Sporadic amyotrophic lateral sclerosis: a case report and literature review

Yin Hang¹, Dong Bo²*, Yang Feng², Yuan Puwei², Kang Wulin², Liu Deyu²

¹The First School of Clinical Medicine, Shaanxi University of Traditional Chinese Medicine, Xianyang, Shaanxi, 712046, China
²Department of Osteopathology, Affiliated Hospital of Shaanxi University of Traditional Chinese Medicine, Xianyang 712000, China
*Corresponding author

Abstract: To summarize the clinical features of a case of sporadic amyotrophic lateral sclerosis (SALS), improve the ability of early diagnosis and differential diagnosis, and summarize the treatment experience. [Methods] The clinical data of a case of sporadic amyotrophic lateral sclerosis (SALS) in our hospital were analyzed, and the diagnosis and treatment experience was summarized according to its history, clinical manifestations, and imaging and electromyogram manifestations, combined with relevant literature reports at home and abroad. [Results] After comprehensive treatment, the patient was in stable condition and discharged without any progress. After discharge, the patient continued to follow up. Although the systemic symptoms progressed, the degree was slight. [Conclusion] The etiology and pathogenesis of SALS are very complex, and the disease continues to progress. At present, there is no particularly effective treatment for this kind of disease. Comprehensive treatment can be adopted to control the development of the disease, so as to better improve the quality of life and survival rate of patients.

Keywords: Amyotrophic lateral sclerosis; Loose hair; Case analysis; combined treatment

Amyotrophic lateral sclerosis (ALS) is the most common type of motor neuron disease. It is a progressive degenerative disease of the nervous system that can involve upper and lower motor neurons. It can be divided into sporadic ALS (sporadic amyotrophic lateral sclerosis, SALS) and familial ALS (familial amyotrophic lateral sclerosis, FALS) [1]. Clinically, it is mainly characterized by progressive muscle weakness, atrophy Muscle bundle tremor and pyramidal tract damage may eventually lead to death due to dysphagia and respiratory failure [2]. Because the etiology and pathogenesis of the disease are not very clear, and the disease progresses rapidly, which seriously affects the quality of daily life and survival rate of the patient, this paper analyzes the clinical data of this patient, and discusses the etiology and pathogenesis, diagnosis, differential diagnosis, treatment and nursing of the disease in combination with relevant literature at home and abroad, so as to improve the understanding of the disease and summarize the treatment experience for clinical reference.

1. Clinical data

The patient, a 39 year old female, developed hoarseness, speech congestion and unclear enunciation in March, 2020. Since then, she felt that her left hand was weak, inflexible, inflexible, weak lower limbs, muscle atrophy of her left limbs, and obvious atrophy of her left thenar and palm muscles. She performed speech rehabilitation training at home, but her symptoms were not significantly improved, and showed progressive aggravation. She had no family history. Physical examination: dysarthria, lingual muscle atrophy, lingual muscle fibrillation, cervical flexor muscle strength grade 4, left triceps brachii muscle strength grade 4+, left abductor pollicis brevis muscle grade 3, left index finger extensor muscle grade 3, left total extensor muscle grade 4, left first interosseous muscle, thenar and hypothenar atrophy. Routine nerve electrophysiological examination: routine nerve conduction test (NCS): the amplitude of left ulnar motor nerve is suspiciously lower than that of the contralateral side, and no obvious abnormality is found in the remaining limb nerves. Imaging examination: when the brain and cervical vertebra were m, there were muscle beats, nasal congestion and drinking water choking cough. The patient was diagnosed as sporadic amyotrophic lateral sclerosis.
2. Treatment

On the basis of multi-disciplinary comprehensive treatment (such as respiratory support, nutritional support and symptomatic treatment), traditional Chinese medicine mainly adopts Jianpi Yifei formula plus or minus oral, massage and chiropractic therapy, warm moxibustion of Du Meridian and Electroacupuncture of Jiaji point, combined with oral drug riluzole tablets and intravenous injection edaravone, to maintain the stability of patients' exercise and respiratory function, improve muscle strength, delay the development of disease and improve the prognosis of the disease. After treatment, the patient's condition was stable. After discharge, follow-up was continued. Although the systemic symptoms were progressing, the degree was relatively light. Continue rehabilitation exercise and massage therapy to delay the disease progress and improve the patient's quality of life and survival rate.

3. Discussion

Sporadic amyotrophic lateral sclerosis (ALS) is a degenerative disease of the nervous system that progressively aggravates the upper and lower motor neurons. It is mainly caused by the involvement of spinal cord anterior horn cells, brain vertebral body cells, brain stem motor nucleus and pyramidal tract. It tends to occur in people after the age of 50, and men are more than women [3]. The incidence rate and prevalence are low, most of them progress slowly, and the onset is hidden. The first symptoms are muscle weakness and muscular atrophy at the distal end of unilateral upper limb. In this case, the onset of the disease in the medulla oblongata is relatively rare. The main manifestations are hoarseness, speech congestion, lingual muscle atrophy and fibrillation. The progress of the disease affects the limb muscles.

The etiology and pathogenesis of the disease are not very clear, which may be related to gene mutation, oxidative stress response, viral immunity, heavy metal poisoning, malignant tumor, trauma and physical labor [4]. A foreign study on genetic factors of SALS found that the polymorphisms of rs2619566, rs10260404 and rs79609816 may be related to the disease and may play a role in the pathogenesis of the disease. SOD1 gene mutation is also related to this, while tardbp mutation is rare in the disease [5]. Researchers also found that changes in the function of nuclear porin (nups) and nuclear pore complex (NPC) are the factors leading to the pathogenesis of genetic forms of neurodegenerative diseases [6]. At present, the genetic basis of the disease is very complex. So far, more than 20 genes have been found to be related to the disease. The presence of multiple gene mutations will increase the susceptibility to the disease, and can further complicate it by interacting with environmental factors [7].

The diagnosis of SALS mainly relies on clinical symptoms and electrophysiological diagnosis, and excludes other diseases with similar clinical symptoms and electrophysiological characteristics. Electromyography, as a supplement to clinical examination, is of great significance for the early diagnosis and differential diagnosis of diseases. Ct/mri of the head and spinal cord in neuroimaging examination can provide diagnostic basis for the differential diagnosis of similar diseases. Clinically, it is mainly related to cervical spondylotic myelopathy, multifocal motor neuron disease Sjogren's syndrome, spinal cord tumor, etc. can be differentiated from cervical spondylotic myelopathy by electromyography of sternocleidomastoid muscle. The positive rate of electromyography of the former is much higher than that of the latter [8]. Muscle biopsy is also an important auxiliary means for the diagnosis and differential diagnosis of this disease. At present, there is no effective biomarker for the diagnosis of this disease. The study on miRNA expression in cerebrospinal fluid of patients with SALS using microRNA sequencing found that 11 kinds of differentially expressed miRNAs were detected in cerebrospinal fluid of patients with SALS. Detecting miRNA biomarkers in CSF of patients with SALS can better understand the physiology and pathology of the disease, and also carry out personalized new therapies [9]. The parameter levels of neurofilament light chain and heavy chain in cerebrospinal fluid and serum of ALS patients can be used for the diagnosis of ALS patients, as well as for the differential diagnosis of ALS. Some studies have compared and analyzed the neurofilament heavy chain and light chain in serum and cerebrospinal fluid of ALS and other nervous systems, and found that CSF NFL and NFH and serum NFL are also suitable for the differential diagnosis of ALS, while the efficacy of serum NFH is slightly lower [10].

To sum up, as the etiology and pathogenesis of the disease are not clear, although there are some drug treatment methods at present, their curative effects are limited, mainly focusing on delaying the progress of the disease. Through this case, combined with relevant literature at home and abroad, it is very important to summarize the early diagnosis of the disease, advocate early discovery and early treatment, and the treatment should still be based on multidisciplinary comprehensive treatment.
combined with traditional Chinese medicine and Western medicine, and pay attention to patient care, in order to better improve the quality of life of patients and prolong the life cycle.

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