

PRE-PUBLICATION PROOF

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# Living with McArdle Disease

Guidance for protecting yourself, starting an exercise programme and improving your techniques.





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**Guidance for protecting yourself,  
starting an exercise programme  
and improving your techniques.**

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International Association for  
Muscle Glycogen Storage Disease

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**Photographs**

*Front cover:* Bottom row, L to R, problems: keeping up in a crowd; stairs; children rush past as a McArdle-ite has a rest. Top row, L to R, solutions: gentle aerobic conditioning, Martha's Vineyard, USA; on-site tutorial, walking course in Wales; Photographs by Andrew Wakelin, Dan Chambers and Shutterstock.

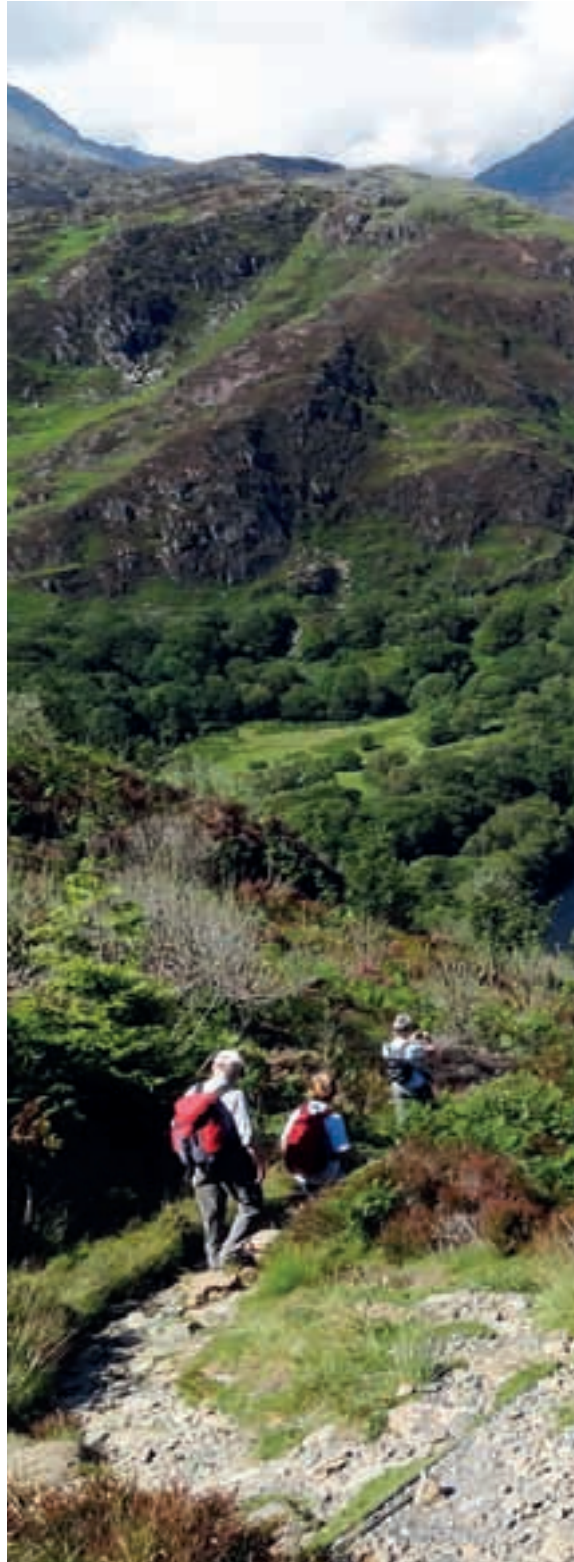
*Frontispiece:* Andrew Wakelin and Stacey Reason, on a "Walking with McArdle's" course. Photo Dan Chambers.

*Opposite:* Top: Descending to Llyn Dinas on a walking course in North Wales. Below: Dan, Andrew, Stacey and Andy on "Walk over Wales" where much of this guidance started to evolve. Photos Dan Chambers.

Our thanks to everyone who pooled photographs over 7 years of walking courses, in particular Dan Chambers, Sally Wakelin, Jos van den Einde, Stacey Reason, Valéry Beaugrand, Andrew Wakelin.

**Acknowledgements**

My thanks to Dr Ros Quinlivan, Dr John Buckley, Dr Richard Godfrey and all the staff past and present of the UK McArdle Disease Service.



Genetically confirmed patients are requested to register with Euromac – the registry for people with McArdle Disease and other very rare glycogenoses.  
[www.euromacregistry.eu](http://www.euromacregistry.eu)

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# Introduction

Walking for the first time with another person with McArdle's sowed a seed for what is now in this book.

I became the AGSD-UK's co-ordinator for McArdle Disease in 2004, but it was in 2006 that I first walked with another person with McArdle's. Jenny Coyne from Australia came to visit me in Wales and we did several short walks together. We were able to compare notes and it was a revelation. It sowed a seed.

Meeting more people through the AGSD-UK and the national McArdle's clinic, I realised that many people seemed not to have understood some of the basics which underpin managing

McArdle's. So I started to analyse how I managed my condition. It was initially hard to do, as I had learnt to adjust and adapt without much conscious thought. I needed to bring it all back to the surface.

I began to publish items of guidance on the AGSD-UK web site and people started to tell me how helpful they found it. Some said it had changed their lives. One man said it had probably saved his life, as he had not realised how life-threatening rhabdomyolysis could be.



*The 'high level' group on day 6 of a walking course. They climbed Tryfan, the mountain behind them.*



*Dr Ros Quinlivan (right) joins Andrew and Stacey on a "Walking with McArdle's" course, Wales, 2011.*

Through sharing with others affected by McArdle's around the world and on the walking courses, by liaising with the medical professionals at the UK Clinic and in Euromac, I have continued to develop the body of guidance for living with this condition. It is now encapsulated in this book.

*Andrew Wakelin*

# Before using this guidance

Read this carefully before you proceed.

Before using the following pages you really need to:

- 1) Make certain that your diagnosis is correct. Ideally this entails having it genetically confirmed by DNA analysis, so if you don't have that already you should request it.
- 2) Seek the opinion and advice of the doctor who looks after you and your muscle GSD.

If you are unable to secure both of the above, then you must make your own decision as to whether it is sensible for you to make use of these guidance notes.

## McArdle Disease (GSD5)

A good start is to read the book "101 Tips for a Good Life with McArdle Disease" (see page 48). Then weigh up each suggestion here and see if it is suitable for you. You

must consider facts such as your level of aerobic fitness and any complicating medical conditions.

## Tarui Disease (GSD7) and other muscle GSDs

If your diagnosis is not McArdle's but one of the other muscle GSDs you need to take extra care in deciding whether to make use of this guidance.

Pompe Disease (GSD2) is not compatible. Tarui Disease (GSD7) is very similar to McArdle's, although it seems the second wind may not apply, or at least take a lot longer to start up. I consider Tarui Disease to be very compatible with this guidance.

The other muscle GSDs have a small number of diagnosed cases and therefore a lot less is known about them. You need to be very cautious in applying this guidance.

# Watch the videos

Instructional tips and reports of the walking courses.

The AGSD-UK McArdle Disease YouTube channel has over 30 videos on various aspects of McArdle's. There are also links to



talks and conference presentations by medical professionals and other organisations. Plus some are for entertainment – try the one of two McArdle-ites on the knife edge mountain ridge of Crib Goch! The aim is to keep improving and expanding it. [www.youtube.com/agsduk](http://www.youtube.com/agsduk)



# McArdle Disease

**McArdle's is a very rare disorder of muscle metabolism.**

**We believe it affects about 1 in 100,000 people, but to date only about 3,000 people have been diagnosed worldwide.**

## Genetic condition

McArdle's is inherited from parents who are both carriers and is not infectious. There is no cure, but much can be achieved with good management.

## Energy shortage

People with McArdle's experience a serious shortage of energy during the first 10 minutes of any activity, and throughout *all* intensive activity. Care has to be taken, as even activities like chewing, drying after a shower and hanging up clothes can cause muscle symptoms.

## Symptoms and risks

McArdle's people appear normal and healthy but activity results in premature fatigue, exaggerated heart rate, pain and muscle spasm. If activity is continued despite the pain, muscles become stiff and swollen and muscle breakdown (rhabdomyolysis) may occur. In severe cases this can lead to life-threatening kidney failure or compartment syndrome requiring urgent surgical intervention.

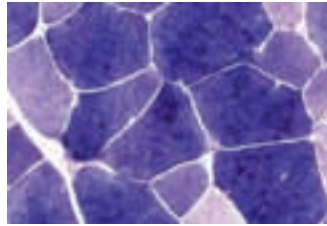
## Longer term risks

Longer term, the risks are of accumulated muscle damage from repeated over-exertion or alternatively of muscle wastage due to the avoidance of activity. If activity is avoided due to the symptoms this can lead to loss of aerobic fitness which makes it much harder to do anything without quite severe symptoms. It can also result in a wasting away of the muscles.

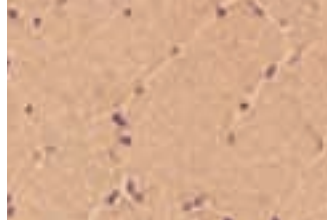
## Adapting activity

To cope with daily tasks, people with McArdle's have to break down any intense activity into very short sections of a few

seconds. Some activities are avoided and others carried out differently. McArdle's people also need to take plenty of gentle exercise to enhance their aerobic capacity. They should plan for at least 45 minutes of exercise, five times per week. The best way is to build activity into your life.



*Normal muscle showing muscle glycogen phosphorylase enzyme.*



*Muscle of a person with McArdle Disease showing absence of the enzyme.*

## The technical bit

McArdle's is also known as Glycogen Storage Disease Type 5 (GSD5). It is inherited in an autosomal recessive fashion. Mutations on the *PYGM* gene on chromosome 11 result in the absence from the muscle cells of an enzyme called myophosphorylase. This enzyme (made up of 842 amino acids) is needed to convert glycogen (fuel stored in the muscle) into energy at the start of activity and throughout intense activity.



# Tarui Disease

Also known as GSD7 or muscle phosphofructokinase deficiency.

## Clinical features

The clinical features of GSD7 are similar to those of GSD5 (McArdle Disease) with onset of more severe fatigue and muscle pain early in exercise. There is said to be no “second wind”. Sometimes there is vomiting after intense exercise.

Symptoms are evident in childhood. Patients may display a hemolytic anemia.

## Energy shortage

GSD7 is caused by a deficiency of the phosphofructokinase enzyme which is needed to facilitate the breakdown of glucose into energy in muscle during exercise.

The body breaks down muscle (rhabdomyolysis) when trying to attain energy, which causes symptoms such as muscle pain, cramping, fatigue and tenderness. The red protein myoglobin is released and red-brown urine may be seen.

## Genetic condition

The enzyme deficiency is due to abnormalities in the muscle phosphofructokinase gene and is inherited as an autosomal recessive genetic disorder.



*Jeremy Michelson, on Snowdon with Tarui.*

## Diagnosis

Diagnosis is by genetic testing or muscle biopsy, which will show a deficiency of muscle phosphofructokinase and a modest accumulation of glycogen.

Treatment primarily consists of avoiding strenuous exercise. Some patients have been helped by a high protein diet, others by a Low Carbohydrate High Fat diet.

It has been reported that only 100 to 200 cases have been identified worldwide.

# Other muscle GSDs

The guide used by IAMGSD’s board is that it wishes to support “McArdle Disease and related disorders of muscle glycogen where exercise intolerance is the major symptom.”

So far it supports McArdle disease (GSD5) and Tarui disease (GSD7). As far as possible it will in future support all other related GSDs with the exception of Pompe Disease (GSD2) for which it defers to the International Pompe Association (IPA).

I believe that GSD3a (Cori Disease), the pure muscle form of GSD9, and GSD13 (beta enolase deficiency) are sufficiently close to McArdle Disease as to be likely that much of the guidance in this booklet will be appropriate. However, people affected by these conditions must discuss this issue with their doctor before making any use of this guidance. The likely relevance to other muscle GSDs is unknown.

# Guidance on McArdle's

Help with protecting yourself, starting an exercise programme and improving your techniques.



*Andrew in the Glydderau, Snowdonia. He has climbed mountains, slowly but surely, for about 60 years.*

**This guidance was prepared by Andrew Wakelin, the McArdle Disease Co-ordinator for AGSD-UK, with the support of many experienced McArdle people. Andrew draws on the management of his own McArdle's and on what he has learnt from professionals in the UK and elsewhere. But most importantly, he draws on over 500 person-days spent walking, talking, supporting, training, working, relaxing and sharing with McArdle people.**



*Andrew (right) aged about 9 with his brother Martin on their first mountain, Moel Hebog 2,569 ft (783 m) in Snowdonia, North Wales. He wasn't diagnosed for another 20 years and had no help for a further 20.*

# Gain without pain

**It is important to remember that the popular exercise-related slogan “No pain, no gain” is completely inappropriate for people with McArdle’s.**

In McArdle’s the predominant muscle pain experienced on exercise is related to the breakdown of muscle. This comes about when the muscle is under heavy demand and finds that it can’t access the glycogen stored in the muscle cells. It looks for alternative fuel sources. It breaks down the muscle itself to obtain protein. The resultant pain is a signal to ease-off or pause for a rest.

Your objective should be to avoid pain during exercise. You should adjust your exertion level up and down to avoid pain, then after about 8-10 minutes you should get into ‘second wind’. Please



*Be wary of a gym, where trainers may push you beyond your limits, risking rhabdomyolysis.*

## Quick summary

- Ignore the slogan “No pain, no gain”.
- When pain comes on, slow down or pause for a rest before it escalates.
- Aim to get into ‘second wind’, when aerobic exercise is easier.

read the section on second wind on page 14.

You need to be very wary of exercising in a gym, due to peer pressure. You may feel embarrassed by the looks from other users if you rest after just a minute or two. Then there is a danger that you will keep going past the point where you should have stopped. There is also a danger that trainers will push you beyond your limits, not realising that McArdle’s pain is different (we do not have a build up of lactic acid) and “No pain, no gain” does not apply.

Of course, just like anyone else, if you do a lengthy exercise session, afterwards you are likely to have some aching muscles. If you exercise regularly you will develop the ability to differentiate between muscles that are aching from doing more than they are used to, and muscles that are experiencing McArdle’s related pain.



*Walking at our own pace is much better exercise for us than risking a workout in a gym.*

# Adjusting activity to protect yourself

Our McArdle's often seems irrational and difficult to understand or predict. In fact, how it affects our activity is explainable, and we can learn to look after ourselves, avoid injury and improve our condition.

Doctors tend to use a simplified approach which has the advantage of being easy for everyone to understand and remember:

**“Don't do any anaerobic activity (lifting heavy weights, sprinting, etc) but do undertake aerobic exercise (walking, etc).”**

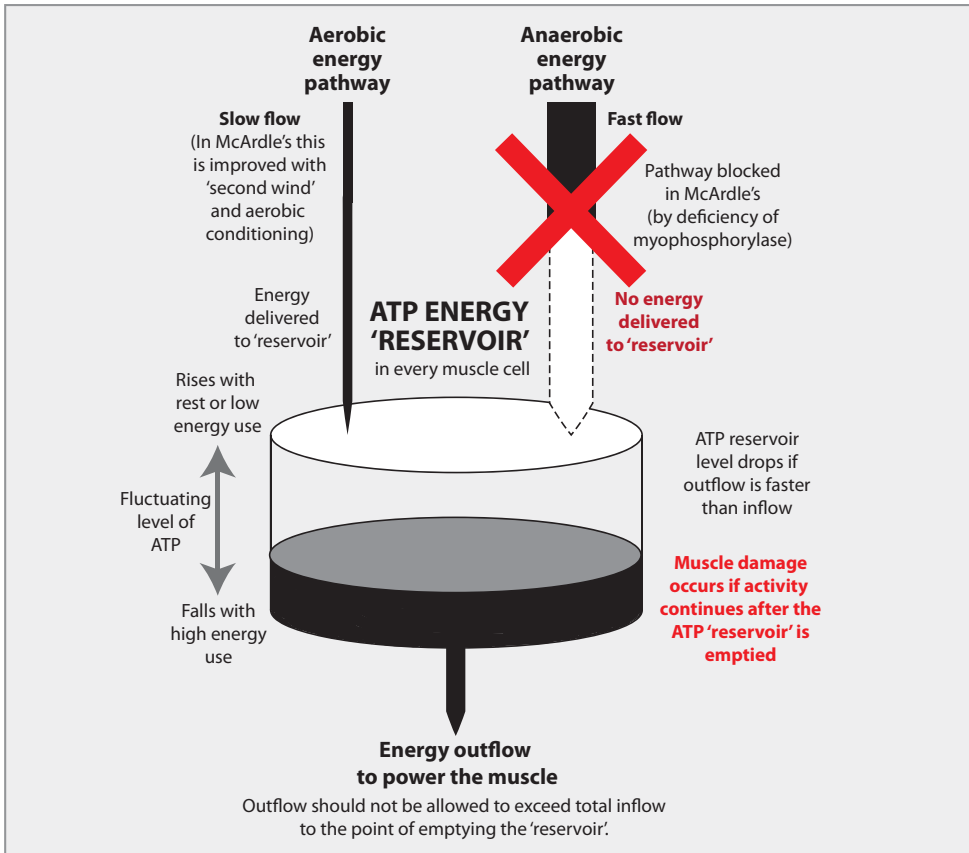
However, to look after ourselves as well as we would like, better advice would be:

**“Don't empty your ATP reservoir.”**

But this needs some explaining before we can understand it and follow it.

## Quick summary

- There is a very small “reservoir” of energy immediately available in the muscle cell.
- An slow (aerobic) pathway tops this up.
- In McArdle's the fast (anaerobic) pathway is blocked.
- We need to limit our demand for energy to avoid emptying the “reservoir”.
- We slow down to balance the ‘in’ and ‘out’.
- If the “reservoir” nears empty we have to pause for 30 seconds to refill it to 80%.



## Energy pathways

The classification of activity into anaerobic and aerobic activity is a simplification. Anaerobic and aerobic actually refer to two energy pathways (ways in which we deliver energy to the muscles). Anaerobic means without oxygen - and this is the pathway which converts muscle glycogen into energy. In McArdle's we can't use this pathway, as we are missing an enzyme (myophosphorylase) which is needed in that process. Aerobic means with oxygen and this pathway converts blood-borne fuels into energy. This is the only energy pathway that people with McArdle's can use.



*An easy stroll on the flat keeps the energy demand low.*

## Energy store in the muscle cell

A chemical called ATP (Adenosine Triphosphate) is used in one of the last steps in activating the muscle. Everyone has a very small store of ATP in each muscle cell. This store of energy is normally topped up by the aerobic and anaerobic pathways. The aerobic pathway is slower at topping up the store than the anaerobic pathway. In McArdle's we can only top up the store through the slower aerobic pathway.

## Reservoir visualisation

You need to visualise the ATP energy store as a "reservoir". The anaerobic and aerobic energy pathways are like two supply pipes bringing water (energy) into the reservoir. The anaerobic pipe is large and people without McArdle's can turn on the tap to a full fast flow very quickly. In McArdle's it is blocked and nothing comes through. The aerobic pipe is narrower and slower flowing but works for us just like for everyone else.

When we exercise it is like taking water (energy) out of the reservoir by an outflow pipe.

In order to not run out of water (energy) we have to ensure that the outflow demand is in balance with the supply. If the outflow is greater than the supply the water (energy) level will drop, and if we keep going like that the water (energy) will run out altogether. This is when we damage our muscles. When they run out of ATP they start to break down the muscle itself as a source of protein to create energy.

## Avoid emptying the ATP reservoir

If we exercise at maximum effort (like sprinting as fast as we can, or lifting something very heavy) we empty our ATP reservoir within about 10 seconds. If we exercise less intensely it takes a bit longer to empty the reservoir. Whilst if we exercise very sedately we can keep our outflow and our inflow in balance and thus never run out of energy.

We can use two tactics to avoid emptying the ATP reservoir. Unfortunately we can't see how full the reservoir is, so we have to learn from experience how to gauge this.

- 1) We can slow down or reduce the intensity of what we are doing so that the demand for energy (the outflow from the reservoir) is slower than the rate at which the reservoir is being topped up by the aerobic inflow.
- 2) If we can't reduce the intensity (like opening a jam jar) then we have to interrupt the demand (the outflow) by stopping for a short rest. 30 seconds rest will allow the inflow to re-fill the reservoir to 80% (see page 18).



*Take care on challenges like this; easy to get a contracture.*

**If you would like to learn more, there is detailed information on the following two pages.**

# More information

## Some practical examples

- 1) When using scissors, secateurs or a pump spray, we can change hands - thus fully resting one hand at a time. The ATP reservoir in muscle cells of the rested hand will fill up again whilst the other hand does the work.
- 2) When carrying something a bit on the heavy side your arms may be using up your ATP quicker than it is replenished. If you can hold the object next to your body, the friction will hold some of the weight and reduce the demand on your muscles. This may be enough to cut your ATP outflow rate to below the inflow rate.
- 3) When walking uphill, which would normally need anaerobic energy, we can slow our pace and/or zig-zag across the slope so as to reduce the effective incline.
- 4) When doing something like gardening or household chores, try to swap back and forth between tasks so that different muscles are used. Maybe do a few minutes of vacuuming (which can be hard on the arms), then swap to say taking something to the other end of the house, which uses your legs and gives your arms a break.

You will soon learn your own techniques.

## How much pain?

People with McArdle's tend to describe the sensation in their muscles as they exercise as "getting heavy", "getting tight", "running out of energy", "being exhausted", or "being painful". Unfortunately these sensations lag behind the damage we are doing. The more we push against it, the more pain we will have.

Over time we can gradually get better at understanding and anticipating the feelings. It should be our aim to never incur pain which, once we stop what we are doing, takes more than a minute or two to subside and disappear. If we get pain that lasts longer than that then we have done too much damage and must learn to ease off or stop and rest before this level is reached.

We should aim never to have fixed contractures (which tend to last for hours or days), and never to have dark urine from myoglobinuria. Of course, it is not always easy to avoid these things, but one fixed contracture per year should be about the limit. If someone is getting more than that, then they are likely to

benefit from a controlled exercise programme. There is some information on starting an exercise programme on page 19. The fitter we get the easier it is to avoid pain.

## Supplements

Some McArdle people report that low-dose creatine supplementation helps them. It may increase the availability of ATP and has been shown to benefit the exercise capacity of healthy individuals undergoing resistance training. However, the Cochrane Review (2014) into pharmacological and nutritional treatments for McArdle Disease reported "Although there was low quality evidence of improvement in some parameters with creatine ... none was sufficiently strong to indicate significant clinical benefit." If you are interested in trying supplements start by reading the latest Cochrane Review.



*Starting level makes it easier to get into second wind.*

## Second wind

At the outset our cardio/respiratory system is at tick-over level so the delivery of oxygenated blood and blood-borne fuels is slow. It takes a few minutes to get this ramped up, so the aerobic energy pathway will be slow to fill the reservoir in the early stages. We are therefore at risk of emptying our ATP stores. We have to be very careful in the first 10 minutes of exercise, until second wind is achieved. (See second wind on page 24.) People unaffected by McArdle's will utilise their anaerobic pathway during this period. When we have achieved what is called "second wind" in McArdle's, our aerobic pathway will be working better than in the early stages of exercise and we are able to exercise much more easily.

# Adverse factors

## Stress and emotion

Stress will tend to make the muscles tense. This interferes with the supply of blood to the muscles and thus activity would normally rely more on the anaerobic pathway – which of course we can't do. We are therefore more prone to muscle damage when under stress, so should perhaps try to postpone significant activity until the stress has passed.



*Tense muscles from excitement like this can be damaging.*

## Temperature

Muscles do not work as well in cold temperatures. This affects us more than others, so keep warm in cold weather and be wary of swimming in cold water (and never in water you can't stand up in).

## Each muscle is independent

Remember that the ATP store is in the muscle cells of every muscle. Any techniques you use are not wholly systemic, but will affect only the



*Soft ground creates a big increase in energy demand...*

particular muscles being used. So slowing down, resting or achieving second wind all need to be considered for each muscle group.

## More that you can do to help

The better conditioned we are the easier all activities will be and less likely we are to incur injury. Regular aerobic exercise is the only available solution known today and probably for the foreseeable future.

## Mitochondria

ATP is made within the muscle cell, in the mitochondria. There are typically about 750 to 1000 mitochondria in each muscle cell. The more mitochondria we have the easier it is to get through our everyday activities and keep up our exercise. The number of mitochondria is increased through muscle conditioning (also called training). At the two extremes, a "couch potato" would increase his or her mitochondria 20-fold by training up to the level of an athlete.

A modest amount of aerobic training to increase the number of mitochondria by say 5-fold would make a significant difference to a McArdle person.

## Cardio/respiratory

The more exercise you do the better your cardio/respiratory system will become. The better you will be at delivering oxygenated blood and blood-borne energy to the muscle cells, thus reducing the risk of injury.



*...as do uneven surfaces.*

# Second wind – an essential tool

**Normally, in the first few minutes of exercise the body wants to start using glycogen stored in the muscle. With McArdle’s we can’t do that, so we struggle until other metabolic pathways start up. That is “second wind”.**

Other people convert the glycogen stored in their muscles into glucose in order to use it as fuel. We have to use glucose and fat which is delivered to the muscle in the blood supply. Our liver starts to release glucose from its store of glycogen, and fat is released from our fat stores.

Switching to these fuel sources enables us to enter a “second wind”, a period of more effective and less difficult exercise, after the initial period during which cramping and pain are likely, but need to be carefully avoided.



*An ideal start, gently downhill on a good track.*

## The benefits of second wind

The main benefit is that you can undertake more activities and exercise with less or no pain. However, a very important secondary benefit is that it enables you to undertake sustained exercise which will ‘train’ the muscles and improve their aerobic capacity. This will make all activity and exercise easier in the long term.

Ideally we should get into second wind and then continue exercising for another 45 minutes, and do this most days.

Exercise is proven to make you feel better and helps to keep your weight down, both of which are very helpful in McArdle’s.

## Quick summary

- Achieving second wind makes exercise easier and improves your aerobic capacity.
- Second wind is universal to all with McArdle’s.
- It is achieved in about 8 minutes after the start of exercise.
- Avoid pain during warm up by slowing and/or pausing and restarting.
- Second wind is not fully systemic, it has to be achieved in each muscle used.
- A break of more than about 30 minutes may lose your second wind.

## Universal to all McArdle people

A few people with McArdle’s feel that they do not get second wind. It can be difficult to recognise in your everyday life as there are many external factors affecting the situation. However, studies have shown that when a person exercises in a controlled manner, wearing a heart rate monitor, it is possible to demonstrate the onset of the second wind. Hundreds of people, in different centres, have exercised in this way and the second wind phenomenon has been found to be universal to McArdle patients.

So if you don’t feel you experience second wind, it is well worth asking for some help to discover it. Once you have learnt to achieve and recognise it, you can make use of it.

## Getting into second wind

It takes approx. 8 to 10 minutes of exercise to get into second wind. This should be the same for everyone, but what you can do in that time will vary depending on your current condition. The objective is to keep exercising but to avoid any significant pain. If you start to experience pain, slow down, if the pain doesn’t abate, then slow down more or even pause for a rest. As long as you pause soon enough the pain should fade off within about 30 seconds and you can then start again. Keep juggling your efforts and the pain in



this way whilst you continue to exercise and after about 8 minutes you should start to find that you can comfortably do more than you could at the start.

There is a talk about getting into second wind in a video on the AGSD-UK Youtube channel.

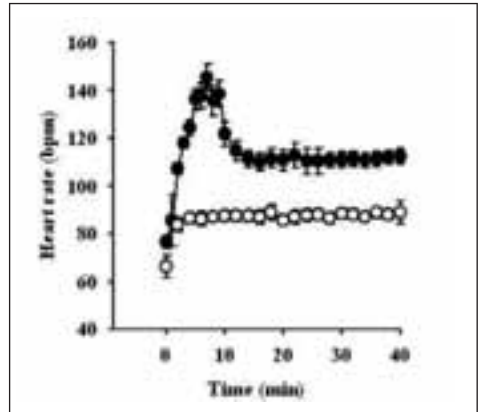
### Avoid pain

Be wary of pushing on when you feel pain start. This pain is as a result of damaging the muscles, and repeated damage will cause problems in the long term. But also this is counterproductive - it will stop you from getting into second wind. By pressing on despite the pain, you start your protein metabolism which then effectively blocks your glucose and fat metabolism.

If ever you get into this situation, you need to stop completely for 30 minutes or more and then start the whole process again.

### Recognising second wind

Unfortunately, achieving second wind is not like flicking a switch; the change comes about gradually. The main signal is that you can carry on free of pain at a rate which would have given you pain at the start. Your heart rate (pulse) is also a very useful signal. In McArdle's, our heart rate tends to increase in what is called an 'inappropriate' response. That is, after the start of exercise it increases much more quickly than would be expected in someone unaffected by McArdle's. Once in second wind the heart rate slows again for the same amount of effort.



Graph copyright Neuromuscular Clinic and Research Unit, Department of Neurology, University of Copenhagen, Rigshospitalet, Copenhagen

In this graph from Prof. John Vissing, the heart rate of McArdle people is shown as black dots and the heart rate of normal (control) people is shown as white dots. People were exercised on a stationary bike at a steady work rate of 65% of their theoretical maximum. You can see how the heart rate of McArdle's people increases dramatically more than the controls. At about 7 minutes it starts to reduce again and by 10 minutes has stabilised, although still higher than the controls. Ratings of perceived pain (not shown) echoed the same curve as the heart rate. In real life you would not press on with your heart rate peaking so much, but this is a very clear illustration of the second wind effect..



Comparing notes with others with McArdle's helps us to understand how to avoid problems and recognise second wind.



*A slope after the flat - slow down or you'll have to pause.*

## Each muscle has to be 'switched on'

The process of getting into second wind isn't systemic to your whole body, but most aspects of it apply to each muscle you use. So, for example, if you walk until you are in second wind your legs will be able to do more without problems than they did at the start, but your arms will not be in that improved state.

If you are walking in second wind and the gradient changes significantly, you will start to use



*We all take a pause. Always respond to the muscle signs.* some different muscles – you need to achieve second wind in these as well.

If you walk for several hours over different gradients you may very well end up with all your walking muscles in second wind. You are likely to feel great and begin to wonder if your McArdle's has been cured. But unfortunately this state is only temporary!

## How long second wind lasts

Once in second wind it will keep working whilst you continue exercising those muscles. If you stop it doesn't disappear straight away, but will fade off. It will be largely gone in about 30 minutes, and is unlikely to be functioning by 45 minutes.

It is best to limit yourself to short stops. For example, if on a day's walk you take a lunch break of more than about 20 minutes you are at risk of having to work at getting back into second wind.

Because the second wind applies to each muscle, if you do a downhill section for more than half an hour, your uphill muscles may lose the second wind. If you do another uphill section second wind will have to be achieved again.



*Limit breaks to 20 minutes or risk losing second wind.*

## To conclude

If you don't already experience "second wind", hopefully you can learn to get into it. If necessary ask for expert guidance to show you how to achieve it. Use it to make your life easier, improve your fitness and feel better.

### Case report

- A woman of 60 was restricted in exercise as she had never experienced second wind.
- She didn't have the confidence to walk on the clinic's treadmill.
- She walked with a physiotherapist on a "shuttle" up and down the corridor.
- Through use of a heart rate monitor and a pain scale she recognised second wind for the first time in her life.
- She can now exercise regularly.

# Six second rule

The “Six Second Rule” is as useful for people with McArdle’s as the “second wind”, although in a very different way. By using it you can help to avoid fixed contractures of the muscle, with their attendant muscle break down (rhabdomyolysis) and protein in the urine (myoglobinuria).

## Immediate energy for 5 to 10 seconds

When doing something like lifting heavy objects or sprinting, there is an energy pathway (the phosphagen system) which is instantly available and lasts for about 5 to 10 seconds†.

This system relies on ATP (adenosine triphosphate) stored in the muscle and creatine phosphate to provide immediate energy. As that is exhausted, people who are not affected by McArdle’s will start to make use of glycogen stored in the muscle, converting it to glucose for energy. However, in McArdle’s an enzyme (myophosphorylase) needed in that process is missing so this energy pathway can’t kick in after the initial 5 to 10 seconds. That is when a painful fixed contracture of the muscle will develop which can last for hours or days.

## Protect yourself in everyday activities

Everyday examples of maximum intensity activities are: opening a new jam jar which is



An example of a maximum intensity activity.

firmly stuck, lifting something heavy; standing on tip toe to get something off a high shelf; rushing up a flight of stairs; and squatting. These are everyday activities which are best avoided, but if you have to do them you need to know how to protect yourself.

## Count to six

To avoid damage when doing something of maximum intensity it is a good idea to time six seconds by saying to yourself “one thousand, two thousand...” up to six. If the task is not completed by six, stop or put it down. Take a break, let the muscles recover and try again later.

## Quick summary

- Avoid maximum intensity activity.
- If you have to do some, the Six Second Rule will help you avoid muscle injury.
- Count seconds whilst doing something of maximum intensity.
- If not complete by six, stop, take a break and try again later.

## Recovery time

Research shows that the ATP will recover in about three minutes, and in fact substantially recovers in about 30 seconds. We don’t really know what factors affect its recovery, whether it varies from one person to another, or whether we can do much to improve its speed of recovery. These are all questions for future research. However, we do know that if our muscles are fit (aerobically conditioned) we will be safer.



An awkward step. Can you complete it in 6 seconds?

† Saltin, B. (1973) *Metabolic fundamentals in exercise. Med & Sci in Sports*, v5, n3, 137-146.

# “30 for 80”

Once you need to pause for a rest, count to 30 seconds.



It is unfortunate that the pain from a McArdle’s cramp lags behind the damage that we do, probably by at least 4 or 5 seconds. So we need to learn to recognise the very early signs and slow down or pause

for a rest before we do any damage.

Our saying comes from the fact that 30 seconds rest will replenish 80% of the immediate ATP energy “reservoir” in our muscle cells. The replenishment slows down as it progresses, so it is not worth waiting to reach 100%, as that takes about three minutes.

With 80% we have some leeway, so that when we set off once more we are not going to run out again straightaway, even if we are going a bit too fast and using more energy than our slow aerobic pathway can deliver.

Most McArdle people tend to move off again too soon, after maybe only 5 or 10 seconds. They

think “Oh, I’ll be OK”, or “I feel a bit better now”, or “Those people must be wondering why I am stood here”. But move off with only say 20% of your immediate ATP replenished and you are at risk of a stop/start scenario that ends up making you think it is a “bad day” and giving up.



*Patience – count to 30 seconds.*

So remember “30 for 80”.

Patently count to 30 seconds to replenish 80% of your ATP before you move off again.

# “Rest before risk”

Before tackling anything with a risk, briefly rest up.

**VITAL FOR SAFETY**

Some activities have inherent risk of accident (such as crossing a road or jumping in a swimming pool), others have a risk of us getting a cramp or contracture (such as climbing stairs or crossing a stile). So when we are about to do something like this which might involve risk, we

should rest for about 30 seconds before proceeding. This is to ensure that we fill the ATP energy “reservoir” (see page 10) and have enough immediate energy available to hopefully get us through without running out or incurring a cramp or contracture.



*Crossing a busy road, climbing stairs, crossing a stile or...*



*climbing over rocks, always rest beforehand for 30 seconds.*

# Starting an exercise programme

**These notes are intended to be of assistance to people with McArdle Disease who are at a low level of aerobic conditioning and need to get started on a regular exercise regime.**

## Basis of suggestion

These are my suggestions drawn from my own experience, the experience of others with McArdle's and from listening to consultants and exercise specialists who work with McArdle's. I am not a doctor, but simply an experienced patient. I have had constructive suggestions from people with McArdle's who have recovered from extremely low levels of aerobic fitness.

## Before you start

Ideally you would be seen by a consultant with expertise in McArdle's and would be given an exercise regime tailored to your particular state of fitness and any other personal factors. (In the UK everyone should be able to get a referral to the UK McArdle Clinic.) If you cannot get to see a suitable consultant, you may decide to use my guidelines at your own risk. Please read the opening statement to this guidance on page 5.



*Personalised guidance from the professionals is best.*

## Quick summary

- The aim is to enhance aerobic fitness whilst minimising anaerobic activity.
- Over weeks or months, aim to build up to sessions of 45 minutes, 5 days per week.
- Make time for exercise and keep a log.
- Use a treadmill or find a flat area outdoors.
- Monitor heart rate,  $65\% \times (220 \text{ minus age})$ .

## Planning

### Objective

Your objective is to build up your aerobic capacity whilst avoiding any anaerobic activity. When you exercise aerobically your body gets better at transporting oxygenated blood and blood-borne fuels to your muscles. More mitochondria grow within the muscle cells and these make it easier for you to exercise. This is the same effect that athletes use when they train, but it is particularly beneficial to those of us with McArdle's.

### Your target

You need to build up gradually to the level of getting into second wind and exercising for 45 minutes, about 5 times per week. Depending on your current conditioning, this may take several months.

### Setting aside time

You need to set aside time to do this. It may be difficult due to work and family commitments, etc., but it has to be prioritised. I suggest you try to find a slot in the day when you can manage it and always do it in that slot – get into a routine. (I am lucky in that I have a treadmill at home and can use it whilst listening to a radio programme I like, so it gives me a set time. But also try listening to an MP3 player or watching TV to help pass the time.) Once you are up to the 45 minutes and it is going well, you may be able to relax the fixed slot routine and instead fit in at least some of the walking with other things that you do. Perhaps visiting a friend or relative, going to the shops, etc.

## Logging your efforts

Keep a log, or use an activity tracker app on your phone, and it will help you to see your progress and encourage you to keep going. We all need some motivation. The exercise will become easier as you go on. You can download a log sheet from the AGSD-UK web site, or see page 23.

## Where to walk

If you have a treadmill available, that is ideal as you can control your speed and incline (but initially *only* on the level). If you use a treadmill in a gym, you must make sure that you take control of your own exercise – never allow an enthusiastic assistant or trainer to set your speed or goals. They simply cannot understand about McArdle's. As experts they may insist that they know best, but do *not* give in. Don't compare yourself to



*For a low level of fitness, walking safely indoors is ideal.*

other people. Ignore their odd looks if you stop for a rest after just two minutes!

If you don't have a treadmill, find somewhere convenient that you can walk on the flat and on a hard surface like a pavement (sidewalk) or surfaced path. You can turn around and walk back and forth, so it could be as little as 100 yards (although you might then need to explain to the neighbours that you haven't gone completely mad!). If you walk outside you may want to walk with a friend at times. If so, you *must* insist that you have to walk at *your* pace and obey *your* need for rests. You must *never* feel obliged to accommodate them or their needs – walk to suit *you*. (Read the section on guidance for walking partners of McArdle people, see page 32.)

## Monitor your heart rate

A heart rate monitor is needed at the outset. Maybe you could borrow one for a few weeks

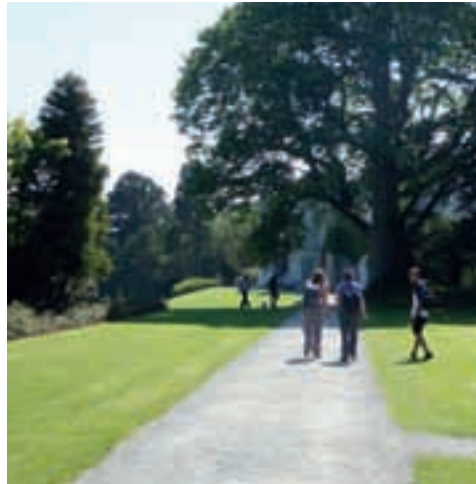
from a friend or a gym. You only need a basic model and can purchase a strapless version (wristwatch style with no chest strap) for about



£40 sterling. You should check your heart rate frequently during exercise and not exceed a set beats per minute. You calculate this as the maximum heart rate of 220 less your age and then take 65% of that. (For example mine is  $(220-68) \times 0.65 = 99$ .)

However, this can only be a guide and you need to see how you feel.

The idea of this is to help make sure that you do not over stress your muscles – your heart rate will increase with muscle stress. You will eventually get used to the sensations and will know by the feel in your muscles when to slow down or stop for a short rest. When you are comfortable with that you will no longer need the heart rate monitor.



*When outdoors, find level, hard surfaces to start with.*

## Case report

- A man had been advised to wear body and leg braces.
- He became very incapacitated.
- Using this guidance he found he could attain “second wind”.
- As his muscle strength improved he soon discarded the braces.
- He further improved to lead a normal life.

## Before you start

### Nutrition

Before you start you need to be on a healthy diet. Which diet is still controversial. At present, major anecdotal evidence from hundreds of people is that a ketogenic diet (about 65% fat, 25% protein, 10% carbohydrates) is far more effective than anything tried before. Clinical trials are awaited.

The latest published study (May 2008) recommends a diet slightly higher than normal in carbohydrates. That diet consists of 20% fat, 15% protein and 65% complex carbohydrates.

In the past, a high protein diet was suggested. A few consultants still promote that, and a small number of people find that suits them best.

This disparity may just highlight that diet is probably less important to us than exercise. See page 42 for more on diet.



*We need to consider nutrition and hydration.*

### Hydration

Take care to drink sufficient water to remain fully hydrated, especially during exercise. Dehydration can be a factor in muscle breakdown.

### Don't exercise when unwell

If you are fighting a cold or 'flu, take a few days off from your exercise routine. It is believed that there is a greater risk of muscle breakdown in these circumstances.

### Avoid anaerobic activity

People with McArdle's must avoid anaerobic activity of more than about 6 seconds (see page 17) – this is high energy-demand activity such as lifting heavy weights, going steeply up hill, sprinting, squatting, etc. Whilst building up your aerobic capacity, if you do any anaerobic activity and get a muscle cramp (fixed contracture) it will put back your training and be a disincentive. So take extra care as you build up your fitness.

## Getting going

### Practice getting into second wind

First of all you need to practice getting into second wind, because exercise without second wind is likely to be damaging rather than beneficial. I must emphasise the need to get into second wind without experiencing any significant pain. You must *not* push through the pain, but keep juggling your effort to avoid the pain. If you feel tightness or pain pause for a rest, it should not take more than 30 seconds to a minute for you to feel OK to continue. If it takes longer than that you were probably going too fast.

Please now read our section about second wind on page 14 for more details before continuing with this section.

If you are at a very low fitness threshold you must start very slowly, about 0.5 mph (0.8 kph). I believe it is more important to learn to respond to the sensations in your muscles than to slavishly follow a formula. However, if you remain pain free you can step up the speed every 2 minutes in increments of about 0.25 mph (0.4 kph). But always taking care to remain pain free, if necessary slow down again or even pause for a short rest.

Once you have improved your fitness, if you find a slow start to be frustrating you might try starting a bit quicker, but do not be tempted to start too quickly. If you press on too fast you may start up other energy pathways which can make it impossible to get into second wind. (It is a bit like taking the wrong turning at a fork in the road – you need to go all the way back to the fork and try again on the right track.) You have to stop completely for about 45 minutes, and try starting again, but more slowly this time.

In McArdle's, no matter how fit we get, the first minutes until getting into second wind are always an issue. You have to accept that.

### Don't overdo it

If you feel great after getting into second wind and exercising, don't be tempted to overdo it. You might then get cramps and that will put you off. Much better to keep to a steady build up of time duration.

If you have aching muscles after exercise it would be sensible to have your plasma Creatine Kinase (CK or CPK) checked by your doctor. A guideline is to ensure that your CK never rises to more than double your normal background level (which is likely to be much higher than the

reference range anyway). If necessary, reduce your exercise for a while. If ever you have a fixed contracture in a muscle after walking, or feel unwell, you should seek the advice of your doctor. In the very unlikely event that you have myoglobinuria (dark coloured urine from muscle breakdown) you should seek emergency medical attention. See McArdle Emergencies on page 43.

## Good days and bad days

A lot of people with McArdle's report having "good days and bad days". They mean that on some days they find it very difficult to get going. The mechanisms behind this are not yet fully understood, although in some cases it may just be that they have been very inactive for a few hours and then have started off walking too fast and can't get into second wind (see above). Whatever the reasons, don't get frustrated and don't be put off by these days. Just leave it till the next day and the chances are that everything will be fine again.



*Getting adventurous... a McArdle group summits Snowdon.*

## Building up to the target duration

Depending on your current fitness you will need to build up to your target time. Start at maybe 10 minutes after second wind – so around 20 minutes total. If you cannot manage that, just get to second wind at about 10 minutes. Certainly don't do more than 20 minutes on the first day. The distance does not matter, just try to get to second wind plus a bit longer. Do this duration for a few days, then extend by 5 minutes and do a few days at that new duration. If you have any problems, reduce again by 5 minutes and keep at that duration for a few days again, then add 5 minutes back on. Keep on in this way until you build up to the 45 minutes. This may take several months depending on how de-conditioned you were at the outset.

# Getting more advanced

## Inclines

Don't think about adding any inclines (hills) into your routine until you can happily manage 45 minutes on the flat. Then gradually increase the steepness of inclines over a period of weeks. Always start a session on the flat and only introduce an incline after getting into second wind. You need to condition the slightly different muscles which are used to go uphill, and this will take time, just as it did for your walking on the level. This is easy to manage on a treadmill, much less so out in the real world.

## Going for a real walk

When you are confident about your abilities you may want to start going for a real walk in the countryside, where your route dictates what you will face in terms of terrain and inclines. We have some notes to help you with this on page 28.

If you have been exercising on a treadmill rather than outside, remember that the surface may be more resistant (loose surfaces, mud, long grass, etc) and you cannot control the incline. Start by being extra careful about getting into second wind and make your first walks outside of shorter duration than you have been used to on the treadmill.

## Keeping in shape

Once you are able to exercise to the full 45 minutes and maybe tackle some inclines, that is not the end of the road. It seems that those of us with McArdle's tend to lose our fitness more quickly than others, and the loss has a greater impact on us. If we lose a lot of aerobic conditioning we can have a hard time and experience repeated muscle injury, flu-like symptoms and even visits to hospital. It is also difficult to get back to fitness. So it is important to keep up with your activity on a regular basis, building it into your life as much as you can.

## Case reports

- A woman increased aerobic fitness to be able to run a marathon in 7 hours.
- A man learned techniques and improved fitness to climb a 14,180 ft mountain.
- A group walked 210 miles across Wales.



Photocopy this page for a motivational log of your treadmill progress.

Date	Workout ref.	Duration minutes	Distance miles	12 minute distance	Max HR	Calories used	Comments

# Stairs and McArdle's

Almost all McArdle people will have an issue with climbing stairs, but there are ways to cope and to improve.



*Take one flight and then take the lift the rest of the way...*



*but stairs are not just indoors of course.*

If I climb stairs at the same pace as other people I can do one domestic-sized flight without problems, but on the second flight I will start to cramp and on the third flight I will become almost unable to move with my heart pounding and my leg muscles very painful. But, conversely, if I go at a slow pace and rest when needed, I can climb three flights of stairs with no symptoms.

## Background to the problem

- It is essential to continue to climb stairs, because if we don't we lose that ability altogether.
- Such a loss has a big impact on quality of life - it is very difficult to take public transport in case there is a flight of stairs around the corner, worried about visiting friends or a restaurant in case they don't have a ground floor toilet.
- To maintain functionality, I recommend tackling at least one flight of stairs each day. If living in a flat or working in an office block, climb one flight of stairs before getting the lift/elevator the rest of the way.

## Quick summary

- All McArdle people find stairs to be a challenge.
- Loss of this ability is a significant quality of life issue.
- Avoid loss by doing one flight each day.
- Go as slowly as necessary, with as many pauses as you need, to avoid symptoms.
- Practice in private to find the pace that works for you.
- Avoid the rush and have a rest at the foot of the stairs before starting up.
- Gradually build up aerobic fitness.
- Always take one flight and then get the elevator/lift the rest of the way.

## Factors involved

- Extra weight has a significantly adverse impact on climbing stairs. Try to achieve the target weight for your height.
- Can you reduce the weight of your bag,

backpack or briefcase?

- Activity (or even getting into 'second wind') before tackling the stairs will help to make them easier.
- Try to plan ahead. Avoid tackling stairs soon after getting out of your chair or a car after sitting for more than about 20 minutes.

## The technique

- Assuming that we have no other mobility issues, just McArdle's, we are likely to be able to rise up one step.
- If we rest until the ATP energy 'reservoir' (the immediate ATP and phosphocreatine system) is restored in the muscle cells, we can then take another step up.
- In this way we can climb a number of stairs, even if very, very slowly.
- The maximum necessary rest per stair is likely to be 30 seconds ("30 for 80" rule – page 16).



*Wait for the main rush to pass before starting off.*

- To start, practice in private at home and perhaps do a few stairs and then reverse down.
- Over days or weeks, gradually increase the number of stairs until you can complete a full flight.
- Most of us will be able to do stairs with a rest of significantly less than 30 seconds on each stair. Experiment to find the length of rest you actually need.
- As fitness improves, you will be able to do them by just going very slowly, perhaps as little as one third of the speed of other people.

- When in public, to accommodate the slow speed, we need to be prepared to:
  - a) wait for the rush of others to go first; and
  - b) allow others to overtake us without embarrassment.
- Even with this slow speed we may start to get some cramping. If so, we need to recognise that and pause for a rest of at least 30 seconds.
- Landings on corners or between floors can be good places to rest, but don't push on against pain to reach one of those. By all means make use of them for a rest BEFORE you really need it, but always stop whenever you feel the need, wherever you may be and however inconvenient it is to others.
- When the cramping sensation has passed, move off again.

## Results

- In theory, in terms of McArdle's this technique allows us to do ANY number of stairs. Of course, normal tiredness can affect us just like anyone else.
- Using this technique on the "Walking with McArdle's" course, participants climbed a flight of over 400 steps in a seaside cliff face with no significant issues; and actually reported enjoying it.
- On the "Children & Parents" event, children of ages 6 to 17 climbed 105 stairs to the spectacular view on top of a castle tower, with just the 6 year old deciding at half way that she had had enough.

## The long term effects

- By climbing some stairs every day we improve and maintain the health and aerobic fitness of the muscles involved.
- Retaining the ability to tackle stairs is an important quality of life issue.
- Aerobic fitness of these muscles will also help when we are faced with an incline in the street.

## Case report

- A young girl with McArdle's (in a wheelchair for two years) used these techniques to include stairs in her exercise plans.
- She can now do two flights of stairs and then takes the lift to her apartment.
- This ability with stairs helped enable her to return to school after 2 years absence.

# Choosing a treadmill

These are guidance notes on trying out, choosing and siting a treadmill for your regular exercise. It is important to get it right, so that nothing discourages you from keeping up your routine.

## Type of treadmill

Perhaps the most important factor is to buy the best, most solidly built treadmill that you can reasonably afford. A vital factor is stability. If you don't feel safe and comfortable on it then you won't use it long term. It is almost certainly best to avoid the folding ones for that reason, and although they are space saving, it is a chore to



Take time to assess several models.

keep folding and unfolding them. A fixed one does of course require a space to leave it out. Bear in mind that a lower quality treadmill is more likely to need repairs at some point, and its absence will interrupt your exercise regime. See also the section below on entertainment regarding placing of a laptop or tablet.

If you are inexperienced in managing your McArdle's you may find a heart rate monitor particularly helpful. Some treadmills have a wireless link to a chest-strap sensor (a bit of a hassle to put on), others sense the heart rate through the handles (you lose the reading when you let go). Or you could use a separate HR monitor that doesn't link to the treadmill. It comes down to personal choice.

## Workout programmes

You don't need amazing software with dozens of workout routines – it is only the fitness fanatics

## Quick summary

- You will need to use this for years, so buy as best as you can afford.
- Avoid the foldaway models, unless space is at an absolute premium.
- Having your heart rate displayed in front of you is very useful.
- Try out a few models at gyms or shops.
- The basic workout programmes are probably all that you need.
- Site the treadmill so as to have no barrier to ease of regular use.
- Spend time learning how to use it to the best effect.
- You will use it more if you have entertainment in front of you.

that need those. You really just want to be able to adjust the speed and the incline very simply whilst walking, as you feel you need to. What we want is to start on the flat at a slow speed, and then increase the speed (and maybe the incline) after about 10 minutes, once in second wind. So buttons for one-touch increase and decrease in speed and incline are essential (see up/down arrows on left and right of the example panel). Increments of 0.1 mph are best for us especially if we have low aerobic fitness, we definitely need increments finer than 0.5 mph. It is worth having some simple workout routines (often called things like "hill" or "random") to give you more variety and to use those once you have improved your aerobic fitness. You want it to allow you to start on the level, and at about 10 minutes switch to one of those workout routines whilst walking, rather than having to stop and restart.

In summary: sturdy, stable, simple programmes, accessible up/down buttons for speed and incline.

## Try out a few to compare

You might visit a couple of gyms to try their treadmills. You can usually have an introductory session at the gym, and use it to try out their model of treadmill. Theirs will tend to be top-end

models of course, and you may not see those models in the shops, but it will give you something to compare the “home” models against. You will be better equipped to make a decision when you try them in a shop. You must try them out - don't buy from the web, unless it is a model you have tried in a gym or shop.



*The control panel can give you lots of feedback.*

### Siting the treadmill

You need a permanent place to keep the treadmill. (Getting it out and putting it away will soon be a chore and your exercise will get neglected.) A spare bedroom would probably be good. Or, if there is space at the side of a living room, that would be even better. Exercise is so important to us, you must be prepared to give it some priority in your home. If you really do not have room, then change to a static exercise bike rather than give up on the idea altogether.

Don't be tempted to put the treadmill in the garage as the cold in winter will make it unappealing, and that is the time that you need it the most as the weather is less amenable to walking outdoors.

### Configuring your treadmill

Take a little time to go through the manual and see what you need to configure. There will be things like choosing between miles per hour and kilometres per hour; also maybe your starting speed and incline (we want it level).

We need to keep our weight under control, as increased weight makes our muscles work harder. So it is important to set up your profile for your gender, age and weight. This will then enable you to obtain accurate figures for calories expended in each session.

If you are using a treadmill in a gym, you may be able to set up a user profile which you can then

log into on each visit. If not, in order to get an accurate report on calories expended you will need to enter gender, age and weight each time.

### Entertainment (or work!)

One factor that people find a problem is boredom, so think about entertainment. Try to set a fixed time slot when there is a programme that you like on the radio, to keep you entertained for 45 minutes. You may be able to have your treadmill facing a TV for the same reason (some very top end models even have a TV built in). Some models offer a holder that allows you place a laptop or tablet/iPad in front of you, or you may be able to purchase an accessory that enables that. Then you can watch what you want and when - to fit in with your treadmill time. It is still a good idea to have a fixed time slot, as that gets you into a regular routine.



*Some people even work at their treadmill, like Dr. Sherry Pagoto, Associate Professor of Medicine at the University of Massachusetts Medical School. [www.fudiet.com/about](http://www.fudiet.com/about)*

### Case report

- A man was very badly affected, with severely low aerobic fitness.
- He installed a treadmill at home.
- He gradually built up to walking on it for 2 miles in the morning and 2 miles again in the evening.
- Years after diagnosis he finally had the confidence to walk in the outdoors.

# Walking tips and hints



*Exercising in fresh air, amongst beautiful surroundings, can give a psychological boost as well as reduce our symptoms.*

**Much is still not known about McArdle Disease, but one thing is clear. Through regular exercise we can boost the aerobic capacity of our muscles and can then achieve incredible things. The following techniques will help you to overcome the barriers put in your way by McArdle's.**

There are videos on some of these subjects on the AGSD-UK Youtube channel. See [Video] marked in the text.

## Slow and steady

This has to be our motto! Slow and steady to get into second wind, slow and steady up the hills. Much better to keep at a pace which we can sustain than push ahead, feel the muscles starting to cramp, the heart starting to race and have to pause for a short break and start again. On up-hill sections we progress at about one third of the rate of 'normal' walkers. On the flat and down-hill we are almost the same, as long as we are in second wind.

We have to know our limits and ensure we stay within them. Starting too quickly and being over exuberant can both lead to problems. 'Slow and steady' is the motto.

## Get into second wind – start on the flat

Whereas most walkers might normally drive to the foot of the mountain and set off straight up,

### Quick summary

- Slow and steady, especially up hill.
- Start on the flat to get into second wind.
- Don't push hard before second wind.
- Use walking poles to help up hill.
- Pause for a rest as soon as your muscles tell you, don't press on to some target.
- Use up hill techniques - zig zag, baby steps, intermediate steps up, etc.
- Limit breaks to 20 minutes to avoid losing your second wind.
- Hydrate and fuel sensibly.
- Avoid exercise with tensed or cramped muscles.

we need to find a car park about a mile away. We need to walk on the flat for about 15 minutes, preferably longer, to make sure we are well into second wind and our cardiovascular system is working well before we encounter an incline. This is not always possible but it should be the aim. If we can't plan such a route then we must take extraordinary care to walk slowly, with frequent

rests, until we get into second wind. With care, this can be achieved on lesser inclines but not on steep inclines. So *never* set off straight up a steep hill. If there is no choice then you must get into second wind elsewhere and then drive to the bottom of the climb, but not for more than about 20 minutes or you are at risk of losing the second wind before you start climbing. [Video]

## Build awareness

The more that someone with McArdle's walks the more familiar they will be with the sensations in their muscles and with their heart rate response. Thus, the more they will understand about how to manage their condition. Eventually this becomes an almost subconscious task.

## Feeling exhausted

Sometimes, even after you have achieved second wind, you can feel exhausted and find that you can only take say 10 or 20 paces before having to pause again. This keeps happening and it doesn't seem to get any better.

This can happen if you are walking at close to the limit of the rate that your aerobic pathway is supplying energy. You are using up your immediate ATP\* and it is not having time to recharge aerobically. (This may be what some people describe as having a "bad day" in terms of their McArdle's.) But don't give up, it is just necessary to have a longer pause. If possible, sit down so that your legs are fully rested, and have something to eat – preferably not refined sugar but something like a banana or a cereal bar. Take perhaps 5 or 10 minutes and then start again slowly. You should now find it possible to keep going steadily without the feeling of exhaustion and those too-frequent rests. This is because your immediate ATP has had time to recharge and as long as you don't push your pace ahead of the energy that your aerobic metabolism can supply, you should be fine from now on.

\*ATP is a late step in creating energy and you have some available in the muscle. Normally it can be recharged aerobically or anaerobically, but in McArdle's only aerobically. See page 10.

## Use walking poles

Many walkers find walking poles very helpful, and they are especially so in McArdle's. Using two walking poles eases some of the demands on the leg muscles particularly uphill, thus helping to keep you in the aerobic phase. They also help reduce the resistance load on the muscles when



*Good technique with walking poles makes a big difference.*

going downhill, and give confidence and stability on difficult terrain.

Telescopic aluminium walking poles are very light weight and comfortable to use. Having someone show you the correct use of them is very helpful. Using walking poles to get your arms into second wind and sustain exercise in that phase may help to avoid the upper-body muscle wasting which appears to be more prevalent amongst McArdle people than the rest of the general population. [Video]

## Choose a good backpack

Choose a backpack with a waist strap and maybe a chest strap. Then load it with the weight high up and you will take most of the weight on your hips, thus reducing the stress on your muscles and



*A stable, well adjusted backpack will save muscle strains.*

enhancing stability. An unstable pack can cause you to make an anaerobic movement to counter the pack's instability.

## Stop as soon as you need to

Due to the fact that McArdle pain lags behind the damage we are doing to our muscles, we need to become very attuned to the signals which we get. With practice it becomes second nature to adjust to the signals and avoid damage. We have to obey the signals. For instance, if we are on a steep slope and our muscles tell us that we have to stop, it is no good thinking "I'll just get to that next rock". We have to stop immediately. Just a few seconds past the stop signal and damage is caused which can last all day, or even longer.

## Slow down on reaching an incline

When an incline is started the demand on the muscles is increased. If you were already walking at your maximum aerobic capacity you are going to have to slow down to avoid your muscles trying to go anaerobic. It is good to make a conscious decision to slow your pace just before the incline starts. If you don't do that you may find the muscles tightening up and must then respond to that by pausing for a short rest.

Another factor in changing to a steeper incline is that you may then be using slightly different muscles which are not in second wind. You have to take the usual care to get those new muscles into second wind.

If you haven't been using your walking poles on the flat, now is a good time to get them out of your pack and put them to use on the slope.



*Finding intermediate places to step up.*



*An ideal wide path for zig-zagging up the incline.*

(Better still, first use them on the flat so as to get your arms into second wind.)

## Steep slopes - zig-zag or contour

It is impossible to get up very steep slopes aerobically, no matter how well trained our muscles are. The most useful technique to assist is to 'zig-zag' back and forth across your route. That way you can, for example, reduce a slope of 1 in 3 to 1 in 6, or even lower if you zig-zag more. You can thus keep within your aerobic capacity. You can use this even on a road or lane – each 'leg' of the zig-zag may be only a few paces, but it still works. However, keep a careful watch out for cars! If the width of the slope allows it you can take a long gently ascending traverse rather than zig-zagging. [Video]

## Stepping up in small steps

On very steep paths and on rock-strewn open terrain there can be high steps to negotiate, which others will manage with no great bother. Each step involves lifting our own body weight vertically for the height of the step, and this is bound to be anaerobic. With McArdle's we need to try to avoid these steps and walk on a graded slope. If this can't be done we need to seek out interim places to put our feet, maybe at the side of the path, so that we lift our weight the same distance in two lifts rather than one. This helps us to keep the activity as aerobic as possible. [Video]

Sometimes on steep slopes slowing the pace and using the above techniques still leaves you working too hard to be fully aerobic. You may still need to take plenty of short rests.





*Surfaces affect our muscles, especially if tensed against slipping – sand, pebbles, seaweed, long grass, wet grass, mud.*

## Environmental factors

With McArdle's we are more affected by environmental factors, so we need to be sensitive to them. A headwind can turn your normal aerobic pace into something anaerobic. Surfaces such as soft ground, mud and long grass can demand quite a lot more energy. Extremes of temperature, both hot and cold, can affect muscle performance.

We need to be sensitive to these factors and adapt our activity, alter our route or slow our pace accordingly.

## Avoid long breaks

Our second wind tends to be lost in about 30 minutes. Therefore it is best to play safe and avoid stops of more than 20 minutes or so. While walking with non-McArdle's people this is a clash of style. You need to stick to your shorter breaks and explain to the others why this is. If the terrain is safe, you might agree to start off before the others and let them catch you up.

## Rest before the top

Many hill walkers will want to get to the top and then take a long break, say for lunch. With McArdle's we instead need lots of little stops on the way up. If there is going to be any more ascent on the route, we need to quickly pass over the top so that we can get to the next ascent before we lose the second wind in our up-hill muscles. If a longer stop is required, best to take it before the top so that we can boost our up-hill muscles again in that last section of ascent. In this way we can minimise the time between up-hill sections and thus reduce the risk of losing the second wind in our up-hill muscles.

## Be up-front about your condition

Walking with others who have McArdle's is a dream compared to walking with people who do not have it. Don't be shy about telling your fellow walkers about your McArdle's. Much better to

explain over a pint in the pub the night before, than start explaining when you are on the walk.

Walkers will tend to be people who are interested in exercise and they will be fascinated by your story. People will not fully understand, but they will realise that it is a serious issue and they will be happy to make allowances for you. You then will not feel under pressure to push yourself beyond your limits and thus will avoid damage.

## Stamina

With regular walking every day you will improve your capacity. Stops will be less frequent and hills will seem easier. You may also find that getting into second wind becomes a bit easier, although it will always take around 7 to 10 minutes.

## Eating and hydration for walking

On a big walk day you may need to consume more calories than you normally do, but otherwise should be eating a healthy balanced diet. Some people find that sugar before starting seems to help them get going, although others find it not necessary. Regular high sugar consumption must be avoided as it will lead to weight gain as you are very unlikely to burn off all the calories.

Because we need to avoid long breaks (see above) we should eat little and often rather than eating all our lunch in one go and then needing to wait for it to be digested before moving on.

Drinking enough fluid is very important, especially in hot weather. Dehydration can lead to muscle damage, so whilst hydration is important for anyone it is extra important for us.

## Avoid tense muscles

Fear or panic (or even over-excitement) can cause the muscles to be tense. In that state almost anything you do will be at least partly anaerobic and thus lead to muscle damage. Know your limits and plan your route to avoid situations which will cause you to be tense.

# Guidance for walking partners

**There are dangers for McArdle people in walking with others who do not have the condition. It is therefore a good idea to brief your unaffected walking partners as to what to expect and how to respond. Suggest that they read this page, or just tell them the key points as set out at the end.**

## “Can I help?”

You certainly can! It is highly beneficial for people with McArdle Disease to have plenty of aerobic exercise such as walking. Yet for them the start of exercise is difficult and has to be carefully managed to avoid pain. You can help them by encouraging them to exercise, preferably for at least 45 minutes at a time, at least five times per week. Calm, patient and unfussed support whenever you can will encourage them a lot.

## “We’ve only just started, surely you can’t be tired already?”

The first minutes of a walk are the most difficult for people with McArdle’s, so you need some patience to put up with a slow start and probably some short rests of 30 seconds or a minute. They need to be very slow and careful with their McArdle’s symptoms in the first 10 minutes, then they should get into “second wind” and it gets a bit easier after that. In fact their symptoms can get even easier during the day. But of course they then get normal tiredness, just like anyone else.

## “This is boring, can’t we start off up that hill?”

Even starting off on the flat can be a challenge for McArdle people. Starting off up hill is madness, unless they are very fit and aerobically conditioned and are extremely careful. It is best to plan any walk to start on the flat for about a mile, so there is a good opportunity to get into “second wind” before tackling an incline. That will make it easier and safer.

## “Are you OK? Do you want to sit down? Shall we turn back?”

Often, when a McArdle person stops for a rest it will be just for a few seconds. There is no need to make a fuss, or even comment, as that just draws attention to the fact that they are holding up others. That in turn is likely to make them feel obliged to set off again before their muscles have recovered sufficiently, or even worse, not to stop

## Quick summary

- Let the McArdle person lead; don’t get ahead of them.
- Expect a slow start, and on the flat.
- Allow time for a very slow pace on up hill sections.
- Rest whenever they want, don’t press on to a better stopping point.
- Stop along with them and don’t fuss over them.
- Avoid a long lunch stop, but have several 10 to 15 minute stops with a small snack.

the next time they need to. So once a McArdle person stops, just stop with them without comment. Carry on the conversation or look at the view. Don’t stand in front of them but to one side, so that they can set off again when they are ready without needing to say anything.

## “It is getting dark, can’t you hurry up?”

McArdle people can walk up hills, but their energy supply is compromised so they have to go very slowly - at perhaps a quarter of the speed of a



*Any challenge is likely to be more challenging for us.*

normal walker. Most find a very slow, steady plod is effective but others go a bit quicker and have many short rests. Either way, they can manage hills. On the flat and downhill they are not very different from any other walker. So allow a bit more time, have some patience on the uphill sections and you'll get there in the end.



*Our rest pattern is very different to that of other people.*

### **“That big rock is a good place to stop for a break, can't you press on to that?”**

When someone with McArdle's decides they have to stop, it has to be right then. The sensations can change from being OK to needing to stop within just a few seconds, and once the signals are felt they have to stop. If they stop at the “right” time the rest might just be for 30 seconds or a minute, if they push on against pain even for just a few yards they will need a much longer rest and can even get a fixed contracture which will put paid to the rest of the walk.

### **“Why do you always have to be in front?”**

A McArdle person should be allowed to set the pace. If someone else is leading they are almost certain to end up some distance ahead, especially when walking up hill. This puts stress on the McArdle person. Stress = tense muscles = anaerobic activity - which is damaging for McArdle people. When they need a rest they will feel under pressure to keep going. And if the person ahead is out of hearing range, if they do take a rest they will drop even further behind. It is much better to allow the McArdle person to be up at the front, to set the pace and choose the rests.

### **“I haven't finished my lunch. Do we have to go so soon?”**

McArdle people will not want to stop for more than about 20 to 30 minutes at a time, as beyond that they are at risk of losing their “second wind”.



*Our lunch break needs to consist of several short stops.*

That would mean another session of slow, careful walking, juggling between pain and pauses in order to get back into “second wind”. So a long leisurely lunch stop of an hour or so is out. Instead they need several short stops with a small snack at each.

### **“Don't they say ‘There's no gain without pain.’?”**

It's a common phrase but for someone with McArdle's it is just plain wrong. For a McArdle person, pushing on against pain will result in a cramp, muscle damage and possibly a fixed contracture with rhabdomyolysis. In severe cases acute renal failure can follow. This phrase may be meant as an encouragement, but it is best left unsaid. See more detail in this section.

## **To conclude**

Of course, other people can have needs as well and it may be that they will need to take precedence over the McArdle person's needs. But if the rest of the group are normal healthy walkers, then all the points above should apply.

### **Case report**

- A woman brought her unaffected husband on a walking course for couples.
- He was able to compare notes with other partners.
- He came to a clearer understanding of all these points.
- They now enjoy country walks together.

# Rules of the road

When people with McArdle's walk together, such as on a walking course, we have some common sense "Rules of the road" to look after everyone.

When walking in a group of people who don't know about our McArdle's, it is very easy for us to damage ourselves as we do not want to hold people back, make a fuss, etc. Even though we know we have to say something, we can sub-consciously suppress that and end up doing too much, going those few yards too far before stopping. The result can be a painful fixed



*Rules of the road in action, looking after each other.*

muscle contracture which can last for hours or days. If it is severe it can make us feel very unwell and possibly even land us in hospital.

All the walkers in a McArdle's walking group need to protect themselves from possible muscle damage. Apart from good techniques, the key to this is a set of rules which we all need to stick to - and we willingly comply as we all understand.

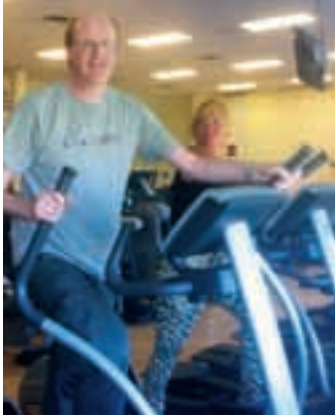
The more that those of us with McArdle's walk, the more we get to understand our muscles, to know the signs, to know what we can do and can't do, to know how to avoid problems. By walking as a group we can compare notes and help each other to a fuller understanding of all these aspects.

## The rules are:

- Always walk at the pace of the slowest person.
- If someone else stops, just carry on slowly, unless...
- If you are the last person, then stop with other person.
- Never mention the stop, just talk about something else.
- The people in front should watch out for those behind and slow down, so as not to put pressure on the ones behind to keep up.
- If the group has spread out more than 150 yards or so, call out for all to stop.
- Usually 30 seconds rest is enough for the sensations to dissipate. However, if the person does not wish to continue after that, the group will take a 5 minute break.
- We will anyway stop frequently for short breaks.
- We will have pick up points wherever we can.
- If anyone needs to, we will all abandon the day's walk without complaint.
- The leader's decision is final!

# Strength training

Researchers in Spain have published the results of trials in strength training for people with McArdle's. This is now offered in clinic.



*Getting both lower and upper body into second wind.*



*Three quick repetitions lifting maybe 100kg, then rest for five minutes.*



*A few repetitions, high resistance, then on to the next equipment.*

It has long been said by doctors that people with McArdle's should not undertake anaerobic activity such as lifting weights.

## Genesis of the six second rule

However, I knew just how often I had done heavy lifting and carrying, but always in very short bursts. I once had to clear large stones in my stream after a flood. I would lift one, hold it tight to myself and as I carried it I would count the seconds. By about 6 or 8 I would drop it, even if I wasn't where I wanted it to go. I would get a break as I walked back and rested a little more before starting again with the next large stone.

After I discussed this event with an exercise physiologist, I developed the "six second rule" to help others understand the limits of what they could do. (See page 17.)

## Strength training and muscle mass

Now a team of researchers led by Alejandro Lucia and Alfredo Santalla in Spain have, in their own separate development, devised training routines that take advantage of this same physiological feature.

This training can build muscle mass and strength. Additional muscle mass is also

beneficial in everyday activities as, there is less stress on the muscle for any given task.

## Rapid repetitions

First the pupil gets into second wind on both the lower and upper limbs.

They can then progress to brief sessions of anaerobic activity, usually just three or four rapid repetitions of high intensity resistance exercise; followed by about 5 minutes rest to fully recharge the ATP and phosphocreatine system in the muscle cells. They can then repeat the earlier repetitions.

If exercising in a gym with suitable equipment available, it is possible to use the rest period to exercise a completely different set of muscles.

## Supervision needed

At present this type of exercise needs to be closely supervised by an expert physiologist who fully understands McArdle's. That is difficult for most of us to achieve, but maybe in the future it could be supervised safely by personal trainers, opening up the benefit to a much greater number of people.

# Reference section

About the walking courses, a reminder to look after ourselves, medical emergencies and further reading.

## About the walking courses

There are seven years experience of running courses. Now IamGSD is building a network of expert patients to offer courses around the world.



*Informal tutorials and discussions along the way.*



*Starting a little experiment!*

After completing 210 miles (338 km) and countless hills, the people who participated in the 'Walk over Wales' in July 2010 - Andrew Wakelin, Stacey Reason, Dan Chambers and Andy Williams - realised that walking as a McArdle's group was enjoyable and confidence boosting. This

I have learnt more about my McArdle's this week than I did in 30 years from my diagnosis.

**GABI JOHANN, GERMANY**

positive experience led to the development of a walking course - 'Walking with McArdle's', starting in 2011.

The focus of these courses is to provide individuals with an opportunity to meet other affected people and share experiences, whilst gaining practical knowledge on how best to manage not just walking but day-to-day life with McArdle's.

### The objectives of the walking courses are to:

- Develop good management of the person's McArdle's.
- Improve aerobic fitness.
- Teach a range of techniques and approaches to improve performance and minimise the risk of injury.
- Identify and eliminate any bad habits that have developed.
- Extend each person's personal boundaries.
- Boost each person's confidence.
- Make walking enjoyable and establish it as a habit.

Originally, anyone with a confirmed diagnosis of McArdle Disease could participate. Now that has been widened to include Tarui Disease and people with other muscle GSDs are welcome to contact course organisers to discuss their possible involvement. People of a wide range of fitness and abilities, and of ages from 5 to 73, have participated.

Through informal discussion and practical experience, these week-long courses aim to develop the best techniques for living with these conditions. Strategies for achieving 'second wind' and learning to

You taught me that I'm capable of steps I never knew I could take – not just steps of walking but those of courage and confidence.

**SIONED HOSSEINIAN, UK**

Just finished my 4 mile walk today. I love the trekking poles that I learnt about on the course. I feel so much better when I walk.

**HARRIET MCGINNIS, USA**

use walking poles are two of the key topics .

With improved aerobic fitness, the risk of muscle injury is lessened – in turn, individuals will gain confidence in their abilities and increase their motivation to undertake regular exercise. Many have considerably extended their boundaries.

The walks are carefully planned to suit the capabilities of all members in the group.

I've never felt so strong, physically capable, hopeful and inspired. There is such benefit in sharing our knowledge and experience.

All that and fun too!

**SIOBHAN MURRAY, USA**

Opt-out points are planned along the way, so that everyone can confidently join the walk for whatever distance best suits them.

Already people from 15 countries have participated. Courses have been offered in UK, USA and Italy. It is planned that this initiative will continue to develop and grow.



Lucy, age 5, on the first "Children & Parents" course.

The course was great - health wise, instructive, fun and meeting new people with McArdle's and sharing experiences.

**WILLIAM O'NEILL, IRELAND**



Enjoying the scenery as well as sharing experiences.



The courses teach all the tips such as "rest before risk".

# The McArdle Mantra

This is a set of phrases which make it easier for us to remember things that are important to looking after ourselves. Some of these concepts will be very familiar to people with McArdle's, but others are perhaps less well known or understood.

- Stroll before Second
- Pause before Pain
- Embrace our Embarrassment
- Rest before Risk
- Stop after Six
- Shorten our Static
- Avoid the Awkward
- Reduce our Repetitions
- Think before Tasks
- Temper our Tension
- Halve our Hurry
- Eat before Effort
- Aim for Activity
- Extend our Exercise
- Watch our Weight
- Mind our Medications
- Carry our Card

## ■ Stroll before Second

“Second Wind” is an essential building block of McArdle's exercise. We must go slow and steady in the first 7 or 8 minutes of exercise or activity until we get into what we know as “Second Wind”. Without this we are at greater risk of injury. Read all about second wind on page 14.



*An easy stroll at the start, to help achieve second wind.*

## ■ Pause before Pain

Whether in that first phase of activity or well into second wind, it is very easy for us to over exert ourselves and empty our ATP stores. As our muscles run out of energy we feel it - our limbs will get heavy and the muscles will start to cramp up. We must react to those signs before they turn into pain. We must pause to allow the ATP reservoir to refill. If we ever incur pain which lasts for more than a minute after we pause, we have overdone it. Read about the ATP reservoir in the page on adapting activity, in this section.

## ■ Embrace our Embarrassment

We all get embarrassed when we have to stop on a slight slope as someone three times our age powers past. Strangers are watching and wondering what's up with us. We have to get over it! What does it matter anyway? Enjoy the moment – keep them guessing, create a bit of mystery and intrigue. Learn to love it.



## ■ Rest before Risk

We can easily be at the point of exhaustion whilst just walking along on the flat. Or maybe we are in second wind but are going slightly up hill or into a headwind. The ATP reservoir in our muscles (see above) is almost empty. Just at that point we simply can't make any extra effort. If we are just about to cross the road, we should stand back from the curb, rest for 30 seconds or so before we cross. Alternatively, if we cross without that deliberate rest we can be sure that a car will come around the corner and we won't be able to get out of the way. Putting the crockery away? Take a rest before lifting the last heavy plates into the top cupboard. Sawing logs? After getting the next log into position take a rest before picking up the chain saw. Rest before risk. See page 18.

## ■ Stop after Six

When we do something at maximum effort (like lifting something heavy or sprinting fast to catch a bus) the immediate energy supply in our muscle cells will be exhausted in about 10 seconds. If we carry on we will get a fixed contracture. It is best to avoid these tasks altogether, but sometimes daily life can be very demanding. If we must do something like open a new jam jar, we should count "one thousand, two thousand" etc as we do it. If by six it is not open, we must stop, put it down and rest or do something gentle for 5 minutes by which time the ATP reservoir should have refilled and we can try again. Read more about the "Six Second Rule" on page 17.

## ■ Shorten our Static

Static exercise (also known as isometric exercise) is the worst kind for us. This is when we are



*Static (isometric) activity like this is the worst for us.*

expending effort but without the muscle moving. It is an anaerobic activity because the blood flow to the muscle is disrupted whilst the muscle is contracted. Take two examples of using our legs. Whereas walking is not static, standing on a ladder is static. For our arms, in DIY, whereas painting a wall is not static, holding up a ceiling tile whilst someone else fixes it is static. We really need to avoid this type of activity, adapt it, or at least shorten the duration. So in the ladder example, we should use a ladder with steps rather than rungs; put more weight on one leg and then alternate between legs every 10 seconds or so; try to break the job into sections with a rest between each; if we can't manage with these adaptations then we must get someone else to do the job.

## ■ Avoid the Awkward

Holding an awkward position can be damaging even if there appears not to be a lot of effort involved. Turning our necks to reverse the car; holding something out at arm's length, squeezing onto a packed train carriage with our arm awkwardly reaching past someone to grab a



*Even here a "pause before pain" may be necessary if our pace is a little too fast.*

handrail; leaning forward to look closely in the mirror to put on make-up. These are examples of activities that will be anaerobic, will start a cramp within seconds and, unless stopped, will cause a fixed contracture within minutes.

## ■ Reduce our Repetitions

Quickly repetitive actions are a problem for us. Getting into second wind beforehand helps. If we can reduce the speed or the number of the repetitions it helps to avoid problems. So if grating cheese – we should do a bit, change hands, do a bit more, take a break and do something else then come back to it in a few minutes. If using a screwdriver, we should do a few turns, take a break, do a few more. Chewing is another problem area; we can't really reduce the number so we just have to take a rest mid chew!



*Repetitive tasks may need to be broken into stages.*

## ■ Think before Tasks

We need to think ahead in order to avoid problems. Many tasks of daily living can pose a threat if we don't think ahead, or at least respond to the sensations in our muscles. Variation is a major help, so we should think through the task in hand and try to break it down into stages so that we can do something different between each stage, using different muscles. (As an example: getting my lawnmower into the boot/trunk of the car to take it for service can cause fixed contractures in my arms. So I first use my arms to open the boot and clear a space. Then I change to using my legs - walking to get the mower and pushing it to the car. I might then take a break or do something else in the garden for 10 minutes. Now I return to the car and lift the mower onto the tailboard. I walk away again because my arms will be almost out of ATP through that lift. Finally, I return and manoeuvre the mower into position in the car.)

Most tasks can be adapted or broken down into stages to enable us to do them despite our McArdle's. If that can't be done safely we must ask for assistance, as otherwise the result can be

painful and cumulative muscle damage will be a problem in the long term.

## ■ Temper our Tension

Not temper as in losing your temper, but temper as in softening metal. If our muscles are tense they have to work anaerobically, as the blood supply is greatly restricted. As we cannot make use of the main anaerobic energy pathway, we are likely to incur muscle damage with remarkably little physical activity. It is easier said than done, but we should try to avoid getting angry, afraid, losing our temper, getting over excited, getting depressed or facing a very stressful situation. All these things can make our muscles tense and then we are at much greater risk of muscle damage.

## ■ Halve our Hurry

Hurrying is one of the major hazards for McArdle people. It could be hurrying for a train, but just as easily it could be hurrying to get dried after a shower or hurrying to get the children to school. When hurrying we are likely to use more energy than is being supplied by our aerobic pathway and thus easily empty our ATP reservoir. As the cramping sensation lags behind the damage we are doing, we may not realise what is happening until we have hurt ourselves. We must plan ahead and allow time. If we find ourselves hurrying, we should ask ourselves 'does it really matter?'

## ■ Eat before Effort

Because we can't use the energy stored in our muscles we are reliant on energy coming to the



*Mostly people need breakfast before activity.*

muscles in the blood supply. Because everyone fasts overnight, for around 8 to 12 hours, blood sugar levels are going to be low in the morning. For McArdle people it is more important than ever to have some breakfast to start fuelling our



*Build activity into your life. You can walk to work rather than trek in the Italian Dolomites.*

system before we get into activity or exercise. Without some food our symptoms are likely to be worse. A similar situation applies during the day - if we are active all day it is better to have several small snacks than either skip lunch or stop for a long lunch break. See page 42 for more on diet.

### ■ Aim for Activity

The times that we find it most difficult to achieve second wind is when we have been inactive for a long period. If we can build a bit of activity into our day it will help when the time comes to start more serious activity or exercise. We need to guard against becoming sedentary in our job and in our day to day life, as that will lead to a loss of aerobic capacity and then almost anything we do is painful and damaging.

### ■ Extend our Exercise

We need to exercise for about 45 minutes about 5 times each week to build and maintain our aerobic capacity. This will not only make exercise easier and extend our abilities, but will also help to protect us against injury. If we are not used to doing that, we need to build up towards it in gradual stages. There is some guidance in this section to help you get started on your exercise programme.

### ■ Watch our Weight

Being overweight is a significant disadvantage for us. It puts extra stress on the muscles and thus makes our symptoms worse. That in turn leads to us finding it harder to exercise, and lack of exercise makes us lose aerobic capacity making our McArdle's worse still. Lack of exercise makes it harder to keep our weight in check. We need to

look up the recommended weight for our height and aim to get close to it and maintain it. There is a chart available on the NHS Livewell web site.

### ■ Mind our Medications

We need to be very wary of medications. For example, statins and steroids may pose more risk to us than to others and should be used very cautiously. Before taking ANY medication we need to read the insert and, if the possible side effects include muscle pain or weakness, ask questions of our doctor. Some doctors prescribe medications and supplements which they think will help with McArdle's, but the Cochrane Review has shown that none are yet *proven* to be effective. We should not take pain medications before activities as they can mask the muscle pain which is the signal to slow down or stop.



### ■ Carry our Card

Keep the McArdle's Information Card handy with its reminder of what to consider if we have overdone things. There are pointers to when we need to seek medical assistance. The card supports us in seeking assistance, such as at an airport, and is a handy way to explain the condition to others. Available from AGSD-UK and support groups in other countries.

# Diet for McArdle's

**In the area of diet and supplements, so far there has not been any convincing scientific evidence of a good answer for McArdle Disease.**

Diet is still a highly controversial area, with no good evidence to support any particular approach. Some patients anecdotally report significant improvement with a high reliance on each of the three macro nutrients. This disparity may just highlight that diet is probably less important to us than exercise.

The starting point is to greatly reduce all processed and manufactured foods. Rely instead on fresh, natural foods as much as possible. After that you may like to try a few things and see what suits you best.

Here is a brief summary of the current position regarding research.

## Higher than normal protein - 1985

In the past, a high protein diet was recommended. That arose from its use in unrelated muscle conditions and a single case report in McArdle's where both diet and exercise were changed at the same time. A few consultants still recommend a high protein diet, and a small number of people find that it suits them.

## High carbohydrate - 2008

The latest published study on diet in McArdle's (May 2008) compared a high protein diet to a high carbohydrate diet. The paper recommends a diet somewhat higher than normal in carbohydrate, consisting of 20% fat, 15% protein and 65% carbohydrates, making use of complex carbohydrates for a prolonged release of energy. The study involved only seven patients and they followed each diet for just three days. So the evidence is not fully convincing. Participant numbers and funding are common problems for such ultra rare diseases as ours.

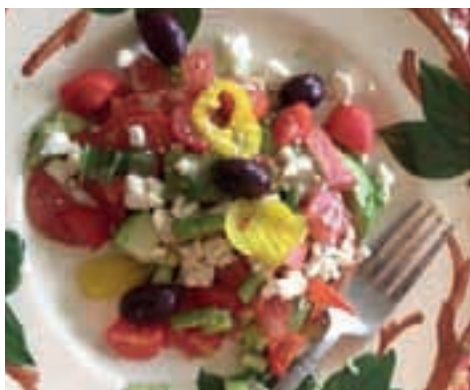
## Nutritional ketosis - from 2005+

There was a single person study that found a ketogenic (high fat) diet to be highly effective (2005). Then a study was run in 2005/6 with five participants on an 80% fat diet, but there were problems and no results were published.

At present, major anecdotal evidence from hundreds of people is accumulating, largely from summer 2014. It appears that a ketogenic diet



*Grilled salmon, courgettes and 'loaded' cauliflower mash.*



*Salad with Feta cheese, olives, avocado, oil dressing.*

(65% fat, 25% protein, 10% carbohydrate) is far more effective than anything tried before. Very significant improvements are currently reported by more people than have been in all the dietary studies ever held in McArdle's.

*The photos above are from people on a ketogenic diet, showing how well they can eat.*

A case series has been published, a pilot trial is in progress and results are awaited of a trial of an oil supplement which is a precursor of ketones. Further trials of a ketogenic diet (also known as LCHF - low carbohydrate, high fat) are being planned.

## Medical advice

For those wishing to try any diet, medical advice and appropriate monitoring is advised.

# McArdle Emergencies

**Guidance in the event of an episode of rhabdomyolysis, with an alert to emergency doctors of the attendant risks. The doctor will treat each episode according to the individual circumstances.**

## Guidance for the McArdle person

If after strenuous or unusual exercise/activity† you have one or more of the following signs:

### Dark coloured urine

This is called myoglobinuria or proteinuria and appears as reddish tea to cola coloured urine. (However, if you have eaten strongly coloured food such as beetroot/beets there is probably no need for concern.)

### Feeling very unwell after activity

'Flu-like symptoms, can be a sign of rhabdomyolysis (muscle breakdown).

### Low volume of urine

Producing a very low volume of urine, or no urine at all, constitutes a medical emergency (unless simply caused by dehydration) as the complications can become life threatening.

†A few people have experienced these symptoms without unusual activity.

### You should:

- Drink plenty of water to help clear your urine.
- Go to hospital promptly for medical assistance.
- Take a urine sample with you, if possible.

**If in doubt, telephone your McArdle Disease consultant or specialist nurse.**

## Information for the Emergency Doctor

**Background:** McArdle Disease (GSD Type V) [1] is a rare metabolic myopathy. Patients experience exercise intolerance and muscle cramps. They are at risk of rhabdomyolysis [2], which can lead to acute renal failure.

**Presentation:** Patients may appear well but have a muscle cramp, myoglobinuria or feel unwell/have fever after activity. There may be an extreme muscle contracture with swelling and often severe pain.

**Investigations:** Urgent assessment for possible rhabdomyolysis. Creatine Kinase usually grossly raised. Consider urine analysis for myoglobinuria and full chemistry panel (including CK, glucose, calcium and bone profile, urea and electrolytes).

**Management:** Consider fluid bolus followed by IV saline at 2x maintenance and (unless diabetic) possibly 10% dextrose to keep blood glucose >3.5 mmol/L. Episodes require monitoring of urine output, CK and electrolyte status.

**Complications:** Oliguria can occur requiring prompt referral for haemodialysis. In rare cases compartment syndrome [3] has developed, requiring urgent surgical referral.

**The McArdle Disease Handbook: (208 pages) is available for searching and browsing via Google Books, free of charge.**

## More information about McArdle Disease episodes

### Fixed contractures

It is likely that you will have a fixed contracture (spasm) in one or more muscles. This is when the muscle contracts and then fails to relax again. It becomes hard or stiff, swells up and usually becomes very painful. You must *not* try to force the muscle – e.g. do not try to forcibly uncurl the

fingers of a “clawed” hand. This may tear the muscle and cause a lot more damage. Cooling the affected muscle, for example with an ice pack, may slow the development of symptoms. If the muscle is not in a fixed contracture but just sore and enlarged, some very gentle massage may also be helpful.

## Myoglobinuria and rhabdomyolysis

Myoglobinuria (myoglobin protein in your urine) is a product of rhabdomyolysis (broken down muscle). If you think you may be having an episode, urinate into a clear or white container so that you can easily monitor the colour of your urine. The depth of colour will depend on the amount of muscle damage you have done. That is, how severe the contracture is and the size of the affected muscle/s. It will also appear darker if you are dehydrated as it will not be so diluted in your urine. The darkness of the colour is what matters, not the length of time that the myoglobinuria lasts. Do not wait to see if it grows paler, if it is dark, get to hospital.

Myoglobin gets into your blood from the damaged muscle and can then block the kidneys as they filter your blood to produce urine. Your kidneys not only remove excess fluids but also regulate various chemicals in your blood. Having your kidneys blocked by myoglobin (acute renal failure) can be life threatening.

### Drink water

Immediately start drinking water at the rate of roughly 500ml (1 pint) per hour to flush through your kidneys and dilute your urine. Remember that over-hydrating can also be dangerous. If available, add a pinch of table salt and a level teaspoon of sugar.



*Hospital visit for IV fluids plus renal function and CK tests.*

### Going to hospital

You should go straight to hospital, not to a family doctor. If you are not producing much urine, immediately on arrival tell them that you are suffering rhabdomyolysis which may be causing acute renal failure. If you can take a urine sample, show this to the triage (assessment) nurse/doctor as soon as possible. A dark urine sample will

immediately highlight to them that urgent treatment is required.

You may hold a stand-by letter from your McArdle consultant for such emergency situations - if so, take that with you, or take your McArdle Information Card. But failing that, draw the attention of the emergency doctor to the notes in the box above.

Another option is to wear a MedicAlert bracelet, which will be engraved with your medical conditions and an emergency phone number for doctors. The call centre will hold what medical records you wish. See MedicAlert's UK website. They also operate in other countries.

### Immediate attention

You should have blood and urine samples taken straight away. You may be put on an intravenous drip (a bottle of fluid connected to a needle in a vein in your arm). This will ensure you have sufficient fluids to flush your kidneys. Your urine output will be monitored and this may involve having a urinary catheter (a drainage tube inserted into the bladder and attached to a clear collection bag) so that your urine is continuously collected and can be easily monitored.

### The test results

Your test results may take an hour or two to come back. The three things which are of particular interest to the doctors are:

#### 1) Myoglobin in the urine

If present this confirms that you are having an episode of rhabdomyolysis.

#### 2) Creatine Kinase (CK) level

The normal range for unaffected people is about 100 to 200 iu/L. However, people with McArdle's have a base level of an average of 2,500 iu/L. The higher the level the more muscle damage you have incurred. Figures over about 10,000 to 20,000 may require you to stay in hospital until the level has fallen again.

#### 3) Kidney function tests

These will help the doctors assess how well your kidneys are dealing with the myoglobin released from your damaged muscle into your blood.

### More about your CK

The CK (also known as CPK) results in these circumstances can be very greatly raised. It is common to see levels of 40,000 and sometimes as high as several hundred thousand. The CK tends to peak at about 24 hours after the injury was

incurred, so the doctors may well want to take samples regularly until the peak has passed. After that they may sample just once every 24 hours. After the peak the CK level starts to drop by about 30% to 50% in each 24 hour period, so you might see a pattern such as: 12 hours 20k, 24 hours 50k; 36 hours 35k; then each day thereafter 24k, 16k, 11k, 7k. Once below about 20k and on a downward trend the hospital may be prepared for you to be discharged.

## Pain medication

Most people find fixed muscle contractures extremely painful and you should be offered pain relief. However, when recovery is well under way you will want to start moving the affected muscles again. At this time it is important not to take regular pain relief which masks the sensations/pain experienced when using the muscles. You need this feedback from the muscles to tell you when you are overdoing it.

## Possible complications

The biggest worry is anuria (not producing urine) or oliguria (producing significantly less urine than you are taking in as fluid), which indicate acute kidney failure. If left untreated this can lead to multiple organ failure and can be life threatening. So it is very important that you go to hospital urgently if you have either of these symptoms, and that you make sure that the hospital treats you promptly.

Without anuria you may still experience acute renal failure. The signs for this are the kidney function tests (from a blood sample) and if you are producing less urine than your fluid intake. This only happens in a percentage of cases (probably less than 30%). The more muscle damage you have done the higher the risk. This needs to be treated by going onto dialysis. As long as this is done promptly, there is normally a full recovery of kidney function.

In a very small number of people something called ‘compartment syndrome’ has developed. This is caused by muscle swelling which leads to increased pressure within the fascia (strong membrane) around the affected muscle group. If untreated it can permanently damage the muscle and affect the circulation. Surgical intervention is usually required to relieve the pressure.

## The recovery

Depending on the level of muscle damage caused, after being discharged from medical care

it may be sensible to take a few days rest and not to exercise the damaged muscle until it feels fully recovered.

As long as you follow the guidelines above, especially about getting prompt attention if you have acute renal failure, there is every reason to expect a full recovery. Kidney function usually returns to normal once the by-products of muscle breakdown have been flushed through. The muscles are very good at recovering, although less so as we get older. However, repeated episodes of major muscle damage should be avoided as they are believed to cause cumulative damage if repeated over many years.

## The future

It should be our aim to avoid emergency episodes by working to improve our aerobic capacity and by learning strategies to cope with life without



*Abseiling, with expert guidance to avoid rhabdomyolysis.*

putting ourselves at risk. See page 10 for help in adapting activity to avoid injury.

Building aerobic capacity is probably the best protection against these painful, inconvenient and costly episodes. If you do not already recognise and use the “second wind” there is information available to help you do that. If you are very de-conditioned, undertaking a careful programme of gentle aerobic exercise, gradually increasing in length and intensity, is the ideal way to achieve a good aerobic capacity. When some isometric activity can't be avoided (like lifting something heavy), adhering to the “six second rule” will help to protect you against injury.

[1] GSD Type V (McArdle Disease) on eMedicine Wayne E Anderson, DO, Assistant Professor of Internal Medicine/Neurology

[2] Rhabdomyolysis on eMedicine Sandy Craig MD, University of Carolina

[3] Compartment syndrome on eMedicine Stephen Wallace, MD, Department of Emergency Medicine, Eastern Idaho Regional Medical Center

# Muscle GSD glossary

You may hear these terms used. We have tried to provide a layman's explanation of each term as it applies to muscle GSDs. For more technical medical definitions, there are free on-line medical dictionaries available.

## Acute renal failure

A sudden decline in kidney function. Requires urgent medical attention. In McArdle disease it can be caused by the breakdown of muscle from anaerobic activity.

## Aerobic activity

Activity which requires oxygen to assist in converting fuel sources into energy. Walking is a good example of aerobic activity. This is good exercise for those with McArdle disease.

## Anaerobic activity

Activity which does not need oxygen to utilise fuel sources – weightlifting or opening a new jam jar. This type of activity requires the conversion of glycogen, which McArdle people cannot do. Anaerobic activity of more than a few seconds must be avoided as it is damaging to the muscles.



*Avoid anaerobic activity of more than a few seconds.*

## ATP (adenosine triphosphate)

A molecule in muscle cells which serves as an energy source for the metabolic process.

## Autosomal recessive

The type of inheritance by which some genetic diseases including McArdle disease are inherited.

## Carbohydrate

A type of compound, such as starches and sugars, found in food. Broken down in the body to form energy.

## Compartment Syndrome

Rare complication of a contracture. Swelling within the fascia enclosing a bundle of muscles

resulting in excessive pressure, restricting the blood flow to the muscles and nerves. A medical emergency which requires urgent treatment.

## Creatine kinase (CK or CPK)

An enzyme which is used in the formation of ATP in muscle. People with McArdle disease tend to have a raised level of CK in their blood and this is often an early sign that something is wrong. In severely damaged muscle from excessive anaerobic activity the CK level can rise to many hundreds of times its normal level.

## ECG (electrocardiogram)

A recording of the beating of the heart made by placing sensors on your chest and limbs. It is printed out as a trace or graph. There is no evidence of heart involvement in McArdle disease but an ECG is a useful general health check.

## Enzyme

A protein which the body uses to make a chemical reaction happen. Myophosphorylase, which is missing in McArdle patients, is an enzyme.

## Fixed contracture

A condition where the muscle has fixed high resistance to movement, or spasm. It takes some hours or days for the muscle to relax again.

## Glucose

The end product of carbohydrate metabolism and also found in certain foods such as fruit. The chief source of energy.

## Glycaemic index (GI)

A number representing the ability of a food to increase the level of glucose in the blood. Low GI foods are beneficial for people with McArdle's.

## Glycogen

The form in which glucose is stored mainly in the muscles and in the liver. It has to be converted back to glucose to be used for energy in the muscles. People with McArdle disease have large stores of glycogen in their muscles as they are unable to convert it back to glucose. Their liver stores are normal.



## Glycolysis

The conversion of glycogen and glucose, via a series of steps, finally into ATP which energises the muscle. The process does not use oxygen and is thus anaerobic. People with McArdle disease have problems with this process.

## Isometric activity/exercise

Muscular action in which tension is developed without contraction of the muscle. Also known as 'static' exercise as there is no movement of the muscle. For example: clenching fists, holding something up or pushing. This is the worst type of activity for McArdle people.



*Isometric (static) activity is the absolute worst for us.*

## Lactic acid

An acid which is made as a byproduct of the muscle using carbohydrate. Normally there is a rise of lactic acid in the bloodstream on exercise, but in McArdle disease it does not rise. The lack of this rise can be a help in diagnosis.

## Malignant hyperthermia

A severe form of fever caused by a reaction to certain anaesthetics and muscle relaxants. Those with McArdle disease are at an increased risk of malignant hyperthermia and should always tell their anaesthetist about their McArdle disease before having a general anaesthetic.

## Metabolism

The process by which energy is made available for use in the body.

## Mitochondria

A very small organ within the cells of the muscle which is responsible for energy production from fuels. Through regular aerobic exercise the number of mitochondria can be increased, which boosts the aerobic capacity of the muscle.

## Myalgia

Pain in a muscle or muscles. This is the main symptom of McArdle disease.

## Myoglobin

A protein found in red skeletal muscle.



*Normal urine through to the very worst myoglobinuria.*

## Myoglobinuria

Presence of myoglobin in the urine. Muscle damage releases myoglobin into the blood and the kidneys remove it from the blood to the urine. An excessive amount of myoglobin in the blood can "block" the kidneys and cause acute renal failure.

## Myopathy

A disease of the muscle. McArdle Disease is a myopathy.

## Myophosphorylase

The muscle type of phosphorylase. There are also brain and neo-natal forms.

## Neuromuscular

Of the muscle and nerves. People with McArdle disease may be diagnosed and/or cared for by a neuromuscular consultant.

## Phosphorylase

An enzyme used in the conversion of stored glycogen to glucose so that it can be utilised for energy.

## Protein

Complex organic compounds found in the body and in foods such as meat and eggs. Consist mainly of amino acids. They serve a number of functions including as enzymes and are involved in oxygen transport and muscle contraction.

## Rhabdomyolysis

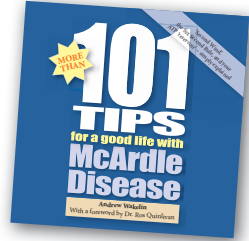
The destruction of cells in the skeletal muscles. In McArdle disease this arises from fixed spasm of the muscle caused by excessive activity such as lifting something heavy – see "anaerobic".

# Further reading

The AGSD-UK has produced these publications about McArdle's. Some are also published in other languages by Euromac.

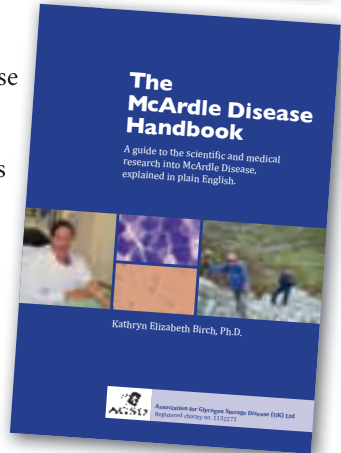
## 101 Tips for a good life with McArdle Disease

This is a handy book for people with McArdle's - "101 Tips for a good life with McArdle Disease". It is an easy to absorb guide to the many things which can be done to avoid problems and improve fitness. This pocket-sized, paperback book of 164 pages is available to order or can be accessed free of charge on-line via the AGSD-UK web site. Also available in a growing list of other languages.



## The McArdle Disease Handbook

For those who want more detail there is the McArdle Disease Handbook, by Kathryn Birch Ph.D. This 208 page large paperback is a guide to the scientific and medical research into McArdle Disease, explained in plain English. As well as printed copies to order, the handbook is available for free access online on Google Books. Also available in German and Italian from the respective patient support groups.



## One Step at a Time

This large paperback (96 pages, 200 colour images) traces the route, thoughts and emotions of a McArdle patient on a life-altering journey of discovery and growth when she walks over 200 miles. It provides a truly awakening narrative for patients, families and health professionals.

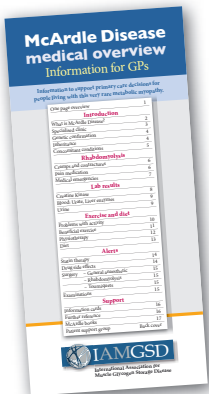


## Medical Overview

A 20 page booklet of referenced medical information to support decisions by general practitioners and other medical professionals. Also published by Euromac in 8 languages.

## McArdle web pages

Check out the IamGSD and AGSD-UK web sites for more information about muscle GSDs at: [www.iamgsd.org](http://www.iamgsd.org) and [ww.agsd.org.uk](http://ww.agsd.org.uk).



## Facebook groups

The main groups in English are "McArdle's Disease", "Ketosis in McArdle's" and "McArdle parents" for parents of diagnosed children. There are also groups in other languages.

### Opposite:

Perhaps the ultimate for McArdle's? After years of training, gaining fitness and a ketogenic diet, Dan Chambers summits Mount Shasta in California at 14,180 feet. Climbing in snow and ice conditions, over 7,000 feet of ascent and descent within 24 hours.



“At age 64 I travelled across the world for the learning, support and friendships of a walking course. I achieved more than I ever thought I could. Others can now similarly gain from this informative book.”

– Lorraine Baguley, Australia

“My walking course provided an opportunity to increase my knowledge, challenge myself, and experience first-hand the bond people with McArdle’s share.”

– Rachel Walker, USA

## Living with McArdle Disease

This book provides people with McArdle’s, and some other muscle GSDs, guidance on topics such as avoiding injury and improving aerobic fitness. There are tips for planning walks, practical techniques, “second wind”, the six second rule, “30 for 80”, the ATP “reservoir”, guidance for non-McArdle walking partners and much, much more. Then, just in case, there is advice on how to handle McArdle emergencies.



### Andrew Wakelin

is the McArdle Disease Co-ordinator for AGSD-UK and a member of the steering committee of the Euromac Registry.

Experiencing symptoms from age four, Andrew was finally diagnosed at 30, but had no help until 50. He has two copies of the common R50X mutation. He credits walking and cycling in childhood for being less badly affected than many. Using techniques he has developed, he has climbed all 188 Welsh

mountains over 2,000ft and Mount Kilimanjaro at 19,340ft.

Andrew has met over 300 McArdle’s people and spoken at conferences and workshops in the UK, USA, Canada, France, Germany, Netherlands, Italy and Spain. He liaises with the UK McArdle’s Clinic to develop their services and research. Through Euromac he liaises with McArdle consultants around the world and contributes to published papers.

Helping people to appreciate how much they can do through building aerobic capacity and using the right techniques, Andrew has devised a week-long residential course to help people develop their walking safely and enjoyably. So far, in seven years people have attended from fifteen countries. The latest development is a “Children & Parents” event.

Andrew’s book “101 Tips for a good life with McArdle Disease” and his booklet “McArdle Disease: Medical Overview” are both published in eight languages, with more in the pipeline.



International Association for  
Muscle Glycogen Storage Disease