PRENATAL DIAGNOSIS

UNIVERSITY OF
ROCHESTER
MEDICAL CENTER

STRONG MEMORIAL HOSPITAL

DIVISION OF REPRODUCTIVE GENETICS
and
DIVISION OF FETAL
MATERNAL MEDICINE

CHORIONIC VILLUS SAMPLING (CVS)

CHORIONIC VILLUS SAMPLING

Genetic testing is usually offered to women who have an increased risk of having a baby with a birth defect. Several different tests are available. Meeting with your obstetrician and genetic counselor will help you determine what test, if any, will help.

INDICATIONS FOR CVS

Patients for whom CVS may be appropriate include those who are at an increased genetic risk for one of several reasons. The most common ones include:

1. Maternal age is 35 or older (at expected date of delivery)
2. One parent is a carrier of a chromosomal rearrangement
3. Both parents are carriers of a recessive gene (i.e., Tay-Sachs, etc.)
4. The mother is a carrier of an X-linked gene (i.e., muscular dystrophy)
5. The family has had a previous child with a chromosome abnormality such as Down syndrome

For patients who are at increased risk for neural tube defects (including spina bifida and anencephaly), amniocentesis rather than CVS is recommended for prenatal diagnosis since CVS can not provide any information about development of the neural tube.

BEFORE YOUR CVS

1. A genetic counseling visit will be scheduled to discuss risks and benefits of the procedure.
2. Your obstetrician will be asked to provide the following information: blood type, results of antibody screen done during the current pregnancy, and the results of a dating ultrasound examination done no more than 10 days before your appointment.

THE CVS PROCEDURE

Chorionic Villus Sampling (CVS) involves obtaining a sample of the chorion, a membrane surrounding the gestational sac which will later become the placenta. The chorion is covered by finger-like projections called villi. The chorionic tissue develops from the same fertilized egg as the fetus itself. Since the chorion tissue is growing actively and rapidly in the first three months (1st trimester) of pregnancy, a sample of these cells can provide chromosomes and other material for rapid study in the laboratory.

CVS is performed as an outpatient procedure usually between the 10th and 12th post-menstrual week. It is always immediately preceded by an ultrasound evaluation of the uterus, placenta, and fetus to determine whether the transabdominal or transcervical approach is preferable.

TRANSCERVICAL CVS

The patient lies down, as she does for routine gynecological examinations. After cleansing the vagina, a very thin tube is curved to the shape required to reach the chorion and gently inserted through the cervix (opening of the womb) to reach the point where the thickened chorion has attached to the wall of the uterus. A syringe is used to remove some chorionic tissue. The patient usually feels little discomfort and no anesthesia is required.

TRANSABDOMINAL CVS

The patient lies flat on her back. The abdomen is cleaned and local anesthesia may be used. Using ultrasound for guidance, a needle is inserted through the wall of the mother's abdomen and gently moved through the chorion with the syringe attached to withdraw a small amount of tissue.

AFTER THE PROCEDURE

Limited activity is advised the day of the procedure. If you are Rh negative, an Rh Immune Globulin injection will be given to prevent Rh sensitization.

RESULTS

Chromosome analysis usually is completed within about two weeks. For other types of studies, more time may be required.

You will be notified of the results by either your physician or your genetic counselor.

Approximately 1-2% of patients who have had CVS will have results that may be difficult to interpret and other studies (including amniocentesis) may be recommended to help provide more information.

RISK OF CVS

Any medical procedure has a potential risk. CVS is a technique that has been widely used and is thought to be safe, but there is no guarantee that the procedure will not cause damage to the mother or the fetus, possibly resulting in subsequent miscarriage. There have been pregnancy losses (usually by intrauterine death of the fetus) several weeks after the procedure where the cause and effect relationship with the procedure is not clear. Recently, some cases of limb abnormalities in pregnancies following CVS have been reported. These reports are controversial, but if the association is real, it appears rare. Some bleeding following the procedure may occur in 1/3 of the patients and is usually limited only to spotting. Some women have heavy bleeding, but experience indicates that this has not been related directly to loss of the pregnancy. Untreated infection can lead to serious complications for the mother. Current published data suggest that the risk for miscarriage related to the CVS procedure may be about 1%. This represents an increased risk over amniocentesis of approximately 0.8%. Additionally, the background miscarriage rate at 10-12 weeks of gestation (even after a "normal" ultrasound) is believed to be approximately 3% even if no prenatal procedures are performed.

FOLLOW-UP

A follow-up ultrasound examination of the pregnancy is required at 16-18 weeks of gestation to evaluate the pregnancy and document continuing safety of the CVS procedure. An alphafetoprotein (AFP) screening test on a blood sample is recommended at 16 weeks gestation to screen for open neural tube defects. If you live out of town, these tests may be scheduled through your physician. If you live locally, we can schedule these tests at your physician's request.

Information about the delivery, and the condition of the infant at birth and at approximately one year of age will be requested. This information is necessary to continue to assess the safety and accuracy of the CVS procedure.

If you have any questions, please call your genetic counselor at (716) 275-3304.