AN UNUSUAL CASE OF HEADACHE AND DYSPHAGIA: ALS DEMENTIA COMPLEX. CASE REPORT

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[Un raro caso di cefalea e disfagia: sindrome SLA e Dementia fronto temporale]

ABSTRACT

Herein we describe a case of typical association of ALS ad dementia, which will help us to enrich the case history and remind us to bear this association in mind in clinical practice

Key words: ALS, dementia, FTD, amyotrophic lateral sclerosis, dysphagia.

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Introduction

Amyotrophic lateral sclerosis (ALS) is a neurodegenerative disease characterized by progressive and combined involvement of the first and second motor neuron that frequently leads to death within a few years of onset.

The association between ALS and dementia, in particular frontotemporal dementia, has shown an incidence over time such that their association has long been considered not to be random, but pathogenically connected, due to the possible concurrence, as it was believed, of a common causal factor at the root of the two epiphenomena, or to the existence of two distinct factors that, for their contiguity, could be contextually involved. In fact, the association between the two pathological conditions, reported for decades in case reports as a possible expression of clinical randomness, has gained over the years the dignity of a causal rather than random association, as proven by the increasing number of cases described.

This association, though reported already a century ago, has been recognized a possible distinct clinical identity, in recent times, by the authors of a famous article in 2003[1]. In confirmation of the earlier suspicion, the conclusion of a multi-center research, coordinated by the Mayo Clinic in Florida, was published in 2006 and it showed a genetic alteration at the root of the association (neuron)[2], which confirms what till then had only been suspected, with the documentation of a GGGGCC esanucleotidic expansion on chromosome 9 of gene C9ORF72, which is closely linked with most of the associations of fronto-temporal dementia and ALS. This result was recently confirmed by an Italian multicenter research in sporadic form[3].

In this context, the authors describe a rare clinical case of ALS combined with Dementia

Case report

F.A. a 44-year-old woman, with no prior neurological pathology, with 8 years of schooling and

no previous clinical history of pathologies involving neurology, internal medicine, surgery, or trauma was referred to our attention in October 2011 with the clinical onset of headache and memory deficits with gradual onset. Dysarthria and dysphagia were also associated.

MRI of the brain and cervical spine were normal, thus excluding a vascular[4] or demyelinating[5] etiology. Esophageal manometry of the pharyngo-oesophageal tract did not show anything abnormal[6]. The electromyographic examination showed signs of denervation in the muscles pertaining to the ulnar and median nerves, bilaterally, more to the right than left. Sporadic fasciculations involving the tongue.

The condition of the patient worsened considerably in the next three months, especially with regard to speech and swallowing; after two months, dysphagia developed for both solids and liquids; significant clinical signs and manifestations of cognitive impairment characterized by lack of interest in personal care, behavioral disorders with stereotypical attitudes, bulimia, and disinhibition.

In June 2012, the above symptoms worsened with the extension of hypotrophy and fasciculations also to the lower limbs, the peroneal region, first right and then left.

Osteotendineal hyperreflexia was present in the 4 limbs; no signs and symptoms of a sensory nature.

The lower limbs developed bilateral foot clonus and bilateral presence of Babinski[7,8]. Cerebellar tests were normal. CSF examination and EEG were negative.

Based on the unremarkable neuroradiology exams and neuropsychological and neurophysiological findings, in addition to clinical signs and symptoms, we made a diagnosis of amyotrophic lateral sclerosis with frontotemporal dementia.

Discussion

The association "Amyotrophic Lateral Sclerosis and Dementia" is a frequent clinical condition in the Mariana Islands (Pacific), and in some areas of Japan.

In Europe, one of the first reports of this syndrome dates back to Zegler[9]. Mitsuyama[10,11] studied the clinical association "ALS-dementia" as an independent nosological complex and Hudson[12] described "familial" and "sporadic" forms of the "ALS-Dementia" complex at times associated with Parkinson's disease. Dementia was clinically equated with Pick's disease and occurred before ALS.

The clinical case described by us is similar to sporadic forms of the association of ALS and dementia, although the age of onset (44 years) is lower than the 53 years reported by Hudson in the literature[12].

Esophageal manometric examination is necessary in the early stages of the disease when it is necessary to carry out an accurate functional assessment of pharyngo-esophageal coordination, since it has been shown that in severe cases some improvement is possible with esophageal myotomy[13]. It is still necessary to always be alert because dysphagia is a very enigmatic symptom for the variety of etiologies and the difficulty of identifying them[14].

Bulbar impairment and the clinical signs that can be correlated with a pathology of the 1st and 2nd motor neuron, and the data of the electromyographic examination of the muscles examined (presence of fibrillation potentials, fasciculation, with normal motor and sensory conduction velocity) have shown, together with absolute negativity to other investigations, a clinical picture that correlates with the diagnosis of ALS with frontotemporal dementia[15].

A neuropsychological examination had shown a significant deterioration of higher cortical functions, correlated with clinical manifestations of frontotemporal dementia.

It seemed interesting to report this case of association of ALS with dementia, typical for its manifestation, which helps to enhance the case history and allows us to take such cases into account in clinical practice, as it is no longer exceptional[16].

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