McArdles Disease

McArdles disease is the type 5 glycogen storage disease which is caused by a defect in skeletal muscle glycogen phosphorylase. Some glycogen is stored in skeletal muscle and the phosphorylase enzyme is necessary to mobilize it during metabolism. In McArdles, patients are unable to break down skeletal muscle glycogen during times of exercise. This results in painful muscle cramps and muscle breakdown during vigorous exercise. After episodes of vigorous exercise, patients can have myoglobinuria. However, patients have normal blood lactic acid levels because the enzyme is only found in skeletal muscle and does not severely affect the body's overall metabolism.



PLAY PICMONIC

Pathophysiology

Type V Glycogen Storage Disease
(5) Hand with Glider
McArdles disease is the type 5 glycogen storage disease.

Muscle Glycogen Phosphorylase Deficiency
Broken Muscle Glider with Phosphorus-P-lace
This enzyme is essential for the first step in glycogen breakdown. If deficient, glycogen will be unable to be mobilized as an energy source when needed.

Signs and Symptoms

Increased Glycogen in Muscle
Up-arrow Glider in Muscle-man
Without glycogen phosphorylase, glycogen is trapped in the skeletal muscles.

Painful Muscle Cramps
Pain-bolt at Muscle Clamp
Painful muscle cramping results from insufficient metabolic supply of glucose.

Myoglobinuria
Mayo-globe-urinal
Without enough glucose, muscles begin to breakdown and release myoglobin into the urine. This can result in kidney damage due to myoglobin's noxious effects on the kidney. Patients display dark-colored, brownish urine.

Normal Lactic Acid
Normal-bell-curve at Lake Acidic-lemon
Because McArdles is localized to only skeletal muscles, the body's overall metabolism is not severely affected. Therefore, lactic acidosis does not severely build up.

Treatment

Vitamin B6
Viking Bee with (6) Sax
Patients with McArdle's disease have a relative Vitamin B6 deficiency, and research suggests that B6 supplementation helps to reduce muscle fatigue. This occurs by enhancing any one of a number of vitamin B6 dependent proceses that increase energy production.

Sign up for FREE at picmonic.com