Informed Consent for NY Clients – Prenatal Microarray Analysis

Instructions: Please obtain patient signature on consent form below. All samples collected in the State of New York must be accompanied by a signed consent form. EGL Genetics (EGL Genetic Diagnostics LLC) is unable to proceed with testing in the absence of a signed consent from the patient. Once completed with signatures of patient/parent and clinician, forward the signed consent to EGL Genetics, either with the transport of the specimen or by fax (see above).

I, (name)__________________________________________________________, voluntarily request of EGL Genetics to perform DNA-based testing on a prenatal specimen from my current pregnancy in an attempt to determine whether the fetus is a carrier of a disease gene or are at increased risk to be affected by a genetic condition. The following points were explained and I understand that:

1. Prenatal microarray 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. Prenatal microarray can identify more than 180 known genetic syndromes.

2. Maternal cell contamination studies are required with all prenatal microarray testing. 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 minimal or no issues in a parent may result in significant physical or developmental problems in a child.

3. This test may be ordered in case of abnormal ultrasound findings, abnormal fetal chromosome findings, history of recurrent miscarriage or stillbirth of unknown cause, previous pregnancy/child with a microarray abnormality, family history of a genetic imbalance, advanced maternal age or parental concern.

4. This is a genetic (DNA-based) test. DNA isolated from the prenatal sample (chorionic villi, amniotic fluid or cord blood) is hybridized to a custom array containing probes across the genome to detect copy number imbalances. FISH analysis or another method, such as G-banding, is used to confirm abnormal findings.

5. I may want genetic counseling before consenting to this test. If the test is positive, I or other family members may wish to have further testing, and I will consult my physician to receive genetic counseling.

6. This analysis can have the following outcomes:

   a. Positive:
      This means that a loss or gain of genetic material that may be clinically significant has been detected.

   b. Negative:
      This means that prenatal microarray analysis 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.
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c. **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.

7. Possible diagnostic errors include sample mix-ups, genotyping errors, rare genetic variants that interfere with analysis and other sources. In addition, due to insufficient specimen size or cell growth, testing may fail to yield results. This testing may yield results that are of unknown clinical significance and that parental blood samples may be also be tested to determine whether the changes were inherited. As a result of parental studies, non-maternity and/or non-paternity may be detected. One may receive a result for which no clinical information exists. By opting to have prenatal microarray testing, one may receive a result relating to an adult onset condition or infertility regarding my fetus.

8. The laboratory does not return the remaining tissue/DNA sample to individuals or physicians; however, in some cases, it may be possible to perform additional studies on the remaining sample. The request for additional studies must be made by the referring physician or other authorized healthcare professional and there will be an additional charge.

9. Remaining DNA samples will be retained in the laboratory in accordance with the laboratory retention policy. Remaining DNA samples may be de-identified and used for internal laboratory purposes with the consent of the patient (see below). The de-identified portion of the sample will not be available for future clinical studies. All original samples (e.g. tissue, blood, etc.) will be destroyed after 60 days of receipt, unless consent is given.

10. I consent to my DNA sample being stored indefinitely and to be used for other laboratory purposes in the future, **PLEASE INITIAL HERE:______________,** I have the right to withdraw this consent at any time, in writing with registered receipt, and any remaining DNA sample will be destroyed.

My signature below acknowledges my voluntary participation in this test and I state that I have been appropriately counseled about the testing process and the different possible outcomes.

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<tr>
<th>Patient/Parent Signature</th>
<th>Date</th>
<th>Printed Name</th>
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<td>Healthcare/Clinician Signature</td>
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