

## A quick overview

- McArdle's is an ultra rare inherited condition which restricts energy for skeletal muscles.
- People with McArdle's cannot utilize the energy stored in the muscles.
- They have a serious shortage of energy early in any activity, and throughout all *intense* activity.
- They need to avoid being in a rush and will be particularly slow and may stop/start at the beginning of activity.
- They can develop pain and cramping. If activity continues in pain, muscles become stiff, swollen and very painful.
- There can occasionally be very serious complications of such 'contractures'.
- They have a 'second wind' which somewhat reduces their symptoms after about 10 careful minutes.
- They need encouragement to maintain an active lifestyle.
- Regular activity will help to keep their muscles functioning and healthy.
- They achieve a lot by utilizing practical and management techniques to protect themselves.
- Some simple adjustments at home will help enhance their safety and their on-going improvement.

*Front cover photographs:* McArdle's people come in all shapes and sizes, from all over the world. They often appear well, but have to cope with their unseen condition. The people shown here range in age from 5 to 68 and are from 7 countries. The question mark represents the many people who remain undiagnosed.

# At home with McArdle's

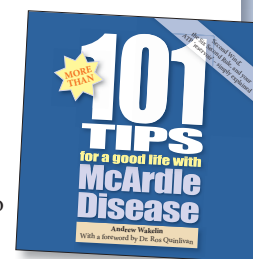
## Friends and family information

A plain language guide to McArdle Disease to help family and friends to understand and support the affected person.

## Further understanding

The person with McArdle's can share this book with you, or you can request a copy from IamGSD.

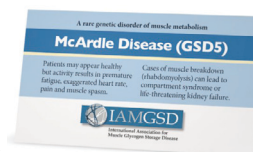
It is an easy to read guide with explanations of 'second wind', the '6 second rule', the ATP energy 'reservoir' and '30 for 80'. There are practical tips for patients, which also illustrate the impact on their every day life. 164 page paperback.



## In the event of overdoing it

Very occasionally, a person with McArdle's may get something wrong and hurt themselves.

- Allow them to rest and have water to drink.
- Ask if you should call a partner or relative.
- In severe cases, ask if they need to be taken to hospital†.
- They carry a card to remind them of the decision points.



† For most people with McArdle's it is a very rare occurrence to need to be hospitalized.



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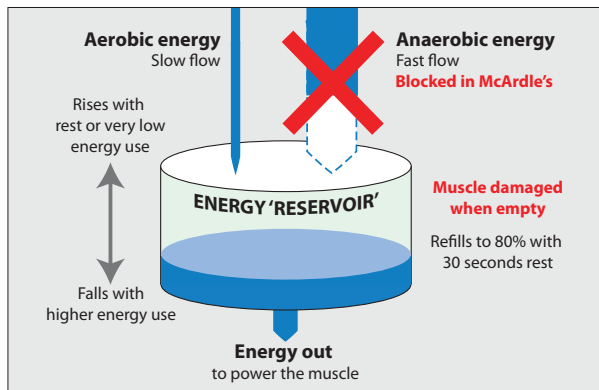
## Their rare genetic condition

McArdle's is very rare, affecting about 1 person in every 100,000. It is inherited from parents who are both carriers but who do not have symptoms themselves. Each of the children of carriers have a 25% chance of having McArdle's.

## Their issue with energy

People with McArdle's cannot use energy which is stored in their muscles, due to a missing enzyme. With intense activity they run out of energy in 6 to 10 seconds. With more gentle activity like walking on the flat it may take a minute or two.

- The 'fast' anaerobic energy pathway is blocked.
- There is a severe shortage of energy supply at times.
- Cramping/pain occurs if the energy 'reservoir' is emptied.



## The risks they face

Pain, cramping, exhaustion and embarrassment are usual. Here are just two examples of the types of issue that arise.

### ■ An everyday problem to avoid

Walking on the level at their friend's pace, in a minute their legs feel heavy. Embarrassed, they struggle on. Cramping starts. They slow down but it *really* hurts. Legs like cement, they can hardly move. Heart pounding, feeling sick and light headed, they grind to a halt.

### ■ A contracture and rare complications

They carry a grocery bag from the car, it's heavy but they manage. Their hand is making a fist, their fingers won't open. More than a cramp, a fixed contracture - muscles swollen, locked solid. The pain is extreme. Next morning their hand is tingling, numb and an odd color. Off to hospital. Intravenous fluids and paid medication are administered. In rare cases, renal failure and/or compartment syndrome are diagnosed.

## Some strategies they use

A person with McArdle's will use these key strategies to help them handle their physical activity effectively and avoid pain.

### Slowing or pausing to balance energy

During activity such as chewing, brushing hair, drying after a shower and walking, people with McArdle's:

- Slow down to ease the level of energy demanded.
- Pause for 30 seconds rest, then continue more slowly.
- Do these frequently in the first 10 minutes of activity.
- Find symptoms easing a little after about 10 minutes, when 'second wind' occurs.
- With each muscle group used, this process is repeated.



### Breaking intense tasks into sections

During maximum intensity tasks (such as carrying a heavy object, opening a new jam jar, squatting or standing on tip toe) people with McArdle's will usually:



- Interrupt the task after 6 seconds.
- Rest for 30 seconds so that the energy 'reservoir' refills, then try again.
- This avoids a painful contracture which may otherwise occur, last for days, and might need medical attention.

### Resting before taking a risk

Before a risk such as crossing a road, climbing a ladder or lifting something heavy, they often first rest for at least 30 seconds.

This should ensure that enough immediate energy is available to get them safely through the activity instead of grinding to a halt as the energy 'reservoir' empties.



### The medical jargon explained

**Aerobic** - activity which requires oxygen in converting fuel sources into energy. Walking is a good example and is good exercise for McArdle's.

**Anaerobic** - activity which does not need oxygen to utilise fuel, e.g. gripping. Energy is produced from muscle glycogen, which McArdle people cannot do.

**Contracture** - when a muscle becomes locked up, swollen and extremely painful. Often lasts for days. Can have very serious complications.

**Inheritance** - McArdle's is inherited in an autosomal recessive pattern.

**Isometric** - muscular action which is 'static', e.g. clenching fists, holding something up, pushing. The worst type of activity for McArdle's people.

**Myophosphorylase** - the enzyme which is deficient or absent in McArdle's.

## Some things they do

The majority of people with McArdle's are able to learn techniques, adapt activities, accommodate the condition, remain active and live *relatively* normal lives.

### Things they do for protection

- Restricting anaerobic and isometric activity to 6 seconds greatly reduces the risk of a getting a muscle contracture.
- Avoiding risky situations, such as swimming in deep water or climbing to heights reduces the risk of accidents.

### Things they do for benefit

- Getting into 'second wind' after about 10 minutes of careful activity, when the muscles start to make more use of sugar released from the liver, and fat, makes activity a little easier.
- Regularly exercising in 'second wind' (45 minutes, five days a week in recommended) improves aerobic conditioning and makes more activity possible with fewer problems.

## How you can help them

- When walking together, allow them to set the pace.
- If they pause for a rest, don't make a fuss or draw attention, just wait patiently.
- Be understanding of their need to rest more frequently early in activity than they do later on.
- Read the "101 Tips" book (see back cover) to get a good idea of what they should and should not do.
- Recognise that even simple things can prove difficult or even risky (e.g. carrying groceries, moving furniture, vacuuming, etc).
- Around the house, divide the chores so that they do the more aerobic ones and you do the more anaerobic ones.
- Watch out for them stubbornly doing something anaerobic, and especially isometric, that they should not do. Offer to take over from them.
- Give them gentle encouragement and support to keep active and build activity into their daily life.

## Why they avoid contractures

- Broken down muscle (myoglobin) released into the bloodstream can cause acute kidney failure which can be life threatening and must be treated urgently.
- Very rarely, a contracture may develop into compartment syndrome, requiring urgent surgery to prevent permanent damage to muscles and nerves.
- Repeated contractures over many years may make activity harder in later life.