Expanded Neuromuscular Disorders: Deletion/Duplication Panel

**Test Code:** MD360
**Turnaround time:** 2 weeks
**CPT Codes:** 81161 x1, 81404 x1, 81405 x1, 81406 x1

**Condition Description**

Neuromuscular disorders (NMDs) collectively refer to the many disorders that affect the peripheral nervous system either by impairing the proper development or functioning of muscles, or by damaging the associated nerves or neuromuscular junctions. NMDs comprise over 200 Mendelian disorders, all of which are rare individually, but have an approximate disease prevalence of 1 in 3,000 altogether. Of the inherited NMDs, muscular dystrophies are the most common. Muscular dystrophies are highly heterogeneous muscle disorders that share clinical, genetic, and pathological characteristics; their major clinical characteristics include muscle degeneration and wasting, progressive muscle weakness, hypotonia, and elevated serum creatine kinase levels.

The expanded neuromuscular panel includes 79 genes which demonstrate a wide range of clinical presentation and heterogeneity. They include muscular dystrophies, congenital myopathies, and congenital myasthenic syndrome. Over the past few years a number of genes with overlapping clinical phenotypes have been identified in neuromuscular disorders. The expanded neuromuscular disorders panel is designed to capture the entire mutation spectrum of these genes.

**References:**

**Genes**

`ACTA1`, `AMPD1`, `ANOS1`, `BAG3`, `BIN1`, `BSDL2`, `CAPN3`, `CAV3`, `CAVIN1`, `CFL2`, `CHAT`, `CHRNA1`, `CHRNB1`, `CHRND`, `CHRNE`, `CHRG`, `COL6A1`, `COL6A2`, `COL6A3`, `COLO`, `CRYAB`, `DAG1`, `DES`, `DM2D`, `DNM2`, `DOK7`, `DYSF`, `EMD`, `FHL1`, `FKRP`, `FKTN`, `FLNC`, `GAA`, `GLES`, `GNE`, `IGHMBP2`, `ISP1`, `ITGA7`, `LAMA2`, `LARGE1`, `LDB3`, `LMNA`, `MTM1`, `MTMR14`, `MUSK`, `MYH2`, `MYH7`, `MYOT`, `NEB`, `PABPN1`, `PLEC`, `PLEKHG5`, `PMM2`, `POMGNT1`, `POMT1`, `POMT2`, `PYGM`, `RAPSN`, `RYR1`, `RYR2`, `SCN4A`, `SELENON`, `SGCA`, `SGCB`, `SGCD`, `SGCE`, `SGCG`, `SIL1`, `SYNE1`, `TCAP`, `TNNT2`, `TNNT1`, `TPM2`, `TPM3`, `TRIM32`, `TTN`, `VRK1`

**Indications**

This test is indicated for:

- Confirmation of a clinical diagnosis of neuromuscular disorders.

**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**

**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

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

- Neuromuscular Disorders Panel.
- Limb-girdle Muscular Dystrophy Panel.
- Congenital Muscular Dystrophy Panel.
- Expanded Neuromuscular Disorders: Sequencing Panel.