INFORMED CONSENT FOR PRENATAL CHROMOSOMAL MICROARRAY ANALYSIS

Patient Name: ______________________________  Submitting Clinician: ______________________________

The following points to consider have been explained to me and I understand that:

• Chromosomal microarray testing requires cells from the fetus. Collection of the fetal sample has an associated risk.

• Additional fetal or parental samples may be required if the sample is depleted or insufficient or if the sample is damaged in shipment.

• Chromosomal microarray testing detects smaller chromosome gains (duplications) and losses (deletions) than routine chromosome analysis.

• If possible, chromosome imbalances will be confirmed by additional testing, such as chromosome analysis or fluorescence in situ hybridization (FISH). Additional tests may be necessary to fully interpret an abnormal result.

• Testing of parents or other family members may be important to determine whether a gain or loss of chromosome material has been inherited or is present only in the fetus. Submission of parental samples with the prenatal sample can expedite results.

• Genetic testing of parents can reveal non-paternity. Misinformation regarding the true biological relationships of the parents can result in significant reporting errors.

• Accurate interpretation of test results is dependent upon the accuracy of the information submitted with the sample. This includes the fetus’ clinical findings, if any, and the family medical history.

• Chromosomal microarray testing may detect a gain or loss of chromosome material that has uncertain clinical significance. Genetic counseling is highly recommended to explain uncertain findings.

• Chromosomal microarray cannot detect balanced chromosome rearrangements, point mutations, or imbalances of regions not included on the microarray.

• The rate of birth defects and intellectual disability in the general population ranges from 3-5%. Therefore, a normal chromosomal microarray result does not guarantee a healthy child.

• Chromosomal microarray may identify a chromosomal abnormality. Identifying this abnormality may be useful in directing additional care, evaluation, or monitoring for your pregnancy or for your child after delivery.

It is the responsibility of the referring clinician to understand the utility and limitations of the testing ordered, and to educate the patient regarding these issues in relation to her pregnancy management.

Due to the complexity of genetic testing and potential implications of test results, results will be reported only to the ordering provider.

The risks, benefits, and limitations of prenatal microarray testing have been explained to me. I have elected to proceed with prenatal chromosomal microarray analysis on a sample from my pregnancy, submitted in an attempt to determine whether my baby has a chromosome imbalance (gain or loss).

_____________________________                 __/___/_______  _______________________________                 __/___/_______
Patient Signature               Date               Clinician Signature               Date

Use of specimens: Emory Genetics Laboratory retains patient samples indefinitely for validation, educational purposes, and/or research. For cytogenetic and molecular genetic tests, the submitted clinical information and test results are also included in a HIPAA-compliant, de-identified public database as part of the National Institute of Health’s effort to improve diagnostic testing and our understanding of the relationships between genetic changes and clinical symptoms (for information about the database visit https://www.iscaconsortium.org/). Confidentiality of each sample is maintained. Patients may request to withdraw consent for the storage of their sample and/or use of the data by: 1) calling the laboratory at 1-800-366-1502 and asking to speak with a laboratory genetic counselor or 2) visiting our website at www.genetics.emory.edu/egl/opt-out. [ ] Refusal for inclusion in these efforts may be indicated by checking this box.