Inherited Motor Neuron Disease Testing Algorithm

Spinal muscular atrophy (SMA) phenotype - SMA types 1-4
Order SMNDX / Spinal Muscular Atrophy Diagnostic Assay by Deletion/ Duplication Analysis

Spinal bulbar muscular atrophy (SBMA) phenotype (Kennedy disease)
Order SBULB / Spinobulbar Muscular Atrophy (Kennedy Disease), Molecular Analysis

Frontal temporal dementia-amyotrophic lateral sclerosis (FTD-ALS) and other ALS suspects
Order C9ORF / C9orf72 Hexanucleotide Repeat, Molecular Analysis

Inherited Motor Neuron Disease suspected*
Sporadic amyotrophic lateral sclerosis (ALS) suspected

CONSIDER

Sporadic amyotrophic lateral sclerosis (ALS) suspected

POSITIVE
NEGATIVE OR INCONCLUSIVE

STOP
Disease-specific management

POSITIVE
NEGATIVE OR INCONCLUSIVE

Consider SMN1Z / SMN1Z Gene, Full Gene Analysis

STOP
Disease-specific management

POSITIVE
NEGATIVE

Order C9ORF / C9orf72 Hexanucleotide Repeat, Molecular Analysis

CONSIDER

Inherited Motor Neuron Disease suspected*

POSITIVE
NEGATIVE OR INCONCLUSIVE

STOP
Disease-specific management

POSITIVE
NEGATIVE

Consider referral to Mayo Clinic or other available ALS Clinic

NEGATIVE OR INCONCLUSIVE

Order C9ORF / C9orf72 Hexanucleotide Repeat, Molecular Analysis

CONSIDER

Inherited Motor Neuron Disease suspected*

POSITIVE
NEGATIVE OR INCONCLUSIVE

STOP
Disease-specific management

POSITIVE
NEGATIVE

Consider referral to Mayo Clinic or other available ALS Clinic

NEGATIVE OR INCONCLUSIVE

Order NMPAN / Neuromuscular Genetic Panels by Next-Generation Sequencing (NGS)
Specify subpanel: Motor Neuron Disease Panel (17 genes)

CONSIDER

Inherited Motor Neuron Disease suspected*

POSITIVE
NEGATIVE OR INCONCLUSIVE

STOP
Disease-specific management

POSITIVE
NEGATIVE

Consider referral to Mayo Clinic or other available ALS Clinic

NEGATIVE OR INCONCLUSIVE

Consider referral to Mayo Clinic or other available ALS Clinic

Consider: WES / Whole Exome Sequencing or whole genome sequencing

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