2001 Workshop - Type V

McArdles Workshop Birmingham 2001

Introduction

Nick Owston, the McArdles representative on the AGSD(UK) Committee introduced himself and John Buckley (Exercise Physiologist from Oswestry Team-see below) and explained about his own history and how the condition had progressed to his present age of 54. Each of the eight McArdleytes present then gave a brief outline of their own progression, experiences and shared symptoms. As we went round the table of eight people it became apparent that there were a number of common features some of which were:

- Late diagnosis; most people had got through school without being diagnosed.
- Dislike of games teachers/ PE instructors and the like; most people had been regarded as lazy
- Most achieved a second wind and immediately understood what that expression meant
- Of the women present most had had children and by normal delivery
- Most found that gentle sustained and controlled exercise was the best therapy for the condition.
- Most agreed that it was all too easy to attribute everything to McArdles (both themselves and their doctors) and one had to bear in mind that other unpleasant conditions could arise quite independently.
- All agreed that there were trials and tribulations in life which were just made harder by McArdles and that it was difficult to get other people to understand what the problem was and just how restricting it could become in some circumstances and yet in other circumstances and subject to other conditions it might be quite manageable.

The introductory explanation made the rest of the session more meaningful and there was a noticeable feeling of common identity and shared problems.

John Buckley's Presentation

John Buckley is an Exercise Physiologist. He is a partner of The Lifestyle Exercise & Physiotherapys Centre, Shrewsbury. He is a Research Fellow (PhD Candidate)/ Associate Lecturer in the Department of Physiotherapy Studies, Keele University, STAFFS.

John explained that the study of people who had been correctly diagnosed as suffering from McArdles Disease was quite a large survey considering the rarity of the condition (which he later estimated as one in a million -but that seems to be from UK information about the incidence of diagnosis).

Twenty people had been involved in the study and some conclusions could be drawn. Those might not be earth shattering from the point of view of the sufferers but they did begin to provide a uniform landscape for management and a database from which to build.

John started by giving a brief nonscientific explanation of the way fuel and energy are used in normal people and how the phosphorylase deficiency in McArdles impinges on that normal formulation.

There are three types of power -three sources of drive, they cover different periods of time and they should interrelate to each other.

1) Immediate term.
This lasts for six to ten seconds and produces / releases immediate ATP (adenosine tri-phosphate)
2) Mid-term Sustenance.  
This lasts from about six seconds into the exercise sequence until about two to four minutes and it is achieved by the carbo/glycogen system.

3) Long Term  
This is the fat/glucose aerobic source and is ever enduring.

For the purposes of understanding people with McArdles we need to concentrate on the activities going on in items two and three and the interaction between all three. If one looks at the two power sources in (2) and (3) [you have to label them if one is not working with pictures]. (2) fails because of the myophosphorylase absence and (3) should in theory work because all it needs is fat and glucose. But to work normally the system requires:-

* A small amount of glucose to prime the fat pump power source. 
Oxygen too and the only supply of this is what the body has left available coupled with what the heart can pump.

**Recommended Therapy – Haller**

This falls into two parts, Exercise Training and Dietary Control.<BR><BR>(1) Exercise Training (2) Dietary Control

(a) Overall fitness Practical physical activity strategies  
(b) Increased oxygen delivery  
(c) Blood-born free fatty acids  
(d) Glucose

**Benefits**

It enhances fat metabolism  
It reduces glycogen demand  

BOTH (1) and (2) help promote an early second wind.

Before the oswestry team start advising on an exercise activity plan they:-

Assess individual ability.  
Consider the perception of the McArdleyte vis a vis their own capabilities.  
Consider that person's individual quality of life (what they had come to tolerate: -conditioning: activity, exercise, work and daily demands)  
NB. "Individual" means just that; it differs from person to person and it is a private matter.

**Thinking about exercise therapy**

Over exertion causes myoglobinuria with all that that entails. HOW DO YOU CONTROL THIS?

Establish a meaningful cap on limits.  
Lose weight if you need to. Carrying extra baggage is making extra work for under-performing muscles.

What was evaluated at oswestry?

Twenty sufferers were asked to exercise for a period of twelve minutes and to go as far as they could during that time but on no account to go over a pain threshold of number Four on a scale of One to Ten. Number Four is a point where the McArdles sufferer will be aware of discomfort and some restriction but it would not have taken the form of real pain. If they had to slow down or stop to avoid going beyond that threshold then they had to do just that. Their pulse rates were monitored at the same time.
Everybody who participated did it from a resting state. In other words there was no warm up period -which is so useful to a McArdles sufferer. This meant that like was being compared to like.

In the twelve minutes the distance covered by the participants varied from 195 meters to 1180. Some had to stop, all slowed down and some clearly began to develop a second wind. No muscle contractures were caused and none were reported prior to the exercise (for a two hour period). Many of the participants were pleased with their performance.

**Conclusions which can be drawn**

- Age and weight made no difference
- Male or female made no difference
- Genetic defect (whether R49X or something else) or whether heterozygous or homozygous made no difference

BUT other genetic factors and the general state of fitness could have made a difference as could the extent of activity that the candidate was conditioned to.

Various graphs were illustrated to record relative performances.

**AS A GENERAL OBSERVATION:**
- People who did not think they would do well sometimes did do well. People who thought they would do well -did so.

The speed that people moved at varied but there was a co-relation between speed, pulse and pain factors which may well lead to a better understanding of exercise techniques and the whole business of achieving a second wind. At around six minutes nearly everybody slowed down, their "pain " levels peaked and their pulse rates also peaked (subject to a last minute spurt from some people -when the saw the end in sight)

John Buckley felt that increases in pulse rate might be associated with pain so that it could be used as a guide for people exercising. In other words once the individual had been assessed it might be possible for the team to assign parameters of exercise and pulse rate which should not be exceeded. The individual could then exercise but not let the pulse rate go above the agreed datum (by slowing down) and thus avoid any serious pain and, much more importantly, any serious damage.

A slight increase in CK (creatine kinase) levels had been recorded but most participants started with a relatively high level in any case. A five per cent increase was discernible. There was no evidence of myoglobin attributable to the exercise evaluation.

Various other tentative CONCLUSIONS could be drawn:

- The more active and fitter people seemed to be better judges of their performance.
- Evidence of a second wind was apparent in most cases at between about six minutes and ten minutes
- The heart rate could not normally be related to walking speed (N.B. I would add that I'm sure this is true in a twelve minute study but if you look at someone with McArdles exercising over say forty minutes and establishing a second wind and settling down to the exercise sequence, the pulse rate may more readily be connected to speed or effort. But it could usefully be used as a guide both to avoiding pain or a contracture but also to nudging oneself into a second wind state.)

Developments on the horizon seemed to be

- Measuring and calibrating the heart rate response in individuals to prevent the onset of contractures and acute pain (and muscle damage) during the warm up period.

- Measure oxygen usage to prove the existence of the second wind but also to help train people how best to exercise and how to bring on a second wind.
Possible use of dietary and pharmaceutical supplements.

N.B. In this third category carnitine was mentioned and many people will know that vitamin B6 has been considered as has glucose and fructose and high protein diets and low carbohydrate diets. There is no doubt that some preparations help some people but there is no scientific proof that any particular supplement helps all McArdles people.

Everybody was most grateful for John Buckley’s efforts in making such a clear and meaningful presentation which showed that he had a good understanding for the considerations which people suffering from McArdles would have in the forefront of their minds when faced with any form of exercise evaluation -albeit NOT exercise tolerance. He and the Oswestry team had a good understanding of the physiology of the condition and they had the confidence of the group.

After lunch by popular demand the group reformed

There was general discussion about management and progression. Andrew Wakelin in particular shared his experiences of very different performances from day to day even after a bit of warm up and when he could identify no dietary or conduct related reason for it.

Karolina Zanoli from Switzerland reported that she keeps the condition under control and is quite active using magnesium tablets but the precise dosage and form was not explained (I have incidentally asked her subsequently to enlarge on this).

Jane Cooper, a nurse, explained how thyroid problems had been at first wrongly attributed to McArdles and Sionned Williams our leading harpist explained not only about the problems of having a non apparent but debilitating disease but life with an orchestra and McArdles.

Andrew highlighted his own experience on the subject of diet. Carbohydrates are essential to him and coming off them and trying the high protein diet not only set him back but did permanent...