

Chorionic Villus Sampling

A couple's decision to have prenatal diagnosis (CVS or amniocentesis) should be based on medical indications as well as a personal evaluation of the risks and benefits of the procedure. Genetic counseling can provide information about the risk of having a child with a particular disorder and can help couples decide on whether or not to have prenatal testing.

Chorionic Villus Sampling (CVS) is a procedure performed from 10-14 weeks gestation. If possible, patients who are considering CVS should consider having an ultrasound examination one to two weeks prior to the procedure to confirm gestational age and fetal viability.

CVS involves taking a small sample of the early placenta for genetic analysis. The cells found here should have the same genetic make-up as the fetus and are rapidly dividing. The cells can be used to diagnose chromosome disorders (such as Down syndrome). In many cases where individuals are at risk for a rare genetic disorder based on family history or ethnic background, CVS can be used to determine if the fetus is affected. When specialized genetic testing is being considered, it is recommended to contact a genetic counselor in advance in order to coordinate genetic testing. However, it is important to keep in mind that a normal CVS result does not rule out all causes for mental retardation or birth defects. Unlike amniocentesis, CVS is not able to test for open neural tube defects (such as spina bifida).

- **Procedure**

Prior to CVS, ultrasound examination is required to identify the locations of the uterus, fetus, and placenta. CVS may be attempted through either a transcervical or transabdominal route. The technique performed will depend upon the placental location as well as the physician and patient preference.

For a transcervical CVS procedure, the patient is asked to rest on her back in a position similar to that used for a gynecological examination. A sterile speculum is inserted into the vagina to allow the cervix to be visualized. The vagina is cleansed with an antiseptic solution and a thin plastic catheter with a bendable guide wire is inserted through the cervical opening into the uterine cavity. Under ultrasound guidance, the catheter is used to remove a small portion of the chorionic villi. Most women describe this test as feeling similar to a pap smear.

A transabdominal CVS is often the preferred route when the placenta is located higher in the uterus. Under ultrasound guidance, a thin needle is inserted through the abdominal wall into the uterus. The needle is guided into the placenta to remove chorionic villi. The method used for this procedure is more similar to an amniocentesis, most women compare this feeling to that of having blood drawn from the arm.

In general, patients are able to leave shortly after the procedure. Vigorous activity should be avoided for 24 hours after the procedure. Usual activities may then be resumed.

- **Complications**

The risk for miscarriage associated with CVS is approximately 1/200 (or 0.5%). This rate appears to be the same for both transcervical CVS and transabdominal CVS. If an adequate sample is not obtained or if cell culture is unsuccessful, a repeat CVS or amniocentesis will be offered.

Shortly after CVS was first offered, studies suggested a possible relationship between CVS and limb defects in the fetus. However, it appears that when CVS is performed after 9 weeks gestation (when the critical period for limb development has passed), the risk of such defects is not more than the risk of such defects in the general population.

- **Results**

In 98% of cases, CVS results will be straightforward and conclusive for the fetal chromosome results. Mosaic results are uncommon after CVS, but more frequent than in amniocentesis. Mosaic results mean that some of the cells in the culture have one chromosome arrangement and some of the cells in the culture have a different chromosome arrangement. If you are working with a genetic counselor, they can help interpret your particular results. Less than 1% of CVS patients will be offered an amniocentesis for clarification of a mosaic result.

- **Contraindications**

CVS should not be done in patients with Rh isoimmunization or who have blood born infectious diseases such as hepatitis B or HIV. All patients must have a current blood type and Rh antibody screen prior to the procedure. It is advised that patients undergo HIV and hepatitis B testing as well. Patients who have a history of recurrent or primary vaginal herpes with an active or recent outbreak should provide this information at the time of the consultation prior to the CVS procedure.