Should I have NIPT?

The decision to have NIPT is a personal one based on your beliefs and concerns. We hope this brochure has helped you to decide whether this option is for you. If you have additional questions please feel free to contact a genetic counselor at Desert Perinatal Associates.

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A helpful guide to Non-Invasive Prenatal Testing (NIPT)

Recent advances in the field of genetics have improved the screening options available to pregnant women. With a simple blood draw, these new screening tests are able to accurately detect certain chromosome disorders affecting the baby during pregnancy.

Chromosomes are the large structures in a cell that store DNA. DNA is the genetic material that controls the growth and development of the cell. Typically, we have 23 pairs of chromosomes. If a pregnancy has an extra copy of a chromosome or is missing a chromosome this can cause miscarriage and/or birth defects.

Non-invasive Prenatal Testing (NIPT) is a blood test that looks at small pieces of fetal DNA found in a pregnant woman's blood. These small pieces of DNA are naturally released from the cells of the placenta during pregnancy. By looking at the amount of this DNA in a woman's blood sample we are able to learn more about possible chromosome disorders affecting the pregnancy.

If there is too much DNA in the maternal blood sample it could mean the baby has an extra chromosome. If there is the correct amount of DNA in the maternal blood sample the chance for a chromosome disorder in the baby is low.

NIPT can be drawn any time after 10 weeks gestation. This test allows for the detection of pregnancies with certain chromosome disorders, specifically Down syndrome, Trisomy 18, and Trisomy 13. The test can also look for Turner syndrome and other sex chromosome abnormalities.

Down syndrome

Down syndrome occurs when a baby has an extra copy of chromosome number 21. Individuals with Down syndrome have mild to severe intellectual disability, characteristic facial features, and low muscle tone. They are also at an increased risk for heart defects. Approximately 1 in 800 babies are born with Down syndrome. Women of any age can have a child with Down syndrome. However, mothers age 35 or older are at a greater risk of having a baby with Down syndrome.

Trisomy 18 and Trisomy 13

Trisomy 18 and trisomy 13 are also caused by extra chromosomes. Like Down syndrome, the chance of having a baby with either trisomy 18 or trisomy 13 increases in mothers over age 35. Babies born with trisomy 18 or trisomy 13 have severe mental retardation. They may also have heart and brain defects. The majority of these babies (90%) die within one year after birth. Trisomy 18 and trisomy 13 occur in about 1 in 6,000 to 1 in 8,000 births.

Turner syndrome

Turner syndrome occurs when there is only one X chromosome and the second sex chromosome is missing. Individuals with Turner syndrome are always female. These girls have short stature, a short neck, low hair line and a wide chest. Many girls with Turner syndrome will have heart defects and kidney abnormalities. Almost all women with Turner syndrome are infertile. Intelligence is usually normal. Turner syndrome can occur with any pregnancy and is not associated with the mother's age.

What Makes NIPT Different?

Many of the traditional maternal serum screening tests available during pregnancy estimate a risk (or statistical probability) for certain birth defects. These tests are known as a Sequential Screen, AFP screen, or a First Trimester Screen. These screening tests are not able to provide a definitive answer.

Amniocentesis and CVS are able to diagnose or rule out a chromosome disorder. However, these tests are invasive and have a very small risk for miscarriage.

Non-Invasive Prenatal Testing (NIPT) provides more definitive information about the pregnancy than a risk estimate and there is not a risk for miscarriage.

Follow up to NIPT

For every pregnancy the background risk for birth defects is 3-5%. Because of this, the doctors at Desert Perinatal Associates will continue to monitor the growth and development of your baby with ultrasound, regardless of your NIPT results. Patients are informed that a normal or reassuring ultrasound reduces the risk of abnormalities in the baby, but is never a guarantee of a normal outcome.

How accurate is NIPT?

NIPT is very reliable, with high detection rates (over 95%) and low false positive rates (less than 1%) for Trisomy 21, 18, and 13.

What does a negative NIPT result mean?

A negative screen result means that the baby is at a very low risk to have Down syndrome, Trisomy 18, or Trisomy 13. However, even with a normal NIPT result there is still a small chance the baby could be affected with a chromosome disorder or other genetic syndrome.

What does a positive NIPT result mean?

A positive screen result does NOT mean that the baby definitely has a problem. However, it does mean that the chance of a chromosome problem is very high. It would be important to have genetic counseling to discuss a positive result in more detail.

After counseling, patients have the option to proceed with a diagnostic test, such as chorionic villi sampling (CVS) or an amniocentesis. These tests are diagnostic and would able to confirm the results from NIPT or rule out a chromosome abnormality. Diagnostic testing is not completely risk free, but in the hands of an experienced physician, such as the Perinatologists at Desert Perinatal Associates, the risk for causing a miscarriage is very small.

The doctors and genetic counselors at Desert Perinatal Associates will help you understand your risk during pregnancy and explain how ultrasound and diagnostic tests can help you learn more.