MFM & GENETICS
OF ADVANTIA

PATIENT INFORMATION

NON-INVASIVE PRENATAL TESTING (NIPT)

What is NIPT?

Non-Invasive Prenatal Testing (NIPT) is a screening test that analyzes fragments of DNA from your baby's placenta that are present in your blood. This test can provide a risk of certain chromosomal conditions as early as 9-10 weeks into pregnancy.

What conditions can NIPT screen for?

NIPT primarily screens for:

- **Trisomy 21 (Down syndrome):** Extra copy of chromosome 21.
- **Trisomy 18 (Edwards syndrome):** Extra copy of chromosome 18.
- **Trisomy 13 (Patau syndrome):** Extra copy of chromosome 13.
- **Sex chromosome conditions:** Such as Turner syndrome (missing an X chromosome) or Klinefelter syndrome (extra X chromosome in a male).

Some labs also offer screening for*:

- Microdeletions (small missing pieces of chromosomes).
- Other rare genetic conditions.

*these additional tests are not recommended to be routinely performed on every pregnancy outside of additional risk factors - genetic counseling is recommended if additional testing is desired

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Who should consider NIPT?

Your Doctor may recommend NIPT for:

- Anyone seeking additional information about their baby's genetic health.
- Women aged 35 or older at the time of delivery.
- Those with a history of a pregnancy with chromosomal abnormalities.
- Women with abnormal findings on ultrasound.
- Families with a known genetic condition.

What are the benefits of NIPT?

- **Non-invasive:** Requires only a blood sample, posing no risk to the baby.
- **Early information:** Can be performed as early as 9-10 weeks into pregnancy.
- **High detection:** 90–99% sensitivity for common conditions like Down syndrome.

Limitations of NIPT

- **Screening test, not diagnostic:** NIPT identifies a high or low risk but cannot confirm or exclude a condition.
- **False positives and negatives:** Results may not reflect the baby's DNA.
- **Does not detect all chromosome or genetic conditions:** A risk for specific chromosomal abnormalities is provided.

What happens if NIPT results show a high risk?

If NIPT indicates an increased risk for a condition, genetic counseling and diagnostic tests may be recommended to confirm the result.

Diagnostic Testing Options

1. **Chorionic Villus Sampling (CVS):**
    - Performed most commonly between 12–13 weeks.
    - Involves collecting placental cells.
    - Can diagnose chromosomal and genetic conditions.
2. **Amniocentesis:**
    - Performed at 16 weeks and beyond.
    - Involves taking a sample of amniotic fluid.
    - Can diagnose chromosomal and genetic conditions, as well as neural tube defects.

Both tests are highly accurate but involve a small risk of miscarriage.