

TYPE 5 MCARDLE'S DISEASE REPORT ON WORKSHOP - 9 OCTOBER 1999

There is a pressing need to report on the AGSD (UK) Workshop at our Annual Conference held in Manchester on 9 October.

I was diagnosed at 29 in 1976 having had a difficult childhood and a father who was a doctor who wouldn't accept that there was anything wrong with me. He went to great lengths to prove that to the school doctors and that was painful! I'm a lawyer and since I qualified (in the same year as I had that muscle biopsy - 1976) life has got better. I have learnt that I have a physical condition and since going to see Dr. Slonim in 1992, I've learnt that I can do something about it too.

At the Conference we were very fortunate in having the only UK doctor who heads up a clinic devoted to McArdle's in the UK, Dr. Ros Quinlivan, and Dr. Alf Slonim flew in directly to Manchester. There were two members of the support team that Ros Quinlivan works with at Oswestry, Tony Twist, a dietician and Jenny Whittal, an exercise therapist. That meant there was a body of informed professionals present who, between them, had seen a relatively large number of McArdle's patients. They had experience of the wide variety of presentations the condition produces and they were able to relate in the right context (sympathetically) to many of the individual problems which people reported.

This is only an interim report because I need to obtain approval from Dr. Quinlivan and Dr. Slonim about the recommended management regime. There was insufficient time for Dr. Quinlivan to respond to the presentation made by Dr. Slonim. As everyone who suffers from McArdle's knows there are many similarities with the condition and many sufferers can identify with other peoples' experiences but no two patients are identical. There are major differences over the severity of the condition. It would be very encouraging if we achieved a management regime that amounted to an agreed protocol on both sides of the Atlantic, but to say this is already "agreed" in the UK would be a bit premature. There may be small differences in approach between the UK message and learning on this subject and the US one. A complete meeting of minds will need a further exchange of information and experience. One way of bringing this goal closer would be for Dr. Quinlivan to visit Dr. Slonim at North Shore University Hospital with a patient who could then be evaluated and set up on an exercise and diet regime. AGSD (UK) may be able to assist financially in that endeavour.

On my count (which is probably wrong) there were seventeen actual sufferers at our Conference but they or others who were there represented a further seven Type V's. So some 24 McArdle's people were represented and it really is true that some very broad trends and similarities were apparent. From a McArdle's standpoint this was a powerful assembly.

With such a relatively large group of people it inevitably took time for everyone to explain who they were and how (and how definitely) they had been diagnosed. Dr.

Slonim gave a fairly detailed explanation of the physical and chemical characteristics of the condition. I will write these up in detail but I would like, ideally, to have both doctors agree. I am seeing Dr. Quinlivan on 29 October and that may provide the opportunity for getting her blessing. There is of, course, no difficulty over the basic cause: the absence of a debranching enzyme, muscle glycogen phosphorylase (myophosphorylase), in skeletal muscle preventing the conversion of glycogen to glucose. But the various theories about the second wind phenomenon, the role that protein plays in the diet and the way that proteins and amino acids work in that process (in the muscle and in the muscle cells) are subjects that the professionals need more time on.

ATP, adenosine triphosphate, is the source of energy for many enzyme reactions in muscle fibre. How it works with impaired bio-chemical reactions where muscles are having to look for a secondary or tertiary source of fuel is simply not understood (as far as I know) but it could have a lot to do with the second wind phenomenon. There are certain identified amino acids which might help in the process of gaining a second wind. The role of certain substrates and the role of the mitochondria in their exercised and, by contrast, their untrained state was briefly mentioned. I think this has so much to do with the way and the ability of some McArdle's people compared to others to get a second wind that it needs consultation with the experts before I say anything more about it and this is only a preliminary report.

The group was generally representative (for the first time at UK AGSD meetings) of what Dr. Slonim describes as McArdle's patients. Even so they were very varied. Three were in (or partly dependent upon) wheelchairs. Most told similar stories about being regarded as lazy and having to find excuses to explain non-apparent physical inadequacies. Many had experienced a similar pattern to the condition. The majority of represented people who were under forty years old were doing fairly well. There was a small number however who simply could not manage exercise at the rate or in the way Dr. Slonim was advocating. Dr. Slonim seemed to accept that in some of those cases there was a complicating factor - like another serious illness. In other cases he wanted to gain the individual's confidence to embark upon a new lifestyle to start taking exercise if exercise ability/tolerance was at a very low ebb.

In other words you can get over the hump without hitting a wall!

Dr. Slonim explained how some of his patients had been in a very weak state and they were almost unable to exercise on his laboratory treadmill. Incidentally, I have done this and it's not half as daunting as it sounds. The main reason for this is that Alf Slonim understands McArdle's patients so he knows what they anticipate and what they are going through. He also has a team of supporters who help him pick up the pieces. When I was on my first trial on the treadmill he insisted on having his heart specialist there (I do have high blood pressure) as well as his exercise therapist and a clutch of other friendly and understanding people. I had my own exercise therapist there for moral and physical support, and they all got on very well. I can assure you that Linda stands for no nonsense! Over a four day period the exercise performance held progressively less apprehension for me. It was always controlled and I never felt they were "testing" me. At the first serious

sign of pain Alf would arrange for the treadmill to go slower and he and they understood what we were all trying to do - get a second wind as simply as possible.

Going back to our Conference, I have to say that almost everyone was in good spirits and there was no evidence of depression. This is apparently one of the things which UK McArdle's people have indicated or complained about as one of the side effects of the condition. People may have been elated about spending a night in Manchester or seeing Dr. Slonim - but at any rate to a layman they seemed generally quite happy. Some may have been putting a brave face on it and if so I would like to know (please).

We ran out of time and therefore a detailed exchange on what sort of diet people should eat and how individuals should exercise simply did not happen. The message from Alf Slonim was very strongly: plenty of controlled exercise coupled with a high protein diet. He has rather increased his idea about how much exercise we should be taking. On close questioning he admitted that the crucial factor was the continuous taking of exercise and the commitment to the regime. He was only saying a minimum of an hour a day, every day, because people were not usually compliant. They gave up and they always did less than they signed up to and promised.

The precise structure of a high protein diet needs to be tailor-made to the individual and that's one of the things they evaluate at North Shore if you go there. However, the percentages of what you eat as between protein, carbohydrate and fat in your diet can be expressed. I will do just that but I would like our Dr. Quinlivan to agree (if she can) before putting the figures out here. Suffice it to say that the exercise element in the Slonim approach seems to have become even more important than it was when I saw him in 1992. Eating a high protein diet without embarking on and submitting to the exercise regime will simply mean you will get out of condition and eventually you will turn the excess of protein to fat (depending on how you are generally nourished).

The best source of protein for McArdle's people according to Dr. Slonim is animal protein. Things like fish (including shellfish), snake, eel, pork, mutton, beef, and lots of flying things including and some things that can no longer properly fly like chickens and ostriches. They are all good for McArdle's people.

But if you are well nourished the exercise is more important. That's the difficult part. That's what takes the effort and that's what you don't get any immediate reward for. As far as the diet is concerned Alf's strongest message would be - cut down on the carbohydrate. If you are not well nourished then you need to get at the protein. That problem probably goes beyond the confines of a note about a McArdle's meeting in Manchester. Alf has modified his thinking about exercise in the sense that he now thinks everyone with the problem needs to exercise for an hour a day every day. Most of us couldn't do that because of other commitments (both to ourselves and others) and I don't know of anybody who has kept it up. I exercise for an hour a day walking to and from work but after five years riding a static bike as well I got bored with it and I now only manage that half hour (but exerting) slot about 75 days a year. If I let myself go the damage of not cycling (but still walking) is apparent after about three weeks.

As to the method of exercise and how it impacts on me at least (although Alf thinks this fairly typical) I find it takes between six and fifteen determined minutes to get a second wind. If I'm pedalling away at my static bike on a measured charge of energy it always takes seven and three quarter minutes! If I'm running after a bus or trying to jack up the car forget it! It can't be measured and it's bound to end in problems - almost no matter how conditioned I am. The method of exercise is a gentle introduction, seven to fourteen minutes of something between slight pain and serious discomfort and then a similar time of exercising on the plateau when if I went harder I might achieve a third wind or I might hit another barrier. Then I try to do a minimum of five minutes wind down which is always no effort at all. It's a relief and very relaxing and that's the best moment of conditioning for your muscles.

As for diet and lifestyle - our Conference was again not specific or prescriptive. Alf advocates high protein and almost no alcohol. There were two McArdle's patients present who were not the worst afflicted in their age ranges who drank rather a lot of alcohol and one of them depended on plenty of carbohydrate to "get going". Suffice it to say that most McArdle's people do find that alcohol damages their performance and there are good bio-chemical reasons for that connected to the way the body breaks down alcohol coupled to the predicted energy pathways which produce a second wind.

There is a lot more which needs to be said on the mechanics of the condition, on the progression of the condition, on the method and way of exercise, on possible dietary supplements (i.e. vitamins), on genetic or enzyme engineering, on the long term management of the condition (what's right at 12 may be damaging at 53 and vice versa), on the tolerance of pain and why some people suffer it and some don't when they're actually exerting themselves. I think those topics should wait for future appraisal because they were only touched on at our Conference and there is so much to tell and so much happening. This is intended to be a preliminary report.

More will follow when I have some reasonably meaningful information from the doctors or if I learn of new developments. I realise that there is a lot left out - like the member we have who can walk for twelve or more miles once a week - or so. Dr. Slonim didn't quite have an answer to that. He also couldn't explain why at least one person needed carbohydrates and survived reasonably well on them. But all that apart, he did identify with the majority of us and he is always ready and prepared to talk.

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