Carolinas Medical Center
Women's Institute
Uncompromising Excellence. Commitment to Care.
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Prenatal Genetic Testing: Chorionic Villus Sampling (CVS) and Amniocentesis

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We are pleased that you have chosen the Center for Maternal and Fetal Care at Carolina Medical Center's Women's Institute for your healthcare needs. Our staff has provided specialty services to women and couples of reproductive age for more than 20 years. If you are considering prenatal genetic testing, we have prepared the following information to answer some frequently asked questions.

Who is a Candidate for Prenatal Genetic Testing?

Deciding whether or not to have prenatal genetic testing is personal. Some of the common reasons for considering prenatal genetic testing include:

1. Maternal age – the chance for having a baby with a chromosome condition, such as Down syndrome, increases as a woman's age increases.
2. Prenatal screening tests have indicated a baby has an increased chance for having a neural tube defect (such as spina bifida), Down syndrome, trisomy 18 or trisomy 13.
3. A parent is a carrier of a chromosome rearrangement (translocation) or he/she has had a child with a chromosome condition.
4. One or both parents carry a gene for an inherited condition, or there is a family history of an inherited condition.
5. A baby has one or more ultrasound findings of a genetic condition.
6. A pregnant woman has a medical condition, such as diabetes, that may affect a baby's development.

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What are the Tests?

Prenatal genetic testing includes both screening tests and diagnostic tests. Screening tests can determine whether a baby has a higher or lower chance of having certain conditions, while diagnostic tests can determine whether a baby *does or does not have* certain conditions. However, no form of testing can detect all genetic conditions. The benefits, risks, and limitations of each kind of test should be carefully considered.

	Timing of Test (weeks after last menstrual period)	**Screening Test** (determines a condition *may or may not* exist)	**Diagnostic Test** (determines a condition *does or does not* exist)	**Does the test have any risks?**
Nuchal translucency screening	11-14	X		No
Maternal serum screening	15-21	X		No
Chorionic Villus Sampling (CVS)	10-13		X	Yes
Amniocentesis	15-20		X	Yes

The Screening Tests

Nuchal translucency (NT) screen – Includes an ultrasound measurement of the skin on the back of the baby's neck AND measurement of pregnancy hormone levels in your blood. This screening test can be done between 11 and 14 weeks after the last menstrual period. The results indicate whether the baby is at higher or lower *chance* for having Down syndrome, trisomy 18, or trisomy 13. Your doctor's office may or may not be able to do this screening test. If it cannot be done at your doctor's office, it can be done at our office.

Maternal serum screen (alpha-fetoprotein (AFP) screen, "triple/quad" screen) – Blood is drawn from your arm at 15-21 weeks after the last menstrual period to measure pregnancy hormone levels. The results indicate whether the baby is at higher or lower *chance* for having spina bifida, Down syndrome or trisomy 18. This screening test can be done at your doctor's office.

Level II ultrasound – Provides a detailed look at the baby's organ development and growth. The types of problems that can be detected by ultrasound depend on how far along you are in the pregnancy and other factors, such as your weight and how much amniotic fluid is around the baby. Ultrasound is not capable of detecting all birth defects and/or all genetic conditions.

The Diagnostic Tests

The diagnostic tests are chorionic villus sampling (CVS) and amniocentesis. Most women who choose diagnostic testing have only CVS or only amniocentesis, not both. These tests must be performed by specially trained physicians, such as our Maternal Fetal Medicine (MFM) doctors at the Women's Institute.

If you are considering having one of these tests, you will meet with a genetic counselor on the day of your appointment to review your reason for referral and your family and pregnancy histories. Detailed information will be provided about the benefits, risks and limitations of each test you are considering. You will have plenty of time to ask questions. If you change your mind and do not want to have these tests, you may do so. If you think it would be helpful to schedule a separate genetic counseling appointment prior to the day of the test, please inform your doctor of this preference.

Chorionic Villus Sampling (CVS)

CVS is an outpatient procedure where a small sample of tissue called chorionic villi is removed from the developing placenta for genetic study. The CVS procedure is ideally performed between 10 and 13 weeks after the last menstrual period. Because the placenta and the baby develop from the same fertilized egg, the cells of the chorionic villi and the cells of the baby are expected to have the same genetic makeup. The benefit of CVS is that it can be performed early in pregnancy so results and information are available early in pregnancy.

If you decide to have a CVS, you will have an ultrasound to determine if the CVS can be performed. Sometimes CVS cannot be performed due to placental position, maternal factors or placental bleeding. There are two ways to perform CVS: transcervically (through the cervix) and transabdominally (through the abdomen). The MFM doctor determines the easiest and safest way to do CVS based on the ultrasound findings.

If transcervical CVS can be performed, a speculum will be placed in your vagina (the same way a Pap smear is done). After the vagina and cervix are cleansed with betadine (please tell us if you have an allergy to iodine or shellfish), a small flexible catheter (thin tube) is inserted through the cervical opening to the uterus, and then into the chorionic villi. Most women say this feels very similar to having a Pap smear. A small sample of the chorionic villi is removed by aspiration (suction). If enough chorionic villi are not obtained on the first attempt, a second attempt may be suggested. In the rare event that enough chorionic villi cannot be obtained for testing, other testing options will be discussed with you.

If transabdominal CVS can be performed, your abdomen is cleansed with a sterile solution containing alcohol and chlorhexadine (please tell us if you have an allergy to these substances). Using the ultrasound for guidance, a thin needle is inserted by the MFM doctor through your abdomen and into the chorionic villi. Then a small sample of chorionic villi is removed. It is not uncommon to feel some cramping during transabdominal CVS.

What happens after CVS?

Processing of the chorionic villi will begin shortly after the sample is obtained. The cells of the chorionic villi will be used for chromosome testing and/or other genetic testing you choose to have based on your particular needs. Chromosome results are usually available within 10-14 days after CVS. The timing of results of DNA or biochemical testing performed in cooperating laboratories may be different (your genetic counselor will tell you when you should expect the results). Your genetic counselor will notify you of the test results and a written report will be sent to your doctor.

The accuracy of chromosome testing is greater than 99 percent. However, in about two to three percent (two to three out of 100) of CVS results, both normal and abnormal cells are found (known as mosaicism). When mosaicism is found, follow-up testing through amniocentesis (see below) is offered to try to determine if the abnormal cells are confined to the placenta or whether they are present in the baby as well. The accuracy of DNA and

--the placenta or whether they are present in the baby as well. The accuracy of DNA and biochemical genetic testing varies based on a number of factors that your genetic counselor will review with you. In most cases, test results will be normal. If an abnormality is found, your genetic counselor is available to meet with you again to review the results and their implications in a supportive, caring environment.

In most cases, a follow-up ultrasound is recommended at 17-18 weeks after the last menstrual period to check the baby's growth and development since normal test results cannot rule out all birth defects. You may also choose to have AFP screening in the mid-second trimester (16-18 weeks after the last menstrual period) to screen for open neural tube defects such as spina bifida (see section on Screening Tests above). AFP screening can be done through your doctor's office.

Amniocentesis
Amniocentesis is an outpatient procedure in which a sample of amniotic fluid is obtained for genetic testing. Amniocentesis for genetic testing is most commonly performed between 15 and 20 weeks after the last menstrual period.

If you choose to have an amniocentesis, an ultrasound is performed to assess the baby's growth and development and to locate an area of accessible amniotic fluid. Then, your abdomen is cleansed with a sterile solution containing alcohol and chlorhexadine (please tell us if you have an allergy to these substances). Using the ultrasound for guidance, a thin needle is inserted by the MFM doctor through your abdomen into your uterus and about two tablespoons of amniotic fluid is withdrawn. This fluid contains skin cells from the baby that can be used for genetic testing. Rarely, the attempt to obtain amniotic fluid on the first try is unsuccessful. If this happens, your options will be discussed with you.

Many women say that they feel pressure or cramping, similar to a menstrual cramp, while the amniocentesis is being performed. You have the option of having your skin numbed with a local anesthetic (lidocaine) before the amniocentesis needle is inserted, but your uterus cannot be numbed.

What happens after Amniocentesis?
Processing of the amniotic fluid will begin shortly after it is obtained. Some of the fluid will be used for testing for spina bifida. The cells in the amniotic fluid will be used for chromosome testing and/or other genetic testing you choose to have based on your particular needs. Chromosome results are usually available within 10-14 days after the amniocentesis. The timing of results of DNA or biochemical testing performed in cooperating laboratories may be different (your genetic counselor will tell you when you should expect the results). Your genetic counselor will notify you of the test results and a written report will be sent to your doctor.

The accuracy of spina bifida testing is 98 percent or greater and the accuracy of chromosome testing is greater than 99 percent. The accuracy of DNA and biochemical genetic testing varies based on a number of factors that your genetic counselor will review with you. In most cases, test results will be normal. If an abnormality is found, your genetic counselor is available to meet with you again to review the results and their implications in a supportive, caring environment.

What are the risks and limitations of CVS and amniocentesis?
Any medical procedure has potential risks. The risk for complications that could lead to pregnancy loss (miscarriage) following CVS or amniocentesis is considered to be about one in 370 (0.27 percent). Following either procedure, you should avoid heavy lifting, strenuous exercise and intercourse for approximately three days. Any fever, cramping, vaginal bleeding similar to menstrual bleeding or vaginal leakage of fluid should be reported to your doctor. Mild spotting after transcervical CVS is not uncommon and it is not typically a sign of problems.

Amniocentesis can be performed in most twin pregnancies, but CVS may or may not be possible in a twin pregnancy. If you have a twin pregnancy, the special risks and limitations of diagnostic testing will be discussed with you.

On rare occasions, the cells obtained from CVS or amniocentesis do not multiply in the laboratory and a test result cannot be obtained. If this happens, your options will be discussed with you.

What should I do before a CVS or amniocentesis appointment?
Before having CVS or amniocentesis, we must have documentation of your blood type and antibody screen from your current pregnancy. These laboratory tests should be done by your

doctor and the results should be sent to our office. If we do not have this information by the time of your appointment, we may have to delay performing the procedure until we have the results. If you are Rh negative, you will be given RhoGam before you leave our office.

If you are planning to have CVS, we ask that you do the following:

1. Have an ultrasound at your doctor's office to confirm that the pregnancy is progressing as expected.
2. If you are taking blood thinners such as aspirin, please call our office at least one week before your appointment for special instructions.
3. Please bring something to drink to the CVS appointment because your bladder needs to be comfortably full for the procedure.

If you are planning to have amniocentesis, a full bladder is not needed.

You may eat your regular meals, and your partner or a friend is welcome to come with you to the appointment. However, we ask that you do not bring your children, if possible. If you wear a belly-button ring, please remove it prior to ultrasound because it can damage our equipment and interfere with our ability to obtain good ultrasound pictures.

Which test is best for me?

There are advantages and disadvantages to any prenatal genetic test. Please realize that no prenatal genetic test can detect all problems. Together with your partner and your doctor, you should decide whether the information provided by these tests is important to have for you and your pregnancy. No two women or couples approach pregnancy-related issues in the same way. We want to provide you with information and support to help you make the choices that are right for you and your family. Prenatal genetic testing, either screening or diagnostic, is always optional.

We hope this information has been helpful. Our primary concern is the welfare of you and your baby, and our goal is to provide you with the most accurate and comprehensive information available. If you need more information, please call one of our offices and ask to speak with a Genetic Counselor.

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