McArdle Disease medical overview

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

IAMGSD

International Association for Muscle Glycogen Storage Disease

IamGSD English edition 1

Using this booklet

- Vital points are highlighted in bold and blue.
- Both medical professional and patient should hold a copy.
- Points are referenced alphabetically for ease of referral.
- See the last pages for further reading and support.

Electronic versions

A PDF may be downloaded free of charge, or refer to flip-through version, both via our website.

Validity

This booklet was originally drafted by AGSD-UK, based on the experience of people with McArdle disease, and updated by Euromac and IamGSD.

The information is intended for use by primary care and other health professionals with the support of a highly specialized service for people with McArdle disease. Please refer to our disclaimer on the Medical menu of our website.

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 our website.

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 through primary care.

- McArdle disease is a very rare autosomal recessive disorder of muscle metabolism.
- **Clinical Practice Guidelines for McArdle's are published with open access (page 5).**
- 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 utilize muscle glycogen.
- Maximal isometric activity must be ceased by about 6 seconds to avoid risk of muscle contracture.
- An energy crisis occurs in all activity, severe in the first 10 minutes and throughout intense activity.
- This causes premature fatigue, exaggerated heart rate, pain, muscle spasm and can lead to 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 recognize 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 disease [1] (Glycogen Storage Disease Type V, GSD5) 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 enzyme [2], caused by mutations in the *PYGM* gene, results in an inability to utilize glycogen in skeletal muscle.**

c) Maximal isometric or anaerobic activity for more than about 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 9).**

f) **Patients must be able to recognize the signs that urgent hospital attendance is required (page 9).**

g) A series of CK tests (page 10c) can help patients understand and avoid future serious episodes.

h) In many cases diagnosis is only achieved approx. 20 years after presentation. Muscle damage or atrophy may have occurred by then, causing disability.

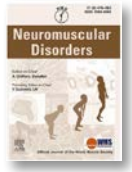
i) Diagnosis in childhood, provision of specialist advice and learning to manage the condition well, can lead to being less severely affected.

j) Patients must learn to attain 'second wind' (page 12f). 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 WF, Illingworth B, Pearson CM, Guillory RJ, Seraydarian K (1956) Proc Natl Acad Sci U S A 45: 791-797.

Clinical Practice Guidelines



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

Visit the following web page to download a free booklet PDF of the 15 page CPG paper, plus the 18 pages of Supplementary Material.
www.iamgsd.org/cpg

McArdle's specialized services

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 center with a multi-disciplinary approach to patient care is therefore highly desirable.

c) **A specialized service for people with McArdle disease and related disorders may be available in your country†. There is a directory of clinicians on the medical menu of the IamGSD website.**

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.

Genetic confirmation

a) Due to the ultra rare nature of McArdle disease, mistakes have been made in diagnosis [1].

b) **If patient has an atypical presentation, check whether they have been genetically confirmed.**

c) DNA analysis of the *PYGM* gene may be undertaken [2] from blood taken locally.

d) Over 150 causative mutations identified to date.

e) There is a significant preponderance of the two most common mutations.

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 pattern [1].**

b) General practices are very unlikely to see a second patient, other than possibly a sibling.

c) Prevalence has been estimated at 1:100,000, based on a carrier frequency of approx. 1:160 [2,3]. In many countries less than half of cases are identified.

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 counseling 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 PJ, Tarnopolsky M, and Vladutiu GD (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 muscle [1] or a sedentary lifestyle may contribute.

Hyperuricemia
Due to high level of purine metabolism, possibly leading to gout and/or renal calculi [2] (page 11).

Obesity (and all its ill effects)
Many are overweight, contributed to by the avoidance of activity due to associated painful cramping if not guided correctly [3].

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

b) Some cases of rhabdomyolysis lead to *acute* renal failure (page 9). However, chronic renal disease is *not* reported to be associated.

c) Data suggests that McArdle's does *not* significantly increase the risk of complications for pregnancy and delivery [3]. Anecdotal evidence shows 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 JG, de Miguel E, Mateos FA, Miranda E, Romera NM, Espinos 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

a) Patients should try to avoid incurring cramps that last for more than a minute or two.

b) **Intense or isometric activity for more than about 6 seconds will risk severe cramps or fixed contractures which last for hours or days.**

c) Such contractures can be incurred accidentally or in extremis (e.g. having to run away from danger).

d) Pain medication will usually be required and medical attention often so.

e) **Muscles recover from fixed contractures but frequently-repeated contractures can accumulate debilitating damage in the long term.**

Pain medication

a) During episodes of fixed contracture or rhabdomyolysis patients are advised to choose pain medications which are not eliminated through the kidneys, as they are stressed due to clearing large amounts of myoglobin.

b) **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 recognize when to slow down or pause for a rest in order to avoid injury [1].**

c) If some muscle injury is incurred, pain relief should be taken *only once activity has ceased.*

d) Patients who start on opioid medications are at risk of dependency and of developing chronic pain [1].

[1] See page 7, reference [3].

Medical emergencies

a) **People with McArdle's are at risk of episodes of rhabdomyolysis with possible acute renal failure and/or compartment syndrome [1].**

b) Episodes cannot be managed in general practice. Patients must understand when to attend the hospital. They should carry an 'Information/ Emergency Card' which has guidance (page 19).

c) Ideally patients should show a letter from their McArdle specialist and the "At hospital" leaflet on arrival, or show their card and/or this page.

d) Kidneys normally recover fully following an episode of rhabdomyolysis and myoglobinuria.

e) **For further information on medical emergencies see: www.iamgsd.org/urgent**

[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 as such, but instead:

Urgent assessment for rhabdomyolysis

- Consider urine analysis for myoglobinuria and full chemistry panel – CK (page 10), glucose, calcium and bone profile, urea and electrolytes.

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.

Potential complications

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

Creatine Kinase

a) **People with McArdle's have raised CK / CPK. Baseline 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 baseline level for 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 go to the hospital. (See *Medical emergencies*, page 9.)

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

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[1] See page 7, reference [3].

Blood

URATE

a) Urate levels are often raised in people with McArdle's, due to increased purine metabolism [1].

b) **There is an increased rate of gout amongst McArdle people compared to the rest of the population [2]. (Approx. 10% compared to 3%.)**

c) Renal stones of uric acid crystals and calcium oxalate can be instigated by raised urate levels [3].

LIVER ENZYMES

d) **Routine screening for unrelated matters may reveal, in people with McArdle's, mildly elevated levels of the liver enzymes ALT and AST [4].**

e) These enzymes are released into the blood when skeletal muscle is damaged [5] 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.

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[1] See page 7, reference [2].

[2] See page 7, reference [3].

[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 (hemoglobin) 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, brushing 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 seconds [2]. 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 recognize it.

h) Tensing muscles (e.g. due to anger, fear or excitement) greatly increases the risk of injury.

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[1] **101 Tips for a Good Life with McArdle Disease.** (2013-2022) Wakelin, Andrew. IamGSD. (See page 19.)

[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 disease [1] – 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 regular exercise routine[2].

d) **At least 45 minutes of aerobic exercise, after attaining 'second wind' (page 12f), five times a week, is strongly recommended [3].**

e) The common mantra 'No pain, no gain' is wrong in McArdle disease.

f) Studies suggest that aerobic training is safe, with improvements in physiological parameters after several weeks. Controlled trials are needed to fully assess the therapeutic effect [4].

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.**

h) **Information to support training of people with McArdle's is available (page 14g).**

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[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 MA, Andreu, AL, et al. (2012) Br J Sports Med doi: 10.1136.

[4] **Exercise and Preexercise Nutrition as Treatment for McArdle Disease.** Nogales-Gadea G, Santalla A, Ballester-Lopez A, Lucia A, et al. (2016) Med Sci Sports Exerc. PMID: 26559449.

Physical therapy

a) **Any physical therapist/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 12e).
- Exercising for short periods without achieving 'second wind' (page 12).
- Advising that pain is acceptable without realizing that McArdle patients are different, see (f) below.
- Failing to monitor CK levels to ensure that muscle damage is not being incurred (page 10).

b) Once a safe and effective exercise routine 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 12 and 13).

d) If physical therapy 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 12e).

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 get what is often described as a "lactic acid burn", as their level of lactate and hydrogen ions does not "rise" on exercise [1].

g) **Web page "Training support" and leaflet "At the gym" are intended for physical therapists/ personal trainers helping people with McArdle's. www.iamgsd.org/training-support**

[1] **A nonischemic forearm exercise test for McArdle Disease.** Kazemi-Esfarjani P, Skomorowska E, Vissing J, et al. 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 7a).

b) **A healthy diet with an appropriate calorie intake to maintain ideal weight is recommended.**

c) Diet remains controversial, further research is required. Ketogenic diet is being investigated.

d) A limited study in 2008 indicates that a carbohydrate-rich diet may be advantageous. (20% fat, 15% protein, 65% carbohydrate – using low glycemic index foods such as vegetables, fruits, pasta, rice, bread and low-fat cheese) [1].

e) 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 minutes [2]. (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 effective [3] 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 ST, Vissing J (2008) J. Neurol. Neurosurg. Psychiatry published online 5 Jun; doi:10.1136/jnnp.2008.146548.

[2] **Effect of oral sucrose shortly before exercise on work capacity in McArdle Disease.** Andersen ST, Haller RG and Vissing J (2008) Arch Neurol 65.

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

Medical providers are alerted to the following areas of risk when treating other conditions in McArdle patients. Care should also be taken when referring patients for physical therapy (page 14).

Statin therapy

a) Although statin medications are generally well-tolerated, the most common side effects relate to skeletal muscle (myalgia, myositis, rhabdomyolysis) [1].

b) **People with McArdle's may be at increased risk of muscle side effects from statins.**

c) If a patient needs a statin, establish their baseline CK level (page 10) before initiating treatment.

d) Monitor symptoms and plasma CK weekly for the first few weeks, then extend the interval in stages to normal practice.

e) Other cholesterol lowering drugs may also worsen myopathy in McArdle patients [2].

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[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

a) **When prescribing for other medical conditions always check for any side effect of rhabdomyolysis (for example succinylcholine).**

b) For a list derived from the British National Formulary [1] see table 12.1, page 145, *The McArdle Disease Handbook* (page 19), free via our website.

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

General anesthetic

a) **McArdle disease may increase the risk of a malignant hyperthermia-type of reaction to anesthetic agents [1].**

b) Rhabdomyolysis, acute renal failure and electrolyte abnormalities may ensue.

c) Although a very small risk, it is advisable to inform the anesthetist of the risk prior to any surgery. Choice of low risk agents and careful monitoring can further lower the risk.

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[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

a) **People with McArdle disease are at increased risk of developing compartment syndrome through use of tourniquets [1].**

b) Prior to surgery, advise the surgeon of the risk.

c) Limit length of use of blood pressure cuffs.

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[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

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

Patient registries

Genetically confirmed patients are requested to register with a patient registry. Details of both at: **www.iamgsd.org/patient-registries**

SANFORD RESEARCH

Sanford CoRDS is a global registry for people with McArdle's and other very rare glycogenoses. It is patient-entered.

EUROMAC

Euromac is the European registry for people with McArdle's and other very rare glycogenoses. Register via your clinician [1].

[1] **Data from the European registry for patients with McArdle disease and other muscle glycogenoses (Euromac).** Scalco RS, Lucia A, Santalla A, Martinuzzi A, Vavla M, Reni G, et al. Orphanet J Rare Dis 2020;15:330. doi:10.1186/s13023-020-01562-x.

Further reading

Some comprehensive papers on McArdle disease:

Phenotype and genotype of 197 British patients with McArdle disease: An observational single-centre study. Pizzamiglio C, Mahroo OA, Khan KN, Patasin M, Quinlivan R. J Inherit Metab Dis. 2021;1-10. doi: 10.1002/jimd.12438

Data from the European registry for patients with McArdle disease and other muscle glycogenoses (Euromac). Scalco RS, Lucia A, Santalla A et al. Orphanet J Rare Dis 15, 330 (2020). doi: 10.1186/s13023-020-01562-x

Centre of Expertise: Comprehensive Care for Patients with McArdle Disease. Reason SL, Cadzow R, Jegier BJ, Wakelin A (2018) . J Rare Disord Diagn Ther. 4:4. doi: 10.21767/2380-7245.100174

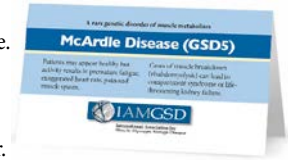
Genotypic and phenotypic features of all Spanish patients with McArdle disease: a 2016 update. Santalla A, Nogales-Gadea G, Encinar AB, et al. BMC Genomics. 2017;18 (Suppl 8):819. Published 2017 Nov 14. doi:10.1186/s12864-017-4188-2

Cards and publications

These publications are available for free on-line browsing and download of free PDFs via our website. **www.iamgsd.org/publications**

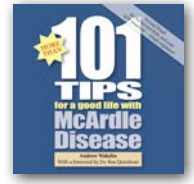
Information/Emergency cards

Patients should carry one of these credit card-sized folding cards in case of an unexpected serious episode. Available from support groups and McArdle's consultants.



- Short description of the condition.
- When patients may need assistance.
- Reminder of the circumstances in which to go to the hospital.
- Link to emergency treatment suggestions, for the hospital doctor.

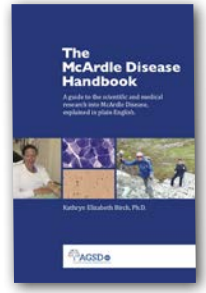
101 Tips for a good life with McArdle Disease



Andrew Wakelin

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, energy reservoir, 30 for 80, etc plus guidance on emergencies. This book will help McArdle people to avoid pain and cramps, reduce hospital visits and get more out of life. *Other language versions are also available.*

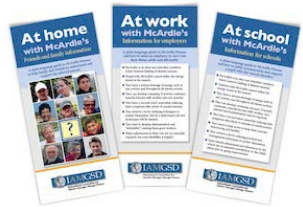
The McArdle Disease Handbook



Kathryn Elizabeth Birch, PhD

Fully referenced to over 260 original research papers. 208 page paperback. A guide to the scientific and medical research. Covers the cause, inheritance, history, symptoms, emotional aspects, treatments, and all the issues which can face McArdle people. Also in German and Italian, printed and PDF, via their GSD support groups.

Leaflets for various situations



A range of leaflets for: attending hospital in an emergency, explaining McArdle's to friends, information for schools and for employers, guidance for physical therapists/personal trainers, etc.

Support groups

About IamGSD

We are a patient-led international group encouraging efforts by research and medical professionals, national support groups and individual patients worldwide.

We have extensive information on our website, together with a range of publications and many videos on our YouTube channel, "IamGSD videos".

National 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. See our website for contact details.

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 2,500 members worldwide. There are also some smaller groups for special interests, such as parents and diet.

IAMGSD
International Association for Muscle Glycogen Storage Disease

www.iamgsd.org

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