Chorionic Villus Sampling Fact Sheet

Palmetto Health USC
OBSTETRICS AND GYNECOLOGY
GENETIC COUNSELING

What is chorionic villus sampling (CVS)?

Chorionic villus sampling (CVS) is a procedure that samples a small portion of the developing placenta, or chorionic villi, in order to learn more about the developing baby (fetus). Because a fertilized egg and sperm form both the fetus and the placenta (afterbirth), sampling the placental tissues allows certain genetic conditions to be detected. CVS is performed in the first trimester of pregnancy between the 10th and the 13th weeks.

Who is offered a CVS?

The most common reason why women are offered CVS is due to maternal age. Women 35 or older at the time of delivery are offered prenatal diagnosis by either CVS or amniocentesis due to the increased chance for the fetus to have a chromosome condition, such as Down syndrome. Chromosomes are the packages in our cells that contain our genetic instructions. Most people have 23 pairs of chromosomes or 46 in all. Extra or missing chromosome material can cause birth defects or developmental delays. For example, an extra copy of the 21st chromosome causes the learning problems and common facial features associated with Down syndrome.

Women who have a positive first-trimester screening test are also offered CVS. There are two blood tests that screen pregnancies in the first trimester to determine if the chance for the fetus to have a chromosome condition may be increased. There may be findings on ultrasound in the first trimester of pregnancy that may also increase the chance for one of these conditions. When a screen shows an increased chance, or is screen positive, for one of these conditions CVS is one of the available tests to confirm or rule out the diagnosis.

CVS may also be offered if a couple has had a previous child with a genetic condition or if the family history suggests an increased chance for a genetic condition for which prenatal testing by CVS is available.

How is CVS done?

CVS can be done in one of two ways. The method used is determined by the physician and depends largely on the physician's experience and on the position of the placenta. In one method, the sample is taken by going through the woman's cervix (transcervical CVS or TC-CVS). An abdominal (on the stomach) ultrasound is used to look at the position of the placenta. A thin tube is inserted through the vagina and cervix to the thick portion of the placenta to take a small sample.

Another method involves sampling the placenta by going through the mother's abdomen. Abdominal ultrasound is again used to determine the best location and a thin needle (guide needle) is inserted through the abdomen (not through the navel) to the placenta to obtain a small sample. In some cases, the procedure may need to be repeated if there is not enough sample to perform an analysis.

Most women who have a CVS have no complications following the procedure. However, with either form of CVS, a woman may experience complications afterwards including spotting (more common after TC-CVS) and cramping (more common after TA-CVS). Following CVS, approximately 1% of women will have significant procedure-related complications that lead to a loss of the pregnancy. This risk is in addition to the 2% to 3% background risk for miscarriage at this point in pregnancy. The sample obtained from CVS is then analyzed in a laboratory. The number and shape of the chromosomes are studied to determine if a genetic condition is present. Typically after 10 to 14 days, a chromosome result is available. If specific DNA or biochemical testing is being offered based on the family history, the amount of time to obtain the result will depend on the test ordered.

Are the results of CVS accurate?

In approximately 98% to 99% of CVS cases, the chromosome results allow for a clear answer about the fetus's chromosomes. The laboratory usually studies 20 cells from each sample. The majority of the time, all the cells studied show the same number of chromosomes and the diagnosis is either negative (normal) or positive for a chromosome condition. Occasionally, some of the cells studied have the normal number of chromosomes, while other cells have an extra or missing chromosome. This mixture of cells is called a mosaic result. Women who have mosaic CVS results are counseled about the possible effects on the fetus and are offered follow up testing. Other ways in which a CVS result might not be accurate include if a mother's cells were accidently sampled instead of the baby's cells, if there is a laboratory error, or if the pregnancy began as twins. Again, these things do not happen very often so that greater than 98% of the time the results are accurate.

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Is CVS safe?

CVS procedures have been performed worldwide since mid 1980s, with the majority of women having good outcomes. CVS is associated with an increased risk for miscarriage that is similar to that of amniocentesis (about 1/200 to 1/100, or 0.5% to 1% above the background rate for miscarriage). It is important to recognize that the background rate of miscarriage between 8 to 12 weeks gestation in pregnancies that do not have CVS is approximately 2% to 3%.

There have been some reports that the risk of a specific type of limb defect called transverse limb defect and oromandibular (mouth and jaw) defect may be increased in pregnancies tested by CVS. The risk for these anomalies is not certain. An analysis by the World Health Organization reports 6 in 10,000 cases of limb defects are due to CVS. This is not significantly different than the occurrence in the general population. It is known, however, that the timing of CVS is important. Procedures performed before 9 weeks gestation had the highest frequency of limb defects. The rate of oromandibular defects is increased when CVS is performed before 7 weeks gestation. Therefore, if the fetus is not yet 10-weeks size by ultrasound dating on the day of the scheduled CVS, it is recommended that another appointment for CVS be made after 10-weeks gestation.

What other testing should be offered after CVS?

Since CVS does not test for openings along a baby's spine (neural tube defect/spina bifida), maternal serum AFP screening for neural tube defects should be offered between 15 to 18 weeks in the pregnancy. In addition, a second trimester ultrasound at 18 to 22 weeks gestation is usually performed to look at the growth and development of the fetus. This will help to screen for other birth defects that CVS cannot test for, such as neural tube or heart defects that are part of any woman's chance when she becomes pregnant. Approximately 3% to 5% of all babies born have some unexpected finding or birth defect.

Is CVS the right test for me?

The decision to have prenatal diagnosis is a personal one. A woman should think about the benefit of knowing the chromosome information about the fetus versus the risk of the procedure. Many people feel that the largest benefit from CVS is the early point in pregnancy that you learn the chromosome information. Amniocentesis is another test that is done later in pregnancy that can detect chromosome conditions and can also detect neural tube defects. It may be helpful to discuss these options and the risks and benefits with your husband/partner, family, physician, and/or genetic counselor. If you have questions about CVS or genetic counseling, an appointment for genetic counseling can be made through your physician.