**Congenital Muscular Dystrophy: Deletion/Duplication Panel**

**Test Code:** MD211  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1, 81406 x1

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**Condition Description**

The congenital muscular dystrophies are a group of genetically and clinically heterogeneous hereditary myopathies characterized by congenital hypotonia and muscle weakness, contractures, and delayed motor development. Muscle biopsy usually reveals a nonspecific dystrophic pattern. The clinical course is broadly variable and can involve the brain and eyes. Initial testing often includes clinical evaluation, muscle imaging, electromyography, and muscle biopsy, followed by targeted genetic testing.

Reference:

- OMIM

**Genes**

- B3GALNT2  
- CHKB  
- COL6A1  
- COL6A2  
- COL6A3  
- CRPPA  
- DAG1  
- DPM1  
- DPM2  
- DPM3  
- FKRP  
- FKTN  
- ITGA7  
- LAMA2  
- LARGE1  
- LMNA  
- POMGNT1  
- POMGNT2  
- POMT1  
- POMT2  
- RYR1  
- SELENON  
- TCAP  
- TMEM5

**Indications**

This test is indicated for:

- Confirmation of a clinical diagnosis of congenital muscular dystrophies (CMD).

**Methodology**

**Deletion/Duplication Analysis:** 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.

**Detection**

**Deletion/Duplication Analysis:** 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 (EDTA)**

**Specimen Requirements:**

- EDTA (Purple Top)  
- Infants and Young Children (2 years of age to 10 years old): 3-5 ml  
- Older Children & Adults: 5-10 ml  
- Autopsy: 2-3 ml unclotted cord or cardiac blood

**Specimen Collection and Shipping:**

Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not freeze.

**Type: DNA, Isolated**

**Specimen Requirements:**

- Microtainer  
- 3µg  
- Isolation using the Perkin Elmer™Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

**Specimen Collection and Shipping:**

Refrigerate until time of shipment in 100 ng/µL in TE buffer. Ship sample at room temperature with overnight delivery.

**Related Tests**

- Single-gene tests.  
- Neuromuscular Disorders Panel.  
- Congenital Muscular Dystrophy: Sequencing Panel.