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# *Difficulties in diagnosis of amyotrophic lateral sclerosis in patient with bulbar symptoms – a case report*

ALS is a disease of unknown cause and pathogenesis. It is called "maladie de Charcot" or "motor neuron disease". It is a progressive and incurable and degenerating disease of the neural system. As for its histopathology, it is labelled as the degeneration of both upper and lower motor neurons. It is characterized by the co-existence of: atrophy and fasciculation of muscles (as a result of destruction of spinal cord front corner cells); spastic syndrome with pyramidal symptoms (as a result of side strings of spinal cord seizure) (1–3).

ALS occurs worldwide with similar frequency, with approximately 50 cases per 1 million people and it concerns male patients more often (2). The beginning of the disease usually happens among patients between 50 and 70 years of age. There are, however, cases of falling ill among under 20-year-olds (4). The etiology of ALS remains unexplained. Among the hypotheses of its etiology there is one concerning exogenetic, toxic and self-immunological factors (5).

Most frequently, the disease starts with weakness and muscular atrophy with noticeable fascicular trembling. At its initial stage one can notice the atrophy of short muscles of one hand – the hypothenar and the thenar. In the course of the disease the symptoms get stronger and more frequent and they move to the muscles of the second limb. The bulbar or the pseudobulbar symptoms – blurred or inarticulate speech, dysarthria or swallowing problems, excessive salivation – happen usually in the further course of the disease but in 25% cases they might be observed at its beginning. These symptoms are often accompanied by emotional disorders such as pathological uncontrolled laughter or cry and, at the final stage of the disease, swallowing and breathing disorders (2, 6). To diagnose the disease, the obligatory criteria of World Federation of Neurology Research Group of Neuromuscular Disease must be observed (3).

## CASE REPORT

A 56-year-old female patient, no previous history of the disease was admitted into MSWiA Hospital in Lublin (Neurological Department). The symptoms were speech disorders (for 6 months until the admission) and right hand muscular strength weakening. When examined, the patient showed atrophy of tongue muscles (the right side), and the tongue swerved to the right, fibrillar trembling over the tongue, atrophy of interosseous muscles of the right hand, especially of the thenar, the weaker handshake, more lively deep reactions in the right upper limb and the knee reaction in the right lower limb, no Babinsky symptom. These are the results of the examination:

Table 1. The results of the examination

| Examination type | Results |
|---|---|
| Electromyography | the tongue muscle showed numerous multiphasic units – neurogenetic features, in the record of the first interosseous muscle, and of the deltoid muscle there are numerous signs of peripheral neuron on the level of spinal cord front corner motor cells |
| The brain MRI examination | on both sides of the white matter round the cerebral ventricles and over the cranial vault there are numerous slight, partly blurring focuses of demyelinizational features; similar focuses can be observed in the bridge on the left |
| The spinal cord MRI | on C5/C6 level the features of the central-right sided rupture with the pressure on the spinal cord and the right nerve root can be observed |
| Cerebral-spinal fluid | no changes |
| Laryngological consultation | limited activity of the forward movements of the tongue, correct palate tension. The larynx is wide, the vocal ligaments act in the right way |

## DISCUSSION

ALS diagnosis causes a number of problems due to little frequency of the disease. Because of that the time of the disease from the first symptoms occurrence until the final diagnosis estimation may last for about a year. The basic examination, the result of which may help to diagnose ALS is the interview, the examination of reflexes (in ALS they are exaggerated), the estimation of the strength and the degree of muscles tension.

The disease's symptoms may sometimes be the cause of diagnostic errors. The unilateral weakness of the hand might be the result of the brachial plexus, medial and elbow nerve, cervical spondylosis. The presence of cervical spondylosis in MRI examination is frequently observed among the patients without clinical symptoms. The wrong diagnoses lead to operations which result in no improvement (7).

The first symptoms of ALS should be differentiated with the inclusive muscle inflammation. The dominant symptoms are of lower motor neuron and they are limited to the muscles controlled by certain nerves or features of single circular nerves damage, and the EMG examination shows no flow (2, 7). Muscle weakening, which imitates lower motor neuron damage and bulbar symptoms might occur in myasthenia, but in this case there should be particular muscle-bound, the reaction to acetylocholinesterasis blocks and typical lesions in EMG (3). Distal paresis and muscle atrophy should be differentiated with the multifocused motor neuropathy, which shows symptoms limited to the muscles controlled by certain nerves, and the features of single circuital nerves damages (8).

Bulbar symptoms might occur in small vessels diseases (lacunar infarctions), in syringobulbia. In this disease there are splitting disorders of facial esthesia, Homer syndrome (7). Apart from that, the bulbar form of ALS should be differentiated from cranial base tumors.

Another disease which should be differentiated with ALS is Kennedy syndrome. That disease is inherited with X chromosome and displays perennial bulbous syndrome with features of lower motor neuron damage. Hormonal disorders, gynaecomastia and impotence are frequent (9). Clumsy hands moving, monotonous speech imitating bulbous might be mistaken with the early stage of Parkinson disease. Multifocal changes might suggest multiple sclerosis, but this disease might be excluded according to the criteria of MS identification by McDonald (10).

In the case of the above mentioned patient, the characteristic clinical form of the disease which was confirmed by the EMG examination made us diagnose ALS. MRI confirmed features of brain damage lead to the additional diagnosis of multifocal demyelinisational damage of the central nervous system.

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## SUMMARY

The aim of this study was to present the difficulties in diagnosing the amyotrophic lateral sclerosis case of a 56-year-old female patient with ALS, whose disease started from non-standard symptoms of bulbar syndrome. It usually starts from muscular weakness and atrophy and only in 25% of cases it starts from bulbar symptoms, which significantly delays proper diagnosis. The case history and other medical documents of the patient referred to the neurological department of MSWiA Hospital in Lublin in July 2006. The patient was admitted with bulbar syndrome as presented. The symptoms of the damage of other neural system structures' deterioration appeared only in the further course of the disease, which made the diagnosis more difficult. The proper diagnosis was stated only after the many-months' observation and repeated additional examinations. This case shows how important is the knowledge of the disease's symptoms, which lets its early diagnosis statement.

### Trudności w diagnostyce stwardnienia bocznego zanikowego u pacjentki z zespołem opuszkowym – opis przypadku

Celem pracy było przedstawienie trudności w ustaleniu rozpoznania stwardnienia bocznego zanikowego u 56-letniej kobiety, u której choroba rozpoczęła się od nietypowych objawów zespołu opuszkowego. Choroba zazwyczaj zaczyna się od osłabienia i zaniku mięśni kończyn, a jedynie u około 25% chorych rozpoczyna się od objawów opuszkowych, co znacznie opóźnia postawienie prawidłowego rozpoznania. W pracy posłużono się dokumentacją medyczną i obserwacją pacjentki, hospitalizowanej w Oddziale Neurologii ZOZ MSWiA w Lublinie w lipcu 2006 r. Chora przyjęta została do szpitala z postępującym zespołem opuszkowym. Objawy uszkodzenia innych struktur

układu nerwowego pojawiły się w późnym okresie choroby, co znacznie utrudniało postawienie diagnozy. Ustalenie właściwego rozpoznania umożliwiła dopiero wielomiesięczna obserwacja i powtarzane badania dodatkowe. Przypadek ten pokazuje, jak ważna jest znajomość wczesnych objawów choroby, pozwalająca na szybkie postawienie właściwego rozpoznania.