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Kennedy's disease with chronic low back pain and muscle weakness : A case report

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INTRODUCTION

Kennedy's disease (KD) is an x-linked recessive spinal muscular atrophy that is characterized by slowly progressing bulbar and proximal atrophy, as opposed to distal, and limb weakness. It presents with symptoms similar to most neuromuscular disorders, but early clinical diagnosis is challenging due to its slow progression and rarity of cases involving sensory loss or neuropathic pain. Moreover, it is characterized by clinical features, such as gynecomastia, testicular atrophy, erectile dysfunction, and diabetes mellitus. This disease has an x-linked recessive inheritance pattern and is thought to be caused by a mutation in the androgen receptor gene, which results in the abnormal aggregation of the androgen receptor and dysfunction of the cytoskeletal system within nerve cells due to impaired tubulin regulation. Although the disease can be confirmed through molecular genetic analysis, the turnaround time is more than a month. The most common symptom of KD is muscle spasms, followed by lower limb muscle weakness, gynecomastia, and upper limb muscle weakness. We report the case of a patient who presented with chronic LBP and mild muscle weakness as chief complaints and suspected with KD based on thorough physical examination and electrophysiological testing. KD was subsequently confirmed through molecular genetic testing.

CASE REPORT

A 52-year-old male patient presented to a secondary hospital in 2018 due to chronic LBP and persistent muscle weakness that began since 2013. In the past few years, chronic LBP was exacerbated, and the patient's exercise endurance declined.
Subsequently, the patient was referred to the rehabilitation medicine department at our hospital in 2023. Physical examination showed mild muscle weakness in some proximal muscles, increased deep tendon reflex, and tongue muscle atrophy, but the patient did not show fasciculation and pathological reflex. Overall, a motor neuron disorder was suspected. Lumbar spine MRI was performed to rule out central nervous lesion and radiculopathy, and there was no significant difference from the previous examination in 2018, such as showing only some degenerative changes (Fig. 1.) In terms of the electrophysiological examination, the motor nerve conduction test showed decreased latency and amplitude in some nerves but was close to normal compared to that in the sensory nerve conduction test. Sensory nerve conduction tests showed decreased latency and amplitude in most nerves (Table 1.). Needle electromyography (EMG) revealed widespread reduction in motor unit action potential recruitment with large amplitude and long duration. Fasciculation potentials, diffuse fibrillation potentials and positive sharp waves were detected. Thus, molecular genetic testing was performed under the suspicion of KD. CAG repeats were increased to 46, based on which KD was confirmed (Fig. 2.).

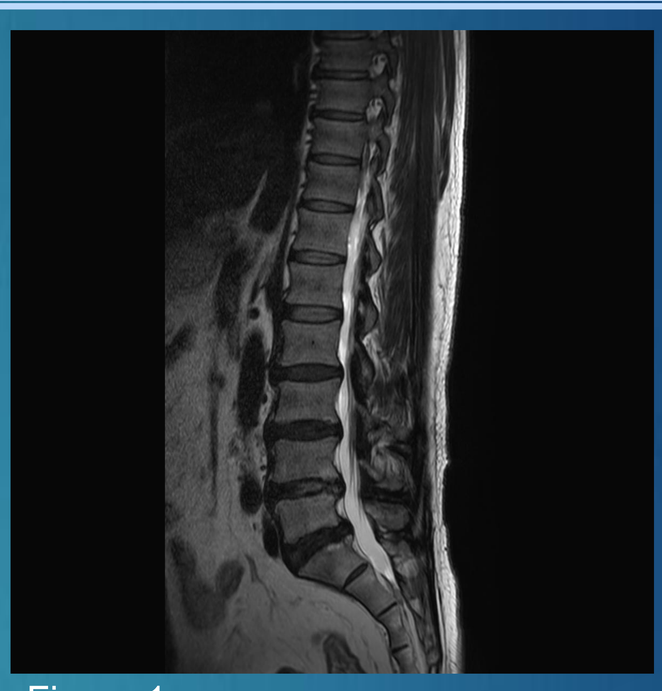

Figure 1.
T2 weighted images of the lumbar spine show degenerative changes.

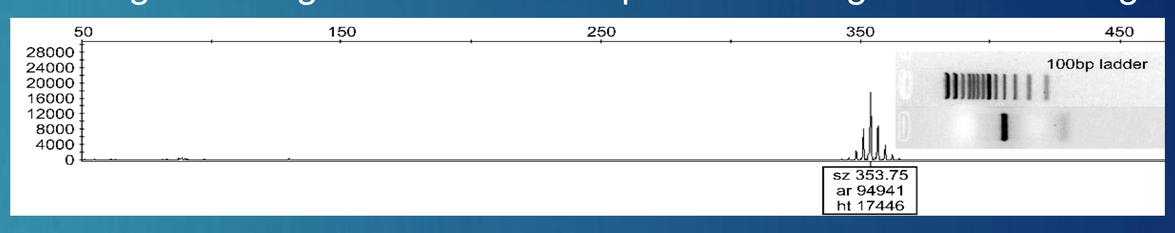

Figure 2.
The number of CAG repeats increased to 46, thereby confirming the diagnosis of Kennedy's disease.

Nerve	Stimulation	Right			Left		
		Latency (ms)	Amplitude (mV)	CV (m/s)	Latency (ms)	Amplitude (mV)	CV (m/s)
Motor							
Median at APB	Wrist	4.43	10.2	-	4.48	9.8	-
	Elbow	8.33	9.2	56.8	8.39	9.6	53.2
Ulnar at ADM	Wrist	3.33	11.8	-	3.02	11.0	-
	Below elbow	6.51	11.6	66.7	6.51	10.3	55.6
Peroneal at EDB	Ankle	4.01	2.2	-	4.90	1.8	-
	Fibular head	10.42	1.5	44.0	10.94	1.9	46.5
Tibial at AH	Ankle	3.91	19.8	-	3.70	18.0	-
	Fibular head	11.67	15.8	45.4	11.30	15.2	46.2
Sensory							
Median at Digit II	Wrist	3.44	6.1	41	3.49	8.8	40
Ulnar at Digit V	Wrist	2.76	7.1	51	2.60	6.3	54
Sural	Calf	3.28	5.1	43	3.23	4.8	43
Superficial peroneal	Lateral leg	3.33	2.5	42	3.39	3.9	41

Table 1. Findings of Nerve Conduction Study

DISCUSSION

The patient in our case had chief complaints of chronic LBP and mild muscle weakness only, without the other common symptoms, such as gynecomastia, testicular atrophy, erectile dysfunction, and diabetes mellitus. Furthermore, the patient also had no notable family history. However, physical examination revealed more significant muscle weakness affecting the proximal than the distal muscles, along with prominent increase in deep tendon reflex, tongue atrophy, and elevated CPK on blood test. Thus, we performed EMG to differentiate myopathy and MND. As characteristic EMG findings of KD were observed, we were able to confirm KD through molecular genetic testing.
A non-negligible percentage (12.8%) of chronic LBP cases is caused by other diseases, including congenital abnormalities and deformities, and 1% of the cases may be caused by a severe disease, including spondylitis, spinal tumor, and genetic disorders, as opposed to a simple structural issue. This highlights the importance of detailed history taking and neurological testing. The reported patient had only received symptomatic treatment for chronic LBP and mild muscle weakness before presenting to our hospital; however, his symptoms progressed without marked improvement. This case is significant since KD, an uncommon genetic disorder, was diagnosed subsequently based on detailed history taking and neurological testing.