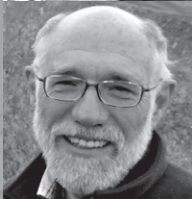


101
tips
for a good life with
McArdle Disease





101 TIPS

for a good life with

McArdle Disease

Andrew Wakelin

With a foreword by Dr. Ros Quinlivan

International English

Edition 1.1

August 2016



Euromac Registry

www.euromacregistry.eu

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Association for Glycogen Storage
Disease (UK) Ltd, 2013-15

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Frontispiece

McArdle's people come in all shapes
and sizes, in all ages, from all over the
world. Generally speaking they appear
well, but have to learn to cope with
their unseen disease.

The question mark represents the many
people who remain undiagnosed.

The people shown range in age from
15 to 64 and have all attended the
McArdle walking courses in Wales.

Left to right, from top:

Andy Williams, Singapore

Stacey Reason, Canada

Dan Chambers, USA

Karmele Ibarra Ereño, Spain

Gabi Johann, Germany

Colin Dearden, England

Cheryl Ellis, England

Andrew Wakelin, Wales

David Thompson, England

Charlton Thear, England

Jennifer Forrest, Scotland

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Arik Weingartz, Germany

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Acknowledgements

The author would like to thank Dr. Ros Quinlivan, Dr. John Buckley and all the staff of the UK McArdle's Clinic; Kathryn Birch; Nancy McVey; Stacey Reason and Terri Chambers. Thanks also to the following for ideas for tips: Dan Chambers, Jenny Coyne, Marilyn Hawker, Alison Thear, David Thompson, Andy Williams, Wendy Wilson and no doubt others from around the world who I can't recall, sorry.

Flick book animation

The story of our lives! Hold in left hand and flick with right thumb from front to back. By Stephanie Von Dressler and Andrew Wakelin.

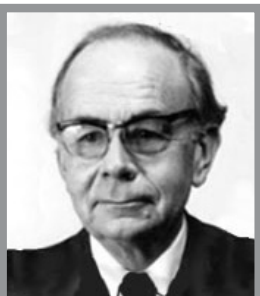
Illustrations

Photos of McArdle's people by: Andrew Wakelin, Dan Chambers, Stacey Reason, Jos van den Einde and Sally Wakelin.

Photo for tip 98: © National Hospital for Neurology and Neurosurgery.

Diagram for tip 82: Genzyme Corporation via Wikimedia Commons.

Illustrations for tips 9, 22, 31, 37, 42, 54, 87, 106, 112, 113 and 124: © shutterstock.com.



Dr. Brian McArdle published a paper in 1951 entitled "Myopathy due to a defect in muscle glycogen breakdown"¹.

The paper described a young man with a lifelong history of exertional muscle pain and stiffness – symptoms that previous doctors had dismissed as imagined. This is what we now know as McArdle Disease, or Glycogen Storage Disease Type V.

Since then the molecular and genetic basis of the disease has been identified.

Dr. McArdle died in 2002, age 91.

Foreword

The impact of McArdle Disease can be confusing. Simple everyday activities such as chewing food or hanging up clothes are enough to bring on symptoms. Yet at another time a McArdle person might hardly notice a problem with a more challenging activity. It can be difficult for an affected person, let alone a parent, partner, teacher or employer to grasp these contradictions.

Many years struggling with exercise intolerance, embarrassment, muscle contractions, maybe even kidney failure, can lead to exercise avoidance, decreased fitness and weight gain.

With careful management, noticing and responding to the sensations in their muscles, individuals can boost their fitness, improve their symptoms and make daily life a lot easier.

A partnership of medical professionals and patient groups can be very effective in advancing the management of rare and chronic conditions. This is a practical guide written by people with the condition. It will help fellow sufferers improve their quality of life.

Dr. Ros Quinlivan MBBS, MD

Consultant in Neuromuscular Disease

MRC Centre for Neuromuscular Diseases

National Hospital for Neurology and Neurosurgery

About McArdle's

A very rare muscle condition

McArdle's is a very rare disorder of muscle metabolism. About 2,000 people are diagnosed worldwide. We believe it affects about 1 in 100,000, so many more may be undiagnosed. It is inherited from parents who are 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 or drying after a shower can pose problems.

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 continues 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.

Adapting activity

To cope with necessary daily activities, 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.

The technical bit

McArdle's is also known as Glycogen Storage Disease Type V. 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.

Using this book

The tips in this book



These tips were collated from many McArdle's people from around the world. We mainly offer practical tips on how to adapt or adjust activities in order to avoid problems and manage more easily.

Hopefully, you will find some tips that are new to you. There is a glossary at the back to help you with any unfamiliar words.

It's up to you

You should first be fully confident that your diagnosis is correct. We are all affected slightly differently due to our past activities, any other complicating conditions and current level of aerobic fitness. Assess each tip individually and work out whether it suits you. Try to apply ideas from the tips in other activities.

Disclaimer

This book cannot replace personalised medical advice from your family doctor or McArdle's specialist. It is simply a supplement to that guidance. Understanding of McArdle's may change over time. On the Euromac website you can read any updates to this edition and see if a later edition is now available.

Daily living.

Adapting and adjusting little things in our daily lives can help us avoid muscle damage. This is important as, despite recovery from each episode, repeated damage can lead to problems later in life.

Six second rule

- When doing something at maximum intensity the instantly available energy lasts for only 5 to 10 seconds².
- Examples: opening a firmly stuck jam jar, standing on tip toe, lifting or pushing a heavy object, rushing up a flight of stairs.
- This relies on the muscle's own fuel store – ATP (adenosine triphosphate) – and creatine phosphate.
- When these are used up people unaffected by McArdle's then convert glycogen stored in the muscle into glucose for energy.
- McArdle's people can't do this due to lacking an enzyme called myophosphorylase.
- A painful fixed contracture of the muscle can then develop and last for hours or days.
- It is best to avoid these activities.
- If you *have* to do them, count up to six seconds.
- If the task is not complete by then, stop or put it down.
- Take a break, let the muscles recover.
- ATP will substantially recover in a minute and fully recover in about three minutes. Then try again.



There is more about the six second rule on the AGSD-UK web site.



Plan to give
yourself
extra time.
Don't rush.



Hurrying is a main enemy of McArdle's. The energy demand is too great and we risk muscle cramps. Try to plan ahead, allow more time and take things steadily.

2

Daily living.



When carrying,
hold close to
your body.



And utilise friction.



Holding items close reduces the effort needed. Pressing them against your body creates friction which transfers weight onto your body instead of your arms. Some items can be comfortably carried on your shoulder.

Swap back and forth between different activities.

And swap left and right arms.

.....

Variety in tasks means variety in the muscles used.
Each time you change, the tired muscles get a chance to recover.
For example: do a few minutes vacuuming, swapping arms,
then take out the rubbish, then return to the vacuuming.

4

Daily living.



At the supermarket
take a trolley not a basket.



Join a queue at
the checkout not
a clear checkout.

If you go in for a just a few items and take a basket you will regret it when you remember some extra items. Joining a queue (line) at the checkout will give your arms a break between unloading the trolley and bagging the scanned items.

To carry shopping bags,
put your hand
through the
handles.



Gripping the handles with your fingers is static and likely to cause a fixed contracture. Instead, put your hand through the loop of the handles to take the load on your whole hand. Or use bags with large loop handles to put over your shoulder.

6

Daily living.

Get comfortable.

Avoid spending a long time in an awkward position.



Being in an awkward position is likely to cause a cramp, so get comfortable in situations like the dentist's chair or standing on a packed bus or train. If that is not possible, regularly change position.

Break down physically demanding tasks into short sections.

For example, carrying suitcases and
packing them into the car.

.....
Limit each section to 6 seconds of maximal effort, or equivalent.

1) Carry to car. 2) Lift in. 3) Manoeuvre into position.

Take a break of 1 to 3 minutes between each section to allow
your energy store to recover. (See Your ATP 'reservoir', page 54.)

8

Daily living.



Resist peer-pressure to
do things which
you know will hurt you.

Don't feel you have to 'keep up', don't do something because it will be embarrassing to not do it. Just be your own person and look after yourself. (See tip 122.)

Try a toddler harness, so they can't run off just as your muscles give out.



If you have a toddler, try out a toddler harness/wrist strap when out and about. It will stop them running off towards traffic or other dangers just as your muscles are about to give out and you can't chase after them.

10

Daily living.

Take the stairs – slowly, of course.



We need to prevent muscle wasting, so don't avoid activities altogether. If you are able to, slowly climb one or two flights of stairs then take the lift the rest of the way. But *not* in a crowd or with anyone you feel you have to keep up with. (See tips 32, 97.)

Make activity part of your daily life.

.....
Organise your life so that you automatically stay active without thinking of it as wasted time. Can you walk to work? Go out at lunchtime rather than sit at your desk? Walk to the shops instead of driving? (See tip 121.)

12

Daily living.



Relax!



Try to avoid exercise
and activity when
tense or stressed.

Tense muscles work more anaerobically than aerobically. So be wary of exercising when tense. If you insist on going on a scary fairground ride, trust in the equipment and don't grip the bar.

If scared of heights, don't walk near edges. (See tips 52, 78.)

Ask for a hotel room on a low floor.












When staying in apartment blocks and hotels – ask for a room on floor 1 or 2. Then in the event of a fire alarm (when the lifts must not be used) you won't have to walk down many flights of stairs.

14

Daily living.

Get help in the kitchen.



 <p>Horizontal handle on potato masher, easier to press.</p>	 <p>Electric tin opener.</p>	 <p>Handle on top of kettle not side. Don't fill kettle more than needed.</p>
 <p>Gripping tool to open jars.</p>	 <p>Long handled dustpan and brush, to save crouching.</p>	 <p>Grater that stands, with robust handle.</p>
 <p>Electric food processor or 'salad shooter'</p>	 <p>Use a smaller frying pan – not a large, heavy cast iron one.</p>	 <p>Electric bottle opener, with push button operation.</p>

Try to avoid gripping, crouching and lifting in the kitchen.
Here are some ideas for tools to help.

Take in some fuel for muscles affected by fits of coughing and sneezing.

They can cause muscle cramps.[†]

.....
Reduce the risk of cramps – support the energy demands of uncontrolled muscle spasms with a sugary drink like fruit juice³.

May be best avoided if you have a condition such as diabetes.

[†]So can laughing, but it's not easy to plan for that.

Use a ladder with
wide steps, not
round rungs.



Ladders are dangerous places and we need to keep as safe as we can. Standing on narrow round rungs will put stress on your calves and the soles of your feet. Stand more comfortably on a wide flat step.

18

Daily living.



Build your self-awareness.

Become familiar with the signals which tell you to slow down or stop for a rest. Eventually you can look after yourself without even thinking about it. You can consider McArdle's just a minor irritation. (See tip 119.)

Leave bars and
nightclubs before
closing time.



.....

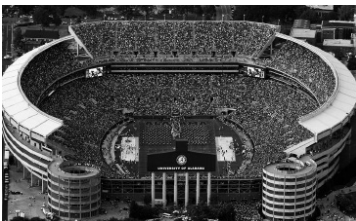
Closing time is when trouble is most likely to start. We can't run to avoid it. And the physical effort of a struggle whilst we are tense will be very damaging to our muscles.

20

Daily living.

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100

Ask for disabled access at sports stadiums, etc.



.....

Venues such as concert halls and sports stadiums have crowds, corridors and stairs – potential problems for us. Ask in advance for disabled access which may have specially located seating, allow use of lifts and enable early entry before the crowds.

House and garden.

Ideas for tools, equipment and techniques that will help you achieve what you need without damage.

21

House and garden.

Two wheels
better than one.



.....
A two-wheel barrow has wheels positioned under the load so it takes the weight of the load on the wheels. A single-wheel barrow involves lifting a large part of the weight with your arms and also needs muscle effort to maintain stability.

Weeding? Cleaning? Sit or kneel rather than squat.



.....

Squatting or crouching is a static activity and uses large muscle groups. It is very demanding of the muscles. McArde's people frequently have to go to hospital if they squat for too long.
Sitting or kneeling down is safer for us.

23

House and garden.

Use secateurs that ratchet up, reducing the effort needed.



Don't attempt to cut anything very thick. The grip needed to prune with secateurs (clippers in the US) can cause a cramp. Use a pair that ratchet up and thus reduce the effort needed. And take regular breaks. (See tip 36.)

Operating a chainsaw?

(or any other potentially
dangerous tools)

First get arms and legs
into 'second wind'.

For any potentially dangerous tool such as a power saw, get into 'second wind' before starting the task. You don't want to run out of energy or get a cramp whilst exposed to danger.

(See 'Second wind', page 48.)

25

House and garden.

— Use fitted sheets
on your bed.

— To reduce the need to
grip and lift the mattress.

When putting clean sheets on the bed, lifting the mattress on each side to tuck in the sheets can pose a hazard, especially if the mattress does not have handles. Fitted sheets go on with much less effort.

Keep a trolley
to help move
heavy items.



A lightweight folding trolley can be tucked away and brought out when you need it. (Or try a basket on wheels.) For example, unloading the shopping from the car, receiving a delivery, moving some furniture or other heavy items around the house.

27

House and garden.

Have a chair or stool
handy to avoid bending.

Like loading the
washing machine
or dishwasher.



Working when bent over is very energy demanding. Keep a chair or stool handy so that when doing these tasks you can sit down to get to the right height for the job.

Plan your house, e.g.:

Low level cupboards for heavy items.

No awkward reaches.

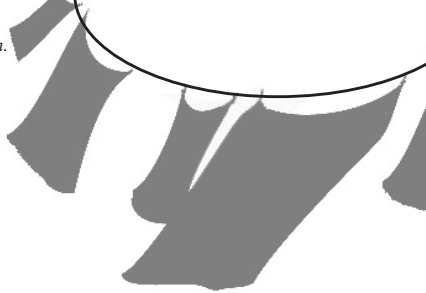
It's good to be walking back and fore.

Plan things so that you are forced to move around the house as much as possible – from desk to make tea, to toilet, to TV, etc.

And try to avoid any heavy lifting, etc.

29

House and garden.



Lower the height of your washing line.

Holding your arms up with the weight of wet washing is anaerobic, which you can only do for a few seconds at a time. A lower washing line needs less energy to reach. Hang large and small items alternately to give your muscles a break.

Swap some
household duties
with your partner.

So they do the ones that
put you at risk.

.....
Swap some regular household duties with your partner – so that they take the ones more likely to put you at risk. You keep the more gentle aerobic ones, which can do you good.

31

House and garden.

— Get your
arms into
'second wind'.

Do the ironing.



We don't have many times when we get our arms into 'second wind', ironing is one. Start slowly to get into 'second wind', then do a whole batch in one session to get the benefit. Avoid rushing to iron an item just as you need it. (See 'Second wind', page 48.)

Choose your trips up the stairs.

.....
Have a safe place at the bottom of the stairs to collect things, so you can take a few of them on your next trip up. That way you can go when you feel up to it and avoid making too many trips.

Never take stairs in a rush. (See tip 10.)

33

House and garden.

— Keep a list of tasks to organise and tackle efficiently.

|||
.....
Keep a list of upcoming household tasks so that you can organise them and tackle them as efficiently as possible – or even pass them on to someone else.

Physical activity.

The textbooks say ‘exercise intolerance’ is the main symptom of McArdle’s. But daily activities are just as problematic as planned exercise. Here are some ideas.

'Second wind'

- When starting activity if you feel your muscles run out of energy, or a cramping sensation, slow down. If the pain doesn't fade, stop for a rest. Once it has faded you can resume activity.
- After 6 to 10 minutes more blood and oxygen is flowing to the muscles, which means they can rely on fat stored in the muscle, and on fat and glucose circulating in the blood.
- Our liver releases glucose from its store of glycogen.
- 'Second wind' partly relieves inaccessibility of muscle glycogen.
- You will learn to recognise it. Heart rate is a good indicator, it will drop as you enter 'second wind'.
- It is not fully systemic. Get each muscle into 'second wind'.
- Even when in 'second wind' if you raise the exercise intensity you can get muscle pain and contracture, so pace yourself.
- Exercising in 'second wind' will train the muscles, improving their aerobic capacity – this makes 'second wind' easier to achieve and enables you to work harder.
- Ideally we should get into 'second wind' and then continue exercising for another 30 to 45 minutes, and do this most days.

There is more about 'second wind' on the AGSD-UK web site.

Learn about 'second wind' and practise getting into it.

The 'second wind' is when light to moderate activity/exercise⁴ becomes noticeably easier. It is a vital aspect of McArdle's.

Studies have shown that 'second wind' is universal to all McArdle's people, but some may need help to recognise it.

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Physical activity.

Grip. Grip. Grip.



No! No! No!

We cannot grip at maximum intensity for more than 6 seconds. So be wary of all of these things. If the jam jar is not open after 6 seconds leave it and come back later. Swap hands regularly whilst filling with fuel. (See 'Six second rule', page 12.)



Learn this tip and use it. It will serve you well in almost all that you do.

Physical activity.

36

Avoid pain and cramps by following
THE FOUR 'R's OF McARDLE'S

- Recognise
- Respond
- Recover
- Resume

.....
Recognise – that your muscles are starting to tighten up.

Respond – slow down, or pause for a rest.

Recover – if your recovery takes more than a minute, you overdid it.

Resume – because aerobic exercise is vitally important to us.

37

Physical activity.

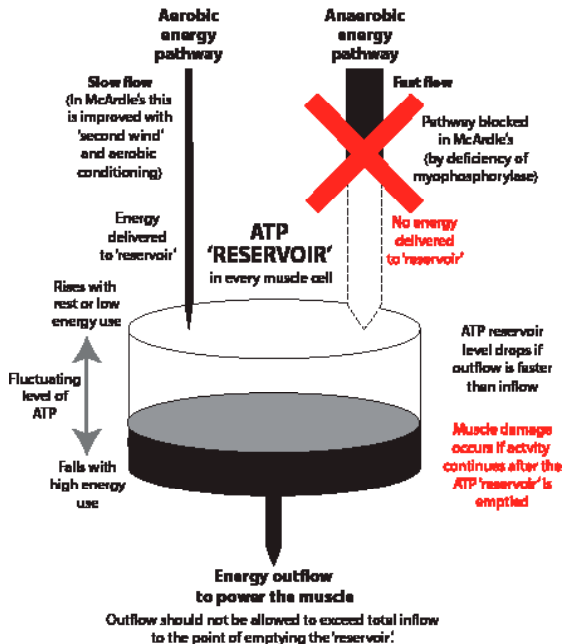


Note increased heart rate as a signal to slow down.

Some people find heart rate a more useful guide than the sensations in their muscles. You need to keep your muscles active, but if your heart rate zooms up⁵, slow down or take a break.

Take care not to
let your 'second wind'
lapse due to inactivity.

.....
Once you have got some muscles into 'second wind', if you stop
it will fade off and be gone completely in about 30 minutes.
So be wary of stopping for long. (See 'Second wind', page 48.)



Your ATP ‘reservoir’

Adapting activity

McArdle’s people have to pace their activity so that their energy usage does not exceed the rate at which the aerobic pathway delivers.

Energy store in the muscle cell

ATP (Adenosine Triphosphate) is used to provide energy for muscle contractions. A small store in each muscle cell is topped up by the fast anaerobic and slow aerobic pathways. In McArdle’s only the slower aerobic pathway works.

Visualise the ATP energy store as a ‘reservoir’.

Picture the anaerobic and aerobic energy pathways as supply pipes bringing energy into the reservoir. The anaerobic pipe is large and can quickly respond to provide a fast flow of energy. In McArdle’s that pipe is blocked. The aerobic pipe is smaller and slower, but still works.

Don’t empty the ‘reservoir’

When we exercise we drain energy out of the ‘reservoir’. So as not to run out of energy we have to ensure that the outflow demand is in balance with, or less than, the inflow. If the outflow is greater than the supply the energy will soon run out. This is to be avoided because when muscles run out of ATP they start to break down.

There is more about adapting activity on the AGSD-UK web site.

39

Physical activity.



Rest before risk.



.....
When you are about to do something which might involve risk, like crossing the road, rest for a minute before proceeding. This is to ensure that you have energy available to get you through.

Physical activity.

40



for 80
% of your ATP

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

Pain in McArdle's lags behind the damage you do. Learn to recognise the early signs and slow or rest before you do damage. 30 seconds rest will replenish 80% of the energy reservoir in your muscles. Then you have some leeway.

41

Physical activity.



Learn how to avoid 'bad days.'

McArdle's people report days
when it is difficult to get going.



Many factors affect this – what you have eaten, stress, the temperature, and most importantly moving too quickly from inactive to active. The longer you have been inactive the more care is needed to get into 'second wind'. (See more on web site.)

Swim only in
water you can
stand up in.

And be wary of waves!



Your muscles can run out of energy so you need always to be able to stand up if that happens. In the sea, waves can knock you over and make it very hard to get up again.

Swim with a friend!

43

Physical activity.



Seek physiotherapy to help you improve any weak muscles.



Some muscle wasting is a known complication of McArdle's, especially in the upper body. If you have some weak muscles, ask for help to work on strengthening them. It is essential that this is from a physiotherapist who knows about McArdle's.

Exercise: good, bad and ugly.

Regular exercise in ‘second wind’ improves our aerobic capacity, so that we can do more without pain and without the risk of damaging our muscles.

Reasons to exercise

Keeping physically active is not *just* good for your McArdle's.

Being more, and regularly, physically active is proven to be of benefit to everyone. In particular it benefits:

- your mental health and wellbeing
- your heart and circulation
- your blood and metabolism
- your muscle endurance and flexibility
- your balance and co-ordination
- your bone mass and strength

We can all suffer from other health conditions, especially as we get older. People with McArdle's often have health issues such as high cholesterol, diabetes or obesity. These conditions all gain benefit from regular exercise.

Currently the only therapy for McArdle's is to improve your fitness and keep your weight down. So keep physically active and take regular exercise.

Make exercise
sociable and
enjoyable.

Then it is much easier
to keep it up.



.....
Arrange to walk with a friend who also needs exercise and explain McArdle's to them. Have regular routes that you enjoy and won't worry about what is around the corner. For the treadmill, fix a time slot when you want to listen to the radio or watch TV.

45

Exercise: good, bad and ugly.



Ignore the saying
“No pain, no gain”
IT’S WRONG!



For McArdle’s people, pain on exercise is the signal that we are overdoing it and need to slow down or take a break.

Next time someone in a gym quotes “no pain, no gain” or “no gain without pain”, ignore them, it’s wrong for us.

Accept the saying
“Use it, or lose it.”
IT’S RIGHT!

If we lose all aerobic capacity in a muscle, exercise becomes very difficult and damaging. We must keep active as it is hard to recover from a very low aerobic capacity. Although it is never too late⁵.

47

Exercise: good, bad and ugly.

Keep a dog.

It will make sure
you get out
for a walk.



A dog will rely on you to take it out for a walk twice a day. Perfect for McArdle's. A well trained dog will be happy to explore each time you stop for a rest, and won't pull you on faster than you want to go.

Get help with planning an exercise programme.

.....


The more unfit you are the more necessary it is to have a personalised programme to help you get started. In the UK this is provided by the McArdle's Clinic. But if you don't have such support, there is some guidance on the AGSD-UK web site.

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Exercise: good, bad and ugly.



Make exercise
more meaningful.



Set yourself a
long term objective.

An objective to gradually work towards over the weeks will give your exercise more meaning. Your progress will encourage you as you build up to your objective such as a duration, a 12-minute distance, or a start speed.

Exercise: good, bad and ugly.

50

Choose the right
treadmill for
exercising at home.



Walking is something that we all use every day, so that is the best exercise to adopt. Choose the best treadmill you can so that you feel secure and it provides the programmes you need.

See the AGSD-UK web site for more information.

51

Exercise: good, bad and ugly.

Try an electrically-assisted bicycle.



Don't get
left
behind!

Electrically-assisted bicycles have improved enormously. One will enable you to cycle with friends without fear of hills or getting left behind. Cycle on your own and you have no worries about getting home.

Take a break from exercise when unwell (or tense).


When the body is under stress from illness, or being tense, exercise can be more risky for us. Take a break from your exercise routine. (See tip 12.)

53

Exercise: good, bad and ugly.



Extend your exercise to make life better.



As we exercise in ‘second wind’ we build more mitochondria in our muscles, helping us utilise our fat stores^{4,6}. But it’s not just about easing our exercise – aerobic fitness makes daily activities much easier for us. (See ‘Second wind’, page 48.)

Choose a sport that competes on skill.



.....

When choosing a sport look for one where you compete on skill rather than physical prowess. Golf is a great example, with lots of walking to keep you aerobically fit. But use a trolley for your clubs, not a shoulder bag.

55

Exercise: good, bad and ugly.

Keep a log of your performance.



	Date	Description or Workout Ref.	Duration (minutes)	Distance (km)	12 minute distance (km)	Comments
M						
T						
W						

 It helps you stay motivated for exercise.

.....
Keeping a simple log will help to keep you focussed, and looking back at your progress will be a great motivator. You can download a log sheet from the AGSD-UK web site.

Walking with McArdle's.

Techniques can be utilised to enable varied and challenging walking to be enjoyed, and our fitness improved.

56

Walking with McArdle's.

Plan your route
to start
on the level.



.....

Start your walk on flat ground (and preferably a firm surface) to make it easier to get into 'second wind'. 1 mile (1.6 km) should be enough. The more aerobically fit you are the less you will need to adopt this approach. (See 'Second wind', page 48.)


Remember –
'slow and steady
all the way'.

.....
At the outset slow and steady will help you get into 'second wind' safely. No rushing, just slow and steady. Come to a slope and you may want to slow further. (See 'Second wind', page 48.)


58

Walking with McArdle's.

Rest whenever you need to.



Don't set artificial targets
such as a number of paces.



.....

We must obey the feedback from our muscles and rest when they tell us. We must not set artificial targets like saying “I’ll do another 50 paces” – that can mislead us into doing too much and potentially getting a fixed contracture.



Learn to use walking poles.

.....
Lightweight, aluminium, telescopic walking poles are great for us. They spread the load onto arms as well as legs, and give your upper body a chance to exercise in 'second wind'. See walking hints on web site and video on AGSD-UK YouTube channel.

60

Walking with McArdle's.

Face sideways when standing on a steep slope.



If taking a rest on a steep slope, don't stand with your feet pointing up hill, as your calf muscles will be stretched and at risk of a contracture. Instead, stand sideways on the slope with one foot higher than the other. Swap legs now and again.

Adjust your pace for different surfaces.

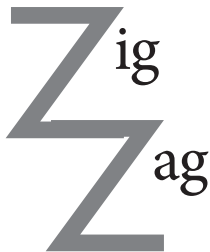


Mud, long grass and loose gravel – McArdle's walking courses, Wales 2011 and 2012

.....
Long grass, loose gravel, soft ground, sand, mud – all these and other surfaces can increase the effort needed. Some can make you tense your muscles as you protect yourself from slipping or falling. Slow your pace or take more frequent rests.

62

Walking with McArdle's.



.....

On a steep slope reduce the effective incline (and thus reduce the effort) by zig-zagging across the width of the path. On a hillside, provided the terrain is safe, follow a continuous gentle angle across the slope instead of a direct ascent.

Take lots of short rests
on the way up and
a short lunch break
before the top.

Other people press on continuously up a hill, then have a long break on top. We need lots of very short rests on the way up. A short lunch break avoids losing 'second wind'. Setting off again we refresh the 'second wind' in our uphill muscles. (See tip 38.)

64

Walking with McArdle's.

Look for small steps up.

They avoid lifting your weight
too much in one step.



Rocky paths are designed for people who don't have McArdle's. Each step is often quite high. Instead, look for intermediate steps, or even a point beside the path, to reduce each lift of your body weight by comparison to the intended step.

Learn from walking with other McArdle's people.



McArdle's walking course, Wales 2011

Most of us never have a chance to meet anyone else with McArdle's. Walking with others who have the condition and understand what you go through is a revelation.

We can all learn from each other.

66

Walking with McArdle's.

Choose
lightweight
walking
boots.



In preparation for country walks requiring boots, make sure you choose a lightweight brand – it makes a big difference. Choose boots that have soles with good grip, so that you feel safe on poor surfaces and don't tense up.

Sit or kneel to put on boots.

It reduces the risk of a cramp in
muscles which are not warmed up.

Before a walk your muscles won't be in 'second wind', so sit or kneel to put on boots rather than stretching your calf by putting your foot up. Take a break between tightening laces on each boot or else your arms or fingers may cramp.

68

Walking with McArdle's.

Slow your pace at a change of slope.



When you come to a hill or slope when walking, slow down, because you will start using some different muscles which will not be in 'second wind'. Slowing your pace also protects against cramping due to the increased effort.

Choose a
good
backpack.



With camera, lunch, flask, first aid kit, map & compass, waterproofs and spare clothes, a backpack soon becomes quite heavy. A fit close to the shoulders and a waistband will make it easier to carry and improve stability.

70

Walking with McArdle's.

Set the pace yourself when walking with non-McArdle's people.



When going for a walk with non-McArdle's people, ask them to let you set the pace. See AGSD-UK web site for further guidance for walking partners.

Personal care.

Simple things like drying after a shower can be a challenge.

Here are tips to help.

And some guidance on when to seek medical help after an episode.

Carry the card.



.....

This four page folding card provides a useful quick explanation of McArdle's and explains when you may need assistance. If you need to go to hospital, show the card when you arrive. Slip one in your pocket or handbag. Available from your consultant.

Guidance on emergencies

Inside the card is a reminder about when to seek medical attention.

If after strenuous or unusual exercise/activity

you have one or more of the following signs. (A few people have experienced these symptoms without exercise.)

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 exercise - perhaps with 'flu-like symptoms, can be a sign of rhabdomyolysis (muscle damage).

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

You should

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

72

Personal care.

Use an electric
toothbrush.

And anything else
electric you can think of.



W

.....
Brushing your teeth requires an intense repetitive action which rapidly exhausts your energy supply, so teeth are not brushed as well as they should be. Electric toothbrush = problem solved.

Think electric screwdriver, tin opener, food mixer...

Take your time having a shower and drying yourself.

Having your arms raised to wash and dry yourself demands a lot of energy. Rush, and your arms may cramp. Don't get too hot as it is harder to get dry. Use a really good towel – maybe two small ones rather than a large, heavy one. (See tip 1.)

If you have long hair, try a wall-mounted hair dryer.



W

One arm holding the dryer and one the brush can be very tiring. This type takes the weight on the wall. Give each arm a break by using the brush alternately in each hand. Or rest your arm on furniture so the weight of the dryer is partly supported.

Never take pain medications before or during exercise.

We need signals from our muscles to tell us when to take a break.

.....
Pain medications interfere with the feedback, so should only be taken *after* exercise. Avoid opioid-based pain meds completely as they cause long term problems. Don't take ibuprofen after muscle damage as it is metabolised in the overloaded kidneys.

76

Personal care.

Be careful with medications – read the data sheet.



Some medications have side effects involving muscle symptoms and are best avoided. Statins (prescribed for high cholesterol) may pose a risk for some McArdle's people⁷ – only try with caution and under close medical supervision with CK testing.



At the dentist's – ask for a 'mouth prop'.

It holds your mouth
open without effort.



If you are in for a long session, this simple plastic wedge will prop your mouth open and eliminate the risk of putting your jaw muscles into a cramp. If you need extensive work, you might ask the dentist to spread it over two visits.

78

Personal care.

When you
get angry:
breathe, and...



**Count
to 10**



Anger makes our muscles tense, and that makes them work anaerobically, which is damaging for people with McArdle's.

So when you start getting angry, breathe deeply and count to 10 to calm down. (See tip 12.)

Use gentle massage only, not deep massage.

On a hard, fixed contracture it is best to avoid massage altogether.

A deep massage can be damaging for our muscles, so use gentle massage only. A muscle which is in a hard, fixed contracture is best not massaged at all, but just left to rest.

80

Personal care.

Limit full-on stretches to six seconds.

Any longer is not recommended for
people with McArdle's.



If you see a physiotherapist for some other problem and are advised to hold a full-on stretch for 30 seconds, instead limit it to six seconds (see 'Six second rule', page 12). Also, stretching to eliminate lactic acid is not needed as we don't make it.

Friends, family and others.

Educating your friends and family about the condition will enable them to help you look after yourself.

81

Family, friends and others.

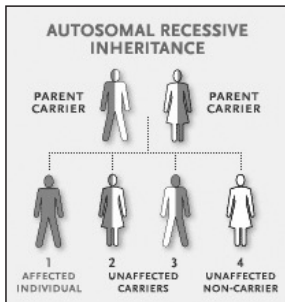
Be open about your
condition, not secretive.

Think of McArdle's more like a
characteristic than a disease.



As a genetic condition it's just a part of who you are, like having ginger hair or blue eyes. It's not infectious, there is no need to hide it. Use the McArdle's Information Card (tip 71), or this book, to help explain this part of you. (See tip 119.)

Consider whether other family members are affected.



.....

It is 3 to 1 against a child of two carriers being affected (see above). For a child of a McArdle person and a carrier it is 2 to 2. But only about 1 in 160 people is a carrier, so the risk is 320 to 1. If symptoms appear in a child, get them checked.

83

Family, friends and others.

Educate your family and friends about McArdle's.

Then they can understand and
help you stay safe.



People are usually very happy to have an understanding of your needs. They will look out for you. This book will help explain what living with McArdle's is like.

Eating and diet.

There is no magic solution, but there are some simple rules. Also, a few things are worth trying to see what works best for you.

84

Eating and diet.

Have a healthy balanced diet.

Include low-glycaemic index
carbohydrates such as
vegetables and wholemeal pasta.



.....
You could try changing the balance to more carbohydrates and less protein, and then try vice versa. See which suits you best.

But avoid excess calories or your weight will increase.

In the UK we can get advice from the McArdle's Clinic dietician.



Keep hydrated
during activity
and exercise.

Muscles contain about 75% water and even a small degree of dehydration can worsen cramps. Symptoms of dehydration include fatigue, moodiness, a 'drained' feeling and thirst.

86

Eating and diet.

Eat before effort (especially breakfast).



Because we can't access the glycogen stored in our muscles we are very dependent on blood-borne glucose produced by the digestion of food. Always eat breakfast to help you get going without cramps and nausea.



Watch your weight.

Excess weight is the
enemy of McArdle's.

Every small amount over our healthy weight puts an extra load on our muscles, which makes cramping more likely. (There is a link on the AGSD-UK web site to the NHS calculator of the healthy weight for your height and age.)

88

Eating and diet.

Try drinking
some coffee.

Caffeine *may* help
aerobic metabolism.



Some research has suggested that caffeine may boost the effectiveness of aerobic metabolism. However, this is controversial. You could try a coffee in the mornings and see if it works for you.

Driving, travel and transport.

We have some tips on driving.
Public transport can pose
unexpected hazards, but there are
ways to be prepared.

89

Driving, travel and transport.

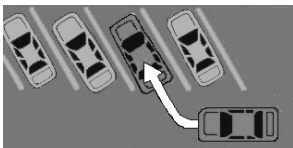
Choose a car with power steering.

And maybe an automatic
rather than a manual gearbox.



The bigger the car, the more essential the power steering. It greatly reduces the strain on your arms when manoeuvring. An automatic rather than manual (stick-shift) gearbox reduces the risk of leg cramps. Never 'ride the clutch' on a manual.

Whenever possible find an angled parking space.



This is the parking with minimum steering wheel movement. The worst is parallel parking on the street into a small space, with cars waiting behind you, when your shoulder and arm muscles can become severely cramped. (See tip 91.)

91

Driving, travel and transport.

Use your
mirrors
when
reversing
your car.



Avoid turning your head to look over your shoulder. That is an awkward position which is static and will result in a muscle cramp within seconds. So use your mirrors. Practice makes perfect. Proximity sensors at the rear also help. (See tip 90.)

When using public transport, plan your route in advance.

Some transport operators will offer advice about routes which avoid stairs or long walks to transfer between platforms, etc. In London www.tfl.gov.uk has an on-line journey planner that can take disability into account, such as step-free routes.

93

Driving, travel and transport.

Don't run for the bus –
there is always another.



Running for a bus (or train) is sprinting. We can only do 6 seconds and after that muscle cramps will come on. Don't be tempted. It is more important to stay out of hospital than to be on time. (See 'Six second rule', page 12.)

On the underground, if an escalator is broken...

don't take the stairs, get back on the train, travel on a stop and walk back.

Long flights of stairs are a major hazard and should only be attempted very slowly and with plenty of rests on the way. Never start on stairs with people behind as they pressure you to keep going. Instead a walk back will be good aerobic exercise.

95

Driving, travel and transport.

Allow plenty of time
at airports.

Or ask ahead for a
wheelchair or buggy.



Walks to flight gates and for inter-connections can be long and rushed. There is usually no problem in getting some assistance if you ask in advance. Be ready to show your McArdle's Information Card (see tip 71).

Reduce the load...
use a suitcase
with four wheels.



Two wheels are good, but four wheels really make a big difference. The weight is entirely taken on the wheels, and it is easy to manoeuvre through awkward places. The wheels need to be ones which freely swivel.

97

Driving, travel and transport.

Be wary of
walking
in crowds.

Try to avoid
rush hour travel.



You need to walk at your own pace, not that of the crowd, and to rest when your muscles tell you to. Being swept along by the crowd and then walking up an incline or steps can bring a high risk of cramping.

Medical.

A confirmed diagnosis,
personalised advice from an expert
and regular monitoring can all help.
But you must take responsibility
for yourself.

98

Medical.



Exercise assessment

Get checked
and advised
by experts
in McArdle's.



Due to the rarity of the disease (c.1 in 100,000 people) there used to be a high rate of misdiagnosis. Get checked by experts and obtain a personal assessment and advice. This is likely to include confirmation of the diagnosis through genetic testing.

Attend for regular
assessment and
keep in touch with
developments.

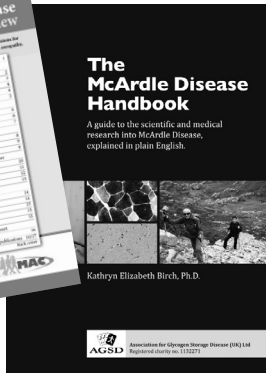
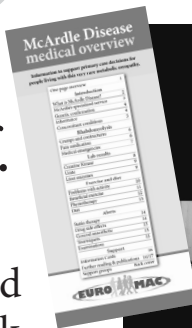
Our symptoms can improve or get worse, especially those linked to aerobic conditioning. As medical science advances, recommendations can change. So keep in touch with your McArdle expert for regular assessment and updates.

100

Medical.

Inform
yourself.

Refer to the
Overview and
the Handbook.



The overview booklet is ideal for primary care doctors. The Handbook covers in plain English all the medical and scientific research. It has references to the key scientific papers. 208 pages, large paperback. Order via the AGSD-UK web site.

Medical.

101



Brief your
general practitioner
about McArdle's.


Don't expect too much. Family doctors will probably never have another McArdle's patient, so cannot spend a lot of time learning about it. There is a medical overview booklet by Euromac, available through your consultant or the ASGD-UK.

102

Medical.

Be clear and express
your concerns.

The professionals can't understand
McArdle's as well as you do.

 We understand what it is like living with McArdle's.
It is hard for the professionals to get that understanding
because they can never experience it first hand.
We have to be our own advocate.

Medical.

103



Build an understanding of your CK[†] levels.

†Creatine Kinase also known as Creatine Phosphokinase or CPK.

CK levels rise after muscle injury. Keep a note of any results to establish your average level. A blood test for CK after an injury will then indicate how much damage you have done. Improve aerobic fitness so that the base level comes down. (See tip 107.)

104 *Medical.*

Limit any time with a tourniquet.



Tourniquets cut off the blood supply to the muscle and will result in injury if left on. This is particularly damaging for people with McArdle's. A short time for a blood sample to be taken or blood pressure to be measured should be OK.



Keep a clear plastic jar
ready by the toilet.

Urinate in it if you feel unwell
after exercise or just think you
have overdone it.

This will enable you to monitor the colour of your urine
without it being diluted in the water of the toilet. Reddish tea
to cola colour indicates considerable muscle damage.
(See 'Guidance on emergencies,' page 93.)

106

Medical.

Resist the temptation to uncurl your fingers on a 'clawed' hand.



This will not speed its recovery and risks tearing the muscle, which will greatly prolong the recovery time. The same applies to any other muscle which is in a fixed contracture.



Relax about
moderately raised levels
of CK, ALT or AST on
routine blood tests.

All these enzymes can be raised because of McArdle's⁸.
ALT and AST are known as liver enzymes but they are also
released by damaged muscle, so a moderately raised level is
unlikely to require further investigation. (See tip 103.)

108

Medical.

Watch out for a high plasma uric acid.

It can lead to gout and/or
kidney stones.

McArdle's people are more prone than the general population to raised uric acid levels in their blood⁹. This can cause attacks of gout, which are very painful. It can also lead to the formation of kidney stones. Alcohol inhibits the clearance of uric acid.



Medical.

109



Keep some gel packs
in your fridge or freezer,
ready to ease a contracture.

You can chill or freeze these medical gel packs and put them on a cramped muscle to help reduce the swelling. Some people microwave the gel pack to use as a hot compress to increase circulation. Both approaches will speed recovery.

110

Medical.



Warn your anaesthetist of the risk of Malignant Hyperthermia.

McArdle people may be at extra risk of a reaction to general anaesthetic similar to Malignant Hyperthermia, whereby your temperature increases out of control¹⁰. Although the risk is very small it is worth mentioning as the condition is serious.



School, college and work.

These make up a very large part of our lives. Making people aware of our limits enables them to make reasonable adjustments.



School, college and work.

Tell your school, college or workplace about your needs.

In many countries, disability rights legislation requires employers and others to make reasonable adjustments to accommodate your needs, so that you do not suffer unfair discrimination. At school, seek permission to use the lift.



Ask your school
for a second set
of textbooks.

And extra time to
transfer between classes.



Try to avoid carrying heavy textbooks to and from school.
A few minutes extra to walk between classes means stairs can
be taken slowly. If there are a lot of stairs, can you get
permission to use the lift?

Exercise in school?

Physical activity is very important for children for both their emotional and physical wellbeing. Physical activity prevents excessive weight gain, conditions muscles for aerobic exercise and lays the foundations for a healthy lifestyle in adulthood.

In early years, PE is not competitive and focuses on developing balance and movement and thus may not present many problems.

In teenage years, sporting activities are competitive and physically demanding. It is important that the child can recognise his/her limitations and must be aware of the importance of McArdle-specific warm up before sport. The child must be allowed to rest as and when required.

Teachers must be well informed of the condition and trained to monitor the child's condition very carefully during activity. It is essential that everyone involved understands the potential risk of rhabdomyolysis.

It is advisable to deal with the school head to ensure that all teachers are aware of the child's condition and monitor closely to ensure that the child is not bullied as a result.

Some children with McArdle's cope well with physical activity at school, but many do not. There are no hard and fast rules, each child is different. Opting out of PE altogether may be preferable.



Discuss PE and sports with school.

McArdle children need to maintain exercise.



If included in PE and sports, the child must be allowed to withdraw whenever they feel the need. As a child gets older it may be safer to exempt them from PE and sports, and instead exercise outside school in a safe environment.

When a child reaches 18 authorise parental notification.

At 18 children legally become adults in the UK. Confidentiality issues arise around providing information to parents. The young person should provide the college/employer with an authorisation to notify a parent in the event of a McArdle's related episode.





Use a
briefcase
on wheels
or a
backpack.



Heavy briefcase? It is much easier to take the load on wheels.
Or use a backpack to carry the load centrally rather than on
an arm and hand.

116

School, college and work.

Say:
“I’ll meet you there”.



When going to a meeting with colleagues, find an excuse to meet them there rather than walk together and risk problems such as walking too fast or going uphill. Plan to be early, so there can be no need to rush.



Choose a career
which does not involve
heavy manual work.

Or any activity which puts you at risk
of cramps, unless you can control it.

.....
It's best to start channelling your interests from an early age.
Look for jobs that won't involve heavy manual work and which
won't put you in danger – like standing on ladders.

118

School, college and work.

Do any physical work
at your own pace.

Have a change or take a break whenever you need to.
Employers have a legal obligation to make reasonable
adjustments to enable you to do your job.



Feeling good.

We need to stay positive to deal with what is a lifelong condition. Acceptance of McArdle's as a part of our make up, and concentrating on what we can do, will help us to feel better.

119

Feeling good.



These people had severe episodes (CK up to 300k) and subsequently climbed 3,000 ft (900 m) mountains.

Accept McArdle's
as part of who you are.

Work with it, don't fight it.

.....
McArdle's can make us dogged and determined. We are likely to be steady and reliable. McArdle's is a part of us. Would we be who we are if we didn't have McArdle's? (See tip 81.)



Feeling good.

120

Concentrate on what
you can do.

Forget about what you can't do.

People with McArdle's can achieve remarkable things and excel in all walks of life⁶.

121

Feeling good.

Lift your mood
by taking
regular exercise.



Exercise produces endorphins which lift our mood.
As our aerobic capacity increases, our symptoms decrease.
The result is less pain and getting through life more easily.
(See 'Reasons to exercise', page 62.)



Feeling good.

122

Get over your embarrassment.

Let them wonder!

.....
Strangers are watching and wondering what's up with us.
What does it matter? Enjoy the moment – keep them guessing,
create a bit of mystery and intrigue. Learn to love it. (See tip 8.)

123

Feeling good.

Look around.
You will see people who
are much worse off.

Managed correctly, McArdle's is not life threatening or life limiting. For the vast majority of us it is not seriously disabling. Many other diseases, including muscular dystrophies, are very much worse. We are really lucky.



Feeling good.

124



Set yourself goals
and strive for them.

Put McArdle's to the back of your mind. Set some goals in life which you want to achieve and focus on them. Consider McArdle's to be only a minor inconvenience.

125

Feeling good.

Share your experiences
with other
McArdle's people.



McArdle's workshop, Denver 2009

It is very affirming to share experiences with people who understand what you go through. Make contact on one of the on-line support groups (including Facebook), or through your country's support group co-ordinator.

The McArdle Mantra

This set of memorable phrases makes it easier for us to remember the things that are important to looking after ourselves. All the tips in this book are encompassed by one or more of these phrases.

- **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**

For anyone who would like to know more, there are some explanatory notes for each phrase on the AGSD-UK web site.

Note your own tips here

A large, empty rectangular box with a thin black border, intended for users to write their own tips. The box is currently blank.

When you are ready email your tips to type5@agsd.org.uk and one day you may see them in 'Another 101 tips....'!

McArdle's glossary

Acute renal failure

Sudden decline in kidney function. Requires urgent medical attention. In McArdle's it is caused by myoglobin blocking the kidneys.

Aerobic exercise

Exercise which requires oxygen in converting fuel sources into energy. Walking is a good example. This is good exercise for McArdle's.

Anaerobic exercise

Exercise which does not need oxygen to utilise fuel sources. Gripping is a good example. The energy for this type of exercise is produced using glycogen, which those with McArdle disease cannot do. Anaerobic exercise must be avoided as it is damaging to our muscles.

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's are inherited.

Carbohydrate

A type of compound, such as starches and sugars, found in food. Broken down in the body to form energy. Low glycaemic index carbohydrates release energy slowly.

Creatine kinase (CK or CPK)

An enzyme which is used in the formation of ATP. People with McArdle disease tend to have a raised level of CK in their blood. When muscle is damaged CK leaks out of muscle cells into the bloodstream. After excessive anaerobic activity CK can rise to hundreds of times normal.

Enzyme

A protein which the body uses to make a chemical reaction. Myophosphorylase, which is missing in McArdle's 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.

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 in the muscles and in the liver. It is converted back to glucose to be used 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. Examples: clenching fists, holding something up or pushing. The worst type of activity for McArdle's people.

Lactic acid

A by-product of muscle using carbohydrate. Exercise leads to a rise of lactic acid in the blood, but in McArdle disease it does not rise.

Malignant hyperthermia (MH)

A severe form of fever caused by a reaction to certain anaesthetics and muscle relaxants. Those with McArdle's are at an increased risk and should tell their anaesthetist.

Metabolism

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

Mitochondria

A small organ within muscle cells, responsible for energy production from fuels. Regular aerobic exercise increases the number of mitochondria, which boosts the aerobic capacity.

Myalgia

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

Myoglobin

A protein found in red skeletal muscle.

Myoglobinuria

Presence of myoglobin in the urine, resulting in a reddish tea to cola colour. Muscle damage releases myoglobin into the blood and the kidneys remove it to the urine. See 'acute renal failure'.

Myopathy

Disease of the muscle. McArdle's is a myopathy.

Myophosphorylase

The muscle type of phosphorylase. McArdle's people are deficient in this

enzyme. There is also a brain type and a liver type.

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.

Protein

Complex organic compounds found in the body and in foods such as meat. Consist mainly of amino acids. Serve as functions including enzymes, in oxygen transport and muscle contraction.

Rhabdomyolysis

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

References

- 1 McArdle, B. (1951) Myopathy due to a defect in muscle glycogen breakdown. *Clin. Sci.* 10: 13-33.
- 2 Saltin, B. (1973) Metabolic fundamentals in exercise. *Med & Sci in Sports*, v5, n3, 137-146.
- 3 Andersen, S.T., Haller, R.G. and Vissing, J. (2008) Effect of oral sucrose shortly before exercise on work capacity in McArdle disease. *Arch Neurol* 65: 786-789.
- 4 Haller, R.G., Wyrick P, Taivassalo P, et al. (2006) Aerobic conditioning: an effective therapy in McArdle's disease. *Ann Neurol* 59:922e8.
- 5 Perez, M. et al. (2006) Exercise capacity in a 78 year old patient with McArdle's disease: it is never too late to start exercising. *Br J Sports Med* 40: 725-726.
- 6 Lucia, A. et al. (2012) The 'McArdle paradox': exercise is good advice for the exercise intolerant. *Br J Sports Med* doi: 10.1136.
- 7 Vladutiu, G.D. et al. (2006) Genetic risk factors associated with lipid-lowering drug induced myopathies. *Musc Nv* 34: 153-162.
- 8 Tuzun, A., Erdil, A., Bagci, S., Bolu, E., Onguru, O., Kurt, I., Yesilova, Z., and Dagalp, K. (2002) McArdle's disease: case report and review of the literature. *Turk J Gastroenterol* 13: 56-59.
- 9 Puig, J.G., de Miguel, E., Mateos, F.A., Miranda, E., Romera, N.M., Espinosa, A., and Gijon, J. (1992) McArdle's disease and gout. *Muscle Nerve* 15: 822-828.
- 10 Bollig, G., Mohr, S., and Raeder, J. (2005) McArdle's disease and anaesthesia: case reports. Review of potential problems and association with malignant hyperthermia. *Acta Anaesthesiol Scand* 49: 1077-1083.

Other papers of particular interest

Quinlivan, R. et al. (2009) McArdle disease: a clinical review. *J Neurol Neurosurg Psychiatry*. doi: 10.1136/jnnp.2009.195040

Quinlivan, R. and Vissing, J. (2007) 144th ENMC International Workshop: Outcome Measures in McArdle Disease, 29 September-1 October 2006, Naarden, The Netherlands. *Neuromuscul Disord* 17: 494-498.

Andersen, S. and Vissing, J. (2008) Carbohydrate- and protein-rich diets in McArdle disease: Effects on exercise capacity. *J. Neurol.*

Neurosurg. Psychiatry doi:10.1136/jnnp.2008.146548

Lucia, A. et al. (2008) McArdle disease: what do neurologists need to know? *Nat Clin Pract Neurol* 4: 568-577.

Cochrane Reviews

Quinlivan, R., Martinuzzi, A., Schoser, B., (2014) Pharmacological and nutritional treatment for McArdle disease

Quinlivan, R., Vissing, J., Hilton-Jones, D., Buckley, J. (2012) Physical training for McArdle disease

Our slow and steady tortoise was adopted as the logo of the 'Walk over Wales' in 2010. A group of McArdle's people met the challenge of walking 210 miles (338 km) in 32 days from north to south across Wales, and over many of its mountains.

The group raised awareness of McArdle's and through their blog inspired many McArdle's people around the world.

"Slow and steady all the way" was their motto, and the tortoise epitomised that.



stephanievondresder.blogspot.co.uk

Further information



Euromac Registry

Euromac is a registry of patients affected by McArdle Disease and by other forms of rare neuromuscular glycogenosis where exercise intolerance is the main symptom. Key aims are: early diagnosis; high quality advice and management, and eventually an effective treatment or even a cure.
www.euromacregistry.eu

The McArdle Disease Handbook

The Handbook, written by Kathryn Birch Ph.D. and published by AGSD-UK, brings together all the medical and scientific research on McArdle's. It explains in layman's terms the cause of the disease, method of inheritance, history, and current and future treatments.

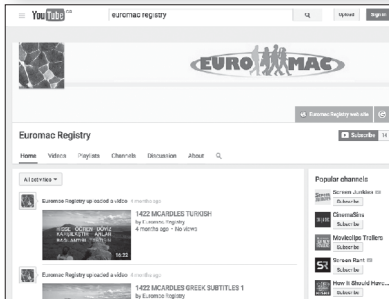
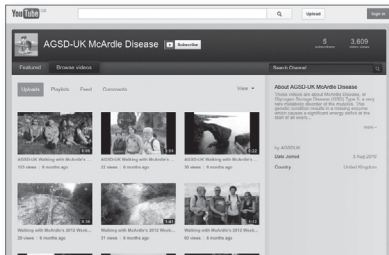
Support group and web site

The AGSD-UK is a support group for people affected by the various Glycogen Storage Diseases, including McArdle's. Although a UK group the information it makes available on its web site, its publications, conferences and activities such as walking courses, are available to all.
www.agsd.org.uk

YouTube channels

Euromac and AGSD-UK each have YouTube channels with over 30 videos on various aspects of McArdle's.

Euromac's videos are available subtitled in 8 languages.



About the author



Andrew Wakelin experienced symptoms from age four. Finally diagnosed in 1980 at age 30, he had no help until age 50. In 1999 the diagnosis was genetically confirmed (two copies of the common R50X mutation).

Andrew credits walking and cycling in childhood for being less badly affected than many. He found he could do a lot by going at his own pace and resting when he felt his muscles tightening up. Using techniques he has developed, he has climbed all 188 Welsh mountains over 2,000ft (600m) and Mount Kilimanjaro at 19,340ft (5,894m).

As AGSD-UK McARDle's co-ordinator Andrew has met around 300 McARDle's people and spoken at conferences in the USA, Germany and Italy. He works with the UK McARDle's Clinic to develop their services, research and clinical trials; liaises with McARDle consultants around the world and has contributed to published papers. He maintains the McARDle's information on the AGSD-UK web site.

In 2010 he led four McARDle's people on the 210 mile (338 km) 'Walk over Wales', raising awareness and funds.

Andrew helps people to appreciate how much they can do through building aerobic capacity and using the right techniques. He has devised a residential course to help people develop their walking safely and enjoyably. To date people have attended from 14 countries.