PRENATAL SCREENING AND TESTING

The following are optional test that are available to you some are offered starting at 10 weeks.

The Non invasive Prenatal Test: (NIPNT) also called Prenatal cell-free DNA (cfDNA) is a genetic blood test (DNA) that looks at fetal cells in the mothers blood. This test can identify Downs Syndrome and Trisomy 18 among other genetically linked disorders. It also can identify the baby's sex. There are a few names for their test: Clari, Panorama and Materna 21 are among a few. Some insurance companies do not reimburse for this test unless there is a risk factor, age over 35 is considered one and history of genetic disorders is the other. It is wise to find out from your insurance company if they will reimburse for this test. This test can be done as earlier as 10 weeks. Please check out link below for details about this test.

The First Trimester Screen: is the Nuchal Translucency and Blood Test. The NT is an ultrasound that looks for a fold of skin behind the baby's neck referred to as the nuchal fold. Looking at the moms age, results of the blood test along with the visual findings of the ultrasound, the risk of Down Syndrome and Trisomy 18 can be gauged. This test is done between 11-14 weeks.

Even though the NIPNT determine the same risk, it is still recommend that the NT is done with the NIPNT. Many women decide to decline it since it is an ultrasound. The NIPNT is also done earlier than in the past. That said, if your insurance company does't want to pick up the tab for the NIPNT and you want this screening the NT is always covered.

Carrier Screen: Fragile X, Spinal Muscle Atrophy (SMS), Cystic Fibrosis, along with other screening depending on your ethnicity: **Tay-Sachs, Sickle Cell etc.** are also available to you at this time. If you were tested for these in the past you do not need to be tested again.

The Quad Screen: is a multiple marker done in the second trimester. Alpha- Fetal Protein (AFP) tests for Neural Tube Defect (spina bifida) it also screens for downs and trisomy 18, if you missed that opportunity you can still do it in the second trimester. Done preferably at 15 weeks (15-20 weeks is OK)

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All genetic testing are screening tests meaning they only are identifying a risk. Amniocentesis is the only diagnostic test, and would be recommended after a positive results and genetic counselling. It is invasive and can take 48 hrs to 2 weeks depending on what tests they do.

Many tests are available early so if there is a problem the decision to terminate the pregnancy is available as early as possible.
This can be a difficult decision for some when deciding, please always keeping in mind the question, what will I do with this information. I suggest to read this, discuss it with your partner, then sit with it and listen to your baby and your intuition, Then make your decision. For your own peace of mind please don't read forums and sites not listed below.

I am available to review this with you verbally, but ultimately the decision is yours.

Non invasive Prenatal Test (NIPNT) also called Prenatal cell-free DNA (cfDNA)
https://www.mayoclinic.org/tests-procedures/noninvasive-prenatal-testing/about/pac-20384574

Cystic Fibrosis, SMA and Fragile X
https://www.clinicallabs.com.au/media/1135/gene-access-carrier-screen-bro-aclmar-bf-nat-01033.pdf

Maternal serum alpha-fetoprotein (MSAFP) and multiple marker screening
https://www.webmd.com/baby/guide/second-trimester-tests#1

PRENATAL SCREENING AND TESTING

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Prenatal Screening and Testing

Prenatal testing may be offered to women during pregnancy to determine if the fetus has a possibility to be born with a genetic condition or birth defect. Performing prenatal testing may be useful in determining different options for the pregnancy or special management of the pregnancy and delivery to improve the outlook for the baby. Several types of prenatal testing are available, depending on which trimester of pregnancy the mother is in and the type of condition in question. This appendix provides an overview of different prenatal tests that may be offered to pregnant women.

Who Is Offered Testing?

All pregnant women, regardless of age, have the option to undergo prenatal testing. However, as women age, the chance of having a baby with a chromosomal abnormality increases. So the age of the mother is the most common reason for prenatal testing. Other reasons that a woman may be offered prenatal testing include:

- Family history or a previous child with a genetic condition
- Parents who are known carriers of a specific genetic condition
- Abnormal ultrasound findings
- Screening test results

Any woman who desires more information about the developing fetus can consider prenatal testing. The decision is an individual choice. A woman should discuss the various options outlined above with her obstetrician or a genetic counselor to determine which are right for her.

How are Tests Performed?

Two main types of prenatal testing are performed during pregnancy. The first type of testing is known as screening. Screening tests are used to identify women with an increased chance to have a baby with certain chromosomal abnormalities. Screening tests do not identify birth defects such as genetic diseases. Results that reveal a chance over a certain cutoff level are called “positive results,” and these women are offered further testing. Screening tests are not diagnostic. And while the majority of fetuses with a chromosomal condition are identified through screening, some affected fetuses with a chromosomal condition receive a normal or “negative” screening result.

The second type of prenatal testing is known as diagnostic testing because these tests can determine definitively if the developing fetus has a certain genetic condition or birth defect.

Screening and diagnostic tests may be performed in either the first or second trimester of pregnancy as follows.

Screening Tests

Screening tests can be performed in both the first and second trimesters of pregnancy. First trimester screening involves an ultrasound examination and a sample of the mother’s blood, while second trimester screening involves just the blood sample. Some women may

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also be offered a combination of first and second trimester screening, known as either "integrated" or "combined" screening. The blood results and ultrasound results are then combined with maternal factors such as age and weight to calculate the chance for certain chromosomal conditions in the current pregnancy.
Screening results are usually available within a week, and those who receive a positive result are offered diagnostic testing. The detection rate for screening tests varies by the type of test performed. The only way to know for certain whether or not a developing baby has a chromosomal condition is by performing a diagnostic test.

Diagnostic Tests

Certain diagnostic tests are procedures that can determine with greater than 99.9 percent accuracy whether or not a developing baby has a chromosomal difference. The two types of diagnostic tests are chorionic villus sampling (CVS) and amniocentesis. Diagnostic tests for specific genetic diseases must be specially requested. These tests have different accuracy rates, depending on which test is ordered.
CVS is performed between 10.5 to 13.5 weeks of pregnancy. During the procedure, a doctor obtains a small tissue sample from the placenta by either inserting a thin needle through the woman's abdomen or by using a small catheter inserted through the cervix. The method used depends on the location of the baby and the placenta.
Amniocentesis is performed from 15 weeks of pregnancy onward. During amniocentesis, a thin needle is inserted through the woman's abdomen into the amniotic sac to withdraw a small sample of fluid from around the developing baby.
The cells collected from either procedure can be used for chromosomal analysis or other genetic tests, as ordered. The results from the chromosomal analysis usually take two weeks; while the results from other genetic tests may take longer, depending on what test has been ordered.
Diagnostic test procedures are associated with a chance for miscarriage, which is estimated to be up to 1 percent for CVS, and less than 1 percent for amniocentesis.

References

1 American College of Medical Genetics www.acmg.net
2 American College of Obstetrics and Gynecology www.acog.org
3 March of Dimes Foundation www.marchofdimes.com
4 National Society of Genetic Counsellors www.nsgc.org

also look at - https://www.genpathdiagnostics.com/womens-health/for-patients/prenatal-for-patients/

Understanding Genetics: A New York, Mid-Atlantic Guide for Patients and Health Professionals.