Myopathies

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Practice Essentials

Myopathy is a muscle disease unrelated to any disorder of innervation or neuromuscular junction. This condition has widely varying etiologies, including congenital or inherited, idiopathic, infectious, metabolic, inflammatory, endocrine, and drug-induced or toxic.

History

Important information to obtain during the patient’s history includes the following:

- Family history: Any periodic paralysis or muscular dystrophy?
- Personal history: Presence of autoimmune disease, endocrinopathy, renal insufficiency, and/or alcoholism?
- Previous episodes of severe weakness (eg, postexercise, after exposure to cold [possibly one of periodic paralyses]; post high-carbohydrate meals [familial hypokalemic periodic paralysis])
- Medications (eg, steroids, lipid lowering agents, retroviral agents, alcohol, colchicine, pentachlorophenol [PCP], heroin)
- Occupational and travel history (potential ingestion of barium chloride or carbonate [acute hypokalemic paralysis])

Signs and symptoms

The common symptoms of myopathy are muscle weakness, impaired function in activities of daily life, and, rarely, muscle pain and tenderness. Significant muscle pain and tenderness without weakness should prompt consideration of other causes.

General signs and symptoms of myopathy include the following:

- Symmetric proximal muscle weakness
- Malaise, fatigue
- Dark colored urine (suggests myoglobinuria) and/or fever
- Absence of sensory complaints or paresthesias; however, deep tendon reflexes (DTRs) may be diminished/absent in hypokalemic paralysis
- Very late findings: Atrophy and hyporeflexia (early presence usually implicates neuropathies)
- Normal level of consciousness
- Gottron papules in dermatomyositis: Pink-to-violaceous scaly areas over knuckles, elbows, and knees

The acuity of symptom onset may aid in the diagnosis, as follows:

- Weakness progressing over hours: Possible toxic etiology or one of episodic paralyses
- Weakness developing over days: May be an acute dermatomyositis or rhabdomyolysis
- Symptom development over a period of weeks: May be polymyositis, steroid myopathy, or myopathy resulting from endocrine causes (eg, hyperthyroidism, hypothyroidism)

Indications of which muscle groups are involved include the following symptoms:

- Proximal muscle weakness: Difficulty rising from chairs, getting out of the bathtub, climbing stairs, and/or shaving or combing the hair
- Weakness of distal muscles: Weak grasp, handwriting problems, and walking difficulties, (eg, flapping gait)

See Clinical Presentation for more detail.

Diagnosis

Laboratory testing

The following laboratory tests may be used to evaluate patients with myopathies:

- Creatine kinase (CK) levels with isoenzymes
- levels of electrolytes, calcium, and magnesium
- Serum myoglobin levels
- Serum creatinine and blood urea nitrogen levels
- Urinalysis: Myoglobinuria indicated by positive urinalysis with few red blood cells on microscopic evaluation
- Complete blood count
- Erythrocyte sedimentation rate
- Thyroid function tests
- Aspartate aminotransferase levels

Other studies may include the following:

- Electrocardiography
- Antinuclear antibody levels
- Genetic testing
- Electromyography
- Magnetic resonance imaging (to assess complications or rule out neurologic disease)
Muscle biopsy

See Workup for more detail.

Management

The treatment of a myopathy is dependent on its etiology and can range from supportive and symptomatic management to therapy for specific conditions. Such treatments may include the following:

- Supportive: Management of airway, breathing, circulation; hydration; intensive care management may be needed in some cases
- Drug therapy
- Physical therapy
- Bracing
- Surgery

See Treatment for more detail.

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References


