

McArdle Disease

What is McArdle Disease?

McArdle Disease is a metabolic disorder that affects your muscles. 'Metabolic' means that there is a problem with the chemical reactions that create energy in your muscles.

McArdle Disease is also called:

- Glycogen Storage Disease Type V
- Myophosphorylase Deficiency
- McArdle Myopathy

In healthy muscles cells, a chemical reaction uses an enzyme (phosphorylase) to help break down carbohydrates stored in the muscles (glycogen). The reaction releases energy that can be used for movement.

With McArdle Disease your body is unable to make this enzyme, so this chemical reaction cannot happen. As a result, your muscle cells are not able to use glycogen as 'fuel' to get energy. This causes fatigue and cramping when you exercise.

What are the signs and symptoms?

The most common symptom of McArdle Disease is **muscle cramping** during exercise, which may last for minutes. However, this can be followed by muscle pain/discomfort for hours or days.

Other signs and symptoms include:

- Muscle weakness – getting tired quickly, usually within 1 or 2 minutes of normal activity.
 - Getting a 'second wind'. After a few minutes of exercise, when you feel tired, stop and rest for 5 to 10 minutes. Then you may notice a 'second wind' - another surge of energy that lets you continue exercising normally.
 - Myoglobinuria (tea- or cola-coloured urine). Strong exercise can break down muscle cells, causing their contents (myoglobin) to enter the blood stream. The kidneys cannot filter all of it out and some gets into the urine, giving the urine a burgundy colour. A long or severe bout of myoglobinuria can lead to kidney failure.
-

Not all people with McArdle Disease are affected in the same way.

Some people have mild symptoms. For example, they may have noticed since childhood that they get tired easily with exercise. It may just seem that they have trouble 'keeping up' or are 'out of shape'. This makes the disease difficult to diagnose. Most people do not find out they have McArdle Disease until they are 20 or 30 years old. However, as more doctors recognize this disease, it is not as rare as it was once thought to be.

How do you know I have McArdle Disease?

Your doctor or specialist can tell if you have McArdle Disease from:

- talking with you about your symptoms and family history
- a physical examination
- reviewing the results of the following tests

Blood and urine tests

- If muscles have broken down after exercise, there may be myoglobin in the urine and elevated muscle enzymes (CK) in the blood.

Muscle biopsy

- A biopsy is a procedure to take a tiny sample of muscle to be examined under a microscope.
- This procedure is done in the clinic and takes about 20 minutes. The sample is usually taken from your thigh muscle (quadriceps). First the area is "frozen" or numbed with a local anesthetic. Then the doctor makes a tiny cut (incision) and removes a sample of muscle with a needle. The incision is closed with a stitch that will need to be removed in a few days.

Forearm exercise testing (ischemic forearm test)

- A small plastic catheter is placed into a vein in your forearm to take a blood sample. Then, you exercise your forearm muscles for 1 minute. Blood tests to measure the amount of lactate and ammonia are done before and after exercise. The level of lactate does not increase, but ammonia increases by a higher than expected amount in people with McArdle Disease.
-

How will having McArdle Disease affect my life?

Having McArdle Disease does not change your life expectancy. The health care team at the Neuromuscular and Neurometabolic Centre can give you the help, information and support you need to live well with McArdle Disease. With certain lifestyle changes, you can prevent or lessen symptoms. This may also help to prevent the disease from getting worse.

Activity

Staying active is important. Keeping fit is the best way to condition your muscles and live normally. It is important to start an exercise program at a low intensity (effort) and to very gradually increase the duration (time) and intensity of activity as your symptoms allow. We have an exercise DVD that can help you with exercise training.

About 10 to 15 minutes before physical activity, have a drink that contains sugar. Research suggests that this may help prevent muscle damage. Always start activity slowly and take rest breaks often. Learn to listen to your body. If you feel tired, stop and rest. Do not over do it! You may gradually increase the intensity and duration of exercise, but never start off exercising strenuously.

It is most important to avoid strenuous exercise, especially:

- **brief, intense exercise such as carrying something heavy**
- **long periods of exercise such as climbing a long hill**

Intense or strenuous exercise can lead to muscle pain and the dark red urine (myoglobinuria) that is a warning sign of muscle breakdown. If you develop myoglobinuria, go to the nearest hospital emergency room as you may need treatment with intravenous fluids.

Nutrition

- A healthy diet is also important. Make sure you get enough protein and carbohydrates. If you find a sugary drink (within 10 to 15 minutes of exercise) helps lessen symptoms, you will need to plan for these extra calories so you do not gain weight. Refer to **Eating Well with Canada's Food Guide** to learn how to eat well. Ask your family doctor for a copy or visit Health Canada's website: www.hc-sc.gc.ca/fn-an/food-guide-aliment/index_e.html
- A supplement of creatine monohydrate may also help relieve symptoms. Take ½ tsp (2ml) in the morning and ½ tsp (2ml) in the evening. Do not take more than this amount (more is not always better). You can add it to a glass of juice or sprinkle it on cereal or yogurt. We also suggest taking a supplement of Vitamin B6 (50 mg once a day).

How did I get McArdle Disease?

McArdle Disease is inherited. For you to get the disease, one copy of the abnormal gene was passed down from each of your parents. This can happen even when they do not have the disease. Your parents may be “carriers”, meaning they have the gene for McArdle Disease, but they do not have the disease. This type of inheritance is called “autosomal recessive”.

Can I pass this disease to my children?

You will pass one copy of the abnormal gene to each of your children. So all of your children will be carriers. A child will only get the disease if your partner is also a carrier of McArdle Disease and passes a second abnormal gene to him or her. This occurs very rarely.

**If you have any questions about McArdle Disease,
please speak with your doctor or nurse
at the Neuromuscular and Neurometabolic Centre.**