MCARDLE'S DISEASE

MUSCULAR DYSTROPHY CANADA
DYSTROPHIE MUSCULAIRE CANADA

What is McArdle's Disease?

McArdle's Disease is a rare, inherited condition that causes severe muscle pain and cramping. It is caused by the inability to produce an enzyme known as phosphorylase or myophosphorylase, which is needed to break down glycogen, the stored form of sugar. Without the enzyme, glycogen can't be used to produce energy during exercise. The condition is also known as Glycogen Storage Disease Type V (GSD V), Phosphorylase Deficiency, and Myophosphorylase Deficiency.

What are the symptoms?

People with McArdle's Disease experience severe pain in their muscles during the first few minutes of exercise. Exercising through the pain can trigger severe muscle spasms or contractures, and lead to muscle damage. Muscle weakness and stiffness are also experienced.

The disease affects skeletal muscles. Symptoms in the legs can occur from activities such as walking. Carrying heavy objects can trigger symptoms in the arms. Other activities such as sawing or digging may affect the back muscles. Some people develop pain in their jaw muscles while chewing.

What is the age of onset?

McArdle's Disease is present from birth, but may not be diagnosed until young adulthood. People with McArdle's Disease often report they had symptoms such as painful muscle spasms during childhood. People often dismiss the muscle pain as being a sign of being 'out of shape' and don't pursue medical follow-up until symptoms are more severe.

How is McArdle's disease diagnosed?

Diagnosis begins with a blood test to check whether a muscle enzyme, creatine kinase (CK), is at high levels. This indicates muscle damage. In the past, muscle biopsy – the removal of a small amount of muscle tissue for examination - was the primary way of diagnosing this condition. Thanks to advances in research, McArdle's Disease can now be diagnosed through genetic testing.

What are the complications of McArdle's Disease?

The most common and potentially serious complication is the breakdown of skeletal muscle tissue. Because people with the disease lack the enzyme to metabolize glycogen (the main source of energy for muscle during activity),

the body turns to other sources for energy. In fact, muscle tissue itself is broken down and used as fuel, a process called rhabdomyolysis. This leads to muscle pain, cramping, fatigue, and muscle tenderness.

When the muscle is broken down, it releases the red protein myoglobin. As a result, urine becomes a dark-red or red-brown in colour. Large amounts of myoglobin can cause temporary kidney failure.

The majority of people with McArdle's Disease will not become disabled. Normally, there is weakness associated with the loss of muscle tissue, but the majority of people remain independent and able to walk. McArdle's Disease does not affect life expectancy.

How is McArdle's Disease inherited?

McArdle's Disease is a recessive genetic disorder, meaning both parents must have a copy of the defective gene. If both parents are carriers, there is a 25% chance in each pregnancy that a child will inherit the disease. If only one parent has the defective gene, then the child is likely to become a carrier. Carriers generally do not have symptoms, but they may pass the gene on to their children. People with a family history may choose to undergo genetic counseling to help understand the risks of passing the gene on to their children.

What treatments are available for McArdle's Disease?

There is no cure for McArdle's Disease. The symptoms can be kept under control by learning techniques for doing exercise, and staying fit. This is done by working with a physiotherapist. Regular moderate exercise, such as walking is recommended. If pain occurs during exercise, slow down or stop until the pain has subsided. Once the pain is gone, try again. Often people with McArdle's Disease will experience a 'second wind', and will be able to exercise more after a short break. Intense exercise such as sprinting or weight lifting should be avoided because of the likelihood of muscle damage.

Special considerations

Health care providers need to be made aware of a diagnosis of McArdle's Disease. Some anaesthetics used during surgery can lead to acute muscle damage or other complications.