Email: somayehmostafaie@yahoo.com
References:


AMYOTROPHIC LATERAL SCLEROSIS OR KENNEDY’S DISEASE: A CASE REPORT OF A PATIENT WITH THREE YEARS OF MISDIAGNOSIS

Aliakbar Taheraghdam¹, Ali Pashapour², Somaiyeh Mostafaei³, Elyar Sadeghi Hokmabadi⁴

Abstract
Kennedy's Disease (KD) Bulbar and spinal muscular atrophy (BSMA) is an adult onset, X-linked, recessive disorder caused by expansion of a polymorphic CAG tandem repeat. Because Kennedy’s clinical symptoms overlap with some other neuromuscular disorders such as amyotrophic lateral sclerosis (ALS) or spinal muscular atrophies, KD sometimes is misdiagnosed or left unnoticed. Here we describe a case of Kennedy’s disease confirmed by genetic testing who had been diagnosed and treated with ALS for three years. We describe a 56-year-old man presented with progressive onset of lower limbs muscular atrophy, weakness, and fasciculations since five years ago. He also complained of instability and fatigue when walking. Since three years ago, he had difficulty in swallowing and talking. He denied any sensory symptoms and sphincter disturbances. On examination at disease beginning he had normal mental status, bilateral facial palsy, tongue atrophy and fasciculation. The patient had lower limb muscle atrophy with mild weakness. The deep tendon reflexes all were depressed and the plantar responses were abolished. There were no sensory and cerebellar signs. Neuroimaging didn't show any significant pathology. Electrodiagnostic examination (EDX) revealed neurogenic pattern with low compound muscle action potentials (CMAP) and acute and chronic denervation pattern in tested muscles, so the patient diagnosed with ALS and treatment started with Riluzole, six months ago we visited him again with new onset sensory symptoms of face and limb parenthesis. Sensory examination revealed mild impairment of pinprick and thermal senses at limbs although senses of vibration and joint position were normal. He had also perioral and facial fasciculations. Since patient's signs and symptoms progression was gradual with developing of sensory findings we questioned the initial diagnosis and reevaluate it later on, EDX study showed decreased lower extremities CMAP with absent F-wave and H-reflects. All sensory parameters were near absent. Genetic studies revealed an increased CAG repeat number (50 normal up to 34), confirming the diagnosis of Kennedy disease. KD is the most common disease which is confused with ALS and recognition of KD is important because its prognosis, natural history, family testing, and management is different from ALS so it is necessary to rule out KD in suspected male cases of ALS.

Keywords: Amyotrophic lateral sclerosis (ALS), Kennedy’s Disease (KD)

Address: Neurology Department, Tabriz University of Medical Sciences, Tabriz, Iran
Email: somaiyehmostafaie@yahoo.com

SOURCE: URMIA MED J 2013: 24(6): 466 ISSN: 1027-3727

¹ Assistant Professor of Neurology, Tabriz University of Medical Sciences, Tabriz, Iran
² Assistant Professor of Neurology, Tabriz University of Medical Sciences, Tabriz, Iran
³ Resident of Neurology, Neuroscience Research Center (NSRC), Tabriz University of Medical Sciences, Tabriz, Iran (Corresponding Author)
⁴ Resident of Neurology, Neuroscience Research Center (NSRC), Tabriz University of Medical Sciences, Tabriz, Iran