Case Report

Amyotrophic Lateral Sclerosis: The Most Common And Lethal Form Of Motor Neuron Disease-a Case Report From Middle East

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ABSTRACT:

A neurodegenerative disorder which is fatal, rapidly progressive and has no effective treatment till date is amyotrophic lateral sclerosis. Almost 90% of all cases occur in the sporadic form, with the rest occurring in the familial form. It is a devastating disease leading to death within 3-5 years in most cases. The diagnosis of AML is difficult to made in spite of acknowledgment for 140 years. It is diagnosed by clinical presentation which is a combination of upper and lower motor neuron signs and electro diagnostic studies which gives information about diffuse motor axonal injury. This neurodegenerative disorder results in degeneration of corticospinal tracts and anterior horn cells and involving motor neurons of the cerebral cortex, brainstem, and spinal cord. There are a variable signs and symptoms of this disease, so the diagnosis is very important for the management and better outcome of the patients. Cause of death in these patients is usually respiratory failure.

KEYWORDS: Amyotrophic Lateral Sclerosis, Motor Neuron disease, Neurodegenerative disorder, MRI

INTRODUCTION:

The most common form of is **Amyotrophic lateral sclerosis (ALS)**, which is also called **Lou Gehrig disease**,[1-3] which is causing progressive weakness and death of the patient due to respiratory failure. Others are also prone to the pseudobulbar affect.[1] It is a neurodegenerative disorder which is rapidly progressive and fatal resulting in degeneration of corticospinal tracts and anterior horn cells and involving motor neurons of the cerebral cortex, brainstem, and spinal cord. The diagnosis of AML is difficult to make in spite of acknowledgment for 140 years. The Escorial criteria are commonly used for obtaining a definitive diagnosis of ALS.[2] Within three years, more than 60% of patients died with AML. The treatment of choice for AML are Riluzole but it is effective for only two to three months.[3] The physician of patients with AML should manage the illnesses which can be treatable along with neuropathy. The underlying etiology is not known. Several genetic mutations have been found to be associated with familial ALS (FALS) which accounts for 10% of ALS.[4] With the combination of pathophysiology and newer treatment options, other diagnostic tests should be considered for the early detection of AML.

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CASE REPORT:

We have encountered a patient with age of 48 years who presented two months after the onset of bilateral lower extremity weakness which was progressive as it was initially started in his right leg and then progressed to his left leg. After that, patient felt muscle twitching. There was no known past medical or family history and no any history of trauma or insect bite. On Physical examination, the patient was thin, cachexic and well oriented with time, place, and person. He was fully conscious and no any loss of memory noted. However, decrease in strength is noted which was mild at the hips and knees but more pronounced at ankles and toes bilaterally, more marked on right side. There was fasciculation in both arms and legs. On sensory examination, sense of vibration was decreased in legs and sense of position was decreased in toes. Achilles tendon reflexes were absent but other reflexes were present and normal. No significant abnormality seen on cerebellar examination. He was unable to walk without support and unable to stand from sitting position. CT scans of the abdomen and chest were done which showed no significant abnormality. So the diagnosis of acquired polyneuropathy was made for which treatment was started with 60 mg steroids and 120 mg azathioprine daily. But no positive change in the disease process was seen and the patient was deteriorated with progressive decrease in strength in arms, legs as well as intrinsic muscles of hands. Further work up was done with MRI brain in axial, coronal and sagittal planes which showed increased signal intensity in corticospinal tracts bilaterally on T2WI and DWI. Thus, on the basis of clinical history, examination and MRI brain findings, diagnosis of Amyotrophic lateral sclerosis was made.

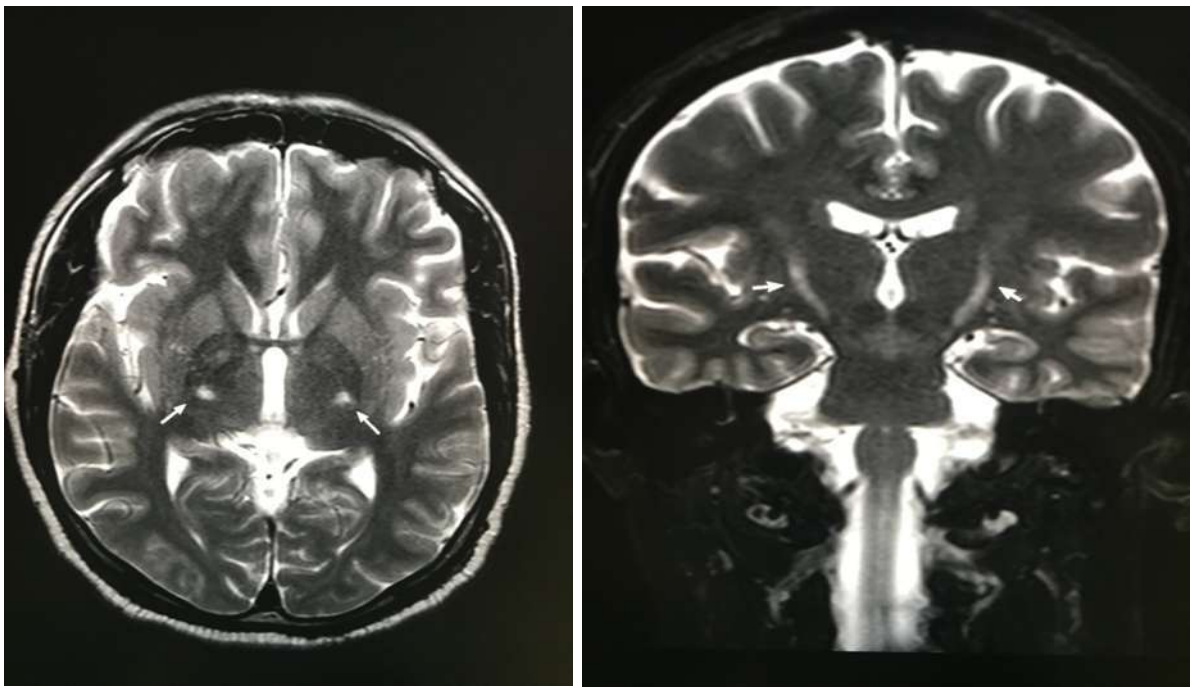

Figure 1: T2WI in Axial and Coronal sequences showing high intensity signals in corticospinal tract (internal capsule).

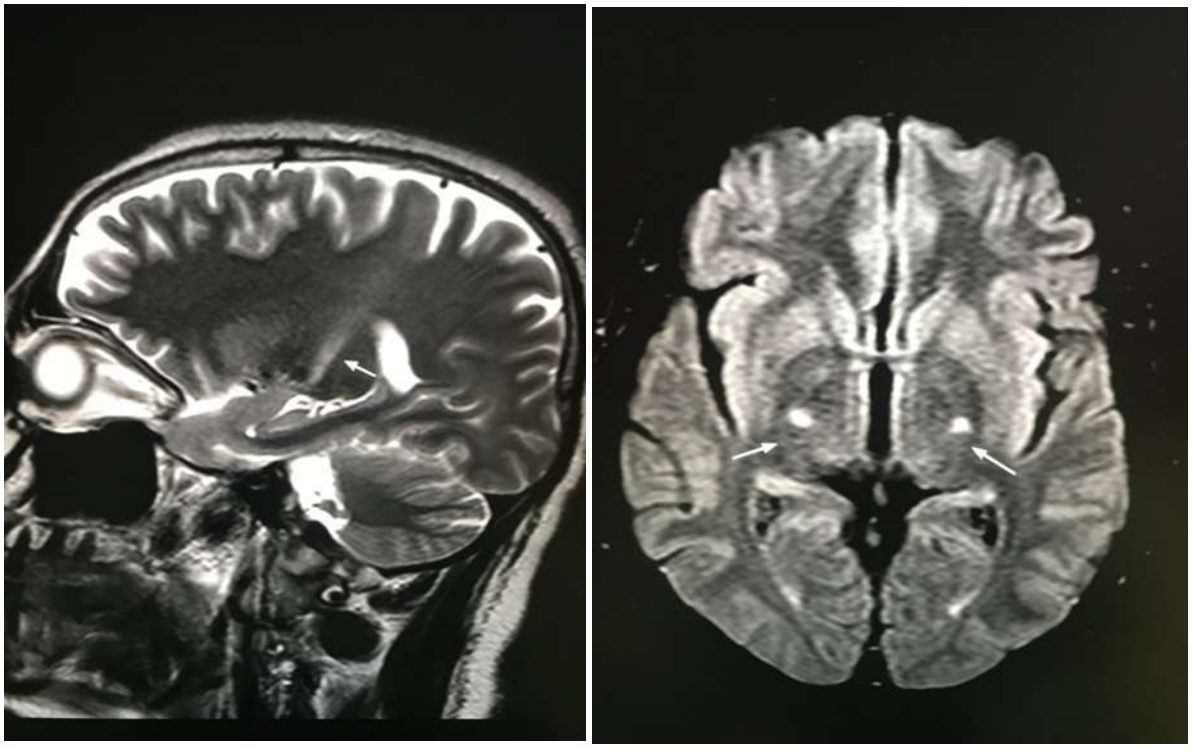

Figure 2: T2WI and DWI in Sagittal and Axial planes showing high intensity signals in corticospinal tract (internal capsule).

DISCUSSION:

ALS is diagnosed on the basis of signs and symptoms of upper and lower motor neurons dysfunction along with laboratory evidence. No any significant test is present for the diagnosis of AML which is widely accepted. It is diagnosed with electro diagnostic studies. Neuro imaging and lab tests are used for the exclusion of other diseases.

Signs and symptoms of ALS include asymmetric weakness of extremities (60%– 80%), bulbar symptoms (20%), respiratory muscle weakness (1%–3%), generalized weakness

in limbs and bulbar muscles (1%–9%), axial onset with head drop or truncal-extension weakness, muscle atrophy, fasciculations, and cramps.[5-6] Although diagnostic criteria for ALS were revised in 2000, the unavailability of the specific diagnostic test is a big hindrance to detect ALS in the early stages.[7]

Weight loss is also seen in AML which is a late finding and is due to difficulty in swallowing and difficulty in respiration during eating along with weakness of muscles of mastication and increase metabolic state.[8-9] Due to the insufficiency of dietary intake and weight loss, the state of catabolism and weakness of respiratory muscles become worsen progressively. So the vicious cycle is continued and the disease get worsen. The poor prognostic factor is weight loss and BMI =18.5 kg/m2. The life of ALS patient is associated with the function of respiratory muscles. The episodes of apnea and dyspnea are poor factor for the quality of life of patients. With the combination of clinical history, physical examination and MRI brain, the diagnosis of AML was made. AML was treated with corticosteroid with symptomatic treatment, but due to the involvement of respiratory muscles the patient is died due to respiratory insufficiency.[10-11]

Various questions in a survey were asked from university students regarding ALS which showed some heard about this disease first time, only some of them have basic information about this disease, some had knowledge about signs and symptoms and only some of them have knowledge regarding its diagnosis and treatment strategies. These types of awareness studies helpful for medical student about disease, symptoms, treatment and precaution etc.[12-17]

CONCLUSION:

In Patients with rapidly progressive muscle weakness and neuropathic symptoms, one should suspect AML. The diagnosis of AML should be made early because the disease should be arrested before progression. The occurrence of the disease is increasing every year, so the attempt should be made for the research and awareness of the disease process.

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