Women's Health Partners, LLC

Diplomates American Board of Obstetrics & Gynecology

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GENETIC TESTING OPTIONS & CONSENT FORM

The American College of Obstetricians and Gynecologists (ACOG) recommends that all pregnant women be offered options for genetic testing. Genetic counseling is recommended to all pregnant women who will be 35 years old or older at delivery and is available to all pregnant women regardless of age if desired.

Babies may be affected with chromosome abnormalities, the most common being Down syndrome, a disorder that leads to intellectual disability and other birth defects. Trisomy 18 is another chromosomal disorder that causes severe intellectual disability and birth defects.

Babies can also be affected by open neural tube defects. The neural tube is a structure that develops into a baby's spinal cord and brain. The tube is open early in the pregnancy and normally closes as the embryo develops. When the tube fails to close properly, it results in an opening. Severity varies depending upon the size and position of the opening, from paralyzed legs to incomplete brain development. The lack of closure below the neck results in a disorder known as Spina Bifida.

Generally, risk of chromosome abnormalities becomes greater as the age of the expectant mother increases. For mothers 35 years of age or more at the time of delivery, the standard recommendation has been to offer CVS or amniocentesis, an invasive test which involves the removal of a sample of the amniotic fluid for analysis. Non-invasive prenatal testing using cell-free fetal DNA from maternal blood samples is now also available.

Non-invasive screening tests using blood samples and/or ultrasound are also available and generally offered to those under the age of 35. These tests are completed during the first and second trimester of the pregnancy and provide information about your risks for carrying a baby affected with Down syndrome, Trisomy 18 and open neural tube defects.

It is important to understand that a screening test is limited; a result that shows increased risk does not mean that the baby actually has an abnormality; a result that is within the normal range does not guarantee that there are no abnormalities present. Mothers whose test results show an increase risk will be offered further evaluation by invasive testing with amniocentesis or chorionic villus sampling (CVS), diagnostic tests that can identify most known chromosomal abnormalities.

It is also important to understand that every pregnancy has some risk (3-5%) for the kinds of birth defects that cannot be diagnosed during pregnancy, such as autism, non-specific intellectual disability, some genetic diseases, and some types of anatomic/physical birth defects.

For women **under** 35 years of age at the time of delivery

We are offering the "**Integrated Screen**" as our standard non-invasive screening test. The test works as follows:

- Approximately between your 11th and 13th week of pregnancy, a small amount of your blood is drawn to evaluate the levels of certain proteins and hormones produced during your pregnancy.
- A special ultrasound measurement is taken of the back of the baby's neck at the same time called the nuchal translucency.
- A second blood test is performed between your 15th and 21st week (optimal time is 16^{th} -18^{th} weeks).
- Results from Part 1 and Part 2 are combined to provide a final, complete result and assessing your risk of having a baby with Down syndrome, Trisomy 18 or an open neural tube defect. There no results before the second blood draw.
- If your screening test shows an increased risk, it does not mean that a problem has been diagnosed. It only means that your baby should be further evaluated. In that case, you will be offered additional tests which can determine whether the baby has a disorder or if there are other explanations for the test result.
- If your screening test shows results in the "normal range", it does not guarantee that your baby is normal. It means that the risk of a chromosome problem is low. Other problems or abnormalities may be present or may develop in the baby.

The "Integrated Screen" has a detection rate for Down syndrome of approximately 92%, for Trisomy 18 of approximately 90%, and for open neural tube defect of approximately 80%.

You may also choose not to undergo any screening test. Some patients who feel that they would not intervene if the baby should have a problem may prefer this option.

For women **over** the age of 35 years of age at the time of delivery

We are offering the "**Non-Invasive Prenatal Test**" as our non-invasive screening test (maternal blood sample) or the genetic amniocentesis as our invasive diagnostic test (an invasive test which involves the removal of a sample of the amniotic fluid for analysis).

The "**Non-Invasive Prenatal Test" or "NIPT"** works as follows:

- We obtain a maternal blood sample after your 10^{th} week of pregnancy.
- A small amount of your blood is analyzed for cell-free fetal DNA, which is used to evaluate for the risk of certain chromosomal abnormalities:
    - trisomy 13, trisomy 18, trisomy 21 (Down Syndrome)
    - Some aneuploidies: 45,X (Turner's Syndrome), 47, XXY (Klinefelter Syndrome), 47, XXX (Triple X), 47, XYY
- This test serves only as a **SCREENING** test and does not replace the precision obtained from diagnostics tests such as chorionic villus sampling (CVS) or amniocentesis.
- False-positive test can occur, and confirmation of a positive test results with either CVS or amniocentesis is recommended.
- False-negative tests can also occur, and a negative test results does not ensure an unaffected pregnancy.
- This test does not screen for open neural tube defects. Therefore, we also recommend a maternal serum alpha-fetoprotein (MSAFP) blood sample at approximately 16-17^{th} week of pregnancy.
- A nuchal translucency ultrasound at 12-13^{th} weeks of pregnancy can also help in screening for other birth defects.

The screening tests and diagnostic tests offer the following performance:

Screen Type	When performed	Down syndrome detection rate	Trisomy 18 detection rate	Trisomy 13 detection rate	Open Neural Tube detection rate	Risk to the baby	False positive rate	Can reveal gender or sex
Integrated Screen	**Part I : 11th – 13th wks. (ultrasound + blood test)** **Part II : 16th – 18th wks. (blood test)**	**92%**	**90%**	**Does not detect**	**80%**	**0%**	**5%**	**No**
Noninvasive Prenatal Testing (NIPT)	**Any time after the 10th wk.** **(blood test)**	**99%**	**98%**	**91%**	**Does not detect**	**0%**	**<1.0%**	**Yes**

Genetic Counseling with a certified genetic counselor is available to all patients at Women's Health Partners. Please discuss these options with your physician or midwife. **These tests are time sensitive** and **must** be done at certain times and will require scheduling in advanced. You must also check with your insurance provider regarding benefits covering the above options.

I understand that there are benefits and limitations for any test, including false positives and false negative results. I have read the above information. All my questions have been satisfactorily answered. I understand that this test is voluntary, and I may decline testing at any point.

If you are less than 35 years-old at delivery, and have not had a previous baby with a chromosome anomaly or an Open Neural Tube Defect (ONTD), I choose:

- ❑ Integrated Screen **(the standard recommendation)**:
  sonogram for nuchal translucency and a blood tests at 11th – 13th wks and at 16th -18th wks.

- ❑ Elective Noninvasive Prenatal (NIPT):
  blood test after the 10th week of pregnancy. It can reveal the presence of certain chromosomal abnormalities like Down's Syndrome and can also reveal the sex of the fetus. **(This is an out-of-pocket test that your insurance company may not cover if you are under the age of 35 years old at delivery and without a medical indication)**

- ❑ No testing at all.

If you are 35 years old or more at the time of delivery, or have had a previous baby with a chromosome anomaly or an Open Neural Tube Defect (ONTD), I choose:

- ❑ Genetic counseling **(the standard recommendation)**:
  After meeting with the genetic counselor, a recommendation will be made. It will most likely involve possible Noninvasive Prenatal (NIPT) in conjunction with screening for open neural tube defect (NT ultrasound and MSAFP) or possible chorionic villus sampling (CVS) or amniocentesis.

- ❑ No testing at all.

_____	_____	_____
Patient Signature	Date	Account #
_____	_____	
Patient Name (Print)	Pregnancy weeks or due date	
_____	_____	
Physician	Date	
_____	_____	
Witness	Date	

While prenatal testing is frequently covered by insurance, you should contact your health insurance plan to obtain specific details about your individual plan benefit and coverage for this laboratory testing, as well as your out of pocket financial responsibility.

When researching your specific benefits and coverage, your health insurance plan may need to know the name/type of test being ordered, and the CPT code that will be used to bill the service

Test Name	CPT Code	Diagnostic Code
Integrated Screen	84163	Z36 (antenatal Screening)
Noninvasive Prenatal Test (NIPT)	81420	Z36 (antenatal Screening)