Inflammatory Myopathies- Polymyositis, Dermatomyositis

The prospect of a muscle disease worries some people because they are afraid of not being able to walk. Yet in many cases, treatment exists for myopathy (muscle disease). Proper diagnosis and treatment raise the chance of living life fully despite this illness.

What are inflammatory myopathies?

Myopathy is the medical term for muscle disease. Some muscle diseases occur when the body's immune system attacks muscles. The result is misdirected inflammation, hence the name inflammatory myopathies. This damages muscle tissue and makes muscles weak.

People with inflammatory myopathies may have these features:

- Weakness in the large muscles around the neck, shoulders and hips
- Trouble climbing stairs, getting up from a seat, or reaching for objects overhead
- Little, if any, pain in the muscles
- Choking while eating or aspiration (intake) of food into the lungs
- Shortness of breath and cough

The inflammatory myopathies include **polymyositis** and **dermatomyositis**. Muscle inflammation and weakness occur in both conditions while patients with dermatomyositis also have a rash. This rash most often appears as purple or red spots on the upper eyelids or as scaly, red bumps over the knuckles, elbows or knees. Children with the disease also may have white calcium deposits in the skin called calcinosis.

Sometimes patients can have the rash with no sign of muscle disease. Doctors call this form of the disease amyopathic dermatomyositis.

People with dermatomyositis may also have lung inflammation that causes cough and shortness of breath. Children with the disease may have an inflammation of the blood vessels (vasculitis) that can result in skin lesions.

Some doctors group a health problem called **inclusion body myositis** with the inflammatory myopathies. Yet, it differs from them. Men get it more often than women, and the patients tend to be older. Most of these patients do not respond to standard treatment. Therefore, this fact sheet will not discuss this disease.

Who gets inflammatory myopathies?

Inflammatory myopathies are rare. Polymyositis and dermatomyositis occur in about one person per 100,000.

All ages can get these diseases. The peak time to get them, though, is ages 5 to 10 in children, and 40 to 50 in adults. Women get inflammatory myopathies about twice as often as men. These diseases affect all ethnic groups. No one can predict who will get an inflammatory myopathy.

What causes inflammatory myopathies?

There are many causes of muscle disease. They include infection, muscle injury due to medicine, inherited diseases that affect muscle function, disorders of electrolyte levels, and thyroid disease.
We do not know what causes inflammatory myopathies. A top theory is that something goes wrong in the immune system that leads to inflammation. This damages muscle cells or the blood vessels that are in the muscle.

**How are inflammatory myopathies diagnosed?**

A doctor suspects myopathy when patients complain of trouble doing tasks that require muscle strength, or when they get certain rashes or breathing problems.

Most people with myopathy have little or no pain in their muscles. This differs from other health problems. Examples are other forms of muscle disease, joint pain due to arthritis and nerve problems that cause numbness or tingling in the hands and feet. Many patients with other illnesses think they feel weak. In fact, they are tired, short of breath or depressed, rather than having true muscle weakness.

A doctor will do a muscle strength exam to find if true muscle weakness is present. The following test may then be needed:

- A blood test to measure the level of various muscle enzymes
- An electromyogram — often referred to as an EMG — to gauge electrical activity in muscle
- A biopsy of a weak muscle (a small piece of muscle tissue is removed for testing)
- Magnetic resonance imaging — more often called MRI — to try to show abnormal muscle

Some patients may have a blood test for myositis-specific antibodies (immune proteins). This test helps detect myopathy. It also gives some information about prognosis (how serious the disease is).

**How are inflammatory myopathies treated?**

Doctors most often treat these diseases with medications.

**Corticosteroids.** Often, the first treatment is an oral (by mouth) corticosteroid, such as prednisone, at a high dose. This reduces inflammation. Blood muscle enzymes tend to return to normal about 4–6 weeks after treatment starts. Most patients regain muscle strength in 2–3 months.

**Combination treatment.** Your doctor likely will add another drug to your treatment plan: methotrexate or azathioprine. This gives better long-range control of the disease and helps avoid long-term side effects of corticosteroids. These side effects include weight gain and redistribution of body fat, thinning of the skin, osteoporosis and cataracts. Even muscle weakness can be a side effect.

Because patients taking prednisone are at risk for osteoporosis, they should receive proper treatment to prevent it.

**Immune treatments.** Patients whose disease is severe or who do not respond to standard treatment have other options. They include intravenous immunoglobulin — sometimes referred to as IVIg — or medicines that suppress the immune system. These immunosuppressive drugs include cyclosporine (Neoral, Sandimmune), tacrolimus (Prograf), mycophenolate mofetil (Cellcept) and rituximab (Rituxan).

**Physical therapy.** Physical therapy and exercise are important in the treatment of muscle disease. Very weak patients who do not walk should receive range of motion exercises. These exercises prevent joint contractures (deformity of the joint). Patients with medium weakness should start an exercise program.
to strengthen their muscles. The goal is to slowly increase in intensity as the patient regains strength. Mildly weak patients should take part in their normal activities.

Broader health impacts of inflammatory myopathies
In adults, dermatomyositis and to a lesser extent polymyositis at times may be related to an underlying cancer. Therefore all adult patients with these disorders should receive appropriate testing to rule out cancer.

Living with inflammatory myopathy
Myopathies are chronic (long-term) diseases. To help control your illness, it is important to practice good health measures. Eat a healthy, well-balanced diet, exercise and try to keep a healthy weight.

If you have a dermatomyositis rash, protect yourself from the sun. This is because the rash gets worse after sun exposure, for reasons that are not clear. Therefore, limit time outdoors, and put on sunscreen when you go outside.

If you have trouble swallowing, eat soft or semisolid foods, or puree your solid foods. To prevent choking if you are bedbound, you should be seated up in bed to eat.

People with myopathy may look healthy and normal. It is important for employers, teachers and family members to understand the limits that muscle weakness causes in people with myopathy.

Points to remember
- Inflammatory Myopathy almost always causes loss of muscle strength.
- Some patients also have rashes or breathing problems, or both.
- While there is no cure for myopathies, treatment is most often effective. Following your doctor's treatment plan is key to having a good outcome.

The role of a rheumatologist in the treatment of inflammatory myopathies
You need a correct diagnosis of myopathy to have the best possible outcome. Rheumatologists are doctors who are experts in diagnosing and treating diseases of the muscles, joints and bones. Therefore, they are more likely to make a proper diagnosis of myopathy. They can also advise patients about the best treatment options available.
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, Muscles require chemical energy to work properly. This energy comes from a chemical called ATP.
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.

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. 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.

**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 commons 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.