Kofinas Perinatal

Providing Care to the Unborn ®

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Chorionic Villous Sampling
Everything you need to know about the procedure

What is chorionic villus sampling?

Chorionic villus sampling (CVS) is a procedure in which a small sample of tissue is removed from the placenta early in a pregnancy. The tissue that is taken has the same genetic information (chromosomes) the baby has.

By analyzing this tissue, genetic specialists can predict early in pregnancy whether the baby is or is not affected by a genetic disorder such as Down syndrome.

The procedure is an alternative to amniocentesis. Amniocentesis, which has been available for many years, is a procedure in which fluid is obtained from the sac surrounding the baby using a needle guided by ultrasound. This fluid contains cells shed from the baby that can be analyzed and provide information regarding genetic defects. CVS is used to test for many of the same disorders that amniocentesis tests for, including Down syndrome. Unlike amniocentesis however, CVS cannot detect neural tube and spinal defects. **However, ultrasound can detect these abnormalities by 16-20 weeks with an accuracy of 100%o when done by an experienced perinatologist.**

When is it used?

You may consider having chorionic villus sampling for one or more of the following reasons:

- You (the mother) will be 35 years old or older on the due date.
- You or your family has a history of pregnancy or child with a chromosome problem, such as Down syndrome.

  -Your family has a history of mental retardation, birth defects, or inherited diseases such as Tay-Sachs, sickle cell anemia, or cystic fibrosis.
- You have had two or more miscarriages or difficulty becoming pregnant.
- You have male relatives with diseases such as hemophilia and muscular dystrophy.
- For the diagnosis of fetal hemoglobinopathies such as sickle cell disease.

- Other genetic conditions as their diagnosis becomes available.

- Abnormal NT Ultrascreen testing.

What happens during the procedure?

CVS can be performed safely any time after 11 weeks gestation and any gestation thereafter. However, at 16 weeks, amniocentesis with a few exceptions is the procedure of choice. This is the earliest time any prenatal invasive genetic testing can be done safely. Amniocentesis is not a choice at 12 weeks due to the high rate of pregnancy loss (as high as 5% at 12 weeks). We perform CVS through the abdomen (trans-abdominal CVS). We have been performing transabdominal CVS since 1986 and have since abandoned the transvaginal approach that has been associated with fetal injury. We are performing CVS at 12 weeks since 1990 and we have experienced 3 pregnancy losses in over 2000 procedures. In addition, there have been no cases of procedure related fetal abnormalities.

A thin but long needle is inserted through the abdomen and uterus into the placenta and a small amount of tissue is withdrawn into a syringe filled with tissue culture media. The procedure is similar to an amniocentesis as far as the mother is concerned but a lot different as far as the fetus is concerned. The amniocentesis causes the perforation of the amniotic sac and this may cause permanent premature rupture of the membranes. The CVS does not. During CVS we withdraw 3-5 mg of placental tissue, which at 12 weeks represent 1/25,000th of the total placental tissue available (the placenta at 12 weeks weighs about 100 Gm or 100,000 mg.

Ultrasound is used with both procedures to locate the chorionic villus and the baby. It also helps to guide placement of the needle with great precision otherwise not attainable. Power color Doppler is used to identify large branches of fetal vessels in order to minimize the risk for placental injury.

What happens after the procedure?

You are advised to avoid strenuous activity for 1 day following the procedure.

Report any unusual symptoms or concerns to Dr Kofinas. You can usually expect a final result within 1 to 2 weeks of the test. For clinically indicated urgent cases, a quick test (Insight) can give results on the most common chromosomal defects and the baby's gender in 48 hours. The final complete reports for all possible chromosomal defects will be completed in 1-2 weeks.

What are the benefits of this procedure?

The main advantage of CVS is that it is performed early in pregnancy compared with amniocentesis, which is not usually done until after the 16th week of pregnancy. This means that with CVS, the results of genetic analysis are usually available before the 14th week of pregnancy. If your baby is found to have an

abnormality, you may wish to terminate the pregnancy. If so, it is safer and easier at that time. It may be also easier on you emotionally if the pregnancy is ended early, before you have started feeling the baby move. Termination later in pregnancy usually requires induction of labor, which is more difficult technically and more emotionally distressing for you and your partner or severely invasive procedures.

What are the risks associated with this procedure?

The biggest risk to the mother is miscarriage after the procedure. Miscarriages in general with chorionic villus sampling take place at about 0.2%, or 1 in 500 cases. This is similar to the risk of amniocentesis. **These statistics are pertinent only to the trans-abdominal CVS as it is performed by us and are different than the statistics pertaining to trans-vaginal CVS, which is more dangerous and has been abandoned by most respected clinicians. We have been fortunate and since we started doing the CVS at 12 weeks we have experienced only three losses in more than 2000 procedures since 1990 that we keep statistics. However, your decision to have or not the CVS should take into account the risk of 1 in 500 or 0.2%, which is the universally accepted risk.**

Other complications that can occur from CVS are bleeding, cramping, and infection. Fortunately, the risk of these is rare, about 1% or less. ***In addition, in a very small number of cases (1% or less), accurate chromosomal analysis may not be possible to obtain from CVS due to local placental mosaicism (mixed normal and abnormal results).*** In such cases, an amniocentesis is usually recommended to clarify the results and evaluate the baby's chromosomes; in the case where the amniocentesis may not resolve the accuracy problem, fetal blood sampling may become necessary. This event has happened to us in four occasions in over 16 years of clinical practice, which makes it very rare.

One of the frequently quoted drawbacks of CVS is that CVS does not help us diagnose spina bifida. However, **the best test for spina bifida is a detailed ultrasound done at 16 weeks and then one done at 22 weeks of your pregnancy**. Amniocentesis alone can detect only open spina bifida, which accounts for 90-95 % of all cases of spina bifida. This means that amniocentesis can miss all cases of closed spina bifida, which represent 5-10% of all spinal defects. Ultrasound by an experienced perinatologist can detect all of these defects.

When should I call the doctor?

Call the doctor immediately if:

- You develop a fever.
- You have any change or worsening of pain or symptoms.
- There is any unusual or bloody drainage from the vagina.

Call the doctor during office hours if:

- You have questions about the procedure or its result.

- You want to make another appointment.

PRECAUTIONS FOLLOWING CVS
Signs to look for:

1. Fluid leaking from the vagina.
2. Bleeding from the vagina.
3. Severe cramping similar to menstrual cramps.
4. Fever.

If you experience any of these signs, get off your feet and call Dr. Kofinas at 718-780-5610 or 516-832-0300. These signs do not mean that you will have a miscarriage; however, your activities should be limited. You should contact our office during regular office hours. You will probably require an ultrasound to ascertain that the pregnancy is not in any serious trouble and the baby is alive.

Provided that you are not experiencing the above-mentioned signs or symptoms, you may return to work the day of the procedure or the following day. You can participate in any of your usual activities with the exception of heavy physical work, which may be resumed within a couple of days.

I _____ have read the above information and all my questions were answered to my satisfaction. I have also received a copy of this document for future reference.

Date:___/___/___ Patient:_____

Date:___/___/___ Witness:_____