

McArdle Disease medical overview

Information to support primary care decisions for people living with this very rare metabolic myopathy.

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One page overview

This booklet provides referenced information to assist GPs and other medical professionals. McArdle Disease should be managed by a specialist service, but this booklet can help with issues arising in primary care.

Using this booklet

- **Vital points are in bold.**
- Patient should hold a copy. Discuss on phone by reference to numbered points.
- See the last pages for further reading and support.

Electronic versions

PDF – may be downloaded free of charge at: www.euromacregistry.eu

Online – free access, search for “McArdle Disease” on books.google.com.

Validity

This booklet was originally drafted by AGSD-UK, based on the experience of people with McArdle Disease.

The information is intended for use by primary care and other health professionals with the support of a highly specialised service for people with McArdle disease.

If in any doubt please refer to a specialist in McArdle Disease.

Updates

Any amendments to this edition, and the announcement of any new edition, will be posted on www.euromacregistry.eu.



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- McArdle Disease is a very rare autosomal recessive disorder of muscle metabolism.
- Specialist services are essential to achieve correct diagnosis and ensure good management.
- Genetic confirmation is necessary to avoid potentially damaging misdiagnosis.
- There is an inability to utilise muscle glycogen.
- Maximal isometric activity must be ceased by approx. 6 seconds to avoid the risk of the muscle entering a fixed contracture.
- A shortage of energy occurs in all activity. It is severe in the first 10 minutes and throughout all intense activity.
- This leads to premature fatigue, exaggerated heart rate, pain, muscle spasm and fixed contracture.
- Diagnosis is often delayed about 20 years, during which time muscle damage or atrophy may occur.
- Patients diagnosed in childhood may learn to manage their condition and be less severely affected – although always at risk of contracture.
- Regular exercise is essential to enhance aerobic metabolism and reduce the risk of muscle damage.
- Patients need to be able to recognise the signs that urgent hospital attendance is required.
- Patients are at risk of some concomitant conditions, but not all issues are related.
- Sharing with other patients through support groups can help considerably.

What is McArdle Disease?

- a) McArdle Disease¹ (Glycogen Storage Disease Type V) is an ultra rare muscle myopathy with an incidence of approx. 1 in 100,000. (Muscle GSDs are very different to liver GSDs.)
- b) **Deficiency of the myophosphorylase enzyme², caused by mutations in the PYGM gene, results in an inability to utilise glycogen in skeletal muscle.**
- c) Maximal isometric activity for more than approx. 6 seconds will risk fixed contracture.
- d) A serious shortage of energy occurs in the first 10 minutes of activity. This leads to premature fatigue, exaggerated heart rate, pain and muscle spasm.
- e) **If activity is continued in the presence of pain, a fixed contracture can occur with risks of rhabdomyolysis, acute renal failure and/or compartment syndrome (page 7).**
- f) **Patients must be able to recognise the signs that urgent hospital attendance is required (page 7).**
- g) A series of CK tests (page 8c) can help patients understand and avoid future serious episodes.
- h) In many cases diagnosis is achieved approx. 20 years after presentation. Muscle damage or atrophy may have occurred by then, causing disability.
- i) Diagnosis in childhood, provision of specialist advice and learning to manage the condition well, can lead to being less severely affected.
- j) Patients must learn to attain 'second wind' (page 10f). Regular exercise enhances aerobic metabolism and reduces risk of muscle damage.

1) Myopathy due to a defect in muscle glycogen breakdown. McArdle, B. (1951) Clin. Sci. 10: 13-33.

2) A functional disorder of muscle associated with the absence of phosphorylase. Mommaerts, W.F., Illingworth, B., Pearson, C.M., Guillory, R.J., Seraydarian, K. (1956) Proc Natl Acad Sci U S A 45: 791-797.

McArdle's specialised service

- a) Due to the rarity of McArdle's, most neurologists will see only one or two patients. They therefore cannot gain a full understanding of the condition.
- b) Referral to an expert centre with a multidisciplinary approach to patient care is therefore highly desirable.
- c) **A specialised service for people with McArdle Disease and related disorders may be available in your country. Visit www.euromacregistry.eu and follow 'Useful info' for a list of McArdle Disease physicians by country.**
- d) Patient support groups often liaise with these specialist services and support patients attending the clinics.

† For other countries, we suggest requesting the names of suitable specialists from national support groups for Glycogen Storage Disease or Muscular Dystrophy. Also try searching PubMed for research papers on McArdle Disease and consider the names and institutes of the contributors.



Genetically confirmed patients are requested to register with Euromac – the registry for people with McArdle Disease and other very rare glycogenoses.

www.euromacregistry.eu

Genetic confirmation

- a) Due to the ultra rare nature of McArdle Disease, mistakes have been made in diagnosis¹.
- b) **If patient has an atypical presentation, check whether they have been genetically confirmed.**
- c) DNA analysis of the *PYGM* gene may be undertaken² from blood taken locally.
- d) Over 150 causative mutations identified to date.
- e) In the UK 96% of people with McArdle's have at least one of the two most common mutations¹.

1) **McArdle Disease: a clinical review.** Quinlivan, R., Buckley, J., James, M, et al. J Neurol Neurosurg Psychiatry. doi: 10.1136/jnnp.2009.195040.

2) **Intron/exon structure of the human gene for the muscle isozyme of glycogen phosphorylase.** Burke, J., Hwang, P., Anderson, L., Lebo, R., Gorin, F., and Fletterick, R. (1987) Proteins 2: 177-187.

Inheritance

- a) **McArdle Disease is inherited in an autosomal recessive pattern¹.**
- b) General practices are very unlikely to see a second patient, other than possibly a sibling.
- c) The prevalence has been estimated to be 1:100,000, based on a carrier frequency of approx. 1:160^{2,3}. In UK it appears less than half of cases are diagnosed.
- d) The risk of a patient partnering with a carrier and their child having McArdle's is approx. 1 in 320.
- e) For patients of childbearing age, referral for genetic counselling may be appropriate.

1) **Online Mendelian Inheritance in Man.** <http://omim.org/entry/232600>

2) **Treatment of McArdle disease.** Haller RG. Arch Neurol 2002; 57:923-4..

3) **A novel mutation in the PYGM gene in a family with pseudo-dominant transmission of McArdle Disease.** Isackson, P.J., Tarnopolsky, M., and Vladutiu, G.D. (2005) Mol Genet Metab, 85: 239-242.

Concomitant conditions

- a) **There are several medical conditions to which people with McArdle Disease are more prone than the rest of the population.**

Insulin resistance

High muscle glycogen concentrations in skeletal muscle¹ or a sedentary lifestyle may contribute.

Hyperuricaemia

Due to high level of purine metabolism, possibly leading to gout and/or renal calculi² (page 9).

Obesity (and all its ill effects)

Many are overweight, contributed to by the avoidance of activity due to associated painful cramping if not guided correctly³.

Depression and anxiety

Factors include living with a chronic condition and worry about severe episodes and the need for hospitalisation³.

- b) Chronic renal failure is *not* reported to be associated. However, some cases of rhabdomyolysis lead to *acute* renal failure (page 7).
- c) Data suggests that McArdle's does *not* significantly increase the risk of complications for pregnancy and delivery³. Anecdotal evidence shows McArdle symptoms being reduced during pregnancy.
- d) **Like anyone else, people with McArdle's may develop other diseases. Care should be taken to properly investigate and not assume that reported symptoms are due to the patient's McArdle Disease.**

1) **Decreased insulin action in skeletal muscle from patients with McArdle's disease.** Nielsen JN, Vissing J, Wojtaszewski JF, Haller RG, Begum N, Richter EA. (2002) Am J Physiol Endocrinol Metab. Jun;282(6):E1267-75.

2) **McArdle's disease and gout.** Puig, J.G., de Miguel, E., Mateos, F.A., Miranda, E., Romera, N.M., Espinosa, A., and Gijon, J. (1992) Muscle Nerve 15: 822-828.

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

Cramps and contractures

- Patients should try to avoid incurring cramps that last for more than a minute or two.
- Intense or isometric activity for more than approx. 6 seconds will risk severe cramps or fixed contractures which last for hours or days.**
- Such contractures can be incurred accidentally or in extremis (e.g. having to run away from danger).
- Pain medication will usually be required and medical attention often so.
- Muscles recover from fixed contractures but frequently-repeated contractures can accumulate debilitating damage in the long term.**

Pain medication

- During episodes of fixed contracture or rhabdomyolysis patients are advised to choose pain medications which are metabolised in the liver. Avoid those metabolised in the kidneys, due to the stress on the kidneys.
- Pain medication should be avoided in the absence of an episode, as it will obscure the signals from the muscles. These are needed for patients to recognise when to slow down or pause for a rest in order to avoid injury¹.**
- If some muscle injury is incurred, pain relief should be taken *only once activity has ceased*.
- Patients who start on opioid medications are at risk of dependency and of developing chronic pain¹.

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

Medical emergencies

- People with McArdle's are at risk of episodes of rhabdomyolysis with possible acute renal failure and/or compartment syndrome¹.**
- Episodes cannot be managed in general practice. Patients must understand when to attend hospital. They should always carry an 'Information Card' which has guidance (page 16).
- Patients should hold a letter from their McArdle specialist to show on arrival at the hospital, advising recommended actions. (In the absence of such a letter patients should show their card and/or the panel below.)
- Kidneys normally recover fully following an episode of rhabdomyolysis and myoglobinuria.

1) **Exertional rhabdomyolysis: a clinical review with a focus on genetic influences.** Landau ME, Kenney K, Deuster P, Campbell W. (2012) J Clin Neuromuscul Dis. Mar;13(3):122-36. doi: 10.1097/CND.

Information for the hospital doctor

Patients may present with muscle cramps, fixed contractures, myoglobinuria, oliguria or anuria or feeling very unwell ('flu like aches and fever) following activity. Do not be concerned about McArdle's per se, but instead:

Urgent assessment for rhabdomyolysis

- Consider urine analysis for myoglobinuria and full chemistry panel – CK (page 8), glucose, calcium and bone profile, urea and electrolytes.

Suggested management

- IV bolus then saline at 2x maintenance and (unless diabetic) 10% Dextrose to keep blood glucose >3.5 mmol/L.
- Monitor urine output, CK and electrolyte status.

Potential complications

- Acute renal failure – prompt referral for haemodialysis.
- Increased swelling causing compartment syndrome – assess the need for urgent surgical intervention.

Creatine Kinase

- a) **People with McArdle's have raised CK. Basal levels can be approx. 2,000–5,000 iu/l (normal values <190)¹ and can vary significantly with activity. With episodes of rhabdomyolysis CK can be much higher, even in excess of 100,000 iu/l.**
- b) **Establish a basal level for the patient by testing several times in the absence of recent injury.**
- c) The results of CK tests following a series of injuries will assist the patient to understand how much damage they are doing. This will help them to manage their condition and avoid serious episodes.
- d) **Set up a mechanism for the patient to be able to access immediate CK testing when they injure themselves. Abnormal results should be urgently communicated to the patient.**
- e) CK tends to peak 24 hours after a McArdle injury, then falls by approx. 30% to 50% per 24 hours.
- f) Be wary of results that do not follow the above pattern, or that are reported as “above” a figure. This may indicate an error by the lab due to a result which is grossly outside the lab's normal experience.
- g) An unusually high CK level after an injury may alert the patient to consider the need to attend hospital. (See *Medical emergencies*, page 7.)
- h) **A high CK need not indicate a cardiac event. If there is any concern, cardiac-specific enzymes (e.g. Troponin) give a clearer indication.**

1) **McArdle Disease: a clinical review.** R Quinlivan, J Buckley, M James, et al. (2010) *J Neurol Neurosurg Psychiatry*. Published online September 22, ; doi: 10.1136/jnnp.2009.195040.

Blood

URATE

- a) Urate levels are often raised in people with McArdle's, due to increased purine metabolism¹.
- b) **There is an increased rate of gout amongst McArdle people compared to the rest of the population². (Approx. 10% compared to 3%).**
- c) Renal stones of uric acid crystals and calcium oxalate can be instigated by raised urate levels³.

LIVER ENZYMES

- d) **Routine screening for unrelated matters may reveal, in people with McArdle's, mildly elevated levels of the liver enzymes ALT and AST⁴.**
- e) These enzymes are released into the blood when skeletal muscle is damaged⁵ and is normally not a matter for concern regarding the liver.
- f) Further investigation may be indicated if the ALT or AST levels are grossly raised and remain so.
- g) Further investigation is also indicated if the ALP or bilirubin levels are significantly raised.

1) **McArdle's disease and gout.** Puig, J.G., de Miguel, E., Mateos, F.A., Miranda, E., Romera, N.M., Espinosa, A., and Gijon, J. (1992) *Muscle Nerve* 15: 822-828.

2) **McArdle Disease: a clinical review.** R Quinlivan, et al. (2010) *J Neurol Neurosurg Psych*. Published online September 22; doi: 10.1136/jnnp.2009.195040.

3) **Kidney stones: pathophysiology and medical management.** Orson W Moe. (2006) *Lancet*; 367: 333–44.

4) **McArdle's disease: case report and review of the literature.** Tuzun, A., et al. (2002) *Turk J Gastroenterol* 13: 56-59.

5) **Serum Alanine Aminotransferase in Skeletal Muscle Diseases.** (2005) Rahul A. et al. *Hepatology*, Volume 41, Issue 2.

Urine

- a) If urine test strip shows blood (haemoglobin) or protein, consider whether this is in fact myoglobin which is often present in McArdle's.

Problems with activity

- a) Everyday activities can cause problems, e.g.: chewing, cleaning teeth, hanging out washing, drying after a shower, standing on tiptoe.
- b) **As the anaerobic metabolic pathway is blocked, people with McArdle Disease should avoid intense activity, especially isometric and repetitive actions.**
- c) During more gentle activity, patients need to recognise the signals from the muscles that warn them to slow down or pause for a rest.
- d) Rushing and sudden activity from a rested state are both likely to lead to muscle cramps.
- e) **'Six second rule'¹ – If a patient has to undertake any activity at maximal effort (e.g. opening a jam jar, running for a bus), they are advised to limit duration to 6 seconds². They can try again after resting for at least 30 seconds.**
- f) **'Second wind'³ – alternative energy pathways (fat, amino acids, glucose from the liver glycogen stores) help to some extent when they start to come into use after about 8 to 10 minutes.**
- g) Patients must learn the techniques for safely achieving 'second wind'³. It is universal to all patients, but some need help to recognise it.
- h) Tensing muscles (e.g. due to anger, fear or excitement) greatly increases the risk of injury.

1) **101 Tips for a Good Life with McArdle Disease.** (2013) Wakelin, Andrew. Association for Glycogen Storage Disease. (See back cover.)

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

3) **Outcome Measures in McArdle Disease.** Quinlivan, R., Vissing, J. (2006) 144th ENMC International Workshop, 29 Sept-1 Oct 2006, Naarden, The Netherlands. *Neuromuscular Disorders* 17: 494-498.

Beneficial exercise

- a) **Relatively gentle aerobic exercise is very beneficial for people with McArdle Disease¹ – it helps to improve their aerobic metabolism.**
- b) Exercise is helpful with the important task of keeping weight under control.
- c) People with McArdle Disease, no matter what age they are, need to have a programme of regular exercise².
- d) **At least 45 minutes of aerobic exercise, after attaining 'second wind' (page 10f), five times a week, is strongly recommended.³**
- e) The common mantra 'No pain, no gain' is wrong in McArdle Disease.
- f) Uncontrolled studies suggest that aerobic training is safe, with improvements in physiological parameters after several weeks. Controlled trials are needed to assess the therapeutic effect⁴.
- g) **If patients are very de-conditioned they need to start their exercise routine very carefully. This should be done under the supervision of a McArdle specialist.**

1) **Aerobic conditioning: an effective therapy in McArdle's Disease.** (2006) Haller RG, Wyrick P, Taivassalo P, et al. *Ann Neurol*; 59: 922e8.

2) **Exercise capacity in a 78 year old patient with McArdle's disease: it is never too late to start exercising.** Perez, M. et al. (2006) *Br J Sports Med* 40: 725-726.

3) **The 'McArdle paradox': exercise is good advice for the exercise intolerant.** Lucia, A., Quinlivan, R., Wakelin, A., Martín, M.A., Andreu, A.L., et al. (2012) *Br J Sports Med* doi: 10.1136.

4) **Cochrane Review: Physical training for McArdle Disease.** Quinlivan, R., Vissing, J., Hilton-Jones, D., Buckley, J. (2012)

Physiotherapy

- a) **Any physiotherapist working with a McArdle Disease patient should be fully briefed by the patient's McArdle specialist prior to first consultation. Common errors include:**
 - Holding stretches too long (page 10e).
 - Exercising for short periods without achieving 'second wind' (page 10).
 - Advising that pain is acceptable without realising that patients do not have a rise in lactic acid, see (f) below.
 - Failing to monitor CK levels to ensure that muscle damage is not being incurred (page 8).
- b) Once a safe and effective exercise programme has been prescribed by the McArdle specialist, local facilities can be used to support patients.
- c) Regular follow up will motivate patients, help to ensure their compliance with the prescribed exercise regime and avoid damaging activities (pages 10 and 11).
- d) If physiotherapy is required for other conditions or following unrelated injuries, stretches should be limited to approx. 6 seconds so that energy demand does not extend beyond the depletion of ATP in the muscles (page 10e).
- e) Should massage be required, this should be gentle, not deep, massage. Massage should not be applied to a muscle which is in a rigid, fixed contracture.
- f) McArdle patients do not experience a lactic acid 'burn' as their lactic acid does not rise on exercise¹.

1) **A nonischemic forearm exercise test for McArdle Disease.** Kazemi-Esfarjani P, Skomorowska E, Jensen TD, Haller RG, Vissing J. *Ann Neurol.* 2002 Aug;52(2):153-9.

Diet

- a) People with McArdle's may have a problem with weight gain due to an aversion to exercise caused by their symptoms (page 10a).
- b) **A healthy diet with an appropriate calorie intake to maintain ideal weight is recommended.**
- c) Diet remains controversial and further research is required.
- d) A limited study in 2008 indicates that a carbohydrate-rich diet may be advantageous. (20% fat, 15% protein, 65% carbohydrate – using low glycaemic index foods such as vegetables, fruits, pasta, rice, bread and low-fat cheese)¹.
- e) A 1985 case report had suggested a high protein diet². Some patients report doing better on a high protein diet, and yet others on high fat.
- f) It is important to keep hydrated during activity.
- g) 37 g of sucrose (143 calories, equivalent to 9 teaspoons of table sugar)†, in a drink 5 minutes before short-term intense activity (such as sexual intercourse) may ease symptoms in the first 15 minutes³. (Some risk of reactive hypoglycaemia.)
- h) Frequency of use of sucrose should be limited, to avoid the risk of weight gain, e.g. twice per week.
- i) To date, no nutritional treatments have been adequately proven to be effective⁴ in the short or long term.

† Equivalent to a 12 oz (355 ml) can of Coca Cola.

1) **Carbohydrate- and protein-rich diets in McArdle Disease: Effects on exercise capacity.** Andersen, S. T., Vissing, J. (2008) *J. Neurol. Neurosurg. Psychiatry* published online 5 Jun; doi:10.1136/jnnp.2008.146548.

2) **Myopathy in McArdle's Syndrome: Improvement with a high-protein diet.** Slonim A.E., Goans, P.J., *New Eng. Jnl. Medicine*, (1985).

3) **Effect of oral sucrose shortly before exercise on work capacity in McArdle Disease.** Andersen, S.T., Haller, R.G. and Vissing, J. (2008) *Arch Neurol* 65.

4) **Cochrane Review: Pharmacological and nutritional treatment for McArdle Disease.** Quinlivan, R., Martinuzzi, A., Schoser, B., (2014).

GPs are alerted to the following areas of risk when treating other conditions in people with McArdle's. Care should also be taken when referring patients for physiotherapy (page 12).

Statin therapy

- Although statin medications are generally well-tolerated, the most common side effects relate to skeletal muscle (myalgia, myositis, rhabdomyolysis)¹.
- People with McArdle's may be at increased risk of muscle side effects from statins.**
- If a patient needs a statin, establish their basal CK level (page 8) before initiating treatment.
- Monitor symptoms and plasma CK weekly for the first few weeks, then extend the interval in stages to normal practice.
- Other cholesterol lowering drugs may also worsen myopathy in McArdle patients².

1) **Genetic risk factors associated with lipid-lowering drug induced myopathies.** Vladutiu, G.D. et al. (2006) *Musc Nv* 34: 153-162.

2) **Worsening myopathy associate with ezetimibe in a patient with McArdle Disease.** (2005) Perez-Calvo J, Civeira-Murillo F, Cabello A. *QJM*;98:461e4.

Drug side effects

- When prescribing for other medical conditions always check for any side effect of rhabdomyolysis (for example succinylcholine).**
- For a list see table 12.1 on page 145 of *The McArdle Disease Handbook* (page 17), free access on Google Books. Derived from British National Formulary¹.

1) **British National Formulary.** Joint Formulary Committee London: British Medical Association and Royal Pharmaceutical Society.

General anaesthetic

- McArdle Disease may increase the risk of a malignant hyperthermia-type of reaction to anaesthetic agents¹.**
- Rhabdomyolysis, acute renal failure and electrolyte abnormalities may ensue.
- Although a very small risk, it is advisable to inform the anaesthetist of the risk prior to any surgery. Choice of low risk agents and careful monitoring can further lower the risk.

1) **McArdle's disease and anaesthesia: case reports. Review of potential problems and association with malignant hyperthermia.** Bollig, G., Mohr, S., and Raeder, J. (2005) *Acta Anaesthesiol Scand* 49: 1077-1083.

Tourniquets

- People with McArdle Disease are at increased risk of developing compartment syndrome through use of tourniquets¹.**
- Prior to surgery, advise the surgeon of the risk.
- Limit length of use of blood pressure cuffs.

1) **Acute compartment syndrome after forearm ischemic work test in a patient with McArdle's disease.** Lindner, A., Reichert, N., Eichhorn, M., and Zierz, S. (2001) *Neurology* 56: , 1779-1780.

Examinations

- Patients may be at risk of cramping when holding an awkward position for examination or treatment.

Information Cards

Patients should carry one of these credit card-sized Information Cards in case of an unexpected serious episode. Available from your support group or McArdle's consultant.



These cards have a:

- Short description of the condition.
- Note of when McArdle people may need assistance.
- Reminder of the circumstances in which to attend hospital.
- Link to emergency treatment suggestions, for hospital doctor.

Further reading

Comprehensive papers on McArdle Disease:

McArdle Disease: a clinical review.

Quinlivan, R., Buckley, J., James, M., et al. (2009) J Neurol Neurosurg Psychiatry. doi: 10.1136/jnnp.2009.195040.

McArdle Disease: what do neurologists need to know?

Lucia, A. et al. (2008) Nat Clin Pract Neurol 4: 568-577.

Genotypic and phenotypic features of McArdle Disease: insights from the Spanish national registry.

Lucia, A., Ruiz, J.R., Santalla, A., et al. J Neurol Neurosurg Psychiatry (2011). doi:10.1136/jnnp-2011-301593

Outcome Measures in McArdle Disease.

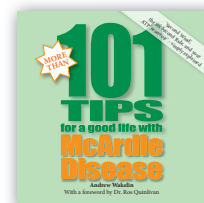
Quinlivan, R., Vissing, J. (2006) 144th ENMC International Workshop, 29 Sept-1 Oct 2006, Naarden, The Netherlands. Neuromuscular Disorders 17: 494-498.

These books are available for free on-line searching and browsing via Google Books. Printed copies can be purchased from: www.agsd.org.uk

101 Tips for a good life with McArdle Disease

Andrew Wakelin, AGSD-UK

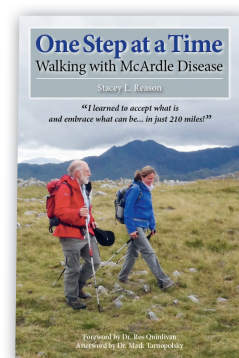
164 page pocket-sized paperback with practical tips which McArdle people have found to be useful. Plus simple explanations of key subjects – ‘second wind’, six second rule, ATP ‘reservoir’ and guidance on emergencies. This book will help McArdle people to avoid pain and cramps, reduce hospital visits and get more out of life. *Some other language versions are also available.*



One Step at a Time Walking with McArdle Disease

Stacey L Reason

This large paperback (128 pages, 250 colour images) traces the route, thoughts and emotions of a McArdle patient on a life-altering journey of discovery and growth when she walks over two hundred miles. It provides a truly awakening narrative for patients, families and health professionals. Complete with an account of the development of McArdle's walking courses and 30 page guidance section.

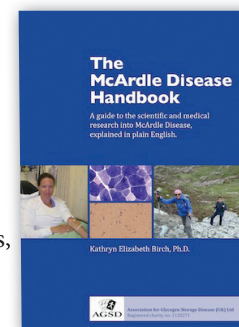


The McArdle Disease Handbook

Kathryn Elizabeth Birch, PhD

Very useful for doctors wanting more in-depth information.

Fully referenced to over 260 original research papers. 208 pages, large paperback. A guide to the scientific and medical research into McArdle Disease. Covers the cause, inheritance, history, symptoms, emotional aspects, treatments, and all the issues which can face McArdle people. Also available in other language editions: German (printed) and Italian (PDF only). **Free access on Google Books.**



Patient support groups

In many countries people affected by McArdle Disease and other rare muscle glycogenoses are assisted by support groups for glycogen storage disease or muscular dystrophy.

These groups can provide information, put people in contact and issue newsletters. Some hold conferences and practical training courses.

Social media groups

The main social media group for McArdle Disease is on Facebook. Search Facebook for 'McArdle's Disease'. It has over 1,500 members worldwide. There are also some smaller groups for special interests, such as parents and diet.

McArdle Disease at AGSD-UK



AGSD The AGSD-UK web site contains extensive information about McArdle's. They publish leaflets, Information Cards and books written in non-medical language (page 17). They produce videos and offer courses for practical guidance.

www.agsd.org.uk

