Neuromuscular Disorders: Deletion/Duplication Panel

**Test Code:** DNEU1  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81161 x1, 81404 x1, 81405 x1, 81406 x1, 81408 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 cardiomypathies. Individual disorders included on this panel are myoadenylate deaminase deficiency, erythrocyte AMP deaminase deficiency, myotubular myopathy, Duchenne/Becker muscular dystrophy, congenital disorder of glycosylation type 1a, malignant hyperthermia susceptibility, myoclonus dystonia, Marfan syndrome, and distal arthrogryposis.

Reference:  

### Genes

ACTA1, AMPD1, ANOS1, CAPN3, CAV3, COL6A1, COL6A2, COL6A3, DES, DMD, DYSF, EMN, FKBP3, FKTN, GAA, GNE, IPSD, ITGA7, LMNA, MYOT, NEB, PLEC, PMM2, POMGNT1, POMT1, POMT2, PYGM, RYR1, RYR2, SELENON, SGCA, SGCB, SGCD, SGCE, SGCG, SIL1, TAC1, TNN1, TNNI1, TPM1, TPM3, TRIM32, TTN

### Indications

- 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 on 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

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.
Specimen Requirements:

In microtainer: 10 µg

Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/µl of TE buffer. Ship sample at room temperature with overnight delivery.

**Related Tests**

- Single-gene testing is available for most genes on this panel.
- Limb-Girdle Muscular Dystrophy: Sequencing Panel.
- Congenital Muscular Dystrophy: Sequencing Panel.
- Bethlem Myopathy/Ullrich Congenital Muscular Dystrophy Panel.
- Expanded Neuromuscular: Sequencing Panel.
- Neuromuscular Disorders: Sequencing Panel.