Prenatal Diagnosis:
What Tests Are Right for You?
Most babies are healthy; only about 2 to 3 percent have a birth defect. Prenatal diagnosis means finding birth defects during pregnancy. Several different tests are available, and this document will help you choose which tests you want. Most likely all of the results will be normal, and you will be reassured.
PRENATAL DIAGNOSIS:
WHAT TESTS ARE RIGHT FOR YOU?

What kinds of birth defects are there?
There are three kinds of birth defects:
- Structural Defects
- Genetic Syndromes
- Chromosomal Disorders

STRUCTURAL DEFECTS

What is a structural defect?
A structural defect is when a specific organ or body part has not formed normally. Some examples include:
- Heart defects
- Spina bifida
- Clubbed foot
- Cleft lip

Who is at risk for giving birth to a baby with a structural defect?
Most structural defects seem to happen randomly. However, some women have an increased risk, such as those with poorly controlled diabetes, a family history of structural defects, and those who are carrying identical twins.

How can you tell if my baby will have a structural defect?
We typically do two ultrasounds during pregnancy. The first one, at 11 to 14 weeks, can sometimes find structural defects. The second one, at 18 to 20 weeks, is specifically to find birth defects and is called a second trimester structural survey.

Do you recommend screening with alpha fetoprotein?
No. Maternal serum alpha fetoprotein (MSAFP) screening has been used in the past to help find spina bifida and certain other birth defects. Since we can find these using a second trimester structural survey, we no longer recommend this test.

Are there structural defects that can’t be found by ultrasound?
Yes, unfortunately, there are.
Is ultrasound covered by my insurance?
Most insurance policies will cover an ultrasound at 11 to 14 weeks as part of screening for chromosome defects (see below) and a structural survey at about 18 to 20 weeks. We recommend you check with your particular plan to understand your coverage and see if there are any out-of-pocket expenses. Our office can help by giving you the correct test code when calling your insurance company.

What may be done if my baby has a structural defect?
We may recommend more tests, such as follow up ultrasounds, amniocentesis, or MRI. We often ask a pediatric cardiologist to look at the baby’s heart.

We may recommend that you have an elective delivery, either by induction of labor or a planned cesarean section.

If your baby needs surgery or other special treatments right after delivery, you should give birth in a hospital that can provide those services.

In rare cases we try to correct the problem before birth.

You may want to make plans to help you care for your child after birth. It is often useful to meet with pediatricians and other specialists before the delivery. You may want to meet with other parents of children with similar problems.

Ending your pregnancy is also an option.
GENETIC SYNDROMES

What are genetic syndromes?
The blueprint for the human body is ‘written’ in DNA. The ‘words’ are called genes. A mistake in the DNA can change how a gene works. This can result in a pattern of abnormalities called a genetic syndrome.

How can you tell if my baby will have a genetic syndrome?
We offer carrier screening for certain common diseases, looking for a copy of a defective gene in apparently healthy people. We also take a careful family history of both parents to see if we can find any other problems.

What syndromes can you find using carrier screening?
• Cystic fibrosis is a disease that mostly affects the lungs, but can also cause problems with digestion. It can be mild to very severe.
• Spinal Muscular Atrophy (SMA) causes muscle weakness that gets worse over time.
• Fragile X syndrome can result in intellectual disability – in the past this was called mental retardation. Boys are more severely affected than girls. Also, women who are carriers are at risk for certain problems themselves.

There are some disorders that are more common in certain ethnic groups.
• Anemias such as sickle cell disease and thalassemia are more common in people from the Mediterranean, Middle East, Africa including African-Americans and the Caribbean, other Hispanics, and Asians.
• Ashkenazi Jewish Diseases are a group of diseases seen more commonly in people of eastern European Jewish descent.
• Tay-Sachs disease is more common in French Canadians.

Does negative carrier screening mean my baby will not have a genetic syndrome?
No. There are many genetic syndromes that don’t have screening tests. Also, a negative screening test reduces – but does not eliminate – the chance that the baby will have that syndrome.

It’s also important to know that when we do find a syndrome, it is often difficult to predict how severely the baby will be affected.
How does carrier screening take place?
Usually the first step is a blood sample from the mother. If she is found to be a carrier of one of these syndromes, the next step is to see if the father is also a carrier for the same syndrome. Chances are he’s not! But if he is, we can see if the fetus is affected, using either a CVS or an amniocentesis, which are described below.

For Fragile X syndrome, we do not need to draw blood from the father.

What if I have more questions about genetic syndromes?
You can speak to a genetic counselor at any point in this process to help you learn more about your particular situation so you can make the best choice about what tests to get.

Are these genetic tests covered by my insurance?
Everyone’s insurance coverage is different, and not all genetic tests are covered by all insurance companies. Contact your insurance company to see which tests are covered and if there are any out-of-pocket costs. This information may help you decide which tests you want done. Our office can help by giving you the correct test code when calling your insurance.

Do I have to have these tests?
No. Many women choose to have screening only for certain genetic syndromes, or none at all.

CHROMOSOMAL DISORDERS

What are chromosomal disorders?
The blueprint of the body is ‘written’ in DNA, and the ‘words’ are called genes. These words are grouped together into chapters of varying length called chromosomes. We get one set of 23 chromosomes from each parent, for a total of 46. When a fetus doesn’t have the correct number of chromosomes, the most common result is a miscarriage. Sometimes the baby is born and can have problems after birth.

What kind of chromosomal disorders are there?
Down syndrome (also called trisomy 21) is the most common chromosomal disorder. People with Down syndrome can have a wide variety of problems including some degree of intellectual disability – in the past this was called mental retardation. Most adults with Down syndrome are unable to live completely independently. The risk of Down syndrome is greater in older women.
Trisomy 13 and 18 are less common than Down syndrome, but are much more severe. Babies are most often stillborn or die within the first year of life; survivors are severely disabled. The risk of these disorders is also greater in older women.

**Sex chromosome abnormalities** are relatively common but seldom result in major mental or physical handicaps.

**Other chromosomal abnormalities** are rare, and can have a variable effect on the baby.

**Microdeletions and microduplications:** We can now use DNA technology to find changes in the chromosomes that are too small to be seen with a microscope. Recognized disorders associated with these changes might be present in up to 1 in 200 (0.5 percent) of all pregnancies. However, we sometimes see a change in the DNA, but are unable to predict its effect on the baby. The presence of a so-called **variant of uncertain significance** can lead to difficult dilemmas for the prospective parents. Therefore, many women do not want us to look for these at all.

**How can you tell if my baby will have a chromosomal disorder?**

We need to examine a sample of fetal cells from the amniotic fluid or the placenta. There are two ways to obtain fetal cells:

**Amniocentesis** (sometimes called amnio) is most often done between 15 and 20 weeks. We insert a needle into the uterus and withdraw a small amount of the fluid that surrounds the baby. The risk that an amniocentesis could cause a miscarriage is so small that it’s hard to measure; we think it is about 2 in 1000, or 0.2 percent. It takes about 2 weeks to get the results.

**CVS** stands for chorionic villus sampling. We take a sample of the placenta typically between 10.5 and 14 weeks, so we can get the results earlier than from an amnio. However, it may be a bit more likely than amnio to cause a miscarriage. We think this happens in up to 10 in 1000 cases, or 1 percent.

**Who should have a CVS or an amnio?**

Due to the risk of miscarriage, most often only women who have an increased risk of having an affected baby choose to have a CVS or amnio. These women are at increased risk:

- Maternal age of 35 or older at delivery
- Family member or previous pregnancy with a chromosomal abnormality
- Abnormality seen on ultrasound
- Worrisome result on a **screening test**
The risk of chromosomal abnormalities gradually increases as a woman gets older, but there is nothing “special” about 35 year olds. For the same reason there is no simple way to separate a “worrisome” from a “reassuring” screening test result.

**What is a screening test?**

We use ultrasound and blood tests from the mother, along with the mother’s age, to estimate the risk of certain chromosome disorders, including Down syndrome. This can help women decide if they want to have an amnio or a CVS.

For example, a 39 year old woman starts out with a risk of Down syndrome of 10 in 1,000 (1 percent). Based on the results of a screening test we might make the risk higher, to perhaps 100 in 1,000 (10 percent); or we might make the risk lower, to perhaps 1 in 1,000 (0.1 percent). The first woman may decide to have an amnio or a CVS, while the second woman may be reassured by the lower risk and choose to avoid a procedure.

**What kind of screening tests are there?**

**ERA, or Early Risk Assessment** is performed between 11.5 and 14 weeks. We perform an ultrasound of the fetus and a maternal blood test. This allows us to calculate a risk for Down syndrome, trisomy 13 and trisomy 18.

If a woman is too far along for ERA, we can do a **quad screen** between 15 and 22 weeks. This is a maternal blood test that gives results that are similar to those obtained from ERA.

**Cell Free DNA** is also a blood test done on the mother. This test is better than ERA at detecting Down syndrome and trisomy 18, but may not be as good at detecting other chromosomal abnormalities. Neither ERA nor cell free DNA is as good as CVS or amnio, because only these invasive tests can reliably detect a wide range of chromosomal abnormalities without the need for additional confirmation.

As of 2015, most insurance policies will cover cell free DNA only for women who are at increased risk for a chromosomal abnormality based on age, family history, ERA, or findings on ultrasound. Low risk women who want this test will have to pay an additional charge.

We think that cell free DNA should be done in addition to, rather than instead of, an 11 to 14 week ultrasound.
What are the downsides of screening?
Screening is not definitive. Although it can tell us that your risk of Down syndrome or trisomy 18 is low, even a very low risk isn’t the same as no risk at all. In addition, screening does not find other chromosomal abnormalities that can be only be detected by CVS or amnio.

I’ve heard the terms “screen positive” and “screen negative.” What does that mean?
Some people want to know if the result of a screening test is “good” or “bad.” It’s a lot more complicated than that, because there is no good way to separate “worrisome” from “reassuring” screening test results. We leave it up to the prospective parents to decide if they want more testing based on their individual risks and preferences.

Are these tests covered by my insurance?
All major insurance companies cover ERA or a quad screen. Also, all major insurance companies cover CVS and amniocentesis, but only for patients who are at high risk based on age, screening results, or other factors. They will not cover these procedures on patients who are low risk.
Coverage for cell free DNA varies greatly and there may be considerable out-of-pocket costs, particularly for low risk patients (younger than 35 without worrisome results from ultrasound or another screening test). If you are considering cell free DNA, we will help you determine your out-of-pocket cost.

Do I have to have one of these tests?
No. Many women choose not to have any testing for chromosomal disorders.
MAKING A DECISION

Before you agree to have a test, you should think about what you will do with the information. Many couples would not have a CVS or amniocentesis under any circumstances. They would not end the pregnancy if the baby is affected, and/or they do not accept the risk of miscarriage from CVS or amnio. These couples might not want to have screening tests.

Think about these questions before deciding to have any form of genetic testing:

- Is this information I want to know before delivery?
- How do I feel about ending the pregnancy?
- How do I feel about raising a child with a disorder?
- What impact will this have on my family?
- Who will care for an adult with special needs when I am no longer able?
- How do I feel about miscarriage?
- If I decide not to have testing, will I worry about this until the baby is born?

Remember that all of these tests are optional and entirely your choice. Discuss your options with your provider or a genetic counselor.

This document has been reviewed for plain language by the staff of the Maxwell & Eleanor Blum Patient and Family Learning Center.
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