CONGENTIAL POSTNATAL CHROMOSOMAL MICROARRAY (CMA) ANALYSIS:

<table>
<thead>
<tr>
<th>BILLING CODE</th>
<th>TEST NAME</th>
<th>SPECIMEN TYPE</th>
<th>COLLECTION</th>
<th>TAT</th>
</tr>
</thead>
<tbody>
<tr>
<td>CTGE</td>
<td>Chromosomal Microarray Analysis</td>
<td>Peripheral Blood</td>
<td>1 tube each whole blood in: Sodium Heparin and EDTA 5 ml each for Adults, 2 ml each for Children/Infants. Transport at room temperature</td>
<td>14 days*</td>
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*TAT may be extended if additional confirmation studies are needed

General Information:
Chromosomal Microarray (CMA) analysis is an array comparative genomic hybridization (array CGH or aCGH) based test that utilizes oligonucleotide probes to examine the entire genome for copy number imbalance. The resolution of this molecular cytogenetic assay is limited to the spacing and the number of probes present on the microarray. This test is able to detect chromosomal gains and losses at a level of resolution more than 10-fold greater than that which can be detected by traditional high-resolution chromosome analysis. CMA can identify aneusomy as well as microscopic and submicroscopic deletions and duplications in areas covered by the microarray. This includes all known microdeletion / microduplication syndromes, subtelomeric regions and pericentromeric regions. CMA can also facilitate identification of marker chromosomes.

Testing Recommendations:
In 2010, the American College of Medical Genetics released new Practice Guidelines in Medical Genetics which recommends array-based technology such as CMA for detection of chromosomal abnormalities. CMA testing for chromosomal imbalances is recommended as a first-line test in the initial evaluation of individuals with the following:

1. Multiple anomalies not specific to a well-delineated genetic syndrome.
2. Apparently non-syndromic developmental delay / intellectual disability.
3. Autism spectrum disorders.

Limitations of the Test:
This test is not designed to detect small copy number changes at the level of individual exons of genes, single basepair mutations, low-level mosaicism, uniparental disomy (UPD), or polyploidy. CMA also cannot detect balanced rearrangements such as; inversions, reciprocal translocations, or insertions that do not result in gain or loss of chromosomal material.

Visualization Studies:
Copy number gains and losses (>300 kb) in size may be visualized by FISH or limited chromosome analyses. Small, submicroscopic gains will be examined by metaphase FISH analysis to determine if the extra material moved to a different chromosomal location. Depending on the size of the copy number gain, it may not be possible to confirm tandem duplications where the extra material is present adjacent to the original copy of the genomic segment.

Familial Studies:
Some copy number changes identified by CMA analysis may require parental follow-up studies. The recommendation for parental studies, if necessary, will appear on the report.

07/12 SLK