Clinical neurophysiology of other hyperkinetic movement disorders

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International Federation of CLINICAL NEUROPHYSIOLOGY

International Parkinson and Movement Disorder Society

Objectives

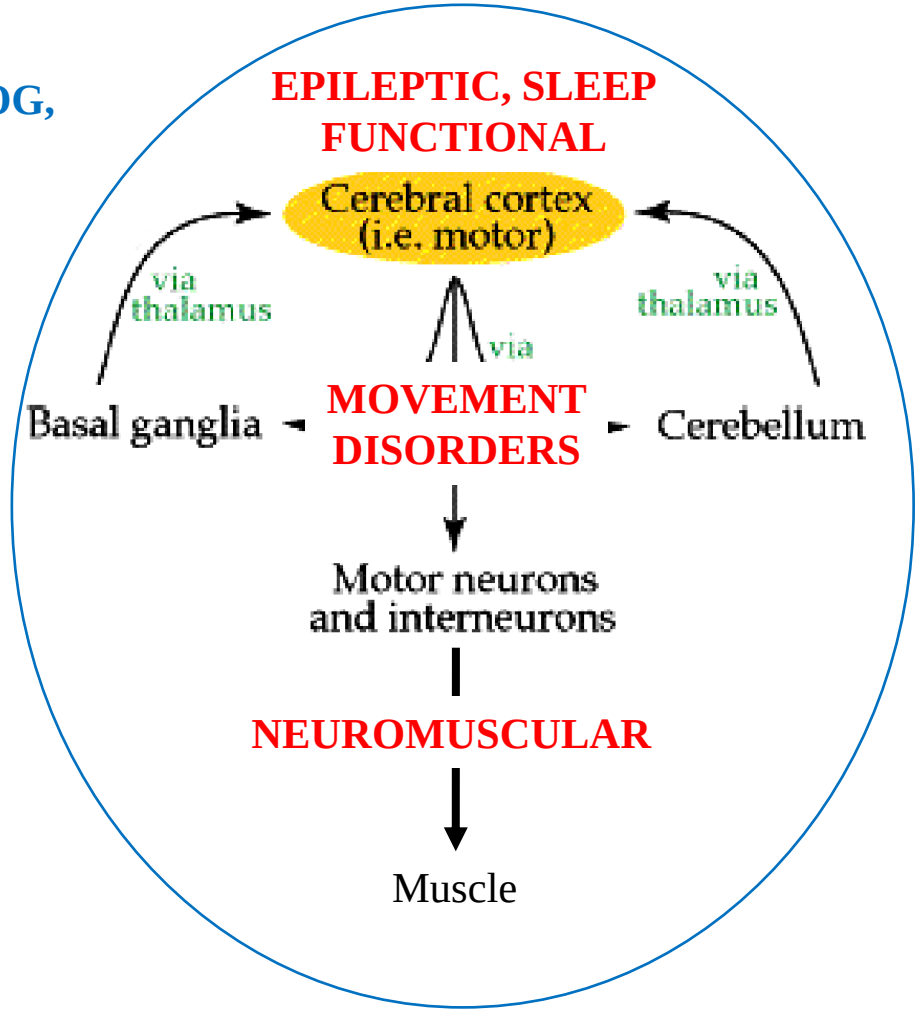
- Identify common neurophysiological patterns underlying disorders of movement of peripheral origin.
- Recognize neurophysiological patterns underlying peripherally induced disorders of movement.

DISORDERS OF MOVEMENT

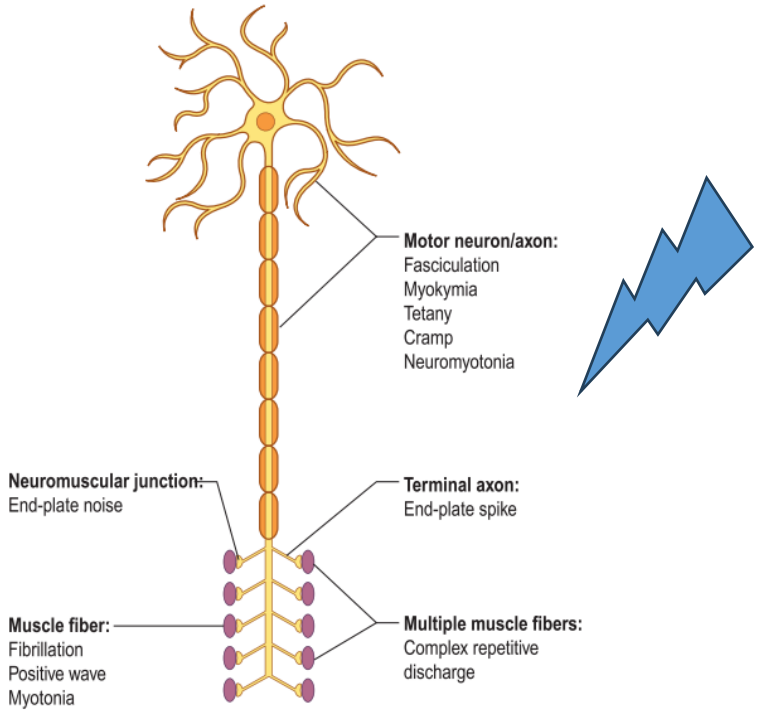
EEG (PSG), ECOG, TMS, etc.

LFPs, MER, etc.

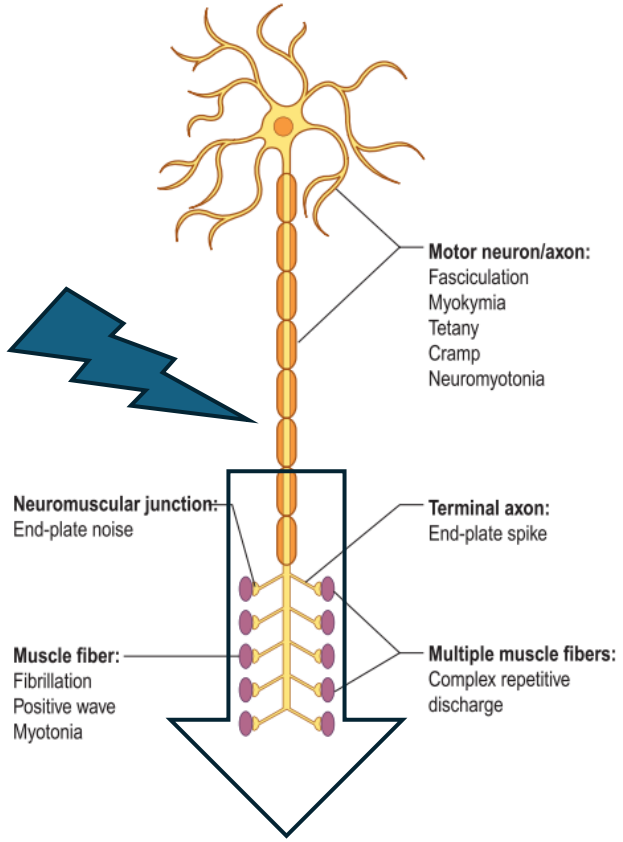
EMG, NCS, etc.



CLINICAL NEUROPHYSIOLOGY



Peripheral disorders of movement



Movement Disorders
CLINICAL PRACTICE

CLINICAL VIGNETTES

Abnormal Movements of the Back Heralding Amyotrophic Lateral Sclerosis

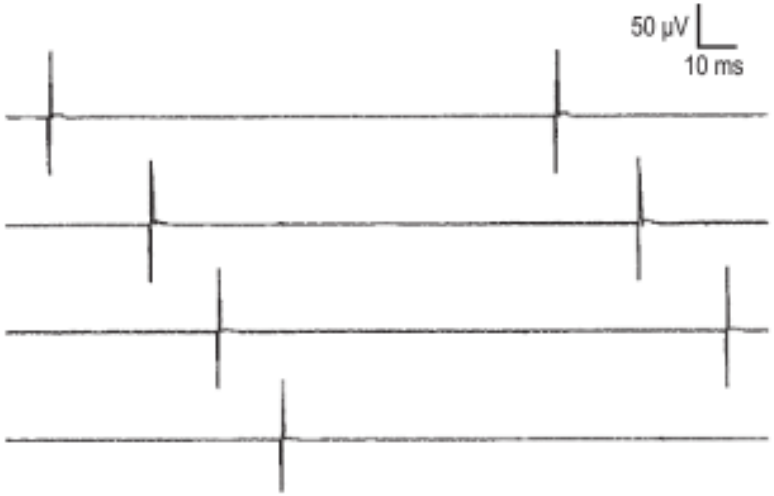
ABG: Chronic, compensated respiratory insufficiency (appropriate for 4300 m.a.s.l.)

Muscle examined	Spontaneous activity	Motor unit action potentials	Recruitment pattern
Right gastrocnemius	Increased insertional activity, 1+ fibrillations, 2+ positive sharp waves, occasional fasciculations	1+ increased amplitude and duration	Moderately reduced
Right tibialis anterior		2+ increased amplitude, 1+ increased duration and polyphasia	Severely reduced
Right vastus medialis	None		Moderately reduced
Right biceps femoris	Increased insertional activity, 1+ fibrillations, 1+ positive sharp waves		
Right thoracic paraspinals (T10-12)		1+ increased amplitude, duration and polyphasia	Unable to asses
Right rectus abdominis		1+ polyphasia	Severely reduced
Right latissimus dorsi	Increased insertional activity, 1+ fibrillations, 1+ positive sharp waves, occasional fasciculations	1+ increased amplitude, duration and polyphasia	
Right first dorsal interosseus			Moderately reduced
Right deltoid			
Rigth upper trapezius	1+ fibrillations		

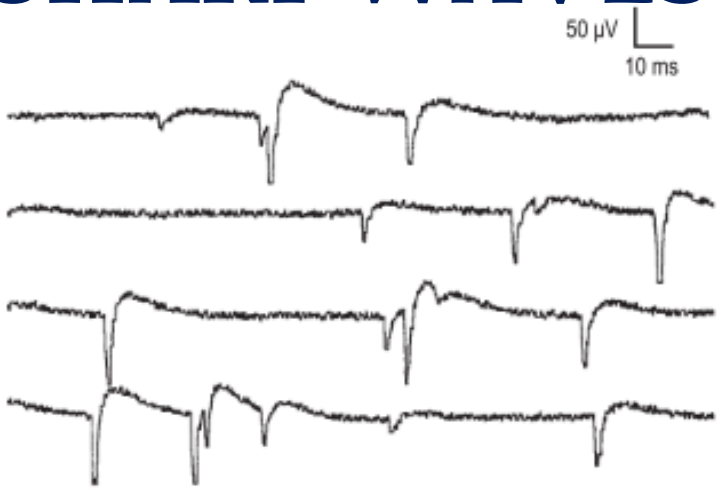
Corcoran, *et al*. Mov Disord Clin Pract. 2021; 430:120028

MUSCLE FIBER DEPOLARIZATION

FIBRILLATIONS

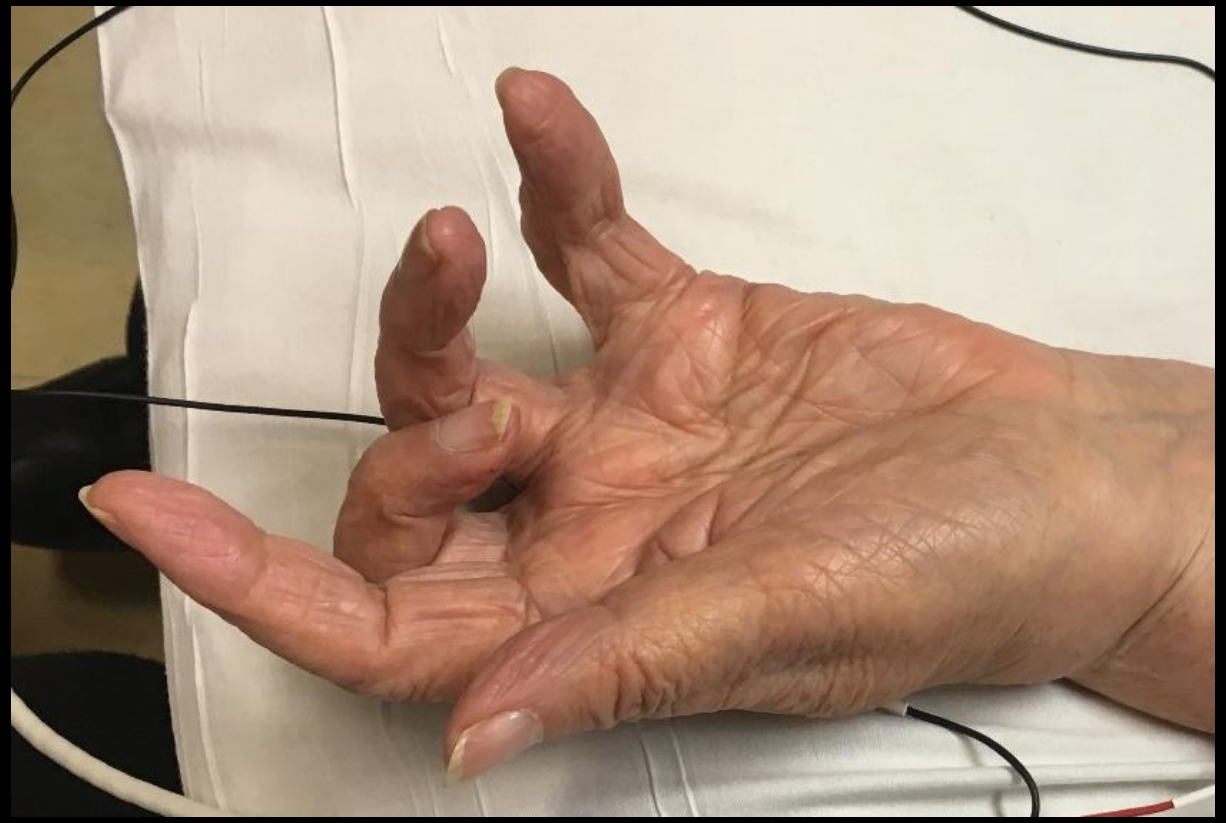


POSITIVE SHARP WAVES



NO MOVEMENT

- 76-year-old woman
- Abnormal flexion of left-sided fingers 3-to-5
- Progressively worsening
- Attempts to open the hand are painful
- Idiopathic focal *dystonia*
- Botulinum toxin injections



MUSCLE FIBER DEPOLARIZATION

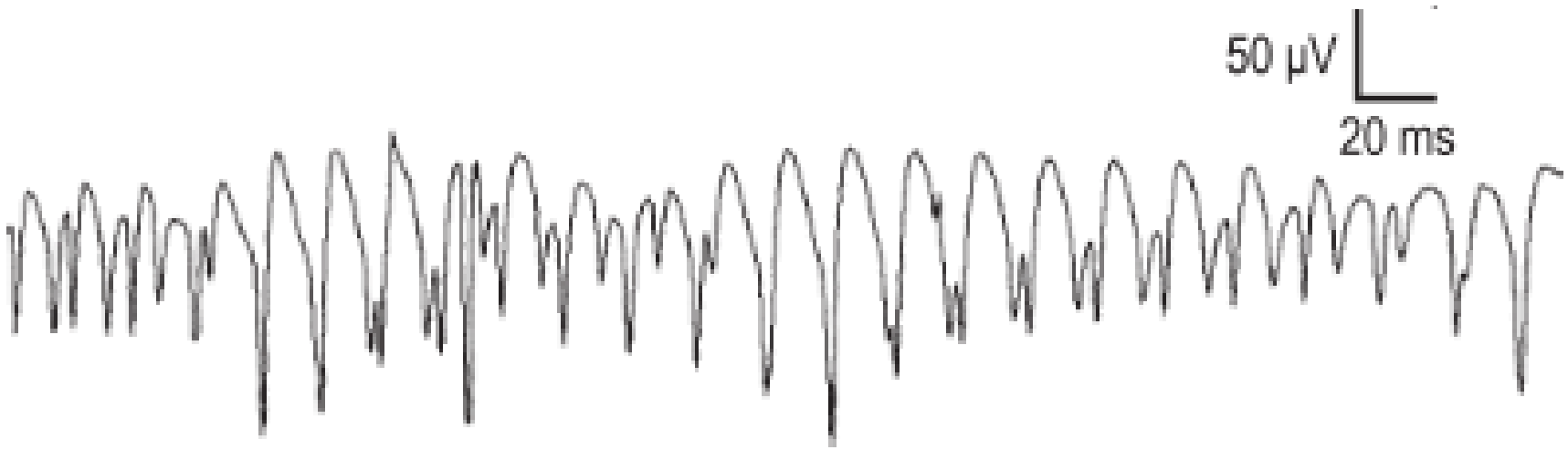
CRDs

NO MOVEMENT

- Precipitants: eating ice cream, cold temperatures, exertion (“hates gym class”)
- Some episodes during afternoon naps after playing in the park

What else would you look for on neurological examination?

ELECTROMYOGRAPHY



MYOTONIC DISCHARGES

- Myotonic dystrophy
- Myotonia congenita
- Paramyotonia congenita
- Hyperkalemic periodic paralysis
- Acid maltase deficiency
- Polymyositis
- Myotubular myopathy

MYOTONIC DISORDERS (Non-dystrophic)

	Myotonia Congenita	Paramyotonia Congenita	Other Sodium Channel Myotonia
Gene	*CLCN1*	*SCN4A*	*SCN4A*
Chromosome	7q35	17q23	17q23
Clinical features	Myotonia	Myotonia, episodic weakness	Myotonia
Triggers	Cold (some patients)	Cold	Potassium (some patients)
Acute treatment	n/a	n/a	n/a
Chronic treatment	Mexiletine, phenytoin, procainamide	Mexiletine, phenytoin, procainamide	Mexiletine, phenytoin, procainamide, acetazolamide
Exercise testing	Short exercise test (SET): Postexercise decrement, rapid return to baseline	SET: Postexercise decrement, facilitated by repetition or cold	SET: Often nondiagnostic
Laboratory features	n/a	n/a	n/a

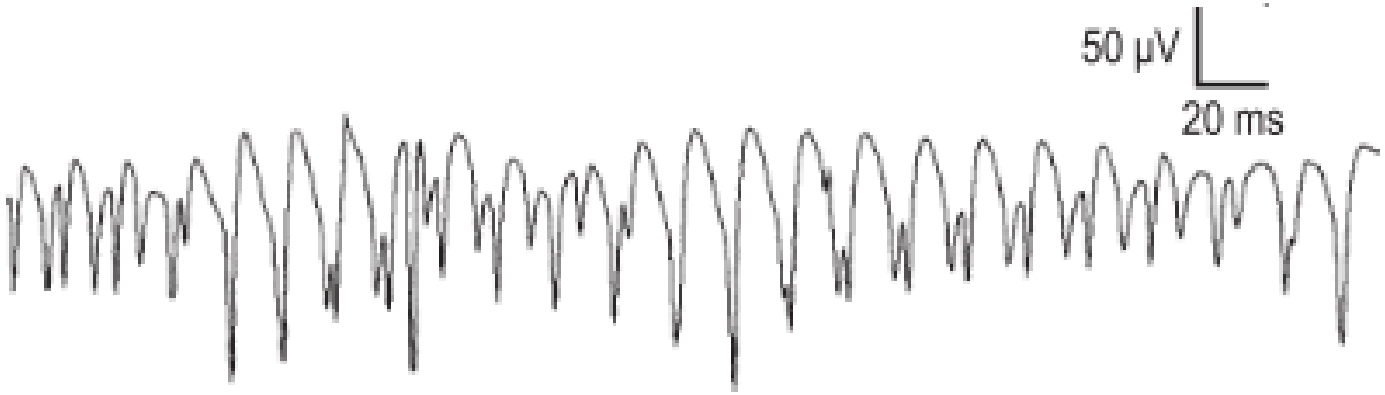
Statland & Barohn, Continuum 2013

Pathogenic heterozygous mutation in **SCN4A** gene (c.3938C>T, p.Thr1313Met)
→ **Paramyotonia congenita**

- Mexiletine – partial response
- **Acetazolamide** – significant improvement
- Trigger avoidance (cold temperature, excessive exertion, fruit juice [hyperK+])

Muscle Fiber Depolarization

MYOTONIA



Abnormal postures, relaxation, stiffening

≠ Dystonia

≠ Paroxysmal kinesigenic dyskinesia

REST OF EXAMINATION

- Weakness
  - FDIs: left 3/5, right 4-/5
  - EDCs: left 4-/5, right 4+/5
- Absent reflexes in BL upper limbs
- No cranial nerves, upper motor neuron or sensory abnormalities

NERVE CONDUCTION STUDIES

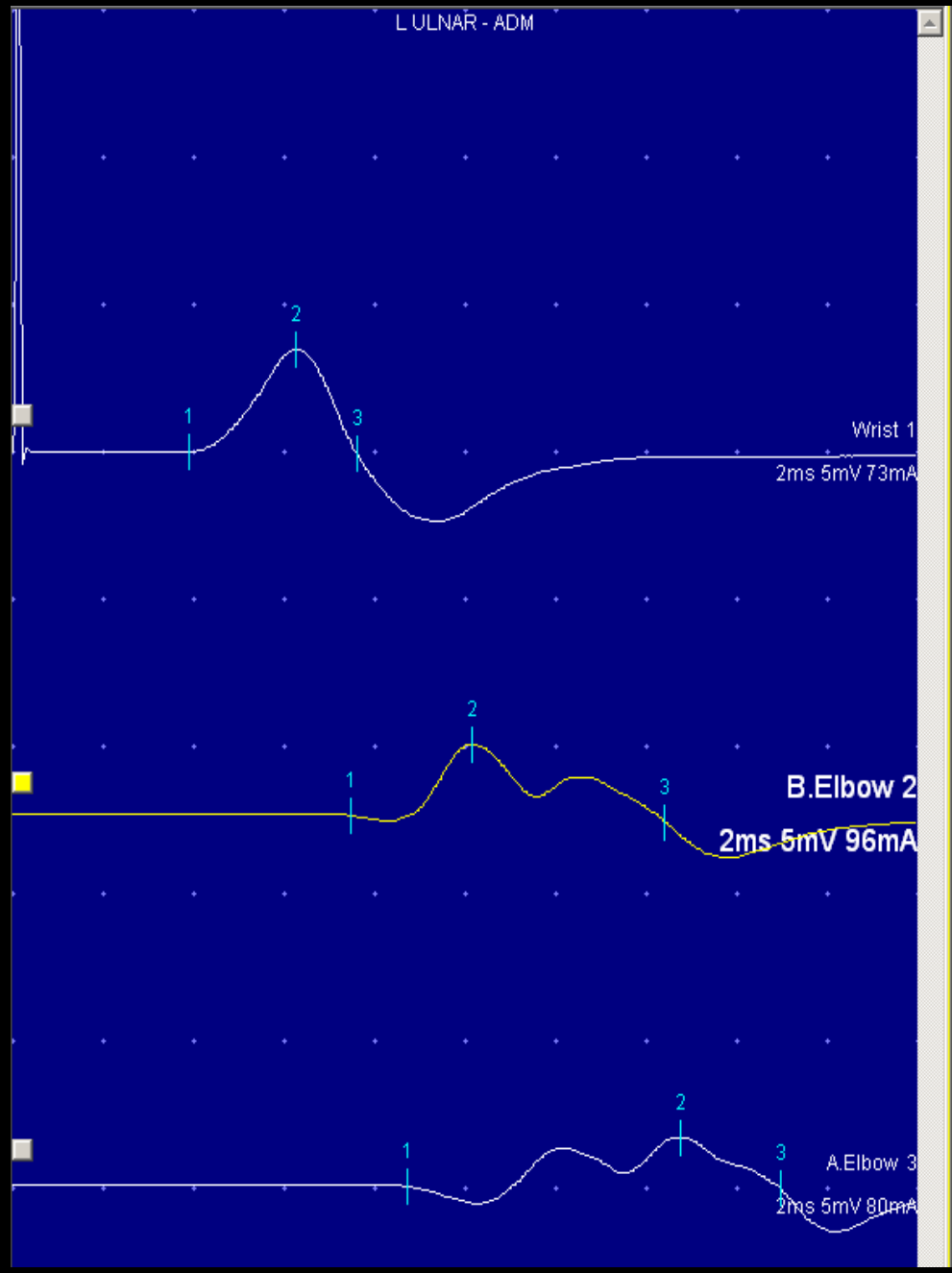
- Motor: Moderate-to-severe axonal neuropathy in bilateral upper extremities
- Left ulnar (forearm): *probable* conduction block (significant temporal dispersion)
- Bilateral lower extremities: Normal
- Sensory: Normal

NERVE CONDUCTION STUDIES

Motor NCS (Temp 32°C)		Lat. (ms)	Amp. (mV)	Rel. Amp. %
R ulnar (ADM)	Wrist	3.85	1.9	100
	B. elbow	8.55	1.5	76.3
	A. elbow	11.05	1.3	68.7
L ulnar (ADM)	Wrist	3.90	**3.5**	**100**
	B. elbow	7.45	2.5	70.7
	A. elbow	8.70	**1.7**	**48.6**

ELECTROMYOGRAPHY

- Bilateral FDIs and left EDC:
  - Moderate-to-severe ongoing denervation (3+ fibs/PSWs)
  - Reduced recruitment of chronically re-innervated units (2+ amp, duration & polyphasia)



L ULNAR - ADM

Sites	Rec. Site	Lat ms	Amp mV	Rel Amp %	SPAR %
Wrist	ADM	3.90	3.5	100	100
B.Elbow	ADM	7.45	2.5	70.7	100
A.Elbow	ADM	8.70	1.7	48.6	100
Axilla	ADM				
Erb's Pt	ADM				

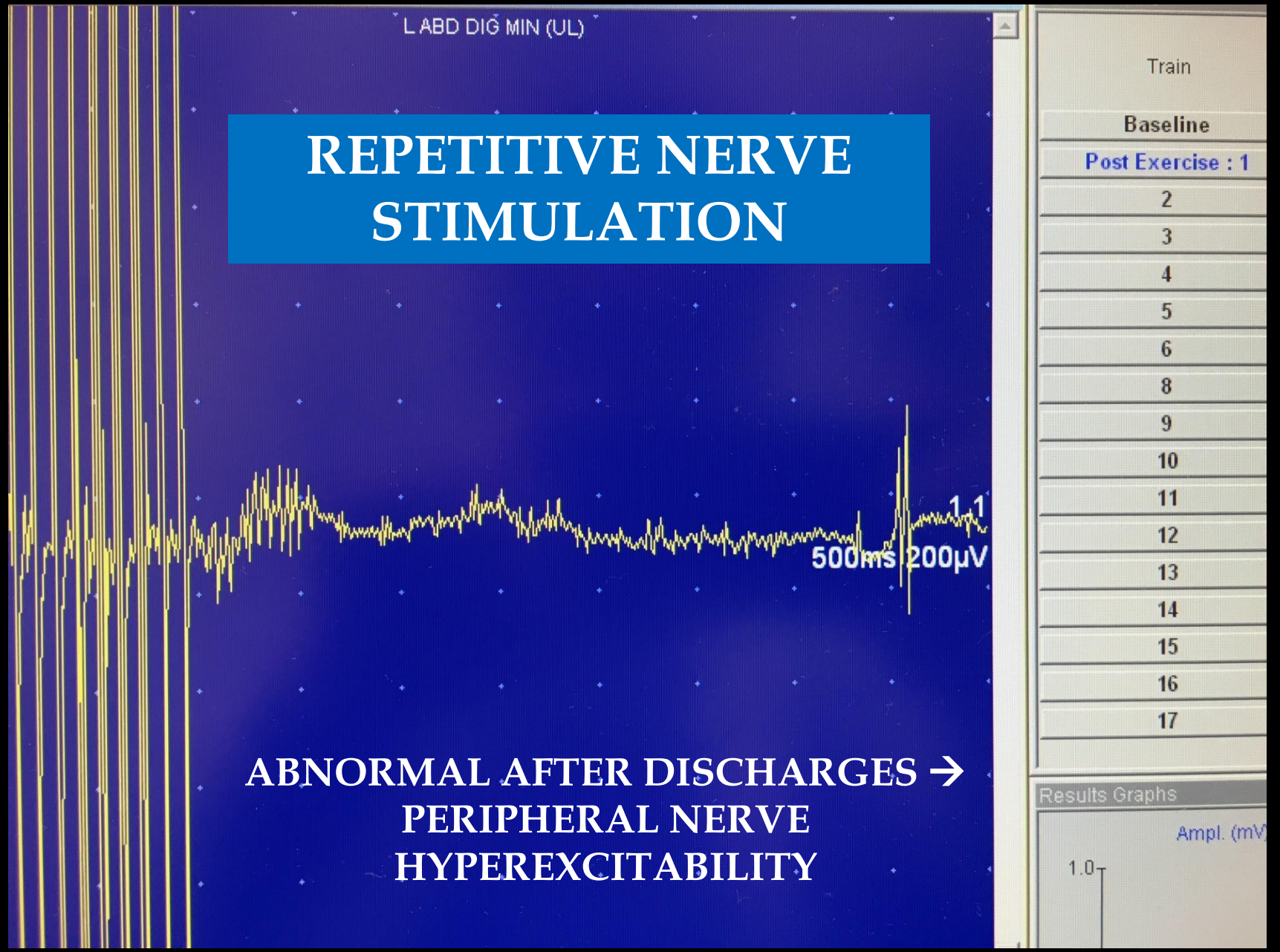
Segments	Dist. cm	Vel m/s	Temp °C
Wrist - ADM	7		20.5
B.Elbow - Wrist	23	64.8	20.8
A.Elbow - B.Elbow	10.5	84.0	20.8
Axilla - A.Elbow			
Erb's Pt - Axilla			

Test Folder

- Motor NCS
- Sensory NCS
- Carpal Tunnel Sensory Index (CSI)
- F Wave
- H Reflex
- Needle EMG
- Rep Nerve Stim
- Anomalous Innervetion
- CMAP with Exercise

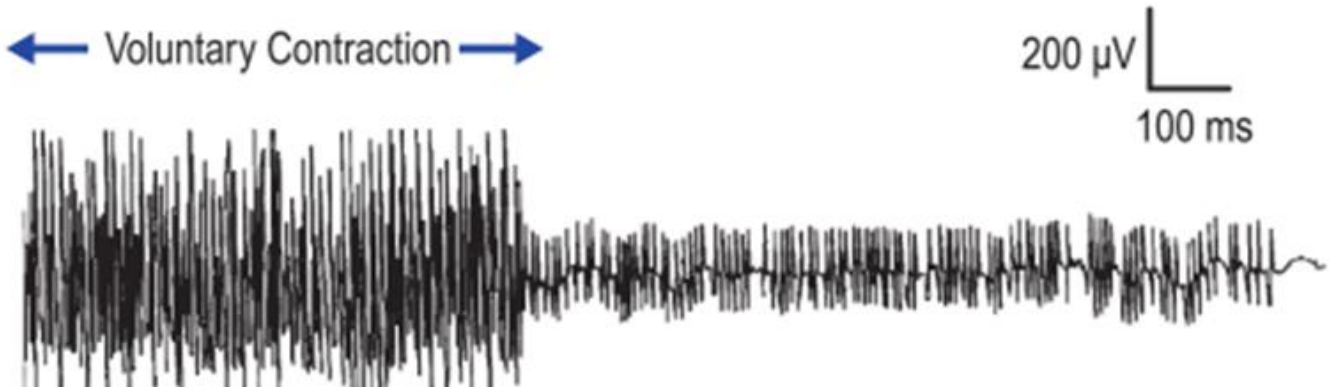
Test History

- Motor NCS R MEDIAN APB
- F Wave R MEDIAN
- Sensory NCS R MEDIAN Dig II Antidr
- Motor NCS L MEDIAN APB
- F Wave L MEDIAN
- Sensory NCS L MEDIAN Dig II Antidr
- Sensory NCS R ULNAR
- **Motor NCS R ULNAR ADM**
- Sensory NCS L ULNAR Dig V Antidr
- Motor NCS L ULNAR ADM


L ABD DIG MIN (UL)
REPETITIVE NERVE STIMULATION
1.1
500ms 200µV
ABNORMAL AFTER DISCHARGES →
PERIPHERAL NERVE HYPEREXCITABILITY
Train
Baseline
Post Exercise : 1
2
3
4
5
6
8
9
10
11
12
13
14
15
16
17
Results Graphs
Ampl. (mV)
1.0

MOTOR UNIT POTENTIALS

CRAMPS



POSTURES / STIFFENING

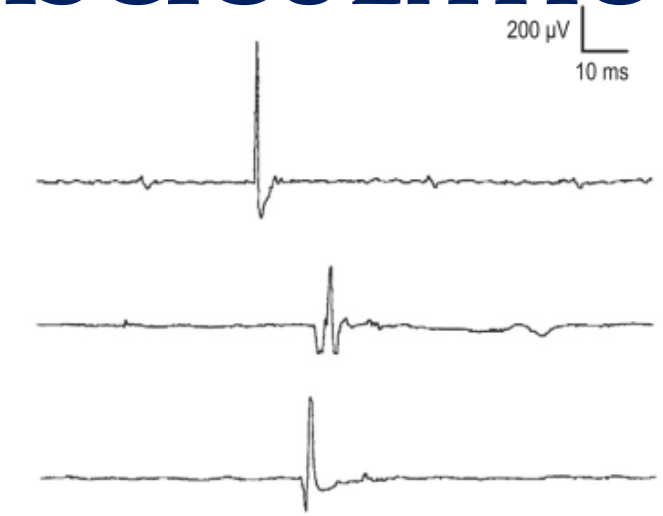
(ABNORMAL RELAXATION)

≠ DYSTONIA

≠ PAROXYSMAL KINESIGENIC DYSKINESIA

MOTOR UNIT POTENTIALS

FASCICULATIONS



FAST, SMALL, IRREGULAR FINGER JERKS

≠ MYOCLONUS

NEUROLOGY 1991;41:1021-1024

Cramp-fasciculation syndrome:
A treatable hyperexcitable peripheral nerve disorder

A.J. Tahmoush, MD; R.J. Alonso, MD; G.P. Tahmoush; and T.D. Heiman-Patterson, MD

Brain (2002), **125,** 1887–1895

Phenotypic variants of autoimmune peripheral nerve hyperexcitability

Ian K. Hart,[1] Paul Maddison,[2] John Newsom-Davis,[2] Angela Vincent[2] and Kerry R. Mills[3]

CRAMP-FASCICULATION SYNDROME ASSOCIATED WITH MONOFOCAL MOTOR NEUROPATHY

NICOLAS J. DUBUISSON, MD,[1,2] VINCENT VAN PESCH, MD, PhD,[2] and PETER Y.K. VAN DEN BERGH, MD, PhD[1,2]

[1]Neuromuscular Reference Center, Cliniques universitaires Saint-Luc Avenue Hippocrate 10/13.11 1200 Brussels, Belgium

MUSCLE & NERVE October 2017

MYOKYMIC DISCHARGES

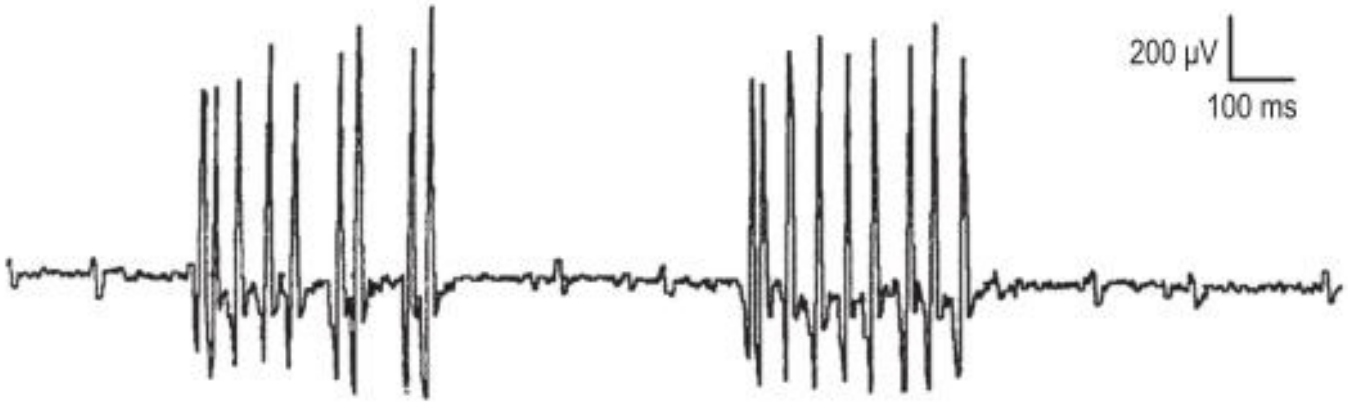
- Radiation injury (usually brachial plexopathy)
- Guillain-Barré syndrome (facial)
- Multiple sclerosis (facial)
- Pontine tumors (facial)
- Hypocalcemia
- Timber rattlesnake envenomization

- Occasionally: GBS (limbs), CIDP, nerve entrapments, radiculopathy

MOTOR UNIT POTENTIALS

MYOKYMIA

(DEMYELINATION → DEPOLARIZATION OR EPHAPTIC)

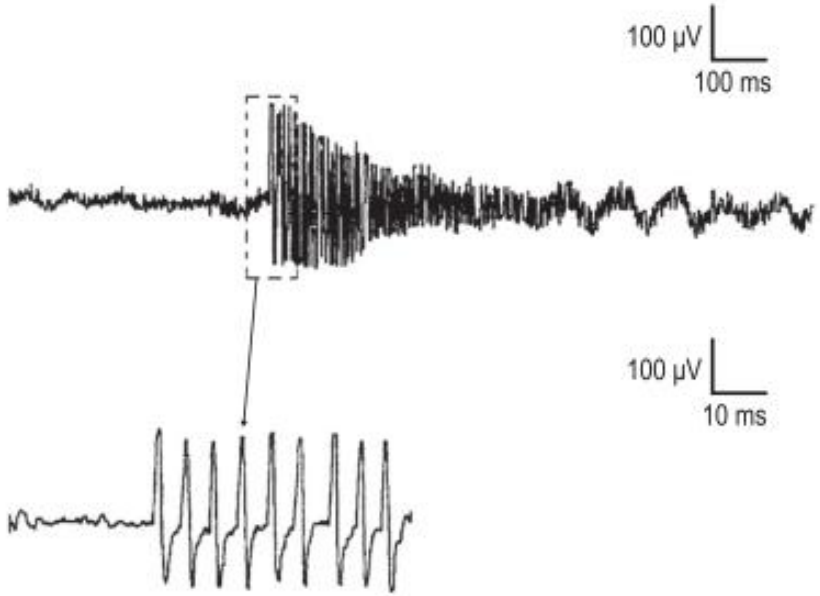


RIPPLING, QUIVERING

≠ TREMOR

MOTOR UNIT POTENTIALS

NEUROMYOTONIA

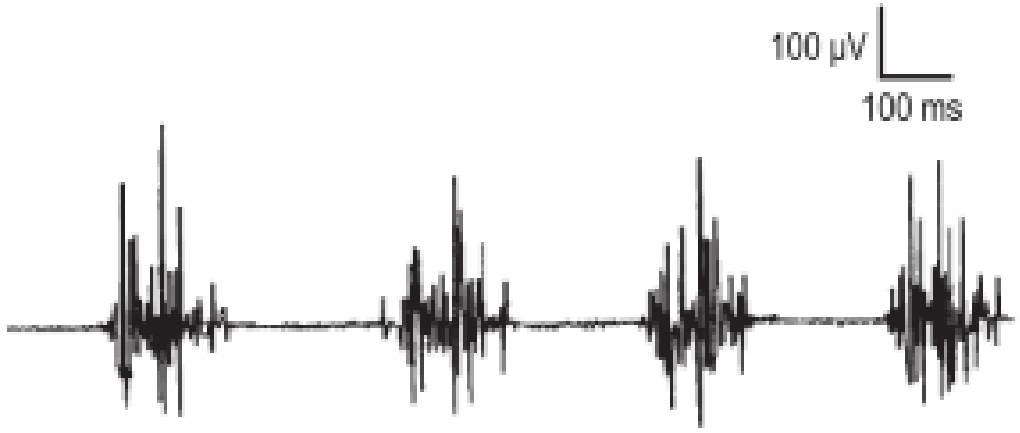


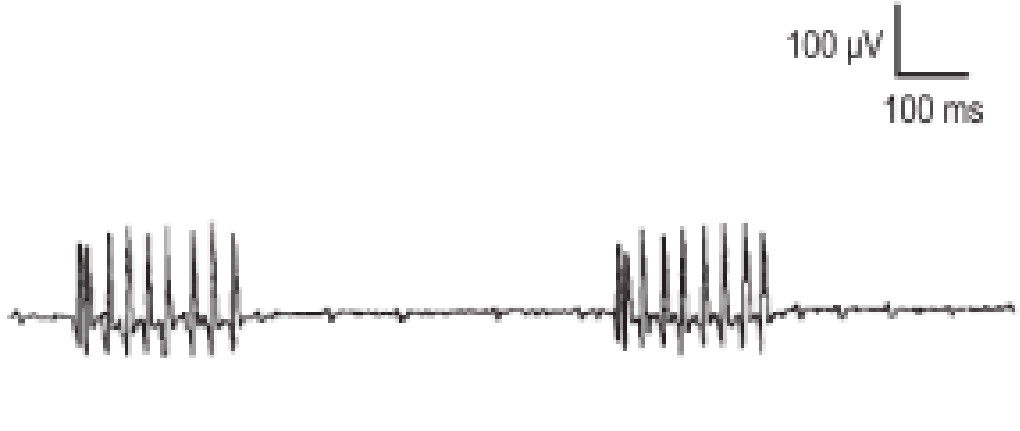
RIPPLING, QUIVERING, POSTURES

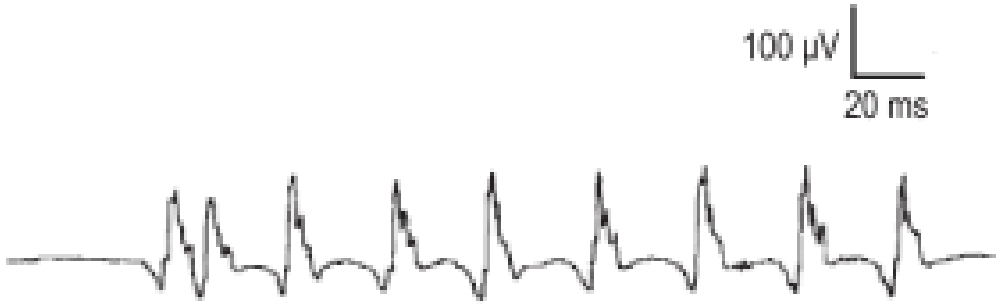
≠ TREMOR, MYOCLONUS

TREMOR

MYOKYMIA

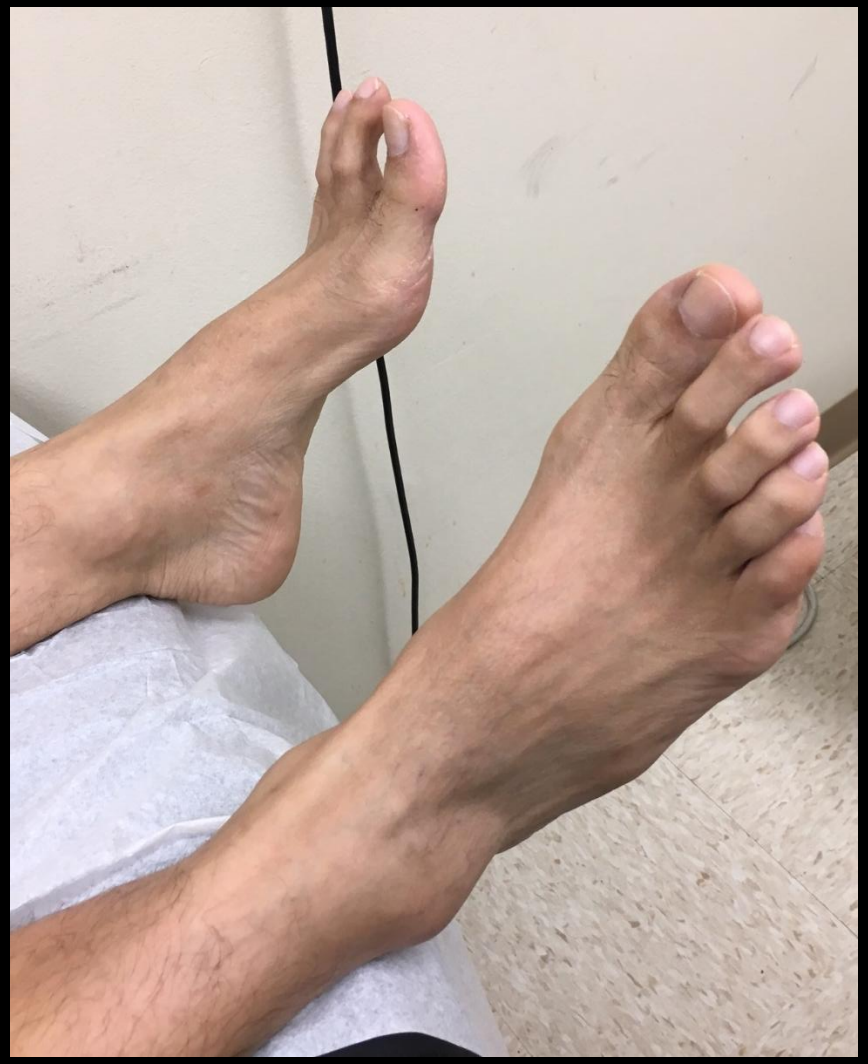




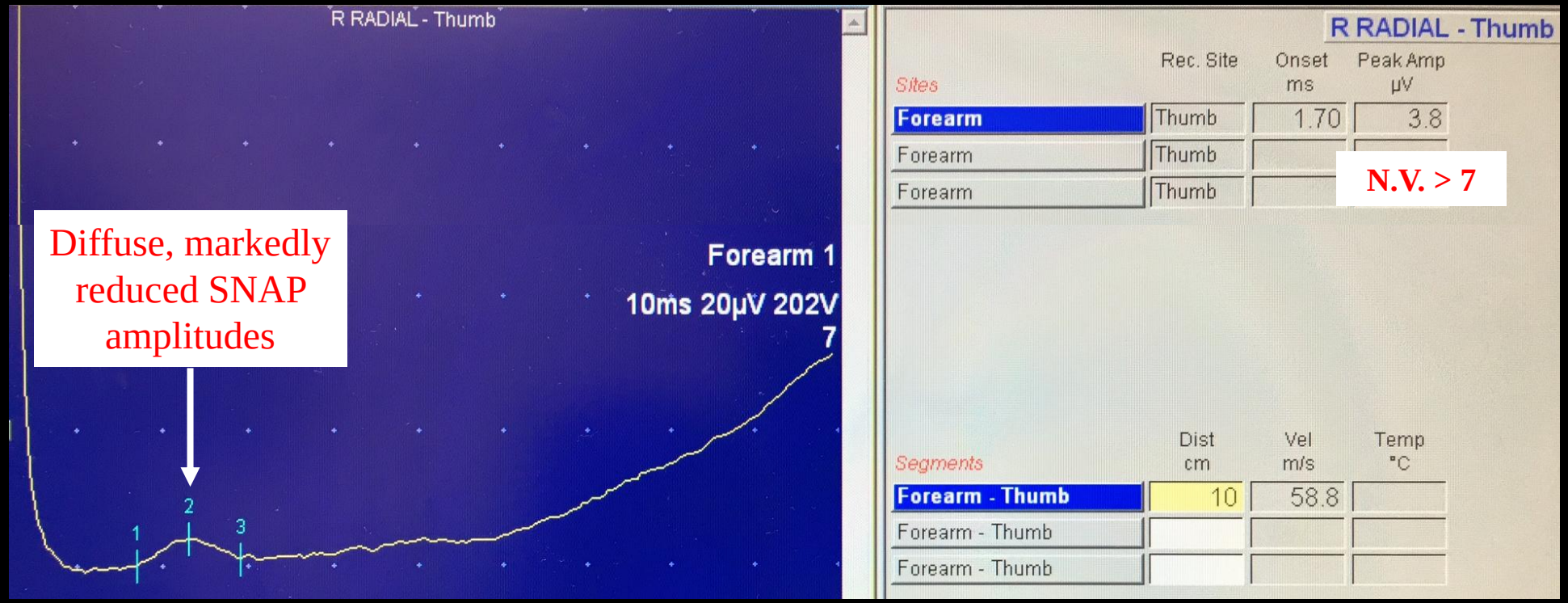


REST OF EXAMINATION

- Mild weakness in proximal upper limbs
- Mild-to-moderate weakness in proximal and distal lower limbs
- Diffuse arreflexia
- Distal sensory loss to all modalities



NERVE CONDUCTION STUDIES



NERVE CONDUCTION STUDIES

Sensory NCS

Nerve / Sites	Rec. Site	Onset ms	Peak Amp µV	Dist cm	Vet m/s	Temp °C
R MEDIAN - Dig II						
Wrist	II	2.75	1.9	14	50.9	32.7
R ULNAR - Dig V						
Wrist	Dig V	2.55	2.1	14	54.9	32.3
R RADIAL - Thumb						
Forearm	Thumb	1.70	3.8	10	58.8	
R SURAL - Lat Mall						
Calf	Lat Mall	3.80	1.5	14	36.8	
L SURAL - Lat Mall						
Calf	Lat Mall	NR	NR	14	NR	32.3
L SUP PERONEAL						
Lat Leg	Ankle	NR	NR			

Diffuse, markedly reduced SNAP amplitudes

NERVE CONDUCTION STUDIES

Motor NCS

Nerve / Sites	Rec. Site	Lat ms	Amp mV	Area mVms	Rel Area %	Distance cm	Vel m/s	Temp. °C	Comment
R MEDIAN - APB									
Wrist	APB	3.85	8.0	20.3	100	7		32	
Elbow	APB	8.60	8.7	24.4	120	25	52.6	32.1	
L PERONEAL - EDB									
Ankle	EDB	5.70	2.2	7.1	100	9		32.5	
FibHead	EDB	6.15	2.4	7.1	101			32.5	

Borderline low amplitudes of CMAP responses in lower extremities.

Diffuse sensory >> motor axonal neuropathy

ELECTROMYOGRAPHY

EMG Summary Table									
	Spontaneous					MUAP			Recruitment
	IA	Fib	PSW	Fasc	Other	Amp	Dur.	Polyphasia	Pattern
L. TIB ANTERIOR	N	None	None	None	None	1+	1+	1+	Sl Reduced
L. GASTROCN (MED)	Incr	None	1+	None	None	2+	2+	1+	Mod. Reduced
L. VAST LATERALIS	Incr	None	1+	None	None	2+	2+	2+	Mod. Reduced
R. DELTOID	N	None	None	Occasional	None	3+	2+	1+	Sev. Reduced
R. TRICEPS	N	None	None	None	None	2+	2+	2+	Mod. Reduced
R. ABD DIG MIN (UL)	N	None	None	Occasional	None	4+	2+	2+	Sev. Reduced
L. MENTALIS	N	None	None	None	None	2+	2+	1+	Mod. Reduced
L. TONGUE	N	None	None	None	None	N	N	1+	Sl Reduced
R. THOR PSP (U)	N	None	None	None	None	N	N	N	N

Very chronic neurogenic changes in all regions including bulbar, with little ongoing denervation.

PCR amplification and high resolution electrophoresis: **44 CAG** repeats in one of the alleles of the **androgen receptor gene** (normal 11–33)

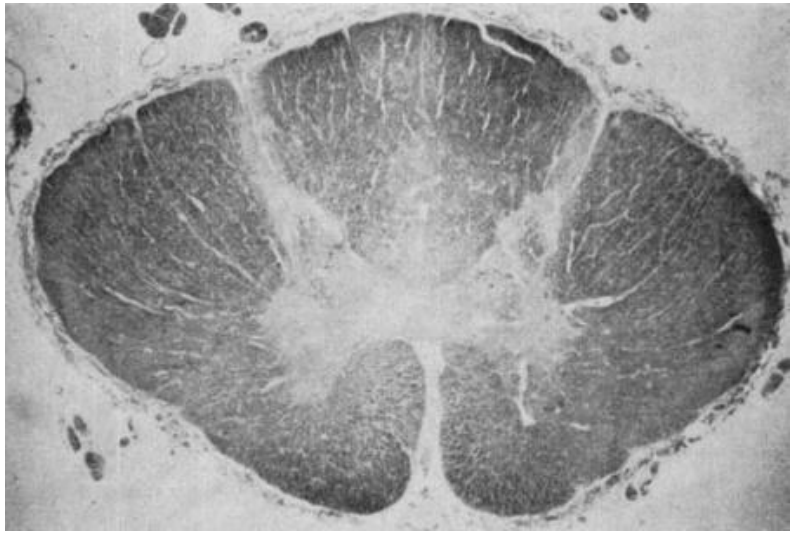
Spinal and bulbar muscular atrophy (Kennedy's disease)

Progressive proximal spinal and bulbar muscular atrophy of late onset

A sex-linked recessive trait

William R. Kennedy, M.D., Milton Alter, M.D., and Joo Ho Sung, M.D.

Neurology / Volume 18 / July 1968 671



- "Other findings included a fine **tremor** of the hands when the arms were outstretched"
- "This man first noted a fine **tremor** of the hands at age 29"
- "A fine rapid **tremor** of the hands developed"
- "**Essential tremor** was present in some affected and unaffected members"

No Videos

Clinical features of spinal and bulbar muscular atrophy

Lindsay E. Rhodes,[1] Brandi K. Freeman,[1] Sungyoung Auh,[2] Angela D. Kokkinis,[1] Alison La Pean,[1] Cheunju Chen,[1] Tanya J. Lehky,[3] Joseph A. Shrader,[4] Ellen W. Levy,[4] Michael Harris-Love,[4,5] Nicholas A. Di Prospero[1] and Kenneth H. Fischbeck[1]

	Number (%)
Presenting symptoms	
Bulbar weakness	1 (1)
Arm weakness	5 (7)
Leg weakness	16 (23)
Breast enlargement	5 (7)
Cramps	22 (32)
Tremor	16 (23)
Other	4 (6)
Area of first muscle weakness	
Bulbar	20 (33)
Arm	10 (17)
Leg	30 (50)

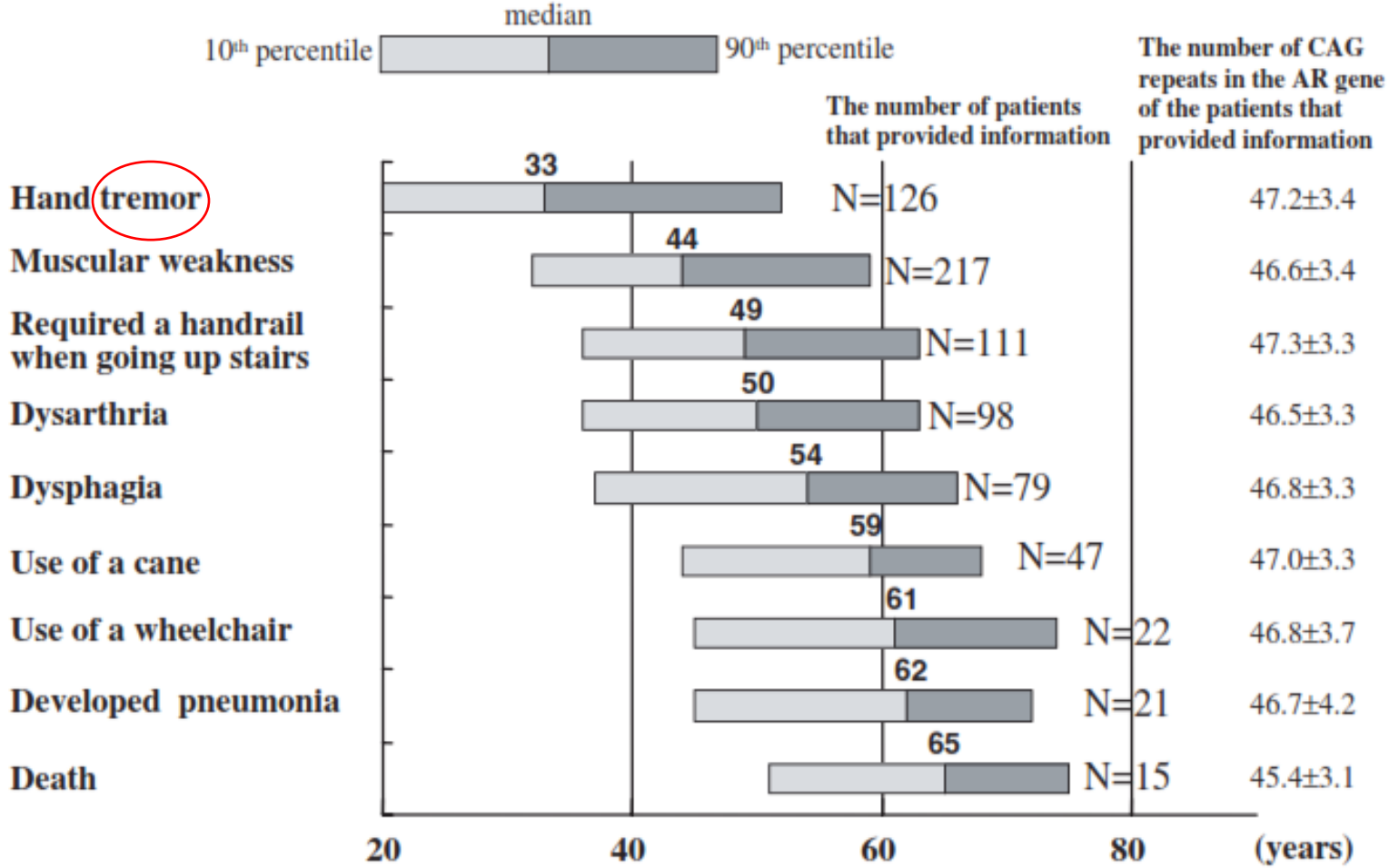
Presenting symptoms were assessed retrospectively for 57 patients at the time of evaluation. Some patients reported more than one symptom at onset. Numbers in parentheses indicate percent of total symptoms reported. Presenting symptoms noted as "other" include choking, muscle twitching (fasciculations) and musculoskeletal pain.

No Videos

doi:10.1093/brain/awl096 Brain (2006), 129, 1446–1455

Natural history of spinal and bulbar muscular atrophy (SBMA): a study of 223 Japanese patients

Naoki Atsuta,[1] Hirohisa Watanabe,[1] Mizuki Ito,[1] Haruhiko Banno,[1] Keisuke Suzuki,[1] Masahisa Katsuno,[1] Fumiaki Tanaka,[1] Akiko Tamakoshi[2] and Gen Sobue[1]



No Videos

doi:10.1093/brain/awl096 Brain (2006), 129, 1446–1455

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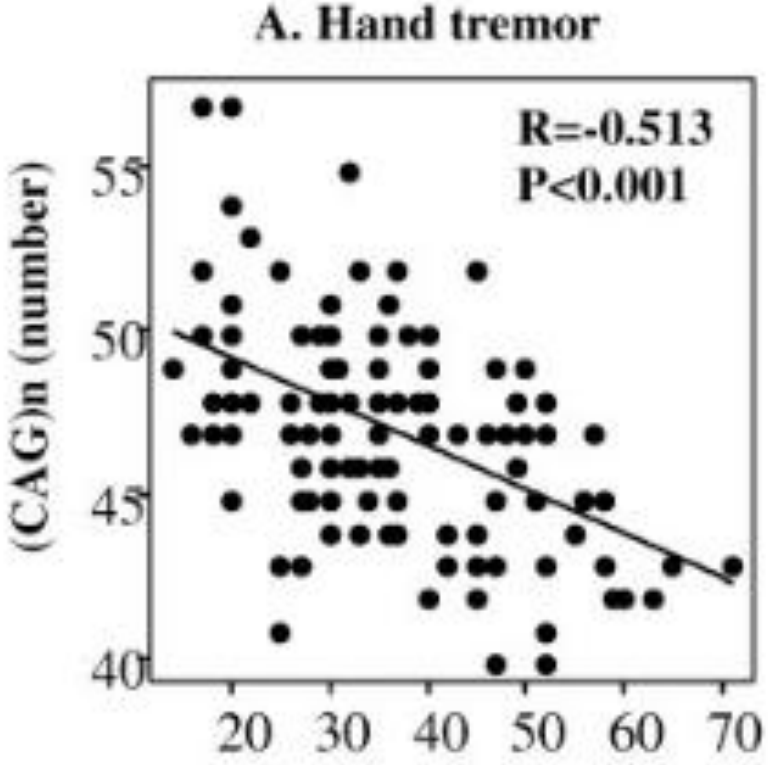
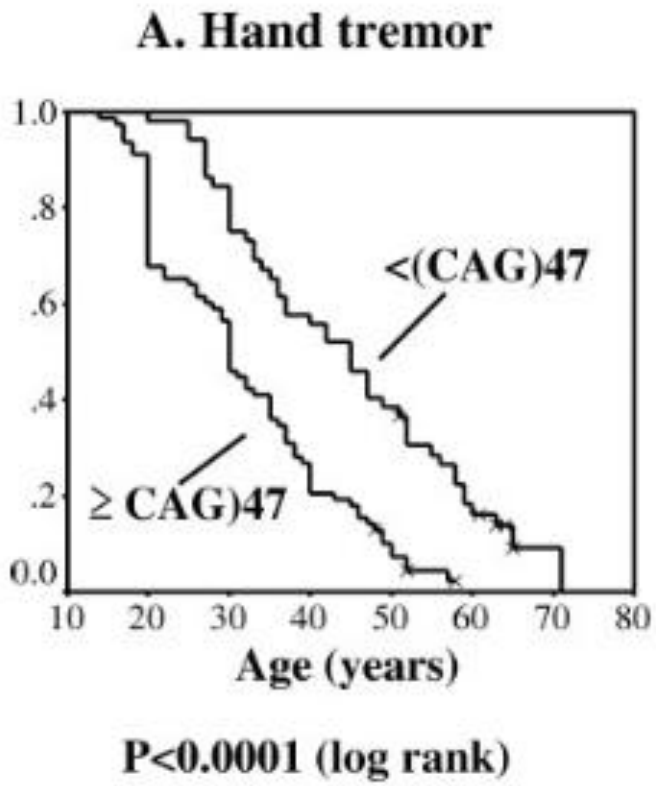
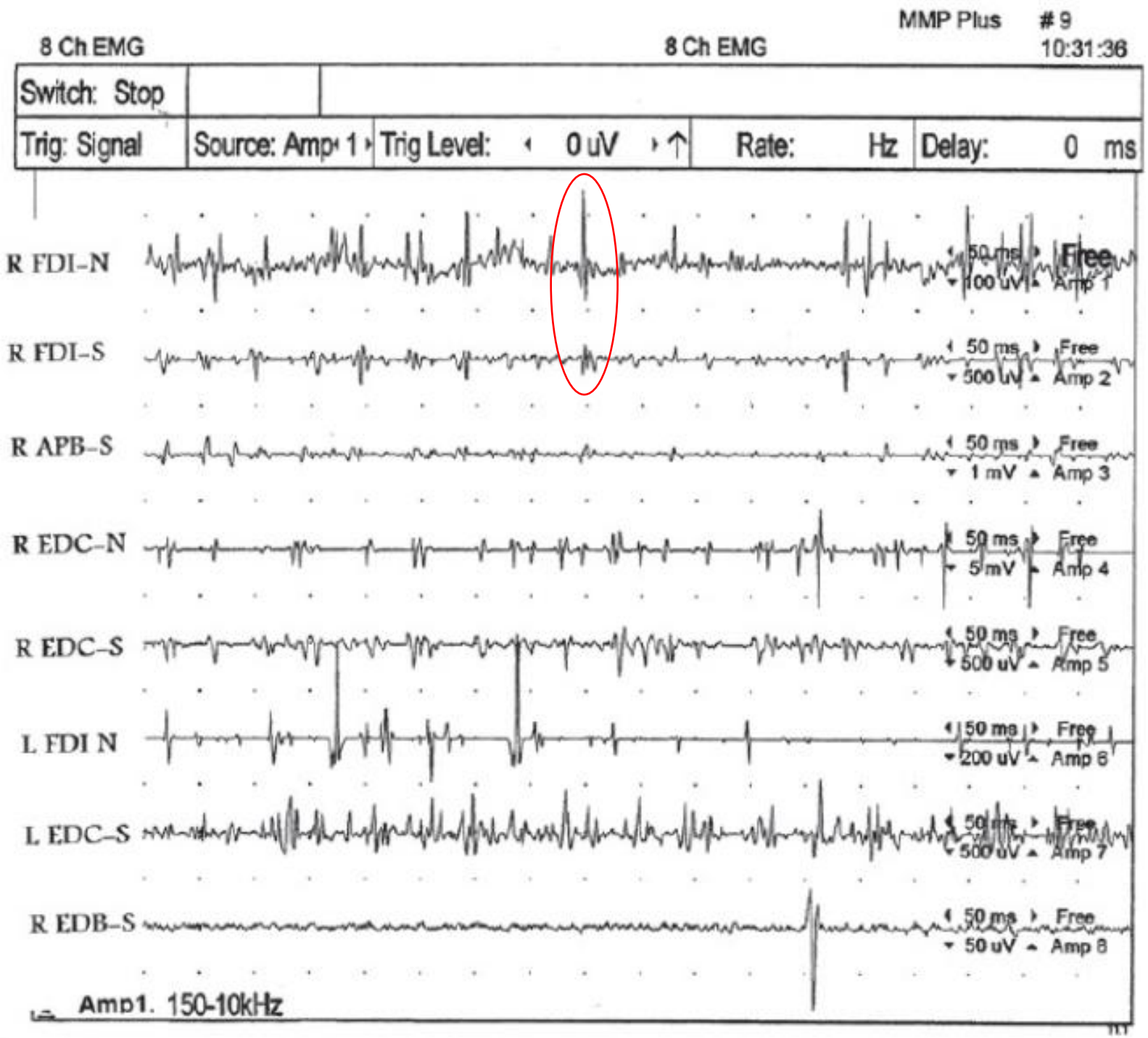
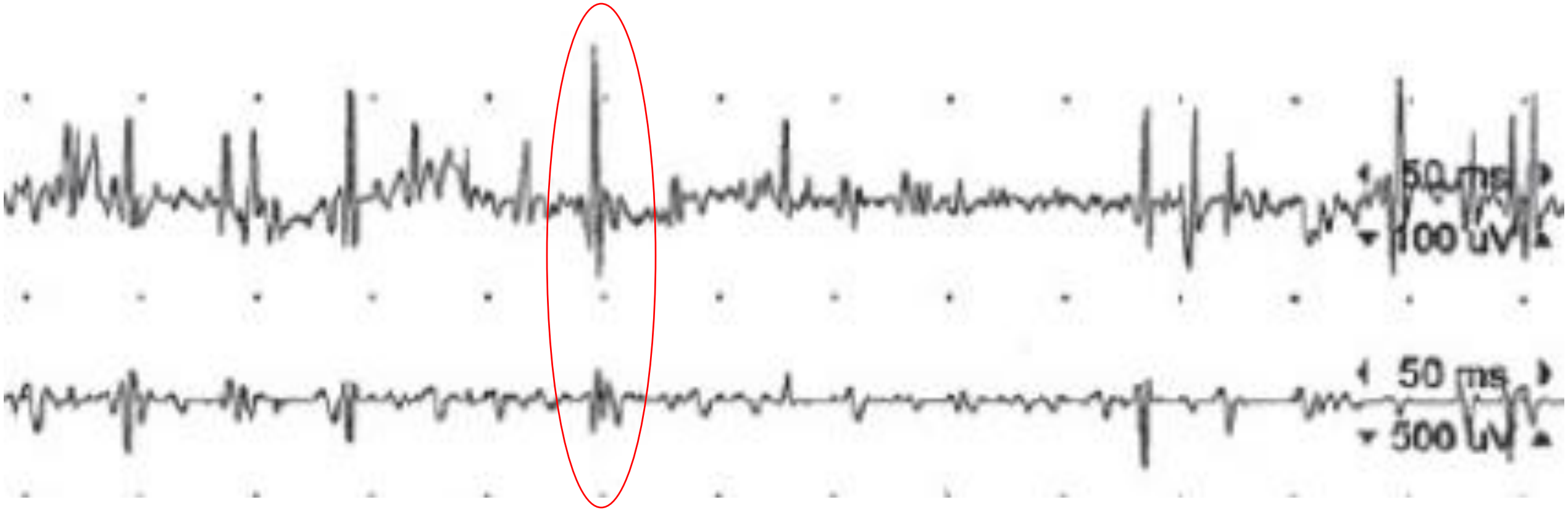




No Videos


MMP Plus # 9
8 Ch EMG
8 Ch EMG
10:31:36
Switch: Stop
Trig: Signal
Source: Amp 1
Trig Level: 0 uV
Rate: Hz
Delay: 0 ms
R FDI-N
R FDI-S
R APB-S
R EDC-N
R EDC-S
L FDI N
L EDC-S
R EDB-S
50 ms Free
100 uV Amp 1
50 ms Free
500 uV Amp 2
50 ms Free
1 mV Amp 3
50 ms Free
5 mV Amp 4
50 ms Free
500 uV Amp 5
50 ms Free
200 uV Amp 6
50 ms Free
500 uV Amp 7
50 ms Free
50 uV Amp 8
Amp1. 150-10kHz


50 ms
100 uV
50 ms
500 uV

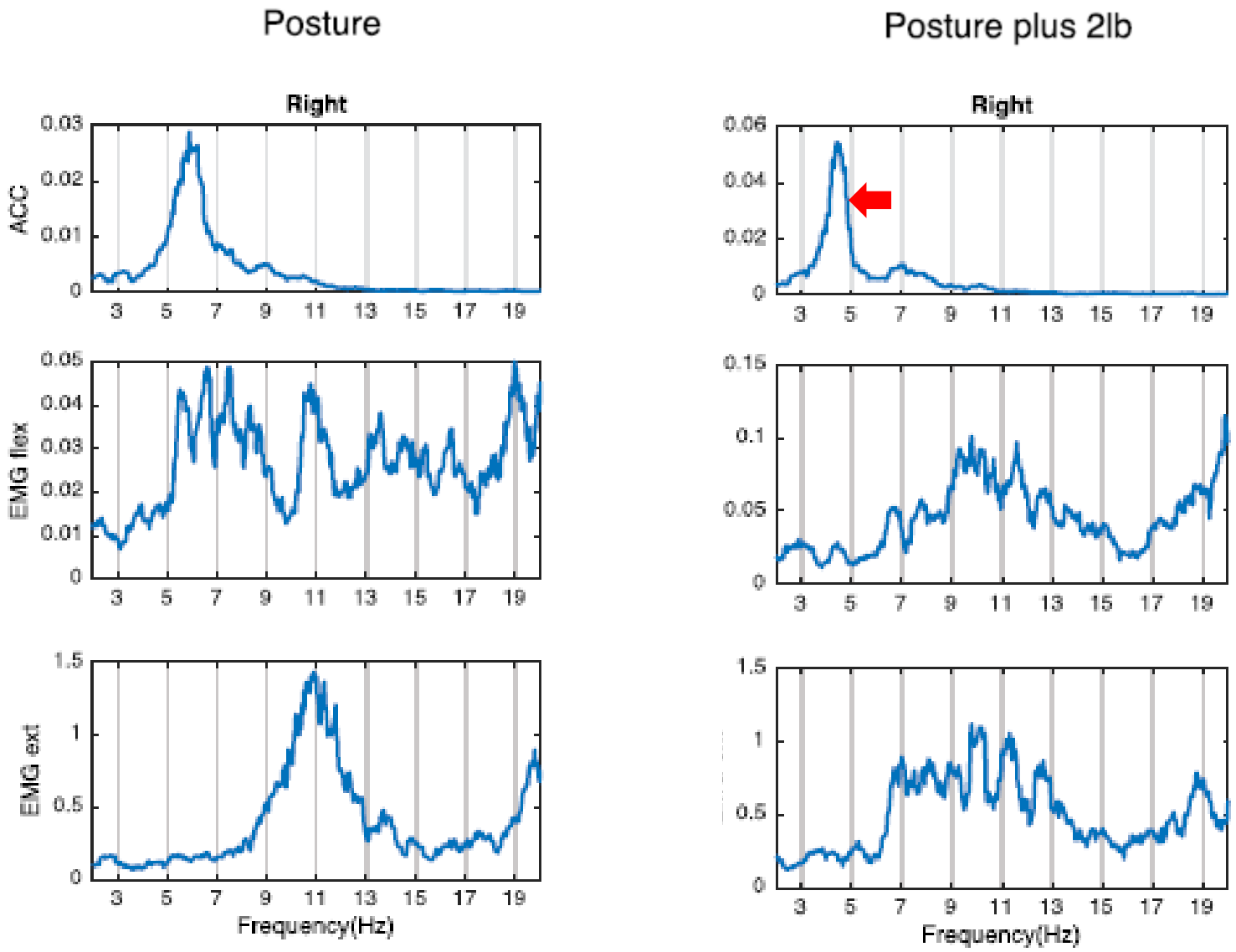
Contraction Pseudotremor of Chronic Denervation

Jack E. Riggs, MD; Ludwig Gutmann, MD; Sydney S. Schochet, Jr, MD

• The tremulousness observed with voluntary muscle contraction in patients with chronic denervating illness has long been described but has been given misleading and confusing labels. The phenomenon is generally attributed to the nonsmooth contraction of a muscle populated by motor units decreased in number and enlarged. *Contraction pseudotremor of chronic denervation* may be a more appropriate label for this useful clinical sign.

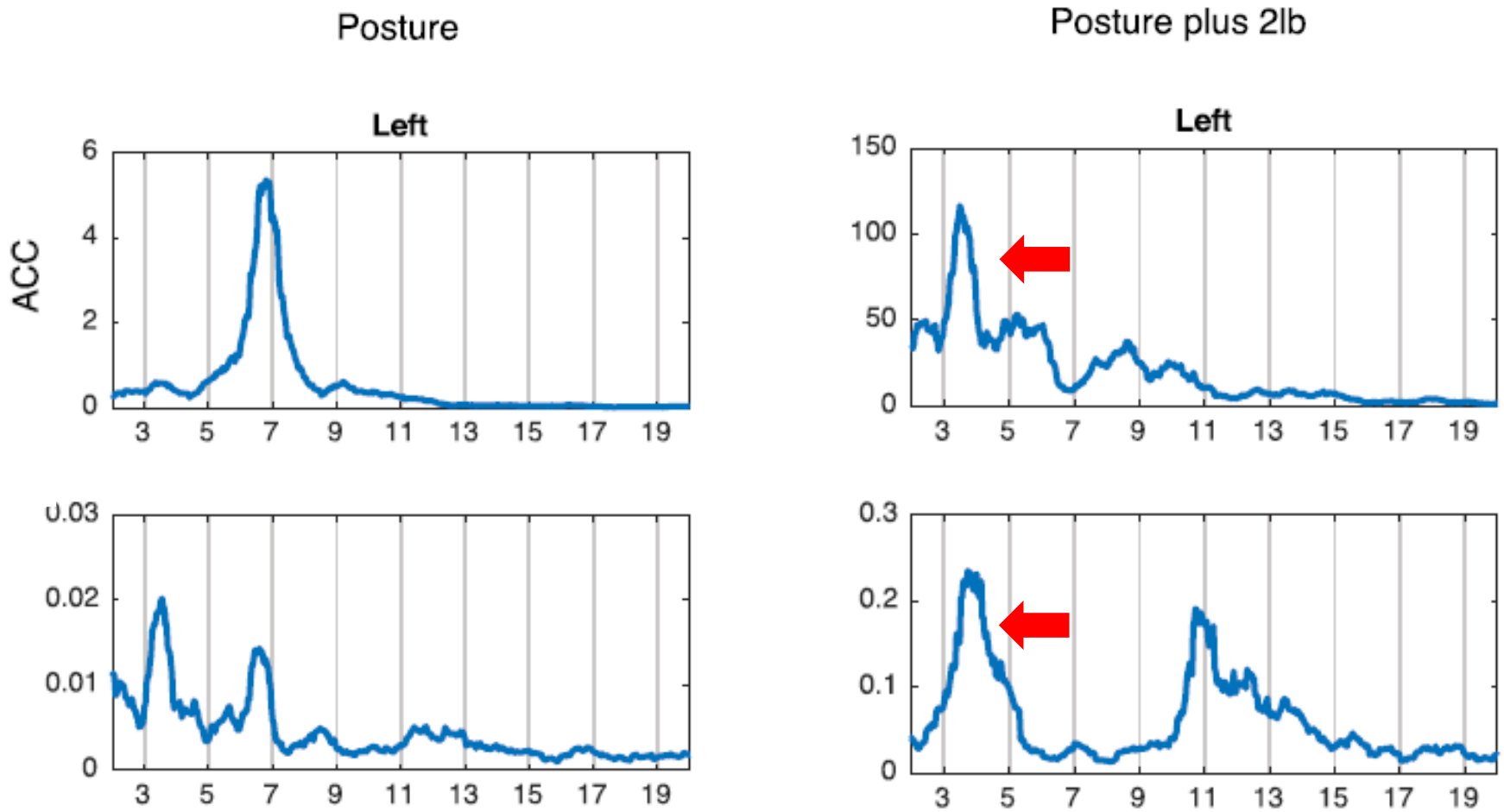
(*Arch Neurol* 1983;40:518-519)

Mechanical tremor



F. Vial et al. / Clinical Neurophysiology Practice 4 (2019) 134–142

Reflex tremor



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Postural Tremor in X-Linked Spinal and Bulbar Muscular Atrophy

Ritsuko Hanajima, MD, PhD,[1*] Yasuo Terao, MD, PhD,[1] Setsu Nakatani-Enomoto, MD,[1,2] Masashi Hamada, MD, PhD,[1] Akihiro Yugeta, MD, PhD,[1] Hideyuki Matsumoto, MD,[1] Tomotaka Yamamoto, MD, PhD,[1] Shoji Tsuji, MD, PhD,[1] and Yoshikazu Ugawa, MD, PhD[2]

TABLE 1. *Clinical features of all patients*

Case	Age (yr)	Onset (yr)	CAG expansion	First symptom	Tremor onset (yr)	MRC scale of EDC	SCV (m/s) Median *N*; Ulnar *N*	SNAP (microV) Median *N*; Ulnar *N*
1	38	28	53	Tremor	28	5–	53; 52	**4.9; 3.1**
2	60	40	>40	Tremor	40	5–	57; 51	**2.5; 1.1**
3	42	20	45	Tremor	20	5–		
4	61	25	50	Tremor	25	5–	52; 51	16.3; 6.4
5	63	43	47	Muscle weakness	51	5–	59; 51	12.5; **1.9**
6	58	45	48	Muscle weakness	45	5–	52; **44**	12.5; **1.9**
7	49	20	46	Tremor	20	5–	52; 47	**6.3; 3.4**
8	47	25	43	Tremor	25	4	56; 59	**7.9; 4.6**

Normal range: SCV, 49–66 m/s and 47.0–69.0 m/s for the median and ulnar nerves; SNAP amplitude, 9.4–40.0 μV and 5.0–36.0 μV for the median and ulnar nerves.

SCV, sensory conduction velocity; SNAP, sensory nerve action potential amplitude, MRC scale, Medical Research Council grade for muscle power.

"All patients had postural **tremor** in the forearm and finger muscles at **6–9 Hz (~ET).** No patients had sensory symptoms."

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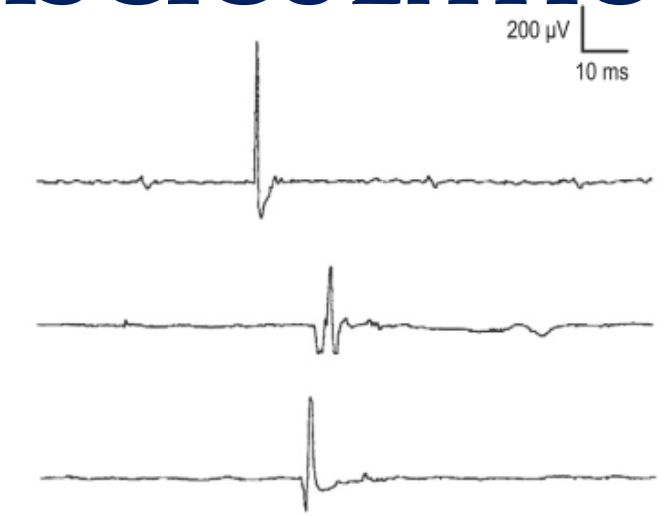
- Phenomenology ≈ ET
- Physiology **≠ ET → Reflex ± mechanical tremor**
  - ET: grouped EMG activity at the same frequency
  - ET: resistant to perturbations (weight, postural changes)

MOTOR UNIT POTENTIALS

FASCICULATIONS



FAST FINGER / FACE JERKS
"MINIPOLYMYOCLONUS"

≠ MYOCLONUS
≠ TREMOR

Thank you