



First trimester screening detects common chromosomal abnormalities in the fetus during pregnancy. This type of screening primarily focuses on the detection of Down syndrome (T21) and Edwards syndrome (T18), with a sensitivity of 85-90% and a false positive rate of 5%.

Technological improvement in DNA analysis has led to the development of non-invasive prenatal tests (NIPT) based on the study of the cell-free fetal (placental) DNA in maternal blood, enabling a more accurate screening of chromosomal conditions with greater sensitivity and specificity, and no risk for the mother or the fetus.

neoBona

neoBona is the new generation of non-invasive prenatal test resulting from the combined expertise of SYNLAB, one of the European leaders in prenatal diagnosis, and Illumina, a world leader in next-generation DNA sequencing and analysis.



SYNLAB has pioneered molecular prenatal diagnosis by incorporating new scientific advances to offer a state-of-the-art non-invasive prenatal test, backed by the expertise and reliability of one of Europe's leading laboratories.

neoBona determines the amount of cell-free fetal DNA present in the maternal blood (fetal fraction), and uniquely includes the analysis of cell-free DNA fragment size, for the highest accuracy.

Scientific background of neoBona

The innovative paired-end sequencing technology of **neoBona** allows the accurate quantification of the fetal fraction and the distinction of maternal and fetal DNA according to the fragment sizes.

The innovative algorithm generates the TSCORE (Trisomy Score) by combining the depth of sequencing, the percentage of cell-free fetal DNA and the quantification on short (fetal) fragments, to obtain reliable results even at very low fetal fractions.

Chromosomal alterations detected

neoBona analyses a blood sample from the mother to determine the risk of chromosomal abnormalities in the fetus during pregnancy. There are three options available in order to adapt the prenatal screening to each patient. **neoBona** and **neoBona Advanced** are performed in SYNLAB European laboratories.

Option available for singleton and twin pregnancies:

- **neoBona:**
 - ✓ Trisomies 21, 18 and 13 + Fetal sex (optional)
 - ✓ Paired-end NGS technology
 - ✓ Fetal fraction reported for both options

In case of twin pregnancies, if the "Fetal sex" option is selected the presence of chromosome Y is determined. If it is detected, at least one of the fetuses is male.

Option available for singleton pregnancies:

- **neoBona Advanced:**
 - ✓ Trisomies 21, 18 and 13 + Fetal sex + Aneuploidies X, Y
 - ✓ Paired-end NGS technology
 - ✓ Fetal fraction
- **neoBona Advanced+:**
 - ✓ Trisomies 21, 18 and 13 + Fetal sex + Aneuploidies X, Y
 - ✓ Trisomies 16 and 9 + Microdeletions panel: DiGeorge, Angelman, Prader-Willi, deletion 1p36, Wolf-Hirschhorn and Cri-du-chat syndromes
 - ✓ Conventional single-read NGS technology
 - ✓ Fetal fraction

neoBona detects aneuploidies and fetal sex, with the confidence of working with a European leader in prenatal diagnosis.

Advantages:

- ✓ Latest-generation sequencing technology with paired-end reads for increased accuracy
- ✓ Determination of fragment size distribution
- ✓ Determination of fetal fraction
- ✓ Innovative TSCORE for unmatched accuracy at low fetal fraction
- ✓ Expertise of one of the European leaders in prenatal diagnosis
- ✓ Largest European network of blood sampling centres
- ✓ Expert consultants available for the doctors

Indications

Pregnant women with at least **10 weeks gestational age** ($\geq 10+0/7$), in the following situations:

- Single or twin (two fetuses) pregnancy
- IVF pregnancy
- Gametes donation
- Vanishing twin

neoBona is a genetic screening test and as such, must be prescribed by a specialist after appropriate counselling.

Requirements

Sample: Dedicated kit provided by the laboratory. Store and send at room temperature.

Documentation: Dedicated test requisition and informed consent form.