A/Prof Lyn Kiers

Department of Neurology and Clinical Neurophysiology

Royal Melbourne Hospital

Mr. SN 43 yo man

- Presented to ED with 3 M history of left hand paresthesia, weakness and wasting
- Awoke with paresthesia L D4/5
- Progressive numbness L D1-3
- Burning pain left hand, worse at night
- Occasional shooting pain from proximal arm to hand
- Weakness left hand, difficulty grasping objects

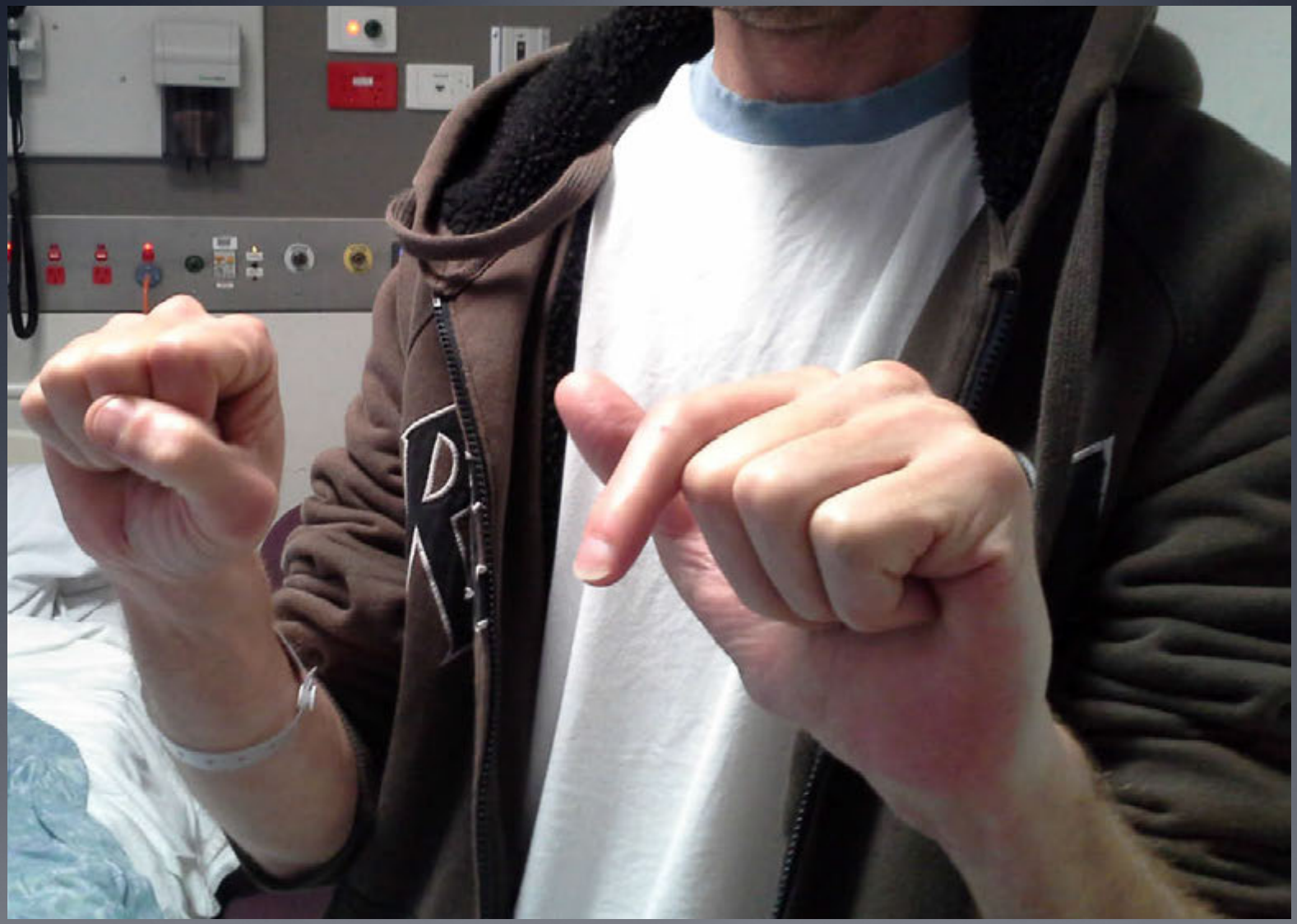
HOPC

- Associated symptoms:
  - 15kg LOW
  - Occasional night sweats
  - Fatigue/lethargy
- Self employed, property maintenance
- Smoker 20 PY; Alcohol 4 SD/W

Referred to Neurology rapid access clinic from ED

Examination

- General examination normal
- Neuro exam normal except LUL
- Tone, reflexes normal
- Weakness median innervated muscles below and including PT
- Reduced LT/pp sensation in entire median N distribution (D1-3, lateral D4, thenar eminence)



? Differential diagnoses

? Further investigation

PMH

- Referred to neurologist in Dec 2008 for investigation of left thumb numbness and dysesthesia
- Symptoms developed after fall with traumatic pneumothorax/GA/thoracic surgery Feb 2008
- Reported numbness left dorsal thumb/index finger/distal forearm; medial aspect right forearm
- Examination normal except reduced pp in left radial and right MABC nerve territories

PMH

- NCS – absent left radial SNAP, MABC not tested
- MRI cervical spine/brachial plexus – normal
- Diagnosis – left radial sensory neuropathy
- Symptoms mild and stable until current presentation 5 yrs later.

? Differential diagnoses

? Further investigation

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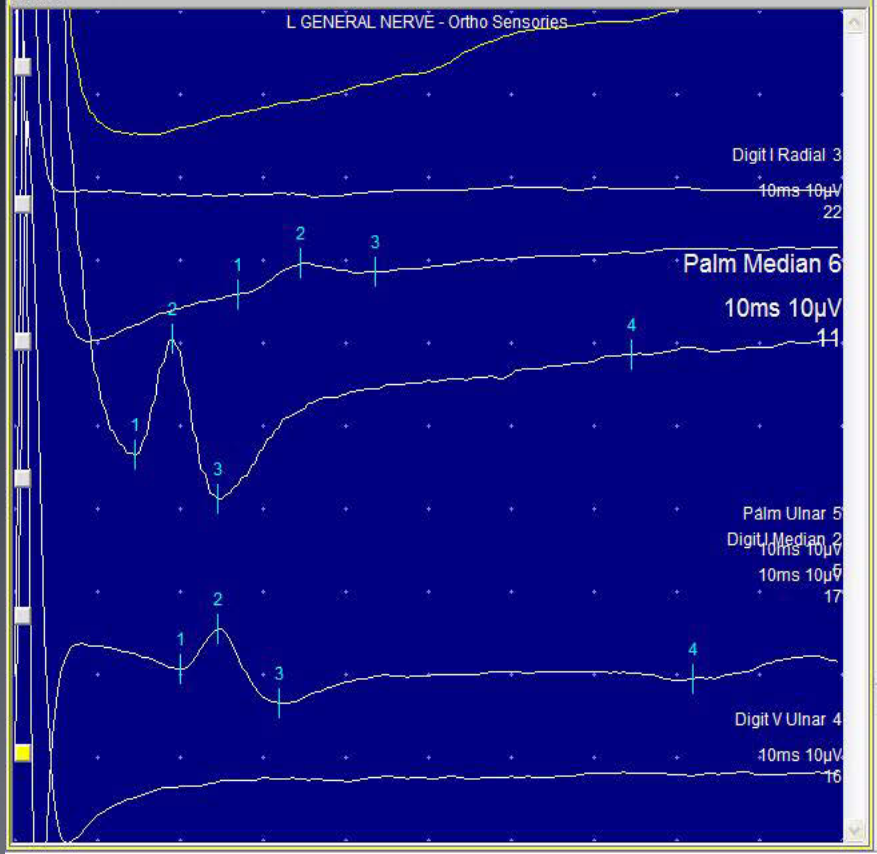
Stores



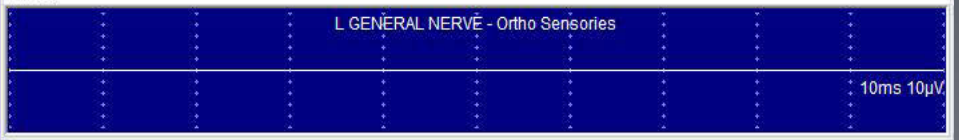
Results Table

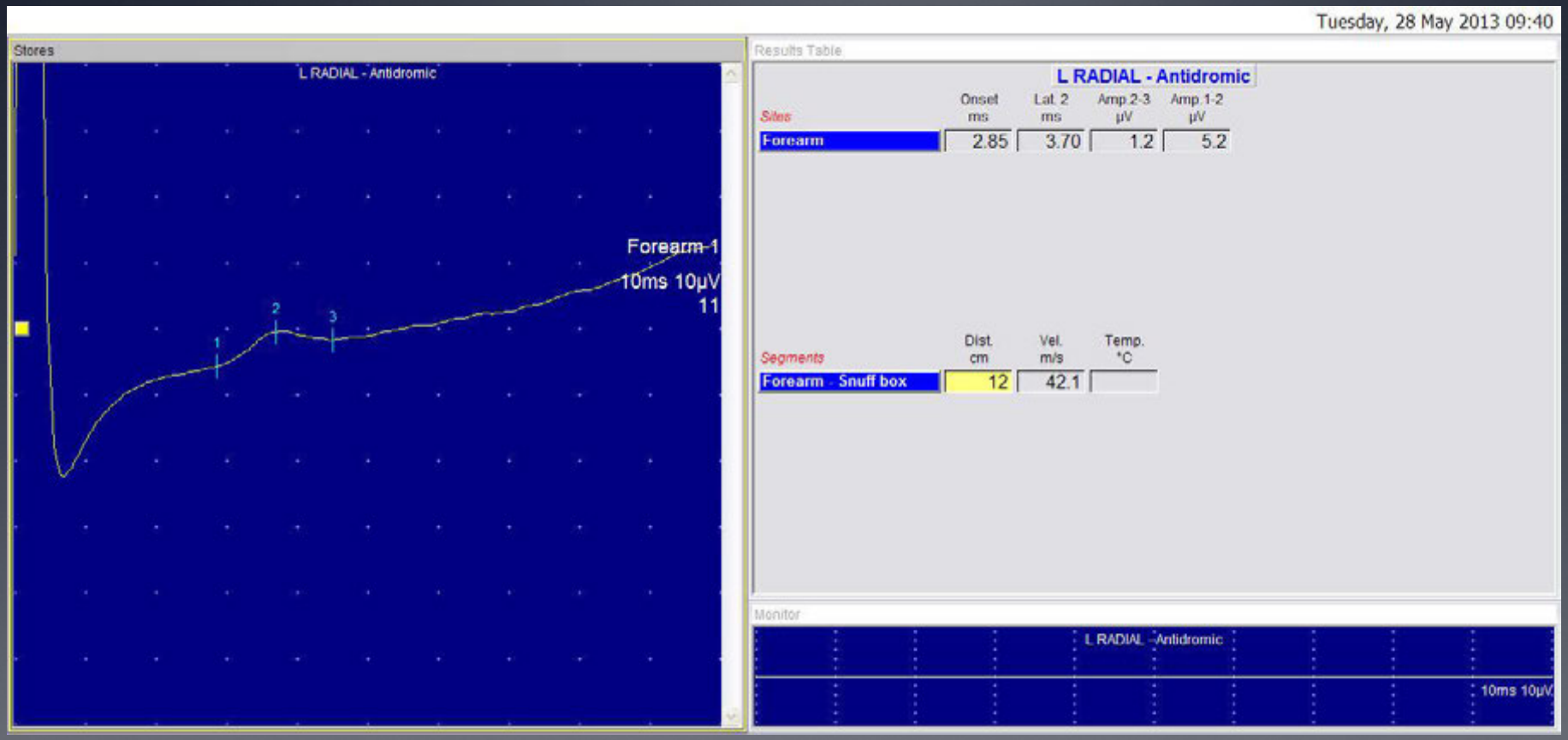
L GENERAL NERVE - Ortho Sensories

Sites	Onset ms	Lat. 2 ms	Amp.2-3 µV	Amp.1-2 µV
Digit II Median				
Digit I Median				
Digit I Radial	2.70	3.45	1.0	3.8
Digit V Ulnar	2.00	2.45	9.2	4.8
Palm Ulnar	1.45	1.90	19.4	14.0
Palm Median				
Digit IV Median				

Segments	Dist. cm	Vel. m/s	Temp. °C
Digit II Median - Wrist			
Digit I Median - Wrist			
Digit I Radial - Wrist	9.5	35.2	
Digit V Ulnar - Wrist	11.3	56.5	
Palm Ulnar - Wrist	8	55.2	
Palm Median - Wrist	8		
Digit IV Median - Wrist			

Monitor

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Stores
L RADIAL - Antidromic
Forearm-1
10ms 10µV
11
1
2
3
Results Table
L RADIAL - Antidromic
Sites
Onset ms
Lat 2 ms
Amp 2-3 µV
Amp 1-2 µV
Forearm
2.85
3.70
1.2
5.2
Segments
Dist. cm
Vel. m/s
Temp. °C
Forearm - Snuff box
12
42.1
Monitor
L RADIAL - Antidromic
10ms 10µV

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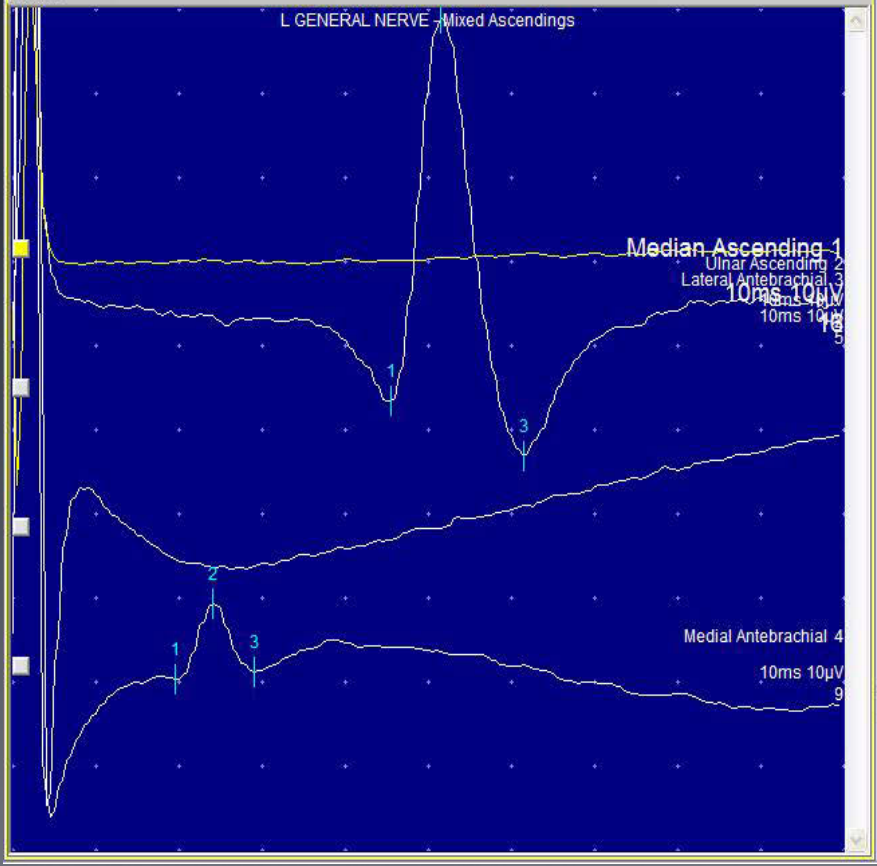
Stores



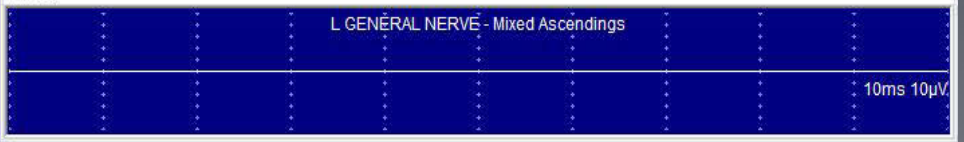
Results Table

L GENERAL NERVE - Mixed Ascendings

Sites	Onset ms	Lat. 2 ms	Amp.2-3 µV	Amp.1-2 µV
Median Ascending				
Ulnar Ascending	4.55	5.15	52.2	45.7
Lateral Antebrachial				
Medial Antebrachial	1.95	2.40	8.2	9.1

Segments	Dist. cm	Vel. m/s	Temp. °C
Median Ascendi - Elbow			
Ulnar Ascending - Elbow	25.5	56.0	
Lateral Antebrac - Forearm			
Medial Antebrac - Forearm	12	61.5	

Monitor



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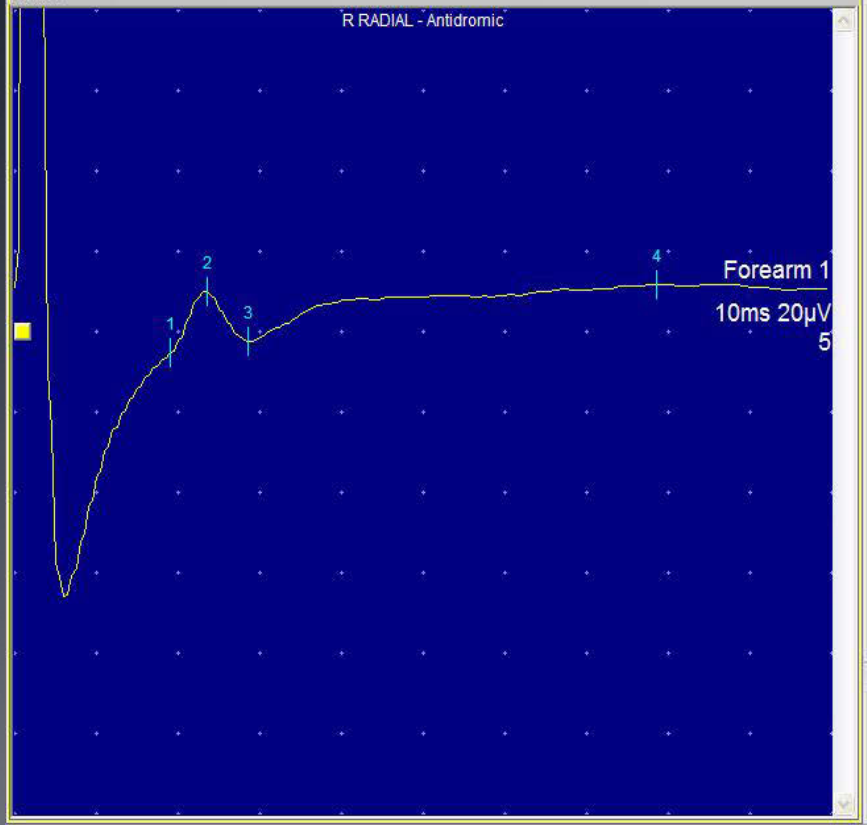

Results Table

R RADIAL - Antidromic

Sites	Onset ms	Lat. 2 ms	Amp.2-3 µV	Amp.1-2 µV
Forearm	1.90	2.35	12.4	15.3

Segments	Dist. cm	Vel. m/s	Temp. °C
Forearm - Snuff box	10.5	55.3	

Monitor

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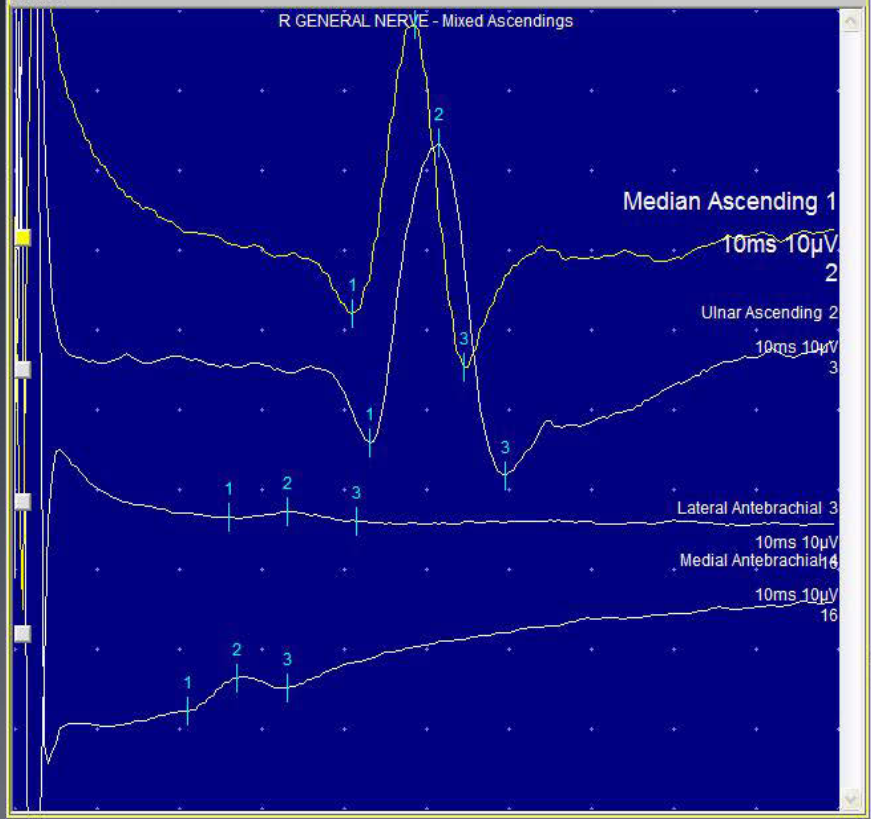
Stores



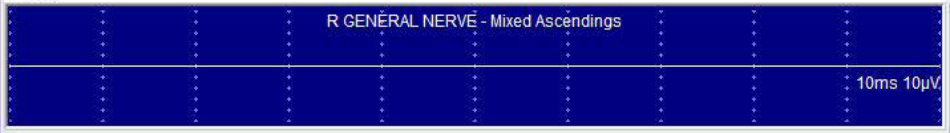
Results Table

R GENERAL NERVE - Mixed Ascendings

Sites	Onset ms	Lat. 2 ms	Amp.2-3 µV	Amp.1-2 µV
Median Ascending	4.10	4.85	43.0	36.3
Ulnar Ascending	4.30	5.15	41.9	37.7
Lateral Antebrachial	2.60	3.30	1.2	0.67
Medial Antebrachial	2.10	2.70	1.2	4.1

Segments	Dist. cm	Vel. m/s	Temp. °C
Median Ascendi - Elbow	24.5	59.8	
Ulnar Ascending - Elbow	25	58.1	
Lateral Antebrac - Forearm	9.5	36.5	
Medial Antebrac - Forearm	10	47.6	

Monitor



Motor NCS

Nerve / Sites	Rec. Site	Lat. ms	Amp.1-2 mV	Dist cm	Vel m/s
L MEDIAN - APB					
Wrist	APB	Absent			
R MEDIAN - APB					
Wrist	APB	3.30	6.0		
Elbow		8.10	5.5	26.5	55.2
L MEDIAN - II Lumb					
MEDIAN	II Lumb	Absent			
ULNAR	4th DI	2.45	5.5		
R ULNAR - ADM 2 point					
Wrist	ADM	2.20	10.0		
L ULNAR - ADM 3 point					
Wrist	ADM	2.25	10.4		
Below Elbow		6.35	9.5	22	53.7
Above Elbow		8.70	8.9	12	51.1
L ULNAR - FDI					
Wrist	FDI	2.95	11.2		

F Wave

Nerve	Fmin ms
L ULNAR	28.35
R MEDIAN	28.95
R ULNAR	28.90

Sensory NCS

Nerve / Sites	Onset ms	Lat. 2 ms	Amp.2-3 µV	Amp.1-2 µV	Dist. cm	Vel. m/s
L - Ortho Sensories						
Digit II Median	Absent					
Digit I Median	Absent					
Digit I Radial	2.70	3.45	1.0	3.8	9.5	35.2
Digit V Ulnar	2.00	2.45	9.2	4.8	11.3	56.5
Palm Ulnar	1.45	1.90	19.4	14.0	8	55.2
Palm Median	Absent				8	

Nerve / Sites	Onset ms	Lat. 2 ms	Amp.2-3 µV	Amp.1-2 µV	Dist. cm	Vel. m/s
R - Ortho Sensories						
Digit II Median	2.30	2.80	10.2	7.5	12.7	55.2
Digit I Radial	1.80	2.35	6.9	4.0	9.5	52.8
Digit V Ulnar	1.95	2.45	8.3	4.6	10.8	55.4
L - Mixed Ascendings						
Median Ascending	Absent					
Ulnar Ascending	4.55	5.15	52.2	45.7	25.5	56.0
Lateral Antebrachial	Absent					
Medial Antebrachial	1.95	2.40	8.2	9.1	12	61.5
R - Mixed Ascendings						
Median Ascending	4.10	4.85	43.0	36.3	24.5	59.8
Ulnar Ascending	4.30	5.15	41.9	37.7	25	58.1
Lateral Antebrachial	2.60	3.30	1.2	0.67	9.5	36.5
Medial Antebrachial	2.10	2.70	1.2	4.1	10	47.6
L RADIAL - Antidromic						
Forearm	2.85	3.70	1.2	5.2	12	42.1
R RADIAL - Antidromic						
Forearm	1.90	2.35	12.4	15.3	10.5	55.3

EMG Summary Table									
	Spontaneous					MUAP			Recruitment
	IA	Fib	PSW	Fasc	H.F.	Amp	Dur.	PPP	Pattern
L. DELTOID	N	None	None	None	None	N	N	N	N
L. BICEPS	N	None	None	None	None	N	N	N	N
L. BRACHIORADIALIS	N	None	None	None	None	N	N	N	N
L. TRICEPS	N	None	None	None	None	N	1+	N	1-
L. EXT DIG COMM	N	None	None	None	None	N	2+	2+	1-
L. FIRST D INTEROSS	N	None	None	None	None	N	1+	1+	1-
L. FLEX POLL LONG	N	4+	3+	None	None	N	N	N	No Activity
L. PRON TERES	N	4+	3+	None	None	N	N	N	No Activity
L. FLEX CARPI RAD	N	3+	3+	None	None	N	2+	2+	2-
L. ABD POLL BREVIS	N	3+	3+	None	None	N	N	N	Discrete

Motor NCS

Nerve / Sites	Rec. Site	Lat. ms	Amp.1-2 mV	Dist cm	Vel m/s
R COMM PERONEAL - EDB					
Ankle	EDB	4.55	4.9		
FibHead		11.45	5.0	31.4	45.5
Knee		13.15	5.0	7.8	45.9
L COMM PERONEAL - EDB					
Ankle	EDB	4.45	7.9		
FibHead		11.35	7.4	32.4	47.0
Knee		13.65	7.8	9.4	40.9
R TIBIAL MALLEOLUS - AH					
Ankle	AH	3.55	10.8		
Knee		12.25	8.8	39.3	45.2
L TIBIAL MALLEOLUS - AH					
Ankle	AH	3.70	10.7		
Knee		12.75	9.4	42	46.4

F Wave

Nerve	Fmin ms
R COMM PERONEAL	51.25
R TIBIAL MALLEOLUS	50.10
L TIBIAL MALLEOLUS	53.10
L COMM PERONEAL	49.40

Sensory NCS

Nerve / Sites	Onset ms	Lat. 2 ms	Amp.2-3 µV	Amp.1-2 µV	Dist. cm	Vel. m/s
R - Leg Sensories						
Sural	2.75	3.50	13.4	16.3	12.7	46.2
Superficial Peroneal	2.30	2.85	6.7	2.8	12.1	52.6
L - Leg Sensories						
Sural	3.35	4.25	12.2	9.5	16.4	49.0
Superficial Peroneal	2.00	2.70	2.2	2.3	10.5	52.5

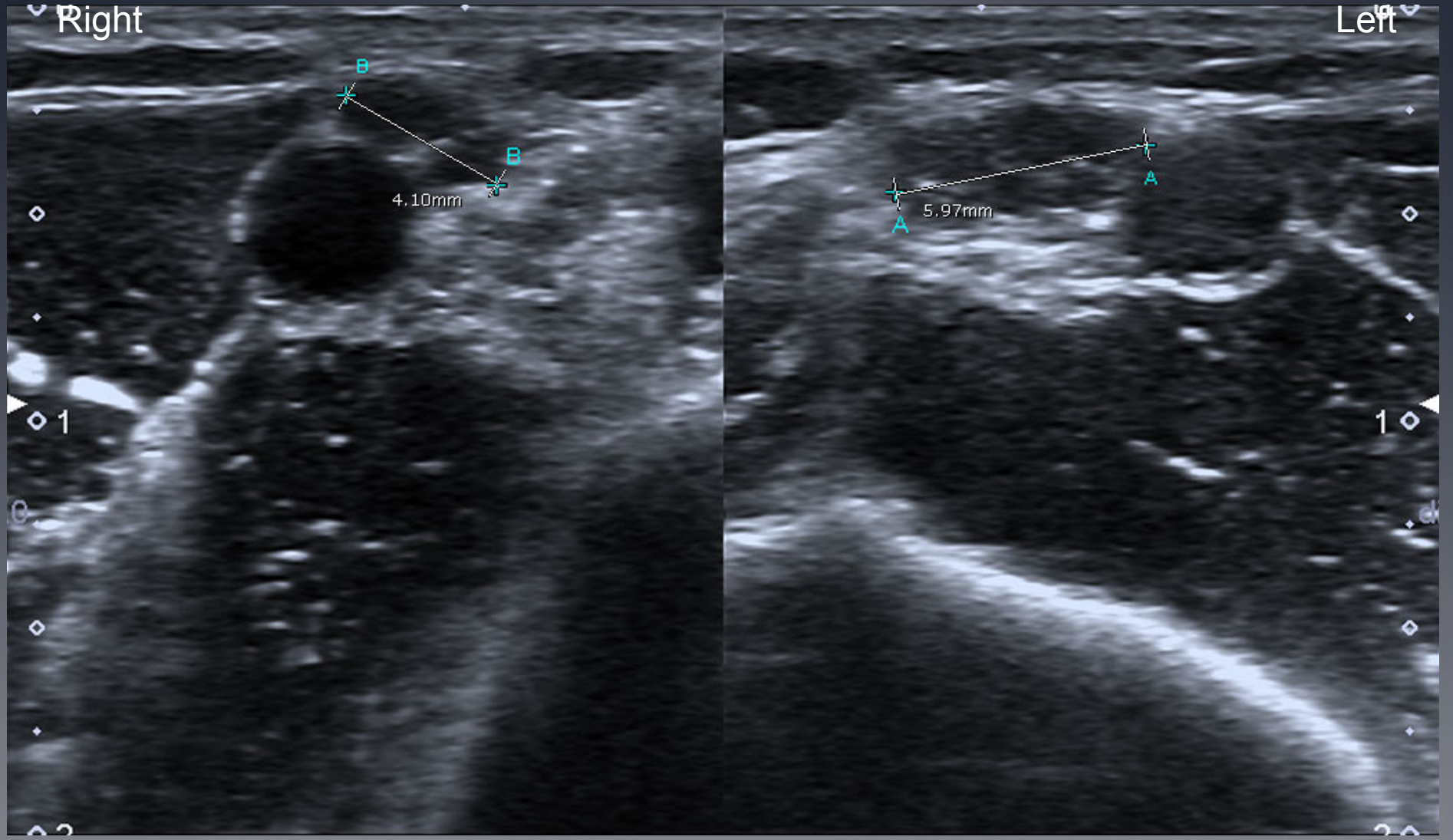
? Differential diagnoses

? Further investigation

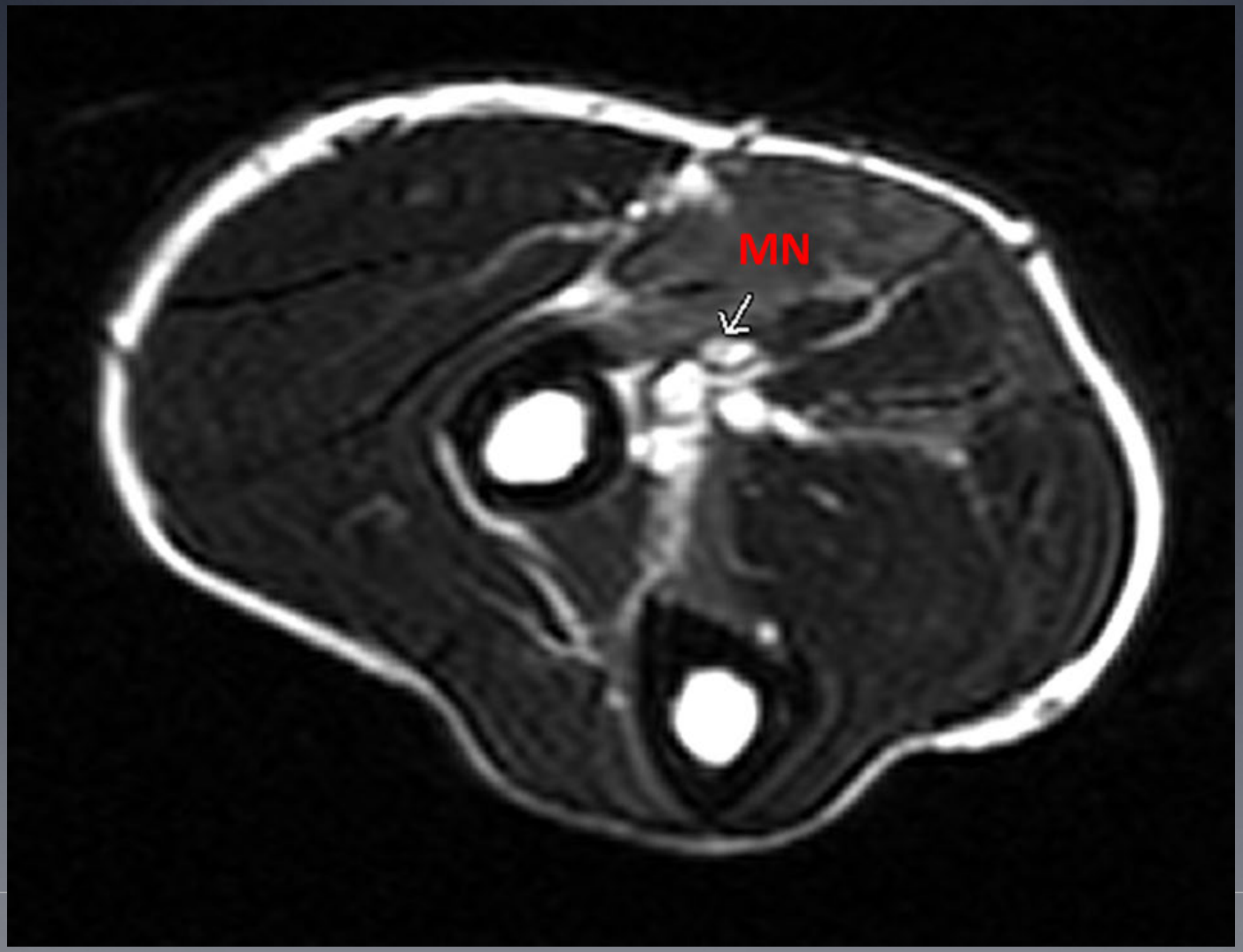
Investigations

- CRP 16 (<5), ESR 2
- C_3 0.71 (0.9-1.8), C_4 0.10 (0.16-0.50)
- Vasculitic screen (RF, ANA, ANCA, ENA anti-dsDNA, urine miscroscopy/cold agglutinins) negative
- Anti-neuronal Ab/tumor markers negative
- HIV/Hep B,C/syphilis/CMV/EBV negative
- LDH 520 (210-420), β2 microglobulin normal
- SPEP: IgMκ paraprotein 3g/L; BJ protein positive in urine (monoclonal free kappa light chains)
- Anti-MAG negative

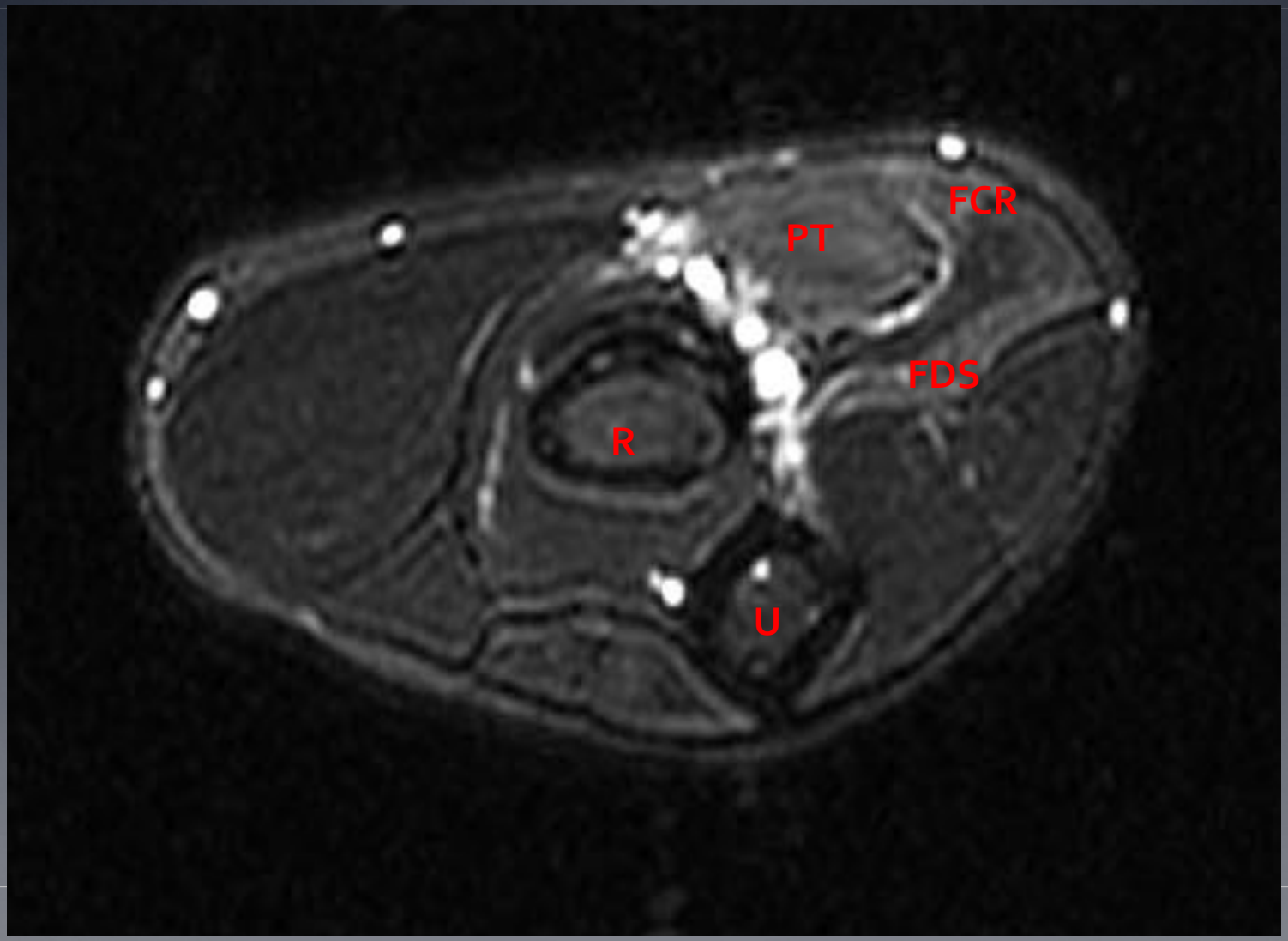
US left arm – mid humerus level



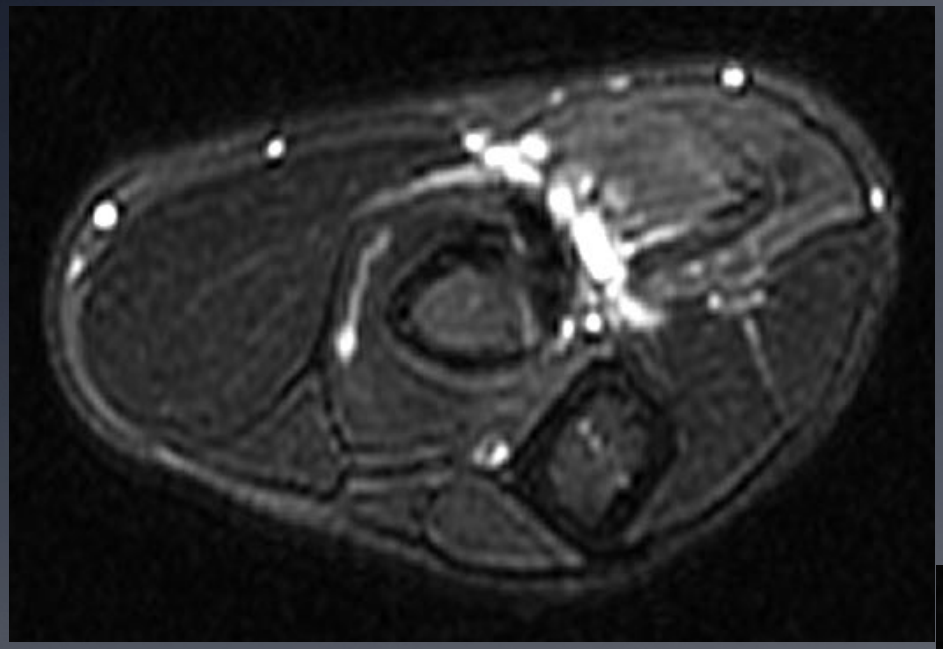
MRI left forearm

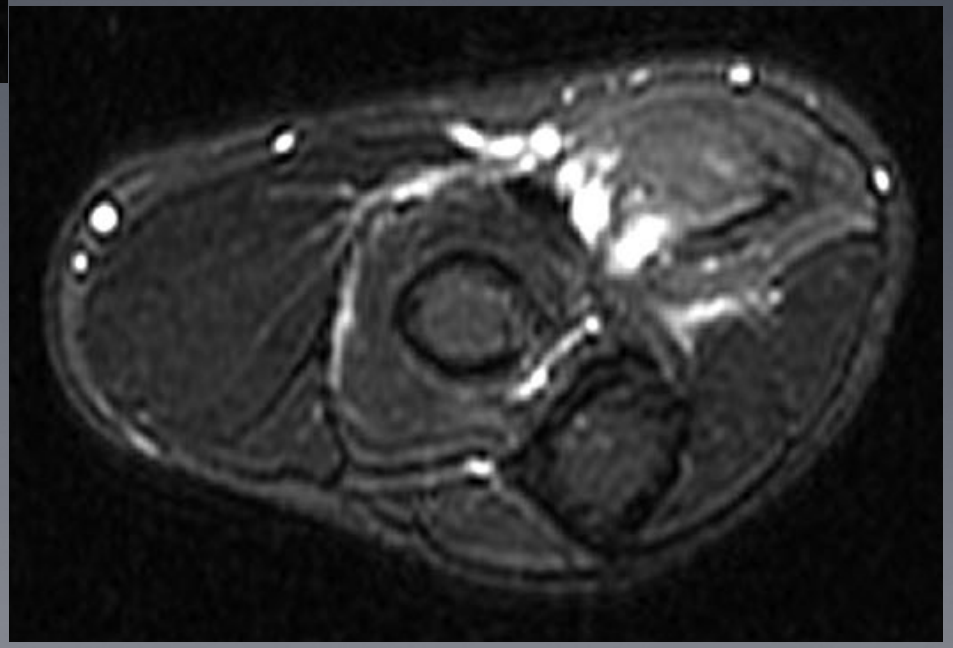


MRI left forearm



MRI left forearm



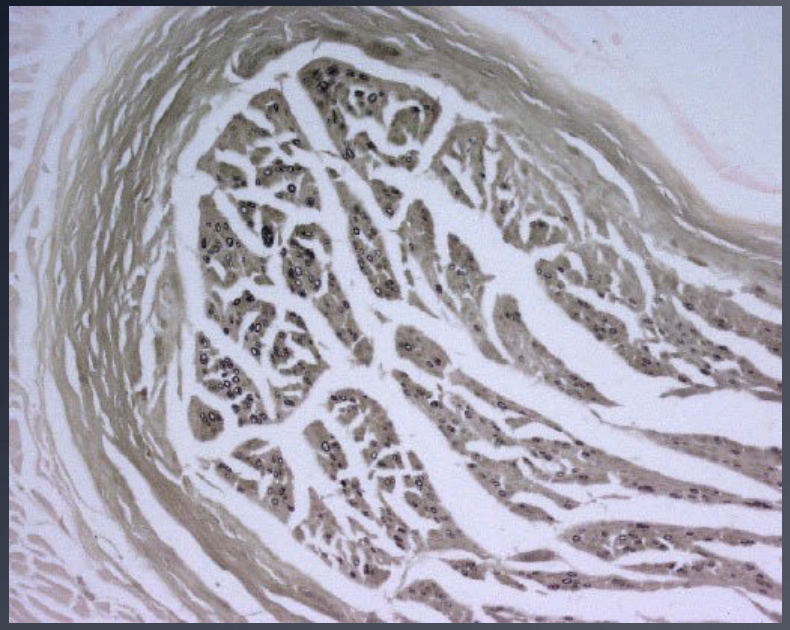


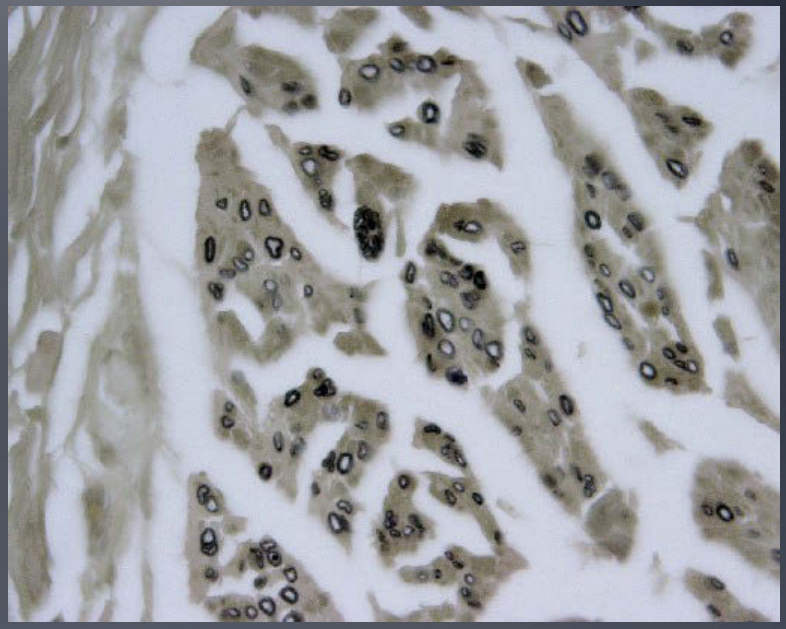
Investigations

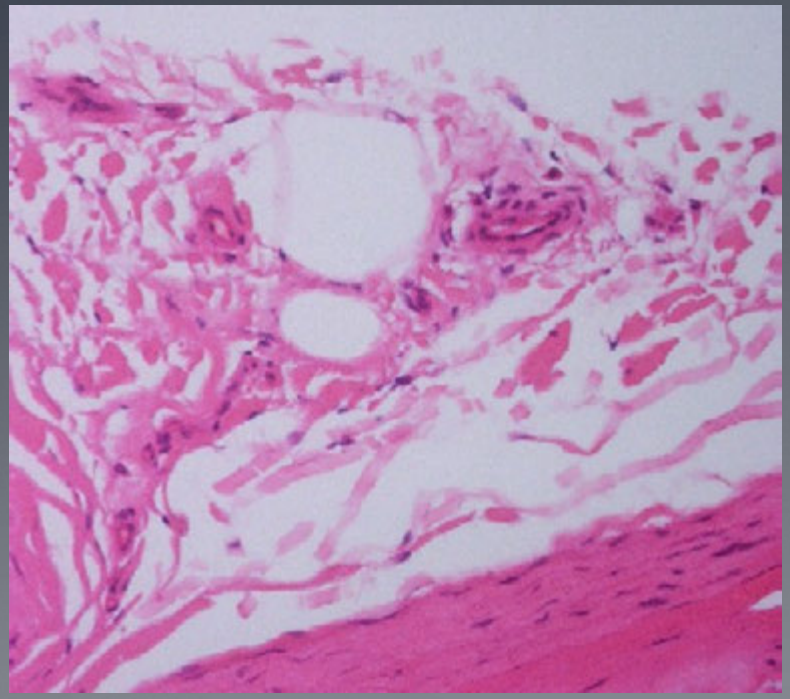
- CT chest/abdomen/pelvis – normal
- Skeletal survey normal
- BMAT – aberrant population plasma cells (~5 %), no abnormal lymphocyte population

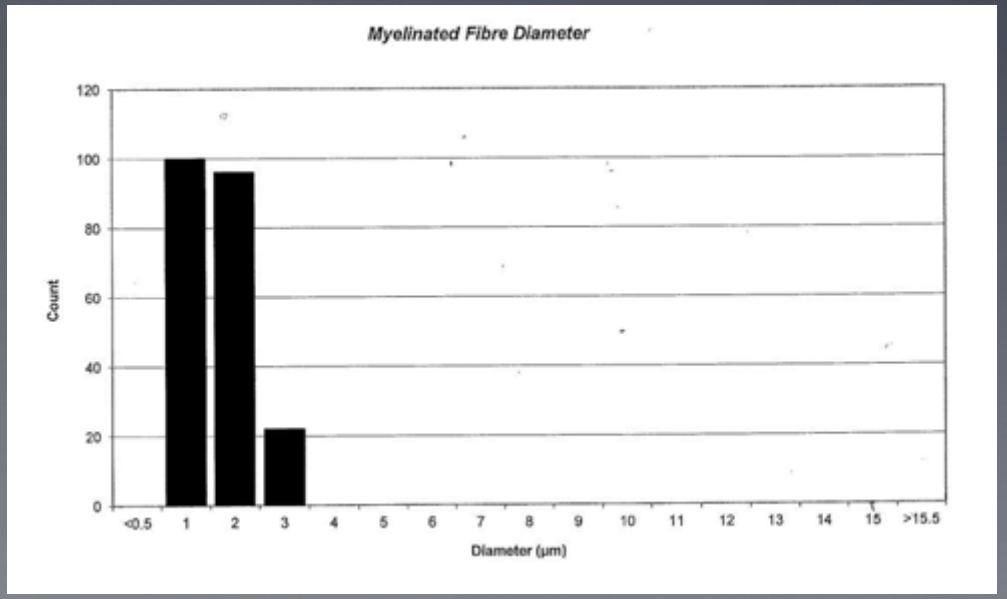
? Further investigation

? To treat or not to treat








Myelinated Fibre Diameter
Count
120
100
80
60
40
20
0
<0.5
1
2
3
4
5
6
7
8
9
10
11
12
13
14
15
>15.5
Diameter (µm)

Progress

- No treatment
- Repeat NCS/EMG unchanged
- Clinically stable – severe, left median neuropathy/ allodynia; multiple cutaneous mononeuropathies

J Peripher Nerv Syst. 2010 Sep;15(3):176-84. doi: 10.1111/j.1529-8027.2010.00281.x.

Peripheral Nerve Society Guideline on the classification, diagnosis, investigation, and immunosuppressive therapy of non-systemic vasculitic neuropathy: executive summary.

Collins MP[1], Dyck PJ, Gronseth GS, Guillevin L, Hadden RD, Heuss D, Léger JM, Notermans NC, Pollard JD, Said G, Sobue G, Vrancken AF, Kissel JT; Peripheral Nerve Society.

Nonsystemic vasculitic neuropathy: insights from a clinical cohort.

Collins MP[1], Periquet MI, Mendell JR, Sahenk Z, Nagaraja HN, Kissel JT.

Clin Exp Rheumatol. 2008 May-Jun;26(3 Suppl 49):S118-30.

Isolated vasculitis of the peripheral nervous system.

Collins MP[1], Periquet MI.

Front Neurol Neurosci. 2009;26:26-66. doi: 10.1159/000212368. Epub 2009 Apr 6.

Nonsystemic vasculitic neuropathy: update on diagnosis, classification, pathogenesis, and treatment.

Collins MP[1], Periquet-Collins I.

Muscle Nerve. 2010 May ; 41(5): 710–715. doi:10.1002/mus.21561.

Primary Amyloidosis Presenting as Upper Limb Multiple Mononeuropathies

Jennifer A. Tracy, M.D., Peter J. Dyck, M.D., and **P. James B. Dyck, M.D.**
Peripheral Neuropathy Research Laboratory, Department of Neurology, Mayo Clinic, 200 First Street SW, Rochester, Minnesota, USA 55905

- Patient with step-wise progressive, multiple upper limb mononeuropathies (left median, bilateral radials) over a 2 year period
- Monoclonal IgM λ (1g/dL)
- Left superficial radial N Bx – axonal deg and amyloid deposition within fascicles and around endoneurial microvessels
- Light chain amyloidosis may be the cause of a multiple mononeuropathy pattern in the absence of a more diffuse peripheral neuropathy
- Primary amyloidosis results from overproduction of monoclonal Ig light chains (λ in 2/3 patients) and subsequent deposition of light chains in vulnerable tissues including nerves
- Systemic complaints, including fatigue and LOW, common

MRI cervical spine + brachial plexus

Acta Neurol Scand. 1997 May;95(5):319-20.

Acute brachial plexus neuropathy as a presenting sign of peripheral nervous system involvement in paraproteinaemia.

Martinelli P, Macrì S, Scaglione C, Stumpo M, Poppi M.

- ABPN as a presenting sign of paraproteinemic neuropathy
- NCS/EMG – focal injury of the upper trunk of BP + mild, diffuse sensorimotor neuropathy
- IgGλ paraprotein
- BMAT – low grade plasmacytoma
- Sural N Bx – demyelinating neuropathy with IgG deposits in Schwann cells; c/w immunomediated mechanism
- ? Coincidental onset

J Neurol Sci. 2010 Apr 15;291(1-2):89-91. doi: 10.1016/j.jns.2010.01.018. Epub 2010 Feb 10.

Light chain deposition in peripheral nerve as a cause of mononeuritis multiplex in Waldenström's macroglobulinaemia.

Luigetti M[1], Frisullo G, Laurenti L, Conte A, Madia F, Profice P, Batocchi AP, Montano N, Tarnani M, Tonali PA, Sabatelli M.

- WM is a form of monoclonal IgM gammopathy a/w B-cell lymphoplasmacytic lymphoma
- Mononeuritis multiplex previously reported
- Nerve biopsy demonstrates light chain depositions which subverts the normal architecture of the endoneurium and epineurium resulting in fascicular hyalinosis and epineurial artery disruption
- Ig deposition is one of several mechanisms of nerve damage in IgM-related neuropathy

NEXT SPEAKER

Another cause for brachial neuritis?

- Richard W Frith

- Sydney Neuromuscular Meeting
- November 2014

“Brachial neuritis”

- Neuralgic amyotrophy
- Parsonage-Turner syndrome
- Brachial plexus neuropathy
- Brachial plexitis

- Acute focal inflammatory neuropathy (AFIN)
- Acute multifocal inflammatory neuropathy (AMFIN)

Brachial neuritis

- Typical syndrome
  - Antecedent event >50%
    - Infection
    - Exercise
    - Surgery
    - Vaccination
    - Trauma
  - Pain > 95%
    - Severe, relentless, nocturnal, mechanical exacerbation
    - Usually 3-6 weeks
    - >8 weeks – 10%
  - Weakness
    - Predominantly individual or multiple peripheral nerves
    - LTN, SSN, AN, AIN, and other motor nerves singly or in combination
  - Sensory symptoms common but comparatively minor

Brachial neuritis

- Immune mediated in some ?in all
- 1.6/100,000
- Familial form (up to 1/3 in some series)
  - Linked to chromosome 17q25, mutations in the septin (SEPT9) gene

- Differential diagnosis
  - Compressive cervical radiculopathy
  - Rotator cuff syndrome
  - Entrapment neuropathies
  - Other causes for focal neuropathy

Brachial neuritis

- Unusual patterns of disease
  - Any nerve or root
  - Lower cranial nerves
  - Phrenic nerve
  - Lower limb nerves - rare

  - ? Painful radiculopathy with normal imaging

Interscalene block & Brachial Neuritis

- 32 F
- 2006
- Acromioplasty & clavicle resection
- Interscalene block
- Typical brachial neuritis
- Axillary & suprascapular neuropathies
- 2010 – mild deficit

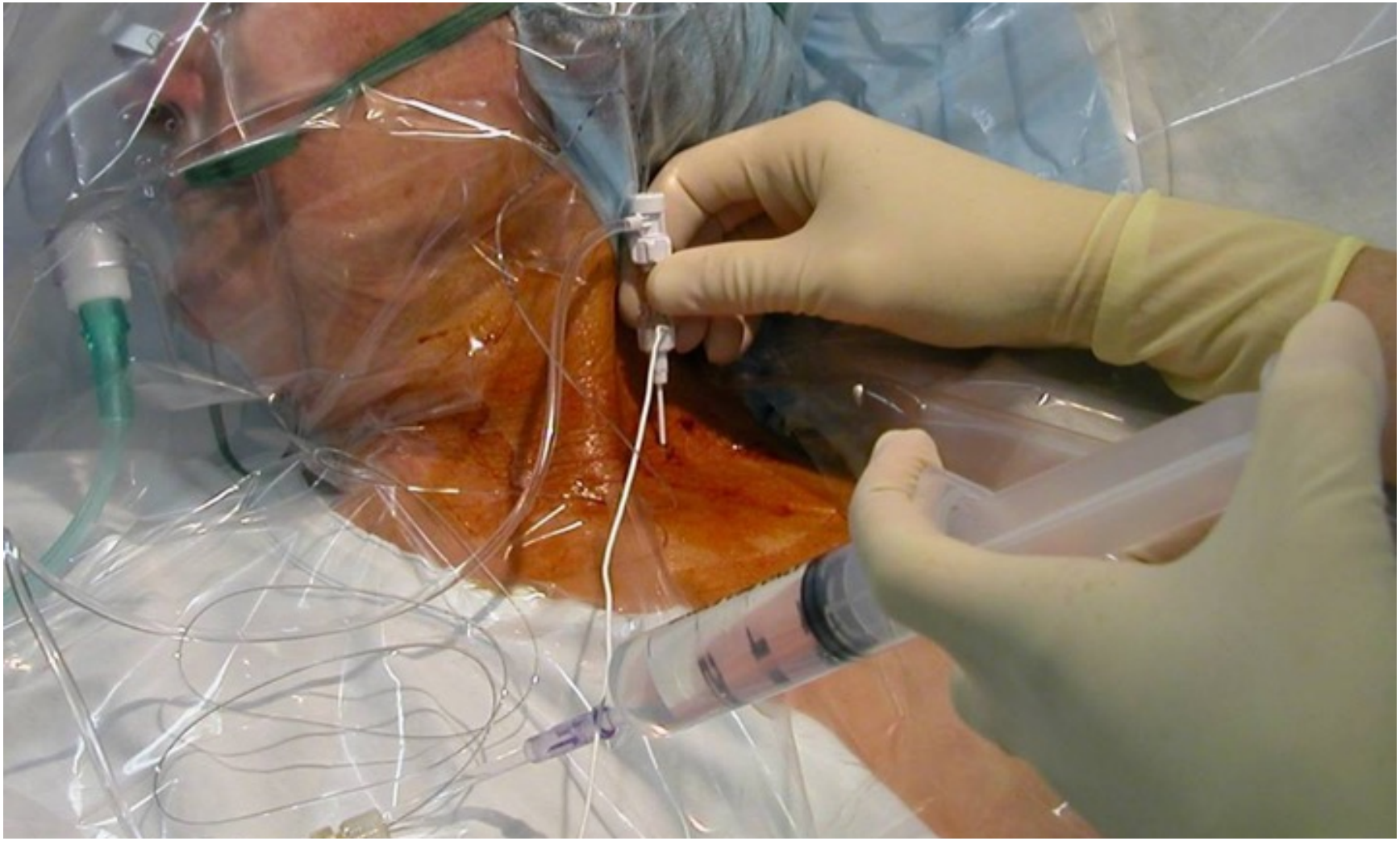
Interscalene block Cases

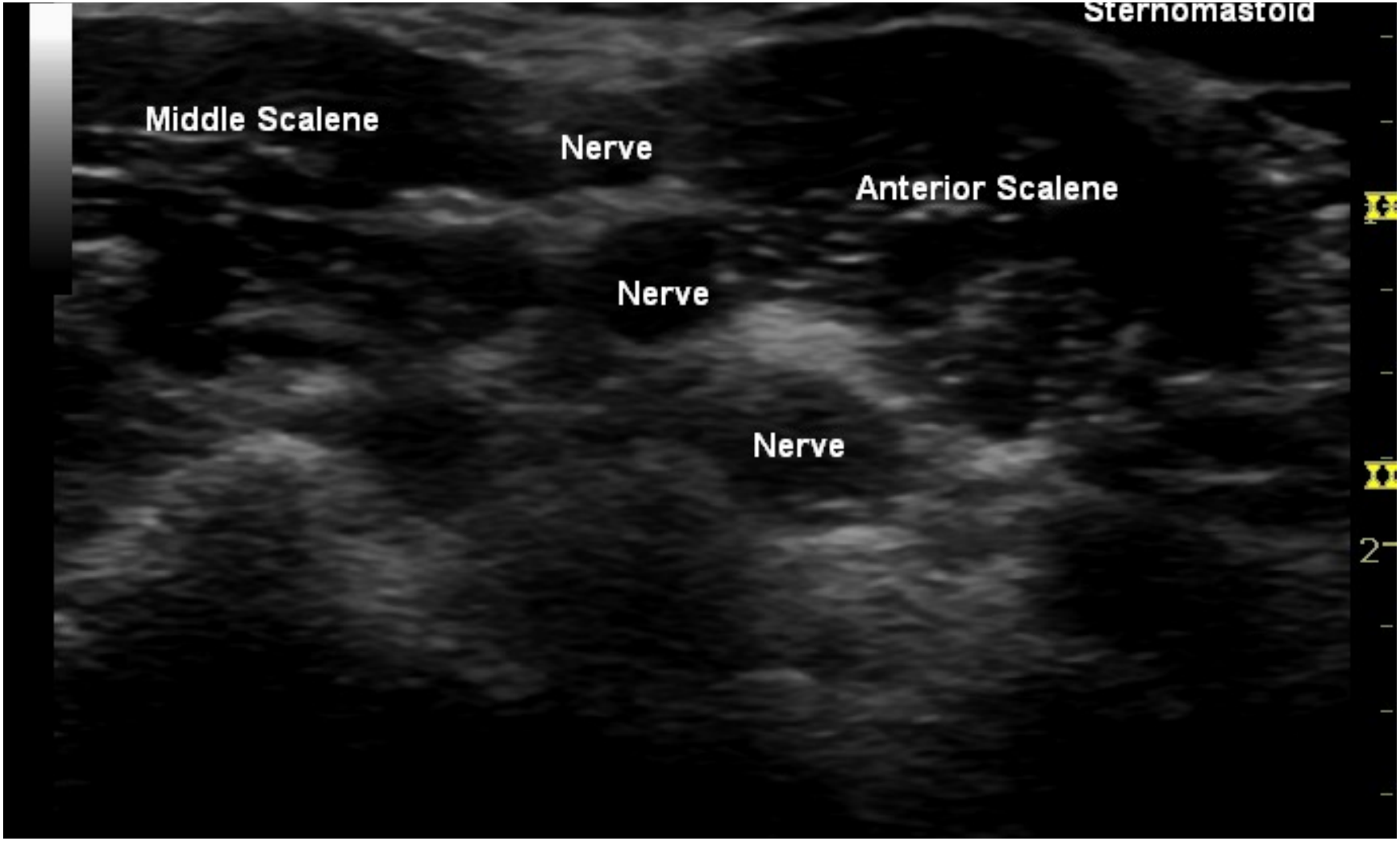
- All from one neurophysiology practice
- Shoulder surgery with interscalene block via catheter +/- elastomeric pump post-procedure
- Bupivicaine or ropivicaine
- Typical brachial neuritis pattern (pain, weakness, neurophysiology)
- Significant neurological deficit
- Comprehensive NCS/EMG

ISB Cases

- Excluded
  - Prior symptoms
  - No deficit
  - Prior trauma
  - Surgical complications
  - ACH/other private practice patients

Interscalene block




Sternomastoid
Middle Scalene
Nerve
Anterior Scalene
Nerve
Nerve
2

Interscalene block & Brachial Neuritis

49 F

AC joint reconstruction

Typical clinical picture for BN

Severe anterior interosseous neuropathy (completely denervated) & patchy median neuropathy

Interscalene block & Brachial Neuritis

63 F

TSJR

Severe median, ulnar & musculocutaneous neuropathies

Pattern – multiple peripheral nerves rather than plexus or root damage

Interscalene block & Brachial Neuritis

44 F

Rotator cuff repair

Severe axillary & SS neuropathies

Interscalene block & Brachial Neuritis

54 F

Rotator cuff repair

Lower trunk plexopathy

Interscalene block & Brachial Neuritis

57 M

Acromioplasty & rotator cuff repair

(Tight sling on arm ?radial neuropathy)

Severe posterior cord, musculocutaneous & SS neuropathies

Interscalene block & Brachial Neuritis

72 F

TSJR

Severe pain

PH uncomplicated SJR/interscalene block

Partial posteror cord lesion

Interscalene block & Brachial Neuritis

35 M

ORIF after clavicle fracture

Severe brachial plexopathy sparing spinati and trapezius

Severe neuropathic pain

Flail arm

Interscalene block & Brachial Neuritis

68 M

SJR

Severe nocturnal neuropathic pain

Hypersensitivity medial arm/forearm

Ulnar neuropathy plus

Brachial Neuritis – no ISB

49 M

Professional sportsman

Elective clavicle plating

No IS block but ropivicaine infusion into operative site

Severe pan-brachial plexopathy

Posterior cord most affected

Neurology review “brachial neuritis”

Brachial Neuritis

2009-2014 57 cases

- Spontaneous 23
- Trauma/repetitive activity 11
- Post-operative (non-shoulder) 5
- Interscalene block 17
- Ropivicaine wound infusion 1

- Contralateral to ISB 0

Interscalene block & Brachial Neuritis

Concerns/criticisms

- Flawed case ascertainment
- Unknown denominator
- Surgical or other trauma
- Coincidental ipsilateral BN
- Related to Interscalene block
  - Operator
  - Technique
  - Equipment
  - Drugs

Literature – ISB complications

- Single case reports
- Lenters et al J Shoulder Elbow Surg 2007;16:379-87

  27 plexus or nerve injuries in 3172 ISBs
- Singh et al J Bone Joint Surg Am. 2012;94:2040-6

  "Brachial plexitis" 3/1319
- Frederickson & Kilfoyle Anaesthesia. 2009 Aug;64(8):836-44

  Nerve injury at 30d 3.7% of 637

NEXT SPEAKER

Limping Along

Dr Timothy J Day
Royal Melbourne Hospital

Case report- 43 F

- May 2011: 43 yr, female scientist
  - PH: R sciatica, obesity surgery,
  - no systemic illness, no CT symptoms
  - FH: Nil neuromuscular, No medications
- 3-4 yrs painless weakness L ankle- limp
  - Can't stand on L toes, occas tripping
  - EX: Mod weakness L ankle PF, depressed AJs
    - Normal R leg, upper limbs, Normal sensation

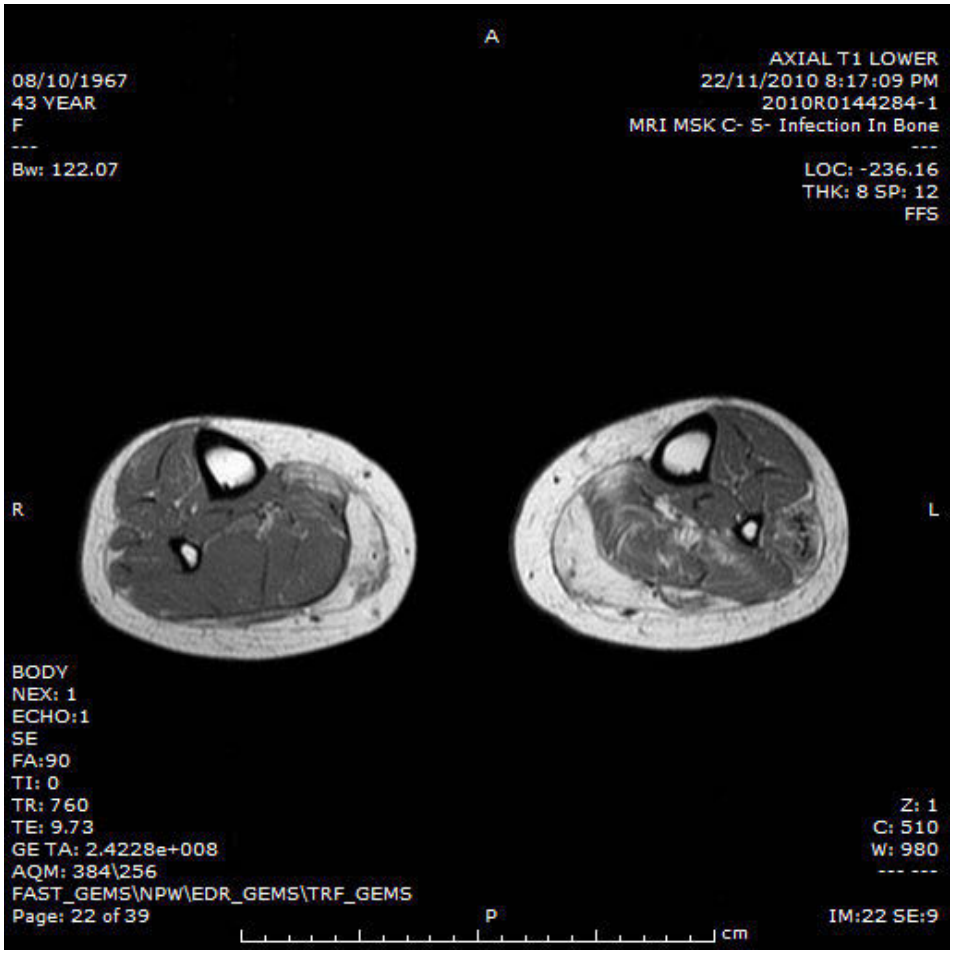
Case report- 43 F

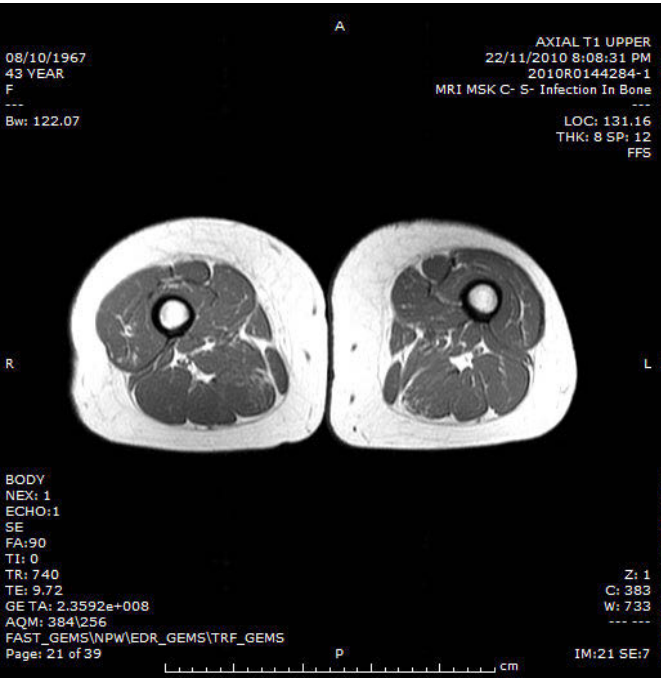
- Investigation:
  - CK 154, RhF: 2700, Abs negative
  - EMG- Patchy irritative myopathy in both calves, also in quads, gluteals, paraspinals
    - Fibrotic gastrocs
  - MRI-atrophy and abnormal signal in
    - posterior calves> ant & post thighs
      - Patchy changes in ant/lateral calf
  - Biopsy: “subtle inflammatory myositis”

EMG Findings

Muscle (Innervation)	Interpretation	Ins.act.	Spontaneous act.			Voluntary act.			
			Fib	PSW	Fasc	Amp	Dur	Poly	IP
Left T10	Normal		Nil	Nil	0				
Left L2	Myopathy		1+	2+	0	Normal	1-	1+	
Left L5	Mod activ Neur		1+	2+	0			1+	
Left Tibialis posterior (Tibial, L5 s1)	Sl activ Neur		1+	2+		Normal	Normal	Normal	Normal
Left Medial Gastroc (Tibial, S1 s2)	Myopathy	Decr.	2+	2+	0	1-	1-	1+	1+ Early
Right Medial Gastroc (Tibial, S1 s2)	Myopathy		2+	3+		1-	1-	2+	1+ Early
Left Peroneus longus (Superficial Peroneal, S1 l5)	Sl activ Neur		1+	2+	0	Normal	1+	1+	+/- Low
Left Gluteus medius (Inferior Gluteal, l4 L5 s1)	Myopathy		Nil	Nil		1-	1-	1+	+/- Low
Left Vastus lateralis (Femoral, l2 l3 L4)	Normal		Nil	Nil	0	Normal	Normal	Normal	Normal
Left Vastus medialis (Femoral, l2 l3 L4)	Myopathy		Nil	1+		1-	1-	1+	+/- Low
Right Vastus medialis (Femoral, l2 l3 L4)			1+	1+	0	Normal	1-	2+	1-
Left Tensor fascia latae (Inferior Gluteal, l4 L5)	Myopathy		1+	2+	0	1-	1-	Normal	+/- Low
Left Tibialis Anterior (Peroneal, l4 L5)	Sl activ Neur		1+	1+	0	Normal	1+	2+	1-
Right Tibialis Anterior (Peroneal, l4 L5)	Myopathy		1+	2+	0	1-	1-	2+	1-

Muscle MRI 2010

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43 YEAR
F

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MRI MSK C- S- Infection In Bone

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BODY
NEX: 1
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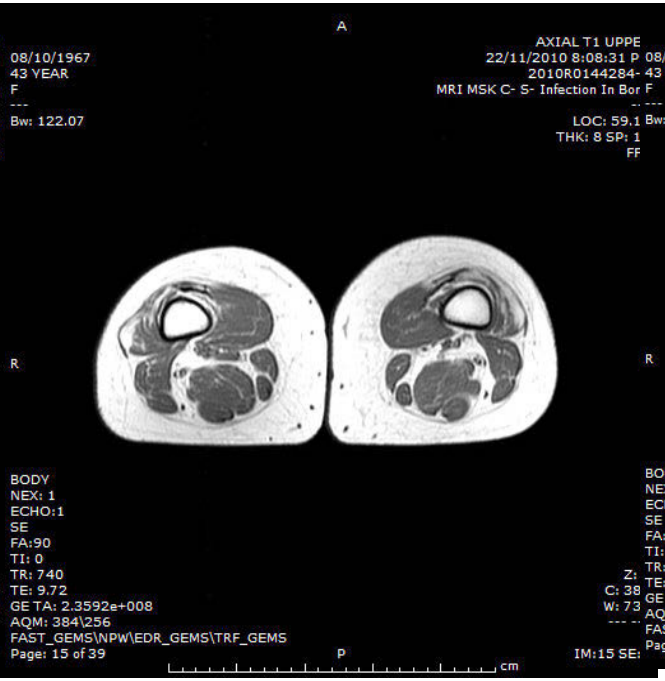
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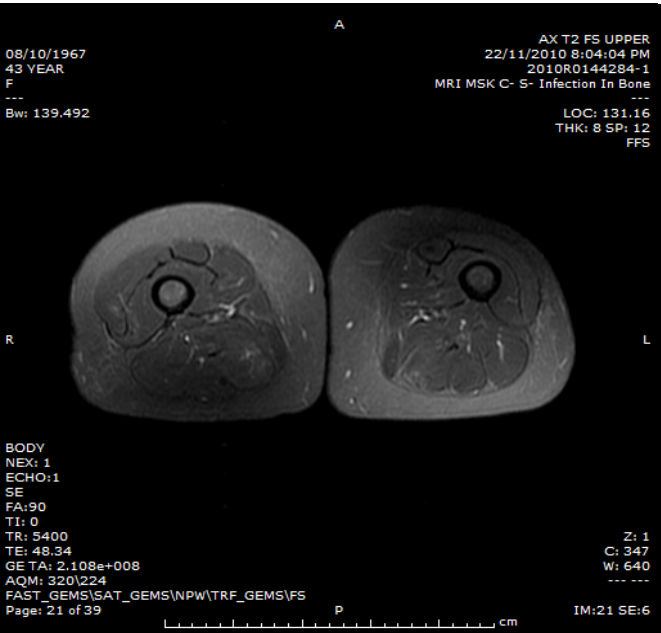
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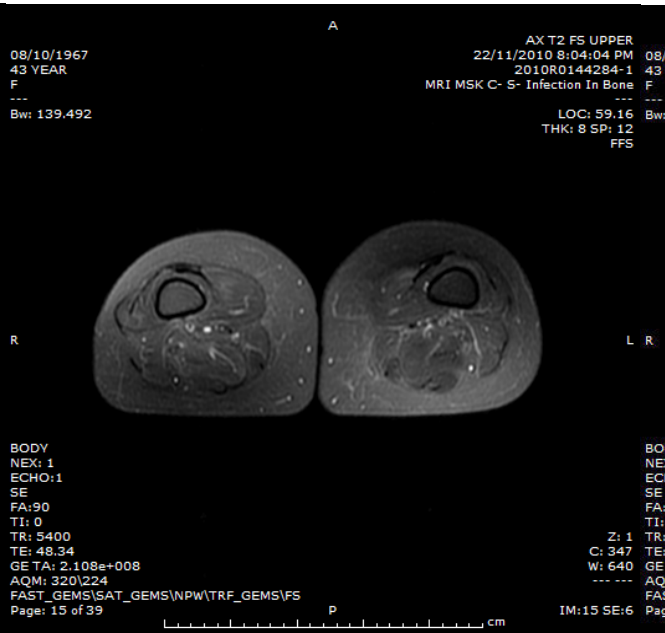
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cm


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08/10/1967
43 YEAR
F

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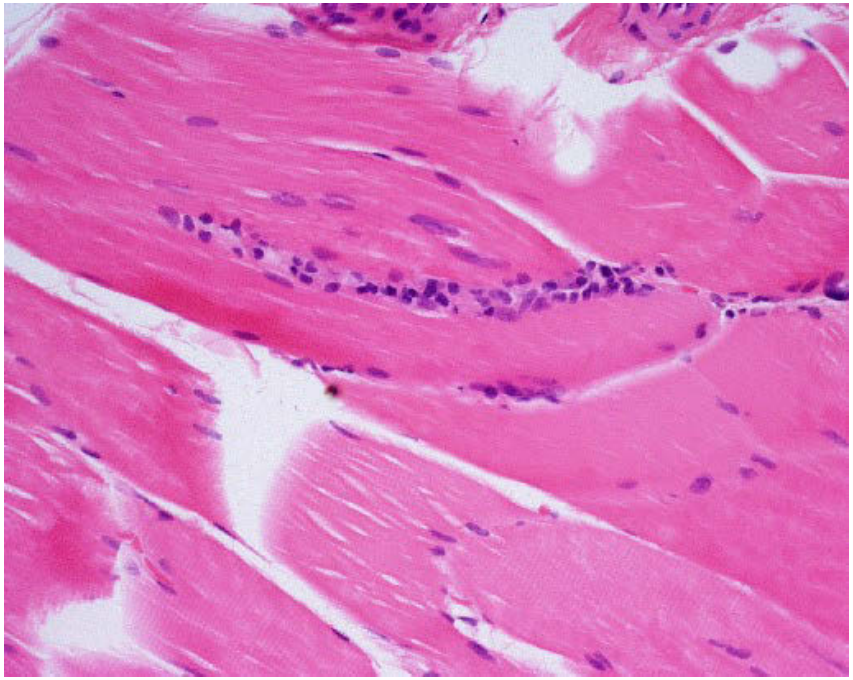
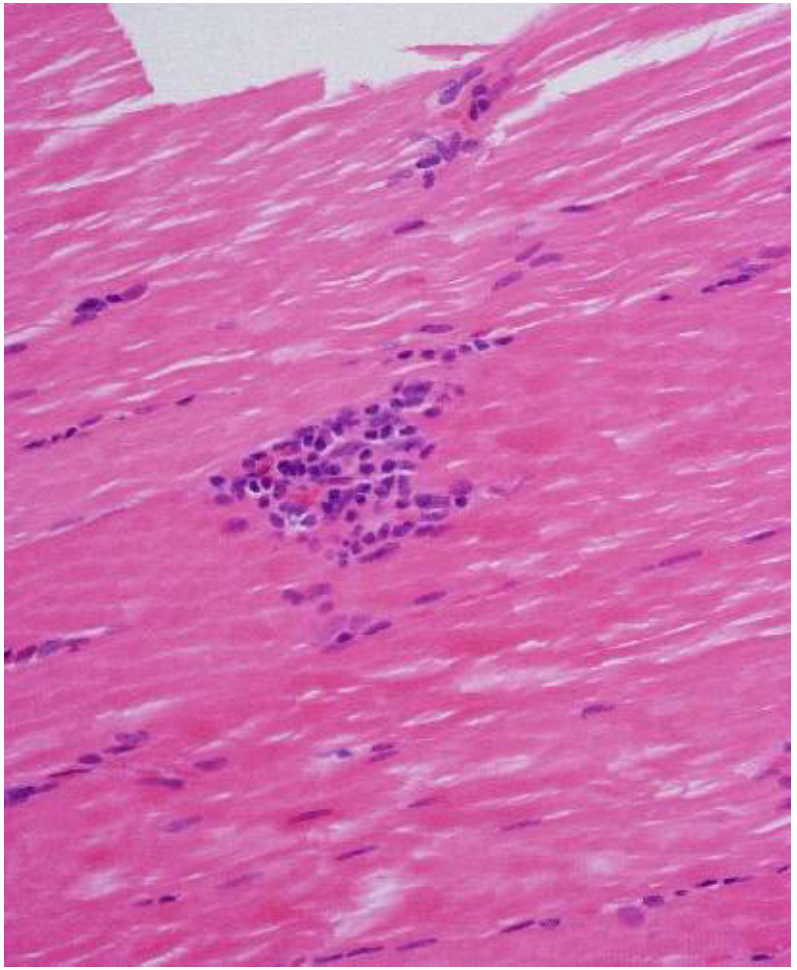
P
cm

Muscle Bx 2010- medial quad





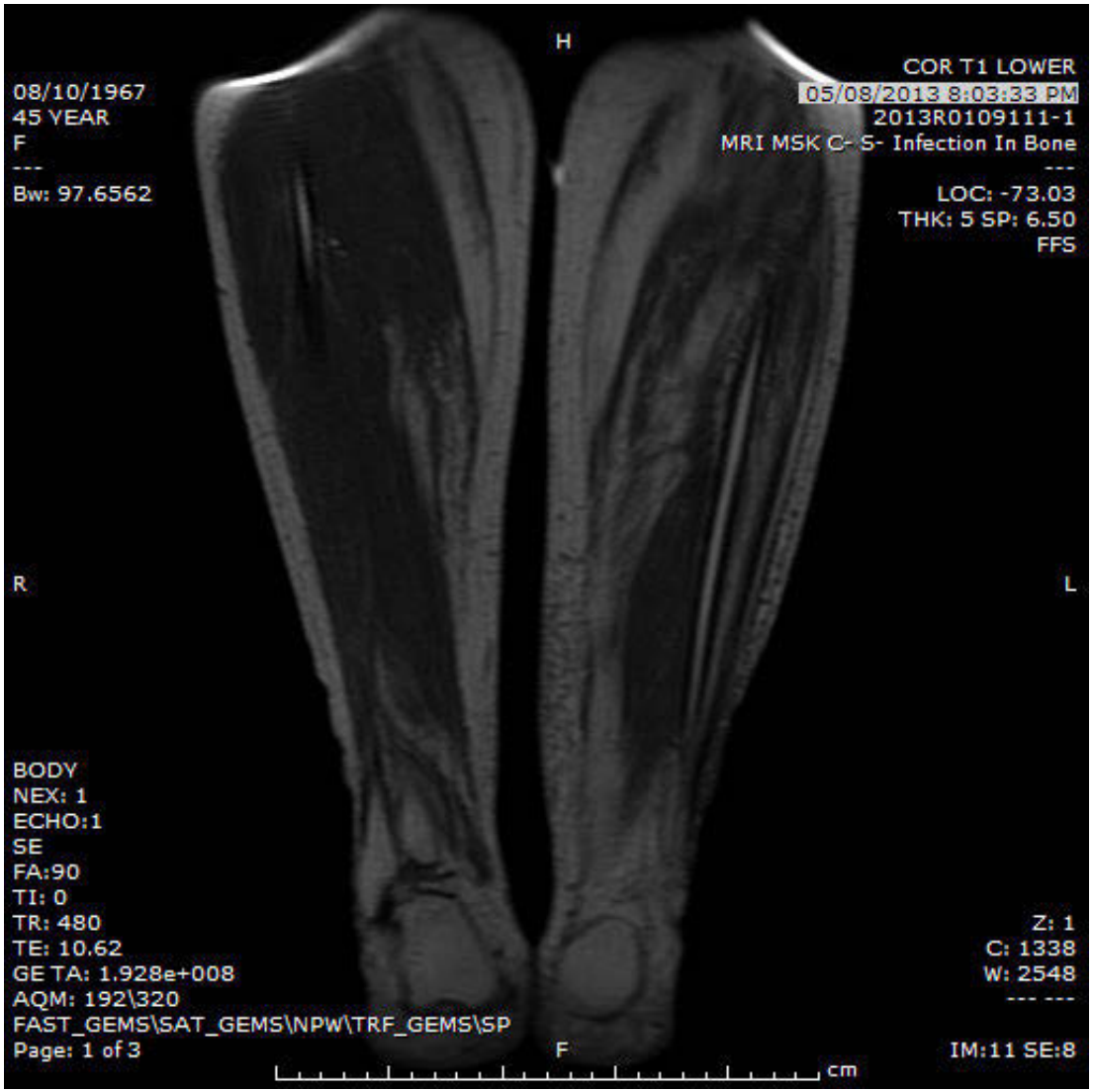
Progress- 2011-2012

- May 2011- mild limp, weak L post calf
  - CK 154 RF 2700 Bx ?polymyositis
  - Rheumatology- No CT disorder, no Rx
- Nov 2011- unchanged on no Rx
  - -CK 167 RF 1870
- Aug 2012- unchanged on no Rx
  - Mild oedema
  - CK 256 RF 2850

Progress- 2013

- Feb 2013- still limping, heaviness in legs, oedema of L calf
  - Ex: mild weakness R plantarflexion ,
    - mod-severe L plantarflexion, otherwise normal.
  - CK 287 RF 4110
  - Rheumatology review- still no features of CT disease, other Ab negative
  - MRI –increased patchy myopathic changes
  - EMG-LL patchy myositic changes

2013 MRI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45 YEAR
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MRI MSK C- S- Infection In Bone

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TE: 8.82
Z: 1
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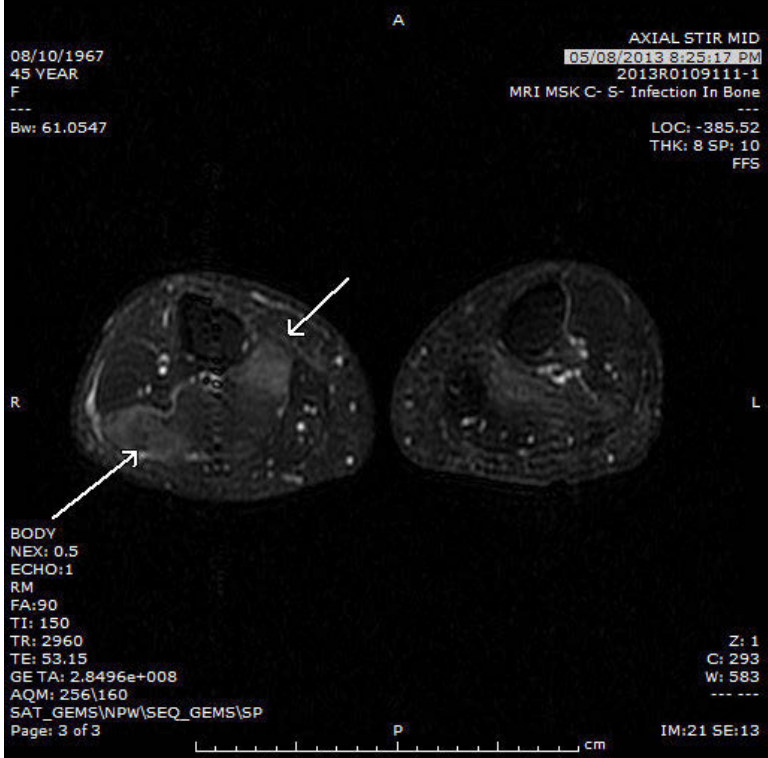
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L
BODY
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RM
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GE TA: 2.8496e+008
AQM: 256\160
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W: 583
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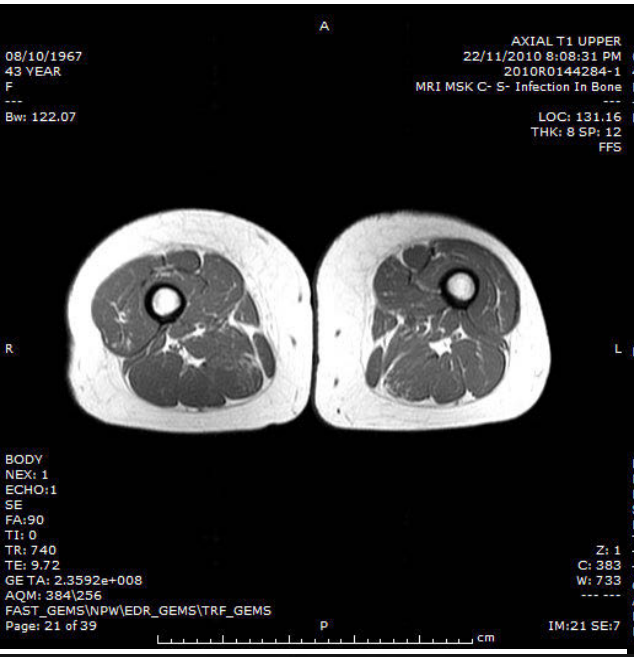
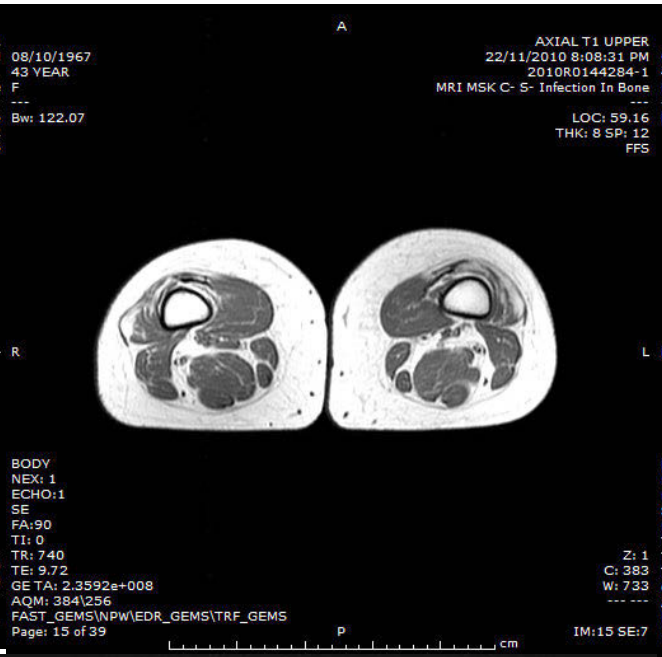
P
cm
IM:21 SE:13

2010 c.f. 2013 MRI



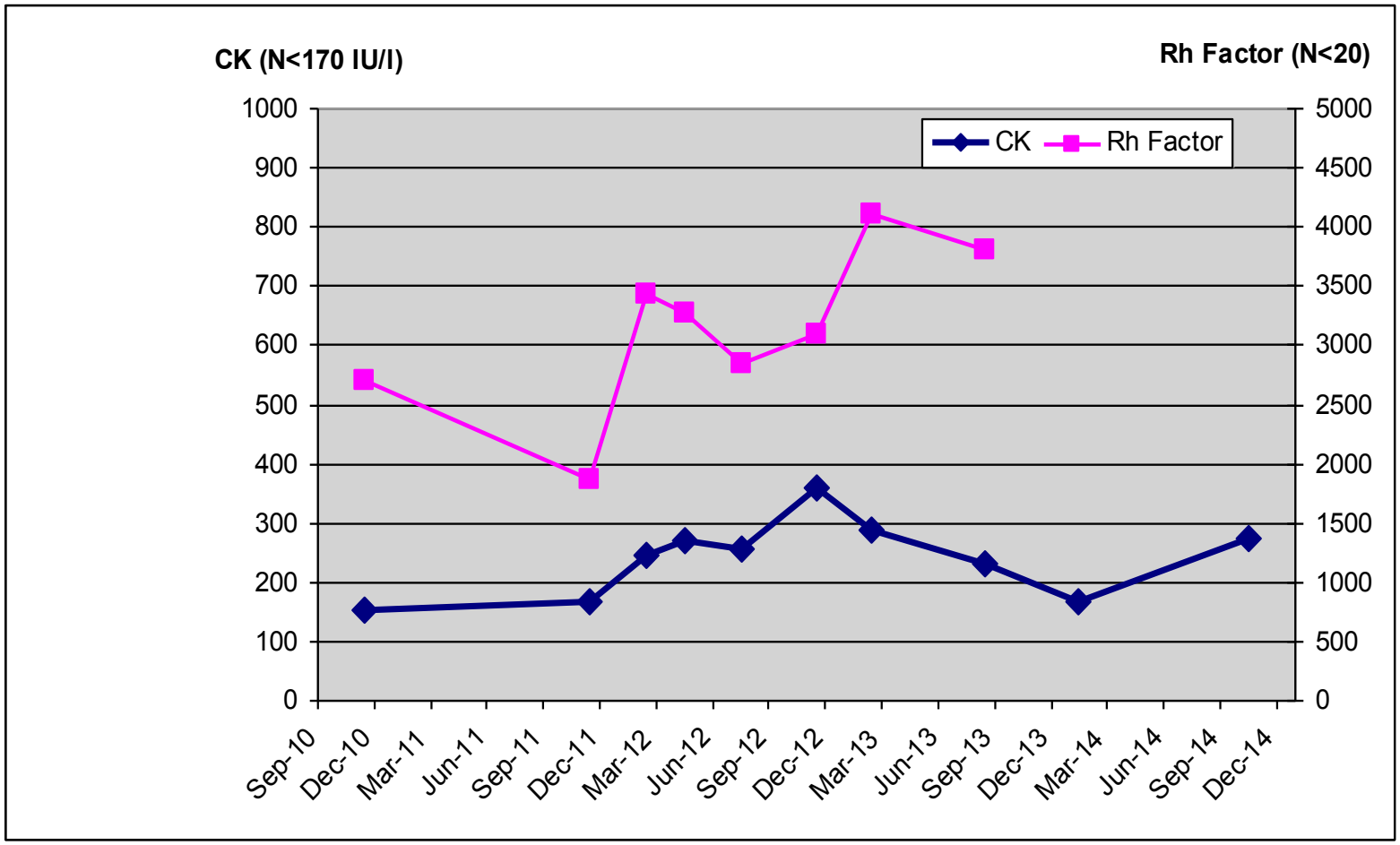


Progress- 2013

- Aug 2013- Incr leg weakness, pain and swelling
  - Ex: mild weakness R plantarflexion , mod-severe L plantarflexion, mild weakness L quads
  - Mild weakness right elbow flexion, finger extension, moderate weakness finger flexors bilaterally
  - CK 231 RF 3810 TRIM21 Ab +, others neg
  - Repeat biopsy- R calf

Progress- 2014

- March 2014- increased leg weakness
  - Problems with stairs, brisk walking
  - EX: Plantarflexion -2/-3
  - Finger flexors -2/-2, Elbow ext -1/-1.
  - CK 169
- Oct 2014- sl worse leg weakness
  - aPF -2/-3 EEx -1/-1 FF -2/-2
  - CK 274 RhF 711


CK (N<170 IU/l)
Rh Factor (N<20)
CK
Rh Factor
1000
900
800
700
600
500
400
300
200
100
0
5000
4500
4000
3500
3000
2500
2000
1500
1000
500
0
Sep-10
Dec-10
Mar-11
Jun-11
Sep-11
Dec-11
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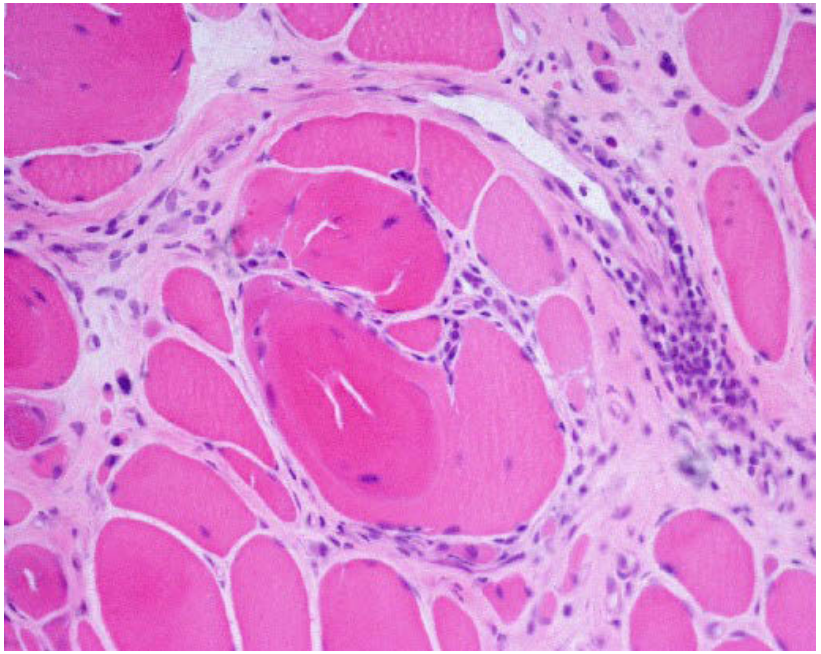
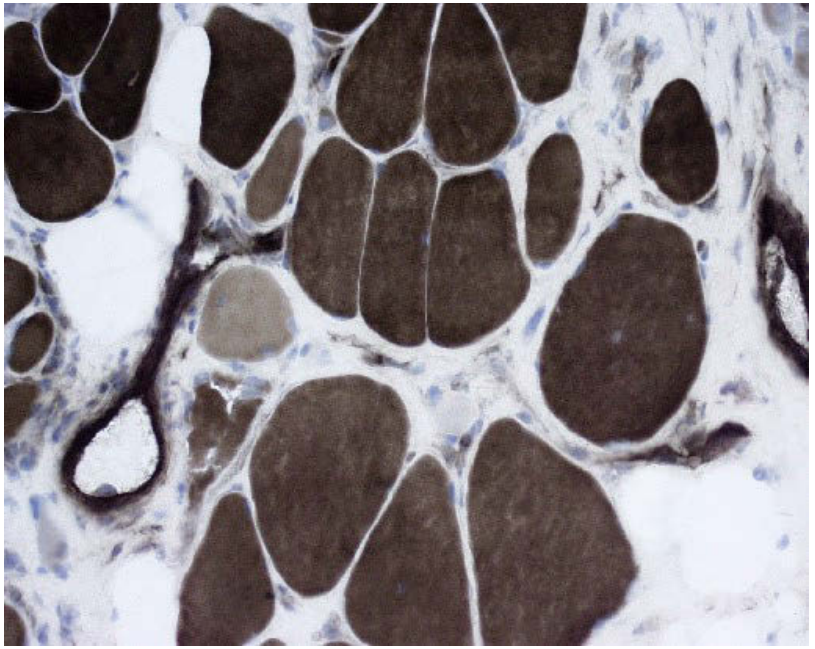
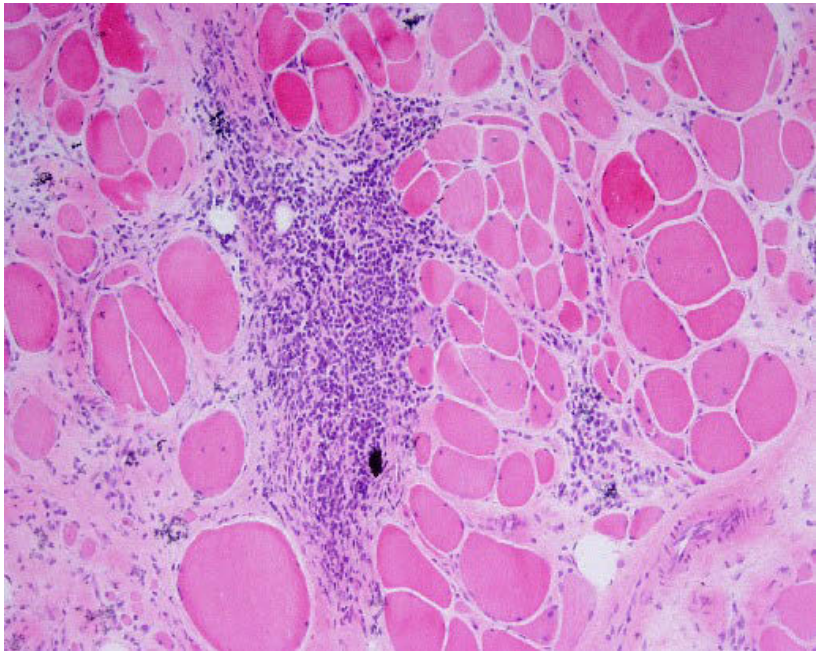
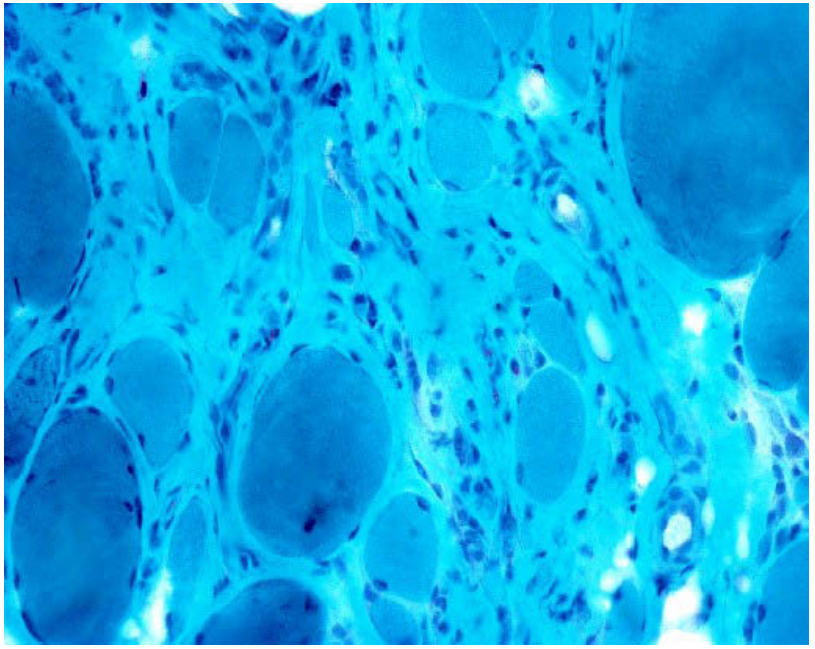
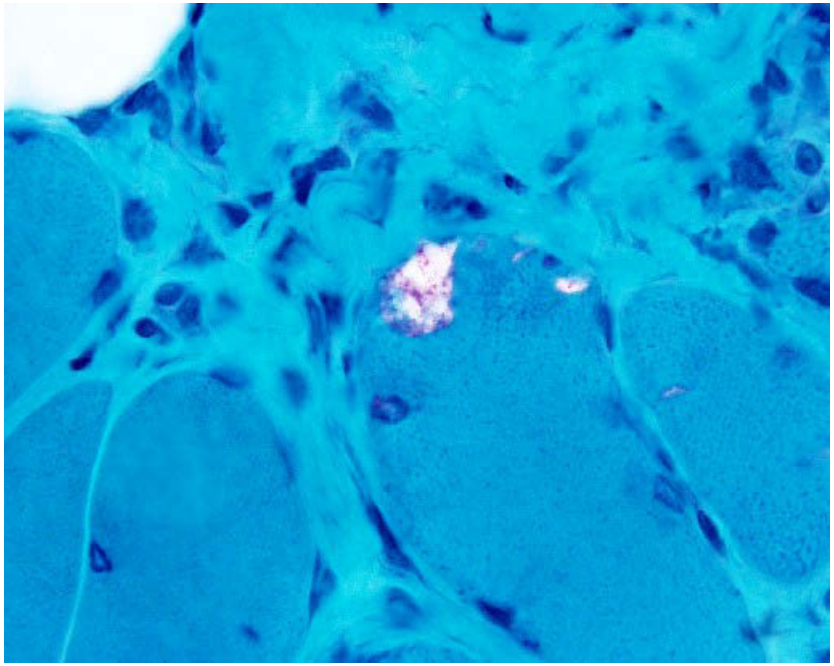
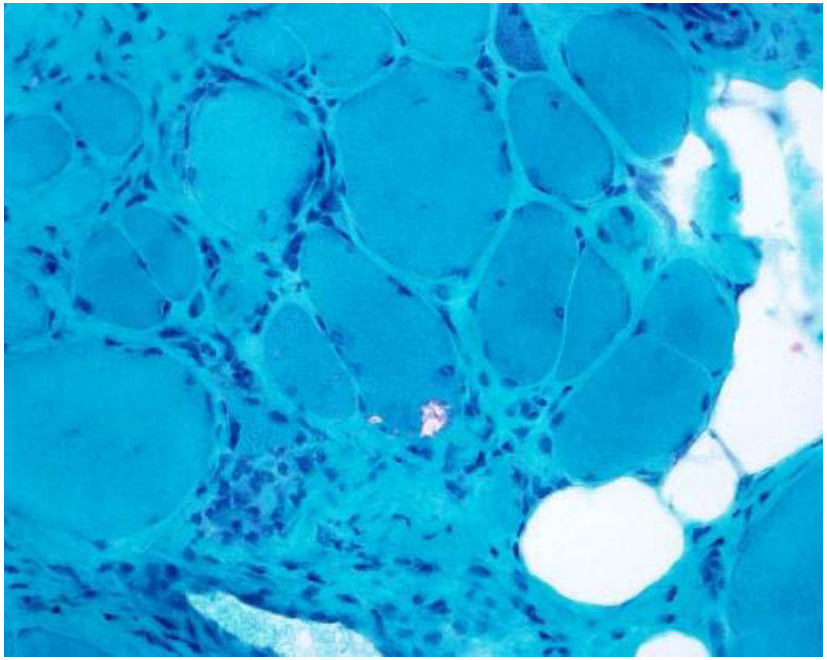
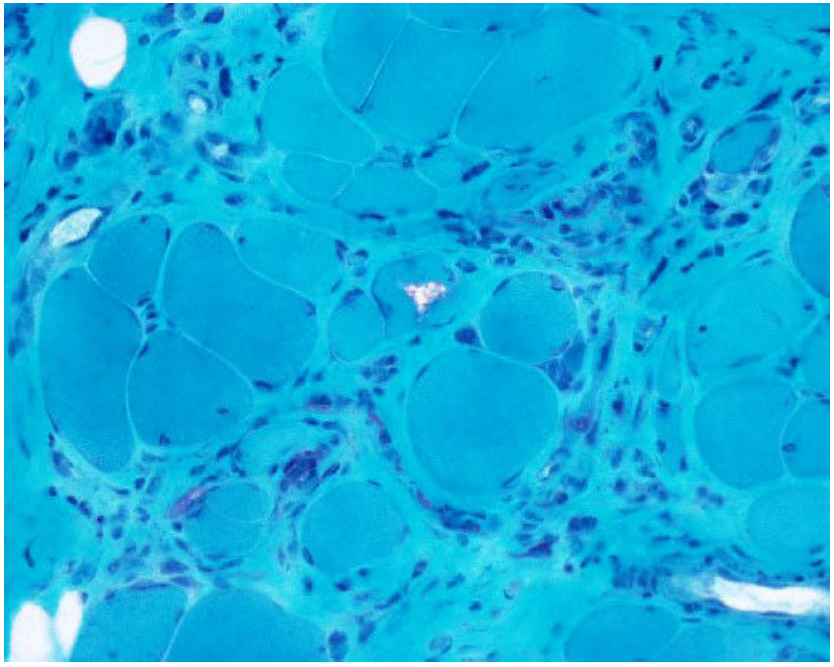
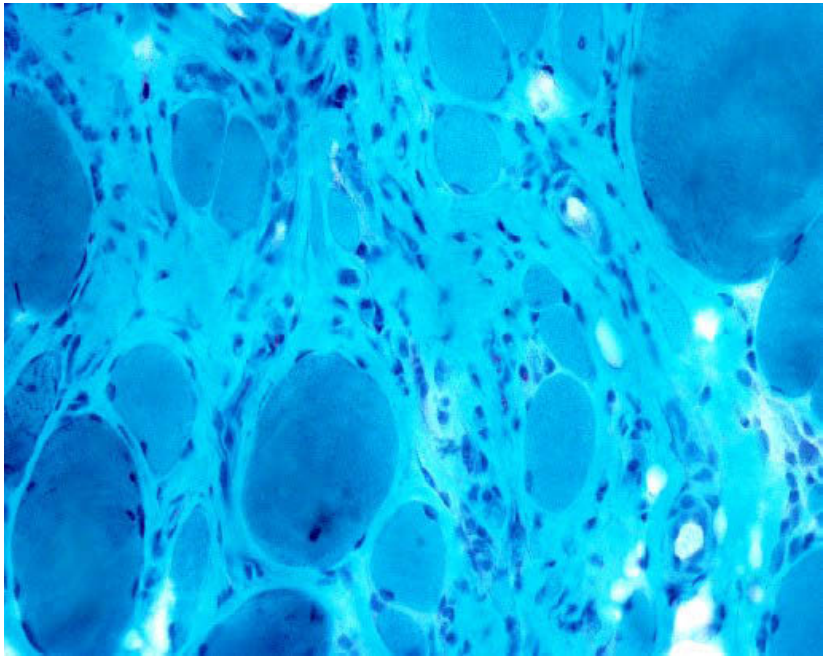
Dec-14





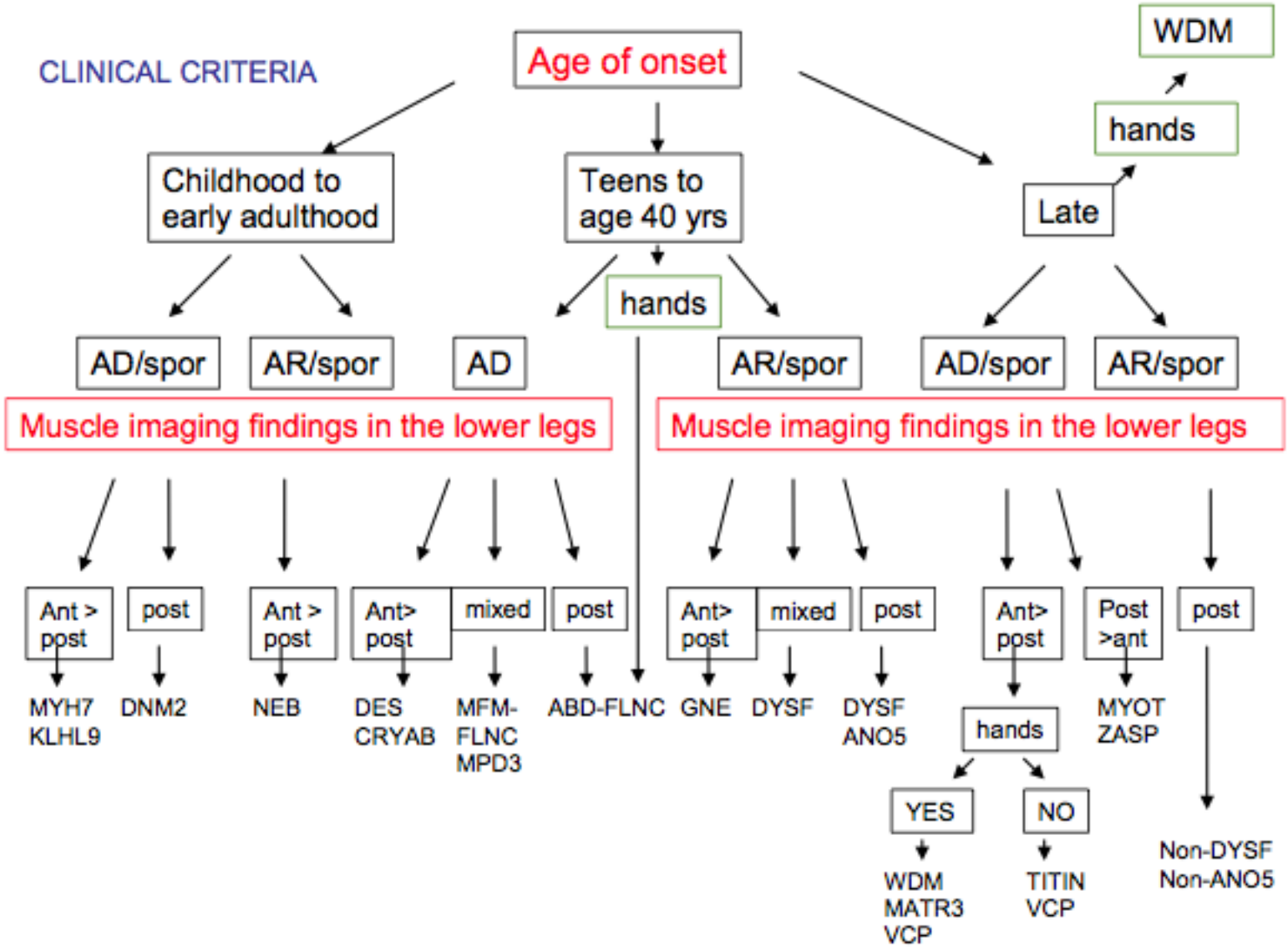




- Selective and indolent presentation
- Dystrophic muscle
- Inflammatory infiltrate and ++RhFactor
- Rimmed vacuoles

- ?Distal dystrophy
- ?atypical IBM


CLINICAL CRITERIA
Age of onset
Childhood to early adulthood
Teens to age 40 yrs
Late
WDM
hands
hands
AD/spor
AR/spor
AD
AR/spor
AD/spor
AR/spor
Muscle imaging findings in the lower legs
Muscle imaging findings in the lower legs
Ant > post
post
Ant > post
Ant> post
mixed
post
Ant> post
mixed
post
Ant> post
Post >ant
post
MYH7 KLHL9
DNM2
NEB
DES CRYAB
MFM-FLNC MPD3
ABD-FLNC
GNE
DYSF
DYSF ANO5
hands
MYOT ZASP
YES
NO
Non-DYSF Non-ANO5
WDM MATR3 VCP
TITIN VCP

Distal myopathy with posterior lower limb weakness

- Myotilinopathy – distal
- ZASP Markesberry-Griggs
- Distal ABD filaminopathy-Williams MPD4
- Miyoshi (dysferlin/anoctamin5)
- Dynamin 2

Distal myopathy with post LL weakness

- Myotilin LGMD 1A
- ZASP MFM4
- Filamin C MPD4 (Williams)
- Miyoshi

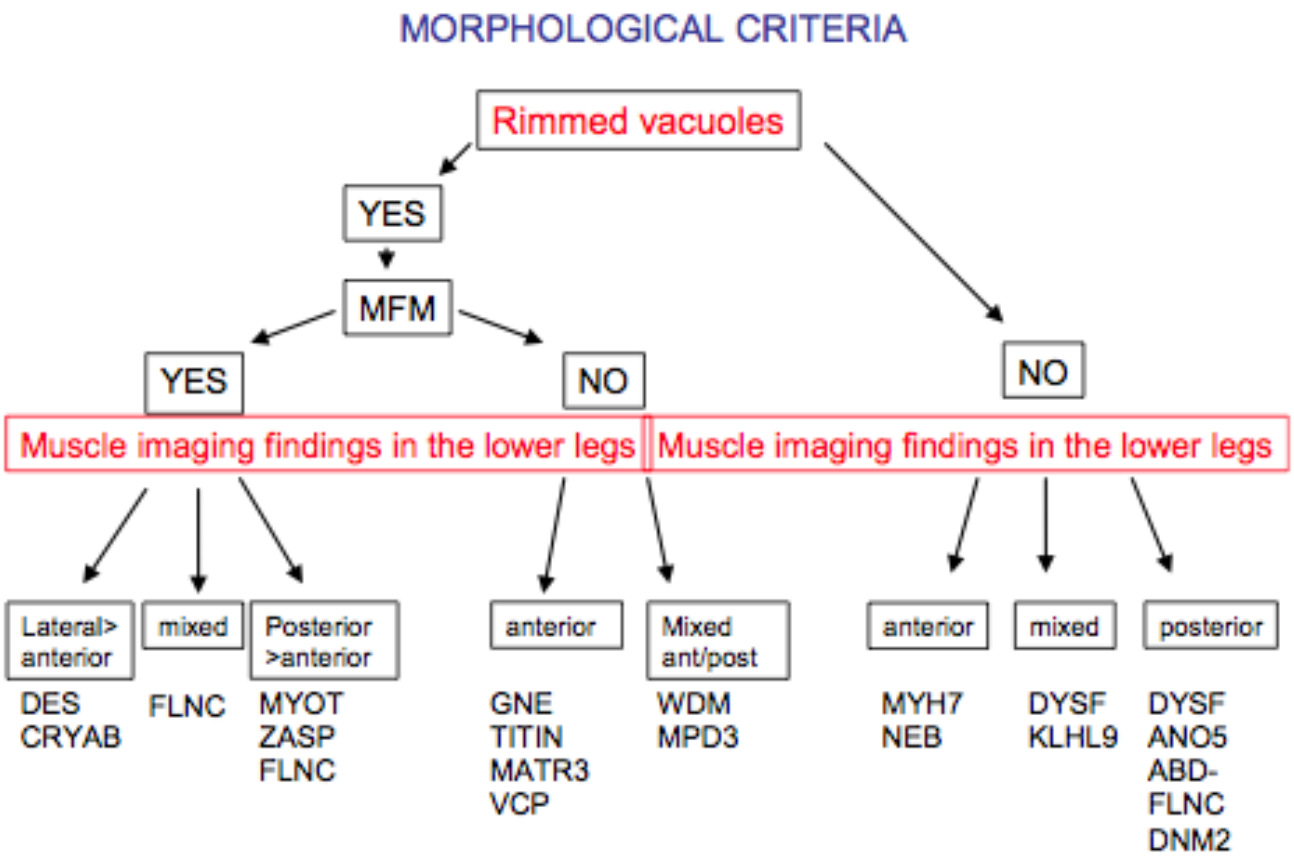
  dysferlin/anoctamin5
- Dynamin 2

. BUT

- Myofibrillar, RV+
- Myofibrillar, rare vacuoles, rarely distal
- Abs AJ but no vacuoles
- CK+++, no vacuoles
- Centronuclear, early onset

Flow chart algorithm for diagnostic purposes aiming at reducing the number of genes qualified for possible molecular genetic testing, using morphological data as the starting point.

From Udd B. Distal myopathies- New genetic entities expand diagnostic challenge Neuromuscular Disorders 22 (2012) 5-12.



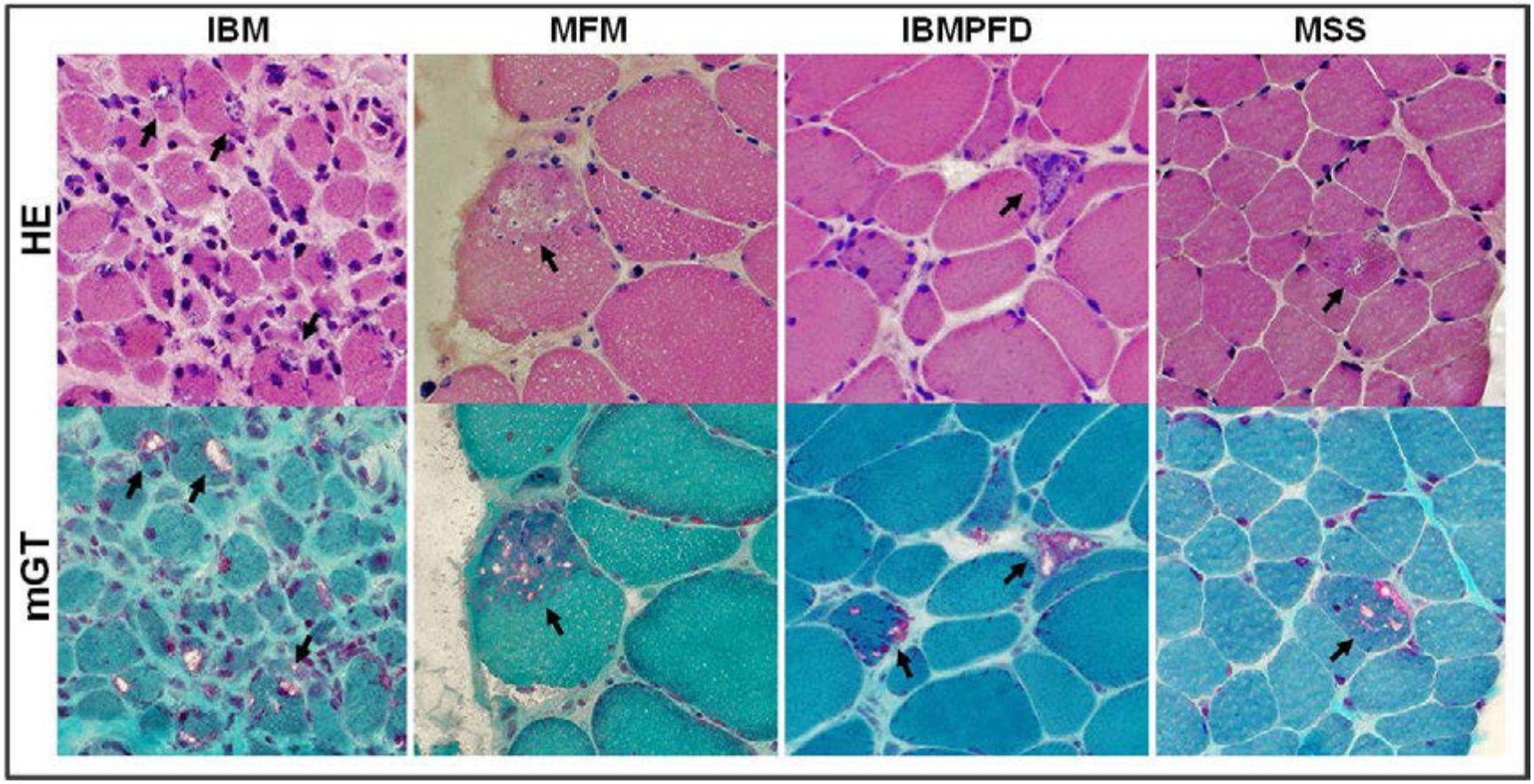
Distal myopathy with Rimmed Vacuoles

- GNE-Nonaka
- Titin- Udd, hIBM+resp failure
- VCP- hIBM+Pagets
- Welander
- MYH7-Laing myopathy
- Matrin3-MPD2
  - + vocal cord, pharyngeal
- MPD3
- LGMD 2G
- OPMD

- Anterior calf
- Anterior calf

- Anterior calf
- Anterior calf, hands
- Anterior calf
- Bulbar involvement

- Anterior & posterior calf
- Limb girdle
- Oculobulbar



- **FIGURE 4.Pathologic findings in myopathies with rimmed vacuoles.**

  Clinically and etiologically different disorders are showing same pathologic features (RVs; arrows) in skeletal muscles. HE-hematoxylin and eosin; mGT-modified Gomori trichrome; IBM-inclusion body myositis; MFM-myofibrillar myopathy; IBMPFD-inclusion body myopathy with Paget's disease of bone and frontotemporal dementia; MSS-Marinesco-Sjögren syndrome.

Best fit???

- MPD3 in non-Finnish family
- Atypical MPD4 with vacuoles
- Atypical Welander- LL onset
- MPD2 before vocal cord onset

- Next-Generation Sequencing
  - No recognised pathological mutations

MPD3 vs MPD4

- Autosomal Dominant
- Epidemiology: Single Finnish family
- Onset ~ 32 to 45 years with clumsiness of hands or legs
- Weakness ?asymmetric
  - Distal LL :Anterior & Posterior
    - TA, EDL, Gastroc, Glut medius; TFL
  - Hands -APB, OP, FDI, ADM
  - Progressive over years to more proximal limbs:
    - Forearm, Triceps, Infraspinatus, Proximal legs
- Lab- EMG: Myopathic
- CK: Normal or Slightly elevated
- Muscle biopsy
  - Myopathy: Severe; Endomysial fibrosis; Fiber size variation
  - Rimmed vacuoles
  - Cytoplasmic inclusion bodies

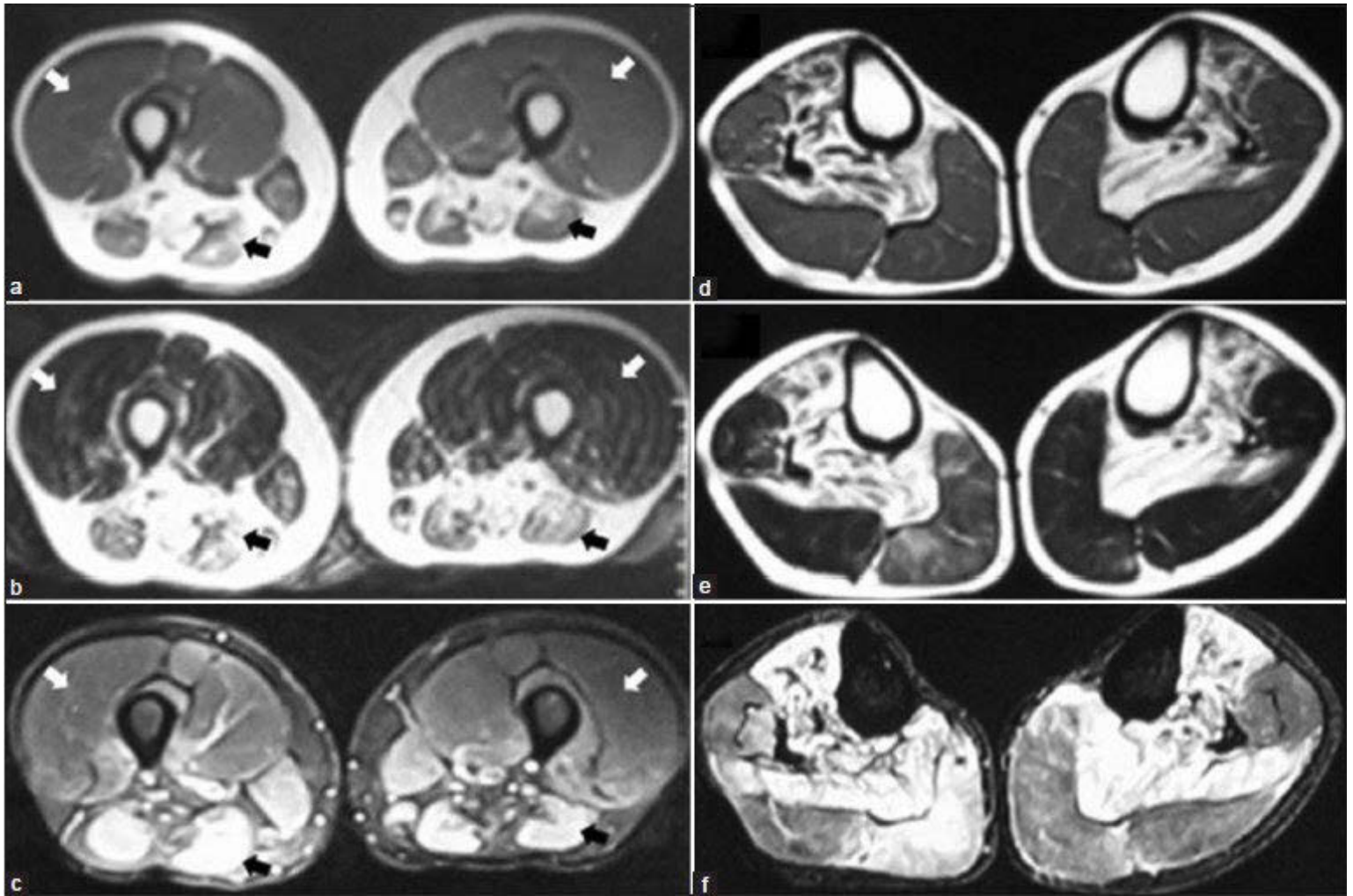
- Autosomal Dominant
- Epidemiology: Australian & Italian families
- Onset up to 30 years
- Weakness symmetrical
  - Distal arm & leg weakness
    - Forearm pronators, Finger flexors, Intrinsic hand muscles
    - Ankle evertors, Plantar-flexors (Calf atrophy)
    - Sparing: Anterior leg; Posterior arm
  - Slowly progressive
- Also-
  - Cramps and myalgia, worse after exercise
  - Tendon reflexes: Absent ankle jerks
  - Also: Cardiomyopathy: 2 patients
    - No respiratory involvement
- Lab- EMG: Myopathic
- CK-normal or mildly elevated
- MRI ?asymmetric involvement of posterior & lateral leg muscles
- Muscle
  - Varied fiber size: Small angular fibers
  - Internal architecture: Irregular in some biopsies
  - No myofibrillar aggregates, vacuoles or inflammation

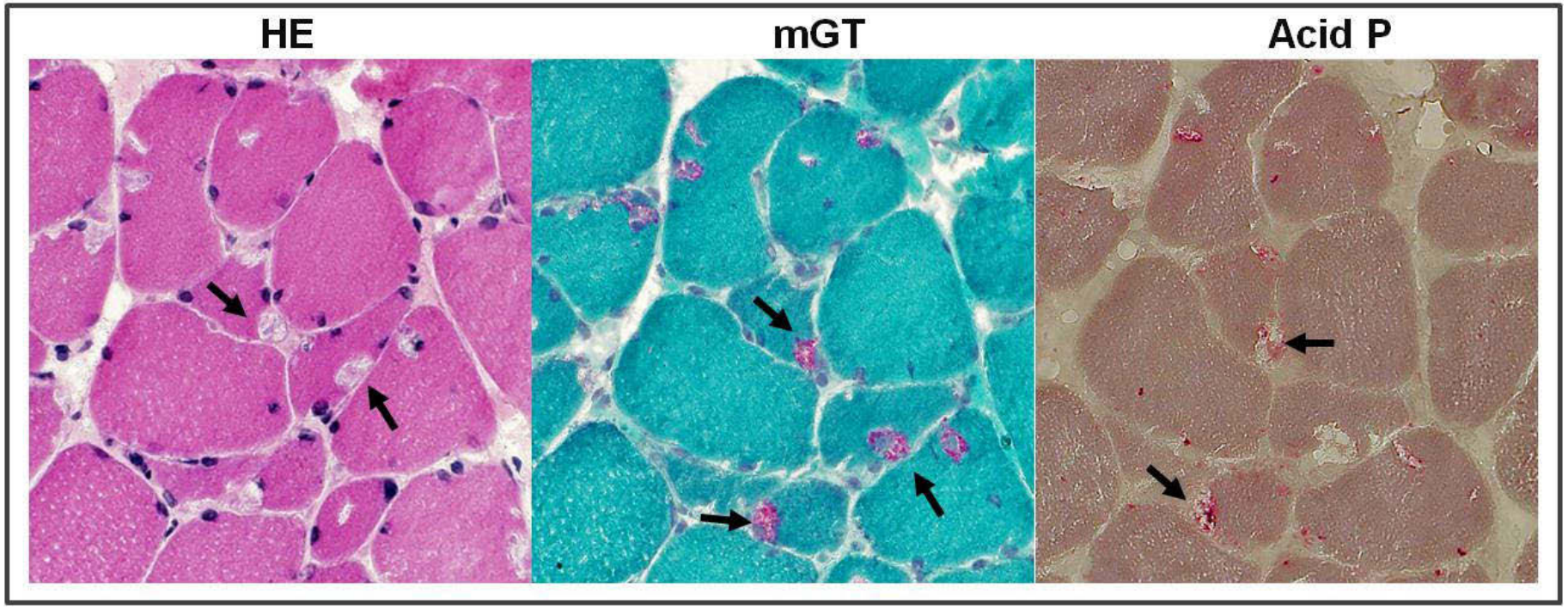
h IBM- ?Nonaka myopathy

- CF: Distal weakness- sparing quads.
- 4 GNE assoc'd myopathies
  - Quadriceps sparing myopathy- Argov
  - Distal myopathy with rimmed vacuoles- Nonaka
  - Proximal weakness with quadriceps sparing
  - Quadriceps-sparing myopathy +inflammation
  - Variants of same disease!

Figure 1: Patient with (a) severe wasting of the anterior leg muscles and mild wasting of the adductors but relatively spared gastrocnemius and quadriceps, (b) severe wasting of tibialis anterior with foot drop and (c) minimal wasting of forearm muscles with preserved small muscles of hands. Had mild disability

Figure 2: T1, T2, STIR (a, b, c respectively) weighted axial images of both thighs showing hyperintensities in hamstring muscles with atrophy (black arrows) sparing quadriceps (white arrowheads). T1, T2, STIR (d, e, f respectively) weighted axial images of both legs showing hyperintensities in tibialis anterior and posterior and gastro-soleous muscles on T2 and STIR

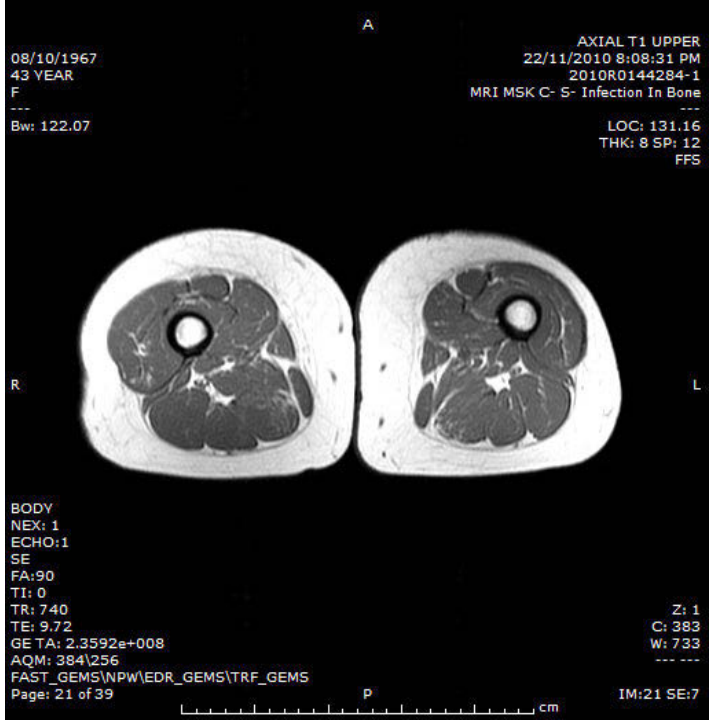




- Muscle pathology (Figure 5) is characterized by the presence of RVs predominantly in atrophic fibers, which are occasionally aggregated and form small groups. These RVs are actually clusters of autophagic vacuoles and multi-lamellar bodies. They often contain congophilic amyloid material and deposits that are immunoreactive to β–amyloid and its precursor protein, ubiquitin, and tau protein. Ultrastructurally, the filamentous inclusions measuring 15-20 nm in diameter are seen in both cytoplasm and nucleus with the presence of autophagic vacuoles. Necrotic and regenerating fibers can be rarely seen in GNE myopathy

- Bx: Filamentous inclusions and autophagic (rimmed) vacuoles
  - No inflammation
- DNA- GNE gene affects sialic acid pathways… sialyation deficiency
  - Mainly mis-sense mutations (>50 recognised)
  - Worldwide distribution, esp Persian Jews, Japanese

- Why sparing of quads?
- Why autophagic vacuoles/inclusions develop
- Can Mannose NAc supplementation help?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08/10/1967
43 YEAR
F

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MRI MSK C- S- Infection In Bone

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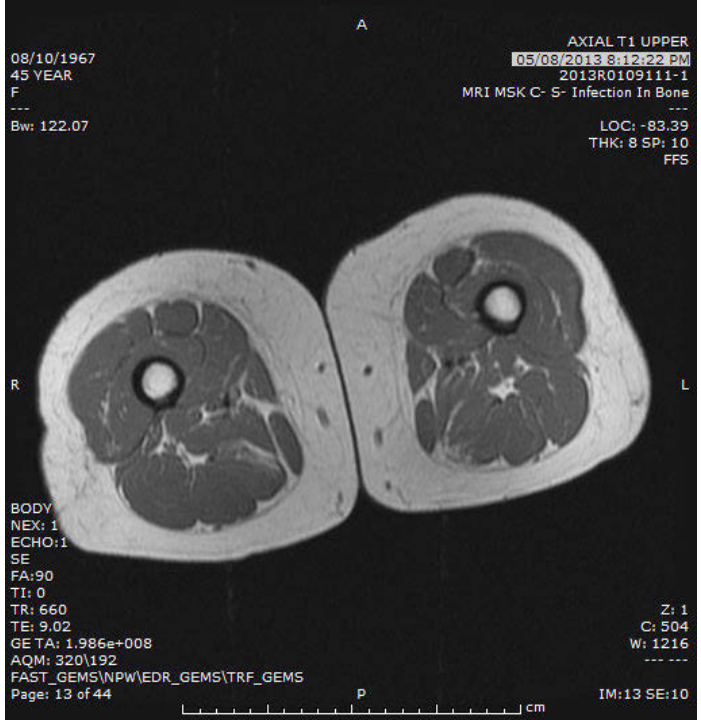
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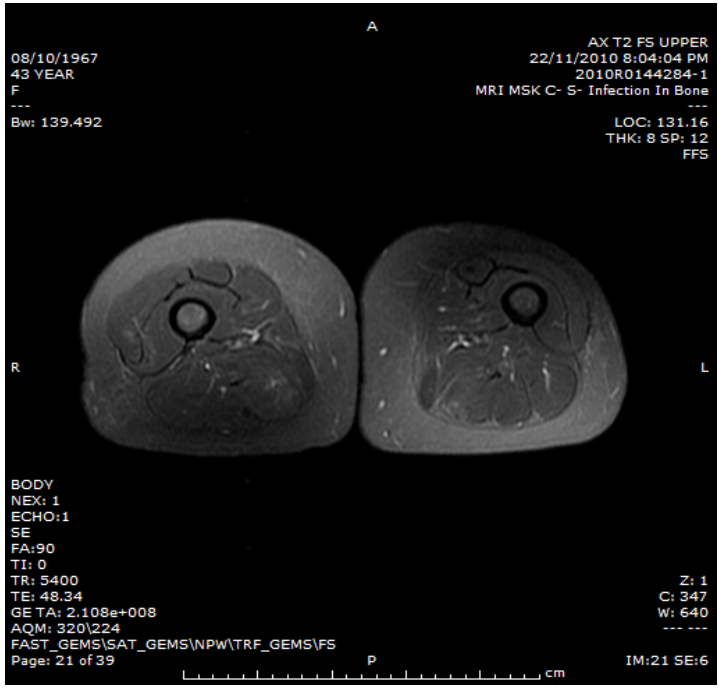
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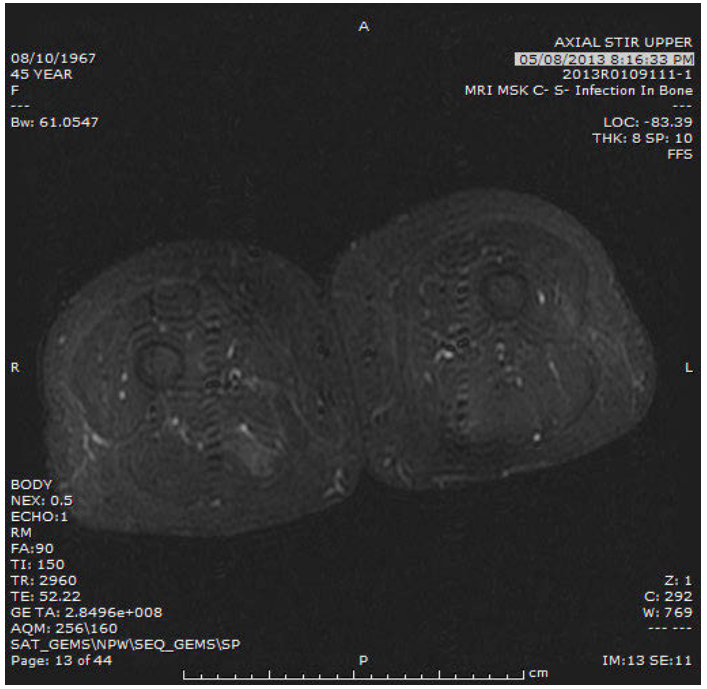
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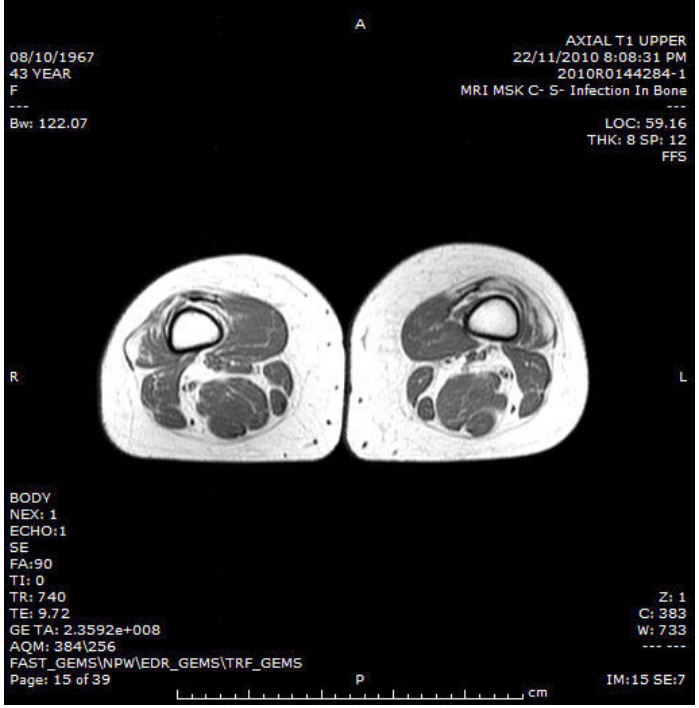
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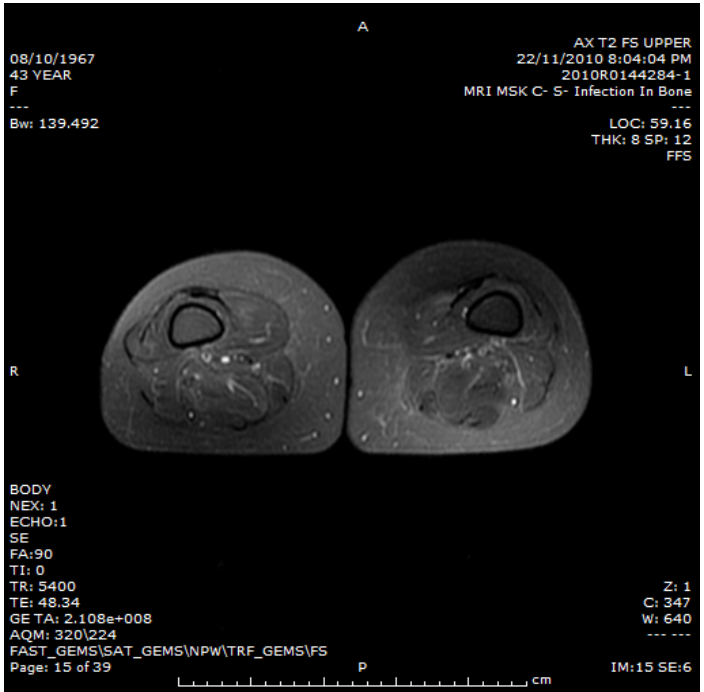
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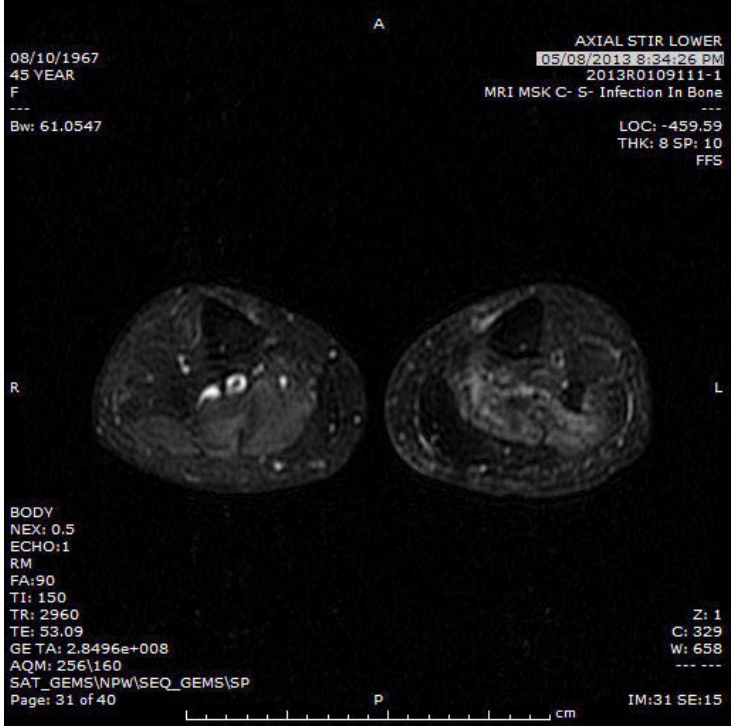
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Page: 31 of 40
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P
IM:31 SE:15
cm

NEXT SPEAKER

Legless

Dr Alexis Selby

Dr Neil Simon

Case

- 18yo right handed female audio engineering student

- 3 months of symptoms
  - Intermittent, increasing right arm, forearm and shoulder pain
  - Associated with numbness of right fourth and fifth digits
  - Woken at night by symptoms
  - Not provoked by position, although shoulder abduction painful at times

- Previous right shoulder dislocation age 12
- Mild intermittent shoulder pain since childhood years

Examination

- Motor examination
  - Normal muscle bulk
  - Normal muscle strength including finger abduction
  - Reflexes 2+ and symmetrical

- Sensation
  - Altered sensation over fifth digit but normal in palm
  - Normal sensation elsewhere in both upper limbs
  - Tinel's phenomenon with percussion of the ulnar nerve at elbow
  - Spurling's test negative

Differential diagnoses?

Differential diagnoses?

- Ulnar nerve entrapment
- Cervical radiculopathy
- Lower trunk brachial plexopathy

Investigations - NCS

- Motor NCS

Nerve / Sites	Latency ms	Amplitude mV	Dur. ms	Velocity m/s	Dist. cm
R MEDIAN					
Wrist	3.8	13.5	7.2		8
Elbow	7.7	13.5	7.2	61.4	24
Axilla	9.7	11.8	7.3	60.0	12
R ULNAR ADM					
Wrist	2.7	11.2	6.5		8
B. Elbow	6.0	10.8	6.7	64.0	21
A. Elbow	7.5	10.6	6.8	66.2	10
Axilla	9.1	9.8	7.0	62.5	10

- Right ulnar nerve inching studies normal

Investigations - NCS

- F waves (Height 1m 80cm)

Nerve / Sites	%F	F lat. ms
R MEDIAN	100	25.9
R ULNAR	100	27.2

Investigations - NCS

- Sensory NCS

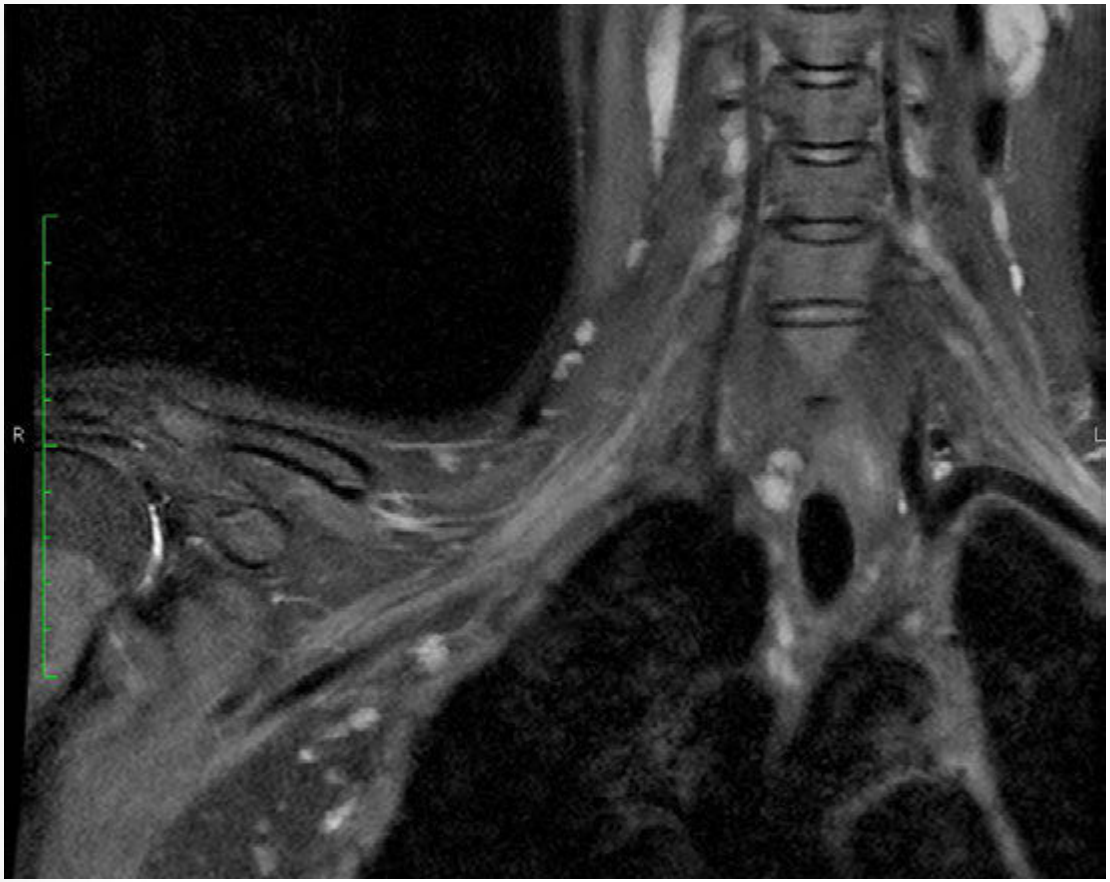
Nerve / Sites	Latency ms	Pk Amp. µV	Distance cm	Velocity m/s
R MEDIAN – Dig II				
Wrist	2.6	78.9	14	53.8
R ULNAR – Dig V				
Wrist	2.5	30.3	14	56.0
L ULNAR – Dig V				
Wrist	2.7	53.5	14	52.7
R RADIAL – Median – Thumb				
Forearm (Radial)	2.4	23.7	10	41.7
Wrist (Median)	2.6	19.1	10	38.4
R MEDIAL AB CUT				
Forearm	2.0	9.7	12	60.6
L MEDIAL AB CUT				
Forearm	1.8	5.1	12	65.8

Investigations - EMG

- EMG

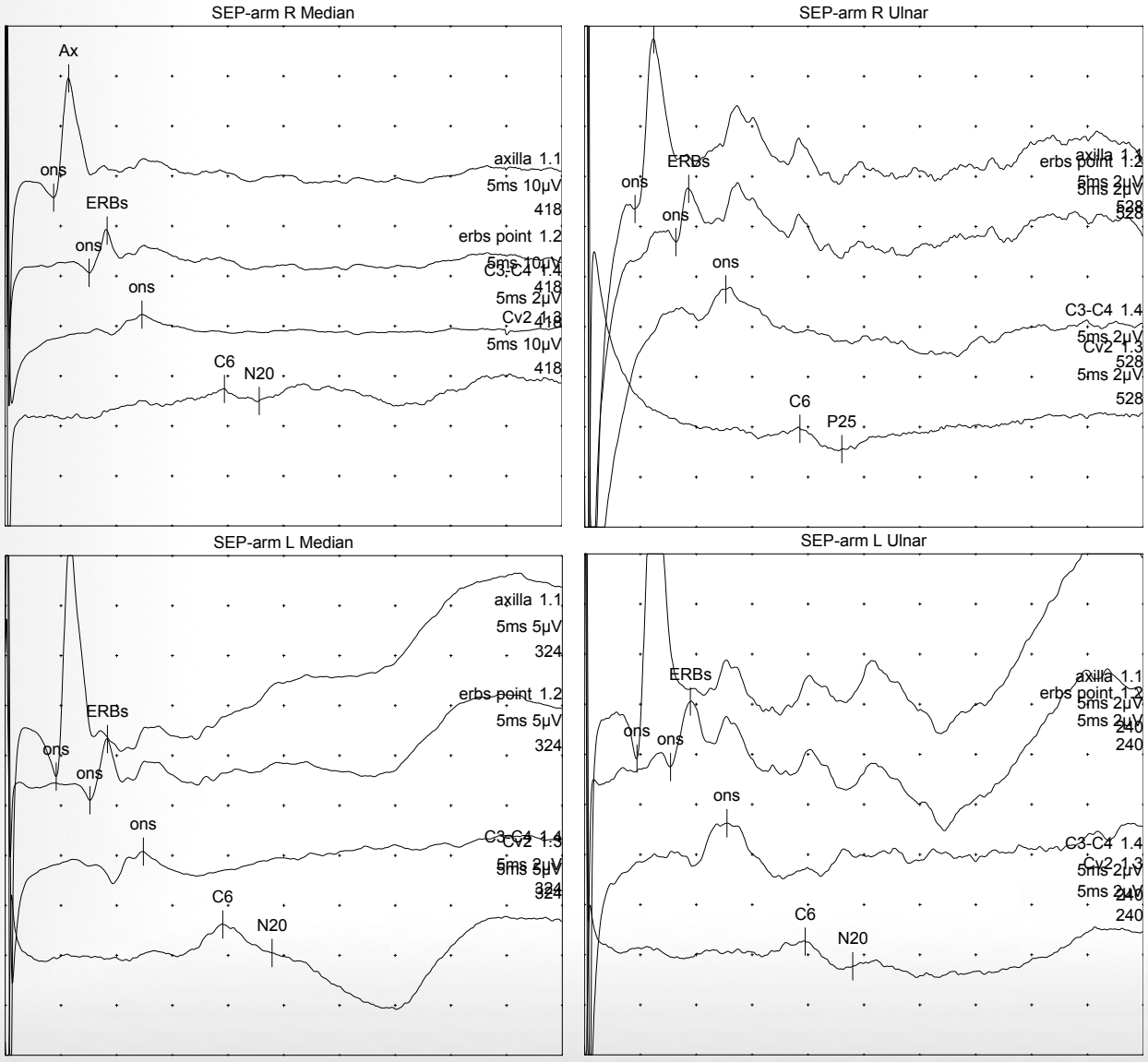
EMG Summary			
	Spontaneous	MUAP	Recruitment
R ABP	No spontaneous activity	MU within normal limits	Recruitment normal
R FDI	No spontaneous activity	MU within normal limits	Recruitment normal

Investigations - MRI



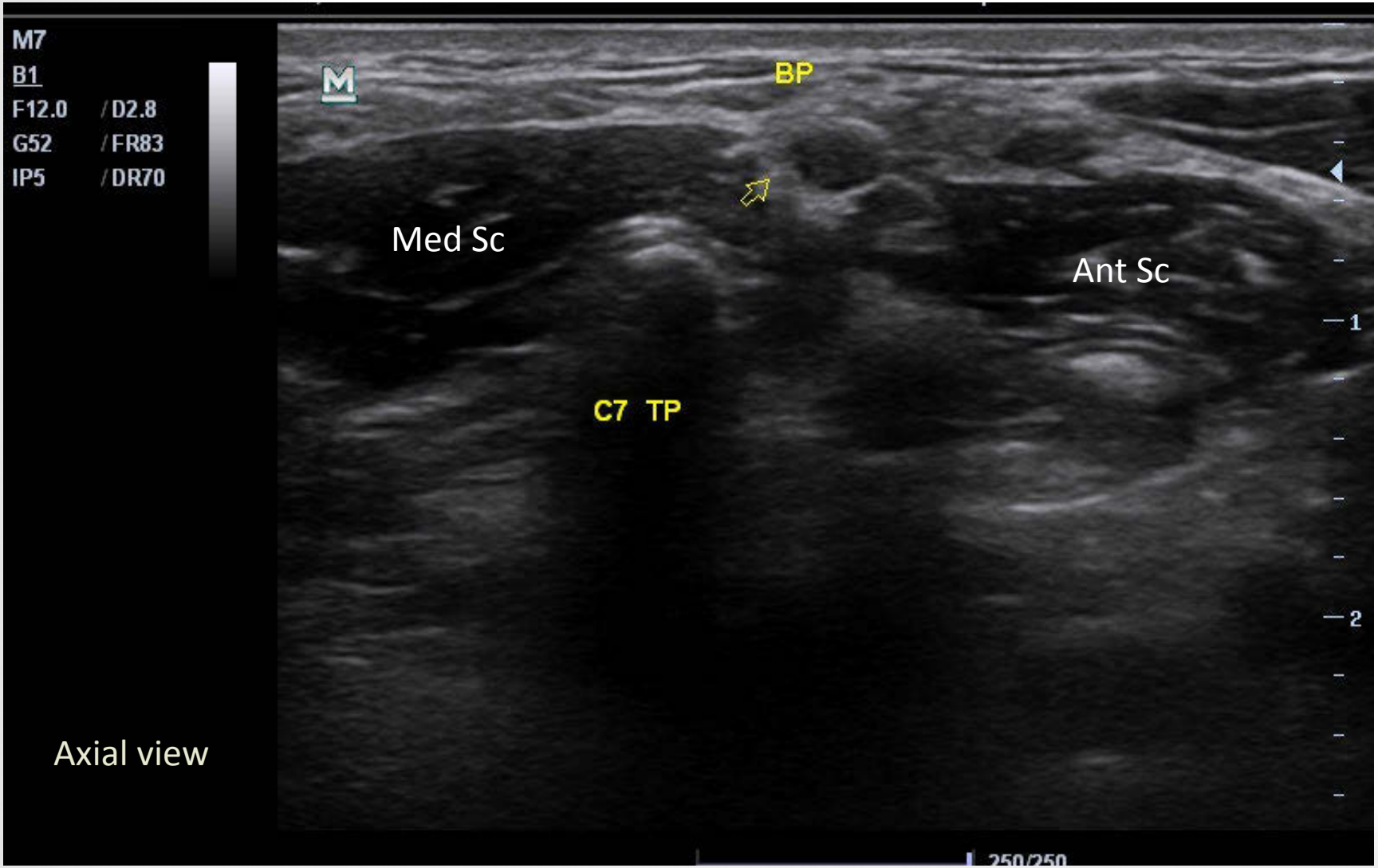
- No evidence of thoracic outlet syndrome

Median and ulnar SSEPs - Normal



Next?

Ultrasound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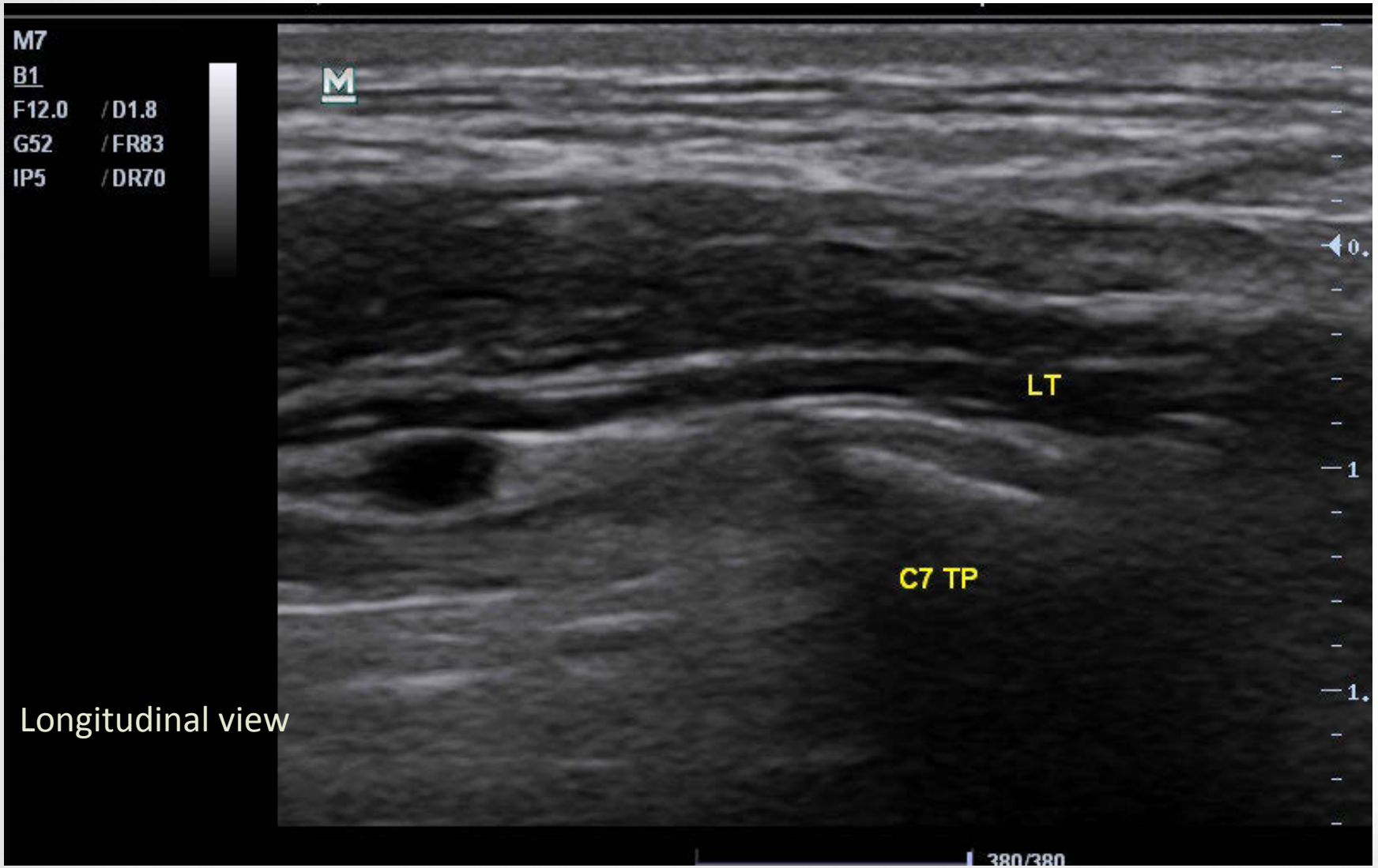
F12.0 / D2.8
G52 / FR83
IP5 / DR70
M
BP
Med Sc
Ant Sc
C7 TP
1
2
250/250
Axial view

Ultrasound

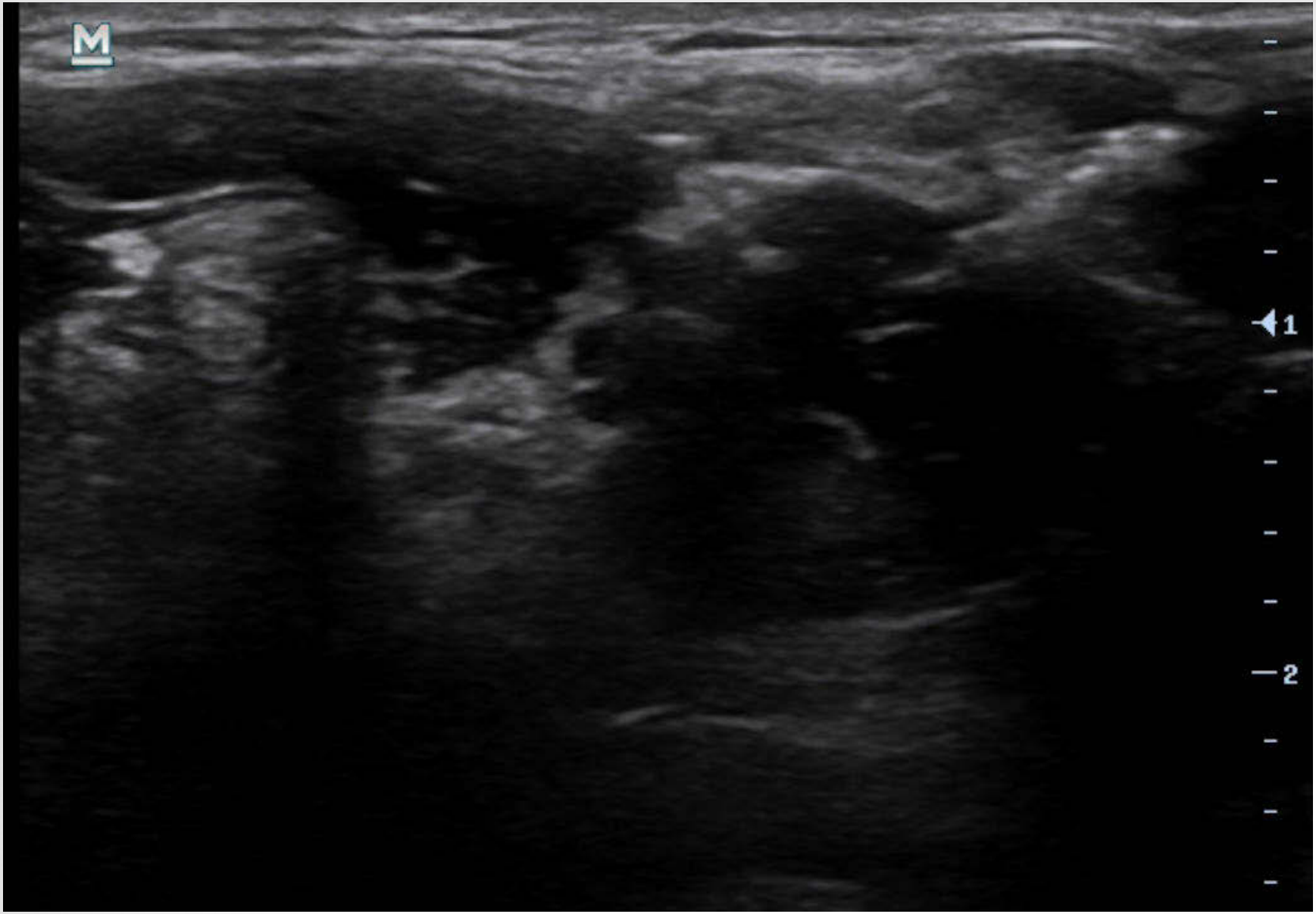


Longitudinal view

Progress

- Ultrasound demonstrated elongated C7 transverse process contacting lower trunk

- 30 units botulinum toxin injected into right anterior scalene muscle
  - Aim to increase size of interscalene triangle
  - Some benefit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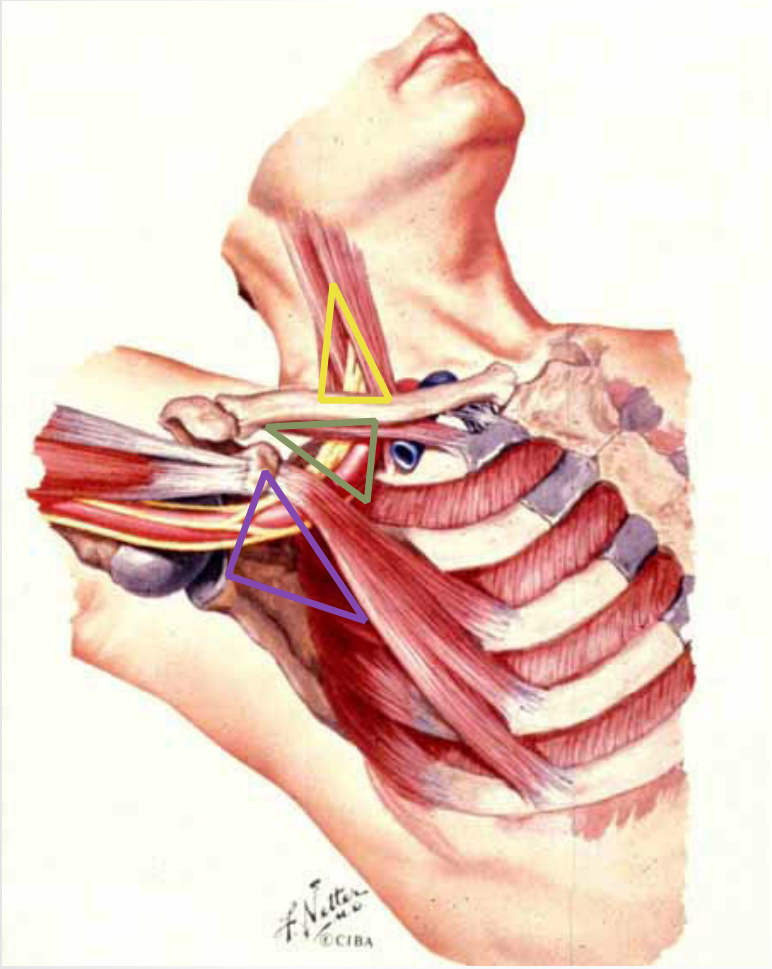
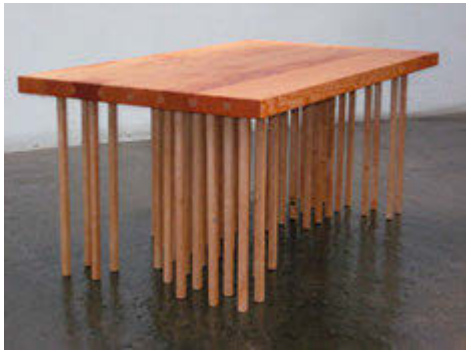
Neurogenic thoracic outlet syndrome



- Compression of brachial plexus elements
  - Bony or soft tissue anomaly

“A table needs at least 3 legs…”

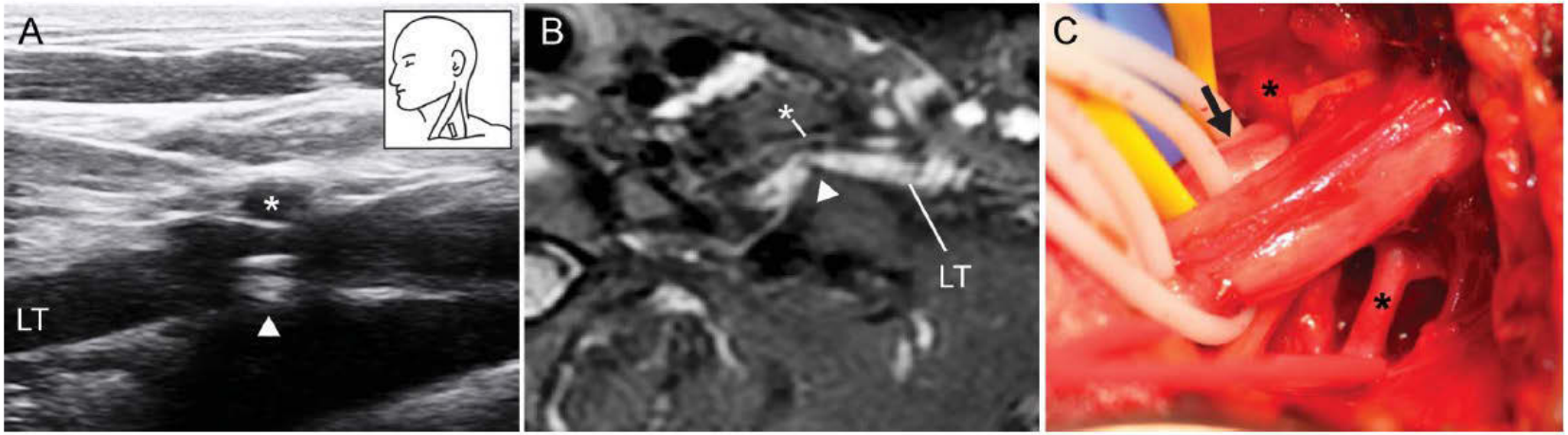
Neurogenic thoracic outlet syndrome



- **‘4 LEGS’ OF THE DIAGNOSIS OF NEUROGENIC TOS**

1. HISTORY
   - Shoulder pain, medial hand and forearm tingling
2. EXAMINATION
   - C8/T1 sensory abnormalities
   - Thenar > intrinsic hand muscle wasting (Gilliatt-Sumner hand)
3. ELECTRODIAGNOSTIC STUDIES
   - Reduced ulnar SNAP, normal median SNAP
   - Reduction median > ulnar CMAP amplitude
   - Reduced MAC SNAP
   - NOTE – ABOVE ONLY PRESENT IF AXONAL LOSS HAS OCCURRED
4. IMAGING
   - Bony or soft tissue abnormality

Ultrasound
– A new leg in the diagnosis of TOS



Correlation between ultrasound (A) and MRI (B) demonstrates compression of the lower trunk (LT) between an artery (*) and fibrous band (arrowhead) arising from an elongated C7 transverse process. At operation (C), the artery (*) passed between the middle and lower trunks. Fibrous bands were resected to release the LT (arrow).

Sonographic diagnosis of true neurogenic thoracic outlet syndrome

Neil G. Simon, Jeffrey W. Ralph, Cynthia Chin, et al.
Neurology 2013;81;1965

Additional diagnostic tools

1. Response to anterior scalene botulinum toxin injection
   - Relevant if anterior scalene abnormality eg hypertrophy
   - Predictor of response to surgery
     - Some surgeons see response as necessary
   - Ultrasound guided

2. Somatosensory evoked potentials
   - Ulnar nerve SSEPs abnormal only if abnormal NCS/examination
   - Normal examination/NCS = normal SSEPs
   - Adjunctive evidence

Somatosensory evoked responses in the diagnosis of thoracic outlet syndrome

C YIANNIKAS, JC WALSH

From the Department of Clinical Neurophysiology, Royal Prince Alfred Hospital, Sydney, Australia

Neurogenic thoracic outlet syndrome

- Are all ‘legs’ required for a diagnosis of neurogenic TOS?
- If not, how much support is required to make the diagnosis?
- Disputed TOS – neurogenic TOS spectrum?
- Is TOS underdiagnosed?

The Thoracic Outlet Syndrome Is Overdiagnosed

Asa J. Wilbourn, MD

References

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