Mount Sinai Genetic Testing Laboratory - Cytogenetics and Cytogenomics Laboratory

Prenatal Array CGH Testing

The Mount Sinai Cytogenetics and Cytogenomics Laboratory is now offering an additional prenatal test which is a microarray-based comparative genomic hybridization called prenatal array CGH. Prenatal 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. Prenatal array CGH can identify over 180 known genetic syndromes.

Possible test results:

- **Negative (Normal) result**: This means that prenatal 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 all prenatal array CGH 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 no or minimal issues in a parent may result in significant physical or developmental problems in a child.

Common reasons for ordering prenatal array include:

- Abnormal ultrasound findings
- Abnormal fetal chromosome findings (such as a chromosome rearrangement or marker chromosome)
- History of recurrent miscarriage or stillbirth of unknown cause
- Previous pregnancy/child with a microarray abnormality
- Parental concern
Specimen Requirements:

- **CVS:** ≥ 8 mg chorionic villi
- **Amniocentesis:** 30 cc clear amniotic fluid (patient optimally <20 weeks gestation; if ≥20 weeks gestation laboratory must be informed)
- **Fetal Blood (PUBS or cord blood):** 2 cc in sodium heparin (green top) tube
- **Maternal blood specimen (5 cc in EDTA (purple top) tube):** must accompany all prenatal specimens in order to perform maternal cell contamination studies.

**Test Cost:** $1629.60 (maximum out-of-pocket expense)

Credit card must be provided at the time that the test is ordered. The patient’s insurance carrier will be billed initially. If the claim is denied, the credit card will then be charged.

**Turnaround time:** approximately 2-4 weeks

Genetic counseling is required prior to ordering prenatal array CGH. Please contact your genetic counselor directly if you would like additional information about this optional test.
Prenatal Array CGH Consent Form

I, ____________________________, hereby request array comparative genomic hybridization (array CGH) testing for my Chorionic Villus Sampling/Amniotic Fluid/Fetal Blood specimen. I have received verbal and written information as described above from my physician or from a genetic counselor and I understand the nature of the array CGH testing that I am about to undergo.

I understand that a prenatal specimen will be taken from me and that the sample will be used in conjunction with chromosome analysis for determining if my fetus is carrying a submicroscopic chromosome abnormality.

No test will be performed and reported on my fetal sample other than the one(s) authorized by my doctor; and any unused portion of my sample will be discarded within 2 months of completion of the testing.

I give consent to have my fetal DNA specimen to be used anonymously by the laboratory for the purposes of quality control or for research related to genetic disease. My sample will be destroyed within 20 years.

☐ I consent to have my sample used anonymously for research by the laboratory.

Initials

The nature of array CGH 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. In addition, due to insufficient specimen size or cell growth, testing may fail to yield results.

I understand that 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. In addition, I understand that I may receive a result for which no clinical information exists. I also understand that by opting to have prenatal array CGH testing, I may receive a result relating to an adult onset condition or infertility regarding my fetus. I also understand that maternal cell contamination studies will be performed on all specimens as part of the testing process, which may result in discovery of non-maternity and/or invalidate the test results due to the presence of maternal DNA.

My prenatal array CGH results 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.

I have had the opportunity to have all of my questions answered and undertake professional genetic counseling prior to signing the form. 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 ____________________________ Date ____________

Witness (Genetic Counselor or Physician) ____________________________ Date ____________

Revised: 9/7/2011