Inherited Motor Neuron Disease Testing Algorithm

Inherited Motor Neuron Suspect?*

YES

- Spinal muscular atrophy (SMA) phenotype -SMA types 1-4
- Spinal bulbar muscular atrophy (SBMA) phenotype (Kennedy disease)
- Frontal temporal dementia-amyotrophic lateral sclerosis (FTD-ALS) and other ALS suspects

NO

- Sporadic ALS suspect

CONSIDER

Order SMNDX / Spinal Muscular Atrophy Diagnostic Assay

POSITIVE

- Consider available therapies

NEGATIVE

Order SBULB / Spinobulbar Muscular Atrophy (Kennedy Disease), Molecular Analysis

POSITIVE

- Consider symptomatic therapies and clinical trials

NEGATIVE

Order C9ORF / C9orf72 Hexanucleotide Repeat, Molecular Analysis

POSITIVE

- Consider referral to available multidisciplinary clinic

NEGATIVE

Order NMPAN / Neuromuscular Genetic Panels by Next-Generation Sequencing (NGS)

select the Motor Neuron Disease Panel (17 genes)

DEFINITIVE POSITIVE

- Consider referral to available/local ALS clinics

NEGATIVE OR INCONCLUSIVE

- Consider: Diagnostic Odyssey Clinic With Whole Exome or Genome sequencing

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