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A Case Showing Specific Electrophysiological Findings of Kennedy's Disease.

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INTRODUCTION

Kennedy disease(KD) is an X-linked recessive form of spinal muscular atrophy characterized by adult onset of slowly progressive bulbar and proximal greater than distal limb weakness and atrophy. As KD has symptoms similar to most neuromuscular diseases, but the progression of symptoms is slow and there are few cases of sensory deterioration or neuropathic pain, so it is difficult to make a rapid diagnosis clinically. KD is caused by a mutation in the androgen receptor gene on chromosome Xq11-12. The mutation is characterized by an increased size of polymorphic tandem expanded CAG repeats within the first exon of androgen receptor gene. It can be confirmed through molecular genetic analysis, but it takes more than a month.

CASE REPORT

A 52-year-old male who has been suffering from limb weakness for 10 years visited a secondary hospital. The patient complained of proximal limb muscle weakness. Blood test showed only an increased CPK, and no abnormal findings on imaging tests and no similar family history. Considering his job moving heavy objects, conservative treatment was performed under suspicion of kidney disease, but no improvement. Symptoms had got worse in recent 5 years, exercise endurance had decreased. He was transferred to our rehabilitation department. Physical examination showed increased tendon reflex and decreased proximal muscle strength, but no pathologic reflex. Tongue muscle had atrophy, but no fasciculation. In summary, motor-neuron disease(MND) was suspected. Spine magnetic resonance imaging(MRI) excluded cord lesions and radiculopathy but found degenerative changes. For electrophysiological examination, the motor nerve conduction study showed a decreased latency and amplitude in some nerves, but it was close to the normal compared to the sensory nerve conduction. Sensory nerve conduction studies showed decreased latency and amplitude in most nerves. (Table 1). Needle electromyography(EMG) revealed a widespread reduction in MUAP recruitment with large amplitude and long duration. Fasciculation potentials, diffuse fibrillation potentials and positive sharp waves were detected. (Table 2) Consequently, KD was most suspected. Molecular genetic test showed the number of CAG repeats increased to 46, confirming the KD (Fig. 1).

MNC

Nerve / Sites	Rec. Site	Lat. ms	Amp.1-2 mV	Dur. ms	Area mVms	Dist. mm	d Lat. ms	Vel. m/s
R Median - APB								
Wrist	APB	4.43	10.2	6.93	35.9	80		
Elbow	APB	8.33	9.2	7.19	36.7	222	3.91	56.8
Palm	APB	2.40	11.8	6.93	39.6	90	-2.05	44.3
L Median - APB								
Wrist	APB	4.48	9.8	5.36	27.1	80		
Elbow	APB	8.39	9.6	5.52	25.5	208	3.91	53.2
Palm	APB	2.45	11.0	5.18	30.5	90	-2.05	44.3
R Ulnar - ADM								
Wrist	ADM	3.33	11.8	6.51	40.2	80		
B.Elbow	ADM	8.51	11.9	6.82	42.8	212	3.18	66.7
A.Elbow	ADM	7.97	11.5	6.67	40.6	100	1.46	68.6
							4.64	
L Ulnar - ADM								
Wrist	ADM	3.02	11.0	7.55	50.0	80		
B.Elbow	ADM	8.51	10.3	7.39	45.8	194	3.49	55.6
A.Elbow	ADM	8.28	10.0	6.98	44.7	100	1.77	56.5
							5.26	
R Peroneal - EDB								
Ankle	EDB	4.01	2.2	5.57	7.6	80		
Fib head	EDB	10.42	1.5	5.61	6.7	282	6.41	44.0
L Peroneal - EDB								
Ankle	EDB	4.80	1.8	4.69	5.2	80		
Fib head	EDB	10.94	1.9	4.64	5.1	281	6.04	46.5
R Tibial - AH								
Ankle	AH	3.91	19.8	5.99	49.7	80		
Pop fossa	AH	11.67	15.8	6.46	42.1	362	7.76	45.4
L Tibial - AH								
Ankle	AH	3.70	18.0	5.78	47.2	80		
Pop fossa	AH	11.30	15.2	5.99	40.8	361	7.60	46.2

SNC

Nerve / Sites	Rec. Site	Onset Lat ms	NP Amp µV	Segments	Distance mm	Peak Diff ms	Velocity m/s
R Median - Digit II (Antidromic)							
Wrist	Dig II	3.44	6.1	Wrist - Dig II	140		41
Mid palm	Dig II	1.35	6.8	Mid palm - Wrist	70	-2.08	34
L Median - Digit II (Antidromic)							
Wrist	Dig II	3.49	6.8	Wrist - Dig II	140		40
Mid palm	Dig II	1.41	9.0	Mid palm - Wrist	70	-2.24	34
R Ulnar - Digit V (Antidromic)							
Wrist	Dig V	2.78	7.1	Wrist - Dig V	140		51
L Ulnar - Digit V (Antidromic)							
Wrist	Dig V	2.60	6.3	Wrist - Dig V	140		54
R Sural - Ankle (Calf)							
Calf	Ankle	3.28	5.1	Calf - Ankle	140		43
L Sural - Ankle (Calf)							
Calf	Ankle	3.23	4.8	Calf - Ankle	140		43
R Superficial peroneal - Ankle							
Lat leg	Ankle	3.33	2.5	Lat leg - Ankle	140		42
L Superficial peroneal - Ankle							
Lat leg	Ankle	3.38	3.9	Lat leg - Ankle	140		41

Table 1.
The motor nerve conduction test of the upper and lower extremities showed a decrease in amplitude and velocity in some nerves, but it was close to the normal compared to the sensory nerve conduction test.

EMG

EMG Summary Table										
	Insertional	Spontaneous Activity			Volitional MUAPs				Maximum Volitional Activity		
	Insertional	Fibs	+Wave	Fasc	Duration	Amplitude	Poly	Config	Recruitment	Amplitude	Pattern	Effort
R. Tibialis anterior	Normal	1+	1+	None	Normal	>10K	Some	Normal	Normal	Normal	Reduced	Max.
R. Peroneus longus	Normal	2+	2+	None	Normal	>10K	Some	Normal	Normal	Normal	Reduced	Max.
R. Gastrocnemius (Medial head)	Normal	None	None	None	Normal	>10K	Normal	Normal	Normal	Normal	Reduced	Max.
R. Vastus medialis	Normal	2+	2+	None	Normal	>10K	Some	Normal	Normal	Normal	Reduced	Max.
L. Tibialis anterior	Normal	None	None	None	Normal	>10K	Some	Normal	Normal	Normal	Reduced	Max.
L. Peroneus longus	Normal	None	None	None	Normal	>20K	Some	Normal	Normal	Normal	Reduced	Max.
L. Gastrocnemius (Medial head)	Normal	1+	1+	None	Normal	>20K	Some	Normal	Normal	Normal	Reduced	Max.
L. Vastus medialis	Normal	2+	2+	Few	Normal	>20K	Some	Normal	Normal	Normal	Reduced	Max.
L. Rectus femoris	Normal	None	None	None	Normal	>20K	Some	Normal	Normal	Normal	Reduced	Max.
L. Biceps brachii	Normal	None	None	Many	Normal	>10K	Some	Normal	Normal	Normal	R/C	Max.
L. Deltoid	Normal	2+	2+	None	Normal	>20K	Some	Normal	Normal	Normal	Reduced	Max.

Table 2.
Needle electromyography revealed a wide spread reduction in MUAP recruitment with large amplitude and long duration. Fasciculation potentials, diffuse fibrillation potentials and positive sharp waves were detected in some muscles examined.

Fig 1.
The number of CAG repeats increased to 46, thereby confirming the diagnosis of Kennedy disease.

DISCUSSION

For diagnosing KD, painful muscle biopsy is often performed, and confirmation test, molecular genetic test is expensive and takes a long time. Without strong suspicion, difficult to patients' consent. Unlike other MNDs, SNAPs in KD are reduced in amplitude or unobtainable and CMAPs are characteristically normal. In needle EMG, fasciculation potentials and abnormal spontaneous activity are prominent findings.
Therefore, KD can be sufficiently suspected only through electromyography without performing muscle biopsy or molecular genetic test.