INDIANA UNIVERSITY
DEPARTMENT OF MEDICAL AND MOLECULAR GENETICS
School of Medicine

Indiana University Genetic Testing Laboratories (IUGTL)
975 W. Walnut St, IB 350, Indianapolis, IN 46202
317/274-2243 (Office) 317/278-1616 (Fax) 317/274-2246 (Lab)

Informed Consent – Prenatal Chromosomal Microarray

Instructions: Please obtain patient signature on the consent form and provide a signed copy to IUGTL to permit testing and processing. If a signed consent is not submitted, the laboratory assumes that the ordering clinician has discussed testing with the patient and obtained the patient's informed consent.

I, (name) _____ , voluntarily request of IUGTL to perform Chromosomal Microarray (CMA) testing on a prenatal specimen from my current pregnancy in an attempt to determine whether the fetus is at increased risk to be affected by a genetic condition. The following points were explained and I understand that:

- Prenatal CMA testing requires cells from the fetus, which has risks associated with obtaining the sample. The volume of sample obtained influences how quickly the laboratory will be able to complete the CMA test and report the results. The test may be unsuccessful on rare occasions. An additional sample may be needed if the test is unsuccessful, or if the sample is damaged in shipment or inaccurately submitted.

- Having the proper amount of genetic material (DNA) in each cell of the body is important for normal growth and development. The DNA is located along the 23 pairs of chromosomes (46 total) in each cell. A basic karyotype analysis can only detect the number of chromosomes in each cell and large structural changes in these chromosomes. CMA is an advanced method of looking at the structure and number of the chromosomes in our body because CMA is able to detect the large changes identified by karyotype, as well as detecting smaller regions of any missing or extra copies (copy number variant, or CNV). These smaller CNVs can also cause abnormal development.

- While the CMA test is very sensitive, not every genetic condition can be diagnosed by a single test. For some conditions, CNVs may represent only a certain percentage of the genetic causes of that condition. Therefore, additional testing methodologies should be appropriately considered.

- When a gain or loss of chromosomal material is inherited from a healthy parent it is much less likely to cause a problem for a fetus or a child. Testing the parents or other family members will tell whether the gain or loss has been inherited or is a new change in the fetus. We often ask for parental blood samples with the prenatal sample to allow parental testing to be done quickly if needed to clarify an observed abnormality.

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- There are several categories of test results that may be reported including:
  - **Normal or Benign/Likely Benign Finding**: A clinically significant abnormality IS NOT detected. However, a normal result does not exclude all genetic conditions, and the clinical diagnosis may still be correct.
  - **Pathogenic/Likely Pathogenic Finding:** A clinically significant abnormality IS detected, which is known to be or likely to be associated with a genetic disorder.
  - **A Result of Uncertain Clinical Significance:** It is not clear if the finding is linked to the fetus's phenotype or associated with disease. Some uncertain findings may be have no impact on fetal development. Others may cause birth defects or developmental disabilities, the extent of which cannot be determined until after delivery. Additional testing of the infant and/or other family members may be recommended to help determine the significance of the result.

- Results of the CMA test may reveal information about yourself, your fetus or your family that is not directly related to the reason for monitoring your pregnancy. This information might relate to diseases with symptoms that may develop in the future in your fetus or possibly yourself or other family members. The test may also identify a different father, the use of an egg or sperm donor, or that the parents are close relatives. It is important to disclose this information before the test to fully understand the potential results.

The CMA test will be performed using materials and protocols developed at the IUGTL and validated by the laboratory. This laboratory is certified by standards set by the Clinical Laboratory Improvement Acts (CLIA) and the College of American Pathologists (CAP).

All of the above has been explained to me to my satisfaction and my signature below attests to the same.

Patient (person being tested):

_____ _____ **Date:** _____
Name of Patient/Authorized Representative Signature of Patient/Authorized Representative

Person obtaining consent:

_____ _____ **Date:** _____
Print Name of Person Obtaining Consent Signature of Person Obtaining Consent