OBSTETRICS,
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Genetic Screening and Testing During Pregnancy

While most babies are born healthy and without birth defects, approximately 3-5% of all babies are born with a birth defect. Some of these babies will have birth defects that are due to or associated with genetic disorders or syndromes.

There are certain birth defects and genetic disorders that all pregnancies can be screened or tested for if a woman wants this information. All of these tests are **optional** and some women choose to do them while others do not. Some of the screening tests can be done prior to pregnancy (preconception), while others can be done only during pregnancy.

The following sections will provide an overview of these optional tests, as well as how to think about these choices:

- Birth defects commonly screened for or tested during pregnancy
- Difference between screening and diagnostic testing
- Screening tests for common birth defects in pregnancy
- Diagnostic tests for common birth defects in pregnancy
- Carrier screening
- How to decide whether or not to have screening or testing

Birth defects commonly screened or tested for during pregnancy
Down syndrome, trisomy 18, and open neural tube defects (like spina bifida) are the birth defects that all women are offered screening or testing for during pregnancy. Some tests will also provide information about trisomy 13 and other chromosome abnormalities.

Down syndrome, trisomy 18(Edward Syndrome), and trisomy 13 are chromosome abnormalities that are caused by a baby accidentally getting an extra chromosome (the chemicals in our cells that carry our DNA and genetic information) in the egg or the sperm. People with Down syndrome have mild to severe mental retardation and an increased risk for other issues such as heart defects. Babies with trisomy 18 or trisomy 13 are very severely affected with birth defects and mental retardation

and the majority do not survive past one year of age. The chance of a baby being born with a chromosome abnormality like Down syndrome, trisomy 18, or trisomy 13 increases gradually as a mother gets older; however, babies can be born with these abnormalities even if a mother is very young.

Open neural tube defects, like spina bifida, happen when the development of a baby's spinal cord is interrupted and does not fully close into a tube. This is usually a randomly occurring birth defect, but if someone in your family or the baby's father's family has spina bifida, the risk to your children may be higher. It is extremely important to take folic acid with your prenatal vitamin (ideally before you even start trying to become pregnant), as this decreases the chance of your baby having spina bifida.

Screening vs. diagnostic testing

Screening tests and diagnostic tests are two different options for getting more information about the chance that your pregnancy has certain birth defects. Screening tests are done with ultrasounds, blood tests, or both. Diagnostic tests are considered "invasive" tests and involve inserting a needle into the area around a pregnancy.

The benefits of the screening tests are that they involve no risk to you or the pregnancy. The limitations of screening tests are they have false positives and false negatives. In other words, a woman can have an abnormal screening test and a normal fetus; or normal screening test and an affected fetus. Most women who have screening tests use them to help decide if they want to proceed with further diagnostic testing.

The benefits of the diagnostic tests are that they diagnose certain birth defects, giving you a "yes or no" answer, and that they look at more birth defects than screening. The risk of diagnostic tests is that, because they are invasive tests, they involve a relatively small risk of miscarriage.

Screening tests for common birth defects in pregnancy

First Trimester Risk Assessment **(the screening test for women less than 35 years of age)**

The first trimester risk assessment is a screening test that can help determine a pregnancy's risk for Down syndrome and trisomy 18. The screen is performed between 10 and 13 weeks of pregnancy. An ultrasound measurement of the fluid at the back of a baby's neck (nuchal translucency) is taken. Five to 7 days before your ultrasound, you will have blood drawn. Information from the blood work and ultrasound will give you a specific risk for Down syndrome and trisomy 18 in the pregnancy. If these risks are considered high risk, you will be offered the option of proceeding to diagnostic testing to get a "yes or no" answer. If you do not elect diagnostic testing after the first trimester risk assessment, then you will have the

option to proceed with cell free DNA testing (see below) Based on these results, you can decide if you want to have diagnostic testing.

Cell Free DNA Testing **(the screening test for women greater than 35 years of age)**

Cell-free fetal DNA testing is a new screening test that indicates if a woman is at increased risk of having a fetus with Down syndrome (trisomy 21), trisomy 18 (Edward Syndrome), trisomy 13 (Patau syndrome) or abnormalities of the X and Y chromosomes. This test is currently being offered to women who will be at least 35 years old at the time of delivery and to women who are found to be at increased risk for Down syndrome or trisomy 18 after the first trimester screen.

With this test, a sample of the woman's blood is taken after 10 weeks of pregnancy. The test measures the relative amount of free fetal DNA in the mother's blood. Cell-free fetal DNA testing is thought to detect greater than 99 percent of all Down syndrome pregnancies and greater than 98 percent of all trisomy 18 pregnancies. It detects about 65 percent of all trisomy 13 pregnancies. Women who are found to be at increased risk can have definitive testing with chorionic villus sampling (CVS), or amniocentesis.

18-week ultrasound, or Anatomy scan

The anatomy scan is an ultrasound that is performed during the second trimester of your pregnancy, usually between 18 and 22 weeks of pregnancy. This ultrasound will look for the presence of multiple structural birth defects, like heart defects, cleft lip, kidney problems, limb abnormalities, and differences in the way the brain is forming. While ultrasound can detect or diagnose a small number of structural abnormalities like cleft lip or club foot, it is also used as a screening test for some genetic disorders like Down syndrome. Most babies with "abnormal" ultrasound findings will be healthy and will not have a genetic disorder. However, some babies with a "normal" ultrasound will end up having birth defects or genetic disorders that we cannot detect using ultrasound.

Diagnostic tests for common birth defects in pregnancy

There are two common diagnostic tests performed during pregnancy:

chorionic villus sampling (CVS)
amniocentesis

Chorionic villus sampling (CVS)

CVS is a diagnostic test that is performed during the first trimester, typically between 10 to 13 weeks of pregnancy. The main purpose of CVS is usually to determine whether or not a baby has a normal number of chromosomes (46).

Therefore, CVS is diagnostic for birth defects like Down syndrome, trisomy 18, trisomy 13, and some other chromosome abnormalities. CVS can also be used to test for some other genetic conditions that your baby may be at risk for based on family history or carrier screening. CVS is generally performed in one of two ways, either by inserting a needle through a mother's abdomen or through the cervix into a pregnancy's placenta. A small piece of the placenta is then removed and sent to the laboratory for genetic testing The risk of miscarriage after a CVS is approximately 1/400.

Amniocentesis

Amniocentesis is a diagnostic test that is performed during the second trimester, typically between 15 and 18 weeks of pregnancy. The main purpose of amniocentesis is usually to determine whether or not a baby has a normal number of chromosomes (46). Therefore, amniocentesis is diagnostic for birth defects like Down syndrome, trisomy 18, trisomy 13, and some other chromosome abnormalities. Amniocentesis can also be used to test for some other genetic conditions that your baby may be at risk for based on family history or carrier screening. Amniocentesis is done by inserting a needle through a mother's abdomen and collecting the cells within the the amniotic fluid around the fetus. The risk of miscarriage after an amniocentesis is 1/400.

Carrier screening

All of us have thousands of genes in our bodies. These genes are made up of DNA and they provide the instructions that our bodies use to grow, develop, and function. We all have two copies of every gene. For certain genetic disorders to occur, a person with that disorder must have two copies of the same gene that are not working correctly. As long as a person has at least one working copy of these types of genes, he or she is healthy and usually has no signs or symptoms of the genetic disorder. People who have one working copy of one of these genes and one non-working copy are called "carriers". Carriers usually do not have a family history of the genetic disorder. It is thought that we all "carry" at least 8-10 of these non-working genes in our DNA. Usually, the fact that we have these non-working genes doesn't matter. However, if we have children with someone who carries a non-working copy of the same gene, these children are at increased risk for having the genetic disorder.

Carrier screening is offered to all people who are planning a pregnancy or who are early in their pregnancy. Your risk of being a carrier for a genetic disorder is not related to age. Some of the common genetic disorders that we have offered carrier screening for are cystic fibrosis (CF), sickle cell anemia, Tay Sachs disease, and spinal muscular atrophy (SMA). Carrier screening is done through a blood test on a pregnant woman or the father of the baby. A new test, called Counsyl, is available which tests for 88 recessive traits (including Cystic Fibrosis and Spinal Muscular Atrophy, Tay Sachs and SMA). This test is acquired through the saliva. Both parents should be tested because in order to have a baby that is affected, BOTH parents need to be carriers for the "bad" gene. We have included a separate hand-

out which provides more details about this test. You may also go to their website to learn more about the test at **Counsyl.com**. If you choose to do the Counsyl testing, you MUST sign up with the company on line for a counseling session with their genetic counselors in order to receive the results of your test. The results of a carrier screen do not change from pregnancy to pregnancy and therefore do not usually need to be repeated.

If both the mother and the father of a baby are carriers for the same genetic disorder, testing can be done prior to or during a pregnancy to determine if that particular pregnancy is affected (ie, the fetus received both "bad" genes).

Fragile X syndrome is a genetic condition that causes a range of developmental problems including learning disabilities and cognitive impairment. Usually, males are more severely affected by this disorder than females.

Affected individuals usually have delayed development of speech and language by age 2. Most males with fragile X syndrome have mild to moderate intellectual disability, while about one-third of affected females are intellectually disabled. Fragile X syndrome occurs in approximately 1 in 4,000 males and 1 in 8,000 females. More information about Fragile X can be found at:

http://ghr.nlm.nih.gov/condition/fragile-x-syndrome

A lot more information about each of these screening and testing options is available for women who want it. If you have questions about your options or want help making a decision, please speak to your doctor, nurse-midwife, or ask to speak with a genetic counselor.

Since insurance coverage varies based on individual plans, some or many of these tests may not be covered. PLEASE CHECK WITH YOUR INSURANCE PLANS BEFORE YOU PROCEED WITH GENETIC TESTING.

Reviewed by health care specialists at Obstetrics, Gynecology, & Menopause Physicians, PC.

This information is for educational purposes only and is not intended to replace the advice of your health care provider. We encourage you to discuss with your doctor or nurse-midwife any questions or concerns you may have.