Neuromuscular Myopathy Testing Algorithm

1. **Inherited myopathy suspected**
   - Adult onset proximal myopathy phenotype
     - Order: MBX / Muscle Pathology Consultation
   - Early onset myopathy or categorical myopathy (see Myopathy subpanels box below) phenotype
     - Order: MBX / Muscle Pathology Consultation
   - Mitochondrial cause suspected
     - Order: MBX / Muscle Pathology Consultation

2. **Duchenne muscular dystrophy phenotype**
   - Inflammatory immune cause found
     - Yes
     - Order: DBMD / Duchenne/Becker Muscular Dystrophy DMD Gene, Large Deletion and Duplication Analysis
     - No or inconclusive
     - Order: MBX / Muscle Pathology Consultation

3. **Definitive cause found**
   - Yes
   - Disorder-specific management
   - Variant supported as causal
     - Yes
     - Disorder-specific management
     - No
     - Consider WES / Whole Exome Sequencing
   - No or inconclusive
     - Disorder-specific management

Myopathy subpanels (categorical myopathies):
- NMPAN / Neuromuscular Genetic Panels by Next-Generation Sequencing (NGS)
  - Myopathy Expanded Panel (141 genes)
  - Congenital Myopathy Panel (36 genes)
  - Distal Myopathy Panel (27 genes)
  - Emery-Dreifuss Panel (5 genes)
  - Metabolic Myopathy Panel (41 genes)
  - Muscular Dystrophy Panel (77 genes, includes Limb-Girdle Muscular Dystrophy)
  - Myofibrillar Myopathy Panel (12 genes)
  - Rhabdomyolysis and Myopathy Panel (31 genes)
  - Skeletal Muscle Channelopathy (6 genes, includes periodic paralysis)
  - Congenital Myasthenic Syndromes Panel (25 genes)
  - Custom Gene Panel (select genes from list) via CGO

Order based on presentation and pattern of inheritance:
- MITOP / Mitochondrial Full Genome Analysis by Next-Generation Sequencing (NGS)
- MITON / Mitochondrial Nuclear Gene Panel by Next-Generation Sequencing (NGS)
- MITOT / Combined Mitochondrial Analysis, Mitochondrial Full Genome and Nuclear Gene Panel (includes genes from both MITOP and MITON)
- Correlate any DNA variants with muscle biopsy, if performed
  - If muscle biopsy not performed, order MBX / Muscle Pathology Consultation
  - Perform recommended functional metabolic assays (per Interpretative Report)

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