

CHORIONIC VILLUS SAMPLING

What is chorionic villus sampling?

Chorionic villus sampling, or CVS, is a procedure in which a small sample of tissue is removed from the placenta. Genetic testing of this tissue can help you determine the health of your fetus. It is performed in early pregnancy, usually between 10 weeks and 13 weeks.

Who should undergo CVS?

If you are at higher than average risk to produce a child with a genetic condition that may be discovered by this testing you should consider CVS. Common reasons for testing include maternal age over 35 at the time of delivery, a prior birth involving or family history of a genetic condition such as Down syndrome or cystic fibrosis, an abnormal finding on ultrasound examination, one or both parents who carry genetic conditions and maternal concern. A patient for whom the identity of the father is in question can utilize DNA from the CVS as part of a paternity test.

What is the chorion?

The chorion is part of the developing placenta. A small amount of this tissue is removed and the cells derived from the chorion are used for genetic testing.

What does a test of the chorion tell me about the health of the fetus?

The cells from the placenta can give us information about the baby's genes and chromosomes because the baby and the placenta formed from the same fertilized egg. When the egg and sperm came together to start the pregnancy they formed the first cell. This cell divided many times. As those cells divided a portion of them became the chorion and some became the fetus. This is why the chorion has the same genetic make-up as the fetus.

What tests are usually included in the genetic testing of CVS tissue?

The test most commonly performed on CVS tissue is a chromosome analysis, a test for disorders such as Down syndrome. Testing for individual genetic disorders such as cystic fibrosis can be done on the same sample. Even identity testing to determine who is the father can be determined from a sample.

What will I need to do prior to the procedure?

You should read over all the material we send you, including this brochure, the consent form and the precautions that we ask you to follow after the test. Please write down any questions you have for us and bring them with you to the clinic. We will need you to

have a partly filled bladder for the ultrasound examination prior to the procedure and for the procedure.

Your physician will explain the CVS procedure and its risks. We will also explain alternatives such as amniocentesis and no invasive testing, and the risks of those choices. You will then be asked to sign the informed consent form. You may decide not to have the procedure up to the time that we perform the procedure.

How is CVS performed?

There are two ways to perform a CVS, transabdominally and transcervically. The approach that we take depends on the location of the villi in the uterus.

The transabdominal approach uses a needle. For this procedure you rest on the examination table. The skin is cleaned with an antiseptic solution. Under ultrasound guidance the needle is passed through your abdominal wall and the wall of the uterus to reach the villi. A syringe attached to the needle is used to suction out a small amount of villi as the needle passes through the villi.

The transcervical approach uses a thin flexible plastic catheter (a hollow tube). For this procedure you rest on your back in a gynecological examination position. A gynecological speculum is inserted into the vagina to enable the cervix (the opening to the uterus) to be seen. The vagina is cleaned with an antiseptic solution, and then the catheter is carefully guided through the cervix under ultrasound guidance to the villi. A syringe attached to the catheter is used to suction out a small amount of villi as the catheter is withdrawn.

If an inadequate amount of tissue is obtained with the first attempt then a second attempt may be undertaken. In studies of CVS the risk of miscarriage rose appreciably with 3 or more attempts (1).

For some individuals the position of the uterus and the villi within the uterus does not permit testing. Waiting one to two weeks for growth to occur may allow testing in many cases. A few individuals will have to wait for an amniocentesis to be performed later in the pregnancy.

If you have had bleeding during this pregnancy, have been told that you have an abnormality of the uterus such as a bicornuate uterus, or have active genital herpes your physician will discuss whether CVS is right for you.

Which CVS approach will I have?

This depends on the position of the uterus and the position of the villi within the uterus. When the villi are anterior (closer to your front) we generally perform the test transabdominally. When the villi are in the lower part of the uterus and closer to your back we generally perform the test transcervically.

Your physician will use an ultrasound examination at the time of the procedure to determine which way will be easiest based on your anatomy, and will discuss the plan with you at that time.

An ultrasound will be performed after the procedure to check the fetus's heartbeat.

Are transabdominal and transcervical CVS equally safe?

Yes. In a study in 1992 it was found that the risk of miscarriage was equal with these two approaches. It was noticed that vaginal bleeding after the procedure was more common with the transcervical approach (2).

What are my alternatives to CVS?

The primary alternative to CVS is amniocentesis. This is ordinarily performed at about 16 weeks gestation. Early amniocentesis can be performed starting at 13 to 14 weeks. The risks of amniocentesis include miscarriage, leakage of amniotic fluid, injury or infection of the fetus, or infection, internal bleeding or injury to other organs in the mother. Amniocentesis can result in the mother developing Rh incompatibility, a blood incompatibility between the fetus and the mother that can affect future pregnancies. Some individuals learn about all of the different prenatal testing options, and then choose not to have any invasive testing.

How long does CVS take and what does it feel like?

When performed transcervically, the procedure usually takes no more than a couple of minutes and women generally experience discomfort similar to that of a pap smear. When performed transabdominally, the procedure usually takes less than a minute and most women experience a heavy pressure sensation in their abdomen. Anesthesia is not needed.

What are the advantages of CVS?

CVS allows you to know about the health of the fetus early in pregnancy before you are showing. This is important to women who may need to make a difficult decision if test results are abnormal and who prefer not to announce the pregnancy until after they get good results. In contrast, when results come back from amniocentesis you will be 17 to 18 weeks into the pregnancy.

What are the risks of CVS?

CVS carries a slightly higher rate of miscarriage than amniocentesis. The rate of miscarriage is 1 in 100 to 1 in 200 for CVS, compared to 1 in 200 to 1 in 400 for amniocentesis.

There is a small chance that we may find a mixture of normal and abnormal cells in your CVS. In most instances these abnormal cells are confined to the placenta and are not in the fetus. In these situations we perform an amniocentesis to determine whether the abnormal cells are present in the fetus.

CVS cannot detect a spina bifida and other neural tube defects and therefore we recommend that women have maternal serum alpha-fetoprotein (AFP) screening in addition to CVS. This blood test is performed at 15 to 18 weeks of pregnancy.

Although substantially proven not to be true, there was some concern a number of years ago about a small increase in the risk of fetal limb abnormalities associated with CVS (3).

What can I expect after a CVS?

Some individuals experience soreness or mild cramping for a day after the procedure. Spotting or vaginal bleeding may also occur. This bleeding may initially soak a pad or more for the first few days. We expect that the bleeding will decrease over several days. The bleeding will initially be red and then change to brown.

We recommend that you take it easy for two days. If you are on your feet a great deal we encourage you to take off for two days or alter your home and/or work schedule to allow you to sit. We can provide documentation of these recommendations for your employer.

If you have any questions or symptoms that are of concern to you after your CVS, please contact us. We particularly ask you to watch for severe cramping, increasing bleeding, leakage of clear fluid, increasing soreness or tenderness in the abdomen and fever. Should any of these occur, please call the Obstetric clinic at 414-805-4777 and ask to speak to a nurse. If you use this number after hours you will be connected with the physician on call.

When will my CVS test results be ready?

Testing for chromosomal abnormalities such as Down syndrome usually take about 10 to 14 days. Other specialized testing such as DNA testing or biochemical analysis may take longer.

We will call you and your physician or midwife with your results. We will also send them a copy of your results.

If your results are abnormal, we will spend time with you to discuss them and any additional testing that is recommended.

Do normal results from a CVS guarantee that I will have a healthy baby?

More than 95 percent of the high-risk women who have CVS or amniocentesis receive reassuring news -- that their unborn babies do not have the disorders for which they are tested. However, no prenatal tests can guarantee the birth of a healthy baby, since only certain birth defects can be ruled out before birth. Three or four out of every 100 babies have a birth defect.

There is no "test" available that guarantees you will have a healthy baby. Normal CVS and amniocentesis results help to exclude some potential problems, so they are reassuring. Only after your baby's birth will you know just how healthy he or she is.

- 1) Rhoads GG, Jackson LG, Schlesselman SE et al. The Safety and efficacy of Chorionic Villus Sampling for Early Prenatal Diagnosis of Cytogenetic Abnormalities. *N Engl J Med* 1989;320:609-617.
- 2) Jackson LG, Zachary JM, Fowler SE et al. A Randomized Comparison of Transcervical and Transabdominal Chorionic Villus Sampling. *N Engl J Med* 1992;327:594-598.
- 3) Evaluation of Chorionic Villus Sampling: WHO/PAHO Consultation on CVS. *Prenatal Diagnosis* 1999; 19:97-99.

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