

A 67-year-old woman presenting
with weakness of both lower
extremities for 1-2 years

가톨릭의대
장대현

F/67

Chief complaint

Gait disturbance with progressive bilateral
motor weakness

(onset : 2021.01)

Present illness

2021.01 경부터 양하지 위약감 지속되며 나타난 보행이상으로

2022.08.03 외부병원에서 실시한 전기진단 검사에서 motor sensory neuropathy 소견 확인되어 Further study 위하여 본원 내원함.

- Weakness는 좌측 하지에서 먼저 시작되었으며 이후 우측 하지, 우측 상지 순서로 진행.
- 감각저하는 비슷한 시기 우측 하지 부터 시작

History

DM/HBP/TBc/Hepatitis (-/-/-/-)

Op Hx (-)

CVA Hx (-)

Fx Hx (-)

Medication Hx (-)

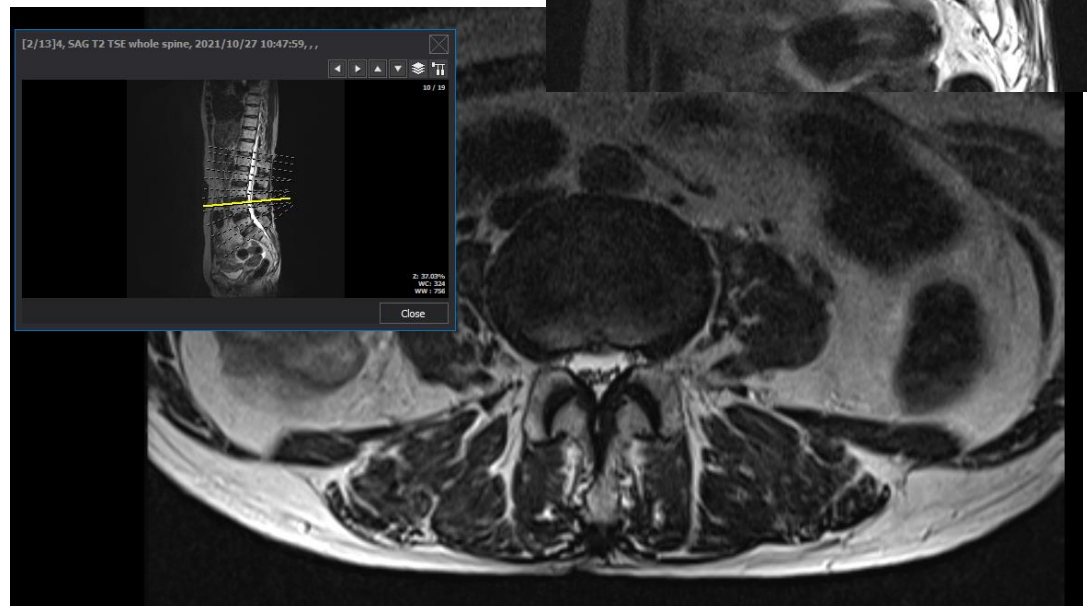
Smoking : denial

Drinking : denial

@ 2021.11.14 건강검진

EGD : gastritis

- 2021.11
 - SNRB, L3,4, bilat



Physical examination

(2022.09.20, onset 1.5YA)

Motor function

U/Ex

Shoulder flexion G/G

Elbow flexion G/G

Elbow extension G/G

Wrist flexion **F+**/G

Wrist extension **F**/G

Finger grasp **F+**/G

L/Ex

Hip flexion G/G

Knee extension G/G

Ankle dorsiflexion **P/P-**

Toe extension **P/P-**

Ankle plantar flexion **P/P-**

Sensory function

Pin-prick, light touch : **Lower extremity diffuse hypoesthesia (Rt. > Lt.) (esp. sole)**

Reflex

BJ(**+/+**) TJ(**+/+**) KJ(**+/+**) AJ(**+/+**)

Pathologic reflex

: Babinski(**-/-**) Ankle clonus(**-/-**) Hoffman's sign(**-/-**)

Muscle atrophy : peroneal muscles, gastrocnemius, intrinsic foot and hand muscles

@ EMG, [REDACTED], 2022.04.27 - [REDACTED]

SNAP : NR

CMAP: NR in tibial, peroneal nn.

decreased amplitudes in median&ulnar nn. , delayed latencies in median n.

F-wave : NR

H-reflex : NR

needle EMG: ASA in Rt FDI, APB, TA, GCM // recruitment-reduced.

G3654 (EC0157)

PMP22 gene duplication/deletion

진 단 명 : Charcot-Marie-Tooth disease

유 전 자 : PMP22 on 17p12.

유전방식 : 상염색체우성유전

검 체 : DNA isolated from Peripheral blood

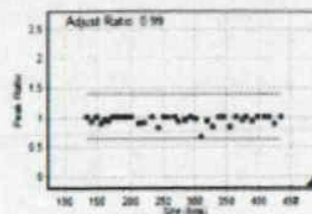
방 법 : MLPA

OMIM : #118220 for disease
★601097 for gene

GenBank accession number :
NG_007949.1
NM_000304.4

검사결과 : No duplicant/deletion of PMP22 gene was detected.

MLPA analysis for PMP22 (MRC-Holland P033-B4 probe set)



] All PMP22 spots located in normal range.

EMG & NCS 2022.08.03 (2nd 외/부병원)

Sensory Nerve Conduction:

Nerve and Site	Peak Latency	Amp.	Segment	Latency Difference	Distance	Conduction Velocity
Median.R						
Mid palm	NE ms	NE ms		ms	mm	m/s
Wrist	NE ms	NE ms		ms	mm	m/s
Median.L						
Mid palm	NE ms	NE ms		ms	mm	m/s
Wrist	NE ms	NE ms		ms	mm	m/s
Ulnar.R						
Wrist(Ulnar)	3.7 ms	6 μ V		ms	mm	m/s
Ulnar.L						
Wrist(Ulnar)	3.8 ms	6 μ V		ms	mm	m/s
Sural.R						
Lower leg	NE ms	NE ms		ms	mm	m/s
Sural.L						
Lower leg	NE ms	NE ms		ms	mm	m/s
Superficial peroneal.R						
Lower leg	NE ms	NE ms		ms	mm	m/s
Superficial peroneal.L						
Lower leg	NE ms	NE ms		ms	mm	m/s

EMG & NCS 2022.08.03 (2nd 외/부병원)

Motor Nerve Conduction:

Nerve and Site	Latency	Amp.	Segment	Distance	Conduction Velocity
Median.R					
Wrist	4.6 ms	1.4 mV		mm	m/s
Elbow	9.2 ms	1.1 mV	Wrist-Elbow	205 mm	45 m/s
Median.L					
Wrist	4.2 ms	3.8 mV		mm	m/s
Elbow	8.6 ms	3.8 mV	Wrist-Elbow	215 mm	49 m/s
Ulnar.R					
Wrist	3.6 ms	2.0 mV		mm	m/s
Below elbow	7.5 ms	1.8 mV	Wrist-Below elbow	208 mm	53 m/s
Above elbow	9.4 ms	1.8 mV	Below elbow-Above elbow	100 mm	53 m/s
Ulnar.L					
Wrist	3.2 ms	5.6 mV		mm	m/s
Below elbow	7.2 ms	5.2 mV	Wrist-Below elbow	198 mm	50 m/s
Above elbow	9.2 ms	4.8 mV	Below elbow-Above elbow	100 mm	50 m/s
Tibial.R					
Ankle	6.3 ms	0.1 mV		mm	m/s
Popliteal fossa	17.7 ms	0.1 mV	Ankle-Popliteal fossa	350 mm	31 m/s
Tibial.L					
Ankle	6.2 ms	0.2 mV		mm	m/s
Popliteal fossa	17.9 ms	0.2 mV	Ankle-Popliteal fossa	360 mm	31 m/s
Peroneal.R -EDB					
Ankle	NR ms	NR mV		mm	m/s
Fibula (head)	NR ms	NR mV	Ankle-Fibula (head)	mm	m/s
accessory peroneal n	NR ms	NR mV	Fibula (head)-accessory peroneal nerve	mm	m/s
Peroneal.L -EDB					
Ankle	NR ms	NR mV		mm	m/s
Fibula (head)	NR ms	NR mV	Ankle-Fibula (head)	mm	m/s
accessory peroneal n	NR ms	NR mV	Fibula (head)-accessory peroneal nerve	mm	m/s
Peroneal.R -TA					
Fibula (head)	3.6 ms	0.6 mV	Tibialis anterior-Fibula (head)	mm	m/s
Popliteal fossa	9.2 ms	0.8 mV	Fibula (head)-Popliteal fossa	100 mm	17 m/s
Peroneal.L -TA					
Fibula (head)	3.7 ms	0.3 mV	Tibialis anterior-Fibula (head)	mm	m/s
Popliteal fossa	10.0 ms	0.4 mV	Fibula (head)-Popliteal fossa	100 mm	16 m/s

EMG & NCS 2022.08.03 (2nd 외/부병원)

Motor Nerve Conduction:

Nerve and Site		Latency	Amp.	Segment
Median.R				
Wrist	4.6 ms	1.4 mV	Wrist-Elbow	
Elbow	9.2 ms	1.1 mV		
Median.L				
Wrist	4.2 ms	3.8 mV	Wrist-Elbow	
Elbow	8.6 ms	3.8 mV		
Ulnar.R				
Wrist	3.6 ms	2.0 mV	Wrist-Below elbow	
Below elbow	7.5 ms	1.8 mV		
Above elbow	9.4 ms	1.8 mV	Below elbow-Above elbow	
Ulnar.L				
Wrist	3.2 ms	5.6 mV	Wrist-Below elbow	
Below elbow	7.2 ms	5.2 mV		
Above elbow	9.2 ms	4.8 mV	Below elbow-Above elbow	
Tibial.R				
Ankle	6.3 ms	0.1 mV	Ankle-Popliteal fossa	
Popliteal fossa	17.7 ms	0.1 mV		
Tibial.L				
Ankle	6.2 ms	0.2 mV		

EMG & NCS 2022.08.03 (2nd 외/부병원)

F-Wave Studies

Nerve	M-Latency	F-Latency
Median.R	4.7	NR
Median.L	8.2	NR
Tibial.R	7.3	NR
Tibial.L	6.5	NR

H-reflex:

Nerve	Latency	Amplitude (max)
Tibial.R		
M-wave:	7.3 ms	0.7 mV
H-wave:	NE	
Tibial.L		
M-wave:	6.9 ms	0.4 mV
H-wave:	NE	

EMG & NCS 2022.08.03 (2nd 외/부병원)

Needle EMG Examination:

Muscle	Insertional	Spontaneous and/or Volitional Activity							Maximum Volitional Activity		
		Fibs	+Wave	Fasc	Duration (ms)	Amplitude (uV)	Poly	Recruitment	Amplitude (mV)	Pattern	Effort
Masseter.R	Normal	None	None	None	Normal	Normal	None	Normal		Full	
Sternocleidomastoid.R	Normal	None	None	None	Normal	Normal	None	Normal		Full	
Trapezius (upper).R	Normal	None	brief runs	None	Normal	Normal	None	Normal		Full	
Trapezius (upper).L	Normal	None	None	None	Normal	Normal	None				
C5-C6 paraspinal.B	Normal	None	None	None	Normal	Normal	None				
C6-C7 paraspinal.B	Normal	None	None	None	Normal	Normal	None				
C7-T1 paraspinal.B	Normal	None	None	None	Normal	Normal	None	Normal		Full	
Biceps brachii.R	Normal	None	brief runs	None	Normal	Normal	None	Normal		Full	
Flexor carpi radialis.R	Normal	None	brief runs	None	Normal	Normal	None	Normal		Full	
1st dorsal interosseous.R	Normal	None	1+	None	Normal	Normal	None	Reduced		Reduced	
Abductor pollicis brevis.R	Normal	None	1+	None	Normal	Normal	None	Reduced		Reduced	
Biceps brachii.L	Normal	None	None	None	Normal	Normal	None	Normal		Full	
Flexor carpi radialis.L	Normal	None	None	None	Normal	Normal	None	Normal		Full	
1st dorsal interosseous.L	Normal	None	1+	None	Normal	Normal	None	Reduced		Reduced	
Abductor pollicis brevis.L	Normal	None	1+	None	Normal	Normal	None	Reduced		Reduced	
Upper thoracic.B	Normal	None	None	None	Normal	Normal	None				
Mid thoracic.B	Normal	None	None	None	Normal	Normal	None				
Lower thoracic.R	Normal	None	None	None	Normal	Normal	None				
Lower thoracic.L	Normal	None	1+	None	Normal	Normal	None				
L1-L2 paraspinal.R	Normal	None	1+	None	Normal	Normal	None				
L2-L3 paraspinal.R	Normal	None	2+	None	Normal	Normal	None				
L3-L4 paraspinal.R	Normal	None	None	None	Normal	Normal	None				
L4-L5 paraspinal.R	Normal	None	None	None	Normal	Normal	None				
L5-S1 paraspinal.R	Normal	None	3+	None	Normal	Normal	None				
L1-L2 paraspinal.L	Normal	None	None	None	Normal	Normal	None				
L2-L3 paraspinal.L	Normal	None	None	None	Normal	Normal	None				
L3-L4 paraspinal.L	Normal	None	1+	None	Normal	Normal	None				
L4-L5 paraspinal.L	CRD	None	1+	None	Normal	Normal	None				
L5-S1 paraspinal.L	Normal	None	None	None	Normal	Normal	None				
Gluteus maximus.R	Normal	None	brief runs	None	Normal	Normal	Poly	Normal		Reduced	
Gluteus medius.R	Normal	None	None	None	Normal	Normal	None	Normal		Reduced	
Vastus medialis.R	Normal	None	brief runs	None	Normal	Normal	None	Reduced		Reduced	
Tibialis anterior.R	Normal	None	2+	None	Normal	Normal	None	Reduced		Discrete	
Peroneus longus.R	Normal	None	1+	None	Normal	Normal	None	Reduced		Discrete	
Gastrocnemius.R	Normal	1+	1+	None	Normal	Normal	None	Reduced		Discrete	
Gluteus maximus.L	Normal	None	None	None	Normal	Normal	Poly	Normal		Reduced	
Gluteus medius.L	Normal	None	None	None	Normal	Normal	None	Normal		Reduced	
Vastus medialis.L	Normal	None	None	None	Normal	Normal	None	Reduced		Reduced	
Tibialis anterior.L	Normal	None	1+	None	Normal	Normal	None	Reduced		Discrete	
Peroneus longus.L	Normal	None	1+	None	Normal	Normal	None	Reduced		Discrete	
Gastrocnemius.L	Normal	None	1+	None	Normal	Normal	None	Reduced		Discrete	

외부병원 전기진단 검사 (2022.8.3)

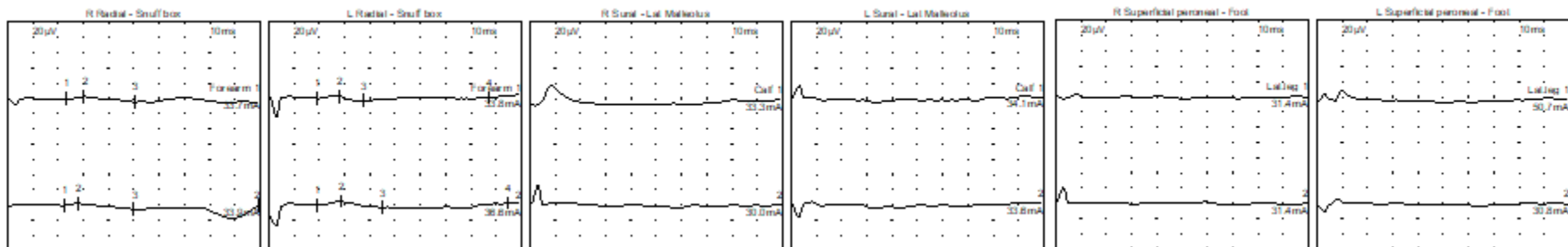
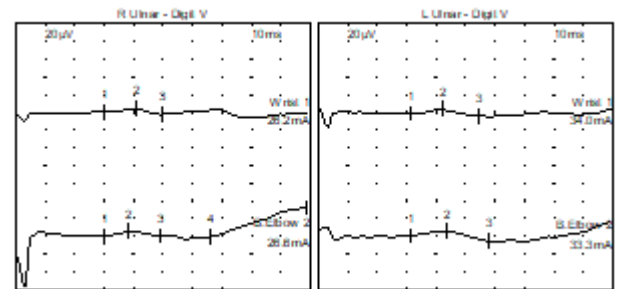
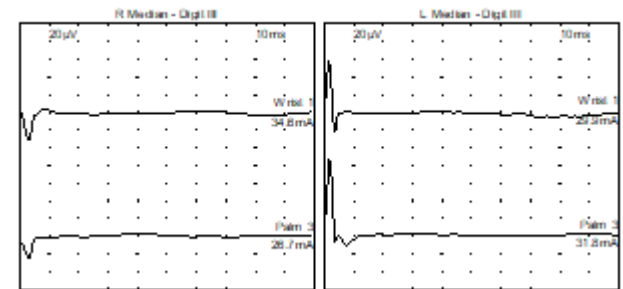
- Distal bilateral (a little asymmetric) motor and sensory polyneuropathy, severe axonal, upper and lower extremity involvement

Nerve conduction studies & Needle EMG

2022.09.28

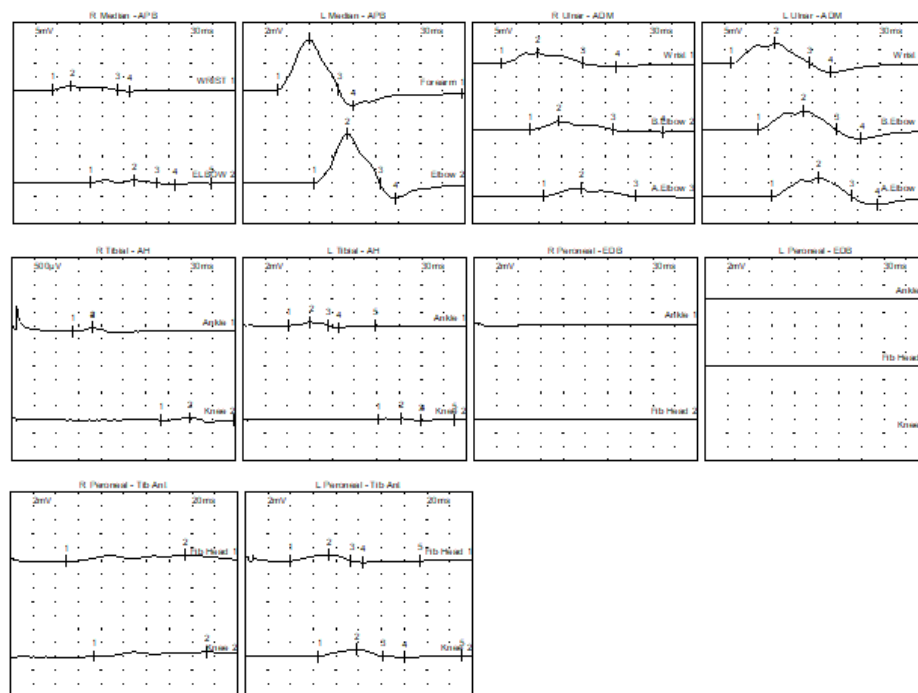
Sensory NCS

Nerve / Sites	Onset ms	Peak.Lat ms	Amp.1-2 μ V	Dist. cm	Vel. Pk m/s
R Median - Digit III					
Wrist	NR	NR	NR		
Palm	NR	NR	NR		
L Median - Digit III					
Wrist	NR	NR	NR		
Palm	NR	NR	NR		
R Ulnar - Digit V					
Wrist	2.97	3.96	4.9	14	35.4
L Ulnar - Digit V					
Wrist	3.13	4.02	5.4	14	34.8
R Radial - Snuff box					
Forearm	2.24	2.81	2.6	10	35.6
L Radial - Snuff box					
Forearm	1.88	2.76	3.4	10	36.2
R Sural - Lat Malleolus					
Calf	NR	NR	NR		
L Sural - Lat Malleolus					
Calf	NR	NR	NR		
R Superficial peroneal - Foot					
Lat.leg	NR	NR	NR		
L Superficial peroneal - Foot					
Lat.leg	NR	NR	NR		



Motor NCS

Nerve / Sites	onset ms	Amp.1-2 mV	Dist. cm	Vel. m/s	Pk-Dur. ms
R Median - APB					
WRIST	4.86	1.5			8.80
ELBOW	9.86	1.0	24	48.0	8.91
L Median - APB					
Forearm	4.69	7.6			8.28
Elbow	9.53	7.4	23	47.5	8.96
R Ulnar - ADM					
Wrist	3.80	3.8			11.09
B.Elbow	7.71	3.2	18	46.1	11.30
A.Elbow	9.58	3.0	10	53.3	12.50
L Ulnar - ADM					
Wrist	3.96	7.5			10.52
B.Elbow	7.60	7.1	17.5	48.0	10.57
A.Elbow	9.43	7.0	10	54.9	10.78
R Tibial - AH					
Ankle	8.13	0.2			
Knee	19.95	0.1	35	29.6	
L Tibial - AH					
Ankle	6.09	0.5			5.36
Knee	18.13	0.2	35	29.1	5.78
R Peroneal - EDB					
Ankle	NR	NR			NR
Fib Head	NR	NR			NR
L Peroneal - EDB					
Ankle	NR	NR			NR
Fib Head	NR	NR			NR
R Peroneal - Tib Ant					
Fib Head	4.90	0.9			
Knee	7.34	0.7	7	28.6	
L Peroneal - Tib Ant					
Fib Head	3.96	0.9			5.36
Knee	6.46	0.9	7	28.0	5.63



F Wave

Nerve	Min F Lat ms
R Median - APB	NR
L Median - APB	NR
R Tibial - AH	NR
L Tibial - AH	NR

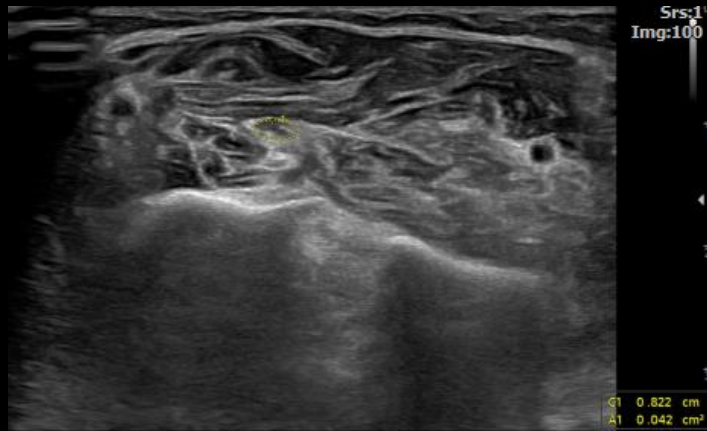
H Reflex

Nerve	H Lat ms
R Tibial - Soleus	NR
L Tibial - Soleus	NR

Motor NCS

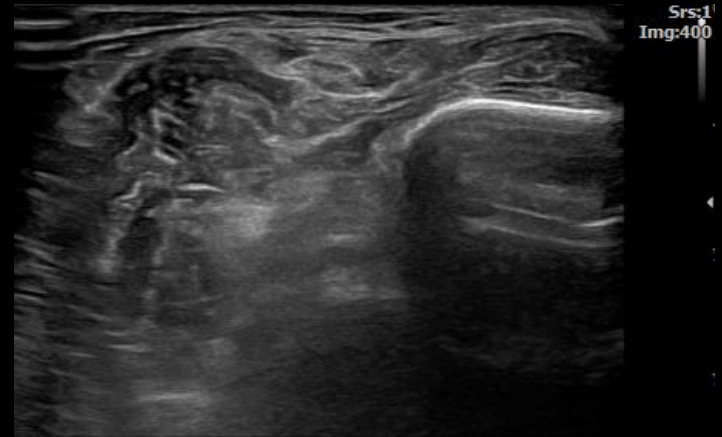
Nerve / Sites	onset ms	Amp.1-2 mV	Dist. cm	Vel. m/s	Pk-Dur. ms
R Median - APB					
WRIST	4.86	1.5			8.80
ELBOW	9.86	1.0	24	48.0	8.91
L Median - APB					
Forearm	4.69	7.6			8.28
Elbow	9.53	7.4	23	47.5	8.96
R Ulnar - ADM					
Wrist	3.80	3.8			11.09
B.Elbow	7.71	3.2	18	46.1	11.30
A.Elbow	9.58	3.0	10	53.3	12.50
L Ulnar - ADM					
Wrist	3.96	7.5			10.52
B.Elbow	7.60	7.1	17.5	48.0	10.57
A.Elbow	9.43	7.0	10	54.9	10.78
R Tibial - AH					
Ankle	8.13	0.2			

Peripheral nerve US (2022.09.28)



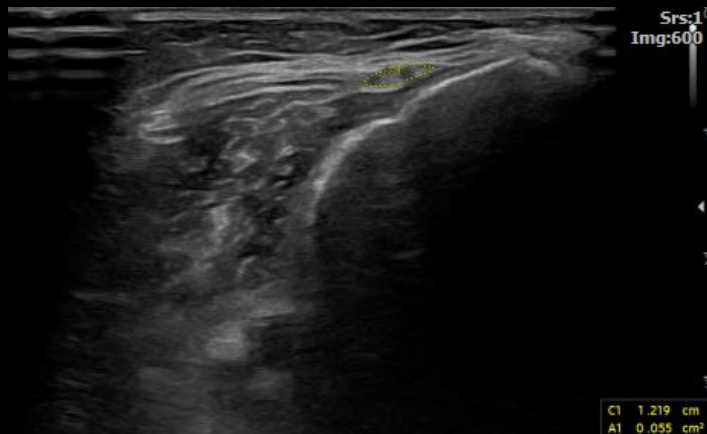
RT MEDIAN N. PROXIMAL

WC: 128
WW: 256
Z: 44.53%



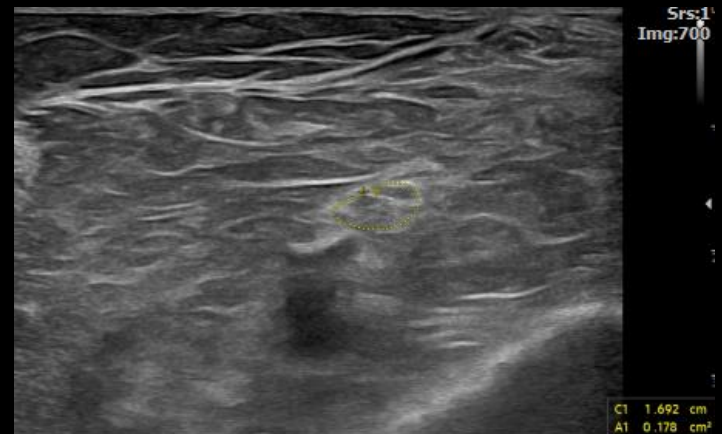
RT ULNAR

WC: 128
WW: 256
Z: 44.53%



LT PERONEAL N.

WC: 128
WW: 256
Z: 44.53%



LT TIBIAL N.

WC: 128
WW: 256
Z: 44.53%

Conclusion

- Distal asymmetric sensorimotor peripheral polyneuropathy or advanced mononeuritis multiplex
- Involving upper and lower extremities, showing axonal loss, clinically which may be due to vasculitis

Comment) Cancer, endocrine, toxin, hereditary, immune-mediated disease 등 다른 polyneuropathy 원인에 대한 감별 필요합니다.

Recommend) vasculitis work up

Needle EMG : 이전 검사 (2022.08.03, 외부병원) 에서 시행하여 추가로 시행하지 않음.

Q and A