INDICATIONS

The non-invasive genetic test for fetal trisomies 13,18,21 is mainly intended for women with an increased risk of fetal trisomy 21 without ultrasound scan abnormalities:

1. Maternal age > 35 years old;
2. Maternal serum markers indicating a risk of trisomy 21 ≥ 250 WITHOUT increased nuchal translucency;
3. Patient with prior medical history of fetal aneuploidy;
4. Presence of a robertsonian translocation involving a chromosome 13 or 21 in one of the parents;
5. Serum markers out of truncation limits, thus underestimating a risk for trisomy 21 initially between 1/250 and 1/500.

This test is indicated as well in case of twin pregnancies and in case of biological risk of trisomy 13 or 18. It can also be performed in the absence of any increased risk of aneuploidy.

RESULTS

The usual turnaround time ranges from 5 to 15 working days at reception of the specimen at Laboratoire Cerba.

Performing this test, regardless its result, does not replace in any case the **follow-up of the pregnancy by ultrasound scan**.

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COFRAC NF EN ISO 15189 ACCREDITATION

N° 8-0945, medical testing
Scope available at www.cofrac.fr

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YOUR HEALTHCARE PROFESSIONAL

INFORMATIONS FOR HEALTHCARE PROFESSIONALS

Cerba

Non invasive prenatal testing for fetal trisomy 21

www.lab-cerba.com

A TEST ON FETAL DNA CIRCULATING IN THE MATERNAL BLOOD

Testing of the fetal DNA circulating in the maternal blood is used since many years in the follow-up of pregnancies, especially for fetal sex determination in the context of genetic diseases linked to chromosome X and for fetal Rhesus D determination in case of feto-maternal incompatibilities.

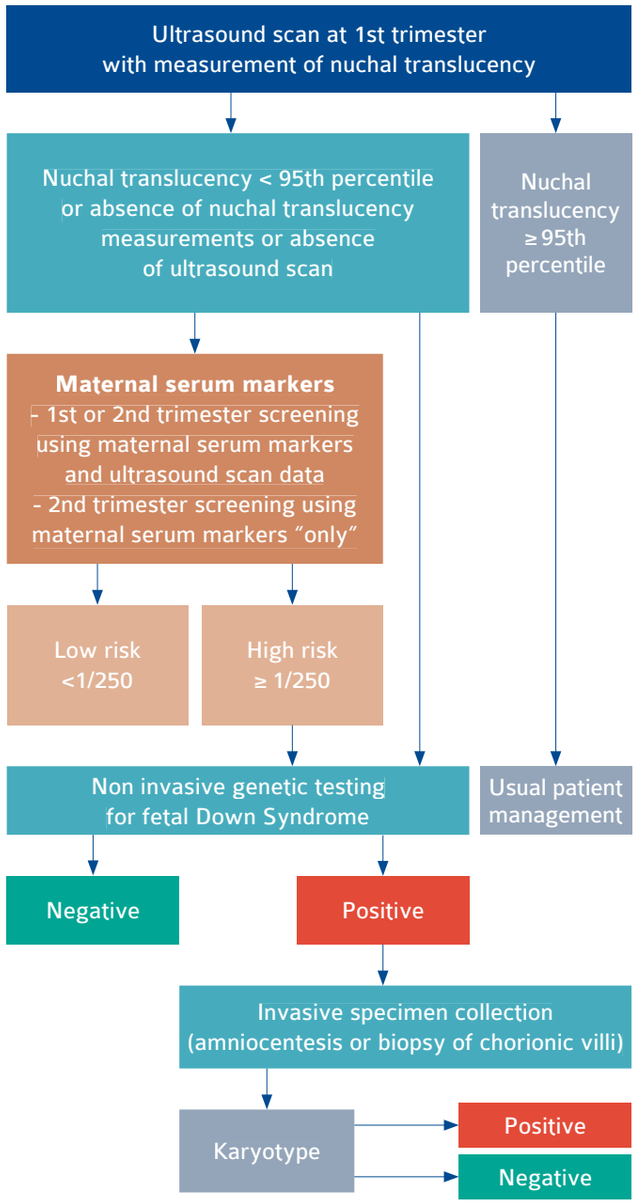
The non invasive prenatal testing for fetal trisomy 21 and other aneuploidies is a new non invasive prenatal test performed on a simple blood sampling from the mother. It enables, as early as the 10th week of gestation in pregnant women, to determine if the fetus carries a trisomy 21 or another aneuploidy of type 13 or 18.

Its sensitivity and specificity are greater than 99% for trisomies 13, 18 and 21.

When applied to the management of patients with increased risk of trisomy 21 without signs at ultrasound scan, it dramatically reduces (around 95%) the number of invasive procedures (amniocentesis or biopsy of chorionic villi) resulting into iatrogenic miscarriages in around 1% of cases.

In case of positive result of the non invasive genetic test for Down Syndrome (or trisomy 13 or 18), a fetal karyotype for confirmation will be performed.

STRATEGY FOR DIAGNOSIS OF DOWN SYNDROME



NATURE AND PRINCIPE OF THE TEST

The non invasive prenatal testing for trisomy 21 and other aneuploidies consists in the analysis of DNA fragments from the fetus(es), those DNA fragments being present in the maternal blood during pregnancy.

Although it is a genetic test, the aim is not to analyze the genome of the fetus, but only to evaluate the relative proportion of each of the chromosomes 13, 18 and 21 in order to detect an excess of 13, 18 or 21 genetic material when the fetus carries a trisomy 13, 18 or 21.

In practice, this over-representation is tiny, considering that the fetal DNA represents only 10% of the DNA present in the maternal blood.

This test thus requires a powerful analysis method, the Next Generation Sequencing (NGS) combined with a high performance computing environnement (bio-informatic pipeline) for quickly analyzing several millions of molecules of DNA, attribute them to their original chromosome, measure their proportion and assess if there is or not an over-representation that is statistically significant. This is what performs the non invasive genetic test for Down Syndrome and other fetal aneuploidies.