

2003 Workshop - Type V

2003 Workshop Report for Type V from the 2003 AGSD(UK) Conference held at the Novotel near Bradford on Saturday 11th October 2003

We started by talking briefly about Nick Owston and his considerable work for AGSD Type V's over the years. He will clearly be greatly missed.

We then went around the room introducing ourselves to each other and listening to each other's synopsis of life with McArdles. Common traits as ever emerged – long and drawn out diagnosis, muscle cramps, fatigue, peoples lack of understanding etc

Six diagnosed Type V's attended and one Type IX (Phospholayse Kinase deficiency). Type IX manifests itself in similar ways to McArdles but is even more rare and mainly affects males.

Those present on the day:

Bill Rome
Linda Osbourne
Sue Fox
Vanessa Gatsby
Andrew Wakelin
William Gibbs

Type 9 present (Phospholayse Kinase deficiency)
Mark

JOHN BUCKLEY, EXERCISE SPECIALIST AT OSWESTRY KINDLY AGREED TO SPEAK AND FIELD QUESTIONS

John began by describing the relationship between the "motors" found inside muscle tissue, these being either aerobic or anaerobic.

People with McArdles are not capable of anaerobic exercise, as we inherently lack glycogen phosphorolayse, which turns glycogen in to glucose. This should in turn produce lactic acid, once again absent in McArdles.

This is where most damage takes place (when there is a breakdown in the anaerobic process), quantified in the form of high CK levels, and it is this process that brings on attacks of myoglobinuria. (Myoglobinuria – results from necrosis, especially acute, of muscle fibres (rhabdomyolysis). Features include muscle swelling, weakness and pain as well as "tea coloured" pee.) (CK, or CPK, stands for creatine phosphokinase – an enzyme that is involved in muscle metabolism. It affects the skeletal muscles, the heart (cardiac muscle), and the tissues of the brain. Injury to these cells results in leakage of CPK into your bloodstream. It is not unheard of for people with McArdles to have readings of CPK, taken from blood samples, resting in the low thousands on a normal day and after severe attacks of myoglobinuria even towards the hundred thousand mark. As a comparison, a healthy adult's CK level rests at anywhere between 30/170 daily)

If you have McArdles it does not mean you are unable to exercise. It means that to exercise comfortably, efficiently and without doing damage you must exercise in a very specific way. Nobody with McArdles, however well conditioned through aerobic exercise, is capable of for example, walking out of their house and immediately walking up a steep incline, as this requires the anaerobic motor to work. Even if through aerobic exercise one has reached second wind, walking up a sudden steep incline is likely to be difficult as the relationship between anaerobic and aerobic exercise is incredibly sensitive and this would require the use of the anaerobic motor.

The most effective way of training the muscles is through aerobic exercise. This traditionally, as performed at the Oswestry (McArdles) clinic, takes place in the form of walking on a treadmill at a slow pace or bicycling gently, until in second wind. By walking at a slow place you avoid the need to use the anaerobic motor and therefore avoid damaging the muscles. Almost all McArdles patients are able to achieve this second

wind. By the time you reach second wind the aerobic process is driving the muscle and it is possible to increase the pace and exercise more “normally”.

The anaerobic process (Glycogen driven) – needs glycogen phosphorylase, absent in McArdles, to turn glycogen in to glucose and power the muscle.

The aerobic process requires the metabolism of free fatty acids and an increase in muscle blood flow, which takes 3-4 minutes to get to the muscle hence start slowly and draw on the other forms of energy used by muscles.

John explained the make up of the Oswestry clinic. Ross Quinlivan is assisted by a dietician, physiotherapist, specialist nurse, family care officer and an exercise specialist (John Buckley). They are hoping to demonstrate that regular exercise is beneficial – ideally this would happen by encouraging people exercise regularly three times a week. We are hoping to see an improvement on our exercise assessment at subsequent clinic appointments. Professor Slonim has already done a lot of work in this area and his results are very encouraging.
Ideally, and more realistically, this would happen by encouraging people to attend their own local physiotherapist department 2/3 times a week to test their performance and CK levels when exercising and then when not exercising.

The issue of fatigue was discussed. As a group we established that exercise helps. Sue Fox told us that she suffered from fatigue but volunteered that this could be due to outside influences. Andrew Wakelin, who famously walks miles per day in the Welsh mountains and is one of the better conditioned people who has McArdles worldwide, said that if he doesn't exercise he doesn't feel like exercising. The group seemed to feel this was common ground.

We discussed glucose drinks taken before exercise as a source of energy. John Buckley advised that this was NOT a wise idea as people with McArdles we may get a 20% boost but the rest would be turned in to fat. Taking on sugar before exercise results in the insulin rebound effect – when glycogen stores are sufficient. This applies to everyone.

John went on to explain that the growth hormone helps repair tissue damage and aids performance, hence top athletes use it. Growth hormone is normally released by the pituitary gland when we are asleep or under stress. Exercise can be a potent stimulant from the brain to release growth hormone. Oswestry have recently measured growth hormone production in patients while they are exercising. Interestingly very low or absent levels were produced by McArdle's patients compared with Olympic athletes. We do not know why this happens and further research will be needed to establish why this happens.

John talked about doing a kreative supplementation trial for people with McArdles. Kreative supplies energy for post six-second activity. For example, the sprinter who wins the Olympic 100 metres has the highest levels of Kreative post six seconds. Would this work for us?

We talked about certain drugs not being suitable for people with McArdles. Mark (Type IX) noted that his problems came to the fore when taking Lariam. It was thought that this might be due to the fact that Lariam can as a side effect cause muscle weakness and depression so this would have exasperated his problem. i.e. Lariam reduces the capacity of normal people so it affects people with McArdles worse.

Following on from that we discussed the fact that certain anaesthetics were not suitable for people with McArdles and may result in malignant hyperthermia. (Malignant hyperthermia is an uncommon and sometimes life threatening reaction to some anaesthetic agents. Extremely high fever, high heart rate, and severe metabolic acidosis are part of the reaction. Just because prior anaesthetics have been uneventful does not mean that MH will not occur so always consult your doctor.) The drug Dantrolene is an effective antidote and treatment for this condition. Malignant hyperthermia appears to be rare in McArdle's disease, but there are a few published reports. It is important to let your anaesthetist know that you have a muscle condition and they will take the

Finally, while exercising it was noted that maximum heart rate achieved should be 200 minus age of the person exercising. Definitions in italics taken from the Muscular Dystrophy Website in the USA.

At present the McArdles clinic at Oswestry has undertaken trials in :

Randomised controlled trial of vitamin B6
Audit of pregnancy and delivery in McArdle's disease
Development and evaluation of 12-minute shuttle test
Cochrane systematic review of treatment in McArdle's disease
Assessment of Growth hormone release with exercise
Empirical use of creatine (not a formal research trial because of a lack of resource)
They are about to undertake a pilot study of brain MRS, this follows an earlier start with 2 patients a few years ago. We hope to recruit 10 patients and controls.

Any volunteers please!

In conclusion, we had a constructive meeting on the day and next year we hope that even more people turn up and listen to our various speakers. More details will be released closer to the time. In the meantime anyone reading this who needs any guidance or help with McArdles, especially in the UK, please do not hesitate to contact myself William Gibbs at gibbs_William@btopeonworld.com.