



EmArray Cyto6000 ©2007

Targeted Array Analysis Combined with Whole Genome Coverage

What is EmArray Cyto6000?

The EmArray Cyto6000 is a Hi-Def Chromosome Microarray Equivalent to a 6,000 Band Karyotype.

Using the latest oligonucleotide microarray technology, Emory is pleased to announce the availability of a high definition microarray with the ability to accurately identify deletions or duplications as small as 500kb anywhere in the genome.

Why Choose EmArray Cyto6000?

The EmArray Cyto6000 features a custom designed oligonucleotide microarray with >44,000 oligos covering the genome. In addition, coverage targets all telomere and centromere regions and known microdeletion/microduplication syndromes. A standard G-banded karyotype (500-650 bands) can detect deletions and duplications of ~5Mb, therefore the EmArray Cyto6000 is equivalent to a 6,000 band karyotype and provides ~10X better resolution. All imbalances are confirmed by a second methodology (usually FISH analysis).

In addition, identification of **clinically relevant abnormalities is maximized** and identification of **benign copy number variants is minimized** by establishing a threshold of 500kb, thereby **reducing the need to obtain parental samples**.

Choose the experts

This new technology and custom design is a giant leap over current chromosome microarray tests on the market, and is brought to you by two expert clinical cytogeneticists with over 45 years combined experience. Drs. Christa Lese Martin and David Ledbetter previously developed telomere FISH testing for the analysis of unexplained mental retardation (in partnership with Vysis/Abbott Molecular), and have now developed a custom hi-def molecular karyotype for postnatal diagnostic applications to be performed in conjunction with a traditional G-banded karyotype. Emory Genetics Laboratory has over 35 years experience in cytogenetic testing, and is supported by a team of clinical geneticists and highly trained genetic counselors who are available to address your questions.

How does the EmArray Cyto6000 work?

This custom array consists of approximately 44,000 oligos (probes) spaced at a minimum of 75kb across the genome, with increased coverage at telomeres, centromeres and clinically relevant regions of the genome.

Oligo array analysis uses DNA isolated from a peripheral blood sample. The patient DNA is fluorescently labeled and mixed in equal ratios with fluorescently labeled opposite gender control DNA. The two DNA samples are hybridized to an array. The competitive hybridization of control DNA to the patient DNA reveals array copy number imbalances in the patient for the chromosomal regions tested. Analysis is performed using quantitative imaging methods to measure each region as normal, deleted or duplicated.

Gains or losses are compared to known copy number variation regions observed in the general population and to previously reported deletions/duplications. In some cases, parental samples are requested to determine whether the gain or loss is inherited or *de novo*. The laboratory will contact the ordering provider directly to discuss these cases. Targeted FISH analysis (or another methodology) is used to confirm any abnormal findings.

Detection is limited to gain of copy number (duplication), loss of copy number (deletion), or normal copy number. Oligo array analysis will not detect balanced chromosome rearrangements, such as translocations or inversions, or smaller deletions, duplications or point mutations that may underlie the clinical presentation of the patient.

Why choose EmArray Cyto6000 testing for your patient?

Approximately 0.6% of liveborn infants have an unbalanced chromosome rearrangement, based on conventional karyotype analysis. The molecular cytogenetic technique, oligonucleotide array-based Comparative Genomic Hybridization (also called oligo array or chromosomal microarray), has been introduced for identifying very small gains or losses of chromosome material. The oligo array has the capability to be a more sensitive, efficient and cost-effective method than currently available techniques, such as standard G-banding with a resolution of ~5Mb, or FISH, which has a higher resolution (~50-100kb), but can only test limited loci at one time. In fact, one array analysis provides results equal to up to hundreds of FISH probes combined. The oligo array currently provides the most robust method for chromosome imbalance detection, and can identify deletions or duplications as small as 500 kb or smaller for targeted genes. This test is currently being used as an adjunct to routine G-banding analysis. G-banding must be performed before, or simultaneously with, microarray analysis.

Indications for Testing

EmArray Cyto6000 is indicated for patients with normal chromosome analysis and:

- Unexplained developmental delay or mental retardation
- Dysmorphic features or congenital anomalies
- Autism spectrum disorders, seizures, or a clinical presentation suggestive of a chromosomal syndrome

EmArray Cyto6000 is also indicated for individuals with a previously identified chromosomal abnormality:

- For unbalanced rearrangements, oligo array analysis can be used to size the deletion or duplication, and identify genes involved
- For 'apparently balanced' rearrangements and an abnormal clinical phenotype, oligo array analysis can be used to test for cryptic deletions/duplications at the breakpoints

Contact the laboratory to discuss cases where a specific deletion/duplication is suspected.

EmArray Cyto6000 Detection and Clinical Sensitivity

The detection of deletions and duplications of 500kb or greater is expected to be very high. Detection is limited to gain of copy number (duplication), loss of copy number (deletion), or normal copy number. Deletions and duplications 500kb or greater are reported. Smaller deletions or duplications in regions of known microdeletion/microduplication syndromes or in targeted genes will also be reported.

The clinical sensitivity for common microdeletion or microduplication syndromes is available in our detection rate chart. The clinical sensitivity for other disorders is dependant on the proportion of cases caused by deletions/duplications compared with other mutations not detectable by array analysis, which will vary by disease. Oligo array analysis will not detect translocations, inversions, smaller imbalances, point mutations, or low level mosaicism (usually less than 25%) that may underlie the clinical presentation of the patient.

Please refer to the website description for the current detection tables.

CPT Codes

83890, 83892(x2), 83894, 83896(x10), 83897, 88384, 88385, 88386, 88230

Contact Emory Genetics Laboratory with your questions by calling (404)778-8500.