



## Chorionic Villus Sampling

**INTRODUCTION** — Chorionic villus sampling (CVS) is a procedure in which small samples of the placenta are obtained for genetic or biochemical analysis to diagnose certain genetic disorders. The placenta is an organ that forms on the inner lining of the mother's uterus through which nourishment and oxygen pass from mother to fetus; the chorionic villi are the tiny units that make up the placenta and have the same chromosomes as the fetus.

CVS can test for diseases such as Down syndrome, Tay Sachs, and cystic fibrosis, and can determine the gender of the fetus. It is generally performed during the first trimester of a woman's pregnancy, at 10 to 12 weeks of gestation. CVS results are available in a few days. Early testing reduces the wait for test results and permits access to pregnancy termination at a safer and more discreet time.

An alternative to CVS, amniocentesis, is a prenatal diagnostic procedure in which a small amount of amniotic fluid from the sac surrounding the fetus is removed and tested for genetic disorders. It is performed later in pregnancy (typically at 15 to 17 weeks).

A pregnant woman may consider CVS testing if:

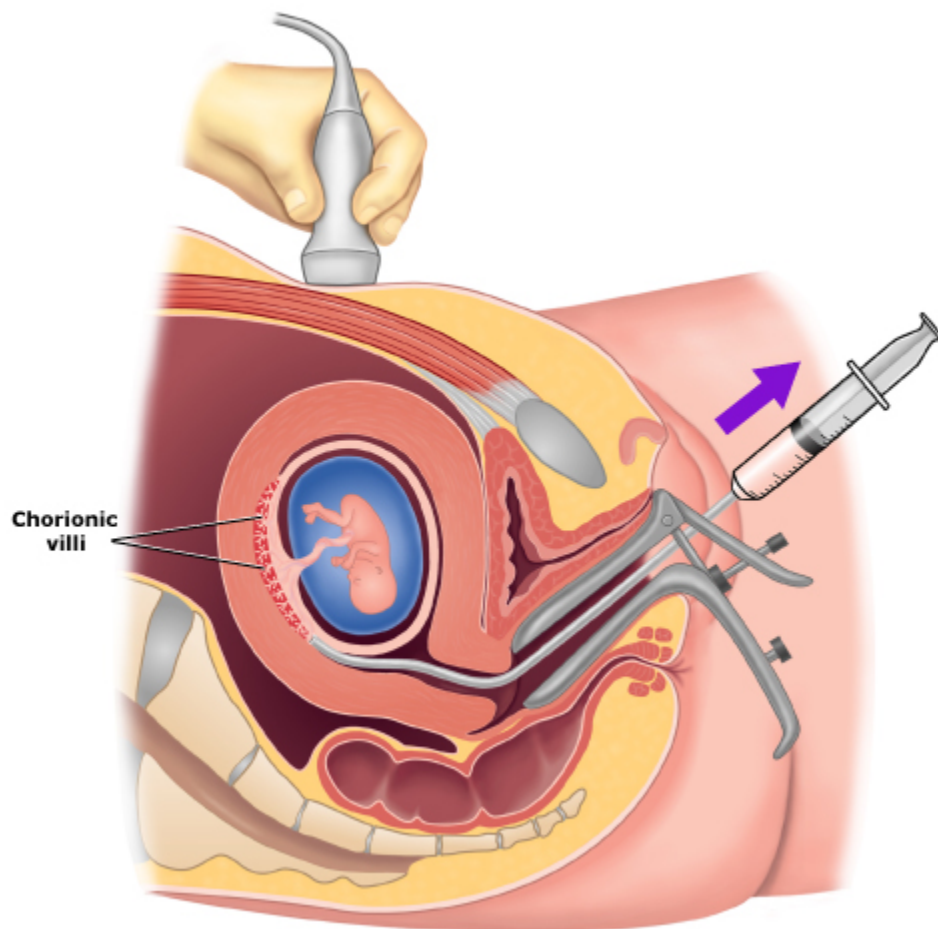
- She desires genetic testing for fetal chromosome anomalies (however, because CVS entails some risk of pregnancy loss, most women consider CVS if the risk of a fetal genetic disease, like Down syndrome, outweigh the risks of the procedure)
- There is a close family or previous pregnancy history of a genetic disorder
- She has had an abnormal screening test result during her first trimester, raising questions about whether the fetus has Down syndrome.

Genetic disorders that can be diagnosed from DNA analysis can be determined with CVS or amniocentesis. Neural tube defects in the fetus cannot be detected by CVS.

**THE PROCEDURE** — An ultrasound is performed before CVS to confirm the gestational age of the fetus. Ultrasound can also determine whether a woman is having twins or multiples, and whether the multiples share a placenta or each has its own. It is important to determine the number of placentas, because each placenta must be sampled separately to have an accurate genetic picture of each fetus.

There are two approaches available to obtain chorionic villi tissue: transcervical (TC) and transabdominal (TA). The choice of the sampling route is based largely upon the placental location.

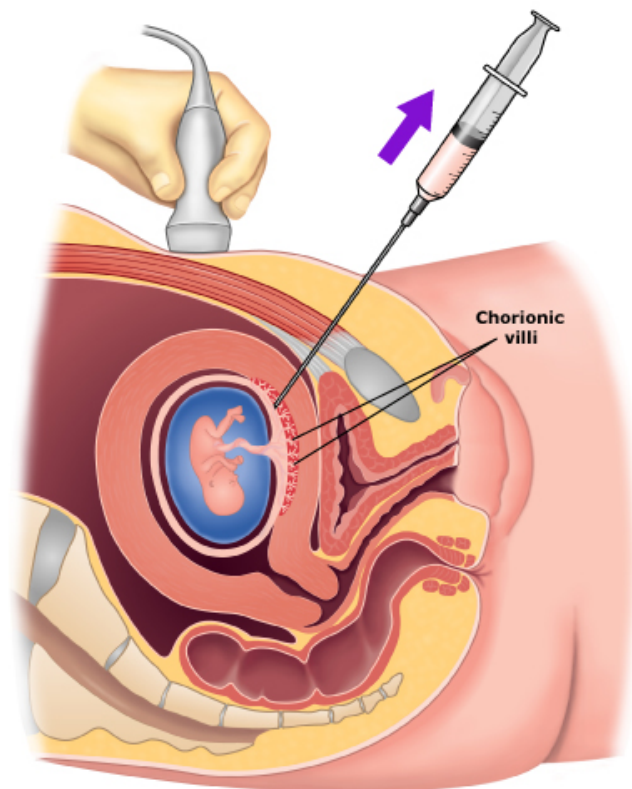
**Transcervical CVS** — In the transcervical CVS (TC CVS) technique, the physician guides a cannula (a small tube used for collection of the chorionic villi) through the vagina into the placenta under ultrasound guidance (Figure 1). After a small amount of tissue is obtained, the cannula is withdrawn.



**Figure 1.** Chorionic Villus Sampling – Transcervical Approach (through the vagina).

**Transabdominal CVS** — In the transabdominal CVS (TA CVS) technique, the physician guides a needle from the patient's abdomen into the placenta under ultrasound guidance (Figure 2). The physician tries to insert the needle so as not to puncture the amniotic sac. Once an adequate specimen is obtained, the needle is withdrawn.

Although most CVS procedures are successful on the first try, second and third attempts are sometimes required to obtain an adequate amount of placental tissue.



**Figure 2.** Chorionic Villus Sampling – Transabdominal Approach.

**Multiple pregnancies** — As mentioned earlier, knowledge of multiple pregnancies and the number of placentas is essential, as it determines the number of samples needed. In addition, the preliminary ultrasound can show where the multiples are located in the uterus to help ensure accurate performance of the CVS.

CVS can be performed through the abdomen, cervix, or a combined approach (abdominal approach for one twin, cervical approach for the other) approach. Only a single sample is required if the twins share a placenta. If there are separate placentas, each is sampled; the procedure is similar to CVS for a singleton pregnancy. DNA analysis is performed for each sample separately.

In CVS of multiple pregnancies, uncertain results occur more frequently than with amniocentesis, which would require further testing. Despite this, CVS offers the advantage of earlier, and therefore safer, selective fetal reduction. Selective reduction may be chosen if the results of the genetic analysis are abnormal.

**DNA analysis** — Rarely, abnormalities are found in the placental cells that are not present in the fetus. In these cases, an amniocentesis is performed to confirm that the abnormalities lie only with the placental cells. This condition is called confined placental mosaicism.

**PRECAUTIONS** — There are a few situations in which transcervical CVS is not recommended:

- Cervical stenosis: a condition where the opening of the cervix is constricted
- Cervical or lower uterine myomas: a myoma is a benign tumor composed of muscle tissue
- Cervical infection
- Severe bending of the uterus that makes the placenta difficult to reach with the cannula

There are few situations that prevent transabdominal CVS, but these include:

- Extreme bending of the uterus, which might cause intestine (between the abdominal wall and the uterus) to be punctured by the CVS needle
- A fetal position that blocks access to the placenta

**COMPLICATIONS** — The most serious complications from CVS are fetal damage and miscarriage. CVS does not affect the risk of stillbirth or infant death.

**Miscarriage** — Studies have shown that CVS increases a woman's risk of miscarriage. For every 100 women who undergo CVS, approximately one woman will have a miscarriage as a result of the procedure. The risk of CVS is higher than that of amniocentesis, after which approximately one in 200 women will have a miscarriage related to the procedure.

There appears to be an even higher risk of miscarriage with the transcervical CVS technique compared to the transabdominal technique. Other factors that further increase the risk of CVS include having the procedure three or more times and having a fetus that is smaller than normal for their age. The physician's skill and experience also play an important role.

**Birth defects** — An increased rate of limb abnormalities have been reported when CVS is performed before nine weeks of gestation. This risk is independent of the expertise of the operator, the route of the procedure (abdominal versus cervical), or the type of needle or cannula used. Therefore, 10 weeks is the generally accepted earliest time for performing CVS procedures.

**Bleeding** — Vaginal spotting after CVS is reported in up to one-third of women; slightly heavier bleeding occurs in fewer than 6 percent of women. Bleeding is more common after the transcervical compared to transabdominal CVS.

**Infection** — Infections as a result of CVS are very rare. Transcervical CVS appears to carry a higher risk of infection than transabdominal CVS.

**Fetal bleeding** — CVS can cause small amounts of fetal blood to be released into the maternal circulation. This can potentially trigger an immune response in mothers who are Rh negative. Rh negative mothers should be given Rhogam (Rh(D) immune globulin) after the procedure to prevent this reaction.

**WHERE TO GET MORE INFORMATION** — Your healthcare provider is the best source of information for questions and concerns related to your medical problem. Because no two patients are exactly alike and recommendations can vary from one person to another, it is important to seek guidance from a provider who is familiar with your individual situation.

This discussion will be updated as needed every four months at the following web site:

[www.uptodate.com/patients](http://www.uptodate.com/patients).

Additional topics as well as selected discussions written for healthcare professionals are also available for those who would like more detailed information.