

Non-Invasive Prenatal Testing (NIPT) for trisomies 21, 18, 13 from week 10 for singleton and twin pregnancies plus X and Y chromosome analysis for singleton pregnancies (all now available for singleton IVF self and non-self donors)

Overview: Non-invasive prenatal testing (NIPT) analyzes cell-free DNA from the fetus circulating in the pregnant mother's blood. This is a new additional prenatal screening test for Down syndrome (trisomy 21) and two other common fetal chromosomal abnormalities (trisomies 18 and 13). Also, testing for X and Y chromosomes is possible upon request (not for twin pregnancies).

Background: DNA from the fetus can be found circulating in the mother's blood. The fetal cells are naturally broken down, resulting in the fetal DNA circulating freely in the mother's blood until several hours after giving birth. This cell-free DNA (cfDNA) can be analyzed to estimate the risk of the fetus having trisomies 21, 18, or 13.

Benefit: Since it is non-invasive there are **no risks to the fetus** as with invasive procedures (amniocentesis, CVS).

Material: Blood sample from the mother (2 x 10 ml whole blood in special tubes)

Results: A 'HIGH RISK' result indicates a high risk for a trisomy, it does not mean that the fetus definitely has a trisomy. However, following up with invasive procedures is recommended in this case. The test identifies more than 99% of fetuses with trisomy 21, 98% of fetuses with trisomy 18, and 80% of fetuses with trisomy 13.

Important note: A 'LOW RISK' result means it is unlikely that the fetus has a trisomy, but it cannot be excluded. Therefore, all results should be interpreted in the context of clinical and familial data and patients are recommended to continue with their first trimester screening/scan.

Who can do the test: Singleton and twin pregnancies, live fetus, WOG > 10 weeks (ultrasound)

X/Y analysis (not possible for twins): Extra, or incomplete copies of one of the sex chromosomes can be detected (XXX, XYY, XXYY, XXYY and a missing X chromosome in a girl).

Preanalytics: Please send your patient to the lab for blood extraction or contact FML for the

special tubes, samples must be shipped on day of blood extraction!

Important note: We can accept samples Saturday-Wednesday before 4pm only.

Turnaround time: 2 weeks

In case of any questions please contact FREIBURG MEDICAL LABORATORY www.fml-dubai.com $\,$