Glycogen Storage Disease V – McArdle Disease

Other names that may be used for this condition are:
- GSD V
- Glycogenosis Type V
- McArdle Disease
- Muscle Glycogen Phosphorylase Deficiency
- Muscle Phosphorylase Deficiency
- Myophosphorylase Deficiency
- PYGM Deficiency

McArdle Disease is a rare metabolic muscle condition belonging to a group known as Glycogen Storage Diseases (GSDs). This form of GSD is caused by a deficiency of the enzyme known as myophosphorylase (muscle phosphorylase) which is needed to break down glycogen into glucose (sugar) needed for energy.

Glycogen is stored in the muscles and is converted through a pathway known as glycolysis into glucose which is used for anaerobic exercise. It is then converted to ATP which allows the muscles to contract. Anaerobic exercise is classed as strenuous exercise, squatting, running, lifting heavy weights and tensing of the muscles. A second pathway is used for aerobic exercise this known as beta oxidation and requires free fatty acids which are formed in the blood from breaking down fat. This also forms ATP. Aerobic exercise is light exercise such as walking.

Treatment for this condition includes avoiding strenuous exercise; however aerobic exercise or light exercise may improve exercise tolerance. It is important that exercise is done safely. Speed should be paced for the first few minutes. When the muscles become tired or discomfort occurs it is important to slow down or stop and wait for the discomfort to improve before continuing gentle exercise. It is recommended that those affected by this disorder do gentle aerobic training for at least 30 minutes, three times a week to improve exercise tolerance. Myoglobinuria should be avoided through pacing exercise and avoiding lifting heavy weights, strenuous activity and tensing muscles. If myoglobinuria does occur and the urine has a slight reddish colour it is advised that the individual drinks plenty of water to flush it through the kidneys. If needed painkillers may be taken but should not be taken routinely. If heavy myoglobinuria is present, the urine will be a dark red, or coca cola colour, and those affected may feel unwell, suffer from vomiting, fever and may even collapse. Those affected should go to hospital immediately where they will receive fluid and pain relief through a drip (intravenously). Urine tests will be given to monitor signs of kidney failure. If this occurs dialysis may be required for a period of time.

Other treatments may include oral glucose immediately before planned exercise. A healthy balanced diet is advised. Excessive weight gain or dieting should be avoided. In the case of general anaesthetics being required it is important to make the
anaesthetist aware of the condition as there is a potential risk of muscle breakdown (rhabdomyolysis).

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This information is fully sourced and referenced, for more detailed information and references please contact CLIMB by email, letter or telephone.

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