Northgate Neurology & ALS Center

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NORTHGATE NEUROLOGY — COMPREHENSIVE CONSULTATION REPORT

Amyotrophic Lateral Sclerosis (ALS) / Motor Neuron Disease Evaluation

Patient Name:	[Sample Patient]	**MRN:**	NNG-0000000
Date of Birth:	March 14, 1968 (Age: 56)	**Date of Visit:**	January 9, 2025
Sex:	Male	**Referring Physician:**	Dr. [Sample Physician], MD (Primary Care)
Insurance:	[Sample Insurance Provider]	**Attending Neurologist:**	Dr. [Sample Neurologist], MD, PhD

REASON FOR CONSULTATION

Mr. [Sample Patient] is a 56-year-old right-handed male, previously in excellent health, presenting for comprehensive ALS specialist evaluation following an abnormal EMG/NCS study obtained by his primary care physician in November 2024. He reports an approximately 14-month history of progressive right hand weakness, fasciculations of bilateral upper and lower extremities, and more recent onset of mild dysarthria. Referral was initiated after PCP noted prominent tongue fasciculations and hyperreflexia on examination.

HISTORY OF PRESENTING ILLNESS

Symptom onset approximately November 2023 with insidious right hand clumsiness noticed during fine motor tasks (buttoning shirts, handwriting). Patient initially attributed symptoms to repetitive strain. Over subsequent months, he developed visible muscle twitching (fasciculations) in bilateral upper arms, thighs, and calves. By mid-2024, he reported occasional tripping on the right foot and mild difficulty with prolonged speech, noting voice fatigue after extended conversations.

He denies sensory symptoms, bowel/bladder dysfunction, significant dysphagia (though notes mild slowing), diplopia, or cognitive/behavioral changes. He has not experienced falls to date. He remains employed as a high school principal but has begun to notice functional limitations at work. He reports unintentional weight loss of approximately 8 lbs over 6 months.

Past Medical History: Hypertension (well-controlled on lisinopril 10mg daily), hyperlipidemia. No prior neurological diagnoses. No history of heavy metal exposure, radiation, or prolonged pesticide exposure. No family history of ALS or other neurodegenerative disease. Non-smoker, occasional alcohol.

PHYSICAL AND NEUROLOGICAL EXAMINATION

Vital Signs	BP 128/76 mmHg, HR 72 bpm, RR 16, SpO2 97% on room air, Wt 178 lbs (BMI 24.2)
General	Alert, oriented, no acute distress. Speech mildly dysarthric with nasal quality on prolonged testing.

Cranial Nerves	Tongue: prominent fasciculations bilaterally, mild atrophy noted on right lateral margin. Jaw jerk: brisk (pathological). Gag reflex: intact but slightly sluggish. Extraocular movements intact. No ptosis.
Motor — UE	Right hand intrinsic atrophy (thenar, hypothenar, first dorsal interosseous). Grip strength R: 3/5, L: 4+/5. Wrist extensors R: 4/5. Shoulder abduction bilaterally 5/5.
Motor — LE	Hip flexors 4+/5 bilaterally. Right ankle dorsiflexion 4/5. Mild right foot drop on heel walking. Quadriceps and hamstrings 5/5.
Reflexes	Biceps R: 3+, L: 2+. Triceps R: 3+, L: 2+. Knee R: 3+, L: 3+. Ankle R: 3+, L: 2+. Plantar responses: EXTENSOR bilaterally (positive Babinski sign). Hoffmann sign: positive right.
Fasciculations	Visible spontaneous fasciculations: bilateral deltoids, bilateral quadriceps, bilateral gastrocnemii, right first dorsal interosseous, tongue.
Sensory	Intact to light touch, pinprick, vibration, proprioception in all four limbs.
Coordination	Mild dysmetria on right finger-nose-finger. Dysdiadochokinesia right hand.
Gait	Mild steppage gait on right. Tandem gait preserved. Romberg negative.
Cognitive	Normal orientation, attention, language. MoCA deferred to neuropsych.

DIAGNOSTIC STUDIES

1. Electromyography / Nerve Conduction Study (EMG/NCS) — Northgate Neurophysiology Lab, December 3, 2024

Nerve conduction studies demonstrated normal sensory nerve action potentials and conduction velocities in bilateral median, ulnar, and sural nerves. Motor nerve conduction studies revealed mildly reduced CMAP amplitudes in the right median (4.1 mV; ref >4.5 mV) and right ulnar (5.2 mV; ref >6.0 mV) nerves, consistent with axonal loss. Conduction velocities and distal latencies were within normal limits, arguing against a primary demyelinating process.

Needle EMG findings (abnormal, summarized):

Muscle	Region	Fibrillations	Positive Waves	Fasciculations	MUP Morphology	Recruitment
1st Dorsal Interosseous (R)	Cervical	2+	2+	Present	Large, polyphasic, long duration	Reduced
Abductor Pollicis Brevis (R)	Cervical	2+	1+	Present	Large, polyphasic	Reduced
Biceps Brachii (R)	Cervical	1+	1+	Present	Mildly enlarged	Mildly reduced
Triceps (R)	Cervical	1+	1+	Absent	Mildly enlarged	Normal
Thoracic Paraspinals (R)	Thoracic	1+	1+	Present	Polyphasic	—
Tibialis Anterior (R)	Lumbosacral	1+	1+	Present	Large, polyphasic	Mildly reduced
Gastrocnemius (R)	Lumbosacral	1+	1+	Present	Enlarged	Normal
Tongue (R)	Bulbar	2+	1+	Present	Polyphasic	Reduced

EMG Impression: Active denervation with chronic reinnervation changes identified in bulbar, cervical, thoracic, and lumbosacral regions. Findings are consistent with diffuse lower motor neuron (LMN) involvement across four regions. In conjunction with upper motor neuron signs on examination, these findings fulfill **El Escorial Definite**

ALS criteria.

2. MRI Brain and Cervical Spine with and without Contrast — Northgate Radiology, December 10, 2024

Brain MRI: Subtle T2/FLAIR hyperintensity along the corticospinal tracts bilaterally from the motor cortex through the posterior limb of the internal capsule and cerebral peduncles, consistent with Wallerian degeneration of upper motor neurons. No cortical atrophy disproportionate to age. No mass, hemorrhage, infarct, or enhancing lesion. Mild diffuse cerebral volume loss consistent with age.

Cervical Spine MRI: Multilevel degenerative disc disease with moderate foraminal narrowing at C5-C6 and C6-C7. No significant central canal stenosis. No cord signal abnormality (no myelomalacia or syrinx). The degenerative changes are not felt to be the primary driver of patient's clinical syndrome given the multifocal nature of EMG abnormalities and presence of UMN signs below the level of any compressive pathology.

3. Pulmonary Function Tests (PFTs) — Northgate Pulmonology, December 18, 2024

Measure	Predicted	Observed	% Predicted	Interpretation
FVC (upright)	4.82 L	3.94 L	82%	Mildly reduced
FVC (supine)	4.82 L	3.61 L	75%	Reduced — orthopnea risk
FEV1	3.78 L	3.12 L	83%	Mildly reduced
FEV1/FVC Ratio	0.78	0.79	101%	Normal
SNIP (Sniff Nasal Insp. Pressure)	≥70 cmH2O	58 cmH2O	83%	Mildly reduced
MIP (Max Inspiratory Pressure)	≥80 cmH2O	62 cmH2O	78%	Borderline reduced
PCF (Peak Cough Flow)	≥360 L/min	310 L/min	—	Below assisted cough threshold

Note: Supine FVC drop of 7% suggests early diaphragmatic weakness. Respiratory monitoring every 3 months recommended. NIV not yet indicated but patient counseled on trajectory.

4. Laboratory and Genetic Panel Results

Test	Result	Reference	Flag
CBC with differential	Within normal limits	—	—
CMP (Comprehensive Metabolic Panel)	Within normal limits	—	—
TSH	1.8 mIU/L	0.4–4.0 mIU/L	—
B12 / Folate	412 pg/mL / 18 ng/mL	Normal ranges	—
ESR / CRP	14 mm/hr / 2.1 mg/L	Normal ranges	—
Heavy metals panel (Pb, Hg, As, Tl)	All within normal limits	—	—
Anti-GM1 antibodies	Negative	Negative	—
Paraneoplastic panel (Hu, Yo, Ri, VGCC)	Negative	Negative	—
HIV / HTLV-I/II	Negative	Negative	—
SOD1 gene sequencing	No pathogenic variant identified	No variant	—
C9orf72 repeat expansion	NEGATIVE — 2 repeats (normal <30)	Normal: <30 repeats	—
FUS gene panel	No pathogenic variant identified	No variant	—

TARDBP (TDP-43) sequencing	No pathogenic variant identified	No variant	—
Creatine Kinase (CK)	487 U/L	55–170 U/L	**HIGH ▲**

5. ALS Functional Rating Scale — Revised (ALSFRS-R)

The ALSFRS-R is a validated 12-item scale (0–48) assessing functional status across bulbar, fine motor, gross motor, and respiratory domains. Higher scores indicate better function.

Domain	Item	Score (0–4)	Notes
Bulbar	Speech	3	Detectable speech disturbance
Bulbar	Salivation	4	Normal
Bulbar	Swallowing	3	Early eating problems
Fine Motor	Handwriting	2	Abnormal, legible
Fine Motor	Cutting food / utensils	2	Cuts with difficulty, slow
Fine Motor	Dressing / hygiene	3	Independent with effort
Gross Motor	Turning in bed	4	Normal
Gross Motor	Walking	3	Early ambulation difficulties
Gross Motor	Climbing stairs	3	Slow
Respiratory	Dyspnea	3	Occurs with one of: walking, bathing, dressing
Respiratory	Orthopnea	3	Some difficulty sleeping, no BiPAP
Respiratory	Respiratory insufficiency	4	None
		37 / 48	**Moderate functional impairment**

DIAGNOSIS

Primary Diagnosis: Amyotrophic Lateral Sclerosis (ALS), Sporadic — ICD-10: G12.21

Diagnostic classification: **Definite ALS** per revised El Escorial / Awaji-shima criteria. Patient demonstrates combined upper and lower motor neuron degeneration in four regions (bulbar, cervical, thoracic, lumbosacral) with progressive course, no alternative diagnosis identified. Genetic testing does not reveal a familial ALS-associated variant; this is classified as sporadic ALS.

Disease stage: Estimated **King's Stage 3** (3 CNS regions clinically involved with early respiratory involvement). Estimated rate of progression: moderate (ALSFRS-R decline ~1.0 pt/month based on symptom timeline).

MANAGEMENT PLAN AND RECOMMENDATIONS

Disease-Modifying Therapy: Initiating Riluzole (Rilutek) 50 mg twice daily with food. Patient counseled on mechanism (glutamate antagonism), expected modest benefit on survival (~2–3 months median), and side effect profile (nausea, fatigue, elevated LFTs). LFTs to be monitored at 1 month, 3 months, then quarterly. Alternative: Edaravone (Radicava) IV or oral formulation discussed; patient to consider after reviewing trial data.

Respiratory: Pulmonary function to be reassessed in 3 months. Referral to UCSF Respiratory Therapy for NIV (BiPAP) education and fitting; initiation recommended when FVC falls below 65% or SNIP <40 cmH2O. Cough assist device (mechanical in-exsufflator) recommended given PCF <360 L/min.

Nutrition: Referral to ALS Dietitian for baseline nutritional assessment. BMI trending down — high-calorie diet counseling initiated. PEG/RIG timing discussion deferred but recommended before FVC <50%. Patient does not require PEG at this time.

Speech / Swallowing: Referral to ALS Speech-Language Pathologist (SLP) for dysarthria management, swallowing evaluation (modified barium swallow if concern increases), and augmentative/alternative communication (AAC) planning. Recommend voice banking via ModelTalker or VocaliD now while voice quality is adequate.

Physical / Occupational Therapy: PT referral for gait assessment, right AFO (ankle-foot orthosis) fitting for right foot drop. OT referral for adaptive equipment for ADLs (buttonhooks, weighted utensils, voice-activated home controls). Aquatic therapy may be considered.

Multidisciplinary ALS Clinic: Patient enrolled in Northgate Multidisciplinary ALS Clinic (every 3 months). Team includes Neurology, Pulmonology, Nutrition, SLP, PT, OT, Social Work, and Palliative Care.

Palliative / Advance Care Planning: Goals of care conversation initiated. Patient expressed desire for aggressive intervention at this time. Advance directive and POLST forms provided. Referral to ALS social worker ([Sample Social Worker], LCSW) for psychosocial support and resource navigation. Caregiver assessment initiated.

Clinical Trials: Patient is potentially eligible for: (1) HEALEY ALS Platform Trial — currently enrolling; (2) Tofersen compassionate use if SOD1+ (not applicable here); (3) Investigational antisense oligonucleotide trials — screening initiated. Research coordinator contact provided.

Support Resources: Referral to ALS Association local chapter. Introduced to Citizen Health platform for medical record organization, community connection, and research participation. Prescription Assistance Program information provided for Riluzole.

FOLLOW-UP

Patient to return to Northgate ALS Multidisciplinary Clinic in **3 months (April 2025)**. LFTs in 4 weeks for Riluzole monitoring. Patient and wife (primary caregiver, [Sample Caregiver]) were present for full discussion. Ample time provided for questions. Both expressed understanding of diagnosis and treatment plan. Emotional support offered; referral to ALS social worker placed today.

Electronically signed by: **Dr. [Sample Neurologist], MD, PhD** Associate Professor of Neurology Northgate ALS and Motor Neuron Disease Program Date: January 9, 2025 — 4:47 PM PST

Report dictated: January 9, 2025 Report finalized: January 10, 2025 Document ID: NNG-NEURO-20250109-0000000 This report has been sent to: • Dr. [Sample Physician] (PCP) • Patient (via Patient Portal)