Polymyositis Masquerading as Motor Neuron Disease

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Background: Several conditions have been reported to mimic motor neuron disease (MND), and misdiagnosis remains a common clinical problem.

Objective: To report a case of bulbar-onset polymyositis where the initial clinical presentation was suggestive of MND.

Case Description: A 73-year-old woman was admitted for investigation of acute-onset dysphagia without dysarthria. Examination revealed nasal dysphonia and severe oropharyngeal weakness. Subtle upper-limb weakness, brisk tendon reflexes, and fasciculations in the right deltoid muscle were also demonstrated. A clinical diagnosis of MND was entertained. The serum creatine kinase value was within the reference range. Findings from electromyographic studies, however, were suggestive of a myopathic rather than a neurodegenerative process, and a muscle biopsy specimen was diagnostic of polymyositis. The dysphagia rapidly resolved upon treatment with corticosteroids and azathioprine.

Conclusions: Bulbar-onset polymyositis may mimic MND, particularly in the absence of inflammatory markers or elevated muscle enzyme levels. Caution should be exercised in the clinical diagnosis of bulbar dysfunction, and further investigations such as electromyography and muscle biopsy are indicated to confirm the diagnosis.

Arch Neurol. 2003;60:1001-1003

A myotrophic lateral sclerosis (ALS) is characterized by progressive degeneration of upper and lower motor neurons, and criteria for diagnosis are based on the original El Escorial1 or revised Airlie House2 criteria. Bulbar-onset disease is a common finding, particularly with advancing age.3 Polymyositis is an idiopathic inflammatory muscle disorder that is also defined by set diagnostic criteria,4,5 and dysphagia at onset is a recognized feature.6,7 Despite these distinctions, atypical cases can be a source of diagnostic confusion. Conditions such as multifocal motor neuropathy, Kennedy disease, and inclusion body myositis8,9 have all been misdiagnosed as ALS. We report an atypical case of dysphagia-onset polymyositis in which the initial clinical presentation was suggestive of ALS. We discuss the evaluation and differential diagnosis of acute-onset dysphagia of neuromuscular origin.

CASE DESCRIPTION

A 75-year-old woman with a history of rheumatoid arthritis presented with acute-onset dysphagia for liquids and solids of 24 hours’ duration. She also complained of nasal regurgitation and dysphonia. Clinical examination revealed nasal dysphonia and severe oropharyngeal weakness. Subtle upper-limb weakness, brisk tendon reflexes, and fasciculations in the right deltoid muscle were also demonstrated. A clinical diagnosis of MND was entertained. The serum creatine kinase value was within the reference range. Findings from electromyographic studies, however, were suggestive of a myopathic rather than a neurodegenerative process, and a muscle biopsy specimen was diagnostic of polymyositis. The dysphagia rapidly resolved upon treatment with corticosteroids and azathioprine.

Arch Neurol. 2003;60:1001-1003

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ness with pooling of secretions and silent aspiration. There was no evidence of cricopharyngeal obstruction.

Nerve conduction studies showed absence of sural sensory nerve action potential and mild loss of median compound motor action potential amplitude. A concentric needle electromyograph of the right extremities and genioglossus showed widespread fibrillation potentials, sparing the tongue. There were no fasciculations. There was a mild excess of complex short-duration polyphasic units with areas of early recruitment. These findings favored a diagnosis of inflammatory myopathy rather than ALS. A biopsy specimen of the right quadriceps revealed type II fiber atrophy. A second biopsy specimen, from the clinically weak left triceps, showed evidence of active ongoing myositis, with focally dense endomysial infiltrate consistent with polymyositis (Figure). In view of the clinical findings, magnetic resonance imaging of the cervical spine was also performed and revealed significant spondylosis, with disc space narrowing in the C3 through C5 region.

A percutaneous enterogastrostomy was temporarily inserted, and the patient began taking high-dose corticosteroids and azathioprine. The response was rapid and sustained. The patient recovered her ability to swallow and, 1 year later, had no further complaints of dysphagia.

A muscle biopsy specimen from the left triceps shows the characteristic endomysial inflammation of polymyositis.

A differential diagnosis for acute dysphagia of neuromuscular origin is given in the Table. Acute stroke, affecting the cortex or brainstem, is a common cause, but it is usually associated with other clinical manifestations. The Arnold-Chiari type I malformation may present as isolated progressive dysphagia before other brainstem signs become evident. Disorders of the anterior horn cell, such as ALS, are frequently associated with swallowing abnormalities, and the dysphagia may be of rapid onset, as in our patient. Guillain-Barré syndrome, in particular the Miller-Fischer variant, may present with varied involvement of the lower cranial nerves and a descending pattern of weakness. Disorders of the neuromuscular junction, such as myasthenia gravis, are relatively common causes of dysphagia and, indeed, may coexist with polymyositis. Our patient underwent an edrophonium chloride test, which was negative, and no acetylcholine receptor antibodies were detected in serum. Botulism can also cause dysphagia. However, the absence of other findings of bulbar dysfunction and dysautonomia, and the preservation of deep tendon reflexes, ruled this out as a diagnosis in this case. Dysphagia in inclusion body myositis is a frequent finding and may precede limb muscle weakness, in some cases by as long as 7 years.

Given the patient's history and the clinical findings of upper and lower motor neuron degeneration (in part attributable to the coexistent cervical spondylosis), a diagnosis of ALS was originally considered. The normal laboratory creatine kinase assay seemed to support this. The serum creatine kinase level may, however, be within the reference range in polymyositis; causes include chronic disease with muscle atrophy, prior treatment with corticosteroids, and low baseline muscle mass. It has also been reported that patients with connective tissue diseases (such as rheumatoid arthritis) tend to have low creatine kinase values. As a result, even though the value lies within the reference range, it may be falsely high for these patients. Thus a “normal” creatine kinase value may be misleading and should not lead one to disregard an inflammatory cause. Rather, continuing investigations are essential when there is clinical suspicion of a neuromuscular condition.

Although electrodiagnostic studies cannot conclusively establish a diagnosis of polymyositis, they proved though dysphagia is frequently reported in polymyositis, it is usually part of a more general clinical picture. Its occurrence in isolation is rare, and such a rapid temporal onset, as occurred in our patient, is distinctly unusual.

A muscle biopsy specimen from the left triceps shows the characteristic endomysial inflammation of polymyositis.
decisive in this case, by focusing our attention on a myopathic rather than a neurodegenerative process. Recruitment patterns were not markedly myopathic, but the presence of diffuse and intense fibrillations, without fasciculations, in the absence of frankly neurogenic (decreased) recruitment, was in favor of myonecrosis rather than motor neuron disease. This interpretation was confirmed by the finding of inflammatory changes on the muscle biopsy specimen. Polymyositis may be a patchy condition, and reliance on the creatine kinase assay and the first (quadriceps) biopsy specimen alone could have led to delayed diagnosis or misdiagnosis. Instead, prompted by clinical and electromyographic findings, a muscle biopsy specimen from a clinically affected area revealed the diagnosis. The advances in ancillary investigations, such as magnetic resonance imaging of muscle, and the availability of laboratory tests for myositis-specific antibodies have significantly augmented the diagnostic modalities available for polymyositis. Nevertheless, careful and complete investigation with existing methods can diagnose even the most atypical examples of this treatable condition. The importance of early diagnosis is underscored by the rapid response, in most cases, to steroid therapy, reducing the risk of serious complications, such as aspiration.

This case illustrates that bulbar-onset polymyositis may mimic ALS, particularly in the presence of normal serum creatine kinase levels, and should be considered in the differential diagnosis of acute-onset dysphagia. Caution should be exercised in the clinical diagnosis of bulbar dysfunction, and further investigations, including magnetic resonance imaging, neurophysiological examination, and muscle biopsy, are essential to confirm (or, in this case, refute) the initial clinical impression.

Accepted for publication November 26, 2002.

Author contributions: Study concept and design (Drs Ryan, Nor, Foley-Nolan, El-Rafie, and Hardiman); acquisition of data (Drs Costigan and Farrell); analysis and interpretation of data (Drs Ryan, Farrell, and Hardiman); drafting of the manuscript (Drs Ryan and Nor); critical revision of the manuscript for important intellectual content (Drs Ryan, Costigan, Foley-Nolan, El-Rafie, and Hardiman); administrative, technical, and material support (Drs Ryan and Nor); study supervision (Drs Costigan, Foley-Nolan, El-Rafie, Farrell, and Hardiman).

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