



# PATIENT FACT SHEET

## Metabolic Myopathies



### CONDITION DESCRIPTION

**Metabolic myopathies are rare, genetic disorders that cause muscle problems.** Metabolic refers to chemical reactions that provide necessary energy and nutrients for healthy muscle growth.

Metabolic myopathies are inherited and tend to run in families. They can occur at any age. Genetic defects interfere with the processes in muscle that generate energy. Muscle cells cannot work properly. Some myopathies can cause rhabdomyolysis. Muscles break

down, and muscle fiber can enter the bloodstream and cause kidney failure.

To work properly, muscles need a chemical called adenosine triphosphate [ATP]. Cells make ATP from sugars and fats on certain pathways in chemical reactions driven by enzymes. Lack of certain enzymes causes metabolic myopathies, which are named according to the pathways affected.



### SIGNS/ SYMPTOMS

**Symptoms of metabolic myopathies include muscle weakness, tiring after exercise or activity, muscle pain after any physical effort, and swollen or tender muscles.**

Some people with metabolic myopathies never have symptoms. Other pathways in their body make the ATP necessary for muscles to work when one pathway does not work. When the body needs to make more energy from that pathway, an ATP deficit can develop and cause symptoms. If cells are moderately low in ATP, signs are fatigue or exercise intolerance.

Rhabdomyolysis, or the death of muscle cells, occurs when cells do not have enough ATP. Low-grade rhabdomyolysis may cause constant weakness.

A rheumatologist can diagnose metabolic myopathies with a muscle tissue biopsy. A needle is used to remove a small sample of tissue. Newer blood testing may also be used to diagnose metabolic myopathies.



### COMMON TREATMENTS

**Treatments vary according to the patient's particular myopathy.** Treatments include changes in physical activity and diet, aerobic exercise training, and the use of various vitamins and supplements. The goal of treatment is to control symptoms.



### CARE/ MANAGEMENT TIPS

**Metabolic myopathies may cause symptoms that are normal among healthy people.** Most people get tired after physical exertion. Metabolic myopathies are rare diseases, so diagnosis and treatment may be delayed. Early detection may happen when patients, family or friends realize there is a reason for these physical limitations.

Because metabolic myopathies are inherited, patients planning a family should get genetic counseling to understand the risks to future children.

Appropriate physical activity, regular aerobic exercise, a healthy diet, and use of vitamins and supplements may help control metabolic myopathy symptoms and improve quality of life.