A Pattern Recognition Approach to Patients with a Suspected Myopathy

Richard J. Barohn, MD[a,*], Mazen M. Dimachkie, MD[b], Carlayne E. Jackson, MD[c]

KEYWORDS

• Myopathy • Limb-girdle • Distal myopathy • Inflammatory myopathy
• Metabolic myopathy • Myotonia

KEY POINTS

- The initial key to the diagnosis of myopathies is recognition of a clinical pattern.
- There are 6 key questions the clinician should consider in arriving at the pattern that fits the patient.
- After arriving at the pattern that fits best, then the clinician can better determine the most appropriate diagnostic tests and management.

INTRODUCTION

Myopathies are disorders affecting the channel, structure, or metabolism of skeletal muscle. Myopathies can be distinguished from other disorders of the motor unit, including the neuromuscular junction, peripheral nerve, or motor neuron, by characteristic clinical and laboratory features. Therefore, the first goal in approaching patients with a suspected muscle disease is to determine the correct *site* of the lesion. Once the lesion is localized to the muscle, the next step is to identify whether the myopathy is caused by a defect in the muscle channel, muscle structure, or a dysfunction in muscle metabolism. The second goal is to determine the *cause* of the myopathy. In general, myopathies can be classified into acquired or hereditary disorders (**Box 1**).

This publication (or project) was supported by an Institutional Clinical and Translational Science Award, National Institutes of Health/National Center for Advancing Translational Sciences grant number UL1TR000001. Its contents are solely the responsibility of the authors and do not necessarily represent the official views of the National Institutes of Health.
[a] Department of Neurology, University of Kansas Medical Center, 3901 Rainbow Boulevard, Mail Stop 2012, Kansas City, KS 66160, USA; [b] Neuromuscular Section, Neurophysiology Division, Department of Neurology, University of Kansas Medical Center, 3901 Rainbow Boulevard, Mail Stop 2012, Kansas City, KS 66160, USA; [c] Division of Neurology, UT Medicine San Antonio, University of Texas Health Science Center, 8300 Floyd Curl Drive, Mail Code 7883, San Antonio, TX 78229-3900, USA
* Corresponding author.
E-mail address: rbarohn@kumc.edu

Neurol Clin 32 (2014) 569–593
http://dx.doi.org/10.1016/j.ncl.2014.04.008 **neurologic.theclinics.com**
0733-8619/14/$ – see front matter

Box 1
Classification of myopathies

- Acquired
  - Drug-induced myopathies
  - Endocrine myopathies
  - Inflammatory/immune myopathies
  - Myopathies associated with other systemic illness
  - Toxic myopathies
- Hereditary
  - Channelopathies
  - Congenital myopathies
  - Metabolic myopathies
  - Mitochondrial myopathies
  - Muscular dystrophies
  - Myotonias

Finally, the third goal in the authors' approach is to determine if there is a specific treatment and, if not, to optimally manage the patients' symptoms in order to maximize their functional abilities and enhance their quality of life.

CLINICAL EVALUATION

The most important component of evaluating patients with a suspected myopathy is obtaining a comprehensive medical history. The history should allow the clinician to make a reasonable preliminary diagnosis that places patients into one of the categories in **Box 1**. The findings on the physical examination, in particular the distribution of muscle weakness, should provide additional information in determining the correct diagnosis. The results of laboratory studies (blood tests, electrodiagnostic studies, muscle biopsy, molecular genetic studies) then play a confirmatory diagnostic role.[1–4]

The first step in this clinical approach is to ask 6 key questions regarding the patients' symptoms.

1. Which Negative and/or Positive Symptoms Do Patients Demonstrate?
2. What is the Temporal Evolution?
3. Is There a Family History of a Myopathic Disorder?
4. Are There Precipitating Factors That Trigger Episodic Weakness or Stiffness?
5. Are There Associated Systemic Symptoms or Signs?
6. What is the Distribution of Weakness?

Which Negative and/or Positive Symptoms Do Patients Demonstrate?

Symptoms of muscle disease (**Box 2**) can be divided into negative complaints, such as exercise intolerance, fatigue, muscle atrophy and weakness, and positive complaints, such as contractures, cramps, myalgias, muscle stiffness, and myoglobinuria.

Weakness is by far the most common negative symptom reported by patients with muscle disease. When the upper extremities are involved, patients notice trouble brushing their teeth, combing their hair, or lifting objects overhead. If the weakness involves the lower extremities, patients will complain of difficulty arising from a low chair