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 (a process when muscle tissue gets damaged and substances from inside the muscle cells enter the bloodstream).

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.

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 or genetic testing. With a biopsy, a needle is used to remove a small sample of tissue.

Treatments vary according to the patient’s particular myopathy. Treatments include changes in physical activity and diet, aerobic and anaerobic (resistance) exercise training, and the use of various vitamins and supplements.

The goal of treatment is to control symptoms and minimize progression of muscle weakness. Physical and occupational therapy are often important in the management of metabolic myopathies.

The best way to prevent Lyme disease is to lower the risk of tick bites. Avoid ticks’ favorite habitats, like tall grasses or brush. Stick to clear paths when hiking. When outdoors, wear light-colored clothes to easily spot ticks, long sleeves and pants tucked in at the hems.

Use insect repellents on skin or clothing when walking outdoors. Regularly check for and remove ticks from skin. Remove a tick by squeezing its head with a tweezers and pulling it off the skin.

If infected, rest and pace activity until symptoms improve with treatment. If symptoms linger, get plenty of rest, good nutrition and regular exercise if possible. Follow up regularly with a rheumatologist to help recovery.