



# McArdle's Disease Patients' Survey 2000

Published 20 November 2004

McArdle's Disease (Glycogen Storage Disease Type V) is a metabolic disorder of skeletal muscle which affects sufferers throughout their lives. The effects on each individual vary enormously from mild pain to totally disabling pain and fixed muscle weakness which can restrict patients to wheelchairs.

***“Please help each other help all of us.”***

So said Nick Owston, the initiator of this survey, in June 2000. He took up the challenge of realising the desire of McArdle's patients to help themselves, and each other, through sharing information which would give us all a better understanding of this disease. Sadly Nick died before he could bring the project to fruition. We are honoured to have the opportunity to complete it in his place and dedicate it to his memory.

This a patient-led survey, conducted by patients, participated in by patients and shared with patients for the benefit of patients. It offers no solutions, it just shares the collated information.

## AGSD (UK)

The Association for Glycogen Storage Disease (UK) is a very small charity supporting patients with glycogen storage disorders. The survey was commissioned by and conducted for AGSD (UK) on behalf of all interested McArdle's patients worldwide. In the event, the UK based patients turned out to be only a small part of the total participants.

## PARTICIPATION

Participation was invited from people with McArdle's Disease all over the world via the GSD Net email support group, personal communication, newsletters, etc. Confidentiality was guaranteed. The only selection criteria was that participants had to have a diagnosis of McArdle's disease.

Our thanks go to the 66 people who responded, without whom the survey would not exist. Technical glitches meant 4 participants managed to answer only a few questions, 2 did not have McArdle's and 6 had a diagnosis which was insufficient (see the Diagnosis section, 'Method by which diagnosed'). Therefore, 54 patient questionnaires were finally included. Those who are excluded from the analysis will be disappointed, but we trust that they will never-the-less appreciate the value of the report.

## ANALYSIS

Except where otherwise stated, percentages refer to the percentage of all 54 included participants. Percentages are normally rounded to the nearest whole number. Some sub-analysis is very slightly skewed where participants did not give answers to some questions.

Due to the unfortunate delay in analysing the collected data, participants may find themselves to be in a different condition now to what they were at the time of the survey. However, much of the data relates to participants' historical information rather than current information. As this is not a report on the current condition of individual patients, but a 'snapshot' of the disease at a point in time, we believe the data is still valid.

## STRENGTHS AND WEAKNESSES

**Strengths:** Participation was open to all. Largest base of participants ever assembled. More comprehensive questions than any known previous survey of McArdle's Disease.

**Weaknesses:** The sample size is small (although it is reasonable considering that so few people are diagnosed with McArdle's Disease). Many questions elicited text responses which had to be analysed into 'data'. Some important questions were not thought of at the time.

## NEW SURVEY IN 2005

One of the best things to come out of the survey is that we now have ideas of how to do it even better! If the results of this pilot survey are well received by participants, then we will carry out another survey during 2005 with a better structure, more extensive and clearer questions, and hopefully many more participants, which will allow better statistical analysis. Any McArdle's sufferer who would like to register their interest in participating, and anyone with expertise or just a desire to help, please email Andrew Wakelin at [andrew@cwmclydisaf.freeserve.co.uk](mailto:andrew@cwmclydisaf.freeserve.co.uk).

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Initiated by:	Nick Owston (UK), with assistance from Sharon Johnston (USA).
Conducted on-line by:	Opinion Surveys, OSDotUK Ltd, June 2000.
On behalf of:	Association for Glycogen Storage Disease (UK), © 2004. Registered Charity No. 327841.
Analysed by:	Andrew Wakelin (UK), with assistance from Jenny Coyne (Australia), November 2004.
For more information about this survey data and report, please email the McArdle's representative to AGSD (UK), Andrew Wakelin, at: <a href="mailto:andrew@cwmclydisaf.freeserve.co.uk">andrew@cwmclydisaf.freeserve.co.uk</a> . Extracts from this survey may be quoted with an accompanying credit to AGSD (UK), please contact us in advance for approval.	

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# HIGHLIGHTS

<b>Diagnosis</b>	80% diagnosed by muscle biopsy. 10% inadequately diagnosed and thus omitted from analysis. 67% had a 'trigger' for diagnosis Muscle cramps were the cause for 53% of those triggers. Only 2 centres had diagnosed multiple cases in the study. 91% of participants were the only participant diagnosed at their centre.
<b>Background</b>	69% had experienced symptoms by age 20. Only 6% of diagnosed participants were aged 20 or under at time of survey. 87% were in the 21-60 age range at time of survey. 39% were wrongly labelled psychosomatic, of those 43% were referred to medical professionals.
<b>Pain on exercise</b>	95% reported pain on exercise or afterwards, only 2% (1 participant) has no pain. 83% experience pain so bad as to stop movement. 74% have a technique to help reduce or control pain on exercise. Of those, 60% use a technique during exercise. 94% have pain during everyday activities such as chewing, writing and housework.
<b>Second wind</b>	83% are able to attain a 'second wind'. Of those able to take regular aerobic exercise 96% attain second wind, whereas of those not able to take regular aerobic exercise only 74% achieve second wind. To assist attainment 36% use 'stop and start', 33% use 'slow down' and 20% use both.
<b>Myoglobinuria and lab results</b>	28% experience myoglobinuria at least once per year, 52% up to 10 times in life. 17% have no myoglobinuria. 41% have myoglobinuria lasting 1 day, 59% have myoglobinuria lasting several days. 19% have suffered kidney malfunction. 6% have needed dialysis. 85% have had abnormal CPK levels.
<b>Management and medical supervision</b>	63% use some kind of management regime. 37% follow a high protein, low carbohydrate diet. 43% take regular aerobic exercise. 37% have medical supervision for their management.
<b>Pregnancy and childbirth</b>	71% of mothers with McArdle's have given birth naturally. 29% gave birth by Caesarian. 21% suffered complications of muscle fatigue, 14% muscle spasm, 14% very high CPK.
<b>Other muscle related symptoms</b>	87% have fixed muscle weakness and wasting. 93% have experienced hard muscles (cramps) in legs and feet. 89% have neck, shoulder and spine issues with 61% reporting progression of this. 74% report head and headache issues. 91% report chest and breathing problems. 81% have other upper body issues.
<b>Related medical conditions</b>	52% reported having other major medical conditions related to McArdle's. 28% have heart, blood pressure and cholesterol issues. 35% have difficulties with hearing. 41% have sight problems. 37% have experienced depression.
<b>Lifetime effects</b>	Pain: 33% suffer from serious to intolerable, 57% manageable, 7% mild. Performance: 35% have serious to intolerable limitation, 54% manageable, 7% mild. Endurance: 54% have serious to intolerable limitation, 33% manageable, 11% mild. There is approx 50% decrease in endurance after age 20.
<b>Current situation</b>	85% experience lack of energy, 63% real exhaustion, 89% 'good days and bad days'. 63% regarded their McArdle's symptoms to be progressing, 33% as constant. In the age range 41 to 60, 82% report their condition to be progressing.
<b>Looking back</b>	41% believe more regular exercise would have helped, whereas 15% believe less exercise would have helped. 41% have been made to do things which they feel have done long term damage. For these participants - lifetime pain, performance and endurance is 20-30% worse than for those who had not be made to do such things. 67% reported that the condition becomes more of a problem as you get older.

# DIAGNOSIS

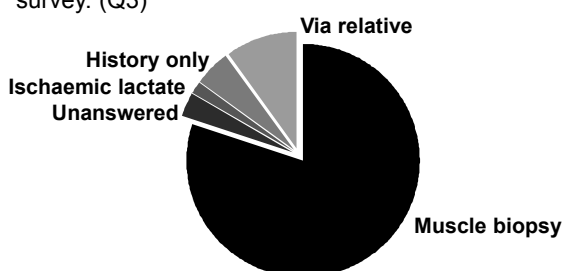
## Method by which diagnosed.

Of 66 participants, 4 answered only one or two questions (probably due to technical glitches) and 2 turned out not to have McArdle's, leaving 60 participants to be considered.

*Included in analysis:*

Method	No. of participants	% of ALL participants
Muscle biopsy	48	80%
Cross reference from a relative	6	10%
TOTAL	54	

Comments: Muscle biopsy is the definitive test for McArdle's as it reveals the absence of myophosphorylase. At the time of the survey DNA testing was not advanced, but where a close relative (especially sibling) has already been properly diagnosed by biopsy, to arrive at a diagnosis on the basis of other evidence is acceptable for inclusion. So 54 out of 60 (90%) valid participants were well diagnosed in terms of this survey. (Q3)



*Excluded from analysis:*

Regrettably, another 6 participants had to be excluded from the analysis as their diagnosis is uncertain. Including them would undermine the validity of the results.

Method	No. of participants	% of ALL participants
Ischaemic lactate test	1	2%
Patient history only	3	5%
Unanswered	2	3%
TOTAL	6	

Comments: It is of concern that 4 participants have been diagnosed with McArdle's only from the ischaemic lactate test or the taking of their history, neither of which are definitive. (Q3)

## Trigger event for diagnosis.

A 'trigger' is defined as an event leading the participant to achieve a diagnosis.

	No. of participants	
A trigger event	36	67%
No trigger event	17	32%
Unanswered	1	2%
TOTAL	54	

Comments: The majority of cases are being picked up because of a trigger event. (Q7a)

## Type of trigger event.

	No. of participants	% of those with trigger
Episode of cramped muscles	19	53%
Unrelated medical investigation	6	17%
Investigation of gout	3	8%
Surgery	2	6%
Friend or relative was a doctor	2	6%
Blood donation	1	3%
Medical for work	1	3%
Wanting to take up sport	1	3%
Death of a relative	1	3%
TOTAL	36	

Comments: 1 episode was witnessed by nurse friends. As well as the 2 cases prompted by a friend or relative who was a doctor, other cases not reporting an 'event' told of friends who pushed them into seeking a diagnosis. 12 cases were diagnosed as a result of medical investigations: of gout, of unrelated symptoms, or prior to or during surgery. (Q7b)

## Location at which diagnosed.

Countries:	No. of participants
Australia	2 (2 cities)
Canada	4 (3 cities)
Denmark	1
Netherlands	1
New Zealand	1
United Kingdom	13 (6 cities)
United States of America	31 (25 cities)
Country not specified	1
TOTAL	54

*Cities with more than one case:*

	No. of participants	No. of hospitals
London	7	5
Chicago	3	3 †
New York	3	3
Rochester, MN	2	1
Halifax, NS	2	1

† Hospital names were not given in full detail, so some uncertainty exists.

*Hospitals with more than one case:*

	No. of participants
National Hospital, Queens Sq, London †	3
Mayo Clinic, Rochester, MN	2

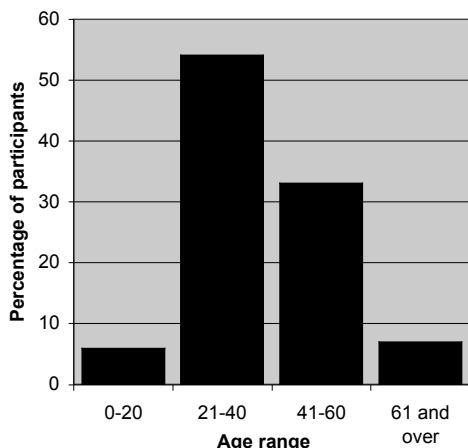
† Dr Brian McArdle, who first identified the disease, worked at this hospital.

Comments: 49 participants (91%) were diagnosed at centres where they were the only case in the survey. The low numbers of cases at centres may contribute to the difficulties which most patients have in achieving diagnosis. (Q2)

# BACKGROUND

## Age of participant.

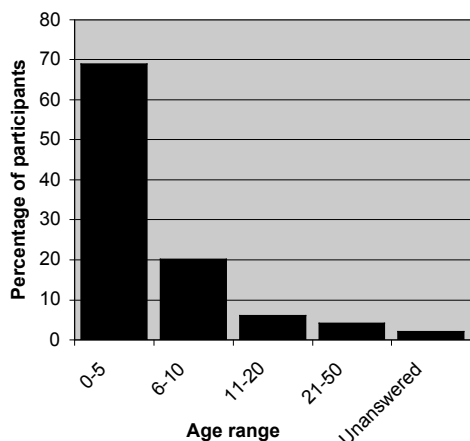
Age range	No. of participants
0 to 20	3 6%
21 to 40	29 54%
41 to 60	18 33%
61 and over	4 7%
<b>TOTAL</b>	<b>54</b>



Comments: The profile reflects the fact that most patients do not manage to obtain a diagnosis until well into adulthood, and that McArdle's disease was first described only in 1951. (Q1)

## Age at which first symptoms experienced.

Age range	No. of participants
0 to 5	39 69%
6 to 10	11 20%
11 to 20	3 6%
21 to 50 †	2 4%
Unanswered	1 2%
<b>TOTAL</b>	<b>54</b>

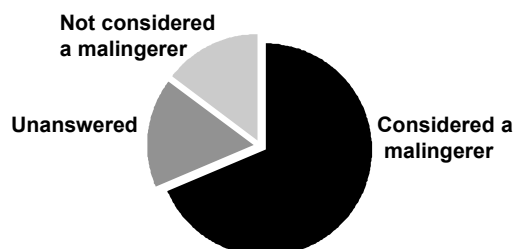


Comments: For those who said "for as long as I can remember" or similar, the age range 0 to 5 is used. Two said they had symptoms from birth or at 3 months. 89% had experienced symptoms by age 10, 95% by age 20. † Both the participants in the 21 to 50 band were diagnosed by biopsy. One said they had "problems from childhood". In a later question neither described their condition as 'late onset'. We conclude that they probably experienced a 'crisis' at the age they declared but experienced minor symptoms earlier. (Q4)

## Considered a malingeringer.

(A person who pretends to be ill in order to avoid having to do something.)

	No. of participants
Considered a malingeringer	37 69%
Not considered a malingeringer	9 17%
Unanswered	8 15%
<b>TOTAL</b>	<b>54</b>



Comments: The perception that the patient is a malingeringer could be a barrier to diagnosis. (Q5a)

## Who considered participant a malingeringer.

	Instances	% of those considered malingeringers
Teachers	28	76%
Doctors	9	24%

Comments: The responses were too varied to allow detailed analysis of 'by whom', but family and friends featured heavily. It is surprising that in so many cases teachers (usually Physical Education) and even doctors feature. (Q5b)

## Condition labelled as psychosomatic.

	No. of participants
Labelled as psychosomatic	21 39%
Not labelled as psychosomatic	33 61%
<b>TOTAL</b>	<b>54</b>

Comments: It was mainly friends, family, teachers, etc who wrongly labelled participants as psychosomatic. There were even a few doctors. This could be a barrier to diagnosis. Three participants referred to depression. (Q6a)

## Referred to a psychologist or psychiatrist.

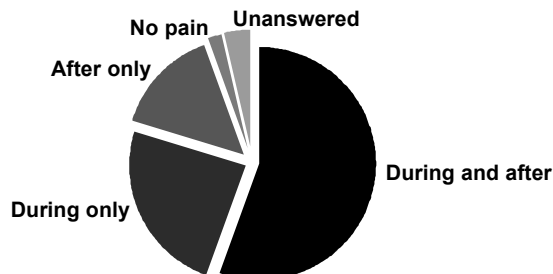
	No. of participants	% of those labelled psychosomatic
Referred	9	43%

Comments: It appears that a large proportion of those considered psychosomatic were referred to professionals. (Q6b)

# PAIN ON EXERCISE

## Pain during and after exertion.

	No. of participants	
Both during and after exertion	30	56%
Only during exertion	13	24%
Only after exertion	8	15%
No pain	1	2%
Unanswered	2	4%
<b>TOTAL</b>	<b>54</b>	



Comments: 80% of participants have pain during exercise. 71% have pain after exercise. The one instance of no pain is that of a child reported by the parent. Descriptions of the pain varied enormously. (Q18)

## Pain so bad that it stops movement altogether.

	No. of participants	
Pain stops movement	45	83%
Does not stop movement	4	7%
Ambivalent	1	2%
Unanswered	4	7%
<b>TOTAL</b>	<b>54</b>	

Comments: Most participants described fixed muscle cramps which prevented movement. (Q19)

## Technique for controlling or reducing the pain.

	No. of participants	
Have technique/s	40	74%
No technique	11	20%
Unanswered	3	6%
<b>TOTAL</b>	<b>54</b>	

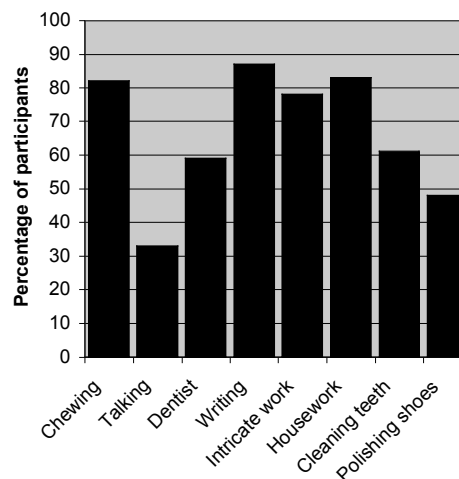
### Phase when technique used:

	Instances	% of those with a technique
Before exercise	7	18%
During exercise	24	60%
After exercise	25	63%

Comments: 26% of participants have been unable to find a technique for controlling or reducing pain. (Q20)

## Muscle fatigue and pain in everyday activities.

Activity:	No. of participants	
Chewing	44	82%
Talking	18	33%
Keeping mouth open at dentist	32	59%
Writing for long periods of time	47	87%
Intricate finger/limb movements	42	78%
Doing housework	45	83%
Cleaning teeth	33	61%
Polishing shoes	26	48%



Comments: 94% of McArdle's participants have fatigue and pain in at least some of these very ordinary everyday activities. (Q24 & Q26)

# SECOND WIND

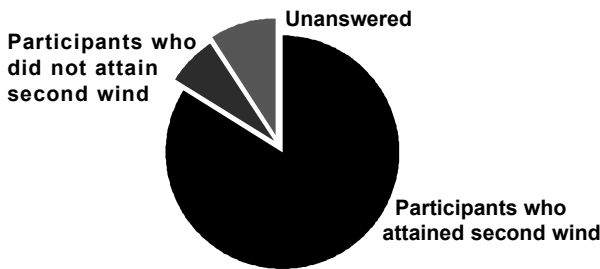
When a McArdle's patient starts exercise their muscle is unable to convert stored glycogen to use as fuel, resulting in pain. The 'second wind' occurs when the exercised muscle switches to use fat for fuel. This typically happens after 8 to 12 minutes†. In this phase exercise may be attained with markedly less pain than in the initial phase.

†Spontaneous "Second Wind" and Glucose-Induced Second "Second Wind" in McArdle Disease; Authors: Ronald G. Haller, MD and John Vissing, MD, PhD; *Archives of Neurology*, 2002; 59: 1395-1402.

<http://archneur.ama-assn.org/cgi/content/abstract/59/9/1395>

## Attainment of 'second wind'.

	No. of participants	
Second wind attained	45	83%
No second wind attained	4	7%
Unanswered	5	9%
<b>TOTAL</b>	<b>54</b>	



Comments: 40 of the 45 participants who attain a second wind use a technique to help them achieve it (see below). Of those able to take regular aerobic exercise 96% attain a second wind, whilst of those not taking such exercise 74% attain a second wind.

*Analysis of medical supervision of management programme, see 'Management and medical supervision' section:*

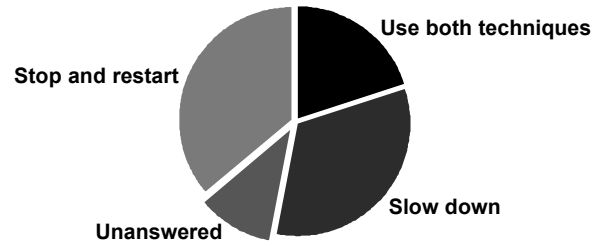
	With supervision		No supervision	
Attaining second wind	17	85%	28	82%
Not attaining second wind	3	15%	6	18%
<b>TOTAL</b>	<b>20</b>		<b>34</b>	

Percentages are of all participants in the 'with supervision' and 'no supervision' groups.

Comments: Having medical supervision of a management programme shows a 3% increase in participants' attainment of second wind. One patient said they had only been able to attain a second wind since they started a programme. (Q21a)

## Techniques for attaining 'second wind'.

	No. of participants	% of those with 2nd wind
Best if stop and restart	16	36%
Best if slow down	15	33%
Use both techniques equally	9	20%
No technique used	5	11%



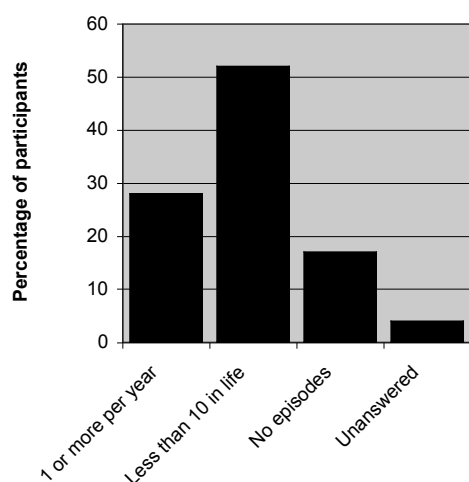
Comments: There is a very even split between preference for stop/start and slow down. (Q21b)

# MYOGLOBINURIA AND LAB RESULTS

NOTE: myoglobinuria shows up as dark coloured urine. It occurs as a result of muscle damage, called rhabdomyolysis. Severe myoglobinuria can cause kidney malfunction as the kidneys try to clear it from the bloodstream.

## Frequency of myoglobinuria episodes.

	No. of participants	
1 or more episodes per year	15	28%
Less than 10 episodes in life	28	52%
No episodes at all	9	17%
Unanswered	2	4%
<b>TOTAL</b>	<b>54</b>	



### Participants with 1 or more episodes per year:

Episodes per year	No. of participants
52 episodes	2
12 episodes	2
10 episodes	4
4 episodes	1
3 episodes	1
2 episodes	3
1 episodes	2
<b>TOTAL</b>	<b>15</b>

Comments: Answers were divided into those who reported lifetime events and those who reported regular events (stated as events per year). The average number of myoglobinuria episodes for participants in the "less than 10 episodes in life" category was 3.1, and for participants in the "1 or more episodes per year" category was 12.2. However, the per year participants included two who said they had events weekly. Excluding those two, the average was 6 events. The reasons why so many participants reported never having had myoglobinuria is worthy of further study, as is the difference between an average of 3 events in a lifetime and an average of 6 per year. (Q9)

## Duration of myoglobinuria episode.

	No. of participants	% of those having an episode
Up to a day	15	41%
Several days	22	59%

Comments: Recovery times were from hours to a week, but could be summarised into two main categories. Clearly, recovery time depends on the severity of the episode as well as the patient's ability to recover. (Q9)

## Experience of kidney malfunction.

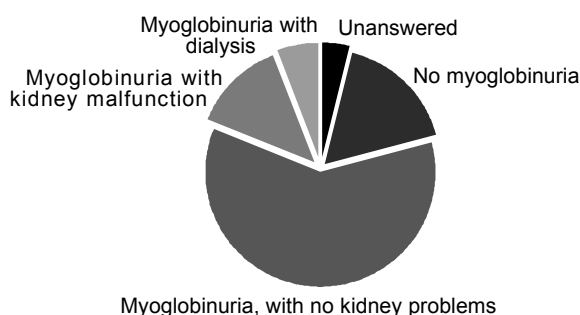
	No. of participants	
Experienced kidney malfunction	10	19%

Comments: Participants may have taken this to mean medically diagnosed kidney malfunction. From the descriptive answers it is deduced that about the same number again had symptoms of kidney overload. (Q9)

## Need for dialysis.

	No. of participants	
Has needed dialysis	3	6%

Comments. This appears to be a very high percentage of participants to require dialysis. (Q9)



## Abnormal laboratory test results.

	No. of participants	
Abnormality reported	46	85%
Nothing unusual	3	6%
Unanswered	5	9%
<b>TOTAL</b>	<b>54</b>	

Lab test	Instances	% of those reporting issues
CPK (CK)†	44	95%
Uric acid	1	2%
Cholesterol	0	0%
Triglycerides	1	2%

† CPK and CK are both abbreviations for Creatine Kinase. Comments: CPK stands out as by far the clearest signal of trouble with 82% of participants aware of raised levels. Regrettably, the software only allowed one, instead of multiple, choices - otherwise the data would probably have shown higher abnormalities for the other lab tests. (Q8)

# MANAGEMENT AND MEDICAL SUPERVISION

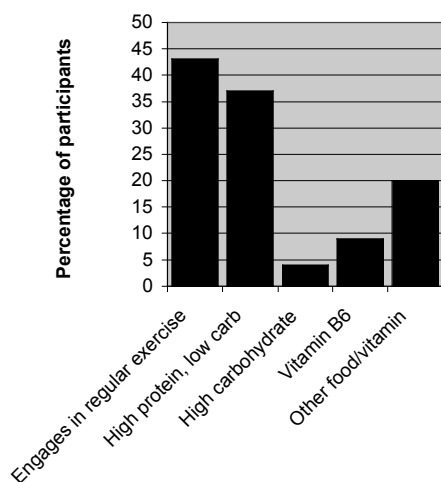
## Use of management, diet or exercise for McArdle's.

	No. of participants	
Management, diet or exercise used	34	63%
None used (or unanswered)	20	37%
TOTAL	54	

Comments: 63% of participants are using one or more of these regimes. (Q11a)

## Type of management regime used.

Regime used:	No. of instances	
Engages in regular aerobic exercise	23	43%
High protein, low carbohydrate diet	20	37%
High carbohydrate diet	2	4%
Vitamin B6 supplement	5	9%
Any other food or vitamin supplement	11	20%



Comments: Some participants used more than one regime. Regular aerobic exercise is the only regime which is agreed to be beneficial by consultants experienced with McArdle's. There is a clear preference for a high-protein rather than high-carbohydrate diet. (Q11b)

## Medical supervision of management.

	No. of participants	
Currently under medical supervision	20	37%
Not under medical supervision	34	63%
TOTAL	54	

### Of those under medical supervision:

Country	No. of participants under supervision	% of participants in country
Denmark	1	100%
United Kingdom	7	54%
United States of America	12	39%

Comments: It is disappointing that only 37% of participants are receiving medical supervision of their management. In only 3 countries are participants reporting medical supervision. Whilst Denmark, with only 1 patient, is statistically not relevant, UK is encouraging with 54% of its reported 13 participants receiving medical supervision, and USA is not far behind at 39% of 31. (Q12)

# PREGNANCY AND CHILDBIRTH

## Experience of pregnancy and childbirth.

	No. of participants
Normal childbirth	10
Child delivered by Caesarian because of McArdle's	4
Complications during pregnancy because of McArdle's	1

Comments: Out of 14 mothers reporting births, 10 or 71% delivered "normally". 12 of the births were in USA (8 normal, 4 Caesarian) and 2 in UK (both normal). (Q38)

## Complications during pregnancy and childbirth.

	Instances	% of all pregnancies
Muscle fatigue and exhaustion	3	21%
Toxaemia	2	14%
Muscle spasm	2	14%
Very high CPK (CK)	2	14%
Dangerously low protein levels	1	7%
Unable to push	1	7%
Unknown "issues"	1	7%

Percentages are of the 14 total reported pregnancies.

Comments: Participants' descriptions were analysed to find the main complication. 9 participants gave an explanation, some had more than one complication. Many participants gave birth before they had been diagnosed with McArdle's. Would their treatment have been different had they received a diagnosis? One participant reported difficulty coping with the extra weight during pregnancy. (Q39)

# OTHER MUSCLE RELATED SYMPTOMS

## Fixed muscle weakness and wasting.

	No. of participants	
Fixed weakness or wasting	47	87%
None experienced	5	9%
Unanswered	2	4%
<b>TOTAL</b>	<b>54</b>	

	Instances	
Upper body muscles less developed than lower body muscles	31	57%
Suffer pain and weakness in the lower back	41	76%
Pain and weakness in the lower back more marked with age	34	63%

Comments: Lower back weakness and wasting is prevalent. (Q40)

## Hard muscles in legs and feet.

	No. of participants	
Experienced hard muscles	50	93%
Not experienced	3	6%
Unanswered	1	2%
<b>TOTAL</b>	<b>54</b>	

Comments: (Q28)

## Pain and weakness in neck, shoulder and spine.

	No. of participants	
Pain and weakness	48	89%
No and unanswered	6	11%
<b>TOTAL</b>	<b>54</b>	

Area:	Instances	
Neck and/or shoulders	42	78%
Middle back	25	46%
Lower back	38	70%

Comments: Perceived muscle weakness in the upper body is a very common problem. (Q27a)

*Progression of these problems:*

	No. of participants	
Progression experienced	33	61%
No progression	12	22%
Unanswered	9	17%
<b>TOTAL</b>	<b>54</b>	

Comments: (Q27b)

## Head and headaches.

	No. of participants	
Problems experienced	40	74%
No and unanswered	14	26%
<b>TOTAL</b>	<b>54</b>	

*Symptoms attributed to McArdle's:*

Symptom	Instances	
Headaches	19	35%
Feeling of tightness in the head	19	35%
Decreased concentration	29	54%

Comments: Headaches may well be caused by cramps in muscles around the head and neck. (Q36)

*Attributed cause of these problems:*

Cause	Instances	% of all causes
Tiredness, fatigue or exhaustion	9	37%
Tight neck and/or head muscles	8	15%
Stress	3	5%
Cramping and other muscle problems	3	5%
Depression	1	2%
Pain	1	2%
Over exertion	1	2%

Comments: Of the 40 participants above who said they had these head symptoms, 23 gave an explanation of why they attributed them to McArdle's. 3 gave more than one cause. (Q37)

## Problems with chest and breathing.

	No. of participants	
Problems experienced	49	91%
No problems	4	7%
Unanswered	1	2%
<b>TOTAL</b>	<b>54</b>	

	Instances	
Weakness in chest	19	35%
Difficulty breathing	17	32%
Shortness of breath	28	52%
Episodes of apnoea† while sleeping	15	28%
Nausea associated with exercise	42	78%
Nausea NOT associated with exercise	20	37%
Inconsistent heart rate	25	46%

Comments: † Temporary cessation of breathing. (Q34 & Q35)

## Muscle fatigue or pain causing other problems.

	No. of participants	
Problems caused	44	81%
No and unanswered	10	19%
<b>TOTAL</b>	<b>54</b>	

Problem:	Instances	% of those with problems
Choking easily	17	32%
Tired voice	24	44%
Other upper body problems	40	74%

Comments: The majority of patients have these problems. (Q25)

# RELATED MEDICAL CONDITIONS

NOTES: a) Some participants may have interpreted this section to be about ALL other medical conditions, whereas others may have taken it to be about only those conditions which medical knowledge associates with McArdle's.  
b) These figures need to be compared to the incidence of these problems within the general population.

## Major medical conditions which may be influenced by McArdle's.

	No. of participants	
Condition/s experienced	28	52%
No conditions	22	41%
Unanswered	4	7%
<b>TOTAL</b>	<b>54</b>	

Conditions:	Instances	
Heart conditions	8	15%
High blood pressure	5	9%
Arthritis and gout	3	6%
Hyperthyroidism	2	4%
High cholesterol level	2	4%
Depression	2	4%
Osteoporosis	2	4%
Cancer	2	4%

In addition, the following conditions involved only one case: Crohn's disease, nephritis, polyneuropathy, esophagus narrowing, hearing loss, fibromyalgia, diabetes, stroke, alcoholism, psoriasis, sleep apnoea (temporary cessation of breathing), environmental sensitivity, gluten allergy, asthma, ankylosing spondylitis, menopause.

*Of those having a major condition:*

Age range	No. of participants	% of participants in range
0 to 20	1	33%
21 to 40	9	31%
41 to 60	19	83%
61 and over	3	75%

Comments: There is a high incidence of heart related issues - heart conditions, high blood pressure and high cholesterol. (Q10)

## High blood pressure.

	No. of participants	
High blood pressure experienced	9	17%
No high blood pressure	45	83%
<b>TOTAL</b>	<b>54</b>	

*Of those having high blood pressure:*

Age range	No. of participants	% of participants in range
21 to 40	2	7%
41 to 60	6	33%
61 and over	2	50%

Comments: The number of participants with high blood pressure here is higher than in the section above. This difference is probably explained by those with high blood pressure for reasons which they do not associate with McArdle's. (Q13)

## Difficulty with hearing.

	No. of participants	
Difficulty with hearing	19	35%
No difficulty	35	65%
<b>TOTAL</b>	<b>54</b>	

*Of those having hearing difficulty:*

Age range	No. of participants	% of participants in range
21 to 40	9	31%
41 to 60	8	44%
61 and over	2	50%

Comments: Hearing difficulties appear to be unusually high in the 21 to 40 age range. Within that range the participants are well spread in age starting at age 22. (Q14)

## Sight problems which participant associates with McArdle's.

	No. of participants	
Sight problems	22	41%
No sight problems	29	54%
Unanswered	3	6%
<b>TOTAL</b>	<b>54</b>	

Age band	No. of participants	% in age band
0 to 20	1	33%
21 to 40	9	31%
41 to 60	11	61%
61 and over	1	25%

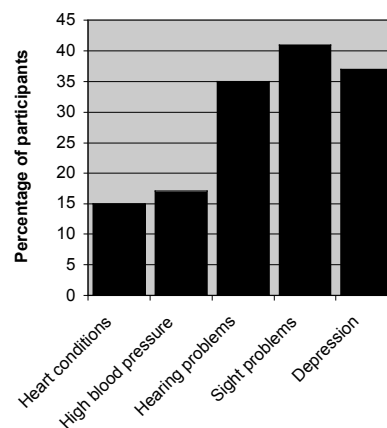
Condition:	Instances	
Focussing your eyes	18	33%
Seeing double	8	15%
Redness	5	9%
Burning	6	11%

Comments: (Q23)

## Depression which participant attributes to McArdle's.

	No. of participants	
Depression experienced	20	37%
No depression	32	59%
Unanswered	2	4%
<b>TOTAL</b>	<b>54</b>	

Comments: The question only elicited a Yes or No answer. These answers are likely to be for perceived depression rather than clinically diagnosed depression. (Q33)

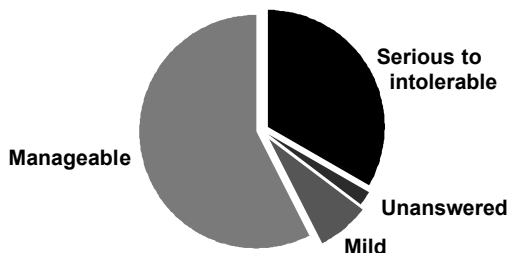


# LIFETIME EFFECTS

NOTE: Participants were given a scale against which to describe their lifetime experience of pain, performance and endurance. This was: 7-10 serious to intolerable and totally inhibiting, 4-6 manageable but getting worse, and 1-3 mild.

## PAIN caused by McArdle's throughout life.

Amount of pain	No. of participants	
Serious to intolerable	18	33%
Manageable	31	57%
Mild	4	7%
Unanswered	1	2%
TOTAL	54	



Comments: The average pain reference was 5.9. 33% of participants have pain which they describe as "serious to intolerable". 90% have "manageable" to "intolerable" pain.

Analysis of taking regular aerobic exercise, see "Management and medical supervision":

Amount of pain	Regular exercise		No regular exercise	
	No.	%	No.	%
Serious to intolerable	4	17%	14	47%
Manageable	17	74%	14	47%
Mild	2	9%	2	7%
Unanswered	0	0%	1	3%
TOTAL	23		31	

Comments: For those unable to take regular exercise 74% have "serious to intolerable" pain; whereas for those able to take regular aerobic exercise only 17% have "serious to intolerable" pain. There is an overall lessening of pain for those able to take regular exercise.

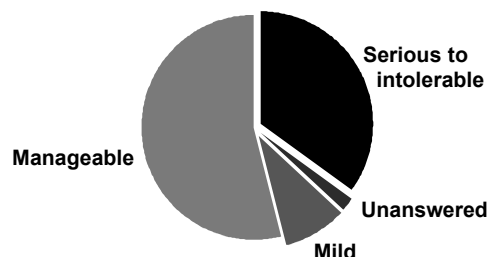
Average pain ref. for participants in age range:

0 to 20	4.6
21 to 40	5.9
41 to 60	6.0
61 and over	5.5

Comments: Average pain levels appear to increase from the lowest age band, but then stabilise. However, Spearman's correlation coefficient shows that there is no significant relationship between age and pain caused by McArdle's ( $\rho = .03$ ,  $p < .05$ ). The older participants do not suffer from more severe pain than the younger participants. (Q15)

## Amount PERFORMANCE was limited by McArdle's throughout life.

Performance limitation	No. of participants	
Serious to intolerable	19	35%
Manageable	29	54%
Mild	5	9%
Unanswered	1	2%
TOTAL	54	



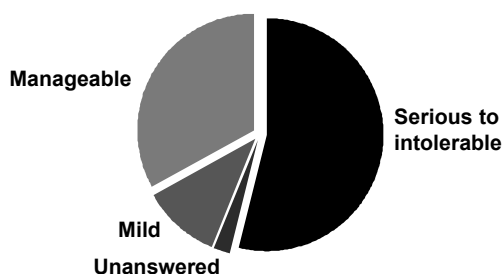
Average performance ref. for participants in age range:

0 to 20	4.3
21 to 40	5.7
41 to 60	5.9
61 and over	6.0

Comments: The question did not define performance; but intended the avoidance of certain tasks due to the disease. The average performance reference was 5.7. 35% of participants have "serious to intolerable" limitation of performance. 89% have "moderate" to "intolerable" limitation to performance. While it may appear that there is a gradual worsening of performance with age, Spearman's correlation coefficient shows that there is no significant relationship between age and performance ( $\rho = .13$ ,  $p < .05$ ). The older participants do not suffer from more limitation of performance than the younger participants. (Q16)

## Amount ENDURANCE was limited by McArdle's throughout life.

Endurance limitation	No. of participants	
Serious to intolerable	29	54%
Manageable	18	33%
Mild	6	11%
Unanswered	1	2%
TOTAL	54	



Average endurance ref. for participants in age range:

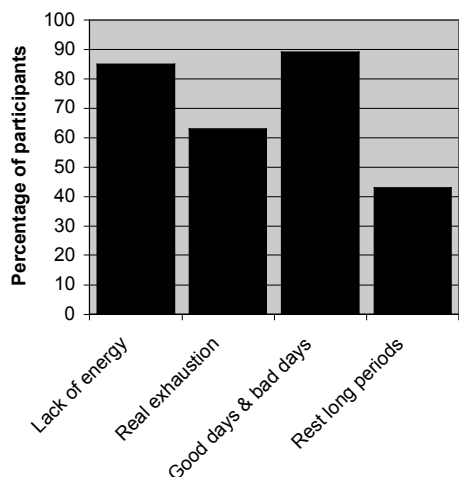
0 to 20	4.3
21 to 40	6.5
41 to 60	6.8
61 and over	5.8

Comments: The average endurance reference was 6.5. 54% of participants have "serious to intolerable" limitation of endurance. 87% have "moderate" to "intolerable" limitation to endurance. While it may appear that there is a decrease in endurance with age, Spearman's correlation coefficient shows that there is no significant relationship between age and endurance ( $\rho = 1.00$ ,  $p < .05$ ). The older participants do not suffer from lower endurance than the younger participants. (Q17)

# CURRENT SITUATION

## General exhaustion and tiredness.

	Instances	
Lack of energy attributable to McArdle's	46	85%
Real exhaustion	34	63%
Good days and bad days	48	89%
Rest/sleep for long periods helps	23	43%



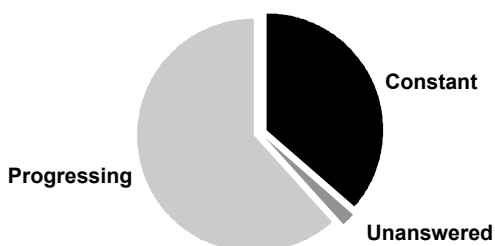
Comments: No participants reported themselves to be free from these problems. 2 left the question unanswered. (Q32)

## Perceived progression of symptoms.

Note: Participants were asked how they regarded their McArdle's "condition" (symptoms) with a choice of 5 options.

	No. of participants	
Constant	18	33%
Progressing	22	41%
Progressing since about age of 40	8	15%
Progressing since about age of 50	4	7%
Late onset	0	0%
Unanswered	2	4%
<b>TOTAL</b>	<b>54</b>	

Comments: "Progressing" is the description chosen by 63% of participants across all ages.



Age band	Constant		Progressing	
0 to 20	2	67%	1	33%
21 to 40	11	39%	17	61%
41 to 60	3	18%	14	82%
61 and over	2	50%	2	50%

Percentages are of the participants in the age band.

Comments: Participants' perception of their symptoms appears to swing from erring towards "Constant" to being definitely "Progressing" as age passes. This peaks at 82% "Progressing" in the 41 to 60 age band. (Q22)

# LOOKING BACK

## Participants aged 20 or over looking back on their case.

	No. of participants	
More regular exercise would have helped	22	41%
Less exercise would have helped	8	15%
Made to do things which have done long term damage †	22	41%
Have always been fairly inactive physically	15	28%

Comments: There is a significant balance in favour of more exercise being viewed as beneficial. (Q41)

† Analysis of long term damage against lifetime effects:

	Long term damage	No long term damage
Lifetime pain	6.6	5.4
Lifetime performance	6.6	5.1
Lifetime endurance	7.5	5.8

Participants who said they had been made to do things which caused long term damage had average references 20% to 30% higher than those who had not be made to do things which caused long term damage.

	No. of participants	
Was very much more active during youth	32	59%
Condition is more of a problem as you get older	36	67%
Condition is less of a problem as you get older	1	2%
Less of a problem as you get older because the demands on you are less	4	7%

Comments: 6 participants (10%) said none of these applied to them, implying that the condition has stayed exactly the same. (Q42)

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# “MY WORST MCARDLE’S EXPERIENCE”

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## The worst McArdle’s event/attack experienced by participants.

The answers to this question would almost make a book! We’ve included here the story from Nick Owston, who was never slow to tell a story against himself if it helped to highlight the dangers for the benefit of others.

“I was on a journey from Australia at 19 years old, on the old Orient Express railway, stopped somewhere in Yugoslavia. On the platform an American girl asked me to look after her scooter for her whilst she made some enquiries. To my horror, the train started an unscheduled departure - all my worldly goods were on it! I decided, rather too late, to abandon the scooter and run after the train. I ran off the end of the platform and along between the rails. At first I was gaining on the train, but then I was gripped by McArdle’s pain. Very soon, unable to continue lifting my feet over the sleepers I collapsed, paralysed and in agony.

Minutes went by, I was in a delirium wondering what to do, when I realised that the train was coming back towards me! Unable to stand, I just managed to roll off the railway line in time to avoid being crushed by it.

The train had just been changing engines. It took me days to recover from the muscle cramps and myoglobinuria. I never knew who the girl was, but she was watching all this and thought I must be mad.”

NICK OWSTON, JUNE 1947 - JUNE 2003

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The following are the main situations/events reported by the other participants. (Q43)

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Collapses in public places.  
Severe fixed muscle spasms, sometimes locked in position, especially squats.  
Cramped abdomen muscles.  
Cramped in water, with risk of drowning.  
Collapse into a lake.  
School runs - vomiting after run and another not even getting out of the school gates.  
Hospitalisation at age 7.  
7 to 10% muscle loss in an ‘event’.  
Very high CPK - up to 250,000.  
Military basic training and school fitness tests.  
Myoglobinuria and 5 days in hospital.  
Skiing, hiking, tennis, softball, tug-of-war.  
Weight-lifting and household/work lifting.  
Embarrassment at having to be carried, once out of the school gym.  
PE classes, dancing, trampoline, gyms, monkey bars.  
Back spasm after fall from wheelchair.  
Unable to get off bike after a race.  
Unable to straighten up after sit ups.  
Swollen kidneys, kidney failure and dialysis.  
Hypotonia at age 3 months.  
School days generally.  
Leading people up two flights of stairs.  
Standing on a pipe - locked muscles.  
Enduring the ischaemic forearm test.

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## NEW SURVEY IN 2005

We plan to organise a new survey during 2005 with a better structure, more extensive and clearer questions, and hopefully many more participants.

Any McArdle’s sufferer who would like to register their interest in participating, please email:

Andrew Wakelin at [andrew@cwmclydisaf.freemove.co.uk](mailto:andrew@cwmclydisaf.freemove.co.uk).

We will advise you when the survey is ready for you on-line.