GENETICS LABORATORIES GENETICS LABORATORIES

Baylor Miraca
Genetics Laboratories

Chromosomal Microarray Analysis (CMA)

Chromosomal Microarray Analysis (CMA) using Array Comparative Genomic Hybridization (aCGH) is available through the Baylor Miraca Genetics Laboratories (BMGL). We are one of the first labs to offer CMA for clinical application, and we remain a leader in the implementation of this new technology. The 180k oligo array contains probes for all the known microdeletion/duplication syndromes as well as the pericentromeric and subtelomeric regions. The newest version, CMA-HR 180k incudes:

- *The most comprehensive array to date, with 1,714 genes covered*
- *All gene coverage is exon by exon focused*
- *High resolution whole genome coverage (first and only array to offer both whole genome coverage and exon coverage)*
- *180,000 of the best performing oligos (increased from 105,000)*
- *Many new genes associated with mental retardation, epilepsy, autism, and heart defects*
- *700 of the most important microRNAs*
- *Complete tiling coverage of the mitochondrial genome*

REASONS FOR REFERRAL

Chromosomal Microarray Analysis may be ordered for all patients with any indication of genomic imbalance which includes: dysmorphic features, unexplained mental retardation/developmental delay, autism spectrum disorder, and/or multiple congenital anomalies. CMA is the more appropriate test for patients who are candidates for subtelomere FISH or multiple individual FISH tests. CMA is well suited for the detection of interstitial duplications that currently can only be detected by interphase FISH. Although CMA can detect some deletions or duplications that cause single gene or contiguous gene phenotypes (e.g. Pelizaeus-Merzbacher), additional testing

methodologies should be appropriately considered. In patients where a duplication is detected by CMA, confirmation testing using chromosome analysis and/or FISH to locate the additional material will be performed at no cost to the patient.

TESTING METHODOLOGY

Chromosomal Microarray Analysis utilizes array-based comparative genomic hybridization (aCGH) with approximately 180,000 oligos covering the whole genome at the average resolution of 30Kb, 1,714 genes with all exons covered, 700 microRNAs and the entire mitochondrial genome. Genomic DNA from the test sample and a control sample are differentially labeled with fluorescent dyes and hybridized to the oligos. Results are analyzed using quantitative imaging methods and analytical software to assist in identifying each targeted-DNA sequence as loss of copy number (deletion), gain of copy number (duplication) or normal copy number. This technology has been validated in our laboratory on many patients with known microdeletions/duplications and other unbalanced karyotypes detected by traditional cytogenetic methods. CMA is limited to detection of gain or loss of genomic material. It will not detect low level mosaicism, balanced translocations, inversions, or point mutations that may be responsible for the clinical phenotype.

SPECIMEN REQUIREMENTS AND TURN AROUND TIME

Blood in both EDTA (purple top) and Sodium Heparin (green top) tubes:

- Adult: 10 cc per tube
- Infant: 2 cc per tube
- Child: 4 cc per tube
- Turn around time: 7 to 10 days

SHIPPING:

Baylor Miraca Genetics Laboratories
Grand Blvd. Receiving Dock
2450 Holcombe Blvd.
Houston, Tx 77021

Prepaid shipping kits are available upon request: Please call us at 1-800-411-gene (4363).

Baylor Miraca
Genetics Laboratories

2450 Holcombe Boulevard, Suite O-100, Houston, Texas 77021
1-800-411-GENE (4363) or 713-798-6555 • www.BMGL.com

3/2015