

A Case Report of Amyotrophic Lateral Sclerosis Presented As Dysarthria and Dysphagia

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Abstract

Amyotrophic lateral sclerosis is a fatal and rare disease of the motor neuron system, caused by the degeneration of the motor neuron and featured by a progressive muscular palsy. This condition gradually progresses and cause spasticity, weakness and wasting of the muscles. This condition can affect gait. The incidence of ALS in different population ranges from one-two cases per 100,000 people per year. Clinical features caused by this condition in upper and lower extremities are more obvious than other types of paralysis caused by other conditions. Major clinical manifestation of MND include dysphagia that cause decreased survival and quality of life. Dysarthria and dysphagia of amyotrophic lateral sclerosis caused by involvement of medulla that in 1/3 cases seen in early stage of the disease. Reevaluation for the dysphagia conclude the history, clinical examination, electromyography, nerve condition velocity and video fluoroscopy swallow study and some other conditions such as cervical spondylosis, myelopathies, multifocal motor neuropathy space occupying lesion, stroke and other related pathology should be excluded. The average life span for this condition is 2-5 years. Treatment is multi-disciplinary as conservative and supportive. Physiotherapy for increased of ability and drug Riluzole is used for prevention of progressive course of this disease are advised. Noninvasive ventilation may increase the span and quality of life. We here report a case of 55 years old lady with dysphagia, dysarthria and muscular cramps since 18 months with gradually progressive course, whom previously was treated for conditions such as ischemic stroke, gastritis, esophagitis and mental conditions, but with help of good clinical evaluation and laboratory exams such as electromyography, nerve conduction velocity, this condition is diagnosed and reported.

Key words: Dysphagia, Amyotrophic Lateral Sclerosis (ALS), bulbar palsy, Dysarthria, Motor Neuron Disease (MND), Lower Motor Neuron (LMN), Upper Motor Neuron (UMN), NCV, EMG and Riluzole.

Introduction

ALS is a fatal and rare form of MND of the nervous system that results from the degeneration of motor neurons, it is otherwise known as Lou Gehrig's disease or motor neuron disease^{1, 2, 8, 10}. This disease is characterized by rapidly progressive muscular paralysis caused by degeneration of motor neurons in the motor cortex, corticospinal tracts, brain stem and spinal cord leads to muscle wasting (atrophy), muscle spasticity, muscle weakness and failure to send messages to the muscles^{1, 8, 9, 10}.

The incidence of the disease in adults reaches 1 - 2 cases per 100,000 populations^{1, 7, 9}. Males are affected more than females with an M: F ratio about 1.5:1¹, although recent data suggests that the gender ratio may be approaching equality¹. The age at onset of the disease varies, with sporadic occurring among young people. Almost most cases occur between the ages of 55 and 65, and only about 5% occur before the age of 30^{1, 7}.

This disease is characterized by the involvement of upper and lower motor neurons in the motor cortex and spinal cord^{1, 2}. Clinical manifestations of the upper and lower neurons of the disease is so typical that it cannot be seen in other diseases that cause paralysis¹. Clinical manifestations of this disease are variable and in two thirds typical cases of ALS have a spinal form of the disease (i.e., LMN) where the onset of the symptoms begins (possibly from distal or proximal upper or lower extremities) with localized weakness and muscle wasting and gradually spasticity progressed in weakened atrophic areas and causes walking problems^{1, 8, 10}. limbs examination showed signs of bilateral pyramidal tract of UMN involvement (extensor plantar bilaterally, spasticity, clonus, positive Jaw jerk and exaggerated deep tendon reflexes) and signs of anterior horn cell damage or lower motor neuron involvement (severe muscle atrophy and wasting)^{4, 10}.

Difficulty in swallowing (Dysphagia) is one of the most important clinical manifestation & frequent features of ALS and can result in reductions in life expectancy and quality of life³. Dysphagia is a commonly encountered symptom in ENT clinical practice but due to amyotrophic lateral sclerosis (ALS) is relatively uncommon in this setting. Patient presented symptoms such as dysphagia for liquids and solids & dysarthria shows bulbar involvement which is associated with hoarseness of voice, muscle atrophy and weakness in the trunk, neck, back and both proximal upper limbs for lower motor signs (pseudo bulbar palsy)^{1, 3, 9, 10}. Fasciculation is not present in involvement of CNS and the sensory system remain intact too¹⁰. Dysphagia is a common symptom of ALS and leads to increased risk of aspiration, malnutrition, weight loss and dehydration, and reduce life expectancy and quality of life⁸. The High Resolution Manometer (HRM) has recently been discovered, which is a safe and effective instrument in the evaluation of assessing the oropharyngeal Neurogenic Dysphagia⁸. The Evaluation of dysphagia may include: history, clinical examinations, Electromyography (EMG), Nerve Conduction Velocity (NCV), Video - Fluoroscopic Swallow Study, Radiological Esophagogram, Flexible Endoscopic Examination, Ultrasound Examination

and Conventional Manometry^{1, 8, 12}. The diagnosis of ALS for evaluation of prognosis & treatment is very important, because the resulting Paralysis is progressive and leads to death due to respiratory failure in ALS cases. If the respiratory failure present, life expectancy is reduced to 2 - 5 years^{1, 4, 8, 9, 10, 14}.

We report here an ALS case in a 55 - year - old woman who had dysphagia associated with disturbances of speech (dysarthria), weakness in the hands, and sometimes feeling of cramps in her legs, which is with a detailed history of completion and EMG / NCV examinations has been diagnosed ALS, and the patient has previously took a variety of medicines under different diagnoses (such as ischemic stroke, gastritis, esophagitis and mental disorders).

Case report

A 55-year-old woman came to Ali Abad teaching hospital for diagnosis and treatment, who presented with progressive dysphagia and hoarseness of voice associated with disturbances of speech (dysarthria) since 18 months and two months history of weakness with a gradual progression in her hands (which recently caused problems with uplifting hands upward) and sometimes having of cramps in her legs, she was admitted to a Neurology & Psychiatry service, although the during of this 18 months she was hospitalized for this purpose under different diagnoses (such as ischemic shock , gastritis, esophagitis and mental disorders) and patient has taken various types of medicines from inside the country and abroad from Pakistan despite the lack of effectiveness of treatment. At first, she complained of swallowing and coughing while eating, and gradually became more difficult to swallow, and now, when she swallows, she chews the solid food until it is liquefied and become relatively fluid. When she swallows it later, it gets into the throat problem and sometimes when swallowed, occasionally she coughs and she each time gets a few mouthfuls of food and walk for 5 - 10 minute to digest the food as she feel that is stuck in the throat and chest.

The patient has a little problem during of speaking such as hoarseness of voice, and spastic Slurred speech associated with a nasal twang, and with torch examination spastic atrophy (because of muscle wasting) and fasciculation visible over tongue. The positive response of swallowing reflex (Gag Reflex) with multiple stimulation is possible. She feels heartburn and sometimes during of swallow she is suffering from coughs and regurgitations of food from the nose, it is also a shocking condition for her (these are confirming and shows the involvement signs of peripheral, central, and bulbar motor neurons).

There is no family history of MNDs, and she has fully conscious with normal vital signs, but she seems to be ill. The patient has not had proper and good nutrition due to dysphagia, and complain about heartburn and epigastric pain. Sometimes she has feeling of weakness in raising her hands upward and having cramps in her legs, by physical examination of abdomen the epigastric area is a little sensitive and painful with palpation. There is no atrophy remarked in the hands of the patient, but in terms of muscular strength the

degree of muscle weakness can be determined between about 4 - 5. Also, the muscular ton of all the muscles of the body is not changed, and all superficial and deep reflexes around the upper and lower limbs are intact.

Except for Electromyography /Nerve conduction Velocity (EMG /NCV) examinations performed on the patient which also helps in diagnosis of ALS, other laboratory examinations do not indicate any specific pathological involvement to helps in diagnosis of ALS and are reported in *Table (1 & 2)* as following:

Table 1: Shows results of different diagnostic examinations.

Examinations	Results
Abdominal Ultrasound	Right Kidney small stone
Echocardiography and ECG	Normal Results
Endoscopy (Gastroscopy)	Mid Gastritis
Brain CT Scan	Normal Study
EMG (Electromyography) & NCV (Nerve Conduction Velocity)	EMG: Pure motor severe axonal neuropathy involve bulbar, cervical and lumbosacral myotomes; in appropriate clinical context consistent with anterior horn cell disorders. NCV: denervation was noted in tibialis anterior, gastrocnemius, vastus medialis, and cervical, thoracic with Para spinal muscles; chronic neurogenic motor units were noted in upper and lower limbs. Sensory nerves are normal.

Table 2. Shows results of blood biochemical and serological tests.

Biochemical tests	Results	Serological tests	Results
FBS	122 mg / dl	H. Pylori Antibody	Negative
Total Bilirubin	0.85 mg / dl		
Direct Bilirubin	0.25 mg / dl		
SGOT	16 U/L	HCV Antibody	Negative
SGPT	14 U/L		
Alkaline Phosphatase	98 U/L		
Triglyceride	214 mg / dl	HBS Antigen	Negative
Total Cholesterol	189 mg / dl		
HDL	34 mg / dl		
LDL	87 mg / dl		
Creatinine	0.9 mg / dl		

As shown in the table above the electromyography (EMG) examination of the patient is abnormal which shows pure severe axonal motor neuropathy involve bulbar, cervical and lumbosacral myotomes with disorders of anterior horn cell. Nerve Conduction Velocity (NCV) recorded denervation in tibialis anterior,

gastrocnemius, vastus medialis, and cervical, thoracic with Para spinal muscles; chronic neurogenic motor units were involved in upper and lower limbs with normal Sensory nerves.

Diagnosis of motor neuron disease–Bulbar type of Amyotrophic Lateral Sclerosis (MND-Bulbar type of ALS) is based on the history and clinical evaluation as mentioned above (indicating central, peripheral associated with bulbar motor neuron involvements). Other brain pathologies such as space occupying lesions, stroke and some other related conditions have been rule out. In this case, which began with bulbar involvement and continued with the hoarseness of the voice, dysphagia and the progress of it to the limbs, she previously for this reason took a variety of medicines according to different diagnoses. Although ALS does not already have specific and effective treatment, it has to be treated to prevent the progress of the disease and improve the quality of life of the patient. Therefore, the principle of speech therapy, sports, physiotherapy and advised along with dietary supplementation is currently recommended for the patient as well as to prevent the progression course of the disease. Treatment of the patient followed by these medications: Riluzole, is prescribed for prevention of progressive course of ALS, Tizanidine prescribed for the legs cramps and spasticity and Gabapentin is prescribed for neuropathy. The patient is now relatively satisfied with her treatment except Riluzole (unfortunately the mentioned medicine is not available inside the country). The patient has been instructed that in the inability of self-feeding, she should insert the nasogastric tube to prevent risk of aspiration, malnutrition, weight loss and dehydration, also if ineffective for enteric feeding she should perform a gastrostomy, percutaneous endoscopic gastrostomy, percutaneous radiologic gastrostomy or radiologically inserted gastrostomy.

Discussion

Amyotrophic lateral sclerosis (ALS) is a rare and fatal disease of the nervous system, that caused by degeneration of motor neurons. The incidence of ALS is reported to be between 1.5 and 2.7 (1 - 2) per 100,000 population/year. However, the prevalence ranges from 2.7 to 7.4 per 100,000 population/year.^{1,2,8,9,10}. Amyotrophic lateral sclerosis is believed to cause by excitotoxicity of glutamate, oxidative stress and axonal injury. Increased glutamate activity, or increased level of free radicals that damages motor neurons which is responsible for the control of the involuntary muscle¹⁰. The best possible way to limit the progression of ALS is early diagnosis together with anti-glutaminergic therapy, physiotherapy and quitting smoking. Sports are very essential to minimize wasting of muscles. Riluzole therapy prevent the progression of disease and Baclofen therapy for limbs spasticity. Life style modifications like regular exercise and quitting smoking proven control over the progression of disease^{4,5,6,10,11}.

Dysphagia is one of the most important prognostic factors and a common symptom in ALS, which occurs in one third of cases at the onset of the disease, although it generally occurs in the later stages of the disease, and leads to increased risk of aspiration, malnutrition, weight loss and dehydration. Therefore, preparation

and careful evaluation and prosecution are important for swallowing tasks^{1, 8, 9, 10, 14}. Pathophysiological mechanism of dysphagia in ALS is as follows: First, submental muscle activity of the laryngeal elevators is significantly prolonged. Second, opening of the cricopharyngeal sphincter muscle is delayed or the muscle closes prematurely. Third, there is a significant lack of coordination between the laryngeal elevator muscles and the cricopharyngeal sphincter muscle. Altogether, these abnormalities may lead to decreased ability to initiate complete reflex swallowing in ALS⁸. Diagnosis is important to evaluate the consequences and treatment of disease¹⁰. Although there are different diagnostic methods for ALS, but in our country the EMG & NCV examinations are good diagnostic method for this disease, because it is not possible to perform other diagnostic methods for the disease and also the diagnosis of UMND and LMND possible with EMG & NCV. Positional changes during meal, dietary modifications and augmented feeding technique with a supplemental tube and gastrostomy were provided to treat dysphagia of the patient^{4, 5, 6, 9, 10, 11, 14}. Among the causes of neurological dysphagia like stroke, accidental or surgical trauma of throat, multiple sclerosis and space-occupying lesions such as tumors, the MND (such as ALS) is relatively rare cause¹. Classic form of ALS include combined UMN and LMN involvement^{1, 10}. Progressive bulbar palsy shows predominant bulbar involvement signs such as dysphagia and dysarthria^{1, 10, 12}.

Progressive muscular atrophy shows predominant LMN involvement. Primary lateral sclerosis shows predominant UMN involvement¹. ALS or Charcot disease or Lou Gehrig disease is the most common MNDs which is associated with the progressive course of the cortico-bulbar and cortico-spinal tracts involvement⁹. In this form of the disease, progressive dysphagia affecting the oral and oropharyngeal stage together with dysarthria and anarthria account for the misery¹⁴.

The mean age of onset for ALS varies between 55 and 65 years with a median age of onset of 64 years. Males are affected more than females with an M:F ratio about 1.5:1^{1, 7}. There is no consistent association between a single environmental factor and risk of developing ALS. It was found that only smoking is likely to be associated with ALS, while other risk factors were weakly related^{1, 13}. Amyotrophy refers to the atrophy of muscle fibers, which are denervated as their corresponding anterior horn cells degenerate, leading to weakness of affected muscles and visible fasciculation^{1, 10}. Lateral sclerosis refers to hardening of the anterior and lateral cortico-spinal tracts as motor neurons in these areas degenerate and is replaced by gliosis¹. Symptoms of ALS include limb muscle weakness, cramps, occasionally fasciculation, disturbances of speech, swallowing, dysarthria, pathological laughter or crying¹. UMN dysfunction leads to stiffness, brisk or abnormally spreading tendon reflexes, presence of abnormal reflexes (hyper reflexic jaw jerk, Babinski sign), and loss of dexterity in the presence of normal strength^{1, 10, 12}. LMN dysfunction manifests as muscle twitching (fasciculation), reduction of muscle bulk (atrophy), foot drop, depressed reflexes, breathing difficulties¹. CT- Scan and MRI is used to rule out structural lesions of muscle¹. Nerve biopsy must be considered if the presentation is atypical, biochemical markers, and genetic studies are also

considered¹. Complications of ALS include progressive inability to perform activities of daily living, including handling utensils for self-feeding, deterioration of ambulation, aspiration pneumonia, deep vein thrombosis (DVT), pulmonary emboli, bed sore, respiratory insufficiency, Wheelchair-bound or bedridden patients are likely to have decubitus ulcers and skin infections¹.

Diagnosis is mainly clinical with both features of UMN and LMN dysfunction such as weakness, atrophy, fasciculation of muscles occurring in combination with increased tone and hyperreflexia¹. In ALS EMG study confirms the diagnosis and helps to exclude other peripheral causes. EMG study shows fibrillation and fasciculation potentials of high amplitude and long duration polyphasic motor units. Nerve conduction study is normal in sensory and abnormal in motor with reduced motor compound muscle action potentials^{1, 5}.

Treatment of ALS is multi-disciplinary as conservative and supportive, include skeletal muscle relaxants for spasticity, Quinidine or dextromethorphan for emotional liability due to the pseudobulbar effect, Riluzole is with modest effect on prolonging life for prevention of progressive course in ALS patients (100 mg probably prolongs median survival by 2-3 months when taken for 18-month duration and liver function should be regularly monitored during therapy), For prevention risk of aspiration, malnutrition, weight loss and dehydration due to dysphagia (which is a common symptom of ALS) the three options available for enteric feeding include percutaneous endoscopic gastrostomy, percutaneous radiologic gastrostomy or radiologically inserted gastrostomy, and nasogastric tube feeding^{1, 5, 6, 9, 11}. There is no cure for progressive dysarthria in ALS, but handwriting can help them and solve their problems, and speech therapy is also recommended. Life style modifications to improve the effectiveness in ALS like regular exercise regimen and quitting smoking proven control over the progression of disease^{1, 10}. ALS is a fatal disease. Overall median survival from onset of symptoms for ALS ranges between 2 and 3 years for bulbar onset cases and 3-5 years for limb onset ALS cases^{1, 14}.

CONCLUSION

The early diagnosis is essential for prevention of disease progression. ALS should be considered in patients with a rapidly progressive, unexplained neuropathic process. In patients of dysphagia focusing only the local signs alone, may miss the diagnosis of a systemic cause, because a complete neurological examination is essential in patients having dysphagia associated with neurological disturbances for precise diagnosis and management of neurodegenerative disorder. Although ALS is incurable, there are treatments that can prolong meaningful quality of life; therefore, diagnosis and its treatment are important to both patient as well as family. The incidence of ALS is increasing every year in the world so efforts must be taken to promote awareness of the disease and encourage the research for ALS management.

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