

Myopathy

Biomedical Engineering - URJC

Rafa Carretero, MD, PhD

Internal Medicine Department

29 February 2024



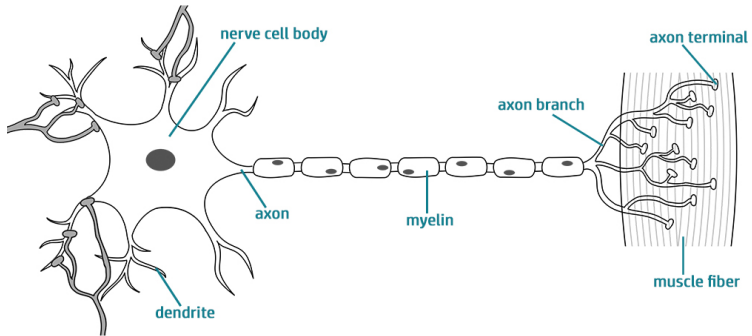
Universidad
Rey Juan Carlos

Myopathies

Introduction

- Myopathy: muscle disease
- Signs and symptoms: weakness, stiffness, cramps, spasms
- Affect skeletal muscle structure, metabolism, or channel function
- Commonly present with muscle weakness and pain

Myopathies



Etiology of Myopathies

- Disruption in muscle tissue integrity and metabolic stability
- Causes: inherited genetic diseases, metabolic errors, drugs, toxins, infections, inflammation, minerals, electrolytes, hormonal irregularities

Types of Myopathies

Inherited Myopathies

- Mitochondrial Myopathies (e.g., MELAS)
- Congenital Myopathies
- Metabolic Myopathies (e.g., Pompe disease)
- Muscular Dystrophies (e.g., Duchenne, Becker)
- Myotonic Muscular Dystrophies (e.g., Steinert's Disease)

Types of Myopathies (Contd.)

Acquired Myopathies

- Toxic Myopathies (statins, antiretrovirals, vincristine)
- Immune-mediated or Idiopathic Inflammatory Myopathies (e.g., Dermatomyositis-Polymyositis)
- Infectious Myopathies
- Endocrine Myopathies
- Electrolyte-mediated

Muscular Dystrophy

- Group of genetic disorders leading to progressive muscle disease
- Duchenne Muscular Dystrophy (DMD)
 - Most severe and common hereditary neuromuscular disease
 - No known cure; palliative treatment
 - Patients usually die in their twenties due to respiratory muscle weakness or cardiomyopathy
- Becker Muscular Dystrophy (BMD)
 - X-linked recessive disorder
 - Less severe than DMD, onset varies widely
 - Supportive management with rehabilitative care

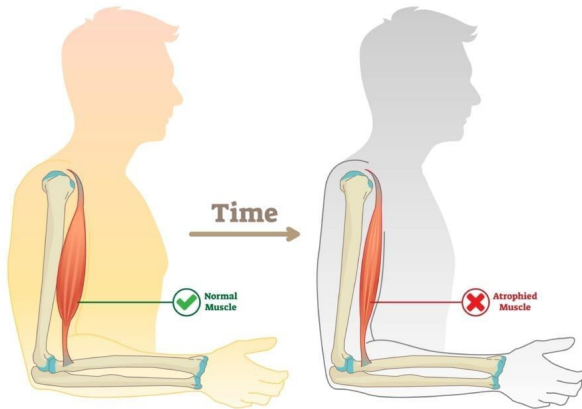
History and Physical Examination

- Myopathies involve motor impairment without sensory symptoms
- Present with proximal muscle weakness, often in pelvic girdle
- Comprehensive history and physical examination crucial for diagnosis
- Symptoms may include difficulty in daily activities, myalgia, rashes, fatigue, or cramps

Polymyositis and Dermatomyositis

- Present with proximal weakness, affecting pelvic girdle more than shoulder girdle
- Polymyositis associated with arthralgia
- Dermatomyositis associated with skin manifestations

MUSCULAR DYSTROPHY



Myopathies

INFLAMMATORY MYOPATHIES

- * POLYMYOSITIS
 - * DERMATOMYOSITIS
 - * INCLUSION BODY MYOSITIS
- } AUTOIMMUNE DISORDERS

ORGAN-SPECIFIC SYMPTOMS

- ~ SUBCUTANEOUS CALCIFICATION in the SKIN
- ~ JOINT PAIN
- ~ ARTHRITIS



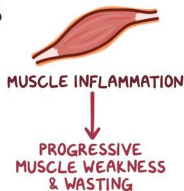
- ~ ARRHYTHMIAS
- ~ MYOCARDITIS
- ~ PERICARDITIS



- ~ GASTROESOPHAGEAL REFLUX DISEASE
- ~ DYSPHAGIA



- ~ ASPIRATION PNEUMONIA

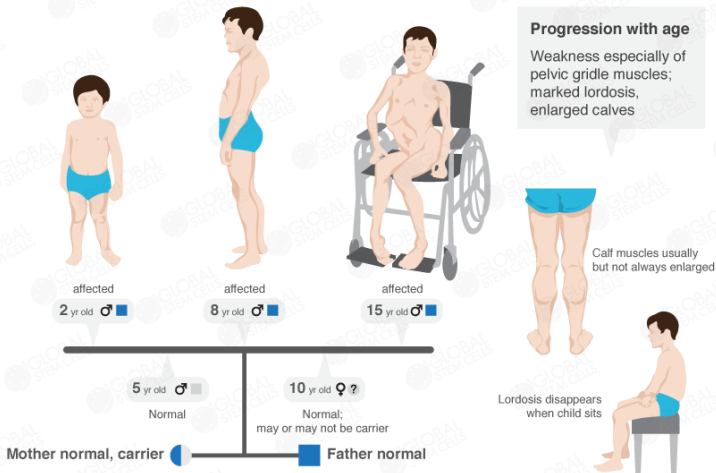


SYSTEMIC SYMPTOMS

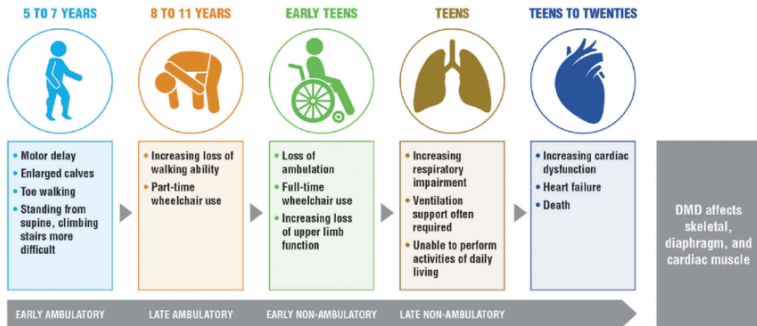
- ~ FEVER
- ~ FATIGUE
- ~ WEIGHT LOSS



Myopathies



Myopathies



Bashir K, Connor E. Clin Invest (Lond) 2011;121:1217-1225; Orat Gazman, et al. Int J Endocrinol 2012;2012:485576.