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GUIDE TO RECOMMENDED TESTS IN PREGNANCY AND TIMEFRAMES FOR TESTING

Please note that the first and second trimester test can only be drawn during specific times in pregnancy. This is due to the fact that levels of the substances measured in these test change during the different stages in pregnancy. Therefore, it is critical that you keep your appointments as scheduled so that our office can help you to coordinate these visits at the proper time. Ultimately, it is your responsibility to make sure you get these test done at the proper time.

NIPT (Non Invasive Prenatal Testing)

NIPT, analyzes cell- free fetal DNA circulating in maternal blood. This testing is done by drawing a sample of the mother's blood. It is typically done ***between 10-22 weeks of pregnancy.*** NIPT is a laboratory- developed test accredited by CAP (College of American Pathologist) and certified by CLIA(Clinical Laboratory Improvement Amendments). NIPT is used to detect Trisomy 21 (**Down 's syndrome**), Trisomy 18 and Trisomy 13. They can also check for sex chromosome abnormalities (and check for gender)!
NIPT can detect 99% of pregnancies with **Down 's syndrome**. Since this test is not 100%, it is possible to miss chromosomal abnormalities. It does **not** screen for many of the other, less common, chromosomal abnormalities.

GENETIC TESTING FOR SPECIFIC GENETIC MUTATIONS

We now have the technology to screen for a variety of inheritable **genetic mutations**. Someone can be **a carrier** of a genetic mutation but not manifest the disease because they only have "bad" copy of the gene. When these mutations are inherited such that an individual inherits two copies of an abnormal gene, then they most often manifest with the disease. So if two carriers come together, they have a 25% chance of producing a child with the disease. If we find that both parents are carriers of a mutation, you will be referred to a genetic counselor to further discuss

diagnostic testing such as chorionic villi sampling (CVS) or amniocentesis. Examples of some of the more common diseases that are due to inheritance of a mutated gene would be cystic fibrosis or spinal muscular atrophy. Caucasian individuals are at the highest risk for being carriers of the cystic fibrosis mutation (1/25) but even individuals of other ethnic descents can carry the gene.

The laboratory that we use to do our genetic mutation is called "Counsyl" so we often refer to this test in our office as the "Counsyl genetic tests". They use a technique called gene sequencing which produces the most accurate result possible for these tests. Couples should understand that this test is completely optional. Dr. Varkey will speak to you at length about this testing at your first pregnancy visit to make sure you understand the benefits and limitations of the tests and to allow you an opportunity to ask further questions in this regard.

The First Trimester Screening Test

This test includes a blood test and an ultrasound exam. The First Trimester Screening Test is done between 11 and 14 weeks of pregnancy to detect the risk of **Down's Syndrome** (Trisomy 21) and **Trisomy 18** (the second most common chromosomal abnormality). This screening is done here as part of a two step process:

1. The first part is a blood test that measures the level of two substances in the mother's blood: PAPP-A and hCG. This test needs to be done ***after 10 weeks but before 14 weeks of pregnancy.*** This blood test can be done in our office during your routine prenatal visit.

2. The second part is an ultrasound exam called "the nuchal translucency screening" and is used to measure the thickness of the back of the fetus' neck. ***This test must be done between 11 -13 6/7 weeks of pregnancy.*** It is performed by a specialist that is trained to measure the nuchal fold in the fetal neck. (So you will need a referral from our office to this specialist).

The final results of this test will combine the results of the blood test, the nuchal fold measurement, and the mother's age to give a final assessment of the risk that this baby will have **Down's Syndrome** or Trisomy 18.

The Second Trimester Screening Test

This test is also known as the "Expanded AFP test" or the Quadruple Marker Test". It tests for **Down's Syndrome**, Trisomy 18, Neural Tube defects and a rare neurological disorder known as "Smith Lemli Opitz Syndrome". It is a blood test that measures the level of four hormones: AFP, Estriol, hCG, and Inhibin-B. It can be drawn in our office but it *must be drawn between 15-20 weeks of pregnancy.*

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Gestational Diabetes Screening

Gestational diabetes is a condition in pregnancy that makes it difficult for us to metabolize glucose (sugar). This test is done between 24-28 weeks of pregnancy. It consists of fasting for two hours and then drinking a glucose drink and having your blood drawn one hour after you finish drinking the glucose water. This is only a screening test. If the test returns showing abnormally high glucose levels in your blood, your physician will ask you to come in and do a three hour test with a more concentrated glucose drink. If this three hour test is also abnormal, then you will be diagnosed with gestational diabetes and referred to a specialist.

Group B- Streptococcus Test (GBS)

This is a bacteria that is normally found in the intestines of many adults. However, if the newborn "catches" this bacteria during vaginal delivery, it could cause an infection for the newborn. Therefore, we test our patients at 35-37weeks with a vaginal/rectal swab. If you are positive, we will give you antibiotics while in labor to reduce the concentration of the bacteria in your body at the time of delivery.

Invasive Testing for Select Patients

Chorionic Villus Sampling and Amniocentesis

These are two invasive tests used in early pregnancy in select patients in order to diagnose fetal chromosomal or genetic abnormalities. They are not recommended for every patient and we will discuss this more with you at your first visit if you are interested or if we believe it is indicated based on your personal risk factors (such as a maternal age greater than 35).

Please be aware that there are OTHER alternatives to these invasive tests – such as NIPT, first and second trimester screening tests previously mentioned. The descriptions below are not meant to imply that all patients over 35 will have to undergo invasive testing.

* Chorionic Villus Sampling (CVS)

This is an invasive test that is performed to obtain fetal cells for the purpose of diagnosing chromosomal abnormalities such as Down's Syndrome. It is generally performed between *11-13 weeks* by passing a needle through the cervix into the placenta under ultrasound guidance. It is performed by a maternal-fetal medicine specialist. It is recommended as an option for women over 35 but can be ordered on any patient who is interested in definitive testing for Down's syndrome or other chromosomal/genetic abnormalities.

* Amniocentesis

This is an invasive test that obtains fetal cells or amniotic fluid for the purposes of diagnosing genetic or fetal chromosomal abnormalities (such as Down's Syndrome) and neural tube defects (ie: spina bifida). It is generally performed after 16 weeks of pregnancy by passing a needle through the maternal abdomen into the amniotic cavity under ultrasound guidance. It is performed by a maternal fetal specialist and generally recommended for women over age 35. Again, it can also be ordered on any patient who is interested in definitive testing for Down's syndrome or other chromosomal/genetic abnormalities.