Weakness and Progressive Muscle Tightness Diagnosed as Kennedy Disease

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ABSTRACT

Kennedy disease is a rare X-linked neurodegenerative disorder that affects patients in 30-50 years of age. It is caused by CAG-repeat in androgen receptor gen. There is no known effective treatment for Kennedy disease. We report a 60-year-old man who had fasciculations and proximal and distal muscle weakness. Physical examination showed involvement of the bulbar musculature accompanied by tongue atrophy and perioral muscle weakness. Furthermore, he had bilateral gynecomastia. Laboratory and imaging findings were normal, except electromyography that showed chronic proximal and distal denervation. Finally, the patient diagnosed with Kennedy disease according to clinical presentation and EMG abnormality that confirmed with genetic study.

Keywords: Spinobulbar muscular atrophy; Kennedy disease; x-linked; Neurodegenerative

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INTRODUCTION

Kennedy disease or spinal-bulbar muscular atrophy (SBMA) is a X-linked neurodegenerative disorder that is caused by CAG trinucleotide repetition on chromosome Xq11-12 [1]. Kennedy disease usually manifests with slowly progressive atrophy and limb muscles weakness in third to fifth decades. Primarily, it is usually characterized by muscle cramps and fasciculation that may progressed to proximal muscles weakness of hip and shoulder. Affected patients commonly lose their ability to walk upstairs, 10-20 years after primary symptoms. Although sexual differentiation and secondary sexual characteristics are normal, patients develop some symptoms related to androgen insensitivity like gynecomastia and testicular atrophy and even infertility [2-4].

We report a 60-year old man with 12 years history of tightness in his legs presented with aggravation in his muscle weakness and twitching, and finally diagnosed with Kennedy disease based on history taking, physical exam, laboratory data and genetic study.

CASE PRESENTATION

A 60-year-old man presented to our neurology department with complaining of progressive muscle weakness, cramping, and twitching. His symptoms first appeared about 12 years ago with cramping and tightness in his legs. Three years later, his shoulders and arms were also affected. The weakness on the left side was greater than right. He had some difficulties in walking and standing from a chair. His problems progressed to the point that he was unable to holding objects in his hands or lifting heavy things. His muscle weakness have flactuating pattern during the day.

He didn't report constipation, bowel or bladder dysfunction, or numbness. He also denied swallowing, speech or visual problems. His past medical history was negative and he was not on any medication. His family history was negative for any neurological disorders.

On physical examination, blood pressure was 130/90 mmHg, heart rate was 70/min, respiratory rate was 17/min, and body temperature was 37°C. Further physical examination showed mild gynecomastia and

mild atrophy of the scapular and hand muscles. Cranial nerve examinations showed decreased labial strength and fasciculation around lips. A fine tremor was observed in both hands symmetrically. Mild atrophy was present in the proximal and distal of upper and lower extremities. He had areflexia in biceps, triceps, brachioradialis, patellar, and ankle joint. Hoffman's sign and jaw jerk was negative. Cerebellar examinations were normal and the patient gait was intact. His mental status was in normal limit.

Laboratory findings including complete blood cells count, biochemistries and electrolytes were in normal range. Brain, cervical, thoracic and lumbosacral Magnetic Resonance Imaging (MRI) revealed no abnormality. Electromyography and nerve conduction study were suggestive for widespread chronic proximal and distal denervation without fibrillation.

There was a scattered 1+ fasciculations in arms and legs accompanied by chronic partial denervation in tongue.

This patient presented with bulbar and spinal muscular atrophy, also he had a mild degree of gynecomastia. Clinical manifestation of the patient and pattern of muscular involvement raised the possible diagnosis of Kennedy's disease that was confirmed by genetic study.

DISCUSSION

Spinal and bulbar muscular atrophy (SBMA) or Kennedy disease is a progressive disease caused by defect in X chromosome with an expansion more than 35 CAG-repeat in exon-1 of the androgen receptor gene [4,5]. Clinically, about 2% of patients with SMBA are misdiagnosed as amyotrophic lateral sclerosis (ALS) [6]; however, CAG-repeat length is normal in ALS [7]. In this report, SBMA was diagnosed in an Iranian patient with typical clinical manifestation and confirmed with genetic study.

In this patient, proximal and distal muscle groups were affected in both upper and lower extremities which this pattern was not consistent with any myopathy.

As shown in this patient, bulbar and spinal muscular involvement, lack of deep tendon reflex and gynecomastia may be seen in Kennedy's disease. Also, affected patients may have testicular atrophy and insulin independent diabetes [4].

Genetic study beside history taking, physical examination and electrophysiological study is necessary for the diagnosis of Kennedy disease [4].

Improving mobility and decreasing complication are two main goals in the management of Kennedy disease. There is no known medical treatment for Kennedy disease, however rehabilitation and behavioral measures play important role in this disease [7].

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