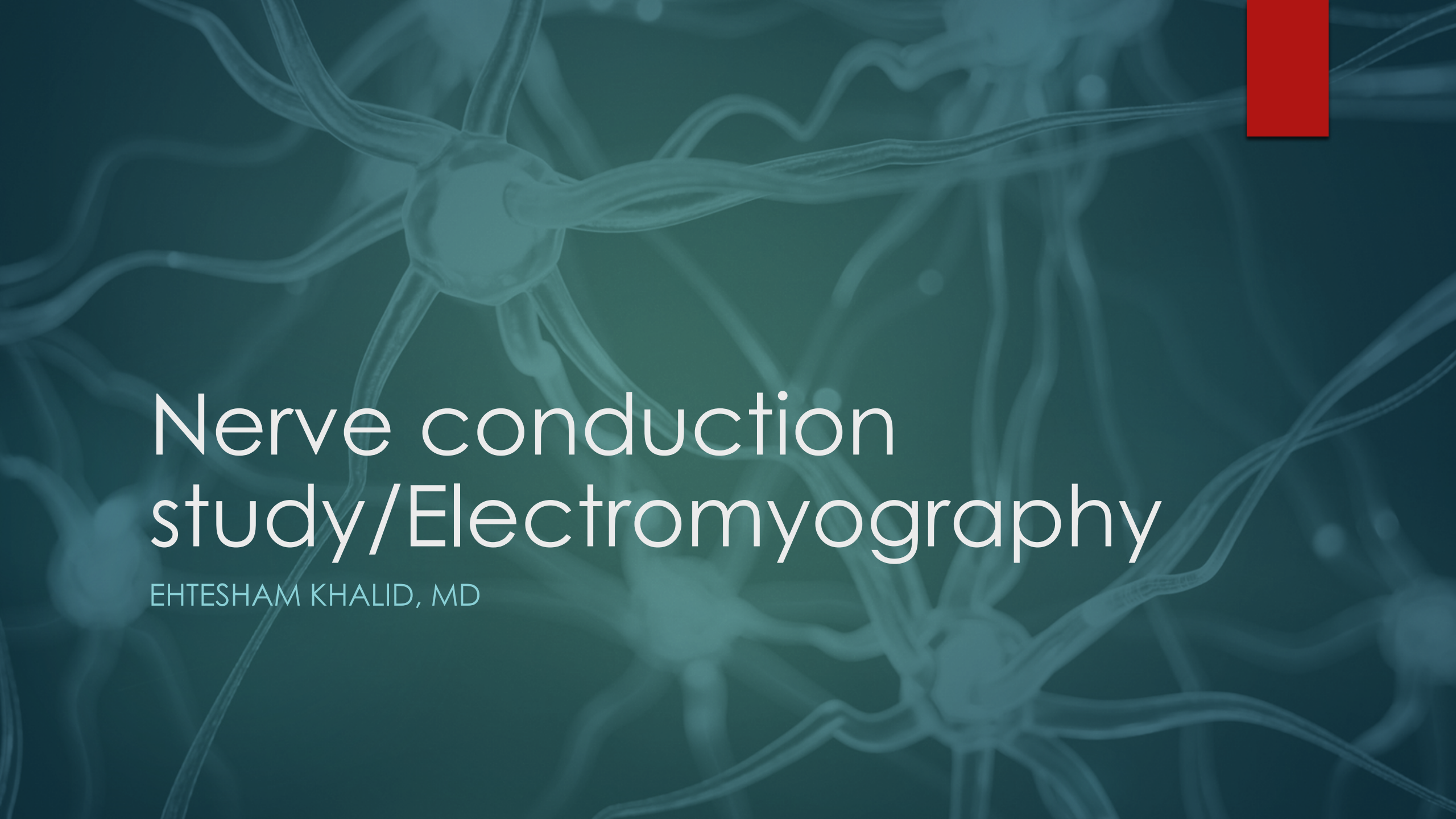




Nerve conduction study/Electromyography

EHTESHAM KHALID, MD



Disclosure

I have no financial disclosure regarding this presentation.

Case 1

- ▶ 33 y.o. female who noticed difficulty with singing and breathing in February 2017. She was born full-term without any need for breathing assistance at the time of birth. She started walking in the age of 7 month, but she was noted to be walking on her toes. She used to do yoga and a lot of walking, but she has never played any active physical sports during high school.
- ▶ She noticed difficulty with dorsiflexing her feet (left more than right) in the spring of 2017. She also noticed some tingling sensation in her toes. There are no symptoms in her hands.
- ▶ She was never diagnosed with scoliosis but she has history of high feet arches. There is strong family history of weakness but nobody ever had a genetic test. Male members are more severely affected than female members of the family.

Sensation:

- Impaired Vibration up to mid foot on right and up to ankle on left side.
- Impaired Pin prick up to ankle bilaterally.

DEEP TENDON REFLEXES:

- 2/2 biceps bilaterally.
- 2/2 triceps bilaterally.
- 0/1 brachioradialis bilaterally.
- 2/2 patellar bilaterally.
- 0/0 ankle bilaterally.
- Flexor plantar responses bilaterally.

Gait & Station:

- Normal casual and difficulty with tandem gait . She can stand with support on toes but she can't stand on heels.

Skeletal features:

- High feet arches.
- Deformed appearance of feet
- Shin muscles are moderately hypertrophic
- No scoliosis

	Right	Left			Right	Left
Arm abduction	5	5		Hip Flexion	5	5
Elbow Flexion	5	5		Knee Flexion	5	5
Elbow Extension	5	5		Knee Extension	5	5
Wrist Flexion	5	5		Dorsiflexion	3	3
Wrist Extension	5	5		Plantar Flexion	5	5
Finger Abduction	-4	+4				
Finger Flexors	5	5				
Finger Extensors	5	5				
APB	5	5				
Neck Flexion	5	5		Toe Extension	2	0
Neck Extension	5	5				

SNC

Nerve / Sites	Rec. Site	Onset Lat ms	Peak Lat ms	NP Amp μ V	PP Amp μ V	Segments	Distance cm	Velocity m/s	Temp. °C
L Sural - Ankle (Calf)									
Calf	Ankle	NR	NR	NR	NR	Calf - Ankle	14	NR	30.4
Ref.			≤ 4.40	≥ 6.0	≥ 6.0	Ref.		≥ 40	
L Ulnar - Digit V (Antidromic)									
Wrist	Dig V	3.33	4.79	10.8	24.6	Wrist - Dig V	14	42	30.7
Ref.			≤ 3.50	≥ 10.0	≥ 15.0	Ref.		≥ 50	
L Radial - Anatomical snuff box (Forearm)									
Forearm	Wrist	2.66	3.44	10.4		Forearm - Wrist	10	38	31.1
Ref.			≤ 2.90	≥ 15.0		Ref.		≥ 50	
R Sural - Ankle (Calf)									
Calf	Ankle	NR	NR	NR	NR	Calf - Ankle	14	NR	
Ref.			≤ 4.40	≥ 6.0	≥ 6.0	Ref.		≥ 40	

MNC

Nerve / Sites	Muscle	Latency ms	Amplitude mV	Rel Amp %	Duration ms	Segments	Distance cm	Lat Diff ms	Velocity m/s	Temp. °C
L Peroneal - EDB										
Ankle	EDB	NR	NR	NR	NR	Ankle - EDB	9			32.5
Ref.		≤ 6.50	≥ 2.0			Ref.				
Fib head	EDB	NR	NR	NR	NR	Fib head - Ankle		NR		32.5
Ref.						Ref.			≥ 44	
Pop fossa	EDB	NR	NR	NR	NR	Pop fossa - Fib head		NR		32.5
L Tibial - AH										
Ankle	AH	7.50	1.6	100	6.35	Ankle - AH	8			32.5
Ref.		≤ 5.80	≥ 4.0			Ref.				
Pop fossa	AH	18.13	1.1	69.9	9.64	Pop fossa - Ankle	32.6	10.63	31	32.5
Ref.						Ref.			≥ 41	
L Ulnar - ADM										
Wrist	ADM	3.59	6.0	100	6.41	Wrist - ADM	7			31.3
Ref.		≤ 3.30	≥ 6.0			Ref.				
B.Elbow	ADM	7.24	5.4	90.1	7.71	B.Elbow - Wrist	16.4	3.65	45	31.5
Ref.						Ref.			≥ 49	
A.Elbow	ADM	9.11	4.6	85.4	7.08	A.Elbow - B.Elbow	9	1.88	48	31.9
L Peroneal - Tib Ant										
Fib Head	Tib Ant	4.90	2.9	100	12.55	Fib Head - Tib Ant	10			31.6
Pop fossa	Tib Ant	6.82	3.0	103	12.50	Pop fossa - Fib Head	8	1.93	42	31.5

R Tibial - AH										
Ankle	AH	10.52	0.9	100	4.01	Ankle - AH	8			
Ref.		≤5.80	≥4.0			Ref.				
Pop fossa	AH	19.69	0.9	107	4.37	Pop fossa - Ankle	35	9.17	38	
Ref.						Ref.			≥41	

F Wave

Nerve	F Lat ms	Ref. ms	M Lat ms	Ref. ms	F-M Lat ms	Min F Lat ms	Ref. ms	Min M Lat ms	Min F-M ms
L Peroneal - EDB	NR	≤58.0	NR	≤32.0	NR	NR	≤58.0	NR	NR
L Tibial - AH	69.7	≤58.0	6.8	≤32.0	62.9	26.0	≤58.0	6.8	19.0
L Ulnar - ADM	28.8	≤32.0	3.5	≤32.0	25.3	28.8	≤32.0	3.5	25.2
R Tibial - AH	25.0	≤58.0	10.5	≤32.0	14.5	25.0	≤58.0	10.5	14.5

EMG

EMG Summary Table														
			Spontaneous					MUAP						
Muscle	Nerve	Roots	IA	Fib	PSW	Fasc	Other	Recruit	Effort	Dur.	Amp	PPP	Comment	
L. Tibialis anterior	Deep peroneal (Fibular)	L4-L5	Increased	1+	1+	None	CRD	Reduced	N	N	Gr Incr	N	9 mV units	
L. Gastrocnemius (Medial head)	Tibial	S1-S2	Increased	1+	None	None	None	N	N	N	Gr Incr	N		
L. Vastus medialis	Femoral	L2-L4	N	None	None	None	None	N	N	N	Mod Incr	N		

GJB1 Mutation- CMT1X

IGHMBP2- VUS

Case 2

- ▶ 42 years old female with scoliosis, and remote cervical cancer s/p resection transferred with acute hypoxemic respiratory failure in the setting of community acquired pneumonia for evaluation of progressive weakness. She noted difficulty in swallowing and significant unexpected weight loss over the past 4 weeks. She was intubated in ED and mechanically ventilated due to poor O2 saturation, extubated and then re-intubated for hypercarbia. On examination, there is restriction of adduction of left eye and upward gaze restriction of right eye. DTRs are relatively brisk in upper and lower extremities with down going plantars. Strength UE: Prox 3/5, Elbow flex 4/5, Exten-2/5, FF 4/5, FE 2/5. Grip 4/5. Strength LE: Prox 3/5, Dist 4/5. Vibration sense is intact. Jaw jerk is relatively brisk and tongue movements are normal.

Nerve conduction study

Sensory NCS

Nerve / Sites	Rec. Site	Onset Lat ms	Peak Lat ms	Ref. ms	NP Amp μV	Ref. μV	Distance cm	Velocity m/s	Ref. m/s	Temp. °C	TC Vel m/s
L Ulnar - Digit V (Antidromic)											
Wrist	Dig V	1.81	2.88	≤3.10	15.5	≥17.0	11	61	≥50	30.7	67

Motor NCS

Nerve / Sites	Muscle	Latency ms	Ref. ms	Amplitude mV	Ref. mV	Segments	Distance cm	Velocity m/s	Ref. m/s	Temp. °C	TC Vel m/s
L Ulnar - ADM											
Wrist	ADM	2.19	≤3.30	7.6	≥6.0	Wrist - ADM	7			23.7	
B.Elbow	ADM	6.10		6.9		B.Elbow - Wrist	23	59	≥49	23.2	79
A.Elbow	ADM	7.94		5.9		A.Elbow - B.Elbow	10	55	≥49	22.9	76
						A.Elbow - Wrist				22.9	

Repetitive Stimulation

Rep Stim

Anatomy / Train	Rate Hz	Ampl. mV	Ampl 4-1 %	Fac Ampl %	Area mVms	Area 4-1 %	Fac Area %	Time
L Ulnar - (ULNAR)								
Baseline @ 1Hz	1	6.4	11.3	100	18.6	16	100	0:00:00
Baseline @ 3 Hz	3	5.1	20	79.3	18.0	26.1	96.9	0:00:37
Post Exercise @ 0:00	3	7.0	0.9	109	23.6	7.4	127	0:01:46
@ 0:30	3	7.0	9.8	109	26.2	10.5	141	0:02:19
@ 1:00	3	6.8	20.4	106	25.9	21.6	140	0:02:52
@ 2:00	3	6.4	22.7	99.2	23.8	26.1	128	0:03:55
@ 3:00	3	6.3	25.1	97.7	23.2	31.1	125	0:04:58
@ 4:00	3	6.1	20.9	95.7	22.0	28.5	119	0:06:02
@5:00	3	6.2	20.9	96	21.6	25.9	116	0:07:05
L Accessory (spinal) - (Accessory spinal)								
Baseline @ 1Hz	1	5.8	28.5	100	37.4	35.4	100	0:00:00
R Accessory (spinal) - (Accessory spinal)								
Baseline @ 1Hz	1	5.3	27.1	100	34.4	33	100	0:00:00
Baseline @ 3Hz	3	4.8	43.5	90.7	30.3	49.1	88.1	0:00:29
Post Exercise @ 0:00	3	7.0	46.2	132	50.3	51.7	146	0:01:46
@ 0:30	3	5.5	42.6	104	40.0	51.1	116	0:02:19
@ 1:00	3	5.8	47.1	110	39.8	54.5	116	0:02:52
@ 2:00	3	5.6	44.6	105	38.6	50.8	112	0:03:56
@ 3:00	3	5.0	41.3	95	36.5	52.4	106	0:04:59
@ 4:00	3	5.7	39.4	108	40.7	51.7	118	0:06:02
@ 5:00	3	5.4	43.3	102	39.5	54.5	115	0:07:05

Case 3

- ▶ 51 years old female with progressive muscle weakness. She was previously diagnosed with scapulo-humeral muscular dystrophy and spinocerebellar ataxia. Her husband noticed walking difficulty in 1994. In 2000, she felt difficulty in going upstairs. She has difficulty in raising arm above head since 2004. She has difficulty in getting up from chair since 2004. Her family noticed scanning speech in 2000. There is no h/o double vision, sleep disturbance, breathing and swallowing difficulties. There is no family h/o muscle disease. Pt has previous h/o steroids intake for 2-3yrs during 2004-5.
- ▶ On examination, mild facial weakness and unable to maintain labial seal. Mild neck flexion and extension weakness. Mild proximal +3/4 upper and 4/5 lower extremities weakness. Anterior calf muscle hypertrophy and posterior calf muscle atrophy. She cannot stand on heels with some dorsiflexion of toes. No winging of scapula
- ▶ CPK 127
- ▶ On NCS/EMG- evidence of chronic myopathic changes predominantly in proximal appendicular muscles.

Repetitive stimulation

Rep Stim

Anatomy / Train	Rate Hz	Ampl. mV	Ampl 4-1 %	Fac Ampl %	Area mVms	Area 4-1 %	Fac Area %	Time
L Ulnar - DMA								
Baseline @3Hz	3	8.7	7.6	100	24.4	9.5	100	0:00:00
Post Exercise @0:00	3	8.7	7.2	100	24.8	8.6	101	0:00:45
@ 0:30	3	9.0	4.5	103	24.0	5.9	98.4	0:01:24
@ 1:00	3	8.8	7.1	102	24.2	7.9	99	0:01:38
@ 2:00	3	9.1	7.4	104	24.6	8.9	101	0:02:02
@ 3:00	3	8.9	6.5	103	24.4	8.2	99.8	0:02:20
@ 4:00	3	9.0	6.5	104	24.7	8.4	101	0:02:35
@ 5:00	3	9.0	5.5	103	24.6	7.5	101	0:02:47

L Facial - Nasalis								
Baseline @3Hz	3	3.8	11.4	100	7.0	14.7	100	0:00:00
Post Exercise @0:00	3	3.7	15.2	99.7	7.1	18.6	101	0:00:34
@ 0:30	3	3.7	13.1	99	7.1	17.4	101	0:01:18
@ 1:00	3	3.8	15.3	102	7.5	20	107	0:02:09
@ 2:00	3	4.7	18.7	126	7.4	3.7	106	0:07:15
@ 3:00	3	4.8	16.9	129	7.6	26.5	109	0:07:41
@ 4:00	3	4.9	18	132	7.6	21.1	108	0:07:59

Genetic test result



RESULT: POSITIVE

One homozygous Pathogenic variant identified in DOK7. DOK7 is associated with autosomal recessive congenital myasthenic syndrome and fetal akinesia deformation sequence.

Additional Variant(s) of Uncertain Significance identified.

GENE	VARIANT	ZYGOSITY	VARIANT CLASSIFICATION
DOK7	c.1124_1127dup (p.Ala378Serfs*30)	homozygous	PATHOGENIC
AGRN	c.2653G>A (p.Ala885Thr)	heterozygous	Uncertain Significance
DAG1	c.815C>G (p.Pro272Arg)	heterozygous	Uncertain Significance
LAMB2	c.1971G>A (Silent)	heterozygous	Uncertain Significance
POMT2	c.242A>G (p.His81Arg)	heterozygous	Uncertain Significance
SYNE2	c.20338C>G (p.Arg6780Gly)	heterozygous	Uncertain Significance

Repetitive Nerve Stimulation (RNS)

- ▶ Concept of Repetitive stimulation and End plate potential:

Change of gears while driving car

- ▶ Maintain Body temperature
- ▶ Skip Pyridostigmine 4-12 hours prior to study.

Repetitive Nerve Stimulation

- ▶ Myasthenia gravis (Baseline CMAP may be low in 15%)
- ▶ Many LEMS patients are misdiagnosed as Myasthenia Gravis.
- ▶ Short duration exercise followed by Repetitive stimulation
- ▶ Sensitivity of RNS for diagnosing MG ranges from 53% -- 100% in generalized MG and 10% -- 17% for ocular MG

Disorders with Electrodecremental response

- ▶ Myasthenia gravis
- ▶ Motor neuron disease
- ▶ Denervating disorders (neuropathies, myopathies)
- ▶ Metabolic myopathies



Any
Questions

Thank You

SF EMG and RNS

Clinical Cases

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Associate Professor of Clinical Neurology
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Disclosure.

- I am a speaker for Alexion.
- I am a consultant for Argenx and UCB

Case 1

- 56-year-old right-handed Gentleman, truck driver, with 5-year h/o weakness and blurry vision.
- PMH: HTN, HLD, DM.
- Follows up with Rheumatology for “Polymyositis”.
- On Methotrexate and prednisone x 5 years. Lisinopril, atorvastatin 20mg.

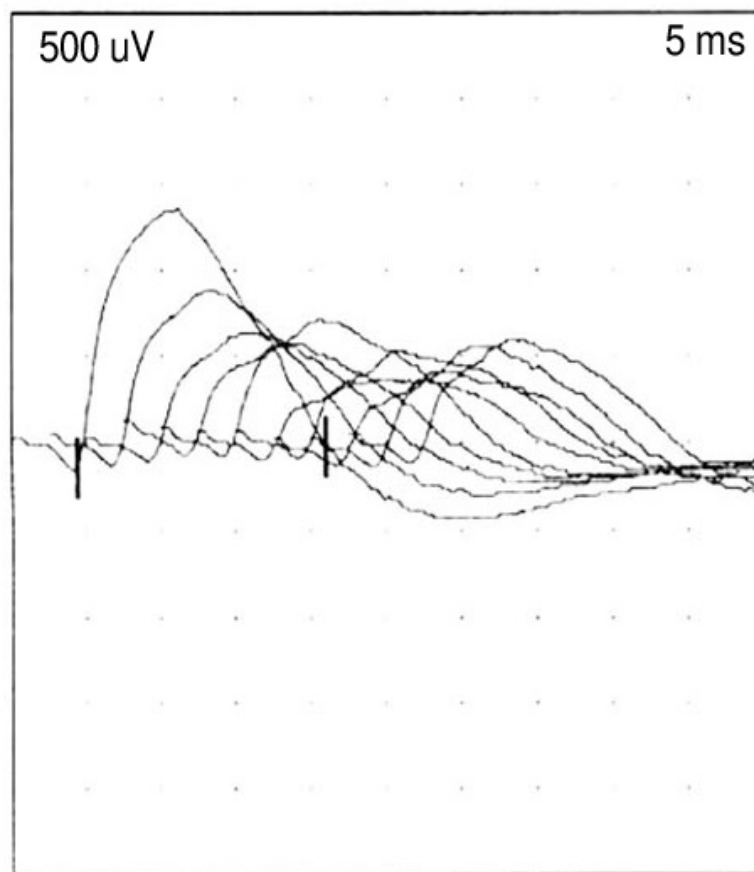
Labs:

- Acetylcholine ab (Binding, modulating, blocking), MUSK, LRP4: Negative
- HgA1c: 7.2.
- Myositis panel: negative.

Exam:

- Eye patch on his right eye. Fatigable EOMs and bifacial and bulbar weakness.
- Proximal weakness in UE and LE.
- Absent Achilles reflexes. Romberg +





Foot Switch: Ready / Stimulate Stop

Stim. Mode: Train / Single

Stim Freq:	2 Hz	No. in Train:	10
Stim Dur:	0.2 ms	Stim Rjct:	50 ms
Time:	11:34:54	Stop Watch:	0:07

Comment:

Pot No.	Peak Amp mV	Amp. Decr %	Area mVms	Area Decr %	Stim Level
1	1.50	0	13.90	0	43.1mA
2	1.03	31	9.38	33	43.1mA
3	0.75	50	7.18	48	43.1mA
4	0.66	56	6.56	53	43.1mA
5	0.80	47	8.10	42	43.1mA
6	0.49	67	4.83	65	43.1mA
7	0.63	58	6.40	54	43.1mA
8	0.55	63	4.84	65	43.1mA
9	0.70	53	6.07	56	43.1mA
10	0.71	53	6.63	52	43.1mA

ADL 10	Composite 15
<p>Talking: 1</p> <p>Double vision: 2</p> <p>Eyelid droop: 1</p> <p>Chewing: 1</p> <p>Swallowing: 1</p> <p>Breathing: Shortness of breath with exertion: 2</p> <p>Impairment in brushing teeth/hair: 1</p> <p>Impairment in ability to rise from chair: 1</p>	<p>Ptois/upward gaze: 1</p> <p>Double vision on lateral gaze: 3</p> <p>Eye closure: 1</p> <p>Talking: Intermittent slurring or nasal speech: 1</p> <p>Chewing: Fatigue with solid food: 1</p> <p>Swallowing: Rare episode of choking: 2</p> <p>Breathing: 2</p> <p>Neck flexion or extension: 1</p> <p>Shoulder abduction: 1</p> <p>Hip flexion: 2</p>

- Treatment: IVIG, CellCept 500mg BID, pred 5mg, ozempic, lisinopril, atorvastatin 20mg.

ADL 3	Composite 3
<p>Talking: 1</p> <p>Double vision: 0</p> <p>Eyelid droop: 0</p> <p>Chewing: 0</p> <p>Swallowing: 0</p> <p>Breathing: Shortness of breath with exertion: 0</p> <p>Impairment in brushing teeth/hair: 1</p> <p>Impairment in ability to rise from chair: 1</p>	<p>Ptosis/upward gaze: 0</p> <p>Double vision on lateral gaze: 0</p> <p>Eye closure: 0</p> <p>Talking: 0</p> <p>Chewing: 0</p> <p>Swallowing: 0</p> <p>Breathing: 0</p> <p>Neck flexion or extension: 1</p> <p>Shoulder abduction: 1</p> <p>Hip flexion: 2</p>

Case 2

- 58-year-old right-handed Lady with 1 year h/o progressive weakness and memory difficulty.
- Weakness reaching overhead. Ambulatory difficulty. Falling. Difficult to climb stairs.
- Progressively worsening memory difficulty.
- PMH: COPD on home oxygen. Uncontrolled DM(was on insulin, then metformin) with Diabetic neuropathy, HTN, HLD(was on statin), CAD, Liver non-alcoholic cirrhosis, portal
- S/H: 20-years h/o smoking.
- Patient has been worked up for PN and found to have severe axonal sensorimotor peripheral polyneuropathy.

Physical Exam:

- CN: Left incomplete Horner syndrome. Pupils reactive to light b/l.
- Weakness 4/5 bilateral deltoids, biceps:+4/5, triceps:4/5, HI: 5/5
- Hip flexion:-4/5, quads:+4/5, Feet DF: 5/5
- Prox LE weakness > Prox UE.
- Reflexes: +3 all over. No Babinski or clonus.
- Severe distal sensory loss. Romberg +++

Labs:

- Pancytopenia
- Low Ca, creatinine, albumin, protein

MRI Brain w Gad:

- No abnormal T2 or FLAIR signal is noted within the brain parenchyma.
- No abnormal enhancement following the administration of IV contrast.
- Mild generalized cortical atrophy is noted. Normal flow voids within major intracranial vessels.
- No foci of restricted diffusion. No foci of abnormal gradient susceptibility.

CT Chest w contrast:

- Patchy ground-glass opacities in the bilateral lung apices may reflect infectious/inflammatory process.
- Nodular contour of the liver compatible with cirrhosis. 1.9 cm right hepatic lobe hypodense focus is nonspecific.
- Splenomegaly and left upper quadrant collaterals consistent with portal hypertension. No ascites.
- Periportal/peripancreatic and retroperitoneal nonspecific lymphadenopathy.
- Sclerotic T6 vertebral body compression fracture.
- 1.5 cm right subcarinal lymph node.

CT ABDOMEN W WO CONTRAST :

- Lung bases: There is an area of scarring versus atelectasis seen within the lingula.
- Liver/gallbladder/biliary: There is a hypodense lesion within segment 7 of the liver that measures approximately 1.9 cm in size. The lesion appears to demonstrate some heterogeneous internal enhancement. This lesion does not have the classic appearance of a HCC.
- Findings are consistent with a LI-RADS 3 lesion based on CT.
- Spleen: The spleen is enlarged.
- Adrenals: There is calcification seen associated with the medial limb of the right adrenal gland that is unchanged in appearance.
- Pancreas, Kidneys and Bowel/Mesentery: Unremarkable.

Impression:

- Hypodense lesion within the right hepatic lobe with some heterogeneity internally within internal solid component not excluded. The lesion is classified as a LI-RADS 3 lesion. MRI may be helpful for further delineation of this lesion in this cirrhotic patient.

Rec: APB Stim: Wrist, Elbow

Switch: N-Rt
Stimulator: 1

Rate: 0

Level:

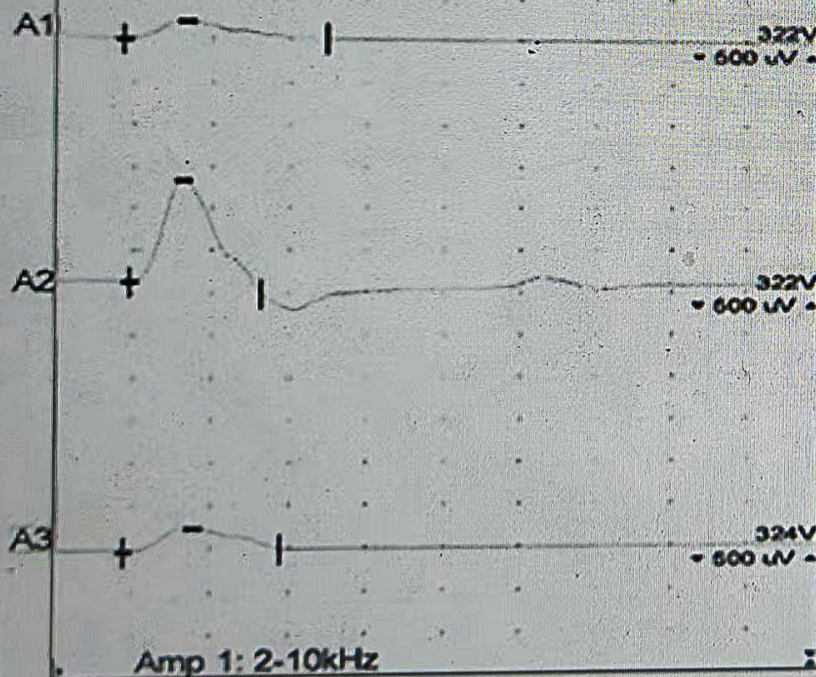
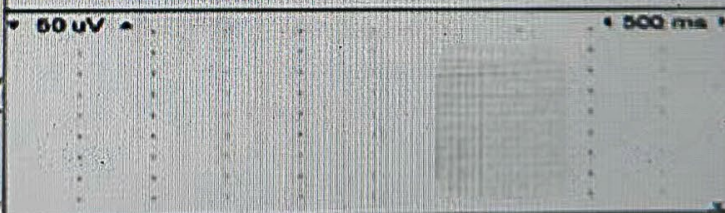
320 V 0.1 ms Single

Step: 3 Average: Off Sig. Enhancer: Off

Recording Site: APB

Stimulus Site	Lat ms	Dur ms	Amp mV	Area mVms	Temp SC
A1: w	4.3	13.0	0.2	1.3	
A2: 20 pe	4.7	8.4	1.2	4.2	
A3: 60 sec pe	4.4	10.0	0.3	1.8	

Segment	Dist mm	Diff ms	CV m/s
APB-w		4.3	
w-20 pe		0.4	
20 pe-60 sec pe		0.3	



Amp 1: 2-10kHz

Level: 25.0 mA

mV

5 ms

Foot Switch: Ready / Stimulate / Stop

Stim Mode: Train / Single

Stim Freq: 50 Hz No. in Train: 100

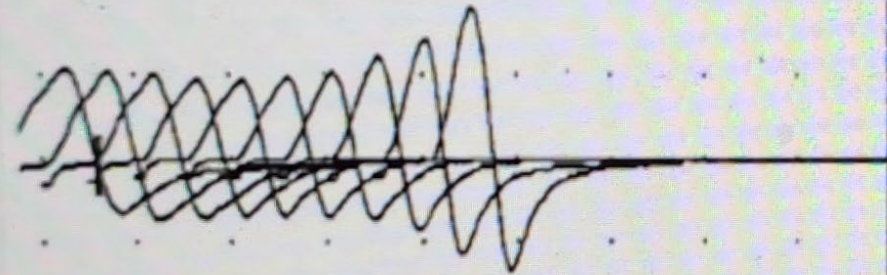
Stim Dur: 0.1 ms Stim Rct: 0.5 ms

Stim Site: Wrist

Time: 16:08:05 Stop Watch: 1:09

Comment:

Pol No.	Peak Amp mV	Amp Decr %	Area mVms	Area Decr %	Stim Level
1	6.00	0	16.60	0	25.0mA
2	6.23	-4	15.70	5	25.0mA
3	5.87	2	15.00	10	25.0mA
5	5.42	10	14.10	15	25.0mA
10	5.20	13	14.10	15	25.0mA
15	5.37	10	14.50	13	25.0mA
20	5.58	7	15.10	9	25.0mA
30	6.69	-12	17.80	-7	25.0mA
50	7.95	-33	21.50	-30	25.0mA
95	9.47	-58	25.40	-53	25.0mA



Jded

Stim: 50 Hz, 0.1 ms, #100

Vanda A. Lennon et al. Calcium-Channel Antibodies in the Lambert-Eaton Syndrome and Other Paraneoplastic Syndromes. N Engl J Med 1995;332:1467-1475

Rapidly progressive dementia–associated N-type voltage-gated calcium channel antibody encephalopathy

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ABSTRACT

Autoimmune encephalopathy is one of the treatable causes of rapidly progressive dementia; however, it is often underdiagnosed. Autoantibodies against voltage-gated calcium channel (VGCC) have been linked to several neurological disorders, including **Lambert-Eaton syndrome**, but VGCC antibody-associated encephalopathy is uncommon. Herein, we present a case of a 74-year-old woman with prominent neuropsychiatric symptoms followed by rapid cognitive decline. Extensive initial studies were nondiagnostic. Subsequently, serum N-type VGCC antibody was positive. After treatment with intravenous immunoglobulin, the patient's cognition and neuropsychiatric symptoms significantly improved.

KEYWORDS Autoimmune encephalopathy; N-type voltage-gated calcium channel; rapidly progressive dementia; VGCC encephalitis

Rapidly progressive dementias (RPDs) are generally defined as dementias that develop in <1 to 2 years after symptom onset.¹ Prion diseases, which are untreatable and associated with poor prognosis, are the prototypical etiology of RPDs. RPD secondary to autoimmune encephalopathies are treatable.² Moreover, voltage-gated calcium channel (VGCC) antibody is not a common autoantibody causing autoimmune encephalopathy. Herein, we present a case of RPD with positive serum N-type VGCC antibody, which presented with prominent psychiatric symptoms followed by rapidly progressive cognitive decline and significantly improved after immunotherapy.

CASE REPORT

A previously healthy 74-year-old, right-handed black woman presented with a 6-month history of progressive cognitive decline and behavioral changes. She had 12 years of

short-term memory problem and inattention. She was brought to the emergency department by a police officer after she was found wandering around a street. She had no history of psychiatric illness or substance abuse and no known family history of dementia. Neurological examination showed no focal neurological deficits. There were no abnormal movements, extrapyramidal dysfunction, akinetic mutism, or visual impairment on the exam. During the bedside examination, she was verbally aggressive. She scored 16/30 on the Mini-Mental State Examination (*Table 1*). Apart from her low body mass index (17 kg/m²), her general physical examination was normal.

Laboratory investigations, including complete blood count, metabolic panel, urine drug screen, serum alcohol, HIV and syphilis screening, thiamin, vitamin B12, folate, thyroid-stimulating hormone, ammonia, erythrocyte sedimentation rate, C-reactive protein, C3, and C4, were within normal limits. Antinuclear antibody, rheumatoid factor,

SFEMG

- Jitter is the variability of arrival time of action potentials to the recording electrode between the consecutive discharges.
- 15–20 pairs from each muscle for voluntary activation and >20 spikes for stimulation studies.

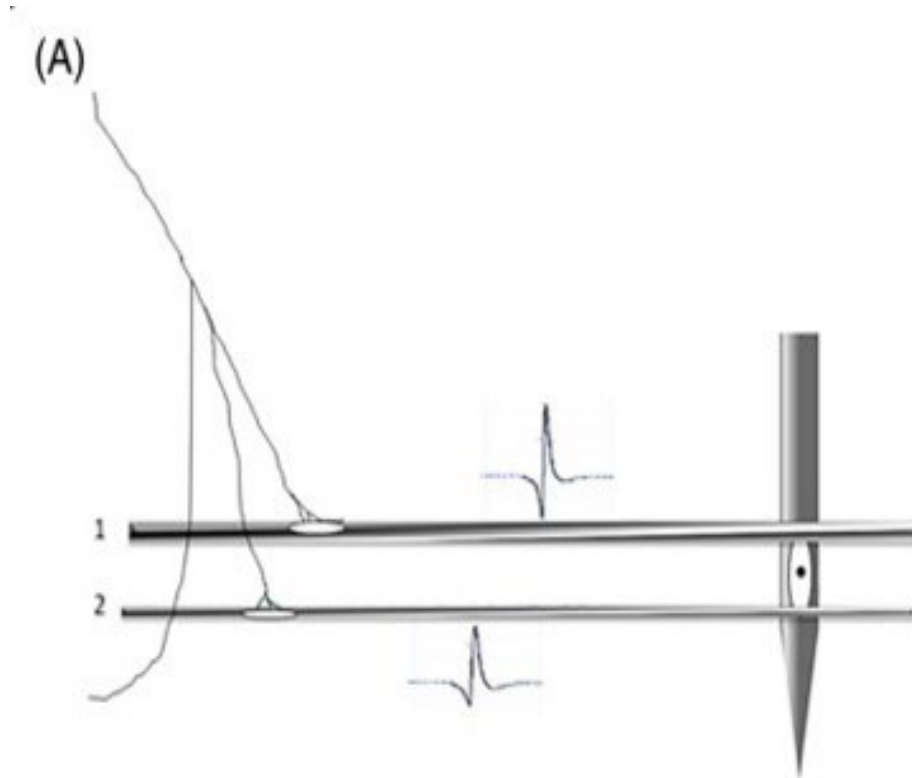
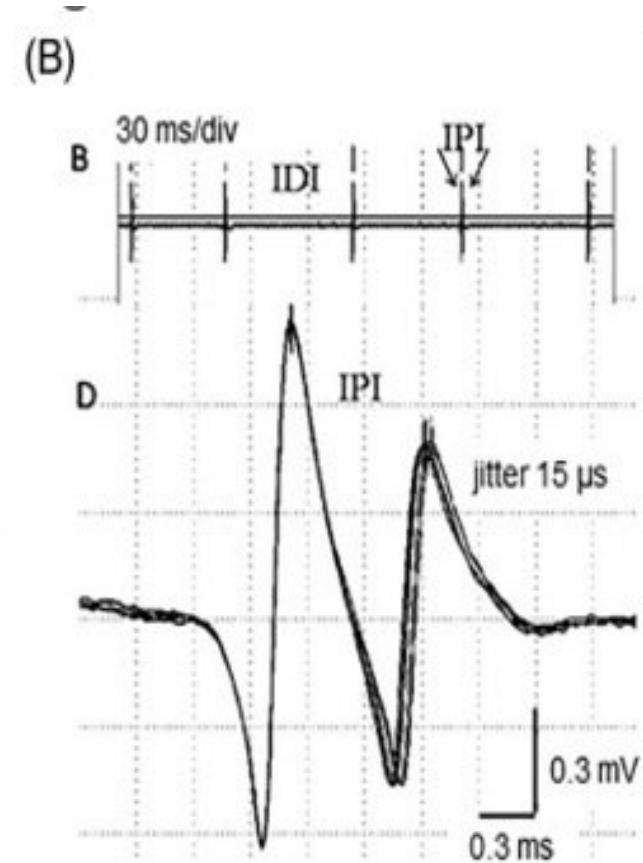
Voluntary activation using Concentric needle SFE

The SFE is positioned to record from a pair of muscle fibers innervated by the same motor axon.

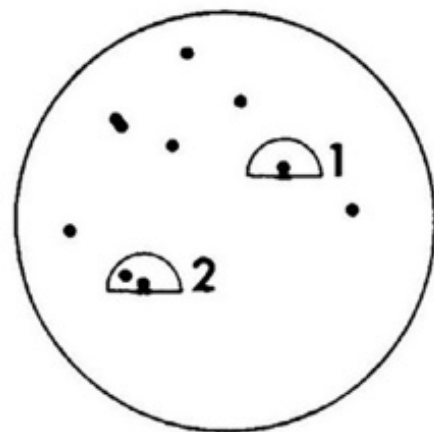
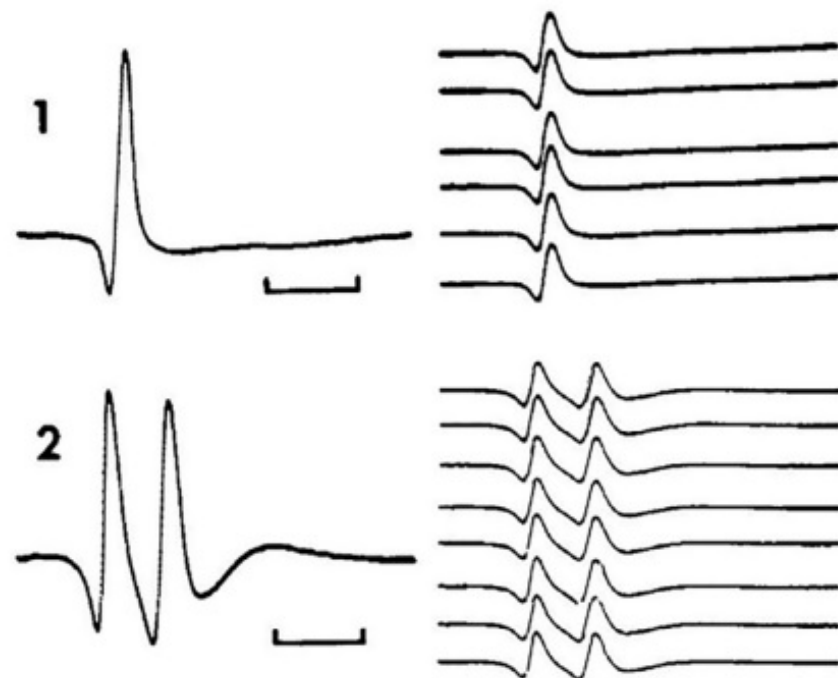
A, The action potential pair firing with slight voluntary effort, with the IDI and IPI indicated.

B, Several superimposed discharges demonstrate changes in the IPI (the neuromuscular jitter).

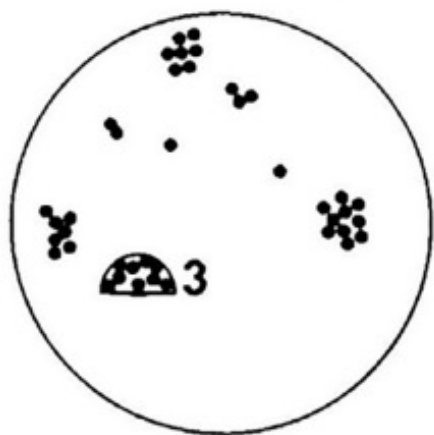
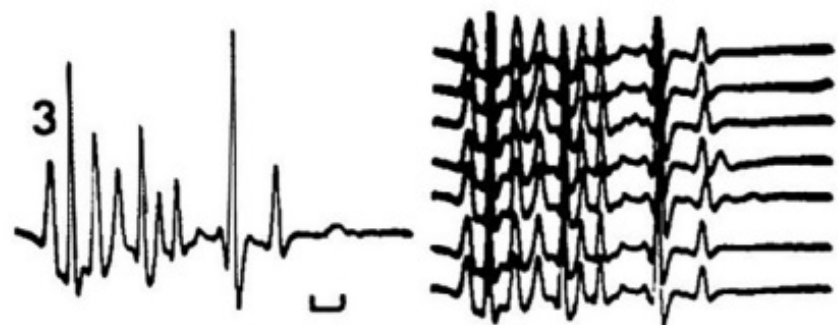
C, Rastered display of the two potentials in B at a higher sweep speed. The sweep is triggered by the first potential.



NORMAL



REINNERVATION



SFEMG:

- Amplitude $> 200\mu\text{V}$
- Rise time $< 300\mu\text{s}$,
- Duration 1ms
- Duration of a pair: 4ms.
- Radius: 300 μm .
- LFF: 500Hz, HFF: 10kHz.

Individual Jitter for single fiber pairs: 30 μs

- To assure adequate sampling of the muscle, recordings should include 3–4 skin insertions.

If the recording contains more than 2 spikes:

- Jitter values are calculated between the triggering spike, and each of the others.

Conduction block:

- When the neuromuscular junction conduction is impaired, jitter increases and some action potentials are lost.

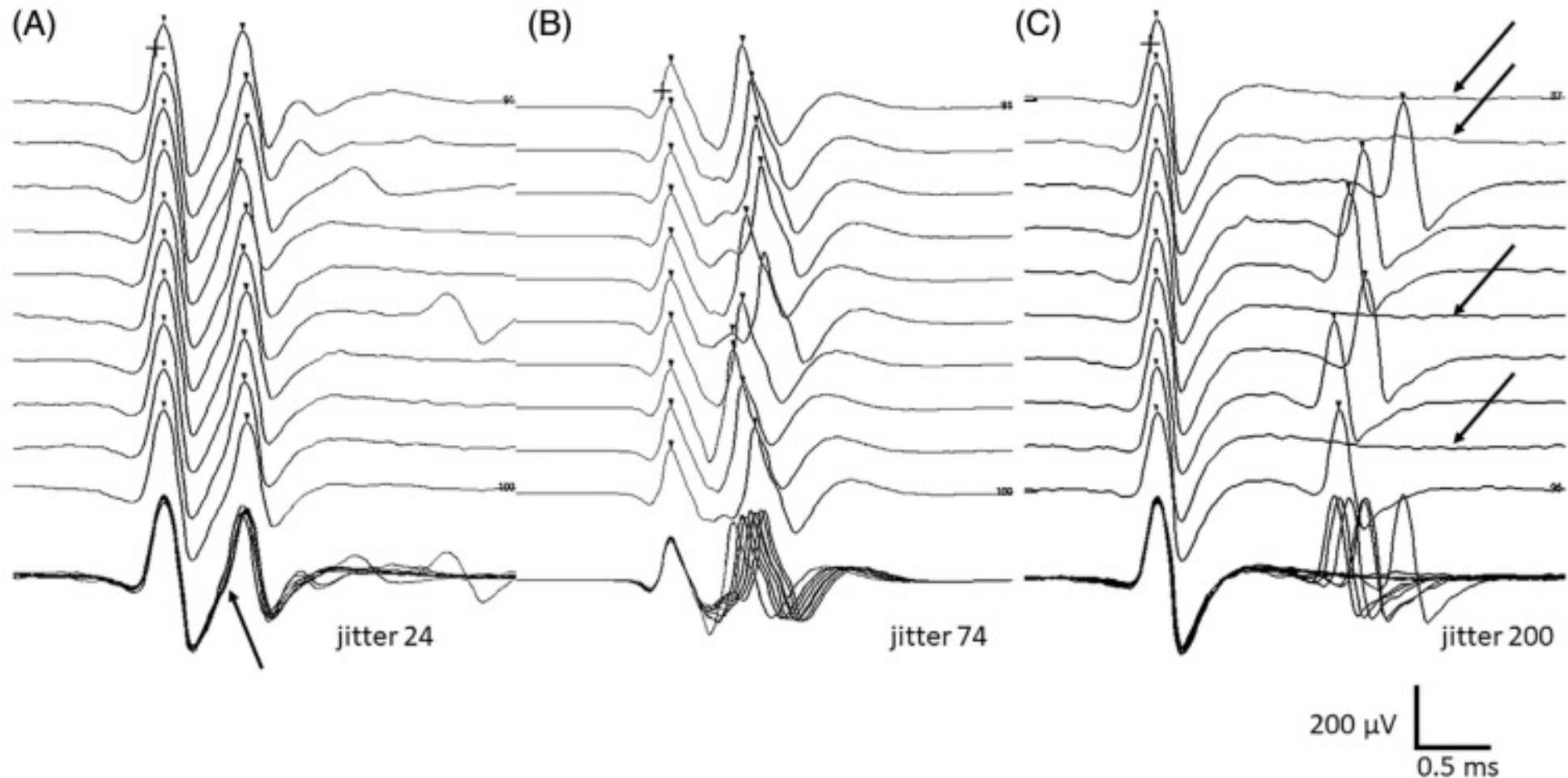
Mean Consecutive Difference:

- To assure adequate sampling of the muscle, recordings should be made from different portions of the muscle using 3–4 skin insertions.
- MCD is usually computed from a series of 50 responses from each endplate.
- A full study in a muscle should include MCD values for 30–40 different endplates (20 signal pairs in a voluntarily activated muscle represents 40 endplates).
- MCD varies among different muscles.
- Jitter can be normal in some endplates and increased in other (affected) endplates !

Jitter during voluntary activation using concentric needle electrodes.

A, Normal jitter B, Increased jitter C, Increased jitter with intermittent blocking.

The rising phase of the second spike in (A) seems to have an inflection (arrow), as a result of arising from a sloping baseline.



Factors causing increased Jitter

- Cooling and ischemia.
- Chronic denervation such as in SMA and PN where there is increased Fiber density + reduced strength.
- In PN, jitter normalizes after 1.5 years of reinnervation.

Case 3

67-year-old right handed lady with right hand weakness.

-

Exam:

- Weak neck flexion.4/5
- Droopy right eyelid, that does not worsen with fatiguing.
- EOMI. Do diplopia or blurry vision.
- R>L hand intrinsic weakness and wasting with fasciculations and split hand sign.
- LE: 5/5
- Hyporeflexia all over.

MRI C-spine: severe anterior disc bulge upon anterior cord.

- Mod C5-6 R and L FN
- Severe C6-7 right and moderate L FN
- C7-T1 : NL

! LCMC HBA1C EXT (NON-POCT)

Order: 126380412

Status: Edited Result - FINAL

! Newer results are available. Click to view them now.

Component	2 yr ago	2 yr ago	3 yr ago	3 yr ago	4 yr ago	4 yr ago	5 yr ago
Ref Range &	(12/23/21)	(6/23/21)	(12/19/20)	(6/13/20)	(11/23/19)	(6/8/19)	(10/13/18)
Units							

Hemoglobin	6.1 ^	5.9 ^	CM	6.0 ^	CM	5.7 ^	CM	6.0 ^	CM	5.9 ^	CM	6.6 ^	CM
A1C													

4.0 - 5.6 %

Comment: ADA Screening Guidelines:

5.7-6.4% Consistent with prediabetes

>or=6.5% Consistent with diabetes

High levels of fetal hemoglobin interfere with the HbA1C assay. Heterozygous hemoglobin variants (HbS, HgC, etc) do not significantly interfere with this assay.

However, presence of multiple variants may affect accuracy

- EXAMEND TIME:6/12/2023 03:02 PM

- CLINICAL HISTORY:

TECHNIQUE: LCMC MRI CERVICAL SPINE WITHOUT CONTRAST

COMPARISON: None available

FINDINGS:

There is straightening of the normal cervical lordosis. No evidence of aggressive osseous lesion or acute compression deformity. Mild loss of disc space height seen at the levels of C4-C7. Benign hemangioma involving the T2 vertebral body.

No evidence of cord signal abnormality.

C2-3: No significant central canal or foraminal stenosis.

C3-4: No significant central canal or foraminal stenosis.

C4-5: Mild disc bulge. Moderate central canal stenosis. Mild bilateral neural foraminal stenosis.

C5-6: Moderate disc bulge. Moderate central canal stenosis. Mild bilateral neural foraminal stenosis.

C6-7: Moderate disc bulge. Moderate central canal stenosis. Mild bilateral neural foraminal stenosis.

C7-T1: Mild disc bulge. Mild bilateral neural foraminal stenosis. No significant central canal stenosis.

Paraspinal soft tissues are unremarkable

Last

Specimen

Resulted: 06/13/23

Collected: 06/12/23

16:16

15:40

- **MRI Brain**

Impression

- No significant intracranial abnormality. No findings to suggest ALS.

Preliminary Report Dictated By: ALISA S TANAKIT, MD

Electronically Signed By: Douglas Casey, MD 6/12/2023 3:18 PM CDT

- **Narrative**

- EXAM END TIME: 6/12/2023 02:51 PM
CLINICAL HISTORY: G12.21 ALS (amyotrophic lateral sclerosis) (CMS/HCC). Motor neuron disease suspected ALS.

TECHNIQUE: LCMC MRI BRAIN WITHOUT CONTRAST

COMPARISON: None available

FINDINGS:

No acute infarct. No acute intracranial hemorrhage. No extra-axial fluid collection. No masses, mass effect, or midline shift.

Parenchymal and ventricular volumes are normal. No confluent parenchymal signal abnormality seen. Vascular flow voids are unremarkable.

Cranium is unremarkable. Orbits and orbital contents are normal. Paranasal sinuses are clear.

Sensory NCS

Nerve / Sites	Rec. Site	Onset Lat ms	Peak Lat ms	NP Amp µV	Segments	Distance cm	Velocity m/s
R Median, Ulnar - Digital Antidromic							
Median Wrist	D1	3.44	4.27	7.3	Median Wrist - D1	13	38
Median Wrist	D2	3.80	4.64	7.8	Median Wrist - D2	14	37
Median Wrist	D3	3.80	4.69	5.8	Median Wrist - D3	14.2	37
Ulnar Wrist	D5	NR	NR	NR	Ulnar Wrist - D5	11	NR
L Median, Ulnar - Digital Antidromic							
Median Wrist	D1	3.07	3.75	9.3	Median Wrist - D1	11.5	37
Median Wrist	D2	3.49	4.48	6.4	Median Wrist - D2	13.6	39
Median Wrist	D3	3.85	4.58	3.7	Median Wrist - D3	14	36
Ulnar Wrist	D5	2.76	3.49	6.7	Ulnar Wrist - D5	10.5	38
R Radial - Anatomical snuff box (Forearm)							
Forearm	Wrist	2.24	2.92	16.0	Forearm - Wrist	10	45
R Sural - Ankle (Calf)							
Calf	Ankle	3.80	4.64	4.9	Calf - Ankle	14	37

Motor NCS

Nerve / Sites	Muscle	Latency ms	Amplitude mV	Amp % %	Duration ms	Area mVms	Segments	Distance cm	Lat Diff ms	Velocity m/s
R Median - APB										
Wrist	APB	5.99	5.4	100	8.13	25.4	Wrist - APB	7		
Elbow	APB	11.67	5.4	100	8.65	26.3	Elbow - Wrist	25.7	5.68	45
L Median - APB										
Wrist	APB	5.10	6.6	100	7.29	24.4	Wrist - APB	7		
Elbow	APB	10.16	4.9	73.8	7.66	21.3	Elbow - Wrist	24	5.05	48
R Ulnar - ADM										
Wrist	ADM	3.70	7.4	100	7.81	32.4	Wrist - ADM	7		
B.Elbow	ADM	8.18	7.0	94.7	8.18	31.3	B.Elbow - Wrist	20.2	4.48	45
A.Elbow	ADM	10.68	6.2	84	8.18	27.9	A.Elbow - B.Elbow	14	2.50	56
L Ulnar - ADM										
Wrist	ADM	3.07	7.4	100	6.20	23.9	Wrist - ADM	7		
B.Elbow	ADM	7.24	7.1	96.4	6.72	25.3	B.Elbow - Wrist	20.4	4.17	49
A.Elbow	ADM	10.16	6.3	85.4	6.98	23.2	A.Elbow - B.Elbow	13.7	2.92	47
L Ulnar - FDI										
Wrist	FDI	3.85	10.8	100	6.41	32.4	Wrist - FDI			
R Ulnar - FDI										
Wrist	FDI	5.00	5.0	100	6.67	17.6	Wrist - FDI			

EMG

EMG Summary Table		Spontaneous				MUP Recruitment					
Muscle		Fib	PSW	Fasc	Other	#	Rate	Polys	Dur	Amp	Effort
R. First dorsal interosseous		None	None	1+	.	4					
R. Abductor pollicis brevis		None	None	2+	.	4					
R. Abductor digiti minimi (manus)		None	None	1+	.	5					
R. Extensor digitorum communis		None	None	1+	.	2-3	Increased	None	mildly inc	Normal	Max
R. Deltoid		None	None	1+	.	Normal	Normal	None	Normal	Normal	Max
R. Tongue		None	None	None	.	Normal	Normal	None	Normal	Normal	.
R. Sternocleidomastoid		None	None	None	.	Normal	Normal	None	Normal	Normal	Max
R. Vastus lateralis		None	None	1+	.	Normal	Normal	None	Normal	Normal	Max
R. Tibialis anterior		None	None	1+	.	Normal	Normal	None	Normal	Normal	Max
R. Gastrocnemius		None	None	1+	.	Normal	Normal	None	Normal	Normal	Max

F Wave

Nerve	Fmin ms
R Median - APB	0.00
R Ulnar - ADM	24.95
L Median - APB	39.43
L Ulnar - ADM	36.20

Sensory NCS

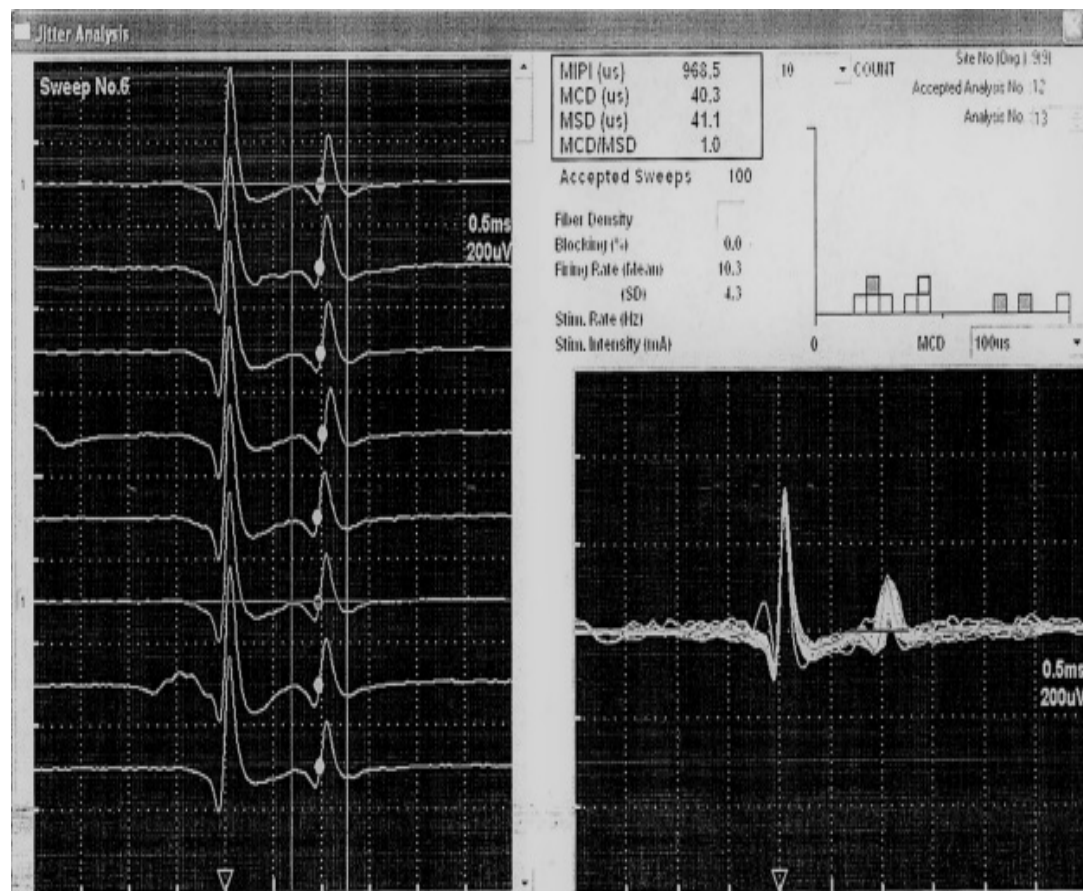
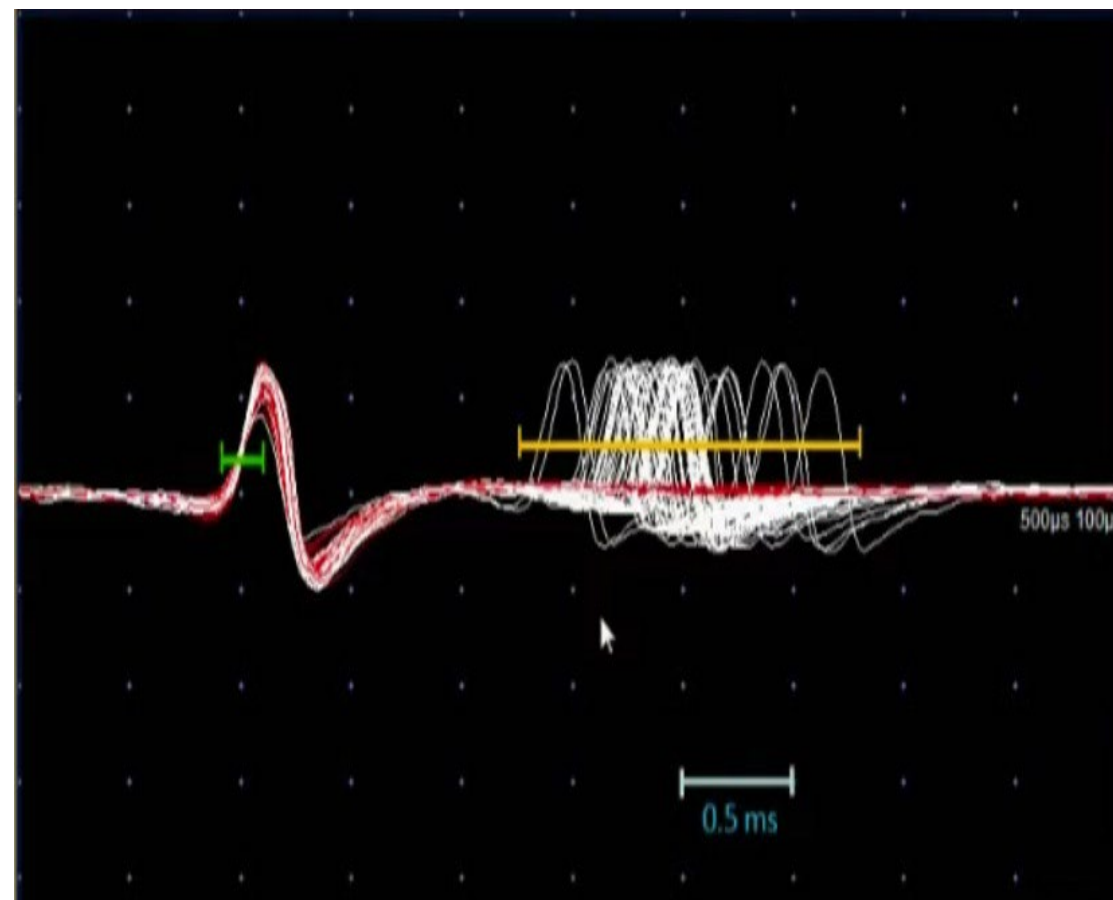
Nerve / Sites	Rec. Site	Onset Lat ms	Peak Lat ms	NP Amp µV	Segments	Distance cm	Velocity m/s
R Median, Ulnar - Digital Antidromic							
Median Wrist	D1	3.59	4.27	7.3	Median Wrist - D1	12	33
Median Wrist	D3	3.80	4.74	7.2	Median Wrist - D3	14.2	37
Ulnar Wrist	D5	NR	NR	NR	Ulnar Wrist - D5	11	NR
L Median, Ulnar - Digital Antidromic							
Median Wrist	D1	2.92	3.59	8.4	Median Wrist - D1	10.5	36
Median Wrist	D2	3.54	4.32	9.0	Median Wrist - D2	13.3	38
Median Wrist	D3	3.96	4.69	4.7	Median Wrist - D3	13.7	35
Ulnar Wrist	D5	2.86	3.49	7.2	Ulnar Wrist - D5	11.6	40
R Radial - Anatomical snuff box (Forearm)							
Forearm	Wrist	2.19	3.02	23.2	Forearm - Wrist	10	46
L Radial - Anatomical snuff box (Forearm)							
Forearm	Wrist	2.14	2.76	17.4	Forearm - Wrist	11.5	54
R Sural - Ankle (Calf)							
Calf	Ankle	NR	NR	NR	Calf - Ankle	14	NR
L Sural - Ankle (Calf)							
Calf	Ankle	NR	NR	NR	Calf - Ankle	14	NR

Motor NCS

Nerve / Sites	Muscle	Latency ms	Amplitude mV	Amp % %	Duration ms	Area mVms	Segments	Distance cm
R Median - APB								
Wrist	APB	6.82	4.1	100	8.39	19.7	Wrist - APB	7
L Median - APB								
Wrist	APB	4.84	6.8	100	6.88	23.9	Wrist - APB	7
R Ulnar - ADM								
Wrist	ADM	3.49	7.6	100	8.54	31.7	Wrist - ADM	7
L Ulnar - ADM								
Wrist	ADM	3.07	7.3	100	6.56	26.9	Wrist - ADM	7
R Peroneal - EDB								
Ankle	EDB	4.32	4.8	100	6.67	18.1	Ankle - EDB	8
L Peroneal - EDB								
Ankle	EDB	4.38	4.3	100	6.82	16.9	Ankle - EDB	8

EMG

EMG Summary Table										
Muscle	Spontaneous				MUP Recruitment					
	Fib	PSW	Fasc	Other	#	Rate	Polys	Dur	Amp	Effort
L. First dorsal interosseous	None	None	3+	.	Normal	Increased	4+	Increased	Increased	Max
L. Abductor pollicis brevis	None	None	3+	CRD	4	Increased		Increased	Increased	Max
L. Extensor digitorum communis	None	None	2+	.	3-4	Increased	2+	Increased	Increased	Max
L. Deltoid	None	None	None	.	2	Increased	3+	Increased	Increased	Max
R. First dorsal interosseous	None	None	1+	.	5					
R. Abductor pollicis brevis	None	None	3+	.	4					
R. Extensor digitorum communis	None	None	None	.	Normal	Increased	3+	Increased	Increased	Max
R. Deltoid	None	None	None	.	1-2	Increased	3+	Increased	Increased	Max
R. Vastus lateralis	None	None	None	.	Normal	Normal	3+	Increased	Normal	Max
R. Tibialis anterior	None	None	1+	.	1-2	Increased	2+	Increased	Increased	Max
R. Peroneus longus	None	None	None	.	Normal	Normal	None	Normal	Normal	Max
R. Gastrocnemius	None	None	None	.	Normal	Normal	None	Normal	Normal	Max
R. Tongue	None	None	None	.	Normal	Normal	None	Normal	Normal	
R. Sternocleidomastoid	None	None	None	.	Normal	Normal	None	Normal	Normal	Max





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Original article

O'Sullivan–McLeod syndrome: Unmasking a rare atypical motor neuron disease



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ABSTRACT

Atypical motor neuron disease represents a rare heterogeneous group of neurodegenerative disorders with clinical, genetic and neuroimaging features distinct from those of the classic spinal or bulbar-onset amyotrophic lateral sclerosis (ALS). O'Sullivan–McLeod syndrome represents an extremely rare lower motor neuronopathy with early adult-onset distal amyotrophy and weakness in the upper limbs with asymmetrical involvement. To add to the few case series and epidemiological and genetic studies describing this variant syndrome, our team here presents a series of seven unrelated Brazilian patients with O'Sullivan–McLeod syndrome in a detailed review of their clinical, neuroimaging, laboratory and neurophysiological findings. A male-to-female ratio of 2.5 to 1 and a mean age at onset of 34.3 years was observed, with a mean time delay of 6.6 years between symptom-onset and a definitive diagnosis. A positive family history was observed in one case, yet whole-exome sequencing results were negative. Neuroimaging studies were unremarkable. All cases presented with chronic denervation restricted to cervical myotomes and normal sensory nerve conduction studies. This case series, one of the largest groups of patients with O'Sullivan–McLeod syndrome reported in the literature, confirms the sporadic nature of the condition and the difficulties faced in arriving at a definite diagnosis, and also expands the age limit in late adult-onset cases.

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- In MND, loss of innervation is partially compensated by peripheral sprouting from intact motor units.
- SF EMG shows the fine parameters of MUP, increase in fiber density and jitter, that indicate reinnervation.
- Fiber Density is calculated by counting the activity of other muscle fibers belonging to the same motor unit, triggered by the trigger potential during activation.

- Variation in the waveform of MUP is called jiggle, observed in the early stages of reinnervation.
- Using concentric needle: MUP peak number indirectly reflect changes in fiber density.
- High jitter values and high peak number in extra-extremity muscles becomes even more important for the differential diagnosis of ALS.

Thank you.

