Chorionic Villus Sampling [1]

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Chorionic villus sampling (CVS) is a test used for prenatal diagnosis [5]. Safe to perform at an earlier stage in pregnancy [6] than amniocentesis, CVS is another invasive prenatal diagnostic test that can be performed as early as ten weeks after the woman’s last menstrual cycle. While this test does carry some risks, it is generally very effective at predicting heritable diseases during or soon after the embryonic stage of development [7].

Jan Mohr was one of the first to attempt and describe the CVS procedure in 1968 but the original trans-cervical technique was rejected as unsafe. By 1982 researchers at the University College [8] Hospital of London had improved the method, and by 1984 a relatively safe trans-abdominal technique was introduced. Shortly thereafter CVS became a popular tool for prenatal diagnosis [5].

CVS is a method of obtaining a karyotype [9], a picture of the chromosomes, of the embryo or fetus [10] to detect genetic defects such as Down’s syndrome and cystic fibrosis [11]. The test consists of retrieving a small sample of the chorionic villi in the placental tissue close to the mother’s bloodstream in one of two ways. To collect the sample a tube can be inserted through the vagina [12] and cervix [13], or a thin tube, also known as a catheter, can be inserted through the abdomen and into the uterus [14]. Both methods are guided by the use of an ultrasound [15] monitor on the abdomen to prevent puncturing the amniotic sac [16]. The tissue samples contain cells with the same DNA and genetic information as the embryo, allowing for accurate predictions of the health of the developing baby. The entire procedure often takes less than twenty minutes and recovery time is usually minimal.

While complications are relatively rare, the risks that CVS carries can be substantial. Miscarriages caused by punctured amniotic sacs or other problems with the procedure take place in approximately 1–2% of all cases. The earlier in the pregnancy [8] CVS is performed, the higher the chance of induced miscarriage [17]. Other possible risks include cramping, bleeding, infection, and rhesus sensitization occurring when an Rh negative mother develops antibodies to the fetus’s blood due to Rh protein entering her bloodstream. In very rare circumstances CVS can cause physical abnormalities in the developing baby.

CVS is not a routine procedure, and it is generally only performed on women with a family history of a serious inheritable disease, women with previous pregnancies affected by fetal health problems, and women over the age of thirty-five. CVS is often preferred over amniocentesis as it can be performed before fifteen weeks into the pregnancy [8] and results can be obtained in one to ten days. While the test can indicate the presence of a genetic defect or chromosomal abnormality with great accuracy, often the severity of the disease cannot be determined until after birth. In some instances, however, treatment for the fetus [10] can begin in utero (i.e. for spina bifida [18]). For women whose fetuses test positive for such diseases, early notification allows time to prepare for a child with such a problem or to obtain an abortion [19].

Because of its relative level of safety and 98% accuracy, CVS is the most common invasive prenatal diagnostic procedure performed before the second trimester [20] of pregnancy [8]. The technique is now an indispensable diagnostic tool for prenatal caregivers and worried parents. CVS practices continue to be developed and improved, and with more widespread use its efficacy is likely to improve, reducing the rate of induced miscarriages and other risks to make it a routine and safe procedure for women with at-risk pregnancies.