# Chapter 1
# Neuromuscular Disease: An Overview

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Although most patients with neuromuscular diseases can be treated as outpatients, some of them complain rapid and progressive muscle weakness and represent real clinical emergencies. Being able to hypothesize the etiopathological mechanism of hyposthenia is essential from the earliest stages of access to an emergency department. In this regard the role of a neurologist in the emergency department is indispensable. A detailed analysis of all the conditions that may occur in an acute phase goes beyond the purpose of this book that is meant to be a quick reference in the daily management of neuromuscular emergencies. The book deals with the main neuromuscular diseases on the basis of anatomo-clinical localizations. We will analyze the diseases affecting the anterior horns of the spinal cord (tetanus) and those that affect the nerve roots and the nerves (GBS, multiple mononeuritis, porphyria), the neuromuscular junction (MG and botulism), and finally the muscles (rhabdomyolysis, malignant hyperthermia, periodic paralysis).

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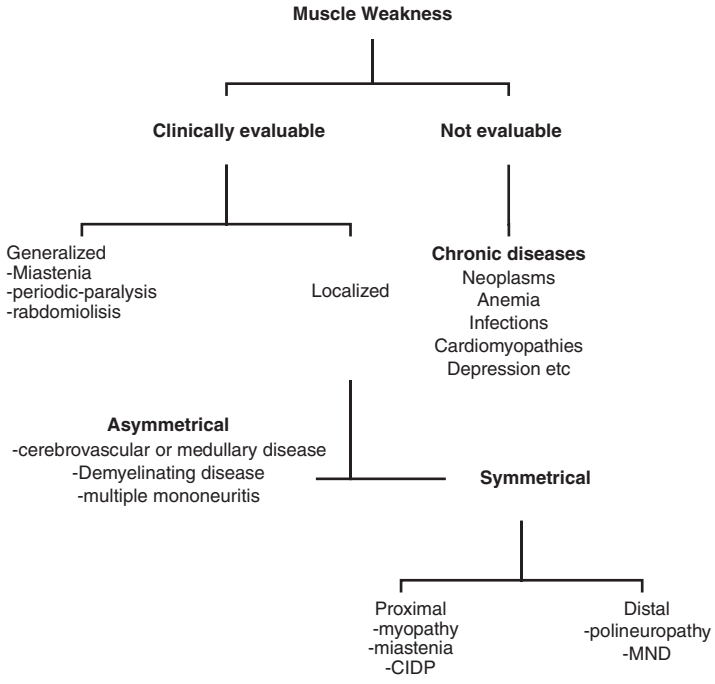


Algorithm 1 Diagnostic algorithm for patients with muscle weakness

In all cases there must be an initial approach aimed at confirming the diagnosis of muscle weakness, which will be followed by an attempt to determine the causes of muscle weakness (Algorithm 1):

- Clinical history analysis
- Clinical exams
- Laboratory exams

The elements of a clinical history that are useful for diagnosis are obviously the preexistence of a neuromuscular disease that may be exacerbated as a result of drugs or bacterial infections; the pre-existence of systemic diseases, medications, or food (botulinum toxin infection, e.g., fish infected with ciguatera, etc.); and the presence of sensory or dysautonomic symptoms.

Collecting a proper drug history is very important to exclude iatrogenic causes:

- Diuretics (hypokalemia)
- Corticosteroids, statins, colchicine, cocaine, cyclosporine, penicillamine (myotoxic effects)
- Amiodarone (demyelinating neuropathy)
- Magnesium-based preparations (hypermagnesemia)

Clinical examination should include evaluation of vital signs and presence of clinical signs of dysautonomia.

The weakness distribution/localization is very important:

- Proximal (basically myopathic) or distal and length dependent (basically neuropathic)
- Symmetrical or asymmetrical
- Involvement or lack of involvement of cranial nerves

Presence or absence of reflexes further helps to clarify the etiology of the symptoms, as well as the presence of dysautonomic sensory disorders. In extremely rapid and acute forms, the need for hospitalization in an ICU must be assessed.

The laboratory tests required in this phase include:

- Blood count (to assess the presence of significant anemia)
- Leukocyte formula (e.g., to highlight a possible eosinophilia)
- ESR and CRP
- CPK (if high it is index of myopathy)
- Indices of hepatic necrosis (transaminases)
- Renal functionality and electrolytes

An EKG should be performed to highlight possible abnormalities related to electrolyte imbalances or dysautonomia.

Our clinical reasoning must answer the following questions:

- Is there a high and imminent risk of respiratory failure?
- Is it necessary to involve the intensive care unit (Table 1.1)?
- Are motor, sensitive, and dysautonomic deficits consistent with our clinical suspicion?
- Is the patient's medical history consistent with our clinical suspicion?

Table 1.1 Factors to be considered when deciding to intubate

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|---|
| *General* |
| – Increasing generalized muscle weakness |
| – Dysphagia |
| – Dysphonia |
| – Dyspnea on exertion and at rest |
| *Subjective* |
| – Rapid shallow breathing |
| – Tachycardia |
| – Weak cough |
| – Interrupted speech (gasping for air) |
| – Use of accessory muscles |
| – Abdominal paradoxical breathing |
| – Orthopnea |
| – Weakness of trapezius and neck muscles: inability to lift head from bed |
| – Inability to perform single-breath count: count from 1 to 10 in single expiration (roughly equal to FVC <1.0 L) |

Table 1.1 (continued)

| | |
|---|---|
| – | Cough after swallowing |
| *Objective* | |
| – | Decreased level of consciousness (have a lower threshold to control the airway if patient requires transfer or movement to unmonitored areas) |
| – | Hypoxemia |
| – | Vital capacity <1 L or 20 mL/kg or 50% decrease in VC in 1 day |
| – | Maximum inspiratory pressure >−30 cm $H_2O$ |
| – | Maximum expiratory pressure <40 cm $H_2O$ |
| – | Hypercarbia (a late finding) |