

McArdle Disease emergencies

Guidance for McArdle patients and an alert to emergency doctors of the risks of a McArdle episode. More detail below. The doctor will treat each episode according to the individual circumstances. [Please read our disclaimer.](#)

Guidance for the McArdle patient

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

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

†A few people have experienced these symptoms without unusual 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 nothing to worry about.)

Feeling very unwell after exercise

Feeling very unwell after exercise (perhaps with '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.

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 exercise. 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 surgical referral.

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 help.

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.

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 above notes (in the red box).

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 produced by your muscle breakdown and released 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 left 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 putting ourselves at risk. We have information available to help you adapt 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.

- **Adapting activity** understanding how to protect ourselves
- **Second wind** - help with beneficial aerobic exercise
- **Starting an exercise programme** - advice for those who are very de-conditioned
- **The six second rule** - help to avoid contractures

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Links to further information

[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

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