



Peripheral Nerves and Skeletal Muscles

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Neuromuscular diseases are a complex group of disorders with numerous inherited and acquired causes that typically present with weakness, muscle pain, or sensory deficits. They can be grouped according to anatomy, the tempo of the disease course, and pathogenesis. Physicians keep all these characteristics in mind when evaluating a patient. This chapter uses an anatomical approach, grouping neuromuscular disorders into those that preferentially affect the peripheral nerves, the neuromuscular junction, or the skeletal muscles. A discussion of neoplasms that arise from peripheral nerves ends the chapter. Conditions that can produce similar clinical presentations but are caused by disorders of the central nervous system are discussed in Chapter 28.

Diseases of Peripheral Nerves

The two main components of peripheral nerves are axons and myelin sheaths made by Schwann cells. Injuries to either of these components may result in a peripheral neuropathy. Before discussing the pathology of these

disorders, a brief review of peripheral nerve structure and function is in order. *Somatic motor function* is carried out by the motor unit, which consists of (1) a lower motor neuron located in the anterior horn of the spinal cord or in the brainstem; (2) an axon that travels to a target muscle as part of a nerve; (3) the neuromuscular junctions; and (4) multiple innervated myofibers (muscle fibers). *Somatic sensory function* depends on (1) the distal nerve endings, which may contain specialized structures that serve to register specific sensory modalities; (2) an axon that travels as part of a peripheral nerve to the dorsal root ganglia; and (3) a proximal axon segment that synapses on neurons in the spinal cord or the brainstem. *Autonomic nerve fibers* outnumber somatic fibers in the peripheral nervous system, but signs and symptoms related to their involvement are generally not prominent features of peripheral neuropathies, with a few important exceptions (e.g., in some cases of diabetic neuropathy, discussed later).

Specific sensations (pain, temperature, touch) and motor signals are each conveyed by axons that can be distinguished based on their diameter. Axonal diameters are in turn correlated with the thickness of their myelin sheaths