



Facts About **Metabolic** Diseases of Muscle

MDA[®] Muscular
Dystrophy
Association
Fighting Muscle Disease

Dear Friends:

If you've just learned that you or a loved one has a metabolic muscle disorder, you're probably both relieved and concerned. That's how I felt when I learned at age 27 that I had muscle phosphorylase deficiency, or McArdle disease.

It was a great relief to have a name and an explanation for a problem I'd had since early childhood. Knowing that my disease is rare and hard to diagnose helped me understand why I'd spent so many years believing I needed to "try harder," but only feeling weaker when I did. It was a relief to know that I wasn't "lazy" and wasn't the only one with this problem.

But getting a diagnosis also raised some questions.

What treatment was there? Would my symptoms get worse? Did the disease affect more than my voluntary muscles? How could I avoid episodes of weakness? Would my children have the same disease?

All those questions are addressed in this booklet from the Muscular Dystrophy Association. MDA offers information and support that will help you move from self-doubt to self-management of your metabolic disorder.

You can, to a great extent, manage your disorder and minimize some of the serious effects. I've learned how to say no to activities that could do harm. I've learned to watch for signs of muscle breakdown so I can avoid kidney failure. I know what treatment I need in case of an emergency.

Metabolic muscle diseases affect each person differently, but for most of us, it doesn't limit our lives as much as you may fear.

I'm in my mid-40s and live a very active lifestyle. My life is full and rewarding with

my beautiful wife and three children. I work full time as a human resource professional, attend college, and spend my spare time remodeling houses, camping and doing the activities I enjoy.

Those whose metabolic disease is more disabling will find much support today. Federal laws guarantee your right to a public education, equal employment opportunity and access to public places. Technology makes it possible for many people to perform work that's suited to their levels of ability and health.

Part of maintaining a healthy lifestyle is learning about your metabolic muscle disease. I take care of myself by avoiding injury, eating healthy and visiting the doctor regularly. Meeting other people with McArdle disease, participating in medical research and talking to doctors have helped me take control of my life. Learning more about your disease is just the beginning of your journey.

"MDA Is Here to Help You," on page 13, describes the many services MDA offers. The Association's scientists are making great progress in understanding metabolic diseases and finding treatments for them. We all pray for the day when no one has to go through the physical and emotional pain that these diseases can cause.

This booklet will give you the basic facts about your metabolic muscle disease, and MDA will help you answer all your questions as they arise. As you face the challenges ahead, please remember that you're not alone.



Keith Stout
Edmond, Oklahoma



Keith Stout

What Are Metabolic Diseases of Muscle?

Metabolic diseases of muscle were first recognized in the second half of the 20th century. Each of these disorders is caused by a different genetic defect that impairs the body's *metabolism*, the collection of chemical changes that occur within cells during normal functioning.

Specifically, the metabolic diseases of muscle interfere with chemical reactions involved in drawing *energy* from food. Normally, fuel molecules derived from food must be broken down further inside each cell before they can be used by the cells' *mitochondria* to make the energy molecule ATP.

The mitochondria inside each cell could be called the cell's "engines." The metabolic muscle diseases are caused by problems in the way certain fuel molecules are processed before they enter the mitochondria, or by the inability to get fuel molecules into mitochondria.

Muscles require a lot of energy in the form of ATP to work properly. When energy levels become too low, muscle weakness and exercise intolerance with muscle pain or cramps may occur.

In a few metabolic muscle disorders, symptoms aren't caused so much by a lack of energy, but rather by unused fuel molecules that build up inside muscle cells. This buildup may damage the cells, leading to chronic weakness.

Metabolic muscle diseases that have their onset in infancy tend to be the most severe, and some forms are fatal. Those that begin in childhood or adulthood tend to be less severe, and changes in diet and lifestyle can help most people with the milder forms adjust.

There are 10 metabolic diseases of muscle (*myopathies*) in MDA's program. Each one gets its name from the substance that's lacking:

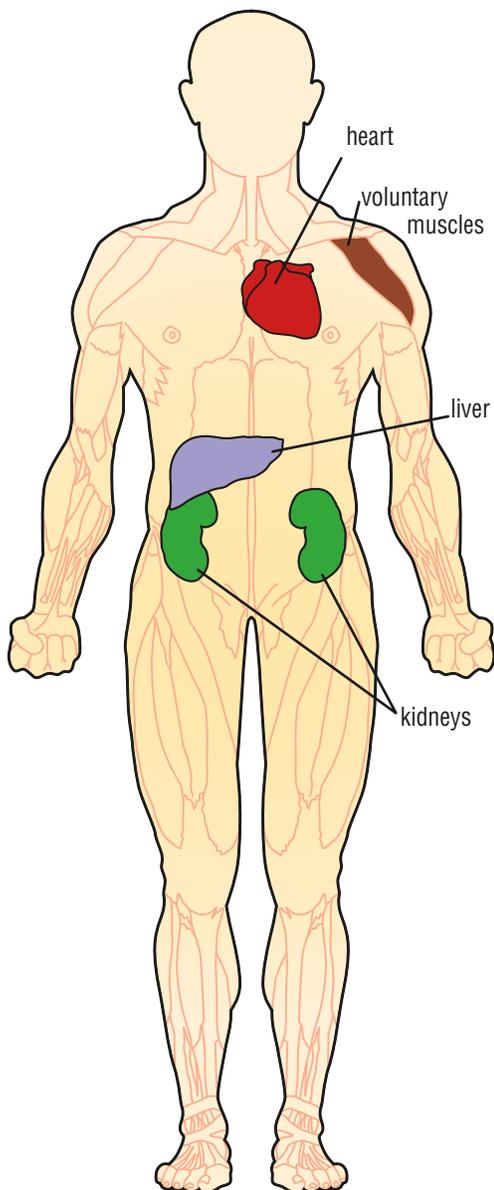
- acid maltase deficiency (Pompe disease)
- carnitine deficiency
- carnitine palmityl transferase deficiency
- debrancher enzyme deficiency (Cori or Forbes disease)
- lactate dehydrogenase deficiency
- myoadenylate deaminase deficiency
- phosphofructokinase deficiency (Tarui disease)
- phosphoglycerate kinase deficiency
- phosphoglycerate mutase deficiency
- phosphorylase deficiency (McArdle disease)

What causes metabolic diseases?

Nine of the diseases in this brochure are caused by defects in the *enzymes* that control chemical reactions used to break down food. Enzyme defects are caused by flaws in the *genes* that govern production of the enzymes.

The 10th disease, *carnitine deficiency*, is caused by lack of a small, naturally occurring molecule that's not an enzyme but is involved in metabolism.

Enzymes are special types of proteins that act like little machines on a microscopic assembly line, each performing a different function to break down food molecules into fuel. When one of the



Metabolic diseases of muscle can affect all the body's voluntary muscles, such as those in the arms, legs and trunk. Some also can involve increased risk of heart or liver diseases, and the effects can damage the kidneys.

The main symptom of most of the metabolic myopathies is difficulty performing some kind of exercise.

enzymes in the line is defective, the process goes more slowly or shuts down entirely.

Our bodies can use carbohydrates (starches and sugars), fats and protein for fuel. Defects in the cells' carbohydrate- and fat-processing pathways usually lead to weakness in the voluntary muscles, but also may affect the heart, kidneys or liver. Although defects in protein-processing pathways can occur as well, these usually lead to different kinds of disorders that affect other organs.

A *gene* is a "recipe" or set of instructions for making a protein, such as an enzyme. A defect in the gene may cause the protein to be made incorrectly or not at all, leading to a deficiency in the amount of that enzyme. Genes are passed from parents to children. Therefore, gene defects can be inherited. (See "Does It Run in the Family?" on page 11.)

The metabolic muscle diseases aren't contagious, and they aren't *caused* by certain kinds of exercise or lack of exercise. However, exercise or fasting (not eating regularly) may bring on episodes of muscle

weakness in a person who has the disease because of a genetic flaw.

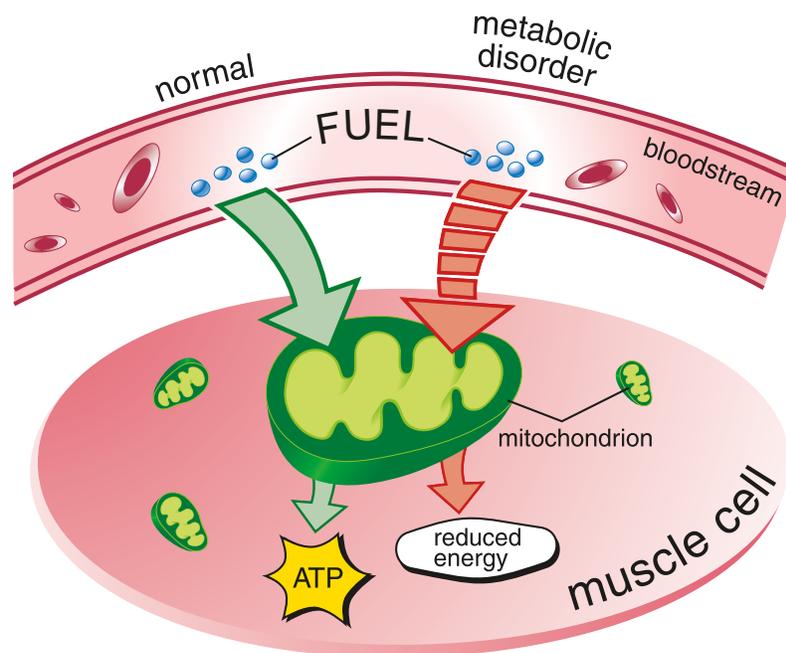
What happens to someone with a metabolic disease?

Exercise intolerance

The main symptom of most of the metabolic myopathies is difficulty performing some types of exercise, a situation known as *exercise intolerance*, in which the person becomes tired very easily.

The degree of exercise intolerance in the metabolic myopathies varies greatly between disorders and even from one individual to the next within a disorder. For instance, some people may run into trouble only when jogging, while others may have trouble after mild exertion such as walking across a parking lot or even blow-drying their hair. Each person must learn his activity limitations.

In general, people with defects in their carbohydrate-processing pathways tend to become very tired at the beginning of exercise but may experience a renewed feeling of energy after 10 or 15 minutes.



In normal metabolism, food provides fuel that's processed inside the cells, producing energy (ATP) for muscle contraction and other cellular functions. In metabolic myopathies, missing enzymes prevent mitochondria from properly processing fuel, and no energy is produced for muscle function.

A person with exercise intolerance also may experience painful muscle cramps and/or pain during or after exercise.

On the other hand, those with carnitine palmitoyl transferase deficiency (CPT) may experience fatigue only after prolonged exercise.

A person with exercise intolerance also may experience painful muscle cramps and/or injury-induced pain during or after exercising.

The exercise-induced cramps (actually sharp contractions that may seem to temporarily “lock” the muscles) are especially noted in many of the disorders of carbohydrate metabolism and, rarely, in myoadenylate deaminase deficiency. The injury-induced pain is caused by acute muscle breakdown, a process called *rhabdomyolysis*, which may occur in any metabolic muscle disorder and is particularly noted in CPT.

Episodes of rhabdomyolysis usually occur when a person with a metabolic myopathy “overdoes it” (sometimes unknowingly). These episodes, often described as “severe muscle pain,” may occur during exercise or several hours afterward. In those with carbohydrate-processing disorders, rhabdomyolysis may be triggered by aerobic exercise (such as running or jumping) or isometric exercise (like pushing or pulling heavy objects, squatting or standing on tip-toes). In people with CPT, rhabdomyolysis is usually brought on by prolonged, moderate exercise, especially if an affected person exercises without eating. In CPT, rhabdomyolysis may also be triggered by illness, cold, fasting, stress or menstruation.

Because rhabdomyolysis is painful and can cause extensive kidney damage, many people with metabolic muscle diseases try to avoid triggering these episodes by modifying their physical activities or diet. Your MDA clinic director can help you work out a lifestyle plan to optimize your health and abilities.

Muscle weakness

In acid maltase deficiency, carnitine deficiency and debrancher enzyme deficiency,

progressive muscle weakness, rather than exercise intolerance, is the primary symptom. Over time, people with acid maltase deficiency or debrancher enzyme deficiency may eventually need a wheelchair to get around and, as respiratory muscles weaken, may require ventilatory assistance to provide extra oxygen at night. All three of these disorders may be associated with heart problems.

It’s important to realize that, although the metabolic muscle diseases characterized by exercise intolerance typically don’t involve muscle weakness, some chronic or permanent weakness *can* develop in response to repeated episodes of rhabdomyolysis and to the normal loss of strength that occurs with aging. The degree of muscle weakness that develops in these disorders is extremely variable and may depend on such factors as genetic background and the number of episodes of rhabdomyolysis experienced. The diseases involving exercise intolerance don’t usually progress to the degree that a wheelchair or any other mechanical assistance is needed.

Special issues in metabolic disorders

- **Myoglobinuria:** Myoglobinuria refers to rust-colored urine caused by the presence of myoglobin (a muscle protein). When overexertion triggers acute muscle breakdown (rhabdomyolysis), muscle proteins like creatine kinase and myoglobin are released into the blood and ultimately appear in the urine. Myoglobinuria can cause severe kidney damage if untreated. Incidences of myoglobinuria should be dealt with as emergencies and may require intravenous fluids to avoid renal failure.

- **Emergencies:** The metabolic muscle diseases are so rare that emergency room staffs are frequently unfamiliar with them. As a result, they may not treat episodes properly (with fluids and pain medica-

For many with metabolic muscle diseases, the only treatment needed is to understand what activities and situations tend to trigger attacks.

tions) or may give the patient food or anesthesia that could trigger further problems.

People with these disorders may want to consider carrying a treatment “protocol” listing their doctor’s phone number, the patient’s current medications and dietary requirements, and guidelines for handling emergency situations. A MedicAlert bracelet also can be worn.

- **Anesthesia:** People with metabolic muscle disorders may be at higher risk for a potentially fatal reaction to certain common general anesthetics (typically combinations of halothane and succinylcholine). This reaction, called *malignant hyperthermia*, can be avoided in planned surgeries by using lower-risk anesthetics. However, it’s a good idea to wear a MedicAlert bracelet stating this susceptibility in case of an emergency.

- **Cardiac care:** People with debrancher enzyme deficiency, carnitine deficiency and acid maltase deficiency may develop significant heart problems. In the case of primary carnitine deficiency, the only symptom may be heart failure; however, this disorder responds well to carnitine supplementation. If you’re at risk for cardiac problems, a cardiologist who’s familiar with your disorder should monitor your heart function.

- **Respiratory care:** Acid maltase deficiency and debrancher enzyme deficiency tend to weaken the respiratory muscles, those that operate the lungs, meaning that a person with one of these disorders may require supplemental oxygen at some point. If you’re at risk for respiratory problems, your breathing should be monitored regularly by a specialist. Also, be conscious of symptoms such as unusual shortness of breath on exertion or morning headaches that may indicate that your breathing is compromised.

How are metabolic diseases of muscle treated?

In April 2006, the U.S. Food and Drug Administration (FDA) approved Myozyme, a synthetic form of the acid maltase enzyme, manufactured by Genzyme of Cambridge, Mass., for the treatment of Pompe disease.

The enzyme replacement therapy requires intravenous infusions of the drug, and has significantly improved survival in patients with infantile-onset Pompe disease. A December 2007 trial of Myozyme in patients who were at least 8 years old showed it improved walking endurance and respiratory function in that group as well.

For many other people with metabolic muscle diseases, the only treatment needed is to understand what activities and situations tend to trigger attacks of rhabdomyolysis. A small percentage of adults with metabolic disorders may experience painful muscle cramps that have no obvious triggers; painkillers and meditation techniques may be effective under these circumstances.

In addition, some people with metabolic disorders have benefited from dietary changes. There’s evidence that those with carbohydrate-processing problems may be helped by a high-protein diet, while those with difficulty processing fats may do well on a diet high in carbohydrates and low in fat. Carnitine supplements are usually given for carnitine deficiency and can be very effective in reversing heart failure in this disorder.

Please consult your doctor before undertaking any special diets. Your MDA clinic director can help you design a specific plan suited for your metabolic disorder and your individual needs.

There’s also emerging evidence that people with some carbohydrate-processing

It's important to get an accurate diagnosis of a specific metabolic myopathy so diet and exercise can be modified and symptoms monitored.

disorders, such as McArdle disease, may benefit from light exercise. Researchers believe that people who are physically fit are better able to use alternative fuel sources to make energy. Because over-exertion can trigger muscle breakdown, you should only undertake an exercise program under the supervision of a doctor who's familiar with your disorder.

It's unclear whether regular exercise is beneficial in the fat-metabolizing disorders, such as carnitine palmityl transferase deficiency.

Because of their rarity, the characteristics of several of these diseases aren't well known.

How are metabolic diseases of muscle diagnosed?

It's important to get an accurate diagnosis of a specific metabolic myopathy so the affected person can modify diet and exercise and monitor potentially serious disease effects. Because these diseases are rare, many people with metabolic disorders of muscle have spent some time trying to find out what caused their muscle weakness, myoglobinuria or other symptoms. The diagnostic process usually begins with a careful medical history, a physical exam and a neurological exam to test reflexes, strength and the distribution of weakness.

Several specialized tests are used to confirm a suspected diagnosis of metabolic disease:

Blood tests can be used to detect the presence of certain chemicals in the blood that may indicate some metabolic diseases.

An *exercise test* is used to monitor a person's response to intense or moderate exercise. Blood samples are taken during exercise for testing.

Electromyography (EMG) uses small needle electrodes to measure the electrical currents in a muscle as it contracts. While an EMG can't definitively diagnose metabolic disease, it can be used to rule out a number of other types of neuromuscular disease that cause similar patterns of weakness.

A *muscle biopsy* requires the removal of a small piece of muscle tissue for microscopic analysis. The procedure is done either surgically, with an incision to expose the target muscle, or with a needle. A *skin biopsy* also is sometimes performed.

Other tests that may be needed include an *electrocardiogram* to test heart function, and *brain imaging studies* such as CT or MRI scans.

Genetic tests, using a blood sample, can analyze the person's genes for particular defects that cause metabolic disease, but these tests often aren't necessary for diagnosis or for determining treatment.

What are the symptoms and characteristics of each type of disease?

CARBOHYDRATE-PROCESSING DISORDERS

These disorders affect the breakdown of glycogen or glucose (complex and simple carbohydrates) and also are called *glycogenesis disorders*.

Acid maltase deficiency

Also called:

Glycogenosis type 2, acid-alpha glucosidase deficiency, Pompe disease, lysosomal storage disease

Onset:

Infancy to adulthood

Inheritance:

Autosomal recessive

In April 2006, the U.S. Food and Drug Administration granted approval for the use of Myozyme as a treatment for Pompe disease.

Symptoms:

Causes slowly progressive weakness, especially of the respiratory muscles and those of the hips, upper legs, shoulders and upper arms. Enlargement of the tongue and liver impairment occur in the infantile form, but rarely in the older forms. Cardiac involvement may occur in the infantile or childhood forms, but is less common in adults. The childhood and adult-onset forms are less severe than the infantile form, but may cause severe weakness and respiratory insufficiency, and, without treatment, shortened life span. Untreated, the infantile form of Pompe disease often leads to death within the first year of life.

Until recently, there was no treatment of this condition and the only remedy was supportive medical care. In April 2006, the U.S. Food and Drug Administration granted approval for the use of Myozyme as a treatment for Pompe disease. The drug was developed by Genzyme Corporation of Cambridge, Mass., with support from MDA. It substitutes for the enzyme missing in Pompe disease and may keep muscle cells from dying.

Your MDA clinic director will keep you abreast of ongoing clinical trials for the disease and work with you to make the best decisions for care.

Debrancher enzyme deficiency

Also called:

Cori or Forbes disease, glycogenosis type 3

Onset:

Childhood to adulthood

Inheritance:

Autosomal recessive

Symptoms:

Principally affects the liver, causing swelling of the liver, slowing of growth, low blood sugar levels and, sometimes, seizures. In children, these symptoms

often improve around puberty. Muscle weakness may develop later in life, and is most pronounced in the muscles of the forearms, hands, lower legs and feet. Weakness often is accompanied by loss of muscle bulk. The heart can be affected as well, and heart function should be monitored closely.

Phosphorylase deficiency

Also called:

Myophosphorylase deficiency, McArdle disease, glycogenosis type 5

Onset:

Childhood to adulthood

Inheritance:

Autosomal recessive

Symptoms:

Causes exercise intolerance, cramps, muscle pain and weakness shortly after the beginning of exercise. A person with this disorder may tolerate light-to-moderate exercise such as walking on level ground, but strenuous exercise will usually bring on symptoms quickly. Resting may lead to a “second wind,” in which activity is then better tolerated. Isometric exercises that require strength, such as lifting heavy objects, squatting or standing on tiptoe, also may cause muscle damage.

The symptoms of McArdle disease vary in severity among people and even within the same person from day to day. Symptoms usually don’t persist between attacks, although fixed weakness later in life is possible.

Phosphofructokinase deficiency

Also called:

Glycogenosis type 7, Tarui disease

Onset:

Childhood to adulthood

Inheritance:

Autosomal recessive

A carbohydrate meal typically worsens exercise capacity in phosphofructokinase deficiency by lowering blood levels of fats.

Symptoms:

Causes exercise intolerance, with pain, cramps and, occasionally, myoglobinuria. Symptoms are very similar to those of phosphorylase deficiency, but people with this disorder are less likely to experience the “second wind” phenomenon.

A carbohydrate meal typically worsens exercise capacity in this condition by lowering blood levels of fats, which are the major muscle energy fuels for those with the disorder. A partial deficiency of phosphofructokinase in the red blood cells results in the breakdown of those cells and an increase in blood levels of bilirubin, though the person usually experiences no symptoms.

Phosphoglycerate kinase deficiency

Also called:

Glycogenosis type 9

Onset:

Infancy to early adulthood

Inheritance:

X-linked recessive

Symptoms:

May cause anemia, enlargement of the spleen, mental retardation and epilepsy. More rarely, weakness, exercise intolerance, muscle cramps and episodes of myoglobinuria also occur.

Phosphoglycerate mutase deficiency

Also called:

Glycogenosis type 10

Onset:

Childhood to early adulthood

Inheritance:

Autosomal recessive

Symptoms:

Causes exercise intolerance, cramps, muscle pain and, sometimes, myoglobinuria. Permanent weakness is rare.

Lactate dehydrogenase deficiency

Also called:

Glycogenosis type 11

Onset:

Early adulthood

Inheritance:

Autosomal recessive

Symptoms:

Causes exercise intolerance and episodes of myoglobinuria. A skin rash is common, probably because skin cells need lactate dehydrogenase.

FAT-PROCESSING DISORDERS

Carnitine deficiency

Onset:

Childhood

Inheritance:

Autosomal recessive

Symptoms:

This slowly progressive disorder causes cardiac disease and muscle weakness in the hips, shoulders, and upper arms and legs. The neck and jaw muscles may also be weak. Carnitine deficiency may occur secondary to other metabolic diseases (*secondary carnitine deficiency*) or in response to a genetic *mutation* (gene defect) in the protein responsible for bringing carnitine into the cell (*primary carnitine deficiency*).

Primary carnitine deficiency can often be treated successfully with carnitine supplements.

Carnitine palmitoyl transferase deficiency

Onset:

Childhood to early adulthood

Inheritance:

Autosomal recessive

Symptoms:

Symptoms usually are brought on by prolonged and intense exercise, especially

in combination with fasting, but may not appear for several hours after activity stops. Short periods of exercise usually don't provoke symptoms. Symptoms also can be brought on by illness, cold, stress or menstruation. This disorder causes muscle pain, stiffness and tenderness, while weakness is less common. Breakdown of muscle tissue during an attack can cause myoglobinuria.

DISORDER AFFECTING ATP RECYCLING

Myoadenylate deaminase deficiency

Onset:

Adulthood

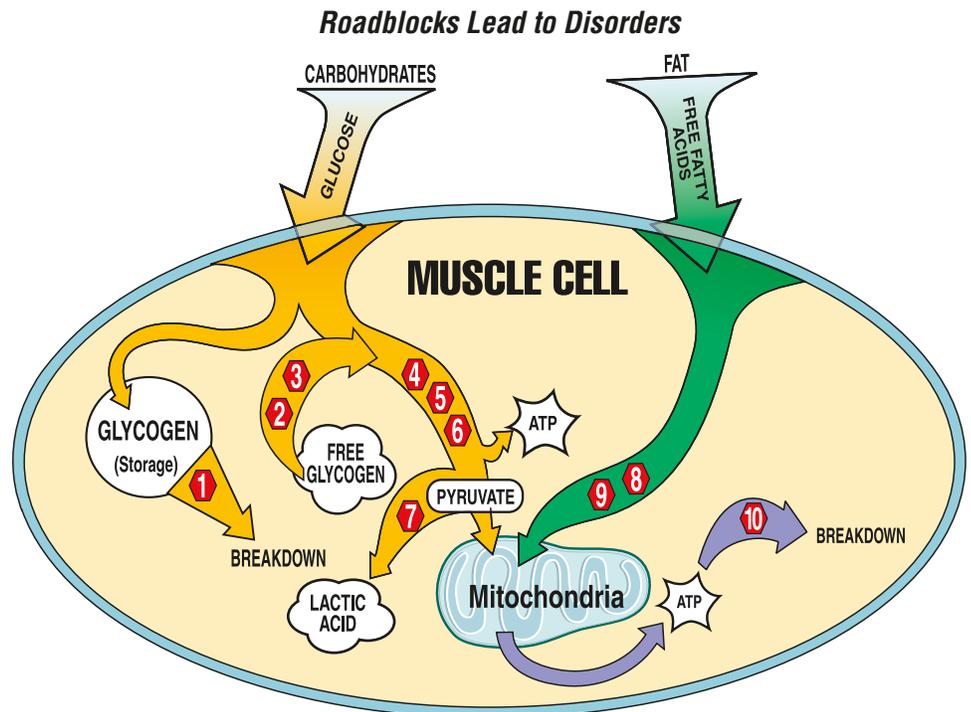
Inheritance:

Autosomal recessive

Symptoms:

Interferes with the recycling of the major energy molecule of the cell (called *ATP*). It may cause exercise intolerance, cramps and muscle pain, although, in many cases, people with deficiencies in this enzyme may experience no symptoms.

Fueling the Muscles



WHERE THE PROBLEMS LIE IN EACH DISEASE

1 Acid maltase deficiency	6 Phosphoglycerate mutase deficiency
2 Muscle phosphorylase deficiency	7 Lactate dehydrogenase deficiency
3 Debrancher enzyme deficiency	8 Carnitine palmityl transferase deficiency
4 Phosphofructokinase deficiency	9 Carnitine deficiency
5 Phosphoglycerate kinase deficiency	10 Myoadenylate deaminase deficiency

Skeletal muscles normally depend on energy from carbohydrates and fats. These fuels can be stored in the muscle (glycogen) or imported directly from the bloodstream (glucose and fatty acids). When a genetic defect (●) interferes with the processing of specific fuels, energy shortages can occur and toxic byproducts may build up. Some people may be able to bypass their defects by adjusting diet or exercise to draw energy more efficiently from unaffected pathways.

Many parents have no idea they're carriers of a disease until they have a child who has the disease.

Does It Run in the Family?

On being told they have a genetic disorder such as a metabolic muscle disease, bewildered patients often ask, "But it doesn't run in the family, so how could it be genetic?"

Metabolic myopathies *can* run in a family, even if only one person in the biological family has it. This is because of the ways in which genetic diseases are inherited.

Most of the metabolic diseases of muscle are inherited in an *autosomal recessive* pattern, meaning that a person needs two defective genes in order to have the disease. One copy is inherited from each parent, neither of whom would normally have symptoms.

Thus, the disease appears to have occurred "out of the blue," but in reality, both parents may be *carriers*, silently harboring the genetic *mutation* (a flaw in the gene). Many parents have no idea they're carriers of a disease until they have a child who has the disease.

Other metabolic disorders have *X-linked* or *autosomal dominant* patterns of inheritance, each of which carries different risks for transmission to children. In some cases, a single disorder is associated with more than one pattern of inheritance.

Finally, metabolic disorders actually can occur "out of the blue" when a new mutation appears with a baby's conception. These are called *spontaneous mutations*, and, after they occur, they can be passed on to the next generation.

The risk of passing on a metabolic myopathy to your children depends on many circumstances, including exactly which type of metabolic disease has been diagnosed.

A good way to find out more about these risks is to talk to your MDA clinic physician or ask to see the genetic counselor at the MDA clinic. Also, see MDA's booklet, "Facts About Genetics and Neuromuscular Diseases."

MDA's Search for Treatments and Cures

The MDA website is constantly updated with the latest information about the neuromuscular diseases in its program. See the latest research news at www.mda.org.

MDA-funded scientists are pursuing a number of promising leads in their quest to understand the causes of the metabolic diseases of muscle.

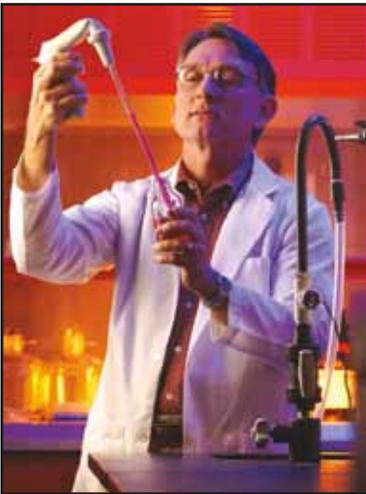
To date, scientists have isolated all of the genes involved in the metabolic myopathies described in this booklet, and their genetic codes have been unraveled, offering insight into how particular gene defects lead to disease. In addition, isolation of genes has allowed researchers to begin experiments with gene therapy, a potential cure for some metabolic diseases.

The knowledge MDA-funded researchers are obtaining about the mechanisms by which metabolic gene defects cause disease may lead to other strategies for prevention and treatment. MDA's extensive research program includes studies to help in developing new treatments for metabolic diseases of muscle, such as strategies designed to replace the enzymes that are missing in these disorders.

Efforts to develop such "enzyme replacement therapies" already have been rewarded with the U.S. Food and Drug Administration's approval in 2006 of Myozyme, a laboratory-modified acid maltase enzyme developed by Genzyme of Cambridge, Mass., with the support of

MDA, for the treatment of Pompe disease. Since Myozyme became available, the outlook for people of all ages with Pompe disease is considerably brighter, though improvement of the drug continues to be the focus of ongoing research.

Targets now being pursued in research include: better diagnosis of metabolic diseases, to allow better identification of at-risk individuals and earlier treatment; continued examination of the role of exercise and diet in metabolic diseases; development of animal models of metabolic diseases, both to improve understanding of the diseases and to test possible treatments; development of enzyme replacement therapies in addition to Myozyme for enzyme deficiencies; and development of gene therapies for metabolic diseases.



MDA Is Here to Help You

The Muscular Dystrophy Association offers a vast array of services to help you and your family deal with metabolic diseases of muscle. The staff at your local MDA office is there to assist you in many ways. The Association's services include:

- nationwide network of clinics staffed by top neuromuscular disease specialists
- MDA summer camps for kids with neuromuscular diseases
- help with locating durable medical equipment through its national equipment program
- financial assistance with repairs or modifications to all types of durable medical equipment
- annual occupational, physical, respiratory or speech therapy consultations
- annual flu shots
- support groups for those affected, spouses, parents or other caregivers
- online support services through the e-community *myMDA* and through *myMuscleTeam*, a program that helps recruit and coordinate in-home help

MDA's public health education program helps you stay abreast of research news, medical findings and disability information through magazines, publications, educational speakers, seminars, videos and newsletters.

MDA's website at www.mda.org contains thousands of pages of valuable information, including disease specifics, research findings, clinical trials and past magazine articles.

Everyone registered with MDA automatically receives *Quest*, MDA's award-winning quarterly magazine. *Quest* publishes detailed articles about research findings, medical and day-to-day care, helpful products and devices, social and family issues, and much more. Other MDA publications can be found at www.mda.org/publications; many booklets are available in Spanish. Ask your local office for "MDA Services for the Individual, Family and Community" and for help with obtaining copies of other publications.

If you have any questions about metabolic diseases of muscle, someone at MDA will help you find the answer. To reach your local MDA office, call (800) 572-1717.



On the cover:

Lateef, who has Pompe disease, composes and produces music in his home studio. He moved from Michigan to California to pursue his music career. He credits MDA "for being there for me. I knew I could come out here and still get the great care and treatment I received in Michigan."

MDA's Purpose and Program

The Muscular Dystrophy Association fights neuromuscular diseases through an unparalleled worldwide research effort. The following diseases are included in MDA's program:

Muscular Dystrophies

Myotonic dystrophy (*Steinert disease*)
Duchenne muscular dystrophy
Becker muscular dystrophy
Limb-girdle muscular dystrophy
Facioscapulohumeral muscular dystrophy
Congenital muscular dystrophy
Oculopharyngeal muscular dystrophy
Distal muscular dystrophy
Emery-Dreifuss muscular dystrophy

Motor Neuron Diseases

Amyotrophic lateral sclerosis (*ALS*)
Infantile progressive spinal muscular atrophy
(*Type 1, Werdnig-Hoffmann disease*)
Intermediate spinal muscular atrophy
(*Type 2*)
Juvenile spinal muscular atrophy
(*Type 3, Kugelberg-Welander disease*)
Adult spinal muscular atrophy (*Type 4*)
Spinal-bulbar muscular atrophy
(*Kennedy disease*)

Inflammatory Myopathies

Polymyositis
Dermatomyositis
Inclusion-body myositis

Diseases of Neuromuscular Junction

Myasthenia gravis
Lambert-Eaton (myasthenic) syndrome
Congenital myasthenic syndromes

Diseases of Peripheral Nerve

Charcot-Marie-Tooth disease
Friedreich's ataxia
Dejerine-Sottas disease

Metabolic Diseases of Muscle

Phosphorylase deficiency (*McArdle disease*)
Acid maltase deficiency (*Pompe disease*)
Phosphofructokinase deficiency
(*Tarui disease*)
Debrancher enzyme deficiency
(*Cori or Forbes disease*)
Mitochondrial myopathy
Carnitine deficiency
Carnitine palmityl transferase deficiency
Phosphoglycerate kinase deficiency
Phosphoglycerate mutase deficiency
Lactate dehydrogenase deficiency
Myoadenylate deaminase deficiency

Myopathies Due to Endocrine Abnormalities

Hyperthyroid myopathy
Hypothyroid myopathy

Other Myopathies

Myotonia congenita
Paramyotonia congenita
Central core disease
Nemaline myopathy
Myotubular myopathy
Periodic paralysis



MDA's website, mda.org, is constantly updated with the latest research news and information about the diseases in its program. Follow MDA on Facebook, Twitter and YouTube.



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