Weakness and Progressive Muscle Tightness Diagnosed as Kennedy Disease

Behdad Behnam, Mehran Arab Ahmadi, Farzad Ashrafi

Functional neurosurgery Research Center, Shahid Beheshti University of Medical Sciences, Tehran, Iran.

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

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 fluctuating 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. Clinically, about 2% of patients with SMBA are
misdiagnosed as amyotrophic lateral sclerosis (ALS); however, CAG-repeat length is normal in ALS. 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.
Genetic study beside history taking, physical
examination and electrophysiological study is necessary
for the diagnosis of Kennedy disease.
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.

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