Inherited Motor Neuron Disease Testing Algorithm

Inherited Motor Neuron Disease suspected

- Spinal muscular atrophy (SMA) phenotype - SMA types 1-4
  - Order SMNDX / Spinal Muscular Atrophy Diagnostic Assay by Deletion/Duplication Analysis
  - POSITIVE: STOP Disease-specific management
  - NEGATIVE OR INCONCLUSIVE: Consider SMN1Z / SMN1Z Gene, Full Gene Analysis
    - POSITIVE: STOP Disease-specific management
    - NEGATIVE OR INCONCLUSIVE: Order NMPAN / Neuromuscular Genetic Panels by Next-Generation Sequencing (NGS)
      Specify subpanel: Motor Neuron Disease Panel (17 genes)
      - POSITIVE: Consider referral to Mayo Clinic or other available ALS Clinic
      - NEGATIVE OR INCONCLUSIVE: Consider referral to Mayo Clinic or other available ALS Clinic

- Spinal bulbar muscular atrophy (SBMA) phenotype (Kennedy disease)
  - Order SBULB / Spinobulbar Muscular Atrophy (Kennedy Disease), Molecular Analysis
  - POSITIVE: STOP Disease-specific management
  - NEGATIVE OR INCONCLUSIVE: Order C9ORF / C9orf72 Hexanucleotide Repeat, Molecular Analysis
    - POSITIVE: Consider referral to Mayo Clinic or other available ALS Clinic
    - NEGATIVE OR INCONCLUSIVE: Order C9ORF / C9orf72 Hexanucleotide Repeat, Molecular Analysis

- Frontal temporal dementia-amyotrophic lateral sclerosis (FTD-ALS) and other ALS suspects
  - Order C9ORF / C9orf72 Hexanucleotide Repeat, Molecular Analysis
  - POSITIVE: Consider referral to Mayo Clinic or other available ALS Clinic
  - NEGATIVE OR INCONCLUSIVE: Consider referral to Mayo Clinic or other available ALS Clinic

Sporadic amyotrophic lateral sclerosis (ALS) suspected

CONSIDER

Inherited Motor Neuron Disease suspected*

*Medical genetic consultation strongly recommended for patients undergoing genetic testing for motor neuron disease.