Medical EmExome Array: Deletion/Duplication Analysis

**Test Code:** EXODD
**Turnaround time:** 2 weeks
**CPT Codes:** 81228 x1

### Condition Description

The Medical EmExome Array: Deletion/Duplication Analysis is designed to identify deletions and duplications within the exons of disease-associated genes, which are missed by exome sequencing platforms. Using a one million probe array with the cytogenetic oligonucleotide array as the backbone, EGL has designed an extremely enriched, targeted deletion/duplication analysis that ensures >98% coverage of all exons in the ~4,600 disease-associated genes of the human genome.

The array is designed to be versatile, with the ability to add additional probes and keep up with the changing medical and scientific understanding of genes associated with disease.

Although this array was designed to complement the Medical EmExome, a highly-sensitive clinical exome sequencing assay, it can also be used following, or in addition to, the mutation analysis or sequencing tests performed at other laboratories.

The Exome Coverage Tool can be used to view typical depth of sequence coverage obtained by exome sequencing performed by EGL. To access this tool, please click here.

### Indications

This test should be considered when:

- Only a single pathogenic variant or VOUS is identified in a patient with an autosomal recessive disorder.
- No pathogenic variants or VOUS were identified on prior exome sequencing.
- The suspected condition(s) have a high incidence of causative deletions/duplications.
- Cytogenetic abnormalities need to be assessed in addition to gene-specific deletions/duplications.

### Methodology

DNA isolated from peripheral blood is hybridized to a gene-targeted CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes that cover the entire genomic region.

Please note that a "backbone" of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

### Detection

Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

### Specimen Requirements

Submit only 1 of the following specimen types

#### Type: Whole Blood

**Specimen Requirements:**

In EDTA (purple top) tube:

- Infants (2 years): 3-5 ml
- Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.

#### Type: Isolated DNA

**Specimen Requirements:**

In microtainer: 60 ug

Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/ul of TE buffer. Ship sample at room temperature with overnight delivery.
Special Instructions

Please submit medical records or clinic summary notes, and a signed consent form when ordering exome testing. Testing will not be initiated until these documents are received.

Related Tests

- Medical EmExome: Clinical Exome Sequencing, Trios
- Medical EmExome: Clinical Exome Sequencing, Proband Only
- Medical EmExome: Clinical Exome Sequencing, Additional Family Member
- Medical EmExome: Sanger Confirmation and Interpretation Only
- Medical EmExome: Interpretation Only (Exome or Genome)