SOUTHWEST OB/GYN

DIAGNOSTIC TESTING AND SCREENING

All pregnancies carry about a 3% risk of either a birth defect or intellectual disability regardless of the mother's age, prenatal exposures, or family history. Some of these birth defects can be detected before birth, but some cannot. There are a number of tests that doctors can use to try to detect certain birth defects before birth. The use of many of these tests is optional. You are not required to have the tests, but if you want to, they are available.

Screening tests for Down Syndrome (Trisomy 21), Trisomy 18, and Trisomy 13

Testing for Trisomy 21, 18, and 13 is optional and falls into two categories: Screening tests and Diagnostic tests. Screening tests are non-invasive tests (no risk to fetus) performed to ESTIMATE risk. Diagnostic tests are invasive tests that can give more PRECISE answers about the presence or absence of these conditions but have a small risk of miscarriage.

1. What is Down syndrome (Trisomy 21)?
   a. A condition that causes varying degrees of mental retardation.
   b. It is caused by an extra copy of the # 21 chromosome. 1/800 fetuses will be affected.
   c. The risk for fetal Down syndrome increases with age.
      1. At age 25 - the risk is 1 in 1200.
      2. At age 35 - the risk is 1 in 270.
      3. At age 40 - the risk is 1 in 80.
2. What is Trisomy 18 (Edwards Syndrome)?
   a. A chromosomal disorder that causes severe developmental delays and affected fetuses may have a cleft palate, abnormal hands and feet. and defects of the lungs, kidneys, stomach, intestines, and heart.
   b. It is caused by an extra copy of chromosome 18. About 1/5000 babies are born with Trisomy 18 and most are female.
   c. Many babies with Trisomy 18 don't survive beyond the second or third trimester.
3. What is Trisomy 13 (Patau syndrome)?
   a. A chromosomal condition which causes severe mental disability and physical abnormalities. Fetuses often have heart defects, brain or spinal cord defects, extra fingers or toes, cleft palate, and weak muscle tone.
   b. It is caused by an extra chromosome 13. 1/16,000 babies are born with trisomy 13.
   c. Many infants die within the first few weeks of life. 5 to 10% live past the first year.

Why would I test for Down Syndrome, Trisomy 18, or 13?

a. If my baby is affected, I want to be aware of the diagnosis so that I can better prepare for delivery and the potential problems the infant may have after birth.
b. If my baby has one of these conditions, I might make the choice not to continue the pregnancy.

Why would I not test for Down Syndrome, Trisomy 18, or 13?

a. I would not change the course of the pregnancy no matter what - if my baby has one of these conditions, it would not matter to me.
b. Even if my baby has a trisomy, I can find out more about it when I deliver - I don't want that information before my delivery.

What are my choices for screening tests?

1. Non-invasive prenatal testing (NIPT) –

    a. Can be done after 10 weeks of pregnancy.
    b. Is a blood test (from the mom) that analyzes circulating cell-free fetal DNA and can detect Trisomy 21, 13, 18, and also X and Y chromosomal abnormalities.
    c. The detection rates are 97- 99% for Down Syndrome, 97 – 99% for Trisomy 18, and 63 – 91% for Trisomy 13.
    d. A positive NIPT increases the chances of having a baby with one of these disorders but the only way to confirm is by amniocentesis or CVS since there can be false positives.
    e. NIPT does not evaluate any other chromosomal or genetic conditions. If you choose to do a NIPT, an AFP blood test will be offered at 16 to 20 weeks to give you a risk for spina bifida.

2. First trimester screen (FTS) –

    a. Performed at 11 to 14 weeks of pregnancy.
    b. Involves an ultrasound of the fetus and a blood test (from mom).
    c. Can detect 85- 91% of Down Syndrome and 85 to 95% of Trisomy 18 or 13 pregnancies. This test will miss 9 to 15% of Down Syndrome pregnancies. FTS may also detect up to 40% of heart defects.
    d. If you choose the First trimester screen, an AFP blood test will be offered at 16 to 20 weeks to give you a risk for spina bifida.

3. QUAD screen –

    a. Done at 15 to 21 weeks of pregnancy.
    b. Is a blood test in the mother.
    c. Detects 75% of Down syndrome pregnancies, 75% of Trisomy 18 pregnancies, and 90% of spina bifida or other neural tube defects.

4. Ultrasound (sonogram) -
    a. The anatomy scan is performed at 20 weeks and can detect some fetuses with Trisomy 21,18, or 13.
    b. Ultrasound alone is not a good screening test for Down Syndrome. Approximately half of fetuses with Down Syndrome will be able to be detected during the 20 week anatomy sonogram.

What do I do with the results of a screening test?

1. If the results show that you are “low risk”, the next test that will be offered to you is your ultrasound at 20 weeks. Being “low risk” does not absolutely rule out Down syndrome, but your risk is small enough that you may not consider additional testing.
2. If the results are “high risk” and show an increased risk for Down Syndrome or another defect , then you will be offered genetic counseling and a diagnostic test (a more precise answer), if desired. Some patients might choose to only have an ultrasound as they do not want invasive testing (which may carry a small risk to the baby).

Important points to remember about screening tests:

1. ALL testing is OPTIONAL - you are not required to do any testing.
2. Screening tests give you an estimate of your risk. If your test results indicate “high risk” it means that your pregnancy is at increased risk for a problem, not that the baby definitely HAS a problem. Many

patients who have results that are "high risk" DO NOT have a baby with Down Syndrome, but their chance of DS is higher than average.

3. If you have a "high risk" screening result, the only way to detect if the baby more precisely has a problem is to do an additional diagnostic test such as amniocentesis or CVS.
4. No screening test will find all cases of Down Syndrome (Trisomy 21), Trisomy 13, or 18.
5. No screening test will find all birth defects.

Diagnostic (invasive) tests will identify essentially 100% of Down Syndrome and other chromosome abnormalities, but carry a small risk. They will be offered if:

1. Your pregnancy is at increased risk for Down Syndrome, Trisomy 13/ 18, or spina bifida based on an abnormal screening test.
2. You wish to skip screening tests and proceed directly to a definitive diagnostic test.
3. An abnormality is found on ultrasound.
4. You have had a prior child with a chromosome abnormality or an inherited disorder that can be diagnosed prenatally.
5. You are a known carrier for a chromosomal or genetic disorder that can be diagnosed prenatally.

Types of invasive tests:

A. CVS (Chorionic Villus sampling) -
   1. Done at 10 to 12 weeks of pregnancy.
   2. A tiny sample of the placenta can be obtained and chromosomal studies are done from the sample.
   3. There is a slightly higher risk of miscarriage(approximately 1%) when compared to amniocentesis. However, it can diagnose an abnormality earlier in pregnancy.

B. Amniocentesis -
   1. Performed at 15 to 16 weeks.
   2. A small amount of fluid that is around the fetus is removed. This fluid contains fetal cells whose chromosomes are then studied. This test evaluates chromosomes and AFP (for spina bifida). Risk of miscarriage is approximately 0.5% (1/200).

Please let your provider know if you are interested in CVS or Amniocentesis.

Carrier Screening

Carrier screening involves a DNA test called the "Universal Genetic Test." Currently, these tests can identify up to 100 genetic conditions, including conditions like as Cystic Fibrosis, Spinal Muscular Atrophy, Fragile X Syndrome, Sickle-Cell Anemia, Tay-Sachs Disease, and many more. While some of these conditions are life threatening, others are not.

For the most part, the genetic condition being tested for is a recessive trait (meaning there is a normal gene and an abnormal gene), also called a carrier state. Because carriers have one "normal gene" and one "abnormal gene" , because of the normal gene, a person who is a carrier is normal and does not have any signs of the genetic condition. Thus, the only way to find out if someone is a carrier is to perform a DNA test on them.

If the mother and the father of an unborn baby both test positive for the same disease, then there is a 1 in 4 (25%) chance that their child could be born with that genetic disease. Statistics show that when we test for over 100 conditions, there is a significant chance that a person will have at least one of these rare genetic mutations. If the mom is a carrier of an abnormal gene, then we would recommend testing the baby's father.

Please feel free to ask us any questions you may have about Genetic Carrier testing.

Other ethnic specific screening

There are a number of other tests available for genetic diseases that vary in frequency between ethnic groups. For example, individuals of African American and Mediterranean descent may be at higher risk for carrying the gene for Sickle Cell Anemia. Individuals of Jewish descent are at higher risk for Tay Sachs and several other genetic diseases. Instead of a full carrier screening panel, your doctor may recommend screening for certain genetic diseases depending on your ethnic background.

Family history

It is important to discuss with your doctor any family history of inherited disease, birth defects, or mental retardation so that we can discuss with you any impact this may have on your pregnancy.

Genetic counseling

Genetic counseling is offered when there are specific risks to the pregnancy or you desire more detailed information about your risk for Down Syndrome or other genetic problems. The following are other indications for genetic counseling:

1. Your pregnancy is at increased risk for Down Syndrome or other disorders because of screening tests.
2. You will be over age 35 at the time of delivery.
3. You have a history of more than 2 miscarriages, a history of a stillbirth, a prior child with birth defects, or there is a family history of these problems.
4. You are concerned that you may have an inherited disorder.
5. Your ethnic background puts you at increased risk for genetic disorders.
6. You feel that your job, lifestyle, or medical history may pose a risk to the pregnancy (i.e. exposure to radiation, medications, chemicals, infection, or drugs).
7. Couples who are first cousins or close relatives might benefit from genetic counseling.
8. Patients who wish more detailed information about diagnostic testing.
9. Genetic counseling is required prior to the performance of CVS or amniocentesis.

Ultrasound

One of the most common procedures done during pregnancy is an ultrasound exam (sonogram). Ultrasound scanning involves the use of a hand held probe, called a transducer, which sends out sound waves of a very high frequency but of very low power. These sound waves bounce off of structures and are reflected back to provide a picture of the baby or pelvic structures. At present there are no known risks to the baby or the mother with an ultrasound exam.

There are many reasons why your doctor might order a sonogram during the pregnancy. In our practice, a sonogram is frequently ordered at about 20 weeks gestation in order to evaluate fetal anatomy, including the heart, brain, and spine. Other information such as placental location, amount of amniotic fluid, and fetal activity can also be assessed. Many, but not all, birth defects can be seen on ultrasound. Approximately half of Down Syndrome babies will be able to be detected with ultrasound alone.

Please remember that a sonogram is a medical diagnostic test, and in order to do the best job possible, the sonographer needs to concentrate fully on obtaining a complete set of images. While the sonogram is often exciting and entertaining, please remember the primary purpose is to confirm the health of the baby and provide you with excellent medical care.

Most common prenatal screening tests & the codes that identify them

Please check with your insurance company, by referring to these codes, as to whether or not these tests are covered.

Cystic Fibrosis
CPT/Procedure code: 81220
Diagnosis code/ICD-9: V77.6 (screening), V26.31, 656.93
Lab test code: 4222

First Trimester screen
CPT/Procedure codes: 84163, 84702, 76801, 76813
Diagnosis code/IDC-9: 656.93 (fetal or placental problem suspected)
Lab test code: 5625

Fragile X
CPT/Procedure code: 81243
Diagnosis code/ICD-9: V22.2 (screening), V26.31
Lab test code: 5217

Quad screen (AFP4)
CPT/ Procedure codes: 82105, 82677, 84702, 86336
Diagnosis code/ICD-9: 656.93 (fetal or placental problem suspected)
Lab test code: 5375

Spinal Muscle Atrophy
CPT/Procedure code: 81401
Diagnosis code/ICD-9: V22.2, V26.31
Lab test code: 5457

Panorama
CPT/Procedure code: 88271
Diagnosis code/ICD-9: V28.89, 659.63, 659.53, 655.13, 796.5
www.panoramatest.com

Counsyl (Carrier Screening)
Universal Panel CPT/Procedure codes: 81200, 81205, 81209, 81220, 81242, 81250, 81251, 81255, 81260, 81290, 81330, 81332, 81400(x5), 81401(x8), 81479, + Fragile X 81243
Diagnosis code/ICD-9: 656.93, V77.6
www.counsyl.com