



Wilmington Maternal-Fetal Medicine

Prenatal Diagnosis

Chorionic villus sampling (CVS) and amniocentesis (Amnio) are prenatal diagnostic tests. These tests can be performed to detect chromosomal abnormalities, such as Down syndrome. They can also be used to detect certain genetic syndromes. Both are considered invasive procedures and carry a small risk to the pregnancy. It is always a woman's decision to have prenatal diagnosis or not.

Chorionic villus sampling is performed between 11 and 13 weeks of pregnancy. Chorionic villi are small projections on the placenta that contain fetal cells. During a CVS, a small sample of these villi are removed from the placenta. The procedure can be done one of two ways: by having a catheter inserted through the cervix into the placenta (transcervical) or by having a needle inserted through the maternal abdomen and into the placenta (transabdominal). The baby's cells collected by the CVS are sent to a lab to be cultured and be analyzed for chromosomal or genetic conditions. CVS does not provide information on neural tube defects, such as spina bifida. CVS has a small risk of 1/500 for a complication that could lead to a miscarriage.

Amniocentesis is most commonly performed between 16 and 20 weeks of pregnancy. Amniotic fluid, which surrounds the baby, contains cells that are shed by the baby during the pregnancy. During amniocentesis, a small amount of amniotic fluid is withdrawn from the sac surrounding the baby under ultrasound guidance. The baby's cells collected in the amniotic fluid are sent to a lab to be cultured and be analyzed for chromosomal or genetic conditions. Amniocentesis also screens for neural tube defects, such as spina bifida. Amniocentesis has a small risk of 1/800 for a complication that could lead to a miscarriage.

The most common reasons expecting parents consider CVS or amniocentesis are:

- Mother will be 35 years or older at the time of delivery
- Mother or father had a previous child with a chromosomal abnormality
- Mother or father are a carrier for a chromosomal rearrangement
- Mother and/or father are carriers of a genetic condition
- Mother had an abnormal maternal serum screen or abnormal NIPS
- Abnormalities were found on fetal ultrasound
- Family history of a genetic condition

Our genetic counselors are available to answer any questions about prenatal diagnosis.