

Safe Prenatal Tests: CVS and Amniocentesis

*By Philip Buchanan, PhD
and Ginger Norris, MGC, CGC*

There are many types of surgical procedures related to the field of genetics. The two most common surgical procedures performed in prenatal genetics are chorionic villus sampling (CVS) and amniocentesis.

CVS and amniocentesis are procedures offered to pregnant couples who have an increased risk for having a baby with a chromosomal defect, like Down syndrome, or a genetic disease, like sickle cell disease or cystic fibrosis. Couples may be at risk because of their ages, a maternal blood screening test, a finding on an ultrasound examination, exposures to medications or chemicals, or because of their family medical histories. Genetic counseling is recommended to review the couple's specific risk and to discuss the different procedures available to them.

CVS is a medical procedure typically performed by an experienced doctor between 10 and 12 weeks of pregnancy. This procedure removes a small portion of the developing placenta (chorionic villus). The doctor will first determine the gestational age of the pregnancy using an ultrasound examination. The doctor will then obtain the sample of chorionic villus by either using a thin, flexible catheter inserted through the vagina and cervix or by inserting a thin needle through the abdomen. This tissue contains cells from the developing placenta, which has the same chromosomes as the baby. The sample is then transported to the genetics laboratory for chromosome analysis and special studies, if needed.

Amniocentesis is a medical procedure routinely performed after 14 weeks of gestation. It may sometimes be performed as early as 12 weeks of gestation. This procedure removes a small portion of amniotic fluid, which surrounds the fetus. This fluid contains skin cells which have been washed off the baby's skin surface into the amniotic fluid. Because these are skin cells from the baby, they contain the baby's chromosomes. The amniotic fluid sample is then transported to the genetics laboratory for chromosome analysis and special studies, if needed.

Most women do not have complications following CVS or amniocentesis. The possible risk of pregnancy loss after CVS is about 0.5% (1 in 200 chances). The chromosome analysis is about 99% accurate. The possible risk of pregnancy loss after amniocentesis is less than 0.5% (1 in 200 chances). The chromosome analysis is over 99.9% accurate for detecting chromosome abnormalities. The chromosome analysis results are available in an average of 7 days. Some special genetic tests may take longer.

Because CVS cannot test for open spine and skull defects (ONTD) by measuring alpha-fetoprotein, we recommend patients have a maternal serum alpha-fetoprotein (MSAFP) blood test to screen for these birth defects around 15 weeks of pregnancy. The amniocentesis procedure allows the genetics laboratory to measure the AFP directly in the amniotic fluid to detect over 99% of open spine and skull defects. Those skull and spine defects which are too small or are covered with skin (closed) cannot be detected by this test.

Genetic counseling is always recommended to couples considering either CVS or amniocentesis because of their ages, family histories, or abnormal prenatal screening results. A genetic counseling session involves a discussion about the specific risks to the pregnancy, the testing available given the current gestational age, and a detailed family medical history in order to assess any other potential risk to the unborn baby.

Dr. Philip Buchanan is the director of GeneCare Medical Genetics Center. Ginger Norris is a certified genetic counselor at GeneCare. GeneCare provides genetic counseling, screening and laboratory services. For more information, contact their website: www.genecare.com; or call (919) 942-0021; (1-800) 277-4363.