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Amyotrophic Lateral Sclerosis with Asymptomatic Syringomyelia: A Case Report

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Abstract: ***Amyotrophic lateral sclerosis (ALS) or also known as Lou Gehrig disease is a neurodegenerative disease that attacks motor neurons. Prevalence in Western countries ranges from an average of 5.2 per 100,000. The average onset of ALS incidence is approximately 60 years, where men affected more frequently than women. The diagnosis of ALS is clinically confirmed. Electrodiagnostic testing contributes to diagnostic accuracy and imaging examination is used to exclude other etiologies. We report a 36-year-old female patient diagnosed with ALS clinically and electrodiagnostically which had an image of an asymptomatic syringomyelia.***

Keywords: Amyotrophic Lateral Sclerosis (ALS), syringomyelia, electrodiagnostic

1. Introduction

Amyotrophic lateral sclerosis (ALS) or also known as Lou Gehrig disease is a neurodegenerative disease attacking motor neurons. Amyotrophy is defined as the presence of muscle fiber atrophy, which innervated by a degenerative anterior horn cell (causing muscle weakness and fasciculation) while lateral sclerosis is defined as hardening of the lateral and anterior corticospinal tracts so that the motor neurons in the region degenerated [1]. Prevalence in Western countries ranges from an average of 5.2 per 100,000 with an average age of ALS events is about 60 years. The incidence rate in men is higher than women with a ratio of 1.5: 1 [2].

About 5-10% of cases of ALS are familial and 90-95% is sporadic. Molecular changes that cause motor neurons degeneration in ALS are still unknown, however, there may be complex interactions of various cellular pathogenic mechanisms such as genetic factors, excitotoxicity, oxidative stress, mitochondrial dysfunction, axon transport disturbances, neurofilament aggregation, protein aggregation, inflammatory dysfunction and non-cell contribution nerves, deficits in neurotrophic factors and signal path dysfunction [2-7].

In ALS disease, the somatosensory component is not disrupted so its clinical manifestations consist of motor disturbances showing signs of UMN and LMN paralysis simultaneously [2]. The diagnosis of ALS is clinically established. Additional examination in the form of electrodiagnostic and imaging tests contribute to diagnostic accuracy and exclude the anatomic abnormalities that cause these clinical manifestations [2,3]. Patients with ALS disease have an average survival rate of 3-5 years. Aspiration of pneumonia and other medical complications of immobilization play an important role in the morbidity of patients with ALS. Until now, ALS has a poor prognosis because it has not cured yet, and the focus of management is to extend the survival and quality of life of patients, therefore early diagnosis is essential for patients and families.

2. Case Report

We report a case of 36 years old woman, a Balinese, who came to the Neurology clinic of Sanglah Hospital on September 8, 2017. The patient came with main complaint of weaknesses in both hands. It initially felt in 2009 then since 5 months before hospitalization. She has difficulty raising her hands. The complaints first felt in the right hand where it feels a bit difficult to hold some objects, then followed by her left hand after 3 years. Complaints worsened gradually and now she complained of difficulty when lifting both hands. These weaknesses were also accompanied by the shrinking of the hands that has been felt since approximately 5 years ago. She had just realized both hands were getting smaller since 5 months ago, where the right hand is felt smaller than the left hand. She was unaware if there were any weaknesses of the limbs. Patient said the legs could still move as normal and she can walk alone. Tingling or numbness is denied. No headache, nausea or vomiting, double glance or blurry, history of head trauma, nor history of fever.

Neurologist examined the patient in 2009 when the first complaint arose. She had cervical MRI with the results were within normal limits. She was diagnosed with ALS, however she never checked herself back until today. The patient finally decided to do the re-examination because she felt her complaints worsened.

Patient denied any history of high blood pressure, diabetes, heart disease, or previous stroke. She said there was no family member from the father or mother who, for her knowledge, suffered the same complaint. Patient is a housewife, only doing light daily activity at home. She has no history of smoking or consuming alcohol.

Examination shows that the patient is compos mentis, blood pressure 100/70 s 36.7°C, 98% oxygen saturation. Neurologic examination show; flaccid tetraparesis of the superior extremities (grade 4-33|34-4) and spastic in the inferior extremities (grade 554+|4+55), atrophy of the superior bilateral extremities muscles; bilateral feet clonus, absence of autonomic disorder and no pathologic reflexes.

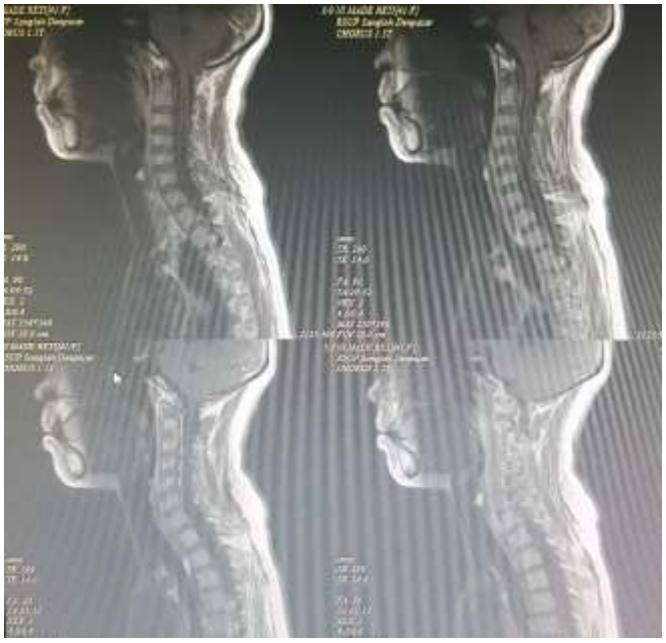
Table 1: EMG result

CMAP	
N. Right Median	Elongated distal latency, shortened amplitude, normal KHS
N. Right Ulnar	No respond
N. Right Radial	Elongated distal latency, shortened amplitude, slowing KHS
SNAP	
N. Right Median	Normal distal latency, normal amplitude, normal KHS
N. Right Ulnar	Normal distal latency, normal amplitude, normal KHS
N. Right Radial	Normal distal latency, normal amplitude, normal KHS
Needle EMG	
M. 1st dorsal interossei of right hand	PSW +4, fibrillation +4, fasiculation +4, MUAP: no respond, decreased recruitment, incomplete IP
M. Right biceps	PSW +4, fibrillation +4, fasiculation +4, MUAP: giant amplitude, widened duration, polyphasic phase, decreased recruitment, incomplete IP..
M. flexor carpi radialis	PSW +4, fibrillation +4, MUAP: giant amplitude, widened duration, polyphasic phase, decreased recruitment, incomplete IP.
M. masseter	PSW +4, fibrillation +4, MUAP: giant amplitude, widened duration, polyphasic phase, decreased recruitment, incomplete IP.
M. 10th Th paraspinal	Normal

CMAP: compound muscle action potential; SNAP: sensory nerve action potential;EMG: electromyography

We were ordering renal function test for feasibility of contrast, cervical MRI with contrast and ENMG examination. Lab test for renal function is within normal limits and there is no contraindication to the use of contrast. Results from EMG on September 19, 2017 showed a lesion in motor neurons with suspicion of ALS (Table 1).

On cervical MRI examination with contrast on September 25, 2017, we found syringomyela as high as 2nd cervical until the encoded area ended in level of 6th thoracic, no solid intramedullary or extramedullary lesions were observed along the area, and intervertebral discs of cervical did not appear abnormal (Figure 1).



Picture 1: Cervical MRI with contrast

The patient diagnosed with ALS with asymptomatic syringomyelia and given supportive therapy with 500 mcg of mecobalamin every 8 hours intraoral. She consulted the medical rehabilitation department and provided with outpatient education for long-term care.

3. Discussion

ALS is a neurodegenerative disease that attacks motor neurons which mixed of Upper Motor Neuron with Lower Motor Neuron lesion occured [1]. The incidence of ALS in 1990 is reported to range from 1.5 to 2.7 per 100,000 population per year (average 1.89 per 100,000/year) in Europe and North America. The incidence rate of ALS is higher in men than women, with an overall ratio is 1.5:1. Based on geographic location, the prevalence of ALS disease in the Western Pacific including Guam, Mariana Islands, Honsu Island and southern West New Guinea is reported 50-100 times higher than other areas [4,8]. Onset of ALS from adolescence to age 80 years, but the peak was at the age of 55-75 years. The onset of the sporadic average age of ALS (SALS) was 65 years, the mean age of ALS (FALS) familial age was 46 years [9].

In this case, the patient is a 41-year-old female. The symptoms initially felt in 2009 when the patient was 33 years old, earlier than the average age of onset of sporadic ALS (65 years). ALS cases in this patient found to be more likely to sporadic ALS compared to familial, because there were no family members of her either from the father or from mother who had the same history. Initial symptoms that appear were weaknesses in upper extremities. This symptom is progressive, chronic and accompanied by shrinking of the upper extremities muscles. The early examination in 2009 found no structural abnormalities in cervical MRI.

ALS diagnosis are based on clinical findings, in which a person allegedly suffering from ALS will generally complain

of gradual or progressive motor function loss in one or more parts of the body, without any sensory impairment and no apparent cause. The progression of ALS occurs slowly so that early symptoms are often overlooked and considered as an aging process [1,2,10].

The body part affected by the initial symptoms of ALS depends on the first affected muscle. Early symptoms frequently affect one leg thus the patient has difficulty walking or running and more likely to stumble than before. Other symptom may be that the patient has difficulty in performing simple movements requiring hand skill, such as buttoning a shirt, writing, or inserting and turning a key in a keyhole. Patient with bulbar symptom usually has problems in speaking (dysarthria) or decreased voice volume, dysphagia, aspiration or choking at meals. This occurs because of the involvement of the cranial nerve nuclei VII, IX, X, XI and XII. In 75-80% of patients, symptoms begin with extremity involvement while 20-25% of patients come with bulbar symptoms. Some patients may also experience cognitive changes, where cognitive dysfunction is experienced by 20-50% of ALS patients and 3-15% develops into dementia (Frontotemporal Lobar Degeneration / FTLD). Cognitive changes can be characterized by changes in personality, irritability, and impaired executive function. In this case, early symptoms of weakness in upper extremities are found. The complaints of weakness are progressive, chronic and accompanied by a shrinking of the upper extremities muscles. However, no structural abnormalities in cervical MRI that conducted in 2009.

The diagnosis of ALS is confirmed by the presence of weakness of UMN and LMN at the same time in the extremities (muscle weakness, muscle atrophy, muscle fasciculation, combined with hypereflex) [1]. The El Escorial Criteria was developed in 1994 by the World Federation of Neurology for research purposes and clinical trials, and then revised to Airlie House criteria in 1998 by incorporating laboratory test criteria. In 2008, the Awaji-Shima Criteria was introduced with the involvement of neurophysiology in diagnostic categorization, thus its use enhanced the diagnostic sensitivity without increasing the positive false [13]. El Escorial and Airlie House criteria are based on the degree of certainty of diagnosis. These are based on a clinical assessment requiring the identification of UMN and LMN in the same topographical area of anatomy in the brainstem, neck, thorax or lumbosacral spinal cord. The exact diagnosis of ALS requires the following signs: identification of clinical, electrophysiological or neuropathological degeneration of LMN; as well as evidence of UMN degeneration in clinical and progression from motor syndrome in a region and/or other region. Diagnosis should also be confirmed by electrophysiological evidence or neuroimaging if symptoms encountered are not caused by other diseases [13,14].

In this case, physical examination showed a mixed lesion of LMN and UMN. If we follow the El Escorial criteria, we find two areas of involvement in the patient that is the upper and lower extremities. Both arms show atrophy and flaccid paresis - an LMN lesion, while there was a spastic paresis showing a UMN lesion in the two inferior extremities. The presence of combination of UMN and LMN lesions in the two regions which the symptoms of UMN appears more rostral than LMN symptoms can already be classified into probable groups of ALS according to El Escorial and World Federation of Neurology (WFN) criteria. There was no clinical evidence of sensory disturbance in this patient so that more diagnosis leads to a motor neuron disease. In EMG examination, an active denervation mark is found in the form of fibrillation and positive sharp wave, and chronic denervation marks of MUAP with giant amplitude at m. right biceps, m. flexor carpi radialis, m. masseter. This depiction is typical of ALS. On cervical MRI examination with contrast, we found abnormalities of syringomyelia as high as the corpus of the 2nd cervical vertebrae until the encoded area ends in the level of 6th thoracic vertebrae corpus. This corresponds to the differential diagnosis of ALS which is syringomyelia but is not accompanied by clinical symptoms of syringomyelia (motor disturbance and sensory) so that the patient is diagnosed with ALS accompanied by an asymptomatic syringomyelia.

Management of ALS is support to patients, palliative and multidisciplinary. Studies recommended administration of riluzole (a glutamate antagonist) 50 mg twice daily, which administered 100 milligrams of oral riluzole daily after 18 months may extended the life expectancy of ALS patients by about three months [15,16].

Symptomatic therapy to overcome spasticity can be given is baclofen or diazepam, and to overcome excessive saliva production may be given trihexyphenidyl, amitriptyline, or botulinum toxin type B injection and radiation therapy [16,17]. The nutritional status of ALS sufferers also needs to be evaluated, given the frequent occurrence of dysphagia and hypermetabolic so that nutritional management includes diet, swallowing strategy, gastrostomy tube placement, vitamin and mineral supplementation [18,19].

Patient treatment in this case is supportive with intraoral mecobalamin. Therapy with riluzole is not currently available in Indonesia so it cannot be given. There were no further symptoms of an ALS found such as dysphagia and weight loss as the consequence of dysphagia, therefore other supportive measures had not been given yet. We consult patient to medical rehabilitation department which she accepts physiotherapy to train the impaired extremities.

ALS is a fatal disease with an average 3–5 years of life expectancy since the onset of clinical weaknesses. About 4–30% of patients with ALS can survive about 5 years and 4% survive for more than 10 years. Long-term survival is often associated with younger age at onset, in men, and in the affected region. ALS exposed to extremities is said to last 3-5 years while bulbar for 2-3 years [20,21].

Patient educated that treatment will last a lifetime and holistic treatment is required. The good prognostic indicator in this case is onset of less than 50 years, and long intervals from the first symptoms appear to be diagnosed (± 3 years), whereas a poor prognostic indicator is patient also accompanied by syringomyelia although it has not clearly manifested clinically at present nor the symptoms overlap with ALS

itself. Since the therapy may still limited, her prognosis is poor.

4. Conclusion

ALS clinically consists of motor movement disorders, which show signs of UMN and LMN paralysis. ALS management is in the form of support to patients, palliative, and multidisciplinary. The average life expectancy is 3-5 years since the onset of clinical weaknesses.

5. Conflicts of Interest

The authors declare no conflict of interest or any financial support. We had consent letter to the patient to publish this case.

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