Neuromuscular Disorders: Deletion/Duplication Panel

**Test Code:** DNEU1  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81404 x1, 81405 x1, 81406 x1, 81408 x1, 81161 x1

### Condition Description

The neuromuscular disorders (NMD) are a group of conditions that affect the peripheral nervous system and muscles. Primarily, they affect the ability to perform voluntary movements. They range in onset from before a child is born to much later in life with the majority beginning during infancy, childhood, or the teenage years. With many of the neuromuscular disorders overlapping in their clinical and/or pathological phenotypes, molecular testing can be necessary to pinpoint the precise disorder a patient has.

The Neuromuscular Disorders Panel includes testing for nemaline myopathy, limb girdle muscular dystrophy, Emery-Dreifuss muscular dystrophy, congenital muscular dystrophy, Zellweger syndrome spectrum, and cardiomyopathies. Individual disorders included on this panel are myoadenylate deaminase deficiency, erythrocyte AMP deaminase deficiency, myofibrillar myopathy, Duchenne/Becker muscular dystrophy, congenital disorder of glycosylation type 1a, malignant hyperthermia susceptibility, myoclonus dystonia, Marinseso-Sjogren syndrome, and distal arthrogryposis.

Reference:

### Genes

ACTA1, AMPD1, ANO5, CAPN3, CAV3, COL6A1, COL6A2, COL6A3, CRPPA, DES, DMD, DYSF, EMD, FKRP, FKTN, GAA, GNE, ITGA7, LAMA2, LARGE1, LAMA4, MYOT, NEB, PLEC, PMM2, POMGNT1, POMT1, POMT2, PYGM, RYR1, RYR2, SELENON, SGCA, SGCB, SGCD, SGCE, SGCG, SILT, TCAP, TENM2, TNNT1, TPM2, TPM3, TRIM32, TTN

### 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.

### 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: DNA, Isolated**

**Specimen Requirements:**
- Microtainer 3µg
- Isolation using the Perkin Elmer™ 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.

**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.

### Related Tests

Disclaimer: This information is confidential and subject to change without notice. It may not be reproduced in whole or part unless authorized in writing by an authorized EGL representative.
- Single-gene testing is available for most genes on this panel.
  - Limb-Girdle Muscular Dystrophy: Sequencing Panel.
  - Congenital Muscular Dystrophy: Sequencing Panel.
  - Bethlem Myopathy/Ulrich Congenital Muscular Dystrophy Panel.
  - Expanded Neuromuscular: Sequencing Panel.
  - Neuromuscular Disorders: Sequencing Panel.