MAYO CLINIC LABORATORIES

Neuromuscular Myopathy Testing Algorithm

Inherited myopathy suspected

Adult onset proximal myopathy phenotype

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Order: MBX / Muscle Pathology Consultation

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Inflammatory immune cause found

- YES → Disorder-specific management
- NO → Order MUPAN / Comprehensive Neuromuscular Gene Panel OR see Tests To Consider box below

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

↓ Order MUPAN / Comprehensive Neuromuscular Gene Panel OR see Tests To Consider box below

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

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Duchenne muscular dystrophy phenotype

- YES → Order DMDZ / *DMD* Gene, Full Gene Analysis
  - POSITIVE → Disorder-specific management
  - NEGATIVE → Order MUPAN / Comprehensive Neuromuscular Gene Panel
- NO → Order MUPAN / Comprehensive Neuromuscular Gene Panel

Mitochondrial cause suspected

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Order: MBX / Muscle Pathology Consultation

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Muscle biopsy supports mitochondrial cause

- YES → Order based on presentation and pattern of inheritance:
  - MITOP / Mitochondrial Full Genome Analysis by Next-Generation Sequencing (NGS)
  - NMITO /Nuclear Mitochondrial Gene Panel, Next-Generation Sequencing

  OR
  - CMITO / Combined Mitochondrial Full Genome and Nuclear Gene Panel (includes genes from both MITOP and MITON)
- NO → Order MUPAN / Comprehensive Neuromuscular Gene Panel

Definitive cause found

- YES → Disorder-specific management
- NO OR INCONCLUSIVE →
  - Correlate any DNA variants with muscle biopsy, if performed
    - If muscle biopsy **not** performed, order MBX
  - Perform recommended functional metabolic assays (per Interpretative Report)

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    Variant supported as causal
    - YES → Disorder-specific management
    - NO → Consider
      - WESDX / Whole Exome Sequencing for Hereditary Disorders
      - NMS1 / Necrotizing Myopathy Evaluation, Serum to assess for potential autoimmune causes

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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
- EDMDP / 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

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Clickable PDF
Click on blue tests to view test catalog information

12/2022