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## **CVS-Chorionic Villus Sampling**

Chorionic villus sampling (CVS) is a relatively new procedure used to diagnose certain birth defects in the first trimester of pregnancy. The test has been performed regularly since 1982, and many thousands have been performed around the world. The CVS procedure involves inserting a small catheter (tube) through the cervix and into the developing placenta. Prior to the procedure, an ultrasound examination is performed to confirm fetal viability, location of the placenta, and length of gestation. Then, the woman will lie on her back with her feet in stirrups while the vagina and cervix are prepared. The catheter is then put through the cervix and a bit of placental tissue is gently suctioned into a syringe. The procedure is performed under ultrasound guidance and most women experience little discomfort during testing. Once the procedure is completed, the patient may resume normal activity. Both placental and fetal tissues originate from the same cell line and are genetically identical. Thus, by obtaining a tiny sample of chorionic villi from the placenta, one can determine certain genetic characteristics of the fetus.

CVS is performed between ten and twelve weeks from the first day of the last menstrual period. CVS can detect chromosome abnormalities such as Down syndrome and may detect certain genetic conditions when there is a family history of the disease. Couples who may wish to consider CVS include:

- Women 35 years of age and older
- Parents who have had a child with Down syndrome or other chromosome abnormality
- Couples who are known carriers of a chromosome rearrangement
- Couples who have a family history of a genetic condition for which testing is available

Until recently, amniocentesis was the accepted mainstay of prenatal diagnosis. Amniocentesis involves inserting a needle into the amniotic sac surrounding the fetus. Amniocentesis is performed between 14-16 weeks gestation and test results take an additional two weeks. One advantage of CVS over amniocentesis is that test results following CVS may be available within the first trimester of pregnancy. For most couples, this means earlier reassurance.

The potential disadvantages of CVS are that minor complications such as vaginal bleeding or cramping occur more frequently following CVS than amniocentesis. Data suggests that the overall miscarriage rate following CVS is 2-5%. However, we recognize that many miscarriages occur naturally in the first trimester, even without CVS. Therefore, the additional risk for miscarriage due to the CVS, based on recent information, is about 0.8%.

Certain defects of the extremities have been reported in infants whose mothers underwent CVS. This type of limb deficiency is known as transverse limb defects and involve the absence of the distal structures of the limb (those furthest from the trunk). It has been hypothesized that these defects may be caused by a disruption of the vascular system of the limb. The overall risk for transverse limb defects following CVS is approximately 0.03%-0.10% (1/3,000-1/1,000). The risk and severity of the defect seems to be related to the timing of the procedure, with procedures performed before nine weeks associated with limb defects more frequently.

Genetic counseling is recommended prior to the day of the procedure. This will allow the couple to make an informed, unhurried decision regarding options for prenatal diagnosis.

If a couple decides to pursue CVS, an ultrasound examination should be performed at 18 to 20 weeks of pregnancy to evaluate fetal growth and anatomy. Also, the option of maternal serum alpha fetoprotein (MSAFP) screening between the 15th and 18th week of pregnancy should be offered. MSAFP is a blood test which screens for neural tube defects such as open spina bifida and anencephaly. Amniotic fluid AFP levels are evaluated when an amniocentesis is performed, and MSAFP screening is not necessary following an amniocentesis.