CHORIONIC VILLUS SAMPLING INFORMED CONSENT AND RELEASE

Chorionic Villus Sampling (CVS) for the prenatal diagnosis of genetic disorders is performed between 10-12 menstrual weeks of pregnancy. Chorionic Villi can be obtained by two different methods. The type of procedure to be performed depends upon the uterine position and the location of the placenta. The transcervical method involves inserting a catheter through the vagina and cervix. The catheter is guided by ultrasound to the developing placenta. A small sample of chorionic villi is obtained by suction through the catheter with a syringe. The transabdominal method involves inserting a thin needle through a sterile area of the abdomen (similar to amniocentesis). The needle is guided by ultrasound to the developing placenta and a small sample of chorionic villi is obtained by suction through the needle into a syringe. CVS has been performed in more than 10,000 cases at Mount Sinai Medical Center and in over 200,000 cases worldwide (May, 1999). While the hazard to the mother or fetus is considered to be extremely small, it cannot be guaranteed the procedure will not cause damage to the mother or fetus. The danger to the mother includes a possible risk of uterine perforation or maternal infection. Infection may require hospitalization, treatment with antibiotics, and termination of the pregnancy. Vaginal spotting (i.e., bleeding in small amounts) is a frequent complication of CVS but it usually transient. The danger to the fetus includes fetal demise or miscarriage. There has been controversy regarding the risk of oromandibular and limb abnormalities when CVS is performed in pregnancies prior to ten (10) weeks gestation. However, evidence of this is limited and the overwhelming experience suggests that the risk is extremely small.

CVS PROCEDURE:
1. At the time of the CVS appointment, an ultrasound is first performed. There are obstetrical considerations such as uterine anatomy, placental position, and vaginal infections which may preclude a woman from one or both of the CVS techniques. The perinatologist performing the procedure will make this determination. Alternatively, a patient may have prenatal diagnosis by amniocentesis, performed at 16 to 18 menstrual weeks of pregnancy.

2. Any particular attempt to obtain villi by chorionic sampling may be unsuccessful. A maximum of two insertions of the sampling catheter or needle will be made to obtain a diagnostic sample. If unsuccessful, a repeat visit may be scheduled the following week if the patient desires and the stage of pregnancy is still appropriate for sampling.

3. An attempt to obtain a viable tissue culture form cells of any particular sample of chorionic villi may be unsuccessful or the chromosome preparation may be of poor quality and unusable, or the biochemical study may be unsuccessful due to technical difficulties. A repeat prenatal diagnostic procedure, amniocentesis or fetal blood sampling, may be offered to provide cells to make the diagnosis.
4. Amniocentesis is available for the verification of chromosomal and/or biochemical and/or molecular test results or for the provision of information to aid in interpretation of results from the CVS studies which might be difficult to interpret, due to factors including placental mosaicism and/or maternal contamination.

5. Rh Sensitization: Sensitization, the development of antibodies to fetal blood in Rh negative women, may occur due to CVS. Therefore, Rhogam, a drug which can prevent Rh sensitization, should be given after the procedure.

6. A follow-up high resolution ultrasound will be performed at 18 to 20 weeks gestation to screen for possible development abnormalities including limb defects.

LABORATORY STUDIES:

1. The nature of chromosome and/or biochemical and/or DNA analyses has been explained to me and the accuracy of the test and its limitations have been detailed. I understand that although the likelihood of an incorrect diagnosis or a misinterpretation of the chromosome, biochemical or molecular study is considered to be extremely small, a complete and correct diagnosis of the condition of the fetus based on the karyotype or biochemical analysis or DNA result obtained cannot be guaranteed and that infrequent errors may occur.

2. An assay for alpha-fetoprotein to screen for open neural tube defects will be made available on request. This will be done from a sample of the mother’s blood obtained at approximately 16 weeks of pregnancy. If the results of the maternal serum alpha-fetoprotein studies are elevated twice, then a confirmatory amniocentesis may be recommended.

3. The finding of a normal chromosome constitution or biochemical/DNA status does not eliminate the possibility that the child may have birth defects and/or mental retardation.

4. No test will be performed on my sample other than the one(s) authorized by my doctor.

5. I give consent to have my specimen be used anonymously by the laboratory for the purposes of quality control or for research related to genetic disease. Please check the box below to consent. If you do not consent your sample will be discarded within 2 months of completion of the testing.

   ☐ I agree to have my sample used anonymously for research by the laboratory.______

   Initials

6. I understand that this testing may yield results that are of unknown clinical significance and that parental blood samples may be also be tested to determine whether a specific finding was inherited.

7. The results of my test will be explained to me by a genetic counselor or by my physician who will have the opportunity to discuss my results with a clinical geneticist.

8. I have had the opportunity to have all of my questions answered and undertake professional genetic counseling prior to signing the form. I understand that this consent is being obtained in order to protect my right to have all of my questions answered before testing. I also understand that the results of this testing will become part of my medical record and may only be disclosed to individuals who have legal access to this record or to individuals who I designate to receive this information.
AUTHORIZATION FOR PROCEDURES AND LABORATORY ANALYSES:

1. I authorize the Mount Sinai Genetic Testing Laboratory and Dr. Eddleman and Dr. Stone and their associates at Mount Sinai Medical Center to perform the following procedures and the appropriate analyses:
   a. Chorionic villus sampling (transcervical or transabdominal)
   b. Amniocentesis
   c. Sonography

2. I acknowledge that I have read, or had explained to me the attached information sheet and the nature and purposes of the procedures described above. In addition, I am aware of any benefits reasonably to be expected and the possibility that complications from both known and unknown causes may arise as a result thereof. I have had the opportunity to ask any questions I had, and all were answered to my satisfaction.

3. I voluntarily accept the risks associated with the use of the above-mentioned procedures with the knowledge and understanding that the extent to which they may be effective in my treatment has not been established, that there may be side-effects and complications from both known and unknown causes and that these procedures may not provide a diagnostic result.

4. I understand that I will be followed to term delivery for the purpose of assessing the outcome of the pregnancy.

5. I understand that I am free to withdraw this consent and to discontinue my involvement with the CVS and prenatal diagnostic procedure at any time. The consequences and risks, if any, which might be involved in the event I later decide to discontinue my participation in the prenatal diagnostic procedures have been explained to me.

____________________________  ________________________
(Signature of Patient)           (Signature of Witness)

____________________________  ________________________
(Print Name)                    (Print Name and Title)

____________________________  ________________________
(Date)                          (Date)

(rev. 9/7/2011)