Icahn School of Medicine at Mount Sinai

MOUNT SINAI GENETIC TESTING LABORATORY

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Mount Sinai Genetic Testing Laboratory - Cytogenetics and Cytogenomics Laboratory

Informed Consent for Chromosome Analysis, FISH and/or Array Comparative Genomic Hybridization on Abortus Tissue

Studies on tissue from first trimester miscarriages indicate that 50 - 60% of these early losses result from chromosome abnormalities and in second trimester losses 20% result from chromosome abnormalities. Most of these are sporadic in nature, and therefore, do not incur an increased risk for chromosomal abnormalities in future conceptions. In a small percentage of couples (less than 5%), one of the parents carries a rearrangement of his/her chromosomes which predisposes future pregnancies to a higher risk for chromosomal abnormalities.

Chromosome studies on this miscarriage have been recommended by your physician as part of his/her evaluation for the cause of your miscarriage. You should be aware that the tissue may not grow in the laboratory. In this event, we will perform fluorescence in situ hybridization (FISH) with a panel of probes that detects approximately 80% of the abnormalities present in abortus specimens. This testing takes approximately two weeks. Additional FISH tests that identify specific genetic syndromes may also be ordered by your physician. This testing will be reported at the same time as your chromosome results.

If the results of the chromosome analysis are normal, your physician may have requested that we perform array Comparative Genomic Hybridization (array CGH) on the specimen. This testing takes an additional week to complete. In a small number of cases, we will not be able to perform chromosome analysis, FISH or array CGH and will be unable to provide an analysis.

Whole genome array CGH analysis is designed to identify very small (submicroscopic) pieces of genetic material (DNA) that are extra and/or missing and cannot be detected with standard chromosome analysis. These submicroscopic chromosome imbalances may cause birth defects, developmental disabilities, and/or behavioral issues. This testing can identify over 180 known genetic syndromes.

Possible test results:

- **Negative (Normal) result**: This means that array CGH did not detect an extra or missing piece of chromosome material that is associated with a known genetic syndrome or has been reported in the literature to be associated with physical or developmental problems. A normal result does not exclude all genetic conditions.

- **Positive (Abnormal) result**: This means that a loss or gain of genetic material that may be clinically significant has been detected.

- **Indeterminate result:** This means that a loss or gain of material of unclear significance has been detected. Please be aware that some such losses or gains of genetic material may be benign, with no impact on fetal development. Other such losses or gains may cause birth defects or developmental disabilities, the extent of which cannot be determined until after delivery.

Maternal cell contamination studies are required with array CGH testing on abortus tissue. In some cases, additional studies (including parental studies) will be recommended to determine whether or not a detected chromosome imbalance is clinically meaningful and/or was inherited. Most inherited changes are benign. Rarely, however, an inherited change which causes no or minimal issues in a parent may result in significant physical or developmental problems in a child.

The nature of cytogenetic testing has been explained to me and the accuracy of the test and its limitations have been detailed. I understand that while results obtained from this testing are usually highly accurate, infrequent errors may occur. The likelihood of this occurring has been estimated to be less than 1%.

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

No test will be performed and reported on my sample other than the one(s) authorized by my physician.

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._____
Initial

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

I have had the opportunity to have all of my questions answered. 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. 01/24/14)