

# Kennedy

## Clinical and Electrodiagnostic Features of Kennedy Disease

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**Backgrounds:** Kennedy disease is a X-linked recessive disease characterized by bulbar symptoms, proximal muscle weakness, and gynecomastia.

**Methods:** We analyzed clinical symptoms and performed electrodiagnostic studies on 6 patients.

**Results:** We found following features: 1) proximal muscle weakness 2) bulbar symptoms, as dysarthria, facial and tongue atrophy 3) hyporeflexia or areflexia 4) fasciculations, predominantly on face, and proximal upper extremities 5) decreased sensory nerve action potentials(SNAPs) 6) chronic neurogenic changes in needle EMG.

**Conclusions:** Kennedy disease is characterized by degenerative process of anterior horn cell and dorsal root ganglion without upper motor neuron dysfunction. Increased triple nucleotide CAG repeats(>38) in androgen receptor gene of Xp21 will confirm early stage of this disease.

**Key Words:** Kennedy disease, Triple nucleotide repeats

Kennedy disease is a X-linked recessive disease characterized by bulbar symptoms, proximal muscle weakness, and gynecomastia. We analyzed clinical symptoms and performed electrodiagnostic studies on 6 patients. We found following features: 1) proximal muscle weakness 2) bulbar symptoms, as dysarthria, facial and tongue atrophy 3) hyporeflexia or areflexia 4) fasciculations, predominantly on face, and proximal upper extremities 5) decreased sensory nerve action potentials(SNAPs) 6) chronic neurogenic changes in needle EMG. Kennedy disease is characterized by degenerative process of anterior horn cell and dorsal root ganglion without upper motor neuron dysfunction. Increased triple nucleotide CAG repeats(>38) in androgen receptor gene of Xp21 will confirm early stage of this disease.

**Key Words:** Kennedy disease, Triple nucleotide repeats

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**Table 1.** Clinical features of Kennedy 's disease and CAG repeat number

Patients	Kj	Yj	Nc	Kd	Sy	Li
Onset of Sx (Age,duration) year	73(3)	67(1)	53(3)	48(5)	47(3)	40(?)
muscle cramps	Y	Y	Y	Y	Y	Y
fasciculation(F/T/   )	Y/Y/Y	Y/Y/Y	Y/Y/Y	Y/Y/Y	Y/Y/Y	Y/Y/Y
gynecomastia	N	N	Y	Y	Y	Y
DM	N	Y	N	N	N	N
bulbar weakness	Y	Y	Y	Y	Y	Y
Weakness (proximal limb; U/L)	U>L	N	L=U	U>L	L>U	U=L
weakness (distal limbs; U/L)	N	L	N	N	U=L	N
Reflexes	D	A	D	A	A	D
Hand tremor	N	Y	N	Y	N	Y
Sensory Sx	N	Y	N	Y	N	Y
Family Hx	?	?	?	Y	N	Y
CPK	304	ND	ND	ND	ND	ND
CAG repeat length	40~43	40~50	43	45~50	45~50	40~45

D,decreased; A,absent;CPK,creatinine phosphokinase;ND,not done

2

2) androgen receptor CAG (Li), 2 (Yj) Kd, 3

Androgen receptor PCR (Yj) CAG, 1 Nc, Kd, Sy, Li, 1

CAG, PCR 가 40~50 Kennedy 가

282bp CAG 21 가 6 4

, 38 66 Kennedy 가

Androgen receptor 5 1 ( Nc) 가 가

6 (compound muscle action potentials, CMAP)

1 ( Li) 가 (posi- tive sharp wave) (fibrillation), (polyphasic potentials), (giant motor unit action potential, giant MUP)가 6

, 3 5 (Table 2).

( Kj, Nc, Li)

**Table 2.** Electrophysiologic findings of Kennedy disease

patients	Kj		Yj		Nc		Kd		Sy		Li	
	M	S	M	S	M	S	M	S	M	S	M	S
R/L	R	R	R	R	R	R	R	R	R	R	R	R
	L	L	L	L	L	L	L	L	L	L	L	L
Median												
TL	4.1		3.9		4.7		3.5		3.8		4.38	
	5.4		3.8		3.8		3.8		3.9		5.10	
Amp									6.9			
Wrist	6.9	0.02	8.1	0.002	8.4	0.000	5.8	0.007	11.	0.014	12	0.002
	0.9	0.003	9.6	0.004	11	8NP	5.4	NP	8	0.009	11	0.001
Elbow	6.9	0.02	7.8	0.009	8.2	0.005	5.1	0.01	6.6	0.021	12	0.015
	0.8	0.006	8.8	0.004	10	0.007	4.5	0.003	10	0.034	10	0.007
NCV												
Wrist		44		40		40		46		42		47
		34		40		NP		NP		44		39
Elbow	60		55		57		58		59		50	
	54		54		55		57		51		56	
Ulnar												
TL	3.0		2.9		3.4		3.1		3.0		2.70	
	3.0		2.9		2.8		3.0		3.8		3.00	
Amp							10.					
Wrist	13	0.009	13	0.002	14	0.002	3	0.006	9.7	0.007	12	0.006
	15	0.006	12	0.004	18	0.003	8.6	0.001	6.4	0.002	12	0.002
BE	13	0.016	11	0.007	12	0.008	9.4	0.004	9.5	0.012	11	0.015
	13	0.02	11	0.01	16	0.008	8.0	0.002	6.0	0.006	12	0.006
NCV												
Wrist		45		32		41		42		41		41
		33		32		41		35		36		36
BE	50		59		56		58		57		56	
	54		59		53		52		53		50	
Peroneal												
TL	4.8		4.4		10		5.8		4.9		5.0	
	4.5		3.9		4.7		5.0		3.8		4.3	
Amp												
Ankle	7.9	NP	3.2	0.004	0.4	NP	0.9	NP	2.7	0.003	7.8	NP
	6.4	0.003	4.2	NP	2.4	NP	1.0	NP	2.0	NP	3.7	NP
Fibular	7.2		2.6		0.3		0.9		2.3		7.3	
	5.3		3.6		2.2		1.0		1.7		3.2	
NCV												
Ankle	43	NP	43	41	26	NP	42	NP	40	34	41	NP
	42	37	42	NP	40	NP	40	NP	45	NP	41	NP
Tibial												
TL	3.8		373		5.1		4.0		3.8/		4.9	
	4.1		50		4.8		4.1		3.8		5.5	
Amp												
Ankle	25		15		3.3		12.4		20		13	
	3		21		13		9.1		16		13	
Popliteal	15		375		1.8		11.5		12		10	
	14		72		10		7.4		14		11	
NCV	48		42		38		47		48		43	
	47		41		40		45		41		48	
Sural												
Amp		0.008		0.001		NP		NP		0.001		0.01
		0.006		0.0007		NP		0.004		0.002		0.01
NCV		49		33		NP		NP		40		32
		56		31		NP		43		39		30

R, right ; L,left ; M, motor ; S,sensory ; Amp, amplitude(mV); NCV, nerve conduction velocity(m/s); TL,terminal latency(ms); BE,below elbow; NP, no potential

6  
Kennedy 가 , ALS<sup>10</sup>  
(bulbar palsy) Matthew  
<sup>2</sup> Matthew 10 20 kj  
(amyotrophic lateral sclerosis,  
ALS) , ALS  
, Kennedy  
<sup>7</sup>  
(autosomal dominant bulbar spinal  
muscular atrophy) 40  
Kennedy<sup>8,9</sup>  
Kennedy 10 20% 가<sup>3</sup>  
1 가  
가  
(progressive muscular  
atrophy) 가  
(myelitis) 6  
가  
가  
Mark Wilbourn<sup>8,10</sup>  
가  
가  
6 3  
Kennedy 가  
<sup>11</sup> Mei<sup>12</sup>  
가  
가  
<sup>10</sup>  
syndrome) (postpolio  
Kenney  
(anterior horn cell)  
, ALS  
, ALS  
<sup>8</sup> Kennedy  
(neurogenic) 가

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