



## Johnson County OB/GYN, Chartered

### Prenatal Tests and Procedures

While you are pregnant your doctor may suggest a number of laboratory tests, ultrasound exams, or other screening tests.

### Laboratory Tests

We perform the following routine blood tests on all of our patients at their first OB appointment:

Rubella immunity  
HIV

Hepatitis B  
Syphilis

### Screening Tests

Screening tests measure the risk of having a baby with some genetic birth defects. Birth defects are caused by problems with a baby's genes, inherited factors passed down from the mother and the father. Birth defects can also occur randomly in people with no family history of that disorder. Women over the age of 35 have the greatest risk of having babies with birth defects.

The benefit of screening tests is that they do not pose any risk to the fetus or mother. But screening tests cannot tell for sure if the baby has a birth defect. So, they do not give a "yes" or "no" answer. Instead, screening tests give the odds of your baby having a birth defect based on your age. Women under the age of 35 will find out if their risk is as high as that of a 35 year old woman. For women over age 35, screening tests will help them find out if their risk for their age is higher or lower than average.

If screening tests or maternal age indicate an increased risk for a genetic birth defect, we will arrange referral for a comprehensive ultrasound with genetic counseling. Based on their risk assessment, further diagnostic testing may then be recommended.

Some common screening tests used during pregnancy include:

#### Targeted ultrasound

The best time to receive this test is between 18 and 20 weeks of pregnancy. Most major problems with the way your baby might be formed can be seen at this time. But some problems like clubbed feet and heart defects can be missed on ultrasound. The ultrasound may also be able to see if your baby has any neural tube defects, such as spina bifida. This test is not the most accurate for finding out whether your baby has Down syndrome. Only 1 in 3 babies with Down syndrome have an abnormal 2nd trimester ultrasound. In most cases, gender can be determined by ultrasound.

#### Maternal serum marker screening test

This blood test can be called by many different names including multiple marker screening test, triple test, quad screen, and others. We offer this test between 15 and 20 weeks of pregnancy. It checks for birth defects such as Down syndrome, trisomy 18, or open neural tube defects. A

sample of blood is checked for 4 chemicals: alpha-fetoprotein (AFP) (made by the liver of the fetus), estriol, human chorionic gonadotropin (hCG) and inhibin-A.

Higher levels of AFP are linked with open neural tube defects. In women age 35 and over, this test finds about 80% of fetuses with Down syndrome, trisomy 18, or an open neural tube defect. In this age group, there is a false positive rate (having a positive result without actually having a fetus with one of these health problems) of 22%. In women under age 35, this test finds about 65% of fetuses with Down syndrome, and there is a false positive rate of about 5%.

### **Nuchal translucency screening (NTS)**

This new type of screening can be done between 11 and 14 weeks of pregnancy. It uses an ultrasound and blood test to calculate the risk of some birth defects. An ultrasound is used to check the thickness of the back of the fetus' neck. Additionally, blood levels of a protein called pregnancy-associated plasma protein and a hormone called human chorionic gonadotropin (hCG) is checked. This information is then used to tell if the fetus has a normal or greater than normal chance of having some birth defects.

In an important recent study, NTS found 87% of cases of Down syndrome when done at 11 weeks of pregnancy. When NTS was followed by another blood test done in the second trimester (maternal serum screening test), 95% of fetuses with Down syndrome were identified.

Like all screening tests, the results are sometimes misleading. In 5% of women who have NTS, results show that their babies have a high risk of having a birth defect when they are actually healthy. This is called a false positive. To find out for sure if the fetus has a birth defect, NTS must be followed by a diagnostic test like chorionic villus sampling or amniocentesis.

If you are interested in NTS, talk to your doctor. We can refer you for testing. You should also call your insurance company to find out if they cover the cost of this procedure since it is new. NTS allows women to find out early if there are potential health problems with the fetus. This may help them decide whether to have follow-up tests.

### **Cystic fibrosis carrier screening**

Cystic fibrosis is a life-long illness that is usually diagnosed in the first few years of life. The disorder causes severe problems with digestion and breathing. Cystic fibrosis does not affect intelligence or appearance.

The purpose of CF carrier testing is to see if a couple is at increased risk for giving birth to a child who will have CF. Cystic fibrosis carrier testing is a laboratory test done on a sample of your blood or saliva. If testing shows that a couple is at high risk, additional testing can be done on the developing baby to see whether or not it will have CF.

You could be a carrier of CF even if no one in your family has CF and even if you already have children without CF. About one of every 30 white people (about 3 in 100 or about 3%) carries the changed gene. If your family background is not white, your chance of being a carrier is less than 1 in 30.

Cystic fibrosis cannot be treated before birth. The purpose of having this information about your developing baby is so you can prepare yourself to care for a child with special health care needs or allow time to terminate the pregnancy if desired.

The American College of Obstetricians and Gynecologists (ACOG) recommends that health care providers make the CF carrier screening test available to all couples. Deciding whether or not to have the test is your own personal choice. The cost of testing is covered by some insurance, but not all. We recommend checking with your insurance company before deciding if you want testing. Carrier screening is available in our office upon request.

## **Diagnostic Tests**

Diagnostic tests can give definite "yes" or "no" answers about whether your baby has a genetic defect. But, unlike screening tests, they are invasive and come with a risk of miscarriage. Amniocentesis and chorionic villus sampling (CVS) are the two most commonly used. Both tests are more than 99% accurate for finding these problems. These tests also can tell you your baby's gender. In most cases, results take about two weeks.

### **Amniocentesis**

This test is performed by a specialist in pregnancies of at least 16 weeks. It involves inserting a thin needle through your abdomen, into your uterus, and into the amniotic sac to take out a small amount of amniotic fluid for testing. The cells from the fluid are grown in a lab to look for problems with chromosomes. The fluid also can be tested for AFP. About 1 in 200 women have a miscarriage as a result of this test.

### **Chorionic villus sampling (CVS)**

This test is performed by a specialist between 10 and 12 weeks of pregnancy. A needle is inserted through your abdomen or through a catheter in your cervix in order to reach the placenta. A sample of cells is taken from the placenta. These cells are used in a lab to look for problems with chromosomes. This test cannot find out whether your baby has open neural tube defects. About 1 in 200 women have a miscarriage as a result of this test.

If you have any further questions or would like more information on any of the above topics, please speak with your doctor.

Please answer the following questions by circling the appropriate response. These questions will help us further assess your risk for birth defects and guide necessary recommendations.

- |   |     |    |
|---|-----|----|
| 1. Will you be age 35 or older at the time of your delivery?  | YES | NO |
| 2. Is the father of the baby age 55 or older?   | YES | NO |
| 3. Do you or the father of the baby suffer from any serious diseases?   | YES | NO |
| 4. Do either you or the baby's father have any birth defects or inheritable diseases?   | YES | NO |
| 5. Is there any history in the family of birth defects or inheritable diseases?   | YES | NO |
| 6. Is there any history in the family of Down's syndrome or mental retardation?   | YES | NO |
| 7. Is there any history in the family of babies born with abnormal development of the spinal cord or brain?   | YES | NO |
| 8. Is there any history of neural tube defects, such as spina bifida?   | YES | NO |
| 9. Have you been exposed to any infectious diseases, such as the measles, during this pregnancy?  | YES | NO |
| 10. Have you received any vaccinations recently?  | YES | NO |
| 11. Have you received any x-rays or taken any medications during this pregnancy? If yes, please list medications and dates take.                        | YES | NO |
| 12. Have you or the father of the baby ever had herpes or any sexually transmitted infections?  | YES | NO |
| 13. Did your mother take any hormonal medication such as DES or estrogen while she was pregnant with you?   | YES | NO |
| 14. Are either you or your husband of Jewish descent? If so, have you been tested for Tay-Sach's disease or Cystic Fibrosis?                            | YES | NO |
| 15. If you are black, have you and your husband been tested for sickle cell disease?  | YES | NO |
| 16. Do you smoke? If yes, how much? _____   | YES | NO |
| 17. Since becoming pregnant, have you consumed any illegal substances, such as cocaine, crack, marijuana, methamphetamines or any other drugs of abuse? | YES | NO |

Signature \_\_\_\_\_ Date \_\_\_\_\_

**Johnson County OB/GYN, Chrtd.**

\_\_\_\_ Yes, I have received information on Prenatal Tests and Procedures.

\_\_\_\_ Yes, I have received a Prenatal Guide (Red Book).

\_\_\_\_ Yes, I have received the hospital registration information.

Signature\_\_\_\_\_

Date\_\_\_\_\_