

AI-01961 Myopathy vs Muscle Weakness

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Explain myopathy. What are types of myopathy? What is the pathophysiology of myopathy?

Myopathy is a general term for any disease that affects the muscles that control voluntary movement in the body. The primary symptom is muscle weakness, which results from a dysfunction of the muscle fibers themselves. Other symptoms can include muscle cramps, stiffness, and pain. Myopathies can be inherited or acquired.

Types of Myopathy

Myopathies are a diverse group of disorders with various causes. They can be broadly categorized into inherited and acquired forms.

Inherited Myopathies: These are caused by genetic mutations that affect the structure or function of muscle proteins. Examples include:

- **Muscular Dystrophies:** A group of genetic disorders characterized by progressive weakness and degeneration of voluntary muscles.
- **Congenital Myopathies:** These are present at birth and often involve developmental delays in motor skills. They typically do not worsen over time.
- **Mitochondrial Myopathies:** Caused by mutations in the mitochondria, the "powerhouses" of the cells, which produce energy. These can affect not only muscles but also the brain, heart, and gastrointestinal system.
- **Metabolic Myopathies:** Result from the body's inability to properly convert food into energy for muscle use. This can lead to episodes of muscle weakness, often triggered by exercise.

- **Channelopathies:** Caused by genetic mutations that impair the function of ion channels, which are crucial for muscle contraction.
Acquired Myopathies: These can develop later in life due to various factors.
Examples include:
- **Inflammatory Myopathies:** These are autoimmune disorders where the body's immune system mistakenly attacks its own muscle fibers, causing inflammation and weakness. Examples include dermatomyositis, polymyositis, and inclusion body myositis.
- **Toxic Myopathies:** Caused by exposure to certain substances, such as alcohol, or side effects from specific medications like statins.
- **Endocrine Myopathies:** Result from hormonal imbalances, such as those caused by thyroid disorders (hypothyroid and hyperthyroid myopathies).
- **Infectious Myopathies:** Caused by viruses, bacteria, or parasites.

Pathophysiology of Myopathy

The pathophysiology of myopathy refers to the functional and structural changes that lead to the disease's symptoms. While the specific mechanisms vary depending on the type of myopathy, the general theme is a disruption in the integrity or function of the skeletal muscle tissue.

- **Disruption of Muscle Integrity and Structure:** Many myopathies involve defects in the proteins that make up the muscle fibers. In muscular dystrophies, for example, a lack of the protein dystrophin leads to muscle fiber damage and progressive weakness.
- **Metabolic Defects:** In metabolic myopathies, the body's inability to process fuel (like carbohydrates or fats) correctly means the muscles don't get the energy they need to function properly. This can lead to muscle weakness, cramping, and pain, especially during or after exercise.
- **Inflammation:** In inflammatory myopathies, the immune system attacks the muscle tissue, causing chronic inflammation and muscle damage.
- **Problems with Excitation-Contraction Coupling:** Some myopathies, such as channelopathies, affect the ion channels and other components that are

essential for the electrical signals to travel through the muscle cells and trigger muscle contraction. This disruption can lead to muscle weakness or stiffness.