PRENATAL DIAGNOSIS VIA CHORIONIC VILLUS SAMPLING
PATIENT INFORMED CONSENT

I, _____________________________, have been referred to the Department of Obstetrics and Gynecology, USC School of Medicine for genetic counseling and prenatal diagnosis by chorionic villus sampling (CVS). CVS involves the insertion of a catheter (small plastic tube) through the cervix (opening to the womb) into the uterus (womb). The catheter will be placed in the area of the developing placenta (afterbirth) and a small amount of the developing placental tissue will be biopsied into the tube. The procedure is performed by an experienced obstetrician between 10 weeks to 13 weeks 6 days gestation.

The following aspects have been explained to me and I understand and accept them:

1. Chorionic villus sampling (CVS) has been performed on over 200,000 pregnancies worldwide. The Department of Obstetrics and Gynecology at USC School of Medicine has thus far performed over 3,000 procedures on continuing pregnancies since 1985. The staff has informed me that, in their opinion, they can perform the procedure of CVS with reasonable expertise.

2. CVS is an accepted method of prenatal diagnosis that is recognized as an alternative to amniocentesis. The safety of CVS has been thoroughly studied and is under continued investigation. Common side effects of CVS may include some spotting or bleeding and light abdominal cramping. These usually resolve within a day or two without further complications. The incidence of amniotic fluid leakage or infection after CVS is less than 0.5% (1 in 200). Rh sensitization, uterine infection, hemorrhage, uterine perforation, and premature labor have been rarely reported.

   CVS is associated with an increased risk for miscarriage that is similar to that of amniocentesis (about 1/200-1/100 or 0.5-1% above the background rate for miscarriage). It is important to recognize that the background rate of miscarriage between 8-12 weeks gestation in pregnancies that do not have CVS is approximately 2-3%.

   There have been some reports that the risk of a specific type of limb defect called transverse limb defect and oromandibular (mouth and jaw) defect may be increased in pregnancies tested by CVS. The risk for these anomalies is not certain. An analysis by the World Health Organization reports 6 in 10,000 cases of limb defects due to CVS. This is not significantly different than the occurrence in the general population. It is known, however, that the timing of CVS is important. Procedures performed before 9 weeks gestation had the highest frequency of limb defects. The rate of oromandibular defects is increased when CVS is performed before 7 weeks gestation. Therefore, if the pregnancy is not yet 10 weeks’ size by ultrasound dating on the day of the scheduled CVS, it is recommended that another appointment for CVS be made after 10 weeks’ gestation. The risk for a limb anomaly (less than 0.06%) or oromandibular defects should be kept in perspective of my risk for a chromosomal or genetic abnormality as well as the approximate 0.5-1% risk for miscarriage.

3. I further understand that any attempt to obtain chorionic tissue may be unsuccessful. No more than two (2) attempts will be made to obtain villus for diagnosis. If I request at any time that the procedure be discontinued, my request will be granted immediately. I realize there are some circumstances which may prevent any attempt at CVS. These may include a “tilted” uterus, the location of the placenta, fibroid tumor, persistent uterine contractions, etc.

   If the sample size or quality of the chromosome preparation is inadequate, a final result may not be available from CVS. The laboratory will make the determination whether the sample received is of sufficient quantity to complete the study. The amount of time to complete the study may take longer when a small amount of villi is obtained. If CVS result is not available, amniocentesis will be offered to complete prenatal diagnosis.
4. I also understand the accuracy of CVS for chromosome analysis is greater than 98%. In some cases, maternal cells from the lining of the womb are unintentionally analyzed with the placental cells (maternal cell contamination), therefore, the chromosome results would not represent fetal (baby) results, but my own. I understand that a blood sample will be obtained from me to be used for comparison with the fetal cells to rule out the possibility of maternal cell contamination if needed.

Approximately 1% of CVS results reveal two or more cell lines with conflicting results (called mosaic results). Amniocentesis will clarify the majority of cases of mosaicism. The possibility of misinterpretation of the chromosome, biochemical, or DNA study does exist. Rare and subtle genetic variations in the fetus such as a chromosome deletion, duplication or translocation or an undiagnosed mutation may remain undetected. With this in mind, I understand that the overall accuracy of prenatal diagnosis via CVS is high; however, the results cannot be guaranteed to be 100% accurate.

5. I understand that this chorionic villi specimen is being sent to ____________________________ for analysis. USC School of Medicine will not perform this test but will serve as contact between me and the laboratory performing the analysis.

When more than one genetic test is indicated based on family history and chorionic villi tissue is not sufficient for testing of both chromosomes and________________, I understand that the priority will be for testing the condition with the highest risk in the pregnancy.

Amniocentesis or other follow-up testing may be necessary if CVS analysis fails or if results are inconclusive.

6. I understand that CVS cannot test for the presence of neural tube defects. I understand that between 15-20 weeks’ gestation, a maternal blood screening test (maternal serum alpha-fetoprotein or MSAFP) along with ultrasound at 18-22 weeks will be available. In pregnancies at an increased risk for a neural tube defect (due to MSAFP results or family history), high-resolution ultrasound and/or amniocentesis may be necessary to test for open neural tube defect in the fetus. These screenings will be arranged through my referring doctor’s office.

7. I understand that if I have genital herpes, I should inform the genetic counselor and the obstetrician prior to the start of the procedure. An active lesion at the time of CVS may risk introducing the virus into the womb. Furthermore, the absence of a lesion does not rule out the presence of the virus. CVS would not be indicated if an active herpes lesion were present. Amniocentesis would be recommended instead of CVS to reduce the risk of passing the infection to the pregnancy.

8. I understand that if I was born with a congenital heart defect such as an atrial septal defect (ASD) or had open heart surgery such as a heart valve replacement, I should inform the genetic counselor and obstetrician prior to performing the procedure. A dose of antibiotics before and following CVS may be recommended.

9. I understand that if I am allergic to Iodine, I should inform the genetic counselor and the obstetrician performing the procedure so that an alternative method for cleansing the vagina will be made available.

10. I understand that if my blood type is Rh negative, I will be given an injection of RhoGam® to prevent sensitization from this procedure as much as possible. I also understand that maximal protection from Rh sensitization will require additional doses later in my pregnancy.
11. The findings of normal chromosome results or of a normal biochemical, molecular or DNA status does not eliminate the possibility that my child might have birth defects and/or intellectual disabilities caused by other disorders for which no tests are performed. Approximately 3-5% of all babies born have a serious birth defect, many of which would not be detected by ultrasound, CVS or amniocentesis.

12. I agree that I have been advised thoroughly regarding alternative means of prenatal diagnosis by amniocentesis. I have read the information provided on chorionic villus sampling and amniocentesis and understand the rationale behind these procedures.

13. I understand that if a twin gestation is confirmed by ultrasound, CVS may not be performed. Instead, second-trimester diagnosis via amniocentesis will be offered. Determination of which procedure is most appropriate will be made on the day of the visit.

14. Following this procedure, I should not do heavy lifting (over 20lb) and should avoid strenuous exercise for the next 72 hours. I should avoid sexual intercourse or anything in the vagina for the next 72 hours. I also understand that some spotting and mild cramping are common side effects after CVS. If any symptoms occur such as heavy bleeding, cramping, abdominal pain, fever, I understand that I should contact the doctor who referred me for the procedure or contact one of the attending physicians at USC Department of Ob/GYN at 803-545-5700.

The indication(s) for prenatal testing in my pregnancy as well as the benefits, risks and limitations of CVS and genetic testing have been explained to me and I have had an opportunity to ask questions. ________________________ has answered my questions. If I have additional questions or concerns, I will discuss these with the obstetrician performing the CVS or the genetic counselor.

☐ I give permission for CVS and prenatal diagnosis. I also understand that I may withdraw my consent at any time prior to performance of the procedure.

☐ I choose not to have the procedure. I understand that prenatal diagnosis by amniocentesis is available to me during the second trimester of pregnancy.

Patient_________________________ Husband/Partner_________________________

Date/Time_______________________ Genetic Counselor_______________________

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