Mount Sinai Genetic Testing Laboratory - Cytogenetics and Cytogenomics Laboratory

Informed Consent for Prenatal Diagnostic Testing of Nine Microdeletion Syndromes by Fluorescence In Situ Hybridization

I, ___________________________________, hereby agree to have prenatal diagnostic testing for nine microdeletion syndromes by fluorescence in situ hybridization (FISH) performed on my fetal sample in addition to routine chromosome studies. The following disorders will be tested for using FISH technology: Angelman syndrome, Cri-du-chat syndrome, DiGeorge/Velo-cardio-facial syndrome, Miller-Dieker syndrome, Prader-Willi syndrome, Smith-Magenis syndrome, Williams syndrome, Wolf-Hirschhorn syndrome and 1p36 deletion syndrome. I understand that fetal cells will be attained through chorionic villus sampling (CVS) or amniocentesis and will be used for the purpose of attempting to determine if my fetus(es) is/are affected with one of these genetic diseases.

I understand that:

1. This testing is performed in conjunction with routine chromosome analysis to detect chromosome abnormalities. The procedure used to obtain fetal cells by chorionic villus sampling or amniocentesis has been explained to me by my genetic counselor/physician ______________________, and I understand the risks and benefits of these procedures.

2. This testing cannot detect all cases of these nine microdeletion syndromes, as the detection rate of these syndromes by FISH testing is not 100%. Therefore, it is possible that my child could have one of these syndromes that was not detected through this testing. Using FISH testing, it is estimated that the detection rates for these nine microdeletion syndromes are as follows: Angelman syndrome (70%), Cri-du-chat syndrome (99%), DiGeorge/Velo-cardio-Facial syndrome (80%), Miller-Dieker syndrome (99%), Prader-Willi syndrome (70%), Smith-Magenis syndrome (99%), Williams syndrome (99%), Wolf-Hirschhorn syndrome (99%) and 1p36 deletion syndrome (95%). For example, this means that 80% of individuals with DiGeorge/Velo-cardio-facial syndrome will be detected by this test, whereas 99% of individuals with Wolf-Hirschhorn syndrome will be detected by this test.

3. By signing this form, I confirm that my genetic counselor/physician has discussed the nature of these nine microdeletion syndromes with me as well as the accuracy of the FISH testing and its limitations. I understand that while results obtained from FISH testing are usually highly accurate, infrequent errors may occur.

4. Even if the results of this testing are normal, my baby(s) could still be born with a birth defect(s) or mental retardation from these or other disorders.

5. If the results are unclear, further testing of parents or family members may be required for clarification.
6. 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.

7. The cytogenetic laboratory may refuse to process a prenatal specimen if there is evidence of maternal cell contamination (the presence of maternal cells in the CVS or amniotic fluid samples). In this situation, a repeat procedure may be recommended/offered.

8. Participation in this testing is completely voluntary. If requested, I will receive a copy of this consent form for future reference. If I change my mind and no longer wish to have my fetal sample tested for the nine microdeletion syndromes, I can contact my genetic counselor/physician to withdraw consent. I will be responsible for charges incurred by the laboratory for the processing of my specimen.

9. Payment is required at the time of submission of the prenatal specimen. This is an optional test that may not be reimbursed by your insurance provider.

10. The cytogenetics laboratory will discard my cultured prenatal specimen within one week of reporting the results of this test.

11. The cytogenetics laboratory is obligated under the Public Health law to retain slides used for cytogenetic diagnosis for 6 years.

I have had the opportunity to have all of my questions answered. If I am signing this form on behalf of a minor for whom I am the legal guardian, I am satisfied that I have received enough information to sign on her behalf. 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.

____________________________________  ____________________
Signature of Person Being Tested (or guardian) Date

____________________________________  ____________________
Witness Date

(rev. 9/7/2011)