# Neuromuscular Myopathy Testing Algorithm

## Inherited myopathy suspected

- **Adult onset proximal myopathy phenotype**
  - Order: MBX / Muscle Pathology Consultation
  - Inflammatory immune cause found
    - YES: Disorder-specific management
    - NO: Order MUPAN / Comprehensive Neuromuscular Gene Panel OR see Tests To Consider box below

- **Specific (other) phenotypes**
  - Limb-girdle muscular dystrophy
  - Distal myopathy
  - Emery-Dreifuss muscular dystrophy
  - Congenital myopathy
  - Congenital muscular dystrophy
  - Myofibrillar myopathy
  - Tubular aggregate myopathy
  - Vascular myopathy
  - Metabolic myopathy
  - Skeletal muscle channelopathy

## Early onset myopathy or categorical myopathy (see Tests To Consider box below) phenotype

- Duchenne muscular dystrophy phenotype
  - YES: Order DMDZ / DMD Gene, Full Gene Analysis
  - NO: Disorder-specific management

## Mitochondrial cause suspected

- Early onset myopathy or categorical myopathy phenotype
  - Order: MBX / Muscle Pathology Consultation
  - Muscle biopsy supports mitochondrial cause
    - YES: Disorder-specific management
    - NO OR INCONCLUSIVE: Definitive cause found

## Tests To Consider

- RABMP / Inherited Rhabdomyolysis/Metabolic Myopathy Panel
- MDYSP / Inherited Muscular Dystrophy Gene Panel
- LGCMP / Inherited Limb-Girdle Muscular Dystrophy and Congenital Myasthenic Syndrome Gene Panel
- CMSP / Inherited Congenital Myasthenic Syndrome Gene Panel
- EDMOP / Inherited Emery-Dreifuss Gene Panel
- SMCP / Inherited Skeletal Muscle Channelopathy Gene Panel
- DMDZ / DMD Gene, Full Gene Analysis
- DBMD / Duchenne/Becker Muscular Dystrophy DMD Gene, Large Deletion and Duplication Analysis
- CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing; stating disease state: Neurologic Disorders

- WESDX / Whole Exome Sequencing for Hereditary Disorders
- NMS1 / Necrotizing Myopathy Evaluation, Serum to assess for potential autoimmune causes