TO PATIENT

When to seek medical attention

You may occasionally get something wrong and hurt yourself. This will normally be due to physical activity. Refer to this leaflet or your emergency card.

- Rest and have water to drink.
- Ask a relative/friend to help assess situation.

Go to hospital if *any* of the following apply, you:

- Have a severe cramp or contracture, or
- Have severe muscle pain, or
- Have cola-colored urine, or
- Have significantly reduced urine output, or
- Feel very unwell ('flu-like aches and fever), or
- All the above are OK and yet you have a CK result many times your baseline level.

If possible take a urine sample with you to show on arrival. If you hold a personalized emergency letter take that also.

On arrival at the hospital

- Immediately book in at emergency registration.
- Explain that you are suffering from rhabdo-myolysis and show your urine sample.
- Show this leaflet (and emergency letter, if held).
- Point out the red panel 1, inside on left.

With the emergency doctor

- Provide a copy of this leaflet.
- Be ready to answer questions about McArdle's.
- Point out references and IamGSD web site.
- Expect to be put on intravenous (IV) fluids.

Rehabilitation

- Following discharge you are likely to still need to rest for at least several days.
- Gradually rebuilding aerobic capacity of the damaged muscles may take some weeks.

References

CLINICAL PRACTICE GUIDELINES

[1] Clinical Practice Guidelines for glycogen storage disease V & VII (McArdle and Tarui) from an international study group. *Neuromuscular Disorders 31* (2021) 1296–1310. https://doi.org/10.1016/j.nmd.2021.10.006

[2] Emergencies, section 6.

[3] Rhabdomyolysis, section 6.1.

[4] Acute Renal Failure, section 6.2.

[5] Compartment Syndrome, section 6.3.

[6] Laboratory testing, section 5.3.

Visit the following page to download free PDFs of the 15 page paper, plus 18 pages of Supplementary Material.

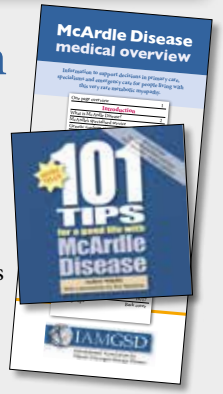
www.iamgsd.org/cpg

Further information

A range of publications is available on our website. Browse on-line or download free PDFs, such as:

Medical overview: A quick guide for medical professionals. 20 pages.

101 Tips for a good life: Simple, practical tips for people living with McArdle's. 164 pages.



IAMGSD
International Association for Muscle Glycogen Storage Disease

www.iamgsd.org

v1 – 05/2022

At hospital with McArdle's

Information for emergency visits

When to seek urgent medical attention. Guidance for assessment and management. References to Clinical Practice Guidelines [1].

Triage needed

- Patient is likely to report with severe muscle cramping or contracture/s and extreme pain [2].
- May develop rhabdomyolysis [3], myoglobinuria and a risk of Acute Renal Failure [4], with severe episodes requiring dialysis.
- Risk of Compartment Syndrome [5] requiring urgent surgical intervention to relieve pressure.

Introducing McArdle Disease

- McArdle disease (Glycogen Storage Disease 5) is an ultra rare genetic metabolic myopathy.
- Myophosphorylase is deficient, so glucose cannot be released from muscle glycogen.
- A severe energy crisis arises early in all physical activity and throughout isometric and anaerobic activity.
- If patient is well managed, urgent medical care is rarely needed. However, patients can make mistakes or be caught out by circumstances.

IAMGSD
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TO DOCTOR

1 Urgent assessment

Following activity, patients may present with:

- Muscle cramps or extreme fixed contractures with swelling and severe pain.
- Myoglobinuria, oliguria or anuria or feeling very unwell ('flu-like aches and fever).

Do not be concerned about the McArdle disease itself, but consider rhabdomyolysis, and:

ASSESS FOR ACUTE RENAL FAILURE [3]

- Urine analysis for myoglobinuria.
- Full chemistry panel – Creatine Kinase (CK or CPK, see right), glucose, calcium and bone profile, urea and electrolytes.

2 Suggested management

- IV fluid bolus, then saline at 2x maintenance and (unless diabetic) consider if 10% dextrose needed to keep blood glucose >3.5 mmol/L (>61 mg/dL).
- Monitor urine output, CK and electrolyte status.
- Consider possible complications of Acute Renal Failure and Compartment Syndrome (see right).
- It is essential to adequately treat pain: no contra indication for intramuscular injections.

3 Guidelines for discharge

Each patient must be assessed individually, but the following are suggested guidelines.

- Renal function lab results are normal.
- Urine volume is fully adequate.
- Patient does not feel unwell or nauseous, and is tolerant of oral fluids.
- CK has trended down for 3 consecutive tests at intervals of approx. 12 to 24 hours.

If all above points are compliant, discharge may be considered with CK many times patient's baseline.

Possible complications

There are two main risks of serious complications, as for any episode of rhabdomyolysis, both are rare.

ACUTE RENAL FAILURE (ARF) [4]

a) Renal dysfunction is predominantly related to myoglobin-direct tubular cytotoxicity, vasoconstriction and tubular obstruction.

b) Severe rhabdomyolysis should be treated with adequate fluid administration to prevent renal impairment, or patient should be put on dialysis if warranted.

c) Take into account the fluid balance to avoid further complications such as hypervolemia and acute pulmonary edema.

d) For patients that develop ARF, consultation with nephrology is required.

COMPARTMENT SYNDROME (CS) [5]

a) Swelling associated with muscles which are in contracture can lead to Compartment Syndrome.

b) Assess for Compartment Syndrome including checking the pressure within the fascia surrounding the affected muscles.

c) Consider the need for surgical intervention and, if necessary, make urgent referral.

For further information on medical emergencies:
www.iamgsd.org/urgent

For references to detailed Clinical Practice Guidelines and further information, see back cover.

A note on: Creatine Kinase

a) The patient may attend due to an unusually high plasma Creatine Kinase (CK / CPK) level reported in primary care following a McArdle injury.

b) Baseline levels can be approx. 2,000–5,000 IU/L [6] and can vary significantly with activity (labs typically quote normal values <200 to <350 IU/L). Patient may be aware of their own baseline level.

c) A high CK need not indicate a cardiac event. If there is any concern, more specific proteins (e.g. Troponin I) can give a clearer indication.

d) With rhabdomyolysis CK can be much higher, even in excess of 100,000 IU/L (see note below).

e) CK usually peaks 24 hours after a McArdle injury, then falls by approx. 30% to 50% per 24 hours.

A note on: Liver Enzymes

a) McArdle patients routinely have mildly elevated plasma levels of the liver enzymes ALT and AST [6].

b) These enzymes are also released when skeletal muscle is damaged, and are therefore normally not a matter for concern regarding the liver.

c) Investigation may be indicated if ALT and AST are grossly elevated disproportionate to the CK, or if ALP or bilirubin levels are significantly raised.

Risk of erroneous lab reports

CREATINE KINASE

a) Be wary of CK results not following the above pattern, and of any reported as ">X" IU/L.

b) Such reports may indicate a mistake made due to the level being outside test limits.

c) This requires dilution of the blood sample and re-test until within the test limit, then the result being multiplied up by the dilution factor.

d) Incorrect reports mask the true course of the episode and can lead to errors in treatment.

MYOGLOBINURIA

e) Differentiate between myoglobinuria and hematuria.