Patient Information: Noninvasive Prenatal Testing

What is Noninvasive Prenatal Testing?
Noninvasive Prenatal Testing is an optional test that is available to certain populations of pregnant women. At this time, it is being offered to:

- Women who will be 35 years old or over at delivery
- Women with a positive screening test (first trimester screen, quad screen)
- Women who have had abnormal ultrasound findings
- Women with personal or family history of chromosomal abnormalities

This test can be done starting at the 10th week of pregnancy. Noninvasive Prenatal Testing is an advanced screening test to identify babies with chromosome abnormalities such as Down syndrome, trisomy 13 or trisomy 18. The test can also identify abnormalities with the sex chromosomes, such as Klinefelter syndrome (XXY), Triple X syndrome (XXX) and Turner syndrome (XO). This test cannot determine for certain if the baby actually has one of these conditions; instead, it helps to find pregnancies that are at a much higher risk. If a higher risk is identified, further tests that can confirm or rule out these conditions are available.

What does the testing involve?
The test involves obtaining a blood sample from the pregnant woman. Two tubes of blood are taken from her arm for the test. There is no risk to the pregnancy with this test.

How does the test work?
Babies with chromosomal abnormalities such as those tested for with this test have extra or missing chromosomes in every cell of their body. Tiny pieces of the baby's chromosomes or DNA are normally present in a woman's blood during pregnancy. This test measures the number of pieces of DNA from each chromosome to determine if there seems to be the normal amount, too much or too little. If the results are abnormal and there is too much or too little DNA, it is highly suggestive that the baby has one of these conditions and further tests are recommended to confirm the result.

When will I get results? How are they reported?
The results are available approximately 10-14 days after the test is performed. Results are reported as "positive" or "negative." A positive test result means that the chance for Down syndrome, trisomy 18, trisomy 13 or a sex chromosome abnormality is greatly increased. A negative result reduces the chance significantly. A genetic counselor will call you with your results as soon as they are available. If the results come back positive, the genetic counselor will recommend another appointment to discuss the results in detail and options for further testing. All results will be forwarded to your doctor or midwife.

What do I need to do if my results are negative?
When the results are negative it means that your risks Down syndrome, trisomy 18, trisomy 13 or a sex chromosome abnormality have been decreased to less than 0.1%. *Noninvasive Prenatal Testing can never eliminate the chance for Down syndrome, trisomy 13, trisomy 18, sex chromosome abnormalities or other health problems. Many people feel reassured by a negative result and may decide to not have further invasive testing such as a* Chorionic Villus Sampling (CVS) *or* Amniocentesis. *Other people may wish to have a definitive diagnosis and so they may choose further testing.*

What do I need to do if my results are positive?
A positive test result means that the chance for Down syndrome, trisomy 18, trisomy 13 or a sex chromosome abnormality is greatly increased. *A positive result does not mean that the baby has one of these conditions but it does make it much more likely for the baby to have Down syndrome, trisomy 18, trisomy 13 or a sex chromosome abnormality*. If your noninvasive prenatal test results come back positive, diagnostic testing is available by either Chorionic Villus Sampling (CVS) until 14 weeks, or Amniocentesis after 16 weeks. Another screening option that can be considered is Targeted Ultrasound. The genetic counselor will discuss these options with you in greater detail.

What is the accuracy of the test?
Noninvasive Prenatal Testing can detect about 99% of pregnancies with Down syndrome or trisomy 18 and approximately 91% of babies with trisomy 13. It can detect about 99% of babies with a sex chromosome abnormality.

What if I have a family history of Down syndrome or other chromosome or genetic conditions?
Patients/couples with a family history of any genetic condition should discuss testing options with their doctor and/or genetic counselor.

What else can Noninvasive Prenatal Testing detect?
As this test identifies the sex chromosomes, it can identify if the baby is a girl or a boy.

Do I still complete the First Trimester Screen or Quad screen if I do Noninvasive Prenatal Testing?
No. If you complete Noninvasive Prenatal Testing, you should not do the blood portion of the First Trimester Screen or the Quad Screen. Noninvasive Prenatal Testing has a higher detection rate for Down syndrome, Trisomy 18 and Trisomy 13 than the First Trimester Screen and the Quad Screen.

What other tests are recommended in addition to Noninvasive Prenatal Testing?
W**e recommend a first trimester** ultrasound to evaluate the fetal nuchal translucency at 12-14 weeks gestation, a blood test called AFP at 15-20 weeks gestation to screen for spina bifida, and a 20 week targeted ultrasound.

Can all birth defects be detected by Noninvasive Prenatal Testing or any other screening?
No. For example, this test will not identify a baby with a heart defect or a cleft lip. No prenatal test can guarantee a child free of birth defects or learning problems. A genetic counselor can discuss the benefits, risks and limitations of prenatal testing.

Where is Noninvasive Prenatal Testing performed?
This test is completed at UnityPoint Health - Meriter Center for Prenatal Care. Valet parking or parking within Meriter's parking ramp are both available free of charge to our patients.

How long does the test take?
In general, we schedule 45 minutes for the appointment. You will first meet with a genetic counselor to review the test process and the blood draw can happen immediately following the appointment in the clinic. The results of the blood test are available in about 10-14 days.

How much does it cost? Does my insurance cover this testing?
It is recommended that you check with your insurance company to see if they will provide coverage. Given that this is a newer test, some companies do not cover this testing and some patient have to pay out of pocket for the testing.

Questions?
Please feel free to contact us if you have any questions or concerns (608) 417-6667. Also, please visit our website (www.unitypoint.org) for more information about other services provided at UnityPoint Health - Meriter's Center for Perinatal Care.

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