

Chorionic Villus Sampling

You are being asked to consider prenatal diagnosis in your current pregnancy. If you are less than twelve weeks pregnant, you have a choice of two different methods of prenatal testing. One method is amniocentesis, traditionally performed between 15 and 18 weeks of pregnancy. In this procedure, a thin needle is inserted through the woman's abdomen and into the uterus. A small amount of amniotic fluid is removed and the cells found floating in this fluid are analyzed.

Chorionic villus sampling (CVS) is the other method used for prenatal testing. This test can be performed even earlier than amniocentesis, thereby offering earlier results. The following will provide you with some basic information on CVS.

Who should consider CVS?

CVS should be considered by women age 33 years or over, individuals who have had a child with a chromosome abnormality, individuals who have a chromosome translocation, and couples at risk for a prenatally diagnosable genetic disease (e.g., hemophilia or sickle cell disease).

CVS may not be appropriate for individuals with a family history of neural tube defects (spina bifida or anencephaly).

When is CVS performed?

CVS is traditionally performed between 10 -12 weeks after a woman's last menstrual period (during the first trimester).

How is CVS performed?

There are two methods for obtaining chorionic villi. For many women, either method can be safely performed. First, an ultrasound evaluation is performed to locate the developing placenta and to date the pregnancy. Often, the placental location determines which method of CVS is more appropriate. There are certain other obstetrical considerations which may make one method preferable, including uterine anatomy and vaginal infections.

- **Transcervical CVS:** A thin catheter (hollow tube) is inserted through the vagina and cervix. Ultrasound is used to guide the catheter to the edge of the developing placenta. A small amount of placental tissue is removed and sent to be analyzed.
- **Transabdominal CVS:** This procedure is similar to amniocentesis. Ultrasound is used to select a site for inserting a needle. After a local anesthetic is given, a needle is inserted, with ultrasound guidance, through the abdomen and placed at the placenta. A small amount of tissue is removed and sent to be analyzed.

Do the procedures hurt?

Most women feel the transcervical CVS is similar to a pelvic exam and only mildly uncomfortable. The transabdominal CVS is very similar to an amniocentesis.

What are the risks of CVS?

Most miscarriages occur early in pregnancy regardless of whether any invasive testing has been performed. A test like an amniocentesis carries a small increased risk of having a miscarriage (about 1/2 of one percent) and the risk after a CVS procedure is slightly higher (about 1/2% to 1%). To help keep this risk as low as possible, women are given precautionary instructions after the procedure.

Several years ago reports appeared in the medical literature regarding women who had undergone CVS and had subsequently delivered infants with limb abnormalities including missing fingers and toes. This association appeared to correlate with, but was not limited to, CVS performed earlier than nine weeks from the onset of their last menstrual period. Recent studies have been performed to provide additional data concerning this issue.

In a recent study conducted by the Center for Disease Control there was no overall increased risk of limb abnormalities among

infants whose mothers had undergone CVS compared to infants whose mothers did not undergo this procedure. However, there was an increased risk for a specific abnormality involving missing fingers or toes (aka "transverse digital deficiency"), with the absolute risk being approximately 1 in 3000 births. This type of birth defect also occurred in infants whose mothers had undergone amniocentesis or who had no form of prenatal testing.

By contrast, an international World Health Organization CVS registry of 138,000 infants born after CVS from 19 medical centers showed no significant difference in the overall frequency or pattern of limb abnormalities compared to the background population frequency.

Based upon currently available information, we conclude the following:

1. CVS is a relatively safe and accurate procedure and is considered an acceptable alternative to genetic amniocentesis.
2. CVS is best performed at 10-12 weeks pregnancy based on the beginning of the last menstrual period.
3. Although it is uncertain whether there is an increased risk of limb abnormalities following CVS, we believe it is important for our patients to understand that this controversy has been raised and that the absolute risk may be in the order of 1 in 3,000 births. Unfortunately, between 2-3% of all children are born with a birth defect regardless of whether or not a woman had prenatal diagnosis during her pregnancy.

Our physicians at the Baylor College of Medicine have been performing CVS since its inception in 1984. It is felt that at our center, as well as others of similar experience, the risk of limb abnormalities (approximately 1 in 3,000) needs to be placed in proper prospective and weighted against the advantages that first trimester prenatal diagnosis offers.

What is analyzed from CVS?

The chorion is the outer membrane of the sac which surrounds the fetus. Early in pregnancy, the chorion is covered by fingerlike projections called villi. In one area, the villi will implant in the uterus to develop into the placenta and the remainder of the villi disappear. The chorionic villi develop from the same fertilized egg as the fetus so the genetic material should be the same. The cells from the villi are cultured (grown) in the laboratory. Chromosome analysis and other special genetic testing can be performed on these cells.

What can be detected through CVS?

CVS detects chromosome abnormalities, such as the kind that causes Down syndrome. If indicated, some special genetic diseases can be diagnosed through DNA or enzyme analysis. The CVS cannot detect neural tube defects such as spina bifida. Therefore, it is recommended that all women who undergo CVS consider having their blood drawn at 15-18 weeks of pregnancy for a spina bifida screening test (maternal serum AFP assay). This can be arranged through your own obstetrician. Many women also wish to pursue a level II ultrasound at 16-20 weeks for further information regarding the physical development of the fetus. No method of prenatal testing (CVS or amniocentesis) can guarantee a baby will be born without birth defects or genetic disease. Prenatal testing can only identify the diagnosable problems for which a couple is known to be at risk. Unfortunately, there are many conditions for which prenatal diagnosis is not yet available.

How accurate are the results from CVS?

Chromosome results from CVS are greater than 99% accurate. Occasionally, these results need to be clarified through blood tests on the parents, ultrasounds, amniocentesis, or fetal blood sampling. Maternal serum AFP screening is greater than 80% accurate in detecting neural tube defects.

When will I receive my test results?

Your CVS results will be available approximately 2 weeks after the procedure. Special studies for other genetic disease may take longer. Maternal serum AFP results are usually available one week after blood is drawn.

How many appointments will I have?

All women will be scheduled for a genetic counseling session to discuss the procedure in detail, determine whether additional genetic testing is appropriate, and ensure that all your concerns have been addressed. Those selecting CVS are scheduled for the procedure between 10-12 weeks. These two appointments may be scheduled on the same day if desired.

What are the advantages of CVS?

The greatest advantage of CVS is timing. Since CVS is performed during the first trimester of pregnancy, the results from the CVS are available earlier (12-15 weeks) than are amniocentesis results (17-20 weeks). If a genetic abnormality is found, families must then decide whether they wish to continue the pregnancy. First trimester pregnancy terminations can be safer,

more easily performed, less expensive, and generally less emotionally difficult than second trimester terminations.

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