Birth, Babies and Beyond, LLC
Information Sheet: Genetic Screening and Diagnostic Testing

This information below is about genetic testing. These are tests that are offered to all women during pregnancy but, as always, you have the choice to decline. These tests look for different congenital anomalies that your baby may have, including Down's Syndrome, neural tube defects and Trisomy 18. All of these anomalies vary in severeness. Many times during your pregnancy (and during your child's life) you will be asked to make important decisions. Choosing whether to do genetic testing is one of those many decisions.There are several options for genetic screening and testing in pregnancy. As always, we encourage you to ask questions and to do your own research. We will support whatever decision you determine is best for you and your family. Below is some information about the different types of genetic screens and tests you could have. Please take a few minutes to read the information below and let us know if you have any questions. First, let's talk about the difference between screening and diagnosis.

Screening vs Diagnosis

Screening test:

- The purpose of these screens is to let you know if your baby is at an increased risk of a particular disorder.
- These are not diagnostic. This means that the results may be true, but they also could be false.
- Most screens are **not** 100% accurate and they can have false positives and false negatives.
- All of the tests used to screen have low risk or side effects to you or your baby.

Screening test results

- A **"positive"** result means that your baby has a higher risk of having a particular disorder. It does not mean that your baby is positive for the disorder. Further testing is required to find out more information.
- A **"negative"** result means your baby has a lower risk of a particular disorder. This does not mean that your baby is negative for the disorder.
- In truth, a woman who screens positive may have a baby with no abnormalities (a false positive) and a woman who tests negative may have a baby with an abnormality (a false negative).

Diagnostic test:

- This kind of test can accurately tell you if a particular disorder is present.
- These tests are very accurate but they do have an increased risk to your baby. (See info below).

Fetal DNA Testing?

There is a new test available called **Fetal DNA test also called Non-Invasive Prenatal Testing (NIPT).** This is done be taking a sample of the pregnant paretns blood and looking for fetal DNA (genetic material) from your baby. It is not considered diagnostic but it is very accurate and has no risk. It is not yet a standardized test so it is expensive and not covered by most insurances. There is more information regarding this test at the end of this hand out.

Birth, Babies and Beyond, LLC
Information Sheet: Genetic Screening and Diagnostic Testing

Screening tests

Which Screen?	*When is the screen performed?*	*How does it work?*	*What does it look for?*
Nuchal translucency screening. (NT)	Around 10-14 weeks of your pregnancy	Ultrasound machine is used to measure the nuchal translucency, the clear space behind the fetal neck.	Down Syndrome, Trisomy 18, Turner Syndrome, neural tube defects and some heart defects
1st trimester maternal serum screen (MSS)	11-14 weeks of your pregnancy.	It is a blood test that looks at certain markers in parents blood as well as an ultrasound	Down Syndrome, Trisomy 18
Maternal serum screen (MSS) Also called Quad or Penta screen	Around 15-18 weeks of your pregnancy	It is a blood test that looks at levels of certain hormones in your blood. Your age, weight, race and diabetes status are also considered.	It is a screen for Down Syndrome, Trisomy 18 and neural tube defects.
Ultrasound/Anatomy screen	Around 18-20 weeks of your pregnancy.	An ultrasound machine uses sound waves to make a picture of your uterus, placenta, and fetus.	Looking for structural defects. Looks at all the organs as well as spine and head and the fetus in general.

Diagnostic Testing

Which Test?	*When is the*	*How does it*	*What does it*	*Risks*

Birth, Babies and Beyond, LLC
Information Sheet: Genetic Screening and Diagnostic Testing

	test performed?	*work?*	*look for?*	
Fetal DNA test also called Non-Invasive Prenatal Testing (NIPT) **More information on this test on the following page.**	As early as 10 weeks	Taking a sample of pregnant parents blood and looking for fetal DNA (genetic material) from your baby	Down's Syndrome and trisomy 18 and 13	none.
Chorionic Villus Sampling (CVS)	10-14 weeks	A small tube is inserted into your vagina and through your cervix and a small piece of the placenta is removed. An ultrasound is also used to monitor where the tube goes.	99% accurate in detecting Down's. Can also detect other malformations. It does not detect neural tube defects.	1/100 risk of miscarriage, discomfort, bleeding, contractions, amniotic fluid may leak, emotional distress, cost if uninsured.
Amniocentesis	15-18 weeks	With ultrasound guidance a needle is inserted into your abdomen to take some amniotic fluid that will be tested in a lab.	99% accurate for detecting genetic anomalies like Down's, Trisomy 18 and neural tube defects	1/200 miscarriage risk, discomfort, bleeding, contractions, amniotic fluid may leak, emotional distress, cost if uninsured, it can take up to 2 weeks to get results.

Birth, Babies and Beyond, LLC
Information Sheet: Genetic Screening and Diagnostic Testing

FETAL DNA TESTING FAQ

(Taken from the OHSU website)
http://www.ohsu.edu/xd/health/services/women/services/pregnancy-and-childbirth/during-your-pregnancy/prenatal-screening-and-genetics/fetal-dna-testing.cfm

What's new?

Testing for some of the more common chromosome abnormalities is now available by taking a sample of pregnant parent's blood and looking for fetal DNA (genetic material) from your baby. These tests are often called "Non-Invasive Prenatal Testing (NIPT)" or "Fetal DNA testing."

What are these tests for?

These tests can tell us the chance, or likelihood, that a baby has certain types of chromosome abnormalities including Down syndrome, trisomy 18 and trisomy 13.

How accurate are these tests?

The accuracy of these new tests appears to be higher than other blood tests currently offered and can be performed any time in pregnancy after 10 weeks. However, these tests are not 100% accurate. They cannot screen for all known chromosome disorders. The results are not as accurate as CVS or amniocentesis.

What do the results mean?

Results generally take 1-2 weeks. A negative results means there is a very low chance a baby has Down syndrome, trisomy 18 or trisomy 13. A positive result means there is a very high chance the baby has Down syndrome, trisomy 18 or trisomy 13. In most cases, a positive result is confirmed with definative testing, such as amniocentesis. About 5% of women that do this test will not get results and may need to have their blood redrawn or will be offered further testing.

Who are the tests for?

These blood tests are currently available for women with high risk pregnancies. This includes women over 35, those with abnormal ultrasound findings, abnormal blood tests or a previous pregnancy with a chromosome abnormality.

Who performs these tests?

Three commercial laboratories currently offer these tests; Sequenom (MaterniT21plus™), Verinata Health (Verifi™ prenatal test), and Ariosa (Harmony™ prenatal test). There may be some out-of-pocket cost for these tests because not all insurance companies cover the cost.

What if I have more questions?

These tests are still very new. Testing options and results are complicated. If you are considering testing or have results to review, you may benefit from meeting with a prenatal genetic counselor. Genetic counselors are trained medical providers who can discuss the pros and cons of screening, help you understand testing results, and support you in your decision-making.

http://www.aruplab.com/files/technical-bulletins/Maternal%20Serum%20Screen,%20First%20Trimester.pdf

Birth, Babies and Beyond, LLC
Information Sheet: Genetic Screening and Diagnostic Testing

Signature Page:Please fill out and return to your midwives.
I have been given information and read the hand out, **Information Sheet: Genetic Screening and Diagnostic Testing**, that is available online at
http://www.birthbabiesbeyond.com/forms.html

I have chosen the following option/s: (please initial next to choice)

_____ To do genetic screening in my pregnancy. Please write which type of screening you would like to do. _____

_____ 20 week ultrasound anatomy screen

_____ No testing of any kind

Signature: _____ Date:_____

Updated 3/21/2017