Congenital Muscular Dystrophy: Deletion/Duplication Panel

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

## 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  
- CHK2  
- COL6A1  
- COL6A2  
- COL6A3  
- DAG1  
- DPM1  
- DPM2  
- DPM3  
- FKRP  
- FKTN  
- ISPD  
- 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. 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: Ship sample at room temperature with overnight delivery.

#### Type: Isolated DNA

Specimen Requirements:

- In microtainer: 10 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.

### Related Tests

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