Chromosomal Microarray: EmArray Cyto

Test Code: VA
Turnaround time: 5 days - 7 days (All abnormal findings are verbally reported immediately. Confirmatory testing for abnormalities will delay final reporting. A written preliminary report is available upon request.)
CPT Codes: 81228 x1

Condition Description

What is EmArray Cyto?
- The EmArray Cyto chromosomal microarray utilizes 60,000 (60K) oligonucleotides to achieve whole genome coverage at a 75 kilobase (kb) spacing. It additionally covers more than 400 targeted regions, including known recurrent microdeletion/microduplication syndromes, centromere and telomere regions and disease-causing genes. The design is based on recommendations from the International Standards for Cytogenomic Arrays (ISCA) Consortium (Bladwin et al. (2008) Genet Med 10(6):415-429). Click here to read more about the CytoScan SNP Array, including case examples.

Why Choose EmArray Cyto?
- When compared to conventional cytogenetic testing by G-banded chromosome analysis, the EmArray Cyto detects more than twice as many clinically significant imbalances. The literature supports offering whole genome chromosomal microarray testing as the first tier test for all genetic evaluations for developmental disabilities including birth defects, developmental delay, dysmorphic features, growth deficiency and intellectual disability (Miller et al. (2010) Am J Hum Genet 86(5):749-764). Testing for chromosomal imbalances by microarray is cost effective given the greater capability to detect imbalances when compared to conventional methods. The EmArray Cyto is roughly equivalent to the cost of chromosome analysis by G-banding plus one targeted FISH study.

References

Indications
Chromosomal microarray is indicated for the following reasons:
- Unexplained developmental delay or intellectual disability
- Autism spectrum disorders
- Epilepsy or seizures
- Dysmorphic features, congenital anomalies or birth defects
- Normal chromosome analysis and an abnormal phenotype
- Apparently balanced chromosome rearrangements and an abnormal phenotype to look for cryptic imbalances at the breakpoints
- Characterization of a previously identified chromosome abnormality

Methodology
DNA isolated from peripheral blood is hybridized to a custom array containing oligonucleotide probes across the genome to detect copy number imbalances. FISH analysis or another method, such as G-banded karyotype, is used to confirm any abnormal findings either at the time of initial testing or upon receipt of parental samples, depending on the abnormality.

Detection
The detection of deletions and duplications of 400 kb 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 of 400 kb or greater are reported. Smaller deletions or duplications in regions of known microdeletion/microduplication syndromes or in clinically relevant genes will also be reported. The clinical sensitivity for known microdeletion or microduplication syndromes is available in our detection rate chart. The clinical sensitivity for other disorders is dependent on the proportion of cases caused by deletions/duplications compared with other mutations not detectable by array analysis. Microarray will not detect balanced translocations, balanced inversions, imbalances smaller than the resolution of this array, point mutations or low level mosaicism (usually less than 25%) that may underlie the clinical presentation of the patient.

Reference Range
Consecutive abnormal oligonucleotide probes are used to identify regions of imbalance. Log2 ratios less than 0.32 are indicative of a deletion or loss of genetic material, while those greater than 0.26 are indicative of a duplication or gain of genetic material.

Specimen Requirements
Additional Specimen Collection/Handling Instructions Required for this Test

If sending whole blood, both tube types are required for this test.

**Type: Whole Blood**

**Specimen Requirements:**

In EDTA (purple top) AND sodium heparin (green top) tubes:

- Infants (Children (>2 years): 3-5 ml in both tubes
- Older Children & Adults: 7-10 ml in both tubes

Specimen Collection and Shipping: Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.

**Special Instructions**

Parental samples may be requested to interpret the clinical significance of some findings.

**Sample Storage and Data Usage:** As a participant in the ISCA (International Standard Cytogenomic Array) Consortium, EGL Genetics retains patient samples indefinitely for validation, educational purposes and/or research. The submitted clinical information and test results are also included in a HIPAA-compliant, de-identified public database as part of the National Institute of Health's effort to improve diagnostic testing and our understanding of the relationships between genetic changes and clinical symptoms (for information about the molecular cytogenetic database visit the consortium website at [https://www.iscaconsortium.org/](https://www.iscaconsortium.org/)). Confidentiality of each sample is maintained.

Patients may request to have their samples discarded upon test completion and to opt-out of participation in the database by:

1) Checking the box provided on the test requisition or consent form
2) Calling the laboratory at (404) 778-8499 and asking to speak with a laboratory genetic counselor
3) Visiting the opt-out page: [http://genetics.emory.edu/egl/opt-out](http://genetics.emory.edu/egl/opt-out)

**Related Tests**

- STAT analysis of chromosomes 13, 18, 21, X or Y and the 22q11 region
- Targeted testing by FISH is available to family members of an individual with a deletion or duplication detected by microarray.
- Prenatal Chromosomal Microarray (EmArray Cyto) (CMPRE)
- Production of Conception (POC) Microarray (EmArray Cyto) (CMPOC)
- EmArray Cyto + SNP (CMSNP)