



Metabolic Myopathies

Metabolic myopathies are genetic diseases, usually inherited, that affect the body's muscles. [Metabolism refers to chemical reactions that provide energy and nutrients, or substances necessary for health and growth.] Some people with a metabolic myopathy (muscle disease) develop weakness; others tire easily with exercise or physical activity, suffer muscle pain after physical effort, and/or experience severely swollen and tender muscles. These symptoms occur when muscle cells don't get enough energy. Without enough energy, the muscle lacks enough fuel to work properly.

Fast facts

- Each of these disorders causes symptoms when the muscle cells cannot make enough energy to move the muscle.
- Not all people with a metabolic myopathy experience the same symptoms.
- Diet, levels of physical activity, and dietary supplements recommended for treatment will vary depending on the myopathy.

What are metabolic myopathies?

Metabolic myopathies are genetic defects that interfere with the energy-generating processes in skeletal muscles. When these defects interfere with muscle function and block energy production, muscle cells cannot work properly. Then the muscles that help the body's movement are unable to communicate ("talk") between bones and joints. These myopathies can cause progressive muscle weakness, fatigue, episodes of pain and cramps after exercise, and/or extensive death and breakdown of muscle tissue.

Some myopathies can cause rhabdomyolysis. In this condition, muscles break down, leaving them very weak, sore, swollen and tender. As muscles break down, portions of muscle fiber enter the bloodstream, which can lead to kidney failure.

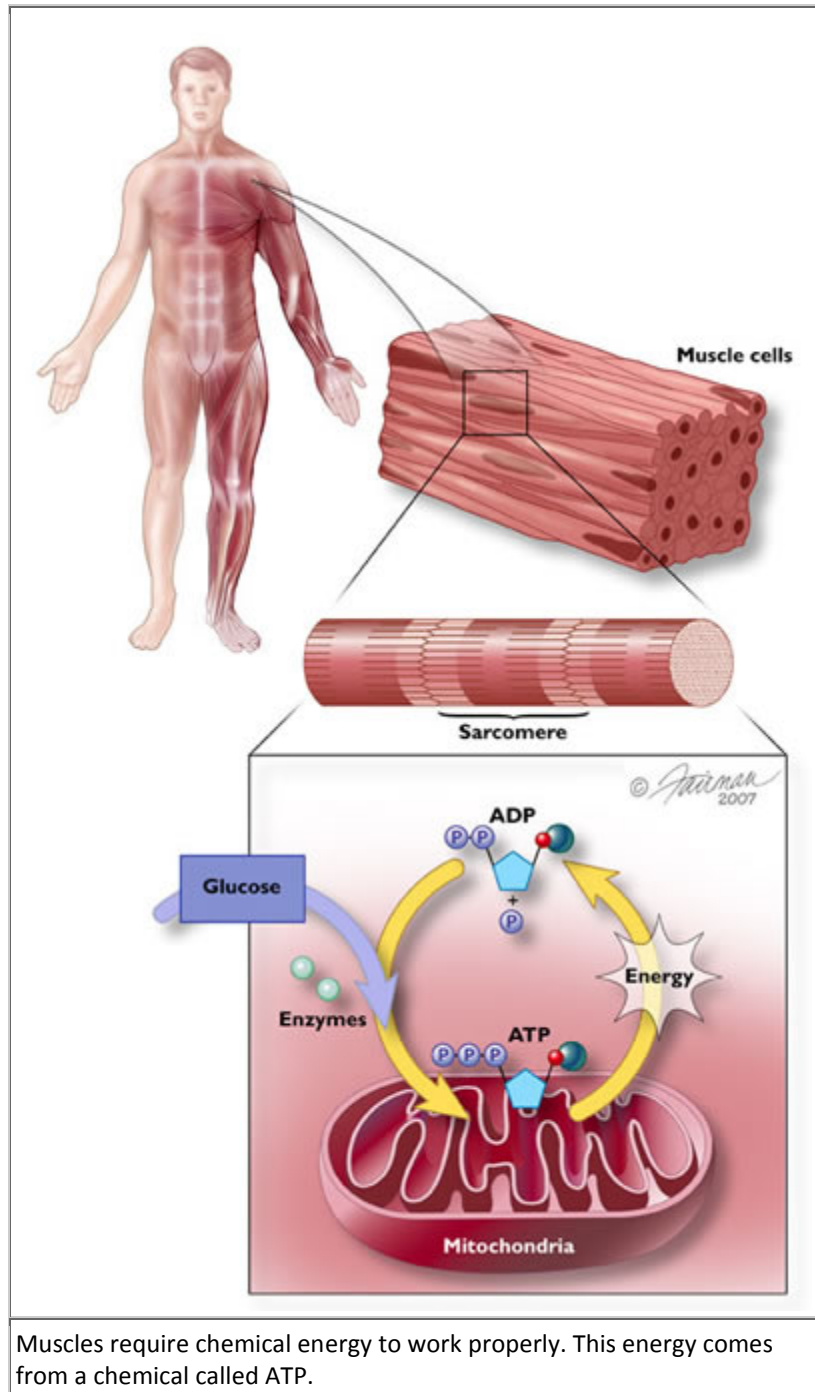


What causes metabolic myopathies?

Muscles require chemical energy to work properly. This energy comes from a chemical called adenosine triphosphate (sometimes called ATP). ATP is made in cells from sugars or fats by chemical reactions called pathways. Normally, most ATP is produced in small cell compartments called mitochondria.

Proteins called enzymes are necessary for each chemical reaction. A metabolic myopathy is caused when there isn't enough of a particular enzyme is not present to cause the necessary reactions.

The name of each metabolic myopathy is based on which pathway has the deficient enzyme. For instance, diseases caused by a defect in sugar metabolism are called glycogen storage diseases. The most common defect in sugar metabolism is McArdle's disease. Lipid storage diseases are due to abnormal fat processing. Finally, the term mitochondrial myopathy is used when the enzyme present in mitochondria is deficient.



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Some people with metabolic myopathies live normal lives and never experience significant symptoms. That is because cells have several ways (pathways) to make ATP. Under usual conditions, normal pathways can produce enough ATP to make up for the deficient pathway. However, when the body needs to make more energy from the deficient pathway, the deficit in ATP can become severe and symptoms develop. When cells are



moderately low in ATP, the major problem may be fatigue or exercise intolerance. [Think of a car engine. If the gas in the tank is good fuel, the engine runs well. If the gas has water or sand in it, the car runs poorly.] Rhabdomyolysis (muscle cell death) occurs when the cells don't have enough ATP (that is, the car runs out of gas). Constant or on-going weakness may develop as a result of repeated low-grade rhabdomyolysis.

Who gets metabolic myopathies?

Most metabolic myopathies tend to run in families, appearing at any age in those who inherit the disease. Either parent or another relative may have been symptomatic. However, metabolic myopathies can develop in a person with no family history of the condition.

How are metabolic myopathies diagnosed?

Most often, physicians diagnose metabolic myopathies by testing a sample of muscle tissue (biopsy), which is taken through a needle or with a small incision under local anesthesia. However, more recent techniques allow diagnosis by testing blood samples.



Treatment is focused on changes in physical activities, aerobic exercise training.

How are metabolic myopathies treated?

Treatment varies by myopathy, but is focused on changes in physical activities, aerobic exercise training, changes in diet, and the use of various vitamins or supplements.

Living with metabolic myopathies

Everyone has run out of energy, become tired when doing physical activities, or experienced muscle pains and cramps after exercising. These are common symptoms. However, for some, these are the symptoms coming from metabolic myopathy. Metabolic myopathies are rare, so diagnosis can be delayed.

How do we know when these common symptoms mean that patients have one of these diseases? First, patients, family and friends realize there is a reason for their limitations. Second, because these are inherited diseases, genetic counseling will help people understand the potential risks for future children. Finally, appropriate activity, regular aerobic exercise, proper diet and use of supplements can help control symptoms.

Points to remember

- Metabolic myopathies are rare genetic diseases that cause muscle problems.
- Diagnosis often requires a muscle biopsy.
- Despite the fact that metabolic myopathies are inherited, treatments can help many people with these diseases live normal lives.



The role of the rheumatologist

Many different diseases and conditions can cause muscle symptoms. Rheumatologists are trained to determine the diagnosis responsible for the symptoms in individual patients.

To find a rheumatologist

For a listing of rheumatologists in your area, [click here](#). Learn more about [rheumatologists](#) and [rheumatology health professionals](#).

For more information

The American College of Rheumatology has compiled this list to give you a starting point for your own additional research. The ACR does not endorse or maintain these Web sites, and is not responsible for any information or claims provided on them. It is always best to talk with your rheumatologist for more information and before making any decisions about your care.



With appropriate activity, changes, regular aerobic exercise, diet and use of supplements, the symptoms often can be controlled.

For more information, contact the Muscular Dystrophy Association www.mda.org.

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Written by Robert L. Wortmann, MD, and reviewed by the American College of Rheumatology Communications and Marketing Committee.

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