Myopathy

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1 Introduction

Myopathy is derived from the Greek words "myo" for muscle, and "pathos" for suffering, which means muscle disease. The most common signs and symptoms of myopathies include weakness, stiffness, cramps, and spasms. Myopathies are a heterogeneous group of disorders primarily affecting the skeletal muscle structure, metabolism, or channel function. They usually present with muscle weakness interfering in daily life activities. Muscle pain is also a common finding and some myopathies are associated with rhabdomyolysis.

2 Etiology

The etiology of the myopathies is usually caused by a disruption in the muscle tissue integrity, and the metabolic stability which may be triggered by inherited genetic diseases, or metabolic errors, certain drugs and toxins, bacterial or viral infections, inflammation, besides minerals, electrolytes, and hormonal irregularities:

- Inherited Myopathies
  - Mitochondrial Myopathies, such as mitochondrial encephalopathy, lactic acidosis, and stroke-like syndrome (MELAS)
  - Congenital Myopathies
  - Metabolic Myopathies, such as Pompe disease
  - Muscular Dystrophies, such as Duchenne muscular dystrophy and Becker muscular dystrophy
  - Myotonic muscular dystrophies, such as Steinert's Disease
- Acquired Myopathies
  - Toxic Myopathies (statins, antiretrovirals, vincristine)
  - Immune-mediated or Idiopathic Inflammatory Myopathies, such as Dermatomyositis-Polymyositis

- Infectious Myopathies
- Endocrine Myopathies
- Electrolyte-mediated

The term "muscular dystrophy" incorporates an assortment of hereditary disorders that lead to progressive, generalized disease of the muscle prompted by inadequate or missing glycoproteins in the muscle cell plasma membrane.

3 History and Physical

Myopathies are typically involving motor impairment without no sensory symptoms. It presents as proximal muscle weakness, mainly in the pelvic girdle or the shoulder girdle muscle groups. However, pelvic muscle group is more common and more severe. Comprehensive history alongside physical examination is mandatory to identify and diagnose myopathies. Patients may complain of difficulty raising up from sitting position, climbing stairs or difficulty brushing their hair, or practicing any above head activities. Some other myopathies will present in different muscle groups like thighs, back muscles, or fingers, and could be possibly associated with other symptoms like myalgia, rashes, fatigue, or cramps.

Duchenne muscular dystrophy (DMD) It is not only one of the most severe forms of inherited muscular dystrophies but also the most common hereditary neuromuscular disease. Sadly, there is no known treatment modality that halts the progression of the disease; available treatment options are palliative. Affected patients usually die in their twenties due to respiratory muscle weakness or cardiomyopathy.

Becker muscular dystrophy (BMD) It is an X-linked recessive disorder involving dystrophin gene mutation, resulting in progressive muscle degeneration. The proximal lower limb muscles are most often affected, while heart failure is a frequent complication among individuals with this condition. BMD is less common and less severe than Duchenne muscular dystrophy (DMD). The onset of BMD symptoms is also late compared to DMD, although it varies widely between 5 and 60 years of age. Management is supportive, with rehabilitative care essential to maintaining function long-term.

Polymyositis and dermatomyositis Both of them present with proximal weakness affects pelvic girdle more than shoulder girdle. Polymyositis is associated with arthralgia while dermatomyositis is associated with other symptoms mainly skin manifestations.