Neuromuscular Diseases: Disorders of the Motor Neuron and Plexus and Peripheral Nerve Disease

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INTRODUCTION

Neuromuscular diseases are classified into four groups, according to which portion of the motor unit is involved (Table 121-1). Motor neuron and peripheral nerve diseases are considered in this chapter; myopathies are considered in Chapter 122, and neuromuscular junction diseases are considered in Chapter 123. The symptoms and signs of the neuromuscular diseases are at times indistinguishable. However, some useful general rules apply to assist with localization based on the distribution of weakness, presence or absence of sensory symptoms, reflex abnormalities, and specific associated clinical features (Table 121-2).

TABLE 121-1 CLASSIFICATION OF NEUROMUSCULAR DISEASES

SITE OF INVOLVEMENT	TYPICAL EXAMPLES
ANTERIOR HORN CELL	
Without upper motor neuron involvement	Spinal muscular atrophy Progressive muscular atrophy Bulbospinal muscular atrophy Poliomyelitis West Nile virus
With upper motor neuron involvement	Amyotrophic lateral sclerosis Primary lateral sclerosis
PERIPHERAL NERVE	
Mononeuropathy	Carpal tunnel syndrome Ulnar palsy Meralgia paresthetica
Multiple mononeuropathies	Mononeuritis multiplex (e.g., polyarteritis nodosa), leprosy, sarcoidosis, amyloidosis
Polyneuropathies	Diabetic neuropathy Charcot-Marie-Tooth disease Guillain-Barré syndrome
NEUROMUSCULAR JUNCTION	
	Myasthenia gravis Lambert-Eaton syndrome
MUSCLE	
	Duchenne muscular dystrophy Dermatomyositis

Electromyography and Nerve Conduction Studies

Electromyography (EMG) and nerve conduction studies can also be useful diagnostic tools in localizing the lesion in a patient with a suspected neuromuscular disease. The measurement of electrical activity arising from muscle fibers is performed by inserting a needle electrode percutaneously into a muscle. Normal muscle is electrically silent at rest. Spontaneous activity during complete relaxation occurs in myotonic disorders, in inflammatory myopathies, and in denervated muscles. Spontaneous activity of a single muscle fiber is called a *fibrillation,* and such activity of part of or an entire motor unit is called a *fasciculation.* In myotonia, repeated muscle depolarization and contraction occur despite voluntary relaxation. Abnormalities in motor unit potentials occur during the course of denervation; with the development of reinnervation, the remaining motor units increase in amplitude and become longer in duration and polyphasic (E-Fig. 121-1). Conversely, in muscle diseases such as the muscular dystrophies and other diseases that destroy scattered fibers within a motor unit, the motor unit action potentials are of lower amplitude and shorter duration and are polyphasic. A reduced recruitment (interference) pattern from maximum voluntary effort occurs in denervation. Conversely, in patients with primary muscle disease, submaximal voluntary effort produces a full recruitment pattern despite marked weakness.

Nerve conduction is studied by stimulating a peripheral nerve (e.g., the ulnar) with surface electrodes placed over the nerve. The resulting action potential is recorded by electrodes placed over the nerve more proximally in the case of large sensory nerve fibers and over the muscle distally in the case of motor nerve fibers in a mixed motor sensory nerve. For sensory nerves, the sensory nerve action potential (SNAP) is quantitated, and for motor nerves, the compound muscle action potential (CMAP) is quantitated.