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, 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.