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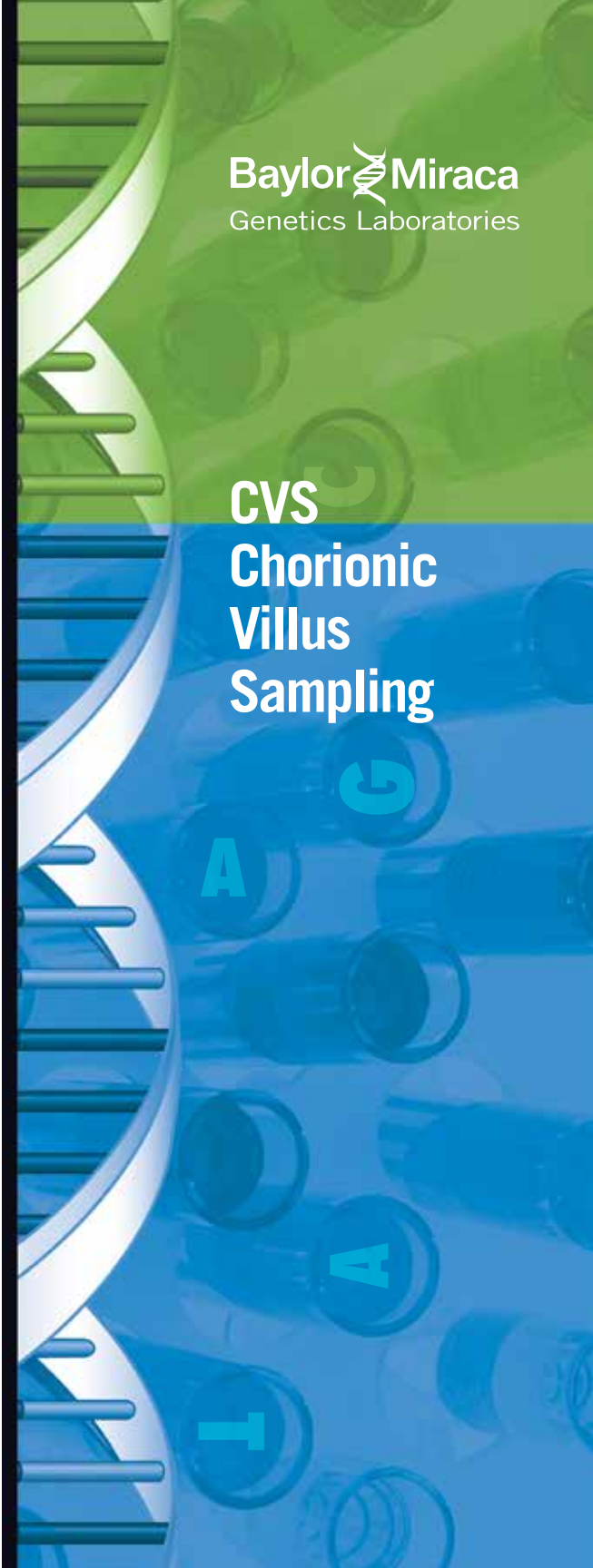
CVS Chorionic Villus Sampling



You are being asked to consider prenatal diagnosis in your current pregnancy. Chorionic villus sampling (CVS) is available as a method of prenatal testing for women who are less than 13 weeks pregnant. This test can be performed earlier than amniocentesis, which is usually performed between 15 and 20 weeks, thereby offering results earlier in the pregnancy. This brochure is to provide you with some basic information on CVS. A separate brochure is available which describes amniocentesis.

Who should consider CVS?

CVS should be considered by women age 35 or older at the time of delivery, individuals who have had a child with a chromosome abnormality, individuals who have a chromosome translocation, and couples at risk for a prenatally diagnosable genetic disease (e.g., hemophilia or sickle cell disease). CVS is not appropriate for individuals with a family history of neural tube defects (spina bifida or anencephaly).

When is CVS performed?

CVS is traditionally performed between 10 - 13 weeks after a woman's last menstrual period (during the first trimester).

How is CVS performed?

There are two methods for obtaining chorionic villi. For many women, either method can be safely performed. First, an ultrasound evaluation is performed to locate the developing placenta and to date the pregnancy. Often, the placental location determines which method of CVS is more appropriate. There are certain other obstetrical considerations which may make one method preferable, including uterine anatomy and vaginal infections.

TRANSCERVICAL CVS: A thin catheter (hollow tube) is inserted into the vagina and through the cervix. Ultrasound is used to guide the catheter to the edge of the developing placenta. A small amount of placental tissue is removed for analysis.

TRANSABDOMINAL CVS: This procedure is similar to amniocentesis. A site is identified on the woman's abdomen and with ultrasound guidance a needle is inserted through the abdomen and placed at the placenta. A small amount of tissue is removed for analysis.

What are the risks of CVS?

Most miscarriages occur early in pregnancy regardless of whether any invasive testing has been performed. A test like an amniocentesis carries a risk for complications, including miscarriage, of about 1 in 200 (0.5%). The risk after a CVS procedure is slightly higher (about 1%). To help keep the risk as low as possible, women are given precautionary instructions after the procedure. Based upon currently available information, the following can be concluded:

1) CVS is a relatively safe and accurate procedure and is considered an acceptable alternative to amniocentesis.

2) CVS is best performed at 10–13 weeks pregnancy based on the beginning of the last menstrual period.

3) CVS is now performed only during or after the 10th week of pregnancy. This is because in 1991 there were several reports of babies with missing or shortened fingers or toes born to women who had undergone CVS before the 10th week of pregnancy. Recent studies suggest that CVS after 10 weeks of pregnancy poses a small or no increase risk for these limb defects. Although it is uncertain whether there is an increased risk of limb abnormalities following CVS, the absolute risk may be the order of one in 3,000 births.

What is analyzed from CVS?

The chorion is the outer membrane of the sac which surrounds the fetus. Early in pregnancy, the chorion is covered by finger-like projections called villi. In one area, the villi will implant in the uterus to develop into the placenta and the remainder of the villi disappear. The chorionic villi develop from the same fertilized egg as the fetus so the genetic material should be the same. The cells from the villi are cultured (grown) in the laboratory. Chromosome analysis and other special genetic testing, when indicated, can be performed on these cells.

What can be detected through CVS?

CVS detects chromosome abnormalities, such as the kind that causes Down syndrome. If indicated, some genetic diseases can be diagnosed through DNA or enzyme analysis. CVS cannot detect neural tube defects such as spina bifida. Therefore, it is recommended that all women who undergo CVS consider having their blood drawn at 15-18 weeks of pregnancy for a spina bifida screening test (maternal serum AFP assay). This can be arranged through your obstetrician. Many women also wish to pursue a level II ultrasound at 18-20 weeks for further information regarding the physical development of the fetus. New testing called prenatal chromosomal microarray analysis (CMA) allows for detection of additional chromosome abnormalities not detectable through standard chromosome analysis. See the prenatal CMA brochure for more details about this testing.

No method of prenatal testing can guarantee a baby will be born without birth defects or genetic disease. It is also important to remember that 2-3% of all children are born with a birth defect regardless of whether or not a women had prenatal testing during her pregnancy. Prenatal testing can only identify the diagnosable problems for which a couple is known to be at risk. Unfortunately, there are many conditions for which prenatal diagnosis is not yet available.

How accurate are the results from CVS?

Chromosome results from CVS are greater than 99% accurate. Occasionally, these results need to be clarified through blood tests on the parents, ultrasounds, amniocentesis, or fetal blood sampling.

When will I receive my test results?

Your CVS results will be available approximately 2 weeks after the procedure. Special studies for other genetic diseases may take longer.

What are the advantages of CVS?

The greatest advantage of CVS is timing. Since CVS is performed during the first trimester of pregnancy, the results from the CVS are available earlier (12-15 weeks) than are amniocentesis results (17-22 weeks).

How Do I Learn More?

We hope this brochure answers some of your questions about CVS. All the information can be discussed in greater detail during a genetic counseling session, thus it is helpful to schedule an appointment for genetic counseling prior to the CVS procedure. The genetic counselor/health-care provider will not only discuss the procedure in greater detail, but will also determine whether additional testing is appropriate and ensure that all of your concerns are addressed. The genetic counselor is also available by phone or in person to follow-up with you after the results of the CVS become available.

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