risk to the fetus.
Ninalia detects the main fœtal aneuploidies i.e. trisomy 21, 18 and 13. They're the only complete and homogenous trisomies found at birth outside of the dysgonosomias.
This test uses next-generation sequencing (NGS) and is performed after the 1st trimester ultrasound.

NEW

Today, this test also makes it possible to screen for other rarer chromosome abnormalities : autosomal abnormalities, deletions and duplications greater than 7 megabases (karyotype-equivalent resolution)

Several test options are available:

- **Ninalia 3** (NIPT) : Trisomies 21, 18 et 13
- **Ninalia 5** (NIPT) : Trisomies 21, 18 et 13
  + fetal gender determination
  + sex chromosomes aneuploidies
- **Ninalia Genomewide** (NIPT) : all autosomes
  + chromosomal imbalance greater than 7Mb Ninalia
- **Genomewide Complete** (NIPT) : all autosomes
  + chromosomal imbalance greater than 7Mb
  + sex chromosomes aneuploidies
  + fetal gender determination

What are the benefits for the patient and her physician?

Autosomal aneuploidies can, for most chromosomes, be found in the form of mosaics. Thus, when an anomaly is observed, the healthcare professional will refer, depending on the anomaly found, to ultrasound monitoring, and/or monitoring of the risk of pre-eclampsia, and/or possibly towards an invasive procedure to study the karyotype.

The result of the test can thus, by directing the monitoring of the pregnancy, make it possible to avoid a fetal loss in late pregnancy.

When an invasive procedure is warranted, knowledge of the suspected abnormality helps the healthcare professional specify the confirmatory techniques to be used (karyotype, FISH, DNA chip) and the type of tissue to be studied (chorionic villi or amniotic fluid).

Finally, a structural imbalance of more than 7 megabases, proven in the fetus, may not necessarily be accompanied by ultrasound call signs at the end of pregnancy but may be associated with clinical signs at birth, or at a later date.

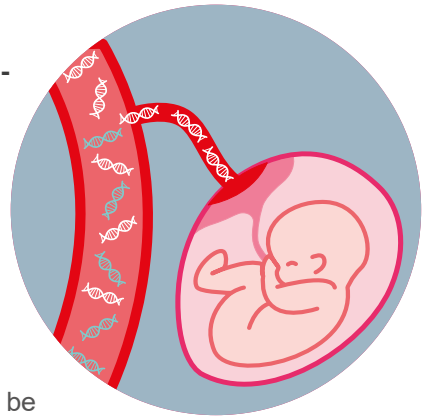
What is cell-free DNA?

During pregnancy, the placenta releases **cell-free DNA** which circulates in the maternal blood stream.

As a result, maternal blood contains a mix of fœtal and maternal DNA in varying proportion. This mix is the **fœtal fraction**

Our laboratory isolates this circulating DNA in order to detect the presence of genetic material.

If the fetal fraction is too low, a new sample may be required for analysis.



Cell-free maternal DNA
Cell-free fœtal DNA

What is the technology used?

Since october 2014, Eurofins Biomnis has chosen to collaborate with the company Illumina, a world leader in DNA sequencing, for non-invasive screening for the main chromosomal abnormalities.

The new VeriSeq method provided by Illumina makes it possible **to extend screening to all autosomes** as well as **deletions and duplications** greater than 7 megabases, for an even more comprehensive management of pregnancy.

Performance of Ninalia?

- Following the change of VeriSeq method, the performance of the test for viable trisomies at birth are improved to a **excellent detection rate.**

Singleton pregnancy	Trisomy 21	Trisomy 18	Trisomy 13
Sensitivity	>99.9%	>99.9%	>99.9%
Specificity	99.9%	99.9%	99.9%

Twin pregnancy	Trisomy 21	Trisomy 18	Trisomy 13
Sensitivity	96.4%	95.7%	93.6%
Specificity	99.9%	>99.9%	>99.9%

Detection of autosomal aneuploidy	
Sensitivity	96.4%
Specificity	99.8%

Detection of chromosomal imbalance greater than 7 megabases	
Sensitivity	74.1%
Specificity	99.8%

- **Fetal sex classification** *(Illumina study of concordance of the results of the sex chromosomes and the standard clinical assessment of 1963 cases):* concordance for fetal sex classification = 100%.

  The presence of a vanishing twin may explain a false positive result for the Y chromosome.

  For multiple pregnancies, the presence of the Y chromosome does not allow to define the number of male fetuses.

Percentage concordance for sex chromosome abnormalities			
XO	90.5%	XXY	100%
XXX	100%	XYY	91.7%

Source: date from the supplier, July 2019

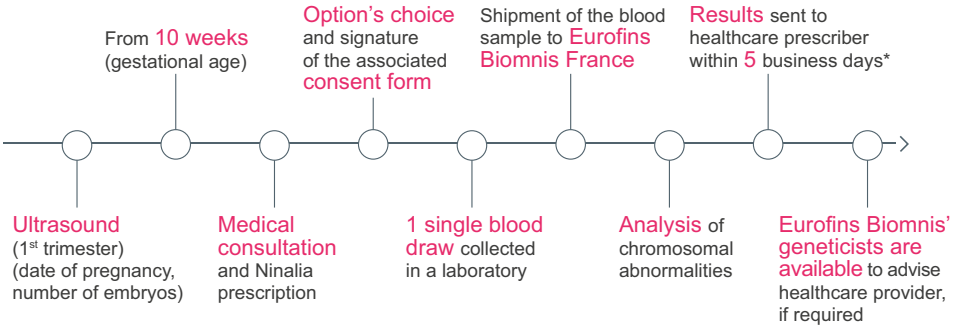
In practice

- A short turnaround time: **5 business days** (upon reception at Eurofins Biomnis)
- A **CE IVD marked** solution

Who should avail of the Ninalia screening test ?

The Ninalia test can be offered to any pregnant woman after the 1st trimester ultrasound and at a genetic counselling consultation.

Step by step guide to Ninalia



** For example, if the sample is received in Eurofins Biomnis on a Tuesday, the results will be sent to the healthcare provider the following Monday*

If you have any questions, we recommend that you contact the international division of Eurofins Biomnis: **international@eurofins-biomnis.com**

About Eurofins Biomnis

Eurofins Biomnis, **European leader in specialised medical pathology**, has its origins in the laboratory established by Marcel Mérieux in 1897. Its primary focus is the performance of highly specialised analyses requiring the use of **high-expertise test procedures.**

Eurofins Biomnis is active in all fields of medical pathology, in particular **fœtal biochemistry and prenatal diagnostics.**

Geneticists, qualified in prenatal diagnosis and responsible for Eurofins Biomnis tests are available to prescribers to provide the best possible support in the implementation of this genetic test.

To know more about Ninalia, please visit:
www.nipt-biomnis.com

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Ninalia: at a glance

- NIPT stands for **Non-Invasive Prenatal Testing**
- Ninalia NIPT is a screening test for pregnant women at risk of **trisomy 21, 18 and 13,** dysgonosomias and fetal sex

NEW

- This test also makes it possible to screen for other rarer chromosome abnormalities : autosomal abnormalities, deletions and duplications greater than 7 megabases

- It is **without risk to the fœtus** since this test requires a simple maternal blood draw collected by healthcare providers
- Ninalia can be performed **from 10 weeks** (gestational age)
- **Results are sent to healthcare providers within 5 business days upon reception at Eurofins Biomnis**
- Highly **accurate test** with a detection rate of 99.9% and an extremely low failure rate
- Over 100,000 Ninalia tests have been performed to date for trisomies 21, 18 and 13
- Ninalia is **NF EN ISO 15189** accredited*
- The assurance of **Eurofins Biomnis expertise.** The laboratory is based in France and is the **European leader in specialised medical pathology.**

**Accreditation delivered by the Cofrac (N#8-1973 & N#8-1100, available on www.cofrac.fr)*

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