Maternal Blood Screening-DNA based Screening (NIPT)

This screen provides assessment of fetal risk for specific chromosome abnormalities using **maternal blood only**.

- **How the test works:** Fragments of placental DNA in the maternal blood are identified and analyzed. This screen can be done anytime in pregnancy after 9 to 10 weeks gestation. It is a single blood draw. On occasion, a repeat specimen is required to complete the test.

- **How good is the test:** This screen provides the highest detection rates with >99% detection for Down syndrome and the lowest false positive rate (0.1%). It also evaluates chromosomes 13, 18, X and Y.

Condition	Detection Rate	False Positive Rate
Down Syndrome	>99%	0.1%
Trisomy 18	>97%	0.1%
Trisomy 13	Variable due to rarity	
Gender	99.4% accuracy	0.6% error

Take-home messages

- All "non-invasive" tests are considered "screening" tests. Screening tests increase or decrease your risk. They cannot diagnose or exclude the possibility of a chromosomal abnormality.
- Diagnostic testing is available to all pregnant women regardless of age or family history.
- It offers information on all 23 pairs of chromosomes (99.9% detection rate) and allows us to evaluate for smaller defects depending on risk factors. Again, LOW RISK DOES NOT MEAN ZERO RISK.
- The two available diagnostic tests are Chorionic Villus Sampling (CVS) and Amniocentesis.

Chorionic Villus Sampling

- Placental cells are sampled through the abdomen or cervix depending on the location of your placenta.
- This can be done between 11 to 14 weeks providing the earliest diagnostic testing.
- Risk of miscarriage about 1/100 (1%) although has been reported as low as 1/300 (0.3%).

Amniocentesis

- Amniotic fluid containing fetal cells is sampled through the abdomen. This can be done after 15 weeks.
- Risk of miscarriage about 1/300 (0.3%) although has been reported as low as 1:1600 (0.06%).

Take-home messages

- All pregnancies are at risk of having a baby with a chromosomal abnormality. Young age and negative family history do not exclude the possibility.
- "Screening" provides us a way to evaluate your individual risk of having a baby with a chromosomal abnormality.
- Screening is not diagnostic. It only increases or decreases your risk for the condition(s) being screened.
- Not all babies with a positive screen are affected. Confirmation of all positive results recommended to exclude the possibility of a false positive.
- False negatives may also occur with screening. Some babies with low risk screens may have a chromosomal abnormality.
- Diagnostic testing is available to everyone. However, it carries a small risk of miscarriage.
- All prenatal testing discussed is considered optional. You may decline any or all testing.

* Detection of Chromosome Abnormalities during Pregnancy - Source: Boston MFM