Distal Myopathies
Case Studies

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KEYWORDS

• Distal • Myopathy • Weakness • Myofibrillar

KEY POINTS

- Distal weakness is an important presentation to the neurology clinics and the differential diagnosis is wide.
- Diagnostic process requires differentiation of myopathic causes of distal weakness from nonmyopathic causes such as neuropathies, myasthenia gravis, and motor neuron diseases.
- The most common causes of myopathic muscle weakness are inclusion body myositis, myotonic dystrophy, and facioscapulohumeral muscular dystrophy.
- Myofibrillar myopathy and distal muscular dystrophy are rare causes of distal myopathy and their differentiation depends on the clinical, pathologic, and genetic profiles.

Video content accompanies this article at http://www.neurologic.theclinics.com.

CASE 1: DYSPHAGIA AND HANDS WEAKNESS (SEE FIG. 1 FOR VIDEO 1)

A 67-year-old woman presented with a 5-year history of gait imbalance and dysphagia. Examination is shown. Creatine kinase (CK) level was 800 U/L and electromyogram (EMG) revealed mixed short and long duration potentials in the proximal and distal arm and leg muscles bilaterally. Discontinuation of atorvastatin and treatment with oral steroids reduced the CK level to 300 U/L and led to improvement of the stamina for a couple of months. No objective changes in the muscle strength were noted.

Regarding the case

1. CK reduction with steroids argues against inclusion body myositis (IBM)
2. The pathologic condition is due to statins
3. Statins may have worsened weakness produced by IBM
4. She is a good candidate for intravenous gamma globulin (IVIG)
5. The pattern of weakness is not typical for IBM

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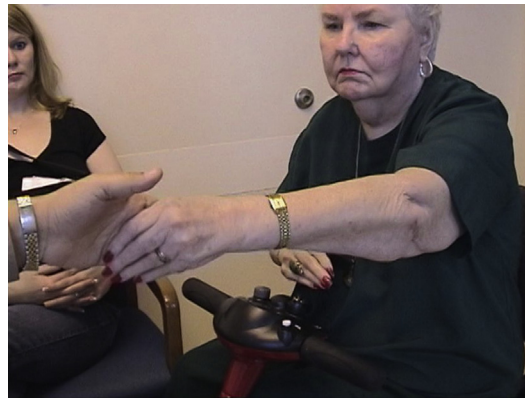

Fig. 1. Dysphagia and hands weakness.

IBM

- The pattern of weakness in this case is very typical for IBM, which should be highly suspected just after shaking hand with the patient (fish mouth handshake appearance due to weakness of finger flexors).

- The practice of adjusting the dose of prednisone based on the CK level is not recommended because steroids stabilize the cell membrane and reduce the CK level regardless of the cause.
  - Strength measurement and functional scales are the most valid outcome measures in monitoring progression and recovery of myopathies.

- Statins can cause different kinds of myopathies, including toxic myopathy and autoimmune necrotizing myopathy, with positive antibodies against 3-hydroxy-3-methylglutaryl coenzyme A (HMG-CoA) reductase.
  - Also, statins may unmask an underlying metabolic or mitochondrial myopathy because 30% of statin-induced myopathies showed evidence of an underlying metabolic defect such as carnitine palmitoyl transferase (CPT)-2, myoadenylate deaminase (MAD), or phosphorylase deficiencies.
  - Some of these myopathies may improve after discontinuation of the statins but it may take several months for maximum recovery. Others need to be treated with steroids and or other immunosuppressive agents.
  - An IBM phenotype is not reported in association with statins therapy. Myopathies may be worsened by statins.
- Neither IVIG nor prednisone, azathioprine, or any other immunomodulatory agent was proven to be effective in treating IBM.
  - Some authorities advocate a 3 months of steroids therapy for patients who show a CK level of more than 20 times normal and intense endomysial inflammation in muscle biopsy. In the author's experience, if improvement happens it is transient and steroids do not change the natural history of the disease.

Reference

Machado PM, Dimachkie MM, Barohn RJ. Inclusion body myositis: a new insight and potential therapy. Curr Opin Neurol 2014;27:591–8.