Mount Sinai Cytogenetics and Cytogenomics Laboratory – Postnatal Array CGH Testing

Physician information:

Testing methodology:
Microarray-based comparative genomic hybridization or array CGH is a molecular cytogenetic test that is generally performed after a patient displays a normal chromosome result on peripheral blood chromosome analysis. It is performed on DNA extracted from a peripheral blood specimen and compares the DNA content of a normal individual to the patient DNA content and can detect gains and losses of chromosomes that are not normally visible by karyotype analysis. The patient and control DNAs are fluorescently labeled in two different colors, mixed together and hybridized to a microarray containing DNA targets. The Mount Sinai Cytogenetics Laboratory uses an Agilent oligonucleotide array that spans the entire genome and includes all of the known microdeletion and microduplication syndrome regions with enhanced coverage at regions of disease regions, which are often involved in unbalanced chromosome rearrangements. The results are analyzed using quantitative imaging methods and analytical software to identify loss of copy number (deletion) or gain of copy number (duplication) in the patient. This technology has been validated in our laboratory on many known normal and abnormal specimens that were detected by karyotype or fluorescence in situ hybridization (FISH) analysis.

Reporting of results:
If a known genetic syndrome or pathogenic chromosome abnormality is detected by array CGH, a second test using FISH will be performed to confirm the result and the abnormal result will be reported to you. If a gain or loss of genetic material is detected, but is of unknown clinical significance, it will be necessary to test parental DNA and/or chromosomes to determine whether the gain or loss was inherited. Most inherited changes are not likely to be pathogenic; however, in rare circumstances, a normal parent or parent with mild clinical manifestations may be found to carry a gain or loss of the genome that has produced a more severe phenotype in their child. In addition, a parent may be found to be a carrier of a balanced rearrangement, such as a translocation or inversion that was transmitted to their child in an unbalanced form.

Indications for referral:
Array CGH testing may be ordered on any patient where there is suspicion of a chromosome imbalance when the karyotype is normal. The findings may include dysmorphic features, unexplained mental retardation/developmental delay, multiple congenital anomalies, seizure disorders, behavioural issues and/or learning disabilities or autism. It is also appropriate for patients that are candidates for subtelomere FISH studies or multiple individual FISH tests. It is important to note that the diagnostic capability of array CGH is limited to the detection of gain or loss of genomic material. It will not detect low level mosaicism, balanced rearrangements, specific gene point mutations or rearrangements below the resolution of the array.

References:
Please complete the information requested below.

Age: _______________   Sex:_______________   Race:_______________

Has chromosome analysis been previously performed? yes______ no_______

Check the appropriate findings and describe in the space below.

___ Dysmorphic features (796.4)
Eyes and ears:______________________________
Facial appearance:__________________________
Other______________________________________

___ Multiple congenital anomalies (759.4)
Cardiac defect:______________________________
Urogenital anomalies:________________________
Skeletal anomalies:__________________________
Other______________________________________

___ Developmental Delay (315.9)
___ Seizure Disorder (345.9)
___ Other _________________

Specimen Requirements:
Blood in both EDTA (purple top) and Na Heparin (Green top) tubes: Please include samples for the individual to be tested and both parents. Failure to collect parental specimens may result in delayed test results.

Adult: 10 cc per tube
Child: 4 cc per tube
Infant: 2 cc per tube

For cultured cells from skin fibroblasts, please send 3 - T25 flasks

Turnaround Time:
7 -14 days

Include the following with each sample:

1. Completed and signed test requisition form, informed consent and this form with clinical information, as well as a copy (front and back) of the patient’s insurance card.

2. Patients will be responsible for any co-insurance and in-network deductible if insurance pays for the testing.

3. We will bill all insurance plans for this testing, with the exception of the following: Empire Blue Cross HMO and POS plans, Medicaid, Managed Medicaid plans.

Please have your patient contact the billing department (212-241-8717) for CPT codes and insurance claim forms/receipts.

Please call 212-241-7518 for specimen pick-up by messenger service (in Manhattan only). Or send specimen for same day or overnight delivery to:
Mount Sinai Genetic Testing Laboratory
1428 Madison Avenue
Atran Laboratory Building
Room AB2-32
New York, NY 10029